MITOCHONDRIAL DISORDERS GENE PANEL DG 3.4.0 (445 genes)

Releasedate: 19-04-2022

Gene	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AARS2	100,0%	100,0%	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
ABAT	100,0%	100,0%	GABA-transaminase deficiency, 613163
ACAD9	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACO2	100,0%	100,0%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACTA1	100,0%	100,0%	?Myopathy, scapulohumeroperoneal, 616852 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310
ADAMTS10	100,0%	100,0%	Weill-Marchesani syndrome 1, recessive, 277600
ADCK2	100,0%	100,0%	No OMIM Disease ID
ADPRS	100,0%	100,0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	100,0%	100,0%	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGK	91,2%	91,2%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AIFM1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614
ALDH1B1	100,0%	100,0%	No OMIM Disease ID
ALKBH1	100,0%	100,0%	No OMIM Disease ID
ANO10	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOO	100,0%	100,0%	No OMIM Disease ID
APTX	100,0%	100,0%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARL2	100,0%	100,0%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082

ARNT2	100,0%	100,0%	?Webb-Dattani syndrome, 615926
ATAD1	100,0%	100,0%	Hyperekplexia 4, 618011
ATAD3A	100,0%	100,0%	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	100,0%	100,0%	No OMIM Disease ID
ATP13A2	100,0%	100,0%	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP5F1A	100,0%	100,0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5F1B	100,0%	100,0%	No OMIM Disease ID
ATP5F1C	100,0%	100,0%	No OMIM Disease ID
ATP5F1D	100,0%	100,0%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100,0%	100,0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5IF1	100,0%	100,0%	No OMIM Disease ID
ATP5MC1	100,0%	100,0%	No OMIM Disease ID
ATP5MC2	100,0%	100,0%	No OMIM Disease ID
ATP5MC3	100,0%	100,0%	Dystonia, early-onset, and/or spastic paraplegia, 619681
ATP5ME	100,0%	100,0%	No OMIM Disease ID
ATP5MF	100,0%	100,0%	No OMIM Disease ID
ATP5MG	100,0%	100,0%	No OMIM Disease ID
ATP5MGL	100,0%	100,0%	No OMIM Disease ID
ATP5MD	100,0%	100,0%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PB	100,0%	100,0%	No OMIM Disease ID
ATP5PD	100,0%	100,0%	No OMIM Disease ID
ATP5PF	100,0%	100,0%	No OMIM Disease ID
ATP5PO	100,0%	100,0%	No OMIM Disease ID
ATPAF1	100,0%	100,0%	No OMIM Disease ID
ATPAF2	100,0%	100,0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
BCAP31	100,0%	100,0%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100,0%	100,0%	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BOLA1	100,0%	100,0%	No OMIM Disease ID
BOLA2	100,0%	100,0%	No OMIM Disease ID
BOLA3	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
C19orf12	100,0%	100,0%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043

C1QBP	100,0%	100,0%	Combined oxidative phosphorylation deficiency 33, 617713
C2orf69	100,0%	100,0%	Combined oxidative phosphorylation deficiency 53, 619423
CA5A	87,7%	87,7%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 27, 616672
CDKL5	92,3%	92,2%	Developmental and epileptic encephalopathy 2, 300672
CEP89	100,0%	100,0%	No OMIM Disease ID
CFAP58	100,0%	100,0%	Spermatogenic failure 49, 619144
CHCHD10	100,0%	100,0%	?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%	Parkinson disease 22, autosomal dominant, 616710
CHKB	100,0%	100,0%	Muscular dystrophy, congenital, megaconial type, 602541
CISD2	100,0%	100,0%	Wolfram syndrome 2, 604928
CLPB	100,0%	100,0%	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%	Perrault syndrome 3, 614129
COA1	100,0%	100,0%	No OMIM Disease ID
COA3	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA5	85,2%	85,2%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	93,5%	93,5%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100,0%	100,0%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COQ2	97,2%	97,2%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	100,0%	100,0%	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100,0%	100,0%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100,0%	100,0%	Nephrotic syndrome, type 9, 615573
COQ9	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX14	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 22, 619355

COX20	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 16, 619060
COX4I2	100,0%	100,0%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX5B	100,0%	100,0%	No OMIM Disease ID
COX6A1	100,0%	100,0%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX6B2	100,0%	100,0%	No OMIM Disease ID
COX6C	100,0%	100,0%	No OMIM Disease ID
COX7A1	100,0%	100,0%	No OMIM Disease ID
COX7A2	100,0%	100,0%	No OMIM Disease ID
COX7B	100,0%	100,0%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	100,0%	100,0%	No OMIM Disease ID
COX7C	100,0%	100,0%	No OMIM Disease ID
COX8A	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
COX8C	100,0%	100,0%	No OMIM Disease ID
CP	100,0%	100,0%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CRAT	100,0%	100,0%	?Neurodegeneration with brain iron accumulation 8, 617917
CTBP1	100,0%	99,4%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CYC1	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	100,0%	100,0%	Thrombocytopenia 4, 612004
DARS2	100,0%	100,0%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF17	100,0%	100,0%	Woodhouse-Sakati syndrome, 241080
DDHD1	100,0%	100,0%	Spastic paraplegia 28, autosomal recessive, 609340
DES	100,0%	100,0%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DGUOK	100,0%	100,0%	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%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DLAT	100,0%	100,0%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100,0%	100,0%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	100,0%	100,0%	Paragangliomas 7, 618475

DMAC1	100,0%	100,0%	No OMIM Disease ID
DMAC2	100,0%	100,0%	No OMIM Disease ID
DMAC2L	100,0%	100,0%	No OMIM Disease ID
DNA2	100,0%	100,0%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJA3	100,0%	100,0%	No OMIM Disease ID
DNAJC19	100,0%	100,0%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	100,0%	100,0%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC30	100,0%	100,0%	Leber hereditary optic neuropathy, autosomal recessive, 619382
DNM1L	100,0%	100,0%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
EARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	100,0%	100,0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	100,0%	100,0%	No OMIM Disease ID
EHHADH	100,0%	100,0%	?Fanconi renotubular syndrome 3, 615605
ELAC2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 17, 615440
EPG5	100,0%	100,0%	Vici syndrome, 242840
ERAL1	100,0%	100,0%	Perrault syndrome 6, 617565
ETFDH	100,0%	100,0%	Glutaric acidemia IIC, 231680
ETHE1	100,0%	100,0%	Ethylmalonic encephalopathy, 602473
EXOSC8	100,0%	100,0%	Pontocerebellar hypoplasia, type 1C, 616081
FA2H	100,0%	100,0%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSB	100,0%	100,0%	Rajab interstitial lung disease with brain calcifications 1, 613658
FASTKD2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 44, 618855
FBXL4	100,0%	100,0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX2	100,0%	100,0%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100,0%	100,0%	Auditory neuropathy and optic atrophy, 617717
FH	100,0%	100,0%	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FOXRED1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FTL	100,0%	100,0%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FXN	100,0%	100,0%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300

GARS1	100,0%	100,0%	Spinal muscular atrophy, infantile, James type, 619042 Neuronopathy, distal hereditary motor, type VA, 600794 Charcot-Marie-Tooth disease, type 2D, 601472
GATB	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100,0%	100,0%	Combined oxidative phosphorylation deficiency 42, 618839
GATM	100,0%	100,0%	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBF1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483
GDAP1	100,0%	100,0%	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%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 39, 618397
GLRX5	100,0%	100,0%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	100,0%	100,0%	Hyperinsulinism-hyperammonemia syndrome, 606762
GMPR	100,0%	100,0%	No OMIM Disease ID
GOT2	100,0%	100,0%	Developmental and epileptic encephalopathy 82, 618721
GPT2	100,0%	100,0%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GTPBP2	100,0%	100,0%	Jaberi-Elahi syndrome, 617988
GTPBP3	100,0%	100,0%	Combined oxidative phosphorylation deficiency 23, 616198
HACE1	100,0%	100,0%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADHA	100,0%	100,0%	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	100,0%	100,0%	Trifunctional protein deficiency, 609015
HARS2	100,0%	100,0%	Perrault syndrome 2, 614926
HCCS	100,0%	100,0%	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	100,0%	100,0%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	100,0%	100,0%	Holocarboxylase synthetase deficiency, 253270
HPDL	100,0%	100,0%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027
HSD17B10	100,0%	100,0%	HSD10 mitochondrial disease, 300438

HSPA9	100,0%	100,0%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	100,0%	100,0%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HTRA2	100,0%	100,0%	3-methylglutaconic aciduria, type VIII, 617248
IARS2	100,0%	100,0%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
ISCA1	95,1%	95,1%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100,0%	100,0%	Myopathy with lactic acidosis, hereditary, 255125
KARS1	100,0%	100,0%	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	98,0%	98,0%	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
LACTB	100,0%	100,0%	No OMIM Disease ID
LARS2	100,0%	100,0%	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LDHD	100,0%	100,0%	D-lactic aciduria with susceptibility to gout, 245450
LIAS	100,0%	100,0%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG3	100,0%	100,0%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780
LIPT1	100,0%	100,0%	Lipoyltransferase 1 deficiency, 616299
LIPT2	100,0%	100,0%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LONP1	100,0%	100,0%	CODAS syndrome, 600373
LRPPRC	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LYRM4	66,3%	66,3%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 8, 615838
MAPT	100,0%	100,0%	Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700
MARS2	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390

MCUR1	100,0%	100,0%	No OMIM Disease ID
MDH1	100,0%	100,0%	?Developmental and epileptic encephalopathy 88, 618959
MDH2	100,0%	100,0%	Developmental and epileptic encephalopathy 51, 617339
MECR	100,0%	100,0%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	100,0%	100,0%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100,0%	100,0%	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%	Mitochondrial DNA depletion syndrome 11, 615084
MICOS13	100,0%	100,0%	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	100,0%	100,0%	Myopathy with extrapyramidal signs, 615673
MICU2	100,0%	100,0%	No OMIM Disease ID
MIEF2	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 49, 619024
MIPEP	100,0%	100,0%	Combined oxidative phosphorylation deficiency 31, 617228
MPC1	100,0%	100,0%	Mitochondrial pyruvate carrier deficiency, 614741
MPV17	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRM2	98,9%	98,9%	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 45, 618951
MRPL24	100,0%	100,0%	No OMIM Disease ID
MRPL3	100,0%	100,0%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	100,0%	100,0%	No OMIM Disease ID
MRPL44	100,0%	100,0%	Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	100,0%	100,0%	No OMIM Disease ID
MRPS14	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100,0%	100,0%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	100,0%	100,0%	Ovarian dysgenesis 7, 618117 Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 46, 618952
MRPS25	82,7%	82,7%	?Combined oxidative phosphorylation deficiency 50, 619025
MRPS28	86,6%	86,6%	?Combined oxidative phosphorylation deficiency 47, 618958
MRPS34	100,0%	100,0%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS36	100,0%	100,0%	No OMIM Disease ID
MRPS7	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	100,0%	100,0%	No OMIM Disease ID
MSTO1	100,0%	100,0%	Myopathy, mitochondrial, and ataxia, 617675

MTFMT	100,0%	100,0%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTO1	94,3%	92,1%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	100,0%	100,0%	?Spastic ataxia 4, autosomal recessive, 613672
C12orf65	100,0%	100,0%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
MTX2	100,0%	100,0%	Mandibuloacral dysplasia progeroid syndrome, 619127
NARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NAXD	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFA1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	100,0%	100,0%	?Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	92,2%	88,6%	No OMIM Disease ID
NDUFA4	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 21, 619065
NDUFA5	100,0%	100,0%	No OMIM Disease ID
NDUFA6	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	100,0%	100,0%	No OMIM Disease ID
NDUFA8	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFA9	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	100,0%	100,0%	No OMIM Disease ID
NDUFAF1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 17, 618239 Fanconi renotubular syndrome 5, 618913
NDUFAF7	100,0%	100,0%	No OMIM Disease ID
NDUFAF8	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	100,0%	100,0%	No OMIM Disease ID
NDUFB10	100,0%	100,0%	?Mitochondrial complex I deficiency, nuclear type 35, 619003

NDUFB11	100,0%	99,9%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	100,0%	100,0%	No OMIM Disease ID
NDUFB3	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	100,0%	100,0%	No OMIM Disease ID
NDUFB5	100,0%	100,0%	No OMIM Disease ID
NDUFB6	100,0%	100,0%	No OMIM Disease ID
NDUFB7	100,0%	100,0%	No OMIM Disease ID
NDUFB8	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	98,7%	98,7%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	100,0%	100,0%	No OMIM Disease ID
NDUFC2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 36, 619170
NDUFS1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	95,3%	91,3%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	100,0%	100,0%	No OMIM Disease ID
NDUFS6	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	100,0%	100,0%	No OMIM Disease ID
NFS1	89,5%	89,5%	Combined oxidative phosphorylation deficiency 52, 619386
NFU1	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	100,0%	100,0%	Congenital disorder of deglycosylation 1, 615273
NME3	100,0%	100,0%	No OMIM Disease ID
NR2F1	100,0%	99,8%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NSUN3	100,0%	100,0%	Combined oxidative phosphorylation deficiency 48, 619012
NUBPL	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 21, 618242
OGDH	100,0%	100,0%	?Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	100,0%	100,0%	Yoon-Bellen neurodevelopmental syndrome, 619701
OPA1	100,0%	100,0%	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500 Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896

OPA3	100,0%	100,0%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OTX2	100,0%	100,0%	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%	No OMIM Disease ID
P4HTM	100,0%	100,0%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PANK2	100,0%	100,0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PARS2	100,0%	100,0%	Developmental and epileptic encephalopathy 75, 618437
PC	100,0%	100,0%	Pyruvate carboxylase deficiency, 266150
PDE2A	100,0%	100,0%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDHA1	100,0%	100,0%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100,0%	100,0%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	100,0%	100,0%	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	100,0%	100,0%	No OMIM Disease ID
PDK2	100,0%	100,0%	No OMIM Disease ID
PDK3	100,0%	100,0%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	100,0%	100,0%	No OMIM Disease ID
PDP1	100,0%	100,0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	97,4%	97,4%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PET117	100,0%	100,0%	?Mitochondrial complex IV deficiency, nuclear type 19, 619063
PIGA	100,0%	100,0%	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%	Liberfarb syndrome, 618889
PITRM1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 30, 619405
PLA2G6	92,3%	92,3%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLPBP	100,0%	100,0%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMPCA	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNPLA8	100,0%	100,0%	?Mitochondrial myopathy with lactic acidosis, 251950

PNPT1	100,0%	100,0%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLG	100,0%	100,0%	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%	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%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLRMT	100,0%	100,0%	Combined oxidative phosphorylation deficiency 55, 619743
PPA2	100,0%	100,0%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	100,0%	100,0%	Cardiomyopathy, dilated, 2C, 618189
PRDX3	100,0%	100,0%	No OMIM Disease ID
PRKAA1	100,0%	100,0%	No OMIM Disease ID
PRORP	100,0%	100,0%	Combined oxidative phosphorylation deficiency 54, 619737
PRPS1	100,0%	100,0%	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%	?Combined oxidative phosphorylation deficiency 51, 619057
PTRH2	100,0%	100,0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	100,0%	99,2%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	100,0%	100,0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100,0%	100,0%	Leukodystrophy, hypomyelinating, 10, 616420
PYROXD1	100,0%	100,0%	Myopathy, myofibrillar, 8, 617258
QRSL1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 40, 618835
RARS2	100,0%	100,0%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	100,0%	100,0%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RRM1	100,0%	100,0%	No OMIM Disease ID
RRM2B	100,0%	100,0%	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%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RYR1	100,0%	99,9%	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 Central core disease, 117000 King-Denborough syndrome, 619542 Minicore myopathy with external ophthalmoplegia, 255320
SACS	100,0%	100,0%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	100,0%	100,0%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS2	100,0%	100,0%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SATB2	100,0%	100,0%	Glass syndrome, 612313
SCO1	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100,0%	100,0%	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	100,0%	100,0%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDHA	100,0%	100,0%	Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial complex II deficiency, nuclear type 1, 252011 Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165
SDHAF1	100,0%	100,0%	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHB	100,0%	100,0%	Paragangliomas 4, 115310 Mitochondrial complex II deficiency, nuclear type 4, 619224 Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864
SDHD	80,1%	80,1%	Paragangliomas 1, with or without deafness, 168000 Paraganglioma and gastric stromal sarcoma, 606864 Mitochondrial complex II deficiency, nuclear type 3, 619167 Pheochromocytoma, 171300
SERAC1	100,0%	100,0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	100,0%	100,0%	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	100,0%	100,0%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	98,7%	98,7%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	100,0%	100,0%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A10	100,0%	100,0%	?Mitochondrial DNA depletion syndrome 19, 618972
SLC25A12	100,0%	100,0%	Developmental and epileptic encephalopathy 39, 612949

SLC25A13	100,0%	100,0%	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471
SLC25A19	100,0%	100,0%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A21	100,0%	100,0%	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A22	100,0%	100,0%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99,7%	99,7%	Fontaine progeroid syndrome, 612289
SLC25A26	100,0%	100,0%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	100,0%	100,0%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100,0%	100,0%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	100,0%	100,0%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100,0%	100,0%	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%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	100,0%	100,0%	Neuropathy, hereditary motor and sensory, type VIB, 616505 Pontocerebellar hypoplasia, type 1E, 619303
SLC39A8	100,0%	100,0%	Congenital disorder of glycosylation, type IIn, 616721
SLC52A2	100,0%	100,0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100,0%	100,0%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC8B1	100,0%	100,0%	No OMIM Disease ID
SLIRP	100,0%	100,0%	No OMIM Disease ID
SOD2	100,0%	100,0%	No OMIM Disease ID
SPART	100,0%	100,0%	Troyer syndrome, 275900
SPATA5	100,0%	100,0%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
SPG7	100,0%	100,0%	Spastic paraplegia 7, autosomal recessive, 607259
SPTBN4	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SQOR	100,0%	100,0%	Sulfide:quinone oxidoreductase deficiency, 619221
SQSTM1	100,0%	100,0%	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%	Optic atrophy 13 with retinal and foveal abnormalities, 165510
STAC3	100,0%	100,0%	Myopathy, congenital, Baily-Bloch, 255995
STAT2	100,0%	100,0%	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636

STXBP1	100,0%	100,0%	Developmental and epileptic encephalopathy 4, 612164
SUCLA2	100,0%	99,9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100,0%	100,0%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	100,0%	100,0%	No OMIM Disease ID
SURF1	100,0%	100,0%	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SZT2	100,0%	100,0%	Developmental and epileptic encephalopathy 18, 615476
TACO1	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TAZ	100,0%	100,0%	Barth syndrome, 302060
TANGO2	100,0%	100,0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	100,0%	100,0%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 21, 615918
TBCK	100,0%	100,0%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TDP2	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TFAM	100,0%	100,0%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFB2M	100,0%	100,0%	No OMIM Disease ID
THG1L	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 28, 618800
TIMM22	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	100,0%	100,0%	No OMIM Disease ID
TIMM50	100,0%	100,0%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	100,0%	100,0%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TK2	100,0%	100,0%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TMEM126A	100,0%	100,0%	Optic atrophy 7, 612989
TMEM126B	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM186	100,0%	100,0%	No OMIM Disease ID
TMEM65	99,8%	97,9%	No OMIM Disease ID
TMEM70	100,0%	100,0%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMX2	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TOMM70	100,0%	100,0%	No OMIM Disease ID
TOP3A	100,0%	100,0%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TPK1	100,0%	100,0%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRAPPC2L	100,0%	100,0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRIT1	100,0%	100,0%	Combined oxidative phosphorylation deficiency 35, 617873

TRMT10C	100,0%	100,0%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100,0%	100,0%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	100,0%	100,0%	Liver failure, transient infantile, 613070
TRNT1	100,0%	100,0%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TSFM	94,9%	94,9%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	100,0%	100,0%	Combined oxidative phosphorylation deficiency 4, 610678
TWNK	100,0%	100,0%	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%	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	100,0%	100,0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UFM1	100,0%	100,0%	Leukodystrophy, hypomyelinating, 14, 617899
UQCC1	100,0%	100,0%	No OMIM Disease ID
UQCC2	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100,0%	100,0%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	100,0%	100,0%	No OMIM Disease ID
UQCR11	100,0%	100,0%	No OMIM Disease ID
UQCRB	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	100,0%	100,0%	Parkinsonism with polyneuropathy, 619279
UQCRC2	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRH	100,0%	100,0%	No OMIM Disease ID
UQCRQ	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 20, 615917
VPS13D	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 4, 607317
WARS2	100,0%	100,0%	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%	Neurodegeneration with brain iron accumulation 5, 300894
YARS2	100,0%	100,0%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	100,0%	100,0%	?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.
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 is the chemistry used for WES analysis.

Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors