Locus 407 (rs17200698)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,469,468-31,565,401
block size	95,934 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.126 [-0.147 – 0.22]	gene(s) hit or close-by	ATP6V1G2, ATP6V1G2-DDX39B, LST1, MCCD1, MICB, NCR3, NFKBIL1, PPIAP9, RPL15P4, XXbac-BPG16N22.5, Y_RNA
phastCons	μ = 0.077 [0 – 0.209]	eQTL gene(s)	XXbac-BPG248L24.12
GERP++	μ = -0.169 [-2.17 – 0.479]	potentially regulated gene(s)	–
CADD score	μ = 4.796 [2.556 – 11.31]	disease gene(s)	NFKBIL1, NCR3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
NFKBIL1	RHEUMATOID ARTHRITIS	OMIM	MIM:180300
NCR3	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	1.41×10^{-6} (p-value)	GTEx Portal V6	6
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	6.98×10^{-7} (p-value)	GTEx Portal V6	6
XXbac-BPG248L24.12	?	ENSG00000271581	blood	2.06×10^{-7} (p-value)	GTEx Portal V6	6

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487851 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	H3K27me3, Rad21
		liver (HepG2)	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00000487872 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		Osteobl	CTCF
		blood (K562)	DNase1, Max, CTCF
		blood (DND-41)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF

endothelium (HUVEC)	H3K36me3, CTCF, Cmyc
liver (HepG2)	CTCF
blood (GM12878)	DNase1, Rad21, CTCF
lung (IMR90)	CTCF
skin (NHEK)	CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ATP6V1G2	upstream gene variant	1714	ENST00000415099	?	ENSP00000390148	1
ATP6V1G2	upstream gene variant	3297	ENST00000303892	NM_130463.3	ENSP00000302194	1
ATP6V1G2	upstream gene variant	3504	ENST00000376151	NM_001204078.1	ENSP00000365321	1
ATP6V1G2	upstream gene variant	3293	ENST00000483251	NM_138282.2	ENSP00000419698	1
ATP6V1G2	upstream gene variant	3548	ENST00000481998	?	?	1
ATP6V1G2	upstream gene variant	3386	ENST00000483170	?	?	1
ATP6V1G2-DDX39B	upstream gene variant	3559	ENST00000376185	?	ENSP00000365356	1
ATP6V1G2-DDX39B	upstream gene variant	3533	ENST00000480131	?	ENSP00000420191	1
ATP6V1G2-DDX39B	upstream gene variant	3548	ENST00000475917	?	?	1
LST1	downstream gene variant	1641	ENST00000376110	?	ENSP00000365278	1
LST1	downstream gene variant	1489	ENST00000376090	?	ENSP00000365258	1
LST1	downstream gene variant	1489	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	downstream gene variant	1641	ENST00000460834	?	?	1
LST1	downstream gene variant	1489	ENST00000376089	?	ENSP00000365257	1
LST1	downstream gene variant	1588	ENST00000211921	?	ENSP00000211921	1
LST1	downstream gene variant	1489	ENST00000303757	NM_205840.2	ENSP00000303649	1
LST1	downstream gene variant	1722	ENST00000490742	?	ENSP00000418578	1
LST1	downstream gene variant	2668	ENST00000433492	?	ENSP00000399869	1
LST1	downstream gene variant	1772	ENST00000396112	?	ENSP00000379418	1
LST1	downstream gene variant	1489	ENST00000396101	?	ENSP00000379408	1
LST1	downstream gene variant	1489	ENST00000376096	?	ENSP00000365264	1
LST1	downstream gene variant	2557	ENST00000419073	?	?	1
LST1	downstream gene variant	1489	ENST00000418507	NM_205838.2	ENSP00000405900	1
LST1	downstream gene variant	1490	ENST00000376086	NM_001166538.1	ENSP00000365254	1
LST1	downstream gene variant	1489	ENST00000376099	?	ENSP00000365267	1
LST1	downstream gene variant	1641	ENST00000464044	?	ENSP00000417645	1
LST1	downstream gene variant	1489	ENST00000464526	?	?	1
LST1	downstream gene variant	1489	ENST00000376093	NM_007161.3	ENSP00000365261	1
LST1	downstream gene variant	1588	ENST00000339530	NM_205837.2	ENSP00000339201	1
LST1	downstream gene variant	1736	ENST00000376092	?	ENSP00000365260	1
MCCD1	upstream gene variant	4235	ENST00000376191	NM_001011700.2	ENSP00000365362	1
MICB	downstream gene variant	4800	ENST00000538442	NM_001289160.1	ENSP00000442345	1

MICB	upstream gene variant	4244	ENST00000494577	?	?	1
MICB	downstream gene variant	4799	ENST00000252229	NM_005931.4	ENSP00000252229	1
MICB	downstream gene variant	4799	ENST00000399150	NM_001289161.1	ENSP00000382103	1
NCR3	upstream gene variant	4639	ENST00000340027	NM_147130.2	ENSP00000342156	1
NCR3	upstream gene variant	4639	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	upstream gene variant	4665	ENST00000491161	?	?	1
NCR3	upstream gene variant	4639	ENST00000376072	NM_001145467.1	ENSP00000365240	1
NCR3	upstream gene variant	4875	ENST00000376071	?	ENSP00000365239	1
NCR3	upstream gene variant	4812	ENST00000495600	?	?	1
PPIAP9	downstream gene variant, upstream gene variant	3557	ENST00000403866	?	?	2
RPL15P4	upstream gene variant	3632	ENST00000416625	?	?	1
XXbac-BPG16N22.5	upstream gene variant	56	ENST00000538358	?	?	1
Y_RNA	upstream gene variant	4901	ENST00000383850	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MICB	ENST00000252229	NM_005931.4	ENSP00000252229	1
MICB	ENST00000538442	NM_001289160.1	ENSP00000442345	1
MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	1
NCR3	ENST00000376071	?	ENSP00000365239	1
NCR3	ENST00000495600	?	?	1
NCR3	ENST00000491161	?	?	1
NCR3	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	ENST00000376072	NM_001145467.1	ENSP00000365240	1
NCR3	ENST00000340027	NM_147130.2	ENSP00000342156	1
NFKBIL1	ENST00000473655	?	?	1
NFKBIL1	ENST00000376148	NM_005007.3	ENSP00000365318	1
NFKBIL1	ENST00000496233	?	ENSP00000437148	1
NFKBIL1	ENST00000376145	NM_001144961.1	ENSP00000365315	1
NFKBIL1	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	1