SNiPAcard

Block annotations

Block info

genomic range	chr16:71,968,701-71,972,060
block size	3,360 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.299 [-0.849 – 0.251]	**gene(s) hit or close-by**	IST1, PKD1L3
phastCons	μ = 0.032 [0 – 0.063]	**eQTL gene(s)**	AP1G1, HP, PHLPP2
GERP++	μ = -0.150 [-0.457 – 0.158]	**potentially regulated gene(s)**	–
CADD score	μ = 5.304 [4.722 – 5.886]	**disease gene(s)**	HP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
gamma-glutamyltyrosine	7.51×10^{-6}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

gene	trait	source DB	source entry/link
HP	ANHAPTOGLOBINEMIA	OMIM	MIM:614081

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PHLPP2	?	ENSG00000040199	testis	2.75×10^{-6} (p-value)	GTEx Portal V6	2
AP1G1	?	ENSG00000166747	aorta	4.33×10^{-6} (p-value)	GTEx Portal V6	1
HP	ENST00000570083	ILMN_1812433	monocyte	2.57×10^{-16} (p-value)	Zeller et al.	1
HP	ENST00000567185					
HP	ENST00000566821					
HP	ENST00000613898					
HP	ENST00000565574					
HP	ENST00000228226					
HP	ENST00000355906					
HP	ENST00000567612					
HP	ENST00000398131					
HP	ENST00000564499					
HP	ENST00000357763					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IST1	downstream gene variant	4159	ENST00000535424	NM_001270976.1	ENSP00000438399	1

IST1	downstream gene variant	4159	ENST00000606369	NM_001270979.1, NM_001270978.1	ENSP00000475853	1
IST1	downstream gene variant	3599	ENST00000378799	NM_001270975.1	ENSP00000368076	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PKD1L3	ENST00000620267	NM_181536.1	ENSP00000480090	2