Locus 175 (rs2516470)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,407,331-31,407,331
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.181	gene(s) hit or close-by	HCP5, LINC01149
phastCons	0.004	eQTL gene(s)	ATP6V1G2, C4A, HCG22, HCG27, HCP5, HLA-B, LST1, LY6G5C, MICA, MICB, NOTCH4, PRRC2A, XXbac-BPG181B23.7, XXbac-BPG248L24.12
GERP++	0.574	potentially regulated gene(s)	–
CADD score	0.943	disease gene(s)	C4A, HLA-B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147
C4A	Behcet disease	OrphaNet	OrphaNet:117
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)
XXbac-BPG181B23.7	?	ENSG00000272221	pancreas	1.41×10^{-6} (p-value)	GTEx Portal V6	1
LST1	ENST00000376099	ILMN_2345353	blood	1.26×10^{-6} (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					

LST1	ENST00000376089					
LST1	ENST00000376096					
LST1	ENST00000418507					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000460834					
LST1	ENST00000211921					
NOTCH4	ENST00000474612	ILMN_1711157	adipocyte	2.39×10-6 (p-value)	MuTHER consortium	1
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
LST1	ENST00000376099	ILMN_1688373	blood	1.10×10-6 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000339530					
LST1	ENST00000419073					
LST1	ENST00000376110					
LST1	ENST00000376086					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000376090					
LST1	ENST00000396101					
LST1	ENST00000376092					
LST1	ENST00000303757					
LST1	ENST00000376093					
LST1	ENST00000376089					
LST1	ENST00000396112					
LST1	ENST00000418507					
LST1	ENST00000376096					
LST1	ENST00000490742					
LST1	ENST00000211921					
LST1	ENST00000460834					
HCG22	ENST00000570223	ILMN_1667229	blood	1.22×10-14 (p-value)	MuTHER consortium	1
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
PRRC2A	ENST00000376033	ILMN_2408179	blood	4.00×10-5 (p-value)	MuTHER consortium	1
PRRC2A	ENST00000487089					
PRRC2A	ENST00000376007					
PRRC2A	ENST00000487839					
PRRC2A	ENST00000462617					
PRRC2A	ENST00000482441					
PRRC2A	ENST00000492691					
HCG27	ENST00000383331	ILMN_1746436	blood	9.73×10-6 (p-value)	MuTHER consortium	1
HCG27	ENST00000415276					
MICA	?	ENSG00000204520	muscularis mucosae	2.27×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	muscularis mucosae	1.74×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	muscularis mucosae	4.11×10-13 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	lung	1.18×10-8 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	lung	5.02×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	lung	5.34×10-14 (p-value)	GTEx Portal V6	1

XXbac-BPG181B23.7	?	ENSG00000272221	atrial appendage	3.15×10^{-9} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	transformed fibroblasts	1.05×10^{-10} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	3.16×10^{-9} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	1.22×10^{-16} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	blood	1.68×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	blood	3.05×10^{-11} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	blood	1.02×10^{-22} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	breast	4.04×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	breast	1.95×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	breast	3.01×10^{-13} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial artery	8.19×10^{-13} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	1.04×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	7.67×10^{-25} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	thyroid	2.48×10^{-12} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	1.15×10^{-8} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	1.74×10^{-18} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	skeletal muscle	9.01×10^{-14} (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	skeletal muscle	2.97×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	5.23×10^{-9} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	skeletal muscle	3.27×10^{-26} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	EBV lymphocytes	3.23×10^{-9} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	transverse colon	1.40×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	transverse colon	1.05×10^{-10} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	ovary	2.40×10^{-7} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	sun exposed skin	6.42×10^{-11} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	1.40×10^{-7} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	1.13×10^{-21} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	unexposed skin	3.40×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	unexposed skin	1.11×10^{-12} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	aorta	2.08×10^{-8} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	aorta	2.04×10^{-11} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	left ventricle	1.18×10^{-8} (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	4.42×10^{-6} (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	left ventricle	3.36×10^{-13} (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	subcutaneous adipocytes	2.68×10^{-10} (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	subcutaneous adipocytes	6.39×10^{-6} (p-value)	GTEx Portal V6	1

XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	1.99×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	3.18×10-19 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	stomach	1.34×10-10 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	visceral adipocytes	5.49×10-12 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	cortex	1.59×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	sigmoid colon	5.87×10-14 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	coronary artery	5.37×10-10 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial nerve	6.18×10-9 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	1.05×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	4.76×10-14 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	tibial nerve	7.59×10-6 (p-value)	GTEx Portal V6	1
HCG22	?	ENSG00000228789	tibial nerve	2.39×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	esophagus mucosa	1.40×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	3.86×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	esophagus mucosa	1.15×10-12 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	gastroesophageal junction	9.01×10-10 (p-value)	GTEx Portal V6	1
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	1.07×10-4 (p-value)	Westra et al.	1
ATP6V1G2	ENST00000303892					
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					
HCP5	ENST00000414046	ILMN_1803945	blood	2.10×10-5 (p-value)	Westra et al.	1
HCP5	ENST00000541196					
LY6G5C	ENST00000383237	ILMN_1763467	blood	1.04×10-5 (p-value)	Westra et al.	1
LY6G5C	ENST00000474395					
LY6G5C	ENST00000375863					
LY6G5C	ENST00000474678					
LY6G5C	ENST00000467098					
LY6G5C	ENST00000460141					
?	?	ILMN_1883997	blood	9.46×10-7 (p-value)	Westra et al.	1
XXbac-BPG181B23.7	?	ENSG00000272221	adrenal gland	1.48×10-7 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	testis	2.87×10-6 (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	testis	6.24×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	testis	5.88×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LINC01149	upstream gene variant	2113	ENST00000430364	?	?	1

Putative effect on transcript

Intron variant

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