# MOVEMENT DISORDERS PANEL DG-5.2.0 (469 GENES)

*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.*
*OMIM disease identifiers and descriptions : October 25th, 2026.*
*This list is accurate for panel version DG 5.2.0*
*non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*
*Ad 1. Blank field signifies a gene without a current OMIM association*
*Ad 2. OMIM phenotype descriptions between {} signify risk factors*
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
*- GQ 20: 99% confidence in the genotype call*
*- GQ 30: 99.9% confidence in the genotype call*

| Gene | WES GQ>20 \| GQ>30* | srWGS GQ>20 \| GQ>30* | lrWGS GQ>20 \| GQ>30* | Associated Phenotype description and OMIM disease ID |
|---|---|---|---|---|
| AARS2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096 |
| ABCB7 | 100% \| 100% | 99% \| 99% | 100% \| 100% | Anemia, sideroblastic, with ataxia, 301310 |
| ABCD1 | 85% \| 84% | 98% \| 98% | 100% \| 100% | Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100 |
| ABHD12 | 99% \| 99% | 95% \| 95% | 99% \| 99% | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 |
| ABHD16A | 98% \| 98% | 96% \| 96% | 99% \| 99% | Spastic paraplegia 86, autosomal recessive, 619735 |
| ACO2 | 85% \| 85% | 97% \| 97% | 99% \| 99% | Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559 |
| ACTB | 100% \| 100% | 97% \| 97% | 98% \| 98% | Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470 |
| ADAR | 100% \| 100% | 96% \| 96% | 99% \| 99% | Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010 |
| ADCY5 | 96% \| 96% | 96% \| 96% | 99% \| 99% | Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647 |
| ADD3 | 98% \| 98% | 98% \| 98% | 99% \| 99% | Cerebral palsy, spastic quadriplegic, 3, 617008 |
| ADGRG1 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752;Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854 |
| ADPRS | 100% \| 100% | 98% \| 98% | 99% \| 99% | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 |
| AFG3L2 | 93% \| 92% | 97% \| 97% | 99% \| 99% | Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246 |

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|---|---|---|---|---|
| AGA | 92% \| 92% | 97% \| 97% | 99% \| 99% | Aspartylglucosaminuria, 208400 |
| AGTPBP1 | 97% \| 97% | 98% \| 98% | 100% \| 100% | Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 |
| AIFM1 | 99% \| 99% | 99% \| 99% | 100% \| 100% | Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614 |
| AIMP1 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Leukodystrophy, hypomyelinating, 3, 260600 |
| ALDH18A1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603 |
| ALDH3A2 | 93% \| 93% | 97% \| 97% | 99% \| 99% | Sjogren-Larsson syndrome, 270200 |
| ALDH5A1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Succinic semialdehyde dehydrogenase deficiency, 271980 |
| ALS2 | 96% \| 96% | 98% \| 98% | 99% \| 99% | Primary lateral sclerosis, juvenile, 606353;Spastic paralysis, infantile onset ascending, 607225;Amyotrophic lateral sclerosis 2, juvenile, 205100 |
| AMACR | 99% \| 99% | 97% \| 97% | 99% \| 99% | Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950 |
| AMFR | 100% \| 100% | 97% \| 97% | 99% \| 98% | Spastic paraplegia 89, autosomal recessive, 620379 |
| AMPD2 | 100% \| 100% | 96% \| 96% | 99% \| 98% | Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686 |
| ANO10 | 96% \| 96% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia, autosomal recessive 10, 613728 |
| ANO3 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Dystonia 24, 615034 |
| AOPEP | 100% \| 100% | 97% \| 97% | 99% \| 99% | Dystonia 31, 619565 |
| AP4B1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Spastic paraplegia 47, autosomal recessive, 614066 |
| AP4E1 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Stuttering, familial persistent, 1, 184450;Spastic paraplegia 51, autosomal recessive, 613744 |
| AP4M1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Spastic paraplegia 50, autosomal recessive, 612936 |
| AP4S1 | 78% \| 78% | 94% \| 94% | 99% \| 99% | Spastic paraplegia 52, autosomal recessive, 614067 |
| AP5Z1 | 91% \| 91% | 96% \| 96% | 98% \| 98% | Spastic paraplegia 48, autosomal recessive, 613647 |
| APTX | 100% \| 100% | 96% \| 96% | 100% \| 100% | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 |
| ARG1 | 93% \| 93% | 98% \| 98% | 100% \| 100% | Argininemia, 207800 |
| ARL6IP1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Spastic paraplegia 61, autosomal recessive, 615685 |
| ARSA | 100% \| 100% | 96% \| 96% | 98% \| 98% | Metachromatic leukodystrophy, 250100 |

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|---|---|---|---|---|
| ARX | 93% \| 93% | 96% \| 96% | 100% \| 100% | Proud syndrome, 300004;Hydranencephaly with abnormal genitalia, 300215;Partington syndrome, 309510;Developmental and epileptic encephalopathy 1, 308350;Lissencephaly, X-linked 2, 300215;Intellectual developmental disorder, X-linked 29, 300419 |
| ASPA | 100% \| 100% | 98% \| 98% | 99% \| 99% | Canavan disease, 271900 |
| ATCAY | 100% \| 100% | 96% \| 96% | 98% \| 98% | Ataxia, cerebellar, Cayman type, 601238 |
| ATG7 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Spinocerebellar ataxia, autosomal recessive 31, 619422 |
| ATL1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708 |
| ATM | 100% \| 100% | 98% \| 98% | 99% \| 99% | Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic |
| ATN1 | 98% \| 98% | 94% \| 94% | 97% \| 97% | Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 |
| ATP13A2 | 99% \| 99% | 96% \| 96% | 98% \| 98% | Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693 |
| ATP1A2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Developmental and epileptic encephalopathy 98, 619605;Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602;Alternating hemiplegia of childhood 1, 104290;Migraine, familial basilar, 602481;Migraine, familial hemiplegic, 2, 602481 |
| ATP1A3 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606 |
| ATP2B3 | 98% \| 98% | 98% \| 98% | 99% \| 99% | ?Spinocerebellar ataxia, X-linked 1, 302500 |
| ATP7B | 100% \| 100% | 97% \| 97% | 99% \| 99% | Wilson disease, 277900 |
| ATP8A2 | 100% \| 100% | 97% \| 97% | 100% \| 100% | Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268 |
| B4GALNT1 | 100% \| 100% | 97% \| 97% | 98% \| 98% | Spastic paraplegia 26, autosomal recessive, 609195 |
| B9D1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | ?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120 |
| BCAP31 | 87% \| 86% | 99% \| 99% | 100% \| 100% | Deafness, dystonia, and cerebral hypomyelination, 300475 |
| BCKDHA | 86% \| 86% | 95% \| 95% | 98% \| 98% | Maple syrup urine disease, type Ia, 248600 |
| BCKDHB | 88% \| 88% | 97% \| 97% | 99% \| 99% | Maple syrup urine disease, type Ib, 620698 |
| BCL11B | 98% \| 98% | 94% \| 94% | 99% \| 99% | Immunodeficiency 49, severe combined, 617237;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 |
| BRAT1 | 94% \| 94% | 96% \| 96% | 96% \| 96% | Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 |

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|---|---|---|---|---|
| BSCL2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | 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 |
| BTD | 95% \| 95% | 98% \| 98% | 99% \| 99% | Biotinidase deficiency, 253260 |
| C19orf12 | 99% \| 99% | 95% \| 95% | 98% \| 98% | Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043 |
| CA8 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia, autosomal recessive 34, 613227 |
| CACNA1A | 99% \| 99% | 95% \| 95% | 98% \| 98% | Developmental and epileptic encephalopathy 42A, 617106;Spinocerebellar ataxia 6, 183086;Episodic ataxia, type 2, 108500;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500;Migraine, familial hemiplegic, 1, 141500;Developmental and epileptic encephalopathy 42B, 621692 |
| CACNA1E | 100% \| 100% | 97% \| 97% | 100% \| 100% | Developmental and epileptic encephalopathy 69, 618285 |
| CACNA1G | 100% \| 100% | 96% \| 96% | 98% \| 98% | Spinocerebellar ataxia 42, 616795;Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 |
| CAMTA1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 |
| CAPN1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Spastic paraplegia 76, autosomal recessive, 616907 |
| CAPRIN1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782;Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636 |
| CC2D2A | 98% \| 98% | 98% \| 98% | 99% \| 99% | COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285 |
| CCT5 | 99% \| 99% | 96% \| 96% | 99% \| 99% | ?Neuropathy, hereditary sensory, with spastic paraplegia, 256840 |
| CHMP1A | 100% \| 100% | 95% \| 95% | 98% \| 98% | Pontocerebellar hypoplasia, type 8, 614961 |
| CLCN2 | 98% \| 98% | 97% \| 97% | 98% \| 98% | Leukoencephalopathy with ataxia, 615651;Hyperaldosteronism, familial, type II, 605635;{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628;{Epilepsy, juvenile absence, susceptibility to, 2}, 607628;{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 |
| CLCN4 | 99% \| 99% | 98% \| 98% | 100% \| 100% | Raynaud-Claes syndrome, 300114 |
| CLCN6 | 98% \| 98% | 96% \| 96% | 99% \| 99% | Ceroid lipofuscinosis, neuronal, 15, 619173 |
| CLN5 | 83% \| 83% | 97% \| 97% | 99% \| 99% | Ceroid lipofuscinosis, neuronal, 5, 256731 |
| CLN6 | 96% \| 96% | 97% \| 97% | 99% \| 99% | Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780 |
| CLN8 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143 |
| CLP1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Pontocerebellar hypoplasia, type 10, 615803 |

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| CLPB | 98% \| 98% | 97% \| 97% | 99% \| 99% | Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 |
| COA7 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 |
| COASY | 100% \| 100% | 96% \| 96% | 98% \| 98% | Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643 |
| COL4A1 | 100% \| 100% | 97% \| 97% | 98% \| 98% | ?Retinal arteries, tortuosity of, 180000;{Hemorrhage, intracerebral, susceptibility to}, 614519;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773;Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564;Brain small vessel disease with or without ocular anomalies, 175780 |
| COL4A2 | 98% \| 98% | 96% \| 96% | 98% \| 98% | Brain small vessel disease 2A, autosomal dominant, 614483;{Hemorrhage, intracerebral, susceptibility to}, 614519;Brain small vessel disease 2B, autosomal recessive, 621414 |
| COL6A1 | 100% \| 100% | 94% \| 94% | 97% \| 96% | Ullrich congenital muscular dystrophy 1A, 254090;Bethlem myopathy 1A, 158810 |
| COL6A2 | 99% \| 99% | 95% \| 95% | 97% \| 97% | ?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1B, 620727;Bethlem myopathy 1B, 620725 |
| COL6A3 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Bethlem myopathy 1C, 620726;Ullrich congenital muscular dystrophy 1C, 620728;Dystonia 27, 616411 |
| COQ2 | 97% \| 97% | 97% \| 96% | 99% \| 99% | {Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426 |
| COQ4 | 97% \| 97% | 96% \| 96% | 98% \| 98% | Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666 |
| COQ8A | 87% \| 87% | 95% \| 95% | 98% \| 98% | Coenzyme Q10 deficiency, primary, 4, 612016 |
| COQ9 | 92% \| 92% | 97% \| 97% | 99% \| 99% | Coenzyme Q10 deficiency, primary, 5, 614654 |
| COX20 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Mitochondrial complex IV deficiency, nuclear type 11, 619054 |
| CP | 100% \| 100% | 97% \| 97% | 99% \| 99% | Aceruloplasminemia, 604290 |
| CSF1R | 97% \| 97% | 96% \| 96% | 99% \| 99% | Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 |
| CSTB | 100% \| 100% | 92% \| 92% | 97% \| 97% | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 |
| CTBP1 | 100% \| 100% | 94% \| 94% | 98% \| 98% | Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 |
| CTC1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Cerebroretinal microangiopathy with calcifications and cysts, 612199 |
| CTNNB1 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Exudative vitreoretinopathy 7, 617572;Pilomatricoma, somatic, 132600;Colorectal cancer, somatic, 114500;Neurodevelopmental disorder with spastic diplegia and visual defects, 615075;Medulloblastoma, somatic, 155255;Ovarian cancer, somatic, 167000;Hepatocellular carcinoma, somatic, 114550 |

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| CTSD | 87% \| 87% | 97% \| 97% | 98% \| 97% | Ceroid lipofuscinosis, neuronal, 10, 610127 |
| CTSF | 100% \| 100% | 97% \| 96% | 98% \| 98% | Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362 |
| CWF19L1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Spinocerebellar ataxia, autosomal recessive 17, 616127 |
| CYP27A1 | 100% \| 100% | 97% \| 97% | 100% \| 100% | Cerebrotendinous xanthomatosis, 213700 |
| CYP2U1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Spastic paraplegia 56, autosomal recessive, 615030 |
| CYP7B1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812 |
| DARS1 | 100% \| 100% | 97% \| 97% | 100% \| 100% | Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 |
| DARS2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105;Charcot-Marie-Tooth disease, axonal, type 2LL, 621485 |
| DBT | 92% \| 92% | 96% \| 96% | 99% \| 99% | Maple syrup urine disease, type II, 620699 |
| DCAF17 | 97% \| 97% | 97% \| 97% | 99% \| 99% | Woodhouse-Sakati syndrome, 241080 |
| DCC | 100% \| 100% | 98% \| 98% | 100% \| 100% | Mirror movements 1 and/or agenesis of the corpus callosum, 157600;Esophageal carcinoma, somatic, 133239;Colorectal cancer, somatic, 114500;Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 |
| DCTN1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641 |
| DDC | 100% \| 100% | 97% \| 97% | 99% \| 99% | Aromatic L-amino acid decarboxylase deficiency, 608643 |
| DDHD1 | 100% \| 100% | 96% \| 95% | 99% \| 99% | Spastic paraplegia 28, autosomal recessive, 609340 |
| DDHD2 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Spastic paraplegia 54, autosomal recessive, 615033 |
| DEGS1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Leukodystrophy, hypomyelinating, 18, 618404 |
| DHDDS | 76% \| 76% | 97% \| 97% | 99% \| 99% | Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 621567;Retinitis pigmentosa 59, 613861 |
| DHPS | 93% \| 93% | 96% \| 96% | 98% \| 98% | Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 |
| DHX30 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Neurodevelopmental disorder with variable motor and speech impairment, 617804 |
| DLAT | 100% \| 100% | 97% \| 97% | 99% \| 99% | Pyruvate dehydrogenase E2 deficiency, 245348 |
| DLD | 100% \| 100% | 98% \| 98% | 99% \| 99% | Dihydrolipoamide dehydrogenase deficiency, 246900 |
| DNAJC12 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 |
| DNAJC3 | 92% \| 92% | 98% \| 98% | 99% \| 99% | Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 |

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| DNAL4 | 100% \| 100% | 96% \| 96% | 99% \| 99% | ?Mirror movements 3, 616059 |
| DNM1L | 100% \| 100% | 98% \| 98% | 99% \| 99% | Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 |
| DNMT1 | 99% \| 99% | 96% \| 96% | 99% \| 99% | Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 |
| DPYS | 100% \| 100% | 96% \| 96% | 99% \| 99% | Dihydropyrimidinuria, 222748 |
| DSTYK | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 23, autosomal recessive, 270750;Congenital anomalies of kidney and urinary tract 1, 610805 |
| DTYMK | 84% \| 84% | 96% \| 96% | 99% \| 99% | Neurodegeneration, childhood-onset, with progressive microcephaly, 619847 |
| EBF3 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Hypotonia, ataxia, and delayed development syndrome, 617330 |
| ECHS1 | 68% \| 68% | 96% \| 96% | 97% \| 97% | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 |
| EIF2AK1 | 94% \| 94% | 96% \| 96% | 99% \| 99% | ?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 |
| EIF2AK2 | 100% \| 100% | 97% \| 97% | 100% \| 100% | Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877;Dystonia 33, 619687 |
| EIF2B1 | 90% \| 90% | 96% \| 96% | 99% \| 99% | Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896 |
| EIF2B2 | 100% \| 100% | 94% \| 94% | 99% \| 99% | Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312 |
| EIF2B3 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313 |
| EIF2B4 | 100% \| 100% | 97% \| 97% | 98% \| 98% | Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314 |
| EIF2B5 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 |
| ELOVL1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 |
| ELOVL4 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 |
| ELOVL5 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia 38, 615957 |
| ENTPD1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 64, autosomal recessive, 615683 |
| EPM2A | 97% \| 97% | 91% \| 91% | 99% \| 99% | Myoclonic epilepsy of Lafora 1, 254780 |
| ERCC2 | 92% \| 91% | 96% \| 96% | 98% \| 98% | Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756 |

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| ERCC4 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272 |
| ERLIN1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Spastic paraplegia 62, autosomal recessive, 615681 |
| ERLIN2 | 100% \| 100% | 97% \| 97% | 100% \| 100% | Spastic paraplegia 18A, autosomal dominant, 620512;Spastic paraplegia 18B, autosomal recessive, 611225 |
| ETHE1 | 77% \| 77% | 94% \| 94% | 99% \| 99% | Ethylmalonic encephalopathy, 602473 |
| EXOSC3 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Pontocerebellar hypoplasia, type 1B, 614678 |
| EXOSC5 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576 |
| EXOSC8 | 100% \| 100% | 96% \| 96% | 100% \| 100% | Pontocerebellar hypoplasia, type 1C, 616081 |
| EXOSC9 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Pontocerebellar hypoplasia, type 1D, 618065 |
| FA2H | 100% \| 100% | 97% \| 97% | 98% \| 98% | Spastic paraplegia 35, autosomal recessive, 612319 |
| FAR1 | 91% \| 91% | 98% \| 98% | 100% \| 100% | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338 |
| FARS2 | 100% \| 100% | 97% \| 97% | 100% \| 100% | Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046 |
| FBXO7 | 99% \| 99% | 97% \| 97% | 100% \| 100% | Parkinson disease 15, 260300 |
| FGF14 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia 27A, 193003;Spinocerebellar ataxia 27B, late-onset, 620174 |
| FICD | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 92, autosomal recessive, 620911 |
| FLVCR1 | 95% \| 95% | 97% \| 97% | 99% \| 99% | Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060 |
| FOLR1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Neurodegeneration due to cerebral folate transport deficiency, 613068 |
| FRMD5 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094 |
| FRMD7 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Nystagmus, infantile periodic alternating, X-linked, 310700;Nystagmus 1, congenital, X-linked, 310700 |
| FTH1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Neurodegeneration with brain iron accumulation 9, 620669;?Hemochromatosis, type 5, 615517 |
| FTL | 97% \| 97% | 97% \| 97% | 98% \| 98% | Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159 |
| FXN | 99% \| 99% | 92% \| 91% | 99% \| 99% | Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300 |
| GALC | 99% \| 99% | 96% \| 96% | 99% \| 99% | Krabbe disease, 245200 |
| GAMT | 91% \| 90% | 96% \| 96% | 97% \| 97% | Cerebral creatine deficiency syndrome 2, 612736 |

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|---|---|---|---|---|
| GAN | 100% \| 100% | 97% \| 97% | 99% \| 99% | Giant axonal neuropathy-1, 256850 |
| GBA1 | 96% \| 95% | 96% \| 95% | 99% \| 99% | {Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600 |
| GBA2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 46, autosomal recessive, 614409 |
| GBE1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570 |
| GCDH | 79% \| 79% | 95% \| 95% | 99% \| 99% | Glutaricaciduria, type I, 231670 |
| GCH1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910 |
| GDAP2 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Spinocerebellar ataxia, autosomal recessive 27, 618369 |
| GEMIN5 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 |
| GFAP | 100% \| 100% | 97% \| 97% | 99% \| 99% | Alexander disease, 203450 |
| GJB1 | 98% \| 98% | 97% \| 97% | 100% \| 100% | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 |
| GJC2 | 95% \| 95% | 94% \| 94% | 99% \| 99% | Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804 |
| GLB1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600 |
| GLRA1 | 100% \| 100% | 97% \| 97% | 100% \| 100% | Hyperekplexia 1, 149400 |
| GLRB | 95% \| 95% | 98% \| 98% | 99% \| 99% | Hyperekplexia 2, 614619 |
| GLS | 96% \| 96% | 97% \| 97% | 99% \| 99% | CASGID syndrome, 618339;Global developmental delay, progressive ataxia, and elevated glutamine, 618412;Developmental and epileptic encephalopathy 71, 618328 |
| GNAL | 100% \| 100% | 97% \| 97% | 99% \| 99% | Dystonia 25, 615073 |
| GNAO1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493 |
| GOSR2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166 |
| GPAA1 | 88% \| 88% | 96% \| 96% | 98% \| 98% | Glycosylphosphatidylinositol biosynthesis defect 15, 617810 |
| GPR143 | 92% \| 92% | 99% \| 99% | 100% \| 100% | Ocular albinism, type I, Nettleship-Falls type, 300500;Nystagmus 6, congenital, X-linked, 300814 |
| GRIA4 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 |

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| GRID2 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Spinocerebellar ataxia, autosomal recessive 18, 616204 |
| GRIK2 | 96% \| 96% | 97% \| 97% | 100% \| 100% | Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580;Intellectual developmental disorder, autosomal recessive 6, 611092 |
| GRIN1 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820;Developmental and epileptic encephalopathy 101, 619814;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 |
| GRIN2B | 100% \| 100% | 97% \| 97% | 100% \| 100% | Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 |
| GRM1 | 100% \| 100% | 97% \| 97% | 100% \| 100% | Spinocerebellar ataxia, autosomal recessive 13, 614831;Spinocerebellar ataxia 44, 617691 |
| GRN | 100% \| 100% | 97% \| 97% | 99% \| 99% | Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706 |
| HACE1 | 96% \| 96% | 97% \| 97% | 100% \| 100% | Spastic paraplegia and psychomotor retardation with or without seizures, 616756 |
| HEPACAM | 100% \| 100% | 97% \| 97% | 99% \| 99% | Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926 |
| HEXA | 100% \| 100% | 95% \| 95% | 99% \| 99% | [Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800 |
| HEXB | 100% \| 100% | 96% \| 96% | 99% \| 99% | Sandhoff disease, infantile, juvenile, and adult forms, 268800 |
| HK1 | 96% \| 96% | 97% \| 97% | 99% \| 99% | 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 |
| HNRNPR | 100% \| 100% | 97% \| 97% | 99% \| 99% | Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073 |
| HPCA | 100% \| 100% | 97% \| 97% | 98% \| 98% | Dystonia 2, torsion, autosomal recessive, 224500 |
| HPDL | 100% \| 100% | 97% \| 97% | 99% \| 99% | Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027 |
| HPRT1 | 89% \| 89% | 99% \| 99% | 100% \| 100% | Hyperuricemia, HPRT-related, 300323;Lesch-Nyhan syndrome, 300322 |
| HSD17B4 | 100% \| 100% | 96% \| 96% | 100% \| 100% | D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400 |
| HSPD1 | 99% \| 98% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233 |
| HTRA1 | 99% \| 99% | 96% \| 96% | 99% \| 99% | Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 2, 616779;Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 2, 600142 |
| HYCC1 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Leukodystrophy, hypomyelinating, 5, 610532 |

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| IBA57 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451 |
| IRF2BPL | 95% \| 95% | 87% \| 87% | 97% \| 97% | Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 |
| ISCA2 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Multiple mitochondrial dysfunctions syndrome 4, 616370 |
| ITPR1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658 |
| JAM2 | 88% \| 88% | 98% \| 98% | 99% \| 99% | Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 |
| JAM3 | 91% \| 91% | 98% \| 98% | 99% \| 99% | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 |
| KARS1 | 94% \| 94% | 96% \| 96% | 99% \| 99% | 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 |
| KATNB1 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Lissencephaly 6, with microcephaly, 616212 |
| KCNA1 | 100% \| 100% | 95% \| 95% | 99% \| 99% | Episodic ataxia/myokymia syndrome, 160120 |
| KCNA2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Developmental and epileptic encephalopathy 32, 616366 |
| KCNC1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Epilepsy, progressive myoclonic 7, 616187 |
| KCNC3 | 97% \| 97% | 88% \| 88% | 96% \| 95% | Spinocerebellar ataxia 13, 605259 |
| KCND3 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399 |
| KCNJ10 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780 |
| KCNJ6 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Keppen-Lubinsky syndrome, 614098 |
| KCNMA1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446;Cerebellar atrophy, developmental delay, and seizures, 617643;Liang-Wang syndrome, 618729 |
| KCNN2 | 100% \| 100% | 95% \| 95% | 99% \| 99% | ?Dystonia 34, myoclonic, 619724;Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 |
| KCTD17 | 100% \| 100% | 95% \| 95% | 98% \| 98% | Dystonia 26, myoclonic, 616398 |
| KCTD7 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 |
| KIDINS220 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501 |
| KIF1A | 100% \| 100% | 96% \| 96% | 98% \| 98% | NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607 |

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| KIF1C | 100% \| 100% | 96% \| 96% | 99% \| 99% | Spastic ataxia 2, autosomal recessive, 611302 |
| KIF5A | 100% \| 100% | 96% \| 96% | 99% \| 99% | Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187 |
| KMT2B | 99% \| 99% | 96% \| 96% | 98% \| 98% | Intellectual developmental disorder, autosomal dominant 68, 619934;Dystonia 28, childhood-onset, 617284 |
| KPNA3 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 88, autosomal dominant, 620106 |
| L1CAM | 97% \| 97% | 99% \| 99% | 99% \| 99% | MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100 |
| LAMA1 | 99% \| 99% | 97% \| 97% | 99% \| 99% | Poretti-Boltshauser syndrome, 615960 |
| LAMB1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Leukoencephalopathy without lacunae, adult-onset, 621424;Leukoencephalopathy with variable cortical brain malformations and/or hydrocephalus, 615191 |
| LMNB1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Microcephaly 26, primary, autosomal dominant, 619179 |
| MAG | 100% \| 100% | 96% \| 96% | 99% \| 99% | Spastic paraplegia 75, autosomal recessive, 616680 |
| MAPK8IP3 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Neurodevelopmental disorder with or without variable brain abnormalities, 618443 |
| MARS2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | ?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390 |
| MBIP | 96% \| 96% | 97% \| 97% | 99% \| 99% | |
| MECP2 | 96% \| 96% | 97% \| 97% | 99% \| 99% | Rett syndrome, atypical, 312750;Encephalopathy, neonatal severe, 300673;Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260;{Autism susceptibility, X-linked 3}, 300496;Intellectual developmental disorder, X-linked syndromic 13, 300055;Rett syndrome, 312750;Rett syndrome, preserved speech variant, 312750 |
| MECR | 94% \| 94% | 97% \| 97% | 99% \| 99% | ?Optic atrophy 16, 620629;Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 |
| MFF | 90% \| 90% | 98% \| 98% | 99% \| 99% | Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 |
| MFSD8 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951 |
| MICU1 | 87% \| 87% | 97% \| 97% | 99% \| 99% | Myopathy with extrapyramidal signs, 615673 |
| MLC1 | 99% \| 99% | 91% \| 90% | 98% \| 98% | Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004 |
| MMADHC | 80% \| 80% | 96% \| 96% | 100% \| 100% | Methylmalonic aciduria and homocystinuria, cblD type, 277410;Methylmalonic aciduria, cblD type, 620953;Homocystinuria-megaloblastic anemia, cblD type, 620952 |
| MRE11 | 98% \| 98% | 98% \| 98% | 99% \| 99% | Ataxia-telangiectasia-like disorder 1, 604391 |

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|---|---|---|---|---|
| MSTO1 | 92% \| 90% | 95% \| 94% | 99% \| 99% | Myopathy, mitochondrial, and ataxia, 617675 |
| MTHFR | 100% \| 100% | 96% \| 96% | 99% \| 99% | {Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634 |
| MTHFS | 100% \| 100% | 96% \| 96% | 99% \| 99% | Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 |
| MTPAP | 97% \| 97% | 95% \| 95% | 99% \| 99% | ?Spastic ataxia 4, autosomal recessive, 613672 |
| MTRFR | 98% \| 98% | 96% \| 96% | 98% \| 98% | Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559 |
| MTTP | 100% \| 100% | 98% \| 98% | 100% \| 100% | Abetalipoproteinemia, 200100 |
| MYBPC1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Congenital myopathy 16, 618524;Lethal congenital contracture syndrome 4, 614915;Arthrogryposis, distal, type 1B, 614335 |
| MYORG | 99% \| 99% | 97% \| 97% | 99% \| 99% | Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 |
| NAA60 | 99% \| 99% | 96% \| 96% | 98% \| 98% | Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786 |
| NANS | 95% \| 95% | 97% \| 97% | 99% \| 99% | Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 |
| NARS2 | 92% \| 92% | 97% \| 97% | 99% \| 99% | Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434 |
| NAXE | 89% \| 88% | 95% \| 95% | 98% \| 98% | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 |
| NDUFS7 | 86% \| 84% | 96% \| 96% | 97% \| 96% | Mitochondrial complex I deficiency, nuclear type 3, 618224 |
| NEFL | 100% \| 100% | 95% \| 95% | 100% \| 100% | Charcot-Marie-Tooth disease, type 1F, 607734;Charcot-Marie-Tooth disease, dominant intermediate G, 617882;Charcot-Marie-Tooth disease, type 2E, 607684 |
| NEU1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Sialidosis, type II, 256550;Sialidosis, type I, 256550 |
| NEXMIF | 100% \| 100% | 99% \| 99% | 100% \| 100% | Intellectual developmental disorder, X-linked 98, 300912 |
| NF2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Meningioma, NF2-related, somatic, 607174;Schwannomatosis, vestibular, 101000;Schwannomatosis, somatic, 101000 |
| NFU1 | 95% \| 94% | 95% \| 95% | 99% \| 99% | Spastic paraplegia 93, autosomal recessive, 620938;Multiple mitochondrial dysfunctions syndrome 1, 605711 |
| NGLY1 | 98% \| 98% | 98% \| 98% | 100% \| 100% | Congenital disorder of deglycosylation 1, 615273 |
| NIPA1 | 99% \| 99% | 96% \| 96% | 99% \| 98% | Spastic paraplegia 6, autosomal dominant, 600363 |
| NKX2-1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Chorea, hereditary benign, 118700;{Thyroid cancer, nonmedullary, 1}, 188550;Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 |
| NKX6-2 | 100% \| 100% | 94% \| 94% | 97% \| 97% | Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 |

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| NPC1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220 |
| NPC2 | 95% \| 95% | 97% \| 97% | 100% \| 100% | Niemann-pick disease, type C2, 607625 |
| NPTX1 | 99% \| 99% | 95% \| 95% | 98% \| 98% | Spinocerebellar ataxia 50, 620158 |
| NR4A2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911 |
| NT5C2 | 91% \| 91% | 98% \| 98% | 99% \| 99% | Spastic paraplegia 45, autosomal recessive, 613162 |
| NUBPL | 89% \| 89% | 98% \| 98% | 99% \| 99% | Mitochondrial complex I deficiency, nuclear type 21, 618242 |
| NUP62 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Striatonigral degeneration, infantile, 271930 |
| NUS1 | 95% \| 95% | 96% \| 96% | 100% \| 100% | Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082 |
| OCLN | 85% \| 82% | 93% \| 90% | 99% \| 99% | Pseudo-TORCH syndrome 1, 251290 |
| OGDHL | 99% \| 99% | 97% \| 97% | 99% \| 99% | |
| OPA1 | 95% \| 95% | 97% \| 97% | 100% \| 100% | Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type), 621481;?Mitochondrial DNA depletion syndrome 14B (cardioencephalomyopathic type), 616896 |
| OPA3 | 100% \| 100% | 96% \| 96% | 99% \| 99% | 3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300 |
| OPHN1 | 93% \| 93% | 99% \| 99% | 100% \| 100% | Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 |
| PACS2 | 77% \| 77% | 97% \| 97% | 99% \| 99% | Developmental and epileptic encephalopathy 66, 618067 |
| PANK2 | 100% \| 100% | 97% \| 97% | 98% \| 98% | Neurodegeneration with brain iron accumulation 1, 234200 |
| PAX6 | 99% \| 99% | 93% \| 93% | 99% \| 99% | Optic nerve hypoplasia, 165550;Cataract with late-onset corneal dystrophy, 106210;Microphthalmia/coloboma 12, 120200;?Coloboma of optic nerve, 120430;Aniridia, 106210;Anterior segment dysgenesis 5, multiple subtypes, 604229;?Morning glory disc anomaly, 120430;Foveal hypoplasia 1, 136520;Keratitis, 148190 |
| PCYT2 | 94% \| 93% | 96% \| 96% | 98% \| 98% | Spastic paraplegia 82, autosomal recessive, 618770 |
| PDE10A | 97% \| 97% | 93% \| 93% | 99% \| 99% | Striatal degeneration, autosomal dominant, 616922;Dyskinesia, limb and orofacial, infantile-onset, 616921 |
| PDE2A | 100% \| 100% | 96% \| 96% | 98% \| 98% | Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 |
| PDE8B | 100% \| 100% | 97% \| 97% | 100% \| 100% | Pigmented nodular adrenocortical disease, primary, 3, 614190;Striatal degeneration, autosomal dominant, 609161 |
| PDGFB | 96% \| 95% | 97% \| 97% | 98% \| 98% | Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483 |

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| PDGFRB | 97% \| 97% | 97% \| 97% | 99% \| 99% | Premature aging syndrome, Penttinen type, 601812;?Ocular pterygium-digital keloid dysplasia syndrome, 621091;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007 |
| PDHA1 | 88% \| 88% | 98% \| 98% | 99% \| 99% | Pyruvate dehydrogenase E1-alpha deficiency, 312170 |
| PDHX | 91% \| 91% | 97% \| 97% | 99% \| 99% | Lacticacidemia due to PDX1 deficiency, 245349 |
| PDSS1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Coenzyme Q10 deficiency, primary, 2, 614651 |
| PDSS2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Coenzyme Q10 deficiency, primary, 3, 614652 |
| PDYN | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia 23, 610245 |
| PEX10 | 100% \| 100% | 96% \| 96% | 97% \| 97% | Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871 |
| PEX2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867 |
| PEX7 | 99% \| 99% | 97% \| 97% | 100% \| 100% | Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879 |
| PHYH | 100% \| 100% | 96% \| 96% | 98% \| 98% | Refsum disease, 266500 |
| PIGG | 100% \| 100% | 97% \| 97% | 99% \| 99% | [Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917 |
| PLA2G6 | 100% \| 99% | 97% \| 97% | 93% \| 87% | Neurodegeneration with brain iron accumulation 2B, 610217;Parkinson disease 14, 612953;Infantile neuroaxonal dystrophy 1, 256600 |
| PLP1 | 93% \| 93% | 99% \| 99% | 100% \| 100% | Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920 |
| PMM2 | 95% \| 95% | 97% \| 97% | 99% \| 99% | Congenital disorder of glycosylation, type Ia, 212065 |
| PMP22 | 100% \| 100% | 97% \| 97% | 99% \| 99% | 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 |
| PMPCA | 96% \| 96% | 96% \| 96% | 99% \| 99% | Spinocerebellar ataxia, autosomal recessive 2, 213200 |
| PNKD | 100% \| 100% | 97% \| 97% | 99% \| 99% | Paroxysmal nonkinesigenic dyskinesia 1, 118800 |
| PNKP | 100% \| 100% | 96% \| 96% | 97% \| 97% | ?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402 |
| PNPLA6 | 100% \| 100% | 95% \| 95% | 98% \| 98% | Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470 |
| PNPT1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | 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% \| 100% | 97% \| 97% | 99% \| 99% | 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 |
| POLR1C | 83% \| 83% | 96% \| 96% | 99% \| 99% | Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390 |
| POLR3A | 100% \| 100% | 97% \| 97% | 99% \| 99% | Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 |
| POLR3B | 95% \| 95% | 97% \| 97% | 99% \| 99% | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 |
| POU4F1 | 91% \| 90% | 91% \| 91% | 99% \| 99% | Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352 |
| PPP1R3F | 95% \| 95% | 98% \| 98% | 99% \| 99% | |
| PPT1 | 90% \| 90% | 96% \| 96% | 99% \| 99% | Ceroid lipofuscinosis, neuronal, 1, 256730 |
| PRDX3 | 97% \| 96% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871 |
| PRF1 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027 |
| PRICKLE1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Epilepsy, progressive myoclonic 1B, 612437 |
| PRKCG | 100% \| 100% | 95% \| 95% | 98% \| 98% | Spinocerebellar ataxia 14, 605361 |
| PRKN | 100% \| 100% | 96% \| 96% | 99% \| 99% | Adenocarcinoma of lung, somatic, 211980;Parkinson disease 2, juvenile-onset, 600116;Ovarian cancer, somatic, 167000 |
| PRKRA | 100% \| 100% | 98% \| 98% | 100% \| 100% | Dystonia 16, 612067 |
| PRRT2 | 99% \| 99% | 95% \| 95% | 97% \| 97% | Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066;Seizures, benign familial infantile, 2, 605751;Episodic kinesigenic dyskinesia 1, 128200 |
| PSAP | 94% \| 94% | 97% \| 97% | 99% \| 99% | Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, susceptibility to}, 619491 |
| PTRH2 | 100% \| 100% | 99% \| 99% | 100% \| 100% | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 |
| PTS | 100% \| 100% | 97% \| 97% | 99% \| 99% | Hyperphenylalaninemia, BH4-deficient, A, 261640 |
| PUM1 | 93% \| 93% | 97% \| 97% | 99% \| 99% | Neurodevelopmental disorder with motor abnormalities, and facial dysmorphism, 620719 |
| PYCR2 | 100% \| 100% | 97% \| 97% | 98% \| 98% | Leukodystrophy, hypomyelinating, 10, 616420 |

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|---|---|---|---|---|
| QDPR | 94% \| 94% | 97% \| 97% | 99% \| 99% | Hyperphenylalaninemia, BH4-deficient, C, 261630 |
| RAB18 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Warburg micro syndrome 3, 614222 |
| RAB3GAP1 | 96% \| 96% | 97% \| 97% | 100% \| 100% | Martsolf syndrome 2, 619420;Warburg micro syndrome 1, 600118 |
| RAB3GAP2 | 94% \| 94% | 97% \| 97% | 99% \| 99% | Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225 |
| RAD51 | 89% \| 89% | 95% \| 95% | 99% \| 99% | Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244 |
| RARS1 | 91% \| 91% | 94% \| 94% | 99% \| 99% | Leukodystrophy, hypomyelinating, 9, 616140 |
| RARS2 | 93% \| 93% | 97% \| 97% | 100% \| 100% | Pontocerebellar hypoplasia, type 6, 611523 |
| REEP1 | 87% \| 87% | 97% \| 97% | 99% \| 99% | Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011;Spastic paraplegia 31, autosomal dominant, 610250;?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751 |
| REEP2 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Spastic paraplegia 72A, autosomal dominant, 615625;?Spastic paraplegia 72B, autosomal recessive, 620606 |
| RFC1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575 |
| RHOBTB2 | 99% \| 99% | 96% \| 96% | 99% \| 99% | Developmental and epileptic encephalopathy 64, 618004 |
| RNASEH2A | 100% \| 100% | 96% \| 96% | 97% \| 95% | Aicardi-Goutieres syndrome 4, 610333 |
| RNASEH2B | 91% \| 91% | 97% \| 97% | 99% \| 99% | Aicardi-Goutieres syndrome 2, 610181 |
| RNASEH2C | 100% \| 100% | 95% \| 95% | 98% \| 98% | Aicardi-Goutieres syndrome 3, 610329 |
| RNF170 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Ataxia, sensory, 1, autosomal dominant, 608984;Spastic paraplegia 85, autosomal recessive, 619686 |
| RNF216 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 |
| RORA | 100% \| 100% | 96% \| 96% | 99% \| 99% | Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 |
| RTN2 | 100% \| 100% | 97% \| 97% | 98% \| 98% | Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854;Spastic paraplegia 12, autosomal dominant, 604805 |
| RUBCN | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia, autosomal recessive 15, 615705 |
| SACS | 99% \| 99% | 98% \| 98% | 99% \| 99% | Spastic ataxia, Charlevoix-Saguenay type, 270550 |
| SAMD9L | 100% \| 100% | 98% \| 98% | 99% \| 99% | Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 |
| SAMHD1 | 91% \| 90% | 98% \| 98% | 99% \| 99% | ?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952 |
| SCN11A | 99% \| 99% | 96% \| 96% | 99% \| 99% | Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548 |

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|---|---|---|---|---|
| SCN1A | 100% \| 100% | 97% \| 97% | 100% \| 100% | Developmental and epileptic encephalopathy 6B, non-Dravet, 619317;Migraine, familial hemiplegic, 3, 609634;Dravet syndrome, 607208;Febrile seizures, familial, 3A, 604403;Generalized epilepsy with febrile seizures plus, type 2, 604403 |
| SCN2A | 100% \| 100% | 98% \| 98% | 100% \| 100% | Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924 |
| SCN8A | 100% \| 100% | 96% \| 96% | 99% \| 99% | ?Myoclonus, familial, 2, 618364;Seizures, benign familial infantile, 5, 617080;Cognitive impairment with or without cerebellar ataxia, 614306;Developmental and epileptic encephalopathy 13, 614558 |
| SCYL1 | 100% \| 100% | 96% \| 96% | 97% \| 97% | Spinocerebellar ataxia, autosomal recessive 21, 616719 |
| SELENOI | 100% \| 100% | 98% \| 98% | 99% \| 99% | Spastic paraplegia 81, autosomal recessive, 618768 |
| SEPSECS | 93% \| 93% | 97% \| 97% | 100% \| 100% | Pontocerebellar hypoplasia type 2D, 613811 |
| SERAC1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 |
| SETX | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433 |
| SGCE | 90% \| 90% | 98% \| 98% | 99% \| 99% | Dystonia-11, myoclonic, 159900 |
| SHQ1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Dystonia 35, childhood-onset, 619921;Neurodevelopmental disorder with dystonia and seizures, 619922 |
| SIL1 | 91% \| 91% | 97% \| 97% | 100% \| 100% | Marinesco-Sjogren syndrome, 248800 |
| SLC12A6 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068 |
| SLC13A3 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 |
| SLC16A2 | 88% \| 88% | 99% \| 99% | 100% \| 100% | Allan-Herndon-Dudley syndrome, 300523 |
| SLC18A2 | 98% \| 96% | 97% \| 97% | 99% \| 99% | Parkinsonism-dystonia, infantile, 2, 618049 |
| SLC19A3 | 99% \| 99% | 97% \| 97% | 99% \| 99% | Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483 |
| SLC1A3 | 94% \| 94% | 97% \| 97% | 99% \| 99% | Episodic ataxia, type 6, 612656 |
| SLC1A4 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 |
| SLC20A2 | 98% \| 98% | 97% \| 97% | 99% \| 99% | Basal ganglia calcification, idiopathic, 1, 213600 |
| SLC25A15 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 |
| SLC25A46 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303 |

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|---|---|---|---|---|
| SLC2A1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Dystonia 9, 601042;GLUT1 deficiency syndrome 1, infantile onset, severe, 606777;Stomatin-deficient cryohydrocytosis with neurologic defects, 608885;{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847;GLUT1 deficiency syndrome 2, childhood onset, 612126 |
| SLC30A10 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Hypermanganesemia with dystonia 1, 613280 |
| SLC33A1 | 93% \| 93% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482 |
| SLC39A14 | 93% \| 93% | 97% \| 97% | 98% \| 98% | ?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013 |
| SLC52A2 | 100% \| 100% | 97% \| 97% | 97% \| 95% | Brown-Vialetto-Van Laere syndrome 2, 614707 |
| SLC52A3 | 99% \| 99% | 96% \| 96% | 99% \| 99% | ?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530 |
| SLC6A3 | 93% \| 93% | 96% \| 96% | 98% \| 98% | Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890 |
| SLC6A5 | 99% \| 99% | 96% \| 96% | 99% \| 99% | Hyperekplexia 3, 614618 |
| SLC9A1 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Lichtenstein-Knorr syndrome, 616291 |
| SMDT1 | 98% \| 98% | 97% \| 97% | 99% \| 99% | |
| SMPD1 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200 |
| SNAPC4 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515 |
| SNORD118 | | | | Leukoencephalopathy, brain calcifications, and cysts, 614561 |
| SNX14 | 95% \| 95% | 98% \| 98% | 99% \| 99% | Spinocerebellar ataxia, autosomal recessive 20, 616354 |
| SOD1 | 96% \| 96% | 98% \| 98% | 99% \| 99% | Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400 |
| SOX10 | 98% \| 98% | 95% \| 95% | 99% \| 99% | Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 |
| SPART | 96% \| 96% | 98% \| 98% | 99% \| 99% | Troyer syndrome, 275900 |
| SPAST | 94% \| 94% | 96% \| 96% | 99% \| 99% | Spastic paraplegia 4, autosomal dominant, 182601 |
| SPG11 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360 |
| SPG21 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Mast syndrome, 248900 |
| SPG7 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Spastic paraplegia 7, autosomal recessive, 607259 |
| SPR | 100% \| 100% | 97% \| 97% | 99% \| 99% | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 |

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| SPTAN1 | 99% \| 99% | 97% \| 97% | 99% \| 99% | 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 |
| SPTBN2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386 |
| SQSTM1 | 100% \| 100% | 97% \| 97% | 98% \| 98% | 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 |
| STAMBP | 97% \| 97% | 98% \| 98% | 100% \| 100% | Microcephaly-capillary malformation syndrome, 614261 |
| STUB1 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Spinocerebellar ataxia 48, 618093;Spinocerebellar ataxia, autosomal recessive 16, 615768 |
| SUCLA2 | 96% \| 96% | 97% \| 97% | 99% \| 99% | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 |
| SUFU | 100% \| 100% | 96% \| 96% | 99% \| 99% | {Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255 |
| SUMF1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Multiple sulfatase deficiency, 272200 |
| SUOX | 100% \| 100% | 96% \| 96% | 99% \| 99% | Sulfite oxidase deficiency, 272300 |
| SYNE1 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Arthrogryposis multiplex congenita 3, myogenic type, 618484;Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998;Spinocerebellar ataxia, autosomal recessive 8, 610743 |
| TACO1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Mitochondrial complex IV deficiency, nuclear type 8, 619052 |
| TANGO2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 |
| TBC1D20 | 93% \| 93% | 97% \| 97% | 99% \| 99% | Warburg micro syndrome 4, 615663 |
| TBC1D23 | 96% \| 96% | 98% \| 98% | 99% \| 99% | Pontocerebellar hypoplasia, type 11, 617695 |
| TBCD | 89% \| 89% | 96% \| 96% | 99% \| 99% | Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 |
| TDP1 | 88% \| 88% | 98% \| 98% | 100% \| 100% | ?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250 |
| TDP2 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spinocerebellar ataxia, autosomal recessive 23, 616949 |
| TECPR2 | 100% \| 100% | 96% \| 96% | 99% \| 99% | Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 |
| TENM4 | 99% \| 99% | 96% \| 96% | 99% \| 99% | Essential tremor, hereditary, 5, 616736 |
| TFG | 100% \| 100% | 97% \| 97% | 100% \| 100% | ?Spastic paraplegia 57, autosomal recessive, 615658;Hereditary motor and sensory neuropathy, Okinawa type, 604484 |

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|---|---|---|---|---|
| TH | 99% \| 99% | 96% \| 96% | 97% \| 96% | Segawa syndrome, recessive, 605407 |
| THAP1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Dystonia 6, torsion, 602629 |
| TIMM8A | 96% \| 96% | 98% \| 98% | 99% \| 99% | Mohr-Tranebjaerg syndrome, 304700 |
| TMEM106B | 100% \| 100% | 98% \| 98% | 100% \| 100% | Leukodystrophy, hypomyelinating, 16, 617964 |
| TMEM240 | 100% \| 100% | 93% \| 93% | 98% \| 98% | Spinocerebellar ataxia 21, 607454 |
| TMEM67 | 96% \| 96% | 97% \| 97% | 99% \| 99% | Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360 |
| TNR | 100% \| 100% | 97% \| 97% | 99% \| 99% | Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653 |
| TOE1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Pontocerebellar hypoplasia, type 7, 614969 |
| TOR1A | 91% \| 91% | 97% \| 97% | 99% \| 99% | {Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100 |
| TPP1 | 100% \| 100% | 97% \| 96% | 99% \| 99% | Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270 |
| TREM2 | 100% \| 100% | 98% \| 98% | 99% \| 99% | {Alzheimer disease 17, susceptibility to}, 615080;Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 |
| TREX1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448 |
| TRIT1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Combined oxidative phosphorylation deficiency 35, 617873 |
| TRPM3 | 98% \| 98% | 97% \| 97% | 99% \| 99% | ?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224 |
| TSEN15 | 100% \| 100% | 95% \| 95% | 99% \| 99% | Pontocerebellar hypoplasia, type 2F, 617026 |
| TSEN2 | 87% \| 87% | 97% \| 97% | 99% \| 99% | Pontocerebellar hypoplasia type 2B, 612389 |
| TSEN54 | 98% \| 98% | 96% \| 96% | 97% \| 97% | Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204 |
| TSPOAP1 | 100% \| 100% | 96% \| 96% | 98% \| 98% | Dystonia 22, juvenile-onset, 620453;?Dystonia 22, adult-onset, 620456 |
| TTBK2 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Spinocerebellar ataxia 11, 604432 |
| TTC19 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Mitochondrial complex III deficiency, nuclear type 2, 615157 |
| TTPA | 88% \| 88% | 95% \| 95% | 100% \| 100% | Ataxia with isolated vitamin E deficiency, 277460 |
| TUBA1A | 100% \| 100% | 97% \| 97% | 99% \| 99% | Cortical dysplasia, complex, with other brain malformations 16, 611603 |

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| TUBA4A | 100% \| 100% | 97% \| 97% | 99% \| 99% | Oocyte/zygote/embryo maturation arrest 23, 621231;Spastic ataxia 11, autosomal dominant, 621226;Frontotemporal dementia and/or amyotrophic lateral sclerosis 9, 616208;Congenital myopathy 26, 621225 |
| TUBB | 100% \| 100% | 98% \| 98% | 100% \| 100% | Symmetric circumferential skin creases, congenital, 1, 156610;Cortical dysplasia, complex, with other brain malformations 6, 615771 |
| TUBB4A | 97% \| 97% | 96% \| 96% | 98% \| 96% | Dystonia 4, torsion, autosomal dominant, 128101;Leukodystrophy, hypomyelinating, 6, 612438 |
| TUBG1 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Cortical dysplasia, complex, with other brain malformations 4, 615412 |
| TWNK | 100% \| 100% | 97% \| 97% | 99% \| 99% | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138 |
| TYROBP | 89% \| 89% | 95% \| 95% | 97% \| 92% | Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 |
| UBA5 | 90% \| 90% | 98% \| 98% | 99% \| 99% | ?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132 |
| UBAP1 | 97% \| 97% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 80, autosomal dominant, 618418 |
| UBTF | 100% \| 100% | 96% \| 96% | 98% \| 98% | Neurodegeneration, childhood-onset, with brain atrophy, 617672 |
| UCHL1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | {?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491 |
| VAC14 | 96% \| 96% | 97% \| 97% | 99% \| 99% | Striatonigral degeneration, childhood-onset, 617054 |
| VAMP1 | 100% \| 100% | 98% \| 98% | 99% \| 99% | Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600 |
| VARS2 | 99% \| 99% | 96% \| 96% | 99% \| 99% | Combined oxidative phosphorylation deficiency 20, 615917 |
| VCP | 99% \| 99% | 97% \| 97% | 100% \| 100% | 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 |
| VLDLR | 100% \| 100% | 98% \| 98% | 100% \| 100% | Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050 |
| VPS11 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683 |
| VPS13A | 100% \| 100% | 98% \| 98% | 99% \| 99% | Choreoacanthocytosis, 200150 |
| VPS13D | 100% \| 100% | 98% \| 98% | 100% \| 100% | Spinocerebellar ataxia, autosomal recessive 4, 607317 |
| VPS16 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Dystonia 30, 619291 |
| VPS37A | 92% \| 92% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 53, autosomal recessive, 614898 |
| VPS41 | 96% \| 96% | 98% \| 98% | 100% \| 100% | Spinocerebellar ataxia, autosomal recessive 29, 619389 |

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| VPS53 | 81% \| 81% | 96% \| 96% | 99% \| 99% | Pontocerebellar hypoplasia, type 2E, 615851 |
| VRK1 | 98% \| 98% | 98% \| 98% | 100% \| 100% | Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542 |
| WASHC5 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Ritscher-Schinzel syndrome 1, 220210;Spastic paraplegia 8, autosomal dominant, 603563 |
| WDR26 | 94% \| 94% | 97% \| 97% | 99% \| 99% | Skraban-Deardorff syndrome, 617616 |
| WDR45 | 97% \| 97% | 99% \| 99% | 100% \| 100% | Neurodegeneration with brain iron accumulation 5, 300894 |
| WDR73 | 99% \| 99% | 96% \| 96% | 99% \| 99% | Galloway-Mowat syndrome 1, 251300 |
| WDR81 | 99% \| 99% | 96% \| 96% | 97% \| 97% | Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185;Hydrocephalus, congenital, 3, with brain anomalies, 617967 |
| WFS1 | 89% \| 89% | 95% \| 95% | 99% \| 99% | Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;Wolfram syndrome 1, 222300 |
| WWOX | 99% \| 99% | 96% \| 96% | 99% \| 99% | Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322 |
| XK | 98% \| 98% | 99% \| 99% | 100% \| 100% | McLeod syndrome, 300842 |
| XPA | 100% \| 100% | 97% \| 97% | 99% \| 99% | Xeroderma pigmentosum, group A, 278700 |
| XPR1 | 100% \| 100% | 98% \| 98% | 100% \| 100% | Basal ganglia calcification, idiopathic, 6, 616413 |
| XRCC1 | 100% \| 100% | 94% \| 94% | 99% \| 99% | ?Spinocerebellar ataxia, autosomal recessive 26, 617633 |
| ZC4H2 | 99% \| 99% | 99% \| 99% | 100% \| 100% | Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041 |
| ZFYVE26 | 100% \| 100% | 97% \| 97% | 99% \| 99% | Spastic paraplegia 15, autosomal recessive, 270700 |