Locus 23 (rs2830585)

SNiPAcard

Block annotations

Block info

genomic range	chr21:28,280,322-28,329,010
block size	48,689 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.713 [-1.94 – 7.487]	gene(s) hit or close-by	ADAMTS5, AP001601.2, MIR4759
phastCons	μ = 0.127 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 0.144 [-6.12 – 6.03]	potentially regulated gene(s)	–
CADD score	μ = 7.436 [1.089 – 22.1]	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)
ADAMTS5	missense variant	ENST00000284987	NM_007038.3	ENSP00000284987	H/R	cAc/cGc	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000612277 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3
		cervix (HeLa-S3)	DNase1, Cjun, Jund, Max
		monocytes (Monocytes-CD14+)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MIR4759	upstream gene variant, downstream gene variant	4	ENST00000584048	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ADAMTS5	ENST00000284987	NM_007038.3	ENSP00000284987	6
AP001601.2	ENST00000426771	?	?	6

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ADAMTS5	ENST00000284987	NM_007038.3	ENSP00000284987	2