# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr5:96,057,882-96,070,205 |
| **block size** | 12,324 bp |
| **variant count** | 5 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.582 [-3.14 – 0.626] | **gene(s) hit or close-by** | CAST |
| **phastCons** | μ = 0.011 [0 – 0.051] | **eQTL gene(s)** | CAST, CTC-506B8.1, CTD-2260A17.1, ERAP1 |
| **GERP++** | μ = 0.491 [0.225 – 1.08] | **potentially regulated gene(s)** | – |
| **CADD score** | μ = 4.473 [2.207 – 8.12] | **disease gene(s)** | – |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| ERAP1 | ? | ENSG00000164307 | pancreas | $1.38\times10^{-7}$ (p-value) | GTEx Portal V6 | 1 |
| CTC-506B8.1 | ? | ENSG00000249180 | muscularis mucosae | $2.46\times10^{-9}$ (p-value) | GTEx Portal V6 | 5 |
| CAST | ? | ENSG00000153113 | muscularis mucosae | $2.49\times10^{-10}$ (p-value) | GTEx Portal V6 | 5 |
| ERAP1 | ? | ENSG00000164307 | lung | $1.19\times10^{-7}$ (p-value) | GTEx Portal V6 | 1 |
| CTC-506B8.1 | ? | ENSG00000249180 | lung | $6.30\times10^{-8}$ (p-value) | GTEx Portal V6 | 3 |
| CAST | ? | ENSG00000153113 | lung | $1.78\times10^{-10}$ (p-value) | GTEx Portal V6 | 5 |
| ERAP1 | ? | ENSG00000164307 | tibial nerve | $1.85\times10^{-5}$ (p-value) | GTEx Portal V6 | 1 |
| CTC-506B8.1 | ? | ENSG00000249180 | tibial nerve | $8.33\times10^{-9}$ (p-value) | GTEx Portal V6 | 5 |
| CAST | ? | ENSG00000153113 | tibial nerve | $7.19\times10^{-11}$ (p-value) | GTEx Portal V6 | 5 |
| ERAP1 | ? | ENSG00000164307 | transformed fibroblasts | $6.20\times10^{-7}$ (p-value) | GTEx Portal V6 | 1 |
| CTC-506B8.1 | ? | ENSG00000249180 | transformed fibroblasts | $1.87\times10^{-8}$ (p-value) | GTEx Portal V6 | 5 |
| CAST | ? | ENSG00000153113 | transformed fibroblasts | $1.87\times10^{-10}$ (p-value) | GTEx Portal V6 | 5 |
| ERAP1 | ? | ENSG00000164307 | blood | $6.95\times10^{-15}$ (p-value) | GTEx Portal V6 | 5 |
| CTC-506B8.1 | ? | ENSG00000249180 | blood | $6.38\times10^{-10}$ (p-value) | GTEx Portal V6 | 5 |
| CAST | ? | ENSG00000153113 | blood | $2.32\times10^{-12}$ (p-value) | GTEx Portal V6 | 5 |
| CTC-506B8.1 | ? | ENSG00000249180 | tibial artery | $2.72\times10^{-10}$ (p-value) | GTEx Portal V6 | 5 |
| CAST | ? | ENSG00000153113 | tibial artery | $4.85\times10^{-8}$ (p-value) | GTEx Portal V6 | 5 |
| ERAP1 | ? | ENSG00000164307 | esophagus mucosa | $9.80\times10^{-7}$ (p-value) | GTEx Portal V6 | 2 |
| CTC-506B8.1 | ? | ENSG00000249180 | thyroid | $5.78\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| CAST | ? | ENSG00000153113 | thyroid | $4.83\times10^{-7}$ (p-value) | GTEx Portal V6 | 2 |
| ERAP1 | ? | ENSG00000164307 | skeletal muscle | $6.41\times10^{-15}$ (p-value) | GTEx Portal V6 | 5 |
| CTD-2260A17.1 | ? | ENSG00000248734 | skeletal muscle | $1.45\times10^{-7}$ (p-value) | GTEx Portal V6 | 5 |

| | | | | | | |
|---|---|---|---|---|---|---|
| ERAP1 | ? | ENSG00000164307 | EBV lymphocytes | 2.27×10-6 (p-value) | GTEx Portal V6 | 1 |
| ERAP1 | ? | ENSG00000164307 | transverse colon | 2.42×10-6 (p-value) | GTEx Portal V6 | 1 |
| CTC-506B8.1 | ? | ENSG00000249180 | aorta | 1.98×10-6 (p-value) | GTEx Portal V6 | 5 |
| ERAP1 | ? | ENSG00000164307 | subcutaneous adipocytes | 2.15×10-6 (p-value) | GTEx Portal V6 | 1 |
| CTC-506B8.1 | ? | ENSG00000249180 | subcutaneous adipocytes | 1.11×10-7 (p-value) | GTEx Portal V6 | 1 |
| CAST | ? | ENSG00000153113 | subcutaneous adipocytes | 9.10×10-7 (p-value) | GTEx Portal V6 | 1 |
| CTD-2260A17.1 | ? | ENSG00000248734 | testis | 5.12×10-7 (p-value) | GTEx Portal V6 | 4 |
| CAST | ? | ENSG00000153113 | adrenal gland | 3.31×10-6 (p-value) | GTEx Portal V6 | 1 |
| ERAP1 | ENST00000296754 | ILMN_1752145 | blood | 4.72×10-9 (p-value) | MuTHER consortium | 1 |
| ERAP1 | ENST00000443439 | ILMN_2336220 | monocyte | 3.15×10-23 (p-value) | Zeller et al. | 1 |
| ERAP1 | ENST00000514604 | | | | | |
| ERAP1 | ENST00000296754 | | | | | |

## Putative effect on regulation

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001285973 (promoter flanking region) | 1 | NHLF | DNase1, H3K36me3 |
| | | embryonic stem cell (H1ESC) | SP1, DNase1 |
| | | blood (K562) | DNase1 |
| | | skin (NHDF-AD) | H3K36me3, DNase1 |
| | | breast (HMEC) | H3K4me2, H3K27ac, DNase1 |
| | | muscle (HSMM) | H3K36me3 |
| | | cervix (HeLa-S3) | H3K36me3 |
| | | monocytes (Monocytes-CD14+) | H3K36me3, H4K20me1 |
| | | lung (IMR90) | H3K36me3 |
| | | blood (GM12878) | H3K36me3 |
| | | nervous (NH-A) | H3K36me3, DNase1 |
| | | A549 | H3K36me3 |
| | | skin (NHEK) | DNase1, H3K36me3, H3K4me2, H3K27ac, H3K4me1 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| CAST | upstream gene variant | 3751 | ENST00000348386 | ? | ? | 1 |
| CAST | downstream gene variant | 2214 | ENST00000514845 | ? | ENSP00000422612 | 2 |
| CAST | upstream gene variant, downstream gene variant | 3360 | ENST00000504532 | ? | ? | 3 |
| CAST | downstream gene variant | 3372 | ENST00000506811 | ? | ENSP00000427092 | 2 |
| CAST | upstream gene variant | 1675 | ENST00000515063 | ? | ? | 2 |
| CAST | downstream gene variant | 3338 | ENST00000514055 | ? | ENSP00000421230 | 2 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| CAST | ENST00000510156 | ? | ENSP00000422325 | 5 |
| CAST | ENST00000395813 | ? | ENSP00000379158 | 5 |
| CAST | ENST00000514055 | ? | ENSP00000421230 | 3 |

| | | | | |
|---|---|---|---|---|
| CAST | ENST00000514055 | ? | ENSP00000421230 | 3 |
| CAST | ENST00000511049 | ? | ENSP00000421130 | 5 |
| CAST | ENST00000309190 | NM_001284212.1, NM_173060.3 | ENSP00000312523 | 5 |
| CAST | ENST00000421689 | ? | ENSP00000396558 | 5 |
| CAST | ENST00000511097 | ? | ENSP00000422951 | 5 |
| CAST | ENST00000506811 | ? | ENSP00000427092 | 3 |
| CAST | ENST00000509903 | ? | ENSP00000426946 | 5 |
| CAST | ENST00000512620 | ? | ENSP00000426422 | 5 |
| CAST | ENST00000338252 | NM_001190442.1 | ENSP00000343421 | 5 |
| CAST | ENST00000508117 | ? | ENSP00000422466 | 5 |
| CAST | ENST00000508608 | ? | ENSP00000422677 | 5 |
| CAST | ENST00000508830 | ? | ENSP00000425721 | 5 |
| CAST | ENST00000511782 | ? | ENSP00000423638 | 5 |
| CAST | ENST00000513666 | ? | ? | 5 |
| CAST | ENST00000507487 | ? | ? | 3 |
| CAST | ENST00000505143 | ? | ENSP00000422957 | 5 |
| CAST | ENST00000395812 | NM_001042440.3 | ENSP00000379157 | 5 |
| CAST | ENST00000514845 | ? | ENSP00000422612 | 3 |
| CAST | ENST00000341926 | ? | ENSP00000339914 | 5 |
| CAST | ENST00000508197 | ? | ENSP00000422831 | 5 |
| CAST | ENST00000504465 | NM_001284213.1 | ENSP00000425670 | 5 |
| CAST | ENST00000510756 | ? | ENSP00000422176 | 5 |
| CAST | ENST00000348386 | ? | ? | 2 |