# Locus 252 (rs672413)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr5:78,278,229-78,278,299 |
| **block size** | 71 bp |
| **variant count** | 2 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.353 [-0.918 – 0.212] | **gene(s) hit or close-by** | ARSB |
| **phastCons** | μ = 0.000 [0 – 0] | **eQTL gene(s)** | ARSB, BHMT |
| **GERP++** | μ = -1.920 [-2.1 – -1.74] | **potentially regulated gene(s)** | – |
| **CADD score** | μ = 2.612 [2.011 – 3.213] | **disease gene(s)** | ARSB |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | variant(s) |
|---|---|---|---|---|
| Blood and toenail selenium levels | $<5.00\times10^{-14}$ | GWAS Catalog | 25343990 | 1 |
| betaine | $2.73\times10^{-5}$ | Metabolomics GWAS Server | 24816252 | 1 |

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| ARSB | mucopolysaccharidosis type 6 (MPS6) | DECIPHER | MIM:253200 |
| ARSB | MULTIPLE SULFATASE DEFICIENCY | OMIM | MIM:272200 |
| ARSB | MUCOPOLYSACCHARIDOSIS TYPE VI | OMIM | MIM:253200 |
| ARSB | Mucopolysaccharidosis type 6, rapidly progressing | OrphaNet | OrphaNet:276212 |
| ARSB | Mucopolysaccharidosis type 6, slowly progressing | OrphaNet | OrphaNet:276223 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| BHMT | ENST00000274353 | ILMN_1656638 | adipocyte | $3.91\times10^{-5}$ (p-value) | MuTHER consortium | 1 |
| ARSB | ENST00000264914 | ILMN_1722045 | blood | $2.82\times10^{-6}$ (p-value) | Westra et al. | 1 |
| ARSB | ENST00000396151 | | | | | |
| ARSB | ENST00000565165 | | | | | |
| ARSB | ENST00000521800 | | | | | |
| ARSB | ENST00000264914 | ILMN_1722045 | liver | $<2.2\times10^{-16}$ (p-value) | Innocenti et al. | 1 |
| ARSB | ENST00000396151 | | | | | |
| ARSB | ENST00000565165 | | | | | |
| ARSB | ENST00000521800 | | | | | |
| ARSB | ? | ENSG00000113273 | liver | $2.04\times10^{-9}$ (p-value) | GTEx Portal V6 | 2 |
| ARSB | ? | ENSG00000113273 | sun exposed skin | $3.05\times10^{-11}$ (p-value) | GTEx Portal V6 | 2 |
| ARSB | ? | ENSG00000113273 | tibial artery | $4.84\times10^{-6}$ (p-value) | GTEx Portal V6 | 2 |

## Putative effect on regulation

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| ARSB | downstream gene variant | 2636 | ENST00000521117 | ? | ENSP00000428611 | 2 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| ARSB | ENST00000264914 | NM_000046.3 | ENSP00000264914 | 2 |
| ARSB | ENST00000396151 | NM_198709.2 | ENSP00000379455 | 2 |
| ARSB | ENST00000565165 | ? | ENSP00000456339 | 2 |