

Block annotations

Block info





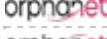

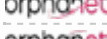
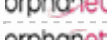

genomic range	chr6:31,407,331-31,407,331 <i>e!</i>
block size	1 bp
variant count	1 variants

Basic features

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

Trait annotations

Disease gene annotation






































gene	trait	source DB	source entry/link
C4A <i>e!</i>	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380 
C4A <i>e!</i>	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374 
C4A <i>e!</i>	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700 
C4A <i>e!</i>	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790 
C4A <i>e!</i>	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147 
C4A <i>e!</i>	Behcet disease	OrphaNet	OrphaNet:117 
HLA-B <i>e!</i>	Takayasu arteritis	OrphaNet	OrphaNet:3287 
HLA-B <i>e!</i>	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426 
HLA-B <i>e!</i>	Behcet disease	OrphaNet	OrphaNet:117 

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG181B23.7 <i>e!</i>	?	ENSG00000272221 <i>e!</i>	pancreas	1.41×10 ⁻⁶ (p-value)	GTEx Portal V6 	1
LST1 <i>e!</i>	ENST00000376099 <i>e!</i>	ILMN_2345353 <i>e!</i>	blood	1.26×10 ⁻⁶ (p-value)	MuTHER consortium 	1
LST1 <i>e!</i>	ENST00000464526 <i>e!</i>					
LST1 <i>e!</i>	ENST00000339530 <i>e!</i>					
LST1 <i>e!</i>	ENST00000419073 <i>e!</i>					
LST1 <i>e!</i>	ENST00000376110 <i>e!</i>					
LST1 <i>e!</i>	ENST00000376086 <i>e!</i>					
LST1 <i>e!</i>	ENST00000433492 <i>e!</i>					
LST1 <i>e!</i>	ENST00000438075 <i>e!</i>					
LST1 <i>e!</i>	ENST00000464044 <i>e!</i>					
LST1 <i>e!</i>	ENST00000376090 <i>e!</i>					
LST1 <i>e!</i>	ENST00000396101 <i>e!</i>					
LST1 <i>e!</i>	ENST00000376092 <i>e!</i>					
LST1 <i>e!</i>	ENST00000303757 <i>e!</i>					
LST1 <i>e!</i>	ENST00000376093 <i>e!</i>					

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

XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	atrial appendage	3.15×10 ⁻⁹ (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	transformed fibroblasts	1.05×10 ⁻¹⁰ (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	transformed fibroblasts	3.16×10 ⁻⁹ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	transformed fibroblasts	1.22×10 ⁻¹⁶ (p-value)	GTEx Portal V6 	1
C4A <i>e!</i> ?	ENSG00000244731 <i>e!</i>	blood	1.68×10 ⁻⁶ (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	blood	3.05×10 ⁻¹¹ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	blood	1.02×10 ⁻²² (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	breast	4.04×10 ⁻⁷ (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	breast	1.95×10 ⁻⁷ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	breast	3.01×10 ⁻¹³ (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	tibial artery	8.19×10 ⁻¹³ (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	tibial artery	1.04×10 ⁻⁷ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	tibial artery	7.67×10 ⁻²⁵ (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	thyroid	2.48×10 ⁻¹² (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	thyroid	1.15×10 ⁻⁸ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	thyroid	1.74×10 ⁻¹⁸ (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	skeletal muscle	9.01×10 ⁻¹⁴ (p-value)	GTEx Portal V6 	1
HLA-B <i>e!</i> ?	ENSG00000234745 <i>e!</i>	skeletal muscle	2.97×10 ⁻⁷ (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	skeletal muscle	5.23×10 ⁻⁹ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	skeletal muscle	3.27×10 ⁻²⁶ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	EBV lymphocytes	3.23×10 ⁻⁹ (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	transverse colon	1.40×10 ⁻⁶ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	transverse colon	1.05×10 ⁻¹⁰ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	ovary	2.40×10 ⁻⁷ (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	sun exposed skin	6.42×10 ⁻¹¹ (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	sun exposed skin	1.40×10 ⁻⁷ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	sun exposed skin	1.13×10 ⁻²¹ (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	unexposed skin	3.40×10 ⁻⁶ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	unexposed skin	1.11×10 ⁻¹² (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	aorta	2.08×10 ⁻⁸ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	aorta	2.04×10 ⁻¹¹ (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	left ventricle	1.18×10 ⁻⁸ (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	left ventricle	4.42×10 ⁻⁶ (p-value)	GTEx Portal V6 	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	left ventricle	3.36×10 ⁻¹³ (p-value)	GTEx Portal V6 	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	subcutaneous adipocytes	2.68×10 ⁻¹⁰ (p-value)	GTEx Portal V6 	1
HLA-B <i>e!</i> ?	ENSG00000234745 <i>e!</i>	subcutaneous adipocytes	6.39×10 ⁻⁶ (p-value)	GTEx Portal V6 	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	subcutaneous adipocytes	1.00×10 ⁻⁶ (p-value)	GTEx Portal V6 	1

XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	subcutaneous adipocytes	1.99×10 ⁻⁶ (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	subcutaneous adipocytes	3.18×10 ⁻¹⁹ (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	stomach	1.34×10 ⁻¹⁰ (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	visceral adipocytes	5.49×10 ⁻¹² (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	cortex	1.59×10 ⁻⁷ (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	sigmoid colon	5.87×10 ⁻¹⁴ (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	coronary artery	5.37×10 ⁻¹⁰ (p-value)	GTEx Portal V6	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	tibial nerve	6.18×10 ⁻⁹ (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	tibial nerve	1.05×10 ⁻⁷ (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	tibial nerve	4.76×10 ⁻¹⁴ (p-value)	GTEx Portal V6	1
MICB <i>e!</i> ?	ENSG00000204516 <i>e!</i>	tibial nerve	7.59×10 ⁻⁶ (p-value)	GTEx Portal V6	1
HCG22 <i>e!</i> ?	ENSG00000228789 <i>e!</i>	tibial nerve	2.39×10 ⁻⁶ (p-value)	GTEx Portal V6	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	esophagus mucosa	1.40×10 ⁻⁶ (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12 <i>e!</i> ?	ENSG00000271581 <i>e!</i>	esophagus mucosa	3.86×10 ⁻⁷ (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	esophagus mucosa	1.15×10 ⁻¹² (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	gastroesophageal junction	9.01×10 ⁻¹⁰ (p-value)	GTEx Portal V6	1
ATP6V1G2 <i>e!</i>	ENST00000481998 <i>e!</i>	ILMN_1654541 <i>e!</i> blood	1.07×10 ⁻⁴ (p-value)	Westra et al.	1
ATP6V1G2 <i>e!</i>	ENST00000303892 <i>e!</i>				
ATP6V1G2 <i>e!</i>	ENST00000483170 <i>e!</i>				
ATP6V1G2 <i>e!</i>	ENST00000376151 <i>e!</i>				
HCP5 <i>e!</i>	ENST00000414046 <i>e!</i>	ILMN_1803945 <i>e!</i> blood	2.10×10 ⁻⁵ (p-value)	Westra et al.	1
HCP5 <i>e!</i>	ENST00000541196 <i>e!</i>				
LY6G5C <i>e!</i>	ENST00000383237 <i>e!</i>	ILMN_1763467 <i>e!</i> blood	1.04×10 ⁻⁵ (p-value)	Westra et al.	1
LY6G5C <i>e!</i>	ENST00000474395 <i>e!</i>				
LY6G5C <i>e!</i>	ENST00000375863 <i>e!</i>				
LY6G5C <i>e!</i>	ENST00000474678 <i>e!</i>				
LY6G5C <i>e!</i>	ENST00000467098 <i>e!</i>				
LY6G5C <i>e!</i>	ENST00000460141 <i>e!</i>				
? ?	ILMN_1883997 <i>e!</i>	blood	9.46×10 ⁻⁷ (p-value)	Westra et al.	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	adrenal gland	1.48×10 ⁻⁷ (p-value)	GTEx Portal V6	1
MICA <i>e!</i> ?	ENSG00000204520 <i>e!</i>	testis	2.87×10 ⁻⁶ (p-value)	GTEx Portal V6	1
HLA-B <i>e!</i> ?	ENSG00000234745 <i>e!</i>	testis	6.24×10 ⁻⁷ (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7 <i>e!</i> ?	ENSG00000272221 <i>e!</i>	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 <i>e!</i>	upstream gene variant	2113	ENST00000430364 <i>e!</i>	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCP5 <i>e!</i>	ENST00000414046 <i>e!</i>	?	?	1

