# Locus 64 (rs7522061)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr1:157,661,848-157,693,722 |
| block size | 31,875 bp |
| variant count | 13 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.795 [-3.247 – 0.025] | gene(s) hit or close-by | FCRL3, RP11-367J7.3, VDAC1P9 |
| phastCons | μ = 0.062 [0 – 0.694] | eQTL gene(s) | ATP1A4, FCRL1, FCRL3, RP11-367J7.3 |
| GERP++ | μ = -1.674 [-10.7 – 1.7] | potentially regulated gene(s) | – |
| CADD score | μ = 2.100 [0.524 – 4.934] | disease gene(s) | FCRL3 |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | variant(s) |
|---|---|---|---|---|
| Graves' disease | $<2.00\times10^{-13}$ | GWAS Catalog | 21841780 | 1 |
| Multiple sclerosis | $<3.00\times10^{-6}$ | GWAS Catalog | 21833088 | 1 |
| Type 1 diabetes autoantibodies | $<1.00\times10^{-11}$ | GWAS Catalog | 21829393 | 1 |
| Alkaline phosphatase | $8.18\times10^{-5}$ | dbGaP | pha001181 | 1 |
| Alkaline phosphatase | $3.25\times10^{-5}$ | dbGaP | pha001179 | 1 |

### Variant annotation

| trait | type | source DB | source entry/link | Variant(s) |
|---|---|---|---|---|
| ? | HGMD curated | HGMD | CR067135 | 1 |
| ? | HGMD curated | HGMD | CR067134 | 1 |
| ? | HGMD curated | HGMD | CR052070 | 1 |

### Disease gene annotation

| gene | trait | source DB | source entry/link | |
|---|---|---|---|---|
| FCRL3 | RHEUMATOID ARTHRITIS | OMIM | MIM:180300 | |

## 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) |
|---|---|---|---|---|---|---|---|---|---|
| FCRL3 | missense variant | ENST00000485028 | ? | ENSP00000434331 | N/D | Aat/Gat | tolerated | benign | 1 |
| FCRL3 | missense variant | ENST00000492769 | ? | ENSP00000435487 | N/D | Aat/Gat | tolerated | benign | 1 |
| FCRL3 | missense variant | ENST00000496769 | ? | ENSP00000473680 | N/D | Aat/Gat | tolerated | benign | 1 |
| FCRL3 | missense variant | ENST00000368186 | ? | ENSP00000357169 | N/D | Aat/Gat | tolerated | benign | 1 |
| FCRL3 | missense variant | ENST00000477837 | ? | ENSP00000433430 | N/D | Aat/Gat | tolerated | benign | 1 |
| FCRL3 | missense variant | ENST00000368184 | NM_052939.3 | ENSP00000357167 | N/D | Aat/Gat | tolerated | benign | 1 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| FCRL3 | ENST00000494724 | ILMN_1691693 | blood | $1.12\times10^{-20}$ (p-value) | MuTHER consortium | 8 |
| FCRL3 | ENST00000485028 | | | | | |
| FCRL3 | ENST00000368186 | | | | | |
| FCRL3 | ENST00000477837 | | | | | |
| FCRL3 | ENST00000492769 | | | | | |
| FCRL3 | ENST00000368184 | | | | | |
| FCRL3 | ENST00000480682 | | | | | |
| FCRL3 | ENST00000494724 | ILMN_1699599 | blood | $9.83\times10^{-16}$ (p-value) | MuTHER consortium | 8 |
| FCRL3 | ENST00000485028 | | | | | |
| FCRL3 | ENST00000477837 | | | | | |
| FCRL3 | ENST00000473231 | | | | | |
| FCRL3 | ENST00000468507 | | | | | |
| FCRL3 | ENST00000480682 | | | | | |
| FCRL3 | ENST00000492769 | | | | | |
| FCRL3 | ENST00000368184 | | | | | |
| FCRL3 | ENST00000473231 | ILMN_1797428 | blood | $9.84\times10^{-17}$ (p-value) | MuTHER consortium | 8 |
| FCRL3 | ENST00000368184 | | | | | |
| FCRL3 | ENST00000468507 | | | | | |
| FCRL3 | ? | ENSG00000160856 | spleen | $1.70\times10^{-8}$ (p-value) | GTEx Portal V6 | 13 |
| RP11-367J7.3 | ? | ENSG00000227217 | spleen | $9.09\times10^{-8}$ (p-value) | GTEx Portal V6 | 5 |
| FCRL3 | ? | ENSG00000160856 | lung | $2.74\times10^{-9}$ (p-value) | GTEx Portal V6 | 13 |
| RP11-367J7.3 | ? | ENSG00000227217 | lung | $5.02\times10^{-8}$ (p-value) | GTEx Portal V6 | 11 |
| RP11-367J7.3 | ? | ENSG00000227217 | blood | $7.86\times10^{-13}$ (p-value) | GTEx Portal V6 | 13 |
| FCRL3 | ? | ENSG00000160856 | blood | $2.00\times10^{-19}$ (p-value) | GTEx Portal V6 | 13 |
| FCRL1 | ? | ENSG00000163534 | blood | $8.70\times10^{-7}$ (p-value) | GTEx Portal V6 | 4 |
| FCRL3 | ? | ENSG00000160856 | esophagus mucosa | $5.44\times10^{-9}$ (p-value) | GTEx Portal V6 | 9 |
| ATP1A4 | ENST00000470705 | ILMN_1705208 | b-cell | $1.09\times10^{-4}$ (p-value) | Fairfax et al. | 3 |
| ATP1A4 | ENST00000466526 | | | | | |
| FCRL3 | ENST00000494724 | ILMN_1691693 | monocyte | $1.27\times10^{-6}$ (p-value) | Fairfax et al. | 3 |
| FCRL3 | ENST00000485028 | | b-cell | $7.43\times10^{-33}$ (p-value) | Fairfax et al. | 3 |
| FCRL3 | ENST00000477837 | | | | | |
| FCRL3 | ENST00000368186 | | | | | |
| FCRL3 | ENST00000492769 | | | | | |
| FCRL3 | ENST00000368184 | | | | | |
| FCRL3 | ENST00000480682 | | | | | |
| FCRL3 | ENST00000494724 | ILMN_1699599 | b-cell | $2.37\times10^{-30}$ (p-value) | Fairfax et al. | 2 |
| FCRL3 | ENST00000485028 | | | | | |
| FCRL3 | ENST00000477837 | | | | | |
| FCRL3 | ENST00000473231 | | | | | |
| FCRL3 | ENST00000480682 | | | | | |
| FCRL3 | ENST00000468507 | | | | | |
| FCRL3 | ENST00000492769 | | | | | |
| FCRL3 | ENST00000368184 | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| FCRL3 | ENST00000368184 | ILMN_1797428 | monocyte | $1.02\times10^{-4}$ (p-value) | Fairfax et al. | 3 |
| FCRL3 | ENST00000473231 | | b-cell | $5.93\times10^{-26}$ (p-value) | Fairfax et al. | 3 |
| FCRL3 | ENST00000468507 | | | | | |
| FCRL3 | ? | ENSG00000160856 | subcutaneous adipocytes | $5.58\times10^{-8}$ (p-value) | GTEx Portal V6 | 7 |

### *trans*-eQTL

| gene | transcript | probe | chromosome | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|---|
| FCRL3 | ENST00000457799 | 231093_at | chr1 | blood | $1.00\times10^{-10}$ (p-value) | Dixon et al. | 3 |
| FCRL3 | ENST00000368184 | | | | | | |

## Putative effect on regulation

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000671247 (CTCF binding site) | 1 | embryonic stem cell (H1ESC) | CTCF, Rad21 |
| | | HSMMtube | CTCF, H3K27me3 |
| | | blood (K562) | CTCF |
| | | skin (NHDF-AD) | CTCF |
| | | muscle (HSMM) | CTCF |
| | | liver (HepG2) | CTCF |
| | | lung (IMR90) | CTCF |
| | | blood (GM12878) | Rad21, CTCF |
| | | nervous (NH-A) | CTCF |
| | | skin (NHEK) | CTCF |
| | | Osteobl | CTCF |
| | | blood (DND-41) | CTCF, H3K4me1 |
| | | breast (HMEC) | CTCF |
| | | cervix (HeLa-S3) | CTCF |
| | | monocytes (Monocytes-CD14+) | H3K27me3 |
| | | endothelium (HUVEC) | H3K36me3, CTCF |
| | | A549 | CTCF, H3K27me3 |
| ENSR00000545969 (open chromatin region) | 1 | embryonic stem cell (H1ESC) | DNase1 |
| | | A549 | H3K27me3 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| FCRL3 | upstream gene variant | 283 | ENST00000485028 | ? | ENSP00000434331 | 1 |
| FCRL3 | downstream gene variant, upstream gene variant | 314 | ENST00000478179 | ? | ? | 7 |
| FCRL3 | upstream gene variant | 296 | ENST00000494724 | ? | ? | 1 |
| FCRL3 | downstream gene variant, upstream gene variant | 281 | ENST00000496769 | ? | ENSP00000473680 | 2 |
| FCRL3 | upstream gene variant | 587 | ENST00000473231 | ? | ? | 6 |
| FCRL3 | upstream gene variant | 2168 | ENST00000468507 | ? | ? | 1 |
| FCRL3 | upstream gene variant | 288 | ENST00000368186 | ? | ENSP00000357169 | 1 |
| FCRL3 | upstream gene variant | 296 | ENST00000477837 | ? | ENSP00000433430 | 1 |
| FCRL3 | upstream gene variant | 296 | ENST00000480682 | ? | ? | 1 |
| FCRL3 | upstream gene variant | 296 | ENST00000492769 | ? | ENSP00000435487 | 1 |
| FCRL3 | upstream gene variant | 169 | ENST00000368184 | NM_052939.3 | ENSP00000357167 | 1 |
| RP11-367J7.3 | downstream gene variant | 395 | ENST00000453692 | ? | ? | 7 |
| VDAC1P9 | downstream gene variant | 248 | ENST00000412766 | ? | ? | 5 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| FCRL3 | ENST00000477837 | ? | ENSP00000433430 | 5 |
| FCRL3 | ENST00000485028 | ? | ENSP00000434331 | 5 |
| FCRL3 | ENST00000480682 | ? | ? | 5 |
| FCRL3 | ENST00000368186 | ? | ENSP00000357169 | 5 |
| FCRL3 | ENST00000494724 | ? | ? | 5 |
| FCRL3 | ENST00000473231 | ? | ? | 2 |
| FCRL3 | ENST00000492769 | ? | ENSP00000435487 | 5 |
| FCRL3 | ENST00000368184 | NM_052939.3 | ENSP00000357167 | 5 |
| FCRL3 | ENST00000496769 | ? | ENSP00000473680 | 3 |
| RP11-367J7.3 | ENST00000453692 | ? | ? | 1 |

### 5'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| FCRL3 | ENST00000477837 | ? | ENSP00000433430 | 1 |
| FCRL3 | ENST00000485028 | ? | ENSP00000434331 | 1 |
| FCRL3 | ENST00000368186 | ? | ENSP00000357169 | 1 |
| FCRL3 | ENST00000492769 | ? | ENSP00000435487 | 1 |
| FCRL3 | ENST00000368184 | NM_052939.3 | ENSP00000357167 | 1 |
| FCRL3 | ENST00000496769 | ? | ENSP00000473680 | 1 |

### Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| FCRL3 | ENST00000480682 | ? | 3 |
| FCRL3 | ENST00000494724 | ? | 3 |
| FCRL3 | ENST00000473231 | ? | 1 |