Locus 149 (rs17206736)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,377,642-31,391,257
block size	13,616 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.284 [-1.521 – 0.426]	gene(s) hit or close-by	HCP5, MICA
phastCons	μ = 0.057 [0 – 0.409]	eQTL gene(s)	CCHCR1, HLA-B, MICA, PSORS1C1, PSORS1C2, Y_RNA
GERP++	μ = -0.195 [-1.75 – 0.536]	potentially regulated gene(s)	MICA, MICA, MICA, MICA, MICA, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2
CADD score	μ = 2.861 [0.826 – 9.209]	disease gene(s)	, , , VARS2, HLA-B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
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)
MICA	?	ENSG00000204520	muscularis mucosae	6.76×10^{-8} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	lung	6.56×10^{-12} (p-value)	GTEx Portal V6	12
PSORS1C2	?	ENSG00000204538	lung	8.23×10^{-7} (p-value)	GTEx Portal V6	1
PSORS1C1	?	ENSG00000204540	lung	7.32×10^{-7} (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	lung	2.71×10^{-8} (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	atrial appendage	1.02×10^{-8} (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	transformed fibroblasts	1.36×10^{-12} (p-value)	GTEx Portal V6	11
HLA-B	?	ENSG00000234745	transformed fibroblasts	5.82×10^{-6} (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	transformed fibroblasts	1.00×10^{-9} (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	tibial artery	7.30×10^{-11} (p-value)	GTEx Portal V6	11
Y_RNA	?	ENSG00000199332	tibial artery	2.46×10^{-12} (p-value)	GTEx Portal V6	12

MICA	?	ENSG00000204520	breast	2.71×10-10 (p-value)	GTEx Portal V6	10
MICA	?	ENSG00000204520	blood	4.42×10-11 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	thyroid	5.40×10-12 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	skeletal muscle	1.18×10-12 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	EBV lymphocytes	2.60×10-8 (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	EBV lymphocytes	3.18×10-7 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	sun exposed skin	3.28×10-11 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	aorta	1.80×10-8 (p-value)	GTEx Portal V6	11
Y_RNA	?	ENSG00000199332	aorta	1.92×10-10 (p-value)	GTEx Portal V6	11
MICA	?	ENSG00000204520	left ventricle	5.23×10-9 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	subcutaneous adipocytes	3.15×10-9 (p-value)	GTEx Portal V6	10
Y_RNA	?	ENSG00000199332	subcutaneous adipocytes	3.69×10-8 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	caudate basal ganglia	5.43×10-8 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	visceral adipocytes	2.59×10-8 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial nerve	9.72×10-14 (p-value)	GTEx Portal V6	11
CCHCR1	?	ENSG00000204536	tibial nerve	2.40×10-6 (p-value)	GTEx Portal V6	1
PSORS1C1	?	ENSG00000204540	tibial nerve	9.33×10-6 (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	tibial nerve	1.35×10-8 (p-value)	GTEx Portal V6	4
Y_RNA	?	ENSG00000199332	sigmoid colon	1.96×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	esophagus mucosa	1.52×10-10 (p-value)	GTEx Portal V6	1
Y_RNA	?	ENSG00000199332	esophagus mucosa	3.49×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	testis	2.92×10-7 (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)
ENCE00000444713	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	39	ENST00000449934	NM_001177519.2	ENSP00000413079	4

MICA	downstream gene variant	41	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	4
MICA	downstream gene variant	39	ENST00000421350	?	ENSP00000402410	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5	ENST00000414046	?	?	13
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	ENST00000421350	?	ENSP00000402410	1