Locus 6 (rs1024948)

SNiPAcard

Block annotations

Block info

genomic range	chr3:190,344,128-190,354,410
block size	10,283 bp
variant count	24 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.442 [-4.686 – 1.233]	**gene(s) hit or close-by**	GCNT1P3, IL1RAP
phastCons	μ = 0.038 [0 – 0.528]	**eQTL gene(s)**	–
GERP++	μ = -0.108 [-6.06 – 2.74]	**potentially regulated gene(s)**	–
CADD score	μ = 3.837 [0.201 – 9.854]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Body height	3.44×10^{-5}	dbGaP	pha000341 dbGaP	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001487397 (CTCF binding site)	1	cervix (HeLa-S3)	H3K36me3
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GCNT1P3	downstream gene variant	1170	ENST00000450607	?	?	16
IL1RAP	downstream gene variant	224	ENST00000422485	NM_134470.3	ENSP00000409352	6
IL1RAP	downstream gene variant	157	ENST00000412080	?	ENSP00000414102	8
IL1RAP	downstream gene variant	224	ENST00000422940	NM_001167930.1	ENSP00000387371	6
IL1RAP	downstream gene variant	265	ENST00000434491	?	ENSP00000391899	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RAP	ENST00000317757	NM_001167931.1	ENSP00000314807	24
IL1RAP	ENST00000413869	?	ENSP00000416296	24
IL1RAP	ENST00000439062	NM_001167928.1	ENSP00000401132	24
IL1RAP	ENST00000447382	NM_002182.3	ENSP00000390541	24
IL1RAP	ENST00000434491	?	ENSP00000391899	17
IL1RAP	ENST00000412080	?	ENSP00000414102	14
IL1RAP	ENST00000422940	NM_001167930.1	ENSP00000387371	17

IL1RAP	ENST00000072516	NM_001167929.1	ENSP00000072516	24
IL1RAP	ENST00000422485	NM_134470.3	ENSP00000409352	17
IL1RAP	ENST00000342550	?	ENSP00000345829	24
IL1RAP	ENST00000412504	?	ENSP00000412053	24
IL1RAP	ENST00000443369	?	ENSP00000408893	24

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL1RAP	ENST00000434491	?	ENSP00000391899	4
IL1RAP	ENST00000412080	?	ENSP00000414102	1
IL1RAP	ENST00000422940	NM_001167930.1	ENSP00000387371	4
IL1RAP	ENST00000422485	NM_134470.3	ENSP00000409352	4