# Locus 83 (rs4859571)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr4:76,852,839-76,985,488 |
| block size | 132,650 bp |
| variant count | 62 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.292 [-4.482 – 2.568] | gene(s) hit or close-by | AC110615.1, AC112719.2, ART3, CXCL11, CXCL9, NAAA, RP11-630D6.1, RP11-630D6.5, SDAD1 |
| phastCons | μ = 0.119 [0 – 1] | eQTL gene(s) | ART3, CXCL10, CXCL11, CXCL9, NAAA, SCARB2 |
| GERP++ | μ = -0.578 [-10.3 – 4.28] | potentially regulated gene(s) | CCDC158, NAAA, NUP54, USO1 |
| CADD score | μ = 4.078 [0.03 – 18.35] | disease gene(s) | SCARB2 |

## Trait annotations

### Variant association

| trait | min(p-value) | source DB | source entry/link | | variant(s) |
|---|---|---|---|---|---|
| Heart rate | $1.52\times10^{-5}$ | dbGaP | pha001394 | dbGaP | 1 |

### Disease gene annotation

| gene | trait | source DB | source entry/link | |
|---|---|---|---|---|
| SCARB2 | EPILEPSY, PROGRESSIVE MYOCLONIC, 4, WITH OR WITHOUT RENAL [...] | OMIM | MIM:254900 | OMIM |
| SCARB2 | Gaucher disease type 1 | OrphaNet | OrphaNet:77259 | orphanet |
| SCARB2 | Action myoclonus - renal failure syndrome | OrphaNet | OrphaNet:163696 | orphanet |
| SCARB2 | Unverricht-Lundborg disease | OrphaNet | OrphaNet:308 | 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) |
|---|---|---|---|---|---|---|---|---|---|
| NAAA | missense variant | ENST00000505594 | ? | ENSP00000426977 | I/V | Atc/Gtc | ? | ? | 1 |
| NAAA | missense variant | ENST00000602782 | ? | ENSP00000473575 | I/V | Atc/Gtc | ? | ? | 1 |
| NAAA | missense variant | ENST00000286733 | NM_014435.3 | ENSP00000286733 | I/V | Atc/Gtc | ? | ? | 1 |
| NAAA | missense variant | ENST00000507956 | ? | ENSP00000427641 | I/V | Atc/Gtc | ? | ? | 1 |
| NAAA | missense variant | ENST00000507187 | ? | ENSP00000423142 | I/V | Atc/Gtc | ? | ? | 1 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| ART3 | ? | ENSG00000156219 | pancreas | $6.72\times10^{-15}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | pancreas | $9.25\times10^{-12}$ (p-value) | GTEx Portal V6 | 62 |

| | | | | | | |
|---|---|---|---|---|---|---|
| CXCL11 | ? | ENSG00000169248 | pancreas | $1.02\times10^{-6}$ (p-value) | GTEx Portal V6 | 24 |
| NAAA | ? | ENSG00000138744 | muscularis mucosae | $1.64\times10^{-18}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | lung | $6.09\times10^{-21}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | transformed fibroblasts | $8.78\times10^{-15}$ (p-value) | GTEx Portal V6 | 62 |
| SCARB2 | ? | ENSG00000138760 | transformed fibroblasts | $1.06\times10^{-6}$ (p-value) | GTEx Portal V6 | 6 |
| NAAA | ? | ENSG00000138744 | blood | $1.63\times10^{-22}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | breast | $2.40\times10^{-15}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | tibial artery | $7.45\times10^{-23}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | thyroid | $6.38\times10^{-15}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | skeletal muscle | $6.09\times10^{-27}$ (p-value) | GTEx Portal V6 | 62 |
| CXCL10 | ? | ENSG00000169245 | EBV lymphocytes | $2.93\times10^{-9}$ (p-value) | GTEx Portal V6 | 42 |
| NAAA | ? | ENSG00000138744 | transverse colon | $1.61\times10^{-11}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | blood | $1.05\times10^{-6}$ (q-value) | SeeQTL DB (HapMap) | 16 |
| CXCL10 | ? | ENSG00000169245 | blood | $7.80\times10^{-4}$ (q-value) | SeeQTL DB (HapMap) | 12 |
| NAAA | ? | ENSG00000138744 | sun exposed skin | $3.52\times10^{-24}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | unexposed skin | $5.99\times10^{-17}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | aorta | $6.37\times10^{-22}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | subcutaneous adipocytes | $4.54\times10^{-18}$ (p-value) | GTEx Portal V6 | 62 |
| ? | ? | ILMN_2067890 | monocyte | $1.57\times10^{-17}$ (p-value) | Zeller et al. | 9 |
| NAAA | ENST00000286733 | ILMN_2391512 | monocyte | $2.13\times10^{-19}$ (p-value) | Zeller et al. | 9 |
| NAAA | ENST00000602782 | | | | | |
| NAAA | ENST00000513045 | | | | | |
| NAAA | ENST00000507956 | | | | | |
| NAAA | ENST00000505594 | | | | | |
| NAAA | ENST00000511606 | | | | | |
| ? | ? | ILMN_1668605 | monocyte | $4.68\times10^{-15}$ (p-value) | Zeller et al. | 5 |
| ? | ? | ILMN_2285568 | monocyte | $4.21\times10^{-87}$ (p-value) | Zeller et al. | 9 |
| NAAA | ? | ENSG00000138744 | caudate basal ganglia | $1.41\times10^{-8}$ (p-value) | GTEx Portal V6 | 52 |
| NAAA | ? | ENSG00000138744 | stomach | $3.34\times10^{-13}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | visceral adipocytes | $2.50\times10^{-21}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | sigmoid colon | $1.05\times10^{-9}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | coronary artery | $6.63\times10^{-10}$ (p-value) | GTEx Portal V6 | 62 |
| ART3 | ? | ENSG00000156219 | tibial nerve | $3.51\times10^{-9}$ (p-value) | GTEx Portal V6 | 60 |
| NAAA | ? | ENSG00000138744 | tibial nerve | $1.59\times10^{-11}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | esophagus mucosa | $8.68\times10^{-25}$ (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | gastroesophageal junction | $2.64\times10^{-12}$ (p-value) | GTEx Portal V6 | 62 |

| | | | | | | |
|---|---|---|---|---|---|---|
| NAAA | ? | ENSG00000138744 | adrenal gland | 1.07×10-16 (p-value) | GTEx Portal V6 | 62 |
| NAAA | ? | ENSG00000138744 | left ventricle | 2.30×10-9 (p-value) | GTEx Portal V6 | 60 |
| NAAA | ? | ENSG00000138744 | atrial appendage | 9.88×10-9 (p-value) | GTEx Portal V6 | 57 |
| NAAA | ? | ENSG00000138744 | ovary | 9.96×10-7 (p-value) | GTEx Portal V6 | 43 |
| NAAA | ? | ENSG00000138744 | spleen | 1.51×10-7 (p-value) | GTEx Portal V6 | 45 |
| NAAA | ? | ENSG00000138744 | pituitary | 3.06×10-7 (p-value) | GTEx Portal V6 | 56 |
| NAAA | ? | ENSG00000138744 | frontal cortex | 2.61×10-7 (p-value) | GTEx Portal V6 | 42 |
| NAAA | ? | ENSG00000138744 | liver | 2.36×10-7 (p-value) | GTEx Portal V6 | 32 |
| NAAA | ENST00000286733 | ILMN_2391512 | blood | 7.55×10-28 (p-value) | MuTHER consortium | 21 |
| NAAA | ENST00000602782 | | | | | |
| NAAA | ENST00000513045 | | | | | |
| NAAA | ENST00000507956 | | | | | |
| NAAA | ENST00000505594 | | | | | |
| NAAA | ENST00000511606 | | | | | |
| CXCL9 | ENST00000264888 | ILMN_1745356 | blood | 1.93×10-6 (p-value) | MuTHER consortium | 16 |
| CXCL10 | ENST00000306602 | ILMN_1791759 | blood | 3.78×10-35 (p-value) | MuTHER consortium | 21 |
| NAAA | ENST00000513045 | ILMN_2391512 | monocyte | 8.05×10-13 (p-value) | Fairfax et al. | 3 |
| NAAA | ENST00000602782 | | b-cell | 1.98×10-6 (p-value) | Fairfax et al. | 3 |
| NAAA | ENST00000286733 | | | | | |
| NAAA | ENST00000507956 | | | | | |
| NAAA | ENST00000505594 | | | | | |
| NAAA | ENST00000511606 | | | | | |
| ? | ? | ILMN_1668605 | monocyte | 1.09×10-12 (p-value) | Fairfax et al. | 6 |
| | | | b-cell | 5.29×10-6 (p-value) | | |
| NAAA | ENST00000602782 | 214765_s_at | blood | 2.10×10-22 (p-value) | Dixon et al. | 7 |
| NAAA | ENST00000286733 | | | | | |
| NAAA | ENST00000513045 | | | | | |
| NAAA | ENST00000507956 | | | | | |
| NAAA | ENST00000505594 | | | | | |
| NAAA | ENST00000511606 | | | | | |
| NAAA | ENST00000286733 | 227135_at | blood | 2.50×10-21 (p-value) | Dixon et al. | 7 |
| NAAA | ENST00000511606 | | | | | |
| CXCL10 | ENST00000306602 | 204533_at | blood | 2.40×10-18 (p-value) | Dixon et al. | 7 |
| NAAA | ? | ENSG00000138744 | cerebellum | 9.13×10-6 (p-value) | GTEx Portal V6 | 3 |
| ? | ? | ILMN_1668605 | liver | 2.89×10-15 (p-value) | Innocenti et al. | 1 |
| CXCL10 | ENST00000306602 | ILMN_1791759 | blood | 3.55×10-5 (p-value) | Westra et al. | 9 |
| NAAA | ? | ENSG00000138744 | uterus | 4.78×10-7 (p-value) | GTEx Portal V6 | 12 |
| NAAA | ? | ENSG00000138744 | anterior cingulate cortex | 1.32×10-6 (p-value) | GTEx Portal V6 | 1 |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000382695 | 1 | ENCP00000041438 | NUP54 |
| | | ENCP00000041452 | CCDC158 |
| | | ENCP00000041415 | NAAA |
| ENCE00000382651 | 1 | ENCP00000041411 | USO1 |
| ENCE00000382654 | 1 | ENCP00000041411 | USO1 |

## Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001432208 (promoter) | 1 | embryonic stem cell (H1ESC) | DNase1, H3K4me3, PolII, Gabp, Sin3Ak20, H3K9ac, Yy1, TAF1, H3K4me2 |
| | | HSMMtube | H3K79me2, H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1 |
| | | blood (K562) | H3K79me2, HEY1, H2AZ, TAF1, H3K4me2, H3K36me3, DNase1, H3K4me3, Cmyc, Gabp, PolII, Max, H3K9ac |
| | | skin (NHDF-AD) | H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac |
| | | muscle (HSMM) | H2AZ, H3K79me2, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K4me3 |
| | | liver (HepG2) | Gabp, PolII, H3K79me2, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, DNase1 |
| | | blood (GM12878) | H3K79me2, H3K4me3, H3K4me2, H3K9ac, ELF1, Gabp, DNase1, Cmyc, PolII, H2AZ |
| | | lung (IMR90) | DNase1, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac |
| | | nervous (NH-A) | H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1 |
| | | skin (NHEK) | DNase1, H3K4me3, H3K4me2, H3K27ac, H3K9ac |
| | | NHLF | H3K27ac, H3K9ac, H3K4me3, DNase1 |
| | | Osteobl | H3K4me2, H3K4me3, H3K27ac, H2AZ |
| | | blood (DND-41) | H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1 |
| | | breast (HMEC) | DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2 |
| | | cervix (HeLa-S3) | DNase1, PolII, Gabp, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2 |
| | | monocytes (Monocytes-CD14+) | H3K36me3, H3K9ac, H3K27ac, H3K4me2, DNase1, H3K4me3, H3K27me3 |
| | | endothelium (HUVEC) | Max, DNase1, H3K4me2, H3K4me3, H3K9ac, PolII, Cmyc |
| | | A549 | H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3 |
| ENSR00001243218 (promoter flanking region) | 2 | NHLF | H3K27ac |
| | | embryonic stem cell (H1ESC) | H3K27me3, DNase1 |
| | | HSMMtube | H3K27me3 |
| | | Osteobl | H3K4me2, H3K27ac |
| | | blood (DND-41) | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | skin (NHDF-AD) | DNase1 |
| | | breast (HMEC) | H3K27ac, H3K4me2 |
| | | blood (GM12878) | H2AZ, H3K27ac, H3K4me2 |
| | | lung (IMR90) | DNase1, H3K18ac, H3K27ac, H3K4ac, H2BK120ac, H2BK20ac, H4K91ac, H3K56ac |
| | | skin (NHEK) | DNase1 |
| ENSR00001243226 (enhancer) | 1 | embryonic stem cell (H1ESC) | CTCF, Yy1, DNase1 |
| | | endothelium (HUVEC) | H3K36me3 |
| | | HSMMtube | H3K27me3 |
| | | skin (NHEK) | CTCF |
| ENSR00001685898 (CTCF binding site) | 1 | endothelium (HUVEC) | H3K36me3 |
| | | embryonic stem cell (H1ESC) | CTCF, Yy1, DNase1 |
| | | HSMMtube | H3K27me3 |
| | | skin (NHEK) | CTCF |
| ENSR00001243231 (enhancer) | 1 | NHLF | H3K27me3 |
| | | HSMMtube | H3K27me3 |
| | | Osteobl | H3K27me3 |
| | | blood (DND-41) | H3K27me3 |
| | | skin (NHDF-AD) | H3K27me3 |
| | | endothelium (HUVEC) | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |
| | | blood (GM12878) | Cjun, BATF, DNase1 |
| | | nervous (NH-A) | H3K27me3 |
| | | A549 | H3K27me3 |
| ENSR00001685900 (promoter flanking region) | 1 | HSMMtube | H3K27me3 |
| | | Osteobl | H3K27me3 |
| | | blood (DND-41) | H3K27me3 |
| | | skin (NHDF-AD) | H3K27me3 |

| | |
|---|---|
| breast (HMEC) | H3K4me2, DNase1 |
| monocytes (Monocytes-CD14+) | H3K4me1, DNase1 |
| endothelium (HUVEC) | H3K27me3 |
| nervous (NH-A) | H3K27me3 |
| A549 | H3K27me3 |
| skin (NHEK) | DNase1, H3K4me1, H3K4me2 |

## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| AC110615.1 | upstream gene variant, downstream gene variant | 122 | ENST00000625176 | ? | ENSP00000485507 | 6 |
| AC112719.2 | upstream gene variant | 1189 | ENST00000629900 | ? | ENSP00000487114 | 2 |
| ART3 | upstream gene variant | 1561 | ENST00000513353 | ? | ENSP00000421345 | 2 |
| ART3 | upstream gene variant | 2474 | ENST00000504914 | ? | ENSP00000421431 | 2 |
| ART3 | upstream gene variant | 1577 | ENST00000341029 | NM_001130017.2 | ENSP00000343843 | 2 |
| ART3 | upstream gene variant | 1577 | ENST00000510669 | ? | ? | 2 |
| ART3 | upstream gene variant | 1626 | ENST00000513122 | ? | ENSP00000422287 | 2 |
| CXCL11 | downstream gene variant, upstream gene variant | 721 | ENST00000306621 | NM_005409.4, NM_001302123.1 | ENSP00000306884 | 4 |
| CXCL11 | downstream gene variant, upstream gene variant | 595 | ENST00000503860 | ? | ENSP00000425819 | 4 |
| CXCL9 | upstream gene variant | 2135 | ENST00000264888 | NM_002416.1 | ENSP00000354901 | 2 |
| NAAA | upstream gene variant | 3046 | ENST00000505594 | ? | ENSP00000426977 | 1 |
| NAAA | upstream gene variant | 2333 | ENST00000507956 | ? | ENSP00000427641 | 1 |
| NAAA | downstream gene variant, upstream gene variant | 653 | ENST00000503636 | ? | ? | 14 |
| NAAA | upstream gene variant | 2255 | ENST00000286733 | NM_014435.3 | ENSP00000286733 | 1 |
| NAAA | upstream gene variant | 5 | ENST00000602782 | ? | ENSP00000473575 | 8 |
| NAAA | downstream gene variant, upstream gene variant | 8 | ENST00000507940 | ? | ? | 16 |
| NAAA | downstream gene variant, upstream gene variant | 168 | ENST00000507187 | ? | ENSP00000423142 | 11 |
| RP11-630D6.1 | downstream gene variant, upstream gene variant | 591 | ENST00000477971 | ? | ? | 3 |
| RP11-630D6.5 | upstream gene variant, downstream gene variant | 1333 | ENST00000501239 | ? | ? | 3 |
| SDAD1 | downstream gene variant | 833 | ENST00000503411 | ? | ENSP00000422368 | 1 |
| SDAD1 | downstream gene variant | 1148 | ENST00000395711 | NM_001288983.1 | ENSP00000379061 | 3 |
| SDAD1 | downstream gene variant, upstream gene variant | 479 | ENST00000515836 | ? | ? | 4 |
| SDAD1 | downstream gene variant | 768 | ENST00000395710 | ? | ENSP00000379060 | 3 |
| SDAD1 | downstream gene variant | 778 | ENST00000356260 | NM_001288984.1, NM_018115.3 | ENSP00000348596 | 3 |
| SDAD1 | downstream gene variant, upstream gene variant | 164 | ENST00000502543 | ? | ? | 13 |
| SDAD1 | downstream gene variant, upstream gene variant | 2587 | ENST00000513089 | ? | ? | 2 |
| SDAD1 | downstream gene variant, upstream gene variant | 1721 | ENST00000507396 | ? | ? | 4 |

# Putative effect on transcript

## Synonymous coding variant

| gene | affected transcript | RefSeq id | protein | AA's | exchanged codons | variant(s) |
|---|---|---|---|---|---|---|
| | ? | | | V | gtA/gtG | 1 |

| | | | | | | |
|---|---|---|---|---|---|---|
| NAAA | ENST00000505594 | | ENSP00000426977 | | | |
| NAAA | ENST00000507956 | ? | ENSP00000427641 | V | gtA/gtG | 1 |
| NAAA | ENST00000507187 | ? | ENSP00000423142 | V | gtA/gtG | 1 |
| NAAA | ENST00000286733 | NM_014435.3 | ENSP00000286733 | V | gtA/gtG | 1 |
| SDAD1 | ENST00000395711 | NM_001288983.1 | ENSP00000379061 | D | gaT/gaC | 1 |
| SDAD1 | ENST00000356260 | NM_001288984.1, NM_018115.3 | ENSP00000348596 | D | gaT/gaC | 1 |

## Intron variant (splice region)

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| NAAA | ENST00000505594 | ? | ENSP00000426977 | 1 |
| NAAA | ENST00000507940 | ? | ? | 1 |
| NAAA | ENST00000507956 | ? | ENSP00000427641 | 1 |
| NAAA | ENST00000507187 | ? | ENSP00000423142 | 1 |
| NAAA | ENST00000286733 | NM_014435.3 | ENSP00000286733 | 1 |

## Non-coding exon variant (splice region)

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| NAAA | ENST00000507940 | ? | 1 |

## Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| AC110615.1 | ENST00000625176 | ? | ENSP00000485507 | 1 |
| ART3 | ENST00000510669 | ? | ? | 19 |
| ART3 | ENST00000504914 | ? | ENSP00000421431 | 19 |
| ART3 | ENST00000513122 | ? | ENSP00000422287 | 19 |
| ART3 | ENST00000341029 | NM_001130017.2 | ENSP00000343843 | 19 |
| ART3 | ENST00000513353 | ? | ENSP00000421345 | 19 |
| CXCL11 | ENST00000503860 | ? | ENSP00000425819 | 3 |
| CXCL11 | ENST00000306621 | NM_005409.4, NM_001302123.1 | ENSP00000306884 | 2 |
| CXCL9 | ENST00000264888 | NM_002416.1 | ENSP00000354901 | 1 |
| NAAA | ENST00000505594 | ? | ENSP00000426977 | 20 |
| NAAA | ENST00000507940 | ? | ? | 6 |
| NAAA | ENST00000507956 | ? | ENSP00000427641 | 20 |
| NAAA | ENST00000507187 | ? | ENSP00000423142 | 10 |
| NAAA | ENST00000602782 | ? | ENSP00000473575 | 14 |
| NAAA | ENST00000286733 | NM_014435.3 | ENSP00000286733 | 20 |
| RP11-630D6.5 | ENST00000501239 | ? | ? | 4 |
| SDAD1 | ENST00000395711 | NM_001288983.1 | ENSP00000379061 | 13 |
| SDAD1 | ENST00000356260 | NM_001288984.1, NM_018115.3 | ENSP00000348596 | 13 |
| SDAD1 | ENST00000504975 | ? | ? | 5 |
| SDAD1 | ENST00000514710 | ? | ENSP00000427443 | 5 |
| SDAD1 | ENST00000503411 | ? | ENSP00000422368 | 5 |
| SDAD1 | ENST00000395710 | ? | ENSP00000379060 | 13 |
| SDAD1 | ENST00000513089 | ? | ? | 7 |
| SDAD1 | ENST00000502543 | ? | ? | 5 |

| | | | | |
|---|---|---|---|---|
| SDAD1 | ENST00000507396 | ? | ? | 3 |
| SDAD1 | ENST00000515836 | ? | ? | 1 |

## 3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| CXCL11 | ENST00000306621 | NM_005409.4, NM_001302123.1 | ENSP00000306884 | 1 |
| NAAA | ENST00000507187 | ? | ENSP00000423142 | 4 |
| SDAD1 | ENST00000395710 | ? | ENSP00000379060 | 1 |

## Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| RP11-630D6.1 | ENST00000477971 | ? | 1 |