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