MITOCHONDRIAL DISORDERS GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AARS2	141.5	99%	99%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABAT	103.8	100%	99%	GABA-transaminase deficiency, 613163
ACAD9	157.1	99%	97%	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACO2	141.2	97%	94%	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ADCK3	161	99%	99%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCK4	104.6	100%	99%	Nephrotic syndrome, type 9, 615573
AFG3L2	124	94%	87%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGK	152.2	99%	97%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AIFM1	126	100%	99%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
ALDH1B1	247.9	100%	100%	No OMIM phenotype Bladder cancer (Nickerson (2014) Clin Cancer Res 20,4935)
ANO10	136.7	99%	98%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOA1BP	88.2	99%	96%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
APOPT1	90.9	87%	86%	Mitochondrial complex IV deficiency, 220110
APTX	127.2	94%	92%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ATAD3A	97.9	89%	85%	Harel-Yoon syndrome, 617183
ATAD3B	92.8	86%	79%	No OMIM phenotype
ATP5A1	84.5	96%	88%	?Combined oxidative phosphorylation deficiency 22, 616045 ?Mitochondrial complex (ATP synthase) deficiency, nuclear type 4, 615228
ATP5B	145.3	100%	99%	No OMIM phenotype
ATP5C1	101.2	97%	92%	No OMIM phenotype
ATP5D	69.5	96%	85%	No OMIM phenotype
ATP5E	199.3	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053

ATP5F1	97.8	98%	91%	No OMIM phenotype
ATP5G1	114.6	100%	99%	No OMIM phenotype
ATP5G2	76.1	100%	95%	No OMIM phenotype
ATP5G3	134	100%	100%	No OMIM phenotype
ATP5H	93.2	94%	78%	No OMIM phenotype
ATP5I	99.9	100%	99%	No OMIM phenotype
ATP5J	73.9	99%	94%	No OMIM phenotype
ATP5J2	134	100%	100%	No OMIM phenotype
ATP5L	137.8	100%	100%	No OMIM phenotype
ATP5L2	191	100%	100%	No OMIM phenotype
ATP5O	124	99%	97%	No OMIM phenotype
ATP5S	126.8	100%	99%	No OMIM phenotype
ATPAF1	100.7	84%	71%	No OMIM phenotype
ATPAF2	115.8	100%	99%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATPIF1	211.5	100%	100%	No OMIM phenotype
BCS1L	199	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BOLA1	124.6	100%	100%	No OMIM phenotype
BOLA2	115.5	100%	99%	No OMIM phenotype ?Autism and developmental delay (Nuttle (2016) Nature 536, 205)
BOLA3	63.6	92%	83%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
C10orf2	192.9	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
C11orf83	101.4	100%	98%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
C12orf65	94.5	98%	93%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	110.3	100%	99%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C19orf70	74	100%	98%	No OMIM phenotype Mitochondrial encephalopathy with liver disease, early-onset fatal (Guarani (2016) Elife 5, e17163)

				Mitochondrial hepato-encephalopathy (Zeharia (2016) Eur J Hum Genet 24,1778)
CARS2	132.6	100%	99%	Combined oxidative phosphorylation deficiency 27, 616672
CEP89	162	99%	98%	No OMIM phenotype Complex IV deficiency,isolated (van Bon (2013) Hum Mol Genet 22,3138) ?Intellectual disability (Vulto-van Silfhout (2013) Hum Mutat 34,1679)
CHCHD10	27.8	65%	42%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHKB	102.6	99%	97%	Muscular dystrophy, congenital, megaconial type, 602541
CLPB	153.8	96%	96%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	140.5	99%	97%	Perrault syndrome 3, 614129
COA1	98.1	100%	99%	No OMIM phenotype
COA3	176.6	100%	100%	No OMIM phenotype Neuropathy,exercise intolerance,obesity and short stature (Ostergaard (2015) J Med Genet 52,203
COA5	64.8	86%	85%	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	76.8	98%	92%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COASY	173	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COQ2	92.8	96%	93%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	103.5	87%	84%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	155.4	99%	97%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	183.6	99%	99%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ9	94.2	99%	96%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	250.6	100%	99%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	144.9	100%	100%	?Mitochondrial complex IV deficiency, 220110
COX15	103.8	100%	98%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	60.1	95%	81%	Mitochondrial complex IV deficiency, 220110
COX4I1	146.7	100%	100%	No OMIM phenotype ?Schizophrenia (Fromer (2014) Nature 506,179)
COX4I2	123.1	100%	99%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	46.3	90%	66%	No OMIM phenotype
COX5B	166.6	100%	100%	No OMIM phenotype

COX6A1	208.3	99%	99%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	52.8	99%	96%	No OMIM phenotype
COX6B1	186.7	100%	100%	Mitochondrial complex IV deficiency, 220110
COX6B2	61.5	100%	99%	No OMIM phenotype
COX6C	150.3	100%	98%	No OMIM phenotype
COX7A1	118.6	100%	99%	No OMIM phenotype
COX7A2	104.5	99%	97%	No OMIM phenotype {insulin secretion,association with} (Olsson (2011) Eur J Endocrinol 164,765)
COX7B	53.1	83%	56%	Linear skin defects with multiple congenital anomalies, 300887
COX7B2	314.5	100%	100%	No OMIM phenotype
COX7C	66.6	99%	97%	No OMIM phenotype
COX8A	136.5	100%	100%	?Mitochondrial complex IV deficiency, 220110
COX8C	178.8	99%	95%	No OMIM phenotype ?Tethered spinal cord syndrome (Zhao (2016) Neural Regen Res 11, 1333)
CYC1	189.9	96%	89%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	89.9	99%	97%	Thrombocytopenia 4, 612004
DARS2	155.1	100%	99%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DDHD1	168.6	97%	95%	Spastic paraplegia 28, autosomal recessive, 609340
DES	145.8	99%	99%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DGUOK	141.9	99%	99%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	156.7	99%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	104.8	99%	98%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	157	99%	98%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	104.4	94%	89%	No OMIM phenotype ?Diaphragmatic hernia,congenital (Yu (2015) Hum Mol Genet 24,4764)
DNA2	161.2	99%	99%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAJC19	115.5	98%	94%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	143.2	100%	99%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192

DNM1L	143.6	100%	98%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
EARS2	110.3	99%	97%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	123.3	100%	99%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	152.6	99%	98%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
ELAC2	133	100%	99%	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ETHE1	92	99%	97%	Ethylmalonic encephalopathy, 602473
FARS2	234.6	100%	100%	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FASTKD2	152.6	99%	98%	?Mitochondrial complex IV deficiency, 220110
FBXL4	231	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX1L	121.3	99%	97%	No OMIM phenotype Mitochondrial muscle myopathy (Spiegel (2014) Eur J Hum Genet 22,902)
FH	183.5	93%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	144.3	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FXN	84.8	86%	76%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GARS	154.1	99%	98%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GATC	128.7	100%	100%	No OMIM phenotype
GATM	173	100%	99%	Cerebral creatine deficiency syndrome 3, 612718
GFER	91.5	99%	92%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	125.6	99%	97%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	154.5	99%	97%	No OMIM phenotype Leigh syndrome with arthrogryposis multiplex congenita (Fukumura (2015) J Hum Genet 60,509) Wolcott-Rallison syndrome (Dixon-Salazar (2012) Sci Transl Med 4,138ra78) {Atorvastatin sensitivity} (Callegari (2012) PLoS Genet 8,e1002755)
GLRX5	105.8	91%	84%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	84.5	94%	85%	Hyperinsulinism-hyperammonemia syndrome, 606762

GTPBP3	139.5	100%	99%	Combined oxidative phosphorylation deficiency 23, 616198
HARS2	197.5	100%	99%	?Perrault syndrome 2, 614926
HCCS	115.6	99%	98%	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	87.8	97%	86%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	182.6	100%	100%	Holocarboxylase synthetase deficiency, 253270
HSD17B10	119.7	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSPD1	105.4	98%	93%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HTRA2	131.8	100%	99%	3-methlyglutaconic aciduria, type VIII,617248 {Parkinson disease 13}, 610297
IARS2	167	100%	99%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	125.4	94%	90%	?Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
ISCA2	91.2	98%	93%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	143.3	100%	99%	Myopathy with lactic acidosis, hereditary, 255125
KARS	141.9	100%	99%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
LACTB	139.3	98%	89%	No OMIM phenotype
LARS2	157.5	100%	100%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LIAS	175	100%	99%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT1	269	100%	99%	Lipoyltransferase 1 deficiency, 616299
LONP1	164.9	98%	97%	CODAS syndrome, 600373
LRPPRC	157.1	99%	97%	Leigh syndrome, French-Canadian type, 220111
LYRM4	75.4	65%	59%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	65.3	92%	78%	Mitochondrial complex III deficiency, nuclear type 8, 615838
MARS2	179.7	100%	100%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MCUR1	66.9	78%	70%	No OMIM phenotype
MDH2	121.1	98%	97%	Epileptic encephalopathy, early infantile, 51, 617339
MFF	119.7	93%	90%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	157.5	100%	100%	Charcot-Marie-Tooth disease, type 2A2A, 609260

				Charcot-Marie-Tooth disease, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MGME1	178.2	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MICU1	134.3	97%	93%	Myopathy with extrapyramidal signs, 615673
MIEF2	132.9	100%	99%	No OMIM phenotype
MPC1	160.7	100%	99%	Mitochondrial pyruvate carrier deficiency,614741
MPV17	112.5	100%	99%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRP63	150.1	100%	98%	No OMIM phenotype
MRPL12	114.5	99%	96%	No OMIM phenotype Growth retardation and neurological deterioration (Serre (2013) Biochim Biophys Acta 1832)
MRPL3	74.4	92%	82%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	116.9	99%	96%	No OMIM phenotype
MRPL44	140.3	100%	98%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS16	159.3	100%	99%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	192.5	100%	99%	No OMIM phenotype
MRPS22	167.4	97%	93%	Combined oxidative phosphorylation deficiency 5, 611719
MRPS7	191	100%	100%	No OMIM phenotype Sensorineural deafness,progressive hepatic and renal failure and lactic acidaemia (Menezes (2015) Hum Mol Genet 24,2297)
MRRF	216.1	100%	99%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
MTFMT	148.9	99%	96%	Combined oxidative phosphorylation deficiency 15, 614947
MTO1	185.3	90%	88%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	133.3	98%	94%	Ataxia, spastic, 4, 613672
NARS2	153	97%	97%	Combined oxidative phosphorylation deficiency 24, 616239
NDUFA1	229	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA10	166.7	98%	96%	?Leigh syndrome, 256000
NDUFA11	95.2	99%	95%	Mitochondrial complex I deficiency, 252010
NDUFA12	175	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA13	146.3	95%	95%	{Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	146.8	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA3	146.8	93%	87%	No OMIM phenotype
NDUFA4	96	100%	97%	No OMIM phenotype Cytochrome c oxidase deficiency (Pitceathly (2013) Cell Rep 3,1795)

				?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA5	89.1	95%	79%	No OMIM phenotype
NDUFA6	238.9	100%	100%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA7	100.9	100%	99%	No OMIM phenotype
NDUFA8	146.4	100%	99%	No OMIM phenotype Complex I deficiency (Bugiani (2004) Biochim Biophys Acta 1659,136)
NDUFA9	154	99%	97%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAB1	130.2	100%	99%	No OMIM phenotype
NDUFAF1	128.7	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	48.3	87%	76%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	130.4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	125.1	99%	97%	Mitochondrial complex I deficiency, 252010
NDUFAF5	108.8	99%	98%	Mitochondrial complex 1 deficiency, 252010
NDUFAF6	106.3	99%	98%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF7	122.6	100%	99%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFB1	41.1	77%	59%	No OMIM phenotype ?Complex I deficiency (Calvo (2012) Nat Genet 42,851)
NDUFB10	141.9	99%	96%	No OMIM phenotype
NDUFB11	108.3	95%	87%	Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB2	102.9	100%	99%	No OMIM phenotype
NDUFB3	29.3	96%	76%	Mitochondrial complex I deficiency, 252010
NDUFB4	128.2	84%	82%	No OMIM phenotype
NDUFB5	119	100%	100%	No OMIM phenotype
NDUFB6	48.4	99%	96%	No OMIM phenotype
NDUFB7	66.4	100%	98%	No OMIM phenotype
NDUFB8	119.4	100%	99%	No OMIM phenotype Psychomotor retardation, Leigh syndrome, leukodystrophy and complex I deficiency (Pronicka (2016) J Transl Med 14,174)
NDUFB9	123.2	99%	97%	?Mitochondrial complex I deficiency, 252010
NDUFC1	91	100%	99%	No OMIM phenotype
NDUFC2	48.4	97%	88%	No OMIM phenotype

				{Insulin secrection,association with} (Olsson (2011) Eur J Endocrinol 164,765)
NDUFS1	165.9	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS2	121.6	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS3	149.4	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	200.6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS5	211.5	100%	100%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFS6	146.2	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS7	141.6	100%	99%	Leigh syndrome, 256000
NDUFS8	146.8	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	158	99%	97%	Mitochondrial complex I deficiency, 252010
NDUFV2	88.7	90%	73%	Mitochondrial complex I deficiency, 252010
NDUFV3	117.9	100%	99%	No OMIM phenotype ?Autistic features,motor problems and macrocephaly (Asadollahi (2014) J Med Genet 51,677) ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NFS1	80.2	87%	84%	No OMIM phenotype Mitochondrial complex II/III deficienc,infantile (Farhan (2014) Mol Genet Genomic Med 2,73)
NFU1	59.7	96%	86%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NSUN3	230.5	100%	100%	No OMIM phenotype Mitochondrial disease (Van Haute (2016) Nat Commun 7)
NUBPL	102.9	92%	88%	Mitochondrial complex I deficiency, 252010
OGDH	227.7	100%	100%	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OPA1	146.6	99%	97%	Behr syndrome,210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type),616896 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	125.8	99%	97%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OXA1L	181.9	100%	99%	No OMIM phenotype
PANK2	178.4	99%	98%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200

PARS2	234.7	100%	100%	No OMIM phenotype Alpers syndrome (Sofou (2015) Mol Genet Genomic Med 3,59)
PC	166.1	99%	97%	Pyruvate carboxylase deficiency, 266150
PDHA1	112.7	98%	92%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	148.5	99%	97%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	133.8	99%	97%	Lacticacidemia due to PDX1 deficiency,245349
PDK1	166.8	97%	95%	No OMIM phenotype
PDK2	185.4	100%	100%	No OMIM phenotype
PDK3	147.1	96%	94%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	140.2	100%	99%	No OMIM phenotype ?Autism spectrum disorder (Matsunami (2014) Mol Autism 5,5)
PDP1	197.1	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	140.2	93%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	146.3	98%	95%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	98.1	98%	86%	Mitochondrial complex IV deficiency, 220110
PET112	122.3	99%	99%	No OMIM phenotype
PET117	126.1	99%	98%	No OMIM phenotype
PIGA	97	94%	86%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PITRM1	128.1	98%	95%	Brunetti et al, EMBO Mol Med 2015
PLA2G6	135.5	99%	97%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PMPCA	135.9	98%	95%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNPT1	67.9	95%	87%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POLG	128.1	100%	99%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	189.4	99%	97%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
PPA2	98.4	98%	92%	No OMIM phenotype

PTRH2	305.6	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	123.1	99%	97%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	103.5	99%	95%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	142.2	99%	96%	Leukodystrophy, hypomyelinating, 10, 616420
QRSL1	115.3	99%	95%	No OMIM phenotype Infantile mitochondrial disorder, lethal (Kohda (2016) PLoS Genet 12, e1005679)
RARS2	137.6	100%	99%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	168.4	99%	98%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	113.9	98%	94%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RRM2B	163	99%	98%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
SARS2	120.4	96%	95%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	122.6	98%	94%	Mitochondrial complex IV deficiency, 220110
SCO2	126.5	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDHA	123.2	84%	79%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	50.5	99%	97%	Mitochondrial complex II deficiency, 252011
SDHB	146.3	100%	99%	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHD	59.2	63%	58%	Carcinoid tumors, intestinal, 114900 Cowden syndrome 3, 615106 Merkel cell carcinoma, somatic Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000

				Pheochromocytoma, 171300
SELRC1	135.7	100%	100%	No OMIM phenotype
SERAC1	139.3	99%	96%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	145.1	99%	99%	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	128.3	99%	98%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	185.8	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	90.7	98%	92%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	183.5	99%	99%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	138.2	99%	96%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	79.7	99%	96%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A22	120.5	99%	97%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A3	160.6	99%	97%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	150.2	100%	100%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A4	142.3	100%	100%	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A46	206.3	97%	90%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC39A8	160.7	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SPATA5	153.8	100%	99%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG20	163.8	99%	98%	Troyer syndrome, 275900
SPG7	138.5	96%	92%	Spastic paraplegia 7, autosomal recessive, 607259
SQSTM1	142.3	99%	96%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Paget disease of bone 3, 167250
STXBP1	146.2	100%	99%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	78.2	94%	86%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	120	100%	99%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	71	94%	83%	No OMIM phenotype ?Methylmalonic aciduria (Chu (2016) Mol Genet Metab 118, 264)

SURF1	98.6	89%	88%	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
TACO1	103.3	97%	93%	Mitochondrial complex IV deficiency, 220110
TANGO2	158.8	100%	100%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TARS2	106.8	99%	96%	?Combined oxidative phosphorylation deficiency 21, 615918
TAZ	123.4	99%	98%	Barth syndrome, 302060
THG1L	154.6	100%	99%	No OMIM phenotype Cerebelar ataxia and developmental delay (Edvardson (2016) Neurogenetics, epub)
TIMM44	144.6	100%	99%	No OMIM phenotype Oncocytic thyroid carcinoma (Bonora (2006) Br J Cancer 95,1529)
TIMM50	125.1	99%	98%	No OMIM phenotype ?Epileptic encephalopathy with Lennox-Gastaut syndrome (Helbig (2016) Genet Med Epub,epub)
TIMM8A	43.7	88%	71%	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	189.8	100%	100%	No OMIM phenotype
TK2	105.4	94%	90%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM126A	112.9	98%	88%	Optic atrophy 7, 612989
TMEM126B	111.8	99%	98%	Mitochondrial complex I deficiency,252010
TMEM70	172	96%	93%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TPK1	126.3	100%	99%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRIT1	151	100%	100%	No OMIM phenotype
TRMT10C	144.7	99%	98%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	228.2	99%	96%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	118.6	100%	99%	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	123.4	98%	94%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TSFM	135.9	100%	99%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	105.8	90%	82%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	151	100%	99%	Combined oxidative phosphorylation deficiency 4, 610678
TXN2	87.8	100%	99%	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	104.7	98%	89%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC1	114.5	100%	100%	No OMIM phenotype

UQCC2	110.5	100%	100%	?Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCR10	174.2	100%	100%	No OMIM phenotype
UQCR11	232.5	100%	100%	No OMIM phenotype
UQCRB	124	99%	97%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	147.9	100%	99%	No OMIM phenotype
UQCRC2	150.8	100%	99%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	150.3	87%	82%	No OMIM phenotype
UQCRH	139.5	100%	99%	No OMIM phenotype
UQCRQ	180.8	100%	100%	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	20.5	68%	43%	Combined oxidative phosphorylation deficiency 20, 615917
YARS2	199.3	99%	99%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	125	98%	95%	?Optic atrophy 11, 617302

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
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.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 14th 2017
This list is accurate for panel version DG 2.9

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors