Locus 46 (rs217181)

SNiPAcard

Block annotations

Block info

genomic range	chr16:72,079,657-72,114,002
block size	34,346 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.184 [-0.937 – 0.205]	gene(s) hit or close-by	HPR, RP11-384M15.3, TXNL4B
phastCons	μ = 0.146 [0.003 – 0.375]	eQTL gene(s)	MARVELD3
GERP++	μ = -0.203 [-0.727 – 0.118]	potentially regulated gene(s)	–
CADD score	μ = 4.410 [0.091 – 11.43]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Metabolite levels	$<1.00\times10^{-36}$	GWAS Catalog	22916037	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MARVELD3	?	ENSG00000140832	sun exposed skin	6.74×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HPR	downstream gene variant	2858	ENST00000356967	?	ENSP00000349451	1
HPR	downstream gene variant	2857	ENST00000540303	NM_020995.3	ENSP00000441828	1
HPR	downstream gene variant	2857	ENST00000561690	?	ENSP00000462916	1
RP11-384M15.3	upstream gene variant	4754	ENST00000561827	?	?	1
TXNL4B	downstream gene variant	4756	ENST00000423037	NM_001142317.1	ENSP00000408130	1
TXNL4B	downstream gene variant	4750	ENST00000268483	NM_017853.2	ENSP00000268483	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HPR	ENST00000561690	?	ENSP00000462916	1
HPR	ENST00000566168	?	?	1
HPR	ENST00000356967	?	ENSP00000349451	1

HPR	ENST00000540303	NM_020995.3	ENSP00000441828	1
TXNL4B	ENST00000562153	?	ENSP00000454635	3

Locus 46 (sentinel SNP rs217181 + 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.