Locus 383 (rs229100)

SNiPAcard

Block annotations

Block info

genomic range	chr21:28,221,167-28,224,836
block size	3,670 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.097 [-2.218 – -0.069]	gene(s) hit or close-by	ADAMTS1
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	GABPA
GERP++	μ = -0.096 [-2.9 – 3]	potentially regulated gene(s)	–
CADD score	μ = 2.243 [1.488 – 3.601]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GABPA	ENST00000400075	ILMN_2175562	monocyte	1.26×10-4 (p-value)	Fairfax et al.	1
GABPA	ENST00000354828					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001668110 (enhancer)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	Rad21
		Osteobl	CTCF
		lung (IMR90)	CTCF
		blood (K562)	CTCF
		blood (DND-41)	CTCF
		muscle (HSMM)	CTCF
ENSR00000612272 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ADAMTS1	upstream gene variant	3439	ENST00000284984	NM_006988.3	ENSP00000284984	1