Locus 231 (rs2193873)

SNiPAcard

Block annotations

Block info

genomic range	chr3:190,313,507-190,314,468
block size	962 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.041 [-0.125 – 0.044]	**gene(s) hit or close-by**	IL1RAP
phastCons	μ = 0.247 [0 – 0.495]	**eQTL gene(s)**	–
GERP++	μ = 1.044 [-0.811 – 2.9]	**potentially regulated gene(s)**	–
CADD score	μ = 7.328 [2.746 – 11.91]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001382725 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K4me1
		skin (NHEK)	H3K4me1
ENSR00001487393 (enhancer)	1	liver (HepG2)	H3K4me1
		skin (NHEK)	H3K4me1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RAP	ENST00000317757	NM_001167931.1	ENSP00000314807	2
IL1RAP	ENST00000434491	?	ENSP00000391899	2
IL1RAP	ENST00000342550	?	ENSP00000345829	2
IL1RAP	ENST00000453359	?	ENSP00000412008	2
IL1RAP	ENST00000072516	NM_001167929.1	ENSP00000072516	2
IL1RAP	ENST00000422940	NM_001167930.1	ENSP00000387371	2
IL1RAP	ENST00000439062	NM_001167928.1	ENSP00000401132	2
IL1RAP	ENST00000422485	NM_134470.3	ENSP00000409352	2
IL1RAP	ENST00000447382	NM_002182.3	ENSP00000390541	2
IL1RAP	ENST00000422625	?	ENSP00000389149	2
IL1RAP	ENST00000413869	?	ENSP00000416296	2
IL1RAP	ENST00000412504	?	ENSP00000412053	2
IL1RAP	ENST00000443369	?	ENSP00000408893	2