Locus 387 (rs2853928)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,242,089-31,328,542
block size	86,454 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.879 [-3.321 – 0.343]	**gene(s) hit or close-by**	HLA-B, HLA-C, MIR6891, RPL3P2, USP8P1, WASF5P, XXbac-BPG248L24.10, XXbac-BPG248L24.12, XXbac-BPG248L24.13
phastCons	μ = 0.045 [0 – 0.398]	**eQTL gene(s)**	ATP6V1G2, BAG6, C4B, CCHCR1, CD248, CSF3R, CYP21A1P, DDX39B, FLOT1, HCG22, HCG27, HLA-B, HLA-C, LINC00243, MICA, MICB, PSORS1C1, PSORS1C2, PSORS1C3, TCF19, TMEM154, TNXB, TSSC4, VARS2, WASF5P, XRCC6, XXbac-BPG181B23.7, XXbac-BPG248L24.12, XXbac-BPG248L24.13, XXbac-BPG299F13.17
GERP++	μ = -0.521 [-3.27 – 0.497]	**potentially regulated gene(s)**	–
CADD score	μ = 2.610 [0.13 – 11.44]	**disease gene(s)**	VARS2, CSF3R, C4B, HLA-B, TNXB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Height	$<4.00\times10^{-12}$	GWAS Catalog	23563607	1
Triglycerides	$<2.00\times10^{-15}$	GWAS Catalog	20686565	1
CD4:CD8 lymphocyte ratio	$<2.00\times10^{-28}$	GWAS Catalog	20045101	1
Blood pressure	7.09×10^{-5}	dbGaP	pha003050	2
Lupus erythematosus, systemic	5.16×10^{-10}	dbGaP	pha002848	1

Disease gene annotation

gene	trait	source DB	source entry/link	
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
CSF3R	NEUTROPHILIA, HEREDITARY	OMIM	MIM:162830	OMIM
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
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
CSF3R	Chronic neutrophilic leukemia	OrphaNet	OrphaNet:86829	orphanet
CSF3R	Hereditary neutrophilia	OrphaNet	OrphaNet:279943	orphanet
CSF3R	Atypical chronic myeloid leukemia	OrphaNet	OrphaNet:98824	orphanet
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839	orphanet
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HLA-C	?	ENSG00000204525	atrial appendage	4.49×10-32 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	atrial appendage	6.95×10-23 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	atrial appendage	3.34×10-7 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	atrial appendage	2.37×10-10 (p-value)	GTEx Portal V6	15
MICA	?	ENSG00000204520	atrial appendage	2.44×10-6 (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	transformed fibroblasts	1.05×10-14 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	transformed fibroblasts	2.71×10-10 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	transformed fibroblasts	2.18×10-71 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	3.70×10-43 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	transformed fibroblasts	4.10×10-7 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	transformed fibroblasts	4.97×10-12 (p-value)	GTEx Portal V6	16
MICA	?	ENSG00000204520	transformed fibroblasts	2.05×10-8 (p-value)	GTEx Portal V6	15
C4B	?	ENSG00000224389	transformed fibroblasts	4.47×10-6 (p-value)	GTEx Portal V6	3
HCG27	?	ENSG00000206344	blood	1.91×10-9 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	blood	1.42×10-53 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	blood	3.70×10-12 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	blood	3.93×10-60 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	blood	8.36×10-16 (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	blood	7.20×10-9 (p-value)	GTEx Portal V6	16
LINC00243	?	ENSG00000214894	blood	7.76×10-8 (p-value)	GTEx Portal V6	16
XXbac-BPG181B23.7	?	ENSG00000272221	blood	7.99×10-7 (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	breast	7.69×10-8 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	breast	2.03×10-37 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	breast	5.30×10-7 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	breast	4.50×10-31 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	breast	2.78×10-14 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	breast	9.37×10-10 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	breast	7.76×10-7 (p-value)	GTEx Portal V6	14
WASF5P	?	ENSG00000231402	breast	1.35×10-6 (p-value)	GTEx Portal V6	8
MICA	?	ENSG00000204520	breast	1.42×10-6 (p-value)	GTEx Portal V6	3
HCG27	?	ENSG00000206344	skeletal muscle	6.99×10-11 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	skeletal muscle	2.75×10-95 (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	skeletal muscle	1.88×10-6 (p-value)	GTEx Portal V6	11
HLA-B	?	ENSG00000234745	skeletal muscle	1.10×10-22 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	4.19×10-64 (p-value)	GTEx Portal V6	16
MICA	?	ENSG00000204520	skeletal muscle	8.21×10-8 (p-value)	GTEx Portal V6	15
PSORS1C1	?	ENSG00000204540	skeletal muscle	9.28×10-7 (p-value)	GTEx Portal V6	15

XXbac-BPG299F13.17	?	ENSG00000272501	skeletal muscle	5.71×10^{-6} (p-value)	GTEx Portal V6	6
HLA-C	?	ENSG00000204525	putamen	1.32×10^{-15} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	putamen	5.13×10^{-16} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	EBV lymphocytes	6.62×10^{-27} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	EBV lymphocytes	2.69×10^{-16} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	EBV lymphocytes	9.14×10^{-18} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	ovary	5.49×10^{-23} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	ovary	2.74×10^{-17} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	ovary	5.36×10^{-11} (p-value)	GTEx Portal V6	15
HCG27	?	ENSG00000206344	sun exposed skin	8.59×10^{-9} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	sun exposed skin	5.17×10^{-72} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	sun exposed skin	1.35×10^{-7} (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	3.48×10^{-51} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	sun exposed skin	1.54×10^{-12} (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	sun exposed skin	2.79×10^{-8} (p-value)	GTEx Portal V6	14
PSORS1C1	?	ENSG00000204540	sun exposed skin	1.53×10^{-16} (p-value)	GTEx Portal V6	16
FLOT1	?	ENSG00000137312	sun exposed skin	3.51×10^{-6} (p-value)	GTEx Portal V6	10
CYP21A1P	?	ENSG00000204338	sun exposed skin	6.43×10^{-7} (p-value)	GTEx Portal V6	14
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	2.10×10^{-6} (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	unexposed skin	1.61×10^{-6} (p-value)	GTEx Portal V6	15
HLA-C	?	ENSG00000204525	unexposed skin	1.31×10^{-43} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	unexposed skin	7.21×10^{-31} (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	unexposed skin	1.05×10^{-9} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	unexposed skin	1.88×10^{-7} (p-value)	GTEx Portal V6	4
HLA-C	?	ENSG00000204525	aorta	3.92×10^{-49} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	aorta	4.29×10^{-19} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	aorta	4.19×10^{-35} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	aorta	2.91×10^{-8} (p-value)	GTEx Portal V6	11
PSORS1C1	?	ENSG00000204540	aorta	6.04×10^{-8} (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	aorta	1.44×10^{-6} (p-value)	GTEx Portal V6	10
MICB	?	ENSG00000204516	aorta	4.90×10^{-6} (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	left ventricle	2.74×10^{-41} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	1.66×10^{-35} (p-value)	GTEx Portal V6	16
HCG27	?	ENSG00000206344	left ventricle	1.89×10^{-6} (p-value)	GTEx Portal V6	10
MICA	?	ENSG00000204520	left ventricle	1.82×10^{-7} (p-value)	GTEx Portal V6	10
HLA-B	?	ENSG00000234745	left ventricle	3.15×10^{-10} (p-value)	GTEx Portal V6	15
PSORS1C1	?	ENSG00000204540	left ventricle	3.33×10^{-7} (p-value)	GTEx Portal V6	15

HCG27	?	ENSG00000206344	subcutaneous adipocytes	2.06×10^{-13} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	7.57×10^{-62} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	1.68×10^{-49} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	subcutaneous adipocytes	2.89×10^{-17} (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	subcutaneous adipocytes	8.99×10^{-8} (p-value)	GTEx Portal V6	11
CYP21A1P	?	ENSG00000204338	subcutaneous adipocytes	8.32×10^{-8} (p-value)	GTEx Portal V6	11
VARS2	?	ENSG00000137411	subcutaneous adipocytes	7.43×10^{-7} (p-value)	GTEx Portal V6	11
PSORS1C1	?	ENSG00000204540	subcutaneous adipocytes	1.13×10^{-15} (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	subcutaneous adipocytes	8.97×10^{-8} (p-value)	GTEx Portal V6	11
XXbac-BPG299F13.17	?	ENSG00000272501	subcutaneous adipocytes	6.48×10^{-6} (p-value)	GTEx Portal V6	5
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	3.86×10^{-8} (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	caudate basal ganglia	2.41×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	caudate basal ganglia	1.68×10^{-17} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	caudate basal ganglia	1.80×10^{-18} (p-value)	GTEx Portal V6	16
XXbac-BPG181B23.7	?	ENSG00000272221	caudate basal ganglia	1.86×10^{-7} (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	visceral adipocytes	4.81×10^{-35} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	visceral adipocytes	1.64×10^{-12} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	6.79×10^{-32} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	visceral adipocytes	3.36×10^{-6} (p-value)	GTEx Portal V6	1
PSORS1C1	?	ENSG00000204540	visceral adipocytes	3.31×10^{-10} (p-value)	GTEx Portal V6	16
HCG27	?	ENSG00000206344	stomach	5.60×10^{-8} (p-value)	GTEx Portal V6	14
PSORS1C2	?	ENSG00000204538	stomach	2.18×10^{-12} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	stomach	1.59×10^{-38} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	stomach	3.57×10^{-29} (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	stomach	7.74×10^{-14} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	stomach	6.50×10^{-10} (p-value)	GTEx Portal V6	15
HLA-C	?	ENSG00000204525	cortex	4.63×10^{-21} (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	cortex	3.42×10^{-18} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	cortex	9.09×10^{-9} (p-value)	GTEx Portal V6	11
HCG27	?	ENSG00000206344	tibial nerve	8.11×10^{-11} (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	tibial nerve	1.32×10^{-50} (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	tibial nerve	2.81×10^{-8} (p-value)	GTEx Portal V6	14
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	4.56×10^{-42} (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	tibial nerve	5.62×10^{-14} (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	tibial nerve	5.37×10^{-9} (p-value)	GTEx Portal V6	14
PSORS1C1	?	ENSG00000204540	tibial nerve	4.02×10^{-10} (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	tibial nerve	6.17×10^{-7} (p-value)	GTEx Portal V6	11

PSORS1C2	?	ENSG00000204538	tibial nerve	1.26×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	1.33×10-5 (p-value)	GTEx Portal V6	1
PSORS1C3	?	ENSG00000204528	esophagus mucosa	3.65×10-6 (p-value)	GTEx Portal V6	7
PSORS1C2	?	ENSG00000204538	esophagus mucosa	1.75×10-13 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	esophagus mucosa	5.36×10-50 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	esophagus mucosa	3.33×10-6 (p-value)	GTEx Portal V6	1
TCF19	?	ENSG00000137310	esophagus mucosa	1.35×10-6 (p-value)	GTEx Portal V6	12
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	1.80×10-39 (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	esophagus mucosa	5.73×10-7 (p-value)	GTEx Portal V6	12
PSORS1C1	?	ENSG00000204540	esophagus mucosa	7.80×10-16 (p-value)	GTEx Portal V6	16
HCG27	?	ENSG00000206344	esophagus mucosa	1.09×10-7 (p-value)	GTEx Portal V6	15
MICA	?	ENSG00000204520	esophagus mucosa	6.28×10-6 (p-value)	GTEx Portal V6	3
HLA-C	?	ENSG00000204525	anterior cingulate cortex	4.70×10-17 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	anterior cingulate cortex	5.96×10-11 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	pituitary	2.35×10-14 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	pituitary	1.06×10-14 (p-value)	GTEx Portal V6	16
WASF5P	?	ENSG00000231402	pituitary	3.26×10-7 (p-value)	GTEx Portal V6	10
HLA-C	?	ENSG00000204525	testis	6.15×10-28 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	testis	6.59×10-24 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	testis	1.24×10-26 (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	testis	9.54×10-8 (p-value)	GTEx Portal V6	15
HLA-C	?	ENSG00000204525	hypothalamus	5.02×10-17 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	hypothalamus	2.34×10-14 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	frontal cortex	1.91×10-19 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	frontal cortex	3.00×10-16 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	frontal cortex	2.36×10-7 (p-value)	GTEx Portal V6	11
HLA-C	?	ENSG00000204525	uterus	2.63×10-16 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	uterus	1.79×10-13 (p-value)	GTEx Portal V6	16
WASF5P	?	ENSG00000231402	pancreas	4.24×10-10 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	pancreas	4.82×10-36 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	pancreas	7.69×10-10 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	pancreas	6.15×10-24 (p-value)	GTEx Portal V6	16
DDX39B	?	ENSG00000198563	pancreas	2.13×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.13	?	ENSG00000256166	pancreas	3.94×10-7 (p-value)	GTEx Portal V6	10
HCG27	?	ENSG00000206344	muscularis mucosae	1.75×10-8 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	muscularis mucosae	1.68×10-7 (p-value)	GTEx Portal V6	11
HLA-C	?	ENSG00000204525	muscularis mucosae	9.42×10-54 (p-value)	GTEx Portal V6	16

XXbac-BPG299F13.17	?	ENSG00000272501	muscularis mucosae	1.02×10-10 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	muscularis mucosae	9.39×10-38 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	muscularis mucosae	5.76×10-17 (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	muscularis mucosae	1.45×10-8 (p-value)	GTEx Portal V6	11
PSORS1C1	?	ENSG00000204540	muscularis mucosae	2.49×10-10 (p-value)	GTEx Portal V6	13
CYP21A1P	?	ENSG00000204338	muscularis mucosae	1.18×10-6 (p-value)	GTEx Portal V6	10
XXbac-BPG181B23.7	?	ENSG00000272221	muscularis mucosae	3.79×10-6 (p-value)	GTEx Portal V6	4
MICA	?	ENSG00000204520	muscularis mucosae	3.42×10-6 (p-value)	GTEx Portal V6	3
HCG27	?	ENSG00000206344	lung	3.97×10-7 (p-value)	GTEx Portal V6	15
PSORS1C2	?	ENSG00000204538	lung	1.93×10-10 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	lung	2.54×10-50 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	lung	7.37×10-7 (p-value)	GTEx Portal V6	8
XXbac-BPG248L24.12	?	ENSG00000271581	lung	8.06×10-45 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	lung	5.60×10-14 (p-value)	GTEx Portal V6	16
CYP21A1P	?	ENSG00000204338	lung	3.52×10-8 (p-value)	GTEx Portal V6	12
PSORS1C1	?	ENSG00000204540	lung	6.21×10-10 (p-value)	GTEx Portal V6	12
MICA	?	ENSG00000204520	lung	3.49×10-7 (p-value)	GTEx Portal V6	15
XXbac-BPG181B23.7	?	ENSG00000272221	lung	5.58×10-7 (p-value)	GTEx Portal V6	6
HCG27	?	ENSG00000206344	tibial artery	5.55×10-9 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	tibial artery	1.06×10-79 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	tibial artery	3.62×10-13 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	4.17×10-48 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	tibial artery	3.17×10-17 (p-value)	GTEx Portal V6	16
CCHCR1	?	ENSG00000204536	tibial artery	5.49×10-13 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	tibial artery	6.30×10-11 (p-value)	GTEx Portal V6	16
XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	1.26×10-6 (p-value)	GTEx Portal V6	4
MICA	?	ENSG00000204520	tibial artery	3.59×10-6 (p-value)	GTEx Portal V6	4
BAG6	?	ENSG00000204463	tibial artery	1.98×10-7 (p-value)	GTEx Portal V6	5
CYP21A1P	?	ENSG00000204338	tibial artery	4.68×10-6 (p-value)	GTEx Portal V6	1
HCG27	?	ENSG00000206344	thyroid	1.62×10-11 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	thyroid	2.85×10-13 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	thyroid	3.56×10-57 (p-value)	GTEx Portal V6	16
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	4.48×10-10 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	3.40×10-50 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	thyroid	7.07×10-17 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	thyroid	9.83×10-15 (p-value)	GTEx Portal V6	16
MICA	?	ENSG00000204520	thyroid	1.53×10-10 (p-value)	GTEx Portal V6	15
CYP21A1P	?	ENSG00000204338	thyroid	1.12×10-6 (p-value)	GTEx Portal V6	11

XXbac-BPG299F13.17	?	ENSG00000272501	thyroid	6.54×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	prostate	1.78×10-10 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	prostate	1.97×10-11 (p-value)	GTEx Portal V6	16
HCG27	?	ENSG00000206344	transverse colon	2.24×10-7 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	transverse colon	1.43×10-8 (p-value)	GTEx Portal V6	12
HLA-C	?	ENSG00000204525	transverse colon	2.23×10-43 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	transverse colon	1.53×10-9 (p-value)	GTEx Portal V6	14
XXbac-BPG248L24.12	?	ENSG00000271581	transverse colon	1.07×10-30 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	transverse colon	5.66×10-11 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	transverse colon	9.90×10-9 (p-value)	GTEx Portal V6	9
XXbac-BPG181B23.7	?	ENSG00000272221	transverse colon	3.62×10-6 (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	transverse colon	1.22×10-6 (p-value)	GTEx Portal V6	3
HLA-C	?	ENSG00000204525	liver	6.43×10-25 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	liver	3.85×10-14 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	terminal ileum	6.70×10-16 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	terminal ileum	8.75×10-12 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	spleen	1.66×10-20 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	spleen	1.14×10-16 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	spleen	4.21×10-6 (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	spleen	9.44×10-7 (p-value)	GTEx Portal V6	4
HLA-C	?	ENSG00000204525	nucleus accumbens	2.12×10-17 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	nucleus accumbens	5.29×10-17 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	sigmoid colon	9.31×10-34 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	sigmoid colon	7.47×10-23 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	sigmoid colon	1.92×10-8 (p-value)	GTEx Portal V6	14
HLA-B	?	ENSG00000234745	sigmoid colon	4.75×10-7 (p-value)	GTEx Portal V6	12
PSORS1C1	?	ENSG00000204540	sigmoid colon	1.05×10-7 (p-value)	GTEx Portal V6	10
PSORS1C2	?	ENSG00000204538	sigmoid colon	1.94×10-6 (p-value)	GTEx Portal V6	6
HLA-C	?	ENSG00000204525	coronary artery	7.07×10-25 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	coronary artery	1.41×10-19 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	coronary artery	2.10×10-6 (p-value)	GTEx Portal V6	5
PSORS1C1	?	ENSG00000204540	coronary artery	1.09×10-6 (p-value)	GTEx Portal V6	10
HLA-C	?	ENSG00000204525	vagina	5.14×10-16 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	vagina	1.75×10-15 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	gastroesophageal junction	7.53×10-35 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	gastroesophageal junction	2.08×10-23 (p-value)	GTEx Portal V6	16
XXbac-BPG299F13.17	?	ENSG00000272501	gastroesophageal junction	1.14×10-6 (p-value)	GTEx Portal V6	11

PSORS1C1	?	ENSG00000204540	gastroesophageal junction	2.66×10-8 (p-value)	GTEx Portal V6	12
PSORS1C2	?	ENSG00000204538	gastroesophageal junction	8.80×10-7 (p-value)	GTEx Portal V6	11
HLA-B	?	ENSG00000234745	gastroesophageal junction	9.26×10-10 (p-value)	GTEx Portal V6	15
HLA-C	?	ENSG00000204525	adrenal gland	1.62×10-30 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	adrenal gland	7.51×10-18 (p-value)	GTEx Portal V6	16
PSORS1C1	?	ENSG00000204540	adrenal gland	2.51×10-8 (p-value)	GTEx Portal V6	16
PSORS1C2	?	ENSG00000204538	adrenal gland	5.60×10-7 (p-value)	GTEx Portal V6	13
TNXB	?	ENSG00000168477	adrenal gland	1.65×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	cerebellar hemisphere	1.11×10-14 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	cerebellar hemisphere	1.21×10-14 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	cerebellar hemisphere	3.42×10-6 (p-value)	GTEx Portal V6	2
WASF5P	?	ENSG00000231402	cerebellum	1.50×10-10 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	cerebellum	3.49×10-22 (p-value)	GTEx Portal V6	16
HLA-B	?	ENSG00000234745	cerebellum	3.72×10-9 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	cerebellum	4.97×10-21 (p-value)	GTEx Portal V6	16
HLA-C	?	ENSG00000204525	hippocampus	2.41×10-18 (p-value)	GTEx Portal V6	16
XXbac-BPG248L24.12	?	ENSG00000271581	hippocampus	6.93×10-14 (p-value)	GTEx Portal V6	16
VARS2	ENST00000321897	ILMN_1737585	skin	1.65×10-8 (p-value)	MuTHER consortium	6
VARS2	ENST00000625423		adipocyte	4.72×10-5 (p-value)	MuTHER consortium	6
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
ATP6V1G2	ENST00000481998	ILMN_1654541	blood	2.93×10-6 (p-value)	MuTHER consortium	6
ATP6V1G2	ENST00000303892					
ATP6V1G2	ENST00000483170					
ATP6V1G2	ENST00000376151					
HCG22	ENST00000570223	ILMN_1667229	blood	2.34×10-5 (p-value)	MuTHER consortium	4
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
MICB	ENST00000538442	ILMN_1708006	skin	2.04×10-5 (p-value)	MuTHER consortium	3
MICB	ENST00000399150					
MICB	ENST00000252229					
HCG27	ENST00000383331	ILMN_1746436	skin	1.35×10-8 (p-value)	MuTHER consortium	6
HCG27	ENST00000415276		adipocyte	3.10×10-15 (p-value)	MuTHER consortium	6
PSORS1C1	ENST00000481450	ILMN_1799669	skin	1.34×10-5 (p-value)	MuTHER consortium	3
PSORS1C1	ENST00000552747					
PSORS1C1	ENST00000547221					
PSORS1C1	ENST00000259881					
PSORS1C1	ENST00000479581					

?	?	ILMN_1893395	blood	6.11×10-7 (p-value)	Westra et al.	5
VARS2	ENST00000321897	ILMN_1737585	monocyte	3.11×10-12 (p-value)	Fairfax et al.	1
VARS2	ENST00000625423		b-cell	2.34×10-8 (p-value)	Fairfax et al.	1
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
CCHCR1	?	ENSG00000204536	blood	3.04×10-2 (q-value)	SeeQTL DB (HapMap)	2
HLA-B	?	ENSG00000234745	blood	3.70×10-2 (q-value)	SeeQTL DB (HapMap)	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
CSF3R	ENST00000464465	ILMN_2371280	chr1	blood	7.65×10-21 (p-value)	Westra et al.	1
CSF3R	ENST00000373104						
CSF3R	ENST00000487540						
CSF3R	ENST00000373106						
CSF3R	ENST00000331941						
CSF3R	ENST00000484762						
CSF3R	ENST00000480825						
CSF3R	ENST00000361632						
CSF3R	ENST00000373103						
TSSC4	ENST00000451491	ILMN_1802753	chr11	blood	1.14×10-8 (p-value)	Westra et al.	1
TSSC4	ENST00000437110						
TSSC4	ENST00000496468						
TSSC4	ENST00000435795						
TSSC4	ENST00000440813						
TSSC4	ENST00000485682						
TSSC4	ENST00000467308						
TSSC4	ENST00000380996						
TSSC4	ENST00000333256						
TMEM154	ENST00000304385	ILMN_2088124	chr4	blood	2.03×10-78 (p-value)	Westra et al.	1
CD248	ENST00000311330	ILMN_1726589	chr11	blood	2.09×10-9 (p-value)	Westra et al.	1
XRCC6	ENST00000405878	ILMN_2166506	chr22	blood	4.99×10-19 (p-value)	Westra et al.	1
XRCC6	ENST00000359308						
XRCC6	ENST00000405506						
XRCC6	ENST00000360079						
XRCC6	ENST00000402580						
XRCC6	ENST00000428575						

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487828 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K27me3, PolII, TAF1, H3K4me2, SP1, H3K4me3
		HSMMtube	H3K27me3, H2AZ, H3K36me3, DNase1, H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H3K4me3, H3K27me3, DNase1
		liver (HepG2)	H3K4me2, H3K4me3, H3K36me3, H3K27me3

blood (GM12878)	H3K79me2, H3K4me3, NFKB, H3K27ac, H4K20me1, Cfos, H3K4me2, H3K9ac, H3K36me3, IRF4, DNase1, TAF1, PU1, PolII, H2AZ
lung (IMR90)	DNase1, H3K27me3, H3K36me3, H3K4me3, CTCF
nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K27me3, DNase1
skin (NHEK)	CTCF, H3K9ac, H3K4me2, H3K4me3, H3K36me3, H3K27me3
NHLF	H3K4me3, H3K9ac, H3K27ac, H3K36me3
Osteobl	H3K27me3, H3K36me3, H3K4me2, H3K4me3
blood (DND-41)	H3K36me3, H3K27me3, H3K9ac
breast (HMEC)	CTCF, DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
cervix (HeLa-S3)	PolII, H3K9ac, H3K4me2, H3K27ac, H3K4me3, DNase1
monocytes (Monocytes-CD14+)	H3K36me3, H3K4me3, H4K20me1, H3K9ac, DNase1, H3K4me2, H3K27ac
endothelium (HUVEC)	H3K4me3, PolII, H3K27me3, DNase1
A549	H3K27me3, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HLA-B	upstream gene variant	1010	ENST00000474381	?	?	5
HLA-B	upstream gene variant	2839	ENST00000497377	?	?	2
HLA-B	upstream gene variant	3109	ENST00000481849	?	?	2
HLA-B	upstream gene variant	2113	ENST00000463574	?	?	5
HLA-B	upstream gene variant	736	ENST00000498007	?	?	5
HLA-B	upstream gene variant	728	ENST00000412585	NM_005514.6	ENSP00000399168	5
HLA-B	upstream gene variant	727	ENST00000434333	?	ENSP00000405931	5
HLA-C	upstream gene variant	757	ENST00000620806	?	ENSP00000480331	5
HLA-C	upstream gene variant	2226	ENST00000376228	NM_002117.5	ENSP00000365402	1
HLA-C	upstream gene variant	2262	ENST00000487245	?	?	1
HLA-C	upstream gene variant	4646	ENST00000466892	?	?	1
HLA-C	upstream gene variant	2226	ENST00000383329	?	ENSP00000372819	1
HLA-C	upstream gene variant	3890	ENST00000470363	?	?	1
HLA-C	upstream gene variant	2222	ENST00000484378	?	?	1
HLA-C	upstream gene variant	2182	ENST00000495835	?	?	1
HLA-C	upstream gene variant	2207	ENST00000376237	?	ENSP00000365412	1
HLA-C	upstream gene variant	2243	ENST00000415537	?	ENSP00000400410	1
MIR6891	upstream gene variant	2599	ENST00000618788	?	?	4
RPL3P2	downstream gene variant	3100	ENST00000413027	?	?	1
USP8P1	upstream gene variant	1260	ENST00000494673	?	?	1
WASF5P	downstream gene variant, upstream gene variant	770	ENST00000428639	?	?	2
XXbac-BPG248L24.10	downstream gene variant	3331	ENST00000421191	?	?	2
XXbac-BPG248L24.12	downstream gene variant	278	ENST00000603274	?	?	5
XXbac-BPG248L24.13	downstream gene variant, upstream gene variant	2148	ENST00000539514	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HLA-C	ENST00000620806	?	ENSP00000480331	11
XXbac-BPG248L24.13	ENST00000539514	?	?	6