# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr19:41,214,932-41,291,872 |
| **block size** | 76,941 bp |
| **variant count** | 30 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.573 [-3.842 – 1.533] | **gene(s) hit or close-by** | ADCK4, C19orf54, ITPKC, MIA, MIA-RAB4B, RAB4B, RAB4B-EGLN2, RNU6-195P, SNRPA |
| **phastCons** | μ = 0.034 [0 – 0.546] | **eQTL gene(s)** | C19orf54, MIA |
| **GERP++** | μ = -1.528 [-9.93 – 1.94] | **potentially regulated gene(s)** | C19orf54 |
| **CADD score** | μ = 5.053 [0.217 – 14.91] | **disease gene(s)** | ADCK4, ITPKC |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| ADCK4 | NEPHROTIC SYNDROME, TYPE 9 | OMIM | MIM:615573 |
| ITPKC | KAWASAKI DISEASE | OMIM | MIM:611775 |
| ADCK4 | Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis | OrphaNet | OrphaNet:93213 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| C19orf54 | ? | ENSG00000188493 | thyroid | 7.69×10-8 (p-value) | GTEx Portal V6 | 21 |
| MIA | ? | ENSG00000261857 | tibial nerve | 1.84×10-7 (p-value) | GTEx Portal V6 | 23 |
| C19orf54 | ? | ENSG00000188493 | blood | 2.44×10-8 (q-value) | SeeQTL DB (HapMap) | 8 |

## Putative effect on regulation

### Transcription factor binding site variation

| transcription factor | binding motif | motif position | highly informative position | score change | variant(s) |
|---|---|---|---|---|---|
| EGR1 | MA0162.2 | 5 | yes | 0.000 | 1 |

### FANTOM5 expressed promoter

| SNiPA promoter id | variant(s) | associated transcript(s) | gene |
|---|---|---|---|
| FFCP00000330481 | 1 | ENST00000470681, ENST00000469741 | C19orf54 |

### ENCODE promoter-associated DHS

| SNiPA promoter id | variant(s) | associated gene(s) |
|---|---|---|
| ENCP00000024392 | | |

## ENCODE promoter-associated distal DHS (Enhancer)

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000228995 | 1 | ENCP00000024393 | C19orf54 |

## Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001648159 (open chromatin region) | 1 | embryonic stem cell (H1ESC) | H3K36me3, DNase1 |
| | | blood (GM12878) | H3K36me3 |
| | | lung (IMR90) | H3K36me3 |
| | | A549 | H3K36me3 |
| | | skin (NHEK) | H3K36me3 |
| ENSR00000346640 (promoter) | 4 | embryonic stem cell (H1ESC) | H3K36me3, CTCF, PolII, p300, Sin3Ak20, H3K27ac, TAF1, H3K4me2, SP1, USF1, H3K9ac, H3K4me3, DNase1 |
| | | HSMMtube | H3K27ac, H3K4me2, DNase1, H2AZ, H3K4me3, H3K9ac, H3K79me2 |
| | | blood (K562) | H3K9ac, Gabp, H3K79me2, H2AZ, ELF1, Cfos, PolII, H3K4me2, H3K27me3, H3K36me3, DNase1, H3K4me3, Max, H3K27ac, Egr1 |
| | | skin (NHDF-AD) | H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac |
| | | muscle (HSMM) | H3K4me3, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K79me2, H2AZ |
| | | liver (HepG2) | Gabp, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H3K4me2, HNF4A, H3K9ac, H3K27ac, H3K4me3, H3K36me3, H3K27me3, DNase1 |
| | | blood (GM12878) | H3K27ac, H3K4me2, H3K9ac, H3K4me3, H3K79me2, Pbx3, Sin3Ak20, PolII, H2AZ, DNase1, Gabp, ELF1 |
| | | lung (IMR90) | DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac |
| | | nervous (NH-A) | DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac |
| | | skin (NHEK) | DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac |
| | | NHLF | DNase1, H3K4me3, H3K9ac, H3K27ac |
| | | Osteobl | H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3 |
| | | blood (DND-41) | H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1 |
| | | breast (HMEC) | DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2 |
| | | cervix (HeLa-S3) | DNase1, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me3, H3K79me2, Gabp, PolII |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3 |
| | | endothelium (HUVEC) | H3K36me3, Cjun, Max, H3K4me3, H3K4me2, H3K9ac, H3K4me1, H3K27ac, PolII, DNase1, Cmyc |
| | | A549 | H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac, H3K36me3 |
| ENSR00000346651 (promoter) | 5 | embryonic stem cell (H1ESC) | TAF1, H3K4me2, SP1, USF1, H3K9ac, H3K4me3, Yy1, ATF3, H4K5ac, H3K36me3, H3K27me3, PolII, TAF7, Sin3Ak20, H3K27ac, DNase1 |
| | | HSMMtube | H3K36me3, H3K4me2, H3K9ac, H3K27ac, DNase1, H2AZ, H3K4me3 |
| | | blood (K562) | Yy1, Egr1, H3K27ac, Nfe2, E2F6, H3K9ac, USF1, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Max, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3 |
| | | skin (NHDF-AD) | H3K36me3, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac |
| | | muscle (HSMM) | H2AZ, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2 |
| | | liver (HepG2) | H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K36me3, USF1, ELF1, Gabp, Yy1, PolII, H3K79me2, TAF1, DNase1 |
| | | blood (GM12878) | H3K36me3, H3K4me1, DNase1, Yy1, ELF1, H3K79me2, H3K4me3, USF1, H3K27ac, H4K20me1, Cfos, H3K4me2, H2AZ, PolII, Tr4, Cmyc, H3K9ac |
| | | lung (IMR90) | H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K4me2, H3K27ac, DNase1, H4K8ac, H3K79me2 |
| | | nervous (NH-A) | DNase1, H3K36me3, H3K27ac, H3K4me3, H3K4me2, H3K9ac |
| | | skin (NHEK) | DNase1, H3K36me3, H3K4me3, H3K4me2, PolII, H3K9ac, H3K27ac |
| | | NHLF | H3K36me3, H3K27ac, H3K9ac, H3K4me3, DNase1 |
| | | Osteobl | H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3 |
| | | blood (DND-41) | H3K36me3, H3K4me3, H3K9ac, H4K20me1, H3K27ac, H3K4me2, H3K4me1 |
| | | breast (HMEC) | H3K4me2, H3K4me1, H3K36me3, H3K9ac, H3K4me3, DNase1, H3K27ac |
| | | cervix (HeLa-S3) | DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me1, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, Cmyc, H3K36me3 |
| | | monocytes (Monocytes-CD14+) | H3K4me3, H3K36me3, H3K9ac, DNase1, H3K4me1, H3K27ac |
| | | endothelium (HUVEC) | H3K36me3, DNase1, Cmyc, PolII, H3K27ac, Max, H3K4me3, H3K9ac |
| | | A549 | H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3 |
| ENSR00001648164 (promoter) | 1 | Osteobl | H3K27me3 |
| | | breast (HMEC) | H3K27ac, H3K4me3, H3K9ac, H3K4me2 |

## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| ADCK4 | upstream gene variant | 1207 | ENST00000594084 | ? | ENSP00000473189 | 2 |
| ADCK4 | upstream gene variant | 1379 | ENST00000594490 | ? | ENSP00000471310 | 3 |

| | | | | | |
|---|---|---|---|---|---|
| ADCK4 | downstream gene variant, upstream gene variant | 943 | ENST00000596357 | ? | ENSP00000472925 4 |
| ADCK4 | upstream gene variant | 930 | ENST00000600080 | ? | ENSP00000473017 3 |
| ADCK4 | downstream gene variant, upstream gene variant | 401 | ENST00000600707 | ? | ENSP00000472978 5 |
| ADCK4 | upstream gene variant | 916 | ENST00000601967 | ? | ENSP00000470916 3 |
| ADCK4 | upstream gene variant | 2529 | ENST00000324464 | NM_024876.3 | ENSP00000315118 2 |
| ADCK4 | upstream gene variant | 2518 | ENST00000594720 | ? | ENSP00000470876 2 |
| ADCK4 | downstream gene variant, upstream gene variant | 398 | ENST00000593723 | ? | ENSP00000472775 5 |
| ADCK4 | upstream gene variant | 941 | ENST00000243583 | NM_001142555.2 | ENSP00000243583 3 |
| ADCK4 | upstream gene variant | 1229 | ENST00000601304 | ? | ENSP00000472519 3 |
| ADCK4 | upstream gene variant | 929 | ENST00000595254 | ? | ENSP00000470894 3 |
| ADCK4 | upstream gene variant | 908 | ENST00000593544 | ? | ? 3 |
| ADCK4 | upstream gene variant | 229 | ENST00000596455 | ? | ? 4 |
| ADCK4 | upstream gene variant | 47 | ENST00000599643 | ? | ENSP00000471192 4 |
| ADCK4 | upstream gene variant | 2529 | ENST00000601451 | ? | ? 2 |
| C19orf54 | upstream gene variant | 93 | ENST00000598352 | ? | ENSP00000473024 5 |
| C19orf54 | upstream gene variant | 22 | ENST00000597507 | ? | ENSP00000472022 3 |
| C19orf54 | downstream gene variant, upstream gene variant | 168 | ENST00000598485 | ? | ENSP00000469330 4 |
| C19orf54 | downstream gene variant, upstream gene variant | 649 | ENST00000470681 | ? | ENSP00000471114 3 |
| C19orf54 | downstream gene variant, upstream gene variant | 145 | ENST00000596940 | ? | ENSP00000472820 6 |
| C19orf54 | upstream gene variant | 104 | ENST00000600139 | ? | ENSP00000472948 5 |
| C19orf54 | downstream gene variant | 4111 | ENST00000594163 | ? | ? 1 |
| C19orf54 | downstream gene variant, upstream gene variant | 61 | ENST00000378313 | NM_198476.3 | ENSP00000367564 5 |
| C19orf54 | downstream gene variant | 4941 | ENST00000596809 | ? | ENSP00000469807 1 |
| C19orf54 | downstream gene variant, upstream gene variant | 588 | ENST00000469741 | ? | ENSP00000471589 3 |
| ITPKC | upstream gene variant | 952 | ENST00000263370 | NM_025194.2 | ENSP00000263370 3 |
| ITPKC | upstream gene variant | 106 | ENST00000597003 | ? | ENSP00000472655 3 |
| MIA | upstream gene variant, downstream gene variant | 190 | ENST00000593317 | ? | ? 7 |
| MIA | upstream gene variant, downstream gene variant | 349 | ENST00000601159 | ? | ENSP00000471439 5 |
| MIA | downstream gene variant | 3612 | ENST00000594436 | NM_001202553.1 | ENSP00000470129 4 |
| MIA | upstream gene variant, downstream gene variant | 346 | ENST00000597140 | ? | ENSP00000470641 5 |
| MIA | upstream gene variant, downstream gene variant | 176 | ENST00000263369 | NM_006533.3 | ENSP00000263369 5 |
| MIA | upstream gene variant, downstream gene variant | 63 | ENST00000597784 | ? | ENSP00000469499 5 |
| MIA | upstream gene variant, downstream gene variant | 34 | ENST00000597600 | ? | ENSP00000472982 3 |
| MIA-RAB4B | upstream gene variant | 1712 | ENST00000600729 | ? | ENSP00000472384 1 |
| RAB4B | upstream gene variant | 478 | ENST00000598430 | ? | ? 5 |
| RAB4B | upstream gene variant | 1615 | ENST00000357052 | NM_016154.4 | ENSP00000349560 3 |
| RAB4B | upstream gene variant | 1705 | ENST00000378307 | ? | ENSP00000367557 3 |
| RAB4B | upstream gene variant | 3781 | ENST00000595728 | ? | ENSP00000471803 2 |
| RAB4B | upstream gene variant, downstream gene variant | 1082 | ENST00000600078 | ? | ? 5 |
| RAB4B | upstream gene variant | 1774 | ENST00000602173 | ? | ENSP00000471504 3 |
| RAB4B | upstream gene variant | 1646 | ENST00000602069 | ? | ? 3 |

| | | | | | | |
|---|---|---|---|---|---|---|
| RAB4B | upstream gene variant | 1612 | ENST00000594800 | ? | ENSP00000470246 | 3 |
| RAB4B | upstream gene variant | 2443 | ENST00000597476 | ? | ? | 3 |
| RAB4B-EGLN2 | upstream gene variant | 3775 | ENST00000596216 | ? | ? | 2 |
| RAB4B-EGLN2 | upstream gene variant | 1638 | ENST00000601949 | ? | ? | 3 |
| RAB4B-EGLN2 | upstream gene variant | 1667 | ENST00000594136 | ? | ENSP00000469872 | 3 |
| RNU6-195P | upstream gene variant | 864 | ENST00000411352 | ? | ? | 2 |
| SNRPA | upstream gene variant, downstream gene variant | 405 | ENST00000599570 | ? | ? | 5 |
| SNRPA | downstream gene variant | 2669 | ENST00000598452 | ? | ? | 2 |
| SNRPA | upstream gene variant, downstream gene variant | 157 | ENST00000600456 | ? | ENSP00000471230 | 6 |
| SNRPA | upstream gene variant, downstream gene variant | 401 | ENST00000599362 | ? | ENSP00000472258 | 5 |
| SNRPA | upstream gene variant, downstream gene variant | 68 | ENST00000601393 | ? | ENSP00000472355 | 5 |
| SNRPA | upstream gene variant, downstream gene variant | 397 | ENST00000601253 | ? | ENSP00000469224 | 5 |
| SNRPA | upstream gene variant | 133 | ENST00000598923 | ? | ? | 4 |
| SNRPA | upstream gene variant | 398 | ENST00000597353 | ? | ENSP00000472449 | 3 |
| SNRPA | upstream gene variant, downstream gene variant | 167 | ENST00000601545 | ? | ENSP00000470534 | 4 |
| SNRPA | upstream gene variant, downstream gene variant | 389 | ENST00000243563 | NM_004596.4 | ENSP00000243563 | 4 |
| SNRPA | upstream gene variant, downstream gene variant | 891 | ENST00000596860 | ? | ? | 4 |

## Putative effect on transcript

### Synonymous coding variant

| gene | affected transcript | RefSeq id | protein | AA's | exchanged codons | variant(s) |
|---|---|---|---|---|---|---|
| C19orf54 | ENST00000378313 | NM_198476.3 | ENSP00000367564 | L | Ttg/Ctg | 1 |
| ITPKC | ENST00000263370 | NM_025194.2 | ENSP00000263370 | A | gcG/gcA | 1 |
| MIA | ENST00000597784 | ? | ENSP00000469499 | P | ccT/ccC | 1 |
| MIA | ENST00000263369 | NM_006533.3 | ENSP00000263369 | P | ccT/ccC | 1 |
| MIA | ENST00000594436 | NM_001202553.1 | ENSP00000470129 | P | ccT/ccC | 1 |
| MIA | ENST00000597140 | ? | ENSP00000470641 | P | ccT/ccC | 1 |
| MIA-RAB4B | ENST00000600729 | ? | ENSP00000472384 | P | ccT/ccC | 1 |

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| ADCK4 | ENST00000596357 | ? | ENSP00000472925 | 1 |
| ADCK4 | ENST00000324464 | NM_024876.3 | ENSP00000315118 | 4 |
| ADCK4 | ENST00000601451 | ? | ? | 4 |
| ADCK4 | ENST00000594490 | ? | ENSP00000471310 | 2 |
| ADCK4 | ENST00000601304 | ? | ENSP00000472519 | 2 |
| ADCK4 | ENST00000596455 | ? | ? | 2 |
| ADCK4 | ENST00000593544 | ? | ? | 2 |
| ADCK4 | ENST00000600080 | ? | ENSP00000473017 | 2 |
| ADCK4 | ENST00000599643 | ? | ENSP00000471192 | 2 |
| ADCK4 | ENST00000595254 | ? | ENSP00000470894 | 2 |
| ADCK4 | ENST00000594720 | ? | ENSP00000470876 | 4 |

| | | | | |
|---|---|---|---|---|
| ADCK4 | ENST00000601967 | ? | ENSP00000470916 | 2 |
| ADCK4 | ENST00000243583 | NM_001142555.2 | ENSP00000243583 | 2 |
| ADCK4 | ENST00000600707 | ? | ENSP00000472978 | 1 |
| ADCK4 | ENST00000594084 | ? | ENSP00000473189 | 4 |
| C19orf54 | ENST00000596809 | ? | ENSP00000469807 | 5 |
| C19orf54 | ENST00000470681 | ? | ENSP00000471114 | 3 |
| C19orf54 | ENST00000597507 | ? | ENSP00000472022 | 2 |
| C19orf54 | ENST00000598485 | ? | ENSP00000469330 | 2 |
| C19orf54 | ENST00000469741 | ? | ENSP00000471589 | 3 |
| ITPKC | ENST00000597003 | ? | ENSP00000472655 | 3 |
| ITPKC | ENST00000263370 | NM_025194.2 | ENSP00000263370 | 7 |
| MIA | ENST00000597784 | ? | ENSP00000469499 | 2 |
| MIA | ENST00000263369 | NM_006533.3 | ENSP00000263369 | 2 |
| MIA | ENST00000601159 | ? | ENSP00000471439 | 2 |
| MIA | ENST00000594436 | NM_001202553.1 | ENSP00000470129 | 3 |
| MIA | ENST00000597140 | ? | ENSP00000470641 | 2 |
| MIA | ENST00000597600 | ? | ENSP00000472982 | 1 |
| MIA-RAB4B | ENST00000600729 | ? | ENSP00000472384 | 9 |
| RAB4B | ENST00000600078 | ? | ? | 4 |
| RAB4B | ENST00000602173 | ? | ENSP00000471504 | 6 |
| RAB4B | ENST00000597476 | ? | ? | 6 |
| RAB4B | ENST00000602069 | ? | ? | 6 |
| RAB4B | ENST00000378307 | ? | ENSP00000367557 | 6 |
| RAB4B | ENST00000594800 | ? | ENSP00000470246 | 6 |
| RAB4B | ENST00000357052 | NM_016154.4 | ENSP00000349560 | 6 |
| RAB4B | ENST00000595728 | ? | ENSP00000471803 | 6 |
| RAB4B-EGLN2 | ENST00000596216 | ? | ? | 6 |
| RAB4B-EGLN2 | ENST00000594136 | ? | ENSP00000469872 | 6 |
| RAB4B-EGLN2 | ENST00000601949 | ? | ? | 6 |
| SNRPA | ENST00000600456 | ? | ENSP00000471230 | 2 |
| SNRPA | ENST00000598923 | ? | ? | 4 |
| SNRPA | ENST00000601545 | ? | ENSP00000470534 | 5 |
| SNRPA | ENST00000599570 | ? | ? | 3 |
| SNRPA | ENST00000601393 | ? | ENSP00000472355 | 4 |
| SNRPA | ENST00000601253 | ? | ENSP00000469224 | 3 |
| SNRPA | ENST00000243563 | NM_004596.4 | ENSP00000243563 | 5 |
| SNRPA | ENST00000597353 | ? | ENSP00000472449 | 5 |
| SNRPA | ENST00000598452 | ? | ? | 1 |
| SNRPA | ENST00000599362 | ? | ENSP00000472258 | 3 |

**5'-UTR variant**

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| C19orf54 | ENST00000596809 | ? | ENSP00000469807 | 3 |
| C19orf54 | ENST00000470681 | ? | ENSP00000471114 | 1 |
| C19orf54 | ENST00000469741 | ? | ENSP00000471589 | 1 |
| MIA | ENST00000594436 | NM_001202553.1 | ENSP00000470129 | 1 |
| SNRPA | ENST00000243563 | NM_004596.4 | ENSP00000243563 | 2 |

## Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| RAB4B | ENST00000598430 | ? | 1 |
| SNRPA | ENST00000599570 | ? | 1 |