# SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| **genomic range** | chr1:196,845,273-196,944,357 |
| **block size** | 99,085 bp |
| **variant count** | 18 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = 0.006 [-1.277 – 0.754] | **gene(s) hit or close-by** | CFHR2, CFHR2, CFHR4, CFHR5, RP4-608O15.4 |
| **phastCons** | μ = 0.095 [0 – 0.492] | **eQTL gene(s)** | CFH, CFHR1, CFHR3, CFHR4 |
| **GERP++** | μ = -0.192 [-3.63 – 1.43] | **potentially regulated gene(s)** | – |
| **CADD score** | μ = 4.973 [0.282 – 13.66] | **disease gene(s)** | CFHR1, CFHR3, CFH, CFHR5 |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | variant(s) |
|---|---|---|---|---|
| End-stage coagulation | $<7.00\times10^{-19}$ | GWAS Catalog | 23381943 | 1 |
| Macular degeneration | $5.49\times10^{-30}$ | dbGaP | pha002890 | 1 |
| Macular degeneration | $5.91\times10^{-34}$ | dbGaP | pha002869 | 1 |
| Macular degeneration | $6.21\times10^{-5}$ | dbGaP | pha002856 | 1 |
| Macular degeneration | $2.51\times10^{-6}$ | dbGaP | pha000001 | 2 |

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| CFHR1 | Atypical hemolytic-uremic syndrome with anti-factor H antibodies | OrphaNet | OrphaNet:93581 |
| CFHR1 | C3 glomerulonephritis | OrphaNet | OrphaNet:329931 |
| CFHR1 | Dense deposit disease | OrphaNet | OrphaNet:93571 |
| CFHR3 | Atypical hemolytic-uremic syndrome with anti-factor H antibodies | OrphaNet | OrphaNet:93581 |
| CFH | Atypical hemolytic uremic syndrome with H factor anomaly | OrphaNet | OrphaNet:93579 |
| CFH | Immunodeficiency with factor H anomaly | OrphaNet | OrphaNet:200421 |
| CFH | Dense deposit disease | OrphaNet | OrphaNet:93571 |
| CFH | Familial drusen | OrphaNet | OrphaNet:75376 |
| CFH | Immunoglobulin-mediated membranoproliferative glomerulonephritis | OrphaNet | OrphaNet:329903 |
| CFHR5 | Atypical hemolytic-uremic syndrome with anti-factor H antibodies | OrphaNet | OrphaNet:93581 |
| CFHR5 | C3 glomerulonephritis | OrphaNet | OrphaNet:329931 |
| CFHR5 | CFHR5 DEFICIENCY | OMIM | MIM:614809 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| CFHR3 | ? | ENSG00000116785 | lung | $4.99\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| CFHR1 | ? | ENSG00000244414 | tibial nerve | $1.40\times10^{-6}$ (p-value) | GTEx Portal V6 | 1 |
| CFHR3 | ? | ENSG00000116785 | tibial nerve | $9.52\times10^{-11}$ (p-value) | GTEx Portal V6 | 12 |
| CFH | ? | ENSG00000000971 | tibial nerve | $2.60\times10^{-6}$ (p-value) | GTEx Portal V6 | 13 |

| | | | | | | |
|---|---|---|---|---|---|---|
| CFHR4 | ? | ENSG00000134365 | liver | 6.84×10-8 (p-value) | GTEx Portal V6 | 16 |
| CFHR1 | ? | ENSG00000244414 | subcutaneous adipocytes | 1.65×10-8 (p-value) | GTEx Portal V6 | 16 |
| CFHR3 | ? | ENSG00000116785 | subcutaneous adipocytes | 7.04×10-11 (p-value) | GTEx Portal V6 | 16 |
| CFHR3 | ? | ENSG00000116785 | thyroid | 4.81×10-6 (p-value) | GTEx Portal V6 | 1 |

## Putative effect on regulation

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| CFHR2 | downstream gene variant | 965 | ENST00000473386 | ? | ? | 1 |
| CFHR2 | downstream gene variant | 970 | ENST00000367421 | ? | ENSP00000356391 | 1 |
| CFHR2 | downstream gene variant | 954 | ENST00000476712 | ? | ENSP00000476677 | 1 |
| CFHR2 | downstream gene variant | 79 | ENST00000489703 | ? | ? | 3 |
| CFHR2 | downstream gene variant | 954 | ENST00000367415 | NM_005666.2 | ENSP00000356385 | 1 |
| CFHR2 | downstream gene variant | 142 | ENST00000485647 | ? | ? | 2 |
| CFHR2 | downstream gene variant | 961 | ENST00000496448 | ? | ? | 1 |
| CFHR4 | upstream gene variant | 1523 | ENST00000251424 | NM_006684.4 | ENSP00000251424 | 1 |
| CFHR4 | upstream gene variant | 1455 | ENST00000367416 | NM_001201551.1, NM_001201550.2 | ENSP00000356386 | 1 |
| CFHR4 | upstream gene variant | 1544 | ENST00000367418 | ? | ENSP00000356388 | 1 |
| CFHR5 | upstream gene variant | 2329 | ENST00000256785 | NM_030787.3 | ENSP00000256785 | 1 |
| RP4-608O15.4 | downstream gene variant | 4556 | ENST00000613613 | ? | ? | 1 |

## Putative effect on transcript

### Synonymous coding variant

| gene | affected transcript | RefSeq id | protein | AA's | exchanged codons | variant(s) |
|---|---|---|---|---|---|---|
| CFHR2 | ENST00000367421 | ? | ENSP00000356391 | C | tgT/tgC | 1 |
| CFHR2 | ENST00000367415 | NM_005666.2 | ENSP00000356385 | C | tgT/tgC | 1 |
| CFHR2 | ENST00000476712 | ? | ENSP00000476677 | C | tgT/tgC | 1 |

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| CFHR2 | ENST00000473386 | ? | ? | 4 |
| CFHR2 | ENST00000367421 | ? | ENSP00000356391 | 16 |
| CFHR2 | ENST00000485647 | ? | ? | 2 |
| CFHR2 | ENST00000367415 | NM_005666.2 | ENSP00000356385 | 4 |
| CFHR2 | ENST00000489703 | ? | ? | 1 |
| CFHR2 | ENST00000476712 | ? | ENSP00000476677 | 4 |
| CFHR2 | ENST00000496448 | ? | ? | 4 |
| CFHR4 | ENST00000251424 | NM_006684.4 | ENSP00000251424 | 6 |
| CFHR4 | ENST00000608469 | ? | ENSP00000477162 | 8 |

| | | | | |
|---|---|---|---|---|
| CFHR4 | ENST00000367418 | ? | ENSP00000356388 | 6 |
| CFHR4 | ENST00000367416 | NM_001201551.1, NM_001201550.2 | ENSP00000356386 | 6 |

## Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| CFHR2 | ENST00000485647 | ? | 1 |
| CFHR2 | ENST00000496448 | ? | 1 |