MITOCHONDRIAL DISORDERS GENE PANEL DG 2.7/DG 2.8

gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	132.9	99%	98%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABAT	103	100%	99%	GABA-transaminase deficiency, 613163
ACAD9	154	99%	96%	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACO2	138.6	96%	92%	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
ADCK3	146	99%	98%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCK4	106.9	100%	99%	Nephrotic syndrome, type 9, 615573
AFG3L2	126.4	92%	85%	Ataxia, spastic, 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGK	137.6	99%	97%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AIFM1	133.5	100%	99%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
ALDH1B1	224.6	100%	100%	No OMIM phenotype Bladder cancer (Nickerson (2014) Clin Cancer Res 20,4935)
ANO10	126.6	99%	96%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOA1BP	95.9	99%	99%	No OMIM phenotype Leukoencephalopathy, lethal infantile (Spiegel (2016) Neurogenetics epub,epub)
APOPT1	80.9	87%	84%	Mitochondrial complex IV deficiency, 220110
APTX	136.6	93%	90%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ATAD3A	93	89%	85%	No OMIM phenotype
ATAD3B	86.3	85%	78%	No OMIM phenotype
ATP5A1	81.9	94%	85%	?Combined oxidative phosphorylation deficiency 22, 616045 ?Mitochondrial complex (ATP synthase) deficiency, nuclear type 4, 615228
ATP5B	142.2	100%	99%	No OMIM phenotype
ATP5C1	101.2	97%	90%	No OMIM phenotype
ATP5D	65.3	96%	86%	No OMIM phenotype

ATP5E	203.2	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	86.1	96%	86%	No OMIM phenotype
ATP5G1	112.3	100%	97%	No OMIM phenotype
ATP5G2	84.3	99%	97%	No OMIM phenotype
ATP5G3	125.7	100%	100%	No OMIM phenotype
ATP5H	106	88%	70%	No OMIM phenotype
ATP5I	91.6	100%	100%	No OMIM phenotype
ATP5J	73.3	99%	92%	No OMIM phenotype
ATP5J2	129.9	99%	99%	No OMIM phenotype
ATP5L	123.5	99%	99%	No OMIM phenotype
ATP5L2	172.7	100%	100%	No OMIM phenotype
ATP5O	119.2	98%	92%	No OMIM phenotype
ATP5S	139.8	100%	99%	No OMIM phenotype
ATPAF1	96.8	82%	71%	No OMIM phenotype
ATPAF2	114.3	100%	99%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATPIF1	197.1	100%	100%	No OMIM phenotype
BCS1L	184.4	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BOLA1	113.6	100%	100%	No OMIM phenotype
BOLA2	105.9	100%	99%	No OMIM phenotype
BOLA3	59	91%	82%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
C10orf2	193.6	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	110.9	100%	98%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
C12orf65	91.3	97%	92%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	100.8	100%	99%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C19orf70	64.2	99%	97%	No OMIM phenotype
CARS2	128.6	100%	99%	Combined oxidative phosphorylation deficiency 27, 616672
CEP89	155.7	99%	97%	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	25.8	58%	38%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHKB	101.2	99%	96%	Muscular dystrophy, congenital, megaconial type, 602541
CLPB	152.7	96%	95%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	128.5	99%	95%	Perrault syndrome 3, 614129
COA1	94.6	100%	99%	No OMIM phenotype
COA3	156.2	100%	100%	No OMIM phenotype Neuropathy,exercise intolerance,obesity and short stature (Ostergaard (2015) J Med Genet 52,203
COA5	52.2	85%	84%	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	76.6	95%	87%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COASY	168	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COQ2	84.5	95%	92%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	94	86%	82%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	154.8	98%	96%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	188.1	99%	98%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ9	105.7	99%	98%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	240.2	100%	99%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	146.5	100%	99%	?Mitochondrial complex IV deficiency, 220110
COX15	105.9	100%	99%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	52.3	90%	73%	Mitochondrial complex IV deficiency, 220110
COX4I1	160.6	100%	100%	No OMIM phenotype ?Schizophrenia (Fromer (2014) Nature 506,179)
COX4I2	107	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	47.2	87%	65%	No OMIM phenotype
COX5B	143.8	100%	99%	No OMIM phenotype
COX6A1	205.9	100%	99%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	51.6	99%	96%	No OMIM phenotype
COX6B1	174.5	100%	100%	Mitochondrial complex IV deficiency, 220110

COX6B2	61	100%	98%	No OMIM phenotype
COX6C	144.3	99%	94%	No OMIM phenotype
COX7A1	109.6	99%	99%	No OMIM phenotype
COX7A2	95	98%	93%	No OMIM phenotype {insulin secretion,association with} (Olsson (2011) Eur J Endocrinol 164,765)
COX7B	60.7	76%	49%	Linear skin defects with multiple congenital anomalies, 300887
COX7B2	305.4	100%	100%	No OMIM phenotype
COX7C	65.4	99%	95%	No OMIM phenotype
COX8A	120.4	100%	100%	?Mitochondrial complex IV deficiency, 220110
COX8C	180.6	99%	96%	No OMIM phenotype
CYC1	198.4	95%	89%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	77.9	99%	95%	Thrombocytopenia 4, 612004
DARS2	137.8	99%	99%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DDHD1	166.1	96%	94%	Spastic paraplegia 28, autosomal recessive, 609340
DES	138.3	99%	98%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DGUOK	134.6	99%	98%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	162.9	99%	97%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	102.4	99%	95%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	142	99%	97%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	105.1	93%	89%	No OMIM phenotype ?Diaphragmatic hernia,congenital (Yu (2015) Hum Mol Genet 24,4764)
DNA2	149.3	99%	97%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAJC19	105.3	97%	90%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	131	99%	98%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	131.5	99%	96%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
EARS2	110.1	99%	97%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	128	100%	99%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	152.8	99%	97%	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	86.4	99%	94%	Ethylmalonic encephalopathy, 602473
FARS2	224.8	100%	99%	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FASTKD2	135.5	99%	97%	?Mitochondrial complex IV deficiency, 220110
FBXL4	227.1	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX1L	116.9	98%	96%	No OMIM phenotype Mitochondrial muscle myopathy (Spiegel (2014) Eur J Hum Genet 22,902)
FH	175.4	92%	88%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	145.1	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FXN	86.1	86%	76%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GARS	147.8	99%	97%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GATM	174.8	100%	99%	Cerebral creatine deficiency syndrome 3, 612718
GFER	91.8	97%	90%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	108.1	98%	94%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	138.1	98%	94%	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	102.2	93%	86%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	82.4	94%	86%	Hyperinsulinism-hyperammonemia syndrome, 606762
GTPBP3	135.8	99%	98%	Combined oxidative phosphorylation deficiency 23, 616198
HARS2	196.9	99%	99%	?Perrault syndrome 2, 614926
HCCS	123.6	100%	98%	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	76.2	91%	72%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	193.1	100%	100%	Holocarboxylase synthetase deficiency, 253270
HSD17B10	120.3	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438

				?Mental retardation, X-linked syndromic 10, 300220
HSPD1	92.7	96%	89%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
IARS2	147.5	100%	99%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	114.8	93%	90%	?Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
ISCA2	89.5	98%	94%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	141	99%	99%	Myopathy with lactic acidosis, hereditary, 255125
KARS	141.2	100%	99%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
LACTB	140.7	96%	85%	No OMIM phenotype
LARS2	147.8	100%	100%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LIAS	159.6	99%	95%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT1	234.6	99%	99%	Lipoyltransferase 1 deficiency, 616299
LONP1	168.1	97%	95%	CODAS syndrome, 600373
LRPPRC	140.1	98%	96%	Leigh syndrome, French-Canadian type, 220111
LYRM4	62	64%	59%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	53.9	83%	65%	Mitochondrial complex III deficiency, nuclear type 8, 615838
MARS2	168.8	100%	100%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MCUR1	63.1	77%	67%	No OMIM phenotype
MFF	107.2	92%	88%	No OMIM phenotype Mitochondrial ecephalomyopathy (Shamseldin (2012) J Med Genet 49,234) Leigh-like encephalopathy, optic atrophy and peripheral neuropathy (Koch (2016) J Med Genet 53, 270)
MFN2	159.7	100%	100%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA, 601152
MGME1	196.8	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MICU1	140	95%	91%	Myopathy with extrapyramidal signs, 615673
MIEF2	120.6	100%	99%	No OMIM phenotype
MPC1	131.6	100%	99%	Mitochondrial pyruvate carrier deficiency,614741
MPV17	119.7	100%	99%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRP63	142.1	99%	97%	No OMIM phenotype

MRPL12	125.9	99%	95%	No OMIM phenotype Growth retardation and neurological deterioration (Serre (2013) Biochim Biophys Acta 1832)
MRPL3	69.9	89%	78%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	118	99%	95%	No OMIM phenotype
MRPL44	119.9	99%	96%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS16	154.4	100%	99%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	179.6	100%	99%	No OMIM phenotype
MRPS22	150.8	95%	91%	Combined oxidative phosphorylation deficiency 5, 611719
MRPS7	185.8	100%	100%	No OMIM phenotype Sensorineural deafness,progressive hepatic and renal failure and lactic acidaemia (Menezes (2015) Hum Mol Genet 24,2297)
MRRF	216.4	100%	98%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
MTFMT	148.4	98%	94%	Combined oxidative phosphorylation deficiency 15, 614947
MTO1	179.8	89%	87%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	133	98%	93%	Ataxia, spastic, 4, 613672
NARS2	155.7	97%	97%	Combined oxidative phosphorylation deficiency 24, 616239
NDUFA1	236.8	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFA10	155.6	99%	96%	?Leigh syndrome, 256000
NDUFA11	95.1	99%	94%	Mitochondrial complex I deficiency, 252010
NDUFA12	166	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA13	131.6	95%	94%	{Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	144.1	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA3	130.5	91%	84%	No OMIM phenotype
NDUFA4	83.6	97%	86%	No OMIM phenotype Cytochrome c oxidase deficiency (Pitceathly (2013) Cell Rep 3,1795) ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA5	83.2	87%	66%	No OMIM phenotype
NDUFA6	276.9	100%	100%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA7	117.6	100%	98%	No OMIM phenotype
NDUFA8	151.6	100%	99%	No OMIM phenotype Complex I deficiency (Bugiani (2004) Biochim Biophys Acta 1659,136)
NDUFA9	156.5	99%	95%	Leigh syndrome due to mitochondrial complex I deficiency, 256000

NDUFAB1	129.9	98%	94%	No OMIM phenotype
NDUFAF1	120.7	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFAF2	59.8	81%	67%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	122.5	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFAF4	103.9	98%	91%	Mitochondrial complex I deficiency, 252010
NDUFAF5	104.7	97%	94%	Mitochondrial complex 1 deficiency, 252010
NDUFAF6	89.6	97%	93%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF7	110.4	99%	98%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFB1	33.1	73%	53%	No OMIM phenotype ?Complex I deficiency (Calvo (2012) Nat Genet 42,851)
NDUFB10	146.4	99%	97%	No OMIM phenotype
NDUFB11	101.4	94%	84%	Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB2	109.8	100%	99%	No OMIM phenotype
NDUFB3	23.3	91%	56%	Mitochondrial complex I deficiency, 252010
NDUFB4	107.3	83%	79%	No OMIM phenotype
NDUFB5	101.5	100%	100%	No OMIM phenotype
NDUFB6	41.6	97%	87%	No OMIM phenotype
NDUFB7	59.1	99%	97%	No OMIM phenotype
NDUFB8	119	100%	100%	No OMIM phenotype
NDUFB9	128.2	99%	97%	?Mitochondrial complex I deficiency, 252010
NDUFC1	95	99%	97%	No OMIM phenotype
NDUFC2	42.3	96%	82%	No OMIM phenotype {Insulin secrection,association with} (Olsson (2011) Eur J Endocrinol 164,765)
NDUFS1	154.7	99%	98%	Mitochondrial complex I deficiency, 252010
NDUFS2	120.4	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS3	151.1	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	175.1	100%	98%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS5	184.5	100%	100%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFS6	138.1	99%	99%	Mitochondrial complex I deficiency, 252010

NDUFS7	132.1	99%	99%	Leigh syndrome, 256000
NDUFS8	145.6	99%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	168.6	99%	97%	Mitochondrial complex I deficiency, 252010
NDUFV2	74.1	84%	62%	Mitochondrial complex I deficiency, 252010
NDUFV3	114	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	82.1	87%	83%	No OMIM phenotype Mitochondrial complex II/III deficienc,infantile (Farhan (2014) Mol Genet Genomic Med 2,73)
NFU1	50.5	93%	78%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NUBPL	101.7	90%	85%	Mitochondrial complex I deficiency, 252010
OGDH	225.1	100%	100%	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OPA1	135.3	98%	91%	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	122	99%	96%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OXA1L	186.8	100%	100%	No OMIM phenotype
PANK2	177.5	99%	96%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PARS2	241.4	100%	100%	No OMIM phenotype Alpers syndrome (Sofou (2015) Mol Genet Genomic Med 3,59)
PC	162.8	99%	97%	Pyruvate carboxylase deficiency, 266150
PDHA1	127.8	97%	92%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	144	98%	95%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	136.1	98%	96%	Lacticacidemia due to PDX1 deficiency,245349
PDK1	153.8	96%	93%	No OMIM phenotype
PDK2	183.5	100%	100%	No OMIM phenotype
PDK3	145.1	95%	94%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	124.1	99%	97%	No OMIM phenotype ?Autism spectrum disorder (Matsunami (2014) Mol Autism 5,5)
PDP1	209.6	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782

PDSS1	134.8	91%	85%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	131.3	97%	93%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	127.6	95%	82%	Mitochondrial complex IV deficiency, 220110
PET112	110.9	99%	98%	No OMIM phenotype
PIGA	102.1	92%	84%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PITRM1	133.2	97%	95%	Brunetti et al, EMBO Mol Med 2015
PLA2G6	132.4	99%	98%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PMPCA	146.2	98%	95%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNPT1	57.7	92%	79%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POLG	126.2	99%	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	179.6	98%	95%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
PPA2	86.9	94%	86%	No OMIM phenotype
PTRH2	315.8	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	150.8	99%	96%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	105.4	99%	94%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	137.6	99%	98%	Leukodystrophy, hypomyelinating, 10, 616420
RARS2	126.3	99%	98%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	142.9	99%	96%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	111.1	97%	93%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RRM2B	148.4	99%	97%	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	116.3	96%	95%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	130.6	97%	93%	Mitochondrial complex IV deficiency, 220110
SCO2	113.3	100%	99%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377

				Myopia 6, 608908
SDHA	117.4	84%	78%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	48.8	99%	98%	Mitochondrial complex II deficiency, 252011
SDHB	144	99%	99%	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHD	59.9	62%	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
SERAC1	125.5	98%	94%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	155.3	99%	98%	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	128.5	99%	98%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	191.3	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	84.7	97%	90%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	165	99%	98%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	125.3	98%	93%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	81.4	99%	95%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A22	117.1	99%	96%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A3	157.9	99%	95%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	132.1	100%	99%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A4	152.1	100%	100%	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283

SLC25A46	191.1	93%	87%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SPATA5	146.6	99%	99%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG20	166.4	99%	97%	Troyer syndrome, 275900
SPG7	127.9	96%	92%	Spastic paraplegia 7, autosomal recessive, 607259
STXBP1	147.9	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	69.4	92%	82%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	111.3	99%	97%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	65.9	91%	79%	No OMIM phenotype
SURF1	97	89%	88%	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
TACO1	104.3	96%	92%	Mitochondrial complex IV deficiency, 220110
TANGO2	161	100%	99%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TARS2	103.6	99%	96%	?Combined oxidative phosphorylation deficiency 21, 615918
TAZ	126.3	100%	98%	Barth syndrome, 302060
THG1L	158.2	100%	99%	No OMIM phenotype
TIMM44	136.3	99%	97%	No OMIM phenotype Oncocytic thyroid carcinoma (Bonora (2006) Br J Cancer 95,1529)
TIMM50	115.1	99%	97%	No OMIM phenotype ?Epileptic encephalopathy with Lennox-Gastaut syndrome (Helbig (2016) Genet Med Epub,epub)
TIMM8A	45.5	87%	70%	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	167.1	100%	100%	No OMIM phenotype
TK2	109.9	92%	87%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM126A	118.9	95%	83%	Optic atrophy 7, 612989
TMEM126B	100.8	99%	97%	Mitochondrial complex I deficiency,252010
TMEM70	152.6	95%	91%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TPK1	127.3	99%	97%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRIT1	149.2	100%	99%	No OMIM phenotype
TRMT10C	123.5	99%	96%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	232.1	98%	93%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	121.1	100%	99%	Liver failure, transient infantile, 613070

				{Deafness, mitochondrial, modifier of}, 580000
TRNT1	111.2	95%	90%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TSFM	148.4	100%	99%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	106.2	90%	81%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	146.1	100%	99%	Combined oxidative phosphorylation deficiency 4, 610678
TXN2	100.5	100%	100%	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	96.7	96%	88%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC1	114.2	100%	99%	No OMIM phenotype
UQCC2	103.8	100%	99%	?Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCR10	186.8	100%	100%	No OMIM phenotype
UQCR11	199.9	100%	100%	No OMIM phenotype
UQCRB	121.7	98%	95%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	143.6	99%	99%	No OMIM phenotype
UQCRC2	154.8	99%	99%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	148.5	88%	83%	No OMIM phenotype
UQCRH	130	99%	98%	No OMIM phenotype
UQCRQ	162.5	100%	99%	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	17.7	62%	35%	Combined oxidative phosphorylation deficiency 20, 615917
YARS2	186.8	99%	98%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561

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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

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