# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr9:136,128,546-136,147,295 |
| **block size** | 18,750 bp |
| **variant count** | 13 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.281 [-2.374 – 0.721] | **gene(s) hit or close-by** | ABO |
| **phastCons** | μ = 0.112 [0 – 0.656] | **eQTL gene(s)** | ABO, GBGT1, SLC2A6, SURF1 |
| **GERP++** | μ = 0.030 [-2.56 – 1.11] | **potentially regulated gene(s)** | – |
| **CADD score** | μ = 2.951 [0.029 – 13.47] | **disease gene(s)** | SURF1 |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | variant(s) |
|---|---|---|---|---|
| ADSGEGDFXAEGGGVR* | $1.17\times10^{-6}$ | Metabolomics GWAS Server | 24816252 | 2 |

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| SURF1 | LEIGH SYNDROME | OMIM | MIM:256000 |
| SURF1 | Complex IV deficiency | DECIPHER | MIM:220110 |
| SURF1 | Leigh Syndrome (nuclear DNA mutation) | DECIPHER | MIM:256000 |
| SURF1 | Fatal infantile cytochrome C oxidase deficiency | OrphaNet | OrphaNet:1561 |
| SURF1 | Leigh syndrome with cardiomyopathy | OrphaNet | OrphaNet:70474 |
| SURF1 | Leigh syndrome with leukodystrophy | OrphaNet | OrphaNet:255241 |
| SURF1 | SURF1-related Charcot-Marie-Tooth disease type 4 | OrphaNet | OrphaNet:391351 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| SURF1 | ? | ENSG00000148290 | sun exposed skin | $3.29\times10^{-6}$ (p-value) | GTEx Portal V6 | 12 |
| ABO | ? | ENSG00000175164 | esophagus mucosa | $2.75\times10^{-7}$ (p-value) | GTEx Portal V6 | 12 |
| ABO | ? | ENSG00000175164 | skeletal muscle | $4.13\times10^{-6}$ (p-value) | GTEx Portal V6 | 7 |
| GBGT1 | ENST00000372040 | ILMN_1652906 | blood | $2.77\times10^{-6}$ (p-value) | Westra et al. | 2 |
| GBGT1 | ENST00000470431 | | | | | |
| GBGT1 | ENST00000472281 | | | | | |
| GBGT1 | ENST00000372043 | | | | | |
| GBGT1 | ENST00000540636 | | | | | |
| SLC2A6 | ENST00000371899 | ILMN_1778321 | blood | $7.27\times10^{-5}$ (p-value) | Westra et al. | 2 |
| SLC2A6 | ENST00000485978 | | | | | |
| SLC2A6 | ENST00000371897 | | | | | |

## Putative effect on regulation

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001475637 (TF binding site) | 1 | embryonic stem cell (H1ESC) | H3K27me3 |
| | | endothelium (HUVEC) | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |
| | | blood (K562) | H3K36me3, H3K4me2, H3K9ac, H3K27ac, DNase1, Brg1, Ini1, PolII |
| | | A549 | H3K27me3 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| ABO | downstream gene variant | 2443 | ENST00000538324 | ? | ENSP00000483018 | 1 |
| ABO | downstream gene variant | 2017 | ENST00000611156 | NM_020469.2 | ENSP00000483265 | 1 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| ABO | ENST00000453660 | ? | ? | 12 |
| ABO | ENST00000538324 | ? | ENSP00000483018 | 12 |
| ABO | ENST00000611156 | NM_020469.2 | ENSP00000483265 | 12 |

### Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| ABO | ENST00000453660 | ? | 1 |