MITOCHONDRIAL DISORDERS GENE PANEL DG 2.15 (385 genes)

Releasedate: 31-01-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AARS2	126.2	100	99.3	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABAT	92.7	100	99.5	GABA-transaminase deficiency, 613163
ACAD9	135.2	98.4	95.7	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACO2	129.3	95.8	91.8	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ADAMTS10	107.8	99.9	98.7	Weill-Marchesani syndrome 1, recessive, 277600
ADPRHL2	163.7	100	99.9	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	121	91.9	84.9	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGK	112.1	99.3	96.4	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AIFM1	106.2	100	99.7	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)
ALKBH1	113.8	100	99.4	No OMIM phenotype Arumugam et al ESHG 2018
ANO10	116.7	98.8	96.5	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOPT1	63.8	81.4	78.1	Mitochondrial complex IV deficiency, 220110
APTX	118.9	94.2	91.1	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ATAD1	59.1	94.9	86.4	Hyperekplexia 4, 618011
ATAD3A	87.9	89	86.2	Harel-Yoon syndrome, 617183
ATAD3B	83.8	88.6	82.8	No OMIM phenotype Late-onset encephalopathy with cerebellar atrophy, ataxia and dystonia (Desai (2017) Brain 140,1595)
ATP13A2	117.4	100	98.8	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225

ATP5A1	85.3	94.8	85.8	?Combined oxidative phosphorylation deficiency 22, 616045 ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228
ATP5B	129.2	100	99.8	No OMIM phenotype
ATP5C1	90.1	95.4	84.9	No OMIM phenotype
ATP5D	66.1	98.8	90.3	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	135.5	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	80.9	96.9	84.5	No OMIM phenotype
ATP5G1	110	100	98.2	No OMIM phenotype
ATP5G2	93.8	100	98.4	No OMIM phenotype
ATP5G3	118.5	100	100	No OMIM phenotype
ATP5H	109.7	93	71.8	No OMIM phenotype
ATP5I	69.6	99.9	97	No OMIM phenotype
ATP5J	66.4	99	90.6	No OMIM phenotype
ATP5J2	109.7	100	99.9	No OMIM phenotype
ATP5L	144.6	100	99.9	No OMIM phenotype
ATP5L2	217.8	100	100	No OMIM phenotype
ATP5O	101.3	99.1	90.9	No OMIM phenotype
ATP5S	133.6	100	100	No OMIM phenotype Complex V deficiency
ATPAF1	79.8	74	68.1	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	99.9	No OMIM phenotype
BOLA2	120.5	100	100	No OMIM phenotype ?Autism and developmental delay (Nuttle (2016) Nature 536, 205)
BOLA3	50.1	92.3	81.7	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
C12orf65	88.2	97.3	91.9	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	93.9	100	99.7	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298

C19orf70	63	100	98.8	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	81.7	71	Combined oxidative phosphorylation deficiency 33, 617713
CA5A	124.1	99.5	94.9	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CARS2	121.1	100	99.8	Combined oxidative phosphorylation deficiency 27, 616672
CEP89	125.5	94.7	91.4	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	35.2	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048
CHCHD2	95.4	99.5	91.9	Parkinson disease 22, autosomal dominant, 616710
CHKB	98.5	100	99	Muscular dystrophy, congenital, megaconial type, 602541
CISD2	127.9	83.4	83.4	Wolfram syndrome 2, 604928
CLPB	140.2	100	99.5	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	115.4	99.8	96.9	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	85.6	84	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	78.7	98.8	91.9	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	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	89.8	88.4	84.9	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	99.3	96	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	158.5	99.7	98.9	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	134.3	100	99.1	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	90.5	100	99.1	Nephrotic syndrome, type 9, 615573

COQ9	91.4	99.9	96.6	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	241.9	100	99.6	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	108.1	100	99.9	?Mitochondrial complex IV deficiency, 220110
COX15	98.6	100	99.7	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	58.1	83	65.4	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	80.2	57.7	No OMIM phenotype
COX5B	126.9	100	100	No OMIM phenotype
COX6A1	180.6	100	99.4	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	32.4	97.5	80.8	No OMIM phenotype
COX6B1	159.6	100	100	Mitochondrial complex IV deficiency, 220110
COX6B2	62.3	100	99.3	No OMIM phenotype
COX6C	131.4	99.2	90.9	No OMIM phenotype
COX7A1	81.7	99.9	98	No OMIM phenotype
COX7A2	82.9	99.3	92.7	No OMIM phenotype {insulin secretion,association with} (Olsson (2011) Eur J Endocrinol 164,765)
COX7B	47.9	73.3	42	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	247.3	100	100	No OMIM phenotype
COX7C	48.9	99	94.4	No OMIM phenotype
COX8A	98.1	100	100	?Mitochondrial complex IV deficiency, 220110
COX8C	159.2	99.9	97.9	No OMIM phenotype ?Tethered spinal cord syndrome (Zhao (2016) Neural Regen Res 11, 1333)
CP	120	93.9	89.6	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CTBP1	93.4	96.1	85	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CYC1	184.5	88.1	86.8	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	72.1	99.6	95	Thrombocytopenia 4, 612004
DARS2	122.3	100	99.6	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF17	91.9	95.6	89.3	Woodhouse-Sakati syndrome, 241080

DDHD1	141.8	97.1	94.8	Spastic paraplegia 28, autosomal recessive, 609340
DES	120.8	99.9	98.1	Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DGUOK	119.2	100	100	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 Portal hypertension, noncirrhotic, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070
DHTKD1	141	99.6	98.2	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	91.6	99.1	96	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	123.5	99.9	98.6	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	95.8	94.2	89.7	No OMIM phenotype ?Diaphragmatic hernia,congenital (Yu (2015) Hum Mol Genet 24,4764)
DMAC1	50.4	99.8	96.3	No OMIM phenotype Complex I assembly factor
DMAC2	135.1	98.3	98.3	No OMIM phenotype Complex I assembly factor
DNA2	123.6	99.8	96.9	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJC19	97.9	98.5	90	3-methylglutaconic aciduria, type V, 610198
DNAJC3	116.3	99.9	98.1	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	123.5	99.7	96.6	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
EARS2	103.4	99.7	98.3	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	112.8	99.8	97.8	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	141.4	99.7	98	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
EHHADH	163.2	100	99.7	?Fanconi renotubular syndrome 3, 615605
ELAC2	123.8	100	99.3	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	99.3	95.8	Ethylmalonic encephalopathy, 602473
FA2H	94.1	87.9	79.9	Spastic paraplegia 35, autosomal recessive, 612319

FARS2	207.7	100	100	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSB	76.6	96	93.4	?Neurodevelopmental disorder with brain, liver, and lung abnormalities, 618007
FASTKD2	118.9	99.5	96.8	?Mitochondrial complex IV deficiency, 220110
FBXL4	189.8	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX2	114.3	99.9	99.3	No OMIM phenotype Mitochondrial muscle myopathy (Spiegel (2014) Eur J Hum Genet 22,902)
FDXR	93.4	100	99.1	Auditory neuropathy and optic atrophy, 617717
FH	146.4	91.7	87.6	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	136.6	100	99.6	Mitochondrial complex I deficiency, nuclear type 19, 618241
FTL	147.7	99	93.2	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FXN	75.2	85.7	75.9	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GARS	125.7	99.9	98.5	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GATB	101.5	99.9	98.9	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	92.9	75.4	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	100.3	99.2	95.3	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	118.6	98.7	93.4	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,e100
GLRX5	108.2	92.6	83.8	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	74.5	94.4	84.3	Hyperinsulinism-hyperammonemia syndrome, 606762
GTPBP2	156.8	96.5	94.7	Jaberi-Elahi syndrome, 617988
GTPBP3	137.4	100	99.7	Combined oxidative phosphorylation deficiency 23, 616198
HARS2	169.7	99.9	99.2	?Perrault syndrome 2, 614926

HCCS	106.6	99.9	99.2	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	67.7	92.7	69.5	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	172.8	100	100	Holocarboxylase synthetase deficiency, 253270
HSD17B10	117.1	100	99.2	HSD10 mitochondrial disease, 300438
HSPA9	91.6	91.1	85.9	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
HSPD1	96.5	98.3	93.2	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HTRA2	122.1	100	99.7	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297
IARS2	131.5	100	99.9	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	113.3	93.3	89.5	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ISCA1	50.8	92.5	80.5	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	92	99.7	96.9	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	111.2	100	99.7	Myopathy with lactic acidosis, hereditary, 255125
KARS	122.6	100	99.3	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KIF1A	114	99.2	96.1	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
LACTB	117.2	93	80.8	No OMIM phenotype
LARS2	143	100	100	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300
LIAS	133.7	99.5	97.1	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT1	227.4	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	92.2	97.3	83.2	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LONP1	141.5	97.9	96.4	CODAS syndrome, 600373
LRPPRC	127.3	99.4	97.2	Leigh syndrome, French-Canadian type, 220111
LYRM4	60.1	63.2	54.3	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	49	87.6	72.4	Mitochondrial complex III deficiency, nuclear type 8, 615838
MARS2	173.2	100	100	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390

MCUR1	65.3	93.5	77.4	No OMIM phenotype
MDH2	123.3	98	97.9	Epileptic encephalopathy, early infantile, 51, 617339
MECR	108.1	98.8	96.1	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	93.7	90.4	87.6	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	150.6	100	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	151.1	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MICU1	134.2	96	88.8	Myopathy with extrapyramidal signs, 615673
MICU2	43	94.5	86	No OMIM phenotype Shamseldin et al,Brain 2017
MIEF2	122.9	100	99.9	No OMIM phenotype mitochondrial medicine meeting 2016,Cambridge
MIPEP	102.1	95.3	88.6	Combined oxidative phosphorylation deficiency 31, 617228
MPC1	121.8	100	99.5	Mitochondrial pyruvate carrier deficiency, 614741
MPV17	108.5	100	99.4	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRM2	122.9	99.7	96.9	No OMIM phenotype Encephalomyopathy, childhood-onset and stroke-like episodes (Garone (2017) Hum Mol Genet 26,4257)
MRPL12	107.3	99.4	93.5	No OMIM phenotype Growth retardation and neurological deterioration (Serre (2013) Biochim Biophys Acta 1832)
MRPL3	66.3	91.2	77.9	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	101.6	99.8	96.5	No OMIM phenotype Paper Rotig
MRPL44	110.5	99.7	97.6	?Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	155	100	99.5	No OMIM phenotype SSIEM 2016
MRPS14	215.9	100	100	No OMIM phenotype Jackson et al ESHG 2018
MRPS16	161.1	100	99.1	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	166.4	99.7	97.9	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	138.8	95.3	91.8	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS23	156.7	100	99.7	No OMIM phenotype Kohda et al,PLoS Genet 2016

MRPS28	122.3	87.9	86.2	No OMIM phenotype Pulman et alHum Mol Gen 2018
MRPS34	132.7	99.9	98.3	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	99.8	97	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	124.6	99.3	96.2	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTO1	173.7	89.5	87.3	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	109.6	98.9	93.5	?Spastic ataxia 4, autosomal recessive, 613672
NARS2	120	97.4	97.1	Combined oxidative phosphorylation deficiency 24, 616239
NAXD	138.7	99.9	99	No OMIM phenotype van Bergen et al Brain 2019 142 50-58
NAXE	81.1	99.7	95.9	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFA1	166.8	100	99.6	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	136.7	98.9	96.8	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	86.9	99.5	95.8	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	160.2	100	100	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	91.4	92.3	91.6	?Mitochondrial complex I deficiency, nuclear type 28, 618249 {Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	133.9	100	100	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	129.7	91.4	87.4	No OMIM phenotype
NDUFA4	79.9	98.8	84.7	No OMIM phenotype Cytochrome c oxidase deficiency (Pitceathly (2013) Cell Rep 3,1795) ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA5	71	87.1	61.3	No OMIM phenotype
NDUFA6	212.4	100	100	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	114.8	100	99.5	No OMIM phenotype
NDUFA8	138.4	100	99.7	No OMIM phenotype Complex I deficiency (Bugiani (2004) Biochim Biophys Acta 1659,136)
NDUFA9	124.7	98.6	93.2	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	116.3	99.9	96.2	No OMIM phenotype
NDUFAF1	115.6	100	100	Mitochondrial complex I deficiency, nuclear type 11, 618234

NDUFAF2	58.6	85.7	70.9	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	120.8	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	79.4	98.9	91.8	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	95.7	98.8	94.5	Mitochondrial complex I deficiency, nuclear type 16, 616238
NDUFAF6	79.3	97.1	85.6	Mitochondrial complex I deficiency, nuclear type 17, 612392
NDUFAF7	101.2	100	98.8	No OMIM phenotype ?Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFAF8	30	60.2	47.4	No OMIM phenotype Alston et al ESHG 2018
NDUFB1	29.3	60.5	53	No OMIM phenotype ?Complex I deficiency (Calvo (2012) Nat Genet 42,851)
NDUFB10	120.2	99.4	95.8	No OMIM phenotype Complex I deficiency (Friederich (2016) Hum Mol Genet)
NDUFB11	109.6	94.4	88	?Mitochondrial complex I deficiency, nuclear type 30, 301021 Linear skin defects with multiple congenital anomalies 3, 300952
NDUFB2	102.4	100	100	No OMIM phenotype
NDUFB3	22.6	91.9	59.2	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	100.6	85	82.9	No OMIM phenotype
NDUFB5	88.5	100	100	No OMIM phenotype
NDUFB6	39.7	99.9	91.4	No OMIM phenotype
NDUFB7	50.4	100	97.4	No OMIM phenotype
NDUFB8	116.6	100	100	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	120.1	99.8	97.4	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	87.2	100	98.3	No OMIM phenotype
NDUFC2	39.9	98	84	No OMIM phenotype
NDUFS1	132.2	99.8	98.6	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	117.8	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	142.4	90.7	90.6	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	147.3	100	99.1	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	196.7	100	100	No OMIM phenotype
NDUFS6	119.1	99.9	99.4	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	118.4	100	99.7	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	141.4	100	99.9	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	136.7	99.7	97.8	Mitochondrial complex I deficiency, nuclear type 4, 618225

NDUFV2	69.5	78.7	53.9	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	119.3	99.9	98.8	No OMIM phenotype
NFS1	82.1	86.4	83.5	No OMIM phenotype
NFU1	47.7	94.9	77.2	Multiple mitochondrial dysfunctions syndrome 1, 605711
NR2F1	201.6	99.9	98.4	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NSUN3	187.1	100	100	No OMIM phenotype
NUBPL	89.8	92.9	85.9	Mitochondrial complex I deficiency, nuclear type 21, 618242
OGDH	201.3	100	100	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OPA1	122.5	99.1	94.1	?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	128	99.5	97.4	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OXA1L	162	100	100	No OMIM phenotype
PANK2	146.6	99.3	93.1	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PARS2	219.1	100	100	No OMIM phenotype
PC	149.3	97.7	94.6	Pyruvate carboxylase deficiency, 266150
PDHA1	109.8	98.1	92.1	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	133	99.3	96.8	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	132.5	98.9	94.6	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	127.4	97.7	94	No OMIM phenotype
PDK2	156.8	100	100	No OMIM phenotype
PDK3	105.1	96.4	94.3	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	110.1	99.8	97.4	No OMIM phenotype
PDP1	209.6	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	116.7	88.8	78.7	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	126.8	96.5	93.5	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	94.5	88.8	74.8	Mitochondrial complex IV deficiency, 220110
PET117	95.3	100	99.8	No OMIM phenotype
PIGA	90.5	90.4	81.3	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818

PISD	163	100	100	No OMIM phenotype Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function (PMID: 30488656)
PITRM1	117.7	97.5	95.7	No OMIM phenotype
PLA2G6	117.5	99.9	98.4	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PMPCA	120.8	99.4	96.8	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121	99.7	97.8	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNPLA8	114.4	100	99.7	?Mitochondrial myopathy with lactic acidosis, 251950
PNPT1	53.7	93.3	80.9	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934
POLG	114.4	100	99.5	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	98.8	96.8	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
PPA2	80.4	94.6	82.5	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	108.7	99.9	98.4	Cardiomyopathy, dilated, 2C, 618189
PRKAA1	119.9	100	99.3	No OMIM phenotype
PTCD3	93.7	97	93.7	No OMIM phenotype
PTRH2	279.6	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	127.2	98.6	93.9	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	86.3	99.4	94.3	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	127.6	100	97.6	Leukodystrophy, hypomyelinating, 10, 616420
PYROXD1	48.4	85.5	70.6	Myopathy, myofibrillar, 8, 617258
QRSL1	96.7	98.7	93.8	No OMIM phenotype
RARS2	107.2	100	99.1	Pontocerebellar hypoplasia, type 6, 611523
RMND1	137.2	99.8	97.3	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	98.6	99.1	95.6	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479

RRM1	132.4	99.8	98.6	No OMIM phenotype
RRM2B	128.6	99.7	97.5	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	99.9	99.1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
SACS	154.5	100	99.7	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	127.9	99.6	96.6	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS2	104.8	94.8	92.7	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	109.6	97.9	94.3	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	99.6	96.5	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDHA	122.2	84.8	80.8	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	42.5	100	96.2	Mitochondrial complex II deficiency, 252011
SDHB	120.3	100	99.3	Gastrointestinal stromal tumor, 606444 Paragangliolas 4, 115310 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SDHD	48.4	55.2	50.4	Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300
SERAC1	112.5	98.8	94.6	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	131.7	100	99.1	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	119.5	99.8	97.8	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	186.4	100	99.9	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	71	92.2	87	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182

SLC25A10	70.2	76.5	68.6	No OMIM phenotype
SLC25A12	150.5	99.8	98.4	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	110.7	95.7	92.3	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	88.6	99.9	98.3	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A21	114	100	98.9	No OMIM phenotype ?Synpolydactyly (Meyertholen (2012) Mol Syndromol 3 25)
SLC25A22	108.7	99.5	96.9	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	115.8	98.6	96.3	Fontaine progeroid syndrome, 612289
SLC25A3	139	99.8	97.6	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	117	100	100	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	111.4	99.8	98.1	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
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	97.8	94.2	No OMIM phenotype Mitochondrial myopathy (Shamseldin (2016) Hum Genet 135,21)
SLC25A46	205.7	95.9	87.3	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC39A8	128.5	100	99.7	Congenital disorder of glycosylation, type IIn, 616721
SLC52A2	177.6	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	119.6	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SPART	132.4	99.8	98.2	Troyer syndrome, 275900
SPATA5	132	99.9	99.2	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG7	119.2	93.3	92.4	Spastic paraplegia 7, autosomal recessive, 607259
SQSTM1	109.1	98.6	94.5	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
STAT2	116	100	99.9	Immunodeficiency 44, 616636
STXBP1	124.5	96.8	96.8	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	64.9	93.3	82.8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic

				aciduria), 612073
SUCLG1	101.3	99.6	95.4	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	57.8	91.1	78.7	No OMIM phenotype ?Methylmalonic aciduria (Chu (2016) Mol Genet Metab 118, 264)
SURF1	96.2	88.3	88.3	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SZT2	149.5	99.5	99.2	Epileptic encephalopathy, early infantile, 18, 615476
TACO1	91.7	97	92.6	Mitochondrial complex IV deficiency, 220110
TANGO2	145.3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TARS2	98.8	99.9	98.5	?Combined oxidative phosphorylation deficiency 21, 615918
TAZ	94	99.9	98.8	Barth syndrome, 302060
TDP2	165.1	99.9	98.8	Spinocerebellar ataxia, autosomal recessive 23, 616949
TFB2M	64.4	99.2	94.1	No OMIM phenotype
THG1L	142.9	100	99.9	No OMIM phenotype
TIMM22	95.4	100	99.9	No OMIM phenotype
TIMM44	123.3	100	98.5	No OMIM phenotype
TIMM50	108.2	98.8	95.3	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	46	94.5	78.8	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	152.2	100	100	Mitochondrial complex I deficiency, nuclear type 31, 618251
TK2	105.7	93.4	89.4	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM126A	120.3	98.4	86.2	Optic atrophy 7, 612989
TMEM126B	79.2	99.8	97.7	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM186	152.4	100	100	No OMIM phenotype
TMEM65	50.2	79.2	65.9	No OMIM phenotype
TMEM70	138.7	94.6	90.3	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMX2	141	100	99.5	No OMIM phenotype
TOP3A	129.8	98.9	96.5	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TPK1	112.7	99.8	97.3	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458

TRAPPC2L	230.2	100	100	No OMIM phenotype
TRIT1	119.4	100	99.8	Combined oxidative phosphorylation deficiency 35, 617873
TRMT10C	131.4	99.8	98.8	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	208.7	99.2	93.9	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	99	100	99.6	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	104.6	97.8	92.3	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	80.6	72.5	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	135.4	100	99.7	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	98.3	85	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC1	96.7	100	100	No OMIM phenotype
UQCC2	96.6	100	99.1	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	95	100	99.2	?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	99.6	96.8	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	151.4	100	99.8	No OMIM phenotype
UQCRC2	122.6	99.9	99.1	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	151.9	87	82.6	No OMIM phenotype
UQCRH	131.1	99.9	98.3	No OMIM phenotype
UQCRQ	131.3	100	99.9	Mitochondrial complex III deficiency, nuclear type 4, 615159
USMG5	16.2	76.5	27.7	No OMIM phenotype
VARS2	110.9	99.9	98.9	Combined oxidative phosphorylation deficiency 20, 615917
VPS13D	158.6	99.9	99.4	Spinocerebellar ataxia, autosomal recessive 4, 607317
WARS2	140.7	100	99.5	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WDR45	75	97.4	90.1	Neurodegeneration with brain iron accumulation 5, 300894

YARS2	173.2	99.8	98.9	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	105.3	97.7	91.9	?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 : December 31st, 2018.
This list is accurate for panel version DG 2.15
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors