Locus 55 (rs376008)

SNiPAcard

Block annotations

Block info

genomic range	chr19:18,178,616-18,189,568
block size	10,953 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.087 [-4.269 – 0.224]	gene(s) hit or close-by	IL12RB1
phastCons	μ = 0.009 [0 – 0.064]	eQTL gene(s)	IL12RB1
GERP++	μ = -1.032 [-7.22 – 1.68]	potentially regulated gene(s)	–
CADD score	μ = 2.669 [0.001 – 10.62]	disease gene(s)	IL12RB1

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM984025	1
?	HGMD curated	HGMD	CM030471	1
?	HGMD curated	HGMD	CM030470	1

Disease gene annotation

gene	trait	source DB	source entry/link	
IL12RB1	IMMUNODEFICIENCY 30	OMIM	MIM:614891	OMIM
IL12RB1	Primary biliary cirrhosis	OrphaNet	OrphaNet:186	orphanet
IL12RB1	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency	OrphaNet	OrphaNet:319552	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IL12RB1	missense variant	ENST00000593993	NM_005535.1, NM_001290023.1	ENSP00000472165	3	3			3
IL12RB1	missense variant	ENST00000600835	NM_001290024.1	ENSP00000470788	3	3			3
IL12RB1	missense variant	ENST00000322153	NM_153701.1	ENSP00000314425	R/Q	cGg/cAg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL12RB1	?	ENSG00000096996	lung	3.11×10^{-9} (p-value)	GTEx Portal V6	21
IL12RB1	?	ENSG00000096996	blood	3.23×10^{-10} (p-value)	GTEx Portal V6	21
IL12RB1	?	ENSG00000096996	transverse colon	2.38×10^{-6} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000344606 (CTCF binding site)	2	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF
		HSMMtube	H3K27me3
		blood (K562)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	CTCF
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL12RB1	downstream gene variant	3476	ENST00000594176	?	ENSP00000473051	1
IL12RB1	downstream gene variant	2140	ENST00000430026	?	ENSP00000403103	5
IL12RB1	downstream gene variant	94	ENST00000322153	NM_153701.1	ENSP00000314425	7
IL12RB1	downstream gene variant	4354	ENST00000597416	?	?	1
IL12RB1	downstream gene variant	3429	ENST00000598019	?	ENSP00000468831	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
IL12RB1	ENST00000593993	NM_005535.1, NM_001290023.1	ENSP00000472165	P	ccT/ccC	1
IL12RB1	ENST00000600835	NM_001290024.1	ENSP00000470788	P	ccT/ccC	1
IL12RB1	ENST00000322153	NM_153701.1	ENSP00000314425	P	ccT/ccC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL12RB1	ENST00000593993	NM_005535.1, NM_001290023.1	ENSP00000472165	17
IL12RB1	ENST00000600835	NM_001290024.1	ENSP00000470788	17
IL12RB1	ENST00000322153	NM_153701.1	ENSP00000314425	12