# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr21:28,280,322-28,329,010 |
| **block size** | 48,689 bp |
| **variant count** | 8 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = 0.713 [-1.94 – 7.487] | **gene(s) hit or close-by** | ADAMTS5, AP001601.2, MIR4759 |
| **phastCons** | μ = 0.127 [0 – 1] | **eQTL gene(s)** | – |
| **GERP++** | μ = 0.144 [-6.12 – 6.03] | **potentially regulated gene(s)** | – |
| **CADD score** | μ = 7.436 [1.089 – 22.1] | **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) |
|---|---|---|---|---|---|---|---|---|---|
| ADAMTS5 | missense variant | ENST00000284987 | NM_007038.3 | ENSP00000284987 | H/R | cAc/cGc | ? | ? | 1 |

## Putative effect on regulation

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000612277 (promoter flanking region) | 1 | embryonic stem cell (H1ESC) | DNase1 |
| | | HSMMtube | DNase1, H3K36me3 |
| | | skin (NHDF-AD) | DNase1 |
| | | muscle (HSMM) | DNase1, H3K36me3 |
| | | cervix (HeLa-S3) | DNase1, Cjun, Jund, Max |
| | | monocytes (Monocytes-CD14+) | H3K27me3 |
| | | lung (IMR90) | DNase1 |
| | | nervous (NH-A) | DNase1 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| MIR4759 | upstream gene variant, downstream gene variant | 4 | ENST00000584048 | ? | ? | 2 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| ADAMTS5 | ENST00000284987 | NM_007038.3 | ENSP00000284987 | 6 |
| AP001601.2 | ENST00000426771 | ? | ? | 6 |

### 3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| ADAMTS5 | ENST00000284987 | NM_007038.3 | ENSP00000284987 | 2 |