Locus 319 (rs8044476)

SNiPAcard

Block annotations

Block info

genomic range	chr16:72,747,685-72,800,567
block size	52,883 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.129 [-2.24 – 1.711]	**gene(s) hit or close-by**	RP5-991G20.1
phastCons	μ = 0.206 [0.001 – 1]	**eQTL gene(s)**	–
GERP++	μ = 0.692 [-0.565 – 3.93]	**potentially regulated gene(s)**	C16orf47
CADD score	μ = 7.917 [0.325 – 17.48]	**disease gene(s)**	–

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000193764	1	ENCP00000021721	C16orf47

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000509797 (TF binding site)	1	embryonic stem cell (H1ESC)	POU5F1, Nanog, BCL11A, H3K4me2, USF1, DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP5-991G20.1	ENST00000563328	?	?	5