MITOCHONDRIAL DISORDERS GENE PANEL DG 2.18 (414 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AARS2	100%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABAT	100%	99,40%	100%	100%	GABA-transaminase deficiency, 613163
ACAD9	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACO2	96,30%	90,30%	100%	100%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACTA1	99,60%	92,30%	100%	100%	Myopathy, actin, congenital, with cores, 161800 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 ?Myopathy, scapulohumeroperoneal, 616852
ADAMTS10	99,90%	98,50%	100%	100%	Weill-Marchesani syndrome 1, recessive, 277600
ADCK2	100%	100%	100%	100%	No OMIM disease ID
ADPRHL2	100%	99,80%	100%	100%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	95,00%	91,10%	100%	99,90%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGK	99,90%	97,60%	100%	100%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AIFM1	99,90%	98,80%	100%	100%	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
ALDH1B1	100%	100%	100%	100%	No OMIM disease ID
ALKBH1	100%	99,90%	100%	100%	No OMIM disease ID
ANO10	99,80%	97,90%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOPT1	81,90%	80,70%	93,50%	93,40%	Mitochondrial complex IV deficiency, 220110
APTX	94,90%	92,50%	100%	100%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARL2	100%	100%	100%	100%	No OMIM disease ID
ARNT2	100%	100%	100%	99,60%	?Webb-Dattani syndrome, 615926
ATAD1	99,60%	95,10%	100%	100%	Hyperekplexia 4, 618011

ATAD3A	91,90%	83,20%	100%	100%	Harel-Yoon syndrome, 617183 ?Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	92,10%	79,20%	100%	100%	No OMIM disease ID
ATP13A2	100%	99,50%	100%	100%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP5A1	95,20%	87,60%	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5B	100%	97,80%	100%	100%	No OMIM disease ID
ATP5C1	98,00%	92,20%	100%	100%	No OMIM disease ID
ATP5D	96,20%	89,30%	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	100%	100%	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	98,90%	90,40%	100%	100%	No OMIM disease ID
ATP5G1	100%	99,90%	100%	100%	No OMIM disease ID
ATP5G2	100%	98,50%	100%	100%	No OMIM disease ID
ATP5G3	100%	100%	100%	100%	No OMIM disease ID
ATP5H	97,30%	80,90%	100%	100%	No OMIM disease ID
ATP5I	100%	100%	100%	100%	No OMIM disease ID
ATP5J	100%	97,50%	100%	100%	No OMIM disease ID
ATP5J2	100%	99,40%	100%	100%	No OMIM disease ID
ATP5L	100%	100%	100%	100%	No OMIM disease ID
ATP5L2	100%	100%	100%	100%	No OMIM disease ID
ATP5O	99,90%	98,00%	100%	100%	No OMIM disease ID
ATP5S	100%	100%	100%	100%	No OMIM disease ID
ATPAF1	78,30%	70,00%	100%	100%	No OMIM disease ID
ATPAF2	100%	100%	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATPIF1	100%	100%	100%	100%	No OMIM disease ID
BCAP31	92,60%	83,20%	100%	99,90%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100%	100%	100%	100%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BOLA1	100%	100%	100%	100%	No OMIM disease ID
BOLA2	100%	100%	100%	100%	No OMIM disease ID
BOLA3	99,40%	90,20%	100%	100%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
C12orf65	99,80%	98,50%	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559

C19orf12	100%	99,80%	100%	100%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C19orf70	100%	99,70%	100%	99,70%	Combined oxidative phosphorylation deficiency 37, 618329
C1QBP	86,90%	77,30%	100%	100%	Combined oxidative phosphorylation deficiency 33, 617713
CA5A	99,70%	97,10%	100%	100%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CARS2	100%	100%	100%	99,20%	Combined oxidative phosphorylation deficiency 27, 616672
CEP89	96,00%	94,50%	100%	100%	No OMIM disease ID
CHCHD10	59,10%	43,90%	100%	100%	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHCHD2	98,40%	83,80%	100%	100%	Parkinson disease 22, autosomal dominant, 616710
CHKB	100%	99,70%	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CISD2	83,40%	83,40%	100%	100%	Wolfram syndrome 2, 604928
CLPB	100%	99,90%	100%	100%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	100%	99,10%	100%	100%	Perrault syndrome 3, 614129
COA1	100%	100%	100%	100%	No OMIM disease ID
COA3	100%	100%	100%	100%	No OMIM disease ID
COA5	99,10%	88,90%	85,20%	85,20%	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	99,90%	98,40%	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COASY	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COQ2	98,00%	95,30%	97,20%	97,20%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	90,90%	89,30%	100%	100%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	100%	100%	100%	100%	No OMIM disease ID
COQ6	99,90%	98,40%	100%	100%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	99,80%	100%	100%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	100%	99,50%	100%	100%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100%	99,30%	100%	100%	Nephrotic syndrome, type 9, 615573
COQ9	100%	97,90%	100%	100%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100%	100%	100%	100%	Mitochondrial complex IV deficiency, 220110 Leigh syndrome due to mitochondrial COX4 deficiency, 256000
COX14	100%	100%	100%	100%	?Mitochondrial complex IV deficiency, 220110
COX15	99,90%	98,80%	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	97,80%	88,30%	100%	100%	Mitochondrial complex IV deficiency, 220110
COX4I1	100%	100%	100%	100%	No OMIM disease ID

COX4I2	100%	100%	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	74,70%	47,10%	100%	100%	No OMIM disease ID
COX5B	100%	100%	100%	100%	No OMIM disease ID
COX6A1	100%	99,50%	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	99,20%	93,70%	100%	100%	Mitochondrial complex IV deficiency, 220110
COX6B1	100%	100%	100%	100%	Mitochondrial complex IV deficiency, 220110
COX6B2	100%	99,80%	100%	100%	No OMIM disease ID
COX6C	100%	97,40%	100%	100%	No OMIM disease ID
COX7A1	100%	99,90%	100%	100%	No OMIM disease ID
COX7A2	100%	99,80%	100%	100%	No OMIM disease ID
COX7B	77,40%	48,80%	100%	100%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	100%	100%	100%	100%	No OMIM disease ID
COX7C	99,30%	86,90%	100%	100%	No OMIM disease ID
COX8A	100%	100%	100%	100%	?Mitochondrial complex IV deficiency, 220110
COX8C	100%	99,90%	100%	100%	No OMIM disease ID
CP	94,80%	88,90%	100%	100%	Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CRAT	100%	99,80%	100%	100%	?Neurodegeneration with brain iron accumulation 8, 617917
CTBP1	93,20%	86,90%	99,50%	98,60%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CYC1	97,50%	89,20%	99,90%	98,70%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	99,10%	94,90%	100%	100%	Thrombocytopenia 4, 612004
DARS2	100%	99,30%	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF17	98,90%	93,30%	100%	100%	Woodhouse-Sakati syndrome, 241080
DDHD1	97,90%	95,80%	100%	100%	Spastic paraplegia 28, autosomal recessive, 609340
DES	100%	99,70%	100%	100%	?Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DGUOK	100%	99,40%	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	99,90%	98,90%	100%	100%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	100%	99,70%	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100%	99,70%	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	96,70%	90,30%	100%	100%	Paragangliomas 7, 618475
DMAC1	100%	100%	100%	100%	No OMIM disease ID
DMAC2	98,30%	98,30%	100%	100%	No OMIM disease ID

DNA2	99,80%	98,30%	100%	100%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJA3	98,60%	96,00%	100%	100%	No OMIM disease ID
DNAJC19	98,90%	96,20%	100%	100%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	100%	99,70%	100%	100%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	99,90%	98,50%	100%	100%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
EARS2	99,80%	97,70%	100%	100%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	99,90%	99,00%	100%	100%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	100%	99,90%	100%	100%	No OMIM disease ID
EHHADH	100%	100%	100%	100%	?Fanconi renotubular syndrome 3, 615605
ELAC2	100%	99,70%	100%	100%	Combined oxidative phosphorylation deficiency 17, 615440
ERAL1	100%	99,70%	100%	100%	Perrault syndrome 6, 617565
ETFDH	100%	99,80%	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	99,90%	97,40%	100%	100%	Ethylmalonic encephalopathy, 602473
FA2H	92,00%	83,10%	100%	100%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	100%	100%	100%	100%	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FARSB	98,80%	94,60%	100%	100%	Rajab interstitial lung disease with brain calcifications, 613658
FASTKD2	99,80%	98,90%	100%	100%	?Mitochondrial complex IV deficiency, 220110
FBXL4	100%	100%	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX2	100%	100%	100%	100%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100%	99,30%	100%	100%	Auditory neuropathy and optic atrophy, 617717
FH	92,10%	88,30%	100%	100%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FTL	98,50%	89,40%	100%	100%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
FXN	95,50%	80,10%	100%	100%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GARS	99,90%	99,10%	100%	100%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuronopathy, distal hereditary motor, type VA, 600794
GATB	100%	99,70%	100%	100%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100%	100%	100%	100%	Combined oxidative phosphorylation deficiency 42, 618839
GATM	100%	100%	100%	100%	Cerebral creatine deficiency syndrome 3, 612718

GFER	99,60%	93,90%	100%	100%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	99,90%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	98,90%	95,20%	100%	100%	Combined oxidative phosphorylation deficiency 39, 618397
GLRX5	97,30%	89,10%	99,60%	95,40%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	94,20%	82,90%	100%	100%	Hyperinsulinism-hyperammonemia syndrome, 606762
GMPR	100%	100%	100%	100%	No OMIM disease ID
GOT2	97,50%	90,90%	100%	100%	Epileptic encephalopathy, early infantile, 82, 618721
GPT2	99,20%	93,60%	100%	99,80%	Mental retardation, autosomal recessive 49, 616281
GTPBP2	100%	99,30%	100%	99,90%	Jaberi-Elahi syndrome, 617988
GTPBP3	100%	99,80%	100%	100%	Combined oxidative phosphorylation deficiency 23, 616198
HACE1	100%	99,30%	100%	100%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADHA	97,10%	91,30%	100%	100%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HADHB	98,80%	89,70%	100%	100%	Trifunctional protein deficiency, 609015
HARS2	100%	100%	100%	100%	?Perrault syndrome 2, 614926
HCCS	99,80%	97,60%	100%	100%	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	98,20%	88,50%	100%	100%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	100%	100%	100%	100%	Holocarboxylase synthetase deficiency, 253270
HSD17B10	100%	99,10%	100%	100%	HSD10 mitochondrial disease, 300438
HSPA9	88,50%	84,50%	100%	100%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	98,80%	93,70%	100%	100%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HTRA2	100%	99,90%	100%	100%	3-methylglutaconic aciduria, type VIII, 617248
IARS2	100%	99,90%	100%	100%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	93,70%	90,10%	100%	100%	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
MRM2	100%	99,50%	99,00%	99,00%	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	100%	98,20%	100%	100%	No OMIM disease ID
MRPL3	93,20%	87,20%	100%	100%	Combined oxidative phosphorylation deficiency 9, 614582

MRPL40	99,90%	96,10%	100%	100%	No OMIM disease ID
MRPL44	99,90%	98,70%	100%	100%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	100%	99,80%	100%	100%	No OMIM disease ID
MRPS14	100%	100%	100%	100%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100%	99,60%	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	99,60%	96,90%	100%	100%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	99,80%	99,10%	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS23	100%	99,60%	100%	100%	No OMIM disease ID
MRPS25	100%	99,90%	82,70%	82,70%	No OMIM disease ID
MRPS28	89,90%	86,80%	86,60%	86,60%	No OMIM disease ID
MRPS34	97,60%	92,00%	100%	100%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS36	95,20%	77,60%	100%	100%	No OMIM disease ID
MRPS7	100%	100%	100%	100%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	100%	100%	100%	100%	No OMIM disease ID
ISCA1	94,20%	85,90%	95,10%	95,10%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100%	98,80%	100%	100%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100%	100%	100%	100%	Myopathy with lactic acidosis, hereditary, 255125
KARS	100%	99,90%	100%	100%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KIF1A	99,40%	97,10%	100%	100%	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
LACTB	99,50%	92,60%	100%	100%	No OMIM disease ID

LARS2	100%	100%	100%	100%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LIAS	100%	99,10%	100%	100%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT1	100%	99,90%	100%	100%	Lipoyltransferase 1 deficiency, 616299
LIPT2	94,90%	75,20%	100%	100%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LONP1	100%	99,80%	100%	100%	CODAS syndrome, 600373
LRPPRC	99,90%	99,10%	100%	100%	Leigh syndrome, French-Canadian type, 220111
LYRM4	68,50%	66,20%	66,30%	66,30%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	95,90%	86,20%	100%	100%	Mitochondrial complex III deficiency, nuclear type 8, 615838
MARS2	100%	100%	100%	100%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MCUR1	99,50%	91,50%	100%	99,80%	No OMIM disease ID
MDH1	100%	99,40%	100%	100%	No OMIM disease ID
MDH2	98,00%	97,90%	100%	100%	Epileptic encephalopathy, early infantile, 51, 617339
MECR	100%	98,90%	100%	100%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	94,30%	89,90%	100%	100%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100%	99,90%	100%	100%	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MGME1	100%	100%	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MICU1	98,90%	95,20%	100%	100%	Myopathy with extrapyramidal signs, 615673
MICU2	97,20%	91,80%	100%	100%	No OMIM disease ID
MIEF2	100%	99,00%	100%	100%	No OMIM disease ID
MIPEP	99,20%	96,50%	100%	100%	Combined oxidative phosphorylation deficiency 31, 617228
MPC1	100%	99,60%	100%	100%	Mitochondrial pyruvate carrier deficiency, 614741
MPV17	100%	97,20%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MSTO1	99,60%	96,70%	100%	100%	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	100%	99,80%	100%	100%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTO1	91,30%	90,40%	91,60%	91,40%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	99,50%	96,10%	100%	100%	?Spastic ataxia 4, autosomal recessive, 613672
NARS2	98,30%	97,40%	100%	100%	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NAXD	100%	100%	100%	100%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100%	99,80%	100%	100%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFA1	99,90%	99,30%	100%	100%	Mitochondrial complex I deficiency, nuclear type 12, 301020

NDUFA10	99,80%	98,60%	100%	100%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	100%	100%	100%	99,80%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	100%	100%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	92,20%	89,20%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	100%	100%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	89,10%	87,90%	88,00%	87,90%	No OMIM disease ID
NDUFA4	100%	96,40%	100%	100%	No OMIM disease ID
NDUFA5	96,50%	81,80%	100%	100%	No OMIM disease ID
NDUFA6	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	100%	99,90%	100%	100%	No OMIM disease ID
NDUFA8	100%	99,00%	100%	100%	No OMIM disease ID
NDUFA9	99,90%	96,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	99,60%	94,90%	100%	100%	No OMIM disease ID
NDUFAF1	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	95,00%	83,40%	100%	99,90%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	99,80%	98,20%	100%	100%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100%	96,80%	100%	100%	Mitochondrial complex I deficiency, nuclear type 17, 618239
NDUFAF7	100%	99,80%	100%	100%	No OMIM disease ID
NDUFAF8	62,60%	61,10%	100%	99,60%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	78,20%	57,80%	100%	100%	No OMIM disease ID
NDUFB10	100%	100%	100%	100%	No OMIM disease ID
NDUFB11	99,50%	96,50%	100%	99,50%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	100%	100%	100%	100%	No OMIM disease ID
NDUFB3	95,80%	80,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	90,20%	85,20%	100%	100%	No OMIM disease ID
NDUFB5	100%	100%	100%	100%	No OMIM disease ID
NDUFB6	98,40%	88,40%	100%	100%	No OMIM disease ID
NDUFB7	99,80%	94,80%	100%	100%	No OMIM disease ID
NDUFB8	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	98,40%	95,50%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	99,60%	99,40%	100%	100%	No OMIM disease ID
NDUFC2	99,80%	96,00%	100%	100%	No OMIM disease ID
NDUFS1	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 6, 618228

NDUFS3	90,70%	90,60%	91,90%	90,70%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	99,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	100%	100%	100%	100%	No OMIM disease ID
NDUFS6	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100%	99,20%	100%	100%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	99,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	98,00%	96,10%	100%	100%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	86,90%	76,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	99,80%	98,00%	100%	100%	No OMIM disease ID
NFS1	87,90%	84,00%	89,50%	89,50%	No OMIM disease ID
NFU1	98,80%	90,80%	100%	100%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	100%	99,80%	100%	100%	Congenital disorder of deglycosylation, 615273
NME3	93,10%	88,50%	100%	100%	No OMIM disease ID
NR2F1	100%	100%	99,10%	95,10%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NSUN3	100%	100%	100%	100%	No OMIM disease ID
NUBPL	99,70%	98,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 21, 618242
OGDH	100%	99,90%	100%	100%	No OMIM disease ID
OPA1	99,70%	97,60%	100%	100%	Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	100%	99,00%	100%	100%	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OTX2	100%	99,70%	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
OXA1L	100%	99,80%	100%	100%	No OMIM disease ID
PANK2	100%	99,30%	100%	100%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PARS2	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 75, 618437
PC	99,80%	97,30%	100%	100%	Pyruvate carboxylase deficiency, 266150
PDHA1	99,40%	97,10%	100%	100%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	99,10%	97,50%	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	99,90%	99,40%	100%	100%	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	99,90%	98,50%	100%	99,90%	No OMIM disease ID
PDK2	100%	100%	100%	100%	No OMIM disease ID
PDK3	99,50%	97,20%	100%	100%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905

PDK4	100%	99,30%	100%	100%	No OMIM disease ID
PDP1	100%	100%	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	94,70%	87,60%	97,30%	96,60%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	99,80%	97,10%	100%	100%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	100%	99,60%	100%	100%	Mitochondrial complex IV deficiency, 220110
PET117	100%	100%	100%	100%	No OMIM disease ID
TACO1	98,40%	93,00%	100%	100%	Mitochondrial complex IV deficiency, 220110
TANGO2	100%	99,30%	100%	100%	metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	99,50%	97,90%	100%	100%	No OMIM disease ID
TARS2	100%	99,30%	100%	100%	?Combined oxidative phosphorylation deficiency 21, 615918
TAZ	99,20%	96,50%	100%	100%	Barth syndrome, 302060
PIGA	93,80%	86,70%	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PISD	100%	100%	100%	100%	No OMIM disease ID
PITRM1	98,40%	96,10%	100%	100%	No OMIM disease ID
PLA2G6	99,90%	98,30%	100%	100%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLPBP	98,20%	90,10%	100%	100%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMPCA	97,70%	94,20%	100%	100%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100%	99,70%	100%	100%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNPLA8	100%	99,80%	100%	100%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPT1	97,70%	89,70%	100%	100%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLG	100%	99,30%	100%	100%	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	99,60%	98,00%	100%	99,90%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528
POLR2A	100%	100%	100%	100%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
PPA2	98,70%	94,00%	100%	100%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	99,80%	99,50%	100%	100%	Cardiomyopathy, dilated, 2C, 618189
PRKAA1	100%	99,50%	100%	100%	No OMIM disease ID

PTCD3	99,20%	97,60%	100%	100%	No OMIM disease ID
PTRH2	100%	100%	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	100%	99,50%	99,60%	97,20%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	99,90%	97,70%	100%	100%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100%	99,10%	100%	100%	Leukodystrophy, hypomyelinating, 10, 616420
PYROXD1	95,20%	83,90%	100%	100%	Myopathy, myofibrillar, 8, 617258
QRSL1	99,20%	93,90%	100%	100%	Combined oxidative phosphorylation deficiency 40, 618835
RARS2	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	100%	98,60%	100%	100%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	98,50%	95,30%	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RRM1	100%	99,50%	100%	100%	No OMIM disease ID
RRM2B	100%	99,70%	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
RTN4IP1	99,90%	98,70%	100%	100%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RYR1	96,90%	93,90%	99,40%	99,00%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
SACS	100%	100%	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	100%	99,60%	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS2	95,80%	94,60%	100%	100%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	97,10%	93,80%	100%	100%	Mitochondrial complex IV deficiency, 220110
SCO2	100%	100%	100%	100%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SCP2	100%	99,20%	100%	100%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDHA	85,80%	80,40%	100%	100%	Leigh syndrome, 256000 Paragangliomas 5, 614165 Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial respiratory chain complex II deficiency, 252011
SDHAF1	99,90%	93,20%	100%	100%	Mitochondrial complex II deficiency, 252011
SDHB	100%	100%	100%	100%	Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paragangliomas 4, 115310 Paraganglioma and gastric stromal sarcoma, 606864

SDHD	54,00%	51,60%	80,10%	80,10%	Paragangliomas 1, with or without deafness, 168000 Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SERAC1	99,90%	99,50%	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	99,90%	98,90%	100%	100%	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	100%	99,70%	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	100%	99,80%	98,70%	98,70%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	95,80%	88,60%	99,50%	97,80%	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A10	76,20%	69,30%	100%	100%	No OMIM disease ID
SLC25A12	99,90%	99,50%	100%	100%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	100%	99,70%	100%	100%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	100%	98,50%	100%	100%	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 Microcephaly, Amish type, 607196
SLC25A21	100%	99,70%	100%	100%	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A22	98,60%	95,80%	100%	100%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	99,40%	99,30%	99,80%	99,80%	Fontaine progeroid syndrome, 612289
SLC25A3	99,80%	98,00%	100%	100%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100%	100%	100%	100%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	99,70%	97,10%	100%	100%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100%	100%	100%	100%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A42	96,50%	93,20%	100%	100%	metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	99,70%	97,30%	100%	100%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC39A8	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC52A2	100%	100%	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SOD2	100%	100%	100%	100%	No OMIM disease ID
SPART	99,70%	96,80%	100%	100%	Troyer syndrome, 275900
SPATA5	100%	99,70%	100%	100%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG7	94,90%	92,60%	100%	100%	Spastic paraplegia 7, autosomal recessive, 607259

SQOR	100%	97,80%	100%	100%	No OMIM disease ID
SQSTM1	98,80%	95,50%	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
SSBP1	99,80%	97,60%	100%	100%	No OMIM disease ID
STAC3	100%	100%	100%	100%	Myopathy, congenital, Baily-Bloch, 255995
STAT2	100%	99,90%	100%	100%	Immunodeficiency 44, 616636
STXBP1	96,80%	96,50%	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	94,30%	86,60%	100%	100%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	99,90%	99,80%	100%	100%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	96,70%	86,30%	100%	100%	No OMIM disease ID
SURF1	89,40%	88,20%	100%	100%	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684
SZT2	99,60%	99,50%	100%	99,90%	Epileptic encephalopathy, early infantile, 18, 615476
TDP2	100%	99,40%	100%	100%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TFB2M	100%	99,10%	100%	100%	No OMIM disease ID
THG1L	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 28, 618800
TIMM22	100%	99,70%	100%	100%	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	100%	99,40%	100%	100%	No OMIM disease ID
TIMM50	98,30%	94,40%	100%	100%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	98,00%	90,10%	100%	100%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TK2	99,20%	96,30%	100%	100%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TMEM126A	96,30%	84,40%	100%	100%	Optic atrophy 7, 612989
TMEM126B	99,80%	97,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM186	100%	100%	100%	100%	No OMIM disease ID
TMEM65	88,00%	81,30%	92,50%	85,30%	No OMIM disease ID
TMEM70	98,00%	93,90%	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMX2	100%	99,80%	100%	100%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TOMM70	100%	99,80%	100%	100%	No OMIM disease ID
TOP3A	100%	98,70%	100%	100%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TPK1	99,80%	99,00%	100%	100%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458

TRAPPC2L	100%	100%	100%	100%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRIT1	100%	100%	100%	100%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT10C	100%	100%	100%	99,90%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100%	99,30%	100%	100%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	100%	100%	100%	99,90%	Liver failure, transient infantile, 613070
TRNT1	99,50%	96,50%	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TSFM	100%	99,50%	94,90%	94,90%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	81,50%	73,80%	100%	99,20%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	100%	99,00%	100%	100%	Combined oxidative phosphorylation deficiency 4, 610678
TWNK	100%	100%	100%	100%	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%	100%	100%	100%	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	100%	97,00%	100%	100%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC1	100%	99,90%	100%	100%	No OMIM disease ID
UQCC2	100%	99,70%	100%	100%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100%	98,70%	100%	100%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	100%	100%	100%	100%	No OMIM disease ID
UQCR11	100%	100%	100%	100%	No OMIM disease ID
UQCRB	99,40%	95,10%	100%	100%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	99,80%	98,40%	100%	100%	No OMIM disease ID
UQCRC2	99,90%	99,30%	100%	100%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	91,90%	84,90%	100%	100%	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRH	100%	98,20%	100%	100%	No OMIM disease ID
UQCRQ	100%	100%	100%	100%	Mitochondrial complex III deficiency, nuclear type 4, 615159
USMG5	82,90%	42,80%	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
VARS2	100%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 20, 615917
VPS13D	100%	99,70%	100%	100%	Spinocerebellar ataxia, autosomal recessive 4, 607317
WARS2	100%	99,40%	100%	100%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WDR45	96,40%	89,70%	100%	100%	Neurodegeneration with brain iron accumulation 5, 300894
YARS2	100%	99,80%	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	99,00%	95,20%	100%	100%	?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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors