SNiPAcard

Block annotations

Block info

genomic range	chr2:309,097-309,097
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.126	**gene(s) hit or close-by**	AC079779.5, RP11-356M6.1
phastCons	0.02	**eQTL gene(s)**	SH3YL1
GERP++	-0.45	**potentially regulated gene(s)**	–
CADD score	1.239	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SH3YL1	?	ENSG00000035115	muscularis mucosae	9.42×10-6 (p-value)	GTEx Portal V6	1
SH3YL1	?	ENSG00000035115	tibial nerve	4.66×10-7 (p-value)	GTEx Portal V6	1
SH3YL1	?	ENSG00000035115	esophagus mucosa	8.54×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000584029 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC079779.5	downstream gene variant	23	ENST00000591927	?	?	1
AC079779.5	downstream gene variant	1199	ENST00000586715	?	?	1
AC079779.5	upstream gene variant	2260	ENST00000591158	?	?	1
AC079779.5	upstream gene variant	2501	ENST00000590255	?	?	1
AC079779.5	upstream gene variant	830	ENST00000587073	?	?	1
AC079779.5	upstream gene variant	1314	ENST00000588334	?	?	1
AC079779.5	upstream gene variant	2575	ENST00000585703	?	?	1
AC079779.5	upstream gene variant	1944	ENST00000591178	?	?	1
AC079779.5	downstream gene variant	124	ENST00000589936	?	?	1
AC079779.5	downstream gene variant	121	ENST00000591244	?	?	1
AC079779.5	upstream gene variant	1940	ENST00000457938	?	?	1
AC079779.5	upstream gene variant	1917	ENST00000588842	?	?	1
AC079779.5	upstream gene variant	2606	ENST00000592090	?	?	1

Putative effect on transcript

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC079779.5	ENST00000586772	?	1
AC079779.5	ENST00000589588	?	1
AC079779.5	ENST00000586311	?	1
AC079779.5	ENST00000586235	?	1
AC079779.5	ENST00000592366	?	1
AC079779.5	ENST00000585783	?	1
AC079779.5	ENST00000591122	?	1
AC079779.5	ENST00000592687	?	1
AC079779.5	ENST00000586673	?	1
AC079779.5	ENST00000590678	?	1
AC079779.5	ENST00000587796	?	1
AC079779.5	ENST00000416685	?	1
AC079779.5	ENST00000612487	?	1
AC079779.5	ENST00000592553	?	1
AC079779.5	ENST00000609984	?	1
AC079779.5	ENST00000591015	?	1
AC079779.5	ENST00000589124	?	1
RP11-356M6.1	ENST00000619113	?	1