SNiPAcard

Block annotations

Block info

genomic range	chr22:33,172,005-33,173,569
block size	1,565 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.759 [-2.942 – 1.425]	**gene(s) hit or close-by**	SYN3
phastCons	μ = 0.003 [0 – 0.005]	**eQTL gene(s)**	–
GERP++	μ = -0.100 [-1.82 – 1.62]	**potentially regulated gene(s)**	–
CADD score	μ = 2.545 [1.118 – 3.972]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000673280 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	DNase1, CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		liver (HepG2)	Rad21, H3K4me1, CTCF
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF
		A549	CTCF
		skin (NHEK)	CTCF

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SYN3	ENST00000462268	?	?	2
SYN3	ENST00000358763	NM_003490.3, NM_001135774.1, NM_133633.2	ENSP00000351614	2