



GENES	PHENOTYPE
ABCB7	X-Linked sideroblastic anemia and ataxia
ACAD8	Isobutyryl-CoA dehydrogenase deficiency
ACADL	Long chain acyl-CoA dehydrogenase deficiency (LCAD)
ACADM	Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
ACADS	Short chain acyl-CoA dehydrogenase deficiency (SCAD)
ACADSB	Short/branched chain acyl-CoA dehydrogenase deficiency
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
ACAT1	b-ketothiolase deficiency
APTX	Ataxia with oculomotor apraxia 1 (AOA1)
ATPAF2 (ATP12)	Neonatal mitochondrial encephalomyopathy, due to ATP synthase deficiency
AUH	3-Methylglutaconic aciduria (MGA) type I
BCS1L	Gracile syndrome; Leigh syndrome; Mitochondrial complex III deficiency, BCS1L related
C10ORF2 (TWINKLE; PEO1)	Progressive external ophthalmoplegia with mtDNA deletions, autosomal dominant 3; PEOA3; Mitochondrial DNA depletion syndrome, hepatocerebral form; Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis; SANDO; Spinocerebellar ataxia, infantile onset
C20ORF7	Mitochondrial complex I deficiency; lethal infantile mitochondrial disease
C8ORF38	Leigh syndrome; LS
CABC1	Coenzyme Q10 deficiency
COQ2	Coenzyme Q10 deficiency
COX10	Leigh syndrome; Mitochondrial complex IV deficiency
COX15	Mitochondrial complex IV deficiency
COX6B1	Mitochondrial complex IV deficiency
COX6C	Leiomyoma uterine, UL
CPT1A	Carnitine palmitoyltransferase IA deficiency
CPT2	Carnitine palmitoyltransferase II deficiency
CYCS	Thrombocytopenia, autosomal dominant 4
DGUOK	Mitochondrial DNA depletion syndrome, hepatocerebral form
DLAT	Pyruvate dehydrogenase E2 deficiency
DLD	Leigh syndrome, Maple syrup urine disease, type III
DNAJC19	3-Methylglutaconic aciduria (MGA) type V
DNM1L (DLP1)	Encephalopathy, lethal due to defective mitochondrial peroxisomal fission
EIF2AK3	Wolcott-Rallison syndrome; epiphyseal dysplasia, multiple, with early onset diabetes mellitus
ETFA	Glutaric aciduria II
ETFB	Glutaric aciduria II
ETFDH	Glutaric aciduria II
ETHE1	Ethylmalonic encephalopathy
GCDH	Glutaric acidemia type I
GCK	GCK-related hyperinsulinism; GCK-related permanent neonatal diabetes mellitus; Maturity-onset diabetes of the young, type II (MODY2)
GFM1(EFG1)	Combined oxidative phosphorylation deficiency 1 (COXPD1)
GPD1L	Brugada syndrome 2
HADH	3-Hydroxyacyl-CoA dehydrogenase deficiency

HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency, mitochondrial trifunctional protein deficiency
HADHB	Mitochondrial trifunctional protein deficiency
HMGCL	HMG-CoA lyase deficiency
HMGCS2	HMG-CoA synthase deficiency
HNF1A (TCF1)	Maturity-onset diabetes of the young, type 3 (MODY3)
HNF1B (TCF2)	Renal cysts and diabetes syndrome
HNF4A	Maturity-onset diabetes of the young, type I (MODY1)
HSD17B10	2-Methyl-3-hydroxybutyryl-CoA dehydrogenase (MHBD) deficiency
L2HGDH	L-2 hydroxyglutaric aciduria
LDHA	Lactate dehydrogenase A deficiency
LDHB	Lactate dehydrogenase B deficiency
LRPPRC	Leigh syndrome, French-Canadian type (LSFC)
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2
MLYCD	Malonic aciduria
MPV17	Mitochondrial DNA depletion syndrome, hepatocerebral form
MRPS16	Combined oxidative phosphorylation deficiency 2 (COXPD2)
MRPS22	Combined oxidative phosphorylation deficiency 5 (COXPD5)
NDUFA1	Mitochondrial complex I deficiency; Leigh syndrome
NDUFA11	Mitochondrial complex I deficiency; encephalopathy, cardiomyopathy and lactic acidosis; Leigh syndrome
NDUFA2	Leigh syndrome; Mitochondrial complex I deficiency
NDUFAF1	Cardioencephalomyopathy and reduced levels and activity of mitochondrial complex I
NDUFAF2 (NDUFA12L)	Mitochondrial complex I deficiency
NDUFAF4 (C6ORF66)	Mitochondrial complex I deficiency
NDUFS1	Leigh syndrome; Mitochondrial complex I deficiency
NDUFS2	Mitochondrial complex I deficiency
NDUFS3	Leigh syndrome; Mitochondrial complex I deficiency
NDUFS4	Mitochondrial complex I deficiency; Leigh syndrome
NDUFS6	Mitochondrial complex I deficiency
NDUFS7	Leigh syndrome; Mitochondrial complex I deficiency
NDUFS8	Leigh syndrome; Mitochondrial complex I deficiency
NDUFV1	Leigh syndrome; Mitochondrial complex I deficiency
NDUFV2	Hypertrophic cardiomyopathy, truncal hypotonia, and encephalopathy; Parkinson disease, susceptibility to; Mitochondrial complex I deficiency
NEUROD1	Maturity-onset diabetes of the young type VI
OPA1	Autosomal dominant optic atrophy, type 1 (OPA1); Optic atrophy, deafness, ophthalmoplegia and myopathy
OPA3	Autosomal dominant optic atrophy, type 3; 3-Methylglutaconic aciduria type III
OTC	Ornithine transcarbamylase deficiency
PC	Pyruvate carboxylase deficiency; Leigh syndrome
PDHA1	Pyruvate dehydrogenase deficiency; Leigh syndrome, X-linked
PDHB	Pyruvate dehydrogenase deficiency
PDHX	Pyruvate dehydrogenase E3-binding protein deficiency
PDSS1	Coenzyme Q10 deficiency
PDSS2	Coenzyme Q10 deficiency
PINK1	Parkinson disease 6, autosomal recessive early-onset; PARK6
POLG	Progressive external ophthalmoplegia with mtDNA deletions, autosomal dominant 1 (PEOA1) or recessive (PEOB); Alpers syndrome; Spinocerebellar ataxia neuropathy, dysarthria, and ophthalmoparesis; SANDO

POLG2	Progressive external ophthalmoplegia with mtDNA deletions, autosomal dominant 4 (adPEO4)
PPARGC1B	Obesity
PUS1	Mitochondrial myopathy and sideroblastic anemia (MLASA)
RMRP (RNase MRP)	Cartilage-hair hypoplasia (CHH); Metaphyseal dysplasia without hypotrichosis; Anauxetic dysplasia; spondylometaphyseal dysplasia, anauxetic type
RRM2B	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy
SCO1	Mitochondrial complex IV deficiency
SCO2	Mitochondrial complex IV deficiency
SDHB	Mitochondrial complex II deficiency; SDHB-related hereditary paraganglioma-pheochromocytoma syndrome
SDHC	SDHC-related hereditary paraganglioma-pheochromocytoma syndrome
SDHD	SDHD-related hereditary paraganglioma-pheochromocytoma syndrome
SLC22A5	Systemic carnitine deficiency
SLC25A13	Citrin deficiency; Citrullinemia type II
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome
SLC25A19	Amish lethal microcephaly
SLC25A20 (CACT)	Carnitine-acylcarnitine translocase deficiency
SLC25A22 (GC1)	Early myoclonic encephalopathy; EME
SLC25A3 (PHC)	Book syndrome
SLC25A4 (ANT1)	Progressive external ophthalmoplegia with mtDNA deletions, autosomal dominant 2 (PEOA2)
SPG7	Spastic paraplegia 7
SUCLA2	Mitochondrial DNA depletion syndrome, myopathic form
SUCLG1	Fatal infantile lactic acidosis
SURF1	Leigh syndrome; Mitochondrial complex IV deficiency
TAZ	3-methylglutaconic aciduria type II; Barth syndrome; Cardioskeletal myopathy with neutropenia and abnormal mitochondria; Endocardial fibroelastosis; Familial isolated noncompaction of left ventricular myocardium; TAZ-related dilated cardiomyopathy
TIMM8A	Deafness-dystonia-optic neuropathy (DDP1) syndrome; Mohr-Tranebjaerg syndrome
TK2	Mitochondrial DNA depletion syndrome, myopathic form
TMEM70	Encephalocardiomyopathy, mitochondrial, neonatal, due to ATP synthase deficiency
TSFM (EFTs)	Combined oxidative phosphorylation deficiency-3 (COXPD3)
TUFM (EFTu)	Combined oxidative phosphorylation deficiency-4 (COXPD4)
TYMP (ECGF1)	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease
UCP3	Type II diabetes; obesity
UQCRB	Mitochondrial complex III deficiency
UQCRCQ	Mitochondrial complex III deficiency
WFS1	Wolfram syndrome 1; diabetes insipidus and mellitus with optic atrophy and deafness