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