MITOCHONDRIAL DISORDERS GENE PANEL DG 2.16 (393 genes)

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Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AARS2	122,7	100.0%	99.8%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABAT	83,2	99.9%	98.3%	GABA-transaminase deficiency, 613163
ACAD9	124,3	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACO2	115,3	95.8%	89.5%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ADAMTS10	122,8	100.0%	99.8%	Weill-Marchesani syndrome 1, recessive, 277600
ADPRHL2	163,8	100.0%	100.0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	98,3	95.9%	86.1%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGK	108,5	99.5%	95.7%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AIFM1	90	99.8%	96.7%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
ALDH1B1	186,5	100.0%	100.0%	No OMIM phenotype Bladder cancer (Nickerson (2014) Clin Cancer Res 20,4935)
ALKBH1	100,5	100.0%	99.5%	No OMIM phenotype
ANO10	106	98.9%	96.3%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOPT1	NC	NC	NC	Mitochondrial complex IV deficiency, 220110
APTX	96,3	94.1%	91.3%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARL2	131,2	100.0%	99.9%	No OMIM phenotype
ATAD1	65,2	99.3%	89.8%	Hyperekplexia 4, 618011
ATAD3A	90,3	93.6%	87.5%	Harel-Yoon syndrome, 617183
ATAD3B	97	92.3%	83.7%	No OMIM phenotype Late-onset encephalopathy with cerebellar atrophy, ataxia and dystonia (Desai (2017) Brain 140,1595)
ATP13A2	134,1	99.9%	99.7%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP5A1	NC	NC	NC	?Combined oxidative phosphorylation deficiency 22, 616045 ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228

ATP5B	NC	NC	NC	No OMIM phenotype
ATP5C1	NC	NC	NC	No OMIM phenotype
ATP5D	NC	NC	NC	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	NC	NC	NC	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	NC	NC	NC	No OMIM phenotype
ATP5G1	NC	NC	NC	No OMIM phenotype
ATP5G2	NC	NC	NC	No OMIM phenotype
ATP5G3	NC	NC	NC	No OMIM phenotype
ATP5H	NC	NC	NC	No OMIM phenotype
ATP5I	NC	NC	NC	No OMIM phenotype
ATP5J	NC	NC	NC	No OMIM phenotype
ATP5J2	NC	NC	NC	No OMIM phenotype
ATP5L	NC	NC	NC	No OMIM phenotype
ATP5L2	NC	NC	NC	No OMIM phenotype
ATP5O	NC	NC	NC	No OMIM phenotype
ATP5S	NC	NC	NC	No OMIM phenotype
ATPAF1	71,7	95.7%	84.8%	No OMIM phenotype
ATPAF2	103,5	100.0%	100.0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATPIF1	NC	NC	NC	No OMIM phenotype
BCS1L	147,9	100.0%	100.0%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BOLA1	117,6	100.0%	100.0%	No OMIM phenotype
BOLA2	112,2	100.0%	100.0%	No OMIM phenotype ?Autism and developmental delay (Nuttle (2016) Nature 536, 205)
BOLA3	48,1	99.9%	92.5%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
C12orf65	110,4	100.0%	99.6%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	104,2	100.0%	99.8%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
C19orf70	NC	NC	NC	Combined oxidative phosphorylation deficiency 37, 618329
C1QBP	66,7	91.9%	79.5%	Combined oxidative phosphorylation deficiency 33, 617713
CA5A	93,2	99.6%	95.7%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CARS2	128,2	100.0%	100.0%	Combined oxidative phosphorylation deficiency 27, 616672
CEP89	127,5	97.4%	94.7%	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	26,1	63.1%	38.4%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048
CHCHD2	69,7	99.9%	93.7%	Parkinson disease 22, autosomal dominant, 616710
CHKB	115,4	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CISD2	113,6	83.4%	83.3%	Wolfram syndrome 2, 604928
CLPB	125,6	99.8%	97.9%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	139,3	100.0%	99.2%	Perrault syndrome 3, 614129
COA1	84,2	100.0%	99.8%	No OMIM phenotype
COA3	159,3	100.0%	100.0%	No OMIM phenotype Neuropathy,exercise intolerance,obesity and short stature (Ostergaard (2015) J Med Genet 52,203
COA5	74,9	86.6%	83.5%	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	112,5	99.4%	97.0%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	122	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COASY	172,7	100.0%	100.0%	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266
COQ2	103,5	97.6%	97.1%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	105	91.3%	90.2%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	168,1	100.0%	100.0%	No OMIM phenotype Cerebellar ataxia and static encephalopmyopathy (Malicdan (2018) Hum Mutat 39,69) Intellectual disability (Najmabadi (2011) Nature 478,57)
COQ6	127,5	99.9%	98.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	138,3	99.9%	99.6%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	161,8	100.0%	99.9%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	99,5	100.0%	99.8%	Nephrotic syndrome, type 9, 615573
COQ9	73,8	100.0%	98.1%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	220,4	100.0%	99.9%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	95,2	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
COX15	87,7	99.9%	98.3%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	66,2	96.4%	85.3%	Mitochondrial complex IV deficiency, 220110
COX4I1	108,8	100.0%	100.0%	No OMIM phenotype ?Schizophrenia (Fromer (2014) Nature 506,179)
COX4I2	116,5	100.0%	99.6%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714

COX5A	29,7	80.0%	47.5%	No OMIM phenotype
COX5B	138,5	100.0%	100.0%	No OMIM phenotype
COX6A1	148,3	100.0%	99.9%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	58,4	99.7%	95.1%	No OMIM phenotype
COX6B1	139,1	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110
COX6B2	93,5	100.0%	99.8%	No OMIM phenotype
COX6C	123	99.8%	96.7%	No OMIM phenotype
COX7A1	125,7	100.0%	99.9%	No OMIM phenotype
COX7A2	89,8	100.0%	99.1%	No OMIM phenotype {insulin secretion,association with} (Olsson (2011) Eur J Endocrinol 164,765)
COX7B	38,6	62.3%	33.6%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	143,8	100.0%	99.9%	No OMIM phenotype
COX7C	37,5	97.5%	83.0%	No OMIM phenotype
COX8A	109,4	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
COX8C	157,3	100.0%	100.0%	No OMIM phenotype ?Tethered spinal cord syndrome (Zhao (2016) Neural Regen Res 11, 1333)
CP	100,6	93.1%	87.4%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CTBP1	101,2	94.3%	86.7%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CYC1	152,2	99.3%	95.6%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	61	99.1%	93.1%	Thrombocytopenia 4, 612004
DARS2	126,8	100.0%	98.6%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF17	90,4	99.9%	97.9%	Woodhouse-Sakati syndrome, 241080
DDHD1	161,6	99.9%	98.4%	Spastic paraplegia 28, autosomal recessive, 609340
DES	125	100.0%	100.0%	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DGUOK	119,4	99.9%	97.9%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DHTKD1	122,4	99.9%	98.8%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	100,2	99.8%	99.2%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	119,2	99.9%	99.7%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	78,6	94.9%	87.3%	No OMIM phenotype ?Diaphragmatic hernia,congenital (Yu (2015) Hum Mol Genet 24,4764)

DMAC1	56	100.0%	98.3%	No OMIM phenotype
DMAC2	123,9	98.3%	98.3%	No OMIM phenotype
DNA2	124,3	99.7%	97.3%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJA3	120,7	99.9%	98.6%	No OMIM phenotype
DNAJC19	93,8	98.4%	92.3%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	137,4	100.0%	99.7%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	119,5	99.9%	98.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
EARS2	99	99.7%	97.8%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	103,8	100.0%	99.7%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	146,1	100.0%	100.0%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
EHHADH	133,4	100.0%	99.7%	?Fanconi renotubular syndrome 3, 615605
ELAC2	109,4	99.9%	99.0%	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ERAL1	156,5	100.0%	100.0%	Perrault syndrome 6, 617565
ETFDH	114,4	100.0%	99.3%	Glutaric acidemia IIC, 231680
ETHE1	97,3	99.9%	97.8%	Ethylmalonic encephalopathy, 602473
FA2H	92,7	98.8%	92.5%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	161,9	100.0%	100.0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSB	78	97.4%	92.7%	Rajab interstitial lung disease with brain calcifications, 613658
FASTKD2	115,5	99.6%	97.9%	?Mitochondrial complex IV deficiency, 220110
FBXL4	168,9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX2	147,2	100.0%	100.0%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	122,3	99.9%	99.1%	Auditory neuropathy and optic atrophy, 617717
FH	128	95.0%	88.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	121	99.8%	98.2%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FTL	145,2	99.7%	96.7%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FXN	64,9	99.7%	96.8%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GARS	128,9	100.0%	99.7%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794

GATB	99,8	100.0%	99.4%	No OMIM phenotype
GATC	141,7	100.0%	100.0%	No OMIM phenotype
GATM	137,3	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GFER	90,6	100.0%	99.6%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	104,2	99.9%	99.0%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	121,3	98.9%	95.6%	Combined oxidative phosphorylation deficiency 39, 618397
GLRX5	137,6	99.6%	96.1%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	65,5	98.1%	87.5%	Hyperinsulinism-hyperammonemia syndrome, 606762
GTPBP2	128,9	99.7%	98.6%	Jaberi-Elahi syndrome, 617988
GTPBP3	164,7	100.0%	100.0%	Combined oxidative phosphorylation deficiency 23, 616198
HARS2	136,6	100.0%	99.9%	?Perrault syndrome 2, 614926
HCCS	92,4	99.2%	95.2%	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	69,9	96.3%	79.8%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	142,3	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HSD17B10	92,4	100.0%	98.4%	HSD10 mitochondrial disease, 300438
HSPA9	82,6	89.5%	84.2%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPD1	74,3	98.1%	92.5%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HTRA2	132,6	100.0%	99.6%	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297
IARS2	142,9	100.0%	99.9%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	137,4	99.3%	95.9%	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ISCA1	67,2	93.5%	84.0%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	105,1	99.8%	95.8%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	117,2	100.0%	99.9%	Myopathy with lactic acidosis, hereditary, 255125
KARS	104,1	100.0%	98.8%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KIF1A	115	99.7%	97.6%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
LACTB	112,1	99.7%	97.7%	No OMIM phenotype
LARS2	122,8	100.0%	100.0%	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300

LIAS	125,3	99.9%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT1	203,2	100.0%	99.9%	Lipoyltransferase 1 deficiency, 616299
LIPT2	91,2	99.9%	99.3%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LONP1	148	100.0%	100.0%	CODAS syndrome, 600373
LRPPRC	129,3	100.0%	99.6%	Leigh syndrome, French-Canadian type, 220111
LYRM4	77,5	67.4%	62.4%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	61,9	98.7%	91.3%	Mitochondrial complex III deficiency, nuclear type 8, 615838
MARS2	178,4	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MCUR1	66,7	99.9%	98.0%	No OMIM phenotype
MDH2	109,4	98.0%	97.9%	Epileptic encephalopathy, early infantile, 51, 617339
MECR	108,2	100.0%	99.7%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	86,2	93.7%	89.6%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	122,8	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MGME1	142,4	100.0%	99.9%	Mitochondrial DNA depletion syndrome 11, 615084
MICU1	103,3	98.8%	96.5%	Myopathy with extrapyramidal signs, 615673
MICU2	58,6	99.4%	95.3%	No OMIM phenotype
MIEF2	138,7	100.0%	99.8%	No OMIM phenotype
MIPEP	99,3	99.4%	96.6%	Combined oxidative phosphorylation deficiency 31, 617228
MPC1	153,1	100.0%	99.6%	Mitochondrial pyruvate carrier deficiency, 614741
MPV17	88,6	100.0%	98.9%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRM2	113,6	99.9%	98.7%	No OMIM phenotype Encephalomyopathy, childhood-onset and stroke-like episodes (Garone (2017) Hum Mol Genet 26,4257)
MRPL12	118,8	100.0%	99.9%	No OMIM phenotype Growth retardation and neurological deterioration (Serre (2013) Biochim Biophys Acta 1832)
MRPL3	63,4	93.2%	81.4%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	84,6	99.1%	94.7%	No OMIM phenotype
MRPL44	126,3	100.0%	99.7%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	222,4	100.0%	99.7%	No OMIM phenotype
MRPS14	168,7	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	127	99.9%	98.5%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	158,6	100.0%	99.9%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	134,6	99.9%	98.7%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117

MRPS23	142,6	99.9%	99.7%	No OMIM phenotype
MRPS25	133,8	100.0%	99.6%	No OMIM phenotype
MRPS28	143,4	87.4%	86.6%	No OMIM phenotype
MRPS34	169	100.0%	99.9%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	153,2	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	141,1	100.0%	100.0%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
MSTO1	101,4	99.3%	96.1%	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	132,5	100.0%	99.8%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTO1	143,7	91.4%	89.6%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	122,3	99.0%	93.2%	?Spastic ataxia 4, autosomal recessive, 613672
NARS2	120,7	97.6%	97.2%	Combined oxidative phosphorylation deficiency 24, 616239
NAXD	132,1	100.0%	99.9%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	81,4	99.7%	97.0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFA1	184,9	99.9%	99.2%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	114,6	99.9%	98.9%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	116	99.8%	97.4%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	160,8	100.0%	100.0%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	121,3	92.3%	91.7%	?Mitochondrial complex I deficiency, nuclear type 28, 618249 {Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	162,6	100.0%	99.6%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	130,1	92.1%	88.3%	No OMIM phenotype
NDUFA4	68	98.9%	89.4%	No OMIM phenotype Cytochrome c oxidase deficiency (Pitceathly (2013) Cell Rep 3,1795) ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA5	62,7	91.4%	70.0%	No OMIM phenotype
NDUFA6	201,9	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	111,2	100.0%	100.0%	No OMIM phenotype
NDUFA8	135,1	100.0%	99.3%	No OMIM phenotype Complex I deficiency (Bugiani (2004) Biochim Biophys Acta 1659,136)
NDUFA9	101,6	99.7%	96.5%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	131,6	98.9%	93.6%	No OMIM phenotype
NDUFAF1	98,5	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	54,1	94.3%	82.0%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	141	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	98,3	99.2%	94.5%	Mitochondrial complex I deficiency, nuclear type 15, 618237

NDUFAF5	124,9	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	91,9	99.8%	98.5%	Mitochondrial complex I deficiency, nuclear type 17, 618239
NDUFAF7	96,6	100.0%	99.8%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFAF8	46,1	77.7%	63.6%	No OMIM phenotype
NDUFB1	41,7	61.2%	52.1%	No OMIM phenotype ?Complex I deficiency (Calvo (2012) Nat Genet 42,851)
NDUFB10	146,6	99.5%	95.9%	No OMIM phenotype Complex I deficiency (Friederich (2016) Hum Mol Genet)
NDUFB11	103,3	98.6%	95.0%	?Mitochondrial complex I deficiency, nuclear type 30, 301021 Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB2	90,3	100.0%	100.0%	No OMIM phenotype
NDUFB3	23,3	89.7%	62.5%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	117,5	86.9%	83.9%	No OMIM phenotype
NDUFB5	102,3	100.0%	100.0%	No OMIM phenotype
NDUFB6	48,3	98.6%	90.5%	No OMIM phenotype
NDUFB7	86,1	100.0%	99.5%	No OMIM phenotype
NDUFB8	105,3	100.0%	99.8%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	105,2	97.8%	93.3%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	102,5	100.0%	99.2%	No OMIM phenotype
NDUFC2	51,8	98.7%	93.2%	No OMIM phenotype {Insulin secrection,association with} (Olsson (2011) Eur J Endocrinol 164,765)
NDUFS1	143,5	99.9%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100,1	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	124,8	90.7%	90.5%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	144,5	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	125,7	100.0%	100.0%	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFS6	111,9	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	140,5	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	156,8	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	141,7	99.9%	98.8%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	74,2	92.4%	77.3%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	132,4	100.0%	99.7%	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	68,6	89.3%	87.6%	No OMIM phenotype

				Mitochondrial complex II/III deficienc,infantile (Farhan (2014) Mol Genet Genomic Med 2,73)
NFU1	61,8	97.4%	82.1%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NME3	146,5	99.6%	94.9%	No OMIM phenotype
NR2F1	222,3	100.0%	100.0%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NSUN3	170,4	100.0%	100.0%	No OMIM phenotype Mitochondrial disease (Van Haute (2016) Nat Commun 7)
NUBPL	102	98.9%	95.5%	Mitochondrial complex I deficiency, nuclear type 21, 618242
OGDH	172,3	99.9%	99.4%	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OPA1	124,7	99.7%	97.4%	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
OPA3	156,6	100.0%	99.2%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OXA1L	137,6	100.0%	99.9%	No OMIM phenotype
PANK2	154,1	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PARS2	177,9	100.0%	100.0%	No OMIM phenotype Alpers syndrome (Sofou (2015) Mol Genet Genomic Med 3,59)
PC	155,4	99.9%	98.7%	Pyruvate carboxylase deficiency, 266150
PDHA1	85,3	98.9%	95.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	111,4	99.2%	97.2%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	129	99.9%	99.5%	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	131,1	99.8%	99.0%	No OMIM phenotype
PDK2	155,5	100.0%	100.0%	No OMIM phenotype
PDK3	108,4	97.4%	94.5%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	114,5	100.0%	99.4%	No OMIM phenotype ?Autism spectrum disorder (Matsunami (2014) Mol Autism 5,5)
PDP1	129,1	100.0%	100.0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	104,8	96.7%	87.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	112,9	99.6%	96.1%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	87,9	98.0%	87.6%	Mitochondrial complex IV deficiency, 220110
PET117	106,9	100.0%	100.0%	No OMIM phenotype
PIGA	70,9	92.9%	84.0%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PISD	160,3	100.0%	99.9%	No OMIM phenotype

PITRM1	102,5	97.4%	94.9%	Brunetti et al, EMBO Mol Med 2015
PLA2G6	111,9	99.8%	98.2%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLPBP	95,3	99.6%	95.3%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMPCA	108,1	99.1%	95.9%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121,6	100.0%	99.2%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNPLA8	121,2	100.0%	99.7%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPT1	56,1	96.2%	84.3%	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POLG	113,9	100.0%	99.6%	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	183,7	99.6%	98.0%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
PPA2	92,3	98.8%	91.7%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	148,5	100.0%	99.3%	Cardiomyopathy, dilated, 2C, 618189
PRKAA1	138,2	99.8%	99.4%	No OMIM phenotype
PTCD3	81,1	99.0%	96.0%	No OMIM phenotype
PTRH2	200,8	100.0%	100.0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	113,3	99.8%	97.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	96	99.7%	97.4%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	116,5	99.7%	96.9%	Leukodystrophy, hypomyelinating, 10, 616420
PYROXD1	48,6	93.0%	78.5%	Myopathy, myofibrillar, 8, 617258
QRSL1	85,2	98.6%	93.1%	No OMIM phenotype Infantile mitochondrial disorder, lethal (Kohda (2016) PLoS Genet 12, e1005679)
RARS2	104	100.0%	99.4%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	132,6	100.0%	99.0%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	101,1	97.6%	92.5%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RRM1	119,7	99.9%	99.4%	No OMIM phenotype
RRM2B	143,9	99.9%	99.4%	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	79,6	100.0%	98.0%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732

RYR1	117,1	98.7%	95.7%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 {Malignant hyperthermia susceptibility 1}, 145600
SACS	150,4	100.0%	99.9%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	133,4	99.8%	98.5%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS2	117,9	95.1%	93.2%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	100,1	99.8%	98.1%	Mitochondrial complex IV deficiency, 220110
SCO2	115,7	100.0%	99.9%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SCP2	107,8	99.7%	96.4%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDHA	88,9	85.1%	77.7%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	83	100.0%	100.0%	Mitochondrial complex II deficiency, 252011
SDHB	114,8	100.0%	99.9%	Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHD	43,7	52.7%	50.6%	Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SERAC1	111	99.7%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	124	100.0%	99.7%	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	101,3	100.0%	99.6%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	134,6	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	103,2	99.3%	95.1%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A10	83,1	81.2%	76.7%	No OMIM phenotype
SLC25A12	150,9	99.9%	99.9%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	120,1	99.8%	98.9%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	77,4	99.9%	97.8%	Microcephaly, Amish type, 607196

				Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A21	122,3	99.9%	99.4%	No OMIM phenotype ?Synpolydactyly (Meyertholen (2012) Mol Syndromol 3 25)
SLC25A22	123,8	100.0%	99.1%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	128,9	99.6%	99.1%	Fontaine progeroid syndrome, 612289
SLC25A3	129,8	99.5%	96.9%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	128,2	100.0%	99.9%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	94,5	99.1%	95.2%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	130,9	100.0%	99.9%	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	130	99.9%	98.5%	No OMIM phenotype Mitochondrial myopathy (Shamseldin (2016) Hum Genet 135,21)
SLC25A46	173	99.8%	98.3%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC39A8	140,9	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC52A2	185,4	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	118,8	100.0%	99.8%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SPART	132,6	99.8%	98.2%	Troyer syndrome, 275900
SPATA5	139,5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG7	115,2	99.3%	96.4%	Spastic paraplegia 7, autosomal recessive, 607259
SQSTM1	117,8	99.9%	99.2%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Paget disease of bone 3, 167250
STAC3	114,7	100.0%	100.0%	Myopathy, congenital, Baily-Bloch, 255995
STAT2	110	100.0%	99.8%	Immunodeficiency 44, 616636
STXBP1	103,7	96.8%	96.4%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	58,8	91.7%	82.6%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	102,9	99.9%	99.6%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	58,1	91.8%	79.3%	No OMIM phenotype ?Methylmalonic aciduria (Chu (2016) Mol Genet Metab 118, 264)
SURF1	84,8	91.3%	88.4%	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SZT2	135,6	99.6%	99.4%	Epileptic encephalopathy, early infantile, 18, 615476
TACO1	93,9	99.5%	95.3%	Mitochondrial complex IV deficiency, 220110
TANGO2	127,3	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration,

				616878
TARS2	89	99.7%	96.4%	?Combined oxidative phosphorylation deficiency 21, 615918
TAZ	114,5	99.3%	95.8%	Barth syndrome, 302060
TDP2	173	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TFB2M	76,6	99.8%	97.1%	No OMIM phenotype
THG1L	134,7	100.0%	100.0%	No OMIM phenotype Cerebelar ataxia and developmental delay (Edvardson (2016) Neurogenetics, epub)
TIMM22	103,7	100.0%	99.5%	No OMIM phenotype
TIMM44	157,1	100.0%	99.8%	No OMIM phenotype Oncocytic thyroid carcinoma (Bonora (2006) Br J Cancer 95,1529)
TIMM50	122,9	99.9%	98.7%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	46,3	94.6%	79.9%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	161,4	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TK2	103,8	100.0%	99.2%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM126A	104,8	96.2%	82.8%	Optic atrophy 7, 612989
TMEM126B	87,8	99.5%	95.9%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM186	121,9	100.0%	100.0%	No OMIM phenotype
TMEM65	70,6	90.6%	84.5%	No OMIM phenotype
TMEM70	117,9	99.8%	97.6%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMX2	116,5	99.9%	98.4%	No OMIM phenotype
TOP3A	121	99.8%	97.5%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TPK1	94	100.0%	98.7%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRAPPC2L	198,9	100.0%	100.0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRIT1	107,9	100.0%	99.9%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT10C	138,5	100.0%	100.0%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	175,7	99.8%	98.7%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	100	100.0%	99.4%	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	101,5	99.2%	96.5%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
TSFM	120	100.0%	99.2%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	83,4	97.0%	82.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	130,6	100.0%	99.2%	Combined oxidative phosphorylation deficiency 4, 610678
TWNK	159,6	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138

				Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
TXN2	61,4	100.0%	99.8%	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	120,9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC1	93,8	100.0%	99.9%	No OMIM phenotype
UQCC2	132,2	100.0%	98.1%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	123	100.0%	99.9%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	173,6	100.0%	100.0%	No OMIM phenotype
UQCR11	212,1	100.0%	100.0%	No OMIM phenotype
UQCRB	106	99.2%	95.4%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	127,3	99.6%	97.7%	No OMIM phenotype
UQCRC2	105,3	99.6%	97.2%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	118,8	96.6%	90.8%	No OMIM phenotype
UQCRH	109,9	99.5%	93.8%	No OMIM phenotype
UQCRQ	158,9	100.0%	100.0%	Mitochondrial complex III deficiency, nuclear type 4, 615159
USMG5	NC	NC	NC	No OMIM phenotype
VARS2	120,1	100.0%	99.8%	Combined oxidative phosphorylation deficiency 20, 615917
VPS13D	138	100.0%	99.7%	Spinocerebellar ataxia, autosomal recessive 4, 607317
WARS2	132,3	99.9%	99.1%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WDR45	68,7	96.8%	88.9%	Neurodegeneration with brain iron accumulation 5, 300894
YARS2	175,2	99.9%	99.6%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	103,9	98.2%	93.5%	?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 : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors