Locus 447 (rs1634718)

SNiPAcard

Block annotations

Block info

genomic range	chr6:30,936,534-30,982,008
block size	45,475 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.217 [-2.088 – 2.249]	gene(s) hit or close-by	MUC21, MUC22
phastCons	μ = 0.021 [0 – 0.116]	eQTL gene(s)	C4A, CCHCR1, CYP21A1P, FLOT1, HCG22, HCP5, HLA-C, LINC00243, MICB, MUC22, TRIM31, VARS2
GERP++	μ = -0.341 [-8.65 – 1.76]	potentially regulated gene(s)	ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, CDSN, CDSN, CDSN, CDSN, CDSN, CDSN, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, GNL1, GNL1, GNL1, GNL1, GNL1, GNL1, GNL1, GNL1, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG22, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, PPP1R10, PPP1R10, PPP1R10, PPP1R10, PPP1R10, PPP1R10, PPP1R10, PRR3, PRR3, PRR3, PRR3, PRR3, PRR3, PRR3, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG118E17.9, XXbac-BPG248L24.13, XXbac-BPG299F13.16
CADD score	μ = 5.289 [1.236 – 12.86]	disease gene(s)	C4A, VARS2, , CDSN

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Lupus erythematosus, systemic	2.64×10^{-8}	dbGaP	pha002867	dbGaP	2

Disease gene annotation

gene	trait	source DB	source entry/link	
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380	OMIM
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790	OMIM
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
CDSN	Hypotrichosis simplex of the scalp	OrphaNet	OrphaNet:90368	orphanet
CDSN	Generalized peeling skin syndrome type B	OrphaNet	OrphaNet:263553	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TRIM31	?	ENSG00000204616	lung	3.73×10^{-6} (p-value)	GTEx Portal V6	3
C4A	?	ENSG00000244731	lung	1.14×10^{-7} (p-value)	GTEx Portal V6	9

HCG22	?	ENSG00000228789	coronary artery	1.07×10^{-6} (p-value)	GTEx Portal V6	6
LINC00243	?	ENSG00000214894	blood	2.13×10^{-9} (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	blood	2.49×10^{-7} (p-value)	GTEx Portal V6	4
MUC22	?	ENSG00000261272	esophagus mucosa	7.09×10^{-9} (p-value)	GTEx Portal V6	15
MICB	?	ENSG00000204516	esophagus mucosa	8.59×10^{-7} (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	skeletal muscle	7.95×10^{-8} (p-value)	GTEx Portal V6	9
C4A	?	ENSG00000244731	skeletal muscle	2.84×10^{-7} (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	cerebellum	6.88×10^{-8} (p-value)	GTEx Portal V6	15
CCHCR1	?	ENSG00000204536	transverse colon	4.24×10^{-8} (p-value)	GTEx Portal V6	9
CCHCR1	?	ENSG00000204536	subcutaneous adipocytes	1.68×10^{-6} (p-value)	GTEx Portal V6	7
HCG22	?	ENSG00000228789	subcutaneous adipocytes	2.19×10^{-6} (p-value)	GTEx Portal V6	5
HCG22	ENST00000570223	ILMN_1667229	blood	3.23×10^{-18} (p-value)	MuTHER consortium	7
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
MICB	ENST00000538442	ILMN_1708006	skin	1.20×10^{-7} (p-value)	MuTHER consortium	7
MICB	ENST00000399150					
MICB	ENST00000252229					
VARS2	ENST00000321897	ILMN_1737585	skin	2.72×10^{-5} (p-value)	MuTHER consortium	1
VARS2	ENST00000625423					
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
HCP5	ENST00000414046	ILMN_1803945	skin	3.52×10^{-5} (p-value)	MuTHER consortium	2
HCP5	ENST00000541196					
FLOT1	?	ENSG00000137312	thyroid	8.04×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	thyroid	4.85×10^{-6} (p-value)	GTEx Portal V6	1
?	?	ILMN_1679389	b-cell	3.75×10^{-4} (p-value)	Fairfax et al.	3
C4A	?	ENSG00000244731	atrial appendage	2.58×10^{-7} (p-value)	GTEx Portal V6	5
C4A	?	ENSG00000244731	tibial nerve	3.53×10^{-7} (p-value)	GTEx Portal V6	9
C4A	?	ENSG00000244731	putamen	2.07×10^{-7} (p-value)	GTEx Portal V6	7
C4A	?	ENSG00000244731	left ventricle	3.19×10^{-7} (p-value)	GTEx Portal V6	9
C4A	?	ENSG00000244731	muscularis mucosae	1.19×10^{-8} (p-value)	GTEx Portal V6	8
CYP21A1P	?	ENSG00000204338	muscularis mucosae	2.21×10^{-7} (p-value)	GTEx Portal V6	2
HLA-C	ENST00000470363	216526_x_at	blood	6.20×10^{-8} (p-value)	Dixon et al.	2
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					

HLA-C	ENST00000466892					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
?	?	ILMN_1893395	monocyte	8.28×10-18 (p-value)	Zeller et al.	2
?	?	ILMN_1883997	monocyte	7.04×10-18 (p-value)	Zeller et al.	2
C4A	?	ENSG00000244731	transformed fibroblasts	3.34×10-6 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	sun exposed skin	1.74×10-7 (p-value)	GTEx Portal V6	2
C4A	?	ENSG00000244731	unexposed skin	1.13×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	EBV lymphocytes	3.03×10-6 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	aorta	2.61×10-6 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	221491_x_at	chr6	blood	6.20×10-8 (p-value)	Dixon et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444432	1	ENCP00000047915	PPP1R10 PPP1R10 PPP1R10 PPP1R10 PPP1R10 PPP1R10 PPP1R10
		ENCP00000047967	VARS2 VARS2 VARS2 VARS2 VARS2 VARS2
		ENCP00000047911	PRR3 PRR3 PRR3 GNL1 GNL1 GNL1 PRR3 GNL1 PRR3 GNL1 GNL1 GNL1 PRR3 PRR3 GNL1
		ENCP00000047978	CDSN CDSN CDSN CDSN CDSN CDSN
		ENCP00000047992	HCG27 HCG27 HCG27

			HCG27 HCG27 HCG27 HCG27
		ENCP00000047974	XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9 XXbac-BPG118E17.9
		ENCP00000047923	ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1
		ENCP00000047975	HCG22 HCG22 HCG22 HCG22 HCG22 HCG22 HCG22 HCG22
		ENCP00000047995	XXbac-BPG299F13.16
		ENCP00000048019	MICB MICB MICB MICB MICB MICB MICB MICB
ENCE00000444471	1	ENCP00000048001	XXbac-BPG248L24.13
		ENCP00000047921	ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1
		ENCP00000047949	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
		ENCP00000047941	FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001703009 (open chromatin region)	2	blood (K562)	H3K27me3
ENSR00001703011 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	DNase1, H3K27me3
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		breast (HMEC)	CTCF, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	CTCF, H3K27me3
		lung (IMR90)	H3K4me2
		nervous (NH-A)	H3K4me2, DNase1
ENSR00001494536 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, H3K27me3, Rad21
		HSMMtube	DNase1, H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	DNase1
		liver (HepG2)	CTCF, H3K27me3
		blood (GM12878)	H3K27me3
		lung (IMR90)	H3K4me2, H3K27me3
		nervous (NH-A)	H3K4me2, DNase1
		NHLF	DNase1
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MUC21	upstream gene variant	475	ENST00000376296	NM_001010909.2	ENSP00000365473	2
MUC21	upstream gene variant, downstream gene variant	475	ENST00000486149	?	ENSP00000457640	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
MUC21	ENST00000376296	NM_001010909.2	ENSP00000365473	T	acA/acG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MUC22	ENST00000561890	NM_001198815.1	ENSP00000455906	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MUC21	ENST00000376296	NM_001010909.2	ENSP00000365473	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MUC21	ENST00000486149	?	ENSP00000457640	1