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MITOCHONDRIAL DISORDERS PANEL DG-5.1.0 (507 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
AARS2	100%	100%	100%	100%	99.5%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ABAT	100%	100%	100%	100%	99.4%	GABA-transaminase deficiency, 613163
ABCB7	100%	100%	99.6%	92.7%	74.7%	Anemia, sideroblastic, with ataxia, 301310
ACAD9	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACO2	90.7%	86.8%	100%	100%	99.7%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
ACTA1	100%	100%	100%	99.8%	97.9%	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265;?Myopathy, scapulohumeroperoneal, 616852;Congenital myopathy 2C, severe infantile, autosomal dominant, 620278;Congenital myopathy 2A, typical, autosomal dominant, 161800
ADAMTS10	100%	100%	100%	100%	99.6%	Weill-Marchesani syndrome 1, recessive, 277600

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ADAR	100%	100%	100%	100%	99.8%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
ADCK2	86.1%	86.1%	100%	100%	98.9%	
ADPRS	100%	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG2A	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AFG3L2	94.2%	92.2%	100%	100%	99.5%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246
AGK	91.6%	88.9%	100%	100%	100%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AIFM1	100%	100%	99.3%	90.3%	72.6%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
AK3	100%	100%	100%	100%	99.9%	
ALDH1B1	100%	100%	100%	100%	99.5%	
ALDH7A1	97.2%	97.2%	100%	100%	99.8%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100
ALKBH1	90.6%	90.6%	100%	100%	99.8%	

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ANO10	94.9%	94.9%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOO	80.2%	80.1%	98.6%	91.1%	74%	
APTX	100%	100%	100%	100%	99.7%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARL2	100%	100%	100%	100%	99%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARNT2	91%	91%	100%	100%	99.5%	?Webb-Dattani syndrome, 615926
ATAD1	96.8%	96.8%	100%	100%	100%	Hyperekplexia 4, 618011
ATAD3A	100%	100%	100%	99.9%	98.7%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	94.5%	94.5%	99.9%	98.7%	96.3%	
ATP13A2	100%	100%	100%	100%	99.5%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP5F1A	100%	100%	100%	99.9%	99.7%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358;?Combined oxidative phosphorylation deficiency 22, 616045;?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228

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ATP5F1B	96.8%	96.6%	100%	100%	99.7%	?Hypermetabolism due to uncoupled mitochondrial oxidative phosphorylation 2, 620085;{Dystonia 38, susceptibility to}, 621502
ATP5F1C	100%	99.7%	100%	100%	99.6%	
ATP5F1D	100%	100%	100%	99.9%	98.9%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100%	100%	100%	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5IF1	89.1%	89.1%	100%	100%	99.4%	
ATP5MC1	86.7%	82.6%	100%	100%	98.8%	
ATP5MC2	100%	100%	100%	100%	99.8%	
ATP5MC3	100%	100%	100%	100%	99.8%	Dystonia, early-onset, and/or spastic paraplegia, 619681
ATP5ME	100%	100%	100%	100%	99%	
ATP5MF	100%	100%	100%	100%	100%	
ATP5MG	87.9%	87.9%	100%	100%	99.6%	
ATP5MGL	100%	100%	100%	100%	98.3%	
ATP5MK	100%	100%	100%	100%	99.8%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PB	100%	100%	100%	100%	99.7%	
ATP5PD	100%	100%	100%	100%	100%	
ATP5PF	100%	100%	100%	100%	99.9%	
ATP5PO	100%	100%	100%	100%	99.7%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359
ATPAF1	100%	100%	100%	99.9%	98.1%	

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ATPAF2	100%	100%	100%	100%	99.5%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BCAP31	91.5%	86.7%	98.7%	88.7%	70.4%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100%	100%	100%	100%	99.6%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BOLA1	100%	100%	100%	100%	99.2%	
BOLA2	100%	100%	100%	100%	100%	
BOLA3	100%	100%	100%	100%	98.9%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BTD	94.2%	94.2%	100%	100%	99.9%	Biotinidase deficiency, 253260
C19orf12	100%	100%	100%	99.9%	98.7%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043
C1QBP	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 33, 617713
C2orf69	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 53, 619423
CA5A	84.7%	84.7%	100%	100%	99.6%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CARS2	100%	99.9%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 27, 616672

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CDKL5	95.7%	95.7%	98.8%	89.3%	70.8%	Developmental and epileptic encephalopathy 2, 300672
CEP89	98.3%	98.3%	100%	100%	99.6%	
CFAP58	100%	100%	100%	100%	99.7%	Spermatogenic failure 49, 619144
CHCHD10	100%	100%	100%	100%	99.5%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHCHD2	90.1%	90.1%	100%	100%	99.8%	Parkinson disease 22, autosomal dominant, 616710
CHCHD4	100%	100%	100%	99.9%	98.8%	
CHKB	100%	100%	100%	100%	99.5%	Muscular dystrophy, congenital, megaconial type, 602541
CISD2	100%	100%	100%	99.9%	99.4%	Wolfram syndrome 2, 604928
CLPB	98.2%	98.2%	100%	100%	99.7%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CLPP	100%	100%	100%	100%	98.9%	Perrault syndrome 3, 614129
COA1	100%	100%	100%	100%	99.7%	
COA3	100%	100%	100%	100%	99.4%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058

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COA5	69.5%	69.5%	100%	99.8%	98.1%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100%	100%	100%	99.8%	99.1%	Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643
COQ2	96.3%	96.3%	100%	100%	99.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	96.3%	96.3%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ5	94.4%	94.4%	100%	100%	99.5%	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	100%	100%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	100%	100%	100%	99.5%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
COQ8A	89.1%	87.2%	100%	99.9%	99.5%	Coenzyme Q10 deficiency, primary, 4, 612016

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COQ8B	92.1%	92.1%	100%	100%	99.6%	Nephrotic syndrome, type 9, 615573
COQ9	91.9%	91.9%	100%	99.9%	99.2%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX11	100%	100%	100%	99.9%	98.7%	Mitochondrial complex IV deficiency, nuclear type 23, 620275
COX14	100%	100%	100%	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	100%	100%	100%	100%	99.4%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	61.9%	61.9%	100%	99.8%	99.4%	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX18	100%	100%	100%	99.9%	99.4%	?Mitochondrial complex IV deficiency, nuclear type 25, 621487;Charcot-Marie-Tooth disease, axonal, type 2MM, 621488
COX20	100%	100%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	100%	100%	100%	100%	99.4%	Mitochondrial complex IV deficiency, nuclear type 16, 619060
COX4I2	100%	99.1%	100%	100%	99.6%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX5B	91.4%	80.9%	100%	100%	99.3%	

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COX6A1	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	100%	100%	100%	99.9%	97.6%	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	80.9%	80.9%	100%	99.8%	98.7%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX6B2	100%	100%	100%	100%	99.4%	
COX6C	100%	100%	100%	100%	99.8%	
COX7A1	100%	100%	100%	100%	99.6%	
COX7A2	66.4%	66.4%	100%	100%	99.6%	
COX7B	100%	100%	98.7%	86.9%	70%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	100%	100%	100%	99.9%	99.3%	
COX7C	78.1%	70.4%	100%	100%	99.6%	
COX8A	100%	100%	100%	99.7%	97.3%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
COX8C	100%	100%	100%	100%	99.7%	
CP	100%	100%	100%	100%	99.9%	Aceruloplasminemia, 604290
CRAT	99.2%	96.9%	100%	99.8%	99.1%	?Neurodegeneration with brain iron accumulation 8, 617917
CRLS1	89.6%	84.4%	100%	99.7%	99.4%	Combined oxidative phosphorylation deficiency 57, 620167
CTBP1	100%	99.6%	100%	99.8%	99.3%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CYC1	96.9%	91.9%	100%	100%	99.3%	Mitochondrial complex III deficiency, nuclear type 6, 615453

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CYCS	100%	100%	100%	100%	99.9%	Thrombocytopenia 4, 612004
D2HGDH	99.9%	98.4%	100%	99.9%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAP3	91.7%	90.8%	100%	100%	99.6%	Perrault syndrome 7, 621101
DARS2	100%	100%	100%	100%	99.8%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105;Charcot-Marie-Tooth disease, axonal, type 2LL, 621485
DCAF17	96.5%	96.5%	100%	99.9%	99.7%	Woodhouse-Sakati syndrome, 241080
DDHD1	100%	100%	100%	100%	99.3%	Spastic paraplegia 28, autosomal recessive, 609340
DES	92.6%	92.6%	100%	99.9%	98.6%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419
DGUOK	100%	100%	100%	100%	99.9%	Portal hypertension, noncirrhotic, 1, 617068;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070;Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	92.7%	92.7%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750

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DLAT	100%	100%	100%	100%	99.4%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100%	100%	100%	100%	99.8%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	100%	100%	100%	99.9%	99.5%	Pheochromocytoma/paraganglioma syndrome 7, 618475
DMAC1	100%	100%	100%	100%	99.6%	
DMAC2	100%	100%	100%	100%	99.4%	
DMAC2L	100%	100%	100%	100%	99.7%	
DNA2	97.1%	97.1%	100%	100%	99.8%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156;Rothmund-Thomson syndrome, type 4, 620819;Seckel syndrome 8, 615807
DNAJA3	100%	100%	100%	100%	99.5%	
DNAJC19	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	92.1%	92.1%	100%	100%	99.8%	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC30	100%	100%	100%	100%	99.7%	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382
DNM1L	100%	100%	100%	100%	99.9%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388

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DNM2	93.3%	91.7%	100%	99.9%	99.4%	Centronuclear myopathy 1, 160150;Charcot-Marie-Tooth disease, axonal type 2M, 606482;Charcot-Marie-Tooth disease, dominant intermediate B, 606482;Lethal congenital contracture syndrome 5, 615368
EARS2	100%	100%	100%	99.9%	99.3%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	71.2%	69%	100%	100%	99.3%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	100%	100%	100%	99.9%	99%	
EHHADH	100%	100%	100%	100%	99.8%	?Fanconi renotubular syndrome 3, 615605
ELAC2	94.8%	94.8%	100%	99.9%	99.5%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440
EMD	93.1%	91.5%	97.5%	87.2%	69.2%	Cardiomyopathy, dilated, 3C, 301163;Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EPG5	100%	100%	100%	100%	99.8%	Vici syndrome, 242840;Neurodevelopmental disorder with parkinsonism or other movement abnormalities, 621506
ERAL1	100%	100%	100%	99.9%	99.3%	Perrault syndrome 6, 617565
ETFDH	94.1%	92.3%	100%	100%	99.9%	Glutaric acidemia IIC, 231680

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ETHE1	77.1%	77.1%	100%	100%	99.7%	Ethylmalonic encephalopathy, 602473
EXOSC8	100%	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 1C, 616081
FA2H	100%	100%	100%	100%	99.7%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FARSB	94%	94%	100%	100%	99.7%	Rajab interstitial lung disease with brain calcifications 1, 613658
FASTKD2	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 44, 618855
FASTKD5	100%	100%	100%	100%	99.9%	Mitochondrial complex IV deficiency, nuclear type 24, 621431
FBXL4	95.6%	95.6%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO7	99.1%	98.1%	100%	100%	99.6%	Parkinson disease 15, autosomal recessive, 260300
FDX2	100%	100%	100%	100%	99.3%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100%	100%	100%	100%	99.4%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717

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FH	100%	100%	100%	100%	99.8%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FLAD1	100%	100%	100%	100%	99.5%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLVCR1	95%	94.6%	100%	100%	99.7%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelop mental disorder with microcephaly, absent speech, and hypotonia, 621060
FOXRED1	100%	100%	100%	100%	99.1%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FTL	100%	100%	100%	100%	99.3%	Hyperferritinemia-catar act syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegenera tion with brain iron accumulation 3, 606159
FXN	100%	100%	100%	100%	98.9%	Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300
GARS1	98.9%	98.9%	100%	100%	99.8%	Spinal muscular atrophy, infantile, James type, 619042;Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794;Charcot-Marie-Tooth disease, type 2D, 601472
GATB	100%	100%	100%	100%	99.7%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	80.1%	80.1%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 42, 618839

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GATM	100%	100%	100%	100%	99.9%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600
GBF1	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483
GCSH	100%	100%	100%	100%	99.9%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GDAP1	86.2%	86.2%	98.3%	98%	97.1%	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706;Charcot-Marie-Tooth disease, recessive intermediate, A, 608340;Charcot-Marie-Tooth disease, axonal, type 2K, 607831;Charcot-Marie-Tooth disease, type 4A, 214400
GFER	100%	100%	100%	99.9%	99.4%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	96.6%	96.6%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 39, 618397
GLRX5	100%	100%	100%	100%	99.7%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	100%	100%	100%	100%	99.8%	Hyperinsulinism-hyperammonemia syndrome, 606762
GMPR	87.3%	87.3%	100%	100%	99.6%	

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GOT2	92.8%	92.8%	100%	100%	99.5%	Developmental and epileptic encephalopathy 82, 618721
GPT2	92.9%	92.9%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GTPBP2	100%	100%	100%	100%	99.9%	Jaberi-Elahi syndrome, 617988
GTPBP3	100%	100%	100%	99.9%	99.3%	Combined oxidative phosphorylation deficiency 23, 616198
GUF1	100%	100%	100%	100%	99.6%	?Developmental and epileptic encephalopathy 40, 617065
GUK1	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 21, 621071
HACE1	95.9%	95.9%	100%	100%	99.7%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADHA	96.1%	95.3%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	94.6%	94.6%	100%	100%	99.7%	Mitochondrial trifunctional protein deficiency 2, 620300
HARS2	100%	100%	100%	99.9%	98.9%	Perrault syndrome 2, 614926
HCCS	100%	100%	99.3%	91.9%	75.5%	Linear skin defects with multiple congenital anomalies 1, 309801

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HIBCH	100%	100%	100%	100%	100%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HK1	98.2%	96.5%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelop mental disorder with visual defects and brain anomalies, 618547
HLCS	100%	100%	100%	100%	99.2%	Holocarboxylase synthetase deficiency, 253270
HPDL	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027
HSD17B10	100%	100%	99.5%	91%	71.8%	HSD10 mitochondrial disease, 300438
HSPA9	100%	100%	100%	100%	99.6%	Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170
HSPD1	99.9%	98.9%	100%	100%	99.9%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystroph y, hypomyelinating, 4, 612233
HTRA2	100%	100%	100%	100%	99.5%	{Parkinson disease 13}, 610297;3-methylglutac onic aciduria, type VIII, 617248

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IARS2	100%	99.7%	100%	99.9%	99.6%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100%	100%	100%	100%	99.5%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451
IDH2	89.7%	89.5%	100%	100%	99.4%	D-2-hydroxyglutaric aciduria 2, 613657
ISCA1	92.4%	92.4%	100%	100%	99.6%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100%	100%	100%	100%	99.7%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100%	100%	100%	100%	99.7%	Myopathy with lactic acidosis, hereditary, 255125
KARS1	94.3%	94.3%	100%	100%	99.8%	Deafness, autosomal recessive 89, 613916;Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147;?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KGD4	100%	100%	100%	99.8%	99.5%	

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KIF1A	100%	100%	100%	100%	99.3%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
LACTB	100%	100%	100%	100%	99.6%	
LARS2	96.2%	96.2%	100%	100%	99.8%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LDHD	100%	100%	100%	100%	99.5%	D-lactic aciduria with susceptibility to gout, 245450
LDLR	100%	100%	100%	100%	99.1%	LDL cholesterol level QTL2, 143890;Hypercholesterolemia, familial, 1, 143890
LETM1	92.4%	89.8%	100%	100%	99.5%	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089
LIAS	100%	100%	100%	100%	99.9%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG3	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780
LIPT1	100%	100%	100%	100%	99.8%	Lipoyltransferase 1 deficiency, 616299
LIPT2	100%	100%	100%	100%	99%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668

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LONP1	99.3%	97%	100%	100%	99.5%	CODAS syndrome, 600373
LRPPRC	96.8%	96.7%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LYRM4	68%	68%	100%	100%	99.5%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	100%	100%	100%	100%	99.4%	Mitochondrial complex III deficiency, nuclear type 8, 615838
MAPT	95.4%	95.4%	100%	99.9%	99.2%	Supranuclear palsy, progressive, 601104;Frontotemporal dementia 1, with or without parkinsonism, 600274;Supranuclear palsy, progressive atypical, 260540;{Parkinson disease, susceptibility to}, 168600;Pick disease, 172700
MARS2	100%	100%	100%	99.9%	99.2%	?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390
MCAT	95.9%	95.9%	100%	100%	99.6%	Optic atrophy 15, 620583
MCUR1	100%	100%	100%	99.9%	99.4%	
MDH1	88.5%	88.5%	100%	100%	99.8%	?Developmental and epileptic encephalopathy 88, 618959
MDH2	100%	100%	100%	100%	98.7%	Developmental and epileptic encephalopathy 51, 617339

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MECR	93.5%	93.5%	100%	100%	99.6%	?Optic atrophy 16, 620629;Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	89.1%	89.1%	100%	100%	99.9%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100%	100%	100%	100%	99.7%	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800;Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260;Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087;Hereditary motor and sensory neuropathy VIA, 601152
MGME1	100%	100%	100%	100%	99.5%	Mitochondrial DNA depletion syndrome 11, 615084
MICOS13	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	86.7%	86.7%	100%	100%	99.8%	Myopathy with extrapyramidal signs, 615673
MICU2	88.6%	85.9%	100%	99.9%	99.5%	
MIEF1	100%	100%	100%	100%	99.3%	Optic atrophy 14, 620550
MIEF2	100%	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 49, 619024
MIPEP	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 31, 617228

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MORC2	100%	100%	100%	100%	99.3%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPC1	100%	100%	100%	100%	99.9%	Mitochondrial pyruvate carrier deficiency, 614741
MPC2	100%	100%	100%	99.9%	99.7%	
MPV17	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRM2	87.4%	87.3%	100%	99.7%	98.3%	Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	100%	100%	100%	100%	99.3%	?Combined oxidative phosphorylation deficiency 45, 618951
MRPL24	100%	100%	100%	99.7%	98.7%	
MRPL3	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL34	100%	100%	100%	100%	98.7%	
MRPL39	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 59, 620646
MRPL40	100%	100%	100%	99.9%	99.5%	
MRPL42	71.3%	71.3%	100%	99.9%	99.6%	
MRPL44	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 16, 615395
MRPL49	100%	100%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 60, 621195

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MRPL50	100%	100%	100%	99.6%	98.7%	
MRPL57	100%	100%	100%	100%	100%	
MRPS14	85.2%	85.2%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100%	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	100%	100%	100%	99.9%	99.6%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	86.6%	86.6%	100%	100%	99.9%	Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	100%	100%	100%	100%	99.7%	?Combined oxidative phosphorylation deficiency 46, 618952
MRPS25	74.2%	74.2%	100%	100%	99.8%	?Combined oxidative phosphorylation deficiency 50, 619025
MRPS28	73.8%	73.8%	100%	99.8%	97.8%	?Combined oxidative phosphorylation deficiency 47, 618958
MRPS34	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	100%	100%	100%	99.9%	99%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	100%	100%	100%	100%	100%	
MSTO1	100%	100%	99.9%	99.1%	97.7%	Myopathy, mitochondrial, and ataxia, 617675
MTERF3	100%	100%	100%	100%	99.9%	

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MTFMT	91.8%	91.8%	100%	99.9%	98.6%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248
MTO1	94.8%	91.9%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	96.6%	96.6%	100%	100%	99.4%	?Spastic ataxia 4, autosomal recessive, 613672
MTRFR	100%	100%	100%	100%	99.7%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
MTX2	87.1%	87.1%	100%	100%	99.2%	Mandibuloacral dysplasia progeroid syndrome, 619127
NADK2	100%	100%	100%	99.9%	99.2%	2,4-dienoyl-CoA reductase deficiency, 616034
NARS2	92.3%	92.3%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NAXD	95.5%	89.7%	100%	100%	99.2%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	97.6%	93.5%	100%	100%	99.5%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFA1	100%	100%	98.8%	90.1%	70.9%	Mitochondrial complex I deficiency, nuclear type 12, 301020

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NDUFA10	82.9%	80.5%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	96.1%	95.9%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	80.4%	80.4%	100%	99.9%	99.7%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	99.1%	94.7%	100%	100%	99.5%	{Thyroid carcinoma, Hurthle cell}, 607464;Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	93.9%	87.2%	100%	100%	98.4%	
NDUFA4	100%	100%	100%	100%	100%	
NDUFA5	75.6%	75.6%	100%	99.9%	99.6%	
NDUFA6	100%	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	78.2%	78.2%	100%	100%	98.6%	
NDUFA8	100%	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFA9	87%	85.2%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	100%	100%	100%	100%	99.5%	
NDUFAF1	91.6%	91.5%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	67.4%	67.4%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240

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NDUFAF4	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	90.9%	90.9%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 17, 618239;Fanconi renotubular syndrome 5, 618913
NDUFAF7	100%	100%	100%	100%	99.9%	
NDUFAF8	100%	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	100%	100%	100%	100%	97.6%	
NDUFB10	100%	100%	100%	100%	99.7%	?Mitochondrial complex I deficiency, nuclear type 35, 619003
NDUFB11	99.9%	99.3%	93.6%	79.3%	59.7%	Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	100%	100%	100%	100%	99.7%	
NDUFB3	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	75%	75%	100%	100%	99.5%	
NDUFB5	100%	100%	100%	100%	99.7%	
NDUFB6	100%	100%	100%	100%	99.9%	
NDUFB7	72.3%	72.3%	100%	100%	99.7%	?Mitochondrial complex I deficiency, nuclear type 39, 620135
NDUFB8	100%	100%	100%	99.7%	98.9%	Mitochondrial complex I deficiency, nuclear type 32, 618252

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NDUFB9	86.8%	86.6%	100%	100%	99.5%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	100%	100%	100%	99.9%	99.2%	
NDUFC2	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 36, 619170
NDUFS1	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	99.9%	98.3%	100%	100%	99.6%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	98%	92.5%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	100%	100%	100%	100%	100%	
NDUFS6	94.7%	86.1%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	89.1%	85%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	100%	100%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100%	100%	100%	100%	98.7%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	100%	100%	100%	100%	99.5%	
NFS1	83.4%	83.4%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 52, 619386

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NFU1	98.9%	94.6%	100%	100%	99.7%	Spastic paraplegia 93, autosomal recessive, 620938;Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	98.4%	97.2%	100%	100%	99.8%	Congenital disorder of deglycosylation 1, 615273
NME3	100%	100%	100%	100%	99.1%	
NR2F1	100%	100%	100%	99.9%	98.5%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRDC	96.9%	96.9%	100%	100%	99.8%	
NRROS	100%	100%	100%	99.9%	99.2%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NSUN3	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 48, 619012
NUBPL	89.8%	89.8%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP54	100%	100%	100%	100%	99.7%	Dystonia 37, early-onset, with striatal lesions, 620427
NUTF2	97.3%	96.9%	100%	99.9%	99.4%	
OGDH	100%	100%	100%	100%	99.5%	Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	100%	100%	100%	100%	99.5%	

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OPA1	94.7%	94.7%	100%	100%	99.7%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type), 621481;?Mitochondrial DNA depletion syndrome 14B (cardioencephalomyopathic type), 616896
OPA3	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300
OTX2	100%	100%	100%	100%	99.9%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
OXA1L	100%	100%	100%	100%	99.7%	
P4HTM	89.4%	89.4%	100%	99.9%	99.2%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PANK2	100%	100%	100%	100%	99.6%	Neurodegeneration with brain iron accumulation 1, 234200
PARS2	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 75, 618437

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PC	99.9%	99.2%	100%	100%	99.5%	Pyruvate carboxylase deficiency, 266150
PDE12	93%	93%	100%	100%	99.5%	
PDE2A	100%	100%	100%	100%	99.3%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDHA1	91.4%	89.5%	99.3%	91.4%	72.9%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100%	100%	100%	100%	99.8%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	91.6%	91.6%	100%	100%	99.6%	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	100%	100%	100%	100%	99.4%	
PDK2	92%	92%	100%	100%	99.3%	
PDK3	100%	100%	99.1%	91%	73.3%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	100%	100%	100%	100%	99.7%	
PDP1	100%	100%	100%	100%	99.9%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	100%	100%	100%	100%	99.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100%	100%	100%	100%	99.4%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	100%	100%	100%	100%	99.2%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PET117	100%	100%	100%	100%	99.5%	?Mitochondrial complex IV deficiency, nuclear type 19, 619063

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PGM1	87.5%	87.5%	100%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PIGA	100%	100%	99.2%	90.8%	73.4%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PISD	100%	100%	100%	99.9%	99.5%	Liberfarb syndrome, 618889
PITRM1	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 30, 619405
PLA2G6	100%	100%	100%	100%	99.4%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLPBP	100%	100%	100%	99.8%	98.6%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PMPCA	96.2%	96.2%	100%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	91.4%	91.4%	100%	100%	99.8%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNPLA8	100%	100%	100%	100%	99.5%	Mitochondrial myopathy with lactic acidosis, 251950

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PNPT1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POLG	100%	100%	100%	100%	99.8%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	95.8%	95.8%	100%	100%	99.5%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131;?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528;?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425
POLR2A	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603

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POLRMT	99.7%	98.7%	100%	100%	99%	Combined oxidative phosphorylation deficiency 55, 619743
PPA2	82.6%	82.6%	100%	100%	99.7%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222
PPCS	100%	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 2C, 618189
PRDX3	100%	98.8%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871
PRKAA1	100%	100%	100%	100%	99.8%	
PRORP	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 54, 619737
PRPS1	100%	100%	99%	89.8%	71.8%	Arts syndrome, 301835;Phosphoribosyl pyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661
PTCD3	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 51, 619057
PTPMT1	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with ataxia and brain abnormalities, 621199
PTRH2	100%	100%	100%	100%	99.9%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263

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PUS1	100%	100%	100%	99.9%	99.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	100%	100%	100%	100%	99.8%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100%	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 10, 616420
PYROXD1	92.6%	92.6%	100%	100%	99.6%	Myopathy, myofibrillar, 8, 617258
PYROXD2	82.2%	79.5%	100%	99.9%	99.1%	
QRSL1	91.9%	89.5%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 40, 618835
RANBP2	100%	100%	100%	100%	99.8%	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RARS2	94.2%	93.8%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	85.8%	85.8%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	88.3%	88.3%	100%	100%	99.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNF213	100%	100%	100%	100%	99.6%	{Moyamoya disease 2, susceptibility to}, 607151
RNU4-2						Retinitis pigmentosa 102, 621560;ReNU syndrome, 620851

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RRM1	100%	100%	100%	100%	99.7%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647
RRM2B	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RTN4IP1	100%	100%	100%	100%	99.8%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732
RYR1	100%	100%	100%	99.9%	99%	Congenital myopathy 1B, autosomal recessive, 255320;Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000;King-Denborough syndrome, 619542;{Malignant hyperthermia susceptibility 1}, 145600
SACS	99%	99%	100%	100%	99.9%	Spastic ataxia, Charlevoix-Saguenay type, 270550

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SAMD9	100%	100%	100%	100%	99.8%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMD9L	100%	100%	100%	100%	99.9%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SAMHD1	92%	89.8%	100%	100%	99.5%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SARS2	100%	100%	100%	100%	99.7%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SATB2	100%	100%	100%	99.9%	99.5%	Glass syndrome, 612313
SCO1	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100%	100%	100%	100%	99.8%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	95.5%	95.5%	100%	100%	99.7%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724

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SDHA	92.6%	89.3%	100%	100%	99.7%	Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma/paraganglioma syndrome 5, 614165
SDHAF1	100%	100%	100%	100%	99.3%	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHB	94%	94%	100%	100%	99.7%	Pheochromocytoma/paraganglioma syndrome 4, 115310;Mitochondrial complex II deficiency, nuclear type 4, 619224;Gastrointestinal stromal tumor, 606764;Paraganglioma and gastric stromal sarcoma, 606864
SDHD	78.9%	78.9%	100%	100%	99.5%	Pheochromocytoma/paraganglioma syndrome 1, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Mitochondrial complex II deficiency, nuclear type 3, 619167
SERAC1	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	100%	100%	100%	99.9%	99.6%	Combined oxidative phosphorylation deficiency 18, 615578
SHMT2	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121

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SIRT5	100%	100%	100%	100%	99.6%	
SLC19A2	100%	100%	100%	100%	99.7%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	99.9%	98.9%	100%	100%	99.7%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483
SLC25A1	100%	100%	100%	99.9%	98.8%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A10	99.7%	97.6%	100%	100%	99.2%	?Mitochondrial DNA depletion syndrome 19, 618972
SLC25A12	90.5%	89.5%	100%	100%	99.9%	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	91.7%	91.7%	100%	100%	99.6%	Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471
SLC25A19	100%	100%	100%	100%	99.8%	Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A21	100%	100%	100%	100%	99.9%	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A22	100%	100%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99.4%	99.4%	99.7%	99.1%	97.9%	Fontaine progeroid syndrome, 612289

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SLC25A26	95.5%	95.5%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	100%	100%	100%	100%	99.6%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100%	100%	100%	100%	99.7%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A36	95%	95%	100%	100%	99.4%	Hyperinsulinemic hypoglycemia, familial, 8, 620211
SLC25A38	90.6%	90.6%	100%	100%	99.4%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283;Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184
SLC25A42	100%	100%	100%	100%	99.2%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	100%	100%	100%	100%	99.7%	Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303
SLC27A3	100%	100%	100%	99.9%	99%	
SLC39A8	77.5%	77.4%	100%	100%	100%	Congenital disorder of glycosylation, type IIn, 616721

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SLC52A2	100%	100%	100%	99.9%	99.7%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	99.4%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC8B1	100%	100%	100%	100%	99.6%	
SLIRP	85.2%	84.3%	100%	100%	99.3%	
SMDT1	100%	100%	100%	100%	99.9%	
SOD2	100%	100%	100%	100%	99.8%	{Microvascular complications of diabetes 6}, 612634
SPART	95.8%	95.8%	100%	100%	99.7%	Troyer syndrome, 275900
SPG7	100%	100%	100%	99.9%	99.3%	Spastic paraplegia 7, autosomal recessive, 607259
SPTBN4	100%	100%	100%	99.9%	98.7%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SQOR	100%	100%	100%	100%	99.7%	Sulfide:quinone oxidoreductase deficiency, 619221
SQSTM1	100%	100%	100%	100%	98.9%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437;Myopathy, distal, with rimmed vacuoles, 617158;Paget disease of bone 3, 167250
SSBP1	100%	100%	100%	100%	100%	Optic atrophy 13 with retinal and foveal abnormalities, 165510
STAC3	100%	100%	100%	100%	99.2%	Congenital myopathy 13, 255995

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STAT2	100%	100%	100%	100%	99.5%	Pseudo-TORCH syndrome 3, 618886;Immunodeficiency 44, 616636
STXBP1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 4, 612164
SUCLA2	96.3%	96.3%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100%	100%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	100%	100%	100%	100%	99.9%	
SUPV3L1	95.3%	95.3%	100%	100%	99.6%	
SURF1	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SZT2	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 18, 615476
TACO1	100%	100%	100%	99.8%	99.3%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TAFAZZIN	100%	100%	98.6%	89.7%	70.1%	Barth syndrome, 302060
TAMM41	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 56, 620139

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TANGO2	100%	100%	100%	100%	99.6%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	100%	100%	100%	100%	99.8%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TARS2	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 21, 615918
TBCK	100%	100%	100%	100%	99.9%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TDP2	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TEFM	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 58, 620451
TFAM	100%	100%	100%	99.9%	99.4%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFB2M	100%	100%	100%	100%	99.8%	
THG1L	100%	100%	100%	99.7%	99.3%	Spinocerebellar ataxia, autosomal recessive 28, 618800
TIMM22	100%	100%	100%	99.8%	98.3%	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	100%	100%	100%	99.8%	98.7%	
TIMM50	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type IX, 617698

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TIMM8A	100%	100%	98.2%	87.9%	71.6%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	100%	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TK2	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TMEM126A	100%	100%	100%	100%	100%	Optic atrophy 7, 612989
TMEM126B	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM186	100%	100%	100%	100%	99.8%	
TMEM63C	100%	100%	100%	100%	99.1%	Spastic paraplegia 87, autosomal recessive, 619966
TMEM65	100%	99.5%	100%	99.7%	98.5%	
TMEM70	100%	100%	100%	100%	99.3%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMX2	99.8%	97%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TOMM40L	100%	100%	100%	100%	99.3%	
TOMM7	100%	100%	100%	100%	99.6%	Garg-Mishra progeroid syndrome, 620601
TOMM70	100%	100%	100%	100%	99.7%	

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TOP3A	95.7%	95%	100%	100%	99.5%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TPK1	93.3%	93.3%	100%	100%	99.9%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRAPPC2L	100%	100%	100%	100%	99.6%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRIT1	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT10C	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100%	100%	100%	100%	99.8%	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539
TRMU	88%	88%	100%	100%	99.4%	{Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070
TRNT1	91.9%	91.9%	100%	100%	99.9%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959

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TSFM	91.5%	91.1%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	100%	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	99.7%	96.7%	100%	99.9%	99.4%	Combined oxidative phosphorylation deficiency 4, 610678
TWNK	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
TXN2	80.8%	80.6%	100%	100%	99.9%	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	100%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UCHL1	100%	100%	100%	100%	99.8%	{?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491
UFM1	100%	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 14, 617899
UPB1	94.3%	91.8%	100%	100%	99.5%	Beta-ureidopropionase deficiency, 613161
UQCC1	100%	100%	100%	100%	99.7%	
UQCC2	100%	100%	100%	100%	99.9%	Mitochondrial complex III deficiency, nuclear type 7, 615824

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UQCC3	100%	100%	100%	100%	99.6%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	100%	100%	100%	100%	99%	
UQCR11	100%	100%	100%	100%	98.8%	
UQCRB	100%	100%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	100%	100%	100%	100%	99.5%	Parkinsonism with polyneuropathy, 619279
UQCRC2	74.7%	74.7%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	100%	100%	100%	100%	99.4%	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRH	78.7%	76%	100%	100%	99.8%	?Mitochondrial complex III deficiency, nuclear type 11, 620137
UQCRQ	100%	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	100%	100%	100%	100%	98.9%	Combined oxidative phosphorylation deficiency 20, 615917
VPS13D	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 4, 607317
WARS2	93.8%	93.8%	100%	100%	100%	Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WDR45	98.4%	98%	98.4%	87.9%	68.6%	Neurodegeneration with brain iron accumulation 5, 300894

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XPNPEP3	100%	100%	100%	99.9%	99.6%	Nephronophthisis-like nephropathy 1, 613159
YARS2	100%	100%	100%	100%	99.8%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	96.7%	96.7%	100%	100%	99.7%	?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.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors