Locus 386 (rs9646944)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,858,921-102,875,540
block size	16,620 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.169 [-0.796 – 1.086]	gene(s) hit or close-by	FAM183DP, IL1RL2
phastCons	μ = 0.010 [0 – 0.027]	eQTL gene(s)	AC007278.3, IL18R1, IL1RL1
GERP++	μ = 0.493 [-0.539 – 1.48]	potentially regulated gene(s)	–
CADD score	μ = 3.754 [1.939 – 6.629]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL1RL1	?	ENSG00000115602	lung	2.92×10^{-6} (p-value)	GTEx Portal V6	5
AC007278.3	?	ENSG00000234389	lung	1.49×10^{-5} (p-value)	GTEx Portal V6	1
AC007278.3	?	ENSG00000234389	blood	1.87×10^{-8} (p-value)	GTEx Portal V6	5
IL18R1	?	ENSG00000115604	cerebellum	8.77×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001544945 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K27me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		liver (HepG2)	H3K27me3, DNase1
		lung (IMR90)	DNase1, H2BK20ac
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FAM183DP	downstream gene variant	442	ENST00000421987	?	?	2
IL1RL2	downstream gene variant	2459	ENST00000264257	NM_003854.2	ENSP00000264257	2
IL1RL2	downstream gene variant	3110	ENST00000441515	?	ENSP00000413348	2
IL1RL2	downstream gene variant	3123	ENST00000481806	?	?	2