MITOCHONDRIAL DISORDERS GENE PANEL DG 2.17 (409 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	135.3	100.0%	100.0%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABAT	86.1	99.9%	98.4%	GABA-transaminase deficiency, 613163
ACAD9	130.9	100.0%	98.8%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACO2	125.5	95.6%	90.3%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ADAMTS10	135.4	100.0%	99.9%	Weill-Marchesani syndrome 1, recessive, 277600
ADCK2	163.7	100.0%	99.9%	No OMIM Disease ID
ADPRHL2	177.4	100.0%	100.0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	100.8	95.7%	85.1%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGK	109.6	99.6%	95.5%	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AIFM1	92.9	99.7%	96.5%	Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Combined oxidative phosphorylation deficiency 6, 300816 Deafness, X-linked 5, 300614
ALDH1B1	205.5	100.0%	100.0%	No OMIM Disease ID
ALKBH1	104.4	100.0%	99.5%	No OMIM Disease ID
ANO10	106.0	98.6%	96.5%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOPT1	80.4	82.1%	82.1%	Mitochondrial complex IV deficiency, 220110
APTX	99.2	94.5%	91.6%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARL2	142.8	100.0%	100.0%	No OMIM Disease ID
ARNT2	127.7	100.0%	100.0%	?Webb-Dattani syndrome, 615926
ATAD1	63.3	98.8%	91.0%	Hyperekplexia 4, 618011
ATAD3A	100.8	93.8%	88.8%	Harel-Yoon syndrome, 617183
ATAD3B	105.7	94.2%	85.9%	No OMIM Disease ID
ATP13A2	149.6	100.0%	99.8%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225

ATP5A1	73.8	93.7%	85.2%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5B	112.2	100.0%	99.2%	No OMIM Disease ID
ATP5C1	82.8	96.2%	89.6%	No OMIM Disease ID
ATP5D	118.5	99.2%	93.8%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5E	146.3	100.0%	100.0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	62.4	96.7%	82.4%	No OMIM Disease ID
ATP5G1	89.2	99.9%	97.6%	No OMIM Disease ID
ATP5G2	84.4	100.0%	97.2%	No OMIM Disease ID
ATP5G3	117.4	100.0%	99.9%	No OMIM Disease ID
ATP5H	77.7	93.9%	77.4%	No OMIM Disease ID
ATP5I	83.2	100.0%	100.0%	No OMIM Disease ID
ATP5J	60.6	99.4%	90.9%	No OMIM Disease ID
ATP5J2	110.2	100.0%	99.8%	No OMIM Disease ID
ATP5L	120.7	100.0%	100.0%	No OMIM Disease ID
ATP5L2	156.3	100.0%	100.0%	No OMIM Disease ID
ATP5O	92.5	99.4%	91.9%	No OMIM Disease ID
ATP5S	108.3	100.0%	100.0%	No OMIM Disease ID
ATPAF1	70.7	96.3%	88.3%	No OMIM Disease ID
ATPAF2	109.2	100.0%	100.0%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATPIF1	199.8	100.0%	100.0%	No OMIM Disease ID
BCS1L	160.0	100.0%	100.0%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BOLA1	131.2	100.0%	100.0%	No OMIM Disease ID
BOLA2	126.2	100.0%	100.0%	No OMIM Disease ID
BOLA3	50.9	99.8%	94.6%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
C12orf65	112.4	100.0%	99.8%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C19orf12	117.5	100.0%	99.9%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C19orf70	84.4	99.9%	99.2%	Combined oxidative phosphorylation deficiency 37, 618329
C1QBP	68.9	92.8%	81.2%	Combined oxidative phosphorylation deficiency 33, 617713
CA5A	99.0	99.9%	97.2%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CARS2	138.8	100.0%	100.0%	Combined oxidative phosphorylation deficiency 27, 616672
CEP89	130.0	98.2%	95.5%	No OMIM disease ID

CHCHD10	28.6	67.7%	41.5%	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHCHD2	75.2	100.0%	96.6%	Parkinson disease 22, autosomal dominant, 616710
CHKB	126.8	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CISD2	116.7	83.4%	83.4%	Wolfram syndrome 2, 604928
CLPB	135.3	99.7%	97.4%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	152.0	100.0%	99.6%	Perrault syndrome 3, 614129
COA1	85.9	100.0%	99.9%	No OMIM Disease ID
COA3	171.5	100.0%	100.0%	No OMIM Disease ID
COA5	81.0	87.4%	83.4%	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500
COA6	125.2	100.0%	97.4%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501
COA7	132.3	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COASY	190.6	100.0%	100.0%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COQ2	107.7	97.7%	97.0%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	116.2	91.7%	90.8%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	176.0	100.0%	100.0%	No OMIM Disease ID
COQ6	136.6	99.6%	97.4%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	142.8	99.9%	99.6%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	177.7	100.0%	100.0%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	109.0	100.0%	99.9%	Nephrotic syndrome, type 9, 615573
COQ9	78.6	99.9%	98.5%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	232.8	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110 Leigh syndrome due to mitochondrial COX4 deficiency, 256000
COX14	103.1	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
COX15	90.4	99.9%	98.7%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
COX20	66.5	93.9%	81.6%	Mitochondrial complex IV deficiency, 220110
COX4I1	114.6	100.0%	100.0%	No OMIM Disease ID
COX4I2	125.6	100.0%	99.9%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	30.2	81.6%	53.1%	No OMIM Disease ID
COX5B	150.3	100.0%	100.0%	No OMIM Disease ID
COX6A1	160.6	100.0%	100.0%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	64.2	99.6%	95.6%	Mitochondrial complex IV deficiency, 220110
COX6B1	143.0	100.0%	100.0%	Mitochondrial complex IV deficiency, 220110
COX6B2	106.9	100.0%	100.0%	No OMIM Disease ID

COX6C	126.5	99.9%	95.8%	No OMIM Disease ID
COX7A1	137.9	100.0%	100.0%	No OMIM Disease ID
COX7A2	95.3	100.0%	99.6%	No OMIM Disease ID
COX7B	39.4	62.3%	31.6%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	154.4	100.0%	100.0%	No OMIM Disease ID
COX7C	36.9	97.6%	85.0%	No OMIM Disease ID
COX8A	120.1	100.0%	100.0%	?Mitochondrial complex IV deficiency, 220110
COX8C	167.7	100.0%	100.0%	No OMIM Disease ID
CP	99.2	92.9%	87.3%	Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CRAT	127.2	100.0%	100.0%	?Neurodegeneration with brain iron accumulation 8, 617917
CTBP1	111.3	95.7%	88.6%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CYC1	167.9	99.7%	97.3%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	60.4	99.5%	93.7%	Thrombocytopenia 4, 612004
DARS2	125.4	100.0%	98.6%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF17	87.5	100.0%	99.2%	Woodhouse-Sakati syndrome, 241080
DDHD1	166.9	100.0%	99.1%	Spastic paraplegia 28, autosomal recessive, 609340
DES	138.0	100.0%	100.0%	Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DGUOK	127.0	100.0%	98.8%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	127.4	99.9%	99.0%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	104.9	100.0%	99.4%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	117.2	100.0%	99.9%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	83.5	94.5%	87.3%	Paragangliomas 7, 618475
DMAC1	62.2	100.0%	99.7%	No OMIM Disease ID
DMAC2	129.0	98.3%	98.3%	No OMIM Disease ID
DNA2	121.9	99.9%	97.8%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJA3	127.5	100.0%	99.3%	No OMIM Disease ID
DNAJC19	92.5	99.1%	90.4%	3-methylglutaconic aciduria, type V, 610198
DNAJC3	136.0	100.0%	99.9%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNM1L	120.8	99.9%	97.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708

EARS2	106.7	99.7%	98.4%	Combined oxidative phosphorylation deficiency 12, 614924
ECHS1	111.6	100.0%	100.0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	160.6	100.0%	100.0%	No OMIM disease ID
EHHADH	138.8	100.0%	99.9%	?Fanconi renotubular syndrome 3, 615605
ELAC2	117.1	100.0%	99.5%	Combined oxidative phosphorylation deficiency 17, 615440
ERAL1	168.0	100.0%	100.0%	Perrault syndrome 6, 617565
ETFDH	112.7	100.0%	99.7%	Glutaric acidemia IIC, 231680
ETHE1	105.9	99.9%	97.9%	Ethylmalonic encephalopathy, 602473
FA2H	101.5	99.3%	95.1%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	169.5	100.0%	100.0%	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FARSB	77.5	96.9%	93.0%	Rajab interstitial lung disease with brain calcifications, 613658
FASTKD2	116.5	99.9%	98.0%	?Mitochondrial complex IV deficiency, 220110
FBXL4	165.9	100.0%	100.0%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX2	164.5	100.0%	100.0%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	134.5	100.0%	99.4%	Auditory neuropathy and optic atrophy, 617717
FH	126.0	95.9%	89.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	129.1	99.9%	99.0%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FTL	164.3	100.0%	98.4%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
FXN	67.4	100.0%	98.3%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GARS	128.2	100.0%	99.6%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GATB	102.6	100.0%	99.9%	No OMIM Disease ID
GATC	153.3	100.0%	100.0%	No OMIM Disease ID
GATM	139.0	100.0%	100.0%	Cerebral creatine deficiency syndrome 3, 612718
GFER	103.0	100.0%	99.9%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	104.4	100.0%	98.9%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	117.8	99.0%	95.2%	Combined oxidative phosphorylation deficiency 39, 618397
GLRX5	149.3	99.8%	97.8%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	66.3	98.0%	88.9%	Hyperinsulinism-hyperammonemia syndrome, 606762
GMPR	127.8	100.0%	100.0%	No OMIM Disease ID
GOT2	85.1	96.6%	91.6%	No OMIM Disease ID

GPT2	130.4	100.0%	99.6%	Mental retardation, autosomal recessive 49, 616281
GTPBP2	138.7	99.9%	99.2%	Jaberi-Elahi syndrome, 617988
GTPBP3	189.1	100.0%	100.0%	Combined oxidative phosphorylation deficiency 23, 616198
HADHA	74.6	96.1%	89.6%	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016 Trifunctional protein deficiency, 609015
HADHB	76.9	96.0%	83.7%	Trifunctional protein deficiency, 609015
HARS2	141.0	100.0%	99.9%	?Perrault syndrome 2, 614926
HCCS	90.4	99.6%	96.6%	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	69.7	95.5%	75.9%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	148.0	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HSD17B10	98.0	100.0%	99.5%	HSD10 mitochondrial disease, 300438
HSPA9	83.8	88.2%	84.2%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	73.7	97.9%	92.1%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HTRA2	145.3	100.0%	99.7%	3-methylglutaconic aciduria, type VIII, 617248
IARS2	145.7	100.0%	100.0%	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	162.1	99.8%	98.5%	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ISCA1	71.9	94.0%	82.9%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	112.2	99.8%	97.4%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	121.3	100.0%	99.9%	Myopathy with lactic acidosis, hereditary, 255125
KARS	109.9	100.0%	99.3%	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KIF1A	125.3	99.8%	98.2%	Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255 Spastic paraplegia 30, autosomal recessive, 610357
LACTB	112.5	99.7%	97.9%	No OMIM Disease ID
LARS2	128.3	100.0%	100.0%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LIAS	124.4	100.0%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT1	199.3	100.0%	99.8%	Lipoyltransferase 1 deficiency, 616299
LIPT2	107.4	100.0%	99.9%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LONP1	164.8	100.0%	100.0%	CODAS syndrome, 600373

LRPPRC	126.3	100.0%	99.7%	Leigh syndrome, French-Canadian type, 220111
LYRM4	78.4	68.0%	63.4%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	58.4	98.2%	87.5%	Mitochondrial complex III deficiency, nuclear type 8, 615838
MARS2	195.2	100.0%	100.0%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MCUR1	70.8	99.9%	97.8%	No OMIM Disease ID
MDH1	103.8	99.9%	99.0%	No OMIM Disease ID
MDH2	116.5	98.0%	98.0%	Epileptic encephalopathy, early infantile, 51, 617339
MECR	114.2	100.0%	99.7%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	86.8	93.5%	88.5%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	130.9	100.0%	99.9%	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MGME1	145.7	100.0%	99.9%	Mitochondrial DNA depletion syndrome 11, 615084
MICU1	103.4	98.7%	96.2%	Myopathy with extrapyramidal signs, 615673
MICU2	55.8	99.2%	94.9%	No OMIM Disease ID
MIEF2	155.0	100.0%	100.0%	No OMIM Disease ID
MIPEP	100.6	99.8%	96.1%	Combined oxidative phosphorylation deficiency 31, 617228
MPC1	161.4	100.0%	99.6%	Mitochondrial pyruvate carrier deficiency, 614741
MPV17	93.2	100.0%	98.5%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRM2	120.4	100.0%	99.4%	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	129.4	100.0%	99.8%	No OMIM Disease ID
MRPL3	66.2	93.4%	81.6%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	92.0	99.8%	95.0%	No OMIM Disease ID
MRPL44	130.4	100.0%	99.9%	?Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	253.5	100.0%	100.0%	No OMIM Disease ID
MRPS14	172.5	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	136.8	100.0%	99.1%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	175.4	100.0%	100.0%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	138.5	100.0%	97.3%	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS23	142.5	100.0%	99.8%	No OMIM Disease ID
MRPS25	142.6	100.0%	99.7%	No OMIM Disease ID
MRPS28	145.5	87.6%	86.8%	No OMIM Disease ID
MRPS34	184.8	100.0%	100.0%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS36	66.8	91.7%	67.3%	No OMIM Disease ID

MRPS7	161.3	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	142.2	100.0%	100.0%	No OMIM Disease ID
MSTO1	107.9	99.5%	97.1%	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	131.5	100.0%	99.7%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTO1	149.2	91.9%	89.5%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	124.6	99.3%	93.9%	?Spastic ataxia 4, autosomal recessive, 613672
NARS2	121.5	97.6%	97.4%	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NAXD	144.4	100.0%	100.0%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	90.7	99.8%	97.2%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFA1	195.4	99.9%	98.7%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	118.3	99.9%	99.2%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	129.7	100.0%	98.5%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	162.1	100.0%	99.9%	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	134.6	92.3%	92.0%	?Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	179.9	100.0%	100.0%	?Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	142.4	94.0%	88.0%	No OMIM Disease ID
NDUFA4	67.4	99.7%	84.3%	No OMIM Disease ID
NDUFA5	64.6	92.8%	68.0%	No OMIM Disease ID
NDUFA6	215.7	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	118.0	100.0%	100.0%	No OMIM Disease ID
NDUFA8	141.6	100.0%	99.8%	No OMIM Disease ID
NDUFA9	105.5	99.7%	96.1%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	139.7	99.6%	94.2%	No OMIM Disease ID
NDUFAF1	100.2	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	56.3	94.0%	79.5%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	159.3	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	98.6	99.4%	93.8%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	128.0	99.8%	99.3%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	92.3	100.0%	98.9%	Mitochondrial complex I deficiency, nuclear type 17, 618239
NDUFAF7	99.8	100.0%	100.0%	No OMIM Disease ID
NDUFAF8	53.8	89.0%	67.0%	No OMIM Disease ID
NDUFB1	43.9	59.7%	51.7%	No OMIM Disease ID
NDUFB10	162.2	99.6%	96.6%	No OMIM Disease ID
NDUFB11	110.0	99.2%	96.2%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021

NDUFB2	94.3	100.0%	100.0%	No OMIM Disease ID
NDUFB3	22.8	89.3%	61.0%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	125.2	86.4%	84.2%	No OMIM Disease ID
NDUFB5	104.0	100.0%	99.9%	No OMIM Disease ID
NDUFB6	46.1	97.9%	83.9%	No OMIM Disease ID
NDUFB7	91.6	100.0%	99.5%	No OMIM Disease ID
NDUFB8	112.6	100.0%	99.8%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	114.2	98.3%	94.1%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	106.7	100.0%	100.0%	No OMIM Disease ID
NDUFC2	55.0	99.4%	92.8%	No OMIM Disease ID
NDUFS1	140.7	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	102.9	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	132.3	90.7%	90.6%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	148.6	100.0%	99.6%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	133.4	100.0%	100.0%	No OMIM Disease ID
NDUFS6	120.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	157.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	171.3	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	154.4	100.0%	99.5%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	71.5	91.8%	77.9%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	140.9	100.0%	99.9%	No OMIM Disease ID
NFS1	70.8	89.4%	88.4%	No OMIM Disease ID
NFU1	61.6	96.2%	77.8%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NME3	171.4	100.0%	98.0%	No OMIM Disease ID
NR2F1	261.9	100.0%	100.0%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NSUN3	168.1	100.0%	100.0%	No OMIM Disease ID
NUBPL	102.4	98.9%	94.2%	Mitochondrial complex I deficiency, nuclear type 21, 618242
OGDH	181.7	100.0%	99.6%	No OMIM disease ID
OPA1	121.4	99.7%	97.5%	Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	171.9	100.0%	99.9%	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OTX2	135.4	100.0%	99.6%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125

OXA1L	144.1	100.0%	99.9%	No OMIM Disease ID
PANK2	161.5	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PARS2	193.2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 75, 618437
PC	170.2	99.9%	98.8%	Pyruvate carboxylase deficiency, 266150
PDHA1	88.0	98.6%	95.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	113.7	99.6%	97.7%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	132.4	100.0%	99.5%	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	134.3	100.0%	99.8%	No OMIM Disease ID
PDK2	167.4	100.0%	100.0%	No OMIM Disease ID
PDK3	109.5	97.4%	94.3%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	111.2	100.0%	99.3%	No OMIM Disease ID
PDP1	134.9	100.0%	100.0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	106.9	97.6%	88.2%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	115.4	99.3%	95.2%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	95.2	99.7%	90.6%	Mitochondrial complex IV deficiency, 220110
PET117	118.8	100.0%	100.0%	No OMIM Disease ID
PIGA	72.9	93.0%	83.4%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PISD	172.5	100.0%	99.9%	No OMIM Disease ID
PITRM1	106.3	97.9%	95.1%	No OMIM Disease ID
PLA2G6	121.0	99.9%	98.6%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLPBP	97.2	99.9%	97.2%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMPCA	113.3	99.6%	97.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	121.6	100.0%	99.3%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNPLA8	118.2	100.0%	100.0%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPT1	54.5	96.4%	83.0%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLG	124.4	100.0%	99.8%	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	190.7	99.8%	98.1%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528

POLR2A	176.1	100.0%	100.0%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
PPA2	92.7	98.0%	90.2%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	166.9	100.0%	99.2%	Cardiomyopathy, dilated, 2C, 618189
PRKAA1	137.0	99.9%	99.3%	No OMIM Disease ID
PTCD3	81.7	98.7%	96.8%	No OMIM Disease ID
PTRH2	209.0	100.0%	100.0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	125.7	99.8%	98.1%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	105.0	100.0%	99.0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	129.0	99.7%	97.6%	Leukodystrophy, hypomyelinating, 10, 616420
PYROXD1	46.8	90.5%	76.6%	Myopathy, myofibrillar, 8, 617258
QRSL1	86.0	98.8%	93.2%	No OMIM Disease ID
RARS2	102.7	100.0%	99.3%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	130.7	100.0%	99.0%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	105.2	98.0%	92.6%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RRM1	120.4	100.0%	99.7%	No OMIM Disease ID
RRM2B	142.6	100.0%	99.4%	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	80.2	99.6%	98.2%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RYR1	128.2	99.3%	96.8%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
SACS	151.1	100.0%	100.0%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	135.4	100.0%	98.7%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS2	129.0	95.4%	93.8%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	105.2	100.0%	99.6%	Mitochondrial complex IV deficiency, 220110
SCO2	134.9	100.0%	100.0%	Myopia 6, 608908 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377
SCP2	107.1	99.8%	96.8%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDHA	94.1	85.1%	78.0%	Leigh syndrome, 256000 Paragangliomas 5, 614165 Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial respiratory chain complex II deficiency, 252011

SDHAF1	104.7	100.0%	100.0%	Mitochondrial complex II deficiency, 252011
SDHB	116.1	100.0%	100.0%	Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paragangliomas 4, 115310 Paraganglioma and gastric stromal sarcoma, 606864
SDHD	45.8	53.1%	50.6%	Paragangliomas 1, with or without deafness, 168000 Mitochondrial complex II deficiency, 252011 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SERAC1	110.4	100.0%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	126.1	100.0%	99.7%	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	103.2	100.0%	99.7%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	139.2	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	114.2	99.8%	97.0%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A10	91.1	82.6%	77.3%	No OMIM Disease ID
SLC25A12	151.7	100.0%	99.7%	Epileptic encephalopathy, early infantile, 39, 612949
SLC25A13	118.2	99.9%	98.1%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A19	82.0	100.0%	98.5%	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 Microcephaly, Amish type, 607196
SLC25A21	121.5	100.0%	99.9%	No OMIM Disease ID
SLC25A22	138.5	100.0%	99.7%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A24	133.1	99.7%	99.5%	Fontaine progeroid syndrome, 612289
SLC25A3	140.1	99.8%	97.0%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	128.5	100.0%	100.0%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	98.5	99.0%	95.5%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	141.9	100.0%	99.9%	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	143.7	100.0%	99.2%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	175.2	99.8%	97.2%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC39A8	144.7	100.0%	99.8%	Congenital disorder of glycosylation, type IIn, 616721
SLC52A2	213.2	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	128.9	100.0%	99.9%	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SOD2	208.3	100.0%	100.0%	No OMIM disease ID

SPART	132.9	100.0%	98.4%	Troyer syndrome, 275900
SPATA5	142.5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG7	123.7	99.8%	97.8%	Spastic paraplegia 7, autosomal recessive, 607259
SQOR	103.2	99.9%	97.9%	No OMIM Disease ID
SQSTM1	129.7	100.0%	99.6%	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	59.4	99.6%	94.1%	No OMIM Disease ID
STAC3	122.8	100.0%	100.0%	Myopathy, congenital, Baily-Bloch, 255995
STAT2	117.1	100.0%	99.9%	Immunodeficiency 44, 616636
STXBP1	108.2	96.8%	96.5%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	57.8	91.5%	82.6%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	104.2	100.0%	99.7%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	57.6	92.1%	79.9%	No OMIM Disease ID
SURF1	89.9	93.5%	89.1%	Leigh syndrome, due to COX IV deficiency, 256000 Charcot-Marie-Tooth disease, type 4K, 616684
SZT2	146.3	99.6%	99.5%	Epileptic encephalopathy, early infantile, 18, 615476
TACO1	103.0	99.9%	95.8%	Mitochondrial complex IV deficiency, 220110
TANGO2	139.6	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	132.6	99.0%	97.3%	No OMIM Disease ID
TARS2	93.9	99.7%	97.1%	?Combined oxidative phosphorylation deficiency 21, 615918
TAZ	125.3	99.3%	96.2%	Barth syndrome, 302060
TDP2	175.8	100.0%	99.9%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TFB2M	76.4	99.9%	96.9%	No OMIM Disease ID
THG1L	136.9	100.0%	100.0%	No OMIM Disease ID
TIMM22	113.4	100.0%	99.7%	No OMIM Disease ID
TIMM44	173.2	100.0%	100.0%	No OMIM Disease ID
TIMM50	133.8	100.0%	99.4%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	50.3	95.4%	80.0%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	167.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TK2	111.5	100.0%	99.8%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TMEM126A	100.7	95.6%	79.5%	Optic atrophy 7, 612989
TMEM126B	87.8	99.6%	96.7%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM186	130.3	100.0%	100.0%	No OMIM Disease ID

TMEM65	74.6	95.2%	86.2%	No OMIM Disease ID
TMEM70	117.3	99.9%	98.5%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMX2	121.3	100.0%	98.1%	No OMIM Disease ID
TOP3A	128.8	99.7%	97.7%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TPK1	96.3	99.7%	97.1%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRAPPC2L	211.9	100.0%	100.0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRIT1	107.1	100.0%	99.9%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT10C	138.5	100.0%	100.0%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	181.7	99.9%	98.2%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	106.5	100.0%	99.5%	Liver failure, transient infantile, 613070
TRNT1	100.7	99.2%	95.3%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TSFM	123.3	100.0%	99.6%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	84.9	98.8%	86.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	141.1	100.0%	99.6%	Combined oxidative phosphorylation deficiency 4, 610678
TWNK	170.3	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	65.7	100.0%	99.8%	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	138.6	100.0%	100.0%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC1	95.6	100.0%	100.0%	No OMIM Disease ID
UQCC2	146.3	99.8%	98.8%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	140.4	100.0%	100.0%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	197.2	100.0%	100.0%	No OMIM Disease ID
UQCR11	220.8	100.0%	100.0%	No OMIM Disease ID
UQCRB	106.1	98.8%	94.8%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	138.0	99.8%	98.4%	No OMIM Disease ID
UQCRC2	105.8	99.9%	98.3%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	124.7	97.7%	92.1%	No OMIM Disease ID
UQCRH	117.7	99.2%	92.3%	No OMIM Disease ID
UQCRQ	172.4	100.0%	100.0%	Mitochondrial complex III deficiency, nuclear type 4, 615159
USMG5	19.0	85.6%	39.2%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
VARS2	130.9	100.0%	99.8%	Combined oxidative phosphorylation deficiency 20, 615917
VPS13D	141.7	100.0%	99.8%	Spinocerebellar ataxia, autosomal recessive 4, 607317
WARS2	142.1	99.9%	99.1%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710

WDR45	74.7	97.1%	90.6%	Neurodegeneration with brain iron accumulation 5, 300894
YARS2	188.2	100.0%	99.4%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	102.3	98.1%	92.4%	?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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors