Locus 286 (rs1888560)

SNiPAcard

Block annotations

Block info

genomic range	chr14:21,164,245-21,164,245
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.241	gene(s) hit or close-by	ANG, RNASE4, RP11-903H12.3, RP11-903H12.5
phastCons	0	eQTL gene(s)	ANG, RNASE4, RP11-903H12.5
GERP++	0.639	potentially regulated gene(s)	–
CADD score	5.488	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)
RP11-903H12.5	ENST00000553909	ILMN_1776602	adipocyte	9.59×10-6 (p-value)	MuTHER consortium	1
RNASE4	ENST00000555597					
RNASE4	ENST00000397995					
RNASE4	ENST00000555835					
RP11-903H12.5	ENST00000553909	ILMN_2408572	blood	1.68×10-6 (p-value)	Westra et al.	1
ANG	ENST00000554073					
RNASE4	ENST00000397995					
RNASE4	ENST00000555597					
RNASE4	ENST00000555835					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANG	downstream gene variant	1900	ENST00000397990	NM_001097577.2	ENSP00000381077	1
ANG	downstream gene variant	1900	ENST00000336811	NM_001145.4	ENSP00000336762	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)

ANG	ENST00000554073	?	?	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
RP11-903H12.3	ENST00000554286	?	?	1
RP11-903H12.5	ENST00000553909	?	ENSP00000477037	1