|  |  |  |
| --- | --- | --- |
| **Gene** | **Description** | **Details** |
| [IGL@](http://en.wikipedia.org/wiki/IGL@) | Asymmetric crying facies (Cayler cardiofacial syndrome) | **Immunoglobulin lambda locus**, also known as **IGL@**, is a region on human [chromosome 22](http://en.wikipedia.org/wiki/Chromosome_22) that contains [genes](http://en.wikipedia.org/wiki/Gene) for the lambda [light chains](http://en.wikipedia.org/wiki/Antibody_light_chain) of [antibodies](http://en.wikipedia.org/wiki/Antibody) (or immunoglobulins).  Immunoglobulins recognize foreign [antigens](http://en.wikipedia.org/wiki/Antigen) and initiate immune responses such as phagocytosis and the complement system. |
| [TBX1](http://en.wikipedia.org/wiki/TBX1) | T-box 1 | **T-box transcription factor TBX1** also known as **T-box protein 1** and **testis-specific T-box protein** is a[protein](http://en.wikipedia.org/wiki/Protein) that in humans is encoded by the **TBX1** [gene](http://en.wikipedia.org/wiki/Gene).[[1]](http://en.wikipedia.org/wiki/TBX1#cite_note-entrez-1) Genes in the [T-box](http://en.wikipedia.org/wiki/T-box) family play important roles in the formation of tissues and organs during embryonic development. |
| [RTN4R](http://en.wikipedia.org/wiki/RTN4R) | [Reticulon 4 receptor](http://en.wikipedia.org/wiki/Reticulon_4_receptor) | This gene encodes the [receptor](http://en.wikipedia.org/wiki/Receptor_(biochemistry)) for [reticulon 4](http://en.wikipedia.org/wiki/Reticulon_4), [oligodendrocytemyelin](http://en.wikipedia.org/wiki/Oligodendrocyte)[glycoprotein](http://en.wikipedia.org/wiki/Glycoprotein) and [myelin-associated glycoprotein](http://en.wikipedia.org/wiki/Myelin-associated_glycoprotein). This receptor mediates [axonal growth](http://en.wikipedia.org/wiki/Axon_guidance) inhibition and may play a role in regulating axonal regeneration and plasticity in the adult central nervous system. |
| [COMT](http://en.wikipedia.org/wiki/COMT) | catechol-O-methyltransferase gene | **Catechol-*O*-methyltransferase** (**COMT**; [EC](http://en.wikipedia.org/wiki/Enzyme_Commission_number) [2.1.1.6](http://enzyme.expasy.org/EC/2.1.1.6)) is one of several [enzymes](http://en.wikipedia.org/wiki/Enzyme) that degrade [catecholamines](http://en.wikipedia.org/wiki/Catecholamine)such as [dopamine](http://en.wikipedia.org/wiki/Dopamine), [epinephrine](http://en.wikipedia.org/wiki/Epinephrine), and [norepinephrine](http://en.wikipedia.org/wiki/Norepinephrine). In humans, catechol-*O*-methyltransferase protein is encoded by the **COMT** [gene](http://en.wikipedia.org/wiki/Gene).[[2]](http://en.wikipedia.org/wiki/Catechol-O-methyl_transferase#cite_note-pmid1572656-2) As the regulation of catecholamines is impaired in a number of medical conditions, several pharmaceutical drugs target COMT to alter its activity and therefore the availability of catecholamines. |
| [NEFH](http://en.wikipedia.org/wiki/NEFH) | neurofilament, heavy polypeptide 200kDa | **NEFH** is a human gene located on [chromosome 22](http://en.wikipedia.org/wiki/Chromosome_22) that stands for "neurofilament, heavy polypeptide". It is the gene for a heavy protein subunit that is combined with medium and light subunits to make neurofilaments, which form the framework for nerve cells. |
| [CHEK2](http://en.wikipedia.org/wiki/CHEK2) | CHK2 checkpoint homolog (S. pombe) | The [protein](http://en.wikipedia.org/wiki/Protein) encoded by this gene, CHK2, is a [protein kinase](http://en.wikipedia.org/wiki/Protein_kinase) that is activated in response to [DNA damage](http://en.wikipedia.org/wiki/DNA_damage) and is involved in [cell cycle arrest](http://en.wikipedia.org/wiki/Cell_cycle_checkpoint). |
| [NF2](http://en.wikipedia.org/wiki/NF2_(gene)) | neurofibromin 2 | **Merlin** (also called **Neurofibromin 2** or **schwannomin**) is a [cytoskeletal](http://en.wikipedia.org/wiki/Cytoskeletal) [protein](http://en.wikipedia.org/wiki/Protein). In humans, it is a [tumor suppressor protein](http://en.wikipedia.org/wiki/Tumor_suppressor_gene) involved in [Neurofibromatosis type II](http://en.wikipedia.org/wiki/Neurofibromatosis_type_II).[[1]](http://en.wikipedia.org/wiki/Merlin_(protein)#cite_note-pmid8379998-1)[[2]](http://en.wikipedia.org/wiki/Merlin_(protein)#cite_note-pmid16324214-2) Sequence data reveal its similarity to the [ERM protein family](http://en.wikipedia.org/wiki/ERM_protein_family). |
| [SOX10](http://en.wikipedia.org/wiki/SOX10) | SRY (sex determining region Y)-box 10 | This gene encodes a member of the [SOX](http://en.wikipedia.org/wiki/SOX_gene_family) (SRY-related HMG-box) family of [transcription factors](http://en.wikipedia.org/wiki/Transcription_factor) involved in the regulation of [embryonic development](http://en.wikipedia.org/wiki/Embryonic_development) and in the determination of the [cell fate](http://en.wikipedia.org/wiki/Cell_fate_determination). The encoded protein may act as a transcriptional activator after forming a protein complex with other proteins. This protein acts as a nucleocytoplasmic shuttle protein and is important for [neural crest](http://en.wikipedia.org/wiki/Neural_crest) and [peripheral nervous system](http://en.wikipedia.org/wiki/Peripheral_nervous_system)development. |
| [APOL1](http://en.wikipedia.org/wiki/APOL1) | Apolipoprotein L1 | Apolipoprotein L1 (apoL1) is a minor apoprotein component of [HDL (High-density lipoprotein)](http://en.wikipedia.org/wiki/High-density_lipoprotein) or 'good cholesterol' which is synthesized in the liver and also in many other tissues, including pancreas, kidney, and brain. |
| [EP300](http://en.wikipedia.org/wiki/EP300) | E1A binding protein p300 | This protein regulates the activity of many genes in tissues throughout the body. It plays an essential role in regulating [cell growth](http://en.wikipedia.org/wiki/Cell_growth) and [division](http://en.wikipedia.org/wiki/Cell_division), prompting cells to mature and assume specialized functions (differentiate), and preventing the growth of cancerous tumors. |
| [WNT7B](http://en.wikipedia.org/wiki/WNT7B) | Wingless-type MMTV integration site family, member 7B | The WNT gene family consists of structurally related genes that encode secreted signaling proteins. These proteins have been implicated in oncogenesis and in several developmental processes, including regulation of cell fate and patterning during embryogenesis. |
| [SHANK3](http://en.wikipedia.org/wiki/SHANK3) | SH3 and multiple ankyrin repeat domains 3 | This gene is a member of the Shank gene family. Shank proteins are multidomain scaffold proteins of the postsynaptic density that connect [neurotransmitter](http://en.wikipedia.org/wiki/Neurotransmitter) receptors, ion channels, and other membrane proteins to the actin cytoskeleton and G-protein-coupled signaling pathways. Shank proteins also play a role in [synapse](http://en.wikipedia.org/wiki/Synapse)formation and [dendritic spine](http://en.wikipedia.org/wiki/Dendritic_spine) maturation. |
| [SULT4A1](http://en.wikipedia.org/wiki/SULT4A1) | sulfotransferase family 4A, member 1 | This gene encodes a member of the sulfotransferase family. The encoded protein is a brain-specific sulfotransferase believed to be involved in the metabolism of neurotransmitters. Polymorphisms in this gene may be associated with susceptibility to [schizophrenia](http://en.wikipedia.org/wiki/Schizophrenia). |
| [PARVB](http://en.wikipedia.org/wiki/PARVB) | parvin beta (cytoskeleton organization and cell adhesion) | Members of the parvin family, including PARVB, are actin-binding proteins associated with focal contacts. |

