Locus 13 (rs2844511)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,378,768-31,390,410
block size	11,643 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.808 [-1.898 – 0.403]	gene(s) hit or close-by	HCP5, MICA
phastCons	μ = 0.008 [0 – 0.054]	eQTL gene(s)	ATP6V1G2, HCG22, HCG27, HLA-B, HLA-C, LTA, MICA, PSORS1C1, PSORS1C2, TAP2, WASF5P, XXbac-BPG181B23.7, XXbac-BPG246D15.9, XXbac-BPG248L24.10, XXbac-BPG248L24.12, XXbac-BPG248L24.13, XXbac-BPG299F13.17, Y_RNA
GERP++	μ = -0.466 [-3.52 – 1.18]	potentially regulated gene(s)	MICA, MICA, MICA, MICA, MICA, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2
CADD score	μ = 2.526 [0.257 – 6.359]	disease gene(s)	HLA-B, TAP2, LTA, , , , , VARS2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
CERVICAL CANCER	$<4.00\times10^{-18}$	GWAS Catalog	23482656		1
Height	$<8.00\times10^{-29}$	GWAS Catalog	20881960		1
cholesterol	4.59×10^{-6}	Metabolomics GWAS Server	24816252		5
Behcet syndrome	4.29×10^{-8}	dbGaP	pha002888	dbGaP	2
Lupus erythematosus, systemic	3.60×10^{-9}	dbGaP	pha002848	dbGaP	2

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
TAP2	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592	orphanet
LTA	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988	OMIM
LTA	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
TAP2	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICA	?	ENSG00000204520	atrial appendage	3.61×10^{-9} (p-value)	GTEx Portal V6	9

HLA-B	?	ENSG00000234745	atrial appendage	1.28×10-12 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	atrial appendage	4.65×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	transformed fibroblasts	2.01×10-26 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	transformed fibroblasts	6.93×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	transformed fibroblasts	3.01×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	1.83×10-6 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	4.38×10-20 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	blood	4.96×10-6 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	blood	4.13×10-17 (p-value)	GTEx Portal V6	9
XXbac-BPG299F13.17	?	ENSG00000272501	blood	2.58×10-6 (p-value)	GTEx Portal V6	8
MICA	?	ENSG00000204520	blood	5.66×10-8 (p-value)	GTEx Portal V6	8
XXbac-BPG248L24.12	?	ENSG00000271581	blood	3.24×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	blood	2.00×10-9 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	breast	1.20×10-10 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	breast	5.82×10-7 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	breast	1.36×10-10 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	breast	2.85×10-6 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	breast	7.91×10-14 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	skeletal muscle	1.53×10-20 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	skeletal muscle	1.22×10-10 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	skeletal muscle	5.77×10-12 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	2.95×10-10 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	skeletal muscle	2.71×10-17 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	putamen	1.12×10-7 (p-value)	GTEx Portal V6	8
MICA	?	ENSG00000204520	EBV lymphocytes	6.51×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	EBV lymphocytes	4.39×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	EBV lymphocytes	1.40×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	ovary	1.40×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	ovary	1.32×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	sun exposed skin	2.64×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	5.30×10-18 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	sun exposed skin	6.34×10-14 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	5.95×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	sun exposed skin	5.96×10-11 (p-value)	GTEx Portal V6	9
PSORS1C1	?	ENSG00000204540	sun exposed skin	4.06×10-9 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	unexposed skin	6.10×10-10 (p-value)	GTEx Portal V6	9

HLA-C	?	ENSG00000204525	unexposed skin	5.60×10^{-6} (p-value)	GTEx Portal V6	3
HLA-B	?	ENSG00000234745	unexposed skin	4.17×10^{-10} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	unexposed skin	1.13×10^{-9} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	unexposed skin	6.69×10^{-12} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	aorta	2.49×10^{-14} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	aorta	7.96×10^{-7} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	aorta	2.87×10^{-13} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	aorta	4.29×10^{-13} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	left ventricle	3.03×10^{-15} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	left ventricle	7.87×10^{-8} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	1.90×10^{-8} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	left ventricle	1.27×10^{-11} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	left ventricle	3.93×10^{-6} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	subcutaneous adipocytes	6.44×10^{-16} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	2.42×10^{-6} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	subcutaneous adipocytes	1.88×10^{-13} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	6.16×10^{-7} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	1.09×10^{-22} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	caudate basal ganglia	1.37×10^{-11} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	caudate basal ganglia	7.92×10^{-12} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	visceral adipocytes	1.94×10^{-10} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	visceral adipocytes	7.68×10^{-8} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	visceral adipocytes	1.41×10^{-11} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	3.58×10^{-7} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	visceral adipocytes	2.01×10^{-11} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	stomach	5.75×10^{-11} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	stomach	1.01×10^{-6} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	stomach	6.69×10^{-8} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	stomach	3.80×10^{-8} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	stomach	5.92×10^{-12} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	cortex	5.99×10^{-8} (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	tibial nerve	9.55×10^{-18} (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	tibial nerve	3.35×10^{-8} (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	tibial nerve	5.58×10^{-13} (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	5.88×10^{-6} (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	1.36×10^{-15} (p-value)	GTEx Portal V6	9
XXbac-BPG299F13.17	?	ENSG00000272501	tibial nerve	6.67×10^{-6} (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	esophagus mucosa	1.45×10^{-10} (p-value)	GTEx Portal V6	9

HLA-C	?	ENSG00000204525	esophagus mucosa	1.45×10-10 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	esophagus mucosa	7.05×10-13 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	esophagus mucosa	4.62×10-13 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	8.48×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	esophagus mucosa	3.06×10-7 (p-value)	GTEx Portal V6	9
PSORS1C1	?	ENSG00000204540	esophagus mucosa	1.73×10-6 (p-value)	GTEx Portal V6	9
HCG22	?	ENSG00000228789	esophagus mucosa	2.62×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	pituitary	2.24×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	testis	5.59×10-13 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	testis	3.13×10-6 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	testis	3.30×10-11 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	frontal cortex	3.60×10-7 (p-value)	GTEx Portal V6	8
XXbac-BPG248L24.10	?	ENSG00000229836	pancreas	6.76×10-7 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	pancreas	4.51×10-6 (p-value)	GTEx Portal V6	6
XXbac-BPG181B23.7	?	ENSG00000272221	pancreas	1.83×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.13	?	ENSG00000256166	pancreas	2.19×10-6 (p-value)	GTEx Portal V6	7
MICA	?	ENSG00000204520	pancreas	6.13×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	pancreas	3.12×10-6 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	pancreas	4.16×10-8 (p-value)	GTEx Portal V6	9
WASF5P	?	ENSG00000231402	pancreas	1.44×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	muscularis mucosae	5.92×10-13 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	muscularis mucosae	7.16×10-7 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	muscularis mucosae	3.10×10-12 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	muscularis mucosae	6.11×10-18 (p-value)	GTEx Portal V6	9
PSORS1C2	?	ENSG00000204538	lung	1.07×10-6 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	lung	4.97×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	lung	2.02×10-17 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	lung	7.89×10-20 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	lung	1.65×10-10 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	lung	3.16×10-12 (p-value)	GTEx Portal V6	9
PSORS1C1	?	ENSG00000204540	lung	5.94×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial artery	7.30×10-24 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	tibial artery	3.23×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	tibial artery	2.88×10-16 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	1.88×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	8.90×10-23 (p-value)	GTEx Portal V6	9
PSORS1C2	?	ENSG00000204538	thyroid	3.56×10-7 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	thyroid	7.34×10-7 (p-value)	GTEx Portal V6	9

XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	6.91×10-19 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	thyroid	1.74×10-20 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	1.48×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	thyroid	1.64×10-11 (p-value)	GTEx Portal V6	9
Y_RNA	?	ENSG00000199332	thyroid	4.47×10-7 (p-value)	GTEx Portal V6	9
PSORS1C1	?	ENSG00000204540	thyroid	5.12×10-8 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	transverse colon	8.76×10-12 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	transverse colon	1.07×10-15 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	liver	9.37×10-10 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	liver	1.25×10-6 (p-value)	GTEx Portal V6	6
XXbac-BPG181B23.7	?	ENSG00000272221	terminal ileum	9.54×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	spleen	2.32×10-6 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	spleen	5.35×10-7 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	sigmoid colon	3.77×10-9 (p-value)	GTEx Portal V6	9
HLA-C	?	ENSG00000204525	sigmoid colon	2.37×10-7 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	sigmoid colon	3.35×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG248L24.12	?	ENSG00000271581	sigmoid colon	9.65×10-7 (p-value)	GTEx Portal V6	7
XXbac-BPG181B23.7	?	ENSG00000272221	sigmoid colon	6.00×10-9 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	coronary artery	1.87×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	coronary artery	1.68×10-9 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	gastroesophageal junction	2.59×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	gastroesophageal junction	7.09×10-8 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	gastroesophageal junction	3.26×10-8 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	cerebellar hemisphere	1.70×10-8 (p-value)	GTEx Portal V6	9
MICA	?	ENSG00000204520	adrenal gland	1.10×10-10 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	adrenal gland	1.45×10-9 (p-value)	GTEx Portal V6	9
HLA-B	?	ENSG00000234745	cerebellum	1.00×10-10 (p-value)	GTEx Portal V6	9
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	7.71×10-7 (p-value)	MuTHER consortium	6
ATP6V1G2	ENST00000303892					
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					
HCG27	ENST00000383331	ILMN_1746436	adipocyte	4.16×10-7 (p-value)	MuTHER consortium	6
HCG27	ENST00000415276					
LTA	ENST00000418386	ILMN_1795464	blood	1.20×10-4 (p-value)	Westra et al.	2
LTA	ENST00000471842					
LTA	ENST00000454783					
LTA	ENST00000489638					
MICA	ENST00000449934	ILMN_3241692	monocyte	4.42×10-5 (p-value)	Fairfax et al.	4
MICA	ENST00000616296					
MICA	ENST00000421350					

TAP2	ENST00000374899	ILMN_1759250	monocyte	4.61×10-4 (p-value)	Fairfax et al.	4
TAP2	ENST00000620123					
TAP2	ENST00000374897					
TAP2	ENST00000464100					
XXbac-BPG246D15.9	ENST00000452392					
XXbac-BPG181B23.7	ENST00000606743	230937_at	blood	1.10×10-10 (p-value)	Dixon et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444714	1	ENCP00000048015	MICA MICA MICA MICA MICA
		ENCP00000047968	VARS2 VARS2 VARS2 VARS2 VARS2 VARS2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MICA	downstream gene variant	2393	ENST00000449934	NM_001177519.2	ENSP00000413079	3
MICA	downstream gene variant	2395	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	3
MICA	downstream gene variant	2393	ENST00000421350	?	ENSP00000402410	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	9
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	4
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	4
MICA	ENST00000421350	?	ENSP00000402410	4