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MONOGENIC DISEASES TESTED

DISEASE	GENE	CLASSIFICATION	SEVERITY
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	*HMGCL*	MET	Severe
3-Methylcrotonyl-CoA Carboxylase Deficiency 1	*MCCC1*	MET	Severe
3-Methylcrotonyl-CoA Carboxylase Deficiency 2	*MCCC2*	MET	Severe
Abetalipoproteinemia	*MTTP*	DIG, NEUR, OPTH, HEM	Severe
Acyl-CoA Oxidase I Deficiency	*ACOX1*	NEUR	Very severe
Aicardi-Goutières Syndrome	*SAMHD1*	NEUR	Severe
Alport Syndrome, X-Linked	*COL4A5*	REN, OPTH, HEAR	Severe
Alstrom Syndrome	*ALMS1*	OPTH, HEAR, REN, CARD	Severe
Andermann Syndrome	*SLC12A6*	MUSC, NEUR	Severe
Aromatase Deficiency	*CYP19A1*	SD	Moderate
Arthrogryposis Mental Retardation Seizures	*SLC35A3*	MET	Severe
Asparagine Synthetase Deficiency	*ASNS*	NEUR	Very severe
Aspartylglycosaminuria	*AGA*	MET, NEUR	Severe
Autosomal Recessive Polycystic Kidney Disease	*PKHD1*	REN	Severe
Bardet-Biedl Syndrome (BBS1-related)	*BBS1*	OPTH, MET, END	Severe
Bardet Biedl Syndrome (BBS12-related)	*BBS12*	OPTH	Severe
Beta Thalassemia	*HBB*	HEM	Very severe
Biotinidase Deficiency	*BTD*	MET	Severe
Canavan Disease	*ASPA*	NEUR	Severe
Carpenter Syndrome	*RAB23*	SKEL	Severe
Choreacanthocytosis	*VPS13A*	NEUR	Moderate
Choroideremia, X-Linked	*CHM*	OPTH	Severe
Citrin Deficiency	*SLC25A13*	MET	Moderate
Combined Oxidative Phosphorylation Deficiency 3	*TSFM*	NEUR, MET, CARD	Very severe
Congenital Disorder of Glycosylation, Type 1A (PMM2-related)	*PMM2*	MET	Severe
Congenital Neutropenia (HAX1-related)	*HAX1*	IMM	Severe
Crigler Najjar Syndrome, Type I	*UGT1A1*	MET	Very severe
Cystic Fibrosis *	*CFTR*	RESP, DIG	Very severe
Factor XI Deficiency	*F11*	HEM	Severe
Familial Dysautonomia	*IKBKAP*	NEUR	Moderate
Fanconi Anemia, Type C	*FANCC*	IMM	Severe
Fanconi Anemia, Type G	*FANCG*	HEM	Severe
Gaucher Disease	*GBA*	NEUR, HEP, CARD	Severe
Glutaric Acidemia, Type 2A	*ETFA*	MET	Moderate
Glycine Encephalopathy (GLDC-related)	*GLDC*	MET	Very severe
Glycogen Storage Disease, Type 1A	*G6PC*	MET	Moderate
Glycogen Storage Disease, Type 1B	*SLC37A4*	MET	Moderate
Glycogen Storage Disease, Type 3	*AGL*	MET	Severe
Glycogen Storage Disease, Type 7	*PFKM*	MET	Severe
GRACILE Syndrome	*BCS1L*	MET	Very severe
Hereditary Fructose Intolerance	*ALDOB*	MET	Moderate
Homocystinuria, Type cblE	*MTRR*	MET	Severe
Hydrolethalus Syndrome	*HYLS1*	NEUR, CARD	Very severe
Inclusion Body Myopathy, Type 2	*GNE*	MUSC	Moderate
Isovaleric Acidemia	*IVD*	MET	Severe
Joubert Syndrome, Type 2	*TMEM216*	NEUR	Severe
Junctional Epidermolysis Bullosa, Herlitz Type	*LAMC2*	SKIN	Severe
Lamellar Ichthyosis, Type 1	*TGM1*	MET	Moderate
Leber Congenital Amaurosis (LCA5-related)	*LCA5*	OPTH	Severe
Leigh Syndrome, French-Canadian Type	*LRPPRC*	NEUR, MUSC	Severe
Leukoencephalopathy with Vanishing White Matter	*EIF2B5*	NEUR	Severe

* tests for mutations that cause the classic Cystic Fibrosis phenotype.

DISEASE	GENE	CLASSIFICATION	SEVERITY
Leydig Cell Hypoplasia [Luteinizing Hormone Resistance]	*LHCGR*	SD	Moderate
Limb Girdle Muscular Dystrophy, Type 2E	*SGCB*	MUSC	Severe
Lipoamide Dehydrogenase Deficiency [Maple Syrup Urine Disease, Type 3]	*DLD*	MET	Severe
Lipoprotein Lipase Deficiency	*LPL*	MET	Moderate
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	*HADHA*	MET	Severe
Lysinuric Protein Intolerance	*SLC7A7*	MET	Severe
Maple Syrup Urine Disease, Type 1B	*BCKDHB*	MET	Severe
Methylmalonic Acidemia (MMAA-related)	*MMAA*	MET	Very severe
Methylmalonic Aciduria, Type Mut(0)	*MUT*	MET	Severe
Methylmalonic Aciduria and Homocystinuria, Type cblC	*MMACHC*	MET	Severe
Methylmalonic Aciduria and Homocystinuria, Type cblD	*MMADHC*	MET	Severe
Mucopolysaccharidosis, Type II [Hunter Syndrome], X-Linked	*IDS*	RESP, CARD	Very severe
Mucopolysaccharidosis, Type IIIC [Sanfilippo C]	*HGSNAT*	MET, NEUR, OPTH	Severe
Multiple Sulfatase Deficiency	*SUMF1*	MET	Very severe
Myotubular Myopathy, X-Linked	*MTM1*	MUSC	Severe
Navajo Neurohepatopathy [MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome]	*MPV17*	NEUR	Severe
Neuronal Ceroid Lipofuscinosis (CLN8-related)	*CLN8*	NEUR	Very severe
Neuronal Ceroid Lipofuscinosis (MFSD8-related)	*MFSD8*	NEUR	Very severe
Neuronal Ceroid Lipofuscinosis (TPP1-related)	*TPP1*	NEUR	Very severe
Nijmegen Breakage Syndrome	*NBN*	NEUR	Severe
Omenn Syndrome (RAG2-related)	*RAG2*	IMM	Very severe
Ornithine Aminotransferase Deficiency	*OAT*	OPTH	Moderate
Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia -Homocitrullinuria (HHH) Syndrome]	*SLC25A15*	MET	Severe
Pendred Syndrome	*SLC26A4*	HEAR, END	Moderate
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related)	*PEX1*	MET	Severe
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related)	*PEX2*	MET	Severe
Phenylketonurea	*PAH*	MET	Very severe
Pontocerebellar Hypoplasia, Type 1A	*VRK1*	NEUR, MUSC	Very severe
Pontocerebellar Hypoplasia, Type 2D	*SEPSECS*	NEUR	Very severe
Pontocerebellar Hypoplasia, Type 2E	*VPS53*	NEUR	Very severe
Primary Ciliary Dyskinesia (DNAH5-related)	*DNAH5*	RESP, INF	Moderate
Primary Ciliary Dyskinesia (DNAI1-related)	*DNAI1*	RESP, INF	Moderate
Primary Hyperoxaluria, Type 3	*HOGA1*	REN, MET	Moderate
Pycnodysostosis	*CTSK*	MET	Severe
Pyruvate Dehydrogenase Deficiency (PDHB-Related)	*PDHB*	NEUR, MET	Severe
Retinal Dystrophy (RLBP1-related) [Bothnia Retinal Dystrophy]	*RLBP1*	OPTH	Severe
Retinitis Pigmentosa 25 (EYS-related)	*EYS*	OPTH	Severe
Retinitis Pigmentosa 59 (DHDDS-related)	*DHDDS*	OPTH	Severe
Sanfilippo Syndrome, Type D [Mucopolysaccharidosis IIID]	*GNS*	MET	Severe
Severe Combined Immunodeficiency, Type Athabaskan	*DCLRE1C*	IMM	Very severe
Severe Combined Immunodeficiency, X-Linked	*IL2RG*	IMM	Very severe
Sickle-Cell Disease	*HBB*	HEM	Very severe
Sjögren-Larsson Syndrome	*ALDH3A2*	MET	Severe
Steroid-Resistant Nephrotic Syndrome	*NPHS2*	REN	Severe
Stuve-Wiedemann Syndrome	*LIFR*	SKEL	Severe
Tay-Sachs Disease	*HEXA*	MET	Very severe
Usher Syndrome, Type 1F	*PCDH15*	HEAR	Moderate
Usher Syndrome, Type 3	*CLRN1*	HEAR, OPTH	Moderate
Wolman Disease	*LIPA*	MET, HEP	Severe

CARD	CARDIAC	DIG	DIGESTIVE	END	ENDOCRINE	HEAR	HEARING	HEM	HEMATOLOGICAL
HEP	HEPATIC	IMM	IMMUNOLOGICAL	INF	INFERTILITY	MET	METABOLIC	MUSC	MUSCULAR
NEUR	NEUROLOGICAL	OPTH	OPTHALMOLOGICAL	REN	RENAL	RESP	RESPIRATORY	SD	SEXUAL DEVELOPMENT
SKEL	SKELETAL	SKIN	SKIN						

A disease may be classified into several types. The classification listed is based on the most common symptoms associated with each condition. Degree of severity of a condition can vary and depends on the specific mutation, signs and symptoms.

Results and possible next steps should always be considered in the context of other clinical criteria and should be fully discussed with your healthcare provider. Genetic counseling is recommended when a high risk result is received.

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