
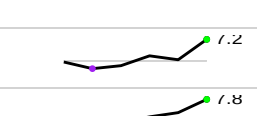



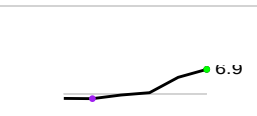
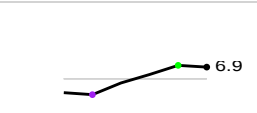
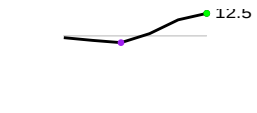
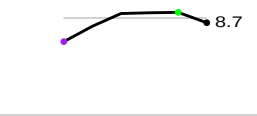
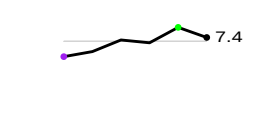
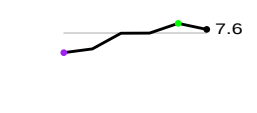


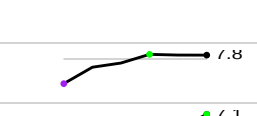
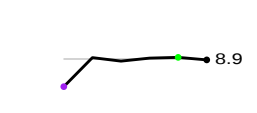








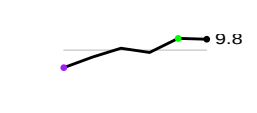



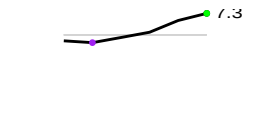




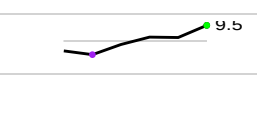

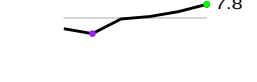


| Upregulated                            |          |   |  | EXPRESSION_OVER_TIME  |
|--|----------|---|--|---|
| List of differentially expressed genes |          |   |  |   |
| TRANSCRIPT                             | GENE     | PROTEIN   | SUMMARY  |   |
| ENST00000243222                        | COL10A1  | collagen type X alpha 1 chain                         | This gene encodes the alpha chain of type X collagen, a short chain collagen expressed by hypertrophic chondrocytes during endochondral ossification. Unlike type VIII collagen, the other short chain collagen, type X collagen is a homotrimer. Mutations in this gene are associated with Schmid type metaphyseal chondrodysplasia (SMCD) and Japanese type spondylometaphyseal dysplasia (SMD). [provided by RefSeq, Jul 2008]   |     |
| ENST00000251296                        | IGSF21   | immunoglobulin superfamily member                     | This gene encodes a protein which has two immunoglobulin (Ig) domains and is a member of the immunoglobulin superfamily. Proteins in this superfamily are usually found on or in cell membranes and act as receptors in immune response pathways. [provided by RefSeq, Sep 2011]   |    |
| ENST00000253109                        | ANGPTL6  | angiotensin like 6                                    |  |    |
| ENST00000255418                        | MYBPH    | myosin binding  |  |    |
| ENST00000259698                        | RIPOR2   | RHO family interacting cell polarization regulator    | This gene encodes an atypical inhibitor of the small G protein RhoA. Inhibition of RhoA activity by the encoded protein mediates myoblast fusion and polarization of T cells and neutrophils. The encoded protein is a component of hair cell stereocilia that is essential for hearing. A splice site mutation in this gene results in hearing loss in human patients. [provided by RefSeq, Sep 2016]   |    |
| ENST00000259889                        | FGFBP2   | fibroblast growth factor binding protein 2            | This gene encodes a member of the fibroblast growth factor binding protein family. The encoded protein is a serum protein that is selectively secreted by cytotoxic lymphocytes and may be involved in cytotoxic lymphocyte-mediated immunity. An increase in the amount of gene product may be associated with atopic asthma and mild extrinsic asthma.[provided by RefSeq Staff, Oct 2008]   |    |
| ENST00000261024                        | SLC40A1  | solute carrier family 40 member 1                     | The protein encoded by this gene is a cell membrane protein that may be involved in iron export from duodenal epithelial cells. Defects in this gene are a cause of hemochromatosis type 4 (HFE4). [provided by RefSeq, Jul 2008]  |    |
| ENST00000265182                        | ENPEP    | glutamyl aminopeptidase                               | The ENPEP gene encodes glutamyl aminopeptidase, a type II integral membrane protein with an extracellular zinc-binding domain. This protein can upregulate blood pressure by cleaving the N-terminal aspartate from angiotensin II, and can regulate blood vessel formation and enhance tumorigenesis in some tissues. Along with ANPEP and DPP4, ENPEP was found to be a candidate co-receptor for the coronavirus SARS-CoV-2, which causes COVID-19. [provided by RefSeq, Apr 2020]  |    |
| ENST00000267484                        | RTN1     | reticulon 1   | This gene belongs to the family of reticulon encoding genes. Reticulons are associated with the endoplasmic reticulum, and are involved in neuroendocrine secretion or in membrane trafficking in neuroendocrine cells. This gene is considered to be a specific marker for neurological diseases and cancer, and is a potential molecular target for therapy. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Jul 2011]  |    |
| ENST00000268053                        | CYP11A1  | cytochrome P450 family 11 subfamily A member 1        | This gene encodes a member of the cytochrome P450 superfamily of enzymes. The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids. This protein localizes to the mitochondrial inner membrane and catalyzes the conversion of cholesterol to pregnenolone, the first and rate-limiting step in the synthesis of the steroid hormones. Two transcript variants encoding different isoforms have been found for this gene. The cellular location of the smaller isoform is unclear since it lacks the mitochondrial-targeting transit peptide. [provided by RefSeq, Jul 2008]  |    |
| ENST00000286758                        | CXCL13   | C-X-C motif chemokine ligand 13                       | B lymphocyte chemoattractant, independently cloned and named Angie, is an antimicrobial peptide and CXO chemokine strongly expressed in the follicles of the spleen, lymph nodes, and Peyer's patches. It preferentially promotes the migration of B lymphocytes (compared to T cells and macrophages), apparently by stimulating calcium influx into, and chemotaxis of, cells expressing Burkitt's lymphoma receptor 1 (BLR-1). It may therefore function in the homing of B lymphocytes to follicles. [provided by RefSeq, Oct 2014]  |    |
| ENST00000288815                        | OLFM2A   | olfactomedin like 2A                                  |  |    |
| ENST00000295095                        | ARHGAP15 | Rho GTPase activating protein 15                      | RHO GTPases (see ARHA- MIM 165390) regulate diverse biologic processes, and their activity is regulated by RHO GTPase-activating proteins (GAPs), such as ARHGAP15 (Seoh et al., 2003 [PubMed 12650940]).[supplied by OMIM, Mar 2008]  |    |
| ENST00000296130                        | CLEC3B   | C-type lectin domain family 3 member B                |  |    |
| ENST00000301908                        | PNOC     | prepronociceptin                                      | This gene encodes a preproprotein that is proteolytically processed to generate multiple protein products. These products include nociceptin, nocistatin, and orphanin FQ2 (OFQ2). Nociceptin, also known as orphanin FQ, is a 17-amino acid neuropeptide that binds to the nociceptin receptor to induce increased pain sensitivity, and may additionally regulate body temperature, learning and memory, and hunger. Another product of the encoded preproprotein, nocistatin, may inhibit the effects of nociceptin. [provided by RefSeq, Jul 2015]   |    |
| ENST00000308873                        | RUNX3    | RUNX family transcription factor 3                    | This gene encodes a member of the runt domain-containing family of transcription factors. A heterodimer of this protein and a beta subunit forms a complex that binds to the core DNA sequence 5'-PYGPyGGT-3' found in a number of enhancers and promoters, and can either activate or suppress transcription. It also interacts with other transcription factors. It functions as a tumor suppressor, and the gene is frequently deleted or transcriptionally silenced in cancer. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Mar 2016]  |    |
| ENST00000318041                        | IL11RA   | interleukin 11 receptor subunit alpha                 | Interleukin 11 is a stromal cell-derived cytokine that belongs to a family of pleiotropic and redundant cytokines that use the gp130 transducing subunit in their high affinity receptors. This gene encodes the IL-11 receptor, which is a member of the hematopoietic cytokine receptor family. This particular receptor is very similar to ciliary neurotrophic factor, since both contain an extracellular region with a 2-domain structure composed of an immunoglobulin-like domain and a cytokine receptor-like domain. Multiple alternatively spliced transcript variants have been found for this gene. [provided by RefSeq, Jun 2012]  |    |
| ENST00000329759                        | FAM43A   | family with sequence similarity 43 member             |  |  |
| ENST00000331898                        | OPOML    | opioid binding protein/cell adhesion molecule like    | This gene encodes a member of the IgLON subfamily in the immunoglobulin protein superfamily of proteins. The encoded preprotein is proteolytically processed to generate the mature protein. This protein is localized in the plasma membrane and may have an accessory role in opioid receptor function. This gene has an ortholog in rat and bovine. The opioid binding-cell adhesion molecule encoded by the rat gene binds opioid alkaloids in the presence of acidic lipids, exhibits selectivity for mu ligands and acts as a GPI-anchored protein. Since the encoded protein is highly conserved in species during evolution, it may have a fundamental role in mammalian systems. Alternative splicing results in multiple transcript variants, at least one of which encodes an isoform that is proteolytically processed. [provided by RefSeq, Jan 2016]   |  |
| ENST00000335295                        | HBB      | hemoglobin subunit beta                               | The alpha (HBA) and beta (HBB) loci determine the structure of the 2 types of polypeptide chains in adult hemoglobin, Hb A. The normal adult hemoglobin tetramer consists of two alpha chains and two beta chains. Mutant beta globin causes sickle cell anemia. Absence of beta chain causes beta-zero-thalassemia. Reduced amounts of detectable beta globin causes beta-plus-thalassemia. The order of the genes in the beta-globin cluster is 5'-epsilon -- gamma-G -- gamma-A -- delta -- beta--3'. [provided by RefSeq, Jul 2008]  |  |
| ENST00000335712                        | BCL11A   | BAF chromatin remodeling complex subunit BCL11A       | This gene encodes a C2H2-type zinc-finger protein by its similarity to the mouse Bcl11a/Evi9 protein. The corresponding mouse gene is a common site of retroviral integration in myeloid leukemia, and may function as a leukemia disease gene, in part, through its interaction with BCL6. During hematopoietic cell differentiation, this gene is down-regulated. It is possibly involved in lymphoma pathogenesis since translocations associated with B-cell malignancies also deregulates its expression. Multiple transcript variants encoding several different isoforms have been found for this gene. [provided by RefSeq, Jul 2008]  |  |
| ENST00000337908                        | MS4A4A   | membrane spanning 4-domains A4A                       | This gene encodes a member of the membrane-spanning 4A gene family. Members of this nascent protein family are characterized by common structural features, similar intron/exon splice boundaries, and display unique expression patterns in hematopoietic cells and nonlymphoid tissues. Alternatively spliced transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Aug 2011]  |  |
| ENST00000338087                        | SLA      | Src like adaptor                                      |  |  |
| ENST00000346867                        | TAC1     | tachykinin precursor 1                                | This gene encodes four products of the tachykinin peptide hormone family: substance P and neurokinin A, as well as the related peptides, neuropeptide K and neuropeptide gamma. These hormones are thought to function as neurotransmitters which interact with nerve receptors and smooth muscle cells. They are known to induce behavioral responses and function as vasodilators and secretagogues. Substance P is an antimicrobial peptide with antibacterial and antifungal properties. Multiple transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Nov 2014]  |  |
| ENST00000357195                        | BCL11B   | BAF chromatin remodeling complex subunit BCL11B       | This gene encodes a C2H2-type zinc finger protein and is closely related to BCL11A, a gene whose translocation may be associated with B-cell malignancies. Although the specific function of this gene has not been determined, the encoded protein is known to be a transcriptional repressor, and is regulated by the NURD nucleosome remodeling and histone deacetylase complex. Four alternatively spliced transcript variants encoding distinct isoforms have been found for this gene. [provided by RefSeq, Aug 2013]  |  |
| ENST00000358511                        | COL6A6   | collagen type VI alpha 6 chain                        | This gene encodes a large protein that contains multiple von Willebrand factor domains and forms a component of the basal lamina of epithelial cells. This protein may regulate epithelial cell-fibronectin interactions. Variation in this gene may be implicated in skin diseases. [provided by RefSeq, May 2017]  |  |
| ENST00000367938                        | NA       | NA  | NA   |  |
| ENST00000368716                        | S100A4   | S100 calcium binding protein A4                       | The protein encoded by this gene is a member of the S100 family of proteins containing 2 EF-hand calcium-binding motifs. S100 proteins are localized in the cytoplasm and/or nucleus of a wide range of cells, and involved in the regulation of a number of cellular processes such as cell cycle progression and differentiation. S100 genes include at least 13 members which are located as a cluster on chromosome 1q21. This protein may function in motility, invasion, and tubulin polymerization. Chromosomal rearrangements and altered expression of this gene have been implicated in tumor metastasis. Multiple alternatively spliced variants, encoding the same protein, have been identified. [provided by RefSeq, Jul 2008]   |  |
| ENST00000368732                        | S100A8   | S100 calcium binding protein A8                       | The protein encoded by this gene is a member of the S100 family of proteins containing 2 EF-hand calcium-binding motifs. S100 proteins are localized in the cytoplasm and/or nucleus of a wide range of cells, and involved in the regulation of a number of cellular processes such as cell cycle progression and differentiation. S100 genes include at least 13 members which are located as a cluster on chromosome 1q21. This protein may function in the inhibition of casein kinase and as a cytokine. Altered expression of this protein is associated with the disease cystic fibrosis. Multiple transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Jan 2016]  |  |
| ENST00000368733                        | S100A8   | S100 calcium binding protein A8                       | The protein encoded by this gene is a member of the S100 family of proteins containing 2 EF-hand calcium-binding motifs. S100 proteins are localized in the cytoplasm and/or nucleus of a wide range of cells, and involved in the regulation of a number of cellular processes such as cell cycle progression and differentiation. S100 genes include at least 13 members which are located as a cluster on chromosome 1q21. This protein may function in the inhibition of casein kinase and as a cytokine. Altered expression of this protein is associated with the disease cystic fibrosis. Multiple transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Jan 2016]  |  |
| ENST00000370031                        | HENMT1   | HEN   |  |  |
| ENST00000371125                        | PRR32    | proline rich 32                                       |  |  |
| ENST00000374429                        | CXCL12   | C-X-C motif chemokine ligand 12                       | This antimicrobial gene encodes a stromal cell-derived alpha chemokine member of the intercrine family. The encoded protein functions as the ligand for the G-protein coupled receptor, chemokine (C-X-C motif) receptor 4, and plays a role in many diverse cellular functions, including embryogenesis, immune surveillance, inflammation response, tissue homeostasis, and tumor growth and metastasis. Mutations in this gene are associated with resistance to human immunodeficiency virus type 1 infections. Multiple transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Sep 2014]   |  |
| ENST00000379780                        | CETP     | cholesteryl ester transfer protein                    | The protein encoded by this gene is found in plasma, where it is involved in the transfer of cholesteryl ester from high density lipoprotein (HDL) to other lipoproteins. Defects in this gene are a cause of hyperalphalipoproteinemia 1 (HALP1). Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Oct 2013]   |  |
| ENST00000381905                        | TNNI2    | troponin 12, fast skeletal type                       | This gene encodes a fast-twitch skeletal muscle protein, a member of the troponin I gene family, and a component of the troponin complex including troponin T, troponin C and troponin I subunits. The troponin complex, along with tropomyosin, is responsible for the calcium-dependent regulation of striated muscle contraction. Mouse studies show that this component is also present in vascular smooth muscle and may play a role in regulation of smooth muscle function. In addition to muscle tissues, this protein is found in corneal epithelium, cartilage where it is an inhibitor of angiogenesis to inhibit tumor growth and metastasis, and mammary gland where it functions as a co-activator of estrogen receptor-related receptor alpha. This protein also suppresses tumor growth in human ovarian carcinoma. Mutations in this gene cause myopathy and distal arthrogryposis type 2B. Alternatively spliced transcript variants have been found for this gene. [provided by RefSeq, Mar 2009]   |  |
| ENST00000394693                        | PCDHA1   | protocadherin alpha 1                                 | This gene is a member of the protocadherin alpha gene cluster; one of three related gene clusters tandemly linked on chromosome five that demonstrate an unusual genomic organization similar to that of B-cell and T-cell receptor gene clusters. The alpha gene cluster is composed of 15 cadherin superfamily genes related to the mouse CNR genes and consists of 13 highly similar and 2 more distantly related coding sequences. The tandem array of 15 N-terminal exons, or variable exons, are followed by downstream C-terminal exons, or constant exons, which are shared by all genes in the cluster. The large, uninterrupted N-terminal exons each encode six cadherin ectodomains while the C-terminal exons encode the cytoplasmic domain. These neural cadherin-like cell adhesion proteins are integral plasma membrane proteins that most likely play a critical role in the establishment and function of specific cell-cell connections in the brain. Alternative splicing has been observed and additional variants have been suggested but their full-length nature has yet to be determined. [provided by RefSeq, Jul 2008] |  |
| ENST00000394695                        | CFI      | complement factor 1                                   | This gene encodes a serine proteinase that is essential for regulating the complement cascade. The encoded preproprotein is cleaved to produce both heavy and light chains, which are linked by disulfide bonds to form a heterodimeric glycoprotein. This heterodimer can cleave and inactivate the complement components C4b and C3b, and it prevents the assembly of the C3 and C5 convertase enzymes. Defects in this gene cause complement factor I deficiency, an autosomal recessive disease associated with a susceptibility to pyogenic infections. Mutations in this gene have been associated with a predisposition to atypical hemolytic uremic syndrome, a disease characterized by acute renal failure, microangiopathic hemolytic anemia and thrombocytopenia. Primary glomerulonephritis with immune deposits and age-related macular degeneration are other conditions associated with mutations of this gene. [provided by RefSeq, Dec 2015]   |  |
| ENST00000395388                        | HLA-DRA  | major histocompatibility complex, class II, DR alpha  | HLA-DRA is one of the HLA class II alpha chain paralogs. This class II molecule is a heterodimer consisting of an alpha and a beta chain, both anchored in the membrane. This molecule is expressed on the surface of various antigen presenting cells such as B lymphocytes, dendritic cells, and monocytes/macrophages, and plays a central role in the immune system and response by presenting peptides derived from extracellular proteins, in particular, pathogen-derived peptides to T cells. The alpha chain is approximately 33-35 kDa and its gene contains 5 exons. Exon 1 encodes the leader peptide, exons 2 and 3 encode the two extracellular domains, and exon 4 encodes the transmembrane domain and the cytoplasmic tail. DRA does not have polymorphisms in the peptide binding part and acts as the sole alpha chain for DRB1, DRB3, DRB4 and DRB5. [provided by RefSeq, Aug 2020]  |  |
| ENST00000395794                        | CXCL12   | C-X-C motif chemokine ligand 12                       | This antimicrobial gene encodes a stromal cell-derived alpha chemokine member of the intercrine family. The encoded protein functions as the ligand for the G-protein coupled receptor, chemokine (C-X-C motif) receptor 4, and plays a role in many diverse cellular functions, including embryogenesis, immune surveillance, inflammation response, tissue homeostasis, and tumor growth and metastasis. Mutations in this gene are associated with resistance to human immunodeficiency virus type 1 infections. Multiple transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Sep 2014]   |  |
| ENST00000399916                        | RUNX3    | RUNX family transcription factor 3                    | This gene encodes a member of the runt domain-containing family of transcription factors. A heterodimer of this protein and a beta subunit forms a complex that binds to the core DNA sequence 5'-PYGPyGGT-3' found in a number of enhancers and promoters, and can either activate or suppress transcription. It also interacts with other transcription factors. It functions as a tumor suppressor, and the gene is frequently deleted or transcriptionally silenced in cancer. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Mar 2016]  |  |
| ENST00000400131                        | CHODL    | chondrolectin   | This gene encodes a type I membrane protein with a carbohydrate recognition domain characteristic of C-type lectins in its extracellular portion. In other proteins, this domain is involved in endocytosis of glycoproteins and exogenous sugar-bearing pathogens. This protein localizes predominantly to the perinuclear region. Several transcript variants encoding a few different isoforms have been found for this gene. [provided by RefSeq, Feb 2011]  |  |
| ENST00000404220                        | IFNAR2   | interferon alpha and beta receptor subunit 2          | The protein encoded by this gene is a type I membrane protein that forms one of the two chains of a receptor for interferons alpha and beta. Binding and activation of the receptor stimulates Janus protein kinases, which in turn phosphorylate several proteins, including STAT1 and STAT2. The type I receptors to the type II cytokine receptor family. Mutations in this gene are associated with Immunodeficiency 45. [provided by RefSeq, Jul 2020]  |  |
| ENST00000409605                        | XIRP2    | zinc actin binding repeat containing 2                |  |  |
| ENST00000424100                        | GPR85    | G protein-coupled receptor 85                         | Members of the G protein-coupled receptor (GPCR) family, such as GPR85, have a similar structure characterized by 7 transmembrane domains. Activation of GPCRs by extracellular stimuli, such as neurotransmitters, hormones, or light, induces an intracellular signaling cascade mediated by heterotrimeric GTP-binding proteins, or G proteins (Matsumoto et al., 2000 [PubMed 10833454]).[supplied by OMIM, Aug 2008]  |  |
| ENST00000432103                        | NA       | NA  | NA   |  |
| ENST00000435365                        | NA       | NA  | NA   |  |
| ENST00000436072                        | SFRP4    | secreted frizzled-related protein 4                   | Secreted frizzled-related protein 4 (SFRP4) is a member of the SFRP family that contains a cysteine-rich domain homologous to the putative Wnt-binding site of Frizzled proteins. SFRPs act as soluble modulators of Wnt signaling. The expression of SFRP4 in ventricular myocardium correlates with apoptosis related gene expression. [provided by RefSeq, Jul 2008]  |  |
| ENST00000437650                        | NA       | NA  | NA   |  |
| ENST00000445105                        | FGF12    | fibroblast growth factor 12                           | The protein encoded by this gene is a member of the fibroblast growth factor (FGF) family. FGF family members possess broad mitogenic and cell survival activities, and are involved in a variety of biological processes, including embryonic development, cell growth, morphogenesis, tissue repair, tumor growth, and invasion. This growth factor lacks the N-terminal signal sequence present in most of the FGF family members, but it contains clusters of basic residues that have been demonstrated to act as a nuclear localization signal. When transfected into mammalian cells, this protein accumulated in the nucleus, but was not secreted. The specific function of this gene has not yet been determined. [provided by RefSeq, Dec 2019]   |  |
| ENST00000446488                        | NA       | NA  | NA   |  |
| ENST00000449591                        | GPR85    | G protein-coupled receptor 85                         | Members of the G protein-coupled receptor (GPCR) family, such as GPR85, have a similar structure characterized by 7 transmembrane domains. Activation of GPCRs by extracellular stimuli, such as neurotransmitters, hormones, or light, induces an intracellular signaling cascade mediated by heterotrimeric GTP-binding proteins, or G proteins (Matsumoto et al., 2000 [PubMed 10833454]).[supplied by OMIM, Aug 2008]  |  |
| ENST00000462700                        | NA       | NA  | NA   |  |
| ENST00000464120                        | NA       | NA  | NA   |  |
| ENST00000464145                        | NA       | NA  | NA   |  |
| ENST00000466315                        | NA       | NA  | NA   |  |
| ENST00000474911                        | RTN1     | reticulon 1   | This gene belongs to the family of reticulon encoding genes. Reticulons are associated with the endoplasmic reticulum, and are involved in neuroendocrine secretion or in membrane trafficking in neuroendocrine cells. This gene is considered to be a specific marker for neurological diseases and cancer, and is a potential molecular target for therapy. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Jul 2011]  |  |
| ENST00000475226                        | NA       | NA  | NA   |  |
| ENST00000476911                        | NA       | NA  | NA   |  |
| ENST00000478001                        | NA       | NA  | NA   |  |
| ENST00000478553                        | NA       | NA  | NA   |  |
| ENST00000478975                        | NA       | NA  | NA   |  |
| ENST00000479026                        | NA       | NA  | NA   |  |
| ENST00000481205                        | NA       | NA  | NA   |  |
| ENST00000481799                        | GGTA1    | glycoprotein alpha-galactosyltransferase 1 (inactive) | Alpha-1,3-galactosyltransferase (GGTA1) is an enzyme present in most mammals except man, apes, and Old World monkeys. This gene is thought to encode a truncated, non-enzymatic form of the GGTA1 protein that lacks the C-terminal catalytic domain. Aberrant expression of the GGTA1 protein in man can lead to autoimmune diseases and sometimes germ cell tumors. [provided by RefSeq, May 2020]   |  |
| ENST00000485743                        | NA       | NA  | NA   |  |
| ENST00000493075                        | LRTM1    | leucine rich repeats and transmembrane domains 1      |  |  |
| ENST00000493434                        | NA       | NA  | NA   |  |
| ENST00000498141                        | NA       | NA  | NA   |  |
| ENST00000504853                        | NA       | NA  | NA   |  |
| ENST00000506143                        | NA       | NA  | NA   |  |
| ENST00000512148                        | CFI      | complement factor 1                                   | This gene encodes a serine proteinase that is essential for regulating the complement cascade. The encoded preproprotein is cleaved to produce both heavy and light chains, which are linked by disulfide bonds to form a heterodimeric glycoprotein. This heterodimer can cleave and inactivate the complement components C4b and C3b, and it prevents the assembly of the C3 and C5 convertase enzymes. Defects in this gene cause complement factor I deficiency, an autosomal recessive disease associated with a susceptibility to pyogenic infections. Mutations in this gene have been associated with a predisposition to atypical hemolytic uremic syndrome, a disease characterized by acute renal failure, microangiopathic hemolytic anemia and thrombocytopenia. Primary glomerulonephritis with immune deposits and age-related macular degeneration are other conditions associated with mutations of this gene. [provided by RefSeq, Dec 2015]   |  |
| ENST00000517638                        | RALYL    | RALY RNA binding protein like                         |  |  |
| ENST00000518566                        | RALYL    | RALY RNA binding protein like                         |  |  |
| ENST00000521376                        | NA       | NA  | NA   |  |
| ENST00000524381                        | OPOML    | opioid binding protein/cell adhesion molecule like    | This gene encodes a member of the IgLON subfamily in the immunoglobulin protein superfamily of proteins. The encoded preprotein is proteolytically processed to generate the mature protein. This protein is localized in the plasma membrane and may have an accessory role in opioid receptor function. This gene has an ortholog in rat and bovine. The opioid binding-cell adhesion molecule encoded by the rat gene binds opioid alkaloids in the presence of acidic lipids, exhibits selectivity for mu ligands and acts as a GPI-anchored protein. Since the encoded protein is highly conserved in species during evolution, it may have a fundamental role in mammalian systems. Alternative splicing results in multiple transcript variants, at least one of which encodes an isoform that is proteolytically processed. [provided by RefSeq, Jan 2016]   |  |
| ENST00000525                           |          |   |  |   |