# MITOCHONDRIAL DISORDERS PANEL DG 3.8.1 (483 GENES)

| *Gene* | *Twist X2 covered >10x* | *Twist X2 covered >20x* | *WGS covered >10x* | *WGS covered >20x* | *Associated Phenotype description and OMIM disease ID* |
|---|---|---|---|---|---|
| AARS2 | 100.0% | 100.0% | 100.0% | 99.9% | Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096 |
| ABAT | 100.0% | 100.0% | 100.0% | 99.6% | GABA-transaminase deficiency, 613163 |
| ABCB7 | 99.8% | 99.3% | 99.3% | 77.3% | Anemia, sideroblastic, with ataxia, 301310 |
| ACAD9 | 100.0% | 100.0% | 100.0% | 99.7% | Mitochondrial complex I deficiency, nuclear type 20, 611126 |
| ACO2 | 100.0% | 100.0% | 100.0% | 99.7% | Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559 |

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| ACTA1 | 100.0% | 100.0% | 100.0% | 99.0% | Congenital myopathy 2B, severe infantile, autosomal recessive, 620265;?Myopathy, scapulohumeroperoneal, 616852;Congenital myopathy 2C, severe infantile, autosomal dominant, 620278;Congenital myopathy 2A, typical, autosomal dominant, 161800 |
| ADAMTS10 | 100.0% | 100.0% | 100.0% | 99.8% | Weill-Marchesani syndrome 1, recessive, 277600 |
| ADAR | 100.0% | 100.0% | 100.0% | 99.1% | Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010 |
| ADCK2 | 100.0% | 100.0% | 100.0% | 99.2% | |
| ADPRS | 100.0% | 100.0% | 100.0% | 99.8% | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 |
| AFG3L2 | 100.0% | 100.0% | 100.0% | 99.1% | Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246 |
| AGK | 91.7% | 91.7% | 100.0% | 99.6% | Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350 |

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| AIFM1 | 100.0% | 99.9% | 98.3% | 73.3% | Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614 |
| AK3 | 100.0% | 100.0% | 100.0% | 99.4% | |
| ALDH1B1 | 100.0% | 100.0% | 100.0% | 99.9% | |
| ALKBH1 | 100.0% | 100.0% | 100.0% | 99.5% | |
| ANO10 | 100.0% | 100.0% | 100.0% | 99.6% | Spinocerebellar ataxia, autosomal recessive 10, 613728 |
| APOO | 100.0% | 100.0% | 98.7% | 75.5% | |
| APTX | 100.0% | 100.0% | 100.0% | 99.5% | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 |
| ARL2 | 100.0% | 100.0% | 100.0% | 99.8% | ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082 |
| ARNT2 | 100.0% | 100.0% | 100.0% | 99.5% | ?Webb-Dattani syndrome, 615926 |
| ATAD1 | 100.0% | 99.7% | 100.0% | 98.9% | Hyperekplexia 4, 618011 |

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| ATAD3A | 100.0% | 100.0% | 100.0% | 99.0% | Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 |
| ATAD3B | 100.0% | 100.0% | 100.0% | 98.4% | |
| ATP13A2 | 100.0% | 100.0% | 100.0% | 99.8% | Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693 |
| ATP5F1A | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358;?Combined oxidative phosphorylation deficiency 22, 616045;?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228 |
| ATP5F1B | 100.0% | 100.0% | 100.0% | 99.7% | ?Hypermetabolism due to uncoupled mitochondrial oxidative phosphorylation 2, 620085 |
| ATP5F1C | 100.0% | 100.0% | 100.0% | 99.2% | |
| ATP5F1D | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial complex V (ATP synthase) deficiency, 618120 |

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| ATP5F1E | 100.0% | 100.0% | 100.0% | 97.8% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 |
| ATP5IF1 | 100.0% | 100.0% | 100.0% | 99.6% | |
| ATP5MC1 | 100.0% | 100.0% | 100.0% | 99.6% | |
| ATP5MC2 | 100.0% | 100.0% | 100.0% | 99.0% | |
| ATP5MC3 | 100.0% | 100.0% | 100.0% | 99.8% | Dystonia, early-onset, and/or spastic paraplegia, 619681 |
| ATP5MD | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683 |
| ATP5ME | 100.0% | 100.0% | 100.0% | 100.0% | |
| ATP5MF | 100.0% | 100.0% | 100.0% | 99.5% | |
| ATP5MG | 100.0% | 100.0% | 100.0% | 99.7% | |
| ATP5MGL | 100.0% | 100.0% | 100.0% | 99.9% | |
| ATP5PB | 100.0% | 100.0% | 100.0% | 99.8% | |
| ATP5PD | 100.0% | 100.0% | 100.0% | 99.3% | |
| ATP5PF | 100.0% | 100.0% | 100.0% | 99.2% | |
| ATP5PO | 100.0% | 100.0% | 100.0% | 99.3% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359 |
| ATPAF1 | 100.0% | 100.0% | 100.0% | 98.8% | |
| ATPAF2 | 100.0% | 100.0% | 100.0% | 99.4% | ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 |

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| BCAP31 | 99.1% | 92.8% | 99.2% | 77.3% | Deafness, dystonia, and cerebral hypomyelination, 300475 |
| BCS1L | 100.0% | 100.0% | 100.0% | 99.9% | GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000 |
| BOLA1 | 100.0% | 100.0% | 100.0% | 99.9% | |
| BOLA2 | 100.0% | 100.0% | 100.0% | 100.0% | |
| BOLA3 | 100.0% | 100.0% | 100.0% | 99.4% | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 |
| BTD | 94.4% | 94.3% | 100.0% | 99.5% | Biotinidase deficiency, 253260 |
| C12orf65 | 100.0% | 100.0% | 100.0% | 98.6% | Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559 |
| C19orf12 | 100.0% | 99.9% | 100.0% | 98.8% | Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043 |
| C1QBP | 100.0% | 100.0% | 100.0% | 99.1% | Combined oxidative phosphorylation deficiency 33, 617713 |

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| C2orf69 | 100.0% | 100.0% | 100.0% | 99.3% | Combined oxidative phosphorylation deficiency 53, 619423 |
| CA5A | 100.0% | 100.0% | 100.0% | 99.1% | Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 |
| CARS2 | 100.0% | 100.0% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 27, 616672 |
| CDKL5 | 95.7% | 95.3% | 98.0% | 71.6% | Developmental and epileptic encephalopathy 2, 300672 |
| CEP89 | 100.0% | 100.0% | 100.0% | 98.3% | |
| CFAP58 | 100.0% | 100.0% | 100.0% | 98.5% | Spermatogenic failure 49, 619144 |
| CHCHD10 | 100.0% | 100.0% | 100.0% | 99.2% | ?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 |
| CHCHD2 | 100.0% | 100.0% | 100.0% | 99.9% | Parkinson disease 22, autosomal dominant, 616710 |
| CHKB | 100.0% | 100.0% | 100.0% | 99.3% | Muscular dystrophy, congenital, megaconial type, 602541 |
| CISD2 | 100.0% | 100.0% | 100.0% | 99.4% | Wolfram syndrome 2, 604928 |

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| CLPB | 100.0% | 100.0% | 100.0% | 98.9% | Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 |
| CLPP | 100.0% | 100.0% | 100.0% | 99.3% | Perrault syndrome 3, 614129 |
| COA1 | 100.0% | 100.0% | 100.0% | 99.4% | |
| COA3 | 100.0% | 100.0% | 100.0% | 99.8% | ?Mitochondrial complex IV deficiency, nuclear type 14, 619058 |
| COA5 | 82.4% | 82.4% | 100.0% | 99.4% | ?Mitochondrial complex IV, deficiency, nuclear type 9, 616500 |
| COA6 | 100.0% | 100.0% | 100.0% | 99.4% | Mitochondrial complex IV deficiency, nuclear type 13, 616501 |
| COA7 | 100.0% | 100.0% | 100.0% | 99.6% | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 |
| COA8 | 100.0% | 99.9% | 100.0% | 98.9% | Mitochondrial complex IV deficiency, nuclear type 17, 619061 |

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| COASY | 100.0% | 100.0% | 100.0% | 99.5% | Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643 |
| COQ2 | 96.3% | 96.3% | 100.0% | 99.7% | {Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426 |
| COQ4 | 100.0% | 100.0% | 100.0% | 99.9% | Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666 |
| COQ5 | 100.0% | 100.0% | 100.0% | 99.0% | ?Coenzyme Q10 deficiency, primary, 9, 619028 |
| COQ6 | 100.0% | 100.0% | 100.0% | 99.5% | Coenzyme Q10 deficiency, primary, 6, 614650 |
| COQ7 | 100.0% | 100.0% | 100.0% | 99.6% | Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 |
| COQ8A | 100.0% | 100.0% | 100.0% | 100.0% | Coenzyme Q10 deficiency, primary, 4, 612016 |
| COQ8B | 100.0% | 100.0% | 100.0% | 99.8% | Nephrotic syndrome, type 9, 615573 |
| COQ9 | 100.0% | 100.0% | 100.0% | 99.6% | Coenzyme Q10 deficiency, primary, 5, 614654 |

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| COX10 | 100.0% | 100.0% | 99.9% | 98.3% | Mitochondrial complex IV deficiency, nuclear type 3, 619046 |
| COX11 | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex IV deficiency, nuclear type 23, 620275 |
| COX14 | 100.0% | 100.0% | 100.0% | 100.0% | ?Mitochondrial complex IV deficiency, nuclear type 10, 619053 |
| COX15 | 100.0% | 100.0% | 100.0% | 99.3% | Mitochondrial complex IV deficiency, nuclear type 6, 615119 |
| COX16 | 100.0% | 100.0% | 100.0% | 99.5% | Mitochondrial complex IV deficiency, nuclear type 22, 619355 |
| COX18 | 100.0% | 100.0% | 100.0% | 99.1% | |
| COX20 | 100.0% | 100.0% | 100.0% | 99.4% | Mitochondrial complex IV deficiency, nuclear type 11, 619054 |
| COX4I1 | 100.0% | 100.0% | 100.0% | 99.9% | Mitochondrial complex IV deficiency, nuclear type 16, 619060 |
| COX4I2 | 100.0% | 100.0% | 100.0% | 98.8% | Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714 |
| COX5A | 100.0% | 100.0% | 100.0% | 99.5% | ?Mitochondrial complex IV deficiency, nuclear type 20, 619064 |
| COX5B | 100.0% | 100.0% | 100.0% | 99.3% | |

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| COX6A1 | 100.0% | 100.0% | 100.0% | 99.4% | Charcot-Marie-Tooth disease, recessive intermediate D, 616039 |
| COX6A2 | 100.0% | 99.6% | 100.0% | 98.8% | Mitochondrial complex IV deficiency, nuclear type 18, 619062 |
| COX6B1 | 100.0% | 100.0% | 100.0% | 99.9% | Mitochondrial complex IV deficiency, nuclear type 7, 619051 |
| COX6B2 | 100.0% | 100.0% | 100.0% | 97.7% | |
| COX6C | 100.0% | 100.0% | 100.0% | 98.9% | |
| COX7A1 | 100.0% | 100.0% | 100.0% | 95.5% | |
| COX7A2 | 100.0% | 100.0% | 100.0% | 98.8% | |
| COX7B | 100.0% | 99.9% | 98.2% | 73.6% | Linear skin defects with multiple congenital anomalies 2, 300887 |
| COX7B2 | 100.0% | 100.0% | 100.0% | 99.8% | |
| COX7C | 100.0% | 100.0% | 100.0% | 99.9% | |
| COX8A | 100.0% | 100.0% | 100.0% | 100.0% | ?Mitochondrial complex IV deficiency, nuclear type 15, 619059 |
| COX8C | 100.0% | 100.0% | 100.0% | 100.0% | |
| CP | 100.0% | 100.0% | 100.0% | 99.0% | Cerebellar ataxia, 604290;[Hypoceruloplasminemia, hereditary], 604290;Hemosiderosis, systemic, due to aceruloplasminemia, 604290 |

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| CRAT | 100.0% | 100.0% | 100.0% | 99.8% | ?Neurodegeneration with brain iron accumulation 8, 617917 |
| CRLS1 | 100.0% | 100.0% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 57, 620167 |
| CTBP1 | 100.0% | 99.5% | 100.0% | 98.3% | Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 |
| CYC1 | 100.0% | 100.0% | 100.0% | 99.7% | Mitochondrial complex III deficiency, nuclear type 6, 615453 |
| CYCS | 100.0% | 100.0% | 100.0% | 99.4% | Thrombocytopenia 4, 612004 |
| D2HGDH | 100.0% | 100.0% | 100.0% | 99.7% | D-2-hydroxyglutaric aciduria, 600721 |
| DARS2 | 100.0% | 100.0% | 100.0% | 98.5% | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 |
| DCAF17 | 100.0% | 100.0% | 100.0% | 99.8% | Woodhouse-Sakati syndrome, 241080 |
| DDHD1 | 100.0% | 100.0% | 100.0% | 99.3% | Spastic paraplegia 28, autosomal recessive, 609340 |

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| DES | 100.0% | 100.0% | 100.0% | 99.8% | Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419 |
| DGUOK | 100.0% | 100.0% | 100.0% | 98.7% | Portal hypertension, noncirrhotic, 1, 617068;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070;Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 |
| DHTKD1 | 100.0% | 100.0% | 100.0% | 99.4% | ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 |
| DLAT | 100.0% | 100.0% | 100.0% | 99.4% | Pyruvate dehydrogenase E2 deficiency, 245348 |
| DLD | 100.0% | 100.0% | 100.0% | 99.3% | Dihydrolipoamide dehydrogenase deficiency, 246900 |
| DLST | 100.0% | 100.0% | 100.0% | 99.8% | Pheochromocytoma/paraganglioma syndrome 7, 618475 |
| DMAC1 | 100.0% | 100.0% | 100.0% | 99.1% | |
| DMAC2 | 100.0% | 100.0% | 100.0% | 99.3% | |

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| DMAC2L | 100.0% | 100.0% | 100.0% | 99.5% | |
| DNA2 | 100.0% | 100.0% | 100.0% | 98.9% | ?Seckel syndrome 8, 615807;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 |
| DNAJA3 | 100.0% | 100.0% | 100.0% | 99.8% | |
| DNAJC19 | 100.0% | 100.0% | 100.0% | 99.6% | 3-methylglutaconic aciduria, type V, 610198 |
| DNAJC3 | 100.0% | 100.0% | 100.0% | 98.5% | Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 |
| DNAJC30 | 100.0% | 100.0% | 100.0% | 100.0% | Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382 |
| DNM1L | 100.0% | 100.0% | 100.0% | 99.5% | Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 |
| DNM2 | 100.0% | 100.0% | 100.0% | 99.6% | Centronuclear myopathy 1, 160150;Charcot-Marie-Tooth disease, axonal type 2M, 606482;Charcot-Marie-Tooth disease, dominant intermediate B, 606482;Lethal congenital contracture syndrome 5, 615368 |

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| EARS2 | 100.0% | 100.0% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 12, 614924 |
| ECHS1 | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 |
| ECSIT | 100.0% | 100.0% | 100.0% | 99.8% | |
| EHHADH | 100.0% | 100.0% | 100.0% | 99.6% | ?Fanconi renotubular syndrome 3, 615605 |
| ELAC2 | 100.0% | 100.0% | 100.0% | 99.7% | {Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440 |
| EMD | 100.0% | 99.5% | 98.9% | 74.6% | Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 |
| EPG5 | 100.0% | 100.0% | 100.0% | 99.3% | Vici syndrome, 242840 |
| ERAL1 | 100.0% | 100.0% | 100.0% | 99.7% | Perrault syndrome 6, 617565 |
| ETFDH | 100.0% | 100.0% | 100.0% | 99.1% | Glutaric acidemia IIC, 231680 |
| ETHE1 | 100.0% | 100.0% | 100.0% | 99.3% | Ethylmalonic encephalopathy, 602473 |
| EXOSC8 | 100.0% | 100.0% | 99.9% | 99.0% | Pontocerebellar hypoplasia, type 1C, 616081 |
| FA2H | 100.0% | 100.0% | 100.0% | 99.8% | Spastic paraplegia 35, autosomal recessive, 612319 |

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| FARS2 | 100.0% | 100.0% | 100.0% | 99.5% | Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046 |
| FARSB | 100.0% | 100.0% | 100.0% | 99.3% | Rajab interstitial lung disease with brain calcifications 1, 613658 |
| FASTKD2 | 100.0% | 100.0% | 100.0% | 98.3% | Combined oxidative phosphorylation deficiency 44, 618855 |
| FBXL4 | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 |
| FDX2 | 100.0% | 100.0% | 100.0% | 99.2% | Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900 |
| FDXR | 100.0% | 100.0% | 100.0% | 99.8% | Auditory neuropathy and optic atrophy, 617717 |
| FH | 100.0% | 100.0% | 100.0% | 99.2% | Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812 |
| FLAD1 | 100.0% | 100.0% | 100.0% | 99.8% | Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 |

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| FOXRED1 | 100.0% | 100.0% | 100.0% | 99.4% | Mitochondrial complex I deficiency, nuclear type 19, 618241 |
| FTL | 100.0% | 100.0% | 100.0% | 99.2% | Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159 |
| FXN | 100.0% | 100.0% | 100.0% | 99.0% | Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300 |
| GARS1 | 100.0% | 100.0% | 100.0% | 99.4% | Spinal muscular atrophy, infantile, James type, 619042;Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794;Charcot-Marie-Tooth disease, type 2D, 601472 |
| GATB | 100.0% | 100.0% | 100.0% | 99.4% | ?Combined oxidative phosphorylation deficiency 41, 618838 |
| GATC | 100.0% | 100.0% | 100.0% | 99.9% | Combined oxidative phosphorylation deficiency 42, 618839 |
| GATM | 100.0% | 100.0% | 100.0% | 99.6% | Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600 |

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| GBF1 | 100.0% | 100.0% | 100.0% | 99.4% | Charcot-Marie-Tooth disease, axonal, type 2GG, 606483 |
| GCSH | 100.0% | 100.0% | 100.0% | 99.3% | Multiple mitochondrial dysfunctions syndrome 7, 620423 |
| GDAP1 | 100.0% | 100.0% | 100.0% | 99.4% | Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706;Charcot-Marie-Tooth disease, recessive intermediate, A, 608340;Charcot-Marie-Tooth disease, axonal, type 2K, 607831;Charcot-Marie-Tooth disease, type 4A, 214400 |
| GFER | 100.0% | 100.0% | 100.0% | 99.8% | Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 |
| GFM1 | 100.0% | 100.0% | 100.0% | 99.3% | Combined oxidative phosphorylation deficiency 1, 609060 |
| GFM2 | 100.0% | 100.0% | 100.0% | 99.3% | Combined oxidative phosphorylation deficiency 39, 618397 |
| GLRX5 | 100.0% | 100.0% | 100.0% | 99.6% | Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859 |

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| GLUD1 | 100.0% | 100.0% | 100.0% | 99.4% | Hyperinsulinism-hyperammonemia syndrome, 606762 |
| GMPR | 100.0% | 100.0% | 100.0% | 99.7% | |
| GOT2 | 100.0% | 100.0% | 100.0% | 99.7% | Developmental and epileptic encephalopathy 82, 618721 |
| GPT2 | 100.0% | 100.0% | 100.0% | 99.8% | Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 |
| GTPBP2 | 100.0% | 100.0% | 100.0% | 99.3% | Jaberi-Elahi syndrome, 617988 |
| GTPBP3 | 100.0% | 100.0% | 100.0% | 99.7% | Combined oxidative phosphorylation deficiency 23, 616198 |
| GUF1 | 100.0% | 100.0% | 100.0% | 98.4% | ?Developmental and epileptic encephalopathy 40, 617065 |
| HACE1 | 100.0% | 100.0% | 100.0% | 99.3% | Spastic paraplegia and psychomotor retardation with or without seizures, 616756 |
| HADHA | 100.0% | 100.0% | 100.0% | 99.3% | HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016 |

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| HADHB | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial trifunctional protein deficiency 2, 620300 |
| HARS2 | 100.0% | 100.0% | 100.0% | 99.6% | Perrault syndrome 2, 614926 |
| HCCS | 100.0% | 100.0% | 98.1% | 72.8% | Linear skin defects with multiple congenital anomalies 1, 309801 |
| HIBCH | 100.0% | 100.0% | 100.0% | 98.8% | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 |
| HLCS | 100.0% | 100.0% | 100.0% | 99.6% | Holocarboxylase synthetase deficiency, 253270 |
| HPDL | 100.0% | 100.0% | 100.0% | 99.4% | Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027 |
| HSD17B10 | 100.0% | 99.8% | 99.6% | 75.5% | HSD10 mitochondrial disease, 300438 |
| HSPA9 | 100.0% | 100.0% | 100.0% | 99.4% | Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170 |
| HSPD1 | 100.0% | 100.0% | 100.0% | 99.2% | Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233 |
| HTRA2 | 100.0% | 100.0% | 100.0% | 99.3% | {Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248 |

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| IARS2 | 100.0% | 100.0% | 100.0% | 99.5% | Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 |
| IBA57 | 100.0% | 100.0% | 100.0% | 100.0% | Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451 |
| IDH2 | 100.0% | 100.0% | 100.0% | 99.6% | D-2-hydroxyglutaric aciduria 2, 613657 |
| ISCA1 | 92.4% | 92.4% | 100.0% | 99.7% | Multiple mitochondrial dysfunctions syndrome 5, 617613 |
| ISCA2 | 100.0% | 100.0% | 100.0% | 99.6% | Multiple mitochondrial dysfunctions syndrome 4, 616370 |
| ISCU | 100.0% | 100.0% | 100.0% | 99.7% | Myopathy with lactic acidosis, hereditary, 255125 |

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| KARS1 | 100.0% | 100.0% | 100.0% | 99.4% | Deafness, autosomal recessive 89, 613916;Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147;?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 |
| KIF1A | 100.0% | 100.0% | 100.0% | 99.9% | NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 610357 |
| LACTB | 100.0% | 100.0% | 99.9% | 98.8% | |
| LARS2 | 100.0% | 100.0% | 100.0% | 99.5% | Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021 |
| LDHD | 100.0% | 100.0% | 100.0% | 99.9% | D-lactic aciduria with susceptibility to gout, 245450 |

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| LETM1 | 100.0% | 100.0% | 100.0% | 99.7% | Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 |
| LIAS | 100.0% | 100.0% | 100.0% | 99.7% | Hyperglycinemia, lactic acidosis, and seizures, 614462 |
| LIG3 | 100.0% | 100.0% | 100.0% | 99.5% | Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 |
| LIPT1 | 100.0% | 100.0% | 100.0% | 98.6% | Lipoyltransferase 1 deficiency, 616299 |
| LIPT2 | 100.0% | 100.0% | 100.0% | 99.6% | Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 |
| LONP1 | 100.0% | 100.0% | 100.0% | 99.8% | CODAS syndrome, 600373 |
| LRPPRC | 100.0% | 100.0% | 100.0% | 99.3% | Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 |
| LYRM4 | 68.0% | 68.0% | 100.0% | 99.8% | ?Combined oxidative phosphorylation deficiency 19, 615595 |
| LYRM7 | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex III deficiency, nuclear type 8, 615838 |

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| MAPT | 100.0% | 100.0% | 100.0% | 99.6% | Supranuclear palsy, progressive, 601104;Supranuclear palsy, progressive atypical, 260540;Dementia, frontotemporal, with or without parkinsonism, 600274;{Parkinson disease, susceptibility to}, 168600;Pick disease, 172700 |
| MARS2 | 100.0% | 100.0% | 100.0% | 99.9% | ?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390 |
| MCAT | 100.0% | 100.0% | 100.0% | 99.6% | Optic atrophy 15, 620583 |
| MCUR1 | 100.0% | 100.0% | 100.0% | 99.2% | |
| MDH1 | 100.0% | 100.0% | 100.0% | 99.6% | ?Developmental and epileptic encephalopathy 88, 618959 |
| MDH2 | 100.0% | 100.0% | 100.0% | 99.3% | Developmental and epileptic encephalopathy 51, 617339 |
| MECR | 100.0% | 100.0% | 100.0% | 99.6% | Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282;Optic atrophy 16, 620629 |
| MFF | 100.0% | 100.0% | 100.0% | 99.6% | Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 |

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| MFN2 | 100.0% | 100.0% | 100.0% | 99.5% | Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800;Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260;Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087;Hereditary motor and sensory neuropathy VIA, 601152 |
| MGME1 | 100.0% | 100.0% | 100.0% | 97.9% | Mitochondrial DNA depletion syndrome 11, 615084 |
| MICOS13 | 100.0% | 100.0% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 37, 618329 |
| MICU1 | 100.0% | 99.9% | 100.0% | 99.7% | Myopathy with extrapyramidal signs, 615673 |
| MICU2 | 100.0% | 100.0% | 100.0% | 97.8% | |
| MIEF2 | 100.0% | 100.0% | 100.0% | 99.9% | ?Combined oxidative phosphorylation deficiency 49, 619024 |
| MIPEP | 100.0% | 100.0% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 31, 617228 |

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| MORC2 | 100.0% | 100.0% | 100.0% | 99.6% | Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 |
| MPC1 | 100.0% | 100.0% | 100.0% | 99.7% | Mitochondrial pyruvate carrier deficiency, 614741 |
| MPC2 | 100.0% | 100.0% | 100.0% | 98.9% | |
| MPV17 | 100.0% | 100.0% | 100.0% | 99.7% | Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 |
| MRM2 | 97.0% | 97.0% | 100.0% | 99.0% | ?Mitochondrial DNA depletion syndrome 17, 618567 |
| MRPL12 | 100.0% | 100.0% | 100.0% | 100.0% | ?Combined oxidative phosphorylation deficiency 45, 618951 |
| MRPL24 | 100.0% | 100.0% | 100.0% | 99.1% | |
| MRPL3 | 100.0% | 100.0% | 100.0% | 99.7% | Combined oxidative phosphorylation deficiency 9, 614582 |
| MRPL39 | 100.0% | 100.0% | 100.0% | 99.4% | Combined oxidative phosphorylation deficiency 59, 620646 |
| MRPL40 | 100.0% | 100.0% | 100.0% | 99.6% | |

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| MRPL44 | 100.0% | 100.0% | 100.0% | 99.4% | Combined oxidative phosphorylation deficiency 16, 615395 |
| MRPL50 | 100.0% | 100.0% | 100.0% | 98.8% | |
| MRPL57 | 100.0% | 100.0% | 100.0% | 100.0% | |
| MRPS14 | 100.0% | 100.0% | 100.0% | 99.8% | ?Combined oxidative phosphorylation deficiency 38, 618378 |
| MRPS16 | 100.0% | 100.0% | 100.0% | 99.9% | Combined oxidative phosphorylation deficiency 2, 610498 |
| MRPS2 | 100.0% | 100.0% | 100.0% | 99.9% | Combined oxidative phosphorylation deficiency 36, 617950 |
| MRPS22 | 100.0% | 100.0% | 100.0% | 98.9% | Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719 |
| MRPS23 | 100.0% | 100.0% | 100.0% | 99.9% | ?Combined oxidative phosphorylation deficiency 46, 618952 |
| MRPS25 | 83.2% | 83.2% | 100.0% | 99.5% | ?Combined oxidative phosphorylation deficiency 50, 619025 |
| MRPS28 | 85.4% | 85.3% | 100.0% | 97.7% | ?Combined oxidative phosphorylation deficiency 47, 618958 |
| MRPS34 | 100.0% | 100.0% | 100.0% | 99.9% | Combined oxidative phosphorylation deficiency 32, 617664 |

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| MRPS36 | 100.0% | 100.0% | 100.0% | 99.0% | |
| MRPS7 | 100.0% | 100.0% | 100.0% | 99.9% | ?Combined oxidative phosphorylation deficiency 34, 617872 |
| MRRF | 100.0% | 100.0% | 100.0% | 99.5% | |
| MSTO1 | 100.0% | 100.0% | 100.0% | 99.3% | Myopathy, mitochondrial, and ataxia, 617675 |
| MTFMT | 100.0% | 100.0% | 100.0% | 99.4% | Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248 |
| MTO1 | 93.7% | 91.1% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 10, 614702 |
| MTPAP | 100.0% | 100.0% | 100.0% | 99.3% | ?Spastic ataxia 4, autosomal recessive, 613672 |
| MTX2 | 100.0% | 99.9% | 100.0% | 99.4% | Mandibuloacral dysplasia progeroid syndrome, 619127 |
| NADK2 | 100.0% | 100.0% | 100.0% | 99.2% | 2,4-dienoyl-CoA reductase deficiency, 616034 |
| NARS2 | 100.0% | 100.0% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434 |

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| NAXD | 100.0% | 100.0% | 100.0% | 99.9% | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 |
| NAXE | 100.0% | 100.0% | 100.0% | 99.7% | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 |
| NDUFA1 | 100.0% | 100.0% | 97.0% | 73.1% | Mitochondrial complex I deficiency, nuclear type 12, 301020 |
| NDUFA10 | 100.0% | 100.0% | 100.0% | 99.1% | Mitochondrial complex I deficiency, nuclear type 22, 618243 |
| NDUFA11 | 100.0% | 98.8% | 100.0% | 98.5% | Mitochondrial complex I deficiency, nuclear type 14, 618236 |
| NDUFA12 | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex I deficiency, nuclear type 23, 618244 |
| NDUFA13 | 100.0% | 100.0% | 100.0% | 99.7% | {Thyroid carcinoma, Hurthle cell}, 607464;?Mitochondrial complex I deficiency, nuclear type 28, 618249 |
| NDUFA2 | 100.0% | 100.0% | 100.0% | 99.7% | Mitochondrial complex I deficiency, nuclear type 13, 618235 |
| NDUFA3 | 91.4% | 86.8% | 100.0% | 99.6% | |

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| NDUFA4 | 100.0% | 100.0% | 100.0% | 98.5% | ?Mitochondrial complex IV deficiency, nuclear type 21, 619065 |
| NDUFA5 | 100.0% | 100.0% | 100.0% | 99.3% | |
| NDUFA6 | 100.0% | 100.0% | 100.0% | 99.9% | Mitochondrial complex I deficiency, nuclear type 33, 618253 |
| NDUFA7 | 100.0% | 100.0% | 100.0% | 99.8% | |
| NDUFA8 | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial complex I deficiency, nuclear type 37, 619272 |
| NDUFA9 | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex I deficiency, nuclear type 26, 618247 |
| NDUFAB1 | 100.0% | 100.0% | 100.0% | 98.6% | |
| NDUFAF1 | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex I deficiency, nuclear type 11, 618234 |
| NDUFAF2 | 100.0% | 100.0% | 100.0% | 99.7% | Mitochondrial complex I deficiency, nuclear type 10, 618233 |
| NDUFAF3 | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex I deficiency, nuclear type 18, 618240 |
| NDUFAF4 | 100.0% | 100.0% | 100.0% | 98.6% | Mitochondrial complex I deficiency, nuclear type 15, 618237 |
| NDUFAF5 | 100.0% | 100.0% | 100.0% | 98.1% | Mitochondrial complex I deficiency, nuclear type 16, 618238 |

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| NDUFAF6 | 100.0% | 100.0% | 100.0% | 98.6% | Mitochondrial complex I deficiency, nuclear type 17, 618239;Fanconi renotubular syndrome 5, 618913 |
| NDUFAF7 | 100.0% | 100.0% | 100.0% | 99.4% | |
| NDUFAF8 | 100.0% | 100.0% | 100.0% | 99.9% | Mitochondrial complex I deficiency, nuclear type 34, 618776 |
| NDUFB1 | 100.0% | 100.0% | 100.0% | 97.6% | |
| NDUFB10 | 100.0% | 100.0% | 100.0% | 98.2% | ?Mitochondrial complex I deficiency, nuclear type 35, 619003 |
| NDUFB11 | 99.7% | 97.9% | 93.6% | 63.2% | Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021 |
| NDUFB2 | 100.0% | 100.0% | 100.0% | 99.8% | |
| NDUFB3 | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial complex I deficiency, nuclear type 25, 618246 |
| NDUFB4 | 100.0% | 100.0% | 100.0% | 98.9% | |
| NDUFB5 | 100.0% | 100.0% | 100.0% | 99.0% | |
| NDUFB6 | 100.0% | 100.0% | 100.0% | 99.2% | |
| NDUFB7 | 100.0% | 100.0% | 100.0% | 99.3% | ?Mitochondrial complex I deficiency, nuclear type 39, 620135 |

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| NDUFB8 | 100.0% | 100.0% | 100.0% | 99.2% | Mitochondrial complex I deficiency, nuclear type 32, 618252 |
| NDUFB9 | 100.0% | 100.0% | 100.0% | 99.4% | ?Mitochondrial complex I deficiency, nuclear type 24, 618245 |
| NDUFC1 | 100.0% | 100.0% | 100.0% | 99.6% | |
| NDUFC2 | 100.0% | 100.0% | 100.0% | 98.9% | Mitochondrial complex I deficiency, nuclear type 36, 619170 |
| NDUFS1 | 100.0% | 100.0% | 100.0% | 99.1% | Mitochondrial complex I deficiency, nuclear type 5, 618226 |
| NDUFS2 | 100.0% | 100.0% | 100.0% | 99.2% | ?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228 |
| NDUFS3 | 96.6% | 91.3% | 100.0% | 99.5% | Mitochondrial complex I deficiency, nuclear type 8, 618230 |
| NDUFS4 | 100.0% | 99.9% | 100.0% | 99.1% | Mitochondrial complex I deficiency, nuclear type 1, 252010 |
| NDUFS5 | 100.0% | 100.0% | 100.0% | 99.6% | |
| NDUFS6 | 100.0% | 100.0% | 99.9% | 99.0% | Mitochondrial complex I deficiency, nuclear type 9, 618232 |

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| NDUFS7 | 100.0% | 100.0% | 100.0% | 100.0% | Mitochondrial complex I deficiency, nuclear type 3, 618224 |
| NDUFS8 | 100.0% | 100.0% | 100.0% | 99.9% | Mitochondrial complex I deficiency, nuclear type 2, 618222 |
| NDUFV1 | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex I deficiency, nuclear type 4, 618225 |
| NDUFV2 | 100.0% | 100.0% | 100.0% | 99.5% | Mitochondrial complex I deficiency, nuclear type 7, 618229 |
| NDUFV3 | 100.0% | 100.0% | 100.0% | 99.3% | |
| NFS1 | 89.8% | 89.8% | 100.0% | 99.7% | Combined oxidative phosphorylation deficiency 52, 619386 |
| NFU1 | 100.0% | 100.0% | 100.0% | 99.4% | Multiple mitochondrial dysfunctions syndrome 1, 605711 |
| NGLY1 | 100.0% | 100.0% | 100.0% | 99.3% | Congenital disorder of deglycosylation 1, 615273 |
| NME3 | 100.0% | 100.0% | 100.0% | 99.9% | |
| NR2F1 | 100.0% | 99.9% | 100.0% | 98.5% | Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 |
| NRROS | 100.0% | 100.0% | 100.0% | 99.8% | Seizures, early-onset, with neurodegeneration and brain calcification, 618875 |

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| NSUN3 | 100.0% | 100.0% | 100.0% | 99.5% | Combined oxidative phosphorylation deficiency 48, 619012 |
| NUBPL | 100.0% | 100.0% | 100.0% | 99.5% | Mitochondrial complex I deficiency, nuclear type 21, 618242 |
| NUP54 | 100.0% | 100.0% | 100.0% | 99.5% | Dystonia 37, early-onset, with striatal lesions, 620427 |
| NUTF2 | 100.0% | 100.0% | 100.0% | 99.4% | |
| OGDH | 100.0% | 100.0% | 100.0% | 99.7% | Oxoglutarate dehydrogenase deficiency, 203740 |
| OGDHL | 100.0% | 100.0% | 100.0% | 99.7% | Yoon-Bellen neurodevelopmental syndrome, 619701 |
| OPA1 | 100.0% | 100.0% | 100.0% | 99.4% | Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 |
| OPA3 | 100.0% | 100.0% | 100.0% | 99.8% | 3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300 |

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| OTX2 | 100.0% | 100.0% | 100.0% | 99.6% | Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125 |
| OXA1L | 100.0% | 100.0% | 100.0% | 99.3% | |
| P4HTM | 100.0% | 100.0% | 100.0% | 99.4% | Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 |
| PANK2 | 100.0% | 100.0% | 100.0% | 99.6% | HARP syndrome, 607236;Neurodegeneration with brain iron accumulation 1, 234200 |
| PARS2 | 100.0% | 100.0% | 100.0% | 99.7% | Developmental and epileptic encephalopathy 75, 618437 |
| PC | 100.0% | 100.0% | 100.0% | 99.8% | Pyruvate carboxylase deficiency, 266150 |
| PDE2A | 100.0% | 100.0% | 100.0% | 99.5% | Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 |
| PDHA1 | 99.7% | 97.5% | 99.1% | 75.1% | Pyruvate dehydrogenase E1-alpha deficiency, 312170 |
| PDHB | 100.0% | 100.0% | 100.0% | 99.2% | Pyruvate dehydrogenase E1-beta deficiency, 614111 |

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| PDHX | 100.0% | 99.8% | 100.0% | 99.4% | Lacticacidemia due to PDX1 deficiency, 245349 |
| PDK1 | 100.0% | 100.0% | 100.0% | 99.3% | |
| PDK2 | 100.0% | 100.0% | 100.0% | 99.8% | |
| PDK3 | 100.0% | 100.0% | 99.2% | 77.9% | ?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905 |
| PDK4 | 100.0% | 100.0% | 100.0% | 99.4% | |
| PDP1 | 100.0% | 100.0% | 100.0% | 99.9% | Pyruvate dehydrogenase phosphatase deficiency, 608782 |
| PDSS1 | 100.0% | 100.0% | 100.0% | 99.6% | Coenzyme Q10 deficiency, primary, 2, 614651 |
| PDSS2 | 100.0% | 100.0% | 100.0% | 99.3% | Coenzyme Q10 deficiency, primary, 3, 614652 |
| PET100 | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial complex IV deficiency, nuclear type 12, 619055 |
| PET117 | 100.0% | 100.0% | 100.0% | 98.8% | ?Mitochondrial complex IV deficiency, nuclear type 19, 619063 |
| PIGA | 100.0% | 100.0% | 98.6% | 74.5% | Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 |

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| PISD | 100.0% | 100.0% | 100.0% | 99.9% | Liberfarb syndrome, 618889 |
| PITRM1 | 100.0% | 100.0% | 100.0% | 99.7% | Spinocerebellar ataxia, autosomal recessive 30, 619405 |
| PLA2G6 | 100.0% | 99.9% | 100.0% | 99.5% | Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600 |
| PLPBP | 100.0% | 100.0% | 100.0% | 99.5% | Epilepsy, early-onset, 1, vitamin B6-dependent, 617290 |
| PMPCA | 100.0% | 100.0% | 100.0% | 99.9% | Spinocerebellar ataxia, autosomal recessive 2, 213200 |
| PMPCB | 100.0% | 100.0% | 100.0% | 99.4% | Multiple mitochondrial dysfunctions syndrome 6, 617954 |
| PNPLA8 | 100.0% | 100.0% | 100.0% | 98.7% | ?Mitochondrial myopathy with lactic acidosis, 251950 |
| PNPT1 | 100.0% | 100.0% | 100.0% | 99.3% | Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932 |

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| POLG | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450 |
| POLG2 | 100.0% | 100.0% | 100.0% | 99.0% | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131;?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528;?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425 |
| POLR2A | 100.0% | 100.0% | 100.0% | 99.9% | Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 |

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| POLRMT | 100.0% | 100.0% | 100.0% | 99.9% | Combined oxidative phosphorylation deficiency 55, 619743 |
| PPA2 | 100.0% | 99.9% | 100.0% | 98.7% | ?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222 |
| PPCS | 100.0% | 100.0% | 100.0% | 99.8% | Cardiomyopathy, dilated, 2C, 618189 |
| PRDX3 | 100.0% | 100.0% | 100.0% | 99.7% | Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871 |
| PRKAA1 | 100.0% | 100.0% | 100.0% | 99.0% | |
| PRORP | 100.0% | 100.0% | 100.0% | 99.1% | Combined oxidative phosphorylation deficiency 54, 619737 |
| PRPS1 | 100.0% | 100.0% | 99.0% | 75.1% | Arts syndrome, 301835;Phosphoribosylpyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661 |
| PTCD3 | 100.0% | 100.0% | 100.0% | 99.5% | Combined oxidative phosphorylation deficiency 51, 619057 |

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| PTPMT1 | 100.0% | 100.0% | 100.0% | 99.6% | |
| PTRH2 | 100.0% | 100.0% | 100.0% | 99.9% | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 |
| PUS1 | 100.0% | 100.0% | 100.0% | 99.8% | Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 |
| PYCR1 | 100.0% | 100.0% | 100.0% | 100.0% | Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940 |
| PYCR2 | 100.0% | 100.0% | 100.0% | 99.9% | Leukodystrophy, hypomyelinating, 10, 616420 |
| PYROXD1 | 100.0% | 100.0% | 100.0% | 99.0% | Myopathy, myofibrillar, 8, 617258 |
| PYROXD2 | 100.0% | 100.0% | 100.0% | 99.6% | |
| QRSL1 | 100.0% | 100.0% | 100.0% | 99.2% | Combined oxidative phosphorylation deficiency 40, 618835 |
| RANBP2 | 100.0% | 100.0% | 100.0% | 98.7% | {Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 |
| RARS2 | 100.0% | 100.0% | 100.0% | 99.5% | Pontocerebellar hypoplasia, type 6, 611523 |
| RMND1 | 100.0% | 100.0% | 100.0% | 99.3% | Combined oxidative phosphorylation deficiency 11, 614922 |

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| RNASEH1 | 100.0% | 100.0% | 100.0% | 99.3% | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479 |
| RNF213 | 100.0% | 100.0% | 100.0% | 99.7% | {Moyamoya disease 2, susceptibility to}, 607151 |
| RRM1 | 100.0% | 100.0% | 100.0% | 99.5% | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647 |
| RRM2B | 100.0% | 100.0% | 100.0% | 98.9% | Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 |
| RTN4IP1 | 100.0% | 100.0% | 100.0% | 99.4% | Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732 |

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| RYR1 | 100.0% | 99.9% | 100.0% | 99.7% | Congenital myopathy 1B, autosomal recessive, 255320;Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000;King-Denborough syndrome, 619542;{Malignant hyperthermia susceptibility 1}, 145600 |
| SACS | 100.0% | 100.0% | 100.0% | 99.2% | Spastic ataxia, Charlevoix-Saguenay type, 270550 |
| SAMHD1 | 100.0% | 100.0% | 100.0% | 99.2% | ?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952 |
| SARS2 | 100.0% | 100.0% | 100.0% | 99.5% | Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 |
| SATB2 | 100.0% | 99.7% | 100.0% | 99.4% | Glass syndrome, 612313 |
| SCO1 | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial complex IV deficiency, nuclear type 4, 619048 |
| SCO2 | 100.0% | 100.0% | 100.0% | 99.8% | Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377 |
| SCP2 | 100.0% | 100.0% | 100.0% | 99.3% | ?Leukoencephalopathy with dystonia and motor neuropathy, 613724 |

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| SDHA | 100.0% | 100.0% | 100.0% | 99.9% | Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma /paraganglioma syndrome 5, 614165 |
| SDHAF1 | 100.0% | 100.0% | 100.0% | 100.0% | Mitochondrial complex II deficiency, nuclear type 2, 619166 |
| SDHB | 100.0% | 100.0% | 100.0% | 99.5% | Pheochromocytoma/paraganglioma syndrome 4, 115310;Mitochondrial complex II deficiency, nuclear type 4, 619224;Gastrointestinal stromal tumor, 606764;Paraganglioma and gastric stromal sarcoma, 606864 |
| SDHD | 78.9% | 78.9% | 100.0% | 99.6% | Pheochromocytoma/paraganglioma syndrome 1, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Mitochondrial complex II deficiency, nuclear type 3, 619167 |
| SERAC1 | 100.0% | 100.0% | 100.0% | 99.1% | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 |

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| SFXN4 | 100.0% | 100.0% | 100.0% | 97.9% | Combined oxidative phosphorylation deficiency 18, 615578 |
| SHMT2 | 100.0% | 100.0% | 100.0% | 99.7% | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 |
| SLC19A2 | 100.0% | 100.0% | 100.0% | 99.6% | Thiamine-responsive megaloblastic anemia syndrome, 249270 |
| SLC19A3 | 99.6% | 98.4% | 100.0% | 99.6% | Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483 |
| SLC25A1 | 100.0% | 100.0% | 100.0% | 99.0% | Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197 |
| SLC25A10 | 100.0% | 100.0% | 100.0% | 100.0% | ?Mitochondrial DNA depletion syndrome 19, 618972 |
| SLC25A12 | 100.0% | 100.0% | 100.0% | 99.4% | Developmental and epileptic encephalopathy 39, 612949 |
| SLC25A13 | 100.0% | 100.0% | 100.0% | 99.5% | Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471 |

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| SLC25A19 | 100.0% | 100.0% | 100.0% | 99.5% | Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 |
| SLC25A21 | 100.0% | 100.0% | 100.0% | 99.1% | ?Mitochondrial DNA depletion syndrome 18, 618811 |
| SLC25A22 | 100.0% | 100.0% | 100.0% | 99.8% | Developmental and epileptic encephalopathy 3, 609304 |
| SLC25A24 | 99.5% | 99.5% | 99.3% | 97.2% | Fontaine progeroid syndrome, 612289 |
| SLC25A26 | 100.0% | 100.0% | 100.0% | 99.2% | Combined oxidative phosphorylation deficiency 28, 616794 |
| SLC25A3 | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial phosphate carrier deficiency, 610773 |
| SLC25A32 | 100.0% | 100.0% | 100.0% | 99.8% | ?Exercise intolerance, riboflavin-responsive, 616839 |
| SLC25A36 | 100.0% | 100.0% | 100.0% | 99.4% | Hyperinsulinemic hypoglycemia, familial, 8, 620211 |
| SLC25A38 | 100.0% | 100.0% | 100.0% | 99.4% | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 |

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| SLC25A4 | 100.0% | 100.0% | 100.0% | 99.5% | Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283;Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 |
| SLC25A42 | 100.0% | 100.0% | 100.0% | 99.9% | Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 |
| SLC25A46 | 100.0% | 100.0% | 100.0% | 99.1% | Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303 |
| SLC39A8 | 100.0% | 100.0% | 100.0% | 99.3% | Congenital disorder of glycosylation, type IIn, 616721 |
| SLC52A2 | 100.0% | 100.0% | 100.0% | 100.0% | Brown-Vialetto-Van Laere syndrome 2, 614707 |
| SLC52A3 | 100.0% | 100.0% | 100.0% | 99.9% | ?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530 |
| SLC8B1 | 100.0% | 100.0% | 100.0% | 99.7% | |

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| SLIRP | 100.0% | 100.0% | 100.0% | 98.0% | |
| SMDT1 | 100.0% | 100.0% | 100.0% | 99.7% | |
| SOD2 | 100.0% | 100.0% | 100.0% | 99.4% | {Microvascular complications of diabetes 6}, 612634 |
| SPART | 100.0% | 100.0% | 100.0% | 99.3% | Troyer syndrome, 275900 |
| SPATA5 | 100.0% | 100.0% | 100.0% | 99.6% | Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 |
| SPG7 | 100.0% | 100.0% | 100.0% | 99.6% | Spastic paraplegia 7, autosomal recessive, 607259 |
| SPTBN4 | 100.0% | 100.0% | 100.0% | 99.5% | Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 |
| SQOR | 100.0% | 100.0% | 100.0% | 99.0% | Sulfide:quinone oxidoreductase deficiency, 619221 |
| SQSTM1 | 100.0% | 100.0% | 100.0% | 99.7% | Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437;Myopathy, distal, with rimmed vacuoles, 617158;Paget disease of bone 3, 167250 |

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| SSBP1 | 100.0% | 100.0% | 100.0% | 99.4% | Optic atrophy 13 with retinal and foveal abnormalities, 165510 |
| STAC3 | 100.0% | 100.0% | 100.0% | 99.3% | Congenital myopathy 13, 255995 |
| STAT2 | 100.0% | 100.0% | 100.0% | 99.7% | Pseudo-TORCH syndrome 3, 618886;Immunodeficiency 44, 616636 |
| STXBP1 | 100.0% | 100.0% | 100.0% | 99.6% | Developmental and epileptic encephalopathy 4, 612164 |
| SUCLA2 | 100.0% | 99.6% | 100.0% | 99.4% | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 |
| SUCLG1 | 100.0% | 100.0% | 100.0% | 98.6% | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 |
| SUCLG2 | 100.0% | 99.8% | 100.0% | 99.1% | |
| SUPV3L1 | 100.0% | 100.0% | 100.0% | 99.4% | |
| SURF1 | 100.0% | 100.0% | 100.0% | 99.4% | Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110 |
| SZT2 | 100.0% | 100.0% | 100.0% | 99.7% | Developmental and epileptic encephalopathy 18, 615476 |

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| TACO1 | 100.0% | 100.0% | 100.0% | 99.5% | Mitochondrial complex IV deficiency, nuclear type 8, 619052 |
| TAMM41 | 100.0% | 100.0% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 56, 620139 |
| TANGO2 | 100.0% | 100.0% | 100.0% | 99.4% | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 |
| TAOK1 | 100.0% | 100.0% | 100.0% | 99.0% | Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575 |
| TARS2 | 100.0% | 100.0% | 100.0% | 99.5% | Combined oxidative phosphorylation deficiency 21, 615918 |
| TAZ | 100.0% | 100.0% | 99.3% | 74.1% | Barth syndrome, 302060 |
| TBCK | 100.0% | 100.0% | 100.0% | 99.5% | Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 |
| TDP2 | 100.0% | 100.0% | 100.0% | 99.6% | Spinocerebellar ataxia, autosomal recessive 23, 616949 |
| TEFM | 100.0% | 100.0% | 100.0% | 99.2% | Combined oxidative phosphorylation deficiency 58, 620451 |

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| TFAM | 100.0% | 100.0% | 100.0% | 99.4% | ?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156 |
| TFB2M | 100.0% | 100.0% | 100.0% | 99.5% | |
| THG1L | 100.0% | 100.0% | 100.0% | 99.2% | Spinocerebellar ataxia, autosomal recessive 28, 618800 |
| TIMM22 | 100.0% | 100.0% | 100.0% | 99.8% | ?Combined oxidative phosphorylation deficiency 43, 618851 |
| TIMM44 | 100.0% | 100.0% | 100.0% | 99.8% | |
| TIMM50 | 100.0% | 100.0% | 100.0% | 99.8% | 3-methylglutaconic aciduria, type IX, 617698 |
| TIMM8A | 100.0% | 99.5% | 97.2% | 69.2% | Mohr-Tranebjaerg syndrome, 304700 |
| TIMMDC1 | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial complex I deficiency, nuclear type 31, 618251 |
| TK2 | 100.0% | 100.0% | 100.0% | 99.5% | Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 |
| TMEM126A | 100.0% | 100.0% | 100.0% | 99.3% | Optic atrophy 7, 612989 |
| TMEM126B | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial complex I deficiency, nuclear type 29, 618250 |

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| TMEM186 | 100.0% | 100.0% | 100.0% | 99.9% | |
| TMEM63C | 100.0% | 100.0% | 100.0% | 99.5% | Spastic paraplegia 87, autosomal recessive, 619966 |
| TMEM65 | 100.0% | 98.6% | 100.0% | 98.9% | |
| TMEM70 | 100.0% | 100.0% | 100.0% | 98.8% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 |
| TMX2 | 100.0% | 100.0% | 100.0% | 99.7% | Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 |
| TOMM40L | 100.0% | 100.0% | 100.0% | 99.7% | |
| TOMM70 | 100.0% | 100.0% | 100.0% | 99.9% | |
| TOP3A | 100.0% | 100.0% | 100.0% | 99.5% | ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098;Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 |
| TPK1 | 100.0% | 100.0% | 100.0% | 99.4% | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 |
| TRAPPC2L | 100.0% | 100.0% | 100.0% | 100.0% | Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 |

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| TRIT1 | 100.0% | 100.0% | 100.0% | 99.7% | Combined oxidative phosphorylation deficiency 35, 617873 |
| TRMT10C | 100.0% | 100.0% | 100.0% | 98.7% | Combined oxidative phosphorylation deficiency 30, 616974 |
| TRMT5 | 100.0% | 100.0% | 100.0% | 99.6% | Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539 |
| TRMU | 100.0% | 100.0% | 100.0% | 99.4% | {Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070 |
| TRNT1 | 100.0% | 100.0% | 100.0% | 99.3% | Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959 |
| TSFM | 94.3% | 94.3% | 100.0% | 99.8% | Combined oxidative phosphorylation deficiency 3, 610505 |
| TTC19 | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex III deficiency, nuclear type 2, 615157 |
| TUFM | 100.0% | 100.0% | 100.0% | 99.7% | Combined oxidative phosphorylation deficiency 4, 610678 |

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| TWNK | 100.0% | 100.0% | 100.0% | 99.7% | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138 |
| TXN2 | 100.0% | 100.0% | 100.0% | 99.7% | ?Combined oxidative phosphorylation deficiency 29, 616811 |
| TYMP | 100.0% | 100.0% | 100.0% | 100.0% | Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 |
| UCHL1 | 100.0% | 100.0% | 100.0% | 99.0% | {?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491 |
| UFM1 | 100.0% | 100.0% | 100.0% | 99.8% | Leukodystrophy, hypomyelinating, 14, 617899 |
| UQCC1 | 100.0% | 100.0% | 100.0% | 98.4% | |
| UQCC2 | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex III deficiency, nuclear type 7, 615824 |

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| UQCC3 | 100.0% | 100.0% | 100.0% | 99.4% | ?Mitochondrial complex III deficiency, nuclear type 9, 616111 |
| UQCR10 | 100.0% | 100.0% | 100.0% | 99.1% | |
| UQCR11 | 100.0% | 100.0% | 100.0% | 100.0% | |
| UQCRB | 100.0% | 100.0% | 100.0% | 99.7% | Mitochondrial complex III deficiency, nuclear type 3, 615158 |
| UQCRC1 | 100.0% | 100.0% | 100.0% | 99.9% | Parkinsonism with polyneuropathy, 619279 |
| UQCRC2 | 100.0% | 100.0% | 100.0% | 99.4% | Mitochondrial complex III deficiency, nuclear type 5, 615160 |
| UQCRFS1 | 100.0% | 100.0% | 100.0% | 100.0% | Mitochondrial complex III deficiency, nuclear type 10, 618775 |
| UQCRH | 100.0% | 100.0% | 100.0% | 99.1% | ?Mitochondrial complex III deficiency, nuclear type 11, 620137 |
| UQCRQ | 100.0% | 100.0% | 100.0% | 98.8% | Mitochondrial complex III deficiency, nuclear type 4, 615159 |
| VARS2 | 100.0% | 100.0% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 20, 615917 |
| VPS13D | 100.0% | 100.0% | 100.0% | 99.3% | Spinocerebellar ataxia, autosomal recessive 4, 607317 |

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| WARS2 | 100.0% | 100.0% | 100.0% | 99.3% | Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 |
| WDR45 | 100.0% | 100.0% | 99.7% | 84.2% | Neurodegeneration with brain iron accumulation 5, 300894 |
| XPNPEP3 | 100.0% | 100.0% | 100.0% | 99.8% | Nephronophthisis-like nephropathy 1, 613159 |
| YARS2 | 100.0% | 100.0% | 100.0% | 99.6% | Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 |
| YME1L1 | 100.0% | 100.0% | 100.0% | 99.0% | ?Optic atrophy 11, 617302 |

*Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.*
*TWIST X2 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry.*
*TWIST X2 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry.*
*srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.*
*srWGS GRCh38 Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.*
*non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*
*OMIM release used for OMIM disease identifiers and descriptions : March 17th, 2023.*
*This list is accurate for panel version DG 3.8.1*

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