SNiPAcard

Block annotations

Block info

genomic range	chr16:68,428,089-68,428,089
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.856	**gene(s) hit or close-by**	SMPD3
phastCons	0	**eQTL gene(s)**	ATP6V0D1, DUS2, TRADD
GERP++	-1.04	**potentially regulated gene(s)**	–
CADD score	2.866	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DUS2	?	ENSG00000167264	blood	7.29×10^{-6} (p-value)	GTEx Portal V6	1
DUS2	ENST00000432752	ILMN_1811650	monocyte	3.56×10^{-14} (p-value)	Zeller et al.	1
DUS2	ENST00000565263					
DUS2	ENST00000561965					
DUS2	ENST00000565980					
DUS2	ENST00000358896					
ATP6V0D1	ENST00000568620	ILMN_1795826	monocyte	6.15×10^{-13} (p-value)	Zeller et al.	1
ATP6V0D1	ENST00000563064					
ATP6V0D1	ENST00000567694					
ATP6V0D1	ENST00000602876					
ATP6V0D1	ENST00000561852					
ATP6V0D1	ENST00000290949					
ATP6V0D1	ENST00000567170					
ATP6V0D1	ENST00000564615					
ATP6V0D1	ENST00000565835					
ATP6V0D1	ENST00000426604					
ATP6V0D1	ENST00000563305					
TRADD	ENST00000486556	ILMN_1765851	monocyte	4.94×10^{-13} (p-value)	Zeller et al.	1
TRADD	ENST00000566247					
TRADD	ENST00000563348					
TRADD	ENST00000345057					

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SMPD3	ENST00000561749	?	ENSP00000457236	1
SMPD3	ENST00000566723	?	?	1
SMPD3	ENST00000219334	NM_018667.3	ENSP00000219334	1