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