Locus 338 (rs11753208)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,005,432-31,010,159
block size	4,728 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.057 [-0.247 – 0.361]	gene(s) hit or close-by	MUC22
phastCons	μ = 0.002 [0 – 0.003]	eQTL gene(s)	CCHCR1, HCG22, HCG27, MICA, PSORS1C1, PSORS1C2, VARS2, XXbac-BPG181B23.7, XXbac-BPG299F13.17
GERP++	μ = 0.700 [-1.26 – 2.66]	potentially regulated gene(s)	–
CADD score	μ = 5.436 [1.051 – 9.822]	disease gene(s)	VARS2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HCG22	ENST00000570223	ILMN_1667229	blood	6.50×10-6 (p-value)	MuTHER consortium	1
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
MICA	?	ENSG00000204520	muscularis mucosae	3.09×10-6 (p-value)	GTEx Portal V6	1
PSORS1C2	?	ENSG00000204538	muscularis mucosae	3.51×10-8 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	muscularis mucosae	8.34×10-7 (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	lung	7.42×10-8 (p-value)	GTEx Portal V6	2
PSORS1C2	?	ENSG00000204538	lung	3.42×10-6 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	lung	1.13×10-7 (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	transformed fibroblasts	8.53×10-7 (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	blood	3.14×10-6 (p-value)	GTEx Portal V6	1
MICA	?	ENSG00000204520	tibial artery	4.53×10-6 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	tibial artery	1.44×10-7 (p-value)	GTEx Portal V6	2
PSORS1C2	?	ENSG00000204538	thyroid	5.72×10-8 (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	thyroid	4.27×10-10 (p-value)	GTEx Portal V6	2

XXbac-BPG299F13.17	?	ENSG00000272501	skeletal muscle	2.90×10^{-7} (p-value)	GTEx Portal V6	2
CCHCR1	?	ENSG00000204536	sun exposed skin	8.04×10^{-7} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	sun exposed skin	1.67×10^{-6} (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	sun exposed skin	1.33×10^{-7} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	aorta	2.63×10^{-8} (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	aorta	4.63×10^{-6} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	subcutaneous adipocytes	3.37×10^{-6} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	subcutaneous adipocytes	1.38×10^{-6} (p-value)	GTEx Portal V6	2
HCG27	ENST00000383331	ILMN_1746436	monocyte	1.68×10^{-22} (p-value)	Zeller et al.	1
HCG27	ENST00000415276					
MICA	?	ENSG00000204520	sigmoid colon	8.14×10^{-7} (p-value)	GTEx Portal V6	2
PSORS1C1	?	ENSG00000204540	sigmoid colon	1.35×10^{-6} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	tibial nerve	4.59×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	tibial nerve	2.84×10^{-8} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	tibial nerve	9.15×10^{-6} (p-value)	GTEx Portal V6	2
MICA	?	ENSG00000204520	esophagus mucosa	2.91×10^{-9} (p-value)	GTEx Portal V6	2
XXbac-BPG299F13.17	?	ENSG00000272501	esophagus mucosa	1.50×10^{-6} (p-value)	GTEx Portal V6	2
HCG22	?	ENSG00000228789	esophagus mucosa	1.60×10^{-9} (p-value)	GTEx Portal V6	2
VARS2	ENST00000321897	ILMN_1737585	blood	4.68×10^{-5} (p-value)	Westra et al.	1
VARS2	ENST00000625423					
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494543 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1, H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me1, H2AZ, H3K27ac, H3K4me2, DNase1
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MUC22	downstream gene variant	2253	ENST00000561890	NM_001198815.1	ENSP00000455906	1

Locus 338 (sentinel SNP rs11753208 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.