SNiPAcard

Block annotations

Block info

genomic range	chr4:15,766,933-15,817,324
block size	50,392 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.995 [-3.896 – 0.774]	**gene(s) hit or close-by**	CD38
phastCons	μ = 0.026 [0 – 0.239]	**eQTL gene(s)**	CD38, FBXL5
GERP++	μ = -0.163 [-2.82 – 2.18]	**potentially regulated gene(s)**	RP11-442P12.1, RP11-442P12.2
CADD score	μ = 3.121 [0.076 – 13.93]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FBXL5	ENST00000511441	ILMN_2378100	blood	3.65×10-6 (p-value)	Westra et al.	4
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
CD38	?	ENSG00000004468	esophagus mucosa	1.41×10-6 (p-value)	GTEx Portal V6	10

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000374279	1	ENCP00000040482	RP11-442P12.1
		ENCP00000040479	RP11-442P12.2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CD38	upstream gene variant	895	ENST00000510674	?	ENSP00000423047	11

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CD38	ENST00000502843	?	ENSP00000427277	11
CD38	ENST00000511430	?	?	11
CD38	ENST00000506191	?	?	11
CD38	ENST00000226279	NM_001775.2	ENSP00000226279	11