# Locus 60 (rs7959452)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr12:69,729,434-69,747,834 |
| block size | 18,401 bp |
| variant count | 8 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.116 [-1.544 – 0.956] | gene(s) hit or close-by | LYZ, RP11-1143G9.4, RP11-1143G9.5 |
| phastCons | μ = 0.171 [0 – 0.922] | eQTL gene(s) | CPM, LYZ, RP11-1143G9.4, YEATS4 |
| GERP++ | μ = -1.254 [-4.79 – 3.4] | potentially regulated gene(s) | – |
| CADD score | μ = 4.343 [0.265 – 18.4] | disease gene(s) | LYZ |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| LYZ | AMYLOIDOSIS, FAMILIAL VISCERAL | OMIM | MIM:105200 |
| LYZ | Familial renal amyloidosis due to lysozyme variant | OrphaNet | OrphaNet:93561 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| YEATS4 | ENST00000247843 | ILMN_1801387 | blood | 8.39×10-6 (p-value) | MuTHER consortium | 5 |
| YEATS4 | ENST00000548020 | | adipocyte | 1.48×10-9 (p-value) | MuTHER consortium | 5 |
| | | | skin | 6.04×10-6 (p-value) | MuTHER consortium | 5 |
| YEATS4 | ? | ENSG00000127337 | uterus | 6.55×10-8 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | muscularis mucosae | 9.16×10-13 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | muscularis mucosae | 5.32×10-15 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | muscularis mucosae | 4.85×10-10 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | lung | 4.55×10-34 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | lung | 3.98×10-11 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | lung | 2.35×10-33 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | atrial appendage | 7.74×10-13 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | atrial appendage | 1.93×10-8 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | atrial appendage | 2.72×10-10 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | tibial artery | 4.52×10-25 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | tibial artery | 3.94×10-17 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | tibial artery | 1.47×10-23 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | breast | 8.36×10-11 (p-value) | GTEx Portal V6 | 8 |

| | | | | | | |
|---|---|---|---|---|---|---|
| YEATS4 | ? | ENSG00000127337 | breast | 3.55×10-15 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | breast | 2.27×10-10 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | blood | 1.00×10-56 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | blood | 1.24×10-15 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | blood | 8.25×10-53 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | thyroid | 1.81×10-16 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | thyroid | 3.11×10-34 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | thyroid | 3.44×10-15 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | skeletal muscle | 3.51×10-13 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | skeletal muscle | 2.36×10-31 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | skeletal muscle | 3.17×10-10 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | transverse colon | 5.76×10-11 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | transverse colon | 5.70×10-9 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | transverse colon | 1.73×10-9 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | EBV lymphocytes | 1.19×10-12 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | unexposed skin | 3.15×10-9 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | unexposed skin | 1.21×10-19 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | unexposed skin | 5.30×10-7 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | sun exposed skin | 3.83×10-16 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | sun exposed skin | 1.00×10-29 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | sun exposed skin | 1.53×10-12 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | left ventricle | 4.12×10-13 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | left ventricle | 1.17×10-12 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | left ventricle | 2.02×10-13 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | aorta | 3.92×10-15 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | aorta | 5.92×10-9 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | aorta | 2.28×10-13 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | subcutaneous adipocytes | 3.53×10-24 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | subcutaneous adipocytes | 1.16×10-31 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | subcutaneous adipocytes | 1.83×10-21 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | visceral adipocytes | 2.86×10-15 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | visceral adipocytes | 2.40×10-14 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | visceral adipocytes | 1.56×10-13 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | stomach | 3.45×10-7 (p-value) | GTEx Portal V6 | 4 |
| YEATS4 | ? | ENSG00000127337 | stomach | 1.23×10-6 (p-value) | GTEx Portal V6 | 4 |
| RP11-1143G9.4 | ? | ENSG00000257764 | stomach | 8.13×10-7 (p-value) | GTEx Portal V6 | 1 |

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|---|---|---|---|---|---|---|
| YEATS4 | ? | ENSG00000127337 | nucleus accumbens | 4.71×10-9 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | spleen | 2.72×10-8 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | spleen | 2.92×10-8 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | tibial nerve | 5.77×10-21 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | tibial nerve | 4.19×10-15 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | tibial nerve | 4.78×10-18 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | coronary artery | 2.17×10-6 (p-value) | GTEx Portal V6 | 7 |
| YEATS4 | ? | ENSG00000127337 | sigmoid colon | 3.81×10-9 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | vagina | 4.28×10-9 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | esophagus mucosa | 3.72×10-8 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | esophagus mucosa | 1.35×10-9 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | esophagus mucosa | 4.97×10-7 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | adrenal gland | 2.06×10-10 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | adrenal gland | 1.12×10-8 (p-value) | GTEx Portal V6 | 8 |
| RP11-1143G9.4 | ? | ENSG00000257764 | adrenal gland | 5.23×10-9 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | testis | 6.72×10-7 (p-value) | GTEx Portal V6 | 8 |
| YEATS4 | ? | ENSG00000127337 | testis | 1.82×10-12 (p-value) | GTEx Portal V6 | 4 |
| RP11-1143G9.4 | ? | ENSG00000257764 | testis | 5.10×10-6 (p-value) | GTEx Portal V6 | 1 |
| YEATS4 | ? | ENSG00000127337 | cerebellum | 4.52×10-6 (p-value) | GTEx Portal V6 | 8 |
| LYZ | ? | ENSG00000090382 | pancreas | 6.60×10-6 (p-value) | GTEx Portal V6 | 1 |
| LYZ | ? | ENSG00000090382 | prostate | 2.08×10-6 (p-value) | GTEx Portal V6 | 1 |
| YEATS4 | ? | ENSG00000127337 | caudate basal ganglia | 5.11×10-7 (p-value) | GTEx Portal V6 | 1 |
| YEATS4 | ENST00000247843 | ILMN_1801387 | monocyte | 1.26×10-13 (p-value) | Fairfax et al. | 3 |
| YEATS4 | ENST00000548020 | | b-cell | 1.55×10-7 (p-value) | Fairfax et al. | 3 |
| LYZ | ENST00000261267 | ILMN_2162972 | monocyte | 8.90×10-78 (p-value) | Fairfax et al. | 3 |
| CPM | ENST00000338356 | ILMN_2299795 | monocyte | 1.58×10-8 (p-value) | Fairfax et al. | 3 |
| CPM | ENST00000551568 | | | | | |
| ? | ? | ILMN_1815205 | monocyte | 2.31×10-17 (p-value) | Fairfax et al. | 3 |
| | | | b-cell | 4.27×10-18 (p-value) | | |
| YEATS4 | ? | ENSG00000127337 | cortex | 1.95×10-6 (p-value) | GTEx Portal V6 | 4 |
| YEATS4 | ? | ENSG00000127337 | cerebellar hemisphere | 3.87×10-6 (p-value) | GTEx Portal V6 | 4 |

## Putative effect on regulation

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001464668 (open chromatin region) | 1 | endothelium (HUVEC) | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |
| ENSR00001612777 | 1 | NHLF | H3K27me3 |

| | | | |
|---|---|---|---|
| ENSR00001612777 (enhancer) | 1 | NHLF | H3K27me3 |
| | | HSMMtube | H3K27me3 |
| | | Osteobl | H3K27me3 |
| | | blood (DND-41) | H3K27me3 |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3 |
| | | endothelium (HUVEC) | H3K27me3 |
| | | liver (HepG2) | H3K4me1 |
| | | lung (IMR90) | H3K27me3 |
| | | A549 | H3K27me3 |
| ENSR00001612779 (enhancer) | 1 | monocytes (Monocytes-CD14+) | H3K4me1, H3K4me2, H3K27ac, H3K4me3 |
| | | liver (HepG2) | H3K27ac, H3K4me1 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| LYZ | upstream gene variant, downstream gene variant | 1297 | ENST00000548839 | ? | ENSP00000449969 | 4 |
| LYZ | upstream gene variant, downstream gene variant | 559 | ENST00000549690 | ? | ENSP00000449898 | 3 |
| LYZ | upstream gene variant | 1252 | ENST00000261267 | NM_000239.2 | ENSP00000261267 | 2 |
| RP11-1143G9.4 | downstream gene variant | 602 | ENST00000548900 | ? | ? | 1 |
| RP11-1143G9.5 | upstream gene variant | 3772 | ENST00000613291 | ? | ? | 1 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| LYZ | ENST00000549690 | ? | ENSP00000449898 | 1 |
| LYZ | ENST00000261267 | NM_000239.2 | ENSP00000261267 | 1 |

### 3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| LYZ | ENST00000261267 | NM_000239.2 | ENSP00000261267 | 1 |

### Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| RP11-1143G9.4 | ENST00000548900 | ? | 1 |