Locus 295 (rs2284178)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,430,799-31,432,125
block size	1,327 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.123 [-2.407 – 0.16]	gene(s) hit or close-by	HCP5, XXbac-BPG181B23.6
phastCons	μ = 0.215 [0 – 0.431]	eQTL gene(s)	AIF1, CCHCR1, CYP21A1P, HLA-C, LST1, MICB, PSORS1C1, PSORS1C2, VARS2, XXbac-BPG248L24.12, XXbac-BPG299F13.17
GERP++	μ = -3.415 [-6.83 – 0]	potentially regulated gene(s)	HCP5, HCP5, HCP5, HCP5
CADD score	μ = 5.348 [0.416 – 10.28]	disease gene(s)	VARS2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Behcet syndrome	3.06×10^{-10}	dbGaP	pha002888	1

Disease gene annotation

gene	trait	source DB	source entry/link
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LST1	ENST00000339530	ILMN_1718936	blood	2.73×10^{-5} (p-value)	MuTHER consortium	2
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
MICB	ENST00000538442	ILMN_1708006	skin	5.07×10^{-10} (p-value)	MuTHER consortium	2
MICB	ENST00000399150					
MICB	ENST00000252229					
HLA-C	?	ENSG00000204525	atrial appendage	2.87×10^{-11} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	transformed fibroblasts	4.29×10^{-15} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	blood	3.23×10^{-19} (p-value)	GTEx Portal V6	2

XXbac-BPG248L24.12	?	ENSG00000271581	blood	4.48×10-9 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	blood	7.80×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	breast	1.02×10-15 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	breast	2.40×10-7 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	breast	1.67×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	skeletal muscle	1.73×10-24 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	2.17×10-11 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	skeletal muscle	9.85×10-9 (p-value)	GTEx Portal V6	2
CCHCR1	?	ENSG00000204536	blood	2.01×10-2 (q-value)	SeeQTL DB (HapMap)	2
HLA-C	?	ENSG00000204525	EBV lymphocytes	6.11×10-10 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	ovary	3.64×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	sun exposed skin	1.82×10-19 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	3.21×10-8 (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	sun exposed skin	7.82×10-6 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	sun exposed skin	2.70×10-8 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	unexposed skin	4.53×10-12 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	aorta	1.51×10-11 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	aorta	8.40×10-9 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	left ventricle	4.44×10-8 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	2.32×10-16 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	3.05×10-8 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	subcutaneous adipocytes	8.87×10-8 (p-value)	GTEx Portal V6	2
VARS2	?	ENSG00000137411	subcutaneous adipocytes	2.89×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	visceral adipocytes	2.02×10-11 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	1.53×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	stomach	1.64×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	cortex	1.79×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	tibial nerve	3.17×10-14 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	tibial nerve	1.46×10-10 (p-value)	GTEx Portal V6	2
PSORS1C2	?	ENSG00000204538	esophagus mucosa	4.95×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	esophagus mucosa	3.38×10-12 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	esophagus mucosa	5.95×10-6 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	esophagus mucosa	2.52×10-7 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	pancreas	4.35×10-10 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	muscularis mucosae	5.10×10-11 (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	muscularis mucosae	2.08×10-6 (p-value)	GTEx Portal V6	2

XXbac-BPG299F13.17	?	ENSG00000272501	muscularis mucosae	3.39×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	lung	1.54×10^{-15} (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	lung	5.45×10^{-6} (p-value)	GTEx Portal V6	2
CYP21A1P	?	ENSG00000204338	lung	3.27×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	lung	6.58×10^{-10} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	tibial artery	7.32×10^{-20} (p-value)	GTEx Portal V6	2
CCHCR1	?	ENSG00000204536	tibial artery	1.16×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	2.96×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	tibial artery	1.68×10^{-14} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	thyroid	6.25×10^{-18} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	thyroid	9.79×10^{-9} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	transverse colon	1.00×10^{-11} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	transverse colon	7.75×10^{-9} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	liver	1.05×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	terminal ileum	1.22×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	nucleus accumbens	1.37×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	sigmoid colon	5.76×10^{-8} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	vagina	2.96×10^{-7} (p-value)	GTEx Portal V6	2
AIF1	ENST00000466820	ILMN_1703538	blood	2.86×10^{-5} (p-value)	Westra et al.	2
AIF1	ENST00000376059					
HLA-C	?	ENSG00000204525	gastroesophageal junction	8.77×10^{-7} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	gastroesophageal junction	2.91×10^{-6} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	adrenal gland	5.91×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	cerebellum	4.73×10^{-9} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	hippocampus	6.32×10^{-8} (p-value)	GTEx Portal V6	2

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	215666_at	chr6	blood	1.40×10^{-9} (p-value)	Dixon et al.	2

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000048017		HCP5
		HCP5
		HCP5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000129758 (promoter flanking region)	2	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, DNase1, TAF7, PolII, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K4me2, H2AZ, H3K36me3
		blood (K562)	H3K27me3, Cfos
		skin (NHDF-AD)	DNase1, H3K27me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		A549	DNase1, H3K4me3
		skin (NHEK)	H3K4me3, DNase1, H3K4me2, H3K9ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HCP5	upstream gene variant	163	ENST00000467369	?	?	1
HCP5	upstream gene variant, downstream gene variant	163	ENST00000460889	?	?	2
HCP5	upstream gene variant	160	ENST00000541196	?	?	1
XXbac-BPG181B23.6	upstream gene variant	1012	ENST00000440087	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	2
HCP5	ENST00000467369	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
HCP5	ENST00000541196	?	1
HCP5	ENST00000414046	?	2
XXbac-BPG181B23.6	ENST00000440087	?	1