MITOCHONDRIAL DISORDERS PANEL DG-4.0.0 (485 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	100.0%	100.0%	100.0%	99.4%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ABAT	100.0%	100.0%	100.0%	98.7%	GABA-transaminase deficiency, 613163
ABCB7	99.8%	99.3%	98.3%	74.8%	Anemia, sideroblastic, with ataxia, 301310
ACAD9	100.0%	100.0%	100.0%	99.6%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACO2	92.4%	89.8%	100.0%	99.3%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559

ACTA1	100.0%	100.0%	100.0%	97.1%	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	100.0%	100.0%	100.0%	99.1%	Weill-Marchesani syndrome 1, recessive, 277600
ADAR	100.0%	100.0%	100.0%	98.2%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
ADCK2	100.0%	100.0%	100.0%	98.2%	
ADPRS	100.0%	100.0%	100.0%	99.2%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG2A	100.0%	100.0%	100.0%	98.9%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AFG3L2	100.0%	100.0%	100.0%	98.4%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246

AGK	91.7%	91.7%	100.0%	98.9%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AIFM1	100.0%	99.8%	97.6%	67.9%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
AK3	100.0%	100.0%	100.0%	98.5%	
ALDH1B1	100.0%	100.0%	100.0%	99.7%	
ALKBH1	100.0%	100.0%	100.0%	97.5%	
ANO10	100.0%	100.0%	100.0%	98.2%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOO	100.0%	100.0%	98.3%	71.2%	
APTX	100.0%	100.0%	100.0%	98.5%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARL2	100.0%	100.0%	100.0%	98.5%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARNT2	100.0%	100.0%	100.0%	98.7%	?Webb-Dattani syndrome, 615926
ATAD1	100.0%	99.7%	100.0%	97.4%	Hyperekplexia 4, 618011

ATAD3A	100.0%	100.0%	99.9%	96.7%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	100.0%	100.0%	99.8%	95.0%	
ATP13A2	100.0%	100.0%	100.0%	99.3%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP5F1A	100.0%	100.0%	100.0%	99.6%	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
ATP5F1B	100.0%	100.0%	100.0%	99.2%	?Hypermetabolism due to uncoupled mitochondrial oxidative phosphorylation 2, 620085
ATP5F1C	100.0%	100.0%	100.0%	97.8%	
ATP5F1D	100.0%	100.0%	100.0%	97.9%	Mitochondrial complex V (ATP synthase) deficiency, 618120

ATP5F1E	100.0%	100.0%	100.0%	96.8%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5IF1	100.0%	100.0%	100.0%	98.6%	
ATP5MC1	100.0%	100.0%	100.0%	99.7%	
ATP5MC2	100.0%	100.0%	100.0%	98.2%	
ATP5MC3	100.0%	100.0%	100.0%	99.3%	Dystonia, early-onset, and/or spastic paraplegia, 619681
ATP5ME	100.0%	100.0%	100.0%	98.7%	
ATP5MF	100.0%	100.0%	100.0%	99.5%	
ATP5MG	95.4%	95.4%	100.0%	97.4%	
ATP5MGL	100.0%	100.0%	100.0%	99.8%	
ATP5MK	100.0%	100.0%	100.0%	97.2%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PB	100.0%	100.0%	100.0%	99.6%	
ATP5PD	100.0%	100.0%	100.0%	98.4%	
ATP5PF	100.0%	100.0%	100.0%	97.4%	
ATP5PO	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359
ATPAF1	100.0%	100.0%	99.9%	92.8%	
ATPAF2	100.0%	100.0%	100.0%	99.1%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273

BCAP31	99.1%	92.8%	98.0%	69.1%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100.0%	100.0%	100.0%	99.2%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BOLA1	100.0%	100.0%	100.0%	100.0%	
BOLA2	100.0%	100.0%	100.0%	99.3%	
BOLA3	100.0%	100.0%	100.0%	97.5%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BTD	94.2%	94.2%	100.0%	99.5%	Biotinidase deficiency, 253260
C19orf12	100.0%	99.8%	100.0%	98.4%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043
C1QBP	100.0%	100.0%	100.0%	98.9%	Combined oxidative phosphorylation deficiency 33, 617713
C2orf69	100.0%	100.0%	99.9%	96.5%	Combined oxidative phosphorylation deficiency 53, 619423
CA5A	100.0%	100.0%	100.0%	98.0%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751

CARS2	100.0%	100.0%	100.0%	99.0%	Combined oxidative phosphorylation deficiency 27, 616672
CDKL5	95.7%	95.3%	97.4%	68.8%	Developmental and epileptic encephalopathy 2, 300672
CEP89	100.0%	100.0%	100.0%	96.9%	
CFAP58	100.0%	100.0%	100.0%	97.2%	Spermatogenic failure 49, 619144
CHCHD10	100.0%	100.0%	100.0%	96.9%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHCHD2	100.0%	100.0%	100.0%	99.6%	Parkinson disease 22, autosomal dominant, 616710
CHKB	100.0%	100.0%	100.0%	98.7%	Muscular dystrophy, congenital, megaconial type, 602541
CISD2	100.0%	100.0%	100.0%	98.0%	Wolfram syndrome 2, 604928

CLPB	100.0%	100.0%	99.9%	98.3%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CLPP	100.0%	100.0%	100.0%	96.3%	Perrault syndrome 3, 614129
COA1	100.0%	100.0%	100.0%	98.6%	
COA3	100.0%	100.0%	100.0%	99.4%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA5	82.4%	82.4%	100.0%	98.4%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	100.0%	100.0%	100.0%	96.5%	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100.0%	100.0%	100.0%	99.2%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	100.0%	99.9%	100.0%	97.0%	Mitochondrial complex IV deficiency, nuclear type 17, 619061

COASY	100.0%	100.0%	100.0%	99.1%	Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643
COQ2	96.3%	96.3%	100.0%	98.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100.0%	100.0%	100.0%	99.6%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ5	100.0%	100.0%	100.0%	97.4%	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	100.0%	100.0%	99.9%	98.4%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100.0%	100.0%	100.0%	98.8%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
COQ8A	100.0%	100.0%	100.0%	99.7%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100.0%	100.0%	100.0%	99.0%	Nephrotic syndrome, type 9, 615573
COQ9	100.0%	100.0%	100.0%	98.8%	Coenzyme Q10 deficiency, primary, 5, 614654

COX10	100.0%	100.0%	100.0%	99.4%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX11	100.0%	100.0%	100.0%	94.9%	Mitochondrial complex IV deficiency, nuclear type 23, 620275
COX14	100.0%	100.0%	100.0%	100.0%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	100.0%	100.0%	100.0%	98.5%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	100.0%	100.0%	99.9%	98.3%	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX18	100.0%	100.0%	100.0%	98.3%	
COX20	100.0%	100.0%	100.0%	98.9%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	100.0%	100.0%	100.0%	99.2%	Mitochondrial complex IV deficiency, nuclear type 16, 619060
COX4I2	100.0%	100.0%	100.0%	98.8%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	100.0%	100.0%	100.0%	98.0%	Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX5B	100.0%	100.0%	100.0%	98.1%	

COX6A1	100.0%	100.0%	100.0%	97.6%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	100.0%	99.6%	100.0%	95.1%	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	100.0%	100.0%	100.0%	99.3%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX6B2	100.0%	100.0%	100.0%	92.8%	
COX6C	100.0%	100.0%	100.0%	98.6%	
COX7A1	100.0%	100.0%	100.0%	92.0%	
COX7A2	100.0%	100.0%	100.0%	95.5%	
COX7B	100.0%	99.9%	98.5%	76.8%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	100.0%	100.0%	100.0%	99.7%	
COX7C	100.0%	100.0%	100.0%	97.1%	
COX8A	100.0%	100.0%	100.0%	99.8%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
COX8C	100.0%	100.0%	100.0%	99.5%	
CP	100.0%	100.0%	100.0%	98.7%	Aceruloplasminemia, 604290
CRAT	100.0%	100.0%	100.0%	99.3%	?Neurodegeneration with brain iron accumulation 8, 617917

CRLS1	100.0%	100.0%	100.0%	95.1%	Combined oxidative phosphorylation deficiency 57, 620167
CTBP1	100.0%	99.5%	99.4%	97.5%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CYC1	100.0%	100.0%	100.0%	96.9%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	100.0%	100.0%	100.0%	98.2%	Thrombocytopenia 4, 612004
D2HGDH	100.0%	100.0%	100.0%	99.2%	D-2-hydroxyglutaric aciduria, 600721
DARS2	100.0%	100.0%	100.0%	96.8%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF17	100.0%	100.0%	99.9%	98.3%	Woodhouse-Sakati syndrome, 241080
DDHD1	100.0%	100.0%	100.0%	97.6%	Spastic paraplegia 28, autosomal recessive, 609340
DES	100.0%	100.0%	100.0%	98.9%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419

DGUOK	100.0%	100.0%	100.0%	98.6%	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	100.0%	100.0%	100.0%	98.0%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DLAT	100.0%	100.0%	100.0%	98.9%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100.0%	100.0%	100.0%	98.7%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	100.0%	100.0%	100.0%	98.9%	Pheochromocytoma/paraganglioma syndrome 7, 618475
DMAC1	100.0%	100.0%	100.0%	96.7%	
DMAC2	100.0%	100.0%	100.0%	99.5%	
DMAC2L	100.0%	100.0%	100.0%	99.2%	

DNA2	100.0%	100.0%	100.0%	97.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156;Rothmund-Thomson syndrome, type 4, 620819;Seckel syndrome 8, 615807
DNAJA3	100.0%	100.0%	100.0%	99.1%	
DNAJC19	100.0%	100.0%	100.0%	98.3%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	100.0%	100.0%	99.9%	97.4%	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC30	100.0%	100.0%	100.0%	99.7%	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382
DNM1L	100.0%	100.0%	100.0%	98.6%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	100.0%	100.0%	100.0%	98.3%	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	100.0%	100.0%	100.0%	98.7%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	100.0%	100.0%	100.0%	96.6%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	100.0%	100.0%	100.0%	99.0%	
EHHADH	100.0%	100.0%	100.0%	99.2%	?Fanconi renotubular syndrome 3, 615605
ELAC2	100.0%	100.0%	100.0%	99.3%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440
EMD	92.9%	90.4%	98.3%	71.2%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EPG5	100.0%	100.0%	100.0%	98.4%	Vici syndrome, 242840
ERAL1	100.0%	100.0%	100.0%	98.3%	Perrault syndrome 6, 617565
ETFDH	93.6%	92.0%	100.0%	98.9%	Glutaric acidemia IIC, 231680
ETHE1	100.0%	100.0%	100.0%	97.9%	Ethylmalonic encephalopathy, 602473
EXOSC8	100.0%	100.0%	100.0%	97.0%	Pontocerebellar hypoplasia, type 1C, 616081
FA2H	100.0%	100.0%	100.0%	98.8%	Spastic paraplegia 35, autosomal recessive, 612319

FARS2	100.0%	100.0%	100.0%	99.1%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FARSB	100.0%	100.0%	100.0%	98.9%	Rajab interstitial lung disease with brain calcifications 1, 613658
FASTKD2	100.0%	100.0%	100.0%	97.2%	Combined oxidative phosphorylation deficiency 44, 618855
FBXL4	100.0%	100.0%	100.0%	99.2%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX2	100.0%	99.6%	100.0%	98.7%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100.0%	100.0%	100.0%	99.4%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717
FH	100.0%	100.0%	100.0%	98.5%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FLAD1	100.0%	100.0%	100.0%	99.4%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100

FOXRED1	100.0%	100.0%	100.0%	98.6%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FTL	100.0%	100.0%	100.0%	96.5%	Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159
FXN	100.0%	100.0%	100.0%	95.9%	Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300
GARS1	98.9%	98.9%	100.0%	98.6%	Spinal muscular atrophy, infantile, James type, 619042;Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794;Charcot-Marie-Tooth disease, type 2D, 601472
GATB	100.0%	100.0%	100.0%	99.1%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100.0%	100.0%	100.0%	99.1%	Combined oxidative phosphorylation deficiency 42, 618839
GATM	100.0%	100.0%	100.0%	97.8%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600

GBF1	100.0%	100.0%	100.0%	99.0%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483
GCSH	100.0%	100.0%	100.0%	98.1%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GDAP1	86.7%	86.7%	98.0%	96.0%	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	100.0%	100.0%	99.6%	91.8%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	100.0%	100.0%	100.0%	98.6%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	100.0%	100.0%	100.0%	98.7%	Combined oxidative phosphorylation deficiency 39, 618397
GLRX5	100.0%	100.0%	100.0%	97.1%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859

GLUD1	100.0%	100.0%	100.0%	94.8%	Hyperinsulinism-hyperammonemia syndrome, 606762
GMPR	100.0%	100.0%	100.0%	99.3%	
GOT2	100.0%	100.0%	100.0%	99.2%	Developmental and epileptic encephalopathy 82, 618721
GPT2	100.0%	100.0%	100.0%	98.3%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GTPBP2	100.0%	100.0%	100.0%	98.3%	Jaberi-Elahi syndrome, 617988
GTPBP3	100.0%	100.0%	100.0%	98.3%	Combined oxidative phosphorylation deficiency 23, 616198
GUF1	100.0%	100.0%	99.9%	97.4%	?Developmental and epileptic encephalopathy 40, 617065
HACE1	100.0%	100.0%	100.0%	97.4%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADHA	100.0%	100.0%	100.0%	98.8%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016

HADHB	100.0%	100.0%	100.0%	99.0%	Mitochondrial trifunctional protein deficiency 2, 620300
HARS2	100.0%	100.0%	100.0%	98.9%	Perrault syndrome 2, 614926
HCCS	100.0%	100.0%	97.8%	69.8%	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	100.0%	100.0%	100.0%	98.2%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	100.0%	100.0%	99.9%	97.6%	Holocarboxylase synthetase deficiency, 253270
HPDL	100.0%	100.0%	100.0%	98.9%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027
HSD17B10	100.0%	99.8%	98.0%	70.3%	HSD10 mitochondrial disease, 300438
HSPA9	100.0%	100.0%	100.0%	98.5%	Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170
HSPD1	99.6%	97.9%	100.0%	98.7%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
HTRA2	100.0%	100.0%	100.0%	98.4%	{Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248

IARS2	100.0%	100.0%	100.0%	98.0%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100.0%	100.0%	100.0%	99.2%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451
IDH2	100.0%	100.0%	100.0%	98.1%	D-2-hydroxyglutaric aciduria 2, 613657
ISCA1	92.4%	92.4%	100.0%	98.9%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100.0%	100.0%	100.0%	98.9%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100.0%	100.0%	100.0%	99.2%	Myopathy with lactic acidosis, hereditary, 255125

KARS1	100.0%	100.0%	100.0%	98.4%	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
KIF1A	100.0%	100.0%	100.0%	99.5%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
LACTB	100.0%	100.0%	100.0%	97.5%	
LARS2	100.0%	100.0%	100.0%	99.1%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LDHD	100.0%	100.0%	100.0%	99.3%	D-lactic aciduria with susceptibility to gout, 245450

LETM1	100.0%	100.0%	100.0%	99.3%	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089
LIAS	100.0%	100.0%	100.0%	99.2%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG3	100.0%	100.0%	100.0%	99.1%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780
LIPT1	100.0%	100.0%	100.0%	96.6%	Lipoyltransferase 1 deficiency, 616299
LIPT2	100.0%	100.0%	100.0%	98.2%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LONP1	100.0%	100.0%	100.0%	99.1%	CODAS syndrome, 600373
LRPPRC	96.8%	96.5%	100.0%	98.1%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LYRM4	68.0%	68.0%	100.0%	98.8%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	100.0%	100.0%	100.0%	98.2%	Mitochondrial complex III deficiency, nuclear type 8, 615838

MAPT	95.4%	95.4%	100.0%	98.0%	Supranuclear palsy, progressive, 601104;Supranuclear palsy, progressive atypical, 260540;Dementia, frontotemporal, with or without parkinsonism, 600274;{Parkinson disease, susceptibility to}, 168600;Pick disease, 172700
MARS2	100.0%	100.0%	100.0%	99.7%	?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390
MCAT	100.0%	100.0%	100.0%	99.3%	Optic atrophy 15, 620583
MCUR1	100.0%	100.0%	100.0%	95.2%	
MDH1	100.0%	100.0%	100.0%	99.2%	?Developmental and epileptic encephalopathy 88, 618959
MDH2	100.0%	100.0%	100.0%	98.6%	Developmental and epileptic encephalopathy 51, 617339
MECR	100.0%	100.0%	100.0%	99.2%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282;Optic atrophy 16, 620629
MFF	95.9%	95.9%	100.0%	98.8%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086

MFN2	100.0%	100.0%	100.0%	98.7%	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	100.0%	100.0%	100.0%	96.3%	Mitochondrial DNA depletion syndrome 11, 615084
MICOS13	100.0%	100.0%	100.0%	99.4%	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	100.0%	99.9%	100.0%	99.0%	Myopathy with extrapyramidal signs, 615673
MICU2	100.0%	100.0%	99.9%	96.8%	
MIEF2	100.0%	100.0%	100.0%	99.5%	?Combined oxidative phosphorylation deficiency 49, 619024
MIPEP	100.0%	100.0%	100.0%	98.7%	Combined oxidative phosphorylation deficiency 31, 617228

MORC2	100.0%	100.0%	100.0%	98.5%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPC1	100.0%	100.0%	100.0%	98.4%	Mitochondrial pyruvate carrier deficiency, 614741
MPC2	100.0%	100.0%	100.0%	96.0%	
MPV17	100.0%	100.0%	100.0%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRM2	97.0%	97.0%	100.0%	98.3%	Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	100.0%	100.0%	100.0%	99.0%	?Combined oxidative phosphorylation deficiency 45, 618951
MRPL24	100.0%	100.0%	100.0%	99.3%	
MRPL3	100.0%	100.0%	100.0%	98.4%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL39	100.0%	100.0%	100.0%	98.2%	Combined oxidative phosphorylation deficiency 59, 620646
MRPL40	100.0%	100.0%	100.0%	98.4%	
MRPL42	100.0%	100.0%	100.0%	98.7%	

MRPL44	100.0%	100.0%	100.0%	98.9%	Combined oxidative phosphorylation deficiency 16, 615395
MRPL50	100.0%	100.0%	100.0%	99.2%	
MRPL57	100.0%	100.0%	100.0%	99.6%	
MRPS14	100.0%	100.0%	100.0%	99.8%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100.0%	100.0%	100.0%	98.9%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	100.0%	100.0%	100.0%	99.0%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	100.0%	99.9%	100.0%	98.2%	Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	100.0%	100.0%	100.0%	99.7%	?Combined oxidative phosphorylation deficiency 46, 618952
MRPS25	74.2%	74.2%	100.0%	98.5%	?Combined oxidative phosphorylation deficiency 50, 619025
MRPS28	85.4%	85.3%	99.9%	96.2%	?Combined oxidative phosphorylation deficiency 47, 618958
MRPS34	100.0%	100.0%	100.0%	99.4%	Combined oxidative phosphorylation deficiency 32, 617664

MRPS36	100.0%	100.0%	100.0%	96.9%	
MRPS7	100.0%	100.0%	100.0%	99.2%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	100.0%	100.0%	100.0%	99.0%	
MSTO1	100.0%	100.0%	100.0%	98.6%	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	100.0%	100.0%	100.0%	97.8%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248
MTO1	93.7%	91.1%	100.0%	98.4%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	100.0%	100.0%	100.0%	98.0%	?Spastic ataxia 4, autosomal recessive, 613672
MTRFR	100.0%	99.7%	99.7%	98.1%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
MTX2	100.0%	99.9%	100.0%	97.8%	Mandibuloacral dysplasia progeroid syndrome, 619127
NADK2	100.0%	100.0%	100.0%	95.8%	2,4-dienoyl-CoA reductase deficiency, 616034

NARS2	92.3%	92.3%	100.0%	98.5%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NAXD	96.8%	92.4%	100.0%	99.2%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	95.8%	91.2%	100.0%	98.5%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFA1	100.0%	100.0%	95.8%	64.4%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	83.4%	81.0%	100.0%	98.6%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	100.0%	98.8%	100.0%	96.7%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	79.4%	79.4%	100.0%	98.0%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	100.0%	100.0%	100.0%	99.1%	{Thyroid carcinoma, Hurthle cell}, 607464;Mitochondrial complex I deficiency, nuclear type 28, 618249

NDUFA2	100.0%	100.0%	100.0%	99.3%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	91.4%	86.8%	100.0%	98.9%	
NDUFA4	100.0%	100.0%	100.0%	95.7%	?Mitochondrial complex IV deficiency, nuclear type 21, 619065
NDUFA5	75.0%	75.0%	100.0%	98.1%	
NDUFA6	100.0%	100.0%	100.0%	98.5%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	100.0%	100.0%	100.0%	98.5%	
NDUFA8	100.0%	100.0%	100.0%	98.8%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFA9	100.0%	100.0%	100.0%	99.0%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	100.0%	100.0%	100.0%	97.5%	
NDUFAF1	100.0%	100.0%	100.0%	98.3%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	67.4%	67.4%	100.0%	97.3%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100.0%	100.0%	100.0%	95.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240

NDUFAF4	100.0%	100.0%	100.0%	95.3%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100.0%	100.0%	99.9%	96.1%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100.0%	100.0%	100.0%	96.3%	Mitochondrial complex I deficiency, nuclear type 17, 618239;Fanconi renotubular syndrome 5, 618913
NDUFAF7	100.0%	100.0%	100.0%	97.5%	
NDUFAF8	100.0%	100.0%	100.0%	99.3%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	100.0%	100.0%	99.9%	93.2%	
NDUFB10	100.0%	100.0%	100.0%	95.2%	?Mitochondrial complex I deficiency, nuclear type 35, 619003
NDUFB11	99.7%	97.9%	88.1%	61.0%	Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	100.0%	100.0%	100.0%	99.5%	
NDUFB3	100.0%	100.0%	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	100.0%	100.0%	100.0%	99.1%	
NDUFB5	100.0%	100.0%	100.0%	98.1%	

NDUFB6	100.0%	100.0%	100.0%	98.8%	
NDUFB7	100.0%	100.0%	99.8%	96.4%	?Mitochondrial complex I deficiency, nuclear type 39, 620135
NDUFB8	100.0%	100.0%	100.0%	97.7%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	100.0%	100.0%	100.0%	98.9%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	100.0%	100.0%	100.0%	97.2%	
NDUFC2	100.0%	100.0%	100.0%	97.7%	Mitochondrial complex I deficiency, nuclear type 36, 619170
NDUFS1	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	99.5%	96.5%	100.0%	98.3%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	96.5%	91.2%	100.0%	99.2%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100.0%	99.9%	100.0%	98.0%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	100.0%	100.0%	100.0%	98.7%	

NDUFS6	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100.0%	100.0%	100.0%	99.6%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100.0%	100.0%	100.0%	99.6%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100.0%	100.0%	99.9%	98.8%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100.0%	100.0%	100.0%	98.5%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	100.0%	100.0%	100.0%	99.0%	
NFS1	89.8%	89.8%	100.0%	99.2%	Combined oxidative phosphorylation deficiency 52, 619386
NFU1	100.0%	100.0%	100.0%	98.4%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	100.0%	100.0%	100.0%	98.7%	Congenital disorder of deglycosylation 1, 615273
NME3	100.0%	100.0%	99.8%	95.9%	
NR2F1	100.0%	99.9%	99.9%	91.8%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722

NRROS	100.0%	100.0%	100.0%	99.6%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NSUN3	100.0%	100.0%	100.0%	98.5%	Combined oxidative phosphorylation deficiency 48, 619012
NUBPL	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP54	100.0%	100.0%	100.0%	98.7%	Dystonia 37, early-onset, with striatal lesions, 620427
NUTF2	97.2%	96.9%	100.0%	98.9%	
OGDH	100.0%	100.0%	100.0%	99.3%	Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	100.0%	100.0%	100.0%	99.2%	Yoon-Bellen neurodevelopmental syndrome, 619701
OPA1	100.0%	100.0%	100.0%	98.5%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	100.0%	100.0%	100.0%	98.6%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300

OTX2	100.0%	100.0%	100.0%	98.2%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
OXA1L	100.0%	100.0%	99.9%	97.9%	
P4HTM	100.0%	100.0%	100.0%	95.5%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PANK2	100.0%	100.0%	100.0%	98.6%	Neurodegeneration with brain iron accumulation 1, 234200
PARS2	100.0%	100.0%	100.0%	99.5%	Developmental and epileptic encephalopathy 75, 618437
PC	100.0%	100.0%	100.0%	99.7%	Pyruvate carboxylase deficiency, 266150
PDE2A	100.0%	100.0%	100.0%	98.3%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDHA1	99.6%	96.5%	97.6%	72.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100.0%	100.0%	100.0%	98.8%	Pyruvate dehydrogenase E1-beta deficiency, 614111

PDHX	100.0%	99.8%	99.9%	98.1%	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	100.0%	100.0%	100.0%	97.4%	
PDK2	100.0%	100.0%	100.0%	98.8%	
PDK3	100.0%	100.0%	98.2%	73.8%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	100.0%	100.0%	100.0%	98.2%	
PDP1	100.0%	100.0%	100.0%	99.4%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	100.0%	100.0%	100.0%	97.3%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100.0%	100.0%	100.0%	98.5%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	100.0%	100.0%	100.0%	99.3%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PET117	100.0%	100.0%	100.0%	93.6%	?Mitochondrial complex IV deficiency, nuclear type 19, 619063
PIGA	100.0%	100.0%	97.7%	73.6%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072

PISD	100.0%	100.0%	100.0%	99.8%	Liberfarb syndrome, 618889
PITRM1	100.0%	100.0%	100.0%	99.0%	Spinocerebellar ataxia, autosomal recessive 30, 619405
PLA2G6	100.0%	99.9%	100.0%	99.2%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLPBP	100.0%	100.0%	100.0%	99.2%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PMPCA	96.0%	96.0%	100.0%	99.0%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	91.4%	91.4%	100.0%	98.2%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNPLA8	100.0%	100.0%	100.0%	97.0%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPT1	100.0%	100.0%	100.0%	98.3%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932

POLG	100.0%	100.0%	100.0%	99.4%	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	100.0%	100.0%	100.0%	97.3%	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	100.0%	100.0%	100.0%	98.7%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603

POLRMT	100.0%	100.0%	100.0%	99.6%	Combined oxidative phosphorylation deficiency 55, 619743
PPA2	100.0%	99.9%	100.0%	96.7%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222
PPCS	100.0%	100.0%	100.0%	98.6%	Cardiomyopathy, dilated, 2C, 618189
PRDX3	100.0%	100.0%	100.0%	98.6%	Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871
PRKAA1	100.0%	100.0%	100.0%	97.4%	
PRORP	100.0%	100.0%	100.0%	97.9%	Combined oxidative phosphorylation deficiency 54, 619737
PRPS1	100.0%	100.0%	96.3%	69.8%	Arts syndrome, 301835;Phosphoribosylpyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661
PTCD3	100.0%	100.0%	100.0%	98.7%	Combined oxidative phosphorylation deficiency 51, 619057

PTPMT1	100.0%	100.0%	99.9%	93.5%	
PTRH2	100.0%	100.0%	100.0%	98.8%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	100.0%	100.0%	100.0%	98.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	100.0%	100.0%	100.0%	99.8%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100.0%	100.0%	100.0%	98.6%	Leukodystrophy, hypomyelinating, 10, 616420
PYROXD1	100.0%	100.0%	100.0%	97.3%	Myopathy, myofibrillar, 8, 617258
PYROXD2	90.6%	87.8%	100.0%	99.1%	
QRSL1	100.0%	100.0%	100.0%	98.4%	Combined oxidative phosphorylation deficiency 40, 618835
RANBP2	100.0%	100.0%	100.0%	97.4%	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RARS2	94.2%	93.1%	100.0%	98.6%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	85.6%	85.6%	100.0%	97.6%	Combined oxidative phosphorylation deficiency 11, 614922

RNASEH1	100.0%	100.0%	100.0%	98.9%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNF213	100.0%	100.0%	100.0%	99.2%	{Moyamoya disease 2, susceptibility to}, 607151
RRM1	100.0%	100.0%	100.0%	98.6%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647
RRM2B	100.0%	100.0%	100.0%	97.7%	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	100.0%	100.0%	100.0%	97.5%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732

RYR1	100.0%	99.9%	100.0%	98.7%	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	99.0%	99.0%	100.0%	98.0%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	100.0%	100.0%	100.0%	98.1%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SARS2	100.0%	100.0%	100.0%	98.6%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SATB2	100.0%	99.7%	100.0%	98.6%	Glass syndrome, 612313
SCO1	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100.0%	100.0%	100.0%	99.5%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	100.0%	100.0%	100.0%	97.9%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724

SDHA	100.0%	100.0%	100.0%	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	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHB	100.0%	100.0%	100.0%	98.4%	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%	100.0%	98.4%	Pheochromocytoma/paraganglioma syndrome 1, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Mitochondrial complex II deficiency, nuclear type 3, 619167
SERAC1	100.0%	100.0%	100.0%	98.3%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739

SFXN4	100.0%	100.0%	100.0%	96.9%	Combined oxidative phosphorylation deficiency 18, 615578
SHMT2	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SIRT5	100.0%	100.0%	100.0%	99.5%	
SLC19A2	100.0%	100.0%	100.0%	99.5%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	99.6%	98.4%	100.0%	98.1%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483
SLC25A1	100.0%	100.0%	100.0%	93.2%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A10	100.0%	100.0%	100.0%	99.8%	?Mitochondrial DNA depletion syndrome 19, 618972
SLC25A12	100.0%	100.0%	100.0%	98.5%	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	100.0%	100.0%	100.0%	98.7%	Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471

SLC25A19	100.0%	100.0%	100.0%	98.7%	Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A21	100.0%	100.0%	100.0%	98.5%	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A22	100.0%	100.0%	100.0%	99.6%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99.5%	99.5%	99.6%	97.2%	Fontaine progeroid syndrome, 612289
SLC25A26	100.0%	100.0%	100.0%	98.8%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	100.0%	100.0%	100.0%	99.0%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100.0%	100.0%	100.0%	98.9%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A36	100.0%	100.0%	100.0%	97.2%	Hyperinsulinemic hypoglycemia, familial, 8, 620211
SLC25A38	100.0%	100.0%	100.0%	99.2%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950

SLC25A4	100.0%	100.0%	100.0%	98.5%	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	100.0%	100.0%	100.0%	99.2%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	100.0%	100.0%	99.9%	98.1%	Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303
SLC39A8	99.9%	99.4%	100.0%	97.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC52A2	100.0%	100.0%	100.0%	99.9%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100.0%	100.0%	100.0%	99.0%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC8B1	100.0%	100.0%	100.0%	99.6%	

SLIRP	100.0%	100.0%	99.9%	95.1%	
SMDT1	100.0%	100.0%	100.0%	99.2%	
SOD2	100.0%	100.0%	100.0%	99.4%	{Microvascular complications of diabetes 6}, 612634
SPART	100.0%	100.0%	100.0%	98.1%	Troyer syndrome, 275900
SPG7	100.0%	100.0%	100.0%	98.6%	Spastic paraplegia 7, autosomal recessive, 607259
SPTBN4	100.0%	100.0%	100.0%	98.3%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SQOR	100.0%	100.0%	100.0%	98.2%	Sulfide:quinone oxidoreductase deficiency, 619221
SQSTM1	100.0%	100.0%	100.0%	99.3%	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	100.0%	100.0%	100.0%	98.6%	Optic atrophy 13 with retinal and foveal abnormalities, 165510

STAC3	100.0%	100.0%	100.0%	98.5%	Congenital myopathy 13, 255995
STAT2	100.0%	100.0%	100.0%	99.0%	Pseudo-TORCH syndrome 3, 618886;Immunodeficiency 44, 616636
STXBP1	100.0%	100.0%	100.0%	98.6%	Developmental and epileptic encephalopathy 4, 612164
SUCLA2	100.0%	99.6%	100.0%	98.8%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100.0%	100.0%	100.0%	96.4%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	100.0%	99.8%	100.0%	97.2%	
SUPV3L1	100.0%	100.0%	100.0%	98.1%	
SURF1	100.0%	100.0%	100.0%	98.7%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SZT2	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 18, 615476
TACO1	100.0%	100.0%	100.0%	98.4%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TAFAZZIN	100.0%	100.0%	96.7%	66.1%	Barth syndrome, 302060

TAMM41	100.0%	100.0%	100.0%	98.7%	Combined oxidative phosphorylation deficiency 56, 620139
TANGO2	100.0%	100.0%	100.0%	99.4%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	100.0%	100.0%	100.0%	98.6%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TARS2	100.0%	100.0%	100.0%	98.4%	Combined oxidative phosphorylation deficiency 21, 615918
TBCK	100.0%	100.0%	100.0%	98.7%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TDP2	100.0%	100.0%	100.0%	97.8%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TEFM	100.0%	100.0%	100.0%	98.6%	Combined oxidative phosphorylation deficiency 58, 620451
TFAM	100.0%	100.0%	100.0%	98.1%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFB2M	100.0%	100.0%	100.0%	97.4%	

THG1L	100.0%	100.0%	100.0%	98.3%	Spinocerebellar ataxia, autosomal recessive 28, 618800
TIMM22	100.0%	100.0%	100.0%	99.1%	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	100.0%	100.0%	100.0%	98.2%	
TIMM50	100.0%	100.0%	100.0%	99.5%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	100.0%	99.5%	97.6%	65.5%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	100.0%	100.0%	100.0%	97.8%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TK2	100.0%	100.0%	100.0%	98.5%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TMEM126A	100.0%	100.0%	100.0%	97.6%	Optic atrophy 7, 612989
TMEM126B	100.0%	100.0%	100.0%	99.0%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM186	100.0%	100.0%	100.0%	99.9%	
TMEM63C	100.0%	100.0%	100.0%	98.9%	Spastic paraplegia 87, autosomal recessive, 619966

TMEM65	100.0%	98.6%	99.9%	91.7%	
TMEM70	100.0%	100.0%	100.0%	97.4%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMX2	100.0%	100.0%	100.0%	99.1%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TOMM40L	100.0%	100.0%	100.0%	98.6%	
TOMM70	100.0%	100.0%	100.0%	99.1%	
TOP3A	100.0%	100.0%	100.0%	98.8%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TPK1	100.0%	100.0%	100.0%	98.0%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRAPPC2L	100.0%	100.0%	100.0%	99.8%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRIT1	100.0%	100.0%	100.0%	98.4%	Combined oxidative phosphorylation deficiency 35, 617873

TRMT10C	100.0%	100.0%	100.0%	98.4%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100.0%	100.0%	100.0%	98.6%	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539
TRMU	100.0%	100.0%	100.0%	97.9%	{Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070
TRNT1	92.0%	91.9%	100.0%	98.7%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959
TSFM	94.3%	94.3%	100.0%	98.5%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	100.0%	100.0%	100.0%	97.2%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	100.0%	100.0%	100.0%	98.5%	Combined oxidative phosphorylation deficiency 4, 610678

TWNK	100.0%	100.0%	100.0%	99.8%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
TXN2	100.0%	100.0%	100.0%	99.7%	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	100.0%	100.0%	100.0%	98.6%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UCHL1	100.0%	100.0%	100.0%	97.9%	{?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491
UFM1	100.0%	100.0%	100.0%	99.0%	Leukodystrophy, hypomyelinating, 14, 617899
UQCC1	100.0%	100.0%	100.0%	96.5%	
UQCC2	100.0%	100.0%	100.0%	99.8%	Mitochondrial complex III deficiency, nuclear type 7, 615824

UQCC3	100.0%	100.0%	100.0%	97.8%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	100.0%	100.0%	100.0%	97.3%	
UQCR11	100.0%	100.0%	100.0%	99.2%	
UQCRB	100.0%	100.0%	100.0%	98.6%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	100.0%	100.0%	100.0%	99.5%	Parkinsonism with polyneuropathy, 619279
UQCRC2	100.0%	100.0%	100.0%	98.4%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	100.0%	100.0%	100.0%	99.0%	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRH	100.0%	100.0%	100.0%	99.0%	?Mitochondrial complex III deficiency, nuclear type 11, 620137
UQCRQ	100.0%	100.0%	100.0%	98.2%	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	100.0%	100.0%	100.0%	99.2%	Combined oxidative phosphorylation deficiency 20, 615917
VPS13D	100.0%	100.0%	100.0%	98.9%	Spinocerebellar ataxia, autosomal recessive 4, 607317

WARS2	100.0%	100.0%	100.0%	99.2%	Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WDR45	100.0%	100.0%	98.9%	76.4%	Neurodegeneration with brain iron accumulation 5, 300894
XPNPEP3	100.0%	100.0%	100.0%	99.2%	Nephronophthisis-like nephropathy 1, 613159
YARS2	100.0%	100.0%	100.0%	97.6%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	100.0%	100.0%	100.0%	97.7%	?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.
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.
srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.
This list is accurate for panel version DG 4.0.0

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