Gene	Pathology
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
AGXT	Primary oxalosis or hyperoxaluria
ARSA	Metachromatic leukodystrophy
ATP7B	Wilson's disease
BTD	Biotinidase deficiency
CBS	Homocystinuria
CFTR	Cystic fibrosis
DHCR7	Smith-Lemli-Opitz syndrome
EMD	Emery-Dreifuss muscular dystrophy
FMR1	Fragile X syndrome
GAA	Glycogen storage disease type 2
GALC	Krabbe disease
GALT	Galactosemia
GBA	Gaucher disease type I-II-III-IIIC-fetal
GJB1	X-linked Charcot-Marie-Tooth neuropathy (CMTX)
GJB2	Recessive non-syndromic sensorineural hearing loss type 1A
GJB6	Recessive non-syndromic sensorineural hearing loss type 1B
GLA	Fabry disease
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HBA1	Alpha thalassemia (HBA1)
HBA2	Alpha thalassemia (HBA2)
HBB	Beta thalassemia/Sickle cell anemia
HEXA	Tay-Sachs disease
MEFV	Familial Mediterranean Fever
MMACHC	Methylmalonic acidemia with homocystinuria
PAH	Phenylketonuria
PMM2	Congenital defect of glycosylation type 1A
SERPINA1	Alpha-1-antitrypsin deficiency
SLC26A2	Achondrogenesis type 1B
SMN1	Spinal Muscular Atrophy (SMA 1)