Locus 318 (rs6865582)

SNiPAcard

Block annotations

Block info

genomic range	chr5:78,176,721-78,201,833
block size	25,113 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.281 [-0.077 – 0.791]	gene(s) hit or close-by	ARSB
phastCons	μ = 0.072 [0 – 0.418]	eQTL gene(s)	ARSB
GERP++	μ = 0.035 [-3.85 – 1.96]	potentially regulated gene(s)	–
CADD score	μ = 4.115 [0.341 – 6.101]	disease gene(s)	ARSB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ARSB	MULTIPLE SULFATASE DEFICIENCY	OMIM	MIM:272200
ARSB	MUCOPOLYSACCHARIDOSIS TYPE VI	OMIM	MIM:253200
ARSB	mucopolysaccharidosis type 6 (MPS6)	DECIPHER	MIM:253200
ARSB	Mucopolysaccharidosis type 6, rapidly progressing	OrphaNet	OrphaNet:276212
ARSB	Mucopolysaccharidosis type 6, slowly progressing	OrphaNet	OrphaNet:276223

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ARSB	?	ENSG00000113273	sun exposed skin	7.74×10-7 (p-value)	GTEx Portal V6	5

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001283744 (promoter flanking region)	3	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, DNase1
		Osteobl	H3K4me2, H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1, H3K4me2, H3K4me3
		muscle (HSMM)	H3K9ac, DNase1, H3K4me2, H3K27ac
		endothelium (HUVEC)	Max, DNase1
		liver (HepG2)	H3K4me1, H3K36me3
		blood (GM12878)	PU1, H3K4me1, H3K27ac, H3K4me2
		lung (IMR90)	DNase1, H3K27ac, H3K4me2
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

ARSB	downstream gene variant, upstream gene variant	375	ENST00000521800	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ARSB	ENST00000264914	NM_000046.3	ENSP00000264914	7
ARSB	ENST00000396151	NM_198709.2	ENSP00000379455	7
ARSB	ENST00000565165	?	ENSP00000456339	7