Gene	Disease
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
AGXT	Hyperoxaluria, primary, type 1
ARSA	Metachromatic leukodystrophy
ATP7B	Wilson disease
BTD	Biotinidase deficiency
CBS	Homocystinuria due to cystathionine beta-synthase
CFTR	Cystic fibrosis
DHCR7	Smith-Lemli-Opitz syndrome
EMD	Emery-Dreifuss muscular dystrophy, type 1 X-linked
FMR1	Fragile X syndrome
GAA	Glycogen storage disease, type 2
GALC	Krabbe disease
GALT	Galactosemia
GBA	Gaucher Disease
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1
GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6
GJB6	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6
GLA	Fabry disease
HADHA	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency
HBA1	Alpha-thalassemia
HBA2	Alpha-thalassemia
HBB	Beta-thalassemia, Sickle cell anemia and other HBB-related hemoglobinopathies
HEXA	Tay-Sachs disease
MEFV	Familial Mediterranean fever
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
PAH	Phenylketonuria
PMM2	Congenital disorder of glycosylation, type 1A
SERPINA1	Alpha-1 antitrypsin deficiency
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)
SMN1	Spinal muscular atrophy