Locus 232 (rs28385609)

SNiPAcard

Block annotations

Block info

genomic range	chr6:123,121,472-123,128,536
block size	7,065 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 1.207 [-1.366 – 6.514]	gene(s) hit or close-by	SMPDL3A
phastCons	μ = 0.250 [0 – 1]	eQTL gene(s)	SMPDL3A
GERP++	μ = 0.172 [-3.94 – 5.72]	potentially regulated gene(s)	SMPDL3A
CADD score	μ = 5.927 [0.363 – 14.62]	disease gene(s)	–

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SMPDL3A	missense variant	ENST00000539041	NM_001286138.1	ENSP00000442152	S/P	Tct/Cct	?	?	1
SMPDL3A	missense variant	ENST00000368440	NM_006714.4	ENSP00000357425	S/P	Tct/Cct	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SMPDL3A	?	ENSG00000172594	tibial nerve	2.43×10^{-5} (p-value)	GTEx Portal V6	1
SMPDL3A	?	ENSG00000172594	transformed fibroblasts	1.28×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000715093	1	ENST00000368440, ENST00000539041	SMPDL3A

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001226031 (promoter flanking region)	2	NHLF	DNase1
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		muscle (HSMM)	DNase1, H3K36me3, H3K4me2
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	DNase1, H3K4me1, H3K4me2, FOXA1, H3K36me3
		lung (IMR90)	DNase1, H3K36me3
		blood (GM12878)	BATF
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K4me2, DNase1

ENSR00001226034 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SMPDL3A	downstream gene variant	2982	ENST00000487215	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SMPDL3A	ENST00000539041	NM_001286138.1	ENSP00000442152	3
SMPDL3A	ENST00000368440	NM_006714.4	ENSP00000357425	3