# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr11:64,876,740-64,936,719 |
| **block size** | 59,980 bp |
| **variant count** | 11 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.540 [-6.639 – 1.312] | **gene(s) hit or close-by** | AP003068.18, AP003068.9, HIGD1AP10, MIR6751, MRPL49, PDCL2P2, RNU2-23P, SPDYC, SYVN1, TM7SF2, VPS51 |
| **phastCons** | μ = 0.041 [0 – 0.105] | **eQTL gene(s)** | ARL2, NAALADL1, SPDYC |
| **GERP++** | μ = -1.022 [-10.4 – 0.632] | **potentially regulated gene(s)** | CDCA5, MEN1, MEN1, SAC3D1, SIPA1, SYVN1 |
| **CADD score** | μ = 4.642 [0.032 – 12.66] | **disease gene(s)** | MEN1 |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| MEN1 | MULTIPLE ENDOCRINE NEOPLASIA, TYPE I | OMIM | MIM:131100 |
| MEN1 | HYPERPARATHYROIDISM 1 | OMIM | MIM:145000 |
| MEN1 | Zollinger-Ellison syndrome | OrphaNet | OrphaNet:913 |
| MEN1 | Familial parathyroid adenoma | OrphaNet | OrphaNet:99877 |
| MEN1 | Multiple endocrine neoplasia, type 1 | OrphaNet | OrphaNet:652 |
| MEN1 | Familial isolated hyperparathyroidism | OrphaNet | OrphaNet:99879 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| SPDYC | ? | ENSG00000204710 | lung | $2.92\times10^{-6}$ (p-value) | GTEx Portal V6 | 10 |
| SPDYC | ? | ENSG00000204710 | blood | $2.22\times10^{-14}$ (p-value) | GTEx Portal V6 | 11 |
| NAALADL1 | ENST00000526516 | ILMN_1770963 | blood | $9.11\times10^{-6}$ (p-value) | Westra et al. | 7 |
| NAALADL1 | ENST00000532802 | | | | | |
| NAALADL1 | ENST00000528977 | | | | | |
| NAALADL1 | ENST00000533340 | | | | | |
| NAALADL1 | ENST00000531746 | | | | | |
| NAALADL1 | ENST00000356632 | | | | | |
| NAALADL1 | ENST00000534568 | | | | | |
| NAALADL1 | ENST00000530995 | | | | | |
| NAALADL1 | ENST00000533842 | | | | | |
| NAALADL1 | ENST00000532450 | | | | | |
| NAALADL1 | ENST00000533753 | | | | | |
| NAALADL1 | ENST00000358658 | | | | | |
| NAALADL1 | ENST00000355721 | | | | | |
| NAALADL1 | ENST00000340252 | | | | | |
| NAALADL1 | ENST00000529274 | | | | | |
| ARL2 | ENST00000529254 | ILMN_1787879 | blood | $2.01\times10^{-5}$ (p-value) | Westra et al. | 7 |

| | | | | | | |
|---|---|---|---|---|---|---|
| ARL2 | ENST00000533729 | | | | | |
| ARL2 | ENST00000529384 | | | | | |
| ARL2 | ENST00000246747 | | | | | |
| ? | ? | ILMN_1777476 | monocyte | 2.43×10-22 (p-value) | Zeller et al. | 2 |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000109797 | 1 | ENCP00000012907 | CDCA5 |
| | | ENCP00000012927 | SYVN1 |
| | | ENCP00000013012 | SIPA1 |
| | | ENCP00000012901 | SAC3D1 |
| | | ENCP00000012872 | MEN1<br>MEN1 |

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000563760 (open chromatin region) | 1 | blood (GM12878) | DNase1 |
| ENSR00000563763 (promoter flanking region) | 1 | endothelium (HUVEC) | DNase1, PolII, H3K27ac, Max, Cjun |
| | | liver (HepG2) | H2AZ |
| | | HSMMtube | H2AZ |
| | | lung (IMR90) | H4K5ac |
| | | blood (GM12878) | PolII, H2AZ |
| | | blood (K562) | Max, H3K9ac, H2AZ, H3K4me3, PolII, H3K4me2, H3K27ac, DNase1 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| AP003068.18 | downstream gene variant | 4869 | ENST00000534819 | ? | ? | 1 |
| AP003068.9 | downstream gene variant | 1446 | ENST00000528887 | ? | ? | 1 |
| HIGD1AP10 | upstream gene variant | 1104 | ENST00000527837 | ? | ? | 1 |
| MIR6751 | upstream gene variant | 776 | ENST00000610479 | ? | ? | 1 |
| MRPL49 | downstream gene variant | 3504 | ENST00000531705 | ? | ENSP00000436740 | 1 |
| MRPL49 | downstream gene variant | 3391 | ENST00000532671 | ? | ? | 1 |
| MRPL49 | downstream gene variant | 3391 | ENST00000526319 | ? | ENSP00000434190 | 1 |
| MRPL49 | downstream gene variant | 4576 | ENST00000533943 | ? | ENSP00000434052 | 1 |
| MRPL49 | downstream gene variant | 3383 | ENST00000279242 | NM_004927.3 | ENSP00000279242 | 1 |
| MRPL49 | downstream gene variant | 4452 | ENST00000528529 | ? | ? | 1 |
| MRPL49 | downstream gene variant | 3398 | ENST00000524482 | ? | ? | 1 |
| MRPL49 | downstream gene variant | 4750 | ENST00000534078 | ? | ENSP00000434222 | 1 |
| PDCL2P2 | downstream gene variant, upstream gene variant | 274 | ENST00000533651 | ? | ? | 5 |
| RNU2-23P | upstream gene variant | 508 | ENST00000410545 | ? | ? | 1 |
| SPDYC | upstream gene variant | 906 | ENST00000377185 | NM_001008778.1 | ENSP00000366390 | 1 |
| SYVN1 | upstream gene variant | 768 | ENST00000530451 | ? | ? | 1 |
| SYVN1 | upstream gene variant | 2950 | ENST00000449943 | ? | ? | 1 |
| SYVN1 | downstream gene variant, upstream gene variant | 1873 | ENST00000525874 | ? | ? | 2 |
| SYVN1 | downstream gene variant, upstream gene variant | 1941 | ENST00000533685 | ? | ? | 2 |

| | | | | | | |
|---|---|---|---|---|---|---|
| SYVN1 | upstream gene variant | 2905 | ENST00000294256 | NM_032431.2 | ENSP00000294256 | 1 |
| SYVN1 | downstream gene variant, upstream gene variant | 241 | ENST00000532771 | ? | ENSP00000432166 | 2 |
| SYVN1 | upstream gene variant | 2904 | ENST00000377190 | NM_172230.2 | ENSP00000366395 | 1 |
| SYVN1 | downstream gene variant, upstream gene variant | 217 | ENST00000531018 | ? | ENSP00000431215 | 2 |
| SYVN1 | upstream gene variant | 2957 | ENST00000307289 | ? | ENSP00000302035 | 1 |
| SYVN1 | downstream gene variant, upstream gene variant | 520 | ENST00000528487 | ? | ENSP00000431720 | 2 |
| SYVN1 | upstream gene variant | 3008 | ENST00000526060 | ? | ENSP00000436984 | 1 |
| SYVN1 | upstream gene variant | 465 | ENST00000527765 | ? | ? | 1 |
| SYVN1 | downstream gene variant, upstream gene variant | 378 | ENST00000529207 | ? | ? | 2 |
| SYVN1 | downstream gene variant, upstream gene variant | 378 | ENST00000527142 | ? | ENSP00000432888 | 2 |
| TM7SF2 | upstream gene variant | 3003 | ENST00000530892 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 2610 | ENST00000529601 | ? | ENSP00000435458 | 1 |
| TM7SF2 | upstream gene variant | 2634 | ENST00000530650 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 2780 | ENST00000524690 | ? | ENSP00000434598 | 1 |
| TM7SF2 | upstream gene variant | 2730 | ENST00000530750 | ? | ENSP00000432413 | 1 |
| TM7SF2 | upstream gene variant | 2577 | ENST00000526809 | ? | ENSP00000432171 | 1 |
| TM7SF2 | upstream gene variant | 4241 | ENST00000528802 | ? | ENSP00000436233 | 1 |
| TM7SF2 | upstream gene variant | 2604 | ENST00000534371 | ? | ENSP00000432187 | 1 |
| TM7SF2 | upstream gene variant | 3946 | ENST00000534667 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 2616 | ENST00000345348 | NM_001277233.1 | ENSP00000329520 | 1 |
| TM7SF2 | upstream gene variant | 4282 | ENST00000526048 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 2622 | ENST00000529414 | ? | ENSP00000433275 | 1 |
| TM7SF2 | upstream gene variant | 3471 | ENST00000527851 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 4228 | ENST00000527968 | ? | ENSP00000431685 | 1 |
| TM7SF2 | upstream gene variant | 2618 | ENST00000531321 | ? | ENSP00000431300 | 1 |
| TM7SF2 | upstream gene variant | 2611 | ENST00000525385 | ? | ENSP00000433325 | 1 |
| TM7SF2 | upstream gene variant | 2621 | ENST00000529292 | ? | ENSP00000436370 | 1 |
| TM7SF2 | upstream gene variant | 2599 | ENST00000524986 | ? | ENSP00000435972 | 1 |
| TM7SF2 | upstream gene variant | 2607 | ENST00000533646 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 3437 | ENST00000529233 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 2604 | ENST00000528026 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 4103 | ENST00000532328 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 2623 | ENST00000526085 | ? | ENSP00000434447 | 1 |
| TM7SF2 | upstream gene variant | 2586 | ENST00000279263 | NM_003273.3 | ENSP00000279263 | 1 |
| TM7SF2 | upstream gene variant | 3376 | ENST00000531029 | ? | ? | 1 |
| TM7SF2 | upstream gene variant | 2604 | ENST00000533766 | ? | ? | 1 |
| VPS51 | upstream gene variant | 50 | ENST00000534591 | ? | ? | 1 |
| VPS51 | downstream gene variant | 918 | ENST00000534124 | ? | ENSP00000436867 | 1 |
| VPS51 | downstream gene variant | 869 | ENST00000529180 | ? | ENSP00000435245 | 1 |
| VPS51 | downstream gene variant | 774 | ENST00000534557 | ? | ENSP00000435691 | 1 |
| VPS51 | downstream gene variant | 949 | ENST00000530773 | ? | ENSP00000434138 | 1 |

| | | | | | | |
|---|---|---|---|---|---|---|
| VPS51 | downstream gene variant | 1640 | ENST00000528588 | ? | ENSP00000436134 | 1 |
| VPS51 | upstream gene variant | 23 | ENST00000533827 | ? | ENSP00000434542 | 1 |
| VPS51 | downstream gene variant | 1383 | ENST00000526578 | ? | ENSP00000431188 | 1 |
| VPS51 | upstream gene variant | 1214 | ENST00000530673 | ? | ENSP00000431317 | 1 |
| VPS51 | downstream gene variant | 1541 | ENST00000533656 | ? | ? | 1 |
| VPS51 | downstream gene variant | 1248 | ENST00000533487 | ? | ENSP00000433579 | 1 |
| VPS51 | upstream gene variant | 76 | ENST00000526856 | ? | ENSP00000435629 | 1 |

## Putative effect on transcript

### Synonymous coding variant

| gene | affected transcript | RefSeq id | protein | AA's | exchanged codons | variant(s) |
|---|---|---|---|---|---|---|
| SYVN1 | ENST00000526060 | ? | ENSP00000436984 | S | tcA/tcG | 1 |
| SYVN1 | ENST00000377190 | NM_172230.2 | ENSP00000366395 | S | tcA/tcG | 1 |
| SYVN1 | ENST00000294256 | NM_032431.2 | ENSP00000294256 | S | tcA/tcG | 1 |
| SYVN1 | ENST00000307289 | ? | ENSP00000302035 | S | tcA/tcG | 1 |

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| VPS51 | ENST00000279281 | NM_013265.3 | ENSP00000279281 | 1 |
| VPS51 | ENST00000527646 | ? | ? | 1 |

### Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| SYVN1 | ENST00000526121 | ? | 1 |
| SYVN1 | ENST00000449943 | ? | 1 |
| VPS51 | ENST00000531146 | ? | 1 |