MITOCHONDRIAL DISORDERS GENE PANEL DG 2.5/2.6

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM disease ID
AARS2	111	99%	97%	Combined oxidative phosphorylation deficiency 8, 614096
ABAT	86.7	100%	97%	Besse et al, Cell Metab 2015
ACAD9	137.4	99%	97%	ACAD9 deficiency, 611126
ACO2	111.4	95%	89%	Infantile cerebellar-retinal degeneration, 614559
ADCK3	114.9	99%	97%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCK4	90.9	100%	100%	Nephrotic syndrome type 9, 615573
AFG3L2	108.2	90%	85%	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487
AGK	115.1	99%	94%	Hyperoxaluria, primary, type 1, 259900
AIFM1	76.8	100%	100%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
ALDH1B1	173.8	100%	100%	No OMIM phenotype Bladder cancer (Nickerson (2014) Clin Cancer Res 20,4935)
ANO10	108.7	96%	95%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOPT1	62.9	87%	82%	Mitochondrial complex IV deficiency,220110
APTX	122.4	92%	86%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ATAD3A	71.7	89%	85%	No OMIM phenotype
ATAD3B	69	85%	66%	No OMIM phenotype
ATP5A1	68.2	97%	84%	?Combined oxidative phosphorylation deficiency 22,616045 ?Mitochondrial complex (ATP synthase) deficiency,nuclear type 4,615228
ATP5B	117.4	100%	98%	No OMIM phenotype
ATP5C1	85.5	94%	76%	No OMIM phenotype
ATP5D	50.7	99%	89%	No OMIM phenotype
ATP5E	167.3	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	68.3	97%	82%	No OMIM phenotype
ATP5G1	88.2	100%	88%	No OMIM phenotype
ATP5G2	61.7	100%	95%	No OMIM phenotype
ATP5G3	112.6	100%	100%	No OMIM phenotype
ATP5H	93.2	65%	60%	No OMIM phenotype

ATP5I	71.7	100%	100%	No OMIM phenotype
ATP5J	54.9	100%	89%	No OMIM phenotype
ATP5J2	91.1	100%	97%	No OMIM phenotype
ATP5L	136	100%	100%	No OMIM phenotype
ATP5L2	149.9	100%	100%	No OMIM phenotype
ATP5O	108.3	99%	96%	No OMIM phenotype
ATP5S	117.5	100%	100%	No OMIM phenotype
ATPAF1	88	82%	68%	No OMIM phenotype
ATPAF2	83.3	100%	98%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATPIF1	173.6	100%	100%	No OMIM phenotype
BCS1L	147	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BOLA1	81	100%	100%	No OMIM phenotype
BOLA2	78.2	100%	100%	No OMIM phenotype
BOLA3	55.4	91%	83%	Multiple mitochondrial dysfunctions syndrome 2, 614299
C10orf2	154	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type),271245 Perrault syndrome 5,616138 Progressive external ophthalmoplegia with mitochondrial DNA depletions, autosomal dominant,609286
C11orf83	93.9	100%	96%	?Mitochondrial complex III deficiency,nuclear type 9,616111
C12orf65	74.3	99%	97%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55,autosomal recessive, 615035
C19orf12	78.4	100%	96%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
CARS2	97	100%	99%	Combined oxidative phosphorylation deficiency 27,616672
CEP89	132.5	100%	98%	No OMIM phenotype Intellectual disability (Vulto-van Silfhout (2013) Hum Mutat 34,1679) Complex IV deficiency,isolated (van Bon (2013) Hum Mol Genet 22,3138)
CHCHD10	18.8	42%	35%	?Myopathy,isolated mitochondrial,autosomal dominant,616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2,615911 Spinal muscular atrophy,Jokela type,615048
CHKB	82.4	98%	90%	Muscular dystrophy, congenital, megaconial type, 602541
CLPB	132.8	96%	96%	3-methylglutaconic aciduria,type VII,with cataracts,neurologic involvement and neutropenia,616271

CLPP	106.7	98%	92%	Perrault syndrome 3, 614129
COA1	68.9	100%	100%	No OMIM phenotype
COA3	122.9	100%	100%	No OMIM phenotype Neuropathy,exercise intolerance,obesity and short stature (Ostergaard (2015) J Med Genet 52,203
COA5	42.1	85%	85%	Mitochondrial complex IV deficiency, 220110
COA6	53	94%	78%	Cardioencephalomyopathy,fatal infantile,due to cytochrome c oxidase deficiency 4,616501
COASY	126.7	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COQ2	71.1	97%	91%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	80.5	84%	81%	Coenzyme Q10 deficiency,primary,7,616276
COQ6	127.8	98%	96%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ9	90.2	100%	97%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	202	100%	98%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	151.3	100%	100%	Mitochondrial complex IV deficiency, 220110
COX15	94.5	100%	99%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	44.1	95%	79%	Mitochondrial complex IV deficiency, 220110
COX4I1	125.8	100%	100%	No OMIM phenotype Schizophrenia (Fromer (2014) Nature 506,179)
COX4I2	93.5	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis,612714
COX5A	38.4	74%	48%	No OMIM phenotype
COX5B	127.1	100%	100%	No OMIM phenotype
COX6A1	160.3	100%	100%	Charcot-Marie-Tooth disease,recessive intermediate D,616039
COX6A2	45.3	99%	99%	No OMIM phenotype
COX6B1	139.4	100%	100%	?Mitochondrial complex IV deficiency,220110
COX6B2	51.7	100%	99%	No OMIM phenotype
COX6C	128.3	99%	99%	No OMIM phenotype
COX7A1	99.4	100%	99%	No OMIM phenotype
COX7A2	76.8	99%	86%	No OMIM phenotype insulin secretion,association with (Olsson (2011) Eur J Endocrinol 164,765)
COX7B	41.8	79%	54%	Linear skin defects with multiple congenital anomalies,300887
COX7B2	274.2	100%	100%	No OMIM phenotype
COX7C	68.5	99%	96%	No OMIM phenotype
COX8A	78.9	100%	100%	?Mitochondrial complex IV deficiency,220110

COX8C	156.9	99%	89%	No OMIM phenotype
CYC1	164.2	97%	86%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	68.2	99%	98%	Thrombocytopenia 4, 612004
DARS2	117.7	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DES	108.8	100%	99%	?Muscular dystrophy, limb-girdle, type 2R, 615325 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400
DGUOK	103.6	100%	97%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	139.3	98%	97%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	85.7	99%	96%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	129.1	98%	98%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	87.6	94%	91%	No OMIM phenotype Diaphragmatic hernia,congenital (Yu (2015) Hum Mol Genet 24,4764)
DNA2	128.6	99%	96%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6, 615156
DNAJC19	96.9	99%	88%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	112.5	100%	100%	?Ataxia,combined cerebellar and peripheral,with hearing loss and diabetes mellitus,616192
DNM1L	109.2	100%	94%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
EARS2	89.3	99%	94%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	107.7	100%	95%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency,616277
ECSIT	114.2	99%	95%	No OMIM phenotype Complex I deficiency (Calvo (2010) Nat Genet 42,851)
ELAC2	113.9	100%	97%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 Combined oxidative phosphorylation deficiency 17, 615440
ETHE1	64.8	99%	88%	Ethylmalonic encephalopathy, 602473
FARS2	190	100%	99%	Combined oxidative phosphorylation deficiency 14, 614946
FASTKD2	125.1	99%	96%	?Mitochondrial complex IV deficiency,220110
FBXL4	204	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX1L	89.6	91%	90%	No OMIM phenotype Mitochondrial muscle myopathy (Spiegel (2014) Eur J Hum Genet 22,902)
FH	160.3	90%	87%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800

FOXRED1	131.8	100%	98%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency,252010
FXN	82	75%	75%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GATM	152.2	100%	100%	Cerebral creatine deficiency syndrome 3, 612718
GFER	69.7	98%	71%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	100.4	99%	95%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	124.6	98%	95%	No OMIM phenotype Atorvastatin sensitivity (Callegari (2012) PLoS Genet 8,e1002755) Leigh syndrome with arthrogryposis multiplex congenita (Fukumura (2015) J Hum Genet 60,509) Wolcott-Rallison syndrome (Dixon-Salazar (2012) Sci Transl Med 4,138ra78)
GLRX5	99.2	90%	84%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	71.2	88%	81%	Hyperinsulinism-hyperammonemia syndrome, 606762
GTPBP3	107.7	100%	98%	Combined oxidative phosphorylation deficiency 23,616198
HARS2	166.6	100%	100%	Perrault syndrome 2, 614926
HCCS	63.8	99%	92%	Microphthalmia, syndromic 7, 309801
HIBCH	75.9	93%	70%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	160.7	100%	100%	Holocarboxylase synthetase deficiency, 253270
HSD17B10	71.2	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked 17/31, microduplication, 300705 Mental retardation, X-linked syndromic 10, 300220
HSPD1	84.8	97%	89%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
IARS2	127.5	100%	99%	?Cataracts,growth hormone deficiency,sensory neuropathy,sensorineural hearing loss and skeletal dysplasia,616007
IBA57	89.8	91%	90%	?Multiple mitochondrial dysfunctions syndrome 3,615330 ?Spastic paraplegia 74,autosomal recessive,616451
ISCA2	73.3	94%	87%	Multiple mitochondrial dysfunctions syndrome 4,616370
ISCU	123.5	100%	99%	Myopathy with lactic acidosis, hereditary, 255125
KARS	116.5	100%	98%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness,autosomal recessive 89,613916
LACTB	119.4	97%	82%	No OMIM phenotype
LARS2	126.7	100%	100%	Perrault syndrome 4, 615300

LIAS	149.8	100%	98%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
LIPT1	210.5	100%	100%	Lipoyltransferase 1 deficiency,616299
LONP1	134.4	95%	89%	CODAS syndrome,600373
LRPPRC	130	98%	94%	Leigh syndrome, French-Canadian type, 220111
LYRM4	54.2	66%	63%	?Combined oxidative phosphorylation deficiency 19,615595
LYRM7	41.3	84%	61%	?Mitochondrial complex III deficiency,nuclear type 8,615838
MARS2	122.6	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MCUR1	52.7	66%	58%	No OMIM phenotype
MFF	87.5	91%	87%	No OMIM phenotype Mitochondrial ecephalomyopathy (Shamseldin (2012) J Med Genet 49,234)
MFN2	138	100%	100%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA,601152
MGME1	168.8	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MPC1	110.7	100%	98%	Mitochondrial pyruvate carrier deficiency,614741
MPV17	103	100%	100%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 -3
MRPL12	98.6	94%	89%	No OMIM phenotype Growth retardation and neurological deterioration (Serre (2013) Biochim Biophys Acta 1832)
MRPL3	61.9	88%	72%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	97.8	97%	94%	No OMIM phenotype paper in press (shoubridge)
MRPL44	99.1	96%	90%	?Combined oxidative phosphorylation deficiency 16,615395
MRPS16	119.4	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	139.6	100%	100%	No OMIM phenotype
MRPS22	129.9	93%	90%	Combined oxidative phosphorylation deficiency 5, 611719
MRPS7	160	100%	100%	No OMIM phenotype Sensorineural deafness,progressive hepatic and renal failure and lactic acidaemia (Menezes (2015) Hum Mol Genet 24,2297)
MRRF	182.6	100%	98%	No OMIM phenotype Complex I deficiency (Calvo (2010) Nat Genet 42,851)
MTFMT	130.8	99%	95%	Combined oxidative phosphorylation deficiency 15, 614947
MTO1	155.5	90%	88%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	115.1	99%	90%	Ataxia, spastic, 4, 613672
NARS2	125.8	97%	97%	Combined oxidative phosphorylation deficiency 24,616239
NDUFA1	138.7	100%	97%	Mitochondrial complex I deficiency, 252010

NDUFA10	137.9	97%	95%	Leigh syndrome, 256000
NDUFA11	74.8	98%	91%	Mitochondrial complex I deficiency, 252010
NDUFA12	138	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA13	100.7	95%	94%	{Thyroid carcinoma,Hurthle cell},607464
NDUFA2	110.5	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA3	110.9	88%	87%	No OMIM phenotype
NDUFA4	66	99%	96%	No OMIM phenotype Complex I deficiency (Calvo (2010) Nat Genet 42,851) Cytochrome c oxidase deficiency (Pitceathly (2013) Cell Rep 3,1795)
NDUFA5	78	85%	68%	No OMIM phenotype
NDUFA6	245.7	100%	100%	No OMIM phenotype Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFA7	115.5	100%	99%	No OMIM phenotype
NDUFA8	115.5	100%	97%	No OMIM phenotype Complex I deficiency (Bugiani (2004) Biochim Biophys Acta 1659,136)
NDUFA9	133	98%	96%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 -3
NDUFAB1	114.4	99%	94%	No OMIM phenotype
NDUFAF1	95.1	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFAF2	43	85%	47%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	106.1	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	80.8	99%	94%	Mitochondrial complex I deficiency, 252010
NDUFAF5	86.5	99%	91%	Mitochondrial complex I deficiency, 252010
NDUFAF6	83.1	100%	95%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF7	96.7	100%	100%	No OMIM phenotype Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFB1	19.5	55%	52%	No OMIM phenotype Complex I deficiency (Calvo (2012) Nat Genet 42,851)
NDUFB10	108.5	98%	96%	No OMIM phenotype
NDUFB11	57	92%	69%	Linear skin defects with multiple congenital anomalies,300952
NDUFB2	105.4	100%	100%	No OMIM phenotype
NDUFB3	26	96%	56%	Mitochondrial complex I deficiency, 252010
NDUFB4	85.6	85%	80%	No OMIM phenotype
NDUFB5	86.1	100%	100%	No OMIM phenotype

NDUFB6	28.1	97%	64%	No OMIM phenotype
NDUFB7	43.2	99%	90%	No OMIM phenotype
NDUFB8	90.3	100%	100%	No OMIM phenotype
NDUFB9	109.1	100%	98%	?Mitochondrial complex I deficiency,252010
NDUFC1	77.1	99%	99%	No OMIM phenotype
NDUFC2	50.3	98%	95%	No OMIM phenotype Insullin secrection,association with (Olsson (2011) Eur J Endocrinol 164,765)
NDUFS1	134.3	97%	97%	Mitochondrial complex I deficiency, 252010
NDUFS2	102.5	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS3	121.8	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	155.3	100%	99%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS5	148.9	100%	100%	No OMIM phenotype Complex I deficiency (Calvo (2010) Nat Genet 42,851)
NDUFS6	124.5	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS7	106.9	100%	99%	Leigh syndrome, 256000
NDUFS8	125.5	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	132.1	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFV2	60.6	83%	54%	Mitochondrial complex I deficiency, 252010
NDUFV3	90.9	100%	100%	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	66.9	85%	83%	No OMIM phenotype Mitochondrial complex II/III deficienc,infantile (Farhan (2014) Mol Genet Genomic Med 2,73)
NFU1	43.4	90%	75%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NUBPL	90.3	82%	82%	Mitochondrial complex I deficiency, 252010
OGDH	191.1	100%	100%	Alpha-ketoglutarate dehydrogenase deficiency,203740
OPA1	115.4	98%	89%	Optic atrophy 1, 165500
OPA3	89.2	99%	93%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OXA1L	149.1	100%	100%	No OMIM phenotype
PANK2	142.9	95%	86%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200

PARS2	198.7	100%	100%	No OMIM phenotype Alpers syndrome (Sofou (2015) Mol Genet Genomic Med 3,59)
PC	134.1	98%	94%	Pyruvate carboxylase deficiency, 266150
PDHA1	72.7	94%	82%	Leigh syndrome, X-linked, 308930 Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	127.9	97%	94%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	115.4	97%	92%	Lacticacidemia due to PDX1 deficiency,245349
PDK1	135.7	95%	92%	No OMIM phenotype
PDK2	153.7	100%	100%	No OMIM phenotype
PDK3	77.7	94%	94%	?Charcot-Marie-Tooth disease,X-linked dominant, 6,300905
PDK4	112	100%	96%	No OMIM phenotype Autism spectrum disorder (Matsunami (2014) Mol Autism 5,5)
PDP1	173.3	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	118.2	92%	83%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	109	97%	92%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	110.5	89%	72%	Mitochondrial complex IV deficiency, 220110
PET112	91.8	100%	97%	No OMIM phenotype
PIGA	56.5	76%	73%	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria,somatic,300818
PITRM1	110.8	96%	96%	Brunetti et al, EMBO Mol Med 2015
PLA2G6	108.7	100%	98%	Infantile neuroaxonal dystrophy 1,256600 Neurodegeneration with brain iron accumulation 2B,610217 Parkinson disease 14,autosomal recessive,612953
PMPCA	135.1	98%	92%	Choquet et al, Brain 2015
PNPT1	48.8	94%	77%	Combined oxidative phosphorylation deficiency 13, 614932
POLG	107.7	100%	98%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome, 607459 Progressive external ophthalmoplegia, autosomal dominant, 157640
POLG2	143.9	99%	97%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
PTRH2	274.1	100%	100%	infantile-onset multisystem neurologic,endocrine and pancreatic disease,616263
PUS1	130	99%	97%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PYCR1	80.4	100%	91%	Cutis laxa, autosomal recessive, type IIB, 612940

				Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	105.9	98%	94%	Leukodystrophy,hypomyelinating,10,616420
RARS2	111.7	100%	99%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	131.5	99%	90%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	94	95%	90%	Progressive external ophthalmoplegia with mitochondrial DNA deletions,autosomal recessive 2,616479
RRM2B	133	98%	96%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075
SARS2	92	96%	94%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	110.2	98%	94%	Mitochondrial complex IV deficiency,220110
SCO2	94.1	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDHA	96.8	81%	73%	Cardiomyopathy, dilated, 1GG, 613642 Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Paragangliomas 5, 614165
SDHAF1	36.9	100%	94%	Mitochondrial complex II deficiency, 252011
SDHB	124.7	100%	100%	Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764 Paraganglioma and gastric stromal sarcoma, 606864 Paragangliomas 4, 115310 Pheochromocytoma, 171300
SDHD	51.2	59%	59%	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	112.7	99%	94%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	132	100%	99%	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	115.7	97%	92%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	168.9	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	64.1	97%	88%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182

SLC25A12	149.7	99%	95%	Hypomyelination, global cerebral, 612949
SLC25A13	103.6	93%	90%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	60.5	100%	95%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A22	91.8	100%	97%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A3	137.6	97%	88%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A4	125.7	100%	100%	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283
SLC25A46	175.2	96%	94%	Neuropathy,hereditary motor and sensory,type VIB,616505
SPG7	109	93%	89%	Spastic paraplegia 7, autosomal recessive, 607259
STXBP1	124.2	100%	100%	Epileptic encephalopathy,early infantile,4,612164
SUCLA2	58	94%	76%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with/without methylmalonic aciduria), 612073
SUCLG1	94.7	100%	98%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	56.4	91%	82%	No OMIM phenotype
SURF1	87.3	88%	88%	Leigh syndrome, due to COX deficiency, 256000
TACO1	90.4	93%	90%	Mitochondrial complex IV deficiency,220110
TANGO2	133.1	99%	97%	Kremer et al, AJHG 2016
TARS2	82.6	99%	92%	?Combined oxidative phosphorylation deficiency 21,615918
TAZ	74.7	100%	99%	Barth syndrome, 302060
TIMM44	106.7	100%	97%	No OMIM phenotype Oncocytic thyroid carcinoma (Bonora (2006) Br J Cancer 95,1529)
TIMM8A	17.5	64%	30%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	141.8	100%	100%	No OMIM phenotype
TK2	99.5	93%	90%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM126A	108.6	99%	91%	Optic atrophy-7, 612989
TMEM126B	93	100%	98%	No OMIM phenotype
TMEM70	121.2	94%	90%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TPK1	117	100%	95%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRIT1	125.2	99%	97%	No OMIM phenotype
TRMU	91.6	100%	97%	{Deafness, mitochondrial, modifier of}, 580000 Liver failure, transient infantile, 613070

TRNT1	93.1	95%	91%	Sideroblastic anemia with B-cell immunodeficiency,periodic fevers, and developmental delay,616084
TSFM	117.4	100%	100%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	90.6	89%	82%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	119	100%	96%	Combined oxidative phosphorylation deficiency 4, 610678
TXN2	86.5	100%	100%	Holzerova et al, Brain 2015
TYMP	74.9	92%	69%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC1	92.4	100%	100%	No OMIM phenotype
UQCC2	90	100%	100%	?Mitochondrial complex III deficiency,nuclear type 7,615824
UQCR10	148.1	100%	100%	No OMIM phenotype
UQCR11	139.4	100%	100%	No OMIM phenotype
UQCRB	101.6	99%	96%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	122	100%	98%	No OMIM phenotype
UQCRC2	137.1	100%	99%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	121.8	86%	81%	No OMIM phenotype
UQCRH	111.6	100%	99%	No OMIM phenotype
UQCRQ	121.5	100%	100%	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	18.3	61%	36%	Combined oxidative phosphorylation deficiency 20,615917
YARS2	157.3	99%	98%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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