SNiPAcard

Block annotations

Block info

genomic range	chr6:31,250,801-31,271,716
block size	20,916 bp
variant count	127 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.758 [-4.394 – 2.486]	**gene(s) hit or close-by**	HLA-C, RPL3P2, USP8P1, WASF5P, XXbac-BPG248L24.10, XXbac-BPG248L24.13
phastCons	μ = 0.063 [0 – 0.998]	**eQTL gene(s)**	C4B, CCHCR1, CD248, DDR1, GPBAR1, HCG22, HCG27, HLA-B, HLA-C, MICA, MICB, PLOD3, POU5F1, PSORS1C1, PSORS1C2, PSORS1C3, TCF19, TMEM154, TRAPPC1, WASF5P, XXbac-BPG248L24.10, XXbac-BPG248L24.12, XXbac-BPG299F13.17
GERP++	μ = -0.144 [-1.75 – 0.718]	**potentially regulated gene(s)**	CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, CCHCR1, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, DDAH2, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, SFTA2, SFTA2, SFTA2, SFTA2, SFTA2, SFTA2, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P, WASF5P
CADD score	μ = 2.579 [0.016 – 17.05]	**disease gene(s)**	HLA-B, C4B, PLOD3

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Protein quantitative trait loci	$<4.00\times10^{-6}$	GWAS Catalog	18464913	1

Disease gene annotation

gene	trait	source DB	source entry/link
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147
PLOD3	Connective tissue disorder due to lysyl hydroxylase-3 deficiency	OrphaNet	OrphaNet:300284
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
PLOD3	BONE FRAGILITY WITH CONTRACTURES, ARTERIAL RUPTURE, AND [...]	OMIM	MIM:612394
PLOD3	LYSYL HYDROXYLASE 3 DEFICIENCY	DECIPHER	MIM:612394

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HCG22	ENST00000570223	ILMN_1667229	blood	5.04×10^{-5} (p-value)	MuTHER consortium	1
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
HCG27	ENST00000383331	ILMN_1746436	adipocyte	6.00×10^{-27} (p-value)	MuTHER consortium	21

HCG27	ENST00000415276		skin	1.40×10-13 (p-value)	MuTHER consortium	21
DDR1	ENST00000376567	ILMN_1812262	blood	3.85×10-7 (p-value)	MuTHER consortium	20
DDR1	ENST00000376570					
DDR1	ENST00000418800					
DDR1	ENST00000376569					
DDR1	ENST00000452441					
DDR1	ENST00000324771					
DDR1	ENST00000482873					
DDR1	ENST00000376568					
DDR1	ENST00000446312					
DDR1	ENST00000454612					
HCG27	?	ENSG00000206344	muscularis mucosae	2.64×10-14 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	muscularis mucosae	5.82×10-6 (p-value)	GTEx Portal V6	22
HCG27	?	ENSG00000206344	lung	4.59×10-9 (p-value)	GTEx Portal V6	104
PSORS1C3	?	ENSG00000204528	lung	1.12×10-7 (p-value)	GTEx Portal V6	95
HLA-C	?	ENSG00000204525	lung	2.82×10-7 (p-value)	GTEx Portal V6	99
XXbac-BPG248L24.12	?	ENSG00000271581	lung	2.21×10-6 (p-value)	GTEx Portal V6	1
PSORS1C3	?	ENSG00000204528	atrial appendage	1.61×10-9 (p-value)	GTEx Portal V6	100
HLA-C	?	ENSG00000204525	atrial appendage	4.10×10-7 (p-value)	GTEx Portal V6	86
HCG27	?	ENSG00000206344	atrial appendage	1.72×10-6 (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	transformed fibroblasts	4.37×10-11 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	transformed fibroblasts	4.61×10-13 (p-value)	GTEx Portal V6	104
C4B	?	ENSG00000224389	transformed fibroblasts	2.95×10-6 (p-value)	GTEx Portal V6	18
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	2.07×10-7 (p-value)	GTEx Portal V6	2
PSORS1C3	?	ENSG00000204528	blood	4.75×10-11 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	blood	4.64×10-15 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	blood	6.84×10-10 (p-value)	GTEx Portal V6	104
XXbac-BPG299F13.17	?	ENSG00000272501	blood	3.88×10-13 (p-value)	GTEx Portal V6	104
POU5F1	?	ENSG00000204531	blood	1.09×10-9 (p-value)	GTEx Portal V6	104
HCG27	?	ENSG00000206344	blood	2.38×10-7 (p-value)	GTEx Portal V6	13
HCG27	?	ENSG00000206344	tibial artery	7.32×10-13 (p-value)	GTEx Portal V6	104
PSORS1C3	?	ENSG00000204528	tibial artery	4.66×10-8 (p-value)	GTEx Portal V6	100
HLA-C	?	ENSG00000204525	tibial artery	9.94×10-12 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	7.57×10-7 (p-value)	GTEx Portal V6	52
HLA-B	?	ENSG00000234745	tibial artery	1.09×10-5 (p-value)	GTEx Portal V6	5
HCG27	?	ENSG00000206344	thyroid	1.80×10-15 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.10	?	ENSG00000229836	thyroid	1.19×10-12 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	thyroid	8.55×10-10 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	2.30×10-8 (p-value)	GTEx Portal V6	90
HLA-B	?	ENSG00000234745	thyroid	7.33×10-8 (p-value)	GTEx Portal V6	96

WASF5P	?	ENSG00000231402	thyroid	1.99×10-6 (p-value)	GTEx Portal V6	96
PSORS1C3	?	ENSG00000204528	thyroid	1.58×10-6 (p-value)	GTEx Portal V6	62
HCG27	?	ENSG00000206344	skeletal muscle	5.07×10-11 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	skeletal muscle	3.36×10-13 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	1.36×10-9 (p-value)	GTEx Portal V6	102
HCG27	?	ENSG00000206344	transverse colon	1.86×10-11 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	transverse colon	1.19×10-6 (p-value)	GTEx Portal V6	27
XXbac-BPG248L24.12	?	ENSG00000271581	transverse colon	3.36×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	liver	1.46×10-7 (p-value)	GTEx Portal V6	81
HCG27	?	ENSG00000206344	sun exposed skin	9.23×10-11 (p-value)	GTEx Portal V6	104
PSORS1C3	?	ENSG00000204528	sun exposed skin	1.22×10-9 (p-value)	GTEx Portal V6	103
HLA-C	?	ENSG00000204525	sun exposed skin	1.05×10-7 (p-value)	GTEx Portal V6	96
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	2.06×10-8 (p-value)	GTEx Portal V6	23
POU5F1	?	ENSG00000204531	sun exposed skin	6.68×10-11 (p-value)	GTEx Portal V6	103
PSORS1C3	?	ENSG00000204528	unexposed skin	3.01×10-7 (p-value)	GTEx Portal V6	88
HLA-C	?	ENSG00000204525	unexposed skin	7.04×10-7 (p-value)	GTEx Portal V6	94
POU5F1	?	ENSG00000204531	unexposed skin	5.17×10-8 (p-value)	GTEx Portal V6	102
HCG27	?	ENSG00000206344	unexposed skin	1.41×10-6 (p-value)	GTEx Portal V6	9
PSORS1C3	?	ENSG00000204528	left ventricle	1.05×10-6 (p-value)	GTEx Portal V6	62
HCG27	?	ENSG00000206344	aorta	6.79×10-7 (p-value)	GTEx Portal V6	87
HLA-C	?	ENSG00000204525	aorta	7.00×10-12 (p-value)	GTEx Portal V6	104
HCG27	?	ENSG00000206344	subcutaneous adipocytes	6.73×10-17 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	4.19×10-10 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	3.75×10-8 (p-value)	GTEx Portal V6	11
HCG27	?	ENSG00000206344	stomach	5.12×10-8 (p-value)	GTEx Portal V6	104
PSORS1C3	?	ENSG00000204528	stomach	3.67×10-7 (p-value)	GTEx Portal V6	93
HCG27	?	ENSG00000206344	visceral adipocytes	1.20×10-7 (p-value)	GTEx Portal V6	101
HLA-C	?	ENSG00000204525	visceral adipocytes	3.81×10-7 (p-value)	GTEx Portal V6	83
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	3.09×10-7 (p-value)	GTEx Portal V6	84
PSORS1C3	?	ENSG00000204528	spleen	7.06×10-7 (p-value)	GTEx Portal V6	86
HCG27	?	ENSG00000206344	nucleus accumbens	9.32×10-8 (p-value)	GTEx Portal V6	88
HCG27	?	ENSG00000206344	sigmoid colon	1.10×10-7 (p-value)	GTEx Portal V6	86
HLA-C	?	ENSG00000204525	sigmoid colon	3.69×10-7 (p-value)	GTEx Portal V6	70
HCG27	?	ENSG00000206344	tibial nerve	3.74×10-18 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	tibial nerve	1.16×10-10 (p-value)	GTEx Portal V6	104
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	8.13×10-6 (p-value)	GTEx Portal V6	1
HLA-B	?	ENSG00000234745	tibial nerve	6.97×10-6 (p-value)	GTEx Portal V6	9

HLA-B	?	ENSG00000234745	tibial nerve	6.97×10-6 (p-value)	GTEx Portal V6	9
PSORS1C3	?	ENSG00000204528	esophagus mucosa	8.85×10-10 (p-value)	GTEx Portal V6	103
PSORS1C2	?	ENSG00000204538	esophagus mucosa	3.96×10-8 (p-value)	GTEx Portal V6	104
HLA-C	?	ENSG00000204525	esophagus mucosa	2.95×10-11 (p-value)	GTEx Portal V6	104
TCF19	?	ENSG00000137310	esophagus mucosa	5.99×10-11 (p-value)	GTEx Portal V6	104
CCHCR1	?	ENSG00000204536	esophagus mucosa	7.87×10-7 (p-value)	GTEx Portal V6	93
POU5F1	?	ENSG00000204531	esophagus mucosa	5.23×10-12 (p-value)	GTEx Portal V6	103
XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	1.20×10-6 (p-value)	GTEx Portal V6	3
?	?	ILMN_1883997	blood	4.92×10-6 (p-value)	Westra et al.	1
MICB	ENST00000538442	ILMN_1708006	blood	9.19×10-7 (p-value)	Westra et al.	13
MICB	ENST00000399150					
MICB	ENST00000252229					
HCG27	?	ENSG00000206344	adrenal gland	2.17×10-7 (p-value)	GTEx Portal V6	38
HCG27	?	ENSG00000206344	testis	2.07×10-7 (p-value)	GTEx Portal V6	87
HLA-C	?	ENSG00000204525	testis	3.85×10-8 (p-value)	GTEx Portal V6	96
PSORS1C1	?	ENSG00000204540	testis	3.06×10-6 (p-value)	GTEx Portal V6	4
HCG27	?	ENSG00000206344	pancreas	2.72×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	EBV lymphocytes	2.10×10-6 (p-value)	GTEx Portal V6	1
CCHCR1	?	ENSG00000204536	blood	1.78×10-2 (q-value)	SeeQTL DB (HapMap)	8
HLA-C	ENST00000470363	ILMN_2150787	monocyte	2.06×10-20 (p-value)	Zeller et al.	4
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	ENST00000466892					
?	?	ILMN_1721113	monocyte	3.03×10-30 (p-value)	Zeller et al.	4
TCF19	ENST00000496421	223274_at	blood	3.20×10-8 (p-value)	Dixon et al.	2
TCF19	ENST00000376257					
TCF19	ENST00000376255					
HLA-C	?	ENSG00000204525	gastroesophageal junction	3.29×10-6 (p-value)	GTEx Portal V6	2
HCG27	?	ENSG00000206344	caudate basal ganglia	1.53×10-6 (p-value)	GTEx Portal V6	4
PSORS1C3	ENST00000412143	ILMN_3309634	b-cell	2.61×10-8 (p-value)	Fairfax et al.	6
MICA	ENST00000449934	ILMN_3241692	monocyte	9.22×10-5 (p-value)	Fairfax et al.	5
MICA	ENST00000616296					
MICA	ENST00000421350					
HCG27	?	ENSG00000206344	breast	2.13×10-6 (p-value)	GTEx Portal V6	52
XXbac-BPG299F13.17	?	ENSG00000272501	frontal cortex	7.70×10-7 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
PLOD3	ENST00000223127	ILMN_1774836	chr7	blood	1.47×10-18 (p-value)	Westra et al.	1

PLOD3	ENST00000454310						
TMEM154	ENST00000304385	ILMN_2088124	chr4	blood	2.87×10^{-25} (p-value)	Westra et al.	1
CD248	ENST00000311330	ILMN_1726589	chr11	blood	1.06×10^{-8} (p-value)	Westra et al.	1
GPBAR1	ENST00000519574	ILMN_2316386	chr2	blood	2.65×10^{-8} (p-value)	Westra et al.	1
GPBAR1	ENST00000479077						
GPBAR1	ENST00000521462						
GPBAR1	ENST00000522678						
TRAPPC1	ENST00000303731	ILMN_1716913	chr17	blood	3.28×10^{-7} (p-value)	Westra et al.	1
TRAPPC1	ENST00000575639						
TRAPPC1	ENST00000571739						
TRAPPC1	ENST00000571947						
TRAPPC1	ENST00000572656						
TRAPPC1	ENST00000573989						
TRAPPC1	ENST00000540486						

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444768	1	ENCP00000047982	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
		ENCP00000048000	WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P WASF5P
		ENCP00000047981	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
ENCE00000444720	2	ENCP00000047970	SFTA2 SFTA2 SFTA2 SFTA2 SFTA2 SFTA2
ENCE00000444721	6	ENCP00000047970	SFTA2 SFTA2 SFTA2 SFTA2 SFTA2 SFTA2
ENCE00000445080	6	ENCP00000048049	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000048087	DDAH2 DDAH2 DDAH2 DDAH2

				DDAH2 DDAH2 DDAH2		

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RPL3P2	downstream gene variant	1505	ENST00000413027	?	?	17
USP8P1	downstream gene variant	4270	ENST00000494673	?	?	9
WASF5P	downstream gene variant, upstream gene variant	206	ENST00000428639	?	?	24
XXbac-BPG248L24.10	downstream gene variant	3876	ENST00000421191	?	?	3
XXbac-BPG248L24.13	downstream gene variant, upstream gene variant	17	ENST00000539514	?	?	16

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HLA-C	ENST00000620806	?	ENSP00000480331	127
XXbac-BPG248L24.13	ENST00000539514	?	?	90

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
WASF5P	ENST00000428639	?	1
XXbac-BPG248L24.13	ENST00000539514	?	5