Gene List – nucSEEK® Focus Nuclear Mitochondrial Disorder Panel

By Gene (alphabetically)

Gene	Disease	Category
AARS2	Combined oxidative phosphorylation deficiency 8	tRNA
ABAT	autosomal recessive neurometabolic disorder and mtDNA depletion syndrome (MDS) http://www.ncbi.nlm.nih.gov/pubmed/?term=25738457	Neurotransmitters
ABCB7	Anemia, sideroblastic, with ataxia	ABC Transporters
ACACA	Acetyl-CoA carboxylase deficiency	Lipid Metabolism
ACAD9	Acyl Co-A dehydrogenase 9 deficiency	Lipid Metabolism
ACADL	Long chain acyl CoA dehydrogenase, no disease association in OMIM	Lipid Metabolism
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of	Lipid Metabolism
ACADVL	VLCAD deficiency	Lipid Metabolism
ACAT	Alpha-methylacetoacetic aciduria	Lipid Metabolism
ACO2	Infantile cerebellar-retinal degeneration	TCA Cycle
ADCK3	Coenzyme Q10 deficiency, primary, 4	Other
AFG3L2	Ataxia, spastic, 5, autosomal recessive	Other
AGK	Cataract 38, autosomal recessive	Lipid Metabolism
AIFM1	Combined oxidative phosphorylation deficiency 6	Complex I
ALAS2	Anemia, sideroblastic, X-linked	Iron Metabolism
ALDH1B1	Aldehyde dehydrogenase family 1, B1 - no associated phenotype in OMIM	Other Enzymes
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	DNA/RNA
ATP5A1	Combined oxidative phosphorylation deficiency 22	Complex V
ATP5E	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3	Complex V
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	Complex V
AUH	3-methylglutaconic aciduria, type I	Cell Function
BCS1L	Mitochondrial complex III deficiency, nuclear type 1	Complex III
BOLA3	Multiple mitochondrial dysfunctions syndrome 2	Iron Metabolism
BTD	Biotinidase deficiency	Other Enzymes
C10ORF2	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	DNA/RNA
C12ORF65	Combined oxidative phosphorylation deficiency 7	Protein Metabolism
C20ORF7	Mitochondrial complex I deficiency	Cell Function
C8ORF38	Leigh syndrome due to mitochondrial complex I deficiency	Other
CISD2	Wolfram syndrome 2	Iron Metabolism
CLPB	Autosomal-Recessive Mitochondrial Disorder	Other Enzymes
COA5	Mitochondrial complex IV deficiency	Complex IV
COQ2	Coenzyme Q10 deficiency, primary, 1	Coenzyme Q
COQ6	Coenzyme Q10 deficiency, primary, 6	Coenzyme Q
COQ9	Coenzyme Q10 deficiency, primary, 5	Coenzyme Q
COX10	Leigh syndrome due to mitochondrial COX4 deficiency	Complex IV
COX14	Mitochondrial complex IV deficiency	Complex IV
COX15	Leigh syndrome due to cytochrome c oxidase deficiency	Complex IV

Gene	Disease	Category
COX20	Mitochondrial complex IV deficiency	Complex IV
COX4I2	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	Complex IV
COX6B1	Mitochondrial complex IV deficiency	Complex IV
CPS1	Carbamoylphosphate synthetase I deficiency	Other Enzymes
CPT1A	CPT deficiency, hepatic, type IA	Lipid Metabolism
CPT2	CPT deficiency, hepatic, type II; lethal neonatal	Lipid Metabolism
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	tRNA
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	Nucleotide Metabolism
DLAT	Pyruvate dehydrogenase E2 deficiency	Pyruvate Dehydrogenase
DLD	Dihydrolipoamide dehydrogenase deficiency	Amino Acid Metabolism
DNAJC19	3-methylglutaconic aciduria, type V	Protein Metabolism
DNM1L	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission	Cell Function
EARS2	Combined oxidative phosphorylation deficiency 12	tRNA
ECHS1	Leigh Disease http://www.ncbi.nlm.nih.gov/pubmed/25393721	Lipid Metabolism
ETFA	Glutaric acidemia IIA	Lipid Metabolism
ETFB	Glutaric acidemia IIB	Lipid Metabolism
ETFDH	Glutaric acidemia IIC	Lipid Metabolism
ETHE1	Ethylmalonic encephalopathy	Amino Acid Metabolism
FARS2	Combined oxidative phosphorylation deficiency 14	tRNA
FASTKD2	Mitochondrial complex IV deficiency	Cell Function
FBP1	Fructose-1,6-bisphosphatase deficiency	Carbohydrate Metabolism
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	Other Enzymes
FH	Fumarase deficiency	TCA Cycle
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
G6PC	Glycogen storage disease Ia	Carbohydrate Metabolism
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	Cell Function
GFM1	Combined oxidative phosphorylation deficiency 1	Protein Metabolism
GFM2	Mitochondrial elongation factor G2 - no associated phenotype in OMIM	Protein Metabolism
GTPBP3	hypertrophic cardiomyopathy, lactic acidosis, and encephalopathy http://www.ncbi.nlm.nih.gov/pubmed/25434004	GTPase
GYS1	Glycogen storage disease 0, muscle	Carbohydrate Metabolism
GYS2	Glycogen storage disease 0, liver	Carbohydrate Metabolism
HADHA	LCHAD deficiency	Lipid Metabolism
HADHB	Trifunctional protein deficiency	Lipid Metabolism
HARS2	Perrault syndrome 2	tRNA
HIBCH	Leigh disease	Lipid Metabolism
HLCS	Holocarboxylase synthetase deficiency	Carbohydrate Metabolism
HSPD1	Leukodystrophy, hypomyelinating, 4	Cell Function
IARS2	Peripheral neuropathy, Leigh syndrome	tRNA
ISCU	Myopathy with lactic acidosis, hereditary	Iron Metabolism
IVD	Isovaleric acidemia	Amino Acid Metabolism
KARS	Charcot-Marie-Tooth disease, recessive intermediate, B	tRNA
LARS	Infantile liver failure syndrome 1	tRNA
LARS2	Infantile liver failure syndrome 1	tRNA
LIAS	Pyruvate dehydrogenase lipoic acid synthetase deficiency	Lipid Metabolism
LRPPRC	Leigh syndrome, French-Canadian type	Cell Function

Gene	Disease	Category
MARS2	Spastic ataxia 3, autosomal recessive	tRNA
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	Lipid Metabolism
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	Lipid Metabolism
MFN2	Charcot-Marie-Tooth disease, type 2A2	Cell Function
MGME1	Mitochondrial DNA depletion syndrome 11	DNA/RNA
MMAA	Methylmalonic aciduria, vitamin B12-responsive	Lipid Metabolism
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	Lipid Metabolism
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	Lipid Metabolism
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	Lipid Metabolism
MPC1	Mitochondrial pyruvate carrier deficiency	Cell Function
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	Antioxidant
MRPL44	Combined oxidative phosphorylation deficiency 16	Ribosome Protein
MRPS16	Combined oxidative phosphorylation deficiency 2	Ribosome Protein
MRPS22	Combined oxidative phosphorylation deficiency 5	Ribosome Protein
MTFMT	Combined oxidative phosphorylation deficiency 15	tRNA
MTO1	Combined oxidative phosphorylation deficiency 10	Protein Metabolism
MTPAP	Ataxia, spastic, 4	DNA/RNA
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	Other Enzymes
MUT	Methylmalonic aciduria, mut(0) type	Lipid Metabolism
NAGS	N-acetylglutamate synthase deficiency	Other Enzymes
NDUFA1	Mitochondrial complex I deficiency	Complex I
NDUFA10	Leigh syndrome	Complex I
NDUFA11	Mitochondrial complex I deficiency	Complex I
NDUFA12	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFA9	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFAF1	Mitochondrial complex I deficiency	Complex I
NDUFAF2	Leigh syndrome	Complex I
NDUFAF3	Mitochondrial complex I deficiency	Complex I
NDUFAF4	Mitochondrial complex I deficiency	Complex I
NDUFAF7	NADH dehydrogenase (ubiquinone) complex 1, assembly factor 7 - no associated phenoytype in OMIM	Complex I
NDUFB3	Mitochondrial complex I deficiency	Complex I
NDUFB9	Mitochondrial complex I deficiency	Complex I
NDUFS1	Mitochondrial complex I deficiency	Complex I
NDUFS2	Mitochondrial complex I deficiency	Complex I
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFS4	Leigh syndrome	Complex I
NDUFS6	Complex I, mitochondrial respiratory chain, deficiency of	Complex I
NDUFS7	Leigh syndrome	Complex I
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFV1	Mitochondrial complex I deficiency	Complex I
NDUFV2	Mitochondrial complex I deficiency	Complex I
NFU1	Multiple mitochondrial dysfunctions syndrome 1	Iron Metabolism
NUBPL	Mitochondrial complex I deficiency	Complex I
OPA1	Optic atrophy 1	DNA/RNA

Gene	Disease	Category
OPA3	Optic atrophy 3 with cataract	DNA/RNA
OTC	Ornithine transcarbamylase deficiency	Other Enzymes
PC	Pyruvate carboxylase deficiency	TCA Cycle
PCCA	Propionic acidemia	Other Enzymes
PCCB	Propionic acidemia	Other Enzymes
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	Pyruvate Dehydrogenase
PDHB	Pyruvate dehydrogenase E1-beta deficiency	Pyruvate Dehydrogenase
PDHX	Lacticacidemia due to PDX1 deficiency	Pyruvate Dehydrogenase
PDP1	Pyruvate dehydrogenase phosphatase deficiency	Pyruvate Dehydrogenase
PDSS1	Coenzyme Q10 deficiency, primary, 2	Coenzyme Q
PDSS2	Coenzyme Q10 deficiency, primary, 3	Coenzyme Q
PGAM2	Glycogen storage disease X	Carbohydrate Metabolism
PNPT1	Combined oxidative phosphorylation deficiency 13	DNA/RNA
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 4B (MNGIE type)	DNA/RNA
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4	DNA/RNA
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	tRNA
RARS2	Pontocerebellar hypoplasia, type 6	tRNA
REEP1	Neuronopathy, distal hereditary motor, type VB	Cell Function
RMND1	Combined oxidative phosphorylation deficiency 11	Cell Function
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	DNA/RNA
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	tRNA
SCO1	Hepatic failure, early onset, and neurologic disorder	Complex IV
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Complex IV
SDHAF1	Mitochondrial complex II deficiency	Complex II
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	Cell Function
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	Solute Carrier
SLC22A5	Carnitine deficiency, systemic primary	Solute Carrier
SLC25A13	Citrullinemia, type II, neonatal-onset	Solute Carrier
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Solute Carrier
SLC25A19	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	Solute Carrier
SLC25A20	Carnitine-acylcarnitine translocase deficiency	Solute Carrier
SLC25A3	Mitochondrial phosphate carrier deficiency	Solute Carrier
SLC25A4	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type)	Solute Carrier
SLC37A4	Glycogen storage disease Ib	Solute Carrier
SPG7	Spastic paraplegia 7, autosomal recessive	Cell Function
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	TCA Cycle
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	TCA Cycle
SURF1	Leigh syndrome, due to COX deficiency	Complex IV
TACO1	Mitochondrial complex IV deficiency	Complex IV
TAZ	Barth syndrome	Other
TIMM8A	Deafness, X-linked 1, progressive	Membrane Protein
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type)	DNA/RNA
TMEM126A	Optic atrophy-7	Membrane Protein
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	Complex V
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	Carbohydrate Metabolism

Gene	Disease	Category
TRMU	Liver failure, transient infantile	tRNA
TSFM	Combined oxidative phosphorylation deficiency 3	DNA/RNA
TTC19	Mitochondrial complex III deficiency, nuclear type 2	Other
TUFM	Combined oxidative phosphorylation deficiency 4	DNA/RNA
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	Cell Function
UQCRB	Mitochondrial complex III deficiency, nuclear type 3	Complex III
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4	Complex III
WFS1	Wolfram syndrome, Wolfram-like syndrome, autosomal dominant	Other
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2	tRNA

By Category

Gene	Disease	Category
ABCB7	Anemia, sideroblastic, with ataxia	ABC Transporters
DLD	Dihydrolipoamide dehydrogenase deficiency	Amino Acid Metabolism
ETHE1	Ethylmalonic encephalopathy	Amino Acid Metabolism
IVD	Isovaleric acidemia	Amino Acid Metabolism
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	Antioxidant
FBP1	Fructose-1,6-bisphosphatase deficiency	Carbohydrate Metabolism
G6PC	Glycogen storage disease Ia	Carbohydrate Metabolism
GYS1	Glycogen storage disease 0, muscle	Carbohydrate Metabolism
GYS2	Glycogen storage disease 0, liver	Carbohydrate Metabolism
HLCS	Holocarboxylase synthetase deficiency	Carbohydrate Metabolism
PGAM2	Glycogen storage disease X	Carbohydrate Metabolism
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	Carbohydrate Metabolism
AUH	3-methylglutaconic aciduria, type I	Cell Function
C20ORF7	Mitochondrial complex I deficiency	Cell Function
DNM1L	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission	Cell Function
FASTKD2	Mitochondrial complex IV deficiency	Cell Function
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	Cell Function
HSPD1	Leukodystrophy, hypomyelinating, 4	Cell Function
LRPPRC	Leigh syndrome, French-Canadian type	Cell Function
MFN2	Charcot-Marie-Tooth disease, type 2A2	Cell Function
MPC1	Mitochondrial pyruvate carrier deficiency	Cell Function
REEP1	Neuronopathy, distal hereditary motor, type VB	Cell Function
RMND1	Combined oxidative phosphorylation deficiency 11	Cell Function
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	Cell Function
SPG7	Spastic paraplegia 7, autosomal recessive	Cell Function
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	Cell Function
COQ2	Coenzyme Q10 deficiency, primary, 1	Coenzyme Q
COQ6	Coenzyme Q10 deficiency, primary, 6	Coenzyme Q
COQ9	Coenzyme Q10 deficiency, primary, 5	Coenzyme Q
PDSS1	Coenzyme Q10 deficiency, primary, 2	Coenzyme Q
PDSS2	Coenzyme Q10 deficiency, primary, 3	Coenzyme Q
AIFM1	Combined oxidative phosphorylation deficiency 6	Complex I
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFA1	Mitochondrial complex I deficiency	Complex I
NDUFA10	Leigh syndrome	Complex I
NDUFA11	Mitochondrial complex I deficiency	Complex I
NDUFA12	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFA9	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFAF1	Mitochondrial complex I deficiency	Complex I
NDUFAF2	Leigh syndrome	Complex I
NDUFAF3	Mitochondrial complex I deficiency	Complex I
NDUFAF4	Mitochondrial complex I deficiency	Complex I
NDUFAF7	NADH dehydrogenase (ubiquinone) complex 1, assembly factor 7 - no associated phenoytype in OMIM	Complex I

Gene	Disease	Category
NDUFB3	Mitochondrial complex I deficiency	Complex I
NDUFB9	Mitochondrial complex I deficiency	Complex I
NDUFS1	Mitochondrial complex I deficiency	Complex I
NDUFS2	Mitochondrial complex I deficiency	Complex I
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFS4	Leigh syndrome	Complex I
NDUFS6	Complex I, mitochondrial respiratory chain, deficiency of	Complex I
NDUFS7	Leigh syndrome	Complex I
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency	Complex I
NDUFV1	Mitochondrial complex I deficiency	Complex I
NDUFV2	Mitochondrial complex I deficiency	Complex I
NUBPL	Mitochondrial complex I deficiency	Complex I
SDHAF1	Mitochondrial complex II deficiency	Complex II
BCS1L	Mitochondrial complex III deficiency, nuclear type 1	Complex III
UQCRB	Mitochondrial complex III deficiency, nuclear type 3	Complex III
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4	Complex III
COA5	Mitochondrial complex IV deficiency	Complex IV
COX10	Leigh syndrome due to mitochondrial COX4 deficiency	Complex IV
COX14	Mitochondrial complex IV deficiency	Complex IV
COX15	Leigh syndrome due to cytochrome c oxidase deficiency	Complex IV
COX20	Mitochondrial complex IV deficiency	Complex IV
COX4I2	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	Complex IV
COX6B1	Mitochondrial complex IV deficiency	Complex IV
SCO1	Hepatic failure, early onset, and neurologic disorder	Complex IV
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Complex IV
SURF1	Leigh syndrome, due to COX deficiency	Complex IV
TACO1	Mitochondrial complex IV deficiency	Complex IV
ATP5A1	Combined oxidative phosphorylation deficiency 22	Complex V
ATP5E	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3	Complex V
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	Complex V
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	Complex V
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	DNA/RNA
C10ORF2	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	DNA/RNA
MGME1	Mitochondrial DNA depletion syndrome 11	DNA/RNA
MTPAP	Ataxia, spastic, 4	DNA/RNA
OPA1	Optic atrophy 1	DNA/RNA
OPA3	Optic atrophy 3 with cataract	DNA/RNA
PNPT1	Combined oxidative phosphorylation deficiency 13	DNA/RNA
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 4B (MNGIE type)	DNA/RNA
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4	DNA/RNA
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	DNA/RNA
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type)	DNA/RNA

Gene	Disease	Category
TSFM	Combined oxidative phosphorylation deficiency 3	DNA/RNA
TUFM	Combined oxidative phosphorylation deficiency 4	DNA/RNA
GTPBP3	hypertrophic cardiomyopathy, lactic acidosis, and encephalopathy http://www.ncbi.nlm.nih.gov/pubmed/25434004	GTPase
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BOLA3	Multiple mitochondrial dysfunctions syndrome 2	Iron Metabolism
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ISCU	Myopathy with lactic acidosis, hereditary	Iron Metabolism
NFU1	Multiple mitochondrial dysfunctions syndrome 1	Iron Metabolism
ACACA	Acetyl-CoA carboxylase deficiency	Lipid Metabolism
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ACADL	Long chain acyl CoA dehydrogenase, no disease association in OMIM	Lipid Metabolism
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CPT2	CPT deficiency, hepatic, type II; lethal neonatal	Lipid Metabolism
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ETFA	Glutaric acidemia IIA	Lipid Metabolism
ETFB	Glutaric acidemia IIB	Lipid Metabolism
ETFDH	Glutaric acidemia IIC	Lipid Metabolism
HADHA	LCHAD deficiency	Lipid Metabolism
HADHB	Trifunctional protein deficiency	Lipid Metabolism
HIBCH	Leigh disease	Lipid Metabolism
LIAS	Pyruvate dehydrogenase lipoic acid synthetase deficiency	Lipid Metabolism
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	Lipid Metabolism
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	Lipid Metabolism
MMAA	Methylmalonic aciduria, vitamin B12-responsive	Lipid Metabolism
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	Lipid Metabolism
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	Lipid Metabolism
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	Lipid Metabolism
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ADCK3	Coenzyme Q10 deficiency, primary, 4	Other
AFG3L2	Ataxia, spastic, 5, autosomal recessive	Other
C8ORF38	Leigh syndrome due to mitochondrial complex I deficiency	Other
TAZ	Barth syndrome	Other
TTC19	Mitochondrial complex III deficiency, nuclear type 2	Other
WFS1	Wolfram syndrome, Wolfram-like syndrome, autosomal dominant	Other

Gene	Disease	Category
ALDH1B1	Aldehyde dehydrogenase family 1, B1 - no associated phenotype in OMIM	Other Enzymes
BTD	Biotinidase deficiency	Other Enzymes
CLPB	Autosomal-Recessive Mitochondrial Disorder	Other Enzymes
CPS1	Carbamoylphosphate synthetase I deficiency	Other Enzymes
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	Other Enzymes
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	Other Enzymes
NAGS	N-acetylglutamate synthase deficiency	Other Enzymes
OTC	Ornithine transcarbamylase deficiency	Other Enzymes
PCCA	Propionic acidemia	Other Enzymes
PCCB	Propionic acidemia	Other Enzymes
C12ORF65	Combined oxidative phosphorylation deficiency 7	Protein Metabolism
DNAJC19	3-methylglutaconic aciduria, type V	Protein Metabolism
GFM1	Combined oxidative phosphorylation deficiency 1	Protein Metabolism
GFM2	Mitochondrial elongation factor G2 - no associated phenotype in OMIM	Protein Metabolism
MTO1	Combined oxidative phosphorylation deficiency 10	Protein Metabolism
DLAT	Pyruvate dehydrogenase E2 deficiency	Pyruvate Dehydrogenase
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	Pyruvate Dehydrogenase
PDHB	Pyruvate dehydrogenase E1-beta deficiency	Pyruvate Dehydrogenase
PDHX	Lacticacidemia due to PDX1 deficiency	Pyruvate Dehydrogenase
PDP1	Pyruvate dehydrogenase phosphatase deficiency	Pyruvate Dehydrogenase
MRPL44	Combined oxidative phosphorylation deficiency 16	Ribosome Protein
MRPS16	Combined oxidative phosphorylation deficiency 2	Ribosome Protein
MRPS22	Combined oxidative phosphorylation deficiency 5	Ribosome Protein
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	Solute Carrier
SLC22A5	Carnitine deficiency, systemic primary	Solute Carrier
SLC25A13	Citrullinemia, type II, neonatal-onset	Solute Carrier
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Solute Carrier
SLC25A19	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	Solute Carrier
SLC25A20	Carnitine-acylcarnitine translocase deficiency	Solute Carrier
SLC25A3	Mitochondrial phosphate carrier deficiency	Solute Carrier
SLC25A4	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type)	Solute Carrier
SLC37A4	Glycogen storage disease Ib	Solute Carrier
ACO2	Infantile cerebellar-retinal degeneration	TCA Cycle
FH	Fumarase deficiency	TCA Cycle
PC	Pyruvate carboxylase deficiency	TCA Cycle
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	TCA Cycle
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	TCA Cycle
AARS2	Combined oxidative phosphorylation deficiency 8	tRNA
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	tRNA
EARS2	Combined oxidative phosphorylation deficiency 12	tRNA
FARS2	Combined oxidative phosphorylation deficiency 14	tRNA
HARS2	Perrault syndrome 2	tRNA

Gene	Disease	Category
IARS2	Peripheral neuropathy, Leigh syndrome	tRNA
KARS	Charcot-Marie-Tooth disease, recessive intermediate, B	tRNA
LARS	Infantile liver failure syndrome 1	tRNA
LARS2	Infantile liver failure syndrome 1	tRNA
MARS2	Spastic ataxia 3, autosomal recessive	tRNA
MTFMT	Combined oxidative phosphorylation deficiency 15	tRNA
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	tRNA
RARS2	Pontocerebellar hypoplasia, type 6	tRNA
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	tRNA
TRMU	Liver failure, transient infantile	tRNA
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2	tRNA