Locus 131 (rs17114699)

SNiPAcard

Block annotations

Block info

genomic range	chr14:21,158,910-21,158,910
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.017	gene(s) hit or close-by	ANG, RNASE4, RP11-903H12.3, RP11-903H12.5
phastCons	0	eQTL gene(s)	RNASE9
GERP++	-0.0912	potentially regulated gene(s)	–
CADD score	1.825	disease gene(s)	ANG

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ANG	AMYOTROPHIC LATERAL SCLEROSIS 9	OMIM	MIM:611895
ANG	Amyotrophic lateral sclerosis	OrphaNet	OrphaNet:803

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RNASE9	ENST00000404716	ILMN_1655474	monocyte	1.81×10^{-4} (p-value)	Fairfax et al.	1
RNASE9	ENST00000429244					
RNASE9	ENST00000338904					
RNASE9	ENST00000611135					
RNASE9	ENST00000556208					
RNASE9	ENST00000555230					
RNASE9	ENST00000553706					
RNASE9	ENST00000553541					
RNASE9	ENST00000557209					
RNASE9	ENST00000557068					
RNASE9	ENST00000554964					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001621075 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	H4K20me1, H3K79me2, Rad21, ELF1, H3K4me1, CTCF, H3K36me3
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF

skin (NHEK)	CTCF, DNase1
NHLF	CTCF
Osteobl	CTCF
blood (DND-41)	CTCF
breast (HMEC)	CTCF
cervix (HeLa-S3)	DNase1, CTCF
monocytes (Monocytes-CD14+)	DNase1, CTCF
endothelium (HUVEC)	CTCF
A549	DNase1, CTCF, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-903H12.3	downstream gene variant	2729	ENST00000554286	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANG	ENST00000336811	NM_001145.4	ENSP00000336762	1
ANG	ENST00000554073	?	?	1
ANG	ENST00000397990	NM_001097577.2	ENSP00000381077	1
RNASE4	ENST00000397995	?	ENSP00000381081	1
RNASE4	ENST00000555597	NM_194431.2	ENSP00000451624	1
RNASE4	ENST00000555835	NM_001282193.1, NM_001282192.1, NM_002937.4	ENSP00000452245	1
RP11-903H12.5	ENST00000553909	?	ENSP00000477037	1