Mito 101 Gene List and Associated Disorders

Gene Name	OMIM #	Associated Disease(s) or Phenotype(s)	Inheritance Pattern
AARS2	*612035	Combined Oxidative Phosphorylation Deficiency 8; Infantile Mitochondrial Cardiomyopathy[159]	Autosomal recessive
ABCB7	*300135	Anemia, Sideroblastic and Spinocerebellar Ataxia[1,2]	X-linked
ACAD9	*611103	Acyl-CoA dehydrogenase 9 deficiency; ACAD9 Deficiency; Mitochondrial complex I deficiency due to ACAD9 Deficiency[3]	Autosomal recessive
ADCK3 (CABC1; (COQ8)	*606980	Coenzyme Q10 Deficiency; Spinocerebellar Ataxia, Autosomal Recessive 9[24,25]	Autosomal recessive
AIFM1	*300169	Combined oxidative phosphorylation deficiency 6; COXPD6; Encephalomyopathy, Mitochondrial, X-Linked[4]	X-linked
ALAS2	*301300	Anemia, Hereditary Sideroblastic, X-linked[1,2] Protoporphyria, erythropoietic, X-linked dominant.	X-linked
APTX	*606350	Ataxia with Oculomotor Apraxia 1 (AOA1); Cerebellar ataxia with Muscle coenzyme Q10 deficiency[5,6]	Autosomal recessive
ATP5E	*606153	Mitochondrial complex V (ATP Synthase) deficiency, Nuclear Type 3; MC5DN3[7]	Autosomal recessive
ATPAF2 (ATP12)	*608918	Mitochondrial complex V (ATP Synthase) Deficiency, Nuclear Type 1; MC5DN1[8]	Autosomal recessive
AUH	*600529	3-Methylglutaconic Aciduria, Type 1; MGA, Type 1; 3-Methylglutaconyl-CoA Hydratase Deficiency[9]	Autosomal recessive
BCS1L	*603647	Bjornstad syndrome; GRACILE syndrome; Mitochondrial Complex III Deficiency; Leigh Syndrome[10,11,12]	Autosomal recessive
C10ORF2 (Twinkle, PEO1)	*606075	C10orf2-Related Ataxia Neuropathy Spectrum Disorders; Mitochondrial DNA Depletion Syndrome 7, Hepatocerebral Form; Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions 3[14,15,16,17,18,19,20]	Autosomal dominant AND Autosomal recessive
C12ORF65	*613541	Combined Oxidative Phosphorylation Deficiency 7; COXPD7[21]	Autosomal recessive
C20ORF7	*612360	Mitochondrial Complex 1 Deficiency[22,23]	Autosomal recessive
C8ORF38	*612392	Leigh syndrome due to mitochondrial complex I deficiency[13]	Autosomal recessive
CISD2 (WFS2)	*611507	Wolfram Syndrome 2 (DIDMOAD: diabetes insipidus, diabetes mellitus, optic atrophy, and deafness)[26,27]	Autosomal recessive
COQ2	*609825	Coenzyme Q10 Deficiency; CoQ10 deficiency, Primary[28]	Autosomal recessive
COQ9	*612837	Coenzyme Q10 Deficiency; CoQ10 Deficiency, Primary[29]	Autosomal recessive
COX10	*602125	Mitochondrial Complex IV Deficiency; Cytochrome c Oxidase Deficiency; Leigh Syndrome[30]	Autosomal recessive
COX15	*603646	Mitochondrial Complex IV Deficiency; Cytochrome c Oxidase Deficiency[31]	Autosomal recessive
COX6B1	*124089	Mitochondrial Complex IV Deficiency; Cytochrome c Oxidase Deficiency[32]	Autosomal recessive
DARS2	*610956	Leukoencephalopathy with Brain Stem and Spinal Cord Involvment and Lactate Elevation[33]	Autosomal recessive
DGUOK	*601465	Mitochondrial DNA Depletion Syndrome 3 (hepatocerebral type)[34,35,36]	Autosomal recessive

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DLAT	*608770	Pyruvate dehydrogenase E2 Deficiency; Leigh syndrome; Lactic Acidemia due to defect of E2 lipoyl transacetylase of the pyruvate dehydrogenase complex[37]	Autosomal recessive
DLD	*238331	E3 deficiency (Maple syrup urine disease, type III); Leigh syndrome[38]	Autosomal recessive
DNAJC19	*608977	3-methylglutaconic aciduria, type V ; dilated cardiomyopathy and ataxia[39]	Autosomal recessive
DNM1L	+603850	Lethal Encephalopathy due to Defective Mitochondrial Fission[40]	Autosomal dominant
ETFA	*608053	Multiple acyl-CoA dehydrogenation deficiency; Glutaricaciduria, type IIA[160,161,162]	Autosomal recessive
ETFB	*130410	Multiple acyl-CoA dehydrogenation deficiency; Glutaricaciduria, type IIB[160,161,162]	Autosomal recessive
ETFDH	*231675	Multiple acyl-CoA Dehydrogenation Deficiency; Glutaricaciduria, Type IIC[160,161,162]	Autosomal recessive
ETHE1	*608451	Ethylmalonic encephalopathy[163,164,165]	Autosomal recessive
FASTKD2	*612322	Mitochondrial Complex IV Deficiency[41]	Autosomal recessive
FBP1	*611570	Fructose-1,6-Bisphosphatase Deficiency; Lactic acidosis[166]	Autosomal recessive
FH	*136850	Fumarate Hydratase Deficiency; Fumaric Aciduria; Fumarase Deficiency[42,43] Leiomyomatosis and renal cell cancer; Multiple cutaneous and uterine leiomyomata	Autosomal recessive; Autosomal dominant
FOXRED1	*613622	Leigh syndrome due to mitochondrial complex I deficiency[44]	Autosomal recessive
G6PC	*613742	Glycogen Storage Disease Ia; Lactic acidosis[169]	Autosomal recessive
GFER	*600924	Myopathy, Mitochondrial Progressive, with Congenital Cataract, Hearing Loss and Developmental Delay[45]	Autosomal recessive
GFM1 (EFG1)	*606639	Combined Oxidative Phosphorylation Deficiency 1; COXPD1; Hepatoencephalopathy, Early Fatal Progressive[46,47,48]	Autosomal recessive
GYS2	*138571	Glycogen Storage Disease, Type 0, Liver ; Lactic acidosis[166,167,168]	Autosomal recessive
ISCU	*611911	Myopathy with Deficiency of ISCU; Iron-Sulfur Cluster Deficiency Myopathy; Myopathy with Deficiency of Succinate Dehydrogenase and Aconitase; Myopathy with Exercise Intolerance, Swedish Type[49,50]	Autosomal recessive
LRPPRC	*607544	Complex IV deficiency; Leigh syndrome, French-Canadian Type[51]	Autosomal recessive
MPV17	*137960	Mitochondrial DNA Depletion Syndrome 6 (Hepatocerebral type)[34]	Autosomal recessive
MRPS16	*609204	Combined Oxidative Phosphorylation Deficiency 2; COXPD2; Agenesis of Corpus Callosum with Dysmorphism and Fatal Lactic Acidosis[55]	Autosomal recessive
MRPS22	*605810	Combined Oxidative Phosphorylation Deficiency 5; COXPD5[56]	Autosomal recessive
NDUFA1	*300078	Mitochondrial Complex I Deficiency; NADH-Coenzyme Q Reductase Deficiency[57,58,59]	X-linked
NDUFA10	*603835	Leigh Syndrome due to Mitochondrial Complex I Deficiency[61]	Autosomal recessive

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NDUFA11	*612638	Mitochondrial complex I deficiency presented with encephalocardiomyopathy or fatal infantile lactic acidemia[62]	Autosomal recessive
NDUFA2	*602137	Leigh syndrome due to mitochondrial complex I deficiency[60]	Autosomal recessive
NDUFAF1	*606934	Mitochondrial complex I deficiency and cardioencephalomyopathy[63,64]	Autosomal recessive
NDUFAF2	*609653	Mitochondrial Complex I Deficiency; Leigh Syndrome[65,66,67]	Autosomal recessive
NDUFAF3 (C3ORF60)	*612911	Mitochondrial Complex I Deficiency[68]	Autosomal recessive
NDUFAF4 (C6ORF66)	*611776	Mitochondrial Complex I Deficiency[68]	Autosomal recessive
NDUFS1	*157655	Mitochondrial Complex I Deficiency[69,70,71,72,73,74] Leigh syndrome or Leigh-like syndrome	Autosomal recessive
NDUFS2	*602985	Mitochondrial Complex I Deficiency[70,75]	Autosomal recessive likely[a]
NDUFS3	*603846	Leigh Syndrome; Leigh Syndrome due to Mitochondrial Complex I Deficiency[73,74]	Autosomal recessive likely[a]
NDUFS4	*602694	Mitochondrial complex I deficiency; Leigh Syndrome[76,77,78,79,80]	Autosomal recessive
NDUFS6	*603848	Mitochondrial complex I deficiency and fatal neonatal lactic acidemia[81,82]	Autosomal recessive
NDUFS7	*601825	Leigh Syndrome due to Mitochondrial Complex I Deficiency[83,84,85]	Autosomal recessive likely[a]
NDUFS8	*602141	Leigh Syndrome due to Mitochondrial Complex I Deficiency[70,72,86,87]	Autosomal recessive likely[a]
NDUFV1	*161015	Alexander Disease; Leigh Syndrome; Mitochondrial Complex I Deficiency[70,71,72,73,74,88]	Autosomal recessive
NDUFV2	*600532	Mitochondrial Complex I Deficiency. Possible association with Parkinson Disease[72,73,89,90]	Autosomal recessive with exception[b]
NUBPL	*613621	Mitochondrial complex I deficiency with mitochondrial encephalomyopathy[44]	Autosomal recessive
OPA1	*605290	Optic Atrophy I; Optic Atrophy with or without deafness, ophthalmoplegia, myopathy, ataxia and neuropathy[91,92]	Autosomal dominant; Semi-dominant
OPA3	*606580	3-Methylglutaconic Aciduria, Type 3; Costeff Syndrome; Optic Atrophy Plus Syndrome[93,94]	Autosomal recessive; Optic Atrophy Autosomal dominant
PC	*608786	Pyruvate carboxylase deficiency; Leigh syndrome[97,98]	Autosomal recessive
PDHA1	*300502	Pyruvate Dehydrogenase E1-alpha Deficiency; Leigh Syndrome, X-linked[99]	X-linked
PDHB	*179060	Pyruvate dehydrogenase (E1-beta) deficiency; Lactic acidosis and neurologic dysfunction[100]	Autosomal recessive
PDHX	*608769	Pyruvate Dehydrogenase E-3 Binding Protein (Component X) Deficiency; Lactic Acidemia due to Defect in Lipoyl-Containing Component X of the Pyruvate Dehydrogenase Complex[101]	Autosomal recessive
PDP1	*605993	Pyruvate dehydrogenase phosphatase deficiency; lactic acidosis resulting in acute respiratory distress and early death[102,103]	Autosomal recessive
PDSS1	*607429	Coenzyme Q10 Deficiency; CoQ10 deficiency, Primary[104]	Autosomal recessive

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PDSS2	*610564	Coenzyme Q10 Deficiency; CoQ10 Deficiency, Primary[105]	Autosomal recessive
POLG (POLG1)	*174763	Mitochondrial DNA Depletion Syndrome (types 4A [Alpers type] and 4B [MNGIE type]); Mitochondrial Recessive Ataxia Syndrome (SANDO and SCAE); Progressive External Opthalmoplegia, Autosomal Recessive AND Autosomal Dominant[107,108,109]	Autosomal dominant AND Autosomal recessive
POLG2	*604983	Progressive External Ophthalmoplegia (PEO) with Mitochondrial DNA Deletions, Autosomal Dominant[110,111]	Autosomal dominant
PUS1	*608109	Myopathy, Lactic Acidosis and Sideroblastic Anemia 1[113,114]	Autosomal recessive
RARS2	*611524	Pontocerebellar Hypoplasia Type 6; PCH6[115]	Autosomal recessive
RRM2B	*604712	Mitochondrial DNA Depletion Syndrome (types 8A and 8B); Progressive External Ophthalmoplegia (PEO) with Mitochondrial DNA Deletions, Autosomal Dominant, 5[116,117,118]	Autosomal dominant AND Autosomal recessive
SARS2 (FBXO17)	*612804	Hyperuricemia, Pulmonary Hypertention, Renal Failure, and Alkalosis; HUPRA Syndrome[119]	Autosomal recessive
SCO1	*603644	Cytochrome Oxidase-Deficiency, Hepatic Failure and Encephalopathy[120]	Autosomal recessive
SCO2	*604272	Cytochrome Oxidase Deficiency and Early Onset, Fatal Hypertrophic Cardiomyopathy (HCM)[121,122]	Autosomal recessive
SDHAF1	*612848	Mitochondrial Complex II Deficiency; Succinate Dehydrogenase Complex Assembly Factor 1 Deficiency[124]	Autosomal recessive
SLC25A3 (PHC)	*600370	Mitochondrial Phosphate Carrier Deficiency[125]	Autosomal recessive
SLC25A4 (ANT1)	*103220	Progressive External Ophthalmoplegia (PEO) with Mitochondrial DNA Deletions 3; Cardiomyopathy, Familial Hypertrophic (CMH)[18,20]	Autosomal dominant (PEO); Autosomal recessive (CMH)
SPG7	*602783	Spastic Paraplegia 7; Hereditary Spastic Paraplegia, Paraplegin Type[128]	Autosomal recessive
SUCLA2	*603921	Mitochondrial DNA Depletion Syndrome 5 (encephalomyopathic type with methylmalonic aciduria)[129]	Autosomal recessive
SUCLG1	*611224	Mitochondrial DNA Depletion Syndrome 9 (encephalomyopathic type with methylmalonic aciduria)[130,131,132]	Autosomal recessive
SURF1	*185620	Leigh Syndrome due to Mitochondrial Complex IV Deficiency[133,134,135,136]	Autosomal recessive
TACO1	*612958	Leigh Syndrome due to Mitochondrial Complex IV Deficiency[137]	Autosomal recessive
TAZ	*300394	Barth syndrome; 3-Methylglutaconic Aciduria Type II; TAZ-related dilated cardiomyopathy[138,139]	X-linked
TIMM8A (DDP1)	*300356	Mohr-Tranebjaerg Syndrome (Deafness-Dystonia-Optic Atrophy Syndrome [DDON]); Jensen Syndrome (Opticoacoustic Nerve Atrophy with Dementia)[140]	X-linked
TK2	*188250	Mitochondrial DNA Depletion Syndrome 2 (myopathic type)[151,142,143,144]	Autosomal recessive
TMEM126A	*612988	Optic Atrophy 7; Nonsyndromic autosomal recessive optic atrophy[146]	Autosomal recessive
TMEM70	*612418	Mitochondrial Complex V (ATP Synthase) Deficiency, Nuclear Type 2; Encephalocardiomyopathy, Mitochondrial, Neonatal, Due to ATP Synthase Deficiency[145]	Autosomal recessive
TRMU	*610230	Liver Failure, Acute Infantile[147]	Autosomal recessive

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TSFM	*604723	Combined Oxidative Phosphorylation Deficiency 3[148]	Autosomal recessive
TTC19	*613814	Mitochondrial Complex III Deficiency [149]	Autosomal recessive
TUFM	*602389	Combined Oxidative Phosphorylation Deficiency 4[150]	Autosomal recessive
TYMP (ECGF1, TP)	*131222	Mitochondrial DNA Depletion Syndrome 1 (MNGIE Type)[151,152,153]	Autosomal recessive
UQCRB	*191330	Mitochondrial Complex III Deficiency, UQCRB-Related[155]	Autosomal recessive
UQCRQ	*612080	Mitochondrial Complex III Deficiency, UQCRQ-Related [155]	Autosomal recessive
WFS1	*606201	WFS1-Related Disorders; Wolfram Syndrome (DIDMOAD: diabetes insipidus, diabetes mellitus, optic atrophhy, and deafness); Wolfram Syndrome-Like Disease; DFNA6/14/38 Nonsyndromic Low-Frequency Sensorineural Hearing Loss[156,157]	Autosomal recessive AND Autosomal dominant
YARS2	*610957	Myopathy, Lactic Acidosis and Sideroblastic Anemia 2[158]	Autosomal recessive

[a] While autosomal recessive inheritance is most likely, these case reports should be noted. See references as indicated for each gene for additional information:
NDUFS2: Single NDUFS2 mutation identified in affected individual who also harbored a single NDUFA8 mutation[70]
NDUFS3: Single NDUFS3 missense mutation identified in individual affected with Complex I Deficiency[73]
NDUFS7: Single heterozygous NDUFS7 mutation identified in individual affected with Complex I Deficiency[70]
NDUFS8: 2 affected individuals identified with single NDUFAS8 mutation; one of these individuals also harbored single NDUFS2 mutation[70,72]
[b] Heterozygous missense change p.A29V in NDUFV2 has been associated with Parkinson Disease[90]

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