# Locus 98 (rs6695321)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr1:196,675,861-196,675,861 |
| block size | 1 bp |
| variant count | 1 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | 0.57 | gene(s) hit or close-by | CFH |
| phastCons | 0.001 | eQTL gene(s) | CFHR3 |
| GERP++ | 0.628 | potentially regulated gene(s) | – |
| CADD score | 2.058 | disease gene(s) | CFHR3, CFH |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| CFHR3 | Atypical hemolytic-uremic syndrome with anti-factor H antibodies | OrphaNet | OrphaNet:93581 |
| CFH | Immunodeficiency with factor H anomaly | OrphaNet | OrphaNet:200421 |
| CFH | Atypical hemolytic uremic syndrome with H factor anomaly | OrphaNet | OrphaNet:93579 |
| CFH | Dense deposit disease | OrphaNet | OrphaNet:93571 |
| CFH | Familial drusen | OrphaNet | OrphaNet:75376 |
| CFH | Immunoglobulin-mediated membranoproliferative glomerulonephritis | OrphaNet | OrphaNet:329903 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| CFHR3 | ? | ENSG00000116785 | atrial appendage | $7.18\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| CFHR3 | ? | ENSG00000116785 | tibial nerve | $7.49\times10^{-10}$ (p-value) | GTEx Portal V6 | 1 |
| CFHR3 | ? | ENSG00000116785 | aorta | $6.75\times10^{-7}$ (p-value) | GTEx Portal V6 | 1 |
| CFHR3 | ? | ENSG00000116785 | tibial artery | $3.96\times10^{-9}$ (p-value) | GTEx Portal V6 | 1 |
| CFHR3 | ? | ENSG00000116785 | subcutaneous adipocytes | $2.39\times10^{-7}$ (p-value) | GTEx Portal V6 | 1 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| CFH | ENST00000466229 | ? | ? | 1 |
| CFH | ENST00000367429 | NM_000186.3 | ENSP00000356399 | 1 |