# Locus 200 (rs550513)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr6:31,905,130-31,928,306 |
| block size | 23,177 bp |
| variant count | 13 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.055 [-3.331 – 5.881] | gene(s) hit or close-by | C2, C2-AS1, CFB, MIR1236, NELFE, SKIV2L, XXbac-BPG116M5.17 |
| phastCons | μ = 0.127 [0 – 1] | eQTL gene(s) | NELFE, NEU1, PRRT1, SKIV2L |
| GERP++ | μ = -0.978 [-10.3 – 5.14] | potentially regulated gene(s) | CFB, CFB, CFB, CFB, CFB, CFB, CFB, CFB, SKIV2L, SKIV2L, SKIV2L, SKIV2L, SKIV2L, SKIV2L |
| CADD score | μ = 5.009 [0.293 – 22.3] | disease gene(s) | SKIV2L, NEU1, , , , , , , C2, CFB, , , , , , |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | | variant(s) |
|---|---|---|---|---|---|
| Age-related macular degeneration | $<2.00\times10^{-10}$ | GWAS Catalog | 23577725 | | 1 |
| Age-related macular degeneration (extreme sampling) | $<3.00\times10^{-7}$ | GWAS Catalog | 23577725 | | 1 |
| Age-related macular degeneration | $<9.00\times10^{-17}$ | GWAS Catalog | 22694956 | | 1 |
| Macular degeneration | $2.70\times10^{-18}$ | dbGaP | pha002890 | dbGaP | 5 |
| Macular degeneration | $2.90\times10^{-19}$ | dbGaP | pha002869 | dbGaP | 5 |
| Macular degeneration | $8.34\times10^{-5}$ | dbGaP | pha002856 | dbGaP | 1 |
| Macular degeneration | $1.38\times10^{-5}$ | dbGaP | pha000001 | dbGaP | 1 |

### Variant annotation

| trait | type | source DB | source entry/link | | Variant(s) |
|---|---|---|---|---|---|
| Age-related macular degeneration 14 | protective | ClinVar | RCV000012914.4 | ClinVar | 1 |
| ? | HGMD curated | HGMD | CM093386 | HGMD | 1 |
| MACULAR DEGENERATION, AGE-RELATED, 14, REDUCED RISK OF | OMIM curated | OMIM | MIM:613927 | OMIM | 1 |

### Disease gene annotation

| gene | trait | source DB | source entry/link | |
|---|---|---|---|---|
| SKIV2L | TRICHOHEPATOENTERIC SYNDROME 2 | DECIPHER | MIM:615602 | OMIM |
| NEU1 | sialidosis (SIALIDOSIS) | DECIPHER | MIM:256550 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | DECIPHER | MIM:615602 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | DECIPHER | MIM:615602 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | DECIPHER | MIM:615602 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | DECIPHER | MIM:615602 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | DECIPHER | MIM:615602 | OMIM |
| SKIV2L | TRICHOHEPATOENTERIC SYNDROME 2 | OMIM | MIM:614602 | OMIM |
| C2 | COMPLEMENT COMPONENT 2 DEFICIENCY | OMIM | MIM:217000 | OMIM |
| C2 | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| NEU1 | NEURAMINIDASE DEFICIENCY | OMIM | MIM:256550 | OMIM |
| CFB | COMPLEMENT FACTOR B DEFICIENCY | OMIM | MIM:615561 | OMIM |
| CFB | HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4 | OMIM | MIM:612924 | OMIM |

| | | | | |
|---|---|---|---|---|
| CFB | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | COMPLEMENT FACTOR B DEFICIENCY | OMIM | MIM:615561 | OMIM |
| | HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4 | OMIM | MIM:612924 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | COMPLEMENT FACTOR B DEFICIENCY | OMIM | MIM:615561 | OMIM |
| | HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4 | OMIM | MIM:612924 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | COMPLEMENT FACTOR B DEFICIENCY | OMIM | MIM:615561 | OMIM |
| | HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4 | OMIM | MIM:612924 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | COMPLEMENT FACTOR B DEFICIENCY | OMIM | MIM:615561 | OMIM |
| | HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4 | OMIM | MIM:612924 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | COMPLEMENT FACTOR B DEFICIENCY | OMIM | MIM:615561 | OMIM |
| | HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4 | OMIM | MIM:612924 | OMIM |
| | MACULAR DEGENERATION, AGE-RELATED, 1 | OMIM | MIM:603075 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | OMIM | MIM:614602 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | OMIM | MIM:614602 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | OMIM | MIM:614602 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | OMIM | MIM:614602 | OMIM |
| | TRICHOHEPATOENTERIC SYNDROME 2 | OMIM | MIM:614602 | OMIM |
| SKIV2L | Syndromic diarrhea | OrphaNet | OrphaNet:84064 | orphanet |
| C2 | Immunodeficiency due to an early component of complement deficiency | OrphaNet | OrphaNet:169147 | orphanet |
| NEU1 | Juvenile sialidosis type 2 | OrphaNet | OrphaNet:93399 | orphanet |
| NEU1 | Sialidosis type 1 | OrphaNet | OrphaNet:812 | orphanet |
| NEU1 | Congenital sialidosis type 2 | OrphaNet | OrphaNet:93400 | orphanet |
| CFB | Atypical hemolytic uremic syndrome with B factor anomaly | OrphaNet | OrphaNet:93578 | orphanet |

## 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) |
|---|---|---|---|---|---|---|---|---|---|
| SKIV2L | missense variant | ENST00000375394 | NM_006929.4 | ENSP00000364543 | R/Q | cGg/cAg | ? | ? | 1 |
| SKIV2L | missense variant | ENST00000461073 | ? | ENSP00000419905 | R/Q | cGg/cAg | ? | ? | 1 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| SKIV2L | ? | ENSG00000204351 | pancreas | 5.47×10-11 (p-value) | GTEx Portal V6 | 13 |
| NELFE | ENST00000492185 | ILMN_1765532 | adipocyte | 1.04×10-7 (p-value) | MuTHER consortium | 6 |
| NELFE | ENST00000488426 | | blood | 6.64×10-5 (p-value) | MuTHER consortium | 6 |
| NELFE | ENST00000481121 | | | | | |
| NELFE | ENST00000375429 | | | | | |
| SKIV2L | ENST00000484835 | ILMN_1666512 | skin | 5.43×10-6 (p-value) | MuTHER consortium | 6 |
| SKIV2L | ENST00000375394 | | blood | 2.47×10-6 (p-value) | MuTHER consortium | 6 |
| SKIV2L | ENST00000474839 | | adipocyte | 2.04×10-6 (p-value) | MuTHER consortium | 6 |
| SKIV2L | ENST00000470453 | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| SKIV2L | ENST00000465703 | | | | | |
| SKIV2L | ENST00000491994 | | | | | |
| SKIV2L | ENST00000471818 | | | | | |
| SKIV2L | ENST00000483553 | | | | | |
| SKIV2L | ENST00000485349 | | | | | |
| SKIV2L | ? | ENSG00000204351 | muscularis mucosae | 1.86×10-27 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | lung | 1.00×10-21 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | atrial appendage | 1.13×10-9 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | transformed fibroblasts | 9.54×10-13 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | blood | 2.49×10-16 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | breast | 2.88×10-10 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | tibial artery | 3.59×10-35 (p-value) | GTEx Portal V6 | 13 |
| NELFE | ? | ENSG00000204356 | tibial artery | 1.15×10-8 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ENST00000484835 | ILMN_1666512 | monocyte | 1.40×10-8 (p-value) | Fairfax et al. | 10 |
| SKIV2L | ENST00000375394 | | b-cell | 4.53×10-10 (p-value) | Fairfax et al. | 10 |
| SKIV2L | ENST00000474839 | | | | | |
| SKIV2L | ENST00000470453 | | | | | |
| SKIV2L | ENST00000465703 | | | | | |
| SKIV2L | ENST00000491994 | | | | | |
| SKIV2L | ENST00000471818 | | | | | |
| SKIV2L | ENST00000483553 | | | | | |
| SKIV2L | ENST00000485349 | | | | | |
| NEU1 | ENST00000495807 | ILMN_1763144 | monocyte | 7.88×10-4 (p-value) | Fairfax et al. | 9 |
| NEU1 | ENST00000375631 | | | | | |
| NEU1 | ENST00000480384 | | | | | |
| NEU1 | ENST00000491768 | | | | | |
| SKIV2L | ? | ENSG00000204351 | thyroid | 1.97×10-16 (p-value) | GTEx Portal V6 | 13 |
| PRRT1 | ? | ENSG00000204314 | thyroid | 7.19×10-6 (p-value) | GTEx Portal V6 | 12 |
| NELFE | ? | ENSG00000204356 | skeletal muscle | 1.10×10-10 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | EBV lymphocytes | 3.42×10-9 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | transverse colon | 5.98×10-12 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | sun exposed skin | 1.82×10-16 (p-value) | GTEx Portal V6 | 13 |
| NELFE | ? | ENSG00000204356 | sun exposed skin | 4.14×10-14 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | aorta | 1.13×10-20 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | left ventricle | 3.58×10-8 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | subcutaneous adipocytes | 1.27×10-25 (p-value) | GTEx Portal V6 | 13 |
| NELFE | ? | ENSG00000204356 | subcutaneous adipocytes | 1.38×10-11 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | stomach | 5.07×10-12 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | visceral adipocytes | 5.78×10-10 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | cortex | 6.27×10-7 (p-value) | GTEx Portal V6 | 13 |

| | | | | | | |
|---|---|---|---|---|---|---|
| SKIV2L | ? | ENSG00000204351 | sigmoid colon | 1.41×10-11 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | coronary artery | 1.46×10-9 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | tibial nerve | 6.52×10-18 (p-value) | GTEx Portal V6 | 13 |
| NELFE | ? | ENSG00000204356 | tibial nerve | 1.87×10-8 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | esophagus mucosa | 5.48×10-12 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | gastroesophageal junction | 1.96×10-10 (p-value) | GTEx Portal V6 | 13 |
| SKIV2L | ? | ENSG00000204351 | testis | 1.51×10-7 (p-value) | GTEx Portal V6 | 13 |
| NELFE | ENST00000492185 | ILMN_1765532 | monocyte | 3.33×10-15 (p-value) | Zeller et al. | 2 |
| NELFE | ENST00000488426 | | | | | |
| NELFE | ENST00000481121 | | | | | |
| NELFE | ENST00000375429 | | | | | |
| SKIV2L | ENST00000484835 | ILMN_1666512 | monocyte | 5.72×10-13 (p-value) | Zeller et al. | 2 |
| SKIV2L | ENST00000375394 | | | | | |
| SKIV2L | ENST00000474839 | | | | | |
| SKIV2L | ENST00000470453 | | | | | |
| SKIV2L | ENST00000465703 | | | | | |
| SKIV2L | ENST00000491994 | | | | | |
| SKIV2L | ENST00000471818 | | | | | |
| SKIV2L | ENST00000483553 | | | | | |
| SKIV2L | ENST00000485349 | | | | | |

## Putative effect on regulation

### ENCODE promoter-associated DHS

| SNiPA promoter id | variant(s) | associated gene(s) |
|---|---|---|
| ENCP00000048142 | | CFB |
| | | CFB |
| | | CFB |
| | | CFB |
| | | CFB |
| | | CFB |
| | | CFB |
| ENCP00000048147 | | SKIV2L |
| | | SKIV2L |
| | | SKIV2L |
| | | SKIV2L |
| | | SKIV2L |

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000264486 (promoter flanking region) | 1 | cervix (HeLa-S3) | DNase1, Cmyc, PolII, Max, Jund, H3K27ac, H3K4me1 |
| | | embryonic stem cell (H1ESC) | DNase1, H3K27me3 |
| | | liver (HepG2) | PolII, H3K79me2, H3K4me1, H3K4me2, H3K36me3 |
| | | nervous (NH-A) | DNase1 |
| | | blood (K562) | H3K4me1, DNase1, PolII, Cjun, Max |
| | | skin (NHDF-AD) | DNase1 |
| ENSR00000025316 (CTCF binding site) | 1 | embryonic stem cell (H1ESC) | H3K36me3, Rad21, CTCF, DNase1 |
| | | HSMMtube | H3K36me3, CTCF |
| | | blood (K562) | Rad21, CTCF, H3K36me3 |
| | | skin (NHDF-AD) | H3K36me3, CTCF |

| | |
|---|---|
| muscle (HSMM) | CTCF |
| liver (HepG2) | PolII, Rad21, CTCF, H3K36me3, DNase1 |
| lung (IMR90) | H3K36me3, CTCF |
| blood (GM12878) | CTCF, H3K36me3 |
| nervous (NH-A) | CTCF, H3K36me3 |
| skin (NHEK) | CTCF, H3K36me3 |
| NHLF | CTCF, H3K36me3 |
| Osteobl | H3K36me3, CTCF |
| blood (DND-41) | CTCF, H3K36me3 |
| breast (HMEC) | H3K36me3, CTCF |
| cervix (HeLa-S3) | CTCF, H3K36me3, DNase1 |
| monocytes (Monocytes-CD14+) | H3K36me3, CTCF |
| endothelium (HUVEC) | H3K36me3, CTCF, DNase1 |
| A549 | H3K36me3, CTCF |

## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| C2 | upstream gene variant, downstream gene variant | 512 | ENST00000468407 | ? | ? | 6 |
| C2 | downstream gene variant | 2295 | ENST00000418949 | NM_001282459.1 | ENSP00000406190 | 7 |
| C2 | downstream gene variant | 3071 | ENST00000494905 | ? | ENSP00000419048 | 5 |
| C2 | downstream gene variant | 1363 | ENST00000484636 | ? | ENSP00000420305 | 7 |
| C2 | downstream gene variant | 3058 | ENST00000452202 | ? | ENSP00000406121 | 5 |
| C2 | downstream gene variant | 92 | ENST00000411571 | ? | ENSP00000388727 | 6 |
| C2 | downstream gene variant | 3108 | ENST00000447952 | ? | ENSP00000391354 | 5 |
| C2 | downstream gene variant | 3502 | ENST00000486124 | ? | ? | 1 |
| C2 | downstream gene variant | 3502 | ENST00000299367 | NM_001282458.1, NM_000063.5 | ENSP00000299367 | 1 |
| C2 | downstream gene variant | 3550 | ENST00000383177 | ? | ENSP00000372664 | 1 |
| C2 | downstream gene variant | 3783 | ENST00000482060 | ? | ENSP00000418332 | 1 |
| C2 | downstream gene variant | 3503 | ENST00000442278 | NM_001145903.2 | ENSP00000395683 | 1 |
| C2 | downstream gene variant | 3653 | ENST00000469372 | NM_001282457.1 | ENSP00000418923 | 1 |
| C2 | downstream gene variant | 4321 | ENST00000497706 | ? | ENSP00000417482 | 1 |
| C2 | downstream gene variant | 3503 | ENST00000452323 | NM_001178063.2 | ENSP00000392322 | 1 |
| C2-AS1 | upstream gene variant | 1437 | ENST00000630806 | ? | ? | 1 |
| CFB | downstream gene variant | 2883 | ENST00000482886 | ? | ? | 2 |
| CFB | upstream gene variant, downstream gene variant | 56 | ENST00000425368 | NM_001710.5 | ENSP00000416561 | 3 |
| CFB | upstream gene variant, downstream gene variant | 1495 | ENST00000452035 | ? | ? | 3 |
| CFB | upstream gene variant, downstream gene variant | 2542 | ENST00000497841 | ? | ? | 3 |
| CFB | upstream gene variant, downstream gene variant | 2054 | ENST00000472581 | ? | ? | 2 |
| CFB | upstream gene variant, downstream gene variant | 2033 | ENST00000475617 | ? | ENSP00000420090 | 3 |
| CFB | upstream gene variant, downstream gene variant | 1205 | ENST00000460718 | ? | ENSP00000417793 | 4 |
| CFB | upstream gene variant, downstream gene variant | 56 | ENST00000482312 | ? | ? | 3 |
| CFB | upstream gene variant, downstream gene variant | 557 | ENST00000467360 | ? | ? | 3 |
| CFB | upstream gene variant, downstream gene variant | 2589 | ENST00000461483 | ? | ? | 3 |
| CFB | upstream gene variant, downstream gene variant | 56 | ENST00000498317 | ? | ENSP00000418833 | 3 |

| | | | | | |
|---|---|---|---|---|---|
| CFB | downstream gene variant | 87 | ENST00000483004 ? | ENSP00000419887 | 2 |
| CFB | downstream gene variant | 2542 | ENST00000467150 ? | ? | 2 |
| CFB | downstream gene variant | 2542 | ENST00000465750 ? | ? | 2 |
| MIR1236 | downstream gene variant, upstream gene variant | 163 | ENST00000408340 ? | ? | 4 |
| NELFE | downstream gene variant, upstream gene variant | 87 | ENST00000492185 ? | ? | 3 |
| NELFE | downstream gene variant, upstream gene variant | 88 | ENST00000488426 ? | ? | 3 |
| NELFE | downstream gene variant, upstream gene variant | 1419 | ENST00000375429 NM_002904.5 | ENSP00000364578 | 2 |
| NELFE | downstream gene variant, upstream gene variant | 848 | ENST00000441998 ? | ENSP00000397914 | 4 |
| NELFE | downstream gene variant, upstream gene variant | 1479 | ENST00000436289 ? | ENSP00000414029 | 3 |
| NELFE | downstream gene variant, upstream gene variant | 1561 | ENST00000492539 ? | ENSP00000419183 | 3 |
| NELFE | downstream gene variant, upstream gene variant | 89 | ENST00000375425 ? | ENSP00000364574 | 3 |
| NELFE | downstream gene variant, upstream gene variant | 88 | ENST00000481121 ? | ? | 3 |
| NELFE | downstream gene variant, upstream gene variant | 1575 | ENST00000494956 ? | ENSP00000420205 | 3 |
| NELFE | downstream gene variant, upstream gene variant | 89 | ENST00000444811 ? | ENSP00000388400 | 3 |
| NELFE | downstream gene variant, upstream gene variant | 297 | ENST00000625905 ? | ENSP00000486884 | 3 |
| NELFE | downstream gene variant, upstream gene variant | 1394 | ENST00000454913 ? | ENSP00000409389 | 3 |
| NELFE | downstream gene variant, upstream gene variant | 1564 | ENST00000491139 ? | ? | 3 |
| NELFE | downstream gene variant, upstream gene variant | 1670 | ENST00000426722 ? | ENSP00000394340 | 3 |
| SKIV2L | upstream gene variant, downstream gene variant | 571 | ENST00000492900 ? | ? | 2 |
| SKIV2L | upstream gene variant | 2068 | ENST00000474839 ? | ENSP00000420470 | 1 |
| SKIV2L | upstream gene variant | 2014 | ENST00000488648 ? | ? | 1 |
| SKIV2L | upstream gene variant | 2054 | ENST00000461073 ? | ENSP00000419905 | 1 |
| SKIV2L | upstream gene variant | 2090 | ENST00000628157 ? | ENSP00000485707 | 1 |
| SKIV2L | upstream gene variant | 968 | ENST00000466290 ? | ? | 2 |
| SKIV2L | upstream gene variant | 1977 | ENST00000375394 NM_006929.4 | ENSP00000364543 | 1 |
| SKIV2L | upstream gene variant | 2038 | ENST00000465703 ? | ? | 1 |
| XXbac-BPG116M5.17 | downstream gene variant | 110 | ENST00000477310 ? | ENSP00000418996 | 2 |
| XXbac-BPG116M5.17 | downstream gene variant | 92 | ENST00000456570 ? | ENSP00000410815 | 2 |

## Putative effect on transcript

### Synonymous coding variant

| gene | affected transcript | RefSeq id | protein | AA's | exchanged codons | variant(s) |
|---|---|---|---|---|---|---|
| C2 | ENST00000469372 | NM_001282457.1 | ENSP00000418923 | A | gcA/gcG | 1 |
| C2 | ENST00000442278 | NM_001145903.2 | ENSP00000395683 | A | gcA/gcG | 1 |
| C2 | ENST00000485690 | ? | ENSP00000417790 | A | gcA/gcG | 1 |
| C2 | ENST00000497706 | ? | ENSP00000417482 | A | gcA/gcG | 1 |
| C2 | ENST00000452323 | NM_001178063.2 | ENSP00000392322 | A | gcA/gcG | 1 |

| | | | | | | |
|---|---|---|---|---|---|---|
| C2 | ENST00000383177 | ? | ENSP00000372664 | A | gcA/gcG | 1 |
| C2 | ENST00000299367 | NM_001282458.1, NM_000063.5 | ENSP00000299367 | A | gcA/gcG | 1 |
| XXbac-BPG116M5.17 | ENST00000456570 | ? | ENSP00000410815 | A | gcA/gcG | 1 |
| XXbac-BPG116M5.17 | ENST00000477310 | ? | ENSP00000418996 | A | gcA/gcG | 1 |

## Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| C2 | ENST00000482060 | ? | ENSP00000418332 | 8 |
| C2 | ENST00000486124 | ? | ? | 8 |
| C2 | ENST00000469372 | NM_001282457.1 | ENSP00000418923 | 8 |
| C2 | ENST00000442278 | NM_001145903.2 | ENSP00000395683 | 8 |
| C2 | ENST00000485690 | ? | ENSP00000417790 | 8 |
| C2 | ENST00000497706 | ? | ENSP00000417482 | 8 |
| C2 | ENST00000452323 | NM_001178063.2 | ENSP00000392322 | 8 |
| C2 | ENST00000383177 | ? | ENSP00000372664 | 8 |
| C2 | ENST00000299367 | NM_001282458.1, NM_000063.5 | ENSP00000299367 | 8 |
| C2-AS1 | ENST00000630806 | ? | ? | 7 |
| CFB | ENST00000461483 | ? | ? | 1 |
| CFB | ENST00000483004 | ? | ENSP00000419887 | 1 |
| CFB | ENST00000425368 | NM_001710.5 | ENSP00000416561 | 1 |
| CFB | ENST00000452035 | ? | ? | 1 |
| CFB | ENST00000497841 | ? | ? | 1 |
| CFB | ENST00000465750 | ? | ? | 1 |
| NELFE | ENST00000481121 | ? | ? | 2 |
| NELFE | ENST00000488426 | ? | ? | 2 |
| NELFE | ENST00000375429 | NM_002904.5 | ENSP00000364578 | 2 |
| NELFE | ENST00000492185 | ? | ? | 2 |
| NELFE | ENST00000491139 | ? | ? | 1 |
| NELFE | ENST00000436289 | ? | ENSP00000414029 | 1 |
| NELFE | ENST00000444811 | ? | ENSP00000388400 | 2 |
| NELFE | ENST00000375425 | ? | ENSP00000364574 | 2 |
| NELFE | ENST00000426722 | ? | ENSP00000394340 | 1 |
| NELFE | ENST00000492539 | ? | ENSP00000419183 | 1 |
| NELFE | ENST00000494956 | ? | ENSP00000420205 | 1 |
| NELFE | ENST00000441998 | ? | ENSP00000397914 | 1 |
| NELFE | ENST00000454913 | ? | ENSP00000409389 | 1 |
| SKIV2L | ENST00000628157 | ? | ENSP00000485707 | 1 |
| SKIV2L | ENST00000474839 | ? | ENSP00000420470 | 1 |
| XXbac-BPG116M5.17 | ENST00000456570 | ? | ENSP00000410815 | 9 |
| XXbac-BPG116M5.17 | ENST00000477310 | ? | ENSP00000418996 | 9 |

## 3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|

| | | | | |
|---|---|---|---|---|
| C2 | ENST00000482060 | ? | ENSP00000418332 | 1 |
| C2 | ENST00000411571 | ? | ENSP00000388727 | 1 |
| NELFE | ENST00000375429 | NM_002904.5 | ENSP00000364578 | 1 |

## Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| C2 | ENST00000486124 | ? | 1 |
| CFB | ENST00000467150 | ? | 1 |
| CFB | ENST00000482886 | ? | 1 |
| SKIV2L | ENST00000488648 | ? | 1 |
| SKIV2L | ENST00000465703 | ? | 1 |