Locus 300 (rs1922291)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,816,695-102,842,124
block size	25,430 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.948 [-1.696 – 0.167]	gene(s) hit or close-by	IL1RL2
phastCons	μ = 0.053 [0 – 0.303]	eQTL gene(s)	IL1RL2
GERP++	μ = -0.858 [-2.87 – 0.466]	potentially regulated gene(s)	–
CADD score	μ = 1.467 [0.89 – 2.161]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL1RL2	?	ENSG00000115598	sun exposed skin	3.32×10^{-8} (p-value)	GTEx Portal V6	7
IL1RL2	?	ENSG00000115598	unexposed skin	2.32×10^{-6} (p-value)	GTEx Portal V6	7
IL1RL2	?	ENSG00000115598	thyroid	4.87×10^{-7} (p-value)	GTEx Portal V6	7

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000595631 (CTCF binding site)	1	liver (HepG2)	H2AZ, H3K4me2, CTCF
		blood (DND-41)	H3K27me3
ENSR00001655312 (enhancer)	1	liver (HepG2)	CTCF, H3K4me2, H2AZ
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL1RL2	downstream gene variant	1007	ENST00000421464	?	ENSP00000387611	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RL2	ENST00000441515	?	ENSP00000413348	7
IL1RL2	ENST00000264257	NM_003854.2	ENSP00000264257	7
IL1RL2	ENST00000481806	?	?	7
IL1RL2	ENST00000421464	?	ENSP00000387611	1