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