

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	Histone Cluster 1 H3 Family Member	Histone Cluster

	Dystrophy, Posterior Polymorphous, 1		
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
			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
RUNDC3B		RUN DOMAIN-CONTAINING PROTEIN 3B	
TMEM35B///ZMYM6		Transmembrane Protein 35	
			initiation 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
MCM5	Meier-Gorlin Syndrome 8 and Melanoacanthoma	Minichromosome Maintenance Complex Component	
ZMYND11	Mental Retardation, Autosomal Dominant 30 and Autosomal Dominant Non-Syndromic Intellectual Disability	Zinc Finger MYND-Type Containing 11	transcriptional repressor
	Immunodeficiency, Common Variable, 12 and Common Variable		
NFKB1	Immunodeficiency	Nuclear Factor Kappa B Subunit 1	pleiotropic transcription factor
PSMB10		Proteasome Subunit Beta 10	proteasome

HIST2H2 BE	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