Locus 412 (rs12788387)

SNiPAcard

Block annotations

Block info

genomic range	chr11:57,851,041-57,913,286
block size	62,246 bp
variant count	46 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.103 [-2.019 – 1.61]	**gene(s) hit or close-by**	OR9I1, OR9I2P, OR9I3P, OR9L1P, OR9Q1
phastCons	μ = 0.090 [0 – 1]	**eQTL gene(s)**	MED19, RN7SL605P, TIMM10
GERP++	μ = -0.303 [-6.36 – 2.96]	**potentially regulated gene(s)**	LPXN, OR5B21, ZDHHC5
CADD score	μ = 4.517 [0.27 – 18.8]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MED19	?	ENSG00000156603	skeletal muscle	4.35×10^{-7} (p-value)	GTEx Portal V6	40
MED19	?	ENSG00000156603	atrial appendage	1.20×10^{-6} (p-value)	GTEx Portal V6	46
MED19	?	ENSG00000156603	transverse colon	3.46×10^{-6} (p-value)	GTEx Portal V6	5
MED19	?	ENSG00000156603	sun exposed skin	2.90×10^{-8} (p-value)	GTEx Portal V6	46
MED19	?	ENSG00000156603	esophagus mucosa	1.29×10^{-9} (p-value)	GTEx Portal V6	46
MED19	?	ENSG00000156603	subcutaneous adipocytes	5.37×10^{-6} (p-value)	GTEx Portal V6	12
MED19	?	ENSG00000156603	thyroid	1.93×10^{-8} (p-value)	GTEx Portal V6	45
TIMM10	ENST00000525587	ILMN_1765332	skin	6.29×10^{-6} (p-value)	MuTHER consortium	18
RN7SL605P	ENST00000577229		adipocyte	7.42×10^{-8} (p-value)	MuTHER consortium	18
TIMM10	ENST00000257245		blood	3.39×10^{-5} (p-value)	MuTHER consortium	18
TIMM10	ENST00000525587	ILMN_1765332	monocyte	7.68×10^{-18} (p-value)	Zeller et al.	5
RN7SL605P	ENST00000577229					
TIMM10	ENST00000257245					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000106527	1	ENCP00000012323	LPXN
		ENCP00000012278	ZDHHC5
		ENCP00000012322	OR5B21

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
OR9I1	downstream gene variant, upstream gene variant	1043	ENST00000302610	NM_001005211.1	ENSP00000302606	6
OR9I2P	upstream gene variant, downstream gene variant	37	ENST00000427941	?	?	12

OR9I3P	downstream gene variant, upstream gene variant	998	ENST00000608484	?	?	4
OR9L1P	upstream gene variant	4966	ENST00000529493	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
OR9Q1	ENST00000335397	NM_001005212.3	ENSP00000334934	46

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
OR9I2P	ENST00000427941	?	1
OR9I3P	ENST00000608484	?	1