

# POLYNEUROPATHIES PANEL<sup>1</sup> DG-5.1.0 (248 GENES)

| Gene   | Twist X2 covered 10x | Twist X2 covered 20x | srWGS covered 10x | srWGS covered 15x | srWGS covered 20x | Associated Phenotype description and OMIM disease ID                                                                                                                                                                           |
|--------|----------------------|----------------------|-------------------|-------------------|-------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| AAAS   | 100%                 | 100%                 | 100%              | 100%              | 99.7%             | Achalasia-addisonianism-alacrimia syndrome, 231550                                                                                                                                                                             |
| AARS1  | 100%                 | 100%                 | 100%              | 100%              | 99.4%             | Developmental and epileptic encephalopathy 29, 616339;Charcot-Marie-Tooth disease, axonal, type 2N, 613287;?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661;Trichothiodystrophy 8, nonphotosensitive, 619691 |
| ABCA1  | 100%                 | 100%                 | 100%              | 100%              | 99.8%             | Tangier disease, 205400;HDL deficiency, familial, 1, 604091                                                                                                                                                                    |
| ABCD1  | 89.3%                | 86.3%                | 98.7%             | 89.2%             | 70.9%             | Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100                                                                                                                                                              |
| ABHD12 | 100%                 | 100%                 | 100%              | 100%              | 99.6%             | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674                                                                                                                                               |
| ACOX1  | 97.9%                | 97.9%                | 100%              | 100%              | 99.7%             | Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470                                                                                                                                                      |
| ADPRS  | 100%                 | 100%                 | 100%              | 100%              | 99.4%             | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170                                                                                                                                  |

|          |       |       |       |       |       |                                                                                                                                                                                                         |
|----------|-------|-------|-------|-------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| AFG3L2   | 94.2% | 92.2% | 100%  | 100%  | 99.5% | Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246                                                                                                |
| AGRN     | 100%  | 100%  | 100%  | 99.9% | 98.9% | Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120                                                                                                                          |
| AGTPBP1  | 96.5% | 96.5% | 100%  | 100%  | 99.7% | Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276                                                                                                                                     |
| AHNAK2   | 97.8% | 97.7% | 95.6% | 94.1% | 91.1% |                                                                                                                                                                                                         |
| AIFM1    | 100%  | 100%  | 99.3% | 90.3% | 72.6% | Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepime taphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614 |
| AMACR    | 100%  | 100%  | 100%  | 99.8% | 99.4% | Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950                                                                                                      |
| APTX     | 100%  | 100%  | 100%  | 100%  | 99.7% | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920                                                                                                                                |
| ARHGAP19 | 94.5% | 94.1% | 100%  | 100%  | 99.6% | Charcot-Marie-Tooth disease, axonal, type 2KK, 621466                                                                                                                                                   |
| ARHGEF10 | 100%  | 100%  | 100%  | 100%  | 99.7% | ?Slowed nerve conduction velocity, AD, 608236                                                                                                                                                           |
| ARHGEF28 | 100%  | 100%  | 100%  | 100%  | 99.7% |                                                                                                                                                                                                         |

|          |       |       |       |       |       |                                                                                                                                    |
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| ARSA     | 100%  | 100%  | 100%  | 99.9% | 99%   | Metachromatic leukodystrophy, 250100                                                                                               |
| ATAD3A   | 100%  | 100%  | 100%  | 99.9% | 98.7% | Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 |
| ATL1     | 100%  | 100%  | 100%  | 100%  | 99.7% | Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708                                  |
| ATL3     | 100%  | 100%  | 100%  | 100%  | 99.7% | Neuropathy, hereditary sensory, type IF, 615632                                                                                    |
| ATP13A2  | 100%  | 100%  | 100%  | 100%  | 99.5% | Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693                                                    |
| ATP1A1   | 97.2% | 95.4% | 100%  | 100%  | 99.6% | Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036    |
| ATP7A    | 95%   | 94.8% | 99.4% | 92.2% | 74.7% | Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400                     |
| B2M      | 100%  | 100%  | 100%  | 100%  | 99.8% | Amyloidosis, hereditary systemic 6, 620659;Immunodeficiency 43, 241600                                                             |
| B4GALNT1 | 100%  | 100%  | 100%  | 100%  | 99.5% | Spastic paraplegia 26, autosomal recessive, 609195                                                                                 |

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| BAG3     | 100% | 100% | 100% | 100%  | 99.5% | ?Neuronopathy, distal hereditary motor, autosomal dominant 15, 621094;Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954;Charcot-Marie-Tooth disease, axonal, type 2JJ, 621095                                       |
| BICD2    | 100% | 100% | 100% | 100%  | 99.4% | Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290                                                                |
| BSCL2    | 100% | 100% | 100% | 100%  | 99.6% | Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924 |
| C19orf12 | 100% | 100% | 100% | 99.9% | 98.7% | Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043                                                                                                                             |
| CADM3    | 100% | 100% | 100% | 100%  | 99.4% | Charcot-Marie-Tooth disease, axonal, type 2FF, 619519                                                                                                                                                                                    |
| CCT5     | 100% | 100% | 100% | 100%  | 99.5% | ?Neuropathy, hereditary sensory, with spastic paraplegia, 256840                                                                                                                                                                         |

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| CD59    | 89.3% | 89.3% | 100% | 100%  | 100%  | Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300                                                                                           |
| CFAP276 | 100%  | 100%  | 100% | 100%  | 99.7% |                                                                                                                                                                                   |
| CHCHD10 | 100%  | 100%  | 100% | 100%  | 99.5% | ?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 |
| CNTNAP1 | 100%  | 100%  | 100% | 100%  | 99.1% | Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186                                                                                |
| COA3    | 100%  | 100%  | 100% | 100%  | 99.4% | ?Mitochondrial complex IV deficiency, nuclear type 14, 619058                                                                                                                     |
| COA7    | 100%  | 100%  | 100% | 100%  | 99.6% | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387                                                                                                     |
| COL6A5  | 100%  | 100%  | 100% | 100%  | 99.8% |                                                                                                                                                                                   |
| COMP    | 96.8% | 96.3% | 100% | 99.9% | 99.3% | Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400                                                                           |
| COX18   | 100%  | 100%  | 100% | 99.9% | 99.4% | ?Mitochondrial complex IV deficiency, nuclear type 25, 621487;Charcot-Marie-Tooth disease, axonal, type 2MM, 621488                                                               |
| COX20   | 100%  | 100%  | 100% | 100%  | 99.8% | Mitochondrial complex IV deficiency, nuclear type 11, 619054                                                                                                                      |

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| COX6A1  | 100%  | 100%  | 100% | 100%  | 99.5% | Charcot-Marie-Tooth disease, recessive intermediate D, 616039                                                                                                                |
| CRYAB   | 100%  | 100%  | 100% | 100%  | 99.7% | Myopathy, myofibrillar, 2B, infantile-onset, 613869;Myopathy, myofibrillar, 2A, adult-onset, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184 |
| CTDP1   | 90.8% | 90.5% | 100% | 100%  | 99.2% | Congenital cataracts, facial dysmorphism, and neuropathy, 604168                                                                                                             |
| CYP27A1 | 100%  | 100%  | 100% | 100%  | 99.5% | Cerebrotendinous xanthomatosis, 213700                                                                                                                                       |
| CYP2U1  | 100%  | 100%  | 100% | 99.9% | 98.8% | Spastic paraplegia 56, autosomal recessive, 615030                                                                                                                           |
| CYP7B1  | 100%  | 100%  | 100% | 100%  | 100%  | Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812                                                                         |
| DARS2   | 100%  | 100%  | 100% | 100%  | 99.8% | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105;Charcot-Marie-Tooth disease, axonal, type 2LL, 621485                          |
| DCAF8   | 96%   | 96%   | 100% | 100%  | 99.7% | ?Giant axonal neuropathy 2, autosomal dominant, 610100                                                                                                                       |
| DCTN1   | 100%  | 100%  | 100% | 100%  | 99%   | Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641                       |
| DCTN2   | 100%  | 100%  | 100% | 100%  | 99.5% |                                                                                                                                                                              |

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|--------|-------|-------|-------|-------|-------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| DGAT2  | 100%  | 100%  | 100%  | 100%  | 99.5% |                                                                                                                                                                                                    |
| DHTKD1 | 92.7% | 92.7% | 100%  | 100%  | 99.4% | ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750                                                                                      |
| DNAJB2 | 100%  | 100%  | 100%  | 99.9% | 99.5% | Neuronopathy, distal hereditary motor, autosomal recessive 5, 614881                                                                                                                               |
| DNAJB5 | 100%  | 100%  | 100%  | 100%  | 99.3% |                                                                                                                                                                                                    |
| DNAJC3 | 92.1% | 92.1% | 100%  | 100%  | 99.8% | Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192                                                                                                        |
| DNM1L  | 100%  | 100%  | 100%  | 100%  | 99.9% | Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388                                                                                       |
| DNM2   | 93.3% | 91.7% | 100%  | 99.9% | 99.4% | 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 |
| DNMT1  | 100%  | 99.3% | 100%  | 100%  | 99.4% | Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121                                                                            |
| DRP2   | 100%  | 100%  | 98.8% | 88.9% | 69.3% |                                                                                                                                                                                                    |

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| DST     | 100%  | 100%  | 100% | 100%  | 99.7% | Congenital myopathy 29 with contractures, 621510;Neuropathy, hereditary sensory and autonomic, type VI, 614653;Lethal congenital contracture syndrome 12, 621511;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425 |
| DYNC1H1 | 99.3% | 99.3% | 100% | 99.9% | 99.4% | Charcot-Marie-Tooth disease, axonal, type 2O, 614228;Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600;Cortical dysplasia, complex, with other brain malformations 13, 614563                                                                         |
| EGR2    | 100%  | 100%  | 100% | 100%  | 99.6% | Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1D, 607678;Hypomyelinating neuropathy, congenital, 1, 605253                                                                                                                                         |
| ELF2    | 100%  | 100%  | 100% | 100%  | 99.7% |                                                                                                                                                                                                                                                                        |
| ELOVL5  | 100%  | 100%  | 100% | 100%  | 99.6% | Spinocerebellar ataxia 38, 615957                                                                                                                                                                                                                                      |
| ELP1    | 100%  | 100%  | 100% | 100%  | 99.8% | {Medulloblastoma}, 155255;Dysautonomia, familial, 223900                                                                                                                                                                                                               |
| EMILIN1 | 100%  | 99.7% | 100% | 100%  | 99.3% | Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080;Arterial tortuosity-bone fragility syndrome, 620908                                                                                                                                               |

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|--------|-------|-------|------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| ERBB2  | 100%  | 100%  | 100% | 100%  | 99.4% | Gastric cancer, somatic, 613659;Adenocarcinoma of lung, somatic, 211980;Ovarian cancer, somatic, 167000;?Visceral neuropathy, familial, 2, autosomal recessive, 619465;Glioblastoma, somatic, 137800 |
| ERBB3  | 100%  | 100%  | 100% | 99.9% | 98.9% | ?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180                             |
| ETFDH  | 94.1% | 92.3% | 100% | 100%  | 99.9% | Glutaric acidemia IIC, 231680                                                                                                                                                                        |
| EXOSC8 | 100%  | 100%  | 100% | 100%  | 99.8% | Pontocerebellar hypoplasia, type 1C, 616081                                                                                                                                                          |
| EXOSC9 | 100%  | 100%  | 100% | 100%  | 99.9% | Pontocerebellar hypoplasia, type 1D, 618065                                                                                                                                                          |
| FAT3   | 100%  | 100%  | 100% | 100%  | 99.8% |                                                                                                                                                                                                      |
| FBLN5  | 92.8% | 92.8% | 100% | 99.9% | 99.4% | Cutis laxa, autosomal recessive, type IA, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;?Cutis laxa, autosomal dominant 2, 614434  |
| FBN2   | 99.2% | 99.2% | 100% | 100%  | 99.6% | Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050                                                                                                            |

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|--------|-------|-------|------|-------|-------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| FBXO38 | 100%  | 100%  | 100% | 100%  | 99.9% | Neuronopathy, distal hereditary motor, autosomal dominant 6, 615575                                                                                                     |
| FGD4   | 100%  | 100%  | 100% | 100%  | 99.8% | Charcot-Marie-Tooth disease, type 4H, 609311                                                                                                                            |
| FIG4   | 98.4% | 98.4% | 100% | 100%  | 99.9% | Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228  |
| FLVCR1 | 95%   | 94.6% | 100% | 100%  | 99.7% | Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060                                     |
| FXN    | 100%  | 100%  | 100% | 100%  | 98.9% | Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300                                                                                              |
| GALC   | 100%  | 100%  | 100% | 100%  | 99.6% | Krabbe disease, 245200                                                                                                                                                  |
| GAN    | 100%  | 100%  | 100% | 99.9% | 99.5% | Giant axonal neuropathy-1, 256850                                                                                                                                       |
| GARS1  | 98.9% | 98.9% | 100% | 100%  | 99.8% | Spinal muscular atrophy, infantile, James type, 619042;Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794;Charcot-Marie-Tooth disease, type 2D, 601472 |
| GBA2   | 100%  | 100%  | 100% | 99.9% | 99.4% | Spastic paraplegia 46, autosomal recessive, 614409                                                                                                                      |

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| GBE1  | 100%  | 100%  | 100%  | 100%  | 99.7% | Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570                                                                                                                                                     |
| GBF1  | 100%  | 100%  | 100%  | 100%  | 99.5% | Charcot-Marie-Tooth disease, axonal, type 2GG, 606483                                                                                                                                                                                 |
| GDAP1 | 86.2% | 86.2% | 98.3% | 98%   | 97.1% | 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 |
| GJB1  | 100%  | 100%  | 97.9% | 84.9% | 64.7% | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800                                                                                                                                                                          |
| GJB3  | 100%  | 100%  | 100%  | 100%  | 99.5% | Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratodermia variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644                                                           |
| GLA   | 91.4% | 91.4% | 99.3% | 91.7% | 72.9% | Fabry disease, cardiac variant, 301500;Fabry disease, 301500                                                                                                                                                                          |
| GLE1  | 100%  | 100%  | 100%  | 100%  | 99.7% | Lethal congenital contracture syndrome 1, 253310;Congenital arthrogryposis with anterior horn cell disease, 611890                                                                                                                    |
| GNB4  | 100%  | 100%  | 100%  | 100%  | 100%  | Charcot-Marie-Tooth disease, dominant intermediate F, 615185                                                                                                                                                                          |

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| GNE   | 100%  | 100%  | 100% | 100%  | 99.8% | Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820                                                                                                                                                          |
| GSN   | 92.1% | 90%   | 100% | 99.8% | 98.9% | Amyloidosis, Finnish type, 105120                                                                                                                                                                                                                      |
| HADHA | 96.1% | 95.3% | 100% | 100%  | 99.6% | HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016                                                                              |
| HADHB | 94.6% | 94.6% | 100% | 100%  | 99.7% | Mitochondrial trifunctional protein deficiency 2, 620300                                                                                                                                                                                               |
| HARS1 | 93.2% | 93.2% | 100% | 99.9% | 99.9% | Charcot-Marie-Tooth disease, axonal, type 2W, 616625;Usher syndrome type 3B, 614504                                                                                                                                                                    |
| HINT1 | 100%  | 100%  | 100% | 100%  | 99.8% | Neuromyotonia and axonal neuropathy, autosomal recessive, 137200                                                                                                                                                                                       |
| HK1   | 98.2% | 96.5% | 100% | 99.9% | 99.5% | Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelopmental disorder with visual defects and brain anomalies, 618547 |

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| HMBS    | 100% | 100% | 100% | 100% | 99.9% | Leukoencephalopathy, porphyria-related, 620711;Encephalopathy, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000 |
| HNRNPA1 | 100% | 100% | 100% | 100% | 99.7% | ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424;?Myopathy, distal, 3, 610099;Amyotrophic lateral sclerosis 20, 615426                    |
| HOXD10  | 100% | 100% | 100% | 100% | 98.6% | Vertical talus, congenital, 192950;Charcot-Marie-Tooth disease, foot deformity of, 192950                                                                                                  |
| HSD17B4 | 100% | 100% | 100% | 100% | 99.9% | D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400                                                                                                                      |
| HSPB1   | 100% | 100% | 100% | 100% | 99.1% | Charcot-Marie-Tooth disease, axonal, type 2F, 606595;Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634                                                                   |
| HSPB3   | 100% | 100% | 100% | 100% | 99.2% | ?Neuronopathy, distal hereditary motor, autosomal dominant 4, 613376                                                                                                                       |

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| HSPB8   | 100%  | 100%  | 100%  | 100%  | 99.7% | Neuronopathy, distal hereditary motor, autosomal dominant 2, 158590;Charcot-Marie-Tooth disease, axonal, type 2L, 608673;Myopathy, myofibrillar, 13, with rimmed vacuoles, 621078 |
| HYCC1   | 100%  | 100%  | 100%  | 100%  | 100%  | Leukodystrophy, hypomyelinating, 5, 610532                                                                                                                                        |
| IARS2   | 100%  | 99.7% | 100%  | 99.9% | 99.6% | Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007                                                              |
| IFRD1   | 100%  | 100%  | 100%  | 100%  | 99.6% |                                                                                                                                                                                   |
| IGHMBP2 | 100%  | 100%  | 100%  | 100%  | 99.4% | Charcot-Marie-Tooth disease, axonal, type 2S, 616155;Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320                                                         |
| INF2    | 100%  | 100%  | 99.9% | 99.5% | 98.1% | Glomerulosclerosis, focal segmental, 5, 613237;Charcot-Marie-Tooth disease, dominant intermediate E, 614455                                                                       |
| INSC    | 94.4% | 94.4% | 100%  | 100%  | 99.5% |                                                                                                                                                                                   |
| ITPR3   | 100%  | 100%  | 100%  | 100%  | 99.4% | Charcot-Marie-Tooth disease, demyelinating, type 1J, 620111;Immunodeficiency 133 with ectodermal dysplasia with or without peripheral neuropathy, 621254                          |

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| JAG1    | 100%  | 100%  | 100% | 100% | 99.6% | ?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500                                                                                       |
| KARS1   | 94.3% | 94.3% | 100% | 100% | 99.8% | 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 |
| KBTBD13 | 100%  | 100%  | 100% | 100% | 99.1% | Nemaline myopathy 6, autosomal dominant, 609273                                                                                                                                                                                                                            |
| KIF1A   | 100%  | 100%  | 100% | 100% | 99.3% | NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607                                                                                              |
| KIF1B   | 94.9% | 94.9% | 100% | 100% | 99.6% | {Neuroblastoma, susceptibility to, 1}, 256700;Charcot-Marie-Tooth disease, type 2A1, 118210                                                                                                                                                                                |

|       |      |      |      |       |       |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
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| KIF5A | 100% | 100% | 100% | 100%  | 99.5% | Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187                                                                                                                                                                                                                                                                                                                           |
| KLC2  | 100% | 100% | 100% | 100%  | 99.4% | Spastic paraplegia, optic atrophy, and neuropathy, 609541                                                                                                                                                                                                                                                                                                                                                                                                                           |
| LAMA2 | 100% | 100% | 100% | 100%  | 99.8% | Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855                                                                                                                                                                                                                                                                                                                                    |
| LITAF | 100% | 100% | 100% | 100%  | 99.3% | Charcot-Marie-Tooth disease, type 1C, 601098                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| LMNA  | 100% | 100% | 100% | 99.9% | 99.3% | Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112 |

|        |       |       |      |      |       |                                                                                                                                                                                                                                                  |
|--------|-------|-------|------|------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| LRSAM1 | 100%  | 100%  | 100% | 100% | 99.4% | Charcot-Marie-Tooth disease, axonal, type 2P, 614436                                                                                                                                                                                             |
| MAG    | 100%  | 100%  | 100% | 100% | 99.3% | Spastic paraplegia 75, autosomal recessive, 616680                                                                                                                                                                                               |
| MARS1  | 89.3% | 88.7% | 100% | 100% | 99.5% | Spastic paraplegia 70, autosomal recessive, 620323;Interstitial lung and liver disease, 615486;?Trichothiodystrophy 9, nonphotosensitive, 619692;Charcot-Marie-Tooth disease, axonal, type 2U, 616280                                            |
| MCM3AP | 100%  | 100%  | 100% | 100% | 99.5% | Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124                                                                                                                                            |
| MFN2   | 100%  | 100%  | 100% | 100% | 99.7% | 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 |
| MME    | 97.7% | 97.7% | 100% | 100% | 99.7% | ?Spinocerebellar ataxia 43, 617018;Charcot-Marie-Tooth disease, axonal, type 2T, 617017                                                                                                                                                          |
| MORC2  | 100%  | 100%  | 100% | 100% | 99.3% | Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090                                                                                                      |

|       |      |      |      |       |       |                                                                                                                                                                                                                                                                                                                    |
|-------|------|------|------|-------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| MPV17 | 100% | 100% | 100% | 100%  | 99.6% | Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810                                                                                                                                                                                         |
| MPZ   | 100% | 100% | 100% | 100%  | 98.9% | Charcot-Marie-Tooth disease, type 2I, 607677;Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1B, 118200;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, dominant intermediate D, 607791;Hypomyelinating neuropathy, congenital, 2, 618184;Charcot-Marie-Tooth disease, type 2J, 607736 |
| MTMR2 | 100% | 100% | 100% | 99.9% | 99.6% | Charcot-Marie-Tooth disease, type 4B1, 601382                                                                                                                                                                                                                                                                      |
| MTRFR | 100% | 100% | 100% | 100%  | 99.7% | Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559                                                                                                                                                                                                         |
| MYH14 | 100% | 100% | 100% | 99.9% | 98.8% | ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369;Deafness, autosomal dominant 4A, 600652                                                                                                                                                                                                     |
| MYO9B | 100% | 100% | 100% | 100%  | 99.2% |                                                                                                                                                                                                                                                                                                                    |
| MYOT  | 100% | 100% | 100% | 100%  | 99.8% | Myopathy, myofibrillar, 3, 609200                                                                                                                                                                                                                                                                                  |

|        |       |       |      |       |       |                                                                                                                                                                                                                                                      |
|--------|-------|-------|------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| NAGLU  | 100%  | 100%  | 100% | 100%  | 99.4% | ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920                                                                                                                                         |
| NARS1  | 100%  | 100%  | 100% | 100%  | 99.5% | Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092;Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091 |
| NDRG1  | 100%  | 100%  | 100% | 100%  | 99.7% | Charcot-Marie-Tooth disease, type 4D, 601455                                                                                                                                                                                                         |
| NDUFA9 | 87%   | 85.2% | 100% | 100%  | 99.9% | Mitochondrial complex I deficiency, nuclear type 26, 618247                                                                                                                                                                                          |
| NDUFS6 | 94.7% | 86.1% | 100% | 100%  | 99.7% | Mitochondrial complex I deficiency, nuclear type 9, 618232                                                                                                                                                                                           |
| NEFH   | 100%  | 100%  | 100% | 100%  | 99.6% | Charcot-Marie-Tooth disease, axonal, type 2CC, 616924;{?Amyotrophic lateral sclerosis, susceptibility to}, 105400                                                                                                                                    |
| NEFL   | 100%  | 100%  | 100% | 99.9% | 98.8% | Charcot-Marie-Tooth disease, type 1F, 607734;Charcot-Marie-Tooth disease, dominant intermediate G, 617882;Charcot-Marie-Tooth disease, type 2E, 607684                                                                                               |

|        |       |       |       |       |       |                                                                                                |
|--------|-------|-------|-------|-------|-------|------------------------------------------------------------------------------------------------|
| NEMF   | 100%  | 100%  | 100%  | 99.8% | 99.3% | Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099 |
| NFASC  | 100%  | 100%  | 100%  | 99.9% | 99.5% | Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356              |
| NGF    | 100%  | 100%  | 100%  | 100%  | 98.9% | Neuropathy, hereditary sensory and autonomic, type V, 608654                                   |
| NHERF1 | 85.8% | 85.8% | 100%  | 100%  | 99.2% | Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287                                      |
| NIPA1  | 100%  | 100%  | 99.8% | 99.2% | 97.4% | Spastic paraplegia 6, autosomal dominant, 600363                                               |
| NMNAT2 | 100%  | 100%  | 100%  | 100%  | 99.7% |                                                                                                |
| NRG1   | 100%  | 100%  | 100%  | 99.9% | 99.3% |                                                                                                |
| NTRK1  | 100%  | 100%  | 100%  | 99.9% | 99.3% | Insensitivity to pain, congenital, with anhidrosis, 256800                                     |
| PDHA1  | 91.4% | 89.5% | 99.3% | 91.4% | 72.9% | Pyruvate dehydrogenase E1-alpha deficiency, 312170                                             |
| PDK3   | 100%  | 100%  | 99.1% | 91%   | 73.3% | ?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905                                     |
| PDXK   | 91.6% | 88.3% | 100%  | 100%  | 99.3% | Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511                 |
| PDYN   | 100%  | 100%  | 100%  | 100%  | 99.9% | Spinocerebellar ataxia 23, 610245                                                              |

|        |       |       |      |       |       |                                                                                                                                                                                      |
|--------|-------|-------|------|-------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| PEX1   | 100%  | 100%  | 100% | 100%  | 99.9% | Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100                                                |
| PEX10  | 100%  | 100%  | 100% | 99.6% | 98.7% | Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871                                                                                      |
| PEX16  | 100%  | 100%  | 100% | 100%  | 99.6% | Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876                                                                                      |
| PEX7   | 97.9% | 97.9% | 100% | 100%  | 99.6% | Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879                                                                                       |
| PHYH   | 100%  | 100%  | 100% | 99.8% | 99.1% | Refsum disease, 266500                                                                                                                                                               |
| PIEZO2 | 100%  | 100%  | 100% | 100%  | 99.7% | Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700 |
| PIGB   | 100%  | 100%  | 100% | 100%  | 99.8% | Developmental and epileptic encephalopathy 80, 618580                                                                                                                                |
| PIGG   | 100%  | 100%  | 100% | 100%  | 99.5% | [Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917                                                    |

|         |       |       |      |       |       |                                                                                                                                                                                                                                                             |
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| PLA2G6  | 100%  | 100%  | 100% | 100%  | 99.4% | Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600                                                                                                       |
| PLD3    | 100%  | 100%  | 100% | 100%  | 99.3% | ?Spinocerebellar ataxia 46, 617770                                                                                                                                                                                                                          |
| PLEKHG5 | 100%  | 100%  | 100% | 99.9% | 99.3% | Neuronopathy, distal hereditary motor, autosomal recessive 4, 611067;Charcot-Marie-Tooth disease, recessive intermediate C, 615376                                                                                                                          |
| PMM2    | 94.6% | 94.6% | 100% | 100%  | 99.7% | Congenital disorder of glycosylation, type Ia, 212065                                                                                                                                                                                                       |
| PMP2    | 100%  | 100%  | 100% | 100%  | 99.7% | Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279                                                                                                                                                                                                 |
| PMP22   | 100%  | 100%  | 100% | 100%  | 99.7% | Charcot-Marie-Tooth disease, type 1A, 118220;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, type 1E, 118300;?Neuropathy, inflammatory demyelinating, 139393;Neuropathy, recurrent, with pressure palsies, 162500;Dejerine-Sottas disease, 145900 |
| PNKP    | 100%  | 100%  | 100% | 99.9% | 99.2% | ?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402                                                                                                                  |

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|-------|-------|-------|------|------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| PNPT1 | 100%  | 100%  | 100% | 100% | 99.8% | Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932                                                                                                                                                          |
| POLG  | 100%  | 100%  | 100% | 100% | 99.8% | Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450 |
| POLG2 | 95.8% | 95.8% | 100% | 100% | 99.5% | 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                                                                                                |

|        |       |       |      |       |       |                                                                                                                                                                                                                                                                                                |
|--------|-------|-------|------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| POLR3A | 100%  | 100%  | 100% | 100%  | 99.6% | Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694                                                                                                                                          |
| POLR3B | 95.4% | 95.4% | 100% | 100%  | 99.7% | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742                                                                                                                       |
| PRDM12 | 97.1% | 92.6% | 100% | 99.9% | 98.8% | Neuropathy, hereditary sensory and autonomic, type VIII, 616488                                                                                                                                                                                                                                |
| PRNP   | 100%  | 100%  | 100% | 100%  | 99.8% | Spongiform encephalopathy with neuropsychiatric features, 606688;Gerstmann-Straussler disease, 137440;Huntington disease-like 1, 603218;Insomnia, fatal familial, 600072;{Kuru, susceptibility to}, 245300;Cerebral amyloid angiopathy, PRNP-related, 137440;Creutzfeldt-Jakob disease, 123400 |
| PRPS1  | 100%  | 100%  | 99%  | 89.8% | 71.8% | Arts syndrome, 301835;Phosphoribosyl pyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661                                                                                         |

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|---------|-------|-------|------|-------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| PRX     | 100%  | 100%  | 100% | 100%  | 99.5% | Charcot-Marie-Tooth disease, type 4F, 614895;Dejerine-Sottas disease, 145900                                                                                                                                                          |
| PSAP    | 99.6% | 97.1% | 100% | 100%  | 99.4% | Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 |
| RAB7A   | 89.2% | 89.2% | 100% | 100%  | 99.5% | Charcot-Marie-Tooth disease, type 2B, 600882                                                                                                                                                                                          |
| RCC1    | 100%  | 100%  | 100% | 99.9% | 99.3% | Infection-induced acute-onset axonal neuropathy, 621333                                                                                                                                                                               |
| REEP1   | 87.5% | 87.5% | 100% | 100%  | 99.7% | Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011;Spastic paraplegia 31, autosomal dominant, 610250;?Neuronopathy , distal hereditary motor, autosomal dominant 12, 614751                                         |
| RETREG1 | 91.3% | 91.3% | 100% | 99.9% | 99.5% | Neuropathy, hereditary sensory and autonomic, type IIB, 613115                                                                                                                                                                        |
| RNF170  | 100%  | 100%  | 100% | 100%  | 99.9% | Ataxia, sensory, 1, autosomal dominant, 608984;Spastic paraplegia 85, autosomal recessive, 619686                                                                                                                                     |

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|--------|-------|-------|------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------------|
| RTN2   | 100%  | 100%  | 100% | 100%  | 99.2% | Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854;Spastic paraplegia 12, autosomal dominant, 604805 |
| SACS   | 99%   | 99%   | 100% | 100%  | 99.9% | Spastic ataxia, Charlevoix-Saguenay type, 270550                                                                                         |
| SAMD9L | 100%  | 100%  | 100% | 100%  | 99.9% | Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270        |
| SBF1   | 100%  | 100%  | 100% | 100%  | 99.2% | Charcot-Marie-Tooth disease, type 4B3, 615284                                                                                            |
| SBF2   | 93.7% | 93.7% | 100% | 100%  | 99.7% | Charcot-Marie-Tooth disease, type 4B2, 604563                                                                                            |
| SCARB2 | 100%  | 100%  | 100% | 100%  | 99.7% | Epilepsy, progressive myoclonic 4, with or without renal failure, 254900                                                                 |
| SCN10A | 100%  | 100%  | 100% | 100%  | 99.8% | Episodic pain syndrome, familial, 2, 615551                                                                                              |
| SCN11A | 100%  | 100%  | 100% | 99.8% | 99.4% | Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548                               |

|         |       |       |      |       |       |                                                                                                                                                                                                               |
|---------|-------|-------|------|-------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| SCN9A   | 100%  | 100%  | 100% | 100%  | 99.5% | Erythralgia, primary, 133020;Insensitivity to pain, congenital, 243000;Small fiber neuropathy, 133020;Paroxysmal extreme pain disorder, 167400;Neuropathy, hereditary sensory and autonomic, type IID, 243000 |
| SCO2    | 100%  | 100%  | 100% | 100%  | 99.8% | Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377                                                                                                                                  |
| SCP2    | 95.5% | 95.5% | 100% | 100%  | 99.7% | ?Leukoencephalopathy with dystonia and motor neuropathy, 613724                                                                                                                                               |
| SCYL1   | 100%  | 100%  | 100% | 99.9% | 99.1% | Spinocerebellar ataxia, autosomal recessive 21, 616719                                                                                                                                                        |
| SEPTIN9 | 100%  | 100%  | 100% | 99.9% | 98.9% | Amyotrophy, hereditary neuralgic, 162100                                                                                                                                                                      |
| SETX    | 100%  | 100%  | 100% | 100%  | 99.8% | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433                                                                               |
| SGPL1   | 95.5% | 95.5% | 100% | 100%  | 99.7% | RENI syndrome, 617575                                                                                                                                                                                         |
| SH3TC2  | 100%  | 100%  | 100% | 100%  | 99.5% | Charcot-Marie-Tooth disease, type 4C, 601596;Mononeuropathy of the median nerve, mild, 613353                                                                                                                 |
| SIGMAR1 | 100%  | 100%  | 100% | 100%  | 100%  | ?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726;?Amyotrophic lateral sclerosis 16, juvenile, 614373                                                                                     |

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| SLC12A6  | 100% | 100% | 100% | 100%  | 99.9% | Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068             |
| SLC25A19 | 100% | 100% | 100% | 100%  | 99.8% | Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710                |
| SLC25A46 | 100% | 100% | 100% | 100%  | 99.7% | Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303                               |
| SLC52A2  | 100% | 100% | 100% | 99.9% | 99.7% | Brown-Vialetto-Van Laere syndrome 2, 614707                                                                                          |
| SLC52A3  | 100% | 100% | 100% | 100%  | 99.4% | ?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530                                                             |
| SLC5A6   | 100% | 100% | 100% | 100%  | 99.5% | Sodium-dependent multivitamin transporter deficiency, 618973;Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903 |
| SLC5A7   | 100% | 100% | 100% | 99.9% | 99.4% | Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143         |
| SOD1     | 100% | 100% | 100% | 100%  | 99.7% | Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400                                 |

|        |       |       |      |       |       |                                                                                                                                                                                                                                                                     |
|--------|-------|-------|------|-------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| SORD   | 83%   | 78.5% | 98%  | 95.9% | 93.1% | Neuronopathy, distal hereditary motor, autosomal recessive 8, 618912                                                                                                                                                                                                |
| SOX10  | 97.8% | 97.8% | 100% | 100%  | 99.2% | Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584                                                                                                                           |
| SPAST  | 94.7% | 94.6% | 100% | 100%  | 99.5% | Spastic paraplegia 4, autosomal dominant, 182601                                                                                                                                                                                                                    |
| SPG11  | 99.6% | 99.6% | 100% | 100%  | 99.7% | Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360                                                                                                           |
| SPG7   | 100%  | 100%  | 100% | 99.9% | 99.3% | Spastic paraplegia 7, autosomal recessive, 607259                                                                                                                                                                                                                   |
| SPTAN1 | 99.2% | 98.8% | 100% | 100%  | 99.5% | Developmental delay with or without epilepsy, 620540;Developmental and epileptic encephalopathy 5, 613477;Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538;Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528 |
| SPTBN4 | 100%  | 100%  | 100% | 99.9% | 98.7% | Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519                                                                                                                                                                                        |

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| SPTLC1 | 84.4% | 83.8% | 100% | 100%  | 99.7% | Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400                                                                        |
| SPTLC2 | 89%   | 89%   | 100% | 100%  | 99.7% | Neuropathy, hereditary sensory and autonomic, type IC, 613640                                                                                                                           |
| SPTLC3 | 91.9% | 91.9% | 100% | 100%  | 99.9% |                                                                                                                                                                                         |
| SURF1  | 100%  | 100%  | 100% | 100%  | 99.4% | Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110                                                                                |
| SYT2   | 100%  | 100%  | 100% | 100%  | 99.4% | Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040;Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461 |
| TBCE   | 96.7% | 96.6% | 100% | 100%  | 99.6% | Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207                |
| TDP1   | 87.9% | 87.5% | 100% | 100%  | 99.8% | ?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250                                                                                                          |
| TDRKH  | 100%  | 100%  | 100% | 99.6% | 98.6% |                                                                                                                                                                                         |
| TECPR2 | 100%  | 100%  | 100% | 99.9% | 99.6% | Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031                                                                                                 |

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| TFG    | 100%  | 100%  | 100% | 100%  | 99.4% | ?Spastic paraplegia 57, autosomal recessive, 615658;Hereditary motor and sensory neuropathy, Okinawa type, 604484                                                                                                                                                                                                                                                                                                                                                                                          |
| TRIM2  | 96.1% | 96.1% | 100% | 100%  | 99.7% | Charcot-Marie-Tooth disease, type 2R, 615490                                                                                                                                                                                                                                                                                                                                                                                                                                                               |
| TRPV4  | 100%  | 100%  | 100% | 99.9% | 99.1% | Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175;Spondylometap hyseal dysplasia, Kozlowski type, 184252;Digital arthropathy-brachydact yly, familial, 606835;[Sodium serum level QTL 1], 613508;SED, Maroteaux type, 184095;Metatropic dysplasia, 156530;Scapuloperone al spinal muscular atrophy, 181405;Hereditary motor and sensory neuropathy, type IIc, 606071;?Avascular necrosis of femoral head, primary, 2, 617383;Parastremmati c dwarfism, 168400;Brachyolmia type 3, 113500 |
| TTR    | 100%  | 100%  | 100% | 100%  | 99.5% | Amyloidosis, hereditary systemic 1, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyreti nemic hyperthyroxinemia], 145680                                                                                                                                                                                                                                                                                                                                                                      |
| TUBB2A | 100%  | 100%  | 100% | 100%  | 99%   | Cortical dysplasia, complex, with other brain malformations 5, 615763                                                                                                                                                                                                                                                                                                                                                                                                                                      |

|        |       |       |      |      |       |                                                                                                                                                                                                      |
|--------|-------|-------|------|------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| TUBB3  | 100%  | 100%  | 100% | 100% | 99.5% | Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039                                                                        |
| TWNK   | 100%  | 100%  | 100% | 100% | 99.6% | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138 |
| TYMP   | 100%  | 100%  | 100% | 100% | 99.4% | Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041                                                                                                                                          |
| UBA5   | 94.1% | 92.2% | 100% | 100% | 99.8% | ?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132                                                                                        |
| UBE3C  | 94.8% | 94.8% | 100% | 100% | 99.6% | Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270                                                                                                     |
| UCHL1  | 100%  | 100%  | 100% | 100% | 99.8% | {?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491                                             |
| UQCRC1 | 100%  | 100%  | 100% | 100% | 99.5% | Parkinsonism with polyneuropathy, 619279                                                                                                                                                             |

|       |       |       |      |       |       |                                                                                                                                                                                                                  |
|-------|-------|-------|------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| VCP   | 100%  | 99.6% | 100% | 100%  | 99.8% | Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954;Charcot-Marie-Tooth disease, type 2Y, 616687;Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 |
| VRK1  | 99.6% | 98%   | 100% | 100%  | 99.9% | Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542                                                                                                 |
| VWA1  | 100%  | 100%  | 100% | 100%  | 99.1% | Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216                                                                                                                                             |
| WARS1 | 91.8% | 91.8% | 100% | 100%  | 99.6% | Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721;Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317                                  |
| WNK1  | 100%  | 100%  | 100% | 99.9% | 99%   | Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoparathyroidism, type IIC, 614492                                                                                                         |
| XRCC1 | 100%  | 100%  | 100% | 100%  | 99.2% | ?Spinocerebellar ataxia, autosomal recessive 26, 617633                                                                                                                                                          |
| YARS1 | 100%  | 98.5% | 100% | 100%  | 99.5% | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323                                                                 |

|         |      |      |      |      |       |                                                          |
|---------|------|------|------|------|-------|----------------------------------------------------------|
| ZFYVE26 | 100% | 100% | 100% | 100% | 99.7% | Spastic paraplegia 15,<br>autosomal recessive,<br>270700 |
|---------|------|------|------|------|-------|----------------------------------------------------------|

Gene symbols used follow HGNC 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 mapped against GRCh38.*  
*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 mapped against GRCh38.*  
*srWGS 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 covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.*  
*srWGS 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 : November 25th, 2024.*  
*This list is accurate for panel version DG 5.1.0*  
*Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*