# Locus 196 (rs540254)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr1:160,759,519-160,767,737 |
| **block size** | 8,219 bp |
| **variant count** | 7 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.349 [-1.062 – 0.48] | **gene(s) hit or close-by** | LY9 |
| **phastCons** | μ = 0.006 [0 – 0.034] | **eQTL gene(s)** | LY9, SLAMF7 |
| **GERP++** | μ = -0.668 [-2.19 – 0.577] | **potentially regulated gene(s)** | – |
| **CADD score** | μ = 4.528 [0.201 – 10.73] | **disease gene(s)** | – |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| SLAMF7 | ENST00000368043 | ILMN_1710923 | blood | $1.14\times10^{-7}$ (p-value) | MuTHER consortium | 2 |
| SLAMF7 | ENST00000368042 | | | | | |
| SLAMF7 | ENST00000458602 | | | | | |
| SLAMF7 | ENST00000444090 | | | | | |
| SLAMF7 | ENST00000441662 | | | | | |
| SLAMF7 | ENST00000621377 | | | | | |
| SLAMF7 | ENST00000359331 | | | | | |
| SLAMF7 | ENST00000484221 | | | | | |
| SLAMF7 | ENST00000458104 | | | | | |
| LY9 | ENST00000392203 | ILMN_1731928 | b-cell | $1.91\times10^{-5}$ (p-value) | Fairfax et al. | 1 |
| LY9 | ENST00000263285 | | | | | |
| LY9 | ENST00000368037 | | | | | |
| LY9 | ENST00000368035 | | | | | |

## Putative effect on regulation

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000167128 (open chromatin region) | 1 | blood (GM12878) | H3K4me1, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K9ac |
| | | blood (K562) | H3K27me3 |
| | | breast (HMEC) | DNase1 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| LY9 | upstream gene variant | 1165 | ENST00000368037 | NM_001261456.1 | ENSP00000357016 | 4 |
| LY9 | upstream gene variant | 1302 | ENST00000485624 | ? | ? | 4 |
| LY9 | upstream gene variant | 1203 | ENST00000392203 | NM_001261457.1 | ENSP00000376039 | 4 |
| LY9 | upstream gene variant | 1197 | ENST00000474998 | ? | ENSP00000476321 | 4 |
| LY9 | upstream gene variant | 1299 | ENST00000471816 | ? | ? | 4 |
| LY9 | upstream gene variant | 1290 | ENST00000480837 | ? | ? | 4 |
| LY9 | upstream gene variant | 1278 | ENST00000368039 | NM_001033667.2 | ENSP00000357018 | 4 |

| | | | | | | |
|---|---|---|---|---|---|---|
| LY9 | upstream gene variant | 1291 | ENST00000490902 | ? | ENSP00000473643 | 4 |
| LY9 | upstream gene variant | 1249 | ENST00000263285 | NM_002348.3 | ENSP00000263285 | 4 |

## Putative effect on transcript

### Synonymous coding variant

| gene | affected transcript | RefSeq id | protein | AA's | exchanged codons | variant(s) |
|---|---|---|---|---|---|---|
| LY9 | ENST00000474998 | ? | ENSP00000476321 | S | tcT/tcC | 1 |

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| LY9 | ENST00000368037 | NM_001261456.1 | ENSP00000357016 | 2 |
| LY9 | ENST00000485624 | ? | ? | 2 |
| LY9 | ENST00000392203 | NM_001261457.1 | ENSP00000376039 | 2 |
| LY9 | ENST00000474998 | ? | ENSP00000476321 | 2 |
| LY9 | ENST00000471816 | ? | ? | 2 |
| LY9 | ENST00000480837 | ? | ? | 2 |
| LY9 | ENST00000368039 | NM_001033667.2 | ENSP00000357018 | 2 |
| LY9 | ENST00000490902 | ? | ENSP00000473643 | 2 |
| LY9 | ENST00000263285 | NM_002348.3 | ENSP00000263285 | 2 |