# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr1:39,297,734-39,380,307 |
| **block size** | 82,574 bp |
| **variant count** | 59 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.217 [-3.141 – 2.757] | **gene(s) hit or close-by** | GJA9, MYCBP, RHBDL2, RP5-864K19.4, RP5-864K19.6, RP5-864K19.7, RRAGC |
| **phastCons** | μ = 0.074 [0 – 1] | **eQTL gene(s)** | GJA9, MYCBP, RHBDL2, RP5-864K19.6, RP5-864K19.7 |
| **GERP++** | μ = -0.291 [-6.75 – 6.03] | **potentially regulated gene(s)** | MACF1, RRAGC |
| **CADD score** | μ = 4.507 [0.029 – 20.2] | **disease gene(s)** | – |

## Direct effect on transcript

### Amino acid sequence alteration

| gene | effect type | affected transcript | RefSeq id | protein | exchanged AA's | exchanged codons | SIFT prediction | PolyPhen prediction | variant(s) |
|---|---|---|---|---|---|---|---|---|---|
| GJA9 | missense variant | ENST00000357771 | NM_030772.4 | ENSP00000350415 | I/V | Ata/Gta | ? | ? | 1 |
| GJA9 | missense variant | ENST00000360786 | ? | ENSP00000354020 | I/V | Ata/Gta | ? | ? | 1 |
| RHBDL2 | missense variant | ENST00000289248 | ? | ENSP00000289248 | M/L | Atg/Ctg | ? | ? | 1 |
| RHBDL2 | missense variant | ENST00000372990 | NM_017821.3, NM_001304746.1 | ENSP00000362081 | M/L | Atg/Ctg | ? | ? | 1 |
| RP5-864K19.6 | missense variant | ENST00000454994 | ? | ENSP00000406846 | Q/R | cAg/cGg | ? | ? | 1 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| RHBDL2 | ? | ENSG00000158315 | esophagus mucosa | $2.02\times10^{-6}$ (p-value) | GTEx Portal V6 | 36 |
| RP5-864K19.6 | ENST00000489803 | ILMN_1710161 | blood | $1.14\times10^{-4}$ (p-value) | Westra et al. | 1 |
| GJA9 | ENST00000357771 | | | | | |
| RP5-864K19.7 | ENST00000622355 | | | | | |
| MYCBP | ENST00000397572 | | | | | |

## Putative effect on regulation

### Transcription factor binding site variation

| transcription factor | binding motif | motif position | highly informative position | score change | variant(s) |
|---|---|---|---|---|---|
| HNF4A | MA0114.2 | 6 | yes | 0.000 | 1 |
| EGR1 | MA0162.2 | 13 | no | 0.000 | 1 |

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000015074 | 1 | ENCP00000001927 | MACF1 |
| | | ENCP00000001918 | RRAGC |

## Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001579011 (promoter) | 1 | embryonic stem cell (H1ESC) | H3K36me3, Jund, Sin3Ak20, H3K9ac, ATF3, TAF1, H3K4me2, SP1, USF1, H3K4me3, DNase1 |
| | | HSMMtube | H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1 |
| | | blood (K562) | Egr1, H3K27ac, Max, SP2, H3K9ac, Nfe2, USF1, Cmyc, H2AZ, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3 |
| | | skin (NHDF-AD) | H3K4me3, DNase1 |
| | | muscle (HSMM) | H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1 |
| | | liver (HepG2) | TAF1, USF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, PolII, ATF3, DNase1 |
| | | blood (GM12878) | DNase1, Egr1, H3K79me2, H3K4me3, USF1, Cfos, H3K4me2, H3K9ac, Pbx3, ATF3, PolII, H2AZ |
| | | lung (IMR90) | DNase1, H3K4me2, H3K36me3, H3K4me3, H3K9ac |
| | | nervous (NH-A) | DNase1, H3K4me3, H3K4me2, H3K9ac |
| | | skin (NHEK) | H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1 |
| | | NHLF | DNase1, H3K4me3, H3K9ac, H3K27ac |
| | | Osteobl | H2AZ, H3K27ac, H3K4me3, H3K4me2 |
| | | blood (DND-41) | H3K36me3, H3K4me3, H3K9ac, H3K4me1, H3K4me2, H3K27ac |
| | | breast (HMEC) | H3K4me3, H3K4me2, H3K27ac, DNase1 |
| | | cervix (HeLa-S3) | DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, Max, H3K4me3, H3K36me3 |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3 |
| | | endothelium (HUVEC) | H3K36me3, Max, H3K4me3, H3K4me2, PolII, DNase1 |
| | | A549 | H3K4me3, H3K4me2, H3K9ac, DNase1 |
| ENSR00000164746 (promoter) | 2 | embryonic stem cell (H1ESC) | H3K9ac, H3K36me3, CTCF, PolII, Rad21, Sin3Ak20, Yy1, H3K4me2, H3K4me3, DNase1 |
| | | HSMMtube | H3K36me3, DNase1, H2AZ, H3K4me3, H3K4me2 |
| | | blood (K562) | H3K4me3, DNase1, H3K27ac, Max, H3K9ac, Gabp, H2AZ, PolII, H3K4me2, H3K36me3 |
| | | skin (NHDF-AD) | H3K4me3, DNase1, H3K9ac, H3K4me2 |
| | | muscle (HSMM) | DNase1, H2AZ, H3K4me3, H3K4me2 |
| | | liver (HepG2) | Gabp, PolII, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1 |
| | | lung (IMR90) | DNase1, H3K4me2, H3K36me3, H3K4me3, H3K9ac |
| | | blood (GM12878) | PolII, H2AZ, DNase1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac |
| | | nervous (NH-A) | H3K4me2, H3K9ac, H3K4me3, DNase1 |
| | | skin (NHEK) | H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1 |
| | | NHLF | DNase1, H3K4me3, H3K9ac |
| | | Osteobl | H3K36me3, H3K4me2, H3K4me3 |
| | | blood (DND-41) | H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3 |
| | | breast (HMEC) | DNase1, H3K4me3, H3K9ac, H3K4me2, CTCF |
| | | cervix (HeLa-S3) | Nrf1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, Gabp, PolII, DNase1 |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3 |
| | | endothelium (HUVEC) | H3K4me3, H3K4me2, PolII, DNase1 |
| | | A549 | H3K36me3, DNase1, H3K4me3, H3K4me2, H3K9ac |
| ENSR00000281600 (CTCF binding site) | 1 | endothelium (HUVEC) | H3K36me3 |
| | | embryonic stem cell (H1ESC) | Rad21, CTCF, DNase1 |
| | | liver (HepG2) | FOXA1, CTCF |
| | | nervous (NH-A) | DNase1 |
| | | blood (K562) | Egr1, CTCF |
| | | skin (NHEK) | H3K36me3 |

## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| GJA9 | downstream gene variant, upstream gene variant | 541 | ENST00000357771 | NM_030772.4 | ENSP00000350415 | 5 |
| GJA9 | downstream gene variant, upstream gene variant | 441 | ENST00000360786 | ? | ENSP00000354020 | 10 |
| MYCBP | downstream gene variant, upstream gene variant | 499 | ENST00000495043 | ? | ? | 7 |
| MYCBP | downstream gene variant, upstream gene variant | 505 | ENST00000397572 | NM_012333.4 | ENSP00000380702 | 7 |
| MYCBP | downstream gene variant, upstream gene variant | 409 | ENST00000494695 | ? | ENSP00000485717 | 7 |
| MYCBP | downstream gene variant, upstream gene variant | 1172 | ENST00000465771 | ? | ? | 7 |

| | | | | | | |
|---|---|---|---|---|---|---|
| RHBDL2 | downstream gene variant | 868 | ENST00000540558 | ? | ENSP00000441097 | 4 |
| RHBDL2 | downstream gene variant | 888 | ENST00000372990 | NM_017821.3, NM_001304746.1 | ENSP00000362081 | 3 |
| RHBDL2 | downstream gene variant | 1377 | ENST00000289248 | ? | ENSP00000289248 | 2 |
| RP5-864K19.4 | downstream gene variant | 630 | ENST00000456813 | ? | ? | 7 |
| RP5-864K19.4 | downstream gene variant | 628 | ENST00000443161 | ? | ? | 7 |
| RP5-864K19.6 | downstream gene variant, upstream gene variant | 896 | ENST00000489803 | ? | ? | 4 |
| RP5-864K19.6 | downstream gene variant, upstream gene variant | 876 | ENST00000454994 | ? | ENSP00000406846 | 5 |
| RP5-864K19.6 | downstream gene variant, upstream gene variant | 891 | ENST00000489575 | ? | ? | 5 |
| RP5-864K19.6 | downstream gene variant, upstream gene variant | 891 | ENST00000621281 | ? | ENSP00000479064 | 5 |
| RP5-864K19.7 | downstream gene variant, upstream gene variant | 116 | ENST00000622355 | ? | ? | 9 |
| RRAGC | downstream gene variant, upstream gene variant | 488 | ENST00000373001 | NM_022157.3, NM_001271851.1 | ENSP00000362092 | 2 |
| RRAGC | downstream gene variant | 2028 | ENST00000474456 | ? | ? | 1 |
| RRAGC | downstream gene variant, upstream gene variant | 2447 | ENST00000493015 | ? | ? | 3 |
| RRAGC | downstream gene variant, upstream gene variant | 642 | ENST00000496778 | ? | ? | 4 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| GJA9 | ENST00000357771 | NM_030772.4 | ENSP00000350415 | 8 |
| MYCBP | ENST00000465771 | ? | ? | 3 |
| MYCBP | ENST00000494695 | ? | ENSP00000485717 | 3 |
| MYCBP | ENST00000397572 | NM_012333.4 | ENSP00000380702 | 5 |
| MYCBP | ENST00000495043 | ? | ? | 3 |
| RHBDL2 | ENST00000372990 | NM_017821.3, NM_001304746.1 | ENSP00000362081 | 32 |
| RHBDL2 | ENST00000289248 | ? | ENSP00000289248 | 32 |
| RP5-864K19.4 | ENST00000456813 | ? | ? | 7 |
| RP5-864K19.4 | ENST00000443161 | ? | ? | 3 |
| RP5-864K19.4 | ENST00000433671 | ? | ? | 50 |
| RP5-864K19.6 | ENST00000489575 | ? | ? | 14 |
| RP5-864K19.6 | ENST00000489803 | ? | ? | 14 |
| RP5-864K19.6 | ENST00000621281 | ? | ENSP00000479064 | 14 |
| RP5-864K19.6 | ENST00000454994 | ? | ENSP00000406846 | 13 |
| RP5-864K19.7 | ENST00000622355 | ? | ? | 13 |
| RRAGC | ENST00000373001 | NM_022157.3, NM_001271851.1 | ENSP00000362092 | 7 |
| RRAGC | ENST00000474456 | ? | ? | 6 |
| RRAGC | ENST00000493015 | ? | ? | 5 |

### 3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| RHBDL2 | ENST00000372990 | NM_017821.3, NM_001304746.1 | ENSP00000362081 | 1 |

| | | | | |
|---|---|---|---|---|
| RHBDL2 | ENST00000289248 | ? | ENSP00000289248 | 1 |

## 5'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| MYCBP | ENST00000397572 | NM_012333.4 | ENSP00000380702 | 2 |

## Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| RP5-864K19.6 | ENST00000489803 | ? | 3 |
| RP5-864K19.7 | ENST00000622355 | ? | 6 |
| RRAGC | ENST00000474456 | ? | 4 |