Locus 198 (rs2248372)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,446,466-31,452,103
block size	5,638 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.604 [-1.421 – -0.238]	gene(s) hit or close-by	HCP5
phastCons	μ = 0.004 [0.001 – 0.006]	eQTL gene(s)	ATP6V1G2, HCG27, HLA-B, HLA-C, LST1, LY6G5C, MICA, MICB, PSORS1C1, PSORS1C2, XXbac-BPG181B23.7, XXbac-BPG248L24.12
GERP++	μ = -0.018 [-0.737 – 0.458]	potentially regulated gene(s)	CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, LY6G6D, LY6G6D, LY6G6D, LY6G6D, LY6G6D, LY6G6D, LY6G6E, LY6G6E, LY6G6E, LY6G6E, LY6G6E, LY6G6E, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P
CADD score	μ = 2.082 [0.871 – 4.161]	disease gene(s)	HLA-B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
gamma-glutamylglutamine	2.92×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PSORS1C1	ENST00000481450	ILMN_1799669	skin	2.59×10^{-5} (p-value)	MuTHER consortium	1
PSORS1C1	ENST00000552747					
PSORS1C1	ENST00000547221					
PSORS1C1	ENST00000259881					
PSORS1C1	ENST00000479581					
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	7.59×10^{-6} (p-value)	MuTHER consortium	2
ATP6V1G2	ENST00000303892		skin	2.90×10^{-5} (p-value)	MuTHER consortium	2
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					
MICB	ENST00000538442	ILMN_1708006	skin	1.42×10^{-9} (p-value)	MuTHER consortium	2
MICB	ENST00000399150					
MICB	ENST00000252229					
HCG27	ENST00000383331	ILMN_1746436	adipocyte	3.10×10^{-5} (p-value)	MuTHER consortium	1
HCG27	ENST00000415276					

XXbac-BPG248L24.12	?	ENSG00000271581	lung	7.99×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	lung	2.96×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	lung	2.36×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	1.29×10-5 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	transformed fibroblasts	1.06×10-8 (p-value)	GTEx Portal V6	3
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	7.57×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	blood	4.00×10-7 (p-value)	GTEx Portal V6	4
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	1.30×10-6 (p-value)	GTEx Portal V6	4
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	1.17×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	1.19×10-8 (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	blood	1.47×10-2 (q-value)	SeeQTL DB (HapMap)	1
MICA	?	ENSG00000204520	sun exposed skin	1.48×10-9 (p-value)	GTEx Portal V6	4
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	1.96×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	3.64×10-9 (p-value)	GTEx Portal V6	4
PSORS1C1	?	ENSG00000204540	sun exposed skin	4.04×10-7 (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	sun exposed skin	6.17×10-6 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	2.30×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	left ventricle	1.79×10-6 (p-value)	GTEx Portal V6	3
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	1.88×10-6 (p-value)	GTEx Portal V6	4
PSORS1C1	?	ENSG00000204540	subcutaneous adipocytes	2.65×10-6 (p-value)	GTEx Portal V6	3
HLA-C	ENST00000470363	ILMN_2150787	monocyte	1.54×10-16 (p-value)	Zeller et al.	1
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	ENST00000466892					
PSORS1C1	?	ENSG00000204540	tibial nerve	5.92×10-9 (p-value)	GTEx Portal V6	4
PSORS1C2	?	ENSG00000204538	esophagus mucosa	7.70×10-8 (p-value)	GTEx Portal V6	4
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	2.02×10-9 (p-value)	GTEx Portal V6	4
PSORS1C1	?	ENSG00000204540	esophagus mucosa	9.36×10-9 (p-value)	GTEx Portal V6	4
LY6G5C	ENST00000383237	ILMN_1763467	blood	1.29×10-4 (p-value)	Westra et al.	1
LY6G5C	ENST00000474395					
LY6G5C	ENST00000375863					
LY6G5C	ENST00000474678					
LY6G5C	ENST00000467098					
LY6G5C	ENST00000460141					
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	1.05×10-4 (p-value)	Westra et al.	1
ATP6V1G2	ENST00000303892					
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					

LST1	ENST00000419073	ILMN_1717127	blood	7.89×10-5 (p-value)	Westra et al.	1
LST1	ENST00000433492					
LST1	ENST00000376089					
LST1	ENST00000376090					
LST1	ENST00000211921					
XXbac-BPG181B23.7	?	ENSG00000272221	testis	8.41×10-8 (p-value)	GTEx Portal V6	4
HLA-B	?	ENSG00000234745	cerebellum	1.63×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	1.16×10-6 (p-value)	GTEx Portal V6	3
PSORS1C1	?	ENSG00000204540	unexposed skin	1.03×10-6 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	breast	2.78×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444780	1	ENCP00000048082	LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D LY6G6D LY6G6E LY6G6D LY6G6D LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D
		ENCP00000047982	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
		ENCP00000048120	SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4 SLC44A4
		ENCP00000048000	WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P
		ENCP00000047981	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

HCP5	downstream gene variant	1183	ENST00000467369	?	?	2