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| **Symbol** | **Disease** | **Full Name** | **Function** |
| FAM57A | membrane-associated protein that promotes lung carcinogenesis | Family With Sequence Similarity 57 Member A | amino acid transport and glutathione metabolism |
| HIST1H2BD | Rahman Syndrome | Histone Cluster 1 H2B Family Member D | Histone Cluster |
| PMF1 | | Polyamine Modulated Factor 1, | Part of the MIS12 complex which is required for normal chromosome alignment and segregation and kinetochore formation during mitosis |
| JADE2 | | Jade Family PHD Finger 2 | Component of the HBO1 complex which has a histone H4-specific acetyltransferase activity |
| LMNB1 | autosomal dominant adult-onset leukodystrophy (ADLD) | Lamin B1 | nuclear lamina |
| AQP3 | | water channel protein aquaporin 3 | promote glycerol permeability and water transport across cell membrane |
| KIAA0368 | | Ecm29 Proteasome Adaptor And Scaffold | couple the proteasome to different compartments including endosome, endoplasmic reticulum and centrosome |
| ENPP2 | Intrahepatic Cholestasis Of Pregnancy and Teratocarcinoma | Ectonucleotide Pyrophosphatase/Phosphodiesterase 2 | The protein encoded by this gene functions as both a phosphodiesterase, which cleaves phosphodiester bonds at the 5' end of oligonucleotides, and a phospholipase, which catalyzes production of lysophosphatidic acid (LPA) in extracellular fluids. LPA evokes growth factor-like responses including stimulation of cell proliferation and chemotaxis. This gene product stimulates the motility of tumor cells and has angiogenic properties, and its expression is upregulated in several kinds of carcinoma |
| SYT11 | Parkinson Disease, Late-Onset | Synaptotagmin 11 | calcium sensors and mediate calcium-dependent regulation of membrane trafficking in synaptic transmission. The encoded protein is also a substrate for ubiquitin-E3-ligase parkin |
| HIST1H3F | Diffuse Intrinsic Pontine Glioma and Corneal Dystrophy, Posterior Polymorphous, 1 | Histone Cluster 1 H3 Family Member | Histone Cluster |
| VWA5A | | Von Willebrand Factor A Domain Containing 5A | May play a role in tumorigenesis as a tumor suppressor. |
| LOC728392 | | | Uncharacterized |
| TRIP6 |  | hyroid Hormone Receptor Interactor 6 | NOD-like receptor signaling pathway and LPA receptor mediated events |
| RUNDC3B | | RUN DOMAIN-CONTAINING PROTEIN 3B | Burmeister et al. (2015) examined the methylation status of 6 regions spanning the CpG island in the RUNDC3B promoter region in cancer cell lines. Lymphoid malignancies exhibited prominent RUNDC3B promoter methylation and did not express RUNDC3B, unlike myeloid malignancies and solid tumors. The authors suggested that DNA methylation in this region may serve as a biomarker for lymphoid malignancie |
| TMEM35B///ZMYM6 | | Transmembrane Protein 35 | |
| MCM5 | Meier-Gorlin Syndrome 8 and Melanoacanthoma | Minichromosome Maintenance Complex Component | nitiation of DNA replication. The encoded protein is a member of the MCM family of chromatin-binding proteins and can interact with at least two other members of this family. The encoded protein is upregulated in the transition from the G0 to G1/S phase of the cell cycle and may actively participate in cell cycle regulation |
| ZMYND11 | Mental Retardation, Autosomal Dominant 30 and Autosomal Dominant Non-Syndromic Intellectual Disability | Zinc Finger MYND-Type Containing 11 | transcriptional repressor |
| NFKB1 | Immunodeficiency, Common Variable, 12 and Common Variable Immunodeficiency | Nuclear Factor Kappa B Subunit 1 | pleiotropic transcription factor |
| PSMB10 | | Proteasome Subunit Beta 10 | proteasome |
| HIST2H2BE | Vaccinia and Systemic Lupus Erythematosus | Histone Cluster 2 H2B Family Member E | Histone Cluster |
| AVEN | Schizoid Personality Disorder and Chromosome 15Q13.3 Deletion Syndrome. | Apoptosis And Caspase Activation Inhibitor | |
| PHF11 | Asthma and Ige Responsiveness, Atopic | PHD Finger Protein 11 | |
| KIF5C | Cortical Dysplasia, Complex, With Other Brain Malformations 2 and Complex Cortical Dysplasia With Other Brain Malformations | Kinesin Family Member 5C | inesin heavy chain subunit involved in the transport of cargo within the central nervous system |
| CORO1A | Immunodeficiency 8 and Coronin-1A Deficiency | Coronin 1A | cell cycle progression, signal transduction, apoptosis, and gene regulation |
| MPDU1 | Congenital Disorder Of Glycosylation, Type If and Congenital Disorder Of Glycosylation, Type Iia | Mannose-P-Dolichol Utilization Defect 1 | endoplasmic reticulum membrane protein that is required for utilization of the mannose donor mannose-P-dolichol in the synthesis of lipid-linked oligosaccharides and glycosylphosphatidylinositols. Mutations in this gene result in congenital disorder of glycosylation type |
| FOS | Acute Pericementitis and Osteoblastoma | Fos Proto-Oncogene, AP-1 Transcription Factor Subunit | regulators of cell proliferation, differentiation, and transformation |