

SNiPAcad

Block annotations

Block info

genomic range	chr16:72,079,657-72,114,002 <i>e!</i>
block size	34,346 bp
variant count	3 variants

Basic features

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


Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Metabolite levels	<1.00×10 ⁻³⁶	GWAS Catalog	22916037 	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MARVELD3 <i>e!</i>	?	ENSG00000140832 <i>e!</i>	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 <i>e!</i>	downstream gene variant	2858	ENST00000356967 <i>e!</i>	?	ENSP00000349451 <i>e!</i>	1
HPR <i>e!</i>	downstream gene variant	2857	ENST00000540303 <i>e!</i>	NM_020995.3	ENSP00000441828 <i>e!</i>	1
HPR <i>e!</i>	downstream gene variant	2857	ENST00000561690 <i>e!</i>	?	ENSP00000462916 <i>e!</i>	1
RP11-384M15.3 <i>e!</i>	upstream gene variant	4754	ENST00000561827 <i>e!</i>	?	?	1
TXNL4B <i>e!</i>	downstream gene variant	4756	ENST00000423037 <i>e!</i>	NM_001142317.1	ENSP00000408130 <i>e!</i>	1
TXNL4B <i>e!</i>	downstream gene variant	4750	ENST00000268483 <i>e!</i>	NM_017853.2	ENSP00000268483 <i>e!</i>	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HPR <i>e!</i>	ENST00000561690 <i>e!</i>	?	ENSP00000462916 <i>e!</i>	1
HPR <i>e!</i>	ENST00000566168 <i>e!</i>	?	?	1
HPR <i>e!</i>	ENST00000356967 <i>e!</i>	?	ENSP00000349451 <i>e!</i>	1

HPR <i>e!</i>	ENST00000540303 <i>e!</i>	NM_020995.3	ENSP00000441828 <i>e!</i>	1
TXNL4B <i>e!</i>	ENST00000562153 <i>e!</i>	?	ENSP00000454635 <i>e!</i>	3
