Locus 240 (rs6668534)

SNiPAcard

Block annotations

Block info

genomic range	chr1:161,644,976-161,671,995
block size	27,020 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.717 [-1.689 – 0.386]	gene(s) hit or close-by	FCGR2B, FCRLA, RP11-25K21.1, RPL31P11, Y_RNA
phastCons	μ = 0.035 [0 – 0.075]	eQTL gene(s)	FCGR2B, FCGR2C, FCGR3B, FCRLA, FCRLB, HSPA6, HSPA7
GERP++	μ = -0.263 [-2.5 – 0.705]	potentially regulated gene(s)	–
CADD score	μ = 2.677 [0.282 – 5.888]	disease gene(s)	FCGR2B

Trait annotations

Disease gene annotation

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

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCGR3B	?	ENSG00000162747	sun exposed skin	3.64×10^{-9} (p-value)	GTEx Portal V6	7
FCGR3B	?	ENSG00000162747	unexposed skin	5.28×10^{-9} (p-value)	GTEx Portal V6	7
FCGR2C	?	ENSG00000244682	tibial nerve	1.38×10^{-6} (p-value)	GTEx Portal V6	1
FCRLA	ENST00000367957	ILMN_1691071	skin	2.19×10^{-8} (p-value)	MuTHER consortium	2
FCRLA	ENST00000236938		blood	1.93×10^{-14} (p-value)	MuTHER consortium	2
FCRLA	ENST00000350710					
FCRLA	ENST00000470841					
FCRLA	ENST00000367949					
FCRLA	ENST00000367959					
FCRLA	ENST00000546024					
FCRLA	ENST00000540521					
FCRLA	ENST00000367953					
?	?	ILMN_1728639	blood	8.15×10^{-5} (p-value)	Westra et al.	2
FCGR2B	ENST00000403078	ILMN_1660027	blood	5.57×10^{-7} (p-value)	Westra et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000403078	ILMN_2382403	monocyte	1.44×10^{-7} (p-value)	Fairfax et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					

FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000480308	ILMN_1660027	monocyte	6.50×10-10 (p-value)	Fairfax et al.	1
FCGR2B	ENST00000403078					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCRLB	ENST00000367948	ILMN_1782015	monocyte	8.39×10-11 (p-value)	Fairfax et al.	1
FCRLB	ENST00000495397					
FCRLB	ENST00000367944					
FCRLB	ENST00000336830					
HSPA7	ENST00000445535	ILMN_3235964	monocyte	8.70×10-8 (p-value)	Fairfax et al.	1
			b-cell	2.28×10-6 (p-value)	Fairfax et al.	1
FCGR2B	ENST00000480308	ILMN_2382403	monocyte	4.50×10-18 (p-value)	Zeller et al.	1
FCGR2B	ENST00000403078					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000403078	ILMN_1660027	monocyte	1.52×10-39 (p-value)	Zeller et al.	1
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCRLB	ENST00000367948	ILMN_1782015	monocyte	2.05×10-24 (p-value)	Zeller et al.	1
FCRLB	ENST00000367944					
FCRLB	ENST00000495397					
FCRLB	ENST00000336830					
HSPA7	ENST00000445535	ILMN_1806165	monocyte	1.02×10-14 (p-value)	Zeller et al.	1
HSPA6	ENST00000309758					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000546488 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		A549	H3K4me3, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR2B	downstream gene variant	1070	ENST00000485778	?	?	1
FCRLA	upstream gene variant	4767	ENST00000540521	NM_001184870.1	ENSP00000442870	1
FCRLA	upstream gene variant	4767	ENST00000367959	NM_001184866.1	ENSP00000356936	1
FCRLA	upstream gene variant	4767	ENST00000546024	NM_001184867.1	ENSP00000439838	1
FCRLA	upstream gene variant	4767	ENST00000236938	NM_032738.3	ENSP00000236938	1
FCRLA	upstream gene variant	4767	ENST00000350710	NM_001184871.1	ENSP00000344808	1
FCRLA	upstream gene variant	4767	ENST00000367949	NM_001184873.1	ENSP00000356926	1

RP11-25K21.1	upstream gene variant	362	ENST00000453111	?	?	1
RP11-25K21.1	upstream gene variant	422	ENST00000626340	?	?	1
RPL31P11	upstream gene variant	4803	ENST00000426558	?	?	1
RPL31P11	upstream gene variant	4846	ENST00000454082	?	?	1
Y_RNA	upstream gene variant, downstream gene variant	2598	ENST00000362725	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2B	ENST00000403078	?	ENSP00000386038	1
FCGR2B	ENST00000358671	NM_004001.4, NM_001002275.2	ENSP00000351497	1
FCGR2B	ENST00000367960	?	ENSP00000356937	1
FCGR2B	ENST00000428605	?	ENSP00000404329	1
FCGR2B	ENST00000236937	NM_001002274.2, NM_001002273.2	ENSP00000236937	1
FCGR2B	ENST00000367961	NM_001190828.1	ENSP00000356938	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FCGR2B	ENST00000480308	?	1