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# INTELLECTUAL DISABILITY / DEVELOPMENTAL DELAY PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.1.0 (2032 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 |
| AASS | 100% | 100% | 100% | 100% | 99.8% | Hyperlysinemia, 238700 |
| ABAT | 100% | 100% | 100% | 100% | 99.4% | GABA-transaminase deficiency, 613163 |
| ABCA2 | 100% | 100% | 100% | 99.9% | 98.7% | Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 |

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| ABCC8 | 100% | 100% | 100% | 100% | 99.5% | Diabetes mellitus, permanent neonatal 3, 618857;Maturity-onset diabetes of the young, type 12, 621196;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450 |
| ABCC9 | 96% | 96% | 100% | 100% | 99.8% | Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719 |
| ABCD1 | 89.3% | 86.3% | 98.7% | 89.2% | 70.9% | Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100 |
| ABCD4 | 100% | 99.6% | 100% | 99.9% | 99.3% | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 |
| ABHD16A | 97.5% | 97.5% | 100% | 100% | 99.4% | Spastic paraplegia 86, autosomal recessive, 619735 |
| ABHD5 | 100% | 100% | 100% | 100% | 99.9% | Chanarin-Dorfman syndrome, 275630 |
| ACAD9 | 100% | 100% | 100% | 100% | 99.7% | Mitochondrial complex I deficiency, nuclear type 20, 611126 |
| ACADS | 100% | 100% | 100% | 100% | 99.5% | Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 |

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| ACADSB | 100% | 100% | 100% | 99.9% | 99.7% | 2-methylbutyrylglycinuria, 610006 |
| ACAT1 | 100% | 100% | 100% | 100% | 99.6% | Alpha-methylacetoacetic aciduria, 203750 |
| ACBD6 | 79.5% | 79.5% | 100% | 99.9% | 99.6% | Neurodevelopmental disorder with progressive movement abnormalities, 620785 |
| ACER3 | 96.3% | 96.3% | 100% | 100% | 99.9% | ?Leukodystrophy, progressive, early childhood-onset, 617762 |
| ACO2 | 90.7% | 86.8% | 100% | 100% | 99.7% | Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559 |
| ACOX1 | 97.9% | 97.9% | 100% | 100% | 99.7% | Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470 |
| ACSF3 | 94.7% | 94.7% | 100% | 100% | 99.3% | Combined malonic and methylmalonic aciduria, 614265 |
| ACSL4 | 100% | 100% | 99.1% | 91.5% | 73.4% | Intellectual developmental disorder, X-linked 63, 300387 |
| ACTB | 100% | 100% | 100% | 99.9% | 99.4% | 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 |

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| ACTG1 | 100% | 100% | 100% | 99.9% | 98.9% | Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583 |
| ACTL6A | 100% | 100% | 100% | 100% | 99.9% | |
| ACTL6B | 95.9% | 94.6% | 100% | 100% | 99.4% | Developmental and epileptic encephalopathy 76, 618468;Intellectual developmental disorder with severe speech and ambulation defects, 618470 |
| ACVR1 | 100% | 100% | 100% | 100% | 99.7% | Fibrodysplasia ossificans progressiva, 135100 |
| ACY1 | 100% | 100% | 100% | 99.9% | 99.4% | Aminoacylase 1 deficiency, 609924 |
| ADAM22 | 98.7% | 98.7% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 61, 617933 |
| ADAR | 100% | 100% | 100% | 100% | 99.8% | Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010 |
| ADARB1 | 94.8% | 94.7% | 100% | 100% | 99.4% | Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 |
| ADAT3 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 |

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| ADCY5 | 97.5% | 96.9% | 100% | 99.9% | 99.1% | Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647 |
| ADD1 | 100% | 100% | 100% | 100% | 99.6% | {Hypertension, essential, salt-sensitive}, 145500 |
| ADD3 | 97.6% | 97.4% | 100% | 100% | 99.6% | Cerebral palsy, spastic quadriplegic, 3, 617008 |
| ADGRG1 | 100% | 100% | 100% | 100% | 99.3% | Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752;Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854 |
| ADGRL1 | 100% | 100% | 100% | 100% | 99.2% | Developmental delay, behavioral abnormalities, and neuropsychiatric disorders, 620065 |
| ADK | 88% | 88% | 100% | 100% | 99.8% | Hypermethioninemia due to adenosine kinase deficiency, 614300 |
| ADNP | 100% | 100% | 100% | 100% | 99.7% | Helsmoortel-van der Aa syndrome, 615873 |
| ADPRS | 100% | 100% | 100% | 100% | 99.4% | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 |
| ADSL | 99.5% | 97.5% | 100% | 100% | 99.7% | Adenylosuccinase deficiency, 103050 |

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| AFF2 | 100% | 100% | 99.4% | 90.5% | 71.8% | Intellectual developmental disorder, X-linked 109, 309548 |
| AFF3 | 100% | 100% | 100% | 100% | 99.4% | KINSSHIP syndrome, 619297 |
| AFF4 | 100% | 100% | 100% | 100% | 99.9% | CHOPS syndrome, 616368 |
| AFG2A | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 |
| AFG2B | 100% | 100% | 100% | 100% | 99.8% | Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616 |
| AFG3L2 | 94.2% | 92.2% | 100% | 100% | 99.5% | Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246 |
| AGA | 91.6% | 91.6% | 100% | 100% | 99.5% | Aspartylglucosaminuria, 208400 |
| AGAP1 | 100% | 100% | 100% | 99.8% | 98.5% | |
| AGMO | 100% | 100% | 100% | 100% | 99.7% | |
| AGO1 | 99.4% | 98.4% | 100% | 100% | 99.7% | Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292 |
| AGO2 | 100% | 100% | 100% | 100% | 99.6% | Lessel-Kreienkamp syndrome, 619149 |
| AGTPBP1 | 96.5% | 96.5% | 100% | 100% | 99.7% | Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 |

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| AHCY | 100% | 100% | 100% | 99.9% | 99.1% | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 |
| AHDC1 | 100% | 100% | 100% | 100% | 98.9% | Xia-Gibbs syndrome, 615829 |
| AHI1 | 98.7% | 98.7% | 100% | 99.9% | 99.6% | Joubert syndrome 3, 608629 |
| AHSG | 100% | 100% | 100% | 100% | 99.8% | ?Alopecia-intellectual disability syndrome 1, 203650 |
| AIFM1 | 100% | 100% | 99.3% | 90.3% | 72.6% | 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% | 100% | 100% | 99.9% | Leukodystrophy, hypomyelinating, 3, 260600 |
| AIMP2 | 100% | 100% | 100% | 99.8% | 99.1% | Leukodystrophy, hypomyelinating, 17, 618006 |
| AJAP1 | 100% | 100% | 100% | 100% | 99.1% | |
| AKT3 | 94.6% | 94.6% | 100% | 100% | 99.7% | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 |
| ALDH18A1 | 100% | 100% | 100% | 100% | 99.6% | 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 |

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| ALDH3A2 | 93.5% | 93.5% | 100% | 100% | 99.7% | Sjogren-Larsson syndrome, 270200 |
| ALDH4A1 | 97.9% | 96.1% | 100% | 100% | 99.5% | Hyperprolinemia, type II, 239510 |
| ALDH5A1 | 100% | 100% | 100% | 100% | 99.5% | Succinic semialdehyde dehydrogenase deficiency, 271980 |
| ALDH7A1 | 97.2% | 97.2% | 100% | 100% | 99.8% | Epilepsy, early-onset, 4, vitamin B6-dependent, 266100 |
| ALG1 | 100% | 100% | 100% | 100% | 99.7% | Congenital disorder of glycosylation, type Ik, 608540 |
| ALG11 | 91% | 91% | 100% | 100% | 99.9% | Congenital disorder of glycosylation, type Ip, 613661 |
| ALG12 | 100% | 100% | 100% | 100% | 99.6% | Congenital disorder of glycosylation, type Ig, 607143 |
| ALG13 | 100% | 100% | 99.2% | 90.7% | 72.3% | Developmental and epileptic encephalopathy 36, 300884 |
| ALG14 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031;Myopathy, epilepsy, and progressive cerebral atrophy, 619036;?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 |
| ALG2 | 100% | 100% | 100% | 100% | 99.8% | Congenital disorder of glycosylation, type Ii, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 |

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| ALG3 | 100% | 100% | 100% | 100% | 99.4% | Congenital disorder of glycosylation, type Id, 601110 |
| ALG6 | 100% | 100% | 100% | 100% | 99.9% | Congenital disorder of glycosylation, type Ic, 603147 |
| ALG8 | 78.4% | 77.9% | 100% | 100% | 99.7% | Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874 |
| ALG9 | 100% | 100% | 100% | 99.9% | 99.5% | Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776 |
| ALKBH8 | 96.3% | 96.3% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal recessive 71, 618504 |
| ALMS1 | 100% | 100% | 100% | 100% | 99.8% | Alstrom syndrome, 203800 |
| ALX3 | 100% | 100% | 100% | 99.5% | 98.7% | Frontonasal dysplasia 1, 136760 |
| ALX4 | 100% | 100% | 100% | 100% | 99.4% | Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451 |
| AMER1 | 100% | 100% | 98.9% | 90.5% | 71.9% | Osteopathia striata with cranial sclerosis, 300373 |
| AMFR | 100% | 100% | 100% | 100% | 99.6% | Spastic paraplegia 89, autosomal recessive, 620379 |
| AMMECR1 | 99.9% | 97.7% | 99.1% | 88.8% | 71.2% | Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 |
| AMOTL1 | 100% | 100% | 100% | 100% | 99.5% | Craniofaciocardiohepatic syndrome, 621192 |

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| AMPD2 | 100% | 100% | 100% | 99.9% | 99.6% | Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686 |
| AMT | 100% | 100% | 100% | 99.9% | 99.3% | Glycine encephalopathy 2, 620398 |
| ANK2 | 100% | 100% | 100% | 100% | 99.7% | Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919 |
| ANK3 | 99.7% | 99.7% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 37, 615493 |
| ANKH | 100% | 100% | 100% | 100% | 99.4% | Chondrocalcinosis 2, 118600;Craniometaphyseal dysplasia, 123000 |
| ANKLE2 | 100% | 100% | 100% | 99.8% | 99.1% | Microcephaly 16, primary, autosomal recessive, 616681 |
| ANKRD11 | 100% | 100% | 100% | 100% | 99.2% | KBG syndrome, 148050 |
| ANKRD17 | 100% | 100% | 100% | 100% | 99.6% | Chopra-Amiel-Gordon syndrome, 619504 |
| ANKS1B | 100% | 100% | 100% | 100% | 99.8% | |
| ANO10 | 94.9% | 94.9% | 100% | 100% | 100% | Spinocerebellar ataxia, autosomal recessive 10, 613728 |
| ANO4 | 100% | 100% | 100% | 100% | 99.7% | |
| ANTXR1 | 100% | 100% | 100% | 99.9% | 99.4% | GAPO syndrome, 230740;{?Hemangioma, capillary infantile, susceptibility to}, 602089 |
| AP1B1 | 93.7% | 91.9% | 100% | 100% | 99.6% | Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150 |

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| AP1G1 | 100% | 100% | 100% | 100% | 99.7% | Usmani-Riazuddin syndrome, autosomal recessive, 619548;Usmani-Riazuddin syndrome, autosomal dominant, 619467 |
| AP1S1 | 100% | 100% | 100% | 100% | 99.2% | MEDNIK syndrome, 609313 |
| AP1S2 | 100% | 100% | 99.3% | 92.3% | 76.1% | Pettigrew syndrome, 304340 |
| AP2M1 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 60, with seizures, 618587 |
| AP2S1 | 82.5% | 82.5% | 100% | 99.9% | 98.8% | Hypocalciuric hypercalcemia, type III, 600740 |
| AP3B1 | 100% | 100% | 100% | 100% | 99.8% | Hermansky-Pudlak syndrome 2, 608233 |
| AP3B2 | 100% | 100% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 48, 617276 |
| AP3D1 | 100% | 100% | 100% | 100% | 99.7% | ?Hermansky-Pudlak syndrome 10, 617050 |
| AP4B1 | 100% | 100% | 100% | 100% | 99.8% | Spastic paraplegia 47, autosomal recessive, 614066 |
| AP4E1 | 100% | 100% | 100% | 100% | 99.9% | Stuttering, familial persistent, 1, 184450;Spastic paraplegia 51, autosomal recessive, 613744 |
| AP4M1 | 100% | 100% | 100% | 100% | 99.7% | Spastic paraplegia 50, autosomal recessive, 612936 |
| AP4S1 | 79.1% | 79.1% | 100% | 99.9% | 99.4% | Spastic paraplegia 52, autosomal recessive, 614067 |

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| APC2 | 100% | 100% | 100% | 100% | 99% | Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169 |
| APTX | 100% | 100% | 100% | 100% | 99.7% | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 |
| ARCN1 | 96.3% | 96.1% | 100% | 100% | 99.7% | Short stature-micrognathia syndrome, 617164 |
| ARF1 | 100% | 100% | 100% | 100% | 99.8% | Periventricular nodular heterotopia 8, 618185 |
| ARF3 | 100% | 100% | 100% | 100% | 99.1% | |
| ARFGEF1 | 100% | 100% | 100% | 100% | 99.9% | Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964 |
| ARFGEF2 | 100% | 100% | 100% | 100% | 99.5% | Periventricular heterotopia with microcephaly, 608097 |
| ARG1 | 93% | 93% | 100% | 100% | 99.8% | Argininemia, 207800 |
| ARHGAP31 | 100% | 100% | 100% | 100% | 99.6% | Adams-Oliver syndrome 1, 100300 |
| ARHGAP35 | 100% | 100% | 100% | 100% | 99.5% | |
| ARHGEF6 | 93.6% | 93.6% | 99.2% | 90.8% | 73.8% | |
| ARHGEF9 | 96.9% | 96.9% | 99.3% | 90.1% | 70.9% | Developmental and epileptic encephalopathy 8, 300607 |
| ARID1A | 100% | 100% | 100% | 99.9% | 98.9% | Coffin-Siris syndrome 2, 614607 |
| ARID1B | 98.7% | 98.4% | 100% | 99.5% | 96.7% | Coffin-Siris syndrome 1, 135900 |
| ARID2 | 100% | 100% | 100% | 100% | 99.7% | Coffin-Siris syndrome 6, 617808 |

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| ARL13B | 93.4% | 93.3% | 100% | 100% | 99.7% | Joubert syndrome 8, 612291 |
| ARL6 | 100% | 100% | 100% | 100% | 99.8% | Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151 |
| ARMC9 | 96.3% | 94.2% | 100% | 100% | 99.3% | Joubert syndrome 30, 617622 |
| ARPC4 | 100% | 100% | 100% | 100% | 99.9% | Developmental delay, language impairment, and ocular abnormalities, 620141 |
| ARSA | 100% | 100% | 100% | 99.9% | 99% | Metachromatic leukodystrophy, 250100 |
| ARSL | 100% | 100% | 98.7% | 90.2% | 71.8% | Chondrodysplasia punctata, X-linked recessive, 302950 |
| ARV1 | 87.3% | 86.5% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 38, 617020 |
| ARX | 99.9% | 98.4% | 96.3% | 80.9% | 59.5% | 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 |
| ASAH1 | 100% | 100% | 100% | 99.9% | 99.3% | Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000 |

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| ASCC3 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 81, 620700 |
| ASH1L | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal dominant 52, 617796 |
| ASL | 100% | 100% | 100% | 100% | 99.1% | Argininosuccinic aciduria, 207900 |
| ASNS | 100% | 100% | 100% | 100% | 99.8% | Asparagine synthetase deficiency, 615574 |
| ASPA | 100% | 100% | 100% | 100% | 99.8% | Canavan disease, 271900 |
| ASPM | 97.9% | 97.9% | 100% | 100% | 99.7% | Microcephaly 5, primary, autosomal recessive, 608716 |
| ASS1 | 92.3% | 90% | 100% | 100% | 99.6% | Citrullinemia, 215700 |
| ASTN1 | 96.6% | 96.6% | 100% | 100% | 99.5% | |
| ASXL1 | 100% | 100% | 100% | 100% | 99.8% | Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039 |
| ASXL2 | 100% | 100% | 100% | 100% | 99.6% | Shashi-Pena syndrome, 617190 |
| ASXL3 | 100% | 100% | 100% | 100% | 99.6% | Bainbridge-Ropers syndrome, 615485 |
| ATAD1 | 96.8% | 96.8% | 100% | 100% | 100% | Hyperekplexia 4, 618011 |
| ATAD3A | 100% | 100% | 100% | 99.9% | 98.7% | Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 |
| ATCAY | 100% | 100% | 100% | 100% | 99.2% | Ataxia, cerebellar, Cayman type, 601238 |
| ATG4D | 100% | 100% | 100% | 100% | 99.2% | Spermatogenic failure 101, 621269 |

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| ATG7 | 100% | 100% | 100% | 100% | 99.8% | Spinocerebellar ataxia, autosomal recessive 31, 619422 |
| ATIC | 100% | 100% | 100% | 100% | 99.9% | AICA-ribosiduria due to ATIC deficiency, 608688 |
| ATL1 | 100% | 100% | 100% | 100% | 99.7% | Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708 |
| ATN1 | 98.2% | 98.2% | 100% | 100% | 98.7% | Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 |
| ATOH1 | 100% | 100% | 100% | 100% | 99.6% | ?Deafness, autosomal dominant 89, 620284 |
| 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 |
| ATP1A2 | 100% | 100% | 100% | 99.9% | 99.4% | 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 |

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| ATP1A3 | 100% | 100% | 100% | 100% | 99.2% | Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606 |
| ATP2A2 | 100% | 100% | 100% | 100% | 99.6% | Acrokeratosis verruciformis, 101900;Darier disease, 124200;{Rhabdomyolysis, susceptibility to, 2}, 621236 |
| ATP2B1 | 100% | 100% | 100% | 99.9% | 99.6% | Intellectual developmental disorder, autosomal dominant 66, 619910 |
| ATP2B2 | 100% | 100% | 100% | 100% | 99.2% | Deafness, autosomal dominant 82, 619804 |
| ATP5F1A | 100% | 100% | 100% | 99.9% | 99.7% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358;?Combined oxidative phosphorylation deficiency 22, 616045;?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228 |
| ATP5PO | 100% | 100% | 100% | 100% | 99.7% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359 |
| ATP6AP1 | 99.6% | 97.5% | 98.9% | 89.5% | 70.5% | Immunodeficiency 47, 300972 |

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|---|---|---|---|---|---|---|
| ATP6AP2 | 100% | 100% | 99.3% | 91.2% | 74.9% | Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045 |
| ATP6V0A1 | 92.9% | 92.9% | 100% | 100% | 99.7% | Neurodevelopmental disorder with epilepsy and brain atrophy, 619971;Developmental and epileptic encephalopathy 104, 619970 |
| ATP6V0A2 | 100% | 100% | 100% | 100% | 99.6% | Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200 |
| ATP6V0C | 100% | 100% | 100% | 100% | 99.8% | Epilepsy, early-onset, 3, with or without developmental delay, 620465 |
| ATP6V1A | 100% | 100% | 100% | 100% | 99.8% | Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012 |
| ATP6V1B2 | 94.3% | 94.3% | 100% | 100% | 99.8% | Zimmermann-Laband syndrome 2, 616455;Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 |
| ATP7A | 95% | 94.8% | 99.4% | 92.2% | 74.7% | Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400 |

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| ATP8A2 | 100% | 100% | 100% | 100% | 99.6% | Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268 |
| ATP9A | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242 |
| ATR | 100% | 100% | 99.9% | 99.7% | 99.2% | Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564 |
| ATRX | 100% | 100% | 99.1% | 91.6% | 74.3% | Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040 |
| ATXN2L | 100% | 100% | 100% | 99.8% | 99.2% | |
| ATXN7L3 | 100% | 100% | 100% | 100% | 99.6% | Harel-Tora neurodevelopmental syndrome, 621377 |
| AUH | 100% | 100% | 100% | 100% | 99.7% | 3-methylglutaconic aciduria, type I, 250950 |
| AUTS2 | 100% | 100% | 100% | 100% | 99.4% | Intellectual developmental disorder, autosomal dominant 26, 615834 |
| AVPR2 | 100% | 100% | 98.8% | 89% | 71.9% | Diabetes insipidus, nephrogenic, 1, 304800;Nephrogenic syndrome of inappropriate antidiuresis, 300539 |

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| AXIN1 | 100% | 100% | 100% | 100% | 99.5% | Hepatocellular carcinoma, somatic, 114550;Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558;?Caudal duplication anomaly, 607864 |
| B3GALNT2 | 92.6% | 92.6% | 100% | 100% | 99.7% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 |
| B3GALT6 | 100% | 98.6% | 100% | 99.9% | 98.6% | Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465 |
| B3GLCT | 100% | 100% | 100% | 100% | 99.9% | Peters-plus syndrome, 261540 |
| B4GALNT1 | 100% | 100% | 100% | 100% | 99.5% | Spastic paraplegia 26, autosomal recessive, 609195 |
| B4GALT1 | 91% | 88.3% | 100% | 100% | 99.6% | Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091 |
| B4GALT7 | 86.8% | 86.8% | 100% | 100% | 99.9% | Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 |
| B4GAT1 | 100% | 100% | 100% | 100% | 99.3% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 |

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| B9D1 | 100% | 100% | 100% | 100% | 99.4% | ?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120 |
| B9D2 | 100% | 100% | 100% | 100% | 99.7% | ?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175 |
| BAIAP2 | 100% | 100% | 100% | 99.9% | 99.3% | Developmental and epileptic encephalopathy 120, 621468 |
| BAP1 | 100% | 100% | 100% | 100% | 99.5% | Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661 |
| BAZ2B | 100% | 100% | 100% | 99.9% | 99.3% | |
| BBS1 | 93.7% | 93.7% | 100% | 100% | 99.3% | Bardet-Biedl syndrome 1, 209900 |
| BBS10 | 100% | 100% | 100% | 100% | 99.8% | Bardet-Biedl syndrome 10, 615987 |
| BBS12 | 100% | 100% | 100% | 100% | 100% | Bardet-Biedl syndrome 12, 615989 |
| BBS2 | 98% | 98% | 100% | 100% | 99.8% | Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981 |
| BBS4 | 100% | 100% | 100% | 100% | 99.7% | Bardet-Biedl syndrome 4, 615982 |
| BBS5 | 100% | 100% | 100% | 100% | 99.9% | Bardet-Biedl syndrome 5, 615983 |
| BBS7 | 100% | 100% | 100% | 100% | 99.9% | Bardet-Biedl syndrome 7, 615984 |
| BBS9 | 91.9% | 91.9% | 100% | 100% | 99.7% | Bardet-Biedl syndrome 9, 615986 |
| BCAP31 | 91.5% | 86.7% | 98.7% | 88.7% | 70.4% | Deafness, dystonia, and cerebral hypomyelination, 300475 |

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| BCAS3 | 100% | 100% | 100% | 100% | 99.8% | Hengel-Maroofian-Schols syndrome, 619641 |
| BCAT2 | 100% | 100% | 100% | 100% | 99.4% | Hypervalinemia and hyperleucine-isoleucinemia, 618850 |
| BCKDHA | 86.3% | 86.3% | 100% | 99.9% | 99.3% | Maple syrup urine disease, type Ia, 248600 |
| BCKDHB | 89.9% | 87.7% | 100% | 99.9% | 99.3% | Maple syrup urine disease, type Ib, 620698 |
| BCKDK | 100% | 100% | 100% | 100% | 99.6% | Branched-chain keto acid dehydrogenase kinase deficiency, 614923 |
| BCL11A | 100% | 100% | 100% | 99.9% | 98.9% | Dias-Logan syndrome, 617101 |
| BCL11B | 100% | 99.8% | 100% | 99.9% | 98% | Immunodeficiency 49, severe combined, 617237;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 |
| BCOR | 100% | 100% | 98.9% | 89.4% | 69.8% | Microphthalmia, syndromic 2, 300166 |
| BCORL1 | 100% | 100% | 98.5% | 87.1% | 67.3% | Shukla-Vernon syndrome, 301029 |
| BCS1L | 100% | 100% | 100% | 100% | 99.6% | GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000 |
| BHLHE22 | 100% | 100% | 100% | 99.8% | 97.3% | |

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| 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 |
| BICRA | 100% | 100% | 100% | 99.8% | 98.4% | Coffin-Siris syndrome 12, 619325 |
| BLM | 94.7% | 94.6% | 100% | 100% | 99.9% | Bloom syndrome, 210900 |
| BLOC1S1 | 100% | 100% | 100% | 100% | 99.1% | |
| BLTP1 | 100% | 100% | 100% | 100% | 99.8% | Alkuraya-Kucinskas syndrome, 617822 |
| BMP4 | 100% | 100% | 100% | 100% | 99.5% | Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932 |
| BOLA3 | 100% | 100% | 100% | 100% | 98.9% | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 |
| BORCS5 | 89.3% | 89.3% | 100% | 100% | 99.5% | |
| BORCS8 | 84.1% | 84.1% | 100% | 99.7% | 98.8% | Neurodegeneration, infantile-onset, with optic atrophy and brain abnormalities, 620987 |
| BPTF | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755;{Kaposi sarcoma, susceptibility to}, 148000 |

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| BRAF | 100% | 100% | 100% | 99.8% | 98.2% | Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980 |
| BRAT1 | 97.3% | 96.5% | 100% | 99.9% | 99.3% | Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 |
| BRD4 | 100% | 100% | 100% | 99.9% | 98.7% | Cornelia de Lange syndrome 6, 620568 |
| BRF1 | 100% | 100% | 100% | 100% | 99.5% | Cerebellofaciodental syndrome, 616202 |
| BRPF1 | 100% | 100% | 100% | 100% | 99.6% | Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 |
| BRSK2 | 100% | 100% | 100% | 99.9% | 99.1% | |
| BRWD3 | 100% | 100% | 99.3% | 91.7% | 73.7% | Intellectual developmental disorder, X-linked 93, 300659 |

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| 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 |
| BSN | 100% | 99.9% | 100% | 99.9% | 99.3% | |
| BTD | 94.2% | 94.2% | 100% | 100% | 99.9% | Biotinidase deficiency, 253260 |
| BUB1 | 97.3% | 97.3% | 100% | 100% | 99.7% | Colorectal cancer with chromosomal instability, somatic, 114500;Microcephaly 30, primary, autosomal recessive, 620183 |
| BUB1B | 96.3% | 96.3% | 100% | 100% | 99.8% | Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300 |
| C12orf57 | 100% | 100% | 100% | 99.8% | 98.3% | Temtamy syndrome, 218340 |
| C2CD3 | 96% | 96% | 100% | 100% | 99.5% | Orofaciodigital syndrome XIV, 615948 |
| C2orf69 | 100% | 100% | 100% | 100% | 99.9% | Combined oxidative phosphorylation deficiency 53, 619423 |
| CA2 | 100% | 100% | 100% | 100% | 99.8% | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 |
| CA5A | 84.7% | 84.7% | 100% | 100% | 99.6% | Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 |

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| CA8 | 100% | 100% | 100% | 100% | 99.9% | Spinocerebellar ataxia, autosomal recessive 34, 613227 |
| CACHD1 | 100% | 100% | 100% | 100% | 99.7% | Davis-Wells syndrome, 621566 |
| CACNA1A | 100% | 100% | 100% | 99.9% | 99.1% | Spinocerebellar ataxia 6, 183086;Episodic ataxia, type 2, 108500;Developmental and epileptic encephalopathy 42, 617106;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500;Migraine, familial hemiplegic, 1, 141500 |
| CACNA1B | 97.2% | 97.2% | 100% | 99.9% | 99% | Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 |
| CACNA1C | 100% | 100% | 100% | 100% | 99.5% | Timothy syndrome, 601005;Long QT syndrome 8, 618447;Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029;Brugada syndrome 3, 611875 |
| CACNA1D | 100% | 100% | 100% | 100% | 99.7% | Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896 |
| CACNA1E | 100% | 100% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 69, 618285 |

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| CACNA1G | 100% | 100% | 100% | 99.9% | 99.4% | Spinocerebellar ataxia 42, 616795;Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 |
| CACNA1I | 100% | 100% | 100% | 99.8% | 98.5% | Neurodevelopmental disorder with speech impairment and with or without seizures, 620114 |
| CACNA2D1 | 100% | 100% | 100% | 100% | 99.4% | Developmental and epileptic encephalopathy 110, 620149 |
| CACNA2D2 | 100% | 100% | 100% | 100% | 99.3% | Cerebellar atrophy with seizures and variable developmental delay, 618501 |
| CAD | 99.7% | 98.5% | 100% | 100% | 99.4% | Developmental and epileptic encephalopathy 50, 616457 |
| CAMK2A | 100% | 100% | 100% | 100% | 99.5% | Intellectual developmental disorder, autosomal dominant 53, 617798;?Intellectual developmental disorder, autosomal recessive 63, 618095 |
| CAMK2B | 100% | 100% | 100% | 99.9% | 99% | Intellectual developmental disorder, autosomal dominant 54, 617799 |
| CAMK2D | 97.2% | 97.2% | 100% | 100% | 99.6% | |
| CAMK2G | 99.7% | 98.1% | 100% | 100% | 99.6% | Intellectual developmental disorder, autosomal dominant 59, 618522 |
| CAMK4 | 100% | 100% | 100% | 100% | 99.7% | |

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| CAMSAP1 | 100% | 100% | 100% | 99.9% | 99.3% | Cortical dysplasia, complex, with other brain malformations 12, 620316 |
| CAMTA1 | 100% | 100% | 100% | 100% | 99.2% | Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 |
| CANT1 | 100% | 100% | 100% | 100% | 99.7% | Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719 |
| CAPN15 | 100% | 100% | 100% | 99.9% | 98.9% | Oculogastrointestinal neurodevelopmental syndrome, 619318 |
| CAPRIN1 | 100% | 100% | 100% | 100% | 99.5% | Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782;Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636 |
| CAPZA2 | 100% | 100% | 100% | 100% | 99.6% | |
| CARS1 | 100% | 100% | 100% | 100% | 99.6% | Microcephaly, developmental delay, and brittle hair syndrome, 618891 |
| CARS2 | 100% | 99.9% | 100% | 100% | 99.2% | Combined oxidative phosphorylation deficiency 27, 616672 |
| CASK | 100% | 100% | 99.2% | 91.1% | 73.2% | Intellectual developmental disorder, with or without nystagmus, 300422;Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749;FG syndrome 4, 300422 |

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| CASP2 | 100% | 99.9% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, 620653 |
| CBL | 100% | 100% | 100% | 100% | 99.7% | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785 |
| CBS | 87.7% | 85.5% | 100% | 100% | 99.5% | Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200 |
| CBX1 | 100% | 100% | 100% | 100% | 99.9% | |
| CC2D1A | 100% | 100% | 100% | 99.9% | 98.9% | Intellectual developmental disorder, autosomal recessive 3, 608443 |
| CC2D2A | 98.2% | 98.2% | 100% | 100% | 99.8% | COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285 |
| CCBE1 | 100% | 100% | 100% | 100% | 99.7% | Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 |
| CCDC174 | 100% | 100% | 100% | 100% | 99.8% | Hypotonia, infantile, with psychomotor retardation, 616816 |
| CCDC186 | 100% | 100% | 100% | 100% | 99.7% | |
| CCDC22 | 100% | 100% | 98.8% | 88% | 69.2% | Ritscher-Schinzel syndrome 2, 300963 |
| CCDC32 | 100% | 100% | 100% | 99.9% | 99.5% | Cardiofacioneurodevelopmental syndrome, 619123 |

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| CCDC47 | 100% | 100% | 100% | 100% | 99.9% | Trichohepatoneurodevelopmental syndrome, 618268 |
| CCDC82 | 100% | 100% | 100% | 100% | 99.8% | |
| CCDC88A | 97.4% | 97.4% | 100% | 100% | 99.7% | PEHO syndrome-like, 617507 |
| CCDC88C | 100% | 100% | 100% | 99.9% | 98.8% | ?Spinocerebellar ataxia 40, 616053;Hydrocephalus, congenital, 1, 236600 |
| CCND2 | 100% | 100% | 100% | 100% | 99.3% | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 |
| CCNK | 100% | 100% | 99.9% | 98.9% | 95.1% | ?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147 |
| CCT3 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with speech or visual impairment and brain hypomyelination, 621034 |
| CCT6A | 100% | 100% | 100% | 99.9% | 99.6% | |
| CDC42 | 100% | 100% | 100% | 100% | 99.6% | Takenouchi-Kosaki syndrome, 616737 |
| CDC42BPB | 100% | 100% | 100% | 100% | 99.5% | Chilton-Okur-Chung neurodevelopmental syndrome, 619841 |
| CDC6 | 100% | 100% | 100% | 100% | 99.4% | Meier-Gorlin syndrome 5, 613805 |
| CDH11 | 100% | 100% | 100% | 100% | 99.9% | Teebi hypertelorism syndrome 2, 619736;Elsahy-Waters syndrome, 211380 |
| CDH15 | 100% | 100% | 100% | 99.9% | 99% | Intellectual developmental disorder, autosomal dominant 3, 612580 |

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| CDH2 | 100% | 100% | 100% | 100% | 99.7% | Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 |
| CDK10 | 100% | 100% | 100% | 100% | 99.6% | Al Kaissi syndrome, 617694 |
| CDK13 | 100% | 100% | 100% | 100% | 99.1% | Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 |
| CDK16 | 100% | 100% | 98.5% | 87.5% | 69.3% | |
| CDK19 | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 87, 618916 |
| CDK5RAP2 | 100% | 100% | 100% | 100% | 99.7% | Microcephaly 3, primary, autosomal recessive, 604804 |
| CDK8 | 92.4% | 92.4% | 100% | 100% | 99.6% | Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 |
| CDKL5 | 95.7% | 95.7% | 98.8% | 89.3% | 70.8% | Developmental and epileptic encephalopathy 2, 300672 |
| CDKN1C | 100% | 100% | 100% | 100% | 99% | IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650 |
| CDON | 100% | 100% | 100% | 100% | 99.9% | Holoprosencephaly 11, 614226 |
| CELF2 | 100% | 100% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 97, 619561 |

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| CELF4 | 100% | 100% | 100% | 99.9% | 99% | |
| CELSR1 | 100% | 100% | 100% | 100% | 99.2% | Lymphatic malformation 9, 619319;Yellow nail syndrome, 153300 |
| CELSR3 | 100% | 100% | 100% | 100% | 99.4% | |
| CENPF | 98.6% | 98.6% | 100% | 100% | 99.8% | Stromme syndrome, 243605 |
| CEP104 | 95.1% | 95.1% | 100% | 100% | 99.6% | Joubert syndrome 25, 616781;Intellectual developmental disorder, autosomal recessive 77, 619988 |
| CEP120 | 100% | 100% | 100% | 100% | 99.6% | Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761 |
| CEP135 | 95.8% | 95.8% | 100% | 100% | 99.7% | Microcephaly 8, primary, autosomal recessive, 614673 |
| CEP152 | 100% | 100% | 100% | 100% | 99.9% | Microcephaly 9, primary, autosomal recessive, 614852;Seckel syndrome 5, 613823 |
| CEP290 | 100% | 100% | 100% | 100% | 99.8% | Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134 |
| CEP295 | 100% | 100% | 100% | 100% | 99.9% | Seckel syndrome 11, 620767 |
| CEP41 | 100% | 100% | 100% | 100% | 99.8% | Joubert syndrome 15, 614464 |

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| CEP55 | 100% | 100% | 100% | 100% | 99.9% | Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 |
| CEP57 | 100% | 100% | 100% | 100% | 99.7% | Mosaic variegated aneuploidy syndrome 2, 614114 |
| CEP63 | 92.8% | 92.8% | 100% | 100% | 99.7% | ?Seckel syndrome 6, 614728 |
| CEP83 | 100% | 100% | 100% | 100% | 99.8% | Nephronophthisis 18, 615862 |
| CEP85L | 100% | 100% | 100% | 100% | 99.7% | Lissencephaly 10, 618873 |
| CEP89 | 98.3% | 98.3% | 100% | 100% | 99.6% | |
| CERT1 | 98.1% | 98.1% | 100% | 99.9% | 99.6% | Neurodevelopmental disorder with hypotonia, speech delay, and dysmorphic facies, 616351 |
| CHAMP1 | 100% | 100% | 100% | 100% | 99.3% | Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579 |
| CHASERR | | | | | | Neurodevelopmental disorder with dysmorphic facies, absent speech and ambulation, and brain abnormalities, 621012 |
| CHD1 | 100% | 100% | 100% | 100% | 99.9% | Pilarowski-Bjornsson syndrome, 617682 |
| CHD2 | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 94, 615369 |
| CHD3 | 100% | 100% | 100% | 99.9% | 99.1% | Snijders Blok-Campeau syndrome, 618205 |
| CHD4 | 100% | 100% | 100% | 100% | 99.5% | Sifrim-Hitz-Weiss syndrome, 617159 |

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| CHD5 | 100% | 100% | 100% | 99.8% | 98.7% | Parenti-Mignot neurodevelopmental syndrome, 619873 |
| CHD7 | 100% | 100% | 100% | 100% | 99.8% | Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800 |
| CHD8 | 100% | 100% | 100% | 100% | 99.5% | Intellectual developmental disorder with autism and macrocephaly, 615032 |
| CHKA | 100% | 100% | 100% | 100% | 99.1% | Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023 |
| CHKB | 100% | 100% | 100% | 100% | 99.5% | Muscular dystrophy, congenital, megaconial type, 602541 |
| CHMP1A | 100% | 100% | 100% | 99.9% | 99.2% | Pontocerebellar hypoplasia, type 8, 614961 |
| CHRM1 | 100% | 100% | 100% | 100% | 99.4% | |
| CHRNA4 | 100% | 100% | 100% | 99.7% | 96.4% | {Nicotine addiction, susceptibility to}, 188890;Epilepsy, nocturnal frontal lobe, 1, 600513 |
| CIAO1 | 100% | 100% | 100% | 99.7% | 98.8% | Multiple mitochondrial dysfunctions syndrome 10, 620960 |
| CIC | 100% | 100% | 100% | 99.9% | 99% | Intellectual developmental disorder, autosomal dominant 45, 617600 |
| CIT | 95.8% | 95.8% | 100% | 100% | 99.4% | Microcephaly 17, primary, autosomal recessive, 617090 |
| CIZ1 | 100% | 100% | 100% | 100% | 99.3% | |
| CKAP2L | 100% | 100% | 100% | 100% | 99.7% | Filippi syndrome, 272440 |

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|---|---|---|---|---|---|---|
| CLCN3 | 100% | 100% | 100% | 100% | 99.8% | ?Neurodevelopmental disorder with seizures and brain abnormalities, 619517;Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512 |
| CLCN4 | 100% | 100% | 98.5% | 88.6% | 71.1% | Raynaud-Claes syndrome, 300114 |
| CLCN6 | 97.7% | 97.7% | 100% | 100% | 99.6% | Ceroid lipofuscinosis, neuronal, 15, 619173 |
| CLDN11 | 100% | 100% | 100% | 99.9% | 99% | Leukodystrophy, hypomyelinating, 22, 619328 |
| CLDN5 | 100% | 100% | 100% | 100% | 99.3% | |
| CLIC2 | 87.2% | 87.2% | 99.7% | 92.8% | 74.4% | |
| CLIP1 | 100% | 100% | 100% | 100% | 99.8% | |
| CLN3 | 93.1% | 93.1% | 100% | 100% | 99.4% | Ceroid lipofuscinosis, neuronal, 3, 204200;Retinitis pigmentosa 101, 621548 |
| CLN5 | 83% | 83% | 100% | 100% | 99.8% | Ceroid lipofuscinosis, neuronal, 5, 256731 |
| CLN6 | 95.1% | 95.1% | 100% | 100% | 99.4% | Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780 |
| CLN8 | 100% | 100% | 100% | 100% | 99.5% | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143 |
| CLP1 | 100% | 100% | 100% | 100% | 99.5% | Pontocerebellar hypoplasia, type 10, 615803 |

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|---|---|---|---|---|---|---|
| CLPB | 98.2% | 98.2% | 100% | 100% | 99.7% | Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 |
| CLTC | 97.3% | 96% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 56, 617854 |
| CMIP | 100% | 100% | 100% | 100% | 99.4% | |
| CNKSR2 | 100% | 100% | 99.2% | 91.3% | 72.7% | Intellectual developmental disorder, X-linked syndromic, Houge type, 301008 |
| CNNM2 | 100% | 100% | 100% | 100% | 99.4% | Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418 |
| CNOT1 | 100% | 100% | 100% | 100% | 99.7% | Vissers-Bodmer syndrome, 619033;Holoprosencephaly 12, with or without pancreatic agenesis, 618500 |
| CNOT2 | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 |
| CNOT3 | 100% | 100% | 100% | 99.9% | 98.8% | Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 |
| CNOT9 | 98.4% | 93.6% | 100% | 100% | 99.9% | |

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| CNPY3 | 99.1% | 95.7% | 100% | 100% | 99.3% | Developmental and epileptic encephalopathy 60, 617929 |
| CNTNAP1 | 100% | 100% | 100% | 100% | 99.1% | Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186 |
| CNTNAP2 | 100% | 100% | 100% | 100% | 99.7% | Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100 |
| COA8 | 100% | 100% | 100% | 100% | 99.7% | Mitochondrial complex IV deficiency, nuclear type 17, 619061 |
| COASY | 100% | 100% | 100% | 99.8% | 99.1% | Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643 |
| COG1 | 100% | 100% | 100% | 100% | 99.4% | Congenital disorder of glycosylation, type IIg, 611209 |
| COG3 | 100% | 100% | 100% | 100% | 99.8% | Congenital disorder of glycosylation, type IIbb, 620546 |
| COG4 | 97% | 97% | 100% | 100% | 99.3% | Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150 |
| COG5 | 96.8% | 96.8% | 100% | 100% | 99.8% | Congenital disorder of glycosylation, type IIi, 613612 |
| COG6 | 100% | 100% | 100% | 100% | 99.9% | Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576 |
| COG7 | 95.1% | 95.1% | 100% | 100% | 99.6% | Congenital disorder of glycosylation, type IIe, 608779 |

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| COG8 | 100% | 100% | 100% | 100% | 99.6% | Congenital disorder of glycosylation, type IIh, 611182 |
| COL18A1 | 99.2% | 98% | 100% | 99.9% | 99.2% | Knobloch syndrome, type 1, 267750;Glaucoma, primary closed-angle, 618880 |
| COL4A1 | 100% | 100% | 100% | 100% | 99.7% | ?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 | 98.6% | 98.5% | 100% | 100% | 99.3% | Brain small vessel disease 2A, autosomal dominant, 614483;{Hemorrhage, intracerebral, susceptibility to}, 614519;Brain small vessel disease 2B, autosomal recessive, 621414 |
| COLEC11 | 100% | 100% | 100% | 100% | 99.6% | 3MC syndrome 2, 265050 |
| COLGALT1 | 96% | 92.4% | 100% | 100% | 99% | Brain small vessel disease 3, 618360 |
| COPB1 | 96.3% | 96.3% | 100% | 99.9% | 99.7% | Baralle-Macken syndrome, 619255 |

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| COPB2 | 96.7% | 96.7% | 100% | 100% | 99.8% | Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884;?Microcephaly 19, primary, autosomal recessive, 617800 |
| COQ2 | 96.3% | 96.3% | 100% | 100% | 99.5% | {Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426 |
| COQ4 | 96.3% | 96.3% | 100% | 100% | 99.6% | Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666 |
| COQ7 | 100% | 100% | 100% | 100% | 99.5% | Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 |
| COQ8A | 89.1% | 87.2% | 100% | 99.9% | 99.5% | Coenzyme Q10 deficiency, primary, 4, 612016 |
| COQ9 | 91.9% | 91.9% | 100% | 99.9% | 99.2% | Coenzyme Q10 deficiency, primary, 5, 614654 |
| COX10 | 100% | 100% | 100% | 100% | 99.7% | Mitochondrial complex IV deficiency, nuclear type 3, 619046 |
| COX11 | 100% | 100% | 100% | 99.9% | 98.7% | Mitochondrial complex IV deficiency, nuclear type 23, 620275 |
| COX15 | 100% | 100% | 100% | 100% | 99.4% | Mitochondrial complex IV deficiency, nuclear type 6, 615119 |
| COX16 | 61.9% | 61.9% | 100% | 99.8% | 99.4% | Mitochondrial complex IV deficiency, nuclear type 22, 619355 |
| COX6B1 | 80.9% | 80.9% | 100% | 99.8% | 98.7% | Mitochondrial complex IV deficiency, nuclear type 7, 619051 |

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| CPAP | 100% | 100% | 100% | 100% | 99.7% | Microcephaly 6, primary, autosomal recessive, 608393;Seckel syndrome 4, 613676 |
| CPE | 97.5% | 97.5% | 100% | 99.9% | 99.1% | BDV syndrome, 619326 |
| CPLANE1 | 100% | 100% | 100% | 100% | 99.8% | Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615 |
| CPLX1 | 100% | 100% | 100% | 100% | 99.6% | Developmental and epileptic encephalopathy 63, 617976 |
| CPS1 | 100% | 100% | 100% | 100% | 99.7% | Carbamoylphosphate synthetase I deficiency, 237300 |
| CPSF3 | 95.9% | 95.9% | 100% | 100% | 99.9% | Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876 |
| CRADD | 100% | 100% | 100% | 100% | 99.4% | Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499 |
| CRBN | 100% | 99.6% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 2, 607417 |
| CREBBP | 100% | 100% | 100% | 99.9% | 99.5% | Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849 |

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| CRELD1 | 100% | 100% | 100% | 100% | 99.6% | Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217 |
| CRIPT | 100% | 100% | 100% | 100% | 99.9% | Rothmund-Thomson syndrome, type 3, 615789 |
| CRLF1 | 94.8% | 90.1% | 99.7% | 98.1% | 93.3% | Cold-induced sweating syndrome 1, 272430 |
| CRLS1 | 89.6% | 84.4% | 100% | 99.7% | 99.4% | Combined oxidative phosphorylation deficiency 57, 620167 |
| CRNKL1 | 100% | 100% | 100% | 100% | 99.8% | Microcephaly, progressive, with simplified gyral pattern and cerebellar hypoplasia, 621436 |
| CRPPA | 100% | 100% | 100% | 100% | 99.3% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 |
| CSDE1 | 97.8% | 97.8% | 100% | 100% | 99.8% | |
| CSF1R | 97.3% | 97.3% | 100% | 100% | 99.6% | Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 |
| CSMD1 | 100% | 100% | 100% | 99.9% | 99.4% | |
| CSNK1G1 | 100% | 100% | 100% | 100% | 99.7% | |

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| CSNK2A1 | 94.2% | 94.2% | 100% | 100% | 99.7% | Okur-Chung neurodevelopmental syndrome, 617062 |
| CSNK2B | 100% | 100% | 100% | 100% | 99.7% | Poirier-Bienvenu neurodevelopmental syndrome, 618732 |
| CSPP1 | 96.9% | 96.9% | 100% | 100% | 99.6% | Joubert syndrome 21, 615636 |
| CSTB | 100% | 100% | 100% | 100% | 99.4% | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 |
| CTBP1 | 100% | 99.6% | 100% | 99.8% | 99.3% | Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 |
| CTC1 | 100% | 100% | 100% | 100% | 99.4% | Cerebroretinal microangiopathy with calcifications and cysts, 612199 |
| CTCF | 100% | 100% | 100% | 100% | 99.4% | Intellectual developmental disorder, autosomal dominant 21, 615502 |
| CTDP1 | 90.8% | 90.5% | 100% | 100% | 99.2% | Congenital cataracts, facial dysmorphism, and neuropathy, 604168 |
| CTNNA2 | 100% | 100% | 100% | 100% | 99.6% | Cortical dysplasia, complex, with other brain malformations 9, 618174 |

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| CTNNB1 | 100% | 100% | 100% | 100% | 99.9% | 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 |
| CTNND1 | 100% | 100% | 100% | 100% | 99.9% | Blepharocheilodontic syndrome 2, 617681 |
| CTNND2 | 100% | 100% | 100% | 99.9% | 98.4% | |
| CTR9 | 100% | 100% | 100% | 100% | 99.6% | |
| CTSA | 100% | 100% | 100% | 100% | 99.5% | Galactosialidosis, 256540;Brain small vessel disease 6 with leukoencephalopathy, 621394 |
| CTSD | 86% | 86% | 100% | 100% | 99.5% | Ceroid lipofuscinosis, neuronal, 10, 610127 |
| CTTNBP2 | 100% | 100% | 100% | 100% | 99.8% | |
| CTU2 | 100% | 100% | 100% | 100% | 99.2% | Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 |
| CUL3 | 97.6% | 97.6% | 100% | 100% | 99.8% | Neurodevelopmental disorder with or without autism or seizures, 619239;Pseudohypoaldosteronism, type IIE, 614496 |
| CUL4B | 96.7% | 96.7% | 99.2% | 91.3% | 74% | Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354 |

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| CUX1 | 100% | 100% | 100% | 99.8% | 98.8% | Neurodevelopmental disorder with developmental delay and with or without motor or speech delay, 618330 |
| CUX2 | 100% | 100% | 100% | 100% | 99.3% | Developmental and epileptic encephalopathy 67, 618141 |
| CWC27 | 82.6% | 82.6% | 100% | 100% | 99.8% | Retinitis pigmentosa with or without skeletal anomalies, 250410 |
| CWF19L1 | 100% | 100% | 100% | 100% | 99.8% | Spinocerebellar ataxia, autosomal recessive 17, 616127 |
| CYB5R3 | 95.7% | 95% | 100% | 100% | 99.2% | Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800 |
| CYFIP2 | 96.2% | 96.2% | 100% | 100% | 99.6% | Developmental and epileptic encephalopathy 65, 618008 |
| CYP27A1 | 100% | 100% | 100% | 100% | 99.5% | Cerebrotendinous xanthomatosis, 213700 |
| CYP2U1 | 100% | 100% | 100% | 99.9% | 98.8% | Spastic paraplegia 56, autosomal recessive, 615030 |
| D2HGDH | 99.9% | 98.4% | 100% | 99.9% | 99.4% | D-2-hydroxyglutaric aciduria, 600721 |
| DAG1 | 100% | 100% | 100% | 100% | 99.3% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 |
| DAGLA | 100% | 100% | 100% | 100% | 99.3% | Neuroocular syndrome 2, paroxysmal type, 168885 |

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| DAP3 | 91.7% | 90.8% | 100% | 100% | 99.6% | Perrault syndrome 7, 621101 |
| DARS1 | 100% | 100% | 100% | 100% | 99.8% | Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 |
| 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 |
| DBT | 100% | 100% | 100% | 100% | 99.9% | Maple syrup urine disease, type II, 620699 |
| DCAF17 | 96.5% | 96.5% | 100% | 99.9% | 99.7% | Woodhouse-Sakati syndrome, 241080 |
| DCC | 100% | 100% | 100% | 100% | 99.7% | 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 |
| DCHS1 | 100% | 100% | 100% | 100% | 99.6% | Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390 |
| DCPS | 100% | 100% | 100% | 100% | 99.6% | Al-Raqad syndrome, 616459 |
| DCX | 100% | 100% | 99% | 89.8% | 73% | Subcortical laminal heterotopia, X-linked, 300067;Lissencephaly, X-linked, 300067 |
| DDB1 | 92.4% | 92.3% | 100% | 100% | 99.6% | White-Kernohan syndrome, 619426 |
| DDC | 100% | 100% | 100% | 100% | 99.7% | Aromatic L-amino acid decarboxylase deficiency, 608643 |

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| DDHD2 | 100% | 100% | 100% | 100% | 99.7% | Spastic paraplegia 54, autosomal recessive, 615033 |
| DDX11 | 100% | 100% | 100% | 100% | 99.5% | Warsaw breakage syndrome, 613398 |
| DDX17 | 100% | 100% | 100% | 100% | 99.5% | |
| DDX23 | 100% | 100% | 100% | 99.9% | 99.7% | |
| DDX39B | 100% | 100% | 100% | 100% | 99.6% | |
| DDX3X | 99.4% | 98.7% | 99.1% | 90.6% | 72.1% | Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 |
| DDX59 | 100% | 100% | 100% | 100% | 99.8% | Orofaciodigital syndrome V, 174300 |
| DDX6 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder with impaired language and dysmorphic facies, 618653 |
| DEAF1 | 93.3% | 88.8% | 100% | 99.9% | 98% | Vulto-van Silfout-de Vries syndrome, 615828;Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171 |
| DEGS1 | 100% | 100% | 100% | 100% | 99.8% | Leukodystrophy, hypomyelinating, 18, 618404 |
| DENND5A | 100% | 100% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 49, 617281 |
| DENND5B | 96% | 96% | 100% | 100% | 99.7% | |
| DEPDC5 | 100% | 100% | 100% | 99.9% | 99.4% | Epilepsy, familial focal, with variable foci 1, 604364;Developmental and epileptic encephalopathy 111, 620504 |

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| DHCR24 | 97.8% | 97.8% | 100% | 99.7% | 98.6% | Desmosterolosis, 602398 |
| DHCR7 | 96.2% | 96.2% | 100% | 100% | 99.6% | Smith-Lemli-Opitz syndrome, 270400 |
| DHDDS | 74.4% | 74.4% | 100% | 100% | 99.8% | Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 621567;Retinitis pigmentosa 59, 613861 |
| DHFR | 100% | 100% | 100% | 100% | 99.9% | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 |
| DHPS | 96.4% | 93.1% | 100% | 99.9% | 99% | Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 |
| DHRS3 | 100% | 100% | 100% | 100% | 99.5% | Craniosynostosis-scoliosis syndrome, 621499 |
| DHRSX | 50% | 50% | 50% | 50% | 49.7% | Congenital disorder of glycosylation, type 1DD, 301133 |
| DHTKD1 | 92.7% | 92.7% | 100% | 100% | 99.4% | ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 |
| DHX16 | 100% | 100% | 100% | 100% | 99.4% | Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 |
| DHX30 | 100% | 100% | 100% | 100% | 99.3% | Neurodevelopmental disorder with variable motor and speech impairment, 617804 |

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| DHX37 | 100% | 100% | 100% | 100% | 99.4% | Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731;46XY sex reversal 11, 273250 |
| DHX9 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 75, 620988 |
| DIAPH1 | 100% | 100% | 100% | 100% | 99.5% | Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632 |
| DIP2B | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630 |
| DIP2C | 100% | 100% | 100% | 100% | 99.4% | |
| DIS3L2 | 100% | 100% | 100% | 99.9% | 99.5% | Perlman syndrome, 267000 |
| DKC1 | 100% | 100% | 99% | 89.5% | 70.6% | ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000 |
| DLAT | 100% | 100% | 100% | 100% | 99.4% | Pyruvate dehydrogenase E2 deficiency, 245348 |
| DLD | 100% | 100% | 100% | 100% | 99.8% | Dihydrolipoamide dehydrogenase deficiency, 246900 |
| DLG3 | 100% | 100% | 98.7% | 89.2% | 70.2% | Intellectual developmental disorder, X-linked 90, 300850 |

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| DLG4 | 100% | 100% | 100% | 100% | 99.4% | Intellectual developmental disorder, autosomal dominant 62, 618793 |
| DLL1 | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 |
| DMAP1 | 92.4% | 92.4% | 100% | 100% | 99.7% | |
| DMD | 99.6% | 99.6% | 99.2% | 91.5% | 73.3% | Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200 |
| DMPK | 100% | 100% | 100% | 99.9% | 99.1% | Myotonic dystrophy 1, 160900 |
| DMXL2 | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113 |
| DNAJC12 | 100% | 100% | 100% | 100% | 99.7% | Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 |
| DNAJC19 | 100% | 100% | 100% | 100% | 99.9% | 3-methylglutaconic aciduria, type V, 610198 |
| DNM1 | 100% | 100% | 100% | 100% | 99.2% | Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352;Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346 |

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| DNM1L | 100% | 100% | 100% | 100% | 99.9% | Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 |
| DNMT3A | 100% | 100% | 100% | 100% | 99.5% | Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724 |
| DNMT3B | 100% | 100% | 100% | 100% | 99.7% | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478 |
| DOCK3 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 |
| DOCK4 | 100% | 100% | 100% | 100% | 99.7% | |
| DOCK6 | 100% | 100% | 100% | 99.9% | 99.3% | Adams-Oliver syndrome 2, 614219 |
| DOCK7 | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 23, 615859 |
| DOHH | 98.4% | 92.9% | 100% | 100% | 99.6% | Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066 |
| DOLK | 100% | 100% | 100% | 100% | 99.6% | Congenital disorder of glycosylation, type Im, 610768 |

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|---|---|---|---|---|---|---|
| DONSON | 100% | 99.7% | 100% | 100% | 99.7% | Microcephaly, short stature, and limb abnormalities, 617604;Microcephaly-micromelia syndrome, 251230;Meier-Gorlin syndrome 10, 621528 |
| DOT1L | 100% | 100% | 100% | 100% | 99.3% | Nil-Deshwar neurodevelopmental syndrome, 621265 |
| DPAGT1 | 100% | 100% | 100% | 100% | 99.4% | Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093 |
| DPF2 | 95.7% | 95.7% | 100% | 99.8% | 99.1% | Coffin-Siris syndrome 7, 618027 |
| DPH1 | 100% | 100% | 100% | 100% | 99.3% | Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 |
| DPH5 | 75.4% | 75.4% | 100% | 100% | 99.9% | Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070 |
| DPM1 | 99.6% | 96.9% | 99.6% | 98.4% | 96.6% | Congenital disorder of glycosylation, type Ie, 608799 |
| DPM2 | 100% | 99.9% | 100% | 100% | 99.5% | Congenital disorder of glycosylation, type Iu, 615042 |
| DPP6 | 100% | 100% | 100% | 100% | 99.5% | Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956 |

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|---|---|---|---|---|---|---|
| DPYD | 86.9% | 86.9% | 100% | 100% | 99.9% | Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270 |
| DPYS | 100% | 100% | 100% | 100% | 99.5% | Dihydropyrimidinuria, 222748 |
| DPYSL2 | 100% | 100% | 100% | 100% | 99.4% | |
| DPYSL5 | 100% | 100% | 100% | 99.9% | 99.6% | Ritscher-Schinzel syndrome 4, 619435 |
| DRG1 | 92.1% | 92.1% | 100% | 100% | 99.7% | Tan-Almurshedi syndrome, 620641 |
| DTYMK | 91.4% | 84.2% | 100% | 100% | 99.1% | Neurodegeneration, childhood-onset, with progressive microcephaly, 619847 |
| DYM | 100% | 100% | 100% | 100% | 99.9% | Smith-McCort dysplasia, 607326;Dyggve-Melchior-Clausen disease, 223800 |
| 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 |
| DYNC1I2 | 92.4% | 92.4% | 100% | 100% | 99.9% | Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492 |
| DYRK1A | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal dominant 7, 614104 |
| EARS2 | 100% | 100% | 100% | 99.9% | 99.3% | Combined oxidative phosphorylation deficiency 12, 614924 |

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|---|---|---|---|---|---|---|
| EBF3 | 100% | 100% | 100% | 99.9% | 99.2% | Hypotonia, ataxia, and delayed development syndrome, 617330 |
| EBP | 100% | 100% | 98.8% | 88.4% | 69.5% | MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960 |
| ECHS1 | 71.2% | 69% | 100% | 100% | 99.3% | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 |
| EDC3 | 100% | 100% | 100% | 100% | 99.5% | ?Intellectual developmental disorder, autosomal recessive 50, 616460 |
| EDEM3 | 100% | 100% | 100% | 100% | 99.8% | Congenital disorder of glycosylation, type IIv, 619493 |
| EED | 99.1% | 99.1% | 100% | 99.6% | 97.9% | Cohen-Gibson syndrome, 617561 |
| EEF1A2 | 99.4% | 95.6% | 100% | 100% | 99.1% | Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393 |
| EEF1D | 100% | 100% | 100% | 100% | 99.5% | Neurodevelopmental disorder with thin corpus callosum, hypotonia, and absent language, 621150 |
| EEFSEC | 100% | 100% | 100% | 100% | 98.7% | Neurodevelopmental disorder with progressive spasticity and brain abnormalities, 621102 |
| EFL1 | 96.3% | 96.3% | 100% | 100% | 99.9% | Shwachman-Diamond syndrome 2, 617941 |
| EFNB1 | 100% | 100% | 98.8% | 87.7% | 69.3% | Craniofrontonasal dysplasia, 304110 |
| EFNB2 | 100% | 100% | 100% | 100% | 99.6% | |

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|---|---|---|---|---|---|---|
| EFTUD2 | 100% | 100% | 100% | 100% | 99.7% | Mandibulofacial dysostosis, Guion-Almeida type, 610536 |
| EHMT1 | 97.7% | 97.7% | 100% | 100% | 99.7% | Kleefstra syndrome 1, 610253 |
| EHMT2 | 97% | 97% | 100% | 99.9% | 99% | |
| EIF1AX | 100% | 100% | 98.9% | 92.3% | 73.7% | |
| EIF2AK1 | 95% | 94.1% | 100% | 100% | 99.7% | ?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 |
| EIF2AK2 | 100% | 100% | 100% | 100% | 99.8% | Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877;Dystonia 33, 619687 |
| EIF2AK3 | 100% | 100% | 100% | 100% | 99.7% | Wolcott-Rallison syndrome, 226980 |
| EIF2B2 | 100% | 100% | 100% | 99.9% | 99.2% | Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312 |
| EIF2B3 | 100% | 100% | 100% | 100% | 99.5% | Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313 |
| EIF2B4 | 100% | 100% | 100% | 100% | 99.9% | Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314 |
| EIF2B5 | 100% | 100% | 100% | 100% | 99.6% | Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 |
| EIF2S3 | 100% | 100% | 99.2% | 91% | 73.9% | MEHMO syndrome, 300148 |

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|---|---|---|---|---|---|---|
| EIF3F | 100% | 100% | 100% | 100% | 99.3% | Intellectual developmental disorder, autosomal recessive 67, 618295 |
| EIF4A2 | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455 |
| EIF4A3 | 100% | 100% | 100% | 100% | 99.4% | Robin sequence with cleft mandible and limb anomalies, 268305 |
| EIF5A | 100% | 100% | 100% | 100% | 99.2% | Faundes-Banka syndrome, 619376 |
| EIPR1 | 91.2% | 91.2% | 100% | 100% | 99.7% | |
| ELAC2 | 94.8% | 94.8% | 100% | 99.9% | 99.5% | {Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440 |
| ELFN1 | 100% | 100% | 100% | 99.9% | 98.5% | Dursun-Ozgul neurodevelopmental syndrome, 621344 |
| ELOVL4 | 100% | 100% | 100% | 100% | 99.8% | Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 |
| ELP2 | 95% | 94.7% | 100% | 100% | 99.9% | Intellectual developmental disorder, autosomal recessive 58, 617270 |
| EMC1 | 99.6% | 98.2% | 100% | 100% | 99.6% | Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 |

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|---|---|---|---|---|---|---|
| EMC10 | 100% | 100% | 100% | 100% | 99.4% | Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264 |
| EML1 | 90.9% | 90.9% | 100% | 100% | 99.4% | Band heterotopia, 600348 |
| EMX2 | 100% | 100% | 100% | 99.7% | 96% | Schizencephaly, 269160 |
| ENTPD1 | 100% | 100% | 100% | 100% | 99.6% | Spastic paraplegia 64, autosomal recessive, 615683 |
| EP300 | 100% | 100% | 100% | 100% | 99.5% | Menke-Hennekam syndrome 2, 618333;Colorectal cancer, somatic, 114500;Rubinstein-Taybi syndrome 2, 613684 |
| EP400 | 100% | 100% | 100% | 100% | 99.4% | |
| EPB41L3 | 99.9% | 99.3% | 100% | 100% | 99.7% | |
| EPG5 | 100% | 100% | 100% | 100% | 99.8% | Vici syndrome, 242840;Neurodevelopmental disorder with parkinsonism or other movement abnormalities, 621506 |
| EPHA7 | 100% | 100% | 100% | 100% | 99.7% | |
| EPRS1 | 96.2% | 96.2% | 100% | 100% | 99.4% | Leukodystrophy, hypomyelinating, 15, 617951 |
| ERCC1 | 100% | 100% | 100% | 99.9% | 99.2% | Cerebrooculofacioskeletal syndrome 4, 610758 |
| ERCC2 | 96.3% | 94.6% | 100% | 100% | 99.3% | Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756 |

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| ERCC3 | 100% | 100% | 100% | 100% | 99.7% | Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651 |
| ERCC5 | 100% | 100% | 100% | 100% | 99.8% | Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 |
| ERCC6 | 100% | 100% | 100% | 100% | 99.6% | UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980 |
| ERCC8 | 95.2% | 95.2% | 100% | 99.9% | 98.8% | UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400 |
| ERF | 100% | 100% | 100% | 100% | 97.9% | Craniosynostosis 4, 600775;Chitayat syndrome, 617180 |
| ERI1 | 100% | 100% | 100% | 100% | 99.6% | Hoxha-Aliu syndrome, 620662;Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663 |
| ERLIN2 | 100% | 100% | 100% | 100% | 99.5% | Spastic paraplegia 18A, autosomal dominant, 620512;Spastic paraplegia 18B, autosomal recessive, 611225 |

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|---|---|---|---|---|---|---|
| ESAM | 99.7% | 96.8% | 100% | 99.9% | 99.1% | Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371 |
| ESCO2 | 100% | 100% | 100% | 100% | 99.8% | Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300 |
| ETFB | 92.9% | 92.9% | 100% | 100% | 99.3% | Glutaric acidemia IIB, 231680 |
| ETHE1 | 77.1% | 77.1% | 100% | 100% | 99.7% | Ethylmalonic encephalopathy, 602473 |
| EXOC2 | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306 |
| EXOC7 | 98.6% | 98.6% | 100% | 100% | 99.6% | Neurodevelopmental disorder with seizures and brain atrophy, 619072 |
| EXOC8 | 100% | 100% | 100% | 100% | 99.5% | ?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076 |
| EXOSC2 | 91.1% | 91% | 100% | 100% | 99.7% | Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 |
| EXOSC3 | 100% | 100% | 100% | 100% | 99.7% | Pontocerebellar hypoplasia, type 1B, 614678 |
| EXOSC5 | 100% | 100% | 100% | 100% | 98.6% | Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576 |
| EXOSC8 | 100% | 100% | 100% | 100% | 99.8% | Pontocerebellar hypoplasia, type 1C, 616081 |

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|---|---|---|---|---|---|---|
| EXOSC9 | 100% | 100% | 100% | 100% | 99.9% | Pontocerebellar hypoplasia, type 1D, 618065 |
| EXT2 | 100% | 100% | 100% | 100% | 99.6% | Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701 |
| EXTL3 | 100% | 99.9% | 100% | 100% | 99.7% | Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 |
| EZH1 | 100% | 100% | 100% | 100% | 99.7% | |
| EZH2 | 100% | 100% | 100% | 100% | 99.7% | Weaver syndrome, 277590 |
| FA2H | 100% | 100% | 100% | 100% | 99.7% | Spastic paraplegia 35, autosomal recessive, 612319 |
| FAM149B1 | 100% | 100% | 100% | 100% | 99.9% | Joubert syndrome 36, 618763 |
| FAM177A1 | 86.5% | 86.5% | 100% | 99.9% | 99.3% | Neurodevelopmental disorder with white matter abnormalities and gait disturbance, 621152 |
| FAM20C | 99.4% | 97.2% | 100% | 100% | 99.2% | Raine syndrome, 259775 |
| FAM50A | 93% | 93% | 98.9% | 88.4% | 67.1% | Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261 |
| FAR1 | 91% | 90.9% | 100% | 100% | 99.9% | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338 |

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| FARS2 | 100% | 100% | 100% | 100% | 99.5% | Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046 |
| FARSA | 100% | 100% | 100% | 100% | 99.8% | ?Rajab interstitial lung disease with brain calcifications 2, 619013 |
| FARSB | 94% | 94% | 100% | 100% | 99.7% | Rajab interstitial lung disease with brain calcifications 1, 613658 |
| FASTKD2 | 100% | 100% | 100% | 100% | 99.8% | Combined oxidative phosphorylation deficiency 44, 618855 |
| FAT4 | 100% | 100% | 100% | 99.9% | 99.5% | Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 |
| FBRSL1 | 100% | 99.1% | 99.6% | 96.9% | 89.1% | |
| FBXL3 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 |
| FBXL4 | 95.6% | 95.6% | 100% | 100% | 99.8% | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 |
| FBXO11 | 100% | 100% | 100% | 99.9% | 99.3% | Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 |
| FBXO22 | 100% | 100% | 100% | 100% | 99.6% | Tayoun-Maawali syndrome, 621184 |
| FBXO28 | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 100, 619777 |

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| FBXO31 | 100% | 100% | 100% | 100% | 99.2% | ?Intellectual developmental disorder, autosomal recessive 45, 615979 |
| FBXW11 | 100% | 100% | 100% | 100% | 99.4% | Neurodevelopmental, jaw, eye, and digital syndrome, 618914 |
| FBXW7 | 100% | 100% | 100% | 100% | 99.6% | Developmental delay, hypotonia, and impaired language, 620012 |
| FCSK | 100% | 100% | 100% | 100% | 99.6% | Congenital disorder of glycosylation with defective fucosylation 2, 618324 |
| FDFT1 | 100% | 98.7% | 100% | 100% | 99.5% | Squalene synthase deficiency, 618156 |
| FEM1B | 100% | 100% | 100% | 100% | 99.5% | Neurodevelopmental disorder with behavioral, ear, and skeletal abnormalities, 621263 |
| FERRY3 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 66, 618221 |
| FEZF2 | 100% | 100% | 100% | 100% | 98.9% | |
| FGD1 | 100% | 100% | 98.4% | 88% | 69.3% | Aarskog-Scott syndrome, 305400 |
| FGF12 | 100% | 100% | 100% | 100% | 99.1% | Developmental and epileptic encephalopathy 47, 617166 |
| FGF13 | 100% | 99.6% | 99.6% | 91.4% | 73.2% | Developmental and epileptic encephalopathy 90, 301058;Intellectual developmental disorder, X-linked 110, 301095 |

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|---|---|---|---|---|---|---|
| FGF14 | 100% | 100% | 100% | 100% | 99.7% | Spinocerebellar ataxia 27A, 193003;Spinocerebellar ataxia 27B, late-onset, 620174 |
| FGFR1 | 100% | 99.2% | 100% | 100% | 99.4% | Pfeiffer syndrome, 101600;Hypogonadotropic hypogonadism 2 with or without anosmia, 147950;Jackson-Weiss syndrome, 123150;Hartsfield syndrome, 615465;Trigonocephaly 1, 190440;Osteoglophonic dysplasia, 166250;Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 |

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| FGFR2 | 100% | 100% | 100% | 100% | 99.7% | Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Scaphocephaly and Axenfeld-Rieger anomaly;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;Craniosynostosis, nonspecific;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400 |

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| FGFR3 | 100% | 100% | 100% | 99.9% | 99.1% | Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247 |
| FH | 100% | 100% | 100% | 100% | 99.8% | Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812 |
| FIBP | 98.7% | 95% | 100% | 100% | 99.5% | Thauvin-Robinet-Faivre syndrome, 617107 |
| 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 |
| FIGN | 100% | 100% | 100% | 100% | 99.6% | |
| FILIP1 | 100% | 100% | 100% | 100% | 99.7% | Neuromuscular disorder, congenital, with dysmorphic facies, 620775 |
| FITM2 | 100% | 100% | 100% | 100% | 99.2% | Siddiqi syndrome, 618635 |

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| FKRP | 100% | 100% | 100% | 100% | 99.3% | Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 |
| FKTN | 95.2% | 94.9% | 100% | 100% | 99.9% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800;Cardiomyopathy, dilated, 1X, 611615;Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152 |

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| FLNA | 100% | 100% | 98.3% | 87.4% | 68.7% | Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620 |
| FLVCR1 | 95% | 94.6% | 100% | 100% | 99.7% | Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060 |
| FLVCR2 | 100% | 100% | 100% | 100% | 99.7% | Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 |
| FMN2 | 100% | 100% | 99.9% | 99.7% | 97.9% | Intellectual developmental disorder, autosomal recessive 47, 616193 |
| FMR1 | 97% | 97% | 99.2% | 92.8% | 75.4% | Fragile X tremor/ataxia syndrome, 300623;Fragile X syndrome, 300624;Premature ovarian failure 1, 311360 |
| FOLR1 | 100% | 100% | 100% | 100% | 99.5% | Neurodegeneration due to cerebral folate transport deficiency, 613068 |

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| FOSL2 | 100% | 100% | 100% | 99.9% | 99.2% | Aplasia cutis-enamel dysplasia syndrome, 620789 |
| FOXG1 | 100% | 100% | 100% | 99.9% | 98.2% | Rett syndrome, congenital variant, 613454 |
| FOXJ1 | 100% | 100% | 100% | 99.9% | 99.4% | Ciliary dyskinesia, primary, 43, 618699 |
| FOXP1 | 100% | 100% | 100% | 100% | 99.9% | Intellectual developmental disorder with language impairment with or without autistic features, 613670 |
| FOXP2 | 100% | 100% | 100% | 100% | 99.7% | Speech-language disorder-1, 602081 |
| FOXP4 | 100% | 100% | 100% | 100% | 99.3% | |
| FOXRED1 | 100% | 100% | 100% | 100% | 99.1% | Mitochondrial complex I deficiency, nuclear type 19, 618241 |
| FRA10AC1 | 92.2% | 92.2% | 100% | 99.9% | 99.5% | Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113 |
| FRAS1 | 100% | 100% | 100% | 100% | 99.7% | Fraser syndrome 1, 219000 |
| FRMD4A | 96.6% | 96.6% | 100% | 99.9% | 99.5% | ?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 |
| FRMD5 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094 |
| FRMPD4 | 100% | 100% | 98.7% | 88.4% | 70.1% | Intellectual developmental disorder, X-linked 104, 300983 |

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| FRRS1L | 100% | 100% | 100% | 100% | 98.2% | Developmental and epileptic encephalopathy 37, 616981 |
| FRYL | 100% | 100% | 100% | 99.9% | 99.6% | Pan-Chung-Bellen syndrome, 621049 |
| FSD1L | 100% | 100% | 100% | 99.9% | 99.6% | |
| FTCD | 100% | 100% | 100% | 99.8% | 98.7% | Glutamate formiminotransferase deficiency, 229100 |
| FTH1 | 100% | 100% | 100% | 100% | 99.7% | Neurodegeneration with brain iron accumulation 9, 620669;?Hemochromatosis, type 5, 615517 |
| FTO | 94.5% | 94.5% | 100% | 100% | 99.9% | Growth retardation, developmental delay, facial dysmorphism, 612938;{Obesity, susceptibility to, BMIQ14}, 612460 |
| FTSJ1 | 100% | 100% | 98.6% | 86.8% | 71.1% | Intellectual developmental disorder, X-linked 9, 309549 |
| FUCA1 | 100% | 100% | 100% | 100% | 99.5% | Fucosidosis, 230000 |
| FUT8 | 97.2% | 97.2% | 100% | 100% | 99.8% | Congenital disorder of glycosylation with defective fucosylation 1, 618005 |
| FZR1 | 99.7% | 97.1% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 109, 620145 |
| GABBR1 | 100% | 100% | 100% | 99.9% | 98.9% | Neurodevelopmental disorder with language delay and variable cognitive abnormalities, 620502 |

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|---|---|---|---|---|---|---|
| GABBR2 | 100% | 100% | 100% | 100% | 99.4% | {Nicotine dependence, protection against}, 188890;{Nicotine dependence, susceptibility to}, 188890;Developmental and epileptic encephalopathy 59, 617904;Neurodevelopmental disorder with poor language and loss of hand skills, 617903 |
| GABRA1 | 100% | 100% | 100% | 100% | 99.4% | {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136;Developmental and epileptic encephalopathy 19, 615744;{Epilepsy, childhood absence, susceptibility to, 4}, 611136 |
| GABRA2 | 95% | 95% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 78, 618557;{Alcohol dependence, susceptibility to}, 103780 |
| GABRA3 | 100% | 100% | 98.7% | 90.1% | 71% | Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091 |
| GABRA4 | 100% | 100% | 100% | 100% | 99.9% | |
| GABRA5 | 100% | 100% | 100% | 99.9% | 99.5% | Developmental and epileptic encephalopathy 79, 618559 |
| GABRB1 | 100% | 100% | 100% | 100% | 99.9% | Developmental and epileptic encephalopathy 45, 617153 |
| GABRB2 | 100% | 100% | 100% | 100% | 99.9% | Developmental and epileptic encephalopathy 92, 617829 |

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| GABRB3 | 100% | 100% | 100% | 100% | 99.7% | {Epilepsy, childhood absence, susceptibility to, 5}, 612269;Developmental and epileptic encephalopathy 43, 617113 |
| GABRD | 100% | 100% | 100% | 99.7% | 98.3% | {?Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060 |
| GABRG2 | 92.9% | 92.9% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 74, 618396;Febrile seizures, familial, 8, 607681;Generalized epilepsy with febrile seizures plus, type 3, 607681 |
| GAD1 | 100% | 100% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 89, 619124 |
| GALC | 100% | 100% | 100% | 100% | 99.6% | Krabbe disease, 245200 |
| GALE | 100% | 100% | 100% | 100% | 99.6% | Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350 |
| GALNT2 | 100% | 100% | 100% | 100% | 99.4% | Congenital disorder of glycosylation, type IIt, 618885 |
| GALT | 100% | 100% | 100% | 100% | 99.5% | Galactosemia, 230400 |
| GAMT | 97% | 93.2% | 100% | 99.8% | 98.8% | Cerebral creatine deficiency syndrome 2, 612736 |
| GATAD2A | 100% | 100% | 100% | 99.9% | 99.5% | |
| GATAD2B | 100% | 100% | 100% | 100% | 99.4% | GAND syndrome, 615074 |

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| GATM | 100% | 100% | 100% | 100% | 99.9% | Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600 |
| GBA1 | 100% | 98.3% | 100% | 100% | 99.5% | {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 |
| GCH1 | 100% | 100% | 100% | 100% | 99.5% | Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910 |
| GCSH | 100% | 100% | 100% | 100% | 99.9% | Multiple mitochondrial dysfunctions syndrome 7, 620423 |
| GDI1 | 100% | 100% | 99% | 88.1% | 69.4% | Intellectual developmental disorder, X-linked 41, 300849 |
| GEMIN4 | 100% | 100% | 100% | 100% | 99.5% | Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913 |
| GEMIN5 | 100% | 100% | 100% | 99.9% | 99.6% | Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 |
| GFAP | 100% | 100% | 100% | 99.9% | 99.4% | Alexander disease, 203450 |

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| GFER | 100% | 100% | 100% | 99.9% | 99.4% | Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 |
| GFM1 | 96.6% | 96.6% | 100% | 100% | 99.7% | Combined oxidative phosphorylation deficiency 1, 609060 |
| GFM2 | 100% | 100% | 100% | 100% | 99.9% | Combined oxidative phosphorylation deficiency 39, 618397 |
| GIGYF1 | 100% | 100% | 100% | 100% | 99% | |
| GJA1 | 100% | 100% | 100% | 100% | 99.7% | Erythrokeratodermia variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850 |
| GJB1 | 100% | 100% | 97.9% | 84.9% | 64.7% | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 |
| GJC2 | 100% | 99.4% | 100% | 99.9% | 98.3% | Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804 |
| GK | 96% | 96% | 99.5% | 92.1% | 73.6% | Glycerol kinase deficiency, 307030 |

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| GLB1 | 100% | 100% | 100% | 100% | 99.5% | GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600 |
| GLDC | 97.3% | 97.3% | 100% | 100% | 99.6% | Glycine encephalopathy1, 605899 |
| GLI2 | 100% | 100% | 100% | 99.9% | 99.3% | Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829 |
| GLI3 | 99.3% | 99.3% | 100% | 99.9% | 99.2% | Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700 |
| GLIS3 | 100% | 100% | 100% | 100% | 99.6% | Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 |
| GLRA2 | 100% | 100% | 99.2% | 91.6% | 73.5% | Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076 |
| GLS | 95.8% | 95.8% | 100% | 100% | 99.8% | CASGID syndrome, 618339;Global developmental delay, progressive ataxia, and elevated glutamine, 618412;Developmental and epileptic encephalopathy 71, 618328 |
| GLUD1 | 100% | 100% | 100% | 100% | 99.8% | Hyperinsulinism-hyperammonemia syndrome, 606762 |

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| GLUL | 100% | 100% | 100% | 99.9% | 99.2% | Glutamine deficiency, congenital, 610015;Developmental and epileptic encephalopathy 116, 620806 |
| GLYCTK | 100% | 100% | 100% | 99.9% | 99.3% | D-glyceric aciduria, 220120 |
| GM2A | 100% | 100% | 100% | 100% | 99.7% | GM2-gangliosidosis, AB variant, 272750 |
| GMNN | 91.4% | 91.4% | 100% | 100% | 99.8% | Meier-Gorlin syndrome 6, 616835 |
| GMPPA | 100% | 100% | 100% | 100% | 98.8% | Alacrima, achalasia, and impaired intellectual development syndrome, 615510 |
| GMPPB | 100% | 100% | 100% | 100% | 99.7% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 |
| GNAI1 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854 |
| GNAO1 | 100% | 100% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493 |

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| GNAS | 100% | 99.7% | 100% | 99.9% | 98.5% | Pituitary adenoma 3, multiple types, somatic, 617686;Pseudohypoparathyroidism Ic, 612462;Pseudohypoparathyroidism Ia, 103580;Osseous heteroplasia, progressive, 166350;McCune-Albright syndrome, somatic mosaic, 174800;Pseudohypoparathyroidism Ib, 603233;Pseudopseudohypoparathyroidism, 612463;ACTH-independent macronodular adrenal hyperplasia 1, somatic, 219080 |
| GNB1 | 94.3% | 94.3% | 100% | 100% | 99.5% | Myelodysplastic syndrome, somatic, 614286;Leukemia, acute lymphoblastic, somatic, 613065;Intellectual developmental disorder, autosomal dominant 42, 616973 |
| GNB2 | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464 |
| GNB5 | 100% | 100% | 100% | 100% | 99.8% | Lodder-Merla syndrome, type 2, with developmental delay and with or without cardiac arrhythmia, 617182;Lodder-Merla syndrome, type 1, with impaired intellectual development and cardiac arrhythmia, 617173 |
| GNPAT | 100% | 100% | 100% | 100% | 100% | Rhizomelic chondrodysplasia punctata, type 2, 222765 |

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| GNPTAB | 100% | 100% | 100% | 100% | 99.8% | Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500 |
| GNPTG | 90.1% | 90.1% | 100% | 99.8% | 98.5% | Mucolipidosis III gamma, 252605 |
| GNS | 94.3% | 94.3% | 100% | 100% | 99.6% | Mucopolysaccharidosis type IIID, 252940 |
| GOLGA2 | 100% | 100% | 100% | 99.9% | 99.1% | Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240 |
| GON4L | 100% | 100% | 100% | 100% | 99.7% | Li-Takada-Miyake syndrome, 621212 |
| GOT2 | 92.8% | 92.8% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 82, 618721 |
| GPAA1 | 88.5% | 88.5% | 100% | 100% | 99.4% | Glycosylphosphatidylinositol biosynthesis defect 15, 617810 |
| GPATCH11 | 100% | 100% | 100% | 100% | 99.8% | |
| GPC3 | 94% | 94% | 98.6% | 88.7% | 70.8% | Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870 |
| GPC4 | 100% | 100% | 99.4% | 91.2% | 70.7% | Keipert syndrome, 301026 |
| GPHN | 95.9% | 95.9% | 100% | 100% | 99.8% | Molybdenum cofactor deficiency C, 615501 |
| GPRC5B | 100% | 100% | 100% | 100% | 99.9% | Megalencephalic leukoencephalopathy with subcortical cysts 3, 620447 |
| GPSM2 | 95.5% | 95.5% | 100% | 100% | 99.8% | Chudley-McCullough syndrome, 604213 |
| GPT2 | 92.9% | 92.9% | 100% | 100% | 99.7% | Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 |

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| GRIA1 | 100% | 100% | 100% | 100% | 99.7% | ?Intellectual developmental disorder, autosomal recessive 76, 619931;Intellectual developmental disorder, autosomal dominant 67, 619927 |
| GRIA2 | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917 |
| GRIA3 | 100% | 100% | 99.2% | 91.4% | 73.7% | Intellectual developmental disorder, X-linked syndromic, Wu type, 300699 |
| GRIA4 | 100% | 100% | 100% | 99.9% | 99.5% | Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 |
| GRID2 | 100% | 100% | 100% | 100% | 99.8% | Spinocerebellar ataxia, autosomal recessive 18, 616204 |
| GRIK2 | 95.3% | 95.3% | 100% | 99.9% | 99.3% | Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580;Intellectual developmental disorder, autosomal recessive 6, 611092 |
| GRIN1 | 100% | 100% | 100% | 100% | 99.2% | 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 |

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| GRIN2A | 100% | 100% | 100% | 100% | 99.5% | Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570 |
| GRIN2B | 100% | 100% | 100% | 100% | 99.6% | Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 |
| GRIN2D | 100% | 99.9% | 100% | 99.8% | 97.9% | Developmental and epileptic encephalopathy 46, 617162 |
| GRIP1 | 100% | 100% | 100% | 100% | 99.9% | Fraser syndrome 3, 617667 |
| GRM1 | 100% | 100% | 100% | 100% | 99.6% | Spinocerebellar ataxia, autosomal recessive 13, 614831;Spinocerebellar ataxia 44, 617691 |
| GRM7 | 100% | 100% | 100% | 99.8% | 98.4% | Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 |
| GRN | 100% | 100% | 100% | 100% | 99.7% | Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706 |
| GSE1 | 100% | 100% | 100% | 99.9% | 99.2% | |
| GSK3B | 100% | 100% | 100% | 100% | 99.8% | |
| GSPT2 | 100% | 100% | 98.9% | 90.3% | 74.5% | |

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| GSS | 100% | 100% | 100% | 99.9% | 99.5% | Anemia, congenital, nonspherocytic hemolytic, 6, glutatione synthetase deficient, 231900;Glutathione synthetase deficiency, 266130 |
| GSX2 | 100% | 100% | 100% | 100% | 99.4% | Diencephalic-mesence phalic junction dysplasia syndrome 2, 618646 |
| GTF2E2 | 78.7% | 78.7% | 100% | 100% | 99.9% | Trichothiodystrophy 6, nonphotosensitive, 616943 |
| GTF2H5 | 44.2% | 44.2% | 100% | 100% | 99.8% | Trichothiodystrophy 3, photosensitive, 616395 |
| GTF3C3 | 98.6% | 98.5% | 100% | 100% | 99.7% | Neurodevelopmental disorder with dysmorphic facies, brain anomalies, and seizures, 621201 |
| GTF3C5 | 100% | 100% | 100% | 99.9% | 99.4% | |
| GTPBP1 | 99.9% | 98.3% | 100% | 99.9% | 98.8% | Neurodevelopmental disorder with characteristic facial and ectodermal features and tetraparesis 1, 620888 |
| GTPBP2 | 100% | 100% | 100% | 100% | 99.9% | Jaberi-Elahi syndrome, 617988 |
| GTPBP3 | 100% | 100% | 100% | 99.9% | 99.3% | Combined oxidative phosphorylation deficiency 23, 616198 |
| GUSB | 98.4% | 98.4% | 100% | 100% | 99.1% | Mucopolysaccharidosis VII, 253220 |
| H1-4 | 100% | 100% | 100% | 100% | 99.3% | Rahman syndrome, 617537 |
| H3-3A | 100% | 100% | 100% | 99.8% | 98.9% | Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720 |
| H3-3B | 100% | 100% | 100% | 100% | 99.1% | Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721 |

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| H4C3 | 100% | 100% | 100% | 99.8% | 98.7% | Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758 |
| H4C5 | 100% | 100% | 100% | 100% | 98.3% | Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950 |
| H4C9 | 100% | 100% | 100% | 99.9% | 98.6% | Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951 |
| HAAO | 100% | 98.9% | 100% | 100% | 99.5% | Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 |
| HACE1 | 95.9% | 95.9% | 100% | 100% | 99.7% | Spastic paraplegia and psychomotor retardation with or without seizures, 616756 |
| HADH | 87.7% | 87.7% | 100% | 100% | 99.6% | Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 |
| 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 |
| HAX1 | 100% | 100% | 100% | 100% | 99.7% | Neutropenia, severe congenital 3, autosomal recessive, 610738 |
| HCCS | 100% | 100% | 99.3% | 91.9% | 75.5% | Linear skin defects with multiple congenital anomalies 1, 309801 |

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| HCFC1 | 100% | 100% | 98.5% | 88.6% | 69.8% | Methylmalonic aciduria and homocysteinemia, cblX type, 309541 |
| HCN1 | 100% | 100% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482 |
| HCN2 | 95.1% | 93.5% | 99.3% | 97.5% | 93% | Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477 |
| HDAC3 | 100% | 100% | 100% | 99.9% | 99.3% | |
| HDAC4 | 100% | 100% | 100% | 100% | 99.3% | Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797 |
| HDAC6 | 94.9% | 94.8% | 98.7% | 89.6% | 71.3% | ?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 |
| HDAC8 | 94.6% | 94.6% | 99.2% | 90.3% | 71.3% | Cornelia de Lange syndrome 5, 300882 |
| HEATR3 | 100% | 100% | 100% | 100% | 99.8% | Diamond-Blackfan anemia 21, 620072 |
| HEATR5B | 100% | 100% | 100% | 100% | 99.8% | |
| HECTD1 | 98.5% | 98.5% | 100% | 100% | 99.8% | |
| HECTD4 | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250 |

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| HECW2 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 |
| HEPACAM | 100% | 100% | 100% | 100% | 99.5% | Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926 |
| HERC1 | 100% | 100% | 100% | 100% | 99.7% | Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 |
| HERC2 | 100% | 100% | 100% | 99.9% | 99.5% | Intellectual developmental disorder, autosomal recessive 38, 615516;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 |
| HESX1 | 100% | 100% | 100% | 100% | 99.7% | Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230 |
| HEXA | 100% | 100% | 100% | 100% | 99.4% | [Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800 |
| HEXB | 100% | 100% | 100% | 99.9% | 99.5% | Sandhoff disease, infantile, juvenile, and adult forms, 268800 |

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| HGSNAT | 83.5% | 83.5% | 100% | 100% | 99.8% | Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544 |
| HIBCH | 100% | 100% | 100% | 100% | 100% | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 |
| HID1 | 100% | 100% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983 |
| HIKESHI | 100% | 100% | 100% | 100% | 99.4% | Leukodystrophy, hypomyelinating, 13, 616881 |
| HIVEP2 | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal dominant 43, 616977 |
| 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;Neurodevelop mental disorder with visual defects and brain anomalies, 618547 |
| HLCS | 100% | 100% | 100% | 100% | 99.2% | Holocarboxylase synthetase deficiency, 253270 |
| HMGB1 | 100% | 100% | 100% | 100% | 99.4% | |
| HMGCL | 76.2% | 76.2% | 100% | 100% | 99.5% | HMG-CoA lyase deficiency, 246450 |
| HNMT | 100% | 100% | 100% | 100% | 99.1% | Intellectual developmental disorder, autosomal recessive 51, 616739;{Asthma, susceptibility to}, 600807 |

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| HNRNPC | 87.6% | 87.6% | 100% | 100% | 99.5% | Intellectual developmental disorder, autosomal dominant 74, 620688 |
| HNRNPD | 100% | 100% | 100% | 100% | 99.9% | |
| HNRNPH1 | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083 |
| HNRNPH2 | 100% | 100% | 99.4% | 91.3% | 73.6% | Intellectual developmental disorder, X-linked syndromic, Bain type, 300986 |
| HNRNPK | 98.7% | 96% | 100% | 100% | 99.7% | Au-Kline syndrome, 616580 |
| HNRNPR | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073 |
| HNRNPU | 98.5% | 98.5% | 100% | 99.9% | 98.8% | Developmental and epileptic encephalopathy 54, 617391 |
| HOXA1 | 100% | 100% | 100% | 100% | 99.5% | Bosley-Salih-Alorainy syndrome, 601536;Athabaskan brainstem dysgenesis syndrome, 601536 |
| HPD | 93.7% | 90.9% | 100% | 99.9% | 98.9% | Hawkinsinuria, 140350;Tyrosinemia, type III, 276710 |
| HPDL | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027 |

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|---|---|---|---|---|---|---|
| HPRT1 | 87.6% | 87.4% | 99% | 91.6% | 74% | Hyperuricemia, HPRT-related, 300323;Lesch-Nyhan syndrome, 300322 |
| HRAS | 100% | 100% | 100% | 99.9% | 99.5% | Bladder cancer, somatic, 109800;Thyroid carcinoma, follicular, somatic, 188470;Congenital myopathy with excess of muscle spindles, 218040;Nevus sebaceous or woolly hair nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040 |
| HS2ST1 | 100% | 100% | 100% | 100% | 99.7% | Neurofacioskeletal syndrome with or without renal agenesis, 619194 |
| HS6ST2 | 100% | 100% | 98.8% | 87.6% | 70% | ?Paganini-Miozzo syndrome, 301025 |
| HSD17B10 | 100% | 100% | 99.5% | 91% | 71.8% | HSD10 mitochondrial disease, 300438 |
| HSD17B4 | 100% | 100% | 100% | 100% | 99.9% | D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400 |
| HSPA9 | 100% | 100% | 100% | 100% | 99.6% | Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170 |
| HSPD1 | 99.9% | 98.9% | 100% | 100% | 99.9% | Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233 |
| HTRA2 | 100% | 100% | 100% | 100% | 99.5% | {Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248 |

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|---|---|---|---|---|---|---|
| HUWE1 | 100% | 100% | 98.9% | 90.1% | 71.6% | Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 |
| HYCC1 | 100% | 100% | 100% | 100% | 100% | Leukodystrophy, hypomyelinating, 5, 610532 |
| HYLS1 | 100% | 100% | 100% | 100% | 99.8% | Hydrolethalus syndrome, 236680 |
| IARS1 | 100% | 100% | 100% | 100% | 99.6% | Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 |
| IARS2 | 100% | 99.7% | 100% | 99.9% | 99.6% | Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 |
| IBA57 | 100% | 100% | 100% | 100% | 99.5% | Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451 |
| IDS | 100% | 100% | 99.4% | 91.4% | 72.9% | Mucopolysaccharidosis II, 309900 |
| IDUA | 96% | 96% | 100% | 99.8% | 98.7% | Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014 |
| IER3IP1 | 90.2% | 74.9% | 100% | 100% | 99.3% | Microcephaly, epilepsy, and diabetes syndrome, 614231 |
| IFIH1 | 100% | 100% | 100% | 100% | 99.8% | Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250 |

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| IFT140 | 100% | 100% | 100% | 100% | 99.6% | {Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180 |
| IFT172 | 100% | 100% | 100% | 100% | 99.7% | Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 |
| IFT27 | 99.8% | 96.5% | 100% | 100% | 99.9% | Bardet-Biedl syndrome 19, 615996 |
| IFT74 | 100% | 100% | 100% | 100% | 100% | Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582 |
| IFT81 | 92.5% | 92.5% | 100% | 100% | 99.7% | Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 |
| IGBP1 | 100% | 100% | 98.8% | 91.9% | 72.1% | ?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472 |
| IGF1 | 100% | 100% | 100% | 99.9% | 99.1% | Insulin-like growth factor I deficiency, 608747 |
| IGF1R | 100% | 100% | 100% | 100% | 99.7% | Insulin-like growth factor I, resistance to, 270450 |
| IGF2 | 100% | 100% | 100% | 100% | 99.7% | Silver-Russell syndrome 3, 616489 |

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|---|---|---|---|---|---|---|
| IKBKG | 94.1% | 93.5% | 99% | 90.5% | 73.6% | Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081 |
| IKZF2 | 100% | 100% | 100% | 100% | 99.9% | Immunodysregulation, craniofacial anomalies, hearing impairment, athelia, and developmental delay, 621234;Immunodysregulation with variable immunodeficiency and autoimmunity, 621233 |
| IL1RAPL1 | 100% | 100% | 99.2% | 90.4% | 71.4% | Intellectual developmental disorder, X-linked 21, 300143 |
| IMPA1 | 81.6% | 81.6% | 100% | 100% | 99.9% | Intellectual developmental disorder, autosomal recessive 59, 617323 |
| INPP4A | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with growth impairment, quadriparesis, and poor or absent speech, 621354 |
| INPP5E | 100% | 100% | 100% | 99.8% | 98.1% | Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300 |
| INPP5K | 100% | 98.5% | 100% | 100% | 99.8% | Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 |

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| INTS1 | 99.6% | 98.7% | 100% | 100% | 99.4% | Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 |
| INTS11 | 100% | 100% | 100% | 99.9% | 98.6% | Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428 |
| INTS13 | 100% | 100% | 100% | 100% | 99.9% | |
| INTS6 | 100% | 100% | 100% | 100% | 99.7% | |
| IPO8 | 97% | 97% | 100% | 100% | 99.8% | VISS syndrome, 619472 |
| IQSEC1 | 100% | 100% | 100% | 99.8% | 98.6% | Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 |
| IQSEC2 | 99.1% | 97.9% | 97.5% | 86% | 67.2% | Intellectual developmental disorder, X-linked 1, 309530;Neurodevelopmental disorder, X-linked, with poor or absent speech and behavioral abnormalities, 301164 |
| IREB2 | 98.6% | 98.6% | 100% | 100% | 99.8% | Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 |
| IRF2BPL | 100% | 100% | 99.8% | 98.6% | 94.6% | Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 |
| IRX5 | 100% | 100% | 99.9% | 99.6% | 97.1% | Hamamy syndrome, 611174 |
| ISCA2 | 100% | 100% | 100% | 100% | 99.7% | Multiple mitochondrial dysfunctions syndrome 4, 616370 |

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| ITGA7 | 100% | 99.9% | 100% | 100% | 99.5% | Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 |
| ITGAV | 99.5% | 99.5% | 100% | 100% | 99.8% | Immune dysregulation, neurodevelopmental defects, and colitis, 621375 |
| ITPA | 76.9% | 76.9% | 100% | 99.9% | 99.4% | [Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647 |
| ITPR1 | 100% | 100% | 100% | 100% | 99.6% | Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658 |
| ITSN1 | 100% | 100% | 100% | 100% | 99.7% | |
| IVD | 90% | 90% | 100% | 100% | 99.6% | Isovaleric acidemia, 243500 |
| 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 |
| JAG2 | 100% | 100% | 100% | 100% | 99.1% | Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566 |
| JAM3 | 98.1% | 93.6% | 100% | 100% | 99.5% | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 |
| JARID2 | 100% | 100% | 100% | 100% | 99.5% | Developmental delay with variable intellectual disability and dysmorphic facies, 620098 |

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| JKAMP | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with seizures and impaired intellectual and language development, 621533 |
| JMJD1C | 100% | 100% | 100% | 100% | 99.6% | |
| KANK1 | 98.1% | 98.1% | 100% | 100% | 99.7% | Cerebral palsy, spastic quadriplegic, 2, 612900 |
| KANSL1 | 98.1% | 98.1% | 100% | 100% | 99.8% | Koolen-De Vries syndrome, 610443 |
| 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 |
| KAT5 | 100% | 100% | 100% | 100% | 99.3% | Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103 |
| KAT6A | 100% | 100% | 100% | 100% | 99.7% | Arboleda-Tham syndrome, 616268 |
| KAT6B | 100% | 100% | 100% | 100% | 99.5% | SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170 |
| KAT8 | 100% | 100% | 100% | 100% | 99.5% | Li-Ghorgani-Weisz-Hubshman syndrome, 618974 |
| KATNB1 | 100% | 100% | 100% | 100% | 99.6% | Lissencephaly 6, with microcephaly, 616212 |
| KATNIP | 100% | 100% | 100% | 100% | 99.4% | Joubert syndrome 26, 616784 |
| KBTBD2 | 100% | 100% | 100% | 100% | 99.9% | |

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| KCNA2 | 100% | 100% | 100% | 100% | 99.3% | Developmental and epileptic encephalopathy 32, 616366 |
| KCNA3 | 100% | 100% | 100% | 99.9% | 99.2% | |
| KCNA4 | 100% | 100% | 100% | 100% | 99.8% | Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 |
| KCNB1 | 100% | 100% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 26, 616056 |
| KCNB2 | 100% | 100% | 100% | 100% | 99.5% | |
| KCNC1 | 100% | 100% | 100% | 99.8% | 99% | Epilepsy, progressive myoclonic 7, 616187 |
| KCNC2 | 100% | 100% | 100% | 100% | 99.3% | Developmental and epileptic encephalopathy 103, 619913 |
| KCNC3 | 100% | 99.5% | 99.8% | 98.4% | 93.4% | Spinocerebellar ataxia 13, 605259 |
| KCND1 | 100% | 100% | 98.6% | 86.3% | 67.7% | |
| KCND2 | 100% | 100% | 100% | 99.9% | 99.7% | |
| KCND3 | 100% | 100% | 100% | 99.9% | 99% | Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399 |
| KCNH1 | 98.6% | 98.6% | 100% | 100% | 99.8% | Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816 |
| KCNH5 | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 112, 620537 |
| KCNJ10 | 100% | 100% | 100% | 100% | 99.7% | Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780 |

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|---|---|---|---|---|---|---|
| KCNJ11 | 100% | 100% | 100% | 100% | 99.5% | Diabetes, permanent neonatal 2, with or without neurologic features, 618856;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820 |
| KCNJ6 | 100% | 100% | 100% | 100% | 99.8% | Keppen-Lubinsky syndrome, 614098 |
| KCNK3 | 100% | 100% | 100% | 100% | 99% | Developmental delay with sleep apnea, 621402;Pulmonary hypertension, primary, 4, 615344 |
| KCNK4 | 99.4% | 99.4% | 100% | 100% | 99.3% | Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381 |
| KCNK9 | 100% | 100% | 100% | 100% | 99.8% | Birk-Barel syndrome, 612292 |
| KCNMA1 | 100% | 100% | 100% | 99.9% | 99.4% | {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 |

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| KCNN2 | 100% | 100% | 100% | 99.9% | 98.9% | ?Dystonia 34, myoclonic, 619724;Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 |
| KCNN3 | 100% | 100% | 100% | 99.9% | 99% | Zimmermann-Laband syndrome 3, 618658 |
| KCNQ2 | 94.3% | 94.1% | 100% | 100% | 99.3% | Developmental and epileptic encephalopathy 7, 613720;Seizures, benign neonatal, 1, 121200;Myokymia, 121200 |
| KCNQ3 | 100% | 100% | 100% | 100% | 99.6% | Seizures, benign neonatal, 2, 121201 |
| KCNQ5 | 100% | 100% | 100% | 100% | 99.2% | Intellectual developmental disorder, autosomal dominant 46, 617601 |
| KCNT1 | 100% | 100% | 100% | 99.9% | 99.2% | Developmental and epileptic encephalopathy 14, 614959;Epilepsy nocturnal frontal lobe, 5, 615005 |
| KCNT2 | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 57, 617771 |
| KCTD3 | 100% | 100% | 100% | 100% | 99.8% | |
| KCTD7 | 100% | 100% | 100% | 100% | 99.9% | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 |

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| KDM1A | 93.5% | 93.5% | 100% | 100% | 99.6% | Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990 |
| KDM2A | 100% | 100% | 100% | 100% | 99.5% | |
| KDM2B | 95.9% | 94% | 100% | 99.9% | 98.8% | Neurodevelopmental disorder with congenital cardiac defects and variable renal and ocular abnormalities, 621474 |
| KDM3B | 100% | 100% | 100% | 100% | 99.6% | Diets-Jongmans syndrome, 618846 |
| KDM4B | 100% | 100% | 100% | 99.9% | 99.2% | Intellectual developmental disorder, autosomal dominant 65, 619320 |
| KDM5A | 100% | 100% | 100% | 100% | 99.8% | El Hayek-Chahrour neurodevelopmental syndrome, 620820 |
| KDM5B | 97.6% | 96.9% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal recessive 65, 618109 |
| KDM5C | 97.9% | 97.8% | 98.9% | 88.7% | 69.9% | Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 |
| KDM6A | 100% | 100% | 99% | 91.6% | 72.4% | Kabuki syndrome 2, 300867 |
| KDM6B | 100% | 100% | 100% | 100% | 98.9% | Stolerman neurodevelopmental syndrome, 618505 |
| KIAA0586 | 95.6% | 95.6% | 100% | 100% | 99.7% | Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490 |

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| KIAA0753 | 100% | 100% | 100% | 100% | 99.8% | ?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479 |
| KICS2 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 83, 621100 |
| KIDINS220 | 100% | 100% | 100% | 100% | 99.8% | Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501 |
| KIF11 | 100% | 100% | 100% | 100% | 99.8% | Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950 |
| KIF14 | 97.6% | 97.6% | 100% | 100% | 99.8% | Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258 |
| 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 |
| KIF21B | 100% | 100% | 100% | 100% | 99.5% | |
| KIF26A | 97.6% | 97.6% | 100% | 99.9% | 99.4% | Cortical dysplasia, complex, with other brain malformations 11, 620156 |

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| KIF2A | 97.9% | 97.1% | 100% | 99.9% | 99.6% | Cortical dysplasia, complex, with other brain malformations 3, 615411 |
| KIF3B | 100% | 100% | 100% | 100% | 99.6% | Retinitis pigmentosa 89, 618955 |
| KIF4A | 96.1% | 96.1% | 99.1% | 91.6% | 73.6% | Taurodontism, microdontia, and dens invaginatus, 313490;Intellectual developmental disorder, X-linked 100, 300923 |
| KIF5A | 100% | 100% | 100% | 100% | 99.5% | Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187 |
| KIF5B | 98.8% | 96.7% | 100% | 100% | 99.6% | |
| KIF5C | 100% | 100% | 100% | 100% | 99.5% | Cortical dysplasia, complex, with other brain malformations 2, 615282 |
| KIF7 | 100% | 100% | 100% | 100% | 99.2% | Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131 |
| KIFBP | 95.9% | 95.9% | 100% | 99.9% | 99.5% | Goldberg-Shprintzen megacolon syndrome, 609460 |
| KIRREL3 | 100% | 100% | 100% | 100% | 99.2% | |
| KLF7 | 100% | 100% | 100% | 100% | 99% | |
| KLHL13 | 100% | 100% | 99.3% | 93% | 75.6% | |

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| KLHL15 | 89.1% | 89.1% | 99.3% | 93.2% | 74.9% | Intellectual developmental disorder, X-linked 103, 300982 |
| KLHL20 | 93.7% | 93.7% | 100% | 100% | 99.9% | Neurodevelopmental disorder with early-onset seizures, facial dysmorphism, and behavioral abnormalities, 621390 |
| KLHL7 | 100% | 100% | 100% | 100% | 99.8% | Retinitis pigmentosa 42, 612943;PERCHING syndrome, 617055 |
| KMT2A | 99.2% | 99.2% | 100% | 100% | 99.5% | Wiedemann-Steiner syndrome, 605130 |
| KMT2B | 100% | 99.7% | 100% | 99.9% | 98.8% | Intellectual developmental disorder, autosomal dominant 68, 619934;Dystonia 28, childhood-onset, 617284 |
| KMT2C | 100% | 100% | 100% | 100% | 99.7% | Kleefstra syndrome 2, 617768 |
| KMT2D | 100% | 100% | 100% | 100% | 99.2% | Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920 |
| KMT2E | 100% | 100% | 100% | 100% | 99.5% | O'Donnell-Luria-Rodan syndrome, 618512 |
| KMT5B | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 51, 617788 |
| KNL1 | 98.7% | 98.7% | 100% | 100% | 99.8% | Microcephaly 4, primary, autosomal recessive, 604321 |
| KPTN | 100% | 100% | 100% | 100% | 99% | Intellectual developmental disorder, autosomal recessive 41, 615637 |

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| KRAS | 100% | 100% | 100% | 100% | 100% | Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;RAS-associated autoimmune leukoproliferative disorder, 614470;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;Bladder cancer, somatic, 109800 |
| L1CAM | 100% | 100% | 98.4% | 88.2% | 69.2% | MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100 |
| L2HGDH | 92.8% | 92.8% | 100% | 100% | 99.6% | L-2-hydroxyglutaric aciduria, 236792 |
| LAGE3 | 100% | 100% | 98.7% | 89% | 67.1% | Galloway-Mowat syndrome 2, X-linked, 301006 |
| LAMA1 | 98.9% | 98.9% | 100% | 100% | 99.6% | Poretti-Boltshauser syndrome, 615960 |

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| LAMA2 | 100% | 100% | 100% | 100% | 99.8% | Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 |
| LAMB1 | 100% | 100% | 100% | 100% | 99.7% | Leukoencephalopathy without lacunae, adult-onset, 621424;Leukoencephalopathy with variable cortical brain malformations and/or hydrocephalus, 615191 |
| LAMB2 | 100% | 100% | 100% | 100% | 99.7% | Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049 |
| LAMC3 | 100% | 100% | 100% | 100% | 99.4% | Cortical malformations, occipital, 614115 |
| LAMP2 | 70.7% | 70.7% | 98.9% | 91% | 71.6% | Danon disease, 300257 |
| LARGE1 | 96.9% | 96.9% | 100% | 100% | 99.6% | Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 |
| LARP1 | 100% | 100% | 100% | 100% | 99.2% | |
| LARP7 | 100% | 100% | 100% | 100% | 99.8% | Alazami syndrome, 615071 |
| LARS1 | 94.3% | 94.3% | 100% | 100% | 99.7% | ?Infantile liver failure syndrome 1, 615438 |
| LARS2 | 96.2% | 96.2% | 100% | 100% | 99.8% | Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021 |

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|---|---|---|---|---|---|---|
| LAS1L | 96% | 96% | 98.3% | 88.6% | 69.8% | Wilson-Turner syndrome, 309585 |
| LDB1 | 100% | 100% | 100% | 99.9% | 99.5% | |
| LETM1 | 92.4% | 89.8% | 100% | 100% | 99.5% | Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 |
| LGI1 | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 121, 621475;Epilepsy, familial temporal lobe, 1, 600512 |
| LGI3 | 100% | 100% | 100% | 100% | 99.4% | Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects, 620007 |
| LHX2 | 100% | 100% | 100% | 99.9% | 99.3% | |
| LIAS | 100% | 100% | 100% | 100% | 99.9% | Hyperglycinemia, lactic acidosis, and seizures, 614462 |
| LIG4 | 100% | 100% | 100% | 100% | 99.8% | LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500 |
| LINGO1 | 100% | 100% | 100% | 99.9% | 98.1% | Intellectual developmental disorder, autosomal recessive 64, 618103 |
| LINS1 | 100% | 100% | 100% | 100% | 99.9% | Intellectual developmental disorder, autosomal recessive 27, 614340 |
| LIPT2 | 100% | 100% | 100% | 100% | 99% | Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 |

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| LMAN2L | 100% | 100% | 100% | 100% | 99.9% | ?Intellectual developmental disorder, autosomal dominant 69, 617863;?Intellectual developmental disorder, autosomal recessive 52, 616887 |
| LMBRD1 | 86.4% | 86.4% | 100% | 99.9% | 99.6% | Methylmalonic aciduria and homocystinuria, cblF type, 277380 |
| LMBRD2 | 100% | 100% | 100% | 100% | 99.9% | Developmental delay with variable neurologic and brain abnormalities, 619694 |
| LMNB1 | 100% | 100% | 100% | 99.9% | 99.2% | Microcephaly 26, primary, autosomal dominant, 619179 |
| LMNB2 | 92.9% | 92.9% | 100% | 99.8% | 98.3% | Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709 |
| LNPK | 85.8% | 85.8% | 100% | 100% | 99.8% | Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090 |
| LONP1 | 99.3% | 97% | 100% | 100% | 99.5% | CODAS syndrome, 600373 |
| LRP2 | 100% | 100% | 100% | 100% | 99.8% | Donnai-Barrow syndrome, 222448 |
| LRPPRC | 96.8% | 96.7% | 100% | 100% | 99.8% | Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 |
| LRRC7 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 77, 621415 |

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| LSM1 | 100% | 100% | 100% | 100% | 99.2% | FICUS syndrome, 621193 |
| LSS | 100% | 100% | 100% | 100% | 99.8% | Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840 |
| LTBP1 | 100% | 100% | 100% | 100% | 99.5% | Cutis laxa, autosomal recessive, type IIE, 619451 |
| LYRM7 | 100% | 100% | 100% | 100% | 99.4% | Mitochondrial complex III deficiency, nuclear type 8, 615838 |
| LYST | 99.5% | 99.5% | 100% | 100% | 99.9% | Chediak-Higashi syndrome, 214500 |
| LZTFL1 | 100% | 100% | 100% | 100% | 99.9% | Bardet-Biedl syndrome 17, 615994 |
| LZTR1 | 100% | 100% | 100% | 100% | 99.6% | Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670 |
| MAB21L1 | 100% | 100% | 100% | 100% | 99.3% | Cerebellar, ocular, craniofacial, and genital syndrome, 618479 |
| MAB21L2 | 100% | 100% | 100% | 100% | 99.2% | Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 |
| MACF1 | 100% | 100% | 100% | 100% | 99.7% | Lissencephaly 9 with complex brainstem malformation, 618325 |
| MADD | 98.7% | 98.7% | 100% | 100% | 99.5% | Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005;DEEAH syndrome, 619004 |
| MAF | 95.1% | 91.7% | 100% | 99.4% | 96.8% | Cataract 21, multiple types, 610202;Ayme-Gripp syndrome, 601088 |

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| MAG | 100% | 100% | 100% | 100% | 99.3% | Spastic paraplegia 75, autosomal recessive, 616680 |
| MAGEL2 | 100% | 100% | 100% | 100% | 99.7% | Schaaf-Yang syndrome, 615547 |
| MAN1B1 | 99.8% | 98.1% | 100% | 100% | 99.6% | Rafiq syndrome, 614202 |
| MAN2A2 | 100% | 100% | 100% | 100% | 99.4% | |
| MAN2B1 | 100% | 100% | 100% | 100% | 99.1% | Mannosidosis, alpha-, types I and II, 248500 |
| MAN2C1 | 98.4% | 98.4% | 100% | 99.9% | 99.5% | Congenital disorder of deglycosylation 2, 619775 |
| MANBA | 99.4% | 97.5% | 100% | 100% | 99.5% | Mannosidosis, beta, 248510 |
| MAOA | 100% | 100% | 99.6% | 92.1% | 73.7% | Brunner syndrome, 300615 |
| MAP1B | 100% | 100% | 100% | 100% | 99.6% | ?Deafness, autosomal dominant 83, 619808;Periventricular nodular heterotopia 9, 618918 |
| MAP2K1 | 95.8% | 95.8% | 100% | 100% | 99.5% | Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950 |
| MAP2K2 | 92.4% | 92.3% | 100% | 100% | 99.5% | Cardiofaciocutaneous syndrome 4, 615280 |
| MAP2K4 | 100% | 100% | 100% | 100% | 99.7% | |
| MAP4K4 | 100% | 100% | 100% | 100% | 99.7% | |
| MAPK1 | 100% | 100% | 100% | 99.9% | 99.4% | Noonan syndrome 13, 619087 |
| MAPK8IP3 | 100% | 100% | 100% | 100% | 99.4% | Neurodevelopmental disorder with or without variable brain abnormalities, 618443 |
| MAPKAPK5 | 100% | 100% | 100% | 100% | 99.4% | Neurocardiofaciodigital syndrome, 619869 |

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|---|---|---|---|---|---|---|
| MAPRE2 | 100% | 100% | 100% | 100% | 99.8% | Symmetric circumferential skin creases, congenital, 2, 616734 |
| MARK2 | 100% | 100% | 100% | 100% | 99.2% | Intellectual developmental disorder, autosomal dominant 76, 621285 |
| 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 |
| MASP1 | 100% | 100% | 100% | 100% | 99.6% | 3MC syndrome 1, 257920 |
| MAST1 | 100% | 100% | 100% | 100% | 99.3% | Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 |
| MAST3 | 100% | 100% | 100% | 99.9% | 99% | Developmental and epileptic encephalopathy 108, 620115 |
| MAST4 | 100% | 100% | 100% | 100% | 99.5% | |
| MAT1A | 100% | 99.9% | 100% | 99.9% | 99.4% | Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850;Methionine adenosyltransferase deficiency, autosomal recessive, 250850 |
| MAU2 | 100% | 100% | 100% | 100% | 99.5% | Cornelia de Lange syndrome 7, 621570 |

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| MAX | 100% | 100% | 100% | 100% | 99.7% | Polydactyly-macrocephaly syndrome, 620712;{Pheochromocytoma, susceptibility to}, 171300 |
| MBD5 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 1, 156200 |
| MBOAT7 | 100% | 100% | 100% | 100% | 99.6% | Intellectual developmental disorder, autosomal recessive 57, 617188 |
| MBTPS2 | 94.6% | 94.6% | 99.3% | 91.4% | 73.6% | Keratosis follicularis spinulosa decalvans, X-linked, 308800;Osteogenesis imperfecta, type XIX, 301014;IFAP syndrome with or without BRESHECK syndrome, 308205;?Olmsted syndrome, X-linked, 300918 |
| MCCC1 | 94.5% | 94.5% | 100% | 100% | 99.8% | 3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 |
| MCCC2 | 93.5% | 93.5% | 100% | 100% | 99.9% | 3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 |
| MCM3AP | 100% | 100% | 100% | 100% | 99.5% | Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 |
| MCM6 | 100% | 100% | 100% | 100% | 99.8% | Lactase persistence/nonpersistence, 223100 |
| MCOLN1 | 98.3% | 95.7% | 100% | 100% | 99.7% | Lisch epithelial corneal dystrophy, 620763;Mucolipidosis IV, 252650 |
| MCPH1 | 94.6% | 92.8% | 100% | 100% | 99.7% | Microcephaly 1, primary, autosomal recessive, 251200 |

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| MDGA2 | 100% | 100% | 100% | 100% | 99.9% | |
| MDH2 | 100% | 100% | 100% | 100% | 98.7% | Developmental and epileptic encephalopathy 51, 617339 |
| MECP2 | 100% | 100% | 99% | 84.8% | 65.9% | 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 | 93.5% | 93.5% | 100% | 100% | 99.6% | ?Optic atrophy 16, 620629;Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 |
| MED11 | 100% | 100% | 100% | 100% | 99.7% | Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327 |
| MED12 | 100% | 99.9% | 98.1% | 87.5% | 68.9% | Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450 |
| MED12L | 100% | 100% | 100% | 100% | 99.7% | Nizon-Isidor syndrome, 618872 |
| MED13 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 61, 618009 |

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| MED13L | 100% | 100% | 100% | 100% | 99.6% | Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 |
| MED16 | 100% | 100% | 99.9% | 99.6% | 97.3% | Guillouet-Gordon syndrome, 621220 |
| MED17 | 100% | 100% | 100% | 100% | 99.6% | Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 |
| MED23 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249 |
| MED25 | 100% | 100% | 100% | 100% | 99.4% | Basel-Vanagaite-Smirin-Yosef syndrome, 616449 |
| MED27 | 100% | 100% | 100% | 100% | 99.5% | Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 |
| MEF2C | 100% | 100% | 100% | 100% | 99.7% | Chromosome 5q14.3 deletion syndrome, 613443;Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443 |
| MEGF8 | 100% | 100% | 100% | 99.8% | 98.9% | Carpenter syndrome 2, 614976 |
| MEIS2 | 89.6% | 88.5% | 100% | 100% | 99.6% | Cleft palate, cardiac defects, and impaired intellectual development, 600987 |
| METTL23 | 100% | 100% | 100% | 100% | 100% | Intellectual developmental disorder, autosomal recessive 44, 615942 |

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| METTL5 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 72, 618665 |
| MFF | 89.1% | 89.1% | 100% | 100% | 99.9% | Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 |
| MFSD2A | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 |
| MFSD8 | 100% | 100% | 100% | 100% | 100% | Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951 |
| MGAT2 | 100% | 100% | 100% | 99.8% | 99% | Congenital disorder of glycosylation, type IIa, 212066 |
| MGP | 100% | 100% | 100% | 100% | 99.7% | Keutel syndrome, 245150 |
| MIA3 | 100% | 100% | 100% | 100% | 99.7% | ?Odontochondrodysplasia 2 with hearing loss and diabetes, 619269 |
| MICOS13 | 100% | 100% | 100% | 100% | 99.6% | Combined oxidative phosphorylation deficiency 37, 618329 |
| MICU1 | 86.7% | 86.7% | 100% | 100% | 99.8% | Myopathy with extrapyramidal signs, 615673 |
| MID1 | 100% | 100% | 98.8% | 88.7% | 69.8% | Opitz GBBB syndrome, 300000 |
| MID2 | 100% | 100% | 98.9% | 90.9% | 72.5% | ?Intellectual developmental disorder, X-linked 101, 300928 |
| MIDEAS | 100% | 100% | 100% | 100% | 99.2% | |

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| MINPP1 | 100% | 100% | 100% | 100% | 99.9% | {Thyroid carcinoma, follicular}, 188470;Pontocerebellar hypoplasia, type 16, 619527 |
| MIR17HG | | | | | | |
| MKKS | 100% | 100% | 100% | 100% | 100% | McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231 |
| MKS1 | 99.1% | 99% | 100% | 100% | 99.7% | Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121 |
| MLC1 | 100% | 100% | 100% | 100% | 99.2% | Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004 |
| MLYCD | 100% | 100% | 100% | 100% | 99.4% | Malonyl-CoA decarboxylase deficiency, 248360 |
| MMAA | 100% | 100% | 100% | 100% | 99.8% | Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 |
| MMAB | 91.8% | 91.8% | 100% | 99.9% | 99.7% | Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 |
| MMACHC | 100% | 100% | 100% | 100% | 99.5% | Methylmalonic aciduria and homocystinuria, cblC type, 277400 |
| MMADHC | 79.3% | 79.3% | 100% | 100% | 99.7% | Methylmalonic aciduria and homocystinuria, cblD type, 277410;Methylmalonic aciduria, cblD type, 620953;Homocystinuria-megaloblastic anemia, cblD type, 620952 |
| MMGT1 | 100% | 100% | 99.4% | 92.8% | 75.4% | |
| MMUT | 100% | 100% | 100% | 100% | 99.7% | Methylmalonic aciduria, mut(0) type, 251000 |

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| MN1 | 100% | 100% | 100% | 99.9% | 98.5% | CEBALID syndrome, 618774;Meningioma, 607174 |
| MOCS1 | 100% | 100% | 100% | 100% | 99.4% | Molybdenum cofactor deficiency A, 252150 |
| MOCS2 | 100% | 100% | 100% | 100% | 99.9% | Molybdenum cofactor deficiency B1, 252160 |
| MOGS | 100% | 100% | 100% | 100% | 99.5% | Congenital disorder of glycosylation, type IIb, 606056 |
| 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 |
| MPDU1 | 100% | 100% | 100% | 100% | 98.8% | Congenital disorder of glycosylation, type If, 609180 |
| MPDZ | 100% | 100% | 100% | 100% | 99.7% | Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 |
| MPLKIP | 100% | 100% | 100% | 100% | 99% | Trichothiodystrophy 4, nonphotosensitive, 234050 |
| MPV17 | 100% | 100% | 100% | 100% | 99.6% | Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 |
| MRAS | 100% | 100% | 100% | 100% | 99.7% | Noonan syndrome 11, 618499 |
| MRPL49 | 100% | 100% | 100% | 100% | 99.3% | Combined oxidative phosphorylation deficiency 60, 621195 |

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| MRPS22 | 86.6% | 86.6% | 100% | 100% | 99.9% | Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719 |
| MRPS34 | 100% | 100% | 100% | 100% | 99.5% | Combined oxidative phosphorylation deficiency 32, 617664 |
| MRTFB | 100% | 100% | 100% | 100% | 99.8% | |
| MSL2 | 92.7% | 92.7% | 100% | 100% | 99.8% | Karayol-Borroto-Haghshenas neurodevelopmental syndrome, 620985 |
| MSL3 | 100% | 100% | 99.3% | 91.2% | 72.6% | Basilicata-Akhtar syndrome, 301032 |
| MSMO1 | 100% | 100% | 100% | 100% | 99.9% | Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 |
| MSTO1 | 100% | 100% | 99.9% | 99.1% | 97.7% | Myopathy, mitochondrial, and ataxia, 617675 |
| MTFMT | 91.8% | 91.8% | 100% | 99.9% | 98.6% | Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248 |
| MTHFR | 100% | 100% | 100% | 100% | 99.5% | {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% | 100% | 100% | 99.7% | Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 |

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| MTO1 | 94.8% | 91.9% | 100% | 100% | 99.7% | Combined oxidative phosphorylation deficiency 10, 614702 |
| MTOR | 100% | 100% | 100% | 100% | 99.6% | Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638 |
| MTR | 100% | 100% | 100% | 100% | 99.7% | {Neural tube defects, folate-sensitive, susceptibility to}, 601634;Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 |
| MTRFR | 100% | 100% | 100% | 100% | 99.7% | Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559 |
| MTRR | 100% | 100% | 100% | 100% | 99.7% | Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634 |
| MTSS2 | 100% | 100% | 100% | 99.9% | 99.1% | Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086 |
| MVK | 100% | 100% | 100% | 100% | 99.8% | Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377 |
| MYCBP2 | 100% | 99.8% | 100% | 100% | 99.8% | |
| MYCN | 100% | 100% | 100% | 100% | 98.9% | Feingold syndrome 1, 164280;Megalencephaly-polydactyly syndrome, 620748 |
| MYH10 | 99.5% | 99.5% | 100% | 100% | 99.6% | |

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| MYH9 | 97.3% | 97% | 100% | 100% | 99.4% | Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622 |
| MYO5A | 99% | 99% | 100% | 100% | 99.7% | Griscelli syndrome, type 1, 214450 |
| MYO9A | 100% | 100% | 100% | 100% | 99.7% | Myasthenic syndrome, congenital, 24, presynaptic, 618198 |
| MYT1L | 100% | 100% | 100% | 100% | 99.4% | Intellectual developmental disorder, autosomal dominant 39, 616521 |
| NAA10 | 100% | 100% | 99% | 89% | 68.4% | Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855 |
| NAA15 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 |
| NAA20 | 100% | 100% | 100% | 100% | 99.9% | Intellectual developmental disorder, autosomal recessive 73, 619717 |
| NAA60 | 100% | 100% | 100% | 100% | 99.5% | Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786 |
| NACC1 | 100% | 100% | 100% | 100% | 99.5% | Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 |
| NAE1 | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210 |

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| NAGA | 90% | 88% | 100% | 100% | 99.5% | Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241 |
| NAGLU | 100% | 100% | 100% | 100% | 99.4% | ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 |
| NALCN | 100% | 100% | 100% | 100% | 99.8% | Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 |
| NANS | 94.8% | 94.3% | 100% | 99.9% | 99.7% | Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 |
| NAPB | 100% | 100% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 107, 620033 |
| 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 |
| NARS2 | 92.3% | 92.3% | 100% | 100% | 99.7% | Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434 |

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| NAV3 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with poor or absent speech, dysmorphic facies, and behavioral abnormalities, 621182 |
| NAXE | 97.6% | 93.5% | 100% | 100% | 99.5% | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 |
| NBEA | 97.6% | 97.6% | 100% | 100% | 99.7% | Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157 |
| NBN | 97.5% | 97.5% | 100% | 100% | 99.8% | Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260 |
| NCAPG2 | 100% | 100% | 100% | 100% | 99.8% | Khan-Khan-Katsanis syndrome, 618460 |
| NCDN | 99.2% | 96.7% | 100% | 100% | 99.4% | Neurodevelopmental disorder with infantile epileptic spasms, 619373 |
| NCKAP1 | 100% | 100% | 100% | 100% | 99.7% | |
| NCOR2 | 100% | 100% | 100% | 99.9% | 98.9% | |
| NDC1 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with achalasia, polyneuropathy, and alacrima, 621328 |
| NDE1 | 93.5% | 93.5% | 100% | 100% | 99.1% | Microhydranencephaly, 605013;Lissencephaly 4 (with microcephaly), 614019 |
| NDP | 100% | 100% | 98.2% | 90.3% | 76.4% | Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600 |

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|---|---|---|---|---|---|---|
| NDST1 | 100% | 100% | 100% | 100% | 99.2% | Intellectual developmental disorder, autosomal recessive 46, 616116 |
| NDUFA1 | 100% | 100% | 98.8% | 90.1% | 70.9% | Mitochondrial complex I deficiency, nuclear type 12, 301020 |
| NDUFA11 | 96.1% | 95.9% | 100% | 100% | 99.2% | Mitochondrial complex I deficiency, nuclear type 14, 618236 |
| NDUFA12 | 80.4% | 80.4% | 100% | 99.9% | 99.7% | Mitochondrial complex I deficiency, nuclear type 23, 618244 |
| NDUFA2 | 100% | 100% | 100% | 100% | 99.7% | Mitochondrial complex I deficiency, nuclear type 13, 618235 |
| NDUFA8 | 100% | 100% | 100% | 100% | 99.6% | Mitochondrial complex I deficiency, nuclear type 37, 619272 |
| NDUFAF2 | 67.4% | 67.4% | 100% | 100% | 99.6% | Mitochondrial complex I deficiency, nuclear type 10, 618233 |
| NDUFAF3 | 100% | 100% | 100% | 100% | 99.9% | Mitochondrial complex I deficiency, nuclear type 18, 618240 |
| NDUFAF5 | 90.9% | 90.9% | 100% | 100% | 99.8% | Mitochondrial complex I deficiency, nuclear type 16, 618238 |
| NDUFAF8 | 100% | 100% | 100% | 100% | 99.4% | Mitochondrial complex I deficiency, nuclear type 34, 618776 |
| NDUFS1 | 100% | 100% | 100% | 100% | 99.8% | Mitochondrial complex I deficiency, nuclear type 5, 618226 |
| NDUFS2 | 99.9% | 98.3% | 100% | 100% | 99.6% | ?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228 |
| NDUFS3 | 98% | 92.5% | 100% | 100% | 99.5% | Mitochondrial complex I deficiency, nuclear type 8, 618230 |

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|---|---|---|---|---|---|---|
| NDUFS4 | 100% | 100% | 100% | 100% | 99.8% | Mitochondrial complex I deficiency, nuclear type 1, 252010 |
| NDUFS6 | 94.7% | 86.1% | 100% | 100% | 99.7% | Mitochondrial complex I deficiency, nuclear type 9, 618232 |
| NDUFS7 | 89.1% | 85% | 100% | 100% | 99.4% | Mitochondrial complex I deficiency, nuclear type 3, 618224 |
| NDUFS8 | 100% | 100% | 100% | 100% | 99.3% | Mitochondrial complex I deficiency, nuclear type 2, 618222 |
| NDUFV1 | 100% | 100% | 100% | 100% | 98.7% | Mitochondrial complex I deficiency, nuclear type 4, 618225 |
| NDUFV2 | 100% | 100% | 100% | 100% | 99.5% | Mitochondrial complex I deficiency, nuclear type 7, 618229 |
| NEDD4L | 100% | 100% | 100% | 100% | 99.6% | Periventricular nodular heterotopia 7, 617201 |
| NEMF | 100% | 100% | 100% | 99.8% | 99.3% | Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099 |
| NEU1 | 100% | 100% | 100% | 99.9% | 99.5% | Sialidosis, type II, 256550;Sialidosis, type I, 256550 |
| NEUROD2 | 100% | 100% | 100% | 99.8% | 97% | Developmental and epileptic encephalopathy 72, 618374 |
| NEUROG1 | 100% | 100% | 100% | 99.6% | 97% | Cranial dysinnervation disorder, congenital, with absent corneal reflex and developmental delay, 620469 |
| NEXMIF | 100% | 100% | 99.3% | 91.3% | 72.3% | Intellectual developmental disorder, X-linked 98, 300912 |

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| NF1 | 99.4% | 99.4% | 100% | 100% | 99.8% | Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321 |
| NFASC | 100% | 100% | 100% | 99.9% | 99.5% | Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 |
| NFE2L2 | 81.2% | 81.2% | 100% | 100% | 99.7% | Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 |
| NFIA | 92.2% | 92.2% | 100% | 99.9% | 99% | Brain malformations with or without urinary tract defects, 613735 |
| NFIB | 100% | 100% | 100% | 100% | 99.6% | Macrocephaly, acquired, with impaired intellectual development, 618286 |
| NFIX | 100% | 100% | 100% | 100% | 98.8% | Marshall-Smith syndrome, 602535;Malan syndrome, 614753 |
| NFU1 | 98.9% | 94.6% | 100% | 100% | 99.7% | Spastic paraplegia 93, autosomal recessive, 620938;Multiple mitochondrial dysfunctions syndrome 1, 605711 |
| NGLY1 | 98.4% | 97.2% | 100% | 100% | 99.8% | Congenital disorder of deglycosylation 1, 615273 |
| NHLRC2 | 100% | 100% | 100% | 100% | 99.8% | FINCA syndrome, 618278 |
| NHS | 100% | 100% | 98.9% | 90.1% | 71.8% | Cataract 40, X-linked, 302200;Nance-Horan syndrome, 302350 |

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| NIPBL | 100% | 100% | 100% | 100% | 99.8% | Cornelia de Lange syndrome 1, 122470 |
| NKAP | 100% | 100% | 99.1% | 89.5% | 71% | Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039 |
| NKX2-1 | 100% | 100% | 100% | 99.9% | 98.4% | Chorea, hereditary benign, 118700;{Thyroid cancer, nonmedullary, 1}, 188550;Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 |
| NKX6-2 | 100% | 100% | 100% | 99.6% | 97.1% | Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 |
| NLGN2 | 100% | 100% | 100% | 99.9% | 98.9% | |
| NLGN3 | 99.5% | 97.9% | 97.8% | 86.9% | 67.8% | {Autism susceptibility, X-linked 1}, 300425 |
| NLGN4X | 100% | 100% | 98.6% | 88.6% | 69.2% | Intellectual developmental disorder, X-linked, 300495;{Autism susceptibility, X-linked 2}, 300495 |
| NONO | 94.7% | 89.1% | 99.4% | 92.9% | 75.5% | Intellectual developmental disorder, X-linked syndromic 34, 300967 |
| NOVA2 | 100% | 100% | 99.8% | 98.6% | 94.2% | Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 |
| NPC1 | 100% | 100% | 100% | 100% | 99.7% | Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220 |

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| NPC2 | 99% | 95.2% | 100% | 100% | 99.6% | Niemann-pick disease, type C2, 607625 |
| NPHP1 | 100% | 100% | 100% | 100% | 99.8% | Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900 |
| NPTN | 85.5% | 85.5% | 100% | 100% | 99.4% | |
| NR2F1 | 100% | 100% | 100% | 99.9% | 98.5% | Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 |
| NR2F2 | 100% | 100% | 100% | 100% | 99.4% | 46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779 |
| NR4A2 | 100% | 100% | 100% | 99.9% | 99.4% | Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911 |
| NRAS | 89.4% | 89.4% | 100% | 100% | 99.9% | Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500 |

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| NRCAM | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833 |
| NRDC | 96.9% | 96.9% | 100% | 100% | 99.8% | |
| NRROS | 100% | 100% | 100% | 99.9% | 99.2% | Seizures, early-onset, with neurodegeneration and brain calcification, 618875 |
| NRXN1 | 100% | 100% | 100% | 100% | 99.5% | Pitt-Hopkins-like syndrome 2, 614325;{Schizophrenia, susceptibility to, 17}, 621407 |
| NSD1 | 100% | 100% | 100% | 100% | 99.5% | Sotos syndrome, 117550 |
| NSD2 | 99.5% | 99.5% | 100% | 100% | 99.6% | Rauch-Steindl syndrome, 619695 |
| NSDHL | 95.9% | 95.9% | 99.3% | 93.1% | 75.4% | CK syndrome, 300831;CHILD syndrome, 308050 |
| NSF | 100% | 100% | 99.8% | 98.1% | 93.5% | Developmental and epileptic encephalopathy 96, 619340 |
| NSRP1 | 91% | 91% | 100% | 100% | 99.5% | Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001 |
| NSUN2 | 98.5% | 98.5% | 100% | 100% | 99.5% | Intellectual developmental disorder, autosomal recessive 5, 611091 |
| NSUN6 | 100% | 100% | 100% | 100% | 99.9% | Intellectual developmental disorder, autosomal recessive 82, 620779 |
| NT5C2 | 91.4% | 91.4% | 100% | 100% | 99.8% | Spastic paraplegia 45, autosomal recessive, 613162 |

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| NTNG2 | 100% | 100% | 100% | 100% | 99.3% | Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 |
| NTRK1 | 100% | 100% | 100% | 99.9% | 99.3% | Insensitivity to pain, congenital, with anhidrosis, 256800 |
| NTRK2 | 89.5% | 89.5% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 58, 617830;Obesity, hyperphagia, and developmental delay, 613886 |
| NUBPL | 89.8% | 89.8% | 100% | 100% | 99.9% | Mitochondrial complex I deficiency, nuclear type 21, 618242 |
| NUDT2 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder with or without peripheral neuropathy, 619844 |
| NUP107 | 97.8% | 97.8% | 100% | 100% | 99.9% | ?Ovarian dysgenesis 6, 618078;Galloway-Mowat syndrome 7, 618348;Nephrotic syndrome, type 11, 616730 |
| NUP133 | 97.9% | 97.9% | 100% | 100% | 99.8% | ?Galloway-Mowat syndrome 8, 618349;Nephrotic syndrome, type 18, 618177 |
| NUP188 | 95.4% | 95.4% | 100% | 100% | 99.6% | Sandestig-Stefanova syndrome, 618804 |
| NUP214 | 100% | 100% | 100% | 100% | 99.5% | Leukemia, T-cell acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, somatic, 601626;{Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 |

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| NUP54 | 100% | 100% | 100% | 100% | 99.7% | Dystonia 37, early-onset, with striatal lesions, 620427 |
| NUP62 | 100% | 100% | 100% | 100% | 99.3% | Striatonigral degeneration, infantile, 271930 |
| NUP85 | 100% | 99.8% | 100% | 100% | 99.4% | Nephrotic syndrome, type 17, 618176 |
| NUS1 | 93.9% | 93.9% | 100% | 100% | 99.4% | Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082 |
| OAT | 100% | 100% | 100% | 99.9% | 99.3% | Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 |
| OCLN | 94.5% | 94.5% | 100% | 99.9% | 99.4% | Pseudo-TORCH syndrome 1, 251290 |
| OCRL | 100% | 100% | 99.3% | 92.4% | 74.8% | Dent disease 2, 300555;Lowe syndrome, 309000 |
| ODC1 | 100% | 100% | 100% | 100% | 99.8% | Bachmann-Bupp syndrome, 619075 |
| OFD1 | 100% | 100% | 99.1% | 91% | 73.2% | Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804 |
| OGDH | 100% | 100% | 100% | 100% | 99.5% | Oxoglutarate dehydrogenase deficiency, 203740 |
| OGDHL | 100% | 100% | 100% | 100% | 99.5% | |
| OGT | 100% | 100% | 99.2% | 92% | 73.4% | Intellectual developmental disorder, X-linked 106, 300997 |

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| OLA1 | 100% | 100% | 100% | 100% | 99.8% | |
| OPA3 | 100% | 100% | 100% | 100% | 99.6% | 3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300 |
| OPHN1 | 94% | 94% | 98.9% | 90.6% | 72.9% | Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 |
| ORC1 | 100% | 100% | 100% | 100% | 99.8% | Meier-Gorlin syndrome 1, 224690 |
| OSGEP | 100% | 100% | 100% | 100% | 99.8% | Galloway-Mowat syndrome 3, 617729 |
| OTC | 100% | 100% | 99.6% | 91.8% | 73.3% | Ornithine transcarbamylase deficiency, 311250 |
| OTUD5 | 100% | 100% | 97.7% | 84.3% | 65.5% | Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056 |
| OTUD6B | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 |
| OTUD7A | 99.9% | 99.1% | 100% | 99.9% | 97.8% | Neurodevelopmental disorder with hypotonia and seizures, 620790 |
| OTX2 | 100% | 100% | 100% | 100% | 99.9% | Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125 |
| OXR1 | 100% | 100% | 100% | 100% | 99.8% | Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 |

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| P4HTM | 89.4% | 89.4% | 100% | 99.9% | 99.2% | Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 |
| PABPC1 | 94.3% | 93.8% | 100% | 100% | 99.7% | |
| PACS1 | 100% | 100% | 100% | 100% | 99.3% | Schuurs-Hoeijmakers syndrome, 615009 |
| PACS2 | 78.8% | 78.8% | 100% | 99.9% | 99.4% | Developmental and epileptic encephalopathy 66, 618067 |
| PAFAH1B1 | 100% | 100% | 100% | 100% | 99.8% | Subcortical laminar heterotopia, 607432;Lissencephaly 1, 607432 |
| PAH | 94.9% | 93.7% | 100% | 100% | 99.8% | [Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600 |
| PAK1 | 100% | 100% | 100% | 100% | 99.9% | Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158 |
| PAK3 | 100% | 100% | 99.3% | 91.9% | 73.7% | Intellectual developmental disorder, X-linked 30, 300558 |
| PALS1 | 100% | 100% | 100% | 100% | 99.7% | |
| PAM16 | 85.8% | 85.5% | 100% | 100% | 99.3% | Spondylometaphyseal dysplasia, Megarbane-Dagher-Me like type, 613320 |
| PAN2 | 100% | 100% | 100% | 100% | 99.7% | Developmental delay with variable cardiac and renal congenital anomalies and dysmorphic facies, 621384 |

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| PANK2 | 100% | 100% | 100% | 100% | 99.6% | Neurodegeneration with brain iron accumulation 1, 234200 |
| PANX1 | 100% | 100% | 100% | 100% | 99.5% | Oocyte/zygote/embryo maturation arrest 7, 618550 |
| PARN | 97.2% | 96% | 100% | 100% | 99.8% | Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371 |
| PARP6 | 100% | 100% | 100% | 100% | 99.6% | |
| PARS2 | 100% | 100% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 75, 618437 |
| PAX1 | 100% | 100% | 100% | 100% | 98.6% | Otofaciocervical syndrome 2 with T-cell deficiency, 615560 |
| PAX7 | 94.2% | 94.2% | 100% | 99.9% | 99.1% | Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220 |
| PAX8 | 100% | 100% | 100% | 99.9% | 99.5% | Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 |
| PBX1 | 100% | 100% | 100% | 100% | 99.7% | Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 |
| PC | 99.9% | 99.2% | 100% | 100% | 99.5% | Pyruvate carboxylase deficiency, 266150 |
| PCCA | 84.6% | 84.4% | 100% | 100% | 99.7% | Propionicacidemia, 606054 |

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| PCCB | 86.8% | 85.3% | 100% | 100% | 99.8% | Propionicacidemia, 606054 |
| PCDH12 | 100% | 100% | 100% | 100% | 99.5% | Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 |
| PCDH19 | 100% | 100% | 98.2% | 86.7% | 65.6% | Developmental and epileptic encephalopathy 9, 300088 |
| PCDHGC4 | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880 |
| PCGF2 | 100% | 100% | 100% | 100% | 98.8% | Turnpenny-Fry syndrome, 618371 |
| PCLO | 100% | 100% | 100% | 100% | 99.5% | Pontocerebellar hypoplasia, type 3, 608027 |
| PCNT | 100% | 100% | 100% | 100% | 99.6% | Microcephalic osteodysplastic primordial dwarfism, type II, 210720 |
| PCYT2 | 99.9% | 97.1% | 100% | 100% | 99.5% | Spastic paraplegia 82, autosomal recessive, 618770 |
| PDCD6IP | 100% | 100% | 100% | 100% | 99.8% | ?Microcephaly 29, primary, autosomal recessive, 620047 |
| PDE2A | 100% | 100% | 100% | 100% | 99.3% | Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 |
| PDE4D | 100% | 100% | 100% | 100% | 99.2% | Acrodysostosis 2, with or without hormone resistance, 614613 |

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| PDGFRB | 99.3% | 97.4% | 100% | 100% | 99.2% | 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 | 91.4% | 89.5% | 99.3% | 91.4% | 72.9% | Pyruvate dehydrogenase E1-alpha deficiency, 312170 |
| PDHB | 100% | 100% | 100% | 100% | 99.8% | Pyruvate dehydrogenase E1-beta deficiency, 614111 |
| PDHX | 91.6% | 91.6% | 100% | 100% | 99.6% | Lacticacidemia due to PDX1 deficiency, 245349 |
| PDP1 | 100% | 100% | 100% | 100% | 99.9% | Pyruvate dehydrogenase phosphatase deficiency, 608782 |
| PDSS1 | 100% | 100% | 100% | 100% | 99.7% | Coenzyme Q10 deficiency, primary, 2, 614651 |
| PDSS2 | 100% | 100% | 100% | 100% | 99.4% | Coenzyme Q10 deficiency, primary, 3, 614652 |
| PDZD8 | 100% | 100% | 100% | 99.8% | 98.5% | Intellectual developmental disorder with autism and dysmorphic facies, 620021 |
| PEPD | 94% | 94% | 100% | 100% | 99.6% | Prolidase deficiency, 170100 |
| PET100 | 100% | 100% | 100% | 100% | 99.2% | Mitochondrial complex IV deficiency, nuclear type 12, 619055 |

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| 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 |
| PEX11B | 100% | 100% | 100% | 99.9% | 99.1% | Peroxisome biogenesis disorder 14B, 614920 |
| PEX12 | 100% | 100% | 100% | 100% | 99.5% | Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859 |
| PEX13 | 92.4% | 92.4% | 100% | 100% | 99.9% | Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885 |
| PEX16 | 100% | 100% | 100% | 100% | 99.6% | Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876 |
| PEX19 | 88.6% | 88.6% | 100% | 100% | 99.9% | Peroxisome biogenesis disorder 12A (Zellweger), 614886 |
| PEX2 | 100% | 100% | 100% | 100% | 99.7% | Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867 |
| PEX26 | 91.9% | 91.7% | 100% | 99.9% | 99.1% | Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872 |

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| PEX3 | 100% | 100% | 100% | 100% | 99.9% | Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370 |
| PEX5 | 100% | 100% | 100% | 99.8% | 99% | Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716 |
| PEX6 | 100% | 100% | 100% | 100% | 99.1% | Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617 |
| PEX7 | 97.9% | 97.9% | 100% | 100% | 99.6% | Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879 |
| PGAP1 | 96.7% | 96.7% | 100% | 100% | 99.9% | Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 |
| PGAP2 | 100% | 100% | 100% | 99.9% | 98.9% | Hyperphosphatasia with impaired intellectual development syndrome 3, 614207 |
| PGAP3 | 100% | 100% | 100% | 100% | 99.4% | Hyperphosphatasia with impaired intellectual development syndrome 4, 615716 |
| PGBD5 | 100% | 100% | 100% | 99.9% | 99.4% | Neurodevelopmental disorder with seizures, hypotonia, and variable spasticity, 621482 |

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|---|---|---|---|---|---|---|
| PGK1 | 92.7% | 92.7% | 99.3% | 90.9% | 71.5% | Phosphoglycerate kinase 1 deficiency, 300653 |
| PGM2L1 | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191 |
| PGM3 | 100% | 100% | 100% | 100% | 99.8% | Immunodeficiency 23, 615816 |
| PHACTR1 | 100% | 100% | 100% | 99.9% | 99.1% | Developmental and epileptic encephalopathy 70, 618298 |
| PHF21A | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 |
| PHF5A | 85% | 85% | 100% | 100% | 99.8% | |
| PHF6 | 100% | 100% | 99.4% | 92.6% | 74.6% | Borjeson-Forssman-Lehmann syndrome, 301900 |
| PHF8 | 100% | 100% | 98.9% | 90.4% | 71.6% | Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263 |
| PHGDH | 100% | 100% | 100% | 100% | 99.6% | Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815 |
| PHIP | 99.4% | 99.3% | 100% | 99.9% | 99.3% | Chung-Jansen syndrome, 617991 |
| PI4K2A | 86.9% | 86.9% | 100% | 100% | 99.1% | Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732 |

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|---|---|---|---|---|---|---|
| PI4KA | 99.3% | 99.2% | 100% | 99.9% | 99% | Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 |
| PIBF1 | 100% | 100% | 100% | 100% | 99.8% | Joubert syndrome 33, 617767 |
| PIDD1 | 100% | 100% | 100% | 100% | 99.6% | Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827 |
| PIGA | 100% | 100% | 99.2% | 90.8% | 73.4% | Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 |
| PIGB | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 80, 618580 |
| PIGC | 100% | 100% | 100% | 100% | 99.8% | Glycosylphosphatidylinositol biosynthesis defect 16, 617816 |
| PIGF | 100% | 100% | 100% | 100% | 99.8% | Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356 |

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|---|---|---|---|---|---|---|
| PIGG | 100% | 100% | 100% | 100% | 99.5% | [Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917 |
| PIGH | 82.9% | 76.2% | 100% | 100% | 99.8% | Glycosylphosphatidylinositol biosynthesis defect 17, 618010 |
| PIGK | 88.2% | 88.2% | 100% | 100% | 99.9% | Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 |
| PIGL | 100% | 100% | 100% | 100% | 99.7% | CHIME syndrome, 280000 |
| PIGN | 95.9% | 94.4% | 100% | 100% | 99.7% | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 |
| PIGO | 100% | 100% | 100% | 100% | 99.8% | Hyperphosphatasia with impaired intellectual development syndrome 2, 614749 |
| PIGP | 100% | 100% | 100% | 99.9% | 99.4% | Developmental and epileptic encephalopathy 55, 617599 |
| PIGQ | 100% | 100% | 100% | 100% | 99.6% | Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548 |
| PIGS | 98.4% | 95.1% | 100% | 99.9% | 99.1% | Developmental and epileptic encephalopathy 95, 618143 |
| PIGT | 100% | 100% | 100% | 100% | 99.5% | ?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 |

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|---|---|---|---|---|---|---|
| PIGU | 97.1% | 97.1% | 100% | 100% | 99.7% | Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 |
| PIGV | 100% | 100% | 100% | 100% | 99.7% | Hyperphosphatasia with impaired intellectual development syndrome 1, 239300 |
| PIGW | 100% | 100% | 100% | 100% | 99.9% | Glycosylphosphatidylinositol biosynthesis defect 11, 616025 |
| PIGY | 100% | 100% | 100% | 99.8% | 99.3% | Hyperphosphatasia with impaired intellectual development syndrome 6, 616809 |
| PIK3C2A | 100% | 100% | 100% | 100% | 99.9% | Oculoskeletodental syndrome, 618440 |

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|---|---|---|---|---|---|---|
| PIK3CA | 100% | 100% | 100% | 100% | 99.9% | Hemifacial myohyperplasia, somatic, 606773;CLOVE syndrome, somatic, 612918;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Cerebral cavernous malformations 4, somatic, 619538;Ovarian cancer, somatic, 167000;Colorectal cancer, somatic, 114500;Macrodactyly, somatic, 155500;CLAPO syndrome, somatic, 613089;Keratosis, seborrheic, somatic, 182000;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Nevus, epidermal, somatic mosaic, 162900;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108 |
| PIK3R2 | 100% | 100% | 100% | 100% | 99.2% | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 |
| PIP5K1C | 90.6% | 90.2% | 100% | 99.9% | 99.4% | Lethal congenital contractural syndrome 3, 611369 |
| PISD | 100% | 100% | 100% | 99.9% | 99.5% | Liberfarb syndrome, 618889 |
| PITRM1 | 100% | 100% | 100% | 100% | 99.6% | Spinocerebellar ataxia, autosomal recessive 30, 619405 |
| PJA1 | 100% | 100% | 98.3% | 86% | 67.4% | |

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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 |
| PLAA | 100% | 100% | 100% | 99.9% | 99.8% | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 |
| PLAAT3 | 65.6% | 63.1% | 100% | 100% | 99.7% | Lipodystrophy, familial partial, type 9, 620683 |
| PLCB1 | 100% | 100% | 100% | 100% | 99.6% | Developmental and epileptic encephalopathy 12, 613722 |
| PLEKHG2 | 100% | 100% | 100% | 100% | 99.4% | Leukodystrophy and acquired microcephaly with or without dystonia, 616763 |
| PLK1 | 100% | 100% | 100% | 100% | 99.3% | |
| PLK4 | 100% | 100% | 100% | 100% | 99.9% | Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 |
| PLP1 | 94.5% | 94.1% | 98.8% | 88.7% | 69.7% | Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920 |
| PLPBP | 100% | 100% | 100% | 99.8% | 98.6% | Epilepsy, early-onset, 1, vitamin B6-dependent, 617290 |
| PLXNA1 | 100% | 100% | 100% | 100% | 99.6% | Dworschak-Punetha neurodevelopmental syndrome, 619955 |
| PLXNA2 | 100% | 100% | 100% | 100% | 99.4% | |
| PLXNB2 | 100% | 100% | 100% | 100% | 99.1% | |

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| PLXND1 | 100% | 100% | 100% | 99.9% | 99.2% | Congenital heart defects, multiple types, 9, 620294 |
| PMM2 | 94.6% | 94.6% | 100% | 100% | 99.7% | Congenital disorder of glycosylation, type Ia, 212065 |
| PMPCA | 96.2% | 96.2% | 100% | 100% | 99.1% | Spinocerebellar ataxia, autosomal recessive 2, 213200 |
| PMPCB | 91.4% | 91.4% | 100% | 100% | 99.8% | Multiple mitochondrial dysfunctions syndrome 6, 617954 |
| 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 |
| PNP | 100% | 100% | 100% | 100% | 99.8% | Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 |
| PNPLA6 | 100% | 100% | 100% | 100% | 99.3% | Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470 |
| PNPLA8 | 100% | 100% | 100% | 100% | 99.5% | Mitochondrial myopathy with lactic acidosis, 251950 |
| PNPO | 100% | 100% | 100% | 100% | 99.6% | Pyridoxamine 5'-phosphate oxidase deficiency, 610090 |

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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 |
| POGZ | 100% | 100% | 100% | 100% | 99.6% | White-Sutton syndrome, 616364 |
| POLA1 | 100% | 100% | 99.3% | 91.7% | 73.3% | Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220;Van Esch-O'Driscoll syndrome, 301030 |
| 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 |
| POLR1A | 100% | 100% | 100% | 100% | 99.7% | Leukodystrophy, hypomyelinating, 27, 620675;Acrofacial dysostosis, Cincinnati type, 616462 |
| POLR1C | 83.6% | 83.5% | 100% | 100% | 99.2% | Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390 |

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| POLR2A | 100% | 100% | 100% | 100% | 99.2% | Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 |
| 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 |
| POLR3K | 100% | 100% | 100% | 100% | 99.6% | Leukodystrophy, hypomyelinating, 21, 619310 |
| POLRMT | 99.7% | 98.7% | 100% | 100% | 99% | Combined oxidative phosphorylation deficiency 55, 619743 |
| POMGNT1 | 100% | 100% | 100% | 100% | 99.6% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151;Retinitis pigmentosa 76, 617123;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 |

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| POMGNT2 | 100% | 100% | 100% | 100% | 99.5% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830;Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 |
| POMK | 100% | 100% | 100% | 100% | 99.8% | ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 |
| POMT1 | 100% | 99.7% | 100% | 100% | 99.3% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155 |
| POMT2 | 94.9% | 94.9% | 100% | 99.8% | 98.4% | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156 |
| PORCN | 100% | 100% | 98.5% | 89.4% | 70.4% | Focal dermal hypoplasia, 305600 |

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| POU1F1 | 100% | 100% | 100% | 100% | 99.7% | Pituitary hormone deficiency, combined or isolated, 1, 613038 |
| POU3F2 | 100% | 100% | 100% | 99.9% | 98.7% | |
| POU3F3 | 100% | 99.8% | 99.9% | 97.9% | 88.4% | Snijders Blok-Fisher syndrome, 618604 |
| POU4F1 | 95.7% | 92.7% | 100% | 99.5% | 96.9% | Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352 |
| PPFIA2 | 100% | 100% | 100% | 100% | 99.8% | |
| PPFIA3 | 100% | 100% | 100% | 99.8% | 98.5% | Paul-Chao neurodevelopmental syndrome, 621122 |
| PPFIBP1 | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024 |
| PPIL1 | 80.1% | 80.1% | 100% | 100% | 99.2% | Pontocerebellar hypoplasia, type 14, 619301 |
| PPM1D | 100% | 100% | 100% | 100% | 99.9% | Breast cancer, somatic, 114480;Jansen-de Vries syndrome, 617450 |
| PPP1CB | 87.8% | 87.6% | 100% | 100% | 99.7% | Noonan syndrome-like disorder with loose anagen hair 2, 617506 |
| PPP1R12A | 100% | 100% | 100% | 100% | 99.8% | Genitourinary and/or/brain malformation syndrome, 618820 |
| PPP1R15B | 100% | 100% | 100% | 100% | 99.8% | Microcephaly, short stature, and impaired glucose metabolism 2, 616817 |
| PPP1R21 | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383 |

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| PPP1R3F | 97% | 97% | 98.6% | 88.8% | 70.5% | |
| PPP2CA | 100% | 100% | 100% | 100% | 99.8% | Houge-Janssens syndrome 3, 618354 |
| PPP2R1A | 94.1% | 94% | 100% | 99.9% | 99.5% | Houge-Janssens syndrome 2, 616362 |
| PPP2R2B | 100% | 100% | 100% | 100% | 99.7% | Spinocerebellar ataxia 12, 604326 |
| PPP2R3C | 100% | 100% | 100% | 100% | 99.9% | Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419 |
| PPP2R5B | 100% | 100% | 100% | 100% | 99.6% | |
| PPP2R5C | 97.8% | 96.9% | 100% | 100% | 99.7% | Houge-Janssens syndrome 4, 621185 |
| PPP2R5D | 100% | 100% | 100% | 100% | 99.3% | Houge-Janssens syndrome 1, 616355 |
| PPP3CA | 100% | 100% | 100% | 100% | 99.6% | Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265;Developmental and epileptic encephalopathy 91, 617711 |
| PPT1 | 90.3% | 90.3% | 100% | 100% | 99.7% | Ceroid lipofuscinosis, neuronal, 1, 256730 |
| PQBP1 | 100% | 100% | 98.6% | 86.7% | 67.3% | Renpenning syndrome, 309500 |
| PRDM13 | 100% | 100% | 100% | 99.9% | 98.7% | Pontocerebellar hypoplasia, type 17, 619909;Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 |
| PRDM15 | 100% | 100% | 100% | 99.9% | 99.2% | |
| PRICKLE2 | 100% | 100% | 100% | 100% | 99.5% | |
| PRKACB | 94.2% | 94.2% | 100% | 99.9% | 99.6% | Cardioacrofacial dysplasia 2, 619143 |

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| PRKAR1A | 100% | 100% | 100% | 100% | 99.6% | Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Adrenocortical tumor, somatic;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960 |
| PRKAR1B | 100% | 100% | 100% | 99.9% | 99.2% | Marbach-Schaaf neurodevelopmental syndrome, 619680 |
| PRMT7 | 100% | 100% | 100% | 100% | 99.6% | Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 |
| PRMT9 | 97.7% | 97.7% | 99.9% | 98.9% | 96.2% | |
| PRODH | 91.9% | 91.9% | 100% | 99.9% | 99.5% | {Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500 |
| PRPF19 | 100% | 100% | 100% | 100% | 99.4% | |
| PRPF8 | 100% | 100% | 100% | 100% | 99.3% | Retinitis pigmentosa 13, 600059 |
| 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 |
| PRR12 | 100% | 100% | 100% | 99.9% | 98.7% | Neuroocular syndrome, 619539 |
| PRSS12 | 100% | 100% | 100% | 100% | 99.3% | Intellectual developmental disorder, autosomal recessive 1, 249500 |

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| PRUNE1 | 93.1% | 93.1% | 100% | 100% | 99.6% | Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 |
| 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 |
| PSAT1 | 100% | 100% | 100% | 100% | 99.9% | Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992 |
| PSMC3 | 100% | 100% | 100% | 99.9% | 99.1% | Ebstein-Bezieau neurodevelopmental syndrome, 621539;?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354 |
| PSMC5 | 100% | 100% | 100% | 100% | 99.2% | Yu-Kury neurodevelopmental syndrome, 621565 |
| PSMD11 | 94.1% | 94.1% | 100% | 100% | 99.8% | |
| PSMD12 | 89.3% | 89.3% | 100% | 100% | 99.8% | Stankiewicz-Isidor syndrome, 617516 |
| PSPH | 100% | 100% | 100% | 100% | 99.8% | Phosphoserine phosphatase deficiency, 614023 |
| PTBP1 | 100% | 99.3% | 100% | 100% | 99.4% | STAD syndrome, 621495 |

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| PTCD3 | 100% | 100% | 100% | 100% | 99.7% | Combined oxidative phosphorylation deficiency 51, 619057 |
| PTCH1 | 100% | 100% | 100% | 100% | 99.2% | Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828 |
| PTCHD1 | 100% | 100% | 98.9% | 89.1% | 69.9% | {Autism, susceptibility to, X-linked 4}, 300830 |
| PTDSS1 | 93% | 93% | 100% | 100% | 100% | Lenz-Majewski hyperostotic dwarfism, 151050 |
| PTEN | 89.6% | 89.6% | 100% | 100% | 99.6% | {Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309 |
| PTF1A | 100% | 100% | 100% | 99.5% | 97.6% | Pancreatic and cerebellar agenesis, 609069;Pancreatic agenesis 2, 615935 |
| PTPA | 100% | 100% | 100% | 100% | 99.1% | Parkinson disease 25, autosomal recessive early-onset, with impaired intellectual development, 620482 |
| PTPMT1 | 100% | 100% | 100% | 100% | 99.2% | Neurodevelopmental disorder with ataxia and brain abnormalities, 621199 |

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| PTPN11 | 89.2% | 89.2% | 100% | 100% | 99.8% | Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785 |
| PTPN23 | 100% | 100% | 100% | 100% | 99% | Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 |
| PTPN4 | 100% | 100% | 100% | 100% | 99.9% | |
| PTRH2 | 100% | 100% | 100% | 100% | 99.9% | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 |
| PTRHD1 | 100% | 100% | 100% | 99.8% | 99.4% | Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747 |
| PTS | 100% | 100% | 100% | 100% | 99.6% | Hyperphenylalaninemia, BH4-deficient, A, 261640 |
| PUF60 | 100% | 100% | 100% | 100% | 99.2% | Verheij syndrome, 615583 |
| PUM1 | 92.4% | 92.4% | 100% | 99.9% | 99.2% | Neurodevelopmental disorder with motor abnormalities, and facial dysmorphism, 620719 |
| PURA | 100% | 100% | 100% | 100% | 99.3% | Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158 |
| PUS1 | 100% | 100% | 100% | 99.9% | 99.5% | Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 |

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| PUS3 | 100% | 100% | 100% | 100% | 100% | Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 |
| PUS7 | 100% | 100% | 100% | 100% | 99.5% | Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 |
| PYCR1 | 100% | 100% | 100% | 100% | 99.8% | Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940 |
| PYCR2 | 100% | 100% | 100% | 100% | 99.5% | Leukodystrophy, hypomyelinating, 10, 616420 |
| QARS1 | 100% | 100% | 100% | 100% | 99.2% | Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 |
| QDPR | 93.7% | 93.6% | 100% | 100% | 99.3% | Hyperphenylalaninemia, BH4-deficient, C, 261630 |
| QRICH1 | 100% | 100% | 100% | 100% | 99.4% | Ververi-Brady syndrome 1, 617982 |
| RAB11A | 92.9% | 92.9% | 100% | 100% | 99.6% | |
| RAB11B | 100% | 100% | 100% | 100% | 99.4% | Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 |
| RAB14 | 100% | 100% | 100% | 100% | 99.6% | |
| RAB18 | 100% | 100% | 100% | 100% | 99.5% | Warburg micro syndrome 3, 614222 |
| RAB23 | 100% | 100% | 100% | 100% | 99.4% | Carpenter syndrome 1, 201000 |
| RAB27A | 100% | 100% | 100% | 100% | 99.6% | Griscelli syndrome, type 2, 607624 |
| RAB34 | 100% | 100% | 100% | 100% | 99.5% | Orofaciodigital syndrome XX, 620718 |

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|---|---|---|---|---|---|---|
| RAB39B | 100% | 100% | 99.4% | 90.8% | 72.7% | Intellectual developmental disorder, X-linked 72, 300271;Waisman syndrome, 311510 |
| RAB3GAP1 | 95.7% | 95.7% | 100% | 100% | 99.6% | Martsolf syndrome 2, 619420;Warburg micro syndrome 1, 600118 |
| RAB3GAP2 | 94.4% | 94.4% | 100% | 100% | 99.8% | Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225 |
| RAB5C | 100% | 100% | 100% | 99.9% | 99.2% | |
| RABGAP1 | 90.8% | 90.2% | 100% | 100% | 99.8% | |
| RAC1 | 78.8% | 78.8% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal dominant 48, 617751 |
| RAC3 | 98.6% | 91.2% | 100% | 100% | 99.2% | Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 |
| RAD21 | 100% | 100% | 100% | 99.9% | 99.6% | Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376 |
| RAF1 | 96.1% | 93.8% | 100% | 100% | 99.7% | Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554 |
| RAI1 | 100% | 100% | 100% | 100% | 99% | Smith-Magenis syndrome, 182290 |
| RALA | 100% | 100% | 100% | 99.9% | 99.8% | Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311 |
| RALGAPA1 | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 |

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| RAP1B | 100% | 100% | 100% | 100% | 99.6% | Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies, 620654 |
| RAPGEF2 | 100% | 100% | 100% | 100% | 99.6% | ?Epilepsy, familial adult myoclonic, 7, 618075 |
| RARB | 98% | 98% | 100% | 100% | 99.7% | Microphthalmia, syndromic 12, 615524 |
| RARS1 | 90.9% | 90.5% | 100% | 99.9% | 99.4% | Leukodystrophy, hypomyelinating, 9, 616140 |
| RARS2 | 94.2% | 93.8% | 100% | 100% | 99.9% | Pontocerebellar hypoplasia, type 6, 611523 |
| RBBP5 | 100% | 100% | 100% | 100% | 99.5% | |
| RBBP8 | 100% | 100% | 100% | 100% | 99.8% | Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic |
| RBFOX1 | 100% | 99.7% | 100% | 99.9% | 99.3% | |
| RBL2 | 100% | 100% | 100% | 100% | 99.6% | Brunet-Wagner neurodevelopmental syndrome, 619690 |
| RBM10 | 95.6% | 95.6% | 98.7% | 88.7% | 70.4% | TARP syndrome, 311900 |
| RBM28 | 100% | 100% | 100% | 100% | 99.7% | ?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 |
| RBPJ | 100% | 100% | 100% | 100% | 99.6% | Adams-Oliver syndrome 3, 614814 |
| RBSN | 96.1% | 96.1% | 100% | 100% | 99.5% | Myelofibrosis, congenital, with anemia, neutropenia, developmental delay, and ocular abnormalities, 620939;Kariminejad neurodevelopmental syndrome, 620937 |

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| RCBTB1 | 100% | 100% | 100% | 100% | 99.7% | Retinal dystrophy with or without extraocular anomalies, 617175 |
| RECQL4 | 100% | 100% | 100% | 100% | 99.4% | Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280 |
| RELN | 100% | 100% | 100% | 100% | 99.8% | {Epilepsy, familial temporal lobe, 7}, 616436;Lissencephaly 2 (Norman-Roberts type), 257320 |
| RERE | 99% | 99% | 100% | 99.8% | 98.2% | Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 |
| REV3L | 100% | 100% | 100% | 100% | 99.8% | |
| RFC4 | 100% | 100% | 100% | 100% | 99.8% | Morimoto-Ryu-Malicdan neuromuscular syndrome, 621010 |
| RFT1 | 84.9% | 84.9% | 100% | 100% | 99.8% | Congenital disorder of glycosylation, type In, 612015 |
| RFX3 | 100% | 100% | 100% | 100% | 99.9% | |
| RFX4 | 100% | 100% | 100% | 99.9% | 99.1% | |
| RFX7 | 100% | 100% | 100% | 100% | 99.4% | Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities, 620330 |
| RHEB | 100% | 100% | 100% | 100% | 99.7% | |
| RHOBTB2 | 98.7% | 98.7% | 100% | 100% | 99.6% | Developmental and epileptic encephalopathy 64, 618004 |
| RIC1 | 100% | 100% | 100% | 100% | 99.8% | CATIFA syndrome, 618761 |
| RICTOR | 100% | 100% | 100% | 100% | 99.7% | |

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| RIMS2 | 99.2% | 99.2% | 100% | 100% | 99.8% | Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 |
| RIT1 | 100% | 100% | 100% | 100% | 99.8% | Noonan syndrome 8, 615355 |
| RLF | 100% | 100% | 100% | 100% | 99.9% | |
| RLIM | 100% | 100% | 98.9% | 91.9% | 75% | Tonne-Kalscheuer syndrome, 300978 |
| RMND1 | 85.8% | 85.8% | 100% | 100% | 99.7% | Combined oxidative phosphorylation deficiency 11, 614922 |
| RMRP | | | | | | Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250 |
| RNASEH2A | 100% | 100% | 100% | 100% | 98.8% | Aicardi-Goutieres syndrome 4, 610333 |
| RNASEH2B | 91.5% | 91.5% | 100% | 99.9% | 99.5% | Aicardi-Goutieres syndrome 2, 610181 |
| RNASEH2C | 100% | 100% | 100% | 99.9% | 98.6% | Aicardi-Goutieres syndrome 3, 610329 |
| RNASET2 | 79.4% | 74.3% | 100% | 100% | 99.4% | Leukoencephalopathy, cystic, without megalencephaly, 612951 |
| RNF113A | 100% | 100% | 99.3% | 87.4% | 70.4% | Trichothiodystrophy 5, nonphotosensitive, 300953 |
| RNF125 | 100% | 100% | 100% | 100% | 99.5% | Tenorio syndrome, 616260 |
| RNF13 | 85.4% | 85.3% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 73, 618379 |
| RNF2 | 100% | 100% | 100% | 100% | 99.5% | Luo-Schoch-Yamamoto syndrome, 619460 |

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| RNF220 | 100% | 100% | 100% | 100% | 99.7% | Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688 |
| RNPC3 | 100% | 100% | 100% | 100% | 99.8% | Pituitary hormone deficiency, combined or isolated, 7, 618160 |
| RNU12 | | | | | | CDAGS syndrome, 603116;?Spinocerebellar ataxia, autosomal recessive 33, 620208 |
| RNU2-2 | | | | | | Developmental and epileptic encephalopathy 119, 621304 |
| RNU4-2 | | | | | | Retinitis pigmentosa 102, 621560;ReNU syndrome, 620851 |
| RNU4ATAC | | | | | | Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710 |
| RNU5B-1 | | | | | | Neurodevelopmental disorder with seizures and joint laxity, 621302 |
| RNU6ATAC | | | | | | |
| RNU7-1 | | | | | | Aicardi-Goutieres syndrome 9, 619487 |
| ROBO1 | 100% | 100% | 100% | 100% | 99.6% | Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculorenal syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400 |
| ROGDI | 100% | 100% | 100% | 99.9% | 98.8% | Kohlschutter-Tonz syndrome, 226750 |

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| ROR2 | 100% | 100% | 100% | 100% | 99.6% | Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive 1, 268310 |
| RORA | 100% | 100% | 100% | 99.8% | 98.6% | Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 |
| RORB | 100% | 100% | 100% | 100% | 99.9% | {Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357 |
| RPGRIP1L | 100% | 100% | 100% | 100% | 99.7% | Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113 |
| RPH3A | 100% | 100% | 100% | 99.9% | 99.7% | |
| RPIA | 100% | 100% | 100% | 100% | 99.7% | Ribose 5-phosphate isomerase deficiency, 608611 |
| RPL10 | 100% | 100% | 99.3% | 91.1% | 69.5% | {Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998 |
| RPS19 | 100% | 100% | 100% | 100% | 99.2% | Diamond-Blackfan anemia 1, 105650 |
| RPS6KA3 | 97.6% | 97.6% | 99.2% | 92.4% | 75.4% | Intellectual developmental disorder, X-linked 19, 300844;Coffin-Lowry syndrome, 303600 |
| RPS6KC1 | 97.6% | 97.6% | 100% | 100% | 99.5% | Neurodevelopmental disorder with spasticity, thin corpus callosum, and decreased brain white matter, 621460 |
| RRAS2 | 95.5% | 95.5% | 100% | 100% | 99.5% | Ovarian carcinoma;Noonan syndrome 12, 618624 |
| RREB1 | 100% | 100% | 100% | 99.9% | 98.9% | |

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| RRM2B | 100% | 100% | 100% | 100% | 99.9% | Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 |
| RRP7A | 100% | 100% | 99.9% | 99.3% | 96.9% | ?Microcephaly 28, primary, autosomal recessive, 619453 |
| RSF1 | 100% | 100% | 100% | 100% | 99.8% | |
| RSPRY1 | 95.5% | 95.5% | 100% | 100% | 99.4% | Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 |
| RSRC1 | 84.2% | 84.2% | 100% | 99.9% | 99.5% | Intellectual developmental disorder, autosomal recessive 70, 618402 |
| RTEL1 | 100% | 100% | 100% | 99.9% | 99.4% | Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 |

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| RTN4IP1 | 100% | 100% | 100% | 100% | 99.8% | Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732 |
| RTTN | 100% | 100% | 100% | 100% | 99.8% | Microcephaly, short stature, and polymicrogyria with seizures, 614833 |
| RUBCN | 100% | 100% | 100% | 99.9% | 99.5% | Spinocerebellar ataxia, autosomal recessive 15, 615705 |
| RUNX1T1 | 100% | 100% | 100% | 100% | 99.7% | |
| RUSC2 | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal recessive 61, 617773 |
| RXYLT1 | 100% | 100% | 100% | 99.8% | 99.3% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 |
| RYBP | 100% | 100% | 100% | 99.9% | 99.4% | |
| SALL1 | 100% | 100% | 100% | 100% | 99.2% | Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480 |
| SAMD9 | 100% | 100% | 100% | 100% | 99.8% | Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053 |
| SAMHD1 | 92% | 89.8% | 100% | 100% | 99.5% | ?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952 |
| SARS1 | 89.1% | 89.1% | 100% | 100% | 99.3% | Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 |

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| SART3 | 100% | 99.6% | 100% | 100% | 99.4% | |
| SATB1 | 100% | 100% | 100% | 100% | 99.7% | den Hoed-de Boer-Voisin syndrome, 619229;Developmental delay with dysmorphic facies and dental anomalies, 619228 |
| SATB2 | 100% | 100% | 100% | 99.9% | 99.5% | Glass syndrome, 612313 |
| SBDS | 100% | 100% | 100% | 100% | 99.9% | {Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400 |
| SC5D | 100% | 100% | 100% | 100% | 99.9% | Lathosterolosis, 607330 |
| SCAF4 | 100% | 100% | 100% | 100% | 99.3% | Fliedner-Zweier syndrome, 620511 |
| SCAMP5 | 100% | 100% | 100% | 100% | 99.5% | |
| SCAPER | 97.9% | 97.9% | 100% | 100% | 99.8% | Intellectual developmental disorder and retinitis pigmentosa, 618195 |
| SCN1A | 100% | 100% | 100% | 100% | 99.7% | 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 |

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| SCN1B | 100% | 100% | 100% | 100% | 99.5% | Generalized epilepsy with febrile seizures plus, type 1, 604233;Developmental and epileptic encephalopathy 52, 617350;Cardiac conduction defect, nonspecific, 612838;Atrial fibrillation, familial, 13, 615377;Brugada syndrome 5, 612838 |
| SCN2A | 100% | 100% | 100% | 100% | 99.8% | Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924 |
| SCN3A | 100% | 100% | 100% | 100% | 99.8% | Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938 |
| SCN8A | 100% | 100% | 100% | 100% | 99.6% | ?Myoclonus, familial, 2, 618364;Seizures, benign familial infantile, 5, 617080;Cognitive impairment with or without cerebellar ataxia, 614306;Developmental and epileptic encephalopathy 13, 614558 |
| SCO1 | 100% | 100% | 100% | 100% | 99.6% | Mitochondrial complex IV deficiency, nuclear type 4, 619048 |
| SCO2 | 100% | 100% | 100% | 100% | 99.8% | Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377 |
| SCUBE3 | 97.5% | 97.5% | 100% | 100% | 99.8% | Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 2, 619184 |

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| SCYL1 | 100% | 100% | 100% | 99.9% | 99.1% | Spinocerebellar ataxia, autosomal recessive 21, 616719 |
| SCYL2 | 97.3% | 97.3% | 100% | 100% | 99.9% | Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766 |
| SDCCAG8 | 100% | 100% | 100% | 100% | 99.9% | Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993 |
| SDHA | 92.6% | 89.3% | 100% | 100% | 99.7% | Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma/paraganglioma syndrome 5, 614165 |
| SDHAF1 | 100% | 100% | 100% | 100% | 99.3% | Mitochondrial complex II deficiency, nuclear type 2, 619166 |
| SEC31A | 96.6% | 96.6% | 100% | 99.9% | 99.7% | ?Halperin-Birk syndrome, 618651 |
| SECISBP2 | 100% | 100% | 100% | 100% | 99.7% | Thyroid hormone metabolism, abnormal, 1, 609698 |
| SEL1L | 96.7% | 96.7% | 100% | 100% | 99.7% | Neurodevelopmental disorder with hypotonia, poor growth, dysmorphic facies, and agammaglobulinemia, 621068;?Neurodevelopmental disorder with poor growth, absent speech, progressive ataxia, and dysmorphic facies, 621067 |
| SEMA3E | 100% | 100% | 100% | 100% | 99.9% | |
| SEMA6A | 100% | 100% | 100% | 100% | 99.6% | |

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| SEMA6B | 100% | 100% | 100% | 99.8% | 98.9% | Epilepsy, progressive myoclonic, 11, 618876 |
| SEPHS1 | 91.3% | 86.6% | 100% | 100% | 99.8% | Ververi-Brady syndrome 2, 621325 |
| SEPSECS | 99.4% | 96.3% | 100% | 100% | 99.5% | Pontocerebellar hypoplasia type 2D, 613811 |
| SEPTIN2 | 93.9% | 93.9% | 100% | 100% | 99.7% | |
| SERAC1 | 100% | 100% | 100% | 100% | 99.9% | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 |
| SET | 100% | 100% | 100% | 99.9% | 99.2% | Intellectual developmental disorder, autosomal dominant 58, 618106 |
| SETBP1 | 100% | 100% | 100% | 100% | 99.5% | Schinzel-Giedion midface retraction syndrome, 269150;Intellectual developmental disorder, autosomal dominant 29, 616078 |
| SETD1A | 100% | 100% | 100% | 100% | 99.1% | Epilepsy, early-onset, 2, with or without developmental delay, 618832;Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056 |
| SETD1B | 100% | 100% | 100% | 99.8% | 98.6% | Intellectual developmental disorder with seizures and language delay, 619000 |
| SETD2 | 100% | 100% | 100% | 100% | 99.7% | Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155 |

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| SETD5 | 99.7% | 99.1% | 100% | 99.9% | 99.5% | Intellectual developmental disorder, autosomal dominant 23, 615761 |
| SF1 | 100% | 100% | 100% | 99.7% | 98.7% | |
| SF3B1 | 100% | 100% | 100% | 100% | 99.8% | Myelodysplastic syndrome, somatic, 614286 |
| SF3B3 | 98% | 96.9% | 100% | 100% | 99.6% | |
| SFXN4 | 100% | 100% | 100% | 99.9% | 99.6% | Combined oxidative phosphorylation deficiency 18, 615578 |
| SGPL1 | 95.5% | 95.5% | 100% | 100% | 99.7% | RENI syndrome, 617575 |
| SGSH | 100% | 100% | 100% | 100% | 99.5% | Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 |
| SGSM3 | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal recessive 84, 620401 |
| SHANK1 | 99.4% | 98.7% | 100% | 99.8% | 97.6% | |
| SHANK2 | 100% | 100% | 100% | 100% | 99% | {Autism susceptibility 17}, 613436 |
| SHANK3 | 99.8% | 99% | 99.9% | 99.7% | 98.4% | Phelan-McDermid syndrome, 606232;{Schizophrenia 15}, 613950 |
| SHH | 100% | 100% | 100% | 99.8% | 98.4% | Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638 |
| SHMT2 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 |
| SHOC2 | 100% | 100% | 100% | 100% | 99.9% | Noonan syndrome-like with loose anagen hair 1, 607721 |

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| SHQ1 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with dystonia and seizures, 619922;?Dystonia 35, childhood-onset, 619921 |
| SHROOM4 | 100% | 100% | 99% | 90.1% | 72.3% | |
| SIAH1 | 100% | 100% | 100% | 100% | 99.8% | Buratti-Harel syndrome, 619314 |
| SIK1 | 100% | 100% | 100% | 100% | 99.3% | Developmental and epileptic encephalopathy 30, 616341 |
| SIL1 | 91.5% | 91.5% | 100% | 100% | 99.5% | Marinesco-Sjogren syndrome, 248800 |
| SIN3A | 97.5% | 97.5% | 100% | 100% | 99.6% | Witteveen-Kolk syndrome, 613406 |
| SIN3B | 99.2% | 97.7% | 100% | 99.9% | 99.3% | |
| SIX3 | 100% | 100% | 100% | 99.4% | 97.2% | Schizencephaly, 269160;Holoprosencephaly 2, 157170 |
| SKI | 100% | 100% | 100% | 99.8% | 98.8% | Shprintzen-Goldberg syndrome, 182212 |
| SKIC3 | 98.9% | 98.9% | 100% | 100% | 99.9% | Trichohepatoenteric syndrome 1, 222470 |
| SKOR2 | 100% | 100% | 100% | 99.8% | 97.1% | Valence-Farazi cerebellar ataxia syndrome, 621386 |
| SLC12A2 | 100% | 100% | 100% | 100% | 99.6% | Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081 |
| SLC12A5 | 100% | 100% | 100% | 99.9% | 99.4% | {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685;Developmental and epileptic encephalopathy 34, 616645 |

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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 |
| SLC12A9 | 100% | 100% | 100% | 100% | 98.5% | |
| SLC13A5 | 100% | 100% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 |
| SLC16A2 | 88.4% | 88.4% | 99% | 91.3% | 72% | Allan-Herndon-Dudley syndrome, 300523 |
| SLC17A5 | 92.7% | 92.7% | 100% | 100% | 99.8% | Salla disease, 604369;Sialic acid storage disorder, infantile, 269920 |
| SLC18A2 | 99.6% | 96.7% | 100% | 100% | 99.8% | Parkinsonism-dystonia, infantile, 2, 618049 |
| SLC19A3 | 99.9% | 98.9% | 100% | 100% | 99.7% | Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483 |
| SLC1A1 | 100% | 100% | 100% | 99.9% | 99.4% | Dicarboxylic aminoaciduria, 222730;{?Schizophrenia susceptibility 18}, 615232 |
| SLC1A2 | 100% | 100% | 100% | 100% | 99.4% | Developmental and epileptic encephalopathy 41, 617105 |
| SLC1A4 | 100% | 100% | 100% | 99.9% | 99.3% | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 |
| SLC25A1 | 100% | 100% | 100% | 99.9% | 98.8% | Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197 |

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| SLC25A12 | 90.5% | 89.5% | 100% | 100% | 99.9% | Developmental and epileptic encephalopathy 39, 612949 |
| SLC25A15 | 100% | 100% | 100% | 100% | 99.8% | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 |
| SLC25A22 | 100% | 100% | 100% | 99.9% | 99.2% | Developmental and epileptic encephalopathy 3, 609304 |
| SLC25A24 | 99.4% | 99.4% | 99.7% | 99.1% | 97.9% | Fontaine progeroid syndrome, 612289 |
| SLC25A42 | 100% | 100% | 100% | 100% | 99.2% | Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 |
| SLC27A3 | 100% | 100% | 100% | 99.9% | 99% | |
| SLC2A1 | 100% | 100% | 100% | 100% | 99.3% | 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 |
| SLC30A9 | 100% | 100% | 100% | 100% | 99.9% | Birk-Landau-Perez syndrome, 617595 |
| SLC31A1 | 100% | 100% | 100% | 99.9% | 99.5% | Neurodegeneration and seizures due to copper transport defect, 620306 |

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| SLC32A1 | 100% | 100% | 100% | 100% | 99.8% | Generalized epilepsy with febrile seizures plus, type 12, 620755;Developmental and epileptic encephalopathy 114, 620774 |
| SLC33A1 | 92.6% | 92.6% | 100% | 100% | 99.7% | Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482 |
| SLC35A1 | 94.9% | 94.9% | 100% | 100% | 100% | Congenital disorder of glycosylation, type IIf, 603585 |
| SLC35A2 | 100% | 100% | 98.9% | 87.9% | 68.3% | Congenital disorder of glycosylation, type IIm, 300896 |
| SLC35A3 | 91% | 87.6% | 100% | 100% | 99.8% | Arthrogryposis, impaired intellectual development, and seizures, 615553 |
| SLC35B2 | 100% | 100% | 100% | 100% | 99.2% | Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269 |
| SLC35C1 | 100% | 100% | 100% | 100% | 98.9% | Congenital disorder of glycosylation, type IIc, 266265 |
| SLC38A3 | 96.3% | 96.3% | 100% | 100% | 99.4% | Developmental and epileptic encephalopathy 102, 619881 |
| SLC39A14 | 94% | 94% | 100% | 100% | 99.5% | ?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013 |
| SLC39A8 | 77.5% | 77.4% | 100% | 100% | 100% | Congenital disorder of glycosylation, type IIn, 616721 |
| SLC45A1 | 100% | 100% | 100% | 100% | 99.4% | Intellectual developmental disorder with neuropsychiatric features, 617532 |

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| SLC46A1 | 100% | 100% | 99.9% | 99.7% | 99% | Folate malabsorption, hereditary, 229050 |
| SLC4A10 | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, 620746 |
| SLC4A4 | 95% | 95% | 100% | 100% | 99.9% | Proximal renal tubular acidosis-ocular anomaly syndrome, 604278 |
| 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 |
| SLC6A1 | 100% | 100% | 100% | 99.9% | 99.3% | Myoclonic-atonic epilepsy, 616421 |
| SLC6A17 | 100% | 100% | 100% | 100% | 99.2% | Intellectual developmental disorder, autosomal recessive 48, 616269 |
| SLC6A19 | 100% | 100% | 100% | 99.9% | 99% | Hartnup disorder, 234500 |
| SLC6A3 | 93.9% | 92.4% | 100% | 100% | 99.6% | Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890 |
| SLC6A8 | 100% | 99.9% | 98.4% | 88.1% | 71.2% | Cerebral creatine deficiency syndrome 1, 300352 |

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| SLC6A9 | 100% | 100% | 100% | 100% | 99.2% | Glycine encephalopathy with normal serum glycine, 617301;{Scoliosis, isolated, susceptibility to, 6}, 621428 |
| SLC7A7 | 100% | 100% | 100% | 100% | 98.8% | Lysinuric protein intolerance, 222700 |
| SLC9A6 | 100% | 100% | 99.1% | 90.8% | 73.1% | Neurodegenerative disorder, X-linked, female-restricted, with parkinsonism and cognitive impairment, 301142;Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 |
| SLC9A7 | 100% | 100% | 99% | 89% | 70.1% | Intellectual developmental disorder, X-linked 108, 301024 |
| SLF2 | 100% | 100% | 100% | 100% | 99.8% | Atelis syndrome 1, 620184 |
| SLITRK2 | 100% | 100% | 99.2% | 90.7% | 71.1% | Intellectual developmental disorder, X-linked 111, 301107 |
| SMAD4 | 95.4% | 95.4% | 100% | 100% | 99.8% | Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 |
| SMARCA1 | 98% | 98% | 99.2% | 91.5% | 73.1% | |
| SMARCA2 | 98% | 97.9% | 100% | 100% | 99.6% | Nicolaides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293 |

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| SMARCA4 | 100% | 100% | 100% | 99.9% | 99.3% | Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792 |
| SMARCA5 | 98.4% | 98.4% | 100% | 100% | 99.8% | |
| SMARCB1 | 95.2% | 95.2% | 100% | 100% | 99.7% | Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322 |
| SMARCC2 | 94.2% | 94.2% | 100% | 99.9% | 99.3% | Coffin-Siris syndrome 8, 618362 |
| SMARCD1 | 100% | 100% | 100% | 100% | 99.5% | Coffin-Siris syndrome 11, 618779 |
| SMARCE1 | 100% | 100% | 100% | 100% | 99.8% | {Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938 |
| SMC1A | 100% | 100% | 99.1% | 88.6% | 70.4% | Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 |
| SMC3 | 100% | 100% | 100% | 100% | 99.9% | Cornelia de Lange syndrome 3, 610759 |
| SMC5 | 97.3% | 97.3% | 100% | 100% | 99.8% | Atelis syndrome 2, 620185 |
| SMG8 | 100% | 100% | 100% | 100% | 99.7% | Alzahrani-Kuwahara syndrome, 619268 |

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| SMG9 | 100% | 100% | 100% | 100% | 99.6% | Heart and brain malformation syndrome, 616920;Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995 |
| SMOC1 | 96% | 96% | 100% | 100% | 99.7% | Microphthalmia with limb anomalies, 206920 |
| SMPD1 | 100% | 100% | 100% | 100% | 99.6% | Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200 |
| SMPD4 | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 |
| SMS | 94.4% | 94.2% | 99.1% | 91.9% | 73.3% | Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 |
| SNAP25 | 89.5% | 89.5% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 117, 616330 |
| SNAP29 | 100% | 100% | 100% | 99.7% | 99.3% | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 |
| SNAPC4 | 100% | 100% | 100% | 100% | 99.3% | Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515 |

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| SNAPIN | 100% | 100% | 100% | 100% | 99.3% | Neurodevelopmental disorder with structural brain abnormalities and craniofacial abnormalities, 621393 |
| SNF8 | 94.6% | 94.6% | 100% | 100% | 99.6% | Developmental and epileptic encephalopathy 115, 620783;Neurodevelopmental disorder plus optic atrophy, 620784 |
| SNIP1 | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501 |
| SNORD118 | | | | | | Leukoencephalopathy, brain calcifications, and cysts, 614561 |
| SNRPB | 90.5% | 82.4% | 100% | 100% | 99.7% | Cerebrocostomandibular syndrome, 117650 |
| SNRPN | 100% | 100% | 100% | 100% | 99.9% | |
| SNX14 | 95% | 95% | 100% | 100% | 99.8% | Spinocerebellar ataxia, autosomal recessive 20, 616354 |
| SNX27 | 100% | 100% | 100% | 100% | 99.5% | |
| SOBP | 100% | 100% | 100% | 99.8% | 97% | ?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671 |
| SON | 100% | 100% | 100% | 100% | 99.7% | ZTTK syndrome, 617140 |
| SOS1 | 98.8% | 98.6% | 100% | 100% | 99.6% | Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300 |
| SOS2 | 95% | 95% | 100% | 100% | 99.5% | Noonan syndrome 9, 616559 |

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| 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 |
| SOX11 | 100% | 100% | 100% | 99.7% | 96% | Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866 |
| SOX2 | 100% | 100% | 100% | 99.9% | 97.8% | Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900 |
| SOX3 | 100% | 100% | 96.1% | 81.1% | 61.5% | Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000 |
| SOX4 | 100% | 100% | 100% | 99.7% | 97.5% | Intellectual developmental disorder with speech delay and dysmorphic facies, 618506 |
| SOX5 | 100% | 100% | 100% | 100% | 99.8% | Lamb-Shaffer syndrome, 616803 |
| SOX6 | 100% | 100% | 100% | 100% | 99.6% | Tolchin-Le Caignec syndrome, 618971 |
| SP9 | 100% | 100% | 100% | 99.9% | 98.4% | |
| SPAG9 | 100% | 100% | 100% | 100% | 99.5% | |
| SPART | 95.8% | 95.8% | 100% | 100% | 99.7% | Troyer syndrome, 275900 |
| SPAST | 94.7% | 94.6% | 100% | 100% | 99.5% | Spastic paraplegia 4, autosomal dominant, 182601 |

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| SPECC1L | 94.6% | 94.6% | 100% | 100% | 99.7% | Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251 |
| SPEN | 100% | 100% | 100% | 100% | 99.6% | Radio-Tartaglia syndrome, 619312 |
| 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 |
| SPOCK1 | 100% | 100% | 100% | 100% | 99.2% | |
| SPOP | 96.4% | 96.4% | 100% | 100% | 99.8% | Nabais Sa-de Vries syndrome, type 1, 618828;Nabais Sa-de Vries syndrome, type 2, 618829 |
| SPOUT1 | 100% | 100% | 100% | 100% | 99.4% | Neurodevelopmental disorder with poor growth, seizures, and brain abnormalities, 621154 |
| SPR | 100% | 100% | 100% | 100% | 99.8% | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 |
| SPRED1 | 97.6% | 97.6% | 100% | 100% | 100% | Legius syndrome, 611431 |
| SPRED2 | 100% | 100% | 100% | 100% | 99.8% | Noonan syndrome 14, 619745 |

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| 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 |
| SPTBN1 | 100% | 100% | 100% | 100% | 99.7% | Developmental delay, impaired speech, and behavioral abnormalities, 619475 |
| SPTBN2 | 100% | 100% | 100% | 99.9% | 99.3% | Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386 |
| SPTBN4 | 100% | 100% | 100% | 99.9% | 98.7% | Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 |
| SRCAP | 100% | 100% | 100% | 100% | 99.3% | Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140 |
| SRD5A3 | 100% | 100% | 100% | 100% | 99.9% | Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379 |
| SRP54 | 96.3% | 96.3% | 100% | 100% | 99.9% | Neutropenia, severe congenital, 8, autosomal dominant, 618752 |
| SRPK3 | 100% | 99.9% | 97.7% | 87.1% | 69.3% | Intellectual developmental disorder, X-linked 114, 301134 |

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| SRPX2 | 100% | 100% | 99.1% | 89.6% | 71.9% | ?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643 |
| SRRM1 | 100% | 100% | 100% | 100% | 99.3% | |
| SRRM2 | 100% | 100% | 100% | 99.9% | 99.1% | Intellectual developmental disorder, autosomal dominant 72, 620439 |
| SRRM4 | 100% | 100% | 100% | 100% | 99.1% | |
| SRSF1 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities, 620489 |
| SSPOP | | | | | | |
| SSR4 | 100% | 100% | 98.9% | 87.6% | 65.5% | Congenital disorder of glycosylation, type Iy, 300934 |
| ST3GAL3 | 89.5% | 88.2% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090 |
| ST3GAL5 | 98.4% | 98.4% | 100% | 100% | 99.6% | Salt and pepper developmental regression syndrome, 609056 |
| STAG1 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 47, 617635 |
| STAG2 | 100% | 100% | 99.3% | 91.5% | 74% | Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022 |
| STAMBP | 96.3% | 96.3% | 100% | 100% | 99.8% | Microcephaly-capillary malformation syndrome, 614261 |

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| STEEP1 | 100% | 100% | 99.1% | 88% | 67.3% | ?Intellectual developmental disorder, X-linked 107, 301013 |
| STIL | 100% | 100% | 100% | 100% | 99.8% | Microcephaly 7, primary, autosomal recessive, 612703 |
| STRA6 | 100% | 100% | 100% | 100% | 99.3% | Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186 |
| STRADA | 100% | 100% | 100% | 99.9% | 99.4% | Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 |
| STT3A | 99.9% | 98.8% | 100% | 100% | 99.8% | Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714;Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596 |
| STT3B | 100% | 100% | 100% | 100% | 99.5% | Congenital disorder of glycosylation, type Ix, 615597 |
| STX1A | 100% | 98.7% | 100% | 100% | 99.2% | |
| STX1B | 100% | 100% | 100% | 99.8% | 98.2% | Generalized epilepsy with febrile seizures plus, type 9, 616172 |
| STXBP1 | 100% | 100% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 4, 612164 |
| SUCLA2 | 96.3% | 96.3% | 100% | 100% | 99.9% | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 |

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| SUCLG1 | 100% | 100% | 100% | 100% | 99.8% | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 |
| SUFU | 100% | 100% | 100% | 100% | 99.6% | {Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255 |
| SUMF1 | 100% | 100% | 100% | 100% | 99.9% | Multiple sulfatase deficiency, 272200 |
| SUOX | 100% | 100% | 100% | 100% | 99.5% | Sulfite oxidase deficiency, 272300 |
| SUPT16H | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480 |
| SUPT4H1 | 100% | 100% | 100% | 100% | 99.5% | |
| SURF1 | 100% | 100% | 100% | 100% | 99.4% | Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110 |
| SUZ12 | 95.6% | 95.6% | 100% | 100% | 99.7% | Imagawa-Matsumoto syndrome, 618786 |
| SVBP | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 |
| SYN1 | 100% | 100% | 98.4% | 86.4% | 66.5% | Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491;Intellectual developmental disorder, X-linked 50, 300115 |
| SYNCRIP | 92.9% | 92.9% | 100% | 100% | 99.9% | |

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| SYNGAP1 | 100% | 100% | 100% | 99.9% | 99% | Intellectual developmental disorder, autosomal dominant 5, 612621 |
| SYNJ1 | 100% | 100% | 100% | 100% | 99.8% | Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389 |
| SYP | 100% | 100% | 98.6% | 89.9% | 71% | Intellectual developmental disorder, X-linked 96, 300802 |
| SYT1 | 96% | 96% | 100% | 100% | 99.9% | Baker-Gordon syndrome, 618218 |
| SZT2 | 100% | 100% | 100% | 100% | 99.6% | Developmental and epileptic encephalopathy 18, 615476 |
| TACO1 | 100% | 100% | 100% | 99.8% | 99.3% | Mitochondrial complex IV deficiency, nuclear type 8, 619052 |
| TAF1 | 98.7% | 98.7% | 98.9% | 89% | 70.8% | Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250 |
| TAF13 | 85.1% | 77.2% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 60, 617432 |
| TAF1C | 100% | 99.2% | 100% | 100% | 99.4% | |
| TAF2 | 96.3% | 96.3% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 40, 615599 |
| TAF4 | 92.3% | 84.8% | 99.9% | 99% | 94.1% | Intellectual developmental disorder, autosomal dominant 73, 620450 |

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| TAF6 | 100% | 100% | 100% | 100% | 99.3% | Alazami-Yuan syndrome, 617126 |
| TAF8 | 85.3% | 85.3% | 100% | 100% | 99.3% | Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972 |
| TANC2 | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906 |
| TANGO2 | 100% | 100% | 100% | 100% | 99.6% | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 |
| TAOK1 | 100% | 100% | 100% | 100% | 99.8% | Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575 |
| TAOK2 | 100% | 100% | 100% | 100% | 99.2% | |
| TARS2 | 100% | 100% | 100% | 100% | 99.8% | Combined oxidative phosphorylation deficiency 21, 615918 |
| TASP1 | 93.2% | 93.2% | 100% | 100% | 99.8% | Suleiman-El-Hattab syndrome, 618950 |
| TAT | 100% | 100% | 100% | 100% | 99.8% | Tyrosinemia, type II, 276600 |
| TBC1D20 | 92.2% | 92.2% | 100% | 100% | 99.7% | Warburg micro syndrome 4, 615663 |
| TBC1D23 | 96.3% | 96.3% | 100% | 100% | 99.8% | Pontocerebellar hypoplasia, type 11, 617695 |

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| TBC1D24 | 100% | 100% | 100% | 100% | 99.4% | Deafness, autosomal recessive 86, 614617;Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021;Deafness, autosomal dominant 65, 616044;Developmental and epileptic encephalopathy 16, 615338;DOORS syndrome, 220500 |
| TBC1D2B | 100% | 100% | 100% | 100% | 99.5% | Neurodevelopmental disorder with seizures and gingival overgrowth, 619323 |
| TBC1D7 | 88.9% | 88.9% | 100% | 100% | 99.4% | Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 |
| TBCB | 77.5% | 77.5% | 100% | 100% | 99.4% | Neurodevelopmental disorder with behavioral abnormalities and childhood onset spastic paraplegia, 621382 |
| TBCD | 91.2% | 90.7% | 100% | 99.9% | 99.2% | Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 |
| 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 |

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| TBCK | 100% | 100% | 100% | 100% | 99.9% | Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 |
| TBL1XR1 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 41, 616944;Pierpont syndrome, 602342 |
| TBP | 100% | 100% | 100% | 100% | 99.3% | Spinocerebellar ataxia 17, 607136;{Parkinson disease, susceptibility to}, 168600 |
| TBR1 | 100% | 100% | 100% | 99.9% | 98.9% | Intellectual developmental disorder with autism and speech delay, 606053 |
| TBX1 | 98.6% | 95.7% | 100% | 99.4% | 96.1% | Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430 |
| TCEAL1 | 100% | 100% | 99.7% | 86.3% | 68.4% | Hijazi-Reis syndrome, 301094 |
| TCF20 | 100% | 100% | 100% | 99.9% | 99.1% | Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 |
| TCF4 | 97.1% | 97.1% | 100% | 100% | 99.6% | Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267 |
| TCF7L2 | 100% | 100% | 100% | 99.9% | 98.6% | {Diabetes mellitus, type 2, susceptibility to}, 125853 |
| TCN2 | 91.8% | 87.7% | 100% | 100% | 99.9% | Transcobalamin II deficiency, 275350 |

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|---|---|---|---|---|---|---|
| TCP1 | 100% | 100% | 100% | 99.9% | 99.7% | Intellectual developmental disorder with polymicrogyria and seizures, 621021 |
| TCTN2 | 98.5% | 98.5% | 100% | 100% | 99.5% | Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885 |
| TCTN3 | 100% | 100% | 100% | 100% | 99.5% | Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860 |
| TDP2 | 100% | 100% | 100% | 100% | 99.8% | Spinocerebellar ataxia, autosomal recessive 23, 616949 |
| TECPR2 | 100% | 100% | 100% | 99.9% | 99.6% | Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 |
| TECR | 87.3% | 87.3% | 100% | 100% | 98.9% | Intellectual developmental disorder, autosomal recessive 14, 614020 |
| TEFM | 100% | 100% | 100% | 100% | 99.8% | Combined oxidative phosphorylation deficiency 58, 620451 |
| TELO2 | 94.4% | 94.3% | 100% | 99.9% | 99.4% | You-Hoover-Fong syndrome, 616954 |
| TENM3 | 99.6% | 99.6% | 100% | 100% | 99.6% | Microphthalmia, syndromic 15, 615145;?Microphthalmia/coloboma 9, 615145 |
| TET3 | 100% | 100% | 100% | 100% | 98.9% | Beck-Fahrner syndrome, 618798 |
| TFAP2A | 100% | 100% | 100% | 100% | 98.8% | Branchiooculofacial syndrome, 113620 |
| TFE3 | 100% | 99.9% | 98.4% | 87.4% | 67.8% | Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066;Renal cell carcinoma, papillary, 1, 300854 |

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|---|---|---|---|---|---|---|
| TGDS | 100% | 100% | 100% | 100% | 99.9% | Catel-Manzke syndrome, 616145 |
| TGFBR1 | 100% | 100% | 100% | 100% | 99.6% | {Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192 |
| TGIF1 | 100% | 100% | 100% | 100% | 99.9% | Holoprosencephaly 4, 142946 |
| TH | 100% | 99.8% | 100% | 100% | 99.2% | Segawa syndrome, recessive, 605407 |
| THG1L | 100% | 100% | 100% | 99.7% | 99.3% | Spinocerebellar ataxia, autosomal recessive 28, 618800 |
| THOC2 | 100% | 100% | 99.2% | 91.7% | 73.7% | Arthrogryposis multiplex congenita 7, X-linked, 301127;Intellectual developmental disorder, X-linked syndromic, Kumar type, 300957 |
| THOC6 | 100% | 100% | 100% | 100% | 99.5% | Beaulieu-Boycott-Innes syndrome, 613680 |
| THRB | 100% | 100% | 100% | 100% | 99.9% | Thyroid hormone resistance, autosomal recessive, 274300;Thyroid hormone resistance, 188570;Thyroid hormone resistance, selective pituitary, 145650 |
| THUMPD1 | 100% | 100% | 100% | 99.8% | 99.4% | Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989 |
| TIAM1 | 98.3% | 98.3% | 100% | 100% | 99.5% | Neurodevelopmental disorder with language delay and seizures, 619908 |
| TIMM50 | 100% | 100% | 100% | 100% | 99.6% | 3-methylglutaconic aciduria, type IX, 617698 |

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|---|---|---|---|---|---|---|
| TIMM8A | 100% | 100% | 98.2% | 87.9% | 71.6% | Mohr-Tranebjaerg syndrome, 304700 |
| TINF2 | 100% | 100% | 100% | 100% | 99.7% | Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130 |
| TKFC | 100% | 100% | 100% | 99.9% | 99.4% | Triokinase and FMN cyclase deficiency syndrome, 618805 |
| TKT | 89% | 89% | 100% | 100% | 99.5% | Short stature, developmental delay, and congenital heart defects, 617044 |
| TLK2 | 100% | 100% | 100% | 100% | 99.9% | Intellectual developmental disorder, autosomal dominant 57, 618050 |
| TM2D3 | 100% | 100% | 100% | 100% | 99.6% | Neurocardiorenal malformation syndrome, 621379 |
| TMCO1 | 72.3% | 72.3% | 100% | 100% | 99.6% | Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980 |
| TMEM106B | 100% | 100% | 100% | 100% | 99.9% | Leukodystrophy, hypomyelinating, 16, 617964 |
| TMEM107 | 100% | 100% | 100% | 100% | 99.2% | Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562 |
| TMEM147 | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075 |
| TMEM163 | 93.9% | 93.9% | 100% | 99.9% | 99.1% | Leukodystrophy, hypomyelinating, 25, 620243 |

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| TMEM165 | 88.9% | 88.9% | 100% | 100% | 99.7% | Congenital disorder of glycosylation, type IIk, 614727 |
| TMEM167A | 100% | 100% | 100% | 100% | 99.7% | |
| TMEM184B | 100% | 100% | 100% | 100% | 99.2% | |
| TMEM216 | 100% | 100% | 100% | 100% | 99.8% | Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194 |
| TMEM218 | 100% | 100% | 100% | 100% | 99.6% | Joubert syndrome 39, 619562 |
| TMEM222 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470 |
| TMEM231 | 93.5% | 93.5% | 100% | 100% | 99.5% | Joubert syndrome 20, 614970;Meckel syndrome 11, 615397 |
| TMEM237 | 98.2% | 98.2% | 100% | 100% | 99.7% | Joubert syndrome 14, 614424 |
| TMEM240 | 100% | 100% | 100% | 99.7% | 97.6% | Spinocerebellar ataxia 21, 607454 |
| TMEM63A | 99.7% | 97.5% | 100% | 100% | 99.6% | Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 |
| TMEM63B | 100% | 100% | 100% | 100% | 99.5% | Developmental and epileptic encephalopathy 118, 621250 |
| TMEM63C | 100% | 100% | 100% | 100% | 99.1% | Spastic paraplegia 87, autosomal recessive, 619966 |

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|---|---|---|---|---|---|---|
| TMEM67 | 96.1% | 96.1% | 100% | 100% | 99.7% | 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 |
| TMEM70 | 100% | 100% | 100% | 100% | 99.3% | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 |
| TMEM94 | 100% | 100% | 100% | 100% | 99.6% | Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 |
| TMLHE | 94.8% | 94.8% | 99.6% | 94.8% | 79.1% | {Autism, susceptibility to, X-linked 6}, 300872 |
| TMTC3 | 100% | 100% | 100% | 100% | 99.7% | Lissencephaly 8, 617255 |
| TMX2 | 99.8% | 97% | 100% | 100% | 99.7% | Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 |
| TNIK | 98.6% | 98.6% | 100% | 100% | 99.6% | Intellectual developmental disorder, autosomal recessive 54, 617028 |
| TNPO2 | 100% | 100% | 100% | 99.9% | 99.2% | Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556 |
| TNR | 100% | 100% | 100% | 99.9% | 99.5% | Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653 |

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| TNRC6B | 100% | 100% | 100% | 100% | 99.6% | Global developmental delay with speech and behavioral abnormalities, 619243 |
| TOE1 | 100% | 100% | 100% | 99.9% | 99.4% | Pontocerebellar hypoplasia, type 7, 614969 |
| TOGARAM1 | 100% | 100% | 100% | 100% | 99.7% | Joubert syndrome 37, 619185 |
| TOMM70 | 100% | 100% | 100% | 100% | 99.7% | |
| TOR1A | 93.1% | 90.7% | 100% | 99.9% | 99.4% | {Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100 |
| TP53RK | 100% | 100% | 100% | 100% | 99.8% | Galloway-Mowat syndrome 4, 617730 |
| TP73 | 100% | 100% | 100% | 100% | 99.5% | Ciliary dyskinesia, primary, 47, and lissencephaly, 619466 |
| TPI1 | 100% | 100% | 100% | 100% | 99.6% | Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 |
| TPO | 100% | 100% | 100% | 100% | 99.5% | Thyroid dyshormonogenesis 2A, 274500 |
| TPP1 | 100% | 100% | 100% | 100% | 99.3% | Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270 |
| TPP2 | 100% | 100% | 100% | 100% | 99.9% | Immunodeficiency 78 with autoimmunity and developmental delay, 619220 |
| TPRKB | 83.2% | 81.2% | 100% | 100% | 99.8% | Galloway-Mowat syndrome 5, 617731 |
| TRA2B | 100% | 100% | 100% | 100% | 99.7% | Ramond-Elliott neurodevelopmental syndrome, 621421 |

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| TRAF7 | 100% | 100% | 100% | 99.9% | 99.2% | Cardiac, facial, and digital anomalies with developmental delay, 618164 |
| TRAIP | 100% | 100% | 100% | 100% | 99.8% | Seckel syndrome 9, 616777 |
| TRAK1 | 100% | 100% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 68, 618201 |
| TRAPPC11 | 100% | 100% | 100% | 100% | 99.8% | Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 |
| TRAPPC12 | 86.3% | 86.3% | 100% | 99.9% | 99% | Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 |
| TRAPPC2L | 100% | 100% | 100% | 100% | 99.6% | Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 |
| TRAPPC4 | 100% | 100% | 100% | 99.9% | 99.3% | Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 |
| TRAPPC6B | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 |
| TRAPPC9 | 100% | 100% | 100% | 100% | 99.5% | Intellectual developmental disorder, autosomal recessive 13, 613192 |

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| TREX1 | 100% | 100% | 100% | 100% | 99.5% | 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 |
| TRH | 100% | 100% | 100% | 100% | 99.5% | Thyrotropin-releasing hormone deficiency, 275120 |
| TRIM32 | 100% | 100% | 100% | 100% | 99.6% | ?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 |
| TRIM71 | 100% | 100% | 100% | 100% | 98.9% | Hydrocephalus, congenital, 4, 618667 |
| TRIM8 | 100% | 100% | 100% | 99.9% | 99.2% | Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428 |
| TRIO | 99.3% | 99.1% | 100% | 100% | 99.5% | Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061;Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825 |
| TRIP12 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 49, 617752 |
| TRIT1 | 100% | 100% | 100% | 100% | 99.8% | Combined oxidative phosphorylation deficiency 35, 617873 |

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| TRMT1 | 100% | 100% | 100% | 100% | 99% | Intellectual developmental disorder, autosomal recessive 68, 618302 |
| TRMT10A | 100% | 100% | 100% | 100% | 99.7% | Microcephaly, short stature, and impaired glucose metabolism 1, 616033 |
| TRMT5 | 100% | 100% | 100% | 100% | 99.8% | Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539 |
| TRNT1 | 91.9% | 91.9% | 100% | 100% | 99.9% | Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959 |
| TRPM3 | 97.8% | 97.8% | 100% | 100% | 99.6% | ?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224 |
| TRPM7 | 100% | 100% | 100% | 100% | 99.8% | {Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500 |
| TRRAP | 100% | 100% | 100% | 100% | 99.5% | ?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454 |

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| TSC1 | 100% | 100% | 100% | 100% | 99.6% | Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690 |
| TSC2 | 100% | 100% | 100% | 100% | 99.3% | Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254 |
| TSEN15 | 100% | 100% | 100% | 100% | 99.8% | Pontocerebellar hypoplasia, type 2F, 617026 |
| TSEN2 | 88.4% | 88.4% | 100% | 100% | 99.6% | Pontocerebellar hypoplasia type 2B, 612389 |
| TSEN54 | 100% | 100% | 100% | 100% | 99.7% | Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204 |
| TSFM | 91.5% | 91.1% | 100% | 100% | 99.3% | Combined oxidative phosphorylation deficiency 3, 610505 |
| TSHB | 100% | 100% | 100% | 100% | 99.7% | Hypothyroidism, congenital, nongoitrous 4, 275100 |
| TSPAN7 | 100% | 100% | 99.2% | 90.3% | 72.7% | Intellectual developmental disorder, X-linked 58, 300210 |
| TSPOAP1 | 100% | 100% | 100% | 100% | 99.5% | Dystonia 22, juvenile-onset, 620453;?Dystonia 22, adult-onset, 620456 |
| TTC19 | 100% | 100% | 100% | 100% | 99.7% | Mitochondrial complex III deficiency, nuclear type 2, 615157 |

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| TTC5 | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244 |
| TTC8 | 91.4% | 91.4% | 100% | 100% | 99.9% | Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464 |
| TTI1 | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445 |
| TTI2 | 100% | 100% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal recessive 39, 615541 |
| TUBA1A | 100% | 100% | 100% | 100% | 99.6% | Lissencephaly 3, 611603 |
| TUBA8 | 95.3% | 95.1% | 100% | 99.8% | 99.2% | Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840 |
| TUBB | 99.8% | 98.9% | 100% | 100% | 99.8% | Symmetric circumferential skin creases, congenital, 1, 156610;Cortical dysplasia, complex, with other brain malformations 6, 615771 |
| TUBB2A | 100% | 100% | 100% | 100% | 99% | Cortical dysplasia, complex, with other brain malformations 5, 615763 |
| TUBB2B | 100% | 100% | 100% | 100% | 99.4% | Cortical dysplasia, complex, with other brain malformations 7, 610031 |
| TUBB3 | 100% | 100% | 100% | 100% | 99.5% | Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039 |

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| TUBB4A | 99.2% | 96.6% | 100% | 100% | 99% | Dystonia 4, torsion, autosomal dominant, 128101;Leukodystrophy, hypomyelinating, 6, 612438 |
| TUBG1 | 100% | 100% | 100% | 100% | 99.5% | Cortical dysplasia, complex, with other brain malformations 4, 615412 |
| TUBGCP2 | 97% | 97% | 100% | 100% | 99.6% | Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 |
| TUBGCP4 | 100% | 100% | 100% | 99.8% | 98.7% | Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 |
| TUBGCP6 | 100% | 100% | 100% | 99.9% | 99.2% | Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 |
| TUSC3 | 93.3% | 93.3% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal recessive 7, 611093 |
| TWIST1 | 100% | 100% | 100% | 100% | 98.5% | Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 |
| 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 |

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| U2AF2 | 100% | 100% | 100% | 99.9% | 99.2% | Developmental delay, dysmorphic facies, and brain anomalies, 620535 |
| UBA2 | 100% | 100% | 100% | 100% | 99.6% | ACCES syndrome, 619959 |
| UBA5 | 94.1% | 92.2% | 100% | 100% | 99.8% | ?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132 |
| UBAP2L | 100% | 100% | 100% | 100% | 99.5% | Neurodevelopmental disorder with impaired language, behavioral abnormalities, and dysmorphic facies, 620494 |
| UBE2A | 95.6% | 91.8% | 99% | 90.8% | 72.2% | Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 |
| UBE3A | 100% | 100% | 100% | 100% | 99.7% | Angelman syndrome, 105830 |
| UBE3B | 100% | 100% | 100% | 100% | 99.8% | Kaufman oculocerebrofacial syndrome, 244450 |
| UBE3C | 94.8% | 94.8% | 100% | 100% | 99.6% | Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270 |
| UBE4A | 100% | 100% | 100% | 100% | 99.7% | Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639 |
| UBR1 | 98% | 97.9% | 100% | 100% | 99.7% | Johanson-Blizzard syndrome, 243800 |
| UBR5 | 100% | 100% | 100% | 100% | 99.8% | Neurodevelopmental disorder with speech delay and behavioral abnormalities, 621372 |

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| UBR7 | 100% | 100% | 100% | 100% | 99.4% | Li-Campeau syndrome, 619189 |
| UBTF | 100% | 100% | 100% | 100% | 99.4% | Neurodegeneration, childhood-onset, with brain atrophy, 617672 |
| UFC1 | 100% | 100% | 100% | 99.9% | 99.6% | Neurodevelopmental disorder with spasticity and poor growth, 618076 |
| UFM1 | 100% | 100% | 100% | 100% | 99.8% | Leukodystrophy, hypomyelinating, 14, 617899 |
| UFSP2 | 100% | 100% | 100% | 100% | 99.8% | ?Hip dysplasia, Beukes type, 142669;Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974;Developmental and epileptic encephalopathy 106, 620028 |
| UGDH | 97% | 94% | 100% | 100% | 99.7% | Developmental and epileptic encephalopathy 84, 618792 |
| UGGT1 | 100% | 100% | 100% | 100% | 99.8% | Congenital disorder of glycosylation, type IICC, 621381 |
| UGP2 | 91.8% | 90.6% | 100% | 100% | 99.8% | Developmental and epileptic encephalopathy 83, 618744 |
| UNC13A | 100% | 100% | 100% | 99.9% | 99% | ?Intellectual development disorder with seizures and dysmorphic facies, 621457;Neurodevelopmental disorder with speech delay, movement abnormalities, and seizures, 621456;Neurodevelopmental disorder with hypotonia, epilepsy, and absent speech, 621455 |

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| UNC45A | 100% | 100% | 100% | 100% | 99.4% | Osteootohepatoenteric syndrome, 619377 |
| UNC79 | 100% | 100% | 100% | 100% | 99.7% | |
| UNC80 | 100% | 100% | 100% | 100% | 99.7% | Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 |
| UPB1 | 94.3% | 91.8% | 100% | 100% | 99.5% | Beta-ureidopropionase deficiency, 613161 |
| UPF1 | 98.7% | 96.5% | 100% | 100% | 99.5% | |
| UPF3B | 92.5% | 92.5% | 99% | 90.2% | 71.7% | Intellectual developmental disorder, X-linked syndromic 14, 300676 |
| UROC1 | 96.6% | 96.6% | 100% | 100% | 99.6% | ?Urocanase deficiency, 276880 |
| USP14 | 100% | 100% | 100% | 100% | 99.8% | |
| USP27X | 100% | 100% | 99.3% | 92.3% | 72.5% | Intellectual developmental disorder, X-linked 105, 300984 |
| USP34 | 100% | 100% | 100% | 100% | 99.9% | |
| USP7 | 100% | 100% | 100% | 100% | 99.3% | Hao-Fountain syndrome, 616863 |
| USP9X | 100% | 100% | 99.4% | 91.8% | 75% | Intellectual developmental disorder, X-linked 99, 300919;Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968 |
| VAMP1 | 100% | 100% | 100% | 99.9% | 98.9% | Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600 |

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| VAMP2 | 100% | 100% | 100% | 100% | 99.5% | Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 |
| VARS1 | 100% | 100% | 100% | 100% | 99.3% | Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 |
| VARS2 | 100% | 100% | 100% | 100% | 98.9% | Combined oxidative phosphorylation deficiency 20, 615917 |
| 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 |
| VIPAS39 | 99.8% | 97.9% | 100% | 100% | 99.7% | Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 |
| VLDLR | 100% | 100% | 100% | 100% | 99.8% | Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050 |
| VMA22 | 100% | 98.5% | 100% | 99.9% | 97.9% | Congenital disorder of glycosylation, type IIo, 616828 |
| VPS11 | 100% | 100% | 100% | 100% | 99.6% | ?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683 |
| VPS13B | 100% | 100% | 100% | 100% | 99.7% | Cohen syndrome, 216550 |
| VPS16 | 100% | 100% | 100% | 100% | 99.6% | Dystonia 30, 619291 |

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|---|---|---|---|---|---|---|
| VPS35L | 93.6% | 93% | 100% | 100% | 99.4% | Ritscher-Schinzel syndrome 3, 619135 |
| VPS37A | 92.3% | 92.3% | 100% | 99.9% | 99.5% | Spastic paraplegia 53, autosomal recessive, 614898 |
| VPS41 | 98.4% | 96.8% | 100% | 100% | 99.9% | Spinocerebellar ataxia, autosomal recessive 29, 619389 |
| VPS4A | 100% | 100% | 100% | 99.9% | 98.9% | CIMDAG syndrome, 619273 |
| VPS50 | 96.2% | 95.3% | 100% | 100% | 99.7% | Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685 |
| VPS51 | 100% | 100% | 100% | 100% | 99% | Pontocerebellar hypoplasia, type 13, 618606 |
| VPS53 | 83.1% | 81.5% | 100% | 99.9% | 99.4% | Pontocerebellar hypoplasia, type 2E, 615851 |
| VRK1 | 99.6% | 98% | 100% | 100% | 99.9% | Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542 |
| VWA3B | 100% | 100% | 100% | 100% | 99.7% | ?Spinocerebellar ataxia, autosomal recessive 22, 616948 |
| WAC | 94% | 94% | 100% | 100% | 99.6% | Desanto-Shinawi syndrome, 616708 |
| WAPL | 97.6% | 97.6% | 100% | 100% | 99.7% | |
| WARS1 | 91.8% | 91.8% | 100% | 100% | 99.6% | Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721;Neurodevelop mental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317 |

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|---|---|---|---|---|---|---|
| WARS2 | 93.8% | 93.8% | 100% | 100% | 100% | Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 |
| WASF1 | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with absent language and variable seizures, 618707 |
| WASHC4 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 43, 615817 |
| WASHC5 | 100% | 100% | 100% | 100% | 99.8% | Ritscher-Schinzel syndrome 1, 220210;Spastic paraplegia 8, autosomal dominant, 603563 |
| WBP4 | 100% | 100% | 100% | 100% | 99.2% | Neurodevelopemental disorder with hypotonia, feeding difficulties, facial dysmorphism, and brain abnormalities, 620852 |
| WDFY3 | 100% | 100% | 100% | 100% | 99.8% | ?Microcephaly 18, primary, autosomal dominant, 617520 |
| WDPCP | 92.1% | 92.1% | 100% | 100% | 99.8% | Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 |
| WDR11 | 100% | 100% | 100% | 100% | 99.9% | Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 |
| WDR13 | 100% | 100% | 98% | 88% | 69.1% | |

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|---|---|---|---|---|---|---|
| WDR26 | 93.8% | 93.8% | 100% | 100% | 99.5% | Skraban-Deardorff syndrome, 617616 |
| WDR37 | 90.2% | 89.9% | 100% | 100% | 99.8% | Neurooculocardiogenitourinary syndrome, 618652 |
| WDR4 | 100% | 100% | 100% | 99.9% | 99.3% | Galloway-Mowat syndrome 6, 618347;Microcephaly, growth deficiency, seizures, and brain malformations, 618346 |
| WDR44 | 91.1% | 91.1% | 99.4% | 92.1% | 73.3% | |
| WDR45 | 98.4% | 98% | 98.4% | 87.9% | 68.6% | Neurodegeneration with brain iron accumulation 5, 300894 |
| WDR45B | 89.6% | 89.6% | 100% | 100% | 99.6% | Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 |
| WDR47 | 100% | 100% | 100% | 100% | 99.9% | |
| WDR5 | 93.9% | 93.9% | 100% | 100% | 99.6% | |
| WDR62 | 98.8% | 98.8% | 100% | 100% | 99.6% | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 |
| WDR73 | 100% | 100% | 100% | 100% | 99.3% | Galloway-Mowat syndrome 1, 251300 |
| WDR81 | 100% | 100% | 100% | 100% | 99.5% | Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185;Hydrocephalus, congenital, 3, with brain anomalies, 617967 |
| WDR83OS | 100% | 100% | 100% | 100% | 98.4% | Neurodevelopmental disorder with variable familial hypercholanemia, 621016 |

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|---|---|---|---|---|---|---|
| WDTC1 | 100% | 100% | 100% | 100% | 99.4% | |
| WFS1 | 91.5% | 90.5% | 100% | 100% | 99.4% | Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;{Diabetes mellitus, noninsulin-dependent, association with}, 125853;Wolfram syndrome 1, 222300 |
| WIPI2 | 100% | 100% | 100% | 100% | 99.5% | ?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453 |
| WLS | 100% | 100% | 100% | 100% | 99.9% | Zaki syndrome, 619648 |
| WNK3 | 100% | 100% | 99.4% | 91.4% | 73.7% | Prieto syndrome, 309610 |
| WNT1 | 100% | 100% | 100% | 99.9% | 99% | {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221;Osteogenesis imperfecta, type XV, 615220 |
| WSB2 | 100% | 100% | 100% | 99.9% | 99.1% | Luo-Agrawal neurodevelopmental syndrome, 621552 |
| WWOX | 100% | 100% | 100% | 100% | 99.7% | Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322 |
| XPA | 100% | 100% | 100% | 100% | 99.9% | Xeroderma pigmentosum, group A, 278700 |
| XPO1 | 100% | 99.9% | 100% | 100% | 99.9% | |
| XPOT | 89.4% | 89.4% | 100% | 100% | 99.9% | |

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|---|---|---|---|---|---|---|
| XRCC4 | 100% | 100% | 100% | 100% | 99.9% | Short stature, microcephaly, and endocrine dysfunction, 616541 |
| XYLT1 | 100% | 100% | 100% | 99.9% | 99.3% | Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800 |
| 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 |
| YIF1B | 90% | 90% | 100% | 99.9% | 99.4% | Kaya-Barakat-Masson syndrome, 619125 |
| YIPF5 | 100% | 100% | 100% | 100% | 100% | Microcephaly, epilepsy, and diabetes syndrome 2, 619278 |
| YME1L1 | 96.7% | 96.7% | 100% | 100% | 99.7% | ?Optic atrophy 11, 617302 |
| YWHAE | 100% | 100% | 100% | 100% | 99.9% | |
| YWHAG | 100% | 100% | 100% | 100% | 99.4% | Developmental and epileptic encephalopathy 56, 617665 |
| YWHAZ | 100% | 100% | 100% | 99.9% | 99.1% | Popov-Chang syndrome, 618428 |
| YY1 | 100% | 100% | 100% | 99.6% | 96.6% | Gabriele-de Vries syndrome, 617557 |
| YY1AP1 | 100% | 100% | 100% | 100% | 99.9% | Grange syndrome, 602531 |
| ZBTB11 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 69, 618383 |
| ZBTB16 | 87.8% | 87.8% | 100% | 100% | 99.3% | Leukemia, acute promyelocytic, PL2F/RARA type |

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|---|---|---|---|---|---|---|
| ZBTB18 | 93.2% | 93.2% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 22, 612337 |
| ZBTB20 | 100% | 100% | 100% | 99.9% | 99.1% | Primrose syndrome, 259050 |
| ZBTB24 | 100% | 100% | 100% | 100% | 99.7% | Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 |
| ZBTB47 | 99.6% | 99.5% | 100% | 99.9% | 98.8% | |
| ZBTB7A | 95.4% | 95.4% | 100% | 99.9% | 97.7% | Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769 |
| ZC3H14 | 100% | 100% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal recessive 56, 617125 |
| ZC4H2 | 100% | 100% | 98.3% | 88.3% | 72.7% | Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041 |
| ZDHHC9 | 100% | 100% | 99% | 89.7% | 72.4% | Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799 |
| ZEB2 | 100% | 100% | 100% | 99.9% | 99.4% | Mowat-Wilson syndrome, 235730 |
| ZFHX3 | 100% | 100% | 100% | 100% | 99.4% | Prostate cancer, somatic, 176807;{Atrial fibrillation 8, susceptibility to}, 613055;Spinocerebellar ataxia 4, 600223;Epilepsy, idiopathic generalized 20, 621500 |
| ZFHX4 | 100% | 100% | 100% | 100% | 99.6% | |

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|---|---|---|---|---|---|---|
| ZFTRAF1 | 75.9% | 72.6% | 100% | 99.6% | 97.8% | |
| ZFX | 100% | 100% | 98.7% | 90.4% | 73.6% | Intellectual developmental disorder, X-linked syndromic 37, 301118 |
| ZFYVE26 | 100% | 100% | 100% | 100% | 99.7% | Spastic paraplegia 15, autosomal recessive, 270700 |
| ZIC1 | 100% | 100% | 100% | 99.9% | 98.6% | ?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 |
| ZIC2 | 100% | 100% | 99.9% | 99.6% | 97% | Holoprosencephaly 5, 609637 |
| ZMIZ1 | 100% | 99.5% | 100% | 100% | 99.3% | Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 |
| ZMYM2 | 100% | 100% | 100% | 100% | 99.9% | Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522 |
| ZMYM3 | 100% | 100% | 98.9% | 89.9% | 71.3% | Intellectual developmental disorder, X-linked 112, 301111 |
| ZMYND11 | 100% | 99.2% | 100% | 100% | 99.7% | Intellectual developmental disorder, autosomal dominant 30, 616083 |
| ZMYND8 | 98% | 98% | 100% | 100% | 99.5% | |
| ZNF142 | 100% | 100% | 100% | 100% | 99.6% | Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 |

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|---|---|---|---|---|---|---|
| ZNF148 | 94.9% | 94.9% | 100% | 100% | 99.8% | Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 |
| ZNF292 | 99.4% | 99.4% | 100% | 100% | 99.8% | Intellectual developmental disorder, autosomal dominant 64, 619188 |
| ZNF335 | 100% | 100% | 100% | 100% | 99.5% | Microcephaly 10, primary, autosomal recessive, 615095 |
| ZNF407 | 100% | 100% | 100% | 100% | 99.8% | SIMHA syndrome, 619557 |
| ZNF41 | 100% | 100% | 99.2% | 91% | 73.3% | |
| ZNF462 | 100% | 100% | 100% | 100% | 99.6% | Weiss-Kruszka syndrome, 618619 |
| ZNF526 | 100% | 100% | 100% | 100% | 99.3% | Dentici-Novelli neurodevelopmental syndrome, 619877 |
| ZNF668 | 100% | 100% | 100% | 100% | 99.4% | Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194 |
| ZNF699 | 100% | 100% | 100% | 99.9% | 99.3% | DEGCAGS syndrome, 619488 |
| ZNF711 | 100% | 100% | 99.3% | 91.1% | 73.2% | Intellectual developmental disorder, X-linked 97, 300803 |
| ZNF865 | 100% | 100% | 100% | 100% | 98.9% | |
| ZNFX1 | 100% | 100% | 100% | 100% | 99.6% | Immunodeficiency 91 and hyperinflammation, 619644 |
| ZNHIT3 | 78.8% | 76.2% | 100% | 100% | 99.4% | PEHO syndrome, 260565 |
| ZNRF3 | 98.4% | 96.9% | 100% | 99.9% | 99.2% | |
| ZRSR2 | 84.4% | 84.4% | 98.7% | 88.9% | 70.2% | Orofaciodigital syndrome XXI, 301132 |

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| ZSCAN10 | 100% | 100% | 100% | 100% | 99.2% | Otofacial neurodevelopmental syndrome, 620910 |
| ZSWIM6 | 97.9% | 96.3% | 99.2% | 98.1% | 96.1% | Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865;Acromelic frontonasal dysostosis, 603671 |

*Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.*

*TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry 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 5.0.0*
*Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*