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