SNiPAcard

Block annotations

Block info

genomic range	chr1:161,490,524-161,519,440
block size	28,917 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.281 [-2.127 – 0.551]	**gene(s) hit or close-by**	FCGR2A, FCGR3A, HSPA6, RP11-25K21.6, RPS23P10
phastCons	μ = 0.001 [0 – 0.003]	**eQTL gene(s)**	–
GERP++	μ = 0.479 [-0.451 – 1.41]	**potentially regulated gene(s)**	–
CADD score	μ = 2.633 [0.417 – 4.352]	**disease gene(s)**	FCGR3A

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
FCGR3A	IMMUNODEFICIENCY 20	OMIM	MIM:615707

Putative effect on regulation

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
ENSR00001587253 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR2A	downstream gene variant	2144	ENST00000461298	?	?	1
FCGR2A	downstream gene variant	1166	ENST00000271450	NM_001136219.1, NM_201563.5	ENSP00000271450	1
FCGR2A	downstream gene variant	2131	ENST00000486608	?	?	1
FCGR2A	downstream gene variant	2176	ENST00000367972	NM_021642.3	ENSP00000356949	1

FCGR2A	downstream gene variant	2169	ENST00000483665	?	ENSP00000440148 1
FCGR2A	downstream gene variant	2379	ENST00000471026	?	? 1
FCGR2A	downstream gene variant	767	ENST00000459885	?	ENSP00000476411 1
FCGR3A	downstream gene variant	3187	ENST00000426740	?	ENSP00000410180 1
FCGR3A	downstream gene variant	3095	ENST00000443193	?	ENSP00000392047 1
FCGR3A	downstream gene variant	3097	ENST00000436743	NM_001127595.1, NM_001127593.1, NM_001127596.1	ENSP00000416607 1
FCGR3A	downstream gene variant	3091	ENST00000367969	NM_000569.6, NM_001127592.1	ENSP00000356946 1
FCGR3A	downstream gene variant	3128	ENST00000367967	?	ENSP00000356944 1
HSPA6	upstream gene variant, downstream gene variant	3806	ENST00000309758	NM_002155.3	ENSP00000310219 2
RPS23P10	downstream gene variant, upstream gene variant	510	ENST00000416185	?	? 2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2A	ENST00000467525	?	ENSP00000476495	1
FCGR2A	ENST00000491841	?	?	1
FCGR3A	ENST00000442336	?	ENSP00000396567	1
FCGR3A	ENST00000367969	NM_000569.6, NM_001127592.1	ENSP00000356946	1
FCGR3A	ENST00000476031	?	?	1
FCGR3A	ENST00000426740	?	ENSP00000410180	1
FCGR3A	ENST00000443193	?	ENSP00000392047	1
FCGR3A	ENST00000436743	NM_001127595.1, NM_001127593.1, NM_001127596.1	ENSP00000416607	1
FCGR3A	ENST00000367967	?	ENSP00000356944	1
RP11-25K21.6	ENST00000537821	?	?	4