Mitokondriesykdommer

Genpanel, versjon v02

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
>x10 Andel av genet som har blitt lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

Gen	Transkript	>10x	Fenotype
AARS	NM_001605.2	100%	Epileptic encephalopathy, early infantile, 29 OMIM PubMed Charcot-Marie-Tooth disease, axonal, type 2N OMIM
AARS2	NM_020745.3	100%	Combined oxidative phosphorylation deficiency 8 OMIM
ABCB7	NM_004299.4	100%	Cerebellar ataxia with or without sideroblastic anemia OMIM
ACAD9	NM_014049.4	100%	Mitochondrial complex I deficiency OMIM
ACO2	NM_001098.2	97%	Infantile cerebellar-retinal degeneration OMIM
ADCK3	NM_020247.4	100%	Coenzyme Q10 deficiency, primary, 4 OMIM
ADCK4	NM_024876.3	100%	Nephrotic syndrome, type 9, with or without seizures, mild mental retardation, retinitis pigmentosa OMIM PubMed
AFG3L2	NM_006796.2	98%	Spinocerebellar ataxia 28 OMIM Ataxia, spastic, 5, autosomal recessive OMIM
AGK	NM_018238.3	100%	Sengers syndrome OMIM
AIFM1	NM_004208.3	100%	Cowchock syndrome OMIM Combined oxidative phosphorylation deficiency 6 OMIM Spondyloepimetaphyseal dysplasia with neurodegeneration PubMed
ANO10	NM_018075.3	100%	Spinocerebellar ataxia, autosomal recessive 10 OMIM
APOPT1	NM_032374.4	100%	Mitochondrial complex IV deficiency OMIM
APTX	NM_175073.2	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia OMIM
ATP5A1	NM_001001937.1	98%	Mitochondrial complex (ATP synthase) deficiency, nuclear type 4 OMIM Combined oxidative phosphorylation deficiency 22 OMIM
ATP5E	NM_006886.3	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3 OMIM

Gen	Transkript	>10x	Fenotype
ATPAF2	NM_145691.3	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1 OMIM
BCS1L	NM_004328.4	100%	Mitochondrial complex III deficiency, nuclear type 1 OMIM Leigh syndrome OMIM GRACILE syndrome OMIM Bjornstad syndrome OMIM
BOLA3	NM_212552.2	97%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia OMIM
C10orf2	NM_021830.4	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions OMIM Perrault syndrome 5 OMIM Mitochondrial DNA depletion syndrome 7 (hepatocerebral type) OMIM
C12orf65	NM_152269.4	100%	Spastic paraplegia 55, autosomal recessive OMIM Combined oxidative phosphorylation deficiency 7 OMIM
C19orf12	NM_001031726.3	100%	Neurodegeneration with brain iron accumulation 4 OMIM Spastic paraplegia 43, autosomal recessive OMIM
CA5A	NM_001739.1	100%	Hyperammonemia due to carbonic anhydrase VA deficiency OMIM
CARS2	NM_024537.2	100%	Combined oxidative phosphorylation deficiency 27 OMIM
CEP89	NM_032816.4	100%	Mitochondrial complex IV deficiency PubMed
CHCHD10	NM_213720.2	90%	Spinal muscular atrophy, Jokela type OMIM Myopathy, isolated mitochondrial, autosomal dominant OMIM
CHKB	NM_005198.4	100%	Muscular dystrophy, congenital, megaconial type OMIM
CISD2	NM_001008388.4	77%	Wolfram syndrome 2 OMIM
CLPB	NM_001258394.1	100%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia OMIM
CLPP	NM_006012.2	100%	Perrault syndrome 3 OMIM
COA3	NM_001040431.2	100%	Mitochondrial complex IV deficiency PubMed
COA5	NM_001008215.2	100%	Cardioencephalomyopathy, fatal infantile OMIM
COA6	NM_001206641.2	100%	Cardioencephalomyopathy, fatal infantile OMIM
COASY	NM_025233.6	100%	Neurodegeneration with brain iron accumulation 6 OMIM

Gen	Transkript	>10x	Fenotype
COQ2	NM_015697.7	100%	Coenzyme Q10 deficiency, primary, 1 OMIM
COQ4	NM_016035.3	100%	Coenzyme Q10 deficiency, primary, 7 OMIM
COQ6	NM_182476.2	100%	Coenzyme Q10 deficiency, primary, 6 OMIM
COQ9	NM_020312.3	100%	Coenzyme Q10 deficiency, primary, 5 OMIM
COX10	NM_001303.3	100%	Mitochondrial complex IV deficiency OMIM Leigh syndrome OMIM
COX14	NM_032901.3	100%	Mitochondrial complex IV deficiency OMIM
COX15	NM_004376.5	100%	Leigh syndrome OMIM Cardioencephalomyopathy, fatal infantile OMIM
COX20	NM_198076.4	99%	Mitochondrial complex IV deficiency OMIM
COX4I2	NM_032609.2	100%	Exocrine pancreatic insufficiency, dyserythropoeitic anemia and calvarial hyperostosis PubMed
COX6A1	NM_004373.3	100%	Charcot-Marie-Tooth disease, recessive intermediate D OMIM
COX6B1	NM_001863.4	100%	Mitochondrial complex IV deficiency OMIM
COX7B	NM_001866.2	84%	Linear skin defects with multiple congenital anomalies OMIM
COX8A	NM_004074.2	100%	Mitochondrial complex IV deficiency OMIM
CYC1	NM_001916.4	100%	Mitochondrial complex III deficiency, nuclear type 6 OMIM
DARS	NM_001349.3	100%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity OMIM
DARS2	NM_018122.4	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation OMIM
DCAF17	NM_025000.3	100%	Woodhouse-Sakati syndrome OMIM
DGUOK	NM_080916.2	100%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type) OMIM

Gen	Transkript	>10x	Fenotype
DHTKD1	NM_018706.6	100%	Charcot-Marie-Tooth disease, axonal, type 2Q OMIM 2-aminoadipic 2-oxoadipic aciduria OMIM
DLAT	NM_001931.4	100%	Pyruvate dehydrogenase E2 deficiency OMIM
DLD	NM_000108.4	100%	Dihydrolipoamide dehydrogenase deficiency OMIM
DNA2	NM_001080449.2	100%	Seckel syndrome 8 OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions OMIM
DNAJC19	NM_145261.3	100%	3-methylglutaconic aciduria, type V OMIM
DNM1L	NM_012062.4	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission OMIM
EARS2	NM_001083614.1	99%	Combined oxidative phosphorylation deficiency 12 OMIM
ECHS1	NM_004092.3	100%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency OMIM
ELAC2	NM_018127.6	100%	Combined oxidative phosphorylation deficiency 17 OMIM
EPG5	NM_020964.2	99%	Vici syndrome OMIM
ETHE1	NM_014297.3	100%	Ethylmalonic encephalopathy OMIM
FARS2	NM_006567.3	100%	Combined oxidative phosphorylation deficiency 14 OMIM
FASTKD2	NM_014929.3	100%	Mitochondrial complex IV deficiency OMIM
FBXL4	NM_012160.4	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type) OMIM
FDX1L	NM_001031734.3	100%	Mitochondrial myopathy with lactic acidosis PubMed
FOXRED1	NM_017547.3	100%	Mitochondrial complex I deficiency OMIM Leigh syndrome OMIM
FTL	NM_000146.3	100%	Neurodegeneration with brain iron accumulation 3 OMIM
GARS	NM_002047.2	100%	Neuropathy, distal hereditary motor, type VA OMIM Charcot-Marie-Tooth disease, type 2D OMIM

Gen	Transkript	>10x	Fenotype
GDAP1	NM_018972.2	100%	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis OMIM Charcot-Marie-Tooth disease, axonal, type 2K OMIM Charcot-Marie-Tooth disease, type 4A OMIM Charcot-Marie-Tooth disease, recessive intermediate, A OMIM
GFER	NM_005262.2	100%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay OMIM
GFM1	NM_024996.5	100%	Combined oxidative phosphorylation deficiency 1 OMIM
GFM2	NM_032380.4	100%	Leigh syndrome with arthrogryposis multiplex congenita PubMed
GLRX5	NM_016417.2	99%	Spasticity, childhood-onset, with hyperglycinemia OMIM
GLUD1	NM_005271.3	98%	Hyperinsulinism-hyperammonemia syndrome OMIM
GTPBP3	NM_133644.3	100%	Combined oxidative phosphorylation deficiency 23 OMIM
HARS2	NM_012208.3	100%	Perrault syndrome 2 OMIM
HCCS	NM_005333.4	100%	Linear skin defects with multiple congenital anomalies 1 OMIM
HIBCH	NM_014362.3	99%	3-hydroxyisobutryl-CoA hydrolase deficiency OMIM
HLCS	NM_000411.6	100%	Holocarboxylase synthetase deficiency OMIM
HSPA9	NM_004134.6	98%	Even-plus syndrome OMIM
HSPD1	NM_002156.4	99%	Leukodystrophy, hypomyelinating, 4 OMIM Spastic paraplegia 13, autosomal dominant OMIM
IARS2	NM_018060.3	100%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia OMIM
IBA57	NM_001010867.2	99%	Spastic paraplegia 74, autosomal recessive OMIM Multiple mitochondrial dysfunctions syndrome 3 OMIM
IDH2	NM_002168.3	100%	D-2-hydroxyglutaric aciduria 2 OMIM
ISCA2	NM_194279.3	99%	Multiple mitochondrial dysfunctions syndrome 4 OMIM
ISCU	NM_213595.3	100%	Myopathy with lactic acidosis, hereditary OMIM

Gen	Transkript	>10x	Fenotype
KARS	NM_001130089.1	100%	Charcot-Marie-Tooth disease, recessive intermediate, B OMIM
LARS2	NM_015340.3	100%	Perrault syndrome 4 OMIM
LIAS	NM_006859.3	100%	Hyperglycinemia, lactic acidosis, and seizures OMIM
LIPT1	NM_145199.2	100%	Lipoyltransferase 1 deficiency OMIM
LONP1	NM_001276480.1	100%	CODAS syndrome OMIM
LRPPRC	NM_133259.3	99%	Leigh syndrome, French-Canadian type OMIM
LYRM4	NM_020408.5	100%	Combined oxidative phosphorylation deficiency 19 OMIM
LYRM7	NM_181705.3	100%	Mitochondrial complex III deficiency, nuclear type 8 OMIM
MARS2	NM_138395.3	100%	Spastic ataxia 3, autosomal recessive OMIM Combined oxidative phosphorylation deficiency 25 OMIM
MFF	NM_020194.5	98%	Developmental delay, microcephaly, lactic acidosis, basal ganglia disease PubMed
MFN2	NM_014874.3	100%	Hereditary motor and sensory neuropathy VIA OMIM Charcot-Marie-Tooth disease, type 2A2 OMIM
MGME1	NM_052865.2	100%	Mitochondrial DNA depletion syndrome 11 OMIM
MICU1	NM_006077.3	100%	Myopathy with extrapyramidal signs OMIM
MPC1	NM_016098.3	100%	Mitochondrial pyruvate carrier deficiency OMIM
MPV17	NM_002437.4	100%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) OMIM
MRPL3	NM_007208.3	97%	Combined oxidative phosphorylation deficiency 9 OMIM
MRPL44	NM_022915.3	100%	Combined oxidative phosphorylation deficiency 16 OMIM
MRPS16	NM_016065.3	100%	Combined oxidative phosphorylation deficiency 2 OMIM

Gen	Transkript	>10x	Fenotype
MRPS22	NM_020191.2	100%	Combined oxidative phosphorylation deficiency 5 OMIM
MRPS23	NM_016070.3	100%	Combined oxidative phosphorylation deficiency PubMed
MRPS7	NM_015971.3	100%	Combined oxidative phosphorylation deficiency PubMed
MTFMT	NM_139242.3	100%	Combined oxidative phosphorylation deficiency 15 OMIM
MTO1	NM_012123.3	99%	Combined oxidative phosphorylation deficiency 10 OMIM
MTPAP	NM_018109.3	100%	Ataxia, spastic, 4 OMIM
NADK2	NM_001287340.1	100%	2,4-dienoyl-CoA reductase (DECR) deficiency OMIM
NARS2	NM_024678.5	97%	Combined oxidative phosphorylation deficiency 24 OMIM
NDUFA1	NM_004541.3	100%	Mitochondrial complex I deficiency OMIM
NDUFA10	NM_004544.3	99%	Leigh syndrome OMIM
NDUFA11	NM_175614.4	100%	Mitochondrial complex I deficiency OMIM
NDUFA12	NM_018838.4	100%	Leigh syndrome due to mitochondrial complex 1 deficiency OMIM
NDUFA13	NM_015965.6	100%	Mitochondrial complex I deficiency PubMed
NDUFA2	NM_002488.4	100%	Leigh syndrome due to mitochondrial complex I deficiency OMIM
NDUFA4	NM_002489.3	100%	Mitochondrial complex IV deficiency, Leigh syndrome PubMed
NDUFA8	NM_014222.2	100%	Combined oxidative phosphorylation deficiency PubMed
NDUFA9	NM_005002.4	100%	Leigh syndrome due to mitochondrial complex I deficiency OMIM
NDUFAF1	NM_016013.3	100%	Mitochondrial complex I deficiency OMIM

Gen	Transkript	>10x	Fenotype
NDUFAF2	NM_174889.4	95%	Mitochondrial complex I deficiency OMIM Leigh syndrome OMIM
NDUFAF3	NM_199069.1	100%	Mitochondrial complex I deficiency OMIM
NDUFAF4	NM_014165.3	100%	Mitochondrial complex I deficiency OMIM
NDUFAF5	NM_024120.4	100%	Mitochondrial complex I deficiency OMIM
NDUFAF6	NM_152416.3	100%	Leigh syndrome OMIM
NDUFB11	NM_001135998.2	96%	Linear skin defects with multiple congenital anomalies 3 OMIM
NDUFB3	NM_002491.2	100%	Mitochondrial complex I deficiency OMIM
NDUFB9	NM_005005.2	100%	Mitochondrial complex I deficiency OMIM
NDUFS1	NM_005006.6	100%	Mitochondrial complex I deficiency OMIM
NDUFS2	NM_004550.4	100%	Mitochondrial complex I deficiency OMIM
NDUFS3	NM_004551.2	100%	Mitochondrial complex I deficiency OMIM Leigh syndrome due to mitochondrial complex I deficiency OMIM
NDUFS4	NM_002495.2	100%	Mitochondrial complex I deficiency OMIM Leigh syndrome OMIM
NDUFS6	NM_004553.4	100%	Mitochondrial complex I deficiency OMIM
NDUFS7	NM_024407.4	100%	Leigh syndrome OMIM
NDUFS8	NM_002496.3	100%	Leigh syndrome due to mitochondrial complex I deficiency OMIM
NDUFV1	NM_007103.3	100%	Mitochondrial complex I deficiency OMIM
NDUFV2	NM_021074.4	100%	Mitochondrial complex I deficiency OMIM
NFS1	NM_021100.4	99%	Mitochondrial complex II/III deficiency, infantile PubMed

Gen	Transkript	>10x	Fenotype
NFU1	NM_001002755.2	100%	Multiple mitochondrial dysfunctions syndrome 1 OMIM
NNT	NM_012343.3	99%	Glucocorticoid deficiency 4 OMIM
NUBPL	NM_025152.2	100%	Mitochondrial complex I deficiency OMIM
OAT	NM_000274.3	95%	Gyrate atrophy of choroid and retina with or without ornithinemia OMIM
OGDH	NM_002541.3	100%	Alpha-ketoglutarate dehydrogenase deficiency OMIM
OPA1	NM_015560.2	100%	Behr syndrome OMIM Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type) OMIM
OPA3	NM_025136.3	100%	3-methylglutaconic aciduria, type III OMIM
OTC	NM_000531.5	100%	Ornithine transcarbamylase deficiency OMIM
OXCT1	NM_000436.3	100%	Succinyl CoA:3-oxoacid CoA transferase deficiency OMIM
PANK2	NM_153638.2	100%	HARP syndrome OMIM Neurodegeneration with brain iron accumulation 1 OMIM
PARS2	NM_152268.3	100%	Combined oxidative phosphorylation deficiency, Alpers syndrome PubMed
PC	NM_000920.3	100%	Pyruvate carboxylase deficiency OMIM
PDHA1	NM_000284.3	99%	Pyruvate dehydrogenase E1-alpha deficiency OMIM
PDHB	NM_000925.3	100%	Pyruvate dehydrogenase E1-beta deficiency OMIM
PDHX	NM_003477.2	100%	Leigh-like syndrome, Pyruvate dehydrogenase complex deficiency OMIM
PDP1	NM_018444.3	100%	Pyruvate dehydrogenase phosphatase deficiency OMIM
PDPR	NM_017990.3	99%	Joubert-like syndrome PubMed
PDSS1	NM_014317.3	99%	Coenzyme Q10 deficiency, primary, 2 OMIM

Gen	Transkript	>10x	Fenotype
PDSS2	NM_020381.3	99%	Coenzyme Q10 deficiency, primary, 3 OMIM
PET100	NM_001171155.1	100%	Mitochondrial complex IV deficiency OMIM
PITRM1	NM_014889.3	99%	Progressive cognitive decline, spinocerebellar ataxia and psychosis PubMed
PMPCA	NM_015160.2	99%	Spinocerebellar ataxia, autosomal recessive 2 OMIM
PNPLA2	NM_020376.3	100%	Neutral lipid storage disease with myopathy OMIM
PNPLA4	NM_004650.2	100%	Combined oxidative phosphorylation deficiency PubMed
PNPLA6	NM_006702.4	99%	Boucher-Neuhauser syndrome OMIM Laurence-Moon syndrome OMIM Spastic paraplegia 39, autosomal recessive OMIM Oliver-McFarlane syndrome OMIM
PNPT1	NM_033109.4	100%	Combined oxidative phosphorylation deficiency 13 OMIM
POLG	NM_002693.2	100%	Progressive external ophthalmoplegia, autosomal recessive 1 OMIM Progressive external ophthalmoplegia, autosomal dominant 1 OMIM Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE) OMIM Mitochondrial DNA depletion syndrome 4B (MNGIE type) OMIM Mitochondrial DNA depletion syndrome 4A (Alpers type) OMIM
POLG2	NM_007215.3	99%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4 OMIM
PTCD1	NM_015545.3	100%	Mitochondrial complex I deficiency PubMed
PTRH2	NM_016077.3	100%	Multisystem neurologic, endocrine, and pancreatic disease (IMNEPD), infantile OMIM PubMed
PUS1	NM_025215.5	100%	Myopathy, lactic acidosis, and sideroblastic anemia 1 OMIM
QARS	NM_005051.2	100%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy OMIM
QRSL1	NM_018292.4	100%	Combined oxidative phosphorylation deficiency, infantile lethal PubMed
RARS2	NM_020320.3	100%	Pontocerebellar hypoplasia, type 6 OMIM
RMND1	NM_017909.3	100%	Combined oxidative phosphorylation deficiency 11 OMIM

Gen	Transkript	>10x	Fenotype
RNASEH1	NM_002936.4	99%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2 OMIM
RRM2B	NM_015713.4	100%	Mitochondrial DNA depletion syndrome 8B (MNGIE type) OMIM Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy) OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5 OMIM
SACS	NM_014363.5	100%	Spastic ataxia, Charlevoix-Saguenay type OMIM
SAMHD1	NM_015474.3	100%	Aicardi-Goutieres syndrome 5 OMIM
SARS2	NM_017827.3	100%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis OMIM
SCO1	NM_004589.3	100%	Mitochondrial complex IV deficiency OMIM
SCO2	NM_005138.2	100%	Cardioencephalomyopathy, fatal infantile OMIM
SDHA	NM_004168.3	86%	Mitochondrial complex II deficiency OMIM Leigh syndrome OMIM
SDHAF1	NM_001042631.2	100%	Mitochondrial complex II deficiency OMIM
SDHD	NM_003002.3	65%	Mitochondrial complex II deficiency OMIM
SERAC1	NM_032861.3	100%	MEGDEL syndrome OMIM
SFXN4	NM_213649.1	100%	Combined oxidative phosphorylation deficiency 18 OMIM
SLC19A2	NM_006996.2	100%	Thiamine-responsive megaloblastic anemia syndrome OMIM
SLC19A3	NM_025243.3	100%	Thiamine metabolism dysfunction syndrome 2 OMIM
SLC25A1	NM_005984.4	99%	Hydroxyglutaric aciduria OMIM Congenital myasthenia PubMed
SLC25A12	NM_003705.4	100%	Hypomyelination, global cerebral OMIM
SLC25A13	NM_014251.2	100%	Citrullinemia, type II, neonatal-onset OMIM Citrullinemia, adult-onset type II OMIM
SLC25A15	NM_014252.3	96%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome OMIM

Gen	Transkript	>10x	Fenotype
SLC25A19	NM_021734.4	99%	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type) OMIM
SLC25A20	NM_000387.5	100%	Carnitine-acylcarnitine translocase deficiency OMIM
SLC25A22	NM_024698.5	100%	Epileptic encephalopathy, early infantile, 3 OMIM
SLC25A26	NM_173471.3	100%	Combined oxidative phosphorylation deficiency 28 OMIM
SLC25A3	NM_005888.3	100%	Mitochondrial phosphate carrier deficiency OMIM
SLC25A38	NM_017875.2	100%	Anemia, sideroblastic, 2, pyridoxine-refractory OMIM
SLC25A4	NM_001151.3	100%	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type) OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions OMIM
SLC25A46	NM_138773.2	100%	Neuropathy, hereditary motor and sensory, type VIB OMIM Optic atrophy and cerebellar degeneration PubMed Charcot-Marie-Tooth disease, type 2, with optic atrophy PubMed
SUCLA2	NM_003850.2	96%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) OMIM
SUCLG1	NM_003849.3	100%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria) OMIM
SURF1	NM_003172.3	95%	Leigh syndrome OMIM Charcot-Marie-Tooth disease, type 4K OMIM
TACO1	NM_016360.3	100%	Mitochondrial complex IV deficiency OMIM
TARS2	NM_025150.4	100%	Combined oxidative phosphorylation deficiency 21 OMIM
TAZ	NM_000116.4	100%	Barth syndrome OMIM
TIMM8A	NM_004085.3	96%	Mohr-Tranebjaerg syndrome OMIM
TK2	NM_004614.4	99%	Mitochondrial DNA depletion syndrome 2 (myopathic type) OMIM
TMEM126A	NM_032273.3	100%	Optic atrophy 7, with auditory neuropathy OMIM
TMEM70	NM_017866.5	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2 OMIM

Gen	Transkript	>10x	Fenotype
TPK1	NM_022445.3	100%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type) OMIM
TRIT1	NM_017646.4	100%	Combined oxidative phosphorylation deficiency PubMed
TRMT5	NM_020810.3	100%	Combined oxidative phosphorylation deficiency 26 OMIM
TRMU	NM_018006.4	100%	Liver failure, transient infantile OMIM
TRNT1	NM_182916.2	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay OMIM Retinitis pigmentosa and erythrocytic microcytosis OMIM
TSFM	NM_001172696.1	100%	Combined oxidative phosphorylation deficiency 3 OMIM
TTC19	NM_017775.3	98%	Mitochondrial complex III deficiency, nuclear type 2 OMIM
TUFM	NM_003321.4	100%	Combined oxidative phosphorylation deficiency 4 OMIM
TXN2	NM_012473.3	100%	Combined oxidative phosphorylation deficiency 29 OMIM
TYMP	NM_001953.4	100%	Mitochondrial DNA depletion syndrome 1 (MNGIE type) OMIM
UQCC2	NM_032340.3	100%	Mitochondrial complex III deficiency, nuclear type 7 OMIM
UQCRB	NM_006294.4	100%	Mitochondrial complex III deficiency, nuclear type 3 OMIM
UQCRC2	NM_003366.2	100%	Mitochondrial complex III deficiency, nuclear type 5 OMIM
UQCRQ	NM_014402.4	100%	Mitochondrial complex III deficiency, nuclear type 4 OMIM
VARS2	NM_001167734.1	100%	Combined oxidative phosphorylation deficiency 20 OMIM
WDR45	NM_007075.3	99%	Neurodegeneration with brain iron acculation 5 OMIM
WFS1	NM_006005.3	100%	Wolfram-like syndrome, autosomal dominant OMIM Wolfram syndrome OMIM
WWOX	NM_016373.3	100%	Epileptic encephalopathy, early infantile, 28 OMIM Spinocerebellar ataxia, autosomal recessive 12 OMIM

Gen	Transkript	>10x	Fenotype
YARS2	NM_001040436.2	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2 OMIM