Locus 281 (rs8049040)

SNiPAcard

Block annotations

Block info

genomic range	chr16:71,465,613-71,506,983
block size	41,371 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.134 [-3.461 – 0.421]	**gene(s) hit or close-by**	AC010547.9, RP11-510M2.4, RP11-510M2.8, ZNF19
phastCons	μ = 0.002 [0 – 0.01]	**eQTL gene(s)**	AC010547.9, MARVELD3, ZNF23
GERP++	μ = -1.694 [-9.66 – 1.52]	**potentially regulated gene(s)**	–
CADD score	μ = 1.501 [0.025 – 4.134]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MARVELD3	?	ENSG00000140832	sun exposed skin	6.69×10^{-8} (p-value)	GTEx Portal V6	6
MARVELD3	?	ENSG00000140832	thyroid	1.68×10^{-8} (p-value)	GTEx Portal V6	6
ZNF23	ENST00000358700	ILMN_1720027	blood	2.93×10^{-6} (p-value)	Westra et al.	2
ZNF23	ENST00000539742					
ZNF23	ENST00000428724					
AC010547.9	ENST00000561908					
ZNF23	ENST00000357254					
ZNF23	ENST00000393539					
ZNF23	ENST00000576258					
ZNF23	ENST00000564528					
ZNF23	ENST00000497160					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-510M2.8	downstream gene variant	3122	ENST00000571789	?	?	3
ZNF19	downstream gene variant	3083	ENST00000565541	?	ENSP00000454549	2
ZNF19	downstream gene variant	1304	ENST00000565100	?	ENSP00000462810	2
ZNF19	downstream gene variant	503	ENST00000288177	NM_006961.3	ENSP00000288177	2
ZNF19	downstream gene variant	1669	ENST00000569717	?	ENSP00000456143	2
ZNF19	downstream gene variant	1299	ENST00000565637	?	ENSP00000454864	2
ZNF19	downstream gene variant	3114	ENST00000568815	?	ENSP00000455796	2
ZNF19	downstream gene variant	1730	ENST00000564230	?	ENSP00000458105	2
ZNF19	downstream gene variant	4806	ENST00000561469	?	ENSP00000462021	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC010547.9	ENST00000561908	?	ENSP00000463741	2
RP11-510M2.4	ENST00000561754	?	?	1
RP11-510M2.4	ENST00000572770	?	?	1
ZNF19	ENST00000567225	?	ENSP00000462566	2