SNiPAcard

Block annotations

Block info

genomic range	chr1:161,500,130-161,508,763
block size	8,634 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -3.023 [-9.199 – -0.106]	**gene(s) hit or close-by**	FCGR3A, HSPA6, RP11-25K21.6, RPS23P10
phastCons	μ = 0.115 [0 – 0.46]	**eQTL gene(s)**	FCGR2B, FCGR2C, FCRLB, HSPA7, RPS23P10, RPS23P9
GERP++	μ = -1.192 [-4.77 – 0.602]	**potentially regulated gene(s)**	HSPA6, TOMM40L, UFC1, USF1
CADD score	μ = 3.328 [0.487 – 6.425]	**disease gene(s)**	USF1, FCGR2B, FCGR3A

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
USF1	HYPERLIPIDEMIA, FAMILIAL COMBINED	OMIM	MIM:144250
USF1	HYPERLIPIDEMIA, COMBINED, 1	OMIM	MIM:602491
FCGR2B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
FCGR2B	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
FCGR3A	IMMUNODEFICIENCY 20	OMIM	MIM:615707

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCGR2C	?	ENSG00000244682	muscularis mucosae	9.34×10^{-7} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	muscularis mucosae	7.78×10^{-9} (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	lung	6.16×10^{-7} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	lung	3.33×10^{-8} (p-value)	GTEx Portal V6	3
RPS23P10	?	ENSG00000224203	blood	2.65×10^{-14} (p-value)	GTEx Portal V6	3
RPS23P9	?	ENSG00000231586	blood	3.15×10^{-7} (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	breast	4.93×10^{-8} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	breast	1.60×10^{-8} (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	tibial artery	2.39×10^{-6} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	tibial artery	8.99×10^{-13} (p-value)	GTEx Portal V6	3
HSPA7	ENST00000445535	ILMN_3235964	monocyte	1.02×10^{-6} (p-value)	Fairfax et al.	1
			b-cell	9.19×10^{-12} (p-value)	Fairfax et al.	1
FCGR2C	?	ENSG00000244682	thyroid	1.90×10^{-6} (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	thyroid	5.16×10^{-9} (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	skeletal muscle	5.52×10^{-9} (p-value)	GTEx Portal V6	3

HSPA7	?	ENSG00000225217	skeletal muscle	8.17×10-13 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	transverse colon	5.46×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	sun exposed skin	1.08×10-12 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	sun exposed skin	8.65×10-16 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	left ventricle	9.76×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	aorta	3.59×10-6 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	aorta	3.45×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	subcutaneous adipocytes	2.16×10-10 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	subcutaneous adipocytes	1.23×10-13 (p-value)	GTEx Portal V6	3
?	?	ILMN_1666932	liver	1.21×10-10 (p-value)	Innocenti et al.	1
HSPA7	?	ENSG00000225217	visceral adipocytes	2.58×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	coronary artery	2.86×10-6 (p-value)	GTEx Portal V6	1
HSPA7	?	ENSG00000225217	coronary artery	5.03×10-8 (p-value)	GTEx Portal V6	3
HSPA7	?	ENSG00000225217	esophagus mucosa	9.65×10-7 (p-value)	GTEx Portal V6	3
FCGR2B	ENST00000480308	ILMN_2382403	blood	2.26×10-5 (p-value)	Westra et al.	3
FCGR2B	ENST00000403078					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCGR2B	ENST00000403078	ILMN_1660027	blood	5.53×10-6 (p-value)	Westra et al.	3
FCGR2B	ENST00000480308					
FCGR2B	ENST00000367960					
FCGR2B	ENST00000236937					
FCGR2B	ENST00000358671					
FCGR2B	ENST00000367961					
FCRLB	ENST00000367948	ILMN_1782015	blood	1.52×10-6 (p-value)	Westra et al.	3
FCRLB	ENST00000367944					
FCRLB	ENST00000495397					
FCRLB	ENST00000336830					
HSPA7	?	ENSG00000225217	testis	4.32×10-7 (p-value)	GTEx Portal V6	3
FCGR2C	?	ENSG00000244682	tibial nerve	8.54×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000041151	1	ENCP00000005224	HSPA6
		ENCP00000005167	USF1
		ENCP00000005179	UFC1
		ENCP00000005164	USF1
		ENCP00000005205	TOMM40L
		ENCP00000005166	USF1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000546473 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, CTCF, PolII, Rad21, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	DNase1, H3K9ac, H3K4me2, H3K27me3
		blood (K562)	H3K27ac, Max, H3K9ac, ETS1, Cmyc, HEY1, H2AZ, TAF1, PolII, H3K4me2, DNase1, H3K4me3, Egr1
		skin (NHDF-AD)	DNase1, H3K4me3
		muscle (HSMM)	H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H2AZ, H3K4me2, H3K4me3, H3K27me3, DNase1
		blood (GM12878)	PolII, H2AZ, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K27ac, H3K9ac, H3K4me3, H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, PolII, H3K4me3, Gabp
		monocytes (Monocytes-CD14+)	H3K27ac, H3K27me3, H3K4me3, H3K4me2, H3K4me1, DNase1
		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, PolII, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR3A	downstream gene variant	2792	ENST00000436743	NM_001127595.1, NM_001127593.1, NM_001127596.1	ENSP00000416607	3
FCGR3A	downstream gene variant	2882	ENST00000426740	?	ENSP00000410180	3
FCGR3A	downstream gene variant	2786	ENST00000367969	NM_000569.6, NM_001127592.1	ENSP00000356946	3
FCGR3A	downstream gene variant	2823	ENST00000367967	?	ENSP00000356944	3
FCGR3A	downstream gene variant	2790	ENST00000443193	?	ENSP00000392047	3
HSPA6	downstream gene variant	3430	ENST00000309758	NM_002155.3	ENSP00000310219	1
RPS23P10	upstream gene variant	669	ENST00000416185	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-25K21.6	ENST00000537821	?	?	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RPS23P10	ENST00000416185	?	1