# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr4:15,749,703-15,756,270 |
| **block size** | 6,568 bp |
| **variant count** | 4 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.788 [-2.591 – 0.219] | **gene(s) hit or close-by** | RP11-442P12.2 |
| **phastCons** | μ = 0.005 [0 – 0.019] | **eQTL gene(s)** | BST1, CD38, RP11-115L11.1 |
| **GERP++** | μ = 0.212 [-2.99 – 2.39] | **potentially regulated gene(s)** | – |
| **CADD score** | μ = 3.061 [2.042 – 4.102] | **disease gene(s)** | – |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| CD38 | ENST00000502843 | ILMN_2233783 | adipocyte | $9.04\times10^{-6}$ (p-value) | MuTHER consortium | 4 |
| CD38 | ENST00000226279 | | | | | |
| RP11-115L11.1 | ? | ENSG00000214846 | transformed fibroblasts | $3.48\times10^{-7}$ (p-value) | GTEx Portal V6 | 4 |
| CD38 | ? | ENSG00000004468 | thyroid | $6.54\times10^{-6}$ (p-value) | GTEx Portal V6 | 2 |
| BST1 | ENST00000382346 | ILMN_1770161 | monocyte | $8.49\times10^{-5}$ (p-value) | Fairfax et al. | 1 |
| BST1 | ENST00000265016 | | | | | |

## Putative effect on regulation

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001237252 (promoter flanking region) | 2 | NHLF | DNase1 |
| | | monocytes (Monocytes-CD14+) | H3K36me3 |
| | | endothelium (HUVEC) | Cjun, DNase1 |
| | | HSMMtube | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | nervous (NH-A) | H3K27ac, DNase1 |
| | | blood (DND-41) | H3K4me1, H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF |
| ENSR00001683638 (CTCF binding site) | 1 | NHLF | DNase1 |
| | | monocytes (Monocytes-CD14+) | H3K36me3 |
| | | endothelium (HUVEC) | Cjun, DNase1 |
| | | blood (K562) | H3K27me3 |
| | | blood (DND-41) | H3K27ac, CTCF, H3K4me1, H3K9ac, H3K4me3 |
| | | nervous (NH-A) | H3K27ac, DNase1 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| RP11-442P12.2 | downstream gene variant | 4498 | ENST00000513715 | ? | ? | 1 |