Locus 170 (rs2524280)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,327,701-31,426,399
block size	98,699 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.409 [-3.036 – 0.31]	gene(s) hit or close-by	HCP5, HLA-B, HLA-C, HLA-S, LINC01149, MICA, MIR6891, XXbac-BPG181B23.6, XXbac-BPG181B23.7, XXbac-BPG248L24.12, Y_RNA
phastCons	μ = 0.148 [0 – 0.796]	eQTL gene(s)	ATP6V1G2-DDX39B, DDX39B, HCG27, HLA-C, LY6G5C, MICA, MICB, POU5F1, PSORS1C3, XXbac-BPG299F13.17, Y_RNA
GERP++	μ = -0.556 [-2.85 – 0.641]	potentially regulated gene(s)	CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, FGFR3P1, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, SLC44A4, TCF19, TCF19, TCF19, TCF19, TCF19, TCF19, TCF19, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P
CADD score	μ = 4.560 [0.038 – 14.35]	disease gene(s)	HLA-B, , VARS2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Beta-2 microglubulin plasma levels	$<2.00\times10^{-12}$	GWAS Catalog	23417110	1
Bipolar disorder	$<1.00\times10^{-8}$	GWAS Catalog	21254220	1

Disease gene annotation

gene	trait	source DB	source entry/link	
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM®
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM®

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HCG27	ENST00000383331	ILMN_1746436	adipocyte	5.34×10^{-6} (p-value)	MuTHER consortium	6
HCG27	ENST00000415276					
MICB	ENST00000538442	ILMN_1708006	skin	3.69×10^{-5} (p-value)	MuTHER consortium	2
MICB	ENST00000399150					
MICB	ENST00000252229					
MICA	?	ENSG00000204520	muscularis mucosae	4.04×10^{-7} (p-value)	GTEx Portal V6	14
Y_RNA	?	ENSG00000199332	muscularis mucosae	2.82×10^{-6} (p-value)	GTEx Portal V6	6
MICA	?	ENSG00000204520	lung	4.47×10^{-9} (p-value)	GTEx Portal V6	14
PSORS1C3	?	ENSG00000204528	lung	1.30×10^{-6} (p-value)	GTEx Portal V6	4

Y_RNA	?	ENSG00000199332	lung	1.27×10-8 (p-value)	GTEx Portal V6	13
MICA	?	ENSG00000204520	transformed fibroblasts	1.29×10-5 (p-value)	GTEx Portal V6	2
Y_RNA	?	ENSG00000199332	transformed fibroblasts	1.19×10-7 (p-value)	GTEx Portal V6	14
PSORS1C3	?	ENSG00000204528	blood	1.47×10-7 (p-value)	GTEx Portal V6	14
MICA	?	ENSG00000204520	blood	1.39×10-6 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	tibial artery	1.10×10-7 (p-value)	GTEx Portal V6	14
Y_RNA	?	ENSG00000199332	tibial artery	3.29×10-8 (p-value)	GTEx Portal V6	14
MICA	?	ENSG00000204520	thyroid	2.25×10-6 (p-value)	GTEx Portal V6	12
PSORS1C3	?	ENSG00000204528	thyroid	2.39×10-9 (p-value)	GTEx Portal V6	15
PSORS1C3	?	ENSG00000204528	skeletal muscle	2.00×10-7 (p-value)	GTEx Portal V6	13
MICA	?	ENSG00000204520	sun exposed skin	1.57×10-9 (p-value)	GTEx Portal V6	15
PSORS1C3	?	ENSG00000204528	sun exposed skin	1.05×10-7 (p-value)	GTEx Portal V6	15
POU5F1	?	ENSG00000204531	sun exposed skin	5.91×10-8 (p-value)	GTEx Portal V6	14
HLA-C	?	ENSG00000204525	sun exposed skin	9.60×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	unexposed skin	1.58×10-6 (p-value)	GTEx Portal V6	4
PSORS1C3	?	ENSG00000204528	left ventricle	9.84×10-9 (p-value)	GTEx Portal V6	13
PSORS1C3	?	ENSG00000204528	subcutaneous adipocytes	1.54×10-6 (p-value)	GTEx Portal V6	13
XXbac-BPG299F13.17	?	ENSG00000272501	subcutaneous adipocytes	4.50×10-6 (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	stomach	4.18×10-7 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	tibial nerve	5.19×10-7 (p-value)	GTEx Portal V6	12
Y_RNA	?	ENSG00000199332	tibial nerve	5.49×10-8 (p-value)	GTEx Portal V6	14
PSORS1C3	?	ENSG00000204528	esophagus mucosa	5.04×10-6 (p-value)	GTEx Portal V6	8
POU5F1	?	ENSG00000204531	esophagus mucosa	5.68×10-7 (p-value)	GTEx Portal V6	14
DDX39B	ENST00000396172	ILMN_1686626	blood	1.27×10-4 (p-value)	Westra et al.	1
DDX39B	ENST00000482195					
DDX39B	ENST00000431908					
DDX39B	ENST00000484566					
DDX39B	ENST00000417023					
ATP6V1G2-DDX39B	ENST00000376185					
DDX39B	ENST00000462256					
DDX39B	ENST00000462421					
DDX39B	ENST00000478365					
DDX39B	ENST00000474961					
DDX39B	ENST00000376177					
DDX39B	ENST00000458640					
MICB	ENST00000538442	ILMN_1708006	blood	4.39×10-6 (p-value)	Westra et al.	1
MICB	ENST00000399150					
MICB	ENST00000252229					
DDX39B	?	ENSG00000198563	testis	3.71×10-6 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	testis	2.37×10-6 (p-value)	GTEx Portal V6	1

HLA-C	?	ENSG00000204525	testis	7.26×10-6 (p-value)	GTEx Portal V6	1
PSORS1C3	ENST00000412143	ILMN_3309634	b-cell	2.64×10-6 (p-value)	Fairfax et al.	3
LY6G5C	ENST00000474395	ILMN_1770209	b-cell	4.42×10-6 (p-value)	Fairfax et al.	3
LY6G5C	ENST00000461572					
LY6G5C	ENST00000467098					
MICA	ENST00000449934	ILMN_3241692	monocyte	3.05×10-8 (p-value)	Fairfax et al.	2
MICA	ENST00000616296		b-cell	1.13×10-5 (p-value)	Fairfax et al.	2
MICA	ENST00000421350					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444777	1	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
		ENCP00000048009	FGFR3P1
ENCE00000444715	1	ENCP00000047985	TCF19 TCF19 TCF19 TCF19 TCF19 TCF19 TCF19
		ENCP00000047968	VARS2 VARS2 VARS2 VARS2 VARS2 VARS2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494562 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1

ENSR00001494563 (promoter flanking region)	1	Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K79me2
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K79me2
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2
		nervous (NH-A)	H4K20me1
		skin (NHEK)	H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HCP5	upstream gene variant	4563	ENST00000467369	?	?	1
HCP5	upstream gene variant	4560	ENST00000541196	?	?	1
HCP5	upstream gene variant	2184	ENST00000414046	?	?	1
HCP5	upstream gene variant	4563	ENST00000460889	?	?	1
HLA-B	upstream gene variant	2736	ENST00000434333	?	ENSP00000405931	1
HLA-B	upstream gene variant	3019	ENST00000474381	?	?	1
HLA-B	upstream gene variant	2745	ENST00000498007	?	?	1
HLA-B	upstream gene variant	4848	ENST00000497377	?	?	1
HLA-B	upstream gene variant	2737	ENST00000412585	NM_005514.6	ENSP00000399168	1
HLA-B	upstream gene variant	4122	ENST00000463574	?	?	1
HLA-C	upstream gene variant	2766	ENST00000620806	?	ENSP00000480331	1
HLA-S	upstream gene variant	4173	ENST00000425174	?	?	1
LINC01149	upstream gene variant, downstream gene variant	2240	ENST00000430364	?	?	2
MICA	upstream gene variant	1266	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICA	upstream gene variant	989	ENST00000449934	NM_001177519.2	ENSP00000413079	2
MICA	upstream gene variant	1082	ENST00000421350	?	ENSP00000402410	2
MIR6891	upstream gene variant	4608	ENST00000618788	?	?	1
XXbac-BPG181B23.6	downstream gene variant	4106	ENST00000440087	?	?	1
XXbac-BPG181B23.7	downstream gene variant, upstream gene variant	2617	ENST00000606743	?	?	2
XXbac-BPG248L24.12	downstream gene variant	2287	ENST00000603274	?	?	1
Y_RNA	downstream gene variant, upstream gene variant	302	ENST00000362462	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	11
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	2
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	4
MICA	ENST00000421350	?	ENSP00000402410	2

Locus 170 (sentinel SNP rs2524280 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.