# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr2:3,639,909-3,648,186 |
| **block size** | 8,278 bp |
| **variant count** | 7 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.830 [-1.637 – 0.331] | **gene(s) hit or close-by** | AC010907.2, COLEC11 |
| **phastCons** | μ = 0.022 [0 – 0.08] | **eQTL gene(s)** | ALLC, COLEC11, RNASEH1-AS1 |
| **GERP++** | μ = 0.320 [-0.397 – 2.32] | **potentially regulated gene(s)** | – |
| **CADD score** | μ = 4.127 [0.417 – 5.959] | **disease gene(s)** | COLEC11 |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| COLEC11 | 3MC SYNDROME 2 | OMIM | MIM:265050 |
| COLEC11 | 3MC SYNDROME 2 | DECIPHER | MIM:265050 |
| COLEC11 | Craniofacial-ulnar-renal syndrome | OrphaNet | OrphaNet:293843 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| ALLC | ? | ENSG00000151360 | liver | $1.92\times10^{-6}$ (p-value) | GTEx Portal V6 | 2 |
| RNASEH1-AS1 | ? | ENSG00000234171 | thyroid | $2.37\times10^{-6}$ (p-value) | GTEx Portal V6 | 3 |
| COLEC11 | ? | ENSG00000118004 | pancreas | $2.78\times10^{-7}$ (p-value) | GTEx Portal V6 | 3 |
| RNASEH1-AS1 | ? | ENSG00000234171 | tibial nerve | $9.49\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| COLEC11 | ENST00000382062 | ILMN_1735192 | liver | $1.68\times10^{-11}$ (p-value) | Innocenti et al. | 1 |
| COLEC11 | ENST00000402922 | | | | | |
| COLEC11 | ENST00000403096 | | | | | |
| COLEC11 | ENST00000402794 | | | | | |
| COLEC11 | ENST00000487365 | | | | | |
| COLEC11 | ENST00000416132 | | | | | |
| COLEC11 | ENST00000460971 | | | | | |
| COLEC11 | ENST00000349077 | | | | | |
| COLEC11 | ENST00000404205 | | | | | |
| COLEC11 | ENST00000236693 | | | | | |
| COLEC11 | ENST00000418971 | | | | | |

## Putative effect on regulation

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|

| | | | |
|---|---|---|---|
| ENSR00000584312 (promoter flanking region) | 1 | monocytes (Monocytes-CD14+) | H3K27me3 |
| | | embryonic stem cell (H1ESC) | DNase1, TAF1, Yy1, Rad21, CTCF, H3K27me3 |
| | | endothelium (HUVEC) | DNase1 |
| | | Osteobl | H3K27me3, H3K4me2 |
| | | blood (GM12878) | H3K36me3 |
| | | blood (K562) | PolII, Max, Rad21, CTCF, DNase1 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| AC010907.2 | downstream gene variant | 2801 | ENST00000456450 | ? | ? | 2 |
| COLEC11 | upstream gene variant | 383 | ENST00000416132 | ? | ENSP00000410919 | 5 |
| COLEC11 | upstream gene variant | 321 | ENST00000349077 | NM_001255982.1, NM_024027.4 | ENSP00000339168 | 5 |
| COLEC11 | upstream gene variant | 184 | ENST00000460971 | ? | ? | 5 |
| COLEC11 | upstream gene variant | 61 | ENST00000382062 | NM_001255983.1, NM_001255984.1 | ENSP00000371494 | 5 |
| COLEC11 | upstream gene variant | 1310 | ENST00000418971 | NM_001255985.1 | ENSP00000411770 | 2 |
| COLEC11 | upstream gene variant | 278 | ENST00000236693 | NM_199235.2 | ENSP00000236693 | 5 |
| COLEC11 | upstream gene variant | 184 | ENST00000487365 | ? | ? | 5 |
| COLEC11 | upstream gene variant | 65 | ENST00000419002 | ? | ? | 5 |
| COLEC11 | upstream gene variant | 1310 | ENST00000438814 | ? | ENSP00000393167 | 2 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| COLEC11 | ENST00000416132 | ? | ENSP00000410919 | 2 |
| COLEC11 | ENST00000460971 | ? | ? | 2 |
| COLEC11 | ENST00000349077 | NM_001255982.1, NM_024027.4 | ENSP00000339168 | 2 |
| COLEC11 | ENST00000487365 | ? | ? | 2 |
| COLEC11 | ENST00000419002 | ? | ? | 2 |
| COLEC11 | ENST00000382062 | NM_001255983.1, NM_001255984.1 | ENSP00000371494 | 2 |
| COLEC11 | ENST00000236693 | NM_199235.2 | ENSP00000236693 | 2 |