# Locus 346 (rs6669284)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr1:206,885,839-206,904,464 |
| block size | 18,626 bp |
| variant count | 7 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.577 [-2.101 – 0.376] | gene(s) hit or close-by | MAPKAPK2 |
| phastCons | μ = 0.001 [0 – 0.006] | eQTL gene(s) | IL19 |
| GERP++ | μ = -0.975 [-4.84 – 2.73] | potentially regulated gene(s) | IKBKE, IL19 |
| CADD score | μ = 4.853 [0.834 – 12.73] | disease gene(s) | – |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| IL19 | ENST00000620365 | ILMN_1799575 | blood | 2.78×10-6 (p-value) | MuTHER consortium | 5 |
| IL19 | ENST00000270218 | | | | | |
| IL19 | ENST00000340758 | | | | | |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000052952 | 1 | ENCP00000006333 | IKBKE |
| | | ENCP00000006352 | IL19 |

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001040819 (promoter flanking region) | 1 | NHLF | H3K27ac |
| | | embryonic stem cell (H1ESC) | DNase1 |
| | | HSMMtube | H3K79me2 |
| | | Osteobl | H3K36me3, H3K27ac |
| | | blood (K562) | H3K79me2, H3K4me1, H3K36me3 |
| | | skin (NHDF-AD) | DNase1 |
| | | muscle (HSMM) | H3K79me2 |
| | | cervix (HeLa-S3) | Jund, DNase1 |
| | | monocytes (Monocytes-CD14+) | DNase1, H4K20me1 |
| | | lung (IMR90) | DNase1, H4K20me1, H3K79me2 |
| | | blood (GM12878) | H3K79me2, H3K27ac |
| | | A549 | H3K4me3, H3K4me2, DNase1, H3K27ac |
| ENSR00000551719 (promoter flanking region) | 2 | HSMMtube | H3K79me2 |
| | | blood (K562) | DNase1, H3K79me2, H3K4me1 |
| | | muscle (HSMM) | H3K79me2 |
| | | monocytes (Monocytes-CD14+) | H4K20me1 |
| | | liver (HepG2) | H3K36me3 |
| | | lung (IMR90) | H4K20me1, H3K79me2 |
| | | blood (GM12878) | Srf, DNase1, H3K79me2, H3K27ac |
| | | skin (NHEK) | H3K36me3 |
| ENSR00000672024 | 1 | embryonic stem cell (H1ESC) | Rad21, CTCF, Tcf12, DNase1 |
| (CTCF binding site) | | HSMMtube | DNase1, CTCF |

| | |
|---|---|
| blood (K562) | DNase1, H3K27me3, Rad21, H4K20me1, CTCF |
| skin (NHDF-AD) | CTCF |
| muscle (HSMM) | H3K79me2, CTCF, H4K20me1, DNase1 |
| liver (HepG2) | CTCF |
| lung (IMR90) | H4K20me1, H3K36me3, CTCF |
| blood (GM12878) | Pax5, CTCF, H3K79me2, Rad21, DNase1, H3K4me1 |
| nervous (NH-A) | H4K20me1, CTCF |
| skin (NHEK) | CTCF, H3K36me3 |
| NHLF | CTCF |
| Osteobl | H4K20me1, CTCF |
| blood (DND-41) | CTCF, H3K36me3 |
| breast (HMEC) | CTCF |
| cervix (HeLa-S3) | CTCF, DNase1, H3K4me2, H3K4me1 |
| monocytes (Monocytes-CD14+) | CTCF, H4K20me1, H3K36me3 |
| endothelium (HUVEC) | H3K36me3, CTCF |
| A549 | H3K4me2, CTCF, DNase1, H3K27ac |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| MAPKAPK2 | upstream gene variant, downstream gene variant | 897 | ENST00000493447 | ? | ? | 4 |
| MAPKAPK2 | upstream gene variant | 169 | ENST00000479009 | ? | ? | 3 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| MAPKAPK2 | ENST00000294981 | NM_004759.4 | ENSP00000294981 | 7 |
| MAPKAPK2 | ENST00000367103 | NM_032960.3 | ENSP00000356070 | 7 |