Locus 271 (rs4418214)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,380,034-31,391,401
block size	11,368 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.000 [–]	gene(s) hit or close-by	HCP5, MICA
phastCons	μ = 0.000 [–]	eQTL gene(s)	MICA, MICB, PSORS1C2, TNXA, TNXB, XXbac-BPG181B23.7
GERP++	μ = -1.225 [-2.45 – 0]	potentially regulated gene(s)	–
CADD score	μ = 4.632 [4.233 – 5.031]	disease gene(s)	TNXB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
HIV-1 susceptibility	$<4.00\times10^{-11}$	GWAS Catalog	23935489	1
HIV-1 control	$<1.00\times10^{-34}$	GWAS Catalog	21051598	1

Disease gene annotation

gene	trait	source DB	source entry/link
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICA	?	ENSG00000204520	pancreas	2.01×10^{-12} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	pancreas	3.75×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	muscularis mucosae	3.13×10^{-10} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	muscularis mucosae	6.11×10^{-13} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	muscularis mucosae	4.75×10^{-7} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	lung	1.75×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	lung	6.14×10^{-14} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	lung	3.14×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	atrial appendage	6.38×10^{-10} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	transformed fibroblasts	6.61×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	2.25×10^{-16} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	transformed fibroblasts	4.12×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	blood	1.23×10^{-11} (p-value)	GTEx Portal V6	2

XXbac-BPG181B23.7	?	ENSG00000272221	blood	7.53×10^{-13} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	blood	1.78×10^{-6} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	breast	1.18×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	breast	3.41×10^{-11} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	breast	8.43×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial artery	4.75×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	1.80×10^{-17} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	tibial artery	7.33×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	thyroid	7.64×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	1.88×10^{-15} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	thyroid	2.47×10^{-9} (p-value)	GTEx Portal V6	2
PSORS1C2	?	ENSG00000204538	thyroid	2.21×10^{-6} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	skeletal muscle	7.03×10^{-10} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	skeletal muscle	9.61×10^{-11} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	prostate	2.29×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	prostate	8.17×10^{-12} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	transverse colon	3.92×10^{-9} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	transverse colon	1.18×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	sun exposed skin	9.48×10^{-16} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	6.43×10^{-16} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	sun exposed skin	6.95×10^{-9} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	unexposed skin	5.77×10^{-9} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	unexposed skin	1.22×10^{-10} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	aorta	4.89×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	aorta	2.71×10^{-9} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	left ventricle	1.81×10^{-11} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	left ventricle	6.68×10^{-15} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	subcutaneous adipocytes	5.34×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	1.04×10^{-12} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	subcutaneous adipocytes	1.29×10^{-9} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	terminal ileum	5.11×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	terminal ileum	3.42×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	caudate basal ganglia	1.06×10^{-6} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	stomach	2.56×10^{-14} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	stomach	2.18×10^{-11} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	visceral adipocytes	4.85×10^{-10} (p-value)	GTEx Portal V6	2

XXbac-BPG181B23.7	?	ENSG00000272221	visceral adipocytes	3.15×10^{-11} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	spleen	1.65×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	sigmoid colon	3.37×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	coronary artery	6.58×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	coronary artery	2.18×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial nerve	8.74×10^{-17} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	3.76×10^{-13} (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	tibial nerve	1.52×10^{-9} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	esophagus mucosa	3.15×10^{-11} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	esophagus mucosa	4.80×10^{-12} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	gastroesophageal junction	1.40×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	gastroesophageal junction	3.56×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	adrenal gland	9.32×10^{-10} (p-value)	GTEx Portal V6	2
TNXB	?	ENSG00000168477	adrenal gland	2.07×10^{-6} (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	adrenal gland	3.08×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	testis	1.12×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	testis	3.86×10^{-12} (p-value)	GTEx Portal V6	2
MICB	ENST00000538442	ILMN_1708006	skin	8.50×10^{-10} (p-value)	MuTHER consortium	1
MICB	ENST00000399150		blood	3.34×10^{-6} (p-value)	MuTHER consortium	1
MICB	ENST00000252229					
MICB	ENST00000399150	ILMN_1708006	monocyte	3.63×10^{-6} (p-value)	Fairfax et al.	1
MICB	ENST00000538442		b-cell	4.35×10^{-9} (p-value)	Fairfax et al.	1
MICB	ENST00000252229					

Putative effect on transcript

Intron variant

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