# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr16:71,465,613-71,506,983 |
| **block size** | 41,371 bp |
| **variant count** | 6 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -1.134 [-3.461 – 0.421] | **gene(s) hit or close-by** | AC010547.9, RP11-510M2.4, RP11-510M2.8, ZNF19 |
| **phastCons** | μ = 0.002 [0 – 0.01] | **eQTL gene(s)** | AC010547.9, MARVELD3, ZNF23 |
| **GERP++** | μ = -1.694 [-9.66 – 1.52] | **potentially regulated gene(s)** | – |
| **CADD score** | μ = 1.501 [0.025 – 4.134] | **disease gene(s)** | – |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| MARVELD3 | ? | ENSG00000140832 | sun exposed skin | $6.69\times10^{-8}$ (p-value) | GTEx Portal V6 | 6 |
| MARVELD3 | ? | ENSG00000140832 | thyroid | $1.68\times10^{-8}$ (p-value) | GTEx Portal V6 | 6 |
| ZNF23 | ENST00000358700 | ILMN_1720027 | blood | $2.93\times10^{-6}$ (p-value) | Westra et al. | 2 |
| ZNF23 | ENST00000539742 | | | | | |
| ZNF23 | ENST00000428724 | | | | | |
| AC010547.9 | ENST00000561908 | | | | | |
| ZNF23 | ENST00000357254 | | | | | |
| ZNF23 | ENST00000393539 | | | | | |
| ZNF23 | ENST00000576258 | | | | | |
| ZNF23 | ENST00000564528 | | | | | |
| ZNF23 | ENST00000497160 | | | | | |

## Putative effect on regulation

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| RP11-510M2.8 | downstream gene variant | 3122 | ENST00000571789 | ? | ? | 3 |
| ZNF19 | downstream gene variant | 3083 | ENST00000565541 | ? | ENSP00000454549 | 2 |
| ZNF19 | downstream gene variant | 1304 | ENST00000565100 | ? | ENSP00000462810 | 2 |
| ZNF19 | downstream gene variant | 503 | ENST00000288177 | NM_006961.3 | ENSP00000288177 | 2 |
| ZNF19 | downstream gene variant | 1669 | ENST00000569717 | ? | ENSP00000456143 | 2 |
| ZNF19 | downstream gene variant | 1299 | ENST00000565637 | ? | ENSP00000454864 | 2 |
| ZNF19 | downstream gene variant | 3114 | ENST00000568815 | ? | ENSP00000455796 | 2 |
| ZNF19 | downstream gene variant | 1730 | ENST00000564230 | ? | ENSP00000458105 | 2 |
| ZNF19 | downstream gene variant | 4806 | ENST00000561469 | ? | ENSP00000462021 | 1 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| AC010547.9 | ENST00000561908 | ? | ENSP00000463741 | 2 |
| RP11-510M2.4 | ENST00000561754 | ? | ? | 1 |
| RP11-510M2.4 | ENST00000572770 | ? | ? | 1 |
| ZNF19 | ENST00000567225 | ? | ENSP00000462566 | 2 |