Set 2

Gene List

Gene	Condition	Type of Condition	More Information	Notes
ARSA	Metachromatic Leukodystrophy	Metabolic Disorder	Metachromatic leukodystrophy: MedlinePlus Genetics	
ASPA	Canavan disease	Metabolic Disorder	Canavan disease: MedlinePlus Genetics	
ATP7A	Menkes Disease	Metabolic Disorder	Menkes syndrome: MedlinePlus Genetics	Males only
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 due to inability to report duplications and deletions <1kb
GALC	Krabbe Disease	Metabolic Disorder	Krabbe disease: MedlinePlus Genetics	
GSS	Glutathione Synthetase Deficiency	Metabolic Disorder	Glutathione synthetase deficiency: MedlinePlus Genetics	
GUSB	Mucopolysaccharidosis type 7	Metabolic Disorder	Mucopolysaccharidosis type VII: MedlinePlus Genetics	
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	
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	

Continued

Gene	Condition	Type of Condition	More Information	Notes
PHGDH	3-phosphoglycerate dehydrogenase deficiency	Metabolic Disorder	Phosphoglycerate dehydrogenase deficiency: MedlinePlus Genetics	
PLPBP	Epilepsy, early-onset, vitamin B6-dependent	Seizure Disorder	PLPBP Deficiency - GeneReviews® - NCBI Bookshelf (nih.gov)	
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	
SLC2A1	Dystonia 9/GLUT1 deficiency syndrome	Metabolic Disorder	GLUT1 deficiency syndrome: 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	
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	