Symbol	Description	Summary	OMIM Phenotype	Phenotype MIM #
GIPC3	GIPC PDZ domain containing family, member 3	The protein encoded by this gene belongs to the GIPC family. Studies in mice suggest that this gene is required for postnatal maturation of the hair bundle and long-term survival of hair cells and spiral ganglion in the ear. Mutations in this gene are associated with autosomal recessive deafness.	Deafness, autosomal recessive 15	601869
TBXA2R	thromboxane A2 receptor	This gene encodes a member of the G protein-coupled receptor family. The protein interacts with thromboxane A2 to induce platelet aggregation and regulate hemostasis. A mutation in this gene results in a bleeding disorder. Multiple transcript variants encoding different isoforms have been found for this gene.	{Bleeding disorder, platelet-type, 13, susceptibility to}	614009
PIP5K1C	phosphatidylinositol-4-phosphate 5-kinase, type I, gamma	This locus encodes a type I phosphatidylinositol 4-phosphate 5-kinase. The encoded protein catalyzes phosphorylation of phosphatidylinositol 4-phosphate, producing phosphatidylinositol 4,5-bisphosphate. This enzyme is found at synapses and has been found to play roles in endocytosis and cell migration. Mutations at this locus have been associated with lethal congenital contractural syndrome. Alternatively spliced transcript variants encoding different isoforms have been described	Lethal congenital contractural syndrome 3	611369
RAX2	retina and anterior neural fold homeobox 2	This gene encodes a homeodomain-containing protein that plays a role in eye development. Mutation of this gene causes age-related macular degeneration type 6, an eye disorder resulting in accumulations of protein and lipid beneath the retinal pigment epithelium and within the Bruch's membrane. Defects in this gene can also cause cone-rod dystrophy type 11, a disease characterized by the initial degeneration of cone photoreceptor cells and resulting in loss of color vision and visual acuity, followed by the degeneration of rod photoreceptor cells, which progresses to night blindness and the loss of peripheral vision.	Cone-rod dystrophy 11 Macular degeneration, age-related, 6	610381 613757
ATCAY	ataxia, cerebellar, Cayman type	This gene encodes a neuron-restricted protein that contains a CRAL-TRIO motif common to proteins that bind small lipophilic molecules. Mutations in this gene are associated with cerebellar ataxia, Cayman type. Autosomal recessive Cayman cerebellar ataxia, identified by Johnson et al. (1978) in a population isolate on Grand Cayman Island, is characterized by marked psychomotor retardation and prominent nonprogressive cerebellar dysfunction including nystagmus, intention tremor, dysarthria, and wide-based ataxic gait. Hypotonia is present from early childhood; retinal abnormalities are absent.	Ataxia, cerebellar, Cayman type	601238
MAP2K2	mitogen-activated protein kinase kinase 2	The protein encoded by this gene is a dual specificity protein kinase that belongs to the MAP kinase kinase family. This kinase is known to play a critical role in mitogen growth factor signal transduction. It phosphorylates and thus activates MAPK1/ERK2 and MAPK2/ERK3. The activation of this kinase itself is dependent on the Ser/Thr phosphorylation by MAP kinase kinase kinases. Mutations in this gene cause cardiofaciocutaneous syndrome (CFC syndrome), a disease characterized by heart defects, mental retardation, and distinctive facial features similar to those found in Noonan syndrome. The inhibition or degradation of this kinase is also found to be involved in the pathogenesis of Yersinia and anthrax. A pseudogene, which is located on chromosome 7, has been identified for this gene.	Cardiofaciocutaneous syndrome	115150
SH3GL1	SH3-domain GRB2-like 1	This gene encodes a member of the endophilin family of Src homology 3 domain-containing proteins. The encoded protein is involved in endocytosis and may also play a role in the cell cycle. Overexpression of this gene may play a role in leukemogenesis, and the encoded protein has been implicated in acute myeloid leukemia as a fusion partner of the myeloid-lymphoid leukemia protein. Pseudogenes of this gene are located on the long arm of chromosomes 11 and 17. Alternatively spliced transcript variants encoding multiple isoforms have been observed for this gene.	Leukemia, acute myeloid	601626