# 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 | $7.30\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| RP11-903H12.5 | ENST00000553909 | ILMN_2408572 | monocyte | $3.74\times10^{-5}$ (p-value) | Fairfax et al. | 1 |
| ANG | ENST00000554073 | | | | | |
| RNASE4 | ENST00000555597 | | | | | |
| RNASE4 | ENST00000397995 | | | | | |
| RNASE4 | ENST00000555835 | | | | | |
| ANG | ENST00000397990 | ILMN_1760727 | monocyte | $4.57\times10^{-9}$ (p-value) | Fairfax et al. | 1 |
| ANG | ENST00000336811 | | | | | |
| ANG | ENST00000397990 | ILMN_2294976 | monocyte | $5.32\times10^{-8}$ (p-value) | Fairfax et al. | 1 |
| RP11-903H12.5 | ENST00000553909 | | | | | |
| ANG | ENST00000336811 | | | | | |
| ANG | ENST00000397990 | ILMN_2294978 | monocyte | $2.65\times10^{-8}$ (p-value) | Fairfax et al. | 1 |
| RP11-903H12.5 | ENST00000553909 | | | | | |
| ANG | ENST00000336811 | | | | | |
| RP11-903H12.5 | ENST00000553909 | ILMN_1776602 | monocyte | $1.03\times10^{-6}$ (p-value) | Fairfax et al. | 1 |
| RNASE4 | ENST00000397995 | | | | | |
| RNASE4 | ENST00000555597 | | | | | |
| RNASE4 | ENST00000555835 | | | | | |
| ANG | ENST00000397990 | ILMN_1696974 | monocyte | $1.31\times10^{-8}$ (p-value) | Fairfax et al. | 1 |
| RP11-903H12.5 | ENST00000553909 | | | | | |
| ANG | ENST00000336811 | | | | | |
| RP11-903H12.5 | ENST00000553909 | ILMN_2408572 | monocyte | $3.45\times10^{-17}$ (p-value) | Zeller et al. | 2 |
| ANG | ENST00000554073 | | | | | |
| RNASE4 | ENST00000397995 | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| RNASE4 | ENST00000555597 | | | | | |
| RNASE4 | ENST00000555835 | | | | | |
| ANG | ENST00000397990 | ILMN_1760727 | monocyte | $5.55\times10^{-19}$ (p-value) | Zeller et al. | 2 |
| ANG | ENST00000336811 | | | | | |
| ANG | ENST00000397990 | ILMN_2294976 | monocyte | $1.01\times10^{-17}$ (p-value) | Zeller et al. | 2 |
| RP11-903H12.5 | ENST00000553909 | | | | | |
| ANG | ENST00000336811 | | | | | |
| ANG | ENST00000397990 | ILMN_2294978 | monocyte | $1.33\times10^{-15}$ (p-value) | Zeller et al. | 2 |
| RP11-903H12.5 | ENST00000553909 | | | | | |
| ANG | ENST00000336811 | | | | | |
| RP11-903H12.5 | ENST00000553909 | ILMN_1776602 | monocyte | $7.17\times10^{-20}$ (p-value) | Zeller et al. | 2 |
| RNASE4 | ENST00000555597 | | | | | |
| RNASE4 | ENST00000397995 | | | | | |
| RNASE4 | ENST00000555835 | | | | | |
| ANG | ENST00000397990 | ILMN_1696974 | monocyte | $2.28\times10^{-16}$ (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 |