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|------------------|--|----------|-----------------|--|
| TUBB <i>e!</i> | CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS [...] | OMIM | MIM:615771 | |
| ZFP57 <i>e!</i> | Diabetes Mellitus, 6q24-Related Transient Neonatal | DECIPHER | MIM:601410 | |
| NDUFS1 <i>e!</i> | Leigh Syndrome (nuclear DNA mutation) | DECIPHER | MIM:256000 | |
| NDUFS1 <i>e!</i> | Mitochondrial Respiratory Chain Complex I Deficiency (nuclear genes) | DECIPHER | MIM:252010 | |
| ZFP57 <i>e!</i> | Transient neonatal diabetes mellitus | OrphaNet | OrphaNet:99886 | |
| HLA-A <i>e!</i> | Birdshot chorioretinopathy | OrphaNet | OrphaNet:179 | |
| NDUFS1 <i>e!</i> | Isolated NADH-CoQ reductase deficiency | OrphaNet | OrphaNet:2609 | |
| NDUFS1 <i>e!</i> | Leigh syndrome with leukodystrophy | OrphaNet | OrphaNet:255241 | |
| C2 <i>e!</i> | Immunodeficiency due to an early component of complement deficiency | OrphaNet | OrphaNet:169147 | |
| CFB <i>e!</i> | Atypical hemolytic uremic syndrome with B factor anomaly | OrphaNet | OrphaNet:93578 | |
| HLA-B <i>e!</i> | Takayasu arteritis | OrphaNet | OrphaNet:3287 | |
| HLA-B <i>e!</i> | Stevens-Johnson syndrome | OrphaNet | OrphaNet:36426 | |
| HLA-B <i>e!</i> | Behcet disease | OrphaNet | OrphaNet:117 | |

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) |
|-------------------|------------------|---------------------------|----------------|---------------------------|----------------|------------------|-----------------|---------------------|------------|
| PPP1R18 <i>e!</i> | missense variant | ENST00000615527 <i>e!</i> | ? | ENSP00000480270 <i>e!</i> | R/G | Agg/Ggg | ? | ? | 1 |
| PPP1R18 <i>e!</i> | missense variant | ENST00000274853 <i>e!</i> | NM_133471.3 | ENSP00000274853 <i>e!</i> | R/G | Agg/Ggg | ? | ? | 1 |
| PPP1R18 <i>e!</i> | missense variant | ENST00000399199 <i>e!</i> | NM_001134870.1 | ENSP00000382150 <i>e!</i> | R/G | Agg/Ggg | ? | ? | 1 |

Direct effect on regulation

cis-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|-----------------------|---------------------------|------------------------|-----------|---------------------------------|-------------------|------------|
| ? | ? | ILMN_1654987 <i>e!</i> | blood | 3.60×10 ⁻⁶ (p-value) | MuTHER consortium | 58 |
| | | | adipocyte | 2.42×10 ⁻⁶ (p-value) | | |
| HLA-L <i>e!</i> | ENST00000463348 <i>e!</i> | ILMN_1675282 <i>e!</i> | blood | 1.92×10 ⁻⁶ (p-value) | MuTHER consortium | 47 |
| ZNRD1 <i>e!</i> | ENST00000471008 <i>e!</i> | ILMN_2398587 <i>e!</i> | blood | 2.39×10 ⁻⁶ (p-value) | MuTHER consortium | 44 |
| ZNRD1 <i>e!</i> | ENST00000359374 <i>e!</i> | | | | | |
| ZNRD1-AS1_3 <i>e!</i> | ENST00000611686 <i>e!</i> | | | | | |
| ZNRD1 <i>e!</i> | ENST00000332435 <i>e!</i> | | | | | |
| ZNRD1 <i>e!</i> | ENST00000376782 <i>e!</i> | | | | | |
| ZNRD1 <i>e!</i> | ENST00000376785 <i>e!</i> | | | | | |
| ZNRD1 <i>e!</i> | ENST00000463141 <i>e!</i> | | | | | |
| TRIM31 <i>e!</i> | ENST00000376734 <i>e!</i> | ILMN_1748685 <i>e!</i> | skin | 4.22×10 ⁻⁶ (p-value) | MuTHER consortium | 25 |
| TRIM31 <i>e!</i> | ENST00000484583 <i>e!</i> | | | | | |
| TRIM31 <i>e!</i> | ENST00000471695 <i>e!</i> | | | | | |
| TRIM31 <i>e!</i> | ENST00000468264 <i>e!</i> | | | | | |
| HLA-V <i>e!</i> | ENST00000446817 <i>e!</i> | ILMN_1660923 <i>e!</i> | blood | 1.32×10 ⁻⁹ (p-value) | MuTHER consortium | 59 |
| HLA-V <i>e!</i> | ENST00000429037 <i>e!</i> | | adipocyte | 1.91×10 ⁻⁶ (p-value) | MuTHER consortium | 59 |
| | | | skin | 4.10×10 ⁻⁶ (p-value) | MuTHER consortium | 59 |
| HCG22 <i>e!</i> | ENST00000570223 <i>e!</i> | ILMN_1667229 <i>e!</i> | blood | 2.57×10 ⁻⁶ (p-value) | MuTHER consortium | 40 |
| HCG22 <i>e!</i> | ENST00000565192 <i>e!</i> | | | | | |
| HCG22 <i>e!</i> | ENST00000615046 <i>e!</i> | | | | | |
| HCG22 <i>e!</i> | ENST00000426185 <i>e!</i> | | | | | |
| HCG22 <i>e!</i> | ENST00000562344 <i>e!</i> | | | | | |

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|---------------------|---------------------------|---------------------------|-------------------------|----------------------------------|-----------------------------|-----|
| GABBR1 <i>e!</i> | ENST00000491829 <i>e!</i> | ILMN_2395375 <i>e!</i> | blood | 1.94×10 ⁻⁵ (p-value) | MuTHER consortium <i>IM</i> | 18 |
| GABBR1 <i>e!</i> | ENST00000377012 <i>e!</i> | | | | | |
| GABBR1 <i>e!</i> | ENST00000377034 <i>e!</i> | | | | | |
| GABBR1 <i>e!</i> | ENST00000478931 <i>e!</i> | | | | | |
| GABBR1 <i>e!</i> | ENST00000472823 <i>e!</i> | | | | | |
| GABBR1 <i>e!</i> | ENST00000377016 <i>e!</i> | | | | | |
| VARS2 <i>e!</i> | ENST00000321897 <i>e!</i> | ILMN_1737585 <i>e!</i> | skin | 2.79×10 ⁻⁷ (p-value) | MuTHER consortium <i>IM</i> | 36 |
| VARS2 <i>e!</i> | ENST00000625423 <i>e!</i> | | | | | |
| VARS2 <i>e!</i> | ENST00000541562 <i>e!</i> | | | | | |
| VARS2 <i>e!</i> | ENST00000542001 <i>e!</i> | | | | | |
| VARS2 <i>e!</i> | ENST00000469358 <i>e!</i> | | | | | |
| VARS2 <i>e!</i> | ENST00000477288 <i>e!</i> | | | | | |
| VARS2 <i>e!</i> | ENST00000473916 <i>e!</i> | | | | | |
| VARS2 <i>e!</i> | ENST00000476162 <i>e!</i> | | | | | |
| HCP5 <i>e!</i> | ENST00000414046 <i>e!</i> | ILMN_1803945 <i>e!</i> | skin | 5.42×10 ⁻⁹ (p-value) | MuTHER consortium <i>IM</i> | 18 |
| HCP5 <i>e!</i> | ENST00000541196 <i>e!</i> | | | | | |
| MICB <i>e!</i> | ENST00000538442 <i>e!</i> | ILMN_1708006 <i>e!</i> | skin | 5.95×10 ⁻⁹ (p-value) | MuTHER consortium <i>IM</i> | 18 |
| MICB <i>e!</i> | ENST00000399150 <i>e!</i> | | | | | |
| MICB <i>e!</i> | ENST00000252229 <i>e!</i> | | | | | |
| IER3 <i>e!</i> | ENST00000376377 <i>e!</i> | ILMN_1682717 <i>e!</i> | skin | 2.84×10 ⁻⁵ (p-value) | MuTHER consortium <i>IM</i> | 1 |
| IER3 <i>e!</i> | ENST00000259874 <i>e!</i> | | | | | |
| ZFP57 <i>e!</i> | ? | ENSG00000204644 <i>e!</i> | muscularis mucosae | 6.78×10 ⁻⁸ (p-value) | GTEX Portal V6 <i>IM</i> | 82 |
| PPP1R18 <i>e!</i> | ? | ENSG00000146112 <i>e!</i> | muscularis mucosae | 2.64×10 ⁻⁶ (p-value) | GTEX Portal V6 <i>IM</i> | 22 |
| HLA-C <i>e!</i> | ? | ENSG00000204525 <i>e!</i> | muscularis mucosae | 1.12×10 ⁻⁶ (p-value) | GTEX Portal V6 <i>IM</i> | 7 |
| HLA-B <i>e!</i> | ? | ENSG00000234745 <i>e!</i> | muscularis mucosae | 1.89×10 ⁻⁶ (p-value) | GTEX Portal V6 <i>IM</i> | 3 |
| FLOT1 <i>e!</i> | ? | ENSG00000137312 <i>e!</i> | lung | 6.00×10 ⁻⁸ (p-value) | GTEX Portal V6 <i>IM</i> | 91 |
| HLA-J <i>e!</i> | ? | ENSG00000204622 <i>e!</i> | lung | 1.58×10 ⁻¹² (p-value) | GTEX Portal V6 <i>IM</i> | 104 |
| HLA-G <i>e!</i> | ? | ENSG00000204632 <i>e!</i> | lung | 2.21×10 ⁻⁷ (p-value) | GTEX Portal V6 <i>IM</i> | 89 |
| HLA-H <i>e!</i> | ? | ENSG00000206341 <i>e!</i> | lung | 2.13×10 ⁻⁸ (p-value) | GTEX Portal V6 <i>IM</i> | 101 |
| ZFP57 <i>e!</i> | ? | ENSG00000204644 <i>e!</i> | lung | 6.53×10 ⁻¹¹ (p-value) | GTEX Portal V6 <i>IM</i> | 82 |
| LINC00243 <i>e!</i> | ? | ENSG00000214894 <i>e!</i> | lung | 4.13×10 ⁻⁶ (p-value) | GTEX Portal V6 <i>IM</i> | 4 |
| HLA-J <i>e!</i> | ? | ENSG00000204622 <i>e!</i> | atrial appendage | 1.61×10 ⁻⁶ (p-value) | GTEX Portal V6 <i>IM</i> | 4 |
| ZFP57 <i>e!</i> | ? | ENSG00000204644 <i>e!</i> | atrial appendage | 6.27×10 ⁻⁸ (p-value) | GTEX Portal V6 <i>IM</i> | 54 |
| ZFP57 <i>e!</i> | ? | ENSG00000204644 <i>e!</i> | transformed fibroblasts | 3.56×10 ⁻⁸ (p-value) | GTEX Portal V6 <i>IM</i> | 81 |
| HLA-J <i>e!</i> | ? | ENSG00000204622 <i>e!</i> | transformed fibroblasts | 3.90×10 ⁻⁶ (p-value) | GTEX Portal V6 <i>IM</i> | 44 |
| HLA-C <i>e!</i> | ? | ENSG00000204525 <i>e!</i> | transformed fibroblasts | 7.51×10 ⁻⁸ (p-value) | GTEX Portal V6 <i>IM</i> | 75 |
| HCG20 <i>e!</i> | ? | ENSG00000228022 <i>e!</i> | transformed fibroblasts | 2.40×10 ⁻⁷ (p-value) | GTEX Portal V6 <i>IM</i> | 10 |
| HLA-H <i>e!</i> | ? | ENSG00000206341 <i>e!</i> | transformed fibroblasts | 2.47×10 ⁻⁵ (p-value) | GTEX Portal V6 <i>IM</i> | 1 |
| HLA-J <i>e!</i> | ? | ENSG00000204622 <i>e!</i> | blood | 4.01×10 ⁻⁹ (p-value) | GTEX Portal V6 <i>IM</i> | 98 |
| HLA-H <i>e!</i> | ? | ENSG00000206341 <i>e!</i> | blood | 7.77×10 ⁻⁹ (p-value) | GTEX Portal V6 <i>IM</i> | 93 |
| ZFP57 <i>e!</i> | ? | ENSG00000204644 <i>e!</i> | blood | 2.24×10 ⁻¹⁰ (p-value) | GTEX Portal V6 <i>IM</i> | 83 |
| ? | ? | | blood | | | 52 |
| HLA-G <i>e!</i> | ? | ENSG00000204632 <i>e!</i> | blood | 9.33×10 ⁻⁸ (p-value) | GTEX Portal V6 <i>IM</i> | |
| HLA-F-AS1 <i>e!</i> | ? | ENSG00000214922 <i>e!</i> | blood | 1.37×10 ⁻⁶ (p-value) | GTEX Portal V6 <i>IM</i> | 70 |

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|-----------|-----------|---|-----------------|-----------|------------------|----------------------------------|---------------|---|-----|
| LINC00243 | <i>e!</i> | ? | ENSG00000214894 | <i>e!</i> | blood | 2.67×10 ⁻¹⁴ (p-value) | GTE Portal V6 |  | 103 |
| HLA-K | <i>e!</i> | ? | ENSG00000230795 | <i>e!</i> | blood | 1.57×10 ⁻⁹ (p-value) | GTE Portal V6 |  | 92 |
| CCHCR1 | <i>e!</i> | ? | ENSG00000204536 | <i>e!</i> | blood | 4.75×10 ⁻⁶ (p-value) | GTE Portal V6 |  | 6 |
| HLA-C | <i>e!</i> | ? | ENSG00000204525 | <i>e!</i> | blood | 8.58×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 16 |
| VARS2 | <i>e!</i> | ? | ENSG00000137411 | <i>e!</i> | blood | 6.90×10 ⁻⁶ (p-value) | GTE Portal V6 |  | 1 |
| HLA-J | <i>e!</i> | ? | ENSG00000204622 | <i>e!</i> | breast | 1.40×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 27 |
| ZFP57 | <i>e!</i> | ? | ENSG00000204644 | <i>e!</i> | breast | 6.42×10 ⁻⁸ (p-value) | GTE Portal V6 |  | 47 |
| HLA-C | <i>e!</i> | ? | ENSG00000204525 | <i>e!</i> | breast | 3.52×10 ⁻⁶ (p-value) | GTE Portal V6 |  | 5 |
| HLA-U | <i>e!</i> | ? | ENSG00000228078 | <i>e!</i> | tibial artery | 5.57×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 72 |
| HLA-J | <i>e!</i> | ? | ENSG00000204622 | <i>e!</i> | tibial artery | 3.63×10 ⁻⁸ (p-value) | GTE Portal V6 |  | 33 |
| HLA-H | <i>e!</i> | ? | ENSG00000206341 | <i>e!</i> | tibial artery | 5.15×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 39 |
| ZFP57 | <i>e!</i> | ? | ENSG00000204644 | <i>e!</i> | tibial artery | 2.39×10 ⁻⁹ (p-value) | GTE Portal V6 |  | 83 |
| HLA-C | <i>e!</i> | ? | ENSG00000204525 | <i>e!</i> | tibial artery | 1.92×10 ⁻⁹ (p-value) | GTE Portal V6 |  | 82 |
| HLA-J | <i>e!</i> | ? | ENSG00000204622 | <i>e!</i> | thyroid | 6.77×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 39 |
| ZFP57 | <i>e!</i> | ? | ENSG00000204644 | <i>e!</i> | thyroid | 2.65×10 ⁻¹⁰ (p-value) | GTE Portal V6 |  | 83 |
| ZNRD1 | <i>e!</i> | ? | ENSG00000066379 | <i>e!</i> | thyroid | 1.59×10 ⁻⁵ (p-value) | GTE Portal V6 |  | 1 |
| FLOT1 | <i>e!</i> | ? | ENSG00000137312 | <i>e!</i> | thyroid | 2.35×10 ⁻¹⁰ (p-value) | GTE Portal V6 |  | 96 |
| CCHCR1 | <i>e!</i> | ? | ENSG00000204536 | <i>e!</i> | thyroid | 8.47×10 ⁻¹⁰ (p-value) | GTE Portal V6 |  | 93 |
| PSORS1C1 | <i>e!</i> | ? | ENSG00000204540 | <i>e!</i> | thyroid | 5.65×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 11 |
| PPP1R18 | <i>e!</i> | ? | ENSG00000146112 | <i>e!</i> | thyroid | 1.44×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 36 |
| HLA-J | <i>e!</i> | ? | ENSG00000204622 | <i>e!</i> | skeletal muscle | 1.24×10 ⁻⁹ (p-value) | GTE Portal V6 |  | 103 |
| HLA-H | <i>e!</i> | ? | ENSG00000206341 | <i>e!</i> | skeletal muscle | 1.43×10 ⁻⁶ (p-value) | GTE Portal V6 |  | 58 |
| ZFP57 | <i>e!</i> | ? | ENSG00000204644 | <i>e!</i> | skeletal muscle | 1.78×10 ⁻⁶ (p-value) | GTE Portal V6 |  | 23 |
| HLA-C | <i>e!</i> | ? | ENSG00000204525 | <i>e!</i> | skeletal muscle | 1.59×10 ⁻¹² (p-value) | GTE Portal V6 |  | 82 |
| ZFP57 | <i>e!</i> | ? | ENSG00000204644 | <i>e!</i> | liver | 2.45×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 5 |
| HLA-J | <i>e!</i> | ? | ENSG00000204622 | <i>e!</i> | sun exposed skin | 2.03×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 20 |
| HLA-H | <i>e!</i> | ? | ENSG00000206341 | <i>e!</i> | sun exposed skin | 9.76×10 ⁻⁶ (p-value) | GTE Portal V6 |  | 2 |
| ZFP57 | <i>e!</i> | ? | ENSG00000204644 | <i>e!</i> | sun exposed skin | 9.47×10 ⁻¹⁰ (p-value) | GTE Portal V6 |  | 82 |
| VARS2 | <i>e!</i> | ? | ENSG00000137411 | <i>e!</i> | sun exposed skin | 6.86×10 ⁻⁹ (p-value) | GTE Portal V6 |  | 42 |
| HLA-C | <i>e!</i> | ? | ENSG00000204525 | <i>e!</i> | sun exposed skin | 3.64×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 26 |
| ZFP57 | <i>e!</i> | ? | ENSG00000204644 | <i>e!</i> | unexposed skin | 1.63×10 ⁻⁶ (p-value) | GTE Portal V6 |  | 4 |
| ZFP57 | <i>e!</i> | ? | ENSG00000204644 | <i>e!</i> | left ventricle | 5.64×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 6 |
| PSORS1C1 | <i>e!</i> | ? | ENSG00000204540 | <i>e!</i> | left ventricle | 3.69×10 ⁻⁷ (p-value) | GTE Portal V6 |  | 9 |
| HLA-H | <i>e!</i> | ? | ENSG00000206341 | <i>e!</i> | left ventricle | 8.80×10 ⁻⁶ (p-value) | GTE Portal V6 |  | 1 |
| ZFP57 | <i>e!</i> | ? | ENSG00000204644 | <i>e!</i> | aorta | 5.31×10 ⁻¹¹ (p-value) | GTE Portal V6 |  | 83 |
| PPP1R18 | <i>e!</i> | ? | ENSG00000146112 | <i>e!</i> | aorta | 5.47×10 ⁻⁸ (p-value) | GTE Portal V6 |  | 77 |
| HLA-B | <i>e!</i> | ? | ENSG00000234745 | <i>e!</i> | aorta | 6.92×10 ⁻⁶ (p-value) | GTE Portal V6 |  | 1 |

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|---------|----|-----------------|-----------------|--------------|-------------------------|----------------------------------|----------------------------------|---|---|---|
| HCG17 | e! | ? | ENSG00000270604 | e! | subcutaneous adipocytes | 3.78×10 ⁻⁸ (p-value) | GTEEx Portal V6 |  | 70 | |
| ZFP57 | e! | ? | ENSG00000204644 | e! | subcutaneous adipocytes | 2.47×10 ⁻⁹ (p-value) | GTEEx Portal V6 |  | 82 | |
| HLA-C | e! | ? | ENSG00000204525 | e! | subcutaneous adipocytes | 6.58×10 ⁻⁷ (p-value) | GTEEx Portal V6 |  | 57 | |
| VARS2 | e! | ? | ENSG00000137411 | e! | subcutaneous adipocytes | 6.34×10 ⁻⁸ (p-value) | GTEEx Portal V6 |  | 49 | |
| CCHCR1 | e! | ? | ENSG00000204536 | e! | subcutaneous adipocytes | 6.45×10 ⁻⁹ (p-value) | GTEEx Portal V6 |  | 46 | |
| ZFP57 | e! | ? | ENSG00000204644 | e! | stomach | 7.56×10 ⁻⁷ (p-value) | GTEEx Portal V6 |  | 65 | |
| HLA-C | e! | ? | ENSG00000204525 | e! | stomach | 1.66×10 ⁻⁷ (p-value) | GTEEx Portal V6 |  | 76 | |
| ZFP57 | e! | ? | ENSG00000204644 | e! | visceral adipocytes | 1.11×10 ⁻⁸ (p-value) | GTEEx Portal V6 |  | 22 | |
| CCHCR1 | e! | ? | ENSG00000204536 | e! | visceral adipocytes | 2.13×10 ⁻⁶ (p-value) | GTEEx Portal V6 |  | 7 | |
| HLA-H | e! | ? | ENSG00000206341 | e! | spleen | 3.40×10 ⁻⁶ (p-value) | GTEEx Portal V6 |  | 4 | |
| ZFP57 | e! | ? | ENSG00000204644 | e! | tibial nerve | 4.33×10 ⁻⁹ (p-value) | GTEEx Portal V6 |  | 81 | |
| CCHCR1 | e! | ? | ENSG00000204536 | e! | tibial nerve | 1.48×10 ⁻⁷ (p-value) | GTEEx Portal V6 |  | 77 | |
| PPP1R18 | e! | ? | ENSG00000146112 | e! | tibial nerve | 2.17×10 ⁻⁶ (p-value) | GTEEx Portal V6 |  | 5 | |
| HLA-C | e! | ? | ENSG00000204525 | e! | tibial nerve | 4.29×10 ⁻⁶ (p-value) | GTEEx Portal V6 |  | 34 | |
| HLA-J | e! | ? | ENSG00000204622 | e! | esophagus mucosa | 1.95×10 ⁻⁷ (p-value) | GTEEx Portal V6 |  | 31 | |
| ZFP57 | e! | ? | ENSG00000204644 | e! | esophagus mucosa | 1.13×10 ⁻¹² (p-value) | GTEEx Portal V6 |  | 83 | |
| HLA-C | e! | ? | ENSG00000204525 | e! | esophagus mucosa | 2.41×10 ⁻⁸ (p-value) | GTEEx Portal V6 |  | 68 | |
| MICB | e! | ? | ENSG00000204516 | e! | esophagus mucosa | 4.51×10 ⁻⁶ (p-value) | GTEEx Portal V6 |  | 4 | |
| HLA-A | e! | ? | ENSG00000206503 | e! | brain | 3.82×10 ⁻⁶ (q-value) | SeeQTL DB (Myers et al.) |  | 4 | |
| ? | | ? | ILMN_1679389 | e! | monocyte | 1.31×10 ⁻¹⁴ (p-value) | Zeller et al. |  | 8 | |
| ? | | ? | ILMN_1666078 | e! | monocyte | 1.81×10 ⁻³⁷ (p-value) | Zeller et al. |  | 8 | |
| ? | | ? | ILMN_1671054 | e! | monocyte | 1.64×10 ⁻⁴⁵ (p-value) | Zeller et al. |  | 8 | |
| HLA-G | e! | ENST00000376828 | e! | ILMN_1656670 | e! | monocyte | 3.91×10 ⁻¹⁹ (p-value) | Zeller et al. |  | 8 |
| HLA-G | e! | ENST00000360323 | e! | | | | | | | |
| HLA-G | e! | ENST00000478519 | e! | | | | | | | |
| HLA-G | e! | ENST00000478355 | e! | | | | | | | |
| HLA-G | e! | ENST00000428701 | e! | | | | | | | |
| HLA-G | e! | ENST00000376815 | e! | | | | | | | |
| HLA-G | e! | ENST00000376818 | e! | | | | | | | |
| BTN3A2 | e! | ENST00000396934 | e! | ILMN_1676528 | e! | monocyte | 1.68×10 ⁻⁴⁰ (p-value) | Zeller et al. |  | 8 |
| BTN3A2 | e! | ENST00000508906 | e! | | | | | | | |
| BTN3A2 | e! | ENST00000356386 | e! | | | | | | | |
| ? | | ? | ILMN_1820787 | e! | monocyte | 2.53×10 ⁻²⁴ (p-value) | Zeller et al. |  | 8 | |
| BTN3A2 | e! | ENST00000524459 | e! | ILMN_1700067 | e! | monocyte | 8.29×10 ⁻²⁸ (p-value) | Zeller et al. |  | 8 |
| BTN3A2 | e! | ENST00000528541 | e! | | | | | | | |
| BTN3A3 | e! | ENST00000467524 | e! | | | | | | | |
| BTN3A3 | e! | ENST00000482451 | e! | | | | | | | |
| BTN3A3 | e! | ENST00000494393 | e! | | | | | | | |
| BTN3A2 | e! | ENST00000527417 | e! | | | | | | | |
| BTN3A3 | e! | ENST00000244519 | e! | | | | | | | |
| BTN3A2 | e! | ENST00000528222 | e! | | | | | | | |
| BTN3A3 | e! | ENST00000474790 | e! | | | | | | | |
| BTN3A2 | e! | ENST00000377708 | e! | | | | | | | |

| | | | | | | | | | |
|--------------|----|-----------------|----|--------------|----|----------|----------------------------------|----------------|--|
| BTN3A3 | e! | ENST00000496719 | e! | | | | | | |
| BTN3A2 | e! | ENST00000396948 | e! | | | | | | |
| BTN3A2 | e! | ENST00000527422 | e! | | | | | | |
| BTN3A2 | e! | ENST00000356386 | e! | | | | | | |
| ? | | ? | | ILMN_1883997 | e! | monocyte | 1.74×10 ⁻⁴⁴ (p-value) | Zeller et al. |  8 |
| HLA-C | e! | ENST00000470363 | e! | ILMN_2150787 | e! | monocyte | 3.78×10 ⁻²³ (p-value) | Zeller et al. |  7 |
| HLA-C | e! | ENST00000383329 | e! | | | | | | |
| HLA-C | e! | ENST00000487245 | e! | | | | | | |
| HLA-C | e! | ENST00000376228 | e! | | | | | | |
| HLA-C | e! | ENST00000376237 | e! | | | | | | |
| HLA-C | e! | ENST00000620806 | e! | | | | | | |
| HLA-C | e! | ENST00000466892 | e! | | | | | | |
| ? | | ? | | ILMN_1893395 | e! | monocyte | 1.16×10 ⁻¹⁴ (p-value) | Zeller et al. |  1 |
| HIST1H3I | e! | ENST00000616365 | e! | ILMN_2207865 | e! | b-cell | 1.43×10 ⁻⁴ (p-value) | Fairfax et al. |  8 |
| ZNRD1 | e! | ENST00000471008 | e! | ILMN_2398587 | e! | b-cell | 2.62×10 ⁻⁴ (p-value) | Fairfax et al. |  2 |
| ZNRD1 | e! | ENST00000359374 | e! | | | | | | |
| ZNRD1-AS1_3 | e! | ENST00000611686 | e! | | | | | | |
| ZNRD1 | e! | ENST00000376785 | e! | | | | | | |
| ZNRD1 | e! | ENST00000376782 | e! | | | | | | |
| ZNRD1 | e! | ENST00000332435 | e! | | | | | | |
| ZNRD1 | e! | ENST00000463141 | e! | | | | | | |
| C2 | e! | ENST00000486124 | e! | ILMN_1710740 | e! | monocyte | 7.85×10 ⁻⁶ (p-value) | Fairfax et al. |  2 |
| C2 | e! | ENST00000469372 | e! | | | | | | |
| CFB | e! | ENST00000425368 | e! | | | | | | |
| C2 | e! | ENST00000497706 | e! | | | | | | |
| C2 | e! | ENST00000442278 | e! | | | | | | |
| C2 | e! | ENST00000452323 | e! | | | | | | |
| C2 | e! | ENST00000383177 | e! | | | | | | |
| C2 | e! | ENST00000299367 | e! | | | | | | |
| C2 | e! | ENST00000482060 | e! | | | | | | |
| ? | | ? | | ILMN_1679389 | e! | b-cell | 4.36×10 ⁻⁶ (p-value) | Fairfax et al. |  17 |
| IER3 | e! | ENST00000376377 | e! | ILMN_1682717 | e! | monocyte | 2.83×10 ⁻⁵ (p-value) | Fairfax et al. |  20 |
| IER3 | e! | ENST00000259874 | e! | | | | | | |
| RPP21 | e! | ENST00000489124 | e! | ILMN_2186597 | e! | b-cell | 4.61×10 ⁻⁴ (p-value) | Fairfax et al. |  1 |
| RPP21 | e! | ENST00000466327 | e! | | | | | | |
| RPP21 | e! | ENST00000436442 | e! | | | | | | |
| TRIM39-RPP21 | e! | ENST00000623385 | e! | | | | | | |
| RPP21 | e! | ENST00000473266 | e! | | | | | | |
| TRIM39-RPP21 | e! | ENST00000513556 | e! | | | | | | |
| RPP21 | e! | ENST00000498414 | e! | | | | | | |
| RPP21 | e! | ENST00000433076 | e! | | | | | | |
| RPP21 | e! | ENST00000442966 | e! | | | | | | |
| RPP21 | e! | ENST00000491477 | e! | | | | | | |
| RPP21 | e! | ENST00000428040 | e! | | | | | | |
| OR5V1 | e! | ENST00000543825 | e! | ILMN_1683352 | e! | b-cell | 3.80×10 ⁻⁶ (p-value) | Fairfax et al. |  10 |
| OR5V1 | e! | ENST00000377154 | e! | | | | | | |
| VARS2 | e! | ENST00000321897 | e! | ILMN_1737585 | e! | monocyte | 5.36×10 ⁻⁷ (p-value) | Fairfax et al. |  2 |
| VARS2 | e! | ENST00000625423 | e! | | | b-cell | 5.12×10 ⁻⁵ (p-value) | Fairfax et al. |  2 |
| VARS2 | e! | ENST00000542001 | e! | | | | | | |
| VARS2 | e! | ENST00000541562 | e! | | | | | | |
| VARS2 | e! | ENST00000469358 | e! | | | | | | |
| VARS2 | e! | ENST00000477320 | e! | | | | | | |

| | | | | | | | |
|--------------------|---------------------------|---------------------------|-----------------------|---------------------------------|---------------------------|----|--|
| VARS2 <i>e!</i> | ENST00000477288 <i>e!</i> | | | | | | |
| VARS2 <i>e!</i> | ENST00000473916 <i>e!</i> | | | | | | |
| VARS2 <i>e!</i> | ENST00000476162 <i>e!</i> | | | | | | |
| ZFP57 <i>e!</i> | ? | ENSG00000204644 <i>e!</i> | transverse colon | 2.63×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 2 | |
| HLA-C <i>e!</i> | ? | ENSG00000204525 <i>e!</i> | transverse colon | 2.66×10 ⁻⁸ (p-value) | GTEEx Portal V6 <i>!M</i> | 66 | |
| TRIM26BP <i>e!</i> | ? | ENSG00000236475 <i>e!</i> | transverse colon | 2.59×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 19 | |
| CCHCR1 <i>e!</i> | ? | ENSG00000204536 <i>e!</i> | transverse colon | 1.82×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 3 | |
| ZFP57 <i>e!</i> | ? | ENSG00000204644 <i>e!</i> | coronary artery | 2.82×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 2 | |
| HLA-J <i>e!</i> | ? | ENSG00000204622 <i>e!</i> | coronary artery | 2.14×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 20 | |
| HLA-C <i>e!</i> | ENST00000470363 <i>e!</i> | 216526_x_at <i>e!</i> | blood | 1.50×10 ⁻⁸ (p-value) | Dixon et al. <i>!M</i> | 4 | |
| HLA-C <i>e!</i> | ENST00000383329 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000487245 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000376228 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000466892 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000376237 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000620806 <i>e!</i> | | | | | | |
| RNF39 <i>e!</i> | ? | ENSG00000204618 <i>e!</i> | pancreas | 2.26×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 1 | |
| WASF5P <i>e!</i> | ? | ENSG00000231402 <i>e!</i> | pancreas | 3.25×10 ⁻⁸ (p-value) | GTEEx Portal V6 <i>!M</i> | 42 | |
| HLA-C <i>e!</i> | ? | ENSG00000204525 <i>e!</i> | pancreas | 1.65×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 4 | |
| MICB <i>e!</i> | ? | ENSG00000204516 <i>e!</i> | cerebellar hemisphere | 1.84×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 1 | |
| MICB <i>e!</i> | ? | ENSG00000204516 <i>e!</i> | cerebellum | 1.25×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 1 | |
| HLA-G <i>e!</i> | ? | ENSG00000204632 <i>e!</i> | EBV lymphocytes | 1.79×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 4 | |
| HCG17 <i>e!</i> | ? | ENSG00000270604 <i>e!</i> | hypothalamus | 1.06×10 ⁻⁶ (p-value) | GTEEx Portal V6 <i>!M</i> | 4 | |

trans-eQTL

| gene | transcript | probe | chromosome | tissue | min(statistic) (type) | source | variant(s) |
|-------------------|---------------------------|---------------------------|------------|----------|----------------------------------|------------------------------------|------------|
| HLA-A <i>e!</i> | ? | ENSG00000206503 <i>e!</i> | chr6 | brain | 6.61×10 ⁻⁴ (q-value) | SeeQTL DB (Myers et al.) <i>!M</i> | 4 |
| ? | ? | ILMN_1752592 <i>e!</i> | chr6 | monocyte | 8.64×10 ⁻²⁹ (p-value) | Zeller et al. <i>!M</i> | 8 |
| HLA-C <i>e!</i> | ENST00000470363 <i>e!</i> | ILMN_2150787 <i>e!</i> | chr6 | monocyte | 2.91×10 ⁻¹⁹ (p-value) | Zeller et al. <i>!M</i> | 1 |
| HLA-C <i>e!</i> | ENST00000383329 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000487245 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000376228 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000376237 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000620806 <i>e!</i> | | | | | | |
| HLA-C <i>e!</i> | ENST00000466892 <i>e!</i> | | | | | | |
| ? | ? | ILMN_1893395 <i>e!</i> | chr6 | monocyte | 1.06×10 ⁻¹² (p-value) | Zeller et al. <i>!M</i> | 4 |
| ? | ? | ILMN_1715169 <i>e!</i> | chr6 | monocyte | 9.75×10 ⁻¹⁵ (p-value) | Zeller et al. <i>!M</i> | 6 |
| ? | ? | ILMN_1661266 <i>e!</i> | chr6 | monocyte | 4.95×10 ⁻¹⁵ (p-value) | Zeller et al. <i>!M</i> | 6 |
| TMEM154 <i>e!</i> | ENST00000304385 <i>e!</i> | ILMN_2088124 <i>e!</i> | chr4 | blood | 1.16×10 ⁻¹⁰ (p-value) | Westra et al. <i>!M</i> | 1 |
| NDUFS1 <i>e!</i> | ENST00000455934 <i>e!</i> | ILMN_1728810 <i>e!</i> | chr2 | blood | 5.94×10 ⁻⁸ (p-value) | Westra et al. <i>!M</i> | 1 |
| NDUFS1 <i>e!</i> | ENST00000233190 <i>e!</i> | | | | | | |
| NDUFS1 <i>e!</i> | ENST00000423725 <i>e!</i> | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000524682 <i>e!</i> | 209846_s_at <i>e!</i> | chr6 | blood | 3.10×10 ⁻¹¹ (p-value) | Dixon et al. <i>!M</i> | 6 |
| BTN3A2 <i>e!</i> | ENST00000604202 <i>e!</i> | | | | | | |

| | | | | | | | | |
|------------------|---------------------------|-----------------------|------|-------|----------------------------------|-----------------------|--|---|
| BTN3A2 <i>e!</i> | ENST00000396934 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000532294 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000508906 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000377708 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000527422 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000531055 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000396948 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000356386 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000396934 <i>e!</i> | 212613_at <i>e!</i> | chr6 | blood | 1.40×10 ⁻⁸ (p-value) | Dixon et al. <i>!</i> | | 3 |
| BTN3A2 <i>e!</i> | ENST00000508906 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000356386 <i>e!</i> | | | | | | | |
| BTN3A3 <i>e!</i> | ENST00000244519 <i>e!</i> | 204820_s_at <i>e!</i> | chr6 | blood | 3.20×10 ⁻¹⁰ (p-value) | Dixon et al. <i>!</i> | | 5 |
| BTN3A2 <i>e!</i> | ENST00000396934 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000508906 <i>e!</i> | | | | | | | |
| BTN3A2 <i>e!</i> | ENST00000356386 <i>e!</i> | | | | | | | |
| BTN3A3 <i>e!</i> | ENST00000480110 <i>e!</i> | | | | | | | |
| HLA-C <i>e!</i> | ENST00000470363 <i>e!</i> | 216526_x_at <i>e!</i> | chr6 | blood | 3.20×10 ⁻⁸ (p-value) | Dixon et al. <i>!</i> | | 1 |
| HLA-C <i>e!</i> | ENST00000383329 <i>e!</i> | | | | | | | |
| HLA-C <i>e!</i> | ENST00000487245 <i>e!</i> | | | | | | | |
| HLA-C <i>e!</i> | ENST00000376228 <i>e!</i> | | | | | | | |
| HLA-C <i>e!</i> | ENST00000466892 <i>e!</i> | | | | | | | |
| HLA-C <i>e!</i> | ENST00000376237 <i>e!</i> | | | | | | | |
| HLA-C <i>e!</i> | ENST00000620806 <i>e!</i> | | | | | | | |

Putative effect on regulation

Transcription factor binding site variation

| transcription factor | binding motif | motif position | highly informative position | score change | variant(s) |
|----------------------|---------------|----------------|-----------------------------|--------------|------------|
| JUND | MA0491.1 | 8 | yes | 0.000 | 1 |
| FOSL1 | MA0477.1 | 4 | yes | 0.000 | 1 |
| SP1 | MA0079.3 | 10 | no | 0.000 | 1 |

ENCODE promoter-associated DHS

| SNiPA promoter id | variant(s) | associated gene(s) |
|---------------------------|------------|--------------------|
| ENCP00000047900 <i>e!</i> | | HCG19P <i>e!</i> |
| | | HCG19P <i>e!</i> |
| | | HCG19P <i>e!</i> |
| | | HCG19P <i>e!</i> |
| | | HCG19P <i>e!</i> |
| | | HCG19P <i>e!</i> |
| ENCP00000047941 <i>e!</i> | | FLOT1 <i>e!</i> |
| | | FLOT1 <i>e!</i> |
| | | FLOT1 <i>e!</i> |
| | | FLOT1 <i>e!</i> |
| | | FLOT1 <i>e!</i> |
| | | FLOT1 <i>e!</i> |

ENCODE promoter-associated distal DHS (Enhancer)

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---------------------------|------------|------------------------------|--------------------------------|
| ENCE00000444288 <i>e!</i> | 1 | ENCP00000047932 | NRM <i>e!</i> NRM <i>e!</i> |

| | | | |
|---------------------------|---|-----------------|--|
| | | | NRM <i>e!</i> NRM <i>e!</i> NRM <i>e!</i> NRM <i>e!</i> NRM <i>e!</i> |
| | | ENCP00000047889 | TRIM26 <i>e!</i> TRIM26 <i>e!</i> TRIM26 <i>e!</i> TRIM26 <i>e!</i> TRIM26 <i>e!</i> TRIM26 <i>e!</i> TRIM26 <i>e!</i> |
| ENCE00000444247 <i>e!</i> | 1 | ENCP00000047877 | RNF39 <i>e!</i> RNF39 <i>e!</i> RNF39 <i>e!</i> RNF39 <i>e!</i> RNF39 <i>e!</i> RNF39 <i>e!</i> RNF39 <i>e!</i> |
| | | ENCP00000047957 | DDR1 <i>e!</i> DDR1 <i>e!</i> DDR1 <i>e!</i> DDR1 <i>e!</i> DDR1 <i>e!</i> DDR1 <i>e!</i> |
| | | ENCP00000047879 | TRIM31 <i>e!</i> TRIM31 <i>e!</i> TRIM31 <i>e!</i> TRIM31 <i>e!</i> TRIM31 <i>e!</i> TRIM31 <i>e!</i> TRIM31 <i>e!</i> |
| ENCE00000444553 <i>e!</i> | 1 | ENCP00000047941 | FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i> |
| ENCE00000444465 <i>e!</i> | 1 | ENCP00000047921 | ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> |

Regulatory feature cluster



| element id | variant(s) | tissue/cell | factors |
|---|------------|--|---|
| ENSR00000487584 <i>e!</i> (CTCF binding site) | 1 | monocytes (Monocytes-CD14+) embryonic stem cell (H1ESC) endothelium (HUVEC) skin (NHDF-AD) | H3K27me3 H3K27me3 H3K27me3 DNase1 |
| ENSR00000487598 <i>e!</i> (promoter flanking region) | 2 | NHLF embryonic stem cell (H1ESC) HSMMtube blood (K562) blood (DND-41) monocytes (Monocytes-CD14+) endothelium (HUVEC) liver (HepG2) lung (IMR90) nervous (NH-A) | H3K27me3 DNase1, H3K27me3 H3K27me3 H3K27me3 H3K27me3 H3K27me3 H3K36me3, H3K27me3 FOSL2, Jund, USF1, H3K4me1, H3K4me2, HNF4A, FOXA1 H3K27me3 DNase1, H3K27me3 |

| | | nervous (NH-A) | DNase1, H3K27me3 |
|---|----------------|-----------------------------|------------------------------------|
| ENSR00001702960 <i>e!</i> (enhancer) | 1 | liver (HepG2) | H3K36me3 |
| | | HSMMtube | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | blood (DND-41) | H3K27me3 |
| | | nervous (NH-A) | H3K27me3 |
| ENSR00001702961 <i>e!</i> (enhancer) | 1 | embryonic stem cell (H1ESC) | H3K27me3, Rad21, CTCF |
| | | HSMMtube | H3K27me3, CTCF |
| | | blood (K562) | H3K27me3, CTCF |
| | | skin (NHDF-AD) | CTCF, H3K27me3 |
| | | muscle (HSMM) | CTCF |
| | | liver (HepG2) | Rad21, CTCF, H3K36me3 |
| | | blood (GM12878) | CTCF |
| | | lung (IMR90) | H3K27me3 |
| | | nervous (NH-A) | H3K27me3 |
| | | skin (NHEK) | CTCF, H3K27me3 |
| | | NHLF | CTCF, H3K27me3 |
| | | Osteobl | H3K27me3, CTCF |
| | | blood (DND-41) | CTCF, H3K27me3 |
| | | breast (HMEC) | CTCF |
| | | cervix (HeLa-S3) | CTCF |
| | | monocytes (Monocytes-CD14+) | CTCF, H3K27me3 |
| | | endothelium (HUVEC) | H3K36me3, CTCF, H3K27me3 |
| A549 | CTCF | | |
| ENSR00000487633 <i>e!</i> (open chromatin region) | 1 | embryonic stem cell (H1ESC) | DNase1 |
| | | endothelium (HUVEC) | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| ENSR00001494491 <i>e!</i> (CTCF binding site) | 1 | embryonic stem cell (H1ESC) | H3K4me2, Rad21, Nrsf, CTCF, DNase1 |
| | | HSMMtube | CTCF, H3K27me3 |
| | | blood (K562) | H3K27me3, Rad21, CTCF, DNase1 |
| | | skin (NHDF-AD) | CTCF |
| | | muscle (HSMM) | CTCF |
| | | liver (HepG2) | Rad21, CTCF, H3K27me3, DNase1 |
| | | blood (GM12878) | DNase1, Rad21, CTCF |
| | | lung (IMR90) | H3K27me3, CTCF |
| | | nervous (NH-A) | H3K27me3, CTCF |
| | | skin (NHEK) | CTCF, H3K27me3, DNase1 |
| | | NHLF | CTCF, H3K27me3 |
| | | Osteobl | CTCF, H3K27me3 |
| | | blood (DND-41) | CTCF, H3K27me3 |
| | | breast (HMEC) | CTCF |
| | | cervix (HeLa-S3) | Nrsf, CTCF, DNase1 |
| | | monocytes (Monocytes-CD14+) | CTCF, H3K27me3 |
| | | endothelium (HUVEC) | H3K36me3, DNase1, H3K27me3, CTCF |
| A549 | H3K27me3, CTCF | | |
| ENSR00001494494 <i>e!</i> (open chromatin region) | 1 | endothelium (HUVEC) | H3K27me3 |
| | | embryonic stem cell (H1ESC) | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | A549 | H3K27me3 |
| | | skin (NHEK) | DNase1, H3K27me3 |
| | | breast (HMEC) | DNase1 |
| ENSR00001032027 <i>e!</i> (promoter flanking region) | 1 | embryonic stem cell (H1ESC) | H3K27me3, DNase1 |
| | | HSMMtube | DNase1, H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | skin (NHDF-AD) | H3K27me3 |
| | | muscle (HSMM) | DNase1 |
| | | liver (HepG2) | H3K4me2, H3K27me3, DNase1 |
| | | lung (IMR90) | H3K27me3 |
| | | nervous (NH-A) | DNase1, H3K27me3 |
| | | skin (NHEK) | H3K27me3, DNase1 |
| | | NHLF | DNase1, H3K27me3 |

| | | | | |
|---|---|-----------------------------|---|--|
| | Osteobl | H3K27me3, H3K4me2 | | |
| | blood (DND-41) | H3K27me3 | | |
| | breast (HMEC) | H3K27me3 | | |
| | cervix (HeLa-S3) | H3K27me3 | | |
| | monocytes (Monocytes-CD14+) | H3K4me1, H3K4me3, H3K27me3 | | |
| | endothelium (HUVEC) | H3K27me3 | | |
| | A549 | H3K27me3 | | |
| ENSR00000487656 <i>e!</i> (promoter flanking region) | 1 | embryonic stem cell (H1ESC) | DNase1, H3K36me3 | |
| | | HSMMtube | H3K27me3, DNase1 | |
| | | skin (NHDF-AD) | DNase1 | |
| | | muscle (HSMM) | DNase1 | |
| | | liver (HepG2) | DNase1, H3K27ac, Cmyc, FOXA1, HNF4A, Jund, HNF4G, HDAC2, H3K4me1, p300 | |
| | | lung (IMR90) | DNase1 | |
| | | blood (GM12878) | DNase1, H3K27ac, H3K4me2 | |
| | | nervous (NH-A) | DNase1 | |
| | | skin (NHEK) | DNase1, H3K27ac | |
| | | NHLF | H3K27ac, DNase1 | |
| | | Osteobl | H3K4me2, H3K27ac | |
| | | breast (HMEC) | DNase1, H3K27ac | |
| | | cervix (HeLa-S3) | DNase1, H3K27me3 | |
| | | monocytes (Monocytes-CD14+) | DNase1 | |
| | | endothelium (HUVEC) | Max, DNase1, H3K27ac, Cjun | |
| | | A549 | DNase1 | |
| | ENSR00000487671 <i>e!</i> (promoter) | 2 | embryonic stem cell (H1ESC) | DNase1, TAF1, H3K4me2, SP1, Srf, H3K4me3, HDAC2, CTCF, H3K27ac, H3K36me3, H3K27me3, PolII, Rad21, Egr1, Jund, Sin3Ak20, H3K9ac |
| | | HSMMtube | DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, CTCF, H3K79me2 | |
| | | blood (K562) | Ini1, Egr1, CTCF, H3K36me3, DNase1, H3K4me3, H3K4me2, PolII, FOSL1, H3K27ac, Max, ZBTB7A, Srf, H3K4me1, H3K9ac, Gabp, Cmyc, H3K79me2, HEY1, H2AZ, Rad21, TAF1, ELF1 | |
| | | skin (NHDF-AD) | H3K36me3, H3K4me1, CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac | |
| | | muscle (HSMM) | H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1, H2AZ | |
| | | liver (HepG2) | DNase1, Gabp, PolII, Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3 | |
| | | lung (IMR90) | DNase1, H4K8ac, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K56ac, CTCF | |
| | | blood (GM12878) | ELF1, Rad21, H3K79me2, H3K4me3, Pax5, H3K27ac, H3K4me2, CTCF, H3K9ac, H3K36me3, Gabp, Yy1, EBF1, PolII, H2AZ, Tcf12, DNase1, Egr1 | |
| | | nervous (NH-A) | DNase1, H3K9ac, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3 | |
| | | skin (NHEK) | H3K36me3, DNase1, CTCF, H3K4me3, H3K4me2, H3K4me1, H3K27ac, H3K9ac | |
| | | NHLF | DNase1, H3K4me3, H3K4me1, CTCF, H3K9ac, H3K27ac, H3K36me3 | |
| | | Osteobl | H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac | |
| | | blood (DND-41) | H3K9ac, H3K4me3, H3K36me3, H3K27ac, CTCF, H3K4me2, H3K4me1 | |
| | | breast (HMEC) | CTCF, DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3 | |
| | | cervix (HeLa-S3) | DNase1, Jund, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, CTCF, H3K36me3, H3K9ac | |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3 | |
| | | endothelium (HUVEC) | H3K36me3, H3K27ac, PolII, CTCF, DNase1, Cmyc, H3K9ac, H3K4me3, H3K4me2, Cjun, Max | |
| | | A549 | H3K36me3, H3K27ac, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3 | |
| ENSR00000487679 <i>e!</i> (promoter) | | 1 | embryonic stem cell (H1ESC) | DNase1, H4K5ac, H3K27me3, PolII, Rad21, Egr1, Jund, TAF7, Sin3Ak20, CTCF, SIX5, H3K27ac, ATF3, Yy1, TAF1, H3K4me2, SP1, USF1, H3K36me3, H3K9ac, H3K4me3 |
| | | | HSMMtube | H3K36me3, H2AZ, H3K4me3, CTCF, H3K27ac, H3K4me2, H3K9ac, DNase1 |
| | | | blood (K562) | H3K27ac, H3K36me3, DNase1, H3K4me3, CTCF, H3K4me2, Max, SP2, PU1, H3K9ac, USF1, H3K79me2, HEY1, H2AZ, Rad21, TAF1, Cfos, CTCFL, Cmyc, PolII, H4K20me1, H3K27me3, Yy1, Ini1, Brg1, Egr1 |
| | | | skin (NHDF-AD) | H3K27ac, H3K4me2, H3K36me3, H3K4me1, CTCF, H4K20me1, H3K4me3, DNase1, H3K9ac |
| | | | muscle (HSMM) | H2AZ, H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1 |
| | | | liver (HepG2) | DNase1, Rad21, TAF1, USF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3, H3K36me3, H3K79me2, PolII, ATF3, Yy1, H4K20me1 |
| | | | lung (IMR90) | DNase1, H4K8ac, H4K20me1, H3K79me2, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K56ac, CTCF |
| | | | blood (GM12878) | Pbx3, TAF1, ATF3, SIX5, PolII, BCLAF1, H2AZ, Tcf12, DNase1, Egr1, Yy1, ELF1, Rad21, H3K79me2, H3K4me3, USF1, H3K27ac, Cfos, H3K4me2, CTCF, H3K9ac, H3K36me3 |
| | | | nervous (NH-A) | DNase1, H3K9ac, H4K20me1, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3 |
| | | skin (NHEK) | H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, H3K36me3, DNase1 | |
| | | NHLF | H3K36me3, H3K27ac, H3K9ac, DNase1, H3K4me3, H3K4me1, CTCF | |
| | | Osteobl | H3K27ac, H2AZ, H3K4me3, H3K4me2, CTCF, H3K36me3 | |
| | | blood (DND-41) | H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K36me3, CTCF, H3K4me2, H3K4me1 | |
| | | breast (HMEC) | H3K36me3, CTCF, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K4me3 | |
| | | cervix (HeLa-S3) | DNase1, H3K9ac, Nrsf, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, PolII, Cmyc, CTCF, Ini1, H3K36me3 | |

| | | | |
|----------------------------|---|-----------------------------|---|
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3 |
| | | endothelium (HUVEC) | H3K36me3, Cmyc, DNase1, CTCF, Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII |
| | | A549 | H3K36me3, H3K27ac, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3 |
| ENSR00000487687 <i>e!</i> | 1 | embryonic stem cell (H1ESC) | DNase1, H3K36me3 |
| (promoter flanking region) | | HSMMtube | H3K36me3 |
| | | blood (K562) | H3K27me3, PU1, H3K36me3 |
| | | skin (NHDF-AD) | H3K36me3 |
| | | muscle (HSMM) | H3K36me3 |
| | | liver (HepG2) | DNase1, H3K4me1, H3K36me3 |
| | | lung (IMR90) | H3K36me3 |
| | | blood (GM12878) | Yy1, PU1, PolII, H3K4me1, DNase1, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3 |
| | | nervous (NH-A) | H3K36me3 |
| | | skin (NHEK) | H3K36me3 |
| | | NHLF | H3K36me3 |
| | | Osteobl | H3K36me3 |
| | | blood (DND-41) | H3K4me1, H3K36me3 |
| | | cervix (HeLa-S3) | H3K27ac, PolII |
| | | monocytes (Monocytes-CD14+) | H3K36me3, H3K4me1 |
| | | endothelium (HUVEC) | PolII, H3K36me3, DNase1 |
| | | A549 | H3K36me3 |
| ENSR00000487711 <i>e!</i> | 1 | monocytes (Monocytes-CD14+) | DNase1, H3K4me1, H3K27ac |
| (promoter flanking region) | | liver (HepG2) | H3K4me1, H3K27ac |
| | | Osteobl | H2AZ, H3K27ac, H3K4me2 |
| | | blood (GM12878) | H2AZ, DNase1, H3K27ac |
| | | lung (IMR90) | DNase1, H3K27ac |
| | | blood (DND-41) | H3K4me1, H3K27ac |
| | | blood (K562) | DNase1, H2AZ |
| ENSR00000487714 <i>e!</i> | 1 | endothelium (HUVEC) | DNase1 |
| (promoter flanking region) | | liver (HepG2) | H3K4me1, FOXA1 |
| | | blood (K562) | H3K4me1 |
| ENSR00000662312 <i>e!</i> | 2 | NHLF | DNase1 |
| (promoter flanking region) | | embryonic stem cell (H1ESC) | DNase1, H3K36me3, H3K27me3, PolII, TAF7 |
| | | Osteobl | H3K27ac |
| | | blood (K562) | USF1, HEY1, H2AZ, Cjun, Cfos, PolII, MEF2A, Max, Ini1, Brg1, DNase1, FOSL1, H3K27ac |
| | | skin (NHDF-AD) | DNase1 |
| | | breast (HMEC) | H3K27ac, DNase1 |
| | | muscle (HSMM) | H2AZ |
| | | cervix (HeLa-S3) | DNase1, PolII, TAF1, H3K9ac, Jun, H3K27ac, H3K4me1 |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me1, H3K27ac |
| | | endothelium (HUVEC) | H3K36me3, Cjun, Max, H3K4me1, H3K27ac, PolII, DNase1, Cmyc |
| | | liver (HepG2) | DNase1, FOSL2, Jun, PolII, H3K4me1 |
| | | lung (IMR90) | DNase1, H2BK12ac, H3K18ac, H2BK120ac, H2BK20ac, H4K91ac |
| | | blood (GM12878) | EBF1, PolII, BATF, DNase1, IRF4, BCL3, BCL11A, Pax5, NFKB |
| | | nervous (NH-A) | DNase1 |
| | | skin (NHEK) | H3K4me1, H3K27ac, DNase1 |

Variation in RISC binding site

| gene | variant(s) | affected transcript(s) | targeting miRNA(s) |
|-----------------|------------|--|--|
| ZNRD1 <i>e!</i> | 1 | ENST00000332435 <i>e!</i> ENST00000359374 <i>e!</i> ENST00000376782 <i>e!</i> ENST00000376785 <i>e!</i> ENST00000463141 <i>e!</i> ENST00000471008 <i>e!</i> | hsa-miR-199a-5p  hsa-miR-199b-5p  |

Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|-----------------|-----------------------|---------------|---------------------------|----------------|-----------------|------------|
| ABCF1 <i>e!</i> | upstream gene variant | 2298 | ENST00000376545 <i>e!</i> | NM_001090.2 | ENSP00000365728 | 1 |
| ABCF1 <i>e!</i> | upstream gene variant | 2338 | ENST00000441867 <i>e!</i> | ? | ENSP00000405512 | 1 |
| ABCF1 <i>e!</i> | upstream gene variant | 1503 | ENST00000542772 <i>e!</i> | ? | ? | 1 |
| ABCF1 <i>e!</i> | upstream gene variant | 2281 | ENST00000326195 <i>e!</i> | NM_001025091.1 | ENSP00000313603 | 1 |

| | | | | |
|----------------------|--|------|---|-------------------|
| ABCF1 <i>e!</i> | upstream gene variant | 2347 | ENST00000468958 ? | ENSP00000440893 1 |
| ABCF1 <i>e!</i> | downstream gene variant | 1451 | ENST00000475993 ? | ENSP00000445100 1 |
| AL662797.1 <i>e!</i> | upstream gene variant, downstream gene variant | 112 | ENST00000620079 ? | ? 3 |
| AL662800.2 <i>e!</i> | downstream gene variant | 2370 | ENST00000583820 ? | ? 1 |
| ATAT1 <i>e!</i> | downstream gene variant | 4727 | ENST00000462304 ? | ? 1 |
| ATAT1 <i>e!</i> | downstream gene variant | 4306 | ENST00000330083 NM_001031722.2 | ENSP00000327832 1 |
| ATAT1 <i>e!</i> | downstream gene variant | 4306 | ENST00000376485 ? | ENSP00000365668 1 |
| ATAT1 <i>e!</i> | downstream gene variant | 4306 | ENST00000471782 ? | ? 1 |
| ATAT1 <i>e!</i> | downstream gene variant | 4306 | ENST00000479562 ? | ? 1 |
| ATAT1 <i>e!</i> | downstream gene variant | 2875 | ENST00000493388 ? | ? 1 |
| ATAT1 <i>e!</i> | downstream gene variant | 4306 | ENST00000468713 ? | ? 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 137 | ENST00000467801 ? | ENSP00000419610 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 3482 | ENST00000488383 ? | ENSP00000419968 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 3848 | ENST00000446773 ? | ENSP00000415810 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 3466 | ENST00000484551 ? | ENSP00000418599 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 3553 | ENST00000463794 ? | ENSP00000418223 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 255 | ENST00000468785 ? | ENSP00000419460 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 3591 | ENST00000376471 NM_145029.3 | ENSP00000365654 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 3466 | ENST00000293604 NM_001161376.1 | ENSP00000293604 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 3500 | ENST00000376473 NM_001109938.2 | ENSP00000365656 1 |
| C6orf136 <i>e!</i> | upstream gene variant | 1482 | ENST00000460172 ? | ? 1 |
| C6orf136 <i>e!</i> | upstream gene variant, downstream gene variant | 1216 | ENST00000493705 ? | ? 2 |
| DHX16 <i>e!</i> | upstream gene variant | 3372 | ENST00000415603 ? | ENSP00000399101 1 |
| DHX16 <i>e!</i> | downstream gene variant | 1990 | ENST00000376437 ? | ENSP00000365620 1 |
| DHX16 <i>e!</i> | downstream gene variant, upstream gene variant | 1990 | ENST00000376442 NM_001164239.1, NM_003587.4 | ENSP00000365625 2 |
| FLOT1 <i>e!</i> | upstream gene variant | 384 | ENST00000476729 ? | ? 2 |
| FLOT1 <i>e!</i> | downstream gene variant | 1925 | ENST00000454845 ? | ENSP00000391341 1 |
| FLOT1 <i>e!</i> | downstream gene variant | 176 | ENST00000484693 ? | ? 2 |
| FLOT1 <i>e!</i> | downstream gene variant | 1577 | ENST00000418160 ? | ENSP00000404300 1 |
| FLOT1 <i>e!</i> | downstream gene variant | 1670 | ENST00000484168 ? | ? 1 |
| FLOT1 <i>e!</i> | upstream gene variant | 325 | ENST00000487376 ? | ? 2 |
| FLOT1 <i>e!</i> | downstream gene variant | 1592 | ENST00000445853 ? | ENSP00000398834 1 |
| FLOT1 <i>e!</i> | downstream gene variant | 1671 | ENST00000416018 ? | ENSP00000412058 1 |
| FLOT1 <i>e!</i> | downstream gene variant | 1574 | ENST00000470643 ? | ? 1 |
| GNL1 <i>e!</i> | downstream gene variant | 4911 | ENST00000487166 ? | ? 1 |
| GNL1 <i>e!</i> | downstream gene variant | 198 | ENST00000376621 NM_005275.3 | ENSP00000365806 2 |

| | | | | | | |
|-----------|----|--|------|-----------------------------|-----------------|----|
| HCG17 | e! | upstream gene variant | 1993 | ENST00000455957 ? | ? | 2 |
| HCG17 | e! | upstream gene variant | 2342 | ENST00000453558 ? | ? | 1 |
| HCG18 | e! | downstream gene variant | 3783 | ENST00000602591 ? | ? | 1 |
| HCG18 | e! | upstream gene variant | 1610 | ENST00000412685 ? | ? | 2 |
| HCG18 | e! | upstream gene variant | 2100 | ENST00000454129 ? | ? | 1 |
| HCG18 | e! | upstream gene variant | 2089 | ENST00000438412 ? | ? | 1 |
| HCG18 | e! | upstream gene variant | 4008 | ENST00000602516 ? | ? | 2 |
| HCG18 | e! | upstream gene variant | 2113 | ENST00000602290 ? | ? | 1 |
| HCG18 | e! | upstream gene variant | 1579 | ENST00000413358 ? | ? | 2 |
| HCG18 | e! | upstream gene variant | 2129 | ENST00000449544 ? | ? | 1 |
| HCG18 | e! | upstream gene variant | 2072 | ENST00000602550 ? | ? | 1 |
| HCG18 | e! | upstream gene variant | 1326 | ENST00000426882 ? | ? | 2 |
| HCG18 | e! | upstream gene variant | 2339 | ENST00000602498 ? | ? | 1 |
| HCG18 | e! | upstream gene variant | 2142 | ENST00000444126 ? | ? | 1 |
| HCG18 | e! | upstream gene variant | 2090 | ENST00000454269 ? | ? | 1 |
| HCG19P | e! | downstream gene variant, upstream gene variant | 151 | ENST00000448756 ? | ? | 14 |
| HCG20 | e! | downstream gene variant | 1545 | ENST00000439406 ? | ? | 1 |
| HCG20 | e! | upstream gene variant, downstream gene variant | 1545 | ENST00000422944 ? | ? | 2 |
| HCG4P3 | e! | downstream gene variant | 2938 | ENST00000458060 ? | ? | 1 |
| HLA-J | e! | upstream gene variant | 3413 | ENST00000495278 ? | ? | 1 |
| HLA-J | e! | upstream gene variant | 3404 | ENST00000462773 ? | ? | 1 |
| HLA-J | e! | upstream gene variant | 3423 | ENST00000494367 ? | ? | 1 |
| HLA-J | e! | upstream gene variant | 3400 | ENST00000469281 ? | ? | 1 |
| HLA-L | e! | upstream gene variant | 2391 | ENST00000482052 ? | ? | 1 |
| HLA-L | e! | upstream gene variant, downstream gene variant | 1288 | ENST00000420110 ? | ? | 2 |
| HLA-L | e! | upstream gene variant | 2404 | ENST00000463348 ? | ? | 1 |
| HLA-N | e! | upstream gene variant, downstream gene variant | 39 | ENST00000437516 ? | ? | 3 |
| IER3 | e! | downstream gene variant, upstream gene variant | 1619 | ENST00000376377 ? | ENSP00000365557 | 4 |
| IER3 | e! | downstream gene variant, upstream gene variant | 1619 | ENST00000259874 NM_003897.3 | ENSP00000259874 | 4 |
| LINC00243 | e! | downstream gene variant | 4859 | ENST00000419357 ? | ? | 1 |
| LINC00243 | e! | downstream gene variant | 75 | ENST00000399196 ? | ? | 1 |
| MDC1 | e! | upstream gene variant | 3623 | ENST00000416571 ? | ENSP00000400979 | 1 |
| MDC1 | e! | upstream gene variant | 3351 | ENST00000425072 ? | ENSP00000396989 | 1 |
| MDC1 | e! | upstream gene variant | 2909 | ENST00000376406 NM_014641.2 | ENSP00000365588 | 1 |
| MDC1 | e! | upstream gene variant | 3723 | ENST00000613547 ? | ENSP00000483601 | 1 |
| MDC1 | e! | upstream gene variant | 3677 | ENST00000422266 ? | ENSP00000411310 | 1 |
| MICC | e! | upstream gene variant | 896 | ENST00000445710 ? | ? | 2 |

| | | | | | |
|---------------------|--|------|--------------------------------|-----------------|---|
| MRPS18B <i>e!</i> | downstream gene variant | 2249 | ENST00000472229 ? | ? | 1 |
| MRPS18B <i>e!</i> | downstream gene variant | 2249 | ENST00000472267 ? | ? | 1 |
| MRPS18B <i>e!</i> | downstream gene variant | 2249 | ENST00000259873 NM_014046.3 | ENSP00000259873 | 1 |
| NRM <i>e!</i> | downstream gene variant | 3043 | ENST00000444096 ? | ENSP00000397892 | 1 |
| NRM <i>e!</i> | downstream gene variant | 3043 | ENST00000259953 NM_007243.2 | ENSP00000259953 | 1 |
| NRM <i>e!</i> | downstream gene variant | 3043 | ENST00000376420 NM_001270709.1 | ENSP00000365602 | 1 |
| NRM <i>e!</i> | downstream gene variant | 3043 | ENST00000470733 ? | ? | 1 |
| NRM <i>e!</i> | downstream gene variant | 3043 | ENST00000462857 ? | ? | 1 |
| NRM <i>e!</i> | downstream gene variant | 3046 | ENST00000376421 NM_001270707.1 | ENSP00000365603 | 1 |
| NRM <i>e!</i> | downstream gene variant | 3043 | ENST00000495946 ? | ? | 1 |
| NRM <i>e!</i> | downstream gene variant | 3043 | ENST00000482141 ? | ? | 1 |
| NRM <i>e!</i> | downstream gene variant | 3043 | ENST00000474864 ? | ? | 1 |
| PAIP1P1 <i>e!</i> | downstream gene variant | 1212 | ENST00000446875 ? | ? | 1 |
| PPP1R10 <i>e!</i> | downstream gene variant | 1770 | ENST00000376511 NM_002714.3 | ENSP00000365694 | 1 |
| PPP1R10 <i>e!</i> | downstream gene variant | 4943 | ENST00000496955 ? | ? | 1 |
| PPP1R11 <i>e!</i> | upstream gene variant, downstream gene variant | 537 | ENST00000376772 NM_021959.2 | ENSP00000365963 | 2 |
| PPP1R11 <i>e!</i> | upstream gene variant, downstream gene variant | 537 | ENST00000376758 ? | ENSP00000365949 | 2 |
| PPP1R11 <i>e!</i> | upstream gene variant, downstream gene variant | 537 | ENST00000376763 ? | ENSP00000365954 | 2 |
| PPP1R11 <i>e!</i> | upstream gene variant, downstream gene variant | 537 | ENST00000376765 ? | ENSP00000365956 | 2 |
| PPP1R11 <i>e!</i> | upstream gene variant, downstream gene variant | 537 | ENST00000376769 ? | ENSP00000365960 | 2 |
| PPP1R11 <i>e!</i> | upstream gene variant, downstream gene variant | 537 | ENST00000376773 ? | ENSP00000365964 | 2 |
| PPP1R18 <i>e!</i> | downstream gene variant, upstream gene variant | 29 | ENST00000399199 NM_001134870.1 | ENSP00000382150 | 2 |
| PPP1R18 <i>e!</i> | downstream gene variant, upstream gene variant | 29 | ENST00000488324 ? | ? | 2 |
| PPP1R18 <i>e!</i> | downstream gene variant, upstream gene variant | 31 | ENST00000615527 ? | ENSP00000480270 | 2 |
| PPP1R18 <i>e!</i> | downstream gene variant, upstream gene variant | 29 | ENST00000274853 NM_133471.3 | ENSP00000274853 | 2 |
| PPP1R18 <i>e!</i> | downstream gene variant, upstream gene variant | 29 | ENST00000467662 ? | ? | 2 |
| PPP1R18 <i>e!</i> | downstream gene variant, upstream gene variant | 29 | ENST00000615892 ? | ENSP00000482578 | 3 |
| PTMAP1 <i>e!</i> | upstream gene variant, downstream gene variant | 1844 | ENST00000455552 ? | ? | 2 |
| RN7SL353P <i>e!</i> | upstream gene variant, downstream gene variant | 1557 | ENST00000579902 ? | ? | 5 |
| RPP21 <i>e!</i> | downstream gene variant | 3693 | ENST00000489124 ? | ? | 2 |
| RPP21 <i>e!</i> | downstream gene variant | 3693 | ENST00000436442 NM_001199121.1 | ENSP00000397778 | 2 |
| RPP21 <i>e!</i> | downstream gene variant | 3693 | ENST00000428040 ? | ENSP00000394320 | 2 |
| RPP21 <i>e!</i> | downstream gene variant | 3693 | ENST00000498414 ? | ? | 2 |
| RPP21 <i>e!</i> | downstream gene variant | 3693 | ENST00000433076 NM_001199120.1 | ENSP00000409799 | 2 |
| RPP21 <i>e!</i> | downstream gene variant | 3666 | ENST00000491477 ? | ? | 2 |
| RPP21 <i>e!</i> | downstream gene variant | 4057 | ENST00000466327 ? | ? | 2 |
| RPP21 <i>e!</i> | downstream gene variant | 3693 | ENST00000442966 NM_024839.2 | ENSP00000403833 | 2 |

| | | | | | |
|-----------------------------|--|------|--------------------------------|-----------------|----|
| RPP21 <i>e!</i> | downstream gene variant | 3693 | ENST00000473266 ? | ? | 2 |
| SUCLA2P1 <i>e!</i> | downstream gene variant | 3275 | ENST00000425839 ? | ? | 1 |
| TRIM10 <i>e!</i> | upstream gene variant | 238 | ENST00000376704 NM_052828.2 | ENSP00000365894 | 1 |
| TRIM10 <i>e!</i> | upstream gene variant | 238 | ENST00000449742 NM_006778.3 | ENSP00000397073 | 1 |
| TRIM15 <i>e!</i> | upstream gene variant | 2034 | ENST00000619857 ? | ENSP00000484001 | 2 |
| TRIM15 <i>e!</i> | upstream gene variant | 703 | ENST00000477944 ? | ? | 1 |
| TRIM15 <i>e!</i> | upstream gene variant | 2044 | ENST00000376694 NM_033229.2 | ENSP00000365884 | 2 |
| TRIM15 <i>e!</i> | upstream gene variant | 2513 | ENST00000376688 ? | ENSP00000365878 | 2 |
| TRIM26 <i>e!</i> | upstream gene variant | 996 | ENST00000437089 ? | ENSP00000395491 | 1 |
| TRIM26 <i>e!</i> | downstream gene variant | 4732 | ENST00000418026 ? | ENSP00000387530 | 1 |
| TRIM26 <i>e!</i> | downstream gene variant | 2777 | ENST00000416596 ? | ENSP00000413673 | 1 |
| TRIM26 <i>e!</i> | downstream gene variant | 4979 | ENST00000434785 ? | ENSP00000400920 | 1 |
| TRIM26 <i>e!</i> | upstream gene variant | 3365 | ENST00000480999 ? | ? | 1 |
| TRIM26BP <i>e!</i> | upstream gene variant | 64 | ENST00000427723 ? | ? | 2 |
| TRIM31 <i>e!</i> | upstream gene variant | 3745 | ENST00000485864 ? | ? | 1 |
| TRIM31 <i>e!</i> | upstream gene variant | 2825 | ENST00000376734 NM_007028.3 | ENSP00000365924 | 1 |
| TRIM31 <i>e!</i> | downstream gene variant, upstream gene variant | 3382 | ENST00000493404 ? | ? | 2 |
| TRIM31 <i>e!</i> | downstream gene variant, upstream gene variant | 910 | ENST00000480808 ? | ? | 2 |
| TRIM31 <i>e!</i> | upstream gene variant | 2396 | ENST00000484583 ? | ? | 1 |
| TRIM31 <i>e!</i> | upstream gene variant | 2179 | ENST00000471695 ? | ? | 1 |
| TRIM31-AS1 <i>e!</i> | downstream gene variant | 1207 | ENST00000440874 ? | ? | 1 |
| TRIM39 <i>e!</i> | upstream gene variant | 1053 | ENST00000420746 ? | ENSP00000388224 | 1 |
| TRIM39 <i>e!</i> | downstream gene variant | 1717 | ENST00000458516 ? | ENSP00000405928 | 1 |
| TRIM39 <i>e!</i> | downstream gene variant | 1973 | ENST00000428404 ? | ENSP00000405498 | 1 |
| TRIM39 <i>e!</i> | downstream gene variant | 1929 | ENST00000440271 ? | ENSP00000394768 | 1 |
| TRIM39 <i>e!</i> | upstream gene variant | 413 | ENST00000396547 ? | ENSP00000379796 | 1 |
| TRIM39 <i>e!</i> | downstream gene variant | 2006 | ENST00000428555 ? | ENSP00000397952 | 1 |
| TRIM39-RPP21 <i>e!</i> | upstream gene variant, downstream gene variant | 431 | ENST00000623385 ? | ENSP00000485378 | 3 |
| TRIM39-RPP21 <i>e!</i> | upstream gene variant, downstream gene variant | 835 | ENST00000513556 NM_001199119.1 | ENSP00000424048 | 3 |
| TUBB <i>e!</i> | upstream gene variant | 869 | ENST00000396389 ? | ENSP00000379672 | 1 |
| TUBB <i>e!</i> | upstream gene variant | 776 | ENST00000330914 NM_001293215.1 | ENSP00000365578 | 1 |
| TUBB <i>e!</i> | upstream gene variant | 901 | ENST00000396384 ? | ENSP00000379668 | 1 |
| UBQLN1P1 <i>e!</i> | downstream gene variant, upstream gene variant | 152 | ENST00000441056 ? | ? | 16 |
| XXbac-BPG252P9.10 <i>e!</i> | upstream gene variant | 1349 | ENST00000607333 ? | ? | 3 |
| XXbac-BPG252P9.9 <i>e!</i> | downstream gene variant | 2307 | ENST00000607476 ? | ? | 1 |
| XXbac-BPG283O16.9 <i>e!</i> | downstream gene variant | 1415 | ENST00000624252 ? | ? | 1 |
| Y_RNA <i>e!</i> | downstream gene variant, upstream gene variant | 2170 | ENST00000365118 ? | ? | 3 |

| | | | | | |
|-----------------------|--|------|-------------------|---|---|
| ZNRD1-AS1 <i>e!</i> | upstream gene variant | 3566 | ENST00000431012 ? | ? | 1 |
| ZNRD1-AS1 <i>e!</i> | upstream gene variant | 3601 | ENST00000376797 ? | ? | 1 |
| ZNRD1-AS1 <i>e!</i> | upstream gene variant | 3817 | ENST00000425604 ? | ? | 1 |
| ZNRD1-AS1 <i>e!</i> | upstream gene variant | 3556 | ENST00000421692 ? | ? | 1 |
| ZNRD1-AS1 <i>e!</i> | upstream gene variant | 3766 | ENST00000452229 ? | ? | 1 |
| ZNRD1-AS1 <i>e!</i> | downstream gene variant, upstream gene variant | 1223 | ENST00000437417 ? | ? | 2 |
| ZNRD1-AS1 <i>e!</i> | upstream gene variant | 3602 | ENST00000448093 ? | ? | 1 |
| ZNRD1-AS1 <i>e!</i> | upstream gene variant | 3561 | ENST00000420251 ? | ? | 1 |
| ZNRD1-AS1_3 <i>e!</i> | downstream gene variant | 3562 | ENST00000611686 ? | ? | 1 |

Putative effect on transcript

Synonymous coding variant

| gene | affected transcript | RefSeq id | protein | AA's | exchanged codons | variant(s) |
|----------------|---------------------------|-------------|---------------------------|------|------------------|------------|
| GNL1 <i>e!</i> | ENST00000376621 <i>e!</i> | NM_005275.3 | ENSP00000365806 <i>e!</i> | A | gcG/gcA | 1 |

Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|--------------------|---------------------------|----------------|---------------------------|------------|
| ABCF1 <i>e!</i> | ENST00000326195 <i>e!</i> | NM_001025091.1 | ENSP00000313603 <i>e!</i> | 1 |
| ABCF1 <i>e!</i> | ENST00000468958 <i>e!</i> | ? | ENSP00000440893 <i>e!</i> | 1 |
| ABCF1 <i>e!</i> | ENST00000441867 <i>e!</i> | ? | ENSP00000405512 <i>e!</i> | 1 |
| ABCF1 <i>e!</i> | ENST00000376545 <i>e!</i> | NM_001090.2 | ENSP00000365728 <i>e!</i> | 1 |
| ATAT1 <i>e!</i> | ENST00000376485 <i>e!</i> | ? | ENSP00000365668 <i>e!</i> | 3 |
| ATAT1 <i>e!</i> | ENST00000493388 <i>e!</i> | ? | ? | 2 |
| ATAT1 <i>e!</i> | ENST00000318999 <i>e!</i> | ? | ENSP00000324222 <i>e!</i> | 3 |
| ATAT1 <i>e!</i> | ENST00000468713 <i>e!</i> | ? | ? | 3 |
| ATAT1 <i>e!</i> | ENST00000376483 <i>e!</i> | ? | ENSP00000365666 <i>e!</i> | 3 |
| ATAT1 <i>e!</i> | ENST00000330083 <i>e!</i> | NM_001031722.2 | ENSP00000327832 <i>e!</i> | 3 |
| ATAT1 <i>e!</i> | ENST00000319027 <i>e!</i> | NM_001254952.1 | ENSP00000324459 <i>e!</i> | 3 |
| ATAT1 <i>e!</i> | ENST00000471782 <i>e!</i> | ? | ? | 3 |
| ATAT1 <i>e!</i> | ENST00000329992 <i>e!</i> | NM_024909.2 | ENSP00000332374 <i>e!</i> | 3 |
| ATAT1 <i>e!</i> | ENST00000479562 <i>e!</i> | ? | ? | 1 |
| ATAT1 <i>e!</i> | ENST00000462304 <i>e!</i> | ? | ? | 3 |
| C6orf136 <i>e!</i> | ENST00000376473 <i>e!</i> | NM_001109938.2 | ENSP00000365656 <i>e!</i> | 1 |
| C6orf136 <i>e!</i> | ENST00000465699 <i>e!</i> | ? | ENSP00000418154 <i>e!</i> | 1 |
| C6orf136 <i>e!</i> | ENST00000293604 <i>e!</i> | NM_001161376.1 | ENSP00000293604 <i>e!</i> | 1 |
| C6orf136 <i>e!</i> | ENST00000488383 <i>e!</i> | ? | ENSP00000419968 <i>e!</i> | 1 |
| C6orf136 <i>e!</i> | ENST00000376471 <i>e!</i> | NM_145029.3 | ENSP00000365654 <i>e!</i> | 1 |
| C6orf136 <i>e!</i> | ENST00000446773 <i>e!</i> | ? | ENSP00000415810 <i>e!</i> | 1 |
| C6orf136 <i>e!</i> | ENST00000487873 <i>e!</i> | ? | ENSP00000418616 <i>e!</i> | 1 |
| C6orf136 <i>e!</i> | ENST00000463794 <i>e!</i> | ? | ENSP00000418223 <i>e!</i> | 1 |

| | | | | |
|---------------------|---------------------------|----------------|---------------------------|----|
| C6orf136 <i>e!</i> | ENST00000484551 <i>e!</i> | ? | ENSP00000418599 <i>e!</i> | 1 |
| FLOT1 <i>e!</i> | ENST00000418160 <i>e!</i> | ? | ENSP00000404300 <i>e!</i> | 2 |
| FLOT1 <i>e!</i> | ENST00000416018 <i>e!</i> | ? | ENSP00000412058 <i>e!</i> | 2 |
| FLOT1 <i>e!</i> | ENST00000470643 <i>e!</i> | ? | ? | 2 |
| FLOT1 <i>e!</i> | ENST00000484168 <i>e!</i> | ? | ? | 2 |
| FLOT1 <i>e!</i> | ENST00000476729 <i>e!</i> | ? | ? | 2 |
| FLOT1 <i>e!</i> | ENST00000413165 <i>e!</i> | ? | ENSP00000395333 <i>e!</i> | 4 |
| FLOT1 <i>e!</i> | ENST00000376389 <i>e!</i> | NM_005803.2 | ENSP00000365569 <i>e!</i> | 4 |
| FLOT1 <i>e!</i> | ENST00000454845 <i>e!</i> | ? | ENSP00000391341 <i>e!</i> | 2 |
| FLOT1 <i>e!</i> | ENST00000438162 <i>e!</i> | ? | ENSP00000400615 <i>e!</i> | 4 |
| FLOT1 <i>e!</i> | ENST00000445853 <i>e!</i> | ? | ENSP00000398834 <i>e!</i> | 2 |
| FLOT1 <i>e!</i> | ENST00000487376 <i>e!</i> | ? | ? | 2 |
| HCG17 <i>e!</i> | ENST00000453558 <i>e!</i> | ? | ? | 11 |
| HCG18 <i>e!</i> | ENST00000454129 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000444126 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000602550 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000438412 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000412685 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000454269 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000426882 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000449544 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000413358 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000602498 <i>e!</i> | ? | ? | 4 |
| HCG18 <i>e!</i> | ENST00000602290 <i>e!</i> | ? | ? | 4 |
| HCG20 <i>e!</i> | ENST00000439406 <i>e!</i> | ? | ? | 2 |
| HLA-L <i>e!</i> | ENST00000463348 <i>e!</i> | ? | ? | 1 |
| HLA-L <i>e!</i> | ENST00000482052 <i>e!</i> | ? | ? | 3 |
| LINC00243 <i>e!</i> | ENST00000419357 <i>e!</i> | ? | ? | 4 |
| LINC00243 <i>e!</i> | ENST00000399196 <i>e!</i> | ? | ? | 2 |
| MICC <i>e!</i> | ENST00000445710 <i>e!</i> | ? | ? | 1 |
| NRM <i>e!</i> | ENST00000474864 <i>e!</i> | ? | ? | 1 |
| NRM <i>e!</i> | ENST00000470733 <i>e!</i> | ? | ? | 1 |
| NRM <i>e!</i> | ENST00000444096 <i>e!</i> | ? | ENSP00000397892 <i>e!</i> | 1 |
| NRM <i>e!</i> | ENST00000376421 <i>e!</i> | NM_001270707.1 | ENSP00000365603 <i>e!</i> | 1 |
| NRM <i>e!</i> | ENST00000482141 <i>e!</i> | ? | ? | 1 |
| NRM <i>e!</i> | ENST00000495946 <i>e!</i> | ? | ? | 1 |
| NRM <i>e!</i> | ENST00000259953 <i>e!</i> | NM_007243.2 | ENSP00000259953 <i>e!</i> | 1 |
| NRM <i>e!</i> | ENST00000462857 <i>e!</i> | ? | ? | 1 |
| NRM <i>e!</i> | ENST00000376420 <i>e!</i> | NM_001270709.1 | ENSP00000365602 <i>e!</i> | 1 |
| PPP1R18 <i>e!</i> | ENST00000274853 <i>e!</i> | NM_133471.3 | ENSP00000274853 <i>e!</i> | 1 |
| PPP1R18 <i>e!</i> | ENST00000467662 <i>e!</i> | ? | ? | 2 |
| PPP1R18 <i>e!</i> | ENST00000399199 <i>e!</i> | NM_001134870.1 | ENSP00000382150 <i>e!</i> | 1 |

| | | | | |
|------------------------|---------------------------|----------------|---------------------------|---|
| PPP1R18 <i>e!</i> | ENST00000615892 <i>e!</i> | ? | ENSP00000482578 <i>e!</i> | 1 |
| PPP1R18 <i>e!</i> | ENST00000615527 <i>e!</i> | ? | ENSP00000480270 <i>e!</i> | 1 |
| PPP1R18 <i>e!</i> | ENST00000488324 <i>e!</i> | ? | ? | 2 |
| TRIM10 <i>e!</i> | ENST00000449742 <i>e!</i> | NM_006778.3 | ENSP00000397073 <i>e!</i> | 2 |
| TRIM10 <i>e!</i> | ENST00000376704 <i>e!</i> | NM_052828.2 | ENSP00000365894 <i>e!</i> | 2 |
| TRIM15 <i>e!</i> | ENST00000376694 <i>e!</i> | NM_033229.2 | ENSP00000365884 <i>e!</i> | 1 |
| TRIM15 <i>e!</i> | ENST00000619857 <i>e!</i> | ? | ENSP00000484001 <i>e!</i> | 1 |
| TRIM15 <i>e!</i> | ENST00000433744 <i>e!</i> | ? | ENSP00000398285 <i>e!</i> | 1 |
| TRIM15 <i>e!</i> | ENST00000376688 <i>e!</i> | ? | ENSP00000365878 <i>e!</i> | 1 |
| TRIM26 <i>e!</i> | ENST00000418026 <i>e!</i> | ? | ENSP00000387530 <i>e!</i> | 2 |
| TRIM26 <i>e!</i> | ENST00000434785 <i>e!</i> | ? | ENSP00000400920 <i>e!</i> | 2 |
| TRIM26 <i>e!</i> | ENST00000487829 <i>e!</i> | ? | ? | 2 |
| TRIM26 <i>e!</i> | ENST00000480999 <i>e!</i> | ? | ? | 1 |
| TRIM26 <i>e!</i> | ENST00000453195 <i>e!</i> | NM_001242783.1 | ENSP00000391879 <i>e!</i> | 4 |
| TRIM26 <i>e!</i> | ENST00000437089 <i>e!</i> | ? | ENSP00000395491 <i>e!</i> | 3 |
| TRIM26 <i>e!</i> | ENST00000454678 <i>e!</i> | NM_003449.4 | ENSP00000410446 <i>e!</i> | 4 |
| TRIM26 <i>e!</i> | ENST00000416596 <i>e!</i> | ? | ENSP00000413673 <i>e!</i> | 2 |
| TRIM31 <i>e!</i> | ENST00000468264 <i>e!</i> | ? | ? | 1 |
| TRIM31 <i>e!</i> | ENST00000485864 <i>e!</i> | ? | ? | 1 |
| TRIM31 <i>e!</i> | ENST00000376734 <i>e!</i> | NM_007028.3 | ENSP00000365924 <i>e!</i> | 1 |
| TRIM31-AS1 <i>e!</i> | ENST00000440874 <i>e!</i> | ? | ? | 1 |
| TRIM39 <i>e!</i> | ENST00000458516 <i>e!</i> | ? | ENSP00000405928 <i>e!</i> | 1 |
| TRIM39 <i>e!</i> | ENST00000396547 <i>e!</i> | ? | ENSP00000379796 <i>e!</i> | 1 |
| TRIM39 <i>e!</i> | ENST00000396548 <i>e!</i> | ? | ENSP00000379797 <i>e!</i> | 2 |
| TRIM39 <i>e!</i> | ENST00000428728 <i>e!</i> | ? | ENSP00000406019 <i>e!</i> | 2 |
| TRIM39 <i>e!</i> | ENST00000428404 <i>e!</i> | ? | ENSP00000405498 <i>e!</i> | 1 |
| TRIM39 <i>e!</i> | ENST00000428555 <i>e!</i> | ? | ENSP00000397952 <i>e!</i> | 1 |
| TRIM39 <i>e!</i> | ENST00000396551 <i>e!</i> | ? | ENSP00000379800 <i>e!</i> | 2 |
| TRIM39 <i>e!</i> | ENST00000376659 <i>e!</i> | NM_172016.2 | ENSP00000365847 <i>e!</i> | 2 |
| TRIM39 <i>e!</i> | ENST00000420746 <i>e!</i> | ? | ENSP00000388224 <i>e!</i> | 1 |
| TRIM39 <i>e!</i> | ENST00000440271 <i>e!</i> | ? | ENSP00000394768 <i>e!</i> | 1 |
| TRIM39 <i>e!</i> | ENST00000376656 <i>e!</i> | NM_021253.3 | ENSP00000365844 <i>e!</i> | 2 |
| TRIM39-RPP21 <i>e!</i> | ENST00000513556 <i>e!</i> | NM_001199119.1 | ENSP00000424048 <i>e!</i> | 1 |
| TRIM39-RPP21 <i>e!</i> | ENST00000623385 <i>e!</i> | ? | ENSP00000485378 <i>e!</i> | 1 |
| TRIM40 <i>e!</i> | ENST00000376724 <i>e!</i> | ? | ENSP00000365914 <i>e!</i> | 1 |
| TRIM40 <i>e!</i> | ENST00000307859 <i>e!</i> | NM_138700.4 | ENSP00000308310 <i>e!</i> | 1 |
| TRIM40 <i>e!</i> | ENST00000396581 <i>e!</i> | NM_001286633.1 | ENSP00000379826 <i>e!</i> | 1 |
| TUBB <i>e!</i> | ENST00000327892 <i>e!</i> | NM_178014.2 | ENSP00000339001 <i>e!</i> | 1 |
| ZNRD1-AS1 <i>e!</i> | ENST00000425604 <i>e!</i> | ? | ? | 2 |
| ZNRD1-AS1 <i>e!</i> | ENST00000420251 <i>e!</i> | ? | ? | 2 |
| ZNRD1-AS1 <i>e!</i> | ENST00000376797 <i>e!</i> | ? | ? | 2 |
| ZNRD1-AS1 <i>e!</i> | ENST00000418002 <i>e!</i> | ? | ? | 2 |

3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|------------------|---------------------------|----------------|---------------------------|------------|
| ATAT1 <i>e!</i> | ENST00000318999 <i>e!</i> | ? | ENSP00000324222 <i>e!</i> | 1 |
| ATAT1 <i>e!</i> | ENST00000376483 <i>e!</i> | ? | ENSP00000365666 <i>e!</i> | 1 |
| ATAT1 <i>e!</i> | ENST00000319027 <i>e!</i> | NM_001254952.1 | ENSP00000324459 <i>e!</i> | 1 |
| ATAT1 <i>e!</i> | ENST00000329992 <i>e!</i> | NM_024909.2 | ENSP00000332374 <i>e!</i> | 1 |
| RNF39 <i>e!</i> | ENST00000376751 <i>e!</i> | NM_170769.2 | ENSP00000365942 <i>e!</i> | 1 |
| RNF39 <i>e!</i> | ENST00000244360 <i>e!</i> | NM_025236.3 | ENSP00000244360 <i>e!</i> | 1 |
| TRIM26 <i>e!</i> | ENST00000453195 <i>e!</i> | NM_001242783.1 | ENSP00000391879 <i>e!</i> | 1 |
| TRIM26 <i>e!</i> | ENST00000437089 <i>e!</i> | ? | ENSP00000395491 <i>e!</i> | 1 |
| TRIM26 <i>e!</i> | ENST00000454678 <i>e!</i> | NM_003449.4 | ENSP00000410446 <i>e!</i> | 1 |
| ZNRD1 <i>e!</i> | ENST00000332435 <i>e!</i> | NM_170783.3 | ENSP00000331111 <i>e!</i> | 1 |
| ZNRD1 <i>e!</i> | ENST00000359374 <i>e!</i> | NM_014596.5 | ENSP00000352333 <i>e!</i> | 1 |
| ZNRD1 <i>e!</i> | ENST00000376782 <i>e!</i> | NM_001278785.1 | ENSP00000365978 <i>e!</i> | 1 |
| ZNRD1 <i>e!</i> | ENST00000376785 <i>e!</i> | NM_001278786.1 | ENSP00000365981 <i>e!</i> | 1 |

Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|-----------------------------|---------------------------|-----------|------------|
| FLOT1 <i>e!</i> | ENST00000484168 <i>e!</i> | ? | 1 |
| FLOT1 <i>e!</i> | ENST00000484693 <i>e!</i> | ? | 1 |
| GNL1 <i>e!</i> | ENST00000462708 <i>e!</i> | ? | 1 |
| GNL1 <i>e!</i> | ENST00000464231 <i>e!</i> | ? | 1 |
| HCG19P <i>e!</i> | ENST00000448756 <i>e!</i> | ? | 2 |
| PAIP1P1 <i>e!</i> | ENST00000446875 <i>e!</i> | ? | 1 |
| TRIM26 <i>e!</i> | ENST00000480999 <i>e!</i> | ? | 2 |
| UBQLN1P1 <i>e!</i> | ENST00000441056 <i>e!</i> | ? | 1 |
| XXbac-BPG283O16.9 <i>e!</i> | ENST00000624252 <i>e!</i> | ? | 1 |
| ZNRD1 <i>e!</i> | ENST00000463141 <i>e!</i> | ? | 1 |
| ZNRD1 <i>e!</i> | ENST00000471008 <i>e!</i> | ? | 1 |
| ZNRD1-AS1 <i>e!</i> | ENST00000444051 <i>e!</i> | ? | 1 |
| ZNRD1-AS1 <i>e!</i> | ENST00000376797 <i>e!</i> | ? | 2 |

