MITOCHONDRIAL DISORDERS GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
AARS2	86.4	99%	96%	Combined oxidative phosphorylation deficiency 8, 614096
ACAD9	91.0	100%	99%	ACAD9 deficiency, 611126
ACO2	77.9	90%	83%	Infantile cerebellar-retinal degeneration, 614559
ADCK3	97.8	100%	95%	Coenzyme Q10 deficiency, primary, 4, 612016
AFG3L2	76.4	95%	91%	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487
AGK	106.6	100%	100%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AIFM1	51.8	97%	75%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
ALDH1B1	131.1	100%	100%	No OMIM phenotype Succinic semialdehyde dehydrogenase deficiency
ANO10	102.8	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOPT1	100.9	100%	99%	Mitochondrial complex IV deficiency, 220110
APTX	121.3	96%	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ATAD3A	41.5	42%	38%	No OMIM phenotype Influence on AIDS progression
ATAD3B	46.1	49%	48%	No OMIM phenotype Influence on AIDS progression
ATP5A1	56.4	93%	80%	?Mitochondrial complex (ATP synthase) deficiency, nuclear type 4, 615228
ATP5B	101.3	100%	100%	No OMIM phenotype
ATP5C1	65.2	94%	88%	No OMIM phenotype
ATP5E	159.8	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5G1	16.8	71%	33%	No OMIM phenotype
ATP5G2	61.8	96%	82%	No OMIM phenotype
ATP5G3	83.1	100%	99%	No OMIM phenotype
ATP5I	72.4	100%	100%	No OMIM phenotype
ATP5J	28.7	90%	64%	No OMIM phenotype
ATP5O	75.2	100%	100%	No OMIM phenotype
ATPAF2	70.9	100%	98%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273

BCS1L	140.9	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358
BOLA1	93.0	100%	100%	No OMIM phenotype
BOLA2	1.1	%	%	No OMIM phenotype
BOLA3	60.2	100%	99%	Multiple mitochondrial dysfunctions syndrome 2, 614299 Hyperglycinaemia, non-ketotic (Baker (2014) Brain 137,366)
C10orf2	145.3	100%	100%	Progressive external ophthalmoplegia, autosomal dominant, 3, 609286 Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245
C12orf65	174.3	100%	100%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	72.8	100%	95%	Neurodegeneration with brain iron accumulation 4, 614298
CARS2	72.6	100%	97%	No OMIM phenotype
CHCHD10	24.0	69%	29%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048
CHKB	91.1	93%	91%	Muscular dystrophy, congenital, megaconial type, 602541
CLPP	90.1	97%	91%	Perrault syndrome 3, 614129
COA1	142.0	100%	100%	No OMIM phenotype
COA5	84.9	99%	96%	Mitochondrial complex IV deficiency, 220110
COA6	76.3	100%	100%	?{Fatal infantile cardiomyopathy, association with}, 604377
COQ2	75.5	99%	96%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	84.4	98%	90%	Coenzyme Q10 deficiency, primary, 607426
COQ6	103.9	99%	94%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ9	86.6	91%	83%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	130.3	100%	97%	Encephalopathy, progressive mitochondrial, with proximal renal tubulopathy due to cytochrome c oxidase deficiency
COX14	129.7	100%	100%	Mitochondrial complex IV deficiency, 220110
COX15	69.4	100%	92%	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119
COX20	59.4	88%	88%	Mitochondrial complex IV deficiency, 220110
COX4I1	53.8	100%	91%	No OMIM phenotype
COX4I2	56.1	99%	89%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, calvarial hyperostosis, 612714

COX5A	70.9	82%	65%	No OMIM phenotype
COX5B	111.5	100%	100%	No OMIM phenotype
COX6A1	99.9	74%	74%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6B1	74.3	100%	100%	Cytochrome c oxidase deficiency, 220110
COX6C	171.1	100%	100%	No OMIM phenotype
COX7A1	93.0	100%	94%	No OMIM phenotype
COX7A2	47.3	83%	76%	No OMIM phenotype
COX7B	32.8	76%	67%	Aplasia cutis congenita, reticulolinear, with microcephaly, facial dysmorphism and other congenital anomalies, 300887
COX7B2	143.6	100%	100%	?{Nasopharyngeal carcinoma, susceptibility to}, 607107
COX7C	25.9	99%	66%	No OMIM phenotype
CYC1	81.4	95%	79%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	49.5	100%	92%	Thrombocytopenia 4, 612004
DARS2	116.2	100%	100%	Leukoencephalopathy (brain stem,spinal cord involvement) and lactate elevation, 611105
DGUOK	98.4	100%	100%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	103.3	100%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	107.0	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	143.7	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	79.6	100%	100%	No OMIM phenotype ?Familial Alzheimer disease
DNA2	115.7	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, 615156
DNAJC19	63.2	74%	74%	3-methylglutaconic aciduria, type V, 610198
DNM1L	102.1	100%	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
EARS2	70.5	93%	91%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	64.0	98%	92%	No OMIM phenotype Leigh disease (Peters (2014) Brain 137, 2903)
ECSIT	95.8	100%	96%	No OMIM phenotype
ELAC2	85.9	100%	100%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 Combined oxidative phosphorylation deficiency 17, 615440
ETHE1	61.0	100%	96%	Ethylmalonic encephalopathy, 602473
FARS2	97.6	98%	94%	Combined oxidative phosphorylation deficiency 14, 614946
FASTKD2	132.3	100%	100%	Mitochondrial complex IV deficiency, 220110
FBXL4	143.6	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX1L	89.9	100%	93%	No OMIM phenotype ?Mitochondrial myopathy with lactic acidosis, association with, 255125

FH	85.7	98%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	95.2	100%	97%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FXN	85.8	92%	87%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GATM	88.8	100%	94%	Cerebral creatine deficiency syndrome 3, 612718
GFER	63.9	99%	94%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	121.8	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GLRX5	29.9	72%	46%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	111.8	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
HARS2	136.3	100%	100%	Perrault syndrome 2, 614926
HCCS	63.9	100%	96%	Microphthalmia, syndromic 7, 309801
HIBCH	68.2	100%	99%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	141.8	100%	100%	Holocarboxylase synthetase deficiency, 253270
HSPD1	14.8	61%	36%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
IARS2	125.7	100%	100%	No OMIM phenotype
IBA57	90.0	100%	94%	?Multiple mitochondrial dysfunctions syndrome 3, 615330
ISCU	93.2	100%	99%	Myopathy with lactic acidosis, hereditary, 255125
KARS	117.9	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
LACTB	104.5	100%	96%	No OMIM phenotype
LARS2	116.2	100%	99%	Perrault syndrome 4, 615300
LIAS	99.9	100%	100%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
LIPT1	200.0	100%	100%	No OMIM phenotype
LRPPRC	100.6	98%	96%	Leigh syndrome, French-Canadian type, 220111
LYRM4	99.6	100%	99%	?Combined oxidative phosphorylation deficiency 19, 615595
MARS2	156.2	100%	100%	Spastic Ataxia 13, autosomal recessive, 611390
MFF	82.1	100%	96%	No OMIM phenotype ?Mitochondrial encephalopathy, 614388
MFN2	103.5	100%	97%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VI, 601152
MGME1	159.2	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MPC1	89.7	100%	100%	Mitochondrial pyruvate carrier deficiency, 614741

MPV17	114.3	100%	100%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 -3
MRPL12	80.8	95%	85%	No OMIM phenotype
MRPL3	74.9	96%	92%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	99.1	100%	100%	No OMIM phenotype
MRPL44	116.8	100%	100%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS16	141.6	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	114.5	95%	90%	No OMIM phenotype
MRPS22	90.7	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719
MTFMT	100.3	100%	100%	Combined oxidative phosphorylation deficiency 15, 614947
MTO1	133.2	100%	98%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	119.0	91%	91%	Ataxia, spastic, 4, 613672
NARS2	117.9	100%	96%	No OMIM phenotype
NDUFA1	116.6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA10	88.8	100%	97%	Leigh syndrome, 256000
NDUFA11	77.0	98%	71%	Mitochondrial complex I deficiency, 252010
NDUFA12	96.2	100%	100%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFA12	96.2	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA13	82.9	100%	100%	{Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	165.3	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA3	92.1	100%	100%	No OMIM phenotype
NDUFA4	51.5	75%	58%	No OMIM phenotype
NDUFA5	36.8	52%	47%	No OMIM phenotype
NDUFA6	203.3	100%	100%	No OMIM phenotype
NDUFA7	79.2	99%	87%	No OMIM phenotype
NDUFA8	86.8	100%	100%	No OMIM phenotype
NDUFA9	99.3	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 -3
NDUFAB1	68.0	100%	100%	No OMIM phenotype
NDUFAF1	116.9	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	55.3	100%	98%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	131.2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	81.2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF5	129.6	100%	100%	Mitochondrial complex 1 deficiency, 252010
NDUFAF6	101.9	100%	97%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFB1	66.8	99%	93%	No OMIM phenotype

NDUFB10	129.1	100%	100%	No OMIM phenotype
NDUFB11	44.9	98%	87%	No OMIM phenotype
NDUFB3	2.3	%	%	Mitochondrial complex I deficiency, 252010
NDUFB4	45.5	88%	68%	No OMIM phenotype
NDUFB7	51.4	99%	69%	No OMIM phenotype
NDUFB8	91.5	100%	100%	No OMIM phenotype
NDUFB9	106.8	100%	100%	?Mitochondrial complex I deficiency, 252010
NDUFC1	78.7	100%	100%	No OMIM phenotype
NDUFC2	67.2	97%	79%	No OMIM phenotype
NDUFS1	79.9	100%	98%	Mitochondrial complex I deficiency, 252010
NDUFS2	123.1	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS3	153.9	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	127.6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS5	161.4	100%	100%	No OMIM phenotype
NDUFS6	118.7	90%	77%	Complex I, mitochondrial respiratory chain, deficiency of, 252010
NDUFS7	100.0	100%	100%	Leigh syndrome, 256000
NDUFS8	107.7	100%	96%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	63.4	100%	92%	Mitochondrial complex I deficiency, 252010
NDUFV2	124.6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFV3	125.3	96%	96%	No OMIM phenotype
NFS1	74.1	100%	94%	No OMIM phenotype Mitochondrial complex II/III deficiency, infantile (Farhan (2014) Mol Gen Gen Med 2, 73)
NFU1	98.5	100%	100%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NUBPL	86.1	100%	100%	Mitochondrial complex I deficiency, 252010
OGDH	114.1	100%	100%	Alpha-ketoglutarate dehydrogenase deficiency, 203740 (1)
OPA1	131.0	99%	98%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
OPA3	101.5	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OXA1L	144.8	100%	98%	No OMIM phenotype
PANK2	143.3	100%	100%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PARS2	153.5	100%	98%	No OMIM phenotype

PDHA1	65.5	98%	93%	Pyruvate dehydrogenase E1-alpha deficiency, 312170 Leigh syndrome, X-linked, 308930
PDHB	98.7	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDK1	96.6	100%	100%	No OMIM phenotype
PDK2	77.2	100%	100%	No OMIM phenotype
PDK3	58.9	100%	98%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	113.2	100%	100%	No OMIM phenotype
PDP1	156.4	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	94.4	90%	86%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	90.7	100%	99%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	70.5	100%	99%	Mitochondrial complex IV deficiency, 220110
PNPT1	103.6	100%	100%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POLG	90.5	98%	92%	Progressive external ophthalmoplegia, autosomal recessive, 258450 Progressive external ophthalmoplegia, autosomal dominant, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpe
POLG2	129.6	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, 610131
PUS1	67.8	100%	98%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PYCR1	88.5	100%	96%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RARS2	83.0	100%	98%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	86.7	95%	92%	Combined oxidative phosphorylation deficiency 11, 614922
RRM2B	113.1	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic renal tubulopathy),612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, 613077 Mitochondrial DNA depletion syndrome 8B
SARS2	66.7	97%	92%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	94.2	96%	95%	Hepatic failure, early onset, and neurologic disorder
SCO2	85.4	100%	100%	Cardioencephalomyopathy, fatal infantile, 604377 Myopia 6, 608908
SDHA	9.1	30%	16%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SDHAF1	91.8	98%	93%	Mitochondrial complex II deficiency, 252011

SDHB	85.5	100%	100%	Paragangliomas 4, 115310 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764
SERAC1	89.8	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, Leigh-like syndrome, 614739
SLC19A2	94.7	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	110.7	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	77.6	88%	82%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	111.6	100%	100%	Hypomyelination, global cerebral, 612949
SLC25A13	99.0	100%	99%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	71.1	100%	97%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A22	75.4	100%	92%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A3	91.8	100%	100%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A4	117.8	100%	98%	Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283 Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418
SPG7	83.9	96%	86%	Spastic paraplegia 7, autosomal recessive, 607259
SUCLA2	81.6	94%	91%	Mitochondrial DNA depletion syndrome 5, 612073
SUCLG1	94.7	95%	91%	Mitochondrial DNA depletion syndrome 9, 245400
SUCLG2	76.2	93%	92%	No OMIM phenotype
SURF1	91.8	88%	88%	Leigh syndrome, due to COX deficiency, 256000
TACO1	90.6	91%	88%	?Mitochondrial complex IV deficiency, 220110
TARS2	101.9	100%	99%	?Combined oxidative phosphorylation deficiency 21, 615918
TAZ	44.9	100%	97%	Barth syndrome, 302060
TIMM44	110.5	100%	100%	No OMIM phenotype
TIMM8A	44.6	100%	99%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TK2	97.0	100%	100%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM126A	85.8	100%	99%	Optic atrophy-7, 612989
TMEM70	176.2	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TPK1	82.4	100%	100%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRIT1	112.5	100%	97%	No OMIM phenotype

TRMU	75.4	100%	99%	{Deafness, mitochondrial, modifier of}, 580000 Liver failure, transient infantile, 613070
TSFM	104.4	100%	98%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	72.4	87%	77%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	100.7	99%	94%	Combined oxidative phosphorylation deficiency 4, 610678
TYMP	89.1	99%	91%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC3	132.0	100%	100%	?Mitochondrial complex III deficiency, nuclear type, 616111
UQCRB	83.4	100%	100%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	91.5	95%	94%	No OMIM phenotype
UQCRC2	88.5	97%	94%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	2.1	24%	13%	No OMIM phenotype
UQCRH	58.3	88%	88%	No OMIM phenotype
UQCRQ	63.3	100%	99%	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	20.5	79%	43%	Combined oxidative phosphorylation deficiency 20, 615917
YARS2	106.9	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors