Locus 1 (rs5498)

SNiPAcard

Block annotations

Block info

genomic range	chr19:10,391,770-10,409,323
block size	17,554 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.064 [-2.744 – 0.21]	gene(s) hit or close-by	CTD-2369P2.5, CTD-2369P2.8, ICAM1, ICAM4, ICAM5
phastCons	μ = 0.040 [0 – 0.256]	eQTL gene(s)	ICAM4, ICAM5
GERP++	μ = -0.089 [-2.57 – 2.66]	potentially regulated gene(s)	ICAM4, KRI1, P2RY11, PPAN, TYK2, TYK2, ZGLP1
CADD score	μ = 4.307 [0.068 – 10.49]	disease gene(s)	ICAM1, ICAM4, TYK2, P2RY11

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Soluble ICAM-1	$<6.00\times10^{-89}$	GWAS Catalog	21533024	1
Soluble levels of adhesion molecules	$<4.00\times10^{-23}$	GWAS Catalog	20167578	1
Soluble ICAM-1	$<5.00\times10^{-25}$	GWAS Catalog	18604267	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM003891	1

Disease gene annotation

gene	trait	source DB	source entry/link
ICAM1	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
ICAM4	BLOOD GROUP SYSTEM, LANDSTEINER-WIENER	OMIM	MIM:111250
TYK2	IMMUNODEFICIENCY 35	OMIM	MIM:611521
ICAM1	Chronic mucocutaneous candidosis	OrphaNet	OrphaNet:1334
TYK2	Oligoarticular juvenile arthritis	OrphaNet	OrphaNet:85410
TYK2	Juvenile rheumatoid factor-negative polyarthritis	OrphaNet	OrphaNet:85408
TYK2	Autosomal recessive hyper-IgE syndrome due to TYK2 deficiency	OrphaNet	OrphaNet:331226
P2RY11	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ICAM1	missense variant	ENST00000423829	?	ENSP00000413124	E/K	Gag/Aag	?	?	1
ICAM1	missense variant	ENST00000264832	NM_000201.2	ENSP00000264832	E/K	Gag/Aag	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ICAM5	?	ENSG00000105376	transformed fibroblasts	3.11×10^{-10} (p-value)	GTEx Portal V6	11
ICAM5	?	ENSG00000105376	thyroid	1.35×10^{-7} (p-value)	GTEx Portal V6	11
ICAM4	?	ENSG00000105371	blood	1.55×10^{-8} (p-value)	GTEx Portal V6	10
ICAM4	ENST00000380770	ILMN_1681296	blood	1.21×10^{-26} (p-value)	MuTHER consortium	4
ICAM4	ENST00000393717					
ICAM4	ENST00000340992					
ICAM4	ENST00000380770	ILMN_1681296	monocyte	5.86×10^{-18} (p-value)	Fairfax et al.	3
ICAM4	ENST00000393717					
ICAM4	ENST00000340992					
?	?	ILMN_2393067	monocyte	2.44×10^{-14} (p-value)	Fairfax et al.	3

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000309935	1	ENST00000380770, ENST00000393717, ENST00000340992	ICAM4

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000223642	1	ENCP00000024048	TYK2 TYK2
		ENCP00000024047	TYK2 TYK2
		ENCP00000024036	PPAN
		ENCP00000024049	TYK2 TYK2
		ENCP00000024035	PPAN
		ENCP00000024037	P2RY11
		ENCP00000024051	KRI1
		ENCP00000024034	PPAN
		ENCP00000024038	ZGLP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001417842 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me2, SP1, USF1, H3K4me3, H3K27me3, H3K36me3
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	H3K4me3, DNase1, H3K36me3, Egr1, H3K27ac, Max, PU1, E2F6, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, CTCF
		skin (NHDF-AD)	H3K4me2, H3K27me3
		muscle (HSMM)	H2AZ, H3K4me2, H3K27me3
		liver (HepG2)	H3K4me1, H3K4me2, H3K4me3, H3K36me3, H3K27me3, DNase1
		blood (GM12878)	DNase1, PU1, PolII, H2AZ, CTCF, Egr1, BCL3, H3K4me3, H3K4me2, H3K36me3
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3
		nervous (NH-A)	DNase1, H3K4me2, H3K27me3
		skin (NHEK)	H3K27me3, H3K4me3, H3K4me2
		NHLF	H3K27me3
		Osteobl	H3K4me2, H3K27me3
		blood (DND-41)	H3K36me3, H3K4me1
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, PolII, H3K4me3, Max, H3K27ac, H3K4me2, H3K9ac, Cmyc, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H4K20me1, H3K36me3, H3K27me3, H3K4me3

		endothelium (HUVEC) A549	H3K36me3, DNase1, H3K27me3, Max DNase1, H3K4me2, H3K4me3, H3K27me3
ENSR00000343265 e! (promoter flanking region)	2	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	CTCF, Rad21, H3K4me2, H3K27me3, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	PolII, H3K36me3
		skin (NHDF-AD)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3, H3K4me1, DNase1
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	H3K4me2, H3K4me3
		blood (GM12878)	PolII, H2AZ, ELF1, H3K4me3, DNase1
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K27me3
		A549	H3K4me2, H3K4me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-2369P2.5 e!	upstream gene variant	1439	ENST00000592893 e!	?	?	2
CTD-2369P2.8 e!	downstream gene variant, upstream gene variant	794	ENST00000589379 e!	?	?	5
ICAM1 e!	upstream gene variant, downstream gene variant	724	ENST00000592686 e!	?	?	6
ICAM1 e!	downstream gene variant	1254	ENST00000588645 e!	?	ENSP00000465680 e!	3
ICAM1 e!	upstream gene variant, downstream gene variant	296	ENST00000585443 e!	?	?	5
ICAM1 e!	downstream gene variant	112	ENST00000264832 e!	NM_000201.2	ENSP00000264832 e!	2
ICAM1 e!	downstream gene variant	964	ENST00000423829 e!	?	ENSP00000413124 e!	3
ICAM4 e!	upstream gene variant, downstream gene variant	247	ENST00000340992 e!	NM_001039132.2	ENSP00000342114 e!	5
ICAM4 e!	upstream gene variant, downstream gene variant	277	ENST00000393717 e!	NM_022377.3	ENSP00000377320 e!	5
ICAM4 e!	upstream gene variant, downstream gene variant	240	ENST00000380770 e!	NM_001544.4	ENSP00000370147 e!	5
ICAM5 e!	upstream gene variant, downstream gene variant	866	ENST00000586480 e!	?	?	5
ICAM5 e!	upstream gene variant, downstream gene variant	555	ENST00000587398 e!	?	ENSP00000464796 e!	5
ICAM5 e!	upstream gene variant, downstream gene variant	3266	ENST00000588912 e!	?	?	4
ICAM5 e!	upstream gene variant, downstream gene variant	3297	ENST00000586004 e!	?	?	4
ICAM5 e!	upstream gene variant, downstream gene variant	864	ENST00000221980 e!	NM_003259.3	ENSP00000221980 e!	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2369P2.5 e!	ENST00000592893 e!	?	?	3
ICAM1 e!	ENST00000423829 e!	?	ENSP00000413124 e!	3
ICAM1 e!	ENST00000264832 e!	NM_000201.2	ENSP00000264832 e!	3
ICAM1 e!	ENST00000588645 e!	?	ENSP00000465680 e!	3
ICAM1 e!	ENST00000585443 e!	?	?	1
ICAM5 e!	ENST00000221980 e!	NM_003259.3	ENSP00000221980 e!	3
ICAM5 e!	ENST00000586004 e!	?	?	1
ICAM5 e!	ENST00000586480 e!	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ICAM1	ENST00000264832	NM_000201.2	ENSP00000264832	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTD-2369P2.5	ENST00000592893	?	1
CTD-2369P2.8	ENST00000589379	?	2
ICAM5	ENST00000588912	?	1

Locus 2 (rs4129267)

SNiPAcard

Block annotations

Block info

genomic range	chr1:154,395,839-154,428,283
block size	32,445 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.413 [-2.774 – 1.935]	gene(s) hit or close-by	IL6R
phastCons	μ = 0.041 [0 – 0.875]	eQTL gene(s)	AQP10, IL6R, POU5F1P4, UBE2Q1
GERP++	μ = -0.744 [-5.29 – 2.48]	potentially regulated gene(s)	C1orf189, IL6R, UBAP2L
CADD score	μ = 3.576 [0.199 – 12.65]	disease gene(s)	IL6R

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Cerebrospinal fluid levels of Alzheimer's disease-related proteins	<6.00×10-63	GWAS Catalog	25340798	1
Fibrinogen	<6.00×10-27	GWAS Catalog	23969696	1
Asthma	<2.00×10-8	GWAS Catalog	21907864	1
C-reactive protein levels	<2.00×10-48	GWAS Catalog	21300955	1
C-reactive protein	<2.00×10-14	GWAS Catalog	19567438	1
Protein quantitative trait loci	<2.00×10-57	GWAS Catalog	18464913	1
PULMONARY FUNCTION	<7.00×10-6	GWAS Catalog	17903307	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CS034756	1

Disease gene annotation

gene	trait	source DB	source entry/link
IL6R	INTERLEUKIN 6, SERUM LEVEL OF, QUANTITATIVE TRAIT LOCUS	OMIM	MIM:614752
IL6R	SOLUBLE INTERLEUKIN-6 RECEPTOR, SERUM LEVEL OF, QUANTITATIVE [...]	OMIM	MIM:614689

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AQP10	ENST00000324978	ILMN_2090004	blood	3.32×10-6 (p-value)	Westra et al.	1
AQP10	ENST00000484864					
UBE2Q1	ENST00000292211	ILMN_1776325	blood	5.52×10-7 (p-value)	Westra et al.	6
IL6R	ENST00000368485	ILMN_1754753	blood	1.45×10-6 (p-value)	Westra et al.	2
IL6R	ENST00000344086					
IL6R	?	ENSG00000160712	transverse colon	2.66×10-6 (p-value)	GTEx Portal V6	18
IL6R	?	ENSG00000160712	blood	1.18×10-6 (p-value)	GTEx Portal V6	19

IL6R	?	ENSG00000160712	tibial artery	6.60×10-6 (p-value)	GTEx Portal V6	16
POU5F1P4	?	ENSG00000237872	tibial artery	2.41×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000004651		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000037853	1	ENCP00000004625	C1orf189
ENCE00000037890	1	ENCP00000004651	IL6R
		ENCP00000004650	IL6R
		ENCP00000004633	UBAP2L

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000545357 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me1
		muscle (HSMM)	DNase1
		liver (HepG2)	FOSL2, Jund, FOXA2, USF1, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, Cmyc, DNase1
		lung (IMR90)	DNase1, H3K18ac, H3K27ac, H3K36me3
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K27me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K36me3, DNase1, H3K4me1
		NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K9ac, H3K4me1
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me1, Max, Cmyc
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H4K20me1, H3K36me3
		endothelium (HUVEC)	Cjun, Max, H3K4me1, DNase1
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac
ENSR00000545365 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	H3K27me3, Egr1, Max
		skin (NHDF-AD)	H3K4me1, DNase1, H3K9ac, H3K27ac
		muscle (HSMM)	DNase1
		liver (HepG2)	H3K36me3, DNase1
		lung (IMR90)	DNase1, H3K36me3
		nervous (NH-A)	DNase1, H3K27me3
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K36me3
		NHLF	DNase1, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1, Jund, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K36me3
		endothelium (HUVEC)	Cjun
ENSR00000545369 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	Egr1, CTCF, DNase1, H3K27me3
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF

		lung (IMR90)	H3K27me3, H3K36me3, CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	CTCF, H3K36me3
		NHLF	DNase1, H3K36me3
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	H3K36me3, H3K27ac, H4K20me1
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF
ENSR00000545370	4	NHLF	H3K36me3, DNase1, H3K4me1, H3K27ac
(promoter flanking region)		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		lung (IMR90)	DNase1, H3K18ac, H3K27ac, H3K4me2, H3K27me3, H3K36me3
		blood (GM12878)	Tcf12, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
ENSR00000545373	2	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K36me3
(promoter flanking region)		HSMMtube	H3K27me3
		lung (IMR90)	H3K36me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K4me1, H3K36me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL6R	upstream gene variant	67	ENST00000507256	?	?	3
IL6R	upstream gene variant	321	ENST00000502679	?	?	2
IL6R	upstream gene variant, downstream gene variant	473	ENST00000515190	?	ENSP00000423036	3
IL6R	downstream gene variant	1399	ENST00000622330	NM_001206866.1	ENSP00000477739	4
IL6R	downstream gene variant	360	ENST00000512471	?	ENSP00000423184	4
IL6R	downstream gene variant	3001	ENST00000476006	?	ENSP00000423668	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL6R	ENST00000507256	?	?	23
IL6R	ENST00000476006	?	ENSP00000423668	23
IL6R	ENST00000344086	NM_181359.2	ENSP00000340589	27
IL6R	ENST00000368485	NM_000565.3	ENSP00000357470	27
IL6R	ENST00000512471	?	ENSP00000423184	3
IL6R	ENST00000515190	?	ENSP00000423036	23
IL6R	ENST00000622330	NM_001206866.1	ENSP00000477739	6
IL6R	ENST00000502679	?	?	1

Locus 3 (rs11553746)

SNiPAcard

Block annotations

Block info

genomic range	chr2:211,317-296,410
block size	85,094 bp
variant count	48 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.104 [-2.209 – 5.667]	gene(s) hit or close-by	AC079779.4, ACP1, FAM150B, SH3YL1
phastCons	μ = 0.120 [0 – 1]	eQTL gene(s)	ACP1, SH3YL1
GERP++	μ = -0.328 [-5.58 – 5.87]	potentially regulated gene(s)	ACP1, SH3YL1
CADD score	μ = 3.895 [0.08 – 19.46]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
3-(cystein-S-yl)acetaminophen*	4.62×10^{-5}	Metabolomics GWAS Server	24816252	14

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Acid phosphatase 1, soluble, a/b polymorphism of	benign	ClinVar	RCV000014681.1	1
ACID PHOSPHATASE 1, SOLUBLE, A/B POLYMORPHISM OF	OMIM curated	OMIM	MIM:171500	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ACP1	missense variant	ENST00000272065	NM_004300.3	ENSP00000272065	R/Q	cGa/cAa	?	?	1
ACP1	missense variant	ENST00000453390	?	ENSP00000411121	I/T	aTa/aCa	?	?	1
ACP1	missense variant	ENST00000405233	?	ENSP00000384307	I/T	aTa/aCa	?	?	1
ACP1	missense variant	ENST00000407983	NM_001040649.2	ENSP00000385404	I/T	aTa/aCa	?	?	1
ACP1	missense variant	ENST00000272067	NM_007099.3	ENSP00000272067	R/Q	cGa/cAa	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SH3YL1	?	ENSG00000035115	pancreas	4.33×10^{-30} (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	muscularis mucosae	2.30×10^{-11} (p-value)	GTEx Portal V6	46
ACP1	?	ENSG00000143727	muscularis mucosae	1.02×10^{-6} (p-value)	GTEx Portal V6	41
SH3YL1	?	ENSG00000035115	lung	2.20×10^{-6} (p-value)	GTEx Portal V6	41

SH3YL1	?	ENSG00000035115	atrial appendage	5.26×10-12 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	tibial artery	3.69×10-6 (p-value)	GTEx Portal V6	31
ACP1	?	ENSG00000143727	tibial artery	1.67×10-6 (p-value)	GTEx Portal V6	19
SH3YL1	?	ENSG00000035115	blood	3.57×10-30 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	thyroid	2.73×10-37 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	skeletal muscle	2.13×10-13 (p-value)	GTEx Portal V6	46
ACP1	?	ENSG00000143727	skeletal muscle	5.67×10-6 (p-value)	GTEx Portal V6	2
SH3YL1	?	ENSG00000035115	transverse colon	3.79×10-9 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	unexposed skin	3.10×10-36 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	sun exposed skin	3.04×10-54 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	left ventricle	1.07×10-12 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	subcutaneous adipocytes	4.80×10-8 (p-value)	GTEx Portal V6	44
SH3YL1	?	ENSG00000035115	stomach	5.68×10-7 (p-value)	GTEx Portal V6	40
SH3YL1	?	ENSG00000035115	sigmoid colon	5.87×10-8 (p-value)	GTEx Portal V6	44
SH3YL1	?	ENSG00000035115	tibial nerve	8.72×10-17 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	esophagus mucosa	3.58×10-75 (p-value)	GTEx Portal V6	46
SH3YL1	?	ENSG00000035115	ovary	7.94×10-7 (p-value)	GTEx Portal V6	37
SH3YL1	ENST00000479739	ILMN_1712231	b-cell	5.27×10-11 (p-value)	Fairfax et al.	5
SH3YL1	ENST00000356150					
SH3YL1	ENST00000463865					
SH3YL1	ENST00000403712					
SH3YL1	ENST00000403657					
SH3YL1	ENST00000473104					
SH3YL1	ENST00000405430					
SH3YL1	ENST00000468321					
SH3YL1	ENST00000626873					
SH3YL1	?	ENSG00000035115	blood	8.90×10-3 (q-value)	SeeQTL DB (HapMap)	8
SH3YL1	?	ENSG00000035115	adrenal gland	1.45×10-6 (p-value)	GTEx Portal V6	27
SH3YL1	?	ENSG00000035115	terminal ileum	1.86×10-6 (p-value)	GTEx Portal V6	1
SH3YL1	?	ENSG00000035115	transformed fibroblasts	1.11×10-5 (p-value)	GTEx Portal V6	1
SH3YL1	?	ENSG00000035115	aorta	8.96×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024883		
ENCP00000024888		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000234638	1	ENCP00000024888	SH3YL1
		ENCP00000024889	ACP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001538827 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K9ac, USF1, H3K4me2, TAF1, Yy1, H3K27ac, TAF7, Egr1, PolII, H3K27me3
		HSMMtube	H3K79me2, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	Egr1, H3K27ac, Max, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, PolII, H3K4me2, H3K36me3, H3K27me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2
		liver (HepG2)	FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K4me2, Yy1, H4K20me1, PolII, H3K79me2, TAF1, H3K4me1, DNase1
		blood (GM12878)	H3K79me2, Yy1, DNase1, H2AZ, PolII, Cmyc, H3K4me3, H3K27ac, H3K36me3, H3K9ac, H3K4me2, ZEB1
		lung (IMR90)	H3K79me2, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	CTCF, H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Nrf1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, PolII, H3K4me2, H3K4me3, Max
		A549	H3K4me3, H3K4me2, H3K36me3, H3K27ac, DNase1, H3K9ac
ENSR00000675428 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, H3K36me3, CTCF
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K79me2, Rad21, CTCF, H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	CTCF
ENSR00001649416 (CTCF binding site)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		skin (NHDF-AD)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00000584027 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
	1		

ACP1	ENST00000272065 ENST00000272067 ENST00000405364 ENST00000413140 ENST00000442386 ENST00000453390 ENST00000463831 ENST00000480874 ENST00000484464	hsa-miR-9-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC079779.4	upstream gene variant	1096	ENST00000427831	?	?	3
AC079779.4	upstream gene variant	948	ENST00000450709	?	?	3
ACP1	upstream gene variant, downstream gene variant	251	ENST00000272065	NM_004300.3	ENSP00000272065	7
ACP1	upstream gene variant, downstream gene variant	2370	ENST00000463831	?	?	6
ACP1	upstream gene variant, downstream gene variant	272	ENST00000413140	?	ENSP00000410331	7
ACP1	upstream gene variant, downstream gene variant	248	ENST00000272067	NM_007099.3	ENSP00000272067	7
ACP1	upstream gene variant, downstream gene variant	295	ENST00000439645	?	ENSP00000408596	6
ACP1	upstream gene variant, downstream gene variant	279	ENST00000442386	?	ENSP00000389681	7
ACP1	upstream gene variant, downstream gene variant	584	ENST00000484464	?	?	7
ACP1	upstream gene variant, downstream gene variant	344	ENST00000453390	?	ENSP00000411121	7
ACP1	upstream gene variant, downstream gene variant	279	ENST00000407983	NM_001040649.2	ENSP00000385404	6
ACP1	upstream gene variant, downstream gene variant	298	ENST00000405364	?	ENSP00000384184	5
ACP1	upstream gene variant, downstream gene variant	121	ENST00000484125	?	?	6
ACP1	upstream gene variant, downstream gene variant	315	ENST00000480874	?	?	5
ACP1	upstream gene variant, downstream gene variant	305	ENST00000405233	?	ENSP00000384307	6
FAM150B	downstream gene variant	468	ENST00000463919	?	?	4
FAM150B	downstream gene variant, upstream gene variant	974	ENST00000452023	?	ENSP00000389939	4
FAM150B	downstream gene variant	2926	ENST00000405290	?	ENSP00000385672	1
FAM150B	downstream gene variant	2926	ENST00000619265	?	ENSP00000478180	1
FAM150B	downstream gene variant	2926	ENST00000401489	?	ENSP00000385214	1
FAM150B	downstream gene variant	2948	ENST00000344414	?	ENSP00000339565	1
FAM150B	downstream gene variant	2763	ENST00000401503	?	ENSP00000385537	1
FAM150B	downstream gene variant	2555	ENST00000403610	NM_001002919.2	ENSP00000384604	2
SH3YL1	downstream gene variant, upstream gene variant	387	ENST00000468321	?	?	7
SH3YL1	downstream gene variant, upstream gene variant	2557	ENST00000472861	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	195	ENST00000403712	NM_001159597.2	ENSP00000384276	7
SH3YL1	downstream gene variant, upstream gene variant	162	ENST00000356150	NM_015677.3	ENSP00000348471	7
SH3YL1	downstream gene variant, upstream gene variant	36	ENST00000471948	?	?	11
SH3YL1	downstream gene variant, upstream gene variant	588	ENST00000403657	?	ENSP00000385668	6
SH3YL1	downstream gene variant, upstream gene variant	1661	ENST00000602998	?	?	2
SH3YL1	upstream gene variant	2557	ENST00000402632	?	ENSP00000384910	4
SH3YL1	downstream gene variant, upstream gene variant	2302	ENST00000477707	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	969	ENST00000462719	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	280	ENST00000403658	NM_001282682.1	ENSP00000383928	7
SH3YL1	downstream gene variant	923	ENST00000481932	?	ENSP00000474411	5

SH3YL1	downstream gene variant, upstream gene variant	48	ENST00000405430	?	ENSP00000384269	7
SH3YL1	downstream gene variant, upstream gene variant	438	ENST00000431160	?	ENSP00000399519	8
SH3YL1	downstream gene variant, upstream gene variant	590	ENST00000473104	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	25	ENST00000472012	?	?	7
SH3YL1	downstream gene variant, upstream gene variant	229	ENST00000463865	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	713	ENST00000415368	?	ENSP00000410235	5
SH3YL1	downstream gene variant, upstream gene variant	411	ENST00000451005	?	ENSP00000416312	5
SH3YL1	downstream gene variant, upstream gene variant	53	ENST00000488979	?	?	7
SH3YL1	downstream gene variant, upstream gene variant	1428	ENST00000454318	?	ENSP00000415723	7
SH3YL1	downstream gene variant, upstream gene variant	1382	ENST00000475027	?	?	7
SH3YL1	downstream gene variant, upstream gene variant	1174	ENST00000497051	?	?	5
SH3YL1	downstream gene variant, upstream gene variant	225	ENST00000488044	?	?	10
SH3YL1	downstream gene variant, upstream gene variant	167	ENST00000460974	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	69	ENST00000626873	NM_001282687.1	ENSP00000485824	7
SH3YL1	downstream gene variant, upstream gene variant	1213	ENST00000605370	?	?	6
SH3YL1	downstream gene variant, upstream gene variant	394	ENST00000479739	?	ENSP00000441266	7
SH3YL1	downstream gene variant, upstream gene variant	2133	ENST00000465733	?	?	6

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ACP1	ENST00000439645	?	ENSP00000408596	D	gaT/gaC	1
ACP1	ENST00000272067	NM_007099.3	ENSP00000272067	D	gaT/gaC	1
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	S	agC/agT	1
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	S	agC/agT	1
SH3YL1	ENST00000405430	?	ENSP00000384269	S	agC/agT	1

Non-coding exon variant (splice region)

gene	affected transcript	RefSeq id	variant(s)
SH3YL1	ENST00000468321	?	1
SH3YL1	ENST00000463865	?	1
SH3YL1	ENST00000472012	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC079779.4	ENST00000450709	?	?	1
AC079779.4	ENST00000427831	?	?	5
ACP1	ENST00000407983	NM_001040649.2	ENSP00000385404	5
ACP1	ENST00000484125	?	?	2
ACP1	ENST00000484464	?	?	7
ACP1	ENST00000439645	?	ENSP00000408596	6
ACP1	ENST00000272067	NM_007099.3	ENSP00000272067	7

ACP1	ENST00000480874	?	?	7
ACP1	ENST00000442386	?	ENSP00000389681	7
ACP1	ENST00000413140	?	ENSP00000410331	7
ACP1	ENST00000453390	?	ENSP00000411121	6
ACP1	ENST00000272065	NM_004300.3	ENSP00000272065	7
ACP1	ENST00000405364	?	ENSP00000384184	7
ACP1	ENST00000405233	?	ENSP00000384307	5
FAM150B	ENST00000463919	?	?	1
FAM150B	ENST00000405290	?	ENSP00000385672	6
FAM150B	ENST00000619265	?	ENSP00000478180	6
FAM150B	ENST00000452023	?	ENSP00000389939	3
FAM150B	ENST00000344414	?	ENSP00000339565	6
FAM150B	ENST00000401503	?	ENSP00000385537	6
FAM150B	ENST00000401489	?	ENSP00000385214	6
FAM150B	ENST00000403610	NM_001002919.2	ENSP00000384604	6
SH3YL1	ENST00000468321	?	?	25
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	24
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	24
SH3YL1	ENST00000463865	?	?	25
SH3YL1	ENST00000479739	?	ENSP00000441266	21
SH3YL1	ENST00000465733	?	?	4
SH3YL1	ENST00000403657	?	ENSP00000385668	22
SH3YL1	ENST00000462719	?	?	4
SH3YL1	ENST00000488044	?	?	14
SH3YL1	ENST00000481932	?	ENSP00000474411	2
SH3YL1	ENST00000451005	?	ENSP00000416312	18
SH3YL1	ENST00000431160	?	ENSP00000399519	8
SH3YL1	ENST00000460974	?	?	1
SH3YL1	ENST00000475027	?	?	13
SH3YL1	ENST00000405430	?	ENSP00000384269	24
SH3YL1	ENST00000415368	?	ENSP00000410235	7
SH3YL1	ENST00000473104	?	?	23
SH3YL1	ENST00000488979	?	?	12
SH3YL1	ENST00000402632	?	ENSP00000384910	13
SH3YL1	ENST00000497051	?	?	6
SH3YL1	ENST00000454318	?	ENSP00000415723	13
SH3YL1	ENST00000471948	?	?	12
SH3YL1	ENST00000472012	?	?	7
SH3YL1	ENST00000403658	NM_001282682.1	ENSP00000383928	25
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	24
SH3YL1	ENST00000472861	?	?	4

Locus 3 (sentinel SNP rs11553746 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

SH3YL1	ENST00000477707	?	?	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACP1	ENST00000442386	?	ENSP00000389681	2
ACP1	ENST00000413140	?	ENSP00000410331	2
ACP1	ENST00000453390	?	ENSP00000411121	1
ACP1	ENST00000405364	?	ENSP00000384184	1
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	5
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	5
SH3YL1	ENST00000479739	?	ENSP00000441266	4
SH3YL1	ENST00000403657	?	ENSP00000385668	5
SH3YL1	ENST00000481932	?	ENSP00000474411	1
SH3YL1	ENST00000405430	?	ENSP00000384269	5
SH3YL1	ENST00000402632	?	ENSP00000384910	3
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	5

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	1
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ACP1	ENST00000484125	?	1
ACP1	ENST00000484464	?	2
ACP1	ENST00000463831	?	2
SH3YL1	ENST00000468321	?	4
SH3YL1	ENST00000602998	?	1
SH3YL1	ENST00000463865	?	5
SH3YL1	ENST00000473104	?	4
SH3YL1	ENST00000471948	?	3

Locus 4 (rs9858542)

SNiPAcard

Block annotations

Block info

genomic range	chr3:49,385,417-49,731,861
block size	346,445 bp
variant count	107 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.217 [-3.849 – 1.994]	**gene(s) hit or close-by**	AC099668.1, AC099668.5, AMT, APEH, BSN, BSN-AS1, BSN-AS2, DAG1, GPX1, MST1, NICN1, RHOA, RHOA-IT1, RNA5SP130, RNF123, TCTA
phastCons	μ = 0.097 [0 – 1]	**eQTL gene(s)**	ACTBP13, AMT, APEH, C3orf62, CDHR4, CISH, DOCK3, FAM212A, GPX1, IP6K2, KLHDC8B, MIR5193, MST1, MST1R, NICN1, QRICH1, RBM6, RN7SL217P, RNF123, RP11-3B7.1, UBA7, USP4, WDR6
GERP++	μ = -0.427 [-10.1 – 4.85]	**potentially regulated gene(s)**	AMT, AMT, APEH, ARIH2, BSN, CAMKV, CDHR4, DAG1, DALRD3, IMPDH2, IP6K1, KLHDC8B, LAMB2, MON1A, MST1, NDUFAF3, QARS, QRICH1, RP11-3B7.1, SLC25A20, TRAIP, UBA7, USP4
CADD score	μ = 4.712 [0.004 – 22.1]	**disease gene(s)**	AMT, KLHDC8B, LAMB2, NDUFAF3, SLC25A20, QARS, DAG1, CISH, MST1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Educational attainment	$<5.00\times10^{-9}$	GWAS Catalog	25201988	1
Ulcerative colitis	$<8.00\times10^{-7}$	GWAS Catalog	25082827	1
Inflammatory bowel disease	$<1.00\times10^{-47}$	GWAS Catalog	23128233	1
Ulcerative colitis	$<2.00\times10^{-17}$	GWAS Catalog	21297633	1
Primary sclerosing cholangitis	$<1.00\times10^{-16}$	GWAS Catalog	21151127	1
Crohn's disease (time to surgery)	$<6.00\times10^{-17}$	GWAS Catalog	21102463	1
Ulcerative colitis	$<4.00\times10^{-9}$	GWAS Catalog	20228799	1
Ulcerative colitis	$<7.00\times10^{-9}$	GWAS Catalog	19915572	1
Crohn's disease (time to surgery)	$<1.00\times10^{-12}$	GWAS Catalog	18587394	1
Crohn's disease (time to surgery)	$<4.00\times10^{-8}$	GWAS Catalog	17554300	1
Crohn's disease (time to surgery)	$<5.00\times10^{-8}$	GWAS Catalog	17554261	1
benzoate	6.67×10^{-5}	Metabolomics GWAS Server	24816252	2

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	likely benign	ClinVar	RCV000116863.2	1
Clinvar: phenotype not specified	not provided	ClinVar	RCV000024450.1	1
Clinvar: phenotype not specified	not provided	ClinVar	RCV000024446.1	1
Glutathione peroxidase polymorphism	benign	ClinVar	RCV000017462.1	1
?	HGMD curated	HGMD	CR045943	1
?	HGMD curated	HGMD	CR045942	1
?	HGMD curated	HGMD	CM087622	1

?	HGMD curated	HGMD	CM061794	HGMD	1
GLUTATHIONE PEROXIDASE POLYMORPHISM	OMIM curated	OMIM	MIM:138320	OMIM	1

Disease gene annotation

gene	trait	source DB	source entry/link	
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899	OMIM
KLHDC8B	LYMPHOMA, HODGKIN, CLASSIC	OMIM	MIM:236000	OMIM
LAMB2	NEPHROTIC SYNDROME, TYPE 5, WITH OR WITHOUT OCULAR [...]	OMIM	MIM:614199	OMIM
LAMB2	PIERSON SYNDROME	OMIM	MIM:609049	OMIM
NDUFAF3	MITOCHONDRIAL COMPLEX I DEFICIENCY	OMIM	MIM:252010	OMIM
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OMIM	MIM:212138	OMIM
QARS	MICROCEPHALY, PROGRESSIVE, WITH SEIZURES AND CEREBRAL AND [...]	OMIM	MIM:615760	OMIM
DAG1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE), TYPE C, [...]	OMIM	MIM:613818	OMIM
CISH	MYCOBACTERIUM TUBERCULOSIS, SUSCEPTIBILITY TOMYCOBACTERIUM TUBERCULOSIS, [...]	OMIM	MIM:607948	OMIM
CISH	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
CISH	BACTEREMIA, SUSCEPTIBILITY TO, 2	OMIM	MIM:614383	OMIM
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899	OMIM
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	DECIPHER	MIM:212138	OMIM
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	DECIPHER	MIM:615760	OMIM
DAG1	muscular dystrophy-dystroglycanopathy limb-girdle type C7 (MDDGC7)	DECIPHER	MIM:613818	OMIM
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860	orphanet
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857	orphanet
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863	orphanet
MST1	Primary sclerosing cholangitis	OrphaNet	OrphaNet:171	orphanet
LAMB2	Synaptic congenital myasthenic syndromes	OrphaNet	OrphaNet:98915	orphanet
LAMB2	LAMB-2-related infantile-onset nephrotic syndrome	OrphaNet	OrphaNet:306507	orphanet
LAMB2	PIERSON SYNDROME	OrphaNet	OrphaNet:2670	orphanet
NDUFAF3	Isolated NADH-CoQ reductase deficiency	OrphaNet	OrphaNet:2609	orphanet
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OrphaNet	OrphaNet:159	orphanet
DAG1	Autosomal recessive limb-girdle muscular dystrophy type 2P	OrphaNet	OrphaNet:280333	orphanet
DAG1	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	OrphaNet	OrphaNet:370997	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
BSN	missense variant	ENST00000296452	NM_003458.3	ENSP00000296452	T/A	Acc/Gcc	?	?	1
GPX1	missense variant	ENST00000620890	?	ENSP00000478837	L/P	cTc/cCc	?	?	1
GPX1	missense variant	ENST00000419783	NM_000581.2	ENSP00000407375	L/P	cTc/cCc	?	?	1
MST1	missense variant	ENST00000449682	NM_020998.3	ENSP00000414287	C/R	Tgc/Cgc	?	?	1
MST1	missense variant	ENST00000448220	?	ENSP00000394756	C/R	Tgc/Cgc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GPX1	?	ENSG00000233276	muscularis mucosae	1.54×10^{-8} (p-value)	GTEx Portal V6	106

RBM6	?	ENSG00000004534	muscularis mucosae	2.15×10-6 (p-value)	GTEx Portal V6	102
NICN1	?	ENSG00000145029	muscularis mucosae	4.84×10-7 (p-value)	GTEx Portal V6	106
RNF123	?	ENSG00000164068	muscularis mucosae	7.42×10-6 (p-value)	GTEx Portal V6	48
IP6K2	?	ENSG00000068745	muscularis mucosae	3.61×10-5 (p-value)	GTEx Portal V6	1
AMT	?	ENSG00000145020	muscularis mucosae	2.79×10-5 (p-value)	GTEx Portal V6	3
RBM6	?	ENSG00000004534	lung	1.43×10-7 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	lung	2.85×10-7 (p-value)	GTEx Portal V6	105
AMT	?	ENSG00000145020	lung	4.83×10-7 (p-value)	GTEx Portal V6	104
AMT	?	ENSG00000145020	transformed fibroblasts	6.52×10-7 (p-value)	GTEx Portal V6	105
IP6K2	?	ENSG00000068745	transformed fibroblasts	2.39×10-8 (p-value)	GTEx Portal V6	80
APEH	?	ENSG00000164062	transformed fibroblasts	6.85×10-13 (p-value)	GTEx Portal V6	106
RNF123	?	ENSG00000164068	transformed fibroblasts	1.48×10-5 (p-value)	GTEx Portal V6	39
RBM6	?	ENSG00000004534	transformed fibroblasts	6.37×10-6 (p-value)	GTEx Portal V6	11
GPX1	?	ENSG00000233276	tibial artery	8.26×10-8 (p-value)	GTEx Portal V6	105
RBM6	?	ENSG00000004534	tibial artery	6.49×10-8 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	tibial artery	1.09×10-10 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	tibial artery	2.71×10-10 (p-value)	GTEx Portal V6	106
IP6K2	?	ENSG00000068745	tibial artery	3.53×10-5 (p-value)	GTEx Portal V6	1
RNF123	?	ENSG00000164068	tibial artery	8.74×10-6 (p-value)	GTEx Portal V6	96
RBM6	?	ENSG00000004534	breast	9.52×10-10 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	breast	1.22×10-5 (p-value)	GTEx Portal V6	2
RBM6	?	ENSG00000004534	blood	4.69×10-8 (p-value)	GTEx Portal V6	106
UBA7	?	ENSG00000182179	blood	3.32×10-8 (p-value)	GTEx Portal V6	104
GPX1	?	ENSG00000233276	thyroid	2.62×10-8 (p-value)	GTEx Portal V6	105
RBM6	?	ENSG00000004534	thyroid	1.97×10-10 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	thyroid	3.22×10-9 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	thyroid	1.04×10-6 (p-value)	GTEx Portal V6	89
RNF123	?	ENSG00000164068	thyroid	6.41×10-6 (p-value)	GTEx Portal V6	87
MST1	?	ENSG00000173531	thyroid	4.85×10-8 (p-value)	GTEx Portal V6	101
FAM212A	?	ENSG00000185614	skeletal muscle	7.82×10-15 (p-value)	GTEx Portal V6	106
IP6K2	?	ENSG00000068745	skeletal muscle	2.44×10-5 (p-value)	GTEx Portal V6	3
RBM6	?	ENSG00000004534	skeletal muscle	2.87×10-5 (p-value)	GTEx Portal V6	2
UBA7	?	ENSG00000182179	transverse colon	9.12×10-9 (p-value)	GTEx Portal V6	106
RBM6	?	ENSG00000004534	transverse colon	9.71×10-6 (p-value)	GTEx Portal V6	2
UBA7	?	ENSG00000182179	unexposed skin	1.69×10-5 (p-value)	GTEx Portal V6	50
MST1	?	ENSG00000173531	unexposed skin	3.70×10-7 (p-value)	GTEx Portal V6	82
RBM6	?	ENSG00000004534	unexposed skin	3.48×10-6 (p-value)	GTEx Portal V6	7

RBM6	?	ENSG00000004534	sun exposed skin	1.84×10-10 (p-value)	GTEx Portal V6	106
KLHDC8B	?	ENSG00000185909	sun exposed skin	1.22×10-5 (p-value)	GTEx Portal V6	32
ACTBP13	?	ENSG00000234667	sun exposed skin	2.94×10-6 (p-value)	GTEx Portal V6	43
APEH	?	ENSG00000164062	sun exposed skin	2.71×10-5 (p-value)	GTEx Portal V6	1
MST1	?	ENSG00000173531	sun exposed skin	2.34×10-6 (p-value)	GTEx Portal V6	32
WDR6	?	ENSG00000178252	sun exposed skin	3.57×10-5 (p-value)	GTEx Portal V6	1
DOCK3	?	ENSG00000088538	sun exposed skin	3.30×10-5 (p-value)	GTEx Portal V6	2
GPX1	?	ENSG00000233276	aorta	5.78×10-6 (p-value)	GTEx Portal V6	33
NICN1	?	ENSG00000145029	aorta	3.82×10-7 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	aorta	3.13×10-6 (p-value)	GTEx Portal V6	87
RBM6	?	ENSG00000004534	aorta	4.07×10-5 (p-value)	GTEx Portal V6	1
RBM6	?	ENSG00000004534	subcutaneous adipocytes	2.38×10-9 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	subcutaneous adipocytes	3.02×10-10 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	subcutaneous adipocytes	4.25×10-11 (p-value)	GTEx Portal V6	106
APEH	?	ENSG00000164062	subcutaneous adipocytes	7.03×10-7 (p-value)	GTEx Portal V6	105
RNF123	?	ENSG00000164068	subcutaneous adipocytes	3.52×10-6 (p-value)	GTEx Portal V6	98
MST1R	?	ENSG00000164078	subcutaneous adipocytes	2.36×10-5 (p-value)	GTEx Portal V6	1
RBM6	?	ENSG00000004534	visceral adipocytes	5.54×10-7 (p-value)	GTEx Portal V6	102
MIR5193	?	ENSG00000263506	tibial nerve	4.13×10-7 (p-value)	GTEx Portal V6	97
RBM6	?	ENSG00000004534	tibial nerve	1.57×10-8 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	tibial nerve	7.68×10-8 (p-value)	GTEx Portal V6	105
AMT	?	ENSG00000145020	tibial nerve	7.07×10-7 (p-value)	GTEx Portal V6	104
MST1	?	ENSG00000173531	tibial nerve	2.47×10-8 (p-value)	GTEx Portal V6	103
RNF123	?	ENSG00000164068	tibial nerve	1.40×10-6 (p-value)	GTEx Portal V6	101
WDR6	?	ENSG00000178252	esophagus mucosa	7.08×10-7 (p-value)	GTEx Portal V6	24
RBM6	?	ENSG00000004534	esophagus mucosa	6.37×10-9 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	esophagus mucosa	1.28×10-6 (p-value)	GTEx Portal V6	72
FAM212A	?	ENSG00000185614	esophagus mucosa	1.66×10-9 (p-value)	GTEx Portal V6	106
APEH	?	ENSG00000164062	esophagus mucosa	1.64×10-7 (p-value)	GTEx Portal V6	103
UBA7	?	ENSG00000182179	esophagus mucosa	2.67×10-5 (p-value)	GTEx Portal V6	1
QRICH1	?	ENSG00000198218	pituitary	1.34×10-6 (p-value)	GTEx Portal V6	67
GPX1	?	ENSG00000233276	cerebellar hemisphere	4.45×10-7 (p-value)	GTEx Portal V6	105
GPX1	?	ENSG00000233276	cerebellum	1.29×10-7 (p-value)	GTEx Portal V6	105
MST1R	?	ENSG00000164078	cerebellum	4.34×10-7 (p-value)	GTEx Portal V6	40
RNF123	?	ENSG00000164068	cerebellum	1.83×10-5 (p-value)	GTEx Portal V6	2
GPX1	?	ENSG00000233276	frontal cortex	6.00×10-6 (p-value)	GTEx Portal V6	62
RNF123	?	ENSG00000164068	atrial appendage	2.55×10-7 (p-value)	GTEx Portal V6	97

RBM6	?	ENSG00000004534	atrial appendage	2.51×10-5 (p-value)	GTEx Portal V6	1
GPX1	?	ENSG00000233276	atrial appendage	1.44×10-5 (p-value)	GTEx Portal V6	2
RBM6	ENST00000442092	ILMN_1738239	monocyte	1.28×10-6 (p-value)	Fairfax et al.	15
RBM6	ENST00000266022		b-cell	2.89×10-6 (p-value)	Fairfax et al.	15
RBM6	ENST00000454079					
RBM6	ENST00000422955					
RBM6	ENST00000419610					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
USP4	ENST00000351842	ILMN_1798712	monocyte	7.21×10-8 (p-value)	Fairfax et al.	14
USP4	ENST00000483212					
C3orf62	ENST00000343010					
USP4	ENST00000485450					
UBA7	ENST00000497908	ILMN_1794612	b-cell	8.34×10-11 (p-value)	Fairfax et al.	1
UBA7	ENST00000333486					
MIR5193	ENST00000584510					
CDHR4	?	ENSG00000187492	testis	1.64×10-5 (p-value)	GTEx Portal V6	64
IP6K2	?	ENSG00000068745	testis	4.07×10-5 (p-value)	GTEx Portal V6	1
RN7SL217P	?	ENSG00000264706	testis	2.24×10-5 (p-value)	GTEx Portal V6	35
WDR6	ENST00000615452	ILMN_1669484	skin	9.77×10-15 (p-value)	MuTHER consortium	38
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
APEH	ENST00000480772	ILMN_1718023	blood	2.51×10-5 (p-value)	MuTHER consortium	35
APEH	ENST00000469362					
APEH	ENST00000296456					
APEH	ENST00000438011					
USP4	ENST00000351842	ILMN_1798712	adipocyte	7.71×10-6 (p-value)	MuTHER consortium	31
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
RP11-3B7.1	ENST00000622888	ILMN_1807244	adipocyte	1.14×10-5 (p-value)	MuTHER consortium	17
RP11-3B7.1	ENST00000440528					
KLHDC8B	ENST00000332780	ILMN_1695246	blood	2.66×10-5 (p-value)	Westra et al.	10
KLHDC8B	ENST00000462582					
FAM212A	ENST00000333323	ILMN_1690454	blood	3.10×10-6 (p-value)	Westra et al.	2
USP4	ENST00000351842	ILMN_1798712	monocyte	1.65×10-13 (p-value)	Zeller et al.	7
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
CISH	?	ENSG00000114737	blood	2.90×10-2 (q-value)	SeeQTL DB (HapMap)	9
MST1	?	ENSG00000173531	brain	4.01×10-2 (q-value)	SeeQTL DB (Myers et al.)	1
RBM6	?	ENSG00000004534	sigmoid colon	1.88×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000338841	1	ENCP00000036479	LAMB2
		ENCP00000036455	DALRD3 NDUFAF3
		ENCP00000036491	RP11-3B7.1
		ENCP00000036487	KLHDC8B
		ENCP00000036545	UBA7
		ENCP00000036459	IMPDH2
		ENCP00000036454	NDUFAF3
		ENCP00000036522	APEH
		ENCP00000036544	UBA7
		ENCP00000036463	QRICH1
		ENCP00000036525	MST1
		ENCP00000036498	USP4
		ENCP00000036434	ARIH2
		ENCP00000036429	SLC25A20
		ENCP00000036496	USP4
		ENCP00000036461	IMPDH2
		ENCP00000036458	DALRD3
		ENCP00000036546	UBA7
ENCE00000338858	1	ENCP00000036479	LAMB2
		ENCP00000036455	DALRD3 NDUFAF3
		ENCP00000036487	KLHDC8B
		ENCP00000036459	IMPDH2
		ENCP00000036454	NDUFAF3
		ENCP00000036522	APEH
		ENCP00000036544	UBA7
		ENCP00000036463	QRICH1
		ENCP00000036525	MST1
		ENCP00000036534	IP6K1
		ENCP00000036498	USP4
		ENCP00000036434	ARIH2
		ENCP00000036496	USP4
		ENCP00000036551	CAMKV
		ENCP00000036461	IMPDH2
		ENCP00000036472	QARS
		ENCP00000036458	DALRD3
		ENCP00000036546	UBA7
ENCE00000338949	1	ENCP00000036468	QARS
		ENCP00000036469	QARS
ENCE00000339059	1	ENCP00000036562	MON1A
		ENCP00000036549	TRAIP
		ENCP00000036518	BSN
		ENCP00000036509	AMT AMT
		ENCP00000036540	CDHR4
		ENCP00000036552	CAMKV
		ENCP00000036515	DAG1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363628 (promoter flanking region)	4	NHLF	DNase1, H3K27ac
		HSMMtube	DNase1
		blood (K562)	Gata2, H3K27ac, DNase1
		skin (NHDF-AD)	H3K4me1, DNase1, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	PolII, H3K4me2, H3K27ac, H3K4me1, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3, H3K9ac
		endothelium (HUVEC)	Cjun, DNase1, H3K27ac, PolII
		liver (HepG2)	H3K4me1, H3K4me2, FOXA1
		blood (GM12878)	PU1, PolII, MEF2A, H2AZ, H3K4me1, Tcf12, DNase1, Yy1, H3K4me3, Pax5, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K27ac, H3K36me3
		nervous (NH-A)	DNase1
		A549	H3K4me3
		skin (NHEK)	H3K4me1
ENSR00001363631 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, Rad21, TAF7, H3K27ac, ATF3, TAF1, H3K4me2, SP1, USF1, H3K36me3, H3K9ac, H3K4me3, H3K27me3, PolII
		HSMMtube	H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac, H3K36me3
		muscle (HSMM)	H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, Gabp, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K36me3
		blood (GM12878)	H3K9ac, H3K36me3, H3K4me2, H3K27ac, USF1, Cmyc, PolII, DNase1, ELF1, H3K79me2, H3K4me3
		lung (IMR90)	H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, DNase1, H3K36me3, H3K4me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, H3K36me3
		cervix (HeLa-S3)	DNase1, Cmyc, H3K79me2, Gabp, PolII, H3K36me3, H3K4me3, Max, TAF1, H3K9ac, Jund, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K4me2, H3K9ac, H3K4me1, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001674821 (enhancer)	1	HSMMtube	H3K79me2
		Osteobl	H3K27ac
		blood (K562)	H3K79me2
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K79me2
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K79me2
		A549	H3K36me3
ENSR00001674822 (enhancer)	1	HSMMtube	H3K79me2
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	H3K79me2
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K36me3
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2
		nervous (NH-A)	H4K20me1
		A549	H3K36me3
ENSR00001363642 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, TAF7, Rad21, H3K36me3, PolII
		HSMMtube	H3K79me2, H3K9ac, H4K20me1, H3K4me2, H3K27ac, H3K4me3, DNase1
		blood (K562)	H3K27ac, Max, H3K9ac, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, DNase1, H3K4me3

		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, DNase1, H3K4me2, H3K4me1, H4K20me1, PolII, H3K79me2, TAF1
		blood (GM12878)	PolII, MEF2A, H2AZ, DNase1, Yy1, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		lung (IMR90)	H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H2AK5ac, H3K9ac, H3K79me2, H3K14ac, DNase1, H4K8ac, H4K20me1
		nervous (NH-A)	H3K9ac, H4K20me1, H3K4me2, H3K4me3, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, H3K4me1, H3K4me2
		breast (HMEC)	H3K4me3, H3K9ac, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	Nrf1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K9ac, H4K20me1, H3K36me3, H3K4me3, DNase1, H3K4me2, H3K27ac
		endothelium (HUVEC)	Max, H3K4me2, H3K4me3, H3K9ac, PolII, DNase1, Cmyc, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001363643 (promoter flanking region)	2	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, DNase1
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K36me3
		breast (HMEC)	CTCF
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	PolII, Rad21, CTCF, H3K36me3
		blood (GM12878)	EBF1, PolII, DNase1, CTCF, Rad21, H3K27ac, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3, CTCF
ENSR00001363645 (enhancer)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	DNase1, H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	Jund, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	FOSL2, Jund, PolII, H3K4me1
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	CTCF
ENSR00001363654 (promoter flanking region)	2	cervix (HeLa-S3)	H3K27ac, H3K4me1, DNase1, CTCF, Jund
		embryonic stem cell (H1ESC)	Rad21, DNase1
		liver (HepG2)	H3K4me1
		HSMMtube	DNase1
		A549	CTCF
		muscle (HSMM)	DNase1
ENSR00001479030 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		blood (DND-41)	H3K36me3
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3, H3K27me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	EBF1, BATF, MEF2A
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001363667 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	DNase1, H3K4me1, H3K4me2, H3K27ac
		breast (HMEC)	H3K27ac
ENSR00001479032 (promoter flanking region)	5	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3, H3K27ac, H3K9ac, DNase1

		blood (K562)	DNase1, H3K27ac, H3K9ac, PolII, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me1
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3, H3K27ac, H3K4me1, H3K4me2, HNF4A, H3K9ac
		lung (IMR90)	DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K4me2, H3K27ac
		NHLF	DNase1, H3K27ac
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me2
		cervix (HeLa-S3)	H3K4me1, H3K36me3, DNase1, H3K27ac, H3K4me2, PolII, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac
		endothelium (HUVEC)	DNase1
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac, H3K36me3
ENSR00001479035 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K27ac, DNase1
		blood (DND-41)	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K27ac
ENSR00001674842 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H3K4me1
		HSMMtube	H3K27ac
ENSR00001363673 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, Nrsf, Sin3Ak20, Yy1, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H2AZ, H3K27me3, DNase1, H3K4me2, H3K4me3
		blood (K562)	H3K4me3, DNase1, Sin3Ak20, Max, H3K9ac, H2AZ, Nrsf, PolII, H3K4me2
		skin (NHDF-AD)	H3K4me3, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2
		liver (HepG2)	H3K27me3, DNase1, H3K4me3, H3K9ac, Nrsf, PolII, Sin3Ak20, TAF1, H3K4me1, H2AZ, H3K4me2
		lung (IMR90)	H3K4me3, DNase1, H3K4me2, H3K27me3
		blood (GM12878)	DNase1, Nrsf, H2AZ, Yy1, H3K4me3, H3K4me2, H3K27me3
		nervous (NH-A)	H3K4me3, H3K27me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K27me3, H3K4me3, H3K4me2
		NHLF	H3K27me3, H3K9ac, H3K4me3
		Osteobl	H3K27me3, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K27me3
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	Ini1, H3K9ac, Nrsf, H3K4me2, H3K27me3, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	PolII, H3K27me3, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, H3K27me3
ENSR00001479043 (enhancer)	2	lung (IMR90)	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHEK)	H3K4me1, DNase1
ENSR00001363687 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (K562)	DNase1, Max, MEF2A, PolII
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		cervix (HeLa-S3)	DNase1, Cjun, Jund, H3K4me1
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	DNase1
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, DNase1
ENSR00001363693 (promoter flanking region)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	DNase1, H3K4me1, FOXA1, CTCF
		lung (IMR90)	H3K27me3

		Osteobl	H3K27me3
		blood (K562)	CTCF
ENSR00001479046 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K27me3, PolII, Gabp, Sin3Ak20, H3K9ac, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, H3K4me3
		HSMMtube	H3K4me3, H2AZ, DNase1, H3K4me2, H3K27ac
		blood (K562)	H3K79me2, ELF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, Cmyc, Gabp, Ini1, Egr1, H3K27ac, Max, CTCF, H3K9ac
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H3K9ac, H3K36me3, DNase1, H3K4me2, H3K27ac, H2AZ, CTCF, H3K4me3
		liver (HepG2)	DNase1, H3K4me3, H3K36me3, Cmyc, H3K27ac, Gabp, CTCF, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H3K4me2, H3K9ac
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3, H3K27ac, DNase1
		blood (GM12878)	EBF1, PolII, H2AZ, DNase1, CTCF, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H3K4me3, H3K36me3, CTCF, H3K4me2
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, CTCF, H3K27ac, Max, H3K4me3, Gabp, PolII, Cmyc, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, Max, H3K4me3, CTCF, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC099668.1	upstream gene variant, downstream gene variant	211	ENST00000630332	?	?	3
AC099668.5	downstream gene variant	115	ENST00000563780	?	?	3
AMT	downstream gene variant, upstream gene variant	99	ENST00000465925	?	?	4
AMT	downstream gene variant, upstream gene variant	100	ENST00000273588	NM_000481.3	ENSP00000273588	4
AMT	downstream gene variant, upstream gene variant	1002	ENST00000473163	?	?	4
AMT	downstream gene variant, upstream gene variant	851	ENST00000399379	?	ENSP00000399943	4
AMT	downstream gene variant, upstream gene variant	305	ENST00000491800	?	?	4
AMT	downstream gene variant, upstream gene variant	767	ENST00000478594	?	?	5
AMT	downstream gene variant, upstream gene variant	1657	ENST00000462048	?	?	5
AMT	downstream gene variant, upstream gene variant	100	ENST00000538581	NM_001164711.1	ENSP00000443200	4
AMT	downstream gene variant, upstream gene variant	721	ENST00000476127	?	?	4
AMT	downstream gene variant, upstream gene variant	99	ENST00000427987	?	ENSP00000403821	4
AMT	downstream gene variant, upstream gene variant	851	ENST00000495436	?	?	4
AMT	downstream gene variant, upstream gene variant	1216	ENST00000430521	?	ENSP00000388068	4
AMT	downstream gene variant, upstream gene variant	100	ENST00000458307	NM_001164710.1	ENSP00000415619	4
AMT	downstream gene variant, upstream gene variant	99	ENST00000395338	NM_001164712.1	ENSP00000378747	4
AMT	downstream gene variant, upstream gene variant	100	ENST00000476226	?	?	4
AMT	downstream gene variant, upstream gene variant	1531	ENST00000485108	?	?	5
AMT	downstream gene variant, upstream gene variant	1449	ENST00000493046	?	?	5
AMT	downstream gene variant, upstream gene variant	735	ENST00000461210	?	?	5
AMT	downstream gene variant, upstream gene variant	819	ENST00000498571	?	?	4
AMT	downstream gene variant, upstream gene variant	725	ENST00000487589	?	?	5

AMT	downstream gene variant, upstream gene variant	1287	ENST00000480957	?	?	5
AMT	downstream gene variant, upstream gene variant	1641	ENST00000476828	?	?	5
APEH	upstream gene variant	968	ENST00000449966	?	ENSP00000414369	1
APEH	upstream gene variant, downstream gene variant	1314	ENST00000446089	?	ENSP00000410149	4
APEH	upstream gene variant, downstream gene variant	440	ENST00000483715	?	?	2
APEH	upstream gene variant, downstream gene variant	286	ENST00000462439	?	?	2
APEH	upstream gene variant, downstream gene variant	1154	ENST00000469362	?	ENSP00000438180	4
APEH	upstream gene variant, downstream gene variant	1339	ENST00000447436	?	ENSP00000401871	4
APEH	upstream gene variant, downstream gene variant	276	ENST00000491799	?	?	2
APEH	upstream gene variant	1333	ENST00000463995	?	?	1
APEH	upstream gene variant, downstream gene variant	830	ENST00000447541	?	ENSP00000404353	3
APEH	upstream gene variant, downstream gene variant	757	ENST00000438011	?	ENSP00000415862	3
APEH	upstream gene variant, downstream gene variant	1806	ENST00000482301	?	?	5
APEH	upstream gene variant, downstream gene variant	1321	ENST00000442186	?	ENSP00000402365	2
APEH	upstream gene variant	1440	ENST00000457042	?	ENSP00000410366	1
APEH	upstream gene variant, downstream gene variant	136	ENST00000296456	NM_001640.3	ENSP00000296456	3
APEH	upstream gene variant, downstream gene variant	560	ENST00000463616	?	?	2
APEH	upstream gene variant, downstream gene variant	305	ENST00000480772	?	?	3
BSN	upstream gene variant, downstream gene variant	1152	ENST00000296452	NM_003458.3	ENSP00000296452	3
BSN	upstream gene variant, downstream gene variant	428	ENST00000467456	?	?	4
BSN-AS1	downstream gene variant, upstream gene variant	2582	ENST00000442384	?	?	4
DAG1	upstream gene variant	3617	ENST00000452060	?	ENSP00000410145	1
DAG1	upstream gene variant, downstream gene variant	1856	ENST00000479935	?	?	4
DAG1	upstream gene variant	3627	ENST00000466701	?	?	1
DAG1	upstream gene variant	2051	ENST00000469139	?	?	1
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000545947	NM_001177634.2	ENSP00000442600	3
DAG1	upstream gene variant	3692	ENST00000496474	?	?	1
DAG1	upstream gene variant, downstream gene variant	1556	ENST00000461492	?	?	3
DAG1	upstream gene variant, downstream gene variant	1760	ENST00000308775	NM_001177636.2, NM_004393.5, NM_001177639.2, NM_001177637.2	ENSP00000312435	3
DAG1	upstream gene variant	3657	ENST00000430636	?	ENSP00000401805	1
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000539901	NM_001177644.2	ENSP00000439334	3
DAG1	upstream gene variant, downstream gene variant	1936	ENST00000421560	?	ENSP00000412067	3
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000515359	NM_001177643.2	ENSP00000440705	3
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000538711	NM_001177640.2, NM_001177642.2, NM_001177635.2	ENSP00000438421	3
DAG1	upstream gene variant	3615	ENST00000452317	?	ENSP00000387859	1
DAG1	upstream gene variant	3615	ENST00000435508	?	ENSP00000415321	1
DAG1	upstream gene variant, downstream gene variant	248	ENST00000461987	?	?	4

DAG1	upstream gene variant	3654	ENST00000419218	?	ENSP00000402443	1
DAG1	upstream gene variant, downstream gene variant	1903	ENST00000431960	?	ENSP00000388833	3
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000541308	NM_001177638.2, NM_001177641.2, NM_001165928.3	ENSP00000440590	3
DAG1	upstream gene variant	3638	ENST00000428779	?	ENSP00000401382	1
DAG1	upstream gene variant	667	ENST00000475424	?	?	2
DAG1	upstream gene variant	3474	ENST00000418588	?	ENSP00000405859	1
GPX1	downstream gene variant, upstream gene variant	207	ENST00000496791	?	?	5
GPX1	downstream gene variant, upstream gene variant	327	ENST00000419783	NM_000581.2	ENSP00000407375	5
GPX1	downstream gene variant, upstream gene variant	41	ENST00000419349	NM_201397.1	ENSP00000391316	6
GPX1	downstream gene variant, upstream gene variant	569	ENST00000620890	?	ENSP00000478837	5
MST1	downstream gene variant	2675	ENST00000481055	?	?	3
MST1	downstream gene variant	496	ENST00000497359	?	?	3
MST1	downstream gene variant, upstream gene variant	2377	ENST00000494828	?	?	4
MST1	downstream gene variant, upstream gene variant	1803	ENST00000492370	?	?	4
MST1	downstream gene variant	226	ENST00000484144	?	?	2
MST1	downstream gene variant, upstream gene variant	3448	ENST00000491943	?	?	4
MST1	downstream gene variant	1651	ENST00000448220	?	ENSP00000394756	1
MST1	downstream gene variant	1042	ENST00000480268	?	?	3
MST1	downstream gene variant	1680	ENST00000492329	?	?	1
MST1	downstream gene variant	1753	ENST00000493836	?	?	1
MST1	downstream gene variant	1380	ENST00000489007	?	?	3
MST1	downstream gene variant	373	ENST00000484269	?	?	3
MST1	downstream gene variant, upstream gene variant	1520	ENST00000484673	?	?	4
MST1	downstream gene variant	1318	ENST00000468847	?	?	3
MST1	downstream gene variant, upstream gene variant	2603	ENST00000490966	?	?	4
MST1	downstream gene variant, upstream gene variant	1680	ENST00000488350	?	?	3
MST1	downstream gene variant	373	ENST00000481930	?	?	3
MST1	downstream gene variant, upstream gene variant	1651	ENST00000449682	NM_020998.3	ENSP00000414287	2
MST1	downstream gene variant	479	ENST00000498021	?	?	3
MST1	downstream gene variant	1680	ENST00000479115	?	?	1
MST1	downstream gene variant	1117	ENST00000494809	?	?	3
NICN1	downstream gene variant, upstream gene variant	3941	ENST00000494057	?	?	2
NICN1	downstream gene variant	4376	ENST00000461015	?	?	1
NICN1	downstream gene variant, upstream gene variant	3947	ENST00000422593	?	?	2
NICN1	downstream gene variant, upstream gene variant	2071	ENST00000423832	?	ENSP00000413115	3
NICN1	downstream gene variant, upstream gene variant	2025	ENST00000615713	?	ENSP00000478287	3
NICN1	downstream gene variant, upstream gene variant	2024	ENST00000273598	NM_032316.3	ENSP00000273598	3

NICN1	downstream gene variant, upstream gene variant	3255	ENST00000436744	?	ENSP00000402335	2
NICN1	downstream gene variant, upstream gene variant	3153	ENST00000497742	?	?	2
RHOA	downstream gene variant, upstream gene variant	691	ENST00000445425	?	ENSP00000408402	6
RHOA	downstream gene variant, upstream gene variant	965	ENST00000454011	?	ENSP00000394483	6
RHOA	downstream gene variant, upstream gene variant	218	ENST00000418115	NM_001664.2	ENSP00000400175	6
RHOA	upstream gene variant	18	ENST00000265538	?	?	4
RHOA	downstream gene variant, upstream gene variant	919	ENST00000431929	?	?	6
RHOA	downstream gene variant, upstream gene variant	1046	ENST00000422781	?	ENSP00000413587	6
RHOA-IT1	downstream gene variant, upstream gene variant	725	ENST00000428083	?	?	5
RNA5SP130	downstream gene variant	453	ENST00000364431	?	?	1
TCTA	downstream gene variant	2975	ENST00000488385	?	?	3
TCTA	downstream gene variant	959	ENST00000493381	?	?	4
TCTA	downstream gene variant	3268	ENST00000497786	?	?	3
TCTA	downstream gene variant	204	ENST00000273590	NM_022171.2	ENSP00000273590	4
TCTA	downstream gene variant	3129	ENST00000482193	?	?	3
TCTA	downstream gene variant	1136	ENST00000487432	?	?	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
AMT	ENST00000430521	?	ENSP00000388068	R	cgA/cgG	1
AMT	ENST00000427987	?	ENSP00000403821	R	cgA/cgG	1
AMT	ENST00000538581	NM_001164711.1	ENSP00000443200	R	cgA/cgG	1
AMT	ENST00000458307	NM_001164710.1	ENSP00000415619	R	cgA/cgG	1
AMT	ENST00000273588	NM_000481.3	ENSP00000273588	R	cgA/cgG	1
AMT	ENST00000395338	NM_001164712.1	ENSP00000378747	R	cgA/cgG	1
APEH	ENST00000296456	NM_001640.3	ENSP00000296456	Y	taC/taT	1
APEH	ENST00000449966	?	ENSP00000414369	Y	taC/taT	1
APEH	ENST00000457042	?	ENSP00000410366	Y	taC/taT	1
APEH	ENST00000438011	?	ENSP00000415862	Y	taC/taT	1
APEH	ENST00000442186	?	ENSP00000402365	Y	taC/taT	1
BSN	ENST00000296452	NM_003458.3	ENSP00000296452	2	2	2
DAG1	ENST00000541308	NM_001177638.2, NM_001177641.2, NM_001165928.3	ENSP00000440590	H	caT/caC	1
DAG1	ENST00000545947	NM_001177634.2	ENSP00000442600	H	caT/caC	1
DAG1	ENST00000308775	NM_001177636.2, NM_004393.5, NM_001177639.2, NM_001177637.2	ENSP00000312435	H	caT/caC	1
DAG1	ENST00000538711	NM_001177640.2, NM_001177642.2, NM_001177635.2	ENSP00000438421	H	caT/caC	1
DAG1		NM_001177644.2	ENSP00000439334	H	caT/caC	1

	ENST00000539901					
DAG1	ENST00000515359	NM_001177643.2	ENSP00000440705	H	caT/caC	1
MST1	ENST00000449682	NM_020998.3	ENSP00000414287	R	cgT/cgG	1
MST1	ENST00000448220	?	ENSP00000394756	R	cgT/cgG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AMT	ENST00000399379	?	ENSP00000399943	3
AMT	ENST00000430521	?	ENSP00000388068	3
AMT	ENST00000473163	?	?	2
AMT	ENST00000427987	?	ENSP00000403821	3
AMT	ENST00000480957	?	?	1
AMT	ENST00000462048	?	?	1
AMT	ENST00000487589	?	?	1
AMT	ENST00000461210	?	?	1
AMT	ENST00000493046	?	?	1
AMT	ENST00000485108	?	?	1
AMT	ENST00000476828	?	?	1
AMT	ENST00000491800	?	?	1
AMT	ENST00000538581	NM_001164711.1	ENSP00000443200	3
AMT	ENST00000458307	NM_001164710.1	ENSP00000415619	3
AMT	ENST00000476127	?	?	2
AMT	ENST00000476226	?	?	3
AMT	ENST00000478594	?	?	1
AMT	ENST00000495436	?	?	3
AMT	ENST00000273588	NM_000481.3	ENSP00000273588	3
AMT	ENST00000465925	?	?	3
AMT	ENST00000395338	NM_001164712.1	ENSP00000378747	3
APEH	ENST00000296456	NM_001640.3	ENSP00000296456	2
APEH	ENST00000463995	?	?	1
APEH	ENST00000449966	?	ENSP00000414369	1
APEH	ENST00000447436	?	ENSP00000401871	1
APEH	ENST00000457042	?	ENSP00000410366	1
APEH	ENST00000491799	?	?	1
APEH	ENST00000446089	?	ENSP00000410149	1
APEH	ENST00000483715	?	?	1
APEH	ENST00000438011	?	ENSP00000415862	2
APEH	ENST00000442186	?	ENSP00000402365	1
APEH	ENST00000462439	?	?	1
APEH	ENST00000469362	?	ENSP00000438180	1
APEH	ENST00000463616	?	?	1
BSN	ENST00000296452	NM_003458.3	ENSP00000296452	30

BSN	ENST00000467456	?	?	3
BSN-AS1	ENST00000442384	?	?	2
BSN-AS2	ENST00000421598	?	?	1
BSN-AS2	ENST00000433882	?	?	1
DAG1	ENST00000421560	?	ENSP00000412067	21
DAG1	ENST00000541308	NM_001177638.2, NM_001177641.2, NM_001165928.3	ENSP00000440590	21
DAG1	ENST00000469139	?	?	17
DAG1	ENST00000461492	?	?	21
DAG1	ENST00000496474	?	?	17
DAG1	ENST00000545947	NM_001177634.2	ENSP00000442600	21
DAG1	ENST00000308775	NM_001177636.2, NM_004393.5, NM_001177639.2, NM_001177637.2	ENSP00000312435	21
DAG1	ENST00000430636	?	ENSP00000401805	17
DAG1	ENST00000466701	?	?	17
DAG1	ENST00000475424	?	?	10
DAG1	ENST00000538711	NM_001177640.2, NM_001177642.2, NM_001177635.2	ENSP00000438421	21
DAG1	ENST00000419218	?	ENSP00000402443	17
DAG1	ENST00000452060	?	ENSP00000410145	17
DAG1	ENST00000539901	NM_001177644.2	ENSP00000439334	21
DAG1	ENST00000515359	NM_001177643.2	ENSP00000440705	21
DAG1	ENST00000431960	?	ENSP00000388833	21
DAG1	ENST00000461987	?	?	4
DAG1	ENST00000435508	?	ENSP00000415321	17
DAG1	ENST00000452317	?	ENSP00000387859	17
DAG1	ENST00000479935	?	?	4
DAG1	ENST00000428779	?	ENSP00000401382	17
DAG1	ENST00000418588	?	ENSP00000405859	17
NICN1	ENST00000423832	?	ENSP00000413115	1
NICN1	ENST00000494057	?	?	1
NICN1	ENST00000422593	?	?	1
NICN1	ENST00000497742	?	?	1
NICN1	ENST00000273598	NM_032316.3	ENSP00000273598	1
NICN1	ENST00000436744	?	ENSP00000402335	1
NICN1	ENST00000461015	?	?	1
NICN1	ENST00000615713	?	ENSP00000478287	1
RHOA	ENST00000445425	?	ENSP00000408402	15
RHOA	ENST00000454011	?	ENSP00000394483	16
RHOA	ENST00000422781	?	ENSP00000413587	16
RHOA	ENST00000418115	NM_001664.2	ENSP00000400175	16
RHOA	ENST00000431929	?	?	11
RNF123	ENST00000486102	?	?	2
RNF123	ENST00000454491	?	ENSP00000408490	2

RNF123	ENST00000457726	?	ENSP00000394369	2
RNF123	ENST00000443204	?	ENSP00000390856	2
RNF123	ENST00000487805	?	?	2
RNF123	ENST00000629802	?	ENSP00000487104	2
RNF123	ENST00000327697	NM_022064.3	ENSP00000328287	2
RNF123	ENST00000432042	?	ENSP00000392443	2
TCTA	ENST00000488385	?	?	1
TCTA	ENST00000493381	?	?	1
TCTA	ENST00000273590	NM_022171.2	ENSP00000273590	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
AMT	ENST00000399379	?	ENSP00000399943	1
APEH	ENST00000447436	?	ENSP00000401871	1
APEH	ENST00000446089	?	ENSP00000410149	1
APEH	ENST00000447541	?	ENSP00000404353	1
BSN	ENST00000296452	NM_003458.3	ENSP00000296452	1
DAG1	ENST00000541308	NM_001177638.2, NM_001177641.2, NM_001165928.3	ENSP00000440590	1
DAG1	ENST00000545947	NM_001177634.2	ENSP00000442600	1
DAG1	ENST00000308775	NM_001177636.2, NM_004393.5, NM_001177639.2, NM_001177637.2	ENSP00000312435	1
DAG1	ENST00000538711	NM_001177640.2, NM_001177642.2, NM_001177635.2	ENSP00000438421	1
DAG1	ENST00000539901	NM_001177644.2	ENSP00000439334	1
DAG1	ENST00000515359	NM_001177643.2	ENSP00000440705	1
GPX1	ENST00000419349	NM_201397.1	ENSP00000391316	1
TCTA	ENST00000273590	NM_022171.2	ENSP00000273590	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPX1	ENST00000620890	?	ENSP00000478837	1
GPX1	ENST00000419783	NM_000581.2	ENSP00000407375	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AMT	ENST00000473163	?	1
AMT	ENST00000476127	?	1
AMT	ENST00000476226	?	2
AMT	ENST00000465925	?	2
APEH	ENST00000482301	?	1
APEH	ENST00000463995	?	1
GPX1	ENST00000496791	?	1
MST1	ENST00000484144	?	1
MST1	ENST00000493836	?	2
MST1	ENST00000479115	?	2
MST1	ENST00000488350	?	2

MST1	ENST00000492329	?	2
TCTA	ENST00000487432	?	1
TCTA	ENST00000497786	?	1
TCTA	ENST00000482193	?	1

Locus 4 (sentinel SNP rs9858542 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 5 (rs17415296)

SNiPAcard

Block annotations

Block info

genomic range	chr1:66,062,325-66,170,362
block size	108,038 bp
variant count	191 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.388 [-3.75 – 2.308]	gene(s) hit or close-by	LEPR
phastCons	μ = 0.082 [0 – 0.971]	eQTL gene(s)	–
GERP++	μ = -0.270 [-5.29 – 4.15]	potentially regulated gene(s)	RP11-397C12.1
CADD score	μ = 3.009 [0.046 – 16.78]	disease gene(s)	LEPR

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Fasting plasma glucose (childhood)	$<5.00\times10^{-8}$	GWAS Catalog	25223902	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Leptin receptor polymorphism	benign	ClinVar	RCV000009050.1	1
?	HGMD curated	HGMD	CM025915	1
LEPTIN RECEPTOR POLYMORPHISM	OMIM curated	OMIM	MIM:601007	1

Disease gene annotation

gene	trait	source DB	source entry/link
LEPR	LEPTIN RECEPTOR DEFICIENCY	OMIM	MIM:614963
LEPR	Obesity due to leptin receptor gene deficiency	OrphaNet	OrphaNet:179494

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
LEPR	missense variant	ENST00000349533	NM_002303.5	ENSP00000330393	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000371058	?	ENSP00000360097	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000616738	NM_001198689.1	ENSP00000483390	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000371060	NM_001003679.3	ENSP00000360099	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000344610	NM_001198688.1	ENSP00000340884	N/K	aaC/aaG	?	?	1
LEPR	missense variant	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	N/K	aaC/aaG	?	?	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000023340	1	ENCP00000002897	RP11-397C12.1
ENCE00000023461	1	ENCP00000002897	RP11-397C12.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001521219 (open chromatin region)	2	blood (K562)	H3K36me3
		muscle (HSMM)	DNase1
ENSR00001580863 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF
		lung (IMR90)	CTCF
ENSR00001521220 (CTCF binding site)	2	blood (K562)	H3K36me3
ENSR00001580864 (CTCF binding site)	2	cervix (HeLa-S3)	H3K27me3
		A549	H3K27me3
ENSR00001521221 (enhancer)	2	cervix (HeLa-S3)	H3K27me3
		A549	H3K27me3
ENSR00001521223 (CTCF binding site)	7	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	Rad21
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LEPR	downstream gene variant	2903	ENST00000406510	?	ENSP00000384025	6
LEPR	upstream gene variant, downstream gene variant	98	ENST00000471762	?	?	8
LEPR	downstream gene variant	183	ENST00000616738	NM_001198689.1	ENSP00000483390	7
LEPR	downstream gene variant	649	ENST00000344610	NM_001198688.1	ENSP00000340884	4
LEPR	downstream gene variant	1	ENST00000462765	?	?	4
LEPR	downstream gene variant	649	ENST00000371058	?	ENSP00000360097	4
LEPR	downstream gene variant	164	ENST00000371060	NM_001003679.3	ENSP00000360099	7
LEPR	downstream gene variant	820	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	5
LEPR	downstream gene variant	1099	ENST00000349533	NM_002303.5	ENSP00000330393	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000371058	?	ENSP00000360097	27
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	37
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	36
LEPR	ENST00000616738	NM_001198689.1	ENSP00000483390	36
LEPR	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	31
LEPR	ENST00000462765	?	?	14
LEPR	ENST00000406510	?	ENSP00000384025	38

LEPR	ENST00000344610	NM_001198688.1	ENSP00000340884	27
LEPR	ENST00000471762	?	?	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	8
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	2
LEPR	ENST00000616738	NM_001198689.1	ENSP00000483390	2

Locus 6 (rs1024948)

SNiPAcard

Block annotations

Block info

genomic range	chr3:190,344,128-190,354,410
block size	10,283 bp
variant count	24 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.442 [-4.686 – 1.233]	**gene(s) hit or close-by**	GCNT1P3, IL1RAP
phastCons	μ = 0.038 [0 – 0.528]	**eQTL gene(s)**	–
GERP++	μ = -0.108 [-6.06 – 2.74]	**potentially regulated gene(s)**	–
CADD score	μ = 3.837 [0.201 – 9.854]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Body height	3.44×10^{-5}	dbGaP	pha000341 dbGaP	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001487397 (CTCF binding site)	1	cervix (HeLa-S3)	H3K36me3
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GCNT1P3	downstream gene variant	1170	ENST00000450607	?	?	16
IL1RAP	downstream gene variant	224	ENST00000422485	NM_134470.3	ENSP00000409352	6
IL1RAP	downstream gene variant	157	ENST00000412080	?	ENSP00000414102	8
IL1RAP	downstream gene variant	224	ENST00000422940	NM_001167930.1	ENSP00000387371	6
IL1RAP	downstream gene variant	265	ENST00000434491	?	ENSP00000391899	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RAP	ENST00000317757	NM_001167931.1	ENSP00000314807	24
IL1RAP	ENST00000413869	?	ENSP00000416296	24
IL1RAP	ENST00000439062	NM_001167928.1	ENSP00000401132	24
IL1RAP	ENST00000447382	NM_002182.3	ENSP00000390541	24
IL1RAP	ENST00000434491	?	ENSP00000391899	17
IL1RAP	ENST00000412080	?	ENSP00000414102	14
IL1RAP	ENST00000422940	NM_001167930.1	ENSP00000387371	17

IL1RAP	ENST00000072516	NM_001167929.1	ENSP00000072516	24
IL1RAP	ENST00000422485	NM_134470.3	ENSP00000409352	17
IL1RAP	ENST00000342550	?	ENSP00000345829	24
IL1RAP	ENST00000412504	?	ENSP00000412053	24
IL1RAP	ENST00000443369	?	ENSP00000408893	24

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RAP	ENST00000434491	?	ENSP00000391899	4
IL1RAP	ENST00000412080	?	ENSP00000414102	1
IL1RAP	ENST00000422940	NM_001167930.1	ENSP00000387371	4
IL1RAP	ENST00000422485	NM_134470.3	ENSP00000409352	4

Locus 7 (rs12459419)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,727,962-51,737,991
block size	10,030 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.887 [-2.575 – 0.29]	gene(s) hit or close-by	CD33
phastCons	μ = 0.145 [0 – 0.744]	eQTL gene(s)	CD33, SIGLEC22P, SIGLECL1
GERP++	μ = -1.199 [-5.72 – 0.919]	potentially regulated gene(s)	SIGLEC12
CADD score	μ = 4.703 [0.881 – 14.88]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Alzheimer's disease (late onset)	$<3.00\times10^{-6}$	GWAS Catalog	24162737	1
Alzheimer's disease (late onset)	$<2.00\times10^{-9}$	GWAS Catalog	21460841	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CD33	missense variant	ENST00000262262	NM_001772.3	ENSP00000262262	V/A	gTc/gCc	?	?	1
CD33	missense variant	ENST00000391796	NM_001177608.1	ENSP00000375673	V/A	gTc/gCc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD33	ENST00000262262	ILMN_1747622	blood	1.88×10^{-7} (p-value)	MuTHER consortium	3
CD33	ENST00000600557					
CD33	ENST00000601785					
CD33	ENST00000421133					
CD33	ENST00000598473					
SIGLEC22P	?	ENSG00000268849	lung	1.91×10^{-7} (p-value)	GTEx Portal V6	6
SIGLECL1	?	ENSG00000179213	testis	1.57×10^{-8} (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000232891	1	ENCP00000024645	SIGLEC12

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000217710 (promoter)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K4me3, H3K4me2, PolII, H2AZ, Gabp, H3K9ac, PU1, DNase1
		cervix (HeLa-S3)	PolII, H3K9ac, Gabp, H3K27me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		liver (HepG2)	H3K27me3, Gabp
		blood (GM12878)	Gabp
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CD33	upstream gene variant	392	ENST00000262262	NM_001772.3	ENSP00000262262	1
CD33	upstream gene variant	358	ENST00000436584	?	ENSP00000403331	1
CD33	upstream gene variant	358	ENST00000421133	NM_001082618.1	ENSP00000410126	1
CD33	upstream gene variant	425	ENST00000601785	?	?	1
CD33	upstream gene variant	745	ENST00000598473	?	?	2
CD33	upstream gene variant	392	ENST00000391796	NM_001177608.1	ENSP00000375673	1
CD33	upstream gene variant	4531	ENST00000600557	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CD33	ENST00000436584	?	ENSP00000403331	5
CD33	ENST00000391796	NM_001177608.1	ENSP00000375673	4
CD33	ENST00000601785	?	?	5
CD33	ENST00000262262	NM_001772.3	ENSP00000262262	4
CD33	ENST00000421133	NM_001082618.1	ENSP00000410126	5
CD33	ENST00000598473	?	?	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CD33	ENST00000601785	?	1

Locus 8 (rs1926447)

SNiPAcard

Block annotations

Block info

genomic range	chr13:46,514,506-46,629,995
block size	115,490 bp
variant count	72 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.652 [-3.814 – 2.601]	gene(s) hit or close-by	CPB2, CPB2-AS1, ZC3H13
phastCons	μ = 0.045 [0 – 0.997]	eQTL gene(s)	LCP1
GERP++	μ = -0.574 [-7.55 – 3]	potentially regulated gene(s)	–
CADD score	μ = 3.565 [0.018 – 11.48]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM013697	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CPB2	missense variant	ENST00000439329	NM_001278541.1	ENSP00000400714	I/T	aTt/aCt	tolerated	benign	1
CPB2	missense variant	ENST00000181383	NM_001872.4	ENSP00000181383	I/T	aTt/aCt	tolerated	benign	1
ZC3H13	missense variant	ENST00000242848	?	ENSP00000242848	E/D	gaA/gaT	?	unknown	1
ZC3H13	missense variant	ENST00000282007	NM_015070.4	ENSP00000282007	E/D	gaA/gaT	?	unknown	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LCP1	ENST00000323076	ILMN_1662932	blood	5.07×10^{-6} (p-value)	Westra et al.	31
LCP1	ENST00000398576					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001509491 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3

	monocytes (Monocytes-CD14+)	H3K36me3
	liver (HepG2)	H3K36me3
	lung (IMR90)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ZC3H13	downstream gene variant, upstream gene variant	594	ENST00000464597	?	ENSP00000480658	13
ZC3H13	downstream gene variant, upstream gene variant	961	ENST00000282007	NM_015070.4	ENSP00000282007	7
ZC3H13	downstream gene variant, upstream gene variant	721	ENST00000242848	?	ENSP00000242848	4
ZC3H13	downstream gene variant, upstream gene variant	981	ENST00000470308	?	?	11
ZC3H13	downstream gene variant, upstream gene variant	2405	ENST00000428921	?	ENSP00000416238	7

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	H	caT/caC	1
ZC3H13	ENST00000242848	?	ENSP00000242848	H	caT/caC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	2
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	2
CPB2-AS1	ENST00000606991	?	?	3
CPB2-AS1	ENST00000606243	?	?	3
CPB2-AS1	ENST00000606351	?	?	3
CPB2-AS1	ENST00000415033	?	?	3
ZC3H13	ENST00000464597	?	ENSP00000480658	9
ZC3H13	ENST00000470308	?	?	13
ZC3H13	ENST00000428921	?	ENSP00000416238	3
ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	59
ZC3H13	ENST00000242848	?	ENSP00000242848	63

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ZC3H13	ENST00000470308	?	1

Locus 9 (rs3806701)

SNiPAcard

Block annotations

Block info

genomic range	chr3:46,596,264-46,631,058
block size	34,795 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.261 [-1.057 – 0.212]	gene(s) hit or close-by	AC104304.1, LRRC2, LRRC2-AS1, TDGF1
phastCons	μ = 0.047 [0 – 0.328]	eQTL gene(s)	ALS2CL, LRRC2, LRRC2-AS1, TDGF1
GERP++	μ = -0.249 [-4.4 – 1.74]	potentially regulated gene(s)	AC104304.1, CCR2, LRRC2, LTF, PRSS44, PRSS50
CADD score	μ = 4.291 [1.295 – 8.84]	disease gene(s)	TDGF1, LTF, CCR2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TDGF1	TERATOCARCINOMA-DERIVED GROWTH FACTOR 1	OMIM	MIM:187395
LTF	SPECIFIC GRANULE DEFICIENCY	OMIM	MIM:245480
CCR2	HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, SUSCEPTIBILITY TO	OMIM	MIM:609423
TDGF1	Semilobar holoprosencephaly	OrphaNet	OrphaNet:220386
TDGF1	Alobar holoprosencephaly	OrphaNet	OrphaNet:93925
TDGF1	Microform holoprosencephaly	OrphaNet	OrphaNet:280200
TDGF1	Midline interhemispheric variant of holoprosencephaly	OrphaNet	OrphaNet:93926
TDGF1	Septopreoptic holoprosencephaly	OrphaNet	OrphaNet:280195
TDGF1	Lobar holoprosencephaly	OrphaNet	OrphaNet:93924

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TDGF1	?	ENSG00000241186	atrial appendage	6.64×10^{-21} (p-value)	GTEx Portal V6	9
ALS2CL	?	ENSG00000178038	transformed fibroblasts	9.66×10^{-7} (p-value)	GTEx Portal V6	5
LRRC2	?	ENSG00000163827	transformed fibroblasts	7.45×10^{-33} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	breast	1.41×10^{-27} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	skeletal muscle	1.27×10^{-57} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	putamen	2.37×10^{-10} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	unexposed skin	1.15×10^{-20} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	sun exposed skin	1.16×10^{-19} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	ovary	5.21×10^{-18} (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	ovary	2.35×10^{-6} (p-value)	GTEx Portal V6	1
TDGF1	?	ENSG00000241186	left ventricle	2.50×10^{-37} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	subcutaneous adipocytes	6.28×10^{-55} (p-value)	GTEx Portal V6	9

TDGF1	?	ENSG00000241186	caudate basal ganglia	2.40×10-12 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	visceral adipocytes	2.25×10-32 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	cortex	5.06×10-15 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	tibial nerve	8.12×10-48 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	pituitary	2.18×10-9 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	anterior cingulate cortex	1.80×10-10 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	esophagus mucosa	2.71×10-27 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	testis	4.56×10-17 (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	testis	1.17×10-7 (p-value)	GTEx Portal V6	9
LRRC2-AS1	?	ENSG00000268324	testis	1.42×10-5 (p-value)	GTEx Portal V6	4
TDGF1	?	ENSG00000241186	hypothalamus	1.44×10-10 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	frontal cortex	7.78×10-15 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	uterus	3.21×10-13 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	muscularis mucosae	1.23×10-40 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	lung	1.26×10-27 (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	tibial artery	1.66×10-8 (p-value)	GTEx Portal V6	7
TDGF1	?	ENSG00000241186	prostate	2.11×10-13 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	transverse colon	3.56×10-7 (p-value)	GTEx Portal V6	5
TDGF1	?	ENSG00000241186	terminal ileum	2.23×10-7 (p-value)	GTEx Portal V6	5
TDGF1	?	ENSG00000241186	nucleus accumbens	1.76×10-12 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	spleen	2.86×10-21 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	coronary artery	1.44×10-14 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	sigmoid colon	1.13×10-25 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	vagina	2.24×10-9 (p-value)	GTEx Portal V6	7
TDGF1	?	ENSG00000241186	gastroesophageal junction	2.89×10-25 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	adrenal gland	1.93×10-27 (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	adrenal gland	8.32×10-27 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	cerebellar hemisphere	5.18×10-8 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	cerebellum	8.08×10-9 (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	cerebellum	9.29×10-7 (p-value)	GTEx Portal V6	5
TDGF1	?	ENSG00000241186	hippocampus	2.99×10-9 (p-value)	GTEx Portal V6	9

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000337772	1	ENCP00000036261	LRRC2
		ENCP00000036273	PRSS50
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036262	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036258	LTF

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363001 (TF binding site)	3	embryonic stem cell (H1ESC)	Rad21, TAF7, H3K27ac, HDAC2, H3K4me2, SP1, H3K4me3, DNase1, PolII, H3K27me3, H3K36me3
		endothelium (HUVEC)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001363008 (TF binding site)	2	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC104304.1	upstream gene variant	4217	ENST00000583198	?	?	1
LRRC2-AS1	upstream gene variant	2624	ENST00000599511	?	?	1
LRRC2-AS1	upstream gene variant	2630	ENST00000614743	?	?	1
TDGF1	upstream gene variant	1394	ENST00000459867	?	?	4
TDGF1	upstream gene variant	3402	ENST00000505797	?	?	2
TDGF1	upstream gene variant	1519	ENST00000493282	?	?	3
TDGF1	upstream gene variant	17	ENST00000471721	?	?	4
TDGF1	upstream gene variant, downstream gene variant	1008	ENST00000296145	NM_003212.3	ENSP00000296145	5
TDGF1	downstream gene variant	4493	ENST00000542931	NM_001174136.1	ENSP00000446375	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LRRC2	ENST00000296144	?	ENSP00000296144	5
LRRC2	ENST00000496388	?	?	1
LRRC2	ENST00000395905	NM_024512.4	ENSP00000379241	1
LRRC2	ENST00000469912	?	?	1
TDGF1	ENST00000542931	NM_001174136.1	ENSP00000446375	4
TDGF1	ENST00000505797	?	?	4

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TDGF1	ENST00000296145	NM_003212.3	ENSP00000296145	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

TDGF1	ENST00000493282	?	1

Locus 9 (sentinel SNP rs3806701 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 10 (rs17413015)

SNiPAcard

Block annotations

Block info

genomic range	chr1:161,640,787-161,646,824
block size	6,038 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.191 [-0.658 – 1.684]	gene(s) hit or close-by	FCGR2B, RP11-25K21.1
phastCons	μ = 0.124 [0 – 0.986]	eQTL gene(s)	FCGR2B, FCGR2C, FCGR3B, FCRLB
GERP++	μ = -0.088 [-2.64 – 2.31]	potentially regulated gene(s)	–
CADD score	μ = 5.474 [0.031 – 14.51]	disease gene(s)	FCGR2B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
FCGR2B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
FCGR2B	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCGR3B	?	ENSG00000162747	sun exposed skin	2.06×10^{-6} (p-value)	GTEx Portal V6	4
FCGR3B	?	ENSG00000162747	unexposed skin	4.97×10^{-7} (p-value)	GTEx Portal V6	4
FCGR2C	?	ENSG00000244682	blood	1.06×10^{-6} (p-value)	GTEx Portal V6	4
FCGR2B	ENST00000403078	ILMN_1660027	blood	5.47×10^{-7} (p-value)	Westra et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000403078	ILMN_2382403	monocyte	1.04×10^{-4} (p-value)	Fairfax et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCRLB	ENST00000367948	ILMN_1782015	monocyte	5.79×10^{-4} (p-value)	Fairfax et al.	1
FCRLB	ENST00000495397		b-cell	9.59×10^{-4} (p-value)	Fairfax et al.	1
FCRLB	ENST00000367944					
FCRLB	ENST00000336830					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001525304 (enhancer)	1	embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H2AZ
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR2B	downstream gene variant	78	ENST00000485778	?	?	5
RP11-25K21.1	downstream gene variant, upstream gene variant	109	ENST00000626340	?	?	6
RP11-25K21.1	downstream gene variant, upstream gene variant	49	ENST00000453111	?	?	6

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
FCGR2B	ENST00000367961	NM_001190828.1	ENSP00000356938	T	acA/acG	1
FCGR2B	ENST00000367960	?	ENSP00000356937	T	acA/acG	1
FCGR2B	ENST00000236937	NM_001002274.2, NM_001002273.2	ENSP00000236937	T	acA/acG	1
FCGR2B	ENST00000403078	?	ENSP00000386038	T	acA/acG	1
FCGR2B	ENST00000428605	?	ENSP00000404329	T	acA/acG	1
FCGR2B	ENST00000358671	NM_004001.4, NM_001002275.2	ENSP00000351497	T	acA/acG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2B	ENST00000367961	NM_001190828.1	ENSP00000356938	7
FCGR2B	ENST00000367960	?	ENSP00000356937	7
FCGR2B	ENST00000480308	?	?	7
FCGR2B	ENST00000236937	NM_001002274.2, NM_001002273.2	ENSP00000236937	7
FCGR2B	ENST00000403078	?	ENSP00000386038	7
FCGR2B	ENST00000428605	?	ENSP00000404329	7
FCGR2B	ENST00000358671	NM_004001.4, NM_001002275.2	ENSP00000351497	7
FCGR2B	ENST00000485778	?	?	2
RP11-25K21.1	ENST00000626340	?	?	2
RP11-25K21.1	ENST00000453111	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FCGR2B	ENST00000480308	?	7
FCGR2B	ENST00000485778	?	2
RP11-25K21.1	ENST00000626340	?	2

Locus 10 (sentinel SNP rs17413015 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 11 (rs26496)

SNiPAcard

Block annotations

Block info

genomic range	chr5:96,123,055-96,151,968
block size	28,914 bp
variant count	25 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.605 [-2.841 – 0.73]	gene(s) hit or close-by	AC008906.1, CTD-2260A17.1, CTD-2260A17.2, CTD-2260A17.3, ERAP1
phastCons	μ = 0.067 [0 – 0.895]	eQTL gene(s)	CTD-2260A17.1, CTD-2260A17.3, ERAP1, ERAP2, LNPEP
GERP++	μ = -0.570 [-10.1 – 2.31]	potentially regulated gene(s)	–
CADD score	μ = 2.945 [0.098 – 9.842]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Ankylosing spondylitis	$<5.00\times10^{-12}$	GWAS Catalog	20062062	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM0911244	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP1	?	ENSG00000164307	pancreas	4.21×10^{-23} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	pancreas	4.98×10^{-6} (p-value)	GTEx Portal V6	3
ERAP1	?	ENSG00000164307	muscularis mucosae	2.83×10^{-8} (p-value)	GTEx Portal V6	24
ERAP1	?	ENSG00000164307	lung	2.55×10^{-33} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	lung	8.96×10^{-9} (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	lung	1.00×10^{-13} (p-value)	GTEx Portal V6	25
CTD-2260A17.3	?	ENSG00000272109	atrial appendage	2.69×10^{-6} (p-value)	GTEx Portal V6	6
ERAP1	?	ENSG00000164307	atrial appendage	1.23×10^{-6} (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	transformed fibroblasts	1.87×10^{-38} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	transformed fibroblasts	2.19×10^{-28} (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	blood	1.62×10^{-45} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	blood	3.22×10^{-17} (p-value)	GTEx Portal V6	25
CTD-2260A17.3	?	ENSG00000272109	blood	4.44×10^{-11} (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	breast	2.06×10^{-10} (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	breast	7.84×10^{-7} (p-value)	GTEx Portal V6	21

ERAP2	?	ENSG00000164308	breast	6.95×10-6 (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	tibial artery	6.71×10-8 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	tibial artery	1.01×10-6 (p-value)	GTEx Portal V6	21
ERAP1	?	ENSG00000164307	thyroid	3.20×10-10 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	thyroid	9.13×10-6 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	skeletal muscle	9.90×10-22 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	skeletal muscle	9.45×10-15 (p-value)	GTEx Portal V6	25
CTD-2260A17.3	?	ENSG00000272109	skeletal muscle	1.31×10-11 (p-value)	GTEx Portal V6	25
ERAP2	?	ENSG00000164308	skeletal muscle	7.63×10-7 (p-value)	GTEx Portal V6	4
ERAP1	?	ENSG00000164307	putamen	8.32×10-7 (p-value)	GTEx Portal V6	20
ERAP1	?	ENSG00000164307	EBV lymphocytes	2.08×10-15 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	EBV lymphocytes	2.47×10-8 (p-value)	GTEx Portal V6	24
ERAP1	?	ENSG00000164307	transverse colon	1.92×10-18 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	transverse colon	3.63×10-7 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	liver	1.32×10-8 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	sun exposed skin	8.36×10-25 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	sun exposed skin	5.11×10-8 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	sun exposed skin	4.99×10-8 (p-value)	GTEx Portal V6	24
ERAP1	?	ENSG00000164307	unexposed skin	1.54×10-18 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	unexposed skin	3.03×10-7 (p-value)	GTEx Portal V6	23
CTD-2260A17.3	?	ENSG00000272109	unexposed skin	1.23×10-8 (p-value)	GTEx Portal V6	25
ERAP2	?	ENSG00000164308	unexposed skin	1.00×10-5 (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	left ventricle	4.06×10-11 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	subcutaneous adipocytes	4.70×10-18 (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	subcutaneous adipocytes	5.21×10-8 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	subcutaneous adipocytes	5.24×10-10 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	caudate basal ganglia	8.05×10-7 (p-value)	GTEx Portal V6	20
ERAP1	?	ENSG00000164307	stomach	2.22×10-15 (p-value)	GTEx Portal V6	24
ERAP1	?	ENSG00000164307	visceral adipocytes	2.96×10-10 (p-value)	GTEx Portal V6	25
ERAP2	?	ENSG00000164308	visceral adipocytes	1.33×10-9 (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	visceral adipocytes	3.80×10-8 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	spleen	2.61×10-7 (p-value)	GTEx Portal V6	16
ERAP1	?	ENSG00000164307	coronary artery	1.88×10-7 (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	sigmoid colon	7.72×10-13 (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	tibial nerve	3.34×10-20 (p-value)	GTEx Portal V6	25

CTD-2260A17.3	?	ENSG00000272109	tibial nerve	1.80×10^{-6} (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	esophagus mucosa	2.25×10^{-25} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	esophagus mucosa	2.32×10^{-7} (p-value)	GTEx Portal V6	24
CTD-2260A17.3	?	ENSG00000272109	esophagus mucosa	1.50×10^{-10} (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	gastroesophageal junction	6.19×10^{-7} (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	adrenal gland	4.65×10^{-13} (p-value)	GTEx Portal V6	25
CTD-2260A17.1	?	ENSG00000248734	adrenal gland	1.73×10^{-6} (p-value)	GTEx Portal V6	11
CTD-2260A17.3	?	ENSG00000272109	testis	1.49×10^{-14} (p-value)	GTEx Portal V6	25
ERAP1	?	ENSG00000164307	frontal cortex	1.45×10^{-7} (p-value)	GTEx Portal V6	23
ERAP2	ENST00000513084	ILMN_1743145	skin	1.67×10^{-9} (p-value)	MuTHER consortium	9
ERAP2	ENST00000512869		blood	7.98×10^{-11} (p-value)	MuTHER consortium	9
ERAP2	ENST00000379904		adipocyte	1.05×10^{-9} (p-value)	MuTHER consortium	9
ERAP2	ENST00000437043					
ERAP1	ENST00000296754	ILMN_1752145	blood	1.25×10^{-5} (p-value)	MuTHER consortium	1
ERAP1	ENST00000296754	214012_at	blood	6.80×10^{-8} (p-value)	Dixon et al.	2
ERAP1	ENST00000443439	209788_s_at	blood	2.70×10^{-32} (p-value)	Dixon et al.	2
ERAP2	ENST00000513084	ILMN_1743145	monocyte	3.89×10^{-7} (p-value)	Fairfax et al.	2
ERAP2	ENST00000512869		b-cell	1.82×10^{-7} (p-value)	Fairfax et al.	2
ERAP2	ENST00000379904					
ERAP2	ENST00000437043					
ERAP1	ENST00000296754	ILMN_1752145	monocyte	3.32×10^{-6} (p-value)	Fairfax et al.	2
ERAP2	?	ENSG00000164308	blood	2.05×10^{-4} (q-value)	SeeQTL DB (HapMap)	3
LNPEP	ENST00000395770	ILMN_1814737	blood	7.60×10^{-6} (p-value)	Westra et al.	9
LNPEP	ENST00000231368					
CTD-2260A17.3	?	ENSG00000272109	aorta	1.22×10^{-5} (p-value)	GTEx Portal V6	2
ERAP2	ENST00000513084	ILMN_1743145	monocyte	6.82×10^{-23} (p-value)	Zeller et al.	2
ERAP2	ENST00000512869					
ERAP2	ENST00000379904					
ERAP2	ENST00000437043					
ERAP1	ENST00000443439	ILMN_2336220	monocyte	1.80×10^{-23} (p-value)	Zeller et al.	2
ERAP1	ENST00000514604					
ERAP1	ENST00000296754					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001412052 (enhancer)	1	blood (K562)	Max
ENSR00001285989 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me3
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, DNase1, PolII, H2AZ, H3K4me1
		blood (K562)	H2AZ
		A549	H3K27me3

blood (DND-41)	H3K9ac, H3K27ac, H3K4me1
skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC008906.1	upstream gene variant, downstream gene variant	7	ENST00000629418	?	?	9
CTD-2260A17.1	downstream gene variant	1353	ENST00000602972	?	?	6
CTD-2260A17.1	downstream gene variant	1353	ENST00000512856	?	?	6
CTD-2260A17.2	downstream gene variant	1954	ENST00000501338	?	?	1
CTD-2260A17.2	downstream gene variant	2100	ENST00000502262	?	?	1
CTD-2260A17.2	downstream gene variant	1958	ENST00000504056	?	?	1
CTD-2260A17.3	upstream gene variant	1195	ENST00000606656	?	?	7
CTD-2260A17.3	upstream gene variant	2509	ENST00000606346	?	?	7
ERAP1	downstream gene variant, upstream gene variant	1217	ENST00000503921	?	ENSP00000427025	6
ERAP1	downstream gene variant, upstream gene variant	2524	ENST00000508227	?	ENSP00000422631	6
ERAP1	upstream gene variant	592	ENST00000514604	?	?	6
ERAP1	upstream gene variant	4165	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	1
ERAP1	upstream gene variant	1576	ENST00000507859	?	?	5
ERAP1	downstream gene variant, upstream gene variant	138	ENST00000503311	?	?	14
ERAP1	downstream gene variant, upstream gene variant	382	ENST00000507154	?	ENSP00000421697	8
ERAP1	upstream gene variant	3974	ENST00000296754	NM_016442.3	ENSP00000296754	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ERAP1	ENST00000296754	NM_016442.3	ENSP00000296754	A	gcT/gcC	1
ERAP1	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	A	gcT/gcC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2260A17.2	ENST00000502262	?	?	1
CTD-2260A17.2	ENST00000504056	?	?	1
CTD-2260A17.2	ENST00000501338	?	?	1
ERAP1	ENST00000296754	NM_016442.3	ENSP00000296754	22
ERAP1	ENST00000508227	?	ENSP00000422631	7
ERAP1	ENST00000507859	?	?	6
ERAP1	ENST00000503311	?	?	2
ERAP1	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	22
ERAP1	ENST00000503921	?	ENSP00000427025	11

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

AC008906.1	ENST00000629418	?	1

Locus 12 (rs3117600)

SNiPAcard

Block annotations

Block info

genomic range	chr4:74,729,616-74,734,668
block size	5,053 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.100 [-3.83 – 0.025]	gene(s) hit or close-by	CXCL1
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	PF4V1
GERP++	μ = -1.685 [-5.79 – 2.96]	potentially regulated gene(s)	AC112518.3, AFM, CXCL6, EREG, MTHFD2L
CADD score	μ = 2.606 [0.082 – 7.402]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PF4V1	?	ENSG00000109272	blood	4.49×10-6 (p-value)	GTEx Portal V6	7
PF4V1	?	ENSG00000109272	breast	4.04×10-6 (p-value)	GTEx Portal V6	7
PF4V1	ENST00000226524	ILMN_1745522	monocyte	3.56×10-17 (p-value)	Zeller et al.	4
PF4V1	?	ENSG00000109272	transformed fibroblasts	8.50×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382197	1	ENCP00000041379	MTHFD2L
		ENCP00000041366	CXCL6
		ENCP00000041383	EREG
		ENCP00000041357	AFM
		ENCP00000041361	AC112518.3

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001432039 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		blood (K562)	Rad21, CTCF
		blood (DND-41)	CTCF
		skin (NHDF-AD)	DNase1
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF, Rad21
		lung (IMR90)	H3K27me3
		blood (GM12878)	CTCF
		A549	CTCF
		skin (NHEK)	CTCF
ENSR00001685779 (enhancer)	1	monocytes (Monocytes-CD14+)	DNase1
		lung (IMR90)	H3K27me3

ENSR00001242853 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K4me3, H3K4me2, DNase1
		skin (NHEK)	H3K4me1, H3K9ac, H3K4me2, CTCF, DNase1
ENSR00001242854 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, H3K4me2, H3K4me3, DNase1
		HSMMtube	H2AZ, H3K27me3
		blood (K562)	CTCF, Rad21, H3K9ac, H2AZ, H3K4me2, H3K4me3, H3K27me3
		skin (NHDF-AD)	H3K4me3, H3K4me2
		muscle (HSMM)	H3K4me3, H2AZ
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3, H3K4me3
		blood (GM12878)	H3K27me3
		nervous (NH-A)	H3K4me2, H3K27me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1, H3K4me1
		NHLF	H3K9ac, H3K4me3
		Osteobl	H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K4me3, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CXCL1	upstream gene variant	442	ENST00000509101	?	?	7
CXCL1	upstream gene variant	453	ENST00000395761	NM_001511.3	ENSP00000379110	7

Locus 13 (rs2844511)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,378,768-31,390,410
block size	11,643 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.808 [-1.898 – 0.403]	gene(s) hit or close-by	HCP5, MICA
phastCons	μ = 0.008 [0 – 0.054]	eQTL gene(s)	ATP6V1G2, HCG22, HCG27, HLA-B, HLA-C, LTA, MICA, PSORS1C1, PSORS1C2, TAP2, WASF5P, XXbac-BPG181B23.7, XXbac-BPG246D15.9, XXbac-BPG248L24.10, XXbac-BPG248L24.12, XXbac-BPG248L24.13, XXbac-BPG299F13.17, Y_RNA
GERP++	μ = -0.466 [-3.52 – 1.18]	potentially regulated gene(s)	MICA, MICA, MICA, MICA, MICA, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2
CADD score	μ = 2.526 [0.257 – 6.359]	disease gene(s)	HLA-B, TAP2, LTA, , , , , VARS2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
CERVICAL CANCER	$<4.00\times10^{-18}$	GWAS Catalog	23482656		1
Height	$<8.00\times10^{-29}$	GWAS Catalog	20881960		1
cholesterol	4.59×10^{-6}	Metabolomics GWAS Server	24816252		5
Behcet syndrome	4.29×10^{-8}	dbGaP	pha002888	dbGaP	2
Lupus erythematosus, systemic	3.60×10^{-9}	dbGaP	pha002848	dbGaP	2

Disease gene annotation

gene	trait	source DB	source entry/link	
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
TAP2	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592	orphanet
LTA	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988	OMIM
LTA	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
TAP2	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICA	?	ENSG00000204520	atrial appendage	3.61×10^{-9} (p-value)	GTEx Portal V6	9

HLA-B	?	ENSG00000234745	atrial appendage	1.28×10-12 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	atrial appendage	4.65×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	transformed fibroblasts	2.01×10-26 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	transformed fibroblasts	6.93×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	transformed fibroblasts	3.01×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	1.83×10-6 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	4.38×10-20 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	blood	4.96×10-6 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	blood	4.13×10-17 (p-value)	GTEx Portal V6	9
XXbac-BPG299F13.17	?	ENSG00000272501	blood	2.58×10-6 (p-value)	GTEx Portal V6	8
MICA	?	ENSG00000204520	blood	5.66×10-8 (p-value)	GTEx Portal V6	8
XXbac-BPG248L24.12	?	ENSG00000271581	blood	3.24×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	blood	2.00×10-9 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	breast	1.20×10-10 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	breast	5.82×10-7 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	breast	1.36×10-10 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	breast	2.85×10-6 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	breast	7.91×10-14 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	skeletal muscle	1.53×10-20 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	skeletal muscle	1.22×10-10 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	skeletal muscle	5.77×10-12 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	2.95×10-10 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	skeletal muscle	2.71×10-17 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	putamen	1.12×10-7 (p-value)	GTEx Portal V6	8
MICA	?	ENSG00000204520	EBV lymphocytes	6.51×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	EBV lymphocytes	4.39×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	EBV lymphocytes	1.40×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	ovary	1.40×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	ovary	1.32×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	sun exposed skin	2.64×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	5.30×10-18 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	sun exposed skin	6.34×10-14 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	5.95×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	sun exposed skin	5.96×10-11 (p-value)	GTEx Portal V6	9
PSORS1C1	?	ENSG00000204540	sun exposed skin	4.06×10-9 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	unexposed skin	6.10×10-10 (p-value)	GTEx Portal V6	9

HLA-C	?	ENSG00000204525	unexposed skin	5.60×10^{-6} (p-value)	GTEx Portal V6	3
HLA-B	?	ENSG00000234745	unexposed skin	4.17×10^{-10} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	unexposed skin	1.13×10^{-9} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	unexposed skin	6.69×10^{-12} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	aorta	2.49×10^{-14} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	aorta	7.96×10^{-7} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	aorta	2.87×10^{-13} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	aorta	4.29×10^{-13} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	left ventricle	3.03×10^{-15} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	left ventricle	7.87×10^{-8} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	1.90×10^{-8} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	left ventricle	1.27×10^{-11} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	left ventricle	3.93×10^{-6} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	subcutaneous adipocytes	6.44×10^{-16} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	2.42×10^{-6} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	subcutaneous adipocytes	1.88×10^{-13} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	6.16×10^{-7} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	1.09×10^{-22} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	caudate basal ganglia	1.37×10^{-11} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	caudate basal ganglia	7.92×10^{-12} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	visceral adipocytes	1.94×10^{-10} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	visceral adipocytes	7.68×10^{-8} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	visceral adipocytes	1.41×10^{-11} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	3.58×10^{-7} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	visceral adipocytes	2.01×10^{-11} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	stomach	5.75×10^{-11} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	stomach	1.01×10^{-6} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	stomach	6.69×10^{-8} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	stomach	3.80×10^{-8} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	stomach	5.92×10^{-12} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	cortex	5.99×10^{-8} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	tibial nerve	9.55×10^{-18} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	tibial nerve	3.35×10^{-8} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	tibial nerve	5.58×10^{-13} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	5.88×10^{-6} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	1.36×10^{-15} (p-value)	GTEx Portal V6	9
XXbac-BPG299F13.17	?	ENSG00000272501	tibial nerve	6.67×10^{-6} (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	esophagus mucosa	1.45×10^{-10} (p-value)	GTEx Portal V6	9

HLA-C	?	ENSG00000204525	esophagus mucosa	1.45×10-10 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	esophagus mucosa	7.05×10-13 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	esophagus mucosa	4.62×10-13 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	8.48×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	esophagus mucosa	3.06×10-7 (p-value)	GTEx Portal V6	9
PSORS1C1	?	ENSG00000204540	esophagus mucosa	1.73×10-6 (p-value)	GTEx Portal V6	9
HCG22	?	ENSG00000228789	esophagus mucosa	2.62×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	pituitary	2.24×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	testis	5.59×10-13 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	testis	3.13×10-6 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	testis	3.30×10-11 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	frontal cortex	3.60×10-7 (p-value)	GTEx Portal V6	8
XXbac-BPG248L24.10	?	ENSG00000229836	pancreas	6.76×10-7 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	pancreas	4.51×10-6 (p-value)	GTEx Portal V6	6
XXbac-BPG181B23.7	?	ENSG00000272221	pancreas	1.83×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.13	?	ENSG00000256166	pancreas	2.19×10-6 (p-value)	GTEx Portal V6	7
MICA	?	ENSG00000204520	pancreas	6.13×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	pancreas	3.12×10-6 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	pancreas	4.16×10-8 (p-value)	GTEx Portal V6	9
WASF5P	?	ENSG00000231402	pancreas	1.44×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	muscularis mucosae	5.92×10-13 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	muscularis mucosae	7.16×10-7 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	muscularis mucosae	3.10×10-12 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	muscularis mucosae	6.11×10-18 (p-value)	GTEx Portal V6	9
PSORS1C2	?	ENSG00000204538	lung	1.07×10-6 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	lung	4.97×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	lung	2.02×10-17 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	lung	7.89×10-20 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	lung	1.65×10-10 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	lung	3.16×10-12 (p-value)	GTEx Portal V6	9
PSORS1C1	?	ENSG00000204540	lung	5.94×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial artery	7.30×10-24 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	tibial artery	3.23×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	tibial artery	2.88×10-16 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	1.88×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	8.90×10-23 (p-value)	GTEx Portal V6	9
PSORS1C2	?	ENSG00000204538	thyroid	3.56×10-7 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	thyroid	7.34×10-7 (p-value)	GTEx Portal V6	9

XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	6.91×10-19 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	thyroid	1.74×10-20 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	1.48×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	thyroid	1.64×10-11 (p-value)	GTEx Portal V6	9
Y_RNA	?	ENSG00000199332	thyroid	4.47×10-7 (p-value)	GTEx Portal V6	9
PSORS1C1	?	ENSG00000204540	thyroid	5.12×10-8 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	transverse colon	8.76×10-12 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	transverse colon	1.07×10-15 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	liver	9.37×10-10 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	liver	1.25×10-6 (p-value)	GTEx Portal V6	6
XXbac-BPG181B23.7	?	ENSG00000272221	terminal ileum	9.54×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	spleen	2.32×10-6 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	spleen	5.35×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	sigmoid colon	3.77×10-9 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	sigmoid colon	2.37×10-7 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	sigmoid colon	3.35×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	sigmoid colon	9.65×10-7 (p-value)	GTEx Portal V6	7
XXbac-BPG181B23.7	?	ENSG00000272221	sigmoid colon	6.00×10-9 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	coronary artery	1.87×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	coronary artery	1.68×10-9 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	gastroesophageal junction	2.59×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	gastroesophageal junction	7.09×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	gastroesophageal junction	3.26×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	cerebellar hemisphere	1.70×10-8 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	adrenal gland	1.10×10-10 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	adrenal gland	1.45×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	cerebellum	1.00×10-10 (p-value)	GTEx Portal V6	9
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	7.71×10-7 (p-value)	MuTHER consortium	6
ATP6V1G2	ENST00000303892					
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					
HCG27	ENST00000383331	ILMN_1746436	adipocyte	4.16×10-7 (p-value)	MuTHER consortium	6
HCG27	ENST00000415276					
LTA	ENST00000418386	ILMN_1795464	blood	1.20×10-4 (p-value)	Westra et al.	2
LTA	ENST00000471842					
LTA	ENST00000454783					
LTA	ENST00000489638					
MICA	ENST00000449934	ILMN_3241692	monocyte	4.42×10-5 (p-value)	Fairfax et al.	4
MICA	ENST00000616296					
MICA	ENST00000421350					

TAP2	ENST00000374899	ILMN_1759250	monocyte	4.61×10-4 (p-value)	Fairfax et al.	4
TAP2	ENST00000620123					
TAP2	ENST00000374897					
TAP2	ENST00000464100					
XXbac-BPG246D15.9	ENST00000452392					
XXbac-BPG181B23.7	ENST00000606743	230937_at	blood	1.10×10-10 (p-value)	Dixon et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444714	1	ENCP00000048015	MICA MICA MICA MICA MICA
		ENCP00000047968	VARS2 VARS2 VARS2 VARS2 VARS2 VARS2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MICA	downstream gene variant	2393	ENST00000449934	NM_001177519.2	ENSP00000413079	3
MICA	downstream gene variant	2395	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	3
MICA	downstream gene variant	2393	ENST00000421350	?	ENSP00000402410	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	9
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	4
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	4
MICA	ENST00000421350	?	ENSP00000402410	4

Locus 14 (rs17482078)

SNiPAcard

Block annotations

Block info

genomic range	chr5:96,118,866-96,161,069
block size	42,204 bp
variant count	23 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.139 [-5.113 – 4.515]	gene(s) hit or close-by	AC008906.1, CAST, CTD-2260A17.1, CTD-2260A17.2, CTD-2260A17.3, ERAP1
phastCons	μ = 0.153 [0 – 1]	eQTL gene(s)	CAST, CTC-506B8.1, CTD-2260A17.1, ERAP1
GERP++	μ = 0.119 [-5.73 – 5.78]	potentially regulated gene(s)	CTD-2215E18.2, ERAP1, ERAP2, LIX1, LNPEP
CADD score	μ = 5.730 [0.159 – 21.1]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Behcet's disease	<4.00×10-8	GWAS Catalog	23291587	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM0911245	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ERAP1	missense variant	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	3	3			3
ERAP1	missense variant	ENST00000296754	NM_016442.3	ENSP00000296754	3	3			3

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP1	ENST00000296754	ILMN_1752145	blood	3.69×10-17 (p-value)	MuTHER consortium	10
			adipocyte	5.78×10-8 (p-value)	MuTHER consortium	10
ERAP1	?	ENSG00000164307	caudate basal ganglia	3.10×10-8 (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	skeletal muscle	2.85×10-21 (p-value)	GTEx Portal V6	23
CTD-2260A17.1	?	ENSG00000248734	skeletal muscle	1.88×10-10 (p-value)	GTEx Portal V6	23
ERAP1	?	ENSG00000164307	putamen	2.52×10-6 (p-value)	GTEx Portal V6	2

ERAP1	?	ENSG00000164307	EBV lymphocytes	6.40×10-7 (p-value)	GTEx Portal V6	2
CTD-2260A17.1	?	ENSG00000248734	ovary	3.35×10-8 (p-value)	GTEx Portal V6	22
ERAP1	?	ENSG00000164307	blood	7.47×10-7 (p-value)	GTEx Portal V6	4
ERAP1	ENST00000296754	ILMN_1752145	monocyte	7.42×10-8 (p-value)	Fairfax et al.	3
ERAP1	ENST00000296754	214012_at	blood	1.40×10-22 (p-value)	Dixon et al.	1
ERAP1	ENST00000443439	209788_s_at	blood	9.40×10-17 (p-value)	Dixon et al.	1
CTC-506B8.1	?	ENSG00000249180	subcutaneous adipocytes	8.39×10-6 (p-value)	GTEx Portal V6	9
ERAP1	ENST00000443439	ILMN_2336220	monocyte	8.06×10-37 (p-value)	Zeller et al.	3
ERAP1	ENST00000514604					
ERAP1	ENST00000296754					
ERAP1	ENST00000296754	ILMN_1752145	monocyte	9.66×10-24 (p-value)	Zeller et al.	3
CAST	ENST00000511782	ILMN_1783627	blood	2.96×10-6 (p-value)	Westra et al.	1
CAST	ENST00000511049					
CAST	ENST00000395813					
CAST	ENST00000421689					
CAST	ENST00000506811					
CAST	ENST00000510156					
CAST	ENST00000507487					
CAST	ENST00000341926					
CAST	ENST00000508117					
CAST	ENST00000508830					
CAST	ENST00000338252					
CAST	ENST00000505143					
CAST	ENST00000514055					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CJUN	MA0489.1	1	no	0.000	1
CJUN	MA0303.1	2	no	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000417173	1	ENCP00000044982	LIX1
ENCE00000417059	1	ENCP00000044978	LNPEP
		ENCP00000044966	ERAP1
		ENCP00000044987	CTD-2215E18.2
		ENCP00000044971	ERAP2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001285985 (promoter flanking region)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1
		cervix (HeLa-S3)	DNase1

monocytes (Monocytes-CD14+)	DNase1, H3K36me3
endothelium (HUVEC)	H3K36me3, Cjun, DNase1
liver (HepG2)	H3K36me3
blood (GM12878)	H3K36me3
lung (IMR90)	H3K36me3
nervous (NH-A)	H3K36me3
A549	H3K36me3
skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC008906.1	upstream gene variant, downstream gene variant	261	ENST00000629418	?	?	9
CAST	downstream gene variant	3567	ENST00000510098	?	ENSP00000427195	2
CTD-2260A17.1	upstream gene variant, downstream gene variant	507	ENST00000512856	?	?	6
CTD-2260A17.1	upstream gene variant, downstream gene variant	507	ENST00000602972	?	?	6
CTD-2260A17.3	upstream gene variant	630	ENST00000606346	?	?	6
CTD-2260A17.3	upstream gene variant	190	ENST00000606656	?	?	5
ERAP1	upstream gene variant	1981	ENST00000512852	?	ENSP00000425381	4
ERAP1	downstream gene variant, upstream gene variant	100	ENST00000507859	?	?	7
ERAP1	downstream gene variant	2331	ENST00000508227	?	ENSP00000422631	3
ERAP1	downstream gene variant	1467	ENST00000503921	?	ENSP00000427025	3
ERAP1	downstream gene variant	177	ENST00000507154	?	ENSP00000421697	6
ERAP1	downstream gene variant, upstream gene variant	161	ENST00000503311	?	?	10
ERAP1	upstream gene variant	966	ENST00000514604	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2260A17.1	ENST00000602972	?	?	2
CTD-2260A17.1	ENST00000512856	?	?	2
CTD-2260A17.2	ENST00000501338	?	?	2
CTD-2260A17.2	ENST00000502262	?	?	2
CTD-2260A17.2	ENST00000504056	?	?	2
CTD-2260A17.3	ENST00000606656	?	?	1
ERAP1	ENST00000508227	?	ENSP00000422631	6
ERAP1	ENST00000443439	NM_001040458.1, NM_001198541.1	ENSP00000406304	18
ERAP1	ENST00000296754	NM_016442.3	ENSP00000296754	18
ERAP1	ENST00000507154	?	ENSP00000421697	2
ERAP1	ENST00000503921	?	ENSP00000427025	9
ERAP1	ENST00000507859	?	?	3
ERAP1	ENST00000514604	?	?	4
ERAP1	ENST00000503311	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

CTD-2260A17.1	ENST00000602972	?	1
CTD-2260A17.1	ENST00000512856	?	1
ERAP1	ENST00000507859	?	2
ERAP1	ENST00000514604	?	4

Locus 14 (sentinel SNP rs17482078 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 15 (rs3776081)

SNiPAcard

Block annotations

Block info

genomic range	chr5:149,519,260-149,537,194
block size	17,935 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.217 [-0.624 – 0.211]	gene(s) hit or close-by	PDGFRB
phastCons	μ = 0.001 [0 – 0.002]	eQTL gene(s)	CDX1, CSF1R, PDGFRB
GERP++	μ = 0.540 [-0.121 – 1.03]	potentially regulated gene(s)	–
CADD score	μ = 6.404 [4.963 – 8.056]	disease gene(s)	PDGFRB, CSF1R

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PDGFRB	Familial Infantile Myofibromatosis	DECIPHER	MIM:228550
PDGFRB	JUVENILE MYELOMONOCYTIC LEUKEMIA	OMIM	MIM:607785
PDGFRB	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 4	OMIM	MIM:615007
PDGFRB	MYOFIBROMATOSIS, INFANTILE, 1	OMIM	MIM:228550
PDGFRB	LEUKEMIA, ACUTE MYELOID	OMIM	MIM:601626
PDGFRB	MYELOPROLIFERATIVE DISORDER, CHRONIC, WITH EOSINOPHILIA	OMIM	MIM:131440
CSF1R	LEUKOENCEPHALOPATHY, DIFFUSE HEREDITARY, WITH SPHEROIDS	OMIM	MIM:221820
PDGFRB	Bilateral striopallidodentate calcinosis	OrphaNet	OrphaNet:1980
PDGFRB	Unclassified chronic myeloproliferative disease	OrphaNet	OrphaNet:86830
PDGFRB	Chronic myelomonocytic leukemia	OrphaNet	OrphaNet:98823
PDGFRB	Idiopathic hypereosinophilic syndrome	OrphaNet	OrphaNet:3260
PDGFRB	Infantile myofibromatosis 1	OrphaNet	OrphaNet:2591
PDGFRB	Myeloid neoplasm associated with PDGFRB rearrangement	OrphaNet	OrphaNet:168950
CSF1R	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia	OrphaNet	OrphaNet:313808

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PDGFRB	ENST00000520579	ILMN_1815057	blood	8.08×10^{-39} (p-value)	MuTHER consortium	1
PDGFRB	ENST00000261799					
CSF1R	ENST00000509861	ILMN_1686623	blood	5.51×10^{-7} (p-value)	Westra et al.	1
CSF1R	ENST00000504875					
CSF1R	ENST00000286301					
CDX1	?	ENSG00000113722	tibial nerve	5.45×10^{-13} (p-value)	GTEx Portal V6	2
PDGFRB	?	ENSG00000113721	EBV lymphocytes	6.13×10^{-8} (p-value)	GTEx Portal V6	2
PDGFRB	ENST00000520579	ILMN_1815057	b-cell	5.54×10^{-6} (p-value)	Fairfax et al.	1
PDGFRB	ENST00000261799					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001293753 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me1, USF1
		HSMMtube	H3K4me1, H3K9ac, H3K27ac, H3K36me3, DNase1
		blood (K562)	H3K27me3, DNase1, Egr1, PU1, Cjun
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, H3K4me1, H3K36me3
		muscle (HSMM)	H3K27ac, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	FOSL2, Jund, H3K4me3, PolII, TAF1, ELF1, USF1, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF, DNase1
		lung (IMR90)	H3K56ac, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4ac, H2BK120ac, H2BK20ac, H2AK5ac, H3K9ac, H3K4me1, H2BK12ac, DNase1, H4K8ac, H4K91ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me1, H3K4me2, H3K36me3, H3K27ac
		skin (NHEK)	H3K27me3, DNase1
		NHLF	H3K27ac, H3K36me3, H3K9ac, H3K4me1, DNase1
		Osteobl	H3K36me3, H3K4me1, H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Cjun, Jund, H3K4me2, H3K27ac, H3K4me1, Max
		monocytes (Monocytes-CD14+)	H3K27ac, DNase1
		endothelium (HUVEC)	H3K27me3
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac
ENSR00001698907 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, Rad21, CTCF
		HSMMtube	DNase1, CTCF
		blood (K562)	H3K27me3, PolII, Egr1, Rad21, H2AZ, H3K4me2, CTCF, DNase1
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, CTCF, H3K4me1
		muscle (HSMM)	H2AZ, CTCF
		liver (HepG2)	Rad21, H2AZ, H3K4me2, CTCF, H3K27me3, DNase1, H3K4me3
		blood (GM12878)	DNase1, EBF1, H2AZ, Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	H3K27me3, CTCF
		NHLF	DNase1, H3K4me1, CTCF, H3K27ac
		Osteobl	CTCF, H3K27ac
		blood (DND-41)	CTCF, H3K27ac, H3K27me3, H3K4me1
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K4me1, CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3, CTCF
		endothelium (HUVEC)	H3K36me3, DNase1, H3K27me3, CTCF
		A549	H3K4me3, H3K4me2, CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDGFRB	upstream gene variant	2260	ENST00000517957	?	ENSP00000430715	1
PDGFRB	upstream gene variant	1771	ENST00000517660	?	?	1
PDGFRB	downstream gene variant, upstream gene variant	867	ENST00000523456	?	?	2
PDGFRB	upstream gene variant	1772	ENST00000520579	?	ENSP00000430026	1
PDGFRB	upstream gene variant	2004	ENST00000517488	?	ENSP00000429218	1
PDGFRB	upstream gene variant	1771	ENST00000261799	NM_002609.3	ENSP00000261799	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDGFRB	ENST00000517660	?	?	2
PDGFRB	ENST00000517488	?	ENSP00000429218	2

PDGFRB	ENST00000520579	?	ENSP00000430026	2
PDGFRB	ENST00000261799	NM_002609.3	ENSP00000261799	2

Locus 15 (sentinel SNP rs3776081 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 16 (rs7551957)

SNiPAcard

Block annotations

Block info

genomic range	chr1:161,469,054-161,479,745
block size	10,692 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.167 [-0.902 – 0.932]	**gene(s) hit or close-by**	FCGR2A, RP11-25K21.6
phastCons	μ = 0.192 [0 – 0.836]	**eQTL gene(s)**	SDHC
GERP++	μ = -0.468 [-4.15 – 1.13]	**potentially regulated gene(s)**	FCGR2A
CADD score	μ = 2.723 [0.63 – 5.131]	**disease gene(s)**	SDHC

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
SYSTEMIC LUPUS ERYTHEMATOSUS	$<9.00\times10^{-6}$	GWAS Catalog	24871463	1
Inflammatory bowel disease	$<2.00\times10^{-38}$	GWAS Catalog	23128233	1
Kawasaki disease	$<7.00\times10^{-11}$	GWAS Catalog	22081228	1
Ulcerative colitis	$<2.00\times10^{-20}$	GWAS Catalog	21297633	1
Ulcerative colitis	$<2.00\times10^{-12}$	GWAS Catalog	19915573	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Malaria, severe, susceptibility to	risk factor	ClinVar	RCV000054529.1	1
Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis	risk factor	ClinVar	RCV000015947.2	1
Lupus nephritis, susceptibility to	risk factor	ClinVar	RCV000015946.2	1
?	HGMD curated	HGMD	CM960640	1
LUPUS NEPHRITIS, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:146790	1

Disease gene annotation

gene	trait	source DB	source entry/link
SDHC	PARAGANGLIOMA AND GASTRIC STROMAL SARCOMA	OMIM	MIM:606864
SDHC	PARAGANGLIOMAS 3	OMIM	MIM:605373
SDHC	COWDEN SYNDROME	OrphaNet	OrphaNet:201
SDHC	Gastrointestinal stromal tumor	OrphaNet	OrphaNet:44890
SDHC	Carney-Stratakis syndrome	OrphaNet	OrphaNet:97286
SDHC	Hereditary pheochromocytoma-paraganglioma	OrphaNet	OrphaNet:29072

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FCGR2A	missense variant	ENST00000367972	NM_021642.3	ENSP00000356949	H/R	cAt/cGt	tolerated	benign	1

FCGR2A	missense variant	ENST00000271450	NM_001136219.1, NM_201563.5	ENSP00000271450	H/R	cAt/cGt	tolerated	benign	1
FCGR2A	missense variant	ENST00000467525	?	ENSP00000476495	H/R	cAt/cGt	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SDHC	ENST00000367975	ILMN_2323366	blood	1.21×10^{-4} (p-value)	Westra et al.	1
?	?	ILMN_1728639	blood	9.71×10^{-5} (p-value)	Westra et al.	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000374341	1	ENST00000367972, ENST00000271450, ENST00000471026, ENST00000467525	FCGR2A

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000286584 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		nervous (NH-A)	H3K27me3
		blood (K562)	H2AZ
		A549	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR2A	upstream gene variant	3422	ENST00000461298	?	?	3
FCGR2A	upstream gene variant	4715	ENST00000367972	NM_021642.3	ENSP00000356949	1
FCGR2A	upstream gene variant, downstream gene variant	2571	ENST00000497474	?	?	4
FCGR2A	upstream gene variant	4715	ENST00000271450	NM_001136219.1, NM_201563.5	ENSP00000271450	1
FCGR2A	upstream gene variant	3943	ENST00000459885	?	ENSP00000476411	3
FCGR2A	upstream gene variant, downstream gene variant	2652	ENST00000473080	?	?	4
FCGR2A	upstream gene variant	4742	ENST00000483665	?	ENSP00000440148	1
FCGR2A	upstream gene variant, downstream gene variant	56	ENST00000536731	?	ENSP00000441156	2
FCGR2A	downstream gene variant	2380	ENST00000482233	?	?	3
FCGR2A	upstream gene variant	4731	ENST00000467654	?	?	1
FCGR2A	upstream gene variant	4665	ENST00000486608	?	?	1
RP11-25K21.6	upstream gene variant	3221	ENST00000537821	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2A	ENST00000367972	NM_021642.3	ENSP00000356949	2
FCGR2A	ENST00000271450	NM_001136219.1, NM_201563.5	ENSP00000271450	2
FCGR2A	ENST00000536731	?	ENSP00000441156	2
FCGR2A	ENST00000471026	?	?	2
FCGR2A	ENST00000467654	?	?	2
FCGR2A	ENST00000483665	?	ENSP00000440148	2
FCGR2A	ENST00000467525	?	ENSP00000476495	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2A	ENST00000483665	?	ENSP00000440148	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FCGR2A	ENST00000471026	?	1
FCGR2A	ENST00000467654	?	1

Locus 17 (rs4525)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,469,142-169,513,436
block size	44,295 bp
variant count	41 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.242 [-2.54 – 1.334]	gene(s) hit or close-by	F5
phastCons	μ = 0.038 [0 – 0.602]	eQTL gene(s)	–
GERP++	μ = -0.719 [-5.59 – 2.81]	potentially regulated gene(s)	RP1-206D15.5, SELL, SELP
CADD score	μ = 2.836 [0.001 – 9.022]	disease gene(s)	F5

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM101722	1

Disease gene annotation

gene	trait	source DB	source entry/link
F5	STROKE, ISCHEMIC	OMIM	MIM:601367
F5	THROMBOPHILIA DUE TO ACTIVATED PROTEIN C RESISTANCE	OMIM	MIM:188055
F5	PREGNANCY LOSS, RECURRENT, SUSCEPTIBILITY TO, 1	OMIM	MIM:614389
F5	FACTOR V DEFICIENCY	OMIM	MIM:227400
F5	BUDD-CHIARI SYNDROME	OMIM	MIM:600880
F5	Congenital factor V deficiency	OrphaNet	OrphaNet:326
F5	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217
F5	BUDD-CHIARI SYNDROME	OrphaNet	OrphaNet:131
F5	East Texas bleeding disorder	OrphaNet	OrphaNet:391320

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
F5	missense variant	ENST00000367796	?	ENSP00000356770	3	3			3
F5	missense variant	ENST00000367797	NM_000130.4	ENSP00000356771	3	3			3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000043595	1	ENCP00000005439	RP1-206D15.5
ENCE00000043639	1	ENCP00000005449	SELL
		ENCP00000005442	SELP

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000547323 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	Rad21, H3K4me1, CTCF
		blood (K562)	H2AZ
ENSR00001525842 (promoter flanking region)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF, H3K4me2, Rad21
		blood (K562)	DNase1, H3K9ac, H2AZ, H3K4me3
ENSR00001525843 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1, Cjun, Jund
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K4me1, H3K36me3
		nervous (NH-A)	DNase1
ENSR00000547326 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		muscle (HSMM)	DNase1
ENSR00000287070 (promoter flanking region)	4	embryonic stem cell (H1ESC)	Rad21, Jund, ATF3, Tcf12, BCL11A, H3K4me2, SP1, USF1, DNase1
		HSMMtube	DNase1
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K4me2
ENSR00001587730 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Cjun, Jund, Cfos
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1
ENSR00000547331 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Cfos, Jund, Cjun
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
F5	downstream gene variant	1454	ENST00000367796	?	ENSP00000356770	3

F5	downstream gene variant, upstream gene variant	1454	ENST00000495481	?	?		7
F5	downstream gene variant	1454	ENST00000367797	NM_000130.4	ENSP00000356771		3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
F5	ENST00000367796	?	ENSP00000356770	4	4	4
F5	ENST00000367797	NM_000130.4	ENSP00000356771	4	4	4

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F5	ENST00000367796	?	ENSP00000356770	25
F5	ENST00000495481	?	?	1
F5	ENST00000367797	NM_000130.4	ENSP00000356771	25

Locus 18 (rs2302465)

SNiPAcard

Block annotations

Block info

genomic range	chr4:15,527,099-15,730,349
block size	203,251 bp
variant count	78 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.038 [-4.028 – 5.711]	gene(s) hit or close-by	BST1, CC2D2A, FAM200B, FBXL5, RP11-115L11.1, RP11-799M12.2
phastCons	μ = 0.086 [0 – 0.995]	eQTL gene(s)	CD38, FAM200B, FBXL5, RP11-799M12.2
GERP++	μ = -0.416 [-7.17 – 5.53]	potentially regulated gene(s)	BST1, C1QTNF7, FAM200B, FBXL5, FGFBP2, RP11-442P12.1, RP11-442P12.2
CADD score	μ = 4.767 [0.048 – 24.4]	disease gene(s)	CC2D2A

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000079447.7	1
?	HGMD curated	HGMD	CM065042	1

Disease gene annotation

gene	trait	source DB	source entry/link
CC2D2A	MECKEL SYNDROME, TYPE 6	OMIM	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	OMIM	MIM:612285
CC2D2A	COACH SYNDROME	OMIM	MIM:216360
CC2D2A	COACH syndrome	DECIPHER	MIM:216360
CC2D2A	MECKEL SYNDROME, TYPE 6	DECIPHER	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	DECIPHER	MIM:612285
CC2D2A	Joubert syndrome with oculorenal defect	OrphaNet	OrphaNet:2318
CC2D2A	Meckel syndrome	OrphaNet	OrphaNet:564
CC2D2A	Joubert syndrome with hepatic defect	OrphaNet	OrphaNet:1454

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
BST1	missense variant	ENST00000382346	?	ENSP00000371783	H/R	cAt/cGt	?	?	1
BST1	missense variant	ENST00000265016	NM_004334.2	ENSP00000265016	H/R	cAt/cGt	?	?	1
BST1	missense variant	ENST00000505785	?	ENSP00000423357	H/R	cAt/cGt	?	?	1
FAM200B	missense variant	ENST00000503617	?	ENSP00000422751	H/D	Cat/Gat	?	?	1
FAM200B	missense variant	ENST00000622362	NM_001145191.1	ENSP00000483930	H/D	Cat/Gat	?	?	1
FAM200B	missense variant	ENST00000422728	?	ENSP00000393017	H/D	Cat/Gat	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FBXL5	ENST00000511441	ILMN_1673370	blood	2.34×10^{-7} (p-value)	MuTHER consortium	7
FBXL5	ENST00000412094		adipocyte	4.43×10^{-5} (p-value)	MuTHER consortium	7
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
CD38	ENST00000502843	ILMN_2233783	adipocyte	1.11×10^{-8} (p-value)	MuTHER consortium	7
CD38	ENST00000226279					
FBXL5	ENST00000511441	ILMN_1673370	monocyte	3.60×10^{-19} (p-value)	Zeller et al.	2
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
FAM200B	?	ENSG00000237765	transformed fibroblasts	6.06×10^{-6} (p-value)	GTEx Portal V6	43
FAM200B	?	ENSG00000237765	EBV lymphocytes	2.63×10^{-6} (p-value)	GTEx Portal V6	50
RP11-799M12.2	?	ENSG00000273133	blood	2.33×10^{-6} (p-value)	GTEx Portal V6	1
FBXL5	ENST00000511441	ILMN_1673370	monocyte	2.17×10^{-4} (p-value)	Fairfax et al.	1
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000040477		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000374034	1	ENCP00000040459	C1QTNF7
		ENCP00000040482	RP11-442P12.1
		ENCP00000040484	FGFBP2
		ENCP00000040479	RP11-442P12.2
ENCE00000374231	1	ENCP00000040475	BST1
ENCE00000374206	1	ENCP00000040473	FBXL5 FAM200B
ENCE00000374192	1	ENCP00000040472	FBXL5
		ENCP00000040476	BST1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001237209 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	H3K36me3
ENSR00001237214 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K36me3
	5	embryonic stem cell (H1ESC)	DNase1, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, H4K5ac, H3K36me3, H3K27me3,

ENSR00001429713 (promoter)			PolII, Rad21, TAF7, CTCF, H3K27ac
		HSMMtube	H3K4me3, H2AZ, DNase1, H3K79me2, CTCF, H3K9ac, H3K4me2, H3K27ac
		blood (K562)	H3K36me3, DNase1, H3K4me3, CTCF, H3K4me2, Egr1, H3K27ac, Max, Rad21, E2F6, H3K9ac, USF1, Cmyc, H3K79me2, H2AZ, PolII
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac
		liver (HepG2)	DNase1, PolII, H3K79me2, Rad21, TAF1, USF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3
		blood (GM12878)	CTCF, H3K9ac, PolII, H2AZ, DNase1, Egr1, Yy1, Rad21, H3K79me2, H3K4me3, USF1, H3K27ac, H3K4me2
		lung (IMR90)	DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K4me3, H3K4ac, H3K9ac, CTCF
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K27ac, H3K9ac
		NHLF	H3K4me3, DNase1, H3K9ac, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K36me3, H3K4me3, H3K9ac, H3K27ac
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, CTCF, PolII, H3K79me2, H3K4me3, H3K27ac, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, CTCF, DNase1
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
ENSR00001429714 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3
		A549	H3K36me3
ENSR00001429715 (promoter flanking region)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	DNase1, H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	DNase1, H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K27ac, Jund
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	Cjun
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		nervous (NH-A)	DNase1, H3K36me3
		skin (NHEK)	DNase1, H3K4me1, H3K36me3
ENSR00001237226 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K9ac, H3K36me3, CTCF, PolII, Rad21, Gabp, TAF7, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, Srf
		HSMMtube	H3K4me3, H2AZ, DNase1, H3K4me2, H3K9ac, H3K79me2
		blood (K562)	Ini1, Egr1, H3K27ac, ZBTB33, Max, Srf, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K4me3, DNase1, H3K9ac
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2
		liver (HepG2)	H3K9ac, H3K27ac, H3K4me3, DNase1, HNF4A, H3K4me2, Gabp, Srf, Sin3Ak20, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, ZBTB33
		lung (IMR90)	H3K4me3, H3K9ac, DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2, H3K36me3
		blood (GM12878)	Sin3Ak20, PU1, PolII, H2AZ, DNase1, ELF1, H3K79me2, H3K4me3, ZBTB33, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K36me3, DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K36me3, PolII, Gabp, H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K9ac, H3K36me3, H3K4me3, H3K27ac, DNase1, H3K4me1, H3K4me2
		endothelium (HUVEC)	PolII, DNase1, H3K36me3, H3K27ac, H3K9ac, Max, H3K4me2, H3K4me3
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001237231	1	NHLF	H3K27ac, DNase1

(promoter flanking region)		embryonic stem cell (H1ESC) HSMMtube	DNase1, H3K36me3 H3K27ac, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K36me3, H3K4me1, DNase1
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3, H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		lung (IMR90)	DNase1, H3K18ac, H3K36me3, H2BK120ac
		nervous (NH-A)	DNase1, H3K9ac, H3K27ac, H3K36me3
		A549	H3K4me3, DNase1, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me2, H3K27ac, H3K4me1, DNase1
ENSR00001237239 (enhancer)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00001429723 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac, H3K36me3
		HSMMtube	DNase1
		nervous (NH-A)	H3K36me3, DNase1
		blood (K562)	H3K27me3
		A549	H3K36me3
		muscle (HSMM)	DNase1, H3K4me2, H3K27ac, H2AZ

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BST1	upstream gene variant	2095	ENST00000514989	?	ENSP00000424761	3
BST1	upstream gene variant	2212	ENST00000505785	?	ENSP00000423357	1
BST1	upstream gene variant	1461	ENST00000382346	?	ENSP00000371783	2
BST1	upstream gene variant	575	ENST00000514445	?	ENSP00000420925	2
BST1	upstream gene variant	1315	ENST00000265016	NM_004334.2	ENSP00000265016	2
CC2D2A	downstream gene variant	1001	ENST00000512202	?	?	5
CC2D2A	downstream gene variant	1080	ENST00000512702	?	ENSP00000422875	1
CC2D2A	downstream gene variant	991	ENST00000389652	?	ENSP00000374303	2
CC2D2A	upstream gene variant, downstream gene variant	1275	ENST00000513035	?	?	2
CC2D2A	downstream gene variant	1166	ENST00000514039	?	?	2
CC2D2A	downstream gene variant	985	ENST00000506643	?	ENSP00000422931	2
CC2D2A	upstream gene variant, downstream gene variant	2143	ENST00000510220	?	?	2
CC2D2A	downstream gene variant	980	ENST00000424120	?	ENSP00000403465	2
CC2D2A	downstream gene variant	980	ENST00000503292	NM_001080522.2	ENSP00000421809	2
FAM200B	upstream gene variant, downstream gene variant	375	ENST00000509022	?	?	6
FAM200B	upstream gene variant, downstream gene variant	964	ENST00000422728	?	ENSP00000393017	5
FAM200B	upstream gene variant, downstream gene variant	915	ENST00000503617	?	ENSP00000422751	5
FAM200B	upstream gene variant, downstream gene variant	914	ENST00000505260	?	?	5
FAM200B	upstream gene variant, downstream gene variant	914	ENST00000515430	?	?	5
FAM200B	upstream gene variant, downstream gene variant	529	ENST00000506610	?	?	5
FAM200B	upstream gene variant, downstream gene variant	1064	ENST00000510186	?	?	4

FAM200B	upstream gene variant, downstream gene variant	1019	ENST00000502856	?	?	4
FAM200B	upstream gene variant, downstream gene variant	413	ENST00000502502	?	?	6
FAM200B	upstream gene variant, downstream gene variant	891	ENST00000515697	?	?	5
FAM200B	upstream gene variant, downstream gene variant	993	ENST00000503600	?	?	5
FAM200B	upstream gene variant, downstream gene variant	983	ENST00000508567	?	?	5
FAM200B	upstream gene variant, downstream gene variant	827	ENST00000504823	?	?	5
FAM200B	upstream gene variant, downstream gene variant	375	ENST00000514803	?	?	6
FAM200B	upstream gene variant, downstream gene variant	914	ENST00000504598	?	?	5
FAM200B	upstream gene variant, downstream gene variant	646	ENST00000622362	NM_001145191.1	ENSP00000483930	5
FAM200B	upstream gene variant, downstream gene variant	914	ENST00000512855	?	?	5
FAM200B	upstream gene variant, downstream gene variant	820	ENST00000510920	?	?	5
FAM200B	upstream gene variant, downstream gene variant	638	ENST00000504137	?	?	5
FAM200B	upstream gene variant, downstream gene variant	585	ENST00000507992	?	?	6
FAM200B	upstream gene variant, downstream gene variant	949	ENST00000507305	?	?	5
FAM200B	upstream gene variant, downstream gene variant	375	ENST00000510032	?	?	6
FAM200B	upstream gene variant, downstream gene variant	964	ENST00000513053	?	?	5
FBXL5	downstream gene variant, upstream gene variant	414	ENST00000512066	?	ENSP00000426993	5
FBXL5	downstream gene variant, upstream gene variant	154	ENST00000507700	?	?	3
FBXL5	downstream gene variant, upstream gene variant	999	ENST00000515679	?	ENSP00000424423	6
FBXL5	downstream gene variant, upstream gene variant	2083	ENST00000514541	?	?	5
FBXL5	downstream gene variant, upstream gene variant	991	ENST00000507899	?	ENSP00000422049	6
FBXL5	downstream gene variant, upstream gene variant	349	ENST00000509314	?	ENSP00000421070	3
FBXL5	downstream gene variant, upstream gene variant	339	ENST00000503196	?	ENSP00000425541	3
FBXL5	downstream gene variant, upstream gene variant	606	ENST00000412094	NM_001193535.1	ENSP00000408679	4
FBXL5	downstream gene variant, upstream gene variant	490	ENST00000510802	?	ENSP00000423961	4
FBXL5	downstream gene variant, upstream gene variant	139	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	4
FBXL5	downstream gene variant, upstream gene variant	443	ENST00000513163	?	ENSP00000425472	6
FBXL5	downstream gene variant, upstream gene variant	606	ENST00000511441	?	ENSP00000425027	4
FBXL5	downstream gene variant, upstream gene variant	718	ENST00000504837	?	?	3
RP11-115L11.1	downstream gene variant	2236	ENST00000498116	?	?	1
RP11-799M12.2	downstream gene variant	2495	ENST00000609724	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BST1	ENST00000265016	NM_004334.2	ENSP00000265016	7
BST1	ENST00000505785	?	ENSP00000423357	5

BST1	ENST00000382346	?	ENSP00000371783	7
BST1	ENST00000514989	?	ENSP00000424761	3
BST1	ENST00000514445	?	ENSP00000420925	5
CC2D2A	ENST00000512202	?	?	1
CC2D2A	ENST00000512702	?	ENSP00000422875	2
CC2D2A	ENST00000389652	?	ENSP00000374303	14
CC2D2A	ENST00000514039	?	?	2
CC2D2A	ENST00000424120	?	ENSP00000403465	14
CC2D2A	ENST00000513811	?	?	3
CC2D2A	ENST00000510220	?	?	1
CC2D2A	ENST00000503292	NM_001080522.2	ENSP00000421809	14
CC2D2A	ENST00000506643	?	ENSP00000422931	12
FAM200B	ENST00000509022	?	?	3
FAM200B	ENST00000422728	?	ENSP00000393017	3
FAM200B	ENST00000505260	?	?	4
FAM200B	ENST00000503617	?	ENSP00000422751	3
FAM200B	ENST00000506610	?	?	1
FAM200B	ENST00000503600	?	?	4
FAM200B	ENST00000510920	?	?	4
FAM200B	ENST00000502502	?	?	3
FAM200B	ENST00000515697	?	?	4
FAM200B	ENST00000512855	?	?	4
FAM200B	ENST00000507992	?	?	3
FAM200B	ENST00000514803	?	?	3
FAM200B	ENST00000622362	NM_001145191.1	ENSP00000483930	3
FAM200B	ENST00000515430	?	?	4
FAM200B	ENST00000504823	?	?	4
FAM200B	ENST00000504598	?	?	8
FAM200B	ENST00000507305	?	?	4
FAM200B	ENST00000508567	?	?	4
FAM200B	ENST00000502856	?	?	4
FAM200B	ENST00000510032	?	?	3
FAM200B	ENST00000510186	?	?	4
FAM200B	ENST00000504137	?	?	4
FAM200B	ENST00000513053	?	?	4
FBXL5	ENST00000509314	?	ENSP00000421070	17
FBXL5	ENST00000504837	?	?	16
FBXL5	ENST00000514541	?	?	13
FBXL5	ENST00000507899	?	ENSP00000422049	27
FBXL5	ENST00000515679	?	ENSP00000424423	27
FBXL5	ENST00000510802	?	ENSP00000423961	29
FBXL5	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	33

FBXL5	ENST00000412094	NM_001193535.1	ENSP00000408679	33
FBXL5	ENST00000507700	?	?	32
FBXL5	ENST00000503196	?	ENSP00000425541	17
FBXL5	ENST00000513163	?	ENSP00000425472	23
FBXL5	ENST00000512066	?	ENSP00000426993	6
FBXL5	ENST00000511441	?	ENSP00000425027	33

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FBXL5	ENST00000509314	?	ENSP00000421070	1
FBXL5	ENST00000507899	?	ENSP00000422049	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FAM200B	ENST00000505260	?	1
FAM200B	ENST00000512855	?	1
FAM200B	ENST00000515430	?	1
FAM200B	ENST00000504137	?	1
FBXL5	ENST00000514541	?	1

Locus 19 (rs2058622)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,918,018-103,093,243
block size	175,226 bp
variant count	94 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.815 [-4.683 – 1.369]	gene(s) hit or close-by	AC007248.7, AC007278.2, AC007278.3, IL18R1, IL18RAP, IL1RL1, MIR4772, SLC9A4
phastCons	μ = 0.030 [0 – 0.992]	eQTL gene(s)	AC007248.7, AC007278.2, AC007278.3, CYP2C19, IL18R1, IL18RAP, IL1RL1, RP11-400G3.5, SLC9A4, ZNF32
GERP++	μ = -1.007 [-9.62 – 4.57]	potentially regulated gene(s)	AC007278.2, AC007278.3, IL18R1, IL18RAP, IL1R2, IL1RL1, SLC9A2, SLC9A4
CADD score	μ = 2.505 [0.044 – 14.43]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Serum protein levels (sST2)	$<1.00\times10^{-35}$	GWAS Catalog	23999434	1
Inflammatory bowel disease	$<3.00\times10^{-20}$	GWAS Catalog	23128233	1
Atopic dermatitis	$<8.00\times10^{-18}$	GWAS Catalog	23042114	1
Crohn's disease (time to surgery)	$<2.00\times10^{-12}$	GWAS Catalog	21102463	1
CELIAC DISEASE	$<1.00\times10^{-15}$	GWAS Catalog	20190752	1
CELIAC DISEASE	$<4.00\times10^{-9}$	GWAS Catalog	18311140	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL18R1	?	ENSG00000115604	caudate basal ganglia	4.85×10^{-7} (p-value)	GTEx Portal V6	6
IL1RL1	?	ENSG00000115602	caudate basal ganglia	3.05×10^{-6} (p-value)	GTEx Portal V6	1
IL18R1	?	ENSG00000115604	lung	1.25×10^{-7} (p-value)	GTEx Portal V6	93
IL1RL1	?	ENSG00000115602	lung	1.32×10^{-8} (p-value)	GTEx Portal V6	92
AC007278.3	?	ENSG00000234389	lung	1.51×10^{-7} (p-value)	GTEx Portal V6	81
IL18RAP	?	ENSG00000115607	lung	9.04×10^{-6} (p-value)	GTEx Portal V6	1
AC007248.7	?	ENSG00000236785	lung	1.19×10^{-5} (p-value)	GTEx Portal V6	2
AC007278.2	?	ENSG00000236525	blood	1.05×10^{-13} (p-value)	GTEx Portal V6	93
IL18RAP	?	ENSG00000115607	blood	4.09×10^{-31} (p-value)	GTEx Portal V6	93
AC007278.3	?	ENSG00000234389	blood	9.36×10^{-17} (p-value)	GTEx Portal V6	93
IL18R1	?	ENSG00000115604	blood	4.67×10^{-6} (p-value)	GTEx Portal V6	38

IL18R1	?	ENSG00000115604	skeletal muscle	7.19×10-7 (p-value)	GTEx Portal V6	92
SLC9A4	?	ENSG00000180251	testis	2.01×10-7 (p-value)	GTEx Portal V6	92
IL18R1	?	ENSG00000115604	sun exposed skin	9.57×10-10 (p-value)	GTEx Portal V6	92
IL18R1	?	ENSG00000115604	left ventricle	2.14×10-6 (p-value)	GTEx Portal V6	55
IL18R1	?	ENSG00000115604	hippocampus	4.34×10-7 (p-value)	GTEx Portal V6	88
IL18R1	?	ENSG00000115604	cerebellum	1.83×10-6 (p-value)	GTEx Portal V6	30
IL18R1	ENST00000409599	ILMN_1781700	blood	6.65×10-5 (p-value)	MuTHER consortium	1
IL18R1	ENST00000233957					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ZNF32	ENST00000395797	ILMN_1680072	chr10	blood	1.31×10-8 (p-value)	Westra et al.	3
ZNF32	ENST00000374433						
CYP2C19	ENST00000371321	ILMN_1729191	chr10	blood	1.18×10-13 (p-value)	Westra et al.	3
RP11-400G3.5	ENST00000464755						

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000493988	1	ENST00000410040	IL18R1
		ENST00000404917, ENST00000473175, ENST00000447231, ENST00000233954	IL1RL1
FFCP00000494102	1	ENST00000409369, ENST00000450855	IL18RAP

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000027320		
ENCP00000027322		
ENCP00000027323		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000257210	2	ENCP00000027298	IL1R2
		ENCP00000027315	IL18R1 IL1RL1
		ENCP00000027324	AC007278.3
		ENCP00000027320	IL18RAP
		ENCP00000027322	IL18RAP
		ENCP00000027326	SLC9A4
		ENCP00000027328	SLC9A2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001655316 (CTCF binding site)	2	monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
ENSR00001045581 (CTCF binding site)	1	HSMMtube	H3K27me3
		blood (K562)	H3K27me3
ENSR00000303075 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		HSMMtube	H3K27me3
		A549	H3K27me3

		blood (K562)	Ini1, DNase1, H2AZ, H3K4me3
ENSR00001655328 (enhancer)	2	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K4me1
		monocytes (Monocytes-CD14+)	H3K4me1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001655330 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27me3
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	DNase1
		blood (GM12878)	BATF, NFKB, H3K4me2
		A549	H3K27me3
ENSR00000595668 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	DNase1
		blood (GM12878)	BATF, NFKB, H3K4me2
		A549	H3K27me3
ENSR00001544959 (promoter flanking region)	1	NHLF	DNase1
		liver (HepG2)	H3K27me3
		HSMMtube	H3K27me3, DNase1
		blood (DND-41)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	DNase1
		muscle (HSMM)	DNase1
ENSR00000677145 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF, H3K27me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00000595671 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF, H3K27me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF

cervix (HeLa-S3)	CTCF
monocytes (Monocytes-CD14+)	CTCF
endothelium (HUVEC)	H3K36me3, CTCF
A549	CTCF, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC007248.7	upstream gene variant, downstream gene variant	600	ENST00000430344	?	?	6
AC007278.2	upstream gene variant, downstream gene variant	406	ENST00000436582	?	?	10
AC007278.3	upstream gene variant, downstream gene variant	152	ENST00000450893	?	?	10
IL18R1	upstream gene variant, downstream gene variant	524	ENST00000409599	?	ENSP00000387211	2
IL18R1	downstream gene variant	740	ENST00000334376	?	ENSP00000334030	5
IL18R1	upstream gene variant, downstream gene variant	263	ENST00000410040	?	ENSP00000386663	4
IL18R1	upstream gene variant, downstream gene variant	640	ENST00000466357	?	?	5
IL18R1	upstream gene variant, downstream gene variant	878	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	2
IL18RAP	upstream gene variant, downstream gene variant	32	ENST00000497795	?	?	5
IL18RAP	upstream gene variant, downstream gene variant	105	ENST00000264260	NM_003853.2	ENSP00000264260	5
IL18RAP	upstream gene variant, downstream gene variant	861	ENST00000450855	?	ENSP00000389815	3
IL18RAP	upstream gene variant, downstream gene variant	1102	ENST00000409369	?	ENSP00000387201	5
IL1RL1	upstream gene variant	1867	ENST00000409584	?	ENSP00000386618	1
IL1RL1	upstream gene variant	1859	ENST00000463990	?	?	1
IL1RL1	upstream gene variant	302	ENST00000447231	?	ENSP00000409437	3
IL1RL1	upstream gene variant	1890	ENST00000482701	?	?	1
IL1RL1	upstream gene variant	1897	ENST00000427077	?	ENSP00000391120	1
IL1RL1	upstream gene variant, downstream gene variant	1757	ENST00000311734	NM_003856.3	ENSP00000310371	3
IL1RL1	upstream gene variant, downstream gene variant	236	ENST00000233954	NM_016232.4	ENSP00000233954	4
IL1RL1	upstream gene variant	263	ENST00000473175	?	?	3
IL1RL1	upstream gene variant, downstream gene variant	236	ENST00000404917	NM_001282408.1	ENSP00000384822	4
MIR4772	upstream gene variant, downstream gene variant	282	ENST00000581495	?	?	10
SLC9A4	upstream gene variant	2993	ENST00000295269	NM_001011552.3	ENSP00000295269	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC007278.2	ENST00000436582	?	?	2
IL18R1	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	22
IL18R1	ENST00000334376	?	ENSP00000334030	11
IL18R1	ENST00000410040	?	ENSP00000386663	27
IL18R1	ENST00000409599	?	ENSP00000387211	22
IL18RAP	ENST00000409369	?	ENSP00000387201	26
IL18RAP	ENST00000264260	NM_003853.2	ENSP00000264260	26

IL18RAP	ENST00000264260	NM_003853.2	ENSP00000264260	26
IL18RAP	ENST00000450855	?	ENSP00000389815	5
IL18RAP	ENST00000497795	?	?	2
IL1RL1	ENST00000404917	NM_001282408.1	ENSP00000384822	3
IL1RL1	ENST00000447231	?	ENSP00000409437	3
IL1RL1	ENST00000233954	NM_016232.4	ENSP00000233954	4
IL1RL1	ENST00000473175	?	?	3
SLC9A4	ENST00000295269	NM_001011552.3	ENSP00000295269	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL18R1	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	1
IL18R1	ENST00000409599	?	ENSP00000387211	1
IL18RAP	ENST00000409369	?	ENSP00000387201	2
IL18RAP	ENST00000264260	NM_003853.2	ENSP00000264260	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC007278.2	ENST00000436582	?	1
AC007278.3	ENST00000450893	?	1

Locus 20 (rs12141375)

SNiPAcard

Block annotations

Block info

genomic range	chr1:203,173,094-203,193,134
block size	20,041 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.357 [-3.015 – 0.381]	**gene(s) hit or close-by**	CHIT1
phastCons	μ = 0.005 [0 – 0.039]	**eQTL gene(s)**	ADORA1, CHIT1
GERP++	μ = -1.325 [-4.67 – 2.4]	**potentially regulated gene(s)**	–
CADD score	μ = 3.377 [0.095 – 11.08]	**disease gene(s)**	CHIT1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CHIT1	CHITOTRIOSIDASE DEFICIENCY	OMIM	MIM:614122

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ADORA1	?	ENSG00000163485	transformed fibroblasts	1.36×10^{-6} (p-value)	GTEx Portal V6	12
CHIT1	?	ENSG00000133063	blood	1.71×10^{-8} (p-value)	GTEx Portal V6	14
CHIT1	?	ENSG00000133063	cortex	3.81×10^{-7} (p-value)	GTEx Portal V6	8

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001527715 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	H3K4me1, H3K27ac, H3K36me3, DNase1
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	DNase1, H3K4me1, H3K18ac, H3K27ac, H4K5ac
		nervous (NH-A)	DNase1
		A549	H3K27me3
ENSR00001527718 (open chromatin region)	2	endothelium (HUVEC)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	H3K27ac, H3K9ac, H3K36me3, DNase1

ENSR00001527719 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	H3K27ac, DNase1
ENSR00001589716 (enhancer)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21
		HSMMtube	CTCF
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF, H3K27me3
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, H3K27me3, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3, DNase1, CTCF
		endothelium (HUVEC)	H3K27me3
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CHIT1	downstream gene variant, upstream gene variant	609	ENST00000506427	?	?	3
CHIT1	downstream gene variant	1032	ENST00000255427	NM_001256125.1	ENSP00000255427	2
CHIT1	downstream gene variant	593	ENST00000484834	?	?	4
CHIT1	downstream gene variant, upstream gene variant	601	ENST00000479483	?	?	6
CHIT1	downstream gene variant	1032	ENST00000491855	?	ENSP00000423778	2
CHIT1	downstream gene variant	441	ENST00000367229	NM_001270509.1, NM_003465.2	ENSP00000356198	2
CHIT1	downstream gene variant	1310	ENST00000503786	?	ENSP00000421617	2
CHIT1	downstream gene variant	1454	ENST00000513472	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CHIT1	ENST00000506427	?	?	1
CHIT1	ENST00000255427	NM_001256125.1	ENSP00000255427	3
CHIT1	ENST00000484834	?	?	4
CHIT1	ENST00000479483	?	?	1
CHIT1	ENST00000491855	?	ENSP00000423778	3
CHIT1	ENST00000367229	NM_001270509.1, NM_003465.2	ENSP00000356198	3
CHIT1	ENST00000503786	?	ENSP00000421617	3
CHIT1	ENST00000513472	?	?	1

Locus 21 (rs9326248)

SNiPAcard

Block annotations

Block info

genomic range	chr11:116,999,225-117,081,974
block size	82,750 bp
variant count	38 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.268 [-2.147 – 0.86]	gene(s) hit or close-by	AP000892.6, PAFAH1B2, PCSK7, RP11-109L13.1, RP11-109L13.5, SIDT2, TAGLN
phastCons	μ = 0.042 [0 – 0.378]	eQTL gene(s)	CBLN2, PAFAH1B2, PCSK7, RP11-109L13.1, SIDT2, TAGLN
GERP++	μ = -0.957 [-6.23 – 2.63]	potentially regulated gene(s)	PCSK7, RP11-109L13.1, SIK3, TAGLN
CADD score	μ = 3.689 [0.184 – 10.77]	disease gene(s)	CBLN2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1,6-anhydroglucose	9.23×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
CBLN2	Heritable pulmonary arterial hypertension	OrphaNet	OrphaNet:275777
CBLN2	Idiopathic pulmonary arterial hypertension	OrphaNet	OrphaNet:275766

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TAGLN	ENST00000525531	ILMN_1778668	blood	2.72×10^{-21} (p-value)	MuTHER consortium	14
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
TAGLN	ENST00000525531	ILMN_2400935	blood	3.94×10^{-7} (p-value)	MuTHER consortium	14
TAGLN	ENST00000529622					
TAGLN	ENST00000278968					
TAGLN	ENST00000530649					
TAGLN	ENST00000529792					
TAGLN	ENST00000532870					
TAGLN	ENST00000392951					
TAGLN	ENST00000533863					
PCSK7	ENST00000540028	ILMN_1699545	skin	5.96×10^{-6} (p-value)	MuTHER consortium	14
PCSK7	ENST00000529458		blood	5.70×10^{-6} (p-value)	MuTHER consortium	14
PCSK7	ENST00000534529					
PCSK7	ENST00000320934					

PCSK7	ENST00000527037					
TAGLN	ENST00000525531	ILMN_1706783	adipocyte	3.60×10-5 (p-value)	MuTHER consortium	8
TAGLN	ENST00000278968					
RP11-109L13.1	?	ENSG00000254851	atrial appendage	8.55×10-30 (p-value)	GTEx Portal V6	35
PCSK7	?	ENSG00000160613	transformed fibroblasts	2.41×10-13 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	transformed fibroblasts	1.29×10-20 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	breast	8.69×10-32 (p-value)	GTEx Portal V6	35
TAGLN	?	ENSG00000149591	blood	6.45×10-45 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	blood	7.27×10-21 (p-value)	GTEx Portal V6	35
SIDT2	?	ENSG00000149577	blood	1.38×10-11 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	skeletal muscle	1.07×10-26 (p-value)	GTEx Portal V6	35
TAGLN	?	ENSG00000149591	EBV lymphocytes	5.99×10-11 (p-value)	GTEx Portal V6	30
RP11-109L13.1	?	ENSG00000254851	unexposed skin	1.92×10-32 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	sun exposed skin	7.36×10-47 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	ovary	4.30×10-14 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	left ventricle	3.41×10-17 (p-value)	GTEx Portal V6	35
PCSK7	?	ENSG00000160613	aorta	7.38×10-15 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	aorta	8.98×10-38 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	subcutaneous adipocytes	5.16×10-61 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	visceral adipocytes	1.37×10-38 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	stomach	2.76×10-26 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	tibial nerve	1.97×10-48 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	esophagus mucosa	1.49×10-36 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	pituitary	5.84×10-13 (p-value)	GTEx Portal V6	35
TAGLN	?	ENSG00000149591	testis	3.46×10-6 (p-value)	GTEx Portal V6	4
RP11-109L13.1	?	ENSG00000254851	testis	5.08×10-15 (p-value)	GTEx Portal V6	34
SIDT2	?	ENSG00000149577	testis	2.87×10-6 (p-value)	GTEx Portal V6	12
TAGLN	?	ENSG00000149591	pancreas	4.73×10-7 (p-value)	GTEx Portal V6	27
RP11-109L13.1	?	ENSG00000254851	pancreas	2.07×10-9 (p-value)	GTEx Portal V6	32
SIDT2	?	ENSG00000149577	pancreas	1.91×10-7 (p-value)	GTEx Portal V6	25
RP11-109L13.1	?	ENSG00000254851	uterus	1.40×10-11 (p-value)	GTEx Portal V6	34
PCSK7	?	ENSG00000160613	muscularis mucosae	4.73×10-11 (p-value)	GTEx Portal V6	34
RP11-109L13.1	?	ENSG00000254851	muscularis mucosae	1.97×10-41 (p-value)	GTEx Portal V6	35
PCSK7	?	ENSG00000160613	lung	8.44×10-13 (p-value)	GTEx Portal V6	34
RP11-109L13.1	?	ENSG00000254851	lung	3.29×10-45 (p-value)	GTEx Portal V6	35
SIDT2	?	ENSG00000149577	lung	9.70×10-11 (p-value)	GTEx Portal V6	32

PAFAH1B2	?	ENSG00000168092	lung	3.46×10-6 (p-value)	GTEx Portal V6	11
TAGLN	?	ENSG00000149591	lung	6.43×10-6 (p-value)	GTEx Portal V6	2
PCSK7	?	ENSG00000160613	tibial artery	7.26×10-18 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	tibial artery	2.13×10-54 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	thyroid	7.60×10-39 (p-value)	GTEx Portal V6	35
SIDT2	?	ENSG00000149577	thyroid	1.81×10-6 (p-value)	GTEx Portal V6	22
RP11-109L13.1	?	ENSG00000254851	prostate	1.94×10-20 (p-value)	GTEx Portal V6	34
RP11-109L13.1	?	ENSG00000254851	transverse colon	7.57×10-24 (p-value)	GTEx Portal V6	35
SIDT2	ENST00000278951	ILMN_1791912	monocyte	2.57×10-124 (p-value)	Zeller et al.	3
SIDT2	ENST00000532062					
SIDT2	ENST00000324225					
SIDT2	ENST00000431081					
SIDT2	ENST00000620360					
TAGLN	ENST00000525531	ILMN_1778668	monocyte	9.39×10-197 (p-value)	Zeller et al.	3
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
TAGLN	ENST00000525531	ILMN_2400935	monocyte	4.10×10-99 (p-value)	Zeller et al.	3
TAGLN	ENST00000529622					
TAGLN	ENST00000278968					
TAGLN	ENST00000530649					
TAGLN	ENST00000529792					
TAGLN	ENST00000532870					
TAGLN	ENST00000392951					
TAGLN	ENST00000533863					
RP11-109L13.1	?	ENSG00000254851	terminal ileum	7.72×10-14 (p-value)	GTEx Portal V6	33
RP11-109L13.1	?	ENSG00000254851	spleen	2.70×10-11 (p-value)	GTEx Portal V6	28
PCSK7	?	ENSG00000160613	coronary artery	8.08×10-8 (p-value)	GTEx Portal V6	6
RP11-109L13.1	?	ENSG00000254851	coronary artery	6.64×10-30 (p-value)	GTEx Portal V6	35
PCSK7	?	ENSG00000160613	sigmoid colon	1.18×10-9 (p-value)	GTEx Portal V6	33
RP11-109L13.1	?	ENSG00000254851	sigmoid colon	2.48×10-23 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	vagina	2.61×10-14 (p-value)	GTEx Portal V6	34
TAGLN	ENST00000525531	ILMN_1706783	blood	5.26×10-7 (p-value)	Westra et al.	14
TAGLN	ENST00000278968					
RP11-109L13.1	?	ENSG00000254851	gastroesophageal junction	5.10×10-26 (p-value)	GTEx Portal V6	35
RP11-109L13.1	?	ENSG00000254851	adrenal gland	1.03×10-21 (p-value)	GTEx Portal V6	35
PAFAH1B2	?	ENSG00000168092	cerebellum	1.07×10-7 (p-value)	GTEx Portal V6	30
SIDT2	ENST00000278951	ILMN_1791912	monocyte	1.60×10-42 (p-value)	Fairfax et al.	4
SIDT2	ENST00000532062		b-cell	1.35×10-29 (p-value)	Fairfax et al.	4
SIDT2	ENST00000324225					

SIDT2	ENST00000431081						
SIDT2	ENST00000620360						
TAGLN	ENST00000525531	ILMN_2400935	monocyte	9.51×10^{-19} (p-value)	Fairfax et al.	4
TAGLN	ENST00000529622		b-cell	1.46×10^{-14} (p-value)	Fairfax et al.	4
TAGLN	ENST00000278968						
TAGLN	ENST00000530649						
TAGLN	ENST00000529792						
TAGLN	ENST00000532870						
TAGLN	ENST00000392951						
TAGLN	ENST00000533863						

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CBLN2	ENST00000581425	ILMN_1796069	chr18	monocyte	1.54×10^{-12} (p-value)	Zeller et al.	2
CBLN2	ENST00000585159						
CBLN2	ENST00000269503						

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000014774		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000123647	1	ENCP00000014794	PCSK7
ENCE00000123461	1	ENCP00000014767	SIK3
ENCE00000123486	1	ENCP00000014768	SIK3
ENCE00000123604	1	ENCP00000014791	PCSK7
		ENCP00000014787	TAGLN

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001607349 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Nrsf, CTCF
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, DNase1
		blood (K562)	ZBTB7A, Rad21, CTCF
		skin (NHDF-AD)	CTCF, H3K4me2
		muscle (HSMM)	H3K4me2, H3K4me3, CTCF
		liver (HepG2)	CTCF
		lung (IMR90)	H3K14ac, H4K8ac, H3K27ac, H3K18ac, H3K4me2, CTCF, H3K56ac, H4K91ac, H3K9ac, H2AK5ac, H3K4ac, H3K36me3, H4K5ac
		blood (GM12878)	CTCF
		nervous (NH-A)	H3K4me2, H3K9ac
		skin (NHEK)	CTCF
		NHLF	H3K27ac, CTCF
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00000569571 (promoter)	4	embryonic stem cell (H1ESC)	DNase1, H3K9ac, CTCF, Yy1, TAF1, H3K4me2, Srf, H3K4me3, H3K36me3, H3K27me3, PolII, Rad21, Jund, TAF7, Nrsf
		HSMMtube	H3K4me2, H3K27ac, H3K36me3, DNase1, H3K9ac
		blood (K562)	ZBTB7A, Rad21, CTCF, H3K36me3

		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K79me2, CTCF, H3K4me3, H3K4me2, H3K36me3
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF, PolII
		lung (IMR90)	H3K27ac, H3K79me2, H3K14ac, H4K8ac, DNase1, H3K18ac, CTCF, H3K56ac, H4K91ac, H3K9ac, H2AK5ac, H3K4ac, H3K4me3, H3K36me3, H4K5ac, H3K4me2
		nervous (NH-A)	DNase1, H3K36me3, H3K4me3, H3K4me2, H4K20me1, H3K9ac
		skin (NHEK)	CTCF, DNase1
		NHLF	H3K9ac, CTCF, H3K4me3, DNase1, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		breast (HMEC)	CTCF, H3K4me3
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00000321754	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, Rad21, CTCF
(CTCF binding site)		HSMMtube	H3K36me3
		blood (K562)	Rad21, H2AZ, CTCF, H3K36me3
		muscle (HSMM)	H3K36me3
		liver (HepG2)	H3K4me3, H3K4me2, CTCF
		lung (IMR90)	H3K36me3
		blood (GM12878)	PolII, H3K36me3
		nervous (NH-A)	H3K36me3, H4K20me1
		skin (NHEK)	H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001573939	1	embryonic stem cell (H1ESC)	H3K36me3, PolII, Rad21, CTCF, DNase1
(CTCF binding site)		HSMMtube	H3K36me3, DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3, H3K27me3
		blood (K562)	CTCF, H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, CTCF
		A549	H3K36me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP000892.6	upstream gene variant, downstream gene variant	755	ENST00000624094	?	?	7
PAFAH1B2	upstream gene variant	2799	ENST00000304808	?	ENSP00000304006	4
PAFAH1B2	upstream gene variant, downstream gene variant	827	ENST00000419197	NM_001184748.1	ENSP00000388742	6
PAFAH1B2	upstream gene variant, downstream gene variant	1564	ENST00000530272	NM_001184746.1	ENSP00000431365	6
PAFAH1B2	downstream gene variant	146	ENST00000533677	?	?	2
PAFAH1B2	upstream gene variant, downstream gene variant	807	ENST00000526888	?	?	6
PAFAH1B2	upstream gene variant	2888	ENST00000533206	?	?	4
PAFAH1B2	upstream gene variant, downstream gene variant	1279	ENST00000529887	NM_001184747.1	ENSP00000434951	6
PAFAH1B2	upstream gene variant	2773	ENST00000527958	NM_002572.3	ENSP00000435289	4
PCSK7	downstream gene variant	2090	ENST00000320934	NM_004716.2	ENSP00000325917	5
PCSK7	downstream gene variant	2681	ENST00000528973	?	?	1
PCSK7	downstream gene variant	2827	ENST00000534529	?	?	5

PCSK7	downstream gene variant, upstream gene variant	36	ENST00000527861	?	?	6
PCSK7	downstream gene variant	2831	ENST00000540028	?	ENSP00000441944	5
PCSK7	downstream gene variant, upstream gene variant	229	ENST00000531573	?	?	4
PCSK7	downstream gene variant, upstream gene variant	782	ENST00000532810	?	?	5
PCSK7	downstream gene variant, upstream gene variant	581	ENST00000529458	?	?	5
PCSK7	downstream gene variant	156	ENST00000527037	?	?	3
PCSK7	downstream gene variant	1783	ENST00000533135	?	?	1
RP11-109L13.1	upstream gene variant, downstream gene variant	944	ENST00000528464	?	?	11
RP11-109L13.5	downstream gene variant, upstream gene variant	1917	ENST00000525475	?	?	7
SIDT2	upstream gene variant, downstream gene variant	618	ENST00000532960	?	ENSP00000431176	4
SIDT2	upstream gene variant, downstream gene variant	900	ENST00000431081	?	ENSP00000399635	7
SIDT2	upstream gene variant, downstream gene variant	999	ENST00000628876	?	ENSP00000486114	6
SIDT2	upstream gene variant, downstream gene variant	587	ENST00000529441	?	?	6
SIDT2	upstream gene variant, downstream gene variant	1987	ENST00000531353	?	ENSP00000435004	3
SIDT2	upstream gene variant, downstream gene variant	189	ENST00000524988	?	?	5
SIDT2	upstream gene variant, downstream gene variant	1668	ENST00000524842	?	ENSP00000436983	2
SIDT2	upstream gene variant, downstream gene variant	975	ENST00000531255	?	?	5
SIDT2	upstream gene variant, downstream gene variant	1480	ENST00000525339	?	?	3
SIDT2	upstream gene variant, downstream gene variant	2048	ENST00000526813	?	?	4
SIDT2	upstream gene variant, downstream gene variant	912	ENST00000529484	?	?	4
SIDT2	upstream gene variant, downstream gene variant	995	ENST00000528397	?	?	4
SIDT2	upstream gene variant, downstream gene variant	972	ENST00000525065	?	?	4
SIDT2	upstream gene variant, downstream gene variant	904	ENST00000620360	?	ENSP00000482762	7
SIDT2	upstream gene variant, downstream gene variant	1066	ENST00000532062	?	ENSP00000432432	7
SIDT2	upstream gene variant, downstream gene variant	901	ENST00000278951	?	ENSP00000278951	7
SIDT2	upstream gene variant, downstream gene variant	479	ENST00000525478	?	ENSP00000435890	6
SIDT2	upstream gene variant, downstream gene variant	903	ENST00000324225	NM_001040455.1	ENSP00000314023	7
SIDT2	upstream gene variant, downstream gene variant	926	ENST00000530948	?	?	5
SIDT2	upstream gene variant, downstream gene variant	155	ENST00000527654	?	?	4
TAGLN	upstream gene variant, downstream gene variant	1049	ENST00000278968	NM_001001522.1	ENSP00000278968	5
TAGLN	upstream gene variant, downstream gene variant	1216	ENST00000530649	?	ENSP00000431941	7
TAGLN	upstream gene variant, downstream gene variant	812	ENST00000533863	?	?	7
TAGLN	upstream gene variant, downstream gene variant	897	ENST00000529622	?	ENSP00000432380	8
TAGLN	upstream gene variant, downstream gene variant	64	ENST00000532870	?	ENSP00000432282	8
TAGLN	upstream gene variant, downstream gene variant	976	ENST00000392951	NM_003186.3	ENSP00000376678	5
TAGLN	upstream gene variant, downstream gene variant	76	ENST00000529792	?	ENSP00000431862	5
TAGLN	upstream gene variant, downstream gene variant	1037	ENST00000525531	?	ENSP00000432054	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PAFAH1B2	ENST00000530272	NM_001184746.1	ENSP00000431365	7
PAFAH1B2	ENST00000529887	NM_001184747.1	ENSP00000434951	7
PAFAH1B2	ENST00000526888	?	?	7
PAFAH1B2	ENST00000419197	NM_001184748.1	ENSP00000388742	7
PAFAH1B2	ENST00000304808	?	ENSP00000304006	7
PAFAH1B2	ENST00000533206	?	?	7
PAFAH1B2	ENST00000527958	NM_002572.3	ENSP00000435289	7
PCSK7	ENST00000532810	?	?	1
PCSK7	ENST00000320934	NM_004716.2	ENSP00000325917	3
PCSK7	ENST00000540028	?	ENSP00000441944	3
PCSK7	ENST00000534529	?	?	3
PCSK7	ENST00000527037	?	?	3
PCSK7	ENST00000533135	?	?	3
SIDT2	ENST00000525339	?	?	5
SIDT2	ENST00000524842	?	ENSP00000436983	4
SIDT2	ENST00000526813	?	?	1
SIDT2	ENST00000532062	?	ENSP00000432432	3
SIDT2	ENST00000528397	?	?	1
SIDT2	ENST00000531353	?	ENSP00000435004	2
SIDT2	ENST00000525478	?	ENSP00000435890	2
SIDT2	ENST00000431081	?	ENSP00000399635	8
SIDT2	ENST00000278951	?	ENSP00000278951	8
SIDT2	ENST00000530948	?	?	1
SIDT2	ENST00000532960	?	ENSP00000431176	1
SIDT2	ENST00000628876	?	ENSP00000486114	7
SIDT2	ENST00000324225	NM_001040455.1	ENSP00000314023	8
SIDT2	ENST00000620360	?	ENSP00000482762	8
SIDT2	ENST00000529441	?	?	1
TAGLN	ENST00000278968	NM_001001522.1	ENSP00000278968	5
TAGLN	ENST00000530649	?	ENSP00000431941	2
TAGLN	ENST00000392951	NM_003186.3	ENSP00000376678	5
TAGLN	ENST00000529792	?	ENSP00000431862	3
TAGLN	ENST00000525531	?	ENSP00000432054	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PCSK7	ENST00000320934	NM_004716.2	ENSP00000325917	1
PCSK7	ENST00000540028	?	ENSP00000441944	1
SIDT2	ENST00000532062	?	ENSP00000432432	1
SIDT2	ENST00000431081	?	ENSP00000399635	1
SIDT2	ENST00000324225	NM_001040455.1	ENSP00000314023	1
SIDT2	ENST00000620360	?	ENSP00000482762	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TAGLN	ENST00000532870	?	ENSP00000432282	1
TAGLN	ENST00000530649	?	ENSP00000431941	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AP000892.6	ENST00000624094	?	2
PAFAH1B2	ENST00000533206	?	1
PCSK7	ENST00000529458	?	1
PCSK7	ENST00000528973	?	3
PCSK7	ENST00000534529	?	4
SIDT2	ENST00000529484	?	1
SIDT2	ENST00000531255	?	1
TAGLN	ENST00000533863	?	2

Locus 22 (rs1993373)

SNiPAcard

Block annotations

Block info

genomic range	chr11:18,277,763-18,287,750
block size	9,988 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.167 [-2.326 – -0.307]	gene(s) hit or close-by	RNA5SP334, SAA1, ST13P5
phastCons	μ = 0.001 [0 – 0.004]	eQTL gene(s)	GTF2H1, SAA1, SAA2, SAA2-SAA4
GERP++	μ = -1.923 [-6.95 – 1.81]	potentially regulated gene(s)	HPS5, PTPN5, SAA1, TSG101
CADD score	μ = 1.247 [0.123 – 3.047]	disease gene(s)	SAA1, HPS5

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Amyloid A serum levels	$<3.00\times10^{-53}$	GWAS Catalog	21124955	1

Disease gene annotation

gene	trait	source DB	source entry/link
SAA1	Secondary amyloidosis	OrphaNet	OrphaNet:85445
HPS5	Hermansky-Pudlak syndrome without pulmonary fibrosis	OrphaNet	OrphaNet:231512
HPS5	HERMANSKY-PUDLAK SYNDROME 5	OMIM	MIM:614074

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SAA1	?	ENSG00000173432	tibial artery	6.31×10^{-7} (p-value)	GTEx Portal V6	6
SAA2	?	ENSG00000134339	tibial artery	3.39×10^{-6} (p-value)	GTEx Portal V6	6
SAA2-SAA4	?	ENSG00000255071	tibial artery	8.31×10^{-6} (p-value)	GTEx Portal V6	6
GTF2H1	ENST00000534641	ILMN_2157957	blood	7.08×10^{-6} (p-value)	Westra et al.	4
GTF2H1	ENST00000265963					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000055282	1	ENST00000356524, ENST00000532858, ENST00000405158	SAA1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000100066	1	ENCP00000011406	HPS5
		ENCP00000011423	TSG101
		ENCP00000011443	PTPN5

Locus 22 (sentinel SNP rs1993373 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000559405 (enhancer)	1	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	H3K4me1
		Osteobl	H3K27me3
		blood (GM12878)	H3K4me1
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001568722 (enhancer)	2	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		breast (HMEC)	H3K4me1
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RNA5SP334	upstream gene variant	190	ENST00000364825	?	?	3
SAA1	upstream gene variant	687	ENST00000356524	NM_199161.3	ENSP00000348918	2
SAA1	upstream gene variant	61	ENST00000405158	NM_000331.4	ENSP00000384906	3
SAA1	upstream gene variant	57	ENST00000532858	NM_001178006.1	ENSP00000436866	3
ST13P5	upstream gene variant, downstream gene variant	1136	ENST00000526214	?	?	6

Putative effect on transcript

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SAA1	ENST00000356524	NM_199161.3	ENSP00000348918	1

Locus 23 (rs2830585)

SNiPAcard

Block annotations

Block info

genomic range	chr21:28,280,322-28,329,010
block size	48,689 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.713 [-1.94 – 7.487]	gene(s) hit or close-by	ADAMTS5, AP001601.2, MIR4759
phastCons	μ = 0.127 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 0.144 [-6.12 – 6.03]	potentially regulated gene(s)	–
CADD score	μ = 7.436 [1.089 – 22.1]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ADAMTS5	missense variant	ENST00000284987	NM_007038.3	ENSP00000284987	H/R	cAc/cGc	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000612277 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3
		cervix (HeLa-S3)	DNase1, Cjun, Jund, Max
		monocytes (Monocytes-CD14+)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MIR4759	upstream gene variant, downstream gene variant	4	ENST00000584048	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ADAMTS5	ENST00000284987	NM_007038.3	ENSP00000284987	6
AP001601.2	ENST00000426771	?	?	6

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ADAMTS5	ENST00000284987	NM_007038.3	ENSP00000284987	2

Locus 24 (rs13294)

SNiPAcard

Block annotations

Block info

genomic range	chr1:150,267,798-150,511,628
block size	243,831 bp
variant count	138 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.549 [-3.729 – 1.45]	gene(s) hit or close-by	AC242988.1, ECM1, FALEC, MRPS21, PRPF3, RPRD2, TARS2
phastCons	μ = 0.061 [0 – 1]	eQTL gene(s)	ADAMTSL4, CDC42SE1, ECM1, ENSA, MRPS21, RP11-54A4.2, RPRD2, TARS2
GERP++	μ = -0.256 [-7.72 – 4.17]	potentially regulated gene(s)	ANXA9, APH1A, MCL1, RP11-235D19.2, VPS45
CADD score	μ = 3.547 [0.014 – 16.23]	disease gene(s)	ADAMTSL4, TARS2, ECM1, PRPF3, VPS45

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ADAMTSL4	ECTOPIA LENTIS ET PUPILLAE	OMIM	MIM:225200
TARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 21	OMIM	MIM:615918
ECM1	LIPOID PROTEINOSIS OF URBACH AND WIETHE	OMIM	MIM:247100
PRPF3	RETINITIS PIGMENTOSA 18	OMIM	MIM:601414
VPS45	NEUTROPENIA, SEVERE CONGENITAL, 5, AUTOSOMAL RECESSIVE	OMIM	MIM:615285
ADAMTSL4	isolated ectopia lentis (EL)	OrphaNet	OrphaNet:1885
ECM1	Lipoid proteinosis	OrphaNet	OrphaNet:530
PRPF3	Retinitis pigmentosa	OrphaNet	OrphaNet:791
VPS45	Recurrent infections-myelofibrosis-nephromegaly syndrome	OrphaNet	OrphaNet:369852

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ECM1	missense variant	ENST00000346569	NM_022664.2	ENSP00000271630	2	2			2
ECM1	missense variant	ENST00000369049	NM_001202858.1	ENSP00000358045	2	2			2
ECM1	missense variant	ENST00000369047	NM_004425.3	ENSP00000358043	2	2			2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MRPS21	ENST00000581066	ILMN_1660292	blood	1.51×10^{-8} (p-value)	MuTHER consortium	32
			adipocyte	2.23×10^{-20} (p-value)	MuTHER consortium	32
MRPS21	ENST00000581066	ILMN_1655765	blood	8.19×10^{-7} (p-value)	MuTHER consortium	32

MRPS21	ENST00000614145					
CDC42SE1	ENST00000540998	ILMN_2349138	blood	5.88×10-5 (p-value)	MuTHER consortium	2
CDC42SE1	ENST00000483763					
CDC42SE1	ENST00000357235					
CDC42SE1	ENST00000439374					
CDC42SE1	ENST00000483763	ILMN_1769027	blood	5.62×10-6 (p-value)	MuTHER consortium	32
CDC42SE1	ENST00000357235					
CDC42SE1	ENST00000540998					
CDC42SE1	ENST00000439374					
TARS2	?	ENSG00000143374	visceral adipocytes	1.00×10-6 (p-value)	GTEx Portal V6	125
RPRD2	?	ENSG00000163125	blood	9.70×10-6 (p-value)	GTEx Portal V6	56
TARS2	?	ENSG00000143374	breast	5.13×10-7 (p-value)	GTEx Portal V6	126
RP11-54A4.2	?	ENSG00000237781	tibial artery	4.92×10-8 (p-value)	GTEx Portal V6	131
ADAMTSL4	?	ENSG00000143382	tibial artery	4.27×10-7 (p-value)	GTEx Portal V6	80
TARS2	?	ENSG00000143374	tibial artery	1.42×10-6 (p-value)	GTEx Portal V6	97
RP11-54A4.2	?	ENSG00000237781	esophagus mucosa	1.06×10-9 (p-value)	GTEx Portal V6	134
ADAMTSL4	?	ENSG00000143382	esophagus mucosa	3.79×10-11 (p-value)	GTEx Portal V6	134
ECM1	?	ENSG00000143369	esophagus mucosa	8.32×10-6 (p-value)	GTEx Portal V6	10
?	?	ILMN_1702231	blood	1.24×10-6 (p-value)	Westra et al.	33
RPRD2	?	ENSG00000163125	testis	2.07×10-8 (p-value)	GTEx Portal V6	134
TARS2	?	ENSG00000143374	skeletal muscle	2.84×10-8 (p-value)	GTEx Portal V6	132
TARS2	?	ENSG00000143374	unexposed skin	4.82×10-8 (p-value)	GTEx Portal V6	129
ADAMTSL4	?	ENSG00000143382	aorta	1.07×10-7 (p-value)	GTEx Portal V6	132
RP11-54A4.2	?	ENSG00000237781	aorta	1.43×10-5 (p-value)	GTEx Portal V6	3
TARS2	?	ENSG00000143374	subcutaneous adipocytes	2.47×10-7 (p-value)	GTEx Portal V6	129
RPRD2	ENST00000401000	ILMN_3238889	monocyte	9.96×10-6 (p-value)	Fairfax et al.	3
			b-cell	6.13×10-6 (p-value)	Fairfax et al.	3
MRPS21	ENST00000581066	ILMN_1660292	monocyte	2.64×10-14 (p-value)	Fairfax et al.	2
			b-cell	1.15×10-4 (p-value)	Fairfax et al.	2
RPRD2	?	ENSG00000163125	lung	1.48×10-5 (p-value)	GTEx Portal V6	42
ENSA	?	ENSG00000143420	liver	5.48×10-6 (p-value)	GTEx Portal V6	1
ADAMTSL4	?	ENSG00000143382	transformed fibroblasts	8.76×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000036149	1	ENCP00000004292	VPS45
		ENCP00000004293	VPS45
ENCE00000036228	2	ENCP00000004307	APH1A

ENCE00000036339	1	ENCP00000004360	ANXA9
		ENCP00000004327	MCL1
ENCE00000036433	1	ENCP00000004346	RP11-235D19.2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001524644 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3, PolII
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		blood (K562)	PolII
		muscle (HSMM)	H3K27ac, DNase1
ENSR00001586440 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K4me2, H3K4me3
		HSMMtube	H3K79me2, H3K4me2
		blood (K562)	H3K27ac, H3K9ac, H3K79me2, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3, H3K9ac, H3K4me2
		muscle (HSMM)	H3K79me2, H3K4me3, H3K4me2
		liver (HepG2)	H3K79me2, H3K4me2, H3K9ac, H3K4me3
		blood (GM12878)	H3K79me2, H3K4me3, H3K27ac, H3K9ac, H3K36me3
		lung (IMR90)	H3K79me2, H3K4me2, H3K36me3, H3K4me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K4me3, H3K27ac
		Osteobl	H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K4me1, H3K4me2, H3K27ac, H3K9ac
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K79me2, H3K4me3, H3K4me2, PolII, H3K9ac, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3
		A549	H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
ENSR00000544743 (promoter)	2	embryonic stem cell (H1ESC)	TAF1, H3K4me2, H3K9ac, H3K4me3, H4K5ac, H3K36me3, CTCF, PolII, Jund, TAF7, H3K27ac, Yy1, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, DNase1
		blood (K562)	CTCFL, PolII, H3K4me2, DNase1, H3K4me3, TAF1, H2AZ, Yy1, H3K27ac, Max, PU1, H3K9ac, Cmyc, H3K79me2, HEY1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		liver (HepG2)	Yy1, PolII, H3K79me2, TAF1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4ac, H3K9ac, H4K5ac, H3K4me2, DNase1, H4K8ac, H3K18ac, H3K27ac
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, H3K79me2, TAF1, PU1, PolII, H2AZ, DNase1, Yy1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Nrf1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001586444 (enhancer)	1	cervix (HeLa-S3)	H3K79me2
		monocytes (Monocytes-CD14+)	H4K20me1
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K79me2
ENSR00001524646 (enhancer)	1	liver (HepG2)	H3K79me2, H3K4me1, FOXA1
		blood (GM12878)	H3K79me2
		blood (DND-41)	H3K36me3
ENSR00001586446 (enhancer)	1	liver (HepG2)	H3K79me2, H3K4me1
		blood (DND-41)	H3K36me3
ENSR00001586448	1	liver (HepG2)	H3K79me2, H3K4me1, H3K4me2, H3K27ac

(enhancer)		blood (GM12878)	H3K79me2
		blood (DND-41)	H3K36me3
ENSR00000544752 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00000544755 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me1, FOXA1
		lung (IMR90)	DNase1, H3K36me3
		blood (K562)	Egr1
		A549	H3K36me3
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, DNase1
		breast (HMEC)	DNase1, H3K27ac, H3K4me2
ENSR00000285713 (promoter flanking region)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		Osteobl	CTCF
		blood (K562)	DNase1, Rad21, H2AZ, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF, H3K4me2, Rad21, H2AZ
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF, DNase1
ENSR00000285718 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, Rad21, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me2, H2AZ, H3K36me3, DNase1
		blood (K562)	H3K27ac, H3K9ac, H2AZ, PolII, H3K4me2, CTCF, H3K4me1, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K36me3, H3K4me1, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K36me3, DNase1
		liver (HepG2)	PolII, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF, H3K4me3, DNase1
		lung (IMR90)	H3K4me3, H3K9ac, H3K36me3, H3K4me2, DNase1
		blood (GM12878)	Pbx3, PolII, H2AZ, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2
		skin (NHEK)	H3K9ac, H3K4me2, H3K36me3, DNase1
		NHLF	H3K36me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K4me1, H3K4me2, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	CTCF, DNase1, Nrf1, H3K9ac, Jund, H3K4me1, PolII
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27me3, DNase1
		endothelium (HUVEC)	H3K4me3, PolII, CTCF, DNase1, Max
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00001524662 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	DNase1, H3K27ac, H3K9ac
		blood (K562)	CTCF
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1
		cervix (HeLa-S3)	CTCF, DNase1
		liver (HepG2)	CTCF, H3K4me1
		lung (IMR90)	DNase1, H3K4me1, H3K18ac, H3K27ac, H4K5ac, H4K91ac
		nervous (NH-A)	DNase1
		skin (NHEK)	CTCF, DNase1
ENSR00001586454 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K9ac, H3K27ac, DNase1
		blood (K562)	CTCF
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1
		cervix (HeLa-S3)	CTCF, DNase1
		liver (HepG2)	CTCF
		lung (IMR90)	DNase1, H3K4me1, H3K18ac, H3K27ac, H4K5ac, H4K91ac

	nervous (NH-A)	DNase1
	skin (NHEK)	CTCF, DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PRPF3	2	ENST00000324862 ENST00000496202	hsa-miR-329-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC242988.1	downstream gene variant, upstream gene variant	893	ENST00000624245	?	ENSP00000485440	4
ECM1	upstream gene variant, downstream gene variant	657	ENST00000498579	?	?	6
ECM1	upstream gene variant, downstream gene variant	727	ENST00000490346	?	?	4
ECM1	upstream gene variant, downstream gene variant	682	ENST00000346569	NM_022664.2	ENSP00000271630	6
ECM1	upstream gene variant, downstream gene variant	651	ENST00000369049	NM_001202858.1	ENSP00000358045	6
ECM1	upstream gene variant, downstream gene variant	637	ENST00000470432	?	?	6
ECM1	upstream gene variant, downstream gene variant	660	ENST00000369047	NM_004425.3	ENSP00000358043	6
ECM1	upstream gene variant, downstream gene variant	327	ENST00000496744	?	?	7
FALEC	upstream gene variant, downstream gene variant	57	ENST00000416894	?	?	7
MRPS21	downstream gene variant	86	ENST00000581066	NM_018997.3	ENSP00000461930	7
MRPS21	downstream gene variant	86	ENST00000614145	NM_031901.5	ENSP00000480129	7
PRPF3	downstream gene variant	266	ENST00000476970	?	?	4
PRPF3	upstream gene variant, downstream gene variant	1435	ENST00000467329	?	?	5
PRPF3	upstream gene variant, downstream gene variant	771	ENST00000324862	NM_004698.2	ENSP00000315379	9
PRPF3	upstream gene variant, downstream gene variant	774	ENST00000496202	?	?	8
PRPF3	downstream gene variant	2184	ENST00000467514	?	?	3
PRPF3	upstream gene variant, downstream gene variant	3359	ENST00000493553	?	?	6
PRPF3	upstream gene variant, downstream gene variant	313	ENST00000470824	?	?	7
RPRD2	upstream gene variant, downstream gene variant	953	ENST00000401000	NM_001297674.1	ENSP00000383785	7
RPRD2	upstream gene variant, downstream gene variant	1366	ENST00000492220	?	?	3
RPRD2	upstream gene variant	1011	ENST00000369067	?	ENSP00000358063	3
RPRD2	upstream gene variant, downstream gene variant	1014	ENST00000369068	NM_015203.3	ENSP00000358064	4
TARS2	upstream gene variant, downstream gene variant	1964	ENST00000438568	?	ENSP00000415002	3
TARS2	upstream gene variant, downstream gene variant	2031	ENST00000369053	?	?	2
TARS2	upstream gene variant, downstream gene variant	293	ENST00000483046	?	?	6
TARS2	upstream gene variant, downstream gene variant	167	ENST00000369054	NM_001271896.1	ENSP00000358050	3
TARS2	upstream gene variant, downstream gene variant	149	ENST00000480070	?	?	3
TARS2	upstream gene variant, downstream gene variant	149	ENST00000463555	?	?	3
TARS2	upstream gene variant, downstream gene variant	2042	ENST00000479372	?	?	2
TARS2	upstream gene variant, downstream gene variant	259	ENST00000462578	?	?	6
TARS2	upstream gene variant, downstream gene variant	1759	ENST00000466989	?	?	7
TARS2	upstream gene variant, downstream gene variant	249	ENST00000460794	?	?	6
TARS2	upstream gene variant, downstream gene variant	242	ENST00000467982	?	ENSP00000475551	3
TARS2	upstream gene variant, downstream gene variant	141	ENST00000606933	NM_001271895.1	ENSP00000475847	3
TARS2	upstream gene variant, downstream gene variant	2035	ENST00000369051	?	ENSP00000358047	3

TARS2	upstream gene variant, downstream gene variant	2017	ENST00000369064	NM_025150.4	ENSP00000358060	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MRPS21	ENST00000614145	NM_031901.5	ENSP00000480129	7
MRPS21	ENST00000581066	NM_018997.3	ENSP00000461930	7
PRPF3	ENST00000470824	?	?	5
PRPF3	ENST00000467514	?	?	2
PRPF3	ENST00000476970	?	?	4
PRPF3	ENST00000496202	?	?	4
PRPF3	ENST00000467329	?	?	13
PRPF3	ENST00000324862	NM_004698.2	ENSP00000315379	15
RPRD2	ENST00000369067	?	ENSP00000358063	47
RPRD2	ENST00000401000	NM_001297674.1	ENSP00000383785	61
RPRD2	ENST00000492220	?	?	62
RPRD2	ENST00000369068	NM_015203.3	ENSP00000358064	61
TARS2	ENST00000467982	?	ENSP00000475551	15
TARS2	ENST00000369053	?	?	1
TARS2	ENST00000480070	?	?	13
TARS2	ENST00000460794	?	?	6
TARS2	ENST00000606933	NM_001271895.1	ENSP00000475847	15
TARS2	ENST00000369051	?	ENSP00000358047	15
TARS2	ENST00000463555	?	?	13
TARS2	ENST00000369064	NM_025150.4	ENSP00000358060	15
TARS2	ENST00000462578	?	?	6
TARS2	ENST00000466989	?	?	1
TARS2	ENST00000483046	?	?	1
TARS2	ENST00000438568	?	ENSP00000415002	15
TARS2	ENST00000369054	NM_001271896.1	ENSP00000358050	15

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MRPS21	ENST00000614145	NM_031901.5	ENSP00000480129	2
MRPS21	ENST00000581066	NM_018997.3	ENSP00000461930	2
RPRD2	ENST00000401000	NM_001297674.1	ENSP00000383785	2
RPRD2	ENST00000369068	NM_015203.3	ENSP00000358064	1
TARS2	ENST00000369051	?	ENSP00000358047	1
TARS2	ENST00000369064	NM_025150.4	ENSP00000358060	1
TARS2	ENST00000438568	?	ENSP00000415002	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ECM1	ENST00000498579	?	1
ECM1	ENST00000470432	?	2
PRPF3	ENST00000470824	?	1
RPRD2	ENST00000492220	?	1
TARS2	ENST00000479372	?	1

Locus 25 (rs16850073)

SNiPAcard

Block annotations

Block info

genomic range	chr4:74,701,512-74,703,999
block size	2,488 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.118 [0.075 – 0.161]	gene(s) hit or close-by	CXCL6
phastCons	μ = 0.519 [0.04 – 0.999]	eQTL gene(s)	CXCL6, PF4V1
GERP++	μ = 1.190 [0 – 2.38]	potentially regulated gene(s)	–
CADD score	μ = 7.585 [1.391 – 13.78]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CXCL6	?	ENSG00000124875	tibial nerve	2.03×10^{-7} (p-value)	GTEx Portal V6	2
CXCL6	?	ENSG00000124875	pancreas	5.79×10^{-7} (p-value)	GTEx Portal V6	1
PF4V1	ENST00000226524	ILMN_1745522	monocyte	1.91×10^{-17} (p-value)	Zeller et al.	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CXCL6	upstream gene variant	1392	ENST00000503446	?	?	1
CXCL6	upstream gene variant, downstream gene variant	845	ENST00000515050	?	ENSP00000424819	2
CXCL6	upstream gene variant	702	ENST00000226317	NM_002993.3	ENSP00000226317	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL6	ENST00000503446	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL6	ENST00000226317	NM_002993.3	ENSP00000226317	1

Locus 26 (rs2304456)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,422,367-186,450,863
block size	28,497 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.131 [-1.563 – 0.542]	gene(s) hit or close-by	KNG1, RP11-573D15.8, RP11-795A2.3
phastCons	μ = 0.003 [0 – 0.006]	eQTL gene(s)	–
GERP++	μ = -2.976 [-9.97 – 0.235]	potentially regulated gene(s)	–
CADD score	μ = 2.767 [1.095 – 4.152]	disease gene(s)	KNG1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
KNG1	HIGH MOLECULAR WEIGHT KININOGEN DEFICIENCY	OMIM	MIM:228960
KNG1	Congenital high-molecular-weight kininogen deficiency	OrphaNet	OrphaNet:483

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
KNG1	missense variant	ENST00000265023	NM_001102416.2	ENSP00000265023	M/I	atG/atT	?	?	1
KNG1	missense variant	ENST00000287611	NM_000893.3	ENSP00000287611	M/I	atG/atT	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	downstream gene variant	3166	ENST00000628020	?	?	2
RP11-573D15.8	upstream gene variant	1472	ENST00000625877	?	?	1
RP11-573D15.8	downstream gene variant	1415	ENST00000628858	?	?	1
RP11-573D15.8	downstream gene variant	3433	ENST00000629734	?	?	1
RP11-573D15.8	downstream gene variant	1304	ENST00000627015	?	?	1
RP11-573D15.8	downstream gene variant	1556	ENST00000626306	?	?	2
RP11-573D15.8	downstream gene variant	1092	ENST00000625710	?	?	1
RP11-573D15.8	downstream gene variant	2775	ENST00000629126	?	?	2
RP11-573D15.8	downstream gene variant	1088	ENST00000627268	?	?	1
RP11-573D15.8	downstream gene variant	3342	ENST00000628831	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KNG1	ENST00000287611	NM_000893.3	ENSP00000287611	2
KNG1	ENST00000265023	NM_001102416.2	ENSP00000265023	2
KNG1	ENST00000447445	NM_001166451.1	ENSP00000396025	3
RP11-573D15.8	ENST00000626151	?	?	3
RP11-573D15.8	ENST00000628190	?	?	4
RP11-573D15.8	ENST00000628253	?	?	4
RP11-573D15.8	ENST00000627919	?	?	4
RP11-573D15.8	ENST00000625386	?	?	4
RP11-573D15.8	ENST00000626649	?	?	1
RP11-573D15.8	ENST00000629451	?	?	4
RP11-573D15.8	ENST00000626633	?	?	4
RP11-573D15.8	ENST00000628601	?	?	3
RP11-573D15.8	ENST00000630178	?	?	4
RP11-573D15.8	ENST00000627469	?	?	4
RP11-573D15.8	ENST00000627268	?	?	2
RP11-573D15.8	ENST00000627830	?	?	4
RP11-573D15.8	ENST00000630331	?	?	4
RP11-573D15.8	ENST00000630864	?	?	4
RP11-573D15.8	ENST00000625839	?	?	4
RP11-573D15.8	ENST00000628505	?	?	4
RP11-573D15.8	ENST00000628728	?	?	4
RP11-573D15.8	ENST00000629106	?	?	4
RP11-573D15.8	ENST00000354642	?	?	3
RP11-573D15.8	ENST00000625741	?	?	4
RP11-573D15.8	ENST00000599314	?	?	3
RP11-573D15.8	ENST00000628858	?	?	2
RP11-573D15.8	ENST00000627551	?	?	3
RP11-573D15.8	ENST00000630726	?	?	1
RP11-573D15.8	ENST00000625303	?	?	4
RP11-573D15.8	ENST00000627015	?	?	2
RP11-573D15.8	ENST00000626845	?	?	4
RP11-573D15.8	ENST00000625710	?	?	2
RP11-795A2.3	ENST00000413467	?	?	1

Locus 27 (rs2097326)

SNiPAcard

Block annotations

Block info

genomic range	chr22:33,159,092-33,167,869
block size	8,778 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.235 [-3.023 – 2.015]	gene(s) hit or close-by	SYN3
phastCons	μ = 0.069 [0 – 0.983]	eQTL gene(s)	–
GERP++	μ = 0.074 [-6.84 – 2.32]	potentially regulated gene(s)	–
CADD score	μ = 4.567 [0.694 – 16.06]	disease gene(s)	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000293419 (promoter flanking region)	3	NHLF	DNase1
		HSMMtube	DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		lung (IMR90)	DNase1, H3K18ac
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SYN3	ENST00000462268	?	?	19
SYN3	ENST00000358763	NM_003490.3, NM_001135774.1, NM_133633.2	ENSP00000351614	19

Locus 28 (rs651007)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,141,870-136,155,000
block size	13,131 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.489 [-4.02 – 0.587]	**gene(s) hit or close-by**	ABO
phastCons	μ = 0.073 [0.002 – 0.373]	**eQTL gene(s)**	ABO, GBGT1, SURF1, SURF6
GERP++	μ = -0.111 [-1.2 – 0.495]	**potentially regulated gene(s)**	ADAMTSL2, CELP, DBH, RALGDS, SARDH
CADD score	μ = 4.265 [0.015 – 8.406]	**disease gene(s)**	SURF1, DBH, ADAMTSL2, SARDH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Iron status biomarkers (ferritin levels)	$<1.00\times10^{-8}$	GWAS Catalog	25352340	1
Blood metabolite levels	$<6.00\times10^{-20}$	GWAS Catalog	24816252	1
Blood metabolite ratios	$<6.00\times10^{-34}$	GWAS Catalog	24816252	2
Urinary metabolites (H-NMR features)	$<1.00\times10^{-28}$	GWAS Catalog	24586186	1
Coronary artery disease or large artery stroke	$<3.00\times10^{-8}$	GWAS Catalog	24262325	1
Coronary Artery Disease	$<2.00\times10^{-7}$	GWAS Catalog	24262325	1
Coronary artery disease or ischemic stroke	$<2.00\times10^{-9}$	GWAS Catalog	24262325	1
Serum alkaline phosphatase levels	$<1.00\times10^{-56}$	GWAS Catalog	24094242	1
End-stage coagulation	$<2.00\times10^{-25}$	GWAS Catalog	23381943	1
Red blood cell traits	$<9.00\times10^{-18}$	GWAS Catalog	23222517	1
Venous thromboembolism	$<3.00\times10^{-16}$	GWAS Catalog	22672568	1
Liver enzyme levels (alkaline phosphatase)	$<3.00\times10^{-123}$	GWAS Catalog	22001757	1
Metabolite levels	$<6.00\times10^{-9}$	GWAS Catalog	21909109	1
Coronary heart disease	$<4.00\times10^{-14}$	GWAS Catalog	21378990	1
Cholesterol, total	$<9.00\times10^{-21}$	GWAS Catalog	20686565	1
LDL cholesterol	$<8.00\times10^{-22}$	GWAS Catalog	20686565	1
Soluble levels of adhesion molecules	$<2.00\times10^{-41}$	GWAS Catalog	20167578	1
E-selectin levels	$<2.00\times10^{-82}$	GWAS Catalog	20147318	1
Red blood cell count	$<3.00\times10^{-12}$	GWAS Catalog	20139978	1
Hematological and biochemical traits	$<1.00\times10^{-11}$	GWAS Catalog	20139978	1
Angiotensin-converting enzyme activity	$<3.00\times10^{-8}$	GWAS Catalog	20066004	1
Soluble E-selectin levels	$<1.00\times10^{-29}$	GWAS Catalog	19729612	1
ADpSGEGDFXAEGGGVR*	6.34×10^{-20}	Metabolomics GWAS Server	24816252	3
E-selectin	1.33×10^{-29}	dbGaP	pha002871	2

Disease gene annotation

gene	trait	source DB	source entry/link
SURF1	LEIGH SYNDROME	OMIM	MIM:256000
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY, CONGENITAL	OMIM	MIM:223360
ADAMTSL2	GELEOPHYSIC DYSPLASIA 1	OMIM	MIM:231050
SARDH	SARCOSINEMIA	OMIM	MIM:268900
SURF1	Complex IV deficiency	DECIPHER	MIM:220110
SURF1	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
SURF1	Fatal infantile cytochrome C oxidase deficiency	OrphaNet	OrphaNet:1561
SURF1	Leigh syndrome with cardiomyopathy	OrphaNet	OrphaNet:70474
SURF1	Leigh syndrome with leukodystrophy	OrphaNet	OrphaNet:255241
SURF1	SURF1-related Charcot-Marie-Tooth disease type 4	OrphaNet	OrphaNet:391351
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY	OrphaNet	OrphaNet:230
ADAMTSL2	Geleophysic dysplasia	OrphaNet	OrphaNet:2623
SARDH	SARCOSINEMIA	OrphaNet	OrphaNet:3129

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABO	?	ENSG00000175164	adrenal gland	2.57×10^{-6} (p-value)	GTEx Portal V6	3
SURF1	?	ENSG00000148290	atrial appendage	4.13×10^{-6} (p-value)	GTEx Portal V6	1
ABO	?	ENSG00000175164	blood	6.42×10^{-10} (p-value)	GTEx Portal V6	7
GBGT1	ENST00000372040	ILMN_1652906	blood	1.60×10^{-6} (p-value)	Westra et al.	3
GBGT1	ENST00000470431					
GBGT1	ENST00000472281					
GBGT1	ENST00000372043					
GBGT1	ENST00000540636					
SURF6	ENST00000372022	ILMN_1778032	blood	8.04×10^{-7} (p-value)	Westra et al.	3
SURF6	ENST00000468290					
SURF6	ENST00000372022	ILMN_1778032	monocyte	4.67×10^{-5} (p-value)	Fairfax et al.	1
SURF6	ENST00000468290		b-cell	1.52×10^{-4} (p-value)	Fairfax et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000558528	1	ENCP00000060534	SARDH
		ENCP00000060486	CELP
		ENCP00000060489	RALGDS
		ENCP00000060527	ADAMTSL2
		ENCP00000060525	ADAMTSL2
		ENCP00000060535	SARDH
		ENCP00000060529	DBH

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000251886 (promoter)	2	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	CTCF, Rad21, Gabp, H3K4me2, H3K4me3, H3K27me3, DNase1
		HSMMtube	H3K27me3

		Osteobl	H3K27me3
		blood (K562)	ZBTB7A, H3K27ac, H3K9ac, Cmyc, H3K79me2, H3K4me3, DNase1, CTCF, H3K4me2, PolII, CTCFL, H2AZ
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001734091 e! (TF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	PolII, H3K9ac, DNase1
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001734092 e! (enhancer)	1	HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	DNase1
		skin (NHDF-AD)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABO e!	upstream gene variant	1140	ENST00000538324 e!	?	ENSP00000483018 e!	5
ABO e!	upstream gene variant	1140	ENST00000611156 e!	NM_020469.2	ENSP00000483265 e!	5
ABO e!	upstream gene variant	1153	ENST00000453660 e!	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO e!	ENST00000538324 e!	?	ENSP00000483018 e!	3
ABO e!	ENST00000611156 e!	NM_020469.2	ENSP00000483265 e!	3
ABO e!	ENST00000453660 e!	?	?	3

Locus 29 (rs11603020)

SNiPAcard

Block annotations

Block info

genomic range	chr11:57,369,008-57,400,462
block size	31,455 bp
variant count	32 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.465 [-3.014 – 1.413]	gene(s) hit or close-by	SERPING1
phastCons	μ = 0.024 [0 – 0.267]	eQTL gene(s)	AP000662.4, MED19, RN7SL605P, TIMM10, YPEL4, ZDHHC5
GERP++	μ = -0.478 [-6.6 – 3.68]	potentially regulated gene(s)	CLP1, PRG2, RP11-872D17.8, RTN4RL2, SERPING1, SERPING1, SLC43A1, SLC43A3, SSRP1, YPEL4, ZDHHC5
CADD score	μ = 3.907 [0.037 – 22.6]	disease gene(s)	SERPING1, CLP1

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	not provided	ClinVar	RCV000077812.1	1

Disease gene annotation

gene	trait	source DB	source entry/link
SERPING1	ANGIOEDEMA, HEREDITARY, TYPE I	OMIM	MIM:106100
CLP1	PONTOCEREBELLAR HYPOPLASIA, TYPE 10	OMIM	MIM:615803
SERPING1	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147
SERPING1	Hereditary angioedema type 1	OrphaNet	OrphaNet:100050
SERPING1	Hereditary angioedema type 2	OrphaNet	OrphaNet:100051
CLP1	Pontocerebellar hypoplasia, type 10	DECIPHER	MIM:615803

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SERPING1	missense variant	ENST00000340687	?	ENSP00000341861	M/V	Atg/Gtg	?	?	1
SERPING1	missense variant	ENST00000278407	NM_001032295.1, NM_000062.2	ENSP00000278407	M/V	Atg/Gtg	?	?	1
SERPING1	missense variant	ENST00000403558	?	ENSP00000384420	M/V	Atg/Gtg	?	?	1
SERPING1	missense variant	ENST00000378324	?	ENSP00000367575	M/V	Atg/Gtg	?	?	1
SERPING1	missense variant	ENST00000378323	?	ENSP00000367574	M/V	Atg/Gtg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TIMM10	?	ENSG00000134809	muscularis mucosae	2.46×10^{-10} (p-value)	GTEx Portal V6	32

TIMM10	?	ENSG00000134809	lung	7.44×10-9 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	tibial nerve	1.51×10-9 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	thyroid	1.81×10-7 (p-value)	GTEx Portal V6	32
AP000662.4	?	ENSG00000254602	thyroid	2.01×10-10 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	esophagus mucosa	1.45×10-11 (p-value)	GTEx Portal V6	32
MED19	?	ENSG00000156603	esophagus mucosa	3.39×10-6 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	skeletal muscle	7.74×10-6 (p-value)	GTEx Portal V6	8
ZDHHC5	?	ENSG00000156599	skeletal muscle	8.30×10-6 (p-value)	GTEx Portal V6	12
TIMM10	?	ENSG00000134809	sun exposed skin	1.11×10-7 (p-value)	GTEx Portal V6	32
MED19	?	ENSG00000156603	sun exposed skin	2.16×10-9 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	subcutaneous adipocytes	7.61×10-7 (p-value)	GTEx Portal V6	24
MED19	?	ENSG00000156603	subcutaneous adipocytes	7.87×10-7 (p-value)	GTEx Portal V6	32
RN7SL605P	ENST00000577229	ILMN_1765332	monocyte	6.00×10-5 (p-value)	Fairfax et al.	5
TIMM10	ENST00000525587		b-cell	1.31×10-4 (p-value)	Fairfax et al.	5
TIMM10	ENST00000257245					
TIMM10	ENST00000525587	ILMN_1765332	skin	3.74×10-7 (p-value)	MuTHER consortium	7
RN7SL605P	ENST00000577229		blood	9.92×10-7 (p-value)	MuTHER consortium	7
TIMM10	ENST00000257245		adipocyte	6.66×10-10 (p-value)	MuTHER consortium	7
TIMM10	ENST00000525587	ILMN_1765332	monocyte	1.71×10-25 (p-value)	Zeller et al.	3
RN7SL605P	ENST00000577229					
TIMM10	ENST00000257245					
YPEL4	ENST00000532314	ILMN_1726624	monocyte	4.38×10-21 (p-value)	Zeller et al.	3
YPEL4	ENST00000528797					
YPEL4	ENST00000300022					
YPEL4	ENST00000524669					
YPEL4	ENST00000524592					
TIMM10	?	ENSG00000134809	stomach	8.05×10-6 (p-value)	GTEx Portal V6	8
TIMM10	?	ENSG00000134809	pancreas	1.55×10-5 (p-value)	GTEx Portal V6	2

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000066147	1	ENST00000403558, ENST00000278407, ENST00000378324, ENST00000340687, ENST00000378323	SERPING1
FFCP00000066188	1	ENST00000403558, ENST00000530113, ENST00000278407, ENST00000378324, ENST00000340687, ENST00000378323	SERPING1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000106368	1	ENCP00000012246	SLC43A3
		ENCP00000012254	SLC43A1
		ENCP00000012242	SLC43A3

		ENCP00000012278	ZDHHC5
		ENCP00000012241	RP11-872D17.8
		ENCP00000012240	PRG2
		ENCP00000012247	SLC43A3
		ENCP00000012244	RP11-872D17.8
		ENCP00000012256	SLC43A1
		ENCP00000012276	CLP1
		ENCP00000012275	YPEL4
		ENCP00000012249	RTN4RL2
ENCE00000106266	2	ENCP00000012246	SLC43A3
		ENCP00000012268	SERPING1 SERPING1
		ENCP00000012254	SLC43A1
		ENCP00000012242	SLC43A3
		ENCP00000012255	SLC43A1
		ENCP00000012278	ZDHHC5
		ENCP00000012235	SSRP1
		ENCP00000012241	RP11-872D17.8
		ENCP00000012240	PRG2
		ENCP00000012247	SLC43A3
		ENCP00000012244	RP11-872D17.8
		ENCP00000012256	SLC43A1
		ENCP00000012276	CLP1
		ENCP00000012275	YPEL4
		ENCP00000012249	RTN4RL2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001570401 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		lung (IMR90)	H3K36me3
		blood (K562)	H3K27me3
ENSR00001603770 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K36me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SERPING1	downstream gene variant	1090	ENST00000403558	?	ENSP00000384420	4
SERPING1	upstream gene variant, downstream gene variant	505	ENST00000531605	?	?	8
SERPING1	downstream gene variant	1323	ENST00000531133	?	ENSP00000435431	4
SERPING1	downstream gene variant	1227	ENST00000405496	?	ENSP00000384561	10
SERPING1	downstream gene variant	1057	ENST00000378324	?	ENSP00000367575	4
SERPING1	upstream gene variant, downstream gene variant	1071	ENST00000528996	?	ENSP00000431226	14
SERPING1	downstream gene variant	1443	ENST00000457869	?	ENSP00000399746	10
SERPING1	upstream gene variant, downstream gene variant	736	ENST00000530113	?	?	10
SERPING1	downstream gene variant	1323	ENST00000619430	?	ENSP00000478572	4
SERPING1	downstream gene variant	1051	ENST00000278407	NM_001032295.1, NM_000062.2	ENSP00000278407	4
SERPING1	downstream gene variant	1058	ENST00000340687	?	ENSP00000341861	4

SERPING1	downstream gene variant	1114	ENST00000531797	?		ENSP00000432554	4
SERPING1	downstream gene variant	1165	ENST00000378323	?		ENSP00000367574	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPING1	ENST00000531133	?	ENSP00000435431	17
SERPING1	ENST00000531797	?	ENSP00000432554	17
SERPING1	ENST00000278407	NM_001032295.1, NM_000062.2	ENSP00000278407	17
SERPING1	ENST00000403558	?	ENSP00000384420	17
SERPING1	ENST00000528996	?	ENSP00000431226	7
SERPING1	ENST00000619430	?	ENSP00000478572	17
SERPING1	ENST00000378323	?	ENSP00000367574	17
SERPING1	ENST00000378324	?	ENSP00000367575	17
SERPING1	ENST00000531605	?	?	8
SERPING1	ENST00000340687	?	ENSP00000341861	17
SERPING1	ENST00000530113	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPING1	ENST00000531133	?	ENSP00000435431	1
SERPING1	ENST00000531797	?	ENSP00000432554	1
SERPING1	ENST00000528996	?	ENSP00000431226	1
SERPING1	ENST00000619430	?	ENSP00000478572	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SERPING1	ENST00000530113	?	1

Locus 30 (rs33995560)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,303,312-34,306,470
block size	3,159 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.168 [-0.463 – 0.853]	**gene(s) hit or close-by**	CCL14, CCL15-CCL14, CCL16, CTB-186H2.3
phastCons	μ = 0.141 [0 – 0.78]	**eQTL gene(s)**	–
GERP++	μ = 0.991 [-0.722 – 3.54]	**potentially regulated gene(s)**	–
CADD score	μ = 4.572 [1.506 – 8.195]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001344901 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		Osteobl	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	DNase1, H3K4me1, CTCF
		blood (GM12878)	CTCF, Rad21
		A549	CTCF
		skin (NHEK)	CTCF
ENSR00001638275 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3
		Osteobl	CTCF
		blood (K562)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		liver (HepG2)	H3K4me1
		blood (GM12878)	CTCF, Rad21
		A549	CTCF, H3K27me3
ENSR00001638276 (enhancer)	3	cervix (HeLa-S3)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	H3K4me1
		blood (K562)	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL14	downstream gene variant	3857	ENST00000614009	?	ENSP00000483186	1
CCL14	downstream gene variant	3860	ENST00000618404	NM_032963.3	ENSP00000481023	1
CCL14	downstream gene variant	4253	ENST00000620991	?	ENSP00000484818	1
CCL14	downstream gene variant	4225	ENST00000622526	NM_032962.4	ENSP00000479097	1
CCL15-CCL14	downstream gene variant	4222	ENST00000610751	?	ENSP00000481940	1
CCL15-CCL14	downstream gene variant	4222	ENST00000616694	?	ENSP00000481402	1

CCL16	downstream gene variant, upstream gene variant	217	ENST00000613642	?	ENSP00000478592	2
CCL16	downstream gene variant	223	ENST00000611905	NM_004590.2	ENSP00000478024	1
CCL16	downstream gene variant	292	ENST00000610493	?	ENSP00000478934	1
CTB-186H2.3	upstream gene variant	4222	ENST00000617687	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL16	ENST00000611905	NM_004590.2	ENSP00000478024	6
CCL16	ENST00000610493	?	ENSP00000478934	6
CCL16	ENST00000613642	?	ENSP00000478592	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL16	ENST00000611905	NM_004590.2	ENSP00000478024	2
CCL16	ENST00000610493	?	ENSP00000478934	2

Locus 31 (rs2228243)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,394,038-186,395,113
block size	1,076 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.758 [-1.01 – -0.507]	gene(s) hit or close-by	HRG, RP11-573D15.8
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	–
GERP++	μ = -1.033 [-1.9 – -0.166]	potentially regulated gene(s)	–
CADD score	μ = 0.532 [0.006 – 1.059]	disease gene(s)	HRG

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HRG	THROMBOPHILIA DUE TO HISTIDINE-RICH GLYCOPROTEIN DEFICIENCY	OMIM	MIM:613116
HRG	Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency	OrphaNet	OrphaNet:217467

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
HRG	missense variant	ENST00000232003	NM_000412.3	ENSP00000232003	R/H	cGt/cAt	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	upstream gene variant	1731	ENST00000626006	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HRG	ENST00000232003	NM_000412.3	ENSP00000232003	1
HRG	ENST00000495413	?	?	2
RP11-573D15.8	ENST00000630331	?	?	2
RP11-573D15.8	ENST00000625741	?	?	2
RP11-573D15.8	ENST00000628253	?	?	2
RP11-573D15.8	ENST00000626649	?	?	2
RP11-573D15.8	ENST00000630864	?	?	2

RP11-573D15.8	ENST00000625386	?	?	2
RP11-573D15.8	ENST00000626633	?	?	2
RP11-573D15.8	ENST00000628190	?	?	2
RP11-573D15.8	ENST00000629451	?	?	2
RP11-573D15.8	ENST00000630315	?	?	2
RP11-573D15.8	ENST00000628505	?	?	2
RP11-573D15.8	ENST00000630178	?	?	2
RP11-573D15.8	ENST00000630726	?	?	2
RP11-573D15.8	ENST00000625303	?	?	2
RP11-573D15.8	ENST00000627469	?	?	2
RP11-573D15.8	ENST00000627830	?	?	2
RP11-573D15.8	ENST00000626845	?	?	2
RP11-573D15.8	ENST00000625877	?	?	2
RP11-573D15.8	ENST00000625839	?	?	2
RP11-573D15.8	ENST00000629106	?	?	2
RP11-573D15.8	ENST00000628728	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
HRG	ENST00000495413	?	1

Locus 32 (rs3827278)

SNiPAcard

Block annotations

Block info

genomic range	chr22:17,595,915-17,596,322
block size	408 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.080 [-0.258 – 0.328]	gene(s) hit or close-by	CECR6, IL17RA
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	AC006946.15, IL17RA
GERP++	μ = -0.095 [-2.8 – 1.55]	potentially regulated gene(s)	–
CADD score	μ = 2.715 [1.84 – 3.715]	disease gene(s)	IL17RA

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
IL17RA	CANDIDIASIS, FAMILIAL, 5	OMIM	MIM:613953
IL17RA	Chronic mucocutaneous candidosis	OrphaNet	OrphaNet:1334

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL17RA	?	ENSG00000177663	pancreas	6.93×10^{-9} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	muscularis mucosae	3.89×10^{-7} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	lung	1.96×10^{-7} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	atrial appendage	1.59×10^{-6} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	transformed fibroblasts	3.23×10^{-15} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	tibial artery	5.99×10^{-9} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	thyroid	3.81×10^{-11} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	skeletal muscle	7.33×10^{-10} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	unexposed skin	5.37×10^{-7} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	sun exposed skin	1.86×10^{-13} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	aorta	1.11×10^{-10} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	left ventricle	1.59×10^{-8} (p-value)	GTEx Portal V6	3
AC006946.15	ENST00000441544	ILMN_1888355	monocyte	8.94×10^{-23} (p-value)	Zeller et al.	1
IL17RA	?	ENSG00000177663	subcutaneous adipocytes	1.47×10^{-15} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	stomach	1.24×10^{-6} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	tibial nerve	4.92×10^{-9} (p-value)	GTEx Portal V6	3

IL17RA	?	ENSG00000177663	esophagus mucosa	1.03×10^{-8} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	gastroesophageal junction	3.12×10^{-6} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CECR6	downstream gene variant	868	ENST00000331437	NM_031890.3	ENSP00000329318	3
CECR6	downstream gene variant	867	ENST00000399875	NM_001163079.1	ENSP00000382764	3

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL17RA	ENST00000612619	NM_001289905.1	ENSP00000479970	3
IL17RA	ENST00000319363	NM_014339.6	ENSP00000320936	3

Locus 33 (rs7221769)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,327,923-34,338,835
block size	10,913 bp
variant count	44 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.596 [-2.574 – 0.251]	gene(s) hit or close-by	CCL15, CCL15-CCL14, CCL23, RP11-104J23.1, RP11-104J23.2
phastCons	μ = 0.145 [0.001 – 0.938]	eQTL gene(s)	–
GERP++	μ = -0.086 [-2.21 – 0.158]	potentially regulated gene(s)	CCL14, CCL14, CCL15, CCL15, CCL16, CCL16
CADD score	μ = 4.547 [0.08 – 16.42]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ENSG00000256515	blood	1.82×10^{-2} (q-value)	SeeQTL DB (HapMap)	2
?	?	ENSG00000006075	blood	4.84×10^{-2} (q-value)	SeeQTL DB (HapMap)	1
?	?	ENSG00000129277	blood	1.08×10^{-2} (q-value)	SeeQTL DB (HapMap)	2
?	?	ENSG00000205021	blood	1.16×10^{-2} (q-value)	SeeQTL DB (HapMap)	2

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	1	no	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000022625		CCL15

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000204082	1	ENCP00000022625	CCL15 CCL15
		ENCP00000022623	CCL16 CCL16
ENCE00000204075	2	ENCP00000022624	CCL14 CCL14
		ENCP00000022622	CCL16 CCL16

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001344912 (promoter)	2	cervix (HeLa-S3)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3

		liver (HepG2)	H3K4me3, PolII, DNase1, RXRA, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K27me3
		blood (DND-41)	H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
ENSR00001344915	4	embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
(CTCF binding site)		Osteobl	CTCF
		blood (K562)	CTCF, Rad21, Max, H3K27me3
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF
		A549	CTCF, H3K27me3
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL15	upstream gene variant	2514	ENST00000614368	?	ENSP00000484262	11
CCL15	upstream gene variant	143	ENST00000617897	NM_032965.4	ENSP00000484078	19
CCL15-CCL14	upstream gene variant	377	ENST00000616694	?	ENSP00000481402	16
CCL15-CCL14	upstream gene variant	447	ENST00000610751	?	ENSP00000481940	15
CCL23	downstream gene variant	1262	ENST00000613876	?	ENSP00000479076	18
CCL23	downstream gene variant	1263	ENST00000615050	NM_005064.4	ENSP00000481357	18
CCL23	downstream gene variant	1261	ENST00000612516	NM_145898.2	ENSP00000484748	18
RP11-104J23.1	upstream gene variant, downstream gene variant	20	ENST00000619334	?	?	9
RP11-104J23.1	upstream gene variant, downstream gene variant	41	ENST00000617328	?	?	19
RP11-104J23.2	upstream gene variant	709	ENST00000617747	?	?	19

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15	ENST00000617897	NM_032965.4	ENSP00000484078	2
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	1
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	1
RP11-104J23.1	ENST00000619334	?	?	34
RP11-104J23.1	ENST00000617328	?	?	3

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15	ENST00000617897	NM_032965.4	ENSP00000484078	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-104J23.1	ENST00000619334	?	1
RP11-104J23.1	ENST00000617328	?	8

Locus 33 (sentinel SNP rs7221769 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 34 (rs10824796)

SNiPAcard

Block annotations

Block info

genomic range	chr10:54,531,685-54,536,083
block size	4,399 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.723 [-2.66 – 0.688]	gene(s) hit or close-by	MBL2
phastCons	μ = 0.001 [0 – 0.006]	eQTL gene(s)	MBL2
GERP++	μ = -0.672 [-4.14 – 2.24]	potentially regulated gene(s)	DKK1, MBL2, MBL2
CADD score	μ = 2.325 [0.134 – 5.949]	disease gene(s)	MBL2, DKK1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1-arachidonoylglycerophosphocholine*	7.59×10^{-5}	Metabolomics GWAS Server	24816252	2

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR952185	1

Disease gene annotation

gene	trait	source DB	source entry/link
MBL2	MANNOSE-BINDING PROTEIN DEFICIENCY	OMIM	MIM:614372
MBL2	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
DKK1	Idiopathic juvenile osteoporosis	OrphaNet	OrphaNet:85193

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MBL2	?	ENSG00000165471	tibial nerve	9.48×10^{-9} (p-value)	GTEx Portal V6	6
MBL2	?	ENSG00000165471	liver	3.29×10^{-10} (p-value)	GTEx Portal V6	6
MBL2	ENST00000373968	ILMN_1762464	liver	$<2.2\times10^{-16}$ (p-value)	Innocenti et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000074894	1	ENCP00000008487	DKK1
		ENCP00000008488	DKK1
		ENCP00000008495	MBL2 MBL2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MBL2	upstream gene variant	225	ENST00000373968	NM_000242.2	ENSP00000363079	6

Locus 35 (rs8112211)

SNiPAcard

Block annotations

Block info

genomic range	chr19:38,792,172-38,856,372
block size	64,201 bp
variant count	28 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.210 [-5.585 – 0.64]	gene(s) hit or close-by	AC005625.1, C19orf33, CATSPERG, CTB-102L5.4, KCNK6, YIF1B
phastCons	μ = 0.005 [0 – 0.054]	eQTL gene(s)	C19orf33, CATSPERG, PPP1R14A, SPINT2, YIF1B
GERP++	μ = -1.340 [-8.19 – 2.29]	potentially regulated gene(s)	PSMD8
CADD score	μ = 3.520 [0.004 – 13.82]	disease gene(s)	SPINT2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SPINT2	DIARRHEA 3, SECRETORY SODIUM, CONGENITAL, WITH OR WITHOUT OTHER [...]	OMIM	MIM:270420
SPINT2	Congenital sodium diarrhea	OrphaNet	OrphaNet:103908

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C19orf33	?	ENSG00000167644	muscularis mucosae	2.60×10^{-7} (p-value)	GTEx Portal V6	19
PPP1R14A	?	ENSG00000167641	muscularis mucosae	6.19×10^{-7} (p-value)	GTEx Portal V6	20
YIF1B	?	ENSG00000167645	tibial nerve	2.84×10^{-6} (p-value)	GTEx Portal V6	17
C19orf33	?	ENSG00000167644	tibial nerve	1.90×10^{-17} (p-value)	GTEx Portal V6	28
PPP1R14A	?	ENSG00000167641	tibial nerve	2.57×10^{-5} (p-value)	GTEx Portal V6	2
YIF1B	?	ENSG00000167645	sun exposed skin	2.70×10^{-9} (p-value)	GTEx Portal V6	28
CATSPERG	?	ENSG00000099338	sun exposed skin	2.74×10^{-6} (p-value)	GTEx Portal V6	3
C19orf33	?	ENSG00000167644	left ventricle	5.01×10^{-10} (p-value)	GTEx Portal V6	28
C19orf33	?	ENSG00000167644	blood	5.45×10^{-10} (p-value)	GTEx Portal V6	28
PPP1R14A	?	ENSG00000167641	blood	9.63×10^{-6} (p-value)	GTEx Portal V6	2
CATSPERG	?	ENSG00000099338	thyroid	2.25×10^{-8} (p-value)	GTEx Portal V6	4
C19orf33	?	ENSG00000167644	subcutaneous adipocytes	6.37×10^{-11} (p-value)	GTEx Portal V6	28
C19orf33	?	ENSG00000167644	spleen	4.01×10^{-6} (p-value)	GTEx Portal V6	1
C19orf33	?	ENSG00000167644	tibial artery	5.06×10^{-6} (p-value)	GTEx Portal V6	16
SPINT2	ENST00000454580	ILMN_1800739	blood	4.75×10^{-5} (p-value)	Westra et al.	1
SPINT2	ENST00000301244					
SPINT2	ENST00000587090					

SPINT2	ENST00000585357					
PPP1R14A	?	ENSG00000167641	sigmoid colon	5.00×10-7 (p-value)	GTEx Portal V6	14
?	?	ILMN_1717793	monocyte	8.66×10-58 (p-value)	Zeller et al.	3
PPP1R14A	ENST00000587515	ILMN_1761968	monocyte	5.24×10-36 (p-value)	Zeller et al.	3
PPP1R14A	ENST00000301242					
PPP1R14A	ENST00000347262					
PPP1R14A	ENST00000591291					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000227970	1	ENCP00000024343	PSMD8

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000346261 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, PolII, Yy1, TAF1, H3K4me2, SP1, H3K4me3, DNase1
		HSMMtube	DNase1
		blood (K562)	Egr1, Max, H3K4me1, PolII, H3K36me3, DNase1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1, H3K36me3
		liver (HepG2)	H3K4me1, H3K4me2, DNase1, CTCF, FOSL2
		blood (GM12878)	H3K36me3
		lung (IMR90)	DNase1, H3K36me3, CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac, H3K4me1
		NHLF	DNase1, CTCF, H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII, DNase1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001647943 (promoter)	1	embryonic stem cell (H1ESC)	H3K4me3, TAF1, H3K4me2, SP1, USF1, DNase1
		HSMMtube	H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K4me3, Gata2, Egr1, H3K27ac, PU1, H3K9ac, H3K79me2, PolII, H3K4me2, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K4me3
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		blood (GM12878)	H3K4me2, Tr4, PolII, H2AZ, DNase1, Egr1, H3K4me3
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K9ac, H3K4me2, DNase1, H3K4me3
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H3K4me3, H3K4me2
		blood (DND-41)	H3K4me2, H3K27ac, H3K4me3, H3K9ac
		breast (HMEC)	H3K4me2, DNase1, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K27ac, H3K4me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27ac, DNase1
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, H3K4me2, Max, H3K4me3
		A549	H3K27me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001647945 (CTCF binding site)	1	embryonic stem cell (H1ESC)	SIX5, Yy1, H3K4me2, SP1, USF1, H3K4me3, Rad21, Sin3Ak20, CTCF, DNase1
		HSMMtube	DNase1, CTCF, H3K4me2
		blood (K562)	Rad21, ELF1, H3K4me2, CTCF, DNase1, H3K4me3, H2AZ, Gabp, Yy1, THAP1, Max
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, DNase1, H3K4me2
		liver (HepG2)	Gabp, Yy1, Rad21, ELF1, H3K4me2, CTCF, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, H3K9ac, CTCF

	blood (GM12878)	H3K27ac, H3K4me2, CTCF, H3K4me3, Rad21, PolII, H2AZ, DNase1, Yy1, Gabp, ELF1
	nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
	skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
	NHLF	DNase1, H3K4me3, CTCF
	Osteobl	H2AZ, H3K4me3, H3K4me2, CTCF
	blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2
	breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2, CTCF
	cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, Gabp, CTCF, DNase1
	monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K4me3
	endothelium (HUVEC)	Max, H3K4me3, PolII, CTCF, DNase1
	A549	DNase1, CTCF, H3K4me2, H3K4me3
ENSR00000346269 2 (promoter)	embryonic stem cell (H1ESC)	H3K4me3, H3K36me3, H3K27me3, Rad21, Sin3Ak20, CTCF, SIX5, Yy1, H3K4me2, SP1, USF1, DNase1
	HSMMtube	DNase1, CTCF, H3K4me2, H2AZ
	blood (K562)	H3K4me2, CTCF, DNase1, H3K4me3, Yy1, THAP1, Egr1, H3K27ac, Max, H3K9ac, Gabp, H2AZ, Rad21, ELF1
	skin (NHDF-AD)	DNase1, H3K4me2, H3K4me3, CTCF
	muscle (HSMM)	DNase1, H3K36me3, H3K4me2, H3K4me3, CTCF, H2AZ
	liver (HepG2)	Yy1, Rad21, ELF1, H3K4me2, CTCF, H3K4me3, Gabp, DNase1
	lung (IMR90)	DNase1, H3K4me2, H3K4me3, H3K9ac, CTCF
	blood (GM12878)	PolII, H2AZ, DNase1, Yy1, Gabp, ELF1, Rad21, H3K4me3, H3K27ac, H3K4me2, CTCF
	nervous (NH-A)	DNase1, H3K4me2, CTCF, H3K9ac, H3K4me3
	skin (NHEK)	H3K9ac, H3K27ac, H3K4me2, DNase1, CTCF, H3K4me3
	NHLF	CTCF, DNase1, H3K4me3
	Osteobl	H2AZ, H3K4me3, H3K4me2, CTCF
	blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
	breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
	cervix (HeLa-S3)	DNase1, CTCF, Gabp, H3K9ac, H3K4me2, H3K27ac, H3K4me3
	monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
	endothelium (HUVEC)	H3K36me3, Max, H3K4me3, PolII, CTCF, DNase1
	A549	DNase1, CTCF, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC005625.1	downstream gene variant, upstream gene variant	1676	ENST00000590304	?	?	5
C19orf33	upstream gene variant, downstream gene variant	18	ENST00000591852	?	?	4
C19orf33	upstream gene variant, downstream gene variant	1540	ENST00000301246	NM_033520.1	ENSP00000301246	3
C19orf33	upstream gene variant, downstream gene variant	1543	ENST00000588605	?	ENSP00000464940	3
C19orf33	upstream gene variant, downstream gene variant	1546	ENST00000589986	?	?	3
CATSPERG	upstream gene variant, downstream gene variant	721	ENST00000477793	?	?	5
CATSPERG	upstream gene variant, downstream gene variant	472	ENST00000470292	?	?	3
CATSPERG	upstream gene variant	2159	ENST00000489693	?	ENSP00000470225	1
CATSPERG	upstream gene variant	1443	ENST00000475646	?	?	1
CATSPERG	upstream gene variant	1892	ENST00000471517	?	ENSP00000468028	3
CATSPERG	upstream gene variant, downstream gene variant	627	ENST00000467739	?	?	3
CATSPERG	upstream gene variant	4772	ENST00000492088	?	?	1
CATSPERG	upstream gene variant	1361	ENST00000412458	?	ENSP00000395093	1
CATSPERG	upstream gene variant, downstream gene variant	1521	ENST00000488473	?	?	2
CATSPERG	upstream gene variant, downstream gene variant	2055	ENST00000409410	?	ENSP00000386950	4
CATSPERG	upstream gene variant	1834	ENST00000410018	?	ENSP00000387057	3
CATSPERG	upstream gene variant, downstream gene variant	262	ENST00000585424	?	?	2
CATSPERG	upstream gene variant	1834	ENST00000409235	NM_021185.4	ENSP00000386962	3

CATSPERG	upstream gene variant	1877	ENST00000312265	?	ENSP00000311314	3
CATSPERG	upstream gene variant, downstream gene variant	1908	ENST00000475343	?	ENSP00000467947	4
CATSPERG	upstream gene variant, downstream gene variant	409	ENST00000466060	?	?	5
CTB-102L5.4	downstream gene variant	1639	ENST00000587519	?	ENSP00000465845	2
CTB-102L5.4	downstream gene variant	1640	ENST00000591889	?	ENSP00000468040	2
KCNK6	upstream gene variant, downstream gene variant	486	ENST00000263372	NM_004823.1	ENSP00000263372	9
KCNK6	upstream gene variant, downstream gene variant	2371	ENST00000588137	?	?	6
YIF1B	downstream gene variant, upstream gene variant	861	ENST00000586319	?	?	4
YIF1B	downstream gene variant, upstream gene variant	218	ENST00000592694	?	ENSP00000466428	7
YIF1B	downstream gene variant, upstream gene variant	396	ENST00000585563	?	?	7
YIF1B	downstream gene variant, upstream gene variant	339	ENST00000329420	NM_001039671.2, NM_001145461.1	ENSP00000329559	7
YIF1B	downstream gene variant, upstream gene variant	148	ENST00000591755	?	ENSP00000465446	7
YIF1B	downstream gene variant, upstream gene variant	209	ENST00000337679	NM_001145463.1	ENSP00000337411	7
YIF1B	downstream gene variant, upstream gene variant	591	ENST00000587039	?	ENSP00000465277	6
YIF1B	downstream gene variant, upstream gene variant	860	ENST00000392124	NM_033557.3	ENSP00000375971	4
YIF1B	downstream gene variant, upstream gene variant	190	ENST00000589644	?	?	7
YIF1B	downstream gene variant, upstream gene variant	333	ENST00000591784	NM_001145462.1	ENSP00000465230	7
YIF1B	downstream gene variant, upstream gene variant	879	ENST00000588002	?	ENSP00000468358	4
YIF1B	downstream gene variant, upstream gene variant	169	ENST00000339413	NM_001039673.2, NM_001039672.2	ENSP00000343435	7
YIF1B	downstream gene variant, upstream gene variant	25	ENST00000589247	?	ENSP00000468463	8
YIF1B	downstream gene variant, upstream gene variant	943	ENST00000592246	?	ENSP00000467489	4
YIF1B	downstream gene variant, upstream gene variant	369	ENST00000587361	?	?	8
YIF1B	downstream gene variant	1107	ENST00000589151	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CATSPERG	ENST00000409410	?	ENSP00000386950	T	acC/acT	1
CATSPERG	ENST00000409235	NM_021185.4	ENSP00000386962	T	acC/acT	1
YIF1B	ENST00000339413	NM_001039673.2, NM_001039672.2	ENSP00000343435	V	gtG/gtA	1
YIF1B	ENST00000588002	?	ENSP00000468358	V	gtG/gtA	1
YIF1B	ENST00000592694	?	ENSP00000466428	V	gtG/gtA	1
YIF1B	ENST00000591784	NM_001145462.1	ENSP00000465230	V	gtG/gtA	1
YIF1B	ENST00000337679	NM_001145463.1	ENSP00000337411	V	gtG/gtA	1
YIF1B	ENST00000329420	NM_001039671.2, NM_001145461.1	ENSP00000329559	V	gtG/gtA	1
YIF1B	ENST00000392124	NM_033557.3	ENSP00000375971	V	gtG/gtA	1
YIF1B	ENST00000592246	?	ENSP00000467489	V	gtG/gtA	1
YIF1B	ENST00000591755	?	ENSP00000465446	V	gtG/gtA	1

YIF1B	[illegible]	?	[illegible]	V	gtG/gtA

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CATSPERG	ENST00000409410	?	ENSP00000386950	10
CATSPERG	ENST00000489693	?	ENSP00000470225	6
CATSPERG	ENST00000585424	?	?	3
CATSPERG	ENST00000412458	?	ENSP00000395093	8
CATSPERG	ENST00000312265	?	ENSP00000311314	11
CATSPERG	ENST00000466060	?	?	3
CATSPERG	ENST00000409235	NM_021185.4	ENSP00000386962	11
CATSPERG	ENST00000410018	?	ENSP00000387057	11
CATSPERG	ENST00000475343	?	ENSP00000467947	3
CATSPERG	ENST00000471517	?	ENSP00000468028	11
CATSPERG	ENST00000470292	?	?	3
CTB-102L5.4	ENST00000591889	?	ENSP00000468040	2
CTB-102L5.4	ENST00000587519	?	ENSP00000465845	2
KCNK6	ENST00000588137	?	?	2
KCNK6	ENST00000263372	NM_004823.1	ENSP00000263372	2
YIF1B	ENST00000587039	?	ENSP00000465277	4
YIF1B	ENST00000587361	?	?	2
YIF1B	ENST00000586319	?	?	1
YIF1B	ENST00000339413	NM_001039673.2, NM_001039672.2	ENSP00000343435	3
YIF1B	ENST00000592694	?	ENSP00000466428	3
YIF1B	ENST00000591784	NM_001145462.1	ENSP00000465230	3
YIF1B	ENST00000337679	NM_001145463.1	ENSP00000337411	3
YIF1B	ENST00000329420	NM_001039671.2, NM_001145461.1	ENSP00000329559	3
YIF1B	ENST00000589644	?	?	2
YIF1B	ENST00000589151	?	?	1
YIF1B	ENST00000392124	NM_033557.3	ENSP00000375971	1
YIF1B	ENST00000592246	?	ENSP00000467489	1
YIF1B	ENST00000589247	?	ENSP00000468463	2
YIF1B	ENST00000591755	?	ENSP00000465446	2
YIF1B	ENST00000585563	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CATSPERG	ENST00000412458	?	ENSP00000395093	1
CATSPERG	ENST00000312265	?	ENSP00000311314	1
CATSPERG	ENST00000471517	?	ENSP00000468028	1
KCNK6	ENST00000263372	NM_004823.1	ENSP00000263372	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

C19orf33	ENST00000301246	NM_033520.1	ENSP00000301246	1
C19orf33	ENST00000588605	?	ENSP00000464940	1
CATSPERG	ENST00000409410	?	ENSP00000386950	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C19orf33	ENST00000589986	?	1
CATSPERG	ENST00000492088	?	1
CATSPERG	ENST00000585424	?	1
CATSPERG	ENST00000470292	?	1
YIF1B	ENST00000586319	?	1
YIF1B	ENST00000589644	?	1
YIF1B	ENST00000589151	?	1
YIF1B	ENST00000585563	?	1

Locus 36 (rs505922)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,137,065-136,149,500
block size	12,436 bp
variant count	35 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.587 [-2.293 – 0.687]	gene(s) hit or close-by	ABO
phastCons	μ = 0.133 [0 – 0.646]	eQTL gene(s)	ABO, GBGT1
GERP++	μ = -0.006 [-0.989 – 1.37]	potentially regulated gene(s)	ABO, ABO, AK8, SARDH, TSC1, TSC1
CADD score	μ = 2.446 [0.195 – 8.142]	disease gene(s)	TSC1, SARDH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
ADpSGEGDFXAEGGGVR*	9.66×10^{-14}	Metabolomics GWAS Server	24816252	11

Disease gene annotation

gene	trait	source DB	source entry/link
TSC1	FOCAL CORTICAL DYSPLASIA OF TAYLOR	OMIM	MIM:607341
TSC1	TUBEROUS SCLEROSIS 1	OMIM	MIM:191100
SARDH	SARCOSINEMIA	OMIM	MIM:268900
TSC1	tuberous sclerosis type 1 (TSC1)	DECIPHER	MIM:191100
TSC1	Tuberous sclerosis	OrphaNet	OrphaNet:805
TSC1	LYMPHANGIOLEIOMYOMATOSIS	OrphaNet	OrphaNet:538
TSC1	Isolated focal cortical dysplasia type IIb	OrphaNet	OrphaNet:269008
SARDH	SARCOSINEMIA	OrphaNet	OrphaNet:3129

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABO	?	ENSG00000175164	pancreas	1.36×10^{-13} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	lung	7.77×10^{-12} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	atrial appendage	8.21×10^{-9} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	thyroid	8.24×10^{-16} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	skeletal muscle	5.22×10^{-8} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	transverse colon	1.80×10^{-9} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	sun exposed skin	4.00×10^{-6} (p-value)	GTEx Portal V6	26
ABO	?	ENSG00000175164	unexposed skin	5.93×10^{-6} (p-value)	GTEx Portal V6	1
ABO	?	ENSG00000175164	ovary	4.63×10^{-13} (p-value)	GTEx Portal V6	34

ABO	?	ENSG00000175164	visceral adipocytes	2.24×10-11 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	tibial nerve	1.62×10-9 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	vagina	2.69×10-9 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	esophagus mucosa	1.79×10-32 (p-value)	GTEx Portal V6	34
GBGT1 GBGT1 GBGT1 GBGT1 GBGT1	ENST00000372040 ENST00000470431 ENST00000472281 ENST00000372043 ENST00000540636	ILMN_1652906	blood	5.72×10-7 (p-value)	Westra et al.	11
ABO	?	ENSG00000175164	adrenal gland	2.39×10-21 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	testis	4.95×10-9 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	subcutaneous adipocytes	2.56×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000558384	1	ENCP00000060472	TSC1 TSC1
ENCE00000558361	1	ENCP00000060497	ABO ABO
		ENCP00000060532	SARDH
		ENCP00000060469	AK8

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000251881 (TF binding site)	1	lung (IMR90)	H3K27me3
		blood (K562)	H3K36me3, DNase1, Brg1, Ini1
		A549	H3K27me3
ENSR00001475637 (TF binding site)	5	embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K36me3, H3K4me2, H3K9ac, H3K27ac, DNase1, Brg1, Ini1, PolII
		A549	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000538324	?	ENSP00000483018	35
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	35
ABO	ENST00000453660	?	?	35

Locus 37 (rs342706)

SNiPAcard

Block annotations

Block info

genomic range	chr13:92,414,715-92,453,051
block size	38,337 bp
variant count	76 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.632 [-5.445 – 1.165]	gene(s) hit or close-by	GPC5
phastCons	μ = 0.023 [0 – 0.991]	eQTL gene(s)	–
GERP++	μ = -0.480 [-5.75 – 2.7]	potentially regulated gene(s)	FABP5P4
CADD score	μ = 2.705 [0.015 – 13.41]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
LUNG CANCER	<6.00×10-6	GWAS Catalog	20304703	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000161614	2	ENCP00000018923	FABP5P4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000518738 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	Rad21, CTCF
		blood (K562)	CTCF

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPC5	ENST00000377067	NM_004466.4	ENSP00000366267	76

Locus 38 (rs3742264)

SNiPAcard

Block annotations

Block info

genomic range	chr13:46,602,162-46,655,449
block size	53,288 bp
variant count	63 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.509 [-2.944 – 1.624]	gene(s) hit or close-by	CPB2, CPB2-AS1, ZC3H13
phastCons	μ = 0.064 [0 – 1]	eQTL gene(s)	CPB2, CPB2-AS1, ZC3H13
GERP++	μ = -0.460 [-5.92 – 2.82]	potentially regulated gene(s)	LCP1, LRRC63
CADD score	μ = 4.005 [0.041 – 18.87]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR080756	1
?	HGMD curated	HGMD	CM063919	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CPB2	missense variant	ENST00000439329	NM_001278541.1	ENSP00000400714	T/A	Aca/Gca	?	?	1
CPB2	missense variant	ENST00000181383	NM_001872.4	ENSP00000181383	T/A	Aca/Gca	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CPB2-AS1	?	ENSG00000235903	nucleus accumbens	5.04×10^{-10} (p-value)	GTEx Portal V6	62
ZC3H13	?	ENSG00000123200	sun exposed skin	5.31×10^{-7} (p-value)	GTEx Portal V6	43
CPB2	?	ENSG00000080618	sun exposed skin	7.22×10^{-6} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	thyroid	4.23×10^{-8} (p-value)	GTEx Portal V6	61
CPB2-AS1	?	ENSG00000235903	cerebellum	2.92×10^{-8} (p-value)	GTEx Portal V6	59
CPB2-AS1	?	ENSG00000235903	cerebellar hemisphere	4.23×10^{-6} (p-value)	GTEx Portal V6	53
CPB2-AS1	?	ENSG00000235903	pituitary	3.78×10^{-7} (p-value)	GTEx Portal V6	54
CPB2-AS1	?	ENSG00000235903	caudate basal ganglia	1.48×10^{-6} (p-value)	GTEx Portal V6	4
CPB2-AS1	?	ENSG00000235903	liver	2.97×10^{-6} (p-value)	GTEx Portal V6	1

CPB2-AS1	?	ENSG00000235903	lung	4.50×10-7 (p-value)	GTEx Portal V6	4
CPB2-AS1	?	ENSG00000235903	hypothalamus	3.14×10-7 (p-value)	GTEx Portal V6	2
CPB2-AS1	?	ENSG00000235903	sigmoid colon	2.59×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000157095	2	ENCP00000018502	LCP1
		ENCP00000018503	LRRC63 LCP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000515658 (promoter)	1	embryonic stem cell (H1ESC)	CTCF, PolII, Gabp, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K9ac, H3K79me2, H3K27ac, DNase1, H2AZ, H3K4me3
		blood (K562)	H3K4me3, H3K4me2, PolII, TAF1, H2AZ, H3K79me2, Gabp, H3K9ac, CTCF, Max, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K9ac, DNase1, H3K4me2, H3K4me3, H2AZ, H3K79me2
		liver (HepG2)	H3K27me3, DNase1, H3K36me3, Gabp, PolII, H3K79me2, ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3
		lung (IMR90)	H3K4me2, H3K4me3, H3K9ac, DNase1
		blood (GM12878)	DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H2AZ, PolII, CTCF, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K9ac, H3K4me3, DNase1, H3K27ac
		Osteobl	H3K4me3, H3K4me2, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac, DNase1, PolII, Gabp, H3K79me2
		monocytes (Monocytes-CD14+)	H3K9ac, H3K27me3, H3K4me3, H3K27ac, H3K4me2, DNase1
		endothelium (HUVEC)	Max, H3K4me2, H3K4me3, H3K9ac, PolII, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001509498 (CTCF binding site)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	Rad21, CTCF, USF1
		liver (HepG2)	H3K36me3, Rad21
		HSMMtube	H3K27me3
		blood (GM12878)	BATF, MEF2A, H2AZ, Rad21, H3K27ac, CTCF
		nervous (NH-A)	DNase1
		blood (K562)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ZC3H13	upstream gene variant	558	ENST00000242848	?	ENSP00000242848	1
ZC3H13	upstream gene variant	619	ENST00000428921	?	ENSP00000416238	1
ZC3H13	upstream gene variant	545	ENST00000282007	NM_015070.4	ENSP00000282007	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	D	gaC/gaT	1

Intron variant

intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	58
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	58
CPB2-AS1	ENST00000606991	?	?	59
CPB2-AS1	ENST00000606351	?	?	59
CPB2-AS1	ENST00000415033	?	?	59
CPB2-AS1	ENST00000606243	?	?	59
ZC3H13	ENST00000428921	?	ENSP00000416238	2
ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	4
ZC3H13	ENST00000242848	?	ENSP00000242848	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	1
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	1

Locus 39 (rs7250849)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,151,087-52,160,765
block size	9,679 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.381 [-1.801 – -0.878]	**gene(s) hit or close-by**	RPL7P51, SIGLEC14, SIGLEC5, SIGLEC5
phastCons	μ = 0.002 [0 – 0.006]	**eQTL gene(s)**	RPL7P51, SIGLEC14, SIGLEC5
GERP++	μ = -1.326 [-2 – -0.513]	**potentially regulated gene(s)**	–
CADD score	μ = 1.757 [0.015 – 5.867]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC14	?	ENSG00000254415	visceral adipocytes	7.21×10^{-9} (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	muscularis mucosae	1.95×10^{-13} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	lung	1.95×10^{-13} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	tibial nerve	2.48×10^{-16} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	sigmoid colon	1.04×10^{-8} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	atrial appendage	9.58×10^{-10} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	tibial artery	1.27×10^{-13} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	blood	7.77×10^{-16} (p-value)	GTEx Portal V6	4
SIGLEC5	?	ENSG00000105501	blood	8.62×10^{-11} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	thyroid	1.55×10^{-14} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	gastroesophageal junction	1.81×10^{-10} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	testis	1.18×10^{-8} (p-value)	GTEx Portal V6	4
SIGLEC5	?	ENSG00000105501	testis	6.00×10^{-6} (p-value)	GTEx Portal V6	1
SIGLEC14	?	ENSG00000254415	sun exposed skin	3.17×10^{-10} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	aorta	6.40×10^{-10} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	left ventricle	5.61×10^{-9} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	subcutaneous adipocytes	2.87×10^{-15} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	breast	3.54×10^{-9} (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	skeletal muscle	3.82×10^{-8} (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	EBV lymphocytes	2.75×10^{-8} (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	transverse colon	1.84×10^{-8} (p-value)	GTEx Portal V6	3

SIGLEC5	?	ENSG00000105501	blood	4.75×10-2 (q-value)	SeeQTL DB (HapMap)	1
SIGLEC14	?	ENSG00000254415	caudate basal ganglia	6.53×10-8 (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	spleen	4.15×10-9 (p-value)	GTEx Portal V6	3
RPL7P51	?	ENSG00000243469	spleen	3.14×10-7 (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	cortex	6.04×10-7 (p-value)	GTEx Portal V6	1
SIGLEC14	?	ENSG00000254415	frontal cortex	4.23×10-9 (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	pancreas	5.09×10-7 (p-value)	GTEx Portal V6	2
SIGLEC14	?	ENSG00000254415	esophagus mucosa	4.64×10-8 (p-value)	GTEx Portal V6	1
RPL7P51	?	ENSG00000243469	nucleus accumbens	1.57×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000641970 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	CTCF, Rad21
ENSR00000199573 (CTCF binding site)	2	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	CTCF, DNase1
		blood (K562)	Max, DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		blood (GM12878)	DNase1, Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	DNase1, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RPL7P51	upstream gene variant	1028	ENST00000490532	?	?	1
SIGLEC14	upstream gene variant	1033	ENST00000360844	NM_001098612.1	ENSP00000354090	1
SIGLEC14	upstream gene variant	2257	ENST00000533866	?	?	1
SIGLEC5	upstream gene variant	2289	ENST00000534261	?	ENSP00000473238	1
SIGLEC5	upstream gene variant	1034	ENST00000599649	?	ENSP00000470259	1
SIGLEC5	upstream gene variant	945	ENST00000429354	?	ENSP00000415200	1

Locus 40 (rs9013)

SNiPAcard

Block annotations

Block info

genomic range	chr2:143,778,115-143,810,757
block size	32,643 bp
variant count	28 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.207 [-2.518 – 1.857]	gene(s) hit or close-by	KYNU
phastCons	μ = 0.063 [0 – 0.402]	eQTL gene(s)	–
GERP++	μ = -0.222 [-6.91 – 4.36]	potentially regulated gene(s)	–
CADD score	μ = 4.571 [0.063 – 13.06]	disease gene(s)	KYNU

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
KYNU	HYDROXYKYNURENINURIA	OMIM	MIM:236800
KYNU	Encephalopathy due to hydroxykynureninuria	OrphaNet	OrphaNet:79155

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
KYNU	missense variant	ENST00000264170	NM_003937.2	ENSP00000264170	E/K	Gaa/Aaa	?	?	1
KYNU	missense variant	ENST00000409512	NM_001199241.1	ENSP00000386731	E/K	Gaa/Aaa	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1746517	monocyte	2.76×10^{-5} (p-value)	Fairfax et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001657233 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF
		endothelium (HUVEC)	H3K36me3
		HSMMtube	H3K27me3
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KYNU	downstream gene variant	166	ENST00000409512	NM_001199241.1	ENSP00000386731	10

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KYNU	ENST00000264170	NM_003937.2	ENSP00000264170	7
KYNU	ENST00000409512	NM_001199241.1	ENSP00000386731	7

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
KYNU	ENST00000264170	NM_003937.2	ENSP00000264170	20

Locus 41 (rs2004318)

SNiPAcard

Block annotations

Block info

genomic range	chr19:55,117,749-55,168,602
block size	50,854 bp
variant count	74 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.557 [-4.065 – 2.368]	**gene(s) hit or close-by**	AC009892.10, AC009892.5, AC009892.9, AC245036.1, AC245036.2, AC245036.3, AC245036.4, AC245036.5, AC245036.6, LILRA1, LILRB1, LILRB4, MIR8061, VN1R105P
phastCons	μ = 0.059 [0 – 0.633]	**eQTL gene(s)**	CTB-83J4.2, LILRA1, LILRA2, LILRB2, LILRB5, LILRP1
GERP++	μ = -0.651 [-4.69 – 2.07]	**potentially regulated gene(s)**	AC008984.5, AC008984.5, AC008984.6, AC008984.6, AC008984.7, AC008984.7, AC009892.10, AC009892.10, AC009892.2, AC009892.2, AC009892.5, AC010518.3, AC010518.3, AC011515.2, AC011515.2, AC012314.19, AC012314.19, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, KIR2DL1, KIR2DL1, KIR2DL1

trait	min(p-value)	source DB	source entry/link	variant(s)
3-(4-hydroxyphenyl)lactate	3.53×10^{-5}	Metabolomics GWAS Server	24816252	3

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
LILRB1	missense variant	ENST00000421584	?	ENSP00000410165	5	5			5
LILRB1	missense variant	ENST00000396315	?	ENSP00000379608	5	5			5
LILRB1	missense variant	ENST00000427581	?	ENSP00000395004	5	5			5
LILRB1	missense variant	ENST00000396332	NM_001081639.2	ENSP00000379623	5	5			5
LILRB1	missense variant	ENST00000396327	NM_001081638.2	ENSP00000379618	5	5			5
LILRB1	missense variant	ENST00000324602	NM_001081637.1	ENSP00000315997	5	5			5
LILRB1	missense variant	ENST00000396317	NM_001278398.1	ENSP00000379610	5	5			5
LILRB1	missense variant	ENST00000396331	NM_006669.5	ENSP00000379622	5	5			5
LILRB1	missense variant	ENST00000418536	?	ENSP00000391514	A/T	Gcc/Acc	?	?	1
LILRB1	missense variant	ENST00000396321	?	ENSP00000379614	A/T	Gcc/Acc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LILRA1	?	ENSG00000104974	lung	4.15×10^{-7} (p-value)	GTEx Portal V6	39
LILRA2	?	ENSG00000239998	lung	1.26×10^{-7} (p-value)	GTEx Portal V6	61
LILRP1	?	ENSG00000186152	EBV lymphocytes	6.26×10^{-8} (p-value)	GTEx Portal V6	13
LILRB5	?	ENSG00000105609	EBV lymphocytes	1.02×10^{-7} (p-value)	GTEx Portal V6	51
LILRB2	?	ENSG00000131042	EBV lymphocytes	5.43×10^{-9} (p-value)	GTEx Portal V6	56
CTB-83J4.2	?	ENSG00000269271	blood	4.50×10^{-8} (p-value)	GTEx Portal V6	38
LILRA1	?	ENSG00000104974	blood	2.28×10^{-9} (p-value)	GTEx Portal V6	52
LILRA2	?	ENSG00000239998	subcutaneous adipocytes	1.86×10^{-8} (p-value)	GTEx Portal V6	5

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000344796	1	ENST00000418536, ENST00000396321	LILRB1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024800		

ENCP00000024805			LILRB1 LILRB1 LILRB1 LILRB1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000233609	1	ENCP00000024802	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024807	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
		ENCP00000024761	AC008984.5 AC008984.5
		ENCP00000024823	KIR2DS4 KIR2DS4
		ENCP00000024769	LAIR1 LAIR1 LAIR1 LAIR1 LAIR1
		ENCP00000024758	LILRB2 LILRB2 LILRB2 LILRB2 LILRB2
		ENCP00000024812	AC011515.2 AC011515.2
		ENCP00000024753	LILRA6 LILRA6 LILRA6 LILRA6 LILRA6
		ENCP00000024817	KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3

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ENCP00000024829	NLRP2
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	NLRP2
	NLRP2
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ENCP00000024788	LENG8
	LENG8
	LENG8
	LENG8
ENCP00000024803	LILRB1
	LILRB1
	LILRB1
	LILRB1
	LILRB1
ENCP00000024773	LAIR1
	LAIR1
	LAIR1
	LAIR1
	LAIR1
ENCP00000024808	VN1R105P
ENCP00000024763	LILRA5
	AC008984.7
	AC008984.7
	LILRA5
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ENCP00000024821	KIR2DL4
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ENCP00000024772	LAIR1
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ENCP00000024825	FCAR
	FCAR
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ENCP00000024771	LAIR1
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ENCP00000024810	LILRB4
	LILRB4
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ENCP00000024759	AC010518.3
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ENCP00000024819	KIR2DL1
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ENCP00000024799	LILRA1
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ENCP00000024751	AC012314.19
	AC012314.19
ENCP00000024811	LILRB4
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ENCP00000024768	LAIR1
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	LAIR1

ENCP00000024756	LILRB2 LILRB2 LILRB2 LILRB2 LILRB2
ENCP00000024764	LILRA5 LILRA5 LILRA5 LILRA5 LILRA5
ENCP00000024822	KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1
ENCP00000024818	KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1
ENCP00000024816	KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3

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		ENCP00000024760	LILRA3
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			LILRA3
			LILRA3
		ENCP00000024806	AC009892.10
			AC009892.10
		ENCP00000024798	LILRA2
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		ENCP00000024797	LILRA2
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		ENCP00000024824	KIR3DL2
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		ENCP00000024800	AC009892.5
		ENCP00000024770	LAIR1
			LAIR1
			LAIR1
			LAIR1
			LAIR1
ENCE00000233672	1	ENCP00000024805	LILRB1
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			LILRB1
		ENCP00000024796	AC009892.2
			AC009892.2

		ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024801	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024826	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1
ENCE00000233675	1	ENCP00000024805	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024796	AC009892.2 AC009892.2
		ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024801	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024826	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1
ENCE00000233680	3	ENCP00000024805	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024796	AC009892.2 AC009892.2
		ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024826	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024801	LILRB1

		LILRB1 LILRB1 LILRB1 LILRB1
	ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348881 (open chromatin region)	6	blood (GM12878)	H3K4me3, H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		A549	H3K27me3
ENSR00000348884 (promoter)	3	cervix (HeLa-S3)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	Yy1, PU1, PolII, BATF, DNase1, IRF4, H3K79me2, H3K4me3, BCL11A, Pax5, H3K27ac, H3K4me2, H3K9ac
		A549	H3K27me3
		blood (K562)	H3K27me3
ENSR00001649225 (promoter)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H3K9ac, DNase1, H3K4me1, H3K4me2, H3K27ac
		blood (GM12878)	PolII, H3K36me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		A549	H3K27me3
ENSR00000348890 (open chromatin region)	2	blood (K562)	USF1, H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC009892.10	downstream gene variant, upstream gene variant	347	ENST00000456337	?	?	14
AC009892.5	upstream gene variant, downstream gene variant	72	ENST00000432400	?	?	15
AC009892.9	upstream gene variant, downstream gene variant	2348	ENST00000429259	?	?	5
AC245036.1	downstream gene variant, upstream gene variant	20	ENST00000578908	?	?	22
AC245036.2	upstream gene variant, downstream gene variant	1613	ENST00000618840	?	?	4
AC245036.3	upstream gene variant, downstream gene variant	2000	ENST00000610849	?	?	4
AC245036.4	upstream gene variant, downstream gene variant	1785	ENST00000622837	?	?	4
AC245036.5	upstream gene variant, downstream gene variant	1871	ENST00000616955	?	?	4
AC245036.6	upstream gene variant, downstream gene variant	2086	ENST00000614474	?	?	4
LILRA1	downstream gene variant	4407	ENST00000473156	?	?	3
LILRA1	downstream gene variant	4194	ENST00000453777	NM_001278318.1	ENSP00000413715	4
LILRB1	upstream gene variant, downstream gene variant	195	ENST00000487425	?	?	22
LILRB1	upstream gene variant	225	ENST00000630741	?	ENSP00000485956	11
LILRB1	upstream gene variant	187	ENST00000418536	?	ENSP00000391514	11
LILRB1	upstream gene variant, downstream gene variant	37	ENST00000480375	?	?	20
LILRB1	upstream gene variant, downstream gene variant	225	ENST00000396315	?	ENSP00000379608	13
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000396332	NM_001081639.2	ENSP00000379623	12
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000324602	NM_001081637.1	ENSP00000315997	12
LILRB1	upstream gene variant	225	ENST00000396321	?	ENSP00000379614	11
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000396331	NM_006669.5	ENSP00000379622	6
LILRB1	upstream gene variant, downstream gene variant	613	ENST00000427581	?	ENSP00000395004	12
LILRB1	upstream gene variant, downstream gene variant	225	ENST00000396317	NM_001278308.1	ENSP00000379610	13

LILRB1	upstream gene variant, downstream gene variant	225	ENST00000396317	NM_001278398.1	ENSP00000379610	13
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000462628	?	?	10
LILRB1	upstream gene variant, downstream gene variant	477	ENST00000473412	?	?	16
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000396327	NM_001081638.2	ENSP00000379618	12
LILRB1	upstream gene variant, downstream gene variant	225	ENST00000421584	?	ENSP00000410165	13
LILRB1	upstream gene variant, downstream gene variant	333	ENST00000480257	?	?	28
LILRB4	upstream gene variant, downstream gene variant	137	ENST00000461839	?	?	2
LILRB4	upstream gene variant	4977	ENST00000391736	NM_001278426.2	ENSP00000375616	1
LILRB4	upstream gene variant	3684	ENST00000270452	?	ENSP00000270452	1
MIR8061	downstream gene variant	1485	ENST00000618936	?	?	3
VN1R105P	upstream gene variant, downstream gene variant	117	ENST00000486595	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	2	3	3
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	2	3	3
LILRB1	ENST00000427581	?	ENSP00000395004	2	3	3
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	2	3	3
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	2	3	3
LILRB1	ENST00000421584	?	ENSP00000410165	2	2	3
LILRB1	ENST00000396315	?	ENSP00000379608	2	3	3
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	2	2	3

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
LILRB1	ENST00000462628	?	?	1
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	1
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	1
LILRB1	ENST00000427581	?	ENSP00000395004	1
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	1
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	1
LILRB1	ENST00000421584	?	ENSP00000410165	1
LILRB1	ENST00000396315	?	ENSP00000379608	1
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC009892.10	ENST00000456337	?	?	7
LILRB1	ENST00000462628	?	?	15
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	39
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	14
LILRB1	ENST00000480257	?	?	1

LILRB1	ENST00000427581	?	ENSP00000395004	14
LILRB1	ENST00000480375	?	?	1
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	14
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	14
LILRB1	ENST00000421584	?	ENSP00000410165	13
LILRB1	ENST00000396315	?	ENSP00000379608	13
LILRB1	ENST00000396321	?	ENSP00000379614	25
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	13
LILRB1	ENST00000418536	?	ENSP00000391514	25
LILRB1	ENST00000487425	?	?	1
LILRB1	ENST00000630741	?	ENSP00000485956	26
LILRB4	ENST00000270452	?	ENSP00000270452	6
LILRB4	ENST00000461839	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LILRB1	ENST00000421584	?	ENSP00000410165	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LILRB1	ENST00000427581	?	ENSP00000395004	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC009892.5	ENST00000432400	?	3
LILRB1	ENST00000462628	?	13
LILRB1	ENST00000473412	?	8
LILRB4	ENST00000461839	?	1

Locus 42 (rs4725617)

SNiPAcard

Block annotations

Block info

genomic range	chr7:143,092,918-143,108,081
block size	15,164 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.574 [-1.838 – 0.994]	gene(s) hit or close-by	EPHA1, EPHA1-AS1, ZYX
phastCons	μ = 0.026 [0 – 0.13]	eQTL gene(s)	EPHA1, EPHA1-AS1, ZYX
GERP++	μ = 0.477 [-1.57 – 3.88]	potentially regulated gene(s)	–
CADD score	μ = 5.976 [2.371 – 12.8]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
EPHA1	missense variant	ENST00000275815	NM_005232.4	ENSP00000275815	V/A	gTg/gCg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
EPHA1-AS1	?	ENSG00000229153	blood	3.02×10^{-8} (p-value)	GTEx Portal V6	2
ZYX	?	ENSG00000159840	blood	2.45×10^{-8} (p-value)	GTEx Portal V6	3
EPHA1	?	ENSG00000146904	sun exposed skin	2.87×10^{-7} (p-value)	GTEx Portal V6	8
EPHA1	?	ENSG00000146904	thyroid	6.98×10^{-7} (p-value)	GTEx Portal V6	8

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001564739 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	DNase1, CTCF
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	DNase1, H3K4me1, PolII
		liver (HepG2)	H3K4me1, H3K4me2, HNF4A, DNase1, RXRA
		lung (IMR90)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000069717 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF, Rad21, H3K36me3
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	CTCF
		breast (HMEC)	H3K36me3
		endothelium (HUVEC)	DNase1, PolII, H3K4me1

		liver (HepG2)	RXRA, H3K4me1, H3K4me2, HNF4A, DNase1
		lung (IMR90)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000635676 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K36me3
ENSR00001718722 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, H3K4me3, PolII, Rad21, Egr1, Nrsf, CTCF, TAF1, SP1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3, Egr1, CTCF, DNase1
		breast (HMEC)	CTCF, H3K4me3
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	CTCF
		liver (HepG2)	H3K4me3, H3K4me2, DNase1
		blood (GM12878)	ZEB1
		lung (IMR90)	CTCF
		nervous (NH-A)	H3K27me3
		A549	H3K4me3, H3K4me2, H3K27me3
		skin (NHEK)	DNase1, CTCF, H3K4me3
ENSR00000635678 (promoter flanking region)	2	monocytes (Monocytes-CD14+)	H3K4me3, H3K27ac, H3K4me1, DNase1
		embryonic stem cell (H1ESC)	H3K4me2, DNase1
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
EPHA1	upstream gene variant	4434	ENST00000458129	?	?	2
EPHA1	downstream gene variant, upstream gene variant	819	ENST00000497891	?	?	6
EPHA1	downstream gene variant, upstream gene variant	1267	ENST00000479459	?	?	4
EPHA1	upstream gene variant	224	ENST00000465208	?	?	3
EPHA1	upstream gene variant	1041	ENST00000275815	NM_005232.4	ENSP00000275815	4
EPHA1	upstream gene variant	1128	ENST00000488068	?	?	4
EPHA1	upstream gene variant	3326	ENST00000494989	?	?	2
EPHA1-AS1	upstream gene variant	1526	ENST00000429289	?	?	2
EPHA1-AS1	upstream gene variant	1546	ENST00000421648	?	?	2
ZYX	downstream gene variant	4716	ENST00000354434	?	ENSP00000346417	1
ZYX	downstream gene variant	4714	ENST00000322764	NM_003461.4, NM_001010972.1	ENSP00000324422	1
ZYX	downstream gene variant	4716	ENST00000392910	?	ENSP00000376642	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
EPHA1	ENST00000497891	?	?	3
EPHA1	ENST00000488068	?	?	5
EPHA1	ENST00000275815	NM_005232.4	ENSP00000275815	5
EPHA1	ENST00000494989	?	?	2

EPHA1-AS1	ENST00000421648	?	?	4
EPHA1-AS1	ENST00000429289	?	?	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
EPHA1	ENST00000497891	?	1
EPHA1	ENST00000488068	?	2

Locus 43 (rs1042445)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,393,547-186,397,474
block size	3,928 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.544 [-1.333 – -0.092]	gene(s) hit or close-by	HRG, RP11-573D15.8
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	–
GERP++	μ = 0.212 [-1.91 – 2.35]	potentially regulated gene(s)	–
CADD score	μ = 5.246 [2.169 – 15.47]	disease gene(s)	HRG

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HRG	THROMBOPHILIA DUE TO HISTIDINE-RICH GLYCOPROTEIN DEFICIENCY	OMIM	MIM:613116
HRG	Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency	OrphaNet	OrphaNet:217467

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
HRG	missense variant	ENST00000232003	NM_000412.3	ENSP00000232003	C/R	Tgt/Cgt	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HRG	downstream gene variant	596	ENST00000495413	?	?	2
HRG	downstream gene variant	587	ENST00000232003	NM_000412.3	ENSP00000232003	2
RP11-573D15.8	upstream gene variant	1240	ENST00000626006	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HRG	ENST00000495413	?	?	3
HRG	ENST00000232003	NM_000412.3	ENSP00000232003	2
RP11-573D15.8	ENST00000627469	?	?	5
RP11-573D15.8	ENST00000626649	?	?	5
RP11-573D15.8	ENST00000625877	?	?	5

RP11-573D15.8	ENST00000625386	?	?	5
RP11-573D15.8	ENST00000630331	?	?	5
RP11-573D15.8	ENST00000628190	?	?	5
RP11-573D15.8	ENST00000630726	?	?	5
RP11-573D15.8	ENST00000630315	?	?	5
RP11-573D15.8	ENST00000626633	?	?	5
RP11-573D15.8	ENST00000630178	?	?	5
RP11-573D15.8	ENST00000629106	?	?	5
RP11-573D15.8	ENST00000627830	?	?	5
RP11-573D15.8	ENST00000625741	?	?	5
RP11-573D15.8	ENST00000628253	?	?	5
RP11-573D15.8	ENST00000625303	?	?	5
RP11-573D15.8	ENST00000629451	?	?	5
RP11-573D15.8	ENST00000626845	?	?	5
RP11-573D15.8	ENST00000628505	?	?	5
RP11-573D15.8	ENST00000625839	?	?	5
RP11-573D15.8	ENST00000630864	?	?	5
RP11-573D15.8	ENST00000628728	?	?	5

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
HRG	ENST00000495413	?	1

Locus 44 (rs12712135)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,917,234-102,954,190
block size	36,957 bp
variant count	73 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.743 [-2.876 – 1.082]	gene(s) hit or close-by	IL18R1, IL1RL1
phastCons	μ = 0.026 [0 – 0.241]	eQTL gene(s)	IL18R1, IL18RAP, IL1RL1, SLC9A4
GERP++	μ = -0.524 [-7.5 – 3.17]	potentially regulated gene(s)	IL18R1, IL1R1, IL1R2, IL1RL1, MAP4K4
CADD score	μ = 3.025 [0.041 – 10.23]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Leprosy	$<2.00\times10^{-23}$	GWAS Catalog	25642632	1
tryptophan	4.17×10^{-5}	Metabolomics GWAS Server	24816252	20

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL18R1	?	ENSG00000115604	caudate basal ganglia	1.01×10^{-18} (p-value)	GTEx Portal V6	67
IL1RL1	?	ENSG00000115602	caudate basal ganglia	2.07×10^{-15} (p-value)	GTEx Portal V6	65
IL18R1	?	ENSG00000115604	cortex	3.07×10^{-14} (p-value)	GTEx Portal V6	66
IL1RL1	?	ENSG00000115602	cortex	4.08×10^{-14} (p-value)	GTEx Portal V6	64
IL18R1	?	ENSG00000115604	lung	1.35×10^{-7} (p-value)	GTEx Portal V6	61
IL1RL1	?	ENSG00000115602	lung	1.61×10^{-33} (p-value)	GTEx Portal V6	67
IL18R1	?	ENSG00000115604	nucleus accumbens	1.85×10^{-14} (p-value)	GTEx Portal V6	64
IL1RL1	?	ENSG00000115602	nucleus accumbens	1.19×10^{-13} (p-value)	GTEx Portal V6	63
IL18RAP	?	ENSG00000115607	blood	1.45×10^{-8} (p-value)	GTEx Portal V6	64
IL18R1	?	ENSG00000115604	anterior cingulate cortex	2.02×10^{-12} (p-value)	GTEx Portal V6	64
IL1RL1	?	ENSG00000115602	anterior cingulate cortex	7.46×10^{-10} (p-value)	GTEx Portal V6	63
IL18R1	?	ENSG00000115604	cerebellar hemisphere	1.51×10^{-12} (p-value)	GTEx Portal V6	65
IL1RL1	?	ENSG00000115602	cerebellar hemisphere	5.24×10^{-9} (p-value)	GTEx Portal V6	63
SLC9A4	?	ENSG00000180251	testis	3.35×10^{-13} (p-value)	GTEx Portal V6	65
IL18R1	?	ENSG00000115604	putamen	2.73×10^{-13} (p-value)	GTEx Portal V6	64
IL1RL1	?	ENSG00000115602	putamen	2.03×10^{-11} (p-value)	GTEx Portal V6	62

IL18R1	?	ENSG00000115604	cerebellum	1.10×10-15 (p-value)	GTEx Portal V6	67
IL1RL1	?	ENSG00000115602	cerebellum	1.21×10-13 (p-value)	GTEx Portal V6	65
IL18R1	?	ENSG00000115604	frontal cortex	1.25×10-9 (p-value)	GTEx Portal V6	63
IL1RL1	?	ENSG00000115602	frontal cortex	3.56×10-11 (p-value)	GTEx Portal V6	65
IL18R1	?	ENSG00000115604	hippocampus	4.51×10-13 (p-value)	GTEx Portal V6	62
IL1RL1	?	ENSG00000115602	hippocampus	1.59×10-10 (p-value)	GTEx Portal V6	58

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000494017	1	ENST00000410040	IL18R1
		ENST00000404917, ENST00000233954	IL1RL1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000257175	1	ENCP00000027297	MAP4K4
ENCE00000257176	2	ENCP00000027297	MAP4K4
ENCE00000257360	2	ENCP00000027305	IL1R1
ENCE00000257274	1	ENCP00000027300	IL1R2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000677139 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	PU1, Rad21, CTCF
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	CTCF
ENSR00001544951 (promoter flanking region)	8	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	Cjun, DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, DNase1
ENSR00001655317 (enhancer)	6	monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
		A549	H3K27me3
ENSR00001655318	1	HSMMtube	H3K27me3, DNase1

(enhancer)		A549	H3K27me3
		blood (DND-41)	H3K27me3
		muscle (HSMM)	DNase1
ENSR00001655321 (enhancer)	3	monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K4me1, H3K27ac
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001655322 (CTCF binding site)	1	HSMMtube	H3K27me3
		blood (K562)	PolII, Ini1, Brg1, Gata2, DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	DNase1, H3K4me2, H3K4me1, H3K27ac
		liver (HepG2)	H3K27me3
		A549	H3K4me2
		skin (NHEK)	DNase1, H3K27ac, H3K4me2
ENSR00000595653 (promoter)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	PolII, Ini1, Brg1, Gata2, Cmyc, DNase1
		skin (NHDF-AD)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	Cjun, DNase1, H3K4me2, H3K4me1, H3K27ac, PolII, CTCF
		liver (HepG2)	H3K27me3, DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me3, H3K4me2
		skin (NHEK)	DNase1, H3K27ac, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL18R1	upstream gene variant	903	ENST00000410040	?	ENSP00000386663	7
IL1RL1	upstream gene variant	876	ENST00000404917	NM_001282408.1	ENSP00000384822	8
IL1RL1	upstream gene variant	681	ENST00000427077	?	ENSP00000391120	8
IL1RL1	upstream gene variant	876	ENST00000233954	NM_016232.4	ENSP00000233954	8
IL1RL1	upstream gene variant	541	ENST00000311734	NM_003856.3	ENSP00000310371	8
IL1RL1	upstream gene variant	651	ENST00000409584	?	ENSP00000386618	8
IL1RL1	upstream gene variant	903	ENST00000473175	?	?	7
IL1RL1	upstream gene variant	643	ENST00000463990	?	?	8
IL1RL1	upstream gene variant	942	ENST00000447231	?	ENSP00000409437	7
IL1RL1	upstream gene variant	674	ENST00000482701	?	?	8

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL18R1	ENST00000410040	?	ENSP00000386663	58
IL1RL1	ENST00000427077	?	ENSP00000391120	1
IL1RL1	ENST00000404917	NM_001282408.1	ENSP00000384822	58
IL1RL1	ENST00000311734	NM_003856.3	ENSP00000310371	1
IL1RL1	ENST00000473175	?	?	58
IL1RL1	ENST00000409584	?	ENSP00000386618	1
IL1RL1	ENST00000233954	NM_016232.4	ENSP00000233954	58

IL1RL1	ENST00000463990	?	?	1
IL1RL1	ENST00000447231	?	ENSP00000409437	58
IL1RL1	ENST00000482701	?	?	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RL1	ENST00000447231	?	ENSP00000409437	1

Locus 45 (rs12151621)

SNiPAcard

Block annotations

Block info

genomic range	chr2:85,933,003-85,938,407
block size	5,405 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.852 [-2.998 – 0.359]	gene(s) hit or close-by	–
phastCons	μ = 0.020 [0 – 0.116]	eQTL gene(s)	AC012454.4, ATOH8, GGCX, GNLY, MLH3, VAMP8
GERP++	μ = -1.305 [-5.72 – 2.01]	potentially regulated gene(s)	AC105053.3, CAPG, IMMT, MAT2A, MRPL35, POLR1A, PTCD3, SFTPB, SH2D6, TGOLN2, TMEM150A, USP39, VAMP5, VAMP8
CADD score	μ = 2.968 [0.191 – 6.026]	disease gene(s)	GGCX, MLH3, SFTPB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
GGCX	VITAMIN K-DEPENDENT CLOTTING FACTORS, COMBINED DEFICIENCY OF, [...]	OMIM	MIM:277450
GGCX	PSEUDOXANTHOMA ELASTICUM-LIKE DISORDER WITH MULTIPLE COAGULATION [...]	OMIM	MIM:610842
MLH3	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 7	OMIM	MIM:614385
MLH3	COLORECTAL CANCER	OMIM	MIM:114500
SFTPB	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 1	OMIM	MIM:265120
SFTPB	RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	OMIM	MIM:267450
GGCX	Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency	OrphaNet	OrphaNet:91135
GGCX	Hereditary combined deficiency of vitamin K-dependent clotting factors	OrphaNet	OrphaNet:98434
MLH3	Hereditary nonpolyposis colon cancer	OrphaNet	OrphaNet:144
SFTPB	Congenital pulmonary alveolar proteinosis	OrphaNet	OrphaNet:264675
SFTPB	Infant acute respiratory distress syndrome	OrphaNet	OrphaNet:70587
SFTPB	Neonatal acute respiratory distress with surfactant metabolism deficiency	OrphaNet	OrphaNet:217563

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GNLY	?	ENSG00000115523	pancreas	8.36×10^{-9} (p-value)	GTEx Portal V6	6
GNLY	ENST00000489214	ILMN_1708779	adipocyte	6.83×10^{-14} (p-value)	MuTHER consortium	5
GNLY	ENST00000409696					
GNLY	ENST00000534351					
GNLY	ENST00000489980					
GNLY	ENST00000533041					
GNLY	ENST00000482900					
GNLY	ENST00000470974					
GNLY	ENST00000464298					
GNLY	ENST00000263863					
GNLY	ENST00000524600					
GNLY	?	ENSG00000115523	muscularis mucosae	1.78×10^{-14} (p-value)	GTEx Portal V6	6

GNLY	?	ENSG00000115523	lung	1.37×10-11 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	atrial appendage	6.52×10-7 (p-value)	GTEx Portal V6	5
ATOH8	?	ENSG00000168874	blood	5.22×10-20 (p-value)	GTEx Portal V6	6
AC012454.4	?	ENSG00000203363	blood	1.46×10-12 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	blood	9.39×10-24 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	thyroid	6.13×10-11 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	skeletal muscle	4.22×10-6 (p-value)	GTEx Portal V6	5
GNLY	?	ENSG00000115523	prostate	9.77×10-9 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	transverse colon	1.42×10-8 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	sun exposed skin	7.34×10-7 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	aorta	2.64×10-8 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	subcutaneous adipocytes	5.31×10-12 (p-value)	GTEx Portal V6	6
?	?	ILMN_2256295	monocyte	7.03×10-22 (p-value)	Zeller et al.	2
?	?	ILMN_1790692	monocyte	1.26×10-24 (p-value)	Zeller et al.	2
ATOH8	ENST00000469442	ILMN_1654065	monocyte	1.32×10-157 (p-value)	Zeller et al.	2
ATOH8	ENST00000306279					
ATOH8	ENST00000489682					
GNLY	ENST00000489214	ILMN_1708779	monocyte	2.86×10-19 (p-value)	Zeller et al.	2
GNLY	ENST00000409696					
GNLY	ENST00000534351					
GNLY	ENST00000489980					
GNLY	ENST00000533041					
GNLY	ENST00000482900					
GNLY	ENST00000470974					
GNLY	ENST00000464298					
GNLY	ENST00000263863					
GNLY	ENST00000524600					
GNLY	?	ENSG00000115523	visceral adipocytes	1.30×10-7 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	stomach	9.04×10-10 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	spleen	3.83×10-8 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	sigmoid colon	3.04×10-6 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	coronary artery	3.38×10-6 (p-value)	GTEx Portal V6	5
GNLY	?	ENSG00000115523	tibial nerve	1.39×10-6 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	esophagus mucosa	2.80×10-16 (p-value)	GTEx Portal V6	6
GNLY	?	ENSG00000115523	gastroesophageal junction	2.82×10-7 (p-value)	GTEx Portal V6	6
GGCX	ENST00000233838	ILMN_1758232	blood	1.22×10-4 (p-value)	Westra et al.	1
VAMP8	ENST00000263864	ILMN_2190084	blood	9.93×10-6 (p-value)	Westra et al.	5
VAMP8	ENST00000432071					
VAMP8	ENST00000409760					

ATOH8	ENST00000306279	ILMN_1654065	monocyte	2.74×10-25 (p-value)	Fairfax et al.	1
ATOH8	ENST00000469442					
ATOH8	ENST00000489682					
GNLY	?	ENSG00000115523	terminal ileum	1.09×10-6 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
MLH3	ENST00000555144	ILMN_2282282	chr14	monocyte	2.52×10-18 (p-value)	Zeller et al.	2
MLH3	ENST00000555499						
MLH3	ENST00000553713						
MLH3	ENST00000556740						
MLH3	ENST00000355774						

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000253865	1	ENCP00000026860	USP39
		ENCP00000026814	TGOLN2
		ENCP00000026873	AC105053.3
		ENCP00000026830	CAPG
		ENCP00000026897	MRPL35
		ENCP00000026853	TMEM150A
		ENCP00000026896	IMMT
ENCE00000253866	2	ENCP00000026854	USP39
		ENCP00000026837	SH2D6
		ENCP00000026840	MAT2A
		ENCP00000026849	TMEM150A
		ENCP00000026888	PTCD3
		ENCP00000026853	TMEM150A
		ENCP00000026863	SFTPB
		ENCP00000026834	SH2D6
		ENCP00000026841	MAT2A
		ENCP00000026864	SFTPB
		ENCP00000026883	POLR1A
		ENCP00000026845	VAMP8
		ENCP00000026858	USP39
		ENCP00000026825	CAPG
		ENCP00000026814	TGOLN2
		ENCP00000026828	CAPG
		ENCP00000026830	CAPG
		ENCP00000026847	VAMP5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000593772 (promoter flanking region)	3	embryonic stem cell (H1ESC)	DNase1, H3K27me3, Rad21, CTCF
		HSMMtube	DNase1, CTCF
		blood (K562)	H3K27me3, DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, H3K27ac, DNase1
		liver (HepG2)	Rad21, CTCF, H3K27me3
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	H3K4me2, CTCF, H3K27ac, Rad21, H3K4me3, PU1, Srf, BATF, H3K4me1, DNase1

		nervous (NH-A)	CTCF, DNase1
		skin (NHEK)	DNase1, H3K4me1, CTCF
		NHLF	CTCF, DNase1
		Osteobl	CTCF, H3K27ac
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	DNase1, H3K4me1, CTCF
		cervix (HeLa-S3)	Jund, H3K27ac, H3K4me1, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K27ac, H3K9ac, H3K4me1, DNase1
		endothelium (HUVEC)	DNase1, CTCF
		A549	H3K4me3, CTCF, DNase1
ENSR00000676793 e! (CTCF binding site)	4	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	DNase1, CTCF, H3K27me3
		blood (K562)	H3K27me3, DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	Rad21, CTCF, H3K27me3
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	H3K9ac, DNase1, H3K4me1, CTCF, H3K27ac
		endothelium (HUVEC)	DNase1, CTCF
		A549	H3K4me3, CTCF, DNase1
ENSR00001544078 e! (enhancer)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac
		liver (HepG2)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K27me3

Locus 46 (rs217181)

SNiPAcard

Block annotations

Block info

genomic range	chr16:72,079,657-72,114,002
block size	34,346 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.184 [-0.937 – 0.205]	gene(s) hit or close-by	HPR, RP11-384M15.3, TXNL4B
phastCons	μ = 0.146 [0.003 – 0.375]	eQTL gene(s)	MARVELD3
GERP++	μ = -0.203 [-0.727 – 0.118]	potentially regulated gene(s)	–
CADD score	μ = 4.410 [0.091 – 11.43]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Metabolite levels	$<1.00\times10^{-36}$	GWAS Catalog	22916037	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MARVELD3	?	ENSG00000140832	sun exposed skin	6.74×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HPR	downstream gene variant	2858	ENST00000356967	?	ENSP00000349451	1
HPR	downstream gene variant	2857	ENST00000540303	NM_020995.3	ENSP00000441828	1
HPR	downstream gene variant	2857	ENST00000561690	?	ENSP00000462916	1
RP11-384M15.3	upstream gene variant	4754	ENST00000561827	?	?	1
TXNL4B	downstream gene variant	4756	ENST00000423037	NM_001142317.1	ENSP00000408130	1
TXNL4B	downstream gene variant	4750	ENST00000268483	NM_017853.2	ENSP00000268483	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HPR	ENST00000561690	?	ENSP00000462916	1
HPR	ENST00000566168	?	?	1
HPR	ENST00000356967	?	ENSP00000349451	1

HPR	ENST00000540303	NM_020995.3	ENSP00000441828	1
TXNL4B	ENST00000562153	?	ENSP00000454635	3

Locus 46 (sentinel SNP rs217181 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 47 (rs45602433)

SNiPAcard

Block annotations

Block info

genomic range	chr9:117,793,413-117,835,899
block size	42,487 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.608 [-0.255 – 1.75]	gene(s) hit or close-by	TNC
phastCons	μ = 0.057 [0 – 0.211]	eQTL gene(s)	–
GERP++	μ = 1.703 [-1.46 – 3.39]	potentially regulated gene(s)	–
CADD score	μ = 8.642 [3.163 – 18.2]	disease gene(s)	TNC

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TNC	DEAFNESS, AUTOSOMAL DOMINANT 56	OMIM	MIM:615629
TNC	Autosomal dominant nonsyndromic sensorineural deafness type DFNA	OrphaNet	OrphaNet:90635

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
TNC	missense variant	ENST00000341037	?	ENSP00000339553	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000537320	?	ENSP00000443478	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000542877	?	ENSP00000442242	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000535648	?	ENSP00000438152	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000350763	NM_002160.3	ENSP00000265131	H/R	cAt/cGt	?	?	1
TNC	missense variant	ENST00000423613	?	ENSP00000411406	H/R	cAt/cGt	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001474309 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3, DNase1
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
TNC	downstream gene variant	4968	ENST00000498724	?	?	1
TNC	downstream gene variant	3545	ENST00000473855	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TNC	ENST00000423613	?	ENSP00000411406	3
TNC	ENST00000460345	?	?	1
TNC	ENST00000542877	?	ENSP00000442242	3
TNC	ENST00000544972	?	ENSP00000445380	3
TNC	ENST00000476680	?	?	2
TNC	ENST00000537320	?	ENSP00000443478	3
TNC	ENST00000535648	?	ENSP00000438152	3
TNC	ENST00000341037	?	ENSP00000339553	3
TNC	ENST00000498724	?	?	2
TNC	ENST00000350763	NM_002160.3	ENSP00000265131	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TNC	ENST00000498724	?	1
TNC	ENST00000473855	?	1

Locus 48 (rs103294)

SNiPAcard

Block annotations

Block info

genomic range	chr19:54,792,761-54,809,343
block size	16,583 bp
variant count	25 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.377 [-4.251 – 0.44]	gene(s) hit or close-by	AC008984.5, AC008984.7, AC010518.2, AC010518.3
phastCons	μ = 0.006 [0 – 0.089]	eQTL gene(s)	AC010518.2, ITIH6, LILRA1, LILRB2, TSEN34
GERP++	μ = -0.450 [-1.87 – 0.713]	potentially regulated gene(s)	AC008753.6, AC008984.5, AC008984.5, AC008984.7, AC008984.7, AC009892.10, AC009892.10, AC009892.5, AC010518.3, AC010518.3, AC011515.2, AC011515.2, AC012314.19, AC012314.19, AC012314.2, KIR2DL1, KIR2DL1, KIR2DL1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LILRB2	ENST00000434421	ILMN_2312340	blood	1.93×10^{-5} (p-value)	Westra et al.	3
LILRB2	ENST00000493242					
LILRB2	ENST00000455108					
LILRB2	ENST00000314446					
LILRB2	ENST00000391748					
TSEN34	ENST00000396383	ILMN_1673111	blood	4.50×10^{-5} (p-value)	Westra et al.	2
TSEN34	ENST00000396388					
TSEN34	ENST00000302937					
TSEN34	ENST00000496583					
TSEN34	ENST00000396383	ILMN_2368292	blood	1.27×10^{-5} (p-value)	Westra et al.	4
TSEN34	ENST00000396388					
TSEN34	ENST00000302937					
TSEN34	ENST00000496583					
LILRB2	?	ENSG00000131042	sun exposed skin	1.90×10^{-8} (p-value)	GTEx Portal V6	22
LILRA1	ENST00000618665	ILMN_1661631	monocyte	2.83×10^{-289} (p-value)	Zeller et al.	2
LILRA1	ENST00000477255					
LILRA1	ENST00000495417					
AC010518.2	ENST00000615364					
?	?	ILMN_1786303	monocyte	4.40×10^{-265} (p-value)	Zeller et al.	2
LILRA1	ENST00000618665	ILMN_1661631	liver	4.88×10^{-15} (p-value)	Innocenti et al.	1
LILRA1	ENST00000477255					
AC010518.2	ENST00000615364					
LILRA1	ENST00000495417					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ITIH6	ENST00000218436	ILMN_1709177	chrX	monocyte	6.95×10^{-24} (p-value)	Zeller et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000233622	2	ENCP00000024752	LILRB3 LILRB3 LILRB3 LILRB3
ENCE00000233400	5	ENCP00000024802	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024761	AC008984.5 AC008984.5
		ENCP00000024807	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
		ENCP00000024769	LAIR1 LAIR1

Locus 48 (sentinel SNP rs103294 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

	LAIR1 LAIR1 LAIR1
ENCP00000024758	LILRB2 LILRB2 LILRB2 LILRB2 LILRB2
ENCP00000024753	LILRA6 LILRA6 LILRA6 LILRA6 LILRA6
ENCP00000024812	AC011515.2 AC011515.2
ENCP00000024705	MYADM
ENCP00000024817	KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3
ENCP00000024721	AC012314.2
ENCP00000024722	OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR
ENCP00000024703	NLRP12
ENCP00000024788	LENG8 LENG8 LENG8 LENG8
ENCP00000024803	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
ENCP00000024808	VN1R105P
ENCP00000024773	LAIR1 LAIR1 LAIR1 LAIR1 LAIR1
ENCP00000024763	LILRA5 AC008984.7 AC008984.7 LILRA5 LILRA5 LILRA5

	LILRA5
ENCP00000024772	LAIR1
	LAIR1
	LAIR1
	LAIR1
	LAIR1
ENCP00000024810	LILRB4
	LILRB4
	LILRB4
	LILRB4
	LILRB4
ENCP00000024759	AC010518.3
	AC010518.3
ENCP00000024771	LAIR1
	LAIR1
	LAIR1
	LAIR1
	LAIR1
ENCP00000024819	KIR2DL1
	KIR2DL1
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ENCP00000024799	LILRA1
	LILRA1
	LILRA1
	LILRA1
ENCP00000024751	AC012314.19
	AC012314.19
ENCP00000024811	LILRB4
	LILRB4
	LILRB4
	LILRB4
	LILRB4
ENCP00000024768	LAIR1
	LAIR1
	LAIR1
	LAIR1
	LAIR1
ENCP00000024764	LILRA5
	LILRA5
	LILRA5
	LILRA5
	LILRA5
ENCP00000024756	LILRB2
	LILRB2
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ENCP00000024818	KIR2DP1

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ENCP00000024816	KIR3DL3
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ENCP00000024709	MYADM
	AC008753.6
ENCP00000024760	LILRA3
	LILRA3
	LILRA3
	LILRA3
ENCP00000024806	AC009892.10
	AC009892.10
ENCP00000024798	LILRA2
	LILRA2
	LILRA2
	LILRA2
ENCP00000024797	LILRA2
	LILRA2
	LILRA2
	LILRA2
ENCP00000024800	AC009892.5
ENCP00000024770	LAIR1
	LAIR1
	LAIR1

			LAIR1
			LAIR1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348830 (open chromatin region)	11	blood (K562)	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC008984.5	downstream gene variant	541	ENST00000514037	?	?	4
AC008984.7	downstream gene variant	3024	ENST00000426257	?	?	3
AC010518.2	downstream gene variant, upstream gene variant	396	ENST00000615364	?	?	8
AC010518.3	downstream gene variant, upstream gene variant	54	ENST00000417373	?	?	23

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC008984.5	ENST00000514037	?	?	2

Locus 49 (rs2691273)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,596,136-51,600,921
block size	4,786 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.365 [-0.94 – 0.209]	gene(s) hit or close-by	CTU1
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	CTU1
GERP++	μ = -4.200 [-4.53 – -3.87]	potentially regulated gene(s)	–
CADD score	μ = 1.494 [1.32 – 1.667]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CTU1	?	ENSG00000142544	thyroid	1.75×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000143070 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K36me3
		blood (K562)	CTCFL, Rad21, CTCF, H3K36me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K36me3, CTCF
		lung (IMR90)	H3K36me3, CTCF
		blood (GM12878)	DNase1, Yy1, CTCF, H3K36me3
		nervous (NH-A)	CTCF
		skin (NHEK)	H3K4me1, CTCF, H3K36me3, DNase1
		NHLF	CTCF
		Osteobl	CTCF, H3K36me3
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	DNase1, CTCF
		cervix (HeLa-S3)	H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, PolII
		A549	H3K36me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTU1	downstream gene variant	4727	ENST00000421832	NM_145232.3	ENSP00000390011	1

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

CTU1	ENST00000421832	NM_145232.3	ENSP00000390011	1

Locus 50 (rs2425143)

SNiPAcard

Block annotations

Block info

genomic range	chr20:34,199,977-34,562,924
block size	362,948 bp
variant count	168 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.159 [-3.59 – 9.577]	gene(s) hit or close-by	CNBD2, COX7BP2, CPNE1, FER1L4, HIGD1AP16, NFS1, PHF20, RBM12, RBM39, RN7SKP271, RNU4-40P, RNU6-759P, RNU6-937P, ROMO1, RP1-309K20.6, RPF2P1, SCAND1, SPAG4, Y_RNA
phastCons	μ = 0.080 [0 – 1]	eQTL gene(s)	CEP250, CPNE1, EPB41L1, ERGIC3, FER1L4, LINC00657, MMP24-AS1, RBM39, RPL36P4, SCAND1
GERP++	μ = -0.096 [-5.54 – 5.32]	potentially regulated gene(s)	CEP250, CPNE1, EPB41L1, ERGIC3, FER1L4, GDF5, HMGB3P2, NFS1, PHF20, RBM12, RBM39, ROMO1, RP11-234K24.3, RP3-477O4.14, RP3-477O4.5, RP4-550H1.4, RP4-550H1.5, SCAND1, SPAG4
CADD score	μ = 4.564 [0.041 – 22.4]	disease gene(s)	EPB41L1, GDF5, NFS1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
EPB41L1	MENTAL RETARDATION, AUTOSOMAL DOMINANT 11	OMIM	MIM:614257
EPB41L1	Autosomal dominant non-syndromic intellectual disability	OrphaNet	OrphaNet:178469
GDF5	Acromesomelic dysplasia, Grebe type	OrphaNet	OrphaNet:2098
GDF5	Acromesomelic dysplasia, Hunter-Thomson type	OrphaNet	OrphaNet:968
GDF5	Proximal symphalangism	OrphaNet	OrphaNet:3250
GDF5	Multiple synostoses syndrome	OrphaNet	OrphaNet:3237
GDF5	Angel-shaped phalango-epiphyseal dysplasia	OrphaNet	OrphaNet:63442
GDF5	BRACHYDACTYLY TYPE C	OrphaNet	OrphaNet:93384
GDF5	Fibular aplasia - complex brachydactyly	OrphaNet	OrphaNet:2639
GDF5	BRACHYDACTYLY, TYPE A2	OrphaNet	OrphaNet:93396
GDF5	Brachydactyly, type A1	OrphaNet	OrphaNet:93388
NFS1	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	OrphaNet	OrphaNet:397593
GDF5	Du Pan syndrome	DECIPHER	MIM:228900
GDF5	multiple synostoses syndrome type 2 (SYNS2)	DECIPHER	MIM:610017
GDF5	brachydactyly type C (BDC)	DECIPHER	MIM:113100
GDF5	brachydactyly type A2 (BDA2)	DECIPHER	MIM:112600
GDF5	brachydactyly type A1 (BDA1)	DECIPHER	MIM:112500
GDF5	symphalangism proximal syndrome (SYM1)	DECIPHER	MIM:185800
GDF5	acromesomelic chondrodysplasia Hunter-Thompson type (AMDH)	DECIPHER	MIM:201250
GDF5	acromesomelic chondrodysplasia Grebe type (AMDG)	DECIPHER	MIM:200700

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CPNE1	missense variant	ENST00000352393	?	ENSP00000336945	R/P	cGt/cCt	?	?	1

CPNE1	missense variant	ENST00000317677	NM_003915.5	ENSP00000317257	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000401607	?	ENSP00000386067	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000397442	?	ENSP00000380584	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000397443	NM_152925.2, NM_001198863.1, NM_152926.2	ENSP00000380585	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000397445	NM_152927.2, NM_152928.2	ENSP00000380587	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000430570	?	ENSP00000390626	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000437340	?	ENSP00000415597	R/P	cGt/cCt	?	?	1
PHF20	missense variant	ENST00000439301	?	ENSP00000410373	M/V	Atg/Gtg	?	?	1
PHF20	missense variant	ENST00000374012	NM_016436.4	ENSP00000363124	M/V	Atg/Gtg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FER1L4	?	ENSG00000088340	atrial appendage	3.70×10-8 (p-value)	GTEx Portal V6	156
RPL36P4	?	ENSG00000224497	atrial appendage	1.15×10-10 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	atrial appendage	1.91×10-46 (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	transformed fibroblasts	1.75×10-8 (p-value)	GTEx Portal V6	165
RPL36P4	?	ENSG00000224497	transformed fibroblasts	8.96×10-18 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	transformed fibroblasts	4.53×10-66 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	breast	3.38×10-8 (p-value)	GTEx Portal V6	154
CPNE1	?	ENSG00000214078	breast	2.47×10-36 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	blood	2.32×10-11 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	blood	2.41×10-71 (p-value)	GTEx Portal V6	167
CEP250	?	ENSG00000126001	blood	4.47×10-7 (p-value)	GTEx Portal V6	1
FER1L4	?	ENSG00000088340	skeletal muscle	3.85×10-14 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	skeletal muscle	1.36×10-7 (p-value)	GTEx Portal V6	125
CPNE1	?	ENSG00000214078	skeletal muscle	2.47×10-46 (p-value)	GTEx Portal V6	167
CEP250	?	ENSG00000126001	skeletal muscle	7.89×10-6 (p-value)	GTEx Portal V6	24
CPNE1	?	ENSG00000214078	putamen	9.62×10-14 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	EBV lymphocytes	1.45×10-23 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	EBV lymphocytes	6.76×10-7 (p-value)	GTEx Portal V6	1
FER1L4	?	ENSG00000088340	unexposed skin	6.74×10-13 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	unexposed skin	4.43×10-14 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	unexposed skin	7.48×10-40 (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	sun exposed skin	1.04×10-11 (p-value)	GTEx Portal V6	165
RPL36P4	?	ENSG00000224497	sun exposed skin	9.86×10-16 (p-value)	GTEx Portal V6	167

CPNE1	?	ENSG00000214078	sun exposed skin	7.80×10-61 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	ovary	5.31×10-10 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	ovary	2.42×10-6 (p-value)	GTEx Portal V6	23
FER1L4	?	ENSG00000088340	left ventricle	9.16×10-11 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	left ventricle	2.47×10-8 (p-value)	GTEx Portal V6	165
CPNE1	?	ENSG00000214078	left ventricle	8.69×10-37 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	aorta	1.46×10-12 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	aorta	6.55×10-47 (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	aorta	7.36×10-6 (p-value)	GTEx Portal V6	12
FER1L4	?	ENSG00000088340	subcutaneous adipocytes	1.21×10-8 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	subcutaneous adipocytes	5.85×10-16 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	subcutaneous adipocytes	2.74×10-64 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	caudate basal ganglia	4.34×10-15 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	visceral adipocytes	1.31×10-12 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	visceral adipocytes	5.53×10-49 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	stomach	8.62×10-10 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	stomach	1.47×10-35 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	cortex	1.60×10-13 (p-value)	GTEx Portal V6	166
MMP24-AS1	?	ENSG00000126005	cortex	7.02×10-6 (p-value)	GTEx Portal V6	1
FER1L4	?	ENSG00000088340	tibial nerve	1.91×10-17 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	tibial nerve	2.79×10-12 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	tibial nerve	3.76×10-69 (p-value)	GTEx Portal V6	167
CEP250	?	ENSG00000126001	tibial nerve	4.38×10-6 (p-value)	GTEx Portal V6	56
RPL36P4	?	ENSG00000224497	pituitary	8.84×10-6 (p-value)	GTEx Portal V6	72
CPNE1	?	ENSG00000214078	pituitary	2.52×10-12 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	anterior cingulate cortex	5.59×10-15 (p-value)	GTEx Portal V6	166
ERGIC3	?	ENSG00000125991	anterior cingulate cortex	1.14×10-5 (p-value)	GTEx Portal V6	9
EPB41L1	?	ENSG00000088367	esophagus mucosa	1.24×10-7 (p-value)	GTEx Portal V6	161
RPL36P4	?	ENSG00000224497	esophagus mucosa	4.55×10-12 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	esophagus mucosa	1.19×10-71 (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	testis	2.75×10-8 (p-value)	GTEx Portal V6	111
RPL36P4	?	ENSG00000224497	testis	1.05×10-8 (p-value)	GTEx Portal V6	165
CPNE1	?	ENSG00000214078	testis	1.01×10-36 (p-value)	GTEx Portal V6	167
LINC00657	?	ENSG00000260032	testis	1.47×10-5 (p-value)	GTEx Portal V6	9
CPNE1	?	ENSG00000214078	hypothalamus	1.38×10-11 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	frontal cortex	6.74×10-14 (p-value)	GTEx Portal V6	166

CPNE1	?	ENSG00000214078	uterus	7.38×10-9 (p-value)	GTEx Portal V6	162
RPL36P4	?	ENSG00000224497	uterus	2.48×10-7 (p-value)	GTEx Portal V6	5
RPL36P4	?	ENSG00000224497	pancreas	9.94×10-12 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	pancreas	2.51×10-38 (p-value)	GTEx Portal V6	167
RBM39	?	ENSG00000131051	pancreas	6.07×10-6 (p-value)	GTEx Portal V6	5
FER1L4	?	ENSG00000088340	muscularis mucosae	1.73×10-13 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	muscularis mucosae	1.28×10-14 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	muscularis mucosae	5.11×10-43 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	lung	4.47×10-20 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	lung	2.11×10-71 (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	tibial artery	2.42×10-11 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	tibial artery	3.68×10-13 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	tibial artery	6.92×10-60 (p-value)	GTEx Portal V6	167
EPB41L1	?	ENSG00000088367	tibial artery	1.87×10-6 (p-value)	GTEx Portal V6	32
FER1L4	?	ENSG00000088340	thyroid	4.16×10-8 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	thyroid	2.29×10-16 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	thyroid	1.79×10-67 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	prostate	9.68×10-13 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	prostate	6.99×10-7 (p-value)	GTEx Portal V6	1
RPL36P4	?	ENSG00000224497	transverse colon	2.04×10-11 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	transverse colon	4.93×10-49 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	terminal ileum	1.26×10-6 (p-value)	GTEx Portal V6	52
CPNE1	?	ENSG00000214078	terminal ileum	4.62×10-19 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	nucleus accumbens	1.11×10-7 (p-value)	GTEx Portal V6	165
RPL36P4	?	ENSG00000224497	nucleus accumbens	8.13×10-6 (p-value)	GTEx Portal V6	70
RPL36P4	?	ENSG00000224497	spleen	1.70×10-6 (p-value)	GTEx Portal V6	113
CPNE1	?	ENSG00000214078	spleen	2.05×10-21 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	coronary artery	4.85×10-10 (p-value)	GTEx Portal V6	155
CPNE1	?	ENSG00000214078	coronary artery	7.97×10-23 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	sigmoid colon	7.42×10-7 (p-value)	GTEx Portal V6	101
CPNE1	?	ENSG00000214078	sigmoid colon	5.99×10-20 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	vagina	3.60×10-12 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	gastroesophageal junction	7.44×10-10 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	gastroesophageal junction	6.13×10-31 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	cerebellar hemisphere	5.12×10-8 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	cerebellar hemisphere	8.05×10-6 (p-value)	GTEx Portal V6	1

FER1L4	?	ENSG00000088340	adrenal gland	1.94×10-8 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	adrenal gland	2.04×10-10 (p-value)	GTEx Portal V6	158
CPNE1	?	ENSG00000214078	adrenal gland	5.78×10-34 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	cerebellum	2.33×10-9 (p-value)	GTEx Portal V6	166
CEP250	?	ENSG00000126001	cerebellum	1.89×10-5 (p-value)	GTEx Portal V6	21
CPNE1	?	ENSG00000214078	hippocampus	7.13×10-9 (p-value)	GTEx Portal V6	164
CPNE1	?	ENSG00000214078	liver	4.29×10-16 (p-value)	GTEx Portal V6	149
SCAND1	ENST00000373991	ILMN_1795317	monocyte	4.35×10-7 (p-value)	Fairfax et al.	13
SCAND1	ENST00000305978	ILMN_2372011	monocyte	2.41×10-6 (p-value)	Fairfax et al.	11
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
CPNE1	ENST00000397443	ILMN_1670841	monocyte	1.48×10-51 (p-value)	Fairfax et al.	13
CPNE1	ENST00000317677		b-cell	1.45×10-18 (p-value)	Fairfax et al.	13
CPNE1	ENST00000498056					
CPNE1	ENST00000397442					
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000498814					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
CPNE1	ENST00000397443	ILMN_2307025	monocyte	5.23×10-40 (p-value)	Fairfax et al.	13
CPNE1	ENST00000317677		b-cell	2.28×10-23 (p-value)	Fairfax et al.	13
CPNE1	ENST00000498056					
CPNE1	ENST00000397445					
CPNE1	ENST00000415920					
CPNE1	ENST00000437340					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
CPNE1	ENST00000397443	ILMN_1670841	skin	1.64×10-29 (p-value)	MuTHER consortium	52
CPNE1	ENST00000317677		blood	8.08×10-69 (p-value)	MuTHER consortium	52
CPNE1	ENST00000498056		adipocyte	4.98×10-34 (p-value)	MuTHER consortium	52
CPNE1	ENST00000397442					
CPNE1	ENST00000397445					
CPNE1	ENST00000415920					
CPNE1	ENST00000437340					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000498814					

CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
CPNE1	ENST00000397443	ILMN_2307025	skin	1.95×10-30 (p-value)	MuTHER consortium	52
CPNE1	ENST00000317677		blood	9.52×10-70 (p-value)	MuTHER consortium	52
CPNE1	ENST00000498056		adipocyte	8.91×10-43 (p-value)	MuTHER consortium	52
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000483495					
CPNE1	ENST00000473373					
SCAND1	ENST00000305978	ILMN_1794230	blood	1.14×10-5 (p-value)	MuTHER consortium	36
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
FER1L4	ENST00000615531	ILMN_2206722	blood	4.69×10-5 (p-value)	MuTHER consortium	1
FER1L4	ENST00000621044					
FER1L4	ENST00000616283					
FER1L4	ENST00000616711					
FER1L4	ENST00000611673					
FER1L4	ENST00000613061					
SCAND1	ENST00000305978	ILMN_2372011	blood	1.54×10-5 (p-value)	MuTHER consortium	32
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
CEP250	ENST00000397527	ILMN_1799011	blood	1.64×10-5 (p-value)	Westra et al.	17
CPNE1	?	ENSG00000214078	blood	3.42×10-21 (q-value)	SeeQTL DB (HapMap)	23
SCAND1	ENST00000373991	ILMN_1795317	monocyte	7.72×10-41 (p-value)	Zeller et al.	10
SCAND1	ENST00000305978	ILMN_1794230	monocyte	1.59×10-13 (p-value)	Zeller et al.	10
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
SCAND1	ENST00000305978	ILMN_2372011	monocyte	8.37×10-17 (p-value)	Zeller et al.	10
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
CPNE1	ENST00000397443	ILMN_1670841	monocyte	1.80×10-150 (p-value)	Zeller et al.	10
CPNE1	ENST00000317677					
CPNE1	ENST00000498056					
CPNE1	ENST00000397442					
CPNE1	ENST00000397445					
CPNE1	ENST00000415920					
CPNE1	ENST00000437340					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000498814					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					

CPNE1	ENST00000483495					
CPNE1	ENST00000397443	ILMN_2307025	monocyte	1.72×10-116 (p-value)	Zeller et al.	10
CPNE1	ENST00000317677					
CPNE1	ENST00000498056					
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000483495					
CPNE1	ENST00000473373					
CPNE1	ENST00000397443	ILMN_1670841	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
CPNE1	ENST00000317677					
CPNE1	ENST00000498056					
CPNE1	ENST00000397442					
CPNE1	ENST00000437340					
CPNE1	ENST00000397445					
CPNE1	ENST00000415920					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000498814					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
CPNE1	ENST00000397443	206918_s_at	blood	1.30×10-33 (p-value)	Dixon et al.	1
CPNE1	ENST00000317677					
CPNE1	ENST00000498056					
CPNE1	ENST00000397445					
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000447127	1	ENST00000397443, ENST00000430570, ENST00000439806, ENST00000397445	CPNE1
		ENST00000435161	RBM12

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000031405		RBM12

ENCP00000031420			
ENCP00000031435			

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000293219	1	ENCP00000031365	GDF5
		ENCP00000031373	RP3-477O4.14
		ENCP00000031370	CEP250
		ENCP00000031438	RP4-550H1.4
ENCE00000293515	1	ENCP00000031431	PHF20
ENCE00000293353	1	ENCP00000031397	SPAG4
		ENCP00000031415	RBM39
		ENCP00000031440	RP4-550H1.5
ENCE00000293517	1	ENCP00000031431	PHF20
ENCE00000293226	1	ENCP00000031446	RP11-234K24.3
		ENCP00000031448	EPB41L1
		ENCP00000031444	EPB41L1
		ENCP00000031410	NFS1 ROMO1
		ENCP00000031385	ERGIC3
		ENCP00000031367	CEP250
		ENCP00000031393	FER1L4
		ENCP00000031380	ERGIC3
		ENCP00000031378	RP3-477O4.5
		ENCP00000031406	NFS1
		ENCP00000031387	FER1L4
		ENCP00000031398	SPAG4
		ENCP00000031389	FER1L4
		ENCP00000031439	HMGB3P2
		ENCP00000031445	EPB41L1
		ENCP00000031392	FER1L4
		ENCP00000031390	FER1L4
		ENCP00000031443	EPB41L1
		ENCP00000031366	CEP250
		ENCP00000031381	ERGIC3
		ENCP00000031450	EPB41L1
		ENCP00000031427	PHF20
		ENCP00000031405	CPNE1 RBM12
ENCE00000293228	1	ENCP00000031439	HMGB3P2
		ENCP00000031446	RP11-234K24.3
		ENCP00000031392	FER1L4
		ENCP00000031393	FER1L4
		ENCP00000031390	FER1L4
		ENCP00000031366	CEP250
		ENCP00000031378	RP3-477O4.5
		ENCP00000031406	NFS1
		ENCP00000031387	FER1L4
		ENCP00000031450	EPB41L1
ENCE00000293397	1	ENCP00000031411	RBM39
ENCE00000293217	1	ENCP00000031364	GDF5
ENCE00000293297	1	ENCP00000031376	CEP250
ENCE00000293340	1	ENCP00000031394	FER1L4

	ENCP00000031395	FER1L4
	ENCP00000031442	EPB41L1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001665301 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, PolII, Rad21, TAF7, H3K9ac, CTCF, TAF1, H3K4me2, SP1, USF1, H3K4me3, DNase1
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	H2AZ, PolII, H3K4me2, CTCF, DNase1, H3K4me3, HEY1, H3K9ac, E2F6, Max, ZBTB7A, Rad21
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	H3K27ac, CTCF, H2AZ, DNase1
		liver (HepG2)	H3K36me3, H3K27me3, H3K4me3, Sin3Ak20, PolII, H3K79me2, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, DNase1
		blood (GM12878)	H3K9ac, CTCF, H3K4me2, EBF1, PolII, BATF, H2AZ, DNase1, H3K4me3, H3K27ac
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, CTCF
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K4me3, H3K4me2
		NHLF	DNase1, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, PolII, H3K4me3, Max, TAF1, H3K27ac, H3K4me2, H3K9ac, Ini1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	CTCF, DNase1, PolII, H3K4me1, H3K4me2, Max, H3K4me3
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
ENSR00000400325 (promoter)	2	embryonic stem cell (H1ESC)	H3K9ac, H3K4me3, H4K5ac, H3K36me3, CTCF, PolII, H4K20me1, H3K27ac, Yy1, TAF1, H3K4me2, DNase1
		HSMMtube	H2AZ, DNase1, H3K4me3, H3K79me2, H3K9ac, H4K20me1, H3K4me2, H3K27ac
		blood (K562)	Brg1, Egr1, H3K27ac, Max, H3K9ac, Cmyc, H3K79me2, HEY1, Yy1, H2AZ, TAF1, ELF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2
		liver (HepG2)	H3K27ac, Cmyc, H3K4me3, H3K27me3, DNase1, H3K9ac, H3K4me2, Yy1, H4K20me1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ
		blood (GM12878)	H3K79me2, TAF1, PolII, H2AZ, DNase1, Yy1, ELF1, H3K4me3, H3K4me2, H3K9ac, H3K36me3, H3K27ac
		lung (IMR90)	H3K9ac, H3K4ac, DNase1, H4K8ac, H4K20me1, H3K79me2, H3K27ac, H3K4me2, H4K5ac, H3K4me3
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac, H4K20me1
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H4K20me1, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K27ac, DNase1, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Nrf1, Ini1, H3K9ac, H3K4me2, H3K27ac, H3K36me3, PolII, H3K79me2, H3K4me3, TAF1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, Max, H3K4me2, H3K4me3, H3K9ac
		A549	H3K36me3, H3K27ac, H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00000400329 (promoter)	1	embryonic stem cell (H1ESC)	H3K4me3, H3K9ac, H3K36me3, CTCF, PolII, Rad21, Gabp, Sin3Ak20, H3K27ac, TAF1, H3K4me2, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K36me3, DNase1
		blood (K562)	H2AZ, ELF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, H3K27ac, Max, CTCF, H3K9ac, Gabp, H3K79me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K27ac, H3K4me2, H3K9ac
		muscle (HSMM)	H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3, H3K36me3, DNase1, PolII, Gabp
		blood (GM12878)	Tr4, H3K36me3, H3K9ac, H3K4me2, PolII, H2AZ, DNase1, CTCF, Gabp, ELF1, H3K4me3, H3K27ac
		lung (IMR90)	H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, CTCF, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, PolII, H3K9ac, H3K27ac, CTCF
		NHLF	H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K27ac, CTCF, H3K36me3, H3K4me2, H3K9ac, H3K4me3

		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF, PolII, Gabp, H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H3K9ac, DNase1, H3K4me1, H3K4me2, H3K27ac
		endothelium (HUVEC)	Max, H3K4me3, H3K4me2, H3K9ac, PolII, DNase1, H3K36me3
		A549	H3K36me3, H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00000400333 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001404744 (open chromatin region)	2	embryonic stem cell (H1ESC)	PolII, H3K36me3
		blood (K562)	H3K36me3, H3K79me2
		breast (HMEC)	H3K36me3
		muscle (HSMM)	H3K79me2, H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H4K20me1, H3K36me3
		blood (GM12878)	H3K36me3, H3K79me2
		A549	H3K36me3
		skin (NHEK)	H3K36me3, DNase1
ENSR00001665311 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII
		HSMMtube	H3K36me3
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K36me3, H3K79me2
		liver (HepG2)	H4K20me1, PolII, H3K79me2, H3K4me1, H3K36me3
		blood (GM12878)	PolII, H3K79me2, H3K4me3, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	H3K36me3, PolII
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00000400351 (enhancer)	1	lung (IMR90)	H3K27me3
ENSR00001448569 (promoter flanking region)	1	embryonic stem cell (H1ESC)	CTCF
		HSMMtube	DNase1
		liver (HepG2)	H3K27me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
ENSR00001665314 (enhancer)	1	liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
ENSR00000234313 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	DNase1, H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00001665315 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (DND-41)	H3K36me3
		blood (K562)	Egr1
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K36me3

		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001448575	1	NHLF	DNase1
(promoter flanking region)		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	DNase1
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K27ac, DNase1
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		cervix (HeLa-S3)	H3K36me3, DNase1, Jund, Cjun
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3, DNase1
		blood (GM12878)	H3K36me3
		nervous (NH-A)	DNase1
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000400367	1	NHLF	DNase1
(promoter flanking region)		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	DNase1, H3K36me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	FOXA1, H3K36me3
		blood (GM12878)	H3K36me3
		nervous (NH-A)	DNase1
		A549	DNase1, H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000400368	1	lung (IMR90)	H3K36me3
(TF binding site)			
ENSR00000400373	3	embryonic stem cell (H1ESC)	H3K36me3, DNase1
(promoter flanking region)		HSMMtube	H3K27ac, H3K36me3, DNase1
		blood (K562)	DNase1, H3K36me3
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	H3K36me3, H3K4me2, H3K9ac, H3K27ac, H3K4me1, DNase1
		NHLF	DNase1, H3K27ac
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27ac, H3K4me2, Jund, PolII, TAF1, Max, Cfos, DNase1, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	DNase1, Cjun
		A549	H3K36me3, DNase1, H3K4me2, H3K4me3
ENSR00000400377	1	embryonic stem cell (H1ESC)	DNase1, CTCF, H3K36me3, Rad21
(promoter flanking region)		HSMMtube	H3K27ac, DNase1, H3K9ac, CTCF
		blood (K562)	Rad21, CTCF, H3K36me3
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me3, CTCF
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, CTCF
		liver (HepG2)	DNase1, H3K36me3, Rad21, CTCF
		blood (GM12878)	Rad21, CTCF, H3K36me3
		lung (IMR90)	H3K27ac, H3K4me2, H3K18ac, H4K5ac, H4K91ac, CTCF, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	DNase1, CTCF, H3K9ac, H3K27ac, H3K4me1

		NHLF	DNase1, CTCF, H3K27ac
		Osteobl	H3K4me2, H3K27ac, CTCF, H3K36me3
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	Cjun, DNase1, H3K36me3, CTCF, PolII, Max, TAF1, H3K27ac, H3K4me2, Jund, H3K9ac
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, Cjun, H3K27ac
		A549	H3K4me3, H3K4me2, CTCF, DNase1, H3K27ac
ENSR00000400379 (open chromatin region)	3	lung (IMR90)	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	DNase1
ENSR00000234322 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K36me3, DNase1
		blood (K562)	H3K27ac, H3K9ac, Nrsf, FOSL1, DNase1, Ini1, PolII
		skin (NHDF-AD)	H3K4me1, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	DNase1, H3K36me3
		nervous (NH-A)	H3K36me3, DNase1
		NHLF	DNase1, H3K27ac
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun
		A549	H3K4me3, DNase1, H3K36me3
ENSR00000649302 (promoter)	2	embryonic stem cell (H1ESC)	PolII, Rad21, Gabp, TAF7, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	CTCF, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K36me3, DNase1
		blood (K562)	H3K79me2, HEY1, H2AZ, TAF1, ELF1, CTCFL, Cmyc, PolII, Gabp, H3K9ac, Rad21, Yy1, Ini1, Egr1, H3K27ac, Max, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	H3K9ac, DNase1, H3K4me3, CTCF, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me3, CTCF, H3K79me2, H2AZ, H3K27ac, H3K4me2, H3K9ac, DNase1, H3K36me3
		liver (HepG2)	Gabp, Yy1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K36me3, H3K27me3, DNase1
		lung (IMR90)	H3K36me3, H3K4me3, H3K9ac, CTCF, H3K4me2, H3K27ac, DNase1
		blood (GM12878)	TAF1, PolII, BCLAF1, H2AZ, H3K9ac, CTCF, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2
		nervous (NH-A)	DNase1, H3K4me2, H3K4me3, H3K27ac, H3K36me3, CTCF, H3K9ac
		skin (NHEK)	H3K4me2, H3K9ac, H3K4me3, CTCF, DNase1
		NHLF	DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K79me2, H3K4me3, Max, TAF1, H3K27ac, H3K4me2, H3K9ac, Ini1, Gabp, PolII, Cmyc, H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, PolII, CTCF, DNase1, Cmyc, H3K27ac, H3K9ac, H3K4me3, Max, H3K4me2
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
ENSR00001665325 (open chromatin region)	1	lung (IMR90)	H3K27me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
NFS1	1	ENST00000374085	hsa-miR-103a-3p
		ENST00000374092	hsa-miR-15a-5p
		ENST00000397425	hsa-miR-15b-5p
		ENST00000440385	hsa-miR-16-5p
		ENST00000480655	hsa-miR-195-5p
		ENST00000498084	hsa-miR-34a-5p
			hsa-miR-424-5p
			hsa-miR-449a
			hsa-miR-497-5p
			hsa-miR-503-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CNBD2	upstream gene variant	651	ENST00000349339	NM_080834.2	ENSP00000340954	1
CNBD2	upstream gene variant, downstream gene variant	36	ENST00000489667	NM_001304367.1	ENSP00000480804	3
CNBD2	upstream gene variant	667	ENST00000463258	?	ENSP00000476014	1
CNBD2	upstream gene variant	121	ENST00000622112	?	ENSP00000479333	3
CNBD2	upstream gene variant	667	ENST00000538900	NM_001207076.1	ENSP00000442729	1
CNBD2	upstream gene variant	634	ENST00000373973	?	ENSP00000363084	1
CNBD2	upstream gene variant, downstream gene variant	264	ENST00000614708	?	ENSP00000478833	5
COX7BP2	downstream gene variant, upstream gene variant	413	ENST00000437976	?	?	3
CPNE1	downstream gene variant, upstream gene variant	97	ENST00000414711	?	ENSP00000409955	5
CPNE1	downstream gene variant, upstream gene variant	73	ENST00000483359	?	?	5
CPNE1	downstream gene variant, upstream gene variant	2289	ENST00000498056	?	?	4
CPNE1	downstream gene variant, upstream gene variant	1639	ENST00000317677	NM_003915.5	ENSP00000317257	5
CPNE1	downstream gene variant, upstream gene variant	51	ENST00000412056	?	ENSP00000416962	5
CPNE1	downstream gene variant, upstream gene variant	59	ENST00000397442	?	ENSP00000380584	5
CPNE1	downstream gene variant, upstream gene variant	778	ENST00000437340	?	ENSP00000415597	4
CPNE1	downstream gene variant, upstream gene variant	295	ENST00000415920	?	ENSP00000396069	4
CPNE1	downstream gene variant, upstream gene variant	52	ENST00000439669	?	ENSP00000409305	5
CPNE1	downstream gene variant, upstream gene variant	57	ENST00000401607	?	ENSP00000386067	5
CPNE1	downstream gene variant, upstream gene variant	778	ENST00000397443	NM_152925.2, NM_001198863.1, NM_152926.2	ENSP00000380585	4
CPNE1	downstream gene variant, upstream gene variant	2	ENST00000430570	?	ENSP00000390626	5
CPNE1	downstream gene variant, upstream gene variant	778	ENST00000397445	NM_152927.2, NM_152928.2	ENSP00000380587	4
CPNE1	downstream gene variant, upstream gene variant	1639	ENST00000498814	?	?	4
CPNE1	downstream gene variant, upstream gene variant	1639	ENST00000462352	?	?	4
CPNE1	downstream gene variant, upstream gene variant	48	ENST00000352393	?	ENSP00000336945	5
CPNE1	downstream gene variant, upstream gene variant	679	ENST00000483495	?	?	3
CPNE1	downstream gene variant, upstream gene variant	1639	ENST00000473373	?	?	4
CPNE1	downstream gene variant, upstream gene variant	52	ENST00000440240	?	ENSP00000397638	5
CPNE1	downstream gene variant, upstream gene variant	34	ENST00000434795	?	ENSP00000409794	5
CPNE1	downstream gene variant, upstream gene variant	71	ENST00000416778	?	ENSP00000389662	5
CPNE1	downstream gene variant, upstream gene variant	34	ENST00000420363	?	ENSP00000401915	5
CPNE1	downstream gene variant, upstream gene variant	34	ENST00000475146	?	?	6
CPNE1	downstream gene variant, upstream gene variant	45	ENST00000458038	?	ENSP00000390141	5
CPNE1	downstream gene variant, upstream gene variant	376	ENST00000435747	?	ENSP00000412806	5
CPNE1	downstream gene variant, upstream gene variant	33	ENST00000437100	?	ENSP00000391483	5
CPNE1	downstream gene variant, upstream gene variant	2	ENST00000439806	?	ENSP00000387434	5
CPNE1	downstream gene variant, upstream gene variant	207	ENST00000486021	?	?	3

CPNE1	downstream gene variant, upstream gene variant	18	ENST00000414664 ?	ENSP00000404355	5
FER1L4	upstream gene variant	4526	ENST00000426117 ?	?	1
FER1L4	upstream gene variant	4561	ENST00000615531 ?	?	1
FER1L4	upstream gene variant	4493	ENST00000430275 ?	?	1
HIGD1AP16	upstream gene variant, downstream gene variant	560	ENST00000450986 ?	?	4
NFS1	upstream gene variant	2373	ENST00000421540 ?	?	2
NFS1	upstream gene variant	2424	ENST00000306750 ?	ENSP00000304740	2
NFS1	upstream gene variant	2539	ENST00000419569 ?	ENSP00000393482	2
NFS1	downstream gene variant	3230	ENST00000480655 ?	?	3
NFS1	downstream gene variant, upstream gene variant	109	ENST00000541387 NM_001198989.1	ENSP00000440897	5
NFS1	downstream gene variant, upstream gene variant	2539	ENST00000374085 ?	ENSP00000363198	5
NFS1	downstream gene variant	3230	ENST00000498084 ?	?	3
NFS1	downstream gene variant, upstream gene variant	2379	ENST00000397425 ?	ENSP00000380570	5
NFS1	downstream gene variant, upstream gene variant	2373	ENST00000440385 ?	ENSP00000392871	5
NFS1	downstream gene variant, upstream gene variant	2321	ENST00000374092 NM_021100.4	ENSP00000363205	5
NFS1	upstream gene variant	2376	ENST00000456462 ?	ENSP00000397572	2
NFS1	downstream gene variant	554	ENST00000471137 ?	?	2
NFS1	upstream gene variant	2376	ENST00000413203 ?	ENSP00000393669	2
NFS1	downstream gene variant, upstream gene variant	861	ENST00000489163 ?	?	3
PHF20	upstream gene variant, downstream gene variant	2735	ENST00000485604 ?	?	4
PHF20	upstream gene variant, downstream gene variant	492	ENST00000496305 ?	?	3
PHF20	upstream gene variant, downstream gene variant	716	ENST00000473943 ?	?	5
PHF20	upstream gene variant, downstream gene variant	445	ENST00000495338 ?	?	6
PHF20	upstream gene variant, downstream gene variant	2064	ENST00000449988 ?	ENSP00000391915	2
PHF20	upstream gene variant, downstream gene variant	361	ENST00000374000 ?	ENSP00000363112	6
PHF20	upstream gene variant, downstream gene variant	1547	ENST00000480940 ?	?	3
PHF20	upstream gene variant, downstream gene variant	2309	ENST00000420233 ?	ENSP00000413712	4
PHF20	upstream gene variant, downstream gene variant	422	ENST00000486408 ?	?	5
PHF20	upstream gene variant, downstream gene variant	361	ENST00000481202 ?	?	6
PHF20	upstream gene variant, downstream gene variant	309	ENST00000461122 ?	?	6
PHF20	upstream gene variant, downstream gene variant	361	ENST00000339089 ?	ENSP00000341900	6
PHF20	upstream gene variant, downstream gene variant	318	ENST00000374012 NM_016436.4	ENSP00000363124	9
PHF20	upstream gene variant, downstream gene variant	357	ENST00000452270 ?	ENSP00000404455	6
PHF20	upstream gene variant, downstream gene variant	368	ENST00000617560 ?	?	8
PHF20	upstream gene variant, downstream gene variant	347	ENST00000461405 ?	?	11
PHF20	upstream gene variant, downstream gene variant	606	ENST00000439301 ?	ENSP00000410373	5
RBM12	downstream gene variant, upstream gene variant	18	ENST00000374114 NM_001198838.1, NM_006047.5	ENSP00000363228	6

RBM12	downstream gene variant, upstream gene variant	46	ENST00000359646	NM_001198840.1	ENSP00000352668 5
RBM12	upstream gene variant	800	ENST00000435161	?	ENSP00000411692 2
RBM12	downstream gene variant, upstream gene variant	34	ENST00000374104	NM_152838.3	ENSP00000363217 5
RBM12	upstream gene variant	45	ENST00000424458	?	ENSP00000411036 3
RBM12	upstream gene variant	778	ENST00000431148	?	ENSP00000392642 2
RBM39	downstream gene variant, upstream gene variant	624	ENST00000433027	?	ENSP00000407466 6
RBM39	downstream gene variant, upstream gene variant	1250	ENST00000463004	?	? 3
RBM39	downstream gene variant, upstream gene variant	2018	ENST00000496183	?	? 4
RBM39	downstream gene variant, upstream gene variant	1967	ENST00000429968	?	ENSP00000395124 3
RBM39	downstream gene variant, upstream gene variant	2072	ENST00000470563	?	? 5
RBM39	downstream gene variant, upstream gene variant	1252	ENST00000403542	?	ENSP00000385977 3
RBM39	downstream gene variant, upstream gene variant	794	ENST00000498280	?	? 9
RBM39	downstream gene variant, upstream gene variant	1967	ENST00000495293	?	? 5
RBM39	downstream gene variant, upstream gene variant	1221	ENST00000493853	?	? 3
RBM39	downstream gene variant, upstream gene variant	525	ENST00000449489	?	ENSP00000390303 6
RBM39	downstream gene variant, upstream gene variant	2046	ENST00000465158	?	? 5
RBM39	downstream gene variant, upstream gene variant	903	ENST00000397370	?	ENSP00000380527 8
RBM39	downstream gene variant, upstream gene variant	2542	ENST00000374038	?	ENSP00000363150 3
RBM39	upstream gene variant	3407	ENST00000434927	?	ENSP00000393493 1
RBM39	downstream gene variant, upstream gene variant	1567	ENST00000455343	?	ENSP00000395563 7
RBM39	downstream gene variant	333	ENST00000481037	?	? 2
RBM39	downstream gene variant, upstream gene variant	1717	ENST00000416108	?	ENSP00000406900 7
RBM39	downstream gene variant, upstream gene variant	1252	ENST00000338163	?	ENSP00000344581 3
RBM39	downstream gene variant, upstream gene variant	274	ENST00000425184	?	ENSP00000389459 3
RBM39	downstream gene variant, upstream gene variant	97	ENST00000477334	?	? 6
RBM39	downstream gene variant, upstream gene variant	164	ENST00000426951	?	ENSP00000405916 6
RBM39	downstream gene variant, upstream gene variant	694	ENST00000490354	?	? 4
RBM39	downstream gene variant, upstream gene variant	48	ENST00000487604	?	? 6
RBM39	downstream gene variant, upstream gene variant	1363	ENST00000468086	?	? 3
RBM39	downstream gene variant, upstream gene variant	495	ENST00000442447	?	? 6
RBM39	downstream gene variant, upstream gene variant	2391	ENST00000471635	?	? 4
RBM39	downstream gene variant, upstream gene variant	823	ENST00000416529	?	ENSP00000407150 8
RBM39	downstream gene variant, upstream gene variant	87	ENST00000615771	?	? 6
RBM39	downstream gene variant, upstream gene variant	546	ENST00000453310	?	ENSP00000397443 8
RBM39	downstream gene variant, upstream gene variant	736	ENST00000475651	?	? 3
RBM39	downstream gene variant, upstream gene variant	1555	ENST00000463098	?	? 2
RBM39	downstream gene variant, upstream gene variant	1645	ENST00000448303	?	ENSP00000394824 9

RBM39	downstream gene variant, upstream gene variant	862	ENST00000461849 ?	?	3
RBM39	downstream gene variant, upstream gene variant	1967	ENST00000444878 ?	ENSP00000394602	3
RBM39	upstream gene variant	2903	ENST00000412738 ?	ENSP00000397917	1
RBM39	downstream gene variant, upstream gene variant	31	ENST00000492779 ?	?	6
RBM39	downstream gene variant, upstream gene variant	1236	ENST00000476806 ?	ENSP00000483837	5
RBM39	downstream gene variant, upstream gene variant	2018	ENST00000482563 ?	?	4
RBM39	downstream gene variant, upstream gene variant	1251	ENST00000361162 NM_004902.3	ENSP00000354437	3
RBM39	downstream gene variant, upstream gene variant	200	ENST00000461283 ?	?	3
RBM39	downstream gene variant, upstream gene variant	1539	ENST00000528062 NM_001242600.1, NM_001242599.1	ENSP00000436747	3
RBM39	downstream gene variant, upstream gene variant	1080	ENST00000490484 ?	?	6
RBM39	downstream gene variant, upstream gene variant	1129	ENST00000494274 ?	?	6
RBM39	downstream gene variant, upstream gene variant	1251	ENST00000253363 NM_184234.2	ENSP00000253363	3
RN7SKP271	upstream gene variant, downstream gene variant	1031	ENST00000410528 ?	?	4
RNU4-40P	upstream gene variant, downstream gene variant	3161	ENST00000364351 ?	?	5
RNU6-759P	downstream gene variant, upstream gene variant	170	ENST00000516740 ?	?	3
RNU6-937P	downstream gene variant	207	ENST00000384325 ?	?	2
ROMO1	upstream gene variant, downstream gene variant	748	ENST00000336695 ?	ENSP00000338293	4
ROMO1	upstream gene variant, downstream gene variant	744	ENST00000374077 ?	ENSP00000363190	4
ROMO1	upstream gene variant, downstream gene variant	744	ENST00000374078 NM_080748.2	ENSP00000363191	4
ROMO1	upstream gene variant, downstream gene variant	750	ENST00000374072 ?	ENSP00000363185	4
ROMO1	upstream gene variant, downstream gene variant	818	ENST00000397416 ?	ENSP00000380561	4
RP1-309K20.6	downstream gene variant	1589	ENST00000441563 ?	ENSP00000411232	2
RP1-309K20.6	downstream gene variant	1589	ENST00000454607 ?	ENSP00000395201	2
RP1-309K20.6	downstream gene variant	1589	ENST00000541176 ?	ENSP00000443983	2
RPF2P1	upstream gene variant, downstream gene variant	461	ENST00000424137 ?	?	11
SCAND1	downstream gene variant, upstream gene variant	577	ENST00000305978 NM_033630.2	ENSP00000301995	5
SCAND1	downstream gene variant, upstream gene variant	1943	ENST00000615116 NM_016558.3	ENSP00000481289	5
SCAND1	downstream gene variant, upstream gene variant	369	ENST00000373991 ?	ENSP00000363103	5
SPAG4	upstream gene variant, downstream gene variant	3180	ENST00000430878 ?	ENSP00000399231	3
SPAG4	upstream gene variant	1537	ENST00000462896 ?	?	2
SPAG4	upstream gene variant, downstream gene variant	2202	ENST00000498203 ?	?	3
SPAG4	upstream gene variant, downstream gene variant	1537	ENST00000454819 ?	ENSP00000396670	4
SPAG4	upstream gene variant, downstream gene variant	405	ENST00000374273 NM_003116.1	ENSP00000363391	4
SPAG4	upstream gene variant	1393	ENST00000463973 ?	?	2
SPAG4	upstream gene variant, downstream gene variant	1559	ENST00000468248 ?	?	4
Y_RNA	downstream gene variant, upstream gene variant	255	ENST00000362806 ?	?	4

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
SCAND1	ENST00000615116	NM_016558.3	ENSP00000481289	1
SCAND1	ENST00000305978	NM_033630.2	ENSP00000301995	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNBD2	ENST00000622112	?	ENSP00000479333	8
CNBD2	ENST00000373973	?	ENSP00000363084	3
CNBD2	ENST00000463258	?	ENSP00000476014	3
CNBD2	ENST00000538900	NM_001207076.1	ENSP00000442729	3
CNBD2	ENST00000349339	NM_080834.2	ENSP00000340954	3
CNBD2	ENST00000489667	NM_001304367.1	ENSP00000480804	1
CPNE1	ENST00000401607	?	ENSP00000386067	10
CPNE1	ENST00000434795	?	ENSP00000409794	9
CPNE1	ENST00000430570	?	ENSP00000390626	10
CPNE1	ENST00000352393	?	ENSP00000336945	10
CPNE1	ENST00000458038	?	ENSP00000390141	9
CPNE1	ENST00000483359	?	?	10
CPNE1	ENST00000414664	?	ENSP00000404355	9
CPNE1	ENST00000414711	?	ENSP00000409955	9
CPNE1	ENST00000415920	?	ENSP00000396069	1
CPNE1	ENST00000483495	?	?	1
CPNE1	ENST00000439669	?	ENSP00000409305	9
CPNE1	ENST00000435747	?	ENSP00000412806	9
CPNE1	ENST00000440240	?	ENSP00000397638	9
CPNE1	ENST00000420363	?	ENSP00000401915	9
CPNE1	ENST00000412056	?	ENSP00000416962	9
CPNE1	ENST00000439806	?	ENSP00000387434	9
CPNE1	ENST00000437340	?	ENSP00000415597	11
CPNE1	ENST00000437100	?	ENSP00000391483	9
CPNE1	ENST00000416778	?	ENSP00000389662	9
CPNE1	ENST00000397442	?	ENSP00000380584	10
CPNE1	ENST00000397445	NM_152927.2, NM_152928.2	ENSP00000380587	11
CPNE1	ENST00000475146	?	?	1
CPNE1	ENST00000397443	NM_152925.2, NM_001198863.1, NM_152926.2	ENSP00000380585	11
CPNE1	ENST00000317677	NM_003915.5	ENSP00000317257	6
NFS1	ENST00000419569	?	ENSP00000393482	2
NFS1	ENST00000480655	?	?	2
NFS1	ENST00000456462	?	ENSP00000397572	2
NFS1	ENST00000498084	?	?	2

NFS1	ENST00000489163	?	?	1
NFS1	ENST00000374092	NM_021100.4	ENSP00000363205	7
NFS1	ENST00000421540	?	?	2
NFS1	ENST00000374085	?	ENSP00000363198	7
NFS1	ENST00000541387	NM_001198989.1	ENSP00000440897	6
NFS1	ENST00000397425	?	ENSP00000380570	7
NFS1	ENST00000413203	?	ENSP00000393669	5
NFS1	ENST00000440385	?	ENSP00000392871	7
NFS1	ENST00000306750	?	ENSP00000304740	2
PHF20	ENST00000496305	?	?	6
PHF20	ENST00000486408	?	?	7
PHF20	ENST00000439301	?	ENSP00000410373	54
PHF20	ENST00000480940	?	?	6
PHF20	ENST00000495338	?	?	17
PHF20	ENST00000617560	?	?	1
PHF20	ENST00000461405	?	?	16
PHF20	ENST00000452270	?	ENSP00000404455	41
PHF20	ENST00000449988	?	ENSP00000391915	9
PHF20	ENST00000374000	?	ENSP00000363112	70
PHF20	ENST00000481202	?	?	70
PHF20	ENST00000461122	?	?	50
PHF20	ENST00000339089	?	ENSP00000341900	70
PHF20	ENST00000374012	NM_016436.4	ENSP00000363124	81
RBM12	ENST00000374114	NM_001198838.1, NM_006047.5	ENSP00000363228	4
RBM12	ENST00000374104	NM_152838.3	ENSP00000363217	4
RBM12	ENST00000435161	?	ENSP00000411692	5
RBM12	ENST00000424458	?	ENSP00000411036	4
RBM12	ENST00000431148	?	ENSP00000392642	5
RBM12	ENST00000359646	NM_001198840.1	ENSP00000352668	4
RBM39	ENST00000492779	?	?	17
RBM39	ENST00000338163	?	ENSP00000344581	22
RBM39	ENST00000468086	?	?	1
RBM39	ENST00000453310	?	ENSP00000397443	2
RBM39	ENST00000615771	?	?	5
RBM39	ENST00000448303	?	ENSP00000394824	13
RBM39	ENST00000494274	?	?	6
RBM39	ENST00000461283	?	?	21
RBM39	ENST00000490484	?	?	6
RBM39	ENST00000397370	?	ENSP00000380527	2
RBM39	ENST00000434927	?	ENSP00000393493	15
RBM39	ENST00000465158	?	?	3
RBM39	ENST00000412738	?	ENSP00000397917	15

RBM39	ENST00000429968	?	ENSP00000395124	22
RBM39	ENST00000374038	?	ENSP00000363150	15
RBM39	ENST00000361162	NM_004902.3	ENSP00000354437	22
RBM39	ENST00000481037	?	?	13
RBM39	ENST00000475651	?	?	4
RBM39	ENST00000425184	?	ENSP00000389459	13
RBM39	ENST00000416529	?	ENSP00000407150	2
RBM39	ENST00000455343	?	ENSP00000395563	9
RBM39	ENST00000416108	?	ENSP00000406900	9
RBM39	ENST00000253363	NM_184234.2	ENSP00000253363	22
RBM39	ENST00000426951	?	ENSP00000405916	7
RBM39	ENST00000403542	?	ENSP00000385977	22
RBM39	ENST00000477334	?	?	6
RBM39	ENST00000444878	?	ENSP00000394602	22
RBM39	ENST00000487604	?	?	5
RBM39	ENST00000463004	?	?	22
RBM39	ENST00000442447	?	?	6
RBM39	ENST00000496183	?	?	1
RBM39	ENST00000449489	?	ENSP00000390303	6
RBM39	ENST00000482563	?	?	1
RBM39	ENST00000433027	?	ENSP00000407466	6
RBM39	ENST00000528062	NM_001242600.1, NM_001242599.1	ENSP00000436747	22
RBM39	ENST00000471635	?	?	2
RBM39	ENST00000495293	?	?	3
RBM39	ENST00000476806	?	ENSP00000483837	3
RBM39	ENST00000470563	?	?	3
RP1-309K20.6	ENST00000441563	?	ENSP00000411232	13
RP1-309K20.6	ENST00000454607	?	ENSP00000395201	13
RP1-309K20.6	ENST00000541176	?	ENSP00000443983	13
SCAND1	ENST00000373991	?	ENSP00000363103	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNBD2	ENST00000614708	?	ENSP00000478833	1
NFS1	ENST00000374092	NM_021100.4	ENSP00000363205	1
NFS1	ENST00000374085	?	ENSP00000363198	1
NFS1	ENST00000397425	?	ENSP00000380570	1
NFS1	ENST00000440385	?	ENSP00000392871	1
PHF20	ENST00000374012	NM_016436.4	ENSP00000363124	1
RBM39	ENST00000426951	?	ENSP00000405916	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPNE1	ENST00000437340	?	ENSP00000415597	1
CPNE1	ENST00000397445	NM_152927.2, NM_152928.2	ENSP00000380587	1
CPNE1	ENST00000397443	NM_152925.2, NM_001198863.1, NM_152926.2	ENSP00000380585	1
RBM12	ENST00000435161	?	ENSP00000411692	1
RBM12	ENST00000431148	?	ENSP00000392642	1
SCAND1	ENST00000373991	?	ENSP00000363103	2
SCAND1	ENST00000615116	NM_016558.3	ENSP00000481289	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CPNE1	ENST00000483359	?	1
CPNE1	ENST00000483495	?	1
NFS1	ENST00000480655	?	1
NFS1	ENST00000498084	?	1
RBM39	ENST00000492779	?	2
RBM39	ENST00000481037	?	3
RBM39	ENST00000463098	?	1
RBM39	ENST00000477334	?	2
RBM39	ENST00000463004	?	3

Locus 51 (rs778986)

SNiPAcard

Block annotations

Block info

genomic range	chr19:5,844,537-5,844,649
block size	113 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 1.409 [-0.147 – 2.964]	gene(s) hit or close-by	AC024592.9, FUT3, FUT6
phastCons	μ = 0.498 [0 – 0.996]	eQTL gene(s)	–
GERP++	μ = 0.964 [-0.403 – 2.33]	potentially regulated gene(s)	–
CADD score	μ = 7.561 [3.992 – 11.13]	disease gene(s)	FUT6

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM970545	1
?	HGMD curated	HGMD	CM930259	1

Disease gene annotation

gene	trait	source DB	source entry/link
FUT6	FUCOSYLTRANSFERASE 6 DEFICIENCY	OMIM	MIM:613852

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FUT3	missense variant	ENST00000589620	NM_001097639.1	ENSP00000465804	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000589714	?	ENSP00000467081	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000589918	NM_001097640.1	ENSP00000468123	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000303225	NM_000149.3	ENSP00000305603	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000458379	NM_001097641.1	ENSP00000416443	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000587048	?	ENSP00000468515	R/W	Cgg/Tgg	tolerated	benign	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC024592.9	upstream gene variant	2829	ENST00000589276	?	?	2
FUT3	downstream gene variant	14	ENST00000587048	?	ENSP00000468515	1
FUT3	downstream gene variant	1927	ENST00000593144	?	?	2
FUT3	downstream gene variant	2169	ENST00000588539	?	?	2

FUT3	downstream gene variant	69	ENST00000585715	?	ENSP00000467633	2
FUT3	downstream gene variant	188	ENST00000587183	?	ENSP00000468795	2
FUT6	upstream gene variant	4809	ENST00000286955	NM_001040701.1	ENSP00000286955	2
FUT6	upstream gene variant	4804	ENST00000591079	?	?	2
FUT6	upstream gene variant	4834	ENST00000524754	?	ENSP00000431708	2
FUT6	upstream gene variant	4795	ENST00000318336	NM_000150.2	ENSP00000313398	2

Locus 52 (rs10935480)

SNiPAcard

Block annotations

Block info

genomic range	chr3:98,402,969-98,434,493
block size	31,525 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.517 [-1.412 – 0.415]	gene(s) hit or close-by	ST3GAL6-AS1
phastCons	μ = 0.007 [0 – 0.029]	eQTL gene(s)	CPOX, DCBLD2, ST3GAL6, ST3GAL6-AS1
GERP++	μ = -0.989 [-5.16 – 0.372]	potentially regulated gene(s)	–
CADD score	μ = 3.151 [0.104 – 9.904]	disease gene(s)	CPOX

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CPOX	COPROPORPHYRIA, HEREDITARY	OMIM	MIM:121300
CPOX	Hereditary coproporphyria	OrphaNet	OrphaNet:79273

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ST3GAL6-AS1	splice donor variant	ENST00000475816	?	?					1
ST3GAL6-AS1	splice donor variant	ENST00000488132	?	?					1
ST3GAL6-AS1	splice donor variant	ENST00000461931	?	?					1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ST3GAL6-AS1	?	ENSG00000239445	pancreas	6.23×10^{-8} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	muscularis mucosae	2.39×10^{-17} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	lung	2.20×10^{-10} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	atrial appendage	3.43×10^{-28} (p-value)	GTEx Portal V6	10
CPOX	?	ENSG00000080819	transformed fibroblasts	1.28×10^{-6} (p-value)	GTEx Portal V6	6
ST3GAL6	?	ENSG00000064225	transformed fibroblasts	8.59×10^{-14} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	transformed fibroblasts	3.69×10^{-12} (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	breast	4.14×10^{-14} (p-value)	GTEx Portal V6	10
ST3GAL6	?	ENSG00000064225	tibial artery	2.47×10^{-8} (p-value)	GTEx Portal V6	9

ST3GAL6-AS1	?	ENSG00000239445	tibial artery	3.76×10-13 (p-value)	GTEx Portal V6	10
DCBLD2	?	ENSG00000057019	thyroid	9.35×10-7 (p-value)	GTEx Portal V6	10
ST3GAL6	?	ENSG00000064225	thyroid	3.05×10-7 (p-value)	GTEx Portal V6	6
ST3GAL6-AS1	?	ENSG00000239445	thyroid	7.65×10-21 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	skeletal muscle	9.61×10-18 (p-value)	GTEx Portal V6	10
ST3GAL6	?	ENSG00000064225	skeletal muscle	4.66×10-6 (p-value)	GTEx Portal V6	5
ST3GAL6-AS1	?	ENSG00000239445	putamen	8.30×10-10 (p-value)	GTEx Portal V6	9
ST3GAL6-AS1	?	ENSG00000239445	liver	1.01×10-7 (p-value)	GTEx Portal V6	9
ST3GAL6-AS1	?	ENSG00000239445	sun exposed skin	1.25×10-16 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	unexposed skin	2.64×10-9 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	aorta	2.81×10-14 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	left ventricle	8.49×10-28 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	subcutaneous adipocytes	1.15×10-16 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	caudate basal ganglia	3.17×10-11 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	visceral adipocytes	2.24×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	cortex	2.18×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	coronary artery	6.20×10-10 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	sigmoid colon	3.28×10-17 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	tibial nerve	1.74×10-22 (p-value)	GTEx Portal V6	10
ST3GAL6	?	ENSG00000064225	esophagus mucosa	1.18×10-15 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	esophagus mucosa	6.81×10-14 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	anterior cingulate cortex	6.57×10-13 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	gastroesophageal junction	3.08×10-11 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	adrenal gland	1.34×10-7 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	cerebellar hemisphere	1.14×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	testis	5.41×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	hypothalamus	2.14×10-13 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	cerebellum	1.45×10-12 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	frontal cortex	1.51×10-13 (p-value)	GTEx Portal V6	10
ST3GAL6-AS1	?	ENSG00000239445	EBV lymphocytes	2.90×10-6 (p-value)	GTEx Portal V6	8
ST3GAL6-AS1	?	ENSG00000239445	hippocampus	3.18×10-9 (p-value)	GTEx Portal V6	8
ST3GAL6-AS1	?	ENSG00000239445	stomach	2.88×10-7 (p-value)	GTEx Portal V6	5
ST3GAL6-AS1	?	ENSG00000239445	transverse colon	8.03×10-6 (p-value)	GTEx Portal V6	3
ST3GAL6-AS1	?	ENSG00000239445	nucleus accumbens	4.55×10-7 (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001676910 (enhancer)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ST3GAL6-AS1	downstream gene variant	617	ENST00000475816	?	?	3
ST3GAL6-AS1	downstream gene variant	615	ENST00000488132	?	?	3
ST3GAL6-AS1	downstream gene variant	1590	ENST00000461931	?	?	3

Locus 53 (rs3138036)

SNiPAcard

Block annotations

Block info

genomic range	chr17:32,647,544-32,660,149
block size	12,606 bp
variant count	23 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.302 [-3.87 – 2.066]	gene(s) hit or close-by	CCL8, RP11-521P1.1
phastCons	μ = 0.079 [0 – 0.941]	eQTL gene(s)	–
GERP++	μ = -0.332 [-5.69 – 3.09]	potentially regulated gene(s)	–
CADD score	μ = 4.854 [0.181 – 22.6]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CCL8	missense variant	ENST00000394620	NM_005623.2	ENSP00000378118	Q/K	Caa/Aaa	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001638184 (CTCF binding site)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		A549	H3K27me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL8	downstream gene variant	1316	ENST00000394620	NM_005623.2	ENSP00000378118	9
RP11-521P1.1	downstream gene variant, upstream gene variant	902	ENST00000582638	?	?	8

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL8	ENST00000394620	NM_005623.2	ENSP00000378118	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-521P1.1	ENST00000582638	?	1

Locus 54 (rs5167)

SNiPAcard

Block annotations

Block info

genomic range	chr19:45,440,529-45,448,465
block size	7,937 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.913 [-2.819 – 1.155]	**gene(s) hit or close-by**	APOC2, APOC4, APOC4-APOC2, CTB-129P6.11
phastCons	μ = 0.016 [0 – 0.043]	**eQTL gene(s)**	APOC1, APOE, CLPTM1
GERP++	μ = 0.266 [-0.38 – 2.07]	**potentially regulated gene(s)**	BCL3, CEACAM19
CADD score	μ = 2.814 [0.159 – 13.66]	**disease gene(s)**	APOE, APOC2

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM065970	1

Disease gene annotation

gene	trait	source DB	source entry/link
APOE	SEA-BLUE HISTIOCYTE DISEASE	OMIM	MIM:269600
APOE	ALZHEIMER DISEASE 2	OMIM	MIM:104310
APOE	HYPERCHOLESTEROLEMIA, FAMILIAL	OMIM	MIM:143890
APOE	LIPOPROTEIN GLOMERULOPATHY	OMIM	MIM:611771
APOE	APOLIPOPROTEIN E	OMIM	MIM:107741
APOC2	APOLIPOPROTEIN C-II DEFICIENCY	OMIM	MIM:207750
APOE	Hyperlipoproteinemia type 3	OrphaNet	OrphaNet:412
APOE	Sea-blue histiocytosis	OrphaNet	OrphaNet:158029
APOE	LIPOPROTEIN GLOMERULOPATHY	OrphaNet	OrphaNet:329481
APOC2	Familial apolipoprotein C-II deficiency	OrphaNet	OrphaNet:309020

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
APOC4	missense variant	ENST00000592954	NM_001646.2	ENSP00000468236	R/L	cGc/cTc	?	?	1
APOC4-APOC2	missense variant	ENST00000585685	?	ENSP00000467185	R/L	cGc/cTc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CLPTM1	?	ENSG00000104853	testis	1.50×10^{-10} (p-value)	GTEx Portal V6	9
APOE	?	ENSG00000130203	lung	3.76×10^{-8} (p-value)	GTEx Portal V6	8

APOC1	?	ENSG00000130208	lung	7.37×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000230231	1	ENCP00000024489	BCL3
		ENCP00000024485	CEACAM19

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
APOC2	upstream gene variant	827	ENST00000592257	?	ENSP00000477261	4
APOC2	upstream gene variant	848	ENST00000591597	?	ENSP00000476835	4
APOC2	upstream gene variant	3078	ENST00000585786	?	ENSP00000465001	1
APOC2	upstream gene variant	1995	ENST00000590360	?	ENSP00000466775	2
APOC4	upstream gene variant, downstream gene variant	192	ENST00000591600	?	ENSP00000466444	10
APOC4	upstream gene variant	849	ENST00000592954	NM_001646.2	ENSP00000468236	10
APOC4-APOC2	upstream gene variant	774	ENST00000252490	NM_000483.4	ENSP00000252490	4
APOC4-APOC2	upstream gene variant	867	ENST00000589057	?	ENSP00000468139	10
APOC4-APOC2	upstream gene variant	849	ENST00000585685	?	ENSP00000467185	10
CTB-129P6.11	downstream gene variant	4836	ENST00000591646	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOC4	ENST00000591600	?	ENSP00000466444	1
APOC4	ENST00000592954	NM_001646.2	ENSP00000468236	1
APOC4-APOC2	ENST00000585685	?	ENSP00000467185	1
APOC4-APOC2	ENST00000589057	?	ENSP00000468139	2

Locus 55 (rs376008)

SNiPAcard

Block annotations

Block info

genomic range	chr19:18,178,616-18,189,568
block size	10,953 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.087 [-4.269 – 0.224]	gene(s) hit or close-by	IL12RB1
phastCons	μ = 0.009 [0 – 0.064]	eQTL gene(s)	IL12RB1
GERP++	μ = -1.032 [-7.22 – 1.68]	potentially regulated gene(s)	–
CADD score	μ = 2.669 [0.001 – 10.62]	disease gene(s)	IL12RB1

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM984025	1
?	HGMD curated	HGMD	CM030471	1
?	HGMD curated	HGMD	CM030470	1

Disease gene annotation

gene	trait	source DB	source entry/link	
IL12RB1	IMMUNODEFICIENCY 30	OMIM	MIM:614891	OMIM
IL12RB1	Primary biliary cirrhosis	OrphaNet	OrphaNet:186	orphanet
IL12RB1	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency	OrphaNet	OrphaNet:319552	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IL12RB1	missense variant	ENST00000593993	NM_005535.1, NM_001290023.1	ENSP00000472165	3	3			3
IL12RB1	missense variant	ENST00000600835	NM_001290024.1	ENSP00000470788	3	3			3
IL12RB1	missense variant	ENST00000322153	NM_153701.1	ENSP00000314425	R/Q	cGg/cAg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL12RB1	?	ENSG00000096996	lung	3.11×10^{-9} (p-value)	GTEx Portal V6	21
IL12RB1	?	ENSG00000096996	blood	3.23×10^{-10} (p-value)	GTEx Portal V6	21
IL12RB1	?	ENSG00000096996	transverse colon	2.38×10^{-6} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000344606 (CTCF binding site)	2	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF
		HSMMtube	H3K27me3
		blood (K562)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	CTCF
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL12RB1	downstream gene variant	3476	ENST00000594176	?	ENSP00000473051	1
IL12RB1	downstream gene variant	2140	ENST00000430026	?	ENSP00000403103	5
IL12RB1	downstream gene variant	94	ENST00000322153	NM_153701.1	ENSP00000314425	7
IL12RB1	downstream gene variant	4354	ENST00000597416	?	?	1
IL12RB1	downstream gene variant	3429	ENST00000598019	?	ENSP00000468831	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
IL12RB1	ENST00000593993	NM_005535.1, NM_001290023.1	ENSP00000472165	P	ccT/ccC	1
IL12RB1	ENST00000600835	NM_001290024.1	ENSP00000470788	P	ccT/ccC	1
IL12RB1	ENST00000322153	NM_153701.1	ENSP00000314425	P	ccT/ccC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL12RB1	ENST00000593993	NM_005535.1, NM_001290023.1	ENSP00000472165	17
IL12RB1	ENST00000600835	NM_001290024.1	ENSP00000470788	17
IL12RB1	ENST00000322153	NM_153701.1	ENSP00000314425	12

Locus 56 (rs36032232)

SNiPAcard

Block annotations

Block info

genomic range	chr15:60,687,152-60,798,590
block size	111,439 bp
variant count	29 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.490 [-3.197 – 1.559]	**gene(s) hit or close-by**	ANXA2, ICE2, RORA, RORA-AS1, RP11-745A24.1, RP11-745A24.2, RP11-745A24.3
phastCons	μ = 0.100 [0 – 0.992]	**eQTL gene(s)**	ICE2
GERP++	μ = -1.141 [-7.15 – 3.05]	**potentially regulated gene(s)**	–
CADD score	μ = 3.973 [0.01 – 13.59]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ICE2	ENST00000261520	ILMN_1666610	blood	4.67×10^{-5} (p-value)	Westra et al.	3
ICE2	ENST00000558181					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001452708 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, Rad21, Jund, Nanog, Yy1, TAF1, H3K4me2, SP1, H3K4me3, PolII, H3K27me3, H3K36me3
		HSMMtube	H3K79me2, H3K4me1, H3K9ac, H3K4me2, DNase1, H3K36me3, H3K4me3, H3K27ac
		blood (K562)	Egr1, H3K27ac, Max, E2F6, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, Cjun, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF, H3K4me1, H3K36me3
		muscle (HSMM)	H3K9ac, H3K36me3, DNase1, H3K4me2, H3K27ac, H3K79me2, CTCF, H4K20me1, H3K4me3
		liver (HepG2)	DNase1, FOSL2, Jund, H3K4me3, PolII, H3K79me2, H3K4me1, H3K4me2, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K36me3
		lung (IMR90)	DNase1, H4K20me1, H3K79me2, H3K27ac, H3K18ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, H4K91ac, H3K56ac, CTCF
		blood (GM12878)	BATF, H2AZ, H3K4me1, DNase1, H3K79me2, H3K4me3, H3K4me2, H3K9ac, H3K36me3, PolII
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me3, H3K4me2, H3K4me1, H4K20me1, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, PolII, H3K9ac, H4K20me1, H3K27ac, H3K4me1, CTCF
		NHLF	DNase1, H3K4me3, H3K4me1, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K4me3, H3K79me2, PolII, Cmyc, CTCF, H3K36me3, Ini1, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me1, TAF1, Max, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, PolII, H3K27ac, H3K4me1, H3K9ac, H3K4me3, H3K4me2, Cjun
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00000409015 (promoter flanking region)	1	cervix (HeLa-S3)	Jund, DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me3
		skin (NHDF-AD)	DNase1
ENSR00000237536	2	embryonic stem cell (H1ESC)	DNase1, H3K36me3

(promoter flanking region)		HSMMtube	DNase1, H3K27ac
		blood (K562)	DNase1, H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K4me1
		lung (IMR90)	DNase1, H3K36me3
		blood (GM12878)	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3, H3K27ac, H3K4me1, DNase1
		NHLF	DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K36me3
		endothelium (HUVEC)	H3K4me2, Cjun
		A549	H3K36me3
ENSR00001628076 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		A549	H3K4me3, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANXA2	upstream gene variant	3802	ENST00000558986	?	ENSP00000453739	1
ANXA2	upstream gene variant	710	ENST00000557906	?	ENSP00000452895	2
ANXA2	upstream gene variant	732	ENST00000558998	?	ENSP00000453925	2
ANXA2	upstream gene variant	653	ENST00000451270	NM_004039.2	ENSP00000387545	2
ANXA2	upstream gene variant	3826	ENST00000559350	?	ENSP00000453663	1
ANXA2	upstream gene variant	710	ENST00000559370	?	ENSP00000453566	2
ANXA2	downstream gene variant, upstream gene variant	692	ENST00000560936	?	?	3
ANXA2	upstream gene variant	723	ENST00000557986	?	ENSP00000453742	2
ANXA2	upstream gene variant	3656	ENST00000560389	?	ENSP00000453579	1
ANXA2	upstream gene variant	710	ENST00000396024	NM_001136015.2	ENSP00000379342	2
ANXA2	upstream gene variant	458	ENST00000560367	?	ENSP00000453556	3
ANXA2	upstream gene variant	710	ENST00000561445	?	?	2
ANXA2	upstream gene variant	710	ENST00000559113	?	ENSP00000453869	2
ANXA2	downstream gene variant, upstream gene variant	708	ENST00000560495	?	?	3
ANXA2	upstream gene variant	718	ENST00000559467	?	ENSP00000453000	2
ANXA2	upstream gene variant	710	ENST00000561022	?	ENSP00000452876	2
ANXA2	upstream gene variant	708	ENST00000559559	?	?	2
ANXA2	upstream gene variant	710	ENST00000558558	?	ENSP00000452981	2
ANXA2	upstream gene variant	710	ENST00000421017	NM_001002857.1	ENSP00000411352	2
ANXA2	upstream gene variant	722	ENST00000559780	?	ENSP00000453770	2
ANXA2	upstream gene variant	663	ENST00000558985	?	ENSP00000453039	2
ANXA2	upstream gene variant	710	ENST00000557937	?	?	2
ANXA2	upstream gene variant	711	ENST00000332680	NM_001002858.2	ENSP00000346032	2
ANXA2	upstream gene variant	710	ENST00000560468	?	ENSP00000452858	2
ANXA2	upstream gene variant	695	ENST00000559725	?	ENSP00000453174	2
ANXA2	upstream gene variant	708	ENST00000558169	?	ENSP00000453945	2
ANXA2	upstream gene variant	728	ENST00000560165	?	ENSP00000452921	2

ANXA2	upstream gene variant	2875	ENST00000559818	?	ENSP00000453859	1
ANXA2	upstream gene variant	710	ENST00000560546	?	?	2
ANXA2	upstream gene variant	712	ENST00000559956	?	ENSP00000453694	2
ANXA2	upstream gene variant	710	ENST00000560466	?	ENSP00000453841	2
ANXA2	upstream gene variant	722	ENST00000558503	?	ENSP00000453356	2
ANXA2	upstream gene variant	710	ENST00000558132	?	ENSP00000454096	2
ANXA2	upstream gene variant	1172	ENST00000557904	?	ENSP00000452958	1
ICE2	downstream gene variant	1253	ENST00000558512	?	ENSP00000452714	1
ICE2	downstream gene variant	4302	ENST00000560668	?	ENSP00000453303	1
ICE2	downstream gene variant, upstream gene variant	1773	ENST00000561328	?	?	4
ICE2	downstream gene variant	603	ENST00000561124	?	?	2
ICE2	downstream gene variant	120	ENST00000558121	?	?	1
ICE2	downstream gene variant	4207	ENST00000561446	?	ENSP00000452713	1
ICE2	downstream gene variant, upstream gene variant	2229	ENST00000558451	?	?	3
ICE2	upstream gene variant	3508	ENST00000561144	?	?	1
ICE2	downstream gene variant	4306	ENST00000561087	?	?	1
ICE2	downstream gene variant	833	ENST00000560072	?	ENSP00000453041	3
ICE2	downstream gene variant	2897	ENST00000560895	?	ENSP00000453820	1
ICE2	downstream gene variant	1359	ENST00000560406	?	ENSP00000453783	3
ICE2	downstream gene variant	1375	ENST00000560520	?	ENSP00000452618	3
ICE2	downstream gene variant	819	ENST00000558654	?	?	3
RORA	downstream gene variant	222	ENST00000335670	NM_134261.2	ENSP00000335087	3
RORA	downstream gene variant	4903	ENST00000551975	?	ENSP00000449482	1
RORA-AS1	upstream gene variant	4017	ENST00000559902	?	?	1
RP11-745A24.1	downstream gene variant	4271	ENST00000475062	?	?	1
RP11-745A24.2	upstream gene variant	408	ENST00000477729	?	?	2
RP11-745A24.3	upstream gene variant	1950	ENST00000620477	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANXA2	ENST00000560936	?	?	1
ANXA2	ENST00000557906	?	ENSP00000452895	2
ANXA2	ENST00000559113	?	ENSP00000453869	2
ANXA2	ENST00000561445	?	?	2
ANXA2	ENST00000559467	?	ENSP00000453000	2
ANXA2	ENST00000560466	?	ENSP00000453841	2
ANXA2	ENST00000559559	?	?	2
ANXA2	ENST00000559370	?	ENSP00000453566	2
ANXA2	ENST00000560367	?	ENSP00000453556	1
ANXA2	ENST00000560468	?	ENSP00000452858	2

ANXA2	ENST00000421017	NM_001002857.1	ENSP00000411352	2
ANXA2	ENST00000558132	?	ENSP00000454096	2
ANXA2	ENST00000332680	NM_001002858.2	ENSP00000346032	2
ANXA2	ENST00000559956	?	ENSP00000453694	2
ANXA2	ENST00000558985	?	ENSP00000453039	2
ANXA2	ENST00000396024	NM_001136015.2	ENSP00000379342	2
ANXA2	ENST00000557986	?	ENSP00000453742	2
ANXA2	ENST00000557937	?	?	2
ANXA2	ENST00000559725	?	ENSP00000453174	2
ANXA2	ENST00000451270	NM_004039.2	ENSP00000387545	2
ANXA2	ENST00000561022	?	ENSP00000452876	2
ANXA2	ENST00000558169	?	ENSP00000453945	2
ANXA2	ENST00000558558	?	ENSP00000452981	2
ANXA2	ENST00000560165	?	ENSP00000452921	2
ANXA2	ENST00000558998	?	ENSP00000453925	2
ANXA2	ENST00000560546	?	?	2
ANXA2	ENST00000559780	?	ENSP00000453770	2
ANXA2	ENST00000557904	?	ENSP00000452958	4
ANXA2	ENST00000559818	?	ENSP00000453859	3
ANXA2	ENST00000560495	?	?	1
ANXA2	ENST00000558503	?	ENSP00000453356	2
ICE2	ENST00000560520	?	ENSP00000452618	1
ICE2	ENST00000558512	?	ENSP00000452714	7
ICE2	ENST00000261520	NM_001018089.2, NM_024611.5	ENSP00000261520	16
ICE2	ENST00000561087	?	?	7
ICE2	ENST00000558121	?	?	8
ICE2	ENST00000560072	?	ENSP00000453041	2
ICE2	ENST00000561124	?	?	8
ICE2	ENST00000561446	?	ENSP00000452713	7
ICE2	ENST00000561328	?	?	3
ICE2	ENST00000560895	?	ENSP00000453820	7
ICE2	ENST00000561114	NM_001276385.1	ENSP00000454162	8
ICE2	ENST00000558181	?	ENSP00000453593	16
ICE2	ENST00000560668	?	ENSP00000453303	7
ICE2	ENST00000560406	?	ENSP00000453783	1
ICE2	ENST00000558654	?	?	2
RORA	ENST00000335670	NM_134261.2	ENSP00000335087	1
RORA	ENST00000559587	?	?	1
RORA	ENST00000449337	NM_134262.2	ENSP00000402971	1
RORA	ENST00000309157	NM_002943.3	ENSP00000309753	1
RORA	ENST00000261523	NM_134260.2	ENSP00000261523	1
RORA-AS1	ENST00000501570	?	?	4

RORA-AS1	ENST00000501579	?	?	4
RORA-AS1	ENST00000558140	?	?	4
RORA-AS1	ENST00000559824	?	?	4
RORA-AS1	ENST00000558235	?	?	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ICE2	ENST00000261520	NM_001018089.2, NM_024611.5	ENSP00000261520	3
ICE2	ENST00000561114	NM_001276385.1	ENSP00000454162	1
ICE2	ENST00000558181	?	ENSP00000453593	3

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANXA2	ENST00000559818	?	ENSP00000453859	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ICE2	ENST00000558121	?	3
ICE2	ENST00000561328	?	2

Locus 57 (rs17284969)

SNiPAcard

Block annotations

Block info

genomic range	chr7:83,004,981-83,063,088
block size	58,108 bp
variant count	30 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.184 [-2.023 – 2.024]	gene(s) hit or close-by	AC079987.2, SEMA3E
phastCons	μ = 0.063 [0 – 1]	eQTL gene(s)	CHI3L2
GERP++	μ = -0.468 [-4.54 – 3.12]	potentially regulated gene(s)	SEMA3E
CADD score	μ = 3.790 [0.019 – 22.4]	disease gene(s)	SEMA3E

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
SEMA3E	CHARGE SYNDROME	OrphaNet	OrphaNet:138	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SEMA3E	missense variant	ENST00000427262	NM_001178129.1	ENSP00000405052	P/R	cCa/cGa	?	?	1
SEMA3E	missense variant	ENST00000307792	NM_012431.2	ENSP00000303212	P/R	cCa/cGa	?	?	1
SEMA3E	missense variant	ENST00000442159	?	ENSP00000412867	P/R	cCa/cGa	?	?	1

Direct effect on regulation

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CHI3L2	?	ENSG00000064886	chr1	blood	2.70×10^{-2} (q-value)	SeeQTL DB (HapMap)	2

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000798976	1	ENST00000427262, ENST00000307792	SEMA3E

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001715349 (enhancer)	1	HSMMtube	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC079987.2	upstream gene variant	4757	ENST00000420181	?	?	1
SEMA3E	downstream gene variant	3322	ENST00000442159	?	ENSP00000412867	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SEMA3E	ENST00000427262	NM_001178129.1	ENSP00000405052	29
SEMA3E	ENST00000307792	NM_012431.2	ENSP00000303212	29
SEMA3E	ENST00000442159	?	ENSP00000412867	5

Locus 58 (rs776905)

SNiPAcard

Block annotations

Block info

genomic range	chr13:113,780,041-113,781,942
block size	1,902 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.869 [-1.9 – 0.01]	gene(s) hit or close-by	F10, F10-AS1, KARSP2
phastCons	μ = 0.001 [0 – 0.002]	eQTL gene(s)	F10, F7
GERP++	μ = -0.050 [-1.29 – 0.635]	potentially regulated gene(s)	–
CADD score	μ = 3.276 [1.392 – 4.335]	disease gene(s)	F10, F7

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
F10	FACTOR X DEFICIENCY	OMIM	MIM:227600
F7	FACTOR VII DEFICIENCY	OMIM	MIM:227500
F10	Congenital factor X deficiency	OrphaNet	OrphaNet:328
F7	Congenital factor VII deficiency	OrphaNet	OrphaNet:327

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
F10	?	ENSG00000126218	lung	1.30×10-6 (p-value)	GTEx Portal V6	1
F10	?	ENSG00000126218	transformed fibroblasts	4.84×10-8 (p-value)	GTEx Portal V6	3
F7	?	ENSG00000057593	sun exposed skin	1.02×10-5 (p-value)	GTEx Portal V6	1
F7	?	ENSG00000057593	unexposed skin	1.63×10-8 (p-value)	GTEx Portal V6	3
F10	?	ENSG00000126218	aorta	7.31×10-8 (p-value)	GTEx Portal V6	3
F10	?	ENSG00000126218	tibial artery	4.98×10-8 (p-value)	GTEx Portal V6	3
F10	?	ENSG00000126218	thyroid	3.89×10-6 (p-value)	GTEx Portal V6	3

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
F10-AS1	downstream gene variant	527	ENST00000424635	?	?	3
KARSP2	upstream gene variant	2788	ENST00000415696	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F10	ENST00000483537	?	?	3
F10	ENST00000477269	?	?	3
F10	ENST00000410083	?	ENSP00000386320	3
F10	ENST00000409306	?	ENSP00000387092	3
F10	ENST00000375559	NM_000504.3	ENSP00000364709	3
F10	ENST00000375551	?	ENSP00000364701	3

Locus 60 (rs7959452)

SNiPAcard

Block annotations

Block info

genomic range	chr12:69,729,434-69,747,834
block size	18,401 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.116 [-1.544 – 0.956]	gene(s) hit or close-by	LYZ, RP11-1143G9.4, RP11-1143G9.5
phastCons	μ = 0.171 [0 – 0.922]	eQTL gene(s)	CPM, LYZ, RP11-1143G9.4, YEATS4
GERP++	μ = -1.254 [-4.79 – 3.4]	potentially regulated gene(s)	–
CADD score	μ = 4.343 [0.265 – 18.4]	disease gene(s)	LYZ

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
LYZ	AMYLOIDOSIS, FAMILIAL VISCERAL	OMIM	MIM:105200
LYZ	Familial renal amyloidosis due to lysozyme variant	OrphaNet	OrphaNet:93561

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
YEATS4	ENST00000247843	ILMN_1801387	blood	8.39×10-6 (p-value)	MuTHER consortium	5
YEATS4	ENST00000548020		adipocyte	1.48×10-9 (p-value)	MuTHER consortium	5
			skin	6.04×10-6 (p-value)	MuTHER consortium	5
YEATS4	?	ENSG00000127337	uterus	6.55×10-8 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	muscularis mucosae	9.16×10-13 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	muscularis mucosae	5.32×10-15 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	muscularis mucosae	4.85×10-10 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	lung	4.55×10-34 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	lung	3.98×10-11 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	lung	2.35×10-33 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	atrial appendage	7.74×10-13 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	atrial appendage	1.93×10-8 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	atrial appendage	2.72×10-10 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	tibial artery	4.52×10-25 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	tibial artery	3.94×10-17 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	tibial artery	1.47×10-23 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	breast	8.36×10-11 (p-value)	GTEx Portal V6	8

YEATS4	?	ENSG00000127337	breast	3.55×10-15 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	breast	2.27×10-10 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	blood	1.00×10-56 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	blood	1.24×10-15 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	blood	8.25×10-53 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	thyroid	1.81×10-16 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	thyroid	3.11×10-34 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	thyroid	3.44×10-15 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	skeletal muscle	3.51×10-13 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	skeletal muscle	2.36×10-31 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	skeletal muscle	3.17×10-10 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	transverse colon	5.76×10-11 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	transverse colon	5.70×10-9 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	transverse colon	1.73×10-9 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	EBV lymphocytes	1.19×10-12 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	unexposed skin	3.15×10-9 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	unexposed skin	1.21×10-19 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	unexposed skin	5.30×10-7 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	sun exposed skin	3.83×10-16 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	sun exposed skin	1.00×10-29 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	sun exposed skin	1.53×10-12 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	left ventricle	4.12×10-13 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	left ventricle	1.17×10-12 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	left ventricle	2.02×10-13 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	aorta	3.92×10-15 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	aorta	5.92×10-9 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	aorta	2.28×10-13 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	subcutaneous adipocytes	3.53×10-24 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	subcutaneous adipocytes	1.16×10-31 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	subcutaneous adipocytes	1.83×10-21 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	visceral adipocytes	2.86×10-15 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	visceral adipocytes	2.40×10-14 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	visceral adipocytes	1.56×10-13 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	stomach	3.45×10-7 (p-value)	GTEx Portal V6	4
YEATS4	?	ENSG00000127337	stomach	1.23×10-6 (p-value)	GTEx Portal V6	4
RP11-1143G9.4	?	ENSG00000257764	stomach	8.13×10-7 (p-value)	GTEx Portal V6	1

YEATS4	?	ENSG00000127337	nucleus accumbens	4.71×10-9 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	spleen	2.72×10-8 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	spleen	2.92×10-8 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	tibial nerve	5.77×10-21 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	tibial nerve	4.19×10-15 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	tibial nerve	4.78×10-18 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	coronary artery	2.17×10-6 (p-value)	GTEx Portal V6	7
YEATS4	?	ENSG00000127337	sigmoid colon	3.81×10-9 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	vagina	4.28×10-9 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	esophagus mucosa	3.72×10-8 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	esophagus mucosa	1.35×10-9 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	esophagus mucosa	4.97×10-7 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	adrenal gland	2.06×10-10 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	adrenal gland	1.12×10-8 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	adrenal gland	5.23×10-9 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	testis	6.72×10-7 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	testis	1.82×10-12 (p-value)	GTEx Portal V6	4
RP11-1143G9.4	?	ENSG00000257764	testis	5.10×10-6 (p-value)	GTEx Portal V6	1
YEATS4	?	ENSG00000127337	cerebellum	4.52×10-6 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	pancreas	6.60×10-6 (p-value)	GTEx Portal V6	1
LYZ	?	ENSG00000090382	prostate	2.08×10-6 (p-value)	GTEx Portal V6	1
YEATS4	?	ENSG00000127337	caudate basal ganglia	5.11×10-7 (p-value)	GTEx Portal V6	1
YEATS4	ENST00000247843	ILMN_1801387	monocyte	1.26×10-13 (p-value)	Fairfax et al.	3
YEATS4	ENST00000548020		b-cell	1.55×10-7 (p-value)	Fairfax et al.	3
LYZ	ENST00000261267	ILMN_2162972	monocyte	8.90×10-78 (p-value)	Fairfax et al.	3
CPM	ENST00000338356	ILMN_2299795	monocyte	1.58×10-8 (p-value)	Fairfax et al.	3
CPM	ENST00000551568					
?	?	ILMN_1815205	monocyte	2.31×10-17 (p-value)	Fairfax et al.	3
			b-cell	4.27×10-18 (p-value)		
YEATS4	?	ENSG00000127337	cortex	1.95×10-6 (p-value)	GTEx Portal V6	4
YEATS4	?	ENSG00000127337	cerebellar hemisphere	3.87×10-6 (p-value)	GTEx Portal V6	4

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001464668 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
ENSR00001612777	1	NHLF	H3K27me3

ENSR00001612777 (enhancer)	1	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me1
		lung (IMR90)	H3K27me3
		A549	H3K27me3
ENSR00001612779 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K4me2, H3K27ac, H3K4me3
		liver (HepG2)	H3K27ac, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LYZ	upstream gene variant, downstream gene variant	1297	ENST00000548839	?	ENSP00000449969	4
LYZ	upstream gene variant, downstream gene variant	559	ENST00000549690	?	ENSP00000449898	3
LYZ	upstream gene variant	1252	ENST00000261267	NM_000239.2	ENSP00000261267	2
RP11-1143G9.4	downstream gene variant	602	ENST00000548900	?	?	1
RP11-1143G9.5	upstream gene variant	3772	ENST00000613291	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LYZ	ENST00000549690	?	ENSP00000449898	1
LYZ	ENST00000261267	NM_000239.2	ENSP00000261267	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LYZ	ENST00000261267	NM_000239.2	ENSP00000261267	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-1143G9.4	ENST00000548900	?	1

Locus 61 (rs17094448)

SNiPAcard

Block annotations

Block info

genomic range	chr14:74,304,567-74,652,932
block size	348,366 bp
variant count	145 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.322 [-4.812 – 3.018]	gene(s) hit or close-by	AC005480.1, ALDH6A1, CCDC176, COQ6, ENTPD5, FAM161B, LIN52, PTGR2, RN7SL530P, RP5-1021I20.4, RP5-1021I20.5, RP5-1021I20.6, RP5-1021I20.7, RP5-1021I20.8, Y_RNA, Y_RNA, ZNF410
phastCons	μ = 0.075 [0 – 1]	eQTL gene(s)	ALDH6A1, CCDC176, ENTPD5, LIN52
GERP++	μ = -0.197 [-9.11 – 3.71]	potentially regulated gene(s)	ABCD4, PNMA1
CADD score	μ = 4.182 [0.01 – 21.5]	disease gene(s)	ALDH6A1, COQ6, ABCD4

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ALDH6A1	METHYLMALONATE SEMIALDEHYDE DEHYDROGENASE DEFICIENCY	OMIM	MIM:614105
COQ6	COENZYME Q10 DEFICIENCY, PRIMARY, 6	OMIM	MIM:614650
ABCD4	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblJ TYPE	OMIM	MIM:614857
ALDH6A1	Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency	OrphaNet	OrphaNet:289307
COQ6	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	OrphaNet	OrphaNet:280406
ABCD4	Methylmalonic acidemia with homocystinuria, type cblJ	OrphaNet	OrphaNet:369955
ABCD4	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblJ TYPE	DECIPHER	MIM:614857

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ENTPD5	ENST00000334696	ILMN_1745849	blood	2.53×10^{-6} (p-value)	Westra et al.	24
LIN52	ENST00000554076	ILMN_1797055	blood	7.71×10^{-7} (p-value)	Westra et al.	10
LIN52	ENST00000555028					
CCDC176	ENST00000394009	ILMN_1806456	blood	4.46×10^{-5} (p-value)	Westra et al.	7
ALDH6A1	ENST00000554501	ILMN_2096985	b-cell	4.66×10^{-4} (p-value)	Fairfax et al.	1
ALDH6A1	ENST00000350259					
ALDH6A1	ENST00000555126					
ALDH6A1	ENST00000553458					
CCDC176	?	ENSG00000119636	blood	9.70×10^{-8} (p-value)	GTEx Portal V6	21
ALDH6A1	ENST00000554501	ILMN_2096985	monocyte	3.63×10^{-14} (p-value)	Zeller et al.	2
ALDH6A1	ENST00000350259					
ALDH6A1	ENST00000555126					
ALDH6A1	ENST00000553458					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000171206	1	ENCP00000019795	PNMA1
		ENCP00000019810	ABCD4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001623807 (CTCF binding site)	1	cervix (HeLa-S3)	DNase1
		embryonic stem cell (H1ESC)	DNase1
		blood (K562)	Egr1
ENSR00000652291 (promoter)	1	embryonic stem cell (H1ESC)	H3K4me3, DNase1, H3K9ac, USF1, H3K27me3, CTCF, PolII, TAF7, H3K27ac, TAF1, H3K4me2
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K4me2, H3K9ac
		blood (K562)	H3K4me3, DNase1, H3K4me2, PolII, TAF1, H2AZ, HEY1, Cmyc, H3K9ac, PU1, Max, H3K27ac
		skin (NHDF-AD)	H3K9ac, DNase1, H3K4me3, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	DNase1, PolII, TAF1, USF1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3
		lung (IMR90)	DNase1, H4K8ac, H3K27ac, H3K18ac, H3K4me2, H4K5ac, H3K4me3, H3K9ac
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, ZBTB33, H3K4me3, H2AZ, DNase1, TAF1, PolII
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, DNase1, H3K4me3
		NHLF	H3K36me3, H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me3, H3K4me2, H3K27ac, H2AZ
		blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, H3K4me1
		breast (HMEC)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, PolII, H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, PolII, DNase1, Max, H3K4me3, H3K4me2, H3K9ac
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00000420786 (promoter)	2	embryonic stem cell (H1ESC)	H3K36me3, PolII, Egr1, Gabp, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K79me2, H3K9ac, H3K4me2, DNase1, H2AZ, H3K4me3, H3K27ac
		blood (K562)	H2AZ, H3K79me2, Cmyc, Gabp, H3K9ac, Max, H3K27ac, Egr1, TAF1, ELF1, PolII, H3K4me3, DNase1, H3K4me2
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, H2AZ
		liver (HepG2)	DNase1, Gabp, PolII, H3K79me2, TAF1, ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3
		lung (IMR90)	DNase1, H3K27ac, H3K18ac, H3K4me2, H3K4me3, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, Egr1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K27ac, H3K9ac
		Osteobl	H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K27ac, H3K4me2, H3K4me1, H3K36me3, H3K4me3, H3K9ac
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K4me3, Max, TAF1, H3K27ac, H3K4me2, H3K9ac, H3K79me2, Gabp, DNase1, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, H3K9ac, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00000420795 (promoter)	2	embryonic stem cell (H1ESC)	Yy1, TAF1, H3K4me2, H3K4me3, DNase1, H3K9ac, Sin3Ak20, H3K27me3, CTCF, PolII, Rad21
		HSMMtube	DNase1, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		blood (K562)	H2AZ, HEY1, Gabp, H3K9ac, H3K27ac, H3K4me3, DNase1, CTCF, H3K4me2, PolII
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K9ac, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, DNase1, H3K4me2, H3K4me1, Gabp, Yy1, PolII, TAF1, ELF1
		lung (IMR90)	CTCF, DNase1, H3K27ac, H3K4me2, H3K4me3, H3K9ac
		blood (GM12878)	DNase1, Yy1, ELF1, H3K4me3, CTCF, H3K9ac, H2AZ, PolII
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, CTCF
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2

		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, PolII, Gabp, H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K4me3, H3K4me2, DNase1
		endothelium (HUVEC)	Cmyc, DNase1, Max, H3K4me3, H3K4me2, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00000652293 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K4me3, PolII, H3K4me1, H3K4me2, H3K9ac, H3K27ac
		lung (IMR90)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00001623814 (enhancer)	2	liver (HepG2)	H3K4me1, H3K27ac
		lung (IMR90)	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00000241136 (enhancer)	1	liver (HepG2)	H3K4me1
		blood (K562)	DNase1
ENSR00000180920 (promoter)	4	embryonic stem cell (H1ESC)	H3K9ac, H3K27me3, Sin3Ak20, H3K4me2, H3K4me3, DNase1
		HSMMtube	DNase1, H2AZ, H3K4me3, H3K4me2, H3K9ac
		blood (K562)	H3K27ac, Max, H3K4me3, DNase1, H3K4me2, PolII, H2AZ, Cmyc, H3K9ac, Srf
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, H3K27ac
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	Srf, PolII, TAF1, H3K4me2, H3K9ac, H3K27ac, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K4me3, H3K9ac
		blood (GM12878)	DNase1, PolII, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	PolII, H3K4me3, H3K27ac, H3K4me2, H3K9ac, Ini1, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	Max, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, DNase1
		A549	H3K27ac, H3K9ac, H3K4me3, H3K4me2
ENSR00000420807 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac
		cervix (HeLa-S3)	DNase1, TAF1, H3K27ac
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		skin (NHEK)	H3K27ac
ENSR00000420808 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K4me1, H3K4me2, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun, H3K4me2
		liver (HepG2)	DNase1, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
ENSR00001623819 (CTCF binding site)	1	cervix (HeLa-S3)	H3K4me1, H3K4me2, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K4me2
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00001623829 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC005480.1	upstream gene variant, downstream gene variant	536	ENST00000624550	?	ENSP00000485283	20
ALDH6A1	downstream gene variant	267	ENST00000555126	?	ENSP00000452081	2
ALDH6A1	upstream gene variant	4119	ENST00000556852	?	?	1
ALDH6A1	downstream gene variant, upstream gene variant	2682	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	3
ALDH6A1	upstream gene variant	4146	ENST00000553814	?	?	1
ALDH6A1	downstream gene variant, upstream gene variant	247	ENST00000350259	NM_001278593.1	ENSP00000342564	3
ALDH6A1	downstream gene variant, upstream gene variant	4074	ENST00000554231	?	?	2
ALDH6A1	downstream gene variant, upstream gene variant	99	ENST00000554501	?	?	3
CCDC176	upstream gene variant, downstream gene variant	1365	ENST00000492247	?	?	4
CCDC176	upstream gene variant	133	ENST00000394009	NM_025057.2	ENSP00000377577	13
CCDC176	upstream gene variant	447	ENST00000492026	?	?	4
CCDC176	upstream gene variant	225	ENST00000463558	?	ENSP00000480689	12
CCDC176	upstream gene variant	223	ENST00000489323	?	?	12
CCDC176	upstream gene variant	130	ENST00000464394	?	ENSP00000451659	13
COQ6	upstream gene variant, downstream gene variant	308	ENST00000555392	?	?	5
COQ6	upstream gene variant, downstream gene variant	257	ENST00000553462	?	?	3
COQ6	upstream gene variant, downstream gene variant	1090	ENST00000554341	?	ENSP00000450736	2
COQ6	upstream gene variant, downstream gene variant	331	ENST00000334571	NM_182476.2	ENSP00000333946	3
COQ6	upstream gene variant, downstream gene variant	348	ENST00000554193	?	?	4
COQ6	upstream gene variant, downstream gene variant	337	ENST00000556300	?	?	3
COQ6	upstream gene variant, downstream gene variant	346	ENST00000554920	?	ENSP00000451562	3
COQ6	upstream gene variant, downstream gene variant	348	ENST00000553922	?	?	4
COQ6	upstream gene variant, downstream gene variant	348	ENST00000557205	?	?	3
COQ6	upstream gene variant, downstream gene variant	364	ENST00000238709	?	ENSP00000238709	3
COQ6	upstream gene variant, downstream gene variant	1076	ENST00000394026	NM_182480.2	ENSP00000377594	2
COQ6	upstream gene variant, downstream gene variant	1809	ENST00000553448	?	?	2
COQ6	upstream gene variant, downstream gene variant	1394	ENST00000556588	?	?	2
COQ6	upstream gene variant, downstream gene variant	339	ENST00000554153	?	ENSP00000451685	3
COQ6	upstream gene variant, downstream gene variant	348	ENST00000555552	?	?	4
COQ6	upstream gene variant, downstream gene variant	191	ENST00000557584	?	ENSP00000450511	3
COQ6	upstream gene variant, downstream gene variant	290	ENST00000629426	?	ENSP00000486650	3
COQ6	upstream gene variant, downstream gene variant	348	ENST00000554217	?	?	3
COQ6	upstream gene variant, downstream gene variant	346	ENST00000555196	?	ENSP00000451301	4
COQ6	upstream gene variant, downstream gene variant	1399	ENST00000556299	?	?	3
COQ6	upstream gene variant, downstream gene	562	ENST00000554320	?	ENSP00000451123	3

	variant					
COQ6	upstream gene variant, downstream gene variant	2347	ENST00000557780	?	?	2
COQ6	upstream gene variant, downstream gene variant	4160	ENST00000555511	?	?	2
ENTPD5	downstream gene variant, upstream gene variant	258	ENST00000555829	?	ENSP00000451499	4
ENTPD5	downstream gene variant, upstream gene variant	695	ENST00000554664	?	?	8
ENTPD5	downstream gene variant, upstream gene variant	432	ENST00000556242	?	ENSP00000451483	8
ENTPD5	downstream gene variant, upstream gene variant	352	ENST00000556108	?	?	9
ENTPD5	downstream gene variant, upstream gene variant	777	ENST00000557681	?	?	16
ENTPD5	downstream gene variant, upstream gene variant	549	ENST00000553284	?	ENSP00000451591	11
ENTPD5	downstream gene variant, upstream gene variant	762	ENST00000334696	NM_001249.2	ENSP00000335246	7
ENTPD5	downstream gene variant, upstream gene variant	317	ENST00000557325	?	ENSP00000451810	15
FAM161B	downstream gene variant, upstream gene variant	492	ENST00000286544	NM_152445.2	ENSP00000286544	6
FAM161B	downstream gene variant, upstream gene variant	174	ENST00000556794	?	ENSP00000450889	7
LIN52	downstream gene variant	46	ENST00000554289	?	?	2
PTGR2	upstream gene variant, downstream gene variant	413	ENST00000555661	?	ENSP00000452280	5
PTGR2	upstream gene variant, downstream gene variant	514	ENST00000267568	NM_152444.2	ENSP00000267568	6
PTGR2	upstream gene variant, downstream gene variant	1738	ENST00000553813	?	ENSP00000450824	4
PTGR2	upstream gene variant, downstream gene variant	438	ENST00000556219	?	?	5
PTGR2	upstream gene variant, downstream gene variant	411	ENST00000555228	NM_001146155.1, NM_001146154.1	ENSP00000450975	6
PTGR2	upstream gene variant, downstream gene variant	108	ENST00000555976	?	?	6
PTGR2	upstream gene variant, downstream gene variant	370	ENST00000553326	?	?	5
RN7SL530P	upstream gene variant	3981	ENST00000485097	?	?	1
RP5-1021I20.4	upstream gene variant, downstream gene variant	538	ENST00000556551	?	ENSP00000451484	4
RP5-1021I20.5	downstream gene variant	1666	ENST00000554009	?	?	1
RP5-1021I20.5	downstream gene variant, upstream gene variant	1565	ENST00000555916	?	?	3
RP5-1021I20.6	upstream gene variant, downstream gene variant	1270	ENST00000602874	?	?	5
RP5-1021I20.7	downstream gene variant, upstream gene variant	86	ENST00000604337	?	?	4
RP5-1021I20.8	downstream gene variant, upstream gene variant	762	ENST00000613926	?	?	8
Y_RNA	downstream gene variant, upstream gene variant	479	ENST00000362602	?	?	5
Y_RNA	downstream gene variant, upstream gene variant	361	ENST00000411368	?	?	5
ZNF410	upstream gene variant, downstream gene variant	488	ENST00000324593	NM_001242926.1	ENSP00000323293	6
ZNF410	upstream gene variant, downstream gene variant	554	ENST00000442160	NM_001242924.1	ENSP00000407130	6
ZNF410	upstream gene variant, downstream gene variant	604	ENST00000554316	?	ENSP00000451748	6
ZNF410	upstream gene variant	544	ENST00000541357	?	?	5
ZNF410	upstream gene variant, downstream gene variant	342	ENST00000555620	?	?	5
ZNF410	upstream gene variant	1994	ENST00000557679	?	?	4
ZNF410	upstream gene variant, downstream gene variant	1176	ENST00000557214	?	ENSP00000451620	4
	upstream gene variant, downstream gene	542	ENST00000557363	?	ENSP00000452199	9

ZNF410	variant					
ZNF410	upstream gene variant, downstream gene variant	415	ENST00000555602	?	?	8
ZNF410	upstream gene variant, downstream gene variant	300	ENST00000540593	NM_001242927.1	ENSP00000442228	6
ZNF410	upstream gene variant	326	ENST00000556179	?	ENSP00000450884	3
ZNF410	upstream gene variant, downstream gene variant	542	ENST00000412490	?	?	7
ZNF410	upstream gene variant, downstream gene variant	542	ENST00000556659	?	ENSP00000452023	8
ZNF410	upstream gene variant, downstream gene variant	554	ENST00000555044	NM_021188.2, NM_001242928.1	ENSP00000451763	6
ZNF410	upstream gene variant, downstream gene variant	2826	ENST00000615736	?	ENSP00000483073	6
ZNF410	upstream gene variant, downstream gene variant	1426	ENST00000554582	?	ENSP00000450650	8
ZNF410	upstream gene variant, downstream gene variant	1227	ENST00000334521	?	ENSP00000334170	3
ZNF410	upstream gene variant, downstream gene variant	554	ENST00000398139	?	ENSP00000381208	6
ZNF410	upstream gene variant, downstream gene variant	1157	ENST00000556797	?	ENSP00000451073	6
ZNF410	upstream gene variant, downstream gene variant	417	ENST00000555730	?	ENSP00000451807	9
ZNF410	upstream gene variant, downstream gene variant	2500	ENST00000554797	?	ENSP00000451116	5
ZNF410	upstream gene variant, downstream gene variant	741	ENST00000556160	?	ENSP00000451458	8
ZNF410	upstream gene variant, downstream gene variant	542	ENST00000557495	?	ENSP00000450569	8
ZNF410	upstream gene variant, downstream gene variant	491	ENST00000556396	?	ENSP00000451867	6

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
ENTPD5	ENST00000557325	?	ENSP00000451810	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ALDH6A1	ENST00000556852	?	?	2
ALDH6A1	ENST00000555126	?	ENSP00000452081	2
ALDH6A1	ENST00000554231	?	?	2
ALDH6A1	ENST00000553814	?	?	2
ALDH6A1	ENST00000350259	NM_001278593.1	ENSP00000342564	4
ALDH6A1	ENST00000554501	?	?	4
ALDH6A1	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	6
CCDC176	ENST00000464394	?	ENSP00000451659	12
CCDC176	ENST00000463558	?	ENSP00000480689	12
CCDC176	ENST00000394009	NM_025057.2	ENSP00000377577	18
CCDC176	ENST00000492026	?	?	8
CCDC176	ENST00000489323	?	?	12
COQ6	ENST00000555552	?	?	1
COQ6	ENST00000553462	?	?	2
COQ6	ENST00000557205	?	?	2

COQ6	ENST00000555196	?	ENSP00000451301	1
COQ6	ENST00000553922	?	?	1
COQ6	ENST00000238709	?	ENSP00000238709	3
COQ6	ENST00000557584	?	ENSP00000450511	2
COQ6	ENST00000554153	?	ENSP00000451685	2
COQ6	ENST00000394026	NM_182480.2	ENSP00000377594	4
COQ6	ENST00000554341	?	ENSP00000450736	4
COQ6	ENST00000334571	NM_182476.2	ENSP00000333946	3
COQ6	ENST00000555511	?	?	2
COQ6	ENST00000557780	?	?	1
COQ6	ENST00000554320	?	ENSP00000451123	3
COQ6	ENST00000554193	?	?	1
COQ6	ENST00000554920	?	ENSP00000451562	3
COQ6	ENST00000556300	?	?	2
COQ6	ENST00000554217	?	?	1
COQ6	ENST00000629426	?	ENSP00000486650	3
ENTPD5	ENST00000334696	NM_001249.2	ENSP00000335246	55
ENTPD5	ENST00000557325	?	ENSP00000451810	46
ENTPD5	ENST00000555829	?	ENSP00000451499	1
ENTPD5	ENST00000553284	?	ENSP00000451591	12
ENTPD5	ENST00000554664	?	?	48
ENTPD5	ENST00000556242	?	ENSP00000451483	48
ENTPD5	ENST00000556108	?	?	40
ENTPD5	ENST00000557681	?	?	19
FAM161B	ENST00000286544	NM_152445.2	ENSP00000286544	5
FAM161B	ENST00000556794	?	ENSP00000450889	1
LIN52	ENST00000554289	?	?	2
LIN52	ENST00000553404	?	?	13
LIN52	ENST00000554938	?	ENSP00000452513	13
LIN52	ENST00000554076	?	?	5
LIN52	ENST00000555028	NM_001024674.2	ENSP00000451812	13
PTGR2	ENST00000553326	?	?	3
PTGR2	ENST00000555661	?	ENSP00000452280	17
PTGR2	ENST00000556219	?	?	14
PTGR2	ENST00000267568	NM_152444.2	ENSP00000267568	16
PTGR2	ENST00000555976	?	?	8
PTGR2	ENST00000555228	NM_001146155.1, NM_001146154.1	ENSP00000450975	16
PTGR2	ENST00000553813	?	ENSP00000450824	14
RP5-1021I20.4	ENST00000556551	?	ENSP00000451484	40
RP5-1021I20.5	ENST00000555916	?	?	13
RP5-1021I20.5	ENST00000554009	?	?	15

ZNF410	ENST00000334521	?	ENSP00000334170	21
ZNF410	ENST00000442160	NM_001242924.1	ENSP00000407130	23
ZNF410	ENST00000555044	NM_021188.2, NM_001242928.1	ENSP00000451763	23
ZNF410	ENST00000557363	?	ENSP00000452199	3
ZNF410	ENST00000557214	?	ENSP00000451620	7
ZNF410	ENST00000541357	?	?	18
ZNF410	ENST00000615736	?	ENSP00000483073	16
ZNF410	ENST00000556396	?	ENSP00000451867	23
ZNF410	ENST00000554797	?	ENSP00000451116	1
ZNF410	ENST00000540593	NM_001242927.1	ENSP00000442228	23
ZNF410	ENST00000556797	?	ENSP00000451073	1
ZNF410	ENST00000412490	?	?	10
ZNF410	ENST00000554316	?	ENSP00000451748	9
ZNF410	ENST00000555602	?	?	7
ZNF410	ENST00000555620	?	?	9
ZNF410	ENST00000555730	?	ENSP00000451807	3
ZNF410	ENST00000557679	?	?	11
ZNF410	ENST00000556659	?	ENSP00000452023	3
ZNF410	ENST00000398139	?	ENSP00000381208	23
ZNF410	ENST00000556179	?	ENSP00000450884	2
ZNF410	ENST00000554582	?	ENSP00000450650	7
ZNF410	ENST00000557495	?	ENSP00000450569	3
ZNF410	ENST00000556160	?	ENSP00000451458	3
ZNF410	ENST00000324593	NM_001242926.1	ENSP00000323293	23

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ALDH6A1	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	3
CCDC176	ENST00000394009	NM_025057.2	ENSP00000377577	3
ENTPD5	ENST00000334696	NM_001249.2	ENSP00000335246	1
FAM161B	ENST00000286544	NM_152445.2	ENSP00000286544	1
PTGR2	ENST00000555661	?	ENSP00000452280	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
COQ6	ENST00000394026	NM_182480.2	ENSP00000377594	1
COQ6	ENST00000554341	?	ENSP00000450736	1
ENTPD5	ENST00000553284	?	ENSP00000451591	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
COQ6	ENST00000553448	?	1
COQ6	ENST00000556588	?	1

ENTPD5	ENST00000554664	?	4
PTGR2	ENST00000556219	?	1
RP5-1021I20.5	ENST00000555916	?	2
RP5-1021I20.6	ENST00000602874	?	1

Locus 62 (rs12651314)

SNiPAcard

Block annotations

Block info

genomic range	chr4:15,749,703-15,756,270
block size	6,568 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.788 [-2.591 – 0.219]	gene(s) hit or close-by	RP11-442P12.2
phastCons	μ = 0.005 [0 – 0.019]	eQTL gene(s)	BST1, CD38, RP11-115L11.1
GERP++	μ = 0.212 [-2.99 – 2.39]	potentially regulated gene(s)	–
CADD score	μ = 3.061 [2.042 – 4.102]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD38	ENST00000502843	ILMN_2233783	adipocyte	9.04×10-6 (p-value)	MuTHER consortium	4
CD38	ENST00000226279					
RP11-115L11.1	?	ENSG00000214846	transformed fibroblasts	3.48×10-7 (p-value)	GTEx Portal V6	4
CD38	?	ENSG00000004468	thyroid	6.54×10-6 (p-value)	GTEx Portal V6	2
BST1	ENST00000382346	ILMN_1770161	monocyte	8.49×10-5 (p-value)	Fairfax et al.	1
BST1	ENST00000265016					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001237252 (promoter flanking region)	2	NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		nervous (NH-A)	H3K27ac, DNase1
		blood (DND-41)	H3K4me1, H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF
ENSR00001683638 (CTCF binding site)	1	NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27ac, CTCF, H3K4me1, H3K9ac, H3K4me3
		nervous (NH-A)	H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-442P12.2	downstream gene variant	4498	ENST00000513715	?	?	1

Locus 63 (rs489286)

SNiPAcard

Block annotations

Block info

genomic range	chr1:160,710,157-160,725,854
block size	15,698 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.419 [-0.939 – 0.166]	gene(s) hit or close-by	SLAMF7
phastCons	μ = 0.002 [0 – 0.018]	eQTL gene(s)	LY9, SLAMF7
GERP++	μ = -1.916 [-4.78 – 0.85]	potentially regulated gene(s)	B4GALT3, LY9, NCSTN, PVRL4, RP11-544M22.3, SLAMF1
CADD score	μ = 4.046 [0.885 – 11.26]	disease gene(s)	NCSTN, PVRL4

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
NCSTN	ACNE INVERSA, FAMILIAL, 1	OMIM	MIM:142690
PVRL4	ECTODERMAL DYSPLASIA-SYNDACTYLY SYNDROME 1	OMIM	MIM:613573
PVRL4	ECTODERMAL DYSPLASIA-SYNDACTYLY SYNDROME 1	DECIPHER	MIM:613573
PVRL4	Ectodermal dysplasia - syndactyly syndrome	OrphaNet	OrphaNet:247820

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SLAMF7	ENST00000368043	ILMN_1710923	blood	1.56×10^{-15} (p-value)	MuTHER consortium	9
SLAMF7	ENST00000368042					
SLAMF7	ENST00000458602					
SLAMF7	ENST00000444090					
SLAMF7	ENST00000441662					
SLAMF7	ENST00000621377					
SLAMF7	ENST00000359331					
SLAMF7	ENST00000484221					
SLAMF7	ENST00000458104					
SLAMF7	?	ENSG00000026751	blood	5.58×10^{-5} (q-value)	SeeQTL DB (HapMap)	6
LY9	ENST00000392203	ILMN_1731928	b-cell	1.04×10^{-4} (p-value)	Fairfax et al.	1
LY9	ENST00000263285					
LY9	ENST00000368037					
LY9	ENST00000368035					
LY9	ENST00000490902	ILMN_1812278	blood	3.44×10^{-6} (p-value)	Westra et al.	1
LY9	ENST00000368039					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
SLAMF7	ENST00000368043	222838_at	chr1	blood	2.70×10^{-11} (p-value)	Dixon et al.	3
SLAMF7	ENST00000368042						
SLAMF7	ENST00000458602						

SLAMF7	ENST00000444090						
SLAMF7	ENST00000621377						
SLAMF7	ENST00000441662						
SLAMF7	ENST00000359331						
SLAMF7	ENST00000484221						
SLAMF7	ENST00000458104						
LY9	ENST00000368039	231124_x_at	chr1	blood	9.00×10^{-8} (p-value)	Dixon et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000040800	1	ENCP00000005127	NCSTN
		ENCP00000005137	SLAMF1
		ENCP00000005188	B4GALT3
ENCE00000041028	1	ENCP00000005160	RP11-544M22.3
		ENCP00000005170	PVRL4
		ENCP00000005147	LY9

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000671302 (promoter)	3	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me3
		embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	H3K27me3
		blood (GM12878)	TAF1, PU1, PolII, DNase1, Yy1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		blood (K562)	H3K27me3
		skin (NHEK)	DNase1, H3K4me3
ENSR00001587157 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K4me3
		embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	H3K27me3
		blood (GM12878)	PolII, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		blood (K562)	H3K27me3
ENSR00001587158 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (GM12878)	PolII, H3K79me2, H3K36me3
		A549	CTCF
		blood (K562)	H3K27me3, CTCF
		skin (NHEK)	DNase1, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SLAMF7	downstream gene variant	1250	ENST00000441662	NM_001282589.1	ENSP00000405605	1
SLAMF7	downstream gene variant	2396	ENST00000488819	?	?	2
SLAMF7	upstream gene variant, downstream gene variant	1243	ENST00000484221	?	?	3
SLAMF7	downstream gene variant	514	ENST00000495334	?	ENSP00000473590	5
SLAMF7	downstream gene variant	1243	ENST00000359331	NM_001282596.1, NM_001282592.1	ENSP00000352281	1
SLAMF7	downstream gene variant	1243	ENST00000368043	NM_021181.4	ENSP00000357022	1
SLAMF7	downstream gene variant	1250	ENST00000444090	NM_001282588.1	ENSP00000416592	1
SLAMF7	downstream gene variant	1250	ENST00000458602	NM_001282591.1	ENSP00000409965	1

SLAMF7	downstream gene variant	1250	ENST00000621377	NM_001282594.1	ENSP00000483774	1
SLAMF7	downstream gene variant	1243	ENST00000368042	NM_001282590.1, NM_001282595.1	ENSP00000357021	1
SLAMF7	downstream gene variant	1250	ENST00000458104	NM_001282593.1	ENSP00000403294	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLAMF7	ENST00000359331	NM_001282596.1, NM_001282592.1	ENSP00000352281	9
SLAMF7	ENST00000368043	NM_021181.4	ENSP00000357022	9
SLAMF7	ENST00000488819	?	?	7
SLAMF7	ENST00000458602	NM_001282591.1	ENSP00000409965	9
SLAMF7	ENST00000458104	NM_001282593.1	ENSP00000403294	9
SLAMF7	ENST00000368042	NM_001282590.1, NM_001282595.1	ENSP00000357021	9
SLAMF7	ENST00000441662	NM_001282589.1	ENSP00000405605	9
SLAMF7	ENST00000444090	NM_001282588.1	ENSP00000416592	9
SLAMF7	ENST00000495334	?	ENSP00000473590	8
SLAMF7	ENST00000621377	NM_001282594.1	ENSP00000483774	9
SLAMF7	ENST00000484221	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLAMF7	ENST00000359331	NM_001282596.1, NM_001282592.1	ENSP00000352281	3
SLAMF7	ENST00000368043	NM_021181.4	ENSP00000357022	3
SLAMF7	ENST00000458602	NM_001282591.1	ENSP00000409965	3
SLAMF7	ENST00000458104	NM_001282593.1	ENSP00000403294	3
SLAMF7	ENST00000368042	NM_001282590.1, NM_001282595.1	ENSP00000357021	3
SLAMF7	ENST00000441662	NM_001282589.1	ENSP00000405605	3
SLAMF7	ENST00000444090	NM_001282588.1	ENSP00000416592	3
SLAMF7	ENST00000621377	NM_001282594.1	ENSP00000483774	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SLAMF7	ENST00000484221	?	4

Locus 64 (rs7522061)

SNiPAcard

Block annotations

Block info

genomic range	chr1:157,661,848-157,693,722
block size	31,875 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.795 [-3.247 – 0.025]	gene(s) hit or close-by	FCRL3, RP11-367J7.3, VDAC1P9
phastCons	μ = 0.062 [0 – 0.694]	eQTL gene(s)	ATP1A4, FCRL1, FCRL3, RP11-367J7.3
GERP++	μ = -1.674 [-10.7 – 1.7]	potentially regulated gene(s)	–
CADD score	μ = 2.100 [0.524 – 4.934]	disease gene(s)	FCRL3

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Graves' disease	$<2.00\times10^{-13}$	GWAS Catalog	21841780	1
Multiple sclerosis	$<3.00\times10^{-6}$	GWAS Catalog	21833088	1
Type 1 diabetes autoantibodies	$<1.00\times10^{-11}$	GWAS Catalog	21829393	1
Alkaline phosphatase	8.18×10^{-5}	dbGaP	pha001181	1
Alkaline phosphatase	3.25×10^{-5}	dbGaP	pha001179	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR067135	1
?	HGMD curated	HGMD	CR067134	1
?	HGMD curated	HGMD	CR052070	1

Disease gene annotation

gene	trait	source DB	source entry/link	
FCRL3	RHEUMATOID ARTHRITIS	OMIM	MIM:180300	

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FCRL3	missense variant	ENST00000485028	?	ENSP00000434331	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000492769	?	ENSP00000435487	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000496769	?	ENSP00000473680	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000368186	?	ENSP00000357169	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000477837	?	ENSP00000433430	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000368184	NM_052939.3	ENSP00000357167	N/D	Aat/Gat	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCRL3	ENST00000494724	ILMN_1691693	blood	1.12×10^{-20} (p-value)	MuTHER consortium	8
FCRL3	ENST00000485028					
FCRL3	ENST00000368186					
FCRL3	ENST00000477837					
FCRL3	ENST00000492769					
FCRL3	ENST00000368184					
FCRL3	ENST00000480682					
FCRL3	ENST00000494724	ILMN_1699599	blood	9.83×10^{-16} (p-value)	MuTHER consortium	8
FCRL3	ENST00000485028					
FCRL3	ENST00000477837					
FCRL3	ENST00000473231					
FCRL3	ENST00000468507					
FCRL3	ENST00000480682					
FCRL3	ENST00000492769					
FCRL3	ENST00000368184					
FCRL3	ENST00000473231	ILMN_1797428	blood	9.84×10^{-17} (p-value)	MuTHER consortium	8
FCRL3	ENST00000368184					
FCRL3	ENST00000468507					
FCRL3	?	ENSG00000160856	spleen	1.70×10^{-8} (p-value)	GTEx Portal V6	13
RP11-367J7.3	?	ENSG00000227217	spleen	9.09×10^{-8} (p-value)	GTEx Portal V6	5
FCRL3	?	ENSG00000160856	lung	2.74×10^{-9} (p-value)	GTEx Portal V6	13
RP11-367J7.3	?	ENSG00000227217	lung	5.02×10^{-8} (p-value)	GTEx Portal V6	11
RP11-367J7.3	?	ENSG00000227217	blood	7.86×10^{-13} (p-value)	GTEx Portal V6	13
FCRL3	?	ENSG00000160856	blood	2.00×10^{-19} (p-value)	GTEx Portal V6	13
FCRL1	?	ENSG00000163534	blood	8.70×10^{-7} (p-value)	GTEx Portal V6	4
FCRL3	?	ENSG00000160856	esophagus mucosa	5.44×10^{-9} (p-value)	GTEx Portal V6	9
ATP1A4	ENST00000470705	ILMN_1705208	b-cell	1.09×10^{-4} (p-value)	Fairfax et al.	3
ATP1A4	ENST00000466526					
FCRL3	ENST00000494724	ILMN_1691693	monocyte	1.27×10^{-6} (p-value)	Fairfax et al.	3
FCRL3	ENST00000485028		b-cell	7.43×10^{-33} (p-value)	Fairfax et al.	3
FCRL3	ENST00000477837					
FCRL3	ENST00000368186					
FCRL3	ENST00000492769					
FCRL3	ENST00000368184					
FCRL3	ENST00000480682					
FCRL3	ENST00000494724	ILMN_1699599	b-cell	2.37×10^{-30} (p-value)	Fairfax et al.	2
FCRL3	ENST00000485028					
FCRL3	ENST00000477837					
FCRL3	ENST00000473231					
FCRL3	ENST00000480682					
FCRL3	ENST00000468507					
FCRL3	ENST00000492769					
FCRL3	ENST00000368184					

FCRL3	ENST00000368184	ILMN_1797428	monocyte	1.02×10^{-4} (p-value)	Fairfax et al.	3
FCRL3	ENST00000473231		b-cell	5.93×10^{-26} (p-value)	Fairfax et al.	3
FCRL3	ENST00000468507					
FCRL3	?	ENSG00000160856	subcutaneous adipocytes	5.58×10^{-8} (p-value)	GTEx Portal V6	7

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
FCRL3	ENST00000457799	231093_at	chr1	blood	1.00×10^{-10} (p-value)	Dixon et al.	3
FCRL3	ENST00000368184						

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000671247 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	CTCF, H3K27me3
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K4me1
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3
ENSR00000545969 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCRL3	upstream gene variant	283	ENST00000485028	?	ENSP00000434331	1
FCRL3	downstream gene variant, upstream gene variant	314	ENST00000478179	?	?	7
FCRL3	upstream gene variant	296	ENST00000494724	?	?	1
FCRL3	downstream gene variant, upstream gene variant	281	ENST00000496769	?	ENSP00000473680	2
FCRL3	upstream gene variant	587	ENST00000473231	?	?	6
FCRL3	upstream gene variant	2168	ENST00000468507	?	?	1
FCRL3	upstream gene variant	288	ENST00000368186	?	ENSP00000357169	1
FCRL3	upstream gene variant	296	ENST00000477837	?	ENSP00000433430	1
FCRL3	upstream gene variant	296	ENST00000480682	?	?	1
FCRL3	upstream gene variant	296	ENST00000492769	?	ENSP00000435487	1
FCRL3	upstream gene variant	169	ENST00000368184	NM_052939.3	ENSP00000357167	1
RP11-367J7.3	downstream gene variant	395	ENST00000453692	?	?	7
VDAC1P9	downstream gene variant	248	ENST00000412766	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCRL3	ENST00000477837	?	ENSP00000433430	5
FCRL3	ENST00000485028	?	ENSP00000434331	5
FCRL3	ENST00000480682	?	?	5
FCRL3	ENST00000368186	?	ENSP00000357169	5
FCRL3	ENST00000494724	?	?	5
FCRL3	ENST00000473231	?	?	2
FCRL3	ENST00000492769	?	ENSP00000435487	5
FCRL3	ENST00000368184	NM_052939.3	ENSP00000357167	5
FCRL3	ENST00000496769	?	ENSP00000473680	3
RP11-367J7.3	ENST00000453692	?	?	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCRL3	ENST00000477837	?	ENSP00000433430	1
FCRL3	ENST00000485028	?	ENSP00000434331	1
FCRL3	ENST00000368186	?	ENSP00000357169	1
FCRL3	ENST00000492769	?	ENSP00000435487	1
FCRL3	ENST00000368184	NM_052939.3	ENSP00000357167	1
FCRL3	ENST00000496769	?	ENSP00000473680	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FCRL3	ENST00000480682	?	3
FCRL3	ENST00000494724	?	3
FCRL3	ENST00000473231	?	1

Locus 65 (rs692804)

SNiPAcard

Block annotations

Block info

genomic range	chr11:120,098,044-120,100,867
block size	2,824 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.208 [-2.697 – 2.393]	gene(s) hit or close-by	OAF
phastCons	μ = 0.129 [0 – 1]	eQTL gene(s)	POU2F3
GERP++	μ = -0.794 [-5.98 – 5.26]	potentially regulated gene(s)	ARHGEF12, CTD-2523D13.2, OAF, POU2F3, PVRL1, TMEM136, TRIM29
CADD score	μ = 4.439 [0.537 – 15.33]	disease gene(s)	PVRL1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PVRL1	CLEFT LIP/PALATE-ECTODERMAL DYSPLASIA SYNDROME	OMIM	MIM:225060
PVRL1	CLEFT LIP/PALATE-ECTODERMAL DYSPLASIA SYNDROME	DECIPHER	MIM:225060
PVRL1	Zlotogora-Ogur syndrome	OrphaNet	OrphaNet:3253

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
OAF	missense variant	ENST00000328965	NM_178507.2	ENSP00000332613	H/R	cAc/cGc	tolerated	benign	1
OAF	missense variant	ENST00000531220	?	ENSP00000431865	H/R	cAc/cGc	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
POU2F3	?	ENSG00000137709	subcutaneous adipocytes	6.18×10^{-8} (p-value)	GTEx Portal V6	8

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000047292	1	ENST00000328965, ENST00000531220	OAF

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000125097	1	ENCP00000015007	CTD-2523D13.2 PVRL1
		ENCP00000015012	TRIM29
		ENCP00000015027	OAF
		ENCP00000015028	POU2F3
		ENCP00000015032	TMEM136
		ENCP00000015034	ARHGEF12

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000112413 (CTCF binding site)	3	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, H3K36me3, PolII, Rad21, CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF, H3K36me3
		nervous (NH-A)	CTCF
		skin (NHEK)	H3K36me3, CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K27me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
OAF	downstream gene variant	571	ENST00000531220	?	ENSP00000431865	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
OAF	ENST00000328965	NM_178507.2	ENSP00000332613	4
OAF	ENST00000531220	?	ENSP00000431865	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
OAF	ENST00000328965	NM_178507.2	ENSP00000332613	3

Locus 66 (rs35395347)

SNiPAcard

Block annotations

Block info

genomic range	chr3:49,277,998-49,335,745
block size	57,748 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.296 [-0.129 – 0.887]	gene(s) hit or close-by	CCDC36, USP4
phastCons	μ = 0.003 [0 – 0.013]	eQTL gene(s)	AMT, APEH, MRPS18AP1, MST1, NICN1, ZNF589
GERP++	μ = 0.732 [-1.39 – 1.88]	potentially regulated gene(s)	–
CADD score	μ = 3.251 [1.691 – 5.129]	disease gene(s)	MST1, AMT

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
MST1	Primary sclerosing cholangitis	OrphaNet	OrphaNet:171
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MRPS18AP1	?	ENSG00000229759	muscularis mucosae	1.57×10-6 (p-value)	GTEx Portal V6	1
MST1	?	ENSG00000173531	tibial nerve	4.13×10-5 (p-value)	GTEx Portal V6	5
MST1	?	ENSG00000173531	transformed fibroblasts	3.39×10-6 (p-value)	GTEx Portal V6	5
APEH	?	ENSG00000164062	transformed fibroblasts	1.99×10-5 (p-value)	GTEx Portal V6	5
MST1	?	ENSG00000173531	esophagus mucosa	2.20×10-6 (p-value)	GTEx Portal V6	5
MRPS18AP1	?	ENSG00000229759	esophagus mucosa	3.17×10-5 (p-value)	GTEx Portal V6	1
ZNF589	?	ENSG00000164048	esophagus mucosa	1.05×10-5 (p-value)	GTEx Portal V6	1
APEH	?	ENSG00000164062	esophagus mucosa	4.03×10-7 (p-value)	GTEx Portal V6	5
MST1	?	ENSG00000173531	subcutaneous adipocytes	2.11×10-6 (p-value)	GTEx Portal V6	5
NICN1	ENST00000273598	ILMN_1807211	blood	1.57×10-6 (p-value)	Westra et al.	1
NICN1	ENST00000615713					
AMT	ENST00000273588					
NICN1	ENST00000423832					
APEH	?	ENSG00000164062	unexposed skin	1.65×10-5 (p-value)	GTEx Portal V6	4
MST1	?	ENSG00000173531	thyroid	3.24×10-5 (p-value)	GTEx Portal V6	4

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001479015 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF
		HSMMtube	H3K36me3
		blood (K562)	Max, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF, H3K36me3
		liver (HepG2)	Rad21, CTCF, H3K36me3
		blood (GM12878)	Rad21, CTCF, H3K36me3
		lung (IMR90)	CTCF, H3K36me3
		nervous (NH-A)	CTCF, H3K36me3
		skin (NHEK)	H3K36me3, CTCF
		NHLF	CTCF
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF, H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	CTCF, H3K36me3
		A549	H3K36me3, CTCF
ENSR00001479016 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K4me1, H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	H3K36me3, DNase1
		muscle (HSMM)	H3K27ac, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K27ac, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	DNase1, H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCDC36	downstream gene variant	3991	ENST00000493870	?	?	1
USP4	upstream gene variant	1738	ENST00000475873	?	?	4
USP4	downstream gene variant	506	ENST00000486549	?	?	3
USP4	downstream gene variant	2283	ENST00000488520	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCDC36	ENST00000366429	?	ENSP00000403700	1
CCDC36	ENST00000296449	NM_178173.3	ENSP00000296449	1

CCDC36	ENST00000452691	NM_001135197.1	ENSP00000407837	1
CCDC36	ENST00000438782	?	ENSP00000391788	1
USP4	ENST00000485450	?	?	4
USP4	ENST00000265560	NM_003363.3	ENSP00000265560	4
USP4	ENST00000431357	?	ENSP00000399079	4
USP4	ENST00000351842	NM_199443.2	ENSP00000341028	4
USP4	ENST00000486549	?	?	1

Locus 66 (sentinel SNP rs35395347 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 67 (rs9302635)

SNiPAcard

Block annotations

Block info

genomic range	chr16:72,078,043-72,227,398
block size	149,356 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.615 [-2.712 – 0.203]	**gene(s) hit or close-by**	DHX38, PMFBP1, TXNL4B
phastCons	μ = 0.119 [0 – 0.83]	**eQTL gene(s)**	DHODH, DHX38, HP, HPR, TAT-AS1
GERP++	μ = -1.104 [-3.54 – 0.149]	**potentially regulated gene(s)**	–
CADD score	μ = 4.966 [0.747 – 11.04]	**disease gene(s)**	DHODH, HP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
gamma-glutamyltyrosine	9.15×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
DHODH	POSTAXIAL ACROFACIAL DYSOSTOSIS	OMIM	MIM:263750
HP	ANHAPTOGLOBINEMIA	OMIM	MIM:614081
DHODH	postaxial acrofacial dysostosis (POADS)	DECIPHER	MIM:263750
DHODH	Postaxial acrofacial dysostosis	OrphaNet	OrphaNet:246

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TAT-AS1	?	ENSG00000260886	testis	2.38×10^{-6} (p-value)	GTEx Portal V6	4
DHODH	?	ENSG00000102967	tibial nerve	1.02×10^{-7} (p-value)	GTEx Portal V6	6
DHODH	?	ENSG00000102967	transformed fibroblasts	3.63×10^{-6} (p-value)	GTEx Portal V6	5
DHODH	?	ENSG00000102967	tibial artery	2.90×10^{-6} (p-value)	GTEx Portal V6	3
HPR	?	ENSG00000261701	left ventricle	4.07×10^{-7} (p-value)	GTEx Portal V6	4
HPR	?	ENSG00000261701	blood	4.70×10^{-15} (p-value)	GTEx Portal V6	7
DHODH	?	ENSG00000102967	muscularis mucosae	1.53×10^{-5} (p-value)	GTEx Portal V6	1
HPR	?	ENSG00000261701	nucleus accumbens	5.64×10^{-6} (p-value)	GTEx Portal V6	1
DHX38	?	ENSG00000140829	EBV lymphocytes	3.30×10^{-6} (p-value)	GTEx Portal V6	1
HP	ENST00000570083	ILMN_1812433	monocyte	1.33×10^{-10} (p-value)	Fairfax et al.	1
HP	ENST00000567185					
HP	ENST00000566821					
HP	ENST00000613898					

HP	ENST00000565574					
HP	ENST00000228226					
HP	ENST00000567612					
HP	ENST00000355906					
HP	ENST00000398131					
HP	ENST00000564499					
HP	ENST00000357763					
HP	ENST00000570083	ILMN_1812433	monocyte	4.52×10^{-48} (p-value)	Zeller et al.	1
HP	ENST00000567185					
HP	ENST00000566821					
HP	ENST00000613898					
HP	ENST00000565574					
HP	ENST00000228226					
HP	ENST00000355906					
HP	ENST00000567612					
HP	ENST00000398131					
HP	ENST00000564499					
HP	ENST00000357763					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DHX38	downstream gene variant	1318	ENST00000566794	?	ENSP00000455939	1
DHX38	downstream gene variant	1397	ENST00000564622	?	?	1
DHX38	downstream gene variant	3112	ENST00000268482	NM_014003.3	ENSP00000268482	1
DHX38	downstream gene variant	3116	ENST00000562774	?	ENSP00000462965	1
DHX38	downstream gene variant	1363	ENST00000566489	?	ENSP00000457887	1
DHX38	upstream gene variant	2852	ENST00000566329	?	?	1
DHX38	upstream gene variant	4871	ENST00000569935	?	?	1
DHX38	downstream gene variant	3385	ENST00000563819	?	ENSP00000462397	1
DHX38	downstream gene variant	3115	ENST00000567142	?	ENSP00000454521	1
DHX38	downstream gene variant	3374	ENST00000567552	?	?	1
DHX38	downstream gene variant	519	ENST00000569952	?	?	1
DHX38	downstream gene variant	3121	ENST00000579387	?	ENSP00000462149	1
PMFBP1	downstream gene variant	1882	ENST00000537792	?	ENSP00000443366	1
PMFBP1	downstream gene variant	3120	ENST00000535461	?	ENSP00000444953	1
PMFBP1	downstream gene variant	3077	ENST00000379073	?	ENSP00000368364	1
PMFBP1	downstream gene variant	2153	ENST00000537392	?	ENSP00000439564	1
PMFBP1	downstream gene variant	3073	ENST00000237353	NM_031293.2	ENSP00000237353	1
PMFBP1	downstream gene variant	3076	ENST00000355636	NM_001160213.1	ENSP00000347854	1
PMFBP1	downstream gene variant	3073	ENST00000537465	?	ENSP00000443817	1
TXNL4B	upstream gene variant	3799	ENST00000426362	NM_001142318.1	ENSP00000392310	1
TXNL4B	upstream gene variant	4361	ENST00000569767	?	ENSP00000457699	1
TXNL4B	upstream gene variant	4579	ENST00000268483	NM_017853.2	ENSP00000268483	1
TXNL4B	downstream gene variant, upstream gene variant	145	ENST00000562153	?	ENSP00000454635	2

TXNL4B	downstream gene variant, upstream gene variant	145	ENST00000562153	?	ENSP00000454635	2
TXNL4B	upstream gene variant	4361	ENST00000423037	NM_001142317.1	ENSP00000408130	1
TXNL4B	upstream gene variant	4678	ENST00000565171	?	ENSP00000455550	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DHX38	ENST00000567142	?	ENSP00000454521	1
DHX38	ENST00000563650	?	?	1
DHX38	ENST00000562774	?	ENSP00000462965	1
DHX38	ENST00000564307	?	?	1
DHX38	ENST00000567552	?	?	1
DHX38	ENST00000579387	?	ENSP00000462149	2
DHX38	ENST00000268482	NM_014003.3	ENSP00000268482	2
DHX38	ENST00000563819	?	ENSP00000462397	1
PMFBP1	ENST00000537792	?	ENSP00000443366	2
PMFBP1	ENST00000379073	?	ENSP00000368364	2
PMFBP1	ENST00000237353	NM_031293.2	ENSP00000237353	2
PMFBP1	ENST00000537465	?	ENSP00000443817	2
PMFBP1	ENST00000355636	NM_001160213.1	ENSP00000347854	2
PMFBP1	ENST00000537392	?	ENSP00000439564	1

Locus 68 (rs4693167)

SNiPAcard

Block annotations

Block info

genomic range	chr4:88,400,924-88,431,892
block size	30,969 bp
variant count	37 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.799 [-3.224 – 0.94]	gene(s) hit or close-by	SPARCL1
phastCons	μ = 0.045 [0 – 0.942]	eQTL gene(s)	HSD17B11, HSD17B13, NUDT9, PPM1K, RP11-529H2.2, SPARCL1
GERP++	μ = 0.298 [-4.01 – 4.25]	potentially regulated gene(s)	RP11-529H2.1, SPARCL1
CADD score	μ = 3.588 [0.322 – 10.4]	disease gene(s)	PPM1K

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PPM1K	MAPLE SYRUP URINE DISEASE, MILD VARIANT	OMIM	MIM:615135
PPM1K	Intermediate maple syrup urine disease	OrphaNet	OrphaNet:268162

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SPARCL1	missense variant	ENST00000458304	?	ENSP00000406251	A/D	gCt/gAt	?	benign	1
SPARCL1	missense variant	ENST00000418378	NM_001128310.1	ENSP00000414856	2	2			2
SPARCL1	missense variant	ENST00000434434	?	ENSP00000416971	2	2			2
SPARCL1	missense variant	ENST00000282470	NM_004684.4	ENSP00000282470	2	2			2
SPARCL1	missense variant	ENST00000535835	?	ENSP00000438188	2	2			2
SPARCL1	missense variant	ENST00000509407	?	ENSP00000423483	2	2			2
SPARCL1	missense variant	ENST00000512317	?	ENSP00000423448	2	2			2
SPARCL1	missense variant	ENST00000543631	?	ENSP00000444832	A/D	gCt/gAt	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SPARCL1	ENST00000282470	ILMN_2218208	adipocyte	1.24×10-6 (p-value)	MuTHER consortium	7
SPARCL1	ENST00000503414					
SPARCL1	ENST00000418378					
SPARCL1	ENST00000282470	ILMN_1795251	adipocyte	1.65×10-6 (p-value)	MuTHER consortium	7
SPARCL1	ENST00000503414					
SPARCL1	ENST00000418378					

NUDT9	ENST00000302174	ILMN_1680239	blood	5.72×10-6 (p-value)	Westra et al.	6
NUDT9	ENST00000473942					
NUDT9	ENST00000440591					
HSD17B11	ENST00000508413	ILMN_1675117	blood	5.44×10-7 (p-value)	Westra et al.	4
HSD17B11	ENST00000358290					
HSD17B11	?	ENSG00000198189	tibial nerve	3.20×10-6 (p-value)	GTEx Portal V6	25
SPARCL1	?	ENSG00000152583	sun exposed skin	5.36×10-7 (p-value)	GTEx Portal V6	26
HSD17B11	?	ENSG00000198189	esophagus mucosa	1.01×10-7 (p-value)	GTEx Portal V6	36
SPARCL1	?	ENSG00000152583	subcutaneous adipocytes	5.55×10-6 (p-value)	GTEx Portal V6	14
HSD17B13	?	ENSG00000170509	thyroid	2.57×10-6 (p-value)	GTEx Portal V6	12
PPM1K	ENST00000608933	ILMN_2070043	b-cell	6.14×10-4 (p-value)	Fairfax et al.	1
PPM1K	ENST00000295908					
HSD17B11	?	ENSG00000198189	pancreas	5.48×10-6 (p-value)	GTEx Portal V6	3
RP11-529H2.2	?	ENSG00000255723	pancreas	1.05×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000660191	1	ENST00000503414, ENST00000541496	SPARCL1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000384981	1	ENCP00000041684	RP11-529H2.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001244943 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF, DNase1, H3K4me2
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF
		skin (NHEK)	CTCF
ENSR00001244946 (promoter flanking region)	4	cervix (HeLa-S3)	DNase1
		muscle (HSMM)	DNase1
ENSR00001686554 (CTCF binding site)	2	cervix (HeLa-S3)	DNase1
		muscle (HSMM)	DNase1
ENSR00001244947 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		liver (HepG2)	CTCF
		blood (K562)	CTCF
		skin (NHEK)	CTCF
ENSR00001432957 (promoter flanking region)	8	NHLF	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	CTCF, DNase1
		HSMMtube	DNase1
		Osteobl	H3K4me2, H3K27ac

blood (K562)	CTCF
skin (NHDF-AD)	H3K4me1, DNase1, H3K4me2, H3K27ac
muscle (HSMM)	H3K4me1, H2AZ, H3K27ac, H3K4me2, DNase1
endothelium (HUVEC)	CTCF
nervous (NH-A)	DNase1
skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SPARCL1	downstream gene variant	2545	ENST00000535835	?	ENSP00000438188	1
SPARCL1	downstream gene variant	2504	ENST00000434434	?	ENSP00000416971	1
SPARCL1	downstream gene variant	49	ENST00000543631	?	ENSP00000444832	2
SPARCL1	downstream gene variant	529	ENST00000458304	?	ENSP00000406251	2
SPARCL1	downstream gene variant	2568	ENST00000512317	?	ENSP00000423448	1
SPARCL1	downstream gene variant	2497	ENST00000541496	?	ENSP00000445678	1
SPARCL1	downstream gene variant	2497	ENST00000509407	?	ENSP00000423483	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SPARCL1	ENST00000543631	?	ENSP00000444832	31
SPARCL1	ENST00000503414	?	ENSP00000422903	35
SPARCL1	ENST00000282470	NM_004684.4	ENSP00000282470	35
SPARCL1	ENST00000512317	?	ENSP00000423448	31
SPARCL1	ENST00000418378	NM_001128310.1	ENSP00000414856	35
SPARCL1	ENST00000458304	?	ENSP00000406251	31
SPARCL1	ENST00000541496	?	ENSP00000445678	31
SPARCL1	ENST00000535835	?	ENSP00000438188	31
SPARCL1	ENST00000434434	?	ENSP00000416971	31
SPARCL1	ENST00000509407	?	ENSP00000423483	31

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SPARCL1	ENST00000503414	?	ENSP00000422903	2
SPARCL1	ENST00000541496	?	ENSP00000445678	2

Locus 69 (rs8176749)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,115,176-136,184,526
block size	69,351 bp
variant count	25 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.373 [-2.969 – 1.925]	gene(s) hit or close-by	ABO, LCN1P2, Y_RNA
phastCons	μ = 0.069 [0 – 0.636]	eQTL gene(s)	ABO, MED22, OBP2B, SLC2A6
GERP++	μ = -0.804 [-8.76 – 2.51]	potentially regulated gene(s)	ABO, ADAMTS13, ADAMTS13, ADAMTSL2, AK8, C9orf9, CELP, DBH, GFI1B, GTF3C5, OBP2B, OBP2B, RALGDS, REXO4, REXO4, SARDH, SURF1, SURF1, SURF2, SURF2, SURF4, SURF4
CADD score	μ = 3.314 [0.003 – 16.8]	disease gene(s)	SURF1, , DBH, ADAMTSL2, ADAMTS13, SARDH, GFI1B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Intraocular pressure	$<3.00\times10^{-11}$	GWAS Catalog	25173106	1
High serum lipase activity	$<1.00\times10^{-22}$	GWAS Catalog	25028398	1
Elevated serum carcinoembryonic antigen levels	$<2.00\times10^{-24}$	GWAS Catalog	24941225	1
Urinary metabolites (H-NMR features)	$<4.00\times10^{-12}$	GWAS Catalog	24586186	1
End-stage coagulation	$<2.00\times10^{-17}$	GWAS Catalog	23381943	1
Tumor biomarkers	$<7.00\times10^{-105}$	GWAS Catalog	23300138	1
Coagulation factor levels	$<2.00\times10^{-138}$	GWAS Catalog	23267103	1
Mean corpuscular hemoglobin concentration	$<4.00\times10^{-8}$	GWAS Catalog	20139978	1
Parkinson disease	8.66×10^{-6}	dbGaP	pha002868	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Abo blood group system, b(a) phenotype	association	ClinVar	RCV000019312.3	1
ABO BLOOD GROUP SYSTEM, CIS-AB PHENOTYPE	OMIM curated	OMIM	MIM:110300	1

Disease gene annotation

gene	trait	source DB	source entry/link
SURF1	Complex IV deficiency	DECIPHER	MIM:220110
SURF1	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
	Complex IV deficiency	DECIPHER	MIM:220110
	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY, CONGENITAL	OMIM	MIM:223360
ADAMTSL2	GELEOPHYSIC DYSPLASIA 1	OMIM	MIM:231050
SURF1	LEIGH SYNDROME	OMIM	MIM:256000
ADAMTS13	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL	OMIM	MIM:274150
SARDH	SARCOSINEMIA	OMIM	MIM:268900
GFI1B	BLEEDING DISORDER, PLATELET-TYPE, 17	OMIM	MIM:187900
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY	OrphaNet	OrphaNet:230

ADAMTSL2	Geleophysic dysplasia	OrphaNet	OrphaNet:2623	orphanet
SURF1	Fatal infantile cytochrome C oxidase deficiency	OrphaNet	OrphaNet:1561	orphanet
SURF1	Leigh syndrome with cardiomyopathy	OrphaNet	OrphaNet:70474	orphanet
SURF1	Leigh syndrome with leukodystrophy	OrphaNet	OrphaNet:255241	orphanet
SURF1	SURF1-related Charcot-Marie-Tooth disease type 4	OrphaNet	OrphaNet:391351	orphanet
ADAMTS13	Congenital thrombotic thrombocytopenic purpura due to ADAMTS-13 deficiency	OrphaNet	OrphaNet:93583	orphanet
SARDH	SARCOSINEMIA	OrphaNet	OrphaNet:3129	orphanet
GFI1B	GRAY PLATELET SYNDROME	OrphaNet	OrphaNet:721	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ABO	missense variant	ENST00000538324	?	ENSP00000483018	3	3			3
ABO	missense variant	ENST00000611156	NM_020469.2	ENSP00000483265	3	3			3

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABO	?	ENSG00000175164	stomach	1.87×10^{-11} (p-value)	GTEx Portal V6	23
ABO	?	ENSG00000175164	lung	1.34×10^{-22} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	tibial nerve	2.83×10^{-18} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	thyroid	2.25×10^{-34} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	esophagus mucosa	4.41×10^{-26} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	skeletal muscle	2.99×10^{-11} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	transverse colon	2.25×10^{-8} (p-value)	GTEx Portal V6	22
ABO	?	ENSG00000175164	unexposed skin	1.58×10^{-26} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	sun exposed skin	4.86×10^{-27} (p-value)	GTEx Portal V6	24
OBP2B	?	ENSG00000171102	sun exposed skin	5.09×10^{-8} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	subcutaneous adipocytes	7.24×10^{-10} (p-value)	GTEx Portal V6	21
ABO	?	ENSG00000175164	muscularis mucosae	1.92×10^{-7} (p-value)	GTEx Portal V6	19
ABO	?	ENSG00000175164	visceral adipocytes	2.37×10^{-8} (p-value)	GTEx Portal V6	17
ABO	?	ENSG00000175164	pancreas	7.12×10^{-7} (p-value)	GTEx Portal V6	15
ABO	?	ENSG00000175164	testis	4.43×10^{-9} (p-value)	GTEx Portal V6	20
ABO	?	ENSG00000175164	left ventricle	3.30×10^{-8} (p-value)	GTEx Portal V6	18
MED22	ENST00000614493	ILMN_1697218	monocyte	1.69×10^{-4} (p-value)	Fairfax et al.	2
MED22	ENST00000610672					
MED22	ENST00000343730					
MED22	ENST00000610888					

SLC2A6	ENST00000371899	ILMN_1778321	blood	5.84×10-7 (p-value)	Westra et al.	6
SLC2A6	ENST00000485978					
SLC2A6	ENST00000371897					
ABO	?	ENSG00000175164	adrenal gland	2.08×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000859024	1	ENST00000538324, ENST00000453660	ABO

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000558431	1	ENCP00000060477	GFI1B
		ENCP00000060525	ADAMTSL2
		ENCP00000060529	DBH
		ENCP00000060490	RALGDS
		ENCP00000060486	CELP
		ENCP00000060489	RALGDS
		ENCP00000060518	ADAMTS13 ADAMTS13
		ENCP00000060527	ADAMTSL2
		ENCP00000060535	SARDH
		ENCP00000060507	SURF2 SURF2 SURF1 SURF1
		ENCP00000060492	RALGDS
ENCE00000558621	1	ENCP00000060495	OBP2B OBP2B
ENCE00000558373	3	ENCP00000060477	GFI1B
		ENCP00000060471	AK8 C9orf9
		ENCP00000060525	ADAMTSL2
		ENCP00000060529	DBH
		ENCP00000060516	REXO4 REXO4
		ENCP00000060509	SURF4 SURF4
		ENCP00000060490	RALGDS
		ENCP00000060486	CELP
		ENCP00000060489	RALGDS
		ENCP00000060518	ADAMTS13 ADAMTS13
		ENCP00000060527	ADAMTSL2
		ENCP00000060535	SARDH
		ENCP00000060507	SURF2 SURF2 SURF1 SURF1
		ENCP00000060492	RALGDS
ENCE00000558506	1	ENCP00000060483	GTF3C5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001734089 (enhancer)	2	liver (HepG2)	H3K4me1, H3K4me2
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K4me3, H3K4me2
		blood (K562)	H3K4me2, H2AZ
		skin (NHDF-AD)	H3K4me2
ENSR00000452813 (promoter flanking region)	2	cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, PolII, DNase1
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
ENSR00001475637 (TF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K36me3, H3K4me2, H3K9ac, H3K27ac, DNase1, Brg1, Ini1, PolII
		A549	H3K27me3
ENSR00001475641 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABO	downstream gene variant	2696	ENST00000453660	?	?	1
ABO	downstream gene variant	248	ENST00000538324	?	ENSP00000483018	7
ABO	downstream gene variant	848	ENST00000611156	NM_020469.2	ENSP00000483265	6
LCN1P2	upstream gene variant	103	ENST00000372030	?	?	1
Y_RNA	downstream gene variant	4516	ENST00000364973	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	2	2	2
ABO	ENST00000538324	?	ENSP00000483018	2	2	2

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000453660	?	?	7
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	7
ABO	ENST00000538324	?	ENSP00000483018	7

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ABO	ENST00000453660	?	12

Locus 70 (rs6138458)

SNiPAcard

Block annotations

Block info

genomic range	chr20:24,899,905-24,977,586
block size	77,682 bp
variant count	61 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.131 [-4.848 – 0.469]	gene(s) hit or close-by	APMAP, CST7, RNU6-1257P, RP4-568C11.4
phastCons	μ = 0.048 [0 – 0.647]	eQTL gene(s)	APMAP, CST7, NINL
GERP++	μ = -0.368 [-8.11 – 1.91]	potentially regulated gene(s)	RP5-1025A1.2
CADD score	μ = 2.517 [0.005 – 11.69]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
APMAP	?	ENSG00000101474	pancreas	1.50×10^{-9} (p-value)	GTEx Portal V6	61
APMAP	?	ENSG00000101474	testis	1.36×10^{-7} (p-value)	GTEx Portal V6	59
APMAP	?	ENSG00000101474	transverse colon	1.12×10^{-7} (p-value)	GTEx Portal V6	50
CST7	?	ENSG00000077984	blood	4.25×10^{-19} (p-value)	GTEx Portal V6	61
APMAP	?	ENSG00000101474	blood	4.94×10^{-7} (p-value)	GTEx Portal V6	8
CST7	?	ENSG00000077984	lung	2.84×10^{-7} (p-value)	GTEx Portal V6	17
NINL	?	ENSG00000101004	subcutaneous adipocytes	1.06×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000291173	1	ENCP00000031115	RP5-1025A1.2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000399319 (CTCF binding site)	2	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	DNase1, H3K27me3, CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, Rad21, CTCF, H3K27me3
		lung (IMR90)	H3K27me3, CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF

		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1
		A549	CTCF, H3K27me3
ENSR00001664641 (CTCF binding site)	3	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		blood (DND-41)	H3K36me3
ENSR00001664642 (enhancer)	2	cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K4me1, H3K4me2
ENSR00001664643 (promoter flanking region)	2	embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	H3K27me3
		Osteobl	CTCF
		blood (K562)	H3K27me3, CTCF
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	CTCF, H3K27me3
		blood (GM12878)	CTCF
		A549	H3K27me3, CTCF
		skin (NHEK)	CTCF, H3K4me2, H3K4me1, DNase1
ENSR00000399320 (CTCF binding site)	5	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, H3K27me3
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF, H3K4me2, H3K4me1, DNase1
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	H3K4me1, CTCF
		breast (HMEC)	CTCF, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3
ENSR00001448170 (enhancer)	2	HSMMtube	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K4me1, H3K27ac
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K4me2
		cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF
		A549	H3K27me3, CTCF
		skin (NHEK)	CTCF, H3K4me2
ENSR00001448175 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, PolII, Rad21, Gabp, TAF7, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	HEY1, H2AZ, ELF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3, Max, H3K9ac, Gabp, Cmyc
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	DNase1, Gabp, PolII, H3K79me2, Rad21, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K27ac, Cmyc, H3K4me3, H3K27me3
		lung (IMR90)	DNase1, H4K8ac, H3K27ac, H3K4me2, H4K5ac, H3K4me3, H3K9ac, CTCF

		blood (GM12878)	H2AZ, DNase1, Cmyc, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, PolII, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac
		skin (NHEK)	CTCF, H3K9ac, H3K4me2, DNase1, H3K4me3
		NHLF	H3K27ac, H3K9ac, CTCF, H3K4me3, DNase1
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K9ac, DNase1, PolII, Gabp, H3K4me3, TAF1, H3K27ac, CTCF, H3K4me2
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K4me3, CTCF, DNase1
		endothelium (HUVEC)	Max, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1, H3K27ac
ENSR00000399325 e! (CTCF binding site)	3	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K4me2, H3K27me3, DNase1
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	H3K4me2, CTCF
		liver (HepG2)	Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3, DNase1
		blood (GM12878)	H3K9ac, DNase1, Pbx3, H2AZ, Yy1, Rad21, H3K4me2, CTCF
		lung (IMR90)	H4K8ac, H3K4me2, H4K5ac, H3K9ac, CTCF
		nervous (NH-A)	H3K4me2, CTCF, H3K9ac
		skin (NHEK)	CTCF, H3K4me3, H3K4me2, DNase1
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2, H3K27ac
		blood (DND-41)	H3K9ac, CTCF, H3K4me2, H3K4me1, H3K4me3
		breast (HMEC)	H3K4me3, H3K27ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K4me2
		monocytes (Monocytes-CD14+)	H3K9ac, DNase1, CTCF, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K4me2, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1, H3K27ac
ENSR00001664649 e! (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	CTCF, H3K27me3, DNase1
		blood (K562)	Rad21, CTCF, H3K27me3
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, H3K4me2, CTCF, DNase1
		lung (IMR90)	H3K4me2, CTCF
		blood (GM12878)	CTCF, Rad21, Yy1, Pbx3, DNase1
		nervous (NH-A)	H3K4me2, CTCF
		skin (NHEK)	CTCF, DNase1
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	CTCF, DNase1
		A549	H3K4me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
APMAP e!	downstream gene variant, upstream gene variant	154	ENST00000217456 e!	NM_020531.2	ENSP00000217456 e!	10
APMAP e!	downstream gene variant, upstream gene variant	491	ENST00000451442 e!	?	ENSP00000395874 e!	10
CST7 e!	upstream gene variant, downstream gene variant	32	ENST00000480798 e!	NM_003650.3	ENSP00000420384 e!	2
RNU6-1257P e!	downstream gene variant	4470	ENST00000384625 e!	?	?	1
RP4-568C11.4 e!	upstream gene variant, downstream gene variant	34	ENST00000620459 e!	?	?	14

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APMAP	ENST00000217456	NM_020531.2	ENSP00000217456	2
APMAP	ENST00000451442	?	ENSP00000395874	2
CST7	ENST00000480798	NM_003650.3	ENSP00000420384	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
APMAP	ENST00000217456	NM_020531.2	ENSP00000217456	1
APMAP	ENST00000451442	?	ENSP00000395874	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP4-568C11.4	ENST00000620459	?	3

Locus 71 (rs646776)

SNiPAcard

Block annotations

Block info

genomic range	chr1:109,817,192-109,822,166
block size	4,975 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.610 [-1.832 – 0.394]	**gene(s) hit or close-by**	CELSR2, PSRC1
phastCons	μ = 0.102 [0 – 0.992]	**eQTL gene(s)**	CELSR2, PSRC1, SORT1
GERP++	μ = -1.693 [-9 – 3.35]	**potentially regulated gene(s)**	AMPD2
CADD score	μ = 4.646 [0.153 – 13.51]	**disease gene(s)**	SORT1, AMPD2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Coronary Artery Disease	$<1.00\times10^{-8}$	GWAS Catalog	24262325	1
Coronary artery disease or large artery stroke	$<8.00\times10^{-17}$	GWAS Catalog	24262325	1
Coronary artery disease or ischemic stroke	$<1.00\times10^{-8}$	GWAS Catalog	24262325	1
Cholesterol, total	$<2.00\times10^{-170}$	GWAS Catalog	24097068	1
LDL cholesterol	$<5.00\times10^{-241}$	GWAS Catalog	24097068	1
Lipoprotein-associated phospholipase A2 activity and mass	$<2.00\times10^{-22}$	GWAS Catalog	23118302	1
Lipoprotein-associated phospholipase A2 activity and mass	$<1.00\times10^{-17}$	GWAS Catalog	22003152	1
Cardiovascular disease risk factors	$<2.00\times10^{-22}$	GWAS Catalog	21943158	1
Metabolite levels	$<2.00\times10^{-19}$	GWAS Catalog	21909109	1
Coronary heart disease	$<3.00\times10^{-10}$	GWAS Catalog	21378990	1
Coronary heart disease	$<6.00\times10^{-10}$	GWAS Catalog	21378988	1
LDL cholesterol	$<9.00\times10^{-29}$	GWAS Catalog	21347282	1
Coronary Artery Disease	$<8.00\times10^{-11}$	GWAS Catalog	21239051	1
Progranulin levels	$<2.00\times10^{-30}$	GWAS Catalog	21087763	1
LDL cholesterol	$<1.00\times10^{-26}$	GWAS Catalog	20864672	1
Cholesterol, total	$<6.00\times10^{-131}$	GWAS Catalog	20686565	1
LDL cholesterol	$<1.00\times10^{-170}$	GWAS Catalog	20686565	1
Lipoprotein-associated phospholipase A2 activity and mass	$<3.00\times10^{-15}$	GWAS Catalog	20442857	1
Response to statin therapy	$<4.00\times10^{-6}$	GWAS Catalog	20339536	1
Lipid metabolism phenotypes	$<2.00\times10^{-53}$	GWAS Catalog	19936222	1
Myocardial infarction (early onset)	$<8.00\times10^{-12}$	GWAS Catalog	19198609	1
Cholesterol, total	$<9.00\times10^{-22}$	GWAS Catalog	19060911	1
LDL cholesterol	$<8.00\times10^{-23}$	GWAS Catalog	19060911	1
LDL cholesterol	$<2.00\times10^{-12}$	GWAS Catalog	19060910	1
LDL cholesterol	$<2.00\times10^{-42}$	GWAS Catalog	19060906	1
LDL cholesterol	$<1.00\times10^{-33}$	GWAS Catalog	18262040	1

LDL cholesterol	$<3.00\times10^{-29}$	GWAS Catalog	18193044	1
LDL cholesterol	$<6.00\times10^{-33}$	GWAS Catalog	18193043	1
LDL cholesterol	$<1.00\times10^{-7}$	GWAS Catalog	18179892	1
Coronary heart disease	$<4.00\times10^{-9}$	GWAS Catalog	17634449	1
cholesterol	5.02×10^{-9}	Metabolomics GWAS Server	24816252	7
Cholesterol	2.68×10^{-5}	dbGaP	pha003078	1
Cholesterol	7.39×10^{-5}	dbGaP	pha003073	1
Lipoproteins, ldl	2.19×10^{-12}	dbGaP	pha002902	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Low density lipoprotein cholesterol level quantitative trait locus 6	association	ClinVar	RCV000007497.1	1
?	HGMD curated	HGMD	CR114612	1
?	HGMD curated	HGMD	CR105220	1
LOW DENSITY LIPOPROTEIN CHOLESTEROL LEVEL QUANTITATIVE TRAIT LOCUS 6	OMIM curated	OMIM	MIM:602458	1

Disease gene annotation

gene	trait	source DB	source entry/link
SORT1	LOW DENSITY LIPOPROTEIN CHOLESTEROL LEVEL QUANTITATIVE TRAIT [...]	OMIM	MIM:613589
AMPD2	PONTOCEREBELLAR HYPOPLASIA, TYPE 9	OMIM	MIM:615809
AMPD2	SPASTIC PARAPLEGIA 63, AUTOSOMAL RECESSIVE	OMIM	MIM:615686
AMPD2	Autosomal recessive spastic paraplegia type 63	OrphaNet	OrphaNet:401805
AMPD2	Pontocerebellar hypoplasia, type 9	OrphaNet	OrphaNet:369920

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PSRC1	?	ENSG00000134222	pancreas	2.83×10^{-7} (p-value)	GTEx Portal V6	10
SORT1	?	ENSG00000134243	pancreas	2.33×10^{-6} (p-value)	GTEx Portal V6	5
PSRC1	?	ENSG00000134222	blood	1.98×10^{-7} (p-value)	GTEx Portal V6	5
PSRC1	?	ENSG00000134222	esophagus mucosa	5.45×10^{-12} (p-value)	GTEx Portal V6	10
CELSR2	?	ENSG00000143126	esophagus mucosa	4.25×10^{-7} (p-value)	GTEx Portal V6	7
PSRC1	ENST00000369907	ILMN_1671843	monocyte	6.65×10^{-18} (p-value)	Fairfax et al.	4
PSRC1	ENST00000369909					
PSRC1	ENST00000369904					
PSRC1	ENST00000409138					
PSRC1	ENST00000409267					
PSRC1	ENST00000492431					
PSRC1	ENST00000369903					
CELSR2	?	ENSG00000143126	skeletal muscle	7.42×10^{-36} (p-value)	GTEx Portal V6	10
PSRC1	?	ENSG00000134222	testis	3.79×10^{-8} (p-value)	GTEx Portal V6	10
SORT1	?	ENSG00000134243	liver	3.42×10^{-27} (p-value)	GTEx Portal V6	10
PSRC1	?	ENSG00000134222	liver	4.39×10^{-19} (p-value)	GTEx Portal V6	10

CELSR2	?	ENSG00000143126	liver	3.42×10-19 (p-value)	GTEx Portal V6	10
CELSR2	?	ENSG00000143126	sun exposed skin	7.33×10-6 (p-value)	GTEx Portal V6	2
CELSR2	ENST00000498157	ILMN_1711208	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
CELSR2	ENST00000271332					
SORT1	ENST00000538502	ILMN_1707077	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
SORT1	ENST00000256637					
PSRC1	?	ENSG00000134222	cortex	8.68×10-7 (p-value)	GTEx Portal V6	6
PSRC1	?	ENSG00000134222	unexposed skin	2.73×10-6 (p-value)	GTEx Portal V6	3
PSRC1	ENST00000369907	ILMN_1671843	monocyte	7.21×10-61 (p-value)	Zeller et al.	2
PSRC1	ENST00000369909					
PSRC1	ENST00000369904					
PSRC1	ENST00000409138					
PSRC1	ENST00000409267					
PSRC1	ENST00000369903					
PSRC1	ENST00000492431					
PSRC1	ENST00000369909	ILMN_2315964	monocyte	3.05×10-13 (p-value)	Zeller et al.	2
PSRC1	ENST00000369904					
PSRC1	ENST00000409267					
PSRC1	ENST00000369907					
PSRC1	ENST00000409138					
PSRC1	ENST00000474126					
PSRC1	ENST00000492431					
PSRC1	ENST00000369903					
PSRC1	?	ENSG00000134222	thyroid	1.44×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000031690	1	ENCP00000003684	AMPD2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001523273 (promoter flanking region)	5	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF, H3K36me3, DNase1, H3K4me1, MEF2A, Max, PU1, H3K9ac
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF, H3K36me3, DNase1
		liver (HepG2)	PolII, Rad21, H3K4me1, H3K4me2, FOXA1, H3K27ac, CTCF, Cmyc, DNase1
		lung (IMR90)	H3K36me3, CTCF
		blood (GM12878)	DNase1, H2AZ, Rad21, CTCF
		nervous (NH-A)	CTCF, H3K4me1, H3K4me2, DNase1
		skin (NHEK)	DNase1, H3K36me3, CTCF
		NHLF	DNase1, CTCF
		Osteobl	H3K4me2, CTCF, H3K36me3
		blood (DND-41)	CTCF, H3K36me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K4me1, Max, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, Max
		A549	DNase1, H3K27ac, H3K36me3, CTCF, H3K4me2, H3K4me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
CELSR2	1	ENST00000271332 ENST00000498157	hsa-miR-4319

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CELSR2	downstream gene variant	158	ENST00000271332	NM_001408.2	ENSP00000271332	6
CELSR2	downstream gene variant	261	ENST00000489018	?	?	8
CELSR2	downstream gene variant	77	ENST00000498157	?	?	7
PSRC1	downstream gene variant	1611	ENST00000418914	?	ENSP00000413186	5
PSRC1	downstream gene variant	13	ENST00000369909	NM_001032291.2, NM_032636.7	ENSP00000358925	10
PSRC1	downstream gene variant	1442	ENST00000429031	?	ENSP00000387791	5
PSRC1	downstream gene variant	12	ENST00000369904	NM_001005290.3	ENSP00000358920	10
PSRC1	downstream gene variant	12	ENST00000369907	?	ENSP00000358923	10
PSRC1	downstream gene variant	21	ENST00000409138	?	ENSP00000474667	10
PSRC1	downstream gene variant	12	ENST00000409267	?	ENSP00000386323	10
PSRC1	downstream gene variant	1265	ENST00000474126	?	ENSP00000474682	6
PSRC1	downstream gene variant	23	ENST00000492431	?	?	10
PSRC1	downstream gene variant	2075	ENST00000471740	?	?	5
PSRC1	downstream gene variant	41	ENST00000369903	?	ENSP00000358919	9
PSRC1	downstream gene variant	1860	ENST00000459765	?	?	5

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CELSR2	ENST00000271332	NM_001408.2	ENSP00000271332	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CELSR2	ENST00000498157	?	3

Locus 72 (rs10494745)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,887,457-197,127,578
block size	240,122 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.197 [-1.519 – 0.704]	gene(s) hit or close-by	ASPM, CFHR2, CFHR4, ZBTB41
phastCons	μ = 0.001 [0 – 0.003]	eQTL gene(s)	–
GERP++	μ = 0.497 [-4.16 – 3.37]	potentially regulated gene(s)	–
CADD score	μ = 5.015 [0.698 – 8.621]	disease gene(s)	ASPM

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
ASPM	MICROCEPHALY 5, PRIMARY, AUTOSOMAL RECESSIVE	OMIM	MIM:608716	OMIM
ASPM	Primary Autosomal Recessive Microcephaly	DECIPHER	MIM:608716	OMIM
ASPM	Autosomal recessive primary microcephaly	OrphaNet	OrphaNet:2512	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CFHR4	missense variant	ENST00000251424	NM_006684.4	ENSP00000251424	E/G	gAa/gGa	?	?	1
CFHR4	missense variant	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	E/G	gAa/gGa	?	?	1
CFHR4	missense variant	ENST00000367418	?	ENSP00000356388	E/G	gAa/gGa	?	?	1
CFHR4	missense variant	ENST00000608469	?	ENSP00000477162	E/G	gAa/gGa	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ASPM	ENST00000612785	?	ENSP00000479244	1
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	1
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	1
ASPM	ENST00000367408	?	ENSP00000356378	1
CFHR2	ENST00000367421	?	ENSP00000356391	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ZBTB41	ENST00000367405	NM_194314.2	ENSP00000356375	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ZBTB41	ENST00000467322	?	1

Locus 73 (rs947409)

SNiPAcard

Block annotations

Block info

genomic range	chr13:47,341,767-47,391,824
block size	50,058 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.443 [-1.875 – 0.869]	gene(s) hit or close-by	ESD
phastCons	μ = 0.008 [0 – 0.077]	eQTL gene(s)	–
GERP++	μ = -0.219 [-3.58 – 4.31]	potentially regulated gene(s)	–
CADD score	μ = 3.585 [0.087 – 11.38]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Iron	3.02×10-4	dbGaP	pha002876 dbGaP	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1720285	liver	1.26×10-12 (p-value)	Innocenti et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001509548 (CTCF binding site)	1	NHLF	H3K36me3, DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
ENSR00001509551 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		breast (HMEC)	DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	DNase1

		nervous (NH-A)	DNase1, H3K27ac	
		skin (NHEK)	DNase1	
ENSR00001509552 (enhancer)	1	blood (DND-41)	H3K27me3	
		muscle (HSMM)	DNase1	

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ESD	downstream gene variant, upstream gene variant	395	ENST00000412582	?	ENSP00000391350	6
ESD	downstream gene variant, upstream gene variant	1856	ENST00000378697	?	ENSP00000367969	5
ESD	downstream gene variant, upstream gene variant	1777	ENST00000378720	NM_001984.1	ENSP00000367992	6
ESD	downstream gene variant, upstream gene variant	398	ENST00000471867	?	ENSP00000476193	3
ESD	downstream gene variant	1110	ENST00000495654	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ESD	ENST00000412582	?	ENSP00000391350	6
ESD	ENST00000378697	?	ENSP00000367969	15
ESD	ENST00000378720	NM_001984.1	ENSP00000367992	15
ESD	ENST00000471867	?	ENSP00000476193	5
ESD	ENST00000495654	?	?	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ESD	ENST00000471867	?	ENSP00000476193	2

Locus 74 (rs4986791)

SNiPAcard

Block annotations

Block info

genomic range	chr9:120,467,576-120,475,602
block size	8,027 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.319 [-0.896 – 2.147]	gene(s) hit or close-by	RNU6-1082P, TLR4
phastCons	μ = 0.011 [0 – 0.051]	eQTL gene(s)	BRINP1, TLR4
GERP++	μ = -0.045 [-5.56 – 4.64]	potentially regulated gene(s)	–
CADD score	μ = 5.743 [1.266 – 12.31]	disease gene(s)	TLR4

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	not provided	ClinVar	RCV000086999.1	1
Endotoxin hyporesponsiveness	pathogenic	ClinVar	RCV000007043.1	1
Colorectal cancer, susceptibility to	risk factor	ClinVar	RCV000007042.1	1
Macular degeneration, age-related, 10, susceptibility to	risk factor	ClinVar	RCV000007041.1	1
Endotoxin hyporesponsiveness	pathogenic	ClinVar	RCV000007040.1	1
?	HGMD curated	HGMD	CM001362	1
?	HGMD curated	HGMD	CM001361	1
ENDOTOXIN HYPORESPONSIVENESS	OMIM curated	OMIM	MIM:603030	2

Disease gene annotation

gene	trait	source DB	source entry/link
TLR4	TOLL-LIKE RECEPTOR 4	OMIM	MIM:603030
TLR4	MACULAR DEGENERATION, AGE-RELATED, 10	OMIM	MIM:611488
TLR4	Behcet disease	OrphaNet	OrphaNet:117

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
TLR4	missense variant	ENST00000355622	NM_138554.4	ENSP00000363089	2	2			2
TLR4	missense variant	ENST00000394487	NM_003266.3	ENSP00000377997	2	2			2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TLR4	ENST00000394487	ILMN_1706217	blood	6.25×10^{-6} (p-value)	Westra et al.	6

TLR4	ENST00000355622						
TLR4	ENST00000472304						
TLR4	ENST00000394487	ILMN_1706217	blood	7.20×10^{-5} (p-value)	MuTHER consortium	2	
TLR4	ENST00000355622						
TLR4	ENST00000472304						
BRINP1	ENST00000265922	ILMN_1741603	monocyte	7.17×10^{-5} (p-value)	Fairfax et al.	1	

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001312883 (promoter)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K4me2, H3K9ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		breast (HMEC)	H3K4me3, H3K4me2
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	PolII, Cmyc, H3K27ac, H3K9ac, H3K4me3, Cjun, DNase1, H3K4me2
		liver (HepG2)	DNase1
		blood (GM12878)	PU1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RNU6-1082P	downstream gene variant, upstream gene variant	724	ENST00000364574	?	?	6
TLR4	upstream gene variant, downstream gene variant	103	ENST00000490685	?	?	8

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TLR4	ENST00000355622	NM_138554.4	ENSP00000363089	6
TLR4	ENST00000472304	?	?	6
TLR4	ENST00000394487	NM_003266.3	ENSP00000377997	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RNU6-1082P	ENST00000364574	?	1
TLR4	ENST00000472304	?	3

Locus 75 (rs8103941)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,540,143-51,546,893
block size	6,751 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.622 [-3.722 – 0.941]	**gene(s) hit or close-by**	CTC-518B2.10, CTC-518B2.9, KLK12
phastCons	μ = 0.031 [0 – 0.142]	**eQTL gene(s)**	CTC-518B2.9, KLK12
GERP++	μ = -0.100 [-1.91 – 0.889]	**potentially regulated gene(s)**	–
CADD score	μ = 2.159 [0.022 – 12.37]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
dihomo-linolenate (20:3n3 or n6)	6.95×10^{-5}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CTC-518B2.9	?	ENSG00000268906	esophagus mucosa	5.39×10^{-13} (p-value)	GTEx Portal V6	16
KLK12	?	ENSG00000186474	esophagus mucosa	1.36×10^{-16} (p-value)	GTEx Portal V6	16
KLK12	?	ENSG00000186474	terminal ileum	1.45×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTC-518B2.10	upstream gene variant	170	ENST00000601280	?	?	11
CTC-518B2.9	downstream gene variant	2551	ENST00000594910	?	?	6
KLK12	upstream gene variant	2024	ENST00000530943	?	ENSP00000434339	6
KLK12	upstream gene variant	2024	ENST00000529888	NM_145895.1	ENSP00000434036	6
KLK12	upstream gene variant	1994	ENST00000250351	NM_019598.2	ENSP00000250351	6
KLK12	upstream gene variant	1994	ENST00000319590	NM_145894.1	ENSP00000324181	6
KLK12	upstream gene variant	2024	ENST00000531374	?	ENSP00000436121	6
KLK12	upstream gene variant	1656	ENST00000526824	?	ENSP00000434604	6
KLK12	upstream gene variant	2145	ENST00000525263	?	ENSP00000436458	6

Locus 76 (rs10801582)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,845,273-196,944,357
block size	99,085 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.006 [-1.277 – 0.754]	**gene(s) hit or close-by**	CFHR2, CFHR2, CFHR4, CFHR5, RP4-608O15.4
phastCons	μ = 0.095 [0 – 0.492]	**eQTL gene(s)**	CFH, CFHR1, CFHR3, CFHR4
GERP++	μ = -0.192 [-3.63 – 1.43]	**potentially regulated gene(s)**	–
CADD score	μ = 4.973 [0.282 – 13.66]	**disease gene(s)**	CFHR1, CFHR3, CFH, CFHR5

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
End-stage coagulation	$<7.00\times10^{-19}$	GWAS Catalog	23381943	1
Macular degeneration	5.49×10^{-30}	dbGaP	pha002890	1
Macular degeneration	5.91×10^{-34}	dbGaP	pha002869	1
Macular degeneration	6.21×10^{-5}	dbGaP	pha002856	1
Macular degeneration	2.51×10^{-6}	dbGaP	pha000001	2

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR1	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFHR1	C3 glomerulonephritis	OrphaNet	OrphaNet:329931
CFHR1	Dense deposit disease	OrphaNet	OrphaNet:93571
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571
CFH	Familial drusen	OrphaNet	OrphaNet:75376
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903
CFHR5	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFHR5	C3 glomerulonephritis	OrphaNet	OrphaNet:329931
CFHR5	CFHR5 DEFICIENCY	OMIM	MIM:614809

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	lung	4.99×10^{-6} (p-value)	GTEx Portal V6	1
CFHR1	?	ENSG00000244414	tibial nerve	1.40×10^{-6} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	tibial nerve	9.52×10^{-11} (p-value)	GTEx Portal V6	12
CFH	?	ENSG00000000971	tibial nerve	2.60×10^{-6} (p-value)	GTEx Portal V6	13

CFHR4	?	ENSG00000134365	liver	6.84×10-8 (p-value)	GTEx Portal V6	16
CFHR1	?	ENSG00000244414	subcutaneous adipocytes	1.65×10-8 (p-value)	GTEx Portal V6	16
CFHR3	?	ENSG00000116785	subcutaneous adipocytes	7.04×10-11 (p-value)	GTEx Portal V6	16
CFHR3	?	ENSG00000116785	thyroid	4.81×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFHR2	downstream gene variant	965	ENST00000473386	?	?	1
CFHR2	downstream gene variant	970	ENST00000367421	?	ENSP00000356391	1
CFHR2	downstream gene variant	954	ENST00000476712	?	ENSP00000476677	1
CFHR2	downstream gene variant	79	ENST00000489703	?	?	3
CFHR2	downstream gene variant	954	ENST00000367415	NM_005666.2	ENSP00000356385	1
CFHR2	downstream gene variant	142	ENST00000485647	?	?	2
CFHR2	downstream gene variant	961	ENST00000496448	?	?	1
CFHR4	upstream gene variant	1523	ENST00000251424	NM_006684.4	ENSP00000251424	1
CFHR4	upstream gene variant	1455	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	1
CFHR4	upstream gene variant	1544	ENST00000367418	?	ENSP00000356388	1
CFHR5	upstream gene variant	2329	ENST00000256785	NM_030787.3	ENSP00000256785	1
RP4-608O15.4	downstream gene variant	4556	ENST00000613613	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CFHR2	ENST00000367421	?	ENSP00000356391	C	tgT/tgC	1
CFHR2	ENST00000367415	NM_005666.2	ENSP00000356385	C	tgT/tgC	1
CFHR2	ENST00000476712	?	ENSP00000476677	C	tgT/tgC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFHR2	ENST00000473386	?	?	4
CFHR2	ENST00000367421	?	ENSP00000356391	16
CFHR2	ENST00000485647	?	?	2
CFHR2	ENST00000367415	NM_005666.2	ENSP00000356385	4
CFHR2	ENST00000489703	?	?	1
CFHR2	ENST00000476712	?	ENSP00000476677	4
CFHR2	ENST00000496448	?	?	4
CFHR4	ENST00000251424	NM_006684.4	ENSP00000251424	6
CFHR4	ENST00000608469	?	ENSP00000477162	8

CFHR4	ENST00000367418	?	ENSP00000356388	6
CFHR4	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CFHR2	ENST00000485647	?	1
CFHR2	ENST00000496448	?	1

Locus 77 (rs1250258)

SNiPAcard

Block annotations

Block info

genomic range	chr2:216,287,093-216,306,635
block size	19,543 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.022 [-1.29 – 3.387]	gene(s) hit or close-by	AC012462.1, FN1
phastCons	μ = 0.133 [0 – 1]	eQTL gene(s)	FN1
GERP++	μ = -0.320 [-7.88 – 5.35]	potentially regulated gene(s)	AC093850.2, FN1
CADD score	μ = 5.604 [0.051 – 22.4]	disease gene(s)	FN1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
LDL cholesterol	<3.00×10-8	GWAS Catalog	24097068	1

Disease gene annotation

gene	trait	source DB	source entry/link
FN1	GLOMERULOPATHY WITH FIBRONECTIN DEPOSITS 2	OMIM	MIM:601894
FN1	Fibronectin glomerulopathy	OrphaNet	OrphaNet:84090

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FN1	missense variant	ENST00000359671	?	ENSP00000352696	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000432072	?	ENSP00000399538	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000357867	NM_212474.1	ENSP00000350534	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000421182	?	ENSP00000394423	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000446046	NM_212478.1	ENSP00000410422	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000443816	?	ENSP00000415018	Q/L	cAg/cTg	tolerated	unknown	1
FN1	missense variant	ENST00000336916	NM_002026.2	ENSP00000338200	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000354785	NM_212482.1	ENSP00000346839	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000323926	?	ENSP00000323534	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000426059	NM_054034.2	ENSP00000398907	Q/L	cAg/cTg	?	benign	1
FN1	missense variant	ENST00000356005	NM_212476.1	ENSP00000348285	Q/L	cAg/cTg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FN1	ENST00000426059	ILMN_1675646	b-cell	1.72×10^{-5} (p-value)	Fairfax et al.	1
?	?	ILMN_1673991	monocyte	5.73×10^{-13} (p-value)	Zeller et al.	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000529793	1	ENST00000357867, ENST00000421182, ENST00000432072, ENST00000443816, ENST00000354785, ENST00000426059, ENST00000336916, ENST00000359671, ENST00000323926, ENST00000446046, ENST00000356005	FN1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000277616	2	ENCP00000029547	FN1
		ENCP00000029544	FN1
		ENCP00000029545	FN1
		ENCP00000029538	FN1
		ENCP00000029543	FN1
		ENCP00000029540	FN1
		ENCP00000029542	FN1
		ENCP00000029541	FN1
ENCE00000277620	1	ENCP00000029547	FN1
		ENCP00000029544	FN1
		ENCP00000029538	FN1
		ENCP00000029545	FN1
		ENCP00000029543	FN1
		ENCP00000029540	FN1
		ENCP00000029542	FN1
		ENCP00000029558	AC093850.2
		ENCP00000029541	FN1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001551486 (promoter flanking region)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (K562)	DNase1
		skin (NHDF-AD)	H3K36me3
		breast (HMEC)	H3K36me3
		muscle (HSMM)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	Max, PolII, H3K36me3
		liver (HepG2)	PolII, H3K4me1, H3K4me2, H3K27ac, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3, H3K4me2, H4K20me1, H3K9ac
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000607008 (promoter)	4	embryonic stem cell (H1ESC)	ATF3, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1, Rad21, PolII, H3K36me3, H3K27me3
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K36me3, DNase1
		blood (K562)	H2AZ, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, H3K36me3
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me3, H3K27ac, H3K4me2

		liver (HepG2)	TAF1, H3K4me1, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K4me3, H3K27me3, H3K36me3, DNase1, H3K79me2, PolII, Jund
		blood (GM12878)	DNase1, H2AZ, H3K4me3, H3K4me2
		lung (IMR90)	DNase1, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me3, H3K4me2, H4K20me1, H3K9ac
		skin (NHEK)	H3K27me3, H3K36me3, H3K4me3, H3K4me2, H3K9ac, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K4me3, H3K4me2, H3K36me3, H3K27ac
		blood (DND-41)	H3K27me3, H3K4me3, H3K4me1
		breast (HMEC)	H3K36me3, H3K4me2, H3K9ac, H3K4me3, H3K27ac
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K27ac, PolII, DNase1, H3K36me3, H3K9ac, H3K4me3, H3K4me2, Cjun
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3, H3K27me3
ENSR00001661218 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K4me1
		skin (NHDF-AD)	DNase1
ENSR00001551491 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	Rad21
		blood (K562)	H2AZ
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K4me1, H2AZ, H3K4me2
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC012462.1	upstream gene variant, downstream gene variant	494	ENST00000412951	?	?	9
FN1	upstream gene variant	2257	ENST00000426059	NM_054034.2	ENSP00000398907	2
FN1	upstream gene variant	2262	ENST00000446046	NM_212478.1	ENSP00000410422	2
FN1	upstream gene variant	2262	ENST00000357867	NM_212474.1	ENSP00000350534	2
FN1	upstream gene variant	2262	ENST00000359671	?	ENSP00000352696	2
FN1	upstream gene variant	2263	ENST00000432072	?	ENSP00000399538	2
FN1	upstream gene variant	1100	ENST00000496542	?	?	2
FN1	upstream gene variant	2263	ENST00000421182	?	ENSP00000394423	2
FN1	upstream gene variant	2262	ENST00000336916	NM_002026.2	ENSP00000338200	2
FN1	upstream gene variant	2257	ENST00000443816	?	ENSP00000415018	2
FN1	upstream gene variant	2262	ENST00000323926	?	ENSP00000323534	2
FN1	upstream gene variant	2158	ENST00000354785	NM_212482.1	ENSP00000346839	3
FN1	upstream gene variant	2262	ENST00000356005	NM_212476.1	ENSP00000348285	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FN1	ENST00000359671	?	ENSP00000352696	10
FN1	ENST00000432072	?	ENSP00000399538	10
FN1	ENST00000357867	NM_212474.1	ENSP00000350534	10
FN1	ENST00000421182	?	ENSP00000394423	10

FN1	ENST00000323926	?	ENSP00000323534	10
FN1	ENST00000443816	?	ENSP00000415018	10
FN1	ENST00000426059	NM_054034.2	ENSP00000398907	10
FN1	ENST00000354785	NM_212482.1	ENSP00000346839	10
FN1	ENST00000446046	NM_212478.1	ENSP00000410422	10
FN1	ENST00000336916	NM_002026.2	ENSP00000338200	10
FN1	ENST00000356005	NM_212476.1	ENSP00000348285	10

Locus 79 (rs7091871)

SNiPAcard

Block annotations

Block info

genomic range	chr10:101,798,405-101,814,910
block size	16,506 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.652 [-2.147 – 0.21]	gene(s) hit or close-by	CPN1
phastCons	μ = 0.019 [0 – 0.152]	eQTL gene(s)	HIF1AN
GERP++	μ = 0.007 [-2.16 – 1.68]	potentially regulated gene(s)	–
CADD score	μ = 2.581 [0.293 – 6.817]	disease gene(s)	CPN1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CPN1	CARBOXYPEPTIDASE N DEFICIENCY	OMIM	MIM:212070

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HIF1AN	?	ENSG00000166135	transverse colon	4.80×10^{-6} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001426512 (enhancer)	1	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me1, H3K4me2, HNF4A, H3K27ac
		blood (GM12878)	H2AZ, H3K4me1
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001426513 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K36me3, Rad21, CTCF
		lung (IMR90)	CTCF, DNase1
		blood (GM12878)	DNase1, H2AZ, Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	CTCF, DNase1

		endothelium (HUVEC)	H3K36me3, CTCF
		A549	DNase1, CTCF
ENSR00000360384 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	CTCF, H3K36me3, Rad21
		blood (GM12878)	DNase1, H2AZ, Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF, H3K4me2, H3K9ac, DNase1, H3K27ac
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF, H3K27ac
		cervix (HeLa-S3)	H3K27ac, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CPN1	downstream gene variant	2217	ENST00000370418	NM_001308.2	ENSP00000359446	2
CPN1	downstream gene variant	2062	ENST00000441382	?	ENSP00000410895	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPN1	ENST00000370418	NM_001308.2	ENSP00000359446	9
CPN1	ENST00000441382	?	ENSP00000410895	7

Locus 80 (rs600324)

SNiPAcard

Block annotations

Block info

genomic range	chr6:160,519,276-160,519,276
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.576	gene(s) hit or close-by	IGF2R, RP11-288H12.3
phastCons	0	eQTL gene(s)	–
GERP++	-2.07	potentially regulated gene(s)	–
CADD score	0.968	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Lipoproteins	4.40×10-5	dbGaP	pha003079 dbGaP	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-288H12.3	downstream gene variant	2032	ENST00000569097	?	?	1
RP11-288H12.3	downstream gene variant	4251	ENST00000366273	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IGF2R	ENST00000356956	NM_000876.2	ENSP00000349437	1
IGF2R	ENST00000475584	?	?	1

Locus 81 (rs882732)

SNiPAcard

Block annotations

Block info

genomic range	chr14:95,022,686-95,040,431
block size	17,746 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.617 [-2.753 – 1.541]	gene(s) hit or close-by	SERPINA4, SERPINA5
phastCons	μ = 0.049 [0 – 0.979]	eQTL gene(s)	SERPINA3, SERPINA3, SERPINA5
GERP++	μ = -1.273 [-6.99 – 2.58]	potentially regulated gene(s)	–
CADD score	μ = 2.503 [0.079 – 9.785]	disease gene(s)	SERPINA3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SERPINA3	Alpha-1-antichymotrypsin deficiency	OrphaNet	OrphaNet:93594

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SERPINA5	?	ENSG00000188488	tibial artery	1.44×10^{-9} (p-value)	GTEx Portal V6	20
SERPINA3	ENST00000467132	ILMN_1788874	adipocyte	2.28×10^{-13} (p-value)	MuTHER consortium	15
SERPINA3	ENST00000393080					
SERPINA3	ENST00000556968					
SERPINA3	ENST00000553947					
SERPINA3	ENST00000482740					
SERPINA3	ENST00000393078					
SERPINA5	?	ENSG00000188488	skeletal muscle	1.75×10^{-8} (p-value)	GTEx Portal V6	10
SERPINA5	?	ENSG00000188488	thyroid	1.90×10^{-6} (p-value)	GTEx Portal V6	6

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001459927 (enhancer)	3	liver (HepG2)	H3K27ac, H3K4me2, H3K4me1
		Osteobl	H2AZ, H3K27ac
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SERPINA4	upstream gene variant, downstream gene variant	57	ENST00000555095	NM_001289032.1	ENSP00000451172	8
SERPINA4	upstream gene variant, downstream gene variant	840	ENST00000557004	NM_006215.3	ENSP00000450838	8
SERPINA4	upstream gene variant, downstream gene variant	62	ENST00000298841	NM_001289033.1	ENSP00000298841	8

SERPINA5	upstream gene variant	57	ENST00000554220	?	ENSP00000450484	3
SERPINA5	upstream gene variant	57	ENST00000553780	?	ENSP00000450837	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
SERPINA4	ENST00000555095	NM_001289032.1	ENSP00000451172	F	ttT/ttC	1
SERPINA4	ENST00000557004	NM_006215.3	ENSP00000450838	F	ttT/ttC	1
SERPINA4	ENST00000298841	NM_001289033.1	ENSP00000298841	F	ttT/ttC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPINA4	ENST00000555095	NM_001289032.1	ENSP00000451172	12
SERPINA4	ENST00000557004	NM_006215.3	ENSP00000450838	12
SERPINA4	ENST00000298841	NM_001289033.1	ENSP00000298841	12
SERPINA5	ENST00000554220	?	ENSP00000450484	17
SERPINA5	ENST00000553780	?	ENSP00000450837	17

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPINA4	ENST00000557004	NM_006215.3	ENSP00000450838	1

Locus 82 (rs2239908)

SNiPAcard

Block annotations

Block info

genomic range	chr17:26,703,682-26,740,139
block size	36,458 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.006 [-1.561 – 1.574]	gene(s) hit or close-by	CTB-96E2.10, CTB-96E2.3, CTB-96E2.6, CTD-2350C19.2, H3F3BP2, SARM1, SLC46A1, VTN
phastCons	μ = 0.067 [0 – 0.654]	eQTL gene(s)	CTB-96E2.3, CTB-96E2.7, POLDIP2, SARM1, TMEM199, TMEM97
GERP++	μ = -0.561 [-6.27 – 2.35]	potentially regulated gene(s)	–
CADD score	μ = 5.301 [0.45 – 10.68]	disease gene(s)	SLC46A1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SLC46A1	hereditary folate malabsorption (HFM)	DECIPHER	MIM:229050
SLC46A1	FOLATE MALABSORPTION, HEREDITARY	OMIM	MIM:229050
SLC46A1	Hereditary folate malabsorption	OrphaNet	OrphaNet:90045

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TMEM199	?	ENSG00000244045	muscularis mucosae	1.58×10^{-7} (p-value)	GTEx Portal V6	15
POLDIP2	?	ENSG00000004142	nucleus accumbens	10.00×10^{-7} (p-value)	GTEx Portal V6	6
TMEM199	?	ENSG00000244045	lung	6.65×10^{-7} (p-value)	GTEx Portal V6	7
CTB-96E2.3	?	ENSG00000258924	lung	1.90×10^{-8} (p-value)	GTEx Portal V6	17
POLDIP2	?	ENSG00000004142	cortex	1.22×10^{-6} (p-value)	GTEx Portal V6	9
CTB-96E2.7	?	ENSG00000265618	tibial nerve	1.77×10^{-5} (p-value)	GTEx Portal V6	6
TMEM199	?	ENSG00000244045	tibial nerve	1.23×10^{-12} (p-value)	GTEx Portal V6	17
POLDIP2	?	ENSG00000004142	tibial nerve	2.60×10^{-10} (p-value)	GTEx Portal V6	18
TMEM199	?	ENSG00000244045	anterior cingulate cortex	2.17×10^{-6} (p-value)	GTEx Portal V6	1
POLDIP2	?	ENSG00000004142	putamen	7.53×10^{-6} (p-value)	GTEx Portal V6	2
TMEM199	?	ENSG00000244045	unexposed skin	4.06×10^{-6} (p-value)	GTEx Portal V6	6
TMEM199	?	ENSG00000244045	sun exposed skin	4.21×10^{-6} (p-value)	GTEx Portal V6	2
TMEM199	?	ENSG00000244045	subcutaneous adipocytes	9.44×10^{-8} (p-value)	GTEx Portal V6	11
POLDIP2	?	ENSG00000004142	subcutaneous adipocytes	1.92×10^{-5} (p-value)	GTEx Portal V6	6

TMEM199	?	ENSG00000244045	EBV lymphocytes	6.74×10-6 (p-value)	GTEx Portal V6	1
SARM1	ENST00000585482	ILMN_1746265	skin	2.09×10-6 (p-value)	MuTHER consortium	7
			blood	5.04×10-12 (p-value)	MuTHER consortium	7
			adipocyte	1.76×10-12 (p-value)	MuTHER consortium	7
TMEM97	ENST00000226230	ILMN_1710962	blood	1.67×10-14 (p-value)	MuTHER consortium	7
TMEM97	ENST00000336687		adipocyte	2.30×10-14 (p-value)	MuTHER consortium	7
TMEM97	ENST00000582384					
POLDIP2	ENST00000540200	ILMN_1743499	blood	7.99×10-5 (p-value)	Westra et al.	1
POLDIP2	?	ENSG00000004142	testis	9.09×10-6 (p-value)	GTEx Portal V6	4
TMEM97	?	ENSG00000109084	blood	2.78×10-3 (q-value)	SeeQTL DB (HapMap)	1
SARM1	ENST00000585482	ILMN_1746265	monocyte	1.61×10-16 (p-value)	Zeller et al.	1
TMEM97	ENST00000226230	ILMN_1710962	monocyte	4.23×10-36 (p-value)	Zeller et al.	1
TMEM97	ENST00000336687					
TMEM97	ENST00000582384					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
MAX	MA0058.2	6	yes	-0.145	1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001343354 (promoter flanking region)	3	cervix (HeLa-S3)	DNase1, H3K4me1
		NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun
		liver (HepG2)	PolII
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00001343356 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K27me3
		liver (HepG2)	PolII, H3K4me1
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTB-96E2.10	downstream gene variant	3101	ENST00000613598	?	?	1
CTB-96E2.6	upstream gene variant	2362	ENST00000623773	?	?	1
CTD-2350C19.2	downstream gene variant	4074	ENST00000580714	?	?	1
H3F3BP2	upstream gene variant	3801	ENST00000583907	?	?	1
SARM1	downstream gene variant	1408	ENST00000582323	?	ENSP00000464166	5
SARM1	downstream gene variant	396	ENST00000577870	?	?	4
SARM1	downstream gene variant	86	ENST00000585453	?	?	2
SARM1	downstream gene variant	183	ENST00000578128	?	ENSP00000462479	4
SARM1	downstream gene variant	616	ENST00000580711	?	?	4

SARM1	downstream gene variant	616	ENST00000580711	?	?	4
SARM1	downstream gene variant	2631	ENST00000585482	NM_015077.3	ENSP00000468032	1
SARM1	downstream gene variant	1429	ENST00000579593	?	ENSP00000462228	2
SARM1	downstream gene variant	114	ENST00000379061	?	?	2
SLC46A1	downstream gene variant, upstream gene variant	470	ENST00000612814	NM_080669.5	ENSP00000480703	9
SLC46A1	upstream gene variant	50	ENST00000584995	?	ENSP00000464190	2
SLC46A1	upstream gene variant	933	ENST00000578217	?	?	1
SLC46A1	downstream gene variant, upstream gene variant	50	ENST00000618626	NM_001242366.2	ENSP00000483652	9
SLC46A1	downstream gene variant, upstream gene variant	618	ENST00000582735	?	ENSP00000463339	9
SLC46A1	upstream gene variant	81	ENST00000584426	?	ENSP00000467416	2
SLC46A1	downstream gene variant	1790	ENST00000619923	?	?	6
SLC46A1	upstream gene variant	3673	ENST00000581516	?	ENSP00000462942	1
SLC46A1	upstream gene variant	512	ENST00000582590	?	?	2
VTN	upstream gene variant	3572	ENST00000542029	?	ENSP00000440439	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTB-96E2.3	ENST00000591482	?	?	1
SARM1	ENST00000585453	?	?	13
SARM1	ENST00000578128	?	ENSP00000462479	11
SARM1	ENST00000585482	NM_015077.3	ENSP00000468032	14
SARM1	ENST00000379061	?	?	14
SARM1	ENST00000579593	?	ENSP00000462228	13
SARM1	ENST00000577870	?	?	11
SARM1	ENST00000580711	?	?	11
SLC46A1	ENST00000581516	?	ENSP00000462942	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SARM1	ENST00000585482	NM_015077.3	ENSP00000468032	8
SARM1	ENST00000579593	?	ENSP00000462228	6
SLC46A1	ENST00000618626	NM_001242366.2	ENSP00000483652	5
SLC46A1	ENST00000612814	NM_080669.5	ENSP00000480703	8

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTD-2350C19.2	ENST00000580714	?	1
SARM1	ENST00000585453	?	2

Locus 83 (rs4859571)

SNiPAcard

Block annotations

Block info

genomic range	chr4:76,852,839-76,985,488
block size	132,650 bp
variant count	62 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.292 [-4.482 – 2.568]	gene(s) hit or close-by	AC110615.1, AC112719.2, ART3, CXCL11, CXCL9, NAAA, RP11-630D6.1, RP11-630D6.5, SDAD1
phastCons	μ = 0.119 [0 – 1]	eQTL gene(s)	ART3, CXCL10, CXCL11, CXCL9, NAAA, SCARB2
GERP++	μ = -0.578 [-10.3 – 4.28]	potentially regulated gene(s)	CCDC158, NAAA, NUP54, USO1
CADD score	μ = 4.078 [0.03 – 18.35]	disease gene(s)	SCARB2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Heart rate	1.52×10^{-5}	dbGaP	pha001394	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
SCARB2	EPILEPSY, PROGRESSIVE MYOCLONIC, 4, WITH OR WITHOUT RENAL [...]	OMIM	MIM:254900	OMIM
SCARB2	Gaucher disease type 1	OrphaNet	OrphaNet:77259	orphanet
SCARB2	Action myoclonus - renal failure syndrome	OrphaNet	OrphaNet:163696	orphanet
SCARB2	Unverricht-Lundborg disease	OrphaNet	OrphaNet:308	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
NAAA	missense variant	ENST00000505594	?	ENSP00000426977	I/V	Atc/Gtc	?	?	1
NAAA	missense variant	ENST00000602782	?	ENSP00000473575	I/V	Atc/Gtc	?	?	1
NAAA	missense variant	ENST00000286733	NM_014435.3	ENSP00000286733	I/V	Atc/Gtc	?	?	1
NAAA	missense variant	ENST00000507956	?	ENSP00000427641	I/V	Atc/Gtc	?	?	1
NAAA	missense variant	ENST00000507187	?	ENSP00000423142	I/V	Atc/Gtc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ART3	?	ENSG00000156219	pancreas	6.72×10^{-15} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	pancreas	9.25×10^{-12} (p-value)	GTEx Portal V6	62

CXCL11	?	ENSG00000169248	pancreas	1.02×10^{-6} (p-value)	GTEx Portal V6	24
NAAA	?	ENSG00000138744	muscularis mucosae	1.64×10^{-18} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	lung	6.09×10^{-21} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	transformed fibroblasts	8.78×10^{-15} (p-value)	GTEx Portal V6	62
SCARB2	?	ENSG00000138760	transformed fibroblasts	1.06×10^{-6} (p-value)	GTEx Portal V6	6
NAAA	?	ENSG00000138744	blood	1.63×10^{-22} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	breast	2.40×10^{-15} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	tibial artery	7.45×10^{-23} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	thyroid	6.38×10^{-15} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	skeletal muscle	6.09×10^{-27} (p-value)	GTEx Portal V6	62
CXCL10	?	ENSG00000169245	EBV lymphocytes	2.93×10^{-9} (p-value)	GTEx Portal V6	42
NAAA	?	ENSG00000138744	transverse colon	1.61×10^{-11} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	blood	1.05×10^{-6} (q-value)	SeeQTL DB (HapMap)	16
CXCL10	?	ENSG00000169245	blood	7.80×10^{-4} (q-value)	SeeQTL DB (HapMap)	12
NAAA	?	ENSG00000138744	sun exposed skin	3.52×10^{-24} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	unexposed skin	5.99×10^{-17} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	aorta	6.37×10^{-22} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	subcutaneous adipocytes	4.54×10^{-18} (p-value)	GTEx Portal V6	62
?	?	ILMN_2067890	monocyte	1.57×10^{-17} (p-value)	Zeller et al.	9
NAAA	ENST00000286733	ILMN_2391512	monocyte	2.13×10^{-19} (p-value)	Zeller et al.	9
NAAA	ENST00000602782					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
?	?	ILMN_1668605	monocyte	4.68×10^{-15} (p-value)	Zeller et al.	5
?	?	ILMN_2285568	monocyte	4.21×10^{-87} (p-value)	Zeller et al.	9
NAAA	?	ENSG00000138744	caudate basal ganglia	1.41×10^{-8} (p-value)	GTEx Portal V6	52
NAAA	?	ENSG00000138744	stomach	3.34×10^{-13} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	visceral adipocytes	2.50×10^{-21} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	sigmoid colon	1.05×10^{-9} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	coronary artery	6.63×10^{-10} (p-value)	GTEx Portal V6	62
ART3	?	ENSG00000156219	tibial nerve	3.51×10^{-9} (p-value)	GTEx Portal V6	60
NAAA	?	ENSG00000138744	tibial nerve	1.59×10^{-11} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	esophagus mucosa	8.68×10^{-25} (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	gastroesophageal junction	2.64×10^{-12} (p-value)	GTEx Portal V6	62

NAAA	?	ENSG00000138744	adrenal gland	1.07×10-16 (p-value)	GTEx Portal V6	62
NAAA	?	ENSG00000138744	left ventricle	2.30×10-9 (p-value)	GTEx Portal V6	60
NAAA	?	ENSG00000138744	atrial appendage	9.88×10-9 (p-value)	GTEx Portal V6	57
NAAA	?	ENSG00000138744	ovary	9.96×10-7 (p-value)	GTEx Portal V6	43
NAAA	?	ENSG00000138744	spleen	1.51×10-7 (p-value)	GTEx Portal V6	45
NAAA	?	ENSG00000138744	pituitary	3.06×10-7 (p-value)	GTEx Portal V6	56
NAAA	?	ENSG00000138744	frontal cortex	2.61×10-7 (p-value)	GTEx Portal V6	42
NAAA	?	ENSG00000138744	liver	2.36×10-7 (p-value)	GTEx Portal V6	32
NAAA	ENST00000286733	ILMN_2391512	blood	7.55×10-28 (p-value)	MuTHER consortium	21
NAAA	ENST00000602782					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
CXCL9	ENST00000264888	ILMN_1745356	blood	1.93×10-6 (p-value)	MuTHER consortium	16
CXCL10	ENST00000306602	ILMN_1791759	blood	3.78×10-35 (p-value)	MuTHER consortium	21
NAAA	ENST00000513045	ILMN_2391512	monocyte	8.05×10-13 (p-value)	Fairfax et al.	3
NAAA	ENST00000602782		b-cell	1.98×10-6 (p-value)	Fairfax et al.	3
NAAA	ENST00000286733					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
?	?	ILMN_1668605	monocyte	1.09×10-12 (p-value)	Fairfax et al.	6
			b-cell	5.29×10-6 (p-value)		
NAAA	ENST00000602782	214765_s_at	blood	2.10×10-22 (p-value)	Dixon et al.	7
NAAA	ENST00000286733					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
NAAA	ENST00000286733	227135_at	blood	2.50×10-21 (p-value)	Dixon et al.	7
NAAA	ENST00000511606					
CXCL10	ENST00000306602	204533_at	blood	2.40×10-18 (p-value)	Dixon et al.	7
NAAA	?	ENSG00000138744	cerebellum	9.13×10-6 (p-value)	GTEx Portal V6	3
?	?	ILMN_1668605	liver	2.89×10-15 (p-value)	Innocenti et al.	1
CXCL10	ENST00000306602	ILMN_1791759	blood	3.55×10-5 (p-value)	Westra et al.	9
NAAA	?	ENSG00000138744	uterus	4.78×10-7 (p-value)	GTEx Portal V6	12
NAAA	?	ENSG00000138744	anterior cingulate cortex	1.32×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382695	1	ENCP00000041438	NUP54
		ENCP00000041452	CCDC158
		ENCP00000041415	NAAA
ENCE00000382651	1	ENCP00000041411	USO1
ENCE00000382654	1	ENCP00000041411	USO1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001432208 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, PolII, Gabp, Sin3Ak20, H3K9ac, Yy1, TAF1, H3K4me2
		HSMMtube	H3K79me2, H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K79me2, HEY1, H2AZ, TAF1, H3K4me2, H3K36me3, DNase1, H3K4me3, Cmyc, Gabp, PolII, Max, H3K9ac
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K4me3
		liver (HepG2)	Gabp, PolII, H3K79me2, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1
		blood (GM12878)	H3K79me2, H3K4me3, H3K4me2, H3K9ac, ELF1, Gabp, DNase1, Cmyc, PolII, H2AZ
		lung (IMR90)	DNase1, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K27ac, H3K9ac
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, PolII, Gabp, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2
		monocytes (Monocytes-CD14+)	H3K36me3, H3K9ac, H3K27ac, H3K4me2, DNase1, H3K4me3, H3K27me3
		endothelium (HUVEC)	Max, DNase1, H3K4me2, H3K4me3, H3K9ac, PolII, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3
ENSR00001243218 (promoter flanking region)	2	NHLF	H3K27ac
		embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K27ac, H3K4me2
		blood (GM12878)	H2AZ, H3K27ac, H3K4me2
		lung (IMR90)	DNase1, H3K18ac, H3K27ac, H3K4ac, H2BK120ac, H2BK20ac, H4K91ac, H3K56ac
		skin (NHEK)	DNase1
ENSR00001243226 (enhancer)	1	embryonic stem cell (H1ESC)	CTCF, Yy1, DNase1
		endothelium (HUVEC)	H3K36me3
		HSMMtube	H3K27me3
		skin (NHEK)	CTCF
ENSR00001685898 (CTCF binding site)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	CTCF, Yy1, DNase1
		HSMMtube	H3K27me3
		skin (NHEK)	CTCF
ENSR00001243231 (enhancer)	1	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	Cjun, BATF, DNase1
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
ENSR00001685900 (promoter flanking region)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3

breast (HMEC)	H3K4me2, DNase1
monocytes (Monocytes-CD14+)	H3K4me1, DNase1
endothelium (HUVEC)	H3K27me3
nervous (NH-A)	H3K27me3
A549	H3K27me3
skin (NHEK)	DNase1, H3K4me1, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC110615.1	upstream gene variant, downstream gene variant	122	ENST00000625176	?	ENSP00000485507	6
AC112719.2	upstream gene variant	1189	ENST00000629900	?	ENSP00000487114	2
ART3	upstream gene variant	1561	ENST00000513353	?	ENSP00000421345	2
ART3	upstream gene variant	2474	ENST00000504914	?	ENSP00000421431	2
ART3	upstream gene variant	1577	ENST00000341029	NM_001130017.2	ENSP00000343843	2
ART3	upstream gene variant	1577	ENST00000510669	?	?	2
ART3	upstream gene variant	1626	ENST00000513122	?	ENSP00000422287	2
CXCL11	downstream gene variant, upstream gene variant	721	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	4
CXCL11	downstream gene variant, upstream gene variant	595	ENST00000503860	?	ENSP00000425819	4
CXCL9	upstream gene variant	2135	ENST00000264888	NM_002416.1	ENSP00000354901	2
NAAA	upstream gene variant	3046	ENST00000505594	?	ENSP00000426977	1
NAAA	upstream gene variant	2333	ENST00000507956	?	ENSP00000427641	1
NAAA	downstream gene variant, upstream gene variant	653	ENST00000503636	?	?	14
NAAA	upstream gene variant	2255	ENST00000286733	NM_014435.3	ENSP00000286733	1
NAAA	upstream gene variant	5	ENST00000602782	?	ENSP00000473575	8
NAAA	downstream gene variant, upstream gene variant	8	ENST00000507940	?	?	16
NAAA	downstream gene variant, upstream gene variant	168	ENST00000507187	?	ENSP00000423142	11
RP11-630D6.1	downstream gene variant, upstream gene variant	591	ENST00000477971	?	?	3
RP11-630D6.5	upstream gene variant, downstream gene variant	1333	ENST00000501239	?	?	3
SDAD1	downstream gene variant	833	ENST00000503411	?	ENSP00000422368	1
SDAD1	downstream gene variant	1148	ENST00000395711	NM_001288983.1	ENSP00000379061	3
SDAD1	downstream gene variant, upstream gene variant	479	ENST00000515836	?	?	4
SDAD1	downstream gene variant	768	ENST00000395710	?	ENSP00000379060	3
SDAD1	downstream gene variant	778	ENST00000356260	NM_001288984.1, NM_018115.3	ENSP00000348596	3
SDAD1	downstream gene variant, upstream gene variant	164	ENST00000502543	?	?	13
SDAD1	downstream gene variant, upstream gene variant	2587	ENST00000513089	?	?	2
SDAD1	downstream gene variant, upstream gene variant	1721	ENST00000507396	?	?	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
	?			V	gtA/gtG	1

NAAA	ENST00000505594		ENSP00000426977			
NAAA	ENST00000507956	?	ENSP00000427641	V	gtA/gtG	1
NAAA	ENST00000507187	?	ENSP00000423142	V	gtA/gtG	1
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	V	gtA/gtG	1
SDAD1	ENST00000395711	NM_001288983.1	ENSP00000379061	D	gaT/gaC	1
SDAD1	ENST00000356260	NM_001288984.1, NM_018115.3	ENSP00000348596	D	gaT/gaC	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
NAAA	ENST00000505594	?	ENSP00000426977	1
NAAA	ENST00000507940	?	?	1
NAAA	ENST00000507956	?	ENSP00000427641	1
NAAA	ENST00000507187	?	ENSP00000423142	1
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	1

Non-coding exon variant (splice region)

gene	affected transcript	RefSeq id	variant(s)
NAAA	ENST00000507940	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC110615.1	ENST00000625176	?	ENSP00000485507	1
ART3	ENST00000510669	?	?	19
ART3	ENST00000504914	?	ENSP00000421431	19
ART3	ENST00000513122	?	ENSP00000422287	19
ART3	ENST00000341029	NM_001130017.2	ENSP00000343843	19
ART3	ENST00000513353	?	ENSP00000421345	19
CXCL11	ENST00000503860	?	ENSP00000425819	3
CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	2
CXCL9	ENST00000264888	NM_002416.1	ENSP00000354901	1
NAAA	ENST00000505594	?	ENSP00000426977	20
NAAA	ENST00000507940	?	?	6
NAAA	ENST00000507956	?	ENSP00000427641	20
NAAA	ENST00000507187	?	ENSP00000423142	10
NAAA	ENST00000602782	?	ENSP00000473575	14
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	20
RP11-630D6.5	ENST00000501239	?	?	4
SDAD1	ENST00000395711	NM_001288983.1	ENSP00000379061	13
SDAD1	ENST00000356260	NM_001288984.1, NM_018115.3	ENSP00000348596	13
SDAD1	ENST00000504975	?	?	5
SDAD1	ENST00000514710	?	ENSP00000427443	5
SDAD1	ENST00000503411	?	ENSP00000422368	5
SDAD1	ENST00000395710	?	ENSP00000379060	13
SDAD1	ENST00000513089	?	?	7
SDAD1	ENST00000502543	?	?	5

SDAD1	ENST00000507396	?	?	3
SDAD1	ENST00000515836	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	1
NAAA	ENST00000507187	?	ENSP00000423142	4
SDAD1	ENST00000395710	?	ENSP00000379060	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-630D6.1	ENST00000477971	?	1

Locus 84 (rs2124910)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,021,247-52,039,859
block size	18,613 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.322 [-4.185 – 0.489]	gene(s) hit or close-by	CTD-3073N11.9, SIGLEC6
phastCons	μ = 0.097 [0 – 0.973]	eQTL gene(s)	SIGLEC12, SIGLEC5, SIGLEC5, SIGLEC6, ZNF175
GERP++	μ = -1.080 [-5.25 – 1.25]	potentially regulated gene(s)	SIGLEC6
CADD score	μ = 4.196 [0.562 – 11.6]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SIGLEC6	missense variant	ENST00000359982	NM_001177548.1	ENSP00000353071	P/S	Cca/Tca	?	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC5	ENST00000534261	ILMN_1740298	blood	2.78×10-8 (p-value)	MuTHER consortium	13
SIGLEC5	ENST00000599649					
SIGLEC5	ENST00000570106					
SIGLEC5	ENST00000429354					
ZNF175	ENST00000436511	ILMN_1675788	blood	1.07×10-5 (p-value)	MuTHER consortium	12
ZNF175	ENST00000262259					
SIGLEC12	?	ENSG00000254521	pancreas	1.93×10-9 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	muscularis mucosae	5.37×10-19 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	lung	3.79×10-31 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	tibial artery	2.23×10-22 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	breast	9.34×10-13 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	blood	3.73×10-35 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	thyroid	1.67×10-10 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	transverse colon	1.09×10-12 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	sun exposed skin	2.23×10-18 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	aorta	3.18×10-17 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	left ventricle	2.15×10-8 (p-value)	GTEx Portal V6	15
SIGLEC12	?	ENSG00000254521	terminal ileum	3.61×10-9 (p-value)	GTEx Portal V6	15

SIGLEC12	?	ENSG00000254521	subcutaneous adipocytes	3.94×10-24 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	visceral adipocytes	8.50×10-8 (p-value)	GTEx Portal V6	15
SIGLEC12	?	ENSG00000254521	stomach	4.48×10-14 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	spleen	5.33×10-16 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	tibial nerve	3.18×10-16 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	coronary artery	4.81×10-7 (p-value)	GTEx Portal V6	9
SIGLEC12	?	ENSG00000254521	vagina	4.25×10-8 (p-value)	GTEx Portal V6	14
SIGLEC12	?	ENSG00000254521	esophagus mucosa	7.00×10-10 (p-value)	GTEx Portal V6	21
SIGLEC5	ENST00000534261	ILMN_1740298	blood	1.47×10-5 (p-value)	Westra et al.	9
SIGLEC5	ENST00000599649					
SIGLEC5	ENST00000570106					
SIGLEC5	ENST00000429354					
SIGLEC6	ENST00000425629	ILMN_2299095	blood	8.57×10-5 (p-value)	Westra et al.	6
SIGLEC6	ENST00000436458					
SIGLEC6	ENST00000391797					
SIGLEC6	ENST00000343300					
SIGLEC6	ENST00000474054					
SIGLEC6	ENST00000346477					
SIGLEC6	ENST00000359982					
SIGLEC12	?	ENSG00000254521	gastroesophageal junction	2.43×10-10 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	adrenal gland	4.48×10-13 (p-value)	GTEx Portal V6	21
SIGLEC5	?	ENSG00000105501	blood	4.65×10-2 (q-value)	SeeQTL DB (HapMap)	1
SIGLEC12	ENST00000291707	ILMN_1676363	monocyte	1.17×10-115 (p-value)	Zeller et al.	3
SIGLEC12	ENST00000596742					
SIGLEC12	ENST00000598614					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024646		

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001419955 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	Max, PU1
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		endothelium (HUVEC)	H3K27me3
		blood (GM12878)	PU1, H3K4me2
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-3073N11.9	upstream gene variant, downstream gene variant	1448	ENST00000598220	?	?	7
SIGLEC6	downstream gene variant, upstream gene variant	646	ENST00000436458	NM_001177547.1	ENSP00000410679	9
SIGLEC6	downstream gene variant, upstream gene variant	646	ENST00000391797	NM_001177549.1	ENSP00000375674	9
SIGLEC6	downstream gene variant, upstream gene variant	105	ENST00000489837	?	?	12
SIGLEC6	downstream gene variant, upstream gene variant	655	ENST00000359982	NM_001177548.1	ENSP00000353071	9
SIGLEC6	downstream gene variant, upstream gene variant	967	ENST00000474054	?	?	7
SIGLEC6	downstream gene variant, upstream gene variant	646	ENST00000425629	NM_001245.5	ENSP00000401502	9
SIGLEC6	downstream gene variant, upstream gene variant	732	ENST00000346477	NM_198845.4	ENSP00000344064	9
SIGLEC6	downstream gene variant, upstream gene variant	593	ENST00000343300	NM_198846.4	ENSP00000345907	9
SIGLEC6	downstream gene variant, upstream gene variant	646	ENST00000496422	?	?	11

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC6	ENST00000496422	?	?	1
SIGLEC6	ENST00000391797	NM_001177549.1	ENSP00000375674	1
SIGLEC6	ENST00000425629	NM_001245.5	ENSP00000401502	1
SIGLEC6	ENST00000343300	NM_198846.4	ENSP00000345907	1
SIGLEC6	ENST00000489837	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC6	ENST00000436458	NM_001177547.1	ENSP00000410679	11
SIGLEC6	ENST00000346477	NM_198845.4	ENSP00000344064	11
SIGLEC6	ENST00000391797	NM_001177549.1	ENSP00000375674	11
SIGLEC6	ENST00000425629	NM_001245.5	ENSP00000401502	11
SIGLEC6	ENST00000474054	?	?	9
SIGLEC6	ENST00000359982	NM_001177548.1	ENSP00000353071	10
SIGLEC6	ENST00000343300	NM_198846.4	ENSP00000345907	11

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC6	ENST00000436458	NM_001177547.1	ENSP00000410679	1
SIGLEC6	ENST00000346477	NM_198845.4	ENSP00000344064	1
SIGLEC6	ENST00000391797	NM_001177549.1	ENSP00000375674	1
SIGLEC6	ENST00000425629	NM_001245.5	ENSP00000401502	1
SIGLEC6	ENST00000359982	NM_001177548.1	ENSP00000353071	1
SIGLEC6	ENST00000343300	NM_198846.4	ENSP00000345907	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SIGLEC6	ENST00000496422	?	2

SIGLEC6	ENST00000474054	?	1
SIGLEC6	ENST00000489837	?	1

Locus 84 (sentinel SNP rs2124910 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 85 (rs10455097)

SNiPAcard

Block annotations

Block info

genomic range	chr6:74,457,833-74,512,687
block size	54,855 bp
variant count	61 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.794 [-5.724 – 1.073]	gene(s) hit or close-by	CD109
phastCons	μ = 0.051 [0 – 1]	eQTL gene(s)	CD109
GERP++	μ = -0.676 [-5.57 – 5.16]	potentially regulated gene(s)	EEF1A1, RP11-554D15.1, RP11-9G10.1
CADD score	μ = 3.302 [0.038 – 15.22]	disease gene(s)	CD109

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Calcium levels	$<3.00\times10^{-6}$	GWAS Catalog	24068962	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM020385	1

Disease gene annotation

gene	trait	source DB	source entry/link
CD109	Fetal and neonatal alloimmune thrombocytopenia	OrphaNet	OrphaNet:853

Direct effect on transcript

Amino acid sequence alteration (splice region)

gene	effect type	affected transcript	RefSeq id	protein	amino acid	codon	SIFT prediction	PolyPhen prediction	varian(s)
CD109	missense variant	ENST00000287097	NM_133493.3	ENSP00000287097	Y/S	tAc/tCc	tolerated	benign	1
CD109	missense variant	ENST00000437994	NM_001159587.1	ENSP00000388062	Y/S	tAc/tCc	tolerated	benign	1
CD109	missense variant	ENST00000422508	NM_001159588.1	ENSP00000404475	Y/S	tAc/tCc	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD109	?	ENSG00000156535	tibial artery	6.50×10^{-8} (p-value)	GTEx Portal V6	60
CD109	?	ENSG00000156535	aorta	9.53×10^{-10} (p-value)	GTEx Portal V6	60

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000454297	1	ENCP00000049135	RP11-9G10.1
ENCE00000454282	2	ENCP00000049134	RP11-554D15.1
ENCE00000454216	1	ENCP00000049134	RP11-554D15.1
		ENCP00000049123	EEF1A1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001705222 (open chromatin region)	1	blood (K562)	H3K27me3
		A549	H3K36me3
ENSR00001705224 (enhancer)	2	skin (NHEK)	H3K36me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K36me3
ENSR00001220456 (promoter flanking region)	5	NHLF	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K36me3, Rad21, CTCF
		HSMMtube	DNase1
		Osteobl	H3K27ac, CTCF
		skin (NHDF-AD)	H3K4me1, CTCF, DNase1
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF, DNase1
		cervix (HeLa-S3)	Cjun, Jund, CTCF
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	DNase1, CTCF
		A549	H3K36me3
		skin (NHEK)	CTCF
ENSR00001705225 (CTCF binding site)	4	NHLF	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K36me3, Rad21, CTCF
		HSMMtube	DNase1
		Osteobl	CTCF, H3K27ac
		skin (NHDF-AD)	H3K4me1, CTCF, DNase1
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF, DNase1
		cervix (HeLa-S3)	Cjun, Jund, CTCF
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1, CTCF
		A549	H3K36me3
		nervous (NH-A)	CTCF, DNase1
		skin (NHEK)	CTCF
ENSR00001220457 (CTCF binding site)	1	skin (NHDF-AD)	H3K4me1
ENSR00001220458 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	CTCF
		skin (NHDF-AD)	DNase1, CTCF, H3K4me1
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	DNase1, CTCF
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3, CTCF
		NHLF	CTCF, DNase1
		Osteobl	H3K27ac, CTCF
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1, CTCF

	A549	H3K36me3, CTCF

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CD109	ENST00000422508	NM_001159588.1	ENSP00000404475	Y	taC/taT	1
CD109	ENST00000437994	NM_001159587.1	ENSP00000388062	Y	taC/taT	1
CD109	ENST00000287097	NM_133493.3	ENSP00000287097	Y	taC/taT	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CD109	ENST00000422508	NM_001159588.1	ENSP00000404475	59
CD109	ENST00000437994	NM_001159587.1	ENSP00000388062	59
CD109	ENST00000287097	NM_133493.3	ENSP00000287097	59
CD109	ENST00000474094	?	?	39

Locus 86 (rs2000999)

SNiPAcard

Block annotations

Block info

genomic range	chr16:72,075,593-72,148,419
block size	72,827 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.185 [-0.934 – 0.628]	**gene(s) hit or close-by**	DHX38, HP, HPR, PMFBP1, TXNL4B
phastCons	μ = 0.025 [0 – 0.128]	**eQTL gene(s)**	DHODH, DHX38, HP, HPR
GERP++	μ = 0.144 [-0.871 – 0.901]	**potentially regulated gene(s)**	–
CADD score	μ = 2.469 [0.059 – 6.223]	**disease gene(s)**	DHODH, HP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Cholesterol, total	$<7.00\times10^{-41}$	GWAS Catalog	24097068	1
LDL cholesterol	$<4.00\times10^{-41}$	GWAS Catalog	24097068	1
Haptoglobin levels	$<8.00\times10^{-59}$	GWAS Catalog	22403646	1
Cholesterol, total	$<3.00\times10^{-24}$	GWAS Catalog	20686565	1
LDL cholesterol	$<2.00\times10^{-22}$	GWAS Catalog	20686565	1

Disease gene annotation

gene	trait	source DB	source entry/link	
DHODH	postaxial acrofacial dysostosis (POADS)	DECIPHER	MIM:263750	OMIM
DHODH	POSTAXIAL ACROFACIAL DYSOSTOSIS	OMIM	MIM:263750	OMIM
HP	ANHAPTOGLOBINEMIA	OMIM	MIM:614081	OMIM
DHODH	Postaxial acrofacial dysostosis	OrphaNet	OrphaNet:246	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DHODH	?	ENSG00000102967	muscularis mucosae	2.49×10^{-7} (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	atrial appendage	9.97×10^{-11} (p-value)	GTEx Portal V6	6
DHODH	?	ENSG00000102967	tibial artery	9.90×10^{-6} (p-value)	GTEx Portal V6	5
HP	?	ENSG00000257017	blood	1.01×10^{-9} (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	blood	4.09×10^{-9} (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	putamen	5.64×10^{-11} (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	left ventricle	1.40×10^{-8} (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	subcutaneous adipocytes	9.37×10^{-6} (p-value)	GTEx Portal V6	4
HPR	?	ENSG00000261701	caudate basal ganglia	2.38×10^{-11} (p-value)	GTEx Portal V6	6

HP	?	ENSG00000257017	caudate basal ganglia	1.00×10-9 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	cortex	3.50×10-17 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	nucleus accumbens	1.10×10-11 (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	nucleus accumbens	2.56×10-6 (p-value)	GTEx Portal V6	1
DHODH	?	ENSG00000102967	tibial nerve	5.77×10-6 (p-value)	GTEx Portal V6	5
HPR	?	ENSG00000261701	anterior cingulate cortex	2.25×10-12 (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	anterior cingulate cortex	2.94×10-8 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	cerebellar hemisphere	3.70×10-15 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	cerebellum	3.66×10-23 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	hypothalamus	2.11×10-11 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	frontal cortex	9.70×10-12 (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	frontal cortex	9.37×10-6 (p-value)	GTEx Portal V6	1
HPR	?	ENSG00000261701	hippocampus	6.43×10-11 (p-value)	GTEx Portal V6	6
DHX38	?	ENSG00000140829	blood	2.46×10-3 (q-value)	SeeQTL DB (HapMap)	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DHX38	downstream gene variant	1612	ENST00000562774	?	ENSP00000462965	1
DHX38	downstream gene variant	1608	ENST00000268482	NM_014003.3	ENSP00000268482	1
DHX38	upstream gene variant, downstream gene variant	1611	ENST00000567142	?	ENSP00000454521	2
DHX38	upstream gene variant, downstream gene variant	1642	ENST00000563819	?	ENSP00000462397	2
DHX38	upstream gene variant	1643	ENST00000564622	?	?	1
DHX38	downstream gene variant	2100	ENST00000569935	?	?	1
DHX38	downstream gene variant	2733	ENST00000566329	?	?	1
DHX38	upstream gene variant, downstream gene variant	1976	ENST00000569952	?	?	2
DHX38	downstream gene variant	1617	ENST00000579387	?	ENSP00000462149	1
DHX38	upstream gene variant, downstream gene variant	1700	ENST00000567552	?	?	2
HP	downstream gene variant	2873	ENST00000562526	?	ENSP00000454413	1
HP	downstream gene variant	2919	ENST00000565574	?	ENSP00000454966	1
HP	downstream gene variant	3038	ENST00000228226	?	ENSP00000228226	1
HP	downstream gene variant	2874	ENST00000398131	NM_001126102.1	ENSP00000381199	1
HP	downstream gene variant	4110	ENST00000576168	?	ENSP00000461999	1
HP	downstream gene variant	2912	ENST00000357763	?	ENSP00000350406	1
HP	downstream gene variant	4740	ENST00000568417	?	ENSP00000462662	1
HP	downstream gene variant	4467	ENST00000569639	?	ENSP00000463755	1
HP	downstream gene variant	2874	ENST00000613898	?	ENSP00000478279	1
HP	downstream gene variant	2874	ENST00000566821	?	?	1

HP	downstream gene variant	2874	ENST00000566821	?	?	1
HP	downstream gene variant	3000	ENST00000567185	?	ENSP00000464070	1
HP	downstream gene variant	2873	ENST00000570083	?	ENSP00000457629	1
HP	downstream gene variant	2874	ENST00000355906	NM_005143.3	ENSP00000348170	1
HP	downstream gene variant	2902	ENST00000567612	?	ENSP00000463491	1
HP	downstream gene variant	3294	ENST00000564499	?	ENSP00000456503	1
PMFBP1	downstream gene variant	4577	ENST00000537465	?	ENSP00000443817	1
PMFBP1	downstream gene variant	4580	ENST00000355636	NM_001160213.1	ENSP00000347854	1
PMFBP1	downstream gene variant	4581	ENST00000379073	?	ENSP00000368364	1
PMFBP1	downstream gene variant	4577	ENST00000237353	NM_031293.2	ENSP00000237353	1
TXNL4B	downstream gene variant	2595	ENST00000562153	?	ENSP00000454635	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DHX38	ENST00000562774	?	ENSP00000462965	1
DHX38	ENST00000268482	NM_014003.3	ENSP00000268482	1
DHX38	ENST00000579387	?	ENSP00000462149	1
HPR	ENST00000566168	?	?	3
HPR	ENST00000561690	?	ENSP00000462916	3
HPR	ENST00000356967	?	ENSP00000349451	3
HPR	ENST00000540303	NM_020995.3	ENSP00000441828	3
PMFBP1	ENST00000537792	?	ENSP00000443366	1
TXNL4B	ENST00000562153	?	ENSP00000454635	3

Locus 88 (rs41341749)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,310,702-34,322,110
block size	11,409 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.125 [-0.459 – 0.909]	gene(s) hit or close-by	CCL14, CCL15, CCL15-CCL14, CCL16, CTB-186H2.3
phastCons	μ = 0.055 [0 – 0.268]	eQTL gene(s)	–
GERP++	μ = -1.285 [-5.68 – 2.44]	potentially regulated gene(s)	CCL14, CCL14
CADD score	μ = 5.865 [3.257 – 9.467]	disease gene(s)	–

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000022624		CCL14

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL14	upstream gene variant	230	ENST00000622526	NM_032962.4	ENSP00000479097	1
CCL14	downstream gene variant, upstream gene variant	21	ENST00000620991	?	ENSP00000484818	2
CCL14	upstream gene variant	121	ENST00000618404	NM_032963.3	ENSP00000481023	1
CCL15	downstream gene variant	1366	ENST00000614368	?	ENSP00000484262	1
CCL15	downstream gene variant	2508	ENST00000617897	NM_032965.4	ENSP00000484078	1
CCL16	upstream gene variant	2205	ENST00000610493	?	ENSP00000478934	3
CCL16	upstream gene variant	2170	ENST00000611905	NM_004590.2	ENSP00000478024	3
CTB-186H2.3	downstream gene variant	169	ENST00000617687	?	?	1
CTB-186H2.3	upstream gene variant, downstream gene variant	40	ENST00000612584	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL14	ENST00000622526	NM_032962.4	ENSP00000479097	2
CCL14	ENST00000620991	?	ENSP00000484818	2
CCL14	ENST00000618404	NM_032963.3	ENSP00000481023	2
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	4
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	4
CTB-186H2.3	ENST00000617687	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL14	ENST00000614009	?	ENSP00000483186	1

CCL14	ENST00000622526	NM_032962.4	ENSP00000479097	1
CCL14	ENST00000618404	NM_032963.3	ENSP00000481023	1
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	1
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL14	ENST00000614009	?	ENSP00000483186	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTB-186H2.3	ENST00000617687	?	1
CTB-186H2.3	ENST00000612584	?	1

Locus 89 (rs1463657)

SNiPAcard

Block annotations

Block info

genomic range	chr3:57,133,138-57,149,229
block size	16,092 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.135 [-1.696 – 0.831]	gene(s) hit or close-by	IL17RD
phastCons	μ = 0.112 [0 – 0.992]	eQTL gene(s)	ARHGEF3, FLNB
GERP++	μ = -0.423 [-4.73 – 2.58]	potentially regulated gene(s)	–
CADD score	μ = 6.125 [0.859 – 19.03]	disease gene(s)	FLNB, IL17RD

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
FLNB	SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME (SCT;)	DECIPHER	MIM:272460
FLNB	atelosteogenesis type 1 (AO1)	DECIPHER	MIM:108720
FLNB	atelosteogenesis type 3 (AO3)	DECIPHER	MIM:108721
FLNB	boomerang dysplasia (BOOMD)	DECIPHER	MIM:112310
FLNB	autosomal dominant Larsen syndrome (LRS1)	DECIPHER	MIM:150250
FLNB	ATELOSTEOGENESIS, TYPE I	OMIM	MIM:108720
FLNB	SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME	OMIM	MIM:272460
FLNB	LARSEN SYNDROME	OMIM	MIM:150250
FLNB	BOOMERANG DYSPLASIA	OMIM	MIM:112310
FLNB	ATELOSTEOGENESIS, TYPE III	OMIM	MIM:108721
IL17RD	HYPOGONADOTROPIC HYPOGONADISM 18 WITH OR WITHOUT ANOSMIA	OMIM	MIM:615267
FLNB	SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME (SCT;)	OrphaNet	OrphaNet:3275
FLNB	ATELOSTEOGENESIS, TYPE II	OrphaNet	OrphaNet:1190
FLNB	BOOMERANG DYSPLASIA	OrphaNet	OrphaNet:1263
FLNB	ATELOSTEOGENESIS, TYPE III	OrphaNet	OrphaNet:56305
FLNB	Autosomal dominant Larsen syndrome	OrphaNet	OrphaNet:503
IL17RD	Kallmann Syndrome	OrphaNet	OrphaNet:478

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IL17RD	missense variant	ENST00000463523	?	ENSP00000417516	T/M	aCg/aTg	tolerated	benign	1
IL17RD	missense variant	ENST00000320057	?	ENSP00000322250	T/M	aCg/aTg	tolerated	benign	1
IL17RD	missense variant	ENST00000296318	NM_017563.3	ENSP00000296318	T/M	aCg/aTg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FLNB	ENST00000484981	ILMN_1664922	monocyte	5.80×10-4 (p-value)	Fairfax et al.	1
FLNB	ENST00000295956					
FLNB	ENST00000429972					
FLNB	ENST00000481470					
FLNB	ENST00000358537					
FLNB	ENST00000419752					
ARHGEF3	?	ENSG00000163947	transformed fibroblasts	2.44×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
MYC, MAX	MA0059.1	6	yes	-0.063	1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001479531 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K27me3
ENSR00001365042 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, Max, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, Max, Cmyc, CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		A549	CTCF
ENSR00001365044 (enhancer)	1	cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL17RD	downstream gene variant	1561	ENST00000467210	?	ENSP00000418368	2
IL17RD	downstream gene variant	4774	ENST00000479825	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL17RD	ENST00000469841	?	?	8
IL17RD	ENST00000467210	?	ENSP00000418368	5

IL17RD	ENST00000320057	?	ENSP00000322250	8
IL17RD	ENST00000296318	NM_017563.3	ENSP00000296318	8
IL17RD	ENST00000463523	?	ENSP00000417516	8
IL17RD	ENST00000479825	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
IL17RD	ENST00000469841	?	1

Locus 90 (rs6138152)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,811,186-23,859,319
block size	48,134 bp
variant count	42 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.608 [-3.824 – 1.415]	gene(s) hit or close-by	CST2, CST5
phastCons	μ = 0.029 [0 – 0.353]	eQTL gene(s)	CST5
GERP++	μ = -0.237 [-3.17 – 1.19]	potentially regulated gene(s)	CST11, CST8, CSTL1, CSTP1, GGTLC1, RP4-564O4.1, RP5-1100I6.1
CADD score	μ = 2.157 [0.003 – 8.957]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Response to TNF antagonist treatment	$<3.00\times10^{-6}$	GWAS Catalog	18615156	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST5	?	ENSG00000170367	lung	1.25×10^{-9} (p-value)	GTEx Portal V6	42

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000290711	1	ENCP00000031098	GGTLC1
		ENCP00000031079	CST11
		ENCP00000031078	CSTL1
		ENCP00000031081	CST8
		ENCP00000031096	CSTP1
ENCE00000290878	1	ENCP00000031101	RP4-564O4.1
		ENCP00000031099	RP5-1100I6.1
ENCE00000290882	1	ENCP00000031101	RP4-564O4.1
		ENCP00000031099	RP5-1100I6.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001664593 (CTCF binding site)	1	HSMMtube	H3K27me3
ENSR00000233887 (open chromatin region)	2	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3

		A549	H3K27me3
ENSR00001664595 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF
		endothelium (HUVEC)	H3K36me3
		HSMMtube	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CST2	upstream gene variant	3818	ENST00000304725	NM_001322.2	ENSP00000307540	3
CST5	downstream gene variant	2403	ENST00000304710	NM_001900.4	ENSP00000307132	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST5	ENST00000304710	NM_001900.4	ENSP00000307132	4

Locus 91 (rs4688759)

SNiPAcard

Block annotations

Block info

genomic range	chr3:49,597,055-50,125,108
block size	528,054 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.424 [-1.408 – 3.578]	gene(s) hit or close-by	AC139451.2, ACTBP13, AMIGO3, BSN, CAMKV, GMPPB, IP6K1, RBM5, RBM6, RN7SL217P, RNF123, RP13-1056D16.2, TRAIP
phastCons	μ = 0.194 [0 – 1]	eQTL gene(s)	AMT, APEH, C3orf62, CCDC36, CCDC71, HYAL3, MIR5193, NCKIPSD, NICN1, RBM6, RP11-3B7.1, SEMA3F, SLC26A6, TMEM89, UBA7, USP4, WDR6
GERP++	μ = 0.601 [-1.52 – 5.04]	potentially regulated gene(s)	–
CADD score	μ = 5.022 [0.248 – 16.95]	disease gene(s)	AMT, GMPPB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899
GMPPB	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN [...]	OMIM	MIM:615350
GMPPB	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL [...]	OMIM	MIM:615351
GMPPB	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE), TYPE C, [...]	OMIM	MIM:615352
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899
GMPPB	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYEANOMALIES), TYPE A, 14	DECIPHER	MIM:615350
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863
GMPPB	Autosomal recessive limb-girdle muscular dystrophy type 2T	OrphaNet	OrphaNet:363623
GMPPB	Congenital muscular dystrophy with intellectual disability	OrphaNet	OrphaNet:370968
GMPPB	Muscle eye brain disease	OrphaNet	OrphaNet:588
GMPPB	Congenital muscular dystrophy with cerebellar involvement	OrphaNet	OrphaNet:370959

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
RNF123	missense variant	ENST00000327697	NM_022064.3	ENSP00000328287	H/R	cAc/cGc	?	?	1
RNF123	missense variant	ENST00000432042	?	ENSP00000392443	H/R	cAc/cGc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)

WDR6	?	ENSG00000178252	visceral adipocytes	1.17×10-7 (p-value)	GTEx Portal V6	16
NCKIPSD	?	ENSG00000213672	visceral adipocytes	4.77×10-6 (p-value)	GTEx Portal V6	5
WDR6	?	ENSG00000178252	stomach	1.76×10-8 (p-value)	GTEx Portal V6	16
WDR6	?	ENSG00000178252	lung	8.08×10-8 (p-value)	GTEx Portal V6	16
SLC26A6	?	ENSG00000225697	lung	1.05×10-5 (p-value)	GTEx Portal V6	4
WDR6	?	ENSG00000178252	tibial nerve	1.54×10-11 (p-value)	GTEx Portal V6	16
SEMA3F	?	ENSG00000001617	tibial nerve	3.87×10-7 (p-value)	GTEx Portal V6	18
WDR6	?	ENSG00000178252	transformed fibroblasts	2.14×10-12 (p-value)	GTEx Portal V6	16
NCKIPSD	?	ENSG00000213672	transformed fibroblasts	3.63×10-5 (p-value)	GTEx Portal V6	4
CCDC36	?	ENSG00000173421	transformed fibroblasts	4.92×10-5 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	tibial artery	9.89×10-9 (p-value)	GTEx Portal V6	16
TMEM89	?	ENSG00000183396	tibial artery	1.54×10-5 (p-value)	GTEx Portal V6	5
NICN1	?	ENSG00000145029	tibial artery	6.59×10-8 (p-value)	GTEx Portal V6	18
AMT	?	ENSG00000145020	tibial artery	1.71×10-6 (p-value)	GTEx Portal V6	10
WDR6	?	ENSG00000178252	breast	3.36×10-7 (p-value)	GTEx Portal V6	14
WDR6	?	ENSG00000178252	blood	1.65×10-10 (p-value)	GTEx Portal V6	16
WDR6	?	ENSG00000178252	esophagus mucosa	1.92×10-33 (p-value)	GTEx Portal V6	16
NCKIPSD	?	ENSG00000213672	esophagus mucosa	3.59×10-8 (p-value)	GTEx Portal V6	5
HYAL3	?	ENSG00000186792	esophagus mucosa	1.61×10-5 (p-value)	GTEx Portal V6	4
RBM6	?	ENSG00000004534	esophagus mucosa	4.58×10-5 (p-value)	GTEx Portal V6	2
CCDC36	?	ENSG00000173421	testis	1.22×10-7 (p-value)	GTEx Portal V6	15
WDR6	?	ENSG00000178252	skeletal muscle	5.26×10-10 (p-value)	GTEx Portal V6	16
SLC26A6	?	ENSG00000225697	skeletal muscle	2.04×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	unexposed skin	1.92×10-22 (p-value)	GTEx Portal V6	16
WDR6	?	ENSG00000178252	sun exposed skin	2.32×10-45 (p-value)	GTEx Portal V6	16
RBM6	?	ENSG00000004534	sun exposed skin	3.04×10-6 (p-value)	GTEx Portal V6	7
WDR6	?	ENSG00000178252	terminal ileum	9.22×10-7 (p-value)	GTEx Portal V6	15
WDR6	?	ENSG00000178252	subcutaneous adipocytes	2.03×10-19 (p-value)	GTEx Portal V6	16
RBM6	?	ENSG00000004534	subcutaneous adipocytes	4.30×10-5 (p-value)	GTEx Portal V6	3
CCDC36	?	ENSG00000173421	muscularis mucosae	1.11×10-6 (p-value)	GTEx Portal V6	10
WDR6	ENST00000615452	ILMN_1669484	skin	1.08×10-38 (p-value)	MuTHER consortium	2
WDR6	ENST00000452875		adipocyte	7.89×10-35 (p-value)	MuTHER consortium	2
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					

Locus 91 (sentinel SNP rs4688759 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	skin	4.51×10-8 (p-value)	MuTHER consortium	3
USP4	ENST00000483212		adipocyte	5.30×10-13 (p-value)	MuTHER consortium	3
USP4	ENST00000485450		blood	6.42×10-5 (p-value)	MuTHER consortium	3
C3orf62	ENST00000343010					
RP11-3B7.1	ENST00000622888	ILMN_1807244	adipocyte	2.17×10-18 (p-value)	MuTHER consortium	3
RP11-3B7.1	ENST00000440528					
WDR6	ENST00000615452	ILMN_1669484	monocyte	1.33×10-29 (p-value)	Zeller et al.	1
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	monocyte	2.05×10-34 (p-value)	Zeller et al.	1
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
APEH	ENST00000480772	ILMN_1718023	blood	1.03×10-4 (p-value)	Westra et al.	2
APEH	ENST00000469362					
APEH	ENST00000296456					
APEH	ENST00000438011					
WDR6	ENST00000615452	ILMN_1669484	monocyte	3.22×10-14 (p-value)	Fairfax et al.	2
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
UBA7	ENST00000497908	ILMN_1794612	b-cell	3.40×10-5 (p-value)	Fairfax et al.	2
UBA7	ENST00000333486					
MIR5193	ENST00000584510					
USP4	ENST00000351842	ILMN_1798712	monocyte	6.64×10-11 (p-value)	Fairfax et al.	2
USP4	ENST00000483212		b-cell	5.42×10-8 (p-value)	Fairfax et al.	2
C3orf62	ENST00000343010					
USP4	ENST00000485450					
CCDC71	ENST00000321895	ILMN_1768433	monocyte	1.86×10-4 (p-value)	Fairfax et al.	1
			b-cell	6.44×10-4 (p-value)	Fairfax et al.	1
SLC26A6	ENST00000420764	ILMN_1785252	b-cell	5.46×10-6 (p-value)	Fairfax et al.	2
SLC26A6	ENST00000395550					
SLC26A6	ENST00000489483					
SLC26A6	ENST00000480524					
SLC26A6	ENST00000455886					
SLC26A6	ENST00000307364					
SLC26A6	ENST00000614797					
SLC26A6	ENST00000358747					
SLC26A6	ENST00000383733					
SLC26A6	ENST00000337000					
RBM6	ENST00000442092	ILMN_1738239	monocyte	2.69×10-4 (p-value)	Fairfax et al.	1

RBM6	ENST00000266022		b-cell	1.61×10-4 (p-value)	Fairfax et al.	1
RBM6	ENST00000454079					
RBM6	ENST00000422955					
RBM6	ENST00000419610					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
HYAL3	?	ENSG00000186792	left ventricle	2.01×10-6 (p-value)	GTEx Portal V6	5

Putative effect on regulation

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
RNF123	1	ENST00000327697 ENST00000432042 ENST00000457726 ENST00000486102 ENST00000487805	hsa-miR-504-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC139451.2	upstream gene variant	2051	ENST00000627736	?	?	1
ACTBP13	downstream gene variant	2442	ENST00000423971	?	?	1
AMIGO3	downstream gene variant	4291	ENST00000320431	NM_198722.2	ENSP00000323096	1
CAMKV	upstream gene variant	1000	ENST00000466535	?	ENSP00000420766	1
CAMKV	upstream gene variant	683	ENST00000477224	NM_024046.3	ENSP00000419195	1
CAMKV	upstream gene variant	974	ENST00000463537	?	ENSP00000417614	1
CAMKV	upstream gene variant	1000	ENST00000498324	?	?	1
CAMKV	upstream gene variant	4491	ENST00000478149	?	?	1
CAMKV	upstream gene variant	970	ENST00000480398	?	ENSP00000420000	1
CAMKV	upstream gene variant	3109	ENST00000483811	?	?	1
CAMKV	upstream gene variant	1000	ENST00000296471	?	ENSP00000296471	1
CAMKV	upstream gene variant	999	ENST00000467248	?	ENSP00000420053	1
CAMKV	upstream gene variant	1000	ENST00000479704	?	?	1
CAMKV	upstream gene variant	996	ENST00000466940	?	ENSP00000420724	1
CAMKV	upstream gene variant	1000	ENST00000476105	?	?	1
CAMKV	upstream gene variant	1002	ENST00000475665	?	?	1
CAMKV	upstream gene variant	3127	ENST00000620470	?	ENSP00000484045	1
CAMKV	upstream gene variant	999	ENST00000488336	?	ENSP00000418809	1
CAMKV	upstream gene variant	3109	ENST00000472895	?	?	1
CAMKV	upstream gene variant	1002	ENST00000487726	?	ENSP00000420139	1
GMPPB	downstream gene variant	4301	ENST00000480687	?	ENSP00000418565	1
RBM5	upstream gene variant	1279	ENST00000441305	?	ENSP00000390711	1
RBM5	upstream gene variant	2695	ENST00000417905	?	ENSP00000406119	1
RBM5	upstream gene variant	1244	ENST00000404526	?	ENSP00000384872	1
RBM5	upstream gene variant	2206	ENST00000437500	?	ENSP00000394622	1
RBM5	upstream gene variant	1280	ENST00000438369	?	ENSP00000414218	1

RBM5	upstream gene variant	1279	ENST00000433556	?	?	1
RBM5	upstream gene variant	1244	ENST00000469838	?	ENSP00000419534	1
RBM5	upstream gene variant	1233	ENST00000347869	NM_005778.3	ENSP00000343054	1
RBM5	upstream gene variant	1300	ENST00000492472	?	?	1
RBM5	upstream gene variant	1288	ENST00000464087	?	?	1
RBM6	downstream gene variant	3031	ENST00000433811	?	ENSP00000389763	1
RBM6	downstream gene variant	933	ENST00000493652	?	?	1
RBM6	downstream gene variant	3327	ENST00000491874	?	?	1
RBM6	downstream gene variant	3320	ENST00000488807	?	?	1
RBM6	downstream gene variant	1423	ENST00000438912	?	ENSP00000403865	1
RBM6	downstream gene variant	42	ENST00000441115	?	?	1
RN7SL217P	upstream gene variant	1855	ENST00000584520	?	?	1
RNF123	upstream gene variant	287	ENST00000433785	?	ENSP00000416156	1
RNF123	upstream gene variant	984	ENST00000444689	?	ENSP00000413794	1
RNF123	upstream gene variant	7	ENST00000469978	?	?	1
RP13-1056D16.2	downstream gene variant	211	ENST00000463176	?	?	2
TRAIP	downstream gene variant	2334	ENST00000488860	?	?	2
TRAIP	downstream gene variant	2344	ENST00000482243	?	ENSP00000419350	2
TRAIP	downstream gene variant	3564	ENST00000489948	?	?	1
TRAIP	downstream gene variant	3699	ENST00000473863	?	?	1
TRAIP	downstream gene variant	2993	ENST00000477546	?	?	1
TRAIP	downstream gene variant	1915	ENST00000482582	?	ENSP00000418544	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
RBM6	ENST00000443081	?	ENSP00000396466	Q	caA/caG	1
RBM6	ENST00000442092	NM_001167582.1	ENSP00000393530	Q	caA/caG	1
RBM6	ENST00000266022	NM_005777.2	ENSP00000266022	Q	caA/caG	1
RBM6	ENST00000422955	?	ENSP00000392939	Q	caA/caG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BSN	ENST00000296452	NM_003458.3	ENSP00000296452	5
CAMKV	ENST00000463537	?	ENSP00000417614	1
CAMKV	ENST00000477224	NM_024046.3	ENSP00000419195	1
CAMKV	ENST00000296471	?	ENSP00000296471	1
CAMKV	ENST00000466535	?	ENSP00000420766	1
CAMKV	ENST00000487726	?	ENSP00000420139	1
CAMKV	ENST00000475665	?	?	1
CAMKV	ENST00000467248	?	ENSP00000420053	1

CAMKV	ENST00000476105	?	?	1
CAMKV	ENST00000480398	?	ENSP00000420000	1
CAMKV	ENST00000479704	?	?	1
CAMKV	ENST00000466940	?	ENSP00000420724	1
CAMKV	ENST00000488336	?	ENSP00000418809	1
CAMKV	ENST00000498324	?	?	1
IP6K1	ENST00000613416	NM_001242829.1	ENSP00000482032	2
IP6K1	ENST00000321599	NM_153273.3	ENSP00000323780	2
IP6K1	ENST00000395238	NM_001006115.2	ENSP00000378659	2
IP6K1	ENST00000468463	?	ENSP00000420467	2
IP6K1	ENST00000498149	?	?	2
IP6K1	ENST00000460540	?	ENSP00000420762	2
RBM6	ENST00000483350	?	?	2
RBM6	ENST00000493652	?	?	2
RBM6	ENST00000441115	?	?	6
RBM6	ENST00000443081	?	ENSP00000396466	6
RBM6	ENST00000464013	?	?	2
RBM6	ENST00000434592	?	ENSP00000399942	7
RBM6	ENST00000488807	?	?	3
RBM6	ENST00000442092	NM_001167582.1	ENSP00000393530	6
RBM6	ENST00000491874	?	?	3
RBM6	ENST00000425608	?	ENSP00000408665	6
RBM6	ENST00000266022	NM_005777.2	ENSP00000266022	6
RBM6	ENST00000454079	?	ENSP00000406548	6
RBM6	ENST00000422955	?	ENSP00000392939	6
RBM6	ENST00000419610	?	ENSP00000396410	6
RBM6	ENST00000433811	?	ENSP00000389763	3
TRAIP	ENST00000475495	?	?	2
TRAIP	ENST00000473195	?	ENSP00000419556	2
TRAIP	ENST00000482582	?	ENSP00000418544	1
TRAIP	ENST00000331456	NM_005879.2	ENSP00000328203	2
TRAIP	ENST00000469027	?	ENSP00000420085	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RBM6	ENST00000434592	?	ENSP00000399942	1
RBM6	ENST00000454079	?	ENSP00000406548	1
RBM6	ENST00000419610	?	ENSP00000396410	1
RNF123	ENST00000457726	?	ENSP00000394369	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RBM6	ENST00000421682	?	ENSP00000392475	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RNF123	ENST00000487805	?	1
RNF123	ENST00000486102	?	1

Locus 92 (rs16971802)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,311,387-34,350,776
block size	39,390 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.338 [0.241 – 0.521]	gene(s) hit or close-by	CCL14, CCL15, CCL15-CCL14, CCL16, CTB-186H2.3
phastCons	μ = 0.140 [0.053 – 0.304]	eQTL gene(s)	–
GERP++	μ = 1.229 [0.158 – 4.05]	potentially regulated gene(s)	–
CADD score	μ = 5.712 [1.539 – 13.93]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CCL14	missense variant	ENST00000614009	?	ENSP00000483186	E/K	Gag/Aag	?	?	1
CCL14	missense variant	ENST00000622526	NM_032962.4	ENSP00000479097	E/K	Gag/Aag	?	?	1
CCL14	missense variant	ENST00000618404	NM_032963.3	ENSP00000481023	E/K	Gag/Aag	?	?	1
CCL14	missense variant	ENST00000620991	?	ENSP00000484818	E/K	Gag/Aag	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL14	upstream gene variant	4965	ENST00000618404	NM_032963.3	ENSP00000481023	1
CCL14	upstream gene variant	4719	ENST00000614009	?	ENSP00000483186	1
CCL15	downstream gene variant	4717	ENST00000614368	?	ENSP00000484262	1
CCL16	upstream gene variant	2855	ENST00000611905	NM_004590.2	ENSP00000478024	1
CCL16	upstream gene variant	2890	ENST00000610493	?	ENSP00000478934	1
CTB-186H2.3	upstream gene variant, downstream gene variant	1234	ENST00000612584	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	2
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	2
CTB-186H2.3	ENST00000617687	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

CCL15-CCL14	ENST00000610751	?	ENSP00000481940	1
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	1

Locus 92 (sentinel SNP rs16971802 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 93 (rs7588285)

SNiPAcard

Block annotations

Block info

genomic range	chr2:3,639,909-3,648,186
block size	8,278 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.830 [-1.637 – 0.331]	gene(s) hit or close-by	AC010907.2, COLEC11
phastCons	μ = 0.022 [0 – 0.08]	eQTL gene(s)	ALLC, COLEC11, RNASEH1-AS1
GERP++	μ = 0.320 [-0.397 – 2.32]	potentially regulated gene(s)	–
CADD score	μ = 4.127 [0.417 – 5.959]	disease gene(s)	COLEC11

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
COLEC11	3MC SYNDROME 2	OMIM	MIM:265050
COLEC11	3MC SYNDROME 2	DECIPHER	MIM:265050
COLEC11	Craniofacial-ulnar-renal syndrome	OrphaNet	OrphaNet:293843

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ALLC	?	ENSG00000151360	liver	1.92×10^{-6} (p-value)	GTEx Portal V6	2
RNASEH1-AS1	?	ENSG00000234171	thyroid	2.37×10^{-6} (p-value)	GTEx Portal V6	3
COLEC11	?	ENSG00000118004	pancreas	2.78×10^{-7} (p-value)	GTEx Portal V6	3
RNASEH1-AS1	?	ENSG00000234171	tibial nerve	9.49×10^{-6} (p-value)	GTEx Portal V6	1
COLEC11	ENST00000382062	ILMN_1735192	liver	1.68×10^{-11} (p-value)	Innocenti et al.	1
COLEC11	ENST00000402922					
COLEC11	ENST00000403096					
COLEC11	ENST00000402794					
COLEC11	ENST00000487365					
COLEC11	ENST00000416132					
COLEC11	ENST00000460971					
COLEC11	ENST00000349077					
COLEC11	ENST00000404205					
COLEC11	ENST00000236693					
COLEC11	ENST00000418971					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors

ENSR00000584312 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, TAF1, Yy1, Rad21, CTCF, H3K27me3
		endothelium (HUVEC)	DNase1
		Osteobl	H3K27me3, H3K4me2
		blood (GM12878)	H3K36me3
		blood (K562)	PolII, Max, Rad21, CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC010907.2	downstream gene variant	2801	ENST00000456450	?	?	2
COLEC11	upstream gene variant	383	ENST00000416132	?	ENSP00000410919	5
COLEC11	upstream gene variant	321	ENST00000349077	NM_001255982.1, NM_024027.4	ENSP00000339168	5
COLEC11	upstream gene variant	184	ENST00000460971	?	?	5
COLEC11	upstream gene variant	61	ENST00000382062	NM_001255983.1, NM_001255984.1	ENSP00000371494	5
COLEC11	upstream gene variant	1310	ENST00000418971	NM_001255985.1	ENSP00000411770	2
COLEC11	upstream gene variant	278	ENST00000236693	NM_199235.2	ENSP00000236693	5
COLEC11	upstream gene variant	184	ENST00000487365	?	?	5
COLEC11	upstream gene variant	65	ENST00000419002	?	?	5
COLEC11	upstream gene variant	1310	ENST00000438814	?	ENSP00000393167	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
COLEC11	ENST00000416132	?	ENSP00000410919	2
COLEC11	ENST00000460971	?	?	2
COLEC11	ENST00000349077	NM_001255982.1, NM_024027.4	ENSP00000339168	2
COLEC11	ENST00000487365	?	?	2
COLEC11	ENST00000419002	?	?	2
COLEC11	ENST00000382062	NM_001255983.1, NM_001255984.1	ENSP00000371494	2
COLEC11	ENST00000236693	NM_199235.2	ENSP00000236693	2

Locus 94 (rs3803884)

SNiPAcard

Block annotations

Block info

genomic range	chr17:48,907,545-48,940,422
block size	32,878 bp
variant count	39 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.454 [-6.153 – 4.074]	gene(s) hit or close-by	RP11-506D12.5, RPL5P33, TOB1, TOB1-AS1, WFIKKN2
phastCons	μ = 0.116 [0 – 1]	eQTL gene(s)	TOB1
GERP++	μ = -1.209 [-7.88 – 5.84]	potentially regulated gene(s)	ABCC3, ACSF2, ANKRD40, CACNA1G, LUC7L3, MYCBPAP, NME2, RP11-294J22.5, RP11-700H6.1, RP11-94C24.8, RSAD1, SPATA20, TOB1, WFIKKN2, XYLT2
CADD score	μ = 4.953 [0.02 – 21.9]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TOB1	ENST00000499247	ILMN_1672004	skin	2.36×10-6 (p-value)	MuTHER consortium	14
TOB1	ENST00000509385		blood	4.66×10-6 (p-value)	MuTHER consortium	14
TOB1	ENST00000268957					
?	?	228834_at	blood	9.00×10-19 (p-value)	Dixon et al.	6

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000023247		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000209627	2	ENCP00000023246	RP11-294J22.5
ENCE00000209535	1	ENCP00000023224	SPATA20
ENCE00000209342	1	ENCP00000023207	RSAD1
		ENCP00000023253	RP11-700H6.1
		ENCP00000023223	SPATA20
		ENCP00000023195	ACSF2
		ENCP00000023218	RP11-94C24.8
		ENCP00000023220	SPATA20
		ENCP00000023250	TOB1
		ENCP00000023222	SPATA20
		ENCP00000023205	RSAD1
		ENCP00000023271	NME2
		ENCP00000023187	XYLT2
		ENCP00000023237	ABCC3

		ENCP00000023241	ANKRD40
		ENCP00000023188	XYLT2
		ENCP00000023226	CACNA1G
		ENCP00000023197	ACSF2
		ENCP00000023196	ACSF2
		ENCP00000023213	MYCBPAP
		ENCP00000023228	CACNA1G
		ENCP00000023242	LUC7L3
		ENCP00000023233	ABCC3
ENCE00000209343	1	ENCP00000023207	RSAD1
		ENCP00000023253	RP11-700H6.1
		ENCP00000023223	SPATA20
		ENCP00000023195	ACSF2
		ENCP00000023218	RP11-94C24.8
		ENCP00000023220	SPATA20
		ENCP00000023250	TOB1
		ENCP00000023222	SPATA20
		ENCP00000023205	RSAD1
		ENCP00000023271	NME2
		ENCP00000023237	ABCC3
		ENCP00000023187	XYLT2
		ENCP00000023241	ANKRD40
		ENCP00000023188	XYLT2
		ENCP00000023226	CACNA1G
		ENCP00000023197	ACSF2
		ENCP00000023196	ACSF2
		ENCP00000023228	CACNA1G
		ENCP00000023213	MYCBPAP
		ENCP00000023242	LUC7L3
		ENCP00000023233	ABCC3
ENCE00000209722	2	ENCP00000023270	NME2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001348919 (enhancer)	1	liver (HepG2)	H3K27me3
		blood (K562)	H2AZ
ENSR00001639631 (enhancer)	1	monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	DNase1
		embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K27me3, DNase1
		blood (K562)	CTCF
ENSR00001639632 (enhancer)	3	embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, CTCF, H3K27me3, DNase1
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	CTCF

		A549	CTCF
ENSR00001348925 (promoter flanking region)	3	embryonic stem cell (H1ESC)	USF1
		HSMMtube	H3K27ac, DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K27ac
		liver (HepG2)	USF1, H3K4me1, H3K4me2, HNF4A, H3K27me3, DNase1
		skin (NHEK)	DNase1
ENSR00001639636 (enhancer)	1	endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me1
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-506D12.5	downstream gene variant, upstream gene variant	52	ENST00000572491	?	?	20
RPL5P33	upstream gene variant, downstream gene variant	634	ENST00000487833	?	?	12
TOB1	downstream gene variant	2373	ENST00000499247	NM_005749.3, NM_001243885.1	ENSP00000427695	3
TOB1	downstream gene variant	308	ENST00000509385	?	?	3
TOB1	downstream gene variant	3130	ENST00000268957	NM_001243877.1	ENSP00000268957	2
TOB1-AS1	upstream gene variant	3698	ENST00000523470	?	?	1
TOB1-AS1	upstream gene variant	4087	ENST00000514358	?	?	1
TOB1-AS1	upstream gene variant	3618	ENST00000416263	?	?	1
WFIKKN2	upstream gene variant, downstream gene variant	22	ENST00000426127	?	ENSP00000405889	18
WFIKKN2	upstream gene variant, downstream gene variant	370	ENST00000311378	NM_175575.5	ENSP00000311184	16

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TOB1	ENST00000499247	NM_005749.3, NM_001243885.1	ENSP00000427695	K	aaA/aaG	1
TOB1	ENST00000268957	NM_001243877.1	ENSP00000268957	K	aaA/aaG	1
WFIKKN2	ENST00000311378	NM_175575.5	ENSP00000311184	S	agT/agC	1
WFIKKN2	ENST00000426127	?	ENSP00000405889	S	agT/agC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
WFIKKN2	ENST00000311378	NM_175575.5	ENSP00000311184	9
WFIKKN2	ENST00000426127	?	ENSP00000405889	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
WFIKKN2	ENST00000311378	NM_175575.5	ENSP00000311184	1

Locus 95 (rs11715835)

SNiPAcard

Block annotations

Block info

genomic range	chr3:48,688,114-49,269,270
block size	581,157 bp
variant count	107 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.089 [-2.931 – 4.247]	**gene(s) hit or close-by**	AC141002.1, ARIH2, ARIH2OS, C3orf84, CCDC36, CCDC71, CELSR3, DALRD3, IMPDH2, IP6K2, KLHDC8B, LAMB2, MIR6890, NCKIPSD, NDUFAF3, P4HTM, PRKAR2A, PRKAR2A-AS1, QARS, QRICH1, RN7SL182P, RP11-694I15.7, RP13-131K19.1, RP13-131K19.2, RP13-131K19.6, SLC25A20, USP19, WDR6
phastCons	μ = 0.113 [0 – 1]	**eQTL gene(s)**	AMT, ARIH2, C3orf62, CCDC36, CCDC71, CELSR3, IP6K2, KLHDC8B, MIR5193, NCKIPSD, NICN1, RBM6, RP11-3B7.1, SEMA3F, SLC26A6, TMA7, TMEM89, UBA7, USP4, WDR6
GERP++	μ = -0.294 [-10 – 4.95]	**potentially regulated gene(s)**	AMT, AMT, ARIH2, ATRIP, CCDC36, CELSR3, COL7A1, COL7A1, DALRD3, IMPDH2, IP6K2, KLHDC8B, NCKIPSD, P4HTM, PRKAR2A, QARS, SLC26A6, SPINK8, UCN2, USP19, WDR6
CADD score	μ = 5.047 [0.048 – 21.7]	**disease gene(s)**	KLHDC8B, AMT, SLC25A20, COL7A1, NDUFAF3, QARS, LAMB2, ATRIP

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
KLHDC8B	LYMPHOMA, HODGKIN, CLASSIC	OMIM	MIM:236000
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OMIM	MIM:212138
COL7A1	NAIL DISORDER, NONSYNDROMIC CONGENITAL, 8	OMIM	MIM:607523
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, PRETIBIAL	OMIM	MIM:131850
COL7A1	EPIDERMOLYSIS BULLOSA PRURIGINOSA	OMIM	MIM:604129
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL DOMINANT	OMIM	MIM:131750
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL RECESSIVE	OMIM	MIM:226600
COL7A1	EPIDERMOLYSIS BULLOSA WITH CONGENITAL LOCALIZED ABSENCE OF SKIN [...]	OMIM	MIM:132000
COL7A1	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN	OMIM	MIM:131705
NDUFAF3	MITOCHONDRIAL COMPLEX I DEFICIENCY	OMIM	MIM:252010
QARS	MICROCEPHALY, PROGRESSIVE, WITH SEIZURES AND CEREBRAL AND [...]	OMIM	MIM:615760
LAMB2	NEPHROTIC SYNDROME, TYPE 5, WITH OR WITHOUT OCULAR [...]	OMIM	MIM:614199
LAMB2	PIERSON SYNDROME	OMIM	MIM:609049
ATRIP	Seckel syndrome	OrphaNet	OrphaNet:808
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OrphaNet	OrphaNet:159
COL7A1	Generalized dominant dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:231568
COL7A1	Dystrophic epidermolysis bullosa pruriginosa	OrphaNet	OrphaNet:89843
COL7A1	Recessive dystrophic epidermolysis bullosa inversa	OrphaNet	OrphaNet:79409
COL7A1	Dystrophic epidermolysis bullosa, nails only	OrphaNet	OrphaNet:158676
COL7A1	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN	OrphaNet	OrphaNet:79411
COL7A1	Centripetalis recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:89841
COL7A1	Acral dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:158673
COL7A1	Recessive dystrophic epidermolysis bullosa-generalized other	OrphaNet	OrphaNet:89842

COL7A1	Epidermolysis bullosa simplex superficialis	OrphaNet	OrphaNet:89839	orphanet
COL7A1	Severe generalized recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79408	orphanet
COL7A1	Pretibial dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79410	orphanet
NDUFAF3	Isolated NADH-CoQ reductase deficiency	OrphaNet	OrphaNet:2609	orphanet
LAMB2	Synaptic congenital myasthenic syndromes	OrphaNet	OrphaNet:98915	orphanet
LAMB2	LAMB-2-related infantile-onset nephrotic syndrome	OrphaNet	OrphaNet:306507	orphanet
LAMB2	PIERSON SYNDROME	OrphaNet	OrphaNet:2670	orphanet
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899	OMIM
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	DECIPHER	MIM:212138	OMIM
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	DECIPHER	MIM:615760	OMIM

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IP6K2	missense variant	ENST00000449610	?	ENSP00000393077	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000431721	NM_001190316.1	ENSP00000414139	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000453202	?	ENSP00000387394	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000443964	?	ENSP00000410950	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000417896	?	ENSP00000388116	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000433104	?	ENSP00000396904	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000446860	NM_001190317.1	ENSP00000399052	G/D	gGt/gAt	?	?	1
QRICH1	missense variant	ENST00000424300	?	ENSP00000412890	S/N	aGt/aAt	?	?	1
QRICH1	missense variant	ENST00000395443	NM_198880.1	ENSP00000378830	S/N	aGt/aAt	?	?	1
QRICH1	missense variant	ENST00000357496	NM_017730.2	ENSP00000350094	S/N	aGt/aAt	?	?	1
USP19	missense variant	ENST00000453664	NM_001199162.1	ENSP00000400090	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000434032	NM_001199160.1	ENSP00000401197	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000398898	?	ENSP00000381872	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000398888	NM_006677.2	ENSP00000381863	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000398892	?	ENSP00000381867	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000417901	NM_001199161.1	ENSP00000395260	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000306026	?	ENSP00000303503	H/D	Cat/Gat	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
WDR6	?	ENSG00000178252	visceral adipocytes	1.53×10-7 (p-value)	GTEx Portal V6	105
NCKIPSD	?	ENSG00000213672	visceral adipocytes	2.83×10-7 (p-value)	GTEx Portal V6	69
WDR6	?	ENSG00000178252	stomach	2.40×10-7 (p-value)	GTEx Portal V6	103
CCDC36	?	ENSG00000173421	muscularis mucosae	1.49×10-7 (p-value)	GTEx Portal V6	92

KLHDC8B	?	ENSG00000185909	muscularis mucosae	8.00×10-6 (p-value)	GTEx Portal V6	10
WDR6	?	ENSG00000178252	lung	1.44×10-10 (p-value)	GTEx Portal V6	107
SLC26A6	?	ENSG00000225697	lung	9.39×10-8 (p-value)	GTEx Portal V6	105
CELSR3	?	ENSG00000008300	lung	2.18×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	cortex	1.66×10-6 (p-value)	GTEx Portal V6	69
WDR6	?	ENSG00000178252	tibial nerve	7.26×10-13 (p-value)	GTEx Portal V6	107
SEMA3F	?	ENSG00000001617	tibial nerve	1.42×10-6 (p-value)	GTEx Portal V6	24
WDR6	?	ENSG00000178252	transformed fibroblasts	7.13×10-18 (p-value)	GTEx Portal V6	107
CCDC36	?	ENSG00000173421	transformed fibroblasts	2.32×10-7 (p-value)	GTEx Portal V6	95
NCKIPSD	?	ENSG00000213672	transformed fibroblasts	3.12×10-8 (p-value)	GTEx Portal V6	95
IP6K2	?	ENSG00000068745	transformed fibroblasts	1.54×10-7 (p-value)	GTEx Portal V6	27
RP11-3B7.1	?	ENSG00000225399	transformed fibroblasts	4.31×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	tibial artery	3.97×10-9 (p-value)	GTEx Portal V6	107
TMEM89	?	ENSG00000183396	tibial artery	1.39×10-7 (p-value)	GTEx Portal V6	105
NICN1	?	ENSG00000145029	tibial artery	5.92×10-8 (p-value)	GTEx Portal V6	78
IP6K2	?	ENSG00000068745	tibial artery	6.46×10-7 (p-value)	GTEx Portal V6	27
AMT	?	ENSG00000145020	tibial artery	6.08×10-7 (p-value)	GTEx Portal V6	78
CCDC36	?	ENSG00000173421	tibial artery	4.02×10-5 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	blood	8.00×10-12 (p-value)	GTEx Portal V6	107
WDR6	?	ENSG00000178252	esophagus mucosa	7.00×10-34 (p-value)	GTEx Portal V6	107
NCKIPSD	?	ENSG00000213672	esophagus mucosa	8.96×10-9 (p-value)	GTEx Portal V6	107
CCDC36	?	ENSG00000173421	testis	8.22×10-8 (p-value)	GTEx Portal V6	106
WDR6	?	ENSG00000178252	skeletal muscle	2.27×10-13 (p-value)	GTEx Portal V6	107
SLC26A6	?	ENSG00000225697	skeletal muscle	3.53×10-10 (p-value)	GTEx Portal V6	73
NCKIPSD	?	ENSG00000213672	skeletal muscle	1.01×10-5 (p-value)	GTEx Portal V6	20
TMA7	?	ENSG00000232112	skeletal muscle	1.82×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	unexposed skin	1.27×10-26 (p-value)	GTEx Portal V6	107
WDR6	?	ENSG00000178252	sun exposed skin	7.41×10-50 (p-value)	GTEx Portal V6	107
RBM6	?	ENSG00000004534	sun exposed skin	1.41×10-5 (p-value)	GTEx Portal V6	38
WDR6	?	ENSG00000178252	subcutaneous adipocytes	2.01×10-21 (p-value)	GTEx Portal V6	107
ARIH2	?	ENSG00000177479	subcutaneous adipocytes	9.11×10-6 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	breast	2.94×10-7 (p-value)	GTEx Portal V6	75
WDR6	?	ENSG00000178252	EBV lymphocytes	9.15×10-7 (p-value)	GTEx Portal V6	27
SLC26A6	?	ENSG00000225697	EBV lymphocytes	6.39×10-6 (p-value)	GTEx Portal V6	27
IP6K2	?	ENSG00000068745	aorta	1.01×10-5 (p-value)	GTEx Portal V6	25
NICN1	?	ENSG00000145029	aorta	5.04×10-5 (p-value)	GTEx Portal V6	1

CCDC71	?	ENSG00000177352	cerebellar hemisphere	5.33×10^{-6} (p-value)	GTEx Portal V6	26
WDR6	ENST00000615452	ILMN_1669484	skin	7.40×10^{-45} (p-value)	MuTHER consortium	15
WDR6	ENST00000452875		adipocyte	8.90×10^{-42} (p-value)	MuTHER consortium	15
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	skin	1.46×10^{-7} (p-value)	MuTHER consortium	15
USP4	ENST00000483212		blood	7.52×10^{-5} (p-value)	MuTHER consortium	15
USP4	ENST00000485450		adipocyte	1.06×10^{-12} (p-value)	MuTHER consortium	15
C3orf62	ENST00000343010					
TMEM89	ENST00000330862	ILMN_1694339	skin	1.23×10^{-6} (p-value)	MuTHER consortium	14
RP11-3B7.1	ENST00000622888	ILMN_1807244	adipocyte	1.03×10^{-19} (p-value)	MuTHER consortium	15
RP11-3B7.1	ENST00000440528					
C3orf62	ENST00000343010	ILMN_1679339	blood	1.87×10^{-5} (p-value)	MuTHER consortium	7
C3orf62	ENST00000479673					
RBM6	ENST00000266022	ILMN_1738239	adipocyte	2.62×10^{-5} (p-value)	MuTHER consortium	2
RBM6	ENST00000442092					
RBM6	ENST00000454079					
RBM6	ENST00000419610					
RBM6	ENST00000422955					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
WDR6	ENST00000615452	ILMN_1669484	monocyte	1.45×10^{-16} (p-value)	Fairfax et al.	16
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
RBM6	ENST00000442092	ILMN_1738239	monocyte	8.89×10^{-5} (p-value)	Fairfax et al.	2
RBM6	ENST00000266022		b-cell	5.55×10^{-5} (p-value)	Fairfax et al.	2
RBM6	ENST00000454079					
RBM6	ENST00000422955					
RBM6	ENST00000419610					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
UBA7	ENST00000497908	ILMN_1794612	b-cell	6.47×10^{-5} (p-value)	Fairfax et al.	16
UBA7	ENST00000333486					
MIR5193	ENST00000584510					
USP4	ENST00000351842	ILMN_1798712	monocyte	1.23×10^{-11} (p-value)	Fairfax et al.	16
USP4	ENST00000483212		b-cell	1.99×10^{-8} (p-value)	Fairfax et al.	16
C3orf62	ENST00000343010					
USP4	ENST00000485450					
SLC26A6	ENST00000420764	ILMN_1785252	b-cell	3.59×10^{-5} (p-value)	Fairfax et al.	16
SLC26A6	ENST00000395550					
SLC26A6	ENST00000489483					

SLC26A6	ENST00000480524					
SLC26A6	ENST00000455886					
SLC26A6	ENST00000307364					
SLC26A6	ENST00000614797					
SLC26A6	ENST00000358747					
SLC26A6	ENST00000383733					
SLC26A6	ENST00000337000					
CCDC71	ENST00000321895	ILMN_1768433	monocyte	3.61×10^{-4} (p-value)	Fairfax et al.	1
			b-cell	2.91×10^{-4} (p-value)	Fairfax et al.	1
WDR6	?	ENSG00000178252	pancreas	3.37×10^{-5} (p-value)	GTEx Portal V6	1
WDR6	ENST00000615452	ILMN_1669484	monocyte	1.21×10^{-38} (p-value)	Zeller et al.	4
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	monocyte	6.52×10^{-34} (p-value)	Zeller et al.	4
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
WDR6	?	ENSG00000178252	terminal ileum	4.57×10^{-6} (p-value)	GTEx Portal V6	6
ARIH2	ENST00000356401	ILMN_1792825	blood	5.24×10^{-5} (p-value)	Westra et al.	3
RBM6	?	ENSG00000004534	thyroid	3.30×10^{-5} (p-value)	GTEx Portal V6	19

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000609678	2	ENST00000332780, ENST00000459846	KLHDC8B

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000036427		
ENCP00000036447		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000338847	1	ENCP00000036433	ARIH2
ENCE00000338585	1	ENCP00000036360	SPINK8
		ENCP00000036439	ARIH2
		ENCP00000036380	ATRIP
		ENCP00000036426	IP6K2
ENCE00000338754	1	ENCP00000036427	PRKAR2A
		ENCP00000036409	SLC26A6
ENCE00000338989	1	ENCP00000036489	CCDC36
ENCE00000338724	1	ENCP00000036395	UCN2

ENCE00000338727	1	ENCP00000036460	IMPDH2
		ENCP00000036397	COL7A1 COL7A1
		ENCP00000036451	DALRD3
ENCE00000338767	2	ENCP00000036414	CELSR3
		ENCP00000036418	NCKIPSD
		ENCP00000036450	WDR6
		ENCP00000036472	QARS
		ENCP00000036507	AMT AMT
		ENCP00000036475	USP19
ENCE00000338922	1	ENCP00000036453	DALRD3
ENCE00000338903	1	ENCP00000036445	P4HTM

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001478981 (promoter flanking region)	3	embryonic stem cell (H1ESC)	PolII, H3K9ac, H3K27ac, H3K4me2, H3K36me3, DNase1, H3K27me3
		HSMMtube	H3K79me2, H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H3K79me2, H3K27ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3, H3K27ac, DNase1
		blood (GM12878)	PolII, H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K27ac, H3K4me1
		NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, H3K4me1, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K4me1, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001363512 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3, H3K79me2
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	H3K79me2, CTCF, H3K36me3
		blood (GM12878)	H3K36me3, H3K79me2
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001363515 (promoter flanking region)	1	NHLF	H3K4me1, H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1, DNase1
		muscle (HSMM)	H3K79me2, H3K36me3
		breast (HMEC)	H3K4me2, H3K4me1, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K36me3, H3K79me2
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K79me2, H3K36me3

		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H4K20me1, H3K79me2, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K4me1, DNase1, H3K36me3, H3K4me2, H3K27ac
ENSR00001363517 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, TAF7, H3K27ac, Yy1, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3
		HSMMtube	DNase1, H3K79me2, H3K9ac, H3K4me2, H3K4me3, H3K36me3
		blood (K562)	HEY1, H2AZ, TAF1, ELF1, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, H3K79me2, H3K9ac, Yy1, H3K27ac, Max
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		liver (HepG2)	DNase1, H3K36me3, H3K27me3, H3K4me3, H3K27ac, Gabp, Yy1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, DNase1, H3K79me2
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K27ac, H3K36me3, H3K9ac, DNase1, H3K4me3
		Osteobl	H3K27me3, H3K36me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	DNase1, H3K36me3, H3K27me3, PolII, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001363520 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H3K4me2
ENSR00001363530 (open chromatin region)	1	endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (K562)	DNase1
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00001478987 (open chromatin region)	1	muscle (HSMM)	DNase1
ENSR00001363541 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, H3K4me2, H3K4me3
		HSMMtube	H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K27ac, Max, H3K9ac, H3K79me2, H2AZ, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K36me3, DNase1
		liver (HepG2)	PolII, H3K79me2, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K4me2, H4K5ac, H3K36me3, H3K4me3
		blood (GM12878)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, PolII, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K36me3, H3K4me2, H3K4me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	PolII, DNase1, H3K79me2, H3K4me3, H3K27ac, Ini1, Nrf1, H3K9ac, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me2, PolII, DNase1, Cmyc, H3K4me3, Max
		A549	H3K4me3, H3K4me2, H3K9ac, H3K36me3
ENSR00001674797 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
ENSR00001674799 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
ENSR00001478995 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		blood (K562)	Egr1, H3K79me2, H3K36me3
		muscle (HSMM)	H3K79me2, H3K36me3
		liver (HepG2)	H3K79me2, H3K36me3
		blood (GM12878)	BATF, H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3, H3K4me1
		NHLF	H3K36me3

		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001363566 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K36me3, CTCF, PolII, Rad21, TAF7, H3K27ac, ATF3, Yy1, TAF1, H3K4me2, SP1, USF1, H3K9ac, H3K4me3
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H2AZ, Max, Cfos, PolII, H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, Brg1, Egr1, H3K27ac, ZBTB33, H3K9ac, USF1, Cmyc, H3K79me2, HEY1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	DNase1, PolII, H3K79me2, TAF1, USF1, H3K4me1, ZBTB33, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K36me3, H3K27me3
		blood (GM12878)	PolII, H2AZ, DNase1, Egr1, CTCF, H3K79me2, H3K4me3, USF1, ZBTB33, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	H3K4me3, H3K9ac, CTCF, H3K36me3, H4K5ac, DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2
		nervous (NH-A)	DNase1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me2, CTCF, H3K9ac, DNase1, H3K27ac, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, H3K9ac, PolII, DNase1
		A549	H3K36me3, H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00001363576 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, Rad21, Egr1, TAF7, Sin3Ak20, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3
		HSMMtube	CTCF, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, DNase1
		blood (K562)	ZBTB33, Max, Rad21, H3K9ac, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3, Yy1, Egr1, H3K27ac
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, CTCF, H3K4me3
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, Yy1, PolII, H3K79me2, Rad21, TAF1, ZBTB33, H3K4me2, H3K9ac, H3K27ac, CTCF, H3K4me3, H3K36me3
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, CTCF
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, Egr1, Yy1, H3K79me2, H3K4me3, ZBTB33, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K36me3, H3K27ac, DNase1, H3K4me3, CTCF, H3K9ac
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K36me3, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, PolII, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1
ENSR00001479006 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	DNase1
		blood (DND-41)	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	PolII, CTCF
		liver (HepG2)	HNF4G, Rad21, HNF4A, CTCF
		blood (GM12878)	Tr4, PolII, Rad21, CTCF
		skin (NHEK)	CTCF
ENSR00001363588 (promoter)	2	embryonic stem cell (H1ESC)	H3K27ac, Rad21, CTCF, H3K4me2, H3K4me3, DNase1
		HSMMtube	DNase1, H3K36me3, CTCF, H3K9ac, H3K4me2, H3K4me3, H2AZ

		blood (K562)	H3K27ac, Max, H3K4me1, Rad21, H3K9ac, Cmyc, H3K79me2, H2AZ, PolII, H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, Ini1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, H3K36me3, DNase1
		liver (HepG2)	PolII, Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, CTCF
		blood (GM12878)	DNase1, H2AZ, Yy1, Rad21, H3K4me3, H3K4me2, CTCF
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, DNase1
		skin (NHEK)	H3K36me3, CTCF, H3K4me3, H3K4me2, H3K9ac, DNase1
		NHLF	DNase1, H3K4me3, CTCF, H3K9ac, H3K36me3
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	CTCF
		breast (HMEC)	H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K4me3
		monocytes (Monocytes-CD14+)	CTCF, H3K4me2, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1, PolII, H3K4me2, Max, H3K4me3
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF
ENSR00001479009	1	NHLF	DNase1
(promoter flanking region)		HSMMtube	H3K27me3
		Osteobl	H3K36me3
		blood (K562)	DNase1, Nfe2
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Jund, Max
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	FOSL2, Jund, DNase1, H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	DNase1, H3K36me3
		skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC141002.1	upstream gene variant, downstream gene variant	1996	ENST00000619404	?	?	7
ARIH2	upstream gene variant, downstream gene variant	1458	ENST00000492077	?	?	4
ARIH2	upstream gene variant	2504	ENST00000488963	?	?	3
ARIH2	upstream gene variant, downstream gene variant	2520	ENST00000433170	?	ENSP00000406063	4
ARIH2	upstream gene variant	2477	ENST00000478224	?	?	3
ARIH2	upstream gene variant	2935	ENST00000487891	?	?	1
ARIH2	upstream gene variant	2479	ENST00000498314	?	?	3
ARIH2	downstream gene variant	4664	ENST00000469038	?	?	1
ARIH2	upstream gene variant	2512	ENST00000486316	?	?	3
ARIH2	downstream gene variant	2338	ENST00000470296	?	?	1
ARIH2	upstream gene variant	2492	ENST00000449376	?	ENSP00000403222	3
ARIH2	upstream gene variant	3230	ENST00000465217	?	?	1
ARIH2	upstream gene variant, downstream gene variant	162	ENST00000490095	?	?	4
ARIH2	upstream gene variant	942	ENST00000459976	?	?	1
ARIH2	upstream gene variant, downstream gene variant	1593	ENST00000463738	?	?	4
ARIH2	upstream gene variant	4358	ENST00000482342	?	?	1
ARIH2	upstream gene variant	2500	ENST00000482427	?	?	3

ARIH2	upstream gene variant	2986	ENST00000472640	?	?	2
ARIH2	upstream gene variant, downstream gene variant	2297	ENST00000452882	?	ENSP00000395560	4
ARIH2	downstream gene variant	3647	ENST00000466850	?	?	1
ARIH2	upstream gene variant	2485	ENST00000474618	?	?	3
ARIH2	upstream gene variant	423	ENST00000463204	?	?	1
ARIH2	upstream gene variant, downstream gene variant	2513	ENST00000449729	?	ENSP00000404838	4
ARIH2	upstream gene variant	43	ENST00000452385	?	ENSP00000403845	2
ARIH2	upstream gene variant	2551	ENST00000474936	?	?	3
ARIH2	upstream gene variant	1104	ENST00000495507	?	?	2
ARIH2	upstream gene variant	2477	ENST00000356401	NM_006321.2	ENSP00000348769	3
ARIH2	upstream gene variant, downstream gene variant	2297	ENST00000420814	?	ENSP00000397225	4
ARIH2	upstream gene variant, downstream gene variant	2498	ENST00000495761	?	?	4
ARIH2	upstream gene variant	2477	ENST00000430423	?	ENSP00000399788	3
ARIH2	upstream gene variant	2500	ENST00000483333	?	?	3
ARIH2OS	downstream gene variant, upstream gene variant	1444	ENST00000408959	NM_001123040.1	ENSP00000386193	4
C3orf84	downstream gene variant, upstream gene variant	1186	ENST00000443990	?	?	4
C3orf84	downstream gene variant, upstream gene variant	202	ENST00000432035	?	ENSP00000455944	4
C3orf84	downstream gene variant, upstream gene variant	166	ENST00000545770	NM_001080528.2	ENSP00000454903	4
CCDC36	upstream gene variant	3314	ENST00000438782	?	ENSP00000391788	1
CCDC36	upstream gene variant	3927	ENST00000452691	NM_001135197.1	ENSP00000407837	1
CCDC36	upstream gene variant	3937	ENST00000366429	?	ENSP00000403700	1
CCDC36	upstream gene variant	4044	ENST00000493870	?	?	1
CCDC36	upstream gene variant	2855	ENST00000296449	NM_178173.3	ENSP00000296449	1
CCDC71	downstream gene variant, upstream gene variant	3339	ENST00000321895	NM_022903.3	ENSP00000319006	3
DALRD3	upstream gene variant	4386	ENST00000492585	?	?	1
DALRD3	upstream gene variant	4384	ENST00000496568	?	ENSP00000485305	1
IMPDH2	upstream gene variant	1093	ENST00000442157	?	ENSP00000403502	2
IMPDH2	upstream gene variant	1998	ENST00000429182	?	ENSP00000393525	1
IMPDH2	upstream gene variant	2329	ENST00000462980	?	?	1
IMPDH2	upstream gene variant	2344	ENST00000491610	?	?	1
IMPDH2	upstream gene variant	1701	ENST00000472328	?	?	1
IMPDH2	downstream gene variant, upstream gene variant	1063	ENST00000496837	?	?	3
IMPDH2	upstream gene variant	1434	ENST00000484872	?	?	1
IMPDH2	downstream gene variant, upstream gene variant	1034	ENST00000485500	?	?	3
IMPDH2	upstream gene variant	1839	ENST00000463903	?	?	1
IMPDH2	upstream gene variant	1262	ENST00000481274	?	?	1
IMPDH2	upstream gene variant	1081	ENST00000326739	NM_000884.2	ENSP00000321584	2
IMPDH2	upstream gene variant	1484	ENST00000466147	?	?	1

IMPDH2	upstream gene variant	1484	ENST00000466147	?	?	1
IP6K2	downstream gene variant, upstream gene variant	195	ENST00000412795	?	ENSP00000389755	4
IP6K2	downstream gene variant	1429	ENST00000433104	?	ENSP00000396904	1
IP6K2	downstream gene variant, upstream gene variant	229	ENST00000412850	?	ENSP00000395819	4
IP6K2	downstream gene variant, upstream gene variant	228	ENST00000431721	NM_001190316.1	ENSP00000414139	4
IP6K2	downstream gene variant, upstream gene variant	83	ENST00000434860	?	ENSP00000388420	6
IP6K2	downstream gene variant, upstream gene variant	251	ENST00000476645	?	?	8
IP6K2	downstream gene variant, upstream gene variant	241	ENST00000416707	?	ENSP00000387759	4
IP6K2	downstream gene variant, upstream gene variant	81	ENST00000440424	?	ENSP00000393797	6
IP6K2	downstream gene variant, upstream gene variant	122	ENST00000424035	?	ENSP00000406357	6
IP6K2	downstream gene variant, upstream gene variant	238	ENST00000417896	?	ENSP00000388116	4
IP6K2	downstream gene variant, upstream gene variant	256	ENST00000454335	?	ENSP00000412121	4
IP6K2	downstream gene variant, upstream gene variant	1915	ENST00000455545	?	ENSP00000410454	4
IP6K2	downstream gene variant, upstream gene variant	166	ENST00000432678	NM_001146178.2, NM_001146179.2	ENSP00000400812	4
IP6K2	downstream gene variant, upstream gene variant	234	ENST00000413298	?	ENSP00000396203	4
IP6K2	downstream gene variant, upstream gene variant	278	ENST00000413654	?	ENSP00000414428	4
IP6K2	downstream gene variant, upstream gene variant	252	ENST00000443964	?	ENSP00000410950	4
IP6K2	downstream gene variant, upstream gene variant	82	ENST00000453202	?	ENSP00000387394	5
IP6K2	downstream gene variant, upstream gene variant	256	ENST00000436134	?	?	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000449563	?	ENSP00000411776	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000446860	NM_001190317.1	ENSP00000399052	4
IP6K2	downstream gene variant, upstream gene variant	252	ENST00000449610	?	ENSP00000393077	4
IP6K2	downstream gene variant, upstream gene variant	194	ENST00000328631	NM_001005909.2, NM_016291.3	ENSP00000331103	4
IP6K2	downstream gene variant, upstream gene variant	1957	ENST00000450045	?	ENSP00000394488	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000443853	?	ENSP00000389761	4
IP6K2	downstream gene variant, upstream gene variant	1455	ENST00000491686	?	?	5
IP6K2	downstream gene variant, upstream gene variant	228	ENST00000340879	NM_001005910.2, NM_001005911.2	ENSP00000341925	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000437427	?	ENSP00000403968	4
IP6K2	downstream gene variant, upstream gene variant	486	ENST00000479914	?	?	4
KLHDC8B	upstream gene variant, downstream gene variant	1195	ENST00000332780	NM_173546.2	ENSP00000327468	4
KLHDC8B	upstream gene variant, downstream gene variant	631	ENST00000476495	?	?	7
KLHDC8B	upstream gene variant, downstream gene variant	411	ENST00000471811	?	?	8
KLHDC8B	upstream gene variant, downstream gene variant	430	ENST00000459846	?	?	5
KLHDC8B	upstream gene variant, downstream gene variant	1533	ENST00000462582	?	?	6
LAMB2	downstream gene variant	4680	ENST00000480640	?	?	1
LAMB2	downstream gene variant	2075	ENST00000484713	?	?	1
LAMB2	downstream gene variant	4919	ENST00000538659	?	?	1

LAMB2	downstream gene variant	2074	ENST00000467506	?	?	1
LAMB2	downstream gene variant	2217	ENST00000498377	?	?	1
LAMB2	upstream gene variant	3867	ENST00000494831	?	ENSP00000444751	1
LAMB2	downstream gene variant, upstream gene variant	2093	ENST00000305544	?	ENSP00000307156	2
LAMB2	downstream gene variant	4285	ENST00000477225	?	?	1
LAMB2	downstream gene variant, upstream gene variant	2074	ENST00000418109	NM_002292.3	ENSP00000388325	2
LAMB2	downstream gene variant	3454	ENST00000469665	?	?	1
MIR6890	downstream gene variant, upstream gene variant	2059	ENST00000620931	?	?	4
NCKIPSD	upstream gene variant	1697	ENST00000416649	NM_184231.2	ENSP00000389059	1
NCKIPSD	upstream gene variant	1220	ENST00000453349	?	ENSP00000408588	1
NCKIPSD	upstream gene variant	1654	ENST00000454134	?	ENSP00000416144	1
NCKIPSD	upstream gene variant	1746	ENST00000439518	?	ENSP00000409675	1
NCKIPSD	upstream gene variant	1654	ENST00000294129	NM_016453.3	ENSP00000294129	1
NCKIPSD	upstream gene variant	1217	ENST00000426678	?	ENSP00000416904	1
NDUFAF3	downstream gene variant	3390	ENST00000326912	NM_199074.1	ENSP00000323003	1
NDUFAF3	downstream gene variant	3182	ENST00000326925	NM_199069.1	ENSP00000323076	1
NDUFAF3	downstream gene variant	3501	ENST00000480392	?	?	1
NDUFAF3	downstream gene variant	3390	ENST00000496152	?	?	1
NDUFAF3	downstream gene variant	3390	ENST00000395458	NM_199073.1	ENSP00000378843	1
NDUFAF3	downstream gene variant	3389	ENST00000451378	NM_199070.1	ENSP00000402465	1
P4HTM	downstream gene variant	2685	ENST00000472796	?	ENSP00000476858	1
P4HTM	downstream gene variant	165	ENST00000491739	?	ENSP00000476303	1
P4HTM	downstream gene variant	165	ENST00000484115	?	?	1
P4HTM	downstream gene variant	2719	ENST00000609406	?	?	1
P4HTM	downstream gene variant	165	ENST00000343546	NM_177938.2	ENSP00000341422	1
P4HTM	downstream gene variant	4304	ENST00000485210	?	?	1
P4HTM	downstream gene variant	126	ENST00000383729	NM_177939.2	ENSP00000373235	1
P4HTM	downstream gene variant	3017	ENST00000444213	?	ENSP00000398554	1
P4HTM	upstream gene variant, downstream gene variant	1180	ENST00000486817	?	?	2
P4HTM	downstream gene variant	3085	ENST00000468374	?	?	1
P4HTM	downstream gene variant	165	ENST00000472301	?	?	1
PRKAR2A	upstream gene variant	700	ENST00000437821	?	ENSP00000400840	2
PRKAR2A	upstream gene variant	1748	ENST00000265563	NM_004157.2	ENSP00000265563	1
PRKAR2A	upstream gene variant	1790	ENST00000454963	?	ENSP00000394041	1
PRKAR2A	upstream gene variant	1761	ENST00000419216	?	ENSP00000411432	1
PRKAR2A	upstream gene variant	1766	ENST00000296446	?	ENSP00000296446	1
PRKAR2A-AS1	upstream gene variant, downstream gene variant	158	ENST00000431705	?	?	2

PRKAR2A-AS1	downstream gene variant	1101	ENST00000435419	?	?	1
PRKAR2A-AS1	upstream gene variant, downstream gene variant	140	ENST00000412171	?	?	2
PRKAR2A-AS1	upstream gene variant, downstream gene variant	178	ENST00000416209	?	?	2
QARS	downstream gene variant, upstream gene variant	940	ENST00000470225	?	?	2
QARS	downstream gene variant	730	ENST00000417025	?	?	1
QARS	downstream gene variant	693	ENST00000418549	?	ENSP00000415247	1
QARS	downstream gene variant, upstream gene variant	1306	ENST00000478561	?	?	4
QARS	downstream gene variant	4624	ENST00000452739	?	ENSP00000392850	1
QARS	downstream gene variant	332	ENST00000482438	?	?	2
QARS	downstream gene variant	634	ENST00000479495	?	?	1
QARS	downstream gene variant	4832	ENST00000482261	?	?	1
QARS	downstream gene variant, upstream gene variant	73	ENST00000494767	?	?	3
QARS	downstream gene variant	226	ENST00000470619	?	?	1
QARS	downstream gene variant, upstream gene variant	1552	ENST00000482468	?	?	4
QARS	downstream gene variant, upstream gene variant	827	ENST00000459870	?	?	3
QARS	downstream gene variant	818	ENST00000464962	?	?	1
QARS	downstream gene variant	812	ENST00000430182	?	ENSP00000389823	1
QARS	downstream gene variant, upstream gene variant	813	ENST00000494984	?	?	3
QARS	downstream gene variant	3193	ENST00000494838	?	?	1
QARS	downstream gene variant, upstream gene variant	873	ENST00000466179	?	?	3
QARS	downstream gene variant, upstream gene variant	812	ENST00000487495	?	?	2
QARS	downstream gene variant, upstream gene variant	811	ENST00000453392	?	ENSP00000396326	3
QARS	downstream gene variant, upstream gene variant	812	ENST00000475599	?	?	2
QARS	downstream gene variant, upstream gene variant	811	ENST00000482248	?	?	3
QARS	downstream gene variant	816	ENST00000414533	NM_001272073.1	ENSP00000390015	1
QARS	downstream gene variant, upstream gene variant	812	ENST00000497635	?	?	2
QARS	downstream gene variant, upstream gene variant	840	ENST00000470113	?	?	3
QARS	downstream gene variant	811	ENST00000306125	NM_005051.2	ENSP00000307567	1
QRICH1	downstream gene variant	2612	ENST00000479449	?	?	1
QRICH1	downstream gene variant, upstream gene variant	411	ENST00000437939	?	ENSP00000416133	6
QRICH1	downstream gene variant, upstream gene variant	849	ENST00000469910	?	?	3
QRICH1	downstream gene variant, upstream gene variant	411	ENST00000450685	?	ENSP00000413051	6
QRICH1	downstream gene variant, upstream gene variant	411	ENST00000411682	?	ENSP00000412870	6
QRICH1	downstream gene variant, upstream gene variant	577	ENST00000430979	?	ENSP00000405505	6
QRICH1	downstream gene variant, upstream gene variant	816	ENST00000477021	?	?	3
QRICH1	downstream gene variant, upstream gene variant	758	ENST00000395443	NM_198880.1	ENSP00000378830	3
QRICH1	downstream gene variant, upstream gene variant	2576	ENST00000498440	?	?	3

QRICH1	downstream gene variant, upstream gene variant	187	ENST00000498392	?	?	4
QRICH1	downstream gene variant, upstream gene variant	1059	ENST00000357496	NM_017730.2	ENSP00000350094	3
QRICH1	downstream gene variant	3698	ENST00000489642	?	?	1
QRICH1	downstream gene variant, upstream gene variant	1510	ENST00000424300	?	ENSP00000412890	3
RN7SL182P	upstream gene variant, downstream gene variant	3852	ENST00000496499	?	?	2
RP11-694I15.7	downstream gene variant	3163	ENST00000603877	?	?	1
RP13-131K19.1	downstream gene variant	143	ENST00000415982	?	?	1
RP13-131K19.1	downstream gene variant	157	ENST00000429681	?	?	1
RP13-131K19.2	upstream gene variant	4960	ENST00000452042	?	?	1
RP13-131K19.6	upstream gene variant, downstream gene variant	765	ENST00000607245	?	?	3
SLC25A20	downstream gene variant	1228	ENST00000430379	?	ENSP00000388986	1
SLC25A20	downstream gene variant	1233	ENST00000319017	NM_000387.5	ENSP00000326305	1
SLC25A20	downstream gene variant	1689	ENST00000440964	?	ENSP00000388563	1
SLC25A20	downstream gene variant	1538	ENST00000479050	?	?	1
USP19	downstream gene variant, upstream gene variant	1062	ENST00000398896	?	ENSP00000381870	2
USP19	downstream gene variant	4367	ENST00000417901	NM_001199161.1	ENSP00000395260	1
USP19	downstream gene variant	4363	ENST00000398898	?	ENSP00000381872	1
USP19	downstream gene variant	4990	ENST00000398892	?	ENSP00000381867	1
USP19	downstream gene variant	4992	ENST00000398888	NM_006677.2	ENSP00000381863	1
USP19	upstream gene variant	1329	ENST00000464931	?	?	1
USP19	upstream gene variant	1252	ENST00000480163	?	?	1
USP19	downstream gene variant	4448	ENST00000453664	NM_001199162.1	ENSP00000400090	1
USP19	upstream gene variant	1682	ENST00000479073	?	ENSP00000428539	1
WDR6	upstream gene variant	4354	ENST00000615452	?	ENSP00000482289	1
WDR6	upstream gene variant	117	ENST00000452875	?	ENSP00000414157	1
WDR6	upstream gene variant	152	ENST00000627177	?	ENSP00000487056	1
WDR6	upstream gene variant	123	ENST00000471162	?	?	1
WDR6	upstream gene variant	113	ENST00000438660	?	ENSP00000387692	1
WDR6	upstream gene variant	331	ENST00000419837	?	ENSP00000390740	1
WDR6	upstream gene variant	328	ENST00000472878	?	?	1
WDR6	upstream gene variant	82	ENST00000488572	?	?	1
WDR6	upstream gene variant	73	ENST00000429900	?	ENSP00000477418	1
WDR6	upstream gene variant	113	ENST00000608424	?	ENSP00000477389	1
WDR6	upstream gene variant	345	ENST00000461687	?	?	1
WDR6	upstream gene variant	113	ENST00000473238	?	?	1
WDR6	upstream gene variant	123	ENST00000420783	?	ENSP00000395993	1
WDR6	upstream gene variant	110	ENST00000462064	?	?	1

WDR6	upstream gene variant	116	ENST00000415265 ?	ENSP00000412195	1
WDR6	upstream gene variant	328	ENST00000448293 ?	ENSP00000413432	1
WDR6	upstream gene variant	786	ENST00000491365 ?	ENSP00000477463	1
WDR6	upstream gene variant	781	ENST00000489427 ?	ENSP00000477087	1
WDR6	upstream gene variant	110	ENST00000489684 ?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
C3orf84	ENST00000545770	NM_001080528.2	ENSP00000454903	P	ccA/ccG	1
C3orf84	ENST00000432035	?	ENSP00000455944	P	ccA/ccG	1
QARS	ENST00000306125	NM_005051.2	ENSP00000307567	H	caT/caC	1
QARS	ENST00000414533	NM_001272073.1	ENSP00000390015	H	caT/caC	1
QARS	ENST00000452739	?	ENSP00000392850	H	caT/caC	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
KLHDC8B	ENST00000332780	NM_173546.2	ENSP00000327468	1
KLHDC8B	ENST00000462582	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ARIH2	ENST00000495507	?	?	4
ARIH2	ENST00000472640	?	?	1
ARIH2	ENST00000449729	?	ENSP00000404838	2
ARIH2	ENST00000495761	?	?	7
ARIH2	ENST00000452385	?	ENSP00000403845	6
ARIH2	ENST00000484999	?	?	7
ARIH2	ENST00000463204	?	?	5
ARIH2	ENST00000498314	?	?	7
ARIH2	ENST00000466850	?	?	2
ARIH2	ENST00000474936	?	?	7
ARIH2	ENST00000420814	?	ENSP00000397225	6
ARIH2	ENST00000463738	?	?	1
ARIH2	ENST00000433170	?	ENSP00000406063	2
ARIH2	ENST00000482427	?	?	7
ARIH2	ENST00000430423	?	ENSP00000399788	7
ARIH2	ENST00000470296	?	?	6
ARIH2	ENST00000452882	?	ENSP00000395560	6
ARIH2	ENST00000490095	?	?	7
ARIH2	ENST00000449376	?	ENSP00000403222	8
ARIH2	ENST00000478224	?	?	7

ARIH2	ENST00000474618	?	?	7
ARIH2	ENST00000356401	NM_006321.2	ENSP00000348769	8
ARIH2	ENST00000488963	?	?	7
ARIH2	ENST00000486316	?	?	7
ARIH2	ENST00000492077	?	?	1
ARIH2	ENST00000483333	?	?	7
C3orf84	ENST00000443990	?	?	2
C3orf84	ENST00000545770	NM_001080528.2	ENSP00000454903	3
C3orf84	ENST00000432035	?	ENSP00000455944	3
CCDC36	ENST00000452691	NM_001135197.1	ENSP00000407837	12
CCDC36	ENST00000296449	NM_178173.3	ENSP00000296449	12
CCDC36	ENST00000438782	?	ENSP00000391788	12
CCDC36	ENST00000366429	?	ENSP00000403700	12
CCDC36	ENST00000493870	?	?	12
CELSR3	ENST00000164024	NM_001407.2	ENSP00000164024	1
IMPDH2	ENST00000326739	NM_000884.2	ENSP00000321584	1
IMPDH2	ENST00000462980	?	?	1
IMPDH2	ENST00000429182	?	ENSP00000393525	1
IMPDH2	ENST00000442157	?	ENSP00000403502	1
IMPDH2	ENST00000491610	?	?	1
IP6K2	ENST00000431721	NM_001190316.1	ENSP00000414139	17
IP6K2	ENST00000440424	?	ENSP00000393797	16
IP6K2	ENST00000432678	NM_001146178.2, NM_001146179.2	ENSP00000400812	18
IP6K2	ENST00000412850	?	ENSP00000395819	18
IP6K2	ENST00000436134	?	?	18
IP6K2	ENST00000434860	?	ENSP00000388420	16
IP6K2	ENST00000446860	NM_001190317.1	ENSP00000399052	17
IP6K2	ENST00000437427	?	ENSP00000403968	18
IP6K2	ENST00000433104	?	ENSP00000396904	27
IP6K2	ENST00000413298	?	ENSP00000396203	18
IP6K2	ENST00000449610	?	ENSP00000393077	17
IP6K2	ENST00000424035	?	ENSP00000406357	16
IP6K2	ENST00000476645	?	?	13
IP6K2	ENST00000454335	?	ENSP00000412121	18
IP6K2	ENST00000450045	?	ENSP00000394488	18
IP6K2	ENST00000412795	?	ENSP00000389755	18
IP6K2	ENST00000443853	?	ENSP00000389761	18
IP6K2	ENST00000413654	?	ENSP00000414428	18
IP6K2	ENST00000340879	NM_001005910.2, NM_001005911.2	ENSP00000341925	18
IP6K2	ENST00000449563	?	ENSP00000411776	18
IP6K2	ENST00000417896	?	ENSP00000388116	17
IP6K2	ENST00000491686	?	?	1

IP6K2	ENST00000455545	?	ENSP00000410454	18
IP6K2	ENST00000328631	NM_001005909.2, NM_016291.3	ENSP00000331103	19
IP6K2	ENST00000443964	?	ENSP00000410950	17
IP6K2	ENST00000416707	?	ENSP00000387759	18
IP6K2	ENST00000479914	?	?	3
KLHDC8B	ENST00000332780	NM_173546.2	ENSP00000327468	1
KLHDC8B	ENST00000459846	?	?	1
KLHDC8B	ENST00000476495	?	?	1
P4HTM	ENST00000468374	?	?	1
P4HTM	ENST00000472301	?	?	1
P4HTM	ENST00000475629	?	ENSP00000477191	1
P4HTM	ENST00000343546	NM_177938.2	ENSP00000341422	1
P4HTM	ENST00000444213	?	ENSP00000398554	1
P4HTM	ENST00000484115	?	?	1
P4HTM	ENST00000383729	NM_177939.2	ENSP00000373235	1
PRKAR2A	ENST00000265563	NM_004157.2	ENSP00000265563	8
PRKAR2A	ENST00000419216	?	ENSP00000411432	6
PRKAR2A	ENST00000454963	?	ENSP00000394041	8
PRKAR2A	ENST00000437821	?	ENSP00000400840	2
PRKAR2A	ENST00000296446	?	ENSP00000296446	8
QARS	ENST00000430182	?	ENSP00000389823	2
QARS	ENST00000494838	?	?	1
QARS	ENST00000470113	?	?	1
QARS	ENST00000453392	?	ENSP00000396326	1
QARS	ENST00000497635	?	?	1
QARS	ENST00000466179	?	?	1
QARS	ENST00000306125	NM_005051.2	ENSP00000307567	2
QARS	ENST00000475599	?	?	1
QARS	ENST00000482248	?	?	1
QARS	ENST00000459870	?	?	1
QARS	ENST00000414533	NM_001272073.1	ENSP00000390015	2
QARS	ENST00000487495	?	?	1
QARS	ENST00000464962	?	?	2
QARS	ENST00000494984	?	?	1
QARS	ENST00000470225	?	?	1
QARS	ENST00000452739	?	ENSP00000392850	1
QARS	ENST00000482261	?	?	1
QRICH1	ENST00000450685	?	ENSP00000413051	2
QRICH1	ENST00000437939	?	ENSP00000416133	2
QRICH1	ENST00000430979	?	ENSP00000405505	2
QRICH1	ENST00000411682	?	ENSP00000412870	2

QRICH1	ENST00000498440	?	?	3
QRICH1	ENST00000424300	?	ENSP00000412890	11
QRICH1	ENST00000395443	NM_198880.1	ENSP00000378830	11
QRICH1	ENST00000479449	?	?	5
QRICH1	ENST00000357496	NM_017730.2	ENSP00000350094	11
QRICH1	ENST00000489642	?	?	3
RP11-694I15.7	ENST00000603877	?	?	3
SLC25A20	ENST00000440964	?	ENSP00000388563	3
SLC25A20	ENST00000319017	NM_000387.5	ENSP00000326305	3
SLC25A20	ENST00000430379	?	ENSP00000388986	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IP6K2	ENST00000431721	NM_001190316.1	ENSP00000414139	1
IP6K2	ENST00000446860	NM_001190317.1	ENSP00000399052	1
IP6K2	ENST00000433104	?	ENSP00000396904	1
IP6K2	ENST00000449610	?	ENSP00000393077	1
IP6K2	ENST00000412795	?	ENSP00000389755	1
IP6K2	ENST00000417896	?	ENSP00000388116	1
IP6K2	ENST00000443964	?	ENSP00000410950	1
IP6K2	ENST00000416707	?	ENSP00000387759	1
QARS	ENST00000430182	?	ENSP00000389823	1
QARS	ENST00000418549	?	ENSP00000415247	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IP6K2	ENST00000433104	?	ENSP00000396904	1
KLHDC8B	ENST00000332780	NM_173546.2	ENSP00000327468	2
PRKAR2A	ENST00000265563	NM_004157.2	ENSP00000265563	1
PRKAR2A	ENST00000419216	?	ENSP00000411432	1
PRKAR2A	ENST00000454963	?	ENSP00000394041	1
PRKAR2A	ENST00000296446	?	ENSP00000296446	1
WDR6	ENST00000610967	NM_018031.3	ENSP00000484799	1
WDR6	ENST00000395474	?	ENSP00000378857	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C3orf84	ENST00000443990	?	1
IP6K2	ENST00000436134	?	1
KLHDC8B	ENST00000459846	?	2
PRKAR2A-AS1	ENST00000435419	?	1
PRKAR2A-AS1	ENST00000416209	?	1
QARS	ENST00000494838	?	1
QARS	ENST00000417025	?	1

QARS	ENST00000464962	?	1
QARS	ENST00000470619	?	1
QARS	ENST00000479495	?	1
QARS	ENST00000482261	?	1
QRICH1	ENST00000498392	?	1
QRICH1	ENST00000489642	?	1
USP19	ENST00000465902	?	1
USP19	ENST00000488993	?	1
USP19	ENST00000491859	?	1

Locus 96 (rs5030062)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,454,180-186,459,927
block size	5,748 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.444 [-0.367 – 1.999]	gene(s) hit or close-by	KNG1, RP11-573D15.8
phastCons	μ = 0.155 [0 – 0.667]	eQTL gene(s)	–
GERP++	μ = 0.739 [-0.875 – 5.61]	potentially regulated gene(s)	–
CADD score	μ = 5.384 [0.567 – 14.24]	disease gene(s)	KNG1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Plasma renin activity levels	$<9.00\times10^{-9}$	GWAS Catalog	25477429	1
Blood metabolite levels	$<6.00\times10^{-13}$	GWAS Catalog	24816252	1
Activated partial thromboplastin time	$<2.00\times10^{-203}$	GWAS Catalog	22703881	1
Activated partial thromboplastin time	$<1.00\times10^{-21}$	GWAS Catalog	20303064	1
bradykinin, des-arg(9)	5.94×10^{-13}	Metabolomics GWAS Server	24816252	4

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM112211	1

Disease gene annotation

gene	trait	source DB	source entry/link
KNG1	HIGH MOLECULAR WEIGHT KININOGEN DEFICIENCY	OMIM	MIM:228960
KNG1	Congenital high-molecular-weight kininogen deficiency	OrphaNet	OrphaNet:483

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
KNG1	missense variant	ENST00000265023	NM_001102416.2	ENSP00000265023	I/T	aTa/aCa	tolerated	benign	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	downstream gene variant	1586	ENST00000628928	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000609652	?	?	4
RP11-573D15.8	downstream gene variant	71	ENST00000631299	?	?	5

RP11-573D15.8	downstream gene variant	1586	ENST00000609726	?	?	4
RP11-573D15.8	downstream gene variant	25	ENST00000628831	?	?	1
RP11-573D15.8	downstream gene variant	1586	ENST00000625869	?	?	4
RP11-573D15.8	downstream gene variant	1567	ENST00000596632	?	?	4
RP11-573D15.8	downstream gene variant	116	ENST00000629734	?	?	1
RP11-573D15.8	downstream gene variant	1586	ENST00000625690	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000628291	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000626520	?	?	4
RP11-573D15.8	downstream gene variant	1565	ENST00000626586	?	?	4
RP11-573D15.8	downstream gene variant	1570	ENST00000596329	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000629201	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000626978	?	?	4
RP11-573D15.8	downstream gene variant	1586	ENST00000629954	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KNG1	ENST00000287611	NM_000893.3	ENSP00000287611	9
KNG1	ENST00000447445	NM_001166451.1	ENSP00000396025	9
KNG1	ENST00000265023	NM_001102416.2	ENSP00000265023	8
RP11-573D15.8	ENST00000627919	?	?	9
RP11-573D15.8	ENST00000629126	?	?	9
RP11-573D15.8	ENST00000628190	?	?	9
RP11-573D15.8	ENST00000629106	?	?	9
RP11-573D15.8	ENST00000625386	?	?	9
RP11-573D15.8	ENST00000626633	?	?	9
RP11-573D15.8	ENST00000627015	?	?	9
RP11-573D15.8	ENST00000626306	?	?	9
RP11-573D15.8	ENST00000626151	?	?	9
RP11-573D15.8	ENST00000627268	?	?	9
RP11-573D15.8	ENST00000599314	?	?	9
RP11-573D15.8	ENST00000630178	?	?	9
RP11-573D15.8	ENST00000626845	?	?	9
RP11-573D15.8	ENST00000628601	?	?	9
RP11-573D15.8	ENST00000630331	?	?	9
RP11-573D15.8	ENST00000625741	?	?	9
RP11-573D15.8	ENST00000628858	?	?	9
RP11-573D15.8	ENST00000627469	?	?	9
RP11-573D15.8	ENST00000627830	?	?	9
RP11-573D15.8	ENST00000627551	?	?	9

RP11-573D15.8	ENST00000354642	?	?	9
RP11-573D15.8	ENST00000628020	?	?	9
RP11-573D15.8	ENST00000630864	?	?	9
RP11-573D15.8	ENST00000625710	?	?	9
RP11-573D15.8	ENST00000628505	?	?	9
RP11-573D15.8	ENST00000629451	?	?	9
RP11-573D15.8	ENST00000625303	?	?	9
RP11-573D15.8	ENST00000629734	?	?	8
RP11-573D15.8	ENST00000625839	?	?	9
RP11-573D15.8	ENST00000628253	?	?	9
RP11-573D15.8	ENST00000628831	?	?	8
RP11-573D15.8	ENST00000628728	?	?	9
RP11-573D15.8	ENST00000631299	?	?	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-573D15.8	ENST00000629126	?	2
RP11-573D15.8	ENST00000628020	?	2
RP11-573D15.8	ENST00000629734	?	1

Locus 97 (rs41271951)

SNiPAcard

Block annotations

Block info

genomic range	chr1:150,616,888-150,737,220
block size	120,333 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.823 [-1.957 – -0.018]	gene(s) hit or close-by	CTSS, GOLPH3L, HORMAD1
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	CTSS, SEMA6C
GERP++	μ = -0.649 [-1.64 – 0.874]	potentially regulated gene(s)	CA14, CTSK
CADD score	μ = 4.745 [2.699 – 6.373]	disease gene(s)	CTSK

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CTSK	PYCNODYSOSTOSIS	OMIM	MIM:265800
CTSK	PYCNODYSOSTOSIS	DECIPHER	MIM:265800
CTSK	PYCNODYSOSTOSIS	OrphaNet	OrphaNet:763

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CTSS	missense variant	ENST00000368985	NM_004079.4	ENSP00000357981	A/V	gCg/gTg	?	?	1
CTSS	missense variant	ENST00000448301	NM_001199739.1	ENSP00000408414	A/V	gCg/gTg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CTSS	?	ENSG00000163131	muscularis mucosae	3.10×10^{-7} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	tibial nerve	2.48×10^{-12} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	transformed fibroblasts	7.49×10^{-10} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	tibial artery	2.61×10^{-13} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	thyroid	4.04×10^{-8} (p-value)	GTEx Portal V6	3
SEMA6C	?	ENSG00000143434	thyroid	2.60×10^{-8} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	skeletal muscle	6.75×10^{-9} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	transverse colon	7.97×10^{-9} (p-value)	GTEx Portal V6	3
SEMA6C	?	ENSG00000143434	cerebellum	3.85×10^{-6} (p-value)	GTEx Portal V6	3

CTSS	?	ENSG00000163131	sun exposed skin	1.83×10-12 (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	subcutaneous adipocytes	5.77×10-13 (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	pancreas	4.73×10-6 (p-value)	GTEx Portal V6	2
CTSS	?	ENSG00000163131	EBV lymphocytes	8.05×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000036221	1	ENCP00000004304	CA14
		ENCP00000004339	CTSK

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000544808 (promoter)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K4me2
		blood (K562)	DNase1, PU1, H3K9ac, H2AZ, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	DNase1, Cjun
		liver (HepG2)	DNase1
		blood (GM12878)	H3K79me2, H3K4me3, H3K27ac, H3K9ac, DNase1, H2AZ, PU1, PolII
		lung (IMR90)	DNase1
		A549	H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K27ac, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTSS	upstream gene variant	42	ENST00000480760	?	?	1
GOLPH3L	downstream gene variant	4025	ENST00000427665	?	ENSP00000410476	1
GOLPH3L	downstream gene variant	1813	ENST00000271732	NM_018178.5	ENSP00000271732	1
HORMAD1	downstream gene variant	488	ENST00000476530	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HORMAD1	ENST00000322343	NM_001199829.1	ENSP00000326489	1
HORMAD1	ENST00000442853	?	ENSP00000401940	1
HORMAD1	ENST00000361824	NM_032132.4	ENSP00000355167	1
HORMAD1	ENST00000368987	?	ENSP00000357983	1
HORMAD1	ENST00000368995	?	ENSP00000357991	1

Locus 98 (rs6695321)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,675,861-196,675,861
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.57	gene(s) hit or close-by	CFH
phastCons	0.001	eQTL gene(s)	CFHR3
GERP++	0.628	potentially regulated gene(s)	–
CADD score	2.058	disease gene(s)	CFHR3, CFH

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571
CFH	Familial drusen	OrphaNet	OrphaNet:75376
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	atrial appendage	7.18×10^{-6} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	tibial nerve	7.49×10^{-10} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	aorta	6.75×10^{-7} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	tibial artery	3.96×10^{-9} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	subcutaneous adipocytes	2.39×10^{-7} (p-value)	GTEx Portal V6	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFH	ENST00000466229	?	?	1
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	1

Locus 99 (rs16845091)

SNiPAcard

Block annotations

Block info

genomic range	chr4:74,735,524-74,837,932
block size	102,409 bp
variant count	53 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.290 [-3.136 – 1.95]	gene(s) hit or close-by	CXCL1, CXCL1P, HNRNPA1P55
phastCons	μ = 0.088 [0 – 0.99]	eQTL gene(s)	CXCL6, PF4
GERP++	μ = -0.443 [-6.02 – 3.73]	potentially regulated gene(s)	AFP, ALB, RP11-622A1.1
CADD score	μ = 3.025 [0.058 – 10.27]	disease gene(s)	ALB, AFP

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ALB	HYPERTHYROXINEMIA, FAMILIAL DYSALBUMINEMIC	OMIM	MIM:615999
ALB	ANALBUMINEMIA	OMIM	MIM:616000
AFP	ALPHA-FETOPROTEIN, HEREDITARY PERSISTENCE OF	OMIM	MIM:615970
AFP	ALPHA-FETOPROTEIN DEFICIENCY	OMIM	MIM:615969
ALB	Congenital analbuminemia	OrphaNet	OrphaNet:86816
AFP	Congenital deficiency in alpha-fetoprotein	OrphaNet	OrphaNet:168612
AFP	Hereditary persistence of alpha-fetoprotein	OrphaNet	OrphaNet:168615

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PF4	?	ENSG00000163737	transformed fibroblasts	4.04×10^{-12} (p-value)	GTEx Portal V6	30
PF4	?	ENSG00000163737	tibial artery	1.00×10^{-6} (p-value)	GTEx Portal V6	3
CXCL6	?	ENSG00000124875	ovary	2.91×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382127	1	ENCP00000041354	AFP
		ENCP00000041348	ALB
		ENCP00000041359	RP11-622A1.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001242854 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, H3K4me2, H3K4me3, DNase1
		HSMMtube	H2AZ, H3K27me3
		blood (K562)	CTCF, Rad21, H3K9ac, H2AZ, H3K4me2, H3K4me3, H3K27me3

		skin (NHDF-AD)	H3K4me3, H3K4me2
		muscle (HSMM)	H3K4me3, H2AZ
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3, H3K4me3
		blood (GM12878)	H3K27me3
		nervous (NH-A)	H3K4me2, H3K27me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1, H3K4me1
		NHLF	H3K9ac, H3K4me3
		Osteobl	H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K4me3, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3
ENSR00001242858 (enhancer)	1	monocytes (Monocytes-CD14+)	DNase1, H3K27ac
		endothelium (HUVEC)	Cjun
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
ENSR00001685780 (open chromatin region)	1	lung (IMR90)	H3K27me3
ENSR00001242860 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		HSMMtube	H3K27me3
		blood (K562)	DNase1
		blood (DND-41)	H3K27me3
ENSR00001432046 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		nervous (NH-A)	H3K27me3
		muscle (HSMM)	DNase1
ENSR00001432047 (promoter flanking region)	1	HSMMtube	H3K27me3, DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001685781 (enhancer)	1	HSMMtube	H3K27me3
ENSR00001685782 (CTCF binding site)	1	NHLF	DNase1
		nervous (NH-A)	DNase1
		blood (K562)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001432049 (promoter flanking region)	3	NHLF	DNase1
		HSMMtube	H3K27me3
		nervous (NH-A)	DNase1
		blood (K562)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001685783 (CTCF binding site)	1	NHLF	DNase1
		HSMMtube	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00001432050 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	H3K27me3
		blood (K562)	DNase1
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001432054 (promoter flanking region)	2	NHLF	DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27ac
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1, H3K36me3
		cervix (HeLa-S3)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, DNase1
ENSR00001432057 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		Osteobl	CTCF
		blood (K562)	Rad21
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		lung (IMR90)	CTCF
		A549	CTCF
ENSR00001242869 (enhancer)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H3K27me3
		Osteobl	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF
		lung (IMR90)	CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CXCL1	downstream gene variant	2957	ENST00000509101	?	?	2
CXCL1	downstream gene variant	1892	ENST00000395761	NM_001511.3	ENSP00000379110	2
CXCL1P	downstream gene variant, upstream gene variant	2721	ENST00000502804	?	?	3
HNRNPA1P55	downstream gene variant, upstream gene variant	667	ENST00000505935	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL1	ENST00000509101	?	?	1
CXCL1	ENST00000395761	NM_001511.3	ENSP00000379110	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
HNRNPA1P55	ENST00000505935	?	1

Locus 100 (rs2518136)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,335,941-186,341,830
block size	5,890 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.386 [-1.316 – 0.255]	gene(s) hit or close-by	AHSG, RP11-573D15.8
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	–
GERP++	μ = 0.464 [-3.04 – 3.81]	potentially regulated gene(s)	–
CADD score	μ = 3.943 [0.153 – 11.52]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Calcium oxalate urolithiasis	association	ClinVar	RCV000128583.1 ClinVar	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001486982 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, Yy1, DNase1
		HSMMtube	DNase1, H3K27me3, CTCF
		blood (K562)	CTCF, Rad21, H3K27me3
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	H3K4me2, CTCF, H2AZ, DNase1
		liver (HepG2)	DNase1, H3K36me3, CTCF, Rad21, PolII, H4K20me1
		blood (GM12878)	DNase1, H2AZ, Rad21, H3K4me2, CTCF
		lung (IMR90)	DNase1, H3K4me2, H3K27me3, CTCF
		nervous (NH-A)	CTCF, H3K4me2, DNase1
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2
		NHLF	CTCF, DNase1
		Osteobl	H2AZ, H3K4me2, CTCF, H3K27me3
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF, H3K4me2, DNase1
		cervix (HeLa-S3)	Max, CTCF, H3K27me3, DNase1, H3K4me2
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3, DNase1
		A549	H3K27me3, DNase1, CTCF, H3K4me3, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AHSG	downstream gene variant	163	ENST00000478441	?	?	3
AHSG	downstream gene variant	2286	ENST00000411641	NM_001622.2	ENSP00000393887	3
AHSG	downstream gene variant	2304	ENST00000273784	?	ENSP00000273784	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AHSG	ENST00000273784	?	ENSP00000273784	3
AHSG	ENST00000411641	NM_001622.2	ENSP00000393887	3
RP11-573D15.8	ENST00000625386	?	?	6
RP11-573D15.8	ENST00000630178	?	?	6
RP11-573D15.8	ENST00000628505	?	?	6

Locus 101 (rs4704846)

SNiPAcard

Block annotations

Block info

genomic range	chr5:156,507,427-156,547,031
block size	39,605 bp
variant count	49 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.820 [-4.17 – 0.597]	gene(s) hit or close-by	CTB-120L21.1, HAVCR2
phastCons	μ = 0.018 [0 – 0.449]	eQTL gene(s)	HAVCR1, HAVCR2, MED7
GERP++	μ = -0.242 [-7.18 – 2.35]	potentially regulated gene(s)	ADAM19, CTB-120L21.1, CTB-4E7.1, HAVCR2
CADD score	μ = 2.684 [0.011 – 10.38]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
HAVCR2	missense variant	ENST00000307851	NM_032782.4	ENSP00000312002	R/L	cGg/cTg	tolerated	benign	1
HAVCR2	missense variant	ENST00000524219	?	ENSP00000430328	R/L	cGg/cTg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HAVCR1	?	ENSG00000113249	visceral adipocytes	6.97×10^{-9} (p-value)	GTEx Portal V6	47
HAVCR1	?	ENSG00000113249	lung	8.25×10^{-11} (p-value)	GTEx Portal V6	47
HAVCR1	?	ENSG00000113249	tibial nerve	8.25×10^{-10} (p-value)	GTEx Portal V6	47
HAVCR1	?	ENSG00000113249	subcutaneous adipocytes	1.56×10^{-10} (p-value)	GTEx Portal V6	47
HAVCR2	ENST00000307851	ILMN_1693826	blood	5.47×10^{-9} (p-value)	MuTHER consortium	22
MED7	ENST00000524289	ILMN_2229170	b-cell	5.84×10^{-5} (p-value)	Fairfax et al.	4
MED7	ENST00000420343					
MED7	ENST00000286317					
HAVCR1	?	ENSG00000113249	coronary artery	4.35×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000681538	1	ENST00000307851, ENST00000521665, ENST00000524219	HAVCR2

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000429834	1	ENCP00000046421	HAVCR2
		ENCP00000046422	HAVCR2
ENCE00000429824	1	ENCP00000046420	CTB-120L21.1
ENCE00000430024	1	ENCP00000046430	CTB-4E7.1
		ENCP00000046453	ADAM19

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001294841 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	PU1, PolII, H2AZ, H3K27ac, H3K4me2
ENSR00001699242 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27ac, DNase1
		blood (GM12878)	H2AZ
		blood (K562)	H3K27me3
ENSR00001294842 (enhancer)	1	blood (GM12878)	H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTB-120L21.1	upstream gene variant, downstream gene variant	63	ENST00000517708	?	?	20
HAVCR2	downstream gene variant, upstream gene variant	151	ENST00000307851	NM_032782.4	ENSP00000312002	9
HAVCR2	downstream gene variant, upstream gene variant	435	ENST00000522593	?	ENSP00000430873	9
HAVCR2	downstream gene variant, upstream gene variant	1648	ENST00000521665	?	?	9
HAVCR2	downstream gene variant, upstream gene variant	179	ENST00000522902	?	?	22
HAVCR2	downstream gene variant	1606	ENST00000524219	?	ENSP00000430328	5
HAVCR2	downstream gene variant, upstream gene variant	446	ENST00000517358	?	?	24

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTB-120L21.1	ENST00000517708	?	?	15
HAVCR2	ENST00000307851	NM_032782.4	ENSP00000312002	26
HAVCR2	ENST00000522593	?	ENSP00000430873	26
HAVCR2	ENST00000524219	?	ENSP00000430328	37
HAVCR2	ENST00000521665	?	?	21

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
HAVCR2	ENST00000307851	NM_032782.4	ENSP00000312002	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
HAVCR2	ENST00000307851	NM_032782.4	ENSP00000312002	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTB-120L21.1	ENST00000517708	?	4
HAVCR2	ENST00000521665	?	5

HAVCR2	ENST00000522902	?	3

Locus 102 (rs4951168)

SNiPAcard

Block annotations

Block info

genomic range	chr1:205,014,930-205,235,781
block size	220,852 bp
variant count	55 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.887 [-3.535 – 2.059]	**gene(s) hit or close-by**	CNTN2, DSTYK, RBBP5, RP11-383G10.1, RP11-383G10.5, RP11-536L3.4, TMCC2, TMEM81
phastCons	μ = 0.040 [0 – 1]	**eQTL gene(s)**	CNTN2, DSTYK, RBBP5, RP11-383G10.5, TMEM81
GERP++	μ = -0.627 [-9.79 – 5.76]	**potentially regulated gene(s)**	CNTN2, DSTYK, RBBP5
CADD score	μ = 3.523 [0.073 – 22.1]	**disease gene(s)**	CNTN2, DSTYK

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Body weight	9.92×10^{-5}	dbGaP	pha003026	1
Waist circumference	7.71×10^{-5}	dbGaP	pha003024	4
Stroke	2.00×10^{-4}	dbGaP	pha002886	2

Disease gene annotation

gene	trait	source DB	source entry/link
CNTN2	EPILEPSY, FAMILIAL ADULT MYOCLONIC, 5	OMIM	MIM:615400
DSTYK	CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT, SUSCEPTIBILITY [...]	OMIM	MIM:610805
CNTN2	Benign adult familial myoclonic epilepsy	OrphaNet	OrphaNet:86814
DSTYK	CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT, CAKUT1	DECIPHER	MIM:610805

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
TMEM81	missense variant	ENST00000367167	NM_203376.1	ENSP00000356135	R/Q	cGg/cAg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CNTN2	?	ENSG00000184144	thyroid	2.07×10^{-8} (p-value)	GTEx Portal V6	36
TMEM81	?	ENSG00000174529	skeletal muscle	9.35×10^{-8} (p-value)	GTEx Portal V6	34
RBBP5	?	ENSG00000117222	tibial nerve	1.19×10^{-5} (p-value)	GTEx Portal V6	4
CNTN2	?	ENSG00000184144	tibial nerve	1.17×10^{-6} (p-value)	GTEx Portal V6	4

CNTN2	?	ENSG00000184144	muscularis mucosae	3.47×10-7 (p-value)	GTEx Portal V6	43
TMEM81	?	ENSG00000174529	muscularis mucosae	1.83×10-5 (p-value)	GTEx Portal V6	2
RP11-383G10.5	?	ENSG00000225063	testis	6.32×10-9 (p-value)	GTEx Portal V6	32
CNTN2	?	ENSG00000184144	sun exposed skin	1.96×10-6 (p-value)	GTEx Portal V6	8
DSTYK	ENST00000367162	ILMN_2352023	monocyte	1.04×10-30 (p-value)	Zeller et al.	5
DSTYK	ENST00000367161					
CNTN2	ENST00000331830	ILMN_1718896	b-cell	3.40×10-4 (p-value)	Fairfax et al.	4

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000052358	2	ENCP00000006264	RBBP5
		ENCP00000006267	DSTYK
		ENCP00000006260	CNTN2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000082308 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Max, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	H2AZ, Rad21, CTCF
		lung (IMR90)	CTCF, DNase1
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	DNase1, CTCF, H3K4me3
ENSR00001589901 (promoter flanking region)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Max, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	H2AZ, Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K4me3, CTCF, DNase1
ENSR00000288565	1	embryonic stem cell (H1ESC)	H3K36me3

(open chromatin region)		lung (IMR90)	H3K36me3
		blood (K562)	DNase1
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00000551432 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, CTCF, DNase1
		HSMMtube	H3K27ac, DNase1
		blood (K562)	DNase1, FOSL1, Cjun, H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K27ac
		muscle (HSMM)	H3K27ac, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3, DNase1
		lung (IMR90)	DNase1, H3K27ac, H3K36me3
		nervous (NH-A)	DNase1, H3K27ac, H3K9ac
		skin (NHEK)	H3K4me1, H3K27ac, DNase1
		NHLF	H3K27ac, H3K4me1, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Max, Cmyc, Cfos, H3K36me3, H3K27ac, Jund, Cjun, H3K9ac
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac, H3K36me3
ENSR00001527878 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001589908 (CTCF binding site)	1	NHLF	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, CTCF
		blood (K562)	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001527883 (enhancer)	1	cervix (HeLa-S3)	H3K27me3
		HSMMtube	H3K27ac, DNase1
		liver (HepG2)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1
ENSR00000288582 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1, Jund
		blood (K562)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CNTN2	downstream gene variant	270	ENST00000481872	?	?	4
CNTN2	upstream gene variant, downstream gene variant	328	ENST00000525433	?	?	7
CNTN2	downstream gene variant	1409	ENST00000530594	?	?	2
CNTN2	downstream gene variant	756	ENST00000331830	NM_005076.3	ENSP00000330633	2
CNTN2	downstream gene variant	2970	ENST00000527340	?	?	1
DSTYK	downstream gene variant, upstream gene variant	704	ENST00000367162	NM_015375.2	ENSP00000356130	4
DSTYK	downstream gene variant, upstream gene variant	704	ENST00000367161	NM_199462.2	ENSP00000356129	4
DSTYK	downstream gene variant, upstream gene variant	849	ENST00000615388	?	ENSP00000478016	3
RBBP5	downstream gene variant, upstream gene variant	108	ENST00000367164	NM_001193272.1	ENSP00000356132	7

RBBP5	downstream gene variant, upstream gene variant	197	ENST00000484379	?	?	4
RBBP5	downstream gene variant, upstream gene variant	108	ENST00000264515	NM_005057.3, NM_001193273.1	ENSP00000264515	7
RP11-383G10.1	upstream gene variant, downstream gene variant	312	ENST00000428506	?	?	6
RP11-383G10.5	downstream gene variant, upstream gene variant	3749	ENST00000452599	?	?	2
RP11-536L3.4	upstream gene variant, downstream gene variant	540	ENST00000603227	?	?	5
TMCC2	upstream gene variant	3480	ENST00000330675	NM_001297611.1	ENSP00000331842	1
TMCC2	upstream gene variant	3356	ENST00000367159	?	ENSP00000356127	1
TMEM81	downstream gene variant, upstream gene variant	1482	ENST00000367167	NM_203376.1	ENSP00000356135	5

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TMEM81	ENST00000367167	NM_203376.1	ENSP00000356135	L	ctC/ctT	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN2	ENST00000331830	NM_005076.3	ENSP00000330633	5
CNTN2	ENST00000481872	?	?	4
DSTYK	ENST00000615388	?	ENSP00000478016	3
DSTYK	ENST00000367161	NM_199462.2	ENSP00000356129	14
DSTYK	ENST00000367162	NM_015375.2	ENSP00000356130	14
RBBP5	ENST00000484379	?	?	4
RBBP5	ENST00000367164	NM_001193272.1	ENSP00000356132	8
RBBP5	ENST00000264515	NM_005057.3, NM_001193273.1	ENSP00000264515	8
RP11-383G10.5	ENST00000452599	?	?	1
TMCC2	ENST00000495538	?	?	6
TMCC2	ENST00000358024	NM_014858.3	ENSP00000350718	6
TMCC2	ENST00000367159	?	ENSP00000356127	2
TMCC2	ENST00000330675	NM_001297611.1	ENSP00000331842	2
TMCC2	ENST00000545499	NM_001242925.1	ENSP00000437943	6
TMCC2	ENST00000329800	NM_001297613.1	ENSP00000329436	1
TMCC2	ENST00000468846	?	?	1
TMCC2	ENST00000481950	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN2	ENST00000331830	NM_005076.3	ENSP00000330633	3
RBBP5	ENST00000367164	NM_001193272.1	ENSP00000356132	2
RBBP5	ENST00000264515	NM_005057.3, NM_001193273.1	ENSP00000264515	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TMCC2	ENST00000495538	?	1

Locus 103 (rs308403)

SNiPAcard

Block annotations

Block info

genomic range	chr4:123,748,616-123,757,748
block size	9,133 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.001 [-0.06 – 0.059]	**gene(s) hit or close-by**	AC021205.1, FGF2, RP11-170N16.2
phastCons	μ = 0.000 [0 – 0]	**eQTL gene(s)**	CETN4P, FGF2
GERP++	μ = 1.940 [1.7 – 2.18]	**potentially regulated gene(s)**	–
CADD score	μ = 8.498 [5.337 – 11.66]	**disease gene(s)**	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR109896	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CETN4P	?	ENSG00000224786	skeletal muscle	8.37×10^{-11} (p-value)	GTEx Portal V6	2
FGF2	?	ENSG00000138685	testis	1.41×10^{-6} (p-value)	GTEx Portal V6	2
FGF2	?	ENSG00000138685	tibial artery	2.49×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001248693 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K27me3, CTCF, PolII, Rad21, Jund, TAF7, Sin3Ak20, H3K27ac, Yy1, HDAC2, Tcf12, TAF1, H3K4me2, SP1, H4K5ac, Srf, USF1, H3K9ac, H3K4me3
		HSMMtube	H3K9ac, H3K4me2
		blood (K562)	Max, H3K9ac, USF1, H3K79me2, H2AZ, H3K4me2, DNase1, H3K4me3, H3K27ac, Egr1, PolII
		skin (NHDF-AD)	H3K36me3, CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3
		liver (HepG2)	H3K4me3, H3K4me2, H3K27me3, DNase1
		blood (GM12878)	EBF1, PolII, Srf, MEF2A, H2AZ, DNase1, H3K4me3, NFKB, H3K4me2
		lung (IMR90)	CTCF, H3K56ac, H3K79me2, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K4me2, H3K4me3, H3K27me3
		NHLF	H3K4me3, DNase1, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, Cjun, H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, PolII, H3K4me3, H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3

		endothelium (HUVEC)	H3K36me3, DNase1, PolII, H3K4me2, H3K4me3
		A549	H3K4me3, H3K4me2, H3K9ac, H3K27ac, H3K36me3
ENSR00001434812 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (K562)	H3K36me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K36me3
		cervix (HeLa-S3)	Nrsf
		monocytes (Monocytes-CD14+)	H3K27me3
		lung (IMR90)	H3K79me2, H3K36me3
		A549	H3K4me2, H3K36me3, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC021205.1	downstream gene variant	371	ENST00000517260	?	?	1
RP11-170N16.2	upstream gene variant	3750	ENST00000468797	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FGF2	ENST00000608478	?	ENSP00000477134	2
FGF2	ENST00000614010	NM_002006.4	ENSP00000478620	2
FGF2	ENST00000264498	?	ENSP00000264498	2

Locus 104 (rs17114671)

SNiPAcard

Block annotations

Block info

genomic range	chr14:21,143,659-21,157,936
block size	14,278 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.527 [-1.866 – 1.605]	gene(s) hit or close-by	ANG, RNASE4, RP11-903H12.3, RP11-903H12.5, Y_RNA
phastCons	μ = 0.085 [0 – 0.813]	eQTL gene(s)	ANG, RNASE4, RP11-903H12.3, RP11-903H12.5
GERP++	μ = -2.344 [-7.85 – 2.33]	potentially regulated gene(s)	ANG, ANG, RNASE4
CADD score	μ = 3.885 [0.054 – 13.28]	disease gene(s)	ANG

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ANG	AMYOTROPHIC LATERAL SCLEROSIS 9	OMIM	MIM:611895
ANG	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-903H12.3	?	ENSG00000258451	lung	7.30×10^{-6} (p-value)	GTEx Portal V6	1
RP11-903H12.5	ENST00000553909	ILMN_2408572	monocyte	3.74×10^{-5} (p-value)	Fairfax et al.	1
ANG	ENST00000554073					
RNASE4	ENST00000555597					
RNASE4	ENST00000397995					
RNASE4	ENST00000555835					
ANG	ENST00000397990	ILMN_1760727	monocyte	4.57×10^{-9} (p-value)	Fairfax et al.	1
ANG	ENST00000336811					
ANG	ENST00000397990	ILMN_2294976	monocyte	5.32×10^{-8} (p-value)	Fairfax et al.	1
RP11-903H12.5	ENST00000553909					
ANG	ENST00000336811					
ANG	ENST00000397990	ILMN_2294978	monocyte	2.65×10^{-8} (p-value)	Fairfax et al.	1
RP11-903H12.5	ENST00000553909					
ANG	ENST00000336811					
RP11-903H12.5	ENST00000553909	ILMN_1776602	monocyte	1.03×10^{-6} (p-value)	Fairfax et al.	1
RNASE4	ENST00000397995					
RNASE4	ENST00000555597					
RNASE4	ENST00000555835					
ANG	ENST00000397990	ILMN_1696974	monocyte	1.31×10^{-8} (p-value)	Fairfax et al.	1
RP11-903H12.5	ENST00000553909					
ANG	ENST00000336811					
RP11-903H12.5	ENST00000553909	ILMN_2408572	monocyte	3.45×10^{-17} (p-value)	Zeller et al.	2
ANG	ENST00000554073					
RNASE4	ENST00000397995					

RNASE4	ENST00000555597						
RNASE4	ENST00000555835						
ANG	ENST00000397990	ILMN_1760727	monocyte	5.55×10^{-19} (p-value)	Zeller et al.	2	
ANG	ENST00000336811						
ANG	ENST00000397990	ILMN_2294976	monocyte	1.01×10^{-17} (p-value)	Zeller et al.	2	
RP11-903H12.5	ENST00000553909						
ANG	ENST00000336811						
ANG	ENST00000397990	ILMN_2294978	monocyte	1.33×10^{-15} (p-value)	Zeller et al.	2	
RP11-903H12.5	ENST00000553909						
ANG	ENST00000336811						
RP11-903H12.5	ENST00000553909	ILMN_1776602	monocyte	7.17×10^{-20} (p-value)	Zeller et al.	2	
RNASE4	ENST00000555597						
RNASE4	ENST00000397995						
RNASE4	ENST00000555835						
ANG	ENST00000397990	ILMN_1696974	monocyte	2.28×10^{-16} (p-value)	Zeller et al.	2	
RP11-903H12.5	ENST00000553909						
ANG	ENST00000336811						

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000019332		RNASE4
		ANG
ENCP00000019333		RNASE4
		ANG

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001455837 (promoter)	4	embryonic stem cell (H1ESC)	POU5F1, CTCF, PolII, Rad21, H3K4me3, H3K9ac, USF1, SP1, H3K4me2, TAF1, Yy1, H3K27ac, Sin3Ak20, TAF7, DNase1
		HSMMtube	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		blood (K562)	DNase1, Egr1, Max, USF1, H2AZ, PolII, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H3K9ac, DNase1, H3K4me2, H3K27ac, H2AZ, H3K4me3
		liver (HepG2)	H3K4me2, HNF4A, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K36me3, H3K27me3, DNase1, p300, H2AZ, Gabp, FOXA2, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, ELF1, PolIII, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K9ac, DNase1, H3K27ac, H3K4me2, H3K4me3
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K4me3, H3K4me2, H3K9ac, H3K27ac, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me2, H3K9ac, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANG	upstream gene variant	96	ENST00000397990	NM_001097577.2	ENSP00000381077	7
ANG	upstream gene variant	169	ENST00000554073	?	?	1
RNASE4	upstream gene variant	138	ENST00000397995	?	ENSP00000381081	1

RNASE4	upstream gene variant	105	ENST00000555597	NM_194431.2	ENSP00000451624	7
RP11-903H12.3	downstream gene variant	3703	ENST00000554286	?	?	2
RP11-903H12.5	upstream gene variant	142	ENST00000553909	?	ENSP00000477037	1
Y_RNA	upstream gene variant, downstream gene variant	3878	ENST00000362591	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANG	ENST00000554073	?	?	7
ANG	ENST00000336811	NM_001145.4	ENSP00000336762	7
ANG	ENST00000397990	NM_001097577.2	ENSP00000381077	1
RNASE4	ENST00000555835	NM_001282193.1, NM_001282192.1, NM_002937.4	ENSP00000452245	7
RNASE4	ENST00000397995	?	ENSP00000381081	7
RNASE4	ENST00000555597	NM_194431.2	ENSP00000451624	1
RP11-903H12.5	ENST00000553909	?	ENSP00000477037	7

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANG	ENST00000336811	NM_001145.4	ENSP00000336762	1
RNASE4	ENST00000555835	NM_001282193.1, NM_001282192.1, NM_002937.4	ENSP00000452245	1

Locus 105 (rs3094005)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,016,550-31,566,168
block size	549,619 bp
variant count	118 variants

Basic features

Conservation/deleteriousness		Linked genes
phyloP	μ = -0.200 [-3.795 – 1.156]	**gene(s) hit or close-by**: AL662801.1, AL671883.1, ATP6V1G2, ATP6V1G2-DDX39B, DDX39B, DDX39B-AS1, DHFRP2, FGFR3P1, HCG22, HCG27, HCP5, HLA-B, HLA-C, HLA-S, LINC01149, LST1, LTA, LTB, MICA, MICB, MIR6891, NCR3, NFKBIL1, PPIAP9, RNU6-283P, SNORD117, SNORD84, TNF, XXbac-BPG16N22.5, XXbac-BPG181B23.6, XXbac-BPG181B23.7, XXbac-BPG248L24.12, XXbac-BPG299F13.14, XXbac-BPG299F13.15, XXbac-BPG299F13.16, Y_RNA, ZDHHC20P2
phastCons	μ = 0.054 [0 – 1]	**eQTL gene(s)**: BTN3A2, C4A, C4B, C6orf10, CCHCR1, CSF3R, CYP21A1P, CYP21A2, DDX39B, FLOT1, HCG22, HCP5, HIST1H2BD, HLA-B, HLA-C, HLA-DPB1, HLA-DQA1, HLA-DQB1, HLA-G, IER3, LINC00243, LST1, LY6G6C, MICA, MICB, NOTCH4, OR5V1, PPP1R18, PSORS1C1, PSORS1C2, RNF5, SKIV2L, SNORD84, STK19, STK19B, TMEM154, TNXA, VARS2, WASF5P, XXbac-BPG248L24.12, XXbac-BPG248L24.13, XXbac-BPG252P9.10
GERP++	μ = -0.098 [-3.15 – 2.1]	**potentially regulated gene(s)**: ABHD16A (×7), BAG6 (×7), C2 (×10), C6orf47 (×8), C6orf48 (×8), CCHCR1 (×8), CFB (×8), CLIC1 (×8), DDR1 (×8), DDX39B (×8), EHMT2 (×8), GPANK1 (×8), GTF2H4 (×8), HLA-B (×8), HSPA1L (×8), LSM2 (×8), LTA (×8), LTB (×8), LY6G5C (×8), LY6G6D (×8), LY6G6E (×8), MICB (×8), MSH5 (×8), NCR3 (×8), NEU1 (×8), POLR2LP (×8), POU5F1 (×8), PRRC2A (×8), PSORS1C2 (×8), SLC44A4 (×8), STK19 (×8), TCF19 (×8), VARS (×8), VARS2 (×8), WASF5P (×8), XXbac-BPG248L24.10 (×8), ZBTB12 (×4), ZDHHC20P2 (×5)
CADD score	μ = 3.433 [0.004 – 16.26]	**disease gene(s)**: C4B, C4A, CYP21A2, VARS2, CSF3R, LTA, NCR3, C2, NFKBIL1, SKIV2L, TNF, CFB, NEU1, HLA-B, HLA-DQB1, HLA-DPB1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Rheumatoid arthritis (ACPA-negative)	$<9.00\times10^{-9}$	GWAS Catalog	24532677	1
Hematology traits	$<5.00\times10^{-7}$	GWAS Catalog	23263863	1
Myasthenia gravis	$<2.00\times10^{-90}$	GWAS Catalog	23055271	1
Metabolic syndrome	$<2.00\times10^{-8}$	GWAS Catalog	22399527	1
Stevens-Johnson syndrome and toxic epidermal necrolysis (SJS-TEN)	$<2.00\times10^{-8}$	GWAS Catalog	21912425	1
Idiopathic membranous nephropathy	$<6.00\times10^{-72}$	GWAS Catalog	21323541	2
Neonatal lupus	$<5.00\times10^{-10}$	GWAS Catalog	20662065	1
Psoriasis	$<1.00\times10^{-9}$	GWAS Catalog	18364390	1
laurylcarnitine	7.88×10^{-5}	Metabolomics GWAS Server	24816252	1
Lupus erythematosus, systemic	1.95×10^{-6}	dbGaP	pha002867	2
Diabetes mellitus, type 1	5.29×10^{-68}	dbGaP	pha002862	1
Breast neoplasms	1.62×10^{-4}	dbGaP	pha002853	1
Lupus erythematosus, systemic	4.00×10^{-15}	dbGaP	pha002848	2

Disease gene annotation

gene	trait	source DB	source entry/link
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
CYP21A2	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
CSF3R	NEUTROPHILIA, HEREDITARY	OMIM	MIM:162830
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
LTA	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988
LTA	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
NCR3	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...]	OMIM	MIM:608579
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000

C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602	OMIM
NFKBIL1	RHEUMATOID ARTHRITIS	OMIM	MIM:180300	OMIM
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
NEU1	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	OrphaNet	OrphaNet:315306	orphanet
CYP21A2	Nonclassic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	OrphaNet	OrphaNet:95698	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	OrphaNet	OrphaNet:315311	orphanet
HLA-DQB1	Bullous pemphigoid	OrphaNet	OrphaNet:703	orphanet
HLA-DQB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
HLA-DQB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465	orphanet
CSF3R	Chronic neutrophilic leukemia	OrphaNet	OrphaNet:86829	orphanet
	Hereditary neutrophilia	OrphaNet	OrphaNet:279943	

CSF3R	Atypical chronic myeloid leukemia	OrphaNet	OrphaNet:98824	
HLA-DPB1	Chronic berylliosis	OrphaNet	OrphaNet:133	
HLA-DPB1	Granulomatosis with polyangiitis	OrphaNet	OrphaNet:900	
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064	
NEU1	Juvenile sialidosis type 2	OrphaNet	OrphaNet:93399	
NEU1	Sialidosis type 1	OrphaNet	OrphaNet:812	
NEU1	Congenital sialidosis type 2	OrphaNet	OrphaNet:93400	
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
NEU1	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG248L24.13	?	ENSG00000256166	pancreas	1.60×10^{-8} (p-value)	GTEx Portal V6	107
C4A	?	ENSG00000244731	pancreas	3.09×10^{-9} (p-value)	GTEx Portal V6	115
WASF5P	?	ENSG00000231402	pancreas	8.19×10^{-12} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	pancreas	1.80×10^{-8} (p-value)	GTEx Portal V6	110
HLA-B	?	ENSG00000234745	pancreas	3.41×10^{-7} (p-value)	GTEx Portal V6	95
RNF5	?	ENSG00000204308	pancreas	1.58×10^{-6} (p-value)	GTEx Portal V6	31
CYP21A1P	?	ENSG00000204338	pancreas	1.35×10^{-7} (p-value)	GTEx Portal V6	18
C4A	?	ENSG00000244731	muscularis mucosae	4.51×10^{-15} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	muscularis mucosae	2.65×10^{-11} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	muscularis mucosae	2.07×10^{-6} (p-value)	GTEx Portal V6	6
HLA-B	?	ENSG00000234745	muscularis mucosae	2.42×10^{-7} (p-value)	GTEx Portal V6	96
CYP21A1P	?	ENSG00000204338	muscularis mucosae	2.75×10^{-10} (p-value)	GTEx Portal V6	116
MICB	?	ENSG00000204516	muscularis mucosae	2.13×10^{-8} (p-value)	GTEx Portal V6	41
PSORS1C1	?	ENSG00000204540	muscularis mucosae	2.08×10^{-6} (p-value)	GTEx Portal V6	2
C4A	?	ENSG00000244731	lung	1.56×10^{-15} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	lung	5.80×10^{-11} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	lung	7.03×10^{-9} (p-value)	GTEx Portal V6	99
CYP21A1P	?	ENSG00000204338	lung	1.64×10^{-9} (p-value)	GTEx Portal V6	116
MICB	?	ENSG00000204516	lung	1.60×10^{-6} (p-value)	GTEx Portal V6	5
MICA	?	ENSG00000204520	lung	6.23×10^{-6} (p-value)	GTEx Portal V6	1

C4A	?	ENSG00000244731	atrial appendage	5.15×10-15 (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	atrial appendage	5.33×10-11 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	atrial appendage	1.42×10-6 (p-value)	GTEx Portal V6	6
C4B	?	ENSG00000224389	transformed fibroblasts	2.51×10-13 (p-value)	GTEx Portal V6	116
C4A	?	ENSG00000244731	transformed fibroblasts	3.99×10-18 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	transformed fibroblasts	1.59×10-13 (p-value)	GTEx Portal V6	116
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	5.25×10-6 (p-value)	GTEx Portal V6	47
MICB	?	ENSG00000204516	transformed fibroblasts	3.71×10-6 (p-value)	GTEx Portal V6	17
C4A	?	ENSG00000244731	blood	1.08×10-10 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	blood	3.47×10-12 (p-value)	GTEx Portal V6	109
C4B	?	ENSG00000224389	blood	7.08×10-16 (p-value)	GTEx Portal V6	116
CYP21A2	?	ENSG00000231852	blood	5.84×10-7 (p-value)	GTEx Portal V6	53
CCHCR1	?	ENSG00000204536	blood	4.72×10-7 (p-value)	GTEx Portal V6	97
CYP21A1P	?	ENSG00000204338	blood	1.78×10-12 (p-value)	GTEx Portal V6	116
LINC00243	?	ENSG00000214894	blood	1.78×10-11 (p-value)	GTEx Portal V6	102
XXbac-BPG252P9.10	?	ENSG00000272273	blood	5.09×10-6 (p-value)	GTEx Portal V6	2
C4A	?	ENSG00000244731	breast	2.53×10-13 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	breast	5.94×10-12 (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	breast	4.14×10-12 (p-value)	GTEx Portal V6	116
MICB	?	ENSG00000204516	breast	7.43×10-7 (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	tibial artery	1.88×10-16 (p-value)	GTEx Portal V6	116
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	6.92×10-7 (p-value)	GTEx Portal V6	78
C4A	?	ENSG00000244731	tibial artery	4.90×10-8 (p-value)	GTEx Portal V6	39
CYP21A1P	?	ENSG00000204338	tibial artery	1.54×10-7 (p-value)	GTEx Portal V6	15
MICB	?	ENSG00000204516	tibial artery	4.39×10-11 (p-value)	GTEx Portal V6	34
C4A	?	ENSG00000244731	thyroid	7.45×10-12 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	thyroid	1.90×10-10 (p-value)	GTEx Portal V6	115
FLOT1	?	ENSG00000137312	thyroid	4.67×10-8 (p-value)	GTEx Portal V6	98
CCHCR1	?	ENSG00000204536	thyroid	1.10×10-11 (p-value)	GTEx Portal V6	110
PPP1R18	?	ENSG00000146112	thyroid	6.71×10-7 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	thyroid	1.77×10-13 (p-value)	GTEx Portal V6	116
PSORS1C1	?	ENSG00000204540	thyroid	5.44×10-12 (p-value)	GTEx Portal V6	116
PSORS1C2	?	ENSG00000204538	thyroid	4.58×10-8 (p-value)	GTEx Portal V6	31
MICA	?	ENSG00000204520	thyroid	1.10×10-5 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	skeletal muscle	2.99×10-13 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	skeletal muscle	8.80×10-21 (p-value)	GTEx Portal V6	116
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	7.46×10-8 (p-value)	GTEx Portal V6	99
CYP21A1P	?	ENSG00000204338	skeletal muscle	7.49×10-7 (p-value)	GTEx Portal V6	51

RNF5	?	ENSG00000204308	skeletal muscle	4.32×10^{-6} (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	skeletal muscle	1.88×10^{-6} (p-value)	GTEx Portal V6	3
PSORS1C1	?	ENSG00000204540	skeletal muscle	3.52×10^{-6} (p-value)	GTEx Portal V6	5
C4A	?	ENSG00000244731	EBV lymphocytes	2.79×10^{-8} (p-value)	GTEx Portal V6	111
HLA-C	?	ENSG00000204525	EBV lymphocytes	1.93×10^{-6} (p-value)	GTEx Portal V6	51
C4A	?	ENSG00000244731	transverse colon	9.56×10^{-11} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	transverse colon	1.03×10^{-9} (p-value)	GTEx Portal V6	111
HLA-C	?	ENSG00000204525	transverse colon	9.57×10^{-10} (p-value)	GTEx Portal V6	116
VARS2	?	ENSG00000137411	transverse colon	2.70×10^{-6} (p-value)	GTEx Portal V6	3
PSORS1C1	?	ENSG00000204540	transverse colon	2.43×10^{-6} (p-value)	GTEx Portal V6	50
C4A	?	ENSG00000244731	liver	3.18×10^{-12} (p-value)	GTEx Portal V6	116
STK19B	?	ENSG00000250535	liver	1.17×10^{-7} (p-value)	GTEx Portal V6	8
CCHCR1	?	ENSG00000204536	liver	1.52×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	sun exposed skin	3.48×10^{-12} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	sun exposed skin	4.28×10^{-13} (p-value)	GTEx Portal V6	116
C4B	?	ENSG00000224389	sun exposed skin	5.52×10^{-10} (p-value)	GTEx Portal V6	116
CYP21A2	?	ENSG00000231852	sun exposed skin	2.48×10^{-9} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	sun exposed skin	2.63×10^{-6} (p-value)	GTEx Portal V6	33
CYP21A1P	?	ENSG00000204338	sun exposed skin	1.00×10^{-8} (p-value)	GTEx Portal V6	115
PSORS1C1	?	ENSG00000204540	sun exposed skin	6.04×10^{-7} (p-value)	GTEx Portal V6	79
VARS2	?	ENSG00000137411	sun exposed skin	7.32×10^{-6} (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	sun exposed skin	7.73×10^{-13} (p-value)	GTEx Portal V6	113
NOTCH4	?	ENSG00000204301	sun exposed skin	1.55×10^{-6} (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	unexposed skin	3.00×10^{-11} (p-value)	GTEx Portal V6	115
HLA-C	?	ENSG00000204525	unexposed skin	6.98×10^{-8} (p-value)	GTEx Portal V6	114
CYP21A1P	?	ENSG00000204338	unexposed skin	1.71×10^{-8} (p-value)	GTEx Portal V6	112
MICB	?	ENSG00000204516	unexposed skin	1.42×10^{-8} (p-value)	GTEx Portal V6	65
CYP21A2	?	ENSG00000231852	unexposed skin	3.27×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	aorta	9.09×10^{-12} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	aorta	2.76×10^{-10} (p-value)	GTEx Portal V6	116
HLA-B	?	ENSG00000234745	aorta	3.56×10^{-7} (p-value)	GTEx Portal V6	99
PPP1R18	?	ENSG00000146112	aorta	2.23×10^{-7} (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	aorta	6.17×10^{-11} (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	aorta	2.66×10^{-9} (p-value)	GTEx Portal V6	116
C4A	?	ENSG00000244731	left ventricle	2.24×10^{-8} (p-value)	GTEx Portal V6	71
HLA-C	?	ENSG00000204525	left ventricle	5.04×10^{-9} (p-value)	GTEx Portal V6	99
CYP21A1P	?	ENSG00000204338	left ventricle	1.33×10^{-7} (p-value)	GTEx Portal V6	17

PSORS1C1	?	ENSG00000204540	left ventricle	1.37×10^{-8} (p-value)	GTEx Portal V6	105
NOTCH4	?	ENSG00000204301	left ventricle	1.30×10^{-7} (p-value)	GTEx Portal V6	32
C4A	?	ENSG00000244731	subcutaneous adipocytes	3.88×10^{-10} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	1.39×10^{-13} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	subcutaneous adipocytes	1.56×10^{-12} (p-value)	GTEx Portal V6	115
CYP21A1P	?	ENSG00000204338	subcutaneous adipocytes	8.99×10^{-10} (p-value)	GTEx Portal V6	108
VARS2	?	ENSG00000137411	subcutaneous adipocytes	1.76×10^{-6} (p-value)	GTEx Portal V6	3
CYP21A2	?	ENSG00000231852	subcutaneous adipocytes	5.06×10^{-9} (p-value)	GTEx Portal V6	114
PSORS1C1	?	ENSG00000204540	subcutaneous adipocytes	3.60×10^{-7} (p-value)	GTEx Portal V6	57
NOTCH4	?	ENSG00000204301	subcutaneous adipocytes	1.38×10^{-6} (p-value)	GTEx Portal V6	77
MICB	?	ENSG00000204516	subcutaneous adipocytes	4.89×10^{-8} (p-value)	GTEx Portal V6	30
C4A	?	ENSG00000244731	stomach	6.74×10^{-14} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	stomach	9.04×10^{-9} (p-value)	GTEx Portal V6	99
PSORS1C1	?	ENSG00000204540	stomach	1.26×10^{-6} (p-value)	GTEx Portal V6	51
CCHCR1	?	ENSG00000204536	visceral adipocytes	1.20×10^{-8} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	visceral adipocytes	7.22×10^{-11} (p-value)	GTEx Portal V6	116
C4A	?	ENSG00000244731	visceral adipocytes	1.27×10^{-8} (p-value)	GTEx Portal V6	111
NOTCH4	?	ENSG00000204301	visceral adipocytes	9.33×10^{-7} (p-value)	GTEx Portal V6	23
CYP21A1P	?	ENSG00000204338	visceral adipocytes	4.77×10^{-7} (p-value)	GTEx Portal V6	4
C4A	?	ENSG00000244731	tibial nerve	2.20×10^{-16} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	tibial nerve	1.52×10^{-12} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	tibial nerve	3.84×10^{-10} (p-value)	GTEx Portal V6	109
CYP21A1P	?	ENSG00000204338	tibial nerve	3.79×10^{-13} (p-value)	GTEx Portal V6	116
PSORS1C1	?	ENSG00000204540	tibial nerve	1.86×10^{-7} (p-value)	GTEx Portal V6	69
MICB	?	ENSG00000204516	tibial nerve	8.80×10^{-8} (p-value)	GTEx Portal V6	21
C4A	?	ENSG00000244731	esophagus mucosa	1.33×10^{-14} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	esophagus mucosa	3.24×10^{-13} (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	esophagus mucosa	1.42×10^{-10} (p-value)	GTEx Portal V6	116
MICB	?	ENSG00000204516	esophagus mucosa	2.18×10^{-10} (p-value)	GTEx Portal V6	116
LY6G6C	?	ENSG00000204421	esophagus mucosa	9.70×10^{-10} (p-value)	GTEx Portal V6	112
PSORS1C1	?	ENSG00000204540	esophagus mucosa	6.36×10^{-7} (p-value)	GTEx Portal V6	7
HLA-C	?	ENSG00000204525	gastroesophageal junction	5.10×10^{-7} (p-value)	GTEx Portal V6	18
C4A	?	ENSG00000244731	gastroesophageal junction	2.08×10^{-7} (p-value)	GTEx Portal V6	32
STK19B	?	ENSG00000250535	adrenal gland	2.85×10^{-11} (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	adrenal gland	6.58×10^{-8} (p-value)	GTEx Portal V6	110
PSORS1C1	?	ENSG00000204540	adrenal gland	3.76×10^{-7} (p-value)	GTEx Portal V6	82
STK19	?	ENSG00000204344	adrenal gland	7.61×10^{-7} (p-value)	GTEx Portal V6	32

HLA-C	?	ENSG00000204525	adrenal gland	1.04×10-6 (p-value)	GTEx Portal V6	42
TNXA	?	ENSG00000248290	adrenal gland	2.42×10-7 (p-value)	GTEx Portal V6	35
C4A	?	ENSG00000244731	adrenal gland	1.49×10-7 (p-value)	GTEx Portal V6	20
C4A	?	ENSG00000244731	cerebellar hemisphere	8.98×10-10 (p-value)	GTEx Portal V6	49
MICB	?	ENSG00000204516	cerebellar hemisphere	8.82×10-8 (p-value)	GTEx Portal V6	71
C4A	?	ENSG00000244731	testis	1.75×10-11 (p-value)	GTEx Portal V6	110
HLA-C	?	ENSG00000204525	testis	6.97×10-7 (p-value)	GTEx Portal V6	48
CYP21A1P	?	ENSG00000204338	testis	2.49×10-9 (p-value)	GTEx Portal V6	100
C6orf10	?	ENSG00000204296	testis	2.15×10-6 (p-value)	GTEx Portal V6	16
VARS2	ENST00000321897	ILMN_1737585	skin	6.13×10-8 (p-value)	MuTHER consortium	22
VARS2	ENST00000625423		adipocyte	3.43×10-6 (p-value)	MuTHER consortium	22
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
HCG22	ENST00000570223	ILMN_1667229	blood	1.28×10-7 (p-value)	MuTHER consortium	22
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
HCP5	ENST00000414046	ILMN_1803945	skin	2.86×10-9 (p-value)	MuTHER consortium	22
HCP5	ENST00000541196		adipocyte	1.34×10-7 (p-value)	MuTHER consortium	22
MICB	ENST00000538442	ILMN_1708006	skin	7.98×10-14 (p-value)	MuTHER consortium	22
MICB	ENST00000399150					
MICB	ENST00000252229					
NOTCH4	ENST00000474612	ILMN_1711157	adipocyte	9.08×10-8 (p-value)	MuTHER consortium	21
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
LST1	ENST00000339530	ILMN_1718936	blood	3.68×10-5 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
SKIV2L	ENST00000484835	ILMN_1666512	skin	2.93×10-5 (p-value)	MuTHER consortium	1
SKIV2L	ENST00000375394					
SKIV2L	ENST00000474839					
SKIV2L	ENST00000470453					
SKIV2L	ENST00000465703					
SKIV2L	ENST00000491994					
SKIV2L	ENST00000471818					
SKIV2L	ENST00000483553					

SKIV2L	ENST00000485349					
?	?	ILMN_1679389	b-cell	1.55×10-8 (p-value)	Fairfax et al.	9
VARS2	ENST00000321897	ILMN_1737585	monocyte	7.69×10-8 (p-value)	Fairfax et al.	9
VARS2	ENST00000625423		b-cell	8.53×10-6 (p-value)	Fairfax et al.	9
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
OR5V1	ENST00000543825	ILMN_1683352	b-cell	8.01×10-7 (p-value)	Fairfax et al.	9
OR5V1	ENST00000377154					
IER3	ENST00000376377	ILMN_1682717	monocyte	1.79×10-5 (p-value)	Fairfax et al.	8
IER3	ENST00000259874					
DDX39B	ENST00000481456	ILMN_3245672	b-cell	6.77×10-4 (p-value)	Fairfax et al.	3
SNORD84	ENST00000584275					
HLA-C	ENST00000470363	216526_x_at	blood	9.00×10-9 (p-value)	Dixon et al.	1
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000466892					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	?	ENSG00000204525	cerebellum	2.07×10-6 (p-value)	GTEx Portal V6	2
CYP21A1P	?	ENSG00000204338	cerebellum	5.38×10-7 (p-value)	GTEx Portal V6	49
MICB	?	ENSG00000204516	cerebellum	4.33×10-11 (p-value)	GTEx Portal V6	110
C4A	?	ENSG00000244731	cerebellum	2.67×10-7 (p-value)	GTEx Portal V6	3
C4A	?	ENSG00000244731	frontal cortex	4.09×10-9 (p-value)	GTEx Portal V6	46
CCHCR1	?	ENSG00000204536	sigmoid colon	1.95×10-6 (p-value)	GTEx Portal V6	4
C4A	?	ENSG00000244731	sigmoid colon	3.19×10-7 (p-value)	GTEx Portal V6	9
?	?	ILMN_1883997	blood	1.67×10-5 (p-value)	Westra et al.	1
HCP5	ENST00000414046	ILMN_1803945	blood	3.07×10-6 (p-value)	Westra et al.	2
HCP5	ENST00000541196					
?	?	ILMN_1893395	monocyte	6.84×10-28 (p-value)	Zeller et al.	2
VARS2	ENST00000321897	ILMN_1737585	monocyte	1.60×10-12 (p-value)	Zeller et al.	1
VARS2	ENST00000625423					
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
?	?	ILMN_1666078	monocyte	4.19×10-19 (p-value)	Zeller et al.	2
HLA-C	ENST00000470363	ILMN_2150787	monocyte	9.48×10-30 (p-value)	Zeller et al.	2
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					

HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						
?	?	ILMN_1671054	monocyte		1.03×10^{-30} (p-value)	Zeller et al.	2
HLA-G	ENST00000376828	ILMN_1656670	monocyte		2.47×10^{-13} (p-value)	Zeller et al.	1
HLA-G	ENST00000360323						
HLA-G	ENST00000478519						
HLA-G	ENST00000478355						
HLA-G	ENST00000428701						
HLA-G	ENST00000376815						
HLA-G	ENST00000376818						
BTN3A2	ENST00000396934	ILMN_1676528	monocyte		2.50×10^{-21} (p-value)	Zeller et al.	2
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
?	?	ILMN_1883997	monocyte		8.89×10^{-89} (p-value)	Zeller et al.	2
CYP21A1P	?	ENSG00000204338	caudate basal ganglia		4.92×10^{-7} (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
HLA-DQB1	ENST00000399082	209480_at	chr6	blood	4.30×10^{-11} (p-value)	Dixon et al.	3
HLA-DQB1	ENST00000399084						
HLA-DQB1	ENST00000434651						
HLA-DQB1	ENST00000460185						
HLA-DQB1	ENST00000399079						
HLA-DQB1	ENST00000443574						
HLA-DQB1	ENST00000374943						
HLA-DQB1	ENST00000487676						
?	?	221491_x_at	chr6	blood	1.20×10^{-14} (p-value)	Dixon et al.	3
HLA-DQA1	ENST00000374949	213831_at	chr6	blood	2.40×10^{-9} (p-value)	Dixon et al.	3
HLA-DQA1	ENST00000496318						
HLA-DQA1	ENST00000460633						
HLA-DQA1	ENST00000482745						
HLA-DQA1	ENST00000343139						
CSF3R	ENST00000464465	ILMN_2371280	chr1	blood	1.51×10^{-8} (p-value)	Westra et al.	1
CSF3R	ENST00000373104						
CSF3R	ENST00000487540						
CSF3R	ENST00000373106						
CSF3R	ENST00000331941						
CSF3R	ENST00000484762						
CSF3R	ENST00000480825						
CSF3R	ENST00000361632						
CSF3R	ENST00000373103						
HIST1H2BD	ENST00000289316	ILMN_1651496	chr6	blood	5.26×10^{-10} (p-value)	Westra et al.	2
TMEM154	ENST00000304385	ILMN_2088124	chr4	blood	7.64×10^{-24} (p-value)	Westra et al.	2
BTN3A2	ENST00000396934	ILMN_1676528	chr6	blood	1.39×10^{-10} (p-value)	Westra et al.	1
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
?	?	ILMN_1752592	chr6	monocyte	3.44×10^{-55} (p-value)	Zeller et al.	2
?	?	ILMN_1715169	chr6	monocyte	1.81×10^{-19} (p-value)	Zeller et al.	2
HLA-DPB1	ENST00000418931	ILMN_1749070	chr6	monocyte	6.72×10^{-18} (p-value)	Zeller et al.	2

HLA-DPB1	ENST00000416804						
?	?	ILMN_1661266	chr6	monocyte	2.70×10-25 (p-value)	Zeller et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444609	2	ENCP00000047951	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
		ENCP00000048082	LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D LY6G6D LY6G6E LY6G6D LY6G6D LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D
		ENCP00000047987	POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1
ENCE00000444610	3	ENCP00000047951	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
		ENCP00000048071	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000047980	PSORS1C2 PSORS1C2 POLR2LP PSORS1C2 POLR2LP PSORS1C2 PSORS1C2 PSORS1C2 POLR2LP POLR2LP POLR2LP POLR2LP
		ENCP00000047987	POU5F1 POU5F1 POU5F1 POU5F1 POU5F1

			POU5F1
			POU5F1
		ENCP00000047988	POU5F1
			POU5F1
			POU5F1
			POU5F1
			POU5F1
			POU5F1
			POU5F1
		ENCP00000047969	VARS2
			VARS2
			VARS2
			VARS2
			VARS2
			VARS2
ENCE00000445031	2	ENCP00000048032	LTA
			LTA
			LTA
			LTA
			LTA
			LTA
			LTA
		ENCP00000048026	DDX39B
			DDX39B
			DDX39B
			DDX39B
			DDX39B
			DDX39B
			DDX39B
			DDX39B
ENCE00000444689	1	ENCP00000048034	LTB
			LTB
			LTB
			LTB
			LTB
			LTB
			LTB
			LTB
		ENCP00000048002	XXbac-BPG248L24.10
			XXbac-BPG248L24.10
			XXbac-BPG248L24.10
			XXbac-BPG248L24.10
			XXbac-BPG248L24.10
			XXbac-BPG248L24.10
			XXbac-BPG248L24.10
		ENCP00000048058	BAG6
			BAG6
			BAG6
			BAG6
			BAG6
			BAG6
			BAG6
		ENCP00000048114	HSPA1L
			HSPA1L
			HSPA1L
			HSPA1L
			HSPA1L
		ENCP00000048038	NCR3
			NCR3
			NCR3
			NCR3
			NCR3
			NCR3
			NCR3
			NCR3
		ENCP00000048003	HLA-B
			HLA-B
			HLA-B
			HLA-B
			HLA-B

Locus 105 (sentinel SNP rs3094005 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

			HLA-B
		ENCP00000048004	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000048039	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3
		ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000048032	LTA LTA LTA LTA LTA LTA LTA
		ENCP00000048063	GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048026	DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B
		ENCP00000047963	GTF2H4 GTF2H4 GTF2H4 GTF2H4 GTF2H4 GTF2H4
		ENCP00000048020	MICB MICB MICB MICB MICB MICB MICB MICB
		ENCP00000048078	ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A
ENCE00000445159	1	ENCP00000048082	LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D

			LY6G6D LY6G6E LY6G6D LY6G6D LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D
ENCE00000444953	1	ENCP00000048010	ZDHHC20P2 ZDHHC20P2 ZDHHC20P2 ZDHHC20P2 ZDHHC20P2
		ENCP00000048049	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCE00000444759	1	ENCP00000048071	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048129	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000047980	PSORS1C2 PSORS1C2 POLR2LP PSORS1C2 POLR2LP PSORS1C2 PSORS1C2 PSORS1C2 POLR2LP POLR2LP POLR2LP POLR2LP
		ENCP00000047987	POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1
		ENCP00000047988	POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1
ENCE00000444783	1	ENCP00000047982	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
		ENCP00000048000	WASF5P WASF5P

			WASF5P WASF5P WASF5P WASF5P WASF5P
		ENCP00000047981	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
ENCE00000444795	1	ENCP00000048115	C6orf48 C6orf48 C6orf48 C6orf48 C6orf48
		ENCP00000048132	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000048076	ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A
		ENCP00000048055	BAG6 BAG6 BAG6 BAG6 BAG6 BAG6 BAG6
		ENCP00000048135	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000048105	VARS VARS VARS VARS VARS VARS VARS
		ENCP00000048130	ZBTB12 ZBTB12 ZBTB12 ZBTB12 ZBTB12
		ENCP00000048102	VARS VARS VARS VARS VARS VARS VARS
		ENCP00000048091	CLIC1 CLIC1 CLIC1 CLIC1 CLIC1

	CLIC1
	CLIC1
ENCP00000048123	EHMT2
	EHMT2
	EHMT2
	EHMT2
	EHMT2
	EHMT2
	EHMT2
ENCP00000048092	CLIC1
	CLIC1
	CLIC1
	CLIC1
	MSH5
	CLIC1
	CLIC1
	CLIC1
	MSH5
	MSH5
	MSH5
	MSH5
	MSH5
	MSH5
	MSH5
ENCP00000048103	VARS
	VARS
	VARS
	VARS
	VARS
	VARS
	VARS
ENCP00000048070	LY6G5C
	LY6G5C
	LY6G5C
	LY6G5C
	LY6G5C
	LY6G5C
	LY6G5C
ENCP00000048054	BAG6
	BAG6
	BAG6
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	BAG6
	BAG6
	BAG6
ENCP00000048140	CFB
	CFB
	CFB
	CFB
	CFB
	CFB
	CFB
	CFB
ENCP00000048077	ABHD16A
	ABHD16A
	ABHD16A
	ABHD16A
	ABHD16A
	ABHD16A
	ABHD16A
ENCP00000048090	CLIC1
	CLIC1
	CLIC1
	CLIC1
	CLIC1
	CLIC1
	CLIC1
ENCP00000047984	TCF19
	TCF19
	TCF19

		TCF19
		TCF19
		TCF19
		TCF19
	ENCP00000048061	C6orf47
		C6orf47
		C6orf47
		C6orf47
		C6orf47
		C6orf47
		C6orf47
	ENCP00000048119	SLC44A4
		SLC44A4
		SLC44A4
		SLC44A4
		SLC44A4
		SLC44A4
		SLC44A4
		SLC44A4
	ENCP00000048117	NEU1
		NEU1
		NEU1
		NEU1
		NEU1
		NEU1
		NEU1
		NEU1
	ENCP00000048111	LSM2
		LSM2
		LSM2
		LSM2
		LSM2
		LSM2
	ENCP00000048127	EHMT2
		EHMT2
		EHMT2
		EHMT2
		EHMT2
		EHMT2
		EHMT2
	ENCP00000048154	STK19
		STK19
		STK19
		STK19
		STK19
		STK19
	ENCP00000048128	C2
		C2
		EHMT2
		EHMT2
		C2
		EHMT2
		C2
		EHMT2
		EHMT2
		C2
		C2
		C2
		EHMT2
		C2
		EHMT2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487811 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	DNase1, H3K4me2
		blood (K562)	Ini1, H3K27ac, Max, H3K9ac, H2AZ, H3K4me3, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		liver (HepG2)	DNase1, FOSL2, Jund, Srf, H3K4me3, PolII, H3K4me1, H3K4me2, FOXA1, H3K27ac, H3K36me3

		blood (GM12878)	IRF4, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1, H2AZ, Cjun, PolII, Srf, BATF
		lung (IMR90)	H3K4me3, H3K4me2, DNase1, H4K8ac
		nervous (NH-A)	DNase1, H3K4me2
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me2
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Cjun, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me1
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me3, H3K9ac, DNase1, H3K4me1, H3K27ac
		endothelium (HUVEC)	H3K36me3, PolII, H3K4me2, Cjun
		A549	DNase1, H3K4me2, H3K4me3
ENSR00001703036 (CTCF binding site)	3	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	CTCF
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (DND-41)	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001404918 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3, H3K36me3
		blood (GM12878)	POU2F2, PolII, H3K4me3, H3K4me2, H3K36me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K36me3, H3K27me3
ENSR00001703038 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, Yy1, H3K4me2, DNase1
		HSMMtube	H3K27me3, CTCF, DNase1
		blood (K562)	H3K27me3, PolII, H2AZ, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF, DNase1, H3K27me3
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	Rad21, H2AZ, H3K4me2, CTCF, H3K27me3, DNase1
		lung (IMR90)	DNase1, H3K27me3, CTCF
		blood (GM12878)	DNase1, H3K4me3, H2AZ, Rad21, H3K4me2, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	H3K4me2, CTCF, H3K27me3
		NHLF	CTCF, H3K27me3
		Osteobl	H3K4me2, CTCF, H3K27me3
		blood (DND-41)	H3K4me1, CTCF, H3K27me3
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF, H3K27me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me3, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3, CTCF
		A549	H3K27me3, DNase1, CTCF, H3K4me3
ENSR00001703043 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00000662334 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, PolII, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF, H3K4me3, H3K4me2
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me2, CTCF, H3K27me3, DNase1
		lung (IMR90)	H3K27me3, CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	CTCF, DNase1
		NHLF	CTCF
		Osteobl	H3K4me2, CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3, CTCF
		A549	CTCF

ENSR00001703045 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00000129758 (promoter flanking region)	4	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, DNase1, TAF7, PolII, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K4me2, H2AZ, H3K36me3
		blood (K562)	H3K27me3, Cfos
		skin (NHDF-AD)	DNase1, H3K27me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		A549	DNase1, H3K4me3
		skin (NHEK)	H3K4me3, DNase1, H3K4me2, H3K9ac
ENSR00000264406 (promoter)	1	embryonic stem cell (H1ESC)	H3K9ac, H3K36me3, H3K27me3, PolII, Rad21, TAF7, CTCF, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	CTCF, H3K4me2, DNase1, H3K27me3, H2AZ
		blood (K562)	Max, ZBTB7A, Rad21, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, CTCF, DNase1, H3K4me3, Ini1, H3K27me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K4me2
		liver (HepG2)	DNase1, H3K27me3, PolII, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3
		lung (IMR90)	H3K4me2, H3K4me3, CTCF
		blood (GM12878)	PolII, H2AZ, DNase1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII
		monocytes (Monocytes-CD14+)	H3K9ac, H4K20me1, H3K4me3, H3K27ac, H3K4me2, DNase1, H3K4me1, CTCF
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, CTCF, DNase1
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000487848 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, PolII, TAF7, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	Max, ZBTB7A, CTCF, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2
		liver (HepG2)	PolII, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4me2
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K4me2, H3K4me3, DNase1, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, CTCF, H3K4me1
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K4me3
		endothelium (HUVEC)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac
ENSR00000487857 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, Egr1, FOSL1, Gabp, Sin3Ak20, CTCF, SIX5, Yy1, TAF1, H3K4me2, SP1, H3K4me3, H3K36me3, H3K27me3, PolII
		HSMMtube	DNase1, H3K36me3, H3K4me2, H3K9ac, CTCF
		blood (K562)	H2AZ, Rad21, TAF1, ELF1, PolII, H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, H3K79me2, Gabp, H3K27me3, Brg1, SIX5, H3K27ac, Max, H3K4me1, H3K9ac
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H3K9ac, H3K36me3, DNase1, CTCF, H3K4me2
		liver (HepG2)	Gabp, PolII, Rad21, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc,

		blood (GM12878)	H3K4me3, H3K36me3, DNase1 H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac, H3K79me2, Rad21, SIX5, PolII, BCLAF1, H2AZ, ETS1, DNase1, Yy1, Gabp, ELF1
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3, CTCF
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2, H3K36me3
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, CTCF, H3K36me3, DNase1
		NHLF	DNase1, CTCF, H3K27ac
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me3, H3K36me3, H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac
		breast (HMEC)	DNase1, H3K4me3, H3K4me1, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, H3K4me3, Gabp, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	CTCF, H3K4me2, H3K36me3, H3K4me3, DNase1
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, PolII, H3K4me2, H3K9ac
		A549	H3K36me3, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00001703058 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Egr1, FOSL1, Gabp, Sin3Ak20, CTCF, Yy1, TAF1, H3K4me2, SP1, H3K4me3
		HSMMtube	CTCF, H3K4me2, DNase1
		blood (K562)	H3K27me3, PolII, Brg1, DNase1, H3K27ac, H2AZ, Rad21, H3K4me3, H3K4me2, CTCF
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	CTCF, H3K4me2, DNase1
		liver (HepG2)	Rad21, H2AZ, H3K4me2, CTCF, H3K4me3, DNase1, Gabp
		lung (IMR90)	CTCF, H3K4me3, H3K4me2, DNase1
		blood (GM12878)	H2AZ, DNase1, Rad21, H3K4me3, H3K4me2, CTCF
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	DNase1, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K4me3, Gabp, CTCF
		monocytes (Monocytes-CD14+)	H3K4me3, DNase1, CTCF, H3K4me2
		endothelium (HUVEC)	H3K36me3, DNase1, H3K4me2, H3K9ac, CTCF
		A549	H3K4me3, H3K4me2, DNase1, CTCF, H3K9ac
ENSR00000487869 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, Nrsf
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	PolII, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K36me3
		skin (NHEK)	DNase1
ENSR00000264421 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, Yy1, DNase1
		HSMMtube	DNase1, CTCF
		blood (K562)	PolII, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, H2AZ, H3K4me2, CTCF, DNase1
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF, Yy1, H2AZ, DNase1
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF, DNase1
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1
		A549	DNase1, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL662801.1	upstream gene variant	4834	ENST00000617927	?	?	1
AL671883.1	downstream gene variant,	2210	ENST00000578513	?	?	3

ATP6V1G2-DDX39B	upstream gene variant downstream gene variant	2711	ENST00000480131 ?	ENSP00000420191	1
ATP6V1G2-DDX39B	downstream gene variant	4495	ENST00000475917 ?	?	1
DDX39B	upstream gene variant	4481	ENST00000428450 ?	ENSP00000405707	1
DDX39B	upstream gene variant	4453	ENST00000462256 ?	?	1
DDX39B	upstream gene variant	4452	ENST00000428098 ?	ENSP00000392672	1
DDX39B	upstream gene variant	2228	ENST00000484566 ?	ENSP00000436220	1
DDX39B	upstream gene variant	4939	ENST00000427214 ?	ENSP00000399371	1
DDX39B	upstream gene variant	4069	ENST00000482195 ?	?	1
DDX39B	upstream gene variant	4986	ENST00000419338 ?	ENSP00000410313	1
DDX39B	upstream gene variant	4469	ENST00000481456 ?	?	1
DDX39B	upstream gene variant	4489	ENST00000431908 ?	ENSP00000408000	1
DDX39B	upstream gene variant	4741	ENST00000449757 ?	ENSP00000409426	1
DDX39B	upstream gene variant	3192	ENST00000417023 ?	ENSP00000406422	1
DDX39B	downstream gene variant, upstream gene variant	1116	ENST00000456976 ?	ENSP00000393984	2
DDX39B	downstream gene variant, upstream gene variant	1128	ENST00000418897 ?	ENSP00000399841	2
DDX39B	upstream gene variant	4022	ENST00000396172 NM_004640.6	ENSP00000379475	1
DDX39B	upstream gene variant	4022	ENST00000458640 NM_080598.5	ENSP00000416269	1
DDX39B	downstream gene variant, upstream gene variant	1179	ENST00000419020 ?	ENSP00000405245	2
DDX39B	upstream gene variant	4533	ENST00000456662 ?	ENSP00000416350	1
DDX39B-AS1	upstream gene variant, downstream gene variant	3343	ENST00000420520 ?	?	2
DDX39B-AS1	upstream gene variant, downstream gene variant	3332	ENST00000416684 ?	?	2
DHFRP2	downstream gene variant, upstream gene variant	756	ENST00000414224 ?	?	8
FGFR3P1	upstream gene variant, downstream gene variant	806	ENST00000449999 ?	?	4
HCG22	upstream gene variant	3076	ENST00000426185 ?	?	1
HCG22	upstream gene variant	2319	ENST00000565192 ?	?	2
HCG22	upstream gene variant	2369	ENST00000562344 ?	?	2
HCG22	upstream gene variant	3071	ENST00000570223 ?	?	1
HCG22	upstream gene variant	3076	ENST00000566475 ?	?	1
HCG22	upstream gene variant	2941	ENST00000615046 ?	?	1
HCG27	downstream gene variant	406	ENST00000383331 ?	ENSP00000453364	1
HCG27	downstream gene variant	1152	ENST00000424675 ?	ENSP00000453627	1
HCG27	downstream gene variant	3890	ENST00000414008 ?	?	1
HCG27	downstream gene variant	1373	ENST00000415276 ?	?	1
HCP5	upstream gene variant, downstream gene variant	215	ENST00000460889 ?	?	10
HCP5	upstream gene variant, downstream gene variant	268	ENST00000467369 ?	?	8
HCP5	upstream gene variant, downstream gene variant	212	ENST00000541196 ?	?	10
HCP5	downstream gene variant	107	ENST00000414046 ?	?	6

HLA-B	downstream gene variant, upstream gene variant	382	ENST00000412585	NM_005514.6	ENSP00000399168	14
HLA-B	downstream gene variant, upstream gene variant	1109	ENST00000434333	?	ENSP00000405931	10
HLA-B	downstream gene variant, upstream gene variant	1632	ENST00000463574	?	?	6
HLA-B	downstream gene variant, upstream gene variant	382	ENST00000497377	?	?	9
HLA-B	downstream gene variant, upstream gene variant	1392	ENST00000474381	?	?	9
HLA-B	downstream gene variant, upstream gene variant	382	ENST00000481849	?	?	9
HLA-B	downstream gene variant, upstream gene variant	1118	ENST00000498007	?	?	10
HLA-C	upstream gene variant	1139	ENST00000620806	?	ENSP00000480331	7
HLA-S	downstream gene variant, upstream gene variant	1332	ENST00000425174	?	?	6
LINC01149	upstream gene variant	1115	ENST00000430364	?	?	1
LST1	downstream gene variant	2115	ENST00000211921	?	ENSP00000211921	1
LST1	downstream gene variant	2263	ENST00000376092	?	ENSP00000365260	1
LST1	downstream gene variant	2016	ENST00000396101	?	ENSP00000379408	1
LST1	downstream gene variant	2016	ENST00000303757	NM_205840.2	ENSP00000303649	1
LST1	downstream gene variant	2016	ENST00000376093	NM_007161.3	ENSP00000365261	1
LST1	downstream gene variant	2016	ENST00000418507	NM_205838.2	ENSP00000405900	1
LST1	downstream gene variant	2016	ENST00000376096	?	ENSP00000365264	1
LST1	downstream gene variant	2016	ENST00000376090	?	ENSP00000365258	1
LST1	downstream gene variant	2115	ENST00000339530	NM_205837.2	ENSP00000339201	1
LST1	downstream gene variant	2168	ENST00000376110	?	ENSP00000365278	1
LST1	downstream gene variant	2168	ENST00000460834	?	?	1
LST1	downstream gene variant	2016	ENST00000464526	?	?	1
LST1	downstream gene variant	2249	ENST00000490742	?	ENSP00000418578	1
LST1	downstream gene variant	3084	ENST00000419073	?	?	1
LST1	downstream gene variant	3195	ENST00000433492	?	ENSP00000399869	1
LST1	downstream gene variant	2016	ENST00000376089	?	ENSP00000365257	1
LST1	downstream gene variant	2016	ENST00000376099	?	ENSP00000365267	1
LST1	downstream gene variant	2016	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	downstream gene variant	2017	ENST00000376086	NM_001166538.1	ENSP00000365254	1
LST1	downstream gene variant	2168	ENST00000464044	?	ENSP00000417645	1
LST1	downstream gene variant	2299	ENST00000396112	?	ENSP00000379418	1
LTA	downstream gene variant	4749	ENST00000489638	?	?	1
LTA	downstream gene variant	4749	ENST00000471842	?	?	1
LTA	downstream gene variant	4749	ENST00000454783	NM_001159740.2	ENSP00000403495	1
LTA	downstream gene variant	4749	ENST00000418386	NM_000595.3	ENSP00000413450	1
LTB	downstream gene variant	1482	ENST00000446745	NM_009588.1	ENSP00000416113	1

LTB	downstream gene variant	1551	ENST00000482429	?	?	1
LTB	downstream gene variant	1452	ENST00000483972	?	?	1
LTB	downstream gene variant	1482	ENST00000429299	NM_002341.1	ENSP00000410481	1
MICA	downstream gene variant	3039	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	downstream gene variant	3039	ENST00000421350	?	ENSP00000402410	1
MICA	downstream gene variant	3041	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICB	upstream gene variant, downstream gene variant	523	ENST00000538442	NM_001289160.1	ENSP00000442345	3
MICB	upstream gene variant, downstream gene variant	845	ENST00000252229	NM_005931.4	ENSP00000252229	4
MICB	upstream gene variant, downstream gene variant	816	ENST00000399150	NM_001289161.1	ENSP00000382103	4
MICB	upstream gene variant	3938	ENST00000494577	?	?	1
MIR6891	downstream gene variant, upstream gene variant	1734	ENST00000618788	?	?	6
NFKBIL1	upstream gene variant	1191	ENST00000473655	?	?	1
NFKBIL1	upstream gene variant	400	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	1
NFKBIL1	upstream gene variant	1143	ENST00000376145	NM_001144961.1	ENSP00000365315	1
NFKBIL1	upstream gene variant	1170	ENST00000496233	?	ENSP00000437148	1
NFKBIL1	upstream gene variant	1111	ENST00000376148	NM_005007.3	ENSP00000365318	1
PPIAP9	downstream gene variant	3776	ENST00000403866	?	?	1
RNU6-283P	upstream gene variant, downstream gene variant	827	ENST00000364788	?	?	6
SNORD117	upstream gene variant	1254	ENST00000364915	?	?	1
SNORD84	downstream gene variant	3398	ENST00000584275	?	?	1
TNF	downstream gene variant	737	ENST00000449264	NM_000594.3	ENSP00000398698	1
XXbac-BPG16N22.5	upstream gene variant	275	ENST00000538358	?	?	2
XXbac-BPG181B23.6	downstream gene variant, upstream gene variant	440	ENST00000440087	?	?	9
XXbac-BPG181B23.7	downstream gene variant	1403	ENST00000606743	?	?	1
XXbac-BPG248L24.12	upstream gene variant, downstream gene variant	660	ENST00000603274	?	?	8
XXbac-BPG299F13.14	upstream gene variant	2456	ENST00000606909	?	?	1
XXbac-BPG299F13.15	downstream gene variant	3713	ENST00000546340	?	?	1
XXbac-BPG299F13.16	upstream gene variant	1748	ENST00000538455	?	?	1
Y_RNA	downstream gene variant, upstream gene variant	480	ENST00000383850	?	?	2
ZDHHC20P2	upstream gene variant, downstream gene variant	1535	ENST00000424108	?	?	5

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V1G2	ENST00000376151	NM_001204078.1	ENSP00000365321	1
ATP6V1G2	ENST00000303892	NM_130463.3	ENSP00000302194	1
ATP6V1G2	ENST00000481998	?	?	1

ATP6V1G2-DDX39B	ENST00000376185	?	ENSP00000365356	1
ATP6V1G2-DDX39B	ENST00000475917	?	?	1
ATP6V1G2-DDX39B	ENST00000480131	?	ENSP00000420191	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V1G2	ENST00000415099	?	ENSP00000390148	1
ATP6V1G2	ENST00000483170	?	?	1
ATP6V1G2	ENST00000483251	NM_138282.2	ENSP00000419698	1
ATP6V1G2-DDX39B	ENST00000376185	?	ENSP00000365356	2
DDX39B	ENST00000427214	?	ENSP00000399371	1
DDX39B	ENST00000376177	?	ENSP00000365347	1
DDX39B	ENST00000449757	?	ENSP00000409426	1
DDX39B	ENST00000456662	?	ENSP00000416350	1
DDX39B	ENST00000462256	?	?	1
DDX39B	ENST00000458640	NM_080598.5	ENSP00000416269	1
DDX39B	ENST00000396172	NM_004640.6	ENSP00000379475	1
DDX39B	ENST00000481456	?	?	1
DDX39B	ENST00000419338	?	ENSP00000410313	1
DDX39B	ENST00000431908	?	ENSP00000408000	1
DDX39B	ENST00000482195	?	?	1
DDX39B	ENST00000428098	?	ENSP00000392672	1
DDX39B	ENST00000428450	?	ENSP00000405707	1
HCP5	ENST00000467369	?	?	7
HCP5	ENST00000414046	?	?	10
HLA-C	ENST00000620806	?	ENSP00000480331	57
LINC01149	ENST00000430364	?	?	2
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	ENST00000421350	?	ENSP00000402410	1
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICB	ENST00000252229	NM_005931.4	ENSP00000252229	2
MICB	ENST00000538442	NM_001289160.1	ENSP00000442345	3
MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	2
NCR3	ENST00000491161	?	?	1
NCR3	ENST00000376071	?	ENSP00000365239	1
NCR3	ENST00000376072	NM_001145467.1	ENSP00000365240	1
NCR3	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	ENST00000340027	NM_147130.2	ENSP00000342156	1
NCR3	ENST00000495600	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
DHFRP2	ENST00000414224	?	1

HCP5	ENST00000414046	?	3
HCP5	ENST00000541196	?	1
LINC01149	ENST00000430364	?	1
MICB	ENST00000494577	?	1
XXbac-BPG181B23.6	ENST00000440087	?	1
ZDHHC20P2	ENST00000424108	?	2

Locus 106 (rs867186)

SNiPAcard

Block annotations

Block info

genomic range	chr20:33,537,157-33,796,192
block size	259,036 bp
variant count	168 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.337 [-3.949 – 2.371]	gene(s) hit or close-by	EDEM2, GSS, MIR499A, MYH7B, PROCR, RNA5SP483, RNU6-407P, RP11-42O4.2, RP4-614O4.12, SNORD56, TRPC4AP
phastCons	μ = 0.091 [0 – 0.976]	eQTL gene(s)	ACSS2, CPNE1, EDEM2, EIF6, MAP1LC3A, MMP24, MMP24-AS1, PROCR, RBL1, RP4-614O4.11, TRPC4AP, UQCC1
GERP++	μ = -0.393 [-8.99 – 2.92]	potentially regulated gene(s)	ACSS2, C20orf173, CEP250, CPNE1, EDEM2, EIF6, ERGIC3, FER1L4, GDF5OS, GGT7, GSS, HMGB3P1, MMP24, MT1P3, MYH7B, NCOA6, NFS1, PIGU, RBM12, RP3-477O4.5, RPL37P1, SPAG4, TP53INP2, TRPC4AP
CADD score	μ = 4.335 [0.006 – 18.9]	disease gene(s)	GSS, MYH7B, NFS1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Protein C levels	$<1.00\times10^{-64}$	GWAS Catalog	25376901	1
Amyotrophic lateral sclerosis (sporadic)	$<4.00\times10^{-6}$	GWAS Catalog	24529757	1
Prothrombin time	$<5.00\times10^{-13}$	GWAS Catalog	22703881	1
Hemostatic factors and hematological phenotypes	$<4.00\times10^{-34}$	GWAS Catalog	22443383	2
Anticoagulant levels	$<4.00\times10^{-9}$	GWAS Catalog	22216198	1
D-dimer levels	$<4.00\times10^{-6}$	GWAS Catalog	21502573	1
Protein C levels	$<2.00\times10^{-200}$	GWAS Catalog	20802025	1
Coagulation factor levels	$<6.00\times10^{-37}$	GWAS Catalog	20231535	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM052909	1

Disease gene annotation

gene	trait	source DB	source entry/link
GSS	GLUTATHIONE SYNTHETASE DEFICIENCY OF ERYTHROCYTES, HEMOLYTIC [...]	OMIM	MIM:231900
GSS	GLUTATHIONE SYNTHETASE DEFICIENCY	OMIM	MIM:266130
GSS	Glutathione synthetase deficiency with 5-oxoprolinuria	OrphaNet	OrphaNet:289846
GSS	Glutathione synthetase deficiency without 5-oxoprolinuria	OrphaNet	OrphaNet:289849
MYH7B	Left ventricular noncompaction	OrphaNet	OrphaNet:54260
NFS1	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	OrphaNet	OrphaNet:397593

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PROCR	missense variant	ENST00000216968	NM_006404.4	ENSP00000216968	G/S	Ggt/Agt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
EIF6	?	ENSG00000242372	skeletal muscle	2.24×10^{-7} (p-value)	GTEx Portal V6	99
RP4-614O4.11	?	ENSG00000261582	skeletal muscle	8.95×10^{-7} (p-value)	GTEx Portal V6	85
RP4-614O4.11	?	ENSG00000261582	lung	2.19×10^{-7} (p-value)	GTEx Portal V6	87
EIF6	?	ENSG00000242372	lung	6.41×10^{-8} (p-value)	GTEx Portal V6	111
PROCR	?	ENSG00000101000	transformed fibroblasts	1.38×10^{-13} (p-value)	GTEx Portal V6	165
TRPC4AP	?	ENSG00000100991	blood	2.31×10^{-10} (p-value)	GTEx Portal V6	135
EIF6	?	ENSG00000242372	blood	1.30×10^{-7} (p-value)	GTEx Portal V6	51
MAP1LC3A	?	ENSG00000101460	muscularis mucosae	3.42×10^{-5} (p-value)	GTEx Portal V6	5
PROCR	ENST00000216968	ILMN_1717262	skin	7.54×10^{-12} (p-value)	MuTHER consortium	28
			adipocyte	3.24×10^{-15} (p-value)	MuTHER consortium	28
ACSS2	ENST00000253382	ILMN_2336595	blood	1.24×10^{-5} (p-value)	Westra et al.	26
ACSS2	ENST00000481284					
ACSS2	ENST00000360596					
ACSS2	ENST00000494727					
ACSS2	ENST00000477932					
ACSS2	ENST00000476922					
ACSS2	ENST00000253382	ILMN_1714197	blood	1.30×10^{-6} (p-value)	Westra et al.	1
ACSS2	ENST00000481284					
ACSS2	ENST00000360596					
ACSS2	ENST00000494727					
ACSS2	ENST00000477932					
ACSS2	ENST00000476922					
EIF6	ENST00000447927	ILMN_1787410	monocyte	1.17×10^{-9} (p-value)	Fairfax et al.	8
EIF6	ENST00000621148					
EIF6	ENST00000374436					
EIF6	ENST00000440766					
EIF6	ENST00000462894					
EIF6	ENST00000374450					
EIF6	ENST00000374443					
?	?	ILMN_2387636	monocyte	3.21×10^{-11} (p-value)	Fairfax et al.	8
CPNE1	ENST00000397443	ILMN_2307025	monocyte	2.55×10^{-4} (p-value)	Fairfax et al.	6
CPNE1	ENST00000317677					
CPNE1	ENST00000498056					
CPNE1	ENST00000397445					
CPNE1	ENST00000415920					
CPNE1	ENST00000437340					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					

CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
UQCC1	ENST00000374384	ILMN_2381121	monocyte	1.44×10-4 (p-value)	Fairfax et al.	1
UQCC1	ENST00000359226					
UQCC1	ENST00000457259					
UQCC1	ENST00000482440					
UQCC1	ENST00000374380					
UQCC1	ENST00000472559					
UQCC1	ENST00000374394					
UQCC1	ENST00000453855					
UQCC1	ENST00000349714					
UQCC1	ENST00000496812					
UQCC1	ENST00000473982					
UQCC1	ENST00000424405					
UQCC1	ENST00000397556					
UQCC1	ENST00000374385					
RBL1	ENST00000373664	ILMN_1782745	monocyte	2.91×10-4 (p-value)	Fairfax et al.	2
EDEM2	?	ENSG00000088298	atrial appendage	1.23×10-8 (p-value)	GTEx Portal V6	28
PROCR	?	ENSG00000101000	EBV lymphocytes	3.94×10-9 (p-value)	GTEx Portal V6	66
RP4-614O4.11	?	ENSG00000261582	sun exposed skin	7.45×10-6 (p-value)	GTEx Portal V6	4
MMP24-AS1	?	ENSG00000126005	sun exposed skin	7.63×10-8 (p-value)	GTEx Portal V6	5
MMP24	?	ENSG00000125966	sun exposed skin	1.20×10-6 (p-value)	GTEx Portal V6	3
EIF6	ENST00000447927	ILMN_1787410	monocyte	1.97×10-24 (p-value)	Zeller et al.	12
EIF6	ENST00000440766					
EIF6	ENST00000374436					
EIF6	ENST00000621148					
EIF6	ENST00000462894					
EIF6	ENST00000374450					
EIF6	ENST00000374443					
?	?	ILMN_2387636	monocyte	1.83×10-26 (p-value)	Zeller et al.	12
TRPC4AP	ENST00000451813	ILMN_2402805	monocyte	3.59×10-17 (p-value)	Zeller et al.	12
TRPC4AP	ENST00000252015					
MMP24	?	ENSG00000125966	esophagus mucosa	4.69×10-6 (p-value)	GTEx Portal V6	2
EDEM2	?	ENSG00000088298	tibial artery	2.09×10-5 (p-value)	GTEx Portal V6	3
PROCR	?	ENSG00000101000	spleen	7.90×10-7 (p-value)	GTEx Portal V6	15
EDEM2	?	ENSG00000088298	left ventricle	1.93×10-5 (p-value)	GTEx Portal V6	5
PROCR	ENST00000216968	ILMN_1717262	liver	1.17×10-11 (p-value)	Innocenti et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000293121	1	ENCP00000031345	MYH7B
ENCE00000293052	1	ENCP00000031340	ACSS2
		ENCP00000031363	GDF5OS
		ENCP00000031370	CEP250
ENCE00000293009	1	ENCP00000031333	HMGB3P1
ENCE00000293100	1	ENCP00000031343	MYH7B
ENCE00000293101	1	ENCP00000031343	MYH7B
ENCE00000293304	1	ENCP00000031379	C20orf173
ENCE00000293169	1	ENCP00000031376	CEP250
		ENCP00000031354	MT1P3
ENCE00000293330	1	ENCP00000031391	RPL37P1
ENCE00000292949	1	ENCP00000031339	ACSS2
		ENCP00000031338	ACSS2
		ENCP00000031376	CEP250
		ENCP00000031371	CEP250
		ENCP00000031324	PIGU
ENCE00000292951	1	ENCP00000031339	ACSS2
		ENCP00000031324	PIGU
		ENCP00000031383	ERGIC3
ENCE00000293241	1	ENCP00000031369	CEP250
		ENCP00000031368	CEP250
ENCE00000292990	1	ENCP00000031386	ERGIC3
		ENCP00000031353	MT1P3
		ENCP00000031385	ERGIC3
		ENCP00000031328	TP53INP2
		ENCP00000031337	ACSS2
		ENCP00000031355	EIF6
		ENCP00000031393	FER1L4
		ENCP00000031367	CEP250
		ENCP00000031332	NCOA6
		ENCP00000031327	TP53INP2
		ENCP00000031380	ERGIC3
		ENCP00000031378	RP3-477O4.5
		ENCP00000031326	PIGU
		ENCP00000031346	MYH7B
		ENCP00000031406	NFS1
		ENCP00000031335	GGT7 ACSS2
		ENCP00000031387	FER1L4
		ENCP00000031398	SPAG4
		ENCP00000031342	GSS
		ENCP00000031389	FER1L4
		ENCP00000031336	ACSS2
		ENCP00000031334	GGT7
		ENCP00000031392	FER1L4
		ENCP00000031390	FER1L4
		ENCP00000031366	CEP250
		ENCP00000031348	EDEM2
		ENCP00000031351	MMP24
		ENCP00000031381	ERGIC3
		ENCP00000031405	CPNE1 RBM12

Locus 106 (sentinel SNP rs867186 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

	ENCP00000031347		TRPC4AP

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001665252 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		Osteobl	CTCF
ENSR00000400200 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
ENSR00000234246 (open chromatin region)	1	lung (IMR90)	H3K27me3
		blood (K562)	DNase1
ENSR00001665257 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	PolII
		A549	H3K36me3
ENSR00000400209 (open chromatin region)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00001448520 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3, DNase1
		blood (GM12878)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		NHLF	DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001665259 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001665262 (open chromatin region)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (K562)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001665263 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3

		blood (K562)	H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		nervous (NH-A)	H3K36me3
ENSR00000400218 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (K562)	H3K79me2, H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3, H3K79me2
		cervix (HeLa-S3)	H3K36me3, H3K79me2
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K79me2, H3K36me3
		lung (IMR90)	H3K79me2, H3K36me3
		blood (GM12878)	H3K79me2
		A549	H3K36me3
ENSR00001665264 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		Osteobl	CTCF
		blood (K562)	CTCF
		blood (DND-41)	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		liver (HepG2)	H2AZ, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	CTCF
		nervous (NH-A)	CTCF
		A549	CTCF
		skin (NHEK)	CTCF
ENSR00001448525 (promoter flanking region)	2	cervix (HeLa-S3)	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K36me3
		A549	H3K36me3
		blood (K562)	H3K27me3
		skin (NHEK)	DNase1
		breast (HMEC)	DNase1
ENSR00001448526 (promoter flanking region)	2	NHLF	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac, H2AZ
		breast (HMEC)	H3K27ac
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1, H3K27ac
		A549	H3K36me3
ENSR00000400229 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		blood (K562)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		NHLF	DNase1, H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3

		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K27ac
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun, H3K4me1, H3K27ac, PolII, DNase1
		A549	H3K36me3
ENSR00001665267 (enhancer)	1	skin (NHDF-AD)	DNase1
ENSR00000234264 (enhancer)	1	embryonic stem cell (H1ESC)	H3K4me1, H3K4me2
		endothelium (HUVEC)	Cmyc, DNase1, PolII, Max, Cjun
		blood (K562)	DNase1
ENSR00001448528 (CTCF binding site)	1	endothelium (HUVEC)	H3K36me3
		blood (DND-41)	H3K27me3
		A549	H3K36me3
ENSR00001665271 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, DNase1, Rad21, CTCF
		skin (NHDF-AD)	H3K4me3, CTCF, DNase1, H3K4me2
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, DNase1, Rad21, H3K4me1, H3K4me2, FOXA1
		lung (IMR90)	CTCF
		blood (GM12878)	CTCF, Rad21
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K4me2, CTCF, H3K36me3
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF, H3K4me2
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1
		A549	CTCF, DNase1, H3K27ac, H3K4me2, H3K4me3
ENSR00000009183 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, DNase1, Rad21, CTCF
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, H3K4me2, FOXA1, CTCF, DNase1
		lung (IMR90)	CTCF
		blood (GM12878)	CTCF, Rad21
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1
		A549	H3K27ac, H3K4me3, H3K4me2, CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
EDEM2	downstream gene variant, upstream gene variant	33	ENST00000374491	NM_001145025.1	ENSP00000363615	12
EDEM2	downstream gene variant, upstream gene variant	26	ENST00000374492	NM_018217.2	ENSP00000363616	12
GSS	upstream gene variant	657	ENST00000216951	NM_000178.2	ENSP00000216951	3
GSS	upstream gene variant	4622	ENST00000451957	?	ENSP00000407517	1
MIR499A	upstream gene variant, downstream gene variant	1528	ENST00000384903	?	?	4
MYH7B	upstream gene variant, downstream gene variant	1387	ENST00000446156	?	ENSP00000395858	6
MYH7B	upstream gene variant, downstream gene variant	579	ENST00000618182	?	ENSP00000483640	9
MYH7B	upstream gene variant, downstream gene variant	1387	ENST00000435272	?	ENSP00000391939	6

MYH7B	upstream gene variant, downstream gene variant	1387	ENST00000435272	?	ENSP00000391939	6
MYH7B	upstream gene variant, downstream gene variant	437	ENST00000481922	?	?	7
MYH7B	upstream gene variant, downstream gene variant	1387	ENST00000453028	?	ENSP00000409103	6
MYH7B	upstream gene variant, downstream gene variant	730	ENST00000262873	NM_020884.3	ENSP00000262873	8
MYH7B	upstream gene variant, downstream gene variant	179	ENST00000470929	?	?	7
MYH7B	upstream gene variant, downstream gene variant	1387	ENST00000433934	?	ENSP00000412594	6
MYH7B	upstream gene variant, downstream gene variant	1387	ENST00000456649	?	ENSP00000396368	6
PROCR	upstream gene variant, downstream gene variant	604	ENST00000216968	NM_006404.4	ENSP00000216968	4
RNA5SP483	upstream gene variant, downstream gene variant	579	ENST00000363431	?	?	4
RNU6-407P	downstream gene variant, upstream gene variant	249	ENST00000365280	?	?	5
RP11-42O4.2	downstream gene variant, upstream gene variant	1832	ENST00000615962	?	?	2
RP4-614O4.12	downstream gene variant	2873	ENST00000594413	?	?	1
SNORD56	downstream gene variant, upstream gene variant	1805	ENST00000364281	?	?	9
TRPC4AP	downstream gene variant, upstream gene variant	1960	ENST00000451813	NM_199368.1	ENSP00000400614	7
TRPC4AP	downstream gene variant, upstream gene variant	1896	ENST00000252015	NM_015638.2	ENSP00000252015	7

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
MYH7B	ENST00000618182	?	ENSP00000483640	2	2	2
MYH7B	ENST00000262873	NM_020884.3	ENSP00000262873	2	2	2

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
EDEM2	ENST00000374491	NM_001145025.1	ENSP00000363615	27
EDEM2	ENST00000374492	NM_018217.2	ENSP00000363616	27
GSS	ENST00000216951	NM_000178.2	ENSP00000216951	2
GSS	ENST00000451957	?	ENSP00000407517	2
MYH7B	ENST00000618182	?	ENSP00000483640	8
MYH7B	ENST00000262873	NM_020884.3	ENSP00000262873	11
MYH7B	ENST00000470929	?	?	3
TRPC4AP	ENST00000252015	NM_015638.2	ENSP00000252015	62
TRPC4AP	ENST00000451813	NM_199368.1	ENSP00000400614	62

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
MYH7B	ENST00000470929	?	2

Locus 107 (rs17368582)

SNiPAcard

Block annotations

Block info

genomic range	chr11:102,719,534-102,777,303
block size	57,770 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.157 [-1.126 – 1.467]	gene(s) hit or close-by	BOLA3P1, MMP12, MMP3, RNU7-159P
phastCons	μ = 0.143 [0 – 0.967]	eQTL gene(s)	–
GERP++	μ = -0.305 [-6.11 – 3.83]	potentially regulated gene(s)	AP000619.6, DYNC2H1, MMP20, MMP3
CADD score	μ = 4.600 [0.023 – 17.09]	disease gene(s)	MMP3, DYNC2H1, MMP20

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Local histogram emphysema pattern	<1.00×10-8	GWAS Catalog	25006744	1

Disease gene annotation

gene	trait	source DB	source entry/link
MMP3	CORONARY HEART DISEASE, SUSCEPTIBILITY TO, 6	OMIM	MIM:614466
DYNC2H1	SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY	OMIM	MIM:613091
MMP20	AMELOGENESIS IMPERFECTA, HYPOMATURATION TYPE, IIA2	OMIM	MIM:612529
DYNC2H1	short rib-polydactyly syndrome type 3 (SRPS3)	DECIPHER	MIM:263510
DYNC2H1	asphyxiating thoracic dystrophy type 3 (ATD3)	DECIPHER	MIM:613091
DYNC2H1	Short rib-polydactyly syndrome, Verma-Naumoff type	OrphaNet	OrphaNet:93271
DYNC2H1	Short rib-polydactyly syndrome, Majewski type	OrphaNet	OrphaNet:93269
DYNC2H1	Jeune syndrome	OrphaNet	OrphaNet:474
MMP20	Hypomaturation amelogenesis imperfecta	OrphaNet	OrphaNet:100033

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000120359	1	ENCP00000014401	MMP3
		ENCP00000014410	DYNC2H1
ENCE00000120215	1	ENCP00000014398	AP000619.6
		ENCP00000014385	MMP20

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000568035 (promoter flanking region)	5	NHLF	DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	DNase1

		nervous (NH-A)	DNase1
		A549	H3K27me3
		skin (NHEK)	DNase1, H3K27ac
ENSR00001606530 (CTCF binding site)	1	NHLF	DNase1
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	DNase1
		A549	H3K27me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1
ENSR00001606534 (enhancer)	1	HSMMtube	H3K27me3
		A549	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00000568041 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BOLA3P1	upstream gene variant	2527	ENST00000530713	?	?	1
MMP12	downstream gene variant, upstream gene variant	27	ENST00000571244	NM_002426.4	ENSP00000458585	4
MMP3	upstream gene variant	5000	ENST00000299855	NM_002422.3	ENSP00000299855	1
RNU7-159P	downstream gene variant, upstream gene variant	161	ENST00000459284	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
MMP12	ENST00000571244	NM_002426.4	ENSP00000458585	P	ccG/ccA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MMP12	ENST00000571244	NM_002426.4	ENSP00000458585	2

Locus 108 (rs1190552)

SNiPAcard

Block annotations

Block info

genomic range	chr14:102,838,957-102,995,068
block size	156,112 bp
variant count	37 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.804 [-3.848 – 1.045]	gene(s) hit or close-by	ANKRD9, RNU6-244P, TECPR2
phastCons	μ = 0.035 [0 – 0.433]	eQTL gene(s)	ANKRD9, CINP, TECPR2, ZNF839
GERP++	μ = -0.650 [-4.97 – 1.33]	potentially regulated gene(s)	ANKRD9
CADD score	μ = 3.867 [0.244 – 18.09]	disease gene(s)	TECPR2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TECPR2	SPASTIC PARAPLEGIA 49, AUTOSOMAL RECESSIVE	OMIM	MIM:615031
TECPR2	Hereditary Spastic Paraparesis	DECIPHER	MIM:615031
TECPR2	Autosomal recessive spastic paraplegia type 49	OrphaNet	OrphaNet:320385

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CINP	?	ENSG00000100865	muscularis mucosae	1.64×10^{-12} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	lung	3.07×10^{-10} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	atrial appendage	4.94×10^{-7} (p-value)	GTEx Portal V6	8
ANKRD9	?	ENSG00000156381	transformed fibroblasts	3.75×10^{-25} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	transformed fibroblasts	1.39×10^{-13} (p-value)	GTEx Portal V6	35
TECPR2	?	ENSG00000196663	tibial artery	2.31×10^{-7} (p-value)	GTEx Portal V6	16
CINP	?	ENSG00000100865	tibial artery	6.65×10^{-9} (p-value)	GTEx Portal V6	34
CINP	?	ENSG00000100865	breast	8.44×10^{-10} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	blood	8.51×10^{-10} (p-value)	GTEx Portal V6	35
CINP	?	ENSG00000100865	thyroid	3.32×10^{-10} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	skeletal muscle	8.18×10^{-14} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	transverse colon	5.61×10^{-8} (p-value)	GTEx Portal V6	33
CINP	?	ENSG00000100865	EBV lymphocytes	1.82×10^{-8} (p-value)	GTEx Portal V6	11
CINP	?	ENSG00000100865	unexposed skin	7.47×10^{-9} (p-value)	GTEx Portal V6	34
CINP	?	ENSG00000100865	sun exposed skin	7.80×10^{-15} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	liver	2.33×10^{-8} (p-value)	GTEx Portal V6	12

CINP	?	ENSG00000100865	liver	3.23×10-8 (p-value)	GTEx Portal V6	13
CINP	?	ENSG00000100865	aorta	3.94×10-9 (p-value)	GTEx Portal V6	11
CINP	?	ENSG00000100865	left ventricle	3.40×10-8 (p-value)	GTEx Portal V6	13
CINP	?	ENSG00000100865	subcutaneous adipocytes	2.21×10-14 (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	visceral adipocytes	1.70×10-12 (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	tibial nerve	4.97×10-10 (p-value)	GTEx Portal V6	35
CINP	?	ENSG00000100865	sigmoid colon	7.70×10-9 (p-value)	GTEx Portal V6	35
CINP	?	ENSG00000100865	esophagus mucosa	2.53×10-15 (p-value)	GTEx Portal V6	35
CINP	?	ENSG00000100865	gastroesophageal junction	7.26×10-8 (p-value)	GTEx Portal V6	17
CINP	?	ENSG00000100865	testis	8.62×10-8 (p-value)	GTEx Portal V6	12
ZNF839	ENST00000558490	ILMN_2186858	skin	1.56×10-7 (p-value)	MuTHER consortium	4
ZNF839	ENST00000420933		adipocyte	9.15×10-6 (p-value)	MuTHER consortium	4
ZNF839	ENST00000557803					
ZNF839	ENST00000559098					
ZNF839	ENST00000561251					
ZNF839	ENST00000558850					
ZNF839	ENST00000559155					
ZNF839	ENST00000559185					
ZNF839	ENST00000442396					
ZNF839	ENST00000560112					
ZNF839	ENST00000558462					
CINP	ENST00000216756	ILMN_1765257	blood	2.69×10-5 (p-value)	Westra et al.	3
CINP	ENST00000541568					
CINP	ENST00000559504					
CINP	ENST00000559514					
CINP	ENST00000536961					
ANKRD9	ENST00000559404	ILMN_1675937	blood	5.57×10-7 (p-value)	Westra et al.	2
ANKRD9	ENST00000286918					
ANKRD9	ENST00000559651					
ANKRD9	ENST00000557902					
ANKRD9	ENST00000560748					
CINP	?	ENSG00000100865	coronary artery	3.44×10-6 (p-value)	GTEx Portal V6	2
ZNF839	ENST00000420933	ILMN_2186858	b-cell	1.27×10-4 (p-value)	Fairfax et al.	1
ZNF839	ENST00000558490					
ZNF839	ENST00000557803					
ZNF839	ENST00000559098					
ZNF839	ENST00000559155					
ZNF839	ENST00000558850					
ZNF839	ENST00000561251					
ZNF839	ENST00000559185					
ZNF839	ENST00000442396					
ZNF839	ENST00000560112					
ZNF839	ENST00000558462					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000159684	1	ENST00000559404, ENST00000557902, ENST00000559651	ANKRD9

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000424047 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27ac, H3K36me3, DNase1
ENSR00001625363 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K36me3
		blood (K562)	H3K27me3
ENSR00000424048 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, Yy1, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, CTCF, H2AZ, Rad21, Egr1
		skin (NHDF-AD)	CTCF, H3K4me2, DNase1, H3K4me3
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, CTCF, Rad21
		blood (GM12878)	CTCF, DNase1, H2AZ, Yy1, Rad21
		lung (IMR90)	H3K4me2, H3K36me3, CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	H3K36me3, CTCF, H3K4me2
		blood (DND-41)	CTCF, H3K36me3
		breast (HMEC)	CTCF, H3K4me2
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		A549	H3K4me3, CTCF, H3K36me3
ENSR00001625365 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, Rad21, CTCF, Yy1, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, CTCF, H2AZ, Rad21, Egr1
		skin (NHDF-AD)	H3K4me3, CTCF, DNase1, H3K4me2
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, DNase1
		blood (GM12878)	DNase1, H2AZ, Yy1, Rad21, CTCF
		lung (IMR90)	H3K4me2, H3K36me3, CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	H3K4me2, CTCF, H3K36me3
		blood (DND-41)	CTCF, H3K36me3
		breast (HMEC)	CTCF, H3K4me2
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		A549	H3K4me3, CTCF, H3K36me3
ENSR00000181837 (CTCF binding site)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	DNase1, CTCF, H3K36me3, Rad21
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	CTCF, Max
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF, H3K4me1, Rad21, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	CTCF, H3K36me3
ENSR00001460390 (promoter flanking region)	1	NHLF	H3K36me3, H3K27ac
		embryonic stem cell (H1ESC)	DNase1

		HSMMtube	H3K36me3
		Osteobl	H3K27ac, H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	Max
		skin (NHDF-AD)	H3K27ac, DNase1
		muscle (HSMM)	H2AZ, H3K27ac, H3K36me3, DNase1
		breast (HMEC)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	H3K27ac, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, Max, Cjun
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3, H3K9ac, H3K27ac, H3K4me1
ENSR00001625371 (promoter)	2	embryonic stem cell (H1ESC)	PolII, H3K9ac, CTCF, Yy1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me3, H2AZ, DNase1
		blood (K562)	H3K4me3, DNase1, H3K4me2, PolII, H2AZ, H3K9ac, H3K27ac, Egr1
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H3K4me2, H3K4me1, PolII, FOXA1, H3K36me3, H3K4me3, H3K27ac, H3K9ac, DNase1
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3, DNase1
		blood (GM12878)	H2AZ, H3K4me3, H3K4me2, DNase1
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, CTCF
		NHLF	DNase1
		Osteobl	H3K27ac, H3K4me3, H3K4me2, DNase1
		breast (HMEC)	H3K4me2, H3K4me3, DNase1
		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF, PolII, H3K4me3, H3K27ac, H3K4me2, H3K9ac, Ini1
		monocytes (Monocytes-CD14+)	H3K4me3, DNase1
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, H3K4me3
		A549	H3K4me3, H3K4me2, DNase1
ENSR00000181844 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, Egr1, CTCF, Yy1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	H3K4me3, CTCF, ELF1, H2AZ, H3K9ac, Rad21, ZBTB7A, Egr1
		skin (NHDF-AD)	CTCF, H3K4me2, H3K4me3
		muscle (HSMM)	H2AZ, CTCF, H3K4me3
		liver (HepG2)	DNase1, H3K4me3, CTCF, H3K4me2, Rad21
		lung (IMR90)	H3K4me2, H3K4me3, CTCF
		blood (GM12878)	DNase1, H2AZ, ELF1, Rad21, H3K4me3, H3K4me2, CTCF
		nervous (NH-A)	DNase1, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
		NHLF	H3K9ac, CTCF
		Osteobl	H3K4me3, H3K4me2, CTCF
		blood (DND-41)	H3K4me2, CTCF, H3K27me3
		breast (HMEC)	H3K4me2, H3K4me3, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K4me3, H3K4me2
		monocytes (Monocytes-CD14+)	CTCF, H3K4me2, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K4me3, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF
ENSR00000424062 (promoter flanking region)	1	embryonic stem cell (H1ESC)	SP1, USF1, H3K4me3, H3K4me2, TAF1, H3K27me3, PolII, Rad21, Gabp, TAF7, CTCF, Yy1, HDAC2, DNase1
		HSMMtube	CTCF, H2AZ, DNase1
		blood (K562)	Cfos, TAF1, H2AZ, HEY1, USF1, H3K9ac, Rad21, H3K27ac, PolII, H3K4me2, CTCF, H3K4me3, DNase1
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	DNase1, H3K4me3, CTCF, H2AZ
		liver (HepG2)	H3K4me2, USF1, TAF1, Rad21, PolII, H3K27ac, CTCF, H3K4me3, DNase1
		blood (GM12878)	EBF1, PolII, H2AZ, DNase1, Yy1, Rad21, H3K4me3, USF1, Cfos, CTCF
		lung (IMR90)	DNase1, H3K4me2, H4K5ac, H3K4me3, H3K9ac, CTCF
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	H3K27ac, H3K4me1, H3K9ac, H3K4me2, DNase1, CTCF, H3K4me3
		NHLF	H3K4me3, DNase1, CTCF, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	CTCF, H3K4me2, H3K4me3, H3K27ac, DNase1

cervix (HeLa-S3)	DNase1, CTCF, PolII, Cfos, Max, TAF1, H3K4me3, H3K27ac, H3K4me2, H3K9ac
monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K27ac, H3K4me3
endothelium (HUVEC)	H3K36me3, DNase1, CTCF, PolII, H3K4me2, H3K4me3, Max
A549	H3K9ac, CTCF, DNase1, H3K27ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANKRD9	downstream gene variant, upstream gene variant	418	ENST00000559651	?	ENSP00000454100	4
ANKRD9	downstream gene variant, upstream gene variant	2632	ENST00000560748	?	ENSP00000453650	2
ANKRD9	downstream gene variant, upstream gene variant	2101	ENST00000286918	NM_152326.2	ENSP00000286918	6
ANKRD9	downstream gene variant, upstream gene variant	324	ENST00000557902	?	ENSP00000454080	4
ANKRD9	downstream gene variant, upstream gene variant	83	ENST00000559404	?	ENSP00000453417	3
RNU6-244P	upstream gene variant, downstream gene variant	2017	ENST00000391028	?	?	6
TECPR2	downstream gene variant	1663	ENST00000561228	?	?	1
TECPR2	upstream gene variant, downstream gene variant	236	ENST00000559124	?	?	6
TECPR2	upstream gene variant	435	ENST00000557786	?	?	1
TECPR2	upstream gene variant, downstream gene variant	486	ENST00000561099	?	?	5
TECPR2	downstream gene variant	1339	ENST00000558678	NM_001172631.1	ENSP00000453671	1
TECPR2	downstream gene variant	1763	ENST00000359520	NM_014844.3	ENSP00000352510	1
TECPR2	upstream gene variant, downstream gene variant	1211	ENST00000560060	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANKRD9	ENST00000560748	?	ENSP00000453650	2
ANKRD9	ENST00000286918	NM_152326.2	ENSP00000286918	2
TECPR2	ENST00000558678	NM_001172631.1	ENSP00000453671	20
TECPR2	ENST00000359520	NM_014844.3	ENSP00000352510	25
TECPR2	ENST00000561099	?	?	1
TECPR2	ENST00000561228	?	?	11
TECPR2	ENST00000560060	?	?	3
TECPR2	ENST00000557786	?	?	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANKRD9	ENST00000286918	NM_152326.2	ENSP00000286918	1
TECPR2	ENST00000359520	NM_014844.3	ENSP00000352510	4

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANKRD9	ENST00000559404	?	ENSP00000453417	1

Locus 109 (rs6697763)

SNiPAcard

Block annotations

Block info

genomic range	chr1:9,037,273-9,037,546
block size	274 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.444 [-1.855 – -1.033]	**gene(s) hit or close-by**	CA6, RN7SL451P
phastCons	μ = 0.004 [0 – 0.008]	**eQTL gene(s)**	–
GERP++	μ = 0.421 [-0.317 – 1.16]	**potentially regulated gene(s)**	–
CADD score	μ = 3.142 [0.731 – 5.553]	**disease gene(s)**	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CA6	downstream gene variant	2770	ENST00000377436	NM_001270500.1	ENSP00000366654	2
CA6	downstream gene variant	2340	ENST00000476083	?	?	2
CA6	downstream gene variant	2122	ENST00000377443	NM_001215.3	ENSP00000366662	2
CA6	downstream gene variant	2510	ENST00000377442	NM_001270501.1	ENSP00000366661	2
RN7SL451P	downstream gene variant	2089	ENST00000583026	?	?	2

Locus 110 (rs6114248)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,692,121-23,731,560
block size	39,440 bp
variant count	41 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.414 [-4.432 – 0.242]	gene(s) hit or close-by	CST1, CST2P1
phastCons	μ = 0.013 [0 – 0.149]	eQTL gene(s)	CST1
GERP++	μ = -0.468 [-3.14 – 0.785]	potentially regulated gene(s)	CST11, CST8, CST9L, CSTL1, CSTP1, GGTLC1, XXyac-YX60D10.1
CADD score	μ = 1.308 [0.008 – 6.138]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CST1	missense variant	ENST00000398402	?	ENSP00000381439	Y/H	Tat/Cat	deleterious	unknown	1
CST1	missense variant	ENST00000304749	NM_001898.2	ENSP00000305731	Y/H	Tat/Cat	deleterious	unknown	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST1	ENST00000398402	ILMN_1753449	skin	3.41×10-6 (p-value)	MuTHER consortium	9
CST1	ENST00000304749					
CST1	?	ENSG00000170373	testis	1.75×10-6 (p-value)	GTEx Portal V6	31

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000290785	1	ENCP00000031084	CST9L
ENCE00000290706	1	ENCP00000031098	GGTLC1
		ENCP00000031079	CST11
		ENCP00000031078	CSTL1
		ENCP00000031093	XXyac-YX60D10.1
		ENCP00000031081	CST8
		ENCP00000031096	CSTP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000008781	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
(CTCF binding site)		HSMMtube	CTCF, H3K27me3, DNase1

		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, DNase1
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3, DNase1
		A549	CTCF, H3K27me3
ENSR00001664585 (enhancer)	2	embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00000399221 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001448111 (CTCF binding site)	5	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CST1	downstream gene variant	135	ENST00000398402	?	ENSP00000381439	4
CST1	downstream gene variant	33	ENST00000304749	NM_001898.2	ENSP00000305731	3
CST2P1	upstream gene variant	78	ENST00000425770	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST1	ENST00000304749	NM_001898.2	ENSP00000305731	9
CST1	ENST00000398402	?	ENSP00000381439	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST1	ENST00000304749	NM_001898.2	ENSP00000305731	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST1	ENST00000304749	NM_001898.2	ENSP00000305731	1

Locus 111 (rs10737680)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,679,455-196,704,632
block size	25,178 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.440 [-1.946 – 0.687]	gene(s) hit or close-by	CFH
phastCons	μ = 0.024 [0 – 0.272]	eQTL gene(s)	CFH, CFHR1, CFHR3
GERP++	μ = -1.327 [-6.86 – 1.73]	potentially regulated gene(s)	–
CADD score	μ = 1.228 [0.015 – 5.622]	disease gene(s)	CFHR1, CFHR3, CFH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Age-related macular degeneration	$<1.00\times10^{-434}$	GWAS Catalog	23455636	1
End-stage coagulation	$<1.00\times10^{-9}$	GWAS Catalog	23381943	1
Age-related macular degeneration	$<3.00\times10^{-64}$	GWAS Catalog	20861866	1
Age-related macular degeneration	$<2.00\times10^{-111}$	GWAS Catalog	20385826	1
Age-related macular degeneration	$<2.00\times10^{-76}$	GWAS Catalog	20385819	1
HWESASXX*	2.20×10^{-8}	Metabolomics GWAS Server	24816252	1
Macular degeneration	6.93×10^{-76}	dbGaP	pha002890	1
Macular degeneration	8.92×10^{-92}	dbGaP	pha002869	1
Macular degeneration	1.35×10^{-16}	dbGaP	pha000002	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Age-related macular degeneration 4	risk factor	ClinVar	RCV000018024.1	1
MACULAR DEGENERATION, AGE-RELATED, 4, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:134370	1

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR1	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFHR1	C3 glomerulonephritis	OrphaNet	OrphaNet:329931
CFHR1	Dense deposit disease	OrphaNet	OrphaNet:93571
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571
CFH	Familial drusen	OrphaNet	OrphaNet:75376
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFH	ENST00000359637	ILMN_1657803	skin	2.03×10^{-7} (p-value)	MuTHER consortium	6
CFH	ENST00000630130		adipocyte	3.54×10^{-7} (p-value)	MuTHER consortium	6
CFHR1	?	ENSG00000244414	transformed fibroblasts	5.58×10^{-6} (p-value)	GTEx Portal V6	19
CFHR1	?	ENSG00000244414	liver	4.07×10^{-10} (p-value)	GTEx Portal V6	19
CFHR3	?	ENSG00000116785	liver	8.03×10^{-7} (p-value)	GTEx Portal V6	19
CFHR1	?	ENSG00000244414	subcutaneous adipocytes	1.52×10^{-6} (p-value)	GTEx Portal V6	19
CFH	?	ENSG00000000971	subcutaneous adipocytes	6.47×10^{-6} (p-value)	GTEx Portal V6	19

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFH	upstream gene variant	1406	ENST00000470918	?	?	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFH	ENST00000466229	?	?	19
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	19

Locus 112 (rs419137)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,696,875-196,696,875
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-1.535	gene(s) hit or close-by	CFH
phastCons	0	eQTL gene(s)	–
GERP++	-4.04	potentially regulated gene(s)	–
CADD score	0.742	disease gene(s)	CFH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Macular degeneration	2.33×10^{-9}	dbGaP	pha002890	dbGaP	1
Macular degeneration	1.06×10^{-10}	dbGaP	pha002869	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421	orphanet
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579	orphanet
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571	orphanet
CFH	Familial drusen	OrphaNet	OrphaNet:75376	orphanet
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903	orphanet

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFH	ENST00000466229	?	?	1
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	1

Locus 113 (rs2545801)

SNiPAcard

Block annotations

Block info

genomic range	chr5:176,836,532-176,842,474
block size	5,943 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.148 [-0.974 – 1.747]	gene(s) hit or close-by	F12, GRK6
phastCons	μ = 0.095 [0.001 – 0.375]	eQTL gene(s)	F12, GRIN2B, MXD3, RAB24, RGS14
GERP++	μ = 1.385 [-0.529 – 4.44]	potentially regulated gene(s)	F12, F12
CADD score	μ = 7.601 [4.209 – 16.43]	disease gene(s)	F12, GRIN2B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Blood metabolite ratios	$<6.00\times10^{-26}$	GWAS Catalog	24816252	1
Circulating vasoactive peptide levels	$<1.00\times10^{-67}$	GWAS Catalog	23381795	1
Metabolite levels	$<3.00\times10^{-14}$	GWAS Catalog	22916037	1
Activated partial thromboplastin time	$<6.00\times10^{-88}$	GWAS Catalog	22703881	1
Metabolite levels	$<9.00\times10^{-11}$	GWAS Catalog	22286219	1
Platelet function and related traits	$<1.00\times10^{-6}$	GWAS Catalog	21546496	1
Activated partial thromboplastin time	$<2.00\times10^{-30}$	GWAS Catalog	20303064	1
bradykinin, des-arg(9)	2.76×10^{-23}	Metabolomics GWAS Server	24816252	3

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Factor xii polymorphism	benign	ClinVar	RCV000001226.2	1
?	HGMD curated	HGMD	CR982412	1
FACTOR XII POLYMORPHISM	OMIM curated	OMIM	MIM:610619	1

Disease gene annotation

gene	trait	source DB	source entry/link	
F12	FACTOR XII DEFICIENCY	OMIM	MIM:234000	
F12	ANGIOEDEMA, HEREDITARY, TYPE III	OMIM	MIM:610618	
GRIN2B	MENTAL RETARDATION, AUTOSOMAL DOMINANT 6	OMIM	MIM:613970	
F12	Hereditary angioedema type 3	OrphaNet	OrphaNet:100054	
F12	Congenital factor XII deficiency	OrphaNet	OrphaNet:330	
GRIN2B	Mental retardation, autosomal dominant 6	DECIPHER	MIM:613970	

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RAB24	ENST00000478234	ILMN_1714393	blood	1.49×10^{-5} (p-value)	MuTHER consortium	3

RAB24	ENST00000393610		adipocyte	4.14×10-5 (p-value)	MuTHER consortium	3
RAB24	ENST00000471466					
RAB24	ENST00000303270					
MXD3	ENST00000503473					
RAB24	ENST00000303251					
RAB24	ENST00000393611					
RAB24	ENST00000495458					
F12	ENST00000253496	ILMN_1671766	blood	1.14×10-6 (p-value)	Westra et al.	3
F12	ENST00000504406					
F12	ENST00000514943					
F12	ENST00000502854					
F12	ENST00000510358					
RGS14	ENST00000506944	ILMN_1696828	blood	1.13×10-6 (p-value)	Westra et al.	3
RGS14	ENST00000514102					
RGS14	ENST00000509289					
RGS14	ENST00000511890					
RGS14	ENST00000503044					
RGS14	ENST00000502731					
RGS14	ENST00000425155					
RGS14	ENST00000408923					
MXD3	?	ENSG00000213347	muscularis mucosae	7.40×10-7 (p-value)	GTEx Portal V6	5
F12	?	ENSG00000131187	liver	3.72×10-13 (p-value)	GTEx Portal V6	5
F12	ENST00000504406	ILMN_1671766	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
F12	ENST00000253496					
F12	ENST00000502854					
F12	ENST00000514943					
F12	ENST00000510358					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
GRIN2B	ENST00000609686	ILMN_1884244	chr12	blood	2.71×10-7 (p-value)	Westra et al.	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000046906		F12

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001297853 (promoter flanking region)	2	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (GM12878)	PolII, H2AZ, H3K4me1, DNase1, H3K27ac
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
F12	upstream gene variant	4550	ENST00000503736	?	?	1
F12	upstream gene variant	3313	ENST00000253496	NM_000505.3	ENSP00000253496	3

F12	upstream gene variant	3313	ENST00000253496	NM_000505.3	ENSP00000253496	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GRK6	ENST00000502598	?	ENSP00000422873	5
GRK6	ENST00000506296	?	ENSP00000421055	5

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
F12	ENST00000253496	NM_000505.3	ENSP00000253496	1

Locus 114 (rs1203119)

SNiPAcard

Block annotations

Block info

genomic range	chr4:3,406,952-3,406,952
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.17	gene(s) hit or close-by	RGS12
phastCons	0	eQTL gene(s)	–
GERP++	-1.39	potentially regulated gene(s)	–
CADD score	1.776	disease gene(s)	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001428857 (open chromatin region)	1	endothelium (HUVEC)	H3K36me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RGS12	ENST00000514268	?	ENSP00000424251	1
RGS12	ENST00000504194	?	?	1
RGS12	ENST00000506631	?	?	1
RGS12	ENST00000508158	?	?	1
RGS12	ENST00000344733	NM_198229.2	ENSP00000339381	1
RGS12	ENST00000336727	?	ENSP00000338509	1
RGS12	ENST00000513784	?	?	1
RGS12	ENST00000505570	?	?	1
RGS12	ENST00000506998	?	?	1
RGS12	ENST00000382788	NM_002926.3	ENSP00000372238	1
RGS12	ENST00000338806	NM_198227.1	ENSP00000342133	1
RGS12	ENST00000512990	?	?	1
RGS12	ENST00000503041	?	?	1

Locus 115 (rs12146727)

SNiPAcard

Block annotations

Block info

genomic range	chr12:7,166,770-7,181,948
block size	15,179 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.153 [-3.322 – 3.781]	gene(s) hit or close-by	C1S
phastCons	μ = 0.061 [0 – 1]	eQTL gene(s)	C1RL, C1S
GERP++	μ = -0.675 [-8.63 – 5.15]	potentially regulated gene(s)	C1S, C1S
CADD score	μ = 4.078 [0.316 – 24.8]	disease gene(s)	C1S

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C1S	COMPLEMENT COMPONENT C1s DEFICIENCY	OMIM	MIM:613783
C1S	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
C1S	missense variant	ENST00000328916	NM_201442.2	ENSP00000328173	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000406697	?	ENSP00000385035	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000403949	?	ENSP00000384464	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000443875	?	ENSP00000397921	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000360817	NM_001734.3	ENSP00000354057	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000617865	?	ENSP00000484657	H/R	cAt/cGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C1S	ENST00000328916	ILMN_1677603	skin	2.11×10-13 (p-value)	MuTHER consortium	14
C1S	ENST00000488701		adipocyte	1.35×10-7 (p-value)	MuTHER consortium	14
C1S	ENST00000403949					
C1S	ENST00000413211					
C1S	ENST00000443875					
C1S	ENST00000402681					
C1S	ENST00000617865					
C1RL	ENST00000504702	ILMN_1733288	blood	6.73×10-7 (p-value)	Westra et al.	2

C1RL	ENST00000266542

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000129238	1	ENST00000328916, ENST00000360817, ENST00000443875, ENST00000406697, ENST00000402681	C1S

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000015893		C1S
ENCP00000015894		C1S

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001461176 (promoter flanking region)	2	NHLF	H3K36me3
		endothelium (HUVEC)	H3K27me3
		HSMMtube	DNase1
		lung (IMR90)	H3K36me3
		blood (DND-41)	H3K27me3
		skin (NHEK)	H3K4me1
		muscle (HSMM)	H2AZ, H3K27ac, DNase1
ENSR00001609158 (CTCF binding site)	2	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		nervous (NH-A)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C1S	upstream gene variant, downstream gene variant	602	ENST00000461983	?	?	13
C1S	upstream gene variant, downstream gene variant	226	ENST00000542978	?	ENSP00000442298	7
C1S	upstream gene variant, downstream gene variant	47	ENST00000413211	?	ENSP00000406643	6
C1S	upstream gene variant, downstream gene variant	464	ENST00000360817	NM_001734.3	ENSP00000354057	7
C1S	upstream gene variant, downstream gene variant	38	ENST00000497061	?	?	12
C1S	upstream gene variant, downstream gene variant	144	ENST00000463798	?	?	8
C1S	upstream gene variant, downstream gene variant	447	ENST00000402681	?	ENSP00000384171	7
C1S	upstream gene variant, downstream gene variant	377	ENST00000443875	?	ENSP00000397921	7
C1S	upstream gene variant, downstream gene variant	460	ENST00000473545	?	?	9
C1S	upstream gene variant, downstream gene variant	877	ENST00000495053	?	?	7
C1S	upstream gene variant, downstream gene variant	375	ENST00000328916	NM_201442.2	ENSP00000328173	7
C1S	upstream gene variant, downstream gene variant	54	ENST00000423384	?	ENSP00000399892	6
C1S	upstream gene variant, downstream gene variant	420	ENST00000541647	?	?	5
C1S	upstream gene variant, downstream gene variant	129	ENST00000489541	?	?	7
C1S	upstream gene variant, downstream gene variant	378	ENST00000617865	?	ENSP00000484657	7
C1S	upstream gene variant, downstream gene variant	224	ENST00000543187	?	?	6
C1S	upstream gene variant, downstream gene variant	475	ENST00000495061	?	?	13
C1S	upstream gene variant, downstream gene variant	224	ENST00000488701	?	?	6
C1S	upstream gene variant, downstream gene variant	594	ENST00000470326	?	?	13

C1S	upstream gene variant, downstream gene variant	400	ENST00000403949	?	ENSP00000384464	5
C1S	downstream gene variant	375	ENST00000406697	?	ENSP00000385035	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1S	ENST00000463798	?	?	2
C1S	ENST00000402681	?	ENSP00000384171	9
C1S	ENST00000328916	NM_201442.2	ENSP00000328173	8
C1S	ENST00000406697	?	ENSP00000385035	9
C1S	ENST00000360817	NM_001734.3	ENSP00000354057	8
C1S	ENST00000542978	?	ENSP00000442298	5
C1S	ENST00000443875	?	ENSP00000397921	8
C1S	ENST00000461983	?	?	3
C1S	ENST00000423384	?	ENSP00000399892	1
C1S	ENST00000541647	?	?	5
C1S	ENST00000543187	?	?	1
C1S	ENST00000617865	?	ENSP00000484657	8
C1S	ENST00000488701	?	?	1
C1S	ENST00000495053	?	?	4
C1S	ENST00000403949	?	ENSP00000384464	3
C1S	ENST00000473545	?	?	1
C1S	ENST00000413211	?	ENSP00000406643	1
C1S	ENST00000495061	?	?	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1S	ENST00000402681	?	ENSP00000384171	1
C1S	ENST00000328916	NM_201442.2	ENSP00000328173	1
C1S	ENST00000406697	?	ENSP00000385035	1
C1S	ENST00000360817	NM_001734.3	ENSP00000354057	1
C1S	ENST00000443875	?	ENSP00000397921	1
C1S	ENST00000617865	?	ENSP00000484657	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1S	ENST00000402681	?	ENSP00000384171	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C1S	ENST00000463798	?	1
C1S	ENST00000541647	?	3
C1S	ENST00000495053	?	2

Locus 115 (sentinel SNP rs12146727 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 116 (rs2842126)

SNiPAcard

Block annotations

Block info

genomic range	chr10:52,017,680-52,029,901
block size	12,222 bp
variant count	22 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.701 [-3.248 – 2.176]	gene(s) hit or close-by	DYNC1I2P1
phastCons	μ = 0.113 [0 – 1]	eQTL gene(s)	FAM21A
GERP++	μ = -0.161 [-7.14 – 1.41]	potentially regulated gene(s)	–
CADD score	μ = 3.195 [0.039 – 11.92]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FAM21A	?	ENSG00000099290	muscularis mucosae	4.66×10^{-6} (p-value)	GTEx Portal V6	14
FAM21A	?	ENSG00000099290	lung	9.80×10^{-11} (p-value)	GTEx Portal V6	18
FAM21A	?	ENSG00000099290	atrial appendage	1.18×10^{-8} (p-value)	GTEx Portal V6	17
FAM21A	?	ENSG00000099290	transformed fibroblasts	2.28×10^{-9} (p-value)	GTEx Portal V6	16
FAM21A	?	ENSG00000099290	breast	2.34×10^{-9} (p-value)	GTEx Portal V6	19
FAM21A	?	ENSG00000099290	tibial artery	1.53×10^{-14} (p-value)	GTEx Portal V6	19
FAM21A	?	ENSG00000099290	thyroid	1.61×10^{-12} (p-value)	GTEx Portal V6	19
FAM21A	?	ENSG00000099290	skeletal muscle	3.93×10^{-19} (p-value)	GTEx Portal V6	19
FAM21A	?	ENSG00000099290	liver	1.87×10^{-9} (p-value)	GTEx Portal V6	17
FAM21A	?	ENSG00000099290	sun exposed skin	1.22×10^{-15} (p-value)	GTEx Portal V6	19
FAM21A	?	ENSG00000099290	unexposed skin	2.21×10^{-8} (p-value)	GTEx Portal V6	17
FAM21A	?	ENSG00000099290	left ventricle	6.73×10^{-8} (p-value)	GTEx Portal V6	16
FAM21A	?	ENSG00000099290	subcutaneous adipocytes	2.62×10^{-15} (p-value)	GTEx Portal V6	19
FAM21A	?	ENSG00000099290	visceral adipocytes	5.71×10^{-13} (p-value)	GTEx Portal V6	19
FAM21A	?	ENSG00000099290	sigmoid colon	3.70×10^{-7} (p-value)	GTEx Portal V6	17
FAM21A	?	ENSG00000099290	tibial nerve	4.95×10^{-10} (p-value)	GTEx Portal V6	18
FAM21A	?	ENSG00000099290	esophagus mucosa	5.48×10^{-12} (p-value)	GTEx Portal V6	19
FAM21A	?	ENSG00000099290	testis	7.29×10^{-9} (p-value)	GTEx Portal V6	17
FAM21A	?	ENSG00000099290	putamen	1.59×10^{-6} (p-value)	GTEx Portal V6	6
FAM21A	?	ENSG00000099290	gastroesophageal junction	7.79×10^{-7} (p-value)	GTEx Portal V6	8
FAM21A	?	ENSG00000099290	hippocampus	2.59×10^{-6} (p-value)	GTEx Portal V6	4

FAM21A	?	ENSG00000099290	frontal cortex	4.18×10^{-6} (p-value)	GTEx Portal V6	1
FAM21A	?	ENSG00000099290	anterior cingulate cortex	1.16×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001423388 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		lung (IMR90)	H3K36me3, H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DYNC1I2P1	upstream gene variant, downstream gene variant	77	ENST00000435916	?	?	16

Putative effect on transcript

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
DYNC1I2P1	ENST00000435916	?	3

Locus 117 (rs9534288)

SNiPAcard

Block annotations

Block info

genomic range	chr13:46,569,846-46,636,652
block size	66,807 bp
variant count	70 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.506 [-3.814 – 2.895]	**gene(s) hit or close-by**	CPB2, CPB2-AS1, ZC3H13
phastCons	μ = 0.033 [0 – 0.978]	**eQTL gene(s)**	CPB2-AS1, LCP1
GERP++	μ = -0.155 [-5.96 – 3.84]	**potentially regulated gene(s)**	LCP1, LRRC63
CADD score	μ = 3.899 [0.032 – 14.11]	**disease gene(s)**	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR080759	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LCP1	ENST00000323076	ILMN_1662932	blood	6.46×10-6 (p-value)	Westra et al.	23
LCP1	ENST00000398576					
CPB2-AS1	?	ENSG00000235903	EBV lymphocytes	4.98×10-6 (p-value)	GTEx Portal V6	13

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000156965	1	ENCP00000018501	LCP1
ENCE00000156967	1	ENCP00000018501	LCP1
		ENCP00000018503	LRRC63 LCP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000515658 (promoter)	1	embryonic stem cell (H1ESC)	CTCF, PolII, Gabp, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K9ac, H3K79me2, H3K27ac, DNase1, H2AZ, H3K4me3
		blood (K562)	H3K4me3, H3K4me2, PolII, TAF1, H2AZ, H3K79me2, Gabp, H3K9ac, CTCF, Max, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K9ac, DNase1, H3K4me2, H3K4me3, H2AZ, H3K79me2
		liver (HepG2)	H3K27me3, DNase1, H3K36me3, Gabp, PolII, H3K79me2, ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3
		lung (IMR90)	H3K4me2, H3K4me3, H3K9ac, DNase1
		blood (GM12878)	DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H2AZ, PolII, CTCF, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac

		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K9ac, H3K4me3, DNase1, H3K27ac
		Osteobl	H3K4me3, H3K4me2, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac, DNase1, PolII, Gabp, H3K79me2
		monocytes (Monocytes-CD14+)	H3K9ac, H3K27me3, H3K4me3, H3K27ac, H3K4me2, DNase1
		endothelium (HUVEC)	Max, H3K4me2, H3K4me3, H3K9ac, PolII, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000515659 (promoter flanking region)	3	liver (HepG2)	H3K36me3
		skin (NHDF-AD)	DNase1
		skin (NHEK)	DNase1
		breast (HMEC)	H3K27ac
ENSR00001509497 (promoter flanking region)	2	liver (HepG2)	H3K36me3
		Osteobl	H3K27ac, H3K4me2
		A549	H3K4me3
		skin (NHEK)	H3K4me2
ENSR00001618259 (CTCF binding site)	1	liver (HepG2)	H3K36me3
		Osteobl	H3K27ac, H3K4me2
		A549	H3K4me3
		skin (NHEK)	H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CPB2	downstream gene variant	1735	ENST00000181383	NM_001872.4	ENSP00000181383	5
CPB2	downstream gene variant	1736	ENST00000439329	NM_001278541.1	ENSP00000400714	5
CPB2-AS1	upstream gene variant	1397	ENST00000606243	?	?	6
CPB2-AS1	upstream gene variant	1451	ENST00000415033	?	?	6
CPB2-AS1	upstream gene variant	1397	ENST00000606991	?	?	6
CPB2-AS1	upstream gene variant	1355	ENST00000606351	?	?	6
ZC3H13	upstream gene variant	599	ENST00000242848	?	ENSP00000242848	4
ZC3H13	downstream gene variant, upstream gene variant	981	ENST00000470308	?	?	11
ZC3H13	downstream gene variant, upstream gene variant	594	ENST00000464597	?	ENSP00000480658	10
ZC3H13	downstream gene variant, upstream gene variant	660	ENST00000428921	?	ENSP00000416238	7
ZC3H13	upstream gene variant	586	ENST00000282007	NM_015070.4	ENSP00000282007	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	15
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	15
CPB2-AS1	ENST00000606351	?	?	15
CPB2-AS1	ENST00000606243	?	?	15
CPB2-AS1	ENST00000606991	?	?	15
CPB2-AS1	ENST00000415033	?	?	15
ZC3H13	ENST00000464597	?	ENSP00000480658	7
ZC3H13	ENST00000470308	?	?	10
ZC3H13	ENST00000428921	?	ENSP00000416238	14
ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	55

ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	55
ZC3H13	ENST00000242848	?	ENSP00000242848	55

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	1
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	1

Locus 118 (rs12134133)

SNiPAcard

Block annotations

Block info

genomic range	chr1:207,411,912-207,529,576
block size	117,665 bp
variant count	79 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.314 [-2.969 – 1.497]	gene(s) hit or close-by	CD55, RP11-6J21.2
phastCons	μ = 0.070 [0 – 0.985]	eQTL gene(s)	CD55, CR1
GERP++	μ = -0.380 [-6.12 – 3.97]	potentially regulated gene(s)	C4BPAP2, C4BPB, IL20, PIGR, RP11-164O23.5
CADD score	μ = 3.646 [0.01 – 11.79]	disease gene(s)	CD55

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
threonine	2.20×10^{-5}	Metabolomics GWAS Server	24816252	8
4-acetaminophen sulfate	8.26×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
CD55	BLOOD GROUP, CROMER SYSTEM	OMIM	MIM:613793

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD55	?	ENSG00000196352	adrenal gland	3.70×10^{-8} (p-value)	GTEx Portal V6	78
CD55	?	ENSG00000196352	tibial artery	1.85×10^{-8} (p-value)	GTEx Portal V6	78
CD55	?	ENSG00000196352	transformed fibroblasts	6.82×10^{-6} (p-value)	GTEx Portal V6	2
CR1	ENST00000367051	ILMN_1767193	blood	2.76×10^{-5} (p-value)	MuTHER consortium	16
CR1	ENST00000367052					
CR1	ENST00000400960					
CR1	ENST00000367049					
CR1	ENST00000529814					
CR1	ENST00000367053					
?	?	ILMN_1742601	blood	3.62×10^{-5} (p-value)	MuTHER consortium	9

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000053173	1	ENCP00000006362	PIGR
		ENCP00000006355	IL20

ENCE00000053309	1	ENCP00000006375	RP11-164O23.5
		ENCP00000006373	C4BPB
		ENCP00000006371	C4BPB
ENCE00000053321	1	ENCP00000006373	C4BPB
ENCE00000053363	1	ENCP00000006378	C4BPAP2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001590164 (enhancer)	1	cervix (HeLa-S3)	DNase1, H3K27ac
		HSMMtube	H3K27me3
		skin (NHEK)	H3K4me1
ENSR00000551826 (promoter flanking region)	10	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Cjun, PolII, Jund, H3K4me2, H3K27ac, Max, Cmyc
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K4me1, FOXA1, H3K27ac, DNase1
		A549	H3K4me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K4me2
ENSR00000551828 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		blood (K562)	CTCF, Rad21
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF
ENSR00000551831 (promoter flanking region)	4	cervix (HeLa-S3)	DNase1
		embryonic stem cell (H1ESC)	DNase1
		endothelium (HUVEC)	Cjun, DNase1, H3K4me2, H3K4me1, H3K27ac
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
ENSR00001528072 (CTCF binding site)	1	Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00000551833 (enhancer)	1	cervix (HeLa-S3)	H3K36me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K4me3
ENSR00000288774 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H2AZ
ENSR00000288776 (promoter flanking region)	5	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	DNase1
		blood (K562)	Brg1, DNase1, H3K27ac, Max, Junb, PU1, MEF2A, Jund, Cmyc, H2AZ, Cjun
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H2AZ, DNase1
		liver (HepG2)	H3K4me2
		blood (GM12878)	H2AZ, PU1
		nervous (NH-A)	DNase1, H3K4me2
		skin (NHEK)	DNase1
		NHLF	DNase1
		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Cjun, PolII, H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, Max, Cfos
		monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1, DNase1

		endothelium (HUVEC)	Cjun, Max, DNase1, H3K4me2, H3K27ac, PolII
		A549	H3K4me3, H3K4me2, DNase1
ENSR00001590170 (CTCF binding site)	1	cervix (HeLa-S3)	DNase1, Jund, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		nervous (NH-A)	DNase1
		blood (K562)	Brg1, DNase1, Junb, MEF2A, Jund, Cjun
ENSR00000551839 (promoter)	2	embryonic stem cell (H1ESC)	PolII, Rad21, Jund, TAF7, Sin3Ak20, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, DNase1, H3K27me3, H3K36me3
		HSMMtube	H3K9ac, H3K4me2, H2AZ, H3K27me3, DNase1
		blood (K562)	H3K79me2, HEY1, H2AZ, TAF1, Cmyc, PolII, H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, H3K9ac, PU1, H3K27ac, Max
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	DNase1, H3K27me3, H3K4me3, PolII, TAF1, H2AZ, H3K4me2, H3K9ac, Cmyc
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	CTCF, H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K27me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K27me3, H3K9ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, PolII, Cmyc, CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, Cmyc, PolII, H3K27ac, Cjun, Max, H3K4me2, H3K4me3, H3K9ac
		A549	DNase1, H3K27ac, H3K36me3, H3K9ac, H3K4me2, H3K4me3
ENSR00001590172 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, PolII, Rad21, TAF7, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, DNase1, H3K9ac, H3K4me3
		HSMMtube	H3K9ac, H3K4me2, H2AZ
		blood (K562)	H3K27ac, Max, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, PolII, H3K4me2, H3K36me3, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, H3K36me3
		muscle (HSMM)	H3K4me2, H3K9ac, H3K36me3, H3K27ac, H2AZ, H3K4me3
		liver (HepG2)	H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3
		blood (GM12878)	PolII, H2AZ, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K27ac, H3K9ac, H3K4me3
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K9ac, H3K27me3
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K79me2, PolII, H3K36me3, H3K4me3, H3K27ac, H3K9ac, DNase1, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CD55	upstream gene variant, downstream gene variant	457	ENST00000343420	?	ENSP00000340631	2
CD55	upstream gene variant	174	ENST00000367064	NM_000574.3	ENSP00000356031	1
CD55	upstream gene variant	399	ENST00000367067	?	ENSP00000356034	1
CD55	upstream gene variant	1107	ENST00000465534	?	?	3
CD55	upstream gene variant, downstream gene variant	353	ENST00000488171	?	?	3
CD55	upstream gene variant	1107	ENST00000476590	?	?	3
CD55	upstream gene variant	309	ENST00000391921	?	ENSP00000375788	1
CD55	upstream gene variant	367	ENST00000314754	NM_001114752.1	ENSP00000316333	1
CD55	upstream gene variant, downstream gene variant	198	ENST00000367063	NM_001300902.1	ENSP00000356030	3

RP11-6J21.2	upstream gene variant, downstream gene variant	1110	ENST00000417084	?	?	10

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-6J21.2	ENST00000417084	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CD55	ENST00000343420	?	ENSP00000340631	3
CD55	ENST00000314754	NM_001114752.1	ENSP00000316333	10
CD55	ENST00000367064	NM_000574.3	ENSP00000356031	10
CD55	ENST00000367067	?	ENSP00000356034	10
CD55	ENST00000367063	NM_001300902.1	ENSP00000356030	3
CD55	ENST00000488171	?	?	3
CD55	ENST00000391921	?	ENSP00000375788	10
CD55	ENST00000465534	?	?	8
CD55	ENST00000476590	?	?	8
RP11-6J21.2	ENST00000417084	?	?	48

Locus 119 (rs556990)

SNiPAcard

Block annotations

Block info

genomic range	chr13:113,818,708-113,910,611
block size	91,904 bp
variant count	79 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.775 [-6.801 – 1.266]	gene(s) hit or close-by	CUL4A, PCID2, PROZ, RP11-98F14.11
phastCons	μ = 0.029 [0 – 0.987]	eQTL gene(s)	ADAMDEC1, CUL4A, F10, PCID2, PROZ, RP11-98F14.11
GERP++	μ = -1.100 [-8.08 – 2.42]	potentially regulated gene(s)	ADPRHL1, ATP11A, ATP4B, CUL4A, DCUN1D2, F7, GRTP1, MCF2L, PCID2, PROZ, TFDP1, TMCO3
CADD score	μ = 3.026 [0.009 – 15.28]	disease gene(s)	PROZ, ATP11A, F10, F7

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CS040554	1

Disease gene annotation

gene	trait	source DB	source entry/link	
PROZ	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217	orphanet
ATP11A	Idiopathic pulmonary fibrosis	OrphaNet	OrphaNet:2032	orphanet
F10	Congenital factor X deficiency	OrphaNet	OrphaNet:328	orphanet
F7	Congenital factor VII deficiency	OrphaNet	OrphaNet:327	orphanet
F10	FACTOR X DEFICIENCY	OMIM	MIM:227600	OMIM
F7	FACTOR VII DEFICIENCY	OMIM	MIM:227500	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PROZ	?	ENSG00000126231	pancreas	1.30×10^{-16} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	muscularis mucosae	1.65×10^{-7} (p-value)	GTEx Portal V6	69
RP11-98F14.11	?	ENSG00000269125	muscularis mucosae	6.60×10^{-6} (p-value)	GTEx Portal V6	1
PROZ	?	ENSG00000126231	stomach	8.50×10^{-12} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	lung	1.04×10^{-15} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	tibial nerve	1.90×10^{-12} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	transformed fibroblasts	2.61×10^{-7} (p-value)	GTEx Portal V6	64
PROZ	?	ENSG00000126231	blood	4.04×10^{-14} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	tibial artery	1.42×10^{-14} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	thyroid	1.85×10^{-16} (p-value)	GTEx Portal V6	78

RP11-98F14.11	?	ENSG00000269125	thyroid	1.43×10-8 (p-value)	GTEx Portal V6	51
PROZ	?	ENSG00000126231	cerebellar hemisphere	1.80×10-6 (p-value)	GTEx Portal V6	3
PROZ	?	ENSG00000126231	adrenal gland	8.50×10-8 (p-value)	GTEx Portal V6	33
PCID2	?	ENSG00000126226	testis	7.95×10-12 (p-value)	GTEx Portal V6	77
PROZ	?	ENSG00000126231	testis	2.75×10-13 (p-value)	GTEx Portal V6	75
RP11-98F14.11	?	ENSG00000269125	testis	3.71×10-7 (p-value)	GTEx Portal V6	19
PROZ	?	ENSG00000126231	cerebellum	8.07×10-9 (p-value)	GTEx Portal V6	78
RP11-98F14.11	?	ENSG00000269125	cerebellum	2.38×10-6 (p-value)	GTEx Portal V6	12
PROZ	?	ENSG00000126231	transverse colon	1.99×10-11 (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	subcutaneous adipocytes	2.56×10-8 (p-value)	GTEx Portal V6	51
PCID2	ENST00000375477	ILMN_1788024	blood	1.85×10-7 (p-value)	MuTHER consortium	30
PCID2	ENST00000375459					
PCID2	ENST00000462653					
PCID2	ENST00000463102					
PCID2	ENST00000473462					
PCID2	ENST00000493650					
PCID2	ENST00000375479					
PCID2	ENST00000622406					
PCID2	ENST00000337344					
PCID2	ENST00000246505					
PCID2	ENST00000375457					
PROZ	?	ENSG00000126231	spleen	1.52×10-9 (p-value)	GTEx Portal V6	47
PROZ	?	ENSG00000126231	esophagus mucosa	4.90×10-8 (p-value)	GTEx Portal V6	76
PROZ	?	ENSG00000126231	atrial appendage	7.13×10-7 (p-value)	GTEx Portal V6	40
PROZ	?	ENSG00000126231	breast	2.74×10-6 (p-value)	GTEx Portal V6	13
PROZ	?	ENSG00000126231	prostate	1.94×10-7 (p-value)	GTEx Portal V6	15
PCID2	ENST00000375477	ILMN_1788024	blood	1.36×10-5 (p-value)	Westra et al.	1
PCID2	ENST00000375459					
PCID2	ENST00000462653					
PCID2	ENST00000463102					
PCID2	ENST00000473462					
PCID2	ENST00000493650					
PCID2	ENST00000375479					
PCID2	ENST00000622406					
PCID2	ENST00000337344					
PCID2	ENST00000246505					
PCID2	ENST00000375457					
F10	?	ENSG00000126218	left ventricle	1.10×10-7 (p-value)	GTEx Portal V6	27
PROZ	?	ENSG00000126231	sigmoid colon	2.45×10-6 (p-value)	GTEx Portal V6	10
CUL4A	?	ENSG00000139842	skeletal muscle	5.73×10-6 (p-value)	GTEx Portal V6	11

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ADAMDEC1	?	ENSG00000134028	chr8	blood	1.07×10^{-2} (q-value)	SeeQTL DB (HapMap)	3

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000019252		
ENCP00000019255		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000165802	1	ENCP00000019270	ADPRHL1
ENCE00000165363	1	ENCP00000019222	ATP11A
		ENCP00000019281	TMCO3
		ENCP00000019218	ATP11A
ENCE00000165706	1	ENCP00000019286	ATP4B
		ENCP00000019258	CUL4A PCID2
ENCE00000165538	1	ENCP00000019231	MCF2L
ENCE00000165742	1	ENCP00000019261	CUL4A
		ENCP00000019279	TMCO3
		ENCP00000019278	TMCO3
ENCE00000165366	1	ENCP00000019222	ATP11A
		ENCP00000019281	TMCO3
		ENCP00000019218	ATP11A
ENCE00000165845	1	ENCP00000019276	DCUN1D2
ENCE00000165690	1	ENCP00000019252	PROZ
ENCE00000165334	1	ENCP00000019241	MCF2L
		ENCP00000019243	MCF2L
		ENCP00000019284	TFDP1
		ENCP00000019230	MCF2L
		ENCP00000019268	GRTP1
		ENCP00000019220	ATP11A
		ENCP00000019286	ATP4B
		ENCP00000019224	MCF2L
		ENCP00000019274	DCUN1D2
		ENCP00000019233	MCF2L
		ENCP00000019223	ATP11A
		ENCP00000019245	F7
		ENCP00000019259	PCID2
		ENCP00000019217	ATP11A
		ENCP00000019251	PROZ

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000520923 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00000520925 (TF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	DNase1, Cjun
		lung (IMR90)	H3K36me3
ENSR00000520926	1	NHLF	H3K36me3

(enhancer)		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K4me1
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000520927 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H4K8ac, H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001036786 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K36me3, DNase1, H3K27ac
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K36me3
		lung (IMR90)	H4K91ac, H3K56ac, H2BK15ac, H3K23ac, H2BK12ac, H3K27ac, H3K18ac, H3K4me2, H4K5ac, H3K36me3, H3K4ac, H2BK120ac, H2AK5ac, H4K8ac, DNase1, H3K9ac
		blood (GM12878)	H3K36me3
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me2, H3K9ac
		skin (NHEK)	H3K36me3
		NHLF	H3K27ac, H3K9ac, H3K4me1, DNase1, H3K36me3
		Osteobl	H3K4me2, H3K36me3, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3, Jund, Cjun
		monocytes (Monocytes-CD14+)	H3K36me3, H3K27ac
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001512527 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K36me3
		breast (HMEC)	H3K36me3
		muscle (HSMM)	H3K36me3, DNase1
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000520931 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K36me3, DNase1
		blood (K562)	H3K36me3, Cjun, Junb, FOSL1
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H3K79me2, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	FOSL2, DNase1, H3K27ac, H3K36me3
		blood (GM12878)	H3K27ac, H3K36me3, DNase1, H3K4me1, Cmyc
		lung (IMR90)	DNase1, H4K8ac, H2BK12ac, H3K27ac, H3K18ac, H3K36me3, H3K4ac, H2BK120ac, H2BK20ac, H4K91ac
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me2, H3K9ac
		skin (NHEK)	H3K36me3, CTCF, DNase1
		NHLF	H3K36me3, H3K27ac, DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3, H3K27ac, DNase1
		cervix (HeLa-S3)	Cjun, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me1, TAF1, PolII, H3K79me2, Gabp, Cfos, CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3, DNase1

		endothelium (HUVEC)	Cmyc, DNase1, PolII, H3K27ac, Max, Cjun, H3K36me3
		A549	DNase1, H3K4me3, H3K36me3
ENSR00001512530 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K36me3
		breast (HMEC)	H3K36me3
		muscle (HSMM)	DNase1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001512532 (enhancer)	2	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (K562)	H3K36me3, H3K79me2
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K79me2, H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K79me2, H3K4me1, H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K79me2, H3K36me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001512533 (promoter)	1	embryonic stem cell (H1ESC)	H3K27ac, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, SIX5, Sin3Ak20, p300, H3K36me3, CTCF, PolII, Egr1, Gabp, Jund, TAF7, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K36me3, DNase1
		blood (K562)	H3K9ac, CTCF, ZBTB7A, Max, H3K27ac, Gabp, USF1, H3K4me3, DNase1, H3K36me3, H3K4me2, PolII, TAF1, HEY1, H3K79me2, Cmyc
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K27ac, H3K4me2, H3K9ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K4me3, H3K36me3, Cmyc, CTCF, H3K27ac, Gabp, Yy1, PolII, H3K79me2, TAF1, USF1, H3K4me1, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K79me2, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	H3K4me2, H3K9ac, H3K36me3, H3K27ac, H3K4me3, H3K79me2, PolII, BCLAF1, Srf, H2AZ, Cmyc, Yy1, Gabp, ELF1, SIX5, TAF1, DNase1
		nervous (NH-A)	DNase1, H3K4me2, H3K9ac, H3K4me3, H3K27ac
		skin (NHEK)	H3K9ac, H3K27ac, H3K4me1, CTCF, H3K4me2, H3K4me3, DNase1, H3K36me3
		NHLF	H3K36me3, H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me2, H3K36me3, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me2, H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K36me3, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K36me3, PolII, Gabp, H3K79me2, Ini1, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, Cmyc
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H4K20me1, H3K9ac, H3K27ac, DNase1
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, PolII, H3K27ac, H3K9ac, H3K4me2, H3K4me3, Max
		A549	H3K4me3, H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2
ENSR00000275938 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, H3K4me3, H3K4me2, DNase1
		HSMMtube	H3K79me2, DNase1
		blood (K562)	H3K79me2, Max, H3K27ac, DNase1, H3K36me3, H3K4me1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K79me2, DNase1
		liver (HepG2)	H4K20me1, H3K79me2, H3K4me1, H3K36me3
		blood (GM12878)	H3K79me2
		lung (IMR90)	DNase1, H3K79me2, H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1
		NHLF	H3K4me1, DNase1
		Osteobl	H3K4me2, H4K20me1
		blood (DND-41)	H3K36me3, H4K20me1
		cervix (HeLa-S3)	H3K79me2, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3

ENSR00000057151 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, H3K36me3, CTCF
		HSMMtube	H3K36me3
		blood (K562)	CTCF
		muscle (HSMM)	H3K36me3
		liver (HepG2)	CTCF, H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3, CTCF
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF, H3K36me3
		cervix (HeLa-S3)	H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CUL4A	upstream gene variant	73	ENST00000472083	?	?	1
CUL4A	upstream gene variant, downstream gene variant	228	ENST00000488558	?	ENSP00000480367	8
CUL4A	upstream gene variant	901	ENST00000375441	NM_001278513.1	ENSP00000364590	5
CUL4A	upstream gene variant, downstream gene variant	1522	ENST00000470067	?	?	3
CUL4A	upstream gene variant, downstream gene variant	15	ENST00000498562	?	?	8
CUL4A	upstream gene variant, downstream gene variant	410	ENST00000463426	?	?	7
CUL4A	upstream gene variant	1480	ENST00000451881	NM_003589.2	ENSP00000389118	5
CUL4A	upstream gene variant	1544	ENST00000617546	?	ENSP00000481782	5
CUL4A	upstream gene variant, downstream gene variant	233	ENST00000494985	?	?	9
CUL4A	upstream gene variant, downstream gene variant	2324	ENST00000474116	?	?	9
CUL4A	upstream gene variant	2252	ENST00000375440	NM_001008895.2	ENSP00000364589	5
CUL4A	upstream gene variant	1402	ENST00000326335	NM_001278514.1	ENSP00000322132	5
PCID2	downstream gene variant, upstream gene variant	339	ENST00000337344	NM_001127202.2	ENSP00000337405	12
PCID2	downstream gene variant, upstream gene variant	976	ENST00000475433	?	?	10
PCID2	downstream gene variant, upstream gene variant	373	ENST00000375479	NM_018386.3, NM_001127203.2	ENSP00000364628	12
PCID2	downstream gene variant, upstream gene variant	374	ENST00000493650	?	?	13
PCID2	downstream gene variant, upstream gene variant	1590	ENST00000491548	?	?	4
PCID2	downstream gene variant, upstream gene variant	1873	ENST00000484641	?	?	8
PCID2	downstream gene variant, upstream gene variant	301	ENST00000622406	NM_001258212.1	ENSP00000479494	12
PCID2	downstream gene variant, upstream gene variant	579	ENST00000462853	?	?	4
PCID2	downstream gene variant, upstream gene variant	374	ENST00000463102	?	?	13
PCID2	downstream gene variant, upstream gene variant	375	ENST00000462653	?	?	13
PCID2	downstream gene variant, upstream gene variant	1034	ENST00000473462	?	?	12
PCID2	downstream gene variant, upstream gene variant	374	ENST00000375459	?	ENSP00000364608	12
PCID2	downstream gene variant, upstream gene variant	373	ENST00000375477	?	ENSP00000364626	12

PCID2	downstream gene variant, upstream gene variant	374	ENST00000246505	?	ENSP00000246505	12
PCID2	downstream gene variant, upstream gene variant	379	ENST00000375457	NM_001258213.1	ENSP00000364606	12
PCID2	downstream gene variant, upstream gene variant	1035	ENST00000480971	?	?	10
PROZ	downstream gene variant	381	ENST00000342783	NM_001256134.1	ENSP00000344458	10
PROZ	upstream gene variant, downstream gene variant	125	ENST00000493630	?	?	7
PROZ	downstream gene variant	378	ENST00000375547	NM_003891.2	ENSP00000364697	10
RP11-98F14.11	downstream gene variant, upstream gene variant	16	ENST00000600642	?	?	17

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
CUL4A	ENST00000470067	?	?	1
CUL4A	ENST00000472083	?	?	1
CUL4A	ENST00000375441	NM_001278513.1	ENSP00000364590	1
CUL4A	ENST00000326335	NM_001278514.1	ENSP00000322132	1
CUL4A	ENST00000375440	NM_001008895.2	ENSP00000364589	1
CUL4A	ENST00000617546	?	ENSP00000481782	1
CUL4A	ENST00000451881	NM_003589.2	ENSP00000389118	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CUL4A	ENST00000470067	?	?	6
CUL4A	ENST00000472083	?	?	6
CUL4A	ENST00000375441	NM_001278513.1	ENSP00000364590	30
CUL4A	ENST00000326335	NM_001278514.1	ENSP00000322132	30
CUL4A	ENST00000498562	?	?	19
CUL4A	ENST00000375440	NM_001008895.2	ENSP00000364589	30
CUL4A	ENST00000463426	?	?	17
CUL4A	ENST00000617546	?	ENSP00000481782	30
CUL4A	ENST00000474116	?	?	4
CUL4A	ENST00000494985	?	?	15
CUL4A	ENST00000451881	NM_003589.2	ENSP00000389118	30
CUL4A	ENST00000488558	?	ENSP00000480367	17
PCID2	ENST00000491548	?	?	12
PCID2	ENST00000246505	?	ENSP00000246505	19
PCID2	ENST00000337344	NM_001127202.2	ENSP00000337405	19
PCID2	ENST00000493650	?	?	6
PCID2	ENST00000462853	?	?	3
PCID2	ENST00000622406	NM_001258212.1	ENSP00000479494	19
PCID2	ENST00000375479	NM_018386.3, NM_001127203.2	ENSP00000364628	19
PCID2	ENST00000473462	?	?	6

PCID2	ENST00000375459	?	ENSP00000364608	19
PCID2	ENST00000375477	?	ENSP00000364626	19
PCID2	ENST00000484641	?	?	17
PCID2	ENST00000375457	NM_001258213.1	ENSP00000364606	19
PCID2	ENST00000480971	?	?	17
PCID2	ENST00000475433	?	?	17
PROZ	ENST00000375547	NM_003891.2	ENSP00000364697	19
PROZ	ENST00000493630	?	?	16
PROZ	ENST00000342783	NM_001256134.1	ENSP00000344458	19

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PCID2	ENST00000491548	?	1
PCID2	ENST00000493650	?	1

Locus 120 (rs149313)

SNiPAcard

Block annotations

Block info

genomic range	chr5:96,057,882-96,070,205
block size	12,324 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.582 [-3.14 – 0.626]	**gene(s) hit or close-by**	CAST
phastCons	μ = 0.011 [0 – 0.051]	**eQTL gene(s)**	CAST, CTC-506B8.1, CTD-2260A17.1, ERAP1
GERP++	μ = 0.491 [0.225 – 1.08]	**potentially regulated gene(s)**	–
CADD score	μ = 4.473 [2.207 – 8.12]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP1	?	ENSG00000164307	pancreas	1.38×10-7 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	muscularis mucosae	2.46×10-9 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	muscularis mucosae	2.49×10-10 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	lung	1.19×10-7 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	lung	6.30×10-8 (p-value)	GTEx Portal V6	3
CAST	?	ENSG00000153113	lung	1.78×10-10 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	tibial nerve	1.85×10-5 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	tibial nerve	8.33×10-9 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	tibial nerve	7.19×10-11 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	transformed fibroblasts	6.20×10-7 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	transformed fibroblasts	1.87×10-8 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	transformed fibroblasts	1.87×10-10 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	blood	6.95×10-15 (p-value)	GTEx Portal V6	5
CTC-506B8.1	?	ENSG00000249180	blood	6.38×10-10 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	blood	2.32×10-12 (p-value)	GTEx Portal V6	5
CTC-506B8.1	?	ENSG00000249180	tibial artery	2.72×10-10 (p-value)	GTEx Portal V6	5
CAST	?	ENSG00000153113	tibial artery	4.85×10-8 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	esophagus mucosa	9.80×10-7 (p-value)	GTEx Portal V6	2
CTC-506B8.1	?	ENSG00000249180	thyroid	5.78×10-6 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	thyroid	4.83×10-7 (p-value)	GTEx Portal V6	2
ERAP1	?	ENSG00000164307	skeletal muscle	6.41×10-15 (p-value)	GTEx Portal V6	5
CTD-2260A17.1	?	ENSG00000248734	skeletal muscle	1.45×10-7 (p-value)	GTEx Portal V6	5

ERAP1	?	ENSG00000164307	EBV lymphocytes	2.27×10-6 (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	transverse colon	2.42×10-6 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	aorta	1.98×10-6 (p-value)	GTEx Portal V6	5
ERAP1	?	ENSG00000164307	subcutaneous adipocytes	2.15×10-6 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	subcutaneous adipocytes	1.11×10-7 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	subcutaneous adipocytes	9.10×10-7 (p-value)	GTEx Portal V6	1
CTD-2260A17.1	?	ENSG00000248734	testis	5.12×10-7 (p-value)	GTEx Portal V6	4
CAST	?	ENSG00000153113	adrenal gland	3.31×10-6 (p-value)	GTEx Portal V6	1
ERAP1	ENST00000296754	ILMN_1752145	blood	4.72×10-9 (p-value)	MuTHER consortium	1
ERAP1	ENST00000443439	ILMN_2336220	monocyte	3.15×10-23 (p-value)	Zeller et al.	1
ERAP1	ENST00000514604					
ERAP1	ENST00000296754					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001285973 (promoter flanking region)	1	NHLF	DNase1, H3K36me3
		embryonic stem cell (H1ESC)	SP1, DNase1
		blood (K562)	DNase1
		skin (NHDF-AD)	H3K36me3, DNase1
		breast (HMEC)	H3K4me2, H3K27ac, DNase1
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		nervous (NH-A)	H3K36me3, DNase1
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K36me3, H3K4me2, H3K27ac, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CAST	upstream gene variant	3751	ENST00000348386	?	?	1
CAST	downstream gene variant	2214	ENST00000514845	?	ENSP00000422612	2
CAST	upstream gene variant, downstream gene variant	3360	ENST00000504532	?	?	3
CAST	downstream gene variant	3372	ENST00000506811	?	ENSP00000427092	2
CAST	upstream gene variant	1675	ENST00000515063	?	?	2
CAST	downstream gene variant	3338	ENST00000514055	?	ENSP00000421230	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CAST	ENST00000510156	?	ENSP00000422325	5
CAST	ENST00000395813	?	ENSP00000379158	5
CAST	ENST00000514055	?	ENSP00000421230	3

CAST	ENST00000514055	?	ENSP00000421230	3
CAST	ENST00000511049	?	ENSP00000421130	5
CAST	ENST00000309190	NM_001284212.1, NM_173060.3	ENSP00000312523	5
CAST	ENST00000421689	?	ENSP00000396558	5
CAST	ENST00000511097	?	ENSP00000422951	5
CAST	ENST00000506811	?	ENSP00000427092	3
CAST	ENST00000509903	?	ENSP00000426946	5
CAST	ENST00000512620	?	ENSP00000426422	5
CAST	ENST00000338252	NM_001190442.1	ENSP00000343421	5
CAST	ENST00000508117	?	ENSP00000422466	5
CAST	ENST00000508608	?	ENSP00000422677	5
CAST	ENST00000508830	?	ENSP00000425721	5
CAST	ENST00000511782	?	ENSP00000423638	5
CAST	ENST00000513666	?	?	5
CAST	ENST00000507487	?	?	3
CAST	ENST00000505143	?	ENSP00000422957	5
CAST	ENST00000395812	NM_001042440.3	ENSP00000379157	5
CAST	ENST00000514845	?	ENSP00000422612	3
CAST	ENST00000341926	?	ENSP00000339914	5
CAST	ENST00000508197	?	ENSP00000422831	5
CAST	ENST00000504465	NM_001284213.1	ENSP00000425670	5
CAST	ENST00000510756	?	ENSP00000422176	5
CAST	ENST00000348386	?	?	2

Locus 121 (rs4150579)

SNiPAcard

Block annotations

Block info

genomic range	chr11:18,309,700-18,392,983
block size	83,284 bp
variant count	48 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.408 [-3.606 – 2.283]	gene(s) hit or close-by	GTF2H1, HPS5
phastCons	μ = 0.075 [0 – 1]	eQTL gene(s)	GTF2H1, HPS5, LDHA, LDHC, SAA1
GERP++	μ = -0.080 [-4.3 – 3.95]	potentially regulated gene(s)	AC084117.3, GTF2H1, HPS5, LDHA, RP11-113D6.3, RP11-613F22.6, TSG101, UEVLD
CADD score	μ = 5.040 [0.089 – 14.37]	disease gene(s)	LDHA, HPS5, SAA1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
PANCREATIC CANCER	<9.00×10-6	GWAS Catalog	23180869	1
Amyloid A serum levels	<2.00×10-51	GWAS Catalog	21124955	1
alpha-hydroxyisovalerate	1.08×10-23	Metabolomics GWAS Server	24816252	25

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	benign	ClinVar	RCV000150827.1	1

Disease gene annotation

gene	trait	source DB	source entry/link
LDHA	GLYCOGEN STORAGE DISEASE XI	OMIM	MIM:612933
HPS5	HERMANSKY-PUDLAK SYNDROME 5	OMIM	MIM:614074
LDHA	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency	OrphaNet	OrphaNet:284426
SAA1	Secondary amyloidosis	OrphaNet	OrphaNet:85445
HPS5	Hermansky-Pudlak syndrome without pulmonary fibrosis	OrphaNet	OrphaNet:231512

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SAA1	ENST00000356524	ILMN_2304512	adipocyte	2.33×10-5 (p-value)	MuTHER consortium	25
SAA1	ENST00000405158					
SAA1	ENST00000532858					
GTF2H1	?	ENSG00000110768	muscularis mucosae	1.75×10-20 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	lung	8.65×10-14 (p-value)	GTEx Portal V6	44
HPS5	?	ENSG00000110756	lung	1.37×10-9 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	atrial appendage	7.22×10-9 (p-value)	GTEx Portal V6	44

GTF2H1	?	ENSG00000110768	transformed fibroblasts	2.71×10-28 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	breast	8.81×10-13 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	tibial artery	4.70×10-15 (p-value)	GTEx Portal V6	44
LDHC	?	ENSG00000166796	tibial artery	1.90×10-5 (p-value)	GTEx Portal V6	1
GTF2H1	?	ENSG00000110768	thyroid	7.29×10-14 (p-value)	GTEx Portal V6	44
LDHA	?	ENSG00000134333	skeletal muscle	1.73×10-8 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	skeletal muscle	2.31×10-7 (p-value)	GTEx Portal V6	40
GTF2H1	?	ENSG00000110768	EBV lymphocytes	5.82×10-7 (p-value)	GTEx Portal V6	25
GTF2H1	?	ENSG00000110768	transverse colon	1.75×10-14 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	liver	2.96×10-8 (p-value)	GTEx Portal V6	43
GTF2H1	?	ENSG00000110768	sun exposed skin	7.28×10-21 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	unexposed skin	7.19×10-20 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	aorta	6.66×10-16 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	left ventricle	1.11×10-9 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	subcutaneous adipocytes	2.66×10-27 (p-value)	GTEx Portal V6	44
GTF2H1	ENST00000534641	ILMN_1726693	monocyte	1.30×10-20 (p-value)	Zeller et al.	7
GTF2H1	ENST00000530496					
GTF2H1	ENST00000265963					
GTF2H1	ENST00000453096					
HPS5	ENST00000438420	ILMN_1655312	monocyte	1.77×10-14 (p-value)	Zeller et al.	7
HPS5	ENST00000349215					
HPS5	ENST00000396253					
HPS5	ENST00000438420	ILMN_2411731	monocyte	1.18×10-12 (p-value)	Zeller et al.	3
HPS5	ENST00000349215					
HPS5	ENST00000396253					
GTF2H1	?	ENSG00000110768	caudate basal ganglia	1.61×10-10 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	stomach	7.82×10-18 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	visceral adipocytes	5.18×10-18 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	cortex	2.04×10-9 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	spleen	4.46×10-10 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	sigmoid colon	1.48×10-6 (p-value)	GTEx Portal V6	14
GTF2H1	?	ENSG00000110768	tibial nerve	2.83×10-15 (p-value)	GTEx Portal V6	44
SAA1	?	ENSG00000173432	tibial nerve	1.62×10-5 (p-value)	GTEx Portal V6	8
GTF2H1	?	ENSG00000110768	esophagus mucosa	8.35×10-37 (p-value)	GTEx Portal V6	44
GTF2H1	ENST00000534641	ILMN_2157957	blood	1.42×10-5 (p-value)	Westra et al.	25
GTF2H1	ENST00000265963					
HPS5	ENST00000438420	ILMN_1655312	blood	7.84×10-7 (p-value)	Westra et al.	1
HPS5	ENST00000349215					
HPS5	ENST00000396253					

HPS5	ENST00000396253					
GTF2H1	?	ENSG00000110768	adrenal gland	1.02×10-19 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	testis	1.27×10-24 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	cerebellum	5.54×10-7 (p-value)	GTEx Portal V6	35
GTF2H1	?	ENSG00000110768	frontal cortex	1.14×10-8 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	blood	2.20×10-2 (q-value)	SeeQTL DB (HapMap)	9
GTF2H1	?	ENSG00000110768	terminal ileum	6.14×10-8 (p-value)	GTEx Portal V6	22
GTF2H1	?	ENSG00000110768	hypothalamus	1.22×10-7 (p-value)	GTEx Portal V6	33
GTF2H1	?	ENSG00000110768	blood	4.85×10-6 (p-value)	GTEx Portal V6	9
GTF2H1	?	ENSG00000110768	nucleus accumbens	1.06×10-6 (p-value)	GTEx Portal V6	15
?	?	ILMN_1726693	liver	6.66×10-15 (p-value)	Innocenti et al.	1

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
MYC, MAX	MA0059.1	10	no	-0.060	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000011407		
ENCP00000011408		HPS5

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000100096	1	ENCP00000011410	GTF2H1
		ENCP00000011419	LDHA
		ENCP00000011415	LDHA
		ENCP00000011420	AC084117.3
		ENCP00000011418	LDHA
ENCE00000100028	1	ENCP00000011426	TSG101 RP11-613F22.6
		ENCP00000011396	RP11-113D6.3
		ENCP00000011430	UEVLD
		ENCP00000011429	UEVLD

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000559412 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	DNase1, H3K36me3, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	Egr1, H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3, H3K4me2
		skin (NHEK)	DNase1, H3K36me3
		NHLF	DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3

		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K27ac, H3K4me1, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001568729 (promoter)	3	embryonic stem cell (H1ESC)	TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, Yy1, ATF3, H4K5ac, H3K36me3, H3K27me3, PolII, Gabp, TAF7, Sin3Ak20, H3K27ac, DNase1
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K79me2
		blood (K562)	Nfe2, E2F6, H3K9ac, Gabp, USF1, H3K79me2, HEY1, H2AZ, TAF1, H3K27ac, Yy1, ELF1, Jund, Max, Cfos, Cmyc, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	ATF3, H3K4me3, H3K27me3, H3K36me3, Cmyc, H3K27ac, H3K9ac, Yy1, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H3K4me2, Gabp, DNase1
		blood (GM12878)	ATF3, PU1, Max, PolII, H2AZ, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, USF1, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H4K8ac, H3K79me2, H3K27ac, H3K18ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K27ac, H3K9ac, H3K4me2
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H2AZ, H3K27ac, H3K4me3
		blood (DND-41)	H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3, H3K4me1
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, PolII, H3K36me3, Gabp, H3K79me2, BAF170, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, PolII, H3K27ac, H3K9ac, H3K4me3, H3K4me2, Max, Cjun, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00000684954 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	DNase1, Nfe2, H3K79me2, H3K36me3
		breast (HMEC)	H3K36me3, H3K4me2, H3K27ac
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K79me2, H3K27ac, BATF, H3K4me3
		A549	H3K36me3
		skin (NHEK)	H3K4me2, H3K36me3
ENSR00000559415 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001568731 (open chromatin region)	1	A549	H3K36me3
		blood (K562)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GTF2H1	upstream gene variant, downstream gene variant	152	ENST00000532227	?	?	12
GTF2H1	upstream gene variant, downstream gene variant	20	ENST00000453096	NM_001142307.1	ENSP00000393638	7

GTF2H1	upstream gene variant, downstream gene variant	646	ENST00000526282	?	?	5
GTF2H1	upstream gene variant, downstream gene variant	2380	ENST00000526630	?	ENSP00000439774	2
GTF2H1	upstream gene variant, downstream gene variant	226	ENST00000418116	?	?	11
GTF2H1	upstream gene variant, downstream gene variant	236	ENST00000534641	?	ENSP00000435375	9
GTF2H1	upstream gene variant, downstream gene variant	216	ENST00000531757	?	?	8
GTF2H1	upstream gene variant, downstream gene variant	238	ENST00000525831	?	ENSP00000431481	8
GTF2H1	upstream gene variant, downstream gene variant	265	ENST00000265963	NM_005316.3	ENSP00000265963	9
GTF2H1	upstream gene variant, downstream gene variant	237	ENST00000526461	?	?	11
GTF2H1	upstream gene variant, downstream gene variant	190	ENST00000528427	?	?	5
GTF2H1	upstream gene variant, downstream gene variant	548	ENST00000607664	?	?	7
GTF2H1	upstream gene variant, downstream gene variant	147	ENST00000534213	?	?	13
GTF2H1	upstream gene variant, downstream gene variant	361	ENST00000524753	?	ENSP00000436575	11
GTF2H1	upstream gene variant, downstream gene variant	2941	ENST00000530496	?	ENSP00000433133	7
GTF2H1	upstream gene variant, downstream gene variant	898	ENST00000543932	?	?	7
HPS5	downstream gene variant, upstream gene variant	97	ENST00000399287	?	?	5
HPS5	upstream gene variant	101	ENST00000438420	NM_181508.1	ENSP00000399590	4
HPS5	upstream gene variant	101	ENST00000349215	NM_181507.1	ENSP00000265967	4
HPS5	downstream gene variant, upstream gene variant	187	ENST00000589545	?	?	4
HPS5	upstream gene variant	101	ENST00000396253	NM_007216.3	ENSP00000379552	4
HPS5	upstream gene variant	1239	ENST00000537258	?	ENSP00000437437	1
HPS5	upstream gene variant	422	ENST00000545561	?	?	2
HPS5	downstream gene variant, upstream gene variant	77	ENST00000531848	?	ENSP00000431758	6

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
HPS5	ENST00000531848	?	ENSP00000431758	L	ctC/ctA	1
HPS5	ENST00000396253	NM_007216.3	ENSP00000379552	L	ctC/ctA	1
HPS5	ENST00000349215	NM_181507.1	ENSP00000265967	L	ctC/ctA	1
HPS5	ENST00000438420	NM_181508.1	ENSP00000399590	L	ctC/ctA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GTF2H1	ENST00000265963	NM_005316.3	ENSP00000265963	30
GTF2H1	ENST00000531757	?	?	9
GTF2H1	ENST00000528427	?	?	6
GTF2H1	ENST00000526630	?	ENSP00000439774	13
GTF2H1	ENST00000526282	?	?	7
GTF2H1	ENST00000530496	?	ENSP00000433133	14
GTF2H1	ENST00000524753	?	ENSP00000436575	7
GTF2H1	ENST00000543932	?	?	7

GTF2H1	ENST00000534213	?	?	3
GTF2H1	ENST00000526461	?	?	5
GTF2H1	ENST00000525831	?	ENSP00000431481	9
GTF2H1	ENST00000453096	NM_001142307.1	ENSP00000393638	30
GTF2H1	ENST00000418116	?	?	26
GTF2H1	ENST00000532227	?	?	5
GTF2H1	ENST00000534641	?	ENSP00000435375	30
HPS5	ENST00000399287	?	?	6
HPS5	ENST00000589545	?	?	6
HPS5	ENST00000531848	?	ENSP00000431758	10
HPS5	ENST00000544218	?	ENSP00000441781	3
HPS5	ENST00000396253	NM_007216.3	ENSP00000379552	13
HPS5	ENST00000543728	?	?	3
HPS5	ENST00000349215	NM_181507.1	ENSP00000265967	13
HPS5	ENST00000352460	?	?	3
HPS5	ENST00000545561	?	?	1
HPS5	ENST00000438420	NM_181508.1	ENSP00000399590	13

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GTF2H1	ENST00000453096	NM_001142307.1	ENSP00000393638	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
GTF2H1	ENST00000528427	?	1
GTF2H1	ENST00000526461	?	2
GTF2H1	ENST00000532227	?	2

Locus 122 (rs1339847)

SNiPAcard

Block annotations

Block info

genomic range	chr1:248,038,212-248,040,104
block size	1,893 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.718 [-1.477 – 0.039]	gene(s) hit or close-by	TRIM58
phastCons	μ = 0.071 [0 – 0.28]	eQTL gene(s)	–
GERP++	μ = -2.271 [-6.98 – 1.88]	potentially regulated gene(s)	–
CADD score	μ = 4.373 [0.202 – 13.74]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Hypertension	5.36×10-5	dbGaP	pha003041	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
TRIM58	missense variant	ENST00000366481	NM_015431.3	ENSP00000355437	I/V	Atc/Gtc	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TRIM58	ENST00000366481	NM_015431.3	ENSP00000355437	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TRIM58	ENST00000366481	NM_015431.3	ENSP00000355437	1

Locus 123 (rs9283893)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,882,421-31,935,928
block size	53,508 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.590 [-1.89 – 0.405]	gene(s) hit or close-by	C2, DXO, MIR1236, NELFE, SKIV2L, STK19, XXbac-BPG116M5.17
phastCons	μ = 0.025 [0 – 0.14]	eQTL gene(s)	SKIV2L
GERP++	μ = 0.497 [-0.607 – 1.57]	potentially regulated gene(s)	–
CADD score	μ = 3.537 [0.258 – 7.157]	disease gene(s)	SKIV2L, C2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SKIV2L	?	ENSG00000204351	muscularis mucosae	3.96×10-6 (p-value)	GTEx Portal V6	1
SKIV2L	?	ENSG00000204351	tibial nerve	6.19×10-7 (p-value)	GTEx Portal V6	3
SKIV2L	?	ENSG00000204351	transverse colon	9.58×10-8 (p-value)	GTEx Portal V6	3
SKIV2L	?	ENSG00000204351	unexposed skin	1.40×10-8 (p-value)	GTEx Portal V6	5
SKIV2L	?	ENSG00000204351	tibial artery	4.93×10-8 (p-value)	GTEx Portal V6	4
SKIV2L	?	ENSG00000204351	thyroid	1.77×10-7 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494593 (enhancer)	1	liver (HepG2)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K79me2, ELF1, H3K4me1
		HSMMtube	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C2	upstream gene variant	2906	ENST00000494905	?	ENSP00000419048	1
C2	upstream gene variant	2111	ENST00000411571	?	ENSP00000388727	2
C2	upstream gene variant, downstream gene variant	548	ENST00000413154	?	ENSP00000403325	3
C2	upstream gene variant	2266	ENST00000484636	?	ENSP00000420305	2
C2	upstream gene variant	4841	ENST00000485690	?	ENSP00000417790	1
DXO	downstream gene variant	1659	ENST00000477826	?	?	1
DXO	downstream gene variant	1693	ENST00000375356	?	ENSP00000364505	1
DXO	downstream gene variant	1666	ENST00000478221	?	?	1
DXO	downstream gene variant	1659	ENST00000491327	?	?	1
DXO	downstream gene variant	2492	ENST00000487914	?	?	1
DXO	downstream gene variant	2291	ENST00000492946	?	?	1
DXO	downstream gene variant	1662	ENST00000375349	?	ENSP00000364498	1
DXO	downstream gene variant	1797	ENST00000474587	?	?	1
DXO	downstream gene variant	1756	ENST00000480240	?	ENSP00000418632	1
DXO	downstream gene variant	1736	ENST00000498357	?	?	1
DXO	downstream gene variant	1659	ENST00000337523	NM_005510.3	ENSP00000337759	1
DXO	downstream gene variant	1664	ENST00000495340	?	ENSP00000419588	1
DXO	downstream gene variant	1659	ENST00000485557	?	?	1
DXO	downstream gene variant	1664	ENST00000473976	?	?	1
DXO	downstream gene variant	2747	ENST00000460058	?	?	1
MIR1236	upstream gene variant	980	ENST00000408340	?	?	1
SKIV2L	upstream gene variant	3577	ENST00000466290	?	?	1
SKIV2L	upstream gene variant	373	ENST00000470453	?	?	1
SKIV2L	upstream gene variant	324	ENST00000491994	?	ENSP00000417586	1
SKIV2L	upstream gene variant	1271	ENST00000492900	?	?	1
SKIV2L	upstream gene variant	1197	ENST00000488648	?	?	1
SKIV2L	upstream gene variant	1251	ENST00000474839	?	ENSP00000420470	1
SKIV2L	upstream gene variant	1160	ENST00000375394	NM_006929.4	ENSP00000364543	1
SKIV2L	upstream gene variant	1221	ENST00000465703	?	?	1
SKIV2L	upstream gene variant	1237	ENST00000461073	?	ENSP00000419905	1
SKIV2L	upstream gene variant	1273	ENST00000628157	?	ENSP00000485707	1
SKIV2L	upstream gene variant	172	ENST00000471818	?	?	1
SKIV2L	upstream gene variant	303	ENST00000485349	?	?	1
STK19	upstream gene variant	3793	ENST00000375333	NM_032454.1	ENSP00000364482	1
STK19	upstream gene variant	4189	ENST00000473983	?	?	1
STK19	upstream gene variant	3015	ENST00000460018	?	ENSP00000418350	1
STK19	upstream gene variant	4431	ENST00000469907	?	?	1
STK19	upstream gene variant	4230	ENST00000483801	?	ENSP00000418866	1
STK19	upstream gene variant	3680	ENST00000375331	NM_004197.1	ENSP00000364480	1
STK19	upstream gene variant	3680	ENST00000479644	?	?	1
STK19	upstream gene variant	3748	ENST00000492583	?	?	1

STK19	upstream gene variant	4427	ENST00000484540	?	?	1
STK19	upstream gene variant	4223	ENST00000463823	?	?	1
STK19	upstream gene variant	4384	ENST00000471028	?	?	1
STK19	upstream gene variant	4230	ENST00000519179	?	ENSP00000454870	1
STK19	upstream gene variant	4246	ENST00000466336	?	?	1
STK19	upstream gene variant	2940	ENST00000466132	?	?	1
STK19	upstream gene variant	4201	ENST00000478486	?	?	1
STK19	upstream gene variant	4404	ENST00000491861	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C2	ENST00000484636	?	ENSP00000420305	1
C2	ENST00000299367	NM_001282458.1, NM_000063.5	ENSP00000299367	1
C2	ENST00000452323	NM_001178063.2	ENSP00000392322	4
C2	ENST00000442278	NM_001145903.2	ENSP00000395683	1
C2	ENST00000452202	?	ENSP00000406121	4
C2	ENST00000482060	?	ENSP00000418332	1
C2	ENST00000469372	NM_001282457.1	ENSP00000418923	4
C2	ENST00000418949	NM_001282459.1	ENSP00000406190	1
C2	ENST00000497706	?	ENSP00000417482	4
C2	ENST00000383177	?	ENSP00000372664	1
C2	ENST00000411571	?	ENSP00000388727	1
C2	ENST00000447952	?	ENSP00000391354	1
NELFE	ENST00000491139	?	?	1
NELFE	ENST00000436289	?	ENSP00000414029	1
NELFE	ENST00000492185	?	?	1
NELFE	ENST00000481121	?	?	1
NELFE	ENST00000426722	?	ENSP00000394340	1
NELFE	ENST00000375429	NM_002904.5	ENSP00000364578	1
NELFE	ENST00000625905	?	ENSP00000486884	1
NELFE	ENST00000375425	?	ENSP00000364574	1
NELFE	ENST00000444811	?	ENSP00000388400	1
NELFE	ENST00000492539	?	ENSP00000419183	1
NELFE	ENST00000441998	?	ENSP00000397914	1
NELFE	ENST00000494956	?	ENSP00000420205	1
NELFE	ENST00000488426	?	?	1
NELFE	ENST00000454913	?	ENSP00000409389	1
SKIV2L	ENST00000484835	?	?	1
SKIV2L	ENST00000375394	NM_006929.4	ENSP00000364543	1

SKIV2L	ENST00000461915	?	?	1
SKIV2L	ENST00000474839	?	ENSP00000420470	1
SKIV2L	ENST00000483553	?	ENSP00000420332	1
SKIV2L	ENST00000465703	?	?	1
XXbac-BPG116M5.17	ENST00000477310	?	ENSP00000418996	1
XXbac-BPG116M5.17	ENST00000456570	?	ENSP00000410815	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SKIV2L	ENST00000494058	?	1

Locus 124 (rs1568003)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,632,705-51,636,001
block size	3,297 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.624 [-2.525 – 0.424]	gene(s) hit or close-by	SIGLEC9
phastCons	μ = 0.035 [0.001 – 0.103]	eQTL gene(s)	–
GERP++	μ = -0.323 [-0.764 – 0.113]	potentially regulated gene(s)	–
CADD score	μ = 3.481 [0.49 – 7.265]	disease gene(s)	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001649020 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac, H3K4me3
		embryonic stem cell (H1ESC)	CTCF
		endothelium (HUVEC)	H3K36me3, DNase1
		blood (K562)	CTCF, DNase1
		A549	H3K27me3
ENSR00000217699 (promoter flanking region)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1
		embryonic stem cell (H1ESC)	CTCF
		A549	H3K27me3
		blood (K562)	CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SIGLEC9	downstream gene variant	490	ENST00000599948	?	ENSP00000472483	3
SIGLEC9	downstream gene variant	93	ENST00000250360	NM_014441.2	ENSP00000250360	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC9	ENST00000440804	NM_001198558.1	ENSP00000413861	3
SIGLEC9	ENST00000250360	NM_014441.2	ENSP00000250360	1

Locus 125 (rs45461791)

SNiPAcard

Block annotations

Block info

genomic range	chr19:55,110,880-55,149,838
block size	38,959 bp
variant count	51 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.429 [-3.615 – 1.303]	gene(s) hit or close-by	AC009892.10, AC009892.5, AC245036.1, LILRA1, LILRB1
phastCons	μ = 0.067 [0 – 0.948]	eQTL gene(s)	LILRB1
GERP++	μ = -0.277 [-3.51 – 2.1]	potentially regulated gene(s)	AC008746.10, AC008746.10, AC008984.2, AC008984.2, AC008984.6, AC008984.6, AC009892.2, AC009892.2, AC009892.5, AC009892.9, AC012314.20, AC012314.20, AC012314.8, AC012314.8, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, KIR3DX1, KIR3DX1, KIR3DX1, KIR3DX1, LAIR1, LAIR1, LAIR1, LAIR1, LAIR1, LAIR2, LAIR2, LAIR2, LAIR2, LILRA4, LILRA4, LILRA4, LILRA4, LILRB1, LILRB1, LILRB1, LILRB1, LILRB1, LILRP2, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TTYH1, TTYH1, TTYH1, TTYH1
CADD score	μ = 3.491 [0.047 – 15.32]	disease gene(s)	, TSEN34, , , , , , , , , CNOT3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
TSEN34	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
TSEN34	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970

	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970	OMIM
TSEN34	Pontocerebellar hypoplasia type 2	OrphaNet	OrphaNet:2524	orphanet
CNOT3	Precursor T-cell acute lymphoblastic leukemia	OrphaNet	OrphaNet:99861	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
AC009892.5	splice donor variant	ENST00000432400	?	?					1
LILRB1	missense variant	ENST00000324602	NM_001081637.1	ENSP00000315997	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396315	?	ENSP00000379608	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396331	NM_006669.5	ENSP00000379622	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000427581	?	ENSP00000395004	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396327	NM_001081638.2	ENSP00000379618	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000421584	?	ENSP00000410165	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396317	NM_001278398.1	ENSP00000379610	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396332	NM_001081639.2	ENSP00000379623	V/L	Gtg/Ctg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LILRB1	ENST00000462628	ILMN_1708248	blood	9.02×10^{-31} (p-value)	MuTHER consortium	1
LILRB1	ENST00000396315					
LILRB1	ENST00000427581					
LILRB1	ENST00000396327					
LILRB1	ENST00000396317					
LILRB1	ENST00000480375					
LILRB1	ENST00000324602					
LILRB1	ENST00000421584					
LILRB1	ENST00000396332					
LILRB1	ENST00000396331					
LILRB1	?	ENSG00000104972	testis	3.99×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000344796	1	ENST00000418536, ENST00000396321	LILRB1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024800		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000233565	2	ENCP00000024749	AC012314.20 AC012314.20
		ENCP00000024780	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024781	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024748	TSEN34 TSEN34 TSEN34 TSEN34 TSEN34 TSEN34 TSEN34 TSEN34 TSEN34
		ENCP00000024778	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024742	MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7
		ENCP00000024745	MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7
		ENCP00000024733	CNOT3 CNOT3 CNOT3 CNOT3 CNOT3 CNOT3 CNOT3 CNOT3 CNOT3
		ENCP00000024804	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024777	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024741	TMC4 TMC4 TMC4

			TMC4 TMC4 TMC4 TMC4 TMC4 TMC4 TMC4
		ENCP00000024728	AC012314.8 AC012314.8
ENCE00000233676	1	ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1
		ENCP00000024826	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024805	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024809	AC009892.9
		ENCP00000024796	AC009892.2 AC009892.2
		ENCP00000024793	LAIR2 LAIR2 LAIR2 LAIR2
		ENCP00000024801	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
ENCE00000233601	1	ENCP00000024767	LILRA4 LILRA4 LILRA4 LILRA4 AC008984.2 AC008984.2
		ENCP00000024744	MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7
		ENCP00000024775	AC008746.10 AC008746.10
		ENCP00000024779	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024792	LAIR2 LAIR2 LAIR2 LAIR2

		ENCP00000024814	AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1
		ENCP00000024815	LILRP2 LILRP2
		ENCP00000024774	LAIR1 LAIR1 LAIR1 LAIR1 LAIR1
ENCE00000233679	1	ENCP00000024815	LILRP2 LILRP2
		ENCP00000024796	AC009892.2 AC009892.2
		ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024793	LAIR2 LAIR2 LAIR2

		ENCP00000024826	LAIR2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024801	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1
ENCE00000233738	1	ENCP00000024793	LAIR2 LAIR2 LAIR2 LAIR2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348881 (open chromatin region)	7	blood (GM12878)	H3K4me3, H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		A549	H3K27me3
ENSR00000348884 (promoter)	1	cervix (HeLa-S3)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	Yy1, PU1, PolII, BATF, DNase1, IRF4, H3K79me2, H3K4me3, BCL11A, Pax5, H3K27ac, H3K4me2, H3K9ac
		A549	H3K27me3
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC009892.10	downstream gene variant	1067	ENST00000456337	?	?	3
AC009892.5	upstream gene variant, downstream gene variant	53	ENST00000432400	?	?	32
AC245036.1	downstream gene variant, upstream gene variant	678	ENST00000578908	?	?	5
LILRA1	downstream gene variant	1211	ENST00000495417	?	?	2
LILRA1	downstream gene variant	1447	ENST00000618665	?	ENSP00000479482	2
LILRA1	downstream gene variant	2156	ENST00000453777	NM_001278318.1	ENSP00000413715	4
LILRA1	downstream gene variant	1215	ENST00000477255	?	?	2
LILRA1	downstream gene variant	3171	ENST00000251372	NM_006863.3	ENSP00000251372	3
LILRA1	downstream gene variant	2369	ENST00000473156	?	?	4
LILRB1	upstream gene variant, downstream gene variant	859	ENST00000324602	NM_001081637.1	ENSP00000315997	3
LILRB1	upstream gene variant, downstream gene variant	315	ENST00000421584	?	ENSP00000410165	4
LILRB1	upstream gene variant, downstream gene variant	859	ENST00000396331	NM_006669.5	ENSP00000379622	4
LILRB1	upstream gene variant	2360	ENST00000396321	?	ENSP00000379614	2
LILRB1	upstream gene variant, downstream gene variant	315	ENST00000396317	NM_001278398.1	ENSP00000379610	4
LILRB1	upstream gene variant, downstream gene variant	859	ENST00000462628	?	?	2
LILRB1	upstream gene variant	2360	ENST00000630741	?	ENSP00000485956	2
LILRB1	upstream gene variant, downstream gene variant	238	ENST00000480375	?	?	6

LILRB1	upstream gene variant, downstream gene variant	859	ENST00000396332	NM_001081639.2	ENSP00000379623	3
LILRB1	upstream gene variant, downstream gene variant	859	ENST00000396327	NM_001081638.2	ENSP00000379618	4
LILRB1	upstream gene variant, downstream gene variant	2230	ENST00000480257	?	?	6
LILRB1	upstream gene variant, downstream gene variant	315	ENST00000396315	?	ENSP00000379608	4
LILRB1	upstream gene variant, downstream gene variant	102	ENST00000473412	?	?	7
LILRB1	upstream gene variant, downstream gene variant	201	ENST00000427581	?	ENSP00000395004	4
LILRB1	upstream gene variant, downstream gene variant	86	ENST00000487425	?	?	4
LILRB1	upstream gene variant	2322	ENST00000418536	?	ENSP00000391514	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC009892.10	ENST00000456337	?	?	3
LILRA1	ENST00000251372	NM_006863.3	ENSP00000251372	1
LILRA1	ENST00000453777	NM_001278318.1	ENSP00000413715	1
LILRA1	ENST00000473156	?	?	1
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	3
LILRB1	ENST00000396321	?	ENSP00000379614	6
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	4
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	4
LILRB1	ENST00000630741	?	ENSP00000485956	6
LILRB1	ENST00000418536	?	ENSP00000391514	6
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	12
LILRB1	ENST00000421584	?	ENSP00000410165	3
LILRB1	ENST00000396315	?	ENSP00000379608	3
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	4
LILRB1	ENST00000462628	?	?	5
LILRB1	ENST00000427581	?	ENSP00000395004	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LILRA1	ENST00000618665	?	ENSP00000479482	1
LILRA1	ENST00000251372	NM_006863.3	ENSP00000251372	2
LILRA1	ENST00000453777	NM_001278318.1	ENSP00000413715	2
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	1
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	2
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	2
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	2
LILRB1	ENST00000421584	?	ENSP00000410165	1
LILRB1	ENST00000396315	?	ENSP00000379608	1
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	2
LILRB1	ENST00000427581	?	ENSP00000395004	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC009892.5	ENST00000432400	?	1
LILRA1	ENST00000477255	?	1
LILRA1	ENST00000495417	?	1
LILRA1	ENST00000473156	?	2
LILRB1	ENST00000462628	?	4
LILRB1	ENST00000487425	?	2

Locus 126 (rs1654439)

SNiPAcard

Block annotations

Block info

genomic range	chr19:55,548,269-55,571,055
block size	22,787 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.333 [-2.99 – 0.001]	gene(s) hit or close-by	CTC-550B14.6, CTC-550B14.7, GP6, RDH13
phastCons	μ = 0.015 [0 – 0.135]	eQTL gene(s)	CTC-550B14.6, CTC-550B14.7, GP6, NLRP2, RDH13
GERP++	μ = -1.023 [-10.2 – 0.367]	potentially regulated gene(s)	–
CADD score	μ = 2.601 [0.974 – 5.875]	disease gene(s)	GP6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
GP6	BLEEDING DISORDER, PLATELET-TYPE, 11	OMIM	MIM:614201
GP6	Bleeding diathesis due to glycoprotein VI deficiency	OrphaNet	OrphaNet:98885

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NLRP2	?	ENSG00000022556	visceral adipocytes	6.13×10-7 (p-value)	GTEx Portal V6	14
CTC-550B14.7	?	ENSG00000267265	lung	1.01×10-10 (p-value)	GTEx Portal V6	16
CTC-550B14.7	?	ENSG00000267265	spleen	8.64×10-11 (p-value)	GTEx Portal V6	14
RDH13	?	ENSG00000160439	spleen	2.66×10-6 (p-value)	GTEx Portal V6	2
NLRP2	?	ENSG00000022556	spleen	1.02×10-7 (p-value)	GTEx Portal V6	3
RDH13	?	ENSG00000160439	blood	1.40×10-7 (p-value)	GTEx Portal V6	7
CTC-550B14.7	?	ENSG00000267265	blood	8.09×10-8 (p-value)	GTEx Portal V6	14
NLRP2	?	ENSG00000022556	blood	6.52×10-8 (p-value)	GTEx Portal V6	12
CTC-550B14.6	?	ENSG00000267149	esophagus mucosa	2.71×10-8 (p-value)	GTEx Portal V6	14
NLRP2	?	ENSG00000022556	esophagus mucosa	6.53×10-6 (p-value)	GTEx Portal V6	1
GP6	?	ENSG00000088053	thyroid	3.22×10-6 (p-value)	GTEx Portal V6	6
NLRP2	?	ENSG00000022556	thyroid	1.73×10-6 (p-value)	GTEx Portal V6	2
CTC-550B14.6	?	ENSG00000267149	skeletal muscle	1.61×10-6 (p-value)	GTEx Portal V6	5
CTC-550B14.6	?	ENSG00000267149	sun exposed skin	2.53×10-20 (p-value)	GTEx Portal V6	16
RDH13	?	ENSG00000160439	sun exposed skin	2.71×10-13 (p-value)	GTEx Portal V6	16
CTC-550B14.6	?	ENSG00000267149	unexposed skin	7.77×10-13 (p-value)	GTEx Portal V6	16
RDH13	?	ENSG00000160439	unexposed skin	2.76×10-6 (p-value)	GTEx Portal V6	6

CTC-550B14.7	?	ENSG00000267265	subcutaneous adipocytes	2.24×10^{-8} (p-value)	GTEx Portal V6	16
NLRP2	ENST00000537859	ILMN_1664878	skin	4.74×10^{-13} (p-value)	MuTHER consortium	8
NLRP2	ENST00000540597		blood	1.39×10^{-20} (p-value)	MuTHER consortium	8
NLRP2	ENST00000263437					
NLRP2	ENST00000391721					
NLRP2	ENST00000339757					
NLRP2	ENST00000543010					
NLRP2	ENST00000448584					
NLRP2	ENST00000427260					
NLRP2	ENST00000542755					
NLRP2	?	ENSG00000022556	blood	1.37×10^{-3} (q-value)	SeeQTL DB (HapMap)	5
NLRP2	ENST00000537859	ILMN_1664878	monocyte	2.07×10^{-19} (p-value)	Zeller et al.	3
NLRP2	ENST00000540597					
NLRP2	ENST00000263437					
NLRP2	ENST00000391721					
NLRP2	ENST00000339757					
NLRP2	ENST00000543010					
NLRP2	ENST00000448584					
NLRP2	ENST00000427260					
NLRP2	ENST00000542755					
NLRP2	?	ENSG00000022556	tibial nerve	8.87×10^{-6} (p-value)	GTEx Portal V6	1
CTC-550B14.6	?	ENSG00000267149	stomach	8.27×10^{-7} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348908 (CTCF binding site)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, CTCF, Rad21
		blood (K562)	H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTC-550B14.6	upstream gene variant	1158	ENST00000585492	?	?	1
CTC-550B14.7	downstream gene variant	92	ENST00000586845	?	?	5
CTC-550B14.7	downstream gene variant	1339	ENST00000586961	?	?	1
CTC-550B14.7	downstream gene variant	2116	ENST00000593060	?	?	2
GP6	upstream gene variant	648	ENST00000333884	NM_001256017.2	ENSP00000334552	5
GP6	upstream gene variant	648	ENST00000310373	NM_001083899.2	ENSP00000308782	5
GP6	upstream gene variant	648	ENST00000417454	NM_016363.5	ENSP00000394922	5
GP6	upstream gene variant	4036	ENST00000468330	?	ENSP00000464922	1

GP6	upstream gene variant	4036	ENST00000468239	?	ENSP00000464922	1
RDH13	downstream gene variant, upstream gene variant	164	ENST00000592423	?	?	8
RDH13	downstream gene variant, upstream gene variant	707	ENST00000591868	?	?	4
RDH13	downstream gene variant	3584	ENST00000591023	?	?	2
RDH13	downstream gene variant	47	ENST00000587026	?	ENSP00000467754	2
RDH13	downstream gene variant	4214	ENST00000587373	?	?	2
RDH13	downstream gene variant	3533	ENST00000588306	?	?	2
RDH13	downstream gene variant	4353	ENST00000587046	?	?	2
RDH13	downstream gene variant	331	ENST00000396247	NM_138412.3	ENSP00000379547	7
RDH13	downstream gene variant	843	ENST00000592573	?	ENSP00000465538	7
RDH13	downstream gene variant	672	ENST00000589305	?	?	1
RDH13	downstream gene variant	331	ENST00000291892	?	ENSP00000291892	7
RDH13	downstream gene variant	1567	ENST00000586945	?	ENSP00000466708	1
RDH13	downstream gene variant	331	ENST00000415061	NM_001145971.1	ENSP00000391121	7
RDH13	downstream gene variant	167	ENST00000591960	?	ENSP00000468422	3
RDH13	downstream gene variant	312	ENST00000610356	?	ENSP00000477732	7
RDH13	downstream gene variant	3821	ENST00000589197	?	?	2
RDH13	downstream gene variant	1757	ENST00000587721	?	ENSP00000467208	6
RDH13	downstream gene variant	4261	ENST00000593134	?	?	2
RDH13	downstream gene variant, upstream gene variant	167	ENST00000586331	?	?	10
RDH13	downstream gene variant	4065	ENST00000590349	?	?	2
RDH13	downstream gene variant, upstream gene variant	32	ENST00000589605	?	?	6
RDH13	downstream gene variant	3354	ENST00000591603	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
RDH13	ENST00000291892	?	ENSP00000291892	G	ggT/ggC	1
RDH13	ENST00000592573	?	ENSP00000465538	G	ggT/ggC	1
RDH13	ENST00000415061	NM_001145971.1	ENSP00000391121	G	ggT/ggC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTC-550B14.6	ENST00000585492	?	?	13
CTC-550B14.7	ENST00000593060	?	?	11
CTC-550B14.7	ENST00000586845	?	?	6
CTC-550B14.7	ENST00000586961	?	?	10
GP6	ENST00000310373	NM_001083899.2	ENSP00000308782	1
GP6	ENST00000417454	NM_016363.5	ENSP00000394922	1
GP6	ENST00000333884	NM_001256017.2	ENSP00000334552	1
RDH13	ENST00000396247	NM_138412.3	ENSP00000379547	6
RDH13	ENST00000589605	?	?	3

RDH13	ENST00000586945	?	ENSP00000466708	2
RDH13	ENST00000291892	?	ENSP00000291892	6
RDH13	ENST00000610356	?	ENSP00000477732	6
RDH13	ENST00000592573	?	ENSP00000465538	6
RDH13	ENST00000415061	NM_001145971.1	ENSP00000391121	6
RDH13	ENST00000591960	?	ENSP00000468422	13
RDH13	ENST00000587721	?	ENSP00000467208	5
RDH13	ENST00000589305	?	?	4
RDH13	ENST00000591868	?	?	6
RDH13	ENST00000587026	?	ENSP00000467754	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RDH13	ENST00000396247	NM_138412.3	ENSP00000379547	1
RDH13	ENST00000586945	?	ENSP00000466708	1
RDH13	ENST00000610356	?	ENSP00000477732	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTC-550B14.7	ENST00000593060	?	3
CTC-550B14.7	ENST00000586845	?	3
CTC-550B14.7	ENST00000586961	?	3
RDH13	ENST00000586331	?	1
RDH13	ENST00000592423	?	3
RDH13	ENST00000589305	?	1

Locus 127 (rs2297363)

SNiPAcard

Block annotations

Block info

genomic range	chr6:160,476,945-160,513,793
block size	36,849 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.604 [-2.574 – 0.77]	gene(s) hit or close-by	IGF2R, RP11-288H12.3
phastCons	μ = 0.001 [0 – 0.006]	eQTL gene(s)	IGF2R, RP11-288H12.3
GERP++	μ = -1.707 [-6.01 – 1.86]	potentially regulated gene(s)	LPA, RP1-276N6.2
CADD score	μ = 2.880 [0.501 – 6.396]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
isobutyrylcarnitine	5.24×10^{-9}	Metabolomics GWAS Server	24816252	8

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-288H12.3	?	ENSG00000213073	ovary	2.23×10^{-6} (p-value)	GTEx Portal V6	1
RP11-288H12.3	?	ENSG00000213073	blood	2.66×10^{-14} (p-value)	GTEx Portal V6	9
RP11-288H12.3	?	ENSG00000213073	transformed fibroblasts	4.66×10^{-8} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	aorta	4.15×10^{-6} (p-value)	GTEx Portal V6	8
RP11-288H12.3	?	ENSG00000213073	thyroid	1.78×10^{-6} (p-value)	GTEx Portal V6	8
RP11-288H12.3	?	ENSG00000213073	EBV lymphocytes	6.38×10^{-7} (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000468394	1	ENCP00000050541	RP1-276N6.2
		ENCP00000050544	LPA

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001232516 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		blood (K562)	H3K36me3
		skin (NHEK)	H3K36me3
		breast (HMEC)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IGF2R	downstream gene variant	2956	ENST00000487607	?	?	1
IGF2R	upstream gene variant	3686	ENST00000475584	?	?	1
IGF2R	upstream gene variant, downstream gene variant	2199	ENST00000475834	?	ENSP00000473880	3
RP11-288H12.3	upstream gene variant	662	ENST00000366273	?	?	2
RP11-288H12.3	upstream gene variant	321	ENST00000569097	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IGF2R	ENST00000356956	NM_000876.2	ENSP00000349437	9
IGF2R	ENST00000487607	?	?	1
IGF2R	ENST00000475834	?	ENSP00000473880	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IGF2R	ENST00000475834	?	ENSP00000473880	1

Locus 128 (rs8071756)

SNiPAcard

Block annotations

Block info

genomic range	chr17:1,574,342-1,587,212
block size	12,871 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.511 [-1.525 – 0.101]	**gene(s) hit or close-by**	PRPF8, RP11-961A15.3
phastCons	μ = 0.077 [0 – 0.922]	**eQTL gene(s)**	–
GERP++	μ = -0.848 [-3.44 – 1.67]	**potentially regulated gene(s)**	–
CADD score	μ = 6.553 [1.41 – 16]	**disease gene(s)**	PRPF8

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PRPF8	RETINITIS PIGMENTOSA 13	OMIM	MIM:600059
PRPF8	Retinitis pigmentosa	OrphaNet	OrphaNet:791

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001338852 (promoter)	1	embryonic stem cell (H1ESC)	SP1, H3K36me3, PolII, H3K9ac, H3K27ac, Yy1, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K9ac, H3K27ac, DNase1, H3K36me3, H2AZ, H3K4me3
		blood (K562)	H3K4me3, DNase1, H3K36me3, H3K4me2, PolII, Cfos, H3K79me2, Cmyc, H3K9ac, Max
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3
		muscle (HSMM)	DNase1, H3K9ac, H2AZ, H3K79me2, H3K4me3, H3K27ac
		liver (HepG2)	H2AZ, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3, DNase1, H3K4me1, TAF1, H3K79me2, Gabp, Yy1, PolII
		lung (IMR90)	H3K9ac, H3K4me3, H3K36me3, H3K4me2, DNase1, H4K8ac, H3K79me2, H3K27ac
		blood (GM12878)	H3K9ac, H3K36me3, H3K4me2, Cfos, H3K27ac, Pbx3, PolII, H2AZ, DNase1, Cmyc, Yy1, Gabp, H3K79me2, H3K4me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K36me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K4me1, H3K4me2, H3K27ac, H4K20me1
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K4me3, H3K79me2, Gabp, PolII, H3K36me3, DNase1, TAF1, H3K27ac, H3K9ac, Jund, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	PolII, DNase1, H3K36me3, H3K9ac, H3K4me2, Max, H3K4me3
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PRPF8	downstream gene variant	358	ENST00000571346	?	?	2
PRPF8	downstream gene variant, upstream gene variant	806	ENST00000573716	?	?	10
PRPF8	downstream gene variant, upstream gene variant	987	ENST00000574217	?	?	10
PRPF8	downstream gene variant	142	ENST00000577001	?	ENSP00000458151	5

PRPF8	downstream gene variant, upstream gene variant	11	ENST00000576958	?	?	8
PRPF8	downstream gene variant, upstream gene variant	1620	ENST00000574728	?	?	5
PRPF8	downstream gene variant, upstream gene variant	364	ENST00000576407	?	?	10
RP11-961A15.3	upstream gene variant	808	ENST00000615078	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
PRPF8	ENST00000572621	?	ENSP00000460348	3	3	3
PRPF8	ENST00000304992	NM_006445.3	ENSP00000304350	3	3	3
PRPF8	ENST00000577001	?	ENSP00000458151	3	3	3

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
PRPF8	ENST00000572621	?	ENSP00000460348	1
PRPF8	ENST00000304992	NM_006445.3	ENSP00000304350	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PRPF8	ENST00000572621	?	ENSP00000460348	10
PRPF8	ENST00000304992	NM_006445.3	ENSP00000304350	10
PRPF8	ENST00000576958	?	?	3
PRPF8	ENST00000577001	?	ENSP00000458151	6
PRPF8	ENST00000571346	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PRPF8	ENST00000574728	?	1
PRPF8	ENST00000574217	?	1
PRPF8	ENST00000576958	?	1

Locus 129 (rs484915)

SNiPAcard

Block annotations

Block info

genomic range	chr11:102,671,697-102,673,248
block size	1,552 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.143 [-0.238 – -0.048]	gene(s) hit or close-by	AP000619.6, MMP1, WTAPP1
phastCons	μ = 0.001 [0.001 – 0.001]	eQTL gene(s)	MMP1, WTAPP1
GERP++	μ = -1.033 [-1.22 – -0.846]	potentially regulated gene(s)	DYNC2H1, MMP27, MMP3, RP11-817J15.3
CADD score	μ = 2.032 [1.406 – 2.658]	disease gene(s)	MMP3, DYNC2H1, MMP1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
MMP3	CORONARY HEART DISEASE, SUSCEPTIBILITY TO, 6	OMIM	MIM:614466
DYNC2H1	SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY	OMIM	MIM:613091
MMP1	Severe generalized recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79408
DYNC2H1	Short rib-polydactyly syndrome, Verma-Naumoff type	OrphaNet	OrphaNet:93271
DYNC2H1	Short rib-polydactyly syndrome, Majewski type	OrphaNet	OrphaNet:93269
DYNC2H1	Jeune syndrome	OrphaNet	OrphaNet:474
DYNC2H1	short rib-polydactyly syndrome type 3 (SRPS3)	DECIPHER	MIM:263510
DYNC2H1	asphyxiating thoracic dystrophy type 3 (ATD3)	DECIPHER	MIM:613091

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MMP1	?	ENSG00000196611	tibial nerve	1.40×10^{-5} (p-value)	GTEx Portal V6	2
MMP1	?	ENSG00000196611	transformed fibroblasts	1.64×10^{-13} (p-value)	GTEx Portal V6	2
WTAPP1	?	ENSG00000255282	transformed fibroblasts	2.04×10^{-11} (p-value)	GTEx Portal V6	2
MMP1	?	ENSG00000196611	left ventricle	2.57×10^{-7} (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000120249	1	ENCP00000014388	MMP27
		ENCP00000014401	MMP3
ENCE00000120235	1	ENCP00000014388	MMP27
		ENCP00000014387	RP11-817J15.3
		ENCP00000014401	MMP3
		ENCP00000014410	DYNC2H1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001572918 (promoter flanking region)	1	endothelium (HUVEC)	Max
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP000619.6	downstream gene variant	4431	ENST00000546014	?	?	1
MMP1	upstream gene variant	2806	ENST00000315274	NM_002421.3, NM_001145938.1	ENSP00000322788	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
WTAPP1	ENST00000525739	?	?	2
WTAPP1	ENST00000371455	?	?	2
WTAPP1	ENST00000544704	?	?	2

Locus 130 (rs13063312)

SNiPAcard

Block annotations

Block info

genomic range	chr3:48,661,985-48,765,168
block size	103,184 bp
variant count	75 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.176 [-2.807 – 4.885]	gene(s) hit or close-by	AC141002.1, CELSR3, CELSR3-AS1, IP6K2, MIR4793, MIR6824, NCKIPSD, RP11-572O6.1, SLC26A6, TMEM89
phastCons	μ = 0.103 [0 – 1]	eQTL gene(s)	ATRIP, C3orf62, CCDC36, CCDC71, CELSR3, IP6K2, KLHDC8B, MIR5193, NCKIPSD, NICN1, PFKFB4, RBM6, RP11-3B7.1, SLC26A6, TMEM89, TREX1, TREX1, UBA7, USP4, WDR6
GERP++	μ = -0.679 [-8.07 – 5.67]	potentially regulated gene(s)	ARIH2, ATRIP, CCDC51, CDC25A, COL7A1, COL7A1, DALRD3, IP6K2, NCKIPSD, PLXNB1, PRKAR2A, QARS, QRICH1, SHISA5, SLC25A20, SLC26A6, SPINK8, WDR6, ZNF589
CADD score	μ = 5.844 [0.048 – 22.1]	disease gene(s)	KLHDC8B, TREX1, TREX1, QARS, COL7A1, SLC25A20, ATRIP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Blood pressure	2.57×10^{-5}	dbGaP	pha003048	2
Blood pressure	9.40×10^{-6}	dbGaP	pha003047	3
Blood pressure	2.30×10^{-5}	dbGaP	pha003046	2
Blood pressure	2.52×10^{-5}	dbGaP	pha003045	2

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM086000	1

Disease gene annotation

gene	trait	source DB	source entry/link
KLHDC8B	LYMPHOMA, HODGKIN, CLASSIC	OMIM	MIM:236000
TREX1	VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY	OMIM	MIM:192315
TREX1	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
TREX1	CHILBLAIN LUPUS 1	OMIM	MIM:610448
TREX1	AICARDI-GOUTIERES SYNDROME 1	OMIM	MIM:225750
TREX1	VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY	OMIM	MIM:192315
TREX1	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
TREX1	CHILBLAIN LUPUS 1	OMIM	MIM:610448
TREX1	AICARDI-GOUTIERES SYNDROME 1	OMIM	MIM:225750
QARS	MICROCEPHALY, PROGRESSIVE, WITH SEIZURES AND CEREBRAL AND [...]	OMIM	MIM:615760
COL7A1	NAIL DISORDER, NONSYNDROMIC CONGENITAL, 8	OMIM	MIM:607523
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, PRETIBIAL	OMIM	MIM:131850
COL7A1	EPIDERMOLYSIS BULLOSA PRURIGINOSA	OMIM	MIM:604129
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL DOMINANT	OMIM	MIM:131750
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL RECESSIVE	OMIM	MIM:226600
COL7A1	EPIDERMOLYSIS BULLOSA WITH CONGENITAL LOCALIZED ABSENCE OF SKIN [...]	OMIM	MIM:132000
COL7A1	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN	OMIM	MIM:131705

SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OMIM	MIM:212138	OMIM
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive	DECIPHER	MIM:225750	OMIM
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive	DECIPHER	MIM:225750	OMIM
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	DECIPHER	MIM:615760	OMIM
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	DECIPHER	MIM:212138	OMIM
ATRIP	Seckel syndrome	OrphaNet	OrphaNet:808	orphanet
TREX1	Aicardi-Goutieres Syndrome	OrphaNet	OrphaNet:51	orphanet
TREX1	HERNS syndrome	OrphaNet	OrphaNet:63261	orphanet
TREX1	CHILBLAIN LUPUS	OrphaNet	OrphaNet:90280	orphanet
TREX1	Cerebroretinal vasculopathy	OrphaNet	OrphaNet:3421	orphanet
TREX1	Hereditary vascular retinopathy	OrphaNet	OrphaNet:71291	orphanet
COL7A1	Generalized dominant dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:231568	orphanet
COL7A1	Dystrophic epidermolysis bullosa pruriginosa	OrphaNet	OrphaNet:89843	orphanet
COL7A1	Recessive dystrophic epidermolysis bullosa inversa	OrphaNet	OrphaNet:79409	orphanet
COL7A1	Dystrophic epidermolysis bullosa, nails only	OrphaNet	OrphaNet:158676	orphanet
COL7A1	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN	OrphaNet	OrphaNet:79411	orphanet
COL7A1	Centripetalis recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:89841	orphanet
COL7A1	Acral dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:158673	orphanet
COL7A1	Recessive dystrophic epidermolysis bullosa-generalized other	OrphaNet	OrphaNet:89842	orphanet
COL7A1	Epidermolysis bullosa simplex superficialis	OrphaNet	OrphaNet:89839	orphanet
COL7A1	Severe generalized recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79408	orphanet
COL7A1	Pretibial dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79410	orphanet
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OrphaNet	OrphaNet:159	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CELSR3	missense variant	ENST00000164024	NM_001407.2	ENSP00000164024	2	2	tolerated, ?	benign, ?	2
IP6K2	missense variant	ENST00000417896	?	ENSP00000388116	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000433104	?	ENSP00000396904	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000443964	?	ENSP00000410950	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000449610	?	ENSP00000393077	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000453202	?	ENSP00000387394	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000446860	NM_001190317.1	ENSP00000399052	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000431721	NM_001190316.1	ENSP00000414139	G/D	gGt/gAt	?	?	1
SLC26A6	missense variant	ENST00000614797	?	ENSP00000483669	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000383733	NM_134426.2	ENSP00000373239	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000395550	NM_022911.2	ENSP00000378920	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000431739	?	ENSP00000401813	H/R	cAt/cGt	?	?	1
SLC26A6	missense variant	ENST00000444531	?	ENSP00000404607	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000455886	NM_001281732.1	ENSP00000401066	H/R	cAt/cGt	?	?	1
SLC26A6	missense variant	ENST00000421649	?	ENSP00000389922	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000358747	NM_001040454.1	ENSP00000351597	M/V	Atg/Gtg	?	?	1
SLC26A6	missense	ENST00000420764	NM_134263.2	ENSP00000404684	M/V	Atg/Gtg	?	?	1

	variant								
SLC26A6	missense variant	ENST00000414944	?	ENSP00000388721	H/R	cAt/cGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
WDR6	ENST00000615452	ILMN_1669484	skin	1.74×10-39 (p-value)	MuTHER consortium	19
WDR6	ENST00000452875		adipocyte	1.88×10-33 (p-value)	MuTHER consortium	19
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	skin	1.46×10-7 (p-value)	MuTHER consortium	19
USP4	ENST00000483212		adipocyte	5.95×10-12 (p-value)	MuTHER consortium	19
USP4	ENST00000485450		blood	7.52×10-5 (p-value)	MuTHER consortium	19
C3orf62	ENST00000343010					
TMEM89	ENST00000330862	ILMN_1694339	skin	2.13×10-7 (p-value)	MuTHER consortium	19
			adipocyte	3.59×10-5 (p-value)	MuTHER consortium	19
RP11-3B7.1	ENST00000622888	ILMN_1807244	adipocyte	4.30×10-16 (p-value)	MuTHER consortium	19
RP11-3B7.1	ENST00000440528					
ATRIP	ENST00000320211	ILMN_2370464	skin	3.13×10-5 (p-value)	MuTHER consortium	1
TREX1	ENST00000436480					
ATRIP	ENST00000346691					
TREX1	ENST00000395677					
TREX1	ENST00000296443					
TREX1	ENST00000424906					
ATRIP	ENST00000412052					
TREX1	ENST00000433541					
TREX1	ENST00000629913					
TREX1	ENST00000357105					
TREX1	ENST00000625293					
WDR6	?	ENSG00000178252	lung	2.56×10-11 (p-value)	GTEx Portal V6	75
SLC26A6	?	ENSG00000225697	lung	5.04×10-8 (p-value)	GTEx Portal V6	75
CELSR3	?	ENSG00000008300	lung	2.09×10-5 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	transformed fibroblasts	7.13×10-18 (p-value)	GTEx Portal V6	75
NCKIPSD	?	ENSG00000213672	transformed fibroblasts	5.36×10-9 (p-value)	GTEx Portal V6	73
CCDC36	?	ENSG00000173421	transformed fibroblasts	8.39×10-7 (p-value)	GTEx Portal V6	51
IP6K2	?	ENSG00000068745	transformed fibroblasts	1.54×10-7 (p-value)	GTEx Portal V6	65
RP11-3B7.1	?	ENSG00000225399	transformed fibroblasts	1.01×10-5 (p-value)	GTEx Portal V6	11
WDR6	?	ENSG00000178252	blood	8.00×10-12 (p-value)	GTEx Portal V6	75
NCKIPSD	?	ENSG00000213672	blood	1.10×10-5 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	tibial artery	4.82×10-9 (p-value)	GTEx Portal V6	75
TMEM89	?	ENSG00000183396	tibial artery	1.56×10-8 (p-value)	GTEx Portal V6	74

IP6K2	?	ENSG00000068745	tibial artery	6.46×10-7 (p-value)	GTEx Portal V6	54
NICN1	?	ENSG00000145029	tibial artery	5.27×10-5 (p-value)	GTEx Portal V6	2
WDR6	ENST00000615452	ILMN_1669484	monocyte	2.40×10-14 (p-value)	Fairfax et al.	13
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
UBA7	ENST00000497908	ILMN_1794612	b-cell	6.47×10-5 (p-value)	Fairfax et al.	8
UBA7	ENST00000333486					
MIR5193	ENST00000584510					
NCKIPSD	ENST00000294129	ILMN_1736623	monocyte	2.22×10-4 (p-value)	Fairfax et al.	4
NCKIPSD	ENST00000416649					
NCKIPSD	ENST00000470006					
USP4	ENST00000351842	ILMN_1798712	monocyte	1.21×10-9 (p-value)	Fairfax et al.	13
USP4	ENST00000483212		b-cell	1.64×10-6 (p-value)	Fairfax et al.	13
C3orf62	ENST00000343010					
USP4	ENST00000485450					
SLC26A6	ENST00000420764	ILMN_1785252	b-cell	3.59×10-5 (p-value)	Fairfax et al.	6
SLC26A6	ENST00000395550					
SLC26A6	ENST00000489483					
SLC26A6	ENST00000480524					
SLC26A6	ENST00000455886					
SLC26A6	ENST00000307364					
SLC26A6	ENST00000614797					
SLC26A6	ENST00000358747					
SLC26A6	ENST00000383733					
SLC26A6	ENST00000337000					
RBM6	ENST00000442092	ILMN_1738239	monocyte	1.47×10-5 (p-value)	Fairfax et al.	7
RBM6	ENST00000266022		b-cell	2.57×10-5 (p-value)	Fairfax et al.	7
RBM6	ENST00000454079					
RBM6	ENST00000422955					
RBM6	ENST00000419610					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
WDR6	?	ENSG00000178252	skeletal muscle	1.54×10-13 (p-value)	GTEx Portal V6	75
SLC26A6	?	ENSG00000225697	skeletal muscle	1.48×10-13 (p-value)	GTEx Portal V6	75
NCKIPSD	?	ENSG00000213672	skeletal muscle	1.01×10-5 (p-value)	GTEx Portal V6	20
SLC26A6	?	ENSG00000225697	EBV lymphocytes	1.46×10-6 (p-value)	GTEx Portal V6	63
WDR6	?	ENSG00000178252	EBV lymphocytes	9.15×10-7 (p-value)	GTEx Portal V6	43
WDR6	?	ENSG00000178252	sun exposed skin	1.46×10-35 (p-value)	GTEx Portal V6	75
WDR6	?	ENSG00000178252	unexposed skin	1.57×10-22 (p-value)	GTEx Portal V6	75
TMEM89	?	ENSG00000183396	aorta	2.36×10-5 (p-value)	GTEx Portal V6	7
IP6K2	?	ENSG00000068745	aorta	1.01×10-5 (p-value)	GTEx Portal V6	25

WDR6	?	ENSG00000178252	subcutaneous adipocytes	2.01×10-21 (p-value)	GTEx Portal V6	75
SLC26A6	?	ENSG00000225697	subcutaneous adipocytes	3.25×10-5 (p-value)	GTEx Portal V6	12
NCKIPSD	?	ENSG00000213672	visceral adipocytes	2.83×10-7 (p-value)	GTEx Portal V6	71
WDR6	?	ENSG00000178252	visceral adipocytes	1.53×10-7 (p-value)	GTEx Portal V6	55
WDR6	?	ENSG00000178252	cortex	5.42×10-7 (p-value)	GTEx Portal V6	68
WDR6	?	ENSG00000178252	tibial nerve	7.26×10-13 (p-value)	GTEx Portal V6	75
SLC26A6	?	ENSG00000225697	tibial nerve	4.59×10-5 (p-value)	GTEx Portal V6	10
WDR6	?	ENSG00000178252	esophagus mucosa	3.87×10-26 (p-value)	GTEx Portal V6	75
NCKIPSD	?	ENSG00000213672	esophagus mucosa	1.58×10-8 (p-value)	GTEx Portal V6	74
PFKFB4	ENST00000232375	ILMN_1653292	blood	6.11×10-5 (p-value)	Westra et al.	9
CCDC36	?	ENSG00000173421	testis	4.13×10-7 (p-value)	GTEx Portal V6	54
CCDC36	?	ENSG00000173421	muscularis mucosae	2.23×10-6 (p-value)	GTEx Portal V6	57
KLHDC8B	?	ENSG00000185909	muscularis mucosae	8.00×10-6 (p-value)	GTEx Portal V6	24
WDR6	?	ENSG00000178252	blood	2.09×10-3 (q-value)	SeeQTL DB (HapMap)	13
WDR6	ENST00000615452	ILMN_1669484	monocyte	5.52×10-35 (p-value)	Zeller et al.	7
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	monocyte	1.59×10-26 (p-value)	Zeller et al.	7
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
WDR6	?	ENSG00000178252	breast	3.91×10-6 (p-value)	GTEx Portal V6	25
SLC26A6	?	ENSG00000225697	thyroid	1.41×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	stomach	9.03×10-7 (p-value)	GTEx Portal V6	32
CCDC71	?	ENSG00000177352	cerebellar hemisphere	5.33×10-6 (p-value)	GTEx Portal V6	36
WDR6	?	ENSG00000178252	pancreas	3.37×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000338496	1	ENCP00000036385	SHISA5
		ENCP00000036431	ARIH2
		ENCP00000036356	ZNF589
		ENCP00000036451	DALRD3
		ENCP00000036473	QARS
		ENCP00000036376	CCDC51

		ENCP00000036448	WDR6
		ENCP00000036353	CDC25A
ENCE00000338642	1	ENCP00000036430	SLC25A20
		ENCP00000036466	QRICH1
		ENCP00000036373	PLXNB1
		ENCP00000036398	COL7A1 COL7A1
		ENCP00000036399	COL7A1 COL7A1
		ENCP00000036419	NCKIPSD
		ENCP00000036367	PLXNB1
		ENCP00000036396	COL7A1 COL7A1
ENCE00000338674	1	ENCP00000036410	SLC26A6
		ENCP00000036434	ARIH2
		ENCP00000036384	SHISA5
ENCE00000338847	1	ENCP00000036433	ARIH2
ENCE00000338585	1	ENCP00000036360	SPINK8
		ENCP00000036439	ARIH2
		ENCP00000036380	ATRIP
		ENCP00000036426	IP6K2
ENCE00000338754	1	ENCP00000036427	PRKAR2A
		ENCP00000036409	SLC26A6

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363494 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, Rad21, TAF7, Sin3Ak20, H3K9ac, CTCF, TAF1, H3K4me2, SP1, H3K4me3, H3K27ac
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	H3K9ac, H2AZ, PolII, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	DNase1, H3K4me2, H3K9ac, H3K27ac, CTCF, H3K4me3, H3K27me3, H3K36me3, H3K4me1, TAF1, PolII, H3K79me2, Rad21
		blood (GM12878)	PolII, H2AZ, DNase1, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac
		lung (IMR90)	CTCF, DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K4me3, H3K4me2, H3K9ac
		NHLF	H3K27ac, H3K9ac, CTCF, DNase1, H3K4me3
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K4me2, H3K9ac, H3K36me3, H3K4me3, DNase1
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1
ENSR00001363497 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me1, H3K4me2, H3K9ac
		lung (IMR90)	H3K36me3
ENSR00001674778 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Egr1, H3K9ac, CTCF, TAF1, H3K4me2, H3K4me3
		HSMMtube	H3K27me3
		skin (NHDF-AD)	CTCF, H3K4me2
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, CTCF, H3K4me3
		lung (IMR90)	CTCF
		blood (GM12878)	DNase1, H3K4me3, CTCF
		nervous (NH-A)	DNase1, H3K4me2
		skin (NHEK)	DNase1, CTCF, H3K4me3
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2

		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, CTCF, DNase1
		A549	H3K4me3, DNase1
ENSR00001674779 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K9ac, H3K4me3, H3K27me3, PolII, Jund, TAF7, p300, Nrsf, Sin3Ak20, Yy1, HDAC2, TAF1, H3K4me2, DNase1
		HSMMtube	H3K4me2, H2AZ, H3K27me3
		blood (K562)	DNase1, Nrsf, PolII, Sin3Ak20, Ini1, Max
		skin (NHDF-AD)	DNase1, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	Yy1, Nrsf, Sin3Ak20, PolII, TAF1, H3K9ac, H3K4me3, DNase1
		lung (IMR90)	H3K4me3, H3K27me3, H3K4me2
		blood (GM12878)	DNase1, Sin3Ak20, Nrsf, PolII, Yy1, H3K4me3
		nervous (NH-A)	H3K4me2
		skin (NHEK)	H3K9ac, H3K4me3, H3K27me3, DNase1
		NHLF	H3K27me3
		Osteobl	H3K27me3, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	Ini1, Nrsf, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27me3
		endothelium (HUVEC)	Max, H3K4me3, H3K27me3, DNase1
		A549	H3K4me3, H3K9ac, DNase1
ENSR00001478981 (promoter flanking region)	3	embryonic stem cell (H1ESC)	PolII, H3K9ac, H3K27ac, H3K4me2, H3K36me3, DNase1, H3K27me3
		HSMMtube	H3K79me2, H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H3K79me2, H3K27ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3, H3K27ac, DNase1
		blood (GM12878)	PolII, H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K27ac, H3K4me1
		NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, H3K4me1, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K4me1, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001363512 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3, H3K79me2
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	H3K79me2, CTCF, H3K36me3
		blood (GM12878)	H3K36me3, H3K79me2
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001363515 (promoter flanking region)	1	NHLF	H3K4me1, H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1, DNase1
		muscle (HSMM)	H3K79me2, H3K36me3
		breast (HMEC)	H3K4me2, H3K4me1, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K36me3, H3K79me2

		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K79me2, H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H4K20me1, H3K79me2, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K4me1, DNase1, H3K36me3, H3K4me2, H3K27ac
ENSR00001363517	2	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, TAF7, H3K27ac, Yy1, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3
(promoter flanking region)		HSMMtube	DNase1, H3K79me2, H3K9ac, H3K4me2, H3K4me3, H3K36me3
		blood (K562)	HEY1, H2AZ, TAF1, ELF1, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, H3K79me2, H3K9ac, Yy1, H3K27ac, Max
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		liver (HepG2)	DNase1, H3K36me3, H3K27me3, H3K4me3, H3K27ac, Gabp, Yy1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, DNase1, H3K79me2
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K27ac, H3K36me3, H3K9ac, DNase1, H3K4me3
		Osteobl	H3K27me3, H3K36me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	DNase1, H3K36me3, H3K27me3, PolII, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC141002.1	upstream gene variant	1996	ENST00000619404	?	?	4
CELSR3	downstream gene variant, upstream gene variant	1483	ENST00000470999	?	?	6
CELSR3	downstream gene variant, upstream gene variant	1568	ENST00000164024	NM_001407.2	ENSP00000164024	8
CELSR3	downstream gene variant, upstream gene variant	1295	ENST00000498057	?	?	7
CELSR3	downstream gene variant, upstream gene variant	1295	ENST00000461362	?	?	7
CELSR3-AS1	upstream gene variant, downstream gene variant	1690	ENST00000421275	?	?	7
IP6K2	downstream gene variant	1429	ENST00000433104	?	ENSP00000396904	1
IP6K2	downstream gene variant, upstream gene variant	122	ENST00000424035	?	ENSP00000406357	6
IP6K2	downstream gene variant, upstream gene variant	252	ENST00000443964	?	ENSP00000410950	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000446860	NM_001190317.1	ENSP00000399052	4
IP6K2	downstream gene variant, upstream gene variant	241	ENST00000416707	?	ENSP00000387759	4
IP6K2	downstream gene variant, upstream gene variant	256	ENST00000454335	?	ENSP00000412121	4
IP6K2	downstream gene variant, upstream gene variant	1957	ENST00000450045	?	ENSP00000394488	4
IP6K2	downstream gene variant, upstream gene variant	256	ENST00000436134	?	?	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000443853	?	ENSP00000389761	4
IP6K2	downstream gene variant, upstream gene variant	252	ENST00000449610	?	ENSP00000393077	4
IP6K2	downstream gene variant, upstream gene variant	228	ENST00000340879	NM_001005910.2, NM_001005911.2	ENSP00000341925	4
IP6K2	downstream gene variant, upstream gene variant	228	ENST00000431721	NM_001190316.1	ENSP00000414139	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000449563	?	ENSP00000411776	4
IP6K2	downstream gene variant, upstream gene	195	ENST00000412795	?	ENSP00000389755	4

	variant					
IP6K2	downstream gene variant, upstream gene variant	81	ENST00000440424	?	ENSP00000393797	6
IP6K2	downstream gene variant, upstream gene variant	278	ENST00000413654	?	ENSP00000414428	4
IP6K2	downstream gene variant, upstream gene variant	83	ENST00000434860	?	ENSP00000388420	6
IP6K2	downstream gene variant, upstream gene variant	166	ENST00000432678	NM_001146178.2, NM_001146179.2	ENSP00000400812	4
IP6K2	downstream gene variant, upstream gene variant	229	ENST00000412850	?	ENSP00000395819	4
IP6K2	downstream gene variant, upstream gene variant	251	ENST00000476645	?	?	8
IP6K2	downstream gene variant, upstream gene variant	1915	ENST00000455545	?	ENSP00000410454	4
IP6K2	downstream gene variant, upstream gene variant	238	ENST00000417896	?	ENSP00000388116	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000437427	?	ENSP00000403968	4
IP6K2	downstream gene variant, upstream gene variant	194	ENST00000328631	NM_001005909.2, NM_016291.3	ENSP00000331103	4
IP6K2	downstream gene variant, upstream gene variant	82	ENST00000453202	?	ENSP00000387394	5
IP6K2	downstream gene variant, upstream gene variant	1455	ENST00000491686	?	?	5
IP6K2	downstream gene variant, upstream gene variant	234	ENST00000413298	?	ENSP00000396203	4
IP6K2	downstream gene variant, upstream gene variant	486	ENST00000479914	?	?	4
MIR4793	downstream gene variant, upstream gene variant	574	ENST00000577502	?	?	4
MIR6824	downstream gene variant, upstream gene variant	242	ENST00000611478	?	?	14
NCKIPSD	downstream gene variant, upstream gene variant	671	ENST00000294129	NM_016453.3	ENSP00000294129	6
NCKIPSD	downstream gene variant, upstream gene variant	577	ENST00000453349	?	ENSP00000408588	7
NCKIPSD	downstream gene variant, upstream gene variant	573	ENST00000454134	?	ENSP00000416144	7
NCKIPSD	downstream gene variant, upstream gene variant	577	ENST00000426678	?	ENSP00000416904	7
NCKIPSD	downstream gene variant, upstream gene variant	236	ENST00000439518	?	ENSP00000409675	7
NCKIPSD	downstream gene variant, upstream gene variant	671	ENST00000416649	NM_184231.2	ENSP00000389059	6
NCKIPSD	downstream gene variant, upstream gene variant	1106	ENST00000470006	?	?	10
NCKIPSD	downstream gene variant, upstream gene variant	436	ENST00000415281	?	ENSP00000406442	8
NCKIPSD	downstream gene variant, upstream gene variant	1193	ENST00000413374	?	ENSP00000396683	10
RP11-572O6.1	upstream gene variant, downstream gene variant	359	ENST00000607025	?	?	12
SLC26A6	downstream gene variant, upstream gene variant	14	ENST00000431213	?	ENSP00000387532	9
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000337000	NM_001281733.1	ENSP00000337648	6
SLC26A6	downstream gene variant, upstream gene variant	722	ENST00000494717	?	?	9
SLC26A6	downstream gene variant, upstream gene variant	973	ENST00000414944	?	ENSP00000388721	8
SLC26A6	downstream gene variant, upstream gene variant	614	ENST00000485361	?	?	9
SLC26A6	downstream gene variant, upstream gene variant	1212	ENST00000469693	?	?	8
SLC26A6	downstream gene variant, upstream gene variant	314	ENST00000482282	?	?	11
SLC26A6	downstream gene variant, upstream gene variant	962	ENST00000444531	?	ENSP00000404607	8
SLC26A6	downstream gene variant, upstream gene variant	5	ENST00000426599	?	ENSP00000405872	9
SLC26A6	downstream gene variant, upstream gene variant	425	ENST00000421649	?	ENSP00000389922	8

SLC26A6	downstream gene variant, upstream gene variant	977	ENST00000431739	?	ENSP00000401813	8
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000395550	NM_022911.2	ENSP00000378920	6
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000383733	NM_134426.2	ENSP00000373239	6
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000358747	NM_001040454.1	ENSP00000351597	9
SLC26A6	downstream gene variant, upstream gene variant	578	ENST00000466257	?	?	9
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000307364	?	ENSP00000307089	6
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000614797	?	ENSP00000483669	6
SLC26A6	downstream gene variant, upstream gene variant	133	ENST00000455886	NM_001281732.1	ENSP00000401066	6
SLC26A6	downstream gene variant, upstream gene variant	74	ENST00000489483	?	?	8
SLC26A6	downstream gene variant, upstream gene variant	114	ENST00000480524	?	?	9
SLC26A6	downstream gene variant, upstream gene variant	667	ENST00000496469	?	?	8
SLC26A6	downstream gene variant, upstream gene variant	1206	ENST00000462009	?	?	8
SLC26A6	downstream gene variant, upstream gene variant	110	ENST00000420764	NM_134263.2	ENSP00000404684	6
TMEM89	upstream gene variant	2697	ENST00000330862	NM_001008269.1	ENSP00000329557	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CELSR3	ENST00000164024	NM_001407.2	ENSP00000164024	3	3	3

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
SLC26A6	ENST00000358747	NM_001040454.1	ENSP00000351597	1
SLC26A6	ENST00000395550	NM_022911.2	ENSP00000378920	1
SLC26A6	ENST00000431739	?	ENSP00000401813	1
SLC26A6	ENST00000489483	?	?	1
SLC26A6	ENST00000426599	?	ENSP00000405872	1
SLC26A6	ENST00000482282	?	?	1
SLC26A6	ENST00000455886	NM_001281732.1	ENSP00000401066	1
SLC26A6	ENST00000337000	NM_001281733.1	ENSP00000337648	1
SLC26A6	ENST00000614797	?	ENSP00000483669	1
SLC26A6	ENST00000307364	?	ENSP00000307089	1
SLC26A6	ENST00000480524	?	?	1
SLC26A6	ENST00000383733	NM_134426.2	ENSP00000373239	1
SLC26A6	ENST00000420764	NM_134263.2	ENSP00000404684	1

5'-UTR variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
NCKIPSD	ENST00000426678	?	ENSP00000416904	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CELSR3	ENST00000164024	NM_001407.2	ENSP00000164024	11
CELSR3	ENST00000470999	?	?	2
CELSR3	ENST00000498057	?	?	5
CELSR3	ENST00000461362	?	?	5
CELSR3-AS1	ENST00000421275	?	?	3
IP6K2	ENST00000449563	?	ENSP00000411776	18
IP6K2	ENST00000455545	?	ENSP00000410454	18
IP6K2	ENST00000436134	?	?	18
IP6K2	ENST00000440424	?	ENSP00000393797	16
IP6K2	ENST00000446860	NM_001190317.1	ENSP00000399052	17
IP6K2	ENST00000454335	?	ENSP00000412121	18
IP6K2	ENST00000433104	?	ENSP00000396904	23
IP6K2	ENST00000413654	?	ENSP00000414428	18
IP6K2	ENST00000443964	?	ENSP00000410950	17
IP6K2	ENST00000432678	NM_001146178.2, NM_001146179.2	ENSP00000400812	18
IP6K2	ENST00000416707	?	ENSP00000387759	18
IP6K2	ENST00000449610	?	ENSP00000393077	17
IP6K2	ENST00000450045	?	ENSP00000394488	18
IP6K2	ENST00000431721	NM_001190316.1	ENSP00000414139	17
IP6K2	ENST00000434860	?	ENSP00000388420	16
IP6K2	ENST00000437427	?	ENSP00000403968	18
IP6K2	ENST00000340879	NM_001005910.2, NM_001005911.2	ENSP00000341925	18
IP6K2	ENST00000424035	?	ENSP00000406357	16
IP6K2	ENST00000417896	?	ENSP00000388116	17
IP6K2	ENST00000476645	?	?	13
IP6K2	ENST00000443853	?	ENSP00000389761	18
IP6K2	ENST00000412795	?	ENSP00000389755	18
IP6K2	ENST00000413298	?	ENSP00000396203	18
IP6K2	ENST00000491686	?	?	1
IP6K2	ENST00000328631	NM_001005909.2, NM_016291.3	ENSP00000331103	19
IP6K2	ENST00000412850	?	ENSP00000395819	18
IP6K2	ENST00000479914	?	?	3
NCKIPSD	ENST00000426678	?	ENSP00000416904	2
NCKIPSD	ENST00000470006	?	?	6
NCKIPSD	ENST00000413374	?	ENSP00000396683	6
NCKIPSD	ENST00000453349	?	ENSP00000408588	2
NCKIPSD	ENST00000416649	NM_184231.2	ENSP00000389059	11
NCKIPSD	ENST00000294129	NM_016453.3	ENSP00000294129	11
NCKIPSD	ENST00000454134	?	ENSP00000416144	2
NCKIPSD	ENST00000439518	?	ENSP00000409675	2
NCKIPSD	ENST00000415281	?	ENSP00000406442	3

SLC26A6	ENST00000358747	NM_001040454.1	ENSP00000351597	5
SLC26A6	ENST00000395550	NM_022911.2	ENSP00000378920	8
SLC26A6	ENST00000431739	?	ENSP00000401813	4
SLC26A6	ENST00000489483	?	?	7
SLC26A6	ENST00000426599	?	ENSP00000405872	4
SLC26A6	ENST00000469693	?	?	3
SLC26A6	ENST00000482282	?	?	2
SLC26A6	ENST00000455886	NM_001281732.1	ENSP00000401066	8
SLC26A6	ENST00000431213	?	ENSP00000387532	2
SLC26A6	ENST00000462009	?	?	3
SLC26A6	ENST00000337000	NM_001281733.1	ENSP00000337648	9
SLC26A6	ENST00000614797	?	ENSP00000483669	8
SLC26A6	ENST00000444531	?	ENSP00000404607	2
SLC26A6	ENST00000307364	?	ENSP00000307089	9
SLC26A6	ENST00000414944	?	ENSP00000388721	2
SLC26A6	ENST00000480524	?	?	6
SLC26A6	ENST00000421649	?	ENSP00000389922	2
SLC26A6	ENST00000496469	?	?	4
SLC26A6	ENST00000383733	NM_134426.2	ENSP00000373239	8
SLC26A6	ENST00000420764	NM_134263.2	ENSP00000404684	8

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CELSR3	ENST00000164024	NM_001407.2	ENSP00000164024	1
IP6K2	ENST00000446860	NM_001190317.1	ENSP00000399052	1
IP6K2	ENST00000433104	?	ENSP00000396904	1
IP6K2	ENST00000443964	?	ENSP00000410950	1
IP6K2	ENST00000416707	?	ENSP00000387759	1
IP6K2	ENST00000449610	?	ENSP00000393077	1
IP6K2	ENST00000431721	NM_001190316.1	ENSP00000414139	1
IP6K2	ENST00000417896	?	ENSP00000388116	1
IP6K2	ENST00000412795	?	ENSP00000389755	1
SLC26A6	ENST00000431213	?	ENSP00000387532	1
SLC26A6	ENST00000307364	?	ENSP00000307089	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IP6K2	ENST00000433104	?	ENSP00000396904	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CELSR3	ENST00000498057	?	3
CELSR3	ENST00000461362	?	3
IP6K2	ENST00000436134	?	1

SLC26A6	ENST00000489483	?	4
SLC26A6	ENST00000482282	?	2
SLC26A6	ENST00000485361	?	1
SLC26A6	ENST00000494717	?	1
SLC26A6	ENST00000480524	?	3

Locus 131 (rs17114699)

SNiPAcard

Block annotations

Block info

genomic range	chr14:21,158,910-21,158,910
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.017	gene(s) hit or close-by	ANG, RNASE4, RP11-903H12.3, RP11-903H12.5
phastCons	0	eQTL gene(s)	RNASE9
GERP++	-0.0912	potentially regulated gene(s)	–
CADD score	1.825	disease gene(s)	ANG

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ANG	AMYOTROPHIC LATERAL SCLEROSIS 9	OMIM	MIM:611895
ANG	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RNASE9	ENST00000404716	ILMN_1655474	monocyte	1.81×10^{-4} (p-value)	Fairfax et al.	1
RNASE9	ENST00000429244					
RNASE9	ENST00000338904					
RNASE9	ENST00000611135					
RNASE9	ENST00000556208					
RNASE9	ENST00000555230					
RNASE9	ENST00000553706					
RNASE9	ENST00000553541					
RNASE9	ENST00000557209					
RNASE9	ENST00000557068					
RNASE9	ENST00000554964					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001621075 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	H4K20me1, H3K79me2, Rad21, ELF1, H3K4me1, CTCF, H3K36me3
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF

skin (NHEK)	CTCF, DNase1
NHLF	CTCF
Osteobl	CTCF
blood (DND-41)	CTCF
breast (HMEC)	CTCF
cervix (HeLa-S3)	DNase1, CTCF
monocytes (Monocytes-CD14+)	DNase1, CTCF
endothelium (HUVEC)	CTCF
A549	DNase1, CTCF, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-903H12.3	downstream gene variant	2729	ENST00000554286	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANG	ENST00000336811	NM_001145.4	ENSP00000336762	1
ANG	ENST00000554073	?	?	1
ANG	ENST00000397990	NM_001097577.2	ENSP00000381077	1
RNASE4	ENST00000397995	?	ENSP00000381081	1
RNASE4	ENST00000555597	NM_194431.2	ENSP00000451624	1
RNASE4	ENST00000555835	NM_001282193.1, NM_001282192.1, NM_002937.4	ENSP00000452245	1
RP11-903H12.5	ENST00000553909	?	ENSP00000477037	1

Locus 132 (rs2736186)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,572,333-31,572,333
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.269	**gene(s) hit or close-by**	–
phastCons	0	**eQTL gene(s)**	XXbac-BPG181B23.7
GERP++	-1.97	**potentially regulated gene(s)**	–
CADD score	0.854	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG181B23.7	?	ENSG00000272221	skeletal muscle	3.60×10^{-6} (p-value)	GTEx Portal V6	1

Locus 133 (rs1800973)

SNiPAcard

Block annotations

Block info

genomic range	chr12:69,711,929-69,744,014
block size	32,086 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.680 [-1.08 – 2.441]	gene(s) hit or close-by	LYZ, RP11-1143G9.4
phastCons	μ = 0.538 [0.095 – 0.981]	eQTL gene(s)	–
GERP++	μ = 2.821 [-0.298 – 5.94]	potentially regulated gene(s)	LYZ
CADD score	μ = 8.722 [0.794 – 16.65]	disease gene(s)	LYZ

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM060322	1

Disease gene annotation

gene	trait	source DB	source entry/link
LYZ	AMYLOIDOSIS, FAMILIAL VISCERAL	OMIM	MIM:105200
LYZ	Familial renal amyloidosis due to lysozyme variant	OrphaNet	OrphaNet:93561

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
LYZ	missense variant	ENST00000548839	?	ENSP00000449969	N/T	aAc/aCc	?	?	1
LYZ	missense variant	ENST00000549690	?	ENSP00000449898	N/T	aAc/aCc	?	?	1
LYZ	missense variant	ENST00000261267	NM_000239.2	ENSP00000261267	N/T	aAc/aCc	?	?	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000127685	1	ENST00000549690, ENST00000261267	LYZ

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000653867 (promoter flanking region)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H2AZ, PU1, DNase1
		blood (DND-41)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3

	liver (HepG2)	H3K4me3, PolII, H3K79me2, DNase1, TAF1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K36me3
	lung (IMR90)	H3K27me3
	A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-1143G9.4	downstream gene variant	3259	ENST00000548900	?	?	1

Locus 134 (rs6714900)

SNiPAcard

Block annotations

Block info

genomic range	chr2:71,292,522-71,327,352
block size	34,831 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.943 [-2.25 – 0.896]	gene(s) hit or close-by	AC007040.8, NAGK, RP11-467P9.1
phastCons	μ = 0.011 [0 – 0.061]	eQTL gene(s)	MCEE, NAGK, OR7E62P, OR7E91P
GERP++	μ = -0.097 [-2.23 – 1.49]	potentially regulated gene(s)	AC007040.6, AC007040.8, CD207, MPHOSPH10, NAGK, ZNF638
CADD score	μ = 3.759 [0.492 – 7.392]	disease gene(s)	MCEE, CD207

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
MCEE	METHYLMALONYL-CoA EPIMERASE DEFICIENCY	DECIPHER	MIM:251120
MCEE	METHYLMALONYL-CoA EPIMERASE DEFICIENCY	OMIM	MIM:251120
CD207	BIRBECK GRANULE DEFICIENCY	OMIM	MIM:613393
MCEE	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	OrphaNet	OrphaNet:308425

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NAGK	?	ENSG00000124357	muscularis mucosae	2.31×10^{-7} (p-value)	GTEx Portal V6	3
NAGK	?	ENSG00000124357	lung	8.00×10^{-13} (p-value)	GTEx Portal V6	8
NAGK	?	ENSG00000124357	spleen	1.41×10^{-7} (p-value)	GTEx Portal V6	2
NAGK	?	ENSG00000124357	tibial nerve	1.12×10^{-8} (p-value)	GTEx Portal V6	8
NAGK	?	ENSG00000124357	blood	8.69×10^{-24} (p-value)	GTEx Portal V6	8
MCEE	?	ENSG00000124370	esophagus mucosa	4.93×10^{-8} (p-value)	GTEx Portal V6	8
OR7E91P	?	ENSG00000205847	esophagus mucosa	2.33×10^{-8} (p-value)	GTEx Portal V6	8
NAGK	?	ENSG00000124357	esophagus mucosa	8.14×10^{-29} (p-value)	GTEx Portal V6	8
NAGK	?	ENSG00000124357	sun exposed skin	1.07×10^{-9} (p-value)	GTEx Portal V6	8
OR7E62P	?	ENSG00000234896	sun exposed skin	2.09×10^{-6} (p-value)	GTEx Portal V6	1
NAGK	?	ENSG00000124357	subcutaneous adipocytes	6.67×10^{-7} (p-value)	GTEx Portal V6	1
MCEE	ENST00000413592	ILMN_1735347	blood	2.66×10^{-6} (p-value)	Westra et al.	3
MCEE	ENST00000244217					
MCEE	ENST00000462609					
NAGK	?	ENSG00000124357	EBV lymphocytes	2.53×10^{-6} (p-value)	GTEx Portal V6	1
NAGK	?	ENSG00000124357	blood	2.95×10^{-3} (q-value)	SeeQTL DB (HapMap)	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000251577	1	ENCP00000026542	MPHOSPH10
		ENCP00000026554	ZNF638
		ENCP00000026534	NAGK
		ENCP00000026508	CD207
		ENCP00000026522	AC007040.6
		ENCP00000026535	NAGK
		ENCP00000026528	AC007040.8
		ENCP00000026553	ZNF638
		ENCP00000026540	MPHOSPH10
		ENCP00000026541	MPHOSPH10

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000592557 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF, Yy1
		HSMMtube	CTCF
		blood (K562)	Rad21, H2AZ, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Yy1, Rad21, CTCF, DNase1
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K4me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC007040.8	upstream gene variant	649	ENST00000434990	?	?	1
NAGK	upstream gene variant	2916	ENST00000531934	?	ENSP00000436326	1
NAGK	upstream gene variant	2909	ENST00000455662	?	ENSP00000389087	1
NAGK	upstream gene variant	3239	ENST00000493102	?	?	1
NAGK	upstream gene variant	3586	ENST00000489309	?	?	1
NAGK	upstream gene variant	3258	ENST00000480411	?	?	1
NAGK	upstream gene variant	2886	ENST00000613852	NM_017567.4	ENSP00000477639	1
NAGK	upstream gene variant	3255	ENST00000450272	?	ENSP00000400941	1
NAGK	upstream gene variant	3258	ENST00000465105	?	?	1
NAGK	upstream gene variant	3230	ENST00000484984	?	?	1
NAGK	upstream gene variant	3196	ENST00000418807	?	ENSP00000396070	1
NAGK	upstream gene variant	2924	ENST00000455197	?	ENSP00000408550	1
NAGK	upstream gene variant	3258	ENST00000475709	?	?	1

NAGK	upstream gene variant	3258	ENST00000475709	?	?	1
NAGK	upstream gene variant, downstream gene variant	233	ENST00000497690	?	?	2
NAGK	upstream gene variant	4908	ENST00000464638	?	?	1
NAGK	upstream gene variant	3230	ENST00000244204	?	ENSP00000244204	1
NAGK	upstream gene variant	3273	ENST00000472519	?	?	1
NAGK	upstream gene variant	3227	ENST00000443938	?	ENSP00000411846	1
NAGK	upstream gene variant	1433	ENST00000478659	?	?	1
NAGK	upstream gene variant	3270	ENST00000533981	?	ENSP00000435207	1
NAGK	upstream gene variant	3200	ENST00000443872	?	ENSP00000404531	1
NAGK	upstream gene variant	939	ENST00000529236	?	ENSP00000431204	1
NAGK	upstream gene variant	4564	ENST00000468601	?	?	1
NAGK	upstream gene variant	2685	ENST00000524537	?	ENSP00000437192	1
NAGK	downstream gene variant	2471	ENST00000531799	?	?	1
NAGK	upstream gene variant	3768	ENST00000490998	?	?	1
NAGK	upstream gene variant	697	ENST00000479854	?	?	1
RP11-467P9.1	downstream gene variant, upstream gene variant	2127	ENST00000608897	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NAGK	ENST00000464638	?	?	1
NAGK	ENST00000455197	?	ENSP00000408550	1
NAGK	ENST00000524736	?	ENSP00000435437	1
NAGK	ENST00000613852	NM_017567.4	ENSP00000477639	1
NAGK	ENST00000533981	?	ENSP00000435207	1
NAGK	ENST00000493102	?	?	1
NAGK	ENST00000443938	?	ENSP00000411846	1
NAGK	ENST00000484984	?	?	1
NAGK	ENST00000531799	?	?	1
NAGK	ENST00000450272	?	ENSP00000400941	1
NAGK	ENST00000475709	?	?	1
NAGK	ENST00000418807	?	ENSP00000396070	1
NAGK	ENST00000428360	?	?	1
NAGK	ENST00000244204	?	ENSP00000244204	1
NAGK	ENST00000531934	?	ENSP00000436326	1
NAGK	ENST00000472519	?	?	1
NAGK	ENST00000455662	?	ENSP00000389087	1
NAGK	ENST00000443872	?	ENSP00000404531	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NAGK	ENST00000480411	?	1

NAGK	ENST00000480411	?	1
NAGK	ENST00000465105	?	1
NAGK	ENST00000489309	?	1

Locus 135 (rs3792900)

SNiPAcard

Block annotations

Block info

genomic range	chr5:135,362,550-135,400,035
block size	37,486 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.648 [-4.435 – 1]	gene(s) hit or close-by	TGFBI
phastCons	μ = 0.003 [0 – 0.03]	eQTL gene(s)	AC009014.3, SMAD5, TGFBI, TRPC7
GERP++	μ = -2.821 [-9.26 – 3.43]	potentially regulated gene(s)	CTC-321K16.4, TGFBI
CADD score	μ = 3.253 [0.008 – 10.2]	disease gene(s)	TGFBI

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Response to cytidine analogues (gemcitabine)	$<4.00\times10^{-6}$	GWAS Catalog	24483146	1

Disease gene annotation

gene	trait	source DB	source entry/link
TGFBI	CORNEAL DYSTROPHY, AVELLINO TYPE	OMIM	MIM:607541
TGFBI	CORNEAL DYSTROPHY OF BOWMAN LAYER, TYPE II	OMIM	MIM:602082
TGFBI	CORNEAL DYSTROPHY, LATTICE TYPE IIIA	OMIM	MIM:608471
TGFBI	CORNEAL DYSTROPHY, GROENOUW TYPE I	OMIM	MIM:121900
TGFBI	CORNEAL DYSTROPHY OF BOWMAN LAYER, TYPE I	OMIM	MIM:608470
TGFBI	CORNEAL DYSTROPHY, LATTICE TYPE I	OMIM	MIM:122200
TGFBI	CORNEAL DYSTROPHY, EPITHELIAL BASEMENT MEMBRANE	OMIM	MIM:121820
TGFBI	Granular corneal dystrophy type I	OrphaNet	OrphaNet:98962
TGFBI	Lattice corneal dystrophy Type I	OrphaNet	OrphaNet:98964
TGFBI	Microcystic corneal dystrophy	OrphaNet	OrphaNet:98956
TGFBI	Granular corneal dystrophy type II	OrphaNet	OrphaNet:98963
TGFBI	Thiel-Behnke corneal dystrophy	OrphaNet	OrphaNet:98960
TGFBI	Reis-Bucklers corneal dystrophy	OrphaNet	OrphaNet:98961

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TGFBI	ENST00000506699	ILMN_1663866	blood	7.17×10^{-7} (p-value)	Westra et al.	17
TGFBI	ENST00000508076					
TGFBI	ENST00000504411					
TGFBI	ENST00000514554					
TGFBI	ENST00000442011					
TGFBI	ENST00000507018					
SMAD5	ENST00000545620	ILMN_2371984	blood	7.87×10^{-5} (p-value)	Westra et al.	2
SMAD5	ENST00000545279					
SMAD5	ENST00000545620	ILMN_1674551	monocyte	1.36×10^{-5} (p-value)	Fairfax et al.	2

SMAD5	ENST00000545279		b-cell	3.19×10-4 (p-value)	Fairfax et al.	2
TRPC7	ENST00000378459	ILMN_1683909	b-cell	3.95×10-4 (p-value)	Fairfax et al.	2
TRPC7	ENST00000509288					
TRPC7	ENST00000513104					
TRPC7	ENST00000502753					
TRPC7	ENST00000352189					
TRPC7	ENST00000514963					
TRPC7	ENST00000503275					
AC009014.3	?	ENSG00000271824	testis	7.88×10-6 (p-value)	GTEx Portal V6	1
SMAD5	ENST00000545279	ILMN_1674551	monocyte	4.13×10-14 (p-value)	Zeller et al.	2
SMAD5	ENST00000545620					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000671130	1	ENST00000509749, ENST00000508767	TGFBI
FFCP00000671129	1	ENST00000509749, ENST00000508767	TGFBI

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000423745	1	ENCP00000045720	CTC-321K16.4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001290929 (promoter flanking region)	4	NHLF	DNase1, H3K36me3
		embryonic stem cell (H1ESC)	H3K27me3, DNase1
		Osteobl	H3K36me3, H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	DNase1, H3K4me1
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K9ac, H3K36me3, DNase1
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001290932 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27ac, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me1, H3K27ac, H3K4me2, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K36me3, Cmyc, FOSL2, Jund, BHLHE40, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac
		lung (IMR90)	DNase1, H4K8ac, H3K18ac, H3K27ac, H3K36me3
		nervous (NH-A)	H3K9ac, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K4me1
		NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	Cjun, Jund, H3K27ac, H3K4me1, Max, Cfos, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3

		endothelium (HUVEC)	Cjun, DNase1
		A549	H3K4me3, H3K4me1, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001290934 (promoter flanking region)	5	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	CTCF, H3K27ac, H3K36me3, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K36me3, H3K4me1, CTCF, DNase1, H3K27ac
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1, CTCF
		liver (HepG2)	FOSL2, Jund, FOXA2, DNase1, H3K4me1, p300, FOXA1, H3K27ac, CTCF, Cmyc, H3K36me3
		lung (IMR90)	DNase1, H3K4me1, H3K18ac, H3K27ac, H3K36me3, H3K4ac, CTCF
		nervous (NH-A)	DNase1, CTCF, H3K4me2, H3K27ac, H3K36me3, H3K9ac
		skin (NHEK)	CTCF, H3K36me3
		NHLF	DNase1, H3K4me1, CTCF, H3K27ac, H3K36me3
		Osteobl	H3K36me3, CTCF, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K36me3, CTCF
		cervix (HeLa-S3)	DNase1, Cjun, Jund, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	Cjun, CTCF
		A549	H3K4me3, CTCF, DNase1, H3K27ac, H3K36me3
ENSR00001697850 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27ac, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me1, CTCF, DNase1, H3K27ac
		muscle (HSMM)	DNase1, CTCF, H3K27ac, H3K36me3
		liver (HepG2)	DNase1, H3K4me1, p300, FOXA1, H3K27ac, CTCF, Jund
		lung (IMR90)	CTCF, H3K4ac, H3K36me3, DNase1, H3K18ac, H3K27ac
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	CTCF, H3K36me3
		NHLF	H3K27ac, CTCF, DNase1
		Osteobl	H3K36me3, CTCF, H3K27ac
		breast (HMEC)	H3K36me3, CTCF
		cervix (HeLa-S3)	Cjun, Jund, CTCF, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	CTCF
		A549	H3K4me3, CTCF, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
TGFBI	upstream gene variant, downstream gene variant	802	ENST00000504185	?	?	6
TGFBI	upstream gene variant, downstream gene variant	530	ENST00000506699	?	?	5
TGFBI	upstream gene variant, downstream gene variant	531	ENST00000508076	?	ENSP00000423935	3
TGFBI	upstream gene variant, downstream gene variant	529	ENST00000442011	NM_000358.2	ENSP00000416330	5
TGFBI	upstream gene variant, downstream gene variant	199	ENST00000509485	?	ENSP00000426589	12
TGFBI	upstream gene variant, downstream gene variant	681	ENST00000507018	?	ENSP00000421540	5
TGFBI	upstream gene variant, downstream gene variant	974	ENST00000503087	?	ENSP00000427137	3
TGFBI	upstream gene variant, downstream gene variant	2260	ENST00000515433	?	?	11
TGFBI	upstream gene variant, downstream gene variant	118	ENST00000604555	?	ENSP00000474155	8
TGFBI	upstream gene variant, downstream gene variant	53	ENST00000509749	?	?	2
TGFBI	upstream gene variant, downstream gene variant	300	ENST00000514242	?	?	12
TGFBI	upstream gene variant, downstream gene variant	24	ENST00000508767	?	ENSP00000423871	5
TGFBI	upstream gene variant, downstream gene variant	210	ENST00000504411	?	?	13
TGFBI	upstream gene variant, downstream gene variant	528	ENST00000514554	?	ENSP00000421440	3
TGFBI	upstream gene variant, downstream gene variant	702	ENST00000513497	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TGFBI	ENST00000442011	NM_000358.2	ENSP00000416330	L	ctC/ctG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TGFBI	ENST00000506699	?	?	20
TGFBI	ENST00000504185	?	?	5
TGFBI	ENST00000508076	?	ENSP00000423935	11
TGFBI	ENST00000442011	NM_000358.2	ENSP00000416330	20
TGFBI	ENST00000509485	?	ENSP00000426589	2
TGFBI	ENST00000507018	?	ENSP00000421540	20
TGFBI	ENST00000513497	?	?	11
TGFBI	ENST00000515433	?	?	2
TGFBI	ENST00000514554	?	ENSP00000421440	14
TGFBI	ENST00000509749	?	?	1
TGFBI	ENST00000514242	?	?	2
TGFBI	ENST00000508767	?	ENSP00000423871	2
TGFBI	ENST00000503087	?	ENSP00000427137	11
TGFBI	ENST00000604555	?	ENSP00000474155	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TGFBI	ENST00000507018	?	ENSP00000421540	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TGFBI	ENST00000506699	?	3
TGFBI	ENST00000515433	?	2
TGFBI	ENST00000504411	?	1

Locus 136 (rs2005199)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,016,530-52,033,572
block size	17,043 bp
variant count	28 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.517 [-3.498 – 0.854]	gene(s) hit or close-by	CTD-3073N11.9, SIGLEC27P, SIGLEC6
phastCons	μ = 0.077 [0 – 0.626]	eQTL gene(s)	LRRC4B, ZNF175
GERP++	μ = -0.741 [-3.21 – 1.89]	potentially regulated gene(s)	SIGLEC10, SIGLEC12, SIGLEC14, SIGLEC6
CADD score	μ = 3.671 [0.042 – 16.32]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SIGLEC6	missense variant	ENST00000425629	NM_001245.5	ENSP00000401502	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000346477	NM_198845.4	ENSP00000344064	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000436458	NM_001177547.1	ENSP00000410679	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000343300	NM_198846.4	ENSP00000345907	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000391797	NM_001177549.1	ENSP00000375674	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000359982	NM_001177548.1	ENSP00000353071	F/L	Ttc/Ctc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ZNF175	ENST00000436511	ILMN_1675788	blood	4.95×10-7 (p-value)	MuTHER consortium	12
ZNF175	ENST00000262259					
ZNF175	?	ENSG00000105497	esophagus mucosa	2.20×10-6 (p-value)	GTEx Portal V6	4
LRRC4B	?	ENSG00000131409	lung	1.70×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000342383	1	ENST00000425629, ENST00000436458, ENST00000391797, ENST00000346477, ENST00000359982	SIGLEC6

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000232795	1	ENCP00000024652	SIGLEC14

	ENCP00000024646	SIGLEC6
	ENCP00000024642	SIGLEC10
	ENCP00000024643	SIGLEC10
	ENCP00000024645	SIGLEC12

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000217752 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	CTCF, H3K27me3
		blood (K562)	CTCF, Rad21, DNase1
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-3073N11.9	upstream gene variant, downstream gene variant	220	ENST00000598220	?	?	16
SIGLEC27P	downstream gene variant	3965	ENST00000594477	?	?	1
SIGLEC6	downstream gene variant	14	ENST00000346477	NM_198845.4	ENSP00000344064	8
SIGLEC6	downstream gene variant, upstream gene variant	225	ENST00000474054	?	?	11
SIGLEC6	downstream gene variant	406	ENST00000489837	?	?	7
SIGLEC6	downstream gene variant	219	ENST00000436458	NM_001177547.1	ENSP00000410679	6
SIGLEC6	downstream gene variant	488	ENST00000391797	NM_001177549.1	ENSP00000375674	6
SIGLEC6	downstream gene variant	219	ENST00000343300	NM_198846.4	ENSP00000345907	6
SIGLEC6	downstream gene variant	219	ENST00000359982	NM_001177548.1	ENSP00000353071	6
SIGLEC6	downstream gene variant	236	ENST00000425629	NM_001245.5	ENSP00000401502	6
SIGLEC6	downstream gene variant	277	ENST00000496422	?	?	7

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC6	ENST00000489837	?	?	2
SIGLEC6	ENST00000436458	NM_001177547.1	ENSP00000410679	18
SIGLEC6	ENST00000346477	NM_198845.4	ENSP00000344064	18
SIGLEC6	ENST00000391797	NM_001177549.1	ENSP00000375674	18
SIGLEC6	ENST00000425629	NM_001245.5	ENSP00000401502	18
SIGLEC6	ENST00000474054	?	?	14
SIGLEC6	ENST00000359982	NM_001177548.1	ENSP00000353071	18

SIGLEC6	ENST00000343300	NM_198846.4	ENSP00000345907	18
SIGLEC6	ENST00000496422	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SIGLEC6	ENST00000489837	?	1
SIGLEC6	ENST00000496422	?	1

Locus 137 (rs2280774)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,900,372-32,057,972
block size	157,601 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.353 [-2.371 – 1.344]	gene(s) hit or close-by	C2, C2-AS1, C4A, C4B, C4B-AS1, CFB, CYP21A2, DXO, MIR1236, NELFE, RNA5SP206, SKIV2L, STK19, TNXB, XXbac-BPG116M5.17
phastCons	μ = 0.032 [0 – 0.43]	eQTL gene(s)	AGPAT1, AIF1, ATF6B, BAG6, CCHCR1, CYP21A1P, CYP21A2, DXO, EHMT2, HLA-B, HLA-C, HLA-DQA2, HLA-DQB1, HLA-DQB1-AS1, HLA-DRB5, HSPA1B, HSPA1L, LST1, MSH5, MSH5-SAPCD1, NELFE, NOTCH4, PRRC2A, PSORS1C3, VARS2, XXbac-BPG248L24.12, XXbac-BPG300A18.13
GERP++	μ = -0.651 [-7.07 – 3.29]	potentially regulated gene(s)	BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1
CADD score	μ = 4.496 [0.002 – 17.42]	disease gene(s)	C2, VARS2, CFB, SKIV2L, CYP21A2, C4A, C4B, BTNL2, HLA-DQB1, HLA-B, TNXB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Ulcerative colitis	$<5.00\times10^{-14}$	GWAS Catalog	24837172	1
Complement C3 and C4 levels	$<1.00\times10^{-22}$	GWAS Catalog	23028341	1

Disease gene annotation

gene	trait	source DB	source entry/link
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
CYP21A2	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387

	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
BTNL2	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
HLA-DQB1	Bullous pemphigoid	OrphaNet	OrphaNet:703	orphanet
HLA-DQB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
HLA-DQB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465	orphanet
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	orphanet
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	OrphaNet	OrphaNet:315306	orphanet
CYP21A2	Nonclassic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	OrphaNet	OrphaNet:95698	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	OrphaNet	OrphaNet:315311	orphanet
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839	orphanet
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285	orphanet
BTNL2	Sarcoidosis	OrphaNet	OrphaNet:797	orphanet
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CYP21A2	splice acceptor variant	ENST00000464325	?	?					1
CYP21A2	missense variant	ENST00000478281	?	ENSP00000419572	K/R	aAg/aGg	?	?	1
CYP21A2	stop retained variant	ENST00000469053	?	ENSP00000418104	*	taA/taG	?	?	1
CYP21A2	missense variant	ENST00000418967	NM_000500.7	ENSP00000408860	K/R	aAg/aGg	?	?	1
CYP21A2	missense variant	ENST00000471671	?	ENSP00000418561	K/R	aAg/aGg	?	?	1
CYP21A2	missense variant	ENST00000435122	NM_001128590.3	ENSP00000415043	K/R	aAg/aGg	?	?	1
CYP21A2	stop retained variant	ENST00000466779	?	ENSP00000417321	*	taA/taG	?	?	1
TNXB	missense variant	ENST00000611016	?	ENSP00000483409	I/N	aTc/aAc	?	?	1
TNXB	missense variant	ENST00000451343	NM_032470.3	ENSP00000407685	I/N	aTc/aAc	?	?	1
TNXB	missense variant	ENST00000375244	NM_019105.6	ENSP00000364393	I/N	aTc/aAc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DXO	ENST00000474587	ILMN_1726990	blood	2.01×10-5 (p-value)	MuTHER consortium	6
DXO	ENST00000491327					
DXO	ENST00000473976					
DXO	ENST00000480240					
DXO	ENST00000498357					

DXO	ENST00000477826					
DXO	ENST00000492946					
DXO	ENST00000375349					
DXO	ENST00000337523					
DXO	ENST00000460058					
DXO	ENST00000375356					
DXO	ENST00000487914					
DXO	ENST00000478221					
DXO	ENST00000485557					
PRRC2A	ENST00000376033	ILMN_2408179	blood	1.51×10-6 (p-value)	MuTHER consortium	7
PRRC2A	ENST00000487089					
PRRC2A	ENST00000376007					
PRRC2A	ENST00000487839					
PRRC2A	ENST00000462617					
PRRC2A	ENST00000482441					
PRRC2A	ENST00000492691					
HSPA1B	ENST00000375650	ILMN_1660436	adipocyte	2.53×10-6 (p-value)	MuTHER consortium	3
AIF1	ENST00000337917	ILMN_1792473	blood	3.51×10-10 (p-value)	MuTHER consortium	7
AIF1	ENST00000466820					
AIF1	ENST00000497362					
AIF1	ENST00000376059					
AIF1	ENST00000376049					
MSH5	ENST00000395853	ILMN_1810834	blood	6.83×10-6 (p-value)	MuTHER consortium	7
MSH5	ENST00000375750					
MSH5	ENST00000429846					
MSH5	ENST00000423982					
MSH5	ENST00000484309					
MSH5-SAPCD1	ENST00000498473					
MSH5	ENST00000375703					
MSH5	ENST00000375740					
MSH5-SAPCD1	ENST00000493662					
MSH5	ENST00000375755					
MSH5-SAPCD1	ENST00000491552					
MSH5	ENST00000463144					
LST1	ENST00000339530	ILMN_1718936	blood	6.85×10-5 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
CYP21A1P	?	ENSG00000204338	muscularis mucosae	9.51×10-12 (p-value)	GTEx Portal V6	18
ATF6B	?	ENSG00000213676	transformed fibroblasts	2.30×10-18 (p-value)	GTEx Portal V6	18
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	5.13×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG300A18.13	?	ENSG00000273333	transformed fibroblasts	2.63×10-6 (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	blood	8.43×10-7 (p-value)	GTEx Portal V6	6
HLA-B	?	ENSG00000234745	blood	5.49×10-6 (p-value)	GTEx Portal V6	2

HLA-DQA2	?	ENSG00000237541	blood	2.78×10-7 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	blood	9.80×10-10 (p-value)	GTEx Portal V6	15
PRRC2A	?	ENSG00000204469	blood	7.42×10-7 (p-value)	GTEx Portal V6	8
ATF6B	?	ENSG00000213676	breast	1.64×10-10 (p-value)	GTEx Portal V6	18
HLA-B	?	ENSG00000234745	breast	1.39×10-6 (p-value)	GTEx Portal V6	3
BAG6	?	ENSG00000204463	tibial artery	7.76×10-12 (p-value)	GTEx Portal V6	18
XXbac-BPG300A18.13	?	ENSG00000273333	tibial artery	1.30×10-9 (p-value)	GTEx Portal V6	18
HLA-C	?	ENSG00000204525	tibial artery	2.02×10-6 (p-value)	GTEx Portal V6	8
CYP21A1P	?	ENSG00000204338	tibial artery	1.82×10-14 (p-value)	GTEx Portal V6	18
VARS2	ENST00000321897	ILMN_1737585	monocyte	1.34×10-4 (p-value)	Fairfax et al.	7
VARS2	ENST00000625423		b-cell	1.90×10-5 (p-value)	Fairfax et al.	7
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
ATF6B	?	ENSG00000213676	skeletal muscle	8.95×10-19 (p-value)	GTEx Portal V6	18
HLA-C	?	ENSG00000204525	skeletal muscle	1.45×10-6 (p-value)	GTEx Portal V6	3
CYP21A1P	?	ENSG00000204338	skeletal muscle	1.12×10-7 (p-value)	GTEx Portal V6	12
HLA-C	?	ENSG00000204525	sun exposed skin	5.42×10-7 (p-value)	GTEx Portal V6	8
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	1.03×10-6 (p-value)	GTEx Portal V6	3
EHMT2	?	ENSG00000204371	sun exposed skin	4.15×10-6 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	sun exposed skin	7.27×10-7 (p-value)	GTEx Portal V6	14
XXbac-BPG300A18.13	?	ENSG00000273333	sun exposed skin	9.84×10-7 (p-value)	GTEx Portal V6	2
ATF6B	?	ENSG00000213676	left ventricle	2.03×10-13 (p-value)	GTEx Portal V6	18
CYP21A1P	?	ENSG00000204338	left ventricle	3.41×10-7 (p-value)	GTEx Portal V6	11
XXbac-BPG300A18.13	?	ENSG00000273333	aorta	4.87×10-7 (p-value)	GTEx Portal V6	18
XXbac-BPG248L24.12	?	ENSG00000271581	aorta	3.80×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	aorta	1.02×10-5 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	aorta	1.09×10-7 (p-value)	GTEx Portal V6	15
HLA-DQB1	?	ENSG00000179344	subcutaneous adipocytes	1.82×10-6 (p-value)	GTEx Portal V6	4
HLA-DQB1-AS1	?	ENSG00000223534	subcutaneous adipocytes	6.49×10-7 (p-value)	GTEx Portal V6	15
CYP21A1P	?	ENSG00000204338	subcutaneous adipocytes	8.10×10-16 (p-value)	GTEx Portal V6	18
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	7.08×10-7 (p-value)	GTEx Portal V6	4
ATF6B	?	ENSG00000213676	subcutaneous adipocytes	2.38×10-7 (p-value)	GTEx Portal V6	14
PSORS1C3	?	ENSG00000204528	stomach	5.16×10-8 (p-value)	GTEx Portal V6	3
ATF6B	?	ENSG00000213676	stomach	6.65×10-7 (p-value)	GTEx Portal V6	1

HLA-C	?	ENSG00000204525	tibial nerve	3.59×10-7 (p-value)	GTEx Portal V6	8
CYP21A1P	?	ENSG00000204338	tibial nerve	4.93×10-14 (p-value)	GTEx Portal V6	18
ATF6B	?	ENSG00000213676	tibial nerve	2.15×10-8 (p-value)	GTEx Portal V6	12
CCHCR1	?	ENSG00000204536	tibial nerve	2.05×10-6 (p-value)	GTEx Portal V6	12
XXbac-BPG300A18.13	?	ENSG00000273333	tibial nerve	2.25×10-6 (p-value)	GTEx Portal V6	10
XXbac-BPG300A18.13	?	ENSG00000273333	esophagus mucosa	1.02×10-8 (p-value)	GTEx Portal V6	18
ATF6B	?	ENSG00000213676	esophagus mucosa	8.01×10-7 (p-value)	GTEx Portal V6	11
DXO	ENST00000474587	ILMN_1726990	blood	1.56×10-5 (p-value)	Westra et al.	2
DXO	ENST00000491327					
DXO	ENST00000473976					
DXO	ENST00000480240					
DXO	ENST00000498357					
DXO	ENST00000477826					
DXO	ENST00000492946					
DXO	ENST00000375349					
DXO	ENST00000337523					
DXO	ENST00000460058					
DXO	ENST00000375356					
DXO	ENST00000487914					
DXO	ENST00000478221					
DXO	ENST00000485557					
HSPA1L	ENST00000375654	ILMN_1654566	blood	3.16×10-6 (p-value)	Westra et al.	2
HSPA1B	ENST00000375650	ILMN_1660436	blood	2.45×10-6 (p-value)	Westra et al.	1
NELFE	ENST00000492185	ILMN_1765532	blood	2.23×10-6 (p-value)	Westra et al.	4
NELFE	ENST00000488426					
NELFE	ENST00000481121					
NELFE	ENST00000375429					
NOTCH4	ENST00000474612	ILMN_1711157	blood	3.19×10-5 (p-value)	Westra et al.	3
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
AGPAT1	ENST00000375107	ILMN_1679520	blood	1.09×10-4 (p-value)	Westra et al.	2
AGPAT1	ENST00000395499					
AGPAT1	ENST00000375104					
AGPAT1	ENST00000395496					
AGPAT1	ENST00000395497					
AGPAT1	ENST00000490711					
AGPAT1	ENST00000336984					
CYP21A1P	?	ENSG00000204338	adrenal gland	4.42×10-10 (p-value)	GTEx Portal V6	18
CYP21A1P	?	ENSG00000204338	testis	6.35×10-11 (p-value)	GTEx Portal V6	18
EHMT2	?	ENSG00000204371	testis	3.76×10-6 (p-value)	GTEx Portal V6	2
CYP21A1P	?	ENSG00000204338	lung	3.80×10-12 (p-value)	GTEx Portal V6	17
CYP21A1P	?	ENSG00000204338	thyroid	4.47×10-12 (p-value)	GTEx Portal V6	17
CYP21A2	?	ENSG00000231852	thyroid	4.28×10-6 (p-value)	GTEx Portal V6	11
ATF6B	?	ENSG00000213676	atrial appendage	7.92×10-9 (p-value)	GTEx Portal V6	14
CYP21A1P	?	ENSG00000204338	atrial appendage	7.65×10-7 (p-value)	GTEx Portal V6	2

CYP21A1P	?	ENSG00000204338	transverse colon	6.50×10-8 (p-value)	GTEx Portal V6	15
CYP21A1P	?	ENSG00000204338	unexposed skin	5.25×10-7 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	cortex	5.11×10-9 (p-value)	GTEx Portal V6	15
CYP21A1P	?	ENSG00000204338	sigmoid colon	4.18×10-8 (p-value)	GTEx Portal V6	13
CYP21A1P	?	ENSG00000204338	cerebellum	3.20×10-8 (p-value)	GTEx Portal V6	13
CYP21A1P	?	ENSG00000204338	caudate basal ganglia	1.05×10-6 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	cerebellar hemisphere	3.59×10-7 (p-value)	GTEx Portal V6	12
HLA-DQB1	?	ENSG00000179344	blood	4.04×10-2 (q-value)	SeeQTL DB (HapMap)	2
HLA-DRB5	ENST00000374975	ILMN_1697499	monocyte	1.61×10-12 (p-value)	Zeller et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445144	1	ENCP00000048200	BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2
		ENCP00000048071	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048171	PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001703098 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001703099 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00000662352 (promoter flanking region)	1	NHLF	DNase1, H3K4me1, H3K27ac
		HSMMtube	DNase1, H3K27me3
		Osteobl	H3K4me2, H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3, Max, DNase1
		skin (NHDF-AD)	H3K4me1, DNase1, H3K27ac
		muscle (HSMM)	DNase1, H3K27me3
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3, DNase1

liver (HepG2)	H3K4me1, H3K27me3
lung (IMR90)	H3K36me3, H3K27me3, DNase1
blood (GM12878)	H3K27me3
nervous (NH-A)	H3K27me3, DNase1
skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C2	upstream gene variant	1688	ENST00000485690	?	ENSP00000417790	1
C2	upstream gene variant, downstream gene variant	3460	ENST00000486124	?	?	2
C2	downstream gene variant	4910	ENST00000497706	?	ENSP00000417482	1
C2	downstream gene variant	4139	ENST00000383177	?	ENSP00000372664	1
C2	downstream gene variant	4092	ENST00000442278	NM_001145903.2	ENSP00000395683	1
C2	downstream gene variant	3701	ENST00000413154	?	ENSP00000403325	1
C2	downstream gene variant	4242	ENST00000469372	NM_001282457.1	ENSP00000418923	1
C2	downstream gene variant	4092	ENST00000452323	NM_001178063.2	ENSP00000392322	1
C2	downstream gene variant	4091	ENST00000468407	?	?	1
C2	downstream gene variant	4372	ENST00000482060	?	ENSP00000418332	1
C2	downstream gene variant	4091	ENST00000299367	NM_001282458.1, NM_000063.5	ENSP00000299367	1
C2-AS1	downstream gene variant	1879	ENST00000630806	?	?	1
C4A	upstream gene variant	4155	ENST00000428956	NM_007293.2	ENSP00000396688	2
C4A	upstream gene variant	4974	ENST00000467749	?	?	1
C4A	upstream gene variant	4199	ENST00000498271	NM_001252204.1	ENSP00000420212	2
C4B	downstream gene variant	3441	ENST00000468936	?	?	2
C4B	downstream gene variant	3427	ENST00000425700	?	ENSP00000391933	2
C4B	downstream gene variant	3426	ENST00000435363	NM_001002029.3	ENSP00000415941	2
C4B	downstream gene variant	3842	ENST00000496065	?	?	2
C4B	downstream gene variant	3443	ENST00000468237	?	?	2
C4B	downstream gene variant	4513	ENST00000473957	?	?	2
C4B	downstream gene variant	3610	ENST00000463249	?	?	2
C4B	downstream gene variant	4239	ENST00000496560	?	?	2
C4B-AS1	upstream gene variant	2586	ENST00000415626	?	?	2
CFB	downstream gene variant	165	ENST00000497841	?	?	2
CFB	upstream gene variant, downstream gene variant	1111	ENST00000482312	?	?	2
CFB	downstream gene variant	506	ENST00000482886	?	?	2
CFB	upstream gene variant, downstream gene variant	1614	ENST00000498317	?	ENSP00000418833	2
CFB	downstream gene variant	1794	ENST00000460718	?	ENSP00000417793	1
CFB	downstream gene variant	2622	ENST00000475617	?	ENSP00000420090	1
CFB	downstream gene variant	212	ENST00000461483	?	?	2
CFB	downstream gene variant	2643	ENST00000472581	?	?	1

CFB	downstream gene variant	165	ENST00000467150	?	?	2
CFB	downstream gene variant	3053	ENST00000452035	?	?	1
CFB	downstream gene variant	1645	ENST00000483004	?	ENSP00000419887	1
CFB	downstream gene variant	1614	ENST00000425368	NM_001710.5	ENSP00000416561	1
CFB	downstream gene variant	165	ENST00000465750	?	?	2
CFB	downstream gene variant	2454	ENST00000467360	?	?	1
CYP21A2	downstream gene variant	3062	ENST00000478281	?	ENSP00000419572	1
CYP21A2	downstream gene variant	825	ENST00000418967	NM_000500.7	ENSP00000408860	1
CYP21A2	downstream gene variant	3011	ENST00000488465	?	?	1
CYP21A2	downstream gene variant	2855	ENST00000469053	?	ENSP00000418104	1
CYP21A2	downstream gene variant	853	ENST00000435122	NM_001128590.3	ENSP00000415043	1
CYP21A2	downstream gene variant	2672	ENST00000466879	?	?	1
CYP21A2	downstream gene variant	853	ENST00000479074	?	?	1
CYP21A2	downstream gene variant	1309	ENST00000483041	?	?	1
CYP21A2	downstream gene variant	2889	ENST00000471671	?	ENSP00000418561	1
CYP21A2	downstream gene variant	853	ENST00000486063	?	?	1
CYP21A2	downstream gene variant	853	ENST00000479730	?	?	1
CYP21A2	downstream gene variant	2739	ENST00000464325	?	?	1
CYP21A2	downstream gene variant	3380	ENST00000480027	?	?	1
CYP21A2	upstream gene variant, downstream gene variant	105	ENST00000462278	?	?	3
CYP21A2	downstream gene variant	2671	ENST00000466779	?	ENSP00000417321	1
DXO	upstream gene variant	4609	ENST00000487914	?	?	1
DXO	upstream gene variant	4462	ENST00000480240	?	ENSP00000418632	1
DXO	upstream gene variant	4503	ENST00000473976	?	?	1
DXO	upstream gene variant	4346	ENST00000477826	?	?	1
DXO	upstream gene variant	4436	ENST00000492946	?	?	1
DXO	upstream gene variant	4915	ENST00000498357	?	?	1
DXO	upstream gene variant	4500	ENST00000474587	?	?	1
DXO	upstream gene variant	4389	ENST00000485557	?	?	1
DXO	upstream gene variant	4360	ENST00000478221	?	?	1
DXO	upstream gene variant	4306	ENST00000337523	NM_005510.3	ENSP00000337759	1
DXO	upstream gene variant	4589	ENST00000460058	?	?	1
DXO	upstream gene variant	4614	ENST00000375356	?	ENSP00000364505	1
DXO	upstream gene variant	4913	ENST00000491327	?	?	1
DXO	upstream gene variant	4308	ENST00000375349	?	ENSP00000364498	1
MIR1236	downstream gene variant, upstream gene variant	3141	ENST00000408340	?	?	2
NELFE	downstream gene variant, upstream gene	1075	ENST00000492539	?	ENSP00000419183	2

	variant					
NELFE	downstream gene variant, upstream gene variant	1804	ENST00000375429	NM_002904.5	ENSP00000364578	2
NELFE	downstream gene variant, upstream gene variant	60	ENST00000441998	?	ENSP00000397914	3
NELFE	downstream gene variant, upstream gene variant	2062	ENST00000375425	?	ENSP00000364574	2
NELFE	downstream gene variant, upstream gene variant	691	ENST00000436289	?	ENSP00000414029	3
NELFE	downstream gene variant, upstream gene variant	606	ENST00000454913	?	ENSP00000409389	3
NELFE	downstream gene variant, upstream gene variant	1386	ENST00000494956	?	ENSP00000420205	2
NELFE	downstream gene variant, upstream gene variant	2468	ENST00000625905	?	ENSP00000486884	2
NELFE	downstream gene variant, upstream gene variant	1122	ENST00000426722	?	ENSP00000394340	2
NELFE	downstream gene variant, upstream gene variant	1952	ENST00000481121	?	?	2
NELFE	downstream gene variant, upstream gene variant	1367	ENST00000491139	?	?	2
NELFE	downstream gene variant, upstream gene variant	1995	ENST00000488426	?	?	2
NELFE	downstream gene variant, upstream gene variant	1954	ENST00000444811	?	ENSP00000388400	2
NELFE	downstream gene variant, upstream gene variant	1876	ENST00000492185	?	?	2
RNA5SP206	upstream gene variant	4248	ENST00000516703	?	?	1
SKIV2L	downstream gene variant	956	ENST00000492900	?	?	1
SKIV2L	upstream gene variant	583	ENST00000466290	?	?	1
STK19	downstream gene variant	4179	ENST00000460018	?	ENSP00000418350	1
STK19	downstream gene variant	3222	ENST00000479644	?	?	3
STK19	upstream gene variant	2522	ENST00000490822	?	?	3
TNXB	downstream gene variant	2046	ENST00000490077	?	?	2
TNXB	downstream gene variant	3019	ENST00000479795	?	ENSP00000418248	1
TNXB	downstream gene variant	3157	ENST00000498094	?	?	2
TNXB	downstream gene variant	2045	ENST00000375244	NM_019105.6	ENSP00000364393	2
TNXB	downstream gene variant	2046	ENST00000451343	NM_032470.3	ENSP00000407685	2
TNXB	downstream gene variant	653	ENST00000613214	?	ENSP00000480067	2
TNXB	downstream gene variant, upstream gene variant	23	ENST00000611016	?	ENSP00000483409	5
XXbac-BPG116M5.17	downstream gene variant	1650	ENST00000456570	?	ENSP00000410815	1
XXbac-BPG116M5.17	downstream gene variant	1668	ENST00000477310	?	ENSP00000418996	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C2	ENST00000442278	NM_001145903.2	ENSP00000395683	1
C2	ENST00000411571	?	ENSP00000388727	1
C2	ENST00000447952	?	ENSP00000391354	1
C2	ENST00000494905	?	ENSP00000419048	1
C2	ENST00000452323	NM_001178063.2	ENSP00000392322	1

C2	ENST00000452202	?	ENSP00000406121	1
C2	ENST00000482060	?	ENSP00000418332	1
C2	ENST00000484636	?	ENSP00000420305	1
C2	ENST00000497706	?	ENSP00000417482	1
C2	ENST00000418949	NM_001282459.1	ENSP00000406190	1
C2	ENST00000383177	?	ENSP00000372664	1
C2	ENST00000469372	NM_001282457.1	ENSP00000418923	1
C2	ENST00000299367	NM_001282458.1, NM_000063.5	ENSP00000299367	1
CFB	ENST00000425368	NM_001710.5	ENSP00000416561	1
CFB	ENST00000452035	?	?	1
CFB	ENST00000483004	?	ENSP00000419887	1
CYP21A2	ENST00000479730	?	?	1
CYP21A2	ENST00000464325	?	?	1
CYP21A2	ENST00000488465	?	?	1
CYP21A2	ENST00000471671	?	ENSP00000418561	1
CYP21A2	ENST00000418967	NM_000500.7	ENSP00000408860	1
CYP21A2	ENST00000479074	?	?	1
CYP21A2	ENST00000478281	?	ENSP00000419572	1
CYP21A2	ENST00000483041	?	?	1
CYP21A2	ENST00000486063	?	?	1
CYP21A2	ENST00000469053	?	ENSP00000418104	1
CYP21A2	ENST00000435122	NM_001128590.3	ENSP00000415043	1
CYP21A2	ENST00000466779	?	ENSP00000417321	1
NELFE	ENST00000492185	?	?	1
NELFE	ENST00000481121	?	?	1
NELFE	ENST00000375429	NM_002904.5	ENSP00000364578	1
NELFE	ENST00000444811	?	ENSP00000388400	1
NELFE	ENST00000375425	?	ENSP00000364574	1
NELFE	ENST00000488426	?	?	1
SKIV2L	ENST00000375394	NM_006929.4	ENSP00000364543	1
SKIV2L	ENST00000461073	?	ENSP00000419905	1
SKIV2L	ENST00000465703	?	?	1
SKIV2L	ENST00000474839	?	ENSP00000420470	1
SKIV2L	ENST00000628157	?	ENSP00000485707	1
SKIV2L	ENST00000488648	?	?	1
STK19	ENST00000484540	?	?	3
STK19	ENST00000492583	?	?	3
STK19	ENST00000491861	?	?	3
STK19	ENST00000478486	?	?	3
STK19	ENST00000375333	NM_032454.1	ENSP00000364482	3
STK19	ENST00000466336	?	?	3

STK19	ENST00000469907	?	?	3
STK19	ENST00000473983	?	?	3
STK19	ENST00000463823	?	?	3
STK19	ENST00000375331	NM_004197.1	ENSP00000364480	3
STK19	ENST00000466132	?	?	3
STK19	ENST00000471028	?	?	3
STK19	ENST00000519179	?	ENSP00000454870	3
STK19	ENST00000483801	?	ENSP00000418866	3
TNXB	ENST00000613214	?	ENSP00000480067	4
TNXB	ENST00000375244	NM_019105.6	ENSP00000364393	9
TNXB	ENST00000611016	?	ENSP00000483409	3
XXbac-BPG116M5.17	ENST00000456570	?	ENSP00000410815	2
XXbac-BPG116M5.17	ENST00000477310	?	ENSP00000418996	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CFB	ENST00000467360	?	1
CYP21A2	ENST00000479730	?	2
CYP21A2	ENST00000488465	?	2
CYP21A2	ENST00000466879	?	2
CYP21A2	ENST00000479074	?	2
CYP21A2	ENST00000483041	?	2
CYP21A2	ENST00000486063	?	2
CYP21A2	ENST00000480027	?	2
TNXB	ENST00000498094	?	1
TNXB	ENST00000490077	?	1

Locus 138 (rs1511802)

SNiPAcard

Block annotations

Block info

genomic range	chr4:187,140,276-187,178,300
block size	38,025 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.067 [-1.778 – 1.154]	gene(s) hit or close-by	KLKB1
phastCons	μ = 0.100 [0 – 1]	eQTL gene(s)	CYP4V2, F11
GERP++	μ = -0.431 [-5.17 – 1.7]	potentially regulated gene(s)	KLKB1
CADD score	μ = 2.994 [0.416 – 10.46]	disease gene(s)	F11, CYP4V2, KLKB1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
bradykinin, des-arg(9)	8.22×10^{-33}	Metabolomics GWAS Server	24816252	7

Disease gene annotation

gene	trait	source DB	source entry/link
F11	FACTOR XI DEFICIENCY	OMIM	MIM:612416
CYP4V2	BIETTI CRYSTALLINE CORNEORETINAL DYSTROPHY	OMIM	MIM:210370
F11	Congenital factor XI deficiency	OrphaNet	OrphaNet:329
CYP4V2	Bietti crystalline dystrophy	OrphaNet	OrphaNet:41751
KLKB1	Congenital prekallikrein deficiency	OrphaNet	OrphaNet:749

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
F11	?	ENSG00000088926	muscularis mucosae	2.06×10^{-11} (p-value)	GTEx Portal V6	15
CYP4V2	?	ENSG00000145476	tibial nerve	7.73×10^{-12} (p-value)	GTEx Portal V6	15
F11	?	ENSG00000088926	tibial artery	2.59×10^{-9} (p-value)	GTEx Portal V6	15
CYP4V2	?	ENSG00000145476	thyroid	6.38×10^{-7} (p-value)	GTEx Portal V6	15
F11	?	ENSG00000088926	skeletal muscle	3.43×10^{-8} (p-value)	GTEx Portal V6	7
CYP4V2	?	ENSG00000145476	skeletal muscle	2.18×10^{-7} (p-value)	GTEx Portal V6	6
F11	?	ENSG00000088926	cortex	9.79×10^{-7} (p-value)	GTEx Portal V6	11
F11	?	ENSG00000088926	transverse colon	3.76×10^{-7} (p-value)	GTEx Portal V6	6
CYP4V2	ENST00000507209	ILMN_2054607	monocyte	3.48×10^{-4} (p-value)	Fairfax et al.	1
CYP4V2	ENST00000502665					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000642205	1	ENST00000511406, ENST00000511608, ENST00000264690, ENST00000513864	KLKB1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KLKB1	downstream gene variant	3011	ENST00000428196	?	ENSP00000412366	1
KLKB1	downstream gene variant	1603	ENST00000446598	?	ENSP00000415563	4
KLKB1	upstream gene variant, downstream gene variant	229	ENST00000467271	?	?	11

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLKB1	ENST00000511608	?	ENSP00000426629	18
KLKB1	ENST00000511406	?	?	17
KLKB1	ENST00000446598	?	ENSP00000415563	1
KLKB1	ENST00000428196	?	ENSP00000412366	1
KLKB1	ENST00000513864	?	ENSP00000424469	17
KLKB1	ENST00000264690	NM_000892.3	ENSP00000264690	17

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
KLKB1	ENST00000467271	?	1

Locus 139 (rs1455434)

SNiPAcard

Block annotations

Block info

genomic range	chr19:41,214,932-41,291,872
block size	76,941 bp
variant count	30 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.573 [-3.842 – 1.533]	gene(s) hit or close-by	ADCK4, C19orf54, ITPKC, MIA, MIA-RAB4B, RAB4B, RAB4B-EGLN2, RNU6-195P, SNRPA
phastCons	μ = 0.034 [0 – 0.546]	eQTL gene(s)	C19orf54, MIA
GERP++	μ = -1.528 [-9.93 – 1.94]	potentially regulated gene(s)	C19orf54
CADD score	μ = 5.053 [0.217 – 14.91]	disease gene(s)	ADCK4, ITPKC

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ADCK4	NEPHROTIC SYNDROME, TYPE 9	OMIM	MIM:615573
ITPKC	KAWASAKI DISEASE	OMIM	MIM:611775
ADCK4	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis	OrphaNet	OrphaNet:93213

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C19orf54	?	ENSG00000188493	thyroid	7.69×10-8 (p-value)	GTEx Portal V6	21
MIA	?	ENSG00000261857	tibial nerve	1.84×10-7 (p-value)	GTEx Portal V6	23
C19orf54	?	ENSG00000188493	blood	2.44×10-8 (q-value)	SeeQTL DB (HapMap)	8

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
EGR1	MA0162.2	5	yes	0.000	1

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000330481	1	ENST00000470681, ENST00000469741	C19orf54

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024392		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000228995	1	ENCP00000024393	C19orf54

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001648159 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000346640 (promoter)	4	embryonic stem cell (H1ESC)	H3K36me3, CTCF, PolII, p300, Sin3Ak20, H3K27ac, TAF1, H3K4me2, SP1, USF1, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K27ac, H3K4me2, DNase1, H2AZ, H3K4me3, H3K9ac, H3K79me2
		blood (K562)	H3K9ac, Gabp, H3K79me2, H2AZ, ELF1, Cfos, PolII, H3K4me2, H3K27me3, H3K36me3, DNase1, H3K4me3, Max, H3K27ac, Egr1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me3, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K79me2, H2AZ
		liver (HepG2)	Gabp, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H3K4me2, HNF4A, H3K9ac, H3K27ac, H3K4me3, H3K36me3, H3K27me3, DNase1
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, H3K79me2, Pbx3, Sin3Ak20, PolII, H2AZ, DNase1, Gabp, ELF1
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me3, H3K79me2, Gabp, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K4me2, H3K9ac, H3K4me1, H3K27ac, PolII, DNase1, Cmyc
		A549	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac, H3K36me3
ENSR00000346651 (promoter)	5	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, USF1, H3K9ac, H3K4me3, Yy1, ATF3, H4K5ac, H3K36me3, H3K27me3, PolII, TAF7, Sin3Ak20, H3K27ac, DNase1
		HSMMtube	H3K36me3, H3K4me2, H3K9ac, H3K27ac, DNase1, H2AZ, H3K4me3
		blood (K562)	Yy1, Egr1, H3K27ac, Nfe2, E2F6, H3K9ac, USF1, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Max, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2
		liver (HepG2)	H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K36me3, USF1, ELF1, Gabp, Yy1, PolII, H3K79me2, TAF1, DNase1
		blood (GM12878)	H3K36me3, H3K4me1, DNase1, Yy1, ELF1, H3K79me2, H3K4me3, USF1, H3K27ac, H4K20me1, Cfos, H3K4me2, H2AZ, PolII, Tr4, Cmyc, H3K9ac
		lung (IMR90)	H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K4me2, H3K27ac, DNase1, H4K8ac, H3K79me2
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, PolII, H3K9ac, H3K27ac
		NHLF	H3K36me3, H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H4K20me1, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K4me2, H3K4me1, H3K36me3, H3K9ac, H3K4me3, DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me1, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, Cmyc, H3K36me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H3K9ac, DNase1, H3K4me1, H3K27ac
		endothelium (HUVEC)	H3K36me3, DNase1, Cmyc, PolII, H3K27ac, Max, H3K4me3, H3K9ac
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001648164 (promoter)	1	Osteobl	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ADCK4	upstream gene variant	1207	ENST00000594084	?	ENSP00000473189	2
ADCK4	upstream gene variant	1379	ENST00000594490	?	ENSP00000471310	3

ADCK4	downstream gene variant, upstream gene variant	943	ENST00000596357 ?	ENSP00000472925	4
ADCK4	upstream gene variant	930	ENST00000600080 ?	ENSP00000473017	3
ADCK4	downstream gene variant, upstream gene variant	401	ENST00000600707 ?	ENSP00000472978	5
ADCK4	upstream gene variant	916	ENST00000601967 ?	ENSP00000470916	3
ADCK4	upstream gene variant	2529	ENST00000324464 NM_024876.3	ENSP00000315118	2
ADCK4	upstream gene variant	2518	ENST00000594720 ?	ENSP00000470876	2
ADCK4	downstream gene variant, upstream gene variant	398	ENST00000593723 ?	ENSP00000472775	5
ADCK4	upstream gene variant	941	ENST00000243583 NM_001142555.2	ENSP00000243583	3
ADCK4	upstream gene variant	1229	ENST00000601304 ?	ENSP00000472519	3
ADCK4	upstream gene variant	929	ENST00000595254 ?	ENSP00000470894	3
ADCK4	upstream gene variant	908	ENST00000593544 ?	?	3
ADCK4	upstream gene variant	229	ENST00000596455 ?	?	4
ADCK4	upstream gene variant	47	ENST00000599643 ?	ENSP00000471192	4
ADCK4	upstream gene variant	2529	ENST00000601451 ?	?	2
C19orf54	upstream gene variant	93	ENST00000598352 ?	ENSP00000473024	5
C19orf54	upstream gene variant	22	ENST00000597507 ?	ENSP00000472022	3
C19orf54	downstream gene variant, upstream gene variant	168	ENST00000598485 ?	ENSP00000469330	4
C19orf54	downstream gene variant, upstream gene variant	649	ENST00000470681 ?	ENSP00000471114	3
C19orf54	downstream gene variant, upstream gene variant	145	ENST00000596940 ?	ENSP00000472820	6
C19orf54	upstream gene variant	104	ENST00000600139 ?	ENSP00000472948	5
C19orf54	downstream gene variant	4111	ENST00000594163 ?	?	1
C19orf54	downstream gene variant, upstream gene variant	61	ENST00000378313 NM_198476.3	ENSP00000367564	5
C19orf54	downstream gene variant	4941	ENST00000596809 ?	ENSP00000469807	1
C19orf54	downstream gene variant, upstream gene variant	588	ENST00000469741 ?	ENSP00000471589	3
ITPKC	upstream gene variant	952	ENST00000263370 NM_025194.2	ENSP00000263370	3
ITPKC	upstream gene variant	106	ENST00000597003 ?	ENSP00000472655	3
MIA	upstream gene variant, downstream gene variant	190	ENST00000593317 ?	?	7
MIA	upstream gene variant, downstream gene variant	349	ENST00000601159 ?	ENSP00000471439	5
MIA	downstream gene variant	3612	ENST00000594436 NM_001202553.1	ENSP00000470129	4
MIA	upstream gene variant, downstream gene variant	346	ENST00000597140 ?	ENSP00000470641	5
MIA	upstream gene variant, downstream gene variant	176	ENST00000263369 NM_006533.3	ENSP00000263369	5
MIA	upstream gene variant, downstream gene variant	63	ENST00000597784 ?	ENSP00000469499	5
MIA	upstream gene variant, downstream gene variant	34	ENST00000597600 ?	ENSP00000472982	3
MIA-RAB4B	upstream gene variant	1712	ENST00000600729 ?	ENSP00000472384	1
RAB4B	upstream gene variant	478	ENST00000598430 ?	?	5
RAB4B	upstream gene variant	1615	ENST00000357052 NM_016154.4	ENSP00000349560	3
RAB4B	upstream gene variant	1705	ENST00000378307 ?	ENSP00000367557	3
RAB4B	upstream gene variant	3781	ENST00000595728 ?	ENSP00000471803	2
RAB4B	upstream gene variant, downstream gene variant	1082	ENST00000600078 ?	?	5
RAB4B	upstream gene variant	1774	ENST00000602173 ?	ENSP00000471504	3
RAB4B	upstream gene variant	1646	ENST00000602069 ?	?	3

RAB4B	upstream gene variant	1612	ENST00000594800	?	ENSP00000470246	3
RAB4B	upstream gene variant	2443	ENST00000597476	?	?	3
RAB4B-EGLN2	upstream gene variant	3775	ENST00000596216	?	?	2
RAB4B-EGLN2	upstream gene variant	1638	ENST00000601949	?	?	3
RAB4B-EGLN2	upstream gene variant	1667	ENST00000594136	?	ENSP00000469872	3
RNU6-195P	upstream gene variant	864	ENST00000411352	?	?	2
SNRPA	upstream gene variant, downstream gene variant	405	ENST00000599570	?	?	5
SNRPA	downstream gene variant	2669	ENST00000598452	?	?	2
SNRPA	upstream gene variant, downstream gene variant	157	ENST00000600456	?	ENSP00000471230	6
SNRPA	upstream gene variant, downstream gene variant	401	ENST00000599362	?	ENSP00000472258	5
SNRPA	upstream gene variant, downstream gene variant	68	ENST00000601393	?	ENSP00000472355	5
SNRPA	upstream gene variant, downstream gene variant	397	ENST00000601253	?	ENSP00000469224	5
SNRPA	upstream gene variant	133	ENST00000598923	?	?	4
SNRPA	upstream gene variant	398	ENST00000597353	?	ENSP00000472449	3
SNRPA	upstream gene variant, downstream gene variant	167	ENST00000601545	?	ENSP00000470534	4
SNRPA	upstream gene variant, downstream gene variant	389	ENST00000243563	NM_004596.4	ENSP00000243563	4
SNRPA	upstream gene variant, downstream gene variant	891	ENST00000596860	?	?	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
C19orf54	ENST00000378313	NM_198476.3	ENSP00000367564	L	Ttg/Ctg	1
ITPKC	ENST00000263370	NM_025194.2	ENSP00000263370	A	gcG/gcA	1
MIA	ENST00000597784	?	ENSP00000469499	P	ccT/ccC	1
MIA	ENST00000263369	NM_006533.3	ENSP00000263369	P	ccT/ccC	1
MIA	ENST00000594436	NM_001202553.1	ENSP00000470129	P	ccT/ccC	1
MIA	ENST00000597140	?	ENSP00000470641	P	ccT/ccC	1
MIA-RAB4B	ENST00000600729	?	ENSP00000472384	P	ccT/ccC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ADCK4	ENST00000596357	?	ENSP00000472925	1
ADCK4	ENST00000324464	NM_024876.3	ENSP00000315118	4
ADCK4	ENST00000601451	?	?	4
ADCK4	ENST00000594490	?	ENSP00000471310	2
ADCK4	ENST00000601304	?	ENSP00000472519	2
ADCK4	ENST00000596455	?	?	2
ADCK4	ENST00000593544	?	?	2
ADCK4	ENST00000600080	?	ENSP00000473017	2
ADCK4	ENST00000599643	?	ENSP00000471192	2
ADCK4	ENST00000595254	?	ENSP00000470894	2
ADCK4	ENST00000594720	?	ENSP00000470876	4

ADCK4	ENST00000601967	?	ENSP00000470916	2
ADCK4	ENST00000243583	NM_001142555.2	ENSP00000243583	2
ADCK4	ENST00000600707	?	ENSP00000472978	1
ADCK4	ENST00000594084	?	ENSP00000473189	4
C19orf54	ENST00000596809	?	ENSP00000469807	5
C19orf54	ENST00000470681	?	ENSP00000471114	3
C19orf54	ENST00000597507	?	ENSP00000472022	2
C19orf54	ENST00000598485	?	ENSP00000469330	2
C19orf54	ENST00000469741	?	ENSP00000471589	3
ITPKC	ENST00000597003	?	ENSP00000472655	3
ITPKC	ENST00000263370	NM_025194.2	ENSP00000263370	7
MIA	ENST00000597784	?	ENSP00000469499	2
MIA	ENST00000263369	NM_006533.3	ENSP00000263369	2
MIA	ENST00000601159	?	ENSP00000471439	2
MIA	ENST00000594436	NM_001202553.1	ENSP00000470129	3
MIA	ENST00000597140	?	ENSP00000470641	2
MIA	ENST00000597600	?	ENSP00000472982	1
MIA-RAB4B	ENST00000600729	?	ENSP00000472384	9
RAB4B	ENST00000600078	?	?	4
RAB4B	ENST00000602173	?	ENSP00000471504	6
RAB4B	ENST00000597476	?	?	6
RAB4B	ENST00000602069	?	?	6
RAB4B	ENST00000378307	?	ENSP00000367557	6
RAB4B	ENST00000594800	?	ENSP00000470246	6
RAB4B	ENST00000357052	NM_016154.4	ENSP00000349560	6
RAB4B	ENST00000595728	?	ENSP00000471803	6
RAB4B-EGLN2	ENST00000596216	?	?	6
RAB4B-EGLN2	ENST00000594136	?	ENSP00000469872	6
RAB4B-EGLN2	ENST00000601949	?	?	6
SNRPA	ENST00000600456	?	ENSP00000471230	2
SNRPA	ENST00000598923	?	?	4
SNRPA	ENST00000601545	?	ENSP00000470534	5
SNRPA	ENST00000599570	?	?	3
SNRPA	ENST00000601393	?	ENSP00000472355	4
SNRPA	ENST00000601253	?	ENSP00000469224	3
SNRPA	ENST00000243563	NM_004596.4	ENSP00000243563	5
SNRPA	ENST00000597353	?	ENSP00000472449	5
SNRPA	ENST00000598452	?	?	1
SNRPA	ENST00000599362	?	ENSP00000472258	3

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C19orf54	ENST00000596809	?	ENSP00000469807	3
C19orf54	ENST00000470681	?	ENSP00000471114	1
C19orf54	ENST00000469741	?	ENSP00000471589	1
MIA	ENST00000594436	NM_001202553.1	ENSP00000470129	1
SNRPA	ENST00000243563	NM_004596.4	ENSP00000243563	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RAB4B	ENST00000598430	?	1
SNRPA	ENST00000599570	?	1

Locus 140 (rs12052479)

SNiPAcard

Block annotations

Block info

genomic range	chr2:173,373,190-173,450,756
block size	77,567 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.005 [-4.066 – 0.826]	gene(s) hit or close-by	AC093818.1, ITGA6, PDK1
phastCons	μ = 0.125 [0 – 0.539]	eQTL gene(s)	PDK1
GERP++	μ = -0.023 [-2.38 – 2]	potentially regulated gene(s)	METAP1D, PDK1
CADD score	μ = 3.881 [0.345 – 10.73]	disease gene(s)	ITGA6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ITGA6	Epidermolysis Bullosa with Pyloric Atresia	DECIPHER	MIM:226730
ITGA6	EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA	OMIM	MIM:226730
ITGA6	Junctional epidermolysis bullosa - pyloric atresia	OrphaNet	OrphaNet:79403

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PDK1	?	ENSG00000152256	skeletal muscle	2.06×10^{-11} (p-value)	GTEx Portal V6	15
PDK1	?	ENSG00000152256	blood	6.90×10^{-7} (p-value)	GTEx Portal V6	12
PDK1	?	ENSG00000152256	atrial appendage	1.89×10^{-6} (p-value)	GTEx Portal V6	3
PDK1	?	ENSG00000152256	left ventricle	4.16×10^{-7} (p-value)	GTEx Portal V6	3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000269288	1	ENCP00000028626	PDK1
		ENCP00000028606	METAP1D

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001658967 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PDK1	1	ENST00000282077	hsa-miR-33a-5p
		ENST00000392571	hsa-miR-33b-5p

ENST00000410055
ENST00000416991
ENST00000431718

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC093818.1	downstream gene variant	4197	ENST00000444919	?	?	2
AC093818.1	downstream gene variant, upstream gene variant	567	ENST00000416080	?	?	2
ITGA6	downstream gene variant	4134	ENST00000416789	?	ENSP00000388435	1
ITGA6	downstream gene variant	4210	ENST00000458358	?	ENSP00000394169	1
ITGA6	downstream gene variant	2232	ENST00000264107	NM_000210.2	ENSP00000264107	1
ITGA6	downstream gene variant	3928	ENST00000409532	?	ENSP00000386614	1
ITGA6	downstream gene variant	2180	ENST00000409080	NM_001079818.1	ENSP00000386896	1
ITGA6	downstream gene variant	4210	ENST00000442250	?	ENSP00000406694	1
PDK1	upstream gene variant	2127	ENST00000466437	?	?	1
PDK1	downstream gene variant	3771	ENST00000436490	?	ENSP00000406608	1
PDK1	downstream gene variant	3385	ENST00000443353	?	ENSP00000399558	1
PDK1	downstream gene variant	3385	ENST00000439519	?	ENSP00000388366	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC093818.1	ENST00000450443	?	?	15
AC093818.1	ENST00000442417	?	?	15
PDK1	ENST00000416991	?	ENSP00000399160	1
PDK1	ENST00000282077	?	ENSP00000282077	3
PDK1	ENST00000392571	NM_001278549.1	ENSP00000376352	3
PDK1	ENST00000410055	NM_002610.4	ENSP00000386985	3
PDK1	ENST00000431718	?	ENSP00000413629	1
PDK1	ENST00000466437	?	?	2

Locus 141 (rs10919544)

SNiPAcard

Block annotations

Block info

genomic range	chr1:161,500,130-161,508,763
block size	8,634 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -3.023 [-9.199 – -0.106]	gene(s) hit or close-by	FCGR3A, HSPA6, RP11-25K21.6, RPS23P10
phastCons	μ = 0.115 [0 – 0.46]	eQTL gene(s)	FCGR2B, FCGR2C, FCRLB, HSPA7, RPS23P10, RPS23P9
GERP++	μ = -1.192 [-4.77 – 0.602]	potentially regulated gene(s)	HSPA6, TOMM40L, UFC1, USF1
CADD score	μ = 3.328 [0.487 – 6.425]	disease gene(s)	USF1, FCGR2B, FCGR3A

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
USF1	HYPERLIPIDEMIA, FAMILIAL COMBINED	OMIM	MIM:144250
USF1	HYPERLIPIDEMIA, COMBINED, 1	OMIM	MIM:602491
FCGR2B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
FCGR2B	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
FCGR3A	IMMUNODEFICIENCY 20	OMIM	MIM:615707

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCGR2C	?	ENSG00000244682	muscularis mucosae	9.34×10^{-7} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	muscularis mucosae	7.78×10^{-9} (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	lung	6.16×10^{-7} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	lung	3.33×10^{-8} (p-value)	GTEx Portal V6	3
RPS23P10	?	ENSG00000224203	blood	2.65×10^{-14} (p-value)	GTEx Portal V6	3
RPS23P9	?	ENSG00000231586	blood	3.15×10^{-7} (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	breast	4.93×10^{-8} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	breast	1.60×10^{-8} (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	tibial artery	2.39×10^{-6} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	tibial artery	8.99×10^{-13} (p-value)	GTEx Portal V6	3
HSPA7	ENST00000445535	ILMN_3235964	monocyte	1.02×10^{-6} (p-value)	Fairfax et al.	1
			b-cell	9.19×10^{-12} (p-value)	Fairfax et al.	1
FCGR2C	?	ENSG00000244682	thyroid	1.90×10^{-6} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	thyroid	5.16×10^{-9} (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	skeletal muscle	5.52×10^{-9} (p-value)	GTEx Portal V6	3

HSPA7	?	ENSG00000225217	skeletal muscle	8.17×10-13 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	transverse colon	5.46×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	sun exposed skin	1.08×10-12 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	sun exposed skin	8.65×10-16 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	left ventricle	9.76×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	aorta	3.59×10-6 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	aorta	3.45×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	subcutaneous adipocytes	2.16×10-10 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	subcutaneous adipocytes	1.23×10-13 (p-value)	GTEx Portal V6	3
?	?	ILMN_1666932	liver	1.21×10-10 (p-value)	Innocenti et al.	1
HSPA7	?	ENSG00000225217	visceral adipocytes	2.58×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	coronary artery	2.86×10-6 (p-value)	GTEx Portal V6	1
HSPA7	?	ENSG00000225217	coronary artery	5.03×10-8 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	esophagus mucosa	9.65×10-7 (p-value)	GTEx Portal V6	3
FCGR2B	ENST00000480308	ILMN_2382403	blood	2.26×10-5 (p-value)	Westra et al.	3
FCGR2B	ENST00000403078					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000403078	ILMN_1660027	blood	5.53×10-6 (p-value)	Westra et al.	3
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCRLB	ENST00000367948	ILMN_1782015	blood	1.52×10-6 (p-value)	Westra et al.	3
FCRLB	ENST00000367944					
FCRLB	ENST00000495397					
FCRLB	ENST00000336830					
HSPA7	?	ENSG00000225217	testis	4.32×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	tibial nerve	8.54×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000041151	1	ENCP00000005224	HSPA6
		ENCP00000005167	USF1
		ENCP00000005179	UFC1
		ENCP00000005164	USF1
		ENCP00000005205	TOMM40L
		ENCP00000005166	USF1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000546473 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, CTCF, PolII, Rad21, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	DNase1, H3K9ac, H3K4me2, H3K27me3
		blood (K562)	H3K27ac, Max, H3K9ac, ETS1, Cmyc, HEY1, H2AZ, TAF1, PolII, H3K4me2, DNase1, H3K4me3, Egr1
		skin (NHDF-AD)	DNase1, H3K4me3
		muscle (HSMM)	H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H2AZ, H3K4me2, H3K4me3, H3K27me3, DNase1
		blood (GM12878)	PolII, H2AZ, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K27ac, H3K9ac, H3K4me3, H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, PolII, H3K4me3, Gabp
		monocytes (Monocytes-CD14+)	H3K27ac, H3K27me3, H3K4me3, H3K4me2, H3K4me1, DNase1
		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, PolII, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR3A	downstream gene variant	2792	ENST00000436743	NM_001127595.1, NM_001127593.1, NM_001127596.1	ENSP00000416607	3
FCGR3A	downstream gene variant	2882	ENST00000426740	?	ENSP00000410180	3
FCGR3A	downstream gene variant	2786	ENST00000367969	NM_000569.6, NM_001127592.1	ENSP00000356946	3
FCGR3A	downstream gene variant	2823	ENST00000367967	?	ENSP00000356944	3
FCGR3A	downstream gene variant	2790	ENST00000443193	?	ENSP00000392047	3
HSPA6	downstream gene variant	3430	ENST00000309758	NM_002155.3	ENSP00000310219	1
RPS23P10	upstream gene variant	669	ENST00000416185	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-25K21.6	ENST00000537821	?	?	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RPS23P10	ENST00000416185	?	1

Locus 143 (rs2290610)

SNiPAcard

Block annotations

Block info

genomic range	chr3:3,136,700-3,141,600
block size	4,901 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.028 [-0.759 – 1.12]	gene(s) hit or close-by	IL5RA, SNORA43
phastCons	μ = 0.097 [0 – 0.571]	eQTL gene(s)	–
GERP++	μ = -0.506 [-4.22 – 3.09]	potentially regulated gene(s)	–
CADD score	μ = 2.925 [0.581 – 11.03]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IL5RA	missense variant	ENST00000383846	NM_175728.2	ENSP00000373358	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000418488	?	ENSP00000388858	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000445701	?	ENSP00000398117	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000430514	NM_175724.2	ENSP00000400400	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000311981	NM_175725.2	ENSP00000309196	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000256452	NM_000564.4	ENSP00000256452	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000456302	NM_175727.2	ENSP00000392059	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000446632	NM_175726.3	ENSP00000412209	V/I	Gtt/Att	?	?	1
IL5RA	missense variant	ENST00000438560	NM_001243099.1	ENSP00000390753	V/I	Gtt/Att	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL5RA	downstream gene variant	833	ENST00000445701	?	ENSP00000398117	4
IL5RA	downstream gene variant	1816	ENST00000427088	?	ENSP00000391274	3
SNORA43	upstream gene variant	2997	ENST00000517240	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL5RA	ENST00000438560	NM_001243099.1	ENSP00000390753	5
IL5RA	ENST00000418488	?	ENSP00000388858	5
IL5RA	ENST00000456302	NM_175727.2	ENSP00000392059	5

IL5RA	ENST00000430514	NM_175724.2	ENSP00000400400	5
IL5RA	ENST00000311981	NM_175725.2	ENSP00000309196	5
IL5RA	ENST00000256452	NM_000564.4	ENSP00000256452	5
IL5RA	ENST00000383846	NM_175728.2	ENSP00000373358	5
IL5RA	ENST00000446632	NM_175726.3	ENSP00000412209	5
IL5RA	ENST00000445701	?	ENSP00000398117	1

Locus 144 (rs288326)

SNiPAcard

Block annotations

Block info

genomic range	chr2:183,702,964-183,703,336
block size	373 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 1.149 [0.053 – 2.246]	gene(s) hit or close-by	FRZB
phastCons	μ = 0.500 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 0.860 [-1.46 – 3.18]	potentially regulated gene(s)	–
CADD score	μ = 18.154 [4.308 – 32]	disease gene(s)	FRZB

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Osteoarthritis susceptibility 1	risk factor	ClinVar	RCV000005531.1	1
?	HGMD curated	HGMD	CM0910593	1
OSTEOARTHRITIS SUSCEPTIBILITY 1	OMIM curated	OMIM	MIM:605083	1

Disease gene annotation

gene	trait	source DB	source entry/link
FRZB	OSTEOARTHRITIS SUSCEPTIBILITY 1	OMIM	MIM:165720

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FRZB	missense variant	ENST00000295113	NM_001463.3	ENSP00000295113	W/R	Tgg/Cgg	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FRZB	ENST00000295113	NM_001463.3	ENSP00000295113	1

Locus 145 (rs3947)

SNiPAcard

Block annotations

Block info

genomic range	chr8:11,685,453-11,712,639
block size	27,187 bp
variant count	38 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.616 [-3.186 – 2.014]	gene(s) hit or close-by	CTSB, FDFT1, RP11-589N15.2
phastCons	μ = 0.019 [0 – 0.216]	eQTL gene(s)	AF131215.9, ANTXR1, CTSB, FAM167A, FAM167A-AS1, FDFT1, OR7E158P, RP11-148O21.3, RP11-148O21.6, RP11-419I17.1
GERP++	μ = -0.897 [-6.68 – 4.09]	potentially regulated gene(s)	BLK, CTSB, DEFB130, DEFB130, FAM167A, FAM66A, FDFT1, RP11-589N15.1, ZNF705C
CADD score	μ = 4.194 [0.347 – 11.93]	disease gene(s)	ANTXR1, BLK

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Parkinson's disease	<1.00×10-6	GWAS Catalog	22451204	1

Disease gene annotation

gene	trait	source DB	source entry/link
ANTXR1	GAPO SYNDROME	OMIM	MIM:230740
ANTXR1	HEMANGIOMA, CAPILLARY INFANTILE	OMIM	MIM:602089
BLK	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 11	OMIM	MIM:613375
ANTXR1	GAPO syndrome	OrphaNet	OrphaNet:2067
ANTXR1	Familial capillary hemangioma	OrphaNet	OrphaNet:91415
BLK	MODY syndrome	OrphaNet	OrphaNet:552

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CTSB	?	ENSG00000164733	pancreas	3.36×10-9 (p-value)	GTEx Portal V6	37
FAM167A-AS1	?	ENSG00000184608	pancreas	3.84×10-6 (p-value)	GTEx Portal V6	1
CTSB	?	ENSG00000164733	caudate basal ganglia	1.97×10-8 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	muscularis mucosae	1.44×10-21 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	spleen	1.03×10-7 (p-value)	GTEx Portal V6	31
CTSB	?	ENSG00000164733	tibial nerve	9.07×10-16 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	atrial appendage	1.32×10-11 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	transformed fibroblasts	4.17×10-10 (p-value)	GTEx Portal V6	38
RP11-148O21.3	?	ENSG00000254774	transformed fibroblasts	4.47×10-7 (p-value)	GTEx Portal V6	10
RP11-148O21.6	?	ENSG00000269954	transformed fibroblasts	3.03×10-8 (p-value)	GTEx Portal V6	10

CTSB	?	ENSG00000164733	tibial artery	4.71×10-28 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	breast	4.68×10-11 (p-value)	GTEx Portal V6	35
CTSB	?	ENSG00000164733	gastroesophageal junction	3.17×10-7 (p-value)	GTEx Portal V6	29
CTSB	?	ENSG00000164733	putamen	3.94×10-8 (p-value)	GTEx Portal V6	31
CTSB	?	ENSG00000164733	sun exposed skin	6.70×10-19 (p-value)	GTEx Portal V6	38
OR7E158P	?	ENSG00000254948	sun exposed skin	6.03×10-6 (p-value)	GTEx Portal V6	1
CTSB	?	ENSG00000164733	aorta	3.92×10-14 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	subcutaneous adipocytes	9.65×10-10 (p-value)	GTEx Portal V6	38
CTSB	ENST00000353047	ILMN_1696360	blood	2.56×10-9 (p-value)	MuTHER consortium	13
CTSB	ENST00000531551		adipocyte	2.14×10-8 (p-value)	MuTHER consortium	13
CTSB	ENST00000420692					
CTSB	ENST00000353047	ILMN_2359742	blood	1.38×10-8 (p-value)	MuTHER consortium	12
CTSB	ENST00000531551		adipocyte	1.04×10-6 (p-value)	MuTHER consortium	12
FDFT1	ENST00000618539	ILMN_2144088	adipocyte	3.12×10-6 (p-value)	MuTHER consortium	13
FDFT1	ENST00000615631					
FDFT1	ENST00000622850					
FDFT1	ENST00000528643					
FDFT1	ENST00000525954					
FDFT1	ENST00000443614					
FDFT1	ENST00000538689					
FDFT1	ENST00000528812					
FDFT1	ENST00000220584					
FDFT1	ENST00000525607					
FDFT1	ENST00000623368					
FDFT1	ENST00000530664					
FDFT1	ENST00000525900					
FDFT1	ENST00000446331					
FAM167A	ENST00000284486	ILMN_1687213	blood	8.36×10-8 (p-value)	MuTHER consortium	13
CTSB	?	ENSG00000164733	unexposed skin	1.38×10-6 (p-value)	GTEx Portal V6	23
CTSB	ENST00000353047	ILMN_1696360	monocyte	6.80×10-25 (p-value)	Zeller et al.	6
CTSB	ENST00000531551					
CTSB	ENST00000420692					
CTSB	ENST00000353047	ILMN_2359742	monocyte	4.67×10-16 (p-value)	Zeller et al.	4
CTSB	ENST00000531551					
FDFT1	ENST00000618539	ILMN_2144088	monocyte	3.93×10-19 (p-value)	Zeller et al.	6
FDFT1	ENST00000615631					
FDFT1	ENST00000622850					
FDFT1	ENST00000528643					
FDFT1	ENST00000525954					
FDFT1	ENST00000443614					
FDFT1	ENST00000538689					
FDFT1	ENST00000528812					
FDFT1	ENST00000220584					
FDFT1	ENST00000525607					
FDFT1	ENST00000623368					
FDFT1	ENST00000530664					

FDFT1	ENST00000525900						
FDFT1	ENST00000446331						
CTSB	?	ENSG00000164733	blood	4.99×10^{-8} (p-value)	GTEx Portal V6	30	
CTSB	?	ENSG00000164733	lung	1.65×10^{-6} (p-value)	GTEx Portal V6	19	
AF131215.9	?	ENSG00000269918	lung	4.39×10^{-7} (p-value)	GTEx Portal V6	4	
CTSB	?	ENSG00000164733	thyroid	3.73×10^{-6} (p-value)	GTEx Portal V6	16	
AF131215.9	?	ENSG00000269918	thyroid	2.78×10^{-6} (p-value)	GTEx Portal V6	1	
CTSB	?	ENSG00000164733	skeletal muscle	2.41×10^{-6} (p-value)	GTEx Portal V6	21	
CTSB	?	ENSG00000164733	sigmoid colon	1.52×10^{-6} (p-value)	GTEx Portal V6	17	
CTSB	ENST00000353047	ILMN_1696360	monocyte	9.33×10^{-6} (p-value)	Fairfax et al.	2	
CTSB	ENST00000531551		b-cell	2.08×10^{-5} (p-value)	Fairfax et al.	2	
CTSB	ENST00000420692						
FDFT1	ENST00000618539	ILMN_1741096	b-cell	1.27×10^{-9} (p-value)	Fairfax et al.	2	
FDFT1	ENST00000622850						
FDFT1	ENST00000615631						
FDFT1	ENST00000528643						
FDFT1	ENST00000525954						
FDFT1	ENST00000538689						
FDFT1	ENST00000443614						
FDFT1	ENST00000528812						
FDFT1	ENST00000220584						
FDFT1	ENST00000525900						
FDFT1	ENST00000530664						
FDFT1	ENST00000525607						
FDFT1	ENST00000623368						
FDFT1	ENST00000446331						
FAM167A	ENST00000284486	ILMN_1687213	monocyte	2.26×10^{-4} (p-value)	Fairfax et al.	2	
			b-cell	2.00×10^{-7} (p-value)	Fairfax et al.	2	
CTSB	?	ENSG00000164733	left ventricle	4.25×10^{-6} (p-value)	GTEx Portal V6	3	
CTSB	?	ENSG00000164733	nucleus accumbens	2.42×10^{-7} (p-value)	GTEx Portal V6	3	
RP11-419I17.1	?	ENSG00000270154	cortex	5.96×10^{-7} (p-value)	GTEx Portal V6	2	

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ANTXR1	ENST00000409829	ILMN_1811877	chr2	monocyte	3.17×10^{-13} (p-value)	Zeller et al.	2

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	2	no	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000507012	1	ENCP00000054977	FDFT1
		ENCP00000054989	RP11-589N15.1
ENCE00000506878	1	ENCP00000054958	BLK
ENCE00000506825	2	ENCP00000054951	FAM167A
		ENCP00000054980	CTSB
ENCE00000506836	1	ENCP00000054985	CTSB
		ENCP00000054953	FAM167A
		ENCP00000054979	CTSB
		ENCP00000055004	ZNF705C DEFB130 FAM66A DEFB130

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001386037 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (K562)	PolII, H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII
		blood (GM12878)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001719967 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	PolII, H3K36me3
		muscle (HSMM)	H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001386041 (CTCF binding site)	4	embryonic stem cell (H1ESC)	H3K36me3, PolII, Rad21, CTCF, DNase1
		HSMMtube	H3K36me3, DNase1
		blood (K562)	PolII, H3K36me3
		skin (NHDF-AD)	H3K36me3, DNase1
		muscle (HSMM)	H3K36me3
		liver (HepG2)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	PolII, H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	CTCF, DNase1, H3K4me1, H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF, H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII
		A549	H3K36me3
ENSR00001386042 (open chromatin region)	3	embryonic stem cell (H1ESC)	H3K36me3, PolII
		blood (K562)	PolII, H3K36me3
		breast (HMEC)	H3K36me3, H3K4me1
		muscle (HSMM)	H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII
		blood (GM12878)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K36me3, H3K4me1
ENSR00001386043 (TF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII, DNase1
		blood (K562)	PolII
		muscle (HSMM)	H3K36me3

		breast (HMEC)	H3K4me1
		endothelium (HUVEC)	H3K36me3, PolII
		blood (GM12878)	H3K36me3, PolII
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K4me1, H3K36me3
ENSR00001719968 (TF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII, DNase1
		blood (K562)	PolII
		breast (HMEC)	H3K4me1
		endothelium (HUVEC)	H3K36me3, PolII
		blood (GM12878)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K4me1, H3K36me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
FDFT1	7	ENST00000220584 ENST00000443614 ENST00000446331 ENST00000525607 ENST00000525777 ENST00000525900 ENST00000525954 ENST00000528643 ENST00000528812 ENST00000530664 ENST00000538689 ENST00000615631 ENST00000618539 ENST00000622850 ENST00000623368	hsa-miR-216b-5p hsa-miR-433-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTSB	downstream gene variant	555	ENST00000531551	?	ENSP00000436456	8
CTSB	downstream gene variant, upstream gene variant	205	ENST00000524654	?	ENSP00000432077	5
CTSB	downstream gene variant	1289	ENST00000534382	?	ENSP00000435260	5
CTSB	downstream gene variant	1289	ENST00000532656	?	ENSP00000431143	5
CTSB	downstream gene variant	1296	ENST00000527215	?	ENSP00000433379	5
CTSB	downstream gene variant, upstream gene variant	45	ENST00000526481	?	ENSP00000473301	18
CTSB	downstream gene variant	45	ENST00000353047	NM_001908.3	ENSP00000345672	6
CTSB	downstream gene variant	1335	ENST00000528965	?	ENSP00000433929	5
CTSB	downstream gene variant	1252	ENST00000531502	?	ENSP00000435886	11
CTSB	downstream gene variant	551	ENST00000453527	NM_147783.2	ENSP00000409917	8
CTSB	downstream gene variant	1224	ENST00000505496	?	ENSP00000435650	12
CTSB	downstream gene variant	1232	ENST00000524500	?	ENSP00000436074	12
CTSB	downstream gene variant	571	ENST00000345125	NM_147781.2	ENSP00000342070	8
CTSB	downstream gene variant, upstream gene variant	950	ENST00000525315	?	?	13
CTSB	downstream gene variant	284	ENST00000530640	?	ENSP00000435105	8
CTSB	downstream gene variant, upstream gene variant	1200	ENST00000532370	?	?	13
CTSB	downstream gene variant	601	ENST00000534510	?	ENSP00000434217	8

CTSB	downstream gene variant	586	ENST00000533455	NM_147780.2, NM_147782.2	ENSP00000432244	8
CTSB	downstream gene variant	700	ENST00000527243	?	ENSP00000434725	18
CTSB	downstream gene variant	701	ENST00000534149	?	ENSP00000436122	18
CTSB	downstream gene variant, upstream gene variant	251	ENST00000420692	?	?	11
CTSB	downstream gene variant	685	ENST00000526195	?	ENSP00000436627	18
CTSB	downstream gene variant, upstream gene variant	122	ENST00000530624	?	?	17
CTSB	downstream gene variant	325	ENST00000531089	?	ENSP00000433215	8
CTSB	downstream gene variant	150	ENST00000533572	?	ENSP00000433995	20
CTSB	downstream gene variant	723	ENST00000526645	?	ENSP00000431518	18
CTSB	downstream gene variant	135	ENST00000534636	?	ENSP00000436159	20
CTSB	downstream gene variant, upstream gene variant	291	ENST00000533110	?	?	21
CTSB	downstream gene variant	126	ENST00000525076	?	?	21
CTSB	downstream gene variant, upstream gene variant	87	ENST00000532409	?	?	22
CTSB	downstream gene variant	165	ENST00000530296	?	ENSP00000435074	20
CTSB	downstream gene variant	2072	ENST00000532392	?	ENSP00000432408	3
CTSB	downstream gene variant, upstream gene variant	328	ENST00000530290	?	?	22
FDFT1	downstream gene variant	1094	ENST00000525607	?	ENSP00000432551	8
FDFT1	downstream gene variant	966	ENST00000525954	?	?	8
FDFT1	downstream gene variant	819	ENST00000528643	NM_001287751.1	ENSP00000431649	8
FDFT1	downstream gene variant	1825	ENST00000529464	?	ENSP00000434770	4
FDFT1	downstream gene variant	973	ENST00000528812	NM_001287747.1	ENSP00000431749	8
FDFT1	downstream gene variant	723	ENST00000446331	?	?	8
FDFT1	downstream gene variant	1150	ENST00000443614	?	ENSP00000390367	8
FDFT1	downstream gene variant	629	ENST00000538689	NM_001287750.1, NM_001287744.1	ENSP00000444248	8
FDFT1	downstream gene variant	624	ENST00000220584	NM_004462.4	ENSP00000220584	8
FDFT1	downstream gene variant	1093	ENST00000530664	NM_001287749.1	ENSP00000432331	8
FDFT1	downstream gene variant	629	ENST00000623368	NM_001287745.1	ENSP00000485229	8
FDFT1	downstream gene variant	629	ENST00000618539	NM_001287742.1	ENSP00000480828	8
FDFT1	downstream gene variant	629	ENST00000622850	NM_001287748.1	ENSP00000484122	8
FDFT1	downstream gene variant	629	ENST00000615631	NM_001287743.1	ENSP00000481481	8
FDFT1	downstream gene variant	1810	ENST00000528729	?	?	4
FDFT1	downstream gene variant	1028	ENST00000525900	?	ENSP00000434714	8
FDFT1	downstream gene variant	1739	ENST00000525283	?	ENSP00000433985	4
FDFT1	downstream gene variant	3131	ENST00000532266	?	ENSP00000435900	2
FDFT1	upstream gene variant, downstream gene variant	1612	ENST00000529521	?	?	5
FDFT1	downstream gene variant	1848	ENST00000531249	?	?	1
FDFT1	downstream gene variant	1320	ENST00000525777	?	ENSP00000436069	6

RP11-589N15.2	downstream gene variant, upstream gene variant	4	ENST00000602711	?	?	27

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTSB	ENST00000530640	?	ENSP00000435105	21
CTSB	ENST00000531089	?	ENSP00000433215	21
CTSB	ENST00000534382	?	ENSP00000435260	1
CTSB	ENST00000532656	?	ENSP00000431143	1
CTSB	ENST00000527215	?	ENSP00000433379	1
CTSB	ENST00000532392	?	ENSP00000432408	1
CTSB	ENST00000528965	?	ENSP00000433929	1
CTSB	ENST00000505496	?	ENSP00000435650	3
CTSB	ENST00000531502	?	ENSP00000435886	3
CTSB	ENST00000524500	?	ENSP00000436074	3
CTSB	ENST00000532370	?	?	2
CTSB	ENST00000525315	?	?	2
CTSB	ENST00000526195	?	ENSP00000436627	4
CTSB	ENST00000527243	?	ENSP00000434725	4
CTSB	ENST00000525076	?	?	4
CTSB	ENST00000534149	?	ENSP00000436122	4
CTSB	ENST00000526645	?	ENSP00000431518	4
CTSB	ENST00000533572	?	ENSP00000433995	5
CTSB	ENST00000530296	?	ENSP00000435074	5
CTSB	ENST00000345125	NM_147781.2	ENSP00000342070	21
CTSB	ENST00000353047	NM_001908.3	ENSP00000345672	24
CTSB	ENST00000453527	NM_147783.2	ENSP00000409917	21
CTSB	ENST00000526481	?	ENSP00000473301	9
CTSB	ENST00000420692	?	?	17
CTSB	ENST00000534636	?	ENSP00000436159	5
CTSB	ENST00000534510	?	ENSP00000434217	21
CTSB	ENST00000531551	?	ENSP00000436456	21
CTSB	ENST00000533455	NM_147780.2, NM_147782.2	ENSP00000432244	21
CTSB	ENST00000530624	?	?	11
CTSB	ENST00000530290	?	?	4
CTSB	ENST00000532409	?	?	4
CTSB	ENST00000533110	?	?	3
FDFT1	ENST00000525900	?	ENSP00000434714	8
FDFT1	ENST00000525777	?	ENSP00000436069	8
FDFT1	ENST00000528812	NM_001287747.1	ENSP00000431749	8
FDFT1	ENST00000443614	?	ENSP00000390367	8

FDFT1	ENST00000443614	?	ENSP00000390367	8
FDFT1	ENST00000538689	NM_001287750.1, NM_001287744.1	ENSP00000444248	8
FDFT1	ENST00000446331	?	?	8
FDFT1	ENST00000528729	?	?	1
FDFT1	ENST00000525607	?	ENSP00000432551	8
FDFT1	ENST00000530664	NM_001287749.1	ENSP00000432331	8
FDFT1	ENST00000623368	NM_001287745.1	ENSP00000485229	8
FDFT1	ENST00000220584	NM_004462.4	ENSP00000220584	8
FDFT1	ENST00000529464	?	ENSP00000434770	1
FDFT1	ENST00000525283	?	ENSP00000433985	1
FDFT1	ENST00000525954	?	?	8
FDFT1	ENST00000532266	?	ENSP00000435900	1
FDFT1	ENST00000528643	NM_001287751.1	ENSP00000431649	8
FDFT1	ENST00000622850	NM_001287748.1	ENSP00000484122	8
FDFT1	ENST00000618539	NM_001287742.1	ENSP00000480828	8
FDFT1	ENST00000615631	NM_001287743.1	ENSP00000481481	8

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTSB	ENST00000530640	?	ENSP00000435105	5
CTSB	ENST00000531089	?	ENSP00000433215	5
CTSB	ENST00000345125	NM_147781.2	ENSP00000342070	5
CTSB	ENST00000353047	NM_001908.3	ENSP00000345672	8
CTSB	ENST00000453527	NM_147783.2	ENSP00000409917	5
CTSB	ENST00000526481	?	ENSP00000473301	8
CTSB	ENST00000534510	?	ENSP00000434217	5
CTSB	ENST00000531551	?	ENSP00000436456	5
CTSB	ENST00000533455	NM_147780.2, NM_147782.2	ENSP00000432244	5

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTSB	ENST00000420692	?	6

Locus 146 (rs2905736)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,430,060-31,816,059
block size	386,000 bp
variant count	45 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.391 [-3.764 – 1.759]	**gene(s) hit or close-by**	AL662801.1, ATP6V1G2, ATP6V1G2-DDX39B, BAG6, C6orf47, C6orf47-AS1, CLIC1, CSNK2B, CSNK2B-LY6G5B--991, DDX39B, DDX39B-AS1, GPANK1, HCP5, LST1, LTA, LTB, LY6G5B, MCCD1, MICB, MSH5, MSH5-SAPCD1, NCR3, NFKBIL1, PPIAP9, PRRC2A, RPL15P4, SNORA38, SNORD117, SNORD84, TNF, VARS, XXbac-BPG16N22.5, XXbac-BPG181B23.6, Y_RNA, Y_RNA, Y_RNA
phastCons	μ = 0.059 [0 – 0.855]	**eQTL gene(s)**	CDSN, DDR1, HLA-DRB6, LST1, MICA, MICB, NOTCH4, PSORS1C2, RNF5, TNXA, XXbac-BPG181B23.7
GERP++	μ = -0.549 [-6.65 – 4.18]	**potentially regulated gene(s)**	AIF1, AIF1, AIF1, AIF1, AIF1, AIF1, ATP6V1G2, ATP6V1G2, ATP6V1G2, ATP6V1G2, ATP6V1G2, ATP6V1G2, ATP6V1G2, C2, C2, C2, C2, C2, C2, C2, C2, C4A, C4A, C4A, C4A, C4A, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, CFB, CFB, CFB, CFB, CFB, CFB, CFB, CFB, CFB, CLIC1, CLIC1, CLIC1, CLIC1, CLIC1, CLIC1, CLIC1, CLIC1, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CYP21A1P, CYP21A1P, CYP21A1P, CYP21A1P, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DHFRP2, DHFRP2, DHFRP2, DHFRP2, DHFRP2, EHMT2, EHMT2, EHMT2, EHMT2, EHMT2, EHMT2, EHMT2, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCP5, HCP5, HCP5, HCP5, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, LSM2, LSM2, LSM2, LSM2, LSM2, LSM2, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LY6G5B, LY6G5B, LY6G5B, LY6G5B, LY6G5B, LY6G5B, LY6G5B, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, MICA, MICA, MICA, MICA, MICA, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, STK19, STK19, STK19, STK19, STK19, STK19, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, VARS, VARS, VARS, VARS, VARS, VARS, VARS, XXbac-BPG16N22.5, XXbac-BPG181B23.6, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.12, XXbac-BPG248L24.13, XXbac-BPG299F13.15, ZBTB12, ZBTB12, ZBTB12, ZBTB12, ZBTB12
CADD score	μ = 4.116 [0.099 – 15.39]	**disease gene(s)**	NCR3, , , ,

Disease gene annotation

gene	trait	source DB	source entry/link
NCR3	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...]	OMIM	MIM:608579
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
NFKBIL1	RHEUMATOID ARTHRITIS	OMIM	MIM:180300
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561

	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380	OMIM
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790	OMIM
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
LTA	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988	OMIM
LTA	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	orphanet
CDSN	Hypotrichosis simplex of the scalp	OrphaNet	OrphaNet:90368	orphanet
CDSN	Generalized peeling skin syndrome type B	OrphaNet	OrphaNet:263553	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICB	?	ENSG00000204516	muscularis mucosae	1.53×10^{-8} (p-value)	GTEx Portal V6	44
MICB	?	ENSG00000204516	lung	6.83×10^{-14} (p-value)	GTEx Portal V6	44
MICA	?	ENSG00000204520	lung	5.56×10^{-6} (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	transformed fibroblasts	1.64×10^{-10} (p-value)	GTEx Portal V6	44
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	1.54×10^{-6} (p-value)	GTEx Portal V6	13

XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	3.83×10-7 (p-value)	GTEx Portal V6	20
MICB	?	ENSG00000204516	tibial artery	1.47×10-6 (p-value)	GTEx Portal V6	20
MICB	?	ENSG00000204516	breast	1.70×10-8 (p-value)	GTEx Portal V6	23
MICB	?	ENSG00000204516	blood	1.27×10-22 (p-value)	GTEx Portal V6	44
XXbac-BPG181B23.7	?	ENSG00000272221	blood	2.73×10-6 (p-value)	GTEx Portal V6	12
MICB	?	ENSG00000204516	thyroid	6.43×10-17 (p-value)	GTEx Portal V6	44
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	4.07×10-6 (p-value)	GTEx Portal V6	2
DDR1	?	ENSG00000204580	thyroid	7.87×10-6 (p-value)	GTEx Portal V6	2
HLA-DRB6	?	ENSG00000229391	thyroid	6.94×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	skeletal muscle	4.97×10-9 (p-value)	GTEx Portal V6	44
RNF5	?	ENSG00000204308	skeletal muscle	3.42×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	transverse colon	9.05×10-7 (p-value)	GTEx Portal V6	32
MICB	?	ENSG00000204516	sun exposed skin	4.30×10-11 (p-value)	GTEx Portal V6	44
MICA	?	ENSG00000204520	sun exposed skin	9.25×10-7 (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	7.10×10-6 (p-value)	GTEx Portal V6	1
CDSN	?	ENSG00000204539	sun exposed skin	7.80×10-6 (p-value)	GTEx Portal V6	5
MICB	?	ENSG00000204516	left ventricle	1.04×10-11 (p-value)	GTEx Portal V6	44
MICB	?	ENSG00000204516	subcutaneous adipocytes	1.42×10-13 (p-value)	GTEx Portal V6	44
NOTCH4	?	ENSG00000204301	subcutaneous adipocytes	1.45×10-6 (p-value)	GTEx Portal V6	27
MICB	?	ENSG00000204516	visceral adipocytes	5.67×10-10 (p-value)	GTEx Portal V6	35
MICB	?	ENSG00000204516	tibial nerve	3.67×10-10 (p-value)	GTEx Portal V6	44
MICA	?	ENSG00000204520	tibial nerve	7.32×10-6 (p-value)	GTEx Portal V6	1
LST1	?	ENSG00000204482	tibial nerve	3.11×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	esophagus mucosa	5.79×10-6 (p-value)	GTEx Portal V6	15
XXbac-BPG181B23.7	?	ENSG00000272221	esophagus mucosa	3.48×10-6 (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	adrenal gland	1.58×10-8 (p-value)	GTEx Portal V6	44
XXbac-BPG181B23.7	?	ENSG00000272221	stomach	2.95×10-6 (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	blood	1.98×10-3 (q-value)	SeeQTL DB (HapMap)	2
PSORS1C2	ENST00000259845	ILMN_1653447	monocyte	2.10×10-5 (p-value)	Fairfax et al.	4
MICB	ENST00000399150	ILMN_1708006	monocyte	2.79×10-14 (p-value)	Fairfax et al.	4
MICB	ENST00000538442		b-cell	5.86×10-33 (p-value)	Fairfax et al.	4
MICB	ENST00000252229					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)

ENCP00000048021

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444839	1	ENCP00000048065	CSNK2B GPANK1 GPANK1 GPANK1 CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048018	XXbac-BPG181B23.6
		ENCP00000048069	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048107	VARS VARS VARS VARS VARS VARS VARS
		ENCP00000048002	XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10
		ENCP00000048013	HCP5 MICA MICA HCP5 MICA HCP5 MICA MICA HCP5
		ENCP00000048038	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3
		ENCP00000048005	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000047994	XXbac-BPG299F13.15
		ENCP00000048017	HCP5 HCP5 HCP5 HCP5
		ENCP00000048050	PRRC2A PRRC2A

	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000047997	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000048039	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3
ENCP00000047996	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000047991	HCG27 HCG27 HCG27 HCG27 HCG27 HCG27 HCG27
ENCP00000048047	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048066	LY6G5B LY6G5B LY6G5B LY6G5B LY6G5B LY6G5B LY6G5B
ENCP00000048046	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048045	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048109	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048033	TNF

			TNF TNF TNF TNF TNF TNF TNF
		ENCP00000048003	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000047998	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
		ENCP00000048004	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000048031	ATP6V1G2 ATP6V1G2 ATP6V1G2 ATP6V1G2 ATP6V1G2 ATP6V1G2 ATP6V1G2
		ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000048063	GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048006	XXbac-BPG248L24.12
		ENCP00000048020	MICB MICB MICB MICB MICB MICB MICB MICB
ENCE00000445135	1	ENCP00000048115	C6orf48 C6orf48 C6orf48 C6orf48 C6orf48
		ENCP00000048132	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000048070	LY6G5C LY6G5C

	LY6G5C
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ENCP00000048140	CFB
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ENCP00000048090	CLIC1
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ENCP00000048135	C2
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ENCP00000048111	LSM2
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ENCP00000048130	ZBTB12
	ZBTB12
	ZBTB12
	ZBTB12
	ZBTB12
ENCP00000048102	VARS
	VARS
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ENCP00000048123	EHMT2
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	EHMT2
ENCP00000048156	C4A
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	C4A
	C4A
	C4A
ENCP00000048092	CLIC1
	CLIC1
	CLIC1
	CLIC1
	MSH5
	CLIC1
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	MSH5
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			MSH5
		ENCP00000048103	VARS VARS VARS VARS VARS VARS VARS
ENCE00000444818	1	ENCP00000048065	CSNK2B GPANK1 GPANK1 GPANK1 CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048001	XXbac-BPG248L24.13
		ENCP00000048008	DHFRP2 DHFRP2 DHFRP2 DHFRP2 DHFRP2
		ENCP00000048041	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
		ENCP00000048069	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048013	HCP5 MICA MICA HCP5 MICA HCP5 MICA MICA HCP5
		ENCP00000048050	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000048017	HCP5 HCP5 HCP5 HCP5
		ENCP00000047997	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
		ENCP00000048023	DDX39B

	DDX39B
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ENCP00000047991	HCG27
	HCG27
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	HCG27
	HCG27
	HCG27
ENCP00000048104	VARS
	VARS
	VARS
	VARS
	VARS
	VARS
	VARS
ENCP00000048052	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
ENCP00000048110	VARS
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ENCP00000048109	VARS
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	VARS
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ENCP00000048051	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
ENCP00000048150	STK19
	STK19
	STK19
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	C6orf48
ENCP00000047998	HLA-C
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ENCP00000048035	LST1
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ENCP00000048043	AIF1
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ENCP00000048007	HLA-B
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ENCP00000048157	CYP21A1P
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ENCP00000048063	GPANK1
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ENCP00000048048	PRRC2A
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ENCP00000048020	MICB
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ENCP00000048037	LST1
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ENCP00000048134	C2
	C2
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		ENCP00000048040	UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1
ENCE00000444842	1	ENCP00000048041	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
		ENCP00000048013	HCP5 MICA MICA HCP5 MICA HCP5 MICA MICA HCP5
		ENCP00000048050	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000047997	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
		ENCP00000048039	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3
		ENCP00000048046	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000048109	VARS VARS VARS VARS VARS VARS VARS
		ENCP00000048051	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000048150	STK19 STK19 STK19

	STK19 STK19 STK19
ENCP00000048003	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048033	TNF TNF TNF TNF TNF TNF TNF TNF
ENCP00000048035	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000048116	C6orf48 C6orf48 C6orf48 C6orf48 C6orf48
ENCP00000048043	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048048	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048093	MSH5 MSH5 MSH5 MSH5 MSH5 MSH5 MSH5 MSH5
ENCP00000048006	XXbac-BPG248L24.12
ENCP00000048134	C2 C2 C2 C2 C2 C2 C2 C2
ENCP00000048037	LST1 LST1 LST1

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ENCP00000048020	MICB
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ENCP00000048065	CSNK2B
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	GPANK1
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ENCP00000048069	LY6G5C
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ENCP00000047991	HCG27
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	ENCP00000048104	VARS
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	ENCP00000048052	PRRC2A
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		PRRC2A
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		PRRC2A
		PRRC2A
	ENCP00000048110	VARS
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		VARS
		VARS
	ENCP00000047998	HLA-C
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	ENCP00000048157	CYP21A1P
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		CYP21A1P
	ENCP00000048063	GPANK1
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		GPANK1
	ENCP00000048040	UQCRHP1
		UQCRHP1
		UQCRHP1
		UQCRHP1
		UQCRHP1
		UQCRHP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000129758 (promoter flanking region)	2	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, DNase1, TAF7, PolII, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K4me2, H2AZ, H3K36me3
		blood (K562)	H3K27me3, Cfos
		skin (NHDF-AD)	DNase1, H3K27me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac

		A549	DNase1, H3K4me3
		skin (NHEK)	H3K4me3, DNase1, H3K4me2, H3K9ac
ENSR00001703053 (enhancer)	1	Osteobl	H3K4me2
		nervous (NH-A)	H3K27me3
		blood (K562)	H3K27me3
ENSR00000264405 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, Nrsf, CTCF, USF1, DNase1
		HSMMtube	CTCF
		Osteobl	CTCF
		blood (K562)	CTCF, Nrsf, PolII, Max, Rad21, USF1
		blood (DND-41)	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		endothelium (HUVEC)	Max, CTCF
		liver (HepG2)	Nrsf, Rad21, USF1, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	CTCF
ENSR00000264406 (promoter)	1	embryonic stem cell (H1ESC)	H3K9ac, H3K36me3, H3K27me3, PolII, Rad21, TAF7, CTCF, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	CTCF, H3K4me2, DNase1, H3K27me3, H2AZ
		blood (K562)	Max, ZBTB7A, Rad21, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, CTCF, DNase1, H3K4me3, Ini1, H3K27me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K4me2
		liver (HepG2)	DNase1, H3K27me3, PolII, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3
		lung (IMR90)	H3K4me2, H3K4me3, CTCF
		blood (GM12878)	PolII, H2AZ, DNase1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII
		monocytes (Monocytes-CD14+)	H3K9ac, H4K20me1, H3K4me3, H3K27ac, H3K4me2, DNase1, H3K4me1, CTCF
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, CTCF, DNase1
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000487848 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, PolII, TAF7, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	Max, ZBTB7A, CTCF, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2
		liver (HepG2)	PolII, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4me2
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K4me2, H3K4me3, DNase1, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, CTCF, H3K4me1
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K4me3
		endothelium (HUVEC)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac
ENSR00001494568 (promoter flanking region)	2	liver (HepG2)	H3K27me3
		HSMMtube	DNase1
		blood (K562)	H3K27me3
ENSR00000487851 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	H3K27me3, Rad21
		liver (HepG2)	H3K27me3

		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00000487856 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, Nrsf, Sin3Ak20, H3K27ac, Yy1, HDAC2, TAF1, H3K4me2, SP1, H3K36me3, H3K9ac, H3K4me3, TAF7, Gabp, H4K5ac, H3K27me3, CTCF, PolII
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		blood (K562)	Yy1, Ini1, Brg1, THAP1, H3K27ac, Max, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Nrsf, Cmyc, PolII, H4K20me1, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K27me3, H3K36me3, H3K4me3, Cmyc, Gabp, Yy1, Nrsf, H4K20me1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac
		lung (IMR90)	DNase1, H4K8ac, H3K14ac, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K56ac
		blood (GM12878)	TAF1, Nrsf, PolII, H2AZ, DNase1, Cmyc, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		nervous (NH-A)	DNase1, H3K36me3, H3K9ac, H4K20me1, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	H3K36me3, H3K4me3, H3K4me2, DNase1, H3K4me1, H3K27ac, H4K20me1, H3K9ac, PolII
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H4K20me1, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K27me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K9ac, Nrsf, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, Cmyc
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1, Cmyc, H3K36me3
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000487858 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21, H3K36me3
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF, H3K36me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF, H3K36me3
		liver (HepG2)	H3K36me3, Rad21, CTCF
		lung (IMR90)	H3K36me3, CTCF
		blood (GM12878)	CTCF, Rad21
		skin (NHEK)	CTCF, H3K36me3
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		A549	H3K36me3
ENSR00000487863 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (K562)	H3K4me1, PU1, FOSL1, H3K27me3, PolII, Ini1, Brg1, DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K4me1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		blood (GM12878)	H3K4me1, H3K27ac
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00001494573 (promoter)	1	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K27me3, H3K36me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	EBF1, POU2F2, Cmyc, PolII, DNase1, Egr1, Yy1, Gabp, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3

		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me2
ENSR00000487869 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, Nrsf
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	PolII, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K36me3
		skin (NHEK)	DNase1
ENSR00000264430 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27ac, Yy1, TAF1, H3K4me2, SP1, USF1, H3K36me3, H3K9ac, H3K4me3, SIX5, Sin3Ak20, H4K5ac, H3K27me3, CTCF, PolII, TAF7
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K4me2, H3K9ac
		blood (K562)	Ini1, H2AZ, TAF1, ELF1, Cjun, PolII, GTF2B, H3K4me2, H3K36me3, DNase1, H3K4me3, HEY1, H3K79me2, Brg1, Egr1, FOSL1, SIX5, H3K27ac, Max, TAF7, H3K4me1, E2F6, H3K9ac, Gabp, Jund, ETS1, USF1, Cmyc, H3K27me3, Yy1
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2
		liver (HepG2)	DNase1, Gabp, Yy1, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3
		blood (GM12878)	H3K9ac, H3K36me3, H3K4me2, H3K27ac, Tr4, SIX5, PolII, Srf, H2AZ, ETS1, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3
		lung (IMR90)	H3K36me3, H3K4me3, H3K9ac, H3K4me2, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K36me3, DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K4me2, PolII, DNase1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000487898 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K4me3, PolII, Rad21, Jund, Nrsf, Sin3Ak20, CTCF, H3K27ac, ATF3, Yy1, H3K4me1, TAF1, H3K4me2, SP1, USF1, H3K9ac
		HSMMtube	H3K9ac, DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2
		blood (K562)	H3K4me3, Jund, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Cjun, Cfos, Cmyc, PolII, H4K20me1, H3K4me2, CTCF, H3K4me1, H3K36me3, H3K9ac, E2F6, Yy1, Ini1, Brg1, Egr1, FOSL1, H3K27ac, Max, SP2, ZBTB7A, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac, CTCF, H3K4me1, H3K36me3
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K4me2, HNF4A, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3, p300, H3K4me1, FOSL2, Jund, HNF4G, FOXA2, PolII, HDAC2, H3K79me2, TAF1, RXRA, ELF1
		lung (IMR90)	H3K9ac, H3K4ac, DNase1, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3
		blood (GM12878)	H3K27ac, Cfos, H3K4me2, H3K9ac, H3K36me3, H3K4me3, H3K79me2, PolII, H2AZ, H3K4me1, DNase1, CTCF, Yy1, ELF1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me2, H3K4me3, H3K36me3, DNase1, H3K9ac, H3K27ac, H3K4me1, CTCF
		NHLF	H3K27ac, H3K36me3, DNase1, H3K4me3, H3K9ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3, CTCF
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, Cmyc, CTCF, H3K36me3, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, Cmyc, CTCF, PolII, H3K27ac, Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K4me1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
HCP5	1	ENST00000414046 ENST00000467369 ENST00000541196	hsa-miR-181a-5p hsa-miR-181b-5p hsa-miR-181d-5p hsa-miR-22-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL662801.1	upstream gene variant	2780	ENST00000617927	?	?	1
ATP6V1G2	downstream gene variant, upstream gene variant	1436	ENST00000483251	NM_138282.2	ENSP00000419698	3
ATP6V1G2	downstream gene variant, upstream gene variant	1482	ENST00000483170	?	?	3
ATP6V1G2	downstream gene variant, upstream gene variant	1474	ENST00000415099	?	ENSP00000390148	4
ATP6V1G2	downstream gene variant, upstream gene variant	660	ENST00000303892	NM_130463.3	ENSP00000302194	3
ATP6V1G2	downstream gene variant, upstream gene variant	1954	ENST00000481998	?	?	3
ATP6V1G2	downstream gene variant, upstream gene variant	660	ENST00000376151	NM_001204078.1	ENSP00000365321	3
ATP6V1G2-DDX39B	downstream gene variant, upstream gene variant	4765	ENST00000480131	?	ENSP00000420191	2
ATP6V1G2-DDX39B	upstream gene variant	4995	ENST00000475917	?	?	1
BAG6	downstream gene variant, upstream gene variant	1459	ENST00000469182	?	?	2
BAG6	downstream gene variant, upstream gene variant	1567	ENST00000462875	?	?	3
BAG6	downstream gene variant, upstream gene variant	1951	ENST00000438149	?	ENSP00000410280	2
BAG6	downstream gene variant, upstream gene variant	598	ENST00000465348	?	?	3
BAG6	downstream gene variant	768	ENST00000456286	?	ENSP00000406436	2
BAG6	downstream gene variant	573	ENST00000428326	?	ENSP00000413561	2
BAG6	upstream gene variant	1835	ENST00000462682	?	?	1
BAG6	upstream gene variant	4259	ENST00000464126	?	?	1
BAG6	downstream gene variant	660	ENST00000424176	?	ENSP00000391783	2
BAG6	downstream gene variant	571	ENST00000456622	?	ENSP00000401031	2
BAG6	downstream gene variant	571	ENST00000434444	?	ENSP00000394856	2
BAG6	downstream gene variant	1892	ENST00000454165	?	ENSP00000396503	2
BAG6	downstream gene variant	584	ENST00000424480	?	ENSP00000411043	2
BAG6	upstream gene variant	3861	ENST00000422948	?	ENSP00000413225	1
BAG6	downstream gene variant	574	ENST00000451898	?	ENSP00000391613	2
BAG6	downstream gene variant	580	ENST00000433828	?	ENSP00000388244	2
BAG6	downstream gene variant	2961	ENST00000437771	?	ENSP00000397978	1
BAG6	downstream gene variant	571	ENST00000452994	?	ENSP00000404971	2
BAG6	downstream gene variant, upstream gene variant	1517	ENST00000453833	?	ENSP00000406434	2
BAG6	downstream gene variant	686	ENST00000441054	?	ENSP00000406302	2
BAG6	upstream gene variant	3570	ENST00000441793	?	ENSP00000416633	1
BAG6	downstream gene variant, upstream gene variant	482	ENST00000470875	?	?	3
C6orf47	upstream gene variant	4933	ENST00000375911	NM_021184.3	ENSP00000365076	1
C6orf47-AS1	downstream gene variant	4984	ENST00000422049	?	?	1
CLIC1	upstream gene variant	3182	ENST00000375784	NM_001288.4	ENSP00000364940	1
CLIC1	upstream gene variant	2431	ENST00000375780	NM_001287593.1	ENSP00000364935	1

CLIC1	upstream gene variant	3204	ENST00000616760	NM_001287594.1	ENSP00000479808 1
CLIC1	upstream gene variant	3177	ENST00000375779	?	ENSP00000364934 1
CSNK2B	upstream gene variant	382	ENST00000465481	?	? 1
CSNK2B	upstream gene variant	359	ENST00000375882	NM_001320.6, NM_001282385.1	ENSP00000365042 1
CSNK2B	upstream gene variant	389	ENST00000481269	?	? 1
CSNK2B	upstream gene variant	376	ENST00000468255	?	? 1
CSNK2B	upstream gene variant	396	ENST00000475875	?	? 1
CSNK2B	upstream gene variant	650	ENST00000375866	?	ENSP00000365026 1
CSNK2B	upstream gene variant	587	ENST00000375865	?	ENSP00000365025 1
CSNK2B-LY6G5B--991	upstream gene variant	175	ENST00000618754	?	ENSP00000478610 1
CSNK2B-LY6G5B--991	upstream gene variant	175	ENST00000622499	?	ENSP00000483024 1
CSNK2B-LY6G5B--991	upstream gene variant	397	ENST00000375880	?	ENSP00000365040 1
CSNK2B-LY6G5B--991	upstream gene variant	3413	ENST00000409691	?	ENSP00000387322 1
CSNK2B-LY6G5B--991	upstream gene variant	1115	ENST00000617558	?	ENSP00000483989 1
DDX39B	upstream gene variant	719	ENST00000419338	?	ENSP00000410313 2
DDX39B	upstream gene variant	1354	ENST00000458640	NM_080598.5	ENSP00000416269 1
DDX39B	upstream gene variant	185	ENST00000428098	?	ENSP00000392672 2
DDX39B	upstream gene variant	1354	ENST00000396172	NM_004640.6	ENSP00000379475 1
DDX39B	upstream gene variant	1138	ENST00000417023	?	ENSP00000406422 1
DDX39B	upstream gene variant	174	ENST00000484566	?	ENSP00000436220 1
DDX39B	upstream gene variant	3905	ENST00000474961	?	? 1
DDX39B	upstream gene variant	1667	ENST00000376177	?	ENSP00000365347 2
DDX39B	upstream gene variant	4714	ENST00000462421	?	? 1
DDX39B	downstream gene variant, upstream gene variant	200	ENST00000419020	?	ENSP00000405245 3
DDX39B	upstream gene variant	3978	ENST00000478365	?	? 1
DDX39B	upstream gene variant	202	ENST00000481456	?	? 2
DDX39B	downstream gene variant, upstream gene variant	212	ENST00000418897	?	ENSP00000399841 3
DDX39B	upstream gene variant	222	ENST00000431908	?	ENSP00000408000 2
DDX39B	downstream gene variant, upstream gene variant	214	ENST00000428450	?	ENSP00000405707 3
DDX39B	upstream gene variant	672	ENST00000427214	?	ENSP00000399371 2
DDX39B	downstream gene variant, upstream gene variant	217	ENST00000456976	?	ENSP00000393984 3
DDX39B	upstream gene variant	266	ENST00000456662	?	ENSP00000416350 2
DDX39B	downstream gene variant, upstream gene variant	474	ENST00000449757	?	ENSP00000409426 3
DDX39B	upstream gene variant	186	ENST00000462256	?	? 2
DDX39B	upstream gene variant	1401	ENST00000482195	?	? 1
DDX39B-AS1	upstream gene variant, downstream gene variant	170	ENST00000416684	?	? 2
DDX39B-AS1	upstream gene variant, downstream	101	ENST00000420520	?	? 2

	gene variant					
GPANK1	upstream gene variant	315	ENST00000375896	NM_033177.3	ENSP00000365060	1
GPANK1	upstream gene variant	75	ENST00000375893	NM_001199240.1	ENSP00000365057	1
GPANK1	upstream gene variant	263	ENST00000458083	?	ENSP00000395307	1
GPANK1	upstream gene variant	515	ENST00000445768	?	ENSP00000409349	1
GPANK1	upstream gene variant	520	ENST00000456540	?	ENSP00000395484	1
HCP5	upstream gene variant, downstream gene variant	488	ENST00000467369	?	?	2
HCP5	upstream gene variant, downstream gene variant	182	ENST00000460889	?	?	2
HCP5	upstream gene variant	899	ENST00000541196	?	?	1
LST1	downstream gene variant	1825	ENST00000376092	?	ENSP00000365260	1
LST1	downstream gene variant	1578	ENST00000376089	?	ENSP00000365257	1
LST1	downstream gene variant	1578	ENST00000376093	NM_007161.3	ENSP00000365261	1
LST1	downstream gene variant	1578	ENST00000418507	NM_205838.2	ENSP00000405900	1
LST1	downstream gene variant	698	ENST00000433492	?	ENSP00000399869	2
LST1	downstream gene variant	1578	ENST00000396101	?	ENSP00000379408	1
LST1	downstream gene variant	1578	ENST00000303757	NM_205840.2	ENSP00000303649	1
LST1	downstream gene variant	1578	ENST00000376090	?	ENSP00000365258	1
LST1	downstream gene variant	1578	ENST00000376099	?	ENSP00000365267	1
LST1	downstream gene variant	1578	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	downstream gene variant	587	ENST00000419073	?	?	2
LST1	downstream gene variant	1811	ENST00000490742	?	ENSP00000418578	1
LST1	downstream gene variant	1730	ENST00000464044	?	ENSP00000417645	1
LST1	downstream gene variant	1730	ENST00000376110	?	ENSP00000365278	1
LST1	downstream gene variant	1579	ENST00000376086	NM_001166538.1	ENSP00000365254	1
LST1	downstream gene variant	1677	ENST00000211921	?	ENSP00000211921	1
LST1	downstream gene variant	1578	ENST00000464526	?	?	1
LST1	downstream gene variant	1861	ENST00000396112	?	ENSP00000379418	1
LST1	downstream gene variant	1677	ENST00000339530	NM_205837.2	ENSP00000339201	1
LST1	downstream gene variant	1578	ENST00000376096	?	ENSP00000365264	1
LST1	downstream gene variant	1730	ENST00000460834	?	?	1
LTA	downstream gene variant	1657	ENST00000489638	?	?	3
LTA	downstream gene variant	1657	ENST00000454783	NM_001159740.2	ENSP00000403495	3
LTA	downstream gene variant	1657	ENST00000418386	NM_000595.3	ENSP00000413450	3
LTA	downstream gene variant	1657	ENST00000471842	?	?	3
LTB	downstream gene variant	1187	ENST00000483972	?	?	4
LTB	downstream gene variant	1217	ENST00000429299	NM_002341.1	ENSP00000410481	4
LTB	downstream gene variant	1217	ENST00000446745	NM_009588.1	ENSP00000416113	4
LTB	downstream gene variant	1286	ENST00000482429	?	?	4

LY6G5B	upstream gene variant	4674	ENST00000409525	?	ENSP00000386365	1
LY6G5B	upstream gene variant	4462	ENST00000375864	NM_021221.2	ENSP00000365024	1
MCCD1	upstream gene variant	3624	ENST00000376191	NM_001011700.2	ENSP00000365362	2
MICB	upstream gene variant, downstream gene variant	59	ENST00000399150	NM_001289161.1	ENSP00000382103	10
MICB	upstream gene variant, downstream gene variant	10	ENST00000538442	NM_001289160.1	ENSP00000442345	10
MICB	upstream gene variant, downstream gene variant	59	ENST00000252229	NM_005931.4	ENSP00000252229	10
MICB	downstream gene variant	320	ENST00000494577	?	?	5
MSH5	upstream gene variant, downstream gene variant	242	ENST00000425703	?	ENSP00000402842	2
MSH5	upstream gene variant	256	ENST00000482280	?	?	1
MSH5	upstream gene variant	202	ENST00000375750	NM_172166.3	ENSP00000364903	1
MSH5	upstream gene variant	4229	ENST00000450148	?	ENSP00000394971	1
MSH5	upstream gene variant	477	ENST00000423982	?	ENSP00000406352	1
MSH5	upstream gene variant	199	ENST00000375755	NM_002441.4	ENSP00000364908	1
MSH5	upstream gene variant	1497	ENST00000463144	?	ENSP00000419648	1
MSH5	upstream gene variant	272	ENST00000375703	NM_172165.3	ENSP00000364855	1
MSH5	upstream gene variant	272	ENST00000375740	NM_025259.5	ENSP00000364892	1
MSH5	upstream gene variant, downstream gene variant	256	ENST00000463094	?	?	2
MSH5	upstream gene variant	4482	ENST00000497269	?	ENSP00000419131	1
MSH5-SAPCD1	upstream gene variant	271	ENST00000493662	?	ENSP00000417871	1
NCR3	downstream gene variant, upstream gene variant	467	ENST00000491161	?	?	2
NCR3	downstream gene variant, upstream gene variant	846	ENST00000376072	NM_001145467.1	ENSP00000365240	2
NCR3	downstream gene variant, upstream gene variant	467	ENST00000376073	NM_001145466.1	ENSP00000365241	2
NCR3	downstream gene variant, upstream gene variant	869	ENST00000376071	?	ENSP00000365239	2
NCR3	downstream gene variant, upstream gene variant	467	ENST00000340027	NM_147130.2	ENSP00000342156	2
NCR3	downstream gene variant, upstream gene variant	467	ENST00000495600	?	?	2
NFKBIL1	upstream gene variant, downstream gene variant	3779	ENST00000376148	NM_005007.3	ENSP00000365318	2
NFKBIL1	upstream gene variant, downstream gene variant	3811	ENST00000376145	NM_001144961.1	ENSP00000365315	2
NFKBIL1	upstream gene variant, downstream gene variant	3068	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	3
NFKBIL1	upstream gene variant	3859	ENST00000473655	?	?	1
NFKBIL1	upstream gene variant	3838	ENST00000496233	?	ENSP00000437148	1
PPIAP9	downstream gene variant, upstream gene variant	3365	ENST00000403866	?	?	4
PRRC2A	downstream gene variant	3549	ENST00000469501	?	?	1
PRRC2A	downstream gene variant	3008	ENST00000487089	?	?	1
PRRC2A	downstream gene variant	1487	ENST00000376007	NM_080686.2	ENSP00000365175	1
PRRC2A	downstream gene variant	1487	ENST00000492691	?	?	1
PRRC2A	downstream gene variant	3942	ENST00000484787	?	?	1
PRRC2A	downstream gene variant	4502	ENST00000460302	?	?	1

PRRC2A	downstream gene variant	2447	ENST00000487839	?	?	1
PRRC2A	upstream gene variant	34	ENST00000464079	?	?	1
PRRC2A	upstream gene variant	886	ENST00000464890	?	?	1
PRRC2A	upstream gene variant	3772	ENST00000483470	?	?	1
PRRC2A	downstream gene variant	1487	ENST00000462617	?	?	1
PRRC2A	downstream gene variant	1487	ENST00000482441	?	?	1
PRRC2A	downstream gene variant	1487	ENST00000376033	NM_004638.3	ENSP00000365201	1
RPL15P4	upstream gene variant	3021	ENST00000416625	?	?	2
SNORA38	downstream gene variant	1658	ENST00000363946	?	?	1
SNORD117	downstream gene variant	725	ENST00000364915	?	?	1
SNORD84	upstream gene variant	1025	ENST00000584275	?	?	2
TNF	downstream gene variant	382	ENST00000449264	NM_000594.3	ENSP00000398698	3
VARS	upstream gene variant	3243	ENST00000463184	?	?	1
VARS	upstream gene variant	705	ENST00000428445	?	ENSP00000415679	1
VARS	upstream gene variant	874	ENST00000461874	?	?	1
VARS	upstream gene variant	55	ENST00000474643	?	?	1
VARS	upstream gene variant	1100	ENST00000482996	?	?	1
VARS	downstream gene variant	1304	ENST00000495010	?	?	1
VARS	upstream gene variant	1544	ENST00000459667	?	?	1
VARS	downstream gene variant	1952	ENST00000489979	?	?	1
VARS	upstream gene variant	865	ENST00000483275	?	?	1
VARS	upstream gene variant	1369	ENST00000479051	?	?	1
XXbac-BPG16N22.5	upstream gene variant	1130	ENST00000538358	?	?	7
XXbac-BPG181B23.6	downstream gene variant, upstream gene variant	445	ENST00000440087	?	?	2
Y_RNA	upstream gene variant	4389	ENST00000364685	?	?	1
Y_RNA	downstream gene variant, upstream gene variant	375	ENST00000383850	?	?	4
Y_RNA	downstream gene variant	2304	ENST00000364337	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V1G2-DDX39B	ENST00000376185	?	ENSP00000365356	3
ATP6V1G2-DDX39B	ENST00000475917	?	?	2
ATP6V1G2-DDX39B	ENST00000480131	?	ENSP00000420191	2
BAG6	ENST00000437771	?	ENSP00000397978	2
BAG6	ENST00000464126	?	?	1
BAG6	ENST00000439687	NM_001199697.1	ENSP00000402856	3
		NM_080703.2, NM_080702.2		3

BAG6	ENST00000211379		ENSP00000211379	
BAG6	ENST00000462682	?	?	1
BAG6	ENST00000375976	NM_001098534.1	ENSP00000365143	3
BAG6	ENST00000464869	?	?	1
BAG6	ENST00000362049	NM_001199698.1	ENSP00000354875	3
BAG6	ENST00000469182	?	?	1
BAG6	ENST00000438149	?	ENSP00000410280	1
BAG6	ENST00000453833	?	ENSP00000406434	1
BAG6	ENST00000441793	?	ENSP00000416633	1
BAG6	ENST00000375964	NM_004639.3	ENSP00000365131	3
BAG6	ENST00000422948	?	ENSP00000413225	1
BAG6	ENST00000435080	?	ENSP00000411083	2
CSNK2B	ENST00000375885	?	ENSP00000365046	1
DDX39B	ENST00000419338	?	ENSP00000410313	1
DDX39B	ENST00000482195	?	?	2
DDX39B	ENST00000428098	?	ENSP00000392672	1
DDX39B	ENST00000396172	NM_004640.6	ENSP00000379475	2
DDX39B	ENST00000462256	?	?	1
DDX39B	ENST00000431908	?	ENSP00000408000	1
DDX39B	ENST00000427214	?	ENSP00000399371	1
DDX39B	ENST00000481456	?	?	1
DDX39B	ENST00000456662	?	ENSP00000416350	1
DDX39B	ENST00000376177	?	ENSP00000365347	1
DDX39B	ENST00000458640	NM_080598.5	ENSP00000416269	2
GPANK1	ENST00000375906	NM_001199237.1	ENSP00000365071	1
HCP5	ENST00000414046	?	?	2
HCP5	ENST00000467369	?	?	1
LST1	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	ENST00000376092	?	ENSP00000365260	1
LST1	ENST00000303757	NM_205840.2	ENSP00000303649	1
LST1	ENST00000211921	?	ENSP00000211921	1
LST1	ENST00000376110	?	ENSP00000365278	1
LST1	ENST00000376093	NM_007161.3	ENSP00000365261	1
LST1	ENST00000376089	?	ENSP00000365257	1
LST1	ENST00000460834	?	?	1
LST1	ENST00000396112	?	ENSP00000379418	1
LST1	ENST00000464044	?	ENSP00000417645	1
LST1	ENST00000376096	?	ENSP00000365264	1
LST1	ENST00000376086	NM_001166538.1	ENSP00000365254	1
LST1	ENST00000418507	NM_205838.2	ENSP00000405900	1
LST1	ENST00000339530	NM_205837.2	ENSP00000339201	1
LST1	ENST00000396101	?	ENSP00000379408	1

LST1	ENST00000376090	?	ENSP00000365258	1
LST1	ENST00000376099	?	ENSP00000365267	1
LST1	ENST00000490742	?	ENSP00000418578	1
MICB	ENST00000538442	NM_001289160.1	ENSP00000442345	4
MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	3
MICB	ENST00000252229	NM_005931.4	ENSP00000252229	3
MSH5	ENST00000463144	?	ENSP00000419648	1
MSH5	ENST00000375755	NM_002441.4	ENSP00000364908	1
MSH5	ENST00000450148	?	ENSP00000394971	1
MSH5	ENST00000375750	NM_172166.3	ENSP00000364903	1
MSH5	ENST00000482280	?	?	1
MSH5	ENST00000375703	NM_172165.3	ENSP00000364855	1
MSH5	ENST00000423982	?	ENSP00000406352	1
MSH5	ENST00000497269	?	ENSP00000419131	1
MSH5	ENST00000375740	NM_025259.5	ENSP00000364892	1
MSH5-SAPCD1	ENST00000493662	?	ENSP00000417871	1
NCR3	ENST00000340027	NM_147130.2	ENSP00000342156	1
NCR3	ENST00000495600	?	?	1
NCR3	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	ENST00000376072	NM_001145467.1	ENSP00000365240	1
NCR3	ENST00000491161	?	?	1
NCR3	ENST00000376071	?	ENSP00000365239	1
NFKBIL1	ENST00000473655	?	?	2
NFKBIL1	ENST00000376148	NM_005007.3	ENSP00000365318	2
NFKBIL1	ENST00000376145	NM_001144961.1	ENSP00000365315	2
NFKBIL1	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	2
NFKBIL1	ENST00000496233	?	ENSP00000437148	2
PRRC2A	ENST00000376033	NM_004638.3	ENSP00000365201	1
PRRC2A	ENST00000376007	NM_080686.2	ENSP00000365175	1
PRRC2A	ENST00000469577	?	?	1
TNF	ENST00000449264	NM_000594.3	ENSP00000398698	1
VARS	ENST00000375663	NM_006295.2	ENSP00000364815	1
VARS	ENST00000461328	?	?	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CLIC1	ENST00000395892	?	ENSP00000379229	1
DDX39B	ENST00000396172	NM_004640.6	ENSP00000379475	1
DDX39B	ENST00000458640	NM_080598.5	ENSP00000416269	1
GPANK1	ENST00000375900	NM_001199238.1, NM_001199239.1	ENSP00000365065	1
GPANK1	ENST00000375895	?	ENSP00000365059	1
MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	1

MICB	ENST00000252229	NM_005931.4	ENSP00000252229	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ATP6V1G2-DDX39B	ENST00000475917	?	1
DDX39B	ENST00000482195	?	1
HCP5	ENST00000414046	?	1
HCP5	ENST00000541196	?	1
LST1	ENST00000464526	?	1
XXbac-BPG16N22.5	ENST00000538358	?	1

Locus 147 (rs4984778)

SNiPAcard

Block annotations

Block info

genomic range	chr16:1,272,472-1,306,040
block size	33,569 bp
variant count	23 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.000 [-3.057 – 0.514]	gene(s) hit or close-by	CACNA1H, PRSS29P, RP11-616M22.12, RP11-616M22.5, TPSAB1, TPSB2, TPSD1, TPSG1
phastCons	μ = 0.004 [0 – 0.019]	eQTL gene(s)	RP11-616M22.3, RP11-616M22.5, TPSAB1, TPSB2, TPSD1, TPSG1
GERP++	μ = -0.650 [-3.81 – 1.1]	potentially regulated gene(s)	CHTF18, TPSD1
CADD score	μ = 2.104 [0.016 – 6.061]	disease gene(s)	CACNA1H

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CACNA1H	EPILEPSY, CHILDHOOD ABSENCE, SUSCEPTIBILITY TO, 6	OMIM	MIM:611942
CACNA1H	Childhood absence epilepsy	OrphaNet	OrphaNet:64280

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TPSD1	?	ENSG00000095917	pancreas	7.27×10-11 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	pancreas	1.36×10-7 (p-value)	GTEx Portal V6	10
TPSD1	?	ENSG00000095917	muscularis mucosae	1.95×10-18 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	muscularis mucosae	2.14×10-19 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	muscularis mucosae	1.74×10-6 (p-value)	GTEx Portal V6	11
TPSD1	?	ENSG00000095917	lung	6.73×10-30 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	lung	1.82×10-26 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	lung	1.88×10-12 (p-value)	GTEx Portal V6	17
TPSAB1	?	ENSG00000172236	blood	2.20×10-7 (p-value)	GTEx Portal V6	10
TPSD1	?	ENSG00000095917	blood	1.98×10-6 (p-value)	GTEx Portal V6	5
TPSD1	?	ENSG00000095917	breast	9.11×10-9 (p-value)	GTEx Portal V6	14
RP11-616M22.5	?	ENSG00000260182	breast	1.46×10-6 (p-value)	GTEx Portal V6	4
TPSB2	?	ENSG00000197253	breast	1.89×10-6 (p-value)	GTEx Portal V6	2
TPSD1	?	ENSG00000095917	tibial artery	1.01×10-17 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	tibial artery	2.69×10-11 (p-value)	GTEx Portal V6	16

TPSD1	?	ENSG00000095917	skeletal muscle	8.05×10-14 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	skeletal muscle	3.31×10-12 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	skeletal muscle	2.00×10-6 (p-value)	GTEx Portal V6	9
TPSD1	?	ENSG00000095917	prostate	1.33×10-12 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	prostate	6.62×10-11 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	putamen	1.27×10-10 (p-value)	GTEx Portal V6	16
TPSD1	?	ENSG00000095917	transverse colon	5.69×10-13 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	transverse colon	5.11×10-10 (p-value)	GTEx Portal V6	15
RP11-616M22.3	?	ENSG00000261294	transverse colon	1.47×10-6 (p-value)	GTEx Portal V6	4
TPSD1	?	ENSG00000095917	sun exposed skin	7.68×10-24 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	sun exposed skin	1.01×10-20 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	sun exposed skin	3.62×10-11 (p-value)	GTEx Portal V6	16
TPSG1	?	ENSG00000116176	sun exposed skin	7.05×10-7 (p-value)	GTEx Portal V6	1
TPSAB1	?	ENSG00000172236	sun exposed skin	5.76×10-6 (p-value)	GTEx Portal V6	1
TPSG1	?	ENSG00000116176	unexposed skin	6.29×10-7 (p-value)	GTEx Portal V6	6
TPSD1	?	ENSG00000095917	unexposed skin	9.92×10-14 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	unexposed skin	7.91×10-13 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	unexposed skin	4.24×10-6 (p-value)	GTEx Portal V6	1
TPSD1	?	ENSG00000095917	aorta	4.69×10-9 (p-value)	GTEx Portal V6	14
TPSB2	?	ENSG00000197253	aorta	4.01×10-7 (p-value)	GTEx Portal V6	7
TPSD1	?	ENSG00000095917	left ventricle	6.33×10-9 (p-value)	GTEx Portal V6	15
TPSD1	?	ENSG00000095917	subcutaneous adipocytes	1.12×10-16 (p-value)	GTEx Portal V6	16
TPSG1	?	ENSG00000116176	subcutaneous adipocytes	7.44×10-10 (p-value)	GTEx Portal V6	17
TPSAB1	?	ENSG00000172236	subcutaneous adipocytes	1.71×10-6 (p-value)	GTEx Portal V6	4
RP11-616M22.5	?	ENSG00000260182	subcutaneous adipocytes	1.07×10-14 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	subcutaneous adipocytes	9.67×10-6 (p-value)	GTEx Portal V6	2
TPSAB1	ENST00000562432	ILMN_1676256	monocyte	2.14×10-21 (p-value)	Zeller et al.	1
TPSAB1	ENST00000561736					
RP11-616M22.5	ENST00000566997					
TPSB2	ENST00000611196					
TPSAB1	ENST00000338844					
TPSB2	ENST00000606293					
TPSAB1	ENST00000461509					
TPSD1	ENST00000211076					
TPSD1	ENST00000397534					
TPSB2	ENST00000612142					
TPSAB1	ENST00000338844	ILMN_2169801	monocyte	1.74×10-19 (p-value)	Zeller et al.	1
TPSAB1	ENST00000461509					
TPSAB1	ENST00000562432					
TPSAB1	ENST00000561736					
TPSB2	?	ENSG00000197253	caudate basal ganglia	1.30×10-15 (p-value)	GTEx Portal V6	17

TPSD1	?	ENSG00000095917	stomach	2.19×10-13 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	stomach	4.06×10-13 (p-value)	GTEx Portal V6	16
TPSD1	?	ENSG00000095917	visceral adipocytes	5.93×10-9 (p-value)	GTEx Portal V6	13
RP11-616M22.5	?	ENSG00000260182	visceral adipocytes	1.96×10-7 (p-value)	GTEx Portal V6	8
TPSB2	?	ENSG00000197253	visceral adipocytes	4.40×10-6 (p-value)	GTEx Portal V6	1
TPSB2	?	ENSG00000197253	nucleus accumbens	4.27×10-12 (p-value)	GTEx Portal V6	15
TPSB2	?	ENSG00000197253	cortex	1.70×10-12 (p-value)	GTEx Portal V6	16
TPSD1	?	ENSG00000095917	tibial nerve	9.35×10-15 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	tibial nerve	7.05×10-11 (p-value)	GTEx Portal V6	16
TPSG1	?	ENSG00000116176	tibial nerve	4.64×10-6 (p-value)	GTEx Portal V6	1
TPSD1	?	ENSG00000095917	esophagus mucosa	1.30×10-22 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	esophagus mucosa	9.90×10-22 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	esophagus mucosa	1.86×10-11 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	anterior cingulate cortex	3.86×10-12 (p-value)	GTEx Portal V6	15
TPSD1	?	ENSG00000095917	gastroesophageal junction	6.16×10-11 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	gastroesophageal junction	1.33×10-9 (p-value)	GTEx Portal V6	13
TPSD1	?	ENSG00000095917	testis	6.86×10-9 (p-value)	GTEx Portal V6	14
RP11-616M22.5	?	ENSG00000260182	testis	2.96×10-10 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	frontal cortex	1.13×10-12 (p-value)	GTEx Portal V6	15
TPSD1	?	ENSG00000095917	atrial appendage	2.56×10-7 (p-value)	GTEx Portal V6	10
RP11-616M22.5	?	ENSG00000260182	atrial appendage	1.76×10-7 (p-value)	GTEx Portal V6	11
TPSB2	?	ENSG00000197253	hypothalamus	4.64×10-10 (p-value)	GTEx Portal V6	14
TPSB2	?	ENSG00000197253	hippocampus	2.27×10-9 (p-value)	GTEx Portal V6	9
TPSD1	?	ENSG00000095917	terminal ileum	1.33×10-6 (p-value)	GTEx Portal V6	1
RP11-616M22.5	?	ENSG00000260182	terminal ileum	3.45×10-7 (p-value)	GTEx Portal V6	1
RP11-616M22.3	?	ENSG00000261294	terminal ileum	2.33×10-7 (p-value)	GTEx Portal V6	2
TPSD1	?	ENSG00000095917	thyroid	9.03×10-7 (p-value)	GTEx Portal V6	8
RP11-616M22.5	?	ENSG00000260182	cerebellum	2.30×10-6 (p-value)	GTEx Portal V6	3
TPSG1	ENST00000564684	ILMN_1769219	skin	4.76×10-8 (p-value)	MuTHER consortium	1
TPSG1	ENST00000234798		adipocyte	2.12×10-5 (p-value)	MuTHER consortium	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000020937		

ENCODE promoter-associated distal DHS (Enhancer)

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000184452	1	ENCP00000020927	CHTF18

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000502225 (open chromatin region)	4	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	DNase1, H3K27me3
		A549	H3K27me3, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CACNA1H	downstream gene variant	704	ENST00000348261	NM_021098.2	ENSP00000334198	4
CACNA1H	downstream gene variant	1388	ENST00000562079	?	ENSP00000454581	1
CACNA1H	downstream gene variant	1388	ENST00000569107	?	ENSP00000454990	1
CACNA1H	downstream gene variant	1388	ENST00000565831	?	ENSP00000455840	1
CACNA1H	downstream gene variant	707	ENST00000564927	?	ENSP00000455512	4
CACNA1H	downstream gene variant	1388	ENST00000564231	?	ENSP00000457555	1
CACNA1H	downstream gene variant	701	ENST00000358590	NM_001005407.1	ENSP00000351401	4
PRSS29P	downstream gene variant	4913	ENST00000568091	?	?	1
RP11-616M22.12	upstream gene variant, downstream gene variant	1167	ENST00000621827	?	?	6
RP11-616M22.5	upstream gene variant	1300	ENST00000566997	?	?	4
TPSAB1	upstream gene variant, downstream gene variant	1027	ENST00000561736	?	ENSP00000456821	9
TPSAB1	upstream gene variant, downstream gene variant	500	ENST00000461509	?	ENSP00000418247	10
TPSAB1	upstream gene variant, downstream gene variant	383	ENST00000562432	?	?	9
TPSAB1	upstream gene variant, downstream gene variant	472	ENST00000338844	NM_003294.3	ENSP00000343577	10
TPSB2	downstream gene variant, upstream gene variant	274	ENST00000612142	?	ENSP00000478695	6
TPSB2	downstream gene variant, upstream gene variant	542	ENST00000611196	?	ENSP00000484461	5
TPSB2	downstream gene variant, upstream gene variant	273	ENST00000606293	NM_024164.5	ENSP00000482743	6
TPSD1	upstream gene variant	20	ENST00000397534	?	ENSP00000380668	4
TPSD1	upstream gene variant	94	ENST00000211076	NM_012217.2	ENSP00000211076	4
TPSG1	downstream gene variant, upstream gene variant	187	ENST00000564684	?	?	6
TPSG1	upstream gene variant	1151	ENST00000234798	NM_012467.3	ENSP00000234798	5

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
TPSB2	ENST00000611196	?	ENSP00000484461	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TPSB2	ENST00000611196	?	ENSP00000484461	1

TPSG1	ENST00000234798	NM_012467.3	ENSP00000234798	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TPSAB1	ENST00000338844	NM_003294.3	ENSP00000343577	1
TPSAB1	ENST00000461509	?	ENSP00000418247	1
TPSAB1	ENST00000561736	?	ENSP00000456821	1

Locus 148 (rs2361355)

SNiPAcard

Block annotations

Block info

genomic range	chr12:94,621,186-94,630,029
block size	8,844 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.564 [-1.928 – 1.142]	**gene(s) hit or close-by**	PLXNC1
phastCons	μ = 0.166 [0 – 0.99]	**eQTL gene(s)**	–
GERP++	μ = -0.797 [-3.36 – 3.49]	**potentially regulated gene(s)**	–
CADD score	μ = 4.859 [0.817 – 16.82]	**disease gene(s)**	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PLXNC1	downstream gene variant	386	ENST00000546659	?	?	7

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLXNC1	ENST00000258526	NM_005761.2	ENSP00000258526	11

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PLXNC1	ENST00000546659	?	1

Locus 149 (rs17206736)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,377,642-31,391,257
block size	13,616 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.284 [-1.521 – 0.426]	gene(s) hit or close-by	HCP5, MICA
phastCons	μ = 0.057 [0 – 0.409]	eQTL gene(s)	CCHCR1, HLA-B, MICA, PSORS1C1, PSORS1C2, Y_RNA
GERP++	μ = -0.195 [-1.75 – 0.536]	potentially regulated gene(s)	MICA, MICA, MICA, MICA, MICA, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2
CADD score	μ = 2.861 [0.826 – 9.209]	disease gene(s)	, , , VARS2, HLA-B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICA	?	ENSG00000204520	muscularis mucosae	6.76×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	lung	6.56×10^{-12} (p-value)	GTEx Portal V6	12
PSORS1C2	?	ENSG00000204538	lung	8.23×10^{-7} (p-value)	GTEx Portal V6	1
PSORS1C1	?	ENSG00000204540	lung	7.32×10^{-7} (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	lung	2.71×10^{-8} (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	atrial appendage	1.02×10^{-8} (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	transformed fibroblasts	1.36×10^{-12} (p-value)	GTEx Portal V6	11
HLA-B	?	ENSG00000234745	transformed fibroblasts	5.82×10^{-6} (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	transformed fibroblasts	1.00×10^{-9} (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	tibial artery	7.30×10^{-11} (p-value)	GTEx Portal V6	11
Y_RNA	?	ENSG00000199332	tibial artery	2.46×10^{-12} (p-value)	GTEx Portal V6	12

MICA	?	ENSG00000204520	breast	2.71×10-10 (p-value)	GTEx Portal V6	10
MICA	?	ENSG00000204520	blood	4.42×10-11 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	thyroid	5.40×10-12 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	skeletal muscle	1.18×10-12 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	EBV lymphocytes	2.60×10-8 (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	EBV lymphocytes	3.18×10-7 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	sun exposed skin	3.28×10-11 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	aorta	1.80×10-8 (p-value)	GTEx Portal V6	11
Y_RNA	?	ENSG00000199332	aorta	1.92×10-10 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	left ventricle	5.23×10-9 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	subcutaneous adipocytes	3.15×10-9 (p-value)	GTEx Portal V6	10
Y_RNA	?	ENSG00000199332	subcutaneous adipocytes	3.69×10-8 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	caudate basal ganglia	5.43×10-8 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	visceral adipocytes	2.59×10-8 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial nerve	9.72×10-14 (p-value)	GTEx Portal V6	11
CCHCR1	?	ENSG00000204536	tibial nerve	2.40×10-6 (p-value)	GTEx Portal V6	1
PSORS1C1	?	ENSG00000204540	tibial nerve	9.33×10-6 (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	tibial nerve	1.35×10-8 (p-value)	GTEx Portal V6	4
Y_RNA	?	ENSG00000199332	sigmoid colon	1.96×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	esophagus mucosa	1.52×10-10 (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	esophagus mucosa	3.49×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	testis	2.92×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444713	1	ENCP00000048015	MICA MICA MICA MICA MICA
		ENCP00000047968	VARS2 VARS2 VARS2 VARS2 VARS2 VARS2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MICA	downstream gene variant	39	ENST00000449934	NM_001177519.2	ENSP00000413079	4

MICA	downstream gene variant	41	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	4
MICA	downstream gene variant	39	ENST00000421350	?	ENSP00000402410	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	13
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	ENST00000421350	?	ENSP00000402410	1

Locus 150 (rs12099358)

SNiPAcard

Block annotations

Block info

genomic range	chr11:116,718,415-116,972,935
block size	254,521 bp
variant count	77 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.215 [-3.588 – 2.455]	**gene(s) hit or close-by**	AP000936.1, AP000936.2, AP000936.4, APOA1-AS, RNY4P6, SIK3, SIK3-IT1
phastCons	μ = 0.049 [0 – 0.909]	**eQTL gene(s)**	APOA1, APOA1-AS, SIDT2
GERP++	μ = -0.360 [-8.15 – 3.25]	**potentially regulated gene(s)**	AP000936.4, BUD13, CEP164, PCSK7, RNF214, SIDT2, SIK3
CADD score	μ = 4.261 [0.03 – 18.86]	**disease gene(s)**	APOA1, CEP164

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
2-palmitoylglycerophosphocholine*	2.41×10-5	Metabolomics GWAS Server	24816252	5
1-oleoylglycerol (1-monoolein)	5.16×10-5	Metabolomics GWAS Server	24816252	4

Disease gene annotation

gene	trait	source DB	source entry/link
APOA1	AMYLOIDOSIS, FAMILIAL VISCERAL	OMIM	MIM:105200
APOA1	HYPOALPHALIPOPROTEINEMIA, PRIMARY	OMIM	MIM:604091
APOA1	TANGIER DISEASE	OMIM	MIM:205400
APOA1	APOLIPOPROTEIN A-I	OMIM	MIM:107680
CEP164	NEPHRONOPHTHISIS 15	OMIM	MIM:614845
APOA1	Primary systemic amyloidosis	OrphaNet	OrphaNet:314701
APOA1	Familial renal amyloidosis due to Apolipoprotein AI variant	OrphaNet	OrphaNet:93560
APOA1	APOLIPOPROTEIN A-I DEFICIENCY	OrphaNet	OrphaNet:425
CEP164	Senior-Loken syndrome	OrphaNet	OrphaNet:3156

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
APOA1	?	ENSG00000118137	left ventricle	6.90×10-11 (p-value)	GTEx Portal V6	76
APOA1-AS	?	ENSG00000235910	left ventricle	1.34×10-9 (p-value)	GTEx Portal V6	74
SIDT2	ENST00000278951	ILMN_1791912	blood	8.62×10-5 (p-value)	Westra et al.	9
SIDT2	ENST00000532062					
SIDT2	ENST00000324225					
SIDT2	ENST00000431081					
SIDT2	ENST00000620360					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000123650	1	ENCP00000014795	PCSK7
ENCE00000123336	1	ENCP00000014749	BUD13
		ENCP00000014780	SIDT2
ENCE00000123493	1	ENCP00000014770	SIK3
ENCE00000123628	1	ENCP00000014793	PCSK7
ENCE00000123403	1	ENCP00000014798	PCSK7 RNF214
		ENCP00000014772	SIK3 AP000936.4
		ENCP00000014804	CEP164

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000569505 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
		liver (HepG2)	H3K36me3
		blood (GM12878)	PU1, H3K4me3, EBF1, DNase1, Yy1, H3K4me1, H3K27ac, H3K36me3, H3K4me2
		lung (IMR90)	H3K36me3, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1, H3K36me3
		NHLF	DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3
		breast (HMEC)	H3K4me1, H3K27ac
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K36me3
		endothelium (HUVEC)	H3K4me1, DNase1
		A549	H3K36me3
ENSR00001573904 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K9ac, H3K27ac, H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1, H3K4me1
		muscle (HSMM)	H3K4me1, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K4me2, H3K36me3
		NHLF	H3K36me3, DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, H3K36me3, H3K4me1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		A549	H3K36me3
ENSR00000190303 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K4me2
		blood (DND-41)	H3K4me1, H3K36me3
		skin (NHDF-AD)	H3K4me3
		breast (HMEC)	H3K4me2
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K36me3

		liver (HepG2)	H3K4me1, H3K4me2, HNF4A
		blood (GM12878)	H3K4me1
		lung (IMR90)	H3K4me2
		skin (NHEK)	H3K4me2
ENSR00000569523 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K4me3, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00001573919 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1
		nervous (NH-A)	DNase1
ENSR00000321726 (enhancer)	1	blood (K562)	DNase1
ENSR00000569533 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, USF1
		HSMMtube	DNase1, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		blood (GM12878)	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1
		NHLF	DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		breast (HMEC)	H3K4me1, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Jund, Cjun
		monocytes (Monocytes-CD14+)	DNase1, H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	DNase1
ENSR00001607329 (enhancer)	1	liver (HepG2)	H3K4me1
ENSR00001573924 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	Jund
		monocytes (Monocytes-CD14+)	H4K20me1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me2, H3K4me3
		skin (NHEK)	DNase1
ENSR00000569539 (enhancer)	2	monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		embryonic stem cell (H1ESC)	DNase1
		blood (K562)	DNase1
		blood (DND-41)	H3K4me1, H3K27ac
ENSR00000569540 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H4K20me1, H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		skin (NHEK)	H3K4me1, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	DNase1
ENSR00000569542 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1

		monocytes (Monocytes-CD14+)	H4K20me1
		endothelium (HUVEC)	Cjun, DNase1
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00000569548 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, CTCF
		HSMMtube	DNase1
		blood (K562)	PU1, Cjun, FOSL1, DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		liver (HepG2)	FOSL2, Jund, DNase1, H3K4me1, FOXA1
		lung (IMR90)	DNase1
		blood (GM12878)	H3K79me2, BATF
		nervous (NH-A)	DNase1, H3K27me3
		skin (NHEK)	H3K27ac, H3K36me3, H3K4me1, DNase1
		NHLF	DNase1
		Osteobl	H3K27ac, H4K20me1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1, Cfos, Jund, Cjun
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H4K20me1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	DNase1, H3K4me2, H3K4me3
ENSR00001573930 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H4K20me1
		embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H4K20me1, FOXA2, HDAC2, DNase1, H3K4me1, FOXA1
		blood (GM12878)	H3K4me2, H3K79me2, H3K4me1
		skin (NHEK)	DNase1
		muscle (HSMM)	H3K36me3
ENSR00000569552 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H4K20me1, H3K79me2
		blood (GM12878)	H3K79me2
		skin (NHEK)	DNase1, H3K4me1, H3K4me2
		breast (HMEC)	H3K4me2, H3K9ac, H3K27ac, DNase1
ENSR00001573933 (enhancer)	2	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H3K79me2, H3K4me1, FOXA1
		blood (GM12878)	H3K36me3, H3K4me2, H3K4me3, H3K79me2
		A549	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2, H3K36me3
ENSR00000569556 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, CTCF, PolII, Rad21, TAF7, Sin3Ak20, H3K27ac, ATF3, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3
		HSMMtube	DNase1, H3K9ac, H3K4me2, H3K27ac, H2AZ
		blood (K562)	USF1, H3K79me2, H2AZ, TAF1, Max, PolII, H3K4me2, CTCF, DNase1, H3K4me3, H3K9ac, Rad21, H3K27me3, Yy1, H3K27ac
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K9ac, DNase1
		liver (HepG2)	Yy1, H3K27me3, H3K4me3, CTCF, H3K79me2, Rad21, TAF1, USF1, H3K4me1, H3K4me2, HNF4A, FOXA1, H3K9ac, H3K27ac, DNase1
		lung (IMR90)	CTCF, H3K9ac, H3K4me3, H3K4me2, H3K27ac, DNase1
		blood (GM12878)	Yy1, DNase1, H3K4me1, H2AZ, PolII, PU1, ATF3, Rad21, H3K79me2, H3K9ac, H3K4me2, H3K27ac, USF1, H3K4me3
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K4me2, H3K4me3, DNase1, H3K9ac, H3K27ac, CTCF
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	CTCF, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me3, H3K27ac, DNase1, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K4me3, PolII, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, DNase1
		A549	H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP000936.1	downstream gene variant, upstream gene variant	2526	ENST00000438127	?	?	3

AP000936.2	downstream gene variant, upstream gene variant	2369	ENST00000584092	?	?	4
AP000936.4	upstream gene variant	905	ENST00000442124	?	?	2
APOA1-AS	downstream gene variant	1020	ENST00000444200	?	?	1
RNY4P6	upstream gene variant, downstream gene variant	2836	ENST00000363667	?	?	4
SIK3	upstream gene variant	4152	ENST00000413553	?	ENSP00000414347	1
SIK3	downstream gene variant, upstream gene variant	738	ENST00000472648	?	?	3
SIK3	downstream gene variant, upstream gene variant	4016	ENST00000497049	?	?	2
SIK3	downstream gene variant	165	ENST00000480468	?	?	3
SIK3	upstream gene variant	3883	ENST00000454905	?	ENSP00000392849	1
SIK3	upstream gene variant	3824	ENST00000445177	?	ENSP00000391295	1
SIK3	upstream gene variant	586	ENST00000488337	?	?	2
SIK3	upstream gene variant	3914	ENST00000485363	?	?	1
SIK3	upstream gene variant	3798	ENST00000375300	NM_001281748.1, NM_025164.4	ENSP00000364449	1
SIK3	upstream gene variant	3782	ENST00000446921	NM_001281749.1	ENSP00000390442	1
SIK3	upstream gene variant	3948	ENST00000415541	?	ENSP00000392761	1
SIK3-IT1	downstream gene variant, upstream gene variant	1178	ENST00000454832	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AP000936.4	ENST00000442124	?	?	1
APOA1-AS	ENST00000444200	?	?	4
SIK3	ENST00000497049	?	?	18
SIK3	ENST00000465421	?	?	5
SIK3	ENST00000446921	NM_001281749.1	ENSP00000390442	76
SIK3	ENST00000480222	?	?	2
SIK3	ENST00000445177	?	ENSP00000391295	76
SIK3	ENST00000454905	?	ENSP00000392849	2
SIK3	ENST00000415541	?	ENSP00000392761	76
SIK3	ENST00000413553	?	ENSP00000414347	23
SIK3	ENST00000488337	?	?	5
SIK3	ENST00000375300	NM_001281748.1, NM_025164.4	ENSP00000364449	76
SIK3	ENST00000485363	?	?	63
SIK3-IT1	ENST00000454832	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
APOA1-AS	ENST00000444200	?	1

Locus 151 (rs7657257)

SNiPAcard

Block annotations

Block info

genomic range	chr4:15,551,298-15,719,439
block size	168,142 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.028 [-3.15 – 3.645]	gene(s) hit or close-by	BST1, CC2D2A, FAM200B, FBXL5
phastCons	μ = 0.184 [0 – 1]	eQTL gene(s)	BST1
GERP++	μ = 0.117 [-2.61 – 3.84]	potentially regulated gene(s)	BST1, FBXL5
CADD score	μ = 5.313 [0.018 – 18.87]	disease gene(s)	CC2D2A

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CC2D2A	MECKEL SYNDROME, TYPE 6	OMIM	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	OMIM	MIM:612285
CC2D2A	COACH SYNDROME	OMIM	MIM:216360
CC2D2A	COACH syndrome	DECIPHER	MIM:216360
CC2D2A	MECKEL SYNDROME, TYPE 6	DECIPHER	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	DECIPHER	MIM:612285
CC2D2A	Joubert syndrome with oculorenal defect	OrphaNet	OrphaNet:2318
CC2D2A	Meckel syndrome	OrphaNet	OrphaNet:564
CC2D2A	Joubert syndrome with hepatic defect	OrphaNet	OrphaNet:1454

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
BST1	missense variant	ENST00000382346	?	ENSP00000371783	Q/R	cAa/cGa	?	?	1
BST1	missense variant	ENST00000265016	NM_004334.2	ENSP00000265016	Q/R	cAa/cGa	?	?	1
BST1	missense variant	ENST00000505785	?	ENSP00000423357	Q/R	cAa/cGa	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
BST1	ENST00000382346	ILMN_1770161	monocyte	1.56×10-4 (p-value)	Fairfax et al.	2
BST1	ENST00000265016					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000634801	1	ENST00000510802, ENST00000515679, ENST00000503196, ENST00000509314	FBXL5

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000040477		

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001237239 (enhancer)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00001237240 (promoter flanking region)	2	NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		Osteobl	H3K27ac
		nervous (NH-A)	DNase1
ENSR00001429723 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac, H3K36me3
		HSMMtube	DNase1
		nervous (NH-A)	H3K36me3, DNase1
		blood (K562)	H3K27me3
		A549	H3K36me3
		muscle (HSMM)	DNase1, H3K4me2, H3K27ac, H2AZ

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BST1	upstream gene variant	4309	ENST00000265016	NM_004334.2	ENSP00000265016	1
BST1	upstream gene variant	846	ENST00000514989	?	ENSP00000424761	3
BST1	upstream gene variant	4177	ENST00000514445	?	ENSP00000420925	1
BST1	upstream gene variant	4455	ENST00000382346	?	ENSP00000371783	1
CC2D2A	upstream gene variant	1063	ENST00000512202	?	?	1
FAM200B	upstream gene variant	954	ENST00000506610	?	?	1
FAM200B	downstream gene variant	2064	ENST00000504598	?	?	1
FBXL5	upstream gene variant	1517	ENST00000507700	?	?	4
FBXL5	upstream gene variant	2174	ENST00000507899	?	ENSP00000422049	1
FBXL5	upstream gene variant	640	ENST00000512066	?	ENSP00000426993	1
FBXL5	downstream gene variant, upstream gene variant	526	ENST00000514541	?	?	5
FBXL5	upstream gene variant	1476	ENST00000412094	NM_001193535.1	ENSP00000408679	4
FBXL5	upstream gene variant	2161	ENST00000510802	?	ENSP00000423961	1
FBXL5	upstream gene variant	1517	ENST00000511441	?	ENSP00000425027	4
FBXL5	upstream gene variant	1563	ENST00000504837	?	?	4
FBXL5	upstream gene variant	669	ENST00000513163	?	ENSP00000425472	1
FBXL5	upstream gene variant	2109	ENST00000515679	?	ENSP00000424423	1
FBXL5	upstream gene variant	1446	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
BST1	ENST00000382346	?	ENSP00000371783	S	tcT/tcC	1
BST1	ENST00000265016	NM_004334.2	ENSP00000265016	S	tcT/tcC	1
BST1	ENST00000514445	?	ENSP00000420925	S	tcT/tcC	1
BST1	ENST00000505785	?	ENSP00000423357	S	tcT/tcC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BST1	ENST00000382346	?	ENSP00000371783	3
BST1	ENST00000265016	NM_004334.2	ENSP00000265016	3
BST1	ENST00000514445	?	ENSP00000420925	3
BST1	ENST00000514989	?	ENSP00000424761	1
BST1	ENST00000505785	?	ENSP00000423357	3
CC2D2A	ENST00000503292	NM_001080522.2	ENSP00000421809	2
CC2D2A	ENST00000389652	?	ENSP00000374303	2
CC2D2A	ENST00000424120	?	ENSP00000403465	2
CC2D2A	ENST00000506643	?	ENSP00000422931	2
CC2D2A	ENST00000512202	?	?	1
FAM200B	ENST00000622362	NM_001145191.1	ENSP00000483930	1
FAM200B	ENST00000422728	?	ENSP00000393017	1
FAM200B	ENST00000503617	?	ENSP00000422751	1
FAM200B	ENST00000505260	?	?	1
FAM200B	ENST00000503600	?	?	1
FAM200B	ENST00000510920	?	?	1
FAM200B	ENST00000509022	?	?	1
FAM200B	ENST00000514803	?	?	1
FAM200B	ENST00000515697	?	?	1
FAM200B	ENST00000502502	?	?	1
FAM200B	ENST00000512855	?	?	1
FAM200B	ENST00000504598	?	?	2
FAM200B	ENST00000504823	?	?	1
FAM200B	ENST00000515430	?	?	1
FAM200B	ENST00000502856	?	?	1
FAM200B	ENST00000510186	?	?	1
FAM200B	ENST00000510032	?	?	1
FAM200B	ENST00000508567	?	?	1
FAM200B	ENST00000507305	?	?	1
FAM200B	ENST00000513053	?	?	1
FAM200B	ENST00000507992	?	?	1
FAM200B	ENST00000504137	?	?	1

FBXL5	ENST00000515679	?	ENSP00000424423	7
FBXL5	ENST00000509314	?	ENSP00000421070	5
FBXL5	ENST00000507899	?	ENSP00000422049	7
FBXL5	ENST00000514541	?	?	2
FBXL5	ENST00000510802	?	ENSP00000423961	7
FBXL5	ENST00000507700	?	?	3
FBXL5	ENST00000412094	NM_001193535.1	ENSP00000408679	3
FBXL5	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	3
FBXL5	ENST00000504837	?	?	1
FBXL5	ENST00000513163	?	ENSP00000425472	2
FBXL5	ENST00000503196	?	ENSP00000425541	5
FBXL5	ENST00000511441	?	ENSP00000425027	3

Locus 152 (rs16923189)

SNiPAcard

Block annotations

Block info

genomic range	chr9:5,505,822-5,516,067
block size	10,246 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.079 [-1.593 – 0.836]	gene(s) hit or close-by	PDCD1LG2
phastCons	μ = 0.008 [0 – 0.023]	eQTL gene(s)	PDCD1LG2
GERP++	μ = 0.014 [-5.63 – 2.98]	potentially regulated gene(s)	KIAA2026, RP11-574F11.3
CADD score	μ = 3.835 [0.078 – 7.74]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PDCD1LG2	?	ENSG00000197646	muscularis mucosae	3.64×10^{-16} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	visceral adipocytes	3.12×10^{-15} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	lung	1.69×10^{-14} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	tibial nerve	4.38×10^{-11} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	transformed fibroblasts	1.40×10^{-9} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	thyroid	1.98×10^{-16} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	skeletal muscle	3.56×10^{-6} (p-value)	GTEx Portal V6	2
PDCD1LG2	?	ENSG00000197646	left ventricle	1.06×10^{-8} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	aorta	5.40×10^{-9} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	subcutaneous adipocytes	2.82×10^{-11} (p-value)	GTEx Portal V6	7
PDCD1LG2	?	ENSG00000197646	atrial appendage	3.21×10^{-6} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	sigmoid colon	2.43×10^{-6} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	breast	1.59×10^{-6} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	gastroesophageal junction	9.59×10^{-8} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	adrenal gland	3.52×10^{-6} (p-value)	GTEx Portal V6	1
PDCD1LG2	?	ENSG00000197646	tibial artery	3.29×10^{-6} (p-value)	GTEx Portal V6	5
PDCD1LG2	?	ENSG00000197646	coronary artery	4.73×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000536431	1	ENCP00000058187	RP11-574F11.3
ENCE00000536432	1	ENCP00000058198	KIAA2026
		ENCP00000058187	RP11-574F11.3

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001299543 (promoter)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K4me2, DNase1
		skin (NHDF-AD)	H3K9ac, DNase1, H3K4me3, H3K4me1, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac
		liver (HepG2)	DNase1, H3K27me3
		lung (IMR90)	H3K4me2, H3K56ac, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H2BK120ac, H2BK20ac, H2AK5ac, H3K9ac, H4K91ac, DNase1, H4K8ac, H3K23ac, H3K14ac, H2BK12ac, H3K27ac, H3K18ac
		blood (GM12878)	BCL11A, H3K27ac, H3K4me2, H3K9ac, H3K4me3, H3K79me2, IRF4, Cjun, PU1, BATF, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K27ac
		skin (NHEK)	DNase1, H3K4me1, H3K4me2, H3K4me3, H3K36me3
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Jund, Nrsf, H3K27ac, Cjun
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	Cjun, Max, DNase1, H3K4me2, H3K9ac, H3K4me1, H3K27ac, PolII
		A549	DNase1, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDCD1LG2	upstream gene variant	4748	ENST00000397747	NM_025239.3	ENSP00000380855	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDCD1LG2	ENST00000397747	NM_025239.3	ENSP00000380855	5

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDCD1LG2	ENST00000397747	NM_025239.3	ENSP00000380855	1

Locus 153 (rs536299)

SNiPAcard

Block annotations

Block info

genomic range	chr7:83,224,626-83,287,607
block size	62,982 bp
variant count	24 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.337 [-2.51 – 0.531]	gene(s) hit or close-by	SEMA3E
phastCons	μ = 0.018 [0 – 0.12]	eQTL gene(s)	SEMA3E
GERP++	μ = -0.310 [-3.15 – 2.13]	potentially regulated gene(s)	–
CADD score	μ = 2.926 [0.263 – 7.555]	disease gene(s)	SEMA3E

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1-eicosadienoylglycerophosphocholine*	9.31×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link	
SEMA3E	CHARGE SYNDROME	OrphaNet	OrphaNet:138	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SEMA3E	?	ENSG00000170381	gastroesophageal junction	2.51×10^{-7} (p-value)	GTEx Portal V6	20
SEMA3E	?	ENSG00000170381	muscularis mucosae	8.25×10^{-10} (p-value)	GTEx Portal V6	23
SEMA3E	?	ENSG00000170381	breast	2.96×10^{-8} (p-value)	GTEx Portal V6	22
SEMA3E	?	ENSG00000170381	subcutaneous adipocytes	2.84×10^{-9} (p-value)	GTEx Portal V6	23
SEMA3E	?	ENSG00000170381	thyroid	1.32×10^{-12} (p-value)	GTEx Portal V6	24
SEMA3E	?	ENSG00000170381	tibial nerve	8.70×10^{-6} (p-value)	GTEx Portal V6	4
SEMA3E	?	ENSG00000170381	transformed fibroblasts	5.95×10^{-7} (p-value)	GTEx Portal V6	9
SEMA3E	?	ENSG00000170381	esophagus mucosa	1.91×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001561427 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K27me3
		Osteobl	H3K27me3, H3K4me2

blood (DND-41)	H3K27me3
skin (NHDF-AD)	H3K4me3, H3K4me2
breast (HMEC)	H3K4me2, H3K4me3
cervix (HeLa-S3)	H3K27me3
monocytes (Monocytes-CD14+)	H3K27me3
endothelium (HUVEC)	H3K36me3, H3K27me3
liver (HepG2)	H3K27me3
nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
A549	H3K4me3, H3K4me2, H3K9ac, H3K27me3
skin (NHEK)	H3K4me2, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SEMA3E	upstream gene variant	101	ENST00000453333	?	ENSP00000415184	3
SEMA3E	upstream gene variant	2987	ENST00000442159	?	ENSP00000412867	2
SEMA3E	upstream gene variant	4879	ENST00000427262	NM_001178129.1	ENSP00000405052	1
SEMA3E	upstream gene variant	567	ENST00000307792	NM_012431.2	ENSP00000303212	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SEMA3E	ENST00000307792	NM_012431.2	ENSP00000303212	19
SEMA3E	ENST00000427262	NM_001178129.1	ENSP00000405052	18
SEMA3E	ENST00000453333	?	ENSP00000415184	19
SEMA3E	ENST00000442159	?	ENSP00000412867	7

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SEMA3E	ENST00000307792	NM_012431.2	ENSP00000303212	1

Locus 154 (rs3756309)

SNiPAcard

Block annotations

Block info

genomic range	chr5:149,504,115-149,505,624
block size	1,510 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.645 [-2.666 – -0.656]	gene(s) hit or close-by	PDGFRB
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	CSF1R, PDGFRB
GERP++	μ = -2.577 [-3.26 – -1.88]	potentially regulated gene(s)	–
CADD score	μ = 4.338 [2.553 – 6.987]	disease gene(s)	PDGFRB, CSF1R

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PDGFRB	JUVENILE MYELOMONOCYTIC LEUKEMIA	OMIM	MIM:607785
PDGFRB	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 4	OMIM	MIM:615007
PDGFRB	LEUKEMIA, ACUTE MYELOID	OMIM	MIM:601626
PDGFRB	MYOFIBROMATOSIS, INFANTILE, 1	OMIM	MIM:228550
PDGFRB	MYELOPROLIFERATIVE DISORDER, CHRONIC, WITH EOSINOPHILIA	OMIM	MIM:131440
CSF1R	LEUKOENCEPHALOPATHY, DIFFUSE HEREDITARY, WITH SPHEROIDS	OMIM	MIM:221820
PDGFRB	Familial Infantile Myofibromatosis	DECIPHER	MIM:228550
PDGFRB	Bilateral striopallidodentate calcinosis	OrphaNet	OrphaNet:1980
PDGFRB	Unclassified chronic myeloproliferative disease	OrphaNet	OrphaNet:86830
PDGFRB	Chronic myelomonocytic leukemia	OrphaNet	OrphaNet:98823
PDGFRB	Idiopathic hypereosinophilic syndrome	OrphaNet	OrphaNet:3260
PDGFRB	Infantile myofibromatosis 1	OrphaNet	OrphaNet:2591
PDGFRB	Myeloid neoplasm associated with PDGFRB rearrangement	OrphaNet	OrphaNet:168950
CSF1R	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia	OrphaNet	OrphaNet:313808

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PDGFRB	ENST00000520579	ILMN_1815057	blood	2.97×10^{-7} (p-value)	MuTHER consortium	2
PDGFRB	ENST00000261799					
CSF1R	ENST00000509861	ILMN_1686623	blood	3.96×10^{-5} (p-value)	Westra et al.	1
CSF1R	ENST00000504875					
CSF1R	ENST00000286301					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDGFRB	upstream gene variant	2315	ENST00000521723	?	?	3

PDGFRB	upstream gene variant	2350	ENST00000519575	?	?	3
PDGFRB	upstream gene variant	223	ENST00000520229	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDGFRB	ENST00000520579	?	ENSP00000430026	3
PDGFRB	ENST00000261799	NM_002609.3	ENSP00000261799	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PDGFRB	ENST00000520229	?	1

Locus 155 (rs515863)

SNiPAcard

Block annotations

Block info

genomic range	chr13:113,830,329-113,911,651
block size	81,323 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.113 [-0.295 – 0.741]	gene(s) hit or close-by	CUL4A, PCID2, PROZ
phastCons	μ = 0.005 [0 – 0.022]	eQTL gene(s)	PROZ, RP11-120K24.4
GERP++	μ = -1.060 [-3.57 – 1.1]	potentially regulated gene(s)	–
CADD score	μ = 2.255 [1.398 – 4.143]	disease gene(s)	PROZ

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PROZ	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-120K24.4	?	ENSG00000269356	sigmoid colon	3.87×10-7 (p-value)	GTEx Portal V6	3
PROZ	?	ENSG00000126231	liver	1.11×10-7 (p-value)	GTEx Portal V6	6
PROZ	ENST00000375547	ILMN_1664565	liver	1.02×10-14 (p-value)	Innocenti et al.	1
PROZ	ENST00000342783					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CUL4A	upstream gene variant	2206	ENST00000375441	NM_001278513.1	ENSP00000364590	1
CUL4A	downstream gene variant	2562	ENST00000470067	?	?	1
CUL4A	upstream gene variant, downstream gene variant	3613	ENST00000488558	?	ENSP00000480367	2
CUL4A	upstream gene variant	2707	ENST00000326335	NM_001278514.1	ENSP00000322132	1
CUL4A	upstream gene variant	3896	ENST00000498562	?	?	1
CUL4A	upstream gene variant	3557	ENST00000375440	NM_001008895.2	ENSP00000364589	1
CUL4A	upstream gene variant	2785	ENST00000451881	NM_003589.2	ENSP00000389118	1
CUL4A	upstream gene variant, downstream gene variant	3846	ENST00000463426	?	?	2
CUL4A	upstream gene variant	2849	ENST00000617546	?	ENSP00000481782	1
CUL4A	upstream gene variant	3629	ENST00000474116	?	?	1

CUL4A	upstream gene variant	3643	ENST00000494985	?	?	1
PCID2	downstream gene variant	1597	ENST00000246505	?	ENSP00000246505	1
PCID2	downstream gene variant	1596	ENST00000375477	?	ENSP00000364626	1
PCID2	downstream gene variant	1596	ENST00000375479	NM_018386.3, NM_001127203.2	ENSP00000364628	1
PCID2	downstream gene variant	1602	ENST00000375457	NM_001258213.1	ENSP00000364606	1
PCID2	downstream gene variant	4146	ENST00000475433	?	?	1
PCID2	downstream gene variant	2257	ENST00000473462	?	?	1
PCID2	downstream gene variant	1597	ENST00000463102	?	?	1
PCID2	downstream gene variant	1598	ENST00000462653	?	?	1
PCID2	downstream gene variant	1597	ENST00000375459	?	ENSP00000364608	1
PCID2	downstream gene variant	4205	ENST00000480971	?	?	1
PCID2	downstream gene variant	1524	ENST00000622406	NM_001258212.1	ENSP00000479494	1
PCID2	downstream gene variant	1597	ENST00000493650	?	?	1
PCID2	downstream gene variant	1562	ENST00000337344	NM_001127202.2	ENSP00000337405	1
PROZ	downstream gene variant	3632	ENST00000375547	NM_003891.2	ENSP00000364697	1
PROZ	downstream gene variant	3635	ENST00000342783	NM_001256134.1	ENSP00000344458	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CUL4A	ENST00000472083	?	?	1
CUL4A	ENST00000451881	NM_003589.2	ENSP00000389118	3
CUL4A	ENST00000326335	NM_001278514.1	ENSP00000322132	3
CUL4A	ENST00000375441	NM_001278513.1	ENSP00000364590	3
CUL4A	ENST00000488558	?	ENSP00000480367	1
CUL4A	ENST00000498562	?	?	2
CUL4A	ENST00000375440	NM_001008895.2	ENSP00000364589	3
CUL4A	ENST00000463426	?	?	1
CUL4A	ENST00000617546	?	ENSP00000481782	3
CUL4A	ENST00000494985	?	?	1
PCID2	ENST00000337344	NM_001127202.2	ENSP00000337405	2
PCID2	ENST00000493650	?	?	1
PCID2	ENST00000491548	?	?	1
PCID2	ENST00000375479	NM_018386.3, NM_001127203.2	ENSP00000364628	2
PCID2	ENST00000622406	NM_001258212.1	ENSP00000479494	2
PCID2	ENST00000475433	?	?	2
PCID2	ENST00000473462	?	?	1
PCID2	ENST00000375459	?	ENSP00000364608	2

PCID2	ENST00000375477	?	ENSP00000364626	2
PCID2	ENST00000484641	?	?	2
PCID2	ENST00000480971	?	?	2
PCID2	ENST00000246505	?	ENSP00000246505	2
PCID2	ENST00000375457	NM_001258213.1	ENSP00000364606	2

Locus 156 (rs8396)

SNiPAcard

Block annotations

Block info

genomic range	chr4:159,621,250-159,648,866
block size	27,617 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.438 [-2.93 – 2.416]	**gene(s) hit or close-by**	ETFDH, PPID, U3
phastCons	μ = 0.111 [0 – 1]	**eQTL gene(s)**	ETFDH, PPID, U3
GERP++	μ = -1.497 [-7.63 – 2.64]	**potentially regulated gene(s)**	–
CADD score	μ = 4.455 [0.066 – 18.16]	**disease gene(s)**	ETFDH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Blood metabolite ratios	$<2.00\times10^{-57}$	GWAS Catalog	24816252	1
Blood metabolite levels	$<1.00\times10^{-31}$	GWAS Catalog	24816252	1
Metabolic traits	$<6.00\times10^{-15}$	GWAS Catalog	21886157	1
Metabolite levels	$<4.00\times10^{-24}$	GWAS Catalog	20037589	1
decanoylcarnitine	8.69×10^{-38}	Metabolomics GWAS Server	24816252	5

Disease gene annotation

gene	trait	source DB	source entry/link
ETFDH	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	OMIM	MIM:231680
ETFDH	glutaric aciduria type 2C	DECIPHER	MIM:231680
ETFDH	Multiple acyl-CoA dehydrogenation deficiency, mild type	OrphaNet	OrphaNet:394532
ETFDH	Multiple acyl-CoA dehydrogenation deficiency, severe neonatal type	OrphaNet	OrphaNet:394529

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PPID	missense variant	ENST00000307720	NM_005038.2	ENSP00000303754	I/L	Ata/Cta	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ETFDH	ENST00000307738	ILMN_1758034	skin	4.25×10^{-10} (p-value)	MuTHER consortium	6
ETFDH	ENST00000511912		adipocyte	3.72×10^{-20} (p-value)	MuTHER consortium	6
ETFDH	ENST00000506422					
PPID	?	ENSG00000171497	pancreas	7.06×10^{-7} (p-value)	GTEx Portal V6	12

U3	?	ENSG00000271817	pancreas	4.72×10-6 (p-value)	GTEx Portal V6	1
PPID	?	ENSG00000171497	muscularis mucosae	1.79×10-8 (p-value)	GTEx Portal V6	15
U3	?	ENSG00000271817	muscularis mucosae	8.28×10-7 (p-value)	GTEx Portal V6	12
ETFDH	?	ENSG00000171503	muscularis mucosae	2.95×10-6 (p-value)	GTEx Portal V6	2
U3	?	ENSG00000271817	lung	7.56×10-9 (p-value)	GTEx Portal V6	16
ETFDH	?	ENSG00000171503	atrial appendage	1.58×10-8 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	transformed fibroblasts	1.07×10-10 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	transformed fibroblasts	1.14×10-14 (p-value)	GTEx Portal V6	16
ETFDH	?	ENSG00000171503	tibial artery	1.13×10-9 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	tibial artery	2.61×10-12 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	tibial artery	2.37×10-6 (p-value)	GTEx Portal V6	6
U3	?	ENSG00000271817	breast	1.62×10-11 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	thyroid	9.17×10-7 (p-value)	GTEx Portal V6	10
PPID	?	ENSG00000171497	thyroid	2.76×10-6 (p-value)	GTEx Portal V6	13
ETFDH	?	ENSG00000171503	skeletal muscle	7.75×10-40 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	transverse colon	1.32×10-8 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	sun exposed skin	1.47×10-11 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	sun exposed skin	1.51×10-10 (p-value)	GTEx Portal V6	16
ETFDH	?	ENSG00000171503	left ventricle	4.25×10-13 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	left ventricle	1.75×10-10 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	subcutaneous adipocytes	1.07×10-19 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	subcutaneous adipocytes	6.66×10-10 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	visceral adipocytes	4.40×10-8 (p-value)	GTEx Portal V6	13
PPID	?	ENSG00000171497	visceral adipocytes	3.13×10-7 (p-value)	GTEx Portal V6	14
PPID	?	ENSG00000171497	stomach	7.84×10-8 (p-value)	GTEx Portal V6	14
ETFDH	?	ENSG00000171503	stomach	2.05×10-7 (p-value)	GTEx Portal V6	1
U3	?	ENSG00000271817	tibial nerve	2.56×10-17 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	tibial nerve	2.96×10-19 (p-value)	GTEx Portal V6	16
U3	?	ENSG00000271817	esophagus mucosa	2.03×10-7 (p-value)	GTEx Portal V6	16
PPID	?	ENSG00000171497	esophagus mucosa	5.64×10-11 (p-value)	GTEx Portal V6	16
ETFDH	?	ENSG00000171503	adrenal gland	5.58×10-8 (p-value)	GTEx Portal V6	14
U3	?	ENSG00000271817	unexposed skin	6.54×10-6 (p-value)	GTEx Portal V6	2
U3	?	ENSG00000271817	aorta	3.30×10-6 (p-value)	GTEx Portal V6	6
U3	?	ENSG00000271817	testis	9.31×10-9 (p-value)	GTEx Portal V6	12
U3	?	ENSG00000271817	gastroesophageal junction	5.99×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001253032 (CTCF binding site)	2	embryonic stem cell (H1ESC)	H3K36me3, CTCF, Rad21
		HSMMtube	CTCF
		skin (NHDF-AD)	H3K36me3, CTCF
		muscle (HSMM)	CTCF, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, CTCF
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		Osteobl	CTCF, H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K36me3
ENSR00001436804 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, H3K4me2, H3K9ac, H3K4me3, TAF1, Yy1, H3K36me3, H3K27me3, CTCF, PolII, Rad21, Egr1, Gabp, TAF7, H3K27ac
		HSMMtube	H3K4me2, H3K27ac, H3K4me3, H2AZ, DNase1, H3K9ac, H3K79me2
		blood (K562)	H3K27ac, Max, E2F6, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Cmyc, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, DNase1
		muscle (HSMM)	H3K9ac, H3K36me3, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K79me2
		liver (HepG2)	ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, TAF1, H3K79me2, Gabp, Sin3Ak20, PolII, DNase1
		lung (IMR90)	H4K8ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	DNase1, PolII, H2AZ, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2
		breast (HMEC)	H3K4me2, H3K27ac, H3K4me3, H3K9ac
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, H3K4me3, Gabp
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, PolII, H3K4me3, H3K4me2, Max
		A549	H3K9ac, DNase1, H3K27ac, H3K36me3, H3K4me2, H3K4me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ETFDH	1	ENST00000511912	hsa-miR-144-3p hsa-miR-340-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ETFDH	downstream gene variant	42	ENST00000511912	NM_004453.3	ENSP00000426638	5
ETFDH	downstream gene variant	1096	ENST00000507475	?	ENSP00000422735	2
ETFDH	downstream gene variant	63	ENST00000307738	NM_001281738.1, NM_001281737.1	ENSP00000303552	4
ETFDH	downstream gene variant	94	ENST00000506422	?	?	4
PPID	downstream gene variant, upstream gene variant	18	ENST00000507213	?	ENSP00000421221	7
PPID	downstream gene variant, upstream gene variant	472	ENST00000307720	NM_005038.2	ENSP00000303754	4
PPID	downstream gene variant, upstream gene	96	ENST00000512699	?	ENSP00000423207	5

	variant					
U3	upstream gene variant	593	ENST00000607547	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ETFDH	ENST00000307738	NM_001281738.1, NM_001281737.1	ENSP00000303552	3
ETFDH	ENST00000511912	NM_004453.3	ENSP00000426638	3
ETFDH	ENST00000506422	?	?	3
PPID	ENST00000307720	NM_005038.2	ENSP00000303754	6
PPID	ENST00000512699	?	ENSP00000423207	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ETFDH	ENST00000511912	NM_004453.3	ENSP00000426638	1
PPID	ENST00000307720	NM_005038.2	ENSP00000303754	1
PPID	ENST00000512699	?	ENSP00000423207	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PPID	ENST00000307720	NM_005038.2	ENSP00000303754	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
U3	ENST00000607547	?	1

Locus 157 (rs1126478)

SNiPAcard

Block annotations

Block info

genomic range	chr3:46,484,261-46,504,316
block size	20,056 bp
variant count	24 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.587 [-4.779 – 1.614]	gene(s) hit or close-by	LTF
phastCons	μ = 0.035 [0 – 0.761]	eQTL gene(s)	CCR3
GERP++	μ = -1.107 [-10.6 – 3.49]	potentially regulated gene(s)	CCR5, LTF
CADD score	μ = 3.650 [0.001 – 10.49]	disease gene(s)	LTF, CCR5

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM096382	1

Disease gene annotation

gene	trait	source DB	source entry/link
LTF	SPECIFIC GRANULE DEFICIENCY	OMIM	MIM:245480
CCR5	WEST NILE VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610379
CCR5	DIABETES MELLITUS, INSULIN-DEPENDENT, 22	OMIM	MIM:612522
CCR5	HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, SUSCEPTIBILITY TO	OMIM	MIM:609423

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
LTF	missense variant	ENST00000431944	?	ENSP00000395234	K/R	aAa/aGa	tolerated	benign	1
LTF	missense variant	ENST00000417439	?	ENSP00000405546	K/R	aAa/aGa	tolerated	benign	1
LTF	missense variant	ENST00000443496	?	ENSP00000397427	K/R	aAa/aGa	tolerated	benign	1
LTF	missense variant	ENST00000426532	NM_001199149.1	ENSP00000405719	K/R	aAa/aGa	tolerated	benign	1
LTF	missense variant	ENST00000231751	NM_002343.4	ENSP00000231751	K/R	aAa/aGa	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CCR3	ENST00000545097	ILMN_1763322	blood	5.43×10^{-7} (p-value)	Westra et al.	2
CCR3	ENST00000452454					
CCR3	ENST00000395942					
CCR3	ENST00000357422					

CCR3	ENST00000395940
CCR3	ENST00000475150

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000607402	1	ENST00000443496, ENST00000417439, ENST00000426532, ENST00000231751	LTF

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000036255		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000337735	1	ENCP00000036250	CCR5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001362955 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K4me1, CTCF, H3K27ac
		skin (NHDF-AD)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	H3K27me3
		A549	CTCF
		skin (NHEK)	CTCF
ENSR00001362956 (CTCF binding site)	1	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	CTCF
ENSR00001362957 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, CTCF, DNase1
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		lung (IMR90)	H3K27me3, CTCF
		blood (GM12878)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF

	A549		CTCF, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LTF	downstream gene variant	579	ENST00000431944	?	ENSP00000395234	2
LTF	upstream gene variant	865	ENST00000493056	?	?	12
LTF	downstream gene variant, upstream gene variant	1552	ENST00000462667	?	?	6
LTF	downstream gene variant	2282	ENST00000498301	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
LTF	ENST00000417439	?	ENSP00000405546	N	aaT/aaC	1
LTF	ENST00000443496	?	ENSP00000397427	N	aaT/aaC	1
LTF	ENST00000426532	NM_001199149.1	ENSP00000405719	N	aaT/aaC	1
LTF	ENST00000231751	NM_002343.4	ENSP00000231751	N	aaT/aaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LTF	ENST00000431944	?	ENSP00000395234	6
LTF	ENST00000478874	?	?	24
LTF	ENST00000417439	?	ENSP00000405546	23
LTF	ENST00000443496	?	ENSP00000397427	23
LTF	ENST00000426532	NM_001199149.1	ENSP00000405719	23
LTF	ENST00000462667	?	?	1
LTF	ENST00000231751	NM_002343.4	ENSP00000231751	23
LTF	ENST00000498301	?	?	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
LTF	ENST00000498301	?	1

Locus 158 (rs6136)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,527,101-169,563,951
block size	36,851 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.550 [-0.777 – 1.877]	gene(s) hit or close-by	F5, SELP
phastCons	μ = 0.019 [0.004 – 0.033]	eQTL gene(s)	F5
GERP++	μ = -0.890 [-3.92 – 2.14]	potentially regulated gene(s)	–
CADD score	μ = 4.322 [3.287 – 5.356]	disease gene(s)	F5

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Soluble levels of adhesion molecules	$<4.00\times10^{-61}$	GWAS Catalog	20167578	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Selectin p polymorphism	benign	ClinVar	RCV000014482.1	1
?	HGMD curated	HGMD	CM981792	1
SELECTIN P POLYMORPHISM	OMIM curated	OMIM	MIM:173610	1

Disease gene annotation

gene	trait	source DB	source entry/link
F5	STROKE, ISCHEMIC	OMIM	MIM:601367
F5	THROMBOPHILIA DUE TO ACTIVATED PROTEIN C RESISTANCE	OMIM	MIM:188055
F5	PREGNANCY LOSS, RECURRENT, SUSCEPTIBILITY TO, 1	OMIM	MIM:614389
F5	FACTOR V DEFICIENCY	OMIM	MIM:227400
F5	BUDD-CHIARI SYNDROME	OMIM	MIM:600880
F5	Congenital factor V deficiency	OrphaNet	OrphaNet:326
F5	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217
F5	BUDD-CHIARI SYNDROME	OrphaNet	OrphaNet:131
F5	East Texas bleeding disorder	OrphaNet	OrphaNet:391320

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SELP	missense variant	ENST00000367788	?	ENSP00000356762	P/T	Cct/Act	?	?	1
SELP	missense variant	ENST00000367786	?	ENSP00000356760	P/T	Cct/Act	?	?	1
SELP	missense variant	ENST00000426706	?	ENSP00000391694	P/T	Cct/Act	?	?	1
SELP	missense variant	ENST00000263686	NM_003005.3	ENSP00000263686	P/T	Cct/Act	?	?	1
SELP	missense	ENST00000458599	?	ENSP00000399368	P/T	Cct/Act	?	?	1

variant			

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
F5	?	ENSG00000198734	esophagus mucosa	4.00×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000547341 (TF binding site)	1	blood (GM12878)	BATF, DNase1, BCL11A
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SELP	upstream gene variant	3143	ENST00000466167	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F5	ENST00000367796	?	ENSP00000356770	1
F5	ENST00000367797	NM_000130.4	ENSP00000356771	1

Locus 160 (rs2555515)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,365,143-186,396,645
block size	31,503 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.461 [-0.432 – 2.533]	gene(s) hit or close-by	FETUB, HRG, RP11-573D15.8
phastCons	μ = 0.011 [0 – 0.027]	eQTL gene(s)	–
GERP++	μ = -1.116 [-5.26 – 2.97]	potentially regulated gene(s)	–
CADD score	μ = 3.494 [1.577 – 6.299]	disease gene(s)	HRG

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HRG	THROMBOPHILIA DUE TO HISTIDINE-RICH GLYCOPROTEIN DEFICIENCY	OMIM	MIM:613116
HRG	Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency	OrphaNet	OrphaNet:217467

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001381859 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1, Jund, H3K4me1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHEK)	H3K4me1, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FETUB	downstream gene variant	2442	ENST00000488561	?	?	1
HRG	downstream gene variant	4058	ENST00000468154	?	?	1
HRG	downstream gene variant	616	ENST00000232003	NM_000412.3	ENSP00000232003	1
HRG	downstream gene variant	625	ENST00000495413	?	?	1
RP11-573D15.8	downstream gene variant	708	ENST00000629451	?	?	2
RP11-573D15.8	downstream gene variant	1266	ENST00000630726	?	?	2
RP11-573D15.8	upstream gene variant	4536	ENST00000455926	?	?	1
RP11-573D15.8	downstream gene variant	4412	ENST00000625839	?	?	1
RP11-573D15.8	downstream gene variant, upstream gene variant	765	ENST00000625826	?	?	3
RP11-573D15.8	downstream gene variant	4485	ENST00000628728	?	?	1
RP11-573D15.8	downstream gene variant	647	ENST00000627469	?	?	2
RP11-573D15.8	upstream gene variant	4338	ENST00000626006	?	?	1
RP11-573D15.8	upstream gene variant	4532	ENST00000428501	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FETUB	ENST00000265029	NM_014375.2	ENSP00000265029	2
FETUB	ENST00000382134	?	ENSP00000371569	2
FETUB	ENST00000435961	?	ENSP00000393851	2
FETUB	ENST00000382136	?	ENSP00000371571	2
FETUB	ENST00000450521	?	ENSP00000404288	2
FETUB	ENST00000420570	?	ENSP00000405438	2
FETUB	ENST00000431018	?	ENSP00000396581	2
HRG	ENST00000232003	NM_000412.3	ENSP00000232003	1
HRG	ENST00000495413	?	?	1
RP11-573D15.8	ENST00000627469	?	?	2
RP11-573D15.8	ENST00000628190	?	?	4
RP11-573D15.8	ENST00000626649	?	?	4
RP11-573D15.8	ENST00000428501	?	?	2
RP11-573D15.8	ENST00000625877	?	?	2
RP11-573D15.8	ENST00000626633	?	?	4
RP11-573D15.8	ENST00000625839	?	?	1
RP11-573D15.8	ENST00000630331	?	?	4
RP11-573D15.8	ENST00000625386	?	?	4
RP11-573D15.8	ENST00000625303	?	?	4
RP11-573D15.8	ENST00000628253	?	?	4
RP11-573D15.8	ENST00000626845	?	?	4
RP11-573D15.8	ENST00000630315	?	?	4
RP11-573D15.8	ENST00000455926	?	?	2
RP11-573D15.8	ENST00000630178	?	?	4
RP11-573D15.8	ENST00000626006	?	?	1
RP11-573D15.8	ENST00000629106	?	?	4
RP11-573D15.8	ENST00000627830	?	?	2
RP11-573D15.8	ENST00000630864	?	?	4
RP11-573D15.8	ENST00000630726	?	?	2
RP11-573D15.8	ENST00000628505	?	?	4
RP11-573D15.8	ENST00000629451	?	?	2
RP11-573D15.8	ENST00000625741	?	?	4
RP11-573D15.8	ENST00000628728	?	?	1

Locus 161 (rs2284367)

SNiPAcard

Block annotations

Block info

genomic range	chr11:34,468,936-34,516,856
block size	47,921 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.298 [-3.369 – 2.261]	**gene(s) hit or close-by**	AL137224.1, CAT, ELF5
phastCons	μ = 0.007 [0 – 0.123]	**eQTL gene(s)**	ABTB2, CAT
GERP++	μ = -0.916 [-8.18 – 3]	**potentially regulated gene(s)**	CAPRIN1, NAT10
CADD score	μ = 4.821 [0.258 – 15.84]	**disease gene(s)**	CAT

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM025323	1

Disease gene annotation

gene	trait	source DB	source entry/link
CAT	ACATALASEMIA	OMIM	MIM:614097
CAT	ACATALASEMIA	OrphaNet	OrphaNet:926

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CAT	?	ENSG00000121691	pancreas	2.40×10^{-10} (p-value)	GTEx Portal V6	20
CAT	ENST00000241052	ILMN_1651705	skin	2.67×10^{-5} (p-value)	MuTHER consortium	10
CAT	ENST00000534710		blood	2.44×10^{-8} (p-value)	MuTHER consortium	10
CAT	?	ENSG00000121691	muscularis mucosae	4.69×10^{-9} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	lung	4.41×10^{-10} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	atrial appendage	3.33×10^{-13} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	transformed fibroblasts	8.63×10^{-26} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	blood	7.56×10^{-13} (p-value)	GTEx Portal V6	20
ABTB2	?	ENSG00000166016	blood	3.68×10^{-7} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	breast	3.41×10^{-12} (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	tibial artery	1.02×10^{-12} (p-value)	GTEx Portal V6	20
CAT	ENST00000241052	ILMN_1651705	monocyte	2.30×10^{-14} (p-value)	Fairfax et al.	10
CAT	ENST00000534710		b-cell	1.88×10^{-12} (p-value)	Fairfax et al.	10
CAT	?	ENSG00000121691	thyroid	2.57×10^{-17} (p-value)	GTEx Portal V6	20

CAT	?	ENSG00000121691	skeletal muscle	1.76×10-12 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	EBV lymphocytes	7.71×10-8 (p-value)	GTEx Portal V6	16
CAT	?	ENSG00000121691	transverse colon	7.97×10-9 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	blood	4.72×10-5 (q-value)	SeeQTL DB (HapMap)	7
CAT	?	ENSG00000121691	ovary	4.17×10-7 (p-value)	GTEx Portal V6	17
CAT	?	ENSG00000121691	sun exposed skin	6.01×10-16 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	unexposed skin	4.61×10-7 (p-value)	GTEx Portal V6	18
CAT	?	ENSG00000121691	aorta	3.71×10-8 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	left ventricle	9.50×10-13 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	subcutaneous adipocytes	3.93×10-12 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	stomach	2.66×10-6 (p-value)	GTEx Portal V6	5
CAT	?	ENSG00000121691	visceral adipocytes	1.66×10-7 (p-value)	GTEx Portal V6	16
CAT	?	ENSG00000121691	tibial nerve	1.18×10-12 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	pituitary	2.88×10-8 (p-value)	GTEx Portal V6	20
CAT	?	ENSG00000121691	esophagus mucosa	4.10×10-12 (p-value)	GTEx Portal V6	20
CAT	ENST00000241052	201432_at	blood	3.80×10-13 (p-value)	Dixon et al.	5
CAT	ENST00000534710					
CAT	ENST00000241052	211922_s_at	blood	1.20×10-9 (p-value)	Dixon et al.	5
CAT	?	ENSG00000121691	testis	1.09×10-9 (p-value)	GTEx Portal V6	20
CAT	ENST00000241052	ILMN_1651705	monocyte	1.61×10-26 (p-value)	Zeller et al.	2
CAT	ENST00000534710					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000102675	1	ENCP00000011718	NAT10
		ENCP00000011715	CAPRIN1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001603033 (enhancer)	1	cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me1
		liver (HepG2)	H3K36me3
		Osteobl	H3K36me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL137224.1	downstream gene variant, upstream gene variant	1272	ENST00000579311	?	?	3
CAT	upstream gene variant, downstream gene variant	1947	ENST00000528104	?	?	4

CAT	upstream gene variant, downstream gene variant	459	ENST00000525707	?	?	2
CAT	upstream gene variant	1724	ENST00000534710	?	?	3
CAT	upstream gene variant, downstream gene variant	1185	ENST00000530343	?	?	5
ELF5	downstream gene variant	2463	ENST00000532417	?	ENSP00000436386	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CAT	ENST00000241052	NM_001752.3	ENSP00000241052	D	gaT/gaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CAT	ENST00000525707	?	?	8
CAT	ENST00000530343	?	?	5
CAT	ENST00000241052	NM_001752.3	ENSP00000241052	14
ELF5	ENST00000528709	?	?	5
ELF5	ENST00000429939	NM_001243080.1	ENSP00000407589	5
ELF5	ENST00000620316	NM_001243081.1	ENSP00000484521	5
ELF5	ENST00000312319	NM_198381.1	ENSP00000311010	5
ELF5	ENST00000257832	NM_001422.3	ENSP00000257832	5
ELF5	ENST00000532417	?	ENSP00000436386	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CAT	ENST00000525707	?	1
CAT	ENST00000530343	?	1

Locus 162 (rs3740909)

SNiPAcard

Block annotations

Block info

genomic range	chr11:125,882,173-125,901,248
block size	19,076 bp
variant count	43 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.312 [-4.048 – 2.849]	gene(s) hit or close-by	CDON
phastCons	μ = 0.064 [0 – 1]	eQTL gene(s)	–
GERP++	μ = -0.344 [-8.68 – 4.03]	potentially regulated gene(s)	–
CADD score	μ = 4.002 [0.107 – 22.5]	disease gene(s)	CDON

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CDON	HOLOPROSENCEPHALY 11	OMIM	MIM:614226
CDON	Holoprosencephaly 11	DECIPHER	MIM:614226
CDON	Semilobar holoprosencephaly	OrphaNet	OrphaNet:220386
CDON	Alobar holoprosencephaly	OrphaNet	OrphaNet:93925
CDON	Microform holoprosencephaly	OrphaNet	OrphaNet:280200
CDON	Midline interhemispheric variant of holoprosencephaly	OrphaNet	OrphaNet:93926
CDON	Septopreoptic holoprosencephaly	OrphaNet	OrphaNet:280195
CDON	Lobar holoprosencephaly	OrphaNet	OrphaNet:93924

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CDON	missense variant	ENST00000392693	NM_001243597.1	ENSP00000376458	K/E	Aaa/Gaa	?	?	1
CDON	missense variant	ENST00000534661	?	ENSP00000436755	K/E	Aaa/Gaa	?	?	1
CDON	missense variant	ENST00000263577	NM_016952.4	ENSP00000263577	K/E	Aaa/Gaa	?	?	1
CDON	missense variant	ENST00000531586	?	ENSP00000434212	K/E	Aaa/Gaa	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001608006 (enhancer)	3	liver (HepG2)	H3K4me1
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CDON	downstream gene variant, upstream gene variant	339	ENST00000525625	?	?	18

CDON	upstream gene variant	244	ENST00000531830	?	ENSP00000432571	11
CDON	downstream gene variant	56	ENST00000527967	?	ENSP00000436940	17
CDON	downstream gene variant	1168	ENST00000534818	?	ENSP00000437176	16
CDON	downstream gene variant	45	ENST00000531586	?	ENSP00000434212	16
CDON	upstream gene variant	289	ENST00000534661	?	ENSP00000436755	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CDON	ENST00000531586	?	ENSP00000434212	16
CDON	ENST00000525625	?	?	22
CDON	ENST00000531830	?	ENSP00000432571	22
CDON	ENST00000534661	?	ENSP00000436755	31
CDON	ENST00000263577	NM_016952.4	ENSP00000263577	42
CDON	ENST00000527967	?	ENSP00000436940	16
CDON	ENST00000392693	NM_001243597.1	ENSP00000376458	42
CDON	ENST00000534818	?	ENSP00000437176	11

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CDON	ENST00000525625	?	4

Locus 163 (rs12938)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,653,795-169,666,871
block size	13,077 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.008 [-2.659 – 0.256]	**gene(s) hit or close-by**	C1orf112, SELL
phastCons	μ = 0.040 [0 – 0.199]	**eQTL gene(s)**	C1orf112, RN7SL333P, SELL
GERP++	μ = -1.263 [-6.06 – 1.21]	**potentially regulated gene(s)**	–
CADD score	μ = 3.621 [0.664 – 7.649]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Hemoglobin a, glycosylated	6.33×10^{-5}	dbGaP	pha002881	dbGaP	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C1orf112	?	ENSG00000000460	cerebellar hemisphere	6.03×10^{-6} (p-value)	GTEx Portal V6	5
RN7SL333P	?	ENSG00000239494	tibial nerve	4.85×10^{-6} (p-value)	GTEx Portal V6	1
C1orf112	?	ENSG00000000460	tibial nerve	4.94×10^{-6} (p-value)	GTEx Portal V6	4
C1orf112	?	ENSG00000000460	atrial appendage	3.68×10^{-8} (p-value)	GTEx Portal V6	5
RN7SL333P	?	ENSG00000239494	tibial artery	3.30×10^{-6} (p-value)	GTEx Portal V6	5
C1orf112	?	ENSG00000000460	thyroid	7.29×10^{-7} (p-value)	GTEx Portal V6	5
SELL	ENST00000236147	ILMN_1724422	blood	1.85×10^{-13} (p-value)	MuTHER consortium	3
SELL	ENST00000497295					
SELL	?	ENSG00000188404	blood	5.74×10^{-7} (p-value)	GTEx Portal V6	4
SELL	ENST00000497295	ILMN_1724422	monocyte	2.11×10^{-15} (p-value)	Fairfax et al.	1
SELL	ENST00000236147		b-cell	1.57×10^{-8} (p-value)	Fairfax et al.	1
SELL	?	ENSG00000188404	blood	5.79×10^{-5} (q-value)	SeeQTL DB (HapMap)	2

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
SELL	ENST00000236147	ILMN_1724422	chr1	monocyte	3.58×10^{-29} (p-value)	Zeller et al.	1
SELL	ENST00000497295						

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000547356 (promoter flanking region)	3	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3, H3K36me3
		embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (GM12878)	H3K27ac, H3K4me1
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SELL	downstream gene variant	41	ENST00000497295	?	?	1
SELL	downstream gene variant	3765	ENST00000463108	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1orf112	ENST00000498289	?	?	5
SELL	ENST00000236147	NM_000655.4	ENSP00000236147	4
SELL	ENST00000497295	?	?	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SELL	ENST00000236147	NM_000655.4	ENSP00000236147	1

Locus 164 (rs2289252)

SNiPAcard

Block annotations

Block info

genomic range	chr4:187,199,005-187,213,883
block size	14,879 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.748 [-2.809 – 0.512]	**gene(s) hit or close-by**	F11, F11-AS1
phastCons	μ = 0.001 [0 – 0.004]	**eQTL gene(s)**	IRF2
GERP++	μ = -1.683 [-6.41 – 0.594]	**potentially regulated gene(s)**	–
CADD score	μ = 1.126 [0.034 – 2.484]	**disease gene(s)**	F11

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Activated partial thromboplastin time	$<6.00\times10^{-43}$	GWAS Catalog	22703881	1
bradykinin, des-arg(9)	9.31×10^{-5}	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link
F11	FACTOR XI DEFICIENCY	OMIM	MIM:612416
F11	Congenital factor XI deficiency	OrphaNet	OrphaNet:329

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IRF2	ENST00000502750	ILMN_2090607	monocyte	2.92×10^{-4} (p-value)	Fairfax et al.	1
IRF2	ENST00000393593					
IRF2	ENST00000505067					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
F11	downstream gene variant	2954	ENST00000492972	?	ENSP00000424479	2
F11	downstream gene variant	3026	ENST00000452239	?	ENSP00000397401	2
F11	downstream gene variant	3242	ENST00000514715	?	?	2
F11	upstream gene variant, downstream gene variant	1362	ENST00000503841	?	?	3
F11	upstream gene variant, downstream gene variant	543	ENST00000264691	?	ENSP00000264691	2
F11	downstream gene variant	3949	ENST00000264692	?	ENSP00000264692	1
F11	downstream gene variant	3949	ENST00000403665	NM_000128.3	ENSP00000384957	1
F11-AS1	downstream gene variant	999	ENST00000505103	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F11	ENST00000452239	?	ENSP00000397401	2
F11	ENST00000264692	?	ENSP00000264692	4
F11	ENST00000403665	NM_000128.3	ENSP00000384957	4
F11	ENST00000264691	?	ENSP00000264691	1
F11-AS1	ENST00000505103	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
F11-AS1	ENST00000505103	?	1

Locus 165 (rs6033)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,513,583-169,521,853
block size	8,271 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.657 [-0.932 – 2.883]	gene(s) hit or close-by	F5
phastCons	μ = 0.363 [0 – 1]	eQTL gene(s)	–
GERP++	μ = -0.075 [-10.6 – 3.18]	potentially regulated gene(s)	–
CADD score	μ = 8.529 [0.002 – 16.68]	disease gene(s)	F5

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM045144	1

Disease gene annotation

gene	trait	source DB	source entry/link
F5	STROKE, ISCHEMIC	OMIM	MIM:601367
F5	THROMBOPHILIA DUE TO ACTIVATED PROTEIN C RESISTANCE	OMIM	MIM:188055
F5	PREGNANCY LOSS, RECURRENT, SUSCEPTIBILITY TO, 1	OMIM	MIM:614389
F5	FACTOR V DEFICIENCY	OMIM	MIM:227400
F5	BUDD-CHIARI SYNDROME	OMIM	MIM:600880
F5	Congenital factor V deficiency	OrphaNet	OrphaNet:326
F5	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217
F5	BUDD-CHIARI SYNDROME	OrphaNet	OrphaNet:131
F5	East Texas bleeding disorder	OrphaNet	OrphaNet:391320

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
F5	missense variant	ENST00000367796	?	ENSP00000356770	T/M	aCg/aTg	?	?	1
F5	missense variant	ENST00000367797	NM_000130.4	ENSP00000356771	T/M	aCg/aTg	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000671446 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF, DNase1, Rad21
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	DNase1, CTCF

		liver (HepG2)	H3K36me3, Rad21, FOXA1, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	DNase1, CTCF
ENSR00001525847 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF, H3K27me3, DNase1
		blood (K562)	CTCF
		blood (DND-41)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	CTCF
		liver (HepG2)	Rad21, CTCF, H3K36me3
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		A549	CTCF
		nervous (NH-A)	CTCF, H3K27me3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
F5	ENST00000367796	?	ENSP00000356770	3	3	3
F5	ENST00000367797	NM_000130.4	ENSP00000356771	3	3	3

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F5	ENST00000367796	?	ENSP00000356770	5
F5	ENST00000367797	NM_000130.4	ENSP00000356771	5

Locus 166 (rs3109167)

SNiPAcard

Block annotations

Block info

genomic range	chr7:82,976,912-82,978,643
block size	1,732 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.437 [0.109 – 0.961]	**gene(s) hit or close-by**	–
phastCons	μ = 0.009 [0.001 – 0.023]	**eQTL gene(s)**	–
GERP++	μ = 0.500 [-0.0267 – 1.41]	**potentially regulated gene(s)**	–
CADD score	μ = 2.549 [0.653 – 4.433]	**disease gene(s)**	–

Locus 167 (rs9469012)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,360,702-31,426,888
block size	66,187 bp
variant count	22 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.290 [-1.89 – 0.318]	**gene(s) hit or close-by**	HCP5, LINC01149, MICA, XXbac-BPG181B23.6, XXbac-BPG181B23.7, Y_RNA
phastCons	μ = 0.118 [0 – 0.47]	**eQTL gene(s)**	HCG22, HLA-B, LY6G5C, MICA, XXbac-BPG181B23.7
GERP++	μ = 0.053 [-0.718 – 0.641]	**potentially regulated gene(s)**	ABHD16A, ABHD16A, ABHD16A, ABHD16A, ABHD16A, ABHD16A, ABHD16A, AIF1, AIF1, AIF1, AIF1, AIF1, AIF1, BAG6, BAG6, BAG6, BAG6, BAG6, BAG6, BAG6, C2, C2, C2, C2, C2, C2, C2, C2, C2, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCP5, HCP5, HCP5, HCP5, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LTB, LTB, LTB, LTB, LTB, LTB, LTB, LTB, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, MICA, MICA, MICA, MICA, MICA, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, VARS, VARS, VARS, VARS, VARS, VARS, VARS, XXbac-BPG299F13.16
CADD score	μ = 3.431 [0.17 – 8.268]	**disease gene(s)**	HLA-B, C2, TNF

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Behcet syndrome	1.25×10^{-6}	dbGaP	pha002888	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...]	OMIM	MIM:608579	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM

	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICA	?	ENSG00000204520	atrial appendage	2.93×10-8 (p-value)	GTEx Portal V6	13
XXbac-BPG181B23.7	?	ENSG00000272221	atrial appendage	1.51×10-7 (p-value)	GTEx Portal V6	21
MICA	?	ENSG00000204520	transformed fibroblasts	8.78×10-16 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	2.90×10-18 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	blood	4.17×10-14 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	blood	1.62×10-33 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	breast	2.51×10-10 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	breast	1.83×10-13 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	skeletal muscle	2.44×10-19 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	skeletal muscle	2.86×10-20 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	putamen	3.55×10-8 (p-value)	GTEx Portal V6	21
MICA	?	ENSG00000204520	putamen	4.08×10-7 (p-value)	GTEx Portal V6	11

XXbac-BPG181B23.7	?	ENSG00000272221	EBV lymphocytes	1.99×10-8 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	sun exposed skin	5.53×10-19 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	1.08×10-20 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	unexposed skin	1.60×10-12 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	unexposed skin	3.90×10-11 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	aorta	1.38×10-10 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	aorta	1.27×10-13 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	left ventricle	7.94×10-9 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	left ventricle	2.20×10-12 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	subcutaneous adipocytes	3.63×10-18 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	1.79×10-20 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	caudate basal ganglia	8.43×10-9 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	caudate basal ganglia	1.67×10-8 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	visceral adipocytes	2.35×10-12 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	visceral adipocytes	2.37×10-11 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	stomach	4.11×10-8 (p-value)	GTEx Portal V6	5
MICA	?	ENSG00000204520	cortex	9.16×10-12 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	cortex	2.89×10-9 (p-value)	GTEx Portal V6	21
MICA	?	ENSG00000204520	tibial nerve	3.93×10-13 (p-value)	GTEx Portal V6	22
HLA-B	?	ENSG00000234745	tibial nerve	1.12×10-7 (p-value)	GTEx Portal V6	12
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	7.42×10-17 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	esophagus mucosa	3.85×10-12 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	esophagus mucosa	1.01×10-11 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	pituitary	3.36×10-9 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	pituitary	1.09×10-9 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	testis	3.08×10-10 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	testis	3.09×10-7 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	frontal cortex	4.84×10-9 (p-value)	GTEx Portal V6	13
XXbac-BPG181B23.7	?	ENSG00000272221	pancreas	3.99×10-10 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	pancreas	1.39×10-7 (p-value)	GTEx Portal V6	18
MICA	?	ENSG00000204520	muscularis mucosae	3.04×10-13 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	muscularis mucosae	2.96×10-12 (p-value)	GTEx Portal V6	22
HCG22	?	ENSG00000228789	muscularis mucosae	4.78×10-6 (p-value)	GTEx Portal V6	7
MICA	?	ENSG00000204520	lung	1.15×10-13 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	lung	3.96×10-21 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	tibial artery	1.35×10-16 (p-value)	GTEx Portal V6	22

HLA-B	?	ENSG00000234745	tibial artery	3.93×10-6 (p-value)	GTEx Portal V6	4
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	1.76×10-19 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	thyroid	2.47×10-16 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	3.66×10-17 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	transverse colon	1.21×10-8 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	transverse colon	1.07×10-11 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	liver	4.70×10-8 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	nucleus accumbens	1.16×10-7 (p-value)	GTEx Portal V6	13
XXbac-BPG181B23.7	?	ENSG00000272221	coronary artery	3.81×10-7 (p-value)	GTEx Portal V6	4
MICA	?	ENSG00000204520	sigmoid colon	3.24×10-11 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	sigmoid colon	4.97×10-10 (p-value)	GTEx Portal V6	22
MICA	?	ENSG00000204520	gastroesophageal junction	1.08×10-10 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	gastroesophageal junction	8.57×10-12 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	adrenal gland	7.92×10-7 (p-value)	GTEx Portal V6	4
MICA	?	ENSG00000204520	hippocampus	1.98×10-8 (p-value)	GTEx Portal V6	22
XXbac-BPG181B23.7	?	ENSG00000272221	hippocampus	4.35×10-8 (p-value)	GTEx Portal V6	22
HCG22	ENST00000570223	ILMN_1667229	blood	4.45×10-7 (p-value)	MuTHER consortium	2
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
MICA	ENST00000449934	ILMN_3241692	monocyte	1.19×10-10 (p-value)	Fairfax et al.	2
MICA	ENST00000616296		b-cell	5.10×10-8 (p-value)	Fairfax et al.	2
MICA	ENST00000421350					
MICA	?	ENSG00000204520	vagina	5.53×10-7 (p-value)	GTEx Portal V6	10
LY6G5C	ENST00000383237	ILMN_1763467	blood	4.51×10-6 (p-value)	Westra et al.	1
LY6G5C	ENST00000474395					
LY6G5C	ENST00000375863					
LY6G5C	ENST00000474678					
LY6G5C	ENST00000467098					
LY6G5C	ENST00000460141					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000048013		MICA
		MICA
		HCP5
		MICA
		HCP5
		MICA

			MICA
			HCP5

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444737	1	ENCP00000048058	BAG6 BAG6 BAG6 BAG6 BAG6 BAG6 BAG6
		ENCP00000048034	LTB LTB LTB LTB LTB LTB LTB
		ENCP00000048025	DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B
		ENCP00000047992	HCG27 HCG27 HCG27 HCG27 HCG27 HCG27 HCG27
		ENCP00000048062	CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B
		ENCP00000048036	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1
		ENCP00000048043	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
		ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000048063	GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1

		ENCP00000047991	HCG27 HCG27 HCG27 HCG27 HCG27 HCG27 HCG27
		ENCP00000047975	HCG22 HCG22 HCG22 HCG22 HCG22 HCG22 HCG22 HCG22
		ENCP00000047995	XXbac-BPG299F13.16
		ENCP00000048019	MICB MICB MICB MICB MICB MICB MICB MICB
		ENCP00000048078	ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A
ENCE00000444880	1	ENCP00000048065	CSNK2B GPANK1 GPANK1 GPANK1 CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048041	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
		ENCP00000048069	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048050	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000048017	HCP5 HCP5 HCP5 HCP5
		ENCP00000047997	HLA-C

	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000048023	DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B
ENCP00000048104	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048052	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048110	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048109	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048051	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048033	TNF TNF TNF TNF TNF TNF TNF TNF
ENCP00000048035	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000047998	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C

		HLA-C
	ENCP00000048116	C6orf48 C6orf48 C6orf48 C6orf48 C6orf48
	ENCP00000048043	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
	ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
	ENCP00000048048	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
	ENCP00000048020	MICB MICB MICB MICB MICB MICB MICB MICB
	ENCP00000048134	C2 C2 C2 C2 C2 C2 C2 C2
	ENCP00000048040	UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1
	ENCP00000048037	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487835 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K9ac, H3K36me3, H3K27me3, PolII, Rad21, Egr1, CTCF, Yy1, Tcf12, TAF1, H3K4me2, SP1, USF1, H3K4me3
		HSMMtube	CTCF, H3K9ac, H3K36me3, H2AZ, H3K27ac, H3K4me2, DNase1
		blood (K562)	Ini1, Brg1, Egr1, H3K27ac, DNase1, H3K4me3, H3K36me3, H3K4me1, Max, Rad21, E2F6, H3K9ac, USF1, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, Nrsf, Cfos, PolII, H3K4me2, CTCF
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, CTCF, H3K4me3, DNase1
		muscle (HSMM)	H3K36me3, DNase1, H3K9ac, H3K4me2, H2AZ, H3K79me2, CTCF, H3K4me3, H3K27ac
		liver (HepG2)	DNase1, H3K4me3, H3K27me3, Cmyc, CTCF, Gabp, PolII, H3K79me2, Rad21, TAF1, USF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac
		lung (IMR90)	H3K4me3, H3K9ac, CTCF, H3K4me2, DNase1, H3K79me2
		blood (GM12878)	PolII, H2AZ, DNase1, Egr1, Yy1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac

		nervous (NH-A)	DNase1, H3K9ac, H4K20me1, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K36me3, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
		NHLF	DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3, CTCF
		cervix (HeLa-S3)	DNase1, PolII, CTCF, Gabp, H3K79me2, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cjun, H3K4me2, H3K4me3, PolII, CTCF, DNase1
		A549	H3K4me3, H3K36me3, H3K27ac, DNase1, CTCF, H3K9ac, H3K4me2
ENSR00001703045 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001703047 (open chromatin region)	2	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K36me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HCP5	upstream gene variant	4071	ENST00000541196	?	?	1
HCP5	upstream gene variant	28	ENST00000414046	?	?	1
HCP5	upstream gene variant	4074	ENST00000460889	?	?	1
HCP5	upstream gene variant	4074	ENST00000467369	?	?	1
LINC01149	downstream gene variant	2	ENST00000430364	?	?	7
MICA	upstream gene variant, downstream gene variant	278	ENST00000421350	?	ENSP00000402410	2
MICA	upstream gene variant, downstream gene variant	278	ENST00000449934	NM_001177519.2	ENSP00000413079	2
MICA	downstream gene variant	280	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
XXbac-BPG181B23.6	downstream gene variant	3617	ENST00000440087	?	?	2
XXbac-BPG181B23.7	downstream gene variant	983	ENST00000606743	?	?	2
Y_RNA	downstream gene variant	1478	ENST00000362462	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	19
LINC01149	ENST00000430364	?	?	3
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICA	ENST00000421350	?	ENSP00000402410	1
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
LINC01149	ENST00000430364	?	2

Locus 167 (sentinel SNP rs9469012 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 168 (rs6543140)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,991,163-103,092,575
block size	101,413 bp
variant count	43 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.097 [-2.8 – 1.633]	gene(s) hit or close-by	AC007248.7, AC007278.2, AC007278.3, IL18R1, IL18RAP, MIR4772, SLC9A4
phastCons	μ = 0.055 [0 – 1]	eQTL gene(s)	AC007278.3, IL18R1, IL18RAP, IL1RL1
GERP++	μ = -0.748 [-6.53 – 3.67]	potentially regulated gene(s)	IL1R2, SLC9A2, SLC9A4
CADD score	μ = 3.166 [0.068 – 20.9]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AC007278.3	?	ENSG00000234389	visceral adipocytes	5.97×10^{-7} (p-value)	GTEx Portal V6	30
AC007278.3	?	ENSG00000234389	lung	3.09×10^{-12} (p-value)	GTEx Portal V6	42
IL1RL1	?	ENSG00000115602	lung	9.84×10^{-8} (p-value)	GTEx Portal V6	41
AC007278.3	?	ENSG00000234389	tibial nerve	6.35×10^{-7} (p-value)	GTEx Portal V6	39
AC007278.3	?	ENSG00000234389	atrial appendage	2.87×10^{-6} (p-value)	GTEx Portal V6	13
IL18RAP	?	ENSG00000115607	blood	1.78×10^{-9} (p-value)	GTEx Portal V6	42
AC007278.3	?	ENSG00000234389	blood	9.14×10^{-20} (p-value)	GTEx Portal V6	42
IL18R1	?	ENSG00000115604	thyroid	1.71×10^{-5} (p-value)	GTEx Portal V6	9
AC007278.3	?	ENSG00000234389	thyroid	6.60×10^{-7} (p-value)	GTEx Portal V6	42
AC007278.3	?	ENSG00000234389	esophagus mucosa	4.79×10^{-7} (p-value)	GTEx Portal V6	42
AC007278.3	?	ENSG00000234389	subcutaneous adipocytes	7.55×10^{-6} (p-value)	GTEx Portal V6	13

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	8	no	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000027325		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000257249	2	ENCP00000027299	IL1R2
		ENCP00000027329	SLC9A2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001655327 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		HSMMtube	H3K27me3
		A549	H3K27me3
		blood (K562)	H2AZ
ENSR00001544959 (promoter flanking region)	1	NHLF	DNase1
		liver (HepG2)	H3K27me3
		HSMMtube	H3K27me3, DNase1
		blood (DND-41)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	DNase1
		muscle (HSMM)	DNase1
ENSR00000595671 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF, H3K27me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3
ENSR00000677146 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	CTCF
		A549	H3K27me3
ENSR00001544961 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3, CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF, H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, H3K27me3
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		A549	CTCF, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC007248.7	downstream gene variant	383	ENST00000430344	?	?	3
AC007278.2	upstream gene variant, downstream gene variant	1679	ENST00000436582	?	?	2
AC007278.3	downstream gene variant	733	ENST00000450893	?	?	1

IL18R1	downstream gene variant	722	ENST00000334376	?	ENSP00000334030	1
IL18R1	downstream gene variant	4746	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	1
IL18R1	downstream gene variant	4763	ENST00000409599	?	ENSP00000387211	1
IL18RAP	upstream gene variant	1942	ENST00000450855	?	ENSP00000389815	2
IL18RAP	upstream gene variant, downstream gene variant	1150	ENST00000264260	NM_003853.2	ENSP00000264260	8
IL18RAP	upstream gene variant, downstream gene variant	1331	ENST00000409369	?	ENSP00000387201	7
MIR4772	upstream gene variant	11	ENST00000581495	?	?	1
SLC9A4	upstream gene variant	189	ENST00000295269	NM_001011552.3	ENSP00000295269	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL18R1	ENST00000409599	?	ENSP00000387211	2
IL18R1	ENST00000410040	?	ENSP00000386663	2
IL18R1	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	2
IL18RAP	ENST00000264260	NM_003853.2	ENSP00000264260	9
IL18RAP	ENST00000409369	?	ENSP00000387201	9
SLC9A4	ENST00000295269	NM_001011552.3	ENSP00000295269	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLC9A4	ENST00000295269	NM_001011552.3	ENSP00000295269	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC007278.3	ENST00000450893	?	1

Locus 169 (rs4950937)

SNiPAcard

Block annotations

Block info

genomic range	chr1:203,170,892-203,194,186
block size	23,295 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.235 [-1.349 – 4.997]	gene(s) hit or close-by	CHIT1
phastCons	μ = 0.184 [0 – 1]	eQTL gene(s)	KRT8P29
GERP++	μ = -0.102 [-3.04 – 2.59]	potentially regulated gene(s)	–
CADD score	μ = 5.938 [0.591 – 16.99]	disease gene(s)	CHIT1

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Chitotriosidase deficiency	pathogenic	ClinVar	RCV000010134.1	1
?	HGMD curated	HGMD	CM065066	1
CHITOTRIOSIDASE DEFICIENCY	OMIM curated	OMIM	MIM:600031	1

Disease gene annotation

gene	trait	source DB	source entry/link
CHIT1	CHITOTRIOSIDASE DEFICIENCY	OMIM	MIM:614122

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CHIT1	missense variant	ENST00000503786	?	ENSP00000421617	S/G	Agc/Ggc	?	?	1
CHIT1	missense variant	ENST00000491855	?	ENSP00000423778	S/G	Agc/Ggc	?	?	1
CHIT1	missense variant	ENST00000367229	NM_001270509.1, NM_003465.2	ENSP00000356198	S/G	Agc/Ggc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
KRT8P29	?	ENSG00000236430	tibial nerve	7.68×10-6 (p-value)	GTEx Portal V6	6

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors

ENSR00001527715 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	H3K4me1, H3K27ac, H3K36me3, DNase1
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	DNase1, H3K4me1, H3K18ac, H3K27ac, H4K5ac
		nervous (NH-A)	DNase1
		A549	H3K27me3
ENSR00001527717 (enhancer)	1	HSMMtube	H3K4me1, H3K27ac, DNase1
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	H3K27ac, DNase1
ENSR00001527722 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K27me3
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CHIT1	downstream gene variant	3506	ENST00000491855	?	ENSP00000423778	1
CHIT1	downstream gene variant	3784	ENST00000503786	?	ENSP00000421617	1
CHIT1	downstream gene variant	2476	ENST00000513472	?	?	2
CHIT1	downstream gene variant	3506	ENST00000255427	NM_001256125.1	ENSP00000255427	1
CHIT1	upstream gene variant	2759	ENST00000479483	?	?	2
CHIT1	downstream gene variant	2915	ENST00000367229	NM_001270509.1, NM_003465.2	ENSP00000356198	1
CHIT1	upstream gene variant	2748	ENST00000506427	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CHIT1	ENST00000367229	NM_001270509.1, NM_003465.2	ENSP00000356198	2
CHIT1	ENST00000255427	NM_001256125.1	ENSP00000255427	3
CHIT1	ENST00000484834	?	?	4
CHIT1	ENST00000479483	?	?	1
CHIT1	ENST00000491855	?	ENSP00000423778	2
CHIT1	ENST00000506427	?	?	2
CHIT1	ENST00000503786	?	ENSP00000421617	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CHIT1	ENST00000484834	?	1
CHIT1	ENST00000513472	?	1

CHIT1	ENST00000513472	?	1

Locus 170 (rs2524280)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,327,701-31,426,399
block size	98,699 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.409 [-3.036 – 0.31]	gene(s) hit or close-by	HCP5, HLA-B, HLA-C, HLA-S, LINC01149, MICA, MIR6891, XXbac-BPG181B23.6, XXbac-BPG181B23.7, XXbac-BPG248L24.12, Y_RNA
phastCons	μ = 0.148 [0 – 0.796]	eQTL gene(s)	ATP6V1G2-DDX39B, DDX39B, HCG27, HLA-C, LY6G5C, MICA, MICB, POU5F1, PSORS1C3, XXbac-BPG299F13.17, Y_RNA
GERP++	μ = -0.556 [-2.85 – 0.641]	potentially regulated gene(s)	CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, FGFR3P1, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, TCF19, TCF19, TCF19, TCF19, TCF19, TCF19, TCF19, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P
CADD score	μ = 4.560 [0.038 – 14.35]	disease gene(s)	HLA-B, , VARS2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Beta-2 microglubulin plasma levels	$<2.00\times10^{-12}$	GWAS Catalog	23417110	1
Bipolar disorder	$<1.00\times10^{-8}$	GWAS Catalog	21254220	1

Disease gene annotation

gene	trait	source DB	source entry/link	
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM®
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM®

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HCG27	ENST00000383331	ILMN_1746436	adipocyte	5.34×10^{-6} (p-value)	MuTHER consortium	6
HCG27	ENST00000415276					
MICB	ENST00000538442	ILMN_1708006	skin	3.69×10^{-5} (p-value)	MuTHER consortium	2
MICB	ENST00000399150					
MICB	ENST00000252229					
MICA	?	ENSG00000204520	muscularis mucosae	4.04×10^{-7} (p-value)	GTEx Portal V6	14
Y_RNA	?	ENSG00000199332	muscularis mucosae	2.82×10^{-6} (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	lung	4.47×10^{-9} (p-value)	GTEx Portal V6	14
PSORS1C3	?	ENSG00000204528	lung	1.30×10^{-6} (p-value)	GTEx Portal V6	4

Y_RNA	?	ENSG00000199332	lung	1.27×10-8 (p-value)	GTEx Portal V6	13
MICA	?	ENSG00000204520	transformed fibroblasts	1.29×10-5 (p-value)	GTEx Portal V6	2
Y_RNA	?	ENSG00000199332	transformed fibroblasts	1.19×10-7 (p-value)	GTEx Portal V6	14
PSORS1C3	?	ENSG00000204528	blood	1.47×10-7 (p-value)	GTEx Portal V6	14
MICA	?	ENSG00000204520	blood	1.39×10-6 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	tibial artery	1.10×10-7 (p-value)	GTEx Portal V6	14
Y_RNA	?	ENSG00000199332	tibial artery	3.29×10-8 (p-value)	GTEx Portal V6	14
MICA	?	ENSG00000204520	thyroid	2.25×10-6 (p-value)	GTEx Portal V6	12
PSORS1C3	?	ENSG00000204528	thyroid	2.39×10-9 (p-value)	GTEx Portal V6	15
PSORS1C3	?	ENSG00000204528	skeletal muscle	2.00×10-7 (p-value)	GTEx Portal V6	13
MICA	?	ENSG00000204520	sun exposed skin	1.57×10-9 (p-value)	GTEx Portal V6	15
PSORS1C3	?	ENSG00000204528	sun exposed skin	1.05×10-7 (p-value)	GTEx Portal V6	15
POU5F1	?	ENSG00000204531	sun exposed skin	5.91×10-8 (p-value)	GTEx Portal V6	14
HLA-C	?	ENSG00000204525	sun exposed skin	9.60×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	unexposed skin	1.58×10-6 (p-value)	GTEx Portal V6	4
PSORS1C3	?	ENSG00000204528	left ventricle	9.84×10-9 (p-value)	GTEx Portal V6	13
PSORS1C3	?	ENSG00000204528	subcutaneous adipocytes	1.54×10-6 (p-value)	GTEx Portal V6	13
XXbac-BPG299F13.17	?	ENSG00000272501	subcutaneous adipocytes	4.50×10-6 (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	stomach	4.18×10-7 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	tibial nerve	5.19×10-7 (p-value)	GTEx Portal V6	12
Y_RNA	?	ENSG00000199332	tibial nerve	5.49×10-8 (p-value)	GTEx Portal V6	14
PSORS1C3	?	ENSG00000204528	esophagus mucosa	5.04×10-6 (p-value)	GTEx Portal V6	8
POU5F1	?	ENSG00000204531	esophagus mucosa	5.68×10-7 (p-value)	GTEx Portal V6	14
DDX39B	ENST00000396172	ILMN_1686626	blood	1.27×10-4 (p-value)	Westra et al.	1
DDX39B	ENST00000482195					
DDX39B	ENST00000431908					
DDX39B	ENST00000484566					
DDX39B	ENST00000417023					
ATP6V1G2-DDX39B	ENST00000376185					
DDX39B	ENST00000462256					
DDX39B	ENST00000462421					
DDX39B	ENST00000478365					
DDX39B	ENST00000474961					
DDX39B	ENST00000376177					
DDX39B	ENST00000458640					
MICB	ENST00000538442	ILMN_1708006	blood	4.39×10-6 (p-value)	Westra et al.	1
MICB	ENST00000399150					
MICB	ENST00000252229					
DDX39B	?	ENSG00000198563	testis	3.71×10-6 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	testis	2.37×10-6 (p-value)	GTEx Portal V6	1

HLA-C	?	ENSG00000204525	testis	7.26×10-6 (p-value)	GTEx Portal V6	1
PSORS1C3	ENST00000412143	ILMN_3309634	b-cell	2.64×10-6 (p-value)	Fairfax et al.	3
LY6G5C	ENST00000474395	ILMN_1770209	b-cell	4.42×10-6 (p-value)	Fairfax et al.	3
LY6G5C	ENST00000461572					
LY6G5C	ENST00000467098					
MICA	ENST00000449934	ILMN_3241692	monocyte	3.05×10-8 (p-value)	Fairfax et al.	2
MICA	ENST00000616296		b-cell	1.13×10-5 (p-value)	Fairfax et al.	2
MICA	ENST00000421350					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444777	1	ENCP00000047982	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
		ENCP00000048120	SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4
		ENCP00000048000	WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P
		ENCP00000047981	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
		ENCP00000048009	FGFR3P1
ENCE00000444715	1	ENCP00000047985	TCF19 TCF19 TCF19 TCF19 TCF19 TCF19 TCF19
		ENCP00000047968	VARS2 VARS2 VARS2 VARS2 VARS2 VARS2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494562 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1

ENSR00001494563 (promoter flanking region)	1	Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K79me2
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K79me2
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2
		nervous (NH-A)	H4K20me1
		skin (NHEK)	H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HCP5	upstream gene variant	4563	ENST00000467369	?	?	1
HCP5	upstream gene variant	4560	ENST00000541196	?	?	1
HCP5	upstream gene variant	2184	ENST00000414046	?	?	1
HCP5	upstream gene variant	4563	ENST00000460889	?	?	1
HLA-B	upstream gene variant	2736	ENST00000434333	?	ENSP00000405931	1
HLA-B	upstream gene variant	3019	ENST00000474381	?	?	1
HLA-B	upstream gene variant	2745	ENST00000498007	?	?	1
HLA-B	upstream gene variant	4848	ENST00000497377	?	?	1
HLA-B	upstream gene variant	2737	ENST00000412585	NM_005514.6	ENSP00000399168	1
HLA-B	upstream gene variant	4122	ENST00000463574	?	?	1
HLA-C	upstream gene variant	2766	ENST00000620806	?	ENSP00000480331	1
HLA-S	upstream gene variant	4173	ENST00000425174	?	?	1
LINC01149	upstream gene variant, downstream gene variant	2240	ENST00000430364	?	?	2
MICA	upstream gene variant	1266	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICA	upstream gene variant	989	ENST00000449934	NM_001177519.2	ENSP00000413079	2
MICA	upstream gene variant	1082	ENST00000421350	?	ENSP00000402410	2
MIR6891	upstream gene variant	4608	ENST00000618788	?	?	1
XXbac-BPG181B23.6	downstream gene variant	4106	ENST00000440087	?	?	1
XXbac-BPG181B23.7	downstream gene variant, upstream gene variant	2617	ENST00000606743	?	?	2
XXbac-BPG248L24.12	downstream gene variant	2287	ENST00000603274	?	?	1
Y_RNA	downstream gene variant, upstream gene variant	302	ENST00000362462	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	11
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	2
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	4
MICA	ENST00000421350	?	ENSP00000402410	2

Locus 170 (sentinel SNP rs2524280 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 171 (rs9787249)

SNiPAcard

Block annotations

Block info

genomic range	chr1:40,203,722-40,226,046
block size	22,325 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.180 [-1.636 – 0.37]	gene(s) hit or close-by	BMP8B, PPIE
phastCons	μ = 0.043 [0 – 0.341]	eQTL gene(s)	BMP8B, OXCT2, PPIE
GERP++	μ = -0.163 [-5.61 – 2.6]	potentially regulated gene(s)	MACF1, PPIE
CADD score	μ = 4.840 [0.066 – 12.78]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PPIE	?	ENSG00000084072	atrial appendage	8.35×10^{-30} (p-value)	GTEx Portal V6	20
BMP8B	?	ENSG00000116985	transformed fibroblasts	6.30×10^{-11} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	transformed fibroblasts	2.17×10^{-32} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	breast	1.32×10^{-44} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	blood	8.23×10^{-51} (p-value)	GTEx Portal V6	20
PPIE	ENST00000497370	ILMN_2326737	monocyte	3.23×10^{-18} (p-value)	Fairfax et al.	1
PPIE	ENST00000372835		b-cell	4.24×10^{-24} (p-value)	Fairfax et al.	1
PPIE	ENST00000495526					
PPIE	ENST00000470018					
PPIE	ENST00000480169					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000372830					
PPIE	ENST00000482751					
PPIE	ENST00000356511					
PPIE	?	ENSG00000084072	skeletal muscle	2.42×10^{-19} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	putamen	1.32×10^{-11} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	blood	2.01×10^{-4} (q-value)	SeeQTL DB (HapMap)	6
PPIE	?	ENSG00000084072	EBV lymphocytes	1.64×10^{-13} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	unexposed skin	6.98×10^{-51} (p-value)	GTEx Portal V6	20
BMP8B	?	ENSG00000116985	sun exposed skin	1.15×10^{-7} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	sun exposed skin	8.53×10^{-86} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	ovary	9.46×10^{-22} (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	left ventricle	9.03×10^{-11} (p-value)	GTEx Portal V6	20

PPIE	?	ENSG00000084072	aorta	4.36×10-42 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	subcutaneous adipocytes	6.11×10-82 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	caudate basal ganglia	2.43×10-14 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	visceral adipocytes	4.07×10-50 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	stomach	6.63×10-39 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	cortex	1.80×10-11 (p-value)	GTEx Portal V6	20
OXCT2	?	ENSG00000198754	tibial nerve	2.10×10-10 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	tibial nerve	1.91×10-69 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	pituitary	4.54×10-19 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	esophagus mucosa	8.49×10-63 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	testis	4.29×10-43 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	hypothalamus	9.76×10-11 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	frontal cortex	4.38×10-7 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	uterus	4.53×10-13 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	pancreas	3.18×10-34 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	muscularis mucosae	1.08×10-36 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	lung	4.70×10-76 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	tibial artery	5.45×10-50 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	thyroid	4.69×10-73 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	prostate	3.73×10-21 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	transverse colon	1.09×10-31 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	liver	1.18×10-18 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	terminal ileum	3.83×10-23 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	nucleus accumbens	1.11×10-11 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	spleen	6.43×10-24 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	coronary artery	2.58×10-22 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	sigmoid colon	3.11×10-19 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	vagina	2.89×10-17 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	gastroesophageal junction	6.49×10-21 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	cerebellar hemisphere	4.63×10-10 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	adrenal gland	1.45×10-18 (p-value)	GTEx Portal V6	20
PPIE	?	ENSG00000084072	cerebellum	1.45×10-10 (p-value)	GTEx Portal V6	20
BMP8B	?	ENSG00000116985	cerebellum	1.45×10-7 (p-value)	GTEx Portal V6	20

PPIE	?	ENSG00000084072	hippocampus	1.14×10^{-10} (p-value)	GTEx Portal V6	20
PPIE	ENST00000497370	ILMN_2326737	skin	7.60×10^{-18} (p-value)	MuTHER consortium	4
PPIE	ENST00000495526		blood	6.33×10^{-35} (p-value)	MuTHER consortium	4
PPIE	ENST00000372835		adipocyte	5.02×10^{-10} (p-value)	MuTHER consortium	4
PPIE	ENST00000470018					
PPIE	ENST00000480169					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000372830					
PPIE	ENST00000482751					
PPIE	ENST00000356511					
BMP8B	ENST00000372827	ILMN_2123415	blood	7.55×10^{-13} (p-value)	MuTHER consortium	4
PPIE	ENST00000497370	ILMN_1680341	skin	9.11×10^{-75} (p-value)	MuTHER consortium	4
PPIE	ENST00000495526		blood	2.84×10^{-35} (p-value)	MuTHER consortium	4
PPIE	ENST00000372835		adipocyte	3.73×10^{-38} (p-value)	MuTHER consortium	4
PPIE	ENST00000470213					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000482751					
PPIE	ENST00000372830					
PPIE	ENST00000356511					
PPIE	ENST00000497370	ILMN_2326737	monocyte	3.68×10^{-64} (p-value)	Zeller et al.	2
PPIE	ENST00000495526					
PPIE	ENST00000372835					
PPIE	ENST00000470018					
PPIE	ENST00000480169					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000372830					
PPIE	ENST00000482751					
PPIE	ENST00000356511					
PPIE	ENST00000497370	ILMN_1680341	monocyte	5.74×10^{-52} (p-value)	Zeller et al.	2
PPIE	ENST00000495526					
PPIE	ENST00000372835					
PPIE	ENST00000470213					
PPIE	ENST00000324379					
PPIE	ENST00000475350					
PPIE	ENST00000474804					
PPIE	ENST00000482751					
PPIE	ENST00000372830					
PPIE	ENST00000356511					
PPIE	ENST00000475350	202494_at	blood	4.50×10^{-16} (p-value)	Dixon et al.	1
PPIE	ENST00000497370					
PPIE	ENST00000372835					
PPIE	ENST00000495526					
PPIE	ENST00000482751					
PPIE	ENST00000470213					
PPIE	ENST00000324379					

BMP8B	ENST00000372827	235275_at	blood	1.60×10-17 (p-value)	Dixon et al.	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000418809	1	ENST00000497370, ENST00000495526, ENST00000372835, ENST00000456578, ENST00000480169, ENST00000470213, ENST00000324379, ENST00000475350, ENST00000474804, ENST00000482751, ENST00000372830, ENST00000485507, ENST00000356511	PPIE

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000001965		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000015243	1	ENCP00000001935	MACF1
		ENCP00000001943	MACF1
		ENCP00000001939	MACF1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001579074 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1, H4K8ac
		blood (K562)	H2AZ
		muscle (HSMM)	DNase1
ENSR00000534967 (promoter)	3	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, PolII, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	Yy1, H3K27ac, Max, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K27me3, DNase1, H3K4me3, H3K9ac, Gabp, PolII, TAF1
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, Gabp, H3K79me2, H3K4me3, H3K27ac, H3K9ac
		lung (IMR90)	DNase1, H4K8ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	H3K36me3, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K4me3, H3K9ac
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, H3K4me3, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PPIE	3	ENST00000324379 ENST00000356511 ENST00000372830 ENST00000372835 ENST00000470213 ENST00000475350 ENST00000482751 ENST00000495526 ENST00000497370	hsa-miR-411-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BMP8B	downstream gene variant	1787	ENST00000372827	NM_001720.3	ENSP00000361915	3
BMP8B	downstream gene variant	3177	ENST00000397360	?	ENSP00000380518	1
PPIE	upstream gene variant	149	ENST00000372830	NM_001195007.1	ENSP00000361918	3
PPIE	upstream gene variant, downstream gene variant	134	ENST00000456578	?	?	5
PPIE	upstream gene variant, downstream gene variant	136	ENST00000495526	?	ENSP00000434989	6
PPIE	upstream gene variant	133	ENST00000356511	NM_203456.2	ENSP00000348904	3
PPIE	upstream gene variant, downstream gene variant	149	ENST00000497370	?	ENSP00000433475	6
PPIE	upstream gene variant, downstream gene variant	149	ENST00000372835	?	ENSP00000361925	6
PPIE	upstream gene variant, downstream gene variant	238	ENST00000480169	?	?	10
PPIE	upstream gene variant, downstream gene variant	127	ENST00000485507	?	?	6
PPIE	upstream gene variant, downstream gene variant	149	ENST00000470213	?	ENSP00000431714	6
PPIE	upstream gene variant, downstream gene variant	138	ENST00000482751	?	?	6
PPIE	upstream gene variant, downstream gene variant	130	ENST00000324379	NM_006112.3	ENSP00000312769	6
PPIE	downstream gene variant	662	ENST00000470018	?	ENSP00000436689	7
PPIE	upstream gene variant, downstream gene variant	156	ENST00000475350	?	ENSP00000434888	6
PPIE	upstream gene variant	825	ENST00000467741	?	?	1
PPIE	upstream gene variant, downstream gene variant	152	ENST00000474804	?	ENSP00000432396	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PPIE	ENST00000482751	?	?	13
PPIE	ENST00000497370	?	ENSP00000433475	13
PPIE	ENST00000475350	?	ENSP00000434888	13
PPIE	ENST00000372835	?	ENSP00000361925	13
PPIE	ENST00000485507	?	?	3
PPIE	ENST00000456578	?	?	3
PPIE	ENST00000470018	?	ENSP00000436689	7
PPIE	ENST00000495526	?	ENSP00000434989	13
PPIE	ENST00000474804	?	ENSP00000432396	10
PPIE	ENST00000480169	?	?	4
PPIE	ENST00000372830	NM_001195007.1	ENSP00000361918	17
PPIE	ENST00000470213	?	ENSP00000431714	13
PPIE	ENST00000356511	NM_203456.2	ENSP00000348904	17
PPIE	ENST00000324379	NM_006112.3	ENSP00000312769	13

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
BMP8B	ENST00000372827	NM_001720.3	ENSP00000361915	1

PPIE	ENST00000495526	?	ENSP00000434989	5

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PPIE	ENST00000480169	?	1

Locus 172 (rs9332701)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,474,397-169,484,767
block size	10,371 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.887 [-1.839 – 3.613]	gene(s) hit or close-by	F5
phastCons	μ = 0.500 [0.001 – 1]	eQTL gene(s)	–
GERP++	μ = 2.580 [-0.451 – 5.61]	potentially regulated gene(s)	–
CADD score	μ = 12.966 [0.632 – 25.3]	disease gene(s)	F5

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
F5	STROKE, ISCHEMIC	OMIM	MIM:601367
F5	THROMBOPHILIA DUE TO ACTIVATED PROTEIN C RESISTANCE	OMIM	MIM:188055
F5	PREGNANCY LOSS, RECURRENT, SUSCEPTIBILITY TO, 1	OMIM	MIM:614389
F5	FACTOR V DEFICIENCY	OMIM	MIM:227400
F5	BUDD-CHIARI SYNDROME	OMIM	MIM:600880
F5	Congenital factor V deficiency	OrphaNet	OrphaNet:326
F5	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217
F5	BUDD-CHIARI SYNDROME	OrphaNet	OrphaNet:131
F5	East Texas bleeding disorder	OrphaNet	OrphaNet:391320

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
F5	missense variant	ENST00000367796	?	ENSP00000356770	T/M	aCg/aTg	?	?	1
F5	missense variant	ENST00000367797	NM_000130.4	ENSP00000356771	T/M	aCg/aTg	?	?	1

Putative effect on transcript

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
F5	ENST00000495481	?	1

Locus 173 (rs2786107)

SNiPAcard

Block annotations

Block info

genomic range	chr1:197,310,777-197,310,777
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.427	gene(s) hit or close-by	CRB1
phastCons	0	eQTL gene(s)	–
GERP++	-1.17	potentially regulated gene(s)	–
CADD score	1.07	disease gene(s)	CRB1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CRB1	LEBER CONGENITAL AMAUROSIS 8	DECIPHER	MIM:613835
CRB1	Retinitis pigmentosa-12, autosomal recessive	DECIPHER	MIM:600105
CRB1	RETINITIS PIGMENTOSA 12	OMIM	MIM:600105
CRB1	LEBER CONGENITAL AMAUROSIS 8	OMIM	MIM:613835
CRB1	PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY	OMIM	MIM:172870
CRB1	RETINITIS PIGMENTOSA	OMIM	MIM:268000
CRB1	Retinitis pigmentosa	OrphaNet	OrphaNet:791
CRB1	Pigmented paravenous retinochoroidal atrophy	OrphaNet	OrphaNet:251295
CRB1	Leber congenital amaurosis	OrphaNet	OrphaNet:65

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CRB1	ENST00000367399	NM_001193640.1	ENSP00000356369	1
CRB1	ENST00000475659	?	?	1
CRB1	ENST00000484075	?	ENSP00000433932	1
CRB1	ENST00000538660	NM_001257966.1	ENSP00000438091	1
CRB1	ENST00000535699	NM_001257965.1	ENSP00000438786	1
CRB1	ENST00000367400	NM_201253.2	ENSP00000356370	1

Locus 174 (rs6916921)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,520,426-31,563,240
block size	42,815 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.904 [-2.113 – 0.076]	gene(s) hit or close-by	ATP6V1G2, LST1, LTA, LTB, NCR3, NFKBIL1, TNF
phastCons	μ = 0.014 [0 – 0.08]	eQTL gene(s)	AIF1, ATP6V1G2, BAG6, HLA-B, LST1, MICA, MICB, NCR3, PSORS1C1, XXbac-BPG181B23.7, XXbac-BPG299F13.17
GERP++	μ = -1.943 [-7.11 – 0.109]	potentially regulated gene(s)	AIF1, AIF1, AIF1, AIF1, AIF1, AIF1, AIF1, C2, C2, C2, C2, C2, C2, C2, C2, C2, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CYP21A1P, CYP21A1P, CYP21A1P, CYP21A1P, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCP5, HCP5, HCP5, HCP5, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, MICA, MICA, MICA, MICA, MICA, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, STK19, STK19, STK19, STK19, STK19, STK19, STK19, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, VARS, VARS, VARS, VARS, VARS, VARS, VARS, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.12
CADD score	μ = 3.095 [0.301 – 4.713]	disease gene(s)	NFKBIL1, TNF, LTA, NCR3, C2, HLA-B

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR993387	1
?	HGMD curated	HGMD	CR082032	1
VASCULAR DEMENTIA, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:191160	1

Disease gene annotation

gene	trait	source DB	source entry/link
NFKBIL1	RHEUMATOID ARTHRITIS	OMIM	MIM:180300
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
LTA	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988
LTA	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
NCR3	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507

	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...]	OMIM	MIM:608579	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ATP6V1C2	ENST00000481908	ILMN_1654541	skin	6.39×10-8 (p-value)	MuTHER consortium	3

Locus 174 (sentinel SNP rs6916921 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

ATP6V1G2	ENST00000481998	ILMN_1654541	skin	6.20×10-9 (p-value)	MuTHER consortium	3
ATP6V1G2	ENST00000303892		blood	1.36×10-6 (p-value)	MuTHER consortium	3
ATP6V1G2	ENST00000483170		adipocyte	2.03×10-5 (p-value)	MuTHER consortium	3
ATP6V1G2	ENST00000376151					
AIF1	ENST00000337917	ILMN_1792473	blood	1.13×10-8 (p-value)	MuTHER consortium	3
AIF1	ENST00000466820					
AIF1	ENST00000497362					
AIF1	ENST00000376059					
AIF1	ENST00000376049					
BAG6	?	ENSG00000204463	muscularis mucosae	2.06×10-9 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	lung	6.51×10-7 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	transformed fibroblasts	1.48×10-9 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	transformed fibroblasts	2.17×10-7 (p-value)	GTEx Portal V6	6
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	6.61×10-7 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	tibial artery	5.46×10-8 (p-value)	GTEx Portal V6	6
LST1	ENST00000303757	ILMN_1718936	monocyte	4.42×10-5 (p-value)	Fairfax et al.	4
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
MICB	ENST00000399150	ILMN_1708006	monocyte	8.70×10-5 (p-value)	Fairfax et al.	3
MICB	ENST00000538442		b-cell	6.58×10-4 (p-value)	Fairfax et al.	3
MICB	ENST00000252229					
MICA	?	ENSG00000204520	thyroid	4.01×10-7 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	skeletal muscle	2.55×10-8 (p-value)	GTEx Portal V6	6
MICB	?	ENSG00000204516	blood	3.91×10-3 (q-value)	SeeQTL DB (HapMap)	2
NCR3	?	ENSG00000204475	blood	9.04×10-5 (q-value)	SeeQTL DB (HapMap)	1
MICA	?	ENSG00000204520	sun exposed skin	2.35×10-10 (p-value)	GTEx Portal V6	6
PSORS1C1	?	ENSG00000204540	sun exposed skin	2.60×10-6 (p-value)	GTEx Portal V6	4
BAG6	?	ENSG00000204463	sun exposed skin	2.55×10-6 (p-value)	GTEx Portal V6	3
MICA	?	ENSG00000204520	unexposed skin	6.49×10-7 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	aorta	1.94×10-6 (p-value)	GTEx Portal V6	5
MICA	?	ENSG00000204520	subcutaneous adipocytes	9.87×10-8 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	subcutaneous adipocytes	7.18×10-9 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	visceral adipocytes	8.77×10-8 (p-value)	GTEx Portal V6	6
BAG6	?	ENSG00000204463	cortex	3.07×10-7 (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	tibial nerve	1.35×10-8 (p-value)	GTEx Portal V6	6

MICA	?	ENSG00000204520	tibial nerve	1.35×10-8 (p-value)	GTEx Portal V6	6
HLA-B	?	ENSG00000234745	tibial nerve	1.10×10-5 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	sigmoid colon	6.30×10-7 (p-value)	GTEx Portal V6	5
MICA	?	ENSG00000204520	esophagus mucosa	3.60×10-8 (p-value)	GTEx Portal V6	6
XXbac-BPG299F13.17	?	ENSG00000272501	esophagus mucosa	5.43×10-6 (p-value)	GTEx Portal V6	5
BAG6	ENST00000375976	ILMN_1705364	blood	4.62×10-6 (p-value)	Westra et al.	2
BAG6	ENST00000362049					
BAG6	ENST00000375964					
BAG6	ENST00000464869					
BAG6	ENST00000422948					
BAG6	ENST00000211379					
BAG6	ENST00000441793					
BAG6	ENST00000439687					
BAG6	ENST00000464126					
BAG6	?	ENSG00000204463	cerebellum	9.53×10-9 (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444842	1	ENCP00000048041	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
		ENCP00000048013	HCP5 MICA MICA HCP5 MICA HCP5 MICA MICA HCP5
		ENCP00000048050	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000047997	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
		ENCP00000048039	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3
		ENCP00000048046	PRRC2A PRRC2A

	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048109	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048051	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048150	STK19 STK19 STK19 STK19 STK19 STK19
ENCP00000048003	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048033	TNF TNF TNF TNF TNF TNF TNF TNF
ENCP00000048035	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000048116	C6orf48 C6orf48 C6orf48 C6orf48 C6orf48
ENCP00000048043	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048048	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A

	PRRC2A
ENCP00000048093	MSH5 MSH5 MSH5 MSH5 MSH5 MSH5 MSH5 MSH5
ENCP00000048006	XXbac-BPG248L24.12
ENCP00000048134	C2 C2 C2 C2 C2 C2 C2 C2
ENCP00000048037	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000048020	MICB MICB MICB MICB MICB MICB MICB MICB
ENCP00000048065	CSNK2B GPANK1 GPANK1 GPANK1 CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B GPANK1 GPANK1 GPANK1 GPANK1
ENCP00000048069	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
ENCP00000048002	XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10
ENCP00000048038	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3

ENCP00000048005	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048017	HCP5 HCP5 HCP5 HCP5
ENCP00000048023	DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B
ENCP00000047991	HCG27 HCG27 HCG27 HCG27 HCG27 HCG27 HCG27
ENCP00000048104	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048052	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048110	VARS VARS VARS VARS VARS VARS VARS
ENCP00000047998	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000048004	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048157	CYP21A1P CYP21A1P CYP21A1P CYP21A1P
ENCP00000048063	GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1

	ENCP00000048040	UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487867 (CTCF binding site)	1	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac
		blood (K562)	CTCF
		monocytes (Monocytes-CD14+)	H3K4me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	BATF, DNase1, CTCF, H3K79me2, H3K27ac, H3K4me2, H3K9ac, PolII, EBF1
		lung (IMR90)	H3K27me3
ENSR00001494573 (promoter)	1	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K27me3, H3K36me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	EBF1, POU2F2, Cmyc, PolII, DNase1, Egr1, Yy1, Gabp, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me2
ENSR00000264419 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K4me2, H3K4me3, H4K20me1, H3K36me3
		liver (HepG2)	H3K27me3
		blood (K562)	H2AZ

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ATP6V1G2	upstream gene variant	4222	ENST00000415099	?	ENSP00000390148	1
LST1	downstream gene variant	24	ENST00000376086	NM_001166538.1	ENSP00000365254	1
LST1	downstream gene variant	256	ENST00000490742	?	ENSP00000418578	1
LST1	downstream gene variant	122	ENST00000211921	?	ENSP00000211921	1
LST1	downstream gene variant	175	ENST00000460834	?	?	1
LST1	downstream gene variant	122	ENST00000339530	NM_205837.2	ENSP00000339201	1
LST1	downstream gene variant	23	ENST00000376096	?	ENSP00000365264	1
LST1	downstream gene variant	1091	ENST00000419073	?	?	1
LST1	downstream gene variant	175	ENST00000376110	?	ENSP00000365278	1
LST1	downstream gene variant	23	ENST00000376089	?	ENSP00000365257	1
LST1	downstream gene variant	23	ENST00000376090	?	ENSP00000365258	1
LST1	downstream gene variant	23	ENST00000396101	?	ENSP00000379408	1
LST1	downstream gene variant	23	ENST00000376093	NM_007161.3	ENSP00000365261	1
LST1	downstream gene variant	270	ENST00000376092	?	ENSP00000365260	1
LST1	downstream gene variant	23	ENST00000303757	NM_205840.2	ENSP00000303649	1

LST1	downstream gene variant	23	ENST00000376099	?	ENSP00000365267	1
LST1	downstream gene variant	23	ENST00000464526	?	?	1
LST1	downstream gene variant	306	ENST00000396112	?	ENSP00000379418	1
LST1	downstream gene variant	23	ENST00000418507	NM_205838.2	ENSP00000405900	1
LST1	downstream gene variant	1202	ENST00000433492	?	ENSP00000399869	1
LST1	downstream gene variant	23	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	downstream gene variant	175	ENST00000464044	?	ENSP00000417645	1
LTA	downstream gene variant	381	ENST00000489638	?	?	2
LTA	downstream gene variant	381	ENST00000454783	NM_001159740.2	ENSP00000403495	2
LTA	downstream gene variant	381	ENST00000418386	NM_000595.3	ENSP00000413450	2
LTA	downstream gene variant	381	ENST00000471842	?	?	2
LTB	downstream gene variant	4505	ENST00000429299	NM_002341.1	ENSP00000410481	1
LTB	downstream gene variant	4574	ENST00000482429	?	?	1
LTB	downstream gene variant	4505	ENST00000446745	NM_009588.1	ENSP00000416113	1
LTB	downstream gene variant	4475	ENST00000483972	?	?	1
NCR3	downstream gene variant, upstream gene variant	342	ENST00000376072	NM_001145467.1	ENSP00000365240	2
NCR3	downstream gene variant, upstream gene variant	365	ENST00000376071	?	ENSP00000365239	2
NCR3	upstream gene variant	2651	ENST00000495600	?	?	1
NCR3	upstream gene variant	2504	ENST00000491161	?	?	1
NCR3	upstream gene variant	2478	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	upstream gene variant	2478	ENST00000340027	NM_147130.2	ENSP00000342156	1
NFKBIL1	downstream gene variant	4415	ENST00000496233	?	ENSP00000437148	1
NFKBIL1	downstream gene variant	4161	ENST00000473655	?	?	1
NFKBIL1	downstream gene variant	3291	ENST00000376148	NM_005007.3	ENSP00000365318	1
NFKBIL1	downstream gene variant	3291	ENST00000376145	NM_001144961.1	ENSP00000365315	1
NFKBIL1	downstream gene variant	3291	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	1
TNF	upstream gene variant	862	ENST00000449264	NM_000594.3	ENSP00000398698	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NFKBIL1	ENST00000473655	?	?	1
NFKBIL1	ENST00000376148	NM_005007.3	ENSP00000365318	1
NFKBIL1	ENST00000496233	?	ENSP00000437148	1
NFKBIL1	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	1
NFKBIL1	ENST00000376145	NM_001144961.1	ENSP00000365315	1
TNF	ENST00000449264	NM_000594.3	ENSP00000398698	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
NCR3	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	ENST00000340027	NM_147130.2	ENSP00000342156	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NCR3	ENST00000491161	?	1
NCR3	ENST00000495600	?	1

Locus 175 (rs2516470)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,407,331-31,407,331
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.181	gene(s) hit or close-by	HCP5, LINC01149
phastCons	0.004	eQTL gene(s)	ATP6V1G2, C4A, HCG22, HCG27, HCP5, HLA-B, LST1, LY6G5C, MICA, MICB, NOTCH4, PRRC2A, XXbac-BPG181B23.7, XXbac-BPG248L24.12
GERP++	0.574	potentially regulated gene(s)	–
CADD score	0.943	disease gene(s)	C4A, HLA-B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147
C4A	Behcet disease	OrphaNet	OrphaNet:117
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG181B23.7	?	ENSG00000272221	pancreas	1.41×10^{-6} (p-value)	GTEx Portal V6	1
LST1	ENST00000376099	ILMN_2345353	blood	1.26×10^{-6} (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					

LST1	ENST00000376089					
LST1	ENST00000376096					
LST1	ENST00000418507					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000460834					
LST1	ENST00000211921					
NOTCH4	ENST00000474612	ILMN_1711157	adipocyte	2.39×10-6 (p-value)	MuTHER consortium	1
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
LST1	ENST00000376099	ILMN_1688373	blood	1.10×10-6 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					
LST1	ENST00000376089					
LST1	ENST00000396112					
LST1	ENST00000418507					
LST1	ENST00000376096					
LST1	ENST00000490742					
LST1	ENST00000211921					
LST1	ENST00000460834					
HCG22	ENST00000570223	ILMN_1667229	blood	1.22×10-14 (p-value)	MuTHER consortium	1
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
PRRC2A	ENST00000376033	ILMN_2408179	blood	4.00×10-5 (p-value)	MuTHER consortium	1
PRRC2A	ENST00000487089					
PRRC2A	ENST00000376007					
PRRC2A	ENST00000487839					
PRRC2A	ENST00000462617					
PRRC2A	ENST00000482441					
PRRC2A	ENST00000492691					
HCG27	ENST00000383331	ILMN_1746436	blood	9.73×10-6 (p-value)	MuTHER consortium	1
HCG27	ENST00000415276					
MICA	?	ENSG00000204520	muscularis mucosae	2.27×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	muscularis mucosae	1.74×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	muscularis mucosae	4.11×10-13 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	lung	1.18×10-8 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	lung	5.02×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	lung	5.34×10-14 (p-value)	GTEx Portal V6	1

XXbac-BPG181B23.7	?	ENSG00000272221	atrial appendage	3.15×10^{-9} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	transformed fibroblasts	1.05×10^{-10} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	3.16×10^{-9} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	1.22×10^{-16} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	blood	1.68×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	blood	3.05×10^{-11} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	blood	1.02×10^{-22} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	breast	4.04×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	breast	1.95×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	breast	3.01×10^{-13} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial artery	8.19×10^{-13} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	1.04×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	7.67×10^{-25} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	thyroid	2.48×10^{-12} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	1.15×10^{-8} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	1.74×10^{-18} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	skeletal muscle	9.01×10^{-14} (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	skeletal muscle	2.97×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	5.23×10^{-9} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	skeletal muscle	3.27×10^{-26} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	EBV lymphocytes	3.23×10^{-9} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	transverse colon	1.40×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	transverse colon	1.05×10^{-10} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	ovary	2.40×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	sun exposed skin	6.42×10^{-11} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	1.40×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	1.13×10^{-21} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	unexposed skin	3.40×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	unexposed skin	1.11×10^{-12} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	aorta	2.08×10^{-8} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	aorta	2.04×10^{-11} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	left ventricle	1.18×10^{-8} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	4.42×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	left ventricle	3.36×10^{-13} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	subcutaneous adipocytes	2.68×10^{-10} (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	subcutaneous adipocytes	6.39×10^{-6} (p-value)	GTEx Portal V6	1

XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	1.99×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	3.18×10-19 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	stomach	1.34×10-10 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	visceral adipocytes	5.49×10-12 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	cortex	1.59×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	sigmoid colon	5.87×10-14 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	coronary artery	5.37×10-10 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial nerve	6.18×10-9 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	1.05×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	4.76×10-14 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	tibial nerve	7.59×10-6 (p-value)	GTEx Portal V6	1
HCG22	?	ENSG00000228789	tibial nerve	2.39×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	esophagus mucosa	1.40×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	3.86×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	esophagus mucosa	1.15×10-12 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	gastroesophageal junction	9.01×10-10 (p-value)	GTEx Portal V6	1
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	1.07×10-4 (p-value)	Westra et al.	1
ATP6V1G2	ENST00000303892					
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					
HCP5	ENST00000414046	ILMN_1803945	blood	2.10×10-5 (p-value)	Westra et al.	1
HCP5	ENST00000541196					
LY6G5C	ENST00000383237	ILMN_1763467	blood	1.04×10-5 (p-value)	Westra et al.	1
LY6G5C	ENST00000474395					
LY6G5C	ENST00000375863					
LY6G5C	ENST00000474678					
LY6G5C	ENST00000467098					
LY6G5C	ENST00000460141					
?	?	ILMN_1883997	blood	9.46×10-7 (p-value)	Westra et al.	1
XXbac-BPG181B23.7	?	ENSG00000272221	adrenal gland	1.48×10-7 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	testis	2.87×10-6 (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	testis	6.24×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	testis	5.88×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LINC01149	upstream gene variant	2113	ENST00000430364	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	1

Locus 176 (rs11578770)

SNiPAcard

Block annotations

Block info

genomic range	chr1:160,834,616-160,899,100
block size	64,485 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.163 [-0.178 – 0.435]	gene(s) hit or close-by	CD244, ITLN1, RP11-312J18.6, RP11-544M22.1
phastCons	μ = 0.032 [0 – 0.099]	eQTL gene(s)	–
GERP++	μ = 0.255 [-0.854 – 1.97]	potentially regulated gene(s)	ARHGAP30, FCER1G, ITLN1, MPZ, MPZ, NIT1, PPOX, RP11-312J18.3, RP11-404F10.2, SLAMF1, SLAMF7, TOMM40L, USF1, USP21
CADD score	μ = 3.439 [1.402 – 9.687]	disease gene(s)	USF1, MPZ, PPOX

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
USF1	HYPERLIPIDEMIA, FAMILIAL COMBINED	OMIM	MIM:144250
USF1	HYPERLIPIDEMIA, COMBINED, 1	OMIM	MIM:602491
MPZ	ADIE PUPIL	OMIM	MIM:103100
MPZ	ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA	OMIM	MIM:180800
MPZ	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B	OMIM	MIM:118200
MPZ	NEUROPATHY, CONGENITAL HYPOMYELINATING OR AMYELINATING, [...]	OMIM	MIM:605253
MPZ	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2I	OMIM	MIM:607677
MPZ	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2J	OMIM	MIM:607736
MPZ	CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE D	OMIM	MIM:607791
MPZ	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS	OMIM	MIM:145900
PPOX	PORPHYRIA VARIEGATA	OMIM	MIM:176200
MPZ	Autosomal dominant Charcot-Marie-Tooth disease type 2J	OrphaNet	OrphaNet:99943
MPZ	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D	OrphaNet	OrphaNet:100046
MPZ	Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain	OrphaNet	OrphaNet:324585
MPZ	ROUSSY-LEVY SYNDROME	OrphaNet	OrphaNet:3115
MPZ	Charcot-Marie-Tooth disease type 1B	OrphaNet	OrphaNet:101082
MPZ	Dejerine-Sottas syndrome	OrphaNet	OrphaNet:64748
MPZ	Autosomal dominant Charcot-Marie-Tooth disease type 2I	OrphaNet	OrphaNet:99942
PPOX	PORPHYRIA VARIEGATA	OrphaNet	OrphaNet:79473

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000040974	1	ENCP00000005175	NIT1
		ENCP00000005210	MPZ MPZ
		ENCP00000005140	RP11-404F10.2
		ENCP00000005151	ITLN1
		ENCP00000005186	PPOX

		ENCP00000005149	RP11-312J18.3
		ENCP00000005138	SLAMF1
		ENCP00000005204	TOMM40L
		ENCP00000005169	ARHGAP30
		ENCP00000005163	USF1
		ENCP00000005143	SLAMF7
		ENCP00000005168	ARHGAP30
		ENCP00000005142	SLAMF7
		ENCP00000005201	FCER1G
		ENCP00000005185	PPOX
ENCE00000041210	1	ENCP00000005182	USP21

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001587177 (open chromatin region)	1	blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CD244	upstream gene variant	2126	ENST00000492063	?	ENSP00000432636	1
CD244	upstream gene variant	2126	ENST00000322302	NM_001166664.1	ENSP00000313619	1
CD244	upstream gene variant	2066	ENST00000368033	NM_001166663.1	ENSP00000357012	1
CD244	upstream gene variant	1971	ENST00000368034	NM_016382.3	ENSP00000357013	1
ITLN1	downstream gene variant	693	ENST00000326245	NM_017625.2	ENSP00000323587	1
ITLN1	downstream gene variant	3466	ENST00000487531	?	?	1
RP11-312J18.6	downstream gene variant	2429	ENST00000427339	?	?	1
RP11-544M22.1	upstream gene variant	3155	ENST00000356006	?	?	1

Locus 177 (rs7529589)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,646,176-196,717,153
block size	70,978 bp
variant count	77 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.104 [-7.227 – 2.829]	**gene(s) hit or close-by**	CFH
phastCons	μ = 0.040 [0 – 0.596]	**eQTL gene(s)**	CFH, CFHR1, CFHR3, RP11-75C23.1
GERP++	μ = -0.621 [-9.79 – 2.77]	**potentially regulated gene(s)**	–
CADD score	μ = 1.853 [0.001 – 8.248]	**disease gene(s)**	CFHR1, CFHR3, CFH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Age-related macular degeneration	$<1.00\times10^{-31}$	GWAS Catalog	23577725	1
Age-related macular degeneration (extreme sampling)	$<9.00\times10^{-24}$	GWAS Catalog	23577725	1
Age-related macular degeneration	$<7.00\times10^{-32}$	GWAS Catalog	23326517	2
Age-related macular degeneration (CNV)	$<1.00\times10^{-108}$	GWAS Catalog	22705344	1
Age-related macular degeneration (GA)	$<5.00\times10^{-67}$	GWAS Catalog	22705344	1
Age-related macular degeneration	$<2.00\times10^{-47}$	GWAS Catalog	22694956	1
Age-related macular degeneration	$<1.00\times10^{-261}$	GWAS Catalog	21665990	1
Age-related macular degeneration	$<4.00\times10^{-117}$	GWAS Catalog	20385826	1
Age-related macular degeneration	$<6.00\times10^{-16}$	GWAS Catalog	20385819	1
Age-related macular degeneration	$<4.00\times10^{-8}$	GWAS Catalog	15761122	1
HWESASXX*	2.58×10^{-6}	Metabolomics GWAS Server	24816252	4
Macular degeneration	9.52×10^{-75}	dbGaP	pha002890	3
Macular degeneration	2.07×10^{-88}	dbGaP	pha002869	3
Macular degeneration	6.04×10^{-18}	dbGaP	pha002856	4
Macular degeneration	1.75×10^{-19}	dbGaP	pha000002	1
Macular degeneration	1.29×10^{-25}	dbGaP	pha000001	4

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Basal laminar drusen	pathogenic	ClinVar	RCV000018016.27	1
Age-related macular degeneration 4	risk factor	ClinVar	RCV000018015.2	1
?	HGMD curated	HGMD	CM057396	1
?	HGMD curated	HGMD	CM051020	1
MACULAR DEGENERATION, AGE-RELATED, 4, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:134370	1

Disease gene annotation

gene	trait	source DB	source entry/link

CFHR1	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581	orphanet
CFHR1	C3 glomerulonephritis	OrphaNet	OrphaNet:329931	orphanet
CFHR1	Dense deposit disease	OrphaNet	OrphaNet:93571	orphanet
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581	orphanet
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579	orphanet
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421	orphanet
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571	orphanet
CFH	Familial drusen	OrphaNet	OrphaNet:75376	orphanet
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CFH	missense variant	ENST00000359637	?	ENSP00000352658	H/Y	Cat/Tat	tolerated	benign	1
CFH	missense variant	ENST00000630130	NM_001014975.2	ENSP00000487250	H/Y	Cat/Tat	tolerated	benign	1
CFH	missense variant	ENST00000367429	NM_000186.3	ENSP00000356399	H/Y	Cat/Tat	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFH	ENST00000359637	ILMN_1657803	skin	9.43×10^{-8} (p-value)	MuTHER consortium	33
CFH	ENST00000630130		adipocyte	1.02×10^{-5} (p-value)	MuTHER consortium	33
CFH	ENST00000466229	ILMN_1810910	blood	4.35×10^{-6} (p-value)	Westra et al.	32
CFH	ENST00000359637					
CFH	ENST00000367429					
CFH	ENST00000630130					
CFHR1	?	ENSG00000244414	liver	6.46×10^{-8} (p-value)	GTEx Portal V6	75
CFHR3	?	ENSG00000116785	thyroid	5.91×10^{-7} (p-value)	GTEx Portal V6	26
CFH	?	ENSG00000000971	tibial nerve	2.39×10^{-7} (p-value)	GTEx Portal V6	36
CFHR1	?	ENSG00000244414	subcutaneous adipocytes	4.65×10^{-8} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	subcutaneous adipocytes	2.27×10^{-8} (p-value)	GTEx Portal V6	1
RP11-75C23.1	?	ENSG00000230260	nucleus accumbens	6.02×10^{-6} (p-value)	GTEx Portal V6	5
CFHR1	?	ENSG00000244414	transformed fibroblasts	3.21×10^{-5} (p-value)	GTEx Portal V6	5
CFHR3	?	ENSG00000116785	adrenal gland	6.26×10^{-6} (p-value)	GTEx Portal V6	4
CFHR3	?	ENSG00000116785	muscularis mucosae	3.73×10^{-6} (p-value)	GTEx Portal V6	3
CFHR1	?	ENSG00000244414	spleen	5.75×10^{-6} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	transverse colon	1.31×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	16	no	0.004	1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000550120 (CTCF binding site)	3	NHLF	CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	DNase1
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K36me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF, H3K36me3
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF
		A549	CTCF
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFH	downstream gene variant	3639	ENST00000496761	?	?	2
CFH	downstream gene variant	519	ENST00000466229	?	?	1
CFH	downstream gene variant	62	ENST00000359637	?	ENSP00000352658	15
CFH	upstream gene variant	112	ENST00000470918	?	?	4
CFH	downstream gene variant	61	ENST00000630130	NM_001014975.2	ENSP00000487250	15
CFH	downstream gene variant	519	ENST00000367429	NM_000186.3	ENSP00000356399	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CFH	ENST00000630130	NM_001014975.2	ENSP00000487250	A	gcA/gcC	1
CFH	ENST00000359637	?	ENSP00000352658	A	gcA/gcC	1
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	A	gcA/gcC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFH	ENST00000630130	NM_001014975.2	ENSP00000487250	36
CFH	ENST00000466229	?	?	74
CFH	ENST00000359637	?	ENSP00000352658	36
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	74

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CFH	ENST00000466229	?	4

Locus 178 (rs2949833)

SNiPAcard

Block annotations

Block info

genomic range	chr7:45,984,820-45,994,378
block size	9,559 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.316 [-0.373 – 1.005]	**gene(s) hit or close-by**	–
phastCons	μ = 0.030 [0.025 – 0.036]	**eQTL gene(s)**	IGFBP3
GERP++	μ = -0.830 [-1.09 – -0.569]	**potentially regulated gene(s)**	–
CADD score	μ = 1.589 [0.255 – 2.923]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IGFBP3	ENST00000275521	ILMN_2396875	blood	6.57×10^{-5} (p-value)	Westra et al.	1
IGFBP3	ENST00000381086					
IGFBP3	ENST00000613132					

Locus 179 (rs2523570)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,329,734-31,333,914
block size	4,181 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.707 [-2.823 – 0.83]	**gene(s) hit or close-by**	DHFRP2, HLA-B, HLA-C, RNU6-283P, XXbac-BPG248L24.12
phastCons	μ = 0.042 [0 – 0.185]	**eQTL gene(s)**	FLOT1, HCG22, HCP5, HLA-B, SKIV2L, XXbac-BPG248L24.12
GERP++	μ = -0.072 [-1.92 – 0.83]	**potentially regulated gene(s)**	CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GTF2H4, GTF2H4, GTF2H4, GTF2H4, GTF2H4, GTF2H4, GTF2H4, HCP5, HCP5, HCP5, HCP5, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PSORS1C1, PSORS1C1, PSORS1C1, PSORS1C1, PSORS1C1, PSORS1C1, PSORS1C1, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, VARS, VARS, VARS, VARS, VARS, VARS, VARS, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.12
CADD score	μ = 2.437 [0.078 – 4.885]	**disease gene(s)**	HLA-B, SKIV2L, , , , , , , TNF, , , , , , VARS2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162

TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...]	OMIM	MIM:608579	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG248L24.12	?	ENSG00000271581	pancreas	1.22×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	muscularis mucosae	8.92×10-12 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	lung	4.02×10-13 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	1.05×10-10 (p-value)	GTEx Portal V6	6
HLA-B	?	ENSG00000234745	blood	2.32×10-7 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	blood	3.21×10-18 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	breast	1.65×10-14 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	2.50×10-11 (p-value)	GTEx Portal V6	11
FLOT1	?	ENSG00000137312	thyroid	3.53×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	7.04×10-11 (p-value)	GTEx Portal V6	11
HLA-B	?	ENSG00000234745	skeletal muscle	1.41×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	1.03×10-18 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	EBV lymphocytes	9.39×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	transverse colon	1.80×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	liver	2.78×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	1.94×10-12 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	unexposed skin	5.48×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	aorta	1.88×10-10 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	1.15×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	7.46×10-13 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	5.61×10-10 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	cortex	1.69×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	2.22×10-10 (p-value)	GTEx Portal V6	11

XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	2.56×10-11 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	gastroesophageal junction	4.28×10-11 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	adrenal gland	4.12×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	testis	4.56×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	frontal cortex	1.01×10-6 (p-value)	GTEx Portal V6	1
HCG22	ENST00000570223	ILMN_1667229	blood	3.80×10-5 (p-value)	MuTHER consortium	2
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
SKIV2L	ENST00000484835	ILMN_1666512	monocyte	3.43×10-4 (p-value)	Fairfax et al.	1
SKIV2L	ENST00000375394					
SKIV2L	ENST00000474839					
SKIV2L	ENST00000470453					
SKIV2L	ENST00000465703					
SKIV2L	ENST00000491994					
SKIV2L	ENST00000471818					
SKIV2L	ENST00000483553					
SKIV2L	ENST00000485349					
?	?	1557242_at	blood	1.30×10-10 (p-value)	Dixon et al.	1
HCP5	ENST00000414046	ILMN_1803945	blood	7.20×10-7 (p-value)	Westra et al.	1
HCP5	ENST00000541196					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444665	1	ENCP00000048065	CSNK2B GPANK1 GPANK1 GPANK1 CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048069	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048002	XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10

ENCP00000048005	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048017	HCP5 HCP5 HCP5 HCP5
ENCP00000048050	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000047997	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000048023	DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B
ENCP00000048104	VARS VARS VARS VARS VARS VARS VARS
ENCP00000047959	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
ENCP00000047963	GTF2H4 GTF2H4 GTF2H4 GTF2H4 GTF2H4 GTF2H4
ENCP00000048052	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048110	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048109	VARS VARS VARS VARS

	VARS VARS VARS
ENCP00000048051	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048003	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048033	TNF TNF TNF TNF TNF TNF TNF
ENCP00000047998	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000048035	LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000048004	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048048	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048063	GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1
ENCP00000047979	PSORS1C1 PSORS1C1 PSORS1C1 PSORS1C1 PSORS1C1

	PSORS1C1
ENCP00000048006	XXbac-BPG248L24.12
ENCP00000048040	UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1
ENCP00000048037	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000048020	MICB MICB MICB MICB MICB MICB MICB MICB
ENCP00000047966	VARS2 VARS2 VARS2 VARS2 VARS2 VARS2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DHFRP2	downstream gene variant	215	ENST00000414224	?	?	11
HLA-B	upstream gene variant	4770	ENST00000412585	NM_005514.6	ENSP00000399168	1
HLA-B	upstream gene variant	4778	ENST00000498007	?	?	1
HLA-B	upstream gene variant	4769	ENST00000434333	?	ENSP00000405931	1
HLA-C	upstream gene variant	4799	ENST00000620806	?	ENSP00000480331	1
RNU6-283P	upstream gene variant	3997	ENST00000364788	?	?	4
XXbac-BPG248L24.12	downstream gene variant	4320	ENST00000603274	?	?	1

Locus 180 (rs3132645)

SNiPAcard

Block annotations

Block info

genomic range	chr6:29,970,960-30,792,117
block size	821,158 bp
variant count	107 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.141 [-3.802 – 4.141]	**gene(s) hit or close-by**	ABCF1, AL662797.1, AL662800.2, ATAT1, C6orf136, DHX16, FLOT1, GNL1, HCG17, HCG18, HCG19P, HCG20, HCG4P3, HLA-J, HLA-L, HLA-N, IER3, LINC00243, MDC1, MICC, MRPS18B, NRM, PAIP1P1, PPP1R10, PPP1R11, PPP1R18, PTMAP1, RN7SL353P, RNF39, RPP21, SUCLA2P1, TRIM10, TRIM15, TRIM26, TRIM26BP, TRIM31, TRIM31-AS1, TRIM39, TRIM39-RPP21, TRIM40, TUBB, UBQLN1P1, XXbac-BPG252P9.10, XXbac-BPG252P9.9, XXbac-BPG283O16.9, Y_RNA, ZNRD1, ZNRD1-AS1, ZNRD1-AS1_3
phastCons	μ = 0.130 [0 – 1]	**eQTL gene(s)**	BTN3A2, BTN3A3, C2, CCHCR1, CFB, FLOT1, GABBR1, HCG17, HCG20, HCG22, HCP5, HIST1H3I, HLA-A, HLA-B, HLA-C, HLA-F-AS1, HLA-G, HLA-H, HLA-J, HLA-K, HLA-L, HLA-U, HLA-V, IER3, LINC00243, MICB, NDUFS1, OR5V1, PPP1R18, PSORS1C1, RNF39, RPP21, TMEM154, TRIM26BP, TRIM31, TRIM39-RPP21, VARS2, WASF5P, ZFP57, ZNRD1, ZNRD1-AS1_3
GERP++	μ = -0.462 [-9.98 – 5.1]	**potentially regulated gene(s)**	ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, HCG19P, HCG19P, HCG19P, HCG19P, HCG19P, HCG19P, HCG19P, NRM, NRM, NRM, NRM, NRM, NRM, NRM, RNF39, RNF39, RNF39, RNF39, RNF39, RNF39, RNF39, RNF39, TRIM26, TRIM26, TRIM26, TRIM26, TRIM26, TRIM26, TRIM26, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31
CADD score	μ = 4.127 [0.015 – 22.4]	**disease gene(s)**	ZFP57, NDUFS1, C2, CFB, VARS2, TUBB, HLA-A, HLA-B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Autism spectrum disorder, attention deficit-hyperactivity disorder, bipolar disorder, major depressive disorder, and schizophrenia (combined)	$<8.00\times10^{-9}$	GWAS Catalog	23453885	1
AIDS progression	$<5.00\times10^{-7}$	GWAS Catalog	19115949	1
3-indoxyl sulfate	8.34×10^{-5}	Metabolomics GWAS Server	24816252	1
Lupus erythematosus, systemic	6.18×10^{-6}	dbGaP	pha002867	3
Lupus erythematosus, systemic	6.09×10^{-10}	dbGaP	pha002848	3

Disease gene annotation

gene	trait	source DB	source entry/link
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410
NDUFS1	MITOCHONDRIAL COMPLEX I DEFICIENCY	OMIM	MIM:252010
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917

TUBB	CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS [...]	OMIM	MIM:615771	OMIM
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410	OMIM
NDUFS1	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000	OMIM
NDUFS1	Mitochondrial Respiratory Chain Complex I Deficiency (nuclear genes)	DECIPHER	MIM:252010	OMIM
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886	orphanet
HLA-A	Birdshot chorioretinopathy	OrphaNet	OrphaNet:179	orphanet
NDUFS1	Isolated NADH-CoQ reductase deficiency	OrphaNet	OrphaNet:2609	orphanet
NDUFS1	Leigh syndrome with leukodystrophy	OrphaNet	OrphaNet:255241	orphanet
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	orphanet
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PPP1R18	missense variant	ENST00000615527	?	ENSP00000480270	R/G	Agg/Ggg	?	?	1
PPP1R18	missense variant	ENST00000274853	NM_133471.3	ENSP00000274853	R/G	Agg/Ggg	?	?	1
PPP1R18	missense variant	ENST00000399199	NM_001134870.1	ENSP00000382150	R/G	Agg/Ggg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1654987	blood	3.60×10-6 (p-value)	MuTHER consortium	58
			adipocyte	2.42×10-6 (p-value)		
HLA-L	ENST00000463348	ILMN_1675282	blood	1.92×10-6 (p-value)	MuTHER consortium	47
ZNRD1	ENST00000471008	ILMN_2398587	blood	2.39×10-6 (p-value)	MuTHER consortium	44
ZNRD1	ENST00000359374					
ZNRD1-AS1_3	ENST00000611686					
ZNRD1	ENST00000332435					
ZNRD1	ENST00000376782					
ZNRD1	ENST00000376785					
ZNRD1	ENST00000463141					
TRIM31	ENST00000376734	ILMN_1748685	skin	4.22×10-6 (p-value)	MuTHER consortium	25
TRIM31	ENST00000484583					
TRIM31	ENST00000471695					
TRIM31	ENST00000468264					
HLA-V	ENST00000446817	ILMN_1660923	blood	1.32×10-9 (p-value)	MuTHER consortium	59
HLA-V	ENST00000429037		adipocyte	1.91×10-6 (p-value)	MuTHER consortium	59
			skin	4.10×10-6 (p-value)	MuTHER consortium	59
HCG22	ENST00000570223	ILMN_1667229	blood	2.57×10-6 (p-value)	MuTHER consortium	40
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					

GABBR1	ENST00000491829	ILMN_2395375	blood	1.94×10-5 (p-value)	MuTHER consortium	18
GABBR1	ENST00000377012					
GABBR1	ENST00000377034					
GABBR1	ENST00000478931					
GABBR1	ENST00000472823					
GABBR1	ENST00000377016					
VARS2	ENST00000321897	ILMN_1737585	skin	2.79×10-7 (p-value)	MuTHER consortium	36
VARS2	ENST00000625423					
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
HCP5	ENST00000414046	ILMN_1803945	skin	5.42×10-9 (p-value)	MuTHER consortium	18
HCP5	ENST00000541196					
MICB	ENST00000538442	ILMN_1708006	skin	5.95×10-9 (p-value)	MuTHER consortium	18
MICB	ENST00000399150					
MICB	ENST00000252229					
IER3	ENST00000376377	ILMN_1682717	skin	2.84×10-5 (p-value)	MuTHER consortium	1
IER3	ENST00000259874					
ZFP57	?	ENSG00000204644	muscularis mucosae	6.78×10-8 (p-value)	GTEx Portal V6	82
PPP1R18	?	ENSG00000146112	muscularis mucosae	2.64×10-6 (p-value)	GTEx Portal V6	22
HLA-C	?	ENSG00000204525	muscularis mucosae	1.12×10-6 (p-value)	GTEx Portal V6	7
HLA-B	?	ENSG00000234745	muscularis mucosae	1.89×10-6 (p-value)	GTEx Portal V6	3
FLOT1	?	ENSG00000137312	lung	6.00×10-8 (p-value)	GTEx Portal V6	91
HLA-J	?	ENSG00000204622	lung	1.58×10-12 (p-value)	GTEx Portal V6	104
HLA-G	?	ENSG00000204632	lung	2.21×10-7 (p-value)	GTEx Portal V6	89
HLA-H	?	ENSG00000206341	lung	2.13×10-8 (p-value)	GTEx Portal V6	101
ZFP57	?	ENSG00000204644	lung	6.53×10-11 (p-value)	GTEx Portal V6	82
LINC00243	?	ENSG00000214894	lung	4.13×10-6 (p-value)	GTEx Portal V6	4
HLA-J	?	ENSG00000204622	atrial appendage	1.61×10-6 (p-value)	GTEx Portal V6	4
ZFP57	?	ENSG00000204644	atrial appendage	6.27×10-8 (p-value)	GTEx Portal V6	54
ZFP57	?	ENSG00000204644	transformed fibroblasts	3.56×10-8 (p-value)	GTEx Portal V6	81
HLA-J	?	ENSG00000204622	transformed fibroblasts	3.90×10-6 (p-value)	GTEx Portal V6	44
HLA-C	?	ENSG00000204525	transformed fibroblasts	7.51×10-8 (p-value)	GTEx Portal V6	75
HCG20	?	ENSG00000228022	transformed fibroblasts	2.40×10-7 (p-value)	GTEx Portal V6	10
HLA-H	?	ENSG00000206341	transformed fibroblasts	2.47×10-5 (p-value)	GTEx Portal V6	1
HLA-J	?	ENSG00000204622	blood	4.01×10-9 (p-value)	GTEx Portal V6	98
HLA-H	?	ENSG00000206341	blood	7.77×10-9 (p-value)	GTEx Portal V6	93
ZFP57	?	ENSG00000204644	blood	2.24×10-10 (p-value)	GTEx Portal V6	83
HLA-G	?	ENSG00000204632	blood	9.33×10-8 (p-value)	GTEx Portal V6	52
HLA-F-AS1	?	ENSG00000214922	blood	1.37×10-6 (p-value)	GTEx Portal V6	70

LINC00243	?	ENSG00000214894	blood	2.67×10-14 (p-value)	GTEx Portal V6	103
HLA-K	?	ENSG00000230795	blood	1.57×10-9 (p-value)	GTEx Portal V6	92
CCHCR1	?	ENSG00000204536	blood	4.75×10-6 (p-value)	GTEx Portal V6	6
HLA-C	?	ENSG00000204525	blood	8.58×10-7 (p-value)	GTEx Portal V6	16
VARS2	?	ENSG00000137411	blood	6.90×10-6 (p-value)	GTEx Portal V6	1
HLA-J	?	ENSG00000204622	breast	1.40×10-7 (p-value)	GTEx Portal V6	27
ZFP57	?	ENSG00000204644	breast	6.42×10-8 (p-value)	GTEx Portal V6	47
HLA-C	?	ENSG00000204525	breast	3.52×10-6 (p-value)	GTEx Portal V6	5
HLA-U	?	ENSG00000228078	tibial artery	5.57×10-7 (p-value)	GTEx Portal V6	72
HLA-J	?	ENSG00000204622	tibial artery	3.63×10-8 (p-value)	GTEx Portal V6	33
HLA-H	?	ENSG00000206341	tibial artery	5.15×10-7 (p-value)	GTEx Portal V6	39
ZFP57	?	ENSG00000204644	tibial artery	2.39×10-9 (p-value)	GTEx Portal V6	83
HLA-C	?	ENSG00000204525	tibial artery	1.92×10-9 (p-value)	GTEx Portal V6	82
HLA-J	?	ENSG00000204622	thyroid	6.77×10-7 (p-value)	GTEx Portal V6	39
ZFP57	?	ENSG00000204644	thyroid	2.65×10-10 (p-value)	GTEx Portal V6	83
ZNRD1	?	ENSG00000066379	thyroid	1.59×10-5 (p-value)	GTEx Portal V6	1
FLOT1	?	ENSG00000137312	thyroid	2.35×10-10 (p-value)	GTEx Portal V6	96
CCHCR1	?	ENSG00000204536	thyroid	8.47×10-10 (p-value)	GTEx Portal V6	93
PSORS1C1	?	ENSG00000204540	thyroid	5.65×10-7 (p-value)	GTEx Portal V6	11
PPP1R18	?	ENSG00000146112	thyroid	1.44×10-7 (p-value)	GTEx Portal V6	36
HLA-J	?	ENSG00000204622	skeletal muscle	1.24×10-9 (p-value)	GTEx Portal V6	103
HLA-H	?	ENSG00000206341	skeletal muscle	1.43×10-6 (p-value)	GTEx Portal V6	58
ZFP57	?	ENSG00000204644	skeletal muscle	1.78×10-6 (p-value)	GTEx Portal V6	23
HLA-C	?	ENSG00000204525	skeletal muscle	1.59×10-12 (p-value)	GTEx Portal V6	82
ZFP57	?	ENSG00000204644	liver	2.45×10-7 (p-value)	GTEx Portal V6	5
HLA-J	?	ENSG00000204622	sun exposed skin	2.03×10-7 (p-value)	GTEx Portal V6	20
HLA-H	?	ENSG00000206341	sun exposed skin	9.76×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	sun exposed skin	9.47×10-10 (p-value)	GTEx Portal V6	82
VARS2	?	ENSG00000137411	sun exposed skin	6.86×10-9 (p-value)	GTEx Portal V6	42
HLA-C	?	ENSG00000204525	sun exposed skin	3.64×10-7 (p-value)	GTEx Portal V6	26
ZFP57	?	ENSG00000204644	unexposed skin	1.63×10-6 (p-value)	GTEx Portal V6	4
ZFP57	?	ENSG00000204644	left ventricle	5.64×10-7 (p-value)	GTEx Portal V6	6
PSORS1C1	?	ENSG00000204540	left ventricle	3.69×10-7 (p-value)	GTEx Portal V6	9
HLA-H	?	ENSG00000206341	left ventricle	8.80×10-6 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	aorta	5.31×10-11 (p-value)	GTEx Portal V6	83
PPP1R18	?	ENSG00000146112	aorta	5.47×10-8 (p-value)	GTEx Portal V6	77
HLA-B	?	ENSG00000234745	aorta	6.92×10-6 (p-value)	GTEx Portal V6	1

HCG17	?	ENSG00000270604	subcutaneous adipocytes	3.78×10-8 (p-value)	GTEx Portal V6	70
ZFP57	?	ENSG00000204644	subcutaneous adipocytes	2.47×10-9 (p-value)	GTEx Portal V6	82
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	6.58×10-7 (p-value)	GTEx Portal V6	57
VARS2	?	ENSG00000137411	subcutaneous adipocytes	6.34×10-8 (p-value)	GTEx Portal V6	49
CCHCR1	?	ENSG00000204536	subcutaneous adipocytes	6.45×10-9 (p-value)	GTEx Portal V6	46
ZFP57	?	ENSG00000204644	stomach	7.56×10-7 (p-value)	GTEx Portal V6	65
HLA-C	?	ENSG00000204525	stomach	1.66×10-7 (p-value)	GTEx Portal V6	76
ZFP57	?	ENSG00000204644	visceral adipocytes	1.11×10-8 (p-value)	GTEx Portal V6	22
CCHCR1	?	ENSG00000204536	visceral adipocytes	2.13×10-6 (p-value)	GTEx Portal V6	7
HLA-H	?	ENSG00000206341	spleen	3.40×10-6 (p-value)	GTEx Portal V6	4
ZFP57	?	ENSG00000204644	tibial nerve	4.33×10-9 (p-value)	GTEx Portal V6	81
CCHCR1	?	ENSG00000204536	tibial nerve	1.48×10-7 (p-value)	GTEx Portal V6	77
PPP1R18	?	ENSG00000146112	tibial nerve	2.17×10-6 (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	tibial nerve	4.29×10-6 (p-value)	GTEx Portal V6	34
HLA-J	?	ENSG00000204622	esophagus mucosa	1.95×10-7 (p-value)	GTEx Portal V6	31
ZFP57	?	ENSG00000204644	esophagus mucosa	1.13×10-12 (p-value)	GTEx Portal V6	83
HLA-C	?	ENSG00000204525	esophagus mucosa	2.41×10-8 (p-value)	GTEx Portal V6	68
MICB	?	ENSG00000204516	esophagus mucosa	4.51×10-6 (p-value)	GTEx Portal V6	4
HLA-A	?	ENSG00000206503	brain	3.82×10-6 (q-value)	SeeQTL DB (Myers et al.)	4
?	?	ILMN_1679389	monocyte	1.31×10-14 (p-value)	Zeller et al.	8
?	?	ILMN_1666078	monocyte	1.81×10-37 (p-value)	Zeller et al.	8
?	?	ILMN_1671054	monocyte	1.64×10-45 (p-value)	Zeller et al.	8
HLA-G	ENST00000376828	ILMN_1656670	monocyte	3.91×10-19 (p-value)	Zeller et al.	8
HLA-G	ENST00000360323					
HLA-G	ENST00000478519					
HLA-G	ENST00000478355					
HLA-G	ENST00000428701					
HLA-G	ENST00000376815					
HLA-G	ENST00000376818					
BTN3A2	ENST00000396934	ILMN_1676528	monocyte	1.68×10-40 (p-value)	Zeller et al.	8
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
?	?	ILMN_1820787	monocyte	2.53×10-24 (p-value)	Zeller et al.	8
BTN3A2	ENST00000524459	ILMN_1700067	monocyte	8.29×10-28 (p-value)	Zeller et al.	8
BTN3A2	ENST00000528541					
BTN3A3	ENST00000467524					
BTN3A3	ENST00000482451					
BTN3A3	ENST00000494393					
BTN3A2	ENST00000527417					
BTN3A3	ENST00000244519					
BTN3A2	ENST00000528222					
BTN3A3	ENST00000474790					
BTN3A2	ENST00000377708					

BTN3A3	ENST00000496719					
BTN3A2	ENST00000396948					
BTN3A2	ENST00000527422					
BTN3A2	ENST00000356386					
?	?	ILMN_1883997	monocyte	1.74×10-44 (p-value)	Zeller et al.	8
HLA-C	ENST00000470363	ILMN_2150787	monocyte	3.78×10-23 (p-value)	Zeller et al.	7
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	ENST00000466892					
?	?	ILMN_1893395	monocyte	1.16×10-14 (p-value)	Zeller et al.	1
HIST1H3I	ENST00000616365	ILMN_2207865	b-cell	1.43×10-4 (p-value)	Fairfax et al.	8
ZNRD1	ENST00000471008	ILMN_2398587	b-cell	2.62×10-4 (p-value)	Fairfax et al.	2
ZNRD1	ENST00000359374					
ZNRD1-AS1_3	ENST00000611686					
ZNRD1	ENST00000376785					
ZNRD1	ENST00000376782					
ZNRD1	ENST00000332435					
ZNRD1	ENST00000463141					
C2	ENST00000486124	ILMN_1710740	monocyte	7.85×10-6 (p-value)	Fairfax et al.	2
C2	ENST00000469372					
CFB	ENST00000425368					
C2	ENST00000497706					
C2	ENST00000442278					
C2	ENST00000452323					
C2	ENST00000383177					
C2	ENST00000299367					
C2	ENST00000482060					
?	?	ILMN_1679389	b-cell	4.36×10-6 (p-value)	Fairfax et al.	17
IER3	ENST00000376377	ILMN_1682717	monocyte	2.83×10-5 (p-value)	Fairfax et al.	20
IER3	ENST00000259874					
RPP21	ENST00000489124	ILMN_2186597	b-cell	4.61×10-4 (p-value)	Fairfax et al.	1
RPP21	ENST00000466327					
RPP21	ENST00000436442					
TRIM39-RPP21	ENST00000623385					
RPP21	ENST00000473266					
TRIM39-RPP21	ENST00000513556					
RPP21	ENST00000498414					
RPP21	ENST00000433076					
RPP21	ENST00000442966					
RPP21	ENST00000491477					
RPP21	ENST00000428040					
OR5V1	ENST00000543825	ILMN_1683352	b-cell	3.80×10-6 (p-value)	Fairfax et al.	10
OR5V1	ENST00000377154					
VARS2	ENST00000321897	ILMN_1737585	monocyte	5.36×10-7 (p-value)	Fairfax et al.	2
VARS2	ENST00000625423		b-cell	5.12×10-5 (p-value)	Fairfax et al.	2
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					

VARS2	ENST00000477288						
VARS2	ENST00000473916						
VARS2	ENST00000476162						
ZFP57	?	ENSG00000204644	transverse colon	2.63×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	transverse colon	2.66×10-8 (p-value)	GTEx Portal V6	66
TRIM26BP	?	ENSG00000236475	transverse colon	2.59×10-6 (p-value)	GTEx Portal V6	19
CCHCR1	?	ENSG00000204536	transverse colon	1.82×10-6 (p-value)	GTEx Portal V6	3
ZFP57	?	ENSG00000204644	coronary artery	2.82×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	coronary artery	2.14×10-6 (p-value)	GTEx Portal V6	20
HLA-C	ENST00000470363	216526_x_at	blood	1.50×10-8 (p-value)	Dixon et al.	4
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000466892					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
RNF39	?	ENSG00000204618	pancreas	2.26×10-6 (p-value)	GTEx Portal V6	1
WASF5P	?	ENSG00000231402	pancreas	3.25×10-8 (p-value)	GTEx Portal V6	42
HLA-C	?	ENSG00000204525	pancreas	1.65×10-6 (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	cerebellar hemisphere	1.84×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	cerebellum	1.25×10-6 (p-value)	GTEx Portal V6	1
HLA-G	?	ENSG00000204632	EBV lymphocytes	1.79×10-6 (p-value)	GTEx Portal V6	4
HCG17	?	ENSG00000270604	hypothalamus	1.06×10-6 (p-value)	GTEx Portal V6	4

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
HLA-A	?	ENSG00000206503	chr6	brain	6.61×10-4 (q-value)	SeeQTL DB (Myers et al.)	4
?	?	ILMN_1752592	chr6	monocyte	8.64×10-29 (p-value)	Zeller et al.	8
HLA-C	ENST00000470363	ILMN_2150787	chr6	monocyte	2.91×10-19 (p-value)	Zeller et al.	1
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						
?	?	ILMN_1893395	chr6	monocyte	1.06×10-12 (p-value)	Zeller et al.	4
?	?	ILMN_1715169	chr6	monocyte	9.75×10-15 (p-value)	Zeller et al.	6
?	?	ILMN_1661266	chr6	monocyte	4.95×10-15 (p-value)	Zeller et al.	6
TMEM154	ENST00000304385	ILMN_2088124	chr4	blood	1.16×10-10 (p-value)	Westra et al.	1
NDUFS1	ENST00000455934	ILMN_1728810	chr2	blood	5.94×10-8 (p-value)	Westra et al.	1
NDUFS1	ENST00000233190						
NDUFS1	ENST00000423725						
BTN3A2	ENST00000524682	209846_s_at	chr6	blood	3.10×10-11 (p-value)	Dixon et al.	6
BTN3A2	ENST00000604202						

BTN3A2	ENST00000396934						
BTN3A2	ENST00000532294						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000377708						
BTN3A2	ENST00000527422						
BTN3A2	ENST00000531055						
BTN3A2	ENST00000396948						
BTN3A2	ENST00000356386						
BTN3A2	ENST00000396934	212613_at	chr6	blood	1.40×10-8 (p-value)	Dixon et al.	3
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000244519	204820_s_at	chr6	blood	3.20×10-10 (p-value)	Dixon et al.	5
BTN3A2	ENST00000396934						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000480110						
HLA-C	ENST00000470363	216526_x_at	chr6	blood	3.20×10-8 (p-value)	Dixon et al.	1
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000466892						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
JUND	MA0491.1	8	yes	0.000	1
FOSL1	MA0477.1	4	yes	0.000	1
SP1	MA0079.3	10	no	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000047900		HCG19P
		HCG19P
		HCG19P
		HCG19P
		HCG19P
		HCG19P
ENCP00000047941		FLOT1
		FLOT1
		FLOT1
		FLOT1
		FLOT1
		FLOT1
		FLOT1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444288	1	ENCP00000047932	NRM
			NRM

			NRM NRM NRM NRM NRM
		ENCP00000047889	TRIM26 TRIM26 TRIM26 TRIM26 TRIM26 TRIM26 TRIM26 TRIM26
ENCE00000444247	1	ENCP00000047877	RNF39 RNF39 RNF39 RNF39 RNF39 RNF39 RNF39 RNF39
		ENCP00000047957	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
		ENCP00000047879	TRIM31 TRIM31 TRIM31 TRIM31 TRIM31 TRIM31 TRIM31 TRIM31
ENCE00000444553	1	ENCP00000047941	FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1
ENCE00000444465	1	ENCP00000047921	ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487584 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00000487598 (promoter flanking region)	2	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	FOSL2, Jund, USF1, H3K4me1, H3K4me2, HNF4A, FOXA1
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1, H3K27me3

		nervous (NH-A)	DNase1, H3K27me3
ENSR00001702960 (enhancer)	1	liver (HepG2)	H3K36me3
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001702961 (enhancer)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF
		HSMMtube	H3K27me3, CTCF
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	CTCF, H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, H3K36me3
		blood (GM12878)	CTCF
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	CTCF, H3K27me3
		NHLF	CTCF, H3K27me3
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		A549	CTCF
ENSR00000487633 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001494491 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K4me2, Rad21, Nrsf, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, H3K27me3, DNase1
		blood (GM12878)	DNase1, Rad21, CTCF
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	H3K27me3, CTCF
		skin (NHEK)	CTCF, H3K27me3, DNase1
		NHLF	CTCF, H3K27me3
		Osteobl	CTCF, H3K27me3
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	Nrsf, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, DNase1, H3K27me3, CTCF
		A549	H3K27me3, CTCF
ENSR00001494494 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	DNase1, H3K27me3
		breast (HMEC)	DNase1
ENSR00001032027 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	DNase1, H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	DNase1
		liver (HepG2)	H3K4me2, H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1, H3K27me3
		skin (NHEK)	H3K27me3, DNase1
		NHLF	DNase1, H3K27me3

		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K4me3, H3K27me3
		endothelium (HUVEC)	H3K27me3
		A549	H3K27me3
ENSR00000487656 e! (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		HSMMtube	H3K27me3, DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		liver (HepG2)	DNase1, H3K27ac, Cmyc, FOXA1, HNF4A, Jund, HNF4G, HDAC2, H3K4me1, p300
		lung (IMR90)	DNase1
		blood (GM12878)	DNase1, H3K27ac, H3K4me2
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K27ac
		NHLF	H3K27ac, DNase1
		Osteobl	H3K4me2, H3K27ac
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K27me3
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Max, DNase1, H3K27ac, Cjun
		A549	DNase1
ENSR00000487671 e! (promoter)	2	embryonic stem cell (H1ESC)	DNase1, TAF1, H3K4me2, SP1, Srf, H3K4me3, HDAC2, CTCF, H3K27ac, H3K36me3, H3K27me3, PolII, Rad21, Egr1, Jund, Sin3Ak20, H3K9ac
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, CTCF, H3K79me2
		blood (K562)	Ini1, Egr1, CTCF, H3K36me3, DNase1, H3K4me3, H3K4me2, PolII, FOSL1, H3K27ac, Max, ZBTB7A, Srf, H3K4me1, H3K9ac, Gabp, Cmyc, H3K79me2, HEY1, H2AZ, Rad21, TAF1, ELF1
		skin (NHDF-AD)	H3K36me3, H3K4me1, CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1, H2AZ
		liver (HepG2)	DNase1, Gabp, PolII, Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3
		lung (IMR90)	DNase1, H4K8ac, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K56ac, CTCF
		blood (GM12878)	ELF1, Rad21, H3K79me2, H3K4me3, Pax5, H3K27ac, H3K4me2, CTCF, H3K9ac, H3K36me3, Gabp, Yy1, EBF1, PolII, H2AZ, Tcf12, DNase1, Egr1
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K36me3, DNase1, CTCF, H3K4me3, H3K4me2, H3K4me1, H3K27ac, H3K9ac
		NHLF	DNase1, H3K4me3, H3K4me1, CTCF, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K9ac, H3K4me3, H3K36me3, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	DNase1, Jund, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, CTCF, H3K36me3, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K27ac, PolII, CTCF, DNase1, Cmyc, H3K9ac, H3K4me3, H3K4me2, Cjun, Max
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000487679 e! (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K27me3, PolII, Rad21, Egr1, Jund, TAF7, Sin3Ak20, CTCF, SIX5, H3K27ac, ATF3, Yy1, TAF1, H3K4me2, SP1, USF1, H3K36me3, H3K9ac, H3K4me3
		HSMMtube	H3K36me3, H2AZ, H3K4me3, CTCF, H3K27ac, H3K4me2, H3K9ac, DNase1
		blood (K562)	H3K27ac, H3K36me3, DNase1, H3K4me3, CTCF, H3K4me2, Max, SP2, PU1, H3K9ac, USF1, H3K79me2, HEY1, H2AZ, Rad21, TAF1, Cfos, CTCFL, Cmyc, PolII, H4K20me1, H3K27me3, Yy1, Ini1, Brg1, Egr1
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K36me3, H3K4me1, CTCF, H4K20me1, H3K4me3, DNase1, H3K9ac
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, Rad21, TAF1, USF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3, H3K36me3, H3K79me2, PolII, ATF3, Yy1, H4K20me1
		lung (IMR90)	DNase1, H4K8ac, H4K20me1, H3K79me2, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K56ac, CTCF
		blood (GM12878)	Pbx3, TAF1, ATF3, SIX5, PolII, BCLAF1, H2AZ, Tcf12, DNase1, Egr1, Yy1, ELF1, Rad21, H3K79me2, H3K4me3, USF1, H3K27ac, Cfos, H3K4me2, CTCF, H3K9ac, H3K36me3
		nervous (NH-A)	DNase1, H3K9ac, H4K20me1, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, H3K36me3, DNase1
		NHLF	H3K36me3, H3K27ac, H3K9ac, DNase1, H3K4me3, H3K4me1, CTCF
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2, CTCF, H3K36me3
		blood (DND-41)	H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K36me3, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	H3K36me3, CTCF, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K9ac, Nrsf, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, PolII, Cmyc, CTCF, Ini1, H3K36me3

		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, CTCF, Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000487687 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		HSMMtube	H3K36me3
		blood (K562)	H3K27me3, PU1, H3K36me3
		skin (NHDF-AD)	H3K36me3
		muscle (HSMM)	H3K36me3
		liver (HepG2)	DNase1, H3K4me1, H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	Yy1, PU1, PolII, H3K4me1, DNase1, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K4me1, H3K36me3
		cervix (HeLa-S3)	H3K27ac, PolII
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me1
		endothelium (HUVEC)	PolII, H3K36me3, DNase1
		A549	H3K36me3
ENSR00000487711 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		liver (HepG2)	H3K4me1, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me2
		blood (GM12878)	H2AZ, DNase1, H3K27ac
		lung (IMR90)	DNase1, H3K27ac
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	DNase1, H2AZ
ENSR00000487714 (promoter flanking region)	1	endothelium (HUVEC)	DNase1
		liver (HepG2)	H3K4me1, FOXA1
		blood (K562)	H3K4me1
ENSR00000662312 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K27me3, PolII, TAF7
		Osteobl	H3K27ac
		blood (K562)	USF1, HEY1, H2AZ, Cjun, Cfos, PolII, MEF2A, Max, Ini1, Brg1, DNase1, FOSL1, H3K27ac
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K27ac, DNase1
		muscle (HSMM)	H2AZ
		cervix (HeLa-S3)	DNase1, PolII, TAF1, H3K9ac, Jund, H3K27ac, H3K4me1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me1, H3K27ac, PolII, DNase1, Cmyc
		liver (HepG2)	DNase1, FOSL2, Jund, PolII, H3K4me1
		lung (IMR90)	DNase1, H2BK12ac, H3K18ac, H2BK120ac, H2BK20ac, H4K91ac
		blood (GM12878)	EBF1, PolII, BATF, DNase1, IRF4, BCL3, BCL11A, Pax5, NFKB
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K27ac, DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ZNRD1	1	ENST00000332435 ENST00000359374 ENST00000376782 ENST00000376785 ENST00000463141 ENST00000471008	hsa-miR-199a-5p hsa-miR-199b-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABCF1	upstream gene variant	2298	ENST00000376545	NM_001090.2	ENSP00000365728	1
ABCF1	upstream gene variant	2338	ENST00000441867	?	ENSP00000405512	1
ABCF1	upstream gene variant	1503	ENST00000542772	?	?	1
ABCF1	upstream gene variant	2281	ENST00000326195	NM_001025091.1	ENSP00000313603	1

ABCF1	upstream gene variant	2347	ENST00000468958	?	ENSP00000440893	1
ABCF1	downstream gene variant	1451	ENST00000475993	?	ENSP00000445100	1
AL662797.1	upstream gene variant, downstream gene variant	112	ENST00000620079	?	?	3
AL662800.2	downstream gene variant	2370	ENST00000583820	?	?	1
ATAT1	downstream gene variant	4727	ENST00000462304	?	?	1
ATAT1	downstream gene variant	4306	ENST00000330083	NM_001031722.2	ENSP00000327832	1
ATAT1	downstream gene variant	4306	ENST00000376485	?	ENSP00000365668	1
ATAT1	downstream gene variant	4306	ENST00000471782	?	?	1
ATAT1	downstream gene variant	4306	ENST00000479562	?	?	1
ATAT1	downstream gene variant	2875	ENST00000493388	?	?	1
ATAT1	downstream gene variant	4306	ENST00000468713	?	?	1
C6orf136	upstream gene variant	137	ENST00000467801	?	ENSP00000419610	1
C6orf136	upstream gene variant	3482	ENST00000488383	?	ENSP00000419968	1
C6orf136	upstream gene variant	3848	ENST00000446773	?	ENSP00000415810	1
C6orf136	upstream gene variant	3466	ENST00000484551	?	ENSP00000418599	1
C6orf136	upstream gene variant	3553	ENST00000463794	?	ENSP00000418223	1
C6orf136	upstream gene variant	255	ENST00000468785	?	ENSP00000419460	1
C6orf136	upstream gene variant	3591	ENST00000376471	NM_145029.3	ENSP00000365654	1
C6orf136	upstream gene variant	3466	ENST00000293604	NM_001161376.1	ENSP00000293604	1
C6orf136	upstream gene variant	3500	ENST00000376473	NM_001109938.2	ENSP00000365656	1
C6orf136	upstream gene variant	1482	ENST00000460172	?	?	1
C6orf136	upstream gene variant, downstream gene variant	1216	ENST00000493705	?	?	2
DHX16	upstream gene variant	3372	ENST00000415603	?	ENSP00000399101	1
DHX16	downstream gene variant	1990	ENST00000376437	?	ENSP00000365620	1
DHX16	downstream gene variant, upstream gene variant	1990	ENST00000376442	NM_001164239.1, NM_003587.4	ENSP00000365625	2
FLOT1	upstream gene variant	384	ENST00000476729	?	?	2
FLOT1	downstream gene variant	1925	ENST00000454845	?	ENSP00000391341	1
FLOT1	downstream gene variant	176	ENST00000484693	?	?	2
FLOT1	downstream gene variant	1577	ENST00000418160	?	ENSP00000404300	1
FLOT1	downstream gene variant	1670	ENST00000484168	?	?	1
FLOT1	upstream gene variant	325	ENST00000487376	?	?	2
FLOT1	downstream gene variant	1592	ENST00000445853	?	ENSP00000398834	1
FLOT1	downstream gene variant	1671	ENST00000416018	?	ENSP00000412058	1
FLOT1	downstream gene variant	1574	ENST00000470643	?	?	1
GNL1	downstream gene variant	4911	ENST00000487166	?	?	1
GNL1	downstream gene variant	198	ENST00000376621	NM_005275.3	ENSP00000365806	2

HCG17	upstream gene variant	1993	ENST00000455957	?	?	2
HCG17	upstream gene variant	2342	ENST00000453558	?	?	1
HCG18	downstream gene variant	3783	ENST00000602591	?	?	1
HCG18	upstream gene variant	1610	ENST00000412685	?	?	2
HCG18	upstream gene variant	2100	ENST00000454129	?	?	1
HCG18	upstream gene variant	2089	ENST00000438412	?	?	1
HCG18	upstream gene variant	4008	ENST00000602516	?	?	2
HCG18	upstream gene variant	2113	ENST00000602290	?	?	1
HCG18	upstream gene variant	1579	ENST00000413358	?	?	2
HCG18	upstream gene variant	2129	ENST00000449544	?	?	1
HCG18	upstream gene variant	2072	ENST00000602550	?	?	1
HCG18	upstream gene variant	1326	ENST00000426882	?	?	2
HCG18	upstream gene variant	2339	ENST00000602498	?	?	1
HCG18	upstream gene variant	2142	ENST00000444126	?	?	1
HCG18	upstream gene variant	2090	ENST00000454269	?	?	1
HCG19P	downstream gene variant, upstream gene variant	151	ENST00000448756	?	?	14
HCG20	downstream gene variant	1545	ENST00000439406	?	?	1
HCG20	upstream gene variant, downstream gene variant	1545	ENST00000422944	?	?	2
HCG4P3	downstream gene variant	2938	ENST00000458060	?	?	1
HLA-J	upstream gene variant	3413	ENST00000495278	?	?	1
HLA-J	upstream gene variant	3404	ENST00000462773	?	?	1
HLA-J	upstream gene variant	3423	ENST00000494367	?	?	1
HLA-J	upstream gene variant	3400	ENST00000469281	?	?	1
HLA-L	upstream gene variant	2391	ENST00000482052	?	?	1
HLA-L	upstream gene variant, downstream gene variant	1288	ENST00000420110	?	?	2
HLA-L	upstream gene variant	2404	ENST00000463348	?	?	1
HLA-N	upstream gene variant, downstream gene variant	39	ENST00000437516	?	?	3
IER3	downstream gene variant, upstream gene variant	1619	ENST00000376377	?	ENSP00000365557	4
IER3	downstream gene variant, upstream gene variant	1619	ENST00000259874	NM_003897.3	ENSP00000259874	4
LINC00243	downstream gene variant	4859	ENST00000419357	?	?	1
LINC00243	downstream gene variant	75	ENST00000399196	?	?	1
MDC1	upstream gene variant	3623	ENST00000416571	?	ENSP00000400979	1
MDC1	upstream gene variant	3351	ENST00000425072	?	ENSP00000396989	1
MDC1	upstream gene variant	2909	ENST00000376406	NM_014641.2	ENSP00000365588	1
MDC1	upstream gene variant	3723	ENST00000613547	?	ENSP00000483601	1
MDC1	upstream gene variant	3677	ENST00000422266	?	ENSP00000411310	1
MICC	upstream gene variant	896	ENST00000445710	?	?	2

MRPS18B	downstream gene variant	2249	ENST00000472229	?	?	1
MRPS18B	downstream gene variant	2249	ENST00000472267	?	?	1
MRPS18B	downstream gene variant	2249	ENST00000259873	NM_014046.3	ENSP00000259873	1
NRM	downstream gene variant	3043	ENST00000444096	?	ENSP00000397892	1
NRM	downstream gene variant	3043	ENST00000259953	NM_007243.2	ENSP00000259953	1
NRM	downstream gene variant	3043	ENST00000376420	NM_001270709.1	ENSP00000365602	1
NRM	downstream gene variant	3043	ENST00000470733	?	?	1
NRM	downstream gene variant	3043	ENST00000462857	?	?	1
NRM	downstream gene variant	3046	ENST00000376421	NM_001270707.1	ENSP00000365603	1
NRM	downstream gene variant	3043	ENST00000495946	?	?	1
NRM	downstream gene variant	3043	ENST00000482141	?	?	1
NRM	downstream gene variant	3043	ENST00000474864	?	?	1
PAIP1P1	downstream gene variant	1212	ENST00000446875	?	?	1
PPP1R10	downstream gene variant	1770	ENST00000376511	NM_002714.3	ENSP00000365694	1
PPP1R10	downstream gene variant	4943	ENST00000496955	?	?	1
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376772	NM_021959.2	ENSP00000365963	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376758	?	ENSP00000365949	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376763	?	ENSP00000365954	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376765	?	ENSP00000365956	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376769	?	ENSP00000365960	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376773	?	ENSP00000365964	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000399199	NM_001134870.1	ENSP00000382150	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000488324	?	?	2
PPP1R18	downstream gene variant, upstream gene variant	31	ENST00000615527	?	ENSP00000480270	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000274853	NM_133471.3	ENSP00000274853	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000467662	?	?	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000615892	?	ENSP00000482578	3
PTMAP1	upstream gene variant, downstream gene variant	1844	ENST00000455552	?	?	2
RN7SL353P	upstream gene variant, downstream gene variant	1557	ENST00000579902	?	?	5
RPP21	downstream gene variant	3693	ENST00000489124	?	?	2
RPP21	downstream gene variant	3693	ENST00000436442	NM_001199121.1	ENSP00000397778	2
RPP21	downstream gene variant	3693	ENST00000428040	?	ENSP00000394320	2
RPP21	downstream gene variant	3693	ENST00000498414	?	?	2
RPP21	downstream gene variant	3693	ENST00000433076	NM_001199120.1	ENSP00000409799	2
RPP21	downstream gene variant	3666	ENST00000491477	?	?	2
RPP21	downstream gene variant	4057	ENST00000466327	?	?	2
RPP21	downstream gene variant	3693	ENST00000442966	NM_024839.2	ENSP00000403833	2

RPP21	downstream gene variant	3693	ENST00000473266	?	?	2
SUCLA2P1	downstream gene variant	3275	ENST00000425839	?	?	1
TRIM10	upstream gene variant	238	ENST00000376704	NM_052828.2	ENSP00000365894	1
TRIM10	upstream gene variant	238	ENST00000449742	NM_006778.3	ENSP00000397073	1
TRIM15	upstream gene variant	2034	ENST00000619857	?	ENSP00000484001	2
TRIM15	upstream gene variant	703	ENST00000477944	?	?	1
TRIM15	upstream gene variant	2044	ENST00000376694	NM_033229.2	ENSP00000365884	2
TRIM15	upstream gene variant	2513	ENST00000376688	?	ENSP00000365878	2
TRIM26	upstream gene variant	996	ENST00000437089	?	ENSP00000395491	1
TRIM26	downstream gene variant	4732	ENST00000418026	?	ENSP00000387530	1
TRIM26	downstream gene variant	2777	ENST00000416596	?	ENSP00000413673	1
TRIM26	downstream gene variant	4979	ENST00000434785	?	ENSP00000400920	1
TRIM26	upstream gene variant	3365	ENST00000480999	?	?	1
TRIM26BP	upstream gene variant	64	ENST00000427723	?	?	2
TRIM31	upstream gene variant	3745	ENST00000485864	?	?	1
TRIM31	upstream gene variant	2825	ENST00000376734	NM_007028.3	ENSP00000365924	1
TRIM31	downstream gene variant, upstream gene variant	3382	ENST00000493404	?	?	2
TRIM31	downstream gene variant, upstream gene variant	910	ENST00000480808	?	?	2
TRIM31	upstream gene variant	2396	ENST00000484583	?	?	1
TRIM31	upstream gene variant	2179	ENST00000471695	?	?	1
TRIM31-AS1	downstream gene variant	1207	ENST00000440874	?	?	1
TRIM39	upstream gene variant	1053	ENST00000420746	?	ENSP00000388224	1
TRIM39	downstream gene variant	1717	ENST00000458516	?	ENSP00000405928	1
TRIM39	downstream gene variant	1973	ENST00000428404	?	ENSP00000405498	1
TRIM39	downstream gene variant	1929	ENST00000440271	?	ENSP00000394768	1
TRIM39	upstream gene variant	413	ENST00000396547	?	ENSP00000379796	1
TRIM39	downstream gene variant	2006	ENST00000428555	?	ENSP00000397952	1
TRIM39-RPP21	upstream gene variant, downstream gene variant	431	ENST00000623385	?	ENSP00000485378	3
TRIM39-RPP21	upstream gene variant, downstream gene variant	835	ENST00000513556	NM_001199119.1	ENSP00000424048	3
TUBB	upstream gene variant	869	ENST00000396389	?	ENSP00000379672	1
TUBB	upstream gene variant	776	ENST00000330914	NM_001293215.1	ENSP00000365578	1
TUBB	upstream gene variant	901	ENST00000396384	?	ENSP00000379668	1
UBQLN1P1	downstream gene variant, upstream gene variant	152	ENST00000441056	?	?	16
XXbac-BPG252P9.10	upstream gene variant	1349	ENST00000607333	?	?	3
XXbac-BPG252P9.9	downstream gene variant	2307	ENST00000607476	?	?	1
XXbac-BPG283O16.9	downstream gene variant	1415	ENST00000624252	?	?	1
Y_RNA	downstream gene variant, upstream gene variant	2170	ENST00000365118	?	?	3

ZNRD1-AS1	upstream gene variant	3566	ENST00000431012	?	?	1
ZNRD1-AS1	upstream gene variant	3601	ENST00000376797	?	?	1
ZNRD1-AS1	upstream gene variant	3817	ENST00000425604	?	?	1
ZNRD1-AS1	upstream gene variant	3556	ENST00000421692	?	?	1
ZNRD1-AS1	upstream gene variant	3766	ENST00000452229	?	?	1
ZNRD1-AS1	downstream gene variant, upstream gene variant	1223	ENST00000437417	?	?	2
ZNRD1-AS1	upstream gene variant	3602	ENST00000448093	?	?	1
ZNRD1-AS1	upstream gene variant	3561	ENST00000420251	?	?	1
ZNRD1-AS1_3	downstream gene variant	3562	ENST00000611686	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
GNL1	ENST00000376621	NM_005275.3	ENSP00000365806	A	gcG/gcA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABCF1	ENST00000326195	NM_001025091.1	ENSP00000313603	1
ABCF1	ENST00000468958	?	ENSP00000440893	1
ABCF1	ENST00000441867	?	ENSP00000405512	1
ABCF1	ENST00000376545	NM_001090.2	ENSP00000365728	1
ATAT1	ENST00000376485	?	ENSP00000365668	3
ATAT1	ENST00000493388	?	?	2
ATAT1	ENST00000318999	?	ENSP00000324222	3
ATAT1	ENST00000468713	?	?	3
ATAT1	ENST00000376483	?	ENSP00000365666	3
ATAT1	ENST00000330083	NM_001031722.2	ENSP00000327832	3
ATAT1	ENST00000319027	NM_001254952.1	ENSP00000324459	3
ATAT1	ENST00000471782	?	?	3
ATAT1	ENST00000329992	NM_024909.2	ENSP00000332374	3
ATAT1	ENST00000479562	?	?	1
ATAT1	ENST00000462304	?	?	3
C6orf136	ENST00000376473	NM_001109938.2	ENSP00000365656	1
C6orf136	ENST00000465699	?	ENSP00000418154	1
C6orf136	ENST00000293604	NM_001161376.1	ENSP00000293604	1
C6orf136	ENST00000488383	?	ENSP00000419968	1
C6orf136	ENST00000376471	NM_145029.3	ENSP00000365654	1
C6orf136	ENST00000446773	?	ENSP00000415810	1
C6orf136	ENST00000487873	?	ENSP00000418616	1
C6orf136	ENST00000463794	?	ENSP00000418223	1

C6orf136	ENST00000484551	?	ENSP00000418599	1
FLOT1	ENST00000418160	?	ENSP00000404300	2
FLOT1	ENST00000416018	?	ENSP00000412058	2
FLOT1	ENST00000470643	?	?	2
FLOT1	ENST00000484168	?	?	2
FLOT1	ENST00000476729	?	?	2
FLOT1	ENST00000413165	?	ENSP00000395333	4
FLOT1	ENST00000376389	NM_005803.2	ENSP00000365569	4
FLOT1	ENST00000454845	?	ENSP00000391341	2
FLOT1	ENST00000438162	?	ENSP00000400615	4
FLOT1	ENST00000445853	?	ENSP00000398834	2
FLOT1	ENST00000487376	?	?	2
HCG17	ENST00000453558	?	?	11
HCG18	ENST00000454129	?	?	4
HCG18	ENST00000444126	?	?	4
HCG18	ENST00000602550	?	?	4
HCG18	ENST00000438412	?	?	4
HCG18	ENST00000412685	?	?	4
HCG18	ENST00000454269	?	?	4
HCG18	ENST00000426882	?	?	4
HCG18	ENST00000449544	?	?	4
HCG18	ENST00000413358	?	?	4
HCG18	ENST00000602498	?	?	4
HCG18	ENST00000602290	?	?	4
HCG20	ENST00000439406	?	?	2
HLA-L	ENST00000463348	?	?	1
HLA-L	ENST00000482052	?	?	3
LINC00243	ENST00000419357	?	?	4
LINC00243	ENST00000399196	?	?	2
MICC	ENST00000445710	?	?	1
NRM	ENST00000474864	?	?	1
NRM	ENST00000470733	?	?	1
NRM	ENST00000444096	?	ENSP00000397892	1
NRM	ENST00000376421	NM_001270707.1	ENSP00000365603	1
NRM	ENST00000482141	?	?	1
NRM	ENST00000495946	?	?	1
NRM	ENST00000259953	NM_007243.2	ENSP00000259953	1
NRM	ENST00000462857	?	?	1
NRM	ENST00000376420	NM_001270709.1	ENSP00000365602	1
PPP1R18	ENST00000274853	NM_133471.3	ENSP00000274853	1
PPP1R18	ENST00000467662	?	?	2
PPP1R18	ENST00000399199	NM_001134870.1	ENSP00000382150	1

PPP1R18	ENST00000615892	?	ENSP00000482578	1
PPP1R18	ENST00000615527	?	ENSP00000480270	1
PPP1R18	ENST00000488324	?	?	2
TRIM10	ENST00000449742	NM_006778.3	ENSP00000397073	2
TRIM10	ENST00000376704	NM_052828.2	ENSP00000365894	2
TRIM15	ENST00000376694	NM_033229.2	ENSP00000365884	1
TRIM15	ENST00000619857	?	ENSP00000484001	1
TRIM15	ENST00000433744	?	ENSP00000398285	1
TRIM15	ENST00000376688	?	ENSP00000365878	1
TRIM26	ENST00000418026	?	ENSP00000387530	2
TRIM26	ENST00000434785	?	ENSP00000400920	2
TRIM26	ENST00000487829	?	?	2
TRIM26	ENST00000480999	?	?	1
TRIM26	ENST00000453195	NM_001242783.1	ENSP00000391879	4
TRIM26	ENST00000437089	?	ENSP00000395491	3
TRIM26	ENST00000454678	NM_003449.4	ENSP00000410446	4
TRIM26	ENST00000416596	?	ENSP00000413673	2
TRIM31	ENST00000468264	?	?	1
TRIM31	ENST00000485864	?	?	1
TRIM31	ENST00000376734	NM_007028.3	ENSP00000365924	1
TRIM31-AS1	ENST00000440874	?	?	1
TRIM39	ENST00000458516	?	ENSP00000405928	1
TRIM39	ENST00000396547	?	ENSP00000379796	1
TRIM39	ENST00000396548	?	ENSP00000379797	2
TRIM39	ENST00000428728	?	ENSP00000406019	2
TRIM39	ENST00000428404	?	ENSP00000405498	1
TRIM39	ENST00000428555	?	ENSP00000397952	1
TRIM39	ENST00000396551	?	ENSP00000379800	2
TRIM39	ENST00000376659	NM_172016.2	ENSP00000365847	2
TRIM39	ENST00000420746	?	ENSP00000388224	1
TRIM39	ENST00000440271	?	ENSP00000394768	1
TRIM39	ENST00000376656	NM_021253.3	ENSP00000365844	2
TRIM39-RPP21	ENST00000513556	NM_001199119.1	ENSP00000424048	1
TRIM39-RPP21	ENST00000623385	?	ENSP00000485378	1
TRIM40	ENST00000376724	?	ENSP00000365914	1
TRIM40	ENST00000307859	NM_138700.4	ENSP00000308310	1
TRIM40	ENST00000396581	NM_001286633.1	ENSP00000379826	1
TUBB	ENST00000327892	NM_178014.2	ENSP00000339001	1
ZNRD1-AS1	ENST00000425604	?	?	2
ZNRD1-AS1	ENST00000420251	?	?	2
ZNRD1-AS1	ENST00000376797	?	?	2
ZNRD1-AS1	ENST00000448093	?	?	2

ZNRD1-AS1	ENST00000448093	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATAT1	ENST00000318999	?	ENSP00000324222	1
ATAT1	ENST00000376483	?	ENSP00000365666	1
ATAT1	ENST00000319027	NM_001254952.1	ENSP00000324459	1
ATAT1	ENST00000329992	NM_024909.2	ENSP00000332374	1
RNF39	ENST00000376751	NM_170769.2	ENSP00000365942	1
RNF39	ENST00000244360	NM_025236.3	ENSP00000244360	1
TRIM26	ENST00000453195	NM_001242783.1	ENSP00000391879	1
TRIM26	ENST00000437089	?	ENSP00000395491	1
TRIM26	ENST00000454678	NM_003449.4	ENSP00000410446	1
ZNRD1	ENST00000332435	NM_170783.3	ENSP00000331111	1
ZNRD1	ENST00000359374	NM_014596.5	ENSP00000352333	1
ZNRD1	ENST00000376782	NM_001278785.1	ENSP00000365978	1
ZNRD1	ENST00000376785	NM_001278786.1	ENSP00000365981	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FLOT1	ENST00000484168	?	1
FLOT1	ENST00000484693	?	1
GNL1	ENST00000462708	?	1
GNL1	ENST00000464231	?	1
HCG19P	ENST00000448756	?	2
PAIP1P1	ENST00000446875	?	1
TRIM26	ENST00000480999	?	2
UBQLN1P1	ENST00000441056	?	1
XXbac-BPG283O16.9	ENST00000624252	?	1
ZNRD1	ENST00000463141	?	1
ZNRD1	ENST00000471008	?	1
ZNRD1-AS1	ENST00000444051	?	1
ZNRD1-AS1	ENST00000376797	?	2

Locus 181 (rs17286411)

SNiPAcard

Block annotations

Block info

genomic range	chr16:71,968,701-71,972,060
block size	3,360 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.299 [-0.849 – 0.251]	gene(s) hit or close-by	IST1, PKD1L3
phastCons	μ = 0.032 [0 – 0.063]	eQTL gene(s)	AP1G1, HP, PHLPP2
GERP++	μ = -0.150 [-0.457 – 0.158]	potentially regulated gene(s)	–
CADD score	μ = 5.304 [4.722 – 5.886]	disease gene(s)	HP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
gamma-glutamyltyrosine	7.51×10^{-6}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
HP	ANHAPTOGLOBINEMIA	OMIM	MIM:614081

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PHLPP2	?	ENSG00000040199	testis	2.75×10^{-6} (p-value)	GTEx Portal V6	2
AP1G1	?	ENSG00000166747	aorta	4.33×10^{-6} (p-value)	GTEx Portal V6	1
HP	ENST00000570083	ILMN_1812433	monocyte	2.57×10^{-16} (p-value)	Zeller et al.	1
HP	ENST00000567185					
HP	ENST00000566821					
HP	ENST00000613898					
HP	ENST00000565574					
HP	ENST00000228226					
HP	ENST00000355906					
HP	ENST00000567612					
HP	ENST00000398131					
HP	ENST00000564499					
HP	ENST00000357763					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IST1	downstream gene variant	4159	ENST00000535424	NM_001270976.1	ENSP00000438399	1

IST1	downstream gene variant	4159	ENST00000606369	NM_001270979.1, NM_001270978.1	ENSP00000475853	1
IST1	downstream gene variant	3599	ENST00000378799	NM_001270975.1	ENSP00000368076	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PKD1L3	ENST00000620267	NM_181536.1	ENSP00000480090	2

Locus 182 (rs17220241)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,809,582-32,861,183
block size	51,602 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.257 [-2.534 – 0.965]	gene(s) hit or close-by	HLA-Z, PSMB8, PSMB8-AS1, PSMB9, TAP1, TAP2, XXbac-BPG246D15.9
phastCons	μ = 0.032 [0 – 0.164]	eQTL gene(s)	–
GERP++	μ = -0.736 [-4.68 – 3.04]	potentially regulated gene(s)	COL11A2, COL11A2, COL11A2, COL11A2, COL11A2, COL11A2, COL11A2, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z
CADD score	μ = 4.633 [0.832 – 13.03]	disease gene(s)	TAP2, TAP1, PSMB8, , , , , , COL11A2, , , , HLA-DPB1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TAP2	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571
TAP1	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571
PSMB8	AUTOINFLAMMATION, LIPODYSTROPHY, AND DERMATOSIS SYNDROME	OMIM	MIM:256040
	WEISSENBACHER-ZWEYMULLER SYNDROME	OMIM	MIM:277610
	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150
	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706
	FIBROCHONDROGENESIS 2	OMIM	MIM:614524
	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868
	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840
	WEISSENBACHER-ZWEYMULLER SYNDROME	OMIM	MIM:277610
	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150
	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706
	FIBROCHONDROGENESIS 2	OMIM	MIM:614524
	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868
	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840
	WEISSENBACHER-ZWEYMULLER SYNDROME	OMIM	MIM:277610
	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150
	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706
	FIBROCHONDROGENESIS 2	OMIM	MIM:614524
	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868
	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840
	WEISSENBACHER-ZWEYMULLER SYNDROME	OMIM	MIM:277610
	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150
	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706
	FIBROCHONDROGENESIS 2	OMIM	MIM:614524
	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868
	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840
COL11A2	WEISSENBACHER-ZWEYMULLER SYNDROME	OMIM	MIM:277610
COL11A2	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150

COL11A2	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706	OMIM
COL11A2	FIBROCHONDROGENESIS 2	OMIM	MIM:614524	OMIM
COL11A2	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868	OMIM
COL11A2	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840	OMIM
PSMB8	Nakajo syndrome	DECIPHER	MIM:256040	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
COL11A2	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
COL11A2	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
COL11A2	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
COL11A2	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
COL11A2	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
TAP2	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592	orphanet
TAP1	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592	orphanet
PSMB8	CANDLE syndrome	OrphaNet	OrphaNet:325004	orphanet
PSMB8	JMP syndrome	OrphaNet	OrphaNet:324999	orphanet
PSMB8	Nakajo-Nishimura syndrome	OrphaNet	OrphaNet:2615	orphanet
HLA-DPB1	Chronic berylliosis	OrphaNet	OrphaNet:133	orphanet
HLA-DPB1	Granulomatosis with polyangiitis	OrphaNet	OrphaNet:900	orphanet
COL11A2	Autosomal recessive nonsyndromic sensorineural deafness type DFNB	OrphaNet	OrphaNet:90636	orphanet
COL11A2	WEISSENBACHER-ZWEYMULLER SYNDROME	OrphaNet	OrphaNet:3450	orphanet
COL11A2	Autosomal dominant nonsyndromic sensorineural deafness type DFNA	OrphaNet	OrphaNet:90635	orphanet
COL11A2	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OrphaNet	OrphaNet:1427	orphanet
COL11A2	Stickler syndrome, type 3	OrphaNet	OrphaNet:166100	orphanet
COL11A2	Fibrochondrogenesis	OrphaNet	OrphaNet:2021	orphanet

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445615	1	ENCP00000048222	HLA-DOB HLA-DOB HLA-DOB HLA-DOB HLA-DOB HLA-DOB
ENCE00000445701	1	ENCP00000048255	HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1
		ENCP00000048262	COL11A2 COL11A2 COL11A2 COL11A2 COL11A2 COL11A2 COL11A2
		ENCP00000048235	HLA-Z HLA-Z HLA-Z HLA-Z HLA-Z HLA-Z HLA-Z HLA-Z

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000025365 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, PolII, Rad21, Nrsf, CTCF, TAF1, H3K4me2, H3K4me3
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	PolII, Max, H3K4me1, Rad21, H3K9ac, Cmyc, H2AZ, Nrsf, CTCFL, H3K4me2, CTCF, H3K27me3, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me3, H3K4me2, DNase1, CTCF, H2AZ
		liver (HepG2)	H3K4me3, H3K27me3, DNase1, Nrsf, PolII, Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF
		blood (GM12878)	Nrsf, PolII, H2AZ, DNase1, Yy1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac, H3K36me3
		lung (IMR90)	CTCF, H3K9ac, DNase1, H3K27ac, H3K4me2, H3K4me3
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, H3K4me3, H3K27me3, DNase1
		skin (NHEK)	H3K4me2, H3K4me3, CTCF, H3K36me3, DNase1, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, CTCF, H3K4me2, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3, CTCF
		cervix (HeLa-S3)	H3K9ac, Nrsf, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me3, H3K9ac, DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, PolII, CTCF, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000025366 (promoter flanking region)	2	embryonic stem cell (H1ESC)	PolII, Rad21, p300, CTCF, Tcf12, TAF1, SP1, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K4me2, CTCF
		blood (GM12878)	H3K36me3
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF

		skin (NHEK)	CTCF
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		A549	CTCF, H3K4me3
ENSR00001703138 (CTCF binding site)	1	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
ENSR00001494626 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3, CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		liver (HepG2)	CTCF, Rad21, H3K27me3
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HLA-Z	upstream gene variant	3010	ENST00000434920	?	?	1
PSMB8-AS1	upstream gene variant, downstream gene variant	2333	ENST00000415067	?	?	2
PSMB8-AS1	upstream gene variant, downstream gene variant	2328	ENST00000413039	?	?	2
PSMB8-AS1	upstream gene variant, downstream gene variant	2303	ENST00000458296	?	?	2
PSMB8-AS1	upstream gene variant, downstream gene variant	2328	ENST00000429600	?	?	2
PSMB8-AS1	upstream gene variant, downstream gene variant	2281	ENST00000453426	?	?	3
PSMB8-AS1	upstream gene variant, downstream gene variant	2329	ENST00000412095	?	?	2
PSMB9	upstream gene variant, downstream gene variant	54	ENST00000467593	?	?	5
PSMB9	upstream gene variant, downstream gene variant	1006	ENST00000395330	?	ENSP00000378739	3
PSMB9	upstream gene variant, downstream gene variant	1691	ENST00000464863	?	?	4
PSMB9	upstream gene variant, downstream gene variant	1006	ENST00000374859	NM_002800.4	ENSP00000363993	4
PSMB9	upstream gene variant, downstream gene variant	339	ENST00000414474	?	ENSP00000394363	4
TAP1	downstream gene variant, upstream gene variant	489	ENST00000354258	NM_000593.5	ENSP00000346206	4
TAP1	downstream gene variant, upstream gene variant	2446	ENST00000487296	?	?	3
TAP1	downstream gene variant, upstream gene variant	534	ENST00000486332	?	?	4
TAP2	upstream gene variant	3035	ENST00000620123	NM_000544.3	ENSP00000481712	1
TAP2	upstream gene variant	3078	ENST00000374899	NM_018833.2	ENSP00000364034	1
TAP2	upstream gene variant	3025	ENST00000374897	NM_001290043.1	ENSP00000364032	1

XXbac-BPG246D15.9	upstream gene variant	2983	ENST00000452392	?	ENSP00000391806 1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PSMB8	ENST00000374881	NM_004159.4	ENSP00000364015	1
PSMB8	ENST00000484003	?	?	1
PSMB8	ENST00000395339	?	ENSP00000378748	1
PSMB8	ENST00000374882	NM_148919.3	ENSP00000364016	1
PSMB9	ENST00000414474	?	ENSP00000394363	4
PSMB9	ENST00000395330	?	ENSP00000378739	5
PSMB9	ENST00000374859	NM_002800.4	ENSP00000363993	3
PSMB9	ENST00000464863	?	?	3
PSMB9	ENST00000467593	?	?	1
TAP1	ENST00000354258	NM_000593.5	ENSP00000346206	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PSMB8	ENST00000490613	?	1
PSMB9	ENST00000464863	?	2

Locus 183 (rs11957503)

SNiPAcard

Block annotations

Block info

genomic range	chr5:35,827,646-35,883,804
block size	56,159 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.527 [-1.238 – 0.295]	gene(s) hit or close-by	CTD-2113L7.1, IL7R
phastCons	μ = 0.001 [0 – 0.005]	eQTL gene(s)	–
GERP++	μ = -0.467 [-2.75 – 1.27]	potentially regulated gene(s)	–
CADD score	μ = 2.893 [0.346 – 5.497]	disease gene(s)	IL7R

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Breast neoplasms	6.57×10-5	dbGaP	pha002853	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
IL7R	T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency	OrphaNet	OrphaNet:169154	orphanet
IL7R	OMENN SYNDROME	OrphaNet	OrphaNet:39041	orphanet

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001278142 (promoter flanking region)	4	NHLF	DNase1, H3K27ac
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
		cervix (HeLa-S3)	Cjun, Jund, H3K27ac, H3K4me1, Cfos, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Cjun
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-2113L7.1	upstream gene variant	526	ENST00000510433	?	?	3
IL7R	downstream gene variant	3471	ENST00000303115	NM_002185.3	ENSP00000306157	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL7R	ENST00000303115	NM_002185.3	ENSP00000306157	1
IL7R	ENST00000514217	?	ENSP00000427688	1
IL7R	ENST00000511031	?	?	1
IL7R	ENST00000511982	?	ENSP00000425309	1
IL7R	ENST00000506850	?	ENSP00000421207	1

Locus 184 (rs289828)

SNiPAcard

Block annotations

Block info

genomic range	chr2:152,085,984-152,186,147
block size	100,164 bp
variant count	40 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.458 [-3.25 – 1.808]	gene(s) hit or close-by	NMI, RBM43
phastCons	μ = 0.042 [0 – 0.765]	eQTL gene(s)	NMI, RBM43, RIF1, TNFAIP6
GERP++	μ = -0.615 [-5.11 – 2.17]	potentially regulated gene(s)	–
CADD score	μ = 3.579 [0.007 – 10.95]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RIF1	ENST00000453091	ILMN_2149053	blood	3.72×10^{-5} (p-value)	Westra et al.	13
RIF1	ENST00000243326					
NMI	?	ENSG00000123609	testis	8.36×10^{-6} (p-value)	GTEx Portal V6	14
RIF1	?	ENSG00000080345	tibial nerve	5.10×10^{-6} (p-value)	GTEx Portal V6	23
NMI	?	ENSG00000123609	transformed fibroblasts	4.50×10^{-12} (p-value)	GTEx Portal V6	36
TNFAIP6	?	ENSG00000123610	transformed fibroblasts	1.07×10^{-11} (p-value)	GTEx Portal V6	40
RIF1	?	ENSG00000080345	transformed fibroblasts	1.66×10^{-5} (p-value)	GTEx Portal V6	1
RBM43	?	ENSG00000184898	blood	1.35×10^{-9} (p-value)	GTEx Portal V6	40
TNFAIP6	?	ENSG00000123610	blood	1.06×10^{-43} (p-value)	GTEx Portal V6	40
NMI	?	ENSG00000123609	skeletal muscle	6.88×10^{-7} (p-value)	GTEx Portal V6	15
NMI	?	ENSG00000123609	thyroid	7.93×10^{-7} (p-value)	GTEx Portal V6	18
NMI	?	ENSG00000123609	spleen	4.32×10^{-6} (p-value)	GTEx Portal V6	14
NMI	?	ENSG00000123609	lung	1.74×10^{-6} (p-value)	GTEx Portal V6	16
?	?	ILMN_1785732	monocyte	2.96×10^{-64} (p-value)	Zeller et al.	5
RBM43	ENST00000331426	ILMN_1858599	monocyte	1.70×10^{-24} (p-value)	Zeller et al.	5
?	?	ILMN_1785732	monocyte	2.00×10^{-12} (p-value)	Fairfax et al.	3
NMI	ENST00000243346	ILMN_1739541	b-cell	4.61×10^{-4} (p-value)	Fairfax et al.	1
RBM43	ENST00000331426	ILMN_3251415	b-cell	3.44×10^{-5} (p-value)	Fairfax et al.	2
RBM43	ENST00000331426	ILMN_1858599	b-cell	4.64×10^{-14} (p-value)	Fairfax et al.	1
RIF1	?	ENSG00000080345	sun exposed skin	5.51×10^{-7} (p-value)	GTEx Portal V6	7
RIF1	?	ENSG00000080345	unexposed skin	1.57×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000600259 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00001547512 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1
		Osteobl	H3K27ac
		A549	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00001547513 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K4me2, TAF1, PolII, Rad21, H3K9ac
		HSMMtube	H3K9ac, H3K4me2, H2AZ, DNase1
		blood (K562)	H2AZ, PolII, H3K4me2, DNase1, H3K4me3, Cmyc, H3K9ac, Brg1, H3K27ac, Max
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		liver (HepG2)	H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		blood (GM12878)	Tr4, PolII, Srf, H2AZ, DNase1, Yy1, Gabp, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K4me3, H3K9ac, H3K4me2, H3K27ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K9ac, H3K27ac, H3K4me3
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, DNase1
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, H3K27ac
ENSR00001547517 (promoter flanking region)	1	liver (HepG2)	H3K27me3
		HSMMtube	H3K27ac, H2AZ, DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H2AZ, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NMI	downstream gene variant, upstream gene variant	101	ENST00000243346	NM_004688.2	ENSP00000243346	2
NMI	downstream gene variant, upstream gene variant	295	ENST00000491771	?	?	5
NMI	downstream gene variant, upstream gene variant	14	ENST00000477072	?	?	11
NMI	downstream gene variant, upstream gene variant	31	ENST00000414946	?	ENSP00000387373	7
RBM43	downstream gene variant	1099	ENST00000331426	NM_198557.2	ENSP00000331211	1
RBM43	downstream gene variant	876	ENST00000409092	?	ENSP00000386440	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
NMI	ENST00000243346	NM_004688.2	ENSP00000243346	Q	caA/caG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NMI	ENST00000243346	NM_004688.2	ENSP00000243346	23
NMI	ENST00000491771	?	?	19
NMI	ENST00000414946	?	ENSP00000387373	13
NMI	ENST00000477072	?	?	9
RBM43	ENST00000331426	NM_198557.2	ENSP00000331211	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RBM43	ENST00000331426	NM_198557.2	ENSP00000331211	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NMI	ENST00000491771	?	1

Locus 185 (rs35030851)

SNiPAcard

Block annotations

Block info

genomic range	chr9:27,196,743-27,214,579
block size	17,837 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.279 [-1.679 – 1.68]	gene(s) hit or close-by	RNA5SP280, RP11-179D22.1, TEK
phastCons	μ = 0.412 [0 – 0.995]	eQTL gene(s)	–
GERP++	μ = -0.588 [-6.48 – 4.64]	potentially regulated gene(s)	TEK
CADD score	μ = 7.173 [0.031 – 17.78]	disease gene(s)	TEK

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TEK	VENOUS MALFORMATIONS, MULTIPLE CUTANEOUS AND MUCOSAL	DECIPHER	MIM:600195
TEK	VENOUS MALFORMATIONS, MULTIPLE CUTANEOUS AND MUCOSAL	OMIM	MIM:600195
TEK	Mucocutaneous venous malformations	OrphaNet	OrphaNet:2451

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
TEK	missense variant	ENST00000519080	?	ENSP00000428337	L/V	Ttg/Gtg	?	?	1
TEK	missense variant	ENST00000406359	NM_001290077.1	ENSP00000383977	L/V	Ttg/Gtg	?	?	1
TEK	missense variant	ENST00000519097	?	ENSP00000430686	L/V	Ttg/Gtg	?	?	1
TEK	missense variant	ENST00000380036	NM_000459.3	ENSP00000369375	L/V	Ttg/Gtg	?	?	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000865827	1	ENST00000406359, ENST00000519097, ENST00000519080, ENST00000380036	TEK

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RNA5SP280	upstream gene variant, downstream gene variant	799	ENST00000411230	?	?	4
RP11-179D22.1	upstream gene variant, downstream gene variant	856	ENST00000422804	?	?	5
TEK	downstream gene variant	119	ENST00000519080	?	ENSP00000428337	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TEK	ENST00000380036	NM_000459.3	ENSP00000369375	Q	caA/caG	1
TEK	ENST00000406359	NM_001290077.1	ENSP00000383977	Q	caA/caG	1
TEK	ENST00000519097	?	ENSP00000430686	Q	caA/caG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TEK	ENST00000380036	NM_000459.3	ENSP00000369375	7
TEK	ENST00000615002	?	ENSP00000480251	7
TEK	ENST00000406359	NM_001290077.1	ENSP00000383977	7
TEK	ENST00000519097	?	ENSP00000430686	7
TEK	ENST00000519080	?	ENSP00000428337	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TEK	ENST00000615002	?	ENSP00000480251	2

Locus 186 (rs34231037)

SNiPAcard

Block annotations

Block info

genomic range	chr4:55,972,946-55,972,946
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	6.349	gene(s) hit or close-by	KDR
phastCons	1	eQTL gene(s)	–
GERP++	5.67	potentially regulated gene(s)	–
CADD score	25.4	disease gene(s)	KDR

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	not provided	ClinVar	RCV000121295.1	1
Hemangioma, capillary infantile, susceptibility to	risk factor	ClinVar	RCV000013111.1	1
?	HGMD curated	HGMD	CM087498	1
HEMANGIOMA, CAPILLARY INFANTILE, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:191306	1

Disease gene annotation

gene	trait	source DB	source entry/link
KDR	HEMANGIOMA, CAPILLARY INFANTILE	OMIM	MIM:602089
KDR	Familial capillary hemangioma	OrphaNet	OrphaNet:91415

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
KDR	missense variant	ENST00000263923	NM_002253.2	ENSP00000263923	R/C	Cgt/Tgt	?	?	1

Putative effect on transcript

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
KDR	ENST00000512566	?	1

Locus 187 (rs11548618)

SNiPAcard

Block annotations

Block info

genomic range	chr4:76,943,947-76,943,947
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	4.616	gene(s) hit or close-by	ART3, CXCL10
phastCons	0.999	eQTL gene(s)	–
GERP++	4.93	potentially regulated gene(s)	–
CADD score	14.66	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CXCL10	missense variant	ENST00000306602	NM_001565.3	ENSP00000305651	C/R	Tgc/Cgc	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ART3	ENST00000510669	?	?	1
ART3	ENST00000513353	?	ENSP00000421345	1
ART3	ENST00000504914	?	ENSP00000421431	1
ART3	ENST00000341029	NM_001130017.2	ENSP00000343843	1
ART3	ENST00000513122	?	ENSP00000422287	1

Locus 188 (rs10858881)

SNiPAcard

Block annotations

Block info

genomic range	chr12:89,825,925-89,938,590
block size	112,666 bp
variant count	76 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.255 [-2.445 – 1.215]	gene(s) hit or close-by	CENPCP1, GALNT4, POC1B, POC1B-GALNT4, RP11-734K2.3, RP11-734K2.4
phastCons	μ = 0.052 [0 – 0.745]	eQTL gene(s)	ATP2B1, GALNT4, POC1B, POC1B-GALNT4, RP11-981P6.1
GERP++	μ = -0.521 [-5.92 – 3.23]	potentially regulated gene(s)	–
CADD score	μ = 4.052 [0.12 – 13.8]	disease gene(s)	POC1B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
POC1B	CONE-ROD DYSTROPHY 20	OMIM	MIM:615973

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-981P6.1	?	ENSG00000258302	muscularis mucosae	2.36×10-7 (p-value)	GTEx Portal V6	73
RP11-981P6.1	?	ENSG00000258302	spleen	5.53×10-9 (p-value)	GTEx Portal V6	75
POC1B	?	ENSG00000139323	tibial nerve	1.80×10-12 (p-value)	GTEx Portal V6	76
POC1B	?	ENSG00000139323	transformed fibroblasts	1.33×10-6 (p-value)	GTEx Portal V6	73
POC1B	?	ENSG00000139323	tibial artery	1.41×10-6 (p-value)	GTEx Portal V6	73
RP11-981P6.1	?	ENSG00000258302	tibial artery	7.99×10-8 (p-value)	GTEx Portal V6	75
RP11-981P6.1	?	ENSG00000258302	thyroid	2.97×10-25 (p-value)	GTEx Portal V6	76
RP11-981P6.1	?	ENSG00000258302	pituitary	5.61×10-7 (p-value)	GTEx Portal V6	66
RP11-981P6.1	?	ENSG00000258302	adrenal gland	1.17×10-9 (p-value)	GTEx Portal V6	75
RP11-981P6.1	?	ENSG00000258302	sun exposed skin	8.82×10-7 (p-value)	GTEx Portal V6	74
POC1B	?	ENSG00000139323	aorta	3.08×10-6 (p-value)	GTEx Portal V6	31
POC1B	?	ENSG00000139323	subcutaneous adipocytes	2.15×10-10 (p-value)	GTEx Portal V6	76
GALNT4	ENST00000529983	ILMN_1739297	skin	4.74×10-8 (p-value)	MuTHER consortium	33
POC1B-GALNT4	ENST00000547474		blood	5.22×10-6 (p-value)	MuTHER consortium	33
POC1B-GALNT4	ENST00000548729					
ATP2B1	ENST00000428670	ILMN_1824297	monocyte	6.85×10-15 (p-value)	Zeller et al.	4

ATP2B1	ENST00000359142					
ATP2B1	ENST00000261173					
RP11-981P6.1	?	ENSG00000258302	lung	7.14×10-6 (p-value)	GTEx Portal V6	61
RP11-981P6.1	?	ENSG00000258302	transverse colon	3.61×10-6 (p-value)	GTEx Portal V6	41
RP11-981P6.1	?	ENSG00000258302	cerebellum	3.05×10-6 (p-value)	GTEx Portal V6	60
RP11-981P6.1	?	ENSG00000258302	stomach	3.58×10-7 (p-value)	GTEx Portal V6	67
POC1B	?	ENSG00000139323	breast	5.43×10-6 (p-value)	GTEx Portal V6	1
POC1B-GALNT4	ENST00000547474	ILMN_1739297	monocyte	3.17×10-8 (p-value)	Fairfax et al.	6
GALNT4	ENST00000529983		b-cell	1.62×10-5 (p-value)	Fairfax et al.	6
POC1B-GALNT4	ENST00000548729					
POC1B	?	ENSG00000139323	blood	9.10×10-6 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
GALNT4	ENST00000529983	ILMN_1739297	chr12	monocyte	4.54×10-12 (p-value)	Zeller et al.	1
POC1B-GALNT4	ENST00000547474						
POC1B-GALNT4	ENST00000548729						

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000434328 (enhancer)	3	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Cjun
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		A549	H3K36me3
ENSR00000434333 (promoter flanking region)	2	embryonic stem cell (H1ESC)	USF1, DNase1
		HSMMtube	H3K27ac, H3K4me2, DNase1, H3K27me3, H3K9ac
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1, H3K27ac
		nervous (NH-A)	DNase1
		NHLF	DNase1, H3K27ac
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, H3K4me1, H3K4me2
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1, H3K4me1, H3K27ac, PolII
		A549	H3K36me3
ENSR00001613708 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00001613711 (enhancer)	1	liver (HepG2)	H3K4me1
ENSR00001613712 (CTCF binding site)	2	monocytes (Monocytes-CD14+)	H3K36me3
		blood (DND-41)	H3K36me3

ENSR00001613713 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
ENSR00000434347 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K27ac, H3K36me3, H3K27me3, PolII, Rad21, Sin3Ak20, H3K9ac, CTCF, Yy1, TAF1, H3K4me2, SP1, Srf, H3K4me3
		HSMMtube	DNase1, H2AZ, H3K4me3, H3K4me2, CTCF
		blood (K562)	H3K27ac, Max, H3K9ac, H2AZ, TAF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K4me3, CTCF, H2AZ
		liver (HepG2)	H3K79me2, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, DNase1, PolII, Gabp, CTCF, Srf
		lung (IMR90)	H2BK120ac, H3K9ac, CTCF, H3K4ac, H3K4me3, H3K36me3, DNase1, H4K8ac, H3K27ac, H3K18ac, H3K4me2, H4K5ac
		blood (GM12878)	DNase1, Gabp, ELF1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac, H2AZ, Srf, PolII, Tr4
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K4me2, H3K4me3, CTCF, H3K36me3, DNase1, H3K9ac
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	CTCF, H3K36me3, H3K4me2, H3K4me3, H2AZ, H3K27ac
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K4me2, H3K9ac, H3K27ac, PolII, H3K4me3, DNase1, H3K36me3, CTCF, Gabp
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, H3K9ac, PolII, CTCF, DNase1
		A549	H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1, H3K36me3, H3K27ac
ENSR00001465661 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF
ENSR00000245284 (TF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	Gata2, DNase1, Egr1, PolII
ENSR00000434348 (promoter flanking region)	1	HSMMtube	DNase1
		A549	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CENPCP1	upstream gene variant, downstream gene variant	450	ENST00000533085	?	?	6
GALNT4	downstream gene variant, upstream gene variant	94	ENST00000529983	NM_003774.4	ENSP00000436604	9
POC1B	downstream gene variant	2282	ENST00000549304	?	?	1
POC1B	downstream gene variant, upstream gene variant	387	ENST00000547274	?	ENSP00000449648	11
POC1B	downstream gene variant	131	ENST00000551819	?	?	2
POC1B	downstream gene variant, upstream gene variant	414	ENST00000539190	?	?	9
POC1B	upstream gene variant	1992	ENST00000546740	?	?	2
POC1B	downstream gene variant, upstream gene variant	407	ENST00000546830	?	ENSP00000449256	11
POC1B	downstream gene variant, upstream gene variant	226	ENST00000552563	?	?	10

	variant					
POC1B	upstream gene variant	383	ENST00000313546	NM_172240.2	ENSP00000323302	8
POC1B	upstream gene variant	2688	ENST00000549591	?	?	1
POC1B	upstream gene variant	407	ENST00000547496	?	ENSP00000447437	8
POC1B	upstream gene variant	586	ENST00000549504	?	ENSP00000450118	8
POC1B	upstream gene variant	228	ENST00000549035	NM_001199777.1	ENSP00000447916	8
POC1B	upstream gene variant	176	ENST00000393179	?	ENSP00000376877	8
POC1B	upstream gene variant	410	ENST00000548715	?	ENSP00000449945	8
POC1B-GALNT4	downstream gene variant, upstream gene variant	145	ENST00000547474	?	ENSP00000447754	10
POC1B-GALNT4	downstream gene variant, upstream gene variant	209	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	9
RP11-734K2.3	downstream gene variant, upstream gene variant	325	ENST00000623264	?	?	8
RP11-734K2.4	upstream gene variant	754	ENST00000605233	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
POC1B	ENST00000551819	?	?	2
POC1B	ENST00000548715	?	ENSP00000449945	52
POC1B	ENST00000539190	?	?	29
POC1B	ENST00000549035	NM_001199777.1	ENSP00000447916	51
POC1B	ENST00000547274	?	ENSP00000449648	20
POC1B	ENST00000546740	?	?	23
POC1B	ENST00000313546	NM_172240.2	ENSP00000323302	52
POC1B	ENST00000549504	?	ENSP00000450118	51
POC1B	ENST00000393179	?	ENSP00000376877	51
POC1B	ENST00000552563	?	?	26
POC1B	ENST00000547496	?	ENSP00000447437	52
POC1B	ENST00000546830	?	ENSP00000449256	17
POC1B	ENST00000549304	?	?	4
POC1B-GALNT4	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	4
POC1B-GALNT4	ENST00000547474	?	ENSP00000447754	2
RP11-734K2.4	ENST00000605233	?	?	20

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GALNT4	ENST00000529983	NM_003774.4	ENSP00000436604	2
POC1B-GALNT4	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-734K2.4	ENST00000605233	?	6

Locus 188 (sentinel SNP rs10858881 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 189 (rs3848570)

SNiPAcard

Block annotations

Block info

genomic range	chr19:8,137,843-8,149,247
block size	11,405 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.143 [-4.325 – 3.177]	gene(s) hit or close-by	ELAVL1, FBN3
phastCons	μ = 0.232 [0 – 1]	eQTL gene(s)	–
GERP++	μ = -0.950 [-7.86 – 2.27]	potentially regulated gene(s)	–
CADD score	μ = 3.837 [0.226 – 22.4]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FBN3	missense variant	ENST00000600128	?	ENSP00000470498	H/R	cAc/cGc	?	?	1
FBN3	missense variant	ENST00000270509	NM_032447.3	ENSP00000270509	H/R	cAc/cGc	?	?	1
FBN3	missense variant	ENST00000601739	?	ENSP00000472324	H/R	cAc/cGc	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FBN3	downstream gene variant, upstream gene variant	987	ENST00000595036	?	?	4
FBN3	downstream gene variant	2841	ENST00000601281	?	ENSP00000472729	1
FBN3	downstream gene variant	3462	ENST00000598269	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
FBN3	ENST00000270509	NM_032447.3	ENSP00000270509	H	caT/caC	1
FBN3	ENST00000600128	?	ENSP00000470498	H	caT/caC	1
FBN3	ENST00000601739	?	ENSP00000472324	H	caT/caC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ELAVL1	ENST00000351593	?	ENSP00000264073	10
FBN3	ENST00000270509	NM_032447.3	ENSP00000270509	9
FBN3	ENST00000600128	?	ENSP00000470498	9
FBN3	ENST00000601739	?	ENSP00000472324	9

FBN3	ENST00000595036	?	?	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FBN3	ENST00000595036	?	2

Locus 190 (rs28362459)

SNiPAcard

Block annotations

Block info

genomic range	chr19:5,844,792-5,844,792
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.986	gene(s) hit or close-by	AC024592.9, FUT3
phastCons	0.002	eQTL gene(s)	–
GERP++	2.33	potentially regulated gene(s)	–
CADD score	8.418	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM930258	1
Le(-) PHENOTYPE	OMIM curated	OMIM	MIM:111100	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FUT3	missense variant	ENST00000589918	NM_001097640.1	ENSP00000468123	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000589620	NM_001097639.1	ENSP00000465804	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000303225	NM_000149.3	ENSP00000305603	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000587048	?	ENSP00000468515	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000585715	?	ENSP00000467633	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000458379	NM_001097641.1	ENSP00000416443	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000589714	?	ENSP00000467081	R/L	cGg/cTg	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC024592.9	upstream gene variant	2686	ENST00000589276	?	?	1
FUT3	downstream gene variant	1784	ENST00000593144	?	?	1
FUT3	downstream gene variant	2026	ENST00000588539	?	?	1
FUT3	downstream gene variant	45	ENST00000587183	?	ENSP00000468795	1

Locus 191 (rs3118663)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,194,812-136,284,450
block size	89,639 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.780 [-3.93 – 0.701]	gene(s) hit or close-by	ADAMTS13, AL593848.2, MED22, REXO4, RP11-244N20.7, RPL7A, SNORD24, SNORD36A, SNORD36B, SNORD36C, SURF1, SURF2, SURF4, SURF6, Y_RNA
phastCons	μ = 0.021 [0 – 0.275]	eQTL gene(s)	ADAMTS13, CACFD1, MED22, REXO4, RP11-244N20.7, SLC2A6, SURF1, SURF2, SURF6
GERP++	μ = -1.493 [-6.07 – 2.05]	potentially regulated gene(s)	AK8, EEF1A1P5, GTF3C5, REXO4, REXO4, SARDH, SURF1, SURF1, SURF2, SURF2
CADD score	μ = 3.308 [0.026 – 12.47]	disease gene(s)	ADAMTS13, SURF1, SARDH,

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Stroke	2.45×10-4	dbGaP	pha002886	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	benign	ClinVar	RCV000128344.1	1

Disease gene annotation

gene	trait	source DB	source entry/link
ADAMTS13	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL	OMIM	MIM:274150
SURF1	LEIGH SYNDROME	OMIM	MIM:256000
SARDH	SARCOSINEMIA	OMIM	MIM:268900
SURF1	Complex IV deficiency	DECIPHER	MIM:220110
SURF1	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
	Complex IV deficiency	DECIPHER	MIM:220110
	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
ADAMTS13	Congenital thrombotic thrombocytopenic purpura due to ADAMTS-13 deficiency	OrphaNet	OrphaNet:93583
SURF1	Fatal infantile cytochrome C oxidase deficiency	OrphaNet	OrphaNet:1561
SURF1	Leigh syndrome with cardiomyopathy	OrphaNet	OrphaNet:70474
SURF1	Leigh syndrome with leukodystrophy	OrphaNet	OrphaNet:255241
SURF1	SURF1-related Charcot-Marie-Tooth disease type 4	OrphaNet	OrphaNet:391351
SARDH	SARCOSINEMIA	OrphaNet	OrphaNet:3129

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SURF6	ENST00000372022	ILMN_1778032	skin	5.13×10-20 (p-value)	MuTHER consortium	10
SURF6	ENST00000468290		blood	1.68×10-33 (p-value)	MuTHER consortium	10

			adipocyte	1.06×10-20 (p-value)	MuTHER consortium	10
SURF1	?	ENSG00000148290	atrial appendage	8.22×10-10 (p-value)	GTEx Portal V6	23
REXO4	?	ENSG00000148300	transformed fibroblasts	4.52×10-14 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	transformed fibroblasts	1.04×10-14 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	blood	4.49×10-40 (p-value)	GTEx Portal V6	23
RP11-244N20.7	?	ENSG00000230064	blood	1.95×10-9 (p-value)	GTEx Portal V6	23
MED22	?	ENSG00000148297	blood	9.70×10-25 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	blood	4.29×10-6 (p-value)	GTEx Portal V6	1
SURF6	?	ENSG00000148296	breast	5.95×10-8 (p-value)	GTEx Portal V6	16
SURF1	?	ENSG00000148290	breast	6.16×10-17 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	blood	1.94×10-4 (q-value)	SeeQTL DB (HapMap)	10
SURF1	?	ENSG00000148290	blood	1.07×10-12 (q-value)	SeeQTL DB (HapMap)	10
SURF1	?	ENSG00000148290	EBV lymphocytes	3.39×10-11 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	sun exposed skin	7.05×10-27 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	sun exposed skin	1.09×10-25 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	sun exposed skin	1.22×10-8 (p-value)	GTEx Portal V6	12
SURF6	?	ENSG00000148296	unexposed skin	1.86×10-19 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	unexposed skin	3.05×10-12 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	aorta	7.43×10-10 (p-value)	GTEx Portal V6	22
SURF6	?	ENSG00000148296	left ventricle	4.00×10-10 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	left ventricle	2.51×10-15 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	subcutaneous adipocytes	4.51×10-19 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	subcutaneous adipocytes	2.85×10-30 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	subcutaneous adipocytes	2.72×10-9 (p-value)	GTEx Portal V6	18
SURF2	?	ENSG00000148291	subcutaneous adipocytes	2.52×10-6 (p-value)	GTEx Portal V6	3
SURF1	?	ENSG00000148290	caudate basal ganglia	6.03×10-10 (p-value)	GTEx Portal V6	21
ADAMTS13	?	ENSG00000160323	caudate basal ganglia	9.38×10-8 (p-value)	GTEx Portal V6	11
SURF6	?	ENSG00000148296	visceral adipocytes	1.99×10-12 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	visceral adipocytes	3.18×10-11 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	stomach	3.98×10-9 (p-value)	GTEx Portal V6	22
ADAMTS13	?	ENSG00000160323	cortex	6.77×10-8 (p-value)	GTEx Portal V6	17
SURF1	?	ENSG00000148290	cortex	2.90×10-9 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	tibial nerve	7.30×10-13 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	tibial nerve	2.93×10-13 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	tibial nerve	4.00×10-7 (p-value)	GTEx Portal V6	15
SURF6	?	ENSG00000148296	esophagus mucosa	4.21×10-14 (p-value)	GTEx Portal V6	23

SURF6	?	ENSG00000148296	esophagus mucosa	4.21×10-14 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	esophagus mucosa	1.14×10-18 (p-value)	GTEx Portal V6	23
CACFD1	?	ENSG00000160325	esophagus mucosa	3.18×10-7 (p-value)	GTEx Portal V6	16
SURF1	?	ENSG00000148290	anterior cingulate cortex	3.43×10-15 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	testis	3.25×10-14 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	frontal cortex	1.25×10-10 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	frontal cortex	2.35×10-8 (p-value)	GTEx Portal V6	2
SURF6	?	ENSG00000148296	pancreas	1.13×10-10 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	muscularis mucosae	2.30×10-16 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	muscularis mucosae	1.47×10-7 (p-value)	GTEx Portal V6	13
SURF6	?	ENSG00000148296	lung	2.37×10-15 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	lung	4.25×10-16 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	lung	4.53×10-6 (p-value)	GTEx Portal V6	1
SURF6	?	ENSG00000148296	tibial artery	2.73×10-11 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	tibial artery	4.09×10-9 (p-value)	GTEx Portal V6	18
SURF6	?	ENSG00000148296	thyroid	9.98×10-10 (p-value)	GTEx Portal V6	22
CACFD1	?	ENSG00000160325	thyroid	2.38×10-11 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	thyroid	4.92×10-14 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	thyroid	2.24×10-7 (p-value)	GTEx Portal V6	13
SURF6	?	ENSG00000148296	transverse colon	1.30×10-10 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	transverse colon	6.90×10-14 (p-value)	GTEx Portal V6	23
SURF6	ENST00000372022	ILMN_1778032	monocyte	1.38×10-191 (p-value)	Zeller et al.	3
SURF6	ENST00000468290					
MED22	ENST00000614493	ILMN_1792860	monocyte	2.23×10-20 (p-value)	Zeller et al.	2
MED22	ENST00000610888					
MED22	ENST00000614493	ILMN_1697218	monocyte	2.87×10-60 (p-value)	Zeller et al.	2
MED22	ENST00000610672					
MED22	ENST00000343730					
MED22	ENST00000610888					
?	?	ILMN_1663407	monocyte	2.76×10-21 (p-value)	Zeller et al.	1
SURF6	?	ENSG00000148296	terminal ileum	1.93×10-7 (p-value)	GTEx Portal V6	8
SURF6	?	ENSG00000148296	spleen	2.60×10-15 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	sigmoid colon	1.89×10-8 (p-value)	GTEx Portal V6	14
SURF6	?	ENSG00000148296	gastroesophageal junction	5.21×10-11 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	adrenal gland	2.40×10-8 (p-value)	GTEx Portal V6	18
SURF1	?	ENSG00000148290	cerebellar hemisphere	1.08×10-9 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	cerebellum	8.00×10-8 (p-value)	GTEx Portal V6	21
SURF1	?	ENSG00000148290	cerebellum	3.12×10-15 (p-value)	GTEx Portal V6	23

SURF1	?	ENSG00000148290	hypothalamus	1.36×10-8 (p-value)	GTEx Portal V6	14
SURF1	?	ENSG00000148290	prostate	1.35×10-7 (p-value)	GTEx Portal V6	6
MED22	ENST00000614493	ILMN_1697218	monocyte	3.91×10-19 (p-value)	Fairfax et al.	2
MED22	ENST00000610672		b-cell	2.08×10-16 (p-value)	Fairfax et al.	2
MED22	ENST00000343730					
MED22	ENST00000610888					
SURF6	ENST00000372022	ILMN_1778032	monocyte	1.29×10-43 (p-value)	Fairfax et al.	2
SURF6	ENST00000468290		b-cell	1.11×10-17 (p-value)	Fairfax et al.	2
?	?	ILMN_1663407	monocyte	6.11×10-5 (p-value)	Fairfax et al.	2
			b-cell	1.67×10-5 (p-value)		
SLC2A6	ENST00000371899	ILMN_1778321	blood	6.13×10-7 (p-value)	Westra et al.	5
SLC2A6	ENST00000485978					
SLC2A6	ENST00000371897					
SURF1	?	ENSG00000148290	skeletal muscle	4.20×10-7 (p-value)	GTEx Portal V6	3
SURF1	?	ENSG00000148290	hippocampus	1.61×10-7 (p-value)	GTEx Portal V6	5
SURF1	?	ENSG00000148290	brain	1.67×10-2 (q-value)	SeeQTL DB (Myers et al.)	1
ADAMTS13	?	ENSG00000160323	pituitary	1.95×10-6 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
MED22	ENST00000614493	ILMN_1792860	chr9	monocyte	5.32×10-23 (p-value)	Zeller et al.	1
MED22	ENST00000610888						
MED22	ENST00000614493	ILMN_1697218	chr9	monocyte	7.47×10-66 (p-value)	Zeller et al.	1
MED22	ENST00000610672						
MED22	ENST00000343730						
MED22	ENST00000610888						
?	?	ILMN_1663407	chr9	monocyte	9.27×10-22 (p-value)	Zeller et al.	2

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000060505		SURF1
ENCP00000060506		SURF2
		SURF1
		SURF1
ENCP00000060514		REXO4

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000558368	1	ENCP00000060532	SARDH
		ENCP00000060469	AK8
ENCE00000558463	1	ENCP00000060478	EEF1A1P5
		ENCP00000060479	GTF3C5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors

element id	variant(s)	tissue/cell	factors
ENSR00000049921 (promoter)	1	embryonic stem cell (H1ESC)	H3K27ac, H3K36me3, CTCF, PolII, Rad21, Egr1, Gabp, Sin3Ak20, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K4me3, DNase1
		blood (K562)	DNase1, H3K4me2, PolII, ELF1, TAF1, HEY1, Cmyc, Gabp, H3K9ac, Max, Egr1, Yy1, H3K4me3
		skin (NHDF-AD)	H3K9ac, H3K4me2, DNase1, H3K4me3
		muscle (HSMM)	H3K9ac, H3K4me2, H3K4me3
		liver (HepG2)	DNase1, H3K4me3, Gabp, Yy1, PolII, TAF1, ELF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	DNase1, PolII, H2AZ, Egr1, Yy1, Gabp, ELF1, H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	CTCF, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	H3K4me3, H3K36me3
		Osteobl	H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K4me1
		breast (HMEC)	DNase1, CTCF, H3K4me2, H3K4me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K9ac, H3K4me3, H3K36me3, H3K27ac, H3K4me2, DNase1
		endothelium (HUVEC)	H3K36me3, H3K4me3, PolII, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, H3K36me3
ENSR00000049926 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K79me2, H3K36me3
		blood (K562)	H3K36me3, H4K20me1, H3K79me2, PolII
		muscle (HSMM)	H3K36me3, H4K20me1, H3K79me2
		liver (HepG2)	H4K20me1, H3K79me2, H3K4me1, H3K36me3
		lung (IMR90)	H4K20me1, H3K79me2, H3K36me3
		blood (GM12878)	H3K36me3, H3K4me2, H3K27ac, PolII, H3K79me2, H3K4me3
		nervous (NH-A)	H4K20me1, H3K36me3
		skin (NHEK)	H3K4me1, H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3, H3K4me2, H4K20me1
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001475646 (promoter)	2	embryonic stem cell (H1ESC)	H3K27me3, CTCF, PolII, Egr1, Gabp, Jund, TAF7, H3K9ac, H3K27ac, TAF1, H3K4me2, SP1, H3K4me3, DNase1
		HSMMtube	H3K79me2, H3K4me2, H2AZ, DNase1
		blood (K562)	H3K27ac, Max, E2F6, H3K9ac, H3K4me3, DNase1, H3K4me2, PolII, TAF1, H2AZ, HEY1, H3K79me2
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K79me2, H2AZ, H3K4me3, H3K27ac, DNase1, H3K9ac, H3K4me2
		liver (HepG2)	DNase1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3, H3K4me2, H3K4me1, Gabp, PolII, H3K79me2, TAF1, ELF1
		lung (IMR90)	DNase1, H3K79me2, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		skin (NHEK)	CTCF, H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1, H3K36me3
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3, H3K36me3, H4K20me1, H3K9ac
		endothelium (HUVEC)	H3K36me3, PolII, DNase1, H3K27ac, H3K9ac, H3K4me3, Max, H3K4me2
		A549	H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ADAMTS13	upstream gene variant	2670	ENST00000371916	?	ENSP00000360984	1
ADAMTS13	upstream gene variant	2995	ENST00000474918	?	ENSP00000435305	1

ADAMTS13	upstream gene variant	2994	ENST00000495234	?	ENSP00000435274	1
ADAMTS13	upstream gene variant	2994	ENST00000371911	?	ENSP00000360979	1
ADAMTS13	upstream gene variant	2994	ENST00000355699	NM_139027.4	ENSP00000347927	1
ADAMTS13	upstream gene variant	2670	ENST00000371929	NM_139025.4	ENSP00000360997	1
ADAMTS13	upstream gene variant	2994	ENST00000356589	NM_139026.4	ENSP00000348997	1
AL593848.2	downstream gene variant, upstream gene variant	527	ENST00000626556	?	?	5
MED22	downstream gene variant, upstream gene variant	4032	ENST00000610672	?	ENSP00000482438	3
MED22	downstream gene variant, upstream gene variant	3660	ENST00000471524	?	?	3
MED22	downstream gene variant, upstream gene variant	1967	ENST00000343730	NM_133640.4	ENSP00000342343	3
MED22	upstream gene variant	3612	ENST00000494177	?	ENSP00000420815	2
MED22	downstream gene variant, upstream gene variant	893	ENST00000482295	?	?	3
MED22	upstream gene variant	3604	ENST00000457204	?	ENSP00000404178	2
MED22	downstream gene variant, upstream gene variant	1967	ENST00000610888	NM_181491.2	ENSP00000478773	3
MED22	downstream gene variant, upstream gene variant	3618	ENST00000371999	?	ENSP00000361067	3
MED22	upstream gene variant	3713	ENST00000446777	?	ENSP00000394781	2
MED22	downstream gene variant, upstream gene variant	1968	ENST00000614493	?	ENSP00000481493	3
REXO4	downstream gene variant, upstream gene variant	187	ENST00000371935	NM_001279349.1	ENSP00000361003	3
REXO4	downstream gene variant, upstream gene variant	190	ENST00000371942	NM_001279350.1, NM_001279351.1, NM_020385.3	ENSP00000361010	3
REXO4	downstream gene variant, upstream gene variant	1134	ENST00000454825	?	ENSP00000394229	3
REXO4	downstream gene variant, upstream gene variant	1371	ENST00000478037	?	?	2
REXO4	downstream gene variant, upstream gene variant	1376	ENST00000494045	?	?	2
REXO4	downstream gene variant, upstream gene variant	733	ENST00000445916	?	ENSP00000391534	3
RP11-244N20.7	upstream gene variant	52	ENST00000439130	?	?	1
RPL7A	upstream gene variant, downstream gene variant	1040	ENST00000496554	?	?	5
RPL7A	upstream gene variant, downstream gene variant	309	ENST00000315731	?	ENSP00000361071	5
RPL7A	upstream gene variant, downstream gene variant	309	ENST00000323345	NM_000972.2	ENSP00000361076	5
RPL7A	upstream gene variant, downstream gene variant	363	ENST00000468019	?	?	5
RPL7A	upstream gene variant, downstream gene variant	309	ENST00000463740	?	?	5
RPL7A	upstream gene variant, downstream gene variant	1352	ENST00000485706	?	?	4
RPL7A	upstream gene variant, downstream gene variant	1417	ENST00000426651	?	ENSP00000416638	4
RPL7A	upstream gene variant, downstream gene variant	350	ENST00000489392	?	?	5
RPL7A	downstream gene variant	322	ENST00000492798	?	?	4
SNORD24	upstream gene variant, downstream gene variant	2265	ENST00000383884	?	?	4
SNORD36A	upstream gene variant, downstream gene variant	1207	ENST00000362874	?	?	4
SNORD36B	upstream gene variant, downstream gene variant	1567	ENST00000363961	?	?	4
SNORD36C	downstream gene variant	823	ENST00000516733	?	?	4

SURF1	downstream gene variant, upstream gene variant	170	ENST00000437995 ?	?	3
SURF1	downstream gene variant, upstream gene variant	5	ENST00000371974 NM_003172.3	ENSP00000361042	3
SURF1	downstream gene variant	470	ENST00000463965 ?	?	4
SURF1	downstream gene variant, upstream gene variant	105	ENST00000495952 ?	?	5
SURF1	downstream gene variant, upstream gene variant	5	ENST00000615505 NM_001280787.1	ENSP00000482067	3
SURF2	upstream gene variant	558	ENST00000486887 ?	?	3
SURF2	upstream gene variant	88	ENST00000495524 ?	?	5
SURF2	upstream gene variant	62	ENST00000371964 NM_017503.4, NM_001278928.1	ENSP00000361032	5
SURF4	downstream gene variant	4960	ENST00000618229 NM_001280790.1	ENSP00000483216	1
SURF4	downstream gene variant	4960	ENST00000545297 NM_001280792.1	ENSP00000446061	1
SURF4	downstream gene variant	4960	ENST00000613129 NM_001280791.1	ENSP00000481666	1
SURF4	downstream gene variant	4959	ENST00000371989 NM_033161.3, NM_001280788.1	ENSP00000361057	1
SURF6	downstream gene variant, upstream gene variant	808	ENST00000468290 ?	?	7
SURF6	downstream gene variant, upstream gene variant	799	ENST00000372022 NM_006753.5	ENSP00000361092	2
Y_RNA	downstream gene variant, upstream gene variant	1114	ENST00000364581 ?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ADAMTS13	ENST00000485925	?	?	2
MED22	ENST00000494177	?	ENSP00000420815	1
MED22	ENST00000610888	NM_181491.2	ENSP00000478773	1
MED22	ENST00000610672	?	ENSP00000482438	1
MED22	ENST00000446777	?	ENSP00000394781	1
MED22	ENST00000457204	?	ENSP00000404178	1
MED22	ENST00000471524	?	?	1
MED22	ENST00000614493	?	ENSP00000481493	1
MED22	ENST00000371999	?	ENSP00000361067	1
MED22	ENST00000343730	NM_133640.4	ENSP00000342343	1
REXO4	ENST00000494045	?	?	1
REXO4	ENST00000478037	?	?	1
REXO4	ENST00000371935	NM_001279349.1	ENSP00000361003	2
REXO4	ENST00000454825	?	ENSP00000394229	2
REXO4	ENST00000371942	NM_001279350.1, NM_001279351.1, NM_020385.3	ENSP00000361010	2
SURF1	ENST00000371974	NM_003172.3	ENSP00000361042	2
SURF1	ENST00000615505	NM_001280787.1	ENSP00000482067	2
SURF1	ENST00000437995	?	?	2
SURF4	ENST00000545297	NM_001280792.1	ENSP00000446061	4
SURF4	ENST00000485435	NM_001280789.1	ENSP00000419853	4

SURF4	ENST00000371989	NM_033161.3, NM_001280788.1	ENSP00000361057	4
SURF4	ENST00000618229	NM_001280790.1	ENSP00000483216	4
SURF4	ENST00000371991	?	ENSP00000361059	4
SURF4	ENST00000371982	?	?	4
SURF4	ENST00000613129	NM_001280791.1	ENSP00000481666	4
SURF4	ENST00000467910	?	?	4
SURF6	ENST00000372022	NM_006753.5	ENSP00000361092	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MED22	ENST00000610672	?	ENSP00000482438	1
SURF6	ENST00000372022	NM_006753.5	ENSP00000361092	5

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SURF1	ENST00000463965	?	1

Locus 192 (rs4494114)

SNiPAcard

Block annotations

Block info

genomic range	chr1:39,297,734-39,380,307
block size	82,574 bp
variant count	59 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.217 [-3.141 – 2.757]	gene(s) hit or close-by	GJA9, MYCBP, RHBDL2, RP5-864K19.4, RP5-864K19.6, RP5-864K19.7, RRAGC
phastCons	μ = 0.074 [0 – 1]	eQTL gene(s)	GJA9, MYCBP, RHBDL2, RP5-864K19.6, RP5-864K19.7
GERP++	μ = -0.291 [-6.75 – 6.03]	potentially regulated gene(s)	MACF1, RRAGC
CADD score	μ = 4.507 [0.029 – 20.2]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
GJA9	missense variant	ENST00000357771	NM_030772.4	ENSP00000350415	I/V	Ata/Gta	?	?	1
GJA9	missense variant	ENST00000360786	?	ENSP00000354020	I/V	Ata/Gta	?	?	1
RHBDL2	missense variant	ENST00000289248	?	ENSP00000289248	M/L	Atg/Ctg	?	?	1
RHBDL2	missense variant	ENST00000372990	NM_017821.3, NM_001304746.1	ENSP00000362081	M/L	Atg/Ctg	?	?	1
RP5-864K19.6	missense variant	ENST00000454994	?	ENSP00000406846	Q/R	cAg/cGg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RHBDL2	?	ENSG00000158315	esophagus mucosa	2.02×10^{-6} (p-value)	GTEx Portal V6	36
RP5-864K19.6	ENST00000489803	ILMN_1710161	blood	1.14×10^{-4} (p-value)	Westra et al.	1
GJA9	ENST00000357771					
RP5-864K19.7	ENST00000622355					
MYCBP	ENST00000397572					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
HNF4A	MA0114.2	6	yes	0.000	1
EGR1	MA0162.2	13	no	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000015074	1	ENCP00000001927	MACF1
		ENCP00000001918	RRAGC

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001579011 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, Jund, Sin3Ak20, H3K9ac, ATF3, TAF1, H3K4me2, SP1, USF1, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	Egr1, H3K27ac, Max, SP2, H3K9ac, Nfe2, USF1, Cmyc, H2AZ, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	TAF1, USF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, PolII, ATF3, DNase1
		blood (GM12878)	DNase1, Egr1, H3K79me2, H3K4me3, USF1, Cfos, H3K4me2, H3K9ac, Pbx3, ATF3, PolII, H2AZ
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K4me1, H3K4me2, H3K27ac
		breast (HMEC)	H3K4me3, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, Max, H3K4me3, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, PolII, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00000164746 (promoter)	2	embryonic stem cell (H1ESC)	H3K9ac, H3K36me3, CTCF, PolII, Rad21, Sin3Ak20, Yy1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K36me3, DNase1, H2AZ, H3K4me3, H3K4me2
		blood (K562)	H3K4me3, DNase1, H3K27ac, Max, H3K9ac, Gabp, H2AZ, PolII, H3K4me2, H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H2AZ, H3K4me3, H3K4me2
		liver (HepG2)	Gabp, PolII, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K4me2, H3K9ac, H3K4me3, DNase1
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	Nrf1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, Gabp, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me3, H3K4me2, PolII, DNase1
		A549	H3K36me3, DNase1, H3K4me3, H3K4me2, H3K9ac
ENSR00000281600 (CTCF binding site)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		liver (HepG2)	FOXA1, CTCF
		nervous (NH-A)	DNase1
		blood (K562)	Egr1, CTCF
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GJA9	downstream gene variant, upstream gene variant	541	ENST00000357771	NM_030772.4	ENSP00000350415	5
GJA9	downstream gene variant, upstream gene variant	441	ENST00000360786	?	ENSP00000354020	10
MYCBP	downstream gene variant, upstream gene variant	499	ENST00000495043	?	?	7
MYCBP	downstream gene variant, upstream gene variant	505	ENST00000397572	NM_012333.4	ENSP00000380702	7
MYCBP	downstream gene variant, upstream gene variant	409	ENST00000494695	?	ENSP00000485717	7
MYCBP	downstream gene variant, upstream gene variant	1172	ENST00000465771	?	?	7

RHBDL2	downstream gene variant	868	ENST00000540558	?	ENSP00000441097	4
RHBDL2	downstream gene variant	888	ENST00000372990	NM_017821.3, NM_001304746.1	ENSP00000362081	3
RHBDL2	downstream gene variant	1377	ENST00000289248	?	ENSP00000289248	2
RP5-864K19.4	downstream gene variant	630	ENST00000456813	?	?	7
RP5-864K19.4	downstream gene variant	628	ENST00000443161	?	?	7
RP5-864K19.6	downstream gene variant, upstream gene variant	896	ENST00000489803	?	?	4
RP5-864K19.6	downstream gene variant, upstream gene variant	876	ENST00000454994	?	ENSP00000406846	5
RP5-864K19.6	downstream gene variant, upstream gene variant	891	ENST00000489575	?	?	5
RP5-864K19.6	downstream gene variant, upstream gene variant	891	ENST00000621281	?	ENSP00000479064	5
RP5-864K19.7	downstream gene variant, upstream gene variant	116	ENST00000622355	?	?	9
RRAGC	downstream gene variant, upstream gene variant	488	ENST00000373001	NM_022157.3, NM_001271851.1	ENSP00000362092	2
RRAGC	downstream gene variant	2028	ENST00000474456	?	?	1
RRAGC	downstream gene variant, upstream gene variant	2447	ENST00000493015	?	?	3
RRAGC	downstream gene variant, upstream gene variant	642	ENST00000496778	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GJA9	ENST00000357771	NM_030772.4	ENSP00000350415	8
MYCBP	ENST00000465771	?	?	3
MYCBP	ENST00000494695	?	ENSP00000485717	3
MYCBP	ENST00000397572	NM_012333.4	ENSP00000380702	5
MYCBP	ENST00000495043	?	?	3
RHBDL2	ENST00000372990	NM_017821.3, NM_001304746.1	ENSP00000362081	32
RHBDL2	ENST00000289248	?	ENSP00000289248	32
RP5-864K19.4	ENST00000456813	?	?	7
RP5-864K19.4	ENST00000443161	?	?	3
RP5-864K19.4	ENST00000433671	?	?	50
RP5-864K19.6	ENST00000489575	?	?	14
RP5-864K19.6	ENST00000489803	?	?	14
RP5-864K19.6	ENST00000621281	?	ENSP00000479064	14
RP5-864K19.6	ENST00000454994	?	ENSP00000406846	13
RP5-864K19.7	ENST00000622355	?	?	13
RRAGC	ENST00000373001	NM_022157.3, NM_001271851.1	ENSP00000362092	7
RRAGC	ENST00000474456	?	?	6
RRAGC	ENST00000493015	?	?	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RHBDL2	ENST00000372990	NM_017821.3, NM_001304746.1	ENSP00000362081	1

RHBDL2	ENST00000289248	?	ENSP00000289248	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MYCBP	ENST00000397572	NM_012333.4	ENSP00000380702	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP5-864K19.6	ENST00000489803	?	3
RP5-864K19.7	ENST00000622355	?	6
RRAGC	ENST00000474456	?	4

Locus 193 (rs241890)

SNiPAcard

Block annotations

Block info

genomic range	chr22:33,242,162-33,242,162
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.451	gene(s) hit or close-by	RP1-309I22.2, SYN3, TIMP3
phastCons	0.008	eQTL gene(s)	–
GERP++	1.6	potentially regulated gene(s)	–
CADD score	2.879	disease gene(s)	TIMP3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TIMP3	FUNDUS DYSTROPHY, PSEUDOINFLAMMATORY, OF SORSBY	OMIM	MIM:136900
TIMP3	Sorsby's fundus dystrophy	OrphaNet	OrphaNet:59181

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SYN3	ENST00000462268	?	?	1
SYN3	ENST00000358763	NM_003490.3, NM_001135774.1, NM_133633.2	ENSP00000351614	1
TIMP3	ENST00000266085	NM_000362.4	ENSP00000266085	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP1-309I22.2	ENST00000624184	?	1

Locus 195 (rs1789)

SNiPAcard

Block annotations

Block info

genomic range	chr4:15,565,736-15,695,363
block size	129,628 bp
variant count	45 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.773 [-5.238 – 1.138]	**gene(s) hit or close-by**	CC2D2A, FAM200B, FBXL5, RP11-799M12.2
phastCons	μ = 0.052 [0 – 0.697]	**eQTL gene(s)**	C1QTNF7, CC2D2A, FBXL5, RP11-484O2.1, RP11-799M12.2
GERP++	μ = -0.372 [-4.89 – 3.14]	**potentially regulated gene(s)**	BST1, FBXL5
CADD score	μ = 4.067 [0.078 – 12.67]	**disease gene(s)**	CC2D2A

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
palmitoyl sphingomyelin	6.85×10^{-5}	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link
CC2D2A	COACH syndrome	DECIPHER	MIM:216360
CC2D2A	MECKEL SYNDROME, TYPE 6	DECIPHER	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	DECIPHER	MIM:612285
CC2D2A	MECKEL SYNDROME, TYPE 6	OMIM	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	OMIM	MIM:612285
CC2D2A	COACH SYNDROME	OMIM	MIM:216360
CC2D2A	Joubert syndrome with oculorenal defect	OrphaNet	OrphaNet:2318
CC2D2A	Meckel syndrome	OrphaNet	OrphaNet:564
CC2D2A	Joubert syndrome with hepatic defect	OrphaNet	OrphaNet:1454

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-799M12.2	?	ENSG00000273133	muscularis mucosae	2.95×10^{-6} (p-value)	GTEx Portal V6	10
RP11-799M12.2	?	ENSG00000273133	lung	2.20×10^{-7} (p-value)	GTEx Portal V6	42
RP11-799M12.2	?	ENSG00000273133	atrial appendage	6.02×10^{-8} (p-value)	GTEx Portal V6	43
RP11-799M12.2	?	ENSG00000273133	tibial nerve	1.21×10^{-6} (p-value)	GTEx Portal V6	35
CC2D2A	?	ENSG00000048342	blood	5.17×10^{-7} (p-value)	GTEx Portal V6	44
RP11-799M12.2	?	ENSG00000273133	blood	7.88×10^{-20} (p-value)	GTEx Portal V6	45
C1QTNF7	?	ENSG00000163145	blood	9.51×10^{-8} (p-value)	GTEx Portal V6	43
RP11-484O2.1	?	ENSG00000251379	blood	1.64×10^{-6} (p-value)	GTEx Portal V6	33
RP11-799M12.2	?	ENSG00000273133	tibial artery	1.05×10^{-9} (p-value)	GTEx Portal V6	45

FBXL5	?	ENSG00000118564	esophagus mucosa	2.34×10-7 (p-value)	GTEx Portal V6	43
FBXL5	?	ENSG00000118564	thyroid	2.81×10-9 (p-value)	GTEx Portal V6	45
RP11-799M12.2	?	ENSG00000273133	thyroid	2.07×10-7 (p-value)	GTEx Portal V6	45
RP11-799M12.2	?	ENSG00000273133	testis	6.83×10-9 (p-value)	GTEx Portal V6	44
RP11-799M12.2	?	ENSG00000273133	sun exposed skin	2.79×10-10 (p-value)	GTEx Portal V6	45
RP11-799M12.2	?	ENSG00000273133	subcutaneous adipocytes	1.67×10-15 (p-value)	GTEx Portal V6	45
FBXL5	ENST00000511441	ILMN_1673370	skin	5.90×10-11 (p-value)	MuTHER consortium	16
FBXL5	ENST00000412094		blood	1.34×10-36 (p-value)	MuTHER consortium	16
FBXL5	ENST00000513163		adipocyte	1.64×10-26 (p-value)	MuTHER consortium	16
FBXL5	ENST00000341285					
FBXL5	ENST00000511441	ILMN_1673370	monocyte	7.94×10-10 (p-value)	Fairfax et al.	6
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
FBXL5	ENST00000511441	209005_at	blood	1.00×10-12 (p-value)	Dixon et al.	4
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
FBXL5	ENST00000511441	ILMN_1673370	monocyte	7.68×10-103 (p-value)	Zeller et al.	2
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000374191	1	ENCP00000040472	FBXL5
		ENCP00000040476	BST1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001429713 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, H4K5ac, H3K36me3, H3K27me3, PolII, Rad21, TAF7, CTCF, H3K27ac
		HSMMtube	H3K4me3, H2AZ, DNase1, H3K79me2, CTCF, H3K9ac, H3K4me2, H3K27ac
		blood (K562)	H3K36me3, DNase1, H3K4me3, CTCF, H3K4me2, Egr1, H3K27ac, Max, Rad21, E2F6, H3K9ac, USF1, Cmyc, H3K79me2, H2AZ, PolII
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac
		liver (HepG2)	DNase1, PolII, H3K79me2, Rad21, TAF1, USF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3
		blood (GM12878)	CTCF, H3K9ac, PolII, H2AZ, DNase1, Egr1, Yy1, Rad21, H3K79me2, H3K4me3, USF1, H3K27ac, H3K4me2
		lung (IMR90)	DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K4me3, H3K4ac, H3K9ac, CTCF
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K27ac, H3K9ac
		NHLF	H3K4me3, DNase1, H3K9ac, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K36me3, H3K4me3, H3K9ac, H3K27ac
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, CTCF, PolII, H3K79me2, H3K4me3, H3K27ac, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3

		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, CTCF, DNase1
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
ENSR00001683631 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27ac, H3K4me2, H3K4me1
		A549	H3K36me3
		nervous (NH-A)	H3K36me3
ENSR00001429716 (enhancer)	2	monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		blood (DND-41)	H3K36me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K36me3, H3K79me2
ENSR00001237226 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K9ac, H3K36me3, CTCF, PolII, Rad21, Gabp, TAF7, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, Srf
		HSMMtube	H3K4me3, H2AZ, DNase1, H3K4me2, H3K9ac, H3K79me2
		blood (K562)	Ini1, Egr1, H3K27ac, ZBTB33, Max, Srf, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K4me3, DNase1, H3K9ac
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2
		liver (HepG2)	H3K9ac, H3K27ac, H3K4me3, DNase1, HNF4A, H3K4me2, Gabp, Srf, Sin3Ak20, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, ZBTB33
		lung (IMR90)	H3K4me3, H3K9ac, DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2, H3K36me3
		blood (GM12878)	Sin3Ak20, PU1, PolII, H2AZ, DNase1, ELF1, H3K79me2, H3K4me3, ZBTB33, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K36me3, DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K36me3, PolII, Gabp, H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K9ac, H3K36me3, H3K4me3, H3K27ac, DNase1, H3K4me1, H3K4me2
		endothelium (HUVEC)	PolII, DNase1, H3K36me3, H3K27ac, H3K9ac, Max, H3K4me2, H3K4me3
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CC2D2A	downstream gene variant	3251	ENST00000503292	NM_001080522.2	ENSP00000421809	1
CC2D2A	downstream gene variant	3262	ENST00000389652	?	ENSP00000374303	1
CC2D2A	upstream gene variant, downstream gene variant	1711	ENST00000513035	?	?	3
CC2D2A	downstream gene variant	3437	ENST00000514039	?	?	1
CC2D2A	downstream gene variant	3251	ENST00000424120	?	ENSP00000403465	1
CC2D2A	downstream gene variant	3256	ENST00000506643	?	ENSP00000422931	1
FAM200B	upstream gene variant	938	ENST00000509022	?	?	2
FAM200B	upstream gene variant	855	ENST00000504823	?	?	2
FAM200B	upstream gene variant	942	ENST00000504598	?	?	2
FAM200B	upstream gene variant	1021	ENST00000503600	?	?	2
FAM200B	upstream gene variant	848	ENST00000510920	?	?	2
FAM200B	upstream gene variant	942	ENST00000512855	?	?	2
FAM200B	upstream gene variant	1011	ENST00000508567	?	?	2

FAM200B	upstream gene variant	982	ENST00000507992	?	?	2
FAM200B	upstream gene variant	992	ENST00000513053	?	?	2
FAM200B	upstream gene variant	977	ENST00000507305	?	?	2
FAM200B	upstream gene variant, downstream gene variant	904	ENST00000504137	?	?	3
FAM200B	upstream gene variant	1092	ENST00000510186	?	?	2
FAM200B	upstream gene variant	919	ENST00000515697	?	?	2
FAM200B	upstream gene variant	940	ENST00000510032	?	?	2
FAM200B	upstream gene variant, downstream gene variant	992	ENST00000422728	?	ENSP00000393017	3
FAM200B	upstream gene variant	943	ENST00000502502	?	?	2
FAM200B	upstream gene variant	1009	ENST00000514803	?	?	2
FAM200B	upstream gene variant, downstream gene variant	1047	ENST00000502856	?	?	3
FAM200B	upstream gene variant	942	ENST00000515430	?	?	2
FAM200B	upstream gene variant, downstream gene variant	915	ENST00000622362	NM_001145191.1	ENSP00000483930	3
FAM200B	upstream gene variant	943	ENST00000503617	?	ENSP00000422751	2
FAM200B	upstream gene variant, downstream gene variant	1593	ENST00000506610	?	?	4
FAM200B	upstream gene variant, downstream gene variant	942	ENST00000505260	?	?	3
FBXL5	downstream gene variant, upstream gene variant	198	ENST00000511441	?	ENSP00000425027	5
FBXL5	downstream gene variant, upstream gene variant	1410	ENST00000514541	?	?	4
FBXL5	downstream gene variant, upstream gene variant	1446	ENST00000507899	?	ENSP00000422049	4
FBXL5	downstream gene variant, upstream gene variant	1454	ENST00000515679	?	ENSP00000424423	4
FBXL5	downstream gene variant, upstream gene variant	3048	ENST00000509314	?	ENSP00000421070	4
FBXL5	downstream gene variant, upstream gene variant	1028	ENST00000512066	?	ENSP00000426993	4
FBXL5	downstream gene variant, upstream gene variant	335	ENST00000504837	?	?	5
FBXL5	downstream gene variant, upstream gene variant	3045	ENST00000503196	?	ENSP00000425541	4
FBXL5	downstream gene variant, upstream gene variant	1522	ENST00000510802	?	ENSP00000423961	3
FBXL5	downstream gene variant, upstream gene variant	289	ENST00000507700	?	?	5
FBXL5	downstream gene variant, upstream gene variant	198	ENST00000412094	NM_001193535.1	ENSP00000408679	5
FBXL5	downstream gene variant, upstream gene variant	198	ENST00000513163	?	ENSP00000425472	4
FBXL5	downstream gene variant, upstream gene variant	218	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CC2D2A	ENST00000506643	?	ENSP00000422931	7
CC2D2A	ENST00000503292	NM_001080522.2	ENSP00000421809	7
CC2D2A	ENST00000514039	?	?	3
CC2D2A	ENST00000389652	?	ENSP00000374303	7
CC2D2A	ENST00000424120	?	ENSP00000403465	7

CC2D2A	ENST00000424120	?	ENSP00000403465	7
FAM200B	ENST00000509022	?	?	1
FAM200B	ENST00000622362	NM_001145191.1	ENSP00000483930	1
FAM200B	ENST00000514803	?	?	1
FAM200B	ENST00000510920	?	?	1
FAM200B	ENST00000512855	?	?	1
FAM200B	ENST00000508567	?	?	1
FAM200B	ENST00000507992	?	?	1
FAM200B	ENST00000507305	?	?	1
FAM200B	ENST00000513053	?	?	1
FAM200B	ENST00000504137	?	?	1
FAM200B	ENST00000510186	?	?	1
FAM200B	ENST00000510032	?	?	1
FAM200B	ENST00000502856	?	?	1
FAM200B	ENST00000503600	?	?	1
FAM200B	ENST00000503617	?	ENSP00000422751	1
FAM200B	ENST00000515697	?	?	1
FAM200B	ENST00000502502	?	?	1
FAM200B	ENST00000422728	?	ENSP00000393017	1
FAM200B	ENST00000504598	?	?	2
FAM200B	ENST00000515430	?	?	1
FAM200B	ENST00000504823	?	?	1
FAM200B	ENST00000505260	?	?	1
FBXL5	ENST00000514541	?	?	9
FBXL5	ENST00000507899	?	ENSP00000422049	16
FBXL5	ENST00000515679	?	ENSP00000424423	16
FBXL5	ENST00000510802	?	ENSP00000423961	19
FBXL5	ENST00000509314	?	ENSP00000421070	10
FBXL5	ENST00000504837	?	?	7
FBXL5	ENST00000512066	?	ENSP00000426993	4
FBXL5	ENST00000503196	?	ENSP00000425541	10
FBXL5	ENST00000507700	?	?	21
FBXL5	ENST00000513163	?	ENSP00000425472	20
FBXL5	ENST00000412094	NM_001193535.1	ENSP00000408679	23
FBXL5	ENST00000511441	?	ENSP00000425027	23
FBXL5	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	23

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FBXL5	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

RP11-799M12.2	ENST00000609724	?	1

Locus 196 (rs540254)

SNiPAcard

Block annotations

Block info

genomic range	chr1:160,759,519-160,767,737
block size	8,219 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.349 [-1.062 – 0.48]	**gene(s) hit or close-by**	LY9
phastCons	μ = 0.006 [0 – 0.034]	**eQTL gene(s)**	LY9, SLAMF7
GERP++	μ = -0.668 [-2.19 – 0.577]	**potentially regulated gene(s)**	–
CADD score	μ = 4.528 [0.201 – 10.73]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SLAMF7	ENST00000368043	ILMN_1710923	blood	1.14×10^{-7} (p-value)	MuTHER consortium	2
SLAMF7	ENST00000368042					
SLAMF7	ENST00000458602					
SLAMF7	ENST00000444090					
SLAMF7	ENST00000441662					
SLAMF7	ENST00000621377					
SLAMF7	ENST00000359331					
SLAMF7	ENST00000484221					
SLAMF7	ENST00000458104					
LY9	ENST00000392203	ILMN_1731928	b-cell	1.91×10^{-5} (p-value)	Fairfax et al.	1
LY9	ENST00000263285					
LY9	ENST00000368037					
LY9	ENST00000368035					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000167128 (open chromatin region)	1	blood (GM12878)	H3K4me1, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		blood (K562)	H3K27me3
		breast (HMEC)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LY9	upstream gene variant	1165	ENST00000368037	NM_001261456.1	ENSP00000357016	4
LY9	upstream gene variant	1302	ENST00000485624	?	?	4
LY9	upstream gene variant	1203	ENST00000392203	NM_001261457.1	ENSP00000376039	4
LY9	upstream gene variant	1197	ENST00000474998	?	ENSP00000476321	4
LY9	upstream gene variant	1299	ENST00000471816	?	?	4
LY9	upstream gene variant	1290	ENST00000480837	?	?	4
LY9	upstream gene variant	1278	ENST00000368039	NM_001033667.2	ENSP00000357018	4

LY9	upstream gene variant	1291	ENST00000490902	?	ENSP00000473643	4
LY9	upstream gene variant	1249	ENST00000263285	NM_002348.3	ENSP00000263285	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
LY9	ENST00000474998	?	ENSP00000476321	S	tcT/tcC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LY9	ENST00000368037	NM_001261456.1	ENSP00000357016	2
LY9	ENST00000485624	?	?	2
LY9	ENST00000392203	NM_001261457.1	ENSP00000376039	2
LY9	ENST00000474998	?	ENSP00000476321	2
LY9	ENST00000471816	?	?	2
LY9	ENST00000480837	?	?	2
LY9	ENST00000368039	NM_001033667.2	ENSP00000357018	2
LY9	ENST00000490902	?	ENSP00000473643	2
LY9	ENST00000263285	NM_002348.3	ENSP00000263285	2

Locus 197 (rs17741049)

SNiPAcard

Block annotations

Block info

genomic range	chr8:86,089,292-86,292,870
block size	203,579 bp
variant count	36 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.154 [-3.447 – 1.323]	gene(s) hit or close-by	C8orf59, CA1, CA13, CA3, E2F5, RP11-219B4.3, RP11-219B4.5, RP11-219B4.6, RP11-219B4.7
phastCons	μ = 0.098 [0 – 1]	eQTL gene(s)	LRRCC1
GERP++	μ = -0.634 [-6.53 – 3.79]	potentially regulated gene(s)	CA1, CA3, E2F5
CADD score	μ = 3.878 [0.014 – 14.87]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
E2F5	missense variant	ENST00000418930	NM_001083588.1	ENSP00000414312	A/G	gCg/gGg	?	?	1
E2F5	missense variant	ENST00000416274	NM_001951.3	ENSP00000398124	A/G	gCg/gGg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LRRCC1	?	ENSG00000133739	esophagus mucosa	5.45×10-6 (p-value)	GTEx Portal V6	33

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
EGR1	MA0341.1	4	yes	0.000	1
EGR1	MA0366.1	4	yes	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000056669		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000521373	1	ENCP00000056670	CA1
ENCE00000521314	1	ENCP00000056657	E2F5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001395450 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, Rad21, TAF7, H3K27ac, TAF1, H3K4me2, H3K9ac, H3K4me3
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	Egr1, Max, H3K9ac, Cmyc, H2AZ, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K4me3, DNase1
		muscle (HSMM)	H2AZ, H3K4me3
		liver (HepG2)	Gabp, PolII, H3K79me2, TAF1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1
		lung (IMR90)	H3K4me2, H3K4me3
		blood (GM12878)	H2AZ, Tcf12, Cmyc, Egr1, Yy1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, PolII, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	CTCF, H3K4me2, H3K4me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	H3K4me2, H3K4me3, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K4me3, PolII, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, H3K4me2, H3K4me3, Max
		A549	H3K36me3, H3K27ac, DNase1, H3K4me3, H3K4me2, H3K9ac
ENSR00001442694 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	H3K27me3, CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3
ENSR00001442695 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C8orf59	downstream gene variant	1417	ENST00000612977	NM_001099671.1, NM_001099670.1	ENSP00000484101	1
C8orf59	downstream gene variant	1432	ENST00000545322	?	ENSP00000444092	1
C8orf59	downstream gene variant	1433	ENST00000611854	?	ENSP00000484811	1

C8orf59	downstream gene variant	1432	ENST00000612809	NM_001099672.1	ENSP00000481745	1
C8orf59	downstream gene variant	1945	ENST00000523281	?	ENSP00000428722	1
C8orf59	downstream gene variant	3743	ENST00000523245	?	?	1
C8orf59	downstream gene variant	1486	ENST00000615697	?	ENSP00000482944	1
C8orf59	downstream gene variant	1432	ENST00000615071	?	?	1
C8orf59	downstream gene variant	1007	ENST00000614462	NM_001293320.1	ENSP00000480167	1
C8orf59	downstream gene variant	1006	ENST00000619594	NM_001099673.1	ENSP00000484492	1
C8orf59	downstream gene variant	4090	ENST00000321777	?	ENSP00000319020	1
CA1	upstream gene variant	2528	ENST00000523022	NM_001738.3, NM_001128830.2, NM_001128831.2, NM_001128829.2	ENSP00000429798	1
CA1	upstream gene variant	2528	ENST00000523858	?	ENSP00000430975	1
CA1	upstream gene variant	2528	ENST00000522814	?	ENSP00000430737	1
CA1	upstream gene variant	2528	ENST00000520990	?	?	1
CA1	upstream gene variant	2528	ENST00000518341	?	?	1
CA1	upstream gene variant	2528	ENST00000517590	?	ENSP00000429843	1
CA1	upstream gene variant	2528	ENST00000519991	?	ENSP00000430543	1
CA1	upstream gene variant	2533	ENST00000521846	?	ENSP00000430471	1
CA1	upstream gene variant	2528	ENST00000522662	?	ENSP00000430372	1
CA1	upstream gene variant	1627	ENST00000626824	?	ENSP00000486171	1
CA1	upstream gene variant	2528	ENST00000524324	?	ENSP00000428923	1
CA1	upstream gene variant	2528	ENST00000519129	?	ENSP00000429688	1
CA1	upstream gene variant	2528	ENST00000520663	?	ENSP00000430571	1
CA1	upstream gene variant	2528	ENST00000520093	?	?	1
CA1	upstream gene variant	2528	ENST00000522579	?	ENSP00000427852	1
CA1	upstream gene variant	1627	ENST00000523953	?	ENSP00000430656	1
CA1	upstream gene variant	2528	ENST00000517429	?	ENSP00000430710	1
CA13	downstream gene variant	3683	ENST00000517298	?	?	1
CA13	upstream gene variant, downstream gene variant	740	ENST00000518392	?	?	6
CA13	upstream gene variant, downstream gene variant	825	ENST00000321764	NM_198584.2	ENSP00000318912	3
CA13	downstream gene variant	1119	ENST00000522631	?	?	2
CA3	upstream gene variant	1127	ENST00000520921	?	ENSP00000429760	2
E2F5	upstream gene variant	978	ENST00000521429	?	ENSP00000428606	1
E2F5	upstream gene variant	978	ENST00000517476	NM_001083589.1	ENSP00000429120	1
E2F5	upstream gene variant	3406	ENST00000521234	?	?	1
E2F5	upstream gene variant	330	ENST00000416274	NM_001951.3	ENSP00000398124	1
E2F5	upstream gene variant	168	ENST00000418930	NM_001083588.1	ENSP00000414312	1
E2F5	upstream gene variant	139	ENST00000256117	?	ENSP00000256117	2
RP11-219B4.3	downstream gene variant, upstream gene variant	49	ENST00000520129	?	?	3

RP11-219B4.5	upstream gene variant	2439	ENST00000546501 ?	?	2
RP11-219B4.5	upstream gene variant	1532	ENST00000549291 ?	?	2
RP11-219B4.6	downstream gene variant	2348	ENST00000551479 ?	?	1
RP11-219B4.7	upstream gene variant	411	ENST00000566000 ?	?	3
RP11-219B4.7	upstream gene variant	16	ENST00000562577 ?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CA1	ENST00000523953	?	ENSP00000430656	3
CA1	ENST00000522579	?	ENSP00000427852	3
CA1	ENST00000519991	?	ENSP00000430543	3
CA1	ENST00000521846	?	ENSP00000430471	3
CA1	ENST00000524324	?	ENSP00000428923	3
CA1	ENST00000523022	NM_001738.3, NM_001128830.2, NM_001128831.2, NM_001128829.2	ENSP00000429798	3
CA1	ENST00000523858	?	ENSP00000430975	3
CA1	ENST00000522814	?	ENSP00000430737	3
CA1	ENST00000519129	?	ENSP00000429688	3
CA1	ENST00000518341	?	?	3
CA1	ENST00000520093	?	?	3
CA1	ENST00000520990	?	?	3
CA1	ENST00000626824	?	ENSP00000486171	3
CA1	ENST00000517590	?	ENSP00000429843	3
CA1	ENST00000520663	?	ENSP00000430571	3
CA1	ENST00000517429	?	ENSP00000430710	3
CA1	ENST00000522662	?	ENSP00000430372	3
CA13	ENST00000517831	?	?	9
CA13	ENST00000517298	?	?	13
CA13	ENST00000522631	?	?	4
CA13	ENST00000321764	NM_198584.2	ENSP00000318912	10
CA3	ENST00000520921	?	ENSP00000429760	1
E2F5	ENST00000521429	?	ENSP00000428606	4
E2F5	ENST00000416274	NM_001951.3	ENSP00000398124	7
E2F5	ENST00000256117	?	ENSP00000256117	7
E2F5	ENST00000518234	?	ENSP00000429669	1
E2F5	ENST00000517476	NM_001083589.1	ENSP00000429120	4
E2F5	ENST00000520225	?	ENSP00000430461	1
E2F5	ENST00000418930	NM_001083588.1	ENSP00000414312	7
RP11-219B4.5	ENST00000546501	?	?	4
RP11-219B4.5	ENST00000549291	?	?	7

RP11-219B4.6	ENST00000551479	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CA13	ENST00000321764	NM_198584.2	ENSP00000318912	1
E2F5	ENST00000256117	?	ENSP00000256117	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-219B4.3	ENST00000520129	?	1
RP11-219B4.7	ENST00000566000	?	1

Locus 198 (rs2248372)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,446,466-31,452,103
block size	5,638 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.604 [-1.421 – -0.238]	gene(s) hit or close-by	HCP5
phastCons	μ = 0.004 [0.001 – 0.006]	eQTL gene(s)	ATP6V1G2, HCG27, HLA-B, HLA-C, LST1, LY6G5C, MICA, MICB, PSORS1C1, PSORS1C2, XXbac-BPG181B23.7, XXbac-BPG248L24.12
GERP++	μ = -0.018 [-0.737 – 0.458]	potentially regulated gene(s)	CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, LY6G6D, LY6G6D, LY6G6D, LY6G6D, LY6G6D, LY6G6D, LY6G6E, LY6G6E, LY6G6E, LY6G6E, LY6G6E, LY6G6E, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P
CADD score	μ = 2.082 [0.871 – 4.161]	disease gene(s)	HLA-B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
gamma-glutamylglutamine	2.92×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PSORS1C1	ENST00000481450	ILMN_1799669	skin	2.59×10^{-5} (p-value)	MuTHER consortium	1
PSORS1C1	ENST00000552747					
PSORS1C1	ENST00000547221					
PSORS1C1	ENST00000259881					
PSORS1C1	ENST00000479581					
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	7.59×10^{-6} (p-value)	MuTHER consortium	2
ATP6V1G2	ENST00000303892		skin	2.90×10^{-5} (p-value)	MuTHER consortium	2
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					
MICB	ENST00000538442	ILMN_1708006	skin	1.42×10^{-9} (p-value)	MuTHER consortium	2
MICB	ENST00000399150					
MICB	ENST00000252229					
HCG27	ENST00000383331	ILMN_1746436	adipocyte	3.10×10^{-5} (p-value)	MuTHER consortium	1
HCG27	ENST00000415276					

XXbac-BPG248L24.12	?	ENSG00000271581	lung	7.99×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	lung	2.96×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	lung	2.36×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	1.29×10-5 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	transformed fibroblasts	1.06×10-8 (p-value)	GTEx Portal V6	3
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	7.57×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	blood	4.00×10-7 (p-value)	GTEx Portal V6	4
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	1.30×10-6 (p-value)	GTEx Portal V6	4
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	1.17×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	1.19×10-8 (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	blood	1.47×10-2 (q-value)	SeeQTL DB (HapMap)	1
MICA	?	ENSG00000204520	sun exposed skin	1.48×10-9 (p-value)	GTEx Portal V6	4
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	1.96×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	3.64×10-9 (p-value)	GTEx Portal V6	4
PSORS1C1	?	ENSG00000204540	sun exposed skin	4.04×10-7 (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	sun exposed skin	6.17×10-6 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	2.30×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	left ventricle	1.79×10-6 (p-value)	GTEx Portal V6	3
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	1.88×10-6 (p-value)	GTEx Portal V6	4
PSORS1C1	?	ENSG00000204540	subcutaneous adipocytes	2.65×10-6 (p-value)	GTEx Portal V6	3
HLA-C	ENST00000470363	ILMN_2150787	monocyte	1.54×10-16 (p-value)	Zeller et al.	1
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	ENST00000466892					
PSORS1C1	?	ENSG00000204540	tibial nerve	5.92×10-9 (p-value)	GTEx Portal V6	4
PSORS1C2	?	ENSG00000204538	esophagus mucosa	7.70×10-8 (p-value)	GTEx Portal V6	4
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	2.02×10-9 (p-value)	GTEx Portal V6	4
PSORS1C1	?	ENSG00000204540	esophagus mucosa	9.36×10-9 (p-value)	GTEx Portal V6	4
LY6G5C	ENST00000383237	ILMN_1763467	blood	1.29×10-4 (p-value)	Westra et al.	1
LY6G5C	ENST00000474395					
LY6G5C	ENST00000375863					
LY6G5C	ENST00000474678					
LY6G5C	ENST00000467098					
LY6G5C	ENST00000460141					
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	1.05×10-4 (p-value)	Westra et al.	1
ATP6V1G2	ENST00000303892					
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					

LST1	ENST00000419073	ILMN_1717127	blood	7.89×10-5 (p-value)	Westra et al.	1
LST1	ENST00000433492					
LST1	ENST00000376089					
LST1	ENST00000376090					
LST1	ENST00000211921					
XXbac-BPG181B23.7	?	ENSG00000272221	testis	8.41×10-8 (p-value)	GTEx Portal V6	4
HLA-B	?	ENSG00000234745	cerebellum	1.63×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	1.16×10-6 (p-value)	GTEx Portal V6	3
PSORS1C1	?	ENSG00000204540	unexposed skin	1.03×10-6 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	breast	2.78×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444780	1	ENCP00000048082	LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D LY6G6D LY6G6E LY6G6D LY6G6D LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D
		ENCP00000047982	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
		ENCP00000048120	SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4
		ENCP00000048000	WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P
		ENCP00000047981	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

HCP5	downstream gene variant	1183	ENST00000467369	?	?	2

Locus 199 (rs3129682)

SNiPAcard

Block annotations

Block info

genomic range	chr6:28,787,384-29,523,957
block size	736,574 bp
variant count	158 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.194 [-5.844 – 4.77]	**gene(s) hit or close-by**	AL645937.1, AL662791.1, GABBR1, HCG14, HCG15, HCG16, KRT18P1, LINC01556, MAS1L, MAS1LP1, OR10C1, OR10C1, OR11A1, OR12D3, OR14J1, OR2AD1P, OR2B3, OR2B4P, OR2H1, OR2H4P, OR2I1P, OR2J2, OR2J3, OR2J4P, OR2N1P, OR2P1P, OR2U2P, OR2W1, OR5V1, RN7SL471P, RNU6-930P, RPL13P, RPS17P1, SAR1P1, TRIM27, UBD, UBDP1, XXbac-BCX196D17.5, XXbac-BPG13B8.10, XXbac-BPG13B8.11, XXbac-BPG258E24.10, XXbac-BPG308J9.3, XXbac-BPG308K3.5, XXbac-BPG308K3.6, ZNF311, ZNF90P2
phastCons	μ = 0.110 [0 – 1]	**eQTL gene(s)**	ABT1, AL022393.7, BTN3A2, BTN3A3, C2, CFB, GABBR1, HIST1H3J, HIST1H4J, HLA-A, HLA-C, HLA-F-AS1, HLA-G, HLA-H, HLA-J, HLA-K, HLA-L, HLA-U, HLA-V, OR2B7P, OR2B8P, RP1-265C24.5, RP1-313I6.12, SLC44A4, TRIM31, XXbac-BPG308K3.5, ZDHHC20P1, ZFP57, ZNF165, ZNF192P1, ZNRD1, ZNRD1-AS1_3, ZSCAN23, ZSCAN26, ZSCAN31
GERP++	μ = -0.271 [-5.5 – 2.32]	**potentially regulated gene(s)**	AL662791.1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GPX5, GPX5, GPX6, GPX6, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, OR12D3, OR12D3, OR12D3, OR12D3, OR12D3, OR12D3, OR12D3, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2B3, OR2B3, OR2B3, OR2B3, OR2B3, OR2B3, OR2B3, OR2B3, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2G1P, OR2G1P, OR2G1P, OR2G1P, OR2G1P, OR2G1P, OR2G1P, OR2H4P, OR2H4P, OR2H4P, OR2H4P, OR2H4P, OR2H4P, OR2H4P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2P1P, OR2P1P, OR2P1P, OR2P1P, OR2P1P, OR2P1P, OR2P1P, OR2U1P, OR2U1P, OR2U1P, OR2U1P, OR2U1P, TRIM27, TRIM27, TRIM27, TRIM27, TRIM27, TRIM27, TRIM27, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5
CADD score	μ = 4.022 [0.006 – 14.6]	**disease gene(s)**	ZFP57, TRIM27, OR2J3, , , , , C2, CFB, HLA-A

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Barrett's esophagus	$<4.00\times10^{-9}$	GWAS Catalog	22961001	1
phenol sulfate	2.11×10^{-6}	Metabolomics GWAS Server	24816252	8
betaine	4.49×10^{-7}	Metabolomics GWAS Server	24816252	20
Lupus erythematosus, systemic	9.64×10^{-6}	dbGaP	pha002867	4
Lupus erythematosus, systemic	8.06×10^{-9}	dbGaP	pha002848	1

Disease gene annotation

gene	trait	source DB	source entry/link
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410
TRIM27	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550
OR2J3	C3HEX, ABILITY TO SMELL	OMIM	MIM:615082

	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550	
	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550	
	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550	
	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550	
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410	
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886	
TRIM27	Papillary or follicular thyroid carcinoma	OrphaNet	OrphaNet:146	
HLA-A	Birdshot chorioretinopathy	OrphaNet	OrphaNet:179	
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
OR12D3	missense variant	ENST00000396806	NM_030959.2	ENSP00000380023	I/T	aTc/aCc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ZFP57	?	ENSG00000204644	muscularis mucosae	4.17×10-8 (p-value)	GTEx Portal V6	153
AL022393.7	?	ENSG00000216901	muscularis mucosae	6.78×10-6 (p-value)	GTEx Portal V6	5
HLA-H	?	ENSG00000206341	muscularis mucosae	3.17×10-6 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	lung	1.19×10-9 (p-value)	GTEx Portal V6	155
HLA-G	?	ENSG00000204632	lung	1.18×10-6 (p-value)	GTEx Portal V6	81
HLA-H	?	ENSG00000206341	lung	2.28×10-7 (p-value)	GTEx Portal V6	98
HLA-J	?	ENSG00000204622	lung	9.19×10-10 (p-value)	GTEx Portal V6	67
ZFP57	?	ENSG00000204644	atrial appendage	2.14×10-9 (p-value)	GTEx Portal V6	126
ZSCAN26	?	ENSG00000197062	transformed fibroblasts	7.66×10-6 (p-value)	GTEx Portal V6	108
ZFP57	?	ENSG00000204644	transformed fibroblasts	1.02×10-7 (p-value)	GTEx Portal V6	134
ZFP57	?	ENSG00000204644	blood	2.31×10-12 (p-value)	GTEx Portal V6	155
HLA-F-AS1	?	ENSG00000214922	blood	1.81×10-7 (p-value)	GTEx Portal V6	101
HLA-H	?	ENSG00000206341	blood	9.34×10-8 (p-value)	GTEx Portal V6	84
HLA-K	?	ENSG00000230795	blood	8.61×10-9 (p-value)	GTEx Portal V6	28
HLA-J	?	ENSG00000204622	blood	5.11×10-7 (p-value)	GTEx Portal V6	19
ZFP57	?	ENSG00000204644	breast	1.59×10-8 (p-value)	GTEx Portal V6	134

ZFP57	?	ENSG00000204644	tibial artery	1.18×10-9 (p-value)	GTEx Portal V6	150
HLA-H	?	ENSG00000206341	tibial artery	5.16×10-7 (p-value)	GTEx Portal V6	97
HLA-U	?	ENSG00000228078	tibial artery	1.32×10-5 (p-value)	GTEx Portal V6	11
HLA-J	?	ENSG00000204622	tibial artery	6.73×10-6 (p-value)	GTEx Portal V6	14
ZSCAN26	?	ENSG00000197062	thyroid	5.59×10-8 (p-value)	GTEx Portal V6	127
OR2B8P	?	ENSG00000182477	thyroid	8.80×10-9 (p-value)	GTEx Portal V6	99
ZFP57	?	ENSG00000204644	thyroid	2.70×10-12 (p-value)	GTEx Portal V6	155
OR2B7P	?	ENSG00000187763	thyroid	9.85×10-8 (p-value)	GTEx Portal V6	96
ZNF192P1	?	ENSG00000226314	thyroid	4.88×10-6 (p-value)	GTEx Portal V6	68
RP1-265C24.5	?	ENSG00000219392	thyroid	1.41×10-5 (p-value)	GTEx Portal V6	1
RP1-265C24.5	?	ENSG00000219392	skeletal muscle	7.35×10-7 (p-value)	GTEx Portal V6	99
AL022393.7	?	ENSG00000216901	skeletal muscle	1.86×10-11 (p-value)	GTEx Portal V6	113
ZSCAN23	?	ENSG00000187987	skeletal muscle	5.54×10-6 (p-value)	GTEx Portal V6	69
ZFP57	?	ENSG00000204644	skeletal muscle	9.19×10-8 (p-value)	GTEx Portal V6	83
HLA-H	?	ENSG00000206341	skeletal muscle	2.11×10-6 (p-value)	GTEx Portal V6	89
HLA-J	?	ENSG00000204622	skeletal muscle	9.59×10-8 (p-value)	GTEx Portal V6	67
ZFP57	?	ENSG00000204644	liver	9.11×10-7 (p-value)	GTEx Portal V6	7
OR2B8P	?	ENSG00000182477	sun exposed skin	6.50×10-8 (p-value)	GTEx Portal V6	97
ZFP57	?	ENSG00000204644	sun exposed skin	4.62×10-10 (p-value)	GTEx Portal V6	154
HLA-H	?	ENSG00000206341	sun exposed skin	1.74×10-5 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	unexposed skin	1.59×10-7 (p-value)	GTEx Portal V6	121
RP1-265C24.5	?	ENSG00000219392	left ventricle	5.52×10-10 (p-value)	GTEx Portal V6	104
AL022393.7	?	ENSG00000216901	left ventricle	1.04×10-5 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	left ventricle	1.56×10-6 (p-value)	GTEx Portal V6	17
ZNF165	?	ENSG00000197279	left ventricle	8.89×10-6 (p-value)	GTEx Portal V6	2
ZNF192P1	?	ENSG00000226314	left ventricle	6.32×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	aorta	2.84×10-9 (p-value)	GTEx Portal V6	154
ZFP57	?	ENSG00000204644	subcutaneous adipocytes	4.22×10-8 (p-value)	GTEx Portal V6	144
RP1-265C24.5	?	ENSG00000219392	subcutaneous adipocytes	3.75×10-6 (p-value)	GTEx Portal V6	55
HIST1H4J	?	ENSG00000197238	subcutaneous adipocytes	3.92×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	tibial nerve	1.32×10-8 (p-value)	GTEx Portal V6	143
AL022393.7	?	ENSG00000216901	tibial nerve	3.35×10-6 (p-value)	GTEx Portal V6	60
ZSCAN26	?	ENSG00000197062	esophagus mucosa	7.64×10-6 (p-value)	GTEx Portal V6	37
ZFP57	?	ENSG00000204644	esophagus mucosa	1.19×10-14 (p-value)	GTEx Portal V6	155
ZNRD1	?	ENSG00000066379	esophagus mucosa	7.11×10-6 (p-value)	GTEx Portal V6	1
AL022393.7	?	ENSG00000216901	cerebellum	4.16×10-6 (p-value)	GTEx Portal V6	104
ZNF192P1	?	ENSG00000226314	cerebellum	2.00×10-5 (p-value)	GTEx Portal V6	1

ZFP57	?	ENSG00000204644	stomach	2.15×10-7 (p-value)	GTEx Portal V6	131
HIST1H3J	?	ENSG00000197153	testis	1.40×10-5 (p-value)	GTEx Portal V6	1
ZDHHC20P1	?	ENSG00000233916	testis	8.22×10-7 (p-value)	GTEx Portal V6	14
RP1-313I6.12	?	ENSG00000272009	testis	4.58×10-6 (p-value)	GTEx Portal V6	14
AL022393.7	?	ENSG00000216901	testis	1.77×10-5 (p-value)	GTEx Portal V6	10
XXbac-BPG308K3.5	?	ENSG00000225173	testis	3.21×10-5 (p-value)	GTEx Portal V6	1
ZSCAN26	ENST00000421553	ILMN_1760982	blood	2.25×10-6 (p-value)	MuTHER consortium	37
ZSCAN26	ENST00000614088					
ZSCAN26	ENST00000316606					
ZSCAN26	ENST00000619937					
ZSCAN31	ENST00000396838	ILMN_1655748	skin	4.43×10-8 (p-value)	MuTHER consortium	41
ZSCAN31	ENST00000344279		adipocyte	1.04×10-5 (p-value)	MuTHER consortium	41
ZSCAN31	ENST00000414429					
ZSCAN31	ENST00000439158					
ZSCAN31	ENST00000611469					
ZSCAN31	ENST00000446474					
HLA-V	ENST00000446817	ILMN_1660923	blood	1.84×10-7 (p-value)	MuTHER consortium	48
HLA-V	ENST00000429037		adipocyte	4.32×10-6 (p-value)	MuTHER consortium	48
?	?	ILMN_1654987	adipocyte	1.04×10-6 (p-value)	MuTHER consortium	39
			blood	7.02×10-6 (p-value)		
TRIM31	ENST00000376734	ILMN_1748685	skin	5.48×10-6 (p-value)	MuTHER consortium	11
TRIM31	ENST00000484583					
TRIM31	ENST00000471695					
TRIM31	ENST00000468264					
HLA-L	ENST00000463348	ILMN_1675282	blood	2.00×10-6 (p-value)	MuTHER consortium	21
ZNRD1	ENST00000471008	ILMN_2398587	blood	1.50×10-5 (p-value)	MuTHER consortium	2
ZNRD1	ENST00000359374					
ZNRD1-AS1_3	ENST00000611686					
ZNRD1	ENST00000332435					
ZNRD1	ENST00000376782					
ZNRD1	ENST00000376785					
ZNRD1	ENST00000463141					
?	?	ILMN_1679389	monocyte	7.45×10-16 (p-value)	Zeller et al.	10
?	?	ILMN_1666078	monocyte	9.45×10-29 (p-value)	Zeller et al.	10
BTN3A2	ENST00000396934	ILMN_1676528	monocyte	7.87×10-51 (p-value)	Zeller et al.	10
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
?	?	ILMN_1820787	monocyte	1.88×10-32 (p-value)	Zeller et al.	10
BTN3A2	ENST00000524459	ILMN_1700067	monocyte	1.85×10-37 (p-value)	Zeller et al.	10
BTN3A2	ENST00000528541					
BTN3A3	ENST00000467524					
BTN3A3	ENST00000482451					
BTN3A3	ENST00000494393					
BTN3A2	ENST00000527417					
BTN3A3	ENST00000244519					
BTN3A2	ENST00000528222					
BTN3A3	ENST00000474790					

BTN3A2	ENST00000377708					
BTN3A3	ENST00000496719					
BTN3A2	ENST00000396948					
BTN3A2	ENST00000527422					
BTN3A2	ENST00000356386					
?	?	ILMN_1883997	monocyte	3.70×10-33 (p-value)	Zeller et al.	10
?	?	ILMN_1671054	monocyte	6.26×10-38 (p-value)	Zeller et al.	9
HLA-G	ENST00000376828	ILMN_1656670	monocyte	2.84×10-16 (p-value)	Zeller et al.	9
HLA-G	ENST00000360323					
HLA-G	ENST00000478519					
HLA-G	ENST00000478355					
HLA-G	ENST00000428701					
HLA-G	ENST00000376815					
HLA-G	ENST00000376818					
ZFP57	?	ENSG00000204644	visceral adipocytes	2.25×10-6 (p-value)	GTEx Portal V6	27
ZFP57	?	ENSG00000204644	pancreas	3.20×10-6 (p-value)	GTEx Portal V6	17
ZSCAN26	?	ENSG00000197062	transverse colon	5.52×10-6 (p-value)	GTEx Portal V6	11
ZFP57	?	ENSG00000204644	transverse colon	5.30×10-7 (p-value)	GTEx Portal V6	14
ABT1	ENST00000274849	ILMN_1658083	monocyte	6.95×10-5 (p-value)	Fairfax et al.	2
SLC44A4	ENST00000487680	ILMN_1719250	b-cell	3.95×10-4 (p-value)	Fairfax et al.	1
SLC44A4	ENST00000544672					
SLC44A4	ENST00000229729					
SLC44A4	ENST00000375562					
ZNRD1	ENST00000471008	ILMN_2398587	b-cell	1.40×10-4 (p-value)	Fairfax et al.	1
ZNRD1	ENST00000359374					
ZNRD1-AS1_3	ENST00000611686					
ZNRD1	ENST00000376785					
ZNRD1	ENST00000376782					
ZNRD1	ENST00000332435					
ZNRD1	ENST00000463141					
C2	ENST00000486124	ILMN_1710740	monocyte	1.43×10-4 (p-value)	Fairfax et al.	1
C2	ENST00000469372					
CFB	ENST00000425368					
C2	ENST00000497706					
C2	ENST00000442278					
C2	ENST00000452323					
C2	ENST00000383177					
C2	ENST00000299367					
C2	ENST00000482060					
RP1-313I6.12	?	ENSG00000272009	adrenal gland	1.12×10-5 (p-value)	GTEx Portal V6	1
ZSCAN26	?	ENSG00000197062	adrenal gland	8.83×10-6 (p-value)	GTEx Portal V6	1
HLA-A	?	ENSG00000206503	brain	2.27×10-5 (q-value)	SeeQTL DB (Myers et al.)	6
GABBR1	ENST00000491829	ILMN_2395375	blood	3.40×10-5 (p-value)	Westra et al.	4
GABBR1	ENST00000377012					
GABBR1	ENST00000377034					
GABBR1	ENST00000478931					
GABBR1	ENST00000472823					
GABBR1	ENST00000377016					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
BTN3A2	?	ENSG00000186470	chr6	blood	3.72×10-6 (q-value)	SeeQTL DB (HapMap)	25
?	?	ILMN_1671054	chr6	monocyte	7.32×10-38 (p-value)	Zeller et al.	1
?	?	ILMN_1752592	chr6	monocyte	5.21×10-20 (p-value)	Zeller et al.	10
HLA-G	ENST00000376828	ILMN_1656670	chr6	monocyte	7.70×10-15 (p-value)	Zeller et al.	1
HLA-G	ENST00000360323						
HLA-G	ENST00000478519						
HLA-G	ENST00000478355						
HLA-G	ENST00000428701						
HLA-G	ENST00000376815						
HLA-G	ENST00000376818						
HLA-C	ENST00000470363	ILMN_2150787	chr6	monocyte	4.47×10-15 (p-value)	Zeller et al.	10
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						
BTN3A2	ENST00000524682	209846_s_at	chr6	blood	1.50×10-12 (p-value)	Dixon et al.	6
BTN3A2	ENST00000604202						
BTN3A2	ENST00000396934						
BTN3A2	ENST00000532294						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000377708						
BTN3A2	ENST00000527422						
BTN3A2	ENST00000531055						
BTN3A2	ENST00000396948						
BTN3A2	ENST00000356386						
BTN3A2	ENST00000396934	212613_at	chr6	blood	8.90×10-10 (p-value)	Dixon et al.	6
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000244519	204820_s_at	chr6	blood	4.50×10-12 (p-value)	Dixon et al.	6
BTN3A2	ENST00000396934						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000480110						
HLA-A	?	ENSG00000206503	chr6	brain	3.16×10-3 (q-value)	SeeQTL DB (Myers et al.)	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000443678	2	ENCP00000047794	OR2U1P
			OR2U1P
			OR2U1P
			OR2B4P
			OR2B4P
			OR2B4P
			OR2B4P
			OR2U1P

			OR2U1P OR2B4P OR2B4P OR2B4P OR2U1P
ENCE00000443629	1	ENCP00000047791	OR2H4P OR2H4P OR2H4P OR2H4P OR2H4P OR2H4P OR2H4P
		ENCP00000047763	GPX5 GPX5
ENCE00000443616	1	ENCP00000047762	GPX6 GPX6
ENCE00000443718	1	ENCP00000047788	OR2P1P OR2P1P XXbac-BCX196D17.5 XXbac-BCX196D17.5 OR2B3 OR2B3 XXbac-BCX196D17.5 OR2B3 OR2P1P OR2B3 XXbac-BCX196D17.5 OR2B3 OR2P1P OR2P1P OR2B3 OR2B3 OR2P1P OR2B3 XXbac-BCX196D17.5 AL662791.1 OR2P1P XXbac-BCX196D17.5 XXbac-BCX196D17.5
ENCE00000443832	1	ENCP00000047793	OR2G1P OR2G1P OR2G1P OR2G1P OR2G1P OR2G1P
		ENCP00000047802	OR12D3 OR12D3 OR12D3 OR12D3 OR12D3 OR12D3 OR12D3
ENCE00000443781	1	ENCP00000047777	TRIM27 TRIM27 TRIM27 TRIM27 TRIM27 TRIM27 TRIM27
		ENCP00000047786	OR2AD1P OR2AD1P OR2AD1P OR2AD1P OR2AD1P OR2AD1P OR2AD1P OR2AD1P
		ENCP00000047778	TRIM27 TRIM27 TRIM27 TRIM27

				TRIM27
				TRIM27
				TRIM27
			ENCP00000047851	HLA-H
				HLA-H
				HLA-H
				HLA-H
				HLA-H
				HLA-H
				HLA-H
				HLA-H
			ENCP00000047789	OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
			ENCP00000047824	GABBR1
				GABBR1
				GABBR1
				GABBR1
				GABBR1
				GABBR1
				GABBR1
ENCE00000443963		1	ENCP00000047854	DDX39BP1
				DDX39BP1
				DDX39BP1
				DDX39BP1
				DDX39BP1
				DDX39BP1
				DDX39BP1
				DDX39BP1
			ENCP00000047817	GABBR1
				GABBR1
				GABBR1
				GABBR1
				GABBR1
				GABBR1
				GABBR1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494418 (promoter)	1	embryonic stem cell (H1ESC)	H3K9ac, CTCF, Yy1, HDAC2, TAF1, H3K4me2, H3K4me3, DNase1, Sin3Ak20, TAF7, H3K27me3, PolII, Rad21, Egr1
		HSMMtube	H3K4me3, H3K4me2, CTCF, H3K27me3, H2AZ, DNase1
		blood (K562)	H3K4me3, DNase1, CTCF, Ini1, PolIII, Egr1, H3K27ac, Max, Rad21, H3K9ac, Cmyc, H2AZ, PolII, H3K4me2
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, H3K27me3, DNase1
		liver (HepG2)	DNase1, H3K27me3, Rad21, H2AZ, H3K4me2, CTCF, Cmyc, H3K4me3
		blood (GM12878)	H3K4me3, CTCF, H2AZ, DNase1, Egr1, Yy1, Rad21
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, CTCF
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K4me2, CTCF, H3K9ac, H3K4me3, H3K27me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K27ac, H3K4me3, PolII, Max, CTCF, DNase1, H3K9ac, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, H3K27me3, CTCF, Max, H3K4me2, H3K4me3
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000264210 (promoter flanking region)	4	embryonic stem cell (H1ESC)	H3K27me3, CTCF, PolII, Rad21, H3K9ac, Yy1, TAF1, H3K4me2, SP1, H3K4me3, DNase1
		HSMMtube	DNase1, H2AZ, H3K4me2
		blood (K562)	HEY1, H2AZ, Cfos, PolII, H3K4me2, CTCF, H3K4me3, Cmyc, H3K9ac, Ini1, Brg1, DNase1, PolIII, Egr1, H3K27ac, Max, ATF3, Rad21, E2F6

		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H3K4me3, H3K4me2, DNase1, H2AZ
		liver (HepG2)	PolII, DNase1, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, Cmyc, H3K4me3, H3K27me3
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		blood (GM12878)	H2AZ, DNase1, Yy1, ELF1, H3K4me3, H3K4me2, H3K9ac, PolII
		nervous (NH-A)	H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	CTCF, DNase1, H3K4me2, H3K4me3
		NHLF	DNase1, H3K4me3
		Osteobl	H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K9ac, H3K4me3, H3K36me3, H3K27ac, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, DNase1, H3K4me3
		cervix (HeLa-S3)	Max, H3K4me3, CTCF, DNase1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	Max, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000662255 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		blood (GM12878)	PolII, Cfos
		nervous (NH-A)	H3K27me3
		blood (DND-41)	H3K4me1
ENSR00000487496 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	H3K4me3, DNase1
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1
ENSR00000264215 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H4K5ac, CTCF, PolII, TAF7, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3
		HSMMtube	H3K9ac, H3K4me2, DNase1, H2AZ, H3K4me3, H3K27ac
		blood (K562)	Ini1, Brg1, PolIII, H3K27ac, ZBTB33, Max, ATF3, H3K9ac, ETS1, HEY1, H2AZ, TAF1, Jund, Cjun, PolII, GTF2B, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	PolII, TAF1, ELF1, H2AZ, ZBTB33, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1, Jund
		blood (GM12878)	PolII, H2AZ, ETS1, DNase1, CTCF, Yy1, ELF1, H3K4me3, ZBTB33, H3K27ac, H3K9ac
		lung (IMR90)	DNase1, H4K8ac, H3K27ac, H3K4me2, H4K5ac, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K4me3, H3K27ac, H3K9ac, H3K4me2
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	Cmyc, DNase1, Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII
		A549	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac
ENSR00001702905 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K4me2, H3K4me3
		blood (DND-41)	H3K4me2, H3K4me3
		blood (K562)	PolII, H3K4me3, H3K27ac, H3K9ac
		cervix (HeLa-S3)	H3K4me2, H3K4me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K4me3
		endothelium (HUVEC)	PolII
		liver (HepG2)	PolII
		A549	H3K36me3, H3K4me3
ENSR00001494427 (promoter flanking region)	1	endothelium (HUVEC)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00000662257 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, PolII
		blood (DND-41)	H3K27me3
		blood (K562)	DNase1, HEY1, PolII
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1

		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		liver (HepG2)	DNase1, FOSL2, PolII, H3K4me1, FOXA1, H3K27ac
		nervous (NH-A)	DNase1
		A549	DNase1
		skin (NHEK)	DNase1
ENSR00000487504 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K4me3, DNase1, H3K4me2, Yy1, H3K27me3
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	DNase1, PolIII, H3K27ac, Max, H3K9ac, H2AZ, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me2, DNase1
		liver (HepG2)	H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3
		blood (GM12878)	DNase1, H2AZ, Yy1, H3K4me3, H3K4me2
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		nervous (NH-A)	H3K4me3, H3K4me2
		skin (NHEK)	DNase1, H3K4me2
		NHLF	H3K4me3
		Osteobl	H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K4me2
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K4me3, H3K27me3, DNase1, H3K4me2
		A549	DNase1, H3K4me2, H3K4me3
ENSR00001494434 (CTCF binding site)	1	NHLF	CTCF
		embryonic stem cell (H1ESC)	PolII, Rad21, CTCF, ATF3, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K4me2
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	CTCF
		blood (GM12878)	CTCF
		lung (IMR90)	H3K4me3, CTCF
		skin (NHEK)	CTCF, DNase1
ENSR00000662259 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		Osteobl	H3K27me3
		breast (HMEC)	H3K4me3, H3K4me2
ENSR00000487523 (enhancer)	1	embryonic stem cell (H1ESC)	DNase1
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL645937.1	upstream gene variant	1970	ENST00000610295	?	?	1
AL662791.1	downstream gene variant	2606	ENST00000625416	?	?	1
GABBR1	downstream gene variant	3852	ENST00000355973	?	ENSP00000348248	1
HCG14	upstream gene variant, downstream gene variant	1429	ENST00000444986	?	?	3
HCG15	downstream gene variant	1878	ENST00000438190	?	?	2
HCG16	upstream gene variant, downstream gene variant	284	ENST00000445544	?	?	3
KRT18P1	downstream gene variant, upstream gene variant	1046	ENST00000452408	?	?	2
LINC01556	upstream gene variant, downstream gene variant	4280	ENST00000377186	?	?	2
MAS1L	downstream gene variant, upstream gene variant	1255	ENST00000377127	NM_052967.1	ENSP00000366331	3
MAS1LP1	downstream gene variant	101	ENST00000419416	?	?	1
OR10C1	downstream gene variant	2678	ENST00000622521	?	ENSP00000481429	2

OR10C1	downstream gene variant	2701	ENST00000444197	?	ENSP00000419119	2
OR10C1	downstream gene variant	2701	ENST00000623086	NM_013941.3	ENSP00000485032	2
OR12D3	downstream gene variant	457	ENST00000396806	NM_030959.2	ENSP00000380023	1
OR14J1	upstream gene variant	331	ENST00000377160	NM_030946.1	ENSP00000366365	1
OR2AD1P	downstream gene variant, upstream gene variant	2298	ENST00000441992	?	?	2
OR2B3	upstream gene variant	2549	ENST00000377173	NM_001005226.2	ENSP00000366378	1
OR2B4P	upstream gene variant, downstream gene variant	1001	ENST00000356293	?	?	2
OR2H1	downstream gene variant	68	ENST00000377136	?	ENSP00000366340	3
OR2H1	downstream gene variant	3310	ENST00000484371	?	?	1
OR2H1	downstream gene variant	68	ENST00000377133	NM_030883.3	ENSP00000366337	3
OR2H1	downstream gene variant	1676	ENST00000473369	?	?	2
OR2H1	downstream gene variant	1676	ENST00000484554	?	?	2
OR2H1	downstream gene variant	1663	ENST00000396792	?	ENSP00000380010	2
OR2H1	downstream gene variant	68	ENST00000377132	?	ENSP00000366336	3
OR2H4P	upstream gene variant	126	ENST00000415595	?	?	2
OR2I1P	upstream gene variant, downstream gene variant	1442	ENST00000453522	?	?	2
OR2J2	downstream gene variant	4352	ENST00000377167	NM_030905.2	ENSP00000366372	1
OR2J3	upstream gene variant	4580	ENST00000377169	NM_001005216.3	ENSP00000366374	1
OR2J4P	upstream gene variant, downstream gene variant	893	ENST00000453647	?	?	3
OR2N1P	upstream gene variant	2604	ENST00000421799	?	?	1
OR2P1P	upstream gene variant	4325	ENST00000413771	?	?	1
OR2U2P	upstream gene variant	3180	ENST00000465951	?	?	1
OR2W1	downstream gene variant, upstream gene variant	553	ENST00000377175	NM_030903.3	ENSP00000366380	3
RN7SL471P	downstream gene variant	2542	ENST00000580476	?	?	1
RNU6-930P	upstream gene variant, downstream gene variant	126	ENST00000390938	?	?	3
RPL13P	upstream gene variant, downstream gene variant	1197	ENST00000396818	?	?	9
RPS17P1	downstream gene variant, upstream gene variant	788	ENST00000396783	?	?	3
SAR1P1	upstream gene variant	1473	ENST00000455287	?	?	1
TRIM27	downstream gene variant, upstream gene variant	140	ENST00000467742	?	?	3
TRIM27	downstream gene variant, upstream gene variant	1808	ENST00000377194	?	ENSP00000366399	3
TRIM27	downstream gene variant, upstream gene variant	1807	ENST00000377199	NM_006510.4	ENSP00000366404	3
TRIM27	downstream gene variant, upstream gene variant	1883	ENST00000481474	?	?	3
TRIM27	downstream gene variant, upstream gene variant	1837	ENST00000498117	?	?	3
TRIM27	downstream gene variant, upstream gene variant	481	ENST00000414543	?	ENSP00000400058	3
UBD	downstream gene variant	3738	ENST00000377050	NM_006398.3	ENSP00000366249	1
UBDP1	downstream gene variant	200	ENST00000457888	?	?	1
XXbac-BCX196D17.5	upstream gene variant, downstream gene	1976	ENST00000419702	?	?	2

	variant					
XXbac-BPG13B8.10	upstream gene variant	2243	ENST00000436804	?	?	1
XXbac-BPG13B8.11	upstream gene variant	2523	ENST00000618350	?	?	2
XXbac-BPG258E24.10	upstream gene variant	2485	ENST00000623946	?	?	1
XXbac-BPG258E24.10	upstream gene variant, downstream gene variant	400	ENST00000610326	?	?	4
XXbac-BPG258E24.10	upstream gene variant, downstream gene variant	445	ENST00000623334	?	?	3
XXbac-BPG308J9.3	upstream gene variant, downstream gene variant	1805	ENST00000606648	?	?	2
XXbac-BPG308J9.3	upstream gene variant	1778	ENST00000441381	?	?	1
XXbac-BPG308K3.5	downstream gene variant, upstream gene variant	958	ENST00000457253	?	?	8
XXbac-BPG308K3.6	downstream gene variant, upstream gene variant	46	ENST00000606624	?	?	6
XXbac-BPG308K3.6	downstream gene variant, upstream gene variant	157	ENST00000606727	?	?	9
XXbac-BPG308K3.6	downstream gene variant, upstream gene variant	484	ENST00000440244	?	?	8
ZNF311	downstream gene variant, upstream gene variant	1228	ENST00000377179	NM_001010877.2	ENSP00000366384	3
ZNF311	downstream gene variant, upstream gene variant	1172	ENST00000483450	?	?	3
ZNF90P2	downstream gene variant, upstream gene variant	804	ENST00000419323	?	?	6

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
UBD	ENST00000377050	NM_006398.3	ENSP00000366249	Y	taC/taT	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCG14	ENST00000444986	?	?	1
HCG15	ENST00000438190	?	?	1
OR11A1	ENST00000377149	?	ENSP00000366354	3
OR5V1	ENST00000377154	?	ENSP00000366359	4
TRIM27	ENST00000414543	?	ENSP00000400058	1
TRIM27	ENST00000377199	NM_006510.4	ENSP00000366404	5
TRIM27	ENST00000481474	?	?	5
TRIM27	ENST00000377194	?	ENSP00000366399	5
TRIM27	ENST00000496091	?	?	3
TRIM27	ENST00000498117	?	?	3
UBDP1	ENST00000457888	?	?	2
XXbac-BCX196D17.5	ENST00000419702	?	?	1
XXbac-BPG13B8.10	ENST00000436804	?	?	1
XXbac-BPG258E24.10	ENST00000623334	?	?	6
XXbac-BPG258E24.10	ENST00000623946	?	?	6
XXbac-BPG308J9.3	ENST00000606648	?	?	10
XXbac-BPG308J9.3	ENST00000441381	?	?	6

XXbac-BPG308K3.5	ENST00000457253	?	?	1
XXbac-BPG308K3.6	ENST00000440244	?	?	1
XXbac-BPG308K3.6	ENST00000606624	?	?	1
XXbac-BPG308K3.6	ENST00000606727	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GABBR1	ENST00000355973	?	ENSP00000348248	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RPL13P	ENST00000396818	?	1
SAR1P1	ENST00000455287	?	1
TRIM27	ENST00000481474	?	1
XXbac-BPG308K3.6	ENST00000440244	?	2
XXbac-BPG308K3.6	ENST00000606727	?	1
ZNF90P2	ENST00000419323	?	4

Locus 200 (rs550513)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,905,130-31,928,306
block size	23,177 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.055 [-3.331 – 5.881]	gene(s) hit or close-by	C2, C2-AS1, CFB, MIR1236, NELFE, SKIV2L, XXbac-BPG116M5.17
phastCons	μ = 0.127 [0 – 1]	eQTL gene(s)	NELFE, NEU1, PRRT1, SKIV2L
GERP++	μ = -0.978 [-10.3 – 5.14]	potentially regulated gene(s)	CFB, CFB, CFB, CFB, CFB, CFB, CFB, CFB, SKIV2L, SKIV2L, SKIV2L, SKIV2L, SKIV2L, SKIV2L
CADD score	μ = 5.009 [0.293 – 22.3]	disease gene(s)	SKIV2L, NEU1, , , , , , , C2, CFB, , , , , ,

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Age-related macular degeneration	$<2.00\times10^{-10}$	GWAS Catalog	23577725		1
Age-related macular degeneration (extreme sampling)	$<3.00\times10^{-7}$	GWAS Catalog	23577725		1
Age-related macular degeneration	$<9.00\times10^{-17}$	GWAS Catalog	22694956		1
Macular degeneration	2.70×10^{-18}	dbGaP	pha002890	dbGaP	5
Macular degeneration	2.90×10^{-19}	dbGaP	pha002869	dbGaP	5
Macular degeneration	8.34×10^{-5}	dbGaP	pha002856	dbGaP	1
Macular degeneration	1.38×10^{-5}	dbGaP	pha000001	dbGaP	1

Variant annotation

trait	type	source DB	source entry/link		Variant(s)
Age-related macular degeneration 14	protective	ClinVar	RCV000012914.4	ClinVar	1
?	HGMD curated	HGMD	CM093386	HGMD	1
MACULAR DEGENERATION, AGE-RELATED, 14, REDUCED RISK OF	OMIM curated	OMIM	MIM:613927	OMIM	1

Disease gene annotation

gene	trait	source DB	source entry/link	
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM
NEU1	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602	OMIM
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
NEU1	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM

CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602	OMIM
	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602	OMIM
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064	orphanet
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
NEU1	Juvenile sialidosis type 2	OrphaNet	OrphaNet:93399	orphanet
NEU1	Sialidosis type 1	OrphaNet	OrphaNet:812	orphanet
NEU1	Congenital sialidosis type 2	OrphaNet	OrphaNet:93400	orphanet
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SKIV2L	missense variant	ENST00000375394	NM_006929.4	ENSP00000364543	R/Q	cGg/cAg	?	?	1
SKIV2L	missense variant	ENST00000461073	?	ENSP00000419905	R/Q	cGg/cAg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SKIV2L	?	ENSG00000204351	pancreas	5.47×10-11 (p-value)	GTEx Portal V6	13
NELFE	ENST00000492185	ILMN_1765532	adipocyte	1.04×10-7 (p-value)	MuTHER consortium	6
NELFE	ENST00000488426		blood	6.64×10-5 (p-value)	MuTHER consortium	6
NELFE	ENST00000481121					
NELFE	ENST00000375429					
SKIV2L	ENST00000484835	ILMN_1666512	skin	5.43×10-6 (p-value)	MuTHER consortium	6
SKIV2L	ENST00000375394		blood	2.47×10-6 (p-value)	MuTHER consortium	6
SKIV2L	ENST00000474839		adipocyte	2.04×10-6 (p-value)	MuTHER consortium	6
SKIV2L	ENST00000470453					

SKIV2L	ENST00000465703					
SKIV2L	ENST00000491994					
SKIV2L	ENST00000471818					
SKIV2L	ENST00000483553					
SKIV2L	ENST00000485349					
SKIV2L	?	ENSG00000204351	muscularis mucosae	1.86×10-27 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	lung	1.00×10-21 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	atrial appendage	1.13×10-9 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	transformed fibroblasts	9.54×10-13 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	blood	2.49×10-16 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	breast	2.88×10-10 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	tibial artery	3.59×10-35 (p-value)	GTEx Portal V6	13
NELFE	?	ENSG00000204356	tibial artery	1.15×10-8 (p-value)	GTEx Portal V6	13
SKIV2L	ENST00000484835	ILMN_1666512	monocyte	1.40×10-8 (p-value)	Fairfax et al.	10
SKIV2L	ENST00000375394		b-cell	4.53×10-10 (p-value)	Fairfax et al.	10
SKIV2L	ENST00000474839					
SKIV2L	ENST00000470453					
SKIV2L	ENST00000465703					
SKIV2L	ENST00000491994					
SKIV2L	ENST00000471818					
SKIV2L	ENST00000483553					
SKIV2L	ENST00000485349					
NEU1	ENST00000495807	ILMN_1763144	monocyte	7.88×10-4 (p-value)	Fairfax et al.	9
NEU1	ENST00000375631					
NEU1	ENST00000480384					
NEU1	ENST00000491768					
SKIV2L	?	ENSG00000204351	thyroid	1.97×10-16 (p-value)	GTEx Portal V6	13
PRRT1	?	ENSG00000204314	thyroid	7.19×10-6 (p-value)	GTEx Portal V6	12
NELFE	?	ENSG00000204356	skeletal muscle	1.10×10-10 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	EBV lymphocytes	3.42×10-9 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	transverse colon	5.98×10-12 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	sun exposed skin	1.82×10-16 (p-value)	GTEx Portal V6	13
NELFE	?	ENSG00000204356	sun exposed skin	4.14×10-14 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	aorta	1.13×10-20 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	left ventricle	3.58×10-8 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	subcutaneous adipocytes	1.27×10-25 (p-value)	GTEx Portal V6	13
NELFE	?	ENSG00000204356	subcutaneous adipocytes	1.38×10-11 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	stomach	5.07×10-12 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	visceral adipocytes	5.78×10-10 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	cortex	6.27×10-7 (p-value)	GTEx Portal V6	13

SKIV2L	?	ENSG00000204351	sigmoid colon	1.41×10-11 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	coronary artery	1.46×10-9 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	tibial nerve	6.52×10-18 (p-value)	GTEx Portal V6	13
NELFE	?	ENSG00000204356	tibial nerve	1.87×10-8 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	esophagus mucosa	5.48×10-12 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	gastroesophageal junction	1.96×10-10 (p-value)	GTEx Portal V6	13
SKIV2L	?	ENSG00000204351	testis	1.51×10-7 (p-value)	GTEx Portal V6	13
NELFE	ENST00000492185	ILMN_1765532	monocyte	3.33×10-15 (p-value)	Zeller et al.	2
NELFE	ENST00000488426					
NELFE	ENST00000481121					
NELFE	ENST00000375429					
SKIV2L	ENST00000484835	ILMN_1666512	monocyte	5.72×10-13 (p-value)	Zeller et al.	2
SKIV2L	ENST00000375394					
SKIV2L	ENST00000474839					
SKIV2L	ENST00000470453					
SKIV2L	ENST00000465703					
SKIV2L	ENST00000491994					
SKIV2L	ENST00000471818					
SKIV2L	ENST00000483553					
SKIV2L	ENST00000485349					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000048142		CFB
		CFB
		CFB
		CFB
		CFB
		CFB
		CFB
ENCP00000048147		SKIV2L
		SKIV2L
		SKIV2L
		SKIV2L
		SKIV2L

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000264486 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1, Cmyc, PolII, Max, Jund, H3K27ac, H3K4me1
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		liver (HepG2)	PolII, H3K79me2, H3K4me1, H3K4me2, H3K36me3
		nervous (NH-A)	DNase1
		blood (K562)	H3K4me1, DNase1, PolII, Cjun, Max
		skin (NHDF-AD)	DNase1
ENSR00000025316 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, DNase1
		HSMMtube	H3K36me3, CTCF
		blood (K562)	Rad21, CTCF, H3K36me3
		skin (NHDF-AD)	H3K36me3, CTCF

muscle (HSMM)	CTCF
liver (HepG2)	PolII, Rad21, CTCF, H3K36me3, DNase1
lung (IMR90)	H3K36me3, CTCF
blood (GM12878)	CTCF, H3K36me3
nervous (NH-A)	CTCF, H3K36me3
skin (NHEK)	CTCF, H3K36me3
NHLF	CTCF, H3K36me3
Osteobl	H3K36me3, CTCF
blood (DND-41)	CTCF, H3K36me3
breast (HMEC)	H3K36me3, CTCF
cervix (HeLa-S3)	CTCF, H3K36me3, DNase1
monocytes (Monocytes-CD14+)	H3K36me3, CTCF
endothelium (HUVEC)	H3K36me3, CTCF, DNase1
A549	H3K36me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C2	upstream gene variant, downstream gene variant	512	ENST00000468407	?	?	6
C2	downstream gene variant	2295	ENST00000418949	NM_001282459.1	ENSP00000406190	7
C2	downstream gene variant	3071	ENST00000494905	?	ENSP00000419048	5
C2	downstream gene variant	1363	ENST00000484636	?	ENSP00000420305	7
C2	downstream gene variant	3058	ENST00000452202	?	ENSP00000406121	5
C2	downstream gene variant	92	ENST00000411571	?	ENSP00000388727	6
C2	downstream gene variant	3108	ENST00000447952	?	ENSP00000391354	5
C2	downstream gene variant	3502	ENST00000486124	?	?	1
C2	downstream gene variant	3502	ENST00000299367	NM_001282458.1, NM_000063.5	ENSP00000299367	1
C2	downstream gene variant	3550	ENST00000383177	?	ENSP00000372664	1
C2	downstream gene variant	3783	ENST00000482060	?	ENSP00000418332	1
C2	downstream gene variant	3503	ENST00000442278	NM_001145903.2	ENSP00000395683	1
C2	downstream gene variant	3653	ENST00000469372	NM_001282457.1	ENSP00000418923	1
C2	downstream gene variant	4321	ENST00000497706	?	ENSP00000417482	1
C2	downstream gene variant	3503	ENST00000452323	NM_001178063.2	ENSP00000392322	1
C2-AS1	upstream gene variant	1437	ENST00000630806	?	?	1
CFB	downstream gene variant	2883	ENST00000482886	?	?	2
CFB	upstream gene variant, downstream gene variant	56	ENST00000425368	NM_001710.5	ENSP00000416561	3
CFB	upstream gene variant, downstream gene variant	1495	ENST00000452035	?	?	3
CFB	upstream gene variant, downstream gene variant	2542	ENST00000497841	?	?	3
CFB	upstream gene variant, downstream gene variant	2054	ENST00000472581	?	?	2
CFB	upstream gene variant, downstream gene variant	2033	ENST00000475617	?	ENSP00000420090	3
CFB	upstream gene variant, downstream gene variant	1205	ENST00000460718	?	ENSP00000417793	4
CFB	upstream gene variant, downstream gene variant	56	ENST00000482312	?	?	3
CFB	upstream gene variant, downstream gene variant	557	ENST00000467360	?	?	3
CFB	upstream gene variant, downstream gene variant	2589	ENST00000461483	?	?	3
CFB	upstream gene variant, downstream gene variant	56	ENST00000498317	?	ENSP00000418833	3

CFB	downstream gene variant	87	ENST00000483004 ?	ENSP00000419887	2
CFB	downstream gene variant	2542	ENST00000467150 ?	?	2
CFB	downstream gene variant	2542	ENST00000465750 ?	?	2
MIR1236	downstream gene variant, upstream gene variant	163	ENST00000408340 ?	?	4
NELFE	downstream gene variant, upstream gene variant	87	ENST00000492185 ?	?	3
NELFE	downstream gene variant, upstream gene variant	88	ENST00000488426 ?	?	3
NELFE	downstream gene variant, upstream gene variant	1419	ENST00000375429 NM_002904.5	ENSP00000364578	2
NELFE	downstream gene variant, upstream gene variant	848	ENST00000441998 ?	ENSP00000397914	4
NELFE	downstream gene variant, upstream gene variant	1479	ENST00000436289 ?	ENSP00000414029	3
NELFE	downstream gene variant, upstream gene variant	1561	ENST00000492539 ?	ENSP00000419183	3
NELFE	downstream gene variant, upstream gene variant	89	ENST00000375425 ?	ENSP00000364574	3
NELFE	downstream gene variant, upstream gene variant	88	ENST00000481121 ?	?	3
NELFE	downstream gene variant, upstream gene variant	1575	ENST00000494956 ?	ENSP00000420205	3
NELFE	downstream gene variant, upstream gene variant	89	ENST00000444811 ?	ENSP00000388400	3
NELFE	downstream gene variant, upstream gene variant	297	ENST00000625905 ?	ENSP00000486884	3
NELFE	downstream gene variant, upstream gene variant	1394	ENST00000454913 ?	ENSP00000409389	3
NELFE	downstream gene variant, upstream gene variant	1564	ENST00000491139 ?	?	3
NELFE	downstream gene variant, upstream gene variant	1670	ENST00000426722 ?	ENSP00000394340	3
SKIV2L	upstream gene variant, downstream gene variant	571	ENST00000492900 ?	?	2
SKIV2L	upstream gene variant	2068	ENST00000474839 ?	ENSP00000420470	1
SKIV2L	upstream gene variant	2014	ENST00000488648 ?	?	1
SKIV2L	upstream gene variant	2054	ENST00000461073 ?	ENSP00000419905	1
SKIV2L	upstream gene variant	2090	ENST00000628157 ?	ENSP00000485707	1
SKIV2L	upstream gene variant	968	ENST00000466290 ?	?	2
SKIV2L	upstream gene variant	1977	ENST00000375394 NM_006929.4	ENSP00000364543	1
SKIV2L	upstream gene variant	2038	ENST00000465703 ?	?	1
XXbac-BPG116M5.17	downstream gene variant	110	ENST00000477310 ?	ENSP00000418996	2
XXbac-BPG116M5.17	downstream gene variant	92	ENST00000456570 ?	ENSP00000410815	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
C2	ENST00000469372	NM_001282457.1	ENSP00000418923	A	gcA/gcG	1
C2	ENST00000442278	NM_001145903.2	ENSP00000395683	A	gcA/gcG	1
C2	ENST00000485690	?	ENSP00000417790	A	gcA/gcG	1
C2	ENST00000497706	?	ENSP00000417482	A	gcA/gcG	1
C2	ENST00000452323	NM_001178063.2	ENSP00000392322	A	gcA/gcG	1

C2	ENST00000383177	?	ENSP00000372664	A	gcA/gcG	1
C2	ENST00000299367	NM_001282458.1, NM_000063.5	ENSP00000299367	A	gcA/gcG	1
XXbac-BPG116M5.17	ENST00000456570	?	ENSP00000410815	A	gcA/gcG	1
XXbac-BPG116M5.17	ENST00000477310	?	ENSP00000418996	A	gcA/gcG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C2	ENST00000482060	?	ENSP00000418332	8
C2	ENST00000486124	?	?	8
C2	ENST00000469372	NM_001282457.1	ENSP00000418923	8
C2	ENST00000442278	NM_001145903.2	ENSP00000395683	8
C2	ENST00000485690	?	ENSP00000417790	8
C2	ENST00000497706	?	ENSP00000417482	8
C2	ENST00000452323	NM_001178063.2	ENSP00000392322	8
C2	ENST00000383177	?	ENSP00000372664	8
C2	ENST00000299367	NM_001282458.1, NM_000063.5	ENSP00000299367	8
C2-AS1	ENST00000630806	?	?	7
CFB	ENST00000461483	?	?	1
CFB	ENST00000483004	?	ENSP00000419887	1
CFB	ENST00000425368	NM_001710.5	ENSP00000416561	1
CFB	ENST00000452035	?	?	1
CFB	ENST00000497841	?	?	1
CFB	ENST00000465750	?	?	1
NELFE	ENST00000481121	?	?	2
NELFE	ENST00000488426	?	?	2
NELFE	ENST00000375429	NM_002904.5	ENSP00000364578	2
NELFE	ENST00000492185	?	?	2
NELFE	ENST00000491139	?	?	1
NELFE	ENST00000436289	?	ENSP00000414029	1
NELFE	ENST00000444811	?	ENSP00000388400	2
NELFE	ENST00000375425	?	ENSP00000364574	2
NELFE	ENST00000426722	?	ENSP00000394340	1
NELFE	ENST00000492539	?	ENSP00000419183	1
NELFE	ENST00000494956	?	ENSP00000420205	1
NELFE	ENST00000441998	?	ENSP00000397914	1
NELFE	ENST00000454913	?	ENSP00000409389	1
SKIV2L	ENST00000628157	?	ENSP00000485707	1
SKIV2L	ENST00000474839	?	ENSP00000420470	1
XXbac-BPG116M5.17	ENST00000456570	?	ENSP00000410815	9
XXbac-BPG116M5.17	ENST00000477310	?	ENSP00000418996	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

C2	ENST00000482060	?	ENSP00000418332	1
C2	ENST00000411571	?	ENSP00000388727	1
NELFE	ENST00000375429	NM_002904.5	ENSP00000364578	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C2	ENST00000486124	?	1
CFB	ENST00000467150	?	1
CFB	ENST00000482886	?	1
SKIV2L	ENST00000488648	?	1
SKIV2L	ENST00000465703	?	1

Locus 201 (rs1336900)

SNiPAcard

Block annotations

Block info

genomic range	chr1:150,614,869-150,889,489
block size	274,621 bp
variant count	178 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.491 [-5.187 – 2.168]	gene(s) hit or close-by	ARNT, CTSK, CTSS, GOLPH3L, HORMAD1, RNU6-1042P, RP11-363I22.3
phastCons	μ = 0.065 [0 – 0.984]	eQTL gene(s)	ADAMTSL4, ANXA9, ARNT, CDC42SE1, CERS2, CTSK, CTSS, GOLPH3L, HORMAD1, RP11-316M1.12, RP11-54A4.2, SETDB1
GERP++	μ = -0.192 [-8.55 – 3.55]	potentially regulated gene(s)	ADAMTSL4, ARNT, BNIPL, CTSK, CTSS, MCL1, PIP5K1A, PRPF3, RP11-316M1.3, RP11-54A4.2, SEMA6C, SETDB1, TARS2
CADD score	μ = 4.046 [0.008 – 22.4]	disease gene(s)	CTSK, ADAMTSL4, PRPF3, TARS2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Fat body mass	<4.00×10-8	GWAS Catalog	24064335	1

Disease gene annotation

gene	trait	source DB	source entry/link
CTSK	PYCNODYSOSTOSIS	OMIM	MIM:265800
ADAMTSL4	ECTOPIA LENTIS ET PUPILLAE	OMIM	MIM:225200
PRPF3	RETINITIS PIGMENTOSA 18	OMIM	MIM:601414
TARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 21	OMIM	MIM:615918
CTSK	PYCNODYSOSTOSIS	DECIPHER	MIM:265800
CTSK	PYCNODYSOSTOSIS	OrphaNet	OrphaNet:763
ADAMTSL4	isolated ectopia lentis (EL)	OrphaNet	OrphaNet:1885
PRPF3	Retinitis pigmentosa	OrphaNet	OrphaNet:791

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ARNT	splice donor variant	ENST00000504358	?	ENSP00000425363					1
CTSS	missense variant	ENST00000368985	NM_004079.4	ENSP00000357981	W/R	Tgg/Cgg	?	?	1
HORMAD1	missense variant	ENST00000361824	NM_032132.4	ENSP00000355167	I/T	aTa/aCa	?	?	1
HORMAD1	missense variant	ENST00000322343	NM_001199829.1	ENSP00000326489	I/T	aTa/aCa	?	?	1
HORMAD1	missense variant	ENST00000368995	?	ENSP00000357991	I/T	aTa/aCa	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CERS2	ENST00000368954	ILMN_2400500	blood	5.07×10-7 (p-value)	MuTHER consortium	36
CERS2	ENST00000271688					
CERS2	ENST00000345896					
CERS2	ENST00000560793					
CERS2	ENST00000460664					
CTSK	ENST00000271651	ILMN_1758895	blood	3.68×10-19 (p-value)	MuTHER consortium	36
ANXA9	ENST00000368947	ILMN_1755721	skin	2.68×10-5 (p-value)	MuTHER consortium	21
HORMAD1	ENST00000361824	ILMN_1769849	skin	8.78×10-42 (p-value)	MuTHER consortium	36
HORMAD1	ENST00000470397					
HORMAD1	ENST00000486497					
HORMAD1	ENST00000322343					
HORMAD1	ENST00000368995					
GOLPH3L	ENST00000271732	ILMN_1706764	skin	3.09×10-14 (p-value)	MuTHER consortium	36
CTSS	ENST00000368985	ILMN_1743032	skin	6.73×10-6 (p-value)	MuTHER consortium	36
			blood	2.03×10-40 (p-value)	MuTHER consortium	36
CERS2	ENST00000368954	ILMN_1726108	blood	2.52×10-6 (p-value)	MuTHER consortium	35
CERS2	ENST00000271688					
CERS2	ENST00000345896					
CERS2	ENST00000560793					
CERS2	ENST00000460664					
RP11-316M1.12	?	ENSG00000259357	atrial appendage	8.27×10-32 (p-value)	GTEx Portal V6	170
ARNT	?	ENSG00000143437	atrial appendage	4.23×10-6 (p-value)	GTEx Portal V6	34
CTSS	?	ENSG00000163131	atrial appendage	5.93×10-6 (p-value)	GTEx Portal V6	1
RP11-316M1.12	?	ENSG00000259357	transformed fibroblasts	1.58×10-57 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	transformed fibroblasts	1.72×10-18 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	transformed fibroblasts	1.70×10-43 (p-value)	GTEx Portal V6	170
RP11-316M1.12	?	ENSG00000259357	blood	1.06×10-37 (p-value)	GTEx Portal V6	170
CTSK	?	ENSG00000143387	blood	3.85×10-7 (p-value)	GTEx Portal V6	124
RP11-316M1.12	?	ENSG00000259357	breast	4.78×10-21 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	breast	1.00×10-13 (p-value)	GTEx Portal V6	170
RP11-54A4.2	?	ENSG00000237781	breast	4.46×10-6 (p-value)	GTEx Portal V6	1
CERS2	ENST00000368954	ILMN_1726108	b-cell	6.65×10-6 (p-value)	Fairfax et al.	13
CERS2	ENST00000271688					
CERS2	ENST00000345896					
CERS2	ENST00000560793					
CERS2	ENST00000460664					
ARNT	ENST00000358595	ILMN_1762582	monocyte	9.14×10-15 (p-value)	Fairfax et al.	12
ARNT	ENST00000354396		b-cell	3.32×10-7 (p-value)	Fairfax et al.	12
ARNT	ENST00000471844					
CTSS	ENST00000368985	ILMN_1743032	monocyte	9.38×10-17 (p-value)	Fairfax et al.	14
			b-cell	4.91×10-9 (p-value)	Fairfax et al.	14
CTSK	ENST00000271651	ILMN_1758895	b-cell	2.23×10-5 (p-value)	Fairfax et al.	14

CERS2	ENST00000368954	ILMN_2400500	b-cell	3.18×10-9 (p-value)	Fairfax et al.	4
CERS2	ENST00000271688					
CERS2	ENST00000345896					
CERS2	ENST00000560793					
CERS2	ENST00000460664					
RP11-316M1.12	?	ENSG00000259357	skeletal muscle	7.41×10-42 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	skeletal muscle	1.84×10-12 (p-value)	GTEx Portal V6	169
HORMAD1	?	ENSG00000143452	skeletal muscle	2.52×10-10 (p-value)	GTEx Portal V6	169
RP11-316M1.12	?	ENSG00000259357	putamen	5.54×10-10 (p-value)	GTEx Portal V6	169
RP11-316M1.12	?	ENSG00000259357	EBV lymphocytes	1.09×10-11 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	EBV lymphocytes	5.94×10-12 (p-value)	GTEx Portal V6	169
CTSS	?	ENSG00000163131	blood	1.14×10-5 (q-value)	SeeQTL DB (HapMap)	37
RP11-316M1.12	?	ENSG00000259357	ovary	3.91×10-10 (p-value)	GTEx Portal V6	170
GOLPH3L	?	ENSG00000143457	sun exposed skin	6.62×10-50 (p-value)	GTEx Portal V6	170
RP11-316M1.12	?	ENSG00000259357	sun exposed skin	6.85×10-29 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	sun exposed skin	2.66×10-10 (p-value)	GTEx Portal V6	168
ANXA9	?	ENSG00000143412	sun exposed skin	5.18×10-11 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	sun exposed skin	3.17×10-70 (p-value)	GTEx Portal V6	170
GOLPH3L	?	ENSG00000143457	unexposed skin	5.37×10-24 (p-value)	GTEx Portal V6	170
RP11-316M1.12	?	ENSG00000259357	unexposed skin	2.15×10-28 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	unexposed skin	6.26×10-44 (p-value)	GTEx Portal V6	170
ANXA9	?	ENSG00000143412	unexposed skin	1.34×10-6 (p-value)	GTEx Portal V6	35
ARNT	?	ENSG00000143437	aorta	2.03×10-9 (p-value)	GTEx Portal V6	168
RP11-316M1.12	?	ENSG00000259357	aorta	2.87×10-34 (p-value)	GTEx Portal V6	170
GOLPH3L	?	ENSG00000143457	aorta	2.88×10-5 (p-value)	GTEx Portal V6	1
RP11-316M1.12	?	ENSG00000259357	left ventricle	3.46×10-32 (p-value)	GTEx Portal V6	170
CERS2	?	ENSG00000143418	left ventricle	2.76×10-7 (p-value)	GTEx Portal V6	64
RP11-316M1.12	?	ENSG00000259357	subcutaneous adipocytes	1.59×10-59 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	subcutaneous adipocytes	1.77×10-9 (p-value)	GTEx Portal V6	169
HORMAD1	?	ENSG00000143452	subcutaneous adipocytes	4.74×10-43 (p-value)	GTEx Portal V6	170
RP11-316M1.12	?	ENSG00000259357	caudate basal ganglia	3.10×10-12 (p-value)	GTEx Portal V6	169
RP11-316M1.12	?	ENSG00000259357	stomach	2.04×10-13 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	stomach	1.52×10-8 (p-value)	GTEx Portal V6	167
RP11-316M1.12	?	ENSG00000259357	visceral adipocytes	3.60×10-41 (p-value)	GTEx Portal V6	170
ARNT	?	ENSG00000143437	visceral adipocytes	1.31×10-5 (p-value)	GTEx Portal V6	1
RP11-316M1.12	?	ENSG00000259357	cortex	6.79×10-10 (p-value)	GTEx Portal V6	169
GOLPH3L	?	ENSG00000143457	tibial nerve	2.31×10-51 (p-value)	GTEx Portal V6	170
SETDB1	?	ENSG00000143379	tibial nerve	8.37×10-7 (p-value)	GTEx Portal V6	162

RP11-316M1.12	?	ENSG00000259357	tibial nerve	2.76×10-52 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	tibial nerve	3.08×10-67 (p-value)	GTEx Portal V6	170
ARNT	?	ENSG00000143437	tibial nerve	3.46×10-7 (p-value)	GTEx Portal V6	105
GOLPH3L	?	ENSG00000143457	esophagus mucosa	9.80×10-14 (p-value)	GTEx Portal V6	170
RP11-54A4.2	?	ENSG00000237781	esophagus mucosa	4.32×10-6 (p-value)	GTEx Portal V6	122
RP11-316M1.12	?	ENSG00000259357	esophagus mucosa	1.04×10-34 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	esophagus mucosa	1.29×10-13 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	esophagus mucosa	7.73×10-67 (p-value)	GTEx Portal V6	170
ADAMTSL4	?	ENSG00000143382	esophagus mucosa	1.61×10-5 (p-value)	GTEx Portal V6	2
RP11-316M1.12	?	ENSG00000259357	anterior cingulate cortex	2.44×10-7 (p-value)	GTEx Portal V6	123
RP11-316M1.12	?	ENSG00000259357	pituitary	1.73×10-19 (p-value)	GTEx Portal V6	170
GOLPH3L	?	ENSG00000143457	testis	3.75×10-10 (p-value)	GTEx Portal V6	170
RP11-316M1.12	?	ENSG00000259357	testis	4.69×10-22 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	testis	3.85×10-10 (p-value)	GTEx Portal V6	169
CTSK	?	ENSG00000143387	testis	6.48×10-6 (p-value)	GTEx Portal V6	40
RP11-316M1.12	?	ENSG00000259357	hypothalamus	7.87×10-8 (p-value)	GTEx Portal V6	163
RP11-316M1.12	?	ENSG00000259357	uterus	2.47×10-10 (p-value)	GTEx Portal V6	168
RP11-316M1.12	?	ENSG00000259357	pancreas	1.93×10-27 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	pancreas	4.42×10-11 (p-value)	GTEx Portal V6	170
RP11-316M1.12	?	ENSG00000259357	muscularis mucosae	6.93×10-47 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	muscularis mucosae	4.18×10-6 (p-value)	GTEx Portal V6	38
ARNT	?	ENSG00000143437	muscularis mucosae	1.25×10-5 (p-value)	GTEx Portal V6	2
RP11-316M1.12	?	ENSG00000259357	lung	1.52×10-54 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	lung	2.55×10-8 (p-value)	GTEx Portal V6	170
GOLPH3L	?	ENSG00000143457	tibial artery	1.21×10-7 (p-value)	GTEx Portal V6	168
RP11-316M1.12	?	ENSG00000259357	tibial artery	1.71×10-53 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	tibial artery	3.29×10-8 (p-value)	GTEx Portal V6	168
ARNT	?	ENSG00000143437	tibial artery	9.81×10-7 (p-value)	GTEx Portal V6	36
ARNT	?	ENSG00000143437	thyroid	5.51×10-10 (p-value)	GTEx Portal V6	170
RP11-316M1.12	?	ENSG00000259357	thyroid	8.88×10-55 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	thyroid	3.87×10-23 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	thyroid	3.07×10-14 (p-value)	GTEx Portal V6	170
CERS2	?	ENSG00000143418	thyroid	1.58×10-6 (p-value)	GTEx Portal V6	38
RP11-316M1.12	?	ENSG00000259357	prostate	1.21×10-11 (p-value)	GTEx Portal V6	169
RP11-316M1.12	?	ENSG00000259357	transverse colon	3.07×10-29 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	transverse colon	2.09×10-6 (p-value)	GTEx Portal V6	144

RP11-316M1.12	?	ENSG00000259357	liver	5.89×10-13 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	liver	2.48×10-6 (p-value)	GTEx Portal V6	30
RP11-316M1.12	?	ENSG00000259357	terminal ileum	2.92×10-7 (p-value)	GTEx Portal V6	163
RP11-54A4.2	?	ENSG00000237781	spleen	2.08×10-7 (p-value)	GTEx Portal V6	165
RP11-316M1.12	?	ENSG00000259357	spleen	1.30×10-19 (p-value)	GTEx Portal V6	170
RP11-316M1.12	?	ENSG00000259357	nucleus accumbens	5.65×10-16 (p-value)	GTEx Portal V6	169
RP11-316M1.12	?	ENSG00000259357	sigmoid colon	2.26×10-22 (p-value)	GTEx Portal V6	170
RP11-316M1.12	?	ENSG00000259357	coronary artery	9.32×10-20 (p-value)	GTEx Portal V6	170
HORMAD1	?	ENSG00000143452	coronary artery	6.82×10-7 (p-value)	GTEx Portal V6	30
RP11-316M1.12	?	ENSG00000259357	vagina	8.66×10-9 (p-value)	GTEx Portal V6	168
HORMAD1	?	ENSG00000143452	vagina	4.25×10-10 (p-value)	GTEx Portal V6	169
RP11-316M1.12	?	ENSG00000259357	gastroesophageal junction	1.36×10-21 (p-value)	GTEx Portal V6	170
ARNT	ENST00000358595	ILMN_2347314	blood	1.36×10-5 (p-value)	Westra et al.	34
ARNT	ENST00000354396					
ARNT	ENST00000471844					
ARNT	ENST00000358595	ILMN_1762582	blood	1.49×10-6 (p-value)	Westra et al.	24
ARNT	ENST00000471844					
ARNT	ENST00000354396					
SETDB1	ENST00000533529	ILMN_1718207	blood	1.07×10-4 (p-value)	Westra et al.	1
SETDB1	ENST00000498193					
SETDB1	ENST00000271640					
SETDB1	ENST00000497314					
SETDB1	ENST00000368969					
CDC42SE1	ENST00000483763	ILMN_1769027	blood	7.47×10-7 (p-value)	Westra et al.	2
CDC42SE1	ENST00000357235					
CDC42SE1	ENST00000540998					
CDC42SE1	ENST00000439374					
RP11-316M1.12	?	ENSG00000259357	adrenal gland	3.87×10-16 (p-value)	GTEx Portal V6	170
CTSS	?	ENSG00000163131	adrenal gland	8.94×10-6 (p-value)	GTEx Portal V6	1
RP11-316M1.12	?	ENSG00000259357	cerebellar hemisphere	1.30×10-13 (p-value)	GTEx Portal V6	170
CTSK	?	ENSG00000143387	cerebellar hemisphere	2.79×10-6 (p-value)	GTEx Portal V6	2
RP11-316M1.12	?	ENSG00000259357	cerebellum	4.99×10-13 (p-value)	GTEx Portal V6	169
CTSK	?	ENSG00000143387	cerebellum	3.44×10-9 (p-value)	GTEx Portal V6	169
CTSS	?	ENSG00000163131	cerebellum	2.22×10-6 (p-value)	GTEx Portal V6	41
ARNT	?	ENSG00000143437	cerebellum	1.22×10-5 (p-value)	GTEx Portal V6	33
RP11-316M1.12	?	ENSG00000259357	frontal cortex	8.50×10-8 (p-value)	GTEx Portal V6	39

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CTSS	ENST00000448301	202902_s_at	chr1	blood	1.80×10-13 (p-value)	Dixon et al.	12
CTSS	ENST00000368985						
CTSS	ENST00000472977						

CTSS ENST00000483930							
ARNT ENST00000358595	230619_at	chr1	blood	3.90×10-14 (p-value)	Dixon et al.	12	
ARNT ENST00000471844							
ARNT ENST00000354396							
CTSS ENST00000368985	ILMN_1743032	chr1	monocyte	5.62×10-34 (p-value)	Zeller et al.	12	

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	16	no	0.000	1

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000363663	1	ENST00000368985, ENST00000448301	CTSS
FFCP00000363734	1	ENST00000368985, ENST00000480760	CTSS
FFCP00000363733	1	ENST00000368985, ENST00000480760	CTSS

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000036493	1	ENCP00000004367	BNIPL
		ENCP00000004381	SEMA6C
ENCE00000036282	1	ENCP00000004342	ARNT
		ENCP00000004352	SETDB1
		ENCP00000004335	CTSS
		ENCP00000004313	PRPF3
		ENCP00000004351	SETDB1
		ENCP00000004357	RP11-316M1.3
		ENCP00000004312	PRPF3
ENCE00000036288	1	ENCP00000004323	ADAMTSL4
		ENCP00000004342	ARNT
		ENCP00000004352	SETDB1
		ENCP00000004313	PRPF3
		ENCP00000004357	RP11-316M1.3
		ENCP00000004351	SETDB1
		ENCP00000004312	PRPF3
ENCE00000036239	1	ENCP00000004337	CTSS
		ENCP00000004345	ARNT
		ENCP00000004350	SETDB1
		ENCP00000004392	PIP5K1A
		ENCP00000004311	PRPF3
		ENCP00000004325	RP11-54A4.2
		ENCP00000004317	TARS2
		ENCP00000004328	MCL1
ENCE00000036391	1	ENCP00000004340	CTSK
ENCE00000036392	1	ENCP00000004340	CTSK
ENCE00000036303	1	ENCP00000004337	CTSS
		ENCP00000004350	SETDB1
		ENCP00000004392	PIP5K1A
		ENCP00000004345	ARNT

	ENCP00000004317	TARS2
	ENCP00000004328	MCL1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000544801 (promoter flanking region)	1	liver (HepG2)	DNase1, H3K4me1, FOXA1
		blood (DND-41)	H3K36me3
		A549	DNase1
ENSR00000285746 (promoter)	2	embryonic stem cell (H1ESC)	H3K36me3, PolII, Rad21, H3K9ac, TAF1, H3K4me2, SP1, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K4me3, DNase1, H3K9ac, H3K79me2, H2AZ, Cfos, PolII, H3K4me2, H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H3K4me3, PolII, ELF1, H2AZ, H3K4me2, H3K27ac, H3K27me3, DNase1
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3
		blood (GM12878)	Pbx3, PolII, H2AZ, DNase1, H3K79me2, H3K4me3, Cfos, H3K4me2, H3K36me3
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3
		NHLF	H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Cfos, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K4me3, PolII, DNase1, H3K4me2
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3
ENSR00000285747 (open chromatin region)	1	blood (K562)	DNase1, H2AZ
ENSR00000285752 (enhancer)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		blood (GM12878)	H3K36me3
		blood (K562)	H2AZ
ENSR00001040051 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		blood (GM12878)	H3K36me3
ENSR00000544808 (promoter)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K4me2
		blood (K562)	DNase1, PU1, H3K9ac, H2AZ, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	DNase1, Cjun
		liver (HepG2)	DNase1
		blood (GM12878)	H3K79me2, H3K4me3, H3K27ac, H3K9ac, DNase1, H2AZ, PU1, PolII
		lung (IMR90)	DNase1
		A549	H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K27ac, H3K4me2
ENSR00001040053 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	PU1
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K4me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	PU1, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ARNT	downstream gene variant	372	ENST00000504358	?	ENSP00000425363	2

ARNT	downstream gene variant	163	ENST00000505979	?	?	2
ARNT	downstream gene variant, upstream gene variant	944	ENST00000468970	?	?	5
ARNT	downstream gene variant	931	ENST00000478972	?	?	3
ARNT	downstream gene variant, upstream gene variant	1730	ENST00000512296	?	?	5
ARNT	upstream gene variant	1699	ENST00000510273	?	?	1
CTSK	downstream gene variant	1720	ENST00000271651	NM_000396.3	ENSP00000271651	1
CTSS	downstream gene variant	9	ENST00000368985	NM_004079.4	ENSP00000357981	6
CTSS	downstream gene variant, upstream gene variant	1928	ENST00000483930	?	ENSP00000475812	9
CTSS	downstream gene variant, upstream gene variant	1022	ENST00000480760	?	?	2
CTSS	downstream gene variant	2326	ENST00000448301	NM_001199739.1	ENSP00000408414	5
CTSS	downstream gene variant, upstream gene variant	277	ENST00000472977	?	ENSP00000475176	10
CTSS	downstream gene variant, upstream gene variant	217	ENST00000607427	?	ENSP00000475557	12
GOLPH3L	downstream gene variant, upstream gene variant	452	ENST00000271732	NM_018178.5	ENSP00000271732	11
GOLPH3L	downstream gene variant, upstream gene variant	332	ENST00000427665	?	ENSP00000410476	9
GOLPH3L	downstream gene variant, upstream gene variant	191	ENST00000479757	?	?	11
HORMAD1	downstream gene variant, upstream gene variant	105	ENST00000476530	?	?	23
HORMAD1	downstream gene variant, upstream gene variant	154	ENST00000322343	NM_001199829.1	ENSP00000326489	14
HORMAD1	downstream gene variant, upstream gene variant	73	ENST00000442853	?	ENSP00000401940	22
HORMAD1	downstream gene variant, upstream gene variant	52	ENST00000368995	?	ENSP00000357991	14
HORMAD1	downstream gene variant, upstream gene variant	383	ENST00000470397	?	?	17
HORMAD1	downstream gene variant, upstream gene variant	59	ENST00000486497	?	?	19
HORMAD1	downstream gene variant, upstream gene variant	15	ENST00000368987	?	ENSP00000357983	23
HORMAD1	downstream gene variant, upstream gene variant	245	ENST00000361824	NM_032132.4	ENSP00000355167	14
RNU6-1042P	upstream gene variant, downstream gene variant	9	ENST00000384204	?	?	20
RP11-363I22.3	upstream gene variant, downstream gene variant	367	ENST00000283110	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ARNT	ENST00000471844	?	ENSP00000425899	19
ARNT	ENST00000505755	NM_178427.2, NM_001197325.1	ENSP00000427571	19
ARNT	ENST00000505979	?	?	13
ARNT	ENST00000358595	NM_001668.3	ENSP00000351407	19
ARNT	ENST00000504358	?	ENSP00000425363	12
ARNT	ENST00000515192	NM_001286035.1	ENSP00000423851	19
ARNT	ENST00000497108	?	?	15
ARNT	ENST00000354396	NM_001286036.1	ENSP00000346372	19
CTSS	ENST00000368985	NM_004079.4	ENSP00000357981	18
CTSS	ENST00000480760	?	?	10
CTSS	ENST00000448301	NM_001199739.1	ENSP00000408414	19

CTSS	ENST00000472977	?	ENSP00000475176	10
CTSS	ENST00000483930	?	ENSP00000475812	11
GOLPH3L	ENST00000271732	NM_018178.5	ENSP00000271732	66
GOLPH3L	ENST00000479757	?	?	2
GOLPH3L	ENST00000427665	?	ENSP00000410476	66
HORMAD1	ENST00000476530	?	?	8
HORMAD1	ENST00000470397	?	?	8
HORMAD1	ENST00000368995	?	ENSP00000357991	41
HORMAD1	ENST00000486497	?	?	15
HORMAD1	ENST00000322343	NM_001199829.1	ENSP00000326489	41
HORMAD1	ENST00000361824	NM_032132.4	ENSP00000355167	41
HORMAD1	ENST00000368987	?	ENSP00000357983	26
HORMAD1	ENST00000442853	?	ENSP00000401940	26

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTSS	ENST00000368985	NM_004079.4	ENSP00000357981	1
GOLPH3L	ENST00000271732	NM_018178.5	ENSP00000271732	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTSS	ENST00000368985	NM_004079.4	ENSP00000357981	1
CTSS	ENST00000448301	NM_001199739.1	ENSP00000408414	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTSS	ENST00000480760	?	1
GOLPH3L	ENST00000479757	?	1
HORMAD1	ENST00000486497	?	1

Locus 202 (rs1003645)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,319,442-34,351,034
block size	31,593 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.338 [-2.377 – 0.716]	gene(s) hit or close-by	CCL15, CCL15-CCL14, CCL23, CTB-186H2.3, RP11-104J23.1, RP11-104J23.2
phastCons	μ = 0.102 [0 – 0.89]	eQTL gene(s)	CCL14, CCL15-CCL14, CCL23
GERP++	μ = 0.013 [-4.46 – 2.02]	potentially regulated gene(s)	–
CADD score	μ = 4.586 [0.258 – 15.76]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CCL23	missense variant	ENST00000615050	NM_005064.4	ENSP00000481357	M/V	Atg/Gtg	tolerated	benign	1
CCL23	missense variant	ENST00000612516	NM_145898.2	ENSP00000484748	M/V	Atg/Gtg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CCL23	ENST00000613876	ILMN_1764030	skin	1.79×10^{-16} (p-value)	MuTHER consortium	9
CCL23	ENST00000615050		adipocyte	2.68×10^{-10} (p-value)	MuTHER consortium	9
CCL23	ENST00000613876	ILMN_1686109	skin	2.24×10^{-19} (p-value)	MuTHER consortium	9
CCL23	ENST00000615050		adipocyte	1.16×10^{-10} (p-value)	MuTHER consortium	9
CCL23	ENST00000612516					
CCL14	ENST00000622526	ILMN_1740609	adipocyte	2.06×10^{-9} (p-value)	MuTHER consortium	9
CCL15-CCL14	ENST00000616694					
CCL15-CCL14	ENST00000610751					
CCL14	ENST00000618404					
CCL14	ENST00000614009					
CCL14	ENST00000620991					
CCL23	ENST00000613876	ILMN_1764030	blood	3.54×10^{-6} (p-value)	Westra et al.	10
CCL23	ENST00000615050					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

CCL15	downstream gene variant, upstream gene variant	473	ENST00000617897	NM_032965.4	ENSP00000484078	4
CCL15	downstream gene variant, upstream gene variant	364	ENST00000614368	?	ENSP00000484262	5
CCL15-CCL14	upstream gene variant	960	ENST00000610751	?	ENSP00000481940	1
CCL15-CCL14	upstream gene variant	890	ENST00000616694	?	ENSP00000481402	1
CCL23	downstream gene variant, upstream gene variant	224	ENST00000612516	NM_145898.2	ENSP00000484748	9
CCL23	downstream gene variant, upstream gene variant	218	ENST00000615050	NM_005064.4	ENSP00000481357	9
CCL23	downstream gene variant, upstream gene variant	261	ENST00000613876	?	ENSP00000479076	9
CTB-186H2.3	downstream gene variant	2136	ENST00000612584	?	?	2
RP11-104J23.1	upstream gene variant, downstream gene variant	1029	ENST00000619334	?	?	7
RP11-104J23.1	upstream gene variant, downstream gene variant	225	ENST00000617328	?	?	7
RP11-104J23.2	upstream gene variant, downstream gene variant	353	ENST00000617747	?	?	9

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15	ENST00000617897	NM_032965.4	ENSP00000484078	2
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	6
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	6
CCL23	ENST00000612516	NM_145898.2	ENSP00000484748	6
CCL23	ENST00000615050	NM_005064.4	ENSP00000481357	6
RP11-104J23.1	ENST00000619334	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15	ENST00000614368	?	ENSP00000484262	2
CCL23	ENST00000613876	?	ENSP00000479076	1

Locus 203 (rs4683702)

SNiPAcard

Block annotations

Block info

genomic range	chr3:142,602,666-142,631,909
block size	29,244 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.009 [-0.853 – 1.31]	gene(s) hit or close-by	PCOLCE2
phastCons	μ = 0.019 [0 – 0.107]	eQTL gene(s)	PCOLCE2
GERP++	μ = 0.012 [-1.66 – 2.8]	potentially regulated gene(s)	ATR, PLS1, RP11-372E1.4, RP11-372E1.7, RP11-383G6.3, RP13-635I23.3, U2SURP
CADD score	μ = 5.080 [0.216 – 12.01]	disease gene(s)	ATR

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1-oleoylglycerophosphoethanolamine	2.36×10^{-6}	Metabolomics GWAS Server	24816252	7

Disease gene annotation

gene	trait	source DB	source entry/link
ATR	SECKEL SYNDROME 1	OMIM	MIM:210600
ATR	CUTANEOUS TELANGIECTASIA AND CANCER SYNDROME, FAMILIAL	OMIM	MIM:614564
ATR	Seckel syndrome type 1 (SCKL1)	DECIPHER	MIM:210600
ATR	Seckel syndrome	OrphaNet	OrphaNet:808
ATR	Familial cutaneous telangiectasia and oropharyngeal predisposition cancer syndrome	OrphaNet	OrphaNet:313846

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PCOLCE2	ENST00000485766	ILMN_1746888	adipocyte	1.81×10^{-15} (p-value)	MuTHER consortium	7
PCOLCE2	ENST00000470795					
PCOLCE2	ENST00000295992					
PCOLCE2	?	ENSG00000163710	aorta	1.11×10^{-11} (p-value)	GTEx Portal V6	12
PCOLCE2	?	ENSG00000163710	left ventricle	5.95×10^{-6} (p-value)	GTEx Portal V6	7

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000357025	1	ENCP00000038521	RP11-372E1.4
		ENCP00000038539	RP13-635I23.3
ENCE00000356905	1	ENCP00000038522	RP11-372E1.7

	ENCP00000038513	PLS1
	ENCP00000038531	U2SURP
	ENCP00000038504	RP11-383G6.3 ATR
	ENCP00000038510	PLS1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001679457 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001679459 (enhancer)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PCOLCE2	downstream gene variant, upstream gene variant	512	ENST00000483454	?	?	7
PCOLCE2	upstream gene variant	2010	ENST00000495732	?	?	2
PCOLCE2	upstream gene variant	2172	ENST00000485766	?	ENSP00000419842	2
PCOLCE2	upstream gene variant	2326	ENST00000493733	?	?	2
PCOLCE2	upstream gene variant	2049	ENST00000461818	?	?	2
PCOLCE2	upstream gene variant	1868	ENST00000295992	NM_013363.3	ENSP00000295992	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PCOLCE2	ENST00000495732	?	?	5
PCOLCE2	ENST00000461818	?	?	5
PCOLCE2	ENST00000485766	?	ENSP00000419842	5
PCOLCE2	ENST00000493733	?	?	5
PCOLCE2	ENST00000295992	NM_013363.3	ENSP00000295992	5

Locus 204 (rs4443656)

SNiPAcard

Block annotations

Block info

genomic range	chr8:21,521,899-21,553,025
block size	31,127 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.111 [-6.714 – 0.285]	gene(s) hit or close-by	GFRA2
phastCons	μ = 0.001 [0 – 0.005]	eQTL gene(s)	GFRA2
GERP++	μ = -1.147 [-5.4 – 4.46]	potentially regulated gene(s)	–
CADD score	μ = 1.791 [0.058 – 4.948]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
xanthine	5.91×10^{-5}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GFRA2	ENST00000524240	ILMN_1656300	skin	2.81×10^{-7} (p-value)	MuTHER consortium	12
GFRA2	ENST00000306793		adipocyte	7.39×10^{-8} (p-value)	MuTHER consortium	12

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001439445 (promoter flanking region)	1	HSMMtube	H3K27me3, DNase1
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K27ac, H3K4me1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
ENSR00001720518 (enhancer)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	Rad21
		liver (HepG2)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me1, CTCF, DNase1, H3K27ac
ENSR00001387351 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1

		Osteobl	H3K27ac
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me1, DNase1, H3K27ac
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	Jund, DNase1
		monocytes (Monocytes-CD14+)	DNase1
		liver (HepG2)	H3K27me3
		nervous (NH-A)	DNase1
		A549	H3K27ac, DNase1, H3K4me2, H3K4me1, H3K4me3
ENSR00001439448 (promoter flanking region)	3	NHLF	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		A549	H3K27ac, DNase1, H3K4me3
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GFRA2	downstream gene variant	1429	ENST00000518077	NM_001165039.1	ENSP00000429206	4
GFRA2	downstream gene variant	512	ENST00000306793	?	ENSP00000306654	6
GFRA2	downstream gene variant	848	ENST00000524240	NM_001495.4	ENSP00000428518	5
GFRA2	downstream gene variant	1337	ENST00000517328	?	ENSP00000429445	4
GFRA2	downstream gene variant	1676	ENST00000517892	NM_001165038.1	ENSP00000429979	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GFRA2	ENST00000517892	NM_001165038.1	ENSP00000429979	1
GFRA2	ENST00000517328	?	ENSP00000429445	1
GFRA2	ENST00000524240	NM_001495.4	ENSP00000428518	1
GFRA2	ENST00000518077	NM_001165039.1	ENSP00000429206	1
GFRA2	ENST00000306793	?	ENSP00000306654	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GFRA2	ENST00000517892	NM_001165038.1	ENSP00000429979	1
GFRA2	ENST00000517328	?	ENSP00000429445	1
GFRA2	ENST00000524240	NM_001495.4	ENSP00000428518	3
GFRA2	ENST00000518077	NM_001165039.1	ENSP00000429206	1
GFRA2	ENST00000306793	?	ENSP00000306654	1

Locus 205 (rs3828922)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,708,147-31,741,490
block size	33,344 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.734 [-3.279 – 0.364]	gene(s) hit or close-by	CLIC1, MSH5, MSH5-SAPCD1, RNU6-850P, SAPCD1, SAPCD1-AS1, VARS, VWA7
phastCons	μ = 0.013 [0 – 0.066]	eQTL gene(s)	AIF1, ATF6B, BAG6, C6orf48, DXO, HSPA1B, LY6G5C, LY6G6C, NELFE, PRRC2A, SKIV2L, SNORD52, TNF
GERP++	μ = -1.588 [-4.48 – 1.66]	potentially regulated gene(s)	–
CADD score	μ = 4.579 [0.264 – 7.987]	disease gene(s)	SKIV2L, TNF

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LY6G5C	?	ENSG00000204428	caudate basal ganglia	2.18×10^{-7} (p-value)	GTEx Portal V6	6
DXO	ENST00000474587	ILMN_1726990	blood	3.95×10^{-10} (p-value)	MuTHER consortium	5
DXO	ENST00000491327		adipocyte	1.65×10^{-5} (p-value)	MuTHER consortium	5
DXO	ENST00000473976					
DXO	ENST00000480240					
DXO	ENST00000498357					
DXO	ENST00000477826					
DXO	ENST00000492946					
DXO	ENST00000375349					
DXO	ENST00000337523					
DXO	ENST00000460058					
DXO	ENST00000375356					
DXO	ENST00000487914					
DXO	ENST00000478221					
DXO	ENST00000485557					
PRRC2A	ENST00000376033	ILMN_2408179	blood	1.95×10^{-7} (p-value)	MuTHER consortium	5
PRRC2A	ENST00000487089					

PRRC2A	ENST00000376007					
PRRC2A	ENST00000487839					
PRRC2A	ENST00000462617					
PRRC2A	ENST00000482441					
PRRC2A	ENST00000492691					
HSPA1B	ENST00000375650	ILMN_1660436	skin	3.11×10^{-6} (p-value)	MuTHER consortium	5
AIF1	ENST00000337917	ILMN_1792473	blood	8.51×10^{-10} (p-value)	MuTHER consortium	5
AIF1	ENST00000466820					
AIF1	ENST00000497362					
AIF1	ENST00000376059					
AIF1	ENST00000376049					
LY6G5C	?	ENSG00000204428	lung	6.94×10^{-7} (p-value)	GTEx Portal V6	6
C6orf48	?	ENSG00000204387	tibial nerve	6.53×10^{-8} (p-value)	GTEx Portal V6	6
LY6G5C	?	ENSG00000204428	blood	1.15×10^{-11} (p-value)	GTEx Portal V6	6
C6orf48	?	ENSG00000204387	blood	2.20×10^{-7} (p-value)	GTEx Portal V6	6
LY6G6C	?	ENSG00000204421	esophagus mucosa	2.20×10^{-14} (p-value)	GTEx Portal V6	6
TNF	ENST00000449264	ILMN_1728106	blood	5.59×10^{-5} (p-value)	Westra et al.	5
NELFE	ENST00000492185	ILMN_1765532	blood	8.69×10^{-6} (p-value)	Westra et al.	5
NELFE	ENST00000488426					
NELFE	ENST00000481121					
NELFE	ENST00000375429					
SKIV2L	ENST00000484835	ILMN_1666512	blood	9.00×10^{-5} (p-value)	Westra et al.	5
SKIV2L	ENST00000375394					
SKIV2L	ENST00000474839					
SKIV2L	ENST00000470453					
SKIV2L	ENST00000465703					
SKIV2L	ENST00000491994					
SKIV2L	ENST00000471818					
SKIV2L	ENST00000483553					
SKIV2L	ENST00000485349					
C6orf48	?	ENSG00000204387	testis	9.76×10^{-7} (p-value)	GTEx Portal V6	6
C6orf48	?	ENSG00000204387	unexposed skin	1.45×10^{-7} (p-value)	GTEx Portal V6	6
?	?	ILMN_1770787	b-cell	4.66×10^{-5} (p-value)	Fairfax et al.	3
?	?	ILMN_2044471	b-cell	1.41×10^{-4} (p-value)	Fairfax et al.	3
C6orf48	ENST00000375638	ILMN_2391765	monocyte	5.43×10^{-10} (p-value)	Fairfax et al.	3
C6orf48	ENST00000375635		b-cell	1.36×10^{-11} (p-value)	Fairfax et al.	3
C6orf48	ENST00000395788					
C6orf48	ENST00000614363					
C6orf48	ENST00000375640					
SNORD52	ENST00000364884					
C6orf48	ENST00000375641					
C6orf48	ENST00000395789					
C6orf48	ENST00000375639					
C6orf48	ENST00000375633					
C6orf48	ENST00000375642					
ATF6B	ENST00000375201	ILMN_3194911	b-cell	5.63×10^{-5} (p-value)	Fairfax et al.	3
ATF6B	ENST00000375203					

?	?	ILMN_2325394	monocyte	1.41×10-13 (p-value)	Zeller et al.	1
C6orf48	ENST00000375638	ILMN_2391765	monocyte	5.44×10-25 (p-value)	Zeller et al.	1
C6orf48	ENST00000375635					
C6orf48	ENST00000395788					
C6orf48	ENST00000614363					
C6orf48	ENST00000375640					
C6orf48	ENST00000395789					
C6orf48	ENST00000375641					
SNORD52	ENST00000364884					
C6orf48	ENST00000375639					
C6orf48	ENST00000375633					
C6orf48	ENST00000375642					
BAG6	?	ENSG00000204463	tibial artery	7.03×10-6 (p-value)	GTEx Portal V6	1
LY6G5C	?	ENSG00000204428	thyroid	3.04×10-6 (p-value)	GTEx Portal V6	2
LY6G5C	?	ENSG00000204428	sun exposed skin	7.88×10-7 (p-value)	GTEx Portal V6	2
LY6G5C	?	ENSG00000204428	nucleus accumbens	4.64×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487898 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K4me3, PolII, Rad21, Jund, Nrsf, Sin3Ak20, CTCF, H3K27ac, ATF3, Yy1, H3K4me1, TAF1, H3K4me2, SP1, USF1, H3K9ac
		HSMMtube	H3K9ac, DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2
		blood (K562)	H3K4me3, Jund, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Cjun, Cfos, Cmyc, PolII, H4K20me1, H3K4me2, CTCF, H3K4me1, H3K36me3, H3K9ac, E2F6, Yy1, Ini1, Brg1, Egr1, FOSL1, H3K27ac, Max, SP2, ZBTB7A, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac, CTCF, H3K4me1, H3K36me3
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K4me2, HNF4A, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3, p300, H3K4me1, FOSL2, Jund, HNF4G, FOXA2, PolII, HDAC2, H3K79me2, TAF1, RXRA, ELF1
		lung (IMR90)	H3K9ac, H3K4ac, DNase1, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3
		blood (GM12878)	H3K27ac, Cfos, H3K4me2, H3K9ac, H3K36me3, H3K4me3, H3K79me2, PolII, H2AZ, H3K4me1, DNase1, CTCF, Yy1, ELF1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me2, H3K4me3, H3K36me3, DNase1, H3K9ac, H3K27ac, H3K4me1, CTCF
		NHLF	H3K27ac, H3K36me3, DNase1, H3K4me3, H3K9ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3, CTCF
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, Cmyc, CTCF, H3K36me3, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, Cmyc, CTCF, PolII, H3K27ac, Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K4me1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CLIC1	upstream gene variant	3803	ENST00000375784	NM_001288.4	ENSP00000364940	1
CLIC1	upstream gene variant	3798	ENST00000375779	?	ENSP00000364934	1
CLIC1	upstream gene variant	607	ENST00000395892	?	ENSP00000379229	3
CLIC1	upstream gene variant	3825	ENST00000616760	NM_001287594.1	ENSP00000479808	1
CLIC1	upstream gene variant	3052	ENST00000375780	NM_001287593.1	ENSP00000364935	1

MSH5	upstream gene variant	884	ENST00000497269	?	ENSP00000419131	2
MSH5	downstream gene variant	924	ENST00000463094	?	?	2
MSH5	upstream gene variant	722	ENST00000429846	?	ENSP00000406849	1
MSH5	downstream gene variant	170	ENST00000425703	?	ENSP00000402842	2
MSH5	upstream gene variant, downstream gene variant	631	ENST00000450148	?	ENSP00000394971	3
MSH5	downstream gene variant	317	ENST00000494646	?	?	1
MSH5	upstream gene variant	876	ENST00000463144	?	ENSP00000419648	1
MSH5	downstream gene variant	1356	ENST00000468602	?	?	1
MSH5	upstream gene variant	1433	ENST00000494458	?	?	1
MSH5	downstream gene variant	988	ENST00000467319	?	?	1
MSH5	downstream gene variant	989	ENST00000468136	?	?	1
MSH5-SAPCD1	upstream gene variant	2506	ENST00000425424	?	ENSP00000413372	1
MSH5-SAPCD1	upstream gene variant	1954	ENST00000476085	?	ENSP00000435414	1
MSH5-SAPCD1	upstream gene variant	230	ENST00000491552	?	?	1
RNU6-850P	upstream gene variant	3437	ENST00000516934	?	?	1
SAPCD1	upstream gene variant	3130	ENST00000494299	?	?	1
SAPCD1	upstream gene variant	2309	ENST00000433778	?	ENSP00000408918	1
SAPCD1	upstream gene variant	2506	ENST00000415669	NM_001039651.1	ENSP00000411948	1
SAPCD1-AS1	downstream gene variant	3820	ENST00000419679	?	?	1
VARS	downstream gene variant	3805	ENST00000470953	?	?	1
VARS	downstream gene variant	3805	ENST00000375663	NM_006295.2	ENSP00000364815	1
VWA7	downstream gene variant	2346	ENST00000497645	?	?	1
VWA7	upstream gene variant	706	ENST00000487013	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MSH5	ENST00000375740	NM_025259.5	ENSP00000364892	5
MSH5	ENST00000463094	?	?	2
MSH5	ENST00000482280	?	?	4
MSH5	ENST00000425703	?	ENSP00000402842	2
MSH5	ENST00000423982	?	ENSP00000406352	5
MSH5	ENST00000375703	NM_172165.3	ENSP00000364855	5
MSH5	ENST00000375755	NM_002441.4	ENSP00000364908	5
MSH5	ENST00000450148	?	ENSP00000394971	2
MSH5	ENST00000375750	NM_172166.3	ENSP00000364903	5
MSH5	ENST00000463144	?	ENSP00000419648	4
MSH5	ENST00000497269	?	ENSP00000419131	2
MSH5	ENST00000395853	?	ENSP00000379194	1
MSH5	ENST00000484309	?	ENSP00000420232	1
MSH5-SAPCD1	ENST00000493662	?	ENSP00000417871	5

MSH5-SAPCD1	ENST00000498473	?	ENSP00000419220	1
VWA7	ENST00000467576	?	?	1
VWA7	ENST00000375688	NM_025258.2	ENSP00000364840	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MSH5	ENST00000423982	?	ENSP00000406352	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
MSH5	ENST00000463094	?	1

Locus 206 (rs1794282)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,218,120-32,776,921
block size	558,802 bp
variant count	30 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.450 [-3.618 – 0.444]	gene(s) hit or close-by	BTNL2, C6orf10, HLA-DOB, HLA-DRA, HLA-DRB9, HNRNPA1P2, RNU6-603P, XXbac-BPG154L12.4, XXbac-BPG154L12.5, XXbac-BPG246D15.9
phastCons	μ = 0.127 [0 – 0.923]	eQTL gene(s)	C4A, C4B, CYP21A1P, CYP21A2, HCP5, HLA-C, HLA-DMA, HLA-DMB, HLA-DPB1, HLA-DQA1, HLA-DQB1, HLA-DQB1-AS1, HLA-DQB2, HLA-DRB5, HLA-DRB6, LY6G6C, MICB, NOTCH4, PSMB9, RNF5, STK19, STK19B, TAP2, TNXA, TNXB, VARS2, WASF5P, XXbac-BPG246D15.9, XXbac-BPG248L24.13
GERP++	μ = -0.271 [-5.03 – 1.82]	potentially regulated gene(s)	BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, C2, C2, C2, C2, C2, C2, C2, C2, C2, C2, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, HCG23, HCG23, HCG23, HCG23, HCG23, HCG23, HCG23, HCG23, HLA-DRB6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6
CADD score	μ = 2.492 [0.026 – 15.24]	disease gene(s)	C4B, C4A, CYP21A2, VARS2, , , , , , , , , , C2, , , , , , , BTNL2, HLA-DRA, TAP2, TNXB, HLA-DPB1, HLA-DQB1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Lupus erythematosus, systemic	2.21×10^{-7}	dbGaP	pha002867	dbGaP	1
Diabetes mellitus, type 1	2.48×10^{-103}	dbGaP	pha002862	dbGaP	1
Lupus erythematosus, systemic	4.01×10^{-18}	dbGaP	pha002848	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380	OMIM
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790	OMIM
CYP21A2	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM

	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
BTNL2	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
HLA-DRA	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
TAP2	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839	orphanet
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285	orphanet
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	OrphaNet	OrphaNet:315306	orphanet
CYP21A2	Nonclassic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	OrphaNet	OrphaNet:95698	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	OrphaNet	OrphaNet:315311	orphanet
HLA-DPB1	Chronic berylliosis	OrphaNet	OrphaNet:133	orphanet
HLA-DPB1	Granulomatosis with polyangiitis	OrphaNet	OrphaNet:900	orphanet
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
BTNL2	Sarcoidosis	OrphaNet	OrphaNet:797	orphanet
HLA-DQB1	Bullous pemphigoid	OrphaNet	OrphaNet:703	orphanet
HLA-DQB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
HLA-DQB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465	orphanet
HLA-DRA	Graham Little-Piccardi-Lassueur syndrome	OrphaNet	OrphaNet:505	orphanet
TAP2	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C4A	?	ENSG00000244731	pancreas	5.71×10^{-13} (p-value)	GTEx Portal V6	24
RNF5	?	ENSG00000204308	pancreas	1.41×10^{-6} (p-value)	GTEx Portal V6	22
XXbac-BPG248L24.13	?	ENSG00000256166	pancreas	3.28×10^{-7} (p-value)	GTEx Portal V6	5
HLA-DMA	?	ENSG00000204257	pancreas	2.02×10^{-11} (p-value)	GTEx Portal V6	24
WASF5P	?	ENSG00000231402	pancreas	4.68×10^{-7} (p-value)	GTEx Portal V6	5
CYP21A1P	?	ENSG00000204338	pancreas	2.84×10^{-7} (p-value)	GTEx Portal V6	14
HLA-DQB2	?	ENSG00000232629	muscularis mucosae	2.71×10^{-7} (p-value)	GTEx Portal V6	19
C4A	?	ENSG00000244731	muscularis mucosae	2.15×10^{-19} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	muscularis mucosae	3.72×10^{-7} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	muscularis mucosae	2.74×10^{-12} (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	lung	7.65×10^{-7} (p-value)	GTEx Portal V6	20

C4A	?	ENSG00000244731	lung	4.85×10-17 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	lung	1.72×10-8 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	lung	5.42×10-10 (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	atrial appendage	6.02×10-8 (p-value)	GTEx Portal V6	19
C4A	?	ENSG00000244731	atrial appendage	6.06×10-16 (p-value)	GTEx Portal V6	25
CYP21A1P	?	ENSG00000204338	atrial appendage	1.27×10-9 (p-value)	GTEx Portal V6	25
HLA-DMA	?	ENSG00000204257	transformed fibroblasts	1.88×10-20 (p-value)	GTEx Portal V6	24
HLA-DMB	?	ENSG00000242574	transformed fibroblasts	5.14×10-13 (p-value)	GTEx Portal V6	21
C4B	?	ENSG00000224389	transformed fibroblasts	1.54×10-15 (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	transformed fibroblasts	2.95×10-17 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	transformed fibroblasts	3.17×10-8 (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	transformed fibroblasts	9.47×10-6 (p-value)	GTEx Portal V6	1
HLA-DMA	?	ENSG00000204257	tibial artery	2.07×10-9 (p-value)	GTEx Portal V6	20
C4A	?	ENSG00000244731	tibial artery	2.30×10-13 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	tibial artery	7.01×10-10 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	tibial artery	1.84×10-12 (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	breast	1.15×10-14 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	breast	9.99×10-10 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	breast	6.62×10-13 (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	breast	1.26×10-7 (p-value)	GTEx Portal V6	10
STK19B	?	ENSG00000250535	breast	6.19×10-7 (p-value)	GTEx Portal V6	1
C4B	?	ENSG00000224389	blood	3.97×10-17 (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	blood	2.06×10-16 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	blood	3.69×10-8 (p-value)	GTEx Portal V6	4
CYP21A2	?	ENSG00000231852	blood	3.10×10-7 (p-value)	GTEx Portal V6	23
CYP21A1P	?	ENSG00000204338	blood	8.43×10-16 (p-value)	GTEx Portal V6	25
HLA-DMA	?	ENSG00000204257	thyroid	1.77×10-18 (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	thyroid	4.56×10-21 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	thyroid	1.36×10-6 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	thyroid	2.70×10-18 (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	thyroid	9.43×10-7 (p-value)	GTEx Portal V6	12
HLA-DRB6	?	ENSG00000229391	thyroid	6.75×10-6 (p-value)	GTEx Portal V6	3
HLA-DQA1	?	ENSG00000196735	skeletal muscle	6.40×10-8 (p-value)	GTEx Portal V6	24
C4A	?	ENSG00000244731	skeletal muscle	5.08×10-20 (p-value)	GTEx Portal V6	25
HLA-DRB5	?	ENSG00000198502	skeletal muscle	6.10×10-7 (p-value)	GTEx Portal V6	20
HLA-C	?	ENSG00000204525	skeletal muscle	1.19×10-13 (p-value)	GTEx Portal V6	4
RNF5	?	ENSG00000204308	skeletal muscle	4.88×10-10 (p-value)	GTEx Portal V6	24

HLA-DRB6	?	ENSG00000229391	skeletal muscle	3.08×10^{-8} (p-value)	GTEx Portal V6	22
CYP21A1P	?	ENSG00000204338	skeletal muscle	6.94×10^{-7} (p-value)	GTEx Portal V6	20
HLA-DQB2	?	ENSG00000232629	skeletal muscle	1.40×10^{-6} (p-value)	GTEx Portal V6	9
HLA-DQB1-AS1	?	ENSG00000223534	skeletal muscle	2.02×10^{-6} (p-value)	GTEx Portal V6	7
HLA-DMA	?	ENSG00000204257	skeletal muscle	5.10×10^{-10} (p-value)	GTEx Portal V6	9
C4A	?	ENSG00000244731	transverse colon	1.71×10^{-12} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	transverse colon	6.93×10^{-7} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	transverse colon	1.98×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	EBV lymphocytes	9.01×10^{-10} (p-value)	GTEx Portal V6	21
HLA-C	?	ENSG00000204525	EBV lymphocytes	3.18×10^{-6} (p-value)	GTEx Portal V6	4
C4A	?	ENSG00000244731	unexposed skin	4.36×10^{-16} (p-value)	GTEx Portal V6	25
CYP21A2	?	ENSG00000231852	unexposed skin	9.86×10^{-7} (p-value)	GTEx Portal V6	18
CYP21A1P	?	ENSG00000204338	unexposed skin	5.73×10^{-10} (p-value)	GTEx Portal V6	24
MICB	?	ENSG00000204516	unexposed skin	2.73×10^{-6} (p-value)	GTEx Portal V6	16
HLA-DMA	?	ENSG00000204257	unexposed skin	1.94×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	sun exposed skin	4.91×10^{-17} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	sun exposed skin	3.99×10^{-8} (p-value)	GTEx Portal V6	4
NOTCH4	?	ENSG00000204301	sun exposed skin	2.52×10^{-9} (p-value)	GTEx Portal V6	21
HLA-DMA	?	ENSG00000204257	sun exposed skin	1.82×10^{-15} (p-value)	GTEx Portal V6	25
C4B	?	ENSG00000224389	sun exposed skin	8.56×10^{-14} (p-value)	GTEx Portal V6	24
CYP21A2	?	ENSG00000231852	sun exposed skin	1.11×10^{-10} (p-value)	GTEx Portal V6	25
MICB	?	ENSG00000204516	sun exposed skin	5.07×10^{-8} (p-value)	GTEx Portal V6	21
CYP21A1P	?	ENSG00000204338	sun exposed skin	2.27×10^{-13} (p-value)	GTEx Portal V6	25
HLA-DMA	?	ENSG00000204257	liver	4.11×10^{-13} (p-value)	GTEx Portal V6	25
STK19B	?	ENSG00000250535	liver	4.66×10^{-8} (p-value)	GTEx Portal V6	21
C4A	?	ENSG00000244731	liver	3.14×10^{-14} (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	left ventricle	7.06×10^{-8} (p-value)	GTEx Portal V6	21
C4A	?	ENSG00000244731	left ventricle	6.97×10^{-17} (p-value)	GTEx Portal V6	25
CYP21A1P	?	ENSG00000204338	left ventricle	1.10×10^{-9} (p-value)	GTEx Portal V6	25
NOTCH4	?	ENSG00000204301	left ventricle	7.25×10^{-8} (p-value)	GTEx Portal V6	24
C4A	?	ENSG00000244731	aorta	7.08×10^{-16} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	aorta	2.49×10^{-7} (p-value)	GTEx Portal V6	4
CYP21A2	?	ENSG00000231852	aorta	1.41×10^{-6} (p-value)	GTEx Portal V6	12
CYP21A1P	?	ENSG00000204338	aorta	1.34×10^{-8} (p-value)	GTEx Portal V6	24
MICB	?	ENSG00000204516	aorta	1.44×10^{-8} (p-value)	GTEx Portal V6	22
NOTCH4	?	ENSG00000204301	aorta	3.98×10^{-6} (p-value)	GTEx Portal V6	1
HLA-DQB2	?	ENSG00000232629	subcutaneous adipocytes	5.08×10^{-11} (p-value)	GTEx Portal V6	24

C4A	?	ENSG00000244731	subcutaneous adipocytes	1.31×10^{-17} (p-value)	GTEx Portal V6	25
HLA-DRB5	?	ENSG00000198502	subcutaneous adipocytes	3.29×10^{-7} (p-value)	GTEx Portal V6	23
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	1.50×10^{-8} (p-value)	GTEx Portal V6	4
NOTCH4	?	ENSG00000204301	subcutaneous adipocytes	3.45×10^{-7} (p-value)	GTEx Portal V6	21
C4B	?	ENSG00000224389	subcutaneous adipocytes	2.32×10^{-6} (p-value)	GTEx Portal V6	10
CYP21A2	?	ENSG00000231852	subcutaneous adipocytes	4.22×10^{-8} (p-value)	GTEx Portal V6	25
CYP21A1P	?	ENSG00000204338	subcutaneous adipocytes	1.25×10^{-14} (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	visceral adipocytes	4.43×10^{-10} (p-value)	GTEx Portal V6	24
C4A	?	ENSG00000244731	visceral adipocytes	5.89×10^{-11} (p-value)	GTEx Portal V6	24
HLA-C	?	ENSG00000204525	visceral adipocytes	5.84×10^{-7} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	visceral adipocytes	2.87×10^{-9} (p-value)	GTEx Portal V6	24
NOTCH4	?	ENSG00000204301	visceral adipocytes	3.60×10^{-9} (p-value)	GTEx Portal V6	22
C4A	?	ENSG00000244731	stomach	3.95×10^{-16} (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	spleen	3.84×10^{-8} (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	tibial nerve	6.98×10^{-23} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	tibial nerve	6.43×10^{-8} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	tibial nerve	1.34×10^{-13} (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	sigmoid colon	1.97×10^{-9} (p-value)	GTEx Portal V6	24
CYP21A1P	?	ENSG00000204338	sigmoid colon	6.24×10^{-7} (p-value)	GTEx Portal V6	1
LY6G6C	?	ENSG00000204421	esophagus mucosa	4.94×10^{-9} (p-value)	GTEx Portal V6	23
C4A	?	ENSG00000244731	esophagus mucosa	4.61×10^{-16} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	esophagus mucosa	2.15×10^{-9} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	esophagus mucosa	1.69×10^{-10} (p-value)	GTEx Portal V6	24
MICB	?	ENSG00000204516	esophagus mucosa	9.26×10^{-9} (p-value)	GTEx Portal V6	23
HLA-DQB1	?	ENSG00000179344	esophagus mucosa	1.49×10^{-6} (p-value)	GTEx Portal V6	6
HLA-DMA	?	ENSG00000204257	esophagus mucosa	2.46×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	gastroesophageal junction	1.02×10^{-12} (p-value)	GTEx Portal V6	24
CYP21A1P	?	ENSG00000204338	gastroesophageal junction	1.06×10^{-8} (p-value)	GTEx Portal V6	24
HLA-DQB2	?	ENSG00000232629	gastroesophageal junction	8.86×10^{-7} (p-value)	GTEx Portal V6	5
C4A	?	ENSG00000244731	cerebellar hemisphere	5.31×10^{-8} (p-value)	GTEx Portal V6	22
TNXB	?	ENSG00000168477	adrenal gland	2.46×10^{-7} (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	adrenal gland	5.08×10^{-8} (p-value)	GTEx Portal V6	19
STK19	?	ENSG00000204344	adrenal gland	2.32×10^{-7} (p-value)	GTEx Portal V6	20
STK19B	?	ENSG00000250535	adrenal gland	7.79×10^{-12} (p-value)	GTEx Portal V6	25
TNXA	?	ENSG00000248290	adrenal gland	5.38×10^{-10} (p-value)	GTEx Portal V6	21
CYP21A1P	?	ENSG00000204338	adrenal gland	2.89×10^{-8} (p-value)	GTEx Portal V6	22
C4A	?	ENSG00000244731	testis	3.99×10^{-17} (p-value)	GTEx Portal V6	25

CYP21A1P	?	ENSG00000204338	testis	1.92×10-12 (p-value)	GTEx Portal V6	25
HLA-DMA	?	ENSG00000204257	cerebellum	2.83×10-9 (p-value)	GTEx Portal V6	20
C4A	?	ENSG00000244731	cerebellum	4.80×10-8 (p-value)	GTEx Portal V6	20
CYP21A1P	?	ENSG00000204338	cerebellum	8.19×10-8 (p-value)	GTEx Portal V6	20
C4A	?	ENSG00000244731	frontal cortex	1.94×10-8 (p-value)	GTEx Portal V6	19
NOTCH4	ENST00000474612	ILMN_1711157	adipocyte	1.20×10-9 (p-value)	MuTHER consortium	7
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
HCP5	ENST00000414046	ILMN_1803945	skin	5.66×10-6 (p-value)	MuTHER consortium	6
HCP5	ENST00000541196		adipocyte	3.41×10-6 (p-value)	MuTHER consortium	6
HLA-DPB1	ENST00000418931	ILMN_1749070	skin	4.66×10-9 (p-value)	MuTHER consortium	7
HLA-DPB1	ENST00000416804		blood	3.81×10-10 (p-value)	MuTHER consortium	7
			adipocyte	1.57×10-8 (p-value)	MuTHER consortium	7
MICB	ENST00000538442	ILMN_1708006	skin	3.44×10-9 (p-value)	MuTHER consortium	6
MICB	ENST00000399150					
MICB	ENST00000252229					
PSMB9	ENST00000467593	ILMN_1798233	blood	1.55×10-7 (p-value)	MuTHER consortium	4
PSMB9	ENST00000464863					
VARS2	ENST00000321897	ILMN_1737585	monocyte	2.87×10-8 (p-value)	Fairfax et al.	3
VARS2	ENST00000625423		b-cell	3.50×10-6 (p-value)	Fairfax et al.	3
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
HLA-DPB1	ENST00000418931	ILMN_1749070	monocyte	3.64×10-8 (p-value)	Fairfax et al.	2
HLA-DPB1	ENST00000416804		b-cell	2.63×10-8 (p-value)	Fairfax et al.	2
C4A	?	ENSG00000244731	putamen	2.67×10-8 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	caudate basal ganglia	2.05×10-8 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	hypothalamus	8.80×10-8 (p-value)	GTEx Portal V6	2
C4A	?	ENSG00000244731	coronary artery	8.43×10-7 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	anterior cingulate cortex	1.39×10-8 (p-value)	GTEx Portal V6	1
TAP2	ENST00000620123	ILMN_1759250	blood	8.02×10-7 (p-value)	Westra et al.	1
TAP2	ENST00000374899					
XXbac-BPG246D15.9	ENST00000452392					
TAP2	ENST00000464100					
TAP2	ENST00000374897					
HLA-DQB1	ENST00000399082	209480_at	blood	4.90×10-12 (p-value)	Dixon et al.	1
HLA-DQB1	ENST00000399084					
HLA-DQB1	ENST00000434651					
HLA-DQB1	ENST00000460185					
HLA-DQB1	ENST00000399079					
HLA-DQB1	ENST00000443574					

HLA-DQB1	ENST00000374943					
HLA-DQB1	ENST00000487676					
?	?	221491_x_at	blood	2.80×10^{-17} (p-value)	Dixon et al.	1
HLA-DQA1	ENST00000374949	213831_at	blood	1.60×10^{-10} (p-value)	Dixon et al.	1
HLA-DQA1	ENST00000496318					
HLA-DQA1	ENST00000460633					
HLA-DQA1	ENST00000482745					
HLA-DQA1	ENST00000343139					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445252	1	ENCP00000048214	XXbac-BPG254F23.6 XXbac-BPG254F23.6 XXbac-BPG254F23.6 XXbac-BPG254F23.6 XXbac-BPG254F23.6 XXbac-BPG254F23.6
		ENCP00000048178	EGFL8 EGFL8 EGFL8 EGFL8 EGFL8 EGFL8 EGFL8
		ENCP00000048131	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000048199	HCG23 HCG23 HCG23 HCG23 HCG23 HCG23 HCG23 HCG23
		ENCP00000048206	HLA-DRB6
		ENCP00000048201	BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BTNL2	upstream gene variant	2924	ENST00000374993	NM_019602.1	ENSP00000364132	2
BTNL2	upstream gene variant	2919	ENST00000454136	?	ENSP00000390613	2
BTNL2	upstream gene variant	2924	ENST00000465865	?	ENSP00000420063	2
BTNL2	downstream gene variant, upstream gene variant	2919	ENST00000446536	?	ENSP00000388434	3
BTNL2	upstream gene variant	2924	ENST00000544175	?	ENSP00000443364	2

C6orf10	upstream gene variant	1187	ENST00000533191	NM_001286474.1	ENSP00000431199	3
C6orf10	upstream gene variant	1182	ENST00000617061	?	ENSP00000482001	3
C6orf10	downstream gene variant, upstream gene variant	1222	ENST00000532023	?	ENSP00000432814	4
C6orf10	downstream gene variant, upstream gene variant	1215	ENST00000534588	?	ENSP00000432566	4
C6orf10	upstream gene variant	1187	ENST00000375015	?	ENSP00000364155	3
C6orf10	downstream gene variant, upstream gene variant	1195	ENST00000442822	?	ENSP00000411164	4
C6orf10	upstream gene variant	1222	ENST00000527965	NM_001286475.1	ENSP00000435103	3
C6orf10	upstream gene variant	1215	ENST00000375007	?	ENSP00000364146	3
C6orf10	upstream gene variant	1215	ENST00000447241	NM_006781.4	ENSP00000415517	3
C6orf10	upstream gene variant	1182	ENST00000612031	?	ENSP00000480403	3
HLA-DOB	downstream gene variant	3619	ENST00000488325	?	ENSP00000436618	1
HLA-DOB	downstream gene variant	3903	ENST00000475235	?	?	1
HLA-DOB	downstream gene variant	3619	ENST00000438763	NM_002120.3	ENSP00000390020	1
HLA-DRA	downstream gene variant	1612	ENST00000374982	?	ENSP00000364121	2
HLA-DRA	downstream gene variant	1612	ENST00000395388	NM_019111.4	ENSP00000378786	2
HLA-DRB9	downstream gene variant	2606	ENST00000449413	?	?	2
HNRNPA1P2	upstream gene variant, downstream gene variant	1697	ENST00000432375	?	?	2
RNU6-603P	downstream gene variant	4802	ENST00000411403	?	?	1
XXbac-BPG154L12.4	upstream gene variant	3590	ENST00000425033	?	?	1
XXbac-BPG154L12.5	upstream gene variant	3163	ENST00000611838	?	?	2
XXbac-BPG246D15.9	downstream gene variant	4623	ENST00000452392	?	ENSP00000391806	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BTNL2	ENST00000454136	?	ENSP00000390613	1
BTNL2	ENST00000374993	NM_019602.1	ENSP00000364132	1
BTNL2	ENST00000544175	?	ENSP00000443364	1
BTNL2	ENST00000465865	?	ENSP00000420063	1
C6orf10	ENST00000534588	?	ENSP00000432566	2
C6orf10	ENST00000533191	NM_001286474.1	ENSP00000431199	8
C6orf10	ENST00000375007	?	ENSP00000364146	8
C6orf10	ENST00000527965	NM_001286475.1	ENSP00000435103	8
C6orf10	ENST00000375015	?	ENSP00000364155	8
C6orf10	ENST00000447241	NM_006781.4	ENSP00000415517	8
C6orf10	ENST00000442822	?	ENSP00000411164	8
C6orf10	ENST00000532023	?	ENSP00000432814	2
C6orf10	ENST00000617061	?	ENSP00000482001	8

C6orf10	ENST00000612031	?	ENSP00000480403	8
HLA-DRB9	ENST00000449413	?	?	1
XXbac-BPG154L12.4	ENST00000425033	?	?	3
XXbac-BPG154L12.5	ENST00000611838	?	?	10

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
XXbac-BPG154L12.4	ENST00000425033	?	1

Locus 207 (rs2505535)

SNiPAcard

Block annotations

Block info

genomic range	chr10:43,552,931-43,605,902
block size	52,972 bp
variant count	42 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.800 [-2.85 – 0.801]	gene(s) hit or close-by	RET
phastCons	μ = 0.048 [0 – 0.998]	eQTL gene(s)	CSGALNACT2, RET, RP11-351D16.3, ZNF33B
GERP++	μ = -0.954 [-6.48 – 2.97]	potentially regulated gene(s)	BMS1, HNRNPF, RASGEF1A, RET, RET, RP11-168L22.2, RP11-517P14.2, ZNF239
CADD score	μ = 3.994 [0.038 – 19.65]	disease gene(s)	RET, BMS1

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	benign	ClinVar	RCV000153833.3	1
Clinvar: phenotype not specified	benign	ClinVar	RCV000127776.1	1
Hirschsprung disease 1	risk factor	ClinVar	RCV000014980.1	1
Hirschsprung disease 1	risk factor	ClinVar	RCV000014967.1	1
?	HGMD curated	HGMD	CR110781	1
?	HGMD curated	HGMD	CR051713	1
?	HGMD curated	HGMD	CM020759	1
HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, 1	OMIM curated	OMIM	MIM:164761	2

Disease gene annotation

gene	trait	source DB	source entry/link
RET	RENAL AGENESIS	DECIPHER	MIM:191830
RET	Multiple endocrine neoplasia IIB	DECIPHER	MIM:162300
BMS1	APLASIA CUTIS CONGENITA, NONSYNDROMIC	OMIM	MIM:107600
RET	PHEOCHROMOCYTOMAPHEOCHROMOCYTOMA, SUSCEPTIBILITY TO	OMIM	MIM:171300
RET	CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL	OMIM	MIM:209880
RET	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIA	OMIM	MIM:171400
RET	THYROID CARCINOMA, FAMILIAL MEDULLARY	OMIM	MIM:155240
RET	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIB	OMIM	MIM:162300
RET	COLORECTAL CANCER	OMIM	MIM:114500
RET	HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, 1	OMIM	MIM:142623
RET	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550
BMS1	Circumscribed cutaneous aplasia of the vertex	OrphaNet	OrphaNet:1114
RET	Unilateral renal dysplasia	OrphaNet	OrphaNet:93172
RET	Bilateral renal dysplasia	OrphaNet	OrphaNet:93173
RET	Haddad syndrome	OrphaNet	OrphaNet:99803
RET	Hirschsprung disease	OrphaNet	OrphaNet:388
RET	Multiple endocrine neoplasia type 2A	OrphaNet	OrphaNet:247698
RET	Papillary or follicular thyroid carcinoma	OrphaNet	OrphaNet:146

RET	Multiple endocrine neoplasia type 2B	OrphaNet	OrphaNet:247709	orphanet
RET	Familial medullary thyroid carcinoma	OrphaNet	OrphaNet:99361	orphanet
RET	Bilateral renal agenesis	OrphaNet	OrphaNet:1848	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CSGALNACT2	?	ENSG00000169826	pancreas	9.10×10^{-8} (p-value)	GTEx Portal V6	34
CSGALNACT2	?	ENSG00000169826	lung	1.31×10^{-8} (p-value)	GTEx Portal V6	38
RET	?	ENSG00000165731	lung	4.00×10^{-8} (p-value)	GTEx Portal V6	39
CSGALNACT2	?	ENSG00000169826	tibial nerve	3.21×10^{-23} (p-value)	GTEx Portal V6	42
RET	?	ENSG00000165731	tibial nerve	8.42×10^{-9} (p-value)	GTEx Portal V6	40
RP11-351D16.3	?	ENSG00000273008	tibial nerve	2.66×10^{-6} (p-value)	GTEx Portal V6	5
CSGALNACT2	?	ENSG00000169826	breast	1.76×10^{-7} (p-value)	GTEx Portal V6	26
RP11-351D16.3	?	ENSG00000273008	tibial artery	1.56×10^{-6} (p-value)	GTEx Portal V6	18
CSGALNACT2	?	ENSG00000169826	tibial artery	5.33×10^{-6} (p-value)	GTEx Portal V6	12
CSGALNACT2	?	ENSG00000169826	pituitary	3.52×10^{-8} (p-value)	GTEx Portal V6	35
CSGALNACT2	?	ENSG00000169826	thyroid	7.04×10^{-11} (p-value)	GTEx Portal V6	41
CSGALNACT2	?	ENSG00000169826	testis	3.34×10^{-7} (p-value)	GTEx Portal V6	30
CSGALNACT2	?	ENSG00000169826	ovary	4.00×10^{-9} (p-value)	GTEx Portal V6	38
CSGALNACT2	?	ENSG00000169826	left ventricle	1.61×10^{-7} (p-value)	GTEx Portal V6	39
RET	?	ENSG00000165731	subcutaneous adipocytes	2.81×10^{-7} (p-value)	GTEx Portal V6	19
ZNF33B	?	ENSG00000196693	subcutaneous adipocytes	1.49×10^{-5} (p-value)	GTEx Portal V6	1
RP11-351D16.3	?	ENSG00000273008	subcutaneous adipocytes	8.39×10^{-6} (p-value)	GTEx Portal V6	9
CSGALNACT2	?	ENSG00000169826	subcutaneous adipocytes	1.48×10^{-6} (p-value)	GTEx Portal V6	22
CSGALNACT2	?	ENSG00000169826	muscularis mucosae	1.73×10^{-6} (p-value)	GTEx Portal V6	26
CSGALNACT2	?	ENSG00000169826	adrenal gland	3.70×10^{-6} (p-value)	GTEx Portal V6	14
CSGALNACT2	?	ENSG00000169826	atrial appendage	7.94×10^{-6} (p-value)	GTEx Portal V6	1
?	?	ILMN_1889106	blood	1.29×10^{-4} (p-value)	Westra et al.	2
RP11-351D16.3	?	ENSG00000273008	sun exposed skin	1.65×10^{-5} (p-value)	GTEx Portal V6	2
CSGALNACT2	?	ENSG00000169826	anterior cingulate cortex	5.75×10^{-7} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000008227		RET

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000072662	1	ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008242	RP11-517P14.2
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
ENCE00000072664	1	ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
		ENCP00000008241	HNRNPF
		ENCP00000008242	RP11-517P14.2
		ENCP00000008234	RASGEF1A
ENCE00000072665	1	ENCP00000008241	HNRNPF
		ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008242	RP11-517P14.2
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
ENCE00000072668	1	ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
		ENCP00000008241	HNRNPF
		ENCP00000008235	RP11-168L22.2
		ENCP00000008242	RP11-517P14.2
		ENCP00000008234	RASGEF1A
ENCE00000072933	1	ENCP00000008246	ZNF239

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001422878 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3, Rad21
ENSR00000115942 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	CTCF, H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3

		nervous (NH-A)	H3K27me3
		skin (NHEK)	CTCF
ENSR00000354005 (TF binding site)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K4me2, H3K27me3, Rad21
		endothelium (HUVEC)	H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		A549	H3K27me3, H3K4me3
		blood (K562)	H3K27me3, DNase1
		breast (HMEC)	H3K4me2
ENSR00000354008 (promoter flanking region)	3	embryonic stem cell (H1ESC)	USF1
		HSMMtube	DNase1, H3K27ac
		blood (K562)	H3K27me3, USF1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	DNase1
		liver (HepG2)	USF1, H3K27me3
		blood (GM12878)	USF1
		skin (NHEK)	CTCF, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RET	upstream gene variant	487	ENST00000355710	NM_020975.4	ENSP00000347942	3
RET	upstream gene variant	15	ENST00000498820	?	ENSP00000419080	4
RET	upstream gene variant	10	ENST00000615310	?	ENSP00000480088	4
RET	upstream gene variant	20	ENST00000340058	NM_020630.4	ENSP00000344798	4
RET	upstream gene variant, downstream gene variant	1235	ENST00000479913	?	?	10

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
RET	ENST00000340058	NM_020630.4	ENSP00000344798	A	gcA/gcG	1
RET	ENST00000615310	?	ENSP00000480088	A	gcA/gcG	1
RET	ENST00000355710	NM_020975.4	ENSP00000347942	A	gcA/gcG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RET	ENST00000340058	NM_020630.4	ENSP00000344798	33
RET	ENST00000615310	?	ENSP00000480088	33
RET	ENST00000355710	NM_020975.4	ENSP00000347942	33
RET	ENST00000498820	?	ENSP00000419080	33
RET	ENST00000479913	?	?	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RET	ENST00000355710	NM_020975.4	ENSP00000347942	1

Locus 208 (rs1801689)

SNiPAcard

Block annotations

Block info

genomic range	chr17:64,195,431-64,305,051
block size	109,621 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 1.057 [-1.088 – 6.439]	gene(s) hit or close-by	APOH, PRKCA, PSMD7P1
phastCons	μ = 0.241 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 1.266 [-2.42 – 5.5]	potentially regulated gene(s)	–
CADD score	μ = 8.965 [0.147 – 26.9]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
LDL cholesterol	<1.00×10-11	GWAS Catalog	24097068	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM970094	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
APOH	missense variant	ENST00000205948	NM_000042.2	ENSP00000205948	G/C	Ggc/Tgc	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001537522 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	FOSL2, Jund, PolII, H3K79me2, DNase1, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K4me3, H3K27me3, H3K36me3
		lung (IMR90)	H3K27me3
		A549	H3K4me3, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
APOH	downstream gene variant, upstream gene variant	20	ENST00000581797	?	ENSP00000463553	3

APOH	upstream gene variant	3460	ENST00000205948	NM_000042.2	ENSP00000205948	1
PRKCA	downstream gene variant	2585	ENST00000583775	?	?	1
PSMD7P1	downstream gene variant	1075	ENST00000579375	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOH	ENST00000205948	NM_000042.2	ENSP00000205948	1
APOH	ENST00000577982	?	ENSP00000464301	2
APOH	ENST00000585162	?	ENSP00000462260	1
PRKCA	ENST00000284384	?	ENSP00000284384	1
PRKCA	ENST00000578063	?	ENSP00000462087	1
PRKCA	ENST00000413366	NM_002737.2	ENSP00000408695	1
PRKCA	ENST00000583361	?	?	1

Locus 209 (rs300756)

SNiPAcard

Block annotations

Block info

genomic range	chr2:233,416-277,995
block size	44,580 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.458 [-0.646 – -0.269]	gene(s) hit or close-by	ACP1, FAM150B, SH3YL1
phastCons	μ = 0.001 [0 – 0.001]	eQTL gene(s)	SH3YL1
GERP++	μ = -3.740 [-6.45 – -1.03]	potentially regulated gene(s)	–
CADD score	μ = 0.765 [0.18 – 1.351]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SH3YL1	?	ENSG00000035115	sun exposed skin	3.77×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ACP1	1	ENST00000272065 ENST00000272067	hsa-miR-300 hsa-miR-381-3p hsa-miR-520c-3p hsa-miR-520d-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ACP1	downstream gene variant	4840	ENST00000405233	?	ENSP00000384307	1
ACP1	downstream gene variant	489	ENST00000413140	?	ENSP00000410331	1
ACP1	downstream gene variant	489	ENST00000463831	?	?	1
ACP1	downstream gene variant	626	ENST00000484464	?	?	1
ACP1	downstream gene variant	2384	ENST00000480874	?	?	1
ACP1	downstream gene variant	734	ENST00000442386	?	ENSP00000389681	1
ACP1	downstream gene variant	2795	ENST00000405364	?	ENSP00000384184	1
ACP1	downstream gene variant	620	ENST00000453390	?	ENSP00000411121	1
FAM150B	downstream gene variant	1934	ENST00000401489	?	ENSP00000385214	1
FAM150B	downstream gene variant	1956	ENST00000344414	?	ENSP00000339565	1
FAM150B	downstream gene variant	1563	ENST00000403610	NM_001002919.2	ENSP00000384604	1
FAM150B	downstream gene variant	1771	ENST00000401503	?	ENSP00000385537	1
FAM150B	downstream gene variant	1934	ENST00000619265	?	ENSP00000478180	1
FAM150B	downstream gene variant	1934	ENST00000405290	?	ENSP00000385672	1
SH3YL1	upstream gene variant	2257	ENST00000472012	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SH3YL1	ENST00000488979	?	?	1
SH3YL1	ENST00000403657	?	ENSP00000385668	1
SH3YL1	ENST00000497051	?	?	1
SH3YL1	ENST00000454318	?	ENSP00000415723	1
SH3YL1	ENST00000475027	?	?	1
SH3YL1	ENST00000405430	?	ENSP00000384269	1
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	1
SH3YL1	ENST00000468321	?	?	1
SH3YL1	ENST00000488044	?	?	1
SH3YL1	ENST00000481932	?	ENSP00000474411	1
SH3YL1	ENST00000463865	?	?	1
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	1
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	1
SH3YL1	ENST00000451005	?	ENSP00000416312	1
SH3YL1	ENST00000473104	?	?	1
SH3YL1	ENST00000479739	?	ENSP00000441266	1
SH3YL1	ENST00000403658	NM_001282682.1	ENSP00000383928	1
SH3YL1	ENST00000471948	?	?	1
SH3YL1	ENST00000431160	?	ENSP00000399519	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACP1	ENST00000272067	NM_007099.3	ENSP00000272067	1
ACP1	ENST00000272065	NM_004300.3	ENSP00000272065	1

Locus 210 (rs626457)

SNiPAcard

Block annotations

Block info

genomic range	chr1:57,375,718-57,407,484
block size	31,767 bp
variant count	33 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.457 [-3.364 – 2.562]	gene(s) hit or close-by	C8A, C8B
phastCons	μ = 0.031 [0 – 0.981]	eQTL gene(s)	C1orf168
GERP++	μ = -0.849 [-7.05 – 4.16]	potentially regulated gene(s)	C1orf168, DAB1
CADD score	μ = 3.405 [0.041 – 17.59]	disease gene(s)	C8A, C8B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C8A	COMPLEMENT COMPONENT 8 DEFICIENCY, TYPE I	OMIM	MIM:613790
C8A	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150
C8B	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C1orf168	?	ENSG00000187889	thyroid	4.86×10-6 (p-value)	GTEx Portal V6	30

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000021496	1	ENCP00000002721	DAB1
		ENCP00000002712	C1orf168
ENCE00000021498	2	ENCP00000002721	DAB1
		ENCP00000002712	C1orf168

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001520596 (enhancer)	1	liver (HepG2)	H3K4me1, FOXA1, H3K9ac, H3K27ac
		HSMMtube	H3K27me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C8A	downstream gene variant	403	ENST00000361249	NM_000562.2	ENSP00000354458	2
C8B	downstream gene variant	154	ENST00000543257	NM_001278543.1	ENSP00000442548	8
C8B	downstream gene variant	154	ENST00000535057	NM_001278544.1	ENSP00000440113	8

C8B	downstream gene variant	152	ENST00000371237	NM_000066.3	ENSP00000360281	8
C8B	downstream gene variant, upstream gene variant	84	ENST00000465658	?	?	20

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C8A	ENST00000361249	NM_000562.2	ENSP00000354458	4
C8B	ENST00000371237	NM_000066.3	ENSP00000360281	18
C8B	ENST00000535057	NM_001278544.1	ENSP00000440113	18
C8B	ENST00000543257	NM_001278543.1	ENSP00000442548	18
C8B	ENST00000465658	?	?	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C8B	ENST00000371237	NM_000066.3	ENSP00000360281	1
C8B	ENST00000535057	NM_001278544.1	ENSP00000440113	1
C8B	ENST00000543257	NM_001278543.1	ENSP00000442548	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C8B	ENST00000465658	?	1

Locus 211 (rs2070856)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,731,412-23,731,412
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.296	gene(s) hit or close-by	CST1
phastCons	0	eQTL gene(s)	CST1
GERP++	-2.91	potentially regulated gene(s)	–
CADD score	0.003	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CST1	missense variant	ENST00000398402	?	ENSP00000381439	L/P	cTg/cCg	?	?	1
CST1	missense variant	ENST00000304749	NM_001898.2	ENSP00000305731	L/P	cTg/cCg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST1	?	ENSG00000170373	tibial nerve	2.38×10^{-6} (p-value)	GTEx Portal V6	1

Locus 212 (rs8178847)

SNiPAcard

Block annotations

Block info

genomic range	chr17:64,190,994-64,235,007
block size	44,014 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.099 [-2.05 – 1.743]	gene(s) hit or close-by	APOH, CEP112, PSMD7P1
phastCons	μ = 0.053 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 0.154 [-2.89 – 3.46]	potentially regulated gene(s)	–
CADD score	μ = 3.663 [0.212 – 9.646]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR033517	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
APOH	missense variant	ENST00000205948	NM_000042.2	ENSP00000205948	2	2			2
APOH	missense variant	ENST00000581797	?	ENSP00000463553	2	2			2
APOH	missense variant	ENST00000577982	?	ENSP00000464301	2	2			2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001537522 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	FOSL2, Jund, PolII, H3K79me2, DNase1, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K4me3, H3K27me3, H3K36me3
		lung (IMR90)	H3K27me3
		A549	H3K4me3, DNase1
ENSR00001537523 (enhancer)	1	HSMMtube	H3K27me3
		liver (HepG2)	H3K27ac, H3K4me2, H3K4me1
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

APOH	downstream gene variant	1109	ENST00000577982	?	ENSP00000464301	5
APOH	downstream gene variant, upstream gene variant	67	ENST00000585162	?	ENSP00000462260	8
APOH	upstream gene variant	197	ENST00000581797	?	ENSP00000463553	4
APOH	downstream gene variant, upstream gene variant	1383	ENST00000205948	NM_000042.2	ENSP00000205948	3
CEP112	upstream gene variant	3007	ENST00000392769	NM_145036.3	ENSP00000376522	2
CEP112	upstream gene variant	3179	ENST00000537949	?	ENSP00000440775	2
CEP112	upstream gene variant	2792	ENST00000580624	?	?	2
CEP112	upstream gene variant	2797	ENST00000535342	NM_001199165.1	ENSP00000442784	2
CEP112	upstream gene variant	3021	ENST00000583358	?	ENSP00000463914	2
PSMD7P1	upstream gene variant, downstream gene variant	1284	ENST00000579375	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOH	ENST00000585162	?	ENSP00000462260	5
APOH	ENST00000581797	?	ENSP00000463553	14
APOH	ENST00000205948	NM_000042.2	ENSP00000205948	15
APOH	ENST00000577982	?	ENSP00000464301	13

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOH	ENST00000205948	NM_000042.2	ENSP00000205948	1
APOH	ENST00000577982	?	ENSP00000464301	1

Locus 213 (rs3763267)

SNiPAcard

Block annotations

Block info

genomic range	chr6:169,562,593-169,598,082
block size	35,490 bp
variant count	24 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.515 [-2.942 – 0.962]	gene(s) hit or close-by	RP11-417E7.1, RP11-417E7.2
phastCons	μ = 0.035 [0 – 0.384]	eQTL gene(s)	RP11-417E7.2
GERP++	μ = -0.601 [-3.58 – 1.47]	potentially regulated gene(s)	RP1-137D17.1, RP11-417E7.1, RP3-495K2.3, THBS2, WDR27, XXyac-YX65C7_A.2
CADD score	μ = 2.299 [0.198 – 6.035]	disease gene(s)	THBS2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
THBS2	INTERVERTEBRAL DISC DISEASE	OMIM	MIM:603932

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-417E7.2	?	ENSG00000261039	aorta	9.21×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000470093	1	ENCP00000050698	RP1-137D17.1
		ENCP00000050686	RP3-495K2.3
		ENCP00000050695	THBS2
ENCE00000470094	1	ENCP00000050689	RP11-417E7.1
		ENCP00000050698	RP1-137D17.1
		ENCP00000050686	RP3-495K2.3
		ENCP00000050695	THBS2
ENCE00000470180	1	ENCP00000050690	XXyac-YX65C7_A.2
ENCE00000470279	1	ENCP00000050700	WDR27

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001233874 (promoter flanking region)	1	NHLF	H3K36me3, H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H2AZ, H3K4me2, DNase1
		Osteobl	H3K27ac, H3K36me3

		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, H3K4me1, DNase1, H3K9ac, H3K27ac, H3K4me2
		muscle (HSMM)	H3K36me3, H3K27ac, DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	DNase1, H3K27me3
		lung (IMR90)	H4K91ac, H3K4ac, H3K36me3, H4K5ac, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K27ac
		A549	DNase1
		skin (NHEK)	H3K4me1, DNase1
ENSR00001502797 (promoter flanking region)	2	NHLF	H3K36me3, H3K27ac, H3K9ac, DNase1
		HSMMtube	DNase1, H3K27ac, H3K9ac
		Osteobl	H3K27ac, H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K27ac
		liver (HepG2)	H3K27me3
		lung (IMR90)	H4K91ac, H2BK120ac, H3K4ac, H3K36me3, H4K5ac, H3K27ac, DNase1
		nervous (NH-A)	H3K27ac, DNase1, H3K9ac
ENSR00001502798 (promoter flanking region)	5	NHLF	DNase1, H3K27ac
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	DNase1, H3K27ac
		Osteobl	H3K27ac, H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me1
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
		liver (HepG2)	H3K27me3
		nervous (NH-A)	DNase1, H3K9ac
ENSR00001502800 (promoter flanking region)	1	cervix (HeLa-S3)	H3K27me3
		HSMMtube	DNase1, H3K36me3, H3K27ac
		liver (HepG2)	H3K27me3
		blood (K562)	H3K27me3, Gabp, H2AZ
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
ENSR00001233884 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K27ac, H3K9ac, H3K4me1
		Osteobl	H3K27ac
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27ac, DNase1, H3K4me1
		muscle (HSMM)	DNase1, H3K9ac, H3K27ac
		cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00001502806 (open chromatin region)	2	muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-417E7.1	upstream gene variant	197	ENST00000449466	?	?	1
RP11-417E7.2	downstream gene variant, upstream gene variant	201	ENST00000564830	?	?	10

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-417E7.1	ENST00000439703	?	?	1
RP11-417E7.2	ENST00000564830	?	?	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-417E7.2	ENST00000564830	?	1

Locus 214 (rs10832169)

SNiPAcard

Block annotations

Block info

genomic range	chr11:14,038,621-14,121,039
block size	82,419 bp
variant count	36 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.230 [-2.046 – 1.303]	gene(s) hit or close-by	SPON1
phastCons	μ = 0.054 [0 – 0.999]	eQTL gene(s)	SPON1
GERP++	μ = 0.078 [-5.51 – 3.28]	potentially regulated gene(s)	COPB1, RP11-21L19.1, RP11-98J9.1, RP11-98J9.3, RRAS2, SPON1
CADD score	μ = 4.253 [0.29 – 19.18]	disease gene(s)	RRAS2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
RRAS2	OVARIAN CANCEROVARIAN CANCER, EPITHELIAL, INCLUDED	OMIM	MIM:167000

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SPON1	ENST00000576479	ILMN_1791890	liver	$<2.2\times10^{-16}$ (p-value)	Innocenti et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000098968	1	ENCP00000011250	SPON1
ENCE00000098923	1	ENCP00000011259	COPB1
		ENCP00000011252	RRAS2
		ENCP00000011246	RP11-98J9.3
		ENCP00000011251	RP11-21L19.1
		ENCP00000011245	RP11-98J9.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000558962 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3, Egr1
		skin (NHDF-AD)	DNase1
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1

		nervous (NH-A)	DNase1
ENSR00001568447 (promoter flanking region)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SPON1	ENST00000576479	NM_006108.3	ENSP00000460236	36

Locus 215 (rs10895987)

SNiPAcard

Block annotations

Block info

genomic range	chr11:64,876,740-64,936,719
block size	59,980 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.540 [-6.639 – 1.312]	gene(s) hit or close-by	AP003068.18, AP003068.9, HIGD1AP10, MIR6751, MRPL49, PDCL2P2, RNU2-23P, SPDYC, SYVN1, TM7SF2, VPS51
phastCons	μ = 0.041 [0 – 0.105]	eQTL gene(s)	ARL2, NAALADL1, SPDYC
GERP++	μ = -1.022 [-10.4 – 0.632]	potentially regulated gene(s)	CDCA5, MEN1, MEN1, SAC3D1, SIPA1, SYVN1
CADD score	μ = 4.642 [0.032 – 12.66]	disease gene(s)	MEN1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
MEN1	MULTIPLE ENDOCRINE NEOPLASIA, TYPE I	OMIM	MIM:131100
MEN1	HYPERPARATHYROIDISM 1	OMIM	MIM:145000
MEN1	Zollinger-Ellison syndrome	OrphaNet	OrphaNet:913
MEN1	Familial parathyroid adenoma	OrphaNet	OrphaNet:99877
MEN1	Multiple endocrine neoplasia, type 1	OrphaNet	OrphaNet:652
MEN1	Familial isolated hyperparathyroidism	OrphaNet	OrphaNet:99879

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SPDYC	?	ENSG00000204710	lung	2.92×10-6 (p-value)	GTEx Portal V6	10
SPDYC	?	ENSG00000204710	blood	2.22×10-14 (p-value)	GTEx Portal V6	11
NAALADL1	ENST00000526516	ILMN_1770963	blood	9.11×10-6 (p-value)	Westra et al.	7
NAALADL1	ENST00000532802					
NAALADL1	ENST00000528977					
NAALADL1	ENST00000533340					
NAALADL1	ENST00000531746					
NAALADL1	ENST00000356632					
NAALADL1	ENST00000534568					
NAALADL1	ENST00000530995					
NAALADL1	ENST00000533842					
NAALADL1	ENST00000532450					
NAALADL1	ENST00000533753					
NAALADL1	ENST00000358658					
NAALADL1	ENST00000355721					
NAALADL1	ENST00000340252					
NAALADL1	ENST00000529274					
ARL2	ENST00000529254	ILMN_1787879	blood	2.01×10-5 (p-value)	Westra et al.	7

ARL2	ENST00000533729					
ARL2	ENST00000529384					
ARL2	ENST00000246747					
?	?	ILMN_1777476	monocyte	2.43×10-22 (p-value)	Zeller et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000109797	1	ENCP00000012907	CDCA5
		ENCP00000012927	SYVN1
		ENCP00000013012	SIPA1
		ENCP00000012901	SAC3D1
		ENCP00000012872	MEN1 MEN1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000563760 (open chromatin region)	1	blood (GM12878)	DNase1
ENSR00000563763 (promoter flanking region)	1	endothelium (HUVEC)	DNase1, PolII, H3K27ac, Max, Cjun
		liver (HepG2)	H2AZ
		HSMMtube	H2AZ
		lung (IMR90)	H4K5ac
		blood (GM12878)	PolII, H2AZ
		blood (K562)	Max, H3K9ac, H2AZ, H3K4me3, PolII, H3K4me2, H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP003068.18	downstream gene variant	4869	ENST00000534819	?	?	1
AP003068.9	downstream gene variant	1446	ENST00000528887	?	?	1
HIGD1AP10	upstream gene variant	1104	ENST00000527837	?	?	1
MIR6751	upstream gene variant	776	ENST00000610479	?	?	1
MRPL49	downstream gene variant	3504	ENST00000531705	?	ENSP00000436740	1
MRPL49	downstream gene variant	3391	ENST00000532671	?	?	1
MRPL49	downstream gene variant	3391	ENST00000526319	?	ENSP00000434190	1
MRPL49	downstream gene variant	4576	ENST00000533943	?	ENSP00000434052	1
MRPL49	downstream gene variant	3383	ENST00000279242	NM_004927.3	ENSP00000279242	1
MRPL49	downstream gene variant	4452	ENST00000528529	?	?	1
MRPL49	downstream gene variant	3398	ENST00000524482	?	?	1
MRPL49	downstream gene variant	4750	ENST00000534078	?	ENSP00000434222	1
PDCL2P2	downstream gene variant, upstream gene variant	274	ENST00000533651	?	?	5
RNU2-23P	upstream gene variant	508	ENST00000410545	?	?	1
SPDYC	upstream gene variant	906	ENST00000377185	NM_001008778.1	ENSP00000366390	1
SYVN1	upstream gene variant	768	ENST00000530451	?	?	1
SYVN1	upstream gene variant	2950	ENST00000449943	?	?	1
SYVN1	downstream gene variant, upstream gene variant	1873	ENST00000525874	?	?	2
SYVN1	downstream gene variant, upstream gene variant	1941	ENST00000533685	?	?	2

SYVN1	upstream gene variant	2905	ENST00000294256	NM_032431.2	ENSP00000294256	1
SYVN1	downstream gene variant, upstream gene variant	241	ENST00000532771	?	ENSP00000432166	2
SYVN1	upstream gene variant	2904	ENST00000377190	NM_172230.2	ENSP00000366395	1
SYVN1	downstream gene variant, upstream gene variant	217	ENST00000531018	?	ENSP00000431215	2
SYVN1	upstream gene variant	2957	ENST00000307289	?	ENSP00000302035	1
SYVN1	downstream gene variant, upstream gene variant	520	ENST00000528487	?	ENSP00000431720	2
SYVN1	upstream gene variant	3008	ENST00000526060	?	ENSP00000436984	1
SYVN1	upstream gene variant	465	ENST00000527765	?	?	1
SYVN1	downstream gene variant, upstream gene variant	378	ENST00000529207	?	?	2
SYVN1	downstream gene variant, upstream gene variant	378	ENST00000527142	?	ENSP00000432888	2
TM7SF2	upstream gene variant	3003	ENST00000530892	?	?	1
TM7SF2	upstream gene variant	2610	ENST00000529601	?	ENSP00000435458	1
TM7SF2	upstream gene variant	2634	ENST00000530650	?	?	1
TM7SF2	upstream gene variant	2780	ENST00000524690	?	ENSP00000434598	1
TM7SF2	upstream gene variant	2730	ENST00000530750	?	ENSP00000432413	1
TM7SF2	upstream gene variant	2577	ENST00000526809	?	ENSP00000432171	1
TM7SF2	upstream gene variant	4241	ENST00000528802	?	ENSP00000436233	1
TM7SF2	upstream gene variant	2604	ENST00000534371	?	ENSP00000432187	1
TM7SF2	upstream gene variant	3946	ENST00000534667	?	?	1
TM7SF2	upstream gene variant	2616	ENST00000345348	NM_001277233.1	ENSP00000329520	1
TM7SF2	upstream gene variant	4282	ENST00000526048	?	?	1
TM7SF2	upstream gene variant	2622	ENST00000529414	?	ENSP00000433275	1
TM7SF2	upstream gene variant	3471	ENST00000527851	?	?	1
TM7SF2	upstream gene variant	4228	ENST00000527968	?	ENSP00000431685	1
TM7SF2	upstream gene variant	2618	ENST00000531321	?	ENSP00000431300	1
TM7SF2	upstream gene variant	2611	ENST00000525385	?	ENSP00000433325	1
TM7SF2	upstream gene variant	2621	ENST00000529292	?	ENSP00000436370	1
TM7SF2	upstream gene variant	2599	ENST00000524986	?	ENSP00000435972	1
TM7SF2	upstream gene variant	2607	ENST00000533646	?	?	1
TM7SF2	upstream gene variant	3437	ENST00000529233	?	?	1
TM7SF2	upstream gene variant	2604	ENST00000528026	?	?	1
TM7SF2	upstream gene variant	4103	ENST00000532328	?	?	1
TM7SF2	upstream gene variant	2623	ENST00000526085	?	ENSP00000434447	1
TM7SF2	upstream gene variant	2586	ENST00000279263	NM_003273.3	ENSP00000279263	1
TM7SF2	upstream gene variant	3376	ENST00000531029	?	?	1
TM7SF2	upstream gene variant	2604	ENST00000533766	?	?	1
VPS51	upstream gene variant	50	ENST00000534591	?	?	1
VPS51	downstream gene variant	918	ENST00000534124	?	ENSP00000436867	1
VPS51	downstream gene variant	869	ENST00000529180	?	ENSP00000435245	1
VPS51	downstream gene variant	774	ENST00000534557	?	ENSP00000435691	1
VPS51	downstream gene variant	949	ENST00000530773	?	ENSP00000434138	1

VPS51	downstream gene variant	1640	ENST00000528588	?	ENSP00000436134	1
VPS51	upstream gene variant	23	ENST00000533827	?	ENSP00000434542	1
VPS51	downstream gene variant	1383	ENST00000526578	?	ENSP00000431188	1
VPS51	upstream gene variant	1214	ENST00000530673	?	ENSP00000431317	1
VPS51	downstream gene variant	1541	ENST00000533656	?	?	1
VPS51	downstream gene variant	1248	ENST00000533487	?	ENSP00000433579	1
VPS51	upstream gene variant	76	ENST00000526856	?	ENSP00000435629	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
SYVN1	ENST00000526060	?	ENSP00000436984	S	tcA/tcG	1
SYVN1	ENST00000377190	NM_172230.2	ENSP00000366395	S	tcA/tcG	1
SYVN1	ENST00000294256	NM_032431.2	ENSP00000294256	S	tcA/tcG	1
SYVN1	ENST00000307289	?	ENSP00000302035	S	tcA/tcG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
VPS51	ENST00000279281	NM_013265.3	ENSP00000279281	1
VPS51	ENST00000527646	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SYVN1	ENST00000526121	?	1
SYVN1	ENST00000449943	?	1
VPS51	ENST00000531146	?	1

Locus 216 (rs17676662)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,262,254-34,262,254
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.135	gene(s) hit or close-by	LYZL6, RDM1
phastCons	0.001	eQTL gene(s)	–
GERP++	0.799	potentially regulated gene(s)	–
CADD score	2.777	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LYZL6	downstream gene variant	1439	ENST00000617692	?	ENSP00000481373	1
RDM1	upstream gene variant	4968	ENST00000615024	?	ENSP00000481648	1
RDM1	upstream gene variant	4523	ENST00000619368	?	ENSP00000478131	1
RDM1	upstream gene variant	4951	ENST00000615378	NM_001163125.1	ENSP00000480131	1
RDM1	upstream gene variant	4497	ENST00000619876	?	ENSP00000483890	1
RDM1	upstream gene variant	4523	ENST00000612980	NM_001163120.1	ENSP00000483387	1
RDM1	upstream gene variant	4968	ENST00000619828	NM_001163122.1	ENSP00000483933	1
RDM1	upstream gene variant	4477	ENST00000620284	NM_145654.3	ENSP00000483549	1
RDM1	upstream gene variant	4523	ENST00000615288	?	ENSP00000477869	1
RDM1	upstream gene variant	4968	ENST00000617591	NM_001163124.1	ENSP00000479622	1
RDM1	upstream gene variant	4968	ENST00000618511	?	ENSP00000477995	1
RDM1	upstream gene variant	4523	ENST00000616596	NM_001163121.1	ENSP00000478915	1
RDM1	upstream gene variant	4481	ENST00000619262	NM_001163130.1	ENSP00000479310	1
RDM1	upstream gene variant	4510	ENST00000613308	NM_001034836.1	ENSP00000482288	1
RDM1	upstream gene variant	4523	ENST00000619193	?	ENSP00000482981	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LYZL6	ENST00000615905	NM_020426.2	ENSP00000483897	1
LYZL6	ENST00000618542	NM_001199951.1	ENSP00000482955	1

Locus 217 (rs6750059)

SNiPAcard

Block annotations

Block info

genomic range	chr2:109,594,969-109,613,765
block size	18,797 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.183 [-1.417 – 2.379]	gene(s) hit or close-by	EDAR
phastCons	μ = 0.081 [0 – 0.895]	eQTL gene(s)	SULT1C3
GERP++	μ = -0.253 [-6.15 – 4.2]	potentially regulated gene(s)	EDAR, LIMS1
CADD score	μ = 5.145 [0.276 – 14.27]	disease gene(s)	EDAR

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
EDAR	HAIR MORPHOLOGY 1	OMIM	MIM:612630
EDAR	ECTODERMAL DYSPLASIA 10B, HYPOHIDROTIC/HAIR/TOOTH TYPE, [...]	OMIM	MIM:224900
EDAR	ECTODERMAL DYSPLASIA 10A, HYPOHIDROTIC/HAIR/NAIL TYPE, AUTOSOMAL [...]	OMIM	MIM:129490
EDAR	Autosomal dominant hypohidrotic ectodermal dysplasia	OrphaNet	OrphaNet:1810
EDAR	Autosomal recessive hypohidrotic ectodermal dysplasia	OrphaNet	OrphaNet:248

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SULT1C3	ENST00000329106	ILMN_1653463	b-cell	3.65×10-4 (p-value)	Fairfax et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000258714	1	ENCP00000027465	EDAR
		ENCP00000027450	LIMS1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
EDAR	upstream gene variant	4040	ENST00000409271	?	ENSP00000386371	2
EDAR	upstream gene variant	3937	ENST00000376651	?	ENSP00000365839	2
EDAR	upstream gene variant	3937	ENST00000258443	NM_022336.3	ENSP00000258443	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
EDAR	ENST00000409271	?	ENSP00000386371	8
EDAR	ENST00000376651	?	ENSP00000365839	8
EDAR	ENST00000258443	NM_022336.3	ENSP00000258443	8

Locus 218 (rs10823500)

SNiPAcard

Block annotations

Block info

genomic range	chr10:71,949,471-72,006,891
block size	57,421 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.160 [-2.43 – 1.545]	**gene(s) hit or close-by**	NPFFR1, PPA1, RP11-367H5.8
phastCons	μ = 0.015 [0 – 0.148]	**eQTL gene(s)**	AIFM2, EIF4EBP2, PPA1, SAR1A, TYSND1
GERP++	μ = -0.574 [-4.12 – 1.88]	**potentially regulated gene(s)**	CEP57L1P1
CADD score	μ = 3.477 [0.062 – 10.56]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PPA1	?	ENSG00000180817	uterus	9.44×10^{-8} (p-value)	GTEx Portal V6	13
PPA1	?	ENSG00000180817	pancreas	6.48×10^{-9} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	muscularis mucosae	2.47×10^{-10} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	muscularis mucosae	3.13×10^{-19} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	lung	8.34×10^{-11} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	lung	9.85×10^{-7} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	atrial appendage	5.31×10^{-16} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	transformed fibroblasts	1.12×10^{-9} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	transformed fibroblasts	4.94×10^{-7} (p-value)	GTEx Portal V6	13
SAR1A	?	ENSG00000079332	tibial artery	2.18×10^{-18} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	tibial artery	1.53×10^{-48} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	blood	2.42×10^{-14} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	thyroid	2.38×10^{-11} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	thyroid	8.16×10^{-11} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	skeletal muscle	2.68×10^{-6} (p-value)	GTEx Portal V6	11
PPA1	?	ENSG00000180817	skeletal muscle	1.70×10^{-63} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	unexposed skin	1.39×10^{-11} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	sun exposed skin	1.29×10^{-6} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	sun exposed skin	2.38×10^{-9} (p-value)	GTEx Portal V6	14
SAR1A	?	ENSG00000079332	aorta	1.20×10^{-7} (p-value)	GTEx Portal V6	13
PPA1	?	ENSG00000180817	aorta	1.49×10^{-25} (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	left ventricle	8.39×10^{-15} (p-value)	GTEx Portal V6	14

SAR1A	?	ENSG00000079332	subcutaneous adipocytes	1.78×10-13 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	caudate basal ganglia	1.64×10-7 (p-value)	GTEx Portal V6	13
SAR1A	?	ENSG00000079332	visceral adipocytes	1.76×10-7 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	stomach	4.51×10-12 (p-value)	GTEx Portal V6	14
AIFM2	?	ENSG00000042286	cortex	1.05×10-12 (p-value)	GTEx Portal V6	13
PPA1	?	ENSG00000180817	cortex	4.92×10-7 (p-value)	GTEx Portal V6	12
SAR1A	?	ENSG00000079332	tibial nerve	2.12×10-13 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	tibial nerve	3.18×10-7 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	sigmoid colon	2.95×10-9 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	pituitary	3.66×10-12 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	gastroesophageal junction	2.42×10-11 (p-value)	GTEx Portal V6	14
PPA1	?	ENSG00000180817	testis	1.35×10-8 (p-value)	GTEx Portal V6	14
TYSND1	ENST00000287078	ILMN_2325185	blood	5.01×10-5 (p-value)	Westra et al.	6
TYSND1	ENST00000335494					
TYSND1	ENST00000494143					
TYSND1	ENST00000479086					
EIF4EBP2	ENST00000373218	ILMN_1728083	blood	1.92×10-6 (p-value)	Westra et al.	6
AIFM2	ENST00000307864	ILMN_1712639	blood	9.47×10-7 (p-value)	Westra et al.	1
AIFM2	ENST00000613322					
PPA1	ENST00000610026	ILMN_1805827	monocyte	8.11×10-58 (p-value)	Zeller et al.	1
PPA1	ENST00000373232					
SAR1A	?	ENSG00000079332	esophagus mucosa	3.61×10-6 (p-value)	GTEx Portal V6	2
AIFM2	?	ENSG00000042286	cerebellum	1.25×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000077597	1	ENCP00000008751	CEP57L1P1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001424487 (promoter flanking region)	1	NHLF	H3K27ac, H3K4me1, DNase1
		HSMMtube	DNase1
		Osteobl	H3K4me1, H3K4me2, H3K27ac
		blood (K562)	Max, H3K27me3
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1
		muscle (HSMM)	H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K4me1, Max
		monocytes (Monocytes-CD14+)	H3K4me1
		lung (IMR90)	DNase1, H3K27ac
		A549	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1
ENSR00001424491	1	embryonic stem cell (H1ESC)	H3K36me3

(promoter flanking region)		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	DNase1
ENSR00000356568	2	embryonic stem cell (H1ESC)	DNase1, USF1, H3K4me3, H3K4me2, TAF1, H3K27me3, PolII, Sin3Ak20, H3K9ac, Yy1
(promoter)		HSMMtube	DNase1, H2AZ, H3K4me3, H3K4me2
		blood (K562)	Egr1, H3K27ac, Max, CTCF, H3K9ac, H3K79me2, HEY1, H2AZ, TAF1, Cmyc, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	PolII, H3K79me2, TAF1, USF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3, H3K36me3, DNase1
		blood (GM12878)	PolII, H3K9ac, H3K36me3, H3K4me2, H3K27ac, H2AZ, DNase1, Cmyc, CTCF, Yy1, H3K79me2, H3K4me3, USF1
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K4me2, H3K4me3, DNase1, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		breast (HMEC)	H3K4me2, DNase1, H3K27ac, H3K4me3, H3K9ac
		cervix (HeLa-S3)	DNase1, H3K79me2, PolII, Cmyc, CTCF, H3K36me3, H3K4me3, Max, TAF1, Nrf1, H3K9ac, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, H3K4me2, DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K9ac, PolII, DNase1, Cmyc
		A549	H3K27ac, H3K36me3, DNase1, H3K9ac, H3K4me3, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NPFFR1	downstream gene variant	194	ENST00000277942	NM_022146.4	ENSP00000277942	3
PPA1	downstream gene variant, upstream gene variant	57	ENST00000460755	?	?	3
PPA1	downstream gene variant	3635	ENST00000625364	?	ENSP00000486162	1
PPA1	downstream gene variant, upstream gene variant	1	ENST00000373230	?	ENSP00000362327	2
PPA1	downstream gene variant	391	ENST00000495346	?	?	1
RP11-367H5.8	upstream gene variant	4613	ENST00000435591	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PPA1	ENST00000610026	?	?	2
PPA1	ENST00000460755	?	?	4
PPA1	ENST00000373232	NM_021129.3	ENSP00000362329	9
PPA1	ENST00000373230	?	ENSP00000362327	6
PPA1	ENST00000625364	?	ENSP00000486162	7
PPA1	ENST00000495346	?	?	6

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PPA1	ENST00000373232	NM_021129.3	ENSP00000362329	1

PPA1	ENST00000625364	?	ENSP00000486162	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PPA1	ENST00000610026	?	1

Locus 219 (rs6509558)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,115,833-52,128,795
block size	12,963 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.206 [-0.505 – 0.179]	gene(s) hit or close-by	RPL9P33, SIGLEC5, SIGLEC5
phastCons	μ = 0.013 [0 – 0.032]	eQTL gene(s)	SIGLEC14
GERP++	μ = -0.546 [-2.66 – 1.26]	potentially regulated gene(s)	–
CADD score	μ = 2.647 [1.725 – 4.652]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC14	?	ENSG00000254415	subcutaneous adipocytes	8.46×10^{-7} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RPL9P33	downstream gene variant	2995	ENST00000484311	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC5	ENST00000599649	?	ENSP00000470259	4
SIGLEC5	ENST00000534261	?	ENSP00000473238	4
SIGLEC5	ENST00000570106	NM_003830.3	ENSP00000455510	4
SIGLEC5	ENST00000429354	?	ENSP00000415200	4

Locus 220 (rs1461672)

SNiPAcard

Block annotations

Block info

genomic range	chr11:99,042,908-99,225,548
block size	182,641 bp
variant count	39 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.530 [-3.042 – 0.496]	gene(s) hit or close-by	CNTN5
phastCons	μ = 0.007 [0 – 0.089]	eQTL gene(s)	–
GERP++	μ = -0.741 [-7.09 – 3.04]	potentially regulated gene(s)	–
CADD score	μ = 2.568 [0.042 – 8.437]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
2-hydroxyisobutyrate	9.87×10-7	Metabolomics GWAS Server	24816252	4

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001606326 (promoter flanking region)	1	skin (NHDF-AD)	DNase1
ENSR00000567685 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Jund, Cjun
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN5	ENST00000528682	NM_001243270.1	ENSP00000436185	39
CNTN5	ENST00000527185	NM_001243271.1	ENSP00000433575	39
CNTN5	ENST00000524871	NM_014361.3	ENSP00000435637	39
CNTN5	ENST00000528727	?	?	39

Locus 221 (rs1710354)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,802,140-51,802,650
block size	511 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.125 [-1.962 – -0.287]	**gene(s) hit or close-by**	–
phastCons	μ = 0.003 [0 – 0.005]	**eQTL gene(s)**	CD33, ETFB
GERP++	μ = -0.726 [-1.13 – -0.323]	**potentially regulated gene(s)**	–
CADD score	μ = 1.387 [1.232 – 1.542]	**disease gene(s)**	ETFB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
oleate (18:1n9)	3.00×10^{-5}	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link	
ETFB	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	OMIM	MIM:231680	OMIM
ETFB	glutaric aciduria type 2B	DECIPHER	MIM:231680	OMIM
ETFB	Multiple acyl-CoA dehydrogenation deficiency, mild type	OrphaNet	OrphaNet:394532	orphanet
ETFB	Multiple acyl-CoA dehydrogenation deficiency, severe neonatal type	OrphaNet	OrphaNet:394529	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD33	ENST00000262262	ILMN_1747622	blood	6.28×10^{-7} (p-value)	Westra et al.	1
CD33	ENST00000600557					
CD33	ENST00000601785					
CD33	ENST00000421133					
CD33	ENST00000598473					
ETFB	ENST00000596253	ILMN_1729374	blood	8.37×10^{-5} (p-value)	Westra et al.	2
ETFB	ENST00000593992					
ETFB	ENST00000309244					

Locus 222 (rs4803378)

SNiPAcard

Block annotations

Block info

genomic range	chr19:41,333,441-41,392,490
block size	59,050 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.487 [-1.433 – 1.377]	gene(s) hit or close-by	CTC-490E21.10, CTC-490E21.11, CTC-490E21.12, CYP2A6, CYP2A7, CYP2F2P, CYP2G1P
phastCons	μ = 0.050 [0 – 0.484]	eQTL gene(s)	CYP2B7P
GERP++	μ = -0.607 [-4.56 – 0.89]	potentially regulated gene(s)	–
CADD score	μ = 2.379 [0.27 – 8.637]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CYP2B7P	?	ENSG00000256612	lung	4.19×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001419215 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K27me3
		Osteobl	H3K4me2, H3K27me3
		blood (K562)	Nrsf, H3K4me3, H3K4me2, H3K4me1, H2AZ, USF1, DNase1, Gata2, H3K27ac, Max, H3K9ac
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Max, Cmyc
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	USF1, H3K4me1, HNF4A, FOXA1, DNase1
		lung (IMR90)	DNase1, H3K36me3
		A549	H3K27me3, DNase1
		nervous (NH-A)	DNase1
ENSR00001648169 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTC-490E21.10	downstream gene variant, upstream gene variant	396	ENST00000596135	?	?	7
CTC-490E21.11	upstream gene variant, downstream gene variant	475	ENST00000623128	?	?	11
CYP2A6	downstream gene variant, upstream gene variant	30	ENST00000599960	?	?	7
CYP2A6	downstream gene variant	1024	ENST00000301141	NM_000762.5	ENSP00000301141	3

CYP2A6	downstream gene variant	91	ENST00000600495	?	ENSP00000472905	4
CYP2A6	downstream gene variant	40	ENST00000596719	?	?	2
CYP2A6	downstream gene variant, upstream gene variant	930	ENST00000610301	?	ENSP00000477899	5
CYP2A7	upstream gene variant	388	ENST00000301146	NM_000764.2	ENSP00000301146	3
CYP2A7	upstream gene variant	3050	ENST00000598264	?	?	1
CYP2A7	upstream gene variant	388	ENST00000291764	NM_030589.2	ENSP00000291764	3
CYP2A7	upstream gene variant	909	ENST00000594332	?	ENSP00000470933	3
CYP2F2P	upstream gene variant	764	ENST00000437853	?	?	3
CYP2G1P	upstream gene variant	4241	ENST00000252909	?	?	2
CYP2G1P	upstream gene variant	4634	ENST00000597833	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTC-490E21.10	ENST00000596135	?	?	4
CTC-490E21.12	ENST00000601627	?	ENSP00000469533	19
CYP2A6	ENST00000596719	?	?	3
CYP2A6	ENST00000301141	NM_000762.5	ENSP00000301141	4
CYP2A6	ENST00000610301	?	ENSP00000477899	4
CYP2A6	ENST00000600495	?	ENSP00000472905	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CYP2A6	ENST00000596719	?	1

Locus 223 (rs10421385)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,677,669-51,704,168
block size	26,500 bp
variant count	28 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.878 [-4.83 – 0.386]	gene(s) hit or close-by	CTD-3187F8.14, CTD-3187F8.7, SIGLEC17P, SIGLEC20P, SIGLEC21P
phastCons	μ = 0.074 [0 – 0.973]	eQTL gene(s)	SIGLEC20P
GERP++	μ = -0.931 [-5.39 – 1.22]	potentially regulated gene(s)	–
CADD score	μ = 2.421 [0.144 – 14.52]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC20P	?	ENSG00000268336	testis	7.34×10^{-6} (p-value)	GTEx Portal V6	6

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001649028 (open chromatin region)	3	embryonic stem cell (H1ESC)	DNase1
		A549	H3K27me3
		blood (K562)	H3K27me3
ENSR00000217706 (open chromatin region)	1	A549	H3K27me3
		blood (K562)	DNase1, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-3187F8.14	upstream gene variant	1388	ENST00000600074	?	?	9
CTD-3187F8.7	downstream gene variant, upstream gene variant	645	ENST00000595913	?	?	12
SIGLEC17P	downstream gene variant	984	ENST00000614626	?	?	2
SIGLEC17P	downstream gene variant	889	ENST00000618545	?	?	2
SIGLEC17P	downstream gene variant	891	ENST00000619985	?	?	2
SIGLEC17P	downstream gene variant	913	ENST00000611992	?	?	2
SIGLEC20P	upstream gene variant, downstream gene variant	35	ENST00000595243	?	?	11
SIGLEC21P	upstream gene variant	220	ENST00000598990	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-3187F8.14	ENST00000600074	?	?	2
CTD-3187F8.7	ENST00000595913	?	?	1

SIGLEC20P	ENST00000595243	?	?	10
SIGLEC21P	ENST00000598990	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SIGLEC20P	ENST00000595243	?	2

Locus 224 (rs922182)

SNiPAcard

Block annotations

Block info

genomic range	chr15:64,275,409-64,284,214
block size	8,806 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.239 [-1.896 – 0.659]	gene(s) hit or close-by	DAPK2
phastCons	μ = 0.041 [0 – 0.404]	eQTL gene(s)	DAPK2, RP11-111E14.1
GERP++	μ = -1.744 [-7.76 – 1.57]	potentially regulated gene(s)	–
CADD score	μ = 6.194 [0.564 – 12.3]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DAPK2	?	ENSG00000035664	aorta	2.07×10^{-13} (p-value)	GTEx Portal V6	9
RP11-111E14.1	?	ENSG00000259351	aorta	2.21×10^{-6} (p-value)	GTEx Portal V6	4
DAPK2	?	ENSG00000035664	muscularis mucosae	4.12×10^{-6} (p-value)	GTEx Portal V6	4
DAPK2	?	ENSG00000035664	tibial artery	2.45×10^{-5} (p-value)	GTEx Portal V6	1
RP11-111E14.1	?	ENSG00000259351	tibial nerve	2.91×10^{-5} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	15	no	0.000	1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000021558 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF
		blood (GM12878)	Rad21, CTCF, H3K36me3
		blood (K562)	Rad21, CTCF
		skin (NHEK)	CTCF
ENSR00000409563 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	DNase1, Rad21, BCL11A, H3K27ac, H3K4me2, H3K4me1, BATF, PU1, PolII
		blood (K562)	DNase1
		skin (NHDF-AD)	DNase1
ENSR00001023121 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	H3K4me1, H3K27ac
		lung (IMR90)	H3K27me3
		blood (K562)	PU1
ENSR00001453015 (promoter flanking region)	1	blood (GM12878)	H3K4me1, H3K27ac
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DAPK2	downstream gene variant	159	ENST00000559306	?	ENSP00000452970	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DAPK2	ENST00000561162	?	ENSP00000453284	11
DAPK2	ENST00000559897	?	?	11
DAPK2	ENST00000261891	?	ENSP00000261891	11
DAPK2	ENST00000559007	?	ENSP00000453639	11
DAPK2	ENST00000457488	NM_014326.3	ENSP00000408277	11
DAPK2	ENST00000558069	?	ENSP00000453235	11
DAPK2	ENST00000558482	?	?	11
DAPK2	ENST00000612884	?	ENSP00000484390	11
DAPK2	ENST00000559306	?	ENSP00000452970	9

Locus 225 (rs135150)

SNiPAcard

Block annotations

Block info

genomic range	chr22:33,168,562-33,175,471
block size	6,910 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.974 [-1.494 – -0.145]	**gene(s) hit or close-by**	LL22NC01-116C6.1, SYN3
phastCons	μ = 0.000 [0 – 0.001]	**eQTL gene(s)**	–
GERP++	μ = -1.872 [-5.06 – 0.149]	**potentially regulated gene(s)**	–
CADD score	μ = 0.806 [0.213 – 1.804]	**disease gene(s)**	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LL22NC01-116C6.1	upstream gene variant	3692	ENST00000444982	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SYN3	ENST00000462268	?	?	5
SYN3	ENST00000358763	NM_003490.3, NM_001135774.1, NM_133633.2	ENSP00000351614	5

Locus 226 (rs7160336)

SNiPAcard

Block annotations

Block info

genomic range	chr14:74,475,796-74,661,497
block size	185,702 bp
variant count	59 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.490 [-3.067 – 2.691]	**gene(s) hit or close-by**	ALDH6A1, CCDC176, ENTPD5, LIN52, RN7SL530P, RP5-892G5.2
phastCons	μ = 0.080 [0 – 1]	**eQTL gene(s)**	ALDH6A1, COQ6, LIN52, PTGR2, RP5-1021I20.2
GERP++	μ = -0.003 [-7.89 – 5.78]	**potentially regulated gene(s)**	–
CADD score	μ = 4.529 [0.029 – 17.19]	**disease gene(s)**	COQ6, ALDH6A1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
COQ6	COENZYME Q10 DEFICIENCY, PRIMARY, 6	OMIM	MIM:614650
ALDH6A1	METHYLMALONATE SEMIALDEHYDE DEHYDROGENASE DEFICIENCY	OMIM	MIM:614105
COQ6	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	OrphaNet	OrphaNet:280406
ALDH6A1	Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency	OrphaNet	OrphaNet:289307

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CCDC176	missense variant	ENST00000464394	?	ENSP00000451659	D/V	gAt/gTt	tolerated	benign	1
CCDC176	missense variant	ENST00000394009	NM_025057.2	ENSP00000377577	D/V	gAt/gTt	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
COQ6	ENST00000554320	ILMN_1783985	skin	7.62×10^{-7} (p-value)	MuTHER consortium	20
COQ6	ENST00000394026		blood	3.46×10^{-6} (p-value)	MuTHER consortium	20
COQ6	ENST00000556299					
COQ6	ENST00000629426					
COQ6	ENST00000555511					
COQ6	ENST00000238709					
COQ6	ENST00000334571					
COQ6	ENST00000556588					
COQ6	ENST00000554341					
COQ6	ENST00000554920	ILMN_1664367	blood	1.25×10^{-5} (p-value)	MuTHER consortium	19
COQ6	ENST00000554193					
COQ6	ENST00000553922					
COQ6	ENST00000555552					

COQ6	ENST00000394026					
COQ6	ENST00000555511					
COQ6	ENST00000334571					
COQ6	ENST00000238709					
COQ6	ENST00000556300					
COQ6	ENST00000555196					
LIN52	ENST00000554076	ILMN_1797055	skin	1.75×10-12 (p-value)	MuTHER consortium	20
LIN52	ENST00000555028		blood	1.95×10-9 (p-value)	MuTHER consortium	20
			adipocyte	5.39×10-10 (p-value)	MuTHER consortium	20
LIN52	?	ENSG00000205659	pancreas	1.25×10-9 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	lung	9.83×10-11 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	lung	1.06×10-7 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	atrial appendage	1.17×10-6 (p-value)	GTEx Portal V6	55
PTGR2	?	ENSG00000140043	atrial appendage	4.62×10-7 (p-value)	GTEx Portal V6	38
LIN52	?	ENSG00000205659	transformed fibroblasts	2.58×10-7 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	tibial artery	2.29×10-8 (p-value)	GTEx Portal V6	58
LIN52	?	ENSG00000205659	breast	2.75×10-7 (p-value)	GTEx Portal V6	57
COQ6	?	ENSG00000119723	thyroid	2.86×10-9 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	thyroid	2.43×10-12 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	thyroid	1.22×10-7 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	skeletal muscle	6.97×10-8 (p-value)	GTEx Portal V6	59
COQ6	?	ENSG00000119723	skeletal muscle	6.68×10-6 (p-value)	GTEx Portal V6	15
LIN52	?	ENSG00000205659	EBV lymphocytes	3.22×10-9 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	unexposed skin	1.03×10-6 (p-value)	GTEx Portal V6	58
LIN52	?	ENSG00000205659	sun exposed skin	3.00×10-11 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	aorta	1.11×10-10 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	aorta	1.23×10-8 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	subcutaneous adipocytes	2.30×10-14 (p-value)	GTEx Portal V6	59
RP5-1021I20.2	?	ENSG00000258586	tibial nerve	1.51×10-8 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	tibial nerve	8.71×10-16 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	esophagus mucosa	6.24×10-8 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	esophagus mucosa	7.60×10-6 (p-value)	GTEx Portal V6	4
LIN52	?	ENSG00000205659	adrenal gland	4.23×10-8 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	testis	1.87×10-11 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	visceral adipocytes	7.09×10-6 (p-value)	GTEx Portal V6	2
LIN52	?	ENSG00000205659	muscularis mucosae	1.67×10-7 (p-value)	GTEx Portal V6	21
LIN52	?	ENSG00000205659	coronary artery	4.72×10-6 (p-value)	GTEx Portal V6	1

LIN52	ENST00000554076	ILMN_1797055	monocyte	2.78×10^{-9} (p-value)	Fairfax et al.	4
LIN52	ENST00000555028		b-cell	1.34×10^{-7} (p-value)	Fairfax et al.	4
ALDH6A1	ENST00000554501	ILMN_2096985	blood	5.18×10^{-5} (p-value)	Westra et al.	1
ALDH6A1	ENST00000350259					
ALDH6A1	ENST00000555126					
ALDH6A1	ENST00000553458					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000420800 (TF binding site)	1	endothelium (HUVEC)	Cjun, DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
ENSR00000098742 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	DNase1
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, DNase1
ENSR00000420807 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac
		cervix (HeLa-S3)	DNase1, TAF1, H3K27ac
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		skin (NHEK)	H3K27ac
ENSR00000420810 (enhancer)	1	endothelium (HUVEC)	DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ALDH6A1	upstream gene variant	97	ENST00000554501	?	?	1
ALDH6A1	upstream gene variant	126	ENST00000553814	?	?	1
ALDH6A1	upstream gene variant	99	ENST00000350259	NM_001278593.1	ENSP00000342564	1
ALDH6A1	upstream gene variant	54	ENST00000554231	?	?	1
ALDH6A1	downstream gene variant, upstream gene variant	99	ENST00000556852	?	?	2

ALDH6A1	upstream gene variant	53	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	1
ALDH6A1	upstream gene variant	2347	ENST00000555126	?	ENSP00000452081	2
CCDC176	downstream gene variant	4691	ENST00000492247	?	?	1
CCDC176	upstream gene variant, downstream gene variant	1683	ENST00000492026	?	?	2
CCDC176	downstream gene variant	2456	ENST00000463558	?	ENSP00000480689	1
CCDC176	upstream gene variant	96	ENST00000477986	?	?	2
LIN52	upstream gene variant, downstream gene variant	959	ENST00000554289	?	?	4
LIN52	upstream gene variant	410	ENST00000554938	?	ENSP00000452513	3
LIN52	upstream gene variant, downstream gene variant	417	ENST00000554076	?	?	6
LIN52	upstream gene variant	414	ENST00000553404	?	?	3
LIN52	upstream gene variant	250	ENST00000555028	NM_001024674.2	ENSP00000451812	3
RN7SL530P	upstream gene variant, downstream gene variant	833	ENST00000485097	?	?	3
RP5-892G5.2	upstream gene variant, downstream gene variant	1971	ENST00000555526	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ALDH6A1	ENST00000553814	?	?	5
ALDH6A1	ENST00000556852	?	?	4
ALDH6A1	ENST00000554501	?	?	9
ALDH6A1	ENST00000554231	?	?	5
ALDH6A1	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	9
ALDH6A1	ENST00000555126	?	ENSP00000452081	4
ALDH6A1	ENST00000350259	NM_001278593.1	ENSP00000342564	9
CCDC176	ENST00000394009	NM_025057.2	ENSP00000377577	12
CCDC176	ENST00000489323	?	?	8
CCDC176	ENST00000463558	?	ENSP00000480689	6
CCDC176	ENST00000492026	?	?	16
CCDC176	ENST00000464394	?	ENSP00000451659	8
ENTPD5	ENST00000556108	?	?	2
ENTPD5	ENST00000557325	?	ENSP00000451810	2
ENTPD5	ENST00000554664	?	?	2
ENTPD5	ENST00000557681	?	?	2
ENTPD5	ENST00000556242	?	ENSP00000451483	2
ENTPD5	ENST00000334696	NM_001249.2	ENSP00000335246	2
LIN52	ENST00000554938	?	ENSP00000452513	38
LIN52	ENST00000555028	NM_001024674.2	ENSP00000451812	38
LIN52	ENST00000554076	?	?	7
LIN52	ENST00000553404	?	?	38
LIN52	ENST00000554280	?	?	2

LIN52	ENST00000554289	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ALDH6A1	ENST00000554501	?	1
CCDC176	ENST00000489323	?	1
CCDC176	ENST00000492026	?	2
CCDC176	ENST00000477986	?	1
ENTPD5	ENST00000557681	?	1

Locus 227 (rs7184253)

SNiPAcard

Block annotations

Block info

genomic range	chr16:67,308,279-67,549,746
block size	241,468 bp
variant count	114 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.038 [-3.819 – 3.611]	gene(s) hit or close-by	AGRP, ATP6V0D1, CTD-2012K14.8, FAM65A, HSD11B2, KCTD19, LRRC36, PLEKHG4, RN7SKP118, RNU1-123P, RP11-297D21.2, RP11-297D21.4, SLC9A5, TPPP3, ZDHHC1
phastCons	μ = 0.206 [0 – 1]	eQTL gene(s)	AGRP, ATP6V0D1, C16orf86, CTCF, DPEP2, DUS2, EXOC3L1, HSD11B2, LCAT, LRRC36, NFATC3, PLA2G15, RANBP10, TRADD, ZDHHC1
GERP++	μ = 0.189 [-11.3 – 5.69]	potentially regulated gene(s)	ATP6V0D1, C16orf70, C16orf86, CBFB, CES4A, EDC4, ELMO3, EXOC3L1, FAM65A, FBXL8, GFOD2, HSD11B2, HSF4, LCAT, NUTF2, SLC12A4, TPPP3, TRADD, ZDHHC1
CADD score	μ = 6.295 [0.006 – 32]	disease gene(s)	PLEKHG4, HSD11B2, LCAT, HSF4, CTCF, CBFB, AGRP

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Leanness, inherited	association	ClinVar	RCV000007754.2	1
Obesity, late-onset	association	ClinVar	RCV000007753.1	1
?	HGMD curated	HGMD	CM057710	1
?	HGMD curated	HGMD	CM023599	1
OBESITY, LATE-ONSET	OMIM curated	OMIM	MIM:602311	1

Disease gene annotation

gene	trait	source DB	source entry/link
PLEKHG4	Spinocerebellar ataxia type 4	OrphaNet	OrphaNet:98765
HSD11B2	APPARENT MINERALOCORTICOID EXCESS	OrphaNet	OrphaNet:320
LCAT	FISH-EYE DISEASE	OrphaNet	OrphaNet:79292
LCAT	Familial LCAT deficiency	OrphaNet	OrphaNet:79293
HSF4	Zonular cataract	OrphaNet	OrphaNet:98995
CTCF	Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome	OrphaNet	OrphaNet:363611
CBFB	Acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)	OrphaNet	OrphaNet:98829
HSD11B2	APPARENT MINERALOCORTICOID EXCESS	OMIM	MIM:218030
AGRP	OBESITYLEANNESS, INCLUDED	OMIM	MIM:601665
LCAT	FISH-EYE DISEASE	OMIM	MIM:136120
LCAT	LECITHIN:CHOLESTEROL ACYLTRANSFERASE DEFICIENCY	OMIM	MIM:245900
HSF4	CATARACT 5, MULTIPLE TYPES	OMIM	MIM:116800
CTCF	MENTAL RETARDATION, AUTOSOMAL DOMINANT 21	OMIM	MIM:615502
HSF4	cataract Marner type (CAM)	DECIPHER	MIM:116800
HSF4	cataract zonular HSF4-related (CZ-HSF4)	DECIPHER	MIM:116800

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
AGRP	missense variant	ENST00000290953	NM_001138.1	ENSP00000290953	T/A	Act/Gct	?	?	1
KCTD19	missense variant	ENST00000304372	NM_001100915.1	ENSP00000305702	K/E	Aag/Gag	?	?	1
LRRC36	missense variant	ENST00000435835	?	ENSP00000411122	2	2	tolerated, ?	benign, ?	2
LRRC36	missense variant	ENST00000565019	?	ENSP00000464675	R/P	cGt/cCt	?	benign	1
LRRC36	missense variant	ENST00000563189	NM_001161575.1	ENSP00000455103	2	3	?, tolerated	benign, ?	3
LRRC36	missense variant	ENST00000569499	?	ENSP00000456814	R/P	cGt/cCt	?	benign	1
LRRC36	missense variant	ENST00000329956	NM_018296.5	ENSP00000329943	2	3	?, tolerated	benign, ?	3
LRRC36	missense variant	ENST00000568010	?	ENSP00000455018	R/P	cGt/cCt	?	benign	1
LRRC36	missense variant	ENST00000561948	?	ENSP00000456347	R/P	cGt/cCt	?	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LRRC36	?	ENSG00000159708	skeletal muscle	3.14×10^{-17} (p-value)	GTEx Portal V6	111
C16orf86	?	ENSG00000159761	skeletal muscle	1.11×10^{-5} (p-value)	GTEx Portal V6	24
RANBP10	?	ENSG00000141084	cortex	2.02×10^{-6} (p-value)	GTEx Portal V6	14
RANBP10	?	ENSG00000141084	tibial nerve	6.59×10^{-11} (p-value)	GTEx Portal V6	111
LRRC36	?	ENSG00000159708	thyroid	3.22×10^{-15} (p-value)	GTEx Portal V6	111
ZDHHC1	?	ENSG00000159714	thyroid	1.15×10^{-7} (p-value)	GTEx Portal V6	105
HSD11B2	?	ENSG00000176387	thyroid	6.59×10^{-17} (p-value)	GTEx Portal V6	110
DUS2	?	ENSG00000167264	thyroid	1.69×10^{-5} (p-value)	GTEx Portal V6	10
RANBP10	?	ENSG00000141084	thyroid	3.72×10^{-5} (p-value)	GTEx Portal V6	1
EXOC3L1	?	ENSG00000179044	thyroid	1.60×10^{-5} (p-value)	GTEx Portal V6	1
LRRC36	?	ENSG00000159708	visceral adipocytes	5.09×10^{-9} (p-value)	GTEx Portal V6	55
LRRC36	?	ENSG00000159708	atrial appendage	3.59×10^{-6} (p-value)	GTEx Portal V6	21
RANBP10	?	ENSG00000141084	tibial artery	1.57×10^{-7} (p-value)	GTEx Portal V6	83
CTCF	?	ENSG00000102974	tibial artery	1.06×10^{-5} (p-value)	GTEx Portal V6	10
EXOC3L1	?	ENSG00000179044	tibial artery	1.61×10^{-5} (p-value)	GTEx Portal V6	3
DPEP2	?	ENSG00000167261	blood	2.93×10^{-8} (p-value)	GTEx Portal V6	22
PLA2G15	?	ENSG00000103066	blood	1.70×10^{-6} (p-value)	GTEx Portal V6	14
DUS2	?	ENSG00000167264	blood	9.27×10^{-9} (p-value)	GTEx Portal V6	36
ATP6V0D1	?	ENSG00000159720	blood	1.22×10^{-15} (p-value)	GTEx Portal V6	58
ZDHHC1	?	ENSG00000159714	transformed fibroblasts	1.65×10^{-5} (p-value)	GTEx Portal V6	11
DUS2	ENST00000432752	ILMN_1811650	monocyte	4.02×10^{-9} (p-value)	Fairfax et al.	10

DUS2	ENST00000565263					
DUS2	ENST00000561965					
DUS2	ENST00000358896					
DUS2	ENST00000565980					
ATP6V0D1	ENST00000568620	ILMN_1795826	monocyte	2.88×10-15 (p-value)	Fairfax et al.	10
ATP6V0D1	ENST00000567694		b-cell	6.99×10-8 (p-value)	Fairfax et al.	10
ATP6V0D1	ENST00000563064					
ATP6V0D1	ENST00000561852					
ATP6V0D1	ENST00000602876					
ATP6V0D1	ENST00000290949					
ATP6V0D1	ENST00000567170					
ATP6V0D1	ENST00000564615					
ATP6V0D1	ENST00000426604					
ATP6V0D1	ENST00000565835					
ATP6V0D1	ENST00000563305					
AGRP	ENST00000290953	ILMN_1714253	monocyte	4.43×10-10 (p-value)	Fairfax et al.	10
DUS2	ENST00000432752	ILMN_1811650	monocyte	4.71×10-45 (p-value)	Zeller et al.	5
DUS2	ENST00000565263					
DUS2	ENST00000561965					
DUS2	ENST00000565980					
DUS2	ENST00000358896					
ZDHHC1	ENST00000565726	ILMN_2094313	monocyte	1.76×10-13 (p-value)	Zeller et al.	3
ZDHHC1	ENST00000566075					
ZDHHC1	ENST00000348579					
TRADD	ENST00000486556	ILMN_1765851	monocyte	8.13×10-26 (p-value)	Zeller et al.	5
TRADD	ENST00000566247					
TRADD	ENST00000563348					
TRADD	ENST00000345057					
C16orf86	ENST00000403458	ILMN_1697800	monocyte	2.94×10-14 (p-value)	Zeller et al.	5
C16orf86	ENST00000459925					
C16orf86	ENST00000602974					
C16orf86	ENST00000602365					
C16orf86	ENST00000445068					
RANBP10	?	ENSG00000141084	subcutaneous adipocytes	4.19×10-8 (p-value)	GTEx Portal V6	35
LRRC36	?	ENSG00000159708	subcutaneous adipocytes	9.09×10-7 (p-value)	GTEx Portal V6	35
RANBP10	?	ENSG00000141084	muscularis mucosae	1.09×10-5 (p-value)	GTEx Portal V6	50
LRRC36	?	ENSG00000159708	muscularis mucosae	7.71×10-7 (p-value)	GTEx Portal V6	32
LRRC36	?	ENSG00000159708	pancreas	3.84×10-15 (p-value)	GTEx Portal V6	35
LRRC36	?	ENSG00000159708	breast	1.39×10-7 (p-value)	GTEx Portal V6	35
ATP6V0D1	?	ENSG00000159720	EBV lymphocytes	6.90×10-7 (p-value)	GTEx Portal V6	35
ATP6V0D1	?	ENSG00000159720	spleen	9.21×10-11 (p-value)	GTEx Portal V6	34
LCAT	ENST00000264005	ILMN_1815102	skin	3.97×10-6 (p-value)	MuTHER consortium	18
LCAT	ENST00000573538					
LCAT	ENST00000570980					
LCAT	ENST00000570369					
AGRP	ENST00000290953	ILMN_1714253	blood	1.29×10-5 (p-value)	MuTHER consortium	17
TRADD	ENST00000486556	ILMN_1765851	skin	4.86×10-8 (p-value)	MuTHER consortium	18

TRADD	ENST00000566247		blood	5.33×10-11 (p-value)	MuTHER consortium	18
TRADD	ENST00000563348		adipocyte	3.93×10-18 (p-value)	MuTHER consortium	18
TRADD	ENST00000345057					
C16orf86	ENST00000403458	ILMN_1697800	blood	2.18×10-8 (p-value)	MuTHER consortium	18
C16orf86	ENST00000459925					
C16orf86	ENST00000602974					
C16orf86	ENST00000602365					
C16orf86	ENST00000445068					
NFATC3	ENST00000346183	ILMN_1800976	skin	1.55×10-5 (p-value)	MuTHER consortium	9
NFATC3	ENST00000329524					
NFATC3	ENST00000539828					
NFATC3	ENST00000566301					
NFATC3	ENST00000379165					
NFATC3	ENST00000563796					
NFATC3	ENST00000570212					
NFATC3	ENST00000563288					
NFATC3	ENST00000549350					
NFATC3	ENST00000349223					
NFATC3	ENST00000563319					
NFATC3	ENST00000575270					
NFATC3	ENST00000569766					
NFATC3	ENST00000567152					
NFATC3	ENST00000553077					
NFATC3	ENST00000562926					
NFATC3	ENST00000535127					
NFATC3	ENST00000346183	ILMN_2360028	skin	4.68×10-6 (p-value)	MuTHER consortium	8
NFATC3	ENST00000329524					
NFATC3	ENST00000539828					
NFATC3	ENST00000379165					
NFATC3	ENST00000566301					
NFATC3	ENST00000563796					
NFATC3	ENST00000570212					
NFATC3	ENST00000563288					
NFATC3	ENST00000549350					
NFATC3	ENST00000575270					
NFATC3	ENST00000349223					
NFATC3	ENST00000563319					
NFATC3	ENST00000569766					
NFATC3	ENST00000567152					
NFATC3	ENST00000553077					
NFATC3	ENST00000562926					
NFATC3	ENST00000535127					
ATP6V0D1	?	ENSG00000159720	esophagus mucosa	6.60×10-6 (p-value)	GTEx Portal V6	2
ZDHHC1	?	ENSG00000159714	testis	3.79×10-6 (p-value)	GTEx Portal V6	6
AGRP	?	ENSG00000159723	blood	2.95×10-3 (q-value)	SeeQTL DB (HapMap)	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ATP6V0D1	ENST00000568620	ILMN_1795826	chr16	monocyte	2.82×10-15 (p-value)	Zeller et al.	5
ATP6V0D1	ENST00000563064						
ATP6V0D1	ENST00000567694						
ATP6V0D1	ENST00000602876						

ATP6V0D1	ENST00000561852						
ATP6V0D1	ENST00000290949						
ATP6V0D1	ENST00000567170						
ATP6V0D1	ENST00000564615						
ATP6V0D1	ENST00000565835						
ATP6V0D1	ENST00000426604						
ATP6V0D1	ENST00000563305						
AGRP	ENST00000290953	ILMN_1714253	chr16	monocyte	5.25×10^{-30} (p-value)	Zeller et al.	5

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
USF1	MA0281.1	5	yes	-0.163	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000192385	1	ENCP00000021593	EXOC3L1
		ENCP00000021592	HSF4
		ENCP00000021577	CES4A
ENCE00000192543	1	ENCP00000021598	TPPP3
ENCE00000192544	1	ENCP00000021598	TPPP3
		ENCP00000021601	ATP6V0D1
ENCE00000192549	1	ENCP00000021614	EDC4
		ENCP00000021601	ATP6V0D1
		ENCP00000021598	TPPP3
ENCE00000192432	1	ENCP00000021581	C16orf70
		ENCP00000021594	ELMO3
		ENCP00000021586	HSF4 FBXL8
		ENCP00000021613	NUTF2
		ENCP00000021599	ZDHHC1
		ENCP00000021603	FAM65A
		ENCP00000021618	LCAT
		ENCP00000021587	HSF4
		ENCP00000021600	HSD11B2
		ENCP00000021580	CBFB
		ENCP00000021588	HSF4
ENCE00000192693	1	ENCP00000021614	EDC4
ENCE00000192435	3	ENCP00000021609	GFOD2
		ENCP00000021581	C16orf70
		ENCP00000021613	NUTF2
		ENCP00000021608	C16orf86
		ENCP00000021618	LCAT
		ENCP00000021603	FAM65A
		ENCP00000021580	CBFB
		ENCP00000021619	SLC12A4
		ENCP00000021583	TRADD FBXL8
		ENCP00000021588	HSF4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001634343 (enhancer)	2	liver (HepG2)	H3K4me1
		HSMMtube	H3K27me3
ENSR00000508958 (promoter flanking region)	1	cervix (HeLa-S3)	H3K27ac, H3K4me1, DNase1
		endothelium (HUVEC)	Cjun
		HSMMtube	H3K27me3
ENSR00000508959 (CTCF binding site)	1	embryonic stem cell (H1ESC)	USF1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	USF1, H3K4me1, H3K27ac, CTCF, DNase1
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1, H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001634345 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001634346 (enhancer)	1	endothelium (HUVEC)	Cjun
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
ENSR00001634348 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001634351 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1, H3K4me1
		HSMMtube	DNase1
		Osteobl	H3K27me3
		blood (K562)	DNase1, H3K4me1
		breast (HMEC)	H3K4me1, H3K4me2
		muscle (HSMM)	H3K4me1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3, H3K9ac, DNase1, H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me2, H3K27me3, DNase1
		blood (GM12878)	H3K27me3
		skin (NHEK)	H3K4me1
ENSR00000508965 (promoter flanking region)	1	embryonic stem cell (H1ESC)	CTCF, Rad21, Sin3Ak20, H3K4me2, H3K27me3, H3K4me1, DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27me3
		blood (K562)	Egr1, H3K4me1, DNase1
		skin (NHDF-AD)	DNase1
		breast (HMEC)	CTCF, H3K4me2, H3K4me1, DNase1
		muscle (HSMM)	H3K4me1, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K27me3, DNase1
		liver (HepG2)	CTCF, H3K4me1, H3K4me2, H3K27me3, DNase1
		blood (GM12878)	H3K27me3
		A549	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, CTCF, DNase1
ENSR00001634352 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, CTCF, Rad21, DNase1
		HSMMtube	DNase1
		liver (HepG2)	CTCF, H3K4me1
		skin (NHEK)	CTCF, DNase1
		breast (HMEC)	CTCF
		muscle (HSMM)	H3K4me1
ENSR00000508968	3	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21

(promoter flanking region)		HSMMtube	H3K4me1, H3K27ac, DNase1
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	H3K4me1, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me1, H2AZ, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	CTCF, H3K27me3
		lung (IMR90)	DNase1, H3K36me3
		nervous (NH-A)	H3K9ac, DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, DNase1
		NHLF	H3K27ac, DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me1, H3K4me2
		cervix (HeLa-S3)	CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	H3K36me3
ENSR00000272763	1	NHLF	H3K27ac
(CTCF binding site)		embryonic stem cell (H1ESC)	Rad21, CTCF
		Osteobl	H3K27ac
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	H3K4me1, DNase1, H3K27ac
		breast (HMEC)	H3K4me1
		muscle (HSMM)	H3K4me1, H3K27ac, H3K36me3
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	CTCF
		skin (NHEK)	H3K4me1
ENSR00000508970	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
(enhancer)		Osteobl	H3K36me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	H3K36me3
		liver (HepG2)	H3K4me1
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00001634357	2	embryonic stem cell (H1ESC)	H4K5ac, H3K27ac, H3K27me3, H3K4me1, H3K4me2
(open chromatin region)		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00000052856	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21, H3K27me3
(CTCF binding site)		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, H3K4me2, H3K9ac, CTCF, H3K27me3, DNase1
		blood (GM12878)	DNase1, Rad21, CTCF
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF, H3K27me3
		Osteobl	CTCF, H3K27me3
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3, DNase1
		A549	CTCF
ENSR00001506017	2	embryonic stem cell (H1ESC)	CTCF, Rad21, H3K36me3
(CTCF binding site)		blood (DND-41)	H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3

		blood (GM12878)	PolII, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001634363 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (K562)	CTCF, H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	HNF4G, RXRA, H3K4me1, H3K4me2, HNF4A, H3K9ac, CTCF, H3K27ac, Cmyc, H3K36me3, DNase1
		lung (IMR90)	H3K36me3, H3K27ac
		blood (GM12878)	DNase1, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3, CTCF
ENSR00001634364 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, Yy1, DNase1
		blood (K562)	CTCF, H3K36me3
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, H3K27ac, CTCF, DNase1
		blood (GM12878)	DNase1, Yy1, CTCF, H3K36me3
		lung (IMR90)	H3K36me3, CTCF
		skin (NHEK)	CTCF, H3K36me3
		NHLF	CTCF
		Osteobl	CTCF, H3K36me3
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H4K20me1, H3K36me3
		endothelium (HUVEC)	DNase1, H3K36me3
		A549	CTCF, H3K36me3
ENSR00000508978 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, Yy1, DNase1
		HSMMtube	DNase1
		blood (K562)	Rad21, CTCF, H3K36me3
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	CTCF, H3K36me3, DNase1, H3K27ac, H3K4me1, Rad21
		lung (IMR90)	H3K27ac, H3K36me3, CTCF
		blood (GM12878)	DNase1, Yy1, Rad21, CTCF, H3K36me3
		skin (NHEK)	CTCF, H3K36me3
		NHLF	CTCF
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1
		A549	CTCF, H3K36me3
ENSR00001634366 (promoter flanking region)	1	NHLF	CTCF
		embryonic stem cell (H1ESC)	CTCF, DNase1
		HSMMtube	DNase1
		Osteobl	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H3K36me3
		skin (NHEK)	CTCF
ENSR00000052861 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, Jund, CTCF, DNase1
		HSMMtube	CTCF, DNase1

		blood (K562)	FOSL1, Rad21, CTCFL, H4K20me1, CTCF, DNase1
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	H3K79me2, CTCF, DNase1
		liver (HepG2)	FOSL2, Jund, Rad21, H3K4me1, CTCF, DNase1
		lung (IMR90)	H3K79me1, H3K36me3, CTCF
		blood (GM12878)	DNase1, Rad21, H3K79me2, CTCF
		nervous (NH-A)	H4K20me1, CTCF
		skin (NHEK)	CTCF, H3K36me3, DNase1
		NHLF	CTCF
		Osteobl	H4K20me1, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1, Jund
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1, DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, Cjun, CTCF, DNase1
		A549	CTCF
ENSR00001634368 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21, Jund, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	FOSL1, Rad21, CTCFL, H4K20me1, CTCF, DNase1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	H3K79me2, CTCF, DNase1
		liver (HepG2)	FOSL2, Jund, Rad21, H3K4me1, CTCF, DNase1
		lung (IMR90)	H3K79me1, CTCF
		blood (GM12878)	CTCF, DNase1, Rad21, H3K79me2
		nervous (NH-A)	H4K20me1, CTCF
		skin (NHEK)	DNase1, CTCF, H3K36me3
		NHLF	CTCF
		Osteobl	H4K20me1, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	Jund, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, CTCF, DNase1
		A549	CTCF
ENSR00000508981 (enhancer)	1	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		lung (IMR90)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001634370 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3
ENSR00001506024 (promoter flanking region)	2	cervix (HeLa-S3)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		embryonic stem cell (H1ESC)	H3K27me3
		blood (DND-41)	H3K36me3
		blood (K562)	Max, USF1, DNase1
		skin (NHDF-AD)	DNase1
ENSR00001634371 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
ENSR00000508990 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCFL, CTCF
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	DNase1, CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	BCLAF1, H3K4me1, DNase1, Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, H3K27ac, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF

		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, DNase1
ENSR00001634373	3	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
(promoter flanking region)		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCFL, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	DNase1, CTCF
		liver (HepG2)	Rad21, CTCF, H3K27me3
		blood (GM12878)	BCLAF1, H3K4me1, DNase1, Rad21, H3K27ac, H3K4me2, CTCF
		lung (IMR90)	CTCF, DNase1
		nervous (NH-A)	DNase1, CTCF, H3K4me2
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	H3K27ac, H3K4me2, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	Jund, H3K27ac, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF
		endothelium (HUVEC)	H3K36me3, Cjun, CTCF
		A549	CTCF, DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ATP6V0D1	1	ENST00000290949 ENST00000563305 ENST00000565835 ENST00000568620 ENST00000602876	hsa-miR-132-3p hsa-miR-212-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AGRP	downstream gene variant, upstream gene variant	1737	ENST00000290953	NM_001138.1	ENSP00000290953	6
ATP6V0D1	downstream gene variant, upstream gene variant	457	ENST00000561852	?	ENSP00000457744	8
ATP6V0D1	downstream gene variant, upstream gene variant	470	ENST00000563064	?	ENSP00000457105	8
ATP6V0D1	downstream gene variant, upstream gene variant	124	ENST00000540149	?	ENSP00000441282	8
ATP6V0D1	downstream gene variant, upstream gene variant	493	ENST00000567170	?	?	7
ATP6V0D1	downstream gene variant, upstream gene variant	1885	ENST00000568298	?	?	6
ATP6V0D1	downstream gene variant	61	ENST00000426604	?	ENSP00000393910	4
ATP6V0D1	downstream gene variant, upstream gene variant	1306	ENST00000561658	?	?	7
ATP6V0D1	downstream gene variant, upstream gene variant	2025	ENST00000563305	?	?	3
ATP6V0D1	downstream gene variant	431	ENST00000568101	?	ENSP00000457104	2
ATP6V0D1	downstream gene variant, upstream gene variant	473	ENST00000564615	?	ENSP00000455802	6
ATP6V0D1	downstream gene variant, upstream gene variant	268	ENST00000565835	?	ENSP00000463328	7
ATP6V0D1	downstream gene variant, upstream gene variant	756	ENST00000564788	?	ENSP00000455695	8
ATP6V0D1	downstream gene variant	701	ENST00000566322	?	ENSP00000455446	4
ATP6V0D1	downstream gene variant, upstream gene variant	1932	ENST00000602876	?	ENSP00000473515	6
ATP6V0D1	downstream gene variant, upstream gene variant	189	ENST00000567694	?	?	7
ATP6V0D1	downstream gene variant, upstream gene variant	1805	ENST00000290949	NM_004691.4	ENSP00000290949	6
ATP6V0D1	downstream gene variant, upstream gene variant	937	ENST00000568620	?	?	5
ATP6V0D1	downstream gene variant, upstream gene variant	736	ENST00000564191	?	ENSP00000456695	8

CTD-2012K14.8	downstream gene variant	2019	ENST00000613438	?	?	3
CTD-2012K14.8	downstream gene variant	2107	ENST00000621378	?	?	3
FAM65A	upstream gene variant	2575	ENST00000562116	?	ENSP00000455239	3
HSD11B2	upstream gene variant, downstream gene variant	1919	ENST00000566606	?	ENSP00000473429	4
HSD11B2	upstream gene variant, downstream gene variant	500	ENST00000326152	NM_000196.3	ENSP00000316786	4
HSD11B2	upstream gene variant, downstream gene variant	1792	ENST00000567684	?	?	4
HSD11B2	upstream gene variant, downstream gene variant	2646	ENST00000569303	?	?	5
KCTD19	downstream gene variant, upstream gene variant	1234	ENST00000566295	?	ENSP00000462732	6
KCTD19	downstream gene variant, upstream gene variant	1040	ENST00000567976	?	ENSP00000458045	4
KCTD19	downstream gene variant, upstream gene variant	592	ENST00000561625	?	?	7
KCTD19	downstream gene variant, upstream gene variant	602	ENST00000569333	?	?	7
KCTD19	downstream gene variant, upstream gene variant	568	ENST00000304372	NM_001100915.1	ENSP00000305702	7
KCTD19	downstream gene variant	3350	ENST00000562841	?	ENSP00000463249	1
KCTD19	downstream gene variant, upstream gene variant	617	ENST00000562721	?	ENSP00000462122	7
KCTD19	downstream gene variant, upstream gene variant	578	ENST00000566392	?	?	7
KCTD19	upstream gene variant	2160	ENST00000562860	?	?	3
KCTD19	downstream gene variant, upstream gene variant	605	ENST00000570049	?	?	7
KCTD19	downstream gene variant, upstream gene variant	571	ENST00000568736	?	?	7
LRRC36	upstream gene variant, downstream gene variant	2848	ENST00000569228	?	?	4
LRRC36	upstream gene variant, downstream gene variant	1510	ENST00000566558	?	?	7
LRRC36	upstream gene variant, downstream gene variant	756	ENST00000435835	?	ENSP00000411122	3
LRRC36	upstream gene variant, downstream gene variant	1155	ENST00000561948	?	ENSP00000456347	5
LRRC36	upstream gene variant, downstream gene variant	464	ENST00000569552	?	ENSP00000454309	2
LRRC36	upstream gene variant, downstream gene variant	1038	ENST00000329956	NM_018296.5	ENSP00000329943	5
LRRC36	upstream gene variant, downstream gene variant	1157	ENST00000570075	?	ENSP00000454794	5
LRRC36	upstream gene variant, downstream gene variant	729	ENST00000563189	NM_001161575.1	ENSP00000455103	3
LRRC36	upstream gene variant, downstream gene variant	1035	ENST00000565019	?	ENSP00000464675	5
LRRC36	upstream gene variant, downstream gene variant	741	ENST00000567823	?	ENSP00000456164	3
LRRC36	upstream gene variant, downstream gene variant	729	ENST00000567723	?	ENSP00000455799	3
LRRC36	upstream gene variant, downstream gene variant	638	ENST00000561821	?	ENSP00000456517	2
LRRC36	upstream gene variant, downstream gene variant	276	ENST00000569121	?	?	6
LRRC36	upstream gene variant, downstream gene variant	721	ENST00000568010	?	ENSP00000455018	3
LRRC36	upstream gene variant, downstream gene variant	33	ENST00000563303	?	?	7
LRRC36	upstream gene variant, downstream gene variant	2468	ENST00000569499	?	ENSP00000456814	4
LRRC36	upstream gene variant, downstream gene variant	72	ENST00000568804	?	ENSP00000464367	6
LRRC36	upstream gene variant, downstream gene variant	703	ENST00000567211	?	ENSP00000455211	2
PLEKHG4	upstream gene variant, downstream gene variant	1949	ENST00000393966	?	ENSP00000462601	3

PLEKHG4	upstream gene variant, downstream gene variant	1489	ENST00000562289	?	?	3
PLEKHG4	upstream gene variant, downstream gene variant	1525	ENST00000563969	?	ENSP00000457086	4
PLEKHG4	upstream gene variant	2561	ENST00000565899	?	ENSP00000455423	2
PLEKHG4	upstream gene variant, downstream gene variant	1525	ENST00000360461	NM_001129727.1, NM_015432.3	ENSP00000353646	4
PLEKHG4	upstream gene variant	3238	ENST00000565773	?	ENSP00000455876	2
PLEKHG4	upstream gene variant	3551	ENST00000562744	?	ENSP00000455622	2
PLEKHG4	upstream gene variant	3957	ENST00000568621	?	ENSP00000456873	1
PLEKHG4	upstream gene variant, downstream gene variant	1867	ENST00000567136	?	ENSP00000455228	3
PLEKHG4	upstream gene variant, downstream gene variant	1525	ENST00000427155	NM_001129728.1	ENSP00000401118	4
PLEKHG4	upstream gene variant, downstream gene variant	1817	ENST00000569875	?	ENSP00000464631	3
PLEKHG4	upstream gene variant	4000	ENST00000567938	?	ENSP00000455740	1
PLEKHG4	upstream gene variant, downstream gene variant	1531	ENST00000379344	NM_001129729.1	ENSP00000368649	3
PLEKHG4	upstream gene variant	2224	ENST00000562144	?	ENSP00000454348	2
PLEKHG4	upstream gene variant, downstream gene variant	1505	ENST00000450733	NM_001129731.1	ENSP00000398030	3
RN7SKP118	downstream gene variant, upstream gene variant	963	ENST00000364331	?	?	4
RNU1-123P	downstream gene variant, upstream gene variant	472	ENST00000458950	?	?	6
RP11-297D21.2	downstream gene variant, upstream gene variant	2887	ENST00000567261	?	?	4
RP11-297D21.4	upstream gene variant, downstream gene variant	1086	ENST00000602592	?	?	9
RP11-297D21.4	upstream gene variant	966	ENST00000602476	?	?	5
RP11-297D21.4	upstream gene variant, downstream gene variant	881	ENST00000602596	?	?	7
SLC9A5	downstream gene variant	2318	ENST00000564812	?	ENSP00000455058	2
SLC9A5	downstream gene variant	2186	ENST00000299798	NM_004594.2	ENSP00000299798	2
SLC9A5	downstream gene variant	2186	ENST00000563723	?	?	2
SLC9A5	downstream gene variant	2186	ENST00000564704	?	?	2
SLC9A5	downstream gene variant	3396	ENST00000566626	?	ENSP00000462404	2
TPPP3	downstream gene variant, upstream gene variant	2036	ENST00000564104	?	ENSP00000462435	6
TPPP3	downstream gene variant, upstream gene variant	471	ENST00000290942	NM_016140.3	ENSP00000290942	7
TPPP3	downstream gene variant, upstream gene variant	498	ENST00000562206	?	ENSP00000457275	7
TPPP3	downstream gene variant, upstream gene variant	454	ENST00000393957	NM_015964.3	ENSP00000377529	7
TPPP3	downstream gene variant, upstream gene variant	260	ENST00000561537	?	?	7
ZDHHC1	downstream gene variant, upstream gene variant	117	ENST00000565726	?	ENSP00000459264	6
ZDHHC1	downstream gene variant, upstream gene variant	15	ENST00000348579	NM_013304.2	ENSP00000340299	5
ZDHHC1	downstream gene variant, upstream gene variant	1203	ENST00000568650	?	?	4
ZDHHC1	downstream gene variant, upstream gene variant	1162	ENST00000567311	?	?	4
ZDHHC1	downstream gene variant, upstream gene variant	1083	ENST00000562122	?	?	7
ZDHHC1	downstream gene variant, upstream gene variant	21	ENST00000566075	?	?	5

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
AGRP	ENST00000290953	NM_001138.1	ENSP00000290953	E	gaA/gaG	1
HSD11B2	ENST00000326152	NM_000196.3	ENSP00000316786	2	2	2
TPPP3	ENST00000290942	NM_016140.3	ENSP00000290942	S	agT/agC	1
TPPP3	ENST00000393957	NM_015964.3	ENSP00000377529	S	agT/agC	1
TPPP3	ENST00000562206	?	ENSP00000457275	S	agT/agC	1
TPPP3	ENST00000564104	?	ENSP00000462435	S	agT/agC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V0D1	ENST00000565835	?	ENSP00000463328	16
ATP6V0D1	ENST00000426604	?	ENSP00000393910	8
ATP6V0D1	ENST00000564191	?	ENSP00000456695	16
ATP6V0D1	ENST00000564615	?	ENSP00000455802	6
ATP6V0D1	ENST00000290949	NM_004691.4	ENSP00000290949	16
ATP6V0D1	ENST00000567170	?	?	1
ATP6V0D1	ENST00000564788	?	ENSP00000455695	16
ATP6V0D1	ENST00000566322	?	ENSP00000455446	8
ATP6V0D1	ENST00000540149	?	ENSP00000441282	16
ATP6V0D1	ENST00000602876	?	ENSP00000473515	16
ATP6V0D1	ENST00000568101	?	ENSP00000457104	7
ATP6V0D1	ENST00000568298	?	?	10
ATP6V0D1	ENST00000563064	?	ENSP00000457105	16
ATP6V0D1	ENST00000561658	?	?	10
ATP6V0D1	ENST00000561852	?	ENSP00000457744	16
KCTD19	ENST00000570049	?	?	9
KCTD19	ENST00000566392	?	?	9
KCTD19	ENST00000562841	?	ENSP00000463249	2
KCTD19	ENST00000569333	?	?	9
KCTD19	ENST00000304372	NM_001100915.1	ENSP00000305702	8
KCTD19	ENST00000562860	?	?	4
KCTD19	ENST00000567976	?	ENSP00000458045	3
KCTD19	ENST00000568736	?	?	6
KCTD19	ENST00000562721	?	ENSP00000462122	6
KCTD19	ENST00000561625	?	?	6
KCTD19	ENST00000566295	?	ENSP00000462732	3
LRRC36	ENST00000435835	?	ENSP00000411122	18
LRRC36	ENST00000569228	?	?	28
LRRC36	ENST00000568804	?	ENSP00000464367	25
LRRC36	ENST00000567723	?	ENSP00000455799	19

LRRC36	ENST00000569552	?	ENSP00000454309	12
LRRC36	ENST00000567211	?	ENSP00000455211	13
LRRC36	ENST00000563189	NM_001161575.1	ENSP00000455103	17
LRRC36	ENST00000329956	NM_018296.5	ENSP00000329943	32
LRRC36	ENST00000561948	?	ENSP00000456347	25
LRRC36	ENST00000569499	?	ENSP00000456814	26
LRRC36	ENST00000565019	?	ENSP00000464675	33
LRRC36	ENST00000570075	?	ENSP00000454794	26
LRRC36	ENST00000568010	?	ENSP00000455018	18
LRRC36	ENST00000563303	?	?	16
LRRC36	ENST00000561821	?	ENSP00000456517	12
LRRC36	ENST00000566558	?	?	15
LRRC36	ENST00000567823	?	ENSP00000456164	19
PLEKHG4	ENST00000563969	?	ENSP00000457086	1
PLEKHG4	ENST00000379344	NM_001129729.1	ENSP00000368649	1
PLEKHG4	ENST00000393966	?	ENSP00000462601	1
PLEKHG4	ENST00000360461	NM_001129727.1, NM_015432.3	ENSP00000353646	1
PLEKHG4	ENST00000427155	NM_001129728.1	ENSP00000401118	1
PLEKHG4	ENST00000450733	NM_001129731.1	ENSP00000398030	1
RP11-297D21.4	ENST00000602476	?	?	3
RP11-297D21.4	ENST00000602596	?	?	2
TPPP3	ENST00000290942	NM_016140.3	ENSP00000290942	1
TPPP3	ENST00000393957	NM_015964.3	ENSP00000377529	1
TPPP3	ENST00000562206	?	ENSP00000457275	1
TPPP3	ENST00000564104	?	ENSP00000462435	1
ZDHHC1	ENST00000568650	?	?	9
ZDHHC1	ENST00000566075	?	?	1
ZDHHC1	ENST00000348579	NM_013304.2	ENSP00000340299	12
ZDHHC1	ENST00000567311	?	?	1
ZDHHC1	ENST00000562122	?	?	3
ZDHHC1	ENST00000565726	?	ENSP00000459264	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V0D1	ENST00000565835	?	ENSP00000463328	1
ATP6V0D1	ENST00000290949	NM_004691.4	ENSP00000290949	2
ATP6V0D1	ENST00000602876	?	ENSP00000473515	2
HSD11B2	ENST00000566606	?	ENSP00000473429	2
LRRC36	ENST00000567723	?	ENSP00000455799	3
LRRC36	ENST00000567211	?	ENSP00000455211	1
LRRC36	ENST00000565019	?	ENSP00000464675	2
LRRC36	ENST00000570075	?	ENSP00000454794	1

LRRC36	ENST00000568010	?	ENSP00000455018	2
LRRC36	ENST00000561821	?	ENSP00000456517	1
LRRC36	ENST00000567823	?	ENSP00000456164	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ATP6V0D1	ENST00000568620	?	2
ATP6V0D1	ENST00000563305	?	2
ATP6V0D1	ENST00000568298	?	2
HSD11B2	ENST00000567684	?	2
KCTD19	ENST00000570049	?	2
KCTD19	ENST00000566392	?	2
KCTD19	ENST00000569333	?	2
LRRC36	ENST00000569228	?	1
RNU1-123P	ENST00000458950	?	1
TPPP3	ENST00000561537	?	1

Locus 228 (rs12997015)

SNiPAcard

Block annotations

Block info

genomic range	chr2:103,042,401-103,042,401
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.217	gene(s) hit or close-by	IL18RAP
phastCons	0	eQTL gene(s)	–
GERP++	-1.87	potentially regulated gene(s)	AC007278.2, IL18R1
CADD score	4.509	disease gene(s)	–

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000257447	1	ENCP00000027319	IL18R1
		ENCP00000027323	AC007278.2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL18RAP	downstream gene variant	2067	ENST00000450855	?	ENSP00000389815	1
IL18RAP	downstream gene variant	1751	ENST00000497795	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL18RAP	ENST00000409369	?	ENSP00000387201	1
IL18RAP	ENST00000264260	NM_003853.2	ENSP00000264260	1

Locus 229 (rs429017)

SNiPAcard

Block annotations

Block info

genomic range	chr5:41,263,091-41,266,229
block size	3,139 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.503 [-2.33 – 0.563]	gene(s) hit or close-by	C6
phastCons	μ = 0.014 [0 – 0.112]	eQTL gene(s)	C7, PLCXD3
GERP++	μ = -0.409 [-6.2 – 3.32]	potentially regulated gene(s)	PLCXD3
CADD score	μ = 4.149 [0.977 – 9.545]	disease gene(s)	C7, C6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
C7	COMPLEMENT COMPONENT 7 DEFICIENCY	OMIM	MIM:610102	OMIM
C6	COMPLEMENT COMPONENT 6 DEFICIENCY	OMIM	MIM:612446	OMIM
C7	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150	orphanet
C6	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PLCXD3	?	ENSG00000182836	muscularis mucosae	2.99×10^{-7} (p-value)	GTEx Portal V6	9
C7	?	ENSG00000112936	skeletal muscle	9.97×10^{-8} (p-value)	GTEx Portal V6	9
PLCXD3	?	ENSG00000182836	tibial nerve	2.08×10^{-7} (p-value)	GTEx Portal V6	9
PLCXD3	?	ENSG00000182836	sun exposed skin	5.99×10^{-6} (p-value)	GTEx Portal V6	9
PLCXD3	?	ENSG00000182836	lung	1.57×10^{-5} (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000407607	1	ENCP00000043972	PLCXD3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C6	upstream gene variant	1551	ENST00000263413	NM_001115131.2	ENSP00000263413	9

Locus 230 (rs7439493)

SNiPAcard

Block annotations

Block info

genomic range	chr4:110,649,891-110,658,856
block size	8,966 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.257 [-2.61 – 0.539]	**gene(s) hit or close-by**	CFI, PLA2G12A
phastCons	μ = 0.021 [0 – 0.118]	**eQTL gene(s)**	CASP6, PLA2G12A
GERP++	μ = -0.596 [-4.35 – 1.04]	**potentially regulated gene(s)**	CFI, CFI
CADD score	μ = 2.770 [0.01 – 4.375]	**disease gene(s)**	CFI

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Macular degeneration	1.59×10-6	dbGaP	pha002890	dbGaP	1
Macular degeneration	1.26×10-6	dbGaP	pha002869	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
CFI	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 3	OMIM	MIM:612923	OMIM
CFI	COMPLEMENT FACTOR I DEFICIENCY	OMIM	MIM:610984	OMIM
CFI	MACULAR DEGENERATION, AGE-RELATED, 13	OMIM	MIM:615439	OMIM
CFI	Immunodeficiency with factor I anomaly	OrphaNet	OrphaNet:200418	orphanet
CFI	Atypical hemolytic uremic syndrome with I factor anomaly	OrphaNet	OrphaNet:93580	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CASP6	?	ENSG00000138794	cerebellar hemisphere	1.87×10-8 (p-value)	GTEx Portal V6	8
CASP6	?	ENSG00000138794	atrial appendage	1.39×10-6 (p-value)	GTEx Portal V6	7
PLA2G12A	?	ENSG00000123739	tibial nerve	6.77×10-22 (p-value)	GTEx Portal V6	8
CASP6	?	ENSG00000138794	cerebellum	1.45×10-8 (p-value)	GTEx Portal V6	8
PLA2G12A	?	ENSG00000123739	transformed fibroblasts	4.97×10-8 (p-value)	GTEx Portal V6	8
PLA2G12A	?	ENSG00000123739	esophagus mucosa	2.23×10-12 (p-value)	GTEx Portal V6	8
PLA2G12A	?	ENSG00000123739	subcutaneous adipocytes	7.24×10-13 (p-value)	GTEx Portal V6	8
PLA2G12A	?	ENSG00000123739	tibial artery	1.79×10-5 (p-value)	GTEx Portal V6	1
PLA2G12A	?	ENSG00000123739	vagina	2.08×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000388317	1	ENCP00000042049	CFI CFI

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001434100 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3
		HSMMtube	H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K27ac, Max, MEF2A, H3K9ac, USF1, H3K79me2, H2AZ, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2
		liver (HepG2)	PolII, TAF1, USF1, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		blood (GM12878)	DNase1, PolII, H2AZ, H3K4me3, USF1, ZEB1, H3K4me2, H3K9ac
		lung (IMR90)	H3K4me2, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K27ac, H3K9ac, H3K4me3
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K9ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K4me3, H3K79me2, PolII, DNase1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K4me2, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, PolII, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFI	downstream gene variant	2994	ENST00000394634	NM_000204.3	ENSP00000378130	1
CFI	downstream gene variant	2994	ENST00000504853	?	?	1
CFI	downstream gene variant	3013	ENST00000618244	?	ENSP00000483416	1
CFI	downstream gene variant	4176	ENST00000515512	?	?	1
CFI	downstream gene variant	3024	ENST00000512148	?	ENSP00000427438	1
CFI	downstream gene variant	2996	ENST00000394635	?	ENSP00000378131	1
PLA2G12A	upstream gene variant	1604	ENST00000502283	?	ENSP00000425274	5
PLA2G12A	upstream gene variant	1501	ENST00000243501	NM_030821.4	ENSP00000243501	5
PLA2G12A	upstream gene variant	1622	ENST00000507961	?	ENSP00000424021	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLA2G12A	ENST00000507961	?	ENSP00000424021	1
PLA2G12A	ENST00000243501	NM_030821.4	ENSP00000243501	1
PLA2G12A	ENST00000502283	?	ENSP00000425274	1

Locus 231 (rs2193873)

SNiPAcard

Block annotations

Block info

genomic range	chr3:190,313,507-190,314,468
block size	962 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.041 [-0.125 – 0.044]	**gene(s) hit or close-by**	IL1RAP
phastCons	μ = 0.247 [0 – 0.495]	**eQTL gene(s)**	–
GERP++	μ = 1.044 [-0.811 – 2.9]	**potentially regulated gene(s)**	–
CADD score	μ = 7.328 [2.746 – 11.91]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001382725 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K4me1
		skin (NHEK)	H3K4me1
ENSR00001487393 (enhancer)	1	liver (HepG2)	H3K4me1
		skin (NHEK)	H3K4me1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RAP	ENST00000317757	NM_001167931.1	ENSP00000314807	2
IL1RAP	ENST00000434491	?	ENSP00000391899	2
IL1RAP	ENST00000342550	?	ENSP00000345829	2
IL1RAP	ENST00000453359	?	ENSP00000412008	2
IL1RAP	ENST00000072516	NM_001167929.1	ENSP00000072516	2
IL1RAP	ENST00000422940	NM_001167930.1	ENSP00000387371	2
IL1RAP	ENST00000439062	NM_001167928.1	ENSP00000401132	2
IL1RAP	ENST00000422485	NM_134470.3	ENSP00000409352	2
IL1RAP	ENST00000447382	NM_002182.3	ENSP00000390541	2
IL1RAP	ENST00000422625	?	ENSP00000389149	2
IL1RAP	ENST00000413869	?	ENSP00000416296	2
IL1RAP	ENST00000412504	?	ENSP00000412053	2
IL1RAP	ENST00000443369	?	ENSP00000408893	2

Locus 232 (rs28385609)

SNiPAcard

Block annotations

Block info

genomic range	chr6:123,121,472-123,128,536
block size	7,065 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 1.207 [-1.366 – 6.514]	gene(s) hit or close-by	SMPDL3A
phastCons	μ = 0.250 [0 – 1]	eQTL gene(s)	SMPDL3A
GERP++	μ = 0.172 [-3.94 – 5.72]	potentially regulated gene(s)	SMPDL3A
CADD score	μ = 5.927 [0.363 – 14.62]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SMPDL3A	missense variant	ENST00000539041	NM_001286138.1	ENSP00000442152	S/P	Tct/Cct	?	?	1
SMPDL3A	missense variant	ENST00000368440	NM_006714.4	ENSP00000357425	S/P	Tct/Cct	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SMPDL3A	?	ENSG00000172594	tibial nerve	2.43×10^{-5} (p-value)	GTEx Portal V6	1
SMPDL3A	?	ENSG00000172594	transformed fibroblasts	1.28×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000715093	1	ENST00000368440, ENST00000539041	SMPDL3A

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001226031 (promoter flanking region)	2	NHLF	DNase1
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		muscle (HSMM)	DNase1, H3K36me3, H3K4me2
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	DNase1, H3K4me1, H3K4me2, FOXA1, H3K36me3
		lung (IMR90)	DNase1, H3K36me3
		blood (GM12878)	BATF
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K4me2, DNase1

ENSR00001226034 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SMPDL3A	downstream gene variant	2982	ENST00000487215	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SMPDL3A	ENST00000539041	NM_001286138.1	ENSP00000442152	3
SMPDL3A	ENST00000368440	NM_006714.4	ENSP00000357425	3

Locus 233 (rs16935432)

SNiPAcard

Block annotations

Block info

genomic range	chr11:18,435,241-18,474,128
block size	38,888 bp
variant count	94 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.590 [-3.912 – 2.718]	gene(s) hit or close-by	LDHAL6A, LDHC, RP11-613F22.7
phastCons	μ = 0.112 [0 – 0.908]	eQTL gene(s)	LDHAL6A, YWHABP2
GERP++	μ = -0.091 [-4.48 – 2.45]	potentially regulated gene(s)	AC084117.3, GTF2H1, LDHA, LDHC, RP11-113D6.3, RP11-137N23.1, RP11-613F22.6, SERGEF, SPTY2D1, TSG101, UEVLD
CADD score	μ = 4.204 [0.024 – 15.14]	disease gene(s)	LDHA

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Amyotrophic lateral sclerosis (sporadic)	<9.00×10-6	GWAS Catalog	24529757	1

Disease gene annotation

gene	trait	source DB	source entry/link
LDHA	GLYCOGEN STORAGE DISEASE XI	OMIM	MIM:612933
LDHA	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency	OrphaNet	OrphaNet:284426

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
YWHABP2	?	ENSG00000256464	skeletal muscle	5.30×10-8 (p-value)	GTEx Portal V6	94
LDHAL6A	?	ENSG00000166800	muscularis mucosae	5.05×10-6 (p-value)	GTEx Portal V6	71
LDHAL6A	?	ENSG00000166800	pituitary	2.69×10-7 (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000099956	1	ENCP00000011421	LDHC
		ENCP00000011426	TSG101 RP11-613F22.6
		ENCP00000011382	SERGEF
		ENCP00000011436	RP11-137N23.1
		ENCP00000011396	RP11-113D6.3
		ENCP00000011430	UEVLD
		ENCP00000011429	UEVLD

ENCE00000100103	1	ENCP00000011410	GTF2H1
		ENCP00000011435	SPTY2D1
		ENCP00000011419	LDHA
		ENCP00000011420	AC084117.3
		ENCP00000011418	LDHA

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LDHAL6A	upstream gene variant	3303	ENST00000280706	NM_144972.4	ENSP00000280706	3
LDHAL6A	upstream gene variant	3243	ENST00000396213	NM_001144071.1	ENSP00000379516	3
LDHAL6A	upstream gene variant	3243	ENST00000615355	?	ENSP00000478764	3
LDHC	downstream gene variant	756	ENST00000396215	?	ENSP00000379518	3
LDHC	downstream gene variant	872	ENST00000535809	?	ENSP00000443997	3
LDHC	downstream gene variant	872	ENST00000546146	?	ENSP00000443414	3
LDHC	downstream gene variant	756	ENST00000545848	?	ENSP00000438196	3
LDHC	downstream gene variant	872	ENST00000536880	?	ENSP00000439555	3
LDHC	downstream gene variant	756	ENST00000280704	NM_017448.3, NM_002301.4	ENSP00000280704	3
LDHC	downstream gene variant	872	ENST00000537486	?	ENSP00000441478	3
LDHC	downstream gene variant	872	ENST00000544105	?	ENSP00000439060	3
LDHC	downstream gene variant	756	ENST00000539369	?	ENSP00000441991	3
LDHC	downstream gene variant	523	ENST00000541669	?	ENSP00000437783	1
RP11-613F22.7	downstream gene variant, upstream gene variant	32	ENST00000535082	?	?	14

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LDHC	ENST00000396215	?	ENSP00000379518	91
LDHC	ENST00000535809	?	ENSP00000443997	91
LDHC	ENST00000280704	NM_017448.3, NM_002301.4	ENSP00000280704	91
LDHC	ENST00000541669	?	ENSP00000437783	91
LDHC	ENST00000545848	?	ENSP00000438196	91
LDHC	ENST00000536880	?	ENSP00000439555	91
LDHC	ENST00000546146	?	ENSP00000443414	91
LDHC	ENST00000537486	?	ENSP00000441478	91
LDHC	ENST00000544105	?	ENSP00000439060	91
LDHC	ENST00000539369	?	ENSP00000441991	91

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LDHC	ENST00000541669	?	ENSP00000437783	2

Locus 234 (rs260692)

SNiPAcard

Block annotations

Block info

genomic range	chr2:109,574,150-109,581,319
block size	7,170 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.658 [-2.622 – 1.528]	gene(s) hit or close-by	EDAR
phastCons	μ = 0.068 [0 – 0.9]	eQTL gene(s)	–
GERP++	μ = -0.616 [-5.82 – 4.76]	potentially regulated gene(s)	–
CADD score	μ = 3.340 [0.472 – 9.224]	disease gene(s)	EDAR

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
EDAR	HAIR MORPHOLOGY 1	OMIM	MIM:612630
EDAR	ECTODERMAL DYSPLASIA 10B, HYPOHIDROTIC/HAIR/TOOTH TYPE, [...]	OMIM	MIM:224900
EDAR	ECTODERMAL DYSPLASIA 10A, HYPOHIDROTIC/HAIR/NAIL TYPE, AUTOSOMAL [...]	OMIM	MIM:129490
EDAR	Autosomal dominant hypohidrotic ectodermal dysplasia	OrphaNet	OrphaNet:1810
EDAR	Autosomal recessive hypohidrotic ectodermal dysplasia	OrphaNet	OrphaNet:248

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000596156 (enhancer)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		skin (NHEK)	H3K4me1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
EDAR	ENST00000409271	?	ENSP00000386371	14
EDAR	ENST00000376651	?	ENSP00000365839	14
EDAR	ENST00000258443	NM_022336.3	ENSP00000258443	14

Locus 235 (rs8283)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,083,300-32,083,300
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.226	gene(s) hit or close-by	ATF6B, TNXB
phastCons	0	eQTL gene(s)	ATF6B, FKBPL, HLA-DRB6, MICB, STK19B, TAP2, TNXA, XXbac-BPG246D15.9
GERP++	3.07	potentially regulated gene(s)	–
CADD score	9.99	disease gene(s)	TAP2, TNXB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TAP2	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571
TAP2	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TAP2	ENST00000620123	ILMN_1759250	blood	5.92×10^{-5} (p-value)	MuTHER consortium	1
TAP2	ENST00000374899					
XXbac-BPG246D15.9	ENST00000452392					
TAP2	ENST00000464100					
TAP2	ENST00000374897					
MICB	ENST00000538442	ILMN_1708006	skin	2.74×10^{-5} (p-value)	MuTHER consortium	1
MICB	ENST00000399150					
MICB	ENST00000252229					
STK19B	?	ENSG00000250535	muscularis mucosae	2.36×10^{-6} (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	muscularis mucosae	2.63×10^{-6} (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	visceral adipocytes	5.27×10^{-7} (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	tibial nerve	2.04×10^{-7} (p-value)	GTEx Portal V6	1
STK19B	?	ENSG00000250535	esophagus mucosa	4.77×10^{-6} (p-value)	GTEx Portal V6	1
STK19B	?	ENSG00000250535	thyroid	9.02×10^{-10} (p-value)	GTEx Portal V6	1
HLA-DRB6	?	ENSG00000229391	skeletal muscle	1.15×10^{-7} (p-value)	GTEx Portal V6	1
STK19B	?	ENSG00000250535	cerebellum	3.12×10^{-6} (p-value)	GTEx Portal V6	1
ATF6B	?	ENSG00000213676	blood	8.78×10^{-3} (p-value)	SeeQTL DB (HapMap)	1

ATF6B	?	ENSG00000213676	blood	8.78×10-5 (q-value)	SeeQTL DB (HapMap)	1
STK19B	?	ENSG00000250535	sun exposed skin	4.28×10-7 (p-value)	GTEx Portal V6	1
FKBPL	?	ENSG00000204315	sun exposed skin	4.94×10-8 (p-value)	GTEx Portal V6	1
HLA-DRB6	?	ENSG00000229391	sun exposed skin	1.81×10-8 (p-value)	GTEx Portal V6	1
HLA-DRB6	?	ENSG00000229391	subcutaneous adipocytes	3.42×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ATF6B	1	ENST00000375201 ENST00000375203 ENST00000494022	hsa-miR-371a-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ATF6B	downstream gene variant	1185	ENST00000453203	?	ENSP00000393419	1
ATF6B	downstream gene variant	3254	ENST00000475705	?	?	1
ATF6B	downstream gene variant	4986	ENST00000485314	?	?	1
ATF6B	downstream gene variant	4919	ENST00000495579	?	?	1
ATF6B	downstream gene variant	3308	ENST00000492342	?	?	1
TNXB	upstream gene variant	189	ENST00000442721	?	ENSP00000389946	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATF6B	ENST00000494022	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATF6B	ENST00000375201	NM_001136153.1	ENSP00000364347	1
ATF6B	ENST00000375203	NM_004381.4	ENSP00000364349	1

Locus 237 (rs1005776)

SNiPAcard

Block annotations

Block info

genomic range	chr14:102,807,088-102,904,179
block size	97,092 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.144 [-4.741 – 1.045]	gene(s) hit or close-by	AL137229.1, CINP, TECPR2, ZNF839
phastCons	μ = 0.025 [0 – 0.295]	eQTL gene(s)	ANKRD9, CINP, TECPR2, ZNF839
GERP++	μ = -0.697 [-5.15 – 2.34]	potentially regulated gene(s)	–
CADD score	μ = 2.528 [0.153 – 6.923]	disease gene(s)	TECPR2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TECPR2	SPASTIC PARAPLEGIA 49, AUTOSOMAL RECESSIVE	OMIM	MIM:615031
TECPR2	Hereditary Spastic Paraparesis	DECIPHER	MIM:615031
TECPR2	Autosomal recessive spastic paraplegia type 49	OrphaNet	OrphaNet:320385

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CINP	?	ENSG00000100865	pancreas	2.73×10^{-6} (p-value)	GTEx Portal V6	4
CINP	?	ENSG00000100865	muscularis mucosae	1.08×10^{-13} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	lung	7.13×10^{-13} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	atrial appendage	1.82×10^{-9} (p-value)	GTEx Portal V6	16
ANKRD9	?	ENSG00000156381	transformed fibroblasts	3.75×10^{-25} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	transformed fibroblasts	4.66×10^{-15} (p-value)	GTEx Portal V6	20
TECPR2	?	ENSG00000196663	tibial artery	6.53×10^{-7} (p-value)	GTEx Portal V6	15
CINP	?	ENSG00000100865	tibial artery	1.15×10^{-10} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	breast	1.45×10^{-10} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	blood	8.51×10^{-10} (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	thyroid	9.94×10^{-12} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	skeletal muscle	1.51×10^{-15} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	transverse colon	1.62×10^{-9} (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	EBV lymphocytes	7.87×10^{-9} (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	unexposed skin	2.69×10^{-11} (p-value)	GTEx Portal V6	20

CINP	?	ENSG00000100865	sun exposed skin	8.24×10-17 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	liver	4.17×10-9 (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	left ventricle	1.20×10-10 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	aorta	1.49×10-11 (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	subcutaneous adipocytes	2.11×10-16 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	visceral adipocytes	5.94×10-14 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	tibial nerve	2.58×10-11 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	coronary artery	8.52×10-8 (p-value)	GTEx Portal V6	8
CINP	?	ENSG00000100865	sigmoid colon	5.46×10-9 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	esophagus mucosa	1.18×10-15 (p-value)	GTEx Portal V6	20
CINP	ENST00000216756	ILMN_1765257	blood	6.74×10-6 (p-value)	Westra et al.	8
CINP	ENST00000541568					
CINP	ENST00000559504					
CINP	ENST00000559514					
CINP	ENST00000536961					
ANKRD9	ENST00000559404	ILMN_1675937	blood	5.57×10-7 (p-value)	Westra et al.	5
ANKRD9	ENST00000286918					
ANKRD9	ENST00000559651					
ANKRD9	ENST00000557902					
ANKRD9	ENST00000560748					
CINP	?	ENSG00000100865	gastroesophageal junction	3.54×10-9 (p-value)	GTEx Portal V6	18
CINP	?	ENSG00000100865	testis	1.38×10-8 (p-value)	GTEx Portal V6	19
ZNF839	ENST00000558490	ILMN_2186858	skin	3.87×10-9 (p-value)	MuTHER consortium	8
ZNF839	ENST00000420933		adipocyte	2.40×10-7 (p-value)	MuTHER consortium	8
ZNF839	ENST00000557803		blood	3.39×10-5 (p-value)	MuTHER consortium	8
ZNF839	ENST00000559098					
ZNF839	ENST00000561251					
ZNF839	ENST00000558850					
ZNF839	ENST00000559155					
ZNF839	ENST00000559185					
ZNF839	ENST00000442396					
ZNF839	ENST00000560112					
ZNF839	ENST00000558462					
ZNF839	ENST00000420933	ILMN_2186858	b-cell	1.12×10-6 (p-value)	Fairfax et al.	2
ZNF839	ENST00000558490					
ZNF839	ENST00000557803					
ZNF839	ENST00000559098					
ZNF839	ENST00000559155					
ZNF839	ENST00000558850					
ZNF839	ENST00000561251					
ZNF839	ENST00000559185					
ZNF839	ENST00000442396					
ZNF839	ENST00000560112					
ZNF839	ENST00000558462					
CINP	?	ENSG00000100865	adrenal gland	4.59×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000424047 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27ac, H3K36me3, DNase1
ENSR00001625363 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K36me3
		blood (K562)	H3K27me3
ENSR00000424048 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, Yy1, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, CTCF, H2AZ, Rad21, Egr1
		skin (NHDF-AD)	CTCF, H3K4me2, DNase1, H3K4me3
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, CTCF, Rad21
		blood (GM12878)	CTCF, DNase1, H2AZ, Yy1, Rad21
		lung (IMR90)	H3K4me2, H3K36me3, CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	H3K36me3, CTCF, H3K4me2
		blood (DND-41)	CTCF, H3K36me3
		breast (HMEC)	CTCF, H3K4me2
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		A549	H3K4me3, CTCF, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL137229.1	upstream gene variant, downstream gene variant	218	ENST00000631034	?	?	3
CINP	downstream gene variant	351	ENST00000559514	?	ENSP00000453839	1
CINP	downstream gene variant	1868	ENST00000559504	?	ENSP00000453846	1
CINP	downstream gene variant, upstream gene variant	2746	ENST00000560326	?	?	3
CINP	downstream gene variant	1712	ENST00000558523	?	?	2
CINP	downstream gene variant	955	ENST00000536961	?	ENSP00000442057	2
CINP	downstream gene variant	2389	ENST00000561468	?	?	1
CINP	downstream gene variant	752	ENST00000216756	NM_032630.2	ENSP00000216756	2
CINP	downstream gene variant	2476	ENST00000558764	?	ENSP00000452982	1
CINP	downstream gene variant	981	ENST00000541568	?	ENSP00000442377	2
TECPR2	downstream gene variant	1663	ENST00000561228	?	?	1
TECPR2	upstream gene variant	4167	ENST00000560060	?	?	2
TECPR2	upstream gene variant	435	ENST00000557786	?	?	1
ZNF839	downstream gene variant	858	ENST00000558850	NM_001267827.1	ENSP00000453363	2
ZNF839	downstream gene variant	520	ENST00000561251	?	ENSP00000453137	3
ZNF839	downstream gene variant	855	ENST00000557803	?	?	2
ZNF839	downstream gene variant	855	ENST00000559155	?	ENSP00000453415	2
ZNF839	downstream gene variant	1246	ENST00000560112	?	ENSP00000453744	2
ZNF839	downstream gene variant	855	ENST00000559098	?	ENSP00000453515	2
ZNF839	downstream gene variant	858	ENST00000558462	?	ENSP00000454159	2

ZNF839	downstream gene variant	855	ENST00000420933	?	?	2
ZNF839	downstream gene variant	1921	ENST00000560568	?	ENSP00000452983	2
ZNF839	downstream gene variant	855	ENST00000558490	?	ENSP00000452819	2
ZNF839	downstream gene variant	855	ENST00000442396	NM_018335.4	ENSP00000399863	2
ZNF839	downstream gene variant	855	ENST00000559185	NM_001267828.1	ENSP00000453109	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CINP	ENST00000559504	?	ENSP00000453846	4
CINP	ENST00000559514	?	ENSP00000453839	4
CINP	ENST00000536961	?	ENSP00000442057	2
CINP	ENST00000216756	NM_032630.2	ENSP00000216756	2
CINP	ENST00000561468	?	?	2
CINP	ENST00000541568	?	ENSP00000442377	2
CINP	ENST00000558764	?	ENSP00000452982	2
CINP	ENST00000560326	?	?	1
TECPR2	ENST00000558678	NM_001172631.1	ENSP00000453671	15
TECPR2	ENST00000359520	NM_014844.3	ENSP00000352510	15
TECPR2	ENST00000560060	?	?	1
TECPR2	ENST00000561228	?	?	11
ZNF839	ENST00000558850	NM_001267827.1	ENSP00000453363	1
ZNF839	ENST00000559098	?	ENSP00000453515	1
ZNF839	ENST00000560112	?	ENSP00000453744	1
ZNF839	ENST00000442396	NM_018335.4	ENSP00000399863	1
ZNF839	ENST00000561251	?	ENSP00000453137	1
ZNF839	ENST00000559185	NM_001267828.1	ENSP00000453109	1
ZNF839	ENST00000557803	?	?	1
ZNF839	ENST00000559155	?	ENSP00000453415	1
ZNF839	ENST00000558462	?	ENSP00000454159	1
ZNF839	ENST00000558490	?	ENSP00000452819	1
ZNF839	ENST00000420933	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CINP	ENST00000559504	?	ENSP00000453846	2
CINP	ENST00000559514	?	ENSP00000453839	2

Locus 238 (rs12520053)

SNiPAcard

Block annotations

Block info

genomic range	chr5:96,163,905-96,171,829
block size	7,925 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.824 [-3.443 – 0.636]	**gene(s) hit or close-by**	CTD-2260A17.2
phastCons	μ = 0.005 [0 – 0.021]	**eQTL gene(s)**	CAST, ERAP2, LNPEP, RP11-254I22.3
GERP++	μ = -0.669 [-4.04 – 0.895]	**potentially regulated gene(s)**	LIX1
CADD score	μ = 3.809 [1.094 – 7.845]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP2	ENST00000513084	ILMN_1743145	blood	6.15×10^{-5} (p-value)	MuTHER consortium	7
ERAP2	ENST00000512869					
ERAP2	ENST00000379904					
ERAP2	ENST00000437043					
LNPEP	ENST00000395770	ILMN_1814737	blood	4.47×10^{-5} (p-value)	Westra et al.	7
LNPEP	ENST00000231368					
CAST	ENST00000511782	ILMN_1783627	blood	4.26×10^{-6} (p-value)	Westra et al.	7
CAST	ENST00000511049					
CAST	ENST00000395813					
CAST	ENST00000421689					
CAST	ENST00000506811					
CAST	ENST00000510156					
CAST	ENST00000507487					
CAST	ENST00000341926					
CAST	ENST00000508117					
CAST	ENST00000508830					
CAST	ENST00000338252					
CAST	ENST00000505143					
CAST	ENST00000514055					
RP11-254I22.3	?	ENSG00000249746	testis	1.12×10^{-5} (p-value)	GTEx Portal V6	7
ERAP2	?	ENSG00000164308	thyroid	2.63×10^{-5} (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000417175	1	ENCP00000044982	LIX1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2260A17.2	ENST00000504056	?	?	10
CTD-2260A17.2	ENST00000502262	?	?	10
CTD-2260A17.2	ENST00000501338	?	?	10

Locus 239 (rs2412776)

SNiPAcard

Block annotations

Block info

genomic range	chr4:57,941,160-57,952,521
block size	11,362 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.191 [-1.557 – 1.293]	gene(s) hit or close-by	IGFBP7, UBE2CP3
phastCons	μ = 0.163 [0 – 0.97]	eQTL gene(s)	–
GERP++	μ = -0.177 [-9.01 – 5.22]	potentially regulated gene(s)	POLR2B
CADD score	μ = 6.193 [0.746 – 18.99]	disease gene(s)	IGFBP7

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
IGFBP7	RETINAL ARTERIAL MACROANEURYSM WITH SUPRAVALVULAR PULMONIC STENOSIS;	DECIPHER	MIM:614224
IGFBP7	RETINAL ARTERIAL MACROANEURYSM WITH SUPRAVALVULAR PULMONIC [...]	OMIM	MIM:614224
IGFBP7	Familial retinal arterial macroaneurysm	OrphaNet	OrphaNet:284247

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000380606	2	ENCP00000041130	POLR2B

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001241735 (promoter flanking region)	5	NHLF	DNase1
		HSMMtube	H3K36me3, DNase1
		Osteobl	H3K27ac, H3K36me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	Max, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1, H3K36me3
		nervous (NH-A)	H3K36me3, DNase1
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K36me3
ENSR00001685382 (enhancer)	2	Osteobl	H3K36me3
		nervous (NH-A)	H3K36me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
UBE2CP3	downstream gene variant	1862	ENST00000502715	?	?	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IGFBP7	ENST00000514062	NM_001253835.1	ENSP00000486293	12
IGFBP7	ENST00000295666	NM_001553.2	ENSP00000295666	12

Locus 240 (rs6668534)

SNiPAcard

Block annotations

Block info

genomic range	chr1:161,644,976-161,671,995
block size	27,020 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.717 [-1.689 – 0.386]	gene(s) hit or close-by	FCGR2B, FCRLA, RP11-25K21.1, RPL31P11, Y_RNA
phastCons	μ = 0.035 [0 – 0.075]	eQTL gene(s)	FCGR2B, FCGR2C, FCGR3B, FCRLA, FCRLB, HSPA6, HSPA7
GERP++	μ = -0.263 [-2.5 – 0.705]	potentially regulated gene(s)	–
CADD score	μ = 2.677 [0.282 – 5.888]	disease gene(s)	FCGR2B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
FCGR2B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
FCGR2B	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCGR3B	?	ENSG00000162747	sun exposed skin	3.64×10^{-9} (p-value)	GTEx Portal V6	7
FCGR3B	?	ENSG00000162747	unexposed skin	5.28×10^{-9} (p-value)	GTEx Portal V6	7
FCGR2C	?	ENSG00000244682	tibial nerve	1.38×10^{-6} (p-value)	GTEx Portal V6	1
FCRLA	ENST00000367957	ILMN_1691071	skin	2.19×10^{-8} (p-value)	MuTHER consortium	2
FCRLA	ENST00000236938		blood	1.93×10^{-14} (p-value)	MuTHER consortium	2
FCRLA	ENST00000350710					
FCRLA	ENST00000470841					
FCRLA	ENST00000367949					
FCRLA	ENST00000367959					
FCRLA	ENST00000546024					
FCRLA	ENST00000540521					
FCRLA	ENST00000367953					
?	?	ILMN_1728639	blood	8.15×10^{-5} (p-value)	Westra et al.	2
FCGR2B	ENST00000403078	ILMN_1660027	blood	5.57×10^{-7} (p-value)	Westra et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000403078	ILMN_2382403	monocyte	1.44×10^{-7} (p-value)	Fairfax et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					

FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000480308	ILMN_1660027	monocyte	6.50×10-10 (p-value)	Fairfax et al.	1
FCGR2B	ENST00000403078					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCRLB	ENST00000367948	ILMN_1782015	monocyte	8.39×10-11 (p-value)	Fairfax et al.	1
FCRLB	ENST00000495397					
FCRLB	ENST00000367944					
FCRLB	ENST00000336830					
HSPA7	ENST00000445535	ILMN_3235964	monocyte	8.70×10-8 (p-value)	Fairfax et al.	1
			b-cell	2.28×10-6 (p-value)	Fairfax et al.	1
FCGR2B	ENST00000480308	ILMN_2382403	monocyte	4.50×10-18 (p-value)	Zeller et al.	1
FCGR2B	ENST00000403078					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000403078	ILMN_1660027	monocyte	1.52×10-39 (p-value)	Zeller et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCRLB	ENST00000367948	ILMN_1782015	monocyte	2.05×10-24 (p-value)	Zeller et al.	1
FCRLB	ENST00000367944					
FCRLB	ENST00000495397					
FCRLB	ENST00000336830					
HSPA7	ENST00000445535	ILMN_1806165	monocyte	1.02×10-14 (p-value)	Zeller et al.	1
HSPA6	ENST00000309758					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000546488 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		A549	H3K4me3, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR2B	downstream gene variant	1070	ENST00000485778	?	?	1
FCRLA	upstream gene variant	4767	ENST00000540521	NM_001184870.1	ENSP00000442870	1
FCRLA	upstream gene variant	4767	ENST00000367959	NM_001184866.1	ENSP00000356936	1
FCRLA	upstream gene variant	4767	ENST00000546024	NM_001184867.1	ENSP00000439838	1
FCRLA	upstream gene variant	4767	ENST00000236938	NM_032738.3	ENSP00000236938	1
FCRLA	upstream gene variant	4767	ENST00000350710	NM_001184871.1	ENSP00000344808	1
FCRLA	upstream gene variant	4767	ENST00000367949	NM_001184873.1	ENSP00000356926	1

RP11-25K21.1	upstream gene variant	362	ENST00000453111	?	?	1
RP11-25K21.1	upstream gene variant	422	ENST00000626340	?	?	1
RPL31P11	upstream gene variant	4803	ENST00000426558	?	?	1
RPL31P11	upstream gene variant	4846	ENST00000454082	?	?	1
Y_RNA	upstream gene variant, downstream gene variant	2598	ENST00000362725	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2B	ENST00000403078	?	ENSP00000386038	1
FCGR2B	ENST00000358671	NM_004001.4, NM_001002275.2	ENSP00000351497	1
FCGR2B	ENST00000367960	?	ENSP00000356937	1
FCGR2B	ENST00000428605	?	ENSP00000404329	1
FCGR2B	ENST00000236937	NM_001002274.2, NM_001002273.2	ENSP00000236937	1
FCGR2B	ENST00000367961	NM_001190828.1	ENSP00000356938	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FCGR2B	ENST00000480308	?	1

Locus 241 (rs10858300)

SNiPAcard

Block annotations

Block info

genomic range	chr9:137,840,809-137,857,978
block size	17,170 bp
variant count	23 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.830 [-4.588 – 0.787]	**gene(s) hit or close-by**	–
phastCons	μ = 0.003 [0 – 0.046]	**eQTL gene(s)**	FCN1, NELFB, OLFM1, RP11-447M12.2
GERP++	μ = -0.548 [-4.14 – 1.89]	**potentially regulated gene(s)**	–
CADD score	μ = 2.698 [0.166 – 6.591]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-447M12.2	?	ENSG00000236403	blood	3.61×10^{-10} (p-value)	GTEx Portal V6	23
FCN1	?	ENSG00000085265	blood	5.18×10^{-18} (p-value)	GTEx Portal V6	23
OLFM1	ENST00000371796	ILMN_1742025	monocyte	2.73×10^{-23} (p-value)	Fairfax et al.	4
OLFM1	ENST00000371793					
OLFM1	ENST00000483042					
OLFM1	ENST00000252854					
OLFM1	ENST00000392991	ILMN_1714709	monocyte	1.15×10^{-19} (p-value)	Fairfax et al.	2
OLFM1	ENST00000277415					
NELFB	ENST00000343053	ILMN_2290007	monocyte	3.73×10^{-4} (p-value)	Fairfax et al.	2
OLFM1	ENST00000392991	ILMN_1714709	monocyte	1.43×10^{-65} (p-value)	Zeller et al.	3
OLFM1	ENST00000277415					
OLFM1	ENST00000371796	ILMN_1742025	monocyte	8.82×10^{-76} (p-value)	Zeller et al.	3
OLFM1	ENST00000371793					
OLFM1	ENST00000252854					
OLFM1	ENST00000483042					

Locus 242 (rs9468569)

SNiPAcard

Block annotations

Block info

genomic range	chr6:29,613,377-29,671,356
block size	57,980 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.431 [-3.465 – 1.743]	gene(s) hit or close-by	MOG, ZDHHC20P1, ZFP57
phastCons	μ = 0.044 [0 – 0.4]	eQTL gene(s)	–
GERP++	μ = 0.146 [-1.75 – 3.54]	potentially regulated gene(s)	GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-F, HLA-F, HLA-F, HLA-F, HLA-F, HLA-F, HLA-F, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, MOG, MOG, MOG, MOG, MOG, MOG, MOG, MOG, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, ZNRD1-AS1
CADD score	μ = 2.954 [0.116 – 10.4]	disease gene(s)	HLA-A, MOG, ZFP57, , , , , , , ,

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
HLA-A	Birdshot chorioretinopathy	OrphaNet	OrphaNet:179	orphanet
MOG	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886	orphanet
MOG	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410	OMIM

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000735239	1	ENST00000376898, ENST00000494692, ENST00000376917, ENST00000490427, ENST00000376888, ENST00000396704, ENST00000416766, ENST00000431798, ENST00000483013, ENST00000376894, ENST00000396701	MOG

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000047827		MOG
		MOG

			MOG
			MOG
			MOG
			MOG
			MOG

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444016	1	ENCP00000047860	HLA-A HLA-A HLA-A HLA-A HLA-A HLA-A HLA-A HLA-A
		ENCP00000047832	HLA-F HLA-F HLA-F HLA-F HLA-F HLA-F HLA-F
		ENCP00000047875	PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11
		ENCP00000047871	ZNRD1-AS1
		ENCP00000047861	HCG4P5 HCG4P5 HCG4P5 HCG4P5 HCG4P5 HCG4P5 HCG4P5 HCG4P5
		ENCP00000047851	HLA-H HLA-H HLA-H HLA-H HLA-H HLA-H HLA-H HLA-H
		ENCP00000047876	PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11
		ENCP00000047824	GABBR1 GABBR1 GABBR1 GABBR1 GABBR1 GABBR1 GABBR1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487538	2	embryonic stem cell (H1ESC)	PolII, Rad21, CTCF, Yy1, H3K4me2, H3K4me3, DNase1

(CTCF binding site)		HSMMtube	H2AZ, H3K4me2, CTCF, DNase1
		blood (K562)	PolII, H3K4me2, CTCF, DNase1, H3K4me3, H2AZ, THAP1, H3K27me3, Yy1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H3K4me3, ELF1, H2AZ, H3K4me2, CTCF, DNase1
		blood (GM12878)	H3K4me3, H3K27ac, H3K4me2, CTCF, PolII, H2AZ, DNase1, Yy1, ELF1
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, CTCF
		nervous (NH-A)	H3K9ac, CTCF, DNase1, H3K4me2
		skin (NHEK)	H3K4me2, CTCF, DNase1
		NHLF	DNase1, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	CTCF, H3K27ac, H3K4me3
		breast (HMEC)	CTCF, H3K4me2, H3K4me3, DNase1
		cervix (HeLa-S3)	H3K4me2, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, H3K4me2, PolII
		A549	DNase1, CTCF, H3K4me2, H3K4me3
ENSR00001494451 e! (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me1, H3K27me3, PolII, Rad21, CTCF, Yy1, H3K4me2, H3K4me3
		HSMMtube	DNase1, H2AZ, H3K4me2, CTCF
		blood (K562)	Yy1, THAP1, H2AZ, PolII, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H3K4me3, ELF1, H2AZ, H3K4me2, CTCF, DNase1
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, CTCF
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, ELF1, H3K4me3, H3K27ac, H3K4me2, CTCF
		nervous (NH-A)	DNase1, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	CTCF, DNase1, H3K4me2
		NHLF	DNase1, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	CTCF, H3K27ac, H3K4me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K4me2, PolII, CTCF
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K4me3, H3K4me2, DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, H3K4me2, PolII, CTCF, DNase1
		A549	DNase1, CTCF, H3K4me2, H3K4me3
ENSR00000487541 e! (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	H4K5ac, H3K27ac, DNase1
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
ENSR00000487543 e! (TF binding site)	5	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K27ac, CTCF, PolII
		blood (K562)	H3K27me3
		A549	H3K27me3
ENSR00000487548 e! (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3, Rad21, CTCF
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MOG e!	upstream gene variant, downstream gene variant	1672	ENST00000416766 e!	?	ENSP00000409394 e!	4
MOG e!	upstream gene variant, downstream gene variant	1672	ENST00000376898 e!	NM_002433.4	ENSP00000366095 e!	4
MOG e!	upstream gene variant, downstream gene variant	1672	ENST00000490427 e!	NM_001170418.1	ENSP00000420350 e!	4
MOG e!	upstream gene variant, downstream gene variant	1266	ENST00000485885 e!	?	?	12
MOG e!	upstream gene variant, downstream gene variant	1672	ENST00000396704 e!	NM_001008228.2	ENSP00000379932 e!	4

MOG	upstream gene variant, downstream gene variant	1443	ENST00000376917	NM_206809.3	ENSP00000366115	4
MOG	upstream gene variant, downstream gene variant	259	ENST00000376889	?	ENSP00000366086	6
MOG	upstream gene variant, downstream gene variant	1554	ENST00000376894	?	ENSP00000366091	4
MOG	upstream gene variant, downstream gene variant	259	ENST00000485211	?	ENSP00000418872	6
MOG	upstream gene variant, downstream gene variant	1338	ENST00000476244	?	?	10
MOG	upstream gene variant, downstream gene variant	1672	ENST00000483013	?	ENSP00000418090	4
MOG	upstream gene variant, downstream gene variant	1592	ENST00000469353	?	?	2
MOG	upstream gene variant, downstream gene variant	921	ENST00000376903	?	ENSP00000366101	9
MOG	upstream gene variant, downstream gene variant	1465	ENST00000376888	NM_206814.5	ENSP00000366085	4
MOG	upstream gene variant, downstream gene variant	1672	ENST00000376891	NM_001008229.2	ENSP00000366088	4
MOG	upstream gene variant, downstream gene variant	1672	ENST00000431798	NM_206812.3	ENSP00000410866	4
MOG	upstream gene variant, downstream gene variant	598	ENST00000469603	?	?	10
MOG	upstream gene variant, downstream gene variant	1672	ENST00000396701	NM_206810.3	ENSP00000379929	4
MOG	upstream gene variant, downstream gene variant	1672	ENST00000494692	NM_206811.3	ENSP00000417405	4
ZDHHC20P1	downstream gene variant	4546	ENST00000435787	?	?	2
ZFP57	downstream gene variant, upstream gene variant	3541	ENST00000488757	NM_001109809.2	ENSP00000418259	8
ZFP57	downstream gene variant, upstream gene variant	3649	ENST00000376881	?	ENSP00000366078	4
ZFP57	downstream gene variant, upstream gene variant	288	ENST00000376883	?	ENSP00000366080	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MOG	ENST00000396701	NM_206810.3	ENSP00000379929	12
MOG	ENST00000490427	NM_001170418.1	ENSP00000420350	12
MOG	ENST00000376888	NM_206814.5	ENSP00000366085	12
MOG	ENST00000396704	NM_001008228.2	ENSP00000379932	12
MOG	ENST00000494692	NM_206811.3	ENSP00000417405	12
MOG	ENST00000483013	?	ENSP00000418090	12
MOG	ENST00000431798	NM_206812.3	ENSP00000410866	12
MOG	ENST00000376903	?	ENSP00000366101	4
MOG	ENST00000485211	?	ENSP00000418872	10
MOG	ENST00000376891	NM_001008229.2	ENSP00000366088	12
MOG	ENST00000469353	?	?	2
MOG	ENST00000469603	?	?	3
MOG	ENST00000416766	?	ENSP00000409394	12
MOG	ENST00000376898	NM_002433.4	ENSP00000366095	12
MOG	ENST00000376894	?	ENSP00000366091	12
MOG	ENST00000476244	?	?	3
MOG	ENST00000376889	?	ENSP00000366086	10
MOG	ENST00000376917	NM_206809.3	ENSP00000366115	12
ZFP57	ENST00000376883	?	ENSP00000366080	3

ZFP57	ENST00000488757	NM_001109809.2	ENSP00000418259	3
ZFP57	ENST00000376881	?	ENSP00000366078	3

Locus 243 (rs4938330)

SNiPAcard

Block annotations

Block info

genomic range	chr11:116,779,499-116,982,821
block size	203,323 bp
variant count	63 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.780 [-4.61 – 2.46]	gene(s) hit or close-by	AP000936.1, AP000936.2, AP000936.4, RNY4P6, SIK3
phastCons	μ = 0.041 [0 – 1]	eQTL gene(s)	APOA1, APOA1-AS, BUD13, PCSK7, RP11-109L13.1, SIDT2, SIK3, TAGLN
GERP++	μ = -0.556 [-5.77 – 3.97]	potentially regulated gene(s)	AP000936.1, BACE1
CADD score	μ = 3.543 [0.006 – 19.14]	disease gene(s)	APOA1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Cholesterol, hdl	9.34×10^{-5}	dbGaP	pha003074	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
APOA1	AMYLOIDOSIS, FAMILIAL VISCERAL	OMIM	MIM:105200	OMIM
APOA1	HYPOALPHALIPOPROTEINEMIA, PRIMARY	OMIM	MIM:604091	OMIM
APOA1	TANGIER DISEASE	OMIM	MIM:205400	OMIM
APOA1	APOLIPOPROTEIN A-I	OMIM	MIM:107680	OMIM
APOA1	Primary systemic amyloidosis	OrphaNet	OrphaNet:314701	orphanet
APOA1	Familial renal amyloidosis due to Apolipoprotein AI variant	OrphaNet	OrphaNet:93560	orphanet
APOA1	APOLIPOPROTEIN A-I DEFICIENCY	OrphaNet	OrphaNet:425	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TAGLN	ENST00000525531	ILMN_1778668	blood	3.34×10^{-8} (p-value)	MuTHER consortium	26
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
RP11-109L13.1	?	ENSG00000254851	visceral adipocytes	7.68×10^{-11} (p-value)	GTEx Portal V6	61
RP11-109L13.1	?	ENSG00000254851	testis	1.98×10^{-7} (p-value)	GTEx Portal V6	59
APOA1	?	ENSG00000118137	left ventricle	5.60×10^{-12} (p-value)	GTEx Portal V6	62
APOA1-AS	?	ENSG00000235910	left ventricle	8.11×10^{-11} (p-value)	GTEx Portal V6	62
RP11-109L13.1	?	ENSG00000254851	thyroid	6.71×10^{-9} (p-value)	GTEx Portal V6	59

SIK3	?	ENSG00000160584	esophagus mucosa	4.60×10-9 (p-value)	GTEx Portal V6	59
PCSK7	?	ENSG00000160613	esophagus mucosa	6.36×10-8 (p-value)	GTEx Portal V6	28
RP11-109L13.1	?	ENSG00000254851	esophagus mucosa	4.73×10-6 (p-value)	GTEx Portal V6	14
RP11-109L13.1	?	ENSG00000254851	muscularis mucosae	7.17×10-7 (p-value)	GTEx Portal V6	17
RP11-109L13.1	?	ENSG00000254851	stomach	8.58×10-8 (p-value)	GTEx Portal V6	42
RP11-109L13.1	?	ENSG00000254851	tibial nerve	3.96×10-8 (p-value)	GTEx Portal V6	21
PCSK7	?	ENSG00000160613	transformed fibroblasts	9.62×10-7 (p-value)	GTEx Portal V6	39
TAGLN	?	ENSG00000149591	blood	6.52×10-12 (p-value)	GTEx Portal V6	60
BUD13	?	ENSG00000137656	blood	5.63×10-6 (p-value)	GTEx Portal V6	4
RP11-109L13.1	?	ENSG00000254851	tibial artery	9.61×10-8 (p-value)	GTEx Portal V6	34
TAGLN	?	ENSG00000149591	EBV lymphocytes	1.81×10-7 (p-value)	GTEx Portal V6	8
RP11-109L13.1	?	ENSG00000254851	sun exposed skin	3.14×10-8 (p-value)	GTEx Portal V6	21
RP11-109L13.1	?	ENSG00000254851	aorta	2.03×10-7 (p-value)	GTEx Portal V6	20
RP11-109L13.1	?	ENSG00000254851	subcutaneous adipocytes	3.42×10-9 (p-value)	GTEx Portal V6	23
RP11-109L13.1	?	ENSG00000254851	atrial appendage	6.58×10-6 (p-value)	GTEx Portal V6	1
RP11-109L13.1	?	ENSG00000254851	unexposed skin	3.78×10-6 (p-value)	GTEx Portal V6	14
SIDT2	ENST00000278951	ILMN_1791912	blood	9.93×10-7 (p-value)	Westra et al.	25
SIDT2	ENST00000532062					
SIDT2	ENST00000324225					
SIDT2	ENST00000431081					
SIDT2	ENST00000620360					
RP11-109L13.1	?	ENSG00000254851	lung	7.78×10-6 (p-value)	GTEx Portal V6	12
RP11-109L13.1	?	ENSG00000254851	skeletal muscle	9.17×10-6 (p-value)	GTEx Portal V6	7
TAGLN	ENST00000525531	ILMN_1778668	monocyte	9.43×10-14 (p-value)	Zeller et al.	2
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000014771		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000123692	1	ENCP00000014802	BACE1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001573920 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me3, H3K27ac, DNase1, H3K4me1
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K4me2
		Osteobl	H3K27ac
		muscle (HSMM)	DNase1
ENSR00001573922 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac
		liver (HepG2)	H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00001607331 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H4K20me1
		blood (GM12878)	H3K4me2, H3K4me1
		A549	H3K4me2, H3K4me3
ENSR00000569539 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		embryonic stem cell (H1ESC)	DNase1
		blood (K562)	DNase1
		blood (DND-41)	H3K4me1, H3K27ac
ENSR00001607333 (enhancer)	1	monocytes (Monocytes-CD14+)	H4K20me1
		Osteobl	H3K36me3
ENSR00000569542 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H4K20me1
		endothelium (HUVEC)	Cjun, DNase1
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00000569548 (promoter flanking region)	8	embryonic stem cell (H1ESC)	DNase1, CTCF
		HSMMtube	DNase1
		blood (K562)	PU1, Cjun, FOSL1, DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		liver (HepG2)	FOSL2, Jund, DNase1, H3K4me1, FOXA1
		lung (IMR90)	DNase1
		blood (GM12878)	H3K79me2, BATF
		nervous (NH-A)	DNase1, H3K27me3
		skin (NHEK)	H3K27ac, H3K36me3, H3K4me1, DNase1
		NHLF	DNase1
		Osteobl	H3K27ac, H4K20me1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1, Cfos, Jund, Cjun
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H4K20me1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	DNase1, H3K4me2, H3K4me3
ENSR00001607339 (CTCF binding site)	2	monocytes (Monocytes-CD14+)	H3K4me1, H4K20me1
		liver (HepG2)	H3K4me1
		A549	H3K4me2, H3K4me3
		skin (NHEK)	H3K4me1, H3K27ac
ENSR00001607340 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H4K20me1
		embryonic stem cell (H1ESC)	CTCF
		blood (GM12878)	H3K79me2
		nervous (NH-A)	H3K27me3
		skin (NHEK)	DNase1
ENSR00001573930 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H4K20me1
		embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H4K20me1, FOXA2, HDAC2, DNase1, H3K4me1, FOXA1
		blood (GM12878)	H3K4me2, H3K79me2, H3K4me1
		skin (NHEK)	DNase1
		muscle (HSMM)	H3K36me3
ENSR00001573932	1	monocytes (Monocytes-CD14+)	H4K20me1

(enhancer)			
ENSR00001573933 (enhancer)	1	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H3K79me2, H3K4me1, FOXA1
		blood (GM12878)	H3K36me3, H3K4me2, H3K4me3, H3K79me2
		A549	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP000936.1	downstream gene variant, upstream gene variant	276	ENST00000438127	?	?	5
AP000936.2	downstream gene variant, upstream gene variant	50	ENST00000584092	?	?	2
AP000936.4	downstream gene variant	1025	ENST00000442124	?	?	2
RNY4P6	upstream gene variant, downstream gene variant	517	ENST00000363667	?	?	2
SIK3	upstream gene variant	361	ENST00000413553	?	ENSP00000414347	2
SIK3	upstream gene variant	341	ENST00000497049	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIK3	ENST00000497049	?	?	4
SIK3	ENST00000446921	NM_001281749.1	ENSP00000390442	61
SIK3	ENST00000485363	?	?	61
SIK3	ENST00000413553	?	ENSP00000414347	4
SIK3	ENST00000445177	?	ENSP00000391295	61
SIK3	ENST00000415541	?	ENSP00000392761	61
SIK3	ENST00000375300	NM_001281748.1, NM_025164.4	ENSP00000364449	61

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AP000936.1	ENST00000438127	?	1

Locus 244 (rs1204656)

SNiPAcard

Block annotations

Block info

genomic range	chr20:33,887,955-34,023,219
block size	135,265 bp
variant count	39 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.104 [-1.732 – 4.902]	gene(s) hit or close-by	GDF5, GDF5OS, UQCC1
phastCons	μ = 0.086 [0 – 1]	eQTL gene(s)	CEP250, CPNE1, EDEM2, EIF6, ERGIC3, FAM83C, GDF5, GGT7, MMP24-AS1, PROCR, RPL36P4, UQCC1
GERP++	μ = -0.206 [-5.99 – 4.04]	potentially regulated gene(s)	ACSS2, CEP250, CPNE1, ERGIC3, FER1L4, GSS, MYH7B, NFS1, RBM12, ROMO1, TRPC4AP
CADD score	μ = 3.939 [0.19 – 20.8]	disease gene(s)	GDF5, MYH7B, NFS1, GSS

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Height	<8.00×10-7	GWAS Catalog	18391951	1

Disease gene annotation

gene	trait	source DB	source entry/link
GDF5	Du Pan syndrome	DECIPHER	MIM:228900
GDF5	multiple synostoses syndrome type 2 (SYNS2)	DECIPHER	MIM:610017
GDF5	brachydactyly type C (BDC)	DECIPHER	MIM:113100
GDF5	brachydactyly type A2 (BDA2)	DECIPHER	MIM:112600
GDF5	brachydactyly type A1 (BDA1)	DECIPHER	MIM:112500
GDF5	symphalangism proximal syndrome (SYM1)	DECIPHER	MIM:185800
GDF5	acromesomelic chondrodysplasia Hunter-Thompson type (AMDH)	DECIPHER	MIM:201250
GDF5	acromesomelic chondrodysplasia Grebe type (AMDG)	DECIPHER	MIM:200700
GDF5	Acromesomelic dysplasia, Grebe type	OrphaNet	OrphaNet:2098
GDF5	Acromesomelic dysplasia, Hunter-Thomson type	OrphaNet	OrphaNet:968
GDF5	Proximal symphalangism	OrphaNet	OrphaNet:3250
GDF5	Multiple synostoses syndrome	OrphaNet	OrphaNet:3237
GDF5	Angel-shaped phalango-epiphyseal dysplasia	OrphaNet	OrphaNet:63442
GDF5	BRACHYDACTYLY TYPE C	OrphaNet	OrphaNet:93384
GDF5	Fibular aplasia - complex brachydactyly	OrphaNet	OrphaNet:2639
GDF5	BRACHYDACTYLY, TYPE A2	OrphaNet	OrphaNet:93396
GDF5	Brachydactyly, type A1	OrphaNet	OrphaNet:93388
MYH7B	Left ventricular noncompaction	OrphaNet	OrphaNet:54260
NFS1	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	OrphaNet	OrphaNet:397593
GSS	Glutathione synthetase deficiency with 5-oxoprolinuria	OrphaNet	OrphaNet:289846
GSS	Glutathione synthetase deficiency without 5-oxoprolinuria	OrphaNet	OrphaNet:289849
GSS	GLUTATHIONE SYNTHETASE DEFICIENCY OF ERYTHROCYTES, HEMOLYTIC [...]	OMIM	MIM:231900
GSS	GLUTATHIONE SYNTHETASE DEFICIENCY	OMIM	MIM:266130

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RPL36P4	?	ENSG00000224497	lung	9.15×10^{-9} (p-value)	GTEx Portal V6	38
UQCC1	?	ENSG00000101019	lung	1.53×10^{-7} (p-value)	GTEx Portal V6	39
GDF5	?	ENSG00000125965	lung	6.73×10^{-8} (p-value)	GTEx Portal V6	39
RPL36P4	?	ENSG00000224497	nucleus accumbens	5.91×10^{-7} (p-value)	GTEx Portal V6	28
RPL36P4	?	ENSG00000224497	tibial nerve	7.87×10^{-9} (p-value)	GTEx Portal V6	38
UQCC1	?	ENSG00000101019	tibial nerve	2.04×10^{-9} (p-value)	GTEx Portal V6	39
RPL36P4	?	ENSG00000224497	transformed fibroblasts	5.34×10^{-13} (p-value)	GTEx Portal V6	39
UQCC1	?	ENSG00000101019	transformed fibroblasts	2.70×10^{-27} (p-value)	GTEx Portal V6	39
ERGIC3	?	ENSG00000125991	transformed fibroblasts	1.57×10^{-6} (p-value)	GTEx Portal V6	18
EIF6	?	ENSG00000242372	transformed fibroblasts	9.71×10^{-6} (p-value)	GTEx Portal V6	1
UQCC1	?	ENSG00000101019	blood	3.08×10^{-7} (p-value)	GTEx Portal V6	39
RPL36P4	?	ENSG00000224497	blood	1.32×10^{-5} (p-value)	GTEx Portal V6	2
UQCC1	?	ENSG00000101019	tibial artery	5.22×10^{-7} (p-value)	GTEx Portal V6	31
RPL36P4	?	ENSG00000224497	tibial artery	4.48×10^{-6} (p-value)	GTEx Portal V6	15
RPL36P4	?	ENSG00000224497	esophagus mucosa	3.72×10^{-9} (p-value)	GTEx Portal V6	39
UQCC1	?	ENSG00000101019	esophagus mucosa	3.14×10^{-7} (p-value)	GTEx Portal V6	39
FAM83C	?	ENSG00000125998	esophagus mucosa	1.64×10^{-6} (p-value)	GTEx Portal V6	24
CEP250	?	ENSG00000126001	esophagus mucosa	9.34×10^{-6} (p-value)	GTEx Portal V6	9
RPL36P4	?	ENSG00000224497	thyroid	5.73×10^{-11} (p-value)	GTEx Portal V6	39
UQCC1	?	ENSG00000101019	thyroid	2.91×10^{-7} (p-value)	GTEx Portal V6	37
CPNE1	?	ENSG00000214078	thyroid	1.35×10^{-5} (p-value)	GTEx Portal V6	1
RPL36P4	?	ENSG00000224497	skeletal muscle	9.81×10^{-7} (p-value)	GTEx Portal V6	32
UQCC1	?	ENSG00000101019	skeletal muscle	1.71×10^{-11} (p-value)	GTEx Portal V6	39
CEP250	?	ENSG00000126001	skeletal muscle	8.25×10^{-11} (p-value)	GTEx Portal V6	38
RPL36P4	?	ENSG00000224497	putamen	1.09×10^{-8} (p-value)	GTEx Portal V6	28
RPL36P4	?	ENSG00000224497	cerebellum	3.29×10^{-5} (p-value)	GTEx Portal V6	9
UQCC1	?	ENSG00000101019	sun exposed skin	5.91×10^{-10} (p-value)	GTEx Portal V6	39
EIF6	?	ENSG00000242372	sun exposed skin	8.34×10^{-6} (p-value)	GTEx Portal V6	6
RPL36P4	?	ENSG00000224497	sun exposed skin	1.18×10^{-6} (p-value)	GTEx Portal V6	31
RPL36P4	?	ENSG00000224497	subcutaneous adipocytes	3.54×10^{-11} (p-value)	GTEx Portal V6	39
UQCC1	?	ENSG00000101019	subcutaneous adipocytes	2.57×10^{-10} (p-value)	GTEx Portal V6	39
UQCC1	ENST00000374384	ILMN_2381121	monocyte	9.15×10^{-6} (p-value)	Fairfax et al.	2
UQCC1	ENST00000359226					
UQCC1	ENST00000457259					
UQCC1	ENST00000482440					
UQCC1	ENST00000374380					

Locus 244 (sentinel SNP rs1204656 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

UQCC1	ENST00000472559					
UQCC1	ENST00000374394					
UQCC1	ENST00000453855					
UQCC1	ENST00000349714					
UQCC1	ENST00000496812					
UQCC1	ENST00000473982					
UQCC1	ENST00000424405					
UQCC1	ENST00000397556					
UQCC1	ENST00000374385					
CPNE1	ENST00000397443	ILMN_1670841	monocyte	6.05×10-6 (p-value)	Fairfax et al.	2
CPNE1	ENST00000317677		b-cell	1.19×10-4 (p-value)	Fairfax et al.	2
CPNE1	ENST00000498056					
CPNE1	ENST00000397442					
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000498814					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
GDF5	?	ENSG00000125965	pituitary	5.46×10-6 (p-value)	GTEx Portal V6	9
CPNE1	?	ENSG00000214078	blood	7.56×10-5 (q-value)	SeeQTL DB (HapMap)	6
RPL36P4	?	ENSG00000224497	adrenal gland	3.68×10-8 (p-value)	GTEx Portal V6	22
UQCC1	ENST00000374384	ILMN_2381121	blood	2.11×10-7 (p-value)	MuTHER consortium	12
UQCC1	ENST00000482440					
UQCC1	ENST00000457259					
UQCC1	ENST00000359226					
UQCC1	ENST00000374380					
UQCC1	ENST00000374394					
UQCC1	ENST00000472559					
UQCC1	ENST00000349714					
UQCC1	ENST00000453855					
UQCC1	ENST00000496812					
UQCC1	ENST00000473982					
UQCC1	ENST00000424405					
UQCC1	ENST00000397556					
UQCC1	ENST00000374385					
GGT7	ENST00000336431	ILMN_1737036	blood	5.13×10-5 (p-value)	MuTHER consortium	5
?	?	ILMN_1786186	skin	7.72×10-6 (p-value)	MuTHER consortium	7
CPNE1	ENST00000397443	ILMN_1670841	blood	1.49×10-9 (p-value)	MuTHER consortium	12
CPNE1	ENST00000317677		adipocyte	3.70×10-6 (p-value)	MuTHER consortium	12
CPNE1	ENST00000498056					
CPNE1	ENST00000397442					
CPNE1	ENST00000397445					
CPNE1	ENST00000415920					
CPNE1	ENST00000437340					
CPNE1	ENST00000462352					

CPNE1	ENST00000401607					
CPNE1	ENST00000498814					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
CPNE1	ENST00000397443	ILMN_2307025	blood	1.05×10^{-9} (p-value)	MuTHER consortium	12
CPNE1	ENST00000317677		adipocyte	8.95×10^{-8} (p-value)	MuTHER consortium	12
CPNE1	ENST00000498056		skin	2.73×10^{-5} (p-value)	MuTHER consortium	12
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000483495					
CPNE1	ENST00000473373					
GDF5	?	ENSG00000125965	muscularis mucosae	1.09×10^{-5} (p-value)	GTEx Portal V6	7
RPL36P4	?	ENSG00000224497	muscularis mucosae	2.23×10^{-5} (p-value)	GTEx Portal V6	7
UQCC1	?	ENSG00000101019	visceral adipocytes	1.02×10^{-7} (p-value)	GTEx Portal V6	12
GDF5	?	ENSG00000125965	sigmoid colon	6.58×10^{-6} (p-value)	GTEx Portal V6	9
UQCC1	ENST00000374384	ILMN_1745152	monocyte	1.61×10^{-24} (p-value)	Zeller et al.	4
UQCC1	ENST00000457259					
UQCC1	ENST00000359226					
UQCC1	ENST00000374380					
UQCC1	ENST00000472559					
UQCC1	ENST00000374394					
UQCC1	ENST00000349714					
UQCC1	ENST00000496812					
UQCC1	ENST00000374385					
MMP24-AS1	ENST00000566203	ILMN_1911503	blood	3.89×10^{-6} (p-value)	Westra et al.	11
CEP250	ENST00000397527	ILMN_1799011	blood	1.12×10^{-6} (p-value)	Westra et al.	9
EDEM2	ENST00000374491	ILMN_1711909	blood	7.10×10^{-7} (p-value)	Westra et al.	2
EDEM2	ENST00000374492					
UQCC1	?	ENSG00000101019	unexposed skin	1.72×10^{-6} (p-value)	GTEx Portal V6	3
PROCR	?	ENSG00000101000	pancreas	4.03×10^{-6} (p-value)	GTEx Portal V6	5
RPL36P4	?	ENSG00000224497	frontal cortex	2.41×10^{-6} (p-value)	GTEx Portal V6	19
RPL36P4	?	ENSG00000224497	transverse colon	1.77×10^{-6} (p-value)	GTEx Portal V6	10
UQCC1	?	ENSG00000101019	transverse colon	4.32×10^{-6} (p-value)	GTEx Portal V6	2
RPL36P4	?	ENSG00000224497	testis	1.43×10^{-5} (p-value)	GTEx Portal V6	4
UQCC1	?	ENSG00000101019	breast	8.32×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000293024	1	ENCP00000031386	ERGIC3
		ENCP00000031342	GSS
		ENCP00000031336	ACSS2
		ENCP00000031392	FER1L4
		ENCP00000031410	NFS1 ROMO1
		ENCP00000031393	FER1L4
		ENCP00000031390	FER1L4
		ENCP00000031366	CEP250
		ENCP00000031346	MYH7B
		ENCP00000031405	CPNE1 RBM12
		ENCP00000031347	TRPC4AP

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001448537 (promoter flanking region)	4	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, Yy1, USF1, DNase1
		HSMMtube	CTCF, H3K4me1, H3K9ac, H3K27ac, DNase1
		blood (K562)	Cjun, Cfos, CTCF, H3K36me3, DNase1, HDAC2, USF1, Rad21, FOSL1, Max
		skin (NHDF-AD)	H3K27ac, DNase1, CTCF, H3K4me1
		muscle (HSMM)	H3K9ac, H3K27ac, CTCF, H3K4me1, DNase1, H3K36me3
		liver (HepG2)	FOSL2, Jund, Rad21, H3K4me1, H3K9ac, H3K27ac, CTCF, H3K36me3, DNase1
		blood (GM12878)	DNase1, Yy1, Rad21, CTCF, H3K36me3
		lung (IMR90)	H4K5ac, H3K36me3, H3K4ac, H2BK120ac, H2BK20ac, H2AK5ac, H4K91ac, CTCF, H3K18ac, H3K27ac, DNase1, H4K8ac, H2BK12ac
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K27ac, H3K36me3
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K9ac, H3K27ac, H3K4me1
		NHLF	H3K36me3, H3K27ac, CTCF, DNase1, H3K4me1
		Osteobl	H3K27ac, H3K36me3, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF, H3K36me3, H3K4me1, H3K9ac, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Gabp, Cmyc, Max, TAF1, H3K4me1, H3K27ac, Jund, PolII, BAF155, Cjun, Ini1, Cfos, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, H3K4me1, Max, Cjun, H3K27ac, PolII, CTCF, Cmyc, DNase1
		A549	H3K4me3, H3K4me1, H3K4me2, CTCF, DNase1, H3K27ac, H3K36me3
ENSR00000400272 (promoter flanking region)	4	NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K4me2, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K27ac, DNase1, H3K4me1
		muscle (HSMM)	H3K27ac, DNase1, H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K4me1, H3K27ac, H3K4me2, PolII
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun
		liver (HepG2)	H3K36me3, FOXA1, H3K4me1
		lung (IMR90)	H2BK120ac, H4K91ac, DNase1, H2BK12ac, H3K27ac, H3K36me3
		A549	H3K4me3, DNase1, H3K27ac, H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K27ac, H3K4me1
ENSR00001665285 (enhancer)	3	monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00000400275 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	Rad21, CTCF, H3K4me1, H3K36me3, DNase1
		skin (NHDF-AD)	DNase1, CTCF

		muscle (HSMM)	H3K27ac, CTCF, H3K36me3, DNase1
		liver (HepG2)	FOSL2, Jund, HNF4G, HDAC2, Rad21, H3K4me1, p300, H3K4me2, HNF4A, FOXA1, H3K27ac, CTCF, Cmyc, H3K36me3, DNase1
		blood (GM12878)	CTCF
		nervous (NH-A)	DNase1, H3K36me3
		skin (NHEK)	H3K36me3, CTCF, H3K4me1, DNase1
		NHLF	H3K36me3, CTCF
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF, Cfos, H3K4me1, Jund, Cjun
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1, Cjun
		A549	H3K4me3, H3K4me2, CTCF, DNase1, H3K27ac, H3K36me3
ENSR00001448545	1	NHLF	DNase1
(promoter flanking region)		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K4me1, H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K36me3
ENSR00000400285	1	embryonic stem cell (H1ESC)	H3K36me3
(promoter flanking region)		HSMMtube	H3K36me3
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1, H3K4me1
		muscle (HSMM)	H3K36me3
		liver (HepG2)	H3K79me2, H3K4me1, H3K36me3
		lung (IMR90)	DNase1, H3K36me3, CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
		NHLF	DNase1, H3K4me1
		Osteobl	H3K27ac, CTCF
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	DNase1, CTCF
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GDF5	downstream gene variant	3202	ENST00000374369	NM_000557.3	ENSP00000363489	3
GDF5	downstream gene variant	3202	ENST00000374372	?	ENSP00000363492	3
GDF5OS	upstream gene variant	2884	ENST00000374375	?	ENSP00000363495	3
UQCC1	upstream gene variant	4981	ENST00000498651	?	?	1
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000374385	NM_018244.4	ENSP00000363506	4
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000374380	NM_001184977.1	ENSP00000363501	4
UQCC1	upstream gene variant	3961	ENST00000491125	?	?	1
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000374394	?	ENSP00000363515	4
UQCC1	downstream gene variant, upstream gene variant	385	ENST00000443429	?	ENSP00000416246	3
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000472559	?	?	5
UQCC1	upstream gene variant	3964	ENST00000491040	?	ENSP00000420584	1
UQCC1	downstream gene variant, upstream gene variant	886	ENST00000453855	?	ENSP00000390334	4

UQCC1	downstream gene variant, upstream gene variant	19	ENST00000457259	?	ENSP00000411024	4
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000349714	?	ENSP00000335364	4
UQCC1	downstream gene variant, upstream gene variant	162	ENST00000497717	?	?	3
UQCC1	downstream gene variant, upstream gene variant	907	ENST00000496812	?	?	5
UQCC1	downstream gene variant, upstream gene variant	455	ENST00000438533	?	ENSP00000398531	3
UQCC1	downstream gene variant, upstream gene variant	1424	ENST00000473982	?	?	4
UQCC1	upstream gene variant	528	ENST00000495752	?	?	1
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000359226	?	ENSP00000352161	4
UQCC1	upstream gene variant	3964	ENST00000397553	?	ENSP00000380685	1
UQCC1	downstream gene variant, upstream gene variant	1247	ENST00000482440	?	?	5
UQCC1	upstream gene variant	3963	ENST00000397554	?	ENSP00000380686	1
UQCC1	downstream gene variant, upstream gene variant	1444	ENST00000424405	?	ENSP00000399713	4
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000374384	NM_199487.2	ENSP00000363505	4
UQCC1	downstream gene variant, upstream gene variant	840	ENST00000397556	?	ENSP00000380688	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GDF5	ENST00000374369	NM_000557.3	ENSP00000363489	1
GDF5	ENST00000374372	?	ENSP00000363492	1
UQCC1	ENST00000397554	?	ENSP00000380686	7
UQCC1	ENST00000443429	?	ENSP00000416246	22
UQCC1	ENST00000472559	?	?	1
UQCC1	ENST00000374380	NM_001184977.1	ENSP00000363501	23
UQCC1	ENST00000374394	?	ENSP00000363515	23
UQCC1	ENST00000497717	?	?	15
UQCC1	ENST00000453855	?	ENSP00000390334	23
UQCC1	ENST00000397553	?	ENSP00000380685	7
UQCC1	ENST00000349714	?	ENSP00000335364	23
UQCC1	ENST00000374385	NM_018244.4	ENSP00000363506	23
UQCC1	ENST00000496812	?	?	1
UQCC1	ENST00000438533	?	ENSP00000398531	22
UQCC1	ENST00000473982	?	?	3
UQCC1	ENST00000491125	?	?	7
UQCC1	ENST00000359226	?	ENSP00000352161	23
UQCC1	ENST00000491040	?	ENSP00000420584	7
UQCC1	ENST00000457259	?	ENSP00000411024	23
UQCC1	ENST00000495752	?	?	3
UQCC1	ENST00000424405	?	ENSP00000399713	23
UQCC1	ENST00000374384	NM_199487.2	ENSP00000363505	23

UQCC1	ENST00000397556	?	ENSP00000380688	21

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GDF5OS	ENST00000374375	?	ENSP00000363495	1

Locus 246 (rs13157656)

SNiPAcard

Block annotations

Block info

genomic range	chr5:40,960,166-40,985,653
block size	25,488 bp
variant count	23 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.153 [-2.108 – 1.183]	gene(s) hit or close-by	C7, RP11-301A5.2
phastCons	μ = 0.069 [0 – 0.496]	eQTL gene(s)	–
GERP++	μ = -0.671 [-4.69 – 3.22]	potentially regulated gene(s)	–
CADD score	μ = 3.816 [0.138 – 11.55]	disease gene(s)	C7

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C7	COMPLEMENT COMPONENT 7 DEFICIENCY	OMIM	MIM:610102
C7	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
C7	missense variant	ENST00000313164	NM_000587.2	ENSP00000322061	P/T	Cca/Aca	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001692911 (enhancer)	2	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001692912 (enhancer)	1	cervix (HeLa-S3)	H3K27me3
		NHLF	DNase1
		lung (IMR90)	DNase1, H3K18ac, H2BK120ac, H4K91ac
		nervous (NH-A)	DNase1
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C7	upstream gene variant, downstream gene variant	252	ENST00000494960	?	?	8
C7	upstream gene variant, downstream gene variant	148	ENST00000486779	?	?	8
C7	upstream gene variant	134	ENST00000513922	?	?	5
C7	upstream gene variant, downstream gene variant	588	ENST00000464864	?	?	10
C7	downstream gene variant	50	ENST00000313164	NM_000587.2	ENSP00000322061	2
RP11-301A5.2	downstream gene variant, upstream gene variant	163	ENST00000504890	?	?	13

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C7	ENST00000494960	?	?	5
C7	ENST00000313164	NM_000587.2	ENSP00000322061	18
C7	ENST00000486779	?	?	10
C7	ENST00000513922	?	?	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C7	ENST00000313164	NM_000587.2	ENSP00000322061	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C7	ENST00000486779	?	1

Locus 247 (rs2871473)

SNiPAcard

Block annotations

Block info

genomic range	chr2:103,105,611-103,166,325
block size	60,715 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.928 [-2.359 – 0.089]	**gene(s) hit or close-by**	SLC9A4
phastCons	μ = 0.001 [0 – 0.002]	**eQTL gene(s)**	AC007278.3, IL18RAP
GERP++	μ = -2.044 [-6.09 – 0.158]	**potentially regulated gene(s)**	–
CADD score	μ = 0.546 [0.126 – 1.397]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AC007278.3	?	ENSG00000234389	atrial appendage	1.67×10^{-7} (p-value)	GTEx Portal V6	5
IL18RAP	?	ENSG00000115607	blood	1.37×10^{-7} (p-value)	GTEx Portal V6	5
AC007278.3	?	ENSG00000234389	blood	3.34×10^{-10} (p-value)	GTEx Portal V6	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLC9A4	ENST00000295269	NM_001011552.3	ENSP00000295269	3

Locus 248 (rs7263120)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,673,017-23,727,735
block size	54,719 bp
variant count	53 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.786 [-4.406 – 0.696]	gene(s) hit or close-by	CST1, CST2P1, CST4
phastCons	μ = 0.015 [0 – 0.164]	eQTL gene(s)	CST1, CST3
GERP++	μ = -0.390 [-2.17 – 1.08]	potentially regulated gene(s)	CST11, CST4, CST8, CSTL1, CSTP1, GGTLC1, XXyac-YX60D10.1
CADD score	μ = 1.573 [0.012 – 7.042]	disease gene(s)	CST3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CST3	CEREBRAL AMYLOID ANGIOPATHY, CST3-RELATED	OMIM	MIM:105150
CST3	MACULAR DEGENERATION, AGE-RELATED, 11	OMIM	MIM:611953
CST3	Hereditary cerebral hemorrhage with amyloidosis, Icelandic type	OrphaNet	OrphaNet:100008

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST1	?	ENSG00000170373	tibial nerve	1.67×10^{-7} (p-value)	GTEx Portal V6	51
CST3	ENST00000398411	ILMN_1800354	adipocyte	2.52×10^{-5} (p-value)	MuTHER consortium	2
CST3	ENST00000376925					
CST3	ENST00000398409					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000290703	1	ENCP00000031098	GGTLC1
		ENCP00000031079	CST11
		ENCP00000031078	CSTL1
		ENCP00000031093	XXyac-YX60D10.1
		ENCP00000031081	CST8
		ENCP00000031096	CSTP1
ENCE00000290742	1	ENCP00000031090	CST4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000399219 (promoter flanking region)	4	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	DNase1
		blood (DND-41)	H3K27me3

		skin (NHDF-AD)	DNase1, H3K4me1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	H3K27me3, DNase1
		lung (IMR90)	DNase1
ENSR00000399220 (enhancer)	2	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		liver (HepG2)	H3K4me1, H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CST1	downstream gene variant	633	ENST00000398402	?	ENSP00000381439	8
CST1	downstream gene variant	455	ENST00000304749	NM_001898.2	ENSP00000305731	8
CST2P1	downstream gene variant, upstream gene variant	407	ENST00000425770	?	?	11
CST4	upstream gene variant	3340	ENST00000217423	NM_001899.2	ENSP00000217423	4

Locus 249 (rs9640161)

SNiPAcard

Block annotations

Block info

genomic range	chr7:150,026,169-150,046,747
block size	20,579 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.994 [-1.551 – -0.031]	gene(s) hit or close-by	LRRC61, ZBED6CL
phastCons	μ = 0.016 [0 – 0.025]	eQTL gene(s)	ACTR3C, LRRC61, RARRES2, REPIN1, RP4-584D14.5, RP4-584D14.7, ZBED6CL
GERP++	μ = -0.366 [-1.05 – 0.0465]	potentially regulated gene(s)	–
CADD score	μ = 5.794 [2.935 – 10.31]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LRRC61	?	ENSG00000127399	atrial appendage	2.53×10^{-9} (p-value)	GTEx Portal V6	3
RP4-584D14.7	?	ENSG00000261305	transformed fibroblasts	4.39×10^{-7} (p-value)	GTEx Portal V6	3
RARRES2	?	ENSG00000106538	transformed fibroblasts	3.72×10^{-15} (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	transformed fibroblasts	1.08×10^{-15} (p-value)	GTEx Portal V6	3
ACTR3C	?	ENSG00000106526	transformed fibroblasts	1.63×10^{-8} (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	breast	4.39×10^{-13} (p-value)	GTEx Portal V6	3
RP4-584D14.7	?	ENSG00000261305	breast	2.16×10^{-6} (p-value)	GTEx Portal V6	1
ZBED6CL	?	ENSG00000188707	blood	7.94×10^{-9} (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	blood	1.66×10^{-12} (p-value)	GTEx Portal V6	3
RARRES2	?	ENSG00000106538	skeletal muscle	3.14×10^{-10} (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	skeletal muscle	1.44×10^{-26} (p-value)	GTEx Portal V6	3
REPIN1	?	ENSG00000214022	skeletal muscle	1.12×10^{-35} (p-value)	GTEx Portal V6	3
ACTR3C	?	ENSG00000106526	skeletal muscle	2.01×10^{-14} (p-value)	GTEx Portal V6	3
RP4-584D14.5	?	ENSG00000240449	skeletal muscle	1.05×10^{-7} (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	putamen	1.51×10^{-7} (p-value)	GTEx Portal V6	1
LRRC61	?	ENSG00000127399	EBV lymphocytes	9.32×10^{-7} (p-value)	GTEx Portal V6	1
LRRC61	?	ENSG00000127399	unexposed skin	2.54×10^{-12} (p-value)	GTEx Portal V6	3
ZBED6CL	?	ENSG00000188707	unexposed skin	2.33×10^{-8} (p-value)	GTEx Portal V6	1
ZBED6CL	?	ENSG00000188707	sun exposed skin	1.15×10^{-7} (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	sun exposed skin	2.69×10^{-21} (p-value)	GTEx Portal V6	3
ZBED6CL	?	ENSG00000188707	ovary	2.48×10^{-10} (p-value)	GTEx Portal V6	3

LRRC61	?	ENSG00000127399	ovary	1.07×10-6 (p-value)	GTEx Portal V6	1
RP4-584D14.7	?	ENSG00000261305	left ventricle	1.55×10-10 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	left ventricle	4.88×10-11 (p-value)	GTEx Portal V6	3
REPIN1	?	ENSG00000214022	left ventricle	9.63×10-9 (p-value)	GTEx Portal V6	3
RP4-584D14.7	?	ENSG00000261305	aorta	6.33×10-8 (p-value)	GTEx Portal V6	3
RARRES2	?	ENSG00000106538	aorta	2.77×10-8 (p-value)	GTEx Portal V6	1
LRRC61	?	ENSG00000127399	aorta	9.45×10-15 (p-value)	GTEx Portal V6	3
ACTR3C	?	ENSG00000106526	aorta	3.56×10-6 (p-value)	GTEx Portal V6	1
RARRES2	?	ENSG00000106538	subcutaneous adipocytes	6.71×10-8 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	subcutaneous adipocytes	1.10×10-15 (p-value)	GTEx Portal V6	3
REPIN1	?	ENSG00000214022	subcutaneous adipocytes	1.51×10-7 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	caudate basal ganglia	4.68×10-13 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	visceral adipocytes	6.40×10-9 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	stomach	3.99×10-12 (p-value)	GTEx Portal V6	3
ZBED6CL	?	ENSG00000188707	stomach	4.15×10-8 (p-value)	GTEx Portal V6	2
LRRC61	?	ENSG00000127399	cortex	1.12×10-7 (p-value)	GTEx Portal V6	1
RP4-584D14.7	?	ENSG00000261305	tibial nerve	2.01×10-6 (p-value)	GTEx Portal V6	3
RARRES2	?	ENSG00000106538	tibial nerve	3.79×10-9 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	tibial nerve	1.74×10-17 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	pituitary	1.58×10-9 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	anterior cingulate cortex	8.22×10-8 (p-value)	GTEx Portal V6	1
RP4-584D14.7	?	ENSG00000261305	esophagus mucosa	2.08×10-9 (p-value)	GTEx Portal V6	3
ZBED6CL	?	ENSG00000188707	esophagus mucosa	3.59×10-20 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	esophagus mucosa	9.34×10-16 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	testis	2.79×10-20 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	hypothalamus	6.03×10-9 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	frontal cortex	8.28×10-10 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	pancreas	7.67×10-11 (p-value)	GTEx Portal V6	3
ZBED6CL	?	ENSG00000188707	pancreas	1.59×10-9 (p-value)	GTEx Portal V6	3
RARRES2	?	ENSG00000106538	pancreas	3.40×10-6 (p-value)	GTEx Portal V6	3
RP4-584D14.7	?	ENSG00000261305	muscularis mucosae	4.59×10-10 (p-value)	GTEx Portal V6	3
ZBED6CL	?	ENSG00000188707	muscularis mucosae	7.78×10-15 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	muscularis mucosae	6.56×10-10 (p-value)	GTEx Portal V6	3
REPIN1	?	ENSG00000214022	muscularis mucosae	1.12×10-6 (p-value)	GTEx Portal V6	2
ZBED6CL	?	ENSG00000188707	lung	5.61×10-9 (p-value)	GTEx Portal V6	3

LRRC61	?	ENSG00000127399	lung	4.84×10-16 (p-value)	GTEx Portal V6	3
RP4-584D14.7	?	ENSG00000261305	tibial artery	2.30×10-8 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	tibial artery	2.26×10-11 (p-value)	GTEx Portal V6	3
ACTR3C	?	ENSG00000106526	tibial artery	2.30×10-7 (p-value)	GTEx Portal V6	1
RP4-584D14.7	?	ENSG00000261305	thyroid	2.83×10-6 (p-value)	GTEx Portal V6	1
ZBED6CL	?	ENSG00000188707	thyroid	3.87×10-10 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	thyroid	2.87×10-26 (p-value)	GTEx Portal V6	3
ZBED6CL	?	ENSG00000188707	prostate	1.20×10-7 (p-value)	GTEx Portal V6	1
LRRC61	?	ENSG00000127399	transverse colon	6.53×10-10 (p-value)	GTEx Portal V6	3
ZBED6CL	?	ENSG00000188707	transverse colon	5.01×10-12 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	liver	2.51×10-9 (p-value)	GTEx Portal V6	1
LRRC61	?	ENSG00000127399	terminal ileum	3.56×10-9 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	nucleus accumbens	2.92×10-11 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	spleen	6.82×10-11 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	coronary artery	5.43×10-8 (p-value)	GTEx Portal V6	1
ZBED6CL	?	ENSG00000188707	sigmoid colon	3.21×10-7 (p-value)	GTEx Portal V6	1
ZBED6CL	?	ENSG00000188707	gastroesophageal junction	9.67×10-10 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	cerebellar hemisphere	1.14×10-11 (p-value)	GTEx Portal V6	3
LRRC61	?	ENSG00000127399	cerebellum	2.66×10-8 (p-value)	GTEx Portal V6	2
LRRC61	?	ENSG00000127399	hippocampus	6.31×10-10 (p-value)	GTEx Portal V6	3
RARRES2	ENST00000466675	ILMN_1810844	skin	3.55×10-9 (p-value)	MuTHER consortium	1
RARRES2	ENST00000482669		blood	1.56×10-12 (p-value)	MuTHER consortium	1
RARRES2	ENST00000223271		adipocyte	5.22×10-13 (p-value)	MuTHER consortium	1
RARRES2	ENST00000478771					
REPIN1	ENST00000425389	ILMN_2404385	adipocyte	1.31×10-19 (p-value)	MuTHER consortium	1
REPIN1	ENST00000397281					
REPIN1	ENST00000489432					
REPIN1	ENST00000444957					
REPIN1	ENST00000479668					
LRRC61	ENST00000359623	ILMN_1669722	blood	5.28×10-25 (p-value)	MuTHER consortium	1
LRRC61	ENST00000323078					
ZBED6CL	ENST00000343855	ILMN_1678863	blood	8.46×10-13 (p-value)	MuTHER consortium	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001564976	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K27me3, CTCF, Rad21, H3K4me1, Tcf12, H3K4me2, H3K27ac
(promoter flanking region)		HSMMtube	DNase1
		blood (K562)	PolII, H3K36me3
		blood (DND-41)	H3K36me3

muscle (HSMM)	DNase1
cervix (HeLa-S3)	PolII, H3K36me3
monocytes (Monocytes-CD14+)	H3K36me3
liver (HepG2)	H3K36me3
blood (GM12878)	PolII, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LRRC61	downstream gene variant	294	ENST00000463441	?	?	1
LRRC61	downstream gene variant	3161	ENST00000471872	?	?	1
ZBED6CL	upstream gene variant	709	ENST00000343855	NM_138434.2	ENSP00000343242	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LRRC61	ENST00000493307	?	ENSP00000420560	1
LRRC61	ENST00000359623	NM_001142928.1	ENSP00000352642	1
LRRC61	ENST00000323078	NM_023942.2	ENSP00000339047	1

Locus 250 (rs10754218)

SNiPAcard

Block annotations

Block info

genomic range	chr1:197,162,526-197,165,267
block size	2,742 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.239 [-2.612 – 0.829]	gene(s) hit or close-by	ZBTB41
phastCons	μ = 0.081 [0 – 0.389]	eQTL gene(s)	CFHR3
GERP++	μ = -0.548 [-4.43 – 1.87]	potentially regulated gene(s)	–
CADD score	μ = 4.600 [0.478 – 14.6]	disease gene(s)	CFHR3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	lung	4.33×10-7 (p-value)	GTEx Portal V6	4

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001527247 (promoter flanking region)	3	cervix (HeLa-S3)	H3K4me2
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (GM12878)	H3K4me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2, H3K4me2
		muscle (HSMM)	H3K36me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ZBTB41	ENST00000467322	?	?	5
ZBTB41	ENST00000367405	NM_194314.2	ENSP00000356375	5

Locus 251 (rs11209026)

SNiPAcard

Block annotations

Block info

genomic range	chr1:67,684,934-67,743,552
block size	58,619 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.033 [-2.084 – 2.716]	**gene(s) hit or close-by**	AL109843.1, C1orf141, IL23R, RNU4ATAC4P, RP11-131O15.2
phastCons	μ = 0.059 [0 – 0.959]	**eQTL gene(s)**	–
GERP++	μ = 0.161 [-4.62 – 5.19]	**potentially regulated gene(s)**	–
CADD score	μ = 3.253 [0.074 – 18.58]	**disease gene(s)**	IL23R

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Inflammatory bowel disease	$<8.00\times10^{-161}$	GWAS Catalog	23128233		1
Crohn's disease (time to surgery)	$<4.00\times10^{-14}$	GWAS Catalog	22936669		1
Crohn's disease (time to surgery)	$<1.00\times10^{-18}$	GWAS Catalog	22412388		1
Crohn's disease (time to surgery)	$<4.00\times10^{-21}$	GWAS Catalog	22293688		1
Ankylosing spondylitis	$<2.00\times10^{-17}$	GWAS Catalog	21743469		1
Ulcerative colitis	$<5.00\times10^{-28}$	GWAS Catalog	21297633		1
Crohn's disease (time to surgery)	$<1.00\times10^{-64}$	GWAS Catalog	21102463		1
Psoriasis	$<7.00\times10^{-7}$	GWAS Catalog	20953190		1
Crohn's disease (time to surgery)	$<1.00\times10^{-6}$	GWAS Catalog	20570966		1
Ankylosing spondylitis	$<9.00\times10^{-14}$	GWAS Catalog	20062062		1
Ulcerative colitis	$<3.00\times10^{-10}$	GWAS Catalog	19915572		1
Ulcerative colitis	$<1.00\times10^{-8}$	GWAS Catalog	19122664		1
Inflammatory bowel disease	$<7.00\times10^{-11}$	GWAS Catalog	18758464		1
Crohn's disease (time to surgery)	$<7.00\times10^{-63}$	GWAS Catalog	18587394		1
Crohn's disease (time to surgery)	$<1.00\times10^{-8}$	GWAS Catalog	17804789		3
Crohn's disease (time to surgery)	$<2.00\times10^{-18}$	GWAS Catalog	17447842		1
Inflammatory bowel diseases	4.59×10^{-11}	dbGaP	pha002847	dbGaP	2

Variant annotation

trait	type	source DB	source entry/link		Variant(s)
Psoriasis, protection against	protective	ClinVar	RCV000003255.2	ClinVar	1
Inflammatory bowel disease 17, protection against	protective	ClinVar	RCV000003254.2	ClinVar	1
?	HGMD curated	HGMD	CM066884	HGMD	1
INFLAMMATORY BOWEL DISEASE 17, PROTECTION AGAINST	OMIM curated	OMIM	MIM:607562	OMIM	1
INFLAMMATORY BOWEL DISEASE 17, PROTECTION AGAINST	UniProt/OMIM curated	UniProt	MIM:612261 (IL23R)	OMIM	1

Disease gene annotation

gene	trait	source DB	source entry/link
IL23R	PSORIASIS 7, SUSCEPTIBILITY TO	OMIM	MIM:605606
IL23R	INFLAMMATORY BOWEL DISEASE 17	OMIM	MIM:612261
IL23R	Behcet disease	OrphaNet	OrphaNet:117

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IL23R	missense variant	ENST00000425614	?	ENSP00000387640	Q/R	cAa/cGa	?	?	1
IL23R	missense variant	ENST00000347310	NM_144701.2	ENSP00000321345	Q/R	cAa/cGa	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000283180 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H3K27me3
		blood (K562)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		blood (GM12878)	Rad21, CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL109843.1	downstream gene variant, upstream gene variant	748	ENST00000408806	?	?	5
C1orf141	upstream gene variant	334	ENST00000371007	NM_001276351.1	ENSP00000360046	3
C1orf141	upstream gene variant	333	ENST00000448166	?	ENSP00000415519	3
IL23R	downstream gene variant	746	ENST00000473881	?	ENSP00000486667	1
IL23R	upstream gene variant, downstream gene variant	306	ENST00000395227	?	ENSP00000378652	2
IL23R	downstream gene variant	1173	ENST00000425614	?	ENSP00000387640	1
IL23R	downstream gene variant	442	ENST00000347310	NM_144701.2	ENSP00000321345	1
RNU4ATAC4P	downstream gene variant, upstream gene variant	772	ENST00000516307	?	?	4
RP11-131O15.2	upstream gene variant	183	ENST00000432257	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1orf141	ENST00000371007	NM_001276351.1	ENSP00000360046	3
C1orf141	ENST00000448166	?	ENSP00000415519	3
IL23R	ENST00000347310	NM_144701.2	ENSP00000321345	16
IL23R	ENST00000425614	?	ENSP00000387640	16

IL23R	ENST00000473881	?	ENSP00000486667	17
IL23R	ENST00000395227	?	ENSP00000378652	16

Locus 251 (sentinel SNP rs11209026 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 252 (rs672413)

SNiPAcard

Block annotations

Block info

genomic range	chr5:78,278,229-78,278,299
block size	71 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.353 [-0.918 – 0.212]	**gene(s) hit or close-by**	ARSB
phastCons	μ = 0.000 [0 – 0]	**eQTL gene(s)**	ARSB, BHMT
GERP++	μ = -1.920 [-2.1 – -1.74]	**potentially regulated gene(s)**	–
CADD score	μ = 2.612 [2.011 – 3.213]	**disease gene(s)**	ARSB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Blood and toenail selenium levels	$<5.00\times10^{-14}$	GWAS Catalog	25343990	1
betaine	2.73×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
ARSB	mucopolysaccharidosis type 6 (MPS6)	DECIPHER	MIM:253200
ARSB	MULTIPLE SULFATASE DEFICIENCY	OMIM	MIM:272200
ARSB	MUCOPOLYSACCHARIDOSIS TYPE VI	OMIM	MIM:253200
ARSB	Mucopolysaccharidosis type 6, rapidly progressing	OrphaNet	OrphaNet:276212
ARSB	Mucopolysaccharidosis type 6, slowly progressing	OrphaNet	OrphaNet:276223

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
BHMT	ENST00000274353	ILMN_1656638	adipocyte	3.91×10^{-5} (p-value)	MuTHER consortium	1
ARSB	ENST00000264914	ILMN_1722045	blood	2.82×10^{-6} (p-value)	Westra et al.	1
ARSB	ENST00000396151					
ARSB	ENST00000565165					
ARSB	ENST00000521800					
ARSB	ENST00000264914	ILMN_1722045	liver	$<2.2\times10^{-16}$ (p-value)	Innocenti et al.	1
ARSB	ENST00000396151					
ARSB	ENST00000565165					
ARSB	ENST00000521800					
ARSB	?	ENSG00000113273	liver	2.04×10^{-9} (p-value)	GTEx Portal V6	2
ARSB	?	ENSG00000113273	sun exposed skin	3.05×10^{-11} (p-value)	GTEx Portal V6	2
ARSB	?	ENSG00000113273	tibial artery	4.84×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ARSB	downstream gene variant	2636	ENST00000521117	?	ENSP00000428611	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ARSB	ENST00000264914	NM_000046.3	ENSP00000264914	2
ARSB	ENST00000396151	NM_198709.2	ENSP00000379455	2
ARSB	ENST00000565165	?	ENSP00000456339	2

Locus 253 (rs33932084)

SNiPAcard

Block annotations

Block info

genomic range	chr6:27,771,106-28,768,698
block size	997,593 bp
variant count	151 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.110 [-3.56 – 5.072]	gene(s) hit or close-by	AL009179.1, AL022393.7, AL022393.9, AL049822.1, AL121932.1, COX11P1, GPX6, HIST1H1B, HIST1H2AI, HIST1H2AJ, HIST1H2AK, HIST1H2AL, HIST1H2AM, HIST1H2BL, HIST1H2BM, HIST1H2BN, HIST1H2BO, HIST1H2BPS2, HIST1H3H, HIST1H3I, HIST1H4J, HIST1H4K, HIST1H4L, HIST1H4PS1, LINC00533, NKAPL, NOP56P1, OR1F12, OR2B2, OR2B7P, OR2B8P, OR2W2P, OR2W4P, OR2W6P, PGBD1, RNU7-26P, RP1-265C24.5, RP5-874C20.6, RP5-874C20.7, RP5-874C20.8, RPLP2P1, RPSAP2, TOB2P1, Z98744.1, Z98744.2, ZBED9, ZKSCAN3, ZKSCAN8, ZNF165, ZNF192P1, ZNF192P2, ZSCAN12, ZSCAN12P1, ZSCAN16, ZSCAN16-AS1, ZSCAN23, ZSCAN26, ZSCAN31, ZSCAN9
phastCons	μ = 0.083 [0 – 1]	eQTL gene(s)	ABT1, AL022393.7, BTN2A2, BTN3A2, BTN3A3, HIST1H2BK, HIST1H3I, HIST1H3J, HIST1H4J, HLA-C, HLA-F-AS1, HLA-G, LINC01556, OR2B7P, OR2B8P, PRSS16, RP1-265C24.5, RP1-313I6.12, ZFP57, ZKSCAN3, ZNF165, ZNF192P1, ZNF391, ZSCAN23, ZSCAN26, ZSCAN31
GERP++	μ = -0.199 [-6.81 – 4.37]	potentially regulated gene(s)	GPX5, GPX5, GPX6, GPX6, HIST1H1B, HIST1H2AI, HIST1H2BL, HIST1H2BPS2, HIST1H4J, HIST1H4K, HIST1H4L, OR2B2, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2U1P, OR2U1P, OR2U1P, OR2U1P, OR2U1P, RSL24D1P1, ZKSCAN3, ZSCAN12P1
CADD score	μ = 4.950 [0.003 – 22.8]	disease gene(s)	ZFP57

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
betaine	7.17×10-6	Metabolomics GWAS Server	24816252		24
phenol sulfate	4.67×10-6	Metabolomics GWAS Server	24816252		5
Lupus erythematosus, systemic	6.28×10-6	dbGaP	pha002867	dbGaP	4
Lupus erythematosus, systemic	1.21×10-9	dbGaP	pha002848	dbGaP	4

Disease gene annotation

gene	trait	source DB	source entry/link	
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410	OMIM®
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410	OMIM®
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
OR2B2	missense variant	ENST00000303324	NM_033057.2	ENSP00000304419	2	2			2

PGBD1	missense variant	ENST00000259883	NM_001184743.1, NM_032507.3	ENSP00000259883	S/N	aGt/aAt	?	?	1
ZSCAN12	missense variant	ENST00000396827	?	ENSP00000380039	T/M	aCg/aTg	?	?	1
ZSCAN12	missense variant	ENST00000361028	?	ENSP00000354305	T/M	aCg/aTg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AL022393.7	?	ENSG00000216901	tibial nerve	2.04×10^{-7} (p-value)	GTEx Portal V6	111
ZFP57	?	ENSG00000204644	tibial nerve	3.03×10^{-7} (p-value)	GTEx Portal V6	39
ZSCAN26	?	ENSG00000197062	thyroid	1.92×10^{-7} (p-value)	GTEx Portal V6	143
OR2B8P	?	ENSG00000182477	thyroid	5.00×10^{-10} (p-value)	GTEx Portal V6	148
OR2B7P	?	ENSG00000187763	thyroid	5.70×10^{-7} (p-value)	GTEx Portal V6	145
ZNF192P1	?	ENSG00000226314	thyroid	3.51×10^{-7} (p-value)	GTEx Portal V6	110
RP1-265C24.5	?	ENSG00000219392	thyroid	3.29×10^{-5} (p-value)	GTEx Portal V6	14
ZFP57	?	ENSG00000204644	thyroid	2.42×10^{-11} (p-value)	GTEx Portal V6	40
RP1-265C24.5	?	ENSG00000219392	skeletal muscle	9.19×10^{-9} (p-value)	GTEx Portal V6	149
AL022393.7	?	ENSG00000216901	skeletal muscle	2.24×10^{-13} (p-value)	GTEx Portal V6	148
ZSCAN23	?	ENSG00000187987	skeletal muscle	2.13×10^{-6} (p-value)	GTEx Portal V6	110
ZNF192P1	?	ENSG00000226314	skeletal muscle	1.02×10^{-5} (p-value)	GTEx Portal V6	3
ZFP57	?	ENSG00000204644	skeletal muscle	5.24×10^{-6} (p-value)	GTEx Portal V6	4
HIST1H3J	?	ENSG00000197153	testis	7.04×10^{-7} (p-value)	GTEx Portal V6	100
AL022393.7	?	ENSG00000216901	testis	1.10×10^{-6} (p-value)	GTEx Portal V6	61
RP1-265C24.5	?	ENSG00000219392	testis	9.76×10^{-8} (p-value)	GTEx Portal V6	56
RP1-313I6.12	?	ENSG00000272009	testis	3.48×10^{-6} (p-value)	GTEx Portal V6	2
OR2B7P	?	ENSG00000187763	testis	5.52×10^{-6} (p-value)	GTEx Portal V6	2
ZSCAN26	?	ENSG00000197062	transverse colon	1.13×10^{-6} (p-value)	GTEx Portal V6	16
AL022393.7	?	ENSG00000216901	cerebellum	1.41×10^{-6} (p-value)	GTEx Portal V6	147
PRSS16	?	ENSG00000112812	cerebellum	2.09×10^{-9} (p-value)	GTEx Portal V6	64
ZNF192P1	?	ENSG00000226314	cerebellum	1.85×10^{-5} (p-value)	GTEx Portal V6	6
OR2B8P	?	ENSG00000182477	sun exposed skin	1.04×10^{-7} (p-value)	GTEx Portal V6	147
ZKSCAN3	?	ENSG00000189298	sun exposed skin	1.94×10^{-5} (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	sun exposed skin	5.44×10^{-9} (p-value)	GTEx Portal V6	40
RP1-265C24.5	?	ENSG00000219392	left ventricle	1.09×10^{-9} (p-value)	GTEx Portal V6	147
AL022393.7	?	ENSG00000216901	left ventricle	2.81×10^{-6} (p-value)	GTEx Portal V6	57
ZNF165	?	ENSG00000197279	left ventricle	1.33×10^{-5} (p-value)	GTEx Portal V6	1
ZNF192P1	?	ENSG00000226314	left ventricle	6.32×10^{-6} (p-value)	GTEx Portal V6	5
RP1-265C24.5	?	ENSG00000219392	subcutaneous adipocytes	3.87×10^{-8} (p-value)	GTEx Portal V6	113

RP1-265C24.5	?	ENSG00000219392	subcutaneous adipocytes	3.87×10-8 (p-value)	GTEx Portal V6	113
HIST1H4J	?	ENSG00000197238	subcutaneous adipocytes	2.04×10-7 (p-value)	GTEx Portal V6	142
RP1-313I6.12	?	ENSG00000272009	subcutaneous adipocytes	1.45×10-5 (p-value)	GTEx Portal V6	14
HIST1H2BK	?	ENSG00000197903	subcutaneous adipocytes	1.14×10-5 (p-value)	GTEx Portal V6	24
ZNF391	?	ENSG00000124613	subcutaneous adipocytes	1.08×10-5 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	subcutaneous adipocytes	6.15×10-7 (p-value)	GTEx Portal V6	34
ZSCAN26	ENST00000421553	ILMN_1760982	blood	1.05×10-6 (p-value)	MuTHER consortium	37
ZSCAN26	ENST00000614088					
ZSCAN26	ENST00000316606					
ZSCAN26	ENST00000619937					
ZSCAN31	ENST00000396838	ILMN_1655748	skin	7.67×10-7 (p-value)	MuTHER consortium	38
ZSCAN31	ENST00000344279		adipocyte	2.79×10-5 (p-value)	MuTHER consortium	38
ZSCAN31	ENST00000414429					
ZSCAN31	ENST00000439158					
ZSCAN31	ENST00000611469					
ZSCAN31	ENST00000446474					
HIST1H3I	ENST00000616365	ILMN_2207865	b-cell	2.79×10-4 (p-value)	Fairfax et al.	6
BTN2A2	ENST00000471116	ILMN_2256894	b-cell	4.07×10-6 (p-value)	Fairfax et al.	14
BTN2A2	ENST00000356709					
BTN2A2	ENST00000494184					
BTN2A2	ENST00000482636					
BTN2A2	ENST00000469230					
BTN2A2	ENST00000416795					
BTN2A2	ENST00000432533					
BTN2A2	ENST00000493275					
ABT1	ENST00000274849	ILMN_1658083	monocyte	4.18×10-5 (p-value)	Fairfax et al.	7
ZSCAN26	?	ENSG00000197062	transformed fibroblasts	1.09×10-5 (p-value)	GTEx Portal V6	116
ZNF192P1	?	ENSG00000226314	transformed fibroblasts	3.66×10-5 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	transformed fibroblasts	2.25×10-6 (p-value)	GTEx Portal V6	33
ZSCAN26	?	ENSG00000197062	adrenal gland	6.33×10-7 (p-value)	GTEx Portal V6	92
HIST1H4J	?	ENSG00000197238	adrenal gland	1.43×10-5 (p-value)	GTEx Portal V6	1
AL022393.7	?	ENSG00000216901	muscularis mucosae	1.61×10-5 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	muscularis mucosae	2.02×10-7 (p-value)	GTEx Portal V6	39
LINC01556	?	ENSG00000204709	aorta	1.17×10-5 (p-value)	GTEx Portal V6	36
ZFP57	?	ENSG00000204644	aorta	8.39×10-8 (p-value)	GTEx Portal V6	39
ZSCAN23	?	ENSG00000187987	tibial artery	3.47×10-5 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	tibial artery	1.37×10-7 (p-value)	GTEx Portal V6	38
BTN3A2	ENST00000396934	ILMN_1676528	monocyte	2.75×10-52 (p-value)	Zeller et al.	3
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
?	?	ILMN_1820787	monocyte	1.60×10-31 (p-value)	Zeller et al.	3
?	?	ILMN_1883997	monocyte	1.36×10-32 (p-value)	Zeller et al.	3
BTN3A2	ENST00000524459	ILMN_1700067	monocyte	2.62×10-37 (p-value)	Zeller et al.	3
BTN3A2	ENST00000528541					

BTN3A3	ENST00000467524					
BTN3A3	ENST00000482451					
BTN3A3	ENST00000494393					
BTN3A2	ENST00000527417					
BTN3A3	ENST00000244519					
BTN3A2	ENST00000528222					
BTN3A3	ENST00000474790					
BTN3A2	ENST00000377708					
BTN3A3	ENST00000496719					
BTN3A2	ENST00000396948					
BTN3A2	ENST00000527422					
BTN3A2	ENST00000356386					
?	?	ILMN_1679389	monocyte	1.99×10-14 (p-value)	Zeller et al.	2
ZFP57	?	ENSG00000204644	pancreas	7.02×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	lung	3.76×10-8 (p-value)	GTEx Portal V6	40
ZFP57	?	ENSG00000204644	blood	9.66×10-10 (p-value)	GTEx Portal V6	40
HLA-F-AS1	?	ENSG00000214922	blood	3.22×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	breast	5.76×10-7 (p-value)	GTEx Portal V6	34
ZFP57	?	ENSG00000204644	unexposed skin	4.21×10-6 (p-value)	GTEx Portal V6	33
ZFP57	?	ENSG00000204644	stomach	5.19×10-7 (p-value)	GTEx Portal V6	34
ZFP57	?	ENSG00000204644	esophagus mucosa	6.01×10-13 (p-value)	GTEx Portal V6	40
ZSCAN26	?	ENSG00000197062	esophagus mucosa	9.75×10-6 (p-value)	GTEx Portal V6	29
ZFP57	?	ENSG00000204644	atrial appendage	3.72×10-7 (p-value)	GTEx Portal V6	33
ZFP57	?	ENSG00000204644	visceral adipocytes	7.12×10-6 (p-value)	GTEx Portal V6	27
ZFP57	?	ENSG00000204644	liver	3.48×10-6 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
BTN3A2	?	ENSG00000186470	chr6	blood	1.67×10-7 (q-value)	SeeQTL DB (HapMap)	15
BTN3A2	ENST00000524682	209846_s_at	chr6	blood	6.40×10-16 (p-value)	Dixon et al.	4
BTN3A2	ENST00000604202						
BTN3A2	ENST00000396934						
BTN3A2	ENST00000532294						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000377708						
BTN3A2	ENST00000527422						
BTN3A2	ENST00000531055						
BTN3A2	ENST00000396948						
BTN3A2	ENST00000356386						
BTN3A2	ENST00000396934	212613_at	chr6	blood	1.40×10-13 (p-value)	Dixon et al.	4
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000244519	204820_s_at	chr6	blood	3.00×10-15 (p-value)	Dixon et al.	4
BTN3A2	ENST00000396934						
BTN3A2	ENST00000508906						

BTN3A2	ENST00000356386						
BTN3A3	ENST00000480110						
?	?	ILMN_1679389	chr6	monocyte	1.50×10-12 (p-value)	Zeller et al.	1
?	?	ILMN_1671054	chr6	monocyte	3.41×10-37 (p-value)	Zeller et al.	3
?	?	ILMN_1666078	chr6	monocyte	4.02×10-28 (p-value)	Zeller et al.	3
?	?	ILMN_1752592	chr6	monocyte	5.77×10-19 (p-value)	Zeller et al.	3
?	?	ILMN_1715169	chr6	monocyte	3.28×10-12 (p-value)	Zeller et al.	1
HLA-G	ENST00000376828	ILMN_1656670	chr6	monocyte	1.65×10-14 (p-value)	Zeller et al.	2
HLA-G	ENST00000360323						
HLA-G	ENST00000478519						
HLA-G	ENST00000478355						
HLA-G	ENST00000428701						
HLA-G	ENST00000376815						
HLA-G	ENST00000376818						
HLA-C	ENST00000470363	ILMN_2150787	chr6	monocyte	5.78×10-14 (p-value)	Zeller et al.	1
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
E2F4	MA0541.1	3	no	0.000	1
MEF2A	MA0558.1	21	no	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000047721		
ENCP00000047738		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000443358	1	ENCP00000047719	HIST1H2BPS2
		ENCP00000047723	HIST1H4L
		ENCP00000047717	HIST1H4K
		ENCP00000047716	HIST1H4J
		ENCP00000047721	HIST1H1B
ENCE00000443289	1	ENCP00000047711	RSL24D1P1
ENCE00000443591	1	ENCP00000047762	GPX6 GPX6
		ENCP00000047757	ZKSCAN3
ENCE00000443338	1	ENCP00000047713	HIST1H2AI HIST1H2BL
ENCE00000443415	1	ENCP00000047727	OR2B2
ENCE00000443677	1	ENCP00000047794	OR2U1P OR2U1P

			OR2U1P OR2B4P OR2B4P OR2B4P OR2B4P OR2U1P OR2B4P OR2B4P OR2B4P OR2U1P
ENCE00000443536	1	ENCP00000047763	GPX5 GPX5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001702839 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K9ac, SP1, H4K5ac, H3K27me3, CTCF, PolII, Jund, TAF7, Nrsf, H3K27ac, TAF1, H3K4me2
		HSMMtube	H2AZ, H3K4me2, DNase1
		blood (K562)	TAF1, Cfos, PolII, GTF2B, H4K20me1, H3K4me2, H3K36me3, H3K4me3, Ini1, Brg1, DNase1, H3K27ac, Max, TAF7, BCLAF1, CTCF, MEF2A, H3K9ac, Cmyc, H3K79me2, HEY1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, DNase1
		liver (HepG2)	H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1, H3K4me2, H2AZ, PolII, TAF1, H3K4me1
		blood (GM12878)	POU2F2, Cmyc, PolII, BCLAF1, H2AZ, DNase1, Yy1, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	CTCF, H3K27ac, H3K9ac, PolII, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	Ini1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	Cjun, Max, H3K4me3, H3K4me2, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00001214443 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K36me3, H3K27me3, PolII, Jund, TAF7, p300, Nrsf, Sin3Ak20, H3K27ac, Yy1, RXRA, Tcf12, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3
		HSMMtube	DNase1, H2AZ
		blood (K562)	H3K4me2, H3K36me3, DNase1, H3K4me3, Ini1, Brg1, H3K27ac, Max, BCLAF1, H3K9ac, H3K79me2, HEY1, TAF1, PolII
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		liver (HepG2)	DNase1, H3K27me3, H3K4me3, Gabp, Jund, PolII, TAF1, ELF1, p300, ZBTB33, H3K4me2, H3K9ac, H3K27ac
		lung (IMR90)	H3K36me3, H3K4me3, H3K9ac, H3K4me2, DNase1
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, ELF1, POU2F2, TAF1, PolII, BCLAF1, Tcf12, DNase1, Yy1
		nervous (NH-A)	H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, H3K4me1
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K36me3, DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII
		monocytes (Monocytes-CD14+)	H3K4me2, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K4me3, H3K4me2, H3K9ac, PolII, DNase1, Cmyc
		A549	H3K27ac, DNase1, H3K9ac, H3K4me3
ENSR00001214446 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K9ac, H4K5ac, H3K27me3, CTCF, PolII, Jund, TAF7, H3K27ac, Yy1, TAF1, H3K4me2, SP1
		HSMMtube	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		blood (K562)	Cfos, Cmyc, PolII, H3K4me2, H3K36me3, H3K4me3, Cjun, TAF1, Ini1, Brg1, DNase1, Egr1, H3K27ac, Max, SP1, H3K9ac, H3K79me2, HEY1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	PolII, H3K79me2, DNase1, TAF1, H3K4me1, H3K4me2, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3
		blood (GM12878)	PolII, DNase1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	H3K9ac, DNase1, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac

		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K27ac, H3K9ac, PolII, H3K4me2, H3K4me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K4me3, Gabp, Cfos, PolII, Cmyc, H3K36me3, Max, TAF1, Ini1, H3K9ac, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K27ac, PolII, DNase1, Cmyc, H3K9ac, Cjun, Max, H3K4me3, H3K4me2
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001494372 (enhancer)	1	endothelium (HUVEC)	H3K27me3
		blood (K562)	PolII, H3K36me3
ENSR00001702845 (CTCF binding site)	1	endothelium (HUVEC)	H3K27me3
		blood (K562)	PolII, H3K36me3
ENSR00001214452 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, SP1, H3K9ac, H3K4me3, H4K5ac, H3K36me3, CTCF, PolII, Rad21, Egr1, Jund, TAF7, Sin3Ak20, H3K27ac, Yy1, BCL11A, TAF1, H3K4me2
		HSMMtube	DNase1, H2AZ, H3K4me2
		blood (K562)	H3K9ac, HEY1, TAF1, Jund, H3K4me3, PolII, GTF2B, H3K4me2, BCLAF1, TAF7, Ini1, Brg1, H3K27ac
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H3K9ac, DNase1, H2AZ, H3K4me3, H3K27ac, H3K4me2
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K9ac, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4me2, DNase1, H3K27ac
		blood (GM12878)	POU2F2, Cmyc, PU1, PolII, BCLAF1, H2AZ, DNase1, Yy1, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac, Cjun
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K27ac, H3K9ac, PolII
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me2, H3K9ac, DNase1, H3K27ac, H3K4me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K9ac, PolII, DNase1, H3K4me2, H3K4me3, Cjun
		A549	H3K4me3, DNase1
ENSR00001214455 (promoter)	1	embryonic stem cell (H1ESC)	H3K9ac, H3K4me3, SP1, H3K4me2, H4K5ac, H3K36me3, H3K27me3, CTCF, PolII, Jund, TAF7, p300, H3K27ac, Yy1, Tcf12, TAF1
		HSMMtube	DNase1, H2AZ, H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		blood (GM12878)	H3K9ac, POU2F2, PolII, BATF, H2AZ, DNase1, ELF1, H3K4me3, H3K27ac, Cfos, H3K4me2, CTCF
		lung (IMR90)	H3K4me2, H4K5ac, H3K4me3, H3K4ac, H2BK120ac, H4K91ac, H3K56ac, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	CTCF, DNase1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K9ac, H3K27ac, H3K4me3, DNase1
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2, CTCF
		monocytes (Monocytes-CD14+)	H3K4me2, H3K27ac, H3K9ac, H3K4me3, DNase1
		endothelium (HUVEC)	Max, DNase1, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, Cmyc
		A549	DNase1, H3K9ac, H3K4me3, H3K27ac
ENSR00001214477 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Gabp, TAF7, CTCF, Yy1, TAF1, H3K4me2, H3K4me3
		HSMMtube	DNase1, H2AZ, H3K4me2
		blood (K562)	H3K27ac, Max, H3K9ac, Gabp, HEY1, H2AZ, TAF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H3K4me3, DNase1, CTCF, H3K9ac, Gabp, PolII, TAF1, H2AZ, H3K4me2
		blood (GM12878)	TAF1, PolII, BCLAF1, H2AZ, DNase1, Gabp, ELF1, H3K4me3, H3K27ac, CTCF, H3K9ac
		lung (IMR90)	H4K5ac, H3K4me3, CTCF, H3K4me2
		nervous (NH-A)	H3K4me3, H3K4me2, H3K9ac, DNase1
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ

		breast (HMEC)	H3K4me2, CTCF, H3K4me3
		cervix (HeLa-S3)	H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF, DNase1, H3K9ac, Ini1
		monocytes (Monocytes-CD14+)	H3K4me3, H3K9ac, CTCF
		endothelium (HUVEC)	H3K4me3, H3K4me2, PolII, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001214498 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, TAF7, Sin3Ak20, H3K9ac, Yy1, TAF1, H3K4me2, H3K4me3, Gabp, Egr1, H3K36me3, H3K27me3, CTCF, PolII, Rad21
		HSMMtube	DNase1, H3K36me3, H3K4me3, H3K4me2
		blood (K562)	Yy1, Egr1, H3K27ac, Max, SP2, H3K9ac, Gabp, Cmyc, H3K79me2, H2AZ, TAF1, ELF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, Gabp, Yy1, PolII, TAF1, ELF1, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K36me3, H3K27me3
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3, H3K9ac, DNase1
		blood (GM12878)	TAF1, PolII, BCLAF1, H2AZ, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K9ac
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K36me3
		Osteobl	H2AZ, H3K4me3, H3K36me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, TAF1, PolII, H3K4me3, Gabp, H3K36me3, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K9ac, PolII, DNase1, Cmyc, H3K4me3, H3K4me2, Max
		A549	DNase1, H3K4me3
ENSR00001214514 (enhancer)	1	liver (HepG2)	H3K4me1, H3K36me3
ENSR00001494388 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, SP1, Srf, H3K9ac, H3K4me3, PolII, TAF7
		HSMMtube	H3K4me3, H3K27ac, H3K4me2, H3K9ac, H2AZ, DNase1
		blood (K562)	Ini1, Brg1, Gata2, Egr1, H3K4me3, H3K27ac, PU1, ZBTB7A, H3K9ac, Gabp, HEY1, H2AZ, TAF1, Nrsf, Cjun, Cfos, PolII, H3K4me2, H3K4me1, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H2AZ, H3K4me3, H3K27ac
		liver (HepG2)	FOSL2, Gabp, Jund, PolII, TAF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K9ac, H4K5ac, H3K4me2, H3K27ac, DNase1, H3K18ac
		blood (GM12878)	PolII, Srf, H2AZ, DNase1, Yy1, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Gabp, PolII, TAF1, H3K4me3, H3K9ac, Jund, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cjun, H3K4me3, H3K4me2, H3K9ac, PolII, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
ENSR00001214525 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	DNase1
		A549	H3K27me3
ENSR00001214530 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, PolII, Rad21, Sin3Ak20, CTCF, H3K27ac, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K9ac, CTCF, H3K4me2, DNase1, H3K36me3, H2AZ, H3K27ac
		blood (K562)	TAF1, ELF1, PolII, H3K4me2, CTCF, H3K4me1, H3K36me3, DNase1, H3K4me3, Rad21, H2AZ, H3K27me3, Ini1, Brg1, H3K27ac, Max, SP2, Nfe2, H3K9ac, Gabp, H3K79me2, HEY1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	CTCF, H3K4me3, H3K27me3, DNase1, H3K27ac, H3K9ac, Gabp, PolII, Rad21, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2
		blood (GM12878)	CTCF, H3K9ac, H3K4me2, H3K27ac, DNase1, PolII, H2AZ, Yy1, Gabp, ELF1, Rad21, H3K79me2, H3K4me3
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3, H3K9ac, CTCF
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF
		NHLF	DNase1, H3K4me3, CTCF, H3K27ac

		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, CTCF
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, H3K4me3, Gabp, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, CTCF, PolII, H3K4me3, H3K4me2, Cjun, DNase1
		A549	H3K36me3, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00001214540 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, Egr1, Gabp, TAF7, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, PolII, H3K27me3, H3K36me3
		HSMMtube	CTCF, DNase1, H3K36me3, H2AZ, H3K4me3, H3K4me2, H3K9ac
		blood (K562)	Ini1, Brg1, Egr1, H3K27ac, Max, SP2, BCLAF1, H3K4me3, DNase1, ZBTB7A, H3K9ac, Gabp, ETS1, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, ELF1, CTCFL, PolII, H3K4me2, CTCF, H3K36me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K36me3
		liver (HepG2)	DNase1, H3K27me3, H3K36me3, H3K4me3, Cmyc, Gabp, PolII, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF
		blood (GM12878)	ELF1, H3K79me2, H3K4me3, H3K27ac, CTCF, H3K9ac, H3K36me3, Gabp, Yy1, TAF1, PolII, BCLAF1, Srf, H2AZ, ETS1, DNase1
		lung (IMR90)	H4K8ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac, CTCF, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K36me3, H3K4me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, H3K36me3
		NHLF	H3K36me3, DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me3, H3K36me3, H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac
		breast (HMEC)	CTCF, H3K4me2, H3K4me3, H3K9ac
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K9ac, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, H3K9ac, PolII, CTCF, H3K27me3, H3K36me3
		A549	H3K36me3, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001494403 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K27me3, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me2, DNase1, H3K27me3, H2AZ, H3K4me3
		blood (K562)	PolII, Ini1, DNase1, PolIII, H3K27ac, Max, H3K9ac, Cmyc, H2AZ, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me2, DNase1
		liver (HepG2)	H2AZ, H3K4me2, H3K4me3, H3K27me3
		blood (GM12878)	H3K4me2, H3K4me3, H2AZ
		lung (IMR90)	H3K4me3, H3K4me2
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K4me2, H3K4me3, H3K27me3
		NHLF	H3K4me3
		Osteobl	H3K27me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K27me3, H3K4me3, H3K4me2
		breast (HMEC)	H3K4me2, H3K4me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3, H3K4me2, H3K4me3
		A549	H3K9ac, H3K4me2, H3K4me3
ENSR00000487476 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K27me3, Egr1, H3K4me2, H3K4me3
		HSMMtube	H3K27me3, DNase1
		blood (K562)	PolII, Ini1, DNase1, PolIII, Egr1, Max, H2AZ, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me3, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me3, H3K4me2, DNase1
		skin (NHEK)	DNase1, H3K4me2
ENSR00001702885 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K27me3, Egr1, H3K4me3, H3K4me2
		HSMMtube	H3K27me3, DNase1

		blood (K562)	PolII, Ini1, DNase1, PolIII, Egr1, Max, H2AZ, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me3, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	DNase1, H3K4me2, H3K4me3
		skin (NHEK)	H3K4me2, DNase1
ENSR00000264184 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K27me3, H3K9ac, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	Ini1, Brg1, DNase1, H3K27ac, Max, H3K9ac, H2AZ, PolII, Cmyc, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me3
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, H3K4me3, Max, Cmyc, H3K9ac, PolII
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K4me1, H2AZ, H3K4me2, H3K4me3, H3K27me3, DNase1
		A549	DNase1, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, H3K4me2, H3K4me3
ENSR00001702887 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3, H3K36me3
		blood (K562)	H3K4me2, H2AZ
ENSR00000487478 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K4me2, H3K27me3, H3K4me3, DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		skin (NHEK)	H3K27me3
		muscle (HSMM)	H3K27me3
		breast (HMEC)	H3K27me3
ENSR00000264195 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K4me2, H3K27me3, H3K4me3, DNase1
		HSMMtube	H3K27me3
		blood (K562)	PolII, Ini1, Brg1, DNase1, PolIII, H3K27ac, Max, H3K9ac, H2AZ, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3
		muscle (HSMM)	DNase1
		liver (HepG2)	H3K4me3, H3K4me2, H3K27me3
		blood (GM12878)	H3K4me3
		lung (IMR90)	H3K4me3
		nervous (NH-A)	H3K27me3, H3K4me2
		skin (NHEK)	DNase1, H3K4me3, H3K27me3
		Osteobl	H3K27me3, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me2
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K4me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		A549	DNase1, H3K4me2, H3K4me3
ENSR00001494411 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, H3K4me2
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27me3
		blood (K562)	PU1, MEF2A, H3K4me2, H3K4me3, Max, PolIII, PolII, Ini1, DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, H3K4me2
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me2
		nervous (NH-A)	H3K27me3
		A549	DNase1, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL009179.1	downstream gene variant	1163	ENST00000625569	?	?	1
AL022393.7	upstream gene variant, downstream gene variant	380	ENST00000405613	?	?	2

	gene variant					
AL022393.9	upstream gene variant, downstream gene variant	1124	ENST00000625142	?	?	3
AL049822.1	downstream gene variant	1626	ENST00000631369	?	?	1
AL121932.1	upstream gene variant	4597	ENST00000577570	?	?	1
COX11P1	upstream gene variant	2809	ENST00000489116	?	?	1
HIST1H1B	downstream gene variant, upstream gene variant	488	ENST00000331442	NM_005322.2	ENSP00000330074	3
HIST1H2AI	upstream gene variant	949	ENST00000358739	NM_003509.2	ENSP00000351589	2
HIST1H2AJ	upstream gene variant	1423	ENST00000333151	NM_021066.2	ENSP00000328484	1
HIST1H2AK	downstream gene variant, upstream gene variant	470	ENST00000618958	NM_003510.2	ENSP00000482431	3
HIST1H2AL	downstream gene variant	560	ENST00000613174	NM_003511.2	ENSP00000482538	4
HIST1H2AM	upstream gene variant	4975	ENST00000359611	NM_003514.2	ENSP00000352627	1
HIST1H2BL	downstream gene variant	194	ENST00000377401	NM_003519.3	ENSP00000366618	2
HIST1H2BM	downstream gene variant	739	ENST00000621112	NM_003521.2	ENSP00000477907	1
HIST1H2BN	upstream gene variant	1068	ENST00000449538	?	ENSP00000446031	1
HIST1H2BN	upstream gene variant, downstream gene variant	509	ENST00000396980	?	ENSP00000380177	5
HIST1H2BN	upstream gene variant	1124	ENST00000606613	?	ENSP00000475942	1
HIST1H2BN	upstream gene variant, downstream gene variant	283	ENST00000612898	NM_003520.3	ENSP00000483903	2
HIST1H2BO	downstream gene variant	4281	ENST00000616182	NM_003527.4	ENSP00000477527	1
HIST1H2BPS2	upstream gene variant	1906	ENST00000402707	?	?	3
HIST1H3H	upstream gene variant, downstream gene variant	2814	ENST00000369163	NM_003536.2	ENSP00000358160	2
HIST1H3I	downstream gene variant, upstream gene variant	833	ENST00000616365	NM_003533.2	ENSP00000483283	3
HIST1H4J	downstream gene variant	1734	ENST00000355057	NM_021968.3	ENSP00000347168	1
HIST1H4K	downstream gene variant	107	ENST00000611927	NM_003541.2	ENSP00000479794	1
HIST1H4L	downstream gene variant	51	ENST00000618305	NM_003546.2	ENSP00000480960	2
HIST1H4PS1	upstream gene variant	3747	ENST00000406085	?	?	1
LINC00533	upstream gene variant	2510	ENST00000456103	?	?	1
NKAPL	upstream gene variant	1787	ENST00000343684	NM_001007531.2	ENSP00000345716	1
NOP56P1	downstream gene variant, upstream gene variant	3129	ENST00000440030	?	?	3
OR1F12	upstream gene variant	2165	ENST00000402365	?	?	1
OR2B2	upstream gene variant	3838	ENST00000303324	NM_033057.2	ENSP00000304419	1
OR2B7P	downstream gene variant	2105	ENST00000429302	?	?	1
OR2B8P	downstream gene variant	3756	ENST00000432841	?	?	1
OR2W2P	downstream gene variant	595	ENST00000406313	?	?	1
OR2W4P	upstream gene variant	1559	ENST00000406496	?	?	1
PGBD1	downstream gene variant	1647	ENST00000259883	NM_001184743.1, NM_032507.3	ENSP00000259883	1
RNU7-26P	upstream gene variant	558	ENST00000458980	?	?	2
RP1-265C24.5	upstream gene variant	1648	ENST00000405929	?	?	4
RP5-874C20.6	downstream gene variant, upstream	1452	ENST00000605757	?	?	3

	gene variant					
RP5-874C20.7	downstream gene variant, upstream gene variant	1496	ENST00000615674	?	?	4
RP5-874C20.8	downstream gene variant	822	ENST00000621053	?	ENSP00000481142	2
RPLP2P1	upstream gene variant	652	ENST00000398220	?	?	1
RPSAP2	upstream gene variant, downstream gene variant	811	ENST00000416785	?	?	3
TOB2P1	upstream gene variant	1220	ENST00000469761	?	?	2
Z98744.1	upstream gene variant, downstream gene variant	480	ENST00000629628	?	?	3
Z98744.2	upstream gene variant	569	ENST00000627472	?	?	4
ZKSCAN3	upstream gene variant	2847	ENST00000377255	NM_001242894.1	ENSP00000366465	1
ZKSCAN3	upstream gene variant	2847	ENST00000341464	NM_001242895.1	ENSP00000341883	1
ZKSCAN3	upstream gene variant	2811	ENST00000252211	NM_024493.3	ENSP00000252211	1
ZKSCAN8	downstream gene variant	3257	ENST00000536028	?	ENSP00000439117	1
ZKSCAN8	downstream gene variant	2263	ENST00000457389	NM_001278119.1	ENSP00000402948	1
ZKSCAN8	downstream gene variant	1981	ENST00000330236	NM_001278122.1, NM_006298.3, NM_001278121.1	ENSP00000332750	4
ZKSCAN8	downstream gene variant	2225	ENST00000606198	?	ENSP00000475589	1
ZNF165	downstream gene variant	1608	ENST00000377325	NM_003447.3	ENSP00000366542	2
ZNF192P1	upstream gene variant, downstream gene variant	116	ENST00000565888	?	?	6
ZNF192P1	upstream gene variant, downstream gene variant	312	ENST00000562227	?	?	5
ZNF192P1	upstream gene variant, downstream gene variant	127	ENST00000440790	?	?	4
ZNF192P1	upstream gene variant, downstream gene variant	338	ENST00000570126	?	?	4
ZNF192P1	upstream gene variant, downstream gene variant	107	ENST00000570042	?	?	4
ZNF192P2	upstream gene variant	4950	ENST00000402215	?	?	1
ZSCAN12	downstream gene variant, upstream gene variant	1000	ENST00000396827	?	ENSP00000380039	2
ZSCAN12	downstream gene variant, upstream gene variant	997	ENST00000361028	?	ENSP00000354305	2
ZSCAN12P1	downstream gene variant	4359	ENST00000529104	?	?	1
ZSCAN12P1	upstream gene variant	1256	ENST00000406489	?	?	1
ZSCAN16	upstream gene variant	758	ENST00000340487	NM_025231.1	ENSP00000366527	2
ZSCAN16-AS1	downstream gene variant	95	ENST00000602810	?	?	1
ZSCAN16-AS1	downstream gene variant	92	ENST00000600652	?	?	1
ZSCAN23	upstream gene variant	662	ENST00000486481	?	?	1
ZSCAN23	upstream gene variant	684	ENST00000481983	?	ENSP00000435430	1
ZSCAN23	upstream gene variant	697	ENST00000289788	NM_001012455.1	ENSP00000289788	1
ZSCAN26	downstream gene variant	4201	ENST00000316606	NM_152736.5	ENSP00000484931	2
ZSCAN26	downstream gene variant	4178	ENST00000619937	?	ENSP00000484018	2
ZSCAN26	downstream gene variant	4181	ENST00000614088	NM_001287421.1	ENSP00000479314	2
ZSCAN26	downstream gene variant	4872	ENST00000623276	NM_001111039.2	ENSP00000485228	1
ZSCAN26	downstream gene variant	4181	ENST00000421553	NM_001023560.3	ENSP00000481707	2

ZSCAN31	downstream gene variant, upstream gene variant	473	ENST00000414431 ?	ENSP00000407529	5
ZSCAN31	upstream gene variant	1721	ENST00000439628 ?	ENSP00000392552	2
ZSCAN31	downstream gene variant, upstream gene variant	671	ENST00000447021 ?	ENSP00000416108	5
ZSCAN31	downstream gene variant, upstream gene variant	293	ENST00000476001 ?	?	3
ZSCAN31	downstream gene variant, upstream gene variant	442	ENST00000344279 NM_030899.4, NM_001243241.1	ENSP00000345339	5
ZSCAN31	upstream gene variant	1721	ENST00000434036 ?	ENSP00000416225	2
ZSCAN31	downstream gene variant, upstream gene variant	330	ENST00000414429 ?	ENSP00000390076	3
ZSCAN31	downstream gene variant, upstream gene variant	232	ENST00000435857 ?	ENSP00000391235	5
ZSCAN31	downstream gene variant	1823	ENST00000481934 ?	?	2
ZSCAN31	downstream gene variant, upstream gene variant	232	ENST00000611469 NM_001243244.1, NM_001243242.1	ENSP00000480254	5
ZSCAN31	downstream gene variant, upstream gene variant	473	ENST00000446474 NM_001243243.1	ENSP00000402937	5
ZSCAN31	downstream gene variant, upstream gene variant	1730	ENST00000396838 ?	ENSP00000380050	4
ZSCAN31	downstream gene variant, upstream gene variant	1721	ENST00000426434 ?	ENSP00000398680	4
ZSCAN31	upstream gene variant	1860	ENST00000444081 ?	ENSP00000415040	2
ZSCAN31	downstream gene variant, upstream gene variant	1156	ENST00000439636 ?	ENSP00000412519	5
ZSCAN31	upstream gene variant	1739	ENST00000446222 ?	ENSP00000411033	2
ZSCAN31	downstream gene variant, upstream gene variant	473	ENST00000439158 NM_001135216.1	ENSP00000413705	5
ZSCAN31	downstream gene variant, upstream gene variant	409	ENST00000453745 ?	ENSP00000389479	5
ZSCAN9	upstream gene variant, downstream gene variant	738	ENST00000527844 ?	ENSP00000436166	4
ZSCAN9	upstream gene variant, downstream gene variant	1167	ENST00000526391 ?	ENSP00000476254	4
ZSCAN9	upstream gene variant, downstream gene variant	271	ENST00000531979 NM_001199480.1	ENSP00000433402	3
ZSCAN9	upstream gene variant, downstream gene variant	15	ENST00000527436 ?	ENSP00000433468	3
ZSCAN9	upstream gene variant, downstream gene variant	287	ENST00000252207 NM_006299.4	ENSP00000252207	3
ZSCAN9	upstream gene variant	1376	ENST00000531941 ?	ENSP00000433307	1
ZSCAN9	upstream gene variant	1754	ENST00000531981 ?	ENSP00000475719	1
ZSCAN9	upstream gene variant, downstream gene variant	287	ENST00000425468 NM_001199479.1	ENSP00000404074	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
HIST1H1B	ENST00000331442	NM_005322.2	ENSP00000330074	G	ggT/ggC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPX6	ENST00000612264	?	ENSP00000483462	1
GPX6	ENST00000361902	NM_182701.1	ENSP00000354581	1
GPX6	ENST00000474923	?	ENSP00000417364	1
GPX6	ENST00000483058	?	?	1
HIST1H2BN	ENST00000606613	?	ENSP00000475942	5

HIST1H2BN	ENST00000449538	?	ENSP00000446031	5
HIST1H2BN	ENST00000396980	?	ENSP00000380177	5
PGBD1	ENST00000259883	NM_001184743.1, NM_032507.3	ENSP00000259883	6
RP5-874C20.8	ENST00000621053	?	ENSP00000481142	1
ZBED9	ENST00000530247	?	?	1
ZKSCAN3	ENST00000377255	NM_001242894.1	ENSP00000366465	4
ZKSCAN3	ENST00000252211	NM_024493.3	ENSP00000252211	4
ZKSCAN3	ENST00000341464	NM_001242895.1	ENSP00000341883	4
ZKSCAN8	ENST00000536028	?	ENSP00000439117	2
ZKSCAN8	ENST00000330236	NM_001278122.1, NM_006298.3, NM_001278121.1	ENSP00000332750	2
ZKSCAN8	ENST00000457389	NM_001278119.1	ENSP00000402948	2
ZKSCAN8	ENST00000606198	?	ENSP00000475589	2
ZNF192P1	ENST00000562227	?	?	2
ZNF192P1	ENST00000570126	?	?	1
ZNF192P1	ENST00000440790	?	?	3
ZNF192P1	ENST00000570042	?	?	2
ZSCAN12	ENST00000396827	?	ENSP00000380039	8
ZSCAN12	ENST00000361028	?	ENSP00000354305	8
ZSCAN16-AS1	ENST00000600652	?	?	1
ZSCAN16-AS1	ENST00000602810	?	?	1
ZSCAN26	ENST00000617168	?	ENSP00000481024	1
ZSCAN26	ENST00000316606	NM_152736.5	ENSP00000484931	1
ZSCAN26	ENST00000614088	NM_001287421.1	ENSP00000479314	1
ZSCAN26	ENST00000421553	NM_001023560.3	ENSP00000481707	1
ZSCAN26	ENST00000611552	NM_001287422.1	ENSP00000478769	1
ZSCAN26	ENST00000619937	?	ENSP00000484018	1
ZSCAN26	ENST00000623276	NM_001111039.2	ENSP00000485228	1
ZSCAN31	ENST00000426756	?	ENSP00000406376	3
ZSCAN31	ENST00000446222	?	ENSP00000411033	5
ZSCAN31	ENST00000414429	?	ENSP00000390076	6
ZSCAN31	ENST00000396838	?	ENSP00000380050	5
ZSCAN31	ENST00000426434	?	ENSP00000398680	5
ZSCAN31	ENST00000439628	?	ENSP00000392552	5
ZSCAN31	ENST00000476001	?	?	4
ZSCAN31	ENST00000434036	?	ENSP00000416225	5
ZSCAN31	ENST00000444081	?	ENSP00000415040	5
ZSCAN9	ENST00000531941	?	ENSP00000433307	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
HIST1H2BN	ENST00000612898	NM_003520.3	ENSP00000483903	1
ZKSCAN8	ENST00000330236	NM_001278122.1, NM_006298.3, NM_001278121.1	ENSP00000332750	1

ZSCAN12	ENST00000361028	?	ENSP00000354305	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ZSCAN9	ENST00000531979	NM_001199480.1	ENSP00000433402	1
ZSCAN9	ENST00000425468	NM_001199479.1	ENSP00000404074	1
ZSCAN9	ENST00000531981	?	ENSP00000475719	1
ZSCAN9	ENST00000252207	NM_006299.4	ENSP00000252207	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AL022393.9	ENST00000625142	?	1
HIST1H4PS1	ENST00000406085	?	1
OR2W4P	ENST00000406496	?	1
OR2W6P	ENST00000439689	?	1
ZNF192P1	ENST00000562227	?	1
ZNF192P1	ENST00000570126	?	1
ZNF192P1	ENST00000440790	?	1
ZNF192P1	ENST00000570042	?	1
ZSCAN12P1	ENST00000406489	?	1
ZSCAN12P1	ENST00000529104	?	2

Locus 254 (rs35070799)

SNiPAcard

Block annotations

Block info

genomic range	chr14:55,569,578-55,882,548
block size	312,971 bp
variant count	198 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.303 [-4.21 – 5.14]	gene(s) hit or close-by	AL158801.1, ATG14, CHMP4BP1, DLGAP5, FBXO34, LGALS3, Metazoa_SRP, RP11-454L9.2, RP11-665C16.1, RP11-665C16.5, RP11-665C16.6, RP11-665C16.8, RP11-698F20.3, RPL21P6, TBPL2
phastCons	μ = 0.090 [0 – 1]	eQTL gene(s)	FBXO34, KTN1, RP11-454L9.2, RP11-665C16.6, SOCS4
GERP++	μ = -0.493 [-10.3 – 5.57]	potentially regulated gene(s)	LGALS3, RPL13AP3
CADD score	μ = 3.476 [0.008 – 13.35]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FBXO34	missense variant	ENST00000440021	NM_152231.1	ENSP00000394117	A/V	gCt/gTt	?	?	1
FBXO34	missense variant	ENST00000555280	?	ENSP00000450877	A/V	gCt/gTt	?	?	1
FBXO34	missense variant	ENST00000313833	NM_017943.3	ENSP00000313159	A/V	gCt/gTt	?	?	1
LGALS3	missense variant	ENST00000254301	NM_002306.3	ENSP00000254301	H/Q	caT/caA	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-665C16.6	?	ENSG00000258413	testis	7.45×10^{-8} (p-value)	GTEx Portal V6	174
RP11-454L9.2	?	ENSG00000259318	testis	3.31×10^{-7} (p-value)	GTEx Portal V6	181
RP11-454L9.2	?	ENSG00000259318	tibial artery	6.32×10^{-6} (p-value)	GTEx Portal V6	59
RP11-454L9.2	?	ENSG00000259318	thyroid	1.20×10^{-6} (p-value)	GTEx Portal V6	164
SOCS4	ENST00000555846	ILMN_1753795	blood	1.38×10^{-5} (p-value)	MuTHER consortium	9
FBXO34	ENST00000440021	ILMN_1765060	blood	2.29×10^{-6} (p-value)	MuTHER consortium	14
FBXO34	ENST00000313833					
FBXO34	ENST00000440021	ILMN_1765060	b-cell	4.58×10^{-7} (p-value)	Fairfax et al.	2
FBXO34	ENST00000313833					
RP11-454L9.2	?	ENSG00000259318	subcutaneous adipocytes	2.02×10^{-6} (p-value)	GTEx Portal V6	103
KTN1	ENST00000395308	ILMN_2339202	monocyte	2.29×10^{-13} (p-value)	Zeller et al.	2
KTN1	ENST00000555172					
KTN1	ENST00000438792					

KTN1	ENST00000395309					
KTN1	ENST00000413890					
KTN1	ENST00000554507					
KTN1	ENST00000555573					
KTN1	ENST00000395311					
KTN1	ENST00000334975					
KTN1	ENST00000554831					
KTN1	ENST00000554294					
KTN1	ENST00000459737					
KTN1	ENST00000395314					
KTN1	ENST00000553710					
KTN1	ENST00000556631					
SOCS4	ENST00000555846	ILMN_1753795	monocyte	6.28×10-13 (p-value)	Zeller et al.	2
FBXO34	?	ENSG00000178974	pancreas	1.06×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000173483	1	ENST00000556438, ENST00000254301	LGALS3

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000169576	1	ENCP00000019674	RPL13AP3
ENCE00000169577	1	ENCP00000019674	RPL13AP3

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000418188 (promoter flanking region)	1	embryonic stem cell (H1ESC)	SP1, TAF1, PolII, Jund, Yy1, DNase1
		HSMMtube	H3K4me1, H3K9ac, H3K4me2, H3K27ac, DNase1
		blood (K562)	H3K36me3, PolII, H2AZ, H3K9ac, Max, H3K27ac, DNase1
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, H3K4me1
		muscle (HSMM)	H3K4me2, H3K9ac, H3K36me3, DNase1, H3K27ac, H3K9me3, H3K4me1, H2AZ
		liver (HepG2)	DNase1, FOSL2, Jund, H3K4me3, PolII, H3K4me1, H3K4me2, H3K9ac, H3K27ac
		lung (IMR90)	H3K18ac, H3K4me2, H4K5ac, H3K4ac, H2BK120ac, H2BK20ac, H4K91ac, H3K27ac, H2BK12ac, DNase1
		blood (GM12878)	PolII, H2AZ, H3K4me1, Tcf12, DNase1, H3K27ac, H3K4me2, H3K9ac, H3K9me3, PU1, H3K4me3, EBF1, POU2F2
		nervous (NH-A)	DNase1, H3K27ac, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K4me2, H3K9ac, H3K27ac, H3K4me1
		NHLF	H3K27ac, DNase1
		Osteobl	H3K27ac
		breast (HMEC)	DNase1, H3K4me2, H3K9ac, H3K27ac
		cervix (HeLa-S3)	TAF1, Max, Cmyc, Cfos, PolII, DNase1, Cjun, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me1
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac
		endothelium (HUVEC)	Cjun, PolII, Max, H3K4me1, H3K27ac, DNase1, Cmyc
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000418190 (promoter flanking region)	2	NHLF	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	Egr1, Jund, DNase1
		HSMMtube	H2AZ, DNase1, H3K27ac, H3K4me2, H3K9ac
		Osteobl	H2AZ, H3K27ac, H3K4me2
		blood (K562)	DNase1, H3K4me1, H2AZ, Cmyc, Jund, H3K9ac, Max, H3K27ac, Egr1
		skin (NHDF-AD)	H3K4me1, H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3
		breast (HMEC)	H3K9ac, H3K27ac, DNase1
		muscle (HSMM)	H3K27ac, H3K9ac, DNase1
		cervix (HeLa-S3)	DNase1, Max, PolII, Cmyc, TAF1, H3K27ac, Cjun, H3K9ac, Jund

		monocytes (Monocytes-CD14+)	H3K27ac, DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K27ac, DNase1, Cmyc
		liver (HepG2)	DNase1, FOSL2, Jund, PolII, TAF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc
		lung (IMR90)	DNase1, H4K8ac, H2BK12ac, H3K4me1, H3K27ac, H3K18ac, H3K4me2, H4K5ac, H3K4ac, H2BK120ac, H2BK20ac, H3K9ac, H4K91ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K9ac, H3K27ac
ENSR00001622651 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27ac
		liver (HepG2)	H3K4me1
ENSR00001457478 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		HSMMtube	H3K27me3
		breast (HMEC)	DNase1
ENSR00000418197 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, CTCF, Rad21, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H2AZ, DNase1
		blood (K562)	DNase1, H3K27me3
		skin (NHDF-AD)	H3K9ac, DNase1, H3K4me3, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		liver (HepG2)	DNase1, H3K27me3, H3K4me3, H3K4me2
		blood (GM12878)	DNase1, PolII, H2AZ, H3K4me3, H3K4me2
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K36me3, H3K27ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K36me3, H3K27me3
		breast (HMEC)	H3K36me3, H3K4me2, H3K9ac, H3K4me3, H3K27ac
		cervix (HeLa-S3)	TAF1, Max, H3K4me3, H3K79me2, PolII, H3K36me3, DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me3, H4K20me1, H3K9ac, DNase1, H3K4me1, H3K4me2, H3K27ac
		endothelium (HUVEC)	H3K4me3, PolII, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K36me3
ENSR00001622659 (enhancer)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		Osteobl	H3K27me3
ENSR00001457486 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		blood (DND-41)	H3K27me3
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
ENSR00000240495 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K4me1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		A549	H3K36me3
		skin (NHEK)	H3K4me2, H3K27ac, H3K4me1, DNase1
ENSR00001622674 (enhancer)	1	Osteobl	H3K4me2
ENSR00001622675 (CTCF binding site)	1	Osteobl	H3K4me2
ENSR00001024215 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K36me3
		blood (GM12878)	H3K4me1, H3K4me2
		blood (DND-41)	H3K36me3
ENSR00000418231 (CTCF binding site)	2	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3

ENSR00001622678 (open chromatin region)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00000240499 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	DNase1, Gata2, PolII
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	PolII, Jund, H3K27ac, H3K4me1, Max, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1
		A549	H3K36me3
ENSR00000418234 (promoter flanking region)	4	NHLF	H3K36me3, DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	DNase1, H3K36me3
		A549	H3K36me3
		nervous (NH-A)	DNase1
ENSR00001457502 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Cjun, Jund
		endothelium (HUVEC)	H3K36me3, Cjun, PolII, Max, DNase1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ATG14	1	ENST00000247178 ENST00000558189	hsa-miR-107

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL158801.1	upstream gene variant, downstream gene variant	2360	ENST00000623123	?	ENSP00000485133	3
ATG14	downstream gene variant, upstream gene variant	498	ENST00000247178	NM_014924.4	ENSP00000247178	8
ATG14	downstream gene variant, upstream gene variant	284	ENST00000558189	?	?	15
CHMP4BP1	upstream gene variant, downstream gene variant	2	ENST00000556017	?	?	7
DLGAP5	downstream gene variant	1564	ENST00000554067	?	ENSP00000452168	4
DLGAP5	downstream gene variant	2257	ENST00000395425	NM_001146015.1	ENSP00000378815	3
DLGAP5	downstream gene variant, upstream gene variant	2188	ENST00000554007	?	?	4

DLGAP5	downstream gene variant	2002	ENST00000557645	?	ENSP00000451747	2
DLGAP5	downstream gene variant	2281	ENST00000247191	NM_014750.4	ENSP00000247191	3
FBXO34	upstream gene variant, downstream gene variant	1126	ENST00000555280	?	ENSP00000450877	6
FBXO34	upstream gene variant, downstream gene variant	276	ENST00000313833	NM_017943.3	ENSP00000313159	4
FBXO34	upstream gene variant, downstream gene variant	845	ENST00000555087	?	?	3
FBXO34	upstream gene variant, downstream gene variant	396	ENST00000554940	?	?	3
FBXO34	upstream gene variant, downstream gene variant	116	ENST00000557647	?	?	5
FBXO34	upstream gene variant, downstream gene variant	1127	ENST00000440021	NM_152231.1	ENSP00000394117	4
LGALS3	upstream gene variant, downstream gene variant	1400	ENST00000554715	?	ENSP00000451381	4
LGALS3	upstream gene variant, downstream gene variant	349	ENST00000556438	?	?	3
LGALS3	upstream gene variant, downstream gene variant	2443	ENST00000556322	?	?	4
LGALS3	upstream gene variant, downstream gene variant	3823	ENST00000556263	?	?	2
LGALS3	upstream gene variant, downstream gene variant	359	ENST00000254301	NM_002306.3	ENSP00000254301	3
LGALS3	upstream gene variant	4006	ENST00000553493	?	ENSP00000451526	1
LGALS3	upstream gene variant, downstream gene variant	2249	ENST00000553755	?	?	4
Metazoa_SRP	upstream gene variant, downstream gene variant	755	ENST00000612004	?	?	7
RP11-454L9.2	downstream gene variant, upstream gene variant	124	ENST00000559320	?	?	20
RP11-665C16.1	downstream gene variant, upstream gene variant	46	ENST00000488313	?	?	6
RP11-665C16.5	downstream gene variant, upstream gene variant	202	ENST00000555514	?	?	11
RP11-665C16.6	downstream gene variant, upstream gene variant	851	ENST00000556183	?	?	4
RP11-665C16.6	downstream gene variant, upstream gene variant	710	ENST00000554650	?	?	4
RP11-665C16.8	downstream gene variant, upstream gene variant	1008	ENST00000605294	?	?	5
RP11-698F20.3	upstream gene variant	1091	ENST00000555643	?	?	2
RPL21P6	downstream gene variant, upstream gene variant	234	ENST00000479171	?	?	6
TBPL2	downstream gene variant	510	ENST00000247219	NM_199047.2	ENSP00000247219	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
AL158801.1	ENST00000623123	?	ENSP00000485133	P	ccA/ccG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATG14	ENST00000558189	?	?	31
ATG14	ENST00000247178	NM_014924.4	ENSP00000247178	54
DLGAP5	ENST00000247191	NM_014750.4	ENSP00000247191	25
DLGAP5	ENST00000395425	NM_001146015.1	ENSP00000378815	25
DLGAP5	ENST00000554067	?	ENSP00000452168	8
DLGAP5	ENST00000557645	?	ENSP00000451747	11
FBXO34	ENST00000555280	?	ENSP00000450877	3
FBXO34	ENST00000557647	?	?	53

FBXO34	ENST00000440021	NM_152231.1	ENSP00000394117	52
FBXO34	ENST00000313833	NM_017943.3	ENSP00000313159	52
FBXO34	ENST00000554940	?	?	52
FBXO34	ENST00000555087	?	?	52
LGALS3	ENST00000553493	?	ENSP00000451526	4
LGALS3	ENST00000254301	NM_002306.3	ENSP00000254301	3
LGALS3	ENST00000554715	?	ENSP00000451381	3
LGALS3	ENST00000553755	?	?	3
RP11-665C16.5	ENST00000555514	?	?	7
RP11-665C16.6	ENST00000556183	?	?	1
RP11-665C16.6	ENST00000554650	?	?	1
RP11-698F20.3	ENST00000555643	?	?	3
TBPL2	ENST00000247219	NM_199047.2	ENSP00000247219	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATG14	ENST00000247178	NM_014924.4	ENSP00000247178	2
FBXO34	ENST00000555280	?	ENSP00000450877	2
TBPL2	ENST00000247219	NM_199047.2	ENSP00000247219	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ATG14	ENST00000558189	?	2
CHMP4BP1	ENST00000556017	?	1
LGALS3	ENST00000556438	?	1
RP11-665C16.8	ENST00000605294	?	2

Locus 255 (rs12475664)

SNiPAcard

Block annotations

Block info

genomic range	chr2:309,097-309,097
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.126	gene(s) hit or close-by	AC079779.5, RP11-356M6.1
phastCons	0.02	eQTL gene(s)	SH3YL1
GERP++	-0.45	potentially regulated gene(s)	–
CADD score	1.239	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SH3YL1	?	ENSG00000035115	muscularis mucosae	9.42×10-6 (p-value)	GTEx Portal V6	1
SH3YL1	?	ENSG00000035115	tibial nerve	4.66×10-7 (p-value)	GTEx Portal V6	1
SH3YL1	?	ENSG00000035115	esophagus mucosa	8.54×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000584029 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC079779.5	downstream gene variant	23	ENST00000591927	?	?	1
AC079779.5	downstream gene variant	1199	ENST00000586715	?	?	1
AC079779.5	upstream gene variant	2260	ENST00000591158	?	?	1
AC079779.5	upstream gene variant	2501	ENST00000590255	?	?	1
AC079779.5	upstream gene variant	830	ENST00000587073	?	?	1
AC079779.5	upstream gene variant	1314	ENST00000588334	?	?	1
AC079779.5	upstream gene variant	2575	ENST00000585703	?	?	1
AC079779.5	upstream gene variant	1944	ENST00000591178	?	?	1
AC079779.5	downstream gene variant	124	ENST00000589936	?	?	1
AC079779.5	downstream gene variant	121	ENST00000591244	?	?	1
AC079779.5	upstream gene variant	1940	ENST00000457938	?	?	1
AC079779.5	upstream gene variant	1917	ENST00000588842	?	?	1
AC079779.5	upstream gene variant	2606	ENST00000592090	?	?	1

Putative effect on transcript

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC079779.5	ENST00000586772	?	1
AC079779.5	ENST00000589588	?	1
AC079779.5	ENST00000586311	?	1
AC079779.5	ENST00000586235	?	1
AC079779.5	ENST00000592366	?	1
AC079779.5	ENST00000585783	?	1
AC079779.5	ENST00000591122	?	1
AC079779.5	ENST00000592687	?	1
AC079779.5	ENST00000586673	?	1
AC079779.5	ENST00000590678	?	1
AC079779.5	ENST00000587796	?	1
AC079779.5	ENST00000416685	?	1
AC079779.5	ENST00000612487	?	1
AC079779.5	ENST00000592553	?	1
AC079779.5	ENST00000609984	?	1
AC079779.5	ENST00000591015	?	1
AC079779.5	ENST00000589124	?	1
RP11-356M6.1	ENST00000619113	?	1

Locus 256 (rs805273)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,665,452-31,665,452
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.647	gene(s) hit or close-by	ABHD16A, MIR4646, XXbac-BPG32J3.20
phastCons	0.001	eQTL gene(s)	LY6G5C, OR10C1, OR10C1, XXbac-BPG181B23.7
GERP++	0.848	potentially regulated gene(s)	–
CADD score	7.531	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG181B23.7	?	ENSG00000272221	skeletal muscle	1.14×10^{-6} (p-value)	GTEx Portal V6	1
LY6G5C	?	ENSG00000204428	sun exposed skin	3.75×10^{-6} (p-value)	GTEx Portal V6	1
OR10C1	ENST00000623086	ILMN_1790706	monocyte	1.01×10^{-5} (p-value)	Fairfax et al.	1
OR10C1	ENST00000444197					
OR10C1	ENST00000622521					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABHD16A	upstream gene variant	4619	ENST00000496579	?	?	1
MIR4646	downstream gene variant	3354	ENST00000580775	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABHD16A	ENST00000440843	NM_001177515.1	ENSP00000410347	1
ABHD16A	ENST00000482224	?	ENSP00000419742	1
ABHD16A	ENST00000395952	NM_021160.2	ENSP00000379282	1
ABHD16A	ENST00000498420	?	ENSP00000420628	1
ABHD16A	ENST00000477462	?	ENSP00000418331	1
ABHD16A	ENST00000492084	?	ENSP00000420585	1
ABHD16A	ENST00000468037	?	ENSP00000417566	1
ABHD16A	ENST00000495769	?	ENSP00000418170	1
XXbac-BPG32J3.20	ENST00000461287	?	ENSP00000475702	1

Locus 256 (sentinel SNP rs805273 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 257 (rs2506)

SNiPAcard

Block annotations

Block info

genomic range	chr10:54,504,733-54,526,622
block size	21,890 bp
variant count	57 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.534 [-3.733 – 1.202]	gene(s) hit or close-by	MBL2, RP11-556E13.1
phastCons	μ = 0.013 [0 – 0.269]	eQTL gene(s)	PRKRIRP3
GERP++	μ = -0.496 [-5.36 – 1.66]	potentially regulated gene(s)	RP11-556E13.1
CADD score	μ = 2.669 [0.013 – 9.19]	disease gene(s)	MBL2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
theophylline	3.37×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
MBL2	MANNOSE-BINDING PROTEIN DEFICIENCY	OMIM	MIM:614372
MBL2	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PRKRIRP3	?	ENSG00000227972	tibial nerve	2.66×10^{-7} (p-value)	GTEx Portal V6	39

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000074939	1	ENCP00000008494	RP11-556E13.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000355146 (CTCF binding site)	5	HSMMtube	H3K27me3
		liver (HepG2)	CTCF
ENSR00001596434 (promoter flanking region)	1	liver (HepG2)	H3K4me2, H3K9ac, H3K27ac, H3K4me1
		HSMMtube	H3K27me3
ENSR00001016746 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	FOSL2, Jund, H3K4me1, p300, H3K4me2, H3K9ac, H3K27ac
		HSMMtube	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	DNase1

		muscle (HSMM)	DNase1
ENSR00001596435 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	FOSL2, Jund, H3K4me1, p300, H3K4me2, H3K9ac, H3K27ac
		A549	H3K27me3
		nervous (NH-A)	DNase1
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MBL2	downstream gene variant	482	ENST00000373968	NM_000242.2	ENSP00000363079	9
RP11-556E13.1	upstream gene variant	421	ENST00000443523	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-556E13.1	ENST00000443523	?	?	36

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MBL2	ENST00000373968	NM_000242.2	ENSP00000363079	8

Locus 258 (rs7119167)

SNiPAcard

Block annotations

Block info

genomic range	chr11:73,090,265-73,293,082
block size	202,818 bp
variant count	32 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.377 [-4.432 – 4.671]	gene(s) hit or close-by	FAM168A, HMGN2P38, RELT, RP11-809N8.2, RP11-809N8.4, RP11-809N8.5, RP11-809N8.6
phastCons	μ = 0.110 [0 – 1]	eQTL gene(s)	ARHGEF17, FAM168A, RELT, RP11-809N8.4
GERP++	μ = 0.507 [-2.7 – 3.72]	potentially regulated gene(s)	ARHGEF17, P2RY2, P2RY6, RAB6A, RELT, RP11-800A3.2, RP11-809N8.4, RP11-809N8.5
CADD score	μ = 5.363 [0.576 – 21.4]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FAM168A	?	ENSG00000054965	blood	4.50×10^{-7} (p-value)	GTEx Portal V6	30
RP11-809N8.4	?	ENSG00000256448	blood	1.17×10^{-5} (p-value)	GTEx Portal V6	1
ARHGEF17	ENST00000543530	ILMN_1754562	monocyte	6.35×10^{-8} (p-value)	Fairfax et al.	3
ARHGEF17	ENST00000263674					
RELT	ENST00000064780	ILMN_1748614	monocyte	8.43×10^{-10} (p-value)	Fairfax et al.	3
RELT	ENST00000545886		b-cell	8.84×10^{-6} (p-value)	Fairfax et al.	3
RELT	ENST00000393580					
RELT	ENST00000539134					
ARHGEF17	ENST00000543530	ILMN_1754562	monocyte	3.23×10^{-15} (p-value)	Zeller et al.	3
ARHGEF17	ENST00000263674					
FAM168A	ENST00000064778	ILMN_1847308	blood	1.28×10^{-6} (p-value)	Westra et al.	7

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000114360	1	ENCP00000013663	RP11-800A3.2
		ENCP00000013698	RAB6A
		ENCP00000013679	RELT
		ENCP00000013669	P2RY6
		ENCP00000013664	P2RY2
		ENCP00000013678	RELT
		ENCP00000013683	RP11-809N8.4
ENCE00000114413	1	ENCP00000013676	ARHGEF17
		ENCP00000013668	P2RY6
		ENCP00000013686	RP11-809N8.5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001605004 (enhancer)	1	cervix (HeLa-S3)	PolII, H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K4me2, H3K27ac, H4K20me1, H3K36me3, H3K4me3
		blood (GM12878)	H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		blood (K562)	H3K79me2
		blood (DND-41)	H3K4me2, H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000565215 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H4K20me1, H3K36me3
		nervous (NH-A)	DNase1
		A549	H3K36me3
ENSR00001605010 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		HSMMtube	H3K27ac
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
ENSR00000565218 (promoter flanking region)	1	NHLF	DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		blood (K562)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	DNase1
		A549	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
ENSR00000565222 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
ENSR00000565232 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac, H3K4me3
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	H2AZ, H3K4me1, H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FAM168A	downstream gene variant	1655	ENST00000064778	NM_001286050.1	ENSP00000064778	2
FAM168A	downstream gene variant	2305	ENST00000356467	NM_015159.2	ENSP00000348852	1
FAM168A	downstream gene variant, upstream gene variant	181	ENST00000432223	?	?	2
FAM168A	downstream gene variant	2305	ENST00000450446	NM_001286051.1	ENSP00000390501	1
HMGN2P38	downstream gene variant	1520	ENST00000423806	?	?	1
RELT	downstream gene variant	1360	ENST00000064780	NM_152222.1	ENSP00000064780	1
RELT	downstream gene variant	591	ENST00000393580	NM_032871.3	ENSP00000377207	2
RELT	upstream gene variant, downstream gene variant	587	ENST00000545886	?	?	3
RELT	downstream gene variant	2110	ENST00000537771	?	?	2
RELT	downstream gene variant	2737	ENST00000544075	?	ENSP00000440562	2
RELT	downstream gene variant	1358	ENST00000539134	?	?	1
RP11-809N8.2	upstream gene variant	795	ENST00000544674	?	?	2
	upstream gene variant, downstream gene variant	1619		?	?	3

RP11-809N8.4			ENST00000536855			
RP11-809N8.4	upstream gene variant, downstream gene variant	1619	ENST00000542598	?	?	3
RP11-809N8.5	upstream gene variant	2522	ENST00000540641	?	?	1
RP11-809N8.6	upstream gene variant, downstream gene variant	1940	ENST00000539481	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
FAM168A	ENST00000064778	NM_001286050.1	ENSP00000064778	H	caT/caC	1
FAM168A	ENST00000356467	NM_015159.2	ENSP00000348852	H	caT/caC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FAM168A	ENST00000064778	NM_001286050.1	ENSP00000064778	26
FAM168A	ENST00000432223	?	?	5
FAM168A	ENST00000450446	NM_001286051.1	ENSP00000390501	26
FAM168A	ENST00000356467	NM_015159.2	ENSP00000348852	26
RELT	ENST00000393580	NM_032871.3	ENSP00000377207	3
RELT	ENST00000545687	?	ENSP00000439352	3
RELT	ENST00000544075	?	ENSP00000440562	3
RELT	ENST00000064780	NM_152222.1	ENSP00000064780	3
RP11-809N8.4	ENST00000536855	?	?	1
RP11-809N8.4	ENST00000542598	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FAM168A	ENST00000064778	NM_001286050.1	ENSP00000064778	1
RELT	ENST00000064780	NM_152222.1	ENSP00000064780	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RELT	ENST00000539134	?	1

Locus 259 (rs12734260)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,719,947-196,937,951
block size	218,005 bp
variant count	70 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.211 [-2.649 – 2.266]	**gene(s) hit or close-by**	CFH, CFHR2, CFHR2, CFHR4, RP4-608O15.4
phastCons	μ = 0.069 [0 – 0.987]	**eQTL gene(s)**	CFH, CFHR1, CFHR3, JUP, ZBTB41
GERP++	μ = -0.271 [-2.54 – 1.61]	**potentially regulated gene(s)**	KCNT2
CADD score	μ = 2.963 [0.062 – 14.82]	**disease gene(s)**	CFHR1, CFHR3, CFH, JUP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
HWESASXX*	1.65×10^{-5}	Metabolomics GWAS Server	24816252	8
Macular degeneration	5.99×10^{-20}	dbGaP	pha002890	5
Macular degeneration	4.93×10^{-21}	dbGaP	pha002869	3
Macular degeneration	4.26×10^{-11}	dbGaP	pha000002	1

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR1	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFHR1	C3 glomerulonephritis	OrphaNet	OrphaNet:329931
CFHR1	Dense deposit disease	OrphaNet	OrphaNet:93571
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571
CFH	Familial drusen	OrphaNet	OrphaNet:75376
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903
JUP	Lethal acantholytic epidermolysis bullosa	OrphaNet	OrphaNet:158687
JUP	NAXOS DISEASE	OrphaNet	OrphaNet:34217
JUP	Familial isolated arrhythmogenic ventricular dysplasia, right dominant form	OrphaNet	OrphaNet:293910
JUP	Familial isolated arrhythmogenic ventricular dysplasia, left dominant form	OrphaNet	OrphaNet:293888
JUP	Familial isolated arrhythmogenic ventricular dysplasia, biventricular form	OrphaNet	OrphaNet:293899
JUP	NAXOS DISEASE	OMIM	MIM:601214
JUP	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 12	OMIM	MIM:611528

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	muscularis mucosae	1.30×10^{-8} (p-value)	GTEx Portal V6	68
CFHR3	?	ENSG00000116785	adrenal gland	5.83×10^{-10} (p-value)	GTEx Portal V6	66

CFHR3	?	ENSG00000116785	lung	1.03×10-13 (p-value)	GTEx Portal V6	68
CFHR1	?	ENSG00000244414	transformed fibroblasts	9.59×10-7 (p-value)	GTEx Portal V6	58
CFHR1	?	ENSG00000244414	liver	3.11×10-9 (p-value)	GTEx Portal V6	67
CFHR3	?	ENSG00000116785	liver	2.62×10-7 (p-value)	GTEx Portal V6	60
CFHR3	?	ENSG00000116785	thyroid	4.09×10-12 (p-value)	GTEx Portal V6	68
CFHR3	?	ENSG00000116785	prostate	3.24×10-6 (p-value)	GTEx Portal V6	4
CFHR3	?	ENSG00000116785	transverse colon	7.15×10-7 (p-value)	GTEx Portal V6	51
CFHR3	?	ENSG00000116785	tibial nerve	3.80×10-6 (p-value)	GTEx Portal V6	46
CFHR3	?	ENSG00000116785	esophagus mucosa	1.67×10-6 (p-value)	GTEx Portal V6	47
CFHR3	?	ENSG00000116785	aorta	4.24×10-8 (p-value)	GTEx Portal V6	53
CFH	ENST00000359637	ILMN_1657803	skin	4.41×10-7 (p-value)	MuTHER consortium	23
CFH	ENST00000630130		adipocyte	3.96×10-7 (p-value)	MuTHER consortium	23
CFHR3	?	ENSG00000116785	stomach	3.95×10-6 (p-value)	GTEx Portal V6	10
ZBTB41	?	ENSG00000177888	subcutaneous adipocytes	2.08×10-5 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
JUP	?	ENSG00000173801	chr17	blood	4.64×10-2 (q-value)	SeeQTL DB (HapMap)	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000048799	1	ENCP00000005933	KCNT2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001527235 (TF binding site)	1	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, H3K4me2, H3K4me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	H3K27me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFH	downstream gene variant	3313	ENST00000466229	?	?	1
CFH	downstream gene variant	3313	ENST00000367429	NM_000186.3	ENSP00000356399	1
CFHR2	upstream gene variant, downstream gene variant	745	ENST00000473386	?	?	4
CFHR2	upstream gene variant, downstream gene variant	714	ENST00000496448	?	?	4
CFHR2	upstream gene variant, downstream gene variant	659	ENST00000485647	?	?	5
CFHR2	downstream gene variant	4905	ENST00000367421	?	ENSP00000356391	1

CFHR2	upstream gene variant, downstream gene variant	668	ENST00000367415	NM_005666.2	ENSP00000356385	4
CFHR2	upstream gene variant, downstream gene variant	659	ENST00000476712	?	ENSP00000476677	4
CFHR2	upstream gene variant, downstream gene variant	704	ENST00000489703	?	?	4
CFHR4	upstream gene variant, downstream gene variant	3978	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	2
CFHR4	downstream gene variant	4220	ENST00000608469	?	ENSP00000477162	1
CFHR4	upstream gene variant, downstream gene variant	4046	ENST00000251424	NM_006684.4	ENSP00000251424	2
CFHR4	upstream gene variant, downstream gene variant	4067	ENST00000367418	?	ENSP00000356388	2
RP4-608O15.4	upstream gene variant	1674	ENST00000613613	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFHR2	ENST00000473386	?	?	4
CFHR2	ENST00000367421	?	ENSP00000356391	65
CFHR2	ENST00000485647	?	?	2
CFHR2	ENST00000476712	?	ENSP00000476677	4
CFHR2	ENST00000489703	?	?	2
CFHR2	ENST00000367415	NM_005666.2	ENSP00000356385	4
CFHR2	ENST00000496448	?	?	4
CFHR4	ENST00000251424	NM_006684.4	ENSP00000251424	12
CFHR4	ENST00000608469	?	ENSP00000477162	51
CFHR4	ENST00000367418	?	ENSP00000356388	12
CFHR4	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	12

Locus 260 (rs2534674)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,464,029-31,472,308
block size	8,280 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.950 [-2.109 – 0.418]	gene(s) hit or close-by	MICB, Y_RNA
phastCons	μ = 0.040 [0 – 0.202]	eQTL gene(s)	HCG22, HCG27, HCP5, HLA-C, HLA-DRB5, HLA-DRB6, LST1, MICB, MUC22, XXbac-BPG248L24.12
GERP++	μ = -0.208 [-1.33 – 0.13]	potentially regulated gene(s)	–
CADD score	μ = 2.638 [0.78 – 7.406]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICB	?	ENSG00000204516	cerebellar hemisphere	2.83×10-6 (p-value)	GTEx Portal V6	7
HCG22	?	ENSG00000228789	visceral adipocytes	1.50×10-6 (p-value)	GTEx Portal V6	8
HCG22	?	ENSG00000228789	lung	7.16×10-7 (p-value)	GTEx Portal V6	9
MICB	?	ENSG00000204516	cerebellum	5.73×10-8 (p-value)	GTEx Portal V6	9
HCP5	?	ENSG00000206337	transformed fibroblasts	2.96×10-6 (p-value)	GTEx Portal V6	7
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	7.88×10-6 (p-value)	GTEx Portal V6	7
MUC22	?	ENSG00000261272	esophagus mucosa	3.50×10-6 (p-value)	GTEx Portal V6	7
LST1	ENST00000376099	ILMN_1688373	blood	5.96×10-9 (p-value)	MuTHER consortium	3
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					
LST1	ENST00000376089					
LST1	ENST00000396112					
LST1	ENST00000418507					
LST1	ENST00000376096					
LST1	ENST00000490742					

LST1	ENST00000211921					
LST1	ENST00000460834					
HCG22	ENST00000570223	ILMN_1667229	blood	1.28×10^{-12} (p-value)	MuTHER consortium	3
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
LST1	ENST00000376099	ILMN_2345353	blood	1.62×10^{-8} (p-value)	MuTHER consortium	3
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					
LST1	ENST00000376089					
LST1	ENST00000376096					
LST1	ENST00000418507					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000460834					
LST1	ENST00000211921					
MICB	ENST00000538442	ILMN_1708006	skin	8.01×10^{-8} (p-value)	MuTHER consortium	3
MICB	ENST00000399150					
MICB	ENST00000252229					
HCG27	ENST00000383331	ILMN_1746436	blood	1.03×10^{-6} (p-value)	MuTHER consortium	3
HCG27	ENST00000415276					
HLA-C	ENST00000470363	ILMN_2150787	blood	2.10×10^{-5} (p-value)	Westra et al.	1
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	ENST00000466892					
LST1	ENST00000419073	ILMN_1717127	blood	4.51×10^{-6} (p-value)	Westra et al.	1
LST1	ENST00000433492					
LST1	ENST00000376089					
LST1	ENST00000376090					
LST1	ENST00000211921					
LST1	?	ENSG00000204482	blood	4.68×10^{-2} (q-value)	SeeQTL DB (HapMap)	1
?	?	ILMN_1893395	monocyte	8.32×10^{-30} (p-value)	Zeller et al.	1
LST1	ENST00000339530	ILMN_1718936	monocyte	1.96×10^{-28} (p-value)	Zeller et al.	1
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					

LST1	ENST00000396112						
LST1	ENST00000490742						
LST1	ENST00000433492						
LST1	ENST00000438075						
LST1	ENST00000464044						
LST1	ENST00000396101						
?	?	ILMN_1721113	monocyte	3.76×10^{-14} (p-value)	Zeller et al.	1	
?	?	ILMN_1883997	monocyte	9.79×10^{-27} (p-value)	Zeller et al.	1	

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_2159694	chr6	monocyte	3.67×10^{-14} (p-value)	Zeller et al.	1
?	?	ILMN_1715169	chr6	monocyte	1.33×10^{-15} (p-value)	Zeller et al.	1
HLA-DRB6	ENST00000437183	ILMN_2066060	chr6	monocyte	1.45×10^{-19} (p-value)	Zeller et al.	1
HLA-DRB6	ENST00000411500						
HLA-DRB6	ENST00000437650						
HLA-DRB5	ENST00000374975	ILMN_1697499	chr6	monocyte	9.17×10^{-25} (p-value)	Zeller et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487848 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, PolII, TAF7, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	Max, ZBTB7A, CTCF, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2
		liver (HepG2)	PolII, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4me2
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K4me2, H3K4me3, DNase1, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, CTCF, H3K4me1
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K4me3
		endothelium (HUVEC)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac
ENSR00000487850 (enhancer)	2	monocytes (Monocytes-CD14+)	H4K20me1
		HSMMtube	H3K27me3
		blood (GM12878)	H3K79me2
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MICB	upstream gene variant	760	ENST00000399150	NM_001289161.1	ENSP00000382103	2
MICB	upstream gene variant	789	ENST00000252229	NM_005931.4	ENSP00000252229	2
MICB	upstream gene variant	1404	ENST00000494577	?	?	6
Y_RNA	downstream gene variant, upstream gene variant	437	ENST00000383850	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MICB	ENST00000538442	NM_001289160.1	ENSP00000442345	9
MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	7
MICB	ENST00000252229	NM_005931.4	ENSP00000252229	7

Locus 261 (rs7972001)

SNiPAcard

Block annotations

Block info

genomic range	chr12:94,620,432-94,634,913
block size	14,482 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.040 [-1.413 – 2.23]	gene(s) hit or close-by	PLXNC1
phastCons	μ = 0.166 [0 – 1]	eQTL gene(s)	UBE2N
GERP++	μ = 0.078 [-7.06 – 3.51]	potentially regulated gene(s)	–
CADD score	μ = 7.383 [2.948 – 17.47]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
ibuprofen	2.09×10^{-5}	Metabolomics GWAS Server	24816252	2

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PLXNC1	missense variant	ENST00000258526	NM_005761.2	ENSP00000258526	S/R	agT/agG	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
UBE2N	?	ENSG00000177889	transformed fibroblasts	2.12×10^{-5} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001614108 (enhancer)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	DNase1, H3K4me1, p300, FOXA1, H3K27ac
		HSMMtube	H3K27me3
		blood (GM12878)	H3K36me3
		Osteobl	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PLXNC1	downstream gene variant	2273	ENST00000546659	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
PLXNC1	ENST00000258526	NM_005761.2	ENSP00000258526	N	aaT/aaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLXNC1	ENST00000258526	NM_005761.2	ENSP00000258526	9

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PLXNC1	ENST00000546659	?	1

Locus 262 (rs17266357)

SNiPAcard

Block annotations

Block info

genomic range	chr4:102,710,688-102,756,731
block size	46,044 bp
variant count	49 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.684 [-3.212 – 0.908]	gene(s) hit or close-by	BANK1
phastCons	μ = 0.017 [0 – 0.16]	eQTL gene(s)	BANK1
GERP++	μ = -0.306 [-4.46 – 2.03]	potentially regulated gene(s)	–
CADD score	μ = 2.185 [0.066 – 7.844]	disease gene(s)	BANK1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
BANK1	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
BANK1	ENST00000428908	ILMN_1661646	blood	2.82×10^{-16} (p-value)	MuTHER consortium	14
BANK1	ENST00000504592					
BANK1	ENST00000444316					
BANK1	ENST00000322953					
BANK1	ENST00000504403					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001433620 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		HSMMtube	H3K27me3
		Osteobl	H3K4me2
		A549	H3K4me3, H3K4me2, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BANK1	upstream gene variant	1076	ENST00000322953	NM_017935.4	ENSP00000320509	1
BANK1	upstream gene variant	1199	ENST00000508653	?	ENSP00000422314	1
BANK1	upstream gene variant	1076	ENST00000428908	NM_001127507.2	ENSP00000412748	1
BANK1	upstream gene variant	4428	ENST00000444316	NM_001083907.2	ENSP00000388817	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BANK1	ENST00000322953	NM_017935.4	ENSP00000320509	48
BANK1	ENST00000508653	?	ENSP00000422314	48
BANK1	ENST00000504592	?	ENSP00000421443	49
BANK1	ENST00000428908	NM_001127507.2	ENSP00000412748	48
BANK1	ENST00000444316	NM_001083907.2	ENSP00000388817	1

Locus 263 (rs28367631)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,250,801-31,271,716
block size	20,916 bp
variant count	127 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.758 [-4.394 – 2.486]	**gene(s) hit or close-by**	HLA-C, RPL3P2, USP8P1, WASF5P, XXbac-BPG248L24.10, XXbac-BPG248L24.13
phastCons	μ = 0.063 [0 – 0.998]	**eQTL gene(s)**	C4B, CCHCR1, CD248, DDR1, GPBAR1, HCG22, HCG27, HLA-B, HLA-C, MICA, MICB, PLOD3, POU5F1, PSORS1C1, PSORS1C2, PSORS1C3, TCF19, TMEM154, TRAPPC1, WASF5P, XXbac-BPG248L24.10, XXbac-BPG248L24.12, XXbac-BPG299F13.17
GERP++	μ = -0.144 [-1.75 – 0.718]	**potentially regulated gene(s)**	CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, SFTA2, SFTA2, SFTA2, SFTA2, SFTA2, SFTA2, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P
CADD score	μ = 2.579 [0.016 – 17.05]	**disease gene(s)**	HLA-B, C4B, PLOD3

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Protein quantitative trait loci	<4.00×10-6	GWAS Catalog	18464913	1

Disease gene annotation

gene	trait	source DB	source entry/link
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147
PLOD3	Connective tissue disorder due to lysyl hydroxylase-3 deficiency	OrphaNet	OrphaNet:300284
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
PLOD3	BONE FRAGILITY WITH CONTRACTURES, ARTERIAL RUPTURE, AND [...]	OMIM	MIM:612394
PLOD3	LYSYL HYDROXYLASE 3 DEFICIENCY	DECIPHER	MIM:612394

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HCG22	ENST00000570223	ILMN_1667229	blood	5.04×10-5 (p-value)	MuTHER consortium	1
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
HCG27	ENST00000383331	ILMN_1746436	adipocyte	6.00×10-27 (p-value)	MuTHER consortium	21

HCG27	ENST00000415276		skin	1.40×10-13 (p-value)	MuTHER consortium	21
DDR1	ENST00000376567	ILMN_1812262	blood	3.85×10-7 (p-value)	MuTHER consortium	20
DDR1	ENST00000376570					
DDR1	ENST00000418800					
DDR1	ENST00000376569					
DDR1	ENST00000452441					
DDR1	ENST00000324771					
DDR1	ENST00000482873					
DDR1	ENST00000376568					
DDR1	ENST00000446312					
DDR1	ENST00000454612					
HCG27	?	ENSG00000206344	muscularis mucosae	2.64×10-14 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	muscularis mucosae	5.82×10-6 (p-value)	GTEx Portal V6	22
HCG27	?	ENSG00000206344	lung	4.59×10-9 (p-value)	GTEx Portal V6	104
PSORS1C3	?	ENSG00000204528	lung	1.12×10-7 (p-value)	GTEx Portal V6	95
HLA-C	?	ENSG00000204525	lung	2.82×10-7 (p-value)	GTEx Portal V6	99
XXbac-BPG248L24.12	?	ENSG00000271581	lung	2.21×10-6 (p-value)	GTEx Portal V6	1
PSORS1C3	?	ENSG00000204528	atrial appendage	1.61×10-9 (p-value)	GTEx Portal V6	100
HLA-C	?	ENSG00000204525	atrial appendage	4.10×10-7 (p-value)	GTEx Portal V6	86
HCG27	?	ENSG00000206344	atrial appendage	1.72×10-6 (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	transformed fibroblasts	4.37×10-11 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	transformed fibroblasts	4.61×10-13 (p-value)	GTEx Portal V6	104
C4B	?	ENSG00000224389	transformed fibroblasts	2.95×10-6 (p-value)	GTEx Portal V6	18
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	2.07×10-7 (p-value)	GTEx Portal V6	2
PSORS1C3	?	ENSG00000204528	blood	4.75×10-11 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	blood	4.64×10-15 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	blood	6.84×10-10 (p-value)	GTEx Portal V6	104
XXbac-BPG299F13.17	?	ENSG00000272501	blood	3.88×10-13 (p-value)	GTEx Portal V6	104
POU5F1	?	ENSG00000204531	blood	1.09×10-9 (p-value)	GTEx Portal V6	104
HCG27	?	ENSG00000206344	blood	2.38×10-7 (p-value)	GTEx Portal V6	13
HCG27	?	ENSG00000206344	tibial artery	7.32×10-13 (p-value)	GTEx Portal V6	104
PSORS1C3	?	ENSG00000204528	tibial artery	4.66×10-8 (p-value)	GTEx Portal V6	100
HLA-C	?	ENSG00000204525	tibial artery	9.94×10-12 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	7.57×10-7 (p-value)	GTEx Portal V6	52
HLA-B	?	ENSG00000234745	tibial artery	1.09×10-5 (p-value)	GTEx Portal V6	5
HCG27	?	ENSG00000206344	thyroid	1.80×10-15 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.10	?	ENSG00000229836	thyroid	1.19×10-12 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	thyroid	8.55×10-10 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	2.30×10-8 (p-value)	GTEx Portal V6	90
HLA-B	?	ENSG00000234745	thyroid	7.33×10-8 (p-value)	GTEx Portal V6	96

WASF5P	?	ENSG00000231402	thyroid	1.99×10-6 (p-value)	GTEx Portal V6	96
PSORS1C3	?	ENSG00000204528	thyroid	1.58×10-6 (p-value)	GTEx Portal V6	62
HCG27	?	ENSG00000206344	skeletal muscle	5.07×10-11 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	skeletal muscle	3.36×10-13 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	1.36×10-9 (p-value)	GTEx Portal V6	102
HCG27	?	ENSG00000206344	transverse colon	1.86×10-11 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	transverse colon	1.19×10-6 (p-value)	GTEx Portal V6	27
XXbac-BPG248L24.12	?	ENSG00000271581	transverse colon	3.36×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	liver	1.46×10-7 (p-value)	GTEx Portal V6	81
HCG27	?	ENSG00000206344	sun exposed skin	9.23×10-11 (p-value)	GTEx Portal V6	104
PSORS1C3	?	ENSG00000204528	sun exposed skin	1.22×10-9 (p-value)	GTEx Portal V6	103
HLA-C	?	ENSG00000204525	sun exposed skin	1.05×10-7 (p-value)	GTEx Portal V6	96
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	2.06×10-8 (p-value)	GTEx Portal V6	23
POU5F1	?	ENSG00000204531	sun exposed skin	6.68×10-11 (p-value)	GTEx Portal V6	103
PSORS1C3	?	ENSG00000204528	unexposed skin	3.01×10-7 (p-value)	GTEx Portal V6	88
HLA-C	?	ENSG00000204525	unexposed skin	7.04×10-7 (p-value)	GTEx Portal V6	94
POU5F1	?	ENSG00000204531	unexposed skin	5.17×10-8 (p-value)	GTEx Portal V6	102
HCG27	?	ENSG00000206344	unexposed skin	1.41×10-6 (p-value)	GTEx Portal V6	9
PSORS1C3	?	ENSG00000204528	left ventricle	1.05×10-6 (p-value)	GTEx Portal V6	62
HCG27	?	ENSG00000206344	aorta	6.79×10-7 (p-value)	GTEx Portal V6	87
HLA-C	?	ENSG00000204525	aorta	7.00×10-12 (p-value)	GTEx Portal V6	104
HCG27	?	ENSG00000206344	subcutaneous adipocytes	6.73×10-17 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	4.19×10-10 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	3.75×10-8 (p-value)	GTEx Portal V6	11
HCG27	?	ENSG00000206344	stomach	5.12×10-8 (p-value)	GTEx Portal V6	104
PSORS1C3	?	ENSG00000204528	stomach	3.67×10-7 (p-value)	GTEx Portal V6	93
HCG27	?	ENSG00000206344	visceral adipocytes	1.20×10-7 (p-value)	GTEx Portal V6	101
HLA-C	?	ENSG00000204525	visceral adipocytes	3.81×10-7 (p-value)	GTEx Portal V6	83
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	3.09×10-7 (p-value)	GTEx Portal V6	84
PSORS1C3	?	ENSG00000204528	spleen	7.06×10-7 (p-value)	GTEx Portal V6	86
HCG27	?	ENSG00000206344	nucleus accumbens	9.32×10-8 (p-value)	GTEx Portal V6	88
HCG27	?	ENSG00000206344	sigmoid colon	1.10×10-7 (p-value)	GTEx Portal V6	86
HLA-C	?	ENSG00000204525	sigmoid colon	3.69×10-7 (p-value)	GTEx Portal V6	70
HCG27	?	ENSG00000206344	tibial nerve	3.74×10-18 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	tibial nerve	1.16×10-10 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	8.13×10-6 (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	tibial nerve	6.97×10-6 (p-value)	GTEx Portal V6	9

HLA-B	?	ENSG00000234745	tibial nerve	6.97×10-6 (p-value)	GTEx Portal V6	9
PSORS1C3	?	ENSG00000204528	esophagus mucosa	8.85×10-10 (p-value)	GTEx Portal V6	103
PSORS1C2	?	ENSG00000204538	esophagus mucosa	3.96×10-8 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	esophagus mucosa	2.95×10-11 (p-value)	GTEx Portal V6	104
TCF19	?	ENSG00000137310	esophagus mucosa	5.99×10-11 (p-value)	GTEx Portal V6	104
CCHCR1	?	ENSG00000204536	esophagus mucosa	7.87×10-7 (p-value)	GTEx Portal V6	93
POU5F1	?	ENSG00000204531	esophagus mucosa	5.23×10-12 (p-value)	GTEx Portal V6	103
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	1.20×10-6 (p-value)	GTEx Portal V6	3
?	?	ILMN_1883997	blood	4.92×10-6 (p-value)	Westra et al.	1
MICB	ENST00000538442	ILMN_1708006	blood	9.19×10-7 (p-value)	Westra et al.	13
MICB	ENST00000399150					
MICB	ENST00000252229					
HCG27	?	ENSG00000206344	adrenal gland	2.17×10-7 (p-value)	GTEx Portal V6	38
HCG27	?	ENSG00000206344	testis	2.07×10-7 (p-value)	GTEx Portal V6	87
HLA-C	?	ENSG00000204525	testis	3.85×10-8 (p-value)	GTEx Portal V6	96
PSORS1C1	?	ENSG00000204540	testis	3.06×10-6 (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	pancreas	2.72×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	EBV lymphocytes	2.10×10-6 (p-value)	GTEx Portal V6	1
CCHCR1	?	ENSG00000204536	blood	1.78×10-2 (q-value)	SeeQTL DB (HapMap)	8
HLA-C	ENST00000470363	ILMN_2150787	monocyte	2.06×10-20 (p-value)	Zeller et al.	4
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	ENST00000466892					
?	?	ILMN_1721113	monocyte	3.03×10-30 (p-value)	Zeller et al.	4
TCF19	ENST00000496421	223274_at	blood	3.20×10-8 (p-value)	Dixon et al.	2
TCF19	ENST00000376257					
TCF19	ENST00000376255					
HLA-C	?	ENSG00000204525	gastroesophageal junction	3.29×10-6 (p-value)	GTEx Portal V6	2
HCG27	?	ENSG00000206344	caudate basal ganglia	1.53×10-6 (p-value)	GTEx Portal V6	4
PSORS1C3	ENST00000412143	ILMN_3309634	b-cell	2.61×10-8 (p-value)	Fairfax et al.	6
MICA	ENST00000449934	ILMN_3241692	monocyte	9.22×10-5 (p-value)	Fairfax et al.	5
MICA	ENST00000616296					
MICA	ENST00000421350					
HCG27	?	ENSG00000206344	breast	2.13×10-6 (p-value)	GTEx Portal V6	52
XXbac-BPG299F13.17	?	ENSG00000272501	frontal cortex	7.70×10-7 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
PLOD3	ENST00000223127	ILMN_1774836	chr7	blood	1.47×10-18 (p-value)	Westra et al.	1

PLOD3	ENST00000454310						
TMEM154	ENST00000304385	ILMN_2088124	chr4	blood	2.87×10^{-25} (p-value)	Westra et al.	1
CD248	ENST00000311330	ILMN_1726589	chr11	blood	1.06×10^{-8} (p-value)	Westra et al.	1
GPBAR1	ENST00000519574	ILMN_2316386	chr2	blood	2.65×10^{-8} (p-value)	Westra et al.	1
GPBAR1	ENST00000479077						
GPBAR1	ENST00000521462						
GPBAR1	ENST00000522678						
TRAPPC1	ENST00000303731	ILMN_1716913	chr17	blood	3.28×10^{-7} (p-value)	Westra et al.	1
TRAPPC1	ENST00000575639						
TRAPPC1	ENST00000571739						
TRAPPC1	ENST00000571947						
TRAPPC1	ENST00000572656						
TRAPPC1	ENST00000573989						
TRAPPC1	ENST00000540486						

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444768	1	ENCP00000047982	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
		ENCP00000048000	WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P
		ENCP00000047981	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
ENCE00000444720	2	ENCP00000047970	SFTA2 SFTA2 SFTA2 SFTA2 SFTA2 SFTA2
ENCE00000444721	6	ENCP00000047970	SFTA2 SFTA2 SFTA2 SFTA2 SFTA2 SFTA2
ENCE00000445080	6	ENCP00000048049	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000048087	DDAH2 DDAH2 DDAH2 DDAH2

				DDAH2 DDAH2 DDAH2		

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RPL3P2	downstream gene variant	1505	ENST00000413027	?	?	17
USP8P1	downstream gene variant	4270	ENST00000494673	?	?	9
WASF5P	downstream gene variant, upstream gene variant	206	ENST00000428639	?	?	24
XXbac-BPG248L24.10	downstream gene variant	3876	ENST00000421191	?	?	3
XXbac-BPG248L24.13	downstream gene variant, upstream gene variant	17	ENST00000539514	?	?	16

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HLA-C	ENST00000620806	?	ENSP00000480331	127
XXbac-BPG248L24.13	ENST00000539514	?	?	90

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
WASF5P	ENST00000428639	?	1
XXbac-BPG248L24.13	ENST00000539514	?	5

Locus 265 (rs2659067)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,484,553-51,487,175
block size	2,623 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.287 [-4.422 – 0.646]	gene(s) hit or close-by	CTB-147C22.9, KLK7
phastCons	μ = 0.001 [0 – 0.002]	eQTL gene(s)	KLK7
GERP++	μ = -0.817 [-6.68 – 2.29]	potentially regulated gene(s)	KLK7
CADD score	μ = 4.290 [1.036 – 8.337]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
KLK7	?	ENSG00000169035	sun exposed skin	3.70×10^{-10} (p-value)	GTEx Portal V6	6
KLK7	?	ENSG00000169035	unexposed skin	4.61×10^{-8} (p-value)	GTEx Portal V6	6

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000341941	1	ENST00000304045, ENST00000391807	KLK7

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KLK7	downstream gene variant	157	ENST00000595638	?	?	4
KLK7	upstream gene variant	5	ENST00000593904	?	ENSP00000470084	1
KLK7	upstream gene variant	104	ENST00000391807	NM_139277.2	ENSP00000375683	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
KLK7	ENST00000595820	NM_005046.3	ENSP00000470538	L	Tta/Cta	1
KLK7	ENST00000391807	NM_139277.2	ENSP00000375683	L	Tta/Cta	1
KLK7	ENST00000304045	NM_001243126.1	ENSP00000304791	L	Tta/Cta	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTB-147C22.9	ENST00000594512	?	?	6
KLK7	ENST00000595820	NM_005046.3	ENSP00000470538	6

KLK7	ENST00000391807	NM_139277.2	ENSP00000375683	5
KLK7	ENST00000593904	?	ENSP00000470084	5
KLK7	ENST00000304045	NM_001243126.1	ENSP00000304791	6
KLK7	ENST00000597707	NM_001207053.1	ENSP00000469950	6

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLK7	ENST00000595820	NM_005046.3	ENSP00000470538	2
KLK7	ENST00000593904	?	ENSP00000470084	1
KLK7	ENST00000304045	NM_001243126.1	ENSP00000304791	2
KLK7	ENST00000597707	NM_001207053.1	ENSP00000469950	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
KLK7	ENST00000595638	?	2

Locus 266 (rs34436026)

SNiPAcard

Block annotations

Block info

genomic range	chr1:161,490,524-161,519,440
block size	28,917 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.281 [-2.127 – 0.551]	gene(s) hit or close-by	FCGR2A, FCGR3A, HSPA6, RP11-25K21.6, RPS23P10
phastCons	μ = 0.001 [0 – 0.003]	eQTL gene(s)	–
GERP++	μ = 0.479 [-0.451 – 1.41]	potentially regulated gene(s)	–
CADD score	μ = 2.633 [0.417 – 4.352]	disease gene(s)	FCGR3A

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
FCGR3A	IMMUNODEFICIENCY 20	OMIM	MIM:615707

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000546473 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, CTCF, PolII, Rad21, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	DNase1, H3K9ac, H3K4me2, H3K27me3
		blood (K562)	H3K27ac, Max, H3K9ac, ETS1, Cmyc, HEY1, H2AZ, TAF1, PolII, H3K4me2, DNase1, H3K4me3, Egr1
		skin (NHDF-AD)	DNase1, H3K4me3
		muscle (HSMM)	H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H2AZ, H3K4me2, H3K4me3, H3K27me3, DNase1
		blood (GM12878)	PolII, H2AZ, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K27ac, H3K9ac, H3K4me3, H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, PolII, H3K4me3, Gabp
		monocytes (Monocytes-CD14+)	H3K27ac, H3K27me3, H3K4me3, H3K4me2, H3K4me1, DNase1
		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, PolII, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001587253 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR2A	downstream gene variant	2144	ENST00000461298	?	?	1
FCGR2A	downstream gene variant	1166	ENST00000271450	NM_001136219.1, NM_201563.5	ENSP00000271450	1
FCGR2A	downstream gene variant	2131	ENST00000486608	?	?	1
FCGR2A	downstream gene variant	2176	ENST00000367972	NM_021642.3	ENSP00000356949	1

FCGR2A	downstream gene variant	2169	ENST00000483665	?	ENSP00000440148 1
FCGR2A	downstream gene variant	2379	ENST00000471026	?	? 1
FCGR2A	downstream gene variant	767	ENST00000459885	?	ENSP00000476411 1
FCGR3A	downstream gene variant	3187	ENST00000426740	?	ENSP00000410180 1
FCGR3A	downstream gene variant	3095	ENST00000443193	?	ENSP00000392047 1
FCGR3A	downstream gene variant	3097	ENST00000436743	NM_001127595.1, NM_001127593.1, NM_001127596.1	ENSP00000416607 1
FCGR3A	downstream gene variant	3091	ENST00000367969	NM_000569.6, NM_001127592.1	ENSP00000356946 1
FCGR3A	downstream gene variant	3128	ENST00000367967	?	ENSP00000356944 1
HSPA6	upstream gene variant, downstream gene variant	3806	ENST00000309758	NM_002155.3	ENSP00000310219 2
RPS23P10	downstream gene variant, upstream gene variant	510	ENST00000416185	?	? 2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2A	ENST00000467525	?	ENSP00000476495	1
FCGR2A	ENST00000491841	?	?	1
FCGR3A	ENST00000442336	?	ENSP00000396567	1
FCGR3A	ENST00000367969	NM_000569.6, NM_001127592.1	ENSP00000356946	1
FCGR3A	ENST00000476031	?	?	1
FCGR3A	ENST00000426740	?	ENSP00000410180	1
FCGR3A	ENST00000443193	?	ENSP00000392047	1
FCGR3A	ENST00000436743	NM_001127595.1, NM_001127593.1, NM_001127596.1	ENSP00000416607	1
FCGR3A	ENST00000367967	?	ENSP00000356944	1
RP11-25K21.6	ENST00000537821	?	?	4

Locus 267 (rs11645898)

SNiPAcard

Block annotations

Block info

genomic range	chr16:72,151,101-72,243,277
block size	92,177 bp
variant count	37 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.280 [-1.415 – 3.761]	gene(s) hit or close-by	DHX38, PMFBP1
phastCons	μ = 0.151 [0 – 1]	eQTL gene(s)	DHODH, DHX38, HP, HPR, PMFBP1, TXNL4B
GERP++	μ = 0.419 [-6.41 – 6.17]	potentially regulated gene(s)	PMFBP1
CADD score	μ = 5.929 [0.052 – 27.3]	disease gene(s)	DHODH, HP

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
DHODH	POSTAXIAL ACROFACIAL DYSOSTOSIS	OMIM	MIM:263750
HP	ANHAPTOGLOBINEMIA	OMIM	MIM:614081
DHODH	postaxial acrofacial dysostosis (POADS)	DECIPHER	MIM:263750
DHODH	Postaxial acrofacial dysostosis	OrphaNet	OrphaNet:246

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PMFBP1	missense variant (splice region), missense variant	ENST00000537465	?	ENSP00000443817	2	2			2
PMFBP1	missense variant (splice region), missense variant	ENST00000355636	NM_001160213.1	ENSP00000347854	2	2			2
PMFBP1	missense variant (splice region), missense variant	ENST00000237353	NM_031293.2	ENSP00000237353	2	2			2

Amino acid sequence alteration (splice region)

gene	effect type	affected transcript	RefSeq id	protein	amino acid	codon	SIFT prediction	PolyPhen prediction	varian(s)
PMFBP1	missense variant	ENST00000537392	?	ENSP00000439564	K/T	aAg/aCg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DHODH	?	ENSG00000102967	muscularis mucosae	1.31×10-7 (p-value)	GTEx Portal V6	37
HPR	?	ENSG00000261701	tibial nerve	8.46×10-12 (p-value)	GTEx Portal V6	6
DHODH	?	ENSG00000102967	tibial nerve	3.29×10-7 (p-value)	GTEx Portal V6	27
HP	?	ENSG00000257017	tibial nerve	4.59×10-10 (p-value)	GTEx Portal V6	6
DHODH	?	ENSG00000102967	transformed fibroblasts	1.15×10-9 (p-value)	GTEx Portal V6	37

DHODH	?	ENSG00000102967	transformed fibroblasts	1.15×10-9 (p-value)	GTEx Portal V6	37
HPR	?	ENSG00000261701	tibial artery	3.19×10-12 (p-value)	GTEx Portal V6	18
DHODH	?	ENSG00000102967	tibial artery	5.31×10-8 (p-value)	GTEx Portal V6	30
HP	?	ENSG00000257017	tibial artery	7.12×10-7 (p-value)	GTEx Portal V6	5
TXNL4B	?	ENSG00000140830	tibial artery	8.53×10-7 (p-value)	GTEx Portal V6	3
PMFBP1	?	ENSG00000118557	esophagus mucosa	9.96×10-8 (p-value)	GTEx Portal V6	33
DHODH	?	ENSG00000102967	skeletal muscle	3.84×10-6 (p-value)	GTEx Portal V6	23
DHODH	?	ENSG00000102967	transverse colon	3.61×10-8 (p-value)	GTEx Portal V6	35
HPR	?	ENSG00000261701	aorta	4.73×10-12 (p-value)	GTEx Portal V6	37
DHODH	?	ENSG00000102967	subcutaneous adipocytes	7.14×10-9 (p-value)	GTEx Portal V6	37
HPR	?	ENSG00000261701	subcutaneous adipocytes	3.60×10-9 (p-value)	GTEx Portal V6	5
PMFBP1	ENST00000355636	ILMN_1713867	adipocyte	1.78×10-5 (p-value)	MuTHER consortium	7
PMFBP1	ENST00000237353					
PMFBP1	ENST00000537465					
PMFBP1	ENST00000379073					
DHX38	ENST00000567552	ILMN_1735679	blood	9.66×10-11 (p-value)	MuTHER consortium	10
DHX38	ENST00000268482					
DHX38	ENST00000563819					
DHX38	ENST00000579387					
HP	ENST00000570083	ILMN_1812433	monocyte	2.30×10-11 (p-value)	Fairfax et al.	1
HP	ENST00000567185					
HP	ENST00000566821					
HP	ENST00000613898					
HP	ENST00000565574					
HP	ENST00000228226					
HP	ENST00000567612					
HP	ENST00000355906					
HP	ENST00000398131					
HP	ENST00000564499					
HP	ENST00000357763					
HP	?	ENSG00000257017	blood	2.98×10-6 (p-value)	GTEx Portal V6	2
HPR	?	ENSG00000261701	sun exposed skin	2.75×10-7 (p-value)	GTEx Portal V6	5
DHODH	?	ENSG00000102967	sun exposed skin	1.39×10-5 (p-value)	GTEx Portal V6	1
PMFBP1	ENST00000355636	ILMN_1713867	monocyte	4.80×10-13 (p-value)	Zeller et al.	4
PMFBP1	ENST00000237353					
PMFBP1	ENST00000537465					
PMFBP1	ENST00000379073					
HP	ENST00000570083	ILMN_1812433	monocyte	1.16×10-40 (p-value)	Zeller et al.	4
HP	ENST00000567185					
HP	ENST00000566821					
HP	ENST00000613898					
HP	ENST00000565574					
HP	ENST00000228226					
HP	ENST00000355906					
HP	ENST00000567612					
HP	ENST00000398131					

HP	ENST00000564499					
HP	ENST00000357763					
HPR	?	ENSG00000261701	thyroid	2.10×10-7 (p-value)	GTEx Portal V6	3
DHX38	ENST00000567552	ILMN_1735679	blood	6.16×10-7 (p-value)	Westra et al.	2
DHX38	ENST00000268482					
DHX38	ENST00000563819					
DHX38	ENST00000579387					
PMFBP1	?	ENSG00000118557	EBV lymphocytes	8.30×10-6 (p-value)	GTEx Portal V6	1
HPR	?	ENSG00000261701	breast	4.30×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000021719		

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000053177 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, USF1, DNase1
		HSMMtube	DNase1, CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, Rad21, USF1, H3K4me1, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H2AZ, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K4me1
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00000509744 (promoter flanking region)	2	cervix (HeLa-S3)	DNase1
		NHLF	DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Cjun
		liver (HepG2)	DNase1
		Osteobl	H3K9me3, H3K27ac
		skin (NHDF-AD)	DNase1
ENSR00000665811 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		liver (HepG2)	Rad21, CTCF
		blood (K562)	CTCF, Rad21, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DHX38	downstream gene variant	4299	ENST00000579387	?	ENSP00000462149	1
DHX38	downstream gene variant	4294	ENST00000562774	?	ENSP00000462965	1
DHX38	downstream gene variant	4293	ENST00000567142	?	ENSP00000454521	1

DHX38	downstream gene variant	4290	ENST00000268482	NM_014003.3	ENSP00000268482	1
DHX38	downstream gene variant	4563	ENST00000563819	?	ENSP00000462397	1
DHX38	downstream gene variant	4552	ENST00000567552	?	?	1
PMFBP1	downstream gene variant, upstream gene variant	344	ENST00000237353	NM_031293.2	ENSP00000237353	4
PMFBP1	upstream gene variant	4214	ENST00000537792	?	ENSP00000443366	1
PMFBP1	downstream gene variant, upstream gene variant	363	ENST00000536211	?	ENSP00000439841	4
PMFBP1	downstream gene variant, upstream gene variant	344	ENST00000537465	?	ENSP00000443817	4
PMFBP1	downstream gene variant, upstream gene variant	347	ENST00000355636	NM_001160213.1	ENSP00000347854	3
PMFBP1	downstream gene variant, upstream gene variant	348	ENST00000379073	?	ENSP00000368364	4
PMFBP1	downstream gene variant, upstream gene variant	107	ENST00000535461	?	ENSP00000444953	3
PMFBP1	downstream gene variant, upstream gene variant	236	ENST00000539172	?	ENSP00000440941	3
PMFBP1	downstream gene variant, upstream gene variant	249	ENST00000543746	?	?	4
PMFBP1	downstream gene variant, upstream gene variant	15	ENST00000537392	?	ENSP00000439564	7
PMFBP1	downstream gene variant, upstream gene variant	43	ENST00000540440	?	ENSP00000440755	7

Putative effect on transcript

3'-UTR variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
PMFBP1	ENST00000379073	?	ENSP00000368364	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PMFBP1	ENST00000536211	?	ENSP00000439841	11
PMFBP1	ENST00000355636	NM_001160213.1	ENSP00000347854	22
PMFBP1	ENST00000537792	?	ENSP00000443366	7
PMFBP1	ENST00000237353	NM_031293.2	ENSP00000237353	22
PMFBP1	ENST00000543746	?	?	11
PMFBP1	ENST00000537465	?	ENSP00000443817	22
PMFBP1	ENST00000535461	?	ENSP00000444953	12
PMFBP1	ENST00000379073	?	ENSP00000368364	22
PMFBP1	ENST00000539172	?	ENSP00000440941	11
PMFBP1	ENST00000537392	?	ENSP00000439564	5
PMFBP1	ENST00000540440	?	ENSP00000440755	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PMFBP1	ENST00000355636	NM_001160213.1	ENSP00000347854	1
PMFBP1	ENST00000237353	NM_031293.2	ENSP00000237353	1
PMFBP1	ENST00000537465	?	ENSP00000443817	1
PMFBP1	ENST00000379073	?	ENSP00000368364	2

Locus 268 (rs990529)

SNiPAcard

Block annotations

Block info

genomic range	chr21:28,346,546-28,346,546
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-1.027	**gene(s) hit or close-by**	–
phastCons	0	**eQTL gene(s)**	–
GERP++	-1.07	**potentially regulated gene(s)**	–
CADD score	1.256	**disease gene(s)**	–

Locus 269 (rs630510)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,149,581-136,150,484
block size	904 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.148 [-1.212 – 0.562]	**gene(s) hit or close-by**	ABO
phastCons	μ = 0.009 [0.001 – 0.016]	**eQTL gene(s)**	ABO, MED22
GERP++	μ = -0.746 [-1.19 – 0.225]	**potentially regulated gene(s)**	ADAMTSL2, CELP, DBH, RALGDS, SARDH
CADD score	μ = 6.844 [5.434 – 8.361]	**disease gene(s)**	DBH, ADAMTSL2, SARDH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
ADpSGEGDFXAEGGGVR*	3.53×10^{-9}	Metabolomics GWAS Server	24816252	1
Pancreatic neoplasms	4.70×10^{-6}	dbGaP	pha002889	1
Pancreatic neoplasms	4.28×10^{-5}	dbGaP	pha002874	1
E-selectin	9.65×10^{-13}	dbGaP	pha002871	1

Disease gene annotation

gene	trait	source DB	source entry/link
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY, CONGENITAL	OMIM	MIM:223360
ADAMTSL2	GELEOPHYSIC DYSPLASIA 1	OMIM	MIM:231050
SARDH	SARCOSINEMIA	OMIM	MIM:268900
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY	OrphaNet	OrphaNet:230
ADAMTSL2	Geleophysic dysplasia	OrphaNet	OrphaNet:2623
SARDH	SARCOSINEMIA	OrphaNet	OrphaNet:3129

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABO	?	ENSG00000175164	pancreas	9.62×10^{-10} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	adrenal gland	6.23×10^{-12} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	tibial nerve	9.48×10^{-9} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	transverse colon	1.58×10^{-7} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	esophagus mucosa	5.75×10^{-11} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	thyroid	6.65×10^{-13} (p-value)	GTEx Portal V6	4
MED22	ENST00000614493	ILMN_1697218	monocyte	1.74×10^{-5} (p-value)	Fairfax et al.	1
MED22	ENST00000610672		b-cell	1.18×10^{-5} (p-value)	Fairfax et al.	1
MED22	ENST00000343730					

MED22	ENST00000610888

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000558528	1	ENCP00000060534	SARDH
		ENCP00000060486	CELP
		ENCP00000060489	RALGDS
		ENCP00000060527	ADAMTSL2
		ENCP00000060525	ADAMTSL2
		ENCP00000060535	SARDH
		ENCP00000060529	DBH

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000251886 (promoter)	4	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	CTCF, Rad21, Gabp, H3K4me2, H3K4me3, H3K27me3, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	ZBTB7A, H3K27ac, H3K9ac, Cmyc, H3K79me2, H3K4me3, DNase1, CTCF, H3K4me2, PolII, CTCFL, H2AZ
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000538324	?	ENSP00000483018	4
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	4
ABO	ENST00000453660	?	?	4

Locus 270 (rs6575449)

SNiPAcard

Block annotations

Block info

genomic range	chr14:95,094,014-95,097,666
block size	3,653 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.513 [-1.412 – -0.013]	gene(s) hit or close-by	ADIPOR1P2, SERPINA3, SERPINA3
phastCons	μ = 0.001 [0 – 0.002]	eQTL gene(s)	–
GERP++	μ = -0.490 [-2.45 – 1.3]	potentially regulated gene(s)	–
CADD score	μ = 2.221 [0.102 – 6.546]	disease gene(s)	SERPINA3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SERPINA3	Alpha-1-antichymotrypsin deficiency	OrphaNet	OrphaNet:93594

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1838581	monocyte	5.48×10-4 (p-value)	Fairfax et al.	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ADIPOR1P2	upstream gene variant	2376	ENST00000554199	?	?	6
SERPINA3	downstream gene variant	3622	ENST00000393080	?	ENSP00000376795	1
SERPINA3	downstream gene variant	3625	ENST00000393078	NM_001085.4	ENSP00000376793	1
SERPINA3	downstream gene variant	3623	ENST00000556968	?	ENSP00000452476	1
SERPINA3	downstream gene variant	3625	ENST00000482740	?	ENSP00000451119	1
SERPINA3	downstream gene variant	3623	ENST00000467132	?	ENSP00000450540	1
SERPINA3	downstream gene variant	3031	ENST00000553947	?	ENSP00000452367	2
SERPINA3	downstream gene variant	4914	ENST00000556388	?	?	1

Locus 271 (rs4418214)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,380,034-31,391,401
block size	11,368 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.000 [–]	gene(s) hit or close-by	HCP5, MICA
phastCons	μ = 0.000 [–]	eQTL gene(s)	MICA, MICB, PSORS1C2, TNXA, TNXB, XXbac-BPG181B23.7
GERP++	μ = -1.225 [-2.45 – 0]	potentially regulated gene(s)	–
CADD score	μ = 4.632 [4.233 – 5.031]	disease gene(s)	TNXB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
HIV-1 susceptibility	$<4.00\times10^{-11}$	GWAS Catalog	23935489	1
HIV-1 control	$<1.00\times10^{-34}$	GWAS Catalog	21051598	1

Disease gene annotation

gene	trait	source DB	source entry/link
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICA	?	ENSG00000204520	pancreas	2.01×10^{-12} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	pancreas	3.75×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	muscularis mucosae	3.13×10^{-10} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	muscularis mucosae	6.11×10^{-13} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	muscularis mucosae	4.75×10^{-7} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	lung	1.75×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	lung	6.14×10^{-14} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	lung	3.14×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	atrial appendage	6.38×10^{-10} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	transformed fibroblasts	6.61×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	2.25×10^{-16} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	transformed fibroblasts	4.12×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	blood	1.23×10^{-11} (p-value)	GTEx Portal V6	2

XXbac-BPG181B23.7	?	ENSG00000272221	blood	7.53×10^{-13} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	blood	1.78×10^{-6} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	breast	1.18×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	breast	3.41×10^{-11} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	breast	8.43×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial artery	4.75×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	1.80×10^{-17} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	tibial artery	7.33×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	thyroid	7.64×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	1.88×10^{-15} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	thyroid	2.47×10^{-9} (p-value)	GTEx Portal V6	2
PSORS1C2	?	ENSG00000204538	thyroid	2.21×10^{-6} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	skeletal muscle	7.03×10^{-10} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	skeletal muscle	9.61×10^{-11} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	prostate	2.29×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	prostate	8.17×10^{-12} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	transverse colon	3.92×10^{-9} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	transverse colon	1.18×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	sun exposed skin	9.48×10^{-16} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	6.43×10^{-16} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	sun exposed skin	6.95×10^{-9} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	unexposed skin	5.77×10^{-9} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	unexposed skin	1.22×10^{-10} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	aorta	4.89×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	aorta	2.71×10^{-9} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	left ventricle	1.81×10^{-11} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	left ventricle	6.68×10^{-15} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	subcutaneous adipocytes	5.34×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	1.04×10^{-12} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	subcutaneous adipocytes	1.29×10^{-9} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	terminal ileum	5.11×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	terminal ileum	3.42×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	caudate basal ganglia	1.06×10^{-6} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	stomach	2.56×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	stomach	2.18×10^{-11} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	visceral adipocytes	4.85×10^{-10} (p-value)	GTEx Portal V6	2

XXbac-BPG181B23.7	?	ENSG00000272221	visceral adipocytes	3.15×10^{-11} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	spleen	1.65×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	sigmoid colon	3.37×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	coronary artery	6.58×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	coronary artery	2.18×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial nerve	8.74×10^{-17} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	3.76×10^{-13} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	tibial nerve	1.52×10^{-9} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	esophagus mucosa	3.15×10^{-11} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	esophagus mucosa	4.80×10^{-12} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	gastroesophageal junction	1.40×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	gastroesophageal junction	3.56×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	adrenal gland	9.32×10^{-10} (p-value)	GTEx Portal V6	2
TNXB	?	ENSG00000168477	adrenal gland	2.07×10^{-6} (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	adrenal gland	3.08×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	testis	1.12×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	testis	3.86×10^{-12} (p-value)	GTEx Portal V6	2
MICB	ENST00000538442	ILMN_1708006	skin	8.50×10^{-10} (p-value)	MuTHER consortium	1
MICB	ENST00000399150		blood	3.34×10^{-6} (p-value)	MuTHER consortium	1
MICB	ENST00000252229					
MICB	ENST00000399150	ILMN_1708006	monocyte	3.63×10^{-6} (p-value)	Fairfax et al.	1
MICB	ENST00000538442		b-cell	4.35×10^{-9} (p-value)	Fairfax et al.	1
MICB	ENST00000252229					

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	2
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	ENST00000421350	?	ENSP00000402410	1

Locus 272 (rs7730934)

SNiPAcard

Block annotations

Block info

genomic range	chr5:55,228,754-55,318,612
block size	89,859 bp
variant count	55 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.273 [-4.321 – 4.663]	gene(s) hit or close-by	AC008914.1, CTD-2031P19.4, CTD-2031P19.5, FLJ31104, IL6ST, snoU13
phastCons	μ = 0.053 [0 – 1]	eQTL gene(s)	IL6ST
GERP++	μ = -0.092 [-4.49 – 5.76]	potentially regulated gene(s)	ANKRD55, CTC-236F12.4, DDX4, IL6ST, RP11-365H8.2, SLC38A9
CADD score	μ = 4.278 [0.005 – 23.9]	disease gene(s)	ANKRD55

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ANKRD55	Oligoarticular juvenile arthritis	OrphaNet	OrphaNet:85410
ANKRD55	Juvenile rheumatoid factor-negative polyarthritis	OrphaNet	OrphaNet:85408

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IL6ST	missense variant	ENST00000381298	NM_002184.3	ENSP00000370698	2	2	?, deleterious	?, benign	2
IL6ST	missense variant	ENST00000381294	NM_001190981.1	ENSP00000370694	2	2	?, deleterious	?, benign	2
IL6ST	missense variant	ENST00000522633	?	ENSP00000435399	G/R	Ggt/Cgt	deleterious	benign	1
IL6ST	missense variant	ENST00000502326	?	ENSP00000462158	2	2	?, deleterious	?, benign	2
IL6ST	missense variant	ENST00000503773	?	ENSP00000426224	G/R	Ggt/Cgt	deleterious	benign	1
IL6ST	missense variant	ENST00000336909	?	ENSP00000338799	2	2	?, deleterious	?, benign	2
IL6ST	missense variant	ENST00000381287	NM_175767.2	ENSP00000370687	G/R	Ggt/Cgt	deleterious	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL6ST	?	ENSG00000134352	tibial nerve	3.31×10-8 (p-value)	GTEx Portal V6	20

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000697123	1	ENST00000381287, ENST00000381298, ENST00000381294, ENST00000336909	IL6ST

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000409418	1	ENCP00000044145	ANKRD55
ENCE00000409253	2	ENCP00000044146	RP11-365H8.2
		ENCP00000044129	DDX4
		ENCP00000044127	SLC38A9
		ENCP00000044151	CTC-236F12.4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001280383 (promoter flanking region)	1	NHLF	H3K36me3, DNase1
		embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	DNase1, CTCF
		Osteobl	H3K36me3
		blood (K562)	CTCF
		skin (NHDF-AD)	DNase1, CTCF
		breast (HMEC)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H4K20me1, H3K36me3
		endothelium (HUVEC)	CTCF, H3K36me3
		liver (HepG2)	H3K79me2, Rad21, CTCF, H3K36me3, DNase1
		blood (GM12878)	Yy1, Cjun, BATF, Tcf12, CTCF, IRF4, Rad21, H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3, CTCF
		A549	H3K36me3
ENSR00001409454 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		Osteobl	H3K36me3
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H3K36me3, CTCF, H3K79me2, Rad21
		blood (GM12878)	CTCF, H3K79me2, H3K4me3
		lung (IMR90)	H3K36me3
		A549	H3K4me3, CTCF
		skin (NHEK)	CTCF
ENSR00001409459 (promoter)	4	embryonic stem cell (H1ESC)	H3K9ac, TAF1, H3K4me2, SP1, H3K4me3, Sin3Ak20, PolII, H3K27ac, CTCF, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		blood (K562)	TAF1, Cfos, PolII, H3K4me2, DNase1, H3K4me3, H2AZ, HEY1, H3K79me2, Egr1, H3K27ac, Max, H3K9ac
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	PolII, H3K79me2, TAF1, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K4me3, H3K9ac, H3K56ac
		blood (GM12878)	H3K9ac, H3K4me2, EBF1, Pbx3, PolII, H2AZ, Tcf12, DNase1, H3K79me2, H3K4me3, BCL11A, Pax5, H3K27ac, Cfos
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, CTCF
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H3K36me3
		blood (DND-41)	H3K4me2, H3K9ac, H3K4me3, H3K27me3
		breast (HMEC)	DNase1, H3K4me2, H3K9ac, H3K4me3, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1, Cmyc, Cjun
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001693442	1	blood (DND-41)	H3K27me3

(enhancer)		skin (NHDF-AD)	H3K4me1, DNase1, H3K27ac
ENSR00001280396 (enhancer)	3	blood (GM12878)	PU1
		blood (DND-41)	H3K27me3
		muscle (HSMM)	H3K36me3
ENSR00001409462 (promoter flanking region)	1	NHLF	DNase1, H3K27ac
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K4me1, DNase1, H3K9ac
		muscle (HSMM)	DNase1, H3K9ac, H3K27ac, H3K4me2
		monocytes (Monocytes-CD14+)	H3K4me1
		liver (HepG2)	H3K4me1, HNF4A, H3K27ac
		lung (IMR90)	DNase1, H3K4me2
		nervous (NH-A)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
IL6ST	1	ENST00000336909 ENST00000381286 ENST00000381287 ENST00000381293 ENST00000381294 ENST00000381298 ENST00000502326 ENST00000503773 ENST00000506241 ENST00000522633	hsa-miR-296-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC008914.1	upstream gene variant	4185	ENST00000624860	?	ENSP00000485310	1
CTD-2031P19.4	upstream gene variant, downstream gene variant	4110	ENST00000582508	?	?	2
CTD-2031P19.5	upstream gene variant	3217	ENST00000576302	?	?	1
FLJ31104	upstream gene variant, downstream gene variant	549	ENST00000500093	?	?	10
IL6ST	downstream gene variant, upstream gene variant	1285	ENST00000381287	NM_175767.2	ENSP00000370687	7
IL6ST	upstream gene variant	1609	ENST00000423954	?	?	4
IL6ST	downstream gene variant, upstream gene variant	2169	ENST00000381298	NM_002184.3	ENSP00000370698	3
IL6ST	downstream gene variant, upstream gene variant	1321	ENST00000396816	?	ENSP00000463896	10
IL6ST	downstream gene variant, upstream gene variant	1219	ENST00000381286	?	ENSP00000370686	8
IL6ST	downstream gene variant, upstream gene variant	1285	ENST00000336909	?	ENSP00000338799	7
IL6ST	downstream gene variant, upstream gene variant	420	ENST00000503773	?	ENSP00000426224	4
IL6ST	downstream gene variant, upstream gene variant	482	ENST00000502326	?	ENSP00000462158	4
IL6ST	downstream gene variant, upstream gene variant	1148	ENST00000583149	?	?	6
IL6ST	downstream gene variant, upstream gene variant	585	ENST00000506241	?	?	6
IL6ST	downstream gene variant, upstream gene variant	977	ENST00000523039	?	?	3
IL6ST	downstream gene variant, upstream gene variant	1816	ENST00000577363	?	?	6
IL6ST	downstream gene variant, upstream gene variant	1219	ENST00000381293	?	ENSP00000370693	8
IL6ST	upstream gene variant	339	ENST00000522633	?	ENSP00000435399	3
IL6ST	downstream gene variant, upstream gene variant	536	ENST00000381294	NM_001190981.1	ENSP00000370694	8
snoU13	downstream gene variant, upstream gene variant	89	ENST00000459441	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2031P19.5	ENST00000576302	?	?	2
FLJ31104	ENST00000500093	?	?	3
IL6ST	ENST00000381294	NM_001190981.1	ENSP00000370694	22
IL6ST	ENST00000336909	?	ENSP00000338799	24
IL6ST	ENST00000577363	?	?	25
IL6ST	ENST00000583149	?	?	1
IL6ST	ENST00000381286	?	ENSP00000370686	24
IL6ST	ENST00000522633	?	ENSP00000435399	29
IL6ST	ENST00000381298	NM_002184.3	ENSP00000370698	37
IL6ST	ENST00000381293	?	ENSP00000370693	24
IL6ST	ENST00000502326	?	ENSP00000462158	35
IL6ST	ENST00000381287	NM_175767.2	ENSP00000370687	25
IL6ST	ENST00000506241	?	?	5
IL6ST	ENST00000396816	?	ENSP00000463896	12
IL6ST	ENST00000503773	?	ENSP00000426224	36

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL6ST	ENST00000336909	?	ENSP00000338799	2
IL6ST	ENST00000522633	?	ENSP00000435399	1
IL6ST	ENST00000381298	NM_002184.3	ENSP00000370698	2
IL6ST	ENST00000381287	NM_175767.2	ENSP00000370687	3
IL6ST	ENST00000503773	?	ENSP00000426224	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
IL6ST	ENST00000506241	?	1
IL6ST	ENST00000423954	?	1

Locus 273 (rs12121180)

SNiPAcard

Block annotations

Block info

genomic range	chr1:154,282,222-154,283,908
block size	1,687 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.457 [-0.764 – -0.15]	gene(s) hit or close-by	RP11-137P24.5
phastCons	μ = 0.006 [0.001 – 0.011]	eQTL gene(s)	HAX1, IL6R, S100A4, UBAP2L
GERP++	μ = 0.050 [-0.135 – 0.235]	potentially regulated gene(s)	–
CADD score	μ = 4.104 [3.269 – 4.939]	disease gene(s)	HAX1, IL6R

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HAX1	NEUTROPENIA, SEVERE CONGENITAL, 3, AUTOSOMAL RECESSIVE	DECIPHER	MIM:610738
HAX1	NEUTROPENIA, SEVERE CONGENITAL, 3, AUTOSOMAL RECESSIVE	OMIM	MIM:610738
IL6R	INTERLEUKIN 6, SERUM LEVEL OF, QUANTITATIVE TRAIT LOCUS	OMIM	MIM:614752
IL6R	SOLUBLE INTERLEUKIN-6 RECEPTOR, SERUM LEVEL OF, QUANTITATIVE [...]	OMIM	MIM:614689
HAX1	Kostmann syndrome	OrphaNet	OrphaNet:99749

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL6R	ENST00000368485	ILMN_1696394	blood	1.01×10-4 (p-value)	Westra et al.	1
IL6R	ENST00000507256					
IL6R	ENST00000502679					
IL6R	ENST00000515190					
UBAP2L	ENST00000484819	ILMN_1814789	blood	9.02×10-5 (p-value)	Westra et al.	1
UBAP2L	ENST00000361546					
UBAP2L	ENST00000428595					
UBAP2L	ENST00000428931					
HAX1	ENST00000531435	ILMN_1750658	blood	6.09×10-5 (p-value)	Westra et al.	1
HAX1	ENST00000447768					
HAX1	ENST00000459914					
HAX1	ENST00000435087					
HAX1	ENST00000328703					
HAX1	ENST00000457918					
HAX1	ENST00000471326					
HAX1	ENST00000483970					
HAX1	ENST00000532105					
HAX1	ENST00000492550					
HAX1	ENST00000477780					
S100A4	?	ENSG00000196154	esophagus mucosa	5.27×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-137P24.5	upstream gene variant	1030	ENST00000612670	?	?	2

Locus 274 (rs3733159)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,359,810-186,360,409
block size	600 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.686 [-2.582 – -0.791]	gene(s) hit or close-by	FETUB, RP11-573D15.8
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	–
GERP++	μ = -1.260 [-1.57 – -0.949]	potentially regulated gene(s)	–
CADD score	μ = 3.387 [0.978 – 5.796]	disease gene(s)	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001681858 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	downstream gene variant	4493	ENST00000630331	?	?	1
RP11-573D15.8	downstream gene variant	254	ENST00000629106	?	?	1
RP11-573D15.8	downstream gene variant	255	ENST00000626845	?	?	1
RP11-573D15.8	downstream gene variant	4499	ENST00000625741	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FETUB	ENST00000265029	NM_014375.2	ENSP00000265029	2
FETUB	ENST00000435961	?	ENSP00000393851	2
FETUB	ENST00000431018	?	ENSP00000396581	2
FETUB	ENST00000450521	?	ENSP00000404288	2
FETUB	ENST00000382134	?	ENSP00000371569	2
FETUB	ENST00000420570	?	ENSP00000405438	2
FETUB	ENST00000382136	?	ENSP00000371571	2
FETUB	ENST00000488561	?	?	2
RP11-573D15.8	ENST00000625386	?	?	2
RP11-573D15.8	ENST00000628505	?	?	2

RP11-573D15.8	ENST00000626649	?	?	2
RP11-573D15.8	ENST00000428501	?	?	2
RP11-573D15.8	ENST00000628190	?	?	2
RP11-573D15.8	ENST00000630864	?	?	2
RP11-573D15.8	ENST00000628253	?	?	2
RP11-573D15.8	ENST00000625303	?	?	2
RP11-573D15.8	ENST00000630315	?	?	2
RP11-573D15.8	ENST00000629106	?	?	1
RP11-573D15.8	ENST00000455926	?	?	2
RP11-573D15.8	ENST00000626633	?	?	2
RP11-573D15.8	ENST00000630178	?	?	2
RP11-573D15.8	ENST00000626845	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-573D15.8	ENST00000625386	?	1
RP11-573D15.8	ENST00000628505	?	1
RP11-573D15.8	ENST00000626649	?	1
RP11-573D15.8	ENST00000428501	?	1
RP11-573D15.8	ENST00000628190	?	1
RP11-573D15.8	ENST00000630864	?	1
RP11-573D15.8	ENST00000628253	?	1
RP11-573D15.8	ENST00000625303	?	1
RP11-573D15.8	ENST00000630315	?	1
RP11-573D15.8	ENST00000455926	?	1
RP11-573D15.8	ENST00000626633	?	1

Locus 275 (rs7325116)

SNiPAcard

Block annotations

Block info

genomic range	chr13:92,554,173-92,604,445
block size	50,273 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.557 [-2.987 – 0.88]	**gene(s) hit or close-by**	GPC5
phastCons	μ = 0.073 [0 – 0.849]	**eQTL gene(s)**	GPC5
GERP++	μ = -1.026 [-5.52 – 1.62]	**potentially regulated gene(s)**	–
CADD score	μ = 2.599 [0.117 – 5.609]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GPC5	?	ENSG00000179399	subcutaneous adipocytes	1.66×10^{-5} (p-value)	GTEx Portal V6	3
GPC5	ENST00000377067	ILMN_1688886	adipocyte	3.94×10^{-7} (p-value)	MuTHER consortium	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPC5	ENST00000377067	NM_004466.4	ENSP00000366267	13

Locus 276 (rs259842)

SNiPAcard

Block annotations

Block info

genomic range	chr2:180,738,840-180,739,450
block size	611 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.499 [0.068 – 0.929]	**gene(s) hit or close-by**	–
phastCons	μ = 0.102 [0 – 0.203]	**eQTL gene(s)**	ZNF385B
GERP++	μ = 0.581 [-0.0672 – 1.23]	**potentially regulated gene(s)**	–
CADD score	μ = 5.518 [4.902 – 6.135]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ZNF385B	?	ENSG00000144331	tibial artery	2.09×10^{-6} (p-value)	GTEx Portal V6	2

Locus 278 (rs6603134)

SNiPAcard

Block annotations

Block info

genomic range	chr19:8,105,520-8,107,703
block size	2,184 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.185 [-2.036 – 0.169]	**gene(s) hit or close-by**	ELAVL1
phastCons	μ = 0.012 [0.002 – 0.024]	**eQTL gene(s)**	–
GERP++	μ = -0.042 [-0.167 – 0]	**potentially regulated gene(s)**	–
CADD score	μ = 1.701 [0.781 – 3.285]	**disease gene(s)**	–

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ELAVL1	ENST00000351593	?	ENSP00000264073	4

Locus 279 (rs9302690)

SNiPAcard

Block annotations

Block info

genomic range	chr16:57,442,191-57,444,002
block size	1,812 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.894 [-0.195 – 2.306]	gene(s) hit or close-by	CCL17
phastCons	μ = 0.060 [0.006 – 0.182]	eQTL gene(s)	–
GERP++	μ = 0.412 [-1.03 – 0.913]	potentially regulated gene(s)	–
CADD score	μ = 8.514 [5.131 – 13.24]	disease gene(s)	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001633924 (CTCF binding site)	2	cervix (HeLa-S3)	CTCF, DNase1
		embryonic stem cell (H1ESC)	Rad21
		HSMMtube	DNase1, H3K27me3
		liver (HepG2)	Rad21, CTCF, H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	CTCF
ENSR00001505514 (enhancer)	2	embryonic stem cell (H1ESC)	Rad21
		HSMMtube	DNase1, H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	CTCF, DNase1
		liver (HepG2)	Rad21, CTCF, H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	H3K4me1
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL17	upstream gene variant	3779	ENST00000616880	?	ENSP00000480147	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL17	ENST00000219244	NM_002987.2	ENSP00000219244	4

Locus 280 (rs17817077)

SNiPAcard

Block annotations

Block info

genomic range	chr18:72,209,543-72,209,543
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.179	gene(s) hit or close-by	CNDP1
phastCons	0	eQTL gene(s)	CNDP1
GERP++	1.25	potentially regulated gene(s)	–
CADD score	5.184	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CNDP1	?	ENSG00000150656	testis	1.15×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001645555 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		HSMMtube	H3K27me3
		liver (HepG2)	HNF4G, PolII, DNase1, Cmyc, H3K27ac, H3K9ac, HNF4A, H3K4me2, H3K4me1
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNDP1	ENST00000358821	NM_032649.5	ENSP00000351682	1
CNDP1	ENST00000582365	?	ENSP00000462096	1
CNDP1	ENST00000585136	?	?	1

Locus 281 (rs8049040)

SNiPAcard

Block annotations

Block info

genomic range	chr16:71,465,613-71,506,983
block size	41,371 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.134 [-3.461 – 0.421]	**gene(s) hit or close-by**	AC010547.9, RP11-510M2.4, RP11-510M2.8, ZNF19
phastCons	μ = 0.002 [0 – 0.01]	**eQTL gene(s)**	AC010547.9, MARVELD3, ZNF23
GERP++	μ = -1.694 [-9.66 – 1.52]	**potentially regulated gene(s)**	–
CADD score	μ = 1.501 [0.025 – 4.134]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MARVELD3	?	ENSG00000140832	sun exposed skin	6.69×10^{-8} (p-value)	GTEx Portal V6	6
MARVELD3	?	ENSG00000140832	thyroid	1.68×10^{-8} (p-value)	GTEx Portal V6	6
ZNF23	ENST00000358700	ILMN_1720027	blood	2.93×10^{-6} (p-value)	Westra et al.	2
ZNF23	ENST00000539742					
ZNF23	ENST00000428724					
AC010547.9	ENST00000561908					
ZNF23	ENST00000357254					
ZNF23	ENST00000393539					
ZNF23	ENST00000576258					
ZNF23	ENST00000564528					
ZNF23	ENST00000497160					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-510M2.8	downstream gene variant	3122	ENST00000571789	?	?	3
ZNF19	downstream gene variant	3083	ENST00000565541	?	ENSP00000454549	2
ZNF19	downstream gene variant	1304	ENST00000565100	?	ENSP00000462810	2
ZNF19	downstream gene variant	503	ENST00000288177	NM_006961.3	ENSP00000288177	2
ZNF19	downstream gene variant	1669	ENST00000569717	?	ENSP00000456143	2
ZNF19	downstream gene variant	1299	ENST00000565637	?	ENSP00000454864	2
ZNF19	downstream gene variant	3114	ENST00000568815	?	ENSP00000455796	2
ZNF19	downstream gene variant	1730	ENST00000564230	?	ENSP00000458105	2
ZNF19	downstream gene variant	4806	ENST00000561469	?	ENSP00000462021	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC010547.9	ENST00000561908	?	ENSP00000463741	2
RP11-510M2.4	ENST00000561754	?	?	1
RP11-510M2.4	ENST00000572770	?	?	1
ZNF19	ENST00000567225	?	ENSP00000462566	2

Locus 282 (rs2749534)

SNiPAcard

Block annotations

Block info

genomic range	chr14:94,809,760-94,841,641
block size	31,882 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.175 [-4.333 – 0.735]	gene(s) hit or close-by	SERPINA1, SERPINA2
phastCons	μ = 0.016 [0 – 0.343]	eQTL gene(s)	–
GERP++	μ = -0.914 [-4.49 – 2.08]	potentially regulated gene(s)	SERPINA1, SERPINA1, SERPINA9
CADD score	μ = 2.358 [0.063 – 7.213]	disease gene(s)	SERPINA1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SERPINA1	ALPHA-1-ANTITRYPSIN DEFICIENCY	OMIM	MIM:613490
SERPINA1	ALPHA-1-ANTITRYPSIN DEFICIENCY	OrphaNet	OrphaNet:60
SERPINA1	Hemorrhagic disease due to alpha-1 antitrypsin Pittsburgh mutation	OrphaNet	OrphaNet:178396

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SERPINA2	start lost	ENST00000553483	?	ENSP00000486888	I/M	atA/atG	?	benign	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000172474	1	ENCP00000019874	SERPINA1 SERPINA1
ENCE00000172475	1	ENCP00000019874	SERPINA1 SERPINA1
ENCE00000172476	1	ENCP00000019874	SERPINA1 SERPINA1
		ENCP00000019876	SERPINA9

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001459896 (promoter flanking region)	1	NHLF	DNase1, H3K27ac
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K4me1, H3K27ac, DNase1
		breast (HMEC)	H3K4me2, H3K27ac, DNase1

		monocytes (Monocytes-CD14+)	DNase1
		liver (HepG2)	H3K27ac, H3K4me2, H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1, H3K4me2
		A549	DNase1, H3K27me3
		skin (NHEK)	DNase1
ENSR00001624849 (enhancer)	2	cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	H3K4me1
ENSR00000422992 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K4me1, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K27me3
		liver (HepG2)	H3K4me2, H3K4me1, Jund, FOSL2, DNase1
		blood (GM12878)	H3K27ac, NFKB, DNase1, H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K27me3
		skin (NHEK)	H3K27me3, H3K4me1, DNase1
ENSR00001459901 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	Rad21, Nrsf, CTCF, Yy1, DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac
		blood (K562)	CTCF, H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K4me1, H3K27ac
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	CTCF, H3K4me1, Rad21, PolII
		lung (IMR90)	DNase1
		A549	CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SERPINA1	downstream gene variant	1443	ENST00000437397	NM_001127706.1, NM_001127702.1, NM_001127707.1	ENSP00000408474	2
SERPINA1	downstream gene variant	1443	ENST00000440909	?	ENSP00000390299	2
SERPINA1	downstream gene variant	1443	ENST00000448921	NM_001127705.1, NM_001127701.1, NM_001002236.2, NM_001127704.1, NM_001127703.1	ENSP00000416066	2
SERPINA1	downstream gene variant	1443	ENST00000355814	NM_001127700.1, NM_001002235.2	ENSP00000348068	2
SERPINA1	downstream gene variant	3145	ENST00000449399	?	ENSP00000416354	1
SERPINA1	downstream gene variant	3085	ENST00000404814	?	ENSP00000385960	1
SERPINA1	downstream gene variant	3072	ENST00000393088	?	ENSP00000376803	1
SERPINA1	downstream gene variant	3045	ENST00000393087	NM_000295.4	ENSP00000376802	1
SERPINA1	downstream gene variant	3809	ENST00000402629	?	ENSP00000386094	1
SERPINA1	downstream gene variant	3069	ENST00000489769	?	ENSP00000451525	1
SERPINA2	downstream gene variant, upstream gene variant	53	ENST00000553483	?	ENSP00000486888	8

Locus 283 (rs17200067)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,370,722-31,376,989
block size	6,268 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.077 [-0.057 – 0.212]	**gene(s) hit or close-by**	HCP5, MICA, Y_RNA
phastCons	μ = 0.117 [0.001 – 0.232]	**eQTL gene(s)**	–
GERP++	μ = -0.392 [-1.04 – 0.257]	**potentially regulated gene(s)**	–
CADD score	μ = 5.005 [4.24 – 5.771]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487837 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, PolII, TAF1, H3K4me2, SP1, USF1, H3K4me3, DNase1
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K27ac, H3K4me2, H3K9ac
		blood (K562)	H3K27ac, Max, E2F6, H3K9ac, USF1, H3K79me2, H2AZ, Cfos, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	USF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1, TAF1, H3K79me2, PolII
		blood (GM12878)	PolII, H2AZ, DNase1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K79me2, H3K4me2, H3K4me3, H3K9ac, DNase1
		nervous (NH-A)	H3K9ac, H4K20me1, H3K4me2, H3K4me3, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me1, H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3, H3K4me2, H3K4me1
		endothelium (HUVEC)	H3K36me3, H3K4me2, H3K4me3, PolII, DNase1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MICA	upstream gene variant	634	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	upstream gene variant	727	ENST00000421350	?	ENSP00000402410	1
Y_RNA	upstream gene variant	695	ENST00000362462	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	2
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	2

MICA	ENST00000421350	?	ENSP00000402410	1

Locus 284 (rs3213636)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,475,131-34,475,131
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-1.953	gene(s) hit or close-by	CTB-91J4.1
phastCons	0	eQTL gene(s)	–
GERP++	-2.76	potentially regulated gene(s)	–
CADD score	0.344	disease gene(s)	–

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTB-91J4.1	ENST00000617914	?	?	1

Locus 285 (rs2523710)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,450,909-31,450,909
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.625	gene(s) hit or close-by	–
phastCons	0.005	eQTL gene(s)	AGER, DXO, HLA-B, LST1, MICB, PSMB8
GERP++	0.207	potentially regulated gene(s)	–
CADD score	5.078	disease gene(s)	PSMB8, HLA-B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PSMB8	AUTOINFLAMMATION, LIPODYSTROPHY, AND DERMATOSIS SYNDROME	OMIM	MIM:256040
PSMB8	Nakajo syndrome	DECIPHER	MIM:256040
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117
PSMB8	CANDLE syndrome	OrphaNet	OrphaNet:325004
PSMB8	JMP syndrome	OrphaNet	OrphaNet:324999
PSMB8	Nakajo-Nishimura syndrome	OrphaNet	OrphaNet:2615

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LST1	ENST00000339530	ILMN_1718936	blood	1.37×10-6 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
LST1	ENST00000376099	ILMN_1688373	blood	4.99×10-5 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					

LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					
LST1	ENST00000376089					
LST1	ENST00000396112					
LST1	ENST00000418507					
LST1	ENST00000376096					
LST1	ENST00000490742					
LST1	ENST00000211921					
LST1	ENST00000460834					
MICB	ENST00000538442	ILMN_1708006	skin	4.40×10-8 (p-value)	MuTHER consortium	1
MICB	ENST00000399150					
MICB	ENST00000252229					
MICB	?	ENSG00000204516	lung	3.70×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	transformed fibroblasts	3.43×10-7 (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	tibial artery	3.49×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	tibial artery	8.97×10-9 (p-value)	GTEx Portal V6	1
DXO	ENST00000474587	ILMN_1726990	monocyte	4.95×10-7 (p-value)	Fairfax et al.	1
DXO	ENST00000473976					
DXO	ENST00000491327					
DXO	ENST00000480240					
DXO	ENST00000477826					
DXO	ENST00000498357					
DXO	ENST00000337523					
DXO	ENST00000460058					
DXO	ENST00000492946					
DXO	ENST00000375349					
DXO	ENST00000375356					
DXO	ENST00000487914					
DXO	ENST00000478221					
DXO	ENST00000485557					
AGER	ENST00000375070	ILMN_2257015	monocyte	2.29×10-4 (p-value)	Fairfax et al.	1
AGER	ENST00000375056					
AGER	ENST00000450110					
AGER	ENST00000438221					
AGER	ENST00000375065					
AGER	ENST00000375076					
AGER	ENST00000473619					
AGER	ENST00000484849					
AGER	ENST00000488669					
AGER	ENST00000375069					
AGER	ENST00000375055					
PSMB8	ENST00000490613	ILMN_1747195	monocyte	1.27×10-5 (p-value)	Fairfax et al.	1
PSMB8	ENST00000374881					
PSMB8	ENST00000484003					
PSMB8	ENST00000395339					
PSMB8	ENST00000374882					
MICB	?	ENSG00000204516	esophagus mucosa	1.80×10-10 (p-value)	GTEx Portal V6	1

MICB	?	ENSG00000204516	cerebellar hemisphere	1.90×10-7 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	sun exposed skin	1.49×10-6 (p-value)	GTEx Portal V6	1

Locus 286 (rs1888560)

SNiPAcard

Block annotations

Block info

genomic range	chr14:21,164,245-21,164,245
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.241	gene(s) hit or close-by	ANG, RNASE4, RP11-903H12.3, RP11-903H12.5
phastCons	0	eQTL gene(s)	ANG, RNASE4, RP11-903H12.5
GERP++	0.639	potentially regulated gene(s)	–
CADD score	5.488	disease gene(s)	ANG

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ANG	AMYOTROPHIC LATERAL SCLEROSIS 9	OMIM	MIM:611895
ANG	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-903H12.5	ENST00000553909	ILMN_1776602	adipocyte	9.59×10-6 (p-value)	MuTHER consortium	1
RNASE4	ENST00000555597					
RNASE4	ENST00000397995					
RNASE4	ENST00000555835					
RP11-903H12.5	ENST00000553909	ILMN_2408572	blood	1.68×10-6 (p-value)	Westra et al.	1
ANG	ENST00000554073					
RNASE4	ENST00000397995					
RNASE4	ENST00000555597					
RNASE4	ENST00000555835					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANG	downstream gene variant	1900	ENST00000397990	NM_001097577.2	ENSP00000381077	1
ANG	downstream gene variant	1900	ENST00000336811	NM_001145.4	ENSP00000336762	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)

ANG	ENST00000554073	?	?	1
RNASE4	ENST00000397995	?	ENSP00000381081	1
RNASE4	ENST00000555597	NM_194431.2	ENSP00000451624	1
RNASE4	ENST00000555835	NM_001282193.1, NM_001282192.1, NM_002937.4	ENSP00000452245	1
RP11-903H12.3	ENST00000554286	?	?	1
RP11-903H12.5	ENST00000553909	?	ENSP00000477037	1

Locus 287 (rs4655537)

SNiPAcard

Block annotations

Block info

genomic range	chr1:66,054,995-66,058,801
block size	3,807 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.479 [-0.627 – -0.331]	gene(s) hit or close-by	LEPR
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	–
GERP++	μ = 0.661 [-0.258 – 1.58]	potentially regulated gene(s)	–
CADD score	μ = 3.693 [2.605 – 4.781]	disease gene(s)	LEPR

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
LEPR	LEPTIN RECEPTOR DEFICIENCY	OMIM	MIM:614963
LEPR	Obesity due to leptin receptor gene deficiency	OrphaNet	OrphaNet:179494

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	2
LEPR	ENST00000344610	NM_001198688.1	ENSP00000340884	2
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	2
LEPR	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	2
LEPR	ENST00000616738	NM_001198689.1	ENSP00000483390	2
LEPR	ENST00000406510	?	ENSP00000384025	2
LEPR	ENST00000462765	?	?	2
LEPR	ENST00000371058	?	ENSP00000360097	2

Locus 288 (rs3856930)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,431,070-186,458,322
block size	27,253 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.023 [-0.404 – 0.53]	gene(s) hit or close-by	KNG1, RP11-573D15.8, RP11-795A2.3
phastCons	μ = 0.002 [0 – 0.008]	eQTL gene(s)	–
GERP++	μ = -0.121 [-1.03 – 0.6]	potentially regulated gene(s)	–
CADD score	μ = 2.264 [0.571 – 4.553]	disease gene(s)	KNG1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
bradykinin, des-arg(9)	1.37×10^{-12}	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link
KNG1	HIGH MOLECULAR WEIGHT KININOGEN DEFICIENCY	OMIM	MIM:228960
KNG1	Congenital high-molecular-weight kininogen deficiency	OrphaNet	OrphaNet:483

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KNG1	upstream gene variant	4067	ENST00000447445	NM_001166451.1	ENSP00000396025	1
KNG1	upstream gene variant	4050	ENST00000265023	NM_001102416.2	ENSP00000265023	1
KNG1	upstream gene variant	3995	ENST00000287611	NM_000893.3	ENSP00000287611	1
RP11-573D15.8	downstream gene variant	3968	ENST00000599314	?	?	1
RP11-573D15.8	downstream gene variant	1167	ENST00000354642	?	?	1
RP11-573D15.8	downstream gene variant	721	ENST00000629126	?	?	3
RP11-573D15.8	downstream gene variant	1379	ENST00000629734	?	?	3
RP11-573D15.8	upstream gene variant	2714	ENST00000626649	?	?	1
RP11-573D15.8	downstream gene variant	678	ENST00000626306	?	?	2
RP11-573D15.8	downstream gene variant	4417	ENST00000627551	?	?	1
RP11-573D15.8	downstream gene variant	1112	ENST00000628020	?	?	3
RP11-573D15.8	downstream gene variant	3189	ENST00000631299	?	?	2
RP11-573D15.8	downstream gene variant	3191	ENST00000609726	?	?	1
RP11-573D15.8	downstream gene variant	3191	ENST00000626520	?	?	1
RP11-573D15.8	downstream gene variant	3172	ENST00000596632	?	?	1
RP11-573D15.8	downstream gene variant	3191	ENST00000626978	?	?	1

RP11-573D15.8	downstream gene variant	3191	ENST00000625690	?	?	1
RP11-573D15.8	downstream gene variant	3175	ENST00000596329	?	?	1
RP11-573D15.8	downstream gene variant	1288	ENST00000628831	?	?	3
RP11-573D15.8	downstream gene variant	3191	ENST00000609652	?	?	1
RP11-573D15.8	downstream gene variant	3191	ENST00000628291	?	?	1
RP11-573D15.8	downstream gene variant	3191	ENST00000625869	?	?	1
RP11-573D15.8	downstream gene variant	3170	ENST00000626586	?	?	1
RP11-573D15.8	downstream gene variant	3191	ENST00000629201	?	?	1
RP11-573D15.8	downstream gene variant	3191	ENST00000628928	?	?	1
RP11-573D15.8	downstream gene variant	3191	ENST00000629954	?	?	1
RP11-795A2.3	downstream gene variant	4707	ENST00000413467	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KNG1	ENST00000287611	NM_000893.3	ENSP00000287611	4
KNG1	ENST00000447445	NM_001166451.1	ENSP00000396025	4
KNG1	ENST00000265023	NM_001102416.2	ENSP00000265023	4
RP11-573D15.8	ENST00000625386	?	?	5
RP11-573D15.8	ENST00000628190	?	?	5
RP11-573D15.8	ENST00000627919	?	?	5
RP11-573D15.8	ENST00000627268	?	?	4
RP11-573D15.8	ENST00000630178	?	?	5
RP11-573D15.8	ENST00000630331	?	?	5
RP11-573D15.8	ENST00000626633	?	?	5
RP11-573D15.8	ENST00000628858	?	?	4
RP11-573D15.8	ENST00000628831	?	?	1
RP11-573D15.8	ENST00000628601	?	?	4
RP11-573D15.8	ENST00000627830	?	?	5
RP11-573D15.8	ENST00000626151	?	?	5
RP11-573D15.8	ENST00000626306	?	?	2
RP11-573D15.8	ENST00000629106	?	?	5
RP11-573D15.8	ENST00000628020	?	?	1
RP11-573D15.8	ENST00000628505	?	?	5
RP11-573D15.8	ENST00000628728	?	?	5
RP11-573D15.8	ENST00000630864	?	?	5
RP11-573D15.8	ENST00000630726	?	?	1
RP11-573D15.8	ENST00000627015	?	?	4
RP11-573D15.8	ENST00000625741	?	?	5
RP11-573D15.8	ENST00000354642	?	?	4

RP11-573D15.8	ENST00000627551	?	?	4
RP11-573D15.8	ENST00000599314	?	?	4
RP11-573D15.8	ENST00000625303	?	?	5
RP11-573D15.8	ENST00000627469	?	?	5
RP11-573D15.8	ENST00000625710	?	?	4
RP11-573D15.8	ENST00000626845	?	?	5
RP11-573D15.8	ENST00000628253	?	?	5
RP11-573D15.8	ENST00000629126	?	?	1
RP11-573D15.8	ENST00000625839	?	?	5
RP11-573D15.8	ENST00000631299	?	?	1
RP11-573D15.8	ENST00000629451	?	?	5
RP11-573D15.8	ENST00000629734	?	?	1

Locus 289 (rs6665631)

SNiPAcard

Block annotations

Block info

genomic range	chr1:8,971,030-8,980,813
block size	9,784 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.162 [-1.676 – 0.529]	**gene(s) hit or close-by**	RP3-477M7.5, RP3-477M7.6
phastCons	μ = 0.022 [0 – 0.112]	**eQTL gene(s)**	–
GERP++	μ = -0.042 [-0.804 – 0.774]	**potentially regulated gene(s)**	–
CADD score	μ = 2.705 [0.013 – 8.856]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000530162 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, DNase1, H3K27ac
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	Max, H3K27me3
		breast (HMEC)	H3K27ac, H3K4me1, H3K4me2
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	USF1, H3K4me1
		nervous (NH-A)	DNase1
ENSR00001576724 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	CTCF, Rad21, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, FOXA1, CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	DNase1, CTCF
		A549	CTCF
ENSR00000074592 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	Rad21, H3K27me3, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, FOXA1, CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF

cervix (HeLa-S3)	CTCF
monocytes (Monocytes-CD14+)	CTCF
endothelium (HUVEC)	DNase1, CTCF
A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP3-477M7.5	upstream gene variant	3227	ENST00000424862	?	?	2
RP3-477M7.6	downstream gene variant	1067	ENST00000431968	?	?	6

Locus 290 (rs6556937)

SNiPAcard

Block annotations

Block info

genomic range	chr5:95,985,282-95,985,282
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.268	gene(s) hit or close-by	CAST
phastCons	0.253	eQTL gene(s)	CAST, CTC-506B8.1, ERAP1
GERP++	2.41	potentially regulated gene(s)	–
CADD score	8.833	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP1	?	ENSG00000164307	pancreas	5.79×10^{-6} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	lung	4.26×10^{-10} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	lung	8.60×10^{-18} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	transformed fibroblasts	7.47×10^{-10} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	transformed fibroblasts	8.00×10^{-15} (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	blood	3.77×10^{-7} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	blood	2.55×10^{-13} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	blood	1.27×10^{-20} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	breast	6.66×10^{-7} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	tibial artery	4.40×10^{-14} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	tibial artery	1.14×10^{-13} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	thyroid	2.03×10^{-7} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	thyroid	7.36×10^{-11} (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	skeletal muscle	3.16×10^{-6} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	EBV lymphocytes	1.74×10^{-6} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	EBV lymphocytes	2.48×10^{-6} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	ovary	2.54×10^{-8} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	ovary	1.78×10^{-7} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	aorta	1.54×10^{-6} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	aorta	7.30×10^{-9} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	subcutaneous adipocytes	2.85×10^{-10} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	subcutaneous adipocytes	5.43×10^{-12} (p-value)	GTEx Portal V6	1

?	?	ILMN_1717234	monocyte	1.20×10-13 (p-value)	Zeller et al.	1
CTC-506B8.1	?	ENSG00000249180	visceral adipocytes	1.16×10-8 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	visceral adipocytes	2.65×10-12 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	sigmoid colon	1.58×10-7 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	tibial nerve	4.62×10-10 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	tibial nerve	2.36×10-20 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	coronary artery	8.08×10-8 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	coronary artery	4.82×10-7 (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	esophagus mucosa	1.48×10-5 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	adrenal gland	9.17×10-7 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	cerebellum	2.42×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001285945 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1, DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CAST	ENST00000505143	?	ENSP00000422957	1

Locus 291 (rs783150)

SNiPAcard

Block annotations

Block info

genomic range	chr6:161,177,237-161,252,795
block size	75,559 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.106 [-1.896 – -0.595]	gene(s) hit or close-by	PLG, RP1-81D8.5
phastCons	μ = 0.105 [0 – 0.276]	eQTL gene(s)	–
GERP++	μ = 0.088 [-0.465 – 0.687]	potentially regulated gene(s)	AGPAT4, PLG, RP1-81D8.4
CADD score	μ = 3.338 [1.084 – 4.486]	disease gene(s)	PLG

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Lipoproteins	1.04×10-11	dbGaP	pha003079	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
PLG	PLASMINOGEN DEFICIENCY, TYPE ILIGNEOUS CONJUNCTIVITIS, [...]	OMIM	MIM:217090	OMIM
PLG	Hypoplasminogenemia	OrphaNet	OrphaNet:722	orphanet
PLG	Ligneous conjunctivitis	OrphaNet	OrphaNet:97231	orphanet

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000468471	1	ENCP00000050551	PLG
		ENCP00000050546	RP1-81D8.4
ENCE00000468646	1	ENCP00000050568	AGPAT4

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PLG	downstream gene variant	2899	ENST00000308192	NM_000301.3	ENSP00000308938	1
PLG	downstream gene variant	2890	ENST00000461414	?	?	1
PLG	downstream gene variant	3952	ENST00000467466	?	?	1
RP1-81D8.5	upstream gene variant	2721	ENST00000612111	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP1-81D8.5	ENST00000612111	?	?	1

Locus 292 (rs753307)

SNiPAcard

Block annotations

Block info

genomic range	chr19:49,504,260-49,524,220
block size	19,961 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.621 [-5.773 – 4.333]	gene(s) hit or close-by	CGB, CTB-60B18.10, CTB-60B18.6, LHB, MIR6798, NTF6A, RUVBL2
phastCons	μ = 0.089 [0 – 1]	eQTL gene(s)	CGB, CTB-60B18.10, RUVBL2
GERP++	μ = -1.376 [-8.36 – 3.27]	potentially regulated gene(s)	NUCB1
CADD score	μ = 4.534 [0.004 – 14.25]	disease gene(s)	LHB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
LHB	LUTEINIZING HORMONE, BETA POLYPEPTIDE	OMIM	MIM:152780
LHB	Leydig cell hypoplasia due to LHB deficiency	OrphaNet	OrphaNet:325448

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CGB	?	ENSG00000104827	pancreas	3.53×10^{-11} (p-value)	GTEx Portal V6	12
CTB-60B18.10	?	ENSG00000268655	testis	3.15×10^{-8} (p-value)	GTEx Portal V6	12
RUVBL2	?	ENSG00000183207	tibial artery	7.13×10^{-7} (p-value)	GTEx Portal V6	9

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000231691	1	ENCP00000024577	NUCB1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000217422 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CGB	downstream gene variant	1906	ENST00000357383	NM_000737.3	ENSP00000349954	1
CTB-60B18.10	downstream gene variant, upstream gene variant	928	ENST00000600007	?	?	6
CTB-60B18.6	downstream gene variant	2183	ENST00000604577	?	ENSP00000474022	1
CTB-60B18.6	downstream gene variant	1990	ENST00000591656	?	ENSP00000466140	1
LHB	downstream gene variant, upstream gene variant	46	ENST00000391869	?	ENSP00000375742	7
LHB	downstream gene variant, upstream gene variant	7	ENST00000221421	NM_000894.2	ENSP00000221421	7
MIR6798	upstream gene variant, downstream gene variant	787	ENST00000612887	?	?	5
NTF6A	downstream gene variant	4498	ENST00000591175	?	?	1
RUVBL2	downstream gene variant	419	ENST00000594017	?	?	5
RUVBL2	upstream gene variant, downstream gene variant	24	ENST00000595002	?	?	9
RUVBL2	upstream gene variant, downstream gene variant	323	ENST00000627972	?	ENSP00000486242	5
RUVBL2	downstream gene variant	877	ENST00000593570	?	ENSP00000469488	4
RUVBL2	downstream gene variant	79	ENST00000594338	?	?	4
RUVBL2	downstream gene variant	81	ENST00000601968	?	ENSP00000471524	4
RUVBL2	downstream gene variant	1441	ENST00000596837	?	ENSP00000469189	4
RUVBL2	downstream gene variant	603	ENST00000595090	NM_006666.1	ENSP00000473172	4
RUVBL2	downstream gene variant	44	ENST00000596247	?	ENSP00000471538	4
RUVBL2	downstream gene variant	4427	ENST00000595811	?	ENSP00000469760	1
RUVBL2	downstream gene variant	47	ENST00000221413	?	ENSP00000221413	4
RUVBL2	downstream gene variant	1639	ENST00000598768	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
LHB	ENST00000391869	?	ENSP00000375742	P	ccA/ccC	1
LHB	ENST00000221421	NM_000894.2	ENSP00000221421	P	ccA/ccC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LHB	ENST00000391869	?	ENSP00000375742	1
LHB	ENST00000221421	NM_000894.2	ENSP00000221421	1
RUVBL2	ENST00000594017	?	?	2
RUVBL2	ENST00000594338	?	?	7
RUVBL2	ENST00000596837	?	ENSP00000469189	1
RUVBL2	ENST00000596247	?	ENSP00000471538	7
RUVBL2	ENST00000601968	?	ENSP00000471524	7
RUVBL2	ENST00000595811	?	ENSP00000469760	1
RUVBL2	ENST00000221413	?	ENSP00000221413	7
RUVBL2	ENST00000627972	?	ENSP00000486242	6
RUVBL2	ENST00000595090	NM_006666.1	ENSP00000473172	7

RUVBL2	ENST00000593570	?	ENSP00000469488	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RUVBL2	ENST00000595090	NM_006666.1	ENSP00000473172	1

Locus 293 (rs6036478)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,569,186-23,622,880
block size	53,695 bp
variant count	118 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.764 [-5.092 – 1.089]	gene(s) hit or close-by	CST3, CST9, RP11-218C14.2, RP11-218C14.8
phastCons	μ = 0.020 [0 – 0.388]	eQTL gene(s)	CST3, CST9, NAPB, RP11-218C14.5, RP11-218C14.8
GERP++	μ = -0.828 [-5.85 – 1.96]	potentially regulated gene(s)	CST11, CST3, CST5, CST8, CSTL1, CSTP1, GGTLC1, GZF1, POM121L3P, RP11-218C14.2
CADD score	μ = 2.443 [0.013 – 10.32]	disease gene(s)	CST3

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Plasma cystastin c levels in acute coronary syndrome	$<1.00\times10^{-26}$	GWAS Catalog	24952865	1
Chronic kidney disease	$<2.00\times10^{-138}$	GWAS Catalog	20383146	1
Cystatin C	$<9.00\times10^{-9}$	GWAS Catalog	17903292	1
chiro-inositol	6.50×10^{-5}	Metabolomics GWAS Server	24816252	1
Blood pressure	7.50×10^{-5}	dbGaP	pha002898	1
Cystatins	8.45×10^{-9}	dbGaP	pha001330	1
Cystatins	1.19×10^{-7}	dbGaP	pha001328	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Age-related macular degeneration 11	pathogenic	ClinVar	RCV000005989.1	1
?	HGMD curated	HGMD	CR040863	1
?	HGMD curated	HGMD	CR040862	1
MACULAR DEGENERATION, AGE-RELATED, 11	OMIM curated	OMIM	MIM:604312	1

Disease gene annotation

gene	trait	source DB	source entry/link
CST3	CEREBRAL AMYLOID ANGIOPATHY, CST3-RELATED	OMIM	MIM:105150
CST3	MACULAR DEGENERATION, AGE-RELATED, 11	OMIM	MIM:611953
CST3	Hereditary cerebral hemorrhage with amyloidosis, Icelandic type	OrphaNet	OrphaNet:100008

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CST3	missense variant	ENST00000398411	NM_001288614.1	ENSP00000381448	T/A	Acc/Gcc	?	?	1

CST3	missense variant	ENST00000376925	NM_000099.3	ENSP00000366124	T/A	Acc/Gcc	?	?	1
CST3	missense variant, splice acceptor variant	ENST00000398409	?	ENSP00000381446	T/A	Acc/Gcc	?	?	2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST3	ENST00000398411	ILMN_1800354	blood	1.17×10^{-26} (p-value)	MuTHER consortium	44
CST3	ENST00000376925					
CST3	ENST00000398409					
NAPB	?	ENSG00000125814	muscularis mucosae	5.77×10^{-7} (p-value)	GTEx Portal V6	26
CST9	?	ENSG00000173335	muscularis mucosae	1.39×10^{-5} (p-value)	GTEx Portal V6	5
CST3	?	ENSG00000101439	lung	1.52×10^{-8} (p-value)	GTEx Portal V6	108
CST3	?	ENSG00000101439	sigmoid colon	3.39×10^{-7} (p-value)	GTEx Portal V6	68
CST3	?	ENSG00000101439	blood	2.38×10^{-8} (p-value)	GTEx Portal V6	111
RP11-218C14.8	?	ENSG00000270001	cerebellar hemisphere	2.39×10^{-7} (p-value)	GTEx Portal V6	104
CST3	?	ENSG00000101439	testis	4.07×10^{-18} (p-value)	GTEx Portal V6	113
RP11-218C14.8	?	ENSG00000270001	testis	2.23×10^{-7} (p-value)	GTEx Portal V6	112
RP11-218C14.8	?	ENSG00000270001	aorta	1.58×10^{-7} (p-value)	GTEx Portal V6	108
CST3	?	ENSG00000101439	tibial nerve	3.46×10^{-10} (p-value)	GTEx Portal V6	110
CST3	ENST00000398411	ILMN_1800354	monocyte	1.81×10^{-25} (p-value)	Zeller et al.	13
CST3	ENST00000376925					
CST3	ENST00000398409					
CST3	ENST00000398411	ILMN_1800354	monocyte	1.49×10^{-14} (p-value)	Fairfax et al.	9
CST3	ENST00000376925					
CST3	ENST00000398409					
CST3	?	ENSG00000101439	atrial appendage	3.21×10^{-6} (p-value)	GTEx Portal V6	3
CST3	?	ENSG00000101439	skeletal muscle	2.70×10^{-6} (p-value)	GTEx Portal V6	9
RP11-218C14.5	?	ENSG00000225056	hypothalamus	7.71×10^{-7} (p-value)	GTEx Portal V6	17
RP11-218C14.8	?	ENSG00000270001	tibial artery	1.20×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	14	no	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000031087		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000290793	1	ENCP00000031085	RP11-218C14.2
ENCE00000290764	1	ENCP00000031095	CST5
ENCE00000290765	1	ENCP00000031095	CST5
ENCE00000290698	1	ENCP00000031098	GGTLC1
		ENCP00000031079	CST11
		ENCP00000031078	CSTL1
		ENCP00000031081	CST8
		ENCP00000031096	CSTP1
ENCE00000290858	1	ENCP00000031097	POM121L3P GGTLC1
ENCE00000290678	1	ENCP00000031087	CST3
		ENCP00000031074	GZF1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001021906 (enhancer)	3	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	H3K27me3
		liver (HepG2)	H3K4me1, DNase1
		blood (K562)	PU1, H2AZ
		A549	H3K27me3
ENSR00001664573 (open chromatin region)	2	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	H3K27me3
		A549	H3K27me3
ENSR00000649134 (CTCF binding site)	3	embryonic stem cell (H1ESC)	CTCF, Yy1, DNase1, Rad21
		HSMMtube	CTCF, H3K27me3, DNase1
		blood (K562)	CTCF, Rad21, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	DNase1, CTCF
		liver (HepG2)	Rad21, CTCF, H3K27me3, DNase1
		blood (GM12878)	CTCF, Rad21, DNase1
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF
ENSR00001664575 (open chromatin region)	2	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
ENSR00001448098 (promoter flanking region)	12	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K27me3, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H3K27ac, H3K4me2, DNase1, H3K9ac
		liver (HepG2)	FOSL2, Jund, H3K4me3, H3K4me1, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	H3K4me2
		blood (GM12878)	EBF1
		nervous (NH-A)	DNase1, H3K4me2, H3K9ac
		skin (NHEK)	H3K4me2, H3K9ac, H3K27ac, DNase1
		NHLF	H3K27ac, DNase1
		Osteobl	H3K4me2, H3K27ac

		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, H3K9ac, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K4me2, Jund
		monocytes (Monocytes-CD14+)	H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, Cjun, H3K4me2, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00000399197 (promoter flanking region)	4	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		liver (HepG2)	H3K4me1
		skin (NHEK)	DNase1
ENSR00001448099 (open chromatin region)	1	muscle (HSMM)	DNase1
ENSR00000399198 (promoter flanking region)	5	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K27ac
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K4me1
		endothelium (HUVEC)	H3K36me3, Cjun, Max, DNase1, Cmyc
		liver (HepG2)	H3K4me1, H3K4me2, HNF4A, H3K9ac, H3K27ac, Cmyc, DNase1, USF1, PolII
		nervous (NH-A)	DNase1
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001664576 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		blood (DND-41)	H3K27me3
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K27ac
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K4me1
		endothelium (HUVEC)	Max, DNase1, Cmyc, Cjun
		liver (HepG2)	PolII, USF1, H3K4me1, H3K4me2, HNF4A, H3K9ac, H3K27ac, Cmyc, DNase1
		nervous (NH-A)	DNase1
ENSR00000399203 (promoter)	4	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, Egr1, Yy1, H3K4me2, H3K4me3, DNase1
		HSMMtube	DNase1, H2AZ, H3K9ac, H3K4me2, H3K4me3
		blood (K562)	Egr1, H3K9ac, H3K79me2, HEY1, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3
		liver (HepG2)	H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3, Gabp, PolII, H3K79me2, TAF1, ELF1, H3K4me1, DNase1
		lung (IMR90)	H3K9ac, DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3
		blood (GM12878)	PolII, H2AZ, ELF1, H3K4me3, H3K4me2, H3K9ac, DNase1
		nervous (NH-A)	H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		skin (NHEK)	H3K4me2, H3K9ac, H3K4me1, H3K4me3, H3K36me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1, H3K36me3
		cervix (HeLa-S3)	DNase1, PolII, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, H3K4me3, H3K4me2, H3K9ac, H3K4me1, H3K27ac, PolII
		A549	H3K36me3, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000649136 (promoter flanking region)	12	embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	H3K27me3, DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1

		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K4me3, PolII, USF1, H3K4me1, H2AZ, H3K4me2, CTCF, DNase1
		lung (IMR90)	DNase1
ENSR00001664577 (CTCF binding site)	2	monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	CTCF, Rad21
		liver (HepG2)	H3K4me1, H3K4me2
		HSMMtube	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CST3	downstream gene variant, upstream gene variant	70	ENST00000398411	NM_001288614.1	ENSP00000381448	32
CST3	downstream gene variant, upstream gene variant	24	ENST00000376925	NM_000099.3	ENSP00000366124	25
CST3	downstream gene variant, upstream gene variant	33	ENST00000398409	?	ENSP00000381446	23
CST9	downstream gene variant, upstream gene variant	171	ENST00000376971	NM_001008693.2	ENSP00000366170	21
RP11-218C14.2	downstream gene variant	2039	ENST00000437612	?	?	5
RP11-218C14.8	downstream gene variant, upstream gene variant	158	ENST00000602977	?	?	16

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST3	ENST00000398409	?	ENSP00000381446	8
CST3	ENST00000398411	NM_001288614.1	ENSP00000381448	14
CST3	ENST00000376925	NM_000099.3	ENSP00000366124	7
CST9	ENST00000376971	NM_001008693.2	ENSP00000366170	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST3	ENST00000398411	NM_001288614.1	ENSP00000381448	4
CST9	ENST00000376971	NM_001008693.2	ENSP00000366170	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST3	ENST00000398409	?	ENSP00000381446	3
CST3	ENST00000398411	NM_001288614.1	ENSP00000381448	1
CST3	ENST00000376925	NM_000099.3	ENSP00000366124	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-218C14.8	ENST00000602977	?	2

Locus 294 (rs2795502)

SNiPAcard

Block annotations

Block info

genomic range	chr10:43,337,842-43,423,626
block size	85,785 bp
variant count	54 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.516 [-3.961 – 2.125]	gene(s) hit or close-by	RP11-124O11.1
phastCons	μ = 0.078 [0 – 0.766]	eQTL gene(s)	ANKRA2
GERP++	μ = -0.233 [-6.18 – 1.58]	potentially regulated gene(s)	BMS1, CCNYL2, HNRNPF, RASGEF1A, RET, RET, RP11-168L22.2, RP11-517P14.2
CADD score	μ = 2.663 [0.012 – 11.37]	disease gene(s)	RET, BMS1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Progressive supranuclear palsy	<3.00×10-7	GWAS Catalog	21685912	1

Disease gene annotation

gene	trait	source DB	source entry/link
RET	RENAL AGENESIS	DECIPHER	MIM:191830
RET	Multiple endocrine neoplasia IIB	DECIPHER	MIM:162300
BMS1	APLASIA CUTIS CONGENITA, NONSYNDROMIC	OMIM	MIM:107600
RET	PHEOCHROMOCYTOMAPHEOCHROMOCYTOMA, SUSCEPTIBILITY TO	OMIM	MIM:171300
RET	CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL	OMIM	MIM:209880
RET	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIA	OMIM	MIM:171400
RET	THYROID CARCINOMA, FAMILIAL MEDULLARY	OMIM	MIM:155240
RET	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIB	OMIM	MIM:162300
RET	COLORECTAL CANCER	OMIM	MIM:114500
RET	HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, 1	OMIM	MIM:142623
RET	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550
BMS1	Circumscribed cutaneous aplasia of the vertex	OrphaNet	OrphaNet:1114
RET	Unilateral renal dysplasia	OrphaNet	OrphaNet:93172
RET	Bilateral renal dysplasia	OrphaNet	OrphaNet:93173
RET	Haddad syndrome	OrphaNet	OrphaNet:99803
RET	Hirschsprung disease	OrphaNet	OrphaNet:388
RET	Multiple endocrine neoplasia type 2A	OrphaNet	OrphaNet:247698
RET	Papillary or follicular thyroid carcinoma	OrphaNet	OrphaNet:146
RET	Multiple endocrine neoplasia type 2B	OrphaNet	OrphaNet:247709
RET	Familial medullary thyroid carcinoma	OrphaNet	OrphaNet:99361
RET	Bilateral renal agenesis	OrphaNet	OrphaNet:1848

Direct effect on regulation

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)

ANKRA2	ENST00000506130	ILMN_1687351	chr5	blood	3.54×10-7 (p-value)	Westra et al.	1
ANKRA2	ENST00000504641						
ANKRA2	ENST00000515804						
ANKRA2	ENST00000514916						
ANKRA2	ENST00000296785						

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000072560	1	ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
		ENCP00000008241	HNRNPF
		ENCP00000008235	RP11-168L22.2
		ENCP00000008242	RP11-517P14.2
		ENCP00000008212	CCNYL2
		ENCP00000008234	RASGEF1A

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000353984 (CTCF binding site)	2	endothelium (HUVEC)	H3K36me3, H3K27me3
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		liver (HepG2)	CTCF
		blood (K562)	CTCF, H3K27me3
ENSR00000353986 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3, CTCF
		blood (DND-41)	H3K27me3
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF, H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		liver (HepG2)	CTCF, H3K27me3
		lung (IMR90)	H3K27me3, CTCF
		A549	CTCF, H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-124O11.1	upstream gene variant	2463	ENST00000421320	?	?	1

Locus 295 (rs2284178)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,430,799-31,432,125
block size	1,327 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.123 [-2.407 – 0.16]	gene(s) hit or close-by	HCP5, XXbac-BPG181B23.6
phastCons	μ = 0.215 [0 – 0.431]	eQTL gene(s)	AIF1, CCHCR1, CYP21A1P, HLA-C, LST1, MICB, PSORS1C1, PSORS1C2, VARS2, XXbac-BPG248L24.12, XXbac-BPG299F13.17
GERP++	μ = -3.415 [-6.83 – 0]	potentially regulated gene(s)	HCP5, HCP5, HCP5, HCP5
CADD score	μ = 5.348 [0.416 – 10.28]	disease gene(s)	VARS2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Behcet syndrome	3.06×10^{-10}	dbGaP	pha002888	1

Disease gene annotation

gene	trait	source DB	source entry/link
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LST1	ENST00000339530	ILMN_1718936	blood	2.73×10^{-5} (p-value)	MuTHER consortium	2
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
MICB	ENST00000538442	ILMN_1708006	skin	5.07×10^{-10} (p-value)	MuTHER consortium	2
MICB	ENST00000399150					
MICB	ENST00000252229					
HLA-C	?	ENSG00000204525	atrial appendage	2.87×10^{-11} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	transformed fibroblasts	4.29×10^{-15} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	blood	3.23×10^{-19} (p-value)	GTEx Portal V6	2

XXbac-BPG248L24.12	?	ENSG00000271581	blood	4.48×10-9 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	blood	7.80×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	breast	1.02×10-15 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	breast	2.40×10-7 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	breast	1.67×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	skeletal muscle	1.73×10-24 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	2.17×10-11 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	skeletal muscle	9.85×10-9 (p-value)	GTEx Portal V6	2
CCHCR1	?	ENSG00000204536	blood	2.01×10-2 (q-value)	SeeQTL DB (HapMap)	2
HLA-C	?	ENSG00000204525	EBV lymphocytes	6.11×10-10 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	ovary	3.64×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	sun exposed skin	1.82×10-19 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	3.21×10-8 (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	sun exposed skin	7.82×10-6 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	sun exposed skin	2.70×10-8 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	unexposed skin	4.53×10-12 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	aorta	1.51×10-11 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	aorta	8.40×10-9 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	left ventricle	4.44×10-8 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	2.32×10-16 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	3.05×10-8 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	subcutaneous adipocytes	8.87×10-8 (p-value)	GTEx Portal V6	2
VARS2	?	ENSG00000137411	subcutaneous adipocytes	2.89×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	visceral adipocytes	2.02×10-11 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	1.53×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	stomach	1.64×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	cortex	1.79×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	tibial nerve	3.17×10-14 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	tibial nerve	1.46×10-10 (p-value)	GTEx Portal V6	2
PSORS1C2	?	ENSG00000204538	esophagus mucosa	4.95×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	esophagus mucosa	3.38×10-12 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	esophagus mucosa	5.95×10-6 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	esophagus mucosa	2.52×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	pancreas	4.35×10-10 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	muscularis mucosae	5.10×10-11 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	muscularis mucosae	2.08×10-6 (p-value)	GTEx Portal V6	2

XXbac-BPG299F13.17	?	ENSG00000272501	muscularis mucosae	3.39×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	lung	1.54×10^{-15} (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	lung	5.45×10^{-6} (p-value)	GTEx Portal V6	2
CYP21A1P	?	ENSG00000204338	lung	3.27×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	lung	6.58×10^{-10} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	tibial artery	7.32×10^{-20} (p-value)	GTEx Portal V6	2
CCHCR1	?	ENSG00000204536	tibial artery	1.16×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	2.96×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	tibial artery	1.68×10^{-14} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	thyroid	6.25×10^{-18} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	thyroid	9.79×10^{-9} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	transverse colon	1.00×10^{-11} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	transverse colon	7.75×10^{-9} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	liver	1.05×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	terminal ileum	1.22×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	nucleus accumbens	1.37×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	sigmoid colon	5.76×10^{-8} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	vagina	2.96×10^{-7} (p-value)	GTEx Portal V6	2
AIF1	ENST00000466820	ILMN_1703538	blood	2.86×10^{-5} (p-value)	Westra et al.	2
AIF1	ENST00000376059					
HLA-C	?	ENSG00000204525	gastroesophageal junction	8.77×10^{-7} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	gastroesophageal junction	2.91×10^{-6} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	adrenal gland	5.91×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	cerebellum	4.73×10^{-9} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	hippocampus	6.32×10^{-8} (p-value)	GTEx Portal V6	2

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	215666_at	chr6	blood	1.40×10^{-9} (p-value)	Dixon et al.	2

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000048017		HCP5
		HCP5
		HCP5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000129758 (promoter flanking region)	2	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, DNase1, TAF7, PolII, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K4me2, H2AZ, H3K36me3
		blood (K562)	H3K27me3, Cfos
		skin (NHDF-AD)	DNase1, H3K27me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		A549	DNase1, H3K4me3
		skin (NHEK)	H3K4me3, DNase1, H3K4me2, H3K9ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HCP5	upstream gene variant	163	ENST00000467369	?	?	1
HCP5	upstream gene variant, downstream gene variant	163	ENST00000460889	?	?	2
HCP5	upstream gene variant	160	ENST00000541196	?	?	1
XXbac-BPG181B23.6	upstream gene variant	1012	ENST00000440087	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	2
HCP5	ENST00000467369	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
HCP5	ENST00000541196	?	1
HCP5	ENST00000414046	?	2
XXbac-BPG181B23.6	ENST00000440087	?	1

Locus 296 (rs9427116)

SNiPAcard

Block annotations

Block info

genomic range	chr1:154,631,123-154,652,119
block size	20,997 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.331 [-0.913 – 1.261]	gene(s) hit or close-by	–
phastCons	μ = 0.126 [0 – 0.533]	eQTL gene(s)	IL6R, RUSC1-AS1
GERP++	μ = 0.161 [-0.815 – 1.54]	potentially regulated gene(s)	–
CADD score	μ = 4.915 [0.849 – 18.13]	disease gene(s)	IL6R

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
IL6R	INTERLEUKIN 6, SERUM LEVEL OF, QUANTITATIVE TRAIT LOCUS	OMIM	MIM:614752
IL6R	SOLUBLE INTERLEUKIN-6 RECEPTOR, SERUM LEVEL OF, QUANTITATIVE [...]	OMIM	MIM:614689

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RUSC1-AS1	ENST00000543656	ILMN_2114185	blood	4.94×10^{-5} (p-value)	MuTHER consortium	4
RUSC1-AS1	ENST00000446880					
RUSC1-AS1	ENST00000450199					
IL6R	ENST00000368485	ILMN_1696394	blood	5.45×10^{-6} (p-value)	Westra et al.	5
IL6R	ENST00000507256					
IL6R	ENST00000502679					
IL6R	ENST00000515190					

Locus 297 (rs494189)

SNiPAcard

Block annotations

Block info

genomic range	chr1:168,696,549-168,729,840
block size	33,292 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.151 [-4.415 – 0.9]	gene(s) hit or close-by	AL049798.1, DPT, RP1-10C16.1
phastCons	μ = 0.007 [0 – 0.091]	eQTL gene(s)	DPT
GERP++	μ = -1.886 [-6.87 – 0.0691]	potentially regulated gene(s)	DPT
CADD score	μ = 1.904 [0.114 – 6.621]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DPT	ENST00000367817	ILMN_1708107	skin	1.26×10^{-5} (p-value)	MuTHER consortium	3
DPT	?	ENSG00000143196	aorta	1.19×10^{-6} (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000043396	1	ENCP00000005421	DPT

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001587687 (enhancer)	1	HSMMtube	H3K27me3
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, H3K4me2
ENSR00000547238 (TF binding site)	1	cervix (HeLa-S3)	DNase1
		embryonic stem cell (H1ESC)	DNase1
		endothelium (HUVEC)	H3K36me3
		A549	H3K4me3, H3K4me2
		nervous (NH-A)	DNase1
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL049798.1	downstream gene variant, upstream gene variant	2456	ENST00000408651	?	?	4
DPT	upstream gene variant	1632	ENST00000367817	NM_001937.4	ENSP00000356791	2
RP1-10C16.1	downstream gene variant	2763	ENST00000457405	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DPT	ENST00000367817	NM_001937.4	ENSP00000356791	3

Locus 298 (rs9651367)

SNiPAcard

Block annotations

Block info

genomic range	chr10:20,210,060-20,272,662
block size	62,603 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.225 [-1.003 – 0.583]	**gene(s) hit or close-by**	PLXDC2
phastCons	μ = 0.004 [0 – 0.027]	**eQTL gene(s)**	–
GERP++	μ = 0.007 [-3.75 – 2.28]	**potentially regulated gene(s)**	–
CADD score	μ = 4.352 [0.412 – 11.7]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001421558 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K27ac, H3K9ac
		Osteobl	H3K4me2, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
		breast (HMEC)	DNase1, H3K27ac, H3K4me1, H3K4me2
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	FOXA1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	DNase1, H3K4me1, H3K27ac
ENSR00000351639 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K4me2, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	H3K4me2, DNase1
ENSR00001594041 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H4K20me1
		endothelium (HUVEC)	Cjun, DNase1
		nervous (NH-A)	DNase1
ENSR00001594043 (enhancer)	1	monocytes (Monocytes-CD14+)	H4K20me1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLXDC2	ENST00000377242	NM_001282736.1	ENSP00000366450	9
PLXDC2	ENST00000377252	NM_032812.8	ENSP00000366460	9

Locus 299 (rs6547631)

SNiPAcard

Block annotations

Block info

genomic range	chr2:85,909,026-85,912,870
block size	3,845 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.129 [-0.867 – 0.579]	gene(s) hit or close-by	AC012454.4, GNLY
phastCons	μ = 0.022 [0.001 – 0.086]	eQTL gene(s)	AC012454.4, ATOH8, GNLY
GERP++	μ = -0.212 [-0.761 – 0.388]	potentially regulated gene(s)	–
CADD score	μ = 4.218 [1.561 – 8.197]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AC012454.4	?	ENSG00000203363	blood	5.76×10-6 (p-value)	GTEx Portal V6	1
GNLY	?	ENSG00000115523	blood	9.30×10-12 (p-value)	GTEx Portal V6	3
GNLY	?	ENSG00000115523	left ventricle	3.43×10-7 (p-value)	GTEx Portal V6	3
GNLY	?	ENSG00000115523	subcutaneous adipocytes	4.26×10-7 (p-value)	GTEx Portal V6	3
?	?	ILMN_1790692	monocyte	5.43×10-20 (p-value)	Zeller et al.	1
ATOH8	ENST00000469442	ILMN_1654065	monocyte	1.82×10-75 (p-value)	Zeller et al.	1
ATOH8	ENST00000306279					
ATOH8	ENST00000489682					
GNLY	ENST00000489214	ILMN_1708779	monocyte	5.56×10-12 (p-value)	Zeller et al.	1
GNLY	ENST00000409696					
GNLY	ENST00000534351					
GNLY	ENST00000489980					
GNLY	ENST00000533041					
GNLY	ENST00000482900					
GNLY	ENST00000470974					
GNLY	ENST00000464298					
GNLY	ENST00000263863					
GNLY	ENST00000524600					
GNLY	?	ENSG00000115523	thyroid	1.18×10-6 (p-value)	GTEx Portal V6	2
GNLY	?	ENSG00000115523	lung	6.10×10-6 (p-value)	GTEx Portal V6	1
GNLY	?	ENSG00000115523	sun exposed skin	8.77×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000593767	1	cervix (HeLa-S3)	H3K4me2, H3K27ac, DNase1

(enhancer)	embryonic stem cell (H1ESC)	H3K27me3
	HSMMtube	DNase1
	liver (HepG2)	H3K27me3
	blood (K562)	H3K27me3
	blood (DND-41)	H3K27me3
	muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC012454.4 e!	downstream gene variant	306	ENST00000443948 e!	?	?	4
GNLY e!	upstream gene variant	669	ENST00000488945 e!	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GNLY e!	ENST00000488945 e!	?	?	2

Locus 300 (rs1922291)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,816,695-102,842,124
block size	25,430 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.948 [-1.696 – 0.167]	gene(s) hit or close-by	IL1RL2
phastCons	μ = 0.053 [0 – 0.303]	eQTL gene(s)	IL1RL2
GERP++	μ = -0.858 [-2.87 – 0.466]	potentially regulated gene(s)	–
CADD score	μ = 1.467 [0.89 – 2.161]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL1RL2	?	ENSG00000115598	sun exposed skin	3.32×10^{-8} (p-value)	GTEx Portal V6	7
IL1RL2	?	ENSG00000115598	unexposed skin	2.32×10^{-6} (p-value)	GTEx Portal V6	7
IL1RL2	?	ENSG00000115598	thyroid	4.87×10^{-7} (p-value)	GTEx Portal V6	7

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000595631 (CTCF binding site)	1	liver (HepG2)	H2AZ, H3K4me2, CTCF
		blood (DND-41)	H3K27me3
ENSR00001655312 (enhancer)	1	liver (HepG2)	CTCF, H3K4me2, H2AZ
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL1RL2	downstream gene variant	1007	ENST00000421464	?	ENSP00000387611	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RL2	ENST00000441515	?	ENSP00000413348	7
IL1RL2	ENST00000264257	NM_003854.2	ENSP00000264257	7
IL1RL2	ENST00000481806	?	?	7
IL1RL2	ENST00000421464	?	ENSP00000387611	1

Locus 301 (rs6003)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,945,050-197,257,556
block size	312,507 bp
variant count	155 variants

Basic features

Conservation/deleteriousness			Linked genes
phyloP	μ = -0.701 [-7.024 – 2.567]	gene(s) hit or close-by	ASPM, CFHR5, CRB1, F13B, RP11-32D17.4, RP11-332L8.1, ZBTB41
phastCons	μ = 0.052 [0 – 1]	eQTL gene(s)	CFHR1, CFHR3, ZBTB41
GERP++	μ = -0.725 [-9.06 – 3.26]	potentially regulated gene(s)	ASPM, CFH, CFH, CFHR1, CFHR1, CFHR2, CFHR2, CFHR3, CFHR3, CFHR4
CADD score	μ = 3.100 [0.004 – 17.66]	disease gene(s)	CFHR5, F13B, ASPM, CRB1, CFHR3, CFHR1, CFH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Macular degeneration	6.24×10^{-11}	dbGaP	pha002890	2
Macular degeneration	7.66×10^{-12}	dbGaP	pha002869	2
Macular degeneration	4.75×10^{-6}	dbGaP	pha002856	9
Macular degeneration	4.57×10^{-9}	dbGaP	pha000002	3
Macular degeneration	2.17×10^{-9}	dbGaP	pha000001	9

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000145187.5	1
Clinvar: phenotype not specified	likely benign	ClinVar	RCV000145113.1	1
Clinvar: phenotype not specified	likely benign	ClinVar	RCV000145090.1	1
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000145076.2	1
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000081938.6	1
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000081936.6	1
Primary autosomal recessive microcephaly 5	benign	ClinVar	RCV000020792.1	1
Primary autosomal recessive microcephaly 5	benign	ClinVar	RCV000020768.1	1
Venous thrombosis, susceptibility to	risk factor	ClinVar	RCV000017984.1	1
?	HGMD curated	HGMD	CR063400	1
?	HGMD curated	HGMD	CR063399	1
?	HGMD curated	HGMD	CM034655	1
VENOUS THROMBOSIS, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:134580	1

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR5	CFHR5 DEFICIENCY	OMIM	MIM:614809
F13B	FACTOR XIII, B SUBUNIT, DEFICIENCY OF	OMIM	MIM:613235

ASPM	MICROCEPHALY 5, PRIMARY, AUTOSOMAL RECESSIVE	OMIM	MIM:608716	OMIM
CRB1	RETINITIS PIGMENTOSA 12	OMIM	MIM:600105	OMIM
CRB1	LEBER CONGENITAL AMAUROSIS 8	OMIM	MIM:613835	OMIM
CRB1	PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY	OMIM	MIM:172870	OMIM
CRB1	RETINITIS PIGMENTOSA	OMIM	MIM:268000	OMIM
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581	orphanet
CFHR5	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581	orphanet
CFHR5	C3 glomerulonephritis	OrphaNet	OrphaNet:329931	orphanet
F13B	Congenital factor XIII deficiency	OrphaNet	OrphaNet:331	orphanet
CFHR1	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581	orphanet
CFHR1	C3 glomerulonephritis	OrphaNet	OrphaNet:329931	orphanet
CFHR1	Dense deposit disease	OrphaNet	OrphaNet:93571	orphanet
ASPM	Autosomal recessive primary microcephaly	OrphaNet	OrphaNet:2512	orphanet
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579	orphanet
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421	orphanet
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571	orphanet
CFH	Familial drusen	OrphaNet	OrphaNet:75376	orphanet
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903	orphanet
CRB1	Retinitis pigmentosa	OrphaNet	OrphaNet:791	orphanet
CRB1	Pigmented paravenous retinochoroidal atrophy	OrphaNet	OrphaNet:251295	orphanet
CRB1	Leber congenital amaurosis	OrphaNet	OrphaNet:65	orphanet
ASPM	Primary Autosomal Recessive Microcephaly	DECIPHER	MIM:608716	OMIM
CRB1	LEBER CONGENITAL AMAUROSIS 8	DECIPHER	MIM:613835	OMIM
CRB1	Retinitis pigmentosa-12, autosomal recessive	DECIPHER	MIM:600105	OMIM

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
F13B	missense variant	ENST00000367412	NM_001994.2	ENSP00000356382	H/R	cAt/cGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	lung	1.59×10^{-9} (p-value)	GTEx Portal V6	133
CFHR3	?	ENSG00000116785	aorta	1.12×10^{-6} (p-value)	GTEx Portal V6	21
ZBTB41	?	ENSG00000177888	subcutaneous adipocytes	8.07×10^{-8} (p-value)	GTEx Portal V6	127
CFHR3	?	ENSG00000116785	thyroid	4.38×10^{-10} (p-value)	GTEx Portal V6	130
CFHR3	?	ENSG00000116785	prostate	3.26×10^{-6} (p-value)	GTEx Portal V6	1
CFHR1	?	ENSG00000244414	transformed fibroblasts	2.75×10^{-5} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	muscularis mucosae	1.98×10^{-5} (p-value)	GTEx Portal V6	2
CFHR3	?	ENSG00000116785	adrenal gland	1.18×10^{-5} (p-value)	GTEx Portal V6	4

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000048862	1	ENCP00000005943	CFHR2 CFHR2
		ENCP00000005941	CFHR1 CFHR1
ENCE00000048832	1	ENCP00000005947	ASPM
		ENCP00000005938	CFH CFH
ENCE00000048835	1	ENCP00000005942	CFHR4
		ENCP00000005939	CFHR3 CFHR3

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001527237 (CTCF binding site)	1	skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001589240 (enhancer)	1	skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001589241 (CTCF binding site)	1	liver (HepG2)	CTCF
ENSR00001527239 (promoter flanking region)	1	HSMMtube	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00000288050 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	DNase1
		A549	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00000550137 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00001527244 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K36me3
ENSR00001589251 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001527245 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001589253 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3

		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001527246 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H2AZ
		blood (K562)	H3K79me2, H3K36me3
		blood (DND-41)	H3K4me2, H3K36me3, H3K4me1
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (GM12878)	H2AZ, H3K79me2, H3K4me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
ENSR00000550147 (promoter flanking region)	4	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (K562)	H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K4me1, HNF4A, FOXA1, H3K27ac
		blood (GM12878)	H3K36me3
		A549	DNase1, H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000550149 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K27ac, CTCF, H3K36me3
		blood (K562)	DNase1, CTCF, H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	DNase1, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		A549	DNase1, H3K27ac, H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001527247 (promoter flanking region)	1	cervix (HeLa-S3)	H3K4me2
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (GM12878)	H3K4me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2, H3K4me2
		muscle (HSMM)	H3K36me3
ENSR00001589259 (enhancer)	1	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ASPM	1	ENST00000294732 ENST00000367408 ENST00000367409	hsa-miR-382-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ASPM	downstream gene variant, upstream gene variant	454	ENST00000294732	NM_001206846.1	ENSP00000294732	6
ASPM	downstream gene variant, upstream gene variant	458	ENST00000367408	?	ENSP00000356378	11
ASPM	downstream gene variant, upstream gene variant	930	ENST00000612785	?	ENSP00000479244	5
ASPM	downstream gene variant, upstream gene variant	454	ENST00000367409	NM_018136.4	ENSP00000356379	6
CFHR5	upstream gene variant, downstream gene variant	140	ENST00000256785	NM_030787.3	ENSP00000256785	9
F13B	downstream gene variant, upstream gene variant	471	ENST00000367412	NM_001994.2	ENSP00000356382	11
F13B	downstream gene variant, upstream gene variant	1471	ENST00000490002	?	?	7
RP11-32D17.4	downstream gene variant, upstream gene variant	256	ENST00000442280	?	?	10
RP11-332L8.1	upstream gene variant	2444	ENST00000417716	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ASPM	ENST00000612785	?	ENSP00000479244	L	ttG/ttA	3
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	1	2	3
ASPM	ENST00000367408	?	ENSP00000356378	S	tcA/tcT	2
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	2	3	3
F13B	ENST00000367412	NM_001994.2	ENSP00000356382	T	acG/acA	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	1
ASPM	ENST00000367408	?	ENSP00000356378	1
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ASPM	ENST00000612785	?	ENSP00000479244	45
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	46
ASPM	ENST00000367408	?	ENSP00000356378	35
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	46
CFHR5	ENST00000256785	NM_030787.3	ENSP00000256785	11
CRB1	ENST00000475659	?	?	4
CRB1	ENST00000538660	NM_001257966.1	ENSP00000438091	4
CRB1	ENST00000535699	NM_001257965.1	ENSP00000438786	4
CRB1	ENST00000484075	?	ENSP00000433932	4
CRB1	ENST00000367400	NM_201253.2	ENSP00000356370	4
CRB1	ENST00000367399	NM_001193640.1	ENSP00000356369	4
F13B	ENST00000490002	?	?	7
F13B	ENST00000367412	NM_001994.2	ENSP00000356382	17
ZBTB41	ENST00000367405	NM_194314.2	ENSP00000356375	24

ZBTB41	ENST00000467322	?	?	24

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	1
ASPM	ENST00000367408	?	ENSP00000356378	1
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFHR5	ENST00000256785	NM_030787.3	ENSP00000256785	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-32D17.4	ENST00000442280	?	1

Locus 302 (rs27303)

SNiPAcard

Block annotations

Block info

genomic range	chr5:96,374,150-96,374,150
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.623	gene(s) hit or close-by	LNPEP
phastCons	0	eQTL gene(s)	ERAP2
GERP++	-3.9	potentially regulated gene(s)	–
CADD score	5.479	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP2	?	ENSG00000164308	pancreas	1.79×10^{-8} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	uterus	1.42×10^{-6} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	muscularis mucosae	9.10×10^{-9} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	lung	9.64×10^{-16} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	transformed fibroblasts	1.50×10^{-12} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	blood	3.15×10^{-10} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	breast	3.08×10^{-6} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	tibial artery	9.57×10^{-9} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	thyroid	1.23×10^{-16} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	skeletal muscle	1.79×10^{-13} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	EBV lymphocytes	2.53×10^{-6} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	transverse colon	2.06×10^{-8} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	liver	4.03×10^{-7} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	sun exposed skin	2.65×10^{-12} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	unexposed skin	2.61×10^{-9} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	left ventricle	8.70×10^{-7} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	aorta	7.76×10^{-8} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	subcutaneous adipocytes	1.29×10^{-15} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	stomach	2.16×10^{-8} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	visceral adipocytes	6.43×10^{-6} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	nucleus accumbens	5.66×10^{-8} (p-value)	GTEx Portal V6	1

ERAP2	?	ENSG00000164308	sigmoid colon	2.55×10^{-8} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	tibial nerve	1.36×10^{-11} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	esophagus mucosa	4.57×10^{-10} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	adrenal gland	3.22×10^{-11} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	cerebellar hemisphere	1.46×10^{-6} (p-value)	GTEx Portal V6	1
ERAP2	?	ENSG00000164308	testis	4.48×10^{-8} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LNPEP	downstream gene variant	931	ENST00000231368	NM_005575.2	ENSP00000231368	1

Locus 303 (rs17354650)

SNiPAcard

Block annotations

Block info

genomic range	chr1:20,390,322-20,390,322
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.128	gene(s) hit or close-by	PLA2G5
phastCons	0	eQTL gene(s)	MRTO4, OTUD3
GERP++	0.13	potentially regulated gene(s)	–
CADD score	4.268	disease gene(s)	PLA2G5

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PLA2G5	FLECK RETINA, FAMILIAL BENIGN	OMIM	MIM:228980
PLA2G5	Familial benign flecked retina	OrphaNet	OrphaNet:363989

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MRTO4	ENST00000330263	ILMN_1689800	monocyte	4.13×10^{-4} (p-value)	Fairfax et al.	1
MRTO4	ENST00000479559					
OTUD3	ENST00000375120	ILMN_1779280	b-cell	3.71×10^{-6} (p-value)	Fairfax et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000531978 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLA2G5	ENST00000465698	?	?	1
PLA2G5	ENST00000489871	?	?	1

PLA2G5	ENST00000469069	?	?	1
PLA2G5	ENST00000498348	?	?	1
PLA2G5	ENST00000486277	?	?	1
PLA2G5	ENST00000460175	?	?	1

Locus 304 (rs722414)

SNiPAcard

Block annotations

Block info

genomic range	chr12:89,826,210-89,940,502
block size	114,293 bp
variant count	94 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.634 [-4.107 – 5.606]	gene(s) hit or close-by	CENPCP1, GALNT4, POC1B, POC1B-GALNT4, RP11-734K2.3, RP11-734K2.4
phastCons	μ = 0.036 [0 – 1]	eQTL gene(s)	ATP2B1, GALNT4, POC1B, POC1B-GALNT4, RP11-981P6.1
GERP++	μ = -0.111 [-9.6 – 4.59]	potentially regulated gene(s)	–
CADD score	μ = 3.623 [0.036 – 20.6]	disease gene(s)	POC1B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1-linoleoylglycerophosphoethanolamine*	1.97×10^{-5}	Metabolomics GWAS Server	24816252	13
ascorbate (Vitamin C)	9.10×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
POC1B	CONE-ROD DYSTROPHY 20	OMIM	MIM:615973

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
GALNT4	missense variant	ENST00000529983	NM_003774.4	ENSP00000436604	I/T	aTa/aCa	tolerated	benign	1
POC1B-GALNT4	missense variant	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	I/T	aTa/aCa	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
POC1B	?	ENSG00000139323	tibial nerve	1.80×10^{-12} (p-value)	GTEx Portal V6	89
GALNT4	ENST00000529983	ILMN_1739297	skin	7.81×10^{-44} (p-value)	MuTHER consortium	36
POC1B-GALNT4	ENST00000547474		blood	5.59×10^{-68} (p-value)	MuTHER consortium	36
POC1B-GALNT4	ENST00000548729		adipocyte	2.57×10^{-41} (p-value)	MuTHER consortium	36
GALNT4	?	ENSG00000257594	blood	7.61×10^{-4} (q-value)	SeeQTL DB (HapMap)	16
POC1B-GALNT4	ENST00000547474	ILMN_1739297	monocyte	4.40×10^{-58} (p-value)	Fairfax et al.	5

GALNT4 / POC1B-GALNT4	ENST00000529983 / ENST00000548729		b-cell	7.78×10-58 (p-value)	Fairfax et al.	5
GALNT4	?	ENSG00000257594	brain	1.13×10-3 (q-value)	SeeQTL DB (Myers et al.)	10
RP11-981P6.1	?	ENSG00000258302	thyroid	1.24×10-6 (p-value)	GTEx Portal V6	1
ATP2B1	ENST00000428670 / ENST00000359142 / ENST00000261173	ILMN_1824297	blood	7.75×10-6 (p-value)	Westra et al.	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
GALNT4 / POC1B-GALNT4	ENST00000529983 / ENST00000547474 / ENST00000548729	ILMN_1739297	chr12	monocyte	7.49×10-107 (p-value)	Zeller et al.	10

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000434333 (promoter flanking region)	2	embryonic stem cell (H1ESC)	USF1, DNase1
		HSMMtube	H3K27ac, H3K4me2, DNase1, H3K27me3, H3K9ac
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1, H3K27ac
		nervous (NH-A)	DNase1
		NHLF	DNase1, H3K27ac
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, H3K4me1, H3K4me2
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1, H3K4me1, H3K27ac, PolII
		A549	H3K36me3
ENSR00000434334 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		lung (IMR90)	CTCF
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF
ENSR00000434337 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	Cjun
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	BATF, MEF2A, H3K4me2
		nervous (NH-A)	DNase1
		A549	H3K36me3
		blood (DND-41)	H3K36me3, H3K4me1
ENSR00000654046 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	DNase1, CTCF
		blood (K562)	CTCF, Rad21
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF

		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00001613709 (enhancer)	1	blood (DND-41)	H3K36me3
ENSR00001613708 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00001465659 (promoter flanking region)	2	cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		Osteobl	H3K4me2, H3K36me3
		lung (IMR90)	DNase1, H3K4me2
		blood (DND-41)	H3K36me3
		A549	H3K36me3
ENSR00000434344 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		blood (DND-41)	H3K36me3
		A549	H3K27ac, DNase1, H3K4me3
ENSR00001465660 (promoter flanking region)	4	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		Osteobl	H3K4me2, H3K36me3
		blood (K562)	H3K36me3
		blood (DND-41)	H3K36me3, H3K4me3, H3K4me2, H3K4me1
		skin (NHDF-AD)	H3K4me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K79me2, H3K4me1, H3K4me2, FOXA1
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K4me3
		A549	H3K36me3, H3K4me2, H3K4me3
		skin (NHEK)	H3K36me3
ENSR00000245284 (TF binding site)	2	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	Gata2, DNase1, Egr1, PolII
ENSR00001613716 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	DNase1, CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF, DNase1
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00001465663 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	DNase1, CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	DNase1, CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	CTCF
		NHLF	CTCF

		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00001613717 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	CTCF, DNase1, H3K27ac
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00000434348 (promoter flanking region)	1	HSMMtube	DNase1
		A549	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CENPCP1	upstream gene variant, downstream gene variant	158	ENST00000533085	?	?	15
GALNT4	downstream gene variant	230	ENST00000529983	NM_003774.4	ENSP00000436604	14
POC1B	upstream gene variant	2973	ENST00000549591	?	?	1
POC1B	downstream gene variant, upstream gene variant	40	ENST00000549304	?	?	11
POC1B	downstream gene variant	793	ENST00000539190	?	?	9
POC1B	downstream gene variant	2049	ENST00000552563	?	?	3
POC1B	downstream gene variant, upstream gene variant	250	ENST00000551819	?	?	4
POC1B	downstream gene variant	2257	ENST00000546830	?	ENSP00000449256	3
POC1B	upstream gene variant	1519	ENST00000546740	?	?	3
POC1B	downstream gene variant	900	ENST00000547274	?	ENSP00000449648	1
POC1B-GALNT4	downstream gene variant	230	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	14
POC1B-GALNT4	downstream gene variant	162	ENST00000547474	?	ENSP00000447754	9
RP11-734K2.3	downstream gene variant, upstream gene variant	543	ENST00000623264	?	?	7
RP11-734K2.4	upstream gene variant	1913	ENST00000605233	?	?	5

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
POC1B	ENST00000548715	?	ENSP00000449945	1
POC1B	ENST00000313546	NM_172240.2	ENSP00000323302	1
POC1B	ENST00000393179	?	ENSP00000376877	1
POC1B	ENST00000549035	NM_001199777.1	ENSP00000447916	1
POC1B	ENST00000547496	?	ENSP00000447437	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
POC1B	ENST00000551819	?	?	3
POC1B	ENST00000546740	?	?	23
POC1B	ENST00000548715	?	ENSP00000449945	82
POC1B	ENST00000313546	NM_172240.2	ENSP00000323302	82
POC1B	ENST00000547274	?	ENSP00000449648	49
POC1B	ENST00000393179	?	ENSP00000376877	82
POC1B	ENST00000549504	?	ENSP00000450118	82
POC1B	ENST00000549035	NM_001199777.1	ENSP00000447916	82
POC1B	ENST00000552563	?	?	56
POC1B	ENST00000549304	?	?	8
POC1B	ENST00000546830	?	ENSP00000449256	44
POC1B	ENST00000547496	?	ENSP00000447437	82
POC1B	ENST00000539190	?	?	59
POC1B-GALNT4	ENST00000547474	?	ENSP00000447754	1
RP11-734K2.4	ENST00000605233	?	?	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GALNT4	ENST00000529983	NM_003774.4	ENSP00000436604	6
POC1B-GALNT4	ENST00000548729	NM_001199782.1, NM_001199781.1	ENSP00000447852	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CENPCP1	ENST00000533085	?	4
RP11-734K2.4	ENST00000605233	?	7

Locus 305 (rs6561333)

SNiPAcard

Block annotations

Block info

genomic range	chr13:47,420,312-47,423,291
block size	2,980 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.566 [-1.342 – -0.171]	gene(s) hit or close-by	HTR2A, HTR2A-AS1
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	–
GERP++	μ = -0.212 [-0.764 – 0.108]	potentially regulated gene(s)	–
CADD score	μ = 2.501 [1.29 – 3.533]	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HTR2A-AS1	upstream gene variant	2987	ENST00000455126	?	?	1
HTR2A-AS1	upstream gene variant	2996	ENST00000452352	?	?	1
HTR2A-AS1	upstream gene variant	2996	ENST00000430913	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HTR2A	ENST00000378688	?	ENSP00000367959	3
HTR2A	ENST00000542664	NM_000621.4	ENSP00000437737	3
HTR2A	ENST00000543956	NM_001165947.2	ENSP00000441861	3

Locus 306 (rs4420638)

SNiPAcard

Block annotations

Block info

genomic range	chr19:45,422,160-45,424,514
block size	2,355 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.205 [-0.072 – 0.427]	**gene(s) hit or close-by**	APOC1, CTB-129P6.7
phastCons	μ = 0.002 [0 – 0.005]	**eQTL gene(s)**	–
GERP++	μ = 0.441 [0 – 1.22]	**potentially regulated gene(s)**	–
CADD score	μ = 2.533 [0.94 – 4.059]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Longevity (90 years and older)	$<3.00\times10^{-36}$	GWAS Catalog	24688116	1
Longevity (85 years and older)	$<2.00\times10^{-26}$	GWAS Catalog	24688116	1
HDL cholesterol	$<2.00\times10^{-21}$	GWAS Catalog	24097068	1
Cholesterol, total	$<1.00\times10^{-149}$	GWAS Catalog	24097068	1
LDL cholesterol	$<2.00\times10^{-178}$	GWAS Catalog	24097068	1
Lipid traits	$<1.00\times10^{-14}$	GWAS Catalog	24023261	1
Age-related macular degeneration	$<2.00\times10^{-20}$	GWAS Catalog	23455636	1
Alzheimer's disease biomarkers	$<4.00\times10^{-12}$	GWAS Catalog	23419831	1
Alzheimer's disease	$<8.00\times10^{-149}$	GWAS Catalog	22832961	1
Cognitive decline	$<4.00\times10^{-27}$	GWAS Catalog	22054870	1
Alzheimer's disease (age of onset)	$<1.00\times10^{-12}$	GWAS Catalog	22005931	1
Lipoprotein-associated phospholipase A2 activity and mass	$<5.00\times10^{-30}$	GWAS Catalog	22003152	1
Longevity	$<2.00\times10^{-16}$	GWAS Catalog	21740922	1
C-reactive protein levels	$<9.00\times10^{-139}$	GWAS Catalog	21300955	1
C-reactive protein	$<3.00\times10^{-7}$	GWAS Catalog	21196492	1
LDL cholesterol	$<2.00\times10^{-40}$	GWAS Catalog	20864672	1
HDL cholesterol	$<4.00\times10^{-21}$	GWAS Catalog	20686565	1
Cholesterol, total	$<5.00\times10^{-111}$	GWAS Catalog	20686565	1
LDL cholesterol	$<9.00\times10^{-147}$	GWAS Catalog	20686565	1
Lipoprotein-associated phospholipase A2 activity and mass	$<6.00\times10^{-24}$	GWAS Catalog	20442857	1
C-reactive protein	$<5.00\times10^{-27}$	GWAS Catalog	19567438	1
Quantitative traits	$<2.00\times10^{-6}$	GWAS Catalog	19197348	1
LDL cholesterol	$<4.00\times10^{-27}$	GWAS Catalog	19060906	1
LDL cholesterol	$<2.00\times10^{-7}$	GWAS Catalog	18802019	1
LDL cholesterol	$<1.00\times10^{-20}$	GWAS Catalog	18262040	1
LDL cholesterol	$<1.00\times10^{-60}$	GWAS Catalog	18193044	1

LDL cholesterol	$<3.00\times10^{-43}$	GWAS Catalog	18193043		1
Alzheimer's disease	$<2.00\times10^{-44}$	GWAS Catalog	17998437		1
Alzheimer's disease	$<1.00\times10^{-39}$	GWAS Catalog	17975299		1
Alzheimer's disease (late onset)	$<1.00\times10^{-39}$	GWAS Catalog	17474819		1
Triglycerides	$<3.00\times10^{-13}$	GWAS Catalog	17463246		1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
APOC1	downstream gene variant	247	ENST00000588750	NM_001645.3	ENSP00000465356	4
APOC1	downstream gene variant	2611	ENST00000589078	?	ENSP00000465710	5
APOC1	downstream gene variant	245	ENST00000592885	?	ENSP00000467368	4
APOC1	downstream gene variant	380	ENST00000592535	?	ENSP00000468276	4
APOC1	downstream gene variant	247	ENST00000590334	?	ENSP00000465190	4
APOC1	downstream gene variant	243	ENST00000586638	?	ENSP00000466146	4
APOC1	downstream gene variant	246	ENST00000589781	?	ENSP00000467504	4
APOC1	downstream gene variant	240	ENST00000592176	?	ENSP00000466227	4
APOC1	downstream gene variant	243	ENST00000588802	?	ENSP00000468029	4
CTB-129P6.7	downstream gene variant	3935	ENST00000623895	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOC1	ENST00000588802	?	ENSP00000468029	1
APOC1	ENST00000589781	?	ENSP00000467504	1
APOC1	ENST00000592885	?	ENSP00000467368	1
APOC1	ENST00000592535	?	ENSP00000468276	1
APOC1	ENST00000588750	NM_001645.3	ENSP00000465356	1
APOC1	ENST00000586638	?	ENSP00000466146	1
APOC1	ENST00000592176	?	ENSP00000466227	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOC1	ENST00000590334	?	ENSP00000465190	1

Locus 307 (rs6032067)

SNiPAcard

Block annotations

Block info

genomic range	chr20:43,763,968-43,888,106
block size	124,139 bp
variant count	150 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.456 [-3.331 – 1.497]	gene(s) hit or close-by	PI3, RP1-300I2.2, RP1-300I2.3, SEMG1, SEMG2, SLPI
phastCons	μ = 0.073 [0 – 0.822]	eQTL gene(s)	DBNDD2, KCNS1, MATN4, PI3, RP3-453C12.8, SEMG1, SLPI, SYS1-DBNDD2, WFDC12, WFDC5
GERP++	μ = -0.394 [-6.16 – 3.49]	potentially regulated gene(s)	DBNDD2, PABPC1L, PI3, RP3-453C12.8, RPL5P2, RPS2P7, SEMG2, SPINT3, STK4, STK4, SYS1, WFDC11, WFDC13, WFDC2, WFDC5, WFDC9
CADD score	μ = 3.639 [0.002 – 14.69]	disease gene(s)	STK4

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM099561	1

Disease gene annotation

gene	trait	source DB	source entry/link	
STK4	T-CELL IMMUNODEFICIENCY, RECURRENT INFECTIONS, AND AUTOIMMUNITY [...]	OMIM	MIM:614868	
STK4	Combined immunodeficiency due to STK4 deficiency	OrphaNet	OrphaNet:314689	

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PI3	missense variant	ENST00000243924	NM_002638.3	ENSP00000243924	2	2			2
SEMG2	missense variant	ENST00000372769	NM_003008.2	ENSP00000361855	2	2			2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MATN4	?	ENSG00000124159	pancreas	1.67×10^{-7} (p-value)	GTEx Portal V6	148
SLPI	?	ENSG00000124107	sun exposed skin	1.26×10^{-14} (p-value)	GTEx Portal V6	150
KCNS1	?	ENSG00000124134	sun exposed skin	1.07×10^{-5} (p-value)	GTEx Portal V6	9
SLPI	?	ENSG00000124107	esophagus mucosa	2.65×10^{-17} (p-value)	GTEx Portal V6	150
SLPI	?	ENSG00000124107	unexposed skin	9.30×10^{-11} (p-value)	GTEx Portal V6	66

WFDC5	?	ENSG00000175121	unexposed skin	2.65×10-6 (p-value)	GTEx Portal V6	10
KCNS1	ENST00000306117	ILMN_1761903	blood	1.92×10-5 (p-value)	Westra et al.	28
DBNDD2	ENST00000372717	ILMN_1730612	skin	3.96×10-7 (p-value)	MuTHER consortium	47
DBNDD2	ENST00000372712					
DBNDD2	ENST00000372720					
DBNDD2	ENST00000372710					
SYS1-DBNDD2	ENST00000475242					
DBNDD2	ENST00000360981					
DBNDD2	ENST00000443296					
DBNDD2	ENST00000372722					
DBNDD2	ENST00000372723					
DBNDD2	ENST00000357275					
SYS1-DBNDD2	ENST00000419593					
WFDC5	ENST00000372789	ILMN_2079042	skin	4.65×10-7 (p-value)	MuTHER consortium	43
WFDC5	ENST00000307971					
KCNS1	?	ENSG00000124134	adrenal gland	3.84×10-6 (p-value)	GTEx Portal V6	41
SEMG1	?	ENSG00000124233	adrenal gland	3.08×10-6 (p-value)	GTEx Portal V6	113
RP3-453C12.8	?	ENSG00000232880	tibial artery	1.01×10-5 (p-value)	GTEx Portal V6	4
PI3	?	ENSG00000124102	blood	5.52×10-6 (p-value)	GTEx Portal V6	2
WFDC12	?	ENSG00000168703	transverse colon	6.65×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000296292	1	ENCP00000031705	WFDC5
ENCE00000296259	1	ENCP00000031698	PABPC1L
ENCE00000296165	1	ENCP00000031720	SYS1
		ENCP00000031724	DBNDD2
		ENCP00000031716	SYS1
		ENCP00000031722	RP3-453C12.8
		ENCP00000031733	RPL5P2
ENCE00000296320	2	ENCP00000031707	PI3
ENCE00000296274	1	ENCP00000031741	WFDC11
		ENCP00000031702	STK4 STK4
ENCE00000296369	1	ENCP00000031709	SEMG2
		ENCP00000031740	WFDC9 RPS2P7
		ENCP00000031742	WFDC13
		ENCP00000031734	SPINT3
		ENCP00000031731	WFDC2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000009720 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3, Max, Rad21, Gabp, H2AZ, CTCFL, CTCF

		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Gabp, Rad21, CTCF, H3K27me3, DNase1
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF, H3K27me3
		skin (NHEK)	CTCF, DNase1
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF, DNase1
		cervix (HeLa-S3)	DNase1, CTCF, Gabp
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3, CTCF
		A549	CTCF, H3K27me3
ENSR00001665870 (open chromatin region)	1	A549	H3K27me3
ENSR00001665871 (open chromatin region)	2	A549	H3K27me3
ENSR00001449109 (promoter flanking region)	2	NHLF	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K27me3
		muscle (HSMM)	DNase1
		A549	H3K27me3
		nervous (NH-A)	DNase1
ENSR00001449110 (open chromatin region)	2	endothelium (HUVEC)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
		breast (HMEC)	DNase1
ENSR00000401582 (TF binding site)	6	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
ENSR00000401583 (TF binding site)	6	blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001665874 (open chromatin region)	1	A549	H3K27me3
		blood (K562)	H3K27me3
ENSR00001449111 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3
		A549	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00000649547 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Yy1, CTCF, Rad21, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF
		skin (NHEK)	CTCF

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PI3	2	ENST00000243924	hsa-miR-28-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PI3	upstream gene variant, downstream gene variant	89	ENST00000243924	NM_002638.3	ENSP00000243924	22
RP1-300I2.2	upstream gene variant, downstream gene variant	1809	ENST00000461248	?	?	8
RP1-300I2.2	upstream gene variant, downstream gene variant	1883	ENST00000486547	?	?	8
RP1-300I2.3	upstream gene variant, downstream gene variant	758	ENST00000422041	?	?	7
SEMG1	upstream gene variant, downstream gene variant	406	ENST00000372781	NM_003007.3	ENSP00000361867	10
SEMG2	upstream gene variant, downstream gene variant	177	ENST00000372769	NM_003008.2	ENSP00000361855	10
SLPI	downstream gene variant, upstream gene variant	791	ENST00000338380	NM_003064.3	ENSP00000342082	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
SEMG2	ENST00000372769	NM_003008.2	ENSP00000361855	Q	caG/caA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PI3	ENST00000243924	NM_002638.3	ENSP00000243924	6
SEMG1	ENST00000372781	NM_003007.3	ENSP00000361867	1
SEMG2	ENST00000372769	NM_003008.2	ENSP00000361855	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PI3	ENST00000243924	NM_002638.3	ENSP00000243924	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SEMG1	ENST00000372781	NM_003007.3	ENSP00000361867	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP1-300I2.2	ENST00000486547	?	1
RP1-300I2.2	ENST00000461248	?	1

Locus 308 (rs3737276)

SNiPAcard

Block annotations

Block info

genomic range	chr1:248,022,712-248,027,609
block size	4,898 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.892 [-2.185 – 0.498]	gene(s) hit or close-by	TRIM58
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	OR2T8, OR2W3, TRIM58
GERP++	μ = -1.524 [-7.78 – 1.54]	potentially regulated gene(s)	–
CADD score	μ = 2.793 [0.034 – 5.429]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
OR2W3	?	ENSG00000238243	tibial nerve	1.89×10^{-8} (p-value)	GTEx Portal V6	10
TRIM58	?	ENSG00000162722	tibial nerve	1.97×10^{-9} (p-value)	GTEx Portal V6	10
OR2T8	?	ENSG00000177462	tibial nerve	4.60×10^{-7} (p-value)	GTEx Portal V6	7
TRIM58	?	ENSG00000162722	blood	2.01×10^{-6} (p-value)	GTEx Portal V6	8
TRIM58	?	ENSG00000162722	muscularis mucosae	4.59×10^{-6} (p-value)	GTEx Portal V6	2
TRIM58	?	ENSG00000162722	unexposed skin	1.60×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000290792 (open chromatin region)	3	blood (K562)	DNase1, H3K9ac, H3K79me2, H3K4me3, H3K4me2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TRIM58	ENST00000366481	NM_015431.3	ENSP00000355437	D	gaT/gaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TRIM58	ENST00000366481	NM_015431.3	ENSP00000355437	9

Locus 309 (rs7097780)

SNiPAcard

Block annotations

Block info

genomic range	chr10:6,008,958-6,027,791
block size	18,834 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.162 [-1.666 – 0.88]	gene(s) hit or close-by	IL15RA
phastCons	μ = 0.048 [0 – 0.265]	eQTL gene(s)	IL15RA
GERP++	μ = -1.081 [-4.45 – 0.737]	potentially regulated gene(s)	GDI2, RP11-318E3.4
CADD score	μ = 3.814 [0.013 – 12.78]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL15RA	?	ENSG00000134470	transformed fibroblasts	9.34×10^{-8} (p-value)	GTEx Portal V6	15
IL15RA	?	ENSG00000134470	blood	4.89×10^{-9} (p-value)	GTEx Portal V6	14

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000065993	1	ENCP00000007580	GDI2
		ENCP00000007581	RP11-318E3.4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000350158 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	H3K27me3
		blood (K562)	DNase1
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	H3K27ac, H3K4me1, H3K36me3, DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	H3K27me3
		blood (GM12878)	H3K4me3, H3K79me2, H3K27ac, H3K4me2
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL15RA	upstream gene variant	2570	ENST00000397248	NM_001256765.1	ENSP00000380421	2
IL15RA	upstream gene variant	656	ENST00000532039	?	ENSP00000432691	4
IL15RA	upstream gene variant	3163	ENST00000447291	?	ENSP00000405128	2
IL15RA	upstream gene variant	2002	ENST00000379974	?	?	2
IL15RA	upstream gene variant	2967	ENST00000453922	?	ENSP00000405107	2
IL15RA	upstream gene variant	1994	ENST00000525219	NM_001243539.1	ENSP00000431529	2

IL15RA	upstream gene variant	2570	ENST00000620345	?	ENSP00000479839	2
IL15RA	upstream gene variant	2570	ENST00000622442	?	ENSP00000480949	2
IL15RA	upstream gene variant	2674	ENST00000379971	?	ENSP00000369306	2
IL15RA	upstream gene variant	2136	ENST00000534292	?	?	2
IL15RA	upstream gene variant	1994	ENST00000379972	?	?	2
IL15RA	upstream gene variant	2570	ENST00000397251	?	ENSP00000380423	2
IL15RA	upstream gene variant	2659	ENST00000528354	NM_172200.2	ENSP00000435454	2
IL15RA	upstream gene variant	2689	ENST00000397255	?	ENSP00000380426	2
IL15RA	upstream gene variant	2346	ENST00000397246	?	ENSP00000380420	2
IL15RA	upstream gene variant	2591	ENST00000379977	NM_002189.3	ENSP00000369312	2
IL15RA	upstream gene variant	2570	ENST00000618528	?	ENSP00000479938	2
IL15RA	upstream gene variant	2659	ENST00000429135	?	ENSP00000395113	2
IL15RA	upstream gene variant	2570	ENST00000620865	?	ENSP00000478525	2
IL15RA	upstream gene variant	2659	ENST00000397250	?	ENSP00000380422	2
IL15RA	upstream gene variant	2689	ENST00000530685	?	ENSP00000435995	2
IL15RA	upstream gene variant	2143	ENST00000532948	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL15RA	ENST00000622442	?	ENSP00000480949	12
IL15RA	ENST00000525219	NM_001243539.1	ENSP00000431529	12
IL15RA	ENST00000528354	NM_172200.2	ENSP00000435454	12
IL15RA	ENST00000534292	?	?	12
IL15RA	ENST00000379971	?	ENSP00000369306	12
IL15RA	ENST00000397248	NM_001256765.1	ENSP00000380421	12
IL15RA	ENST00000379972	?	?	12
IL15RA	ENST00000397250	?	ENSP00000380422	12
IL15RA	ENST00000620345	?	ENSP00000479839	12
IL15RA	ENST00000453922	?	ENSP00000405107	12
IL15RA	ENST00000618528	?	ENSP00000479938	12
IL15RA	ENST00000397255	?	ENSP00000380426	12
IL15RA	ENST00000397246	?	ENSP00000380420	12
IL15RA	ENST00000379977	NM_002189.3	ENSP00000369312	12
IL15RA	ENST00000530685	?	ENSP00000435995	12
IL15RA	ENST00000429135	?	ENSP00000395113	12
IL15RA	ENST00000397251	?	ENSP00000380423	12
IL15RA	ENST00000379974	?	?	12
IL15RA	ENST00000620865	?	ENSP00000478525	12
IL15RA	ENST00000532948	?	?	12

Locus 309 (sentinel SNP rs7097780 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 310 (rs11260013)

SNiPAcard

Block annotations

Block info

genomic range	chr19:7,758,154-7,759,715
block size	1,562 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.585 [-2.035 – 0.325]	gene(s) hit or close-by	FCER2
phastCons	μ = 0.005 [0 – 0.02]	eQTL gene(s)	FCER2
GERP++	μ = -0.886 [-3.83 – 1.32]	potentially regulated gene(s)	CCL25
CADD score	μ = 2.513 [0.088 – 6.7]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCER2	?	ENSG00000104921	blood	1.13×10^{-7} (p-value)	GTEx Portal V6	11
FCER2	ENST00000597921	ILMN_1662451	b-cell	3.36×10^{-5} (p-value)	Fairfax et al.	1
FCER2	ENST00000346664					
FCER2	ENST00000598803					
FCER2	ENST00000597934					
FCER2	ENST00000597312					
FCER2	ENST00000360067					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
EBF1	MA0154.2	1	no	0.009	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000223034	1	ENCP00000024014	CCL25

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000342882 (TF binding site)	2	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3
		blood (GM12878)	Tr4, Yy1, PolII, DNase1, H3K79me2, H3K4me3, Pax5, H3K4me2, H3K36me3
		blood (K562)	H3K27me3
		A549	H3K27me3
ENSR00000342883 (TF binding site)	3	embryonic stem cell (H1ESC)	H3K27me3
		blood (GM12878)	H3K4me2, H3K36me3, EBF1, PolII, DNase1, H3K79me2, H3K4me3, H3K27ac
		A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCER2	ENST00000597921	?	ENSP00000471974	12
FCER2	ENST00000593418	?	ENSP00000472067	12
FCER2	ENST00000360067	NM_001207019.2	ENSP00000353178	12
FCER2	ENST00000346664	NM_002002.4, NM_001220500.1	ENSP00000264072	12
FCER2	ENST00000597934	?	?	12
FCER2	ENST00000598803	?	?	12
FCER2	ENST00000597312	?	?	12

Locus 311 (rs4356932)

SNiPAcard

Block annotations

Block info

genomic range	chr4:76,922,988-76,997,258
block size	74,271 bp
variant count	265 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.627 [-4.521 – 1.846]	gene(s) hit or close-by	ART3, CXCL10, CXCL11, CXCL9, RP11-630D6.1, RP11-630D6.5
phastCons	μ = 0.100 [0 – 1]	eQTL gene(s)	ART3, CXCL10, CXCL11, NAAA, NUP54, SLC6A20
GERP++	μ = -0.174 [-6.98 – 3.47]	potentially regulated gene(s)	ART3, CCDC158, CXCL11, CXCL9, G3BP2, NAAA, NUP54, PPEF2, RP11-630D6.5, SCARB2, SDAD1, USO1
CADD score	μ = 3.773 [0.088 – 15.97]	disease gene(s)	SLC6A20, SCARB2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SLC6A20	HYPERGLYCINURIA	OMIM	MIM:138500
SLC6A20	IMINOGLYCINURIA	OMIM	MIM:242600
SCARB2	EPILEPSY, PROGRESSIVE MYOCLONIC, 4, WITH OR WITHOUT RENAL [...]	OMIM	MIM:254900
SLC6A20	IMINOGLYCINURIA	OrphaNet	OrphaNet:42062
SCARB2	Gaucher disease type 1	OrphaNet	OrphaNet:77259
SCARB2	Action myoclonus - renal failure syndrome	OrphaNet	OrphaNet:163696
SCARB2	Unverricht-Lundborg disease	OrphaNet	OrphaNet:308

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NAAA	ENST00000286733	ILMN_2391512	skin	1.23×10^{-6} (p-value)	MuTHER consortium	55
NAAA	ENST00000602782		blood	2.46×10^{-35} (p-value)	MuTHER consortium	55
NAAA	ENST00000513045		adipocyte	8.04×10^{-14} (p-value)	MuTHER consortium	55
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
CXCL10	ENST00000306602	ILMN_1791759	blood	4.51×10^{-11} (p-value)	MuTHER consortium	55
NAAA	?	ENSG00000138744	atrial appendage	2.28×10^{-10} (p-value)	GTEx Portal V6	255
NAAA	?	ENSG00000138744	transformed fibroblasts	5.01×10^{-41} (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	blood	3.49×10^{-22} (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	breast	2.46×10^{-17} (p-value)	GTEx Portal V6	256
NAAA	ENST00000513045	ILMN_2391512	monocyte	6.55×10^{-14} (p-value)	Fairfax et al.	10
NAAA	ENST00000602782		b-cell	4.77×10^{-11} (p-value)	Fairfax et al.	10
NAAA	ENST00000286733					

NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
?	?	ILMN_1668605	monocyte	8.79×10-11 (p-value)	Fairfax et al.	10
			b-cell	1.35×10-10 (p-value)		
CXCL10	ENST00000306602	ILMN_1791759	monocyte	3.84×10-4 (p-value)	Fairfax et al.	10
NAAA	?	ENSG00000138744	skeletal muscle	3.24×10-37 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	EBV lymphocytes	6.31×10-8 (p-value)	GTEx Portal V6	250
CXCL10	?	ENSG00000169245	EBV lymphocytes	5.98×10-6 (p-value)	GTEx Portal V6	1
NAAA	?	ENSG00000138744	blood	4.02×10-8 (q-value)	SeeQTL DB (HapMap)	25
CXCL10	?	ENSG00000169245	blood	2.54×10-2 (q-value)	SeeQTL DB (HapMap)	5
NAAA	?	ENSG00000138744	ovary	1.86×10-11 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	sun exposed skin	8.67×10-36 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	unexposed skin	1.46×10-18 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	aorta	2.26×10-20 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	left ventricle	3.49×10-13 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	subcutaneous adipocytes	2.09×10-28 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	stomach	5.02×10-11 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	visceral adipocytes	1.47×10-15 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	cortex	5.43×10-8 (p-value)	GTEx Portal V6	212
ART3	?	ENSG00000156219	tibial nerve	1.44×10-63 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	tibial nerve	2.52×10-24 (p-value)	GTEx Portal V6	256
NUP54	?	ENSG00000138750	tibial nerve	1.17×10-5 (p-value)	GTEx Portal V6	4
NAAA	?	ENSG00000138744	esophagus mucosa	8.13×10-21 (p-value)	GTEx Portal V6	256
ART3	?	ENSG00000156219	pituitary	2.55×10-11 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	pituitary	2.63×10-6 (p-value)	GTEx Portal V6	22
NUP54	?	ENSG00000138750	pituitary	7.59×10-9 (p-value)	GTEx Portal V6	243
NAAA	?	ENSG00000138744	testis	1.89×10-17 (p-value)	GTEx Portal V6	256
ART3	?	ENSG00000156219	pancreas	4.23×10-42 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	pancreas	1.06×10-22 (p-value)	GTEx Portal V6	256
CXCL11	?	ENSG00000169248	pancreas	4.64×10-21 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	muscularis mucosae	4.58×10-30 (p-value)	GTEx Portal V6	256
ART3	?	ENSG00000156219	muscularis mucosae	1.11×10-5 (p-value)	GTEx Portal V6	1
NAAA	?	ENSG00000138744	lung	1.67×10-23 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	tibial artery	9.98×10-31 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	thyroid	2.39×10-13 (p-value)	GTEx Portal V6	256
NUP54	?	ENSG00000138750	thyroid	5.69×10-6 (p-value)	GTEx Portal V6	17

NAAA	?	ENSG00000138744	transverse colon	1.13×10-9 (p-value)	GTEx Portal V6	255
NAAA	?	ENSG00000138744	terminal ileum	7.67×10-8 (p-value)	GTEx Portal V6	233
NAAA	?	ENSG00000138744	coronary artery	5.31×10-15 (p-value)	GTEx Portal V6	256
ART3	?	ENSG00000156219	sigmoid colon	3.42×10-8 (p-value)	GTEx Portal V6	244
NAAA	?	ENSG00000138744	sigmoid colon	2.81×10-13 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	vagina	3.01×10-8 (p-value)	GTEx Portal V6	234
NAAA	?	ENSG00000138744	gastroesophageal junction	1.32×10-12 (p-value)	GTEx Portal V6	256
NAAA	?	ENSG00000138744	adrenal gland	3.14×10-18 (p-value)	GTEx Portal V6	256
NAAA	ENST00000602782	214765_s_at	blood	1.40×10-23 (p-value)	Dixon et al.	16
NAAA	ENST00000286733					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
NAAA	ENST00000286733	227135_at	blood	7.30×10-21 (p-value)	Dixon et al.	16
NAAA	ENST00000511606					
CXCL10	ENST00000306602	204533_at	blood	2.60×10-9 (p-value)	Dixon et al.	14
NAAA	?	ENSG00000138744	frontal cortex	5.99×10-7 (p-value)	GTEx Portal V6	91
?	?	ILMN_2067890	monocyte	4.60×10-64 (p-value)	Zeller et al.	14
NAAA	ENST00000286733	ILMN_2391512	monocyte	5.69×10-38 (p-value)	Zeller et al.	14
NAAA	ENST00000602782					
NAAA	ENST00000513045					
NAAA	ENST00000507956					
NAAA	ENST00000505594					
NAAA	ENST00000511606					
?	?	ILMN_1668605	monocyte	2.02×10-35 (p-value)	Zeller et al.	14
?	?	ILMN_2285568	monocyte	1.10×10-33 (p-value)	Zeller et al.	14
CXCL10	ENST00000306602	ILMN_1791759	monocyte	2.69×10-12 (p-value)	Zeller et al.	9
NAAA	?	ENSG00000138744	cerebellum	3.44×10-6 (p-value)	GTEx Portal V6	12
NAAA	?	ENSG00000138744	uterus	4.67×10-7 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
SLC6A20	ENST00000353278	ILMN_1698585	chr3	monocyte	6.46×10-13 (p-value)	Zeller et al.	11
SLC6A20	ENST00000456124						
SLC6A20	ENST00000358525						

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000655593	1	ENST00000504914, ENST00000513353, ENST00000341029, ENST00000513122	ART3

FFCP00000655602	1	ENST00000504914, ENST00000513353, ENST00000341029, ENST00000513122	ART3

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000041424		
ENCP00000041429		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382631	1	ENCP00000041405	G3BP2
		ENCP00000041421	SDAD1
		ENCP00000041417	NAAA
		ENCP00000041404	G3BP2
ENCE00000382671	1	ENCP00000041434	ART3
		ENCP00000041413	PPEF2
		ENCP00000041412	USO1
		ENCP00000041414	PPEF2
ENCE00000382734	1	ENCP00000041421	SDAD1
		ENCP00000041417	NAAA
		ENCP00000041440	NUP54
ENCE00000382736	1	ENCP00000041421	SDAD1
		ENCP00000041417	NAAA
ENCE00000382737	2	ENCP00000041422	RP11-630D6.5
		ENCP00000041421	SDAD1
		ENCP00000041443	SCARB2
		ENCP00000041417	NAAA
		ENCP00000041440	NUP54
ENCE00000382672	1	ENCP00000041434	ART3
		ENCP00000041413	PPEF2
		ENCP00000041412	USO1
		ENCP00000041414	PPEF2
ENCE00000382653	2	ENCP00000041411	USO1
		ENCP00000041425	ART3
ENCE00000382696	1	ENCP00000041438	NUP54
		ENCP00000041415	NAAA
		ENCP00000041452	CCDC158
ENCE00000382738	2	ENCP00000041443	SCARB2
		ENCP00000041421	SDAD1
		ENCP00000041417	NAAA

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001685894 (enhancer)	1	HSMMtube	H3K27me3
ENSR00001243218 (promoter flanking region)	2	NHLF	H3K27ac
		embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K27ac, H3K4me2
		blood (GM12878)	H2AZ, H3K27ac, H3K4me2
		lung (IMR90)	DNase1, H3K18ac, H3K27ac, H3K4ac, H2BK120ac, H2BK20ac, H4K91ac, H3K56ac
		skin (NHEK)	DNase1

ENSR00001243224 (promoter flanking region)	2	HSMMtube	H3K27me3
		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me3
		monocytes (Monocytes-CD14+)	H3K4me3
		endothelium (HUVEC)	H3K4me2
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K4me2
ENSR00001685897 (enhancer)	2	monocytes (Monocytes-CD14+)	DNase1
		Osteobl	H3K27me3
		A549	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001243225 (enhancer)	1	endothelium (HUVEC)	Cjun, DNase1
		HSMMtube	H3K27me3
ENSR00001243226 (enhancer)	1	embryonic stem cell (H1ESC)	CTCF, Yy1, DNase1
		endothelium (HUVEC)	H3K36me3
		HSMMtube	H3K27me3
		skin (NHEK)	CTCF
ENSR00001685898 (CTCF binding site)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	CTCF, Yy1, DNase1
		HSMMtube	H3K27me3
		skin (NHEK)	CTCF
ENSR00001243231 (enhancer)	1	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	Cjun, BATF, DNase1
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
ENSR00001432212 (enhancer)	7	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	H3K4me2
		monocytes (Monocytes-CD14+)	H3K4me1
		endothelium (HUVEC)	H3K27me3
		blood (GM12878)	Cjun, BATF, MEF2A, Jund
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K4me1, H3K4me2
ENSR00001685900 (promoter flanking region)	4	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	H3K4me2, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, DNase1
		endothelium (HUVEC)	H3K27me3
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	DNase1, H3K4me1, H3K4me2
ENSR00001432213 (CTCF binding site)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	DNase1, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		endothelium (HUVEC)	H3K27me3
		nervous (NH-A)	H3K27me3

	A549	H3K27me3
	skin (NHEK)	DNase1, H3K4me1, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ART3	upstream gene variant	941	ENST00000510423	?	ENSP00000425327	2
ART3	upstream gene variant	352	ENST00000341029	NM_001130017.2	ENSP00000343843	2
ART3	upstream gene variant	336	ENST00000513353	?	ENSP00000421345	2
ART3	upstream gene variant	76	ENST00000504112	?	?	2
ART3	upstream gene variant	1249	ENST00000504914	?	ENSP00000421431	2
ART3	upstream gene variant	401	ENST00000513122	?	ENSP00000422287	2
ART3	upstream gene variant	352	ENST00000510669	?	?	2
ART3	upstream gene variant	76	ENST00000395688	?	?	2
ART3	upstream gene variant	76	ENST00000506667	?	?	2
ART3	upstream gene variant	400	ENST00000355810	NM_001130016.2	ENSP00000348064	2
ART3	upstream gene variant	403	ENST00000513494	?	?	2
ART3	upstream gene variant	400	ENST00000349321	NM_001179.5	ENSP00000304313	2
CXCL10	downstream gene variant, upstream gene variant	452	ENST00000306602	NM_001565.3	ENSP00000305651	21
CXCL11	downstream gene variant, upstream gene variant	38	ENST00000503860	?	ENSP00000425819	73
CXCL11	downstream gene variant, upstream gene variant	208	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	46
CXCL9	upstream gene variant	560	ENST00000264888	NM_002416.1	ENSP00000354901	3
RP11-630D6.1	downstream gene variant, upstream gene variant	117	ENST00000477971	?	?	48
RP11-630D6.5	downstream gene variant	2106	ENST00000501239	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ART3	ENST00000513494	?	?	2
ART3	ENST00000341029	NM_001130017.2	ENSP00000343843	261
ART3	ENST00000504112	?	?	1
ART3	ENST00000513353	?	ENSP00000421345	261
ART3	ENST00000504914	?	ENSP00000421431	261
ART3	ENST00000510669	?	?	261
ART3	ENST00000513122	?	ENSP00000422287	261
ART3	ENST00000506667	?	?	1
ART3	ENST00000395688	?	?	1
ART3	ENST00000349321	NM_001179.5	ENSP00000304313	2
ART3	ENST00000510423	?	ENSP00000425327	2
ART3	ENST00000355810	NM_001130016.2	ENSP00000348064	2
CXCL10	ENST00000306602	NM_001565.3	ENSP00000305651	7
CXCL11	ENST00000503860	?	ENSP00000425819	25

CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	15
CXCL9	ENST00000264888	NM_002416.1	ENSP00000354901	2
RP11-630D6.5	ENST00000501239	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL10	ENST00000306602	NM_001565.3	ENSP00000305651	2
CXCL11	ENST00000503860	?	ENSP00000425819	1
CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	11
CXCL9	ENST00000264888	NM_002416.1	ENSP00000354901	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CXCL11	ENST00000503860	?	ENSP00000425819	2
CXCL11	ENST00000306621	NM_005409.4, NM_001302123.1	ENSP00000306884	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-630D6.1	ENST00000477971	?	1

Locus 312 (rs2236293)

SNiPAcard

Block annotations

Block info

genomic range	chr9:35,779,571-35,860,942
block size	81,372 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.196 [-1.18 – 3.035]	gene(s) hit or close-by	FAM221B, HINT2, LINC00950, NPR2, OR13E1P, RP11-112J3.16, SPAG8, TMEM8B
phastCons	μ = 0.072 [0 – 0.999]	eQTL gene(s)	GBA2, HRCT1, MSMP, NPR2, RP11-112J3.16, RP11-182N22.8, SPAG8, TMEM8B, TPM2
GERP++	μ = 0.459 [-5.31 – 4.04]	potentially regulated gene(s)	–
CADD score	μ = 6.165 [0.737 – 17.84]	disease gene(s)	NPR2, GBA2, TPM2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
2-aminobutyrate	5.07×10^{-5}	Metabolomics GWAS Server	24816252	3

Disease gene annotation

gene	trait	source DB	source entry/link
NPR2	acromesomelic dysplasia Maroteaux type (AMDM)	DECIPHER	MIM:602875
NPR2	ACROMESOMELIC DYSPLASIA, MAROTEAUX TYPE	OMIM	MIM:602875
NPR2	EPIPHYSEAL CHONDRODYSPLASIA, MIURA TYPE	OMIM	MIM:615923
GBA2	SPASTIC PARAPLEGIA 46, AUTOSOMAL RECESSIVE	OMIM	MIM:614409
TPM2	ARTHROGRYPOSIS, DISTAL, TYPE 1A	OMIM	MIM:108120
TPM2	ARTHROGRYPOSIS, DISTAL, TYPE 2B	OMIM	MIM:601680
TPM2	NEMALINE MYOPATHY 4	OMIM	MIM:609285
NPR2	ACROMESOMELIC DYSPLASIA, MAROTEAUX TYPE	OrphaNet	OrphaNet:40
NPR2	Tall stature - scoliosis - macrodactyly of the great toes	OrphaNet	OrphaNet:329191
GBA2	Autosomal recessive cerebellar ataxia with late-onset spasticity	OrphaNet	OrphaNet:352641
GBA2	Autosomal recessive spastic paraplegia type 46	OrphaNet	OrphaNet:320391
TPM2	Sheldon-Hall syndrome	OrphaNet	OrphaNet:1147
TPM2	Congenital fiber-type disproportion myopathy	OrphaNet	OrphaNet:2020
TPM2	Trismus - pseudocamptodactyly	OrphaNet	OrphaNet:3377
TPM2	Cap myopathy	OrphaNet	OrphaNet:171881
TPM2	Typical nemaline myopathy	OrphaNet	OrphaNet:171436
TPM2	Childhood-onset nemaline myopathy	OrphaNet	OrphaNet:171439
TPM2	Digitotalar dysmorphism	OrphaNet	OrphaNet:1146

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HRCT1	ENST00000354323	ILMN_2189675	adipocyte	6.09×10^{-8} (p-value)	MuTHER consortium	5
MSMP	ENST00000436428	ILMN_2115135	blood	1.08×10^{-7} (p-value)	MuTHER consortium	10

MSMP	ENST00000414286					
NPR2	ENST00000421267	ILMN_1681994	skin	1.81×10-5 (p-value)	MuTHER consortium	5
NPR2	ENST00000469249					
NPR2	ENST00000464810					
TMEM8B	?	ENSG00000137103	muscularis mucosae	3.39×10-8 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	lung	1.62×10-10 (p-value)	GTEx Portal V6	17
GBA2	?	ENSG00000070610	atrial appendage	6.96×10-8 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	atrial appendage	8.94×10-7 (p-value)	GTEx Portal V6	5
TMEM8B	?	ENSG00000137103	transformed fibroblasts	8.32×10-10 (p-value)	GTEx Portal V6	17
GBA2	?	ENSG00000070610	blood	1.72×10-8 (p-value)	GTEx Portal V6	14
TMEM8B	?	ENSG00000137103	breast	9.66×10-6 (p-value)	GTEx Portal V6	2
TMEM8B	?	ENSG00000137103	tibial artery	7.01×10-14 (p-value)	GTEx Portal V6	17
NPR2	?	ENSG00000159899	tibial artery	2.55×10-6 (p-value)	GTEx Portal V6	7
MSMP	ENST00000436428	ILMN_2115135	b-cell	5.37×10-7 (p-value)	Fairfax et al.	7
MSMP	ENST00000414286					
GBA2	ENST00000486797	ILMN_1674560	monocyte	1.95×10-5 (p-value)	Fairfax et al.	1
GBA2	ENST00000378103					
GBA2	ENST00000467252					
GBA2	ENST00000378088					
GBA2	ENST00000378094					
NPR2	?	ENSG00000159899	thyroid	1.02×10-5 (p-value)	GTEx Portal V6	4
TMEM8B	?	ENSG00000137103	thyroid	5.05×10-10 (p-value)	GTEx Portal V6	17
RP11-182N22.8	?	ENSG00000234181	thyroid	1.38×10-5 (p-value)	GTEx Portal V6	2
GBA2	?	ENSG00000070610	skeletal muscle	2.77×10-12 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	skeletal muscle	1.27×10-19 (p-value)	GTEx Portal V6	17
RP11-112J3.16	?	ENSG00000227388	skeletal muscle	1.70×10-5 (p-value)	GTEx Portal V6	1
TMEM8B	?	ENSG00000137103	sun exposed skin	6.07×10-11 (p-value)	GTEx Portal V6	17
HRCT1	?	ENSG00000196196	sun exposed skin	1.30×10-5 (p-value)	GTEx Portal V6	1
TMEM8B	?	ENSG00000137103	unexposed skin	8.29×10-9 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	aorta	4.86×10-12 (p-value)	GTEx Portal V6	17
NPR2	?	ENSG00000159899	left ventricle	4.74×10-7 (p-value)	GTEx Portal V6	7
GBA2	?	ENSG00000070610	left ventricle	2.30×10-11 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	left ventricle	1.06×10-7 (p-value)	GTEx Portal V6	8
TMEM8B	?	ENSG00000137103	subcutaneous adipocytes	1.42×10-7 (p-value)	GTEx Portal V6	17
SPAG8	ENST00000489063	ILMN_1754370	monocyte	2.27×10-18 (p-value)	Zeller et al.	4
SPAG8	ENST00000340291					
SPAG8	ENST00000475644					
SPAG8	ENST00000460836					
SPAG8	ENST00000463889					
TMEM8B	?	ENSG00000137103	coronary artery	1.67×10-8 (p-value)	GTEx Portal V6	17

NPR2	?	ENSG00000159899	tibial nerve	2.31×10-6 (p-value)	GTEx Portal V6	9
GBA2	?	ENSG00000070610	tibial nerve	1.01×10-6 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	tibial nerve	1.58×10-14 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	esophagus mucosa	5.83×10-11 (p-value)	GTEx Portal V6	17
HRCT1	?	ENSG00000196196	esophagus mucosa	3.92×10-6 (p-value)	GTEx Portal V6	3
NPR2	?	ENSG00000159899	adrenal gland	4.43×10-8 (p-value)	GTEx Portal V6	14
TMEM8B	?	ENSG00000137103	adrenal gland	1.58×10-7 (p-value)	GTEx Portal V6	9
SPAG8	ENST00000489063	ILMN_1754370	blood	7.98×10-5 (p-value)	Westra et al.	1
SPAG8	ENST00000340291					
SPAG8	ENST00000475644					
SPAG8	ENST00000460836					
SPAG8	ENST00000463889					
TPM2	ENST00000378300	ILMN_1789196	blood	1.10×10-6 (p-value)	Westra et al.	6
TPM2	ENST00000329305					
TPM2	ENST00000378292					
TMEM8B	?	ENSG00000137103	stomach	4.38×10-7 (p-value)	GTEx Portal V6	8
TMEM8B	?	ENSG00000137103	uterus	2.09×10-7 (p-value)	GTEx Portal V6	7

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001470336 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	Ini1, DNase1, Max, USF1
		breast (HMEC)	DNase1
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3
		endothelium (HUVEC)	Max, DNase1, Cjun
		lung (IMR90)	DNase1, H3K36me3
		A549	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FAM221B	downstream gene variant	1762	ENST00000377984	?	ENSP00000367222	1
HINT2	upstream gene variant	2561	ENST00000259667	NM_032593.2	ENSP00000259667	1
HINT2	upstream gene variant	3888	ENST00000471774	?	?	1
HINT2	upstream gene variant	2590	ENST00000472085	?	?	1
HINT2	upstream gene variant	2228	ENST00000474848	?	?	1
HINT2	upstream gene variant	2230	ENST00000474908	?	?	1
HINT2	upstream gene variant	2561	ENST00000461169	?	?	1
HINT2	upstream gene variant	3812	ENST00000490578	?	?	1
LINC00950	upstream gene variant	1111	ENST00000629651	?	?	1
NPR2	upstream gene variant	1933	ENST00000447210	?	ENSP00000393029	1

NPR2	upstream gene variant	4249	ENST00000469249	?	?	1
NPR2	upstream gene variant	1378	ENST00000421267	?	ENSP00000399204	1
NPR2	upstream gene variant	2381	ENST00000448821	?	ENSP00000402902	1
OR13E1P	downstream gene variant, upstream gene variant	503	ENST00000417104	?	?	3
RP11-112J3.16	downstream gene variant	1871	ENST00000574939	?	?	1
RP11-112J3.16	downstream gene variant	4484	ENST00000431981	?	?	1
SPAG8	downstream gene variant	3893	ENST00000463889	?	?	1
SPAG8	downstream gene variant	3895	ENST00000475644	?	ENSP00000418530	1
SPAG8	downstream gene variant	3893	ENST00000340291	NM_172312.1	ENSP00000340982	1
SPAG8	downstream gene variant	3979	ENST00000489063	?	?	1
SPAG8	downstream gene variant	3893	ENST00000460836	?	?	1
TMEM8B	downstream gene variant	2781	ENST00000377988	NM_001042590.2	ENSP00000367227	1
TMEM8B	downstream gene variant	525	ENST00000472010	?	?	2
TMEM8B	downstream gene variant	2780	ENST00000377991	NM_001042589.2	ENSP00000367230	1
TMEM8B	downstream gene variant	390	ENST00000464519	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NPR2	ENST00000464810	?	?	3
NPR2	ENST00000342694	NM_003995.3	ENSP00000341083	3
RP11-112J3.16	ENST00000431981	?	?	3
RP11-112J3.16	ENST00000574939	?	?	1
TMEM8B	ENST00000472010	?	?	5
TMEM8B	ENST00000377988	NM_001042590.2	ENSP00000367227	7
TMEM8B	ENST00000377996	?	ENSP00000367235	8
TMEM8B	ENST00000439587	NM_016446.3	ENSP00000395810	7
TMEM8B	ENST00000464519	?	?	6
TMEM8B	ENST00000473947	?	?	7
TMEM8B	ENST00000377991	NM_001042589.2	ENSP00000367230	7
TMEM8B	ENST00000490199	?	?	7

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FAM221B	ENST00000388950	?	ENSP00000373602	1
FAM221B	ENST00000423537	NM_001012446.3	ENSP00000415299	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TMEM8B	ENST00000377988	NM_001042590.2	ENSP00000367227	1
TMEM8B	ENST00000377996	?	ENSP00000367235	1
TMEM8B	ENST00000439587	NM_016446.3	ENSP00000395810	1

TMEM8B	ENST00000377991	NM_001042589.2	ENSP00000367230	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
LINC00950	ENST00000629651	?	2
TMEM8B	ENST00000464519	?	1

Locus 313 (rs6442112)

SNiPAcard

Block annotations

Block info

genomic range	chr3:48,221,109-48,316,034
block size	94,926 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.964 [-2.748 – -1.105]	gene(s) hit or close-by	CAMP, CDC25A, NDUFB1P1, RNU7-128P, ZNF589
phastCons	μ = 0.002 [0 – 0.007]	eQTL gene(s)	AMT, ARIH2OS, CDC25A, FCF1P2, KLHDC8B, MRPS18AP1, NCKIPSD, NICN1, NME6, QRICH1, SCAP, TMA7, WDR6, ZNF589
GERP++	μ = -0.371 [-1.44 – 0.624]	potentially regulated gene(s)	FBXW12, MAP4, SHISA5, UQCRC1
CADD score	μ = 1.210 [0.443 – 1.735]	disease gene(s)	AMT, KLHDC8B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899	OMIM
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899	OMIM
KLHDC8B	LYMPHOMA, HODGKIN, CLASSIC	OMIM	MIM:236000	OMIM
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860	orphanet
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857	orphanet
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NME6	?	ENSG00000172113	muscularis mucosae	3.38×10-7 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	muscularis mucosae	8.74×10-8 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	muscularis mucosae	5.58×10-6 (p-value)	GTEx Portal V6	3
NCKIPSD	?	ENSG00000213672	muscularis mucosae	5.12×10-6 (p-value)	GTEx Portal V6	2
QRICH1	?	ENSG00000198218	lung	2.11×10-6 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	lung	3.98×10-7 (p-value)	GTEx Portal V6	3
SCAP	?	ENSG00000114650	atrial appendage	4.01×10-6 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	atrial appendage	1.18×10-5 (p-value)	GTEx Portal V6	2
NME6	?	ENSG00000172113	transformed fibroblasts	1.48×10-7 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	transformed fibroblasts	4.53×10-10 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	transformed fibroblasts	3.74×10-7 (p-value)	GTEx Portal V6	2
NCKIPSD	?	ENSG00000213672	transformed fibroblasts	4.27×10-7 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	tibial artery	4.55×10-9 (p-value)	GTEx Portal V6	3

ZNF589	?	ENSG00000164048	tibial artery	4.23×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	tibial artery	1.31×10-5 (p-value)	GTEx Portal V6	1
CDC25A	?	ENSG00000164045	tibial artery	1.43×10-5 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	blood	2.78×10-8 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	blood	2.49×10-6 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	thyroid	3.43×10-8 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	thyroid	1.66×10-5 (p-value)	GTEx Portal V6	2
NCKIPSD	?	ENSG00000213672	skeletal muscle	1.88×10-6 (p-value)	GTEx Portal V6	3
NME6	?	ENSG00000172113	skeletal muscle	2.34×10-6 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	skeletal muscle	5.12×10-10 (p-value)	GTEx Portal V6	3
TMA7	?	ENSG00000232112	skeletal muscle	8.64×10-7 (p-value)	GTEx Portal V6	1
MRPS18AP1	?	ENSG00000229759	prostate	7.45×10-6 (p-value)	GTEx Portal V6	1
MRPS18AP1	?	ENSG00000229759	transverse colon	3.40×10-6 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	transverse colon	2.11×10-6 (p-value)	GTEx Portal V6	1
ZNF589	?	ENSG00000164048	transverse colon	2.46×10-5 (p-value)	GTEx Portal V6	1
MRPS18AP1	?	ENSG00000229759	sun exposed skin	8.13×10-7 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	sun exposed skin	7.28×10-7 (p-value)	GTEx Portal V6	3
NCKIPSD	?	ENSG00000213672	sun exposed skin	1.16×10-5 (p-value)	GTEx Portal V6	1
QRICH1	?	ENSG00000198218	sun exposed skin	2.12×10-5 (p-value)	GTEx Portal V6	1
FCF1P2	?	ENSG00000228638	aorta	1.09×10-6 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	aorta	7.30×10-6 (p-value)	GTEx Portal V6	1
NCKIPSD	?	ENSG00000213672	subcutaneous adipocytes	4.90×10-10 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	subcutaneous adipocytes	5.86×10-9 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	subcutaneous adipocytes	8.77×10-6 (p-value)	GTEx Portal V6	2
QRICH1	?	ENSG00000198218	subcutaneous adipocytes	2.87×10-7 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	subcutaneous adipocytes	6.18×10-8 (p-value)	GTEx Portal V6	3
NME6	?	ENSG00000172113	subcutaneous adipocytes	6.66×10-6 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	visceral adipocytes	1.14×10-6 (p-value)	GTEx Portal V6	3
NME6	?	ENSG00000172113	visceral adipocytes	8.33×10-7 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	spleen	1.08×10-5 (p-value)	GTEx Portal V6	1
NCKIPSD	?	ENSG00000213672	tibial nerve	8.80×10-8 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	tibial nerve	1.33×10-9 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	tibial nerve	1.67×10-10 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	tibial nerve	4.95×10-7 (p-value)	GTEx Portal V6	2
ZNF589	?	ENSG00000164048	esophagus mucosa	1.33×10-6 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	esophagus mucosa	5.92×10-6 (p-value)	GTEx Portal V6	1

QRICH1	?	ENSG00000198218	esophagus mucosa	1.60×10-5 (p-value)	GTEx Portal V6	1
ZNF589	?	ENSG00000164048	adrenal gland	4.93×10-7 (p-value)	GTEx Portal V6	3
QRICH1	?	ENSG00000198218	testis	5.34×10-6 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	testis	6.49×10-8 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	testis	7.45×10-7 (p-value)	GTEx Portal V6	3
ZNF589	ENST00000354698	ILMN_1654612	skin	7.37×10-8 (p-value)	MuTHER consortium	2
ZNF589	ENST00000457782					
ZNF589	ENST00000448461					
WDR6	ENST00000615452	ILMN_1669484	adipocyte	4.99×10-9 (p-value)	MuTHER consortium	2
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
ZNF589	ENST00000354698	ILMN_1654819	blood	2.46×10-6 (p-value)	MuTHER consortium	2
ZNF589	ENST00000457782					
ZNF589	ENST00000448461					
SCAP	ENST00000428413	ILMN_1677534	blood	3.37×10-5 (p-value)	MuTHER consortium	1
SCAP	ENST00000545718					
SCAP	ENST00000320017					
SCAP	ENST00000441517					
SCAP	ENST00000265565					
KLHDC8B	ENST00000332780	ILMN_1695246	blood	2.13×10-9 (p-value)	MuTHER consortium	2
KLHDC8B	ENST00000462582					
AMT	ENST00000476226	ILMN_1665331	b-cell	5.98×10-5 (p-value)	Fairfax et al.	1
AMT	ENST00000495436					
AMT	ENST00000473163					
AMT	ENST00000427987					
AMT	ENST00000399379					
AMT	ENST00000395338					
AMT	ENST00000476127					
AMT	ENST00000273588					
AMT	ENST00000465925					
AMT	ENST00000458307					
AMT	ENST00000538581					
WDR6	ENST00000615452	ILMN_1669484	monocyte	6.65×10-9 (p-value)	Fairfax et al.	1
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
ARIH2OS	ENST00000408959	ILMN_3241670	monocyte	4.42×10-4 (p-value)	Fairfax et al.	1
QRICH1	?	ENSG00000198218	unexposed skin	1.51×10-5 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	left ventricle	5.05×10-6 (p-value)	GTEx Portal V6	1
QRICH1	?	ENSG00000198218	left ventricle	2.08×10-5 (p-value)	GTEx Portal V6	1

NME6	ENST00000415053	ILMN_2229032	blood	1.95×10-5 (p-value)	Westra et al.	2
NME6	ENST00000418431					
NME6	ENST00000426689					
NME6	ENST00000442597					
NME6	ENST00000415644					
NME6	ENST00000447314					
NME6	ENST00000456495					
NME6	ENST00000421967					
NME6	ENST00000452211					
NME6	ENST00000444069					
NME6	ENST00000494209					
NME6	ENST00000425930					
WDR6	ENST00000615452	ILMN_1669484	monocyte	2.13×10-25 (p-value)	Zeller et al.	1
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
NCKIPSD	ENST00000294129	ILMN_1736623	monocyte	1.15×10-12 (p-value)	Zeller et al.	1
NCKIPSD	ENST00000416649					
NCKIPSD	ENST00000470006					
MRPS18AP1	?	ENSG00000229759	coronary artery	1.70×10-5 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
NICN1	ENST00000273598	ILMN_1807211	chr3	monocyte	1.15×10-20 (p-value)	Zeller et al.	1
NICN1	ENST00000615713						
AMT	ENST00000273588						
NICN1	ENST00000423832						

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000338474	1	ENCP00000036343	MAP4
		ENCP00000036362	FBXW12
		ENCP00000036405	UQCRC1
		ENCP00000036386	SHISA5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363389 (TF binding site)	1	embryonic stem cell (H1ESC)	PolII, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CAMP	downstream gene variant	1528	ENST00000296435	NM_004345.4	ENSP00000296435	1
CDC25A	upstream gene variant	4866	ENST00000459900	?	?	1
CDC25A	downstream gene variant	3310	ENST00000437972	?	ENSP00000404285	1

NDUFB1P1	upstream gene variant	1928	ENST00000435191	?	?	1
RNU7-128P	upstream gene variant	709	ENST00000517247	?	?	1
ZNF589	downstream gene variant	3557	ENST00000354698	NM_016089.2	ENSP00000346729	1
ZNF589	downstream gene variant	3554	ENST00000457782	?	ENSP00000401106	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CDC25A	ENST00000443342	?	ENSP00000416483	1
CDC25A	ENST00000351231	NM_201567.1	ENSP00000343166	1
CDC25A	ENST00000302506	NM_001789.2	ENSP00000303706	1
ZNF589	ENST00000427617	?	ENSP00000392719	1
ZNF589	ENST00000412564	?	ENSP00000404398	1
ZNF589	ENST00000440261	?	ENSP00000408719	1
ZNF589	ENST00000448461	?	ENSP00000404592	1

Locus 314 (rs7713972)

SNiPAcard

Block annotations

Block info

genomic range	chr5:40,496,930-40,570,075
block size	73,146 bp
variant count	29 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.853 [-4.636 – 1.79]	**gene(s) hit or close-by**	–
phastCons	μ = 0.047 [0 – 0.998]	**eQTL gene(s)**	PTGER4
GERP++	μ = 0.045 [-4.53 – 4.26]	**potentially regulated gene(s)**	–
CADD score	μ = 4.166 [0.028 – 19.99]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PTGER4	?	ENSG00000171522	transformed fibroblasts	1.57×10^{-6} (p-value)	GTEx Portal V6	23

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001408868 (enhancer)	1	muscle (HSMM)	DNase1
ENSR00001278871 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K4me2, H3K27ac
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K4me2, H3K27ac
		muscle (HSMM)	H3K27ac, H3K4me2, DNase1
		cervix (HeLa-S3)	Cjun, Jund, Max, Cfos, DNase1
		endothelium (HUVEC)	DNase1, Cjun, H3K4me2, H3K4me1, H3K27ac
		lung (IMR90)	DNase1, H4K8ac, H3K27ac, H3K4ac, H2BK120ac, H2BK20ac
		A549	H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3, H3K4me3
		nervous (NH-A)	H3K9ac, H3K27ac, DNase1
		skin (NHEK)	DNase1, H3K4me1, H3K27ac
ENSR00001278876 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		skin (NHDF-AD)	H3K4me1, DNase1
		muscle (HSMM)	H3K4me1, DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Cjun, DNase1, H3K4me1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		blood (GM12878)	BATF, Pax5

Locus 315 (rs9295123)

SNiPAcard

Block annotations

Block info

genomic range	chr6:160,570,013-160,578,271
block size	8,259 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.253 [-1.372 – 1.03]	**gene(s) hit or close-by**	SLC22A1
phastCons	μ = 0.007 [0 – 0.023]	**eQTL gene(s)**	MAS1, SLC22A1
GERP++	μ = -0.266 [-7.06 – 1.96]	**potentially regulated gene(s)**	–
CADD score	μ = 2.965 [0.586 – 6.336]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
isobutyrylcarnitine	2.40×10^{-14}	Metabolomics GWAS Server	24816252	11
Lipoproteins	7.06×10^{-5}	dbGaP	pha003079	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MAS1	?	ENSG00000130368	ovary	7.01×10^{-7} (p-value)	GTEx Portal V6	3
SLC22A1	?	ENSG00000175003	tibial artery	2.64×10^{-6} (p-value)	GTEx Portal V6	12

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001232535 (CTCF binding site)	1	blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SLC22A1	upstream gene variant	1001	ENST00000478607	?	?	7

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLC22A1	ENST00000324965	NM_153187.1	ENSP00000318103	14
SLC22A1	ENST00000460902	?	ENSP00000439274	14
SLC22A1	ENST00000366963	NM_003057.2	ENSP00000355930	14

SLC22A1	ENST00000539263	?	ENSP00000443245	14
SLC22A1	ENST00000457470	?	ENSP00000409557	14
SLC22A1	ENST00000478607	?	?	2

Locus 315 (sentinel SNP rs9295123 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 316 (rs11568819)

SNiPAcard

Block annotations

Block info

genomic range	chr11:102,397,005-102,401,633
block size	4,629 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.454 [-1.095 – 0.187]	gene(s) hit or close-by	MMP7
phastCons	μ = 0.348 [0 – 0.696]	eQTL gene(s)	MMP7
GERP++	μ = -2.955 [-6.39 – 0.48]	potentially regulated gene(s)	–
CADD score	μ = 6.064 [1.978 – 10.15]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR013972	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MMP7	?	ENSG00000137673	tibial nerve	3.83×10-7 (p-value)	GTEx Portal V6	2
MMP7	?	ENSG00000137673	thyroid	1.23×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000567974 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	H3K27me3
		A549	H3K4me2, H3K4me3
		nervous (NH-A)	DNase1
ENSR00001606494 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K4me2
		muscle (HSMM)	DNase1

cervix (HeLa-S3)	DNase1
endothelium (HUVEC)	Cjun, DNase1
liver (HepG2)	H3K27me3
A549	H3K4me2, H3K4me3
nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MMP7	downstream gene variant, upstream gene variant	155	ENST00000531200	?	?	2
MMP7	upstream gene variant	152	ENST00000533366	?	?	1
MMP7	upstream gene variant	149	ENST00000260227	NM_002423.3	ENSP00000260227	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MMP7	ENST00000533366	?	?	1
MMP7	ENST00000260227	NM_002423.3	ENSP00000260227	1

Locus 317 (rs7083)

SNiPAcard

Block annotations

Block info

genomic range	chr11:117,106,280-117,163,765
block size	57,486 bp
variant count	40 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.709 [-3.083 – 0.486]	**gene(s) hit or close-by**	BACE1, BACE1-AS, CMB9-94B1.2, PCSK7, RNF214, SCARNA11
phastCons	μ = 0.072 [0 – 0.446]	**eQTL gene(s)**	BACE1, PCSK7, RNF214, RP11-109L13.1, SIDT2, TAGLN
GERP++	μ = -0.418 [-9.68 – 1.67]	**potentially regulated gene(s)**	–
CADD score	μ = 4.405 [0.738 – 12.24]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-109L13.1	?	ENSG00000254851	sigmoid colon	1.19×10^{-6} (p-value)	GTEx Portal V6	25
RNF214	?	ENSG00000167257	subcutaneous adipocytes	4.10×10^{-6} (p-value)	GTEx Portal V6	9
RP11-109L13.1	?	ENSG00000254851	subcutaneous adipocytes	6.06×10^{-6} (p-value)	GTEx Portal V6	3
TAGLN	ENST00000525531	ILMN_1778668	blood	1.05×10^{-7} (p-value)	MuTHER consortium	10
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
TAGLN	ENST00000525531	ILMN_2400935	blood	8.90×10^{-6} (p-value)	Westra et al.	10
TAGLN	ENST00000529622					
TAGLN	ENST00000278968					
TAGLN	ENST00000530649					
TAGLN	ENST00000529792					
TAGLN	ENST00000532870					
TAGLN	ENST00000392951					
TAGLN	ENST00000533863					
PCSK7	ENST00000540028	ILMN_1699545	blood	5.70×10^{-7} (p-value)	Westra et al.	5
PCSK7	ENST00000529458					
PCSK7	ENST00000534529					
PCSK7	ENST00000320934					
PCSK7	ENST00000527037					
SIDT2	ENST00000278951	ILMN_1791912	monocyte	5.17×10^{-19} (p-value)	Zeller et al.	2
SIDT2	ENST00000532062					
SIDT2	ENST00000324225					
SIDT2	ENST00000431081					
SIDT2	ENST00000620360					
TAGLN	ENST00000525531	ILMN_1778668	monocyte	1.08×10^{-23} (p-value)	Zeller et al.	3
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					

TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
TAGLN	ENST00000525531	ILMN_2400935	monocyte	6.54×10-17 (p-value)	Zeller et al.	2
TAGLN	ENST00000529622					
TAGLN	ENST00000278968					
TAGLN	ENST00000530649					
TAGLN	ENST00000529792					
TAGLN	ENST00000532870					
TAGLN	ENST00000392951					
TAGLN	ENST00000533863					
BACE1	?	ENSG00000186318	testis	1.77×10-7 (p-value)	GTEx Portal V6	10
BACE1	ENST00000528053	ILMN_1797804	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
BACE1	ENST00000392937					
BACE1	ENST00000313005					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000321759 (open chromatin region)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (K562)	DNase1
		A549	H3K36me3
ENSR00000569588 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		lung (IMR90)	H3K36me3
ENSR00001573947 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		A549	H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	H3K36me3
ENSR00000569591 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		HSMMtube	H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, DNase1
		nervous (NH-A)	DNase1, H3K27ac
		skin (NHEK)	DNase1, H3K36me3
		NHLF	H3K36me3, DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

gene	variant type	imm(distance)	transcript	RefSeq id	protein	variant(s)
BACE1	downstream gene variant	1515	ENST00000528053	?	ENSP00000431848	1
BACE1	downstream gene variant	3651	ENST00000510630	NM_001207049.1	ENSP00000422461	1
BACE1	downstream gene variant	3699	ENST00000513780	NM_138972.3	ENSP00000424536	1
BACE1	downstream gene variant	3882	ENST00000392937	NM_001207048.1	ENSP00000475405	1
BACE1	downstream gene variant	822	ENST00000514464	?	?	1
BACE1	downstream gene variant, upstream gene variant	1052	ENST00000509916	?	?	2
BACE1	downstream gene variant	3699	ENST00000445823	NM_138971.3	ENSP00000403685	1
BACE1	downstream gene variant	22	ENST00000504995	?	ENSP00000434486	1
BACE1	downstream gene variant	3699	ENST00000428381	NM_138973.3	ENSP00000402228	1
BACE1	downstream gene variant	4798	ENST00000530824	?	?	1
BACE1	downstream gene variant	3383	ENST00000313005	NM_012104.4	ENSP00000318585	1
CMB9-94B1.2	downstream gene variant	3956	ENST00000613535	?	?	1
PCSK7	upstream gene variant	3039	ENST00000320934	NM_004716.2	ENSP00000325917	3
PCSK7	upstream gene variant	3469	ENST00000540028	?	ENSP00000441944	3
PCSK7	upstream gene variant	3039	ENST00000530269	?	ENSP00000433252	3
PCSK7	upstream gene variant	3696	ENST00000524507	?	ENSP00000433841	2
PCSK7	upstream gene variant	3490	ENST00000532301	?	ENSP00000436459	3
RNF214	downstream gene variant	148	ENST00000531452	NM_001077239.1	ENSP00000431643	1
RNF214	upstream gene variant, downstream gene variant	590	ENST00000534709	?	?	2
RNF214	downstream gene variant	150	ENST00000300650	NM_207343.3	ENSP00000300650	1
RNF214	downstream gene variant	672	ENST00000531287	NM_001278249.1	ENSP00000435361	1
RNF214	upstream gene variant, downstream gene variant	2632	ENST00000524917	?	?	2
RNF214	downstream gene variant	1996	ENST00000529869	?	?	2
SCARNA11	upstream gene variant, downstream gene variant	93	ENST00000517183	?	?	18

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
BACE1	ENST00000313005	NM_012104.4	ENSP00000318585	1
BACE1	ENST00000392937	NM_001207048.1	ENSP00000475405	1
BACE1	ENST00000513780	NM_138972.3	ENSP00000424536	1
BACE1	ENST00000428381	NM_138973.3	ENSP00000402228	1
BACE1	ENST00000510915	?	ENSP00000475171	1
BACE1	ENST00000445823	NM_138971.3	ENSP00000403685	1
BACE1	ENST00000530824	?	?	1
BACE1	ENST00000510630	NM_001207049.1	ENSP00000422461	1
BACE1	ENST00000528053	?	ENSP00000431848	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BACE1-AS	ENST00000614401	?	?	1

RNF214	ENST00000524917	?	?	1
RNF214	ENST00000529869	?	?	6
RNF214	ENST00000530849	?	ENSP00000432903	38
RNF214	ENST00000531287	NM_001278249.1	ENSP00000435361	38
RNF214	ENST00000300650	NM_207343.3	ENSP00000300650	38
RNF214	ENST00000534428	?	ENSP00000434186	5
RNF214	ENST00000531452	NM_001077239.1	ENSP00000431643	38

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
BACE1	ENST00000313005	NM_012104.4	ENSP00000318585	1
BACE1	ENST00000392937	NM_001207048.1	ENSP00000475405	1
RNF214	ENST00000530849	?	ENSP00000432903	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
BACE1-AS	ENST00000618857	?	1
RNF214	ENST00000534709	?	1

Locus 318 (rs6865582)

SNiPAcard

Block annotations

Block info

genomic range	chr5:78,176,721-78,201,833
block size	25,113 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.281 [-0.077 – 0.791]	gene(s) hit or close-by	ARSB
phastCons	μ = 0.072 [0 – 0.418]	eQTL gene(s)	ARSB
GERP++	μ = 0.035 [-3.85 – 1.96]	potentially regulated gene(s)	–
CADD score	μ = 4.115 [0.341 – 6.101]	disease gene(s)	ARSB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ARSB	MULTIPLE SULFATASE DEFICIENCY	OMIM	MIM:272200
ARSB	MUCOPOLYSACCHARIDOSIS TYPE VI	OMIM	MIM:253200
ARSB	mucopolysaccharidosis type 6 (MPS6)	DECIPHER	MIM:253200
ARSB	Mucopolysaccharidosis type 6, rapidly progressing	OrphaNet	OrphaNet:276212
ARSB	Mucopolysaccharidosis type 6, slowly progressing	OrphaNet	OrphaNet:276223

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ARSB	?	ENSG00000113273	sun exposed skin	7.74×10-7 (p-value)	GTEx Portal V6	5

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001283744 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, DNase1
		Osteobl	H3K4me2, H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1, H3K4me2, H3K4me3
		muscle (HSMM)	H3K9ac, DNase1, H3K4me2, H3K27ac
		endothelium (HUVEC)	Max, DNase1
		liver (HepG2)	H3K4me1, H3K36me3
		blood (GM12878)	PU1, H3K4me1, H3K27ac, H3K4me2
		lung (IMR90)	DNase1, H3K27ac, H3K4me2
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

ARSB	downstream gene variant, upstream gene variant	375	ENST00000521800	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ARSB	ENST00000264914	NM_000046.3	ENSP00000264914	7
ARSB	ENST00000396151	NM_198709.2	ENSP00000379455	7
ARSB	ENST00000565165	?	ENSP00000456339	7

Locus 319 (rs8044476)

SNiPAcard

Block annotations

Block info

genomic range	chr16:72,747,685-72,800,567
block size	52,883 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.129 [-2.24 – 1.711]	**gene(s) hit or close-by**	RP5-991G20.1
phastCons	μ = 0.206 [0.001 – 1]	**eQTL gene(s)**	–
GERP++	μ = 0.692 [-0.565 – 3.93]	**potentially regulated gene(s)**	C16orf47
CADD score	μ = 7.917 [0.325 – 17.48]	**disease gene(s)**	–

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000193764	1	ENCP00000021721	C16orf47

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000509797 (TF binding site)	1	embryonic stem cell (H1ESC)	POU5F1, Nanog, BCL11A, H3K4me2, USF1, DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP5-991G20.1	ENST00000563328	?	?	5

Locus 320 (rs4905179)

SNiPAcard

Block annotations

Block info

genomic range	chr14:94,784,618-94,818,078
block size	33,461 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.179 [-0.31 – 1.321]	gene(s) hit or close-by	SERPINA6
phastCons	μ = 0.005 [0 – 0.021]	eQTL gene(s)	SERPINA1
GERP++	μ = -0.556 [-4.11 – 1.31]	potentially regulated gene(s)	–
CADD score	μ = 2.992 [0.741 – 8.487]	disease gene(s)	SERPINA6, SERPINA1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Breast size	<8.00×10-7	GWAS Catalog	22747683	1

Disease gene annotation

gene	trait	source DB	source entry/link
SERPINA6	CORTICOSTEROID-BINDING GLOBULIN DEFICIENCY	OMIM	MIM:611489
SERPINA1	ALPHA-1-ANTITRYPSIN DEFICIENCY	OMIM	MIM:613490
SERPINA6	CORTICOSTEROID-BINDING GLOBULIN DEFICIENCY	OrphaNet	OrphaNet:199247
SERPINA1	ALPHA-1-ANTITRYPSIN DEFICIENCY	OrphaNet	OrphaNet:60
SERPINA1	Hemorrhagic disease due to alpha-1 antitrypsin Pittsburgh mutation	OrphaNet	OrphaNet:178396

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SERPINA1	ENST00000393087	ILMN_1764980	blood	2.42×10-5 (p-value)	Westra et al.	2
SERPINA1	ENST00000557361					
SERPINA1	ENST00000554720					
SERPINA1	ENST00000557093					
SERPINA1	ENST00000556091					
SERPINA1	ENST00000393087	ILMN_2256050	monocyte	5.08×10-15 (p-value)	Zeller et al.	1
SERPINA1	ENST00000557361					
SERPINA1	ENST00000555289					
SERPINA1	ENST00000557093					
SERPINA1	ENST00000393088					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000652586	1	liver (HepG2)	H3K4me3, H3K27ac, H3K9ac, H3K4me2, H3K4me1, H3K79me2, PolII

(promoter flanking region)		HSMMtube	H3K27me3
		A549	DNase1
		blood (DND-41)	H3K27me3
		skin (NHEK)	DNase1
ENSR00000422985 (CTCF binding site)	1	liver (HepG2)	CTCF
		HSMMtube	H3K27me3
ENSR00000422992 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K4me1, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K27me3
		liver (HepG2)	H3K4me2, H3K4me1, Jund, FOSL2, DNase1
		blood (GM12878)	H3K27ac, NFKB, DNase1, H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K27me3
		skin (NHEK)	H3K27me3, H3K4me1, DNase1
ENSR00001624850 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K4me1, H3K27ac
		cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	H3K4me2, H3K4me1
		blood (GM12878)	H3K4me1, DNase1, NFKB, H3K27ac
		A549	H3K27me3
		skin (NHEK)	H3K27me3, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SERPINA6	upstream gene variant	2714	ENST00000341584	NM_001756.3	ENSP00000342850	1
SERPINA6	upstream gene variant	2789	ENST00000555056	?	ENSP00000451045	1
SERPINA6	upstream gene variant	2719	ENST00000557225	?	ENSP00000452018	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPINA6	ENST00000341584	NM_001756.3	ENSP00000342850	2
SERPINA6	ENST00000555056	?	ENSP00000451045	2
SERPINA6	ENST00000557225	?	ENSP00000452018	2

Locus 321 (rs338419)

SNiPAcard

Block annotations

Block info

genomic range	chr15:60,476,248-60,519,685
block size	43,438 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.168 [-1.523 – 1.224]	**gene(s) hit or close-by**	–
phastCons	μ = 0.005 [0 – 0.032]	**eQTL gene(s)**	–
GERP++	μ = -0.311 [-5.42 – 3.25]	**potentially regulated gene(s)**	–
CADD score	μ = 2.754 [0.377 – 7.678]	**disease gene(s)**	–

Locus 322 (rs2729354)

SNiPAcard

Block annotations

Block info

genomic range	chr11:57,283,988-57,358,343
block size	74,356 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.373 [-5.515 – 0.209]	**gene(s) hit or close-by**	RPS4XP13, SLC43A1, SMTNL1, UBE2L6
phastCons	μ = 0.025 [0 – 0.132]	**eQTL gene(s)**	BTBD18, RN7SL605P, RP11-624G17.3, SERPING1, SLC43A1, SMTNL1, TIMM10, YPEL4, ZDHHC5
GERP++	μ = -0.744 [-6.2 – 1.34]	**potentially regulated gene(s)**	CLP1, PRG2, RP11-872D17.8, RTN4RL2, SLC43A1, SLC43A3, YPEL4, ZDHHC5
CADD score	μ = 2.266 [0.264 – 4.946]	**disease gene(s)**	SERPING1, CLP1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SERPING1	ANGIOEDEMA, HEREDITARY, TYPE I	OMIM	MIM:106100
CLP1	PONTOCEREBELLAR HYPOPLASIA, TYPE 10	OMIM	MIM:615803
CLP1	Pontocerebellar hypoplasia, type 10	DECIPHER	MIM:615803
SERPING1	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147
SERPING1	Hereditary angioedema type 1	OrphaNet	OrphaNet:100050
SERPING1	Hereditary angioedema type 2	OrphaNet	OrphaNet:100051

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TIMM10	?	ENSG00000134809	atrial appendage	1.07×10^{-24} (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	atrial appendage	1.54×10^{-6} (p-value)	GTEx Portal V6	9
TIMM10	?	ENSG00000134809	transformed fibroblasts	3.08×10^{-47} (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	transformed fibroblasts	3.76×10^{-11} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	blood	1.60×10^{-8} (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	breast	1.91×10^{-19} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	skeletal muscle	2.01×10^{-29} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	putamen	9.09×10^{-13} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	EBV lymphocytes	4.67×10^{-27} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	ovary	3.38×10^{-12} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	sun exposed skin	3.82×10^{-24} (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	sun exposed skin	2.11×10^{-7} (p-value)	GTEx Portal V6	16

BTBD18	?	ENSG00000233436	unexposed skin	1.93×10-5 (p-value)	GTEx Portal V6	1
TIMM10	?	ENSG00000134809	unexposed skin	3.89×10-25 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	aorta	5.62×10-36 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	aorta	7.26×10-6 (p-value)	GTEx Portal V6	2
TIMM10	?	ENSG00000134809	left ventricle	5.09×10-26 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	left ventricle	1.42×10-5 (p-value)	GTEx Portal V6	3
TIMM10	?	ENSG00000134809	subcutaneous adipocytes	1.03×10-29 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	subcutaneous adipocytes	1.90×10-8 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	caudate basal ganglia	6.91×10-9 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	visceral adipocytes	1.54×10-23 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	stomach	5.18×10-23 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	cortex	3.61×10-9 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	tibial nerve	4.16×10-25 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	tibial nerve	1.71×10-10 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	esophagus mucosa	8.37×10-55 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	anterior cingulate cortex	1.85×10-10 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	pituitary	3.78×10-10 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	testis	1.16×10-30 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	testis	4.08×10-6 (p-value)	GTEx Portal V6	4
TIMM10	?	ENSG00000134809	hypothalamus	5.83×10-8 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	uterus	3.71×10-9 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	pancreas	5.35×10-20 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	muscularis mucosae	1.59×10-43 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	muscularis mucosae	7.49×10-6 (p-value)	GTEx Portal V6	3
TIMM10	?	ENSG00000134809	lung	6.47×10-19 (p-value)	GTEx Portal V6	16
YPEL4	?	ENSG00000166793	lung	3.64×10-7 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	lung	2.00×10-10 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	tibial artery	1.08×10-40 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	tibial artery	1.51×10-8 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	thyroid	1.02×10-31 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	thyroid	8.44×10-7 (p-value)	GTEx Portal V6	13
TIMM10	?	ENSG00000134809	prostate	2.26×10-11 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	transverse colon	3.33×10-30 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	liver	4.85×10-11 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	terminal ileum	1.23×10-8 (p-value)	GTEx Portal V6	15

TIMM10	?	ENSG00000134809	spleen	1.69×10-8 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	nucleus accumbens	2.59×10-9 (p-value)	GTEx Portal V6	15
SLC43A1	?	ENSG00000149150	nucleus accumbens	1.18×10-7 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	sigmoid colon	4.44×10-20 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	coronary artery	3.72×10-15 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	vagina	7.78×10-7 (p-value)	GTEx Portal V6	3
TIMM10	?	ENSG00000134809	gastroesophageal junction	3.99×10-26 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	adrenal gland	8.70×10-17 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	adrenal gland	3.43×10-9 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	hippocampus	2.94×10-10 (p-value)	GTEx Portal V6	16
TIMM10	ENST00000525587	ILMN_1765332	skin	8.47×10-79 (p-value)	MuTHER consortium	6
RN7SL605P	ENST00000577229		blood	7.64×10-69 (p-value)	MuTHER consortium	6
TIMM10	ENST00000257245		adipocyte	2.20×10-97 (p-value)	MuTHER consortium	6
TIMM10	?	ENSG00000134809	brain	5.71×10-8 (q-value)	SeeQTL DB (Myers et al.)	3
TIMM10	?	ENSG00000134809	blood	3.36×10-6 (q-value)	SeeQTL DB (HapMap)	4
TIMM10	ENST00000525587	ILMN_1765332	monocyte	<1.0×10-298 (p-value)	Zeller et al.	4
RN7SL605P	ENST00000577229					
TIMM10	ENST00000257245					
YPEL4	ENST00000532314	ILMN_1726624	monocyte	3.05×10-16 (p-value)	Zeller et al.	4
YPEL4	ENST00000528797					
YPEL4	ENST00000300022					
YPEL4	ENST00000524669					
YPEL4	ENST00000524592					
SERPING1	ENST00000278407	ILMN_1670305	blood	1.14×10-6 (p-value)	Westra et al.	5
SERPING1	ENST00000528996					
SERPING1	ENST00000531797					
SERPING1	ENST00000403558					
SERPING1	ENST00000530113					
SERPING1	ENST00000619430					
SERPING1	ENST00000378324					
SERPING1	ENST00000340687					
SERPING1	ENST00000531133					
SERPING1	ENST00000378323					
ZDHHC5	ENST00000529480	ILMN_1679358	blood	3.36×10-6 (p-value)	Westra et al.	6
ZDHHC5	ENST00000287169					
SMTNL1	?	ENSG00000214872	frontal cortex	3.97×10-6 (p-value)	GTEx Portal V6	9
SMTNL1	?	ENSG00000214872	cerebellar hemisphere	3.21×10-6 (p-value)	GTEx Portal V6	12
RP11-624G17.3	?	ENSG00000255301	cerebellar hemisphere	6.56×10-6 (p-value)	GTEx Portal V6	1
SLC43A1	?	ENSG00000149150	cerebellum	9.33×10-6 (p-value)	GTEx Portal V6	8
ZDHHC5	ENST00000529480	ILMN_1679358	b-cell	2.23×10-5 (p-value)	Fairfax et al.	2
ZDHHC5	ENST00000287169					
YPEL4	ENST00000528797	ILMN_1726624	monocyte	2.62×10-4 (p-value)	Fairfax et al.	2

YPEL4	ENST00000532314					
YPEL4	ENST00000524669					
YPEL4	ENST00000300022					
YPEL4	ENST00000524592					
RN7SL605P	ENST00000577229	ILMN_1765332	monocyte	1.02×10-63 (p-value)	Fairfax et al.	2
TIMM10	ENST00000525587		b-cell	5.82×10-46 (p-value)	Fairfax et al.	2
TIMM10	ENST00000257245					
RN7SL605P	ENST00000577229	ILMN_1765332	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
TIMM10	ENST00000525587					
TIMM10	ENST00000257245					
TIMM10	ENST00000525158	1555764_s_at	blood	1.50×10-21 (p-value)	Dixon et al.	1
TIMM10	ENST00000525587					
TIMM10	ENST00000257245					
TIMM10	ENST00000525158	218408_at	blood	2.30×10-17 (p-value)	Dixon et al.	1
TIMM10	ENST00000525587					
TIMM10	ENST00000257245					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
TIMM10	?	ENSG00000134809	chr11	brain	1.29×10-5 (q-value)	SeeQTL DB (Myers et al.)	3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000106365	1	ENCP00000012246	SLC43A3
		ENCP00000012254	SLC43A1
		ENCP00000012242	SLC43A3
		ENCP00000012255	SLC43A1
		ENCP00000012278	ZDHHC5
		ENCP00000012240	PRG2
		ENCP00000012241	RP11-872D17.8
		ENCP00000012247	SLC43A3
		ENCP00000012244	RP11-872D17.8
		ENCP00000012256	SLC43A1
		ENCP00000012276	CLP1
		ENCP00000012275	YPEL4
		ENCP00000012249	RTN4RL2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000319083 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (K562)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RPS4XP13	upstream gene variant	2321	ENST00000483219	?	?	1
SLC43A1	upstream gene variant	1682	ENST00000534105	?	ENSP00000432241	1
SLC43A1	upstream gene variant	1664	ENST00000530159	?	ENSP00000434312	1
SLC43A1	upstream gene variant	735	ENST00000524995	?	ENSP00000432361	1

SLC43A1	upstream gene variant	735	ENST00000524995	?	ENSP00000432361	1
SLC43A1	upstream gene variant	729	ENST00000278426	NM_003627.5	ENSP00000278426	1
SLC43A1	upstream gene variant	1630	ENST00000528450	NM_001198810.1	ENSP00000435673	1
SLC43A1	upstream gene variant	1900	ENST00000533066	?	ENSP00000435647	1
SLC43A1	upstream gene variant	829	ENST00000533263	?	ENSP00000435486	1
SMTNL1	downstream gene variant	3575	ENST00000527972	NM_001105565.2	ENSP00000432651	1
SMTNL1	downstream gene variant	3575	ENST00000399154	?	ENSP00000382108	1
UBE2L6	upstream gene variant	2661	ENST00000528275	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
UBE2L6	ENST00000528275	?	?	4
UBE2L6	ENST00000526659	?	ENSP00000434348	5
UBE2L6	ENST00000287156	NM_004223.4	ENSP00000287156	5
UBE2L6	ENST00000527022	?	ENSP00000435265	5
UBE2L6	ENST00000340573	NM_198183.2	ENSP00000341980	5

Locus 323 (rs5850)

SNiPAcard

Block annotations

Block info

genomic range	chr7:23,272,144-23,332,280
block size	60,137 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.333 [-2.465 – 1.258]	gene(s) hit or close-by	GPNMB
phastCons	μ = 0.058 [0 – 0.521]	eQTL gene(s)	AC005082.12, GPNMB, KLHL7, KLHL7-AS1, NUPL2
GERP++	μ = -0.903 [-9.76 – 2.64]	potentially regulated gene(s)	–
CADD score	μ = 3.494 [0.044 – 13.92]	disease gene(s)	KLHL7

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
KLHL7	RETINITIS PIGMENTOSA 42	OMIM	MIM:612943	OMIM
KLHL7	Retinitis pigmentosa	OrphaNet	OrphaNet:791	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NUPL2	?	ENSG00000136243	atrial appendage	8.06×10-19 (p-value)	GTEx Portal V6	21
KLHL7	?	ENSG00000122550	atrial appendage	1.73×10-8 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	atrial appendage	2.53×10-18 (p-value)	GTEx Portal V6	21
GPNMB	?	ENSG00000136235	atrial appendage	1.80×10-11 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	atrial appendage	2.25×10-11 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	transformed fibroblasts	6.48×10-45 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	transformed fibroblasts	6.10×10-9 (p-value)	GTEx Portal V6	7
KLHL7-AS1	?	ENSG00000230658	blood	3.10×10-15 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	blood	1.06×10-6 (p-value)	GTEx Portal V6	17
AC005082.12	?	ENSG00000226816	blood	5.54×10-10 (p-value)	GTEx Portal V6	10
NUPL2	?	ENSG00000136243	breast	1.32×10-12 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	breast	2.15×10-21 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	breast	5.17×10-11 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	skeletal muscle	2.51×10-12 (p-value)	GTEx Portal V6	21
KLHL7	?	ENSG00000122550	skeletal muscle	5.66×10-8 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	skeletal muscle	3.06×10-28 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	putamen	1.28×10-11 (p-value)	GTEx Portal V6	20

GPNMB	?	ENSG00000136235	putamen	2.39×10-11 (p-value)	GTEx Portal V6	19
KLHL7-AS1	?	ENSG00000230658	EBV lymphocytes	7.03×10-17 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	ovary	7.03×10-11 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	sun exposed skin	1.28×10-47 (p-value)	GTEx Portal V6	21
GPNMB	?	ENSG00000136235	sun exposed skin	6.40×10-9 (p-value)	GTEx Portal V6	13
AC005082.12	?	ENSG00000226816	sun exposed skin	2.13×10-9 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	sun exposed skin	5.33×10-6 (p-value)	GTEx Portal V6	6
KLHL7-AS1	?	ENSG00000230658	unexposed skin	1.12×10-24 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	unexposed skin	6.75×10-9 (p-value)	GTEx Portal V6	20
NUPL2	?	ENSG00000136243	unexposed skin	9.48×10-6 (p-value)	GTEx Portal V6	1
NUPL2	?	ENSG00000136243	aorta	4.40×10-8 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	aorta	1.67×10-29 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	aorta	4.41×10-8 (p-value)	GTEx Portal V6	18
NUPL2	?	ENSG00000136243	left ventricle	3.04×10-21 (p-value)	GTEx Portal V6	21
KLHL7	?	ENSG00000122550	left ventricle	7.65×10-10 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	left ventricle	5.19×10-19 (p-value)	GTEx Portal V6	21
GPNMB	?	ENSG00000136235	left ventricle	2.84×10-15 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	left ventricle	1.96×10-10 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	subcutaneous adipocytes	1.00×10-19 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	subcutaneous adipocytes	7.79×10-38 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	subcutaneous adipocytes	5.80×10-14 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	caudate basal ganglia	4.60×10-17 (p-value)	GTEx Portal V6	21
GPNMB	?	ENSG00000136235	caudate basal ganglia	8.89×10-12 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	caudate basal ganglia	2.97×10-6 (p-value)	GTEx Portal V6	2
AC005082.12	?	ENSG00000226816	caudate basal ganglia	3.86×10-6 (p-value)	GTEx Portal V6	1
NUPL2	?	ENSG00000136243	visceral adipocytes	7.93×10-12 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	visceral adipocytes	4.09×10-19 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	visceral adipocytes	5.40×10-13 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	stomach	2.03×10-20 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	cortex	2.91×10-11 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	cortex	1.98×10-14 (p-value)	GTEx Portal V6	21
GPNMB	?	ENSG00000136235	cortex	9.58×10-15 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	tibial nerve	5.80×10-19 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	tibial nerve	2.62×10-45 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	tibial nerve	2.35×10-6 (p-value)	GTEx Portal V6	4
KLHL7-AS1	?	ENSG00000230658	esophagus mucosa	4.36×10-26 (p-value)	GTEx Portal V6	21

AC005082.12	?	ENSG00000226816	esophagus mucosa	4.08×10-12 (p-value)	GTEx Portal V6	21
GPNMB	?	ENSG00000136235	anterior cingulate cortex	8.00×10-9 (p-value)	GTEx Portal V6	16
KLHL7-AS1	?	ENSG00000230658	anterior cingulate cortex	2.23×10-7 (p-value)	GTEx Portal V6	15
KLHL7-AS1	?	ENSG00000230658	pituitary	6.13×10-16 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	pituitary	7.95×10-8 (p-value)	GTEx Portal V6	14
KLHL7-AS1	?	ENSG00000230658	testis	1.68×10-18 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	hypothalamus	3.37×10-12 (p-value)	GTEx Portal V6	20
GPNMB	?	ENSG00000136235	hypothalamus	5.24×10-10 (p-value)	GTEx Portal V6	12
KLHL7-AS1	?	ENSG00000230658	frontal cortex	1.62×10-14 (p-value)	GTEx Portal V6	21
GPNMB	?	ENSG00000136235	frontal cortex	2.91×10-12 (p-value)	GTEx Portal V6	19
NUPL2	?	ENSG00000136243	frontal cortex	9.27×10-7 (p-value)	GTEx Portal V6	5
KLHL7-AS1	?	ENSG00000230658	uterus	4.92×10-12 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	pancreas	8.33×10-21 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	muscularis mucosae	1.24×10-7 (p-value)	GTEx Portal V6	18
KLHL7-AS1	?	ENSG00000230658	muscularis mucosae	1.90×10-32 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	muscularis mucosae	7.94×10-10 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	lung	4.43×10-13 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	lung	3.40×10-34 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	lung	1.57×10-8 (p-value)	GTEx Portal V6	18
NUPL2	?	ENSG00000136243	tibial artery	9.46×10-8 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	tibial artery	1.63×10-35 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	tibial artery	1.99×10-8 (p-value)	GTEx Portal V6	21
NUPL2	?	ENSG00000136243	thyroid	7.77×10-9 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	thyroid	1.16×10-38 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	thyroid	4.29×10-10 (p-value)	GTEx Portal V6	21
KLHL7	?	ENSG00000122550	thyroid	4.39×10-8 (p-value)	GTEx Portal V6	20
KLHL7-AS1	?	ENSG00000230658	prostate	1.25×10-11 (p-value)	GTEx Portal V6	18
GPNMB	?	ENSG00000136235	prostate	2.17×10-6 (p-value)	GTEx Portal V6	3
NUPL2	?	ENSG00000136243	transverse colon	7.39×10-11 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	transverse colon	10.00×10-20 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	liver	2.37×10-8 (p-value)	GTEx Portal V6	14
KLHL7-AS1	?	ENSG00000230658	terminal ileum	6.77×10-10 (p-value)	GTEx Portal V6	16
NUPL2	?	ENSG00000136243	spleen	5.25×10-8 (p-value)	GTEx Portal V6	15
KLHL7-AS1	?	ENSG00000230658	spleen	3.33×10-10 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	nucleus accumbens	3.92×10-13 (p-value)	GTEx Portal V6	21
GPNMB	?	ENSG00000136235	nucleus accumbens	7.93×10-9 (p-value)	GTEx Portal V6	11

KLHL7	?	ENSG00000122550	nucleus accumbens	3.93×10-7 (p-value)	GTEx Portal V6	3
NUPL2	?	ENSG00000136243	nucleus accumbens	4.49×10-6 (p-value)	GTEx Portal V6	1
KLHL7-AS1	?	ENSG00000230658	sigmoid colon	1.85×10-17 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	sigmoid colon	2.51×10-8 (p-value)	GTEx Portal V6	18
KLHL7-AS1	?	ENSG00000230658	coronary artery	2.13×10-19 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	coronary artery	6.25×10-7 (p-value)	GTEx Portal V6	19
KLHL7-AS1	?	ENSG00000230658	vagina	8.80×10-9 (p-value)	GTEx Portal V6	15
NUPL2	?	ENSG00000136243	gastroesophageal junction	1.36×10-9 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	gastroesophageal junction	3.21×10-19 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	gastroesophageal junction	5.44×10-8 (p-value)	GTEx Portal V6	11
NUPL2	?	ENSG00000136243	adrenal gland	2.41×10-9 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	adrenal gland	1.71×10-15 (p-value)	GTEx Portal V6	21
AC005082.12	?	ENSG00000226816	adrenal gland	5.11×10-8 (p-value)	GTEx Portal V6	18
NUPL2	?	ENSG00000136243	cerebellar hemisphere	6.32×10-16 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	cerebellar hemisphere	5.96×10-9 (p-value)	GTEx Portal V6	20
GPNMB	?	ENSG00000136235	cerebellar hemisphere	6.64×10-11 (p-value)	GTEx Portal V6	19
NUPL2	?	ENSG00000136243	cerebellum	2.56×10-16 (p-value)	GTEx Portal V6	21
KLHL7-AS1	?	ENSG00000230658	cerebellum	2.05×10-9 (p-value)	GTEx Portal V6	21
GPNMB	?	ENSG00000136235	cerebellum	7.27×10-8 (p-value)	GTEx Portal V6	19
KLHL7-AS1	?	ENSG00000230658	hippocampus	1.16×10-8 (p-value)	GTEx Portal V6	20
NUPL2	?	ENSG00000136243	hippocampus	7.91×10-7 (p-value)	GTEx Portal V6	3
GPNMB	?	ENSG00000136235	hippocampus	3.46×10-7 (p-value)	GTEx Portal V6	5
NUPL2	ENST00000477844	ILMN_2115154	blood	1.42×10-6 (p-value)	MuTHER consortium	11
NUPL2	ENST00000489145		adipocyte	7.35×10-8 (p-value)	MuTHER consortium	11
NUPL2	ENST00000413919					
NUPL2	ENST00000437140					
NUPL2	ENST00000485250					
NUPL2	ENST00000438012					
NUPL2	ENST00000258742					
NUPL2	ENST00000477844	ILMN_1789616	adipocyte	1.21×10-6 (p-value)	MuTHER consortium	11
NUPL2	ENST00000489145					
NUPL2	ENST00000413919					
NUPL2	ENST00000437140					
NUPL2	ENST00000485250					
NUPL2	ENST00000438012					
NUPL2	ENST00000258742					
GPNMB	ENST00000478451	ILMN_1801205	blood	3.87×10-7 (p-value)	MuTHER consortium	11
GPNMB	ENST00000381990		skin	6.43×10-8 (p-value)	MuTHER consortium	11
GPNMB	ENST00000468723					
GPNMB	ENST00000463011					
GPNMB	ENST00000258733					
GPNMB	ENST00000470994					
GPNMB	ENST00000478451	ILMN_2407389	blood	2.81×10-5 (p-value)	MuTHER consortium	11

GPNMB	ENST00000478451	ILMN_2407389	blood	2.81×10-5 (p-value)	MuTHER consortium	11
GPNMB	ENST00000381990		skin	1.34×10-8 (p-value)	MuTHER consortium	11
GPNMB	ENST00000468723					
GPNMB	ENST00000463011					
GPNMB	ENST00000258733					
GPNMB	ENST00000470994					
NUPL2	ENST00000477844	ILMN_2115154	monocyte	4.52×10-19 (p-value)	Fairfax et al.	5
NUPL2	ENST00000489145		b-cell	1.25×10-10 (p-value)	Fairfax et al.	5
NUPL2	ENST00000413919					
NUPL2	ENST00000485250					
NUPL2	ENST00000437140					
NUPL2	ENST00000438012					
NUPL2	ENST00000258742					
NUPL2	ENST00000477844	ILMN_1789616	monocyte	8.72×10-14 (p-value)	Fairfax et al.	5
NUPL2	ENST00000489145		b-cell	3.25×10-7 (p-value)	Fairfax et al.	5
NUPL2	ENST00000413919					
NUPL2	ENST00000485250					
NUPL2	ENST00000437140					
NUPL2	ENST00000438012					
NUPL2	ENST00000258742					
GPNMB	ENST00000478451	ILMN_1801205	monocyte	1.93×10-20 (p-value)	Fairfax et al.	5
GPNMB	ENST00000381990					
GPNMB	ENST00000463011					
GPNMB	ENST00000468723					
GPNMB	ENST00000258733					
GPNMB	ENST00000470994					
GPNMB	ENST00000478451	ILMN_2407389	monocyte	1.34×10-5 (p-value)	Fairfax et al.	5
GPNMB	ENST00000381990					
GPNMB	ENST00000463011					
GPNMB	ENST00000468723					
GPNMB	ENST00000258733					
GPNMB	ENST00000470994					
GPNMB	?	ENSG00000136235	blood	1.10×10-2 (q-value)	SeeQTL DB (HapMap)	4
KLHL7	ENST00000409689	ILMN_1760792	blood	2.13×10-6 (p-value)	Westra et al.	11
KLHL7	ENST00000469845					
KLHL7	ENST00000339077					
KLHL7	ENST00000521082					
KLHL7	ENST00000409689	ILMN_1674458	blood	8.65×10-5 (p-value)	Westra et al.	4
KLHL7	ENST00000339077					
KLHL7	ENST00000521082					
NUPL2	ENST00000477844	ILMN_2115154	monocyte	6.98×10-47 (p-value)	Zeller et al.	3
NUPL2	ENST00000489145					
NUPL2	ENST00000413919					
NUPL2	ENST00000437140					
NUPL2	ENST00000485250					
NUPL2	ENST00000438012					
NUPL2	ENST00000258742					
NUPL2	ENST00000477844	ILMN_1789616	monocyte	7.00×10-24 (p-value)	Zeller et al.	3
NUPL2	ENST00000489145					
NUPL2	ENST00000413919					
NUPL2	ENST00000437140					
NUPL2	ENST00000485250					

NUPL2	ENST00000438012					
NUPL2	ENST00000258742					
GPNMB	ENST00000478451	ILMN_2407389	monocyte	2.73×10-19 (p-value)	Zeller et al.	3
GPNMB	ENST00000381990					
GPNMB	ENST00000468723					
GPNMB	ENST00000463011					
GPNMB	ENST00000258733					
GPNMB	ENST00000470994					
GPNMB	ENST00000478451	ILMN_1801205	monocyte	3.67×10-44 (p-value)	Zeller et al.	3
GPNMB	ENST00000381990					
GPNMB	ENST00000468723					
GPNMB	ENST00000463011					
GPNMB	ENST00000258733					
GPNMB	ENST00000470994					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000623151 (promoter flanking region)	1	Osteobl	H3K4me2
		blood (K562)	H2AZ
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	USF1, H3K27me3
		blood (GM12878)	DNase1, H2AZ, CTCF, H3K27ac, H3K4me2
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me2, DNase1
ENSR00000065472 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, Rad21, CTCF
		HSMMtube	DNase1, H3K27me3, CTCF
		blood (K562)	PolII, Ini1, Egr1, Nfe2, Rad21, CTCF, DNase1, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, DNase1, H3K4me2, CTCF
		lung (IMR90)	CTCF, H3K27me3, DNase1
		blood (GM12878)	Rad21, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac, DNase1, H2AZ, PolII
		nervous (NH-A)	DNase1, CTCF, H3K27me3
		skin (NHEK)	CTCF
		NHLF	DNase1, CTCF
		Osteobl	CTCF, H3K36me3, H3K27me3
		blood (DND-41)	H3K4me1, CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	Jund, H3K4me2, TAF1, Max, Cmyc, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1
		A549	H3K4me3, H3K4me2, CTCF, DNase1, H3K27ac
ENSR00000065473 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	H3K36me3, CTCF
		NHLF	CTCF
		Osteobl	CTCF

blood (DND-41)	CTCF
breast (HMEC)	CTCF
cervix (HeLa-S3)	CTCF
monocytes (Monocytes-CD14+)	CTCF
endothelium (HUVEC)	H3K36me3, CTCF
A549	CTCF

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
GPNMB	1	ENST00000258733 ENST00000381990	hsa-miR-103a-3p hsa-miR-107 hsa-miR-503-5p hsa-miR-508-3p hsa-miR-876-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GPNMB	upstream gene variant, downstream gene variant	863	ENST00000492858	?	?	3
GPNMB	upstream gene variant, downstream gene variant	213	ENST00000463011	?	?	9
GPNMB	upstream gene variant, downstream gene variant	822	ENST00000465673	?	?	3
GPNMB	upstream gene variant, downstream gene variant	543	ENST00000470994	?	?	7
GPNMB	downstream gene variant	4001	ENST00000474157	?	?	1
GPNMB	downstream gene variant	913	ENST00000381990	NM_001005340.1	ENSP00000371420	4
GPNMB	downstream gene variant	1886	ENST00000459927	?	?	1
GPNMB	upstream gene variant, downstream gene variant	387	ENST00000479625	?	?	6
GPNMB	downstream gene variant	941	ENST00000492512	?	?	2
GPNMB	upstream gene variant, downstream gene variant	199	ENST00000478451	?	?	9
GPNMB	downstream gene variant	1886	ENST00000487890	?	?	1
GPNMB	upstream gene variant, downstream gene variant	378	ENST00000468723	?	?	9
GPNMB	downstream gene variant	748	ENST00000409458	?	ENSP00000386476	1
GPNMB	downstream gene variant	912	ENST00000258733	NM_002510.2	ENSP00000258733	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPNMB	ENST00000258733	NM_002510.2	ENSP00000258733	11
GPNMB	ENST00000381990	NM_001005340.1	ENSP00000371420	11
GPNMB	ENST00000468723	?	?	1
GPNMB	ENST00000409458	?	ENSP00000386476	3
GPNMB	ENST00000470994	?	?	4
GPNMB	ENST00000465673	?	?	4
GPNMB	ENST00000492858	?	?	3
GPNMB	ENST00000487890	?	?	3
GPNMB	ENST00000479625	?	?	2
GPNMB	ENST00000459927	?	?	2
GPNMB	ENST00000478451	?	?	1

GPNMB	ENST00000492512	?	?	2
GPNMB	ENST00000463011	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPNMB	ENST00000258733	NM_002510.2	ENSP00000258733	1
GPNMB	ENST00000381990	NM_001005340.1	ENSP00000371420	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
GPNMB	ENST00000470994	?	1
GPNMB	ENST00000479625	?	1

Locus 324 (rs11624221)

SNiPAcard

Block annotations

Block info

genomic range	chr14:21,128,050-21,128,050
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.024	gene(s) hit or close-by	–
phastCons	0.001	eQTL gene(s)	ANG, RNASE6, RP11-903H12.5
GERP++	-1.02	potentially regulated gene(s)	–
CADD score	3.852	disease gene(s)	ANG

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ANG	AMYOTROPHIC LATERAL SCLEROSIS 9	OMIM	MIM:611895
ANG	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RNASE6	ENST00000304677	ILMN_1780533	blood	1.61×10^{-5} (p-value)	Westra et al.	1
ANG	ENST00000397990	ILMN_2294978	blood	1.15×10^{-5} (p-value)	Westra et al.	1
RP11-903H12.5	ENST00000553909					
ANG	ENST00000336811					

Locus 325 (rs17412403)

SNiPAcard

Block annotations

Block info

genomic range	chr1:65,931,394-65,966,564
block size	35,171 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.065 [-2.435 – 2.666]	gene(s) hit or close-by	LEPR, RP4-630A11.3
phastCons	μ = 0.077 [0 – 0.995]	eQTL gene(s)	LEPR, LEPROT
GERP++	μ = 0.179 [-3.2 – 2.26]	potentially regulated gene(s)	AK4, DNAJC6, RP4-535B20.1
CADD score	μ = 4.010 [0.187 – 14.56]	disease gene(s)	LEPR, DNAJC6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
LEPR	LEPTIN RECEPTOR DEFICIENCY	OMIM	MIM:614963
DNAJC6	PARKINSON DISEASE 19, JUVENILE-ONSET	OMIM	MIM:615528
LEPR	Obesity due to leptin receptor gene deficiency	OrphaNet	OrphaNet:179494
DNAJC6	Atypical juvenile parkinsonism	OrphaNet	OrphaNet:391411

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LEPR	?	ENSG00000116678	blood	2.43×10^{-7} (p-value)	GTEx Portal V6	19
LEPR	?	ENSG00000116678	subcutaneous adipocytes	1.63×10^{-7} (p-value)	GTEx Portal V6	20
LEPROT	ENST00000488747	ILMN_1661537	skin	4.64×10^{-11} (p-value)	MuTHER consortium	11
LEPROT	ENST00000475108		blood	7.39×10^{-16} (p-value)	MuTHER consortium	11
LEPROT	ENST00000613538		adipocyte	2.44×10^{-16} (p-value)	MuTHER consortium	11
LEPROT	ENST00000371065					
LEPR	ENST00000349533	ILMN_1696391	adipocyte	4.17×10^{-6} (p-value)	MuTHER consortium	3
LEPR	ENST00000616738					
LEPR	ENST00000371060					
LEPR	ENST00000406510					
LEPR	ENST00000616738	ILMN_2234956	monocyte	3.79×10^{-20} (p-value)	Zeller et al.	6
LEPR	ENST00000371060					
LEPROT	ENST00000488747	ILMN_1661537	monocyte	3.38×10^{-15} (p-value)	Fairfax et al.	2
LEPROT	ENST00000613538		b-cell	2.00×10^{-15} (p-value)	Fairfax et al.	2
LEPROT	ENST00000475108					
LEPROT	ENST00000371065					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000023290	1	ENCP00000002874	RP4-535B20.1
ENCE00000023303	1	ENCP00000002886	DNAJC6
		ENCP00000002877	AK4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000538286 (promoter flanking region)	5	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1, H3K36me3
		HSMMtube	DNase1
		Osteobl	H3K27ac
		blood (K562)	DNase1, Max, CTCF, H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K4me1
		endothelium (HUVEC)	DNase1
		liver (HepG2)	HNF4G, DNase1, USF1, H3K4me1, p300, H3K4me2, HNF4A, FOXA1, H3K27ac, CTCF
		nervous (NH-A)	DNase1
		A549	H3K4me3
ENSR00000670217 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me1, FOXA1, H3K27ac
		blood (K562)	H3K27ac, H3K9ac, H3K79me2, PolII, H3K4me2, H3K36me3, H3K4me3
ENSR00001521210 (promoter flanking region)	4	liver (HepG2)	H3K4me1
		Osteobl	H3K4me2
		blood (K562)	H3K4me2, H3K36me3, H3K4me3, PolII, Max, DNase1, H3K27ac, H3K9ac, USF1, Cmyc, H3K79me2
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP4-630A11.3	upstream gene variant	192	ENST00000429871	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	20
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	20
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	20
LEPR	ENST00000406510	?	ENSP00000384025	20
RP4-630A11.3	ENST00000429871	?	?	3

Locus 326 (rs35908703)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,675,239-196,692,940
block size	17,702 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.300 [-2.22 – -0.379]	gene(s) hit or close-by	CFH
phastCons	μ = 0.004 [0 – 0.007]	eQTL gene(s)	–
GERP++	μ = -1.285 [-1.44 – -1.13]	potentially regulated gene(s)	–
CADD score	μ = 0.389 [0.055 – 0.723]	disease gene(s)	CFH

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571
CFH	Familial drusen	OrphaNet	OrphaNet:75376
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000550120 (CTCF binding site)	1	NHLF	CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	DNase1
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K36me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF, H3K36me3
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF
		A549	CTCF
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFH	downstream gene variant	4544	ENST00000359637	?	ENSP00000352658	1
CFH	downstream gene variant	4543	ENST00000630130	NM_001014975.2	ENSP00000487250	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFH	ENST00000466229	?	?	2
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	2

Locus 327 (rs5754266)

SNiPAcard

Block annotations

Block info

genomic range	chr22:33,172,005-33,173,569
block size	1,565 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.759 [-2.942 – 1.425]	**gene(s) hit or close-by**	SYN3
phastCons	μ = 0.003 [0 – 0.005]	**eQTL gene(s)**	–
GERP++	μ = -0.100 [-1.82 – 1.62]	**potentially regulated gene(s)**	–
CADD score	μ = 2.545 [1.118 – 3.972]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000673280 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	DNase1, CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		liver (HepG2)	Rad21, H3K4me1, CTCF
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF
		A549	CTCF
		skin (NHEK)	CTCF

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SYN3	ENST00000462268	?	?	2
SYN3	ENST00000358763	NM_003490.3, NM_001135774.1, NM_133633.2	ENSP00000351614	2

Locus 328 (rs9577481)

SNiPAcard

Block annotations

Block info

genomic range	chr13:113,860,900-113,860,900
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.45	gene(s) hit or close-by	CUL4A, PCID2
phastCons	0.005	eQTL gene(s)	PCID2
GERP++	-0.587	potentially regulated gene(s)	–
CADD score	2.865	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PCID2	ENST00000375477	ILMN_1788024	blood	3.79×10^{-6} (p-value)	Westra et al.	1
PCID2	ENST00000375459					
PCID2	ENST00000462653					
PCID2	ENST00000463102					
PCID2	ENST00000473462					
PCID2	ENST00000493650					
PCID2	ENST00000375479					
PCID2	ENST00000622406					
PCID2	ENST00000337344					
PCID2	ENST00000246505					
PCID2	ENST00000375457					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CUL4A	upstream gene variant	3014	ENST00000488558	?	ENSP00000480367	1
CUL4A	upstream gene variant	3030	ENST00000474116	?	?	1
CUL4A	upstream gene variant	3044	ENST00000494985	?	?	1
CUL4A	upstream gene variant	2108	ENST00000326335	NM_001278514.1	ENSP00000322132	1
CUL4A	upstream gene variant	2250	ENST00000617546	?	ENSP00000481782	1
CUL4A	upstream gene variant	3297	ENST00000498562	?	?	1
CUL4A	upstream gene variant	2958	ENST00000375440	NM_001008895.2	ENSP00000364589	1
CUL4A	upstream gene variant	1607	ENST00000375441	NM_001278513.1	ENSP00000364590	1
CUL4A	upstream gene variant	3247	ENST00000463426	?	?	1
CUL4A	upstream gene variant	2186	ENST00000451881	NM_003589.2	ENSP00000389118	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PCID2	ENST00000337344	NM_001127202.2	ENSP00000337405	1
PCID2	ENST00000484641	?	?	1
PCID2	ENST00000480971	?	?	1
PCID2	ENST00000375477	?	ENSP00000364626	1
PCID2	ENST00000375457	NM_001258213.1	ENSP00000364606	1
PCID2	ENST00000375459	?	ENSP00000364608	1
PCID2	ENST00000622406	NM_001258212.1	ENSP00000479494	1
PCID2	ENST00000475433	?	?	1
PCID2	ENST00000246505	?	ENSP00000246505	1
PCID2	ENST00000375479	NM_018386.3, NM_001127203.2	ENSP00000364628	1
PCID2	ENST00000491548	?	?	1

Locus 329 (rs11746309)

SNiPAcard

Block annotations

Block info

genomic range	chr5:149,472,918-149,481,535
block size	8,618 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.294 [0.136 – 0.621]	**gene(s) hit or close-by**	CSF1R, RPL7P1, Y_RNA
phastCons	μ = 0.017 [0.002 – 0.042]	**eQTL gene(s)**	CSF1R, PDGFRB
GERP++	μ = 0.287 [-0.693 – 1.97]	**potentially regulated gene(s)**	–
CADD score	μ = 2.375 [1.134 – 3.976]	**disease gene(s)**	CSF1R, PDGFRB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CSF1R	LEUKOENCEPHALOPATHY, DIFFUSE HEREDITARY, WITH SPHEROIDS	OMIM	MIM:221820
PDGFRB	JUVENILE MYELOMONOCYTIC LEUKEMIA	OMIM	MIM:607785
PDGFRB	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 4	OMIM	MIM:615007
PDGFRB	LEUKEMIA, ACUTE MYELOID	OMIM	MIM:601626
PDGFRB	MYOFIBROMATOSIS, INFANTILE, 1	OMIM	MIM:228550
PDGFRB	MYELOPROLIFERATIVE DISORDER, CHRONIC, WITH EOSINOPHILIA	OMIM	MIM:131440
CSF1R	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia	OrphaNet	OrphaNet:313808
PDGFRB	Bilateral striopallidodentate calcinosis	OrphaNet	OrphaNet:1980
PDGFRB	Unclassified chronic myeloproliferative disease	OrphaNet	OrphaNet:86830
PDGFRB	Chronic myelomonocytic leukemia	OrphaNet	OrphaNet:98823
PDGFRB	Idiopathic hypereosinophilic syndrome	OrphaNet	OrphaNet:3260
PDGFRB	Infantile myofibromatosis 1	OrphaNet	OrphaNet:2591
PDGFRB	Myeloid neoplasm associated with PDGFRB rearrangement	OrphaNet	OrphaNet:168950
PDGFRB	Familial Infantile Myofibromatosis	DECIPHER	MIM:228550

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PDGFRB	ENST00000520579	ILMN_1815057	blood	1.19×10^{-11} (p-value)	MuTHER consortium	4
PDGFRB	ENST00000261799					
CSF1R	?	ENSG00000182578	stomach	4.89×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RPL7P1	upstream gene variant, downstream gene variant	947	ENST00000468161	?	?	2
Y_RNA	downstream gene variant	589	ENST00000362539	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CSF1R	ENST00000286301	NM_005211.3, NM_001288705.1	ENSP00000286301	6
CSF1R	ENST00000511344	?	ENSP00000421174	6

Locus 330 (rs35555795)

SNiPAcard

Block annotations

Block info

genomic range	chr6:26,454,363-26,949,672
block size	495,310 bp
variant count	57 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.372 [-9.929 – 5.311]	gene(s) hit or close-by	ABT1, BTN1A1, BTN2A1, BTN3A3, CTA-14H9.5, GUSBP2, HCG11, HMGN4, LINC00240, POM121L6P, RP11-457M11.6, RP11-457M11.7, ZNF322
phastCons	μ = 0.107 [0 – 1]	eQTL gene(s)	BTN2A2, BTN3A2, BTN3A3, HIST1H2BD, HIST1H4H, HIST1H4J, PRSS16, SCGN, TRIM38, ZNF391
GERP++	μ = -0.435 [-5.9 – 3.15]	potentially regulated gene(s)	ABT1, BTN1A1, BTN3A1, BTN3A2, BTN3A3, HCG11, HIST1H2AD, HIST1H2BD, HIST1H2BF, HIST1H3F, HIST1H4C, HIST1H4D, HIST1H4E, HMGN4, VN1R13P
CADD score	μ = 5.289 [0.005 – 24.6]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
phenol sulfate	8.78×10^{-7}	Metabolomics GWAS Server	24816252		5
Lupus erythematosus, systemic	7.66×10^{-6}	dbGaP	pha002867	dbGaP	1
Lupus erythematosus, systemic	1.17×10^{-10}	dbGaP	pha002848	dbGaP	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
BTN1A1	missense variant	ENST00000244513	NM_001732.2	ENSP00000244513	S/P	Tcc/Ccc	?	?	1
BTN1A1	missense variant	ENST00000613186	?	ENSP00000484707	S/P	Tcc/Ccc	?	?	1
BTN2A1	missense variant	ENST00000429381	NM_078476.3	ENSP00000416945	3	3			3
BTN2A1	missense variant	ENST00000541522	NM_001197233.2	ENSP00000443909	5	5			5
BTN2A1	missense variant	ENST00000312541	NM_007049.4	ENSP00000312158	5	5			5
BTN2A1	missense variant	ENST00000469185	NM_001197234.2	ENSP00000419043	3	3			3

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
BTN3A2	?	ENSG00000186470	atrial appendage	1.06×10^{-18} (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	transformed fibroblasts	1.95×10^{-29} (p-value)	GTEx Portal V6	49

ZNF391	?	ENSG00000124613	transformed fibroblasts	1.19×10-5 (p-value)	GTEx Portal V6	19
BTN3A2	?	ENSG00000186470	breast	2.31×10-16 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	blood	3.20×10-54 (p-value)	GTEx Portal V6	49
TRIM38	?	ENSG00000112343	blood	5.60×10-6 (p-value)	GTEx Portal V6	2
BTN3A2	?	ENSG00000186470	skeletal muscle	3.00×10-43 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	EBV lymphocytes	1.45×10-9 (p-value)	GTEx Portal V6	28
BTN3A2	?	ENSG00000186470	unexposed skin	4.12×10-14 (p-value)	GTEx Portal V6	49
BTN2A2	?	ENSG00000124508	sun exposed skin	4.82×10-6 (p-value)	GTEx Portal V6	11
BTN3A2	?	ENSG00000186470	sun exposed skin	2.65×10-35 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	ovary	1.34×10-11 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	left ventricle	4.58×10-34 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	aorta	5.55×10-26 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	subcutaneous adipocytes	1.65×10-32 (p-value)	GTEx Portal V6	49
HIST1H4J	?	ENSG00000197238	subcutaneous adipocytes	1.69×10-6 (p-value)	GTEx Portal V6	4
BTN3A2	?	ENSG00000186470	caudate basal ganglia	5.23×10-6 (p-value)	GTEx Portal V6	28
BTN3A2	?	ENSG00000186470	visceral adipocytes	1.56×10-12 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	stomach	1.77×10-21 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	cortex	6.44×10-8 (p-value)	GTEx Portal V6	31
BTN3A2	?	ENSG00000186470	tibial nerve	4.81×10-36 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	pituitary	3.74×10-7 (p-value)	GTEx Portal V6	31
BTN3A2	?	ENSG00000186470	esophagus mucosa	1.14×10-33 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	testis	6.84×10-17 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	hypothalamus	6.80×10-8 (p-value)	GTEx Portal V6	31
BTN3A2	?	ENSG00000186470	frontal cortex	1.34×10-11 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	uterus	6.66×10-12 (p-value)	GTEx Portal V6	38
BTN3A2	?	ENSG00000186470	pancreas	5.39×10-18 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	muscularis mucosae	8.36×10-32 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	lung	1.27×10-42 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	tibial artery	2.40×10-39 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	thyroid	2.94×10-30 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	transverse colon	7.87×10-18 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	liver	5.30×10-9 (p-value)	GTEx Portal V6	47
BTN3A2	?	ENSG00000186470	nucleus accumbens	1.93×10-7 (p-value)	GTEx Portal V6	31
BTN3A2	?	ENSG00000186470	spleen	2.37×10-10 (p-value)	GTEx Portal V6	47

BTN3A2	?	ENSG00000186470	coronary artery	6.37×10-16 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	vagina	3.81×10-15 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	gastroesophageal junction	1.27×10-20 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	adrenal gland	2.01×10-11 (p-value)	GTEx Portal V6	49
BTN3A2	?	ENSG00000186470	cerebellum	1.03×10-8 (p-value)	GTEx Portal V6	47
PRSS16	?	ENSG00000112812	cerebellum	2.12×10-7 (p-value)	GTEx Portal V6	18
SCGN	ENST00000334979	ILMN_1789648	adipocyte	9.10×10-6 (p-value)	MuTHER consortium	4
SCGN	ENST00000612225					
SCGN	ENST00000377961					
BTN2A2	ENST00000471116	ILMN_2256894	blood	5.64×10-5 (p-value)	Westra et al.	3
BTN2A2	ENST00000356709					
BTN2A2	ENST00000469230					
BTN2A2	ENST00000482636					
BTN2A2	ENST00000494184					
BTN2A2	ENST00000416795					
BTN2A2	ENST00000432533					
BTN2A2	ENST00000493275					
HIST1H4H	ENST00000377727	ILMN_1751120	blood	8.40×10-6 (p-value)	Westra et al.	3
BTN3A3	?	ENSG00000111801	blood	4.34×10-4 (q-value)	SeeQTL DB (HapMap)	2
BTN3A2	?	ENSG00000186470	blood	4.56×10-12 (q-value)	SeeQTL DB (HapMap)	2
HIST1H2BD	ENST00000289316	ILMN_1651496	monocyte	2.65×10-12 (p-value)	Zeller et al.	1
BTN3A2	ENST00000396934	ILMN_1676528	monocyte	8.02×10-83 (p-value)	Zeller et al.	2
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
?	?	ILMN_1820787	monocyte	2.91×10-58 (p-value)	Zeller et al.	2
?	?	ILMN_1883997	monocyte	5.68×10-16 (p-value)	Zeller et al.	2
BTN3A2	ENST00000524459	ILMN_1700067	monocyte	5.72×10-65 (p-value)	Zeller et al.	2
BTN3A2	ENST00000528541					
BTN3A3	ENST00000467524					
BTN3A3	ENST00000482451					
BTN3A3	ENST00000494393					
BTN3A2	ENST00000527417					
BTN3A3	ENST00000244519					
BTN3A2	ENST00000528222					
BTN3A3	ENST00000474790					
BTN3A2	ENST00000377708					
BTN3A3	ENST00000496719					
BTN3A2	ENST00000396948					
BTN3A2	ENST00000527422					
BTN3A2	ENST00000356386					
BTN3A2	ENST00000524682	209846_s_at	blood	7.30×10-22 (p-value)	Dixon et al.	1
BTN3A2	ENST00000604202					
BTN3A2	ENST00000396934					
BTN3A2	ENST00000532294					
BTN3A2	ENST00000508906					
BTN3A2	ENST00000377708					

BTN3A2	ENST00000527422					
BTN3A2	ENST00000531055					
BTN3A2	ENST00000396948					
BTN3A2	ENST00000356386					
BTN3A2	ENST00000396934	212613_at	blood	1.40×10-19 (p-value)	Dixon et al.	1
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
BTN3A3	ENST00000244519	204820_s_at	blood	3.50×10-21 (p-value)	Dixon et al.	1
BTN3A2	ENST00000396934					
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
BTN3A3	ENST00000480110					
BTN2A2	ENST00000471116	ILMN_2256894	b-cell	1.99×10-5 (p-value)	Fairfax et al.	1
BTN2A2	ENST00000356709					
BTN2A2	ENST00000494184					
BTN2A2	ENST00000482636					
BTN2A2	ENST00000469230					
BTN2A2	ENST00000416795					
BTN2A2	ENST00000432533					
BTN2A2	ENST00000493275					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1671054	chr6	monocyte	9.58×10-22 (p-value)	Zeller et al.	2
?	?	ILMN_1752592	chr6	monocyte	1.68×10-13 (p-value)	Zeller et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000442938	1	ENCP00000047667	BTN3A2
		ENCP00000047679	BTN1A1
ENCE00000442976	1	ENCP00000047676	BTN3A3
ENCE00000442878	1	ENCP00000047649	HIST1H2AD HIST1H2BF
		ENCP00000047646	HIST1H4D
		ENCP00000047671	BTN3A1
		ENCP00000047680	HCG11
		ENCP00000047665	BTN3A2
		ENCP00000047640	HIST1H4C
		ENCP00000047644	HIST1H2BD
		ENCP00000047681	HMGN4
		ENCP00000047658	HIST1H3F
		ENCP00000047682	ABT1
		ENCP00000047674	BTN3A3
		ENCP00000047651	HIST1H4E
ENCE00000443031	1	ENCP00000047691	VN1R13P

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494264	2	monocytes (Monocytes-CD14+)	H3K36me3

(enhancer)		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1, DNase1
ENSR00001214106 (enhancer)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me1
		blood (K562)	PolII, Ini1, DNase1, PU1, H2AZ, H3K4me1
ENSR00001214131 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K27ac, PolII, Rad21, TAF7, Yy1, TAF1, H3K4me2, H3K4me3
		HSMMtube	H3K4me2, DNase1, H2AZ
		blood (K562)	PolII, Ini1, Brg1, DNase1, PolIII, THAP1, H3K27ac, Max, H3K9ac, Cmyc, H2AZ, Nrsf, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	DNase1, H3K4me2
		liver (HepG2)	Gabp, PolII, H2AZ, H3K4me2, H3K9ac, Cmyc, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me2, H3K4me3, DNase1
		blood (GM12878)	PolII, MEF2A, H2AZ, DNase1, Yy1, PolIII, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K4me2
		NHLF	H3K4me3, DNase1
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, Max, H3K4me3, Gabp, H3K27ac, H3K4me2, H3K9ac, Jund
		monocytes (Monocytes-CD14+)	DNase1, H3K27me3, H3K4me3
		endothelium (HUVEC)	Cjun, H3K4me2, PolII, DNase1
		A549	H3K9ac, H3K4me2, H3K4me3, DNase1
ENSR00001494272 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, H3K4me2, H3K4me3, DNase1
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2, CTCF
		blood (K562)	PolII, Ini1, DNase1, PolIII, Egr1, H3K27ac, Max, Rad21, H3K9ac, H2AZ, H3K4me2, CTCF, H3K4me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	DNase1, H3K27me3, Rad21, H2AZ, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3
		lung (IMR90)	DNase1, H3K4me3, CTCF
		blood (GM12878)	H2AZ, DNase1, Yy1, Rad21, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		NHLF	DNase1, H3K4me3, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K4me3
		breast (HMEC)	CTCF, H3K4me3, DNase1
		cervix (HeLa-S3)	PolII, H3K9ac, H3K4me2, H3K27ac, H3K4me3, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K4me2, H3K4me3, CTCF, DNase1, Cmyc
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00001214144 (CTCF binding site)	2	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	CTCF, H2AZ
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001702766 (CTCF binding site)	1	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, H3K4me3, DNase1, HDAC2, CTCF, H3K27ac, PolII, Rad21, Gabp, TAF7, H3K9ac
		HSMMtube	CTCF, H2AZ, DNase1
		blood (K562)	ELF1, Cfos, CTCFL, PolII, H3K4me2, CTCF, DNase1, H3K4me3, TAF1, H2AZ, H3K27ac, Max, Rad21, H3K9ac, Gabp, Cmyc, HEY1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3
		liver (HepG2)	DNase1, Gabp, H3K4me3, PolII, TAF1, ELF1, H3K4me2, H3K9ac, H3K27ac, CTCF
		lung (IMR90)	CTCF, DNase1, H3K4me2, H3K4me3
		blood (GM12878)	DNase1, Gabp, ELF1, Rad21, H3K4me3, H3K27ac, Cfos, CTCF, H3K9ac, PolII, H2AZ
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K9ac, H3K4me3, CTCF

		NHLF	H3K9ac, H3K27ac, CTCF, H3K4me3, DNase1
		Osteobl	CTCF, H3K4me2, H3K4me3
		blood (DND-41)	CTCF, H3K27ac, H3K9ac
		breast (HMEC)	H3K27ac, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me3, PolII, CTCF, DNase1
		A549	H3K27ac, DNase1, CTCF, H3K4me2, H3K4me3
ENSR00001214204 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K27ac, PolII, Rad21, Gabp, TAF7, H3K9ac, CTCF, HDAC2, TAF1, H3K4me2, SP1
		HSMMtube	H2AZ, CTCF, DNase1
		blood (K562)	H3K27ac, Max, Rad21, H3K9ac, Gabp, Cmyc, HEY1, H2AZ, TAF1, ELF1, Cfos, CTCFL, PolII, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, CTCF, H3K4me3
		muscle (HSMM)	H2AZ, CTCF, H3K4me3
		liver (HepG2)	DNase1, Gabp, H3K4me3, FOXA2, PolII, TAF1, ELF1, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF
		blood (GM12878)	CTCF, H3K9ac, Cfos, H3K27ac, PolII, H2AZ, DNase1, Gabp, ELF1, Rad21, H3K4me3
		lung (IMR90)	H3K4me3, CTCF, H3K4me2, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K9ac, H3K4me3, CTCF
		NHLF	DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3
		blood (DND-41)	CTCF, H3K27ac, H3K9ac
		breast (HMEC)	H3K27ac, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII
		monocytes (Monocytes-CD14+)	CTCF, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me3, PolII, CTCF, DNase1
		A549	H3K27ac, DNase1, CTCF, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABT1	upstream gene variant, downstream gene variant	2952	ENST00000274849	NM_013375.3	ENSP00000274849	3
BTN1A1	upstream gene variant, downstream gene variant	1094	ENST00000244513	NM_001732.2	ENSP00000244513	3
BTN1A1	upstream gene variant, downstream gene variant	1581	ENST00000613186	?	ENSP00000484707	3
BTN2A1	upstream gene variant	2079	ENST00000312541	NM_007049.4	ENSP00000312158	4
BTN2A1	upstream gene variant	2115	ENST00000429381	NM_078476.3	ENSP00000416945	4
BTN2A1	upstream gene variant	2058	ENST00000541522	NM_001197233.2	ENSP00000443909	4
BTN2A1	upstream gene variant	2076	ENST00000377600	?	ENSP00000366825	4
BTN2A1	upstream gene variant, downstream gene variant	1405	ENST00000480218	?	ENSP00000418936	6
BTN2A1	upstream gene variant, downstream gene variant	93	ENST00000493173	?	ENSP00000420447	8
BTN2A1	upstream gene variant, downstream gene variant	1403	ENST00000469185	NM_001197234.2	ENSP00000419043	5
BTN3A3	downstream gene variant	1320	ENST00000361232	NM_197974.2	ENSP00000355238	4
BTN3A3	downstream gene variant	720	ENST00000244519	NM_006994.4	ENSP00000244519	4
BTN3A3	downstream gene variant	721	ENST00000480110	?	?	4
BTN3A3	downstream gene variant	2218	ENST00000490254	NM_001242803.1	ENSP00000419736	4
BTN3A3	downstream gene variant	2220	ENST00000497681	?	?	4
BTN3A3	downstream gene variant	1974	ENST00000483179	?	?	4
BTN3A3	downstream gene variant	3921	ENST00000477388	?	?	2
CTA-14H9.5	downstream gene variant	2258	ENST00000567732	?	?	2
GUSBP2	upstream gene variant	24	ENST00000479900	?	?	2
GUSBP2	downstream gene variant	2834	ENST00000492832	?	?	1
GUSBP2	downstream gene variant, upstream gene	2183	ENST00000463434	?	?	2

	variant					
HCG11	downstream gene variant	3083	ENST00000411553	?	?	2
HMGN4	downstream gene variant	2730	ENST00000377575	NM_006353.2	ENSP00000366798	2
LINC00240	upstream gene variant	415	ENST00000606878	?	?	2
LINC00240	upstream gene variant, downstream gene variant	414	ENST00000607607	?	?	3
POM121L6P	downstream gene variant	4071	ENST00000441186	?	?	1
RP11-457M11.6	downstream gene variant	2142	ENST00000602994	?	?	1
RP11-457M11.7	upstream gene variant, downstream gene variant	3100	ENST00000616994	?	?	2
RP11-457M11.7	upstream gene variant, downstream gene variant	3100	ENST00000611708	?	?	2
ZNF322	downstream gene variant	910	ENST00000622479	NM_001242797.1	ENSP00000482607	2
ZNF322	downstream gene variant	900	ENST00000415922	NM_024639.4	ENSP00000418897	2
ZNF322	downstream gene variant	2808	ENST00000471278	?	ENSP00000419728	2
ZNF322	downstream gene variant	4993	ENST00000480036	?	ENSP00000420301	1
ZNF322	downstream gene variant	910	ENST00000607204	NM_001242798.1	ENSP00000483223	2
ZNF322	downstream gene variant	910	ENST00000456172	NM_001242799.1	ENSP00000478899	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BTN2A1	ENST00000377600	?	ENSP00000366825	4
BTN2A1	ENST00000312541	NM_007049.4	ENSP00000312158	2
BTN2A1	ENST00000429381	NM_078476.3	ENSP00000416945	4
BTN2A1	ENST00000469185	NM_001197234.2	ENSP00000419043	4
BTN2A1	ENST00000541522	NM_001197233.2	ENSP00000443909	2
BTN2A1	ENST00000480218	?	ENSP00000418936	2
BTN2A1	ENST00000493173	?	ENSP00000420447	2
GUSBP2	ENST00000479900	?	?	9
GUSBP2	ENST00000463434	?	?	3
GUSBP2	ENST00000492832	?	?	1
LINC00240	ENST00000607607	?	?	3
LINC00240	ENST00000606878	?	?	5
RP11-457M11.7	ENST00000611708	?	?	1
ZNF322	ENST00000415922	NM_024639.4	ENSP00000418897	1
ZNF322	ENST00000622479	NM_001242797.1	ENSP00000482607	1
ZNF322	ENST00000465674	?	?	1
ZNF322	ENST00000471278	?	ENSP00000419728	1
ZNF322	ENST00000461899	?	?	1
ZNF322	ENST00000607204	NM_001242798.1	ENSP00000483223	1
ZNF322	ENST00000480036	?	ENSP00000420301	1
ZNF322	ENST00000456172	NM_001242799.1	ENSP00000478899	1

3'-UTR variant

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABT1	ENST00000274849	NM_013375.3	ENSP00000274849	1
BTN2A1	ENST00000377600	?	ENSP00000366825	5
BTN2A1	ENST00000429381	NM_078476.3	ENSP00000416945	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
LINC00240	ENST00000607607	?	1
RP11-457M11.7	ENST00000616994	?	1

Locus 331 (rs2989736)

SNiPAcard

Block annotations

Block info

genomic range	chr9:137,797,175-137,797,175
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-3.233	**gene(s) hit or close-by**	FCN1
phastCons	0	**eQTL gene(s)**	–
GERP++	-1.67	**potentially regulated gene(s)**	–
CADD score	0.312	**disease gene(s)**	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCN1	downstream gene variant	4256	ENST00000371806	NM_002003.3	ENSP00000360871	1
FCN1	downstream gene variant	561	ENST00000616356	?	ENSP00000479379	1

Locus 332 (rs2255336)

SNiPAcard

Block annotations

Block info

genomic range	chr12:10,517,808-10,644,573
block size	126,766 bp
variant count	73 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.740 [-4.39 – 0.939]	**gene(s) hit or close-by**	KLRC1, KLRC2, KLRC3, KLRC4, KLRC4-KLRK1, KLRK1, RP11-277P12.20, RP11-277P12.6, RP11-277P12.9
phastCons	μ = 0.054 [0 – 0.865]	**eQTL gene(s)**	GABARAPL1, KLRC1, KLRC2, KLRC3, KLRC4-KLRK1, KLRD1, KLRK1, RP11-277P12.10, RP11-277P12.20, TAS2R10, YBX3
GERP++	μ = -0.537 [-6.29 – 1.55]	**potentially regulated gene(s)**	CLEC1A, KLRC2, KLRC3, KLRD1, KLRK1
CADD score	μ = 3.036 [0.042 – 10.47]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Obesity-related traits	$<6.00\times10^{-6}$	GWAS Catalog	23251661	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM099345	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
KLRC4-KLRK1	missense variant	ENST00000616233	NM_001199805.1	ENSP00000480609	T/A	Aca/Gca	tolerated	benign	1
KLRK1	missense variant	ENST00000396451	?	ENSP00000379726	T/A	Aca/Gca	tolerated	?	1
KLRK1	missense variant	ENST00000240618	NM_007360.3	ENSP00000240618	T/A	Aca/Gca	tolerated	benign	1
KLRK1	missense variant	ENST00000540818	?	ENSP00000446003	T/A	Aca/Gca	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
KLRC2	?	ENSG00000205809	pancreas	1.05×10^{-6} (p-value)	GTEx Portal V6	36
GABARAPL1	ENST00000539170	ILMN_2151281	blood	8.21×10^{-18} (p-value)	MuTHER consortium	18
GABARAPL1	ENST00000541960					
GABARAPL1	ENST00000540424					
GABARAPL1	ENST00000266458					

GABARAPL1	ENST00000542722					
GABARAPL1	ENST00000546017					
GABARAPL1	ENST00000535576					
KLRD1	ENST00000381908	ILMN_1799134	blood	1.75×10-5 (p-value)	MuTHER consortium	14
KLRD1	ENST00000336164					
KLRD1	ENST00000344825					
KLRD1	ENST00000539374					
KLRD1	ENST00000544747					
KLRD1	ENST00000350274					
KLRD1	ENST00000543777					
KLRD1	ENST00000540271					
KLRD1	ENST00000539792					
KLRC2	?	ENSG00000205809	muscularis mucosae	2.54×10-10 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	lung	9.82×10-20 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	transformed fibroblasts	1.20×10-11 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	blood	3.58×10-13 (p-value)	GTEx Portal V6	68
KLRC3	?	ENSG00000205810	blood	2.21×10-6 (p-value)	GTEx Portal V6	15
KLRC2	?	ENSG00000205809	breast	1.41×10-9 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	thyroid	1.32×10-11 (p-value)	GTEx Portal V6	68
RP11-277P12.20	?	ENSG00000245648	thyroid	3.93×10-6 (p-value)	GTEx Portal V6	1
KLRC2	?	ENSG00000205809	transverse colon	1.12×10-8 (p-value)	GTEx Portal V6	60
KLRK1	?	ENSG00000213809	blood	5.02×10-3 (q-value)	SeeQTL DB (HapMap)	5
RP11-277P12.10	?	ENSG00000256288	sun exposed skin	4.53×10-7 (p-value)	GTEx Portal V6	40
RP11-277P12.20	?	ENSG00000245648	sun exposed skin	3.53×10-7 (p-value)	GTEx Portal V6	44
RP11-277P12.20	?	ENSG00000245648	unexposed skin	4.51×10-10 (p-value)	GTEx Portal V6	67
KLRC2	?	ENSG00000205809	subcutaneous adipocytes	2.11×10-8 (p-value)	GTEx Portal V6	62
KLRC2	?	ENSG00000205809	terminal ileum	1.84×10-8 (p-value)	GTEx Portal V6	24
KLRC2	?	ENSG00000205809	stomach	1.10×10-15 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	spleen	7.02×10-11 (p-value)	GTEx Portal V6	64
KLRC2	?	ENSG00000205809	nucleus accumbens	7.35×10-9 (p-value)	GTEx Portal V6	55
KLRC2	?	ENSG00000205809	vagina	1.21×10-9 (p-value)	GTEx Portal V6	60
KLRC2	?	ENSG00000205809	esophagus mucosa	3.88×10-10 (p-value)	GTEx Portal V6	68
RP11-277P12.20	?	ENSG00000245648	esophagus mucosa	1.10×10-27 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	anterior cingulate cortex	2.84×10-11 (p-value)	GTEx Portal V6	68
RP11-277P12.20	?	ENSG00000245648	testis	2.48×10-11 (p-value)	GTEx Portal V6	68
KLRC2	?	ENSG00000205809	testis	1.63×10-6 (p-value)	GTEx Portal V6	19
KLRC2	?	ENSG00000205809	hypothalamus	4.78×10-9 (p-value)	GTEx Portal V6	56
KLRC2	?	ENSG00000205809	frontal cortex	1.07×10-13 (p-value)	GTEx Portal V6	68

KLRC4-KLRK1	ENST00000591546	1555691_a_at	blood	1.00×10-9 (p-value)	Dixon et al.	2
KLRC4-KLRK1	ENST00000586581					
KLRC4-KLRK1	ENST00000543572					
KLRC4-KLRK1	ENST00000590131					
KLRC4-KLRK1	ENST00000543812					
KLRC4-KLRK1	ENST00000585507					
KLRK1	ENST00000540818					
KLRC4-KLRK1	ENST00000539370					
KLRK1	ENST00000544449					
KLRK1	ENST00000240618					
KLRC4-KLRK1	ENST00000588263					
KLRC4-KLRK1	ENST00000539300					
KLRK1	ENST00000540267					
KLRC4-KLRK1	ENST00000616233					
KLRC4-KLRK1	ENST00000585711					
KLRC4-KLRK1	ENST00000591937					
KLRC4-KLRK1	ENST00000588447					
KLRK1	ENST00000396451					
KLRC4-KLRK1	ENST00000590323					
KLRC4-KLRK1	ENST00000591546	205821_at	blood	1.90×10-12 (p-value)	Dixon et al.	2
KLRC4-KLRK1	ENST00000586581					
KLRC4-KLRK1	ENST00000543572					
KLRC4-KLRK1	ENST00000585711					
KLRC4-KLRK1	ENST00000590131					
KLRC4-KLRK1	ENST00000543812					
KLRK1	ENST00000544449					
KLRK1	ENST00000240618					
KLRC4-KLRK1	ENST00000539300					
KLRK1	ENST00000540267					
KLRC4-KLRK1	ENST00000616233					
KLRC2	?	ENSG00000205809	tibial nerve	2.43×10-9 (p-value)	GTEx Portal V6	57
YBX3	ENST00000279550	ILMN_1782788	monocyte	1.40×10-6 (p-value)	Fairfax et al.	2
YBX3	ENST00000540447					
YBX3	ENST00000228251					
YBX3	ENST00000366286					
YBX3	ENST00000540975					
YBX3	ENST00000536823					
YBX3	ENST00000546164					
KLRC2	?	ENSG00000205809	cortex	1.77×10-7 (p-value)	GTEx Portal V6	8
KLRC2	?	ENSG00000205809	cerebellar hemisphere	3.01×10-6 (p-value)	GTEx Portal V6	4
KLRC2	?	ENSG00000205809	hippocampus	7.43×10-8 (p-value)	GTEx Portal V6	14
?	?	ILMN_1707328	liver	9.23×10-13 (p-value)	Innocenti et al.	1
KLRC2	?	ENSG00000205809	cerebellum	2.71×10-7 (p-value)	GTEx Portal V6	29
KLRC2	?	ENSG00000205809	putamen	7.07×10-7 (p-value)	GTEx Portal V6	3
KLRC1	ENST00000347831	ILMN_1752647	blood	5.64×10-5 (p-value)	Westra et al.	1
KLRC1	ENST00000359151					
KLRC1	ENST00000544822					
KLRC1	ENST00000536188					
KLRC1	ENST00000408006					

KLRC2	?	ENSG00000205809	visceral adipocytes	1.04×10-6 (p-value)	GTEx Portal V6	23
TAS2R10	?	ENSG00000121318	EBV lymphocytes	1.55×10-6 (p-value)	GTEx Portal V6	1
KLRC2	?	ENSG00000205809	caudate basal ganglia	3.48×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000133909	1	ENCP00000016103	CLEC1A
ENCE00000133997	1	ENCP00000016122	KLRK1
		ENCP00000016121	KLRK1
		ENCP00000016129	KLRC2
ENCE00000133946	1	ENCP00000016116	KLRD1
		ENCP00000016127	KLRC3

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000426296 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K27ac, CTCF, Rad21, FOSL1, Yy1, TAF1, H3K4me2, SP1, H3K4me3
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	DNase1, H3K27ac, H3K9ac, H2AZ, PolII, H3K4me2, CTCF, H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	CTCF, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3
		blood (GM12878)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, IRF4, DNase1, H2AZ, BCLAF1, PolII, PU1, EBF1
		lung (IMR90)	CTCF, H3K9ac, H2BK120ac, H3K4me3, H4K5ac, H3K4me2, H3K18ac, H3K27ac, H4K8ac, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	CTCF, DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, CTCF
		blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac
		cervix (HeLa-S3)	H3K9ac, DNase1, H3K4me2, H3K27ac, H3K4me3, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	PolII, H3K4me3, H3K4me2
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF
ENSR00001609380 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H2AZ
		blood (K562)	H3K27ac, H3K9ac, H2AZ, H3K4me3, H3K4me2, CTCF, DNase1
		skin (NHDF-AD)	H3K4me2, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2
		liver (HepG2)	CTCF, Rad21, H2AZ, H3K4me2, H3K4me3
		lung (IMR90)	CTCF, H4K5ac, H3K4me2, H3K18ac, H3K27ac, H4K8ac
		blood (GM12878)	H2AZ, Rad21
		nervous (NH-A)	H3K4me2
		skin (NHEK)	H3K27ac, H3K4me2, CTCF, H3K4me1
		Osteobl	H2AZ, H3K4me2, CTCF
		blood (DND-41)	H3K4me3, H3K9ac, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, H3K4me3, CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K4me3, H3K4me2
		A549	CTCF
ENSR00001609383 (enhancer)	1	cervix (HeLa-S3)	H3K36me3
		lung (IMR90)	H3K36me3
ENSR00001461319 (promoter flanking region)	2	cervix (HeLa-S3)	DNase1, H3K4me2, H3K36me3
		NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3

		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001609384 (CTCF binding site)	1	cervix (HeLa-S3)	DNase1, H3K4me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K27me3
		NHLF	DNase1
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00000653098 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K4me2, H3K9ac, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K36me3, PolII, H3K9ac, DNase1, Nrsf, H3K4me2, H3K27ac, H3K4me1, TAF1, H3K4me3, H3K79me2, Gabp
		monocytes (Monocytes-CD14+)	H3K27me3, H3K27ac, DNase1
		liver (HepG2)	FOXA1
		blood (GM12878)	BCL11A, DNase1, PU1
		A549	H3K4me3, H3K4me2, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KLRC1	downstream gene variant, upstream gene variant	20	ENST00000544822	NM_213658.2	ENSP00000438038	10
KLRC1	downstream gene variant, upstream gene variant	10	ENST00000347831	NM_007328.3	ENSP00000256965	10
KLRC1	downstream gene variant, upstream gene variant	560	ENST00000543893	?	ENSP00000442545	10
KLRC1	upstream gene variant	1137	ENST00000536188	NM_001304448.1	ENSP00000441432	4
KLRC1	downstream gene variant, upstream gene variant	7	ENST00000359151	NM_002259.4	ENSP00000352064	10
KLRC1	downstream gene variant, upstream gene variant	323	ENST00000537699	?	?	11
KLRC1	downstream gene variant, upstream gene variant	7	ENST00000408006	NM_213657.2	ENSP00000385304	10
KLRC2	downstream gene variant	369	ENST00000535069	?	ENSP00000438983	1
KLRC2	downstream gene variant	2105	ENST00000537017	?	ENSP00000445240	4
KLRC2	downstream gene variant	1684	ENST00000381902	NM_002260.3	ENSP00000371327	4
KLRC2	downstream gene variant, upstream gene variant	2065	ENST00000536833	?	ENSP00000444754	13
KLRC2	downstream gene variant	1694	ENST00000619500	?	ENSP00000479152	4
KLRC2	downstream gene variant	1694	ENST00000381901	?	ENSP00000371326	4
KLRC3	downstream gene variant	2288	ENST00000396439	NM_002261.2	ENSP00000379716	2
KLRC4	downstream gene variant, upstream gene variant	130	ENST00000309384	NM_013431.2	ENSP00000310216	4
KLRC4-KLRK1	downstream gene variant	3563	ENST00000616233	NM_001199805.1	ENSP00000480609	5
KLRC4-KLRK1	downstream gene variant	4201	ENST00000543812	?	ENSP00000457500	4
KLRC4-KLRK1	downstream gene variant, upstream gene variant	2121	ENST00000543572	?	ENSP00000456286	7
KLRC4-KLRK1	downstream gene variant, upstream gene variant	338	ENST00000586581	?	ENSP00000467228	5
KLRC4-KLRK1	downstream gene variant	4269	ENST00000591546	?	ENSP00000468432	3
KLRC4-KLRK1	downstream gene variant, upstream gene variant	338	ENST00000539300	?	ENSP00000455951	6
KLRC4-KLRK1	downstream gene variant	188	ENST00000585507	?	ENSP00000465434	2

KLRC4-KLRK1	downstream gene variant, upstream gene variant	69	ENST00000539370	?	?	3
KLRC4-KLRK1	downstream gene variant, upstream gene variant	2128	ENST00000585711	?	ENSP00000467316	5
KLRC4-KLRK1	downstream gene variant	4292	ENST00000590131	?	ENSP00000467152	3
KLRC4-KLRK1	downstream gene variant	39	ENST00000591937	?	ENSP00000466326	3
KLRK1	downstream gene variant, upstream gene variant	1452	ENST00000396451	?	ENSP00000379726	5
KLRK1	downstream gene variant, upstream gene variant	715	ENST00000544449	?	?	7
KLRK1	downstream gene variant, upstream gene variant	170	ENST00000540818	?	ENSP00000446003	9
KLRK1	downstream gene variant, upstream gene variant	722	ENST00000240618	NM_007360.3	ENSP00000240618	8
KLRK1	downstream gene variant, upstream gene variant	97	ENST00000540267	?	?	11
RP11-277P12.20	downstream gene variant	3880	ENST00000500682	?	?	1
RP11-277P12.6	downstream gene variant	2288	ENST00000539033	?	ENSP00000437563	2
RP11-277P12.9	upstream gene variant	4164	ENST00000535911	?	?	3

Putative effect on transcript

Non-coding exon variant (splice region)

gene	affected transcript	RefSeq id	variant(s)
RP11-277P12.20	ENST00000500682	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLRC1	ENST00000544822	NM_213658.2	ENSP00000438038	6
KLRC1	ENST00000359151	NM_002259.4	ENSP00000352064	6
KLRC1	ENST00000536188	NM_001304448.1	ENSP00000441432	12
KLRC1	ENST00000543893	?	ENSP00000442545	2
KLRC1	ENST00000347831	NM_007328.3	ENSP00000256965	6
KLRC1	ENST00000408006	NM_213657.2	ENSP00000385304	6
KLRC2	ENST00000535069	?	ENSP00000438983	3
KLRC4-KLRK1	ENST00000588447	?	ENSP00000465254	25
KLRC4-KLRK1	ENST00000585507	?	ENSP00000465434	23
KLRC4-KLRK1	ENST00000543812	?	ENSP00000457500	25
KLRC4-KLRK1	ENST00000591546	?	ENSP00000468432	25
KLRC4-KLRK1	ENST00000590323	?	ENSP00000467880	25
KLRC4-KLRK1	ENST00000539300	?	ENSP00000455951	23
KLRC4-KLRK1	ENST00000543572	?	ENSP00000456286	23
KLRC4-KLRK1	ENST00000590131	?	ENSP00000467152	25
KLRC4-KLRK1	ENST00000586581	?	ENSP00000467228	23
KLRC4-KLRK1	ENST00000616233	NM_001199805.1	ENSP00000480609	25
KLRC4-KLRK1	ENST00000539370	?	?	22
KLRC4-KLRK1	ENST00000588263	?	ENSP00000468074	25
KLRC4-KLRK1	ENST00000591937	?	ENSP00000466326	23
KLRC4-KLRK1	ENST00000585711	?	ENSP00000467316	23

KLRK1	ENST00000396451	?	ENSP00000379726	19
KLRK1	ENST00000540267	?	?	9
KLRK1	ENST00000240618	NM_007360.3	ENSP00000240618	18
KLRK1	ENST00000544449	?	?	18
KLRK1	ENST00000540818	?	ENSP00000446003	16
RP11-277P12.20	ENST00000500682	?	?	32
RP11-277P12.6	ENST00000539033	?	ENSP00000437563	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLRC1	ENST00000544822	NM_213658.2	ENSP00000438038	2
KLRC1	ENST00000359151	NM_002259.4	ENSP00000352064	2
KLRC1	ENST00000347831	NM_007328.3	ENSP00000256965	2
KLRC1	ENST00000408006	NM_213657.2	ENSP00000385304	2
KLRC4-KLRK1	ENST00000588447	?	ENSP00000465254	1
KLRC4-KLRK1	ENST00000585507	?	ENSP00000465434	1
KLRC4-KLRK1	ENST00000543812	?	ENSP00000457500	1
KLRC4-KLRK1	ENST00000591546	?	ENSP00000468432	1
KLRC4-KLRK1	ENST00000590323	?	ENSP00000467880	1
KLRC4-KLRK1	ENST00000539300	?	ENSP00000455951	1
KLRC4-KLRK1	ENST00000543572	?	ENSP00000456286	1
KLRC4-KLRK1	ENST00000590131	?	ENSP00000467152	1
KLRC4-KLRK1	ENST00000586581	?	ENSP00000467228	1
KLRC4-KLRK1	ENST00000588263	?	ENSP00000468074	1
KLRC4-KLRK1	ENST00000585711	?	ENSP00000467316	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
KLRC4-KLRK1	ENST00000539370	?	2
KLRK1	ENST00000540267	?	3
KLRK1	ENST00000544449	?	3
RP11-277P12.20	ENST00000500682	?	3

Locus 333 (rs9901901)

SNiPAcard

Block annotations

Block info

genomic range	chr17:26,610,513-26,610,513
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.216	gene(s) hit or close-by	KRT18P55
phastCons	0.996	eQTL gene(s)	SARM1, TMEM199, TMEM97
GERP++	-2.77	potentially regulated gene(s)	–
CADD score	1.06	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SARM1	ENST00000585482	ILMN_1746265	adipocyte	1.70×10^{-6} (p-value)	MuTHER consortium	1
TMEM97	ENST00000226230	ILMN_1710962	blood	1.33×10^{-5} (p-value)	MuTHER consortium	1
TMEM97	ENST00000336687		adipocyte	4.53×10^{-7} (p-value)	MuTHER consortium	1
TMEM97	ENST00000582384					
TMEM199	ENST00000483505	ILMN_1748481	blood	2.43×10^{-5} (p-value)	Westra et al.	1
TMEM199	ENST00000509083					
TMEM199	ENST00000585027					
TMEM199	ENST00000292114					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001535058 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KRT18P55	ENST00000577198	?	?	1
KRT18P55	ENST00000578578	?	?	1

KRT18P55	ENST00000581956	?	?	1

Locus 335 (rs10860794)

SNiPAcard

Block annotations

Block info

genomic range	chr12:102,216,673-102,225,751
block size	9,079 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.568 [-1.416 – 0.689]	gene(s) hit or close-by	GNPTAB, RNA5SP369
phastCons	μ = 0.002 [0 – 0.014]	eQTL gene(s)	CHPT1, DRAM1
GERP++	μ = 0.015 [-1.61 – 3.03]	potentially regulated gene(s)	CCDC53
CADD score	μ = 3.994 [0.729 – 7.826]	disease gene(s)	GNPTAB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
GNPTAB	mucolipidosis type III complementation group A (MLIIIA)	DECIPHER	MIM:252600
GNPTAB	mucolipidosis type II (MLII)	DECIPHER	MIM:252500
GNPTAB	MUCOLIPIDOSIS II ALPHA/BETA	OMIM	MIM:252500
GNPTAB	MUCOLIPIDOSIS III ALPHA/BETA	OMIM	MIM:252600
GNPTAB	Mucolipidosis type 3	OrphaNet	OrphaNet:577
GNPTAB	Mucolipidosis type 2	OrphaNet	OrphaNet:576

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CHPT1	?	ENSG00000111666	muscularis mucosae	3.15×10^{-6} (p-value)	GTEx Portal V6	9
DRAM1	?	ENSG00000136048	muscularis mucosae	2.61×10^{-12} (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	tibial nerve	5.47×10^{-6} (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	transformed fibroblasts	7.69×10^{-15} (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	tibial artery	2.87×10^{-14} (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	thyroid	2.40×10^{-11} (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	skeletal muscle	1.82×10^{-7} (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	sun exposed skin	4.17×10^{-8} (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	aorta	2.91×10^{-9} (p-value)	GTEx Portal V6	9
CHPT1	?	ENSG00000111666	subcutaneous adipocytes	1.16×10^{-8} (p-value)	GTEx Portal V6	9
DRAM1	ENST00000258534	ILMN_1669376	blood	2.44×10^{-8} (p-value)	MuTHER consortium	3
CHPT1	ENST00000229266	ILMN_2202940	adipocyte	4.19×10^{-6} (p-value)	MuTHER consortium	3
CHPT1	ENST00000549872					
CHPT1	ENST00000552215					

CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
CHPT1	ENST00000549872	230364_at	blood	2.30×10-9 (p-value)	Dixon et al.	2
CHPT1	ENST00000552215					
CHPT1	ENST00000229266	221675_s_at	blood	1.70×10-9 (p-value)	Dixon et al.	2
CHPT1	ENST00000552215					
CHPT1	ENST00000549872					
CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
CHPT1	?	ENSG00000111666	blood	6.59×10-3 (q-value)	SeeQTL DB (HapMap)	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000143578	1	ENCP00000017129	CCDC53
		ENCP00000017128	CCDC53

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001614475 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K4me2, H3K27ac, DNase1
		lung (IMR90)	H3K36me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K36me3
ENSR00001614476 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, Sin3Ak20, CTCF, USF1, DNase1
		blood (DND-41)	H3K4me1, CTCF, H3K27ac
		blood (K562)	USF1, DNase1, Max, CTCF
		skin (NHDF-AD)	DNase1
		cervix (HeLa-S3)	H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	DNase1
		blood (GM12878)	CTCF
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GNPTAB	upstream gene variant	1155	ENST00000549940	?	ENSP00000449150	1
GNPTAB	upstream gene variant	1294	ENST00000549165	?	ENSP00000450413	1
GNPTAB	upstream gene variant	1130	ENST00000392919	?	ENSP00000376651	1
GNPTAB	upstream gene variant	1035	ENST00000299314	NM_024312.4	ENSP00000299314	1
RNA5SP369	upstream gene variant	4847	ENST00000411114	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GNPTAB	ENST00000549165	?	ENSP00000450413	8
GNPTAB	ENST00000549940	?	ENSP00000449150	8
GNPTAB	ENST00000392919	?	ENSP00000376651	8
GNPTAB	ENST00000299314	NM_024312.4	ENSP00000299314	8

Locus 335 (sentinel SNP rs10860794 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 336 (rs5754249)

SNiPAcard

Block annotations

Block info

genomic range	chr22:33,147,483-33,147,526
block size	44 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.167 [-0.015 – 0.349]	gene(s) hit or close-by	SYN3
phastCons	μ = 0.003 [0.001 – 0.004]	eQTL gene(s)	–
GERP++	μ = 0.480 [-0.479 – 1.44]	potentially regulated gene(s)	–
CADD score	μ = 5.849 [4.562 – 7.137]	disease gene(s)	–

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SYN3	ENST00000462268	?	?	2
SYN3	ENST00000358763	NM_003490.3, NM_001135774.1, NM_133633.2	ENSP00000351614	2

Locus 337 (rs1513890)

SNiPAcard

Block annotations

Block info

genomic range	chr4:76,830,353-76,840,698
block size	10,346 bp
variant count	61 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.564 [-3.774 – 0.968]	gene(s) hit or close-by	NAAA
phastCons	μ = 0.071 [0 – 0.993]	eQTL gene(s)	CDKL2, NAAA, PPEF2
GERP++	μ = 0.145 [-3.31 – 3.69]	potentially regulated gene(s)	ART3, CXCL9, SDAD1
CADD score	μ = 3.814 [0.197 – 13.39]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PPEF2	?	ENSG00000156194	tibial nerve	3.33×10-6 (p-value)	GTEx Portal V6	56
NAAA	?	ENSG00000138744	tibial nerve	4.32×10-6 (p-value)	GTEx Portal V6	2
CDKL2	?	ENSG00000138769	tibial nerve	6.76×10-6 (p-value)	GTEx Portal V6	1
PPEF2	?	ENSG00000156194	cerebellum	3.98×10-9 (p-value)	GTEx Portal V6	60
PPEF2	?	ENSG00000156194	subcutaneous adipocytes	1.07×10-6 (p-value)	GTEx Portal V6	58
NAAA	?	ENSG00000138744	subcutaneous adipocytes	8.70×10-6 (p-value)	GTEx Portal V6	1
PPEF2	?	ENSG00000156194	cerebellar hemisphere	7.72×10-6 (p-value)	GTEx Portal V6	1
NAAA	?	ENSG00000138744	unexposed skin	8.29×10-6 (p-value)	GTEx Portal V6	4

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382796	2	ENCP00000041424	CXCL9
		ENCP00000041435	ART3
ENCE00000382774	2	ENCP00000041420	SDAD1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001432205 (promoter flanking region)	2	monocytes (Monocytes-CD14+)	H3K27ac
		muscle (HSMM)	DNase1
ENSR00001685887 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		blood (K562)	H3K27me3
ENSR00001243211 (CTCF binding site)	3	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF

		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	CTCF
		A549	CTCF, H3K36me3
ENSR00001685888 (promoter flanking region)	5	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K36me3
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		lung (IMR90)	CTCF, DNase1
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF, H3K36me3
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NAAA	downstream gene variant	227	ENST00000286733	NM_014435.3	ENSP00000286733	32
NAAA	downstream gene variant	50	ENST00000507956	?	ENSP00000427641	26
NAAA	downstream gene variant	154	ENST00000511606	?	?	34
NAAA	downstream gene variant	256	ENST00000602782	?	ENSP00000473575	24
NAAA	downstream gene variant	94	ENST00000513045	?	ENSP00000423397	34
NAAA	downstream gene variant	10	ENST00000505594	?	ENSP00000426977	24

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	25
NAAA	ENST00000507956	?	ENSP00000427641	12
NAAA	ENST00000511606	?	?	25
NAAA	ENST00000602782	?	ENSP00000473575	17
NAAA	ENST00000513045	?	ENSP00000423397	25

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
NAAA	ENST00000286733	NM_014435.3	ENSP00000286733	10
NAAA	ENST00000507956	?	ENSP00000427641	6

NAAA	ENST00000505594	?	ENSP00000426977	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NAAA	ENST00000511606	?	2

Locus 338 (rs11753208)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,005,432-31,010,159
block size	4,728 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.057 [-0.247 – 0.361]	gene(s) hit or close-by	MUC22
phastCons	μ = 0.002 [0 – 0.003]	eQTL gene(s)	CCHCR1, HCG22, HCG27, MICA, PSORS1C1, PSORS1C2, VARS2, XXbac-BPG181B23.7, XXbac-BPG299F13.17
GERP++	μ = 0.700 [-1.26 – 2.66]	potentially regulated gene(s)	–
CADD score	μ = 5.436 [1.051 – 9.822]	disease gene(s)	VARS2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HCG22	ENST00000570223	ILMN_1667229	blood	6.50×10-6 (p-value)	MuTHER consortium	1
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
MICA	?	ENSG00000204520	muscularis mucosae	3.09×10-6 (p-value)	GTEx Portal V6	1
PSORS1C2	?	ENSG00000204538	muscularis mucosae	3.51×10-8 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	muscularis mucosae	8.34×10-7 (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	lung	7.42×10-8 (p-value)	GTEx Portal V6	2
PSORS1C2	?	ENSG00000204538	lung	3.42×10-6 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	lung	1.13×10-7 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	transformed fibroblasts	8.53×10-7 (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	blood	3.14×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial artery	4.53×10-6 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	tibial artery	1.44×10-7 (p-value)	GTEx Portal V6	2
PSORS1C2	?	ENSG00000204538	thyroid	5.72×10-8 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	thyroid	4.27×10-10 (p-value)	GTEx Portal V6	2

XXbac-BPG299F13.17	?	ENSG00000272501	skeletal muscle	2.90×10^{-7} (p-value)	GTEx Portal V6	2
CCHCR1	?	ENSG00000204536	sun exposed skin	8.04×10^{-7} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	sun exposed skin	1.67×10^{-6} (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	sun exposed skin	1.33×10^{-7} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	aorta	2.63×10^{-8} (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	aorta	4.63×10^{-6} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	subcutaneous adipocytes	3.37×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	1.38×10^{-6} (p-value)	GTEx Portal V6	2
HCG27	ENST00000383331	ILMN_1746436	monocyte	1.68×10^{-22} (p-value)	Zeller et al.	1
HCG27	ENST00000415276					
MICA	?	ENSG00000204520	sigmoid colon	8.14×10^{-7} (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	sigmoid colon	1.35×10^{-6} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	tibial nerve	4.59×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	2.84×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	tibial nerve	9.15×10^{-6} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	esophagus mucosa	2.91×10^{-9} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	esophagus mucosa	1.50×10^{-6} (p-value)	GTEx Portal V6	2
HCG22	?	ENSG00000228789	esophagus mucosa	1.60×10^{-9} (p-value)	GTEx Portal V6	2
VARS2	ENST00000321897	ILMN_1737585	blood	4.68×10^{-5} (p-value)	Westra et al.	1
VARS2	ENST00000625423					
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494543 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1, H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me1, H2AZ, H3K27ac, H3K4me2, DNase1
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MUC22	downstream gene variant	2253	ENST00000561890	NM_001198815.1	ENSP00000455906	1

Locus 338 (sentinel SNP rs11753208 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 339 (rs17708574)

SNiPAcard

Block annotations

Block info

genomic range	chr5:149,521,238-149,521,238
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.925	gene(s) hit or close-by	PDGFRB
phastCons	0	eQTL gene(s)	–
GERP++	-0.18	potentially regulated gene(s)	–
CADD score	7.735	disease gene(s)	PDGFRB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PDGFRB	Familial Infantile Myofibromatosis	DECIPHER	MIM:228550
PDGFRB	JUVENILE MYELOMONOCYTIC LEUKEMIA	OMIM	MIM:607785
PDGFRB	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 4	OMIM	MIM:615007
PDGFRB	MYOFIBROMATOSIS, INFANTILE, 1	OMIM	MIM:228550
PDGFRB	LEUKEMIA, ACUTE MYELOID	OMIM	MIM:601626
PDGFRB	MYELOPROLIFERATIVE DISORDER, CHRONIC, WITH EOSINOPHILIA	OMIM	MIM:131440
PDGFRB	Bilateral striopallidodentate calcinosis	OrphaNet	OrphaNet:1980
PDGFRB	Unclassified chronic myeloproliferative disease	OrphaNet	OrphaNet:86830
PDGFRB	Chronic myelomonocytic leukemia	OrphaNet	OrphaNet:98823
PDGFRB	Idiopathic hypereosinophilic syndrome	OrphaNet	OrphaNet:3260
PDGFRB	Infantile myofibromatosis 1	OrphaNet	OrphaNet:2591
PDGFRB	Myeloid neoplasm associated with PDGFRB rearrangement	OrphaNet	OrphaNet:168950

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001293753 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me1, USF1
		HSMMtube	H3K4me1, H3K9ac, H3K27ac, H3K36me3, DNase1
		blood (K562)	H3K27me3, DNase1, Egr1, PU1, Cjun
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, H3K4me1, H3K36me3
		muscle (HSMM)	H3K27ac, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	FOSL2, Jund, H3K4me3, PolII, TAF1, ELF1, USF1, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF, DNase1
		lung (IMR90)	H3K56ac, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4ac, H2BK120ac, H2BK20ac, H2AK5ac, H3K9ac, H3K4me1, H2BK12ac, DNase1, H4K8ac, H4K91ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me1, H3K4me2, H3K36me3, H3K27ac
		skin (NHEK)	H3K27me3, DNase1
		NHLF	H3K27ac, H3K36me3, H3K9ac, H3K4me1, DNase1
		Osteobl	H3K36me3, H3K4me1, H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Cjun, Jund, H3K4me2, H3K27ac, H3K4me1, Max
		monocytes (Monocytes-CD14+)	H3K27ac, DNase1
		endothelium (HUVEC)	H3K27me3
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDGFRB	upstream gene variant	4238	ENST00000517957	?	ENSP00000430715	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDGFRB	ENST00000520579	?	ENSP00000430026	1
PDGFRB	ENST00000517488	?	ENSP00000429218	1
PDGFRB	ENST00000517660	?	?	1
PDGFRB	ENST00000261799	NM_002609.3	ENSP00000261799	1

Locus 340 (rs2147190)

SNiPAcard

Block annotations

Block info

genomic range	chr13:92,031,820-92,058,888
block size	27,069 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.417 [-2.725 – 1.103]	**gene(s) hit or close-by**	AL138714.1, GPC5
phastCons	μ = 0.010 [0 – 0.12]	**eQTL gene(s)**	GPC5
GERP++	μ = -1.921 [-8.41 – 1.94]	**potentially regulated gene(s)**	–
CADD score	μ = 3.144 [0.08 – 6.815]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GPC5	?	ENSG00000179399	caudate basal ganglia	2.25×10-6 (p-value)	GTEx Portal V6	4
GPC5	?	ENSG00000179399	lung	4.24×10-6 (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001511261 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	CTCF, Rad21
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF
ENSR00001511262 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, Nrsf, TAF1, H3K4me2, H3K4me3, H3K27me3, DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	H3K4me3, DNase1, H3K27me3, H3K4me2, Nrsf, H2AZ, H3K4me1, H3K9ac, Max, Egr1
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	H3K27me3
		liver (HepG2)	H3K4me3, H3K4me2, H3K27me3, DNase1
		blood (GM12878)	H3K4me2, H3K4me3, H2AZ, DNase1, PolII, Nrsf
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3, H3K4me2
		skin (NHEK)	H3K27me3, H3K4me3, H3K4me2, DNase1
		NHLF	H3K27me3
		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K27ac, TAF1, PolII, H3K4me3, H3K79me2, H3K4me2, Nrsf, H3K9ac
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27me3, H3K4me2, H3K4me1
		endothelium (HUVEC)	H3K36me3, H3K27me3, DNase1
		A549	H3K4me3, H3K4me2, H3K27me3

ENSR00001511263 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	DNase1, H3K27me3
		blood (K562)	DNase1, CTCF, Rad21
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	H3K79me2, CTCF
		endothelium (HUVEC)	CTCF
		A549	H3K27me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL138714.1	downstream gene variant, upstream gene variant	29	ENST00000408499	?	?	5
GPC5	upstream gene variant	526	ENST00000377067	NM_004466.4	ENSP00000366267	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPC5	ENST00000377067	NM_004466.4	ENSP00000366267	3

Locus 342 (rs2523987)

SNiPAcard

Block annotations

Block info

genomic range	chr6:30,075,103-30,103,360
block size	28,258 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.710 [-5.32 – 0.731]	gene(s) hit or close-by	SNORA48, TRIM31, TRIM31-AS1, TRIM40
phastCons	μ = 0.018 [0 – 0.413]	eQTL gene(s)	BTN3A2, BTN3A3, HCG22, HCG4P11, HIST1H3I, HLA-A, HLA-C, HLA-DQA1, HLA-DQB1, HLA-F, HLA-F-AS1, HLA-G, HLA-H, HLA-J, HLA-K, HLA-U, HLA-V, IER3, LINC00243, PPP1R11, PPP1R18, TRIM31, TRIM31-AS1, ZFP57, ZNRD1-AS1
GERP++	μ = -0.849 [-5.14 – 1.74]	potentially regulated gene(s)	TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31
CADD score	μ = 4.371 [0.332 – 13.43]	disease gene(s)	HLA-DQB1, ZFP57, HLA-A

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HLA-DQB1	Bullous pemphigoid	OrphaNet	OrphaNet:703
HLA-DQB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073
HLA-DQB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886
HLA-A	Birdshot chorioretinopathy	OrphaNet	OrphaNet:179
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
TRIM31	missense variant	ENST00000376734	NM_007028.3	ENSP00000365924	I/V	Att/Gtt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1654987	blood	6.16×10-6 (p-value)	MuTHER consortium	2
			adipocyte	4.86×10-6 (p-value)		
TRIM31	ENST00000376734	ILMN_1748685	skin	6.26×10-7 (p-value)	MuTHER consortium	17
TRIM31	ENST00000484583					
TRIM31	ENST00000471695					
TRIM31	ENST00000468264					
HLA-V	ENST00000446817	ILMN_1660923	adipocyte	6.57×10-7 (p-value)	MuTHER consortium	18

HLA-V	ENST00000429037		blood	3.42×10-6 (p-value)	MuTHER consortium	18
HCG22	ENST00000570223	ILMN_1667229	blood	2.42×10-5 (p-value)	MuTHER consortium	1
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
HLA-J	?	ENSG00000204622	lung	4.30×10-12 (p-value)	GTEx Portal V6	27
TRIM31-AS1	?	ENSG00000231226	lung	6.84×10-6 (p-value)	GTEx Portal V6	1
HLA-H	?	ENSG00000206341	lung	2.51×10-6 (p-value)	GTEx Portal V6	13
TRIM31	?	ENSG00000204616	lung	3.26×10-8 (p-value)	GTEx Portal V6	8
HLA-U	?	ENSG00000228078	lung	6.51×10-6 (p-value)	GTEx Portal V6	2
HLA-G	?	ENSG00000204632	lung	8.97×10-7 (p-value)	GTEx Portal V6	3
ZFP57	?	ENSG00000204644	lung	8.97×10-7 (p-value)	GTEx Portal V6	1
ZNRD1-AS1	?	ENSG00000204623	tibial nerve	1.75×10-10 (p-value)	GTEx Portal V6	26
HLA-U	?	ENSG00000228078	tibial nerve	3.89×10-6 (p-value)	GTEx Portal V6	18
HLA-F-AS1	?	ENSG00000214922	blood	1.11×10-9 (p-value)	GTEx Portal V6	27
LINC00243	?	ENSG00000214894	blood	4.32×10-9 (p-value)	GTEx Portal V6	22
HLA-J	?	ENSG00000204622	blood	2.28×10-6 (p-value)	GTEx Portal V6	12
HLA-H	?	ENSG00000206341	blood	3.17×10-7 (p-value)	GTEx Portal V6	20
ZFP57	?	ENSG00000204644	blood	3.49×10-7 (p-value)	GTEx Portal V6	2
HLA-K	?	ENSG00000230795	blood	3.41×10-6 (p-value)	GTEx Portal V6	2
ZNRD1-AS1	?	ENSG00000204623	tibial artery	6.13×10-9 (p-value)	GTEx Portal V6	26
HLA-U	?	ENSG00000228078	tibial artery	2.93×10-9 (p-value)	GTEx Portal V6	26
HLA-J	?	ENSG00000204622	tibial artery	7.46×10-8 (p-value)	GTEx Portal V6	24
ZFP57	?	ENSG00000204644	tibial artery	1.09×10-5 (p-value)	GTEx Portal V6	1
HLA-J	?	ENSG00000204622	thyroid	2.05×10-7 (p-value)	GTEx Portal V6	26
ZNRD1-AS1	?	ENSG00000204623	thyroid	1.18×10-5 (p-value)	GTEx Portal V6	12
ZFP57	?	ENSG00000204644	thyroid	1.78×10-8 (p-value)	GTEx Portal V6	18
PPP1R18	?	ENSG00000146112	thyroid	8.21×10-8 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	skeletal muscle	2.77×10-11 (p-value)	GTEx Portal V6	27
HLA-U	?	ENSG00000228078	skeletal muscle	5.12×10-6 (p-value)	GTEx Portal V6	2
HLA-H	?	ENSG00000206341	skeletal muscle	4.48×10-6 (p-value)	GTEx Portal V6	13
HLA-F	?	ENSG00000204642	cerebellum	5.51×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	cerebellum	3.63×10-6 (p-value)	GTEx Portal V6	5
HCG4P11	?	ENSG00000225864	cerebellum	9.28×10-6 (p-value)	GTEx Portal V6	1
HLA-H	?	ENSG00000206341	transformed fibroblasts	1.75×10-6 (p-value)	GTEx Portal V6	7
ZFP57	?	ENSG00000204644	transformed fibroblasts	1.94×10-5 (p-value)	GTEx Portal V6	1
HLA-J	?	ENSG00000204622	breast	1.76×10-7 (p-value)	GTEx Portal V6	15

?	?	ILMN_1679389	b-cell	3.71×10-5 (p-value)	Fairfax et al.	3
IER3	ENST00000376377	ILMN_1682717	monocyte	1.18×10-4 (p-value)	Fairfax et al.	6
IER3	ENST00000259874					
HIST1H3I	ENST00000616365	ILMN_2207865	b-cell	2.37×10-4 (p-value)	Fairfax et al.	1
PPP1R11	ENST00000376765	ILMN_1747598	blood	1.31×10-6 (p-value)	Westra et al.	7
PPP1R11	ENST00000376769					
PPP1R11	ENST00000376763					
PPP1R11	ENST00000376773					
PPP1R11	ENST00000376758					
PPP1R11	ENST00000376772					
ZNRD1-AS1	?	ENSG00000204623	sun exposed skin	7.33×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	sun exposed skin	3.65×10-7 (p-value)	GTEx Portal V6	24
ZFP57	?	ENSG00000204644	sun exposed skin	2.39×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	left ventricle	3.77×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	aorta	4.32×10-7 (p-value)	GTEx Portal V6	6
HLA-H	?	ENSG00000206341	aorta	1.43×10-5 (p-value)	GTEx Portal V6	1
HLA-U	?	ENSG00000228078	aorta	9.62×10-7 (p-value)	GTEx Portal V6	17
ZFP57	?	ENSG00000204644	aorta	2.15×10-8 (p-value)	GTEx Portal V6	18
?	?	ILMN_1666078	monocyte	2.04×10-21 (p-value)	Zeller et al.	3
?	?	ILMN_1671054	monocyte	1.92×10-32 (p-value)	Zeller et al.	3
BTN3A2	ENST00000396934	ILMN_1676528	monocyte	1.37×10-25 (p-value)	Zeller et al.	3
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
?	?	ILMN_1820787	monocyte	4.51×10-17 (p-value)	Zeller et al.	3
BTN3A2	ENST00000524459	ILMN_1700067	monocyte	8.40×10-22 (p-value)	Zeller et al.	3
BTN3A2	ENST00000528541					
BTN3A3	ENST00000467524					
BTN3A3	ENST00000482451					
BTN3A3	ENST00000494393					
BTN3A2	ENST00000527417					
BTN3A3	ENST00000244519					
BTN3A2	ENST00000528222					
BTN3A3	ENST00000474790					
BTN3A2	ENST00000377708					
BTN3A3	ENST00000496719					
BTN3A2	ENST00000396948					
BTN3A2	ENST00000527422					
BTN3A2	ENST00000356386					
?	?	ILMN_1883997	monocyte	1.50×10-36 (p-value)	Zeller et al.	3
?	?	ILMN_1679389	monocyte	4.26×10-12 (p-value)	Zeller et al.	1
ZNRD1-AS1	?	ENSG00000204623	spleen	1.62×10-6 (p-value)	GTEx Portal V6	11
HLA-J	?	ENSG00000204622	esophagus mucosa	9.67×10-6 (p-value)	GTEx Portal V6	12
ZFP57	?	ENSG00000204644	esophagus mucosa	1.51×10-8 (p-value)	GTEx Portal V6	17
ZNRD1-AS1	?	ENSG00000204623	muscularis mucosae	1.12×10-5 (p-value)	GTEx Portal V6	4

ZNRD1-AS1	?	ENSG00000204623	subcutaneous adipocytes	1.34×10^{-5} (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	subcutaneous adipocytes	1.16×10^{-5} (p-value)	GTEx Portal V6	1
HLA-A	?	ENSG00000206503	brain	4.30×10^{-2} (q-value)	SeeQTL DB (Myers et al.)	1
ZFP57	?	ENSG00000204644	visceral adipocytes	7.76×10^{-6} (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
BTN3A2	ENST00000524682	209846_s_at	chr6	blood	7.60×10^{-9} (p-value)	Dixon et al.	1
BTN3A2	ENST00000604202						
BTN3A2	ENST00000396934						
BTN3A2	ENST00000532294						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000377708						
BTN3A2	ENST00000527422						
BTN3A2	ENST00000531055						
BTN3A2	ENST00000396948						
BTN3A2	ENST00000356386						
HLA-DQB1	ENST00000399082	209480_at	chr6	blood	1.80×10^{-9} (p-value)	Dixon et al.	3
HLA-DQB1	ENST00000399084						
HLA-DQB1	ENST00000434651						
HLA-DQB1	ENST00000460185						
HLA-DQB1	ENST00000399079						
HLA-DQB1	ENST00000443574						
HLA-DQB1	ENST00000374943						
HLA-DQB1	ENST00000487676						
HLA-DQA1	ENST00000374949	213831_at	chr6	blood	7.10×10^{-8} (p-value)	Dixon et al.	1
HLA-DQA1	ENST00000496318						
HLA-DQA1	ENST00000460633						
HLA-DQA1	ENST00000482745						
HLA-DQA1	ENST00000343139						
HLA-C	ENST00000470363	216526_x_at	chr6	blood	9.60×10^{-8} (p-value)	Dixon et al.	1
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000466892						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
?	?	ILMN_1752592	chr6	monocyte	6.28×10^{-21} (p-value)	Zeller et al.	3
HLA-C	ENST00000470363	ILMN_2150787	chr6	monocyte	3.65×10^{-13} (p-value)	Zeller et al.	3
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)

ENCP00000047880	TRIM31
	TRIM31
	TRIM31
	TRIM31
	TRIM31
	TRIM31
	TRIM31

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487603 (promoter flanking region)	5	embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	H3K27me3
		liver (HepG2)	H3K4me1, H3K4me2
		lung (IMR90)	H3K27me3
		blood (GM12878)	EBF1
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K27me3
		NHLF	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		A549	H3K4me3
ENSR00001702952 (enhancer)	2	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me1, H3K4me2
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001702953 (CTCF binding site)	4	monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SNORA48	upstream gene variant	2617	ENST00000516419	?	?	1
TRIM31	downstream gene variant, upstream gene variant	510	ENST00000480808	?	?	17
TRIM31	upstream gene variant	30	ENST00000485864	?	?	19
TRIM31	downstream gene variant, upstream gene variant	22	ENST00000493404	?	?	22
TRIM31	upstream gene variant	2299	ENST00000468264	?	?	9
TRIM31	upstream gene variant	1669	ENST00000471695	?	?	3
TRIM31	upstream gene variant	1886	ENST00000484583	?	?	1
TRIM31	upstream gene variant	73	ENST00000376734	NM_007028.3	ENSP00000365924	16
TRIM31-AS1	downstream gene variant	187	ENST00000440874	?	?	10
TRIM40	upstream gene variant	525	ENST00000396581	NM_001286633.1	ENSP00000379826	1
TRIM40	upstream gene variant	541	ENST00000489892	?	?	1

TRIM40	upstream gene variant	1427	ENST00000307859	NM_138700.4	ENSP00000308310	1
TRIM40	upstream gene variant	1148	ENST00000376724	?	ENSP00000365914	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TRIM31	ENST00000376734	NM_007028.3	ENSP00000365924	3	3	3

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TRIM31	ENST00000376734	NM_007028.3	ENSP00000365924	8
TRIM31	ENST00000485864	?	?	6
TRIM31	ENST00000468264	?	?	1
TRIM31	ENST00000480808	?	?	5
TRIM31	ENST00000493404	?	?	3
TRIM31-AS1	ENST00000440874	?	?	15

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TRIM31	ENST00000485864	?	4
TRIM31	ENST00000480808	?	4
TRIM31	ENST00000493404	?	2

Locus 343 (rs11022119)

SNiPAcard

Block annotations

Block info

genomic range	chr11:12,038,874-12,050,652
block size	11,779 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.783 [0.562 – 1.004]	gene(s) hit or close-by	RP13-631K18.2
phastCons	μ = 0.040 [0.009 – 0.072]	eQTL gene(s)	DKK3, USP47
GERP++	μ = 1.406 [-0.558 – 3.37]	potentially regulated gene(s)	MICALCL, RP11-483L5.1
CADD score	μ = 4.825 [2.084 – 7.565]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DKK3	?	ENSG00000050165	gastroesophageal junction	2.41×10^{-6} (p-value)	GTEx Portal V6	2
DKK3	?	ENSG00000050165	muscularis mucosae	4.15×10^{-13} (p-value)	GTEx Portal V6	2
DKK3	?	ENSG00000050165	skeletal muscle	3.01×10^{-6} (p-value)	GTEx Portal V6	1
DKK3	?	ENSG00000050165	tibial artery	1.80×10^{-32} (p-value)	GTEx Portal V6	2
USP47	?	ENSG00000170242	tibial artery	4.65×10^{-6} (p-value)	GTEx Portal V6	1
DKK3	?	ENSG00000050165	aorta	1.52×10^{-23} (p-value)	GTEx Portal V6	2
USP47	?	ENSG00000170242	aorta	3.77×10^{-16} (p-value)	GTEx Portal V6	2
DKK3	?	ENSG00000050165	thyroid	6.76×10^{-11} (p-value)	GTEx Portal V6	2
DKK3	?	ENSG00000050165	subcutaneous adipocytes	1.77×10^{-7} (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000098145	1	ENCP00000011204	MICALCL
		ENCP00000011165	RP11-483L5.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000317680 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	Rad21
		Osteobl	H3K4me2
		blood (K562)	H3K27me3, DNase1, MEF2A
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3

		endothelium (HUVEC)	DNase1, Cjun
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00000317681 e! (open chromatin region)	1	blood (K562)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP13-631K18.2 e!	upstream gene variant	1770	ENST00000531559 e!	?	?	1

Locus 344 (rs2338747)

SNiPAcard

Block annotations

Block info

genomic range	chr10:48,005,225-48,039,886
block size	34,662 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP		**gene(s) hit or close-by**	–
phastCons		**eQTL gene(s)**	–
GERP++		**potentially regulated gene(s)**	–
CADD score		**disease gene(s)**	–

Locus 345 (rs2565722)

SNiPAcard

Block annotations

Block info

genomic range	chr6:161,201,658-161,276,911
block size	75,254 bp
variant count	36 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.186 [-3.076 – 0.897]	gene(s) hit or close-by	RP1-81D8.6
phastCons	μ = 0.067 [0 – 0.519]	eQTL gene(s)	–
GERP++	μ = -0.218 [-1.83 – 0.924]	potentially regulated gene(s)	AGPAT4, RP11-235G24.2
CADD score	μ = 3.584 [0.031 – 10.36]	disease gene(s)	–

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000468570	1	ENCP00000050568	AGPAT4
		ENCP00000050555	RP11-235G24.2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001232628 (open chromatin region)	1	blood (K562)	DNase1
ENSR00001232632 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21, Yy1, DNase1
		liver (HepG2)	CTCF
		HSMMtube	DNase1, H3K27me3
		blood (GM12878)	H3K27ac, Rad21, EBF1
		Osteobl	H3K4me2
		blood (K562)	H2AZ, CTCF
		skin (NHEK)	H3K27ac, H3K4me3
ENSR00001232633 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	H3K27ac
		blood (K562)	PolII, DNase1, H3K27ac, H3K9ac, H2AZ, H3K4me3, H3K4me2
		skin (NHEK)	H3K27ac
ENSR00001232639 (open chromatin region)	1	A549	H3K27me3
		blood (K562)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP1-81D8.6	downstream gene variant	2404	ENST00000619095	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP1-81D8.6	ENST00000619095	?	?	3

Locus 346 (rs6669284)

SNiPAcard

Block annotations

Block info

genomic range	chr1:206,885,839-206,904,464
block size	18,626 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.577 [-2.101 – 0.376]	gene(s) hit or close-by	MAPKAPK2
phastCons	μ = 0.001 [0 – 0.006]	eQTL gene(s)	IL19
GERP++	μ = -0.975 [-4.84 – 2.73]	potentially regulated gene(s)	IKBKE, IL19
CADD score	μ = 4.853 [0.834 – 12.73]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL19	ENST00000620365	ILMN_1799575	blood	2.78×10-6 (p-value)	MuTHER consortium	5
IL19	ENST00000270218					
IL19	ENST00000340758					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000052952	1	ENCP00000006333	IKBKE
		ENCP00000006352	IL19

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001040819 (promoter flanking region)	1	NHLF	H3K27ac
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K79me2
		Osteobl	H3K36me3, H3K27ac
		blood (K562)	H3K79me2, H3K4me1, H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K79me2
		cervix (HeLa-S3)	Jund, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H4K20me1
		lung (IMR90)	DNase1, H4K20me1, H3K79me2
		blood (GM12878)	H3K79me2, H3K27ac
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac
ENSR00000551719 (promoter flanking region)	2	HSMMtube	H3K79me2
		blood (K562)	DNase1, H3K79me2, H3K4me1
		muscle (HSMM)	H3K79me2
		monocytes (Monocytes-CD14+)	H4K20me1
		liver (HepG2)	H3K36me3
		lung (IMR90)	H4K20me1, H3K79me2
		blood (GM12878)	Srf, DNase1, H3K79me2, H3K27ac
		skin (NHEK)	H3K36me3
ENSR00000672024	1	embryonic stem cell (H1ESC)	Rad21, CTCF, Tcf12, DNase1
(CTCF binding site)		HSMMtube	DNase1, CTCF

blood (K562)	DNase1, H3K27me3, Rad21, H4K20me1, CTCF
skin (NHDF-AD)	CTCF
muscle (HSMM)	H3K79me2, CTCF, H4K20me1, DNase1
liver (HepG2)	CTCF
lung (IMR90)	H4K20me1, H3K36me3, CTCF
blood (GM12878)	Pax5, CTCF, H3K79me2, Rad21, DNase1, H3K4me1
nervous (NH-A)	H4K20me1, CTCF
skin (NHEK)	CTCF, H3K36me3
NHLF	CTCF
Osteobl	H4K20me1, CTCF
blood (DND-41)	CTCF, H3K36me3
breast (HMEC)	CTCF
cervix (HeLa-S3)	CTCF, DNase1, H3K4me2, H3K4me1
monocytes (Monocytes-CD14+)	CTCF, H4K20me1, H3K36me3
endothelium (HUVEC)	H3K36me3, CTCF
A549	H3K4me2, CTCF, DNase1, H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MAPKAPK2	upstream gene variant, downstream gene variant	897	ENST00000493447	?	?	4
MAPKAPK2	upstream gene variant	169	ENST00000479009	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MAPKAPK2	ENST00000294981	NM_004759.4	ENSP00000294981	7
MAPKAPK2	ENST00000367103	NM_032960.3	ENSP00000356070	7

Locus 347 (rs939421)

SNiPAcard

Block annotations

Block info

genomic range	chr3:46,576,081-46,585,153
block size	9,073 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.659 [-3.044 – -0.916]	**gene(s) hit or close-by**	LRRC2
phastCons	μ = 0.003 [0 – 0.009]	**eQTL gene(s)**	LRRC2, LRRC2-AS1, TDGF1
GERP++	μ = -1.157 [-2.2 – 0.158]	**potentially regulated gene(s)**	–
CADD score	μ = 1.710 [0.009 – 2.758]	**disease gene(s)**	TDGF1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TDGF1	TERATOCARCINOMA-DERIVED GROWTH FACTOR 1	OMIM	MIM:187395
TDGF1	Semilobar holoprosencephaly	OrphaNet	OrphaNet:220386
TDGF1	Alobar holoprosencephaly	OrphaNet	OrphaNet:93925
TDGF1	Microform holoprosencephaly	OrphaNet	OrphaNet:280200
TDGF1	Midline interhemispheric variant of holoprosencephaly	OrphaNet	OrphaNet:93926
TDGF1	Septopreoptic holoprosencephaly	OrphaNet	OrphaNet:280195
TDGF1	Lobar holoprosencephaly	OrphaNet	OrphaNet:93924

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TDGF1	?	ENSG00000241186	skeletal muscle	2.49×10^{-7} (p-value)	GTEx Portal V6	3
LRRC2-AS1	?	ENSG00000268324	skeletal muscle	1.91×10^{-10} (p-value)	GTEx Portal V6	3
LRRC2	?	ENSG00000163827	testis	3.46×10^{-9} (p-value)	GTEx Portal V6	3
LRRC2-AS1	?	ENSG00000268324	testis	7.24×10^{-12} (p-value)	GTEx Portal V6	3
TDGF1	?	ENSG00000241186	tibial nerve	2.97×10^{-5} (p-value)	GTEx Portal V6	2
LRRC2	?	ENSG00000163827	transformed fibroblasts	1.11×10^{-11} (p-value)	GTEx Portal V6	3
TDGF1	?	ENSG00000241186	left ventricle	9.43×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001362985 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		Osteobl	H3K4me2, H3K27ac, H2AZ
		blood (K562)	H3K27me3

blood (DND-41)	H3K4me1, H3K27ac, H3K9ac
skin (NHDF-AD)	DNase1, H3K4me2
muscle (HSMM)	H3K36me3
monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K4me3
endothelium (HUVEC)	H3K27me3
liver (HepG2)	DNase1
blood (GM12878)	PU1, MEF2C, BATF, MEF2A, H3K4me1, Tcf12, DNase1, IRF4, BCL11A, Pax5, H3K27ac, H3K4me2
lung (IMR90)	DNase1, H3K36me3
A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LRRC2	downstream gene variant	1488	ENST00000496388	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LRRC2	ENST00000296144	?	ENSP00000296144	3
LRRC2	ENST00000395905	NM_024512.4	ENSP00000379241	3

Locus 348 (rs3134906)

SNiPAcard

Block annotations

Block info

genomic range	chr7:142,503,071-142,506,685
block size	3,615 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.812 [-1.269 – -0.306]	gene(s) hit or close-by	TRBV30
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	EPHB6, TRBV30
GERP++	μ = -2.489 [-5.14 – -0.256]	potentially regulated gene(s)	TRBC2, TRBJ2-1, TRBJ2-2, TRBJ2-2P, TRBJ2-4, TRBJ2-5, TRBJ2-6, TRBJ2-7, TRBV10-2, TRBV22-1, TRBV24-1, TRBV25-1, TRBV27, TRBV28, TRBV29-1, TRBV3-1, TRBV4-1, TRBV4-2, TRBV5-1, TRBV5-3, TRBV7-1, TRBV7-3, TRBV7-6, TRBV7-7, TRBV8-1, TRBVA, TRPV5, TRPV5
CADD score	μ = 2.120 [0.315 – 4.347]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
EPHB6	ENST00000486511	ILMN_1802646	adipocyte	2.99×10-6 (p-value)	MuTHER consortium	2
EPHB6	ENST00000616380					
EPHB6	ENST00000611578					
EPHB6	ENST00000411471					
EPHB6	ENST00000617632					
EPHB6	ENST00000614832					
EPHB6	ENST00000476059					
EPHB6	ENST00000619012					
EPHB6	ENST00000471581					
EPHB6	ENST00000422643					
?	?	ILMN_1823172	blood	1.07×10-6 (p-value)	Westra et al.	3
TRBV30	?	ENSG00000237254	blood	2.45×10-7 (p-value)	GTEx Portal V6	5
?	?	ILMN_1822307	b-cell	2.90×10-5 (p-value)	Fairfax et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000500137	1	ENCP00000054229	TRBVA
		ENCP00000054196	TRBV4-2
		ENCP00000054234	TRBV28
		ENCP00000054225	TRBV22-1
		ENCP00000054193	TRBV5-1
		ENCP00000054242	TRBC2
		ENCP00000054233	TRBV27
		ENCP00000054191	TRBV3-1

		ENCP00000054240	TRBJ2-1 TRBJ2-2 TRBJ2-2P
		ENCP00000054192	TRBV4-1
		ENCP00000054203	TRBV7-6
		ENCP00000054228	TRBV25-1
		ENCP00000054221	TRBV8-1
		ENCP00000054227	TRBV24-1
		ENCP00000054212	TRBV10-2
		ENCP00000054247	TRPV5 TRPV5
ENCE00000500138	1	ENCP00000054191	TRBV3-1
		ENCP00000054229	TRBVA
		ENCP00000054196	TRBV4-2
		ENCP00000054234	TRBV28
		ENCP00000054240	TRBJ2-1 TRBJ2-2 TRBJ2-2P
		ENCP00000054192	TRBV4-1
		ENCP00000054203	TRBV7-6
		ENCP00000054225	TRBV22-1
		ENCP00000054228	TRBV25-1
		ENCP00000054193	TRBV5-1
		ENCP00000054221	TRBV8-1
		ENCP00000054242	TRBC2
		ENCP00000054233	TRBV27
		ENCP00000054247	TRPV5 TRPV5
ENCE00000500143	1	ENCP00000054229	TRBVA
		ENCP00000054196	TRBV4-2
		ENCP00000054234	TRBV28
		ENCP00000054225	TRBV22-1
		ENCP00000054193	TRBV5-1
		ENCP00000054236	TRBV29-1
		ENCP00000054242	TRBC2
		ENCP00000054233	TRBV27
		ENCP00000054191	TRBV3-1
		ENCP00000054240	TRBJ2-1 TRBJ2-2 TRBJ2-2P
		ENCP00000054192	TRBV4-1
		ENCP00000054203	TRBV7-6
		ENCP00000054228	TRBV25-1
		ENCP00000054221	TRBV8-1
		ENCP00000054212	TRBV10-2
		ENCP00000054227	TRBV24-1
		ENCP00000054247	TRPV5 TRPV5
ENCE00000500145	1	ENCP00000054229	TRBVA
		ENCP00000054234	TRBV28
		ENCP00000054225	TRBV22-1
		ENCP00000054195	TRBV7-1
		ENCP00000054193	TRBV5-1
		ENCP00000054236	TRBV29-1
		ENCP00000054241	TRBJ2-5 TRBJ2-4 TRBJ2-6

	TRBJ2-7
ENCP00000054242	TRBC2
ENCP00000054233	TRBV27
ENCP00000054191	TRBV3-1
ENCP00000054240	TRBJ2-1 TRBJ2-2 TRBJ2-2P
ENCP00000054192	TRBV4-1
ENCP00000054203	TRBV7-6
ENCP00000054228	TRBV25-1
ENCP00000054221	TRBV8-1
ENCP00000054219	TRBV7-3
ENCP00000054200	TRBV7-7
ENCP00000054217	TRBV5-3
ENCP00000054227	TRBV24-1
ENCP00000054212	TRBV10-2
ENCP00000054247	TRPV5 TRPV5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001718692 (enhancer)	2	endothelium (HUVEC)	PolII
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	DNase1
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3
		A549	H3K27me3
ENSR00000635620 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3, DNase1, Egr1, FOSL1, Max, PU1, ETS1
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3
		cervix (HeLa-S3)	DNase1, H3K27ac
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, Cmyc, PolII, H3K27ac, H3K4me1, DNase1, Cjun
		blood (GM12878)	H3K9ac, H3K4me2, PU1, PolII, BCLAF1, BATF, MEF2A, H3K4me1, ETS1, Tcf12, DNase1, Egr1, H3K4me3, BCL11A, Pax5, H3K27ac, ZEB1
		A549	CTCF, H3K27me3
		nervous (NH-A)	DNase1, H3K9ac, H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
TRBV30	downstream gene variant	3585	ENST00000417977	?	ENSP00000397118	1

Locus 349 (rs8106657)

SNiPAcard

Block annotations

Block info

genomic range	chr19:7,779,733-7,789,796
block size	10,064 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.795 [-1.99 – 0.075]	gene(s) hit or close-by	CLEC4G
phastCons	μ = 0.023 [0 – 0.077]	eQTL gene(s)	CD209, CLEC4G, FCER2, MCEMP1, TGFBR3L, XAB2
GERP++	μ = -0.107 [-0.858 – 0.13]	potentially regulated gene(s)	C19orf45, CCL25
CADD score	μ = 2.398 [0.127 – 5.545]	disease gene(s)	CD209

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CD209	DENGUE VIRUS, SUSCEPTIBILITY TODENGUE FEVER, SUSCEPTIBILITY TO, [...]	OMIM	MIM:614371
CD209	MYCOBACTERIUM TUBERCULOSIS, SUSCEPTIBILITY TOMYCOBACTERIUM TUBERCULOSIS, [...]	OMIM	MIM:607948

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TGFBR3L	?	ENSG00000260001	prostate	3.22×10^{-7} (p-value)	GTEx Portal V6	3
XAB2	?	ENSG00000076924	left ventricle	3.84×10^{-6} (p-value)	GTEx Portal V6	1
FCER2	?	ENSG00000104921	tibial nerve	3.82×10^{-6} (p-value)	GTEx Portal V6	1
FCER2	?	ENSG00000104921	subcutaneous adipocytes	9.02×10^{-7} (p-value)	GTEx Portal V6	2
FCER2	?	ENSG00000104921	visceral adipocytes	3.72×10^{-6} (p-value)	GTEx Portal V6	1
CLEC4G	ENST00000620128	ILMN_2193817	monocyte	4.97×10^{-7} (p-value)	Fairfax et al.	1
CLEC4G	ENST00000328853					
MCEMP1	ENST00000333598	ILMN_1762713	monocyte	7.18×10^{-5} (p-value)	Fairfax et al.	1
MCEMP1	ENST00000597445					
MCEMP1	ENST00000598851					
CLEC4G	ENST00000620128	ILMN_2193817	monocyte	7.61×10^{-45} (p-value)	Zeller et al.	1
CLEC4G	ENST00000328853					
CD209	ENST00000315599	ILMN_1676372	monocyte	1.36×10^{-17} (p-value)	Zeller et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000222798	1	ENCP00000024006	C19orf45
ENCE00000223049	1	ENCP00000024014	CCL25

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CLEC4G	downstream gene variant	4048	ENST00000620128	?	ENSP00000483109	1
CLEC4G	downstream gene variant	4047	ENST00000328853	NM_198492.3, NM_001244856.1	ENSP00000327599	1

Locus 350 (rs2227955)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,777,157-31,834,197
block size	57,041 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.384 [-1.518 – 6.28]	gene(s) hit or close-by	C6orf48, HSPA1A, HSPA1L, LSM2, NEU1, SLC44A4, SNORD48, SNORD52
phastCons	μ = 0.170 [0 – 1]	eQTL gene(s)	–
GERP++	μ = -1.083 [-8.35 – 4.79]	potentially regulated gene(s)	AGER, AGER, AGER, AGER, AGER, AGER, AGER, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, CFB, CFB, CFB, CFB, CFB, CFB, CFB
CADD score	μ = 4.804 [0.077 – 21.2]	disease gene(s)	, , , , , CFB, NEU1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
NEU1	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578
NEU1	Juvenile sialidosis type 2	OrphaNet	OrphaNet:93399
NEU1	Sialidosis type 1	OrphaNet	OrphaNet:812
NEU1	Congenital sialidosis type 2	OrphaNet	OrphaNet:93400
NEU1	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550

Direct effect on transcript

Amino acid sequence alteration

gene	effect	affected	RefSeq id	protein	exchanged	exchanged	SIFT	PolyPhen	variant(s)

	type	transcript			AA's	codons	prediction	prediction	
HSPA1L	missense variant	ENST00000375654	NM_005527.3	ENSP00000364805	A/E	gCa/gAa	?	?	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000048116		C6orf48
		C6orf48
		C6orf48
		C6orf48

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445257	1	ENCP00000048184	AGER
			AGER
			AGER
			AGER
			AGER
			AGER
			AGER
		ENCP00000048138	CFB
			CFB
			CFB
			CFB
			CFB
			CFB
			CFB
			CFB

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000662343 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, H3K4me2, SP1, Srf, H3K4me3, TAF1, H3K9ac, H3K27ac, H3K36me3, H3K27me3, PolII, Nrsf, Sin3Ak20
		HSMMtube	H3K27ac, H3K4me2, DNase1
		blood (K562)	Nrsf, Cfos, PolII, GTF2B, H4K20me1, H3K4me2, CTCF, H3K4me1, H3K36me3, DNase1, H3K4me3, Ini1, Brg1, Egr1, H3K27ac, Max, ZBTB7A, H3K9ac, Gabp, Cmyc, H3K79me2, HEY1, H2AZ, TAF1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, DNase1
		liver (HepG2)	PolII, H3K79me2, TAF1, H3K4me1, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		blood (GM12878)	Nrsf, PolII, H2AZ, DNase1, Egr1, Yy1, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H3K4me3, H3K4ac, H3K9ac, H3K56ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	CTCF, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, Nrsf, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000662344 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K4me3, H3K27me3, CTCF, PolII, Rad21, TAF7, Sin3Ak20, H3K27ac, Yy1, HDAC2, Tcf12, TAF1, H3K4me2, SP1, H3K36me3, H3K9ac
		HSMMtube	H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, CTCF, DNase1
		blood (K562)	H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, Brg1, Egr1, H3K27ac, Max, TAF7, H3K4me1, E2F6, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, PolII, GTF2B, Ini1, Yy1, H4K20me1
		skin (NHDF-AD)	H3K36me3, CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H4K20me1, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1

liver (HepG2)	DNase1, Yy1, H4K20me1, PolII, H3K79me2, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3, H3K36me3
lung (IMR90)	H3K4ac, H3K9ac, CTCF, H3K4me3, H3K36me3, H3K4me2, DNase1, H4K8ac, H4K20me1, H3K79me2, H3K27ac
blood (GM12878)	H3K4me2, H3K9ac, H3K36me3, DNase1, TAF1, PolII, H2AZ, CTCF, Yy1, Rad21, H3K79me2, H3K4me3, H3K27ac
nervous (NH-A)	DNase1, H3K9ac, H4K20me1, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3
skin (NHEK)	H3K36me3, DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3, CTCF
blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
breast (HMEC)	H3K4me2, H3K36me3, H3K9ac, H3K4me3, H3K27ac
cervix (HeLa-S3)	DNase1, PolII, Cmyc, CTCF, H3K36me3, H3K79me2, H3K4me3, H3K9ac, H3K4me2, H3K27ac, TAF1, Max
monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
endothelium (HUVEC)	H3K36me3, CTCF, DNase1, PolII, H3K27ac, H3K9ac, Max, H3K4me2, H3K4me3
A549	H3K4me3, H3K4me2, H3K9ac, H3K36me3, H3K27ac, DNase1, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C6orf48	upstream gene variant	447	ENST00000395788	?	ENSP00000379134	1
HSPA1A	upstream gene variant	377	ENST00000608703	?	ENSP00000477378	2
HSPA1A	upstream gene variant	298	ENST00000375651	NM_005345.5	ENSP00000364802	2
HSPA1L	downstream gene variant, upstream gene variant	101	ENST00000375654	NM_005527.3	ENSP00000364805	2
LSM2	upstream gene variant	2528	ENST00000470086	?	?	2
LSM2	upstream gene variant	2563	ENST00000470083	?	?	2
LSM2	upstream gene variant	2396	ENST00000375661	NM_021177.4	ENSP00000364813	2
LSM2	upstream gene variant	2720	ENST00000493387	?	?	2
LSM2	upstream gene variant	2778	ENST00000475835	?	?	2
LSM2	upstream gene variant	2721	ENST00000491421	?	?	2
LSM2	upstream gene variant	2414	ENST00000477182	?	?	2
NEU1	downstream gene variant, upstream gene variant	3514	ENST00000375631	NM_000434.3	ENSP00000364782	3
NEU1	downstream gene variant, upstream gene variant	2723	ENST00000495807	?	?	3
NEU1	downstream gene variant, upstream gene variant	3615	ENST00000480384	?	?	3
NEU1	downstream gene variant, upstream gene variant	3598	ENST00000491768	?	ENSP00000433127	3
SLC44A4	upstream gene variant	1740	ENST00000487680	?	?	1
SLC44A4	downstream gene variant	3176	ENST00000475563	?	?	1
SNORD48	downstream gene variant	1070	ENST00000364953	?	?	1
SNORD52	upstream gene variant	680	ENST00000364884	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C6orf48	ENST00000375640	?	ENSP00000364791	1
C6orf48	ENST00000375638	NM_001040437.2, NM_001287486.1, NM_001287482.1, NM_001287485.1	ENSP00000364789	1
C6orf48	ENST00000614363	NM_001040438.2, NM_001287484.1	ENSP00000480389	1
C6orf48	ENST00000375635	NM_001287487.1, NM_001287488.1	ENSP00000364786	1
C6orf48	ENST00000375641	?	ENSP00000364792	1
C6orf48	ENST00000375639	NM_001287483.1	ENSP00000364790	1

C6orf48	ENST00000375633	?	ENSP00000364784	1
HSPA1L	ENST00000375654	NM_005527.3	ENSP00000364805	1
SLC44A4	ENST00000229729	NM_025257.2	ENSP00000229729	1
SLC44A4	ENST00000544672	NM_001178045.1	ENSP00000444109	1
SLC44A4	ENST00000414427	?	ENSP00000398901	1
SLC44A4	ENST00000479777	?	?	1
SLC44A4	ENST00000375562	NM_001178044.1	ENSP00000364712	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C6orf48	ENST00000395789	?	ENSP00000379135	1
C6orf48	ENST00000375642	?	ENSP00000364793	1

Locus 351 (rs1051738)

SNiPAcard

Block annotations

Block info

genomic range	chr19:10,547,226-10,591,142
block size	43,917 bp
variant count	22 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.580 [-2.858 – 0.464]	gene(s) hit or close-by	PDE4A
phastCons	μ = 0.023 [0 – 0.128]	eQTL gene(s)	ICAM4, KEAP1, S1PR5, SLC44A2, TYK2
GERP++	μ = -0.510 [-4.39 – 0.392]	potentially regulated gene(s)	KRI1, P2RY11, PPAN, PPAN-P2RY11, TYK2, TYK2, ZGLP1
CADD score	μ = 2.559 [0.035 – 10.39]	disease gene(s)	TYK2, ICAM4, P2RY11

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TYK2	IMMUNODEFICIENCY 35	OMIM	MIM:611521
ICAM4	BLOOD GROUP SYSTEM, LANDSTEINER-WIENER	OMIM	MIM:111250
TYK2	Oligoarticular juvenile arthritis	OrphaNet	OrphaNet:85410
TYK2	Juvenile rheumatoid factor-negative polyarthritis	OrphaNet	OrphaNet:85408
TYK2	Autosomal recessive hyper-IgE syndrome due to TYK2 deficiency	OrphaNet	OrphaNet:331226
P2RY11	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PDE4A	missense variant	ENST00000380702	NM_001111307.1	ENSP00000370078	E/A	gAg/gCg	?	?	1
PDE4A	missense variant	ENST00000344979	NM_006202.2	ENSP00000341007	E/A	gAg/gCg	?	?	1
PDE4A	missense variant	ENST00000440014	NM_001111309.1	ENSP00000394754	E/A	gAg/gCg	?	?	1
PDE4A	missense variant	ENST00000293683	NM_001111308.1	ENSP00000293683	E/A	gAg/gCg	?	?	1
PDE4A	missense variant	ENST00000592685	NM_001243121.1	ENSP00000468507	E/A	gAg/gCg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TYK2	?	ENSG00000105397	breast	4.56×10^{-6} (p-value)	GTEx Portal V6	1
KEAP1	ENST00000590237	ILMN_2410771	blood	7.39×10^{-6} (p-value)	Westra et al.	1
KEAP1	ENST00000592478					
KEAP1	ENST00000590593					
KEAP1	ENST00000393623					
KEAP1	ENST00000171111					

ICAM4	ENST00000380770	ILMN_1681296	blood	7.31×10-6 (p-value)	Westra et al.	1
ICAM4	ENST00000393717					
ICAM4	ENST00000340992					
KEAP1	ENST00000592478	ILMN_2410772	blood	1.09×10-5 (p-value)	Westra et al.	1
KEAP1	ENST00000590237					
KEAP1	ENST00000590593					
KEAP1	ENST00000171111					
KEAP1	ENST00000393623					
SLC44A2	ENST00000589561	ILMN_1771987	blood	5.04×10-5 (p-value)	Westra et al.	1
SLC44A2	ENST00000586078					
SLC44A2	ENST00000586549					
SLC44A2	ENST00000407327					
SLC44A2	ENST00000335757					
S1PR5	ENST00000333430	ILMN_2073184	blood	1.07×10-4 (p-value)	Westra et al.	1
S1PR5	ENST00000617721					
S1PR5	ENST00000439028					
ICAM4	ENST00000380770	ILMN_1681296	monocyte	5.54×10-4 (p-value)	Fairfax et al.	1
ICAM4	ENST00000393717					
ICAM4	ENST00000340992					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000223450	1	ENCP00000024048	TYK2 TYK2
		ENCP00000024033	PPAN
		ENCP00000024032	PPAN
		ENCP00000024047	TYK2 TYK2
		ENCP00000024049	TYK2 TYK2
		ENCP00000024036	PPAN
		ENCP00000024035	PPAN
		ENCP00000024037	P2RY11
		ENCP00000024031	PPAN-P2RY11 PPAN
		ENCP00000024051	KRI1
		ENCP00000024034	PPAN
		ENCP00000024038	ZGLP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000343297 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	H3K36me3
		Osteobl	CTCF
		blood (DND-41)	CTCF

breast (HMEC)	CTCF
cervix (HeLa-S3)	CTCF
monocytes (Monocytes-CD14+)	DNase1, CTCF
endothelium (HUVEC)	H3K36me3, CTCF
A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDE4A	downstream gene variant	327	ENST00000380702	NM_001111307.1	ENSP00000370078	5
PDE4A	downstream gene variant	174	ENST00000586275	?	?	5
PDE4A	downstream gene variant	2337	ENST00000293683	NM_001111308.1	ENSP00000293683	1
PDE4A	downstream gene variant	1211	ENST00000591971	?	ENSP00000467137	5
PDE4A	upstream gene variant, downstream gene variant	1783	ENST00000590407	?	?	3
PDE4A	downstream gene variant	2337	ENST00000440014	NM_001111309.1	ENSP00000394754	1
PDE4A	downstream gene variant	2593	ENST00000589073	?	ENSP00000465792	1
PDE4A	downstream gene variant	2027	ENST00000344979	NM_006202.2	ENSP00000341007	1
PDE4A	downstream gene variant	2309	ENST00000592685	NM_001243121.1	ENSP00000468507	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDE4A	ENST00000591971	?	ENSP00000467137	4
PDE4A	ENST00000293683	NM_001111308.1	ENSP00000293683	11
PDE4A	ENST00000344979	NM_006202.2	ENSP00000341007	9
PDE4A	ENST00000380702	NM_001111307.1	ENSP00000370078	11
PDE4A	ENST00000440014	NM_001111309.1	ENSP00000394754	11
PDE4A	ENST00000592685	NM_001243121.1	ENSP00000468507	11
PDE4A	ENST00000589073	?	ENSP00000465792	9
PDE4A	ENST00000586275	?	?	2
PDE4A	ENST00000590407	?	?	7

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDE4A	ENST00000589073	?	ENSP00000465792	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PDE4A	ENST00000590407	?	1

Locus 352 (rs17412738)

SNiPAcard

Block annotations

Block info

genomic range	chr3:132,185,652-132,410,214
block size	224,563 bp
variant count	68 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.141 [-4.075 – 3.286]	gene(s) hit or close-by	ACAD11, ACKR4, DNAJC13, HSPA8P19, NPHP3, NPHP3-ACAD11, UBA5
phastCons	μ = 0.087 [0 – 1]	eQTL gene(s)	DNAJC13, NPHP3, NPHP3-AS1
GERP++	μ = -0.410 [-10.8 – 5.69]	potentially regulated gene(s)	ACAD11, DNAJC13
CADD score	μ = 3.787 [0.022 – 14.29]	disease gene(s)	NPHP3, DNAJC13

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
methionine	2.75×10^{-6}	Metabolomics GWAS Server	24816252	9
Coronary artery disease	5.43×10^{-5}	dbGaP	pha003031	1

Disease gene annotation

gene	trait	source DB	source entry/link
NPHP3	RENAL-HEPATIC-PANCREATIC DYSPLASIA 1	OMIM	MIM:208540
NPHP3	MECKEL SYNDROME, TYPE 7	OMIM	MIM:267010
NPHP3	NEPHRONOPHTHISIS 3	OMIM	MIM:604387
NPHP3	Meckel syndrome type 7 (MKS7)	DECIPHER	MIM:267010
NPHP3	nephronophthisis type 3 (NPHP3)	DECIPHER	MIM:604387
NPHP3	renal-hepatic-pancreatic dysplasia (RHPD)	DECIPHER	MIM:208540
NPHP3	Late-onset autosomal recessive medullary cystic kidney disease	OrphaNet	OrphaNet:93589
NPHP3	Senior-Loken syndrome	OrphaNet	OrphaNet:3156
NPHP3	NPHP3-related Meckel-like syndrome	OrphaNet	OrphaNet:3032
NPHP3	RENAL-HEPATIC-PANCREATIC DYSPLASIA	OrphaNet	OrphaNet:294415
DNAJC13	Young adult-onset Parkinsonism	OrphaNet	OrphaNet:2828

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ACAD11	missense variant	ENST00000264990	NM_032169.4	ENSP00000264990	T/K	aCg/aAg	?	?	1
ACAD11	missense variant	ENST00000481970	?	ENSP00000420907	T/K	aCg/aAg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NPHP3	?	ENSG00000113971	muscularis mucosae	2.27×10-7 (p-value)	GTEx Portal V6	63
DNAJC13	?	ENSG00000138246	muscularis mucosae	3.42×10-6 (p-value)	GTEx Portal V6	3
NPHP3-AS1	?	ENSG00000248724	transformed fibroblasts	2.27×10-9 (p-value)	GTEx Portal V6	66
NPHP3	?	ENSG00000113971	transformed fibroblasts	1.28×10-13 (p-value)	GTEx Portal V6	66
DNAJC13	?	ENSG00000138246	transformed fibroblasts	5.95×10-6 (p-value)	GTEx Portal V6	37
NPHP3	?	ENSG00000113971	tibial artery	1.54×10-9 (p-value)	GTEx Portal V6	66
NPHP3	?	ENSG00000113971	left ventricle	8.29×10-8 (p-value)	GTEx Portal V6	62
NPHP3	?	ENSG00000113971	blood	1.65×10-9 (p-value)	GTEx Portal V6	66
NPHP3	?	ENSG00000113971	subcutaneous adipocytes	1.71×10-10 (p-value)	GTEx Portal V6	66
DNAJC13	ENST00000509279	ILMN_1752281	blood	1.07×10-5 (p-value)	Westra et al.	6
DNAJC13	ENST00000260818					
NPHP3	?	ENSG00000113971	lung	2.22×10-6 (p-value)	GTEx Portal V6	28
NPHP3	?	ENSG00000113971	aorta	3.78×10-7 (p-value)	GTEx Portal V6	30
NPHP3-AS1	?	ENSG00000248724	tibial nerve	1.81×10-5 (p-value)	GTEx Portal V6	3
NPHP3	?	ENSG00000113971	skeletal muscle	1.16×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000038218		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000354562	1	ENCP00000038224	ACAD11

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001483665 (promoter flanking region)	1	NHLF	H3K36me3, H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (K562)	DNase1
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K27ac
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1, H2BK12ac, H3K18ac, H3K36me3
		A549	H3K36me3
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2
		skin (NHEK)	H3K36me3, H3K4me1
ENSR00001374079 (promoter flanking region)	2	NHLF	DNase1
		blood (K562)	DNase1, H2AZ
		blood (DND-41)	H3K27ac, H3K36me3, H3K4me1

		skin (NHDF-AD)	H3K4me1, DNase1
		endothelium (HUVEC)	H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1
ENSR00001374085 (promoter flanking region)	1	NHLF	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K4me2, H3K27ac
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K4me2, H3K27ac
		breast (HMEC)	H3K27ac
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H2BK12ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K4ac, H2BK120ac, H2BK20ac, H4K91ac, H4K8ac, DNase1
		nervous (NH-A)	H3K4me2, DNase1
		skin (NHEK)	H3K27ac, H3K4me1, DNase1
ENSR00001678918 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	Yy1
		blood (K562)	H3K27me3
ENSR00001678920 (CTCF binding site)	1	Osteobl	H3K36me3
		blood (DND-41)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ACAD11	upstream gene variant	1185	ENST00000264990	NM_032169.4	ENSP00000264990	1
ACAD11	upstream gene variant	3987	ENST00000507705	?	?	1
ACAD11	upstream gene variant	2259	ENST00000496418	?	?	1
ACAD11	downstream gene variant, upstream gene variant	1588	ENST00000489991	?	?	3
ACAD11	upstream gene variant	1842	ENST00000485198	?	ENSP00000419973	1
ACAD11	downstream gene variant, upstream gene variant	2064	ENST00000481970	?	ENSP00000420907	2
ACAD11	upstream gene variant	1826	ENST00000469042	?	?	1
ACAD11	downstream gene variant, upstream gene variant	781	ENST00000510100	?	ENSP00000423796	2
ACAD11	upstream gene variant	1783	ENST00000487024	?	?	2
ACKR4	upstream gene variant, downstream gene variant	535	ENST00000509820	?	ENSP00000425557	2
ACKR4	upstream gene variant	4933	ENST00000249887	NM_178445.2, NM_016557.3	ENSP00000249887	1
DNAJC13	upstream gene variant	626	ENST00000464766	?	?	5
DNAJC13	upstream gene variant, downstream gene variant	319	ENST00000513822	?	?	4
DNAJC13	downstream gene variant	85	ENST00000260818	NM_015268.3	ENSP00000260818	2
DNAJC13	upstream gene variant	307	ENST00000463038	?	?	4
DNAJC13	downstream gene variant	67	ENST00000509279	?	ENSP00000426240	3
DNAJC13	upstream gene variant, downstream gene variant	1710	ENST00000506813	?	ENSP00000421735	10
DNAJC13	downstream gene variant	2468	ENST00000486798	?	?	4
HSPA8P19	downstream gene variant	887	ENST00000505359	?	?	1
NPHP3	downstream gene variant	2677	ENST00000465756	?	ENSP00000419907	1
NPHP3	downstream gene variant	473	ENST00000515289	?	?	2

NPHP3	downstream gene variant	1661	ENST00000474871	?	?	1
NPHP3	downstream gene variant, upstream gene variant	2334	ENST00000493732	?	?	2
NPHP3	downstream gene variant	2680	ENST00000337331	NM_153240.4	ENSP00000338766	1
NPHP3	downstream gene variant	2483	ENST00000490993	?	?	1
NPHP3	downstream gene variant, upstream gene variant	2680	ENST00000512094	?	ENSP00000427666	2
UBA5	downstream gene variant	1759	ENST00000493720	?	ENSP00000417879	1
UBA5	downstream gene variant	2195	ENST00000473651	?	ENSP00000424984	1
UBA5	downstream gene variant	963	ENST00000480955	?	?	1
UBA5	downstream gene variant	1004	ENST00000356232	NM_024818.3	ENSP00000348565	2
UBA5	upstream gene variant, downstream gene variant	851	ENST00000264991	NM_198329.2	ENSP00000264991	3
UBA5	downstream gene variant	1166	ENST00000494238	?	ENSP00000418807	2
UBA5	upstream gene variant	3831	ENST00000489361	?	ENSP00000417905	1
UBA5	downstream gene variant	2407	ENST00000494112	?	?	1
UBA5	downstream gene variant	1166	ENST00000468227	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ACAD11	ENST00000264990	NM_032169.4	ENSP00000264990	I	atT/atC	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
DNAJC13	ENST00000464766	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACAD11	ENST00000489991	?	?	8
ACAD11	ENST00000481970	?	ENSP00000420907	12
ACAD11	ENST00000485198	?	ENSP00000419973	22
ACAD11	ENST00000264990	NM_032169.4	ENSP00000264990	21
ACAD11	ENST00000510100	?	ENSP00000423796	8
ACAD11	ENST00000487024	?	?	11
ACAD11	ENST00000477604	?	?	4
ACAD11	ENST00000496418	?	?	22
ACAD11	ENST00000469042	?	?	22
ACAD11	ENST00000507705	?	?	9
ACKR4	ENST00000249887	NM_178445.2, NM_016557.3	ENSP00000249887	1
ACKR4	ENST00000509820	?	ENSP00000425557	4
DNAJC13	ENST00000509279	?	ENSP00000426240	3
DNAJC13	ENST00000506813	?	ENSP00000421735	2
DNAJC13	ENST00000464766	?	?	11

DNAJC13	ENST00000464766	?	?	11
DNAJC13	ENST00000486798	?	?	1
DNAJC13	ENST00000260818	NM_015268.3	ENSP00000260818	32
HSPA8P19	ENST00000505359	?	?	3
NPHP3	ENST00000515289	?	?	2
NPHP3	ENST00000490993	?	?	5
NPHP3	ENST00000474871	?	?	5
NPHP3	ENST00000465756	?	ENSP00000419907	5
NPHP3	ENST00000337331	NM_153240.4	ENSP00000338766	5
NPHP3	ENST00000512094	?	ENSP00000427666	1
NPHP3-ACAD11	ENST00000471702	?	ENSP00000419763	29
UBA5	ENST00000494238	?	ENSP00000418807	1
UBA5	ENST00000264991	NM_198329.2	ENSP00000264991	1
UBA5	ENST00000473651	?	ENSP00000424984	1
UBA5	ENST00000505777	?	ENSP00000427233	1
UBA5	ENST00000493720	?	ENSP00000417879	1
UBA5	ENST00000356232	NM_024818.3	ENSP00000348565	1
UBA5	ENST00000464068	?	ENSP00000420055	1
UBA5	ENST00000468022	?	ENSP00000418569	1
UBA5	ENST00000464101	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACAD11	ENST00000481970	?	ENSP00000420907	1
ACAD11	ENST00000485198	?	ENSP00000419973	3
DNAJC13	ENST00000260818	NM_015268.3	ENSP00000260818	1
NPHP3-ACAD11	ENST00000471702	?	ENSP00000419763	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ACAD11	ENST00000487024	?	1
ACAD11	ENST00000496418	?	3
ACAD11	ENST00000469042	?	3
ACAD11	ENST00000507705	?	1
HSPA8P19	ENST00000505359	?	1
NPHP3	ENST00000493732	?	1

Locus 353 (rs1354034)

SNiPAcard

Block annotations

Block info

genomic range	chr3:56,849,749-56,849,749
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	1.794	gene(s) hit or close-by	ARHGEF3
phastCons	1	eQTL gene(s)	–
GERP++	4.93	potentially regulated gene(s)	ARHGEF3, CCDC66
CADD score	13.91	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Platelet counts	$<6.00\times10^{-24}$	GWAS Catalog	24026423	1
Mean platelet volume	$<9.00\times10^{-34}$	GWAS Catalog	24026423	1
Platelet counts	$<1.00\times10^{-19}$	GWAS Catalog	23263863	1
Platelet counts	$<3.00\times10^{-54}$	GWAS Catalog	22139419	1
Mean platelet volume	$<3.00\times10^{-69}$	GWAS Catalog	22139419	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000341329	1	ENCP00000036876	ARHGEF3
		ENCP00000036866	CCDC66
		ENCP00000036864	CCDC66
		ENCP00000036861	CCDC66

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ARHGEF3	ENST00000477833	?	ENSP00000418984	1
ARHGEF3	ENST00000481422	?	?	1
ARHGEF3	ENST00000468466	?	ENSP00000418469	1
ARHGEF3	ENST00000477440	?	?	1
ARHGEF3	ENST00000496106	NM_001289698.1	ENSP00000420420	1
ARHGEF3	ENST00000338458	NM_001128615.1	ENSP00000341071	1
ARHGEF3	ENST00000486829	?	ENSP00000420633	1
ARHGEF3	ENST00000498517	?	?	1

ARHGEF3	ENST00000473779	?	ENSP00000420402	1
ARHGEF3	ENST00000468727	?	ENSP00000417087	1

Locus 354 (rs8176720)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,128,467-136,132,873
block size	4,407 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.049 [-2.334 – 0.014]	**gene(s) hit or close-by**	ABO
phastCons	μ = 0.039 [0 – 0.19]	**eQTL gene(s)**	ABO, SURF6
GERP++	μ = -1.687 [-8.39 – 0.522]	**potentially regulated gene(s)**	–
CADD score	μ = 2.053 [0.112 – 6.094]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Serum alkaline phosphatase levels	$<3.00\times10^{-21}$	GWAS Catalog	24094242	1
ADpSGEGDFXAEGGGVR*	2.01×10^{-7}	Metabolomics GWAS Server	24816252	3
E-selectin	7.24×10^{-7}	dbGaP	pha002871	2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABO	?	ENSG00000175164	blood	6.00×10^{-6} (p-value)	GTEx Portal V6	3
SURF6	ENST00000372022	ILMN_1778032	skin	1.54×10^{-5} (p-value)	MuTHER consortium	3
SURF6	ENST00000468290					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABO	downstream gene variant	96	ENST00000538324	?	ENSP00000483018	5
ABO	downstream gene variant	551	ENST00000611156	NM_020469.2	ENSP00000483265	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	T	acG/acA	1
ABO	ENST00000538324	?	ENSP00000483018	T	acG/acA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000453660	?	?	5
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	5
ABO	ENST00000538324	?	ENSP00000483018	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ABO	ENST00000453660	?	6

Locus 355 (rs11672111)

SNiPAcard

Block annotations

Block info

genomic range	chr19:55,562,797-55,566,258
block size	3,462 bp
variant count	22 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.703 [-2.297 – 0.425]	**gene(s) hit or close-by**	CTC-550B14.6, CTC-550B14.7, RDH13
phastCons	μ = 0.016 [0 – 0.209]	**eQTL gene(s)**	GP6
GERP++	μ = 0.034 [-0.765 – 0.547]	**potentially regulated gene(s)**	AC009892.10, AC009892.10, AC009892.5, AC011515.2, AC011515.2, CTC-550B14.1, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, KIR2DL1, KIR2DL1, KIR2DL1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GP6	?	ENSG00000088053	sun exposed skin	1.22×10^{-7} (p-value)	GTEx Portal V6	21
GP6	?	ENSG00000088053	unexposed skin	1.93×10^{-9} (p-value)	GTEx Portal V6	21

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000233760	1	ENCP00000024802	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024807	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
		ENCP00000024823	KIR2DS4 KIR2DS4
		ENCP00000024813	LILRP1 LILRP1

ENCP00000024799	LILRA1 LILRA1 LILRA1 LILRA1
ENCP00000024812	AC011515.2 AC011515.2
ENCP00000024817	KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3
ENCP00000024811	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
ENCP00000024829	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
ENCP00000024822	KIR3DL1 KIR3DL1
ENCP00000024818	KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1

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ENCP00000024816	KIR3DL3
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ENCP00000024803	LILRB1
	LILRB1
	LILRB1
	LILRB1
	LILRB1
ENCP00000024808	VN1R105P
ENCP00000024798	LILRA2
	LILRA2
	LILRA2
	LILRA2
ENCP00000024806	AC009892.10
	AC009892.10
ENCP00000024824	KIR3DL2
	KIR3DL2
	KIR3DL2
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ENCP00000024797	LILRA2
	LILRA2
	LILRA2
	LILRA2
ENCP00000024800	AC009892.5
ENCP00000024821	KIR2DL4
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ENCP00000024825	FCAR
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ENCP00000024810	LILRB4
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ENCP00000024819	KIR2DL1
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ENCE00000233761	1	ENCP00000024802	LILRB1
			LILRB1
			LILRB1
			LILRB1
			LILRB1
		ENCP00000024807	LILRB4
			LILRB4
			LILRB4
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			LILRB4
		ENCP00000024823	KIR2DS4
			KIR2DS4
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		ENCP00000024813	LILRP1
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ENCP00000024799	LILRA1 LILRA1 LILRA1 LILRA1
ENCP00000024812	AC011515.2 AC011515.2
ENCP00000024817	KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3
ENCP00000024811	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
ENCP00000024822	KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1
ENCP00000024818	KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1

	KIR2DP1
ENCP00000024803	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
ENCP00000024827	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 CTC-550B14.1 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
ENCP00000024808	VN1R105P
ENCP00000024798	LILRA2 LILRA2 LILRA2 LILRA2
ENCP00000024806	AC009892.10 AC009892.10
ENCP00000024797	LILRA2 LILRA2 LILRA2 LILRA2
ENCP00000024824	KIR3DL2 KIR3DL2
ENCP00000024800	AC009892.5
ENCP00000024821	KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4

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ENCP00000024825	FCAR
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ENCP00000024810	LILRB4
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ENCP00000024819	KIR2DL1
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Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTC-550B14.6	upstream gene variant	56	ENST00000585492	?	?	9
CTC-550B14.7	downstream gene variant	3343	ENST00000593060	?	?	3
RDH13	downstream gene variant	1763	ENST00000586945	?	ENSP00000466708	21
RDH13	downstream gene variant	4363	ENST00000587026	?	ENSP00000467754	8

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTC-550B14.6	ENST00000585492	?	?	13
RDH13	ENST00000589305	?	?	22
RDH13	ENST00000591960	?	ENSP00000468422	22
RDH13	ENST00000291892	?	ENSP00000291892	22
RDH13	ENST00000396247	NM_138412.3	ENSP00000379547	22
RDH13	ENST00000587721	?	ENSP00000467208	22
RDH13	ENST00000592573	?	ENSP00000465538	22
RDH13	ENST00000589605	?	?	22
RDH13	ENST00000415061	NM_001145971.1	ENSP00000391121	22
RDH13	ENST00000610356	?	ENSP00000477732	22

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTC-550B14.6	ENST00000585492	?	1

Locus 357 (rs520354)

SNiPAcard

Block annotations

Block info

genomic range	chr2:21,239,862-21,262,427
block size	22,566 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.553 [-4.111 – 6.554]	gene(s) hit or close-by	AC010872.1, APOB
phastCons	μ = 0.250 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 0.025 [-4.21 – 5.73]	potentially regulated gene(s)	–
CADD score	μ = 5.580 [0.276 – 18.42]	disease gene(s)	APOB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Lipoproteins, ldl	9.04×10-5	dbGaP	pha002902	dbGaP	1

Variant annotation

trait	type	source DB	source entry/link		Variant(s)
Clinvar: phenotype not specified	likely benign	ClinVar	RCV000116384.2	ClinVar	1
?	HGMD curated	HGMD	CM984191	HGMD®	1

Disease gene annotation

gene	trait	source DB	source entry/link	
APOB	Homozygous familial hypercholesterolemia	OrphaNet	OrphaNet:391665	orphanet
APOB	Familial hypobetalipoproteinemia	OrphaNet	OrphaNet:426	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
APOB	missense variant	ENST00000616098	?	ENSP00000477990	V/A	gTt/gCt	?	?	1
APOB	missense variant	ENST00000233242	NM_000384.2	ENSP00000233242	V/A	gTt/gCt	?	?	1
APOB	missense variant	ENST00000399256	?	ENSP00000382200	V/A	gTt/gCt	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC010872.1	downstream gene variant	3872	ENST00000624225	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOB	ENST00000616098	?	ENSP00000477990	3
APOB	ENST00000233242	NM_000384.2	ENSP00000233242	3
APOB	ENST00000399256	?	ENSP00000382200	2

Locus 358 (rs12202350)

SNiPAcard

Block annotations

Block info

genomic range	chr6:160,329,616-160,418,050
block size	88,435 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.275 [-3.036 – 1.111]	gene(s) hit or close-by	IGF2R, MAS1
phastCons	μ = 0.020 [0 – 0.072]	eQTL gene(s)	IGF2R, RP11-288H12.3, SLC22A1, WTAP
GERP++	μ = 0.277 [-3.11 – 2.66]	potentially regulated gene(s)	SOD2, TCP1
CADD score	μ = 4.532 [0.785 – 13.46]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
isobutyrylcarnitine	5.07×10^{-18}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SLC22A1	?	ENSG00000175003	skeletal muscle	5.46×10^{-6} (p-value)	GTEx Portal V6	3
RP11-288H12.3	?	ENSG00000213073	skeletal muscle	7.15×10^{-9} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	skeletal muscle	3.10×10^{-38} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	tibial nerve	3.07×10^{-9} (p-value)	GTEx Portal V6	8
RP11-288H12.3	?	ENSG00000213073	tibial nerve	1.75×10^{-5} (p-value)	GTEx Portal V6	1
IGF2R	?	ENSG00000197081	atrial appendage	4.52×10^{-9} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	left ventricle	1.39×10^{-7} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	subcutaneous adipocytes	1.16×10^{-9} (p-value)	GTEx Portal V6	8
WTAP	ENST00000358372	ILMN_2279339	blood	1.99×10^{-5} (p-value)	Westra et al.	1
WTAP	ENST00000621533					
IGF2R	?	ENSG00000197081	muscularis mucosae	6.22×10^{-9} (p-value)	GTEx Portal V6	3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000468216	1	ENCP00000050519	TCP1
		ENCP00000050516	SOD2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001502170 (open chromatin region)	1	lung (IMR90)	H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K27me3
		muscle (HSMM)	DNase1
ENSR00001232486 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, CTCF, PolII, Rad21, Jund, Sin3Ak20, ATF3, TAF1, H3K4me2, USF1, H3K9ac
		HSMMtube	H3K4me2, H3K9ac, H3K79me2, H3K27ac, DNase1
		blood (K562)	H2AZ, TAF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, HEY1, H3K79me2, Brg1, Egr1, H3K27ac, Max, Nfe2, E2F6, H3K9ac, USF1, Cmyc
		skin (NHDF-AD)	H3K4me1, H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, H2AZ
		liver (HepG2)	DNase1, H3K9ac, H3K27ac, H3K4me3, H3K36me3, Gabp, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H2AZ, H3K4me2
		blood (GM12878)	ATF3, PolII, H2AZ, DNase1, ELF1, H3K79me2, H3K4me3, USF1, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K9ac, H3K4me3, H3K36me3, H3K4me2, H3K79me2, H4K20me1, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me2, H3K4me3, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K36me3, PolII, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, H3K27me3, PolII, H3K4me2, H3K4me3, Max
		A549	H3K4me3, H3K4me2, H3K9ac
ENSR00001232498 (promoter flanking region)	1	NHLF	DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		blood (K562)	PolII, Ini1, Brg1, Egr1, H3K27ac, Max, PU1, Nfe2, H3K9ac, USF1, H3K79me2, H3K4me3, H3K4me2, H3K4me1, H3K36me3, DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	DNase1, H3K79me2, H3K36me3
		blood (GM12878)	PU1, H3K79me2, H3K27ac, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IGF2R	upstream gene variant	1571	ENST00000356956	NM_000876.2	ENSP00000349437	1
MAS1	downstream gene variant	1213	ENST00000252660	NM_002377.2	ENSP00000252660	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IGF2R	ENST00000356956	NM_000876.2	ENSP00000349437	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MAS1	ENST00000252660	NM_002377.2	ENSP00000252660	1

Locus 359 (rs35871176)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,366,820-34,375,832
block size	9,013 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.167 [-0.156 – 0.434]	**gene(s) hit or close-by**	–
phastCons	μ = 0.208 [0.013 – 0.709]	**eQTL gene(s)**	–
GERP++	μ = -0.199 [-0.794 – 0]	**potentially regulated gene(s)**	–
CADD score	μ = 2.542 [0.796 – 5.671]	**disease gene(s)**	–

Locus 360 (rs13318932)

SNiPAcard

Block annotations

Block info

genomic range	chr3:98,396,350-98,400,767
block size	4,418 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.086 [0.056 – 0.116]	**gene(s) hit or close-by**	–
phastCons	μ = 0.213 [0.056 – 0.371]	**eQTL gene(s)**	DCBLD2, ST3GAL6
GERP++	μ = 0.000 [0 – 0]	**potentially regulated gene(s)**	–
CADD score	μ = 4.716 [0.656 – 8.777]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DCBLD2	?	ENSG00000057019	transformed fibroblasts	8.90×10^{-7} (p-value)	GTEx Portal V6	1
ST3GAL6	?	ENSG00000064225	transformed fibroblasts	4.10×10^{-6} (p-value)	GTEx Portal V6	1
DCBLD2	?	ENSG00000057019	thyroid	1.60×10^{-5} (p-value)	GTEx Portal V6	1

Locus 361 (rs12411860)

SNiPAcard

Block annotations

Block info

genomic range	chr10:52,032,893-52,043,336
block size	10,444 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.432 [-3.86 – 0.643]	**gene(s) hit or close-by**	–
phastCons	μ = 0.010 [0 – 0.177]	**eQTL gene(s)**	–
GERP++	μ = -0.877 [-5.99 – 3.01]	**potentially regulated gene(s)**	–
CADD score	μ = 2.725 [0.089 – 7.564]	**disease gene(s)**	–

Locus 362 (rs9534338)

SNiPAcard

Block annotations

Block info

genomic range	chr13:46,706,598-46,723,294
block size	16,697 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.108 [-0.764 – 0.273]	**gene(s) hit or close-by**	LCP1
phastCons	μ = 0.001 [0 – 0.002]	**eQTL gene(s)**	–
GERP++	μ = 0.349 [-1.92 – 3.66]	**potentially regulated gene(s)**	–
CADD score	μ = 5.530 [3.363 – 9.165]	**disease gene(s)**	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LCP1	downstream gene variant	3622	ENST00000416500	?	ENSP00000408052	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LCP1	ENST00000494531	?	?	1
LCP1	ENST00000323076	NM_002298.4	ENSP00000315757	3
LCP1	ENST00000398576	?	ENSP00000381581	3
LCP1	ENST00000469227	?	?	1

Locus 363 (rs858267)

SNiPAcard

Block annotations

Block info

genomic range	chr7:23,206,329-23,321,521
block size	115,193 bp
variant count	33 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.086 [-3.04 – 1.42]	gene(s) hit or close-by	GPNMB, KLHL7
phastCons	μ = 0.123 [0 – 0.996]	eQTL gene(s)	AC005082.12
GERP++	μ = -0.219 [-6.54 – 3.09]	potentially regulated gene(s)	GPNMB, TOMM7
CADD score	μ = 5.205 [0.224 – 17.15]	disease gene(s)	KLHL7

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
KLHL7	RETINITIS PIGMENTOSA 42	OMIM	MIM:612943
KLHL7	Retinitis pigmentosa	OrphaNet	OrphaNet:791

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AC005082.12	?	ENSG00000226816	blood	3.64×10^{-11} (p-value)	GTEx Portal V6	29
AC005082.12	?	ENSG00000226816	subcutaneous adipocytes	5.45×10^{-8} (p-value)	GTEx Portal V6	8

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000781966	1	ENST00000381990, ENST00000258733, ENST00000465673	GPNMB
FFCP00000781967	1	ENST00000381990, ENST00000258733, ENST00000465673	GPNMB

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000476393	1	ENCP00000051286	TOMM7

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000065469 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, H3K27me3, H3K4me2, USF1
		HSMMtube	DNase1, H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1
		liver (HepG2)	H3K4me2, H3K27me3
		lung (IMR90)	H3K27me3

		blood (GM12878)	H3K4me2, H3K27me3, PU1, H2AZ
		nervous (NH-A)	DNase1, H3K4me2, H3K27me3
		skin (NHEK)	DNase1, H3K4me1, H3K4me2, H3K4me3, H3K36me3
		NHLF	H3K4me3, H3K27me3, DNase1
		Osteobl	H3K4me2, H2AZ, H3K27ac, H3K4me3
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, H3K4me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K27me3
		A549	H3K27me3
ENSR00001558007 (promoter flanking region)	1	HSMMtube	DNase1, H3K27me3
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K36me3
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K36me3
ENSR00000065473 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	H3K36me3, CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GPNMB	downstream gene variant	1404	ENST00000409458	?	ENSP00000386476	3
GPNMB	downstream gene variant	786	ENST00000459927	?	?	3
GPNMB	downstream gene variant	889	ENST00000474157	?	?	2
GPNMB	downstream gene variant	926	ENST00000492858	?	?	3
GPNMB	upstream gene variant, downstream gene variant	1782	ENST00000492512	?	?	4
GPNMB	downstream gene variant	43	ENST00000487890	?	?	4
GPNMB	downstream gene variant	778	ENST00000465673	?	?	3
GPNMB	upstream gene variant, downstream gene variant	1455	ENST00000478451	?	?	5
GPNMB	downstream gene variant	1077	ENST00000381990	NM_001005340.1	ENSP00000371420	2
GPNMB	upstream gene variant, downstream gene variant	25	ENST00000468723	?	?	6
GPNMB	upstream gene variant, downstream gene variant	223	ENST00000479625	?	?	6
GPNMB	downstream gene variant	1076	ENST00000258733	NM_002510.2	ENSP00000258733	2
GPNMB	upstream gene variant, downstream gene variant	650	ENST00000470994	?	?	6
GPNMB	upstream gene variant, downstream gene variant	323	ENST00000463011	?	?	6

GPNMB	upstream gene variant, downstream gene variant	323	ENST00000463011	?	?	6
KLHL7	downstream gene variant	625	ENST00000469576	?	?	10
KLHL7	upstream gene variant, downstream gene variant	885	ENST00000469845	?	?	11

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
GPNMB	ENST00000409458	?	ENSP00000386476	D	gaT/gaC	1
GPNMB	ENST00000258733	NM_002510.2	ENSP00000258733	D	gaT/gaC	1
GPNMB	ENST00000381990	NM_001005340.1	ENSP00000371420	D	gaT/gaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPNMB	ENST00000409458	?	ENSP00000386476	4
GPNMB	ENST00000487890	?	?	3
GPNMB	ENST00000258733	NM_002510.2	ENSP00000258733	14
GPNMB	ENST00000479625	?	?	3
GPNMB	ENST00000492858	?	?	3
GPNMB	ENST00000492512	?	?	1
GPNMB	ENST00000381990	NM_001005340.1	ENSP00000371420	14
GPNMB	ENST00000459927	?	?	2
GPNMB	ENST00000470994	?	?	3
GPNMB	ENST00000465673	?	?	5
KLHL7	ENST00000521082	?	ENSP00000430351	12
KLHL7	ENST00000469576	?	?	2
KLHL7	ENST00000339077	NM_001031710.2	ENSP00000343273	12
KLHL7	ENST00000409689	NM_018846.4	ENSP00000386263	12

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPNMB	ENST00000409458	?	ENSP00000386476	1
GPNMB	ENST00000258733	NM_002510.2	ENSP00000258733	2
GPNMB	ENST00000381990	NM_001005340.1	ENSP00000371420	2
KLHL7	ENST00000521082	?	ENSP00000430351	1
KLHL7	ENST00000339077	NM_001031710.2	ENSP00000343273	1
KLHL7	ENST00000409689	NM_018846.4	ENSP00000386263	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
GPNMB	ENST00000478451	?	1
GPNMB	ENST00000492858	?	1
GPNMB	ENST00000465673	?	1

Locus 364 (rs3795559)

SNiPAcard

Block annotations

Block info

genomic range	chr1:204,987,953-204,997,928
block size	9,976 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.246 [-0.256 – 1.049]	gene(s) hit or close-by	NFASC
phastCons	μ = 0.011 [0 – 0.048]	eQTL gene(s)	–
GERP++	μ = -1.266 [-6.36 – 0.72]	potentially regulated gene(s)	LRRN2, NFASC, RP11-383G10.1, TMCC2
CADD score	μ = 5.745 [2.233 – 14.53]	disease gene(s)	–

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000391750	1	ENST00000495396, ENST00000503221	NFASC

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000052164	1	ENCP00000006256	NFASC
		ENCP00000006273	TMCC2
		ENCP00000006255	NFASC
		ENCP00000006243	LRRN2
		ENCP00000006265	RP11-383G10.1
		ENCP00000006269	TMCC2
		ENCP00000006247	NFASC

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NFASC	downstream gene variant	454	ENST00000404076	?	ENSP00000385676	4
NFASC	downstream gene variant	454	ENST00000425360	?	ENSP00000395664	4
NFASC	downstream gene variant	543	ENST00000401399	?	ENSP00000385637	2
NFASC	downstream gene variant	86	ENST00000447819	?	ENSP00000416891	4
NFASC	downstream gene variant	543	ENST00000495396	?	?	2
NFASC	downstream gene variant	2439	ENST00000413225	?	ENSP00000393290	3
NFASC	downstream gene variant	544	ENST00000339876	NM_001005388.2	ENSP00000344786	2
NFASC	downstream gene variant	454	ENST00000404907	?	ENSP00000384061	4
NFASC	downstream gene variant	1887	ENST00000513543	?	ENSP00000425908	3
NFASC	downstream gene variant	454	ENST00000367173	?	ENSP00000356141	4
NFASC	downstream gene variant	544	ENST00000360049	NM_015090.3	ENSP00000353154	2
NFASC	downstream gene variant	1887	ENST00000504476	?	ENSP00000422524	3
NFASC	downstream gene variant	544	ENST00000539706	NM_001160332.1	ENSP00000438614	2
NFASC	downstream gene variant	454	ENST00000430393	NM_001160331.1	ENSP00000415031	4
NFASC	downstream gene variant	2334	ENST00000503221	?	?	1

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
NFASC	ENST00000539706	NM_001160332.1	ENSP00000438614	3
NFASC	ENST00000360049	NM_015090.3	ENSP00000353154	3
NFASC	ENST00000401399	?	ENSP00000385637	3
NFASC	ENST00000339876	NM_001005388.2	ENSP00000344786	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NFASC	ENST00000495396	?	3
NFASC	ENST00000503221	?	3

Locus 365 (rs1013579)

SNiPAcard

Block annotations

Block info

genomic range	chr1:57,422,484-57,422,484
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.535	gene(s) hit or close-by	C8B
phastCons	0.998	eQTL gene(s)	–
GERP++	-0.165	potentially regulated gene(s)	–
CADD score	17.12	disease gene(s)	C8B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
C8B	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
C8B	missense variant	ENST00000535057	NM_001278544.1	ENSP00000440113	G/R	Gga/Aga	tolerated	benign	1
C8B	missense variant	ENST00000543257	NM_001278543.1	ENSP00000442548	G/R	Gga/Aga	tolerated	benign	1
C8B	missense variant	ENST00000371237	NM_000066.3	ENSP00000360281	G/R	Gga/Aga	tolerated	benign	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C8B	downstream gene variant	44	ENST00000468990	?	?	1
C8B	downstream gene variant	3252	ENST00000494324	?	?	1

Locus 366 (rs2056492)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,318,565-34,352,415
block size	33,851 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.150 [-0.96 – 1.902]	gene(s) hit or close-by	CCL14, CCL15, CCL15-CCL14, CCL23, CTB-186H2.3, RP11-104J23.1, RP11-104J23.2
phastCons	μ = 0.144 [0.001 – 0.834]	eQTL gene(s)	–
GERP++	μ = 0.306 [-0.237 – 2.23]	potentially regulated gene(s)	–
CADD score	μ = 4.766 [0.246 – 14.22]	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL14	upstream gene variant	4771	ENST00000618404	NM_032963.3	ENSP00000481023	1
CCL14	upstream gene variant	4525	ENST00000614009	?	ENSP00000483186	1
CCL14	upstream gene variant	4880	ENST00000622526	NM_032962.4	ENSP00000479097	1
CCL14	upstream gene variant	4854	ENST00000620991	?	ENSP00000484818	1
CCL15	downstream gene variant, upstream gene variant	1297	ENST00000617897	NM_032965.4	ENSP00000484078	3
CCL15	downstream gene variant	155	ENST00000614368	?	ENSP00000484262	3
CCL15-CCL14	upstream gene variant	2301	ENST00000616694	?	ENSP00000481402	1
CCL15-CCL14	upstream gene variant	2371	ENST00000610751	?	ENSP00000481940	1
CCL23	downstream gene variant	592	ENST00000613876	?	ENSP00000479076	3
CCL23	downstream gene variant	593	ENST00000615050	NM_005064.4	ENSP00000481357	3
CCL23	downstream gene variant	591	ENST00000612516	NM_145898.2	ENSP00000484748	3
CTB-186H2.3	downstream gene variant	4819	ENST00000617687	?	?	1
CTB-186H2.3	downstream gene variant	1259	ENST00000612584	?	?	2
RP11-104J23.1	upstream gene variant, downstream gene variant	580	ENST00000617328	?	?	3
RP11-104J23.1	downstream gene variant	2053	ENST00000619334	?	?	1
RP11-104J23.2	upstream gene variant	39	ENST00000617747	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	3
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	3
RP11-104J23.1	ENST00000619334	?	?	3

Locus 366 (sentinel SNP rs2056492 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 367 (rs1403694)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,437,296-186,439,998
block size	2,703 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.876 [-3.124 – 0.439]	gene(s) hit or close-by	KNG1, RP11-573D15.8
phastCons	μ = 0.009 [0 – 0.152]	eQTL gene(s)	KNG1
GERP++	μ = -0.343 [-6.02 – 1.81]	potentially regulated gene(s)	RP11-48F14.1
CADD score	μ = 2.657 [0.161 – 8.36]	disease gene(s)	KNG1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
bradykinin, des-arg(9)	2.21×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
KNG1	HIGH MOLECULAR WEIGHT KININOGEN DEFICIENCY	OMIM	MIM:228960
KNG1	Congenital high-molecular-weight kininogen deficiency	OrphaNet	OrphaNet:483

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
KNG1	?	ENSG00000113889	frontal cortex	2.73×10^{-9} (p-value)	GTEx Portal V6	20
KNG1	?	ENSG00000113889	cortex	1.16×10^{-6} (p-value)	GTEx Portal V6	13

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000365623	2	ENCP00000039492	RP11-48F14.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001381866 (open chromatin region)	2	blood (K562)	H3K27me3, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	upstream gene variant	1003	ENST00000630726	?	?	21

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	ENST00000628601	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KNG1	ENST00000265023	NM_001102416.2	ENSP00000265023	21
KNG1	ENST00000447445	NM_001166451.1	ENSP00000396025	21
KNG1	ENST00000287611	NM_000893.3	ENSP00000287611	21
RP11-573D15.8	ENST00000626633	?	?	21
RP11-573D15.8	ENST00000630331	?	?	21
RP11-573D15.8	ENST00000625303	?	?	21
RP11-573D15.8	ENST00000629106	?	?	21
RP11-573D15.8	ENST00000625386	?	?	21
RP11-573D15.8	ENST00000628190	?	?	21
RP11-573D15.8	ENST00000626845	?	?	21
RP11-573D15.8	ENST00000627919	?	?	21
RP11-573D15.8	ENST00000629451	?	?	21
RP11-573D15.8	ENST00000630178	?	?	21
RP11-573D15.8	ENST00000630864	?	?	21
RP11-573D15.8	ENST00000627830	?	?	21
RP11-573D15.8	ENST00000625741	?	?	21
RP11-573D15.8	ENST00000626151	?	?	21
RP11-573D15.8	ENST00000627469	?	?	21
RP11-573D15.8	ENST00000354642	?	?	21
RP11-573D15.8	ENST00000627551	?	?	21
RP11-573D15.8	ENST00000628601	?	?	21
RP11-573D15.8	ENST00000628253	?	?	21
RP11-573D15.8	ENST00000628505	?	?	21
RP11-573D15.8	ENST00000625839	?	?	21
RP11-573D15.8	ENST00000599314	?	?	21
RP11-573D15.8	ENST00000628728	?	?	21

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-573D15.8	ENST00000628601	?	2

Locus 369 (rs16971895)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,383,107-34,391,237
block size	8,131 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.784 [-2.213 – 0.281]	**gene(s) hit or close-by**	CCL18
phastCons	μ = 0.008 [0 – 0.027]	**eQTL gene(s)**	–
GERP++	μ = -2.376 [-4.62 – 0.235]	**potentially regulated gene(s)**	–
CADD score	μ = 1.184 [0.023 – 3.986]	**disease gene(s)**	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL18	upstream gene variant	403	ENST00000616054	NM_002988.2	ENSP00000479955	3

Locus 370 (rs7259090)

SNiPAcard

Block annotations

Block info

genomic range	chr19:55,392,646-55,392,646
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-2.068	gene(s) hit or close-by	FCAR
phastCons	0.09	eQTL gene(s)	–
GERP++	0	potentially regulated gene(s)	–
CADD score	2.603	disease gene(s)	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348895 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		Osteobl	CTCF
		blood (K562)	DNase1, H3K27me3, CTCF, Rad21
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3, DNase1, CTCF, H4K20me1
		endothelium (HUVEC)	CTCF
		liver (HepG2)	CTCF, Rad21, DNase1
		blood (GM12878)	CTCF
		A549	H3K27me3, DNase1, CTCF

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCAR	ENST00000482092	?	?	1
FCAR	ENST00000355524	NM_002000.2	ENSP00000347714	1
FCAR	ENST00000472634	?	?	1
FCAR	ENST00000488066	?	ENSP00000474512	1
FCAR	ENST00000391726	NM_133273.2	ENSP00000375606	1
FCAR	ENST00000353758	NM_133277.2	ENSP00000338058	1
FCAR	ENST00000345937	NM_133271.2	ENSP00000338257	1
FCAR	ENST00000391724	NM_133278.2	ENSP00000375604	1
FCAR	ENST00000469767	NM_133279.2	ENSP00000473814	1
FCAR	ENST00000359272	NM_133272.2	ENSP00000352218	1
FCAR	ENST00000471750	?	?	1
FCAR	ENST00000391723	NM_133274.2	ENSP00000375603	1
FCAR	ENST00000391725	NM_133269.2	ENSP00000375605	1

Locus 371 (rs4962130)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,195,607-136,201,191
block size	5,585 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.777 [-0.783 – -0.771]	gene(s) hit or close-by	MED22, RP11-244N20.7, SURF6, Y_RNA
phastCons	μ = 0.005 [0 – 0.011]	eQTL gene(s)	–
GERP++	μ = -0.451 [-0.451 – -0.451]	potentially regulated gene(s)	–
CADD score	μ = 2.429 [0.053 – 4.804]	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MED22	downstream gene variant	3968	ENST00000610672	?	ENSP00000482438	1
RP11-244N20.7	upstream gene variant	4635	ENST00000439130	?	?	1
SURF6	downstream gene variant	3	ENST00000372022	NM_006753.5	ENSP00000361092	1
SURF6	downstream gene variant, upstream gene variant	1416	ENST00000468290	?	?	2
Y_RNA	downstream gene variant	3373	ENST00000364581	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SURF6	ENST00000372022	NM_006753.5	ENSP00000361092	1

Locus 372 (rs2239805)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,380,208-32,428,862
block size	48,655 bp
variant count	71 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.425 [-2.801 – 2.075]	**gene(s) hit or close-by**	HLA-DRA, HLA-DRB9
phastCons	μ = 0.034 [0 – 0.454]	**eQTL gene(s)**	APOM, C4A, C4B, C6orf25, CYP21A1P, CYP21A2, HCP5, HLA-DMA, HLA-DMB, HLA-DOB, HLA-DPB1, HLA-DQA2, HLA-DRA, HLA-DRB1, HLA-DRB5, HLA-DRB6, HLA-DRB9, LIMS1, LST1, LY6G6C, MICB, NOTCH4, PSMB9, RNF5, TNXA, XXbac-BPG246D15.9, XXbac-BPG300A18.13
GERP++	μ = -0.137 [-3.41 – 3.16]	**potentially regulated gene(s)**	BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, C4B, C4B, C4B, C4B, C4B, C4B, CYP21A2, CYP21A2, CYP21A2, CYP21A2, CYP21A2, CYP21A2, FKBPL, FKBPL, FKBPL, FKBPL, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DQA1, HLA-DQA1, HLA-DQA1, HLA-DQA1, HLA-DQA1, HLA-DQA1, HLA-DQB1, HLA-DQB1, HLA-DQB1, HLA-DQB1, HLA-DQB1, HLA-DQB1, HLA-DQB1, HLA-DQB2, HLA-DQB2, HLA-DQB2, HLA-DQB2, HLA-DQB2, HLA-DQB2, HLA-DQB2, HLA-DQB2, HLA-DRA, HLA-DRB5, PBX2, PBX2, PBX2, PBX2, PBX2, PBX2, PBX2, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1, PSMB9, PSMB9, PSMB9, PSMB9, PSMB9, PSMB9, PSMB9, PSMB9, PSMB9, TAP1, TAP1, TAP1, TAP1, TAP1, TAP1, TAP1, TAP1, TAP1
CADD score	μ = 3.146 [0.008 – 11.41]	**disease gene(s)**	C4B, C4A, CYP21A2, HLA-DRA, , , , , , , , , BTNL2, , , , , , , , ,

	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
BTNL2	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
TAP1	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	OrphaNet	OrphaNet:315306	orphanet
CYP21A2	Nonclassic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	OrphaNet	OrphaNet:95698	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	OrphaNet	OrphaNet:315311	orphanet
HLA-DPB1	Chronic berylliosis	OrphaNet	OrphaNet:133	orphanet
HLA-DPB1	Granulomatosis with polyangiitis	OrphaNet	OrphaNet:900	orphanet
HLA-DRB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
HLA-DRB1	Limited systemic sclerosis	OrphaNet	OrphaNet:220407	orphanet
HLA-DRB1	Limited cutaneous systemic sclerosis	OrphaNet	OrphaNet:220402	orphanet
HLA-DRB1	Follicular lymphoma	OrphaNet	OrphaNet:545	orphanet
HLA-DRB1	Bullous pemphigoid	OrphaNet	OrphaNet:703	orphanet
HLA-DRB1	Diffuse cutaneous systemic sclerosis	OrphaNet	OrphaNet:220393	orphanet
HLA-DRB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465	orphanet
HLA-DRB1	Sarcoidosis	OrphaNet	OrphaNet:797	orphanet
HLA-DRA	Graham Little-Piccardi-Lassueur syndrome	OrphaNet	OrphaNet:505	orphanet
BTNL2	Sarcoidosis	OrphaNet	OrphaNet:797	orphanet
HLA-DQB1	Bullous pemphigoid	OrphaNet	OrphaNet:703	orphanet
HLA-DQB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
HLA-DQB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465	orphanet
TAP1	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NOTCH4	ENST00000474612	ILMN_1711157	adipocyte	1.12×10-10 (p-value)	MuTHER consortium	36
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
HLA-DPB1	ENST00000418931	ILMN_1749070	skin	4.45×10-9 (p-value)	MuTHER consortium	36
HLA-DPB1	ENST00000416804		blood	1.14×10-7 (p-value)	MuTHER consortium	36
			adipocyte	2.96×10-7 (p-value)	MuTHER consortium	36
MICB	ENST00000538442	ILMN_1708006	skin	1.36×10-8 (p-value)	MuTHER consortium	36
MICB	ENST00000399150					
MICB	ENST00000252229					
PSMB9	ENST00000467593	ILMN_1798233	blood	2.24×10-8 (p-value)	MuTHER consortium	36
PSMB9	ENST00000464863					
HLA-DMA	ENST00000395305	ILMN_1695311	blood	5.67×10-6 (p-value)	MuTHER consortium	32
HLA-DMA	ENST00000480785					
HLA-DMA	ENST00000464392					
HLA-DMA	ENST00000456800					
HLA-DMA	ENST00000395303					
HLA-DMA	ENST00000374843					
HLA-DMA	ENST00000477541					
HLA-DMB	ENST00000487996	ILMN_1761733	blood	3.21×10-5 (p-value)	MuTHER consortium	1
HLA-DMB	ENST00000498020					
HLA-DMB	ENST00000477537					
HLA-DMB	ENST00000418107					
HLA-DMB	ENST00000414017					
HLA-DMB	ENST00000438510					
HCP5	ENST00000414046	ILMN_1803945	adipocyte	3.50×10-5 (p-value)	MuTHER consortium	1
HCP5	ENST00000541196					
C4A	?	ENSG00000244731	muscularis mucosae	1.81×10-12 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	muscularis mucosae	2.03×10-6 (p-value)	GTEx Portal V6	10
C4A	?	ENSG00000244731	lung	1.13×10-9 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	lung	1.08×10-7 (p-value)	GTEx Portal V6	64
HLA-DRB6	?	ENSG00000229391	lung	4.18×10-6 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	atrial appendage	5.98×10-11 (p-value)	GTEx Portal V6	69
C4B	?	ENSG00000224389	transformed fibroblasts	4.39×10-8 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	transformed fibroblasts	9.62×10-14 (p-value)	GTEx Portal V6	69
C4B	?	ENSG00000224389	blood	4.64×10-10 (p-value)	GTEx Portal V6	67

C4A	?	ENSG00000244731	blood	3.57×10-11 (p-value)	GTEx Portal V6	69
HLA-DRB6	?	ENSG00000229391	blood	1.25×10-9 (p-value)	GTEx Portal V6	69
HLA-DQA2	?	ENSG00000237541	blood	3.38×10-8 (p-value)	GTEx Portal V6	19
HLA-DRB5	?	ENSG00000198502	blood	5.32×10-6 (p-value)	GTEx Portal V6	4
PSMB9	ENST00000467593	ILMN_1798233	monocyte	2.30×10-5 (p-value)	Fairfax et al.	20
PSMB9	ENST00000464863		b-cell	1.37×10-6 (p-value)	Fairfax et al.	20
HLA-DPB1	ENST00000418931	ILMN_1749070	monocyte	2.88×10-5 (p-value)	Fairfax et al.	18
HLA-DPB1	ENST00000416804		b-cell	3.20×10-6 (p-value)	Fairfax et al.	18
HLA-DOB	ENST00000438763	ILMN_1700428	monocyte	9.14×10-5 (p-value)	Fairfax et al.	19
HLA-DOB	ENST00000488325					
HLA-DOB	ENST00000475235					
XXbac-BPG246D15.9	ENST00000452392					
C4A	?	ENSG00000244731	thyroid	5.52×10-12 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	thyroid	7.18×10-13 (p-value)	GTEx Portal V6	69
RNF5	?	ENSG00000204308	thyroid	4.39×10-7 (p-value)	GTEx Portal V6	58
HLA-DRB1	?	ENSG00000196126	thyroid	1.01×10-5 (p-value)	GTEx Portal V6	3
HLA-DMA	?	ENSG00000204257	thyroid	3.20×10-9 (p-value)	GTEx Portal V6	69
CYP21A1P	?	ENSG00000204338	thyroid	5.40×10-9 (p-value)	GTEx Portal V6	63
HLA-DRB6	?	ENSG00000229391	thyroid	3.59×10-6 (p-value)	GTEx Portal V6	10
C4A	?	ENSG00000244731	skeletal muscle	4.15×10-9 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	skeletal muscle	6.15×10-10 (p-value)	GTEx Portal V6	69
HLA-DRB6	?	ENSG00000229391	skeletal muscle	7.72×10-7 (p-value)	GTEx Portal V6	61
APOM	?	ENSG00000204444	skeletal muscle	4.71×10-6 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	transverse colon	7.43×10-10 (p-value)	GTEx Portal V6	63
HLA-DMA	?	ENSG00000204257	sun exposed skin	3.28×10-7 (p-value)	GTEx Portal V6	52
C4B	?	ENSG00000224389	sun exposed skin	4.54×10-10 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	sun exposed skin	1.31×10-8 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	sun exposed skin	2.17×10-6 (p-value)	GTEx Portal V6	12
CYP21A2	?	ENSG00000231852	sun exposed skin	2.20×10-9 (p-value)	GTEx Portal V6	62
HLA-DRB6	?	ENSG00000229391	sun exposed skin	1.86×10-6 (p-value)	GTEx Portal V6	25
HLA-DRB9	?	ENSG00000196301	sun exposed skin	3.11×10-6 (p-value)	GTEx Portal V6	7
C4A	?	ENSG00000244731	unexposed skin	1.89×10-11 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	left ventricle	2.32×10-14 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	left ventricle	2.45×10-7 (p-value)	GTEx Portal V6	61

CYP21A1P	?	ENSG00000204338	left ventricle	9.13×10-9 (p-value)	GTEx Portal V6	69
NOTCH4	?	ENSG00000204301	left ventricle	7.47×10-8 (p-value)	GTEx Portal V6	61
C4A	?	ENSG00000244731	aorta	2.96×10-8 (p-value)	GTEx Portal V6	67
HLA-DRB5	?	ENSG00000198502	aorta	7.96×10-7 (p-value)	GTEx Portal V6	15
CYP21A2	?	ENSG00000231852	aorta	8.94×10-7 (p-value)	GTEx Portal V6	48
MICB	?	ENSG00000204516	aorta	3.81×10-7 (p-value)	GTEx Portal V6	7
C4A	?	ENSG00000244731	subcutaneous adipocytes	1.06×10-10 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	subcutaneous adipocytes	2.11×10-11 (p-value)	GTEx Portal V6	69
CYP21A2	?	ENSG00000231852	subcutaneous adipocytes	1.64×10-6 (p-value)	GTEx Portal V6	17
NOTCH4	?	ENSG00000204301	subcutaneous adipocytes	3.67×10-6 (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	stomach	3.64×10-13 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	visceral adipocytes	5.84×10-9 (p-value)	GTEx Portal V6	67
HLA-DRB5	?	ENSG00000198502	visceral adipocytes	7.47×10-8 (p-value)	GTEx Portal V6	66
C4A	?	ENSG00000244731	tibial nerve	1.49×10-13 (p-value)	GTEx Portal V6	69
XXbac-BPG300A18.13	?	ENSG00000273333	tibial nerve	4.12×10-6 (p-value)	GTEx Portal V6	38
C4A	?	ENSG00000244731	esophagus mucosa	2.22×10-12 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	esophagus mucosa	7.42×10-9 (p-value)	GTEx Portal V6	66
LY6G6C	?	ENSG00000204421	esophagus mucosa	1.01×10-6 (p-value)	GTEx Portal V6	58
C6orf25	?	ENSG00000204420	esophagus mucosa	7.14×10-8 (p-value)	GTEx Portal V6	57
MICB	?	ENSG00000204516	esophagus mucosa	6.07×10-6 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	gastroesophageal junction	1.45×10-7 (p-value)	GTEx Portal V6	41
?	?	221491_x_at	blood	1.80×10-21 (p-value)	Dixon et al.	3
?	?	215666_at	blood	4.10×10-9 (p-value)	Dixon et al.	2
C4A	?	ENSG00000244731	testis	1.97×10-11 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	pancreas	6.53×10-7 (p-value)	GTEx Portal V6	45
C4A	?	ENSG00000244731	breast	1.35×10-8 (p-value)	GTEx Portal V6	62
C4A	?	ENSG00000244731	tibial artery	5.99×10-7 (p-value)	GTEx Portal V6	42
CYP21A1P	?	ENSG00000204338	tibial artery	5.54×10-7 (p-value)	GTEx Portal V6	46
HLA-DRB5	?	ENSG00000198502	tibial artery	4.29×10-7 (p-value)	GTEx Portal V6	6
HLA-DRB6	?	ENSG00000229391	tibial artery	7.53×10-6 (p-value)	GTEx Portal V6	4
C4A	?	ENSG00000244731	EBV lymphocytes	1.74×10-9 (p-value)	GTEx Portal V6	55
HLA-DRB5	?	ENSG00000198502	EBV lymphocytes	3.58×10-7 (p-value)	GTEx Portal V6	15

C4A	?	ENSG00000244731	liver	2.18×10-9 (p-value)	GTEx Portal V6	66
TNXA	?	ENSG00000248290	adrenal gland	1.46×10-8 (p-value)	GTEx Portal V6	67
HLA-DMA	?	ENSG00000204257	cerebellum	3.89×10-7 (p-value)	GTEx Portal V6	67
C4A	?	ENSG00000244731	cerebellum	1.80×10-7 (p-value)	GTEx Portal V6	50
?	?	ILMN_2159694	monocyte	3.60×10-21 (p-value)	Zeller et al.	9
?	?	ILMN_1893395	monocyte	2.44×10-20 (p-value)	Zeller et al.	9
?	?	ILMN_1752592	monocyte	8.41×10-62 (p-value)	Zeller et al.	9
HLA-DRA	ENST00000374982	ILMN_2157441	monocyte	2.49×10-20 (p-value)	Zeller et al.	9
HLA-DRA	ENST00000395388					
HLA-DRB6	ENST00000437183	ILMN_2066060	monocyte	1.15×10-33 (p-value)	Zeller et al.	9
HLA-DRB6	ENST00000411500					
HLA-DRB6	ENST00000437650					
PSMB9	ENST00000467593	ILMN_1798233	monocyte	1.82×10-12 (p-value)	Zeller et al.	1
PSMB9	ENST00000464863					
HLA-DOB	ENST00000438763	ILMN_1700428	monocyte	9.48×10-15 (p-value)	Zeller et al.	7
HLA-DOB	ENST00000475235					
HLA-DOB	ENST00000488325					
XXbac-BPG246D15.9	ENST00000452392					
?	?	ILMN_1715169	monocyte	4.42×10-44 (p-value)	Zeller et al.	9
HLA-DPB1	ENST00000418931	ILMN_1749070	monocyte	9.64×10-17 (p-value)	Zeller et al.	9
HLA-DPB1	ENST00000416804					
HLA-DRB5	ENST00000374975	ILMN_1697499	monocyte	3.78×10-17 (p-value)	Zeller et al.	9
?	?	ILMN_1883997	monocyte	2.12×10-26 (p-value)	Zeller et al.	9
LST1	ENST00000419073	ILMN_1717127	monocyte	1.49×10-13 (p-value)	Zeller et al.	2
LST1	ENST00000433492					
LST1	ENST00000376089					
LST1	ENST00000376090					
LST1	ENST00000211921					
HLA-DRB5	?	ENSG00000198502	brain	9.16×10-4 (q-value)	SeeQTL DB (Myers et al.)	7
C4A	?	ENSG00000244731	spleen	9.55×10-7 (p-value)	GTEx Portal V6	1
HLA-DRB5	?	ENSG00000198502	coronary artery	4.40×10-7 (p-value)	GTEx Portal V6	5
HLA-DRB5	?	ENSG00000198502	sigmoid colon	1.20×10-6 (p-value)	GTEx Portal V6	3

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
LIMS1	ENST00000410093	ILMN_1675387	chr2	monocyte	4.06×10-12 (p-value)	Zeller et al.	1
LIMS1	ENST00000542845						

LIMS1	ENST00000338045
LIMS1	ENST00000480744
LIMS1	ENST00000393310
LIMS1	ENST00000332345
LIMS1	ENST00000409441
LIMS1	ENST00000544547

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000739297	1	ENST00000374982, ENST00000395388	HLA-DRA

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445325	3	ENCP00000048170	FKBPL FKBPL FKBPL FKBPL
		ENCP00000048221	HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2
ENCE00000445329	1	ENCP00000048200	BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2
		ENCP00000048171	PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1
ENCE00000445291	1	ENCP00000048233	PSMB9 PSMB9 PSMB9 PSMB9 PSMB9 PSMB9 PSMB9 PSMB9
		ENCP00000048222	HLA-DOB HLA-DOB HLA-DOB HLA-DOB HLA-DOB HLA-DOB
		ENCP00000048159	CYP21A2 CYP21A2 C4B CYP21A2 C4B C4B

			CYP21A2 C4B C4B CYP21A2 C4B CYP21A2
ENCE00000445363	2	ENCP00000048231	TAP1 TAP1 TAP1 TAP1 TAP1 TAP1 TAP1 TAP1
		ENCP00000048185	PBX2 PBX2 PBX2 PBX2 PBX2 PBX2 PBX2
		ENCP00000048216	HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1
		ENCP00000048209	HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1
		ENCP00000048215	HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1
		ENCP00000048211	HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1
		ENCP00000048204	HLA-DRB5
		ENCP00000048187	PBX2 PBX2 PBX2 PBX2 PBX2 PBX2 PBX2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000025344 (CTCF binding site)	9	embryonic stem cell (H1ESC)	H3K4me2, Rad21, CTCF, H3K4me3, DNase1
		HSMMtube	H3K27me3, CTCF
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF, H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		nervous (NH-A)	CTCF, H3K4me2
		skin (NHEK)	CTCF

		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K4me2, CTCF
		A549	H3K27me3, CTCF
ENSR00001703124 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K4me2, Rad21, CTCF, H3K4me3, DNase1
		HSMMtube	H3K27me3, CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF, H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF, H3K4me2
		skin (NHEK)	CTCF
		NHLF	CTCF, H3K27me3
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K27me3
		endothelium (HUVEC)	H3K4me2, CTCF, H3K27me3
		A549	H3K27me3, CTCF
ENSR00000488005 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
ENSR00000488009 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me2, SP1
		HSMMtube	H3K27me3
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, Yy1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac, H3K36me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HLA-DRA	upstream gene variant, downstream gene variant	1044	ENST00000374982	?	ENSP00000364121	3
HLA-DRA	upstream gene variant, downstream gene variant	1008	ENST00000395388	NM_019111.4	ENSP00000378786	3
HLA-DRB9	downstream gene variant	1327	ENST00000449413	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HLA-DRA	ENST00000374982	?	ENSP00000364121	8
HLA-DRA	ENST00000395388	NM_019111.4	ENSP00000378786	8
HLA-DRB9	ENST00000449413	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
HLA-DRA	ENST00000374982	?	ENSP00000364121	1

HLA-DRA	ENST00000395388	NM_019111.4	ENSP00000378786	1

Locus 373 (rs854658)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,352,666-34,352,666
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.07	gene(s) hit or close-by	–
phastCons	0.16	eQTL gene(s)	CCL14, CCL15-CCL14, CCL23
GERP++	-2	potentially regulated gene(s)	–
CADD score	1.631	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CCL23	ENST00000613876	ILMN_1764030	skin	3.00×10^{-5} (p-value)	MuTHER consortium	1
CCL23	ENST00000615050					
CCL23	ENST00000613876	ILMN_1686109	skin	2.01×10^{-5} (p-value)	MuTHER consortium	1
CCL23	ENST00000615050		adipocyte	2.10×10^{-5} (p-value)	MuTHER consortium	1
CCL23	ENST00000612516					
CCL14	ENST00000622526	ILMN_1740609	adipocyte	6.68×10^{-8} (p-value)	MuTHER consortium	1
CCL15-CCL14	ENST00000616694					
CCL15-CCL14	ENST00000610751					
CCL14	ENST00000618404					
CCL14	ENST00000614009					
CCL14	ENST00000620991					

Locus 374 (rs12724450)

SNiPAcard

Block annotations

Block info

genomic range	chr1:150,273,976-150,514,747
block size	240,772 bp
variant count	69 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.253 [-4.164 – 2.641]	gene(s) hit or close-by	AC242988.1, ECM1, FALEC, MRPS21, PRPF3, RPRD2, TARS2
phastCons	μ = 0.096 [0 – 0.998]	eQTL gene(s)	C1orf54, CTSS, ECM1, FALEC, HORMAD1, RPRD2, TARS2
GERP++	μ = -0.311 [-5.83 – 4.94]	potentially regulated gene(s)	ADAMTSL4, ANP32E, CTSS, MCL1, PRPF3, RP11-458I7.1, RP11-54A4.2, TARS2, VPS45
CADD score	μ = 4.125 [0.007 – 23.2]	disease gene(s)	ECM1, ADAMTSL4, PRPF3, TARS2, VPS45

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ECM1	LIPOID PROTEINOSIS OF URBACH AND WIETHE	OMIM	MIM:247100
ADAMTSL4	ECTOPIA LENTIS ET PUPILLAE	OMIM	MIM:225200
PRPF3	RETINITIS PIGMENTOSA 18	OMIM	MIM:601414
TARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 21	OMIM	MIM:615918
VPS45	NEUTROPENIA, SEVERE CONGENITAL, 5, AUTOSOMAL RECESSIVE	OMIM	MIM:615285
ECM1	Lipoid proteinosis	OrphaNet	OrphaNet:530
ADAMTSL4	isolated ectopia lentis (EL)	OrphaNet	OrphaNet:1885
PRPF3	Retinitis pigmentosa	OrphaNet	OrphaNet:791
VPS45	Recurrent infections-myelofibrosis-nephromegaly syndrome	OrphaNet	OrphaNet:369852

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
MRPS21	missense variant	ENST00000581066	NM_018997.3	ENSP00000461930	Q/R	cAg/cGg	?	?	1
MRPS21	missense variant	ENST00000614145	NM_031901.5	ENSP00000480129	Q/R	cAg/cGg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HORMAD1	ENST00000361824	ILMN_1769849	skin	2.23×10^{-5} (p-value)	MuTHER consortium	7
HORMAD1	ENST00000470397					
HORMAD1	ENST00000486497					
HORMAD1	ENST00000322343					
HORMAD1	ENST00000368995					
C1orf54	?	ENSG00000118292	tibial nerve	1.93×10^{-7} (p-value)	GTEx Portal V6	47

FALEC	?	ENSG00000228126	tibial nerve	6.10×10-6 (p-value)	GTEx Portal V6	26
ECM1	?	ENSG00000143369	transformed fibroblasts	3.99×10-10 (p-value)	GTEx Portal V6	66
TARS2	?	ENSG00000143374	transformed fibroblasts	3.66×10-5 (p-value)	GTEx Portal V6	1
ECM1	?	ENSG00000143369	cerebellum	2.37×10-6 (p-value)	GTEx Portal V6	36
ECM1	?	ENSG00000143369	EBV lymphocytes	4.17×10-9 (p-value)	GTEx Portal V6	66
FALEC	?	ENSG00000228126	tibial artery	6.15×10-6 (p-value)	GTEx Portal V6	36
C1orf54	?	ENSG00000118292	tibial artery	8.11×10-6 (p-value)	GTEx Portal V6	3
HORMAD1	?	ENSG00000143452	esophagus mucosa	1.36×10-8 (p-value)	GTEx Portal V6	67
CTSS	?	ENSG00000163131	esophagus mucosa	1.01×10-5 (p-value)	GTEx Portal V6	4
ECM1	?	ENSG00000143369	sun exposed skin	2.08×10-6 (p-value)	GTEx Portal V6	48
FALEC	?	ENSG00000228126	aorta	4.10×10-6 (p-value)	GTEx Portal V6	27
ECM1	?	ENSG00000143369	thyroid	9.15×10-7 (p-value)	GTEx Portal V6	45
HORMAD1	?	ENSG00000143452	subcutaneous adipocytes	2.88×10-5 (p-value)	GTEx Portal V6	1
ECM1	?	ENSG00000143369	blood	1.15×10-6 (p-value)	GTEx Portal V6	23
?	?	ILMN_1702231	blood	3.16×10-5 (p-value)	Westra et al.	1
RPRD2	ENST00000401000	ILMN_3238889	b-cell	7.98×10-4 (p-value)	Fairfax et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000036275	1	ENCP00000004323	ADAMTSL4
		ENCP00000004313	PRPF3
		ENCP00000004312	PRPF3
ENCE00000036337	1	ENCP00000004327	MCL1
ENCE00000036132	1	ENCP00000004337	CTSS
		ENCP00000004311	PRPF3
		ENCP00000004291	VPS45
		ENCP00000004300	ANP32E
		ENCP00000004317	TARS2
		ENCP00000004293	VPS45
		ENCP00000004287	RP11-458I7.1
ENCE00000036329	1	ENCP00000004323	ADAMTSL4
ENCE00000036323	1	ENCP00000004325	RP11-54A4.2
		ENCP00000004324	ADAMTSL4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001524644 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		blood (K562)	PolII
		muscle (HSMM)	H3K27ac, DNase1

ENSR00000544743 (promoter)	1	embryonic stem cell (H1ESC)	TAF1, H3K4me2, H3K9ac, H3K4me3, H4K5ac, H3K36me3, CTCF, PolII, Jund, TAF7, H3K27ac, Yy1, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, DNase1
		blood (K562)	CTCFL, PolII, H3K4me2, DNase1, H3K4me3, TAF1, H2AZ, Yy1, H3K27ac, Max, PU1, H3K9ac, Cmyc, H3K79me2, HEY1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		liver (HepG2)	Yy1, PolII, H3K79me2, TAF1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4ac, H3K9ac, H4K5ac, H3K4me2, DNase1, H4K8ac, H3K18ac, H3K27ac
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, H3K79me2, TAF1, PU1, PolII, H2AZ, DNase1, Yy1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Nrf1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001586441 (enhancer)	1	cervix (HeLa-S3)	H3K79me2
		embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2
		blood (DND-41)	H3K36me3
		blood (K562)	H3K27me3, H3K79me2
		muscle (HSMM)	H3K79me2
ENSR00001524646 (enhancer)	1	liver (HepG2)	H3K79me2, H3K4me1, FOXA1
		blood (GM12878)	H3K79me2
		blood (DND-41)	H3K36me3
ENSR00000544755 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me1, FOXA1
		lung (IMR90)	DNase1, H3K36me3
		blood (K562)	Egr1
		A549	H3K36me3
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, DNase1
		breast (HMEC)	DNase1, H3K27ac, H3K4me2
ENSR00000079774 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII, Gabp, H3K9ac, Yy1, TAF1, H3K4me2, SP1, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H2AZ, H3K36me3, DNase1
		blood (K562)	H3K4me3, DNase1, H3K36me3, H3K27ac, Max, SP2, MEF2A, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, Cfos, PolII, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	H3K79me2, TAF1, ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K36me3, DNase1, Gabp, PolII
		lung (IMR90)	DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	ELF1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K9ac, H3K36me3, Gabp, Yy1, Pbx3, PolII, BCLAF1, MEF2A, H2AZ, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1, H3K27ac
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, Cfos, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00001524653 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K4me2, DNase1
		blood (K562)	H3K27ac, H3K9ac, Jund, PolII, H3K4me2, H3K36me3, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H3K79me2, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	PolII, H3K36me3

		lung (IMR90)	DNase1, H3K18ac, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac
		nervous (NH-A)	H3K9ac, H3K4me2, DNase1
		skin (NHEK)	H3K4me1, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	H3K4me3, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me1, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun, PolII, DNase1
		A549	H3K36me3
ENSR00000285718 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, Rad21, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me2, H2AZ, H3K36me3, DNase1
		blood (K562)	H3K27ac, H3K9ac, H2AZ, PolII, H3K4me2, CTCF, H3K4me1, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K36me3, H3K4me1, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K36me3, DNase1
		liver (HepG2)	PolII, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF, H3K4me3, DNase1
		lung (IMR90)	H3K4me3, H3K9ac, H3K36me3, H3K4me2, DNase1
		blood (GM12878)	Pbx3, PolII, H2AZ, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2
		skin (NHEK)	H3K9ac, H3K4me2, H3K36me3, DNase1
		NHLF	H3K36me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K4me1, H3K4me2, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	CTCF, DNase1, Nrf1, H3K9ac, Jund, H3K4me1, PolII
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27me3, DNase1
		endothelium (HUVEC)	H3K4me3, PolII, CTCF, DNase1, Max
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00001524662 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	DNase1, H3K27ac, H3K9ac
		blood (K562)	CTCF
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1
		cervix (HeLa-S3)	CTCF, DNase1
		liver (HepG2)	CTCF, H3K4me1
		lung (IMR90)	DNase1, H3K4me1, H3K18ac, H3K27ac, H4K5ac, H4K91ac
		nervous (NH-A)	DNase1
		skin (NHEK)	CTCF, DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PRPF3	3	ENST00000324862 ENST00000496202	hsa-miR-329-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC242988.1	downstream gene variant, upstream gene variant	835	ENST00000624245	?	ENSP00000485440	5
ECM1	upstream gene variant, downstream gene variant	976	ENST00000496744	?	?	2
ECM1	upstream gene variant, downstream gene variant	1207	ENST00000490346	?	?	3
ECM1	upstream gene variant, downstream gene variant	2448	ENST00000369049	NM_001202858.1	ENSP00000358045	2
ECM1	upstream gene variant, downstream gene variant	2213	ENST00000369047	NM_004425.3	ENSP00000358043	2
ECM1	upstream gene variant, downstream gene variant	382	ENST00000498579	?	?	3
ECM1	upstream gene variant, downstream gene variant	12	ENST00000346569	NM_022664.2	ENSP00000271630	3
ECM1	upstream gene variant, downstream gene variant	2449	ENST00000470432	?	?	2
FALEC	upstream gene variant, downstream gene variant	4229	ENST00000416894	?	?	2
MRPS21	downstream gene variant	1144	ENST00000614145	NM_031901.5	ENSP00000480129	1

MRPS21	downstream gene variant	1144	ENST00000581066	NM_018997.3	ENSP00000461930	1
PRPF3	upstream gene variant, downstream gene variant	1806	ENST00000324862	NM_004698.2	ENSP00000315379	3
PRPF3	upstream gene variant, downstream gene variant	225	ENST00000470824	?	?	7
PRPF3	upstream gene variant, downstream gene variant	967	ENST00000493553	?	?	7
PRPF3	upstream gene variant, downstream gene variant	1806	ENST00000467329	?	?	7
PRPF3	upstream gene variant, downstream gene variant	3321	ENST00000496202	?	?	3
PRPF3	upstream gene variant, downstream gene variant	3372	ENST00000467514	?	?	5
PRPF3	upstream gene variant	198	ENST00000476970	?	?	1
RPRD2	upstream gene variant, downstream gene variant	1444	ENST00000369067	?	ENSP00000358063	2
RPRD2	upstream gene variant, downstream gene variant	515	ENST00000401000	NM_001297674.1	ENSP00000383785	3
RPRD2	downstream gene variant	3525	ENST00000492220	?	?	2
RPRD2	upstream gene variant, downstream gene variant	1447	ENST00000369068	NM_015203.3	ENSP00000358064	3
TARS2	upstream gene variant	203	ENST00000479372	?	?	2
TARS2	upstream gene variant, downstream gene variant	178	ENST00000369064	NM_025150.4	ENSP00000358060	4
TARS2	upstream gene variant, downstream gene variant	175	ENST00000369054	NM_001271896.1	ENSP00000358050	4
TARS2	upstream gene variant, downstream gene variant	928	ENST00000463555	?	?	3
TARS2	upstream gene variant, downstream gene variant	450	ENST00000460794	?	?	2
TARS2	upstream gene variant, downstream gene variant	928	ENST00000480070	?	?	3
TARS2	upstream gene variant	752	ENST00000466989	?	?	2
TARS2	upstream gene variant, downstream gene variant	460	ENST00000462578	?	?	2
TARS2	upstream gene variant, downstream gene variant	631	ENST00000467982	?	ENSP00000475551	4
TARS2	upstream gene variant, downstream gene variant	181	ENST00000606933	NM_001271895.1	ENSP00000475847	4
TARS2	upstream gene variant, downstream gene variant	196	ENST00000369051	?	ENSP00000358047	4
TARS2	upstream gene variant	192	ENST00000369053	?	?	2
TARS2	downstream gene variant	1160	ENST00000483046	?	?	2
TARS2	upstream gene variant, downstream gene variant	125	ENST00000438568	?	ENSP00000415002	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ECM1	ENST00000369049	NM_001202858.1	ENSP00000358045	T	acA/acG	1
ECM1	ENST00000369047	NM_004425.3	ENSP00000358043	T	acA/acG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ECM1	ENST00000346569	NM_022664.2	ENSP00000271630	1
MRPS21	ENST00000581066	NM_018997.3	ENSP00000461930	4
MRPS21	ENST00000614145	NM_031901.5	ENSP00000480129	4
PRPF3	ENST00000324862	NM_004698.2	ENSP00000315379	15
PRPF3	ENST00000467329	?	?	10
PRPF3	ENST00000467514	?	?	3

PRPF3	ENST00000476970	?	?	4
PRPF3	ENST00000496202	?	?	8
RPRD2	ENST00000401000	NM_001297674.1	ENSP00000383785	27
RPRD2	ENST00000369068	NM_015203.3	ENSP00000358064	27
RPRD2	ENST00000369067	?	ENSP00000358063	19
RPRD2	ENST00000492220	?	?	27
TARS2	ENST00000606933	NM_001271895.1	ENSP00000475847	2
TARS2	ENST00000480070	?	?	1
TARS2	ENST00000483046	?	?	1
TARS2	ENST00000369064	NM_025150.4	ENSP00000358060	2
TARS2	ENST00000369051	?	ENSP00000358047	2
TARS2	ENST00000467982	?	ENSP00000475551	2
TARS2	ENST00000438568	?	ENSP00000415002	2
TARS2	ENST00000463555	?	?	1
TARS2	ENST00000369054	NM_001271896.1	ENSP00000358050	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ECM1	ENST00000369049	NM_001202858.1	ENSP00000358045	1
ECM1	ENST00000369047	NM_004425.3	ENSP00000358043	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ECM1	ENST00000496744	?	1
ECM1	ENST00000470432	?	2
ECM1	ENST00000498579	?	1
FALEC	ENST00000416894	?	1
RPRD2	ENST00000492220	?	1

Locus 375 (rs4473651)

SNiPAcard

Block annotations

Block info

genomic range	chr4:15,766,933-15,817,324
block size	50,392 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.995 [-3.896 – 0.774]	gene(s) hit or close-by	CD38
phastCons	μ = 0.026 [0 – 0.239]	eQTL gene(s)	CD38, FBXL5
GERP++	μ = -0.163 [-2.82 – 2.18]	potentially regulated gene(s)	RP11-442P12.1, RP11-442P12.2
CADD score	μ = 3.121 [0.076 – 13.93]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FBXL5	ENST00000511441	ILMN_2378100	blood	3.65×10-6 (p-value)	Westra et al.	4
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
CD38	?	ENSG00000004468	esophagus mucosa	1.41×10-6 (p-value)	GTEx Portal V6	10

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000374279	1	ENCP00000040482	RP11-442P12.1
		ENCP00000040479	RP11-442P12.2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CD38	upstream gene variant	895	ENST00000510674	?	ENSP00000423047	11

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CD38	ENST00000502843	?	ENSP00000427277	11
CD38	ENST00000511430	?	?	11
CD38	ENST00000506191	?	?	11
CD38	ENST00000226279	NM_001775.2	ENSP00000226279	11

Locus 376 (rs9869826)

SNiPAcard

Block annotations

Block info

genomic range	chr3:50,562,042-51,196,423
block size	634,382 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.302 [-2.21 – 0.611]	gene(s) hit or close-by	DOCK3, MAPKAPK3, ST13P14
phastCons	μ = 0.054 [0 – 0.294]	eQTL gene(s)	C3orf62, HYAL3, MAPKAPK3, NAT6, RPL29, USP4, WDR6
GERP++	μ = -0.348 [-8.17 – 2]	potentially regulated gene(s)	–
CADD score	μ = 4.951 [0.255 – 7.854]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HYAL3	?	ENSG00000186792	prostate	5.22×10^{-6} (p-value)	GTEx Portal V6	1
MAPKAPK3	ENST00000446044	ILMN_1732452	blood	9.73×10^{-5} (p-value)	Westra et al.	1
MAPKAPK3	ENST00000357955					
MAPKAPK3	ENST00000621469					
NAT6	?	ENSG00000243477	coronary artery	3.52×10^{-6} (p-value)	GTEx Portal V6	3
WDR6	ENST00000615452	ILMN_1669484	monocyte	2.08×10^{-6} (p-value)	Fairfax et al.	1
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	monocyte	2.75×10^{-7} (p-value)	Fairfax et al.	1
USP4	ENST00000483212					
C3orf62	ENST00000343010					
USP4	ENST00000485450					
USP4	ENST00000351842	ILMN_1798712	monocyte	1.00×10^{-17} (p-value)	Zeller et al.	1
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
RPL29	?	ENSG00000162244	liver	6.15×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363979 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac

		blood (K562)	H3K36me3
		blood (DND-41)	H4K20me1, H3K36me3
		skin (NHDF-AD)	H3K4me1, DNase1
		muscle (HSMM)	DNase1, H3K4me1, H3K27ac, H3K36me3
		breast (HMEC)	H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K36me3, DNase1
ENSR00001364012 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ST13P14	upstream gene variant	1868	ENST00000474095	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DOCK3	ENST00000266037	NM_004947.4	ENSP00000266037	9
MAPKAPK3	ENST00000457064	?	ENSP00000402045	1
MAPKAPK3	ENST00000446044	NM_001243926.1	ENSP00000396467	1
MAPKAPK3	ENST00000357955	?	ENSP00000350639	1
MAPKAPK3	ENST00000430409	?	ENSP00000410970	1
MAPKAPK3	ENST00000621469	NM_001243925.1, NM_004635.4	ENSP00000478922	1

Locus 377 (rs983545)

SNiPAcard

Block annotations

Block info

genomic range	chr3:16,947,451-16,972,211
block size	24,761 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.151 [-1.745 – 1.014]	gene(s) hit or close-by	MIR3714, PLCL2
phastCons	μ = 0.033 [0 – 0.422]	eQTL gene(s)	PLCL2
GERP++	μ = -1.075 [-7.38 – 3.22]	potentially regulated gene(s)	PLCL2
CADD score	μ = 3.650 [0.087 – 10.54]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Primary biliary cirrhosis	<2.00×10-8	GWAS Catalog	21399635	1
caproate (6:0)	8.64×10-6	Metabolomics GWAS Server	24816252	2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PLCL2	ENST00000432376	ILMN_1737025	adipocyte	3.87×10-8 (p-value)	MuTHER consortium	12
PLCL2	ENST00000615277					
PLCL2	?	ENSG00000154822	transformed fibroblasts	4.04×10-7 (p-value)	GTEx Portal V6	16
PLCL2	?	ENSG00000154822	blood	2.38×10-10 (p-value)	GTEx Portal V6	17
PLCL2	ENST00000615277	ILMN_1737025	monocyte	4.25×10-9 (p-value)	Fairfax et al.	2
PLCL2	ENST00000432376		b-cell	2.47×10-4 (p-value)	Fairfax et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000331323	1	ENCP00000035615	PLCL2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001673018 (enhancer)	2	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		blood (GM12878)	H3K79me2
		blood (K562)	H3K27me3
ENSR00001673019 (promoter flanking region)	6	monocytes (Monocytes-CD14+)	H4K20me1
		endothelium (HUVEC)	PolII
		blood (GM12878)	H3K79me2

		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1
ENSR00001477059 e! (CTCF binding site)	4	monocytes (Monocytes-CD14+)	H4K20me1
		Osteobl	H3K4me2
		blood (GM12878)	H3K79me2
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MIR3714 e!	upstream gene variant	2477	ENST00000579507 e!	?	?	1
PLCL2 e!	upstream gene variant	2371	ENST00000432376 e!	NM_015184.5	ENSP00000412836 e!	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLCL2 e!	ENST00000460467 e!	?	?	18
PLCL2 e!	ENST00000615277 e!	NM_001144382.1	ENSP00000478458 e!	18

Locus 378 (rs3798911)

SNiPAcard

Block annotations

Block info

genomic range	chr6:161,410,911-161,466,180
block size	55,270 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.059 [-0.379 – 0.364]	gene(s) hit or close-by	MAP3K4, RP3-428L16.2
phastCons	μ = 0.099 [0 – 0.351]	eQTL gene(s)	MAP3K4
GERP++	μ = 0.739 [-0.629 – 2.91]	potentially regulated gene(s)	–
CADD score	μ = 5.009 [1.938 – 8.622]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MAP3K4	ENST00000366919	ILMN_1678087	blood	2.80×10^{-5} (p-value)	Westra et al.	4
MAP3K4	ENST00000348824					
MAP3K4	ENST00000544041					
MAP3K4	ENST00000490904					
MAP3K4	ENST00000392142					
MAP3K4	ENST00000366920					
MAP3K4	ENST00000536852					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MAP3K4	upstream gene variant	903	ENST00000544041	?	ENSP00000445018	2
MAP3K4	upstream gene variant	903	ENST00000490904	NM_001291958.1	ENSP00000446303	2
MAP3K4	upstream gene variant	828	ENST00000542952	?	ENSP00000439377	2
MAP3K4	upstream gene variant	873	ENST00000366920	NM_001301072.1	ENSP00000355887	2
MAP3K4	upstream gene variant	786	ENST00000366919	NM_006724.2	ENSP00000355886	2
MAP3K4	upstream gene variant	873	ENST00000348824	?	ENSP00000297332	2
MAP3K4	upstream gene variant	843	ENST00000392142	NM_005922.2	ENSP00000375986	2
RP3-428L16.2	downstream gene variant, upstream gene variant	439	ENST00000608721	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MAP3K4	ENST00000348824	?	ENSP00000297332	5
MAP3K4	ENST00000366919	NM_006724.2	ENSP00000355886	5
MAP3K4	ENST00000542952	?	ENSP00000439377	5

MAP3K4	ENST00000544041	?	ENSP00000445018	5
MAP3K4	ENST00000490904	NM_001291958.1	ENSP00000446303	5
MAP3K4	ENST00000392142	NM_005922.2	ENSP00000375986	5
MAP3K4	ENST00000366920	NM_001301072.1	ENSP00000355887	5
MAP3K4	ENST00000446500	?	?	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP3-428L16.2	ENST00000608721	?	1

Locus 379 (rs204888)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,021,852-32,097,876
block size	76,025 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.132 [-1.693 – 1.09]	gene(s) hit or close-by	ATF6B, FKBPL, TNXB
phastCons	μ = 0.167 [0 – 1]	eQTL gene(s)	ABCF1, EHMT2, LST1, SKIV2L, TNXA
GERP++	μ = -1.180 [-5.78 – 4.69]	potentially regulated gene(s)	CFB, CFB, CFB, CFB, CFB, CFB, CFB, CFB, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, TNXB, TNXB, TNXB, TNXB, TNXB, TNXB, TNXB
CADD score	μ = 10.688 [1.389 – 21.6]	disease gene(s)	SKIV2L, , , , , , CFB, TNXB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)

SKIV2L	?	ENSG00000204351	muscularis mucosae	1.17×10^{-11} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	lung	9.55×10^{-11} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	sigmoid colon	6.17×10^{-10} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	blood	2.43×10^{-8} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	tibial artery	3.15×10^{-11} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	esophagus mucosa	1.32×10^{-8} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	aorta	4.81×10^{-8} (p-value)	GTEx Portal V6	4
TNXA	?	ENSG00000248290	subcutaneous adipocytes	6.09×10^{-7} (p-value)	GTEx Portal V6	6
SKIV2L	?	ENSG00000204351	atrial appendage	3.10×10^{-8} (p-value)	GTEx Portal V6	5
SKIV2L	?	ENSG00000204351	tibial nerve	1.85×10^{-7} (p-value)	GTEx Portal V6	2
SKIV2L	?	ENSG00000204351	thyroid	5.06×10^{-9} (p-value)	GTEx Portal V6	5
SKIV2L	?	ENSG00000204351	cerebellar hemisphere	1.71×10^{-8} (p-value)	GTEx Portal V6	5
SKIV2L	?	ENSG00000204351	transverse colon	3.72×10^{-7} (p-value)	GTEx Portal V6	1
SKIV2L	?	ENSG00000204351	unexposed skin	2.03×10^{-7} (p-value)	GTEx Portal V6	2
EHMT2	ENST00000375537	ILMN_1751081	blood	1.80×10^{-5} (p-value)	MuTHER consortium	4
EHMT2	ENST00000375528					
EHMT2	ENST00000461880					
EHMT2	ENST00000478491					
EHMT2	ENST00000494816					
EHMT2	ENST00000480912					
EHMT2	ENST00000395728					
EHMT2	ENST00000375530					
LST1	ENST00000419073	ILMN_1717127	skin	1.73×10^{-5} (p-value)	MuTHER consortium	4
LST1	ENST00000433492					
LST1	ENST00000376089					
LST1	ENST00000376090					
LST1	ENST00000211921					
ABCF1	ENST00000326195	ILMN_1763875	b-cell	2.59×10^{-4} (p-value)	Fairfax et al.	4
ABCF1	ENST00000376545					
ABCF1	ENST00000479542					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
E2F4	MA0541.1	6	yes	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445099	1	ENCP00000048143	CFB CFB CFB CFB CFB

	CFB
	CFB
	CFB
ENCP00000048138	CFB
	CFB
	CFB
	CFB
	CFB
	CFB
	CFB
ENCP00000048087	DDAH2
	DDAH2
	DDAH2
	DDAH2
	DDAH2
	DDAH2
	DDAH2
ENCP00000048049	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
	PRRC2A
ENCP00000048163	TNXB
	TNXB
	TNXB
	TNXB
	TNXB
	TNXB
	TNXB
	TNXB

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001703098 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00000264509 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, TAF1, H3K4me2, SP1, Srf, USF1, H3K9ac, H3K4me3, Yy1, H3K27ac, H3K36me3, CTCF, PolII, Gabp, Sin3Ak20
		HSMMtube	H3K4me3, H3K4me2, H3K9ac, H2AZ, DNase1
		blood (K562)	Max, Srf, H3K9ac, Gabp, USF1, H3K79me2, H2AZ, ELF1, Nrsf, Cfos, PolII, H3K4me2, DNase1, H3K4me3, Egr1, H3K27ac, ZBTB33
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	DNase1, ZBTB33, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, H2AZ, H3K4me1, Gabp, Yy1, Srf, PolII, TAF1, ELF1, USF1
		blood (GM12878)	Tr4, SIX5, PolII, Srf, H2AZ, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, ZBTB33, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	H3K9ac, DNase1, H3K18ac, H3K27ac, H3K4me2, H3K36me3, H3K4me3
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K4me1, H3K4me2, H3K27ac
		breast (HMEC)	H3K4me3, H3K9ac, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, Gabp, PolII, Nrf1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, Cjun, Max, H3K4me3, H3K4me2
		A549	H3K36me3, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ATF6B	downstream gene variant, upstream gene variant	1859	ENST00000375203	NM_004381.4	ENSP00000364349	2
ATF6B	upstream gene variant	1346	ENST00000475705	?	?	1

ATF6B	upstream gene variant	945	ENST00000492342	?	?	1
ATF6B	downstream gene variant, upstream gene variant	1846	ENST00000375201	NM_001136153.1	ENSP00000364347	2
ATF6B	upstream gene variant	3755	ENST00000494022	?	?	1
ATF6B	downstream gene variant, upstream gene variant	3474	ENST00000453203	?	ENSP00000393419	2
ATF6B	upstream gene variant	1877	ENST00000495579	?	?	1
ATF6B	upstream gene variant	2332	ENST00000485314	?	?	1
ATF6B	downstream gene variant, upstream gene variant	1881	ENST00000468502	?	?	2
TNXB	upstream gene variant	290	ENST00000479795	?	ENSP00000418248	2
TNXB	upstream gene variant	382	ENST00000486148	?	?	2
TNXB	upstream gene variant	229	ENST00000375244	NM_019105.6	ENSP00000364393	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATF6B	ENST00000494022	?	?	3
ATF6B	ENST00000375203	NM_004381.4	ENSP00000364349	1
ATF6B	ENST00000485314	?	?	1
ATF6B	ENST00000375201	NM_001136153.1	ENSP00000364347	1
ATF6B	ENST00000495579	?	?	1
TNXB	ENST00000442721	?	ENSP00000389946	3
TNXB	ENST00000479795	?	ENSP00000418248	1
TNXB	ENST00000486148	?	?	1
TNXB	ENST00000375244	NM_019105.6	ENSP00000364393	2
TNXB	ENST00000611016	?	ENSP00000483409	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FKBPL	ENST00000375156	NM_022110.3	ENSP00000364298	1

Locus 381 (rs17660992)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,163,948-52,165,394
block size	1,447 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.183 [-0.401 – 0.036]	**gene(s) hit or close-by**	–
phastCons	μ = 0.000 [0 – 0]	**eQTL gene(s)**	–
GERP++	μ = -0.621 [-1.34 – 0.0977]	**potentially regulated gene(s)**	–
CADD score	μ = 6.307 [4.247 – 8.367]	**disease gene(s)**	–

Locus 382 (rs1110258)

SNiPAcard

Block annotations

Block info

genomic range	chr4:15,742,716-15,752,676
block size	9,961 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.026 [-0.295 – 0.242]	**gene(s) hit or close-by**	BST1, RP11-442P12.2
phastCons	μ = 0.119 [0 – 0.239]	**eQTL gene(s)**	–
GERP++	μ = -0.134 [-1.02 – 0.753]	**potentially regulated gene(s)**	–
CADD score	μ = 4.558 [2.04 – 7.076]	**disease gene(s)**	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BST1	downstream gene variant	2785	ENST00000514445	?	ENSP00000420925	1
BST1	downstream gene variant	2780	ENST00000514989	?	ENSP00000424761	1
RP11-442P12.2	upstream gene variant	2103	ENST00000513715	?	?	1

Locus 383 (rs229100)

SNiPAcard

Block annotations

Block info

genomic range	chr21:28,221,167-28,224,836
block size	3,670 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.097 [-2.218 – -0.069]	gene(s) hit or close-by	ADAMTS1
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	GABPA
GERP++	μ = -0.096 [-2.9 – 3]	potentially regulated gene(s)	–
CADD score	μ = 2.243 [1.488 – 3.601]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GABPA	ENST00000400075	ILMN_2175562	monocyte	1.26×10-4 (p-value)	Fairfax et al.	1
GABPA	ENST00000354828					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001668110 (enhancer)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	Rad21
		Osteobl	CTCF
		lung (IMR90)	CTCF
		blood (K562)	CTCF
		blood (DND-41)	CTCF
		muscle (HSMM)	CTCF
ENSR00000612272 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ADAMTS1	upstream gene variant	3439	ENST00000284984	NM_006988.3	ENSP00000284984	1

Locus 384 (rs41265665)

SNiPAcard

Block annotations

Block info

genomic range	chr4:74,331,266-74,483,608
block size	152,343 bp
variant count	52 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.421 [-3.218 – 1.469]	gene(s) hit or close-by	AFM, RASSF6, RP11-622A1.1, RP11-622A1.2
phastCons	μ = 0.062 [0 – 0.979]	eQTL gene(s)	RASSF6
GERP++	μ = -0.510 [-6.47 – 3.2]	potentially regulated gene(s)	AFM, AFP
CADD score	μ = 3.743 [0.044 – 14.92]	disease gene(s)	AFP

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
AFP	ALPHA-FETOPROTEIN, HEREDITARY PERSISTENCE OF	OMIM	MIM:615970
AFP	ALPHA-FETOPROTEIN DEFICIENCY	OMIM	MIM:615969
AFP	Congenital deficiency in alpha-fetoprotein	OrphaNet	OrphaNet:168612
AFP	Hereditary persistence of alpha-fetoprotein	OrphaNet	OrphaNet:168615

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
AFM	missense variant	ENST00000226355	NM_001133.2	ENSP00000226355	H/R	cAt/cGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RASSF6	ENST00000395777	ILMN_1657381	b-cell	5.47×10^{-18} (p-value)	Fairfax et al.	1
RASSF6	ENST00000307439					
RASSF6	ENST00000512591					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000041356		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000382154	1	ENCP00000041353	AFP

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001242774 (enhancer)	1	skin (NHEK)	H3K36me3
ENSR00001432007 (promoter flanking region)	6	embryonic stem cell (H1ESC)	DNase1
		blood (DND-41)	H3K4me1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K27ac, H3K4me1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		blood (GM12878)	Cjun, H3K4me3, PU1, PolII, BATF, DNase1, IRF4, BCL11A, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K4me2
ENSR00001685745 (CTCF binding site)	4	embryonic stem cell (H1ESC)	DNase1
		blood (DND-41)	H3K4me1
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K27ac, H3K4me1
		muscle (HSMM)	DNase1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		blood (GM12878)	PU1, PolII, BATF, DNase1, IRF4, BCL11A, H3K27ac, H3K4me2, Cjun
		skin (NHEK)	H3K4me1, H3K27ac, H3K4me2
ENSR00001685748 (enhancer)	1	A549	H3K36me3
		breast (HMEC)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AFM	upstream gene variant, downstream gene variant	219	ENST00000505794	?	?	5
AFM	upstream gene variant, downstream gene variant	184	ENST00000226355	NM_001133.2	ENSP00000226355	10
RP11-622A1.1	upstream gene variant, downstream gene variant	3438	ENST00000511117	?	?	3
RP11-622A1.2	upstream gene variant, downstream gene variant	1361	ENST00000508147	?	?	6
RP11-622A1.2	upstream gene variant, downstream gene variant	1326	ENST00000504368	?	?	6
RP11-622A1.2	upstream gene variant, downstream gene variant	1562	ENST00000502291	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AFM	ENST00000505794	?	?	3
AFM	ENST00000226355	NM_001133.2	ENSP00000226355	10
RASSF6	ENST00000307439	NM_177532.4	ENSP00000303877	15
RASSF6	ENST00000342081	NM_201431.2	ENSP00000340578	15
RASSF6	ENST00000335049	NM_001270392.1	ENSP00000335582	15
RASSF6	ENST00000512591	?	?	1
RASSF6	ENST00000395777	NM_001270391.1	ENSP00000379123	15
RP11-622A1.2	ENST00000508147	?	?	8
RP11-622A1.2	ENST00000502291	?	?	8
RP11-622A1.2	ENST00000504368	?	?	9

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AFM	ENST00000505794	?	2
RP11-622A1.2	ENST00000502291	?	1

Locus 385 (rs3745542)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,587,288-51,587,893
block size	606 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -2.387 [-3.363 – -1.015]	gene(s) hit or close-by	KLK14
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	–
GERP++	μ = -0.833 [-2.23 – 1.69]	potentially regulated gene(s)	–
CADD score	μ = 4.350 [0.518 – 7.659]	disease gene(s)	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348484 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		A549	H3K27me3
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
KLK14	upstream gene variant	141	ENST00000156499	?	ENSP00000156499	2
KLK14	upstream gene variant	141	ENST00000391802	NM_022046.4	ENSP00000375678	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLK14	ENST00000156499	?	ENSP00000156499	1
KLK14	ENST00000391802	NM_022046.4	ENSP00000375678	1

Locus 386 (rs9646944)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,858,921-102,875,540
block size	16,620 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.169 [-0.796 – 1.086]	gene(s) hit or close-by	FAM183DP, IL1RL2
phastCons	μ = 0.010 [0 – 0.027]	eQTL gene(s)	AC007278.3, IL18R1, IL1RL1
GERP++	μ = 0.493 [-0.539 – 1.48]	potentially regulated gene(s)	–
CADD score	μ = 3.754 [1.939 – 6.629]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL1RL1	?	ENSG00000115602	lung	2.92×10^{-6} (p-value)	GTEx Portal V6	5
AC007278.3	?	ENSG00000234389	lung	1.49×10^{-5} (p-value)	GTEx Portal V6	1
AC007278.3	?	ENSG00000234389	blood	1.87×10^{-8} (p-value)	GTEx Portal V6	5
IL18R1	?	ENSG00000115604	cerebellum	8.77×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001544945 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K27me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		liver (HepG2)	H3K27me3, DNase1
		lung (IMR90)	DNase1, H2BK20ac
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FAM183DP	downstream gene variant	442	ENST00000421987	?	?	2
IL1RL2	downstream gene variant	2459	ENST00000264257	NM_003854.2	ENSP00000264257	2
IL1RL2	downstream gene variant	3110	ENST00000441515	?	ENSP00000413348	2
IL1RL2	downstream gene variant	3123	ENST00000481806	?	?	2

Locus 387 (rs2853928)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,242,089-31,328,542
block size	86,454 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.879 [-3.321 – 0.343]	**gene(s) hit or close-by**	HLA-B, HLA-C, MIR6891, RPL3P2, USP8P1, WASF5P, XXbac-BPG248L24.10, XXbac-BPG248L24.12, XXbac-BPG248L24.13
phastCons	μ = 0.045 [0 – 0.398]	**eQTL gene(s)**	ATP6V1G2, BAG6, C4B, CCHCR1, CD248, CSF3R, CYP21A1P, DDX39B, FLOT1, HCG22, HCG27, HLA-B, HLA-C, LINC00243, MICA, MICB, PSORS1C1, PSORS1C2, PSORS1C3, TCF19, TMEM154, TNXB, TSSC4, VARS2, WASF5P, XRCC6, XXbac-BPG181B23.7, XXbac-BPG248L24.12, XXbac-BPG248L24.13, XXbac-BPG299F13.17
GERP++	μ = -0.521 [-3.27 – 0.497]	**potentially regulated gene(s)**	–
CADD score	μ = 2.610 [0.13 – 11.44]	**disease gene(s)**	VARS2, CSF3R, C4B, HLA-B, TNXB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Height	$<4.00\times10^{-12}$	GWAS Catalog	23563607	1
Triglycerides	$<2.00\times10^{-15}$	GWAS Catalog	20686565	1
CD4:CD8 lymphocyte ratio	$<2.00\times10^{-28}$	GWAS Catalog	20045101	1
Blood pressure	7.09×10^{-5}	dbGaP	pha003050	2
Lupus erythematosus, systemic	5.16×10^{-10}	dbGaP	pha002848	1

Disease gene annotation

gene	trait	source DB	source entry/link	
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
CSF3R	NEUTROPHILIA, HEREDITARY	OMIM	MIM:162830	OMIM
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
CSF3R	Chronic neutrophilic leukemia	OrphaNet	OrphaNet:86829	orphanet
CSF3R	Hereditary neutrophilia	OrphaNet	OrphaNet:279943	orphanet
CSF3R	Atypical chronic myeloid leukemia	OrphaNet	OrphaNet:98824	orphanet
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839	orphanet
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HLA-C	?	ENSG00000204525	atrial appendage	4.49×10-32 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	atrial appendage	6.95×10-23 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	atrial appendage	3.34×10-7 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	atrial appendage	2.37×10-10 (p-value)	GTEx Portal V6	15
MICA	?	ENSG00000204520	atrial appendage	2.44×10-6 (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	transformed fibroblasts	1.05×10-14 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	transformed fibroblasts	2.71×10-10 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	transformed fibroblasts	2.18×10-71 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	3.70×10-43 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	transformed fibroblasts	4.10×10-7 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	transformed fibroblasts	4.97×10-12 (p-value)	GTEx Portal V6	16
MICA	?	ENSG00000204520	transformed fibroblasts	2.05×10-8 (p-value)	GTEx Portal V6	15
C4B	?	ENSG00000224389	transformed fibroblasts	4.47×10-6 (p-value)	GTEx Portal V6	3
HCG27	?	ENSG00000206344	blood	1.91×10-9 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	blood	1.42×10-53 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	blood	3.70×10-12 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	blood	3.93×10-60 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	blood	8.36×10-16 (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	blood	7.20×10-9 (p-value)	GTEx Portal V6	16
LINC00243	?	ENSG00000214894	blood	7.76×10-8 (p-value)	GTEx Portal V6	16
XXbac-BPG181B23.7	?	ENSG00000272221	blood	7.99×10-7 (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	breast	7.69×10-8 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	breast	2.03×10-37 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	breast	5.30×10-7 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	breast	4.50×10-31 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	breast	2.78×10-14 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	breast	9.37×10-10 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	breast	7.76×10-7 (p-value)	GTEx Portal V6	14
WASF5P	?	ENSG00000231402	breast	1.35×10-6 (p-value)	GTEx Portal V6	8
MICA	?	ENSG00000204520	breast	1.42×10-6 (p-value)	GTEx Portal V6	3
HCG27	?	ENSG00000206344	skeletal muscle	6.99×10-11 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	skeletal muscle	2.75×10-95 (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	skeletal muscle	1.88×10-6 (p-value)	GTEx Portal V6	11
HLA-B	?	ENSG00000234745	skeletal muscle	1.10×10-22 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	4.19×10-64 (p-value)	GTEx Portal V6	16
MICA	?	ENSG00000204520	skeletal muscle	8.21×10-8 (p-value)	GTEx Portal V6	15
PSORS1C1	?	ENSG00000204540	skeletal muscle	9.28×10-7 (p-value)	GTEx Portal V6	15

XXbac-BPG299F13.17	?	ENSG00000272501	skeletal muscle	5.71×10^{-6} (p-value)	GTEx Portal V6	6
HLA-C	?	ENSG00000204525	putamen	1.32×10^{-15} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	putamen	5.13×10^{-16} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	EBV lymphocytes	6.62×10^{-27} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	EBV lymphocytes	2.69×10^{-16} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	EBV lymphocytes	9.14×10^{-18} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	ovary	5.49×10^{-23} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	ovary	2.74×10^{-17} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	ovary	5.36×10^{-11} (p-value)	GTEx Portal V6	15
HCG27	?	ENSG00000206344	sun exposed skin	8.59×10^{-9} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	sun exposed skin	5.17×10^{-72} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	sun exposed skin	1.35×10^{-7} (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	3.48×10^{-51} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	sun exposed skin	1.54×10^{-12} (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	sun exposed skin	2.79×10^{-8} (p-value)	GTEx Portal V6	14
PSORS1C1	?	ENSG00000204540	sun exposed skin	1.53×10^{-16} (p-value)	GTEx Portal V6	16
FLOT1	?	ENSG00000137312	sun exposed skin	3.51×10^{-6} (p-value)	GTEx Portal V6	10
CYP21A1P	?	ENSG00000204338	sun exposed skin	6.43×10^{-7} (p-value)	GTEx Portal V6	14
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	2.10×10^{-6} (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	unexposed skin	1.61×10^{-6} (p-value)	GTEx Portal V6	15
HLA-C	?	ENSG00000204525	unexposed skin	1.31×10^{-43} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	unexposed skin	7.21×10^{-31} (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	unexposed skin	1.05×10^{-9} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	unexposed skin	1.88×10^{-7} (p-value)	GTEx Portal V6	4
HLA-C	?	ENSG00000204525	aorta	3.92×10^{-49} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	aorta	4.29×10^{-19} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	aorta	4.19×10^{-35} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	aorta	2.91×10^{-8} (p-value)	GTEx Portal V6	11
PSORS1C1	?	ENSG00000204540	aorta	6.04×10^{-8} (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	aorta	1.44×10^{-6} (p-value)	GTEx Portal V6	10
MICB	?	ENSG00000204516	aorta	4.90×10^{-6} (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	left ventricle	2.74×10^{-41} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	1.66×10^{-35} (p-value)	GTEx Portal V6	16
HCG27	?	ENSG00000206344	left ventricle	1.89×10^{-6} (p-value)	GTEx Portal V6	10
MICA	?	ENSG00000204520	left ventricle	1.82×10^{-7} (p-value)	GTEx Portal V6	10
HLA-B	?	ENSG00000234745	left ventricle	3.15×10^{-10} (p-value)	GTEx Portal V6	15
PSORS1C1	?	ENSG00000204540	left ventricle	3.33×10^{-7} (p-value)	GTEx Portal V6	15

HCG27	?	ENSG00000206344	subcutaneous adipocytes	2.06×10^{-13} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	7.57×10^{-62} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	1.68×10^{-49} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	subcutaneous adipocytes	2.89×10^{-17} (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	subcutaneous adipocytes	8.99×10^{-8} (p-value)	GTEx Portal V6	11
CYP21A1P	?	ENSG00000204338	subcutaneous adipocytes	8.32×10^{-8} (p-value)	GTEx Portal V6	11
VARS2	?	ENSG00000137411	subcutaneous adipocytes	7.43×10^{-7} (p-value)	GTEx Portal V6	11
PSORS1C1	?	ENSG00000204540	subcutaneous adipocytes	1.13×10^{-15} (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	subcutaneous adipocytes	8.97×10^{-8} (p-value)	GTEx Portal V6	11
XXbac-BPG299F13.17	?	ENSG00000272501	subcutaneous adipocytes	6.48×10^{-6} (p-value)	GTEx Portal V6	5
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	3.86×10^{-8} (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	caudate basal ganglia	2.41×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	caudate basal ganglia	1.68×10^{-17} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	caudate basal ganglia	1.80×10^{-18} (p-value)	GTEx Portal V6	16
XXbac-BPG181B23.7	?	ENSG00000272221	caudate basal ganglia	1.86×10^{-7} (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	visceral adipocytes	4.81×10^{-35} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	visceral adipocytes	1.64×10^{-12} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	6.79×10^{-32} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	visceral adipocytes	3.36×10^{-6} (p-value)	GTEx Portal V6	1
PSORS1C1	?	ENSG00000204540	visceral adipocytes	3.31×10^{-10} (p-value)	GTEx Portal V6	16
HCG27	?	ENSG00000206344	stomach	5.60×10^{-8} (p-value)	GTEx Portal V6	14
PSORS1C2	?	ENSG00000204538	stomach	2.18×10^{-12} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	stomach	1.59×10^{-38} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	stomach	3.57×10^{-29} (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	stomach	7.74×10^{-14} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	stomach	6.50×10^{-10} (p-value)	GTEx Portal V6	15
HLA-C	?	ENSG00000204525	cortex	4.63×10^{-21} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	cortex	3.42×10^{-18} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	cortex	9.09×10^{-9} (p-value)	GTEx Portal V6	11
HCG27	?	ENSG00000206344	tibial nerve	8.11×10^{-11} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	tibial nerve	1.32×10^{-50} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	tibial nerve	2.81×10^{-8} (p-value)	GTEx Portal V6	14
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	4.56×10^{-42} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	tibial nerve	5.62×10^{-14} (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	tibial nerve	5.37×10^{-9} (p-value)	GTEx Portal V6	14
PSORS1C1	?	ENSG00000204540	tibial nerve	4.02×10^{-10} (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	tibial nerve	6.17×10^{-7} (p-value)	GTEx Portal V6	11

PSORS1C2	?	ENSG00000204538	tibial nerve	1.26×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	1.33×10-5 (p-value)	GTEx Portal V6	1
PSORS1C3	?	ENSG00000204528	esophagus mucosa	3.65×10-6 (p-value)	GTEx Portal V6	7
PSORS1C2	?	ENSG00000204538	esophagus mucosa	1.75×10-13 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	esophagus mucosa	5.36×10-50 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	esophagus mucosa	3.33×10-6 (p-value)	GTEx Portal V6	1
TCF19	?	ENSG00000137310	esophagus mucosa	1.35×10-6 (p-value)	GTEx Portal V6	12
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	1.80×10-39 (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	esophagus mucosa	5.73×10-7 (p-value)	GTEx Portal V6	12
PSORS1C1	?	ENSG00000204540	esophagus mucosa	7.80×10-16 (p-value)	GTEx Portal V6	16
HCG27	?	ENSG00000206344	esophagus mucosa	1.09×10-7 (p-value)	GTEx Portal V6	15
MICA	?	ENSG00000204520	esophagus mucosa	6.28×10-6 (p-value)	GTEx Portal V6	3
HLA-C	?	ENSG00000204525	anterior cingulate cortex	4.70×10-17 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	anterior cingulate cortex	5.96×10-11 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	pituitary	2.35×10-14 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	pituitary	1.06×10-14 (p-value)	GTEx Portal V6	16
WASF5P	?	ENSG00000231402	pituitary	3.26×10-7 (p-value)	GTEx Portal V6	10
HLA-C	?	ENSG00000204525	testis	6.15×10-28 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	testis	6.59×10-24 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	testis	1.24×10-26 (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	testis	9.54×10-8 (p-value)	GTEx Portal V6	15
HLA-C	?	ENSG00000204525	hypothalamus	5.02×10-17 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	hypothalamus	2.34×10-14 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	frontal cortex	1.91×10-19 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	frontal cortex	3.00×10-16 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	frontal cortex	2.36×10-7 (p-value)	GTEx Portal V6	11
HLA-C	?	ENSG00000204525	uterus	2.63×10-16 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	uterus	1.79×10-13 (p-value)	GTEx Portal V6	16
WASF5P	?	ENSG00000231402	pancreas	4.24×10-10 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	pancreas	4.82×10-36 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	pancreas	7.69×10-10 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	pancreas	6.15×10-24 (p-value)	GTEx Portal V6	16
DDX39B	?	ENSG00000198563	pancreas	2.13×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.13	?	ENSG00000256166	pancreas	3.94×10-7 (p-value)	GTEx Portal V6	10
HCG27	?	ENSG00000206344	muscularis mucosae	1.75×10-8 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	muscularis mucosae	1.68×10-7 (p-value)	GTEx Portal V6	11
HLA-C	?	ENSG00000204525	muscularis mucosae	9.42×10-54 (p-value)	GTEx Portal V6	16

XXbac-BPG299F13.17	?	ENSG00000272501	muscularis mucosae	1.02×10-10 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	muscularis mucosae	9.39×10-38 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	muscularis mucosae	5.76×10-17 (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	muscularis mucosae	1.45×10-8 (p-value)	GTEx Portal V6	11
PSORS1C1	?	ENSG00000204540	muscularis mucosae	2.49×10-10 (p-value)	GTEx Portal V6	13
CYP21A1P	?	ENSG00000204338	muscularis mucosae	1.18×10-6 (p-value)	GTEx Portal V6	10
XXbac-BPG181B23.7	?	ENSG00000272221	muscularis mucosae	3.79×10-6 (p-value)	GTEx Portal V6	4
MICA	?	ENSG00000204520	muscularis mucosae	3.42×10-6 (p-value)	GTEx Portal V6	3
HCG27	?	ENSG00000206344	lung	3.97×10-7 (p-value)	GTEx Portal V6	15
PSORS1C2	?	ENSG00000204538	lung	1.93×10-10 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	lung	2.54×10-50 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	lung	7.37×10-7 (p-value)	GTEx Portal V6	8
XXbac-BPG248L24.12	?	ENSG00000271581	lung	8.06×10-45 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	lung	5.60×10-14 (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	lung	3.52×10-8 (p-value)	GTEx Portal V6	12
PSORS1C1	?	ENSG00000204540	lung	6.21×10-10 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	lung	3.49×10-7 (p-value)	GTEx Portal V6	15
XXbac-BPG181B23.7	?	ENSG00000272221	lung	5.58×10-7 (p-value)	GTEx Portal V6	6
HCG27	?	ENSG00000206344	tibial artery	5.55×10-9 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	tibial artery	1.06×10-79 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	tibial artery	3.62×10-13 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	4.17×10-48 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	tibial artery	3.17×10-17 (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	tibial artery	5.49×10-13 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	tibial artery	6.30×10-11 (p-value)	GTEx Portal V6	16
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	1.26×10-6 (p-value)	GTEx Portal V6	4
MICA	?	ENSG00000204520	tibial artery	3.59×10-6 (p-value)	GTEx Portal V6	4
BAG6	?	ENSG00000204463	tibial artery	1.98×10-7 (p-value)	GTEx Portal V6	5
CYP21A1P	?	ENSG00000204338	tibial artery	4.68×10-6 (p-value)	GTEx Portal V6	1
HCG27	?	ENSG00000206344	thyroid	1.62×10-11 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	thyroid	2.85×10-13 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	thyroid	3.56×10-57 (p-value)	GTEx Portal V6	16
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	4.48×10-10 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	3.40×10-50 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	thyroid	7.07×10-17 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	thyroid	9.83×10-15 (p-value)	GTEx Portal V6	16
MICA	?	ENSG00000204520	thyroid	1.53×10-10 (p-value)	GTEx Portal V6	15
CYP21A1P	?	ENSG00000204338	thyroid	1.12×10-6 (p-value)	GTEx Portal V6	11

XXbac-BPG299F13.17	?	ENSG00000272501	thyroid	6.54×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	prostate	1.78×10-10 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	prostate	1.97×10-11 (p-value)	GTEx Portal V6	16
HCG27	?	ENSG00000206344	transverse colon	2.24×10-7 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	transverse colon	1.43×10-8 (p-value)	GTEx Portal V6	12
HLA-C	?	ENSG00000204525	transverse colon	2.23×10-43 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	transverse colon	1.53×10-9 (p-value)	GTEx Portal V6	14
XXbac-BPG248L24.12	?	ENSG00000271581	transverse colon	1.07×10-30 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	transverse colon	5.66×10-11 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	transverse colon	9.90×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	transverse colon	3.62×10-6 (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	transverse colon	1.22×10-6 (p-value)	GTEx Portal V6	3
HLA-C	?	ENSG00000204525	liver	6.43×10-25 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	liver	3.85×10-14 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	terminal ileum	6.70×10-16 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	terminal ileum	8.75×10-12 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	spleen	1.66×10-20 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	spleen	1.14×10-16 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	spleen	4.21×10-6 (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	spleen	9.44×10-7 (p-value)	GTEx Portal V6	4
HLA-C	?	ENSG00000204525	nucleus accumbens	2.12×10-17 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	nucleus accumbens	5.29×10-17 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	sigmoid colon	9.31×10-34 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	sigmoid colon	7.47×10-23 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	sigmoid colon	1.92×10-8 (p-value)	GTEx Portal V6	14
HLA-B	?	ENSG00000234745	sigmoid colon	4.75×10-7 (p-value)	GTEx Portal V6	12
PSORS1C1	?	ENSG00000204540	sigmoid colon	1.05×10-7 (p-value)	GTEx Portal V6	10
PSORS1C2	?	ENSG00000204538	sigmoid colon	1.94×10-6 (p-value)	GTEx Portal V6	6
HLA-C	?	ENSG00000204525	coronary artery	7.07×10-25 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	coronary artery	1.41×10-19 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	coronary artery	2.10×10-6 (p-value)	GTEx Portal V6	5
PSORS1C1	?	ENSG00000204540	coronary artery	1.09×10-6 (p-value)	GTEx Portal V6	10
HLA-C	?	ENSG00000204525	vagina	5.14×10-16 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	vagina	1.75×10-15 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	gastroesophageal junction	7.53×10-35 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	gastroesophageal junction	2.08×10-23 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	gastroesophageal junction	1.14×10-6 (p-value)	GTEx Portal V6	11

PSORS1C1	?	ENSG00000204540	gastroesophageal junction	2.66×10-8 (p-value)	GTEx Portal V6	12
PSORS1C2	?	ENSG00000204538	gastroesophageal junction	8.80×10-7 (p-value)	GTEx Portal V6	11
HLA-B	?	ENSG00000234745	gastroesophageal junction	9.26×10-10 (p-value)	GTEx Portal V6	15
HLA-C	?	ENSG00000204525	adrenal gland	1.62×10-30 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	adrenal gland	7.51×10-18 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	adrenal gland	2.51×10-8 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	adrenal gland	5.60×10-7 (p-value)	GTEx Portal V6	13
TNXB	?	ENSG00000168477	adrenal gland	1.65×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	cerebellar hemisphere	1.11×10-14 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	cerebellar hemisphere	1.21×10-14 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	cerebellar hemisphere	3.42×10-6 (p-value)	GTEx Portal V6	2
WASF5P	?	ENSG00000231402	cerebellum	1.50×10-10 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	cerebellum	3.49×10-22 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	cerebellum	3.72×10-9 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	cerebellum	4.97×10-21 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	hippocampus	2.41×10-18 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	hippocampus	6.93×10-14 (p-value)	GTEx Portal V6	16
VARS2	ENST00000321897	ILMN_1737585	skin	1.65×10-8 (p-value)	MuTHER consortium	6
VARS2	ENST00000625423		adipocyte	4.72×10-5 (p-value)	MuTHER consortium	6
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	2.93×10-6 (p-value)	MuTHER consortium	6
ATP6V1G2	ENST00000303892					
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					
HCG22	ENST00000570223	ILMN_1667229	blood	2.34×10-5 (p-value)	MuTHER consortium	4
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
MICB	ENST00000538442	ILMN_1708006	skin	2.04×10-5 (p-value)	MuTHER consortium	3
MICB	ENST00000399150					
MICB	ENST00000252229					
HCG27	ENST00000383331	ILMN_1746436	skin	1.35×10-8 (p-value)	MuTHER consortium	6
HCG27	ENST00000415276		adipocyte	3.10×10-15 (p-value)	MuTHER consortium	6
PSORS1C1	ENST00000481450	ILMN_1799669	skin	1.34×10-5 (p-value)	MuTHER consortium	3
PSORS1C1	ENST00000552747					
PSORS1C1	ENST00000547221					
PSORS1C1	ENST00000259881					
PSORS1C1	ENST00000479581					

?	?	ILMN_1893395	blood	6.11×10-7 (p-value)	Westra et al.	5
VARS2	ENST00000321897	ILMN_1737585	monocyte	3.11×10-12 (p-value)	Fairfax et al.	1
VARS2	ENST00000625423		b-cell	2.34×10-8 (p-value)	Fairfax et al.	1
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
CCHCR1	?	ENSG00000204536	blood	3.04×10-2 (q-value)	SeeQTL DB (HapMap)	2
HLA-B	?	ENSG00000234745	blood	3.70×10-2 (q-value)	SeeQTL DB (HapMap)	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CSF3R	ENST00000464465	ILMN_2371280	chr1	blood	7.65×10-21 (p-value)	Westra et al.	1
CSF3R	ENST00000373104						
CSF3R	ENST00000487540						
CSF3R	ENST00000373106						
CSF3R	ENST00000331941						
CSF3R	ENST00000484762						
CSF3R	ENST00000480825						
CSF3R	ENST00000361632						
CSF3R	ENST00000373103						
TSSC4	ENST00000451491	ILMN_1802753	chr11	blood	1.14×10-8 (p-value)	Westra et al.	1
TSSC4	ENST00000437110						
TSSC4	ENST00000496468						
TSSC4	ENST00000435795						
TSSC4	ENST00000440813						
TSSC4	ENST00000485682						
TSSC4	ENST00000467308						
TSSC4	ENST00000380996						
TSSC4	ENST00000333256						
TMEM154	ENST00000304385	ILMN_2088124	chr4	blood	2.03×10-78 (p-value)	Westra et al.	1
CD248	ENST00000311330	ILMN_1726589	chr11	blood	2.09×10-9 (p-value)	Westra et al.	1
XRCC6	ENST00000405878	ILMN_2166506	chr22	blood	4.99×10-19 (p-value)	Westra et al.	1
XRCC6	ENST00000359308						
XRCC6	ENST00000405506						
XRCC6	ENST00000360079						
XRCC6	ENST00000402580						
XRCC6	ENST00000428575						

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487828 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K27me3, PolII, TAF1, H3K4me2, SP1, H3K4me3
		HSMMtube	H3K27me3, H2AZ, H3K36me3, DNase1, H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H3K4me3, H3K27me3, DNase1
		liver (HepG2)	H3K4me2, H3K4me3, H3K36me3, H3K27me3

blood (GM12878)	H3K79me2, H3K4me3, NFKB, H3K27ac, H4K20me1, Cfos, H3K4me2, H3K9ac, H3K36me3, IRF4, DNase1, TAF1, PU1, PolII, H2AZ
lung (IMR90)	DNase1, H3K27me3, H3K36me3, H3K4me3, CTCF
nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K27me3, DNase1
skin (NHEK)	CTCF, H3K9ac, H3K4me2, H3K4me3, H3K36me3, H3K27me3
NHLF	H3K4me3, H3K9ac, H3K27ac, H3K36me3
Osteobl	H3K27me3, H3K36me3, H3K4me2, H3K4me3
blood (DND-41)	H3K36me3, H3K27me3, H3K9ac
breast (HMEC)	CTCF, DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
cervix (HeLa-S3)	PolII, H3K9ac, H3K4me2, H3K27ac, H3K4me3, DNase1
monocytes (Monocytes-CD14+)	H3K36me3, H3K4me3, H4K20me1, H3K9ac, DNase1, H3K4me2, H3K27ac
endothelium (HUVEC)	H3K4me3, PolII, H3K27me3, DNase1
A549	H3K27me3, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HLA-B	upstream gene variant	1010	ENST00000474381	?	?	5
HLA-B	upstream gene variant	2839	ENST00000497377	?	?	2
HLA-B	upstream gene variant	3109	ENST00000481849	?	?	2
HLA-B	upstream gene variant	2113	ENST00000463574	?	?	5
HLA-B	upstream gene variant	736	ENST00000498007	?	?	5
HLA-B	upstream gene variant	728	ENST00000412585	NM_005514.6	ENSP00000399168	5
HLA-B	upstream gene variant	727	ENST00000434333	?	ENSP00000405931	5
HLA-C	upstream gene variant	757	ENST00000620806	?	ENSP00000480331	5
HLA-C	upstream gene variant	2226	ENST00000376228	NM_002117.5	ENSP00000365402	1
HLA-C	upstream gene variant	2262	ENST00000487245	?	?	1
HLA-C	upstream gene variant	4646	ENST00000466892	?	?	1
HLA-C	upstream gene variant	2226	ENST00000383329	?	ENSP00000372819	1
HLA-C	upstream gene variant	3890	ENST00000470363	?	?	1
HLA-C	upstream gene variant	2222	ENST00000484378	?	?	1
HLA-C	upstream gene variant	2182	ENST00000495835	?	?	1
HLA-C	upstream gene variant	2207	ENST00000376237	?	ENSP00000365412	1
HLA-C	upstream gene variant	2243	ENST00000415537	?	ENSP00000400410	1
MIR6891	upstream gene variant	2599	ENST00000618788	?	?	4
RPL3P2	downstream gene variant	3100	ENST00000413027	?	?	1
USP8P1	upstream gene variant	1260	ENST00000494673	?	?	1
WASF5P	downstream gene variant, upstream gene variant	770	ENST00000428639	?	?	2
XXbac-BPG248L24.10	downstream gene variant	3331	ENST00000421191	?	?	2
XXbac-BPG248L24.12	downstream gene variant	278	ENST00000603274	?	?	5
XXbac-BPG248L24.13	downstream gene variant, upstream gene variant	2148	ENST00000539514	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HLA-C	ENST00000620806	?	ENSP00000480331	11
XXbac-BPG248L24.13	ENST00000539514	?	?	6

Locus 388 (rs6687487)

SNiPAcard

Block annotations

Block info

genomic range	chr1:23,060,951-23,066,767
block size	5,817 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.019 [-1.222 – 0.84]	gene(s) hit or close-by	EPHB2
phastCons	μ = 0.031 [0 – 0.123]	eQTL gene(s)	EPHB2
GERP++	μ = 1.218 [-1.12 – 3.76]	potentially regulated gene(s)	C1QA, C1QA, C1QB, C1QB
CADD score	μ = 6.540 [0.389 – 15.18]	disease gene(s)	EPHB2, C1QA, C1QB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
EPHB2	PROSTATE CANCER/BRAIN CANCER SUSCEPTIBILITY	OMIM	MIM:603688
EPHB2	PROSTATE CANCER	OMIM	MIM:176807
C1QA	C1q DEFICIENCY	OMIM	MIM:613652
C1QB	C1q DEFICIENCY	OMIM	MIM:613652
EPHB2	Familial prostate cancer	OrphaNet	OrphaNet:1331
C1QA	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147
C1QB	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
EPHB2	?	ENSG00000133216	blood	4.78×10^{-8} (p-value)	GTEx Portal V6	5
EPHB2	ENST00000400191	ILMN_2367384	monocyte	1.48×10^{-17} (p-value)	Fairfax et al.	1
EPHB2	ENST00000400191	ILMN_1785061	monocyte	1.93×10^{-10} (p-value)	Fairfax et al.	1
EPHB2	ENST00000400191	ILMN_1785061	monocyte	7.42×10^{-17} (p-value)	Zeller et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000008694	1	ENCP00000001089	C1QA C1QA
		ENCP00000001092	C1QB C1QB

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001518245	2	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac

(promoter flanking region)		NHLF	DNase1
		Osteobl	H3K27ac
		blood (K562)	H3K27me3
ENSR00001518246	1	NHLF	DNase1
(promoter flanking region)		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (K562)	DNase1, H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	DNase1, Cjun, H3K4me1
		lung (IMR90)	H4K5ac, DNase1, H3K18ac, H3K27ac, H3K4me2
		nervous (NH-A)	DNase1, H3K27ac, H3K9ac
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
EPHB2	ENST00000400191	?	ENSP00000383053	5
EPHB2	ENST00000374632	NM_004442.6	ENSP00000363763	5
EPHB2	ENST00000544305	?	ENSP00000444174	5
EPHB2	ENST00000374630	NM_017449.3	ENSP00000363761	5

Locus 389 (rs2559856)

SNiPAcard

Block annotations

Block info

genomic range	chr12:102,080,976-102,093,459
block size	12,484 bp
variant count	41 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.275 [-3.362 – 1.909]	gene(s) hit or close-by	CHPT1, MYBPC1, RP11-285E23.2
phastCons	μ = 0.029 [0 – 0.492]	eQTL gene(s)	CHPT1, GNPTAB
GERP++	μ = -0.432 [-4.69 – 2.62]	potentially regulated gene(s)	–
CADD score	μ = 2.917 [0.018 – 8.437]	disease gene(s)	GNPTAB, MYBPC1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
GNPTAB	mucolipidosis type III complementation group A (MLIIIA)	DECIPHER	MIM:252600
GNPTAB	mucolipidosis type II (MLII)	DECIPHER	MIM:252500
GNPTAB	MUCOLIPIDOSIS II ALPHA/BETA	OMIM	MIM:252500
GNPTAB	MUCOLIPIDOSIS III ALPHA/BETA	OMIM	MIM:252600
MYBPC1	ARTHROGRYPOSIS, DISTAL, TYPE 1B	OMIM	MIM:614335
MYBPC1	LETHAL CONGENITAL CONTRACTURE SYNDROME 4	OMIM	MIM:614915
GNPTAB	Mucolipidosis type 3	OrphaNet	OrphaNet:577
GNPTAB	Mucolipidosis type 2	OrphaNet	OrphaNet:576
MYBPC1	lethal congenital contracture syndrome type 3	OrphaNet	OrphaNet:137783
MYBPC1	Digitotalar dysmorphism	OrphaNet	OrphaNet:1146

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CHPT1	?	ENSG00000111666	pancreas	2.04×10^{-10} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	lung	2.66×10^{-10} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	tibial nerve	5.21×10^{-10} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	transformed fibroblasts	8.22×10^{-40} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	blood	2.02×10^{-13} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	tibial artery	8.25×10^{-33} (p-value)	GTEx Portal V6	41
GNPTAB	?	ENSG00000111670	tibial artery	1.43×10^{-6} (p-value)	GTEx Portal V6	5
CHPT1	?	ENSG00000111666	esophagus mucosa	1.12×10^{-14} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	thyroid	2.38×10^{-16} (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	gastroesophageal junction	2.55×10^{-6} (p-value)	GTEx Portal V6	2
CHPT1	?	ENSG00000111666	skeletal muscle	6.95×10^{-24} (p-value)	GTEx Portal V6	41

CHPT1	?	ENSG00000111666	transverse colon	1.05×10-9 (p-value)	GTEx Portal V6	39
CHPT1	?	ENSG00000111666	sun exposed skin	1.16×10-25 (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	unexposed skin	1.36×10-12 (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	aorta	4.81×10-17 (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	subcutaneous adipocytes	3.43×10-27 (p-value)	GTEx Portal V6	41
CHPT1	?	ENSG00000111666	muscularis mucosae	1.26×10-9 (p-value)	GTEx Portal V6	40
CHPT1	?	ENSG00000111666	atrial appendage	1.10×10-7 (p-value)	GTEx Portal V6	37
CHPT1	?	ENSG00000111666	EBV lymphocytes	4.35×10-9 (p-value)	GTEx Portal V6	40
CHPT1	?	ENSG00000111666	left ventricle	6.67×10-8 (p-value)	GTEx Portal V6	35
CHPT1	?	ENSG00000111666	visceral adipocytes	1.23×10-10 (p-value)	GTEx Portal V6	40
CHPT1	?	ENSG00000111666	sigmoid colon	6.14×10-6 (p-value)	GTEx Portal V6	3
CHPT1	?	ENSG00000111666	coronary artery	1.63×10-9 (p-value)	GTEx Portal V6	40
CHPT1	?	ENSG00000111666	cerebellum	4.38×10-8 (p-value)	GTEx Portal V6	36
CHPT1	?	ENSG00000111666	blood	6.05×10-16 (q-value)	SeeQTL DB (HapMap)	7
CHPT1	ENST00000229266	ILMN_2202940	adipocyte	2.25×10-5 (p-value)	MuTHER consortium	7
CHPT1	ENST00000549872					
CHPT1	ENST00000552215					
CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
?	?	ILMN_1729112	monocyte	3.62×10-40 (p-value)	Zeller et al.	4
CHPT1	ENST00000229266	ILMN_2202940	monocyte	7.88×10-17 (p-value)	Zeller et al.	4
CHPT1	ENST00000549872					
CHPT1	ENST00000552215					
CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
CHPT1	ENST00000549872	230364_at	blood	1.30×10-9 (p-value)	Dixon et al.	2
CHPT1	ENST00000552215					
CHPT1	ENST00000229266	221675_s_at	blood	9.00×10-17 (p-value)	Dixon et al.	2
CHPT1	ENST00000552215					
CHPT1	ENST00000549872					
CHPT1	ENST00000552351					
CHPT1	ENST00000549128					
?	?	1559739_at	blood	3.10×10-8 (p-value)	Dixon et al.	1
?	?	ILMN_1729112	monocyte	8.28×10-7 (p-value)	Fairfax et al.	2
			b-cell	3.32×10-4 (p-value)		
CHPT1	?	ENSG00000111666	uterus	5.49×10-7 (p-value)	GTEx Portal V6	3
CHPT1	?	ENSG00000111666	hippocampus	1.23×10-6 (p-value)	GTEx Portal V6	1
CHPT1	?	ENSG00000111666	pituitary	3.42×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000090464 (promoter)	4	embryonic stem cell (H1ESC)	DNase1, H3K27me3, PolII, Rad21, H3K9ac, Yy1, TAF1, H3K4me2, H3K4me3
		HSMMtube	H3K9ac, H3K4me2, H2AZ
		blood (K562)	TAF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3, H2AZ, H3K79me2, H3K27ac, Max, ZBTB7A, H3K9ac, Cmyc
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H3K4me2, H3K4me3, H2AZ
		liver (HepG2)	PolII, H3K79me2, TAF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, CTCF, H3K4me3, DNase1
		lung (IMR90)	H3K4me2, H3K4me3
		blood (GM12878)	DNase1, PolII, H2AZ, CTCF, Yy1, H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K27ac, H3K4me3, H3K9ac
		Osteobl	H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K27me3, H3K4me3, H3K9ac, H3K4me2
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, CTCF, PolII, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K36me3, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CHPT1	upstream gene variant	71	ENST00000549872	?	ENSP00000448766	26
CHPT1	upstream gene variant	26	ENST00000229266	NM_020244.2	ENSP00000229266	26
CHPT1	upstream gene variant	91	ENST00000549128	?	ENSP00000446994	26
CHPT1	upstream gene variant	479	ENST00000550385	?	?	24
CHPT1	upstream gene variant	253	ENST00000546490	?	?	26
CHPT1	upstream gene variant	91	ENST00000552351	?	ENSP00000447887	26
CHPT1	upstream gene variant	364	ENST00000552215	?	ENSP00000448831	26
MYBPC1	downstream gene variant	1350	ENST00000441232	?	ENSP00000388989	11
MYBPC1	downstream gene variant	1318	ENST00000550501	?	?	11
MYBPC1	downstream gene variant	1583	ENST00000541119	NM_001254720.1	ENSP00000442847	11
MYBPC1	downstream gene variant	1180	ENST00000549145	?	ENSP00000447660	12
MYBPC1	downstream gene variant	1328	ENST00000361466	NM_002465.3	ENSP00000354849	11
MYBPC1	downstream gene variant	1392	ENST00000392934	NM_001254723.1	ENSP00000376665	11
MYBPC1	downstream gene variant	1387	ENST00000549608	?	?	11
MYBPC1	downstream gene variant	1329	ENST00000452455	NM_001254718.1	ENSP00000400908	11
MYBPC1	downstream gene variant	1386	ENST00000547509	?	ENSP00000447362	11
MYBPC1	downstream gene variant	1329	ENST00000551300	?	ENSP00000447116	11
MYBPC1	downstream gene variant	1322	ENST00000361685	NM_206819.2	ENSP00000354845	11
MYBPC1	downstream gene variant	2803	ENST00000550270	NM_206820.2	ENSP00000449702	10
MYBPC1	downstream gene variant	1387	ENST00000547405	NM_001254722.1	ENSP00000448175	11
MYBPC1	downstream gene variant	1583	ENST00000536007	NM_001254721.1	ENSP00000446128	11
MYBPC1	downstream gene variant	1583	ENST00000545503	NM_001254719.1	ENSP00000440034	11
MYBPC1	downstream gene variant	1328	ENST00000553190	NM_206821.2	ENSP00000447900	11
RP11-285E23.2	downstream gene variant, upstream gene variant	18	ENST00000616668	?	?	27

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CHPT1	ENST00000229266	NM_020244.2	ENSP00000229266	2
CHPT1	ENST00000550385	?	?	4
CHPT1	ENST00000552215	?	ENSP00000448831	2
CHPT1	ENST00000549872	?	ENSP00000448766	2
CHPT1	ENST00000546490	?	?	2
CHPT1	ENST00000552351	?	ENSP00000447887	2
CHPT1	ENST00000549128	?	ENSP00000446994	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-285E23.2	ENST00000616668	?	2

Locus 390 (rs4834770)

SNiPAcard

Block annotations

Block info

genomic range	chr4:120,224,565-120,244,029
block size	19,465 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.608 [-3.553 – 0.489]	gene(s) hit or close-by	C4orf3, FABP2
phastCons	μ = 0.074 [0 – 0.763]	eQTL gene(s)	C4orf3, FABP2, KLHL2P1, PDE5A, RP11-33B1.1, RP11-33B1.4, RP11-384K6.2, RP11-384K6.6, RP11-548H18.2, USP53
GERP++	μ = -0.928 [-5.65 – 2.35]	potentially regulated gene(s)	–
CADD score	μ = 2.442 [0.004 – 7.632]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
2-aminobutyrate	8.22×10^{-5}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-33B1.4	?	ENSG00000260091	lung	4.08×10^{-8} (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	lung	9.81×10^{-15} (p-value)	GTEx Portal V6	11
RP11-384K6.6	?	ENSG00000260404	lung	1.65×10^{-6} (p-value)	GTEx Portal V6	10
FABP2	?	ENSG00000145384	atrial appendage	6.63×10^{-8} (p-value)	GTEx Portal V6	1
RP11-33B1.1	?	ENSG00000245958	atrial appendage	4.43×10^{-6} (p-value)	GTEx Portal V6	8
RP11-33B1.1	?	ENSG00000245958	transformed fibroblasts	3.88×10^{-15} (p-value)	GTEx Portal V6	11
RP11-384K6.6	?	ENSG00000260404	transformed fibroblasts	2.68×10^{-6} (p-value)	GTEx Portal V6	10
RP11-33B1.1	?	ENSG00000245958	tibial artery	3.95×10^{-10} (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	breast	4.40×10^{-7} (p-value)	GTEx Portal V6	11
RP11-33B1.4	?	ENSG00000260091	blood	1.09×10^{-8} (p-value)	GTEx Portal V6	11
C4orf3	?	ENSG00000164096	blood	5.54×10^{-9} (p-value)	GTEx Portal V6	11
FABP2	?	ENSG00000145384	blood	7.55×10^{-10} (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	blood	5.93×10^{-7} (p-value)	GTEx Portal V6	9
RP11-384K6.6	?	ENSG00000260404	blood	1.20×10^{-11} (p-value)	GTEx Portal V6	11
RP11-384K6.2	?	ENSG00000225892	blood	1.82×10^{-6} (p-value)	GTEx Portal V6	3
RP11-33B1.1	?	ENSG00000245958	thyroid	9.67×10^{-17} (p-value)	GTEx Portal V6	11

KLHL2P1	?	ENSG00000250412	thyroid	1.88×10-5 (p-value)	GTEx Portal V6	1
RP11-33B1.1	?	ENSG00000245958	skeletal muscle	2.15×10-10 (p-value)	GTEx Portal V6	11
RP11-384K6.6	?	ENSG00000260404	skeletal muscle	1.29×10-6 (p-value)	GTEx Portal V6	10
FABP2	?	ENSG00000145384	transverse colon	1.67×10-9 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	transverse colon	7.12×10-12 (p-value)	GTEx Portal V6	11
RP11-548H18.2	?	ENSG00000249244	EBV lymphocytes	6.00×10-8 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	EBV lymphocytes	7.49×10-9 (p-value)	GTEx Portal V6	11
PDE5A	?	ENSG00000138735	EBV lymphocytes	6.40×10-7 (p-value)	GTEx Portal V6	10
RP11-33B1.1	?	ENSG00000245958	unexposed skin	2.81×10-6 (p-value)	GTEx Portal V6	1
FABP2	?	ENSG00000145384	left ventricle	9.80×10-8 (p-value)	GTEx Portal V6	2
RP11-33B1.1	?	ENSG00000245958	left ventricle	4.44×10-11 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	aorta	1.37×10-7 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	subcutaneous adipocytes	4.04×10-12 (p-value)	GTEx Portal V6	11
RP11-548H18.2	?	ENSG00000249244	subcutaneous adipocytes	3.75×10-6 (p-value)	GTEx Portal V6	10
RP11-33B1.1	?	ENSG00000245958	visceral adipocytes	1.16×10-7 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	cortex	9.60×10-7 (p-value)	GTEx Portal V6	1
RP11-33B1.1	?	ENSG00000245958	nucleus accumbens	2.09×10-8 (p-value)	GTEx Portal V6	11
RP11-548H18.2	?	ENSG00000249244	tibial nerve	5.69×10-10 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	tibial nerve	6.99×10-14 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	sigmoid colon	5.88×10-6 (p-value)	GTEx Portal V6	1
RP11-548H18.2	?	ENSG00000249244	anterior cingulate cortex	9.29×10-9 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	esophagus mucosa	1.03×10-12 (p-value)	GTEx Portal V6	11
FABP2	?	ENSG00000145384	adrenal gland	2.28×10-8 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	adrenal gland	1.44×10-6 (p-value)	GTEx Portal V6	8
RP11-33B1.4	?	ENSG00000260091	testis	3.93×10-10 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	testis	2.29×10-9 (p-value)	GTEx Portal V6	11
RP11-548H18.2	?	ENSG00000249244	frontal cortex	4.00×10-6 (p-value)	GTEx Portal V6	2
RP11-33B1.1	?	ENSG00000245958	frontal cortex	5.39×10-6 (p-value)	GTEx Portal V6	7
RP11-33B1.4	?	ENSG00000260091	pancreas	6.52×10-6 (p-value)	GTEx Portal V6	7
USP53	ENST00000450251	ILMN_1795275	b-cell	4.35×10-7 (p-value)	Fairfax et al.	1
USP53	ENST00000509769					
USP53	ENST00000274030					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001434619 (promoter flanking region)	3	embryonic stem cell (H1ESC)	Rad21, CTCF
		Osteobl	CTCF, H3K27ac
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF, Rad21
		A549	H3K27me3, CTCF
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C4orf3	upstream gene variant	2489	ENST00000504110	NM_001001701.3	ENSP00000485697	1
FABP2	downstream gene variant, upstream gene variant	177	ENST00000274024	NM_000134.3	ENSP00000274024	5

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
FABP2	ENST00000274024	NM_000134.3	ENSP00000274024	N	aaT/aaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C4orf3	ENST00000399075	NM_001170330.1	ENSP00000382026	1
FABP2	ENST00000274024	NM_000134.3	ENSP00000274024	3

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FABP2	ENST00000274024	NM_000134.3	ENSP00000274024	2

Locus 391 (rs6842047)

SNiPAcard

Block annotations

Block info

genomic range	chr4:187,122,904-187,179,486
block size	56,583 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.796 [-2.537 – 0.27]	gene(s) hit or close-by	CYP4V2, KLKB1
phastCons	μ = 0.029 [0 – 0.386]	eQTL gene(s)	CYP4V2, KLKB1
GERP++	μ = -1.076 [-8.01 – 2.8]	potentially regulated gene(s)	F11, FAT1
CADD score	μ = 2.122 [0.111 – 6.993]	disease gene(s)	F11, CYP4V2, KLKB1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
F11	FACTOR XI DEFICIENCY	OMIM	MIM:612416
CYP4V2	BIETTI CRYSTALLINE CORNEORETINAL DYSTROPHY	OMIM	MIM:210370
F11	Congenital factor XI deficiency	OrphaNet	OrphaNet:329
CYP4V2	Bietti crystalline dystrophy	OrphaNet	OrphaNet:41751
KLKB1	Congenital prekallikrein deficiency	OrphaNet	OrphaNet:749

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CYP4V2	?	ENSG00000145476	caudate basal ganglia	1.15×10^{-6} (p-value)	GTEx Portal V6	2
CYP4V2	?	ENSG00000145476	pancreas	4.50×10^{-13} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	stomach	1.94×10^{-6} (p-value)	GTEx Portal V6	5
CYP4V2	?	ENSG00000145476	lung	1.59×10^{-10} (p-value)	GTEx Portal V6	18
KLKB1	?	ENSG00000164344	tibial nerve	5.79×10^{-13} (p-value)	GTEx Portal V6	11
CYP4V2	?	ENSG00000145476	blood	5.18×10^{-12} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	breast	2.79×10^{-7} (p-value)	GTEx Portal V6	6
CYP4V2	?	ENSG00000145476	esophagus mucosa	1.17×10^{-13} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	thyroid	7.67×10^{-18} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	skeletal muscle	4.60×10^{-10} (p-value)	GTEx Portal V6	17
CYP4V2	?	ENSG00000145476	liver	1.13×10^{-6} (p-value)	GTEx Portal V6	4
CYP4V2	?	ENSG00000145476	sun exposed skin	1.72×10^{-9} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	unexposed skin	4.57×10^{-9} (p-value)	GTEx Portal V6	18
CYP4V2	?	ENSG00000145476	subcutaneous adipocytes	2.19×10^{-7} (p-value)	GTEx Portal V6	16

CYP4V2 ENST00000502665		ILMN_2054607	monocyte	2.98×10-21 (p-value)	Zeller et al.	4
CYP4V2 ENST00000507209						
KLKB1 ENST00000511406		ILMN_1800468	adipocyte	2.44×10-5 (p-value)	MuTHER consortium	4
KLKB1 ENST00000511608						
KLKB1 ENST00000264690						
KLKB1 ENST00000513864						
CYP4V2 ENST00000502665		ILMN_2054607	blood	6.23×10-7 (p-value)	MuTHER consortium	5
CYP4V2 ENST00000507209						
CYP4V2	?	ENSG00000145476	adrenal gland	2.13×10-6 (p-value)	GTEx Portal V6	2
CYP4V2 ENST00000507209		ILMN_2054607	monocyte	2.13×10-12 (p-value)	Fairfax et al.	3
CYP4V2 ENST00000502665						

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000399558	1	ENCP00000043224	FAT1
		ENCP00000043223	FAT1
		ENCP00000043226	FAT1
		ENCP00000043217	F11
		ENCP00000043225	FAT1
		ENCP00000043222	FAT1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001256081 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, USF1, Yy1, CTCF, Rad21
		Osteobl	CTCF
		blood (K562)	Egr1, Max, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	DNase1, CTCF, Rad21
		lung (IMR90)	CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		A549	CTCF, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CYP4V2	downstream gene variant	204	ENST00000378802	NM_207352.3	ENSP00000368079	10
CYP4V2	upstream gene variant, downstream gene variant	331	ENST00000513354	?	?	10
CYP4V2	downstream gene variant	964	ENST00000507209	?	?	6
CYP4V2	upstream gene variant, downstream gene variant	519	ENST00000502665	?	?	7
KLKB1	upstream gene variant	2414	ENST00000513864	?	ENSP00000424469	2
KLKB1	upstream gene variant	1334	ENST00000428196	?	ENSP00000412366	2
KLKB1	upstream gene variant	2298	ENST00000264690	NM_000892.3	ENSP00000264690	2
KLKB1	upstream gene variant, downstream gene variant	40	ENST00000511406	?	?	3
KLKB1	upstream gene variant	2403	ENST00000446598	?	ENSP00000415563	2

KLKB1	upstream gene variant	2403	ENST00000446598	?	ENSP00000415363 2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CYP4V2	ENST00000507209	?	?	8
CYP4V2	ENST00000502665	?	?	7
CYP4V2	ENST00000378802	NM_207352.3	ENSP00000368079	4
CYP4V2	ENST00000513354	?	?	4
KLKB1	ENST00000264690	NM_000892.3	ENSP00000264690	2
KLKB1	ENST00000511608	?	ENSP00000426629	17
KLKB1	ENST00000511406	?	?	2
KLKB1	ENST00000513864	?	ENSP00000424469	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
KLKB1	ENST00000264690	NM_000892.3	ENSP00000264690	1
KLKB1	ENST00000511608	?	ENSP00000426629	1
KLKB1	ENST00000513864	?	ENSP00000424469	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CYP4V2	ENST00000507209	?	5
CYP4V2	ENST00000502665	?	4
CYP4V2	ENST00000513354	?	1

Locus 392 (rs17801525)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,612,262-51,618,167
block size	5,906 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.815 [-3.587 – 0.21]	gene(s) hit or close-by	CTU1
phastCons	μ = 0.054 [0.001 – 0.184]	eQTL gene(s)	SIGLEC9
GERP++	μ = -0.193 [-1.14 – 0.362]	potentially regulated gene(s)	SHANK1
CADD score	μ = 3.017 [0.628 – 7.501]	disease gene(s)	SHANK1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SHANK1	AUTISM	DECIPHER	MIM:209850 OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC9	?	ENSG00000129450	cerebellar hemisphere	6.90×10-7 (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000232477	1	ENCP00000024630	SHANK1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001419940 (promoter)	1	embryonic stem cell (H1ESC)	Gabp, Sin3Ak20, SIX5, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	Max, H3K27ac, H3K9ac, H3K4me3, DNase1, H3K36me3, CTCF, H3K4me2, PolII, Cmyc, CTCFL, ELF1, H2AZ, ETS1, Gabp
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K4me3, H2AZ
		liver (HepG2)	DNase1, H3K4me3, Cmyc, H3K27ac, H3K9ac, H3K4me2, ELF1, PolII, Gabp
		blood (GM12878)	ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, EBF1, SIX5, PolII, H2AZ, ETS1, DNase1, Cmyc, Gabp
		lung (IMR90)	H3K4me2, H3K4me3, DNase1
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K4me3, DNase1, H3K4me2, H3K9ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me3, H3K27ac, H2AZ, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, PolII, Max, H3K4me3, H3K79me2, Gabp, Cmyc

	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
	endothelium (HUVEC)	Max, H3K4me3, PolII, Cmyc, DNase1
	A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000217695 2 (CTCF binding site)	embryonic stem cell (H1ESC)	Rad21, CTCF
	HSMMtube	H3K27me3
	blood (K562)	CTCF, H3K27me3
	blood (DND-41)	H3K27me3
	skin (NHDF-AD)	CTCF
	breast (HMEC)	CTCF
	cervix (HeLa-S3)	CTCF
	monocytes (Monocytes-CD14+)	H3K4me1
	endothelium (HUVEC)	H3K27me3
	lung (IMR90)	H3K27me3
	A549	H3K27me3
	skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTU1	upstream gene variant	635	ENST00000421832	NM_145232.3	ENSP00000390011	6

Locus 393 (rs10473230)

SNiPAcard

Block annotations

Block info

genomic range	chr5:40,959,959-40,986,374
block size	26,416 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.031 [-2.44 – 3.368]	gene(s) hit or close-by	C7, RP11-301A5.2
phastCons	μ = 0.032 [0 – 0.34]	eQTL gene(s)	C6
GERP++	μ = -0.511 [-7.22 – 4.05]	potentially regulated gene(s)	C7, C7
CADD score	μ = 5.222 [0.192 – 14.47]	disease gene(s)	C7, C6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C7	COMPLEMENT COMPONENT 7 DEFICIENCY	OMIM	MIM:610102
C6	COMPLEMENT COMPONENT 6 DEFICIENCY	OMIM	MIM:612446
C7	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150
C6	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
C7	missense variant	ENST00000313164	NM_000587.2	ENSP00000322061	S/T	Tct/Act	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C6	ENST00000263413	ILMN_1688242	adipocyte	4.98×10^{-5} (p-value)	MuTHER consortium	1
C6	ENST00000337836					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000043959		C7

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000407549	1	ENCP00000043958	C7
			C7

	ENCP00000043961	C7 C7

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001692910 (enhancer)	1	embryonic stem cell (H1ESC)	H3K27me3
ENSR00001408896 (enhancer)	2	cervix (HeLa-S3)	H3K27me3
		NHLF	DNase1
		HSMMtube	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C7	upstream gene variant, downstream gene variant	163	ENST00000464864	?	?	7
C7	upstream gene variant, downstream gene variant	1145	ENST00000486779	?	?	6
C7	downstream gene variant	926	ENST00000513922	?	?	2
C7	downstream gene variant	1579	ENST00000494960	?	?	5
C7	downstream gene variant	16	ENST00000313164	NM_000587.2	ENSP00000322061	4
RP11-301A5.2	downstream gene variant	625	ENST00000504890	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C7	ENST00000513922	?	?	1
C7	ENST00000486779	?	?	2
C7	ENST00000313164	NM_000587.2	ENSP00000322061	10
C7	ENST00000494960	?	?	3
C7	ENST00000464864	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C7	ENST00000313164	NM_000587.2	ENSP00000322061	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C7	ENST00000486779	?	1

Locus 394 (rs1846899)

SNiPAcard

Block annotations

Block info

genomic range	chr3:186,472,816-186,475,666
block size	2,851 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.641 [-1.457 – 0.176]	**gene(s) hit or close-by**	PSMD10P2, RP11-573D15.8
phastCons	μ = 0.250 [0 – 0.5]	**eQTL gene(s)**	–
GERP++	μ = -1.788 [-3.68 – 0.104]	**potentially regulated gene(s)**	–
CADD score	μ = 6.903 [2.027 – 11.78]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001381876 (promoter flanking region)	1	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K4me2, H3K9ac, H3K27ac, H3K4me1, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PSMD10P2	upstream gene variant	3227	ENST00000432890	?	?	1
PSMD10P2	upstream gene variant	2816	ENST00000626135	?	?	1
PSMD10P2	upstream gene variant	3137	ENST00000627340	?	?	1
RP11-573D15.8	upstream gene variant	1272	ENST00000596632	?	?	2
RP11-573D15.8	upstream gene variant	150	ENST00000626586	?	?	1
RP11-573D15.8	downstream gene variant	1998	ENST00000608173	?	?	1
RP11-573D15.8	upstream gene variant	358	ENST00000625690	?	?	1
RP11-573D15.8	upstream gene variant	1563	ENST00000609652	?	?	2
RP11-573D15.8	upstream gene variant	659	ENST00000629201	?	?	1
RP11-573D15.8	upstream gene variant	209	ENST00000599314	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-573D15.8	ENST00000625869	?	?	2
RP11-573D15.8	ENST00000596329	?	?	2
RP11-573D15.8	ENST00000625690	?	?	1
RP11-573D15.8	ENST00000626520	?	?	2
RP11-573D15.8	ENST00000629954	?	?	2

RP11-573D15.8	ENST00000599314	?	?	1
RP11-573D15.8	ENST00000626586	?	?	1
RP11-573D15.8	ENST00000626978	?	?	2
RP11-573D15.8	ENST00000629201	?	?	1
RP11-573D15.8	ENST00000628291	?	?	2
RP11-573D15.8	ENST00000608173	?	?	1

Locus 395 (rs2075746)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,325,532-34,325,532
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.706	gene(s) hit or close-by	CCL15, CCL15-CCL14, RP11-104J23.1
phastCons	0	eQTL gene(s)	–
GERP++	1.62	potentially regulated gene(s)	–
CADD score	5.599	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL15	upstream gene variant	126	ENST00000614368	?	ENSP00000484262	1
RP11-104J23.1	upstream gene variant	2923	ENST00000619334	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL15	ENST00000617897	NM_032965.4	ENSP00000484078	1
CCL15-CCL14	ENST00000616694	?	ENSP00000481402	1
CCL15-CCL14	ENST00000610751	?	ENSP00000481940	1

Locus 396 (rs3743392)

SNiPAcard

Block annotations

Block info

genomic range	chr15:89,436,085-89,449,137
block size	13,053 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.677 [-2.626 – 1.105]	**gene(s) hit or close-by**	HAPLN3, MFGE8
phastCons	μ = 0.015 [0 – 0.2]	**eQTL gene(s)**	HAPLN3, MFGE8
GERP++	μ = -0.415 [-2.92 – 1.63]	**potentially regulated gene(s)**	MFGE8
CADD score	μ = 3.569 [0.602 – 8.556]	**disease gene(s)**	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
HAPLN3	stop lost	ENST00000562889	?	ENSP00000457180	*/Q	Tag/Cag	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HAPLN3	ENST00000558770	ILMN_1654319	blood	2.57×10^{-6} (p-value)	Westra et al.	6
HAPLN3	ENST00000359595					
MFGE8	?	ENSG00000140545	lung	9.36×10^{-10} (p-value)	GTEx Portal V6	12
MFGE8	?	ENSG00000140545	tibial nerve	7.12×10^{-8} (p-value)	GTEx Portal V6	4
MFGE8	?	ENSG00000140545	transformed fibroblasts	6.07×10^{-9} (p-value)	GTEx Portal V6	12
MFGE8	?	ENSG00000140545	esophagus mucosa	5.76×10^{-6} (p-value)	GTEx Portal V6	3
MFGE8	?	ENSG00000140545	left ventricle	3.44×10^{-6} (p-value)	GTEx Portal V6	5
MFGE8	?	ENSG00000140545	testis	2.47×10^{-6} (p-value)	GTEx Portal V6	4

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000215481	1	ENST00000268151, ENST00000558018, ENST00000268150	MFGE8
FFCP00000215480	1	ENST00000558018, ENST00000268151, ENST00000268150	MFGE8

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000413115	1	embryonic stem cell (H1ESC)	PolII, H3K36me3, DNase1

(TF binding site)	lung (IMR90)	H3K36me3
	A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HAPLN3	upstream gene variant	3688	ENST00000563808	?	?	2
HAPLN3	upstream gene variant	3668	ENST00000562281	?	ENSP00000456985	2
HAPLN3	upstream gene variant	3575	ENST00000359595	NM_178232.2	ENSP00000352606	2
HAPLN3	upstream gene variant	3670	ENST00000558770	?	ENSP00000456458	2
HAPLN3	upstream gene variant	3604	ENST00000562889	?	ENSP00000457180	2
MFGE8	downstream gene variant	114	ENST00000542878	?	ENSP00000444332	1
MFGE8	downstream gene variant	2113	ENST00000613965	?	ENSP00000478952	2
MFGE8	downstream gene variant	157	ENST00000558352	?	?	10
MFGE8	downstream gene variant, upstream gene variant	97	ENST00000560937	?	?	11
MFGE8	downstream gene variant, upstream gene variant	1260	ENST00000617199	?	?	3
MFGE8	downstream gene variant	4298	ENST00000558773	?	?	1
MFGE8	downstream gene variant	709	ENST00000559770	?	?	10
MFGE8	downstream gene variant	1521	ENST00000559259	?	?	9
MFGE8	downstream gene variant	1538	ENST00000559997	?	?	9
MFGE8	downstream gene variant, upstream gene variant	4	ENST00000560553	?	?	11
MFGE8	downstream gene variant	4874	ENST00000557944	?	?	1
MFGE8	downstream gene variant	1481	ENST00000558029	?	ENSP00000452926	2
MFGE8	downstream gene variant	3886	ENST00000559143	?	?	1

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
MFGE8	ENST00000558029	?	ENSP00000452926	1
MFGE8	ENST00000268151	NM_001114614.1	ENSP00000268151	1
MFGE8	ENST00000542878	?	ENSP00000444332	1
MFGE8	ENST00000558018	?	ENSP00000452734	1
MFGE8	ENST00000613965	?	ENSP00000478952	1
MFGE8	ENST00000268150	NM_005928.2	ENSP00000268150	1
MFGE8	ENST00000566497	?	ENSP00000456281	1
MFGE8	ENST00000559259	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HAPLN3	ENST00000359595	NM_178232.2	ENSP00000352606	2
HAPLN3	ENST00000558770	?	ENSP00000456458	2
HAPLN3	ENST00000563808	?	?	2
HAPLN3	ENST00000562889	?	ENSP00000457180	1
HAPLN3	ENST00000562281	?	ENSP00000456985	2

MFGE8	ENST00000560937	?	?	1
MFGE8	ENST00000559997	?	?	1
MFGE8	ENST00000558029	?	ENSP00000452926	10
MFGE8	ENST00000268151	NM_001114614.1	ENSP00000268151	12
MFGE8	ENST00000542878	?	ENSP00000444332	11
MFGE8	ENST00000558018	?	ENSP00000452734	12
MFGE8	ENST00000613965	?	ENSP00000478952	10
MFGE8	ENST00000268150	NM_005928.2	ENSP00000268150	12
MFGE8	ENST00000566497	?	ENSP00000456281	12

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MFGE8	ENST00000268151	NM_001114614.1	ENSP00000268151	1
MFGE8	ENST00000558018	?	ENSP00000452734	1
MFGE8	ENST00000268150	NM_005928.2	ENSP00000268150	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
MFGE8	ENST00000617199	?	9
MFGE8	ENST00000560553	?	1

Locus 397 (rs7121784)

SNiPAcard

Block annotations

Block info

genomic range	chr11:120,159,052-120,159,052
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	1.227	gene(s) hit or close-by	POU2F3
phastCons	0.117	eQTL gene(s)	–
GERP++	0.843	potentially regulated gene(s)	–
CADD score	2.745	disease gene(s)	–

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
POU2F3	ENST00000260264	NM_001244682.1	ENSP00000260264	1
POU2F3	ENST00000533620	?	ENSP00000435738	1
POU2F3	ENST00000543440	NM_014352.3	ENSP00000441687	1

Locus 398 (rs4851551)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,781,649-102,851,837
block size	70,189 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.487 [-3.749 – 0.596]	gene(s) hit or close-by	AC007271.3, IL1R1, IL1RL2
phastCons	μ = 0.008 [0 – 0.112]	eQTL gene(s)	–
GERP++	μ = -0.849 [-7.17 – 2.63]	potentially regulated gene(s)	AC007271.3, IL1R1, IL1RL1, IL1RL2
CADD score	μ = 3.879 [0.348 – 11.97]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
myo-inositol	5.22×10^{-5}	Metabolomics GWAS Server	24816252	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IL1R1	missense variant	ENST00000430171	?	ENSP00000408101	G/A	gGa/gCa	?	?	1
IL1R1	missense variant	ENST00000410023	NM_000877.3	ENSP00000386380	G/A	gGa/gCa	?	?	1
IL1R1	missense variant	ENST00000409589	?	ENSP00000386555	G/A	gGa/gCa	?	?	1
IL1R1	missense variant	ENST00000409288	?	ENSP00000386478	G/A	gGa/gCa	?	?	1
IL1R1	missense variant	ENST00000409329	?	ENSP00000387131	G/A	gGa/gCa	?	?	1
IL1R1	missense variant	ENST00000424272	?	ENSP00000415366	G/A	gGa/gCa	?	?	1
IL1R1	missense variant	ENST00000409929	NM_001288706.1	ENSP00000386776	G/A	gGa/gCa	?	?	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000257390	1	ENCP00000027309	IL1R1
		ENCP00000027310	IL1R1
		ENCP00000027316	IL1RL1
ENCE00000257391	2	ENCP00000027309	IL1R1
		ENCP00000027310	IL1R1
		ENCP00000027313	IL1RL2
		ENCP00000027311	AC007271.3
		ENCP00000027316	IL1RL1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000595628 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		Osteobl	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	CTCF
		liver (HepG2)	CTCF
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF
ENSR00000595632 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me2
		HSMMtube	DNase1, H3K9ac, H3K27ac
		blood (K562)	Cfos, Max, Jund, H2AZ, Cjun, PolII, DNase1
		skin (NHDF-AD)	DNase1, H3K9ac, H3K27ac
		muscle (HSMM)	DNase1, H3K27ac
		liver (HepG2)	FOSL2, Jund, H3K4me1, H2AZ, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	DNase1, H3K27ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K27ac
		skin (NHEK)	H3K4me1, H3K27ac, H3K4me2, H3K36me3, DNase1
		NHLF	DNase1, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, PolII, H3K9ac, Jund, H3K4me2, H3K27ac, Max, Cfos
		monocytes (Monocytes-CD14+)	H3K27ac, DNase1, H3K4me1
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me2, H3K4me1, H3K27ac, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00000014951 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, DNase1, Rad21, H3K27me3
		HSMMtube	DNase1, CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	Rad21, H3K4me1, CTCF, DNase1
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC007271.3	downstream gene variant, upstream gene variant	1662	ENST00000428188	?	?	3
IL1R1	downstream gene variant	27	ENST00000442590	?	ENSP00000393296	3
IL1R1	downstream gene variant	2561	ENST00000413623	?	ENSP00000407017	3
IL1R1	downstream gene variant	236	ENST00000452403	?	ENSP00000401646	3
IL1R1	downstream gene variant	2644	ENST00000409929	NM_001288706.1	ENSP00000386776	3
IL1R1	downstream gene variant	2644	ENST00000424272	?	ENSP00000415366	3
IL1R1	downstream gene variant	91	ENST00000410023	NM_000877.3	ENSP00000386380	3
IL1R1	downstream gene variant	3453	ENST00000430171	?	ENSP00000408101	1
IL1R1	downstream gene variant	53	ENST00000450319	?	ENSP00000411627	3
IL1R1	downstream gene variant	3092	ENST00000422532	?	ENSP00000390349	1
IL1R1	downstream gene variant	3026	ENST00000428279	?	ENSP00000410461	3

IL1R1	downstream gene variant	3026	ENST00000428279	?	ENSP00000410461	3
IL1R1	downstream gene variant	3644	ENST00000409288	?	ENSP00000386478	3
IL1R1	downstream gene variant	99	ENST00000409589	?	ENSP00000386555	3
IL1R1	downstream gene variant	3586	ENST00000409329	?	ENSP00000387131	3
IL1RL2	upstream gene variant	1142	ENST00000441515	?	ENSP00000413348	2
IL1RL2	upstream gene variant	1100	ENST00000264257	NM_003854.2	ENSP00000264257	2
IL1RL2	upstream gene variant	1133	ENST00000481806	?	?	2
IL1RL2	upstream gene variant, downstream gene variant	1221	ENST00000421464	?	ENSP00000387611	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC007271.3	ENST00000428188	?	?	2
IL1R1	ENST00000409589	?	ENSP00000386555	4
IL1R1	ENST00000422532	?	ENSP00000390349	3
IL1R1	ENST00000409929	NM_001288706.1	ENSP00000386776	3
IL1R1	ENST00000413623	?	ENSP00000407017	4
IL1R1	ENST00000409288	?	ENSP00000386478	3
IL1R1	ENST00000430171	?	ENSP00000408101	1
IL1R1	ENST00000428279	?	ENSP00000410461	4
IL1R1	ENST00000424272	?	ENSP00000415366	3
IL1R1	ENST00000410023	NM_000877.3	ENSP00000386380	4
IL1R1	ENST00000409329	?	ENSP00000387131	3
IL1RL2	ENST00000441515	?	ENSP00000413348	8
IL1RL2	ENST00000264257	NM_003854.2	ENSP00000264257	8
IL1RL2	ENST00000481806	?	?	8
IL1RL2	ENST00000421464	?	ENSP00000387611	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1R1	ENST00000409589	?	ENSP00000386555	1
IL1R1	ENST00000410023	NM_000877.3	ENSP00000386380	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1R1	ENST00000428279	?	ENSP00000410461	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC007271.3	ENST00000428188	?	2

Locus 399 (rs4585101)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,810,857-23,848,024
block size	37,168 bp
variant count	37 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.968 [-3.14 – 0.428]	**gene(s) hit or close-by**	CST2
phastCons	μ = 0.017 [0 – 0.339]	**eQTL gene(s)**	CST1, CST2
GERP++	μ = -0.151 [-2.13 – 1.63]	**potentially regulated gene(s)**	RP4-564O4.1, RP5-1100I6.1
CADD score	μ = 1.878 [0.04 – 5.506]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST2	?	ENSG00000170369	lung	6.42×10^{-8} (p-value)	GTEx Portal V6	6
CST1	ENST00000398402	ILMN_1753449	blood	6.62×10^{-5} (p-value)	Westra et al.	10
CST1	ENST00000304749					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000290875	1	ENCP00000031101	RP4-564O4.1
		ENCP00000031099	RP5-1100I6.1
ENCE00000290877	1	ENCP00000031101	RP4-564O4.1
		ENCP00000031099	RP5-1100I6.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001664592 (CTCF binding site)	2	HSMMtube	H3K27me3
ENSR00001664594 (TF binding site)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, USF1
		endothelium (HUVEC)	H3K36me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CST2	upstream gene variant	3489	ENST00000304725	NM_001322.2	ENSP00000307540	3

Locus 400 (rs3765967)

SNiPAcard

Block annotations

Block info

genomic range	chr1:9,004,120-9,011,627
block size	7,508 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.432 [-1.57 – 4.333]	gene(s) hit or close-by	CA6
phastCons	μ = 0.180 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 0.327 [-4.55 – 3.39]	potentially regulated gene(s)	CA6, RERE
CADD score	μ = 7.190 [0.168 – 23.5]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CA6	missense variant	ENST00000377436	NM_001270500.1	ENSP00000366654	A/G	gCc/gGc	?	?	1
CA6	missense variant	ENST00000377443	NM_001215.3	ENSP00000366662	A/G	gCc/gGc	?	?	1
CA6	missense variant	ENST00000480186	?	ENSP00000435280	A/G	gCc/gGc	?	?	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000435401	1	ENST00000377436, ENST00000480186, ENST00000377443	CA6
FFCP00000435402	1	ENST00000377436, ENST00000480186, ENST00000377443	CA6

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000003284	1	ENCP00000000458	RERE

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000530170 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		blood (K562)	H3K27me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CA6	upstream gene variant	1826	ENST00000377436	NM_001270500.1	ENSP00000366654	1
CA6	upstream gene variant	1806	ENST00000549778	?	ENSP00000447108	1
CA6	upstream gene variant	1822	ENST00000377443	NM_001215.3	ENSP00000366662	1
CA6	upstream gene variant	1821	ENST00000480186	?	ENSP00000435280	1
CA6	upstream gene variant	1807	ENST00000476083	?	?	1

CA6	upstream gene variant	1826	ENST00000377442	NM_001270501.1	ENSP00000366661	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CA6	ENST00000377436	NM_001270500.1	ENSP00000366654	G	ggG/ggC	2
CA6	ENST00000480186	?	ENSP00000435280	2	2	2
CA6	ENST00000377443	NM_001215.3	ENSP00000366662	G	ggG/ggC	2

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CA6	ENST00000377436	NM_001270500.1	ENSP00000366654	4
CA6	ENST00000377442	NM_001270501.1	ENSP00000366661	5
CA6	ENST00000480186	?	ENSP00000435280	4
CA6	ENST00000377443	NM_001215.3	ENSP00000366662	4
CA6	ENST00000476083	?	?	5
CA6	ENST00000549778	?	ENSP00000447108	5

Locus 402 (rs1462372)

SNiPAcard

Block annotations

Block info

genomic range	chr4:88,478,750-88,481,387
block size	2,638 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.475 [-0.199 – 0.83]	**gene(s) hit or close-by**	–
phastCons	μ = 0.018 [0 – 0.051]	**eQTL gene(s)**	HSD17B13, SPARCL1
GERP++	μ = 0.249 [-1.55 – 1.44]	**potentially regulated gene(s)**	–
CADD score	μ = 2.523 [1 – 3.358]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
stearidonate (18:4n3)	1.92×10^{-5}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SPARCL1	?	ENSG00000152583	tibial nerve	8.50×10^{-6} (p-value)	GTEx Portal V6	3
SPARCL1	?	ENSG00000152583	subcutaneous adipocytes	6.18×10^{-6} (p-value)	GTEx Portal V6	2
HSD17B13	?	ENSG00000170509	thyroid	4.47×10^{-6} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001686562 (enhancer)	1	blood (DND-41)	H3K27me3

Locus 403 (rs3135063)

SNiPAcard

Block annotations

Block info

genomic range	chr4:3,287,052-3,291,401
block size	4,350 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.490 [-0.944 – -0.147]	gene(s) hit or close-by	RGS12
phastCons	μ = 0.001 [0 – 0.004]	eQTL gene(s)	HTT-AS
GERP++	μ = -0.385 [-0.721 – 0.158]	potentially regulated gene(s)	AC141928.1, ADD1, ADRA2C, HGFAC, LRPAP1, MFSD10, RGS12, SH3BP2
CADD score	μ = 2.217 [0.588 – 5.087]	disease gene(s)	LRPAP1, SH3BP2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
LRPAP1	Extreme myopia;Myopia 23, autosomal recessive	DECIPHER	MIM:615431
SH3BP2	CHERUBISM	OMIM	MIM:118400
LRPAP1	MYOPIA 23, AUTOSOMAL RECESSIVE	OMIM	MIM:615431
SH3BP2	CHERUBISM	OrphaNet	OrphaNet:184
LRPAP1	Rare isolated myopia	OrphaNet	OrphaNet:98619

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HTT-AS	?	ENSG00000251075	aorta	8.73×10^{-8} (p-value)	GTEx Portal V6	3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000370653	1	ENCP00000040189	RGS12
		ENCP00000040129	SH3BP2
		ENCP00000040203	ADRA2C
		ENCP00000040193	HGFAC
		ENCP00000040188	RGS12
		ENCP00000040179	RGS12
		ENCP00000040176	RGS12
		ENCP00000040198	LRPAP1
		ENCP00000040142	ADD1
		ENCP00000040191	HGFAC
		ENCP00000040132	SH3BP2
ENCE00000370721	1	ENCP00000040141	SH3BP2

	ENCP00000040154	MFSD10
	ENCP00000040155	MFSD10
	ENCP00000040183	RGS12
	ENCP00000040202	AC141928.1
	ENCP00000040153	MFSD10
	ENCP00000040140	SH3BP2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001235104 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K4me2
		breast (HMEC)	H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me3
		liver (HepG2)	PolII, H3K79me2, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	PU1, H2AZ, H3K4me1, DNase1, H3K4me2
		nervous (NH-A)	H3K27me3, DNase1
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RGS12	upstream gene variant	3354	ENST00000506631	?	?	1

Locus 404 (rs757158)

SNiPAcard

Block annotations

Block info

genomic range	chr7:94,954,696-94,955,528
block size	833 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.623 [-0.924 – -0.321]	gene(s) hit or close-by	PON1
phastCons	μ = 0.001 [0 – 0.001]	eQTL gene(s)	ASB4, PON1, PON2
GERP++	μ = 0.224 [-0.317 – 0.764]	potentially regulated gene(s)	–
CADD score	μ = 3.414 [2.109 – 4.72]	disease gene(s)	PON1, PON2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Paraoxonase activity	$<5.00\times10^{-116}$	GWAS Catalog	22982463	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR000232	1

Disease gene annotation

gene	trait	source DB	source entry/link
PON1	MICROVASCULAR COMPLICATIONS OF DIABETES, SUSCEPTIBILITY TO, [...]	OMIM	MIM:612633
PON1	PARAOXONASE 1	OMIM	MIM:168820
PON2	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803
PON1	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PON1	?	ENSG00000005421	pancreas	2.91×10^{-9} (p-value)	GTEx Portal V6	2
ASB4	?	ENSG00000005981	tibial nerve	5.98×10^{-8} (p-value)	GTEx Portal V6	2
PON2	?	ENSG00000105854	tibial nerve	6.61×10^{-10} (p-value)	GTEx Portal V6	2
PON1	?	ENSG00000005421	pituitary	2.82×10^{-7} (p-value)	GTEx Portal V6	1
ASB4	?	ENSG00000005981	thyroid	7.13×10^{-10} (p-value)	GTEx Portal V6	2
PON2	?	ENSG00000105854	thyroid	3.32×10^{-7} (p-value)	GTEx Portal V6	2
PON1	?	ENSG00000005421	adrenal gland	5.05×10^{-20} (p-value)	GTEx Portal V6	2
ASB4	?	ENSG00000005981	unexposed skin	2.84×10^{-9} (p-value)	GTEx Portal V6	2
PON1	?	ENSG00000005421	liver	4.60×10^{-11} (p-value)	GTEx Portal V6	2
ASB4	?	ENSG00000005981	sun exposed skin	4.86×10^{-13} (p-value)	GTEx Portal V6	2

ASB4	?	ENSG00000005981	sun exposed skin	4.88×10-15 (p-value)	GTEx Portal V6	2
PON2	?	ENSG00000105854	left ventricle	3.73×10-7 (p-value)	GTEx Portal V6	2
PON2	?	ENSG00000105854	muscularis mucosae	2.07×10-5 (p-value)	GTEx Portal V6	1
PON2	?	ENSG00000105854	esophagus mucosa	2.16×10-5 (p-value)	GTEx Portal V6	1
ASB4	?	ENSG00000005981	ovary	1.24×10-6 (p-value)	GTEx Portal V6	1
ASB4	?	ENSG00000005981	subcutaneous adipocytes	2.18×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PON1	upstream gene variant	677	ENST00000222381	NM_000446.5	ENSP00000222381	2
PON1	upstream gene variant	908	ENST00000433729	?	ENSP00000407359	2

Locus 405 (rs6050091)

SNiPAcard

Block annotations

Block info

genomic range	chr20:24,788,418-24,897,155
block size	108,738 bp
variant count	60 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.308 [-4.928 – 0.939]	gene(s) hit or close-by	–
phastCons	μ = 0.066 [0 – 0.743]	eQTL gene(s)	ABHD12
GERP++	μ = -0.366 [-3.16 – 0.823]	potentially regulated gene(s)	ACSS1, CST7, PYGB
CADD score	μ = 3.159 [0.037 – 15.94]	disease gene(s)	ABHD12

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ABHD12	POLYNEUROPATHY, HEARING LOSS, ATAXIA, RETINITIS PIGMENTOSA, AND [...]	OMIM	MIM:612674
ABHD12	Polyneuropathy - hearing loss - ataxia - retinitis pigmentosa - cataract	OrphaNet	OrphaNet:171848

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABHD12	ENST00000465694	ILMN_1745116	monocyte	6.09×10-4 (p-value)	Fairfax et al.	1
ABHD12	ENST00000376542					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000291041	1	ENCP00000031107	CST7
ENCE00000291312	1	ENCP00000031129	PYGB
ENCE00000291043	1	ENCP00000031112	ACSS1
		ENCP00000031111	ACSS1
		ENCP00000031107	CST7

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000399317 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		Osteobl	H3K27me3
ENSR00000399318 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, DNase1
		A549	H3K4me3, H3K27me3

Locus 406 (rs11219769)

SNiPAcard

Block annotations

Block info

genomic range	chr11:124,619,407-124,620,147
block size	741 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.805 [-1.378 – -0.232]	gene(s) hit or close-by	ESAM, NRGN, RP11-677M14.2, VSIG2
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	MSANTD2, VSIG2
GERP++	μ = 0.020 [-2.81 – 2.85]	potentially regulated gene(s)	–
CADD score	μ = 6.803 [3.496 – 10.11]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
VSIG2	?	ENSG00000019102	transformed fibroblasts	9.20×10^{-6} (p-value)	GTEx Portal V6	2
MSANTD2	ENST00000526629	ILMN_1773148	monocyte	2.65×10^{-4} (p-value)	Fairfax et al.	1
MSANTD2	ENST00000533514					
MSANTD2	ENST00000374979					
MSANTD2	ENST00000239614					
MSANTD2	ENST00000524950					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000570678 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me1
		endothelium (HUVEC)	Cjun, H3K4me1, PolII, DNase1, Cmyc
		liver (HepG2)	H3K27me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ESAM	downstream gene variant	2948	ENST00000464067	?	?	2
ESAM	downstream gene variant	3994	ENST00000435477	?	ENSP00000415893	2
ESAM	downstream gene variant	2332	ENST00000444566	?	ENSP00000406689	2
ESAM	downstream gene variant	2872	ENST00000485116	?	?	2
ESAM	downstream gene variant	2872	ENST00000417453	?	ENSP00000389235	2
ESAM	downstream gene variant	2872	ENST00000278927	NM_138961.2	ENSP00000278927	2
NRGN	downstream gene variant	2301	ENST00000284292	NM_001126181.1, NM_006176.2	ENSP00000284292	2
NRGN	downstream gene variant	2305	ENST00000412681	?	ENSP00000399591	2

RP11-677M14.2	upstream gene variant	3174	ENST00000531241	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
VSIG2	ENST00000403470	?	ENSP00000385013	2
VSIG2	ENST00000326621	NM_014312.3	ENSP00000318684	2

Locus 407 (rs17200698)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,469,468-31,565,401
block size	95,934 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.126 [-0.147 – 0.22]	gene(s) hit or close-by	ATP6V1G2, ATP6V1G2-DDX39B, LST1, MCCD1, MICB, NCR3, NFKBIL1, PPIAP9, RPL15P4, XXbac-BPG16N22.5, Y_RNA
phastCons	μ = 0.077 [0 – 0.209]	eQTL gene(s)	XXbac-BPG248L24.12
GERP++	μ = -0.169 [-2.17 – 0.479]	potentially regulated gene(s)	–
CADD score	μ = 4.796 [2.556 – 11.31]	disease gene(s)	NFKBIL1, NCR3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
NFKBIL1	RHEUMATOID ARTHRITIS	OMIM	MIM:180300
NCR3	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	1.41×10^{-6} (p-value)	GTEx Portal V6	6
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	6.98×10^{-7} (p-value)	GTEx Portal V6	6
XXbac-BPG248L24.12	?	ENSG00000271581	blood	2.06×10^{-7} (p-value)	GTEx Portal V6	6

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487851 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	H3K27me3, Rad21
		liver (HepG2)	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00000487872 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		Osteobl	CTCF
		blood (K562)	DNase1, Max, CTCF
		blood (DND-41)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF

endothelium (HUVEC)	H3K36me3, CTCF, Cmyc
liver (HepG2)	CTCF
blood (GM12878)	DNase1, Rad21, CTCF
lung (IMR90)	CTCF
skin (NHEK)	CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ATP6V1G2	upstream gene variant	1714	ENST00000415099	?	ENSP00000390148	1
ATP6V1G2	upstream gene variant	3297	ENST00000303892	NM_130463.3	ENSP00000302194	1
ATP6V1G2	upstream gene variant	3504	ENST00000376151	NM_001204078.1	ENSP00000365321	1
ATP6V1G2	upstream gene variant	3293	ENST00000483251	NM_138282.2	ENSP00000419698	1
ATP6V1G2	upstream gene variant	3548	ENST00000481998	?	?	1
ATP6V1G2	upstream gene variant	3386	ENST00000483170	?	?	1
ATP6V1G2-DDX39B	upstream gene variant	3559	ENST00000376185	?	ENSP00000365356	1
ATP6V1G2-DDX39B	upstream gene variant	3533	ENST00000480131	?	ENSP00000420191	1
ATP6V1G2-DDX39B	upstream gene variant	3548	ENST00000475917	?	?	1
LST1	downstream gene variant	1641	ENST00000376110	?	ENSP00000365278	1
LST1	downstream gene variant	1489	ENST00000376090	?	ENSP00000365258	1
LST1	downstream gene variant	1489	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	downstream gene variant	1641	ENST00000460834	?	?	1
LST1	downstream gene variant	1489	ENST00000376089	?	ENSP00000365257	1
LST1	downstream gene variant	1588	ENST00000211921	?	ENSP00000211921	1
LST1	downstream gene variant	1489	ENST00000303757	NM_205840.2	ENSP00000303649	1
LST1	downstream gene variant	1722	ENST00000490742	?	ENSP00000418578	1
LST1	downstream gene variant	2668	ENST00000433492	?	ENSP00000399869	1
LST1	downstream gene variant	1772	ENST00000396112	?	ENSP00000379418	1
LST1	downstream gene variant	1489	ENST00000396101	?	ENSP00000379408	1
LST1	downstream gene variant	1489	ENST00000376096	?	ENSP00000365264	1
LST1	downstream gene variant	2557	ENST00000419073	?	?	1
LST1	downstream gene variant	1489	ENST00000418507	NM_205838.2	ENSP00000405900	1
LST1	downstream gene variant	1490	ENST00000376086	NM_001166538.1	ENSP00000365254	1
LST1	downstream gene variant	1489	ENST00000376099	?	ENSP00000365267	1
LST1	downstream gene variant	1641	ENST00000464044	?	ENSP00000417645	1
LST1	downstream gene variant	1489	ENST00000464526	?	?	1
LST1	downstream gene variant	1489	ENST00000376093	NM_007161.3	ENSP00000365261	1
LST1	downstream gene variant	1588	ENST00000339530	NM_205837.2	ENSP00000339201	1
LST1	downstream gene variant	1736	ENST00000376092	?	ENSP00000365260	1
MCCD1	upstream gene variant	4235	ENST00000376191	NM_001011700.2	ENSP00000365362	1
MICB	downstream gene variant	4800	ENST00000538442	NM_001289160.1	ENSP00000442345	1

MICB	upstream gene variant	4244	ENST00000494577	?	?	1
MICB	downstream gene variant	4799	ENST00000252229	NM_005931.4	ENSP00000252229	1
MICB	downstream gene variant	4799	ENST00000399150	NM_001289161.1	ENSP00000382103	1
NCR3	upstream gene variant	4639	ENST00000340027	NM_147130.2	ENSP00000342156	1
NCR3	upstream gene variant	4639	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	upstream gene variant	4665	ENST00000491161	?	?	1
NCR3	upstream gene variant	4639	ENST00000376072	NM_001145467.1	ENSP00000365240	1
NCR3	upstream gene variant	4875	ENST00000376071	?	ENSP00000365239	1
NCR3	upstream gene variant	4812	ENST00000495600	?	?	1
PPIAP9	downstream gene variant, upstream gene variant	3557	ENST00000403866	?	?	2
RPL15P4	upstream gene variant	3632	ENST00000416625	?	?	1
XXbac-BPG16N22.5	upstream gene variant	56	ENST00000538358	?	?	1
Y_RNA	upstream gene variant	4901	ENST00000383850	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MICB	ENST00000252229	NM_005931.4	ENSP00000252229	1
MICB	ENST00000538442	NM_001289160.1	ENSP00000442345	1
MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	1
NCR3	ENST00000376071	?	ENSP00000365239	1
NCR3	ENST00000495600	?	?	1
NCR3	ENST00000491161	?	?	1
NCR3	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	ENST00000376072	NM_001145467.1	ENSP00000365240	1
NCR3	ENST00000340027	NM_147130.2	ENSP00000342156	1
NFKBIL1	ENST00000473655	?	?	1
NFKBIL1	ENST00000376148	NM_005007.3	ENSP00000365318	1
NFKBIL1	ENST00000496233	?	ENSP00000437148	1
NFKBIL1	ENST00000376145	NM_001144961.1	ENSP00000365315	1
NFKBIL1	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	1

Locus 408 (rs3771167)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,910,447-103,080,056
block size	169,610 bp
variant count	32 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.298 [-2.21 – 0.904]	**gene(s) hit or close-by**	AC007248.7, AC007278.2, AC007278.3, IL18R1, IL18RAP, IL1RL1, MIR4772
phastCons	μ = 0.014 [0 – 0.072]	**eQTL gene(s)**	AC007278.3, IL18R1, IL1RL1
GERP++	μ = -0.950 [-7.42 – 2.39]	**potentially regulated gene(s)**	IL18R1, IL1R1, IL1RL1
CADD score	μ = 2.876 [0.38 – 8.592]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Serum protein levels (sST2)	$<2.00\times10^{-30}$	GWAS Catalog	23999434	1
1-oleoylglycerophosphoethanolamine	6.45×10^{-5}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL1RL1	?	ENSG00000115602	lung	2.76×10^{-6} (p-value)	GTEx Portal V6	18
IL18R1	?	ENSG00000115604	blood	3.61×10^{-7} (p-value)	GTEx Portal V6	13
AC007278.3	?	ENSG00000234389	blood	6.13×10^{-7} (p-value)	GTEx Portal V6	23
IL18R1	?	ENSG00000115604	thyroid	6.38×10^{-6} (p-value)	GTEx Portal V6	16
IL18R1	?	ENSG00000115604	subcutaneous adipocytes	2.17×10^{-7} (p-value)	GTEx Portal V6	26
?	?	ILMN_1679967	b-cell	7.64×10^{-4} (p-value)	Fairfax et al.	1
IL18R1	?	ENSG00000115604	skeletal muscle	2.71×10^{-6} (p-value)	GTEx Portal V6	10

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000027319		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000257360	1	ENCP00000027305	IL1R1

ENCE00000257426	1	ENCP00000027316	IL1RL1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000595650 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
		A549	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001655321 (enhancer)	2	monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K4me1, H3K27ac
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00000595653 (promoter)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	PolII, Ini1, Brg1, Gata2, Cmyc, DNase1
		skin (NHDF-AD)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	Cjun, DNase1, H3K4me2, H3K4me1, H3K27ac, PolII, CTCF
		liver (HepG2)	H3K27me3, DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me3, H3K4me2
		skin (NHEK)	DNase1, H3K27ac, H3K4me2
ENSR00000123377 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K27me3, Rad21, Jund, CTCF, Yy1, H3K4me2
		HSMMtube	DNase1, CTCF, H2AZ, H3K27me3
		blood (K562)	PU1, H3K9ac, Rad21, H2AZ, CTCF, DNase1, H3K4me3, Max
		skin (NHDF-AD)	CTCF, H3K4me3, H3K4me2
		muscle (HSMM)	CTCF, H2AZ
		liver (HepG2)	Rad21, H3K4me2, CTCF, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me2, H3K27me3, H3K4me3, CTCF
		blood (GM12878)	PU1, PolII, H2AZ, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2
		skin (NHEK)	CTCF, DNase1, H3K4me3, H3K9ac, H3K4me2
		NHLF	CTCF
		Osteobl	H3K4me3, H3K4me2, CTCF, H3K27me3
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, DNase1
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K27me3, H3K4me3
		endothelium (HUVEC)	DNase1, H3K4me3, H3K4me2, H3K9ac, PolII, CTCF
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF
ENSR00001655328 (enhancer)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K4me1
		monocytes (Monocytes-CD14+)	H3K4me1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC007248.7	downstream gene variant	3621	ENST00000430344	?	?	1
AC007278.2	downstream gene variant	2777	ENST00000436582	?	?	1
AC007278.3	upstream gene variant	596	ENST00000450893	?	?	1
IL18R1	upstream gene variant, downstream gene variant	1642	ENST00000466357	?	?	6
IL18R1	upstream gene variant	209	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	3
IL18R1	upstream gene variant	3305	ENST00000410040	?	ENSP00000386663	1

IL18R1	upstream gene variant	1582	ENST00000409599	?	ENSP00000387211	2
IL18RAP	upstream gene variant, downstream gene variant	3331	ENST00000497795	?	?	2
IL18RAP	upstream gene variant, downstream gene variant	450	ENST00000450855	?	ENSP00000389815	4
IL18RAP	upstream gene variant, downstream gene variant	606	ENST00000264260	NM_003853.2	ENSP00000264260	3
IL18RAP	upstream gene variant, downstream gene variant	691	ENST00000409369	?	ENSP00000387201	4
IL1RL1	upstream gene variant	164	ENST00000311734	NM_003856.3	ENSP00000310371	3
IL1RL1	upstream gene variant	274	ENST00000409584	?	ENSP00000386618	3
IL1RL1	upstream gene variant	297	ENST00000482701	?	?	3
IL1RL1	upstream gene variant, downstream gene variant	266	ENST00000463990	?	?	4
IL1RL1	upstream gene variant, downstream gene variant	2885	ENST00000447231	?	ENSP00000409437	2
IL1RL1	upstream gene variant	304	ENST00000427077	?	ENSP00000391120	3
IL1RL1	upstream gene variant, downstream gene variant	2180	ENST00000233954	NM_016232.4	ENSP00000233954	4
IL1RL1	upstream gene variant, downstream gene variant	1580	ENST00000473175	?	?	2
IL1RL1	upstream gene variant	3278	ENST00000404917	NM_001282408.1	ENSP00000384822	1
MIR4772	upstream gene variant	4768	ENST00000581495	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL18R1	ENST00000410040	?	ENSP00000386663	19
IL18R1	ENST00000334376	?	ENSP00000334030	7
IL18R1	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	8
IL18R1	ENST00000409599	?	ENSP00000387211	9
IL18RAP	ENST00000497795	?	?	1
IL18RAP	ENST00000450855	?	ENSP00000389815	1
IL18RAP	ENST00000409369	?	ENSP00000387201	3
IL18RAP	ENST00000264260	NM_003853.2	ENSP00000264260	4
IL1RL1	ENST00000447231	?	ENSP00000409437	7
IL1RL1	ENST00000482701	?	?	2
IL1RL1	ENST00000404917	NM_001282408.1	ENSP00000384822	8
IL1RL1	ENST00000463990	?	?	1
IL1RL1	ENST00000473175	?	?	7
IL1RL1	ENST00000427077	?	ENSP00000391120	2
IL1RL1	ENST00000409584	?	ENSP00000386618	2
IL1RL1	ENST00000233954	NM_016232.4	ENSP00000233954	8
IL1RL1	ENST00000311734	NM_003856.3	ENSP00000310371	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

IL18R1	ENST00000409599	?	ENSP00000387211	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
IL18R1	ENST00000466357	?	1

Locus 409 (rs3917254)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,776,518-102,776,518
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.364	gene(s) hit or close-by	IL1R1
phastCons	0.004	eQTL gene(s)	–
GERP++	0.235	potentially regulated gene(s)	–
CADD score	2.731	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL1R1	upstream gene variant	4716	ENST00000422532	?	ENSP00000390349	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1R1	ENST00000409929	NM_001288706.1	ENSP00000386776	1
IL1R1	ENST00000452403	?	ENSP00000401646	1
IL1R1	ENST00000450319	?	ENSP00000411627	1
IL1R1	ENST00000442590	?	ENSP00000393296	1
IL1R1	ENST00000409288	?	ENSP00000386478	1
IL1R1	ENST00000430171	?	ENSP00000408101	1
IL1R1	ENST00000424272	?	ENSP00000415366	1
IL1R1	ENST00000410023	NM_000877.3	ENSP00000386380	1
IL1R1	ENST00000409589	?	ENSP00000386555	1
IL1R1	ENST00000428279	?	ENSP00000410461	1
IL1R1	ENST00000409329	?	ENSP00000387131	1
IL1R1	ENST00000413623	?	ENSP00000407017	1

Locus 410 (rs17844003)

SNiPAcard

Block annotations

Block info

genomic range	chr13:46,670,347-46,670,347
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.476	gene(s) hit or close-by	CPB2, CPB2-AS1
phastCons	0.706	eQTL gene(s)	CPB2-AS1, ZC3H13
GERP++	0.449	potentially regulated gene(s)	–
CADD score	10.2	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CPB2-AS1	?	ENSG00000235903	caudate basal ganglia	3.11×10^{-9} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	cortex	5.83×10^{-8} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	nucleus accumbens	6.06×10^{-10} (p-value)	GTEx Portal V6	1
ZC3H13	?	ENSG00000123200	tibial nerve	2.55×10^{-6} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	tibial nerve	4.81×10^{-7} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	transformed fibroblasts	2.80×10^{-7} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	anterior cingulate cortex	9.99×10^{-8} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	cerebellar hemisphere	1.82×10^{-7} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	putamen	3.18×10^{-12} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	hypothalamus	2.78×10^{-8} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	cerebellum	1.33×10^{-7} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	frontal cortex	1.78×10^{-8} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	sun exposed skin	2.05×10^{-9} (p-value)	GTEx Portal V6	1
ZC3H13	?	ENSG00000123200	unexposed skin	4.94×10^{-6} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	hippocampus	2.41×10^{-8} (p-value)	GTEx Portal V6	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	1
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	1
CPB2-AS1	ENST00000606991	?	?	1

CPB2-AS1	ENST00000606243	?	?	1
CPB2-AS1	ENST00000415033	?	?	1
CPB2-AS1	ENST00000606351	?	?	1

Locus 411 (rs12585823)

SNiPAcard

Block annotations

Block info

genomic range	chr13:46,536,291-46,536,291
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.774	gene(s) hit or close-by	ZC3H13
phastCons	0.001	eQTL gene(s)	–
GERP++	2.91	potentially regulated gene(s)	–
CADD score	4.899	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ZC3H13	downstream gene variant	20	ENST00000282007	NM_015070.4	ENSP00000282007	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ZC3H13	ENST00000242848	?	ENSP00000242848	1

Locus 412 (rs12788387)

SNiPAcard

Block annotations

Block info

genomic range	chr11:57,851,041-57,913,286
block size	62,246 bp
variant count	46 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.103 [-2.019 – 1.61]	**gene(s) hit or close-by**	OR9I1, OR9I2P, OR9I3P, OR9L1P, OR9Q1
phastCons	μ = 0.090 [0 – 1]	**eQTL gene(s)**	MED19, RN7SL605P, TIMM10
GERP++	μ = -0.303 [-6.36 – 2.96]	**potentially regulated gene(s)**	LPXN, OR5B21, ZDHHC5
CADD score	μ = 4.517 [0.27 – 18.8]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MED19	?	ENSG00000156603	skeletal muscle	4.35×10^{-7} (p-value)	GTEx Portal V6	40
MED19	?	ENSG00000156603	atrial appendage	1.20×10^{-6} (p-value)	GTEx Portal V6	46
MED19	?	ENSG00000156603	transverse colon	3.46×10^{-6} (p-value)	GTEx Portal V6	5
MED19	?	ENSG00000156603	sun exposed skin	2.90×10^{-8} (p-value)	GTEx Portal V6	46
MED19	?	ENSG00000156603	esophagus mucosa	1.29×10^{-9} (p-value)	GTEx Portal V6	46
MED19	?	ENSG00000156603	subcutaneous adipocytes	5.37×10^{-6} (p-value)	GTEx Portal V6	12
MED19	?	ENSG00000156603	thyroid	1.93×10^{-8} (p-value)	GTEx Portal V6	45
TIMM10	ENST00000525587	ILMN_1765332	skin	6.29×10^{-6} (p-value)	MuTHER consortium	18
RN7SL605P	ENST00000577229		adipocyte	7.42×10^{-8} (p-value)	MuTHER consortium	18
TIMM10	ENST00000257245		blood	3.39×10^{-5} (p-value)	MuTHER consortium	18
TIMM10	ENST00000525587	ILMN_1765332	monocyte	7.68×10^{-18} (p-value)	Zeller et al.	5
RN7SL605P	ENST00000577229					
TIMM10	ENST00000257245					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000106527	1	ENCP00000012323	LPXN
		ENCP00000012278	ZDHHC5
		ENCP00000012322	OR5B21

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
OR9I1	downstream gene variant, upstream gene variant	1043	ENST00000302610	NM_001005211.1	ENSP00000302606	6
OR9I2P	upstream gene variant, downstream gene variant	37	ENST00000427941	?	?	12

OR9I3P	downstream gene variant, upstream gene variant	998	ENST00000608484	?	?	4
OR9L1P	upstream gene variant	4966	ENST00000529493	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
OR9Q1	ENST00000335397	NM_001005212.3	ENSP00000334934	46

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
OR9I2P	ENST00000427941	?	1
OR9I3P	ENST00000608484	?	1

Locus 413 (rs9332653)

SNiPAcard

Block annotations

Block info

genomic range	chr1:169,490,772-169,510,475
block size	19,704 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.045 [-0.268 – 0.251]	gene(s) hit or close-by	F5
phastCons	μ = 0.023 [0 – 0.069]	eQTL gene(s)	–
GERP++	μ = -0.358 [-1.42 – 0.198]	potentially regulated gene(s)	–
CADD score	μ = 1.928 [0.002 – 3.434]	disease gene(s)	F5

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
F5	STROKE, ISCHEMIC	OMIM	MIM:601367
F5	THROMBOPHILIA DUE TO ACTIVATED PROTEIN C RESISTANCE	OMIM	MIM:188055
F5	PREGNANCY LOSS, RECURRENT, SUSCEPTIBILITY TO, 1	OMIM	MIM:614389
F5	FACTOR V DEFICIENCY	OMIM	MIM:227400
F5	BUDD-CHIARI SYNDROME	OMIM	MIM:600880
F5	Congenital factor V deficiency	OrphaNet	OrphaNet:326
F5	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217
F5	BUDD-CHIARI SYNDROME	OrphaNet	OrphaNet:131
F5	East Texas bleeding disorder	OrphaNet	OrphaNet:391320

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
F5	missense variant	ENST00000367796	?	ENSP00000356770	I/L	Att/Ctt	?	?	1
F5	missense variant	ENST00000367797	NM_000130.4	ENSP00000356771	I/L	Att/Ctt	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
F5	ENST00000367796	?	ENSP00000356770	2
F5	ENST00000367797	NM_000130.4	ENSP00000356771	2

Locus 414 (rs2506028)

SNiPAcard

Block annotations

Block info

genomic range	chr10:43,435,628-43,447,847
block size	12,220 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.714 [-2.896 – 0.183]	gene(s) hit or close-by	–
phastCons	μ = 0.058 [0 – 0.377]	eQTL gene(s)	RET
GERP++	μ = -2.080 [-8.59 – 0.481]	potentially regulated gene(s)	AL022344.5, CCNYL2, RET, RET, RP11-124O11.1
CADD score	μ = 3.098 [0.535 – 14.03]	disease gene(s)	RET

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
RET	PHEOCHROMOCYTOMAPHEOCHROMOCYTOMA, SUSCEPTIBILITY TO	OMIM	MIM:171300
RET	CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL	OMIM	MIM:209880
RET	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIA	OMIM	MIM:171400
RET	THYROID CARCINOMA, FAMILIAL MEDULLARY	OMIM	MIM:155240
RET	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIB	OMIM	MIM:162300
RET	COLORECTAL CANCER	OMIM	MIM:114500
RET	HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, 1	OMIM	MIM:142623
RET	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550
RET	RENAL AGENESIS	DECIPHER	MIM:191830
RET	Multiple endocrine neoplasia IIB	DECIPHER	MIM:162300
RET	Unilateral renal dysplasia	OrphaNet	OrphaNet:93172
RET	Bilateral renal dysplasia	OrphaNet	OrphaNet:93173
RET	Haddad syndrome	OrphaNet	OrphaNet:99803
RET	Hirschsprung disease	OrphaNet	OrphaNet:388
RET	Multiple endocrine neoplasia type 2A	OrphaNet	OrphaNet:247698
RET	Papillary or follicular thyroid carcinoma	OrphaNet	OrphaNet:146
RET	Multiple endocrine neoplasia type 2B	OrphaNet	OrphaNet:247709
RET	Familial medullary thyroid carcinoma	OrphaNet	OrphaNet:99361
RET	Bilateral renal agenesis	OrphaNet	OrphaNet:1848

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RET	?	ENSG00000165731	unexposed skin	1.48×10-7 (p-value)	GTEx Portal V6	12

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000072563	1	ENCP00000008221	AL022344.5
		ENCP00000008225	RP11-124O11.1
		ENCP00000008213	CCNYL2
		ENCP00000008212	CCNYL2
		ENCP00000008227	RET RET

Locus 414 (sentinel SNP rs2506028 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 415 (rs4851016)

SNiPAcard

Block annotations

Block info

genomic range	chr2:103,098,191-103,120,889
block size	22,699 bp
variant count	56 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.463 [-5.522 – 2.646]	gene(s) hit or close-by	SLC9A4
phastCons	μ = 0.013 [0 – 0.34]	eQTL gene(s)	AC007278.2, AC007278.3, IL18R1, IL18RAP
GERP++	μ = -0.487 [-4.99 – 3.15]	potentially regulated gene(s)	SLC9A4, TMEM182
CADD score	μ = 2.505 [0.007 – 12.24]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AC007278.3	?	ENSG00000234389	lung	5.32×10^{-7} (p-value)	GTEx Portal V6	54
IL18R1	?	ENSG00000115604	sun exposed skin	6.92×10^{-6} (p-value)	GTEx Portal V6	18
AC007278.2	?	ENSG00000236525	blood	1.65×10^{-8} (p-value)	GTEx Portal V6	54
IL18RAP	?	ENSG00000115607	blood	1.07×10^{-14} (p-value)	GTEx Portal V6	54
AC007278.3	?	ENSG00000234389	blood	3.12×10^{-12} (p-value)	GTEx Portal V6	54
AC007278.3	?	ENSG00000234389	subcutaneous adipocytes	4.21×10^{-6} (p-value)	GTEx Portal V6	33

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000027326		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000257544	2	ENCP00000027332	TMEM182

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001544967 (promoter flanking region)	3	NHLF	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3

	nervous (NH-A)	DNase1
	A549	H3K27me3
	skin (NHEK)	DNase1, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SLC9A4	upstream gene variant	872	ENST00000492154	?	?	28

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLC9A4	ENST00000295269	NM_001011552.3	ENSP00000295269	56
SLC9A4	ENST00000492154	?	?	7

Locus 416 (rs870825)

SNiPAcard

Block annotations

Block info

genomic range	chr4:185,571,123-185,608,591
block size	37,469 bp
variant count	89 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.622 [-3.857 – 1.623]	gene(s) hit or close-by	CASP3, PRIMPOL
phastCons	μ = 0.027 [0 – 0.937]	eQTL gene(s)	CASP3, CENPU, PRIMPOL
GERP++	μ = -0.734 [-7.23 – 3.23]	potentially regulated gene(s)	ACSL1, IRF2, PRIMPOL, RP11-162O12.2, RP11-242J7.1, RP11-386B13.3, RP11-701P16.5
CADD score	μ = 3.236 [0.009 – 10.91]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PRIMPOL	missense variant	ENST00000508001	?	ENSP00000424639	N/S	aAt/aGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PRIMPOL	?	ENSG00000164306	tibial nerve	2.48×10-8 (p-value)	GTEx Portal V6	72
CENPU	?	ENSG00000151725	tibial nerve	1.81×10-7 (p-value)	GTEx Portal V6	6
PRIMPOL	?	ENSG00000164306	transformed fibroblasts	2.85×10-6 (p-value)	GTEx Portal V6	25
CENPU	?	ENSG00000151725	transformed fibroblasts	2.12×10-7 (p-value)	GTEx Portal V6	3
CENPU	?	ENSG00000151725	tibial artery	2.13×10-9 (p-value)	GTEx Portal V6	81
PRIMPOL	?	ENSG00000164306	breast	2.48×10-8 (p-value)	GTEx Portal V6	71
CASP3	?	ENSG00000164305	blood	1.80×10-8 (p-value)	GTEx Portal V6	51
PRIMPOL	?	ENSG00000164306	thyroid	3.10×10-7 (p-value)	GTEx Portal V6	28
CENPU	?	ENSG00000151725	thyroid	2.30×10-8 (p-value)	GTEx Portal V6	73
CENPU	?	ENSG00000151725	esophagus mucosa	4.40×10-15 (p-value)	GTEx Portal V6	86
PRIMPOL	?	ENSG00000164306	skeletal muscle	1.04×10-8 (p-value)	GTEx Portal V6	75
CENPU	?	ENSG00000151725	skeletal muscle	3.37×10-6 (p-value)	GTEx Portal V6	16
PRIMPOL	?	ENSG00000164306	unexposed skin	6.96×10-9 (p-value)	GTEx Portal V6	74
CENPU	?	ENSG00000151725	unexposed skin	2.46×10-9 (p-value)	GTEx Portal V6	7
PRIMPOL	?	ENSG00000164306	sun exposed skin	2.24×10-6 (p-value)	GTEx Portal V6	25

CENPU	?	ENSG00000151725	sun exposed skin	9.04×10-13 (p-value)	GTEx Portal V6	86
PRIMPOL	?	ENSG00000164306	subcutaneous adipocytes	6.42×10-7 (p-value)	GTEx Portal V6	44
CENPU	?	ENSG00000151725	subcutaneous adipocytes	2.51×10-8 (p-value)	GTEx Portal V6	77
CENPU	ENST00000502461	ILMN_1679438	skin	3.40×10-9 (p-value)	MuTHER consortium	11
CENPU	ENST00000281453					
CASP3	ENST00000393585	ILMN_1731639	blood	1.19×10-7 (p-value)	MuTHER consortium	11
CASP3	ENST00000517513					
CASP3	ENST00000308394					
CASP3	ENST00000613118					
CENPU	?	ENSG00000151725	lung	3.92×10-7 (p-value)	GTEx Portal V6	7
CENPU	ENST00000502461	ILMN_1679438	blood	1.92×10-5 (p-value)	Westra et al.	11
CENPU	ENST00000281453					
CENPU	?	ENSG00000151725	testis	3.50×10-11 (p-value)	GTEx Portal V6	74
CENPU	?	ENSG00000151725	blood	4.39×10-3 (q-value)	SeeQTL DB (HapMap)	3
CASP3	ENST00000393585	ILMN_1731639	monocyte	2.71×10-17 (p-value)	Zeller et al.	6
CASP3	ENST00000517513					
CASP3	ENST00000308394					
CASP3	ENST00000613118					
CENPU	?	ENSG00000151725	EBV lymphocytes	3.08×10-7 (p-value)	GTEx Portal V6	4
CENPU	?	ENSG00000151725	visceral adipocytes	1.55×10-6 (p-value)	GTEx Portal V6	1
CENPU	?	ENSG00000151725	atrial appendage	1.10×10-6 (p-value)	GTEx Portal V6	2
CENPU	?	ENSG00000151725	transverse colon	1.33×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000640933	1	ENST00000515774, ENST00000314970, ENST00000509002, ENST00000503752, ENST00000512834	PRIMPOL

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000398855	1	ENCP00000043126	RP11-242J7.1
ENCE00000398670	2	ENCP00000043153	RP11-386B13.3
		ENCP00000043119	RP11-162O12.2
		ENCP00000043145	ACSL1
		ENCP00000043138	ACSL1
		ENCP00000043147	RP11-701P16.5
		ENCP00000043124	IRF2
		ENCP00000043144	ACSL1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001690770	1	embryonic stem cell (H1ESC)	DNase1, CTCF, PolII, Rad21, TAF7, H3K27ac, TAF1, H3K4me2, H3K9ac, H3K4me3
(promoter)		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, DNase1
		blood (K562)	Egr1, H3K27ac, Max, Rad21, H3K9ac, HEY1, H2AZ, TAF1, PolII, H3K4me2, CTCF, DNase1,

			H3K4me3
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	Gabp, PolII, TAF1, ELF1, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	CTCF, H3K9ac, H3K4me3, H3K4me2, H3K27ac, DNase1
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, Egr1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me3, H3K4me2, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K27ac, H3K9ac, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00001438105 (promoter flanking region)	4	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	DNase1
		A549	H3K36me3
ENSR00001438106 (enhancer)	1	endothelium (HUVEC)	H3K36me3
		Osteobl	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	DNase1
ENSR00001690773 (enhancer)	5	NHLF	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001255743 (promoter flanking region)	2	monocytes (Monocytes-CD14+)	H3K27ac, DNase1
		blood (K562)	Ini1, Brg1, Gata2, DNase1, Egr1, H3K27ac, H3K4me3, PolII, H3K4me2
		blood (DND-41)	H3K36me3
		nervous (NH-A)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PRIMPOL	2	ENST00000314970 ENST00000503752 ENST00000506278 ENST00000508001 ENST00000509002 ENST00000512834 ENST00000515152 ENST00000515774	hsa-miR-134-5p hsa-miR-302a-3p hsa-miR-302b-3p hsa-miR-302c-3p hsa-miR-302d-3p hsa-miR-329-3p hsa-miR-362-3p hsa-miR-372-3p hsa-miR-373-3p hsa-miR-494-3p hsa-miR-520a-3p hsa-miR-520b hsa-miR-520c-3p hsa-miR-520d-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CASP3	upstream gene variant	522	ENST00000447121	?	ENSP00000407142	6
CASP3	upstream gene variant	514	ENST00000393588	?	ENSP00000377213	6
CASP3	upstream gene variant	522	ENST00000517513	?	ENSP00000428372	6
CASP3	upstream gene variant	460	ENST00000523916	NM_032991.2	ENSP00000428929	6
CASP3	upstream gene variant	494	ENST00000308394	NM_004346.3	ENSP00000311032	6
CASP3	upstream gene variant	1377	ENST00000393585	?	ENSP00000377210	3
CASP3	upstream gene variant	1377	ENST00000613118	?	ENSP00000478339	3
PRIMPOL	downstream gene variant	278	ENST00000506278	?	ENSP00000423409	19
PRIMPOL	downstream gene variant	277	ENST00000515152	?	ENSP00000422199	9
PRIMPOL	downstream gene variant	980	ENST00000509538	?	?	2
PRIMPOL	upstream gene variant	506	ENST00000508001	?	ENSP00000424639	10
PRIMPOL	upstream gene variant	3931	ENST00000512658	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
PRIMPOL	ENST00000512834	NM_001300768.1	ENSP00000425316	Q	caA/caG	1
PRIMPOL	ENST00000314970	NM_152683.2	ENSP00000313816	Q	caA/caG	1
PRIMPOL	ENST00000503752	?	ENSP00000420860	Q	caA/caG	1
PRIMPOL	ENST00000515774	NM_001300767.1	ENSP00000421913	Q	caA/caG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PRIMPOL	ENST00000509002	?	ENSP00000423353	89
PRIMPOL	ENST00000512834	NM_001300768.1	ENSP00000425316	89
PRIMPOL	ENST00000314970	NM_152683.2	ENSP00000313816	89
PRIMPOL	ENST00000503752	?	ENSP00000420860	89
PRIMPOL	ENST00000515774	NM_001300767.1	ENSP00000421913	89
PRIMPOL	ENST00000506278	?	ENSP00000423409	62
PRIMPOL	ENST00000515152	?	ENSP00000422199	48
PRIMPOL	ENST00000509538	?	?	10
PRIMPOL	ENST00000508001	?	ENSP00000424639	27

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PRIMPOL	ENST00000506278	?	ENSP00000423409	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PRIMPOL	ENST00000509538	?	1

Locus 416 (sentinel SNP rs870825 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 417 (rs16873418)

SNiPAcard

Block annotations

Block info

genomic range	chr8:106,578,977-106,593,207
block size	14,231 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.260 [-0.31 – 0.853]	gene(s) hit or close-by	ZFPM2
phastCons	μ = 0.110 [0 – 0.982]	eQTL gene(s)	CXCL5
GERP++	μ = 0.121 [-1.7 – 3.49]	potentially regulated gene(s)	–
CADD score	μ = 5.068 [1.506 – 9.561]	disease gene(s)	ZFPM2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Platelet counts	$<4.00\times10^{-17}$	GWAS Catalog	22139419	1
Vascular endothelial growth factor levels	$<5.00\times10^{-23}$	GWAS Catalog	21757650	1

Disease gene annotation

gene	trait	source DB	source entry/link
ZFPM2	CONOTRUNCAL HEART MALFORMATIONS	OMIM	MIM:217095
ZFPM2	46,XY SEX REVERSAL 9	OMIM	MIM:616067
ZFPM2	TETRALOGY OF FALLOT	OMIM	MIM:187500
ZFPM2	DIAPHRAGMATIC HERNIA 3	OMIM	MIM:610187
ZFPM2	TETRALOGY OF FALLOT	DECIPHER	MIM:187500
ZFPM2	DIAPHRAGMATIC HERNIA 3	DECIPHER	MIM:610187
ZFPM2	Congenital diaphragmatic hernia	OrphaNet	OrphaNet:2140
ZFPM2	TETRALOGY OF FALLOT	OrphaNet	OrphaNet:3303

Direct effect on regulation

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CXCL5	ENST00000296027	ILMN_2171384	chr4	blood	1.28×10^{-7} (p-value)	Westra et al.	1
?	?	ILMN_1884950	chr7	blood	1.23×10^{-7} (p-value)	Westra et al.	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001398577 (enhancer)	1	lung (IMR90)	H3K36me3
		blood (K562)	DNase1
ENSR00001398582 (promoter flanking region)	1	lung (IMR90)	H3K4me2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ZFPM2	ENST00000517361	?	ENSP00000428720	9
ZFPM2	ENST00000511341	?	?	9
ZFPM2	ENST00000520492	?	ENSP00000430757	9
ZFPM2	ENST00000520027	?	ENSP00000428149	9
ZFPM2	ENST00000407775	NM_012082.3	ENSP00000384179	9

Locus 418 (rs11072996)

SNiPAcard

Block annotations

Block info

genomic range	chr15:81,529,410-81,529,410
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.006	gene(s) hit or close-by	IL16
phastCons	0	eQTL gene(s)	–
GERP++	-1.53	potentially regulated gene(s)	–
CADD score	3.993	disease gene(s)	–

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL16	ENST00000560241	?	ENSP00000452738	1
IL16	ENST00000394660	NM_001172128.1	ENSP00000378155	1
IL16	ENST00000302987	NM_172217.3	ENSP00000302935	1
IL16	ENST00000559383	?	ENSP00000453250	1
IL16	ENST00000360547	?	ENSP00000456972	1

Locus 419 (rs1046668)

SNiPAcard

Block annotations

Block info

genomic range	chr2:152,206,464-152,239,059
block size	32,596 bp
variant count	35 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.613 [-3.041 – 1.071]	gene(s) hit or close-by	MIR4773-2, RN7SL124P, TNFAIP6
phastCons	μ = 0.086 [0 – 0.997]	eQTL gene(s)	NMI, RBM43
GERP++	μ = -0.363 [-4.58 – 3.46]	potentially regulated gene(s)	TNFAIP6
CADD score	μ = 4.274 [0.025 – 14.86]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
TNFAIP6	missense variant	ENST00000243347	NM_007115.3	ENSP00000243347	R/Q	cGa/cAa	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RBM43	ENST00000331426	ILMN_1858599	blood	6.08×10^{-6} (p-value)	Westra et al.	8
NMI	ENST00000243346	ILMN_1739541	blood	7.25×10^{-5} (p-value)	Westra et al.	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000505495	1	ENST00000243347, ENST00000460812	TNFAIP6

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001547521 (promoter flanking region)	2	NHLF	DNase1, H3K27ac
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	PolII
		monocytes (Monocytes-CD14+)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00000304445 (enhancer)	2	Osteobl	H3K27ac
		skin (NHDF-AD)	DNase1

ENSR00000600272 (promoter flanking region)	4	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27ac, H3K4me2
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	DNase1
		blood (GM12878)	BATF
		A549	DNase1
		nervous (NH-A)	DNase1
ENSR00001657792 (enhancer)	2	muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MIR4773-2	upstream gene variant, downstream gene variant	244	ENST00000585225	?	?	11
RN7SL124P	upstream gene variant, downstream gene variant	119	ENST00000498656	?	?	16
TNFAIP6	upstream gene variant, downstream gene variant	1707	ENST00000243347	NM_007115.3	ENSP00000243347	5
TNFAIP6	upstream gene variant, downstream gene variant	691	ENST00000460812	?	?	12

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TNFAIP6	ENST00000243347	NM_007115.3	ENSP00000243347	26
TNFAIP6	ENST00000460812	?	?	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TNFAIP6	ENST00000243347	NM_007115.3	ENSP00000243347	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TNFAIP6	ENST00000460812	?	1

Locus 420 (rs2493151)

SNiPAcard

Block annotations

Block info

genomic range	chr1:230,873,488-230,879,511
block size	6,024 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.444 [-2.133 – -0.493]	**gene(s) hit or close-by**	CAPN9, RP11-99J16__A.2
phastCons	μ = 0.000 [0 – 0.001]	**eQTL gene(s)**	–
GERP++	μ = -1.241 [-2.88 – 0.756]	**potentially regulated gene(s)**	–
CADD score	μ = 1.572 [0.279 – 3.222]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000554770 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	Rad21
		liver (HepG2)	Rad21
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CAPN9	upstream gene variant	3619	ENST00000271971	NM_006615.2	ENSP00000271971	2
CAPN9	upstream gene variant	3619	ENST00000366666	?	ENSP00000355626	2
CAPN9	upstream gene variant	3650	ENST00000354537	NM_016452.1	ENSP00000346538	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-99J16__A.2	ENST00000412344	?	?	3

Locus 421 (rs5994176)

SNiPAcard

Block annotations

Block info

genomic range	chr22:17,616,715-17,618,164
block size	1,450 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.697 [-1.53 – -0.194]	**gene(s) hit or close-by**	AC006946.15, CECR5
phastCons	μ = 0.002 [0 – 0.005]	**eQTL gene(s)**	CECR6
GERP++	μ = -0.386 [-2.83 – 1.56]	**potentially regulated gene(s)**	–
CADD score	μ = 4.606 [3.14 – 5.743]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CECR6	?	ENSG00000183307	sun exposed skin	4.38×10^{-8} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC006946.15	downstream gene variant	3721	ENST00000441544	?	?	1
CECR5	downstream gene variant	237	ENST00000155674	NM_017829.5	ENSP00000155674	3
CECR5	downstream gene variant	248	ENST00000477157	?	?	3
CECR5	downstream gene variant	308	ENST00000486462	?	?	3
CECR5	downstream gene variant	305	ENST00000399852	?	ENSP00000382745	3
CECR5	downstream gene variant	246	ENST00000336737	NM_033070.2	ENSP00000337358	3

Locus 422 (rs668555)

SNiPAcard

Block annotations

Block info

genomic range	chr1:57,412,848-57,428,589
block size	15,742 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.766 [-1.388 – 0.056]	gene(s) hit or close-by	AL161740.1, C8B, RP5-1103B4.3
phastCons	μ = 0.001 [0 – 0.003]	eQTL gene(s)	–
GERP++	μ = -2.208 [-4.95 – 1.85]	potentially regulated gene(s)	–
CADD score	μ = 2.995 [0.285 – 6.988]	disease gene(s)	C8B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C8B	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL161740.1	upstream gene variant	3364	ENST00000408664	?	?	1
C8B	downstream gene variant	2805	ENST00000494324	?	?	1
RP5-1103B4.3	upstream gene variant	970	ENST00000417420	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C8B	ENST00000468990	?	?	3
C8B	ENST00000543257	NM_001278543.1	ENSP00000442548	6
C8B	ENST00000535057	NM_001278544.1	ENSP00000440113	6
C8B	ENST00000371237	NM_000066.3	ENSP00000360281	6
C8B	ENST00000494324	?	?	2

Locus 423 (rs9368624)

SNiPAcard

Block annotations

Block info

genomic range	chr6:30,080,036-30,194,873
block size	114,838 bp
variant count	30 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.388 [-2.652 – 1.382]	gene(s) hit or close-by	PAIP1P1, TRIM10, TRIM15, TRIM26, TRIM31, TRIM31-AS1, TRIM40
phastCons	μ = 0.053 [0 – 0.997]	eQTL gene(s)	HCG18, HLA-G, HLA-H, HLA-K, HLA-L, HLA-T, HLA-U, HLA-V, HLA-W, MICA, PPP1R11, TRIM10, TRIM15
GERP++	μ = -1.347 [-6.82 – 2.41]	potentially regulated gene(s)	PTMAP1, PTMAP1, PTMAP1, PTMAP1, PTMAP1, PTMAP1, PTMAP1, TRIM10, TRIM10, TRIM10, TRIM10, TRIM10, TRIM10, TRIM10, TRIM15, TRIM15, TRIM15, TRIM15, TRIM15, TRIM15, TRIM15, TRIM15, TRIM39, TRIM39, TRIM39, TRIM39, TRIM39, TRIM39, TRIM40, TRIM40, TRIM40, TRIM40, TRIM40, TRIM40, TRIM40
CADD score	μ = 3.632 [0.104 – 9.267]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
TRIM15	missense variant	ENST00000376688	?	ENSP00000365878	V/I	Gtt/Att	?	?	1
TRIM15	missense variant	ENST00000376694	NM_033229.2	ENSP00000365884	V/I	Gtt/Att	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HLA-U	?	ENSG00000228078	atrial appendage	7.47×10^{-14} (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	atrial appendage	5.23×10^{-9} (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	atrial appendage	2.24×10^{-12} (p-value)	GTEx Portal V6	29
HLA-W	?	ENSG00000235290	transformed fibroblasts	2.51×10^{-9} (p-value)	GTEx Portal V6	29
HCG18	?	ENSG00000231074	transformed fibroblasts	1.30×10^{-7} (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	transformed fibroblasts	1.35×10^{-25} (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	transformed fibroblasts	1.20×10^{-9} (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	transformed fibroblasts	4.73×10^{-21} (p-value)	GTEx Portal V6	29
HLA-W	?	ENSG00000235290	blood	9.09×10^{-13} (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	blood	3.43×10^{-36} (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	blood	7.62×10^{-12} (p-value)	GTEx Portal V6	29
HLA-T	?	ENSG00000231130	blood	6.27×10^{-13} (p-value)	GTEx Portal V6	29

HLA-H	?	ENSG00000206341	blood	3.90×10-15 (p-value)	GTEx Portal V6	29
TRIM15	?	ENSG00000204610	blood	1.28×10-7 (p-value)	GTEx Portal V6	12
HLA-W	?	ENSG00000235290	breast	4.28×10-7 (p-value)	GTEx Portal V6	26
HLA-U	?	ENSG00000228078	breast	1.16×10-19 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	breast	1.69×10-8 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	breast	1.60×10-12 (p-value)	GTEx Portal V6	29
MICA	ENST00000449934	ILMN_3241692	monocyte	4.23×10-5 (p-value)	Fairfax et al.	6
MICA	ENST00000616296					
MICA	ENST00000421350					
HLA-W	?	ENSG00000235290	skeletal muscle	6.79×10-6 (p-value)	GTEx Portal V6	23
HLA-U	?	ENSG00000228078	skeletal muscle	1.37×10-31 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	skeletal muscle	3.62×10-12 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	skeletal muscle	1.76×10-25 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	putamen	1.39×10-7 (p-value)	GTEx Portal V6	27
HLA-U	?	ENSG00000228078	putamen	1.37×10-6 (p-value)	GTEx Portal V6	3
HLA-U	?	ENSG00000228078	EBV lymphocytes	7.71×10-15 (p-value)	GTEx Portal V6	29
HLA-T	?	ENSG00000231130	EBV lymphocytes	8.40×10-8 (p-value)	GTEx Portal V6	27
HLA-H	?	ENSG00000206341	EBV lymphocytes	1.67×10-9 (p-value)	GTEx Portal V6	29
HLA-W	?	ENSG00000235290	sun exposed skin	3.07×10-10 (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	sun exposed skin	1.12×10-23 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	sun exposed skin	2.44×10-9 (p-value)	GTEx Portal V6	29
HLA-T	?	ENSG00000231130	sun exposed skin	8.52×10-9 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	sun exposed skin	5.20×10-15 (p-value)	GTEx Portal V6	29
HLA-W	?	ENSG00000235290	unexposed skin	6.58×10-7 (p-value)	GTEx Portal V6	15
HLA-U	?	ENSG00000228078	unexposed skin	5.38×10-22 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	unexposed skin	6.77×10-10 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	unexposed skin	4.07×10-12 (p-value)	GTEx Portal V6	29
HLA-W	?	ENSG00000235290	aorta	1.14×10-5 (p-value)	GTEx Portal V6	14
HLA-U	?	ENSG00000228078	aorta	5.14×10-17 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	aorta	4.69×10-8 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	aorta	6.58×10-17 (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	left ventricle	3.72×10-17 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	left ventricle	1.11×10-6 (p-value)	GTEx Portal V6	16
HLA-H	?	ENSG00000206341	left ventricle	1.28×10-11 (p-value)	GTEx Portal V6	29
HLA-W	?	ENSG00000235290	subcutaneous adipocytes	6.36×10-7 (p-value)	GTEx Portal V6	26
HLA-U	?	ENSG00000228078	subcutaneous adipocytes	3.17×10-24 (p-value)	GTEx Portal V6	29
	?		subcutaneous adipocytes			16
HLA-G		ENSG00000204632		2.05×10-6 (p-value)	GTEx Portal V6	
HLA-T	?	ENSG00000231130	subcutaneous adipocytes	5.56×10-8 (p-value)	GTEx Portal V6	27

HLA-H	?	ENSG00000206341	subcutaneous adipocytes	5.09×10^{-17} (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	caudate basal ganglia	3.57×10^{-10} (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	caudate basal ganglia	4.13×10^{-7} (p-value)	GTEx Portal V6	22
HLA-H	?	ENSG00000206341	caudate basal ganglia	1.56×10^{-7} (p-value)	GTEx Portal V6	15
HLA-W	?	ENSG00000235290	visceral adipocytes	4.87×10^{-10} (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	visceral adipocytes	2.42×10^{-17} (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	visceral adipocytes	7.92×10^{-8} (p-value)	GTEx Portal V6	27
HLA-H	?	ENSG00000206341	visceral adipocytes	2.82×10^{-14} (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	stomach	7.24×10^{-12} (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	stomach	3.99×10^{-9} (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	tibial nerve	1.22×10^{-20} (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	tibial nerve	1.47×10^{-7} (p-value)	GTEx Portal V6	27
HLA-H	?	ENSG00000206341	tibial nerve	3.79×10^{-11} (p-value)	GTEx Portal V6	29
HLA-W	?	ENSG00000235290	esophagus mucosa	1.99×10^{-6} (p-value)	GTEx Portal V6	16
HLA-U	?	ENSG00000228078	esophagus mucosa	4.81×10^{-17} (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	esophagus mucosa	4.58×10^{-7} (p-value)	GTEx Portal V6	28
HLA-H	?	ENSG00000206341	esophagus mucosa	6.18×10^{-13} (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	pituitary	1.20×10^{-9} (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	pituitary	6.01×10^{-9} (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	testis	3.08×10^{-12} (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	testis	9.27×10^{-9} (p-value)	GTEx Portal V6	29
HLA-K	?	ENSG00000230795	testis	3.36×10^{-6} (p-value)	GTEx Portal V6	24
HLA-W	?	ENSG00000235290	testis	1.25×10^{-5} (p-value)	GTEx Portal V6	8
HLA-U	?	ENSG00000228078	hypothalamus	7.20×10^{-9} (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	hypothalamus	4.05×10^{-7} (p-value)	GTEx Portal V6	22
HLA-H	?	ENSG00000206341	hypothalamus	9.97×10^{-7} (p-value)	GTEx Portal V6	12
HLA-U	?	ENSG00000228078	frontal cortex	3.98×10^{-11} (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	frontal cortex	2.43×10^{-6} (p-value)	GTEx Portal V6	12
HLA-H	?	ENSG00000206341	frontal cortex	7.72×10^{-7} (p-value)	GTEx Portal V6	8
HLA-U	?	ENSG00000228078	pancreas	8.06×10^{-8} (p-value)	GTEx Portal V6	27
HLA-U	?	ENSG00000228078	muscularis mucosae	1.19×10^{-17} (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	muscularis mucosae	6.55×10^{-18} (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	lung	8.11×10^{-19} (p-value)	GTEx Portal V6	29
HLA-V	?	ENSG00000181126	lung	2.79×10^{-6} (p-value)	GTEx Portal V6	27
HLA-W	?	ENSG00000235290	lung	2.23×10^{-10} (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	lung	3.29×10^{-26} (p-value)	GTEx Portal V6	29
HLA-T	?	ENSG00000231130	lung	9.14×10^{-14} (p-value)	GTEx Portal V6	29

HLA-T	?	ENSG00000231130	lung	9.14×10-14 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	lung	1.36×10-11 (p-value)	GTEx Portal V6	29
HLA-K	?	ENSG00000230795	lung	1.04×10-6 (p-value)	GTEx Portal V6	27
HLA-W	?	ENSG00000235290	tibial artery	7.97×10-7 (p-value)	GTEx Portal V6	27
HLA-U	?	ENSG00000228078	tibial artery	6.96×10-25 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	tibial artery	3.39×10-8 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	tibial artery	3.22×10-19 (p-value)	GTEx Portal V6	29
HLA-W	?	ENSG00000235290	thyroid	9.36×10-8 (p-value)	GTEx Portal V6	27
HLA-U	?	ENSG00000228078	thyroid	1.25×10-21 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	thyroid	9.21×10-11 (p-value)	GTEx Portal V6	29
HLA-T	?	ENSG00000231130	thyroid	7.54×10-6 (p-value)	GTEx Portal V6	23
HLA-H	?	ENSG00000206341	thyroid	9.90×10-19 (p-value)	GTEx Portal V6	29
HLA-L	?	ENSG00000243753	thyroid	6.64×10-7 (p-value)	GTEx Portal V6	12
HLA-U	?	ENSG00000228078	prostate	1.33×10-8 (p-value)	GTEx Portal V6	29
HLA-W	?	ENSG00000235290	transverse colon	3.42×10-7 (p-value)	GTEx Portal V6	27
HLA-U	?	ENSG00000228078	transverse colon	1.22×10-12 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	transverse colon	2.41×10-6 (p-value)	GTEx Portal V6	15
HLA-H	?	ENSG00000206341	transverse colon	2.69×10-10 (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	terminal ileum	6.38×10-10 (p-value)	GTEx Portal V6	25
HLA-H	?	ENSG00000206341	terminal ileum	3.45×10-8 (p-value)	GTEx Portal V6	15
HLA-U	?	ENSG00000228078	nucleus accumbens	3.84×10-9 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	nucleus accumbens	4.30×10-7 (p-value)	GTEx Portal V6	22
HLA-U	?	ENSG00000228078	spleen	1.32×10-7 (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	sigmoid colon	1.10×10-12 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	sigmoid colon	3.91×10-10 (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	coronary artery	3.14×10-10 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	coronary artery	9.05×10-9 (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	gastroesophageal junction	4.37×10-12 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	gastroesophageal junction	6.45×10-11 (p-value)	GTEx Portal V6	29
PPP1R11	ENST00000376765	ILMN_1747598	blood	2.51×10-6 (p-value)	Westra et al.	9
PPP1R11	ENST00000376769					
PPP1R11	ENST00000376763					
PPP1R11	ENST00000376773					
PPP1R11	ENST00000376758					
PPP1R11	ENST00000376772					
HLA-L	ENST00000463348	ILMN_1675282	blood	5.75×10-7 (p-value)	Westra et al.	4
HLA-W	?	ENSG00000235290	adrenal gland	4.02×10-7 (p-value)	GTEx Portal V6	22
HLA-U	?	ENSG00000228078	adrenal gland	6.10×10-11 (p-value)	GTEx Portal V6	29
HLA-H	?	ENSG00000206341	adrenal gland	8.14×10-10 (p-value)	GTEx Portal V6	29

HLA-U	?	ENSG00000228078	cerebellar hemisphere	4.61×10-8 (p-value)	GTEx Portal V6	29
HLA-G	?	ENSG00000204632	cerebellar hemisphere	6.29×10-7 (p-value)	GTEx Portal V6	27
HLA-U	?	ENSG00000228078	cerebellum	5.06×10-9 (p-value)	GTEx Portal V6	29
HLA-U	?	ENSG00000228078	hippocampus	1.99×10-8 (p-value)	GTEx Portal V6	26
HLA-H	?	ENSG00000206341	hippocampus	1.19×10-6 (p-value)	GTEx Portal V6	2
HLA-U	?	ENSG00000228078	cortex	5.39×10-7 (p-value)	GTEx Portal V6	12
?	?	240182_at	blood	8.00×10-10 (p-value)	Dixon et al.	1
HLA-U	?	ENSG00000228078	liver	1.35×10-6 (p-value)	GTEx Portal V6	2
TRIM10	ENST00000376704	ILMN_1746788	skin	2.24×10-5 (p-value)	MuTHER consortium	1
TRIM10	ENST00000449742					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000735626	1	ENST00000376688, ENST00000376694	TRIM15
FFCP00000735631	1	ENST00000376688, ENST00000376694	TRIM15

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444263	1	ENCP00000047883	TRIM10 TRIM10 TRIM10 TRIM10 TRIM10 TRIM10 TRIM10 TRIM10
		ENCP00000047882	TRIM40 TRIM40 TRIM40 TRIM40 TRIM40 TRIM40 TRIM40 TRIM40
		ENCP00000047922	PTMAP1 PTMAP1 PTMAP1 PTMAP1 PTMAP1 PTMAP1 PTMAP1
		ENCP00000047897	TRIM39 TRIM39 TRIM39 TRIM39 TRIM39 TRIM39 TRIM39
		ENCP00000047884	TRIM15 TRIM15 TRIM15 TRIM15 TRIM15

TRIM15
TRIM15
TRIM15

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001702952 (enhancer)	2	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me1, H3K4me2
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00000487613 (promoter)	6	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1, H3K27me3
		blood (K562)	H3K27me3, Egr1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	DNase1, H3K27ac, Cmyc, H3K4me3, H3K27me3, Gabp, PolII, H3K79me2, TAF1, H2AZ, H3K4me2, HNF4A, H3K9ac
		lung (IMR90)	H3K27me3
		A549	H3K4me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K27me3
ENSR00001032837 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		blood (K562)	H3K27me3, PU1
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	PU1, Max, IRF4, H3K79me2, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001702964 (CTCF binding site)	1	blood (DND-41)	H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me1, DNase1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3, H3K27ac, H3K9ac, H3K79me2, H3K4me1, H3K4me2
		blood (GM12878)	H3K79me2
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K4me1, H3K36me3
ENSR00001494480 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, USF1
		blood (K562)	H3K27me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1, DNase1
		muscle (HSMM)	H3K79me2
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	DNase1, HNF4G, H3K4me3, PolII, H3K79me2, USF1, H3K4me1, H3K4me2, HNF4A, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K79me2, H3K4me2, H3K36me3
		A549	H3K36me3

		skin (NHEK)	H3K4me1, H3K4me2, H3K36me3
ENSR00000487623 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, PolII, TAF7, H3K27ac, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3, DNase1
		HSMMtube	H2AZ, H3K4me3, H3K4me2, DNase1, H3K9ac, H3K79me2
		blood (K562)	H3K27me3, H3K9ac, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		liver (HepG2)	Gabp, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K9ac, H3K4me3, H3K4me2, DNase1
		blood (GM12878)	H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, ELF1, DNase1, PolII, H2AZ
		nervous (NH-A)	H3K4me2, H3K9ac, H3K4me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K27ac, H3K9ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K27me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	Ini1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, DNase1, H3K4me2
		endothelium (HUVEC)	H3K4me2, H3K4me3, H3K9ac, PolII, DNase1, Max
		A549	H3K36me3, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PAIP1P1	downstream gene variant	224	ENST00000446875	?	?	2
TRIM10	downstream gene variant, upstream gene variant	2412	ENST00000376704	NM_052828.2	ENSP00000365894	8
TRIM10	downstream gene variant, upstream gene variant	2412	ENST00000449742	NM_006778.3	ENSP00000397073	8
TRIM15	upstream gene variant, downstream gene variant	113	ENST00000433744	?	ENSP00000398285	9
TRIM15	downstream gene variant	576	ENST00000619857	?	ENSP00000484001	1
TRIM15	downstream gene variant	569	ENST00000376694	NM_033229.2	ENSP00000365884	1
TRIM15	upstream gene variant, downstream gene variant	2113	ENST00000477944	?	?	3
TRIM15	upstream gene variant, downstream gene variant	137	ENST00000376688	?	ENSP00000365878	3
TRIM26	upstream gene variant	672	ENST00000418026	?	ENSP00000387530	1
TRIM26	upstream gene variant	692	ENST00000487829	?	?	1
TRIM26	upstream gene variant	638	ENST00000416596	?	ENSP00000413673	1
TRIM26	downstream gene variant, upstream gene variant	199	ENST00000437089	?	ENSP00000395491	3
TRIM26	upstream gene variant	691	ENST00000434785	?	ENSP00000400920	1
TRIM26	downstream gene variant	199	ENST00000480999	?	?	1
TRIM26	downstream gene variant, upstream gene variant	199	ENST00000454678	NM_003449.4	ENSP00000410446	2
TRIM26	downstream gene variant, upstream gene variant	199	ENST00000453195	NM_001242783.1	ENSP00000391879	2
TRIM31	upstream gene variant	4060	ENST00000468264	?	?	1
TRIM31	upstream gene variant	553	ENST00000480808	?	?	3
TRIM31	upstream gene variant	73	ENST00000485864	?	?	3
TRIM31	upstream gene variant	1378	ENST00000493404	?	?	2
TRIM31	upstream gene variant	747	ENST00000376734	NM_007028.3	ENSP00000365924	2
TRIM40	downstream gene variant	259	ENST00000307859	NM_138700.4	ENSP00000308310	1
TRIM40	downstream gene variant	259	ENST00000376724	?	ENSP00000365914	1
TRIM40	downstream gene variant	259	ENST00000396581	NM_001286633.1	ENSP00000379826	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TRIM15	ENST00000433744	?	ENSP00000398285	1
TRIM15	ENST00000619857	?	ENSP00000484001	6
TRIM15	ENST00000376688	?	ENSP00000365878	6
TRIM15	ENST00000376694	NM_033229.2	ENSP00000365884	6
TRIM26	ENST00000480999	?	?	2
TRIM26	ENST00000453195	NM_001242783.1	ENSP00000391879	7
TRIM26	ENST00000454678	NM_003449.4	ENSP00000410446	7
TRIM26	ENST00000434785	?	ENSP00000400920	5
TRIM26	ENST00000418026	?	ENSP00000387530	5
TRIM26	ENST00000437089	?	ENSP00000395491	5
TRIM26	ENST00000487829	?	?	5
TRIM26	ENST00000416596	?	ENSP00000413673	5
TRIM31	ENST00000376734	NM_007028.3	ENSP00000365924	1
TRIM31	ENST00000493404	?	?	1
TRIM31-AS1	ENST00000440874	?	?	3
TRIM40	ENST00000396581	NM_001286633.1	ENSP00000379826	2
TRIM40	ENST00000376724	?	ENSP00000365914	2
TRIM40	ENST00000307859	NM_138700.4	ENSP00000308310	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TRIM15	ENST00000619857	?	ENSP00000484001	3
TRIM15	ENST00000376694	NM_033229.2	ENSP00000365884	2
TRIM26	ENST00000453195	NM_001242783.1	ENSP00000391879	1
TRIM26	ENST00000454678	NM_003449.4	ENSP00000410446	1
TRIM26	ENST00000437089	?	ENSP00000395491	1
TRIM26	ENST00000416596	?	ENSP00000413673	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PAIP1P1	ENST00000446875	?	1
TRIM15	ENST00000477944	?	1
TRIM26	ENST00000487829	?	1

Locus 424 (rs7535099)

SNiPAcard

Block annotations

Block info

genomic range	chr1:66,030,445-66,053,063
block size	22,619 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.094 [-2.963 – 1.639]	gene(s) hit or close-by	LEPR, RP11-430H12.2
phastCons	μ = 0.089 [0 – 1]	eQTL gene(s)	LEPR, LEPROT
GERP++	μ = -0.541 [-6.47 – 2.49]	potentially regulated gene(s)	RP11-430H12.2
CADD score	μ = 3.503 [0.504 – 9.95]	disease gene(s)	LEPR

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
3-carboxy-4-methyl-5-propyl-2-furanpropanoate (CMPF)	4.14×10^{-5}	Metabolomics GWAS Server	24816252	4
Body mass index	9.45×10^{-5}	dbGaP	pha003009	1

Disease gene annotation

gene	trait	source DB	source entry/link
LEPR	LEPTIN RECEPTOR DEFICIENCY	OMIM	MIM:614963
LEPR	Obesity due to leptin receptor gene deficiency	OrphaNet	OrphaNet:179494

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LEPR	ENST00000616738	ILMN_2234956	monocyte	3.09×10^{-17} (p-value)	Zeller et al.	4
LEPR	ENST00000371060					
LEPROT	ENST00000488747	ILMN_1661537	blood	1.52×10^{-6} (p-value)	Westra et al.	1
LEPROT	ENST00000475108					
LEPROT	ENST00000613538					
LEPROT	ENST00000371065					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000002892		

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000670220 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	CTCF, Rad21
		endothelium (HUVEC)	H3K36me3, CTCF

	liver (HepG2)	Rad21, CTCF
	lung (IMR90)	CTCF
	blood (K562)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LEPR	upstream gene variant	784	ENST00000371058	?	ENSP00000360097	1
RP11-430H12.2	downstream gene variant, upstream gene variant	2814	ENST00000426714	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LEPR	ENST00000371058	?	ENSP00000360097	15
LEPR	ENST00000616738	NM_001198689.1	ENSP00000483390	16
LEPR	ENST00000406510	?	ENSP00000384025	16
LEPR	ENST00000344610	NM_001198688.1	ENSP00000340884	16
LEPR	ENST00000349533	NM_002303.5	ENSP00000330393	16
LEPR	ENST00000462765	?	?	16
LEPR	ENST00000371059	NM_001198687.1, NM_001003680.3	ENSP00000360098	16
LEPR	ENST00000371060	NM_001003679.3	ENSP00000360099	16

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-430H12.2	ENST00000426714	?	1

Locus 425 (rs41265930)

SNiPAcard

Block annotations

Block info

genomic range	chr6:160,964,135-160,964,135
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	1.009	gene(s) hit or close-by	LPA
phastCons	0.012	eQTL gene(s)	–
GERP++	0.476	potentially regulated gene(s)	–
CADD score	2.631	disease gene(s)	–

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LPA	ENST00000316300	NM_005577.2	ENSP00000321334	1

Locus 426 (rs7896471)

SNiPAcard

Block annotations

Block info

genomic range	chr10:101,786,600-101,792,462
block size	5,863 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.834 [-2.774 – 0.224]	**gene(s) hit or close-by**	–
phastCons	μ = 0.109 [0.004 – 0.353]	**eQTL gene(s)**	–
GERP++	μ = -0.108 [-0.317 – 0.113]	**potentially regulated gene(s)**	–
CADD score	μ = 2.163 [0.156 – 7.061]	**disease gene(s)**	–

Locus 427 (rs6808835)

SNiPAcard

Block annotations

Block info

genomic range	chr3:46,275,570-46,482,980
block size	207,411 bp
variant count	90 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.456 [-4.588 – 1.248]	gene(s) hit or close-by	CCR2, CCR3, CCR5, CCRL2, LTF, RP11-24F11.2, UQCRC2P1
phastCons	μ = 0.052 [0 – 0.59]	eQTL gene(s)	CCR3, LIMD1, LIMD1-AS1, LRRC2
GERP++	μ = -0.526 [-10.7 – 3.83]	potentially regulated gene(s)	AC104304.1, ALS2CL, CCR1, CCR2, CCR3, CCR5, CCR9, CCRL2, CXCR6, LRRC2, LTF, LZTFL1, PRSS44, PRSS50, RP11-24F11.2, XCR1
CADD score	μ = 3.969 [0.056 – 10.62]	disease gene(s)	LZTFL1, CCR5, LTF, CCR2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Cerebrospinal fluid levels of Alzheimer's disease-related proteins	$<2.00\times10^{-13}$	GWAS Catalog	25340798	1

Disease gene annotation

gene	trait	source DB	source entry/link
LZTFL1	BARDET-BIEDL SYNDROME 17	OMIM	MIM:615994
CCR5	WEST NILE VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610379
CCR5	DIABETES MELLITUS, INSULIN-DEPENDENT, 22	OMIM	MIM:612522
CCR5	HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, SUSCEPTIBILITY TO	OMIM	MIM:609423
LTF	SPECIFIC GRANULE DEFICIENCY	OMIM	MIM:245480
CCR2	HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, SUSCEPTIBILITY TO	OMIM	MIM:609423
LZTFL1	BARDET-BIEDL SYNDROME	OrphaNet	OrphaNet:110

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CCRL2	missense variant	ENST00000399036	NM_003965.4	ENSP00000381994	M/V	Atg/Gtg	?	?	1
CCRL2	missense variant	ENST00000357392	NM_001130910.1	ENSP00000349967	M/V	Atg/Gtg	?	?	1
CCRL2	missense variant	ENST00000400880	?	ENSP00000383677	M/V	Atg/Gtg	?	?	1
CCRL2	missense variant	ENST00000400882	?	ENSP00000383678	M/V	Atg/Gtg	?	?	1
CCRL2	missense variant	ENST00000433848	?	ENSP00000414957	M/V	Atg/Gtg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LIMD1-AS1	?	ENSG00000230530	tibial nerve	5.71×10-6 (p-value)	GTEx Portal V6	23
LIMD1	?	ENSG00000144791	tibial nerve	9.94×10-7 (p-value)	GTEx Portal V6	48
LRRC2	?	ENSG00000163827	transformed fibroblasts	4.59×10-8 (p-value)	GTEx Portal V6	84
LIMD1-AS1	?	ENSG00000230530	tibial artery	9.71×10-6 (p-value)	GTEx Portal V6	8
LIMD1	?	ENSG00000144791	tibial artery	2.16×10-6 (p-value)	GTEx Portal V6	31
CCR3	?	ENSG00000183625	blood	6.66×10-6 (p-value)	GTEx Portal V6	8

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000607398	1	ENST00000443496, ENST00000417439, ENST00000493056, ENST00000426532, ENST00000478874, ENST00000231751	LTF

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000036249		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000337650	1	ENCP00000036251	RP11-24F11.2 CCRL2
		ENCP00000036271	ALS2CL
		ENCP00000036243	CCR3
		ENCP00000036252	CCRL2
		ENCP00000036239	XCR1
		ENCP00000036242	CCR1
ENCE00000337509	1	ENCP00000036231	LZTFL1
		ENCP00000036227	LZTFL1
		ENCP00000036250	CCR5
		ENCP00000036230	CCR9
		ENCP00000036232	LZTFL1
		ENCP00000036234	CXCR6
ENCE00000337805	1	ENCP00000036249	CCR5
ENCE00000337856	1	ENCP00000036254	LTF
ENCE00000337664	1	ENCP00000036251	RP11-24F11.2 CCRL2
		ENCP00000036243	CCR3
		ENCP00000036271	ALS2CL
		ENCP00000036252	CCRL2
		ENCP00000036239	XCR1
		ENCP00000036242	CCR1
ENCE00000337563	3	ENCP00000036231	LZTFL1
		ENCP00000036250	CCR5
		ENCP00000036230	CCR9
		ENCP00000036232	LZTFL1
		ENCP00000036234	CXCR6
ENCE00000337711	1	ENCP00000036261	LRRC2

		ENCP00000036273	PRSS50
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036262	AC104304.1
		ENCP00000036244	CCR3
ENCE00000337859	1	ENCP00000036254	LTF
ENCE00000337713	1	ENCP00000036261	LRRC2
		ENCP00000036273	PRSS50
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036262	AC104304.1
		ENCP00000036244	CCR3
ENCE00000337671	1	ENCP00000036243	CCR3
		ENCP00000036271	ALS2CL
		ENCP00000036239	XCR1
		ENCP00000036242	CCR1
ENCE00000337715	2	ENCP00000036261	LRRC2
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036262	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036244	CCR3
		ENCP00000036258	LTF
		ENCP00000036273	PRSS50

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001478830 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		embryonic stem cell (H1ESC)	CTCF, Rad21
		endothelium (HUVEC)	H3K36me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
ENSR00001362902 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
ENSR00001362911 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1
		lung (IMR90)	H3K27me3
		blood (GM12878)	H3K4me1
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	H3K27me3
		skin (NHEK)	DNase1
ENSR00001674590 (enhancer)	1	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac
		muscle (HSMM)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1
		blood (GM12878)	PU1, H3K4me1, H3K27ac
		lung (IMR90)	H3K27me3
		A549	H3K27me3
ENSR00001362921 (promoter flanking region)	1	HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1, H3K27me3

		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3, H3K36me3
		endothelium (HUVEC)	H3K27me3
		blood (GM12878)	BATF, H3K4me1
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001478841 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	CTCF
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1, CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, H3K27me3
		A549	H3K27me3
ENSR00001478842 (promoter flanking region)	1	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	H3K27me3
ENSR00001674606 (enhancer)	3	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001478843 (promoter flanking region)	3	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		blood (K562)	H3K27me3, DNase1
		blood (DND-41)	H3K4me1, H3K27ac
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1
ENSR00001674607 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	Max, DNase1, H3K27me3
		blood (DND-41)	H3K27ac, H3K4me1
		nervous (NH-A)	H3K27me3
ENSR00001362934 (promoter flanking region)	1	cervix (HeLa-S3)	PolII, H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		blood (K562)	H3K27me3, DNase1, Max
		blood (DND-41)	H3K4me1, H3K27ac
ENSR00001362946	1	HSMMtube	H3K27me3

(enhancer)		liver (HepG2)	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	DNase1, H2AZ
		blood (DND-41)	H3K4me1
ENSR00001362947 (open chromatin region)	3	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
ENSR00001362952 (open chromatin region)	2	A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	DNase1, H3K4me1, H3K4me2, H3K27ac
ENSR00001674619 (enhancer)	1	lung (IMR90)	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCR2	downstream gene variant	1779	ENST00000445132	NM_001123396.1	ENSP00000399285	1
CCR2	downstream gene variant	276	ENST00000292301	NM_001123041.2	ENSP00000292301	1
CCR2	downstream gene variant	269	ENST00000400888	?	ENSP00000383681	1
CCR2	downstream gene variant	2509	ENST00000465202	?	?	1
CCR2	downstream gene variant	679	ENST00000421659	?	ENSP00000396736	2
CCR3	upstream gene variant	2491	ENST00000457243	?	ENSP00000401822	2
CCR3	upstream gene variant	2461	ENST00000395940	NM_178329.2, NM_001837.3	ENSP00000379271	2
CCR3	upstream gene variant	2411	ENST00000545097	NM_178328.1, NM_001164680.1	ENSP00000441600	2
CCR3	upstream gene variant	2491	ENST00000484025	?	?	2
CCR3	upstream gene variant	2490	ENST00000452454	?	ENSP00000389336	2
CCR3	upstream gene variant	3681	ENST00000395942	?	ENSP00000379273	1
CCR3	upstream gene variant	1943	ENST00000475150	?	?	2
CCR5	upstream gene variant, downstream gene variant	684	ENST00000292303	NM_001100168.1, NM_000579.3	ENSP00000292303	4
CCR5	upstream gene variant	2500	ENST00000445772	?	ENSP00000404881	2
CCRL2	upstream gene variant, downstream gene variant	489	ENST00000400882	?	ENSP00000383678	15
CCRL2	upstream gene variant, downstream gene variant	411	ENST00000495870	?	?	16
CCRL2	upstream gene variant, downstream gene variant	488	ENST00000357392	NM_001130910.1	ENSP00000349967	15
CCRL2	upstream gene variant, downstream gene variant	488	ENST00000399036	NM_003965.4	ENSP00000381994	16
CCRL2	upstream gene variant, downstream gene variant	279	ENST00000400880	?	ENSP00000383677	15
CCRL2	upstream gene variant, downstream gene variant	662	ENST00000433848	?	ENSP00000414957	15
CCRL2	upstream gene variant, downstream gene variant	121	ENST00000441909	?	?	19
LTF	downstream gene variant	2014	ENST00000231751	NM_002343.4	ENSP00000231751	4
LTF	downstream gene variant	2388	ENST00000426532	NM_001199149.1	ENSP00000405719	4
LTF	downstream gene variant	2570	ENST00000443496	?	ENSP00000397427	3
LTF	downstream gene variant	2562	ENST00000417439	?	ENSP00000405546	3
LTF	downstream gene variant	1695	ENST00000493056	?	?	5
LTF	downstream gene variant	1756	ENST00000478874	?	?	5

RP11-24F11.2	downstream gene variant, upstream gene variant	1314	ENST00000451485	?	?	9
UQCRC2P1	downstream gene variant, upstream gene variant	1366	ENST00000417777	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CCR2	ENST00000445132	NM_001123396.1	ENSP00000399285	T	acA/acG	1
CCRL2	ENST00000357392	NM_001130910.1	ENSP00000349967	G	ggT/ggG	1
CCRL2	ENST00000400882	?	ENSP00000383678	G	ggT/ggG	1
CCRL2	ENST00000433848	?	ENSP00000414957	G	ggT/ggG	1
CCRL2	ENST00000400880	?	ENSP00000383677	G	ggT/ggG	1
CCRL2	ENST00000399036	NM_003965.4	ENSP00000381994	G	ggT/ggG	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
LTF	ENST00000417439	?	ENSP00000405546	1
LTF	ENST00000443496	?	ENSP00000397427	1
LTF	ENST00000426532	NM_001199149.1	ENSP00000405719	1
LTF	ENST00000231751	NM_002343.4	ENSP00000231751	1
LTF	ENST00000493056	?	?	1
LTF	ENST00000478874	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCR2	ENST00000292301	NM_001123041.2	ENSP00000292301	1
CCR2	ENST00000400888	?	ENSP00000383681	1
CCR3	ENST00000452454	?	ENSP00000389336	6
CCR3	ENST00000457243	?	ENSP00000401822	6
CCR3	ENST00000484025	?	?	6
CCR3	ENST00000545097	NM_178328.1, NM_001164680.1	ENSP00000441600	6
CCR3	ENST00000357422	?	ENSP00000350003	11
CCR3	ENST00000395940	NM_178329.2, NM_001837.3	ENSP00000379271	6
CCRL2	ENST00000441909	?	?	5
LTF	ENST00000417439	?	ENSP00000405546	3
LTF	ENST00000443496	?	ENSP00000397427	3
LTF	ENST00000426532	NM_001199149.1	ENSP00000405719	3
LTF	ENST00000231751	NM_002343.4	ENSP00000231751	3
LTF	ENST00000493056	?	?	1
LTF	ENST00000478874	?	?	1
RP11-24F11.2	ENST00000451485	?	?	30

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

gene	affected transcript	RefSeq id	protein	variant(s)
CCRL2	ENST00000357392	NM_001130910.1	ENSP00000349967	1
CCRL2	ENST00000400882	?	ENSP00000383678	1
CCRL2	ENST00000399036	NM_003965.4	ENSP00000381994	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CCR2	ENST00000465202	?	1

Locus 428 (rs35931837)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,033,158-52,033,158
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.302	gene(s) hit or close-by	SIGLEC6
phastCons	0.083	eQTL gene(s)	SIGLEC12
GERP++	1.55	potentially regulated gene(s)	–
CADD score	8.342	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SIGLEC6	missense variant	ENST00000425629	NM_001245.5	ENSP00000401502	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000359982	NM_001177548.1	ENSP00000353071	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000436458	NM_001177547.1	ENSP00000410679	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000343300	NM_198846.4	ENSP00000345907	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000391797	NM_001177549.1	ENSP00000375674	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000346477	NM_198845.4	ENSP00000344064	S/G	Agc/Ggc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC12	?	ENSG00000254521	blood	5.91×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SIGLEC6	upstream gene variant	1457	ENST00000474054	?	?	1

Putative effect on transcript

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SIGLEC6	ENST00000489837	?	1
SIGLEC6	ENST00000496422	?	1

Locus 428 (sentinel SNP rs35931837 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 429 (rs3213958)

SNiPAcard

Block annotations

Block info

genomic range	chr3:98,487,206-98,487,206
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	1.589	gene(s) hit or close-by	ST3GAL6
phastCons	0.01	eQTL gene(s)	–
GERP++	4.14	potentially regulated gene(s)	–
CADD score	9.889	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ST3GAL6	upstream gene variant	2693	ENST00000485145	?	ENSP00000419202	1
ST3GAL6	upstream gene variant	2027	ENST00000477574	?	ENSP00000419987	1
ST3GAL6	upstream gene variant	3880	ENST00000474595	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ST3GAL6	ENST00000483910	NM_001271146.1	ENSP00000417376	1
ST3GAL6	ENST00000394162	NM_006100.3	ENSP00000377717	1
ST3GAL6	ENST00000469105	?	ENSP00000419690	1
ST3GAL6	ENST00000490684	?	?	1
ST3GAL6	ENST00000466482	?	?	1
ST3GAL6	ENST00000497621	?	ENSP00000418581	1
ST3GAL6	ENST00000476833	?	ENSP00000417109	1
ST3GAL6	ENST00000485391	?	ENSP00000418650	1
ST3GAL6	ENST00000489112	?	?	1
ST3GAL6	ENST00000486334	?	ENSP00000418896	1
ST3GAL6	ENST00000495376	?	ENSP00000420693	1
ST3GAL6	ENST00000613264	NM_001271145.1	ENSP00000480884	1
ST3GAL6	ENST00000460774	?	ENSP00000419209	1
ST3GAL6	ENST00000468553	?	ENSP00000420474	1
ST3GAL6	ENST00000492254	?	ENSP00000417201	1
ST3GAL6	ENST00000486249	?	ENSP00000420047	1
ST3GAL6	ENST00000462152	?	?	1
ST3GAL6	ENST00000497008	?	ENSP00000417584	1

ST3GAL6	ENST00000265261	NM_001271142.1, NM_001271147.1	ENSP00000265261	1
ST3GAL6	ENST00000480133	?	?	1

Locus 429 (sentinel SNP rs3213958 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 430 (rs300703)

SNiPAcard

Block annotations

Block info

genomic range	chr2:92,999-286,812
block size	193,814 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.867 [-4.969 – 0.641]	gene(s) hit or close-by	AC079779.4, AC079779.7, ACP1, FAM150B, SH3YL1
phastCons	μ = 0.037 [0 – 0.676]	eQTL gene(s)	AC079779.4, AC079779.7, SH3YL1
GERP++	μ = -0.316 [-2.61 – 1.65]	potentially regulated gene(s)	AC093326.2, AC105393.2, SH3YL1, TMEM18
CADD score	μ = 3.032 [0.03 – 10.34]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SH3YL1	?	ENSG00000035115	muscularis mucosae	9.02×10^{-7} (p-value)	GTEx Portal V6	18
AC079779.7	?	ENSG00000227061	testis	1.95×10^{-7} (p-value)	GTEx Portal V6	17
SH3YL1	?	ENSG00000035115	left ventricle	5.23×10^{-6} (p-value)	GTEx Portal V6	1
AC079779.4	?	ENSG00000228643	thyroid	1.90×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024884		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000234694	1	ENCP00000024903	TMEM18
		ENCP00000024897	AC105393.2
		ENCP00000024899	AC093326.2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001538823 (open chromatin region)	1	A549	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ACP1	2	ENST00000272065 ENST00000272067 ENST00000405233 ENST00000405364 ENST00000413140 ENST00000442386 ENST00000453390	hsa-miR-216b-5p hsa-miR-9-5p

ENST00000463831
ENST00000480874
ENST00000484464

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC079779.7	downstream gene variant	388	ENST00000437798	?	?	2
ACP1	upstream gene variant, downstream gene variant	440	ENST00000439645	?	ENSP00000408596	3
ACP1	upstream gene variant	870	ENST00000484125	?	?	1
ACP1	upstream gene variant, downstream gene variant	1649	ENST00000405364	?	ENSP00000384184	2
ACP1	upstream gene variant, downstream gene variant	1503	ENST00000463831	?	?	2
ACP1	upstream gene variant, downstream gene variant	1602	ENST00000272065	NM_004300.3	ENSP00000272065	2
ACP1	upstream gene variant, downstream gene variant	1599	ENST00000272067	NM_007099.3	ENSP00000272067	2
ACP1	upstream gene variant, downstream gene variant	1935	ENST00000484464	?	?	2
ACP1	upstream gene variant, downstream gene variant	1630	ENST00000442386	?	ENSP00000389681	2
ACP1	upstream gene variant, downstream gene variant	1695	ENST00000453390	?	ENSP00000411121	2
ACP1	upstream gene variant, downstream gene variant	1623	ENST00000413140	?	ENSP00000410331	2
ACP1	upstream gene variant, downstream gene variant	1297	ENST00000480874	?	?	2
ACP1	upstream gene variant, downstream gene variant	1656	ENST00000405233	?	ENSP00000384307	2
ACP1	upstream gene variant, downstream gene variant	513	ENST00000407983	NM_001040649.2	ENSP00000385404	3
FAM150B	downstream gene variant	5000	ENST00000463919	?	?	1
FAM150B	downstream gene variant	3021	ENST00000619265	?	ENSP00000478180	1
FAM150B	downstream gene variant	3021	ENST00000401489	?	ENSP00000385214	1
FAM150B	downstream gene variant	2858	ENST00000401503	?	ENSP00000385537	1
FAM150B	downstream gene variant	3043	ENST00000344414	?	ENSP00000339565	1
FAM150B	downstream gene variant	2172	ENST00000452023	?	ENSP00000389939	1
FAM150B	downstream gene variant	3021	ENST00000405290	?	ENSP00000385672	1
FAM150B	downstream gene variant	2650	ENST00000403610	NM_001002919.2	ENSP00000384604	1
SH3YL1	downstream gene variant	4428	ENST00000460974	?	?	1
SH3YL1	downstream gene variant	4664	ENST00000454318	?	ENSP00000415723	1
SH3YL1	downstream gene variant	3679	ENST00000602998	?	?	1
SH3YL1	downstream gene variant, upstream gene variant	484	ENST00000473104	?	?	2
SH3YL1	upstream gene variant	2978	ENST00000605370	?	?	1
SH3YL1	upstream gene variant	2562	ENST00000479739	?	ENSP00000441266	1
SH3YL1	downstream gene variant	1873	ENST00000471948	?	?	1
SH3YL1	downstream gene variant	2632	ENST00000488044	?	?	1
SH3YL1	downstream gene variant	4847	ENST00000626873	NM_001282687.1	ENSP00000485824	1
SH3YL1	downstream gene variant	4618	ENST00000475027	?	?	1
SH3YL1	downstream gene variant	4851	ENST00000405430	?	ENSP00000384269	1
SH3YL1	downstream gene variant	3504	ENST00000415368	?	ENSP00000410235	1
SH3YL1	downstream gene variant, upstream gene variant	1947	ENST00000403657	?	ENSP00000385668	2
SH3YL1	downstream gene variant	145	ENST00000402632	?	ENSP00000384910	3
SH3YL1	downstream gene variant, upstream gene variant	1874	ENST00000481932	?	ENSP00000474411	3
SH3YL1	downstream gene variant	4614	ENST00000488079	?	?	1

SH3YL1	downstream gene variant	4614	ENST00000488979	?	?	1
SH3YL1	downstream gene variant	1389	ENST00000431160	?	ENSP00000399519	1
SH3YL1	downstream gene variant, upstream gene variant	2427	ENST00000497051	?	?	2
SH3YL1	downstream gene variant	4856	ENST00000356150	NM_015677.3	ENSP00000348471	1
SH3YL1	downstream gene variant	4834	ENST00000403712	NM_001159597.2	ENSP00000384276	1
SH3YL1	downstream gene variant	4847	ENST00000463865	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC079779.4	ENST00000427831	?	?	1
AC079779.4	ENST00000450709	?	?	1
ACP1	ENST00000484464	?	?	2
ACP1	ENST00000405364	?	ENSP00000384184	1
ACP1	ENST00000272065	NM_004300.3	ENSP00000272065	2
ACP1	ENST00000463831	?	?	1
ACP1	ENST00000272067	NM_007099.3	ENSP00000272067	2
ACP1	ENST00000480874	?	?	1
ACP1	ENST00000442386	?	ENSP00000389681	2
ACP1	ENST00000413140	?	ENSP00000410331	2
ACP1	ENST00000453390	?	ENSP00000411121	2
FAM150B	ENST00000452023	?	ENSP00000389939	1
FAM150B	ENST00000401489	?	ENSP00000385214	2
FAM150B	ENST00000619265	?	ENSP00000478180	2
FAM150B	ENST00000405290	?	ENSP00000385672	2
FAM150B	ENST00000401503	?	ENSP00000385537	2
FAM150B	ENST00000403610	NM_001002919.2	ENSP00000384604	2
FAM150B	ENST00000344414	?	ENSP00000339565	2
FAM150B	ENST00000463919	?	?	1
SH3YL1	ENST00000405430	?	ENSP00000384269	9
SH3YL1	ENST00000463865	?	?	9
SH3YL1	ENST00000415368	?	ENSP00000410235	2
SH3YL1	ENST00000488044	?	?	8
SH3YL1	ENST00000403657	?	ENSP00000385668	8
SH3YL1	ENST00000479739	?	ENSP00000441266	8
SH3YL1	ENST00000402632	?	ENSP00000384910	5
SH3YL1	ENST00000468321	?	?	9
SH3YL1	ENST00000462719	?	?	1
SH3YL1	ENST00000473104	?	?	8
SH3YL1	ENST00000475027	?	?	8
SH3YL1	ENST00000403712	NM_001159597.2	ENSP00000384276	9

SH3YL1	ENST00000488979	?	?	8
SH3YL1	ENST00000451005	?	ENSP00000416312	8
SH3YL1	ENST00000471948	?	?	8
SH3YL1	ENST00000431160	?	ENSP00000399519	7
SH3YL1	ENST00000497051	?	?	6
SH3YL1	ENST00000626873	NM_001282687.1	ENSP00000485824	9
SH3YL1	ENST00000403658	NM_001282682.1	ENSP00000383928	9
SH3YL1	ENST00000454318	?	ENSP00000415723	8
SH3YL1	ENST00000356150	NM_015677.3	ENSP00000348471	9
SH3YL1	ENST00000472012	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACP1	ENST00000405233	?	ENSP00000384307	1
SH3YL1	ENST00000402632	?	ENSP00000384910	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SH3YL1	ENST00000405430	?	ENSP00000384269	1
SH3YL1	ENST00000415368	?	ENSP00000410235	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SH3YL1	ENST00000465733	?	1
SH3YL1	ENST00000471948	?	1
SH3YL1	ENST00000472861	?	1
SH3YL1	ENST00000477707	?	1

Locus 431 (rs3788504)

SNiPAcard

Block annotations

Block info

genomic range	chr22:33,160,397-33,160,397
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.753	gene(s) hit or close-by	SYN3
phastCons	0	eQTL gene(s)	–
GERP++	-2.08	potentially regulated gene(s)	–
CADD score	0.809	disease gene(s)	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000293419 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		lung (IMR90)	DNase1, H3K18ac
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SYN3	ENST00000462268	?	?	1
SYN3	ENST00000358763	NM_003490.3, NM_001135774.1, NM_133633.2	ENSP00000351614	1

Locus 432 (rs41264869)

SNiPAcard

Block annotations

Block info

genomic range	chr1:205,028,581-205,087,683
block size	59,103 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.211 [-1.802 – 1.411]	gene(s) hit or close-by	AL583832.1, CNTN2, RBBP5, RP11-536L3.4, TMEM81
phastCons	μ = 0.014 [0 – 0.121]	eQTL gene(s)	CNTN2, DSTYK, RBBP5, TMCC2
GERP++	μ = -0.422 [-5.23 – 3.62]	potentially regulated gene(s)	CNTN2
CADD score	μ = 3.701 [0.3 – 8.736]	disease gene(s)	CNTN2, DSTYK

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CNTN2	EPILEPSY, FAMILIAL ADULT MYOCLONIC, 5	OMIM	MIM:615400
DSTYK	CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT, SUSCEPTIBILITY [...]	OMIM	MIM:610805
CNTN2	Benign adult familial myoclonic epilepsy	OrphaNet	OrphaNet:86814
DSTYK	CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT, CAKUT1	DECIPHER	MIM:610805

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RBBP5	?	ENSG00000117222	transformed fibroblasts	7.58×10^{-6} (p-value)	GTEx Portal V6	1
RBBP5	?	ENSG00000117222	tibial artery	4.05×10^{-6} (p-value)	GTEx Portal V6	1
TMCC2	?	ENSG00000133069	tibial artery	1.25×10^{-5} (p-value)	GTEx Portal V6	2
RBBP5	?	ENSG00000117222	thyroid	7.76×10^{-6} (p-value)	GTEx Portal V6	12
DSTYK	ENST00000367162	ILMN_2352023	monocyte	1.18×10^{-32} (p-value)	Zeller et al.	1
DSTYK	ENST00000367161					
CNTN2	?	ENSG00000184144	unexposed skin	9.06×10^{-6} (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
RBBP5	ENST00000264515	205169_at	chr1	blood	7.50×10^{-8} (p-value)	Dixon et al.	1
RBBP5	ENST00000367164						

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000006260		

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000167977 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	Rad21, CTCF, DNase1, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3, DNase1
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL583832.1	upstream gene variant, downstream gene variant	274	ENST00000515887	?	?	4
CNTN2	downstream gene variant	855	ENST00000331830	NM_005076.3	ENSP00000330633	2
CNTN2	upstream gene variant, downstream gene variant	486	ENST00000525433	?	?	5
CNTN2	downstream gene variant	1039	ENST00000532366	?	?	4
CNTN2	upstream gene variant, downstream gene variant	100	ENST00000527340	?	?	3
CNTN2	upstream gene variant, downstream gene variant	1937	ENST00000530594	?	?	5
CNTN2	downstream gene variant	758	ENST00000481872	?	?	4
CNTN2	downstream gene variant	281	ENST00000530117	?	?	2
RP11-536L3.4	downstream gene variant	460	ENST00000603227	?	?	2
TMEM81	downstream gene variant	3641	ENST00000367167	NM_203376.1	ENSP00000356135	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN2	ENST00000481872	?	?	8
CNTN2	ENST00000525433	?	?	1
CNTN2	ENST00000530117	?	?	2
CNTN2	ENST00000331830	NM_005076.3	ENSP00000330633	8
CNTN2	ENST00000527340	?	?	2
RBBP5	ENST00000264515	NM_005057.3, NM_001193273.1	ENSP00000264515	7
RBBP5	ENST00000367164	NM_001193272.1	ENSP00000356132	7
RBBP5	ENST00000484379	?	?	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN2	ENST00000331830	NM_005076.3	ENSP00000330633	2

CNTNAP2	ENST00000538538	NM_014141.6	ENSP00000438699	2

Locus 432 (sentinel SNP rs41264869 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

Locus 433 (rs2228467)

SNiPAcard

Block annotations

Block info

genomic range	chr3:42,906,116-42,906,116
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	2.284	gene(s) hit or close-by	ACKR2, CYP8B1, KRBOX1, RP11-136C24.3, RP11-141M3.6
phastCons	0.985	eQTL gene(s)	–
GERP++	5.52	potentially regulated gene(s)	–
CADD score	10.24	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Cerebrospinal fluid levels of Alzheimer's disease-related proteins	$<4.00\times10^{-18}$	GWAS Catalog	25340798	1
Monocyte count	$<2.00\times10^{-7}$	GWAS Catalog	23314186	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ACKR2	missense variant	ENST00000493193	?	ENSP00000476581	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000442925	?	ENSP00000396150	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000497921	?	ENSP00000476901	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000494619	?	ENSP00000477157	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000422265	NM_001296.4	ENSP00000416996	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000273145	?	ENSP00000273145	A/V	gCg/gTg	?	?	1
ACKR2	missense variant	ENST00000492609	?	ENSP00000477475	A/V	gCg/gTg	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ACKR2	downstream gene variant	90	ENST00000463699	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)

CYP8B1	ENST00000437102	?	ENSP00000404499	1
KRBOX1	ENST00000426937	?	ENSP00000413859	1
RP11-136C24.3	ENST00000451200	?	?	1
RP11-141M3.6	ENST00000487368	?	?	1
RP11-141M3.6	ENST00000471537	?	?	1
RP11-141M3.6	ENST00000496604	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ACKR2	ENST00000460855	?	1
ACKR2	ENST00000498111	?	1

Locus 434 (rs2246662)

SNiPAcard

Block annotations

Block info

genomic range	chr1:204,943,947-204,943,991
block size	45 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.410 [-2.081 – 1.261]	gene(s) hit or close-by	NFASC
phastCons	μ = 0.500 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 1.000 [-3.8 – 5.8]	potentially regulated gene(s)	–
CADD score	μ = 8.738 [3.806 – 13.67]	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NFASC	downstream gene variant	136	ENST00000514644	?	?	2
NFASC	upstream gene variant	4525	ENST00000471392	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
NFASC	ENST00000339876	NM_001005388.2	ENSP00000344786	V	gtA/gtC	1
NFASC	ENST00000404076	?	ENSP00000385676	V	gtA/gtC	1
NFASC	ENST00000513543	?	ENSP00000425908	V	gtA/gtC	1
NFASC	ENST00000403080	NM_001005389.1, NM_001160333.1	ENSP00000384875	V	gtA/gtC	1
NFASC	ENST00000360049	NM_015090.3	ENSP00000353154	V	gtA/gtC	1
NFASC	ENST00000401399	?	ENSP00000385637	V	gtA/gtC	1
NFASC	ENST00000367173	?	ENSP00000356141	V	gtA/gtC	1
NFASC	ENST00000404907	?	ENSP00000384061	V	gtA/gtC	1
NFASC	ENST00000430393	NM_001160331.1	ENSP00000415031	V	gtA/gtC	1
NFASC	ENST00000539706	NM_001160332.1	ENSP00000438614	V	gtA/gtC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NFASC	ENST00000339876	NM_001005388.2	ENSP00000344786	1
NFASC	ENST00000404076	?	ENSP00000385676	1
NFASC	ENST00000513543	?	ENSP00000425908	1
NFASC	ENST00000404977	?	?	1
NFASC	ENST00000504476	?	ENSP00000422524	1
NFASC	ENST00000403080	NM_001005389.1, NM_001160333.1	ENSP00000384875	1
NFASC	ENST00000360049	NM_015090.3	ENSP00000353154	1

NFASC	ENST00000401399	?	ENSP00000385637	1
NFASC	ENST00000367173	?	ENSP00000356141	1
NFASC	ENST00000404907	?	ENSP00000384061	1
NFASC	ENST00000430393	NM_001160331.1	ENSP00000415031	1
NFASC	ENST00000539706	NM_001160332.1	ENSP00000438614	1
NFASC	ENST00000512826	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
NFASC	ENST00000504476	?	ENSP00000422524	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NFASC	ENST00000404977	?	1
NFASC	ENST00000512826	?	1

Locus 435 (rs530022)

SNiPAcard

Block annotations

Block info

genomic range	chr1:168,664,450-168,675,971
block size	11,522 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.033 [-0.449 – 0.632]	gene(s) hit or close-by	DPT
phastCons	μ = 0.002 [0 – 0.007]	eQTL gene(s)	DPT
GERP++	μ = -0.783 [-2.66 – 1.57]	potentially regulated gene(s)	RP5-1018K9.1, TBX19
CADD score	μ = 2.879 [1.17 – 5.142]	disease gene(s)	TBX19

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TBX19	ACTH DEFICIENCY, ISOLATED	OMIM	MIM:201400
TBX19	Congenital isolated ACTH deficiency	OrphaNet	OrphaNet:199296

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DPT	ENST00000367817	ILMN_1708107	skin	1.29×10-6 (p-value)	MuTHER consortium	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000043270	1	ENCP00000005412	TBX19
		ENCP00000005426	RP5-1018K9.1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DPT	downstream gene variant	247	ENST00000367817	NM_001937.4	ENSP00000356791	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DPT	ENST00000367817	NM_001937.4	ENSP00000356791	9

Locus 436 (rs4745216)

SNiPAcard

Block annotations

Block info

genomic range	chr9:75,714,313-75,789,730
block size	75,418 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.458 [-2.261 – 0.537]	gene(s) hit or close-by	ANXA1
phastCons	μ = 0.037 [0 – 0.534]	eQTL gene(s)	ANXA1
GERP++	μ = -1.387 [-9 – 2.1]	potentially regulated gene(s)	ANXA1
CADD score	μ = 3.607 [0.004 – 11.09]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Schizophrenia, bipolar disorder and depression (combined)	$<3.00\times10^{-6}$	GWAS Catalog	20713499	1
2-methoxyacetaminophen sulfate*	5.59×10^{-6}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ANXA1	?	ENSG00000135046	blood	1.37×10^{-3} (q-value)	SeeQTL DB (HapMap)	3

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CJUN	MA0489.1	10	no	0.000	1
CFOS	MA0476.1	6	no	0.000	1
JUND	MA0491.1	6	no	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000543116	1	ENCP00000058973	ANXA1
		ENCP00000058972	ANXA1
		ENCP00000058974	ANXA1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001305737 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac

		blood (DND-41)	H3K4me1, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Jund, H3K27ac
		endothelium (HUVEC)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me3, DNase1, H3K36me3
		skin (NHEK)	H3K27ac, DNase1
ENSR00001305741	2	embryonic stem cell (H1ESC)	DNase1
(promoter flanking region)		HSMMtube	H3K9ac, H3K4me2, H3K27ac, DNase1
		blood (K562)	DNase1, H3K27ac, MEF2A, H3K9ac, H2AZ, H3K4me3, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	H3K27me3
		blood (GM12878)	PolII, H2AZ, H3K4me3, H3K27ac, H3K9ac
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		nervous (NH-A)	H3K4me2, H3K4me3, H3K27ac, DNase1, H3K9ac
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, DNase1, H3K4me2, H3K27ac, TAF1, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K9ac, H3K27ac, PolII, H3K4me2, DNase1, Cjun
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00001730160	1	cervix (HeLa-S3)	CTCF, DNase1, Cjun, Jund
(CTCF binding site)		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		A549	H3K4me3, H3K4me2, DNase1
ENSR00001471274	1	cervix (HeLa-S3)	Jund, DNase1, CTCF, Cjun
(promoter flanking region)		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		A549	H3K4me3, H3K4me2, DNase1
		skin (NHEK)	DNase1, H3K27ac, H3K4me2
		breast (HMEC)	DNase1, H3K27ac
ENSR00001305755	1	embryonic stem cell (H1ESC)	DNase1
(promoter)		HSMMtube	H3K36me3, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K79me2, H3K9ac
		blood (K562)	PolII, H3K4me2, H3K36me3, H3K4me3, H2AZ, H3K79me2, Jund, Ini1, Brg1, DNase1, H3K27ac, H3K9ac
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H3K36me3, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		blood (GM12878)	IRF4, BATF, PU1, Cjun
		lung (IMR90)	DNase1, H3K79me2, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	DNase1, H3K27ac, H3K9ac, PolII, H3K4me2, H3K4me3, H3K36me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me1
		breast (HMEC)	H3K4me2, H3K36me3, DNase1, H3K27ac, H3K4me3, H3K9ac
		cervix (HeLa-S3)	DNase1, H3K36me3, PolII, Cjun, H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Cfos
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	Cjun, DNase1, H3K4me2, H3K4me3, H3K9ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001305768	1	embryonic stem cell (H1ESC)	USF1, DNase1
(CTCF binding site)		HSMMtube	H3K36me3
		blood (K562)	PolII, Brg1
		skin (NHDF-AD)	H3K36me3
		muscle (HSMM)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	PolII, H3K36me3

skin (NHEK)	PolII, H3K36me3
NHLF	H3K36me3
Osteobl	H3K36me3
blood (DND-41)	H3K36me3
breast (HMEC)	H3K36me3
cervix (HeLa-S3)	H3K36me3, PolII
monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
endothelium (HUVEC)	H3K36me3
A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANXA1	upstream gene variant, downstream gene variant	744	ENST00000257497	NM_000700.1	ENSP00000257497	5
ANXA1	upstream gene variant, downstream gene variant	2167	ENST00000415424	?	ENSP00000414013	5
ANXA1	downstream gene variant	744	ENST00000376911	?	ENSP00000366109	3
ANXA1	upstream gene variant, downstream gene variant	169	ENST00000456643	?	ENSP00000412489	4
ANXA1	upstream gene variant, downstream gene variant	744	ENST00000491192	?	?	5
ANXA1	upstream gene variant, downstream gene variant	2216	ENST00000468639	?	?	5
ANXA1	downstream gene variant	3268	ENST00000495713	?	?	1

Putative effect on transcript

5'-UTR variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
ANXA1	ENST00000257497	NM_000700.1	ENSP00000257497	1
ANXA1	ENST00000415424	?	ENSP00000414013	1
ANXA1	ENST00000456643	?	ENSP00000412489	1

Non-coding exon variant (splice region)

gene	affected transcript	RefSeq id	variant(s)
ANXA1	ENST00000468639	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANXA1	ENST00000257497	NM_000700.1	ENSP00000257497	3
ANXA1	ENST00000376911	?	ENSP00000366109	3
ANXA1	ENST00000456643	?	ENSP00000412489	1
ANXA1	ENST00000495713	?	?	3
ANXA1	ENST00000489109	?	?	3

Locus 437 (rs7260110)

SNiPAcard

Block annotations

Block info

genomic range	chr19:14,500,714-14,504,025
block size	3,312 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.292 [0.197 – 0.445]	gene(s) hit or close-by	ADGRE5
phastCons	μ = 0.166 [0.06 – 0.337]	eQTL gene(s)	–
GERP++	μ = 0.982 [0.113 – 2.33]	potentially regulated gene(s)	–
CADD score	μ = 7.166 [1.559 – 16.86]	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ADGRE5	downstream gene variant	2016	ENST00000587606	?	ENSP00000465800	1
ADGRE5	upstream gene variant	4267	ENST00000586849	?	?	1
ADGRE5	upstream gene variant	3037	ENST00000591737	?	?	1
ADGRE5	upstream gene variant	4201	ENST00000593028	?	?	1
ADGRE5	upstream gene variant	4520	ENST00000587319	?	?	1
ADGRE5	upstream gene variant	4520	ENST00000590567	?	?	1
ADGRE5	downstream gene variant	1721	ENST00000591080	?	?	1
ADGRE5	upstream gene variant	4520	ENST00000592341	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ADGRE5	ENST00000358600	NM_001784.4	ENSP00000351413	3
ADGRE5	ENST00000586517	?	ENSP00000467351	3
ADGRE5	ENST00000587728	?	?	3
ADGRE5	ENST00000242786	NM_078481.3	ENSP00000242786	3
ADGRE5	ENST00000591080	?	?	2
ADGRE5	ENST00000357355	NM_001025160.2	ENSP00000349918	3
ADGRE5	ENST00000592261	?	ENSP00000467478	3
ADGRE5	ENST00000587606	?	ENSP00000465800	2
ADGRE5	ENST00000587535	?	?	3

Locus 438 (rs12241254)

SNiPAcard

Block annotations

Block info

genomic range	chr10:101,329,158-101,329,158
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-2.395	gene(s) hit or close-by	–
phastCons	0	eQTL gene(s)	DNMBP-AS1
GERP++	-2.75	potentially regulated gene(s)	–
CADD score	1.456	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DNMBP-AS1	?	ENSG00000227695	cerebellum	1.44×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000360276 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3

Locus 439 (rs9811674)

SNiPAcard

Block annotations

Block info

genomic range	chr3:190,287,162-190,290,917
block size	3,756 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.081 [-0.149 – -0.013]	gene(s) hit or close-by	IL1RAP
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	–
GERP++	μ = 1.190 [0 – 2.38]	potentially regulated gene(s)	–
CADD score	μ = 6.082 [3.796 – 8.369]	disease gene(s)	–

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL1RAP	downstream gene variant	4856	ENST00000461629	?	?	1
IL1RAP	downstream gene variant	4955	ENST00000482913	?	?	1
IL1RAP	downstream gene variant	4705	ENST00000473566	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RAP	ENST00000317757	NM_001167931.1	ENSP00000314807	2
IL1RAP	ENST00000439062	NM_001167928.1	ENSP00000401132	2
IL1RAP	ENST00000342550	?	ENSP00000345829	2
IL1RAP	ENST00000453359	?	ENSP00000412008	2
IL1RAP	ENST00000072516	NM_001167929.1	ENSP00000072516	2
IL1RAP	ENST00000422940	NM_001167930.1	ENSP00000387371	2
IL1RAP	ENST00000434491	?	ENSP00000391899	2
IL1RAP	ENST00000422485	NM_134470.3	ENSP00000409352	2
IL1RAP	ENST00000447382	NM_002182.3	ENSP00000390541	2
IL1RAP	ENST00000422625	?	ENSP00000389149	2
IL1RAP	ENST00000413869	?	ENSP00000416296	2
IL1RAP	ENST00000412504	?	ENSP00000412053	2
IL1RAP	ENST00000443369	?	ENSP00000408893	2

Locus 440 (rs10503709)

SNiPAcard

Block annotations

Block info

genomic range	chr8:21,437,574-21,479,725
block size	42,152 bp
variant count	94 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.257 [-2.032 – 1.342]	gene(s) hit or close-by	–
phastCons	μ = 0.053 [0 – 0.999]	eQTL gene(s)	GFRA2
GERP++	μ = -0.220 [-5.17 – 3.73]	potentially regulated gene(s)	FAM160B2, FGF17, NPM2
CADD score	μ = 2.968 [0.028 – 20.2]	disease gene(s)	FGF17

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
erythronate*	4.06×10-5	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link
FGF17	HYPOGONADOTROPIC HYPOGONADISM 20 WITH OR WITHOUT ANOSMIA	OMIM	MIM:615270
FGF17	Kallmann Syndrome	OrphaNet	OrphaNet:478
FGF17	Normosmic congenital hypogonadotropic hypogonadism	OrphaNet	OrphaNet:432

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GFRA2	?	ENSG00000168546	gastroesophageal junction	1.84×10-16 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	muscularis mucosae	6.70×10-31 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	stomach	4.20×10-10 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	atrial appendage	2.58×10-12 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	sigmoid colon	4.15×10-12 (p-value)	GTEx Portal V6	93
GFRA2	?	ENSG00000168546	transverse colon	1.00×10-7 (p-value)	GTEx Portal V6	87
GFRA2	?	ENSG00000168546	esophagus mucosa	3.25×10-17 (p-value)	GTEx Portal V6	93

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000509140	1	ENCP00000055225	FGF17
		ENCP00000055220	NPM2

ENCE00000509287	1	ENCP00000055235	FAM160B2
		ENCP00000055236	FAM160B2
ENCE00000509141	1	ENCP00000055225	FGF17
		ENCP00000055220	NPM2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001439442 (open chromatin region)	1	A549	H3K4me3
ENSR00001720509 (open chromatin region)	4	embryonic stem cell (H1ESC)	Rad21
		A549	DNase1
		blood (K562)	H3K27me3
ENSR00001387343 (promoter flanking region)	1	cervix (HeLa-S3)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHEK)	H3K4me1, DNase1

Locus 441 (rs6662572)

SNiPAcard

Block annotations

Block info

genomic range	chr1:45,954,632-46,252,418
block size	297,787 bp
variant count	44 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.187 [-2.036 – 1.388]	gene(s) hit or close-by	AL355480.1, AL355480.3, AL355480.4, CCDC163P, CCDC17, GPBP1L1, IPP, MAST2, MMACHC, NASP, PRDX1, RP11-630I5.1, RP11-767N6.2, RP11-767N6.7, RPS15AP10, TESK2, TMEM69
phastCons	μ = 0.085 [0 – 0.978]	eQTL gene(s)	MAST2, MMACHC, PIK3R3, PRDX1, TMEM69
GERP++	μ = -0.131 [-3.98 – 2.7]	potentially regulated gene(s)	AKR1A1, GPBP1L1, HPDL, MAST2, PIK3R3, TESK2, TOE1
CADD score	μ = 3.765 [0.207 – 17.61]	disease gene(s)	MMACHC

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Blood metabolite levels	$<5.00\times10^{-11}$	GWAS Catalog	24816252	1
erythritol	4.99×10^{-11}	Metabolomics GWAS Server	24816252	7

Disease gene annotation

gene	trait	source DB	source entry/link
MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE	OMIM	MIM:277400
MMACHC	HOMOCYSTEINEMIA	OMIM	MIM:603174
MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE	DECIPHER	MIM:277400
MMACHC	Methylmalonic acidemia with homocystinuria, type cblC	OrphaNet	OrphaNet:79282

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TMEM69	?	ENSG00000159596	transformed fibroblasts	1.37×10^{-8} (p-value)	GTEx Portal V6	41
MAST2	?	ENSG00000086015	transformed fibroblasts	3.76×10^{-8} (p-value)	GTEx Portal V6	40
PIK3R3	?	ENSG00000117461	transformed fibroblasts	2.79×10^{-5} (p-value)	GTEx Portal V6	1
MAST2	?	ENSG00000086015	tibial artery	7.72×10^{-10} (p-value)	GTEx Portal V6	41
PRDX1	?	ENSG00000117450	tibial artery	8.10×10^{-7} (p-value)	GTEx Portal V6	27
PRDX1	?	ENSG00000117450	thyroid	1.86×10^{-10} (p-value)	GTEx Portal V6	40
MMACHC	?	ENSG00000132763	subcutaneous adipocytes	4.36×10^{-8} (p-value)	GTEx Portal V6	38
MAST2	?	ENSG00000086015	left ventricle	4.91×10^{-7} (p-value)	GTEx Portal V6	36
MMACHC	ENST00000616135	ILMN_1803005	blood	1.08×10^{-6} (p-value)	MuTHER consortium	8
MMACHC	ENST00000401061		adipocyte	4.91×10^{-10} (p-value)	MuTHER consortium	8

MMACHC	ENST00000477188					
PRDX1	ENST00000372079	ILMN_2366388	monocyte	6.74×10-4 (p-value)	Fairfax et al.	1
PRDX1	ENST00000319248					
PRDX1	ENST00000262746					
MAST2	?	ENSG00000086015	tibial nerve	4.13×10-6 (p-value)	GTEx Portal V6	4

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000017982	1	ENCP00000002314	GPBP1L1
		ENCP00000002293	TESK2
		ENCP00000002331	MAST2
		ENCP00000002305	AKR1A1
		ENCP00000002336	MAST2
ENCE00000018297	1	ENCP00000002340	PIK3R3
ENCE00000017914	1	ENCP00000002318	GPBP1L1
		ENCP00000002286	HPDL
ENCE00000017958	1	ENCP00000002291	TOE1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000669883 (promoter)	1	embryonic stem cell (H1ESC)	H3K79me2, H4K5ac, H3K36me3, H3K27me3, CTCF, PolII, Rad21, TAF7, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K79me2, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K79me2, HEY1, H2AZ, TAF1, ELF1, PolII, GTF2B, H4K20me1, H3K4me2, H3K36me3, DNase1, H3K4me3, Cmyc, Gabp, Yy1, Egr1, H3K27ac, Max, E2F6, H3K9ac
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H2AZ, H3K79me2, H3K4me3, H3K27ac
		liver (HepG2)	H3K4me3, DNase1, H3K27ac, H3K9ac, Gabp, Yy1, H4K20me1, PolII, H3K79me2, TAF1, ELF1, H2AZ, H3K4me2
		blood (GM12878)	H3K9ac, H3K4me2, Cmyc, Max, PolII, H2AZ, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac
		lung (IMR90)	DNase1, H3K79me2, H3K18ac, H3K4me2, H3K4me3, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K9ac, H3K4me3, H3K36me3, H4K20me1, H3K27ac, H3K4me1, H3K4me2
		breast (HMEC)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	DNase1, Max, H3K4me3, H3K4me2, H3K9ac, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001519843 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2, H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00000535923 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3, Yy1
		HSMMtube	H3K79me2

		Osteobl	H3K4me2
		blood (K562)	H3K79me2, H4K20me1
		blood (DND-41)	H4K20me1, H3K36me3
		muscle (HSMM)	H3K79me2
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K4me2
		liver (HepG2)	H4K20me1, H3K79me2, H3K4me1
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
ENSR00000282137 (enhancer)	1	blood (K562)	DNase1
ENSR00000165101 (enhancer)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K4me1
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL355480.1	downstream gene variant, upstream gene variant	2577	ENST00000629041	?	?	2
AL355480.3	downstream gene variant, upstream gene variant	2250	ENST00000630128	?	?	2
AL355480.4	downstream gene variant, upstream gene variant	277	ENST00000626823	?	?	3
CCDC163P	downstream gene variant	4906	ENST00000626177	?	ENSP00000485784	1
CCDC163P	downstream gene variant	4966	ENST00000625766	?	ENSP00000486505	1
CCDC163P	downstream gene variant	4977	ENST00000629009	?	?	1
CCDC17	downstream gene variant, upstream gene variant	1512	ENST00000445048	?	ENSP00000411335	3
CCDC17	downstream gene variant, upstream gene variant	1333	ENST00000372044	?	ENSP00000361114	3
CCDC17	downstream gene variant, upstream gene variant	1642	ENST00000479529	?	ENSP00000437072	3
CCDC17	downstream gene variant, upstream gene variant	1334	ENST00000421127	NM_001190182.1	ENSP00000389415	3
CCDC17	downstream gene variant, upstream gene variant	337	ENST00000464739	?	?	4
CCDC17	downstream gene variant, upstream gene variant	1400	ENST00000528266	NM_001114938.2	ENSP00000432172	3
CCDC17	downstream gene variant, upstream gene variant	1333	ENST00000482416	?	?	3
CCDC17	downstream gene variant, upstream gene variant	1333	ENST00000491755	?	?	3
CCDC17	downstream gene variant, upstream gene variant	1624	ENST00000525599	?	?	4
GPBP1L1	downstream gene variant	4062	ENST00000480941	?	?	1
GPBP1L1	downstream gene variant	611	ENST00000290795	?	ENSP00000290795	1
GPBP1L1	downstream gene variant	611	ENST00000479235	?	?	1
GPBP1L1	downstream gene variant, upstream gene variant	309	ENST00000496278	?	?	3
GPBP1L1	downstream gene variant	3949	ENST00000488278	?	?	1
GPBP1L1	downstream gene variant, upstream gene variant	602	ENST00000467032	?	?	2
GPBP1L1	downstream gene variant	4330	ENST00000460859	?	?	1
GPBP1L1	upstream gene variant	1222	ENST00000498128	?	?	1
GPBP1L1	downstream gene variant	2895	ENST00000468724	?	?	1
GPBP1L1	downstream gene variant, upstream gene variant	2287	ENST00000495616	?	?	3
GPBP1L1	downstream gene variant	613	ENST00000355105	NM_021639.4	ENSP00000347224	1

GPBP1L1	downstream gene variant	1535	ENST00000487436	?	?	1
GPBP1L1	downstream gene variant	4209	ENST00000493083	?	?	1
GPBP1L1	downstream gene variant	4081	ENST00000480083	?	?	1
IPP	downstream gene variant, upstream gene variant	863	ENST00000495072	?	?	3
IPP	upstream gene variant	1171	ENST00000359942	NM_001145349.1	ENSP00000353024	1
IPP	downstream gene variant	863	ENST00000461718	?	?	2
IPP	downstream gene variant, upstream gene variant	906	ENST00000396478	NM_005897.2	ENSP00000379739	4
MAST2	upstream gene variant	241	ENST00000470809	?	?	3
MMACHC	downstream gene variant	1508	ENST00000477188	?	?	1
MMACHC	downstream gene variant	948	ENST00000616135	?	ENSP00000478859	1
NASP	upstream gene variant, downstream gene variant	2319	ENST00000527932	?	?	4
NASP	downstream gene variant	2628	ENST00000527359	?	ENSP00000436790	2
NASP	downstream gene variant	2571	ENST00000528238	?	ENSP00000432289	2
NASP	downstream gene variant	3917	ENST00000525515	?	ENSP00000436939	2
NASP	upstream gene variant, downstream gene variant	467	ENST00000530073	?	?	3
NASP	downstream gene variant	2903	ENST00000437901	?	ENSP00000400792	2
NASP	downstream gene variant	3447	ENST00000464190	?	?	1
NASP	downstream gene variant	2903	ENST00000437362	?	ENSP00000388010	2
NASP	downstream gene variant	2594	ENST00000527470	?	ENSP00000437241	1
NASP	downstream gene variant	2463	ENST00000534101	?	?	1
NASP	upstream gene variant, downstream gene variant	427	ENST00000481782	?	?	4
NASP	downstream gene variant	464	ENST00000351223	NM_152298.3	ENSP00000255121	2
NASP	upstream gene variant, downstream gene variant	498	ENST00000531612	?	ENSP00000437116	3
NASP	downstream gene variant	465	ENST00000372052	?	ENSP00000361122	2
NASP	downstream gene variant	3595	ENST00000528084	?	ENSP00000435363	1
NASP	downstream gene variant	465	ENST00000472408	?	?	2
NASP	upstream gene variant, downstream gene variant	464	ENST00000534450	?	ENSP00000434240	3
NASP	downstream gene variant	1763	ENST00000350030	NM_002482.3	ENSP00000255120	1
NASP	downstream gene variant	2632	ENST00000529333	?	ENSP00000436388	1
NASP	downstream gene variant	1753	ENST00000537798	NM_001195193.1	ENSP00000438871	1
NASP	downstream gene variant	991	ENST00000531532	?	ENSP00000436778	1
NASP	downstream gene variant	2155	ENST00000470768	?	ENSP00000436924	1
NASP	downstream gene variant	3729	ENST00000530840	?	ENSP00000435548	1
NASP	downstream gene variant	2680	ENST00000453748	?	ENSP00000403157	2
PRDX1	downstream gene variant, upstream gene variant	1419	ENST00000319248	NM_181697.2	ENSP00000361152	2
PRDX1	downstream gene variant, upstream gene variant	3540	ENST00000483583	?	?	2
PRDX1	downstream gene variant, upstream gene variant	1408	ENST00000262746	NM_181696.2, NM_002574.3, NM_001202431.1	ENSP00000262746	2

PRDX1	downstream gene variant, upstream gene variant	2360	ENST00000424390 ?	ENSP00000389047	3
PRDX1	downstream gene variant, upstream gene variant	1423	ENST00000372079 ?	ENSP00000361150	2
PRDX1	downstream gene variant, upstream gene variant	2494	ENST00000447184 ?	ENSP00000407034	3
RP11-630I5.1	upstream gene variant, downstream gene variant	1526	ENST00000463059 ?	?	4
RP11-767N6.2	upstream gene variant	895	ENST00000437848 ?	?	3
RP11-767N6.7	upstream gene variant	730	ENST00000430643 ?	?	1
RPS15AP10	downstream gene variant	394	ENST00000432472 ?	?	1
TMEM69	downstream gene variant	1819	ENST00000496366 ?	?	3
TMEM69	downstream gene variant	1221	ENST00000372025 NM_016486.3	ENSP00000361095	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CCDC17	ENST00000479529	?	ENSP00000437072	T	acA/acG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCDC17	ENST00000528266	NM_001114938.2	ENSP00000432172	1
CCDC17	ENST00000491755	?	?	1
CCDC17	ENST00000421127	NM_001190182.1	ENSP00000389415	1
CCDC17	ENST00000445048	?	ENSP00000411335	1
CCDC17	ENST00000372044	?	ENSP00000361114	1
CCDC17	ENST00000482416	?	?	1
GPBP1L1	ENST00000480083	?	?	5
GPBP1L1	ENST00000488278	?	?	5
GPBP1L1	ENST00000493083	?	?	5
GPBP1L1	ENST00000498128	?	?	3
GPBP1L1	ENST00000460859	?	?	5
GPBP1L1	ENST00000468724	?	?	1
GPBP1L1	ENST00000290795	?	ENSP00000290795	10
GPBP1L1	ENST00000479235	?	?	1
GPBP1L1	ENST00000487436	?	?	1
GPBP1L1	ENST00000480941	?	?	5
GPBP1L1	ENST00000355105	NM_021639.4	ENSP00000347224	10
IPP	ENST00000396478	NM_005897.2	ENSP00000379739	10
IPP	ENST00000359942	NM_001145349.1	ENSP00000353024	13
IPP	ENST00000461718	?	?	1
IPP	ENST00000495072	?	?	2
MMACHC	ENST00000616135	?	ENSP00000478859	1
MMACHC	ENST00000477188	?	?	1

NASP	ENST00000481782	?	?	1
NASP	ENST00000528084	?	ENSP00000435363	3
NASP	ENST00000351223	NM_152298.3	ENSP00000255121	5
NASP	ENST00000530840	?	ENSP00000435548	2
NASP	ENST00000453748	?	ENSP00000403157	4
NASP	ENST00000531612	?	ENSP00000437116	1
NASP	ENST00000437901	?	ENSP00000400792	4
NASP	ENST00000525515	?	ENSP00000436939	4
NASP	ENST00000470768	?	ENSP00000436924	3
NASP	ENST00000372052	?	ENSP00000361122	5
NASP	ENST00000472408	?	?	1
NASP	ENST00000528238	?	ENSP00000432289	4
NASP	ENST00000534450	?	ENSP00000434240	1
NASP	ENST00000527359	?	ENSP00000436790	4
NASP	ENST00000531532	?	ENSP00000436778	2
NASP	ENST00000537798	NM_001195193.1	ENSP00000438871	5
NASP	ENST00000350030	NM_002482.3	ENSP00000255120	5
NASP	ENST00000437362	?	ENSP00000388010	4
NASP	ENST00000629893	?	ENSP00000486336	1
NASP	ENST00000534101	?	?	3
NASP	ENST00000529333	?	ENSP00000436388	3
NASP	ENST00000527470	?	ENSP00000437241	3
NASP	ENST00000464190	?	?	3
NASP	ENST00000530073	?	?	2
PRDX1	ENST00000319248	NM_181697.2	ENSP00000361152	1
PRDX1	ENST00000262746	NM_181696.2, NM_002574.3, NM_001202431.1	ENSP00000262746	1
PRDX1	ENST00000372079	?	ENSP00000361150	1
RP11-767N6.7	ENST00000430643	?	?	2
TESK2	ENST00000372086	NM_007170.2	ENSP00000361158	1
TESK2	ENST00000486676	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MMACHC	ENST00000401061	NM_015506.2	ENSP00000383840	2
NASP	ENST00000537798	NM_001195193.1	ENSP00000438871	1
NASP	ENST00000350030	NM_002482.3	ENSP00000255120	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NASP	ENST00000530073	?	1

Locus 442 (rs7857390)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,128,546-136,147,295
block size	18,750 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.281 [-2.374 – 0.721]	gene(s) hit or close-by	ABO
phastCons	μ = 0.112 [0 – 0.656]	eQTL gene(s)	ABO, GBGT1, SLC2A6, SURF1
GERP++	μ = 0.030 [-2.56 – 1.11]	potentially regulated gene(s)	–
CADD score	μ = 2.951 [0.029 – 13.47]	disease gene(s)	SURF1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
ADSGEGDFXAEGGGVR*	1.17×10-6	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link
SURF1	LEIGH SYNDROME	OMIM	MIM:256000
SURF1	Complex IV deficiency	DECIPHER	MIM:220110
SURF1	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
SURF1	Fatal infantile cytochrome C oxidase deficiency	OrphaNet	OrphaNet:1561
SURF1	Leigh syndrome with cardiomyopathy	OrphaNet	OrphaNet:70474
SURF1	Leigh syndrome with leukodystrophy	OrphaNet	OrphaNet:255241
SURF1	SURF1-related Charcot-Marie-Tooth disease type 4	OrphaNet	OrphaNet:391351

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SURF1	?	ENSG00000148290	sun exposed skin	3.29×10-6 (p-value)	GTEx Portal V6	12
ABO	?	ENSG00000175164	esophagus mucosa	2.75×10-7 (p-value)	GTEx Portal V6	12
ABO	?	ENSG00000175164	skeletal muscle	4.13×10-6 (p-value)	GTEx Portal V6	7
GBGT1	ENST00000372040	ILMN_1652906	blood	2.77×10-6 (p-value)	Westra et al.	2
GBGT1	ENST00000470431					
GBGT1	ENST00000472281					
GBGT1	ENST00000372043					
GBGT1	ENST00000540636					
SLC2A6	ENST00000371899	ILMN_1778321	blood	7.27×10-5 (p-value)	Westra et al.	2
SLC2A6	ENST00000485978					
SLC2A6	ENST00000371897					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001475637 (TF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K36me3, H3K4me2, H3K9ac, H3K27ac, DNase1, Brg1, Ini1, PolII
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABO	downstream gene variant	2443	ENST00000538324	?	ENSP00000483018	1
ABO	downstream gene variant	2017	ENST00000611156	NM_020469.2	ENSP00000483265	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000453660	?	?	12
ABO	ENST00000538324	?	ENSP00000483018	12
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	12

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ABO	ENST00000453660	?	1

Locus 443 (rs7610301)

SNiPAcard

Block annotations

Block info

genomic range	chr3:46,641,719-46,647,381
block size	5,663 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.535 [-2.28 – 0.735]	gene(s) hit or close-by	TDGF1
phastCons	μ = 0.164 [0 – 0.712]	eQTL gene(s)	ATRIP, PRSS46, TDGF1, TREX1, TREX1
GERP++	μ = -0.251 [-1.53 – 0.559]	potentially regulated gene(s)	AC104304.1, CCR2, CCR3, LRRC2, PRSS44, PRSS50
CADD score	μ = 4.290 [0.163 – 14.08]	disease gene(s)	TDGF1, TREX1, TREX1, CCR2, ATRIP

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TDGF1	TERATOCARCINOMA-DERIVED GROWTH FACTOR 1	OMIM	MIM:187395
TREX1	VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY	OMIM	MIM:192315
TREX1	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
TREX1	CHILBLAIN LUPUS 1	OMIM	MIM:610448
TREX1	AICARDI-GOUTIERES SYNDROME 1	OMIM	MIM:225750
TREX1	VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY	OMIM	MIM:192315
TREX1	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
TREX1	CHILBLAIN LUPUS 1	OMIM	MIM:610448
TREX1	AICARDI-GOUTIERES SYNDROME 1	OMIM	MIM:225750
CCR2	HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, SUSCEPTIBILITY TO	OMIM	MIM:609423
TDGF1	Semilobar holoprosencephaly	OrphaNet	OrphaNet:220386
TDGF1	Alobar holoprosencephaly	OrphaNet	OrphaNet:93925
TDGF1	Microform holoprosencephaly	OrphaNet	OrphaNet:280200
TDGF1	Midline interhemispheric variant of holoprosencephaly	OrphaNet	OrphaNet:93926
TDGF1	Septopreoptic holoprosencephaly	OrphaNet	OrphaNet:280195
TDGF1	Lobar holoprosencephaly	OrphaNet	OrphaNet:93924
ATRIP	Seckel syndrome	OrphaNet	OrphaNet:808
TREX1	Aicardi-Goutieres Syndrome	OrphaNet	OrphaNet:51
TREX1	HERNS syndrome	OrphaNet	OrphaNet:63261
TREX1	CHILBLAIN LUPUS	OrphaNet	OrphaNet:90280
TREX1	Cerebroretinal vasculopathy	OrphaNet	OrphaNet:3421
TREX1	Hereditary vascular retinopathy	OrphaNet	OrphaNet:71291
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive	DECIPHER	MIM:225750
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive	DECIPHER	MIM:225750

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PRSS46	?	ENSG00000261603	muscularis mucosae	1.66×10-6 (p-value)	GTEx Portal V6	19
TDGF1	?	ENSG00000241186	skeletal muscle	1.34×10-6 (p-value)	GTEx Portal V6	16

TREX1	ENST00000436480	ILMN_2370464	b-cell	4.74×10-4 (p-value)	Fairfax et al.	1
ATRIP	ENST00000320211					
ATRIP	ENST00000346691					
TREX1	ENST00000395677					
TREX1	ENST00000296443					
TREX1	ENST00000424906					
ATRIP	ENST00000412052					
TREX1	ENST00000433541					
TREX1	ENST00000629913					
TREX1	ENST00000357105					
TREX1	ENST00000625293					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000337725	1	ENCP00000036261	LRRC2
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036262	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036244	CCR3
		ENCP00000036274	PRSS50
		ENCP00000036273	PRSS50

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001674640 (enhancer)	4	HSMMtube	H3K27me3
		blood (K562)	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TDGF1	ENST00000505797	?	?	20

Locus 444 (rs12079353)

SNiPAcard

Block annotations

Block info

genomic range	chr1:159,530,662-159,582,487
block size	51,826 bp
variant count	47 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.526 [-3.513 – 0.931]	gene(s) hit or close-by	APCS, OR10AE1P, OR10J6P, RP11-740C1.2
phastCons	μ = 0.043 [0 – 0.289]	eQTL gene(s)	CXCL10, CXCL11
GERP++	μ = -1.045 [-7.3 – 2.62]	potentially regulated gene(s)	APCS, CADM3, IGSF8, OR10J1, OR10J2P, OR10J5, OR10J6P, OR10J8P, RP11-550P17.5, SLAMF8
CADD score	μ = 2.474 [0.219 – 7.616]	disease gene(s)	–

Direct effect on regulation

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CXCL10	?	ENSG00000169245	chr4	brain	5.31×10^{-5} (q-value)	SeeQTL DB (Myers et al.)	1
CXCL11	?	ENSG00000169248	chr4	brain	7.53×10^{-6} (q-value)	SeeQTL DB (Myers et al.)	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000040241	1	ENCP00000005063	OR10J1
		ENCP00000005060	OR10J8P
		ENCP00000005066	OR10J5
		ENCP00000005055	OR10J2P
ENCE00000040220	1	ENCP00000005069	OR10J6P
		ENCP00000005065	RP11-550P17.5
		ENCP00000005076	SLAMF8
ENCE00000040180	1	ENCP00000005050	CADM3
		ENCP00000005068	APCS
		ENCP00000005097	IGSF8

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000546120 (enhancer)	3	endothelium (HUVEC)	DNase1
		blood (DND-41)	H3K27me3
ENSR00001525176 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K27me3
		muscle (HSMM)	H3K27ac, H3K4me2, H3K9ac, DNase1
		cervix (HeLa-S3)	Cjun
		endothelium (HUVEC)	Cmyc, H3K27ac, H3K4me1, Cjun, DNase1
		lung (IMR90)	DNase1, H3K4ac, H2BK120ac, H4K91ac

A549	DNase1, H3K27ac, H3K4me3
nervous (NH-A)	H3K9ac, H3K27ac, DNase1
skin (NHEK)	DNase1, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
APCS	upstream gene variant, downstream gene variant	100	ENST00000255040	NM_001639.3	ENSP00000255040	8
OR10AE1P	downstream gene variant, upstream gene variant	112	ENST00000606962	?	?	13
OR10J6P	upstream gene variant, downstream gene variant	118	ENST00000289731	?	?	8
RP11-740C1.2	downstream gene variant	2672	ENST00000435442	?	?	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
APCS	ENST00000255040	NM_001639.3	ENSP00000255040	P	ccA/ccG	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
OR10AE1P	ENST00000606962	?	1

Locus 445 (rs4453827)

SNiPAcard

Block annotations

Block info

genomic range	chr3:53,741,287-53,789,698
block size	48,412 bp
variant count	25 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.528 [-3.374 – 1.263]	gene(s) hit or close-by	CACNA1D
phastCons	μ = 0.022 [0 – 0.396]	eQTL gene(s)	CHDH, IL17RB
GERP++	μ = -1.717 [-8.88 – 4.79]	potentially regulated gene(s)	DCP1A
CADD score	μ = 3.756 [0.136 – 16.38]	disease gene(s)	CACNA1D

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities	DECIPHER	MIM:615474
CACNA1D	Sinoatrial node dysfunction and deafness	DECIPHER	MIM:614896
CACNA1D	SINOATRIAL NODE DYSFUNCTION AND DEAFNESS	OMIM	MIM:614896
CACNA1D	PRIMARY ALDOSTERONISM, SEIZURES, AND NEUROLOGIC [...]	OMIM	MIM:615474
CACNA1D	Sinoatrial node dysfunction and deafness	OrphaNet	OrphaNet:324321
CACNA1D	Aldosterone-producing adenoma with seizures and neurological abnormalities	OrphaNet	OrphaNet:369929

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CHDH	?	ENSG00000016391	tibial nerve	4.41×10^{-8} (p-value)	GTEx Portal V6	24
IL17RB	?	ENSG00000056736	tibial nerve	3.84×10^{-9} (p-value)	GTEx Portal V6	24

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000340690	1	ENCP00000036821	DCP1A

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001479254 (enhancer)	3	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001364519 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3
		blood (GM12878)	H3K4me1, H3K27ac, H3K4me2
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CACNA1D	downstream gene variant	2904	ENST00000481085	?	ENSP00000418045	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CACNA1D	ENST00000350061	NM_001128840.2	ENSP00000288133	25
CACNA1D	ENST00000288139	NM_000720.3	ENSP00000288139	25
CACNA1D	ENST00000481085	?	ENSP00000418045	16
CACNA1D	ENST00000422281	NM_001128839.2	ENSP00000409174	25
CACNA1D	ENST00000481478	?	ENSP00000418014	25

Locus 446 (rs8051517)

SNiPAcard

Block annotations

Block info

genomic range	chr16:68,428,089-68,428,089
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.856	gene(s) hit or close-by	SMPD3
phastCons	0	eQTL gene(s)	ATP6V0D1, DUS2, TRADD
GERP++	-1.04	potentially regulated gene(s)	–
CADD score	2.866	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DUS2	?	ENSG00000167264	blood	7.29×10^{-6} (p-value)	GTEx Portal V6	1
DUS2	ENST00000432752	ILMN_1811650	monocyte	3.56×10^{-14} (p-value)	Zeller et al.	1
DUS2	ENST00000565263					
DUS2	ENST00000561965					
DUS2	ENST00000565980					
DUS2	ENST00000358896					
ATP6V0D1	ENST00000568620	ILMN_1795826	monocyte	6.15×10^{-13} (p-value)	Zeller et al.	1
ATP6V0D1	ENST00000563064					
ATP6V0D1	ENST00000567694					
ATP6V0D1	ENST00000602876					
ATP6V0D1	ENST00000561852					
ATP6V0D1	ENST00000290949					
ATP6V0D1	ENST00000567170					
ATP6V0D1	ENST00000564615					
ATP6V0D1	ENST00000565835					
ATP6V0D1	ENST00000426604					
ATP6V0D1	ENST00000563305					
TRADD	ENST00000486556	ILMN_1765851	monocyte	4.94×10^{-13} (p-value)	Zeller et al.	1
TRADD	ENST00000566247					
TRADD	ENST00000563348					
TRADD	ENST00000345057					

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SMPD3	ENST00000561749	?	ENSP00000457236	1
SMPD3	ENST00000566723	?	?	1
SMPD3	ENST00000219334	NM_018667.3	ENSP00000219334	1

Locus 447 (rs1634718)

SNiPAcard

Block annotations

Block info

genomic range	chr6:30,936,534-30,982,008
block size	45,475 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.217 [-2.088 – 2.249]	gene(s) hit or close-by	MUC21, MUC22
phastCons	μ = 0.021 [0 – 0.116]	eQTL gene(s)	C4A, CCHCR1, CYP21A1P, FLOT1, HCG22, HCP5, HLA-C, LINC00243, MICB, MUC22, TRIM31, VARS2
GERP++	μ = -0.341 [-8.65 – 1.76]	potentially regulated gene(s)	ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, CDSN, CDSN, CDSN, CDSN, CDSN, CDSN, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, GNL1, GNL1, GNL1, GNL1, GNL1, GNL1, GNL1, GNL1, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, PPP1R10, PPP1R10, PPP1R10, PPP1R10, PPP1R10, PPP1R10, PPP1R10, PRR3, PRR3, PRR3, PRR3, PRR3, PRR3, PRR3, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG248L24.13, XXbac-BPG299F13.16
CADD score	μ = 5.289 [1.236 – 12.86]	disease gene(s)	C4A, VARS2, , CDSN

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Lupus erythematosus, systemic	2.64×10^{-8}	dbGaP	pha002867	dbGaP	2

Disease gene annotation

gene	trait	source DB	source entry/link	
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380	OMIM
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790	OMIM
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
CDSN	Hypotrichosis simplex of the scalp	OrphaNet	OrphaNet:90368	orphanet
CDSN	Generalized peeling skin syndrome type B	OrphaNet	OrphaNet:263553	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TRIM31	?	ENSG00000204616	lung	3.73×10^{-6} (p-value)	GTEx Portal V6	3
C4A	?	ENSG00000244731	lung	1.14×10^{-7} (p-value)	GTEx Portal V6	9

HCG22	?	ENSG00000228789	coronary artery	1.07×10^{-6} (p-value)	GTEx Portal V6	6
LINC00243	?	ENSG00000214894	blood	2.13×10^{-9} (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	blood	2.49×10^{-7} (p-value)	GTEx Portal V6	4
MUC22	?	ENSG00000261272	esophagus mucosa	7.09×10^{-9} (p-value)	GTEx Portal V6	15
MICB	?	ENSG00000204516	esophagus mucosa	8.59×10^{-7} (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	skeletal muscle	7.95×10^{-8} (p-value)	GTEx Portal V6	9
C4A	?	ENSG00000244731	skeletal muscle	2.84×10^{-7} (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	cerebellum	6.88×10^{-8} (p-value)	GTEx Portal V6	15
CCHCR1	?	ENSG00000204536	transverse colon	4.24×10^{-8} (p-value)	GTEx Portal V6	9
CCHCR1	?	ENSG00000204536	subcutaneous adipocytes	1.68×10^{-6} (p-value)	GTEx Portal V6	7
HCG22	?	ENSG00000228789	subcutaneous adipocytes	2.19×10^{-6} (p-value)	GTEx Portal V6	5
HCG22	ENST00000570223	ILMN_1667229	blood	3.23×10^{-18} (p-value)	MuTHER consortium	7
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
MICB	ENST00000538442	ILMN_1708006	skin	1.20×10^{-7} (p-value)	MuTHER consortium	7
MICB	ENST00000399150					
MICB	ENST00000252229					
VARS2	ENST00000321897	ILMN_1737585	skin	2.72×10^{-5} (p-value)	MuTHER consortium	1
VARS2	ENST00000625423					
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
HCP5	ENST00000414046	ILMN_1803945	skin	3.52×10^{-5} (p-value)	MuTHER consortium	2
HCP5	ENST00000541196					
FLOT1	?	ENSG00000137312	thyroid	8.04×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	thyroid	4.85×10^{-6} (p-value)	GTEx Portal V6	1
?	?	ILMN_1679389	b-cell	3.75×10^{-4} (p-value)	Fairfax et al.	3
C4A	?	ENSG00000244731	atrial appendage	2.58×10^{-7} (p-value)	GTEx Portal V6	5
C4A	?	ENSG00000244731	tibial nerve	3.53×10^{-7} (p-value)	GTEx Portal V6	9
C4A	?	ENSG00000244731	putamen	2.07×10^{-7} (p-value)	GTEx Portal V6	7
C4A	?	ENSG00000244731	left ventricle	3.19×10^{-7} (p-value)	GTEx Portal V6	9
C4A	?	ENSG00000244731	muscularis mucosae	1.19×10^{-8} (p-value)	GTEx Portal V6	8
CYP21A1P	?	ENSG00000204338	muscularis mucosae	2.21×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	ENST00000470363	216526_x_at	blood	6.20×10^{-8} (p-value)	Dixon et al.	2
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					

HLA-C	ENST00000466892					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
?	?	ILMN_1893395	monocyte	8.28×10-18 (p-value)	Zeller et al.	2
?	?	ILMN_1883997	monocyte	7.04×10-18 (p-value)	Zeller et al.	2
C4A	?	ENSG00000244731	transformed fibroblasts	3.34×10-6 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	sun exposed skin	1.74×10-7 (p-value)	GTEx Portal V6	2
C4A	?	ENSG00000244731	unexposed skin	1.13×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	EBV lymphocytes	3.03×10-6 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	aorta	2.61×10-6 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	221491_x_at	chr6	blood	6.20×10-8 (p-value)	Dixon et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444432	1	ENCP00000047915	PPP1R10 PPP1R10 PPP1R10 PPP1R10 PPP1R10 PPP1R10 PPP1R10
		ENCP00000047967	VARS2 VARS2 VARS2 VARS2 VARS2 VARS2
		ENCP00000047911	PRR3 PRR3 PRR3 GNL1 GNL1 GNL1 PRR3 GNL1 PRR3 GNL1 GNL1 GNL1 PRR3 PRR3 GNL1
		ENCP00000047978	CDSN CDSN CDSN CDSN CDSN CDSN
		ENCP00000047992	HCG27 HCG27 HCG27

			HCG27 HCG27 HCG27 HCG27
		ENCP00000047974	XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9
		ENCP00000047923	ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1
		ENCP00000047975	HCG22 HCG22 HCG22 HCG22 HCG22 HCG22 HCG22 HCG22
		ENCP00000047995	XXbac-BPG299F13.16
		ENCP00000048019	MICB MICB MICB MICB MICB MICB MICB MICB
ENCE00000444471	1	ENCP00000048001	XXbac-BPG248L24.13
		ENCP00000047921	ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1
		ENCP00000047949	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
		ENCP00000047941	FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001703009 (open chromatin region)	2	blood (K562)	H3K27me3
ENSR00001703011 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	DNase1, H3K27me3
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		breast (HMEC)	CTCF, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	CTCF, H3K27me3
		lung (IMR90)	H3K4me2
		nervous (NH-A)	H3K4me2, DNase1
ENSR00001494536 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, H3K27me3, Rad21
		HSMMtube	DNase1, H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	DNase1
		liver (HepG2)	CTCF, H3K27me3
		blood (GM12878)	H3K27me3
		lung (IMR90)	H3K4me2, H3K27me3
		nervous (NH-A)	H3K4me2, DNase1
		NHLF	DNase1
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MUC21	upstream gene variant	475	ENST00000376296	NM_001010909.2	ENSP00000365473	2
MUC21	upstream gene variant, downstream gene variant	475	ENST00000486149	?	ENSP00000457640	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
MUC21	ENST00000376296	NM_001010909.2	ENSP00000365473	T	acA/acG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MUC22	ENST00000561890	NM_001198815.1	ENSP00000455906	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MUC21	ENST00000376296	NM_001010909.2	ENSP00000365473	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MUC21	ENST00000486149	?	ENSP00000457640	1

Locus 448 (rs1159787)

SNiPAcard

Block annotations

Block info

genomic range	chr9:120,327,400-120,341,514
block size	14,115 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.006 [-0.331 – 0.323]	**gene(s) hit or close-by**	–
phastCons	μ = 0.332 [0 – 0.99]	**eQTL gene(s)**	–
GERP++	μ = 0.340 [-1.25 – 2.12]	**potentially regulated gene(s)**	ASTN2
CADD score	μ = 5.697 [0.778 – 14.58]	**disease gene(s)**	–

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000552648	1	ENCP00000059825	ASTN2

Locus 449 (rs4239252)

SNiPAcard

Block annotations

Block info

genomic range	chr17:34,087,704-34,261,831
block size	174,128 bp
variant count	53 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.665 [-5.315 – 2.165]	**gene(s) hit or close-by**	7SK, AC015849.12, AC015849.13, AC015849.14, AC015849.16, AC015849.19, AC015849.2, C17orf50, CCL5, HEATR9, LRRC37A9P, LYZL6, MMP28, RDM1, RP1-161P9.5, TAF15
phastCons	μ = 0.071 [0 – 1]	**eQTL gene(s)**	CCL5
GERP++	μ = -0.087 [-6.19 – 4.15]	**potentially regulated gene(s)**	CCL23, CCL23, CCL5, CCL5, RP11-1094M14.1
CADD score	μ = 3.563 [0.191 – 22.3]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
N-acetylglycine	5.76×10^{-5}	Metabolomics GWAS Server	24816252	2
pyridoxate	9.63×10^{-5}	Metabolomics GWAS Server	24816252	3

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
C17orf50	missense variant	ENST00000604830	?	ENSP00000474618	D/E	gaC/gaG	?	unknown	1
C17orf50	missense variant	ENST00000603305	?	ENSP00000474048	R/G	Cgc/Ggc	?	unknown	1
C17orf50	missense variant	ENST00000605587	NM_145272.3	ENSP00000475146	D/E	gaC/gaG	tolerated	benign	1
LYZL6	missense variant	ENST00000618542	NM_001199951.1	ENSP00000482955	S/F	tCc/tTc	?	?	1
LYZL6	missense variant	ENST00000615905	NM_020426.2	ENSP00000483897	S/F	tCc/tTc	?	?	1
RDM1	missense variant	ENST00000615378	NM_001163125.1	ENSP00000480131	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000612980	NM_001163120.1	ENSP00000483387	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619828	NM_001163122.1	ENSP00000483933	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619876	?	ENSP00000483890	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000615288	?	ENSP00000477869	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619368	?	ENSP00000478131	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000615024	?	ENSP00000481648	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000617591	NM_001163124.1	ENSP00000479622	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000620284	NM_145654.3	ENSP00000483549	W/C	tgG/tgT	?	?	1

RDM1	missense variant	ENST00000616596	NM_001163121.1	ENSP00000478915	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619193	?	ENSP00000482981	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000613308	NM_001034836.1	ENSP00000482288	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000618511	?	ENSP00000477995	W/C	tgG/tgT	?	?	1
RDM1	missense variant	ENST00000619262	NM_001163130.1	ENSP00000479310	W/C	tgG/tgT	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ENSG00000187456	blood	1.75×10^{-5} (q-value)	SeeQTL DB (HapMap)	12
?	?	ENSG00000161570	blood	4.48×10^{-5} (q-value)	SeeQTL DB (HapMap)	7
CCL5	ENST00000603197	ILMN_2098126	blood	1.34×10^{-11} (p-value)	MuTHER consortium	22
CCL5	ENST00000605140					
CCL5	ENST00000603197	ILMN_1773352	blood	4.39×10^{-11} (p-value)	MuTHER consortium	22
CCL5	ENST00000605140					
CCL5	ENST00000605509					
?	?	241885_at	blood	1.00×10^{-9} (p-value)	Dixon et al.	7
CCL5	ENST00000603197	ILMN_2098126	blood	1.56×10^{-5} (p-value)	Westra et al.	12
CCL5	ENST00000605140					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1761314	chr20	monocyte	1.82×10^{-13} (p-value)	Zeller et al.	3

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000022616		CCL5

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000204092	1	ENCP00000022627	CCL23 CCL23
ENCE00000203983	1	ENCP00000022610	RP11-1094M14.1
		ENCP00000022616	CCL5 CCL5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001344848 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, Rad21, TAF7, CTCF, Yy1, H3K4me2, H3K4me3
		HSMMtube	CTCF, H3K27ac, H2AZ, DNase1
		blood (K562)	H3K4me3, DNase1, CTCF, H2AZ, Rad21, Max, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	H2AZ, CTCF, DNase1
		liver (HepG2)	H3K27me3, DNase1, CTCF, H3K4me2, H3K4me3, Rad21
		blood (GM12878)	DNase1, H2AZ, Rad21, H3K4me3, H3K4me2, CTCF

		lung (IMR90)	H3K4me2, H3K27me3, CTCF
		nervous (NH-A)	DNase1, H3K27me3, H3K4me2, CTCF
		skin (NHEK)	CTCF, DNase1, H3K4me2
		NHLF	DNase1, CTCF
		Osteobl	H3K4me2, CTCF, H3K27me3
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3, DNase1, CTCF, H3K4me2
		endothelium (HUVEC)	DNase1, Max, H3K4me2, CTCF
		A549	CTCF, DNase1, H3K4me2, H3K4me3
ENSR00001638253 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, Rad21, TAF7, CTCF, Yy1, H3K4me2
		HSMMtube	CTCF, H2AZ, DNase1
		blood (K562)	H3K4me3, DNase1, CTCF, H2AZ, Rad21, Max
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	H2AZ, CTCF
		liver (HepG2)	H3K4me3, Rad21, H3K4me2, CTCF, DNase1
		blood (GM12878)	DNase1, H2AZ, Rad21, H3K4me3, H3K4me2, CTCF
		lung (IMR90)	H3K4me2, CTCF
		nervous (NH-A)	CTCF, H3K4me2, DNase1
		skin (NHEK)	DNase1, CTCF, H3K4me2
		NHLF	DNase1, CTCF
		Osteobl	H3K4me2, CTCF
		blood (DND-41)	H3K9ac, H3K4me3, H3K27ac, CTCF, H3K4me1, H3K4me2
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27me3, H3K4me3
		endothelium (HUVEC)	Max, H3K4me2, CTCF, DNase1
		A549	DNase1, H3K4me3, H3K4me2, CTCF
ENSR00001344864 (promoter flanking region)	1	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	DNase1
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	PolII, DNase1
		lung (IMR90)	H3K27me3
ENSR00001344879 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1
		HSMMtube	H3K27me3
		blood (GM12878)	MEF2A, DNase1, H3K4me3, H3K27ac, PolII
		blood (DND-41)	H3K27me3
		blood (K562)	H3K4me1, H2AZ, DNase1
ENSR00001638269 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		blood (K562)	H2AZ

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
TAF15	1	ENST00000603067 ENST00000603393 ENST00000603427 ENST00000603777 ENST00000604360 ENST00000604841 ENST00000604879 ENST00000605197 ENST00000605649 ENST00000605844	hsa-miR-18a-5p

Variation proximal to gene

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
7SK	upstream gene variant, downstream gene variant	4434	ENST00000603504	?	?	2
AC015849.12	downstream gene variant, upstream gene variant	535	ENST00000605738	?	?	2
AC015849.13	upstream gene variant, downstream gene variant	1758	ENST00000603678	?	?	4
AC015849.14	downstream gene variant, upstream gene variant	447	ENST00000605454	?	?	3
AC015849.19	downstream gene variant, upstream gene variant	76	ENST00000603981	?	?	3
AC015849.2	upstream gene variant	438	ENST00000615709	?	?	2
AC015849.2	upstream gene variant	4565	ENST00000605548	?	?	1
C17orf50	upstream gene variant	212	ENST00000604830	?	ENSP00000474618	1
C17orf50	upstream gene variant	225	ENST00000605587	NM_145272.3	ENSP00000475146	1
C17orf50	upstream gene variant	224	ENST00000603305	?	ENSP00000474048	1
CCL5	upstream gene variant	268	ENST00000603197	NM_002985.2	ENSP00000474412	2
CCL5	upstream gene variant	485	ENST00000605509	NM_001278736.1	ENSP00000474141	2
CCL5	upstream gene variant	4331	ENST00000605140	?	ENSP00000475057	1
HEATR9	downstream gene variant, upstream gene variant	356	ENST00000604376	?	?	4
HEATR9	upstream gene variant	3798	ENST00000604400	?	?	2
HEATR9	upstream gene variant	3810	ENST00000603323	?	ENSP00000474391	2
HEATR9	upstream gene variant	3746	ENST00000605080	?	ENSP00000474893	2
HEATR9	upstream gene variant	3766	ENST00000603870	?	ENSP00000473760	2
HEATR9	upstream gene variant	3747	ENST00000603852	?	ENSP00000474899	2
HEATR9	downstream gene variant, upstream gene variant	2260	ENST00000603218	?	ENSP00000474337	3
HEATR9	upstream gene variant	3746	ENST00000604834	NM_152781.2	ENSP00000473941	2
LRRC37A9P	upstream gene variant	1228	ENST00000603850	?	?	8
LYZL6	downstream gene variant	1542	ENST00000615905	NM_020426.2	ENSP00000483897	7
LYZL6	downstream gene variant	1529	ENST00000618542	NM_001199951.1	ENSP00000482955	7
LYZL6	downstream gene variant	1862	ENST00000617692	?	ENSP00000481373	4
MMP28	downstream gene variant, upstream gene variant	1011	ENST00000605424	NM_024302.3	ENSP00000473853	2
MMP28	downstream gene variant, upstream gene variant	969	ENST00000615136	?	ENSP00000483299	2
MMP28	downstream gene variant, upstream gene variant	850	ENST00000612292	?	ENSP00000484562	2
MMP28	upstream gene variant	886	ENST00000612672	NM_001032278.1	ENSP00000483539	1
MMP28	upstream gene variant	793	ENST00000616383	?	?	1
MMP28	upstream gene variant	886	ENST00000619655	?	ENSP00000481051	1
MMP28	upstream gene variant	4410	ENST00000615317	?	ENSP00000482385	1
RDM1	upstream gene variant	721	ENST00000615024	?	ENSP00000481648	5
RDM1	downstream gene variant, upstream gene variant	704	ENST00000615378	NM_001163125.1	ENSP00000480131	12
RDM1	upstream gene variant	234	ENST00000619262	NM_001163130.1	ENSP00000479310	5
RDM1	upstream gene variant	276	ENST00000612980	NM_001163120.1	ENSP00000483387	5
RDM1	upstream gene variant	276	ENST00000616596	NM_001163121.1	ENSP00000478915	5
RDM1	upstream gene variant	721	ENST00000617591	NM_001163124.1	ENSP00000479622	5
RDM1	downstream gene variant, upstream gene variant	250	ENST00000619876	?	ENSP00000483890	12
RDM1	upstream gene variant	276	ENST00000615288	?	ENSP00000477869	5
RDM1	upstream gene variant	721	ENST00000619828	NM_001163122.1	ENSP00000483933	5

RDM1	upstream gene variant	721	ENST00000618511	?	ENSP00000477995	5
RDM1	upstream gene variant	276	ENST00000619193	?	ENSP00000482981	5
RDM1	upstream gene variant	230	ENST00000620284	NM_145654.3	ENSP00000483549	5
RDM1	upstream gene variant	276	ENST00000619368	?	ENSP00000478131	5
RDM1	downstream gene variant, upstream gene variant	263	ENST00000613308	NM_001034836.1	ENSP00000482288	12
RP1-161P9.5	upstream gene variant	2360	ENST00000603816	?	?	1
TAF15	upstream gene variant, downstream gene variant	1528	ENST00000604434	?	?	2
TAF15	upstream gene variant, downstream gene variant	664	ENST00000604360	?	?	4
TAF15	upstream gene variant, downstream gene variant	1552	ENST00000605197	?	?	3
TAF15	upstream gene variant	1501	ENST00000605844	NM_139215.2	ENSP00000474096	1
TAF15	upstream gene variant, downstream gene variant	197	ENST00000604879	?	ENSP00000475044	3
TAF15	upstream gene variant	3439	ENST00000603967	?	ENSP00000474047	1
TAF15	upstream gene variant	1530	ENST00000603777	?	ENSP00000474522	1
TAF15	upstream gene variant, downstream gene variant	2337	ENST00000603346	?	?	3
TAF15	downstream gene variant	2943	ENST00000603067	?	?	1
TAF15	upstream gene variant	2581	ENST00000604694	?	ENSP00000475147	1
TAF15	upstream gene variant	1923	ENST00000604195	?	?	1
TAF15	upstream gene variant	1535	ENST00000604841	NM_003487.3	ENSP00000474609	1
TAF15	upstream gene variant, downstream gene variant	1530	ENST00000605649	?	?	3
TAF15	upstream gene variant, downstream gene variant	703	ENST00000603427	?	ENSP00000474436	4
TAF15	upstream gene variant, downstream gene variant	1263	ENST00000603393	?	ENSP00000474653	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC015849.13	ENST00000603678	?	?	2
AC015849.16	ENST00000604647	?	?	1
AC015849.16	ENST00000604907	?	?	3
AC015849.19	ENST00000603981	?	?	4
AC015849.2	ENST00000605548	?	?	4
CCL5	ENST00000605140	?	ENSP00000475057	3
CCL5	ENST00000603197	NM_002985.2	ENSP00000474412	2
CCL5	ENST00000605509	NM_001278736.1	ENSP00000474141	2
HEATR9	ENST00000605080	?	ENSP00000474893	3
HEATR9	ENST00000603218	?	ENSP00000474337	1
HEATR9	ENST00000604400	?	?	3
HEATR9	ENST00000603852	?	ENSP00000474899	3
HEATR9	ENST00000604834	NM_152781.2	ENSP00000473941	3
HEATR9	ENST00000603870	?	ENSP00000473760	3
HEATR9	ENST00000603323	?	ENSP00000474391	3

MMP28	ENST00000619655	?	ENSP00000481051	4
MMP28	ENST00000612672	NM_001032278.1	ENSP00000483539	1
MMP28	ENST00000605424	NM_024302.3	ENSP00000473853	2
MMP28	ENST00000615317	?	ENSP00000482385	2
MMP28	ENST00000615136	?	ENSP00000483299	2
MMP28	ENST00000612292	?	ENSP00000484562	1
RDM1	ENST00000613308	NM_001034836.1	ENSP00000482288	10
RDM1	ENST00000619876	?	ENSP00000483890	10
RDM1	ENST00000612980	NM_001163120.1	ENSP00000483387	17
RDM1	ENST00000616596	NM_001163121.1	ENSP00000478915	17
RDM1	ENST00000615024	?	ENSP00000481648	17
RDM1	ENST00000615378	NM_001163125.1	ENSP00000480131	10
RDM1	ENST00000617591	NM_001163124.1	ENSP00000479622	17
RDM1	ENST00000619193	?	ENSP00000482981	17
RDM1	ENST00000619262	NM_001163130.1	ENSP00000479310	17
RDM1	ENST00000618511	?	ENSP00000477995	17
RDM1	ENST00000615288	?	ENSP00000477869	17
RDM1	ENST00000619368	?	ENSP00000478131	17
RDM1	ENST00000619828	NM_001163122.1	ENSP00000483933	17
RDM1	ENST00000620284	NM_145654.3	ENSP00000483549	17
TAF15	ENST00000605844	NM_139215.2	ENSP00000474096	12
TAF15	ENST00000604841	NM_003487.3	ENSP00000474609	12
TAF15	ENST00000604879	?	ENSP00000475044	7
TAF15	ENST00000604360	?	?	3
TAF15	ENST00000605649	?	?	4
TAF15	ENST00000603777	?	ENSP00000474522	12
TAF15	ENST00000603346	?	?	3
TAF15	ENST00000603067	?	?	17
TAF15	ENST00000603967	?	ENSP00000474047	5
TAF15	ENST00000604694	?	ENSP00000475147	2
TAF15	ENST00000605197	?	?	4
TAF15	ENST00000603393	?	ENSP00000474653	10
TAF15	ENST00000603427	?	ENSP00000474436	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TAF15	ENST00000603967	?	ENSP00000474047	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL5	ENST00000605140	?	ENSP00000475057	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

gene	affected transcript	RefSeq id	variant(s)
TAF15	ENST00000604195	?	1

Locus 450 (rs9462846)

SNiPAcard

Block annotations

Block info

genomic range	chr6:42,824,756-42,892,495
block size	67,740 bp
variant count	71 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.793 [-7.379 – 3.635]	gene(s) hit or close-by	C6orf226, CNPY3, GLTSCR1L, PTCRA, RPL7L1
phastCons	μ = 0.074 [0 – 1]	eQTL gene(s)	C6orf226, PEX6, RPL7L1, RPL7P21
GERP++	μ = -0.003 [-4.25 – 4.82]	potentially regulated gene(s)	CUL9, PRPH2, PTCRA, PTK7, RP11-480N24.4
CADD score	μ = 4.608 [0.014 – 25.7]	disease gene(s)	PEX6, PRPH2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PEX6	PEROXISOME BIOGENESIS DISORDER 4A (ZELLWEGER)	OMIM	MIM:614862
PEX6	PEROXISOME BIOGENESIS DISORDER 4B	OMIM	MIM:614863
PEX6	peroxisome biogenesis disorder complementation group 4 (PBD-CG4)	DECIPHER	MIM:601498
PEX6	Zellweger syndrome (ZWS)	DECIPHER	MIM:214100
PEX6	NEONATAL ADRENOLEUKODYSTROPHY	OrphaNet	OrphaNet:44
PEX6	Infantile Refsum disease	OrphaNet	OrphaNet:772
PEX6	ZELLWEGER SYNDROME	OrphaNet	OrphaNet:912
PRPH2	Butterfly-shaped pigment dystrophy	OrphaNet	OrphaNet:99001
PRPH2	Retinitis pigmentosa	OrphaNet	OrphaNet:791
PRPH2	Central areolar choroidal dystrophy	OrphaNet	OrphaNet:75377
PRPH2	Adult-onset foveomacular vitelliform dystrophy	OrphaNet	OrphaNet:99000
PRPH2	RETINITIS PUNCTATA ALBESCENS	OrphaNet	OrphaNet:52427
PRPH2	FUNDUS ALBIPUNCTATUS	OrphaNet	OrphaNet:227796
PRPH2	Cone-rod dystrophy	OrphaNet	OrphaNet:1872

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PTCRA	missense variant	ENST00000616441	NM_001243168.1	ENSP00000477815	I/V	Atc/Gtc	?	?	1
PTCRA	missense variant	ENST00000304672	NM_138296.2	ENSP00000304447	I/V	Atc/Gtc	?	?	1
PTCRA	missense variant	ENST00000441198	NM_001243169.1	ENSP00000409550	I/V	Atc/Gtc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RPL7L1	?	ENSG00000146223	muscularis mucosae	2.73×10-10 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	muscularis mucosae	7.85×10-6 (p-value)	GTEx Portal V6	5

RPL7L1	?	ENSG00000146223	lung	1.53×10-8 (p-value)	GTEx Portal V6	42
PEX6	?	ENSG00000124587	lung	2.54×10-12 (p-value)	GTEx Portal V6	66
RPL7L1	?	ENSG00000146223	transformed fibroblasts	4.14×10-24 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	transformed fibroblasts	2.36×10-10 (p-value)	GTEx Portal V6	61
RPL7L1	?	ENSG00000146223	tibial artery	9.04×10-17 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	tibial artery	5.99×10-9 (p-value)	GTEx Portal V6	33
RPL7L1	?	ENSG00000146223	breast	1.18×10-13 (p-value)	GTEx Portal V6	65
PEX6	?	ENSG00000124587	breast	2.13×10-7 (p-value)	GTEx Portal V6	23
RPL7L1	?	ENSG00000146223	blood	3.26×10-9 (p-value)	GTEx Portal V6	62
PEX6	?	ENSG00000124587	blood	2.64×10-9 (p-value)	GTEx Portal V6	33
RPL7L1	?	ENSG00000146223	thyroid	2.79×10-8 (p-value)	GTEx Portal V6	41
PEX6	?	ENSG00000124587	thyroid	8.77×10-9 (p-value)	GTEx Portal V6	33
PEX6	ENST00000304611	ILMN_1683279	monocyte	2.16×10-14 (p-value)	Fairfax et al.	9
PEX6	ENST00000244546		b-cell	9.71×10-13 (p-value)	Fairfax et al.	9
RPL7L1	?	ENSG00000146223	skeletal muscle	1.23×10-19 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	skeletal muscle	4.96×10-10 (p-value)	GTEx Portal V6	50
PEX6	?	ENSG00000124587	blood	5.06×10-6 (q-value)	SeeQTL DB (HapMap)	12
RPL7L1	?	ENSG00000146223	transverse colon	5.22×10-11 (p-value)	GTEx Portal V6	65
PEX6	?	ENSG00000124587	transverse colon	3.92×10-6 (p-value)	GTEx Portal V6	7
RPL7L1	?	ENSG00000146223	unexposed skin	5.02×10-10 (p-value)	GTEx Portal V6	55
PEX6	?	ENSG00000124587	unexposed skin	8.55×10-9 (p-value)	GTEx Portal V6	55
RPL7L1	?	ENSG00000146223	sun exposed skin	7.59×10-18 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	sun exposed skin	1.57×10-11 (p-value)	GTEx Portal V6	64
C6orf226	?	ENSG00000221821	sun exposed skin	8.24×10-6 (p-value)	GTEx Portal V6	5
RPL7L1	?	ENSG00000146223	aorta	3.25×10-12 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	aorta	8.93×10-7 (p-value)	GTEx Portal V6	5
RPL7L1	?	ENSG00000146223	left ventricle	3.73×10-12 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	left ventricle	5.40×10-7 (p-value)	GTEx Portal V6	6
RPL7L1	?	ENSG00000146223	subcutaneous adipocytes	6.97×10-16 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	subcutaneous adipocytes	4.18×10-10 (p-value)	GTEx Portal V6	65
RPL7L1	?	ENSG00000146223	tibial nerve	2.06×10-11 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	tibial nerve	5.09×10-12 (p-value)	GTEx Portal V6	65
C6orf226	?	ENSG00000221821	tibial nerve	2.55×10-6 (p-value)	GTEx Portal V6	6
RPL7L1	?	ENSG00000146223	esophagus mucosa	9.20×10-13 (p-value)	GTEx Portal V6	46
PEX6	?	ENSG00000124587	esophagus mucosa	1.28×10-9 (p-value)	GTEx Portal V6	64

RPL7L1	?	ENSG00000146223	gastroesophageal junction	3.82×10-8 (p-value)	GTEx Portal V6	58
RPL7L1	?	ENSG00000146223	testis	3.65×10-13 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	testis	1.26×10-8 (p-value)	GTEx Portal V6	64
PEX6	ENST00000304611	ILMN_1683279	skin	1.78×10-13 (p-value)	MuTHER consortium	22
PEX6	ENST00000244546		blood	6.63×10-18 (p-value)	MuTHER consortium	22
			adipocyte	2.33×10-16 (p-value)	MuTHER consortium	22
PEX6	ENST00000304611	ILMN_1683279	monocyte	2.55×10-60 (p-value)	Zeller et al.	6
PEX6	ENST00000244546					
?	?	ILMN_1736238	monocyte	2.91×10-21 (p-value)	Zeller et al.	6
RPL7L1	ENST00000493763	ILMN_1705908	monocyte	6.06×10-13 (p-value)	Zeller et al.	2
RPL7P21	ENST00000478287					
RPL7L1	ENST00000397415					
RPL7L1	ENST00000462348					
RPL7L1	?	ENSG00000146223	nucleus accumbens	4.29×10-7 (p-value)	GTEx Portal V6	28
PEX6	?	ENSG00000124587	atrial appendage	1.62×10-6 (p-value)	GTEx Portal V6	10
RPL7L1	?	ENSG00000146223	cerebellar hemisphere	7.46×10-11 (p-value)	GTEx Portal V6	60
RPL7L1	?	ENSG00000146223	pancreas	2.81×10-6 (p-value)	GTEx Portal V6	29
RPL7L1	?	ENSG00000146223	frontal cortex	1.58×10-6 (p-value)	GTEx Portal V6	4
RPL7L1	?	ENSG00000146223	visceral adipocytes	4.28×10-6 (p-value)	GTEx Portal V6	13
PEX6	?	ENSG00000124587	stomach	1.53×10-6 (p-value)	GTEx Portal V6	10
RPL7L1	?	ENSG00000146223	cerebellum	7.31×10-6 (p-value)	GTEx Portal V6	1
PEX6	?	ENSG00000124587	brain	2.62×10-5 (q-value)	SeeQTL DB (Myers et al.)	2
PEX6	ENST00000304611	204545_at	blood	6.70×10-8 (p-value)	Dixon et al.	1
PEX6	ENST00000244546					
PEX6	ENST00000304611	320_at	blood	2.60×10-8 (p-value)	Dixon et al.	1
PEX6	ENST00000244546					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
PEX6	?	ENSG00000124587	chr6	brain	3.58×10-3 (q-value)	SeeQTL DB (Myers et al.)	2

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
EGR1	MA0341.1	2	yes	0.000	1
EGR1	MA0366.1	2	yes	0.000	1
EGR1	MA0162.2	11	yes	0.000	1

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000746055	1	ENST00000304672, ENST00000441198	PTCRA

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000048629		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000449464	1	ENCP00000048621	PRPH2
		ENCP00000048658	CUL9
		ENCP00000048660	CUL9
ENCE00000449466	1	ENCP00000048621	PRPH2
		ENCP00000048658	CUL9
		ENCP00000048660	CUL9
ENCE00000449798	1	ENCP00000048674	RP11-480N24.4
ENCE00000449690	1	ENCP00000048649	PTK7
ENCE00000449547	1	ENCP00000048629	PTCRA
		ENCP00000048651	PTK7

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001216922 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K36me3, H3K27me3, PolII, Jund, TAF7, p300, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3
		HSMMtube	H3K36me3, DNase1, H2AZ, H3K4me3, H3K27ac, H3K9ac, H3K4me2
		blood (K562)	PolII, H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K36me3, PolII, Jund, Gabp
		blood (GM12878)	DNase1, Gabp, ELF1, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, H2AZ, PolII
		lung (IMR90)	H3K36me3, H3K4me3, H3K9ac, H3K4me2, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ, H3K36me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	Cmyc, H3K36me3, DNase1, Max, H3K4me3, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001216923 (promoter flanking region)	6	monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me3, H3K4me1, H3K4me2
		blood (GM12878)	H2AZ, PU1
		blood (K562)	DNase1, Max, PU1
ENSR00001703966 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF

		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF
ENSR00001216924 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF
ENSR00001495377 (enhancer)	1	Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF
		blood (K562)	DNase1, CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	Rad21, CTCF
		A549	CTCF
		skin (NHEK)	CTCF
ENSR00001216925 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, H2AZ, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K4me3, Rad21, H2AZ, H3K4me2, CTCF, DNase1
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	H2AZ, Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K4me2, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF, H3K4me2
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, DNase1, H3K27me3
ENSR00001703967 (promoter flanking region)	4	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	H2AZ, Rad21, CTCF, DNase1, PU1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K4me3, PolII, Rad21, H3K4me1, H2AZ, H3K4me2, CTCF, DNase1
		blood (GM12878)	H2AZ, Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K4me2, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF, DNase1

		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K27me3, DNase1, CTCF
ENSR00001216929 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K9ac, PolII, Rad21, Egr1, CTCF, Yy1, HDAC2, Tcf12, TAF1, H3K4me2, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	DNase1, H3K4me3, H3K4me2
		Osteobl	H3K4me2
		nervous (NH-A)	DNase1
ENSR00001216930 (open chromatin region)	1	lung (IMR90)	H3K27me3
		blood (GM12878)	DNase1, H2AZ
		blood (K562)	H3K27me3
ENSR00001216931 (enhancer)	1	liver (HepG2)	H3K4me1
ENSR00001216932 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		blood (K562)	DNase1, H3K36me3
		muscle (HSMM)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
RPL7L1	1	ENST00000304734 ENST00000397415 ENST00000459829 ENST00000483998 ENST00000493763	hsa-miR-1271-5p hsa-miR-96-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C6orf226	downstream gene variant, upstream gene variant	176	ENST00000408925	NM_001008739.1	ENSP00000386146	19
CNPY3	upstream gene variant	4443	ENST00000372836	NM_006586.3	ENSP00000361926	1
CNPY3	upstream gene variant	4435	ENST00000394142	?	ENSP00000377698	1
GLTSCR1L	downstream gene variant	2764	ENST00000314073	?	ENSP00000313933	4
GLTSCR1L	downstream gene variant	2934	ENST00000614467	?	ENSP00000482211	3
GLTSCR1L	downstream gene variant	2766	ENST00000394168	NM_015349.1	ENSP00000377723	4
PTCRA	upstream gene variant	58	ENST00000446507	NM_001243170.1	ENSP00000392288	4
PTCRA	upstream gene variant	58	ENST00000304672	NM_138296.2	ENSP00000304447	4
PTCRA	upstream gene variant	58	ENST00000441198	NM_001243169.1	ENSP00000409550	4
PTCRA	upstream gene variant	58	ENST00000616441	NM_001243168.1	ENSP00000477815	4
RPL7L1	downstream gene variant	1568	ENST00000487619	?	?	5
RPL7L1	downstream gene variant	173	ENST00000497417	?	?	6
RPL7L1	downstream gene variant	446	ENST00000492836	?	?	8
RPL7L1	downstream gene variant	164	ENST00000304734	?	ENSP00000346063	11
RPL7L1	downstream gene variant	614	ENST00000602561	?	ENSP00000473356	5
RPL7L1	downstream gene variant	1460	ENST00000483998	?	?	7
RPL7L1	downstream gene variant	1751	ENST00000397415	?	?	7
RPL7L1	downstream gene variant	193	ENST00000493763	NM_198486.2	ENSP00000418221	11
RPL7L1	upstream gene variant, downstream gene variant	130	ENST00000459829	?	?	15
RPL7L1	downstream gene variant	509	ENST00000462348	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GLTSCR1L	ENST00000314073	?	ENSP00000313933	5
GLTSCR1L	ENST00000614467	?	ENSP00000482211	5
GLTSCR1L	ENST00000394168	NM_015349.1	ENSP00000377723	5
PTCRA	ENST00000441198	NM_001243169.1	ENSP00000409550	7
PTCRA	ENST00000446507	NM_001243170.1	ENSP00000392288	8
PTCRA	ENST00000304672	NM_138296.2	ENSP00000304447	7
PTCRA	ENST00000616441	NM_001243168.1	ENSP00000477815	7
RPL7L1	ENST00000304734	?	ENSP00000346063	9
RPL7L1	ENST00000602561	?	ENSP00000473356	8
RPL7L1	ENST00000462348	?	?	8
RPL7L1	ENST00000459829	?	?	1
RPL7L1	ENST00000397415	?	?	9
RPL7L1	ENST00000483998	?	?	9
RPL7L1	ENST00000493763	NM_198486.2	ENSP00000418221	9
RPL7L1	ENST00000487619	?	?	6

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RPL7L1	ENST00000304734	?	ENSP00000346063	4
RPL7L1	ENST00000493763	NM_198486.2	ENSP00000418221	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RPL7L1	ENST00000497417	?	3
RPL7L1	ENST00000397415	?	6
RPL7L1	ENST00000487619	?	5

Locus 451 (rs10135681)

SNiPAcard

Block annotations

Block info

genomic range	chr14:95,007,744-95,007,744
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-1.604	**gene(s) hit or close-by**	–
phastCons	0	**eQTL gene(s)**	–
GERP++	0.617	**potentially regulated gene(s)**	–
CADD score	3.096	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000423034 (promoter flanking region)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	CTCF, DNase1
		Osteobl	CTCF
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF, H3K4me2, H3K4me1, Rad21, H3K4me3
		A549	H3K27me3