MITOCHONDRIAL DISORDERS GENE PANEL DG 2.4.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AARS2	94.8	99%	98%	Combined oxidative phosphorylation deficiency 8, 614096
ACAD9	85.4	100%	99%	ACAD9 deficiency, 611126
ACO2	85.4	93%	86%	Infantile cerebellar-retinal degeneration, 614559
ADCK3	101.2	100%	98%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCK4	71.4	100%	96%	Nephrotic syndrome type 9, 615573
AFG3L2	79.7	94%	91%	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487
AGK	110.3	99%	99%	Hyperoxaluria, primary, type 1, 259900
AIFM1	110.9	100%	97%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
ALDH1B1	114.3	100%	100%	No OMIM phenotype
ANO10	104	98%	96%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOPT1	85	87%	87%	Mitochondrial complex IV deficiency,220110
APTX	128.2	99%	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ATAD3A	42.3	48%	38%	No OMIM phenotype
ATAD3B	45	49%	45%	No OMIM phenotype
ATP5A1	52.9	92%	84%	?Combined oxidative phosphorylation deficiency 22,616045 ?Mitochondrial complex (ATP synthase) deficiency,nuclear type 4,615228
ATP5B	97	100%	100%	No OMIM phenotype
ATP5C1	64.8	94%	85%	No OMIM phenotype
ATP5D	59.5	84%	68%	No OMIM phenotype
ATP5E	155.4	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	72.5	66%	66%	No OMIM phenotype
ATP5G1	13.6	57%	28%	No OMIM phenotype
ATP5G2	58	94%	87%	No OMIM phenotype
ATP5G3	80.8	100%	91%	No OMIM phenotype
ATP5H	105.8	99%	83%	No OMIM phenotype
ATP5I	69.3	100%	100%	No OMIM phenotype

ATP5J	38.9	90%	82%	No OMIM phenotype
ATP5J2	44.3	91%	69%	No OMIM phenotype
ATP5L	50.1	70%	69%	No OMIM phenotype
ATP5L2	40.4	100%	77%	No OMIM phenotype
ATP5O	79.2	100%	100%	No OMIM phenotype
ATP5S	90.8	100%	97%	No OMIM phenotype
ATPAF1	75.2	88%	73%	No OMIM phenotype
ATPAF2	76.4	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATPIF1	197.1	100%	100%	No OMIM phenotype
BCS1L	159.5	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358
BOLA1	110	100%	100%	No OMIM phenotype
BOLA2	0.8	0%	0%	No OMIM phenotype
BOLA3	57.8	100%	94%	Multiple mitochondrial dysfunctions syndrome 2, 614299
C10orf2	145.4	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type),271245 Perrault syndrome 5,616138 Progressive external ophthalmoplegia with mitochondrial DNA depletions, dominant,609286
C11orf83	132.4	100%	100%	?Mitochondrial complex III deficiency,nuclear type 9,616111
C12orf65	177.7	100%	100%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55,autosomal recessive, 615035
C19orf12	88.2	100%	99%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
CARS2	74.9	99%	97%	Combined oxidative phosphorylation deficiency 27,616672
CEP89	87.4	99%	95%	No OMIM phenotype
CHCHD10	24	78%	31%	?Myopathy,isolated mitochondrial,autosomal dominant,616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2,615911 Spinal muscular atrophy,Jokela type,615048
CHKB	92.9	93%	89%	Muscular dystrophy, congenital, megaconial type, 602541
CLPB	102.4	94%	92%	3-methylglutaconic aciduria,type VII,with cataracts,neurologic involvement and neutropenia,616271
CLPP	88.4	99%	90%	Perrault syndrome 3, 614129
COA1	127.4	100%	100%	No OMIM phenotype

COA3	86.5	100%	99%	No OMIM phenotype
COA5	65.8	85%	84%	Mitochondrial complex IV deficiency, 220110
COA6	71.7	100%	97%	Cardioencephalomyopathy,fatal infantile,due to cytochrome c oxidase deficiency 4,616501
COASY	136.1	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COQ2	76	99%	88%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	81.2	90%	77%	Coenzyme Q10 deficiency,primary,7,616276
COQ6	114.6	98%	96%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ9	85.4	99%	85%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	122.7	100%	95%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX14	138.5	100%	100%	Mitochondrial complex IV deficiency, 220110
COX15	80.5	100%	99%	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119
COX20	64.8	89%	89%	Mitochondrial complex IV deficiency, 220110
COX4I1	62.6	91%	91%	No OMIM phenotype
COX4I2	49.3	99%	90%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis,612714
COX5A	59.1	68%	61%	No OMIM phenotype
COX5B	98.4	100%	100%	No OMIM phenotype
COX6A1	85.9	74%	74%	Charcot-Marie-Tooth disease,recessive intermediate D,616039
COX6A2	48.3	98%	71%	No OMIM phenotype
COX6B1	79.4	100%	100%	?Mitochondrial complex IV deficiency,220110
COX6B2	81.8	90%	78%	No OMIM phenotype
COX6C	151.7	100%	100%	No OMIM phenotype
COX7A1	80.8	100%	94%	No OMIM phenotype
COX7A2	53.9	83%	69%	No OMIM phenotype
COX7B	44.6	99%	92%	Linear skin defects with multiple congenital anomalies,300887
COX7B2	135.4	100%	100%	No OMIM phenotype
COX7C	24.8	79%	46%	No OMIM phenotype
COX8A	129.4	100%	100%	No OMIM phenotype
COX8C	30.1	42%	42%	No OMIM phenotype
CYC1	93.4	92%	79%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	49.7	100%	98%	Thrombocytopenia 4, 612004
DARS2	107.7	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105

DES	101.1	94%	92%	?Muscular dystrophy,limb-girdle,type 2R,615325 Cardiomyopathy,dilated,1I,604765 Myopathy,myofibrillar,1,601419 Scapuloperoneal syndrome,neurogenic,Kaeser type,181400
DGUOK	101.2	100%	100%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	103.9	100%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	101.6	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	131.7	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	90.6	100%	100%	No OMIM phenotype
DNA2	107.7	100%	98%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, AD, 6, 615156
DNAJC19	51.1	79%	78%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	94.9	100%	100%	?Ataxia,combined cerebellar and peripheral,with hearing loss and diabetes mellitus,616192
DNM1L	91.9	100%	99%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
EARS2	79.1	97%	91%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	63.3	95%	89%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency,616277
ECSIT	108.6	100%	97%	No OMIM phenotype
ELAC2	86.3	100%	100%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 Combined oxidative phosphorylation deficiency 17, 615440
ETHE1	58.5	99%	95%	Ethylmalonic encephalopathy, 602473
FARS2	104.6	98%	95%	Combined oxidative phosphorylation deficiency 14, 614946
FASTKD2	121.6	100%	100%	?Mitochondrial complex IV deficiency,220110
FBXL4	134.2	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX1L	93.4	100%	99%	No OMIM phenotype
FH	85.1	92%	86%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	94.5	99%	96%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency,252010
FXN	76.2	89%	79%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GATM	84.3	98%	92%	Cerebral creatine deficiency syndrome 3, 612718
GFER	63.9	95%	89%	Myopathy, mitochondrial progressive,cataract, hearing loss, and developmental delay, 613076
GFM1	120.5	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	103.3	100%	100%	No OMIM phenotype

GLRX5	39.7	87%	56%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	107.2	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GTPBP3	95.8	100%	99%	Combined oxidative phosphorylation deficiency 23,616198
HARS2	140.9	100%	100%	Perrault syndrome 2, 614926
HCCS	113.5	100%	99%	Microphthalmia, syndromic 7, 309801
HIBCH	60.1	100%	94%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	137.6	100%	100%	Holocarboxylase synthetase deficiency, 253270
HSD17B10	108.7	99%	92%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSPD1	16.4	61%	39%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
IARS2	119.4	100%	100%	?Cataracts,growth hormone deficiency,sensory neuropathy, sensorineural hearing loss and skeletal dysplasia,616007
IBA57	95	94%	92%	?Multiple mitochondrial dysfunctions syndrome 3,615330 ?Spastic paraplegia 74,autosomal recessive,616451
ISCA2	93.8	100%	100%	Multiple mitochondrial dysfunctions syndrome 4,616370
ISCU	93	100%	99%	Myopathy with lactic acidosis, hereditary, 255125
KARS	117.3	100%	100%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness,autosomal recessive 89,613916
LACTB	105.8	98%	94%	No OMIM phenotype
LARS2	117.6	100%	100%	Perrault syndrome 4, 615300
LIAS	103.8	100%	100%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
LIPT1	159	100%	100%	Lipoyltransferase 1 deficiency,616299
LONP1	83.7	97%	95%	CODAS syndrome,600373
LRPPRC	92.2	100%	98%	Leigh syndrome, French-Canadian type, 220111
LYRM4	54.4	66%	66%	?Combined oxidative phosphorylation deficiency 19,615595
LYRM7	78.1	100%	98%	?Mitochondrial complex III deficiency,nuclear type 8,615838
MARS2	172	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MCUR1	47.3	81%	71%	No OMIM phenotype
MFF	80.5	98%	94%	No OMIM phenotype
MFN2	107.2	99%	97%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA,601152
MGME1	149.4	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084

MPC1	87.4	100%	100%	Mitochondrial pyruvate carrier deficiency,614741
MPV17	109.1	100%	100%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 -3
MRPL12	74.5	94%	86%	No OMIM phenotype
MRPL3	76.2	98%	97%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	115	100%	95%	No OMIM phenotype paper in press (shoubridge)
MRPL44	114.9	100%	99%	?Combined oxidative phosphorylation deficiency 16,615395
MRPS16	144.2	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	108.6	100%	94%	No OMIM phenotype
MRPS22	89.9	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719
MRPS7	118.2	100%	100%	No OMIM phenotype
MRRF	148.8	100%	100%	No OMIM phenotype
MTFMT	96.2	100%	98%	Combined oxidative phosphorylation deficiency 15, 614947
MTO1	120.1	97%	93%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	96.6	93%	91%	Ataxia, spastic, 4, 613672
NARS2	113.4	100%	99%	Combined oxidative phosphorylation deficiency 24,616239
NDUFA1	214	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA10	85.4	99%	96%	previous assignment to chr. 12 Leigh syndrome, 256000
NDUFA11	104.2	90%	79%	Mitochondrial complex I deficiency, 252010
NDUFA12	87	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA13	73.4	95%	95%	{Thyroid carcinoma,Hurthle cell},607464
NDUFA2	168.4	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA3	90.9	100%	95%	No OMIM phenotype
NDUFA4	49.8	55%	53%	No OMIM phenotype
NDUFA5	44.1	54%	34%	No OMIM phenotype
NDUFA6	182	100%	100%	No OMIM phenotype
NDUFA7	69.2	99%	87%	No OMIM phenotype
NDUFA8	87.1	98%	98%	No OMIM phenotype
NDUFA9	85.2	96%	94%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 -3
NDUFAB1	67	100%	100%	No OMIM phenotype
NDUFAF1	107.6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	45	100%	92%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	143.7	100%	100%	Mitochondrial complex I deficiency, 252010

NDUFAF4	63.5	100%	98%	Mitochondrial complex I deficiency, 252010
NDUFAF5	119.1	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF6	92.3	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF7	79.3	100%	100%	No OMIM phenotype
NDUFB1	57.2	99%	87%	No OMIM phenotype
NDUFB10	132.5	100%	97%	No OMIM phenotype
NDUFB11	95	99%	98%	Linear skin defects with multiple congenital anomalies,300952
NDUFB2	88.8	100%	95%	No OMIM phenotype
NDUFB3	0.5	0%	0%	Mitochondrial complex I deficiency, 252010
NDUFB4	43	76%	68%	No OMIM phenotype
NDUFB5	85.1	100%	100%	No OMIM phenotype
NDUFB6	108	100%	100%	No OMIM phenotype
NDUFB7	52.8	91%	72%	No OMIM phenotype
NDUFB8	95.6	100%	100%	No OMIM phenotype
NDUFB9	109.6	100%	100%	?Mitochondrial complex I deficiency,252010
NDUFC1	79.3	100%	100%	No OMIM phenotype
NDUFC2	67	93%	85%	No OMIM phenotype
NDUFS1	78.3	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS2	128.6	98%	94%	Mitochondrial complex I deficiency, 252010
NDUFS3	133	93%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	111.5	100%	99%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS5	130.5	100%	100%	No OMIM phenotype
NDUFS6	133.4	92%	82%	Mitochondrial complex I deficiency, 252010
NDUFS7	106.1	100%	100%	Leigh syndrome, 256000
NDUFS8	107.2	100%	96%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	67	98%	94%	Mitochondrial complex I deficiency, 252010
NDUFV2	105.4	98%	98%	Mitochondrial complex I deficiency, 252010
NDUFV3	139.6	100%	96%	No OMIM phenotype
NFS1	64.3	89%	81%	No OMIM phenotype
NFU1	80.8	99%	91%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NUBPL	87	100%	99%	Mitochondrial complex I deficiency, 252010
OGDH	119.2	100%	100%	Alpha-ketoglutarate dehydrogenase deficiency,203740

OPA1	126.8	99%	99%	Optic atrophy 1, 165500
OPA3	98.5	100%	99%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OXA1L	141.4	99%	97%	No OMIM phenotype
PANK2	114.6	100%	89%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PARS2	169.2	100%	100%	No OMIM phenotype
PC	103.1	94%	91%	Pyruvate carboxylase deficiency, 266150
PDHA1	118	100%	99%	Pyruvate dehydrogenase E1-alpha deficiency, 312170 Leigh syndrome, X-linked, 308930
PDHB	105.2	100%	97%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	106.7	100%	100%	Lacticacidemia due to PDX1 deficiency,245349
PDK1	82	100%	100%	No OMIM phenotype
PDK2	88	100%	98%	No OMIM phenotype
PDK3	108.2	100%	99%	?Charcot-Marie-Tooth disease,X-linked dominant, 6,300905
PDK4	110.9	100%	100%	No OMIM phenotype
PDP1	158.5	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	94.4	89%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	95	100%	97%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	69.2	100%	91%	Mitochondrial complex IV deficiency, 220110
PET112	89	100%	99%	No OMIM phenotype
PIGA	138.2	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria,somatic,300818
PNPT1	99.6	100%	98%	Combined oxidative phosphorylation deficiency 13, 614932
POLG	90.7	99%	95%	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	129.9	100%	99%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, 4, 610131
PTRH2	141.4	100%	100%	infantile-onset multisystem neurologic,endocrine and pancreatic disease,616263
PUS1	70.6	100%	98%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PYCR1	87.6	100%	98%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
PYCR2	87.5	99%	94%	Leukodystrophy,hypomyelinating,10,616420

RARS2	80.8	100%	99%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	71.7	94%	87%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	71.7	91%	81%	Progressive external ophthalmoplegia with mitochondrial DNA deletions,AR 2,616479
RRM2B	111.1	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic with renal tubulopathy), 612075
SARS2	74.8	96%	90%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	85.1	100%	92%	Mitochondrial complex IV deficiency,220110
SCO2	99	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDHA	8.9	29%	14%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SDHAF1	92.7	100%	99%	Mitochondrial complex II deficiency, 252011
SDHB	91.4	100%	100%	Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764
SDHD	40.7	41%	35%	Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300 Carcinoid tumors, intestinal, 114900 Merkel cell carcinoma, somatic Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 3, 615106
SERAC1	88.6	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	87	100%	99%	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	94.1	100%	98%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	118	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	87.7	87%	85%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	114.2	100%	100%	Hypomyelination, global cerebral, 612949
SLC25A13	96.7	100%	98%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	81.2	100%	100%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A22	81.3	100%	97%	Epileptic encephalopathy, early infantile, 3, 609304

SLC25A3	68.8	88%	85%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A4	129.6	99%	91%	Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283 Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418
SLC25A46	97.2	100%	100%	Neuropathy,hereditary motor and sensory,type VIB,616505
SPG7	85.9	97%	90%	Spastic paraplegia 7, autosomal recessive, 607259
STXBP1	95.2	100%	98%	Epileptic encephalopathy,early infantile,4,612164
SUCLA2	74.4	91%	82%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with/without methylmalonic aciduria), 612073
SUCLG1	83.4	99%	92%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	82.1	100%	95%	No OMIM phenotype
SURF1	91.3	88%	88%	Leigh syndrome, due to COX deficiency, 256000
TACO1	86.6	93%	87%	Mitochondrial complex IV deficiency,220110
TARS2	99.1	100%	98%	?Combined oxidative phosphorylation deficiency 21,615918
TAZ	102.8	100%	100%	Barth syndrome, 302060
TIMM44	114.5	100%	100%	No OMIM phenotype
TIMM8A	64.4	99%	90%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TIMMDC1	148	100%	100%	No OMIM phenotype
TK2	89.6	100%	97%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM126A	83.7	100%	95%	Optic atrophy-7, 612989
TMEM126B	73.5	100%	98%	No OMIM phenotype
TMEM70	179.5	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TPK1	77.3	100%	100%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRIT1	119.2	98%	95%	No OMIM phenotype
TRMU	76.4	100%	97%	{Deafness, mitochondrial, modifier of}, 580000 Liver failure, transient infantile, 613070
TRNT1	99.3	100%	100%	Sideroblastic anemia with B-cell immunodeficiency,periodic fevers, and developmental delay,616084
TSFM	108.1	92%	86%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	61	86%	76%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	108.1	100%	95%	Combined oxidative phosphorylation deficiency 4, 610678
TYMP	90.5	100%	97%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041

UQCC1	106.4	100%	99%	No OMIM phenotype
UQCC2	103.2	100%	100%	?Mitochondrial complex III deficiency,nuclear type 7,615824
UQCR10	79.3	100%	100%	No OMIM phenotype
UQCR11	92.3	100%	100%	No OMIM phenotype
UQCRB	127.8	100%	100%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	103.7	95%	93%	No OMIM phenotype
UQCRC2	82	98%	91%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	2.8	28%	13%	No OMIM phenotype
UQCRH	61.7	88%	88%	No OMIM phenotype
UQCRQ	60.3	100%	98%	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	18.8	70%	36%	Combined oxidative phosphorylation deficiency 20,615917
YARS2	101.2	100%	100%	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
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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