# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr20:24,899,905-24,977,586 |
| **block size** | 77,682 bp |
| **variant count** | 61 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -1.131 [-4.848 – 0.469] | **gene(s) hit or close-by** | APMAP, CST7, RNU6-1257P, RP4-568C11.4 |
| **phastCons** | μ = 0.048 [0 – 0.647] | **eQTL gene(s)** | APMAP, CST7, NINL |
| **GERP++** | μ = -0.368 [-8.11 – 1.91] | **potentially regulated gene(s)** | RP5-1025A1.2 |
| **CADD score** | μ = 2.517 [0.005 – 11.69] | **disease gene(s)** | – |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| APMAP | ? | ENSG00000101474 | pancreas | $1.50\times10^{-9}$ (p-value) | GTEx Portal V6 | 61 |
| APMAP | ? | ENSG00000101474 | testis | $1.36\times10^{-7}$ (p-value) | GTEx Portal V6 | 59 |
| APMAP | ? | ENSG00000101474 | transverse colon | $1.12\times10^{-7}$ (p-value) | GTEx Portal V6 | 50 |
| CST7 | ? | ENSG00000077984 | blood | $4.25\times10^{-19}$ (p-value) | GTEx Portal V6 | 61 |
| APMAP | ? | ENSG00000101474 | blood | $4.94\times10^{-7}$ (p-value) | GTEx Portal V6 | 8 |
| CST7 | ? | ENSG00000077984 | lung | $2.84\times10^{-7}$ (p-value) | GTEx Portal V6 | 17 |
| NINL | ? | ENSG00000101004 | subcutaneous adipocytes | $1.06\times10^{-5}$ (p-value) | GTEx Portal V6 | 1 |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000291173 | 1 | ENCP00000031115 | RP5-1025A1.2 |

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000399319 (CTCF binding site) | 2 | embryonic stem cell (H1ESC) | H3K27me3, Rad21, CTCF, DNase1 |
| | | HSMMtube | DNase1, H3K27me3, CTCF |
| | | blood (K562) | Rad21, CTCF |
| | | skin (NHDF-AD) | CTCF |
| | | muscle (HSMM) | CTCF |
| | | liver (HepG2) | DNase1, Rad21, CTCF, H3K27me3 |
| | | lung (IMR90) | H3K27me3, CTCF |
| | | blood (GM12878) | Rad21, CTCF |
| | | nervous (NH-A) | CTCF |
| | | skin (NHEK) | CTCF |
| | | NHLF | CTCF |
| | | Osteobl | H3K27me3, CTCF, H3K4me2 |
| | | blood (DND-41) | CTCF |
| | | breast (HMEC) | CTCF |

| | | | |
|---|---|---|---|
| | | cervix (HeLa-S3) | DNase1, CTCF |
| | | monocytes (Monocytes-CD14+) | CTCF |
| | | endothelium (HUVEC) | H3K36me3, CTCF, DNase1 |
| | | A549 | CTCF, H3K27me3 |
| ENSR00001664641 (CTCF binding site) | 3 | embryonic stem cell (H1ESC) | H3K27me3, DNase1 |
| | | HSMMtube | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | A549 | H3K27me3 |
| | | blood (DND-41) | H3K36me3 |
| ENSR00001664642 (enhancer) | 2 | cervix (HeLa-S3) | H3K27me3 |
| | | liver (HepG2) | H3K27me3 |
| | | HSMMtube | H3K27me3 |
| | | Osteobl | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |
| | | A549 | H3K27me3 |
| | | skin (NHEK) | H3K4me1, H3K4me2 |
| ENSR00001664643 (promoter flanking region) | 2 | embryonic stem cell (H1ESC) | CTCF, Rad21, DNase1 |
| | | HSMMtube | H3K27me3 |
| | | Osteobl | CTCF |
| | | blood (K562) | H3K27me3, CTCF |
| | | muscle (HSMM) | CTCF |
| | | breast (HMEC) | CTCF, H3K4me2, H3K27ac, DNase1 |
| | | cervix (HeLa-S3) | H3K27me3 |
| | | endothelium (HUVEC) | H3K36me3 |
| | | liver (HepG2) | CTCF, H3K27me3 |
| | | blood (GM12878) | CTCF |
| | | A549 | H3K27me3, CTCF |
| | | skin (NHEK) | CTCF, H3K4me2, H3K4me1, DNase1 |
| ENSR00000399320 (CTCF binding site) | 5 | embryonic stem cell (H1ESC) | Rad21, CTCF, DNase1 |
| | | HSMMtube | CTCF, H3K27me3 |
| | | blood (K562) | H3K27me3, CTCF |
| | | skin (NHDF-AD) | CTCF |
| | | muscle (HSMM) | CTCF |
| | | liver (HepG2) | Rad21, CTCF, H3K27me3 |
| | | blood (GM12878) | CTCF, Rad21 |
| | | lung (IMR90) | CTCF |
| | | nervous (NH-A) | CTCF |
| | | skin (NHEK) | CTCF, H3K4me2, H3K4me1, DNase1 |
| | | Osteobl | H3K27me3, CTCF |
| | | blood (DND-41) | H3K4me1, CTCF |
| | | breast (HMEC) | CTCF, H3K4me2, H3K27ac, DNase1 |
| | | cervix (HeLa-S3) | H3K27me3 |
| | | endothelium (HUVEC) | H3K36me3, CTCF |
| | | A549 | CTCF, H3K27me3 |
| ENSR00001448170 (enhancer) | 2 | HSMMtube | CTCF |
| | | Osteobl | CTCF |
| | | blood (DND-41) | CTCF, H3K4me1, H3K27ac |
| | | blood (K562) | CTCF, H3K27me3 |
| | | skin (NHDF-AD) | CTCF |
| | | muscle (HSMM) | DNase1 |
| | | breast (HMEC) | H3K4me2 |
| | | cervix (HeLa-S3) | H3K27me3 |
| | | liver (HepG2) | CTCF |
| | | blood (GM12878) | CTCF |
| | | A549 | H3K27me3, CTCF |
| | | skin (NHEK) | CTCF, H3K4me2 |
| ENSR00001448175 (promoter) | 1 | embryonic stem cell (H1ESC) | H3K27me3, PolII, Rad21, Gabp, TAF7, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1 |
| | | HSMMtube | H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1 |
| | | blood (K562) | HEY1, H2AZ, ELF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3, Max, H3K9ac, Gabp, Cmyc |
| | | skin (NHDF-AD) | H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF |
| | | muscle (HSMM) | DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ |
| | | liver (HepG2) | DNase1, Gabp, PolII, H3K79me2, Rad21, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K27ac, Cmyc, H3K4me3, H3K27me3 |
| | | lung (IMR90) | DNase1, H4K8ac, H3K27ac, H3K4me2, H4K5ac, H3K4me3, H3K9ac, CTCF |

| | | | |
|---|---|---|---|
| | | blood (GM12878) | H2AZ, DNase1, Cmyc, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, PolII, H3K27ac, H3K4me2, CTCF, H3K9ac |
| | | nervous (NH-A) | H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac |
| | | skin (NHEK) | CTCF, H3K9ac, H3K4me2, DNase1, H3K4me3 |
| | | NHLF | H3K27ac, H3K9ac, CTCF, H3K4me3, DNase1 |
| | | Osteobl | CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ |
| | | blood (DND-41) | H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1 |
| | | breast (HMEC) | CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1 |
| | | cervix (HeLa-S3) | H3K9ac, DNase1, PolII, Gabp, H3K4me3, TAF1, H3K27ac, CTCF, H3K4me2 |
| | | monocytes (Monocytes-CD14+) | H3K27ac, H3K9ac, H3K4me3, CTCF, DNase1 |
| | | endothelium (HUVEC) | Max, H3K4me3, H3K4me2, PolII, CTCF, DNase1 |
| | | A549 | H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1, H3K27ac |
| ENSR00000399325 (CTCF binding site) | 3 | embryonic stem cell (H1ESC) | Rad21, CTCF, DNase1 |
| | | HSMMtube | CTCF, H3K4me2, H3K27me3, DNase1 |
| | | blood (K562) | H3K27me3, Rad21, CTCF |
| | | skin (NHDF-AD) | H3K4me2, DNase1, H3K4me3, CTCF |
| | | muscle (HSMM) | H3K4me2, CTCF |
| | | liver (HepG2) | Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3, DNase1 |
| | | blood (GM12878) | H3K9ac, DNase1, Pbx3, H2AZ, Yy1, Rad21, H3K4me2, CTCF |
| | | lung (IMR90) | H4K8ac, H3K4me2, H4K5ac, H3K9ac, CTCF |
| | | nervous (NH-A) | H3K4me2, CTCF, H3K9ac |
| | | skin (NHEK) | CTCF, H3K4me3, H3K4me2, DNase1 |
| | | NHLF | CTCF |
| | | Osteobl | CTCF, H3K4me2, H3K27ac |
| | | blood (DND-41) | H3K9ac, CTCF, H3K4me2, H3K4me1, H3K4me3 |
| | | breast (HMEC) | H3K4me3, H3K27ac, H3K4me2, CTCF |
| | | cervix (HeLa-S3) | DNase1, CTCF, H3K4me2 |
| | | monocytes (Monocytes-CD14+) | H3K9ac, DNase1, CTCF, H3K27ac, H3K4me3 |
| | | endothelium (HUVEC) | H3K36me3, CTCF, H3K4me2, DNase1 |
| | | A549 | H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1, H3K27ac |
| ENSR00001664649 (promoter flanking region) | 1 | embryonic stem cell (H1ESC) | DNase1, CTCF, Rad21 |
| | | HSMMtube | CTCF, H3K27me3, DNase1 |
| | | blood (K562) | Rad21, CTCF, H3K27me3 |
| | | skin (NHDF-AD) | H3K4me2, DNase1, H3K4me3, CTCF |
| | | muscle (HSMM) | CTCF |
| | | liver (HepG2) | Rad21, H3K4me1, H3K4me2, CTCF, DNase1 |
| | | lung (IMR90) | H3K4me2, CTCF |
| | | blood (GM12878) | CTCF, Rad21, Yy1, Pbx3, DNase1 |
| | | nervous (NH-A) | H3K4me2, CTCF |
| | | skin (NHEK) | CTCF, DNase1 |
| | | NHLF | CTCF |
| | | Osteobl | CTCF, H3K4me2 |
| | | blood (DND-41) | CTCF |
| | | breast (HMEC) | CTCF |
| | | cervix (HeLa-S3) | DNase1, CTCF |
| | | monocytes (Monocytes-CD14+) | DNase1, CTCF |
| | | endothelium (HUVEC) | CTCF, DNase1 |
| | | A549 | H3K4me3, CTCF |

## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| APMAP | downstream gene variant, upstream gene variant | 154 | ENST00000217456 | NM_020531.2 | ENSP00000217456 | 10 |
| APMAP | downstream gene variant, upstream gene variant | 491 | ENST00000451442 | ? | ENSP00000395874 | 10 |
| CST7 | upstream gene variant, downstream gene variant | 32 | ENST00000480798 | NM_003650.3 | ENSP00000420384 | 2 |
| RNU6-1257P | downstream gene variant | 4470 | ENST00000384625 | ? | ? | 1 |
| RP4-568C11.4 | upstream gene variant, downstream gene variant | 34 | ENST00000620459 | ? | ? | 14 |

## Putative effect on transcript

## Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| APMAP | ENST00000217456 | NM_020531.2 | ENSP00000217456 | 2 |
| APMAP | ENST00000451442 | ? | ENSP00000395874 | 2 |
| CST7 | ENST00000480798 | NM_003650.3 | ENSP00000420384 | 2 |

## 3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| APMAP | ENST00000217456 | NM_020531.2 | ENSP00000217456 | 1 |
| APMAP | ENST00000451442 | ? | ENSP00000395874 | 1 |

## Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| RP4-568C11.4 | ENST00000620459 | ? | 3 |