

# MITOCHONDRIAL DISORDERS PANEL DG-4.0.0 (485 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%           | 99.4%            | Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096 |
| ABAT  | 100.0%                | 100.0%                | 100.0%           | 98.7%            | GABA-transaminase deficiency, 613163                                                                                   |
| ABCB7 | 99.8%                 | 99.3%                 | 98.3%            | 74.8%            | Anemia, sideroblastic, with ataxia, 301310                                                                             |
| ACAD9 | 100.0%                | 100.0%                | 100.0%           | 99.6%            | Mitochondrial complex I deficiency, nuclear type 20, 611126                                                            |
| ACO2  | 92.4%                 | 89.8%                 | 100.0%           | 99.3%            | Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559                                              |

|          |        |        |        |       |                                                                                                                                                                                                                                                 |
|----------|--------|--------|--------|-------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| ACTA1    | 100.0% | 100.0% | 100.0% | 97.1% | 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% | 99.1% | Weill-Marchesani syndrome 1, recessive, 277600                                                                                                                                                                                                  |
| ADAR     | 100.0% | 100.0% | 100.0% | 98.2% | Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010                                                                                                                                                              |
| ADCK2    | 100.0% | 100.0% | 100.0% | 98.2% |                                                                                                                                                                                                                                                 |
| ADPRS    | 100.0% | 100.0% | 100.0% | 99.2% | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170                                                                                                                                                   |
| AFG2A    | 100.0% | 100.0% | 100.0% | 98.9% | Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577                                                                                                                                                        |
| AFG3L2   | 100.0% | 100.0% | 100.0% | 98.4% | Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246                                                                                                                                        |

|         |        |        |        |       |                                                                                                                                                                                                        |
|---------|--------|--------|--------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| AGK     | 91.7%  | 91.7%  | 100.0% | 98.9% | Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350                                                                                                                                      |
| AIFM1   | 100.0% | 99.8%  | 97.6%  | 67.9% | 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% | 98.5% |                                                                                                                                                                                                        |
| ALDH1B1 | 100.0% | 100.0% | 100.0% | 99.7% |                                                                                                                                                                                                        |
| ALKBH1  | 100.0% | 100.0% | 100.0% | 97.5% |                                                                                                                                                                                                        |
| ANO10   | 100.0% | 100.0% | 100.0% | 98.2% | Spinocerebellar ataxia, autosomal recessive 10, 613728                                                                                                                                                 |
| APOO    | 100.0% | 100.0% | 98.3%  | 71.2% |                                                                                                                                                                                                        |
| APTX    | 100.0% | 100.0% | 100.0% | 98.5% | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920                                                                                                                               |
| ARL2    | 100.0% | 100.0% | 100.0% | 98.5% | ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082                                                                                                                         |
| ARNT2   | 100.0% | 100.0% | 100.0% | 98.7% | ?Webb-Dattani syndrome, 615926                                                                                                                                                                         |
| ATAD1   | 100.0% | 99.7%  | 100.0% | 97.4% | Hyperekplexia 4, 618011                                                                                                                                                                                |

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| ATAD3A  | 100.0% | 100.0% | 99.9%  | 96.7% | Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810                                                                                                     |
| ATAD3B  | 100.0% | 100.0% | 99.8%  | 95.0% |                                                                                                                                                                                                                                        |
| ATP13A2 | 100.0% | 100.0% | 100.0% | 99.3% | 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% | 99.2% | ?Hypermetabolism due to uncoupled mitochondrial oxidative phosphorylation 2, 620085                                                                                                                                                    |
| ATP5F1C | 100.0% | 100.0% | 100.0% | 97.8% |                                                                                                                                                                                                                                        |
| ATP5F1D | 100.0% | 100.0% | 100.0% | 97.9% | Mitochondrial complex V (ATP synthase) deficiency, 618120                                                                                                                                                                              |

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|---------|--------|--------|--------|-------|----------------------------------------------------------------------------|
| ATP5F1E | 100.0% | 100.0% | 100.0% | 96.8% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053  |
| ATP5IF1 | 100.0% | 100.0% | 100.0% | 98.6% |                                                                            |
| ATP5MC1 | 100.0% | 100.0% | 100.0% | 99.7% |                                                                            |
| ATP5MC2 | 100.0% | 100.0% | 100.0% | 98.2% |                                                                            |
| ATP5MC3 | 100.0% | 100.0% | 100.0% | 99.3% | Dystonia, early-onset, and/or spastic paraplegia, 619681                   |
| ATP5ME  | 100.0% | 100.0% | 100.0% | 98.7% |                                                                            |
| ATP5MF  | 100.0% | 100.0% | 100.0% | 99.5% |                                                                            |
| ATP5MG  | 95.4%  | 95.4%  | 100.0% | 97.4% |                                                                            |
| ATP5MGL | 100.0% | 100.0% | 100.0% | 99.8% |                                                                            |
| ATP5MK  | 100.0% | 100.0% | 100.0% | 97.2% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683  |
| ATP5PB  | 100.0% | 100.0% | 100.0% | 99.6% |                                                                            |
| ATP5PD  | 100.0% | 100.0% | 100.0% | 98.4% |                                                                            |
| ATP5PF  | 100.0% | 100.0% | 100.0% | 97.4% |                                                                            |
| ATP5PO  | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359  |
| ATPAF1  | 100.0% | 100.0% | 99.9%  | 92.8% |                                                                            |
| ATPAF2  | 100.0% | 100.0% | 100.0% | 99.1% | ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 |

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| BCAP31   | 99.1%  | 92.8%  | 98.0%  | 69.1%  | Deafness, dystonia, and cerebral hypomyelination, 300475                                                         |
| BCS1L    | 100.0% | 100.0% | 100.0% | 99.2%  | GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000 |
| BOLA1    | 100.0% | 100.0% | 100.0% | 100.0% |                                                                                                                  |
| BOLA2    | 100.0% | 100.0% | 100.0% | 99.3%  |                                                                                                                  |
| BOLA3    | 100.0% | 100.0% | 100.0% | 97.5%  | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299                                      |
| BTD      | 94.2%  | 94.2%  | 100.0% | 99.5%  | Biotinidase deficiency, 253260                                                                                   |
| C19orf12 | 100.0% | 99.8%  | 100.0% | 98.4%  | Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043     |
| C1QBP    | 100.0% | 100.0% | 100.0% | 98.9%  | Combined oxidative phosphorylation deficiency 33, 617713                                                         |
| C2orf69  | 100.0% | 100.0% | 99.9%  | 96.5%  | Combined oxidative phosphorylation deficiency 53, 619423                                                         |
| CA5A     | 100.0% | 100.0% | 100.0% | 98.0%  | Hyperammonemia due to carbonic anhydrase VA deficiency, 615751                                                   |

|         |        |        |        |       |                                                                                                                                                                                   |
|---------|--------|--------|--------|-------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| CARS2   | 100.0% | 100.0% | 100.0% | 99.0% | Combined oxidative phosphorylation deficiency 27, 616672                                                                                                                          |
| CDKL5   | 95.7%  | 95.3%  | 97.4%  | 68.8% | Developmental and epileptic encephalopathy 2, 300672                                                                                                                              |
| CEP89   | 100.0% | 100.0% | 100.0% | 96.9% |                                                                                                                                                                                   |
| CFAP58  | 100.0% | 100.0% | 100.0% | 97.2% | Spermatogenic failure 49, 619144                                                                                                                                                  |
| CHCHD10 | 100.0% | 100.0% | 100.0% | 96.9% | ?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% | 99.6% | Parkinson disease 22, autosomal dominant, 616710                                                                                                                                  |
| CHKB    | 100.0% | 100.0% | 100.0% | 98.7% | Muscular dystrophy, congenital, megaconial type, 602541                                                                                                                           |
| CISD2   | 100.0% | 100.0% | 100.0% | 98.0% | Wolfram syndrome 2, 604928                                                                                                                                                        |

|      |        |        |        |       |                                                                                                                                                                                                      |
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| CLPB | 100.0% | 100.0% | 99.9%  | 98.3% | 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% | 96.3% | Perrault syndrome 3, 614129                                                                                                                                                                          |
| COA1 | 100.0% | 100.0% | 100.0% | 98.6% |                                                                                                                                                                                                      |
| COA3 | 100.0% | 100.0% | 100.0% | 99.4% | ?Mitochondrial complex IV deficiency, nuclear type 14, 619058                                                                                                                                        |
| COA5 | 82.4%  | 82.4%  | 100.0% | 98.4% | ?Mitochondrial complex IV, deficiency, nuclear type 9, 616500                                                                                                                                        |
| COA6 | 100.0% | 100.0% | 100.0% | 96.5% | Mitochondrial complex IV deficiency, nuclear type 13, 616501                                                                                                                                         |
| COA7 | 100.0% | 100.0% | 100.0% | 99.2% | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387                                                                                                                        |
| COA8 | 100.0% | 99.9%  | 100.0% | 97.0% | Mitochondrial complex IV deficiency, nuclear type 17, 619061                                                                                                                                         |

|       |        |        |        |       |                                                                                                                  |
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| COASY | 100.0% | 100.0% | 100.0% | 99.1% | Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643             |
| COQ2  | 96.3%  | 96.3%  | 100.0% | 98.5% | {Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426                 |
| COQ4  | 100.0% | 100.0% | 100.0% | 99.6% | Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666                       |
| COQ5  | 100.0% | 100.0% | 100.0% | 97.4% | ?Coenzyme Q10 deficiency, primary, 9, 619028                                                                     |
| COQ6  | 100.0% | 100.0% | 99.9%  | 98.4% | Coenzyme Q10 deficiency, primary, 6, 614650                                                                      |
| COQ7  | 100.0% | 100.0% | 100.0% | 98.8% | Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 |
| COQ8A | 100.0% | 100.0% | 100.0% | 99.7% | Coenzyme Q10 deficiency, primary, 4, 612016                                                                      |
| COQ8B | 100.0% | 100.0% | 100.0% | 99.0% | Nephrotic syndrome, type 9, 615573                                                                               |
| COQ9  | 100.0% | 100.0% | 100.0% | 98.8% | Coenzyme Q10 deficiency, primary, 5, 614654                                                                      |

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|--------|--------|--------|--------|--------|-------------------------------------------------------------------------------------------------|
| COX10  | 100.0% | 100.0% | 100.0% | 99.4%  | Mitochondrial complex IV deficiency, nuclear type 3, 619046                                     |
| COX11  | 100.0% | 100.0% | 100.0% | 94.9%  | 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% | 98.5%  | Mitochondrial complex IV deficiency, nuclear type 6, 615119                                     |
| COX16  | 100.0% | 100.0% | 99.9%  | 98.3%  | Mitochondrial complex IV deficiency, nuclear type 22, 619355                                    |
| COX18  | 100.0% | 100.0% | 100.0% | 98.3%  |                                                                                                 |
| COX20  | 100.0% | 100.0% | 100.0% | 98.9%  | Mitochondrial complex IV deficiency, nuclear type 11, 619054                                    |
| COX4I1 | 100.0% | 100.0% | 100.0% | 99.2%  | 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% | 98.0%  | Mitochondrial complex IV deficiency, nuclear type 20, 619064                                    |
| COX5B  | 100.0% | 100.0% | 100.0% | 98.1%  |                                                                                                 |

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| COX6A1 | 100.0% | 100.0% | 100.0% | 97.6% | Charcot-Marie-Tooth disease, recessive intermediate D, 616039    |
| COX6A2 | 100.0% | 99.6%  | 100.0% | 95.1% | Mitochondrial complex IV deficiency, nuclear type 18, 619062     |
| COX6B1 | 100.0% | 100.0% | 100.0% | 99.3% | Mitochondrial complex IV deficiency, nuclear type 7, 619051      |
| COX6B2 | 100.0% | 100.0% | 100.0% | 92.8% |                                                                  |
| COX6C  | 100.0% | 100.0% | 100.0% | 98.6% |                                                                  |
| COX7A1 | 100.0% | 100.0% | 100.0% | 92.0% |                                                                  |
| COX7A2 | 100.0% | 100.0% | 100.0% | 95.5% |                                                                  |
| COX7B  | 100.0% | 99.9%  | 98.5%  | 76.8% | Linear skin defects with multiple congenital anomalies 2, 300887 |
| COX7B2 | 100.0% | 100.0% | 100.0% | 99.7% |                                                                  |
| COX7C  | 100.0% | 100.0% | 100.0% | 97.1% |                                                                  |
| COX8A  | 100.0% | 100.0% | 100.0% | 99.8% | ?Mitochondrial complex IV deficiency, nuclear type 15, 619059    |
| COX8C  | 100.0% | 100.0% | 100.0% | 99.5% |                                                                  |
| CP     | 100.0% | 100.0% | 100.0% | 98.7% | Aceruloplasminemia, 604290                                       |
| CRAT   | 100.0% | 100.0% | 100.0% | 99.3% | ?Neurodegeneration with brain iron accumulation 8, 617917        |

|        |        |        |        |       |                                                                                                                                 |
|--------|--------|--------|--------|-------|---------------------------------------------------------------------------------------------------------------------------------|
| CRLS1  | 100.0% | 100.0% | 100.0% | 95.1% | Combined oxidative phosphorylation deficiency 57, 620167                                                                        |
| CTBP1  | 100.0% | 99.5%  | 99.4%  | 97.5% | Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915                                                |
| CYC1   | 100.0% | 100.0% | 100.0% | 96.9% | Mitochondrial complex III deficiency, nuclear type 6, 615453                                                                    |
| CYCS   | 100.0% | 100.0% | 100.0% | 98.2% | Thrombocytopenia 4, 612004                                                                                                      |
| D2HGDH | 100.0% | 100.0% | 100.0% | 99.2% | D-2-hydroxyglutaric aciduria, 600721                                                                                            |
| DARS2  | 100.0% | 100.0% | 100.0% | 96.8% | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105                                   |
| DCAF17 | 100.0% | 100.0% | 99.9%  | 98.3% | Woodhouse-Sakati syndrome, 241080                                                                                               |
| DDHD1  | 100.0% | 100.0% | 100.0% | 97.6% | Spastic paraplegia 28, autosomal recessive, 609340                                                                              |
| DES    | 100.0% | 100.0% | 100.0% | 98.9% | Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419 |

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| DGUOK  | 100.0% | 100.0% | 100.0% | 98.6% | 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% | 98.0% | ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750                                                                                                          |
| DLAT   | 100.0% | 100.0% | 100.0% | 98.9% | Pyruvate dehydrogenase E2 deficiency, 245348                                                                                                                                                                           |
| DLD    | 100.0% | 100.0% | 100.0% | 98.7% | Dihydrolipoamide dehydrogenase deficiency, 246900                                                                                                                                                                      |
| DLST   | 100.0% | 100.0% | 100.0% | 98.9% | Pheochromocytoma/paraganglioma syndrome 7, 618475                                                                                                                                                                      |
| DMAC1  | 100.0% | 100.0% | 100.0% | 96.7% |                                                                                                                                                                                                                        |
| DMAC2  | 100.0% | 100.0% | 100.0% | 99.5% |                                                                                                                                                                                                                        |
| DMAC2L | 100.0% | 100.0% | 100.0% | 99.2% |                                                                                                                                                                                                                        |

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|---------|--------|--------|--------|-------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| DNA2    | 100.0% | 100.0% | 100.0% | 97.4% | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156;Rothmund-Thomson syndrome, type 4, 620819;Seckel syndrome 8, 615807                            |
| DNAJA3  | 100.0% | 100.0% | 100.0% | 99.1% |                                                                                                                                                                                                    |
| DNAJC19 | 100.0% | 100.0% | 100.0% | 98.3% | 3-methylglutaconic aciduria, type V, 610198                                                                                                                                                        |
| DNAJC3  | 100.0% | 100.0% | 99.9%  | 97.4% | Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192                                                                                                        |
| DNAJC30 | 100.0% | 100.0% | 100.0% | 99.7% | Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382                                                                                                                              |
| DNM1L   | 100.0% | 100.0% | 100.0% | 98.6% | Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388                                                                                       |
| DNM2    | 100.0% | 100.0% | 100.0% | 98.3% | 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 |

|        |        |        |        |       |                                                                                                                      |
|--------|--------|--------|--------|-------|----------------------------------------------------------------------------------------------------------------------|
| EARS2  | 100.0% | 100.0% | 100.0% | 98.7% | Combined oxidative phosphorylation deficiency 12, 614924                                                             |
| ECHS1  | 100.0% | 100.0% | 100.0% | 96.6% | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277                                                   |
| ECSIT  | 100.0% | 100.0% | 100.0% | 99.0% |                                                                                                                      |
| EHHADH | 100.0% | 100.0% | 100.0% | 99.2% | ?Fanconi renotubular syndrome 3, 615605                                                                              |
| ELAC2  | 100.0% | 100.0% | 100.0% | 99.3% | {Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440 |
| EMD    | 92.9%  | 90.4%  | 98.3%  | 71.2% | Emery-Dreifuss muscular dystrophy 1, X-linked, 310300                                                                |
| EPG5   | 100.0% | 100.0% | 100.0% | 98.4% | Vici syndrome, 242840                                                                                                |
| ERAL1  | 100.0% | 100.0% | 100.0% | 98.3% | Perrault syndrome 6, 617565                                                                                          |
| ETFDH  | 93.6%  | 92.0%  | 100.0% | 98.9% | Glutaric acidemia IIC, 231680                                                                                        |
| ETHE1  | 100.0% | 100.0% | 100.0% | 97.9% | Ethylmalonic encephalopathy, 602473                                                                                  |
| EXOSC8 | 100.0% | 100.0% | 100.0% | 97.0% | Pontocerebellar hypoplasia, type 1C, 616081                                                                          |
| FA2H   | 100.0% | 100.0% | 100.0% | 98.8% | Spastic paraplegia 35, autosomal recessive, 612319                                                                   |

|         |        |        |        |       |                                                                                                             |
|---------|--------|--------|--------|-------|-------------------------------------------------------------------------------------------------------------|
| FARS2   | 100.0% | 100.0% | 100.0% | 99.1% | Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046 |
| FARSB   | 100.0% | 100.0% | 100.0% | 98.9% | Rajab interstitial lung disease with brain calcifications 1, 613658                                         |
| FASTKD2 | 100.0% | 100.0% | 100.0% | 97.2% | Combined oxidative phosphorylation deficiency 44, 618855                                                    |
| FBXL4   | 100.0% | 100.0% | 100.0% | 99.2% | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471                                   |
| FDX2    | 100.0% | 99.6%  | 100.0% | 98.7% | Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900             |
| FDXR    | 100.0% | 100.0% | 100.0% | 99.4% | Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717       |
| FH      | 100.0% | 100.0% | 100.0% | 98.5% | Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812                                    |
| FLAD1   | 100.0% | 100.0% | 100.0% | 99.4% | Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100                     |

|         |        |        |        |       |                                                                                                                                                                         |
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| FOXRED1 | 100.0% | 100.0% | 100.0% | 98.6% | Mitochondrial complex I deficiency, nuclear type 19, 618241                                                                                                             |
| FTL     | 100.0% | 100.0% | 100.0% | 96.5% | 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% | 95.9% | Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300                                                                                              |
| GARS1   | 98.9%  | 98.9%  | 100.0% | 98.6% | 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% | 99.1% | ?Combined oxidative phosphorylation deficiency 41, 618838                                                                                                               |
| GATC    | 100.0% | 100.0% | 100.0% | 99.1% | Combined oxidative phosphorylation deficiency 42, 618839                                                                                                                |
| GATM    | 100.0% | 100.0% | 100.0% | 97.8% | Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600                                                                                  |

|       |        |        |        |       |                                                                                                                                                                                                                                       |
|-------|--------|--------|--------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| GBF1  | 100.0% | 100.0% | 100.0% | 99.0% | Charcot-Marie-Tooth disease, axonal, type 2GG, 606483                                                                                                                                                                                 |
| GCSH  | 100.0% | 100.0% | 100.0% | 98.1% | Multiple mitochondrial dysfunctions syndrome 7, 620423                                                                                                                                                                                |
| GDAP1 | 86.7%  | 86.7%  | 98.0%  | 96.0% | 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% | 99.6%  | 91.8% | Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076                                                                                                                                         |
| GFM1  | 100.0% | 100.0% | 100.0% | 98.6% | Combined oxidative phosphorylation deficiency 1, 609060                                                                                                                                                                               |
| GFM2  | 100.0% | 100.0% | 100.0% | 98.7% | Combined oxidative phosphorylation deficiency 39, 618397                                                                                                                                                                              |
| GLRX5 | 100.0% | 100.0% | 100.0% | 97.1% | Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859                                                                                                                     |

|        |        |        |        |       |                                                                                                                                                                           |
|--------|--------|--------|--------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| GLUD1  | 100.0% | 100.0% | 100.0% | 94.8% | Hyperinsulinism-hyperammonemia syndrome, 606762                                                                                                                           |
| GMPR   | 100.0% | 100.0% | 100.0% | 99.3% |                                                                                                                                                                           |
| GOT2   | 100.0% | 100.0% | 100.0% | 99.2% | Developmental and epileptic encephalopathy 82, 618721                                                                                                                     |
| GPT2   | 100.0% | 100.0% | 100.0% | 98.3% | Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281                                                                                              |
| GTPBP2 | 100.0% | 100.0% | 100.0% | 98.3% | Jaberi-Elahi syndrome, 617988                                                                                                                                             |
| GTPBP3 | 100.0% | 100.0% | 100.0% | 98.3% | Combined oxidative phosphorylation deficiency 23, 616198                                                                                                                  |
| GUF1   | 100.0% | 100.0% | 99.9%  | 97.4% | ?Developmental and epileptic encephalopathy 40, 617065                                                                                                                    |
| HACE1  | 100.0% | 100.0% | 100.0% | 97.4% | Spastic paraplegia and psychomotor retardation with or without seizures, 616756                                                                                           |
| HADHA  | 100.0% | 100.0% | 100.0% | 98.8% | HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016 |

|          |        |        |        |       |                                                                                                                                                         |
|----------|--------|--------|--------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------|
| HADHB    | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial trifunctional protein deficiency 2, 620300                                                                                                |
| HARS2    | 100.0% | 100.0% | 100.0% | 98.9% | Perrault syndrome 2, 614926                                                                                                                             |
| HCCS     | 100.0% | 100.0% | 97.8%  | 69.8% | Linear skin defects with multiple congenital anomalies 1, 309801                                                                                        |
| HIBCH    | 100.0% | 100.0% | 100.0% | 98.2% | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620                                                                                                     |
| HLCS     | 100.0% | 100.0% | 99.9%  | 97.6% | Holocarboxylase synthetase deficiency, 253270                                                                                                           |
| HPDL     | 100.0% | 100.0% | 100.0% | 98.9% | Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027 |
| HSD17B10 | 100.0% | 99.8%  | 98.0%  | 70.3% | HSD10 mitochondrial disease, 300438                                                                                                                     |
| HSPA9    | 100.0% | 100.0% | 100.0% | 98.5% | Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170                                                                                             |
| HSPD1    | 99.6%  | 97.9%  | 100.0% | 98.7% | Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233                                                            |
| HTRA2    | 100.0% | 100.0% | 100.0% | 98.4% | {Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248                                                                           |

|       |        |        |        |       |                                                                                                                      |
|-------|--------|--------|--------|-------|----------------------------------------------------------------------------------------------------------------------|
| IARS2 | 100.0% | 100.0% | 100.0% | 98.0% | Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 |
| IBA57 | 100.0% | 100.0% | 100.0% | 99.2% | Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451           |
| IDH2  | 100.0% | 100.0% | 100.0% | 98.1% | D-2-hydroxyglutaric aciduria 2, 613657                                                                               |
| ISCA1 | 92.4%  | 92.4%  | 100.0% | 98.9% | Multiple mitochondrial dysfunctions syndrome 5, 617613                                                               |
| ISCA2 | 100.0% | 100.0% | 100.0% | 98.9% | Multiple mitochondrial dysfunctions syndrome 4, 616370                                                               |
| ISCU  | 100.0% | 100.0% | 100.0% | 99.2% | Myopathy with lactic acidosis, hereditary, 255125                                                                    |

|       |        |        |        |       |                                                                                                                                                                                                                                                                            |
|-------|--------|--------|--------|-------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| KARS1 | 100.0% | 100.0% | 100.0% | 98.4% | 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% | 99.5% | NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607                                                                                              |
| LACTB | 100.0% | 100.0% | 100.0% | 97.5% |                                                                                                                                                                                                                                                                            |
| LARS2 | 100.0% | 100.0% | 100.0% | 99.1% | Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021                                                                                                                                                                                     |
| LDHD  | 100.0% | 100.0% | 100.0% | 99.3% | D-lactic aciduria with susceptibility to gout, 245450                                                                                                                                                                                                                      |

|        |        |        |        |       |                                                                                                           |
|--------|--------|--------|--------|-------|-----------------------------------------------------------------------------------------------------------|
| LETM1  | 100.0% | 100.0% | 100.0% | 99.3% | Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 |
| LIAS   | 100.0% | 100.0% | 100.0% | 99.2% | Hyperglycinemia, lactic acidosis, and seizures, 614462                                                    |
| LIG3   | 100.0% | 100.0% | 100.0% | 99.1% | Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780                                              |
| LIPT1  | 100.0% | 100.0% | 100.0% | 96.6% | Lipoyltransferase 1 deficiency, 616299                                                                    |
| LIPT2  | 100.0% | 100.0% | 100.0% | 98.2% | Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668                     |
| LONP1  | 100.0% | 100.0% | 100.0% | 99.1% | CODAS syndrome, 600373                                                                                    |
| LRPPRC | 96.8%  | 96.5%  | 100.0% | 98.1% | Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111                            |
| LYRM4  | 68.0%  | 68.0%  | 100.0% | 98.8% | ?Combined oxidative phosphorylation deficiency 19, 615595                                                 |
| LYRM7  | 100.0% | 100.0% | 100.0% | 98.2% | Mitochondrial complex III deficiency, nuclear type 8, 615838                                              |

|       |        |        |        |       |                                                                                                                                                                                                                             |
|-------|--------|--------|--------|-------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| MAPT  | 95.4%  | 95.4%  | 100.0% | 98.0% | 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% | 99.7% | ?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390                                                                                                                     |
| MCAT  | 100.0% | 100.0% | 100.0% | 99.3% | Optic atrophy 15, 620583                                                                                                                                                                                                    |
| MCUR1 | 100.0% | 100.0% | 100.0% | 95.2% |                                                                                                                                                                                                                             |
| MDH1  | 100.0% | 100.0% | 100.0% | 99.2% | ?Developmental and epileptic encephalopathy 88, 618959                                                                                                                                                                      |
| MDH2  | 100.0% | 100.0% | 100.0% | 98.6% | Developmental and epileptic encephalopathy 51, 617339                                                                                                                                                                       |
| MECR  | 100.0% | 100.0% | 100.0% | 99.2% | Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282;Optic atrophy 16, 620629                                                                                                              |
| MFF   | 95.9%  | 95.9%  | 100.0% | 98.8% | Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086                                                                                                                                             |

|         |        |        |        |       |                                                                                                                                                                                                                                                  |
|---------|--------|--------|--------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| MFN2    | 100.0% | 100.0% | 100.0% | 98.7% | 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% | 96.3% | Mitochondrial DNA depletion syndrome 11, 615084                                                                                                                                                                                                  |
| MICOS13 | 100.0% | 100.0% | 100.0% | 99.4% | Combined oxidative phosphorylation deficiency 37, 618329                                                                                                                                                                                         |
| MICU1   | 100.0% | 99.9%  | 100.0% | 99.0% | Myopathy with extrapyramidal signs, 615673                                                                                                                                                                                                       |
| MICU2   | 100.0% | 100.0% | 99.9%  | 96.8% |                                                                                                                                                                                                                                                  |
| MIEF2   | 100.0% | 100.0% | 100.0% | 99.5% | ?Combined oxidative phosphorylation deficiency 49, 619024                                                                                                                                                                                        |
| MIPEP   | 100.0% | 100.0% | 100.0% | 98.7% | Combined oxidative phosphorylation deficiency 31, 617228                                                                                                                                                                                         |

|        |        |        |        |       |                                                                                                                                             |
|--------|--------|--------|--------|-------|---------------------------------------------------------------------------------------------------------------------------------------------|
| MORC2  | 100.0% | 100.0% | 100.0% | 98.5% | 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% | 98.4% | Mitochondrial pyruvate carrier deficiency, 614741                                                                                           |
| MPC2   | 100.0% | 100.0% | 100.0% | 96.0% |                                                                                                                                             |
| MPV17  | 100.0% | 100.0% | 100.0% | 99.4% | Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810                  |
| MRM2   | 97.0%  | 97.0%  | 100.0% | 98.3% | Mitochondrial DNA depletion syndrome 17, 618567                                                                                             |
| MRPL12 | 100.0% | 100.0% | 100.0% | 99.0% | ?Combined oxidative phosphorylation deficiency 45, 618951                                                                                   |
| MRPL24 | 100.0% | 100.0% | 100.0% | 99.3% |                                                                                                                                             |
| MRPL3  | 100.0% | 100.0% | 100.0% | 98.4% | Combined oxidative phosphorylation deficiency 9, 614582                                                                                     |
| MRPL39 | 100.0% | 100.0% | 100.0% | 98.2% | Combined oxidative phosphorylation deficiency 59, 620646                                                                                    |
| MRPL40 | 100.0% | 100.0% | 100.0% | 98.4% |                                                                                                                                             |
| MRPL42 | 100.0% | 100.0% | 100.0% | 98.7% |                                                                                                                                             |

|        |        |        |        |       |                                                                                      |
|--------|--------|--------|--------|-------|--------------------------------------------------------------------------------------|
| MRPL44 | 100.0% | 100.0% | 100.0% | 98.9% | Combined oxidative phosphorylation deficiency 16, 615395                             |
| MRPL50 | 100.0% | 100.0% | 100.0% | 99.2% |                                                                                      |
| MRPL57 | 100.0% | 100.0% | 100.0% | 99.6% |                                                                                      |
| MRPS14 | 100.0% | 100.0% | 100.0% | 99.8% | ?Combined oxidative phosphorylation deficiency 38, 618378                            |
| MRPS16 | 100.0% | 100.0% | 100.0% | 98.9% | Combined oxidative phosphorylation deficiency 2, 610498                              |
| MRPS2  | 100.0% | 100.0% | 100.0% | 99.0% | Combined oxidative phosphorylation deficiency 36, 617950                             |
| MRPS22 | 100.0% | 99.9%  | 100.0% | 98.2% | Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719 |
| MRPS23 | 100.0% | 100.0% | 100.0% | 99.7% | ?Combined oxidative phosphorylation deficiency 46, 618952                            |
| MRPS25 | 74.2%  | 74.2%  | 100.0% | 98.5% | ?Combined oxidative phosphorylation deficiency 50, 619025                            |
| MRPS28 | 85.4%  | 85.3%  | 99.9%  | 96.2% | ?Combined oxidative phosphorylation deficiency 47, 618958                            |
| MRPS34 | 100.0% | 100.0% | 100.0% | 99.4% | Combined oxidative phosphorylation deficiency 32, 617664                             |

|        |        |        |        |       |                                                                                                                      |
|--------|--------|--------|--------|-------|----------------------------------------------------------------------------------------------------------------------|
| MRPS36 | 100.0% | 100.0% | 100.0% | 96.9% |                                                                                                                      |
| MRPS7  | 100.0% | 100.0% | 100.0% | 99.2% | ?Combined oxidative phosphorylation deficiency 34, 617872                                                            |
| MRRF   | 100.0% | 100.0% | 100.0% | 99.0% |                                                                                                                      |
| MSTO1  | 100.0% | 100.0% | 100.0% | 98.6% | Myopathy, mitochondrial, and ataxia, 617675                                                                          |
| MTFMT  | 100.0% | 100.0% | 100.0% | 97.8% | Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248 |
| MTO1   | 93.7%  | 91.1%  | 100.0% | 98.4% | Combined oxidative phosphorylation deficiency 10, 614702                                                             |
| MTPAP  | 100.0% | 100.0% | 100.0% | 98.0% | ?Spastic ataxia 4, autosomal recessive, 613672                                                                       |
| MTRFR  | 100.0% | 99.7%  | 99.7%  | 98.1% | Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559           |
| MTX2   | 100.0% | 99.9%  | 100.0% | 97.8% | Mandibuloacral dysplasia progeroid syndrome, 619127                                                                  |
| NADK2  | 100.0% | 100.0% | 100.0% | 95.8% | 2,4-dienoyl-CoA reductase deficiency, 616034                                                                         |

|         |        |        |        |       |                                                                                                       |
|---------|--------|--------|--------|-------|-------------------------------------------------------------------------------------------------------|
| NARS2   | 92.3%  | 92.3%  | 100.0% | 98.5% | Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434    |
| NAXD    | 96.8%  | 92.4%  | 100.0% | 99.2% | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321      |
| NAXE    | 95.8%  | 91.2%  | 100.0% | 98.5% | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186         |
| NDUFA1  | 100.0% | 100.0% | 95.8%  | 64.4% | Mitochondrial complex I deficiency, nuclear type 12, 301020                                           |
| NDUFA10 | 83.4%  | 81.0%  | 100.0% | 98.6% | Mitochondrial complex I deficiency, nuclear type 22, 618243                                           |
| NDUFA11 | 100.0% | 98.8%  | 100.0% | 96.7% | Mitochondrial complex I deficiency, nuclear type 14, 618236                                           |
| NDUFA12 | 79.4%  | 79.4%  | 100.0% | 98.0% | Mitochondrial complex I deficiency, nuclear type 23, 618244                                           |
| NDUFA13 | 100.0% | 100.0% | 100.0% | 99.1% | {Thyroid carcinoma, Hurthle cell}, 607464;Mitochondrial complex I deficiency, nuclear type 28, 618249 |

|         |        |        |        |       |                                                               |
|---------|--------|--------|--------|-------|---------------------------------------------------------------|
| NDUFA2  | 100.0% | 100.0% | 100.0% | 99.3% | Mitochondrial complex I deficiency, nuclear type 13, 618235   |
| NDUFA3  | 91.4%  | 86.8%  | 100.0% | 98.9% |                                                               |
| NDUFA4  | 100.0% | 100.0% | 100.0% | 95.7% | ?Mitochondrial complex IV deficiency, nuclear type 21, 619065 |
| NDUFA5  | 75.0%  | 75.0%  | 100.0% | 98.1% |                                                               |
| NDUFA6  | 100.0% | 100.0% | 100.0% | 98.5% | Mitochondrial complex I deficiency, nuclear type 33, 618253   |
| NDUFA7  | 100.0% | 100.0% | 100.0% | 98.5% |                                                               |
| NDUFA8  | 100.0% | 100.0% | 100.0% | 98.8% | Mitochondrial complex I deficiency, nuclear type 37, 619272   |
| NDUFA9  | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial complex I deficiency, nuclear type 26, 618247   |
| NDUFAB1 | 100.0% | 100.0% | 100.0% | 97.5% |                                                               |
| NDUFAF1 | 100.0% | 100.0% | 100.0% | 98.3% | Mitochondrial complex I deficiency, nuclear type 11, 618234   |
| NDUFAF2 | 67.4%  | 67.4%  | 100.0% | 97.3% | Mitochondrial complex I deficiency, nuclear type 10, 618233   |
| NDUFAF3 | 100.0% | 100.0% | 100.0% | 95.9% | Mitochondrial complex I deficiency, nuclear type 18, 618240   |

|         |        |        |        |       |                                                                                                                               |
|---------|--------|--------|--------|-------|-------------------------------------------------------------------------------------------------------------------------------|
| NDUFAF4 | 100.0% | 100.0% | 100.0% | 95.3% | Mitochondrial complex I deficiency, nuclear type 15, 618237                                                                   |
| NDUFAF5 | 100.0% | 100.0% | 99.9%  | 96.1% | Mitochondrial complex I deficiency, nuclear type 16, 618238                                                                   |
| NDUFAF6 | 100.0% | 100.0% | 100.0% | 96.3% | Mitochondrial complex I deficiency, nuclear type 17, 618239;Fanconi renotubular syndrome 5, 618913                            |
| NDUFAF7 | 100.0% | 100.0% | 100.0% | 97.5% |                                                                                                                               |
| NDUFAF8 | 100.0% | 100.0% | 100.0% | 99.3% | Mitochondrial complex I deficiency, nuclear type 34, 618776                                                                   |
| NDUFB1  | 100.0% | 100.0% | 99.9%  | 93.2% |                                                                                                                               |
| NDUFB10 | 100.0% | 100.0% | 100.0% | 95.2% | ?Mitochondrial complex I deficiency, nuclear type 35, 619003                                                                  |
| NDUFB11 | 99.7%  | 97.9%  | 88.1%  | 61.0% | Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021 |
| NDUFB2  | 100.0% | 100.0% | 100.0% | 99.5% |                                                                                                                               |
| NDUFB3  | 100.0% | 100.0% | 100.0% | 99.9% | Mitochondrial complex I deficiency, nuclear type 25, 618246                                                                   |
| NDUFB4  | 100.0% | 100.0% | 100.0% | 99.1% |                                                                                                                               |
| NDUFB5  | 100.0% | 100.0% | 100.0% | 98.1% |                                                                                                                               |

|        |        |        |        |       |                                                                                                                                   |
|--------|--------|--------|--------|-------|-----------------------------------------------------------------------------------------------------------------------------------|
| NDUFB6 | 100.0% | 100.0% | 100.0% | 98.8% |                                                                                                                                   |
| NDUFB7 | 100.0% | 100.0% | 99.8%  | 96.4% | ?Mitochondrial complex I deficiency, nuclear type 39, 620135                                                                      |
| NDUFB8 | 100.0% | 100.0% | 100.0% | 97.7% | Mitochondrial complex I deficiency, nuclear type 32, 618252                                                                       |
| NDUFB9 | 100.0% | 100.0% | 100.0% | 98.9% | ?Mitochondrial complex I deficiency, nuclear type 24, 618245                                                                      |
| NDUFC1 | 100.0% | 100.0% | 100.0% | 97.2% |                                                                                                                                   |
| NDUFC2 | 100.0% | 100.0% | 100.0% | 97.7% | Mitochondrial complex I deficiency, nuclear type 36, 619170                                                                       |
| NDUFS1 | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex I deficiency, nuclear type 5, 618226                                                                        |
| NDUFS2 | 99.5%  | 96.5%  | 100.0% | 98.3% | ?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228 |
| NDUFS3 | 96.5%  | 91.2%  | 100.0% | 99.2% | Mitochondrial complex I deficiency, nuclear type 8, 618230                                                                        |
| NDUFS4 | 100.0% | 99.9%  | 100.0% | 98.0% | Mitochondrial complex I deficiency, nuclear type 1, 252010                                                                        |
| NDUFS5 | 100.0% | 100.0% | 100.0% | 98.7% |                                                                                                                                   |

|        |        |        |        |       |                                                            |
|--------|--------|--------|--------|-------|------------------------------------------------------------|
| NDUFS6 | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex I deficiency, nuclear type 9, 618232 |
| NDUFS7 | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex I deficiency, nuclear type 3, 618224 |
| NDUFS8 | 100.0% | 100.0% | 100.0% | 99.6% | Mitochondrial complex I deficiency, nuclear type 2, 618222 |
| NDUFV1 | 100.0% | 100.0% | 99.9%  | 98.8% | Mitochondrial complex I deficiency, nuclear type 4, 618225 |
| NDUFV2 | 100.0% | 100.0% | 100.0% | 98.5% | Mitochondrial complex I deficiency, nuclear type 7, 618229 |
| NDUFV3 | 100.0% | 100.0% | 100.0% | 99.0% |                                                            |
| NFS1   | 89.8%  | 89.8%  | 100.0% | 99.2% | Combined oxidative phosphorylation deficiency 52, 619386   |
| NFU1   | 100.0% | 100.0% | 100.0% | 98.4% | Multiple mitochondrial dysfunctions syndrome 1, 605711     |
| NGLY1  | 100.0% | 100.0% | 100.0% | 98.7% | Congenital disorder of deglycosylation 1, 615273           |
| NME3   | 100.0% | 100.0% | 99.8%  | 95.9% |                                                            |
| NR2F1  | 100.0% | 99.9%  | 99.9%  | 91.8% | Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722       |

|       |        |        |        |       |                                                                                                                                                                                                                          |
|-------|--------|--------|--------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| NRROS | 100.0% | 100.0% | 100.0% | 99.6% | Seizures, early-onset, with neurodegeneration and brain calcification, 618875                                                                                                                                            |
| NSUN3 | 100.0% | 100.0% | 100.0% | 98.5% | Combined oxidative phosphorylation deficiency 48, 619012                                                                                                                                                                 |
| NUBPL | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex I deficiency, nuclear type 21, 618242                                                                                                                                                              |
| NUP54 | 100.0% | 100.0% | 100.0% | 98.7% | Dystonia 37, early-onset, with striatal lesions, 620427                                                                                                                                                                  |
| NUTF2 | 97.2%  | 96.9%  | 100.0% | 98.9% |                                                                                                                                                                                                                          |
| OGDH  | 100.0% | 100.0% | 100.0% | 99.3% | Oxoglutarate dehydrogenase deficiency, 203740                                                                                                                                                                            |
| OGDHL | 100.0% | 100.0% | 100.0% | 99.2% | Yoon-Bellen neurodevelopmental syndrome, 619701                                                                                                                                                                          |
| OPA1  | 100.0% | 100.0% | 100.0% | 98.5% | 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% | 98.6% | 3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300                                                                                                                                      |

|       |        |        |        |       |                                                                                                                                                                     |
|-------|--------|--------|--------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| OTX2  | 100.0% | 100.0% | 100.0% | 98.2% | 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% | 99.9%  | 97.9% |                                                                                                                                                                     |
| P4HTM | 100.0% | 100.0% | 100.0% | 95.5% | Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493                                                |
| PANK2 | 100.0% | 100.0% | 100.0% | 98.6% | Neurodegeneration with brain iron accumulation 1, 234200                                                                                                            |
| PARS2 | 100.0% | 100.0% | 100.0% | 99.5% | Developmental and epileptic encephalopathy 75, 618437                                                                                                               |
| PC    | 100.0% | 100.0% | 100.0% | 99.7% | Pyruvate carboxylase deficiency, 266150                                                                                                                             |
| PDE2A | 100.0% | 100.0% | 100.0% | 98.3% | Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150                                                                                  |
| PDHA1 | 99.6%  | 96.5%  | 97.6%  | 72.4% | Pyruvate dehydrogenase E1-alpha deficiency, 312170                                                                                                                  |
| PDHB  | 100.0% | 100.0% | 100.0% | 98.8% | Pyruvate dehydrogenase E1-beta deficiency, 614111                                                                                                                   |

|        |        |        |        |       |                                                                                                                                                                                                |
|--------|--------|--------|--------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| PDHX   | 100.0% | 99.8%  | 99.9%  | 98.1% | Lacticacidemia due to PDX1 deficiency, 245349                                                                                                                                                  |
| PDK1   | 100.0% | 100.0% | 100.0% | 97.4% |                                                                                                                                                                                                |
| PDK2   | 100.0% | 100.0% | 100.0% | 98.8% |                                                                                                                                                                                                |
| PDK3   | 100.0% | 100.0% | 98.2%  | 73.8% | ?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905                                                                                                                                     |
| PDK4   | 100.0% | 100.0% | 100.0% | 98.2% |                                                                                                                                                                                                |
| PDP1   | 100.0% | 100.0% | 100.0% | 99.4% | Pyruvate dehydrogenase phosphatase deficiency, 608782                                                                                                                                          |
| PDSS1  | 100.0% | 100.0% | 100.0% | 97.3% | Coenzyme Q10 deficiency, primary, 2, 614651                                                                                                                                                    |
| PDSS2  | 100.0% | 100.0% | 100.0% | 98.5% | Coenzyme Q10 deficiency, primary, 3, 614652                                                                                                                                                    |
| PET100 | 100.0% | 100.0% | 100.0% | 99.3% | Mitochondrial complex IV deficiency, nuclear type 12, 619055                                                                                                                                   |
| PET117 | 100.0% | 100.0% | 100.0% | 93.6% | ?Mitochondrial complex IV deficiency, nuclear type 19, 619063                                                                                                                                  |
| PIGA   | 100.0% | 100.0% | 97.7%  | 73.6% | Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 |

|        |        |        |        |       |                                                                                                                                                                                    |
|--------|--------|--------|--------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| PISD   | 100.0% | 100.0% | 100.0% | 99.8% | Liberfarb syndrome, 618889                                                                                                                                                         |
| PITRM1 | 100.0% | 100.0% | 100.0% | 99.0% | Spinocerebellar ataxia, autosomal recessive 30, 619405                                                                                                                             |
| PLA2G6 | 100.0% | 99.9%  | 100.0% | 99.2% | 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% | 99.2% | Epilepsy, early-onset, 1, vitamin B6-dependent, 617290                                                                                                                             |
| PMPCA  | 96.0%  | 96.0%  | 100.0% | 99.0% | Spinocerebellar ataxia, autosomal recessive 2, 213200                                                                                                                              |
| PMPCB  | 91.4%  | 91.4%  | 100.0% | 98.2% | Multiple mitochondrial dysfunctions syndrome 6, 617954                                                                                                                             |
| PNPLA8 | 100.0% | 100.0% | 100.0% | 97.0% | ?Mitochondrial myopathy with lactic acidosis, 251950                                                                                                                               |
| PNPT1  | 100.0% | 100.0% | 100.0% | 98.3% | 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% | 99.4% | 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% | 97.3% | 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% | 98.7% | Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603                                                                                                                                                                                                                                   |

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| POLRMT | 100.0% | 100.0% | 100.0% | 99.6% | Combined oxidative phosphorylation deficiency 55, 619743                                                                                                                                               |
| PPA2   | 100.0% | 99.9%  | 100.0% | 96.7% | ?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222                                                                                                             |
| PPCS   | 100.0% | 100.0% | 100.0% | 98.6% | Cardiomyopathy, dilated, 2C, 618189                                                                                                                                                                    |
| PRDX3  | 100.0% | 100.0% | 100.0% | 98.6% | Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871                                                                            |
| PRKAA1 | 100.0% | 100.0% | 100.0% | 97.4% |                                                                                                                                                                                                        |
| PRORP  | 100.0% | 100.0% | 100.0% | 97.9% | Combined oxidative phosphorylation deficiency 54, 619737                                                                                                                                               |
| PRPS1  | 100.0% | 100.0% | 96.3%  | 69.8% | Arts syndrome, 301835;Phosphoribosylpyro phosphate 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% | 98.7% | Combined oxidative phosphorylation deficiency 51, 619057                                                                                                                                               |

|         |        |        |        |       |                                                                                                      |
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| PTPMT1  | 100.0% | 100.0% | 99.9%  | 93.5% |                                                                                                      |
| PTRH2   | 100.0% | 100.0% | 100.0% | 98.8% | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263                    |
| PUS1    | 100.0% | 100.0% | 100.0% | 98.5% | Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462                                        |
| PYCR1   | 100.0% | 100.0% | 100.0% | 99.8% | Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940 |
| PYCR2   | 100.0% | 100.0% | 100.0% | 98.6% | Leukodystrophy, hypomyelinating, 10, 616420                                                          |
| PYROXD1 | 100.0% | 100.0% | 100.0% | 97.3% | Myopathy, myofibrillar, 8, 617258                                                                    |
| PYROXD2 | 90.6%  | 87.8%  | 100.0% | 99.1% |                                                                                                      |
| QRSL1   | 100.0% | 100.0% | 100.0% | 98.4% | Combined oxidative phosphorylation deficiency 40, 618835                                             |
| RANBP2  | 100.0% | 100.0% | 100.0% | 97.4% | {Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033                             |
| RARS2   | 94.2%  | 93.1%  | 100.0% | 98.6% | Pontocerebellar hypoplasia, type 6, 611523                                                           |
| RMND1   | 85.6%  | 85.6%  | 100.0% | 97.6% | Combined oxidative phosphorylation deficiency 11, 614922                                             |

|         |        |        |        |       |                                                                                                                                                                                                                                                                                                                                                          |
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| RNASEH1 | 100.0% | 100.0% | 100.0% | 98.9% | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479                                                                                                                                                                                                                                                     |
| RNF213  | 100.0% | 100.0% | 100.0% | 99.2% | {Moyamoya disease 2, susceptibility to}, 607151                                                                                                                                                                                                                                                                                                          |
| RRM1    | 100.0% | 100.0% | 100.0% | 98.6% | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647                                                                                                                                                                                                                                                     |
| RRM2B   | 100.0% | 100.0% | 100.0% | 97.7% | 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% | 97.5% | Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732                                                                                                                                                                                                                                                          |

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| RYR1   | 100.0% | 99.9%  | 100.0% | 98.7% | 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   | 99.0%  | 99.0%  | 100.0% | 98.0% | Spastic ataxia, Charlevoix-Saguenay type, 270550                                                                                                                                                                                         |
| SAMHD1 | 100.0% | 100.0% | 100.0% | 98.1% | ?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952                                                                                                                                                                          |
| SARS2  | 100.0% | 100.0% | 100.0% | 98.6% | Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845                                                                                                                                                              |
| SATB2  | 100.0% | 99.7%  | 100.0% | 98.6% | Glass syndrome, 612313                                                                                                                                                                                                                   |
| SCO1   | 100.0% | 100.0% | 100.0% | 98.7% | Mitochondrial complex IV deficiency, nuclear type 4, 619048                                                                                                                                                                              |
| SCO2   | 100.0% | 100.0% | 100.0% | 99.5% | Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377                                                                                                                                                             |
| SCP2   | 100.0% | 100.0% | 100.0% | 97.9% | ?Leukoencephalopathy with dystonia and motor neuropathy, 613724                                                                                                                                                                          |

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| SDHA   | 100.0% | 100.0% | 100.0% | 99.7% | 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% | 98.7% | Mitochondrial complex II deficiency, nuclear type 2, 619166                                                                                                                                                            |
| SDHB   | 100.0% | 100.0% | 100.0% | 98.4% | 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% | 98.4% | 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% | 98.3% | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739                                                                                                                             |

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| SFXN4    | 100.0% | 100.0% | 100.0% | 96.9% | Combined oxidative phosphorylation deficiency 18, 615578                                                        |
| SHMT2    | 100.0% | 100.0% | 100.0% | 99.6% | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121                    |
| SIRT5    | 100.0% | 100.0% | 100.0% | 99.5% |                                                                                                                 |
| SLC19A2  | 100.0% | 100.0% | 100.0% | 99.5% | Thiamine-responsive megaloblastic anemia syndrome, 249270                                                       |
| SLC19A3  | 99.6%  | 98.4%  | 100.0% | 98.1% | Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483      |
| SLC25A1  | 100.0% | 100.0% | 100.0% | 93.2% | Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197 |
| SLC25A10 | 100.0% | 100.0% | 100.0% | 99.8% | ?Mitochondrial DNA depletion syndrome 19, 618972                                                                |
| SLC25A12 | 100.0% | 100.0% | 100.0% | 98.5% | Developmental and epileptic encephalopathy 39, 612949                                                           |
| SLC25A13 | 100.0% | 100.0% | 100.0% | 98.7% | Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471                       |

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| SLC25A19 | 100.0% | 100.0% | 100.0% | 98.7% | Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 |
| SLC25A21 | 100.0% | 100.0% | 100.0% | 98.5% | ?Mitochondrial DNA depletion syndrome 18, 618811                                                                      |
| SLC25A22 | 100.0% | 100.0% | 100.0% | 99.6% | Developmental and epileptic encephalopathy 3, 609304                                                                  |
| SLC25A24 | 99.5%  | 99.5%  | 99.6%  | 97.2% | Fontaine progeroid syndrome, 612289                                                                                   |
| SLC25A26 | 100.0% | 100.0% | 100.0% | 98.8% | Combined oxidative phosphorylation deficiency 28, 616794                                                              |
| SLC25A3  | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial phosphate carrier deficiency, 610773                                                                    |
| SLC25A32 | 100.0% | 100.0% | 100.0% | 98.9% | ?Exercise intolerance, riboflavin-responsive, 616839                                                                  |
| SLC25A36 | 100.0% | 100.0% | 100.0% | 97.2% | Hyperinsulinemic hypoglycemia, familial, 8, 620211                                                                    |
| SLC25A38 | 100.0% | 100.0% | 100.0% | 99.2% | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950                                                               |

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| SLC25A4  | 100.0% | 100.0% | 100.0% | 98.5% | 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% | 99.2% | Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416                                                                                                                                                  |
| SLC25A46 | 100.0% | 100.0% | 99.9%  | 98.1% | Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303                                                                                                                                                    |
| SLC39A8  | 99.9%  | 99.4%  | 100.0% | 97.8% | Congenital disorder of glycosylation, type II <sub>n</sub> , 616721                                                                                                                                                                                       |
| SLC52A2  | 100.0% | 100.0% | 100.0% | 99.9% | Brown-Vialetto-Van Laere syndrome 2, 614707                                                                                                                                                                                                               |
| SLC52A3  | 100.0% | 100.0% | 100.0% | 99.0% | ?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530                                                                                                                                                                                  |
| SLC8B1   | 100.0% | 100.0% | 100.0% | 99.6% |                                                                                                                                                                                                                                                           |

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|--------|--------|--------|--------|-------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| SLIRP  | 100.0% | 100.0% | 99.9%  | 95.1% |                                                                                                                                                                                                                                        |
| SMDT1  | 100.0% | 100.0% | 100.0% | 99.2% |                                                                                                                                                                                                                                        |
| SOD2   | 100.0% | 100.0% | 100.0% | 99.4% | {Microvascular complications of diabetes 6}, 612634                                                                                                                                                                                    |
| SPART  | 100.0% | 100.0% | 100.0% | 98.1% | Troyer syndrome, 275900                                                                                                                                                                                                                |
| SPG7   | 100.0% | 100.0% | 100.0% | 98.6% | Spastic paraplegia 7, autosomal recessive, 607259                                                                                                                                                                                      |
| SPTBN4 | 100.0% | 100.0% | 100.0% | 98.3% | Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519                                                                                                                                                           |
| SQOR   | 100.0% | 100.0% | 100.0% | 98.2% | Sulfide:quinone oxidoreductase deficiency, 619221                                                                                                                                                                                      |
| SQSTM1 | 100.0% | 100.0% | 100.0% | 99.3% | 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 |
| SSBP1  | 100.0% | 100.0% | 100.0% | 98.6% | Optic atrophy 13 with retinal and foveal abnormalities, 165510                                                                                                                                                                         |

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| STAC3   | 100.0% | 100.0% | 100.0% | 98.5% | Congenital myopathy 13, 255995                                                                             |
| STAT2   | 100.0% | 100.0% | 100.0% | 99.0% | Pseudo-TORCH syndrome 3, 618886;Immunodeficiency 44, 616636                                                |
| STXBP1  | 100.0% | 100.0% | 100.0% | 98.6% | Developmental and epileptic encephalopathy 4, 612164                                                       |
| SUCLA2  | 100.0% | 99.6%  | 100.0% | 98.8% | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 |
| SUCLG1  | 100.0% | 100.0% | 100.0% | 96.4% | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400       |
| SUCLG2  | 100.0% | 99.8%  | 100.0% | 97.2% |                                                                                                            |
| SUPV3L1 | 100.0% | 100.0% | 100.0% | 98.1% |                                                                                                            |
| SURF1   | 100.0% | 100.0% | 100.0% | 98.7% | Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110   |
| SZT2    | 100.0% | 100.0% | 100.0% | 99.3% | Developmental and epileptic encephalopathy 18, 615476                                                      |
| TACO1   | 100.0% | 100.0% | 100.0% | 98.4% | Mitochondrial complex IV deficiency, nuclear type 8, 619052                                                |
| TFAZZIN | 100.0% | 100.0% | 96.7%  | 66.1% | Barth syndrome, 302060                                                                                     |

|        |        |        |        |       |                                                                                                                         |
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| TAMM41 | 100.0% | 100.0% | 100.0% | 98.7% | 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% | 98.6% | Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575                         |
| TARS2  | 100.0% | 100.0% | 100.0% | 98.4% | Combined oxidative phosphorylation deficiency 21, 615918                                                                |
| TBCK   | 100.0% | 100.0% | 100.0% | 98.7% | Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900                                  |
| TDP2   | 100.0% | 100.0% | 100.0% | 97.8% | Spinocerebellar ataxia, autosomal recessive 23, 616949                                                                  |
| TEFM   | 100.0% | 100.0% | 100.0% | 98.6% | Combined oxidative phosphorylation deficiency 58, 620451                                                                |
| TFAM   | 100.0% | 100.0% | 100.0% | 98.1% | ?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156                                                  |
| TFB2M  | 100.0% | 100.0% | 100.0% | 97.4% |                                                                                                                         |

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| THG1L    | 100.0% | 100.0% | 100.0% | 98.3% | Spinocerebellar ataxia, autosomal recessive 28, 618800                                                                                                                |
| TIMM22   | 100.0% | 100.0% | 100.0% | 99.1% | ?Combined oxidative phosphorylation deficiency 43, 618851                                                                                                             |
| TIMM44   | 100.0% | 100.0% | 100.0% | 98.2% |                                                                                                                                                                       |
| TIMM50   | 100.0% | 100.0% | 100.0% | 99.5% | 3-methylglutaconic aciduria, type IX, 617698                                                                                                                          |
| TIMM8A   | 100.0% | 99.5%  | 97.6%  | 65.5% | Mohr-Tranebjaerg syndrome, 304700                                                                                                                                     |
| TIMMDC1  | 100.0% | 100.0% | 100.0% | 97.8% | Mitochondrial complex I deficiency, nuclear type 31, 618251                                                                                                           |
| TK2      | 100.0% | 100.0% | 100.0% | 98.5% | 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% | 97.6% | Optic atrophy 7, 612989                                                                                                                                               |
| TMEM126B | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial complex I deficiency, nuclear type 29, 618250                                                                                                           |
| TMEM186  | 100.0% | 100.0% | 100.0% | 99.9% |                                                                                                                                                                       |
| TMEM63C  | 100.0% | 100.0% | 100.0% | 98.9% | Spastic paraplegia 87, autosomal recessive, 619966                                                                                                                    |

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| TMEM65   | 100.0% | 98.6%  | 99.9%  | 91.7% |                                                                                                                                                                                          |
| TMEM70   | 100.0% | 100.0% | 100.0% | 97.4% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052                                                                                                                |
| TMX2     | 100.0% | 100.0% | 100.0% | 99.1% | Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730                                                                                            |
| TOMM40L  | 100.0% | 100.0% | 100.0% | 98.6% |                                                                                                                                                                                          |
| TOMM70   | 100.0% | 100.0% | 100.0% | 99.1% |                                                                                                                                                                                          |
| TOP3A    | 100.0% | 100.0% | 100.0% | 98.8% | Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 |
| TPK1     | 100.0% | 100.0% | 100.0% | 98.0% | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458                                                                                                        |
| TRAPPC2L | 100.0% | 100.0% | 100.0% | 99.8% | Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331                                                                                                           |
| TRIT1    | 100.0% | 100.0% | 100.0% | 98.4% | Combined oxidative phosphorylation deficiency 35, 617873                                                                                                                                 |

|         |        |        |        |       |                                                                                                                                                                |
|---------|--------|--------|--------|-------|----------------------------------------------------------------------------------------------------------------------------------------------------------------|
| TRMT10C | 100.0% | 100.0% | 100.0% | 98.4% | Combined oxidative phosphorylation deficiency 30, 616974                                                                                                       |
| TRMT5   | 100.0% | 100.0% | 100.0% | 98.6% | Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539                                                          |
| TRMU    | 100.0% | 100.0% | 100.0% | 97.9% | {Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070                                                                      |
| TRNT1   | 92.0%  | 91.9%  | 100.0% | 98.7% | 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% | 98.5% | Combined oxidative phosphorylation deficiency 3, 610505                                                                                                        |
| TTC19   | 100.0% | 100.0% | 100.0% | 97.2% | Mitochondrial complex III deficiency, nuclear type 2, 615157                                                                                                   |
| TUFM    | 100.0% | 100.0% | 100.0% | 98.5% | Combined oxidative phosphorylation deficiency 4, 610678                                                                                                        |

|       |        |        |        |       |                                                                                                                                                                                                      |
|-------|--------|--------|--------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| TWNK  | 100.0% | 100.0% | 100.0% | 99.8% | 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% | 98.6% | Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041                                                                                                                                          |
| UCHL1 | 100.0% | 100.0% | 100.0% | 97.9% | {?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% | 99.0% | Leukodystrophy, hypomyelinating, 14, 617899                                                                                                                                                          |
| UQCC1 | 100.0% | 100.0% | 100.0% | 96.5% |                                                                                                                                                                                                      |
| UQCC2 | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial complex III deficiency, nuclear type 7, 615824                                                                                                                                         |

|         |        |        |        |       |                                                                |
|---------|--------|--------|--------|-------|----------------------------------------------------------------|
| UQCC3   | 100.0% | 100.0% | 100.0% | 97.8% | ?Mitochondrial complex III deficiency, nuclear type 9, 616111  |
| UQCR10  | 100.0% | 100.0% | 100.0% | 97.3% |                                                                |
| UQCR11  | 100.0% | 100.0% | 100.0% | 99.2% |                                                                |
| UQCRB   | 100.0% | 100.0% | 100.0% | 98.6% | Mitochondrial complex III deficiency, nuclear type 3, 615158   |
| UQCRC1  | 100.0% | 100.0% | 100.0% | 99.5% | Parkinsonism with polyneuropathy, 619279                       |
| UQCRC2  | 100.0% | 100.0% | 100.0% | 98.4% | Mitochondrial complex III deficiency, nuclear type 5, 615160   |
| UQCRFS1 | 100.0% | 100.0% | 100.0% | 99.0% | Mitochondrial complex III deficiency, nuclear type 10, 618775  |
| UQCRH   | 100.0% | 100.0% | 100.0% | 99.0% | ?Mitochondrial complex III deficiency, nuclear type 11, 620137 |
| UQCRQ   | 100.0% | 100.0% | 100.0% | 98.2% | Mitochondrial complex III deficiency, nuclear type 4, 615159   |
| VARS2   | 100.0% | 100.0% | 100.0% | 99.2% | Combined oxidative phosphorylation deficiency 20, 615917       |
| VPS13D  | 100.0% | 100.0% | 100.0% | 98.9% | Spinocerebellar ataxia, autosomal recessive 4, 607317          |

|         |        |        |        |       |                                                                                                                                                                            |
|---------|--------|--------|--------|-------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| WARS2   | 100.0% | 100.0% | 100.0% | 99.2% | 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% | 98.9%  | 76.4% | Neurodegeneration with brain iron accumulation 5, 300894                                                                                                                   |
| XPNPEP3 | 100.0% | 100.0% | 100.0% | 99.2% | Nephronophthisis-like nephropathy 1, 613159                                                                                                                                |
| YARS2   | 100.0% | 100.0% | 100.0% | 97.6% | Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561                                                                                                              |
| YME1L1  | 100.0% | 100.0% | 100.0% | 97.7% | ?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 4.0.0

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