MITOCHONDRIAL DISORDERS GENE PANEL DG 2.13 (356 genes)

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AARS2	126.2	100	99	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABAT	92.7	100	99	GABA-transaminase deficiency, 613163
ACAD9	135.2	98	95	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126
ACO2	129.3	95	91	Infantile cerebellar-retinal degeneration, 614559 ?Optic atrophy 9, 616289
AFG3L2	121	91	84	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGK	112.1	99	96	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AIFM1	106.2	100	99	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
ALDH1B1	206.6	100	100	No OMIM phenotype Bladder cancer (Nickerson (2014) Clin Cancer Res 20,4935)
ANO10	116.7	98	96	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOPT1	63.8	81	78	Mitochondrial complex IV deficiency, 220110
APTX	118.9	94	91	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ATAD3A	87.9	89	86	Harel-Yoon syndrome, 617183
ATAD3B	83.8	88	82	No OMIM phenotype Late-onset encephalopathy with cerebellar atrophy, ataxia and dystonia (Desai (2017) Brain 140,1595)
ATP13A2	117.4	100	98	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693
ATP5A1	85.3	94	85	?Combined oxidative phosphorylation deficiency 22, 616045 ?Mitochondrial complex (ATP synthase) deficiency, nuclear type 4, 615228
ATP5B	129.2	100	99	No OMIM phenotype
ATP5C1	90.1	95	84	No OMIM phenotype
ATP5D	66.1	98	90	No OMIM phenotype

ATP5E	135.5	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	80.9	96	84	No OMIM phenotype
ATP5G1	110	100	98	No OMIM phenotype
ATP5G2	93.8	100	98	No OMIM phenotype
ATP5G3	118.5	100	100	No OMIM phenotype
ATP5H	109.7	93	71	No OMIM phenotype
ATP5I	69.6	99	97	No OMIM phenotype
ATP5J	66.4	99	90	No OMIM phenotype
ATP5J2	109.7	100	99	No OMIM phenotype
ATP5L	144.6	100	99	No OMIM phenotype
ATP5L2	217.8	100	100	No OMIM phenotype
ATP5O	101.3	99	90	No OMIM phenotype
ATP5S	133.6	100	100	No OMIM phenotype
ATPAF1	79.8	74	68	No OMIM phenotype
ATPAF2	101.4	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATPIF1	174.3	100	100	No OMIM phenotype
BCS1L	182.3	100	100	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BOLA1	110.6	100	99	No OMIM phenotype
BOLA2	120.5	100	100	No OMIM phenotype ?Autism and developmental delay (Nuttle (2016) Nature 536, 205)
BOLA3	50.1	92	81	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
C12orf65	88.2	97	91	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	93.9	100	99	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C19orf70	63	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)
C1QBP	80.7	81	71	Combined oxidative phosphorylation deficiency 33, 617713
CA5A	124.1	99	94	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CARS2	121.1	100	99	Combined oxidative phosphorylation deficiency 27, 616672

CEP89	125.5	94	91	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	20	43	35	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHKB	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CLPB	140.2	100	99	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	115.4	99	96	Perrault syndrome 3, 614129
COA1	89.4	100	100	No OMIM phenotype
COA3	143.3	100	100	No OMIM phenotype Neuropathy,exercise intolerance,obesity and short stature (Ostergaard (2015) J Med Genet 52,203
COA5	59.2	85	84	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	78.7	98	91	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	146	100	100	?Mitochondrial complex IV deficiency, 220110
COASY	168.5	100	100	Neurodegeneration with brain iron accumulation 6, 615643
COQ2	89.3	96	93	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	89.8	88	84	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	184.4	100	100	No OMIM phenotype Cerebellar ataxia and static encephalopmyopathy (Malicdan (2018) Hum Mutat 39,69) Intellectual disability (Najmabadi (2011) Nature 478,57)
COQ6	143.9	99	96	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	158.5	99	98	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	134.3	100	99	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	90.5	100	99	Nephrotic syndrome, type 9, 615573
COQ9	91.4	99	96	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	241.9	100	99	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	108.1	100	99	?Mitochondrial complex IV deficiency, 220110
COX15	98.6	100	99	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	58.1	83	65	Mitochondrial complex IV deficiency, 220110

COX4I1	133.9	100	100	No OMIM phenotype ?Schizophrenia (Fromer (2014) Nature 506,179)
COX4I2	120.1	100	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	37.6	80	57	No OMIM phenotype
COX5B	126.9	100	100	No OMIM phenotype
COX6A1	180.6	100	99	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	32.4	97	80	No OMIM phenotype
COX6B1	159.6	100	100	Mitochondrial complex IV deficiency, 220110
COX6B2	62.3	100	99	No OMIM phenotype
COX6C	131.4	99	90	No OMIM phenotype
COX7A1	81.7	99	98	No OMIM phenotype
COX7A2	82.9	99	92	No OMIM phenotype {insulin secretion,association with} (Olsson (2011) Eur J Endocrinol 164,765)
COX7B	47.9	73	42	Linear skin defects with multiple congenital anomalies, 300887
COX7B2	247.3	100	100	No OMIM phenotype
COX7C	48.9	99	94	No OMIM phenotype
COX8A	98.1	100	100	?Mitochondrial complex IV deficiency, 220110
COX8C	159.2	99	97	No OMIM phenotype ?Tethered spinal cord syndrome (Zhao (2016) Neural Regen Res 11, 1333)
CP	120	93	89	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CTBP1	93.4	96	85	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CYC1	184.5	88	86	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	72.1	99	95	Thrombocytopenia 4, 612004
DARS2	122.3	100	99	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF17	91.9	95	89	Woodhouse-Sakati syndrome, 241080
DDHD1	141.8	97	94	Spastic paraplegia 28, autosomal recessive, 609340
DES	120.8	99	98	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 ?Muscular dystrophy, limb-girdle, type 2R, 615325
DGUOK	119.2	100	100	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	141	99	98	2-aminoadipic 2-oxoadipic aciduria, 204750

				?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	91.6	99	96	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	123.5	99	98	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	95.8	94	89	No OMIM phenotype ?Diaphragmatic hernia,congenital (Yu (2015) Hum Mol Genet 24,4764)
DMAC1	50.4	99	96	No OMIM phenotype
DMAC2	135.1	98	98	No OMIM phenotype
DNA2	123.6	99	96	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAJC19	97.9	98	90	3-methylglutaconic aciduria, type V, 610198
DNAJC3	116.3	99	98	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	123.5	99	96	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
EARS2	103.4	99	98	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	112.8	99	97	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	141.4	99	98	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
EHHADH	163.2	100	99	?Fanconi renotubular syndrome 3, 615605
ELAC2	123.8	100	99	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ERAL1	181.5	100	100	Perrault syndrome 6, 617565
ETHE1	85.5	99	95	Ethylmalonic encephalopathy, 602473
FA2H	94.1	87	79	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	207.7	100	100	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FASTKD2	118.9	99	96	?Mitochondrial complex IV deficiency, 220110
FBXL4	189.8	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX2	114.3	99	99	No OMIM phenotype Mitochondrial muscle myopathy (Spiegel (2014) Eur J Hum Genet 22,902)
FDXR	93.4	100	99	Auditory neuropathy and optic atrophy, 617717
FH	146.4	91	87	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	136.6	100	99	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010

FTL	147.7	99	93	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FXN	75.2	85	75	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GARS	125.7	99	98	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GATB	101.5	99	98	No OMIM phenotype
GATC	126.2	100	100	No OMIM phenotype
GATM	150.6	100	100	Cerebral creatine deficiency syndrome 3, 612718
GFER	76.1	92	75	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	100.3	99	95	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	118.6	98	93	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	108.2	92	83	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	74.5	94	84	Hyperinsulinism-hyperammonemia syndrome, 606762
GTPBP2	156.8	96	94	No OMIM phenotype
GTPBP3	137.4	100	99	Combined oxidative phosphorylation deficiency 23, 616198
HARS2	169.7	99	99	?Perrault syndrome 2, 614926
HCCS	106.6	99	99	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	67.7	92	69	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	172.8	100	100	Holocarboxylase synthetase deficiency, 253270
HSD17B10	117.1	100	99	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSPA9	91.6	91	85	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPD1	96.5	98	93	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HTRA2	122.1	100	99	3-methlyglutaconic aciduria, type VIII,617248 {Parkinson disease 13}, 610297

IARS2	131.5	100	99	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	113.3	93	89	?Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
ISCA2	92	99	96	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	111.2	100	99	Myopathy with lactic acidosis, hereditary, 255125
KARS	122.6	100	99	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
LACTB	117.2	93	80	No OMIM phenotype
LARS2	143	100	100	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LIAS	133.7	99	97	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT1	227.4	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	92.2	97	83	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LONP1	141.5	97	96	CODAS syndrome, 600373
LRPPRC	127.3	99	97	Leigh syndrome, French-Canadian type, 220111
LYRM4	60.1	63	54	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	49	87	72	Mitochondrial complex III deficiency, nuclear type 8, 615838
MARS2	173.2	100	100	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MCUR1	65.3	93	77	No OMIM phenotype
MDH2	123.3	98	97	Epileptic encephalopathy, early infantile, 51, 617339
MECR	108.1	98	96	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	93.7	90	87	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	150.6	100	99	Charcot-Marie-Tooth disease, type 2A2A, 609260 Charcot-Marie-Tooth disease, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MGME1	151.1	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MICU1	134.2	96	88	Myopathy with extrapyramidal signs, 615673
MICU2	43	94	86	No OMIM phenotype
MIEF2	122.9	100	99	No OMIM phenotype
MIPEP	102.1	95	88	Combined oxidative phosphorylation deficiency 31, 617228
MPC1	121.8	100	99	Mitochondrial pyruvate carrier deficiency,614741
MPV17	108.5	100	99	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810

MRM2	122.9	99	96	No OMIM phenotype Encephalomyopathy, childhood-onset and stroke-like episodes (Garone (2017) Hum Mol Genet 26,4257)
MRPL12	107.3	99	93	No OMIM phenotype Growth retardation and neurological deterioration (Serre (2013) Biochim Biophys Acta 1832)
MRPL3	66.3	91	77	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	101.6	99	96	No OMIM phenotype
MRPL44	110.5	99	97	?Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	155	100	99	No OMIM phenotype
MRPS16	161.1	100	99	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	166.4	99	97	No OMIM phenotype
MRPS22	138.8	95	91	Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	156.7	100	99	No OMIM phenotype
MRPS34	132.7	99	98	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	173.5	100	100	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	190.2	100	100	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
MSTO1	140	99	97	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	124.6	99	96	Combined oxidative phosphorylation deficiency 15, 614947
MTO1	173.7	89	87	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	109.6	98	93	Ataxia, spastic, 4, 613672
NARS2	120	97	97	Combined oxidative phosphorylation deficiency 24, 616239
NAXE	81.1	99	95	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFA1	166.8	100	99	Mitochondrial complex I deficiency, 252010
NDUFA10	136.7	98	96	?Leigh syndrome, 256000
NDUFA11	86.9	99	95	Mitochondrial complex I deficiency, 252010
NDUFA12	160.2	100	100	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA13	91.4	92	91	{Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	133.9	100	100	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA3	129.7	91	87	No OMIM phenotype
NDUFA4	79.9	98	84	No OMIM phenotype Cytochrome c oxidase deficiency (Pitceathly (2013) Cell Rep 3,1795) ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA5	71	87	61	No OMIM phenotype

NDUFA6	212.4	100	100	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA7	114.8	100	99	No OMIM phenotype
NDUFA8	138.4	100	99	No OMIM phenotype Complex I deficiency (Bugiani (2004) Biochim Biophys Acta 1659,136)
NDUFA9	124.7	98	93	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAB1	116.3	99	96	No OMIM phenotype
NDUFAF1	115.6	100	100	Mitochondrial complex I deficiency, 252010
NDUFAF2	58.6	85	70	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	120.8	100	100	Mitochondrial complex I deficiency, 252010
NDUFAF4	79.4	98	91	Mitochondrial complex I deficiency, 252010
NDUFAF5	95.7	98	94	Mitochondrial complex 1 deficiency, 252010
NDUFAF6	79.3	97	85	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF7	101.2	100	98	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFB1	29.3	60	53	No OMIM phenotype ?Complex I deficiency (Calvo (2012) Nat Genet 42,851)
NDUFB10	120.2	99	95	No OMIM phenotype Complex I deficiency (Friederich (2016) Hum Mol Genet)
NDUFB11	109.6	94	88	Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB2	102.4	100	100	No OMIM phenotype
NDUFB3	22.6	91	59	Mitochondrial complex I deficiency, 252010
NDUFB4	100.6	85	82	No OMIM phenotype
NDUFB5	88.5	100	100	No OMIM phenotype
NDUFB6	39.7	99	91	No OMIM phenotype
NDUFB7	50.4	100	97	No OMIM phenotype
NDUFB8	116.6	100	100	No OMIM phenotype Psychomotor retardation, Leigh syndrome, leukodystrophy and complex I deficiency (Pronicka (2016) J Transl Med 14,174)
NDUFB9	120.1	99	97	?Mitochondrial complex I deficiency, 252010
NDUFC1	87.2	100	98	No OMIM phenotype
NDUFC2	39.9	98	84	No OMIM phenotype {Insulin secrection,association with} (Olsson (2011) Eur J Endocrinol 164,765)

NDUFS1	132.2	99	98	Mitochondrial complex I deficiency, 252010
NDUFS2	117.8	100	100	Mitochondrial complex I deficiency, 252010
NDUFS3	142.4	90	90	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	147.3	100	99	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS5	196.7	100	100	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFS6	119.1	99	99	Mitochondrial complex I deficiency, 252010
NDUFS7	118.4	100	99	Leigh syndrome, 256000
NDUFS8	141.4	100	99	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	136.7	99	97	Mitochondrial complex I deficiency, 252010
NDUFV2	69.5	78	53	Mitochondrial complex I deficiency, 252010
NDUFV3	119.3	99	98	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	86	83	No OMIM phenotype Mitochondrial complex II/III deficienc,infantile (Farhan (2014) Mol Genet Genomic Med 2,73)
NFU1	47.7	94	77	Multiple mitochondrial dysfunctions syndrome 1, 605711
NSUN3	187.1	100	100	No OMIM phenotype Mitochondrial disease (Van Haute (2016) Nat Commun 7)
NUBPL	89.8	92	85	Mitochondrial complex I deficiency, 252010
OGDH	201.3	100	100	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OPA1	122.5	99	94	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	128	99	97	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OXA1L	162	100	100	No OMIM phenotype
PANK2	146.6	99	93	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PARS2	219.1	100	100	No OMIM phenotype

				Alpers syndrome (Sofou (2015) Mol Genet Genomic Med 3,59)
PC	149.3	97	94	Pyruvate carboxylase deficiency, 266150
PDHA1	109.8	98	92	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	133	99	96	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	132.5	98	94	Lacticacidemia due to PDX1 deficiency,245349
PDK1	127.4	97	94	No OMIM phenotype
PDK2	156.8	100	100	No OMIM phenotype
PDK3	105.1	96	94	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	110.1	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	116.7	88	78	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	126.8	96	93	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	94.5	88	74	Mitochondrial complex IV deficiency, 220110
PET117	95.3	100	99	No OMIM phenotype
PIGA	90.5	90	81	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PITRM1	117.7	97	95	Brunetti et al, EMBO Mol Med 2015
PLA2G6	117.5	99	98	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PMPCA	120.8	99	96	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121	99	97	No OMIM phenotype
PNPT1	53.7	93	80	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POLG	114.4	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	157.5	98	96	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
PPA2	80.4	94	82	?Sudden cardiac failure, infantile, 617222 ?Sudden cardiac failure, alcohol-induced,617223

PRKAA1	119.9	100	99	No OMIM phenotype
PTRH2	279.6	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	127.2	98	93	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	86.3	99	94	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	127.6	100	97	Leukodystrophy, hypomyelinating, 10, 616420
QRSL1	96.7	98	93	No OMIM phenotype Infantile mitochondrial disorder, lethal (Kohda (2016) PLoS Genet 12, e1005679)
RARS2	107.2	100	99	Pontocerebellar hypoplasia, type 6, 611523
RMND1	137.2	99	97	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	98.6	99	95	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RRM2B	128.6	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
RTN4IP1	98.1	99	99	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
SACS	154.5	100	99	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	127.9	99	96	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SARS2	104.8	94	92	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	109.6	97	94	Mitochondrial complex IV deficiency, 220110
SCO2	113.1	100	100	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	106.9	99	96	Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDHA	122.2	84	80	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	42.5	100	96	Mitochondrial complex II deficiency, 252011
SDHB	120.3	100	99	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864

				Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHD	48.4	55	50	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	112.5	98	94	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	131.7	100	99	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	119.5	99	97	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	186.4	100	99	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	71	92	87	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A10	70.2	76	68	No OMIM phenotype
SLC25A12	150.5	99	98	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	110.7	95	92	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	88.6	99	98	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A21	114	100	98	No OMIM phenotype ?Synpolydactyly (Meyertholen (2012) Mol Syndromol 3 25)
SLC25A22	108.7	99	96	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	115.8	98	96	Fontaine progeroid syndrome ,612289
SLC25A3	139	99	97	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	117	100	100	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A4	134.1	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
SLC25A42	116.9	97	94	No OMIM phenotype Mitochondrial myopathy (Shamseldin (2016) Hum Genet 135,21)
SLC25A46	205.7	95	87	Neuropathy, hereditary motor and sensory, type VIB, 616505

SLC39A8	128.5	100	99	Congenital disorder of glycosylation, type IIn, 616721
SLC52A2	177.6	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	119.6	100	100	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SPART	132.4	99	98	Troyer syndrome, 275900
SPATA5	132	99	99	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG7	119.2	93	92	Spastic paraplegia 7, autosomal recessive, 607259
SQSTM1	109.1	98	94	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Paget disease of bone 3, 167250
STAT2	116	100	99	Immunodeficiency 44, 616636
STXBP1	124.5	96	96	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	64.9	93	82	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	101.3	99	95	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	57.8	91	78	No OMIM phenotype ?Methylmalonic aciduria (Chu (2016) Mol Genet Metab 118, 264)
SURF1	96.2	88	88	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
TACO1	91.7	97	92	Mitochondrial complex IV deficiency, 220110
TANGO2	145.3	100	100	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TARS2	98.8	99	98	?Combined oxidative phosphorylation deficiency 21, 615918
TAZ	94	99	98	Barth syndrome, 302060
THG1L	142.9	100	99	No OMIM phenotype Cerebelar ataxia and developmental delay (Edvardson (2016) Neurogenetics, epub)
TIMM44	123.3	100	98	No OMIM phenotype Oncocytic thyroid carcinoma (Bonora (2006) Br J Cancer 95,1529)
TIMM50	108.2	98	95	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	46	94	78	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	152.2	100	100	Mitochondrial complex I deficiency, 252010
TK2	105.7	93	89	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM126A	120.3	98	86	Optic atrophy 7, 612989

TMEM126B	79.2	99	97	Mitochondrial complex I deficiency,252010
TMEM186	152.4	100	100	No OMIM phenotype
TMEM65	50.2	79	65	No OMIM phenotype
TMEM70	138.7	94	90	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TPK1	112.7	99	97	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRIT1	119.4	100	99	Combined oxidative phosphorylation deficiency 35, 617873
TRMT10C	131.4	99	98	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	208.7	99	93	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	99	100	99	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	104.6	97	92	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TSFM	127.2	100	100	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	92.1	80	72	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	135.4	100	99	Combined oxidative phosphorylation deficiency 4, 610678
TWNK	178.8	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
TXN2	81.2	100	100	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	95.2	98	85	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC1	96.7	100	100	No OMIM phenotype
UQCC2	96.6	100	99	?Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	95	100	99	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	189.2	100	100	No OMIM phenotype
UQCR11	158.7	100	100	No OMIM phenotype
UQCRB	107.6	99	96	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	151.4	100	99	No OMIM phenotype
UQCRC2	122.6	99	99	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	151.9	87	82	No OMIM phenotype
UQCRH	131.1	99	98	No OMIM phenotype
UQCRQ	131.3	100	99	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	110.9	99	98	Combined oxidative phosphorylation deficiency 20, 615917

VPS13D	158.6	99	99	No OMIM phenotype Schizophrenia (McCarthy (2014) Mol Psychiatry 19, 652)
WARS2	140.7	100	99	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WDR45	75	97	90	Neurodegeneration with brain iron accululation 5, 300894
YARS2	173.2	99	98	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	105.3	97	91	?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 18th, 2018.
This list is accurate for panel version DG 2.13
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors