# Locus 65 (rs692804)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr11:120,098,044-120,100,867 |
| block size | 2,824 bp |
| variant count | 8 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.208 [-2.697 – 2.393] | gene(s) hit or close-by | OAF |
| phastCons | μ = 0.129 [0 – 1] | eQTL gene(s) | POU2F3 |
| GERP++ | μ = -0.794 [-5.98 – 5.26] | potentially regulated gene(s) | ARHGEF12, CTD-2523D13.2, OAF, POU2F3, PVRL1, TMEM136, TRIM29 |
| CADD score | μ = 4.439 [0.537 – 15.33] | disease gene(s) | PVRL1 |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| PVRL1 | CLEFT LIP/PALATE-ECTODERMAL DYSPLASIA SYNDROME | OMIM | MIM:225060 |
| PVRL1 | CLEFT LIP/PALATE-ECTODERMAL DYSPLASIA SYNDROME | DECIPHER | MIM:225060 |
| PVRL1 | Zlotogora-Ogur syndrome | OrphaNet | OrphaNet:3253 |

## 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) |
|---|---|---|---|---|---|---|---|---|---|
| OAF | missense variant | ENST00000328965 | NM_178507.2 | ENSP00000332613 | H/R | cAc/cGc | tolerated | benign | 1 |
| OAF | missense variant | ENST00000531220 | ? | ENSP00000431865 | H/R | cAc/cGc | tolerated | benign | 1 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| POU2F3 | ? | ENSG00000137709 | subcutaneous adipocytes | $6.18\times10^{-8}$ (p-value) | GTEx Portal V6 | 8 |

## Putative effect on regulation

### FANTOM5 expressed promoter

| SNiPA promoter id | variant(s) | associated transcript(s) | gene |
|---|---|---|---|
| FFCP00000047292 | 1 | ENST00000328965, ENST00000531220 | OAF |

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|

| | | | |
|---|---|---|---|
| ENCE00000125097 | 1 | ENCP00000015007 | CTD-2523D13.2<br>PVRL1 |
| | | ENCP00000015012 | TRIM29 |
| | | ENCP00000015027 | OAF |
| | | ENCP00000015028 | POU2F3 |
| | | ENCP00000015032 | TMEM136 |
| | | ENCP00000015034 | ARHGEF12 |

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000112413<br>(CTCF binding site) | 3 | embryonic stem cell (H1ESC) | H3K27me3, Rad21, CTCF, DNase1 |
| | | HSMMtube | CTCF, H3K27me3 |
| | | blood (K562) | H3K27me3, Rad21, CTCF |
| | | skin (NHDF-AD) | CTCF |
| | | muscle (HSMM) | CTCF |
| | | liver (HepG2) | DNase1, H3K36me3, PolII, Rad21, CTCF |
| | | blood (GM12878) | CTCF, Rad21 |
| | | lung (IMR90) | CTCF, H3K36me3 |
| | | nervous (NH-A) | CTCF |
| | | skin (NHEK) | H3K36me3, CTCF |
| | | NHLF | CTCF |
| | | Osteobl | CTCF |
| | | blood (DND-41) | CTCF |
| | | breast (HMEC) | CTCF |
| | | cervix (HeLa-S3) | CTCF |
| | | monocytes (Monocytes-CD14+) | CTCF, H3K36me3 |
| | | endothelium (HUVEC) | H3K36me3, CTCF |
| | | A549 | H3K27me3, CTCF |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| OAF | downstream gene variant | 571 | ENST00000531220 | ? | ENSP00000431865 | 3 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| OAF | ENST00000328965 | NM_178507.2 | ENSP00000332613 | 4 |
| OAF | ENST00000531220 | ? | ENSP00000431865 | 4 |

### 3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| OAF | ENST00000328965 | NM_178507.2 | ENSP00000332613 | 3 |