

SNiPACard

Block annotations

Block info


genomic range	chr21:28,221,167-28,224,836 <i>e!</i>
block size	3,670 bp
variant count	3 variants

Basic features

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

Direct effect on regulation

cis-eQTL

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

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001668110 <i>e!</i> (enhancer)	1	cervix (HeLa-S3) embryonic stem cell (H1ESC) Osteobl lung (IMR90) blood (K562) blood (DND-41) muscle (HSMM)	CTCF Rad21 CTCF CTCF CTCF CTCF CTCF
ENSR00000612272 <i>e!</i> (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 <i>e!</i>	upstream gene variant	3439	ENST00000284984 <i>e!</i>	NM_006988.3	ENSP00000284984 <i>e!</i>	1

