Locus 309 (rs7097780)

SNiPAcard

Block annotations

Block info

genomic range	chr10:6,008,958-6,027,791
block size	18,834 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.162 [-1.666 – 0.88]	gene(s) hit or close-by	IL15RA
phastCons	μ = 0.048 [0 – 0.265]	eQTL gene(s)	IL15RA
GERP++	μ = -1.081 [-4.45 – 0.737]	potentially regulated gene(s)	GDI2, RP11-318E3.4
CADD score	μ = 3.814 [0.013 – 12.78]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL15RA	?	ENSG00000134470	transformed fibroblasts	9.34×10^{-8} (p-value)	GTEx Portal V6	15
IL15RA	?	ENSG00000134470	blood	4.89×10^{-9} (p-value)	GTEx Portal V6	14

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000065993	1	ENCP00000007580	GDI2
		ENCP00000007581	RP11-318E3.4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000350158 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	H3K27me3
		blood (K562)	DNase1
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	H3K27ac, H3K4me1, H3K36me3, DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	H3K27me3
		blood (GM12878)	H3K4me3, H3K79me2, H3K27ac, H3K4me2
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL15RA	upstream gene variant	2570	ENST00000397248	NM_001256765.1	ENSP00000380421	2
IL15RA	upstream gene variant	656	ENST00000532039	?	ENSP00000432691	4
IL15RA	upstream gene variant	3163	ENST00000447291	?	ENSP00000405128	2
IL15RA	upstream gene variant	2002	ENST00000379974	?	?	2
IL15RA	upstream gene variant	2967	ENST00000453922	?	ENSP00000405107	2
IL15RA	upstream gene variant	1994	ENST00000525219	NM_001243539.1	ENSP00000431529	2

IL15RA	upstream gene variant	2570	ENST00000620345	?	ENSP00000479839	2
IL15RA	upstream gene variant	2570	ENST00000622442	?	ENSP00000480949	2
IL15RA	upstream gene variant	2674	ENST00000379971	?	ENSP00000369306	2
IL15RA	upstream gene variant	2136	ENST00000534292	?	?	2
IL15RA	upstream gene variant	1994	ENST00000379972	?	?	2
IL15RA	upstream gene variant	2570	ENST00000397251	?	ENSP00000380423	2
IL15RA	upstream gene variant	2659	ENST00000528354	NM_172200.2	ENSP00000435454	2
IL15RA	upstream gene variant	2689	ENST00000397255	?	ENSP00000380426	2
IL15RA	upstream gene variant	2346	ENST00000397246	?	ENSP00000380420	2
IL15RA	upstream gene variant	2591	ENST00000379977	NM_002189.3	ENSP00000369312	2
IL15RA	upstream gene variant	2570	ENST00000618528	?	ENSP00000479938	2
IL15RA	upstream gene variant	2659	ENST00000429135	?	ENSP00000395113	2
IL15RA	upstream gene variant	2570	ENST00000620865	?	ENSP00000478525	2
IL15RA	upstream gene variant	2659	ENST00000397250	?	ENSP00000380422	2
IL15RA	upstream gene variant	2689	ENST00000530685	?	ENSP00000435995	2
IL15RA	upstream gene variant	2143	ENST00000532948	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL15RA	ENST00000622442	?	ENSP00000480949	12
IL15RA	ENST00000525219	NM_001243539.1	ENSP00000431529	12
IL15RA	ENST00000528354	NM_172200.2	ENSP00000435454	12
IL15RA	ENST00000534292	?	?	12
IL15RA	ENST00000379971	?	ENSP00000369306	12
IL15RA	ENST00000397248	NM_001256765.1	ENSP00000380421	12
IL15RA	ENST00000379972	?	?	12
IL15RA	ENST00000397250	?	ENSP00000380422	12
IL15RA	ENST00000620345	?	ENSP00000479839	12
IL15RA	ENST00000453922	?	ENSP00000405107	12
IL15RA	ENST00000618528	?	ENSP00000479938	12
IL15RA	ENST00000397255	?	ENSP00000380426	12
IL15RA	ENST00000397246	?	ENSP00000380420	12
IL15RA	ENST00000379977	NM_002189.3	ENSP00000369312	12
IL15RA	ENST00000530685	?	ENSP00000435995	12
IL15RA	ENST00000429135	?	ENSP00000395113	12
IL15RA	ENST00000397251	?	ENSP00000380423	12
IL15RA	ENST00000379974	?	?	12
IL15RA	ENST00000620865	?	ENSP00000478525	12
IL15RA	ENST00000532948	?	?	12

Locus 309 (sentinel SNP rs7097780 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.