# Locus 374 (rs12724450)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr1:150,273,976-150,514,747 |
| block size | 240,772 bp |
| variant count | 69 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.253 [-4.164 – 2.641] | gene(s) hit or close-by | AC242988.1, ECM1, FALEC, MRPS21, PRPF3, RPRD2, TARS2 |
| phastCons | μ = 0.096 [0 – 0.998] | eQTL gene(s) | C1orf54, CTSS, ECM1, FALEC, HORMAD1, RPRD2, TARS2 |
| GERP++ | μ = -0.311 [-5.83 – 4.94] | potentially regulated gene(s) | ADAMTSL4, ANP32E, CTSS, MCL1, PRPF3, RP11-458I7.1, RP11-54A4.2, TARS2, VPS45 |
| CADD score | μ = 4.125 [0.007 – 23.2] | disease gene(s) | ECM1, ADAMTSL4, PRPF3, TARS2, VPS45 |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| ECM1 | LIPOID PROTEINOSIS OF URBACH AND WIETHE | OMIM | MIM:247100 |
| ADAMTSL4 | ECTOPIA LENTIS ET PUPILLAE | OMIM | MIM:225200 |
| PRPF3 | RETINITIS PIGMENTOSA 18 | OMIM | MIM:601414 |
| TARS2 | COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 21 | OMIM | MIM:615918 |
| VPS45 | NEUTROPENIA, SEVERE CONGENITAL, 5, AUTOSOMAL RECESSIVE | OMIM | MIM:615285 |
| ECM1 | Lipoid proteinosis | OrphaNet | OrphaNet:530 |
| ADAMTSL4 | isolated ectopia lentis (EL) | OrphaNet | OrphaNet:1885 |
| PRPF3 | Retinitis pigmentosa | OrphaNet | OrphaNet:791 |
| VPS45 | Recurrent infections-myelofibrosis-nephromegaly syndrome | OrphaNet | OrphaNet:369852 |

## 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) |
|---|---|---|---|---|---|---|---|---|---|
| MRPS21 | missense variant | ENST00000581066 | NM_018997.3 | ENSP00000461930 | Q/R | cAg/cGg | ? | ? | 1 |
| MRPS21 | missense variant | ENST00000614145 | NM_031901.5 | ENSP00000480129 | Q/R | cAg/cGg | ? | ? | 1 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| HORMAD1 | ENST00000361824 | ILMN_1769849 | skin | $2.23\times10^{-5}$ (p-value) | MuTHER consortium | 7 |
| HORMAD1 | ENST00000470397 | | | | | |
| HORMAD1 | ENST00000486497 | | | | | |
| HORMAD1 | ENST00000322343 | | | | | |
| HORMAD1 | ENST00000368995 | | | | | |
| C1orf54 | ? | ENSG00000118292 | tibial nerve | $1.93\times10^{-7}$ (p-value) | GTEx Portal V6 | 47 |

| | | | | | | |
|---|---|---|---|---|---|---|
| FALEC | ? | ENSG00000228126 | tibial nerve | 6.10×10-6 (p-value) | GTEx Portal V6 | 26 |
| ECM1 | ? | ENSG00000143369 | transformed fibroblasts | 3.99×10-10 (p-value) | GTEx Portal V6 | 66 |
| TARS2 | ? | ENSG00000143374 | transformed fibroblasts | 3.66×10-5 (p-value) | GTEx Portal V6 | 1 |
| ECM1 | ? | ENSG00000143369 | cerebellum | 2.37×10-6 (p-value) | GTEx Portal V6 | 36 |
| ECM1 | ? | ENSG00000143369 | EBV lymphocytes | 4.17×10-9 (p-value) | GTEx Portal V6 | 66 |
| FALEC | ? | ENSG00000228126 | tibial artery | 6.15×10-6 (p-value) | GTEx Portal V6 | 36 |
| C1orf54 | ? | ENSG00000118292 | tibial artery | 8.11×10-6 (p-value) | GTEx Portal V6 | 3 |
| HORMAD1 | ? | ENSG00000143452 | esophagus mucosa | 1.36×10-8 (p-value) | GTEx Portal V6 | 67 |
| CTSS | ? | ENSG00000163131 | esophagus mucosa | 1.01×10-5 (p-value) | GTEx Portal V6 | 4 |
| ECM1 | ? | ENSG00000143369 | sun exposed skin | 2.08×10-6 (p-value) | GTEx Portal V6 | 48 |
| FALEC | ? | ENSG00000228126 | aorta | 4.10×10-6 (p-value) | GTEx Portal V6 | 27 |
| ECM1 | ? | ENSG00000143369 | thyroid | 9.15×10-7 (p-value) | GTEx Portal V6 | 45 |
| HORMAD1 | ? | ENSG00000143452 | subcutaneous adipocytes | 2.88×10-5 (p-value) | GTEx Portal V6 | 1 |
| ECM1 | ? | ENSG00000143369 | blood | 1.15×10-6 (p-value) | GTEx Portal V6 | 23 |
| ? | ? | ILMN_1702231 | blood | 3.16×10-5 (p-value) | Westra et al. | 1 |
| RPRD2 | ENST00000401000 | ILMN_3238889 | b-cell | 7.98×10-4 (p-value) | Fairfax et al. | 1 |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000036275 | 1 | ENCP00000004323 | ADAMTSL4 |
| | | ENCP00000004313 | PRPF3 |
| | | ENCP00000004312 | PRPF3 |
| ENCE00000036337 | 1 | ENCP00000004327 | MCL1 |
| ENCE00000036132 | 1 | ENCP00000004337 | CTSS |
| | | ENCP00000004311 | PRPF3 |
| | | ENCP00000004291 | VPS45 |
| | | ENCP00000004300 | ANP32E |
| | | ENCP00000004317 | TARS2 |
| | | ENCP00000004293 | VPS45 |
| | | ENCP00000004287 | RP11-458I7.1 |
| ENCE00000036329 | 1 | ENCP00000004323 | ADAMTSL4 |
| ENCE00000036323 | 1 | ENCP00000004325 | RP11-54A4.2 |
| | | ENCP00000004324 | ADAMTSL4 |

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001524644 (open chromatin region) | 1 | embryonic stem cell (H1ESC) | H3K36me3, PolII |
| | | lung (IMR90) | H3K36me3 |
| | | A549 | H3K36me3 |
| | | blood (K562) | PolII |
| | | muscle (HSMM) | H3K27ac, DNase1 |

| | | | |
|---|---|---|---|
| ENSR00000544743 (promoter) | 1 | embryonic stem cell (H1ESC) | TAF1, H3K4me2, H3K9ac, H3K4me3, H4K5ac, H3K36me3, CTCF, PolII, Jund, TAF7, H3K27ac, Yy1, DNase1 |
| | | HSMMtube | H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, DNase1 |
| | | blood (K562) | CTCFL, PolII, H3K4me2, DNase1, H3K4me3, TAF1, H2AZ, Yy1, H3K27ac, Max, PU1, H3K9ac, Cmyc, H3K79me2, HEY1 |
| | | skin (NHDF-AD) | H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac |
| | | muscle (HSMM) | DNase1, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac |
| | | liver (HepG2) | Yy1, PolII, H3K79me2, TAF1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, DNase1 |
| | | lung (IMR90) | H3K4me3, H3K4ac, H3K9ac, H4K5ac, H3K4me2, DNase1, H4K8ac, H3K18ac, H3K27ac |
| | | blood (GM12878) | H3K27ac, H3K4me2, H3K9ac, H3K4me3, H3K79me2, TAF1, PU1, PolII, H2AZ, DNase1, Yy1 |
| | | nervous (NH-A) | H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1 |
| | | skin (NHEK) | H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1 |
| | | NHLF | DNase1, H3K4me3, H3K9ac, H3K27ac |
| | | Osteobl | H2AZ, H3K27ac, H3K4me3, H3K4me2 |
| | | blood (DND-41) | H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3 |
| | | breast (HMEC) | DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2 |
| | | cervix (HeLa-S3) | DNase1, Nrf1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3 |
| | | endothelium (HUVEC) | Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1, Cmyc |
| | | A549 | H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3 |
| ENSR00001586441 (enhancer) | 1 | cervix (HeLa-S3) | H3K79me2 |
| | | embryonic stem cell (H1ESC) | H3K36me3 |
| | | liver (HepG2) | H3K79me2 |
| | | blood (GM12878) | H3K79me2 |
| | | blood (DND-41) | H3K36me3 |
| | | blood (K562) | H3K27me3, H3K79me2 |
| | | muscle (HSMM) | H3K79me2 |
| ENSR00001524646 (enhancer) | 1 | liver (HepG2) | H3K79me2, H3K4me1, FOXA1 |
| | | blood (GM12878) | H3K79me2 |
| | | blood (DND-41) | H3K36me3 |
| ENSR00000544755 (promoter flanking region) | 2 | embryonic stem cell (H1ESC) | DNase1 |
| | | liver (HepG2) | H3K4me1, FOXA1 |
| | | lung (IMR90) | DNase1, H3K36me3 |
| | | blood (K562) | Egr1 |
| | | A549 | H3K36me3 |
| | | skin (NHEK) | H3K4me1, H3K27ac, H3K9ac, H3K4me2, DNase1 |
| | | breast (HMEC) | DNase1, H3K27ac, H3K4me2 |
| ENSR00000079774 (promoter) | 1 | embryonic stem cell (H1ESC) | H3K36me3, PolII, Gabp, H3K9ac, Yy1, TAF1, H3K4me2, SP1, H3K4me3, DNase1 |
| | | HSMMtube | H3K9ac, H3K4me2, H2AZ, H3K36me3, DNase1 |
| | | blood (K562) | H3K4me3, DNase1, H3K36me3, H3K27ac, Max, SP2, MEF2A, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, Cfos, PolII, H3K4me2 |
| | | skin (NHDF-AD) | H3K4me3, DNase1, H3K9ac, H3K4me2 |
| | | muscle (HSMM) | H2AZ, H3K4me3, H3K4me2, H3K9ac, DNase1 |
| | | liver (HepG2) | H3K79me2, TAF1, ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K36me3, DNase1, Gabp, PolII |
| | | lung (IMR90) | DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac |
| | | blood (GM12878) | ELF1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K9ac, H3K36me3, Gabp, Yy1, Pbx3, PolII, BCLAF1, MEF2A, H2AZ, DNase1 |
| | | nervous (NH-A) | H3K9ac, H3K4me2, H3K4me3, DNase1 |
| | | skin (NHEK) | H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1, H3K27ac |
| | | NHLF | DNase1, H3K4me3, H3K27ac |
| | | Osteobl | H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3 |
| | | blood (DND-41) | H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3 |
| | | breast (HMEC) | CTCF, H3K4me2, H3K9ac, H3K4me3 |
| | | cervix (HeLa-S3) | DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, Cfos, PolII |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3 |
| | | endothelium (HUVEC) | H3K4me2, H3K4me3, PolII, DNase1, Cmyc |
| | | A549 | H3K4me3, H3K4me2, H3K9ac, DNase1 |
| ENSR00001524653 (promoter) | 1 | embryonic stem cell (H1ESC) | H3K36me3, DNase1 |
| | | HSMMtube | H3K4me2, DNase1 |
| | | blood (K562) | H3K27ac, H3K9ac, Jund, PolII, H3K4me2, H3K36me3, DNase1 |
| | | skin (NHDF-AD) | H3K4me3, DNase1, H3K9ac, H3K4me2 |
| | | muscle (HSMM) | H3K79me2, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1 |
| | | liver (HepG2) | H3K36me3 |
| | | blood (GM12878) | PolII, H3K36me3 |

| | | | |
|---|---|---|---|
| | | lung (IMR90) | DNase1, H3K18ac, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac |
| | | nervous (NH-A) | H3K9ac, H3K4me2, DNase1 |
| | | skin (NHEK) | H3K4me1, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1 |
| | | NHLF | H3K4me3, H3K27ac, H3K36me3 |
| | | Osteobl | H3K27ac, H3K4me2, H3K36me3 |
| | | blood (DND-41) | H3K36me3 |
| | | cervix (HeLa-S3) | H3K9ac, H3K4me2, H3K27ac, H3K4me1, H3K36me3, DNase1 |
| | | monocytes (Monocytes-CD14+) | H3K36me3 |
| | | endothelium (HUVEC) | Cjun, PolII, DNase1 |
| | | A549 | H3K36me3 |
| ENSR00000285718 (promoter) | 1 | embryonic stem cell (H1ESC) | H3K36me3, H3K27me3, CTCF, Rad21, TAF1, H3K4me2, H3K4me3, DNase1 |
| | | HSMMtube | H3K4me2, H2AZ, H3K36me3, DNase1 |
| | | blood (K562) | H3K27ac, H3K9ac, H2AZ, PolII, H3K4me2, CTCF, H3K4me1, H3K36me3, DNase1, H3K4me3 |
| | | skin (NHDF-AD) | H3K36me3, H3K4me1, H3K4me3, DNase1, H3K9ac, H3K4me2 |
| | | muscle (HSMM) | H2AZ, H3K36me3, DNase1 |
| | | liver (HepG2) | PolII, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF, H3K4me3, DNase1 |
| | | lung (IMR90) | H3K4me3, H3K9ac, H3K36me3, H3K4me2, DNase1 |
| | | blood (GM12878) | Pbx3, PolII, H2AZ, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac |
| | | nervous (NH-A) | DNase1, H3K9ac, H3K4me2 |
| | | skin (NHEK) | H3K9ac, H3K4me2, H3K36me3, DNase1 |
| | | NHLF | H3K36me3, DNase1 |
| | | Osteobl | H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3 |
| | | blood (DND-41) | H3K4me1, H3K4me2, H3K9ac, H3K4me3, H3K36me3 |
| | | breast (HMEC) | DNase1, H3K4me3, H3K4me2 |
| | | cervix (HeLa-S3) | CTCF, DNase1, Nrf1, H3K9ac, Jund, H3K4me1, PolII |
| | | monocytes (Monocytes-CD14+) | H3K4me3, H3K27me3, DNase1 |
| | | endothelium (HUVEC) | H3K4me3, PolII, CTCF, DNase1, Max |
| | | A549 | H3K4me3, H3K4me2, H3K9ac, DNase1 |
| ENSR00001524662 (promoter flanking region) | 1 | NHLF | DNase1 |
| | | embryonic stem cell (H1ESC) | DNase1, Rad21, CTCF |
| | | HSMMtube | DNase1, H3K27ac, H3K9ac |
| | | blood (K562) | CTCF |
| | | skin (NHDF-AD) | DNase1 |
| | | muscle (HSMM) | H3K27ac, DNase1 |
| | | cervix (HeLa-S3) | CTCF, DNase1 |
| | | liver (HepG2) | CTCF, H3K4me1 |
| | | lung (IMR90) | DNase1, H3K4me1, H3K18ac, H3K27ac, H4K5ac, H4K91ac |
| | | nervous (NH-A) | DNase1 |
| | | skin (NHEK) | CTCF, DNase1 |

## Variation in RISC binding site

| gene | variant(s) | affected transcript(s) | targeting miRNA(s) |
|---|---|---|---|
| PRPF3 | 3 | ENST00000324862<br>ENST00000496202 | hsa-miR-329-3p |

## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| AC242988.1 | downstream gene variant, upstream gene variant | 835 | ENST00000624245 | ? | ENSP00000485440 | 5 |
| ECM1 | upstream gene variant, downstream gene variant | 976 | ENST00000496744 | ? | ? | 2 |
| ECM1 | upstream gene variant, downstream gene variant | 1207 | ENST00000490346 | ? | ? | 3 |
| ECM1 | upstream gene variant, downstream gene variant | 2448 | ENST00000369049 | NM_001202858.1 | ENSP00000358045 | 2 |
| ECM1 | upstream gene variant, downstream gene variant | 2213 | ENST00000369047 | NM_004425.3 | ENSP00000358043 | 2 |
| ECM1 | upstream gene variant, downstream gene variant | 382 | ENST00000498579 | ? | ? | 3 |
| ECM1 | upstream gene variant, downstream gene variant | 12 | ENST00000346569 | NM_022664.2 | ENSP00000271630 | 3 |
| ECM1 | upstream gene variant, downstream gene variant | 2449 | ENST00000470432 | ? | ? | 2 |
| FALEC | upstream gene variant, downstream gene variant | 4229 | ENST00000416894 | ? | ? | 2 |
| MRPS21 | downstream gene variant | 1144 | ENST00000614145 | NM_031901.5 | ENSP00000480129 | 1 |

| | | | | | | |
|---|---|---|---|---|---|---|
| MRPS21 | downstream gene variant | 1144 | ENST00000581066 | NM_018997.3 | ENSP00000461930 | 1 |
| PRPF3 | upstream gene variant, downstream gene variant | 1806 | ENST00000324862 | NM_004698.2 | ENSP00000315379 | 3 |
| PRPF3 | upstream gene variant, downstream gene variant | 225 | ENST00000470824 | ? | ? | 7 |
| PRPF3 | upstream gene variant, downstream gene variant | 967 | ENST00000493553 | ? | ? | 7 |
| PRPF3 | upstream gene variant, downstream gene variant | 1806 | ENST00000467329 | ? | ? | 7 |
| PRPF3 | upstream gene variant, downstream gene variant | 3321 | ENST00000496202 | ? | ? | 3 |
| PRPF3 | upstream gene variant, downstream gene variant | 3372 | ENST00000467514 | ? | ? | 5 |
| PRPF3 | upstream gene variant | 198 | ENST00000476970 | ? | ? | 1 |
| RPRD2 | upstream gene variant, downstream gene variant | 1444 | ENST00000369067 | ? | ENSP00000358063 | 2 |
| RPRD2 | upstream gene variant, downstream gene variant | 515 | ENST00000401000 | NM_001297674.1 | ENSP00000383785 | 3 |
| RPRD2 | downstream gene variant | 3525 | ENST00000492220 | ? | ? | 2 |
| RPRD2 | upstream gene variant, downstream gene variant | 1447 | ENST00000369068 | NM_015203.3 | ENSP00000358064 | 3 |
| TARS2 | upstream gene variant | 203 | ENST00000479372 | ? | ? | 2 |
| TARS2 | upstream gene variant, downstream gene variant | 178 | ENST00000369064 | NM_025150.4 | ENSP00000358060 | 4 |
| TARS2 | upstream gene variant, downstream gene variant | 175 | ENST00000369054 | NM_001271896.1 | ENSP00000358050 | 4 |
| TARS2 | upstream gene variant, downstream gene variant | 928 | ENST00000463555 | ? | ? | 3 |
| TARS2 | upstream gene variant, downstream gene variant | 450 | ENST00000460794 | ? | ? | 2 |
| TARS2 | upstream gene variant, downstream gene variant | 928 | ENST00000480070 | ? | ? | 3 |
| TARS2 | upstream gene variant | 752 | ENST00000466989 | ? | ? | 2 |
| TARS2 | upstream gene variant, downstream gene variant | 460 | ENST00000462578 | ? | ? | 2 |
| TARS2 | upstream gene variant, downstream gene variant | 631 | ENST00000467982 | ? | ENSP00000475551 | 4 |
| TARS2 | upstream gene variant, downstream gene variant | 181 | ENST00000606933 | NM_001271895.1 | ENSP00000475847 | 4 |
| TARS2 | upstream gene variant, downstream gene variant | 196 | ENST00000369051 | ? | ENSP00000358047 | 4 |
| TARS2 | upstream gene variant | 192 | ENST00000369053 | ? | ? | 2 |
| TARS2 | downstream gene variant | 1160 | ENST00000483046 | ? | ? | 2 |
| TARS2 | upstream gene variant, downstream gene variant | 125 | ENST00000438568 | ? | ENSP00000415002 | 4 |

## Putative effect on transcript

### Synonymous coding variant

| gene | affected transcript | RefSeq id | protein | AA's | exchanged codons | variant(s) |
|---|---|---|---|---|---|---|
| ECM1 | ENST00000369049 | NM_001202858.1 | ENSP00000358045 | T | acA/acG | 1 |
| ECM1 | ENST00000369047 | NM_004425.3 | ENSP00000358043 | T | acA/acG | 1 |

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| ECM1 | ENST00000346569 | NM_022664.2 | ENSP00000271630 | 1 |
| MRPS21 | ENST00000581066 | NM_018997.3 | ENSP00000461930 | 4 |
| MRPS21 | ENST00000614145 | NM_031901.5 | ENSP00000480129 | 4 |
| PRPF3 | ENST00000324862 | NM_004698.2 | ENSP00000315379 | 15 |
| PRPF3 | ENST00000467329 | ? | ? | 10 |
| PRPF3 | ENST00000467514 | ? | ? | 3 |

| | | | | |
|---|---|---|---|---|
| PRPF3 | ENST00000476970 | ? | ? | 4 |
| PRPF3 | ENST00000496202 | ? | ? | 8 |
| RPRD2 | ENST00000401000 | NM_001297674.1 | ENSP00000383785 | 27 |
| RPRD2 | ENST00000369068 | NM_015203.3 | ENSP00000358064 | 27 |
| RPRD2 | ENST00000369067 | ? | ENSP00000358063 | 19 |
| RPRD2 | ENST00000492220 | ? | ? | 27 |
| TARS2 | ENST00000606933 | NM_001271895.1 | ENSP00000475847 | 2 |
| TARS2 | ENST00000480070 | ? | ? | 1 |
| TARS2 | ENST00000483046 | ? | ? | 1 |
| TARS2 | ENST00000369064 | NM_025150.4 | ENSP00000358060 | 2 |
| TARS2 | ENST00000369051 | ? | ENSP00000358047 | 2 |
| TARS2 | ENST00000467982 | ? | ENSP00000475551 | 2 |
| TARS2 | ENST00000438568 | ? | ENSP00000415002 | 2 |
| TARS2 | ENST00000463555 | ? | ? | 1 |
| TARS2 | ENST00000369054 | NM_001271896.1 | ENSP00000358050 | 2 |

## 5'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| ECM1 | ENST00000369049 | NM_001202858.1 | ENSP00000358045 | 1 |
| ECM1 | ENST00000369047 | NM_004425.3 | ENSP00000358043 | 1 |

## Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| ECM1 | ENST00000496744 | ? | 1 |
| ECM1 | ENST00000470432 | ? | 2 |
| ECM1 | ENST00000498579 | ? | 1 |
| FALEC | ENST00000416894 | ? | 1 |
| RPRD2 | ENST00000492220 | ? | 1 |