MITOCHONDRIAL GENE TABLE

Note: This panel can be ordered from the Mitochondrial/Metabolic test requisition form.

Gene Name	OMIM #	Associated Disease(s)/Phenotypes	Inheritance Pattern
AARS2	612035	Combined oxidative phosphorylation deficiency 8; Progressive leukoencephalopathy with ovarian failure	Autosomal recessive
ABCB7	300135	Sideroblastic anemia with spinocerebellar ataxia	X-linked
ACAD9	611103	Mitochondrial complex I deficiency due to ACAD9 deficiency	Autosomal recessive
ACO2	100850	Optic atrophy 8; Infantile cerebellar-retinal degeneration	Autosomal recessive
AFG3L2	604581	Spinocerebellar ataxia-28; Autosomal recessive spastic ataxia-5	Autosomal dominant and Autosomal recessive
AGK	610345	Autosomal recessive cataract-38; Sengers syndrome or cardiomyopathic mitochondrial DNA depletion syndrome-10	Autosomal recessive
AIFM1	300169	Combined oxidative phosphorylation deficiency-6; Cowchock syndrome or X-linked recessive Charcot-Marie-Tooth disease-4	X-linked
ALAS2	301300	X-linked erythropoietic protoporphyria; X-linked sideroblastic anemia	X-linked
APOPT1	616003	Mitochondrial complex IV deficiency	Autosomal recessive
ATP5A1	164360	Mitochondrial complex V deficiency nuclear type 4; Combined oxidative phosphorylation deficiency-22	Autosomal recessive
ATP5E	606153	Mitochondrial complex V (ATP synthase) deficiency nuclear type 3	Autosomal recessive
ATP7B	606882	Wilson disease	Autosomal recessive
ATPAF2	608918	Mitochondrial complex V (ATP synthase) deficiency nuclear type 1	Autosomal recessive
AUH	600529	3-Methylglutaconic aciduria type I	Autosomal recessive
BCS1L	603647	Mitochondrial complex III deficiency nuclear type 1; Bjornstad syndrome; GRACILE syndrome; Leigh syndrome	Autosomal recessive
BOLA3	613183	Multiple mitochondrial dysfunctions syndrome-2	Autosomal recessive
C12orf65	613541	Autosomal recessive spastic paraplegia-55; Combined oxidative phosphorylation deficiency-7	Autosomal recessive
C19orf12	614297	Neurodegeneration with brain iron accumulation-4; Autosomal recessive spastic paraplegia-43	Autosomal recessive
CARS2	612800	Progressive myoclonic epilepsy[1]	Autosomal recessive
CLPB	616254	3-Methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia or 3-methylglutaconic aciduria type VII	Autosomal recessive
COA5	613920	Mitochondrial complex IV deficiency	Autosomal recessive
COA6	614772	Hypertrophic cardiomyopathy and complex IV deficiency[2, 3, 4]	Autosomal recessive
COASY	609855	Neurodegeneration with brain iron accumulation-6	Autosomal recessive
COQ2	609825	Primary coenzyme Q10 deficiency-1	Autosomal recessive
COQ4	612898	Primary coenzyme Q10 deficiency-7	Autosomal recessive
COQ6	614647	Primary coenzyme Q10 deficiency-6	Autosomal recessive
COQ7	601683	Primary coenzyme Q10 deficiency-8	Autosomal recessive
COQ8A	606980	Primary coenzyme Q10 deficiency-4	Autosomal recessive
COQ8B	615567	Nephrotic syndrome type 9	Autosomal recessive
COQ9	612837	Primary coenzyme Q10 deficiency-8	Autosomal recessive
COX10	602125	Mitochondrial complex IV deficiency; Leigh syndrome due to mitochondrial complex IV deficiency	Autosomal recessive
COX14	614478	Mitochondrial complex IV deficiency	Autosomal recessive
COX15	603646	Fatal infantile cardioencephalomyopathy due to cytochrome c oxidase deficiency; Leigh syndrome due to cytochrome c oxidase deficiency	Autosomal recessive
COX20	614698	Mitochondrial complex IV deficiency	Autosomal recessive

MITOCHONDRIAL GENE TABLE

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Gene Name	OMIM #	Associated Disease(s)/Phenotypes	Inheritance Pattern
COX6A1	602072	Autosomal recessive intermediate Charcot-Marie-Tooth disease D	Autosomal recessive
COX6B1	124089	Mitochondrial complex IV deficiency	Autosomal recessive
COX8A	123870	Mitochondrial complex IV deficiency	Autosomal recessive
CYC1	123980	Mitochondrial complex III deficiency nuclear type 6	Autosomal recessive
DARS2	610956	Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation	Autosomal recessive
DGUOK	601465	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	Autosomal recessive
DLAT	608770	Pyruvate dehydrogenase E2 deficiency	Autosomal recessive
DLD	238331	Dihydrolipoamide dehydrogenase deficiency	Autosomal recessive
DNA2	601810	Autosomal dominant progressive external ophthalmoplegia-6; Seckel syndrome-8	Autosomal dominant and Autosomal recessive
DNAJC19	608977	3-Methylglutaconic aciduria type V or Dilated cardiomyopathy with ataxia	Autosomal recessive
DNM1L	603850	Lethal encephalopathy due to defective mitochondrial and peroxisomal fission	Autosomal dominant and Autosomal recessive
EARS2	612799	Combined oxidative phosphorylation deficiency-12	Autosomal recessive
ECHS1	602292	Mitochondrial short-chain enoyl-CoA hydratase-1 deficiency	Autosomal recessive
ELAC2	605367	Combined oxidative phosphorylation deficiency-17	Autosomal recessive
ETFA	608053	Glutaric aciduria II	Autosomal recessive
ETFB	130410	Glutaric aciduria II	Autosomal recessive
ETFDH	231675	Glutaric aciduria II	Autosomal recessive
ETHE1	608451	Ethylmalonic encephalopathy	Autosomal recessive
FARS2	611592	Combined oxidative phosphorylation deficiency-14	Autosomal recessive
FASTKD2	612322	Mitochondrial complex IV deficiency	Autosomal recessive
FBXL4	605654	Encephalomyopathic mitochondrial DNA depletion syndrome-13	Autosomal recessive
FDX1L	614585	Mitochondrial muscle myopathy[5]	Autosomal recessive
FH	136850	Fumarase deficiency; Hereditary leiomyomatosis and renal cell cancer	Autosomal recessive and Autosomal dominant
FLAD1	610595	Muscle defects with respiratory insufficiency	Autosomal recessive
FOXRED1	613622	Leigh syndrome due to mitochondrial complex I deficiency; Mitochondrial complex I deficiency	Autosomal recessive
GARS	600287	Charcot-Marie-Tooth disease type 2D; Distal hereditary motor neuropathy type VA	Autosomal dominant
GCDH	608801	Glutaric acidemia I	Autosomal recessive
GFER	600924	Progressive mitochondrial myopathy with congenital cataract, hearing loss and developmental delay	Autosomal recessive
GFM1	606639	Combined oxidative phosphorylation deficiency 1	Autosomal recessive
GFM2	606544	Leigh syndrome	Autosomal recessive
GLRX5	609588	Pyridoxine-refractory sideroblastic anemia	Autosomal recessive
GTPBP3	608536	Combined oxidative phosphorylation deficiency 23	Autosomal recessive
GYG2	300198	Leigh syndrome	X-linked
HARS2	600783	Perrault syndrome 2	Autosomal recessive
HMGCL	613898	HMG-CoA lyase deficiency	Autosomal recessive
HTRA2	606441	3-Methylglutaconic aciduria type VIII	Autosomal recessive

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Gene Name	OMIM #	Associated Disease(s)/Phenotypes	Inheritance Pattern
IARS2	612801	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	Autosomal recessive
IBA57	615316	Autosomal recessive spastic paraplegia 74; Multiple mitochondrial dysfunctions syndrome	Autosomal recessive
ISCA2	615317	Multiple mitochondrial dysfunctions syndrome 4	Autosomal recessive
ISCU	611911	Hereditary myopathy with lactic acidosis	Autosomal recessive
LAMP2	309060	Danon disease	X-linked
LARS	151350	Infantile liver failure syndrome 1	Autosomal recessive
LARS2	604544	Perrault syndrome 4	Autosomal recessive
LIAS	607031	Pyruvate dehydrogenase lipoic acid synthetase deficiency	Autosomal recessive
LIPT1	610284	Lipoyltransferase 1 deficiency	Autosomal recessive
LRPPRC	607544	Leigh syndrome, French-Canadian type	Autosomal recessive
LYRM4	613311	Combined oxidative phosphorylation deficiency 19	Autosomal recessive
LYRM7	615831	Mitochondrial complex III deficiency, nuclear type 8	Autosomal recessive
MARS2	609728	Combined oxidative phosphorylation deficiency 25; Spastic ataxia 3	Autosomal recessive
MFF	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission 2	Autosomal recessive
MFN2	608507	Charcot-Marie-Tooth disease, type2A2; Hereditary motor and sensory neuropathy VI	Autosomal dominant and Autosomal recessive
MGME1	615076	Mitochondrial DNA depletion syndrome 11	Autosomal recessive
MICU1	605084	Myopathy with extrapyramidal signs	Autosomal recessive
MPC1	614738	Mitochondrial pyruvate carrier deficiency	Autosomal recessive
MPV17	137960	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	Autosomal recessive
MRPL12	602375	Mitochondrial ribosomal protein L12	Autosomal recessive
MRPL3	607118	Combined oxidative phosphorylation deficiency 9	Autosomal recessive
MRPL44	611849	Combined oxidative phosphorylation deficiency 16	Autosomal recessive
MRPS16	609204	Combined oxidative phosphorylation deficiency 2	Autosomal recessive
MRPS22	605810	Combined oxidative phosphorylation deficiency 5	Autosomal recessive
MRPS7	611974	Mitochondrial ribosomal protein S7	Autosomal recessive
MTFMT	611766	Combined oxidative phosphorylation deficiency 15	Autosomal recessive
MTO1	614667	Combined oxidative phosphorylation deficiency 10	Autosomal recessive
MTPAP	613669	Spastic ataxia 4	Autosomal recessive
NARS2	612803	Combined oxidative phosphorylation deficiency 24	Autosomal recessive
NDUFA1	300078	Mitochondrial complex I deficiency	X-linked
NDUFA10	603835	Leigh syndrome	Autosomal recessive
NDUFA11	612638	Mitochondrial complex I deficiency	Autosomal recessive
NDUFA12	614530	Leigh syndrome due to mitochondrial complex I deficiency	Autosomal recessive
NDUFA2	602137	Leigh syndrome due to mitochondrial complex I deficiency	Autosomal recessive
NDUFA4	603833	Cytochrome c oxidase deficiency	Autosomal recessive
NDUFA9	603834	Leigh syndrome due to mitochondrial complex I deficiency	Autosomal recessive
NDUFAF1	606934	Mitochondrial complex I deficiency	Autosomal recessive

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Gene Name	OMIM #	Associated Disease(s)/Phenotypes	Inheritance Pattern
NDUFAF2	609653	Leigh syndrome; Mitochondrial complex I deficiency	Autosomal recessive
NDUFAF3	612911	Mitochondrial complex I deficiency	Autosomal recessive
NDUFAF4	611776	Mitochondrial complex I deficiency	Autosomal recessive
NDUFAF5	612360	Mitochondrial complex I deficiency	Autosomal recessive
NDUFAF6	612392	Leigh syndrome due to mitochondrial complex I deficiency	Autosomal recessive
NDUFAF7	615898	Mitochondrial complex I deficiency;	Autosomal recessive
NDUFB11	300403	Mitochondrial complex I deficiency: Linear skin defects with multiple congenital anomalies 3	X-linked
NDUFB3	603839	Mitochondrial complex I deficiency	Autosomal recessive
NDUFB9	601445	Mitochondrial Complex I deficiency	Autosomal recessive
NDUFS1	157655	Mitochondrial complex I deficiency	Autosomal recessive
NDUFS2	602985	Mitochondrial complex I deficiency	Autosomal recessive
NDUFS3	603846	Mitochondrial complex I deficiency	Autosomal recessive
NDUFS4	602694	Leigh syndrome; Mitochondrial complex I deficiency	Autosomal recessive
NDUFS6	603848	Mitochondrial complex I deficiency	Autosomal recessive
NDUFS7	601825	Leigh syndrome	Autosomal recessive
NDUFS8	602141	Leigh syndrome due to mitochondrial complex I deficiency	Autosomal recessive
NDUFV1	161015	Mitochondrial complex I deficiency	Autosomal recessive
NDUFV2	600532	Mitochondrial complex I deficiency	Autosomal recessive
NFS1	603485	Mitochondrial complex II/III deficiency, infantile	Autosomal recessive
NFU1	608100	Multiple mitochondrial dysfunctions syndrome 1	Autosomal recessive
NR2F1	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome	Autosomal dominant
NUBPL	613621	Mitochondrial complex I deficiency	Autosomal recessive
OPA1	605290	Optic atrophy 1; Optic atrophy plus syndrome	Autosomal dominant
OPA3	606580	Optic atrophy 3 with cataracts; 3-Methylglutaconic aciduria type III	Autosomal dominant and Autosomal recessive
OTC	300461	Ornithine transcarbamylase deficiency	X-linked
PARS2	612036	Alpers syndrome	Autosomal recessive
PC	608786	Pyruvate carboxylase deficiency	Autosomal recessive
PCCA	232000	Propionic acidaemia	Autosomal recessive
PCCB	232050	Propionic acidaemia	Autosomal recessive
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency	Autosomal recessive
PDHX	608769	Lactic acidemia due to PDX1 deficiency	Autosomal recessive
PDP1	605993	Pyruvate dehydrogenase phosphatase deficiency	Autosomal recessive
PDSS1	607429	Primary coenzyme Q10 deficiency-2	Autosomal recessive
PDSS2	610564	Primary coenzyme Q10 deficiency-3	Autosomal recessive
PET100	614770	Mitochondrial complex IV deficiency	Autosomal recessive
PNPT1	610316	Combined oxidative phosphorylation deficiency 13; Autosomal recessive deafness 70	Autosomal recessive

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Gene Name	OMIM #	Associated Disease(s)/Phenotypes	Inheritance Pattern
POLG	174763	Mitochondrial DNA depletion syndrome 4A (Alpers type); Mitochondrial DNA depletion syndrome 4B (MNGIE type); Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE); Progressive external ophthalmoplegia, autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant	Autosomal recessive and Autosomal dominant
POLG2	604983	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 4	Autosomal dominant
PRKAG2	602743	Hypertrophic cardiomyopathy 6; Lethal congenital glycogen storage disease of heart; Wolff-Parkinson-White syndrome	Autosomal dominant
PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia-1	Autosomal recessive
QARS	603727	Progressive microcephaly with seizures and cerebral and cerebellar atrophy	Autosomal recessive
RARS	107820	Hypomyelinating leukodystrophy-9	Autosomal recessive
RARS2	611524	Pontocerebellar hypoplasia type 6	Autosomal recessive
RMND1	614917	Combined oxidative phosphorylation deficiency 11	Autosomal recessive
RNASEH1	604123	Autosomal recessive progressive external ophthalmoplegia with mitochondrial DNA deletions-2	Autosomal recessive
RRM2B	604712	Mitochondrial DNA depletion syndrome 8A; Mitochondrial DNA depletion syndrome 8B; Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions-5	Autosomal recessive and Autosomal dominant
SARS2	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome	Autosomal recessive
SCO1	603644	Cytochrome c oxidase deficiency; Infantile encephalopathy	Autosomal recessive
SCO2	604272	Fatal infantile cardioencephalomyopathy due to cytochrome c oxidase deficiency-1; Myopia-6	Autosomal recessive and Autosomal dominant
SDHA	600857	Mitochondrial complex II deficiency; Leigh syndrome; Hereditary Paraganglioma and Pheochromocytoma Syndrome	Autosomal recessive and Autosomal dominant
SDHAF1	612848	Mitochondrial complex II deficiency	Autosomal recessive
SERAC1	614725	3-Methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	Autosomal recessive
SFXN4	615564	Combined oxidative phosphorylation deficiency-18	Autosomal recessive
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	Autosomal recessive
SLC19A3	606152	Biotin-responsive basal ganglia disease; Thiamine metabolism dysfunction syndrome-2	Autosomal recessive
SLC22A5	603377	Primary/systemic carnitine deficiency	Autosomal recessive
SLC25A26	611037	Combined oxidative phosphorylation deficiency 28	Autosomal recessive
SLC25A3	600370	Mitochondrial phosphate carrier deficiency	Autosomal recessive
SLC25A38	610819	Autosomal recessive pyridoxine-refractory sideroblastic anemia	Autosomal recessive
SLC25A4	103220	Mitochondrial DNA depletion syndrome-12; Autosomal dominant progressive external ophthalmoplegia (adPEO) with mitochondrial DNA (mtDNA) deletions-2	Autosomal recessive and Autosomal dominant
SLC25A46	610826	Hereditary motor and sensory neuropathy type VIB	Autosomal recessive
SPAST	604277	Autosomal dominant spastic paraplegia-4	Autosomal dominant
SPG7	602783	Autosomal recessive spastic paraplegia-7	Autosomal recessive
SUCLA2	603921	Mitochondrial DNA depletion syndrome-5	Autosomal recessive
SUCLG1	611224	Mitochondrial DNA depletion syndrome-9	Autosomal recessive
SURF1	185620	Leigh syndrome, due to mitochondrial complex IV deficiency	Autosomal recessive
TACO1	612958	Mitochondrial complex IV deficiency; Leigh syndrome due to mitochondrial complex IV deficiency	Autosomal recessive
TARS2	612805	Combined oxidative phosphorylation deficiency-21	Autosomal recessive
TAZ	300394	Barth syndrome or 3-methylglutaconic aciduria type II	X-linked

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Gene Name	OMIM #	Associated Disease(s)/Phenotypes	Inheritance Pattern
TFAM	600438	Mitochondrial DNA depletion syndrome 15 (hepatocerebral type)	Autosomal recessive
TIMM8A	300356	Jensen syndrome; Mohr-Tranebjaerg syndrome	X-linked
TK2	188250	Mitochondrial DNA (mtDNA) depletion syndrome-2	Autosomal recessive
TMEM126A	612988	Optic atrophy-7	Autosomal recessive
TMEM126B	615533	Mitochondrial complex I deficiency	Autosomal recessive
TMEM70	612418	Mitochondrial complex V (ATP synthase) deficiency nuclear type 2	Autosomal recessive
TPK1	606370	Thiamine pyrophosphokinase deficiency or thiamine metabolism dysfunction syndrome-5	Autosomal recessive
TRIT1	N/A	Encephalopathy and myoclonic epilepsy associated with a disorder of mitochondrial translation[6]	Autosomal recessive
TRMT10C	615423	Combined oxidative phosphorylation deficiency 30	Autosomal recessive
TRMU	610230	Transient infantile liver failure	Autosomal recessive
TRNT1	612907	Congenital sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	Autosomal recessive
TSFM	604723	Combined oxidative phosphorylation deficiency-3	Autosomal recessive
TTC19	613814	Mitochondrial complex III deficiency nuclear type 2	Autosomal recessive
TUFM	602389	Combined oxidative phosphorylation deficiency-4	Autosomal recessive
TWNK	606075	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA (mtDNA) deletions-3; Mitochondrial DNA depletion syndrome-7; Perrault syndrome-5	Autosomal dominant and Autosomal recessive
TYMP	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	Autosomal recessive
UQCC2	614461	Mitochondrial complex III deficiency nuclear type 7	Autosomal recessive
UQCC3	616097	Mitochondrial complex III deficiency nuclear type 9	Autosomal recessive
UQCRB	191330	Mitochondrial complex III deficiency nuclear type 3	Autosomal recessive
UQCRC2	191329	Mitochondrial complex III deficiency nuclear type 5	Autosomal recessive
UQCRQ	612080	Mitochondrial complex III deficiency nuclear type 4	Autosomal recessive
VARS2	612802	Combined oxidative phosphorylation deficiency-20	Autosomal recessive
WDR45	300526	Neurodegeneration with brain iron accumulation-5	X-linked
WFS1	606201	Wolfram syndrome; Autosomal dominant Wolfram-like syndrome; DFNA6/14/38 nonsyndromic low-frequency sensorineural hearing loss	Autosomal recessive and Autosomal dominant
YARS2	610957	Myopathy, lactic acidosis, and sideroblastic anemia-2	Autosomal recessive

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