Locus 104 (rs17114671)

SNiPAcard

Block annotations

Block info

genomic range	chr14:21,143,659-21,157,936
block size	14,278 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.527 [-1.866 – 1.605]	gene(s) hit or close-by	ANG, RNASE4, RP11-903H12.3, RP11-903H12.5, Y_RNA
phastCons	μ = 0.085 [0 – 0.813]	eQTL gene(s)	ANG, RNASE4, RP11-903H12.3, RP11-903H12.5
GERP++	μ = -2.344 [-7.85 – 2.33]	potentially regulated gene(s)	ANG, ANG, RNASE4
CADD score	μ = 3.885 [0.054 – 13.28]	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)
RP11-903H12.3	?	ENSG00000258451	lung	7.30×10^{-6} (p-value)	GTEx Portal V6	1
RP11-903H12.5	ENST00000553909	ILMN_2408572	monocyte	3.74×10^{-5} (p-value)	Fairfax et al.	1
ANG	ENST00000554073					
RNASE4	ENST00000555597					
RNASE4	ENST00000397995					
RNASE4	ENST00000555835					
ANG	ENST00000397990	ILMN_1760727	monocyte	4.57×10^{-9} (p-value)	Fairfax et al.	1
ANG	ENST00000336811					
ANG	ENST00000397990	ILMN_2294976	monocyte	5.32×10^{-8} (p-value)	Fairfax et al.	1
RP11-903H12.5	ENST00000553909					
ANG	ENST00000336811					
ANG	ENST00000397990	ILMN_2294978	monocyte	2.65×10^{-8} (p-value)	Fairfax et al.	1
RP11-903H12.5	ENST00000553909					
ANG	ENST00000336811					
RP11-903H12.5	ENST00000553909	ILMN_1776602	monocyte	1.03×10^{-6} (p-value)	Fairfax et al.	1
RNASE4	ENST00000397995					
RNASE4	ENST00000555597					
RNASE4	ENST00000555835					
ANG	ENST00000397990	ILMN_1696974	monocyte	1.31×10^{-8} (p-value)	Fairfax et al.	1
RP11-903H12.5	ENST00000553909					
ANG	ENST00000336811					
RP11-903H12.5	ENST00000553909	ILMN_2408572	monocyte	3.45×10^{-17} (p-value)	Zeller et al.	2
ANG	ENST00000554073					
RNASE4	ENST00000397995					

RNASE4	ENST00000555597						
RNASE4	ENST00000555835						
ANG	ENST00000397990	ILMN_1760727	monocyte	5.55×10^{-19} (p-value)	Zeller et al.	2	
ANG	ENST00000336811						
ANG	ENST00000397990	ILMN_2294976	monocyte	1.01×10^{-17} (p-value)	Zeller et al.	2	
RP11-903H12.5	ENST00000553909						
ANG	ENST00000336811						
ANG	ENST00000397990	ILMN_2294978	monocyte	1.33×10^{-15} (p-value)	Zeller et al.	2	
RP11-903H12.5	ENST00000553909						
ANG	ENST00000336811						
RP11-903H12.5	ENST00000553909	ILMN_1776602	monocyte	7.17×10^{-20} (p-value)	Zeller et al.	2	
RNASE4	ENST00000555597						
RNASE4	ENST00000397995						
RNASE4	ENST00000555835						
ANG	ENST00000397990	ILMN_1696974	monocyte	2.28×10^{-16} (p-value)	Zeller et al.	2	
RP11-903H12.5	ENST00000553909						
ANG	ENST00000336811						

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000019332		RNASE4
		ANG
ENCP00000019333		RNASE4
		ANG

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001455837 (promoter)	4	embryonic stem cell (H1ESC)	POU5F1, CTCF, PolII, Rad21, H3K4me3, H3K9ac, USF1, SP1, H3K4me2, TAF1, Yy1, H3K27ac, Sin3Ak20, TAF7, DNase1
		HSMMtube	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		blood (K562)	DNase1, Egr1, Max, USF1, H2AZ, PolII, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H3K9ac, DNase1, H3K4me2, H3K27ac, H2AZ, H3K4me3
		liver (HepG2)	H3K4me2, HNF4A, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K36me3, H3K27me3, DNase1, p300, H2AZ, Gabp, FOXA2, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, ELF1, PolIII, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K9ac, DNase1, H3K27ac, H3K4me2, H3K4me3
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K4me3, H3K4me2, H3K9ac, H3K27ac, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me2, H3K9ac, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANG	upstream gene variant	96	ENST00000397990	NM_001097577.2	ENSP00000381077	7
ANG	upstream gene variant	169	ENST00000554073	?	?	1
RNASE4	upstream gene variant	138	ENST00000397995	?	ENSP00000381081	1

RNASE4	upstream gene variant	105	ENST00000555597	NM_194431.2	ENSP00000451624	7
RP11-903H12.3	downstream gene variant	3703	ENST00000554286	?	?	2
RP11-903H12.5	upstream gene variant	142	ENST00000553909	?	ENSP00000477037	1
Y_RNA	upstream gene variant, downstream gene variant	3878	ENST00000362591	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANG	ENST00000554073	?	?	7
ANG	ENST00000336811	NM_001145.4	ENSP00000336762	7
ANG	ENST00000397990	NM_001097577.2	ENSP00000381077	1
RNASE4	ENST00000555835	NM_001282193.1, NM_001282192.1, NM_002937.4	ENSP00000452245	7
RNASE4	ENST00000397995	?	ENSP00000381081	7
RNASE4	ENST00000555597	NM_194431.2	ENSP00000451624	1
RP11-903H12.5	ENST00000553909	?	ENSP00000477037	7

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANG	ENST00000336811	NM_001145.4	ENSP00000336762	1
RNASE4	ENST00000555835	NM_001282193.1, NM_001282192.1, NM_002937.4	ENSP00000452245	1