# Locus 22 (rs1993373)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr11:18,277,763-18,287,750 |
| block size | 9,988 bp |
| variant count | 7 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -1.167 [-2.326 – -0.307] | gene(s) hit or close-by | RNA5SP334, SAA1, ST13P5 |
| phastCons | μ = 0.001 [0 – 0.004] | eQTL gene(s) | GTF2H1, SAA1, SAA2, SAA2-SAA4 |
| GERP++ | μ = -1.923 [-6.95 – 1.81] | potentially regulated gene(s) | HPS5, PTPN5, SAA1, TSG101 |
| CADD score | μ = 1.247 [0.123 – 3.047] | disease gene(s) | SAA1, HPS5 |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | variant(s) |
|---|---|---|---|---|
| Amyloid A serum levels | $<3.00\times10^{-53}$ | GWAS Catalog | 21124955 | 1 |

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| SAA1 | Secondary amyloidosis | OrphaNet | OrphaNet:85445 |
| HPS5 | Hermansky-Pudlak syndrome without pulmonary fibrosis | OrphaNet | OrphaNet:231512 |
| HPS5 | HERMANSKY-PUDLAK SYNDROME 5 | OMIM | MIM:614074 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| SAA1 | ? | ENSG00000173432 | tibial artery | $6.31\times10^{-7}$ (p-value) | GTEx Portal V6 | 6 |
| SAA2 | ? | ENSG00000134339 | tibial artery | $3.39\times10^{-6}$ (p-value) | GTEx Portal V6 | 6 |
| SAA2-SAA4 | ? | ENSG00000255071 | tibial artery | $8.31\times10^{-6}$ (p-value) | GTEx Portal V6 | 6 |
| GTF2H1 | ENST00000534641 | ILMN_2157957 | blood | $7.08\times10^{-6}$ (p-value) | Westra et al. | 4 |
| GTF2H1 | ENST00000265963 | | | | | |

## Putative effect on regulation

### FANTOM5 expressed promoter

| SNiPA promoter id | variant(s) | associated transcript(s) | gene |
|---|---|---|---|
| FFCP00000055282 | 1 | ENST00000356524, ENST00000532858, ENST00000405158 | SAA1 |

### ENCODE promoter-associated distal DHS (Enhancer)

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000100066 | 1 | ENCP00000011406 | HPS5 |
| | | ENCP00000011423 | TSG101 |
| | | ENCP00000011443 | PTPN5 |

Locus 22 (sentinel SNP rs1993373 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

## Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000559405 (enhancer) | 1 | endothelium (HUVEC) | H3K27me3 |
| | | HSMMtube | H3K27me3 |
| | | liver (HepG2) | H3K4me1 |
| | | Osteobl | H3K27me3 |
| | | blood (GM12878) | H3K4me1 |
| | | blood (K562) | H3K27me3 |
| | | blood (DND-41) | H3K27me3 |
| ENSR00001568722 (enhancer) | 2 | NHLF | H3K27me3 |
| | | HSMMtube | H3K27me3 |
| | | Osteobl | H3K27me3 |
| | | blood (DND-41) | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | breast (HMEC) | H3K4me1 |
| | | muscle (HSMM) | DNase1 |
| | | endothelium (HUVEC) | H3K27me3 |
| | | liver (HepG2) | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |

## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| RNA5SP334 | upstream gene variant | 190 | ENST00000364825 | ? | ? | 3 |
| SAA1 | upstream gene variant | 687 | ENST00000356524 | NM_199161.3 | ENSP00000348918 | 2 |
| SAA1 | upstream gene variant | 61 | ENST00000405158 | NM_000331.4 | ENSP00000384906 | 3 |
| SAA1 | upstream gene variant | 57 | ENST00000532858 | NM_001178006.1 | ENSP00000436866 | 3 |
| ST13P5 | upstream gene variant, downstream gene variant | 1136 | ENST00000526214 | ? | ? | 6 |

# Putative effect on transcript

## 5'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| SAA1 | ENST00000356524 | NM_199161.3 | ENSP00000348918 | 1 |