MITOCHONDRIAL GENE PANEL DG 2.3.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
AARS2	86,4	99%	96%	Combined oxidative phosphorylation deficiency 8, 614096
ABCB7	60,4	100%	94%	Anemia, sideroblastic, with ataxia, 301310
ACAD8	87,2	97%	92%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	91	100%	99%	ACAD9 deficiency, 611126
ACADSB	85,7	98%	96%	2-methylbutyrylglycinuria, 610006
ACAT1	108,9	100%	98%	Alpha-methylacetoacetic aciduria, 203750
ACO2	78,6	90%	84%	Infantile cerebellar-retinal degeneration, 614559
ACSF2	88,5	100%	98%	No OMIM phenotype
ACSF3	77	100%	99%	Combined malonic and methylmalonic aciduria, 614265
ADAR	133,6	99%	98%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADCK3	97,8	100%	95%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCK4	64	100%	91%	Nephrotic syndrome type 9, 615573
AFG3L2	76,4	95%	91%	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487
AGK	107,5	99%	99%	Hyperoxaluria, primary, type 1, 259900
AIFM1	58,2	99%	87%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
AKR1B15	81,9	65%	65%	No OMIM phenotype
ALDH1B1	131,1	100%	100%	No OMIM phenotype
ANO10	99,3	99%	97%	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOPT1	88,4	87%	87%	Mitochondrial complex IV deficiency,220110
APTX	120,7	96%	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ATAD3A	38,8	42%	35%	No OMIM phenotype
ATAD3B	44	47%	45%	No OMIM phenotype
ATP5A1	56,4	93%	80%	?Combined oxidative phosphorylation deficiency 22,616045 ?Mitochondrial complex (ATP synthase) deficiency,nuclear type 4,615228
ATP5B	101,3	100%	100%	No OMIM phenotype

ATP5C1	65,3	94%	88%	No OMIM phenotype
ATP5D	57,5	81%	59%	No OMIM phenotype
ATP5E	159,8	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5F1	79,5	66%	66%	No OMIM phenotype
ATP5G1	16,8	71%	33%	No OMIM phenotype
ATP5G2	61,8	96%	82%	No OMIM phenotype
ATP5G3	83,1	100%	99%	No OMIM phenotype
ATP5H	89,7	99%	82%	No OMIM phenotype
ATP5I	72,4	100%	100%	No OMIM phenotype
ATP5J	50,5	93%	72%	No OMIM phenotype
ATP5J2	41	99%	78%	No OMIM phenotype
ATP5L	52,8	70%	69%	No OMIM phenotype
ATP5L2	52,4	100%	99%	No OMIM phenotype
ATP5O	75,2	100%	100%	No OMIM phenotype
ATP5S	83,2	100%	97%	No OMIM phenotype
ATPAF1	80,8	96%	71%	No OMIM phenotype
ATPAF2	70,9	100%	98%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATPIF1	185,4	100%	100%	No OMIM phenotype
BCKDHA	104,7	100%	98%	Maple syrup urine disease, type Ia, 248600
BCKDHB	86,1	98%	83%	Maple syrup urine disease, type Ib, 248600
BCS1L	140,9	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358
BOLA1	93	100%	100%	No OMIM phenotype
BOLA2	0	0%	0%	No OMIM phenotype
BOLA3	57,8	100%	99%	Multiple mitochondrial dysfunctions syndrome 2, 614299
C10orf2	138,3	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type),271245 Perrault syndrome 5,616138 Progressive external ophthalmoplegia with mitochondrial DNA depletions, autosomal dominant,609286
C12orf65	174,3	100%	100%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55,autosomal recessive, 615035
C14orf2	89,5	100%	99%	No OMIM phenotype

C19orf12	72,8	100%	95%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
CAMKMT	65,2	99%	94%	No OMIM phenotype
CARS2	72,6	100%	97%	No OMIM phenotype
CEP89	86,6	100%	97%	No OMIM phenotype
CHCHD10	22,5	66%	28%	?Myopathy,isolated mitochondrial,autosomal dominant,616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2,615911 Spinal muscular atrophy,Jokela type,615048
CHCHD4	44,6	100%	86%	No OMIM phenotype
CHKB	91,1	93%	91%	Muscular dystrophy, congenital, megaconial type, 602541
CLPB	101,9	96%	95%	3-methylglutaconic aciduria,type VII,with cataracts,neurologic involvement and neutropenia,616271
CLPP	90,1	97%	91%	Perrault syndrome 3, 614129
COA1	142	100%	100%	No OMIM phenotype
COA3	66,7	100%	95%	No OMIM phenotype
COA5	72,3	85%	82%	Mitochondrial complex IV deficiency, 220110
COA6	72,5	100%	100%	Cardioencephalomyopathy,fatal infantile,due to cytochrome c oxidase deficiency 4,616501
COASY	126,1	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COQ10B	133	100%	100%	No OMIM phenotype
COQ2	73,6	96%	93%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ3	114	100%	100%	No OMIM phenotype
COQ4	75	86%	77%	Coenzyme Q10 deficiency,primary,7,616276
COQ5	136,9	100%	100%	No OMIM phenotype
COQ6	109,6	99%	95%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	96,5	100%	100%	No OMIM phenotype
COQ9	86,6	91%	83%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	130,6	100%	97%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX11	135,5	100%	100%	No OMIM phenotype
COX14	129,7	100%	100%	Mitochondrial complex IV deficiency, 220110
COX15	75	100%	93%	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119
COX17	79,3	100%	100%	No OMIM phenotype
COX18	96	100%	100%	No OMIM phenotype
COX19	81,7	100%	99%	No OMIM phenotype

COX20	67	89%	89%	Mitochondrial complex IV deficiency, 220110
COX4I1	48,2	91%	84%	No OMIM phenotype
COX4I2	56,1	99%	89%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis,612714
COX5A	55,7	64%	51%	No OMIM phenotype
COX5B	111,5	100%	100%	No OMIM phenotype
COX6A1	99,9	74%	74%	Charcot-Marie-Tooth disease,recessive intermediate D,616039
COX6A2	42,1	77%	71%	No OMIM phenotype
COX6B1	74,3	100%	100%	?Mitochondrial complex IV deficiency,220110
COX6B2	81,9	87%	74%	No OMIM phenotype
COX6C	171,1	100%	100%	No OMIM phenotype
COX7A1	93	100%	94%	No OMIM phenotype
COX7A2	57	86%	81%	No OMIM phenotype
COX7A2L	103	100%	100%	No OMIM phenotype
COX7B	32,8	76%	67%	Linear skin defects with multiple congenital anomalies,300887
COX7B2	143,6	100%	100%	No OMIM phenotype
COX7C	29,4	99%	67%	No OMIM phenotype
COX8A	122	100%	100%	No OMIM phenotype
COX8C	28,2	42%	42%	No OMIM phenotype
CYC1	81,4	95%	79%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	49,5	100%	92%	Thrombocytopenia 4, 612004
DAP3	95,7	100%	100%	No OMIM phenotype
DARS2	116,2	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	111,8	100%	100%	Maple syrup urine disease, type II, 248600
DGUOK	96,6	100%	100%	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	103,3	100%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DLAT	107	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	143,7	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	79,6	100%	100%	No OMIM phenotype
DNA2	117,9	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6, 615156
DNAJC19	57,8	79%	78%	3-methylglutaconic aciduria, type V, 610198
DNM1L	104,1	100%	100%	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
EARS2	70	94%	91%	Combined oxidative phosphorylation deficiency 12, 614924

ECHS1	64	98%	92%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency,616277
ECSIT	97,5	100%	96%	No OMIM phenotype
ELAC2	87	100%	100%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 Combined oxidative phosphorylation deficiency 17, 615440
ETFA	117,6	100%	100%	Glutaric acidemia IIA, 231680
ETFB	107,8	100%	99%	Glutaric acidemia 2B, 231680
ETFDH	132,6	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	57	99%	88%	Ethylmalonic encephalopathy, 602473
FARS2	97,6	98%	94%	Combined oxidative phosphorylation deficiency 14, 614946
FASTKD2	132,3	100%	100%	?Mitochondrial complex IV deficiency,220110
FBXL4	143,6	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX1L	89,9	100%	93%	No OMIM phenotype
FH	85,7	98%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FOXRED1	93,4	100%	97%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency,252010
FXN	87	93%	87%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GARS	108	98%	95%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy,distal hereditary motor,type VA,600794
GATM	89,2	100%	94%	Cerebral creatine deficiency syndrome 3, 612718
GCDH	76,3	93%	89%	Glutaricaciduria, type I, 231670
GFER	65,6	99%	94%	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076
GFM1	121,1	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	109,7	100%	100%	No OMIM phenotype
GLRX5	29,9	72%	46%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
GLUD1	111,8	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GRPEL1	89,7	100%	99%	No OMIM phenotype
HADH	85	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975

HADHA	93,2	96%	88%	LCHAD deficiency, 609016 Trifunctional protein deficiency, 609015 HELLP syndrome, maternal, of pregnancy, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	93,9	100%	99%	Trifunctional protein deficiency, 609015
HARS2	136,3	100%	100%	Perrault syndrome 2, 614926
HCCS	63,9	100%	96%	Microphthalmia, syndromic 7, 309801
HIBCH	68,2	100%	99%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	141,8	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMGCL	102,5	100%	99%	HMG-CoA lyase deficiency, 246450
HMGCS2	117,6	100%	99%	HMG-CoA synthase-2 deficiency, 605911
HOGA1	64	100%	93%	Hyperoxaluria, primary, type III, 613616
HSD17B10	54,1	95%	91%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSPA9	76,3	95%	87%	No OMIM phenotype
HSPD1	15,4	62%	38%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HSPE1	12,2	57%	11%	No OMIM phenotype
IARS2	125,7	100%	100%	?Cataracts,growth hormone deficiency,sensory neuropathy,sensorineural hearing loss and skeletal dysplasia,616007
IBA57	90	100%	94%	?Multiple mitochondrial dysfunctions syndrome 3,615330 ?Spastic paraplegia 74,autosomal recessive,616451
ISCA1	42,1	69%	68%	No OMIM phenotype
ISCA2	84,6	100%	96%	Multiple mitochondrial dysfunctions syndrome 4,616370
ISCU	81	100%	95%	Myopathy with lactic acidosis, hereditary, 255125
KARS	116,2	100%	100%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness,autosomal recessive 89,613916
LACTB	104,5	100%	96%	No OMIM phenotype
LARS2	116,2	100%	99%	Perrault syndrome 4, 615300
LIAS	101,7	100%	100%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
LIPT1	200	100%	100%	Lipoyltransferase 1 deficiency,616299
LRPPRC	100,6	98%	96%	Leigh syndrome, French-Canadian type, 220111
LYRM4	63,9	66%	66%	?Combined oxidative phosphorylation deficiency 19,615595

LYRM7	75,9	100%	99%	?Mitochondrial complex III deficiency,nuclear type 8,615838
MARS2	156,2	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MCCC2	113,6	98%	92%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MFF	82,1	100%	96%	Samheldin J Med Genet
MFN1	97,1	98%	91%	No OMIM phenotype
MFN2	103,5	100%	97%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA,601152
MGME1	158,4	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MPC1	89,4	100%	100%	Mitochondrial pyruvate carrier deficiency,614741
MPV17	102,7	100%	100%	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 -3
MRPL1	172,3	98%	96%	No OMIM phenotype
MRPL10	57,4	90%	89%	No OMIM phenotype
MRPL11	91,1	85%	81%	No OMIM phenotype
MRPL12	80,8	95%	85%	No OMIM phenotype
MRPL13	115,2	100%	100%	No OMIM phenotype
MRPL14	137,4	100%	100%	No OMIM phenotype
MRPL15	99,9	100%	100%	No OMIM phenotype
MRPL16	127,4	100%	100%	No OMIM phenotype
MRPL17	109,5	100%	97%	No OMIM phenotype
MRPL18	144,8	100%	100%	No OMIM phenotype
MRPL19	131,3	94%	94%	No OMIM phenotype
MRPL2	165,8	100%	100%	No OMIM phenotype
MRPL20	123,1	100%	95%	No OMIM phenotype
MRPL21	73,6	93%	87%	No OMIM phenotype
MRPL22	120,7	100%	100%	No OMIM phenotype
MRPL23	80,1	100%	98%	No OMIM phenotype
MRPL24	110,5	100%	100%	No OMIM phenotype
MRPL27	160,9	100%	100%	No OMIM phenotype
MRPL28	95,1	98%	95%	No OMIM phenotype
MRPL3	81,6	97%	92%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL30	123,8	100%	100%	No OMIM phenotype
MRPL32	134	100%	100%	No OMIM phenotype
MRPL33	93,5	100%	100%	No OMIM phenotype

MRPL34	81,3	100%	100%	No OMIM phenotype
MRPL35	49,9	99%	83%	No OMIM phenotype
MRPL36	16,3	69%	47%	No OMIM phenotype
MRPL37	162,6	100%	100%	No OMIM phenotype
MRPL38	78,6	100%	97%	No OMIM phenotype
MRPL39	107,3	100%	100%	No OMIM phenotype
MRPL4	91,5	75%	75%	No OMIM phenotype
MRPL40	99,1	100%	100%	No OMIM phenotype paper in press (shoubridge)
MRPL41	170,9	100%	100%	No OMIM phenotype
MRPL42	65,6	84%	84%	No OMIM phenotype
MRPL43	123,8	100%	98%	No OMIM phenotype
MRPL44	116,8	100%	100%	?Combined oxidative phosphorylation deficiency 16,615395
MRPL45	121,6	80%	80%	No OMIM phenotype
MRPL46	103,4	100%	100%	No OMIM phenotype
MRPL47	128	100%	100%	No OMIM phenotype
MRPL48	68,7	91%	91%	No OMIM phenotype
MRPL49	86,2	100%	100%	No OMIM phenotype
MRPL50	104	100%	100%	No OMIM phenotype
MRPL51	104,6	100%	99%	No OMIM phenotype
MRPL52	78,5	92%	91%	No OMIM phenotype
MRPL53	124,5	100%	100%	No OMIM phenotype
MRPL54	63	100%	100%	No OMIM phenotype
MRPL55	63,9	100%	100%	No OMIM phenotype
MRPL9	89,1	99%	97%	No OMIM phenotype
MRPS10	66,4	97%	92%	No OMIM phenotype
MRPS11	103,4	99%	87%	No OMIM phenotype
MRPS12	180,4	100%	100%	No OMIM phenotype
MRPS14	87,5	99%	81%	No OMIM phenotype
MRPS15	89	100%	100%	No OMIM phenotype
MRPS16	139,8	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS17	106,1	100%	100%	No OMIM phenotype
MRPS18A	115,3	100%	99%	No OMIM phenotype

MRPS18B	19,1	80%	41%	No OMIM phenotype
MRPS18C	61,2	94%	84%	No OMIM phenotype
MRPS2	114,5	95%	90%	No OMIM phenotype
MRPS21	34,1	88%	61%	No OMIM phenotype
MRPS22	92,5	100%	100%	Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	112	100%	98%	No OMIM phenotype
MRPS24	128,7	100%	96%	No OMIM phenotype
MRPS25	122,7	100%	100%	No OMIM phenotype
MRPS26	37,9	79%	72%	No OMIM phenotype
MRPS27	122,6	100%	100%	No OMIM phenotype
MRPS28	112	86%	86%	No OMIM phenotype
MRPS30	127,9	100%	100%	No OMIM phenotype
MRPS31	104,7	95%	91%	No OMIM phenotype
MRPS33	91,3	100%	100%	No OMIM phenotype
MRPS34	99,5	100%	100%	No OMIM phenotype
MRPS35	109,6	100%	100%	No OMIM phenotype
MRPS36	61,8	85%	75%	No OMIM phenotype
MRPS5	91,8	100%	100%	No OMIM phenotype
MRPS6	97,8	100%	93%	No OMIM phenotype
MRPS7	113,7	100%	100%	No OMIM phenotype
MRPS9	95,8	100%	98%	No OMIM phenotype
MRRF	158,2	100%	100%	No OMIM phenotype
MTCH1	90,4	100%	97%	No OMIM phenotype
MTCH2	81,7	99%	94%	No OMIM phenotype
MTERF	168,6	100%	100%	No OMIM phenotype
MTFMT	100,3	100%	100%	Combined oxidative phosphorylation deficiency 15, 614947
MTG1	84,1	100%	92%	No OMIM phenotype
MTHFD1L	76,1	88%	80%	No OMIM phenotype
MTO1	125,3	100%	98%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	105,9	93%	93%	Ataxia, spastic, 4, 613672
NARS2	117,9	100%	96%	Combined oxidative phosphorylation deficiency 24,616239
NDUFA1	116,6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA10	81,5	100%	96%	previous assignment to chr. 12 Leigh syndrome, 256000

NDUFA11	96,6	93%	79%	Mitochondrial complex I deficiency, 252010
NDUFA12	92,8	100%	100%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFA13	74,4	95%	91%	{Thyroid carcinoma,Hurthle cell},607464
NDUFA2	160,5	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFA3	89,3	98%	92%	No OMIM phenotype
NDUFA4	51,5	75%	58%	No OMIM phenotype
NDUFA5	44,4	55%	50%	No OMIM phenotype
NDUFA6	203,3	100%	100%	No OMIM phenotype
NDUFA7	79,2	99%	87%	No OMIM phenotype
NDUFA8	85,8	98%	98%	No OMIM phenotype
NDUFA9	84,9	97%	94%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 -3
NDUFAB1	68	100%	100%	No OMIM phenotype
NDUFAF1	116,9	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	53,9	100%	98%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	131,2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	81,2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF5	129,3	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF6	97,9	100%	97%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFAF7	86,3	100%	98%	No OMIM phenotype
NDUFB1	66,8	99%	93%	No OMIM phenotype
NDUFB10	115,4	100%	95%	No OMIM phenotype
NDUFB11	44,9	98%	87%	Linear skin defects with multiple congenital anomalies,300952
NDUFB2	86,7	100%	93%	No OMIM phenotype
NDUFB3	2,3	0%	0%	Mitochondrial complex I deficiency, 252010
NDUFB4	43,7	89%	71%	No OMIM phenotype
NDUFB5	90	100%	100%	No OMIM phenotype
NDUFB6	111,6	100%	100%	No OMIM phenotype
NDUFB7	51,4	99%	69%	No OMIM phenotype
NDUFB8	89,4	100%	100%	No OMIM phenotype
NDUFB9	116,4	100%	100%	?Mitochondrial complex I deficiency,252010
NDUFC1	78,7	100%	100%	No OMIM phenotype
NDUFC2	73,3	97%	82%	No OMIM phenotype

NDUFS1	80,4	100%	98%	Mitochondrial complex I deficiency, 252010
NDUFS2	123	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS3	143,5	96%	94%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	127,6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS5	161,4	100%	100%	No OMIM phenotype
NDUFS6	127,2	93%	85%	Mitochondrial complex I deficiency, 252010
NDUFS7	95,7	100%	100%	Leigh syndrome, 256000
NDUFS8	107,7	100%	96%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	63,4	100%	92%	Mitochondrial complex I deficiency, 252010
NDUFV2	119,2	98%	98%	Mitochondrial complex I deficiency, 252010
NDUFV3	125,3	96%	96%	No OMIM phenotype
NFS1	66,2	89%	84%	No OMIM phenotype
NFU1	86	98%	91%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NNT	96,8	100%	100%	Glucocorticoid deficiency 4, 614736
NRF1	98,4	100%	99%	No OMIM phenotype
NUBPL	87,4	100%	100%	Mitochondrial complex I deficiency, 252010
OGDH	113,8	100%	100%	No OMIM phenotype
OPA1	131,1	99%	99%	Optic atrophy 1, 165500
OPA3	108	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OXA1L	143,4	99%	97%	No OMIM phenotype
OXCT1	96,2	100%	98%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
PAM16	33,3	59%	52%	Spondylometaphyseal dysplasia,Megarbane-Dagher-Melike type,613320
PANK2	112,1	90%	86%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PARS2	153,5	100%	98%	No OMIM phenotype
PC	96,3	95%	92%	Pyruvate carboxylase deficiency, 266150
PCK2	127,8	100%	100%	PEPCK deficiency,mitochondrial,261650
PDHA1	64,3	98%	90%	Pyruvate dehydrogenase E1-alpha deficiency, 312170 Leigh syndrome, X-linked, 308930
PDHB	99,6	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	96,8	100%	100%	Lacticacidemia due to PDX1 deficiency,245349

PDK1	95,6	100%	100%	No OMIM phenotype
PDK2	74,8	100%	100%	No OMIM phenotype
PDK3	58,9	100%	98%	?Charcot-Marie-Tooth disease,X-linked dominant, 6,300905
PDK4	113,2	100%	100%	No OMIM phenotype
PDP1	154,5	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDP2	168,2	100%	100%	No OMIM phenotype
PDPR	120,3	59%	58%	No OMIM phenotype
PDSS1	94,4	90%	86%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	95,5	100%	99%	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	76,3	100%	99%	Mitochondrial complex IV deficiency, 220110
PET112	88,8	100%	100%	No OMIM phenotype
PINK1	78	93%	87%	Parkinson disease 6, early onset, 605909
PNPT1	103,6	100%	100%	Combined oxidative phosphorylation deficiency 13, 614932
POLG	90,5	98%	92%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome, 607459 Progressive external ophthalmoplegia, autosomal dominant, 157640 Progressive external ophthalmoplegia, autosomal recessive, 258450
POLG2	129,6	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131
POP1	97,3	100%	97%	No OMIM phenotype
PUS1	65,8	100%	98%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PYCR1	83	100%	96%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RARS2	83	100%	98%	Pontocerebellar hypoplasia, type 6, 611523
RMND1	86,7	95%	92%	Combined oxidative phosphorylation deficiency 11, 614922
RRM2B	113,1	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075
SAMM50	116,3	100%	100%	No OMIM phenotype
SARS2	73,6	94%	89%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	94,2	96%	95%	Mitochondrial complex IV deficiency,220110
SCO2	85,4	100%	100%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908

SDHA	9,1	30%	16%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SDHAF1	91,8	98%	93%	Mitochondrial complex II deficiency, 252011
SDHAF2	103,5	98%	94%	Paragangliomas 2, 601650
SDHB	85,5	100%	100%	Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 2, 612359 Gastrointestinal stromal tumor, 606764
SDHC	28,4	53%	45%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	42,3	52%	33%	Paragangliomas 1, with or without deafness, 168000 Pheochromocytoma, 171300 Carcinoid tumors, intestinal, 114900 Merkel cell carcinoma, somatic Paraganglioma and gastric stromal sarcoma, 606864 Cowden syndrome 3, 615106
SERAC1	89,3	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	92,5	100%	100%	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	94,7	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	105,7	100%	100%	Thiamine metabolism dysfunction syndrome 2, 607483
SLC25A1	77,6	88%	82%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A10	79	94%	82%	No OMIM phenotype
SLC25A11	118	100%	100%	No OMIM phenotype
SLC25A12	111,6	100%	100%	Hypomyelination, global cerebral, 612949
SLC25A13	99	100%	99%	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A14	52,5	99%	93%	No OMIM phenotype
SLC25A15	100,6	88%	83%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A16	110,1	100%	100%	No OMIM phenotype
SLC25A17	72,6	98%	88%	No OMIM phenotype
SLC25A18	65,5	99%	95%	No OMIM phenotype

SLC25A19	71,1	100%	97%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A2	151,2	100%	100%	No OMIM phenotype
SLC25A20	75,1	100%	100%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	112,7	100%	100%	No OMIM phenotype
SLC25A22	75,4	100%	92%	Epileptic encephalopathy, early infantile, 3, 609304
SLC25A23	107	100%	100%	No OMIM phenotype
SLC25A24	111,5	100%	95%	No OMIM phenotype
SLC25A25	102,9	100%	99%	No OMIM phenotype
SLC25A26	103,9	100%	99%	No OMIM phenotype
SLC25A27	109,5	100%	100%	No OMIM phenotype
SLC25A28	103	94%	90%	No OMIM phenotype
SLC25A29	88,1	100%	99%	No OMIM phenotype
SLC25A3	71,9	88%	85%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A30	145,9	100%	100%	No OMIM phenotype
SLC25A31	131,7	100%	100%	No OMIM phenotype
SLC25A32	110,1	100%	100%	No OMIM phenotype
SLC25A33	41,9	60%	56%	No OMIM phenotype
SLC25A34	68,6	100%	99%	No OMIM phenotype
SLC25A35	90,3	100%	99%	No OMIM phenotype
SLC25A36	93	92%	87%	No OMIM phenotype
SLC25A37	159,3	100%	100%	No OMIM phenotype
SLC25A38	74,4	100%	96%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
SLC25A39	70,4	96%	89%	No OMIM phenotype
SLC25A4	117,8	100%	98%	Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283 Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418
SLC25A40	97,8	100%	100%	No OMIM phenotype
SLC25A41	80,9	100%	97%	No OMIM phenotype
SLC25A42	61,3	100%	96%	No OMIM phenotype
SLC25A43	54,2	89%	76%	No OMIM phenotype
SLC25A44	129,7	97%	94%	No OMIM phenotype
SLC25A45	111,1	100%	97%	No OMIM phenotype
SLC25A46	110,1	100%	100%	Neuropathy,hereditary motor and sensory,type VIB,616505

SLC25A47	94,4	100%	99%	No OMIM phenotype
SLC25A48	88,6	100%	100%	No OMIM phenotype
SLC25A5	30,3	75%	64%	No OMIM phenotype
SLC25A51	2	39%	34%	No OMIM phenotype
SLC25A52	4	40%	27%	No OMIM phenotype
SLC25A53	63,4	100%	100%	No OMIM phenotype
SLC3A1	132	96%	96%	Cystinuria, 220100
SPG7	84,4	96%	87%	Spastic paraplegia 7, autosomal recessive, 607259
SUCLA2	81,6	94%	91%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with/without methylmalonic aciduria), 612073
SUCLG1	94,7	95%	91%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	84	94%	93%	No OMIM phenotype
SURF1	91,8	88%	88%	Leigh syndrome, due to COX deficiency, 256000
TACO1	90,6	91%	88%	Mitochondrial complex IV deficiency,220110
TARS2	101,9	100%	99%	?Combined oxidative phosphorylation deficiency 21,615918
TAZ	47,3	100%	98%	Barth syndrome, 302060
TFAM	68,2	100%	94%	No OMIM phenotype
TFB1M	113,3	100%	100%	No OMIM phenotype
TFB2M	97,1	100%	99%	No OMIM phenotype
TIMM10	135,2	100%	100%	No OMIM phenotype
TIMM13	70,9	100%	100%	No OMIM phenotype
TIMM17A	84,3	100%	96%	No OMIM phenotype
TIMM17B	53,6	100%	100%	No OMIM phenotype
TIMM21	114,6	92%	92%	No OMIM phenotype
TIMM22	89,4	100%	100%	No OMIM phenotype
TIMM23	14,3	18%	18%	No OMIM phenotype
TIMM44	110,5	100%	100%	No OMIM phenotype
TIMM50	106,8	100%	97%	No OMIM phenotype
TIMM8A	26,2	73%	66%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TIMM8B	67,1	62%	44%	No OMIM phenotype
TIMM9	76,8	100%	100%	No OMIM phenotype

TIMMDC1	138,6	100%	100%	No OMIM phenotype
TK2	91,6	100%	99%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560
TMEM126A	85,8	100%	99%	Optic atrophy-7, 612989
TMEM126B	76,8	100%	100%	Uli Brandt, paper in press
TMEM70	210,6	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TOMM20	56,6	89%	82%	No OMIM phenotype
TOMM22	73,7	100%	97%	No OMIM phenotype
TOMM34	106,1	100%	100%	No OMIM phenotype
TOMM40	30,5	81%	54%	No OMIM phenotype
TOMM40L	116,9	100%	100%	No OMIM phenotype
TOMM5	92,7	100%	100%	No OMIM phenotype
TOMM6	79,9	100%	100%	No OMIM phenotype
TOMM7	50,1	78%	69%	No OMIM phenotype
TOMM70A	81,9	96%	85%	No OMIM phenotype
TPK1	83,6	100%	100%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRIT1	113,1	100%	97%	No OMIM phenotype
TRMU	75,4	100%	99%	{Deafness, mitochondrial, modifier of}, 580000 Liver failure, transient infantile, 613070
TSFM	92,5	93%	87%	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	70,2	90%	83%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	100,7	99%	94%	Combined oxidative phosphorylation deficiency 4, 610678
TYMP	88,5	99%	91%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UCP1	91,8	100%	90%	{Obesity,susceptibility to},601665
UCP2	89	100%	100%	{Obesity,susceptibility to,BMIQ4},607447
UCP3	76,5	100%	93%	{Obesity,severe,and type II diabetes},601665
UQCC1	113,5	100%	100%	No OMIM phenotype
UQCC2	95,7	100%	100%	?Mitochondrial complex III deficiency,nuclear type 7,615824
UQCR10	79,8	100%	100%	No OMIM phenotype
UQCR11	108,6	100%	100%	No OMIM phenotype
UQCRB	145,3	100%	100%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	91,5	95%	94%	No OMIM phenotype
UQCRC2	88,7	97%	94%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	2,1	24%	13%	No OMIM phenotype

UQCRH	58,3	88%	88%	No OMIM phenotype
UQCRQ	63,3	100%	99%	Mitochondrial complex III deficiency, nuclear type 4, 615159
USMG5	76,3	100%	90%	No OMIM phenotype
VARS2	15,4	60%	27%	Combined oxidative phosphorylation deficiency 20,615917
VDAC1	23,3	63%	42%	No OMIM phenotype
VDAC2	56,9	88%	81%	No OMIM phenotype
VDAC3	99,2	100%	99%	No OMIM phenotype
WARS2	110,9	100%	100%	No OMIM phenotype
WFS1	145,6	100%	98%	?Cataract 41,116400 Deafness,autosomal dominant 6/14/38,600965 Wolfram syndrome,222300 Wolfram-like syndrome,autosomal dominant,614296 {Diabetes mellitus,noninsulin-dependent,association with},125853
YARS2	106,9	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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
OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015
This list is accurate for all panel versions starting with DG 2.3. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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