Group 2

Gene List

Gene	Condition	Type of Condition	More Information	Notes
22q11.2 microdeletion	DiGeorge syndrome	Multisystem disorder (Cardiac, immune system, neurologic)	22q11 deletion syndrome: MedlinePlus Genetics	Added 6/24/2024
APRT	Adenine phosphoribosyltransferase deficiency	Kidney Disorder	Adenine phosphoribosyltransferase deficiency: MedlinePlus Genetics	Added 6/24/2024
ASPA	Canavan disease	Metabolic Disorder	Canavan disease: MedlinePlus Genetics	
ATP7A	Menkes Disease	Metabolic Disorder	Menkes syndrome: MedlinePlus Genetics	Males only
CA5A	Carbonic anhydrase VA deficiency	Metabolic Disorder	Carbonic anhydrase VA deficiency: MedlinePlus Genetics	Added 6/24/2024; Reduced sensitivity
CDKL5	Developmental and epileptic encephalopathy 2	Neurologic Disorder	CDKL5 deficiency disorder: MedlinePlus Genetics	Males and females
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	Endocrine System Disorder	17 alpha-hydroxylase/17,20-lyase deficiency: MedlinePlus Genetics	
DHCR7	Smith-Lemli-Opitz syndrome	Multisystem disorder (learning/behavior, cardiac, kidneys, muscle tone)	Smith-Lemli-Opitz syndrome: MedlinePlus Genetics	
DMD	Duchenne muscular dystrophy	Musculoskeletal Disorder	Duchenne and Becker muscular dystrophy: MedlinePlus Genetics	Males only; reduced sensitivity
GALC	Krabbe Disease	Metabolic Disorder	Krabbe disease: MedlinePlus Genetics	
GLB1	GM1 Gangliosidosis	Metabolic Disorder	GM1 gangliosidosis: MedlinePlus Genetics	Added 1/6/2025
GSS	Glutathione Synthetase Deficiency	Metabolic Disorder	Glutathione synthetase deficiency: MedlinePlus Genetics	
GUSB	Mucopolysaccharidosis type 7	Metabolic Disorder	Mucopolysaccharidosis type VII: MedlinePlus Genetics	
HEXA	Tay-Sachs Disease	Metabolic Disorder	Tay-Sachs Disease | MedlinePlus	Added 1/6/2025
HEXB	Sandhoff Disease	Metabolic Disorder	Sandhoff disease: MedlinePlus Genetics	Added 1/6/2025

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MECP2	Rett syndrome	Neurologic Disorder (learning/behavior)	Rett syndrome: MedlinePlus Genetics	Males and females
MOCS1	Molybdenum cofactor deficiency A	Metabolic Disorder	Molybdenum cofactor deficiency: MedlinePlus Genetics	
NGLY1	NGLY1 deficiency syndrome	Multisystem Disorder (Neurologic, Hepatic, Vision)	NGLY1-congenital disorder of deglycosylation: MedlinePlus Genetics	Added 1/6/2025
NPC1	Niemann-Pick Disease, types C1 and D	Metabolic Disorder	Niemann-Pick disease: MedlinePlus Genetics	
NPC2	Niemann-Pick Disease, type C2	Metabolic Disorder	Niemann-Pick disease: MedlinePlus Genetics	
OAT	Ornithine aminotransferase deficiency	Eye Disorder	Gyrate atrophy of the choroid and retina: MedlinePlus Genetics	
PHGDH	3-phosphoglycerate dehydrogenase deficiency	Metabolic Disorder	Phosphoglycerate dehydrogenase deficiency: MedlinePlus Genetics	
PIK3CD	Activated PI3K-delta syndrome (APDS)	Immunodeficiency Disorder	Activated PI3K-delta syndrome: MedlinePlus Genetics	Added 1/6/2025
PIK3R1	Activated PI3K-delta syndrome (APDS)	Immunodeficiency Disorder	Activated PI3K-delta syndrome: MedlinePlus Genetics	Added 1/6/2025
PLP1	Pelizaeus-Merzbacher disease	Neurologic Disorder	Pelizaeus-Merzbacher disease: MedlinePlus Genetics	Added 1/6/2025
RPE65	Leber congenital amaurosis 2	Eye Disorder	Leber congenital amaurosis: MedlinePlus Genetics	
SCN1A	Dravet syndrome	Seizure Disorder	Genetic epilepsy with febrile seizures plus: MedlinePlus Genetics	
SGSH	Mucopolysaccharidosis type 3A	Metabolic Disorder	Mucopolysaccharidosis type III: MedlinePlus Genetics	
SLC25A15	Ornithine translocase deficiency	Metabolic Disorder	Ornithine translocase deficiency: MedlinePlus Genetics	
SLC25A20	Carnitine-Acylcarnitine Translocase Deficiency	Metabolic Disorder	Carnitine-acylcarnitine translocase deficiency: MedlinePlus Genetics	
SLC6A8	Creatine transporter deficiency	Metabolic Disorder	X-linked creatine deficiency: MedlinePlus Genetics	Males and females
SMPD1	Niemann-Pick diseae type A	Metabolic Disorder	Niemann-Pick disease: MedlinePlus Genetics	
TCF4	Pitt-Hopkins syndrome	Multisystem Disorder (learning/behavior, seizures)	Pitt-Hopkins syndrome: MedlinePlus Genetics	

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TP53	Li-Fraumeni syndrome	Inherited Cancer Predisposition Syndrome	Li-Fraumeni syndrome: MedlinePlus Genetics	Added 1/6/2025
TSC1	Tuberous sclerosis 1	Multisystem disorder (learning/behavior, kidneys, heart, vision)	Tuberous sclerosis complex: MedlinePlus Genetics	
TSC2	Tuberous sclerosis 2	Multisystem disorder (learning/behavior, kidneys, heart, vision)	Tuberous sclerosis complex: MedlinePlus Genetics	
TTPA	Ataxia with isolated vitamin E deficiency	Metabolic Disorder	Ataxia with vitamin E deficiency: MedlinePlus Genetics	
UBE3A; Isodisomy of Chr15	Angelman syndrome	Multisystem Disorder (learning/behavior, seizures)	Angelman syndrome: MedlinePlus Genetics	Added 6/24/2024
15q11q13 microdeletion; Isodisomy of Chr15	Prader-Willi syndrome	Multisystem Disorder (learning/behavior, endocrine)	Prader-Willi syndrome: MedlinePlus Genetics	Added 6/24/2024
duplication of 15q11-q13	15q11-q13 duplication syndrome	Multisystem Disorder (learning/behavior, seizures)	15q11-q13 duplication syndrome: MedlinePlus Genetics	Added 1/6/2025
Removed from panel 2				
ARSA	Metachromatic Leukodystrophy	Metabolic Disorder	Metachromatic leukodystrophy: MedlinePlus Genetics	Moved to panel 1 6/24/2024
PLPBP	Epilepsy, early-onset, vitamin B6-dependent	Seizure Disorder	PLPBP Deficiency - GeneReviews® - NCBI Bookshelf (nih.gov)	Moved to panel 1 6/24/2024
SLC2A1	Dystonia 9/GLUT1 deficiency syndrome	Metabolic Disorder	GLUT1 deficiency syndrome: MedlinePlus Genetics	Moved to panel 1 6/24/2024