MITOCHONDRIAL DISORDERS GENE PANEL DGD20062014

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	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
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	100%	100%	No OMIM phenotype
BOLA2	1,1	0%	0%	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
CHKB	91,1	93%	91%	Muscular dystrophy, congenital, megaconial type, 602541
CLPP	90,1	97%	91%	Perrault syndrome 3, 614129
COA1	142	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, and 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	100%	94%	No OMIM phenotype
COX7A2	47,3	83%	76%	No OMIM phenotype

COX7B	32,8	76%	67%	Aplasia cutis congenita, reticulolinear, with mmicrocephaly, 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 with brain stem and 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	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, autosomal dominant, 6, 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
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	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	89,4	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	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	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	91%	91%	Ataxia, spastic, 4, 613672
NDUFA1	116,6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA10	88,8	100%	97%	Leigh syndrome, 256000
NDUFA11	77	98%	71%	Mitochondrial complex I deficiency, 252010
NDUFA12	96,2	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA12	96,2	100%	100%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 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	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	0%	0%	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	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
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	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	157,7	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
PET112	86,9	100%	100%	No OMIM phenotype
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 (Alpers type), 203700 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459
POLG2	129,6	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 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	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 type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5, 613077 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
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, due to cytochrome c oxidase deficiency 1, 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, and 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	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 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	94,7	95%	91%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 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	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
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
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 October 2013
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding region that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding region that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors