Locus 10 (rs17413015)

SNiPAcard

Block annotations

Block info

genomic range	chr1:161,640,787-161,646,824
block size	6,038 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.191 [-0.658 – 1.684]	gene(s) hit or close-by	FCGR2B, RP11-25K21.1
phastCons	μ = 0.124 [0 – 0.986]	eQTL gene(s)	FCGR2B, FCGR2C, FCGR3B, FCRLB
GERP++	μ = -0.088 [-2.64 – 2.31]	potentially regulated gene(s)	–
CADD score	μ = 5.474 [0.031 – 14.51]	disease gene(s)	FCGR2B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
FCGR2B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
FCGR2B	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCGR3B	?	ENSG00000162747	sun exposed skin	2.06×10^{-6} (p-value)	GTEx Portal V6	4
FCGR3B	?	ENSG00000162747	unexposed skin	4.97×10^{-7} (p-value)	GTEx Portal V6	4
FCGR2C	?	ENSG00000244682	blood	1.06×10^{-6} (p-value)	GTEx Portal V6	4
FCGR2B	ENST00000403078	ILMN_1660027	blood	5.47×10^{-7} (p-value)	Westra et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000403078	ILMN_2382403	monocyte	1.04×10^{-4} (p-value)	Fairfax et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCRLB	ENST00000367948	ILMN_1782015	monocyte	5.79×10^{-4} (p-value)	Fairfax et al.	1
FCRLB	ENST00000495397		b-cell	9.59×10^{-4} (p-value)	Fairfax et al.	1
FCRLB	ENST00000367944					
FCRLB	ENST00000336830					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001525304 (enhancer)	1	embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H2AZ
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR2B	downstream gene variant	78	ENST00000485778	?	?	5
RP11-25K21.1	downstream gene variant, upstream gene variant	109	ENST00000626340	?	?	6
RP11-25K21.1	downstream gene variant, upstream gene variant	49	ENST00000453111	?	?	6

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
FCGR2B	ENST00000367961	NM_001190828.1	ENSP00000356938	T	acA/acG	1
FCGR2B	ENST00000367960	?	ENSP00000356937	T	acA/acG	1
FCGR2B	ENST00000236937	NM_001002274.2, NM_001002273.2	ENSP00000236937	T	acA/acG	1
FCGR2B	ENST00000403078	?	ENSP00000386038	T	acA/acG	1
FCGR2B	ENST00000428605	?	ENSP00000404329	T	acA/acG	1
FCGR2B	ENST00000358671	NM_004001.4, NM_001002275.2	ENSP00000351497	T	acA/acG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2B	ENST00000367961	NM_001190828.1	ENSP00000356938	7
FCGR2B	ENST00000367960	?	ENSP00000356937	7
FCGR2B	ENST00000480308	?	?	7
FCGR2B	ENST00000236937	NM_001002274.2, NM_001002273.2	ENSP00000236937	7
FCGR2B	ENST00000403078	?	ENSP00000386038	7
FCGR2B	ENST00000428605	?	ENSP00000404329	7
FCGR2B	ENST00000358671	NM_004001.4, NM_001002275.2	ENSP00000351497	7
FCGR2B	ENST00000485778	?	?	2
RP11-25K21.1	ENST00000626340	?	?	2
RP11-25K21.1	ENST00000453111	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FCGR2B	ENST00000480308	?	7
FCGR2B	ENST00000485778	?	2
RP11-25K21.1	ENST00000626340	?	2

Locus 10 (sentinel SNP rs17413015 + 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.