# INTELLECTUAL DISABILITY GENE PANEL DG 3.5.0 (1649 genes)

Releasedate: 05-12-2022

| Gene | TWIST X2 covered >10x | TWIST X2 covered >20x | Associated Phenotype description and OMIM disease ID |
|---|---|---|---|
| AAAS | 100% | 100% | Achalasia-addisonianism-alacrimia syndrome, 231550 |
| AARS1 | 100% | 100% | Developmental and epileptic encephalopathy 29, 616339<br>Charcot-Marie-Tooth disease, axonal, type 2N, 613287<br>?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661<br>Trichothiodystrophy 8, nonphotosensitive, 619691 |
| AASS | 100% | 100% | Hyperlysinemia, 238700 |
| ABAT | 100% | 100% | GABA-transaminase deficiency, 613163 |
| ABCA2 | 100% | 100% | Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 |
| ABCC8 | 100% | 100% | Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857<br>Diabetes mellitus, transient neonatal 2, 610374<br>Diabetes mellitus, noninsulin-dependent, 125853<br>Hypoglycemia of infancy, leucine-sensitive, 240800<br>Hyperinsulinemic hypoglycemia, familial, 1, 256450 |
| ABCC9 | 100% | 100% | Cardiomyopathy, dilated, 1O, 608569<br>Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850<br>?Atrial fibrillation, familial, 12, 614050<br>Intellectual disability and myopathy syndrome, 619719 |
| ABCD1 | 100% | 100% | Adrenoleukodystrophy, 300100<br>Adrenomyeloneuropathy, adult, 300100 |
| ABCD4 | 100% | 100% | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 |
| ABHD16A | 100% | 100% | Spastic paraplegia 86, autosomal recessive, 619735 |
| ABHD5 | 100% | 100% | Chanarin-Dorfman syndrome, 275630 |
| ACAD9 | 100% | 100% | Mitochondrial complex I deficiency, nuclear type 20, 611126 |
| ACADS | 100% | 100% | Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 |
| ACADSB | 100% | 100% | 2-methylbutyrylglycinuria, 610006 |
| ACAT1 | 100% | 100% | Alpha-methylacetoacetic aciduria, 203750 |
| ACER3 | 100% | 100% | ?Leukodystrophy, progressive, early childhood-onset, 617762 |
| ACO2 | 100% | 100% | Optic atrophy 9, 616289<br>Infantile cerebellar-retinal degeneration, 614559 |
| ACOX1 | 100% | 100% | Mitchell syndrome, 618960<br>Peroxisomal acyl-CoA oxidase deficiency, 264470 |
| ACSF3 | 100% | 100% | Combined malonic and methylmalonic aciduria, 614265 |
| ACSL4 | 100% | 100% | Intellectual developmental disorder, X-linked 63, 300387 |
| ACTB | 100% | 100% | Baraitser-Winter syndrome 1, 243310<br>?Dystonia, juvenile-onset, 607371 |

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| *ACTG1* | *100%* | *100%* | *Deafness, autosomal dominant 20/26, 604717*<br>*Baraitser-Winter syndrome 2, 614583* |
| *ACTL6A* | *100%* | *100%* | *No OMIM disease ID* |
| *ACTL6B* | *100%* | *100%* | *Developmental and epileptic encephalopathy 76, 618468*<br>*Intellectual developmental disorder with severe speech and ambulation defects, 618470* |
| *ACVR1* | *100%* | *100%* | *Fibrodysplasia ossificans progressiva, 135100* |
| *ACY1* | *100%* | *100%* | *Aminoacylase 1 deficiency, 609924* |
| *ADAM22* | *100%* | *100%* | *Developmental and epileptic encephalopathy 61, 617933* |
| *ADAR* | *100%* | *100%* | *Dyschromatosis symmetrica hereditaria, 127400*<br>*Aicardi-Goutieres syndrome 6, 615010* |
| *ADARB1* | *95%* | *95%* | *Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862* |
| *ADAT3* | *100%* | *100%* | *Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286* |
| *ADD1* | *100%* | *100%* | *No OMIM disease ID* |
| *ADD3* | *100%* | *100%* | *Cerebral palsy, spastic quadriplegic, 3, 617008* |
| *ADGRG1* | *100%* | *100%* | *Polymicrogyria, bilateral frontoparietal, 606854*<br>*Polymicrogyria, bilateral perisylvian, 615752* |
| *ADK* | *91%* | *91%* | *Hypermethioninemia due to adenosine kinase deficiency, 614300* |
| *ADNP* | *100%* | *100%* | *Helsmoortel-van der Aa syndrome, 615873* |
| *ADPRS* | *100%* | *100%* | *Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170* |
| *ADSL* | *100%* | *100%* | *Adenylosuccinase deficiency, 103050* |
| *AFF2* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 109, 309548* |
| *AFF3* | *100%* | *100%* | *KINSSHIP syndrome, 619297* |
| *AFF4* | *100%* | *100%* | *CHOPS syndrome, 616368* |
| *AFG3L2* | *100%* | *100%* | *Spastic ataxia 5, autosomal recessive, 614487*<br>*Optic atrophy 12, 618977*<br>*Spinocerebellar ataxia 28, 610246* |
| *AGA* | *100%* | *100%* | *Aspartylglucosaminuria, 208400* |
| *AGAP1* | *100%* | *100%* | *No OMIM disease ID* |
| *AGMO* | *100%* | *100%* | *No OMIM disease ID* |
| *AGO1* | *100%* | *100%* | *No OMIM disease ID* |
| *AGO2* | *100%* | *100%* | *Lessel-Kreienkamp syndrome, 619149* |
| *AGTPBP1* | *100%* | *100%* | *Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276* |
| *AHCY* | *100%* | *100%* | *Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752* |
| *AHDC1* | *100%* | *100%* | *Xia-Gibbs syndrome, 615829* |
| *AHI1* | *100%* | *100%* | *Joubert syndrome 3, 608629* |

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| *AHSG* | *100%* | *100%* | *?Alopecia-intellectual disability syndrome 1, 203650* |
| *AIFM1* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 6, 300816*<br>*Cowchock syndrome, 310490*<br>*Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232*<br>*Deafness, X-linked 5, 300614* |
| *AIMP1* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 3, 260600* |
| *AIMP2* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 17, 618006* |
| *AKT3* | *100%* | *100%* | *Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937* |
| *ALDH18A1* | *100%* | *100%* | *Spastic paraplegia 9A, autosomal dominant, 601162*<br>*Cutis laxa, autosomal recessive, type IIIA, 219150*<br>*Spastic paraplegia 9B, autosomal recessive, 616586*<br>*Cutis laxa, autosomal dominant 3, 616603* |
| *ALDH3A2* | *94%* | *94%* | *Sjogren-Larsson syndrome, 270200* |
| *ALDH4A1* | *100%* | *100%* | *Hyperprolinemia, type II, 239510* |
| *ALDH5A1* | *100%* | *100%* | *Succinic semialdehyde dehydrogenase deficiency, 271980* |
| *ALDH7A1* | *100%* | *100%* | *Epilepsy, pyridoxine-dependent, 266100* |
| *ALG1* | *100%* | *100%* | *Congenital disorder of glycosylation, type Ik, 608540* |
| *ALG11* | *96%* | *96%* | *Congenital disorder of glycosylation, type Ip, 613661* |
| *ALG12* | *100%* | *100%* | *Congenital disorder of glycosylation, type Ig, 607143* |
| *ALG13* | *100%* | *99%* | *Developmental and epileptic encephalopathy 36, 300884* |
| *ALG14* | *100%* | *100%* | *Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031*<br>*Myopathy, epilepsy, and progressive cerebral atrophy, 619036*<br>*?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227* |
| *ALG2* | *100%* | *100%* | *Congenital disorder of glycosylation, type Ii, 607906*<br>*Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228* |
| *ALG3* | *100%* | *100%* | *Congenital disorder of glycosylation, type Id, 601110* |
| *ALG6* | *100%* | *100%* | *Congenital disorder of glycosylation, type Ic, 603147* |
| *ALG8* | *96%* | *96%* | *Congenital disorder of glycosylation, type Ih, 608104*<br>*Polycystic liver disease 3 with or without kidney cysts, 617874* |
| *ALG9* | *100%* | *100%* | *Gillessen-Kaesbach-Nishimura syndrome, 263210*<br>*Congenital disorder of glycosylation, type Il, 608776* |
| *ALKBH8* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 71, 618504* |
| *ALMS1* | *100%* | *100%* | *Alstrom syndrome, 203800* |
| *ALX3* | *100%* | *100%* | *Frontonasal dysplasia 1, 136760* |
| *ALX4* | *100%* | *100%* | *Parietal foramina 2, 609597*<br>*Frontonasal dysplasia 2, 613451* |

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| *AMER1* | *100%* | *100%* | *Osteopathia striata with cranial sclerosis, 300373* |
| *AMMECR1* | *100%* | *100%* | *Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990* |
| *AMPD2* | *100%* | *100%* | *?Spastic paraplegia 63, 615686*<br>*Pontocerebellar hypoplasia, type 9, 615809* |
| *AMT* | *100%* | *100%* | *Glycine encephalopathy, 605899* |
| *ANK2* | *100%* | *100%* | *Long QT syndrome 4, 600919*<br>*Cardiac arrhythmia, ankyrin-B-related, 600919* |
| *ANK3* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 37, 615493* |
| *ANKH* | *100%* | *100%* | *Chondrocalcinosis 2, 118600*<br>*Craniometaphyseal dysplasia, 123000* |
| *ANKLE2* | *100%* | *100%* | *Microcephaly 16, primary, autosomal recessive, 616681* |
| *ANKRD11* | *100%* | *100%* | *KBG syndrome, 148050* |
| *ANKRD17* | *100%* | *100%* | *Chopra-Amiel-Gordon syndrome, 619504* |
| *ANKS1B* | *100%* | *100%* | *No OMIM disease ID* |
| *ANO10* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 10, 613728* |
| *ANTXR1* | *100%* | *100%* | *GAPO syndrome, 230740* |
| *AP1G1* | *100%* | *100%* | *Usmani-Riazuddin syndrome, autosomal recessive, 619548*<br>*Usmani-Riazuddin syndrome, autosomal dominant, 619467* |
| *AP1S1* | *100%* | *100%* | *MEDNIK syndrome, 609313* |
| *AP1S2* | *100%* | *100%* | *Pettigrew syndrome, 304340* |
| *AP2M1* | *100%* | *100%* | *Intellectual developmental disorder 60 with seizures, 618587* |
| *AP2S1* | *100%* | *100%* | *Hypocalciuric hypercalcemia, type III, 600740* |
| *AP3B1* | *100%* | *100%* | *Hermansky-Pudlak syndrome 2, 608233* |
| *AP3B2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 48, 617276* |
| *AP3D1* | *100%* | *100%* | *?Hermansky-Pudlak syndrome 10, 617050* |
| *AP4B1* | *100%* | *100%* | *Spastic paraplegia 47, autosomal recessive, 614066* |
| *AP4E1* | *100%* | *100%* | *Stuttering, familial persistent, 1, 184450*<br>*Spastic paraplegia 51, autosomal recessive, 613744* |
| *AP4M1* | *100%* | *100%* | *Spastic paraplegia 50, autosomal recessive, 612936* |
| *AP4S1* | *87%* | *87%* | *Spastic paraplegia 52, autosomal recessive, 614067* |
| *APC2* | *100%* | *100%* | *Cortical dysplasia, complex, with other brain malformations 10, 618677*<br>*Intellectual developmental disorder, autosomal recessive 74, 617169* |
| *APTX* | *100%* | *100%* | *Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920* |
| *ARCN1* | *100%* | *100%* | *Short stature-micrognathia syndrome, 617164* |
| *ARF1* | *100%* | *100%* | *Periventricular nodular heterotopia 8, 618185* |

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| *ARF3* | *100%* | *100%* | *No OMIM disease ID* |
| *ARFGEF1* | *100%* | *100%* | *Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964* |
| *ARFGEF2* | *100%* | *100%* | *Periventricular heterotopia with microcephaly, 608097* |
| *ARG1* | *93%* | *93%* | *Argininemia, 207800* |
| *ARHGAP31* | *100%* | *100%* | *Adams-Oliver syndrome 1, 100300* |
| *ARHGAP35* | *100%* | *100%* | *No OMIM disease ID* |
| *ARHGEF6* | *100%* | *100%* | *No OMIM disease ID* |
| *ARHGEF9* | *97%* | *96%* | *Developmental and epileptic encephalopathy 8, 300607* |
| *ARID1A* | *100%* | *100%* | *Coffin-Siris syndrome 2, 614607* |
| *ARID1B* | *99%* | *98%* | *Coffin-Siris syndrome 1, 135900* |
| *ARID2* | *100%* | *100%* | *Coffin-Siris syndrome 6, 617808* |
| *ARL13B* | *100%* | *100%* | *Joubert syndrome 8, 612291* |
| *ARL6* | *100%* | *100%* | *Retinitis pigmentosa 55, 613575*<br>*Bardet-Biedl syndrome 3, 600151* |
| *ARMC9* | *100%* | *100%* | *Joubert syndrome 30, 617622* |
| *ARPC4* | *100%* | *100%* | *No OMIM disease ID* |
| *ARSA* | *100%* | *100%* | *Metachromatic leukodystrophy, 250100* |
| *ARSL* | *100%* | *100%* | *Chondrodysplasia punctata, X-linked recessive, 302950* |
| *ARV1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 38, 617020* |
| *ARX* | *99%* | *97%* | *Proud syndrome, 300004*<br>*Hydranencephaly with abnormal genitalia, 300215*<br>*Partington syndrome, 309510*<br>*Developmental and epileptic encephalopathy 1, 308350*<br>*Lissencephaly, X-linked 2, 300215*<br>*Intellectual developmental disorder, X-linked 29, 300419* |
| *ASAH1* | *100%* | *100%* | *Spinal muscular atrophy with progressive myoclonic epilepsy, 159950*<br>*Farber lipogranulomatosis, 228000* |
| *ASH1L* | *99%* | *99%* | *Intellectual developmental disorder, autosomal dominant 52, 617796* |
| *ASL* | *100%* | *100%* | *Argininosuccinic aciduria, 207900* |
| *ASNS* | *100%* | *100%* | *Asparagine synthetase deficiency, 615574* |
| *ASPA* | *100%* | *100%* | *Canavan disease, 271900* |
| *ASPM* | *100%* | *100%* | *Microcephaly 5, primary, autosomal recessive, 608716* |
| *ASS1* | *100%* | *100%* | *Citrullinemia, 215700* |
| *ASXL1* | *100%* | *100%* | *Myelodysplastic syndrome, somatic, 614286*<br>*Bohring-Opitz syndrome, 605039* |

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| *ASXL2* | *100%* | *100%* | *Shashi-Pena syndrome, 617190* |
| *ASXL3* | *100%* | *100%* | *Bainbridge-Ropers syndrome, 615485* |
| *ATAD1* | *100%* | *100%* | *Hyperekplexia 4, 618011* |
| *ATAD3A* | *100%* | *100%* | *Harel-Yoon syndrome, 617183*<br>*Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810* |
| *ATG7* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 31, 619422* |
| *ATIC* | *100%* | *100%* | *AICA-ribosiduria due to ATIC deficiency, 608688* |
| *ATL1* | *100%* | *100%* | *Spastic paraplegia 3A, autosomal dominant, 182600*<br>*Neuropathy, hereditary sensory, type ID, 613708* |
| *ATN1* | *100%* | *100%* | *Dentatorubral-pallidoluysian atrophy, 125370*<br>*Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494* |
| *ATP13A2* | *100%* | *100%* | *Spastic paraplegia 78, autosomal recessive, 617225*<br>*Kufor-Rakeb syndrome, 606693* |
| *ATP1A1* | *100%* | *100%* | *Hypomagnesemia, seizures, and impaired intellectual development 2, 618314*<br>*Charcot-Marie-Tooth disease, axonal, type 2DD, 618036* |
| *ATP1A2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 98, 619605*<br>*Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602*<br>*Alternating hemiplegia of childhood 1, 104290*<br>*Migraine, familial basilar, 602481*<br>*Migraine, familial hemiplegic, 2, 602481* |
| *ATP1A3* | *100%* | *100%* | *Alternating hemiplegia of childhood 2, 614820*<br>*Dystonia-12, 128235*<br>*CAPOS syndrome, 601338*<br>*Developmental and epileptic encephalopathy 99, 619606* |
| *ATP2A2* | *100%* | *100%* | *Acrokeratosis verruciformis, 101900*<br>*Darier disease, 124200* |
| *ATP2B1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 66, 619910* |
| *ATP6AP1* | *100%* | *100%* | *Immunodeficiency 47, 300972* |
| *ATP6AP2* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423*<br>*?Parkinsonism with spasticity, X-linked, 300911*<br>*Congenital disorder of glycosylation, type IIr, 301045* |
| *ATP6V0A1* | *100%* | *100%* | *Neurodevelopmental disorder with epilepsy and brain atrophy, 619971*<br>*Developmental and epileptic encephalopathy 104, 619970* |
| *ATP6V0A2* | *100%* | *100%* | *Wrinkly skin syndrome, 278250*<br>*Cutis laxa, autosomal recessive, type IIA, 219200* |
| *ATP6V0C* | *100%* | *100%* | *No OMIM disease ID* |

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| *ATP6V1A* | *100%* | *100%* | *Cutis laxa, autosomal recessive, type IID, 617403*<br>*Developmental and epileptic encephalopathy 93, 618012* |
| *ATP6V1B2* | *100%* | *100%* | *Zimmermann-Laband syndrome 2, 616455*<br>*Deafness, congenital, with onychodystrophy, autosomal dominant, 124480* |
| *ATP7A* | *100%* | *100%* | *Occipital horn syndrome, 304150*<br>*Spinal muscular atrophy, distal, X-linked 3, 300489*<br>*Menkes disease, 309400* |
| *ATP8A2* | *100%* | *100%* | *?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268* |
| *ATP9A* | *100%* | *100%* | *No OMIM disease ID* |
| *ATR* | *100%* | *100%* | *Seckel syndrome 1, 210600*<br>*?Cutaneous telangiectasia and cancer syndrome, familial, 614564* |
| *ATRX* | *100%* | *100%* | *Alpha-thalassemia/mental retardation syndrome, 301040*<br>*Alpha-thalassemia myelodysplasia syndrome, somatic, 300448*<br>*Intellectual disability-hypotonic facies syndrome, X-linked, 309580* |
| *ATXN2L* | *100%* | *100%* | *No OMIM disease ID* |
| *AUH* | *100%* | *100%* | *3-methylglutaconic aciduria, type I, 250950* |
| *AUTS2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 26, 615834* |
| *AVPR2* | *100%* | *100%* | *Diabetes insipidus, nephrogenic, 1, 304800*<br>*Nephrogenic syndrome of inappropriate antidiuresis, 300539* |
| *B3GALNT2* | *92%* | *92%* | *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181* |
| *B3GALT6* | *100%* | *98%* | *Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349*<br>*Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640*<br>*Al-Gazali syndrome, 609465* |
| *B3GLCT* | *100%* | *100%* | *Peters-plus syndrome, 261540* |
| *B4GALNT1* | *100%* | *100%* | *Spastic paraplegia 26, autosomal recessive, 609195* |
| *B4GALT1* | *100%* | *100%* | *Congenital disorder of glycosylation, type IId, 607091* |
| *B4GALT7* | *100%* | *100%* | *Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070* |
| *B4GAT1* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287* |
| *B9D1* | *100%* | *100%* | *?Meckel syndrome 9, 614209*<br>*Joubert syndrome 27, 617120* |
| *B9D2* | *100%* | *100%* | *?Meckel syndrome 10, 614175*<br>*Joubert syndrome 34, 614175* |
| *BAP1* | *100%* | *100%* | *Kury-Isidor syndrome, 619762*<br>*Tumor predisposition syndrome 1, 614327* |
| *BAZ2B* | *100%* | *100%* | *No OMIM disease ID* |
| *BBS1* | *100%* | *100%* | *Bardet-Biedl syndrome 1, 209900* |

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| *BBS10* | *100%* | *100%* | *Bardet-Biedl syndrome 10, 615987* |
| *BBS12* | *100%* | *100%* | *Bardet-Biedl syndrome 12, 615989* |
| *BBS2* | *100%* | *100%* | *Retinitis pigmentosa 74, 616562*<br>*Bardet-Biedl syndrome 2, 615981* |
| *BBS4* | *100%* | *100%* | *Bardet-Biedl syndrome 4, 615982* |
| *BBS5* | *100%* | *100%* | *Bardet-Biedl syndrome 5, 615983* |
| *BBS7* | *100%* | *100%* | *Bardet-Biedl syndrome 7, 615984* |
| *BBS9* | *96%* | *96%* | *Bardet-Biedl syndrome 9, 615986* |
| *BCAP31* | *99%* | *93%* | *Deafness, dystonia, and cerebral hypomyelination, 300475* |
| *BCAS3* | *100%* | *100%* | *Hengel-Maroofian-Schols syndrome, 619641* |
| *BCKDHA* | *100%* | *100%* | *Maple syrup urine disease, type Ia, 248600* |
| *BCKDHB* | *100%* | *100%* | *Maple syrup urine disease, type Ib, 248600* |
| *BCKDK* | *100%* | *100%* | *Branched-chain keto acid dehydrogenase kinase deficiency, 614923* |
| *BCL11A* | *100%* | *100%* | *Dias-Logan syndrome, 617101* |
| *BCL11B* | *100%* | *100%* | *Immunodeficiency 49, severe combined, 617237*<br>*Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092* |
| *BCOR* | *100%* | *100%* | *Microphthalmia, syndromic 2, 300166* |
| *BCORL1* | *100%* | *100%* | *Shukla-Vernon syndrome, 301029* |
| *BCS1L* | *100%* | *100%* | *GRACILE syndrome, 603358*<br>*Mitochondrial complex III deficiency, nuclear type 1, 124000*<br>*Bjornstad syndrome, 262000* |
| *BICRA* | *100%* | *100%* | *Coffin-Siris syndrome 12, 619325* |
| *BLM* | *100%* | *100%* | *Bloom syndrome, 210900* |
| *BLOC1S1* | *100%* | *100%* | *No OMIM disease ID* |
| *KIAA1109* | *100%* | *100%* | *Alkuraya-Kucinskas syndrome, 617822* |
| *BOLA3* | *100%* | *100%* | *Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299* |
| *BPTF* | *100%* | *100%* | *Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755* |
| *BRAF* | *100%* | *100%* | *Melanoma, malignant, somatic, 155600*<br>*LEOPARD syndrome 3, 613707*<br>*Cardiofaciocutaneous syndrome, 115150*<br>*Adenocarcinoma of lung, somatic, 211980*<br>*Noonan syndrome 7, 613706*<br>*Colorectal cancer, somatic, 114500*<br>*Nonsmall cell lung cancer, somatic, 211980* |
| *BRAT1* | *100%* | *100%* | *Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056*<br>*Rigidity and multifocal seizure syndrome, lethal neonatal, 614498* |

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| *BRF1* | *100%* | *100%* | *Cerebellofaciodental syndrome, 616202* |
| *BRPF1* | *100%* | *100%* | *Intellectual developmental disorder with dysmorphic facies and ptosis, 617333* |
| *BRSK2* | *100%* | *100%* | *No OMIM disease ID* |
| *BRWD3* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 93, 300659* |
| *BSCL2* | *100%* | *100%* | *Lipodystrophy, congenital generalized, type 2, 269700*<br>*Neuropathy, distal hereditary motor, type VC, 619112*<br>*Silver spastic paraplegia syndrome, 270685*<br>*Encephalopathy, progressive, with or without lipodystrophy, 615924* |
| *BTD* | *94%* | *94%* | *Biotinidase deficiency, 253260* |
| *BUB1B* | *100%* | *100%* | *Colorectal cancer, somatic, 114500*<br>*Mosaic variegated aneuploidy syndrome 1, 257300* |
| *C12orf4* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 66, 618221* |
| *C12orf57* | *100%* | *100%* | *Temtamy syndrome, 218340* |
| *C2CD3* | *96%* | *96%* | *Orofaciodigital syndrome XIV, 615948* |
| *CA2* | *100%* | *100%* | *Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730* |
| *CA5A* | *100%* | *100%* | *Hyperammonemia due to carbonic anhydrase VA deficiency, 615751* |
| *CA8* | *100%* | *100%* | *Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227* |
| *CACNA1A* | *100%* | *100%* | *Spinocerebellar ataxia 6, 183086*<br>*Episodic ataxia, type 2, 108500*<br>*Developmental and epileptic encephalopathy 42, 617106*<br>*Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500*<br>*Migraine, familial hemiplegic, 1, 141500* |
| *CACNA1B* | *100%* | *100%* | *Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497* |
| *CACNA1C* | *100%* | *100%* | *Timothy syndrome, 601005*<br>*Long QT syndrome 8, 618447*<br>*Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029*<br>*Brugada syndrome 3, 611875* |
| *CACNA1D* | *100%* | *100%* | *Primary aldosteronism, seizures, and neurologic abnormalities, 615474*<br>*Sinoatrial node dysfunction and deafness, 614896* |
| *CACNA1E* | *100%* | *100%* | *Developmental and epileptic encephalopathy 69, 618285* |
| *CACNA1G* | *100%* | *100%* | *Spinocerebellar ataxia 42, 616795*<br>*Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087* |
| *CACNA1I* | *100%* | *100%* | *Neurodevelopmental disorder with speech impairment and with or without seizures, 620114* |
| *CACNA2D1* | *100%* | *100%* | *No OMIM disease ID* |
| *CACNA2D2* | *100%* | *100%* | *Cerebellar atrophy with seizures and variable developmental delay, 618501* |
| *CAD* | *100%* | *100%* | *Developmental and epileptic encephalopathy 50, 616457* |

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| *CAMK2A* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 53, 617798*<br>*?Intellectual developmental disorder, autosomal recessive 63, 618095* |
| *CAMK2B* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 54, 617799* |
| *CAMK2G* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 59, 618522* |
| *CAMK4* | *100%* | *100%* | *No OMIM disease ID* |
| *CAMTA1* | *100%* | *100%* | *Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756* |
| *CANT1* | *100%* | *100%* | *Desbuquois dysplasia 1, 251450*<br>*Epiphyseal dysplasia, multiple, 7, 617719* |
| *CAPN15* | *100%* | *100%* | *Oculogastrointestinal neurodevelopmental syndrome, 619318* |
| *CARS1* | *100%* | *100%* | *Microcephaly, developmental delay, and brittle hair syndrome, 618891* |
| *CARS2* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 27, 616672* |
| *CASK* | *100%* | *100%* | *Intellectual developmental disorder, with or without nystagmus, 300422*<br>*Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749*<br>*FG syndrome 4, 300422* |
| *CBL* | *100%* | *100%* | *Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563*<br>*?Juvenile myelomonocytic leukemia, 607785* |
| *CBS* | *100%* | *100%* | *Thrombosis, hyperhomocysteinemic, 236200*<br>*Homocystinuria, B6-responsive and nonresponsive types, 236200* |
| *CC2D1A* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 3, 608443* |
| *CC2D2A* | *98%* | *98%* | *COACH syndrome 2, 619111*<br>*Retinitis pigmentosa 93, 619845*<br>*Meckel syndrome 6, 612284*<br>*Joubert syndrome 9, 612285* |
| *CCBE1* | *100%* | *100%* | *Hennekam lymphangiectasia-lymphedema syndrome 1, 235510* |
| *CCDC115* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIo, 616828* |
| *CCDC174* | *100%* | *100%* | *Hypotonia, infantile, with psychomotor retardation, 616816* |
| *CCDC186* | *100%* | *100%* | *No OMIM disease ID* |
| *CCDC22* | *100%* | *100%* | *Ritscher-Schinzel syndrome 2, 300963* |
| *CCDC32* | *100%* | *100%* | *Cardiofacioneurodevelopmental syndrome, 619123* |
| *CCDC47* | *100%* | *100%* | *Trichohepatoneurodevelopmental syndrome, 618268* |
| *CCDC88A* | *97%* | *97%* | *?PEHO syndrome-like, 617507* |
| *CCDC88C* | *100%* | *100%* | *?Spinocerebellar ataxia 40, 616053*<br>*Hydrocephalus, congenital, 1, 236600* |
| *CCND2* | *100%* | *100%* | *Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938* |
| *CCNK* | *99%* | *96%* | *?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147* |
| *CDC42* | *100%* | *100%* | *Takenouchi-Kosaki syndrome, 616737* |

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| *CDC42BPB* | *100%* | *100%* | *Chilton-Okur-Chung neurodevelopmental syndrome, 619841* |
| *CDC6* | *100%* | *100%* | *?Meier-Gorlin syndrome 5, 613805* |
| *CDH11* | *100%* | *100%* | *Teebi hypertelorism syndrome 2, 619736*<br>*Elsahy-Waters syndrome, 211380* |
| *CDH15* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 3, 612580* |
| *CDH2* | *100%* | *100%* | *Arrhythmogenic right ventricular dysplasia, familial, 14, 618920*<br>*?Attention deficit-hyperactivity disorder 8, 619957*<br>*Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929* |
| *CDK10* | *100%* | *100%* | *Al Kaissi syndrome, 617694* |
| *CDK13* | *100%* | *100%* | *Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360* |
| *CDK19* | *100%* | *100%* | *Developmental and epileptic encephalopathy 87, 618916* |
| *CDK5RAP2* | *100%* | *100%* | *Microcephaly 3, primary, autosomal recessive, 604804* |
| *CDK8* | *100%* | *100%* | *Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748* |
| *CDKL5* | *96%* | *95%* | *Developmental and epileptic encephalopathy 2, 300672* |
| *CDKN1C* | *100%* | *100%* | *IMAGE syndrome, 614732*<br>*Beckwith-Wiedemann syndrome, 130650* |
| *CDON* | *100%* | *100%* | *Holoprosencephaly 11, 614226* |
| *CELF2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 97, 619561* |
| *CENPF* | *100%* | *100%* | *Stromme syndrome, 243605* |
| *CENPJ* | *100%* | *100%* | *Microcephaly 6, primary, autosomal recessive, 608393*<br>*?Seckel syndrome 4, 613676* |
| *CEP104* | *100%* | *100%* | *Joubert syndrome 25, 616781*<br>*Intellectual developmental disorder, autosomal recessive 77, 619988* |
| *CEP120* | *100%* | *100%* | *Short-rib thoracic dysplasia 13 with or without polydactyly, 616300*<br>*Joubert syndrome 31, 617761* |
| *CEP135* | *100%* | *100%* | *Microcephaly 8, primary, autosomal recessive, 614673* |
| *CEP152* | *100%* | *100%* | *Microcephaly 9, primary, autosomal recessive, 614852*<br>*Seckel syndrome 5, 613823* |
| *CEP290* | *100%* | *100%* | *Leber congenital amaurosis 10, 611755*<br>*Joubert syndrome 5, 610188*<br>*Senior-Loken syndrome 6, 610189*<br>*?Bardet-Biedl syndrome 14, 615991*<br>*Meckel syndrome 4, 611134* |
| *CEP41* | *100%* | *100%* | *Joubert syndrome 15, 614464* |
| *CEP55* | *100%* | *100%* | *Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500* |
| *CEP57* | *100%* | *100%* | *Mosaic variegated aneuploidy syndrome 2, 614114* |

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| *CEP63* | *100%* | *100%* | *?Seckel syndrome 6, 614728* |
| *CEP83* | *100%* | *100%* | *Nephronophthisis 18, 615862* |
| *CEP85L* | *100%* | *100%* | *Lissencephaly 10, 618873* |
| *CEP89* | *100%* | *100%* | *No OMIM disease ID* |
| *CERT1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 34, 616351* |
| *CHAMP1* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579* |
| *CHD1* | *100%* | *100%* | *Pilarowski-Bjornsson syndrome, 617682* |
| *CHD2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 94, 615369* |
| *CHD3* | *100%* | *100%* | *Snijders Blok-Campeau syndrome, 618205* |
| *CHD4* | *100%* | *100%* | *Sifrim-Hitz-Weiss syndrome, 617159* |
| *CHD5* | *100%* | *100%* | *Parenti-Mignot neurodevelopmental syndrome, 619873* |
| *CHD7* | *100%* | *100%* | *Hypogonadotropic hypogonadism 5 with or without anosmia, 612370*<br>*CHARGE syndrome, 214800* |
| *CHD8* | *100%* | *100%* | *Intellectual developmental disorder with autism and macrocephaly, 615032* |
| *CHKA* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023* |
| *CHKB* | *100%* | *100%* | *Muscular dystrophy, congenital, megaconial type, 602541* |
| *CHMP1A* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 8, 614961* |
| *CHRM1* | *100%* | *100%* | *No OMIM disease ID* |
| *CHRNA4* | *100%* | *100%* | *Epilepsy, nocturnal frontal lobe, 1, 600513* |
| *CIC* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 45, 617600* |
| *CIT* | *100%* | *100%* | *Microcephaly 17, primary, autosomal recessive, 617090* |
| *CKAP2L* | *100%* | *100%* | *Filippi syndrome, 272440* |
| *CLCN3* | *97%* | *97%* | *Neurodevelopmental disorder with seizures and brain abnormalities, 619517*<br>*Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512* |
| *CLCN4* | *100%* | *100%* | *Raynaud-Claes syndrome, 300114* |
| *CLDN11* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 22, 619328* |
| *CLIC2* | *100%* | *100%* | *?Intellectual developmental disorder, X-linked syndromic 32, 300886* |
| *CLIP1* | *100%* | *100%* | *No OMIM disease ID* |
| *CLN3* | *93%* | *93%* | *Ceroid lipofuscinosis, neuronal, 3, 204200* |
| *CLN5* | *83%* | *83%* | *Ceroid lipofuscinosis, neuronal, 5, 256731* |
| *CLN6* | *100%* | *100%* | *Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300*<br>*Ceroid lipofuscinosis, neuronal, 6A, 601780* |
| *CLN8* | *100%* | *100%* | *Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003*<br>*Ceroid lipofuscinosis, neuronal, 8, 600143* |

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| *CLP1* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 10, 615803* |
| *CLPB* | *100%* | *100%* | *Neutropenia, severe congenital, 9, autosomal dominant, 619813*<br>*3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271*<br>*3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835* |
| *CLTC* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 56, 617854* |
| *CNKSR2* | *100%* | *99%* | *Intellectual developmental disorder, X-linked syndromic, Houge type, 301008* |
| *CNNM2* | *100%* | *100%* | *Hypomagnesemia 6, renal, 613882*<br>*Hypomagnesemia, seizures, and impaired intellectual development 1, 616418* |
| *CNOT1* | *100%* | *100%* | *Vissers-Bodmer syndrome, 619033*<br>*Holoprosencephaly 12, with or without pancreatic agenesis, 618500* |
| *CNOT2* | *100%* | *100%* | *Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608* |
| *CNOT3* | *100%* | *100%* | *Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672* |
| *CNPY3* | *100%* | *100%* | *Developmental and epileptic encephalopathy 60, 617929* |
| *CNTNAP1* | *100%* | *100%* | *Lethal congenital contracture syndrome 7, 616286*<br>*Hypomyelinating neuropathy, congenital, 3, 618186* |
| *CNTNAP2* | *100%* | *100%* | *Pitt-Hopkins like syndrome 1, 610042* |
| *COA8* | *100%* | *100%* | *Mitochondrial complex IV deficiency, nuclear type 17, 619061* |
| *COASY* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 12, 618266*<br>*Neurodegeneration with brain iron accumulation 6, 615643* |
| *COG1* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIg, 611209* |
| *COG4* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIj, 613489*<br>*Saul-Wilson syndrome, 618150* |
| *COG5* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIi, 613612* |
| *COG6* | *100%* | *100%* | *Shaheen syndrome, 615328*<br>*Congenital disorder of glycosylation, type III, 614576* |
| *COG7* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIe, 608779* |
| *COG8* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIh, 611182* |
| *COL4A1* | *100%* | *100%* | *?Retinal arteries, tortuosity of, 180000*<br>*Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773*<br>*Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564*<br>*Brain small vessel disease with or without ocular anomalies, 175780* |
| *COL4A2* | *100%* | *100%* | *Brain small vessel disease 2, 614483* |
| *COLEC11* | *100%* | *100%* | *3MC syndrome 2, 265050* |
| *COPB1* | *100%* | *100%* | *Baralle-Macken syndrome, 619255* |
| *COPB2* | *100%* | *100%* | *Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884*<br>*?Microcephaly 19, primary, autosomal recessive, 617800* |

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| *COQ2* | *96%* | *96%* | *Coenzyme Q10 deficiency, primary, 1, 607426* |
| *COQ4* | *100%* | *100%* | *Coenzyme Q10 deficiency, primary, 7, 616276* |
| *COQ8A* | *100%* | *100%* | *Coenzyme Q10 deficiency, primary, 4, 612016* |
| *COQ9* | *100%* | *100%* | *Coenzyme Q10 deficiency, primary, 5, 614654* |
| *COX10* | *100%* | *100%* | *Mitochondrial complex IV deficiency, nuclear type 3, 619046* |
| *COX15* | *100%* | *100%* | *Mitochondrial complex IV deficiency, nuclear type 6, 615119* |
| *COX16* | *100%* | *100%* | *Mitochondrial complex IV deficiency, nuclear type 22, 619355* |
| *COX6B1* | *100%* | *100%* | *Mitochondrial complex IV deficiency, nuclear type 7, 619051* |
| *CPE* | *100%* | *100%* | *BDV syndrome, 619326* |
| *CPLANE1* | *100%* | *100%* | *Orofaciodigital syndrome VI, 277170*<br>*Joubert syndrome 17, 614615* |
| *CPLX1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 63, 617976* |
| *CPS1* | *100%* | *100%* | *Carbamoylphosphate synthetase I deficiency, 237300* |
| *CPSF3* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876* |
| *CRADD* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499* |
| *CRBN* | *100%* | *99%* | *Intellectual developmental disorder, autosomal recessive 2, 607417* |
| *CREBBP* | *100%* | *100%* | *Menke-Hennekam syndrome 1, 618332*<br>*Rubinstein-Taybi syndrome 1, 180849* |
| *CRLF1* | *100%* | *99%* | *Cold-induced sweating syndrome 1, 272430* |
| *CRPPA* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052*<br>*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643* |
| *CSDE1* | *100%* | *100%* | *No OMIM disease ID* |
| *CSF1R* | *100%* | *100%* | *Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476*<br>*Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820* |
| *CSNK1G1* | *100%* | *100%* | *No OMIM disease ID* |
| *CSNK2A1* | *94%* | *94%* | *Okur-Chung neurodevelopmental syndrome, 617062* |
| *CSNK2B* | *100%* | *100%* | *Poirier-Bienvenu neurodevelopmental syndrome, 618732* |
| *CSPP1* | *100%* | *100%* | *Joubert syndrome 21, 615636* |
| *CSTB* | *100%* | *100%* | *Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800* |
| *CTBP1* | *100%* | *100%* | *Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915* |
| *CTC1* | *100%* | *100%* | *Cerebroretinal microangiopathy with calcifications and cysts, 612199* |
| *CTCF* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 21, 615502* |
| *CTDP1* | *100%* | *100%* | *Congenital cataracts, facial dysmorphism, and neuropathy, 604168* |
| *CTNNA2* | *100%* | *99%* | *Cortical dysplasia, complex, with other brain malformations 9, 618174* |

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| *CTNNB1* | *100%* | *100%* | *Exudative vitreoretinopathy 7, 617572*<br>*Pilomatricoma, somatic, 132600*<br>*Colorectal cancer, somatic, 114500*<br>*Neurodevelopmental disorder with spastic diplegia and visual defects, 615075*<br>*Medulloblastoma, somatic, 155255*<br>*Ovarian cancer, somatic, 167000*<br>*Hepatocellular carcinoma, somatic, 114550* |
| *CTNND1* | *100%* | *100%* | *Blepharocheilodontic syndrome 2, 617681* |
| *CTNND2* | *100%* | *100%* | *No OMIM disease ID* |
| *CTSA* | *100%* | *100%* | *Galactosialidosis, 256540* |
| *CTSD* | *100%* | *100%* | *Ceroid lipofuscinosis, neuronal, 10, 610127* |
| *CTTNBP2* | *100%* | *100%* | *No OMIM disease ID* |
| *CTU2* | *100%* | *100%* | *Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142* |
| *CUL3* | *100%* | *100%* | *Neurodevelopmental disorder with or without autism or seizures, 619239*<br>*Pseudohypoaldosteronism, type IIE, 614496* |
| *CUL4B* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354* |
| *CUX1* | *100%* | *100%* | *Global developmental delay with or without impaired intellectual development, 618330* |
| *CUX2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 67, 618141* |
| *CWC27* | *100%* | *100%* | *Retinitis pigmentosa with or without skeletal anomalies, 250410* |
| *CWF19L1* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 17, 616127* |
| *CYB5R3* | *100%* | *100%* | *Methemoglobinemia, type I, 250800*<br>*Methemoglobinemia, type II, 250800* |
| *CYFIP2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 65, 618008* |
| *CYP27A1* | *100%* | *100%* | *Cerebrotendinous xanthomatosis, 213700* |
| *CYP2U1* | *100%* | *100%* | *Spastic paraplegia 56, autosomal recessive, 615030* |
| *D2HGDH* | *100%* | *100%* | *D-2-hydroxyglutaric aciduria, 600721* |
| *DAG1* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538*<br>*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818* |
| *DARS1* | *100%* | *100%* | *Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281* |
| *DARS2* | *100%* | *100%* | *Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105* |
| *DBT* | *100%* | *100%* | *Maple syrup urine disease, type II, 248600* |
| *DCAF17* | *100%* | *100%* | *Woodhouse-Sakati syndrome, 241080* |
| *DCC* | *100%* | *100%* | *Mirror movements 1 and/or agenesis of the corpus callosum, 157600*<br>*Esophageal carcinoma, somatic, 133239* |

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| | | | *Colorectal cancer, somatic, 114500*<br>*Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542* |
| *DCHS1* | *100%* | *100%* | *Mitral valve prolapse 2, 607829*<br>*Van Maldergem syndrome 1, 601390* |
| *DCPS* | *100%* | *100%* | *Al-Raqad syndrome, 616459* |
| *DCX* | *99%* | *99%* | *Subcortical laminal heterotopia, X-linked, 300067*<br>*Lissencephaly, X-linked, 300067* |
| *DDB1* | *100%* | *100%* | *White-Kernohan syndrome, 619426* |
| *DDC* | *100%* | *100%* | *Aromatic L-amino acid decarboxylase deficiency, 608643* |
| *DDHD2* | *100%* | *100%* | *Spastic paraplegia 54, autosomal recessive, 615033* |
| *DDX11* | *100%* | *100%* | *Warsaw breakage syndrome, 613398* |
| *DDX23* | *100%* | *100%* | *No OMIM disease ID* |
| *DDX3X* | *99%* | *98%* | *Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958* |
| *DDX59* | *100%* | *100%* | *Orofaciodigital syndrome V, 174300* |
| *DDX6* | *100%* | *100%* | *Intellectual developmental disorder with impaired language and dysmorphic facies, 618653* |
| *DEAF1* | *100%* | *100%* | *Vulto-van Silfout-de Vries syndrome, 615828*<br>*Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171* |
| *DEGS1* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 18, 618404* |
| *DENND5A* | *100%* | *100%* | *Developmental and epileptic encephalopathy 49, 617281* |
| *DEPDC5* | *100%* | *100%* | *Epilepsy, familial focal, with variable foci 1, 604364* |
| *DHCR24* | *100%* | *100%* | *Desmosterolosis, 602398* |
| *DHCR7* | *100%* | *100%* | *Smith-Lemli-Opitz syndrome, 270400* |
| *DHDDS* | *94%* | *94%* | *Developmental delay and seizures with or without movement abnormalities, 617836*<br>*?Congenital disorder of glycosylation, type 1bb, 613861*<br>*Retinitis pigmentosa 59, 613861* |
| *DHFR* | *100%* | *100%* | *Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839* |
| *DHPS* | *97%* | *93%* | *Neurodevelopmental disorder with seizures and speech and walking impairment, 618480* |
| *DHTKD1* | *100%* | *100%* | *?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025*<br>*Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750* |
| *DHX16* | *100%* | *100%* | *Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733* |
| *DHX30* | *100%* | *100%* | *Neurodevelopmental disorder with severe motor impairment and absent language, 617804* |
| *DHX37* | *100%* | *100%* | *Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731*<br>*46, XY sex reversal 11, 273250* |
| *DIAPH1* | *100%* | *100%* | *Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900*<br>*Seizures, cortical blindness, microcephaly syndrome, 616632* |
| *DIP2B* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630* |

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|---|---|---|---|
| *DIS3L2* | *100%* | *100%* | *Perlman syndrome, 267000* |
| *DKC1* | *100%* | *100%* | *Dyskeratosis congenita, X-linked, 305000* |
| *DLAT* | *100%* | *100%* | *Pyruvate dehydrogenase E2 deficiency, 245348* |
| *DLD* | *100%* | *100%* | *Dihydrolipoamide dehydrogenase deficiency, 246900* |
| *DLG3* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 90, 300850* |
| *DLG4* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 62, 618793* |
| *DLL1* | *100%* | *100%* | *Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709* |
| *DMD* | *100%* | *99%* | *Becker muscular dystrophy, 300376*<br>*Cardiomyopathy, dilated, 3B, 302045*<br>*Duchenne muscular dystrophy, 310200* |
| *DMPK* | *100%* | *100%* | *Myotonic dystrophy 1, 160900* |
| *DMXL2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 81, 618663*<br>*?Deafness, autosomal dominant 71, 617605*<br>*?Polyendocrine-polyneuropathy syndrome, 616113* |
| *DNAJC12* | *100%* | *100%* | *Hyperphenylalaninemia, mild, non-BH4-deficient, 617384* |
| *DNAJC19* | *100%* | *100%* | *3-methylglutaconic aciduria, type V, 610198* |
| *DNM1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 31, 616346* |
| *DNM1L* | *100%* | *100%* | *Optic atrophy 5, 610708*<br>*Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388* |
| *DNMT3A* | *100%* | *100%* | *Tatton-Brown-Rahman syndrome, 615879*<br>*Acute myeloid leukemia, somatic, 601626*<br>*Heyn-Sproul-Jackson syndrome, 618724* |
| *DNMT3B* | *100%* | *100%* | *Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860*<br>*Facioscapulohumeral muscular dystrophy 4, digenic, 619478* |
| *DOCK3* | *100%* | *100%* | *Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292* |
| *DOCK6* | *100%* | *100%* | *Adams-Oliver syndrome 2, 614219* |
| *DOCK7* | *100%* | *100%* | *Developmental and epileptic encephalopathy 23, 615859* |
| *DOHH* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066* |
| *DOLK* | *100%* | *100%* | *Congenital disorder of glycosylation, type Im, 610768* |
| *DONSON* | *100%* | *100%* | *Microcephaly, short stature, and limb abnormalities, 617604*<br>*Microcephaly-micromelia syndrome, 251230* |
| *DPAGT1* | *100%* | *100%* | *Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750*<br>*Congenital disorder of glycosylation, type Ij, 608093* |
| *DPF2* | *100%* | *100%* | *Coffin-Siris syndrome 7, 618027* |
| *DPH1* | *100%* | *100%* | *Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901* |
| *DPH5* | *100%* | *100%* | *Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070* |

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| *DPM1* | *99%* | *97%* | *Congenital disorder of glycosylation, type Ie, 608799* |
| *DPM2* | *100%* | *100%* | *Congenital disorder of glycosylation, type Iu, 615042* |
| *DPP6* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 33, 616311* |
| *DPYD* | *100%* | *100%* | *Dihydropyrimidine dehydrogenase deficiency, 274270*<br>*5-fluorouracil toxicity, 274270* |
| *DPYS* | *100%* | *100%* | *Dihydropyrimidinuria, 222748* |
| *DPYSL5* | *100%* | *100%* | *Ritscher-Schinzel syndrome 4, 619435* |
| *DTYMK* | *100%* | *100%* | *Neurodegeneration, childhood-onset, with progressive microcephaly, 619847* |
| *DYM* | *100%* | *100%* | *Smith-McCort dysplasia, 607326*<br>*Dyggve-Melchior-Clausen disease, 223800* |
| *DYNC1H1* | *100%* | *100%* | *Charcot-Marie-Tooth disease, axonal, type 2O, 614228*<br>*Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600*<br>*Intellectual developmental disorder, autosomal dominant 13, 614563* |
| *DYNC1I2* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492* |
| *DYRK1A* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 7, 614104* |
| *EARS2* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 12, 614924* |
| *EBF3* | *100%* | *100%* | *Hypotonia, ataxia, and delayed development syndrome, 617330* |
| *EBP* | *100%* | *100%* | *MEND syndrome, 300960*<br>*Chondrodysplasia punctata, X-linked dominant, 302960* |
| *ECHS1* | *100%* | *100%* | *Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277* |
| *EDC3* | *100%* | *100%* | *?Intellectual developmental disorder, autosomal recessive 50, 616460* |
| *EDEM3* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIv, 619493* |
| *EED* | *100%* | *100%* | *Cohen-Gibson syndrome, 617561* |
| *EEF1A2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 33, 616409*<br>*Intellectual developmental disorder, autosomal dominant 38, 616393* |
| *EFNB2* | *100%* | *100%* | *No OMIM disease ID* |
| *EFTUD2* | *100%* | *100%* | *Mandibulofacial dysostosis, Guion-Almeida type, 610536* |
| *EHMT1* | *100%* | *100%* | *Kleefstra syndrome 1, 610253* |
| *EIF2AK1* | *100%* | *100%* | *?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878* |
| *EIF2AK2* | *100%* | *100%* | *Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877*<br>*Dystonia 33, 619687* |
| *EIF2AK3* | *100%* | *100%* | *Wolcott-Rallison syndrome, 226980* |
| *EIF2B4* | *100%* | *100%* | *Ovarioleukodystrophy, 603896*<br>*Leukoencephalopathy with vanishing white matter, 603896* |
| *EIF2B5* | *100%* | *100%* | *Ovarioleukodystrophy, 603896*<br>*Leukoencephalopathy with vanishing white matter, 603896* |

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| *EIF2S3* | *100%* | *100%* | *MEHMO syndrome, 300148* |
| *EIF3F* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 67, 618295* |
| *EIF4A3* | *100%* | *100%* | *Robin sequence with cleft mandible and limb anomalies, 268305* |
| *EIF5A* | *100%* | *100%* | *Faundes-Banka syndrome, 619376* |
| *ELAC2* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 17, 615440* |
| *ELOVL4* | *100%* | *100%* | *Spinocerebellar ataxia 34, 133190*<br>*Stargardt disease 3, 600110*<br>*Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457* |
| *ELP2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 58, 617270* |
| *EMC1* | *100%* | *100%* | *Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875* |
| *EMC10* | *100%* | *100%* | *Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264* |
| *EML1* | *100%* | *100%* | *Band heterotopia, 600348* |
| *EMX2* | *100%* | *100%* | *Schizencephaly, 269160* |
| *ENTPD1* | *100%* | *100%* | *Spastic paraplegia 64, autosomal recessive, 615683* |
| *EP300* | *100%* | *100%* | *Menke-Hennekam syndrome 2, 618333*<br>*Colorectal cancer, somatic, 114500*<br>*Rubinstein-Taybi syndrome 2, 613684* |
| *EPG5* | *100%* | *100%* | *Vici syndrome, 242840* |
| *EPHA7* | *100%* | *100%* | *No OMIM disease ID* |
| *ERCC1* | *100%* | *100%* | *Cerebrooculofacioskeletal syndrome 4, 610758* |
| *ERCC2* | *100%* | *100%* | *Xeroderma pigmentosum, group D, 278730*<br>*Trichothiodystrophy 1, photosensitive, 601675*<br>*?Cerebrooculofacioskeletal syndrome 2, 610756* |
| *ERCC3* | *100%* | *100%* | *Trichothiodystrophy 2, photosensitive, 616390*<br>*Xeroderma pigmentosum, group B, 610651* |
| *ERCC5* | *100%* | *100%* | *Xeroderma pigmentosum, group G, 278780*<br>*Cerebrooculofacioskeletal syndrome 3, 616570*<br>*Xeroderma pigmentosum, group G/Cockayne syndrome, 278780* |
| *ERCC6* | *100%* | *100%* | *UV-sensitive syndrome 1, 600630*<br>*Cerebrooculofacioskeletal syndrome 1, 214150*<br>*?De Sanctis-Cacchione syndrome, 278800*<br>*Cockayne syndrome, type B, 133540*<br>*Premature ovarian failure 11, 616946* |
| *ERCC8* | *100%* | *100%* | *UV-sensitive syndrome 2, 614621*<br>*Cockayne syndrome, type A, 216400* |

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|---|---|---|---|
| *ERLIN2* | *100%* | *100%* | *Spastic paraplegia 18, autosomal recessive, 611225* |
| *ESCO2* | *100%* | *100%* | *Juberg-Hayward syndrome, 216100*<br>*Roberts-SC phocomelia syndrome, 268300* |
| *ETFB* | *100%* | *100%* | *Glutaric acidemia IIB, 231680* |
| *ETHE1* | *100%* | *100%* | *Ethylmalonic encephalopathy, 602473* |
| *EXOC2* | *100%* | *100%* | *Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306* |
| *EXOC7* | *100%* | *100%* | *Neurodevelopmental disorder with seizures and brain atrophy, 619072* |
| *EXOSC2* | *100%* | *100%* | *Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763* |
| *EXOSC3* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 1B, 614678* |
| *EXOSC8* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 1C, 616081* |
| *EXOSC9* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 1D, 618065* |
| *EXTL3* | *100%* | *100%* | *Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425* |
| *EZH2* | *100%* | *100%* | *Weaver syndrome, 277590* |
| *FA2H* | *100%* | *100%* | *Spastic paraplegia 35, autosomal recessive, 612319* |
| *FAM149B1* | *100%* | *100%* | *Joubert syndrome 36, 618763* |
| *FAM20C* | *100%* | *100%* | *Raine syndrome, 259775* |
| *FAM50A* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261* |
| *FAR1* | *100%* | *100%* | *Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154*<br>*Cataracts, spastic paraparesis, and speech delay, 619338* |
| *FARS2* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 14, 614946*<br>*Spastic paraplegia 77, autosomal recessive, 617046* |
| *FARSB* | *100%* | *100%* | *Rajab interstitial lung disease with brain calcifications 1, 613658* |
| *FAT4* | *100%* | *100%* | *Van Maldergem syndrome 2, 615546*<br>*Hennekam lymphangiectasia-lymphedema syndrome 2, 616006* |
| *FBRSL1* | *100%* | *99%* | *No OMIM disease ID* |
| *FBXL3* | *100%* | *100%* | *Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220* |
| *FBXL4* | *100%* | *100%* | *Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471* |
| *FBXO11* | *100%* | *100%* | *Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089* |
| *FBXO28* | *100%* | *100%* | *Developmental and epileptic encephalopathy 100, 619777* |
| *FBXO31* | *100%* | *100%* | *?Intellectual developmental disorder, autosomal recessive 45, 615979* |
| *FBXW11* | *100%* | *100%* | *Neurodevelopmental, jaw, eye, and digital syndrome, 618914* |
| *FBXW7* | *100%* | *98%* | *Developmental delay, hypotonia, and impaired language, 620012* |
| *FDFT1* | *100%* | *100%* | *Squalene synthase deficiency, 618156* |
| *FGD1* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic 16, 305400*<br>*Aarskog-Scott syndrome, 305400* |

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|---|---|---|---|
| *FGF12* | *100%* | *100%* | *Developmental and epileptic encephalopathy 47, 617166* |
| *FGF13* | *100%* | *100%* | *Developmental and epileptic encephalopathy 90, 301058* |
| *FGF14* | *100%* | *100%* | *Spinocerebellar ataxia 27, 193003* |
| *FGFR1* | *100%* | *100%* | *Pfeiffer syndrome, 101600*<br>*Hypogonadotropic hypogonadism 2 with or without anosmia, 147950*<br>*Jackson-Weiss syndrome, 123150*<br>*Hartsfield syndrome, 615465*<br>*Trigonocephaly 1, 190440*<br>*Osteoglophonic dysplasia, 166250*<br>*Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001* |
| *FGFR2* | *100%* | *100%* | *Bent bone dysplasia syndrome, 614592*<br>*LADD syndrome, 149730*<br>*Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410*<br>*Jackson-Weiss syndrome, 123150*<br>*Gastric cancer, somatic, 613659*<br>*Craniofacial-skeletal-dermatologic dysplasia, 101600*<br>*Apert syndrome, 101200*<br>*Pfeiffer syndrome, 101600*<br>*?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579*<br>*Beare-Stevenson cutis gyrata syndrome, 123790*<br>*Crouzon syndrome, 123500*<br>*Saethre-Chotzen syndrome, 101400*<br>*Scaphocephaly and Axenfeld-Rieger anomaly,*<br>*Craniosynostosis, nonspecific,* |
| *FGFR3* | *100%* | *100%* | *Muenke syndrome, 602849*<br>*SADDAN, 616482*<br>*Hypochondroplasia, 146000*<br>*LADD syndrome, 149730*<br>*Thanatophoric dysplasia, type II, 187601*<br>*Nevus, epidermal, somatic, 162900*<br>*CATSHL syndrome, 610474*<br>*Thanatophoric dysplasia, type I, 187600*<br>*Spermatocytic seminoma, somatic, 273300*<br>*Bladder cancer, somatic, 109800*<br>*Achondroplasia, 100800*<br>*Cervical cancer, somatic, 603956*<br>*Colorectal cancer, somatic, 114500*<br>*Crouzon syndrome with acanthosis nigricans, 612247* |

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|---|---|---|---|
| *FH* | *100%* | *100%* | *Leiomyomatosis and renal cell cancer, 150800*<br>*Fumarase deficiency, 606812* |
| *FIBP* | *100%* | *100%* | *Thauvin-Robinet-Faivre syndrome, 617107* |
| *FIG4* | *100%* | *100%* | *Yunis-Varon syndrome, 216340*<br>*?Polymicrogyria, bilateral temporooccipital, 612691*<br>*Amyotrophic lateral sclerosis 11, 612577*<br>*Charcot-Marie-Tooth disease, type 4J, 611228* |
| *FIGN* | *100%* | *100%* | *No OMIM disease ID* |
| *FKRP* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612*<br>*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155*<br>*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153* |
| *FKTN* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588*<br>*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800*<br>*Cardiomyopathy, dilated, 1X, 611615*<br>*Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152* |
| *FLNA* | *100%* | *100%* | *Otopalatodigital syndrome, type II, 304120*<br>*Intestinal pseudoobstruction, neuronal, 300048*<br>*Cardiac valvular dysplasia, X-linked, 314400*<br>*?FG syndrome 2, 300321*<br>*Melnick-Needles syndrome, 309350*<br>*Terminal osseous dysplasia, 300244*<br>*Congenital short bowel syndrome, 300048*<br>*Otopalatodigital syndrome, type I, 311300*<br>*Heterotopia, periventricular, 1, 300049*<br>*Frontometaphyseal dysplasia 1, 305620* |
| *FLVCR1* | *100%* | *100%* | *Ataxia, posterior column, with retinitis pigmentosa, 609033* |
| *FLVCR2* | *100%* | *100%* | *Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790* |
| *FMN2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 47, 616193* |
| *FMR1* | *100%* | *100%* | *Fragile X tremor/ataxia syndrome, 300623*<br>*Fragile X syndrome, 300624*<br>*Premature ovarian failure 1, 311360* |
| *FOLR1* | *100%* | *100%* | *Neurodegeneration due to cerebral folate transport deficiency, 613068* |
| *FOXG1* | *100%* | *100%* | *Rett syndrome, congenital variant, 613454* |
| *FOXJ1* | *100%* | *100%* | *Ciliary dyskinesia, primary, 43, 618699* |
| *FOXP1* | *100%* | *100%* | *Intellectual developmental disorder with language impairment with or without autistic features, 613670* |
| *FOXP2* | *100%* | *100%* | *Speech-language disorder-1, 602081* |
| *FOXRED1* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 19, 618241* |

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| *FRAS1* | *100%* | *100%* | *Fraser syndrome 1, 219000* |
| *FRMD4A* | *97%* | *97%* | *?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819* |
| *FRMPD4* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 104, 300983* |
| *FRRS1L* | *100%* | *100%* | *Developmental and epileptic encephalopathy 37, 616981* |
| *FTCD* | *100%* | *100%* | *Glutamate formiminotransferase deficiency, 229100* |
| *FTO* | *95%* | *95%* | *Growth retardation, developmental delay, facial dysmorphism, 612938* |
| *FTSJ1* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 9, 309549* |
| *FUCA1* | *100%* | *100%* | *Fucosidosis, 230000* |
| *FUT8* | *100%* | *100%* | *Congenital disorder of glycosylation with defective fucosylation 1, 618005* |
| *FZR1* | *100%* | *100%* | *No OMIM disease ID* |
| *GABBR2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 59, 617904*<br>*Neurodevelopmental disorder with poor language and loss of hand skills, 617903* |
| *GABRA1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 19, 615744* |
| *GABRA2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 78, 618557* |
| *GABRA3* | *100%* | *100%* | *Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091* |
| *GABRA5* | *100%* | *100%* | *Developmental and epileptic encephalopathy 79, 618559* |
| *GABRB1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 45, 617153* |
| *GABRB2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 92, 617829* |
| *GABRB3* | *100%* | *100%* | *Developmental and epileptic encephalopathy 43, 617113* |
| *GABRD* | *100%* | *100%* | *No OMIM disease ID* |
| *GABRG2* | *93%* | *93%* | *Developmental and epileptic encephalopathy 74, 618396*<br>*Febrile seizures, familial, 8, 607681*<br>*Generalized epilepsy with febrile seizures plus, type 3, 607681* |
| *GAD1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 89, 619124* |
| *GALC* | *100%* | *100%* | *Krabbe disease, 245200* |
| *GALE* | *100%* | *100%* | *Galactose epimerase deficiency, 230350* |
| *GALNT2* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIt, 618885* |
| *GALT* | *100%* | *100%* | *Galactosemia, 230400* |
| *GAMT* | *100%* | *100%* | *Cerebral creatine deficiency syndrome 2, 612736* |
| *GATAD2B* | *100%* | *100%* | *GAND syndrome, 615074* |
| *GATM* | *100%* | *100%* | *Cerebral creatine deficiency syndrome 3, 612718*<br>*Fanconi renotubular syndrome 1, 134600* |
| *GCH1* | *100%* | *100%* | *Dystonia, DOPA-responsive, 128230*<br>*Hyperphenylalaninemia, BH4-deficient, B, 233910* |
| *GCSH* | *100%* | *100%* | *?Glycine encephalopathy, 605899* |

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|---|---|---|---|
| *GDI1* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 41, 300849* |
| *GEMIN5* | *100%* | *100%* | *Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333* |
| *GFAP* | *100%* | *100%* | *Alexander disease, 203450* |
| *GFER* | *100%* | *100%* | *Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076* |
| *GFM1* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 1, 609060* |
| *GFM2* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 39, 618397* |
| *GIGYF1* | *100%* | *100%* | *No OMIM disease ID* |
| *GJA1* | *100%* | *100%* | *Erythrokeratodermia variabilis et progressiva 3, 617525*<br>*Craniometaphyseal dysplasia, autosomal recessive, 218400*<br>*Oculodentodigital dysplasia, 164200*<br>*Hypoplastic left heart syndrome 1, 241550*<br>*Palmoplantar keratoderma with congenital alopecia, 104100*<br>*Syndactyly, type III, 186100*<br>*Oculodentodigital dysplasia, autosomal recessive, 257850*<br>*Atrioventricular septal defect 3, 600309* |
| *GJB1* | *100%* | *100%* | *Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800* |
| *GJC2* | *100%* | *99%* | *Lymphatic malformation 3, 613480*<br>*?Spastic paraplegia 44, autosomal recessive, 613206*<br>*Leukodystrophy, hypomyelinating, 2, 608804* |
| *GK* | *100%* | *100%* | *Glycerol kinase deficiency, 307030* |
| *GLB1* | *100%* | *100%* | *GM1-gangliosidosis, type I, 230500*<br>*GM1-gangliosidosis, type III, 230650*<br>*Mucopolysaccharidosis type IVB (Morquio), 253010*<br>*GM1-gangliosidosis, type II, 230600* |
| *GLDC* | *100%* | *100%* | *Glycine encephalopathy, 605899* |
| *GLI2* | *100%* | *100%* | *Culler-Jones syndrome, 615849*<br>*Holoprosencephaly 9, 610829* |
| *GLI3* | *100%* | *100%* | *Greig cephalopolysyndactyly syndrome, 175700*<br>*Polydactyly, postaxial, types A1 and B, 174200*<br>*Pallister-Hall syndrome, 146510*<br>*Polydactyly, preaxial, type IV, 174700* |
| *GLIS3* | *100%* | *100%* | *Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199* |
| *GLRA2* | *100%* | *98%* | *Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076* |
| *GLS* | *100%* | *100%* | *Global developmental delay, progressive ataxia, and elevated glutamine, 618412*<br>*?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339*<br>*Developmental and epileptic encephalopathy 71, 618328* |

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|---|---|---|---|
| *GLUD1* | *100%* | *100%* | *Hyperinsulinism-hyperammonemia syndrome, 606762* |
| *GLUL* | *100%* | *100%* | *Glutamine deficiency, congenital, 610015* |
| *GLYCTK* | *100%* | *100%* | *D-glyceric aciduria, 220120* |
| *GM2A* | *100%* | *100%* | *GM2-gangliosidosis, AB variant, 272750* |
| *GMNN* | *100%* | *100%* | *Meier-Gorlin syndrome 6, 616835* |
| *GMPPA* | *100%* | *100%* | *Alacrima, achalasia, and impaired intellectual development syndrome, 615510* |
| *GMPPB* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352*<br>*Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351*<br>*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350* |
| *GNAI1* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854* |
| *GNAO1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 17, 615473*<br>*Neurodevelopmental disorder with involuntary movements, 617493* |
| *GNAS* | *100%* | *100%* | *ACTH-independent macronodular adrenal hyperplasia, 219080*<br>*Pituitary adenoma 3, multiple types, somatic, 617686*<br>*Pseudohypoparathyroidism Ic, 612462*<br>*Pseudohypoparathyroidism Ia, 103580*<br>*Osseous heteroplasia, progressive, 166350*<br>*Pseudohypoparathyroidism Ib, 603233*<br>*McCune-Albright syndrome, somatic, mosaic, 174800*<br>*Pseudopseudohypoparathyroidism, 612463* |
| *GNB1* | *100%* | *100%* | *Myelodysplastic syndrome, somatic, 614286*<br>*Leukemia, acute lymphoblastic, somatic, 613065*<br>*Intellectual developmental disorder, autosomal dominant 42, 616973* |
| *GNB2* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503*<br>*?Sick sinus syndrome 4, 619464* |
| *GNB5* | *100%* | *100%* | *Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182*<br>*Intellectual developmental disorder with cardiac arrhythmia, 617173* |
| *GNPAT* | *100%* | *100%* | *Rhizomelic chondrodysplasia punctata, type 2, 222765* |
| *GNPTAB* | *100%* | *100%* | *Mucolipidosis III alpha/beta, 252600*<br>*Mucolipidosis II alpha/beta, 252500* |
| *GNPTG* | *100%* | *100%* | *Mucolipidosis III gamma, 252605* |
| *GNS* | *100%* | *100%* | *Mucopolysaccharidosis type IIID, 252940* |
| *GOLGA2* | *100%* | *100%* | *No OMIM disease ID* |
| *GOT2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 82, 618721* |
| *GPAA1* | *100%* | *100%* | *Glycosylphosphatidylinositol biosynthesis defect 15, 617810* |

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| *GPC3* | *100%* | *99%* | *Wilms tumor, somatic, 194070*<br>*Simpson-Golabi-Behmel syndrome, type 1, 312870* |
| *GPC4* | *100%* | *100%* | *Keipert syndrome, 301026* |
| *GPHN* | *100%* | *100%* | *Molybdenum cofactor deficiency C, 615501* |
| *GPSM2* | *100%* | *100%* | *Chudley-McCullough syndrome, 604213* |
| *GPT2* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281* |
| *GRIA2* | *100%* | *100%* | *Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917* |
| *GRIA3* | *100%* | *99%* | *Intellectual developmental disorder, X-linked syndromic, Wu type, 300699* |
| *GRIA4* | *100%* | *100%* | *Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864* |
| *GRID2* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 18, 616204* |
| *GRIK2* | *96%* | *96%* | *Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580*<br>*Intellectual developmental disorder, autosomal recessive 6, 611092* |
| *GRIN1* | *100%* | *100%* | *Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820*<br>*Developmental and epileptic encephalopathy 101, 619814*<br>*Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254* |
| *GRIN2A* | *100%* | *99%* | *Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570* |
| *GRIN2B* | *100%* | *100%* | *Developmental and epileptic encephalopathy 27, 616139*<br>*Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970* |
| *GRIN2D* | *100%* | *99%* | *Developmental and epileptic encephalopathy 46, 617162* |
| *GRIP1* | *100%* | *100%* | *Fraser syndrome 3, 617667* |
| *GRM1* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 13, 614831*<br>*Spinocerebellar ataxia 44, 617691* |
| *GRM7* | *100%* | *100%* | *Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922* |
| *GRN* | *100%* | *100%* | *Aphasia, primary progressive, 607485*<br>*Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485*<br>*Ceroid lipofuscinosis, neuronal, 11, 614706* |
| *GSE1* | *100%* | *100%* | *No OMIM disease ID* |
| *GSS* | *100%* | *100%* | *Hemolytic anemia due to glutathione synthetase deficiency, 231900*<br>*Glutathione synthetase deficiency, 266130* |
| *GTF2E2* | *100%* | *100%* | *Trichothiodystrophy 6, nonphotosensitive, 616943* |
| *GTF2H5* | *70%* | *70%* | *Trichothiodystrophy 3, photosensitive, 616395* |
| *GTPBP2* | *100%* | *100%* | *Jaberi-Elahi syndrome, 617988* |
| *GTPBP3* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 23, 616198* |
| *GUSB* | *100%* | *100%* | *Mucopolysaccharidosis VII, 253220* |
| *H1-4* | *100%* | *100%* | *Rahman syndrome, 617537* |
| *H3-3B* | *100%* | *100%* | *Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721* |

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|---|---|---|---|
| *H4C3* | *100%* | *100%* | *Tessadori-van Haaften neurodevelopmental syndrome 1, 619758* |
| *HAAO* | *100%* | *100%* | *Vertebral, cardiac, renal, and limb defects syndrome 1, 617660* |
| *HACE1* | *100%* | *100%* | *Spastic paraplegia and psychomotor retardation with or without seizures, 616756* |
| *HADH* | *100%* | *100%* | *Hyperinsulinemic hypoglycemia, familial, 4, 609975*<br>*3-hydroxyacyl-CoA dehydrogenase deficiency, 231530* |
| *HADHA* | *100%* | *100%* | *HELLP syndrome, maternal, of pregnancy, 609016*<br>*Mitochondrial trifunctional protein deficiency, 609015*<br>*LCHAD deficiency, 609016*<br>*Fatty liver, acute, of pregnancy, 609016* |
| *HADHB* | *100%* | *100%* | *Trifunctional protein deficiency, 609015* |
| *HAX1* | *100%* | *100%* | *Neutropenia, severe congenital 3, autosomal recessive, 610738* |
| *HCCS* | *100%* | *100%* | *Linear skin defects with multiple congenital anomalies 1, 309801* |
| *HCFC1* | *100%* | *100%* | *Methylmalonic aciduria and homocysteinemia, cblX type, 309541* |
| *HCN1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 24, 615871*<br>*Generalized epilepsy with febrile seizures plus, type 10, 618482* |
| *HDAC4* | *100%* | *100%* | *Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797* |
| *HDAC6* | *100%* | *100%* | *?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863* |
| *HDAC8* | *98%* | *97%* | *Cornelia de Lange syndrome 5, 300882* |
| *HEATR3* | *100%* | *100%* | *Diamond-Blackfan anemia 21, 620072* |
| *HEATR5B* | *100%* | *100%* | *No OMIM disease ID* |
| *HECW2* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268* |
| *HEPACAM* | *100%* | *100%* | *Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925*<br>*Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926* |
| *HERC1* | *100%* | *100%* | *Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011* |
| *HERC2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 38, 615516* |
| *HESX1* | *100%* | *100%* | *Pituitary hormone deficiency, combined, 5, 182230*<br>*Septooptic dysplasia, 182230*<br>*Growth hormone deficiency with pituitary anomalies, 182230* |
| *HEXA* | *100%* | *100%* | *GM2-gangliosidosis, several forms, 272800*<br>*Tay-Sachs disease, 272800* |
| *HEXB* | *100%* | *100%* | *Sandhoff disease, infantile, juvenile, and adult forms, 268800* |
| *HGSNAT* | *92%* | *92%* | *Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930*<br>*Retinitis pigmentosa 73, 616544* |
| *HIBCH* | *100%* | *100%* | *3-hydroxyisobutryl-CoA hydrolase deficiency, 250620* |
| *HID1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983* |
| *HIVEP2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 43, 616977* |

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| *HK1* | *100%* | *100%* | *Retinitis pigmentosa 79, 617460*<br>*Neuropathy, hereditary motor and sensory, Russe type, 605285*<br>*Neurodevelopmental disorder with visual defects and brain anomalies, 618547*<br>*Hemolytic anemia due to hexokinase deficiency, 235700* |
| *HLCS* | *100%* | *100%* | *Holocarboxylase synthetase deficiency, 253270* |
| *HMGB1* | *100%* | *100%* | *No OMIM disease ID* |
| *HMGCL* | *100%* | *100%* | *HMG-CoA lyase deficiency, 246450* |
| *HNMT* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 51, 616739* |
| *HNRNPD* | *100%* | *100%* | *No OMIM disease ID* |
| *HNRNPH1* | *100%* | *100%* | *Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083* |
| *HNRNPH2* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Bain type, 300986* |
| *HNRNPK* | *100%* | *100%* | *Au-Kline syndrome, 616580* |
| *HNRNPU* | *100%* | *100%* | *Developmental and epileptic encephalopathy 54, 617391* |
| *HOXA1* | *100%* | *100%* | *Bosley-Salih-Alorainy syndrome, 601536*<br>*Athabaskan brainstem dysgenesis syndrome, 601536* |
| *HPD* | *100%* | *100%* | *Hawkinsinuria, 140350*<br>*Tyrosinemia, type III, 276710* |
| *HPDL* | *100%* | *100%* | *Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026*<br>*Spastic paraplegia 83, autosomal recessive, 619027* |
| *HPRT1* | *100%* | *100%* | *Hyperuricemia, HRPT-related, 300323*<br>*Lesch-Nyhan syndrome, 300322* |
| *HRAS* | *100%* | *100%* | *Bladder cancer, somatic, 109800*<br>*Thyroid carcinoma, follicular, somatic, 188470*<br>*Congenital myopathy with excess of muscle spindles, 218040*<br>*Nevus sebaceous or woolly hair nevus, somatic, 162900*<br>*Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200*<br>*Spitz nevus or nevus spilus, somatic, 137550*<br>*Costello syndrome, 218040* |
| *HS2ST1* | *100%* | *100%* | *Neurofacioskeletal syndrome with or without renal agenesis, 619194* |
| *HSD17B10* | *100%* | *100%* | *HSD10 mitochondrial disease, 300438* |
| *HSD17B4* | *97%* | *97%* | *D-bifunctional protein deficiency, 261515*<br>*Perrault syndrome 1, 233400* |
| *HSPA9* | *100%* | *100%* | *Even-plus syndrome, 616854*<br>*Anemia, sideroblastic, 4, 182170* |
| *HSPD1* | *100%* | *100%* | *Spastic paraplegia 13, autosomal dominant, 605280*<br>*Leukodystrophy, hypomyelinating, 4, 612233* |

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| *HTRA2* | *100%* | *100%* | *3-methylglutaconic aciduria, type VIII, 617248* |
| *HUWE1* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Turner type, 309590* |
| *FAM126A* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 5, 610532* |
| *HYLS1* | *100%* | *100%* | *Hydrolethalus syndrome, 236680* |
| *IARS1* | *100%* | *100%* | *Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093* |
| *IARS2* | *100%* | *100%* | *Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007* |
| *IBA57* | *100%* | *100%* | *Multiple mitochondrial dysfunctions syndrome 3, 615330*<br>*?Spastic paraplegia 74, autosomal recessive, 616451* |
| *IDS* | *100%* | *100%* | *Mucopolysaccharidosis II, 309900* |
| *IDUA* | *100%* | *100%* | *Mucopolysaccharidosis Is, 607016*<br>*Mucopolysaccharidosis Ih/s, 607015*<br>*Mucopolysaccharidosis Ih, 607014* |
| *IER3IP1* | *100%* | *100%* | *Microcephaly, epilepsy, and diabetes syndrome, 614231* |
| *IFIH1* | *100%* | *100%* | *Immunodeficiency 95, 619773*<br>*Aicardi-Goutieres syndrome 7, 615846*<br>*Singleton-Merten syndrome 1, 182250* |
| *IFT172* | *100%* | *100%* | *Retinitis pigmentosa 71, 616394*<br>*Bardet-Biedl syndrome 20, 619471*<br>*Short-rib thoracic dysplasia 10 with or without polydactyly, 615630* |
| *IFT27* | *100%* | *100%* | *Bardet-Biedl syndrome 19, 615996* |
| *IFT74* | *100%* | *100%* | *Bardet-Biedl syndrome 22, 617119*<br>*Spermatogenic failure 58, 619585*<br>*Joubert syndrome 40, 619582* |
| *IFT81* | *95%* | *95%* | *Short-rib thoracic dysplasia 19 with or without polydactyly, 617895* |
| *IGBP1* | *100%* | *100%* | *?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472* |
| *IGF1* | *100%* | *100%* | *Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747* |
| *IGF1R* | *100%* | *100%* | *Insulin-like growth factor I, resistance to, 270450* |
| *IKBKG* | *100%* | *98%* | *Incontinentia pigmenti, 308300*<br>*Ectodermal dysplasia and immunodeficiency 1, 300291*<br>*Immunodeficiency 33, 300636*<br>*Autoinflammatory disease, systemic, X-linked, 301081* |
| *IL1RAPL1* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 21, 300143* |
| *IMPA1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 59, 617323* |
| *INPP5E* | *100%* | *100%* | *Joubert syndrome 1, 213300*<br>*Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156* |

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| *INPP5K* | *100%* | *100%* | *Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404* |
| *INTS1* | *100%* | *100%* | *Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571* |
| *IPO8* | *100%* | *100%* | *VISS syndrome, 619472* |
| *IQSEC1* | *100%* | *100%* | *Intellectual developmental disorder with short stature and behavioral abnormalities, 618687* |
| *IQSEC2* | *100%* | *98%* | *Intellectual developmental disorder, X-linked 1, 309530* |
| *IREB2* | *100%* | *100%* | *Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451* |
| *IRF2BPL* | *100%* | *100%* | *Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088* |
| *IRX5* | *100%* | *100%* | *Hamamy syndrome, 611174* |
| *ISCA2* | *100%* | *100%* | *Multiple mitochondrial dysfunctions syndrome 4, 616370* |
| *ITGA7* | *100%* | *100%* | *Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204* |
| *ITPA* | *100%* | *100%* | *Developmental and epileptic encephalopathy 35, 616647* |
| *ITPR1* | *100%* | *100%* | *Gillespie syndrome, 206700*<br>*Spinocerebellar ataxia 29, congenital nonprogressive, 117360*<br>*Spinocerebellar ataxia 15, 606658* |
| *IVD* | *100%* | *100%* | *Isovaleric acidemia, 243500* |
| *JAG1* | *100%* | *100%* | *?Deafness, congenital heart defects, and posterior embryotoxon, 617992*<br>*Charcot-Marie-Tooth disease, axonal, type 2HH, 619574*<br>*Alagille syndrome 1, 118450*<br>*Tetralogy of Fallot, 187500* |
| *JAG2* | *100%* | *100%* | *Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566* |
| *JAM3* | *100%* | *100%* | *Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730* |
| *JARID2* | *100%* | *100%* | *Developmental delay with variable intellectual disability and dysmorphic facies, 620098* |
| *JMJD1C* | *100%* | *100%* | *No OMIM disease ID* |
| *KANK1* | *100%* | *100%* | *Cerebral palsy, spastic quadriplegic, 2, 612900* |
| *KANSL1* | *100%* | *100%* | *Koolen-De Vries syndrome, 610443* |
| *KAT5* | *100%* | *100%* | *Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103* |
| *KAT6A* | *100%* | *100%* | *Arboleda-Tham syndrome, 616268* |
| *KAT6B* | *100%* | *100%* | *SBBYSS syndrome, 603736*<br>*Genitopatellar syndrome, 606170* |
| *KAT8* | *100%* | *100%* | *Li-Ghorgani-Weisz-Hubshman syndrome, 618974* |
| *KATNB1* | *100%* | *100%* | *Lissencephaly 6, with microcephaly, 616212* |
| *KCNA2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 32, 616366* |
| *KCNA4* | *100%* | *100%* | *Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284* |
| *KCNB1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 26, 616056* |
| *KCNC1* | *100%* | *100%* | *Epilepsy, progressive myoclonic 7, 616187* |

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| *KCNC3* | *100%* | *98%* | *Spinocerebellar ataxia 13, 605259* |
| *KCNH1* | *99%* | *99%* | *Zimmermann-Laband syndrome 1, 135500*<br>*Temple-Baraitser syndrome, 611816* |
| *KCNJ10* | *100%* | *100%* | *Enlarged vestibular aqueduct, digenic, 600791*<br>*SESAME syndrome, 612780* |
| *KCNJ11* | *100%* | *100%* | *Diabetes, permanent neonatal 2, with or without neurologic features, 618856*<br>*Maturity-onset diabetes of the young, type 13, 616329*<br>*Diabetes mellitus, transient neonatal 3, 610582*<br>*Hyperinsulinemic hypoglycemia, familial, 2, 601820* |
| *KCNJ6* | *100%* | *100%* | *Keppen-Lubinsky syndrome, 614098* |
| *KCNK4* | *100%* | *100%* | *Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381* |
| *KCNK9* | *100%* | *100%* | *Birk-Barel syndrome, 612292* |
| *KCNMA1* | *100%* | *100%* | *Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446*<br>*Cerebellar atrophy, developmental delay, and seizures, 617643*<br>*Liang-Wang syndrome, 618729* |
| *KCNN2* | *100%* | *100%* | *?Dystonia 34, myoclonic, 619724*<br>*Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725* |
| *KCNN3* | *100%* | *100%* | *Zimmermann-Laband syndrome 3, 618658* |
| *KCNQ2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 7, 613720*<br>*Seizures, benign neonatal, 1, 121200*<br>*Myokymia, 121200* |
| *KCNQ3* | *100%* | *100%* | *Seizures, benign neonatal, 2, 121201* |
| *KCNQ5* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 46, 617601* |
| *KCNT1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 14, 614959*<br>*Epilepsy nocturnal frontal lobe, 5, 615005* |
| *KCNT2* | *100%* | *99%* | *Developmental and epileptic encephalopathy 57, 617771* |
| *KCTD3* | *100%* | *100%* | *No OMIM disease ID* |
| *KCTD7* | *100%* | *100%* | *Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726* |
| *KDM1A* | *100%* | *100%* | *Cleft palate, psychomotor retardation, and distinctive facial features, 616728* |
| *KDM3B* | *100%* | *100%* | *Diets-Jongmans syndrome, 618846* |
| *KDM4B* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 65, 619320* |
| *KDM5B* | *98%* | *96%* | *Intellectual developmental disorder, autosomal recessive 65, 618109* |
| *KDM5C* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534* |
| *KDM6A* | *100%* | *100%* | *Kabuki syndrome 2, 300867* |
| *KDM6B* | *100%* | *100%* | *Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505* |

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|---|---|---|---|
| *KIAA0586* | *96%* | *96%* | *Short-rib thoracic dysplasia 14 with polydactyly, 616546*<br>*Joubert syndrome 23, 616490* |
| *KIDINS220* | *100%* | *100%* | *Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296*<br>*Ventriculomegaly and arthrogryposis, 619501* |
| *KIF11* | *100%* | *100%* | *Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950* |
| *KIF14* | *100%* | *100%* | *Microcephaly 20, primary, autosomal recessive, 617914*<br>*?Meckel syndrome 12, 616258* |
| *KIF1A* | *100%* | *100%* | *NESCAV syndrome, 614255*<br>*Neuropathy, hereditary sensory, type IIC, 614213*<br>*Spastic paraplegia 30, autosomal dominant, 610357*<br>*Spastic paraplegia 30, autosomal recessive, 610357* |
| *KIF21B* | *100%* | *100%* | *No OMIM disease ID* |
| *KIF2A* | *100%* | *100%* | *Cortical dysplasia, complex, with other brain malformations 3, 615411* |
| *KIF3B* | *100%* | *100%* | *Retinitis pigmentosa 89, 618955* |
| *KIF4A* | *100%* | *100%* | *?Intellectual developmental disorder, X-linked 100, 300923* |
| *KIF5A* | *100%* | *100%* | *Myoclonus, intractable, neonatal, 617235*<br>*Spastic paraplegia 10, autosomal dominant, 604187* |
| *KIF5C* | *99%* | *99%* | *Cortical dysplasia, complex, with other brain malformations 2, 615282* |
| *KIF7* | *100%* | *100%* | *Joubert syndrome 12, 200990*<br>*Acrocallosal syndrome, 200990*<br>*?Hydrolethalus syndrome 2, 614120*<br>*?Al-Gazali-Bakalinova syndrome, 607131* |
| *KIFBP* | *96%* | *96%* | *Goldberg-Shprintzen megacolon syndrome, 609460* |
| *KIRREL3* | *100%* | *100%* | *No OMIM disease ID* |
| *KLF7* | *100%* | *100%* | *No OMIM disease ID* |
| *KLHL15* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 103, 300982* |
| *KMT2A* | *100%* | *100%* | *Wiedemann-Steiner syndrome, 605130* |
| *KMT2B* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 68, 619934*<br>*Dystonia 28, childhood-onset, 617284* |
| *KMT2C* | *100%* | *100%* | *Kleefstra syndrome 2, 617768* |
| *KMT2D* | *100%* | *100%* | *Kabuki syndrome 1, 147920* |
| *KMT2E* | *100%* | *100%* | *O'Donnell-Luria-Rodan syndrome, 618512* |
| *KMT5B* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 51, 617788* |
| *KNL1* | *99%* | *99%* | *Microcephaly 4, primary, autosomal recessive, 604321* |
| *KPTN* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 41, 615637* |

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| *KRAS* | *100%* | *100%* | *Gastric cancer, somatic, 613659*<br>*Oculoectodermal syndrome, somatic, 600268*<br>*Breast cancer, somatic, 114480*<br>*Noonan syndrome 3, 609942*<br>*RAS-associated autoimmune leukoproliferative disorder, 614470*<br>*Arteriovenous malformation of the brain, somatic, 108010*<br>*Lung cancer, somatic, 211980*<br>*Pancreatic carcinoma, somatic, 260350*<br>*Leukemia, acute myeloid, somatic, 601626*<br>*Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200*<br>*Cardiofaciocutaneous syndrome 2, 615278*<br>*Bladder cancer, somatic, 109800* |
| *L1CAM* | *100%* | *100%* | *MASA syndrome, 303350*<br>*Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000*<br>*Corpus callosum, partial agenesis of, 304100*<br>*CRASH syndrome, 303350*<br>*Hydrocephalus with Hirschsprung disease, 307000*<br>*Hydrocephalus due to aqueductal stenosis, 307000* |
| *L2HGDH* | *100%* | *100%* | *L-2-hydroxyglutaric aciduria, 236792* |
| *LAMA1* | *100%* | *100%* | *Poretti-Boltshauser syndrome, 615960* |
| *LAMA2* | *100%* | *100%* | *Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138*<br>*Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855* |
| *LAMB1* | *100%* | *100%* | *Lissencephaly 5, 615191* |
| *LAMB2* | *100%* | *100%* | *Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199*<br>*Pierson syndrome, 609049* |
| *LAMC3* | *100%* | *100%* | *Cortical malformations, occipital, 614115* |
| *LAMP2* | *100%* | *100%* | *Danon disease, 300257* |
| *LARGE1* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840*<br>*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154* |
| *LARP7* | *100%* | *100%* | *Alazami syndrome, 615071* |
| *LARS1* | *100%* | *100%* | *?Infantile liver failure syndrome 1, 615438* |
| *LAS1L* | *100%* | *100%* | *Wilson-Turner syndrome, 309585* |
| *LIAS* | *100%* | *100%* | *Hyperglycinemia, lactic acidosis, and seizures, 614462* |
| *LIG4* | *100%* | *100%* | *LIG4 syndrome, 606593* |
| *LINGO1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 64, 618103* |
| *LINS1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 27, 614340* |

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| *LMAN2L* | *100%* | *100%* | *?Intellectual developmental disorder, autosomal dominant 69, 617863*<br>*?Intellectual developmental disorder, autosomal recessive 52, 616887* |
| *LMBRD2* | *100%* | *100%* | *Developmental delay with variable neurologic and brain abnormalities, 619694* |
| *LMNB1* | *100%* | *100%* | *Leukodystrophy, adult-onset, autosomal dominant, 169500*<br>*Microcephaly 26, primary, autosomal dominant, 619179* |
| *LMNB2* | *100%* | *100%* | *Microcephaly 27, primary, autosomal dominant, 619180*<br>*?Epilepsy, progressive myoclonic, 9, 616540* |
| *LONP1* | *100%* | *100%* | *CODAS syndrome, 600373* |
| *LRP2* | *100%* | *100%* | *Donnai-Barrow syndrome, 222448* |
| *LRPPRC* | *100%* | *100%* | *Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111* |
| *LSS* | *100%* | *100%* | *Hypotrichosis 14, 618275*<br>*Cataract 44, 616509*<br>*Alopecia-intellectual disability syndrome 4, 618840* |
| *LYRM7* | *100%* | *100%* | *Mitochondrial complex III deficiency, nuclear type 8, 615838* |
| *LYST* | *100%* | *100%* | *Chediak-Higashi syndrome, 214500* |
| *LZTFL1* | *100%* | *100%* | *Bardet-Biedl syndrome 17, 615994* |
| *LZTR1* | *100%* | *100%* | *Noonan syndrome 2, 605275*<br>*Noonan syndrome 10, 616564* |
| *MAB21L1* | *100%* | *100%* | *Cerebellar, ocular, craniofacial, and genital syndrome, 618479* |
| *MAB21L2* | *100%* | *100%* | *Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877* |
| *MACF1* | *100%* | *100%* | *Lissencephaly 9 with complex brainstem malformation, 618325* |
| *MADD* | *100%* | *100%* | *Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005*<br>*DEEAH syndrome, 619004* |
| *MAF* | *94%* | *90%* | *Cataract 21, multiple types, 610202*<br>*Ayme-Gripp syndrome, 601088* |
| *MAG* | *100%* | *100%* | *Spastic paraplegia 75, autosomal recessive, 616680* |
| *MAGEL2* | *100%* | *100%* | *Schaaf-Yang syndrome, 615547* |
| *MAN1B1* | *100%* | *100%* | *Rafiq syndrome, 614202* |
| *MAN2B1* | *100%* | *100%* | *Mannosidosis, alpha-, types I and II, 248500* |
| *MAN2C1* | *100%* | *100%* | *Congenital disorder of deglycosylation 2, 619775* |
| *MANBA* | *100%* | *100%* | *Mannosidosis, beta, 248510* |
| *MAOA* | *99%* | *99%* | *Brunner syndrome, 300615* |
| *MAP1B* | *100%* | *100%* | *?Deafness, autosomal dominant 83, 619808*<br>*Periventricular nodular heterotopia 9, 618918* |
| *MAP2K1* | *100%* | *100%* | *Cardiofaciocutaneous syndrome 3, 615279*<br>*Melorheostosis, isolated, somatic mosaic, 155950* |

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| *MAP2K2* | *100%* | *100%* | *Cardiofaciocutaneous syndrome 4, 615280* |
| *MAPK1* | *100%* | *100%* | *Noonan syndrome 13, 619087* |
| *MAPK8IP3* | *100%* | *100%* | *Neurodevelopmental disorder with or without variable brain abnormalities, 618443* |
| *MAPKAPK5* | *100%* | *100%* | *Neurocardiofaciodigital syndrome, 619869* |
| *MAPRE2* | *100%* | *100%* | *Symmetric circumferential skin creases, congenital, 2, 616734* |
| *MASP1* | *100%* | *100%* | *3MC syndrome 1, 257920* |
| *MAST1* | *100%* | *100%* | *Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273* |
| *MAT1A* | *100%* | *100%* | *Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850*<br>*Methionine adenosyltransferase deficiency, autosomal recessive, 250850* |
| *MBD5* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 1, 156200* |
| *MBOAT7* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 57, 617188* |
| *MBTPS2* | *100%* | *100%* | *Keratosis follicularis spinulosa decalvans, X-linked, 308800*<br>*Osteogenesis imperfecta, type XIX, 301014*<br>*IFAP syndrome with or without BRESHECK syndrome, 308205*<br>*?Olmsted syndrome, X-linked, 300918* |
| *MCCC1* | *100%* | *100%* | *3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200* |
| *MCCC2* | *100%* | *100%* | *3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210* |
| *MCOLN1* | *100%* | *100%* | *Mucolipidosis IV, 252650* |
| *MCPH1* | *100%* | *100%* | *Microcephaly 1, primary, autosomal recessive, 251200* |
| *MDH2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 51, 617339* |
| *MECP2* | *100%* | *100%* | *Rett syndrome, atypical, 312750*<br>*Encephalopathy, neonatal severe, 300673*<br>*Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260*<br>*Intellectual developmental disorder, X-linked syndromic 13, 300055*<br>*Rett syndrome, 312750*<br>*Rett syndrome, preserved speech variant, 312750* |
| *MECR* | *100%* | *100%* | *Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282* |
| *MED12* | *100%* | *100%* | *Lujan-Fryns syndrome, 309520*<br>*Ohdo syndrome, X-linked, 300895*<br>*Hardikar syndrome, 301068*<br>*Opitz-Kaveggia syndrome, 305450* |
| *MED12L* | *100%* | *100%* | *Nizon-Isidor syndrome, 618872* |
| *MED13* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 61, 618009* |
| *MED13L* | *100%* | *100%* | *Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789* |
| *MED17* | *100%* | *100%* | *Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668* |
| *MED23* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249* |

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| *MED25* | *100%* | *100%* | *Basel-Vanagait-Smirin-Yosef syndrome, 616449* |
| *MED27* | *100%* | *100%* | *Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286* |
| *MEF2C* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443* |
| *MEGF8* | *100%* | *100%* | *Carpenter syndrome 2, 614976* |
| *MEIS2* | *100%* | *100%* | *Cleft palate, cardiac defects, and mental retardation, 600987* |
| *METTL23* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 44, 615942* |
| *METTL5* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 72, 618665* |
| *MFF* | *100%* | *100%* | *Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086* |
| *MFSD2A* | *100%* | *100%* | *Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486* |
| *MFSD8* | *100%* | *100%* | *Macular dystrophy with central cone involvement, 616170*<br>*Ceroid lipofuscinosis, neuronal, 7, 610951* |
| *MGAT2* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIa, 212066* |
| *MGP* | *100%* | *100%* | *Keutel syndrome, 245150* |
| *MIA3* | *100%* | *100%* | *?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269* |
| *MICU1* | *100%* | *100%* | *Myopathy with extrapyramidal signs, 615673* |
| *MID1* | *100%* | *99%* | *Opitz GBBB syndrome, 300000* |
| *MID2* | *100%* | *100%* | *?Intellectual developmental disorder, X-linked 101, 300928* |
| *MINPP1* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 16, 619527* |
| *MKKS* | *100%* | *100%* | *McKusick-Kaufman syndrome, 236700*<br>*Bardet-Biedl syndrome 6, 605231* |
| *MKS1* | *100%* | *100%* | *Bardet-Biedl syndrome 13, 615990*<br>*Meckel syndrome 1, 249000*<br>*Joubert syndrome 28, 617121* |
| *MLC1* | *100%* | *100%* | *Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004* |
| *MLYCD* | *100%* | *100%* | *Malonyl-CoA decarboxylase deficiency, 248360* |
| *MMAA* | *100%* | *100%* | *Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100* |
| *MMAB* | *100%* | *100%* | *Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110* |
| *MMACHC* | *100%* | *100%* | *Methylmalonic aciduria and homocystinuria, cblC type, 277400* |
| *MMADHC* | *89%* | *89%* | *Methylmalonic aciduria, cblD type, variant 2, 277410*<br>*Methylmalonic aciduria and homocystinuria, cblD type, 277410*<br>*Homocystinuria, cblD type, variant 1, 277410* |
| *MMGT1* | *100%* | *100%* | *No OMIM disease ID* |
| *MMUT* | *100%* | *100%* | *Methylmalonic aciduria, mut(0) type, 251000* |
| *MN1* | *100%* | *100%* | *CEBALID syndrome, 618774*<br>*Meningioma, 607174* |

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| *MOCS1* | *100%* | *100%* | *Molybdenum cofactor deficiency A, 252150* |
| *MOCS2* | *100%* | *100%* | *Molybdenum cofactor deficiency B, 252160* |
| *MOGS* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIb, 606056* |
| *MORC2* | *100%* | *100%* | *Charcot-Marie-Tooth disease, axonal, type 2Z, 616688*<br>*Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090* |
| *MPDU1* | *100%* | *100%* | *Congenital disorder of glycosylation, type If, 609180* |
| *MPDZ* | *100%* | *99%* | *Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219* |
| *MPLKIP* | *100%* | *100%* | *Trichothiodystrophy 4, nonphotosensitive, 234050* |
| *MPV17* | *100%* | *100%* | *Charcot-Marie-Tooth disease, axonal, type 2EE, 618400*<br>*Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810* |
| *MRAS* | *100%* | *100%* | *Noonan syndrome 11, 618499* |
| *MRPS22* | *100%* | *100%* | *Ovarian dysgenesis 7, 618117*<br>*Combined oxidative phosphorylation deficiency 5, 611719* |
| *MRPS34* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 32, 617664* |
| *MSL2* | *100%* | *100%* | *No OMIM disease ID* |
| *MSL3* | *100%* | *100%* | *Basilicata-Akhtar syndrome, 301032* |
| *MSMO1* | *100%* | *100%* | *Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834* |
| *MTFMT* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 15, 614947*<br>*Mitochondrial complex I deficiency, nuclear type 27, 618248* |
| *MTHFR* | *100%* | *100%* | *Homocystinuria due to MTHFR deficiency, 236250* |
| *MTHFS* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367* |
| *MTO1* | *94%* | *91%* | *Combined oxidative phosphorylation deficiency 10, 614702* |
| *MTOR* | *100%* | *100%* | *Focal cortical dysplasia, type II, somatic, 607341*<br>*Smith-Kingsmore syndrome, 616638* |
| *MTR* | *100%* | *100%* | *Homocystinuria-megaloblastic anemia, cblG complementation type, 250940* |
| *C12orf65* | *100%* | *100%* | *Spastic paraplegia 55, autosomal recessive, 615035*<br>*Combined oxidative phosphorylation deficiency 7, 613559* |
| *MTRR* | *100%* | *100%* | *Homocystinuria-megaloblastic anemia, cbl E type, 236270* |
| *MTSS2* | *100%* | *100%* | *Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086* |
| *MVK* | *90%* | *90%* | *Hyper-IgD syndrome, 260920*<br>*Porokeratosis 3, multiple types, 175900*<br>*Mevalonic aciduria, 610377* |
| *MYCN* | *100%* | *100%* | *Feingold syndrome 1, 164280* |
| *MYH9* | *100%* | *100%* | *Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100*<br>*Deafness, autosomal dominant 17, 603622* |

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| *MYO5A* | *100%* | *100%* | *Griscelli syndrome, type 1, 214450* |
| *MYO9A* | *100%* | *100%* | *Myasthenic syndrome, congenital, 24, presynaptic, 618198* |
| *MYT1L* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 39, 616521* |
| *NAA10* | *100%* | *100%* | *Microphthalmia, syndromic 1, 309800*<br>*Ogden syndrome, 300855* |
| *NAA15* | *97%* | *97%* | *Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787* |
| *NAA20* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 73, 619717* |
| *NACC1* | *100%* | *100%* | *Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393* |
| *NAGA* | *100%* | *100%* | *Schindler disease, type I, 609241*<br>*Kanzaki disease, 609242*<br>*Schindler disease, type III, 609241* |
| *NAGLU* | *100%* | *100%* | *?Charcot-Marie-Tooth disease, axonal, type 2V, 616491*<br>*Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920* |
| *NALCN* | *100%* | *100%* | *Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266*<br>*Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419* |
| *NANS* | *100%* | *100%* | *Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442* |
| *NAPB* | *100%* | *100%* | *Developmental and epileptic encephalopathy 107, 620033* |
| *NARS1* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092*<br>*Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091* |
| *NARS2* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 24, 616239*<br>*?Deafness, autosomal recessive 94, 618434* |
| *NAXE* | *100%* | *100%* | *Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186* |
| *NBEA* | *100%* | *99%* | *Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157* |
| *NBN* | *100%* | *100%* | *Leukemia, acute lymphoblastic, 613065*<br>*Aplastic anemia, 609135*<br>*Nijmegen breakage syndrome, 251260* |
| *NCAPG2* | *100%* | *100%* | *Khan-Khan-Katsanis syndrome, 618460* |
| *NCDN* | *100%* | *100%* | *Neurodevelopmental disorder with infantile epileptic spasms, 619373* |
| *NCKAP1* | *100%* | *100%* | *No OMIM disease ID* |
| *NDE1* | *100%* | *100%* | *Microhydranencephaly, 605013*<br>*Lissencephaly 4 (with microcephaly), 614019* |
| *NDP* | *100%* | *100%* | *Exudative vitreoretinopathy 2, X-linked, 305390*<br>*Norrie disease, 310600* |
| *NDST1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 46, 616116* |
| *NDUFA1* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 12, 301020* |
| *NDUFA11* | *100%* | *99%* | *Mitochondrial complex I deficiency, nuclear type 14, 618236* |

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| *NDUFA12* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 23, 618244* |
| *NDUFA2* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 13, 618235* |
| *NDUFA8* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 37, 619272* |
| *NDUFAF3* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 18, 618240* |
| *NDUFAF5* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 16, 618238* |
| *NDUFAF8* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 34, 618776* |
| *NDUFS1* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 5, 618226* |
| *NDUFS2* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 6, 618228* |
| *NDUFS3* | *97%* | *91%* | *Mitochondrial complex I deficiency, nuclear type 8, 618230* |
| *NDUFS4* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 1, 252010* |
| *NDUFS6* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 9, 618232* |
| *NDUFS7* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 3, 618224* |
| *NDUFS8* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 2, 618222* |
| *NDUFV1* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 4, 618225* |
| *NDUFV2* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 7, 618229* |
| *NEDD4L* | *100%* | *100%* | *Periventricular nodular heterotopia 7, 617201* |
| *NEMF* | *100%* | *100%* | *Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099* |
| *NEU1* | *100%* | *100%* | *Sialidosis, type II, 256550*<br>*Sialidosis, type I, 256550* |
| *NEUROD2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 72, 618374* |
| *NEXMIF* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 98, 300912* |
| *NF1* | *100%* | *100%* | *Watson syndrome, 193520*<br>*Leukemia, juvenile myelomonocytic, 607785*<br>*Neurofibromatosis, familial spinal, 162210*<br>*Neurofibromatosis, type 1, 162200*<br>*Neurofibromatosis-Noonan syndrome, 601321* |
| *NFE2L2* | *100%* | *100%* | *Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744* |
| *NFIA* | *100%* | *100%* | *Brain malformations with or without urinary tract defects, 613735* |
| *NFIB* | *100%* | *100%* | *Macrocephaly, acquired, with impaired intellectual development, 618286* |
| *NFIX* | *100%* | *100%* | *Marshall-Smith syndrome, 602535*<br>*Malan syndrome, 614753* |
| *NFU1* | *100%* | *100%* | *Multiple mitochondrial dysfunctions syndrome 1, 605711* |
| *NGLY1* | *100%* | *100%* | *Congenital disorder of deglycosylation 1, 615273* |
| *NHLRC2* | *100%* | *100%* | *FINCA syndrome, 618278* |

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|---|---|---|---|
| *NHS* | *100%* | *100%* | *Cataract 40, X-linked, 302200*<br>*Nance-Horan syndrome, 302350* |
| *NIPBL* | *100%* | *100%* | *Cornelia de Lange syndrome 1, 122470* |
| *NKAP* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039* |
| *NKX2-1* | *100%* | *100%* | *Chorea, hereditary benign, 118700*<br>*Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978* |
| *NLGN2* | *100%* | *100%* | *No OMIM disease ID* |
| *NLGN3* | *100%* | *100%* | *No OMIM disease ID* |
| *NLGN4X* | *100%* | *100%* | *Intellectual developmental disorder, X-linked, 300495* |
| *NONO* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic 34, 300967* |
| *NOVA2* | *100%* | *100%* | *Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859* |
| *NPC1* | *100%* | *100%* | *Niemann-Pick disease, type C1, 257220*<br>*Niemann-Pick disease, type D, 257220* |
| *NPC2* | *100%* | *100%* | *Niemann-pick disease, type C2, 607625* |
| *NPHP1* | *100%* | *100%* | *Joubert syndrome 4, 609583*<br>*Nephronophthisis 1, juvenile, 256100*<br>*Senior-Loken syndrome-1, 266900* |
| *NR2F1* | *100%* | *100%* | *Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722* |
| *NR4A2* | *100%* | *100%* | *Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911* |
| *NRAS* | *100%* | *100%* | *Noonan syndrome 6, 613224*<br>*?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470*<br>*Melanocytic nevus syndrome, congenital, somatic, 137550*<br>*Epidermal nevus, somatic, 162900*<br>*Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200*<br>*Thyroid carcinoma, follicular, somatic, 188470*<br>*Neurocutaneous melanosis, somatic, 249400*<br>*Colorectal cancer, somatic, 114500* |
| *NRCAM* | *100%* | *100%* | *Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833* |
| *NRROS* | *100%* | *100%* | *Seizures, early-onset, with neurodegeneration and brain calcification, 618875* |
| *NRXN1* | *100%* | *100%* | *Pitt-Hopkins-like syndrome 2, 614325* |
| *NSD1* | *100%* | *100%* | *Sotos syndrome, 117550* |
| *NSD2* | *100%* | *100%* | *Rauch-Steindl syndrome, 619695* |
| *NSDHL* | *100%* | *100%* | *CK syndrome, 300831*<br>*CHILD syndrome, 308050* |
| *NSF* | *100%* | *100%* | *Developmental and epileptic encephalopathy 96, 619340* |
| *NSRP1* | *91%* | *91%* | *Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001* |

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|---|---|---|---|
| *NSUN2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 5, 611091* |
| *NT5C2* | *100%* | *100%* | *Spastic paraplegia 45, autosomal recessive, 613162* |
| *NTNG2* | *100%* | *100%* | *Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718* |
| *NTRK1* | *100%* | *100%* | *Insensitivity to pain, congenital, with anhidrosis, 256800* |
| *NTRK2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 58, 617830*<br>*Obesity, hyperphagia, and developmental delay, 613886* |
| *NUBPL* | *100%* | *100%* | *Mitochondrial complex I deficiency, nuclear type 21, 618242* |
| *NUDT2* | *100%* | *100%* | *Intellectual developmental disorder with or without peripheral neuropathy, 619844* |
| *NUP107* | *100%* | *100%* | *?Ovarian dysgenesis 6, 618078*<br>*Galloway-Mowat syndrome 7, 618348*<br>*Nephrotic syndrome, type 11, 616730* |
| *NUP188* | *100%* | *100%* | *Sandestig-Stefanova syndrome, 618804* |
| *NUP214* | *100%* | *100%* | *Leukemia, T-cell acute lymphoblastic, somatic, 613065*<br>*Leukemia, acute myeloid, somatic, 601626* |
| *NUP62* | *100%* | *100%* | *Striatonigral degeneration, infantile, 271930* |
| *NUP85* | *100%* | *100%* | *Nephrotic syndrome, type 17, 618176* |
| *NUS1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831*<br>*?Congenital disorder of glycosylation, type 1aa, 617082* |
| *OAT* | *100%* | *100%* | *Gyrate atrophy of choroid and retina with or without ornithinemia, 258870* |
| *OCLN* | *100%* | *100%* | *Pseudo-TORCH syndrome 1, 251290* |
| *OCRL* | *100%* | *100%* | *Dent disease 2, 300555*<br>*Lowe syndrome, 309000* |
| *ODC1* | *100%* | *100%* | *Bachmann-Bupp syndrome, 619075* |
| *OFD1* | *100%* | *100%* | *Simpson-Golabi-Behmel syndrome, type 2, 300209*<br>*?Retinitis pigmentosa 23, 300424*<br>*Orofaciodigital syndrome I, 311200*<br>*Joubert syndrome 10, 300804* |
| *OGDHL* | *100%* | *100%* | *Yoon-Bellen neurodevelopmental syndrome, 619701* |
| *OGT* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 106, 300997* |
| *OPA3* | *100%* | *100%* | *3-methylglutaconic aciduria, type III, 258501*<br>*Optic atrophy 3 with cataract, 165300* |
| *OPHN1* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486* |
| *ORC1* | *100%* | *100%* | *Meier-Gorlin syndrome 1, 224690* |
| *OSGEP* | *100%* | *100%* | *Galloway-Mowat syndrome 3, 617729* |
| *OTC* | *100%* | *100%* | *Ornithine transcarbamylase deficiency, 311250* |

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| *OTUD5* | *100%* | *100%* | *Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056* |
| *OTUD6B* | *100%* | *100%* | *Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452* |
| *OTUD7A* | *100%* | *98%* | *No OMIM disease ID* |
| *OTX2* | *100%* | *100%* | *Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125*<br>*Pituitary hormone deficiency, combined, 6, 613986*<br>*Microphthalmia, syndromic 5, 610125* |
| *OXR1* | *100%* | *100%* | *Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000* |
| *P4HTM* | *100%* | *100%* | *Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493* |
| *PACS1* | *100%* | *100%* | *Schuurs-Hoeijmakers syndrome, 615009* |
| *PACS2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 66, 618067* |
| *PAFAH1B1* | *100%* | *100%* | *Subcortical laminar heterotopia, 607432*<br>*Lissencephaly 1, 607432* |
| *PAH* | *100%* | *100%* | *Phenylketonuria, 261600* |
| *PAK1* | *100%* | *100%* | *Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158* |
| *PAK3* | *100%* | *99%* | *Intellectual developmental disorder, X-linked 30, 300558* |
| *MPP5* | *100%* | *100%* | *No OMIM disease ID* |
| *PAM16* | *85%* | *85%* | *Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320* |
| *PAN2* | *100%* | *100%* | *No OMIM disease ID* |
| *PANK2* | *100%* | *100%* | *HARP syndrome, 607236*<br>*Neurodegeneration with brain iron accumulation 1, 234200* |
| *PANX1* | *100%* | *100%* | *Oocyte maturation defect 7, 618550* |
| *PARN* | *97%* | *96%* | *Dyskeratosis congenita, autosomal recessive 6, 616353*<br>*Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371* |
| *PARP6* | *100%* | *100%* | *No OMIM disease ID* |
| *PAX1* | *100%* | *100%* | *Otofaciocervical syndrome 2, 615560* |
| *PAX6* | *100%* | *100%* | *Optic nerve hypoplasia, 165550*<br>*Cataract with late-onset corneal dystrophy, 106210*<br>*?Coloboma, ocular, 120200*<br>*?Coloboma of optic nerve, 120430*<br>*Aniridia, 106210*<br>*Anterior segment dysgenesis 5, multiple subtypes, 604229*<br>*?Morning glory disc anomaly, 120430*<br>*Foveal hypoplasia 1, 136520*<br>*Keratitis, 148190* |

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| *PAX7* | *100%* | *100%* | *Rhabdomyosarcoma 2, alveolar, 268220*<br>*Myopathy, congenital, progressive, with scoliosis, 618578* |
| *PAX8* | *100%* | *100%* | *Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700* |
| *PBX1* | *100%* | *100%* | *Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641* |
| *PC* | *100%* | *100%* | *Pyruvate carboxylase deficiency, 266150* |
| *PCCA* | *100%* | *100%* | *Propionicacidemia, 606054* |
| *PCCB* | *100%* | *98%* | *Propionicacidemia, 606054* |
| *PCDH12* | *100%* | *100%* | *Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280* |
| *PCDH19* | *100%* | *100%* | *Developmental and epileptic encephalopathy 9, 300088* |
| *PCDHGC4* | *100%* | *100%* | *Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880* |
| *PCGF2* | *100%* | *100%* | *Turnpenny-Fry syndrome, 618371* |
| *PCLO* | *100%* | *100%* | *?Pontocerebellar hypoplasia, type 3, 608027* |
| *PCNT* | *100%* | *100%* | *Microcephalic osteodysplastic primordial dwarfism, type II, 210720* |
| *PCYT2* | *100%* | *100%* | *Spastic paraplegia 82, autosomal recessive, 618770* |
| *PDE2A* | *100%* | *100%* | *Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150* |
| *PDE4D* | *100%* | *100%* | *Acrodysostosis 2, with or without hormone resistance, 614613* |
| *PDGFRB* | *100%* | *100%* | *Premature aging syndrome, Penttinen type, 601812*<br>*Kosaki overgrowth syndrome, 616592*<br>*Myofibromatosis, infantile, 1, 228550*<br>*Basal ganglia calcification, idiopathic, 4, 615007* |
| *PDHA1* | *100%* | *98%* | *Pyruvate dehydrogenase E1-alpha deficiency, 312170* |
| *PDHB* | *100%* | *100%* | *Pyruvate dehydrogenase E1-beta deficiency, 614111* |
| *PDHX* | *100%* | *100%* | *Lacticacidemia due to PDX1 deficiency, 245349* |
| *PDP1* | *100%* | *100%* | *Pyruvate dehydrogenase phosphatase deficiency, 608782* |
| *PDSS1* | *100%* | *100%* | *Coenzyme Q10 deficiency, primary, 2, 614651* |
| *PDSS2* | *100%* | *100%* | *Coenzyme Q10 deficiency, primary, 3, 614652* |
| *PDZD8* | *100%* | *100%* | *Intellectual developmental disorder with autism and dysmorphic facies, 620021* |
| *PEPD* | *100%* | *100%* | *Prolidase deficiency, 170100* |
| *PET100* | *100%* | *100%* | *Mitochondrial complex IV deficiency, nuclear type 12, 619055* |
| *PEX1* | *100%* | *100%* | *Heimler syndrome 1, 234580*<br>*Peroxisome biogenesis disorder 1B (NALD/IRD), 601539*<br>*Peroxisome biogenesis disorder 1A (Zellweger), 214100* |
| *PEX10* | *100%* | *100%* | *Peroxisome biogenesis disorder 6A (Zellweger), 614870*<br>*Peroxisome biogenesis disorder 6B, 614871* |

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| *PEX11B* | *100%* | *100%* | *Peroxisome biogenesis disorder 14B, 614920* |
| *PEX12* | *100%* | *100%* | *Peroxisome biogenesis disorder 3B, 266510*<br>*Peroxisome biogenesis disorder 3A (Zellweger), 614859* |
| *PEX13* | *100%* | *100%* | *Peroxisome biogenesis disorder 11A (Zellweger), 614883*<br>*Peroxisome biogenesis disorder 11B, 614885* |
| *PEX16* | *100%* | *100%* | *Peroxisome biogenesis disorder 8B, 614877*<br>*Peroxisome biogenesis disorder 8A (Zellweger), 614876* |
| *PEX19* | *100%* | *100%* | *Peroxisome biogenesis disorder 12A (Zellweger), 614886* |
| *PEX2* | *100%* | *100%* | *Peroxisome biogenesis disorder 5A (Zellweger), 614866*<br>*Peroxisome biogenesis disorder 5B, 614867* |
| *PEX26* | *100%* | *100%* | *Peroxisome biogenesis disorder 7B, 614873*<br>*Peroxisome biogenesis disorder 7A (Zellweger), 614872* |
| *PEX3* | *100%* | *100%* | *Peroxisome biogenesis disorder 10A (Zellweger), 614882*<br>*?Peroxisome biogenesis disorder 10B, 617370* |
| *PEX5* | *100%* | *100%* | *Peroxisome biogenesis disorder 2B, 202370*<br>*Peroxisome biogenesis disorder 2A (Zellweger), 214110*<br>*Rhizomelic chondrodysplasia punctata, type 5, 616716* |
| *PEX6* | *100%* | *100%* | *Peroxisome biogenesis disorder 4B, 614863*<br>*Peroxisome biogenesis disorder 4A (Zellweger), 614862*<br>*Heimler syndrome 2, 616617* |
| *PEX7* | *91%* | *91%* | *Rhizomelic chondrodysplasia punctata, type 1, 215100*<br>*Peroxisome biogenesis disorder 9B, 614879* |
| *PGAP1* | *100%* | *100%* | *Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802* |
| *PGAP2* | *100%* | *100%* | *Hyperphosphatasia with impaired intellectual development syndrome 3, 614207* |
| *PGAP3* | *100%* | *100%* | *Hyperphosphatasia with impaired intellectual development syndrome 4, 615716* |
| *PGK1* | *100%* | *100%* | *Phosphoglycerate kinase 1 deficiency, 300653* |
| *PGM2L1* | *100%* | *100%* | *No OMIM disease ID* |
| *PGM3* | *100%* | *100%* | *Immunodeficiency 23, 615816* |
| *PHACTR1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 70, 618298* |
| *PHF21A* | *100%* | *100%* | *Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725* |
| *PHF6* | *100%* | *100%* | *Borjeson-Forssman-Lehmann syndrome, 301900* |
| *PHF8* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263* |
| *PHGDH* | *100%* | *100%* | *Neu-Laxova syndrome 1, 256520*<br>*Phosphoglycerate dehydrogenase deficiency, 601815* |
| *PHIP* | *100%* | *100%* | *Chung-Jansen syndrome, 617991* |

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| *PI4KA* | *100%* | *100%* | *Spastic paraplegia 84, autosomal recessive, 619621*<br>*Gastrointestinal defects and immunodeficiency syndrome 2, 619708*<br>*Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531* |
| *PIBF1* | *100%* | *100%* | *Joubert syndrome 33, 617767* |
| *PIDD1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827* |
| *PIGA* | *100%* | *100%* | *Paroxysmal nocturnal hemoglobinuria, somatic, 300818*<br>*Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868*<br>*Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072* |
| *PIGB* | *100%* | *100%* | *Developmental and epileptic encephalopathy 80, 618580* |
| *PIGC* | *100%* | *100%* | *Glycosylphosphatidylinositol biosynthesis defect 16, 617816* |
| *PIGF* | *100%* | *100%* | *Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356* |
| *PIGG* | *100%* | *100%* | *Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917* |
| *PIGH* | *81%* | *75%* | *Glycosylphosphatidylinositol biosynthesis defect 17, 618010* |
| *PIGK* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879* |
| *PIGL* | *100%* | *100%* | *CHIME syndrome, 280000* |
| *PIGN* | *100%* | *100%* | *Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080* |
| *PIGO* | *100%* | *100%* | *Hyperphosphatasia with impaired intellectual development syndrome 2, 614749* |
| *PIGP* | *100%* | *100%* | *Developmental and epileptic encephalopathy 55, 617599* |
| *PIGQ* | *100%* | *100%* | *Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548* |
| *PIGS* | *100%* | *100%* | *Developmental and epileptic encephalopathy 95, 618143* |
| *PIGT* | *100%* | *100%* | *?Paroxysmal nocturnal hemoglobinuria 2, 615399*<br>*Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398* |
| *PIGU* | *100%* | *100%* | *Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590* |
| *PIGV* | *100%* | *100%* | *Hyperphosphatasia with impaired intellectual development syndrome 1, 239300* |
| *PIGW* | *100%* | *100%* | *Glycosylphosphatidylinositol biosynthesis defect 11, 616025* |
| *PIGY* | *100%* | *100%* | *Hyperphosphatasia with impaired intellectual development syndrome 6, 616809* |
| *PIK3CA* | *100%* | *100%* | *CLOVE syndrome, somatic, 612918*<br>*Hepatocellular carcinoma, somatic, 114550*<br>*Breast cancer, somatic, 114480*<br>*Cerebral cavernous malformations 4, somatic, 619538*<br>*Ovarian cancer, somatic, 167000*<br>*Colorectal cancer, somatic, 114500*<br>*Macrodactyly, somatic, 155500*<br>*CLAPO syndrome, somatic, 613089*<br>*Keratosis, seborrheic, somatic, 182000*<br>*Nevus, epidermal, somatic, 162900* |

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| | | | *Gastric cancer, somatic, 613659*<br>*Nonsmall cell lung cancer, somatic, 211980*<br>*Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501*<br>*Cowden syndrome 5, 615108* |
| *PIK3R2* | *100%* | *100%* | *Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387* |
| *PISD* | *100%* | *100%* | *Liberfarb syndrome, 618889* |
| *PITRM1* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 30, 619405* |
| *PJA1* | *100%* | *100%* | *No OMIM disease ID* |
| *PLA2G6* | *100%* | *100%* | *Parkinson disease 14, autosomal recessive, 612953*<br>*Neurodegeneration with brain iron accumulation 2B, 610217*<br>*Infantile neuroaxonal dystrophy 1, 256600* |
| *PLAA* | *100%* | *100%* | *Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527* |
| *PLCB1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 12, 613722* |
| *PLK1* | *100%* | *100%* | *No OMIM disease ID* |
| *PLK4* | *100%* | *100%* | *Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171* |
| *PLP1* | *100%* | *99%* | *Pelizaeus-Merzbacher disease, 312080*<br>*Spastic paraplegia 2, X-linked, 312920* |
| *PLPBP* | *100%* | *100%* | *Epilepsy, early-onset, vitamin B6-dependent, 617290* |
| *PLXNA1* | *100%* | *100%* | *Dworschak-Punetha neurodevelopmental syndrome, 619955* |
| *PLXNA2* | *100%* | *100%* | *No OMIM disease ID* |
| *PLXND1* | *100%* | *100%* | *No OMIM disease ID* |
| *PMM2* | *100%* | *100%* | *Congenital disorder of glycosylation, type Ia, 212065* |
| *PMPCA* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 2, 213200* |
| *PMPCB* | *100%* | *100%* | *Multiple mitochondrial dysfunctions syndrome 6, 617954* |
| *PNKP* | *100%* | *100%* | *?Charcot-Marie-Tooth disease, type 2B2, 605589*<br>*Ataxia-oculomotor apraxia 4, 616267*<br>*Microcephaly, seizures, and developmental delay, 613402* |
| *PNP* | *100%* | *100%* | *Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179* |
| *PNPLA6* | *100%* | *100%* | *Spastic paraplegia 39, autosomal recessive, 612020*<br>*Oliver-McFarlane syndrome, 275400*<br>*?Laurence-Moon syndrome, 245800*<br>*Boucher-Neuhauser syndrome, 215470* |
| *POGZ* | *100%* | *100%* | *White-Sutton syndrome, 616364* |
| *POLA1* | *100%* | *99%* | *Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220*<br>*Van Esch-O'Driscoll syndrome, 301030* |

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| *POLG* | *100%* | *100%* | *Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459*<br>*Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662*<br>*Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700*<br>*Progressive external ophthalmoplegia, autosomal dominant 1, 157640*<br>*Progressive external ophthalmoplegia, autosomal recessive 1, 258450* |
| *POLR1C* | *83%* | *83%* | *Leukodystrophy, hypomyelinating, 11, 616494*<br>*Treacher Collins syndrome 3, 248390* |
| *POLR2A* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603* |
| *POLR3A* | *100%* | *100%* | *Wiedemann-Rautenstrauch syndrome, 264090*<br>*Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694* |
| *POLR3B* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381*<br>*Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742* |
| *POLRMT* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 55, 619743* |
| *POMGNT1* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157*<br>*Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151*<br>*Retinitis pigmentosa 76, 617123*<br>*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280* |
| *POMGNT2* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830*<br>*Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135* |
| *POMK* | *100%* | *100%* | *?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094*<br>*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249* |
| *POMT1* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670*<br>*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308*<br>*Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155* |
| *POMT2* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158*<br>*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150*<br>*Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156* |
| *PORCN* | *100%* | *100%* | *Focal dermal hypoplasia, 305600* |
| *POU1F1* | *100%* | *100%* | *Pituitary hormone deficiency, combined or isolated, 1, 613038* |
| *POU3F3* | *100%* | *98%* | *Snijders Blok-Fisher syndrome, 618604* |
| *PPIL1* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 14, 619301* |
| *PPM1D* | *100%* | *100%* | *Breast cancer, somatic, 114480*<br>*Jansen de Vries syndrome, 617450* |
| *PPP1CB* | *100%* | *100%* | *Noonan syndrome-like disorder with loose anagen hair 2, 617506* |

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| *PPP1R12A* | *100%* | *99%* | *Genitourinary and/or/brain malformation syndrome, 618820* |
| *PPP1R15B* | *100%* | *100%* | *Microcephaly, short stature, and impaired glucose metabolism 2, 616817* |
| *PPP1R21* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383* |
| *PPP2CA* | *100%* | *100%* | *Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354* |
| *PPP2R1A* | *94%* | *94%* | *Intellectual developmental disorder, autosomal dominant 36, 616362* |
| *PPP2R3C* | *100%* | *100%* | *Spermatogenic failure 36, 618420*<br>*Myoectodermal gonadal dysgenesis syndrome, 618419* |
| *PPP2R5B* | *100%* | *100%* | *No OMIM disease ID* |
| *PPP2R5C* | *100%* | *100%* | *No OMIM disease ID* |
| *PPP2R5D* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 35, 616355* |
| *PPP3CA* | *100%* | *100%* | *Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265*<br>*Developmental and epileptic encephalopathy 91, 617711* |
| *PPT1* | *90%* | *90%* | *Ceroid lipofuscinosis, neuronal, 1, 256730* |
| *PQBP1* | *100%* | *100%* | *Renpenning syndrome, 309500* |
| *PRDM13* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 17, 619909*<br>*Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761* |
| *PRDM15* | *100%* | *100%* | *No OMIM disease ID* |
| *PRICKLE2* | *100%* | *100%* | *No OMIM disease ID* |
| *PRKACB* | *100%* | *99%* | *Cardioacrofacial dysplasia 2, 619143* |
| *PRKAR1A* | *100%* | *100%* | *Pigmented nodular adrenocortical disease, primary, 1, 610489*<br>*Acrodysostosis 1, with or without hormone resistance, 101800*<br>*Carney complex, type 1, 160980*<br>*Myxoma, intracardiac, 255960*<br>*Adrenocortical tumor, somatic,* |
| *PRKAR1B* | *100%* | *100%* | *Marbach-Schaaf neurodevelopmental syndrome, 619680* |
| *PRMT7* | *100%* | *100%* | *Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157* |
| *PRODH* | *100%* | *100%* | *Hyperprolinemia, type I, 239500* |
| *PRPF8* | *100%* | *100%* | *Retinitis pigmentosa 13, 600059* |
| *PRPS1* | *100%* | *100%* | *Arts syndrome, 301835*<br>*Phosphoribosylpyrophosphate synthetase superactivity, 300661*<br>*Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070*<br>*Deafness, X-linked 1, 304500*<br>*Gout, PRPS-related, 300661* |
| *PRR12* | *100%* | *100%* | *Neuroocular syndrome, 619539* |
| *PRSS12* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 1, 249500* |

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| *PRUNE1* | *93%* | *93%* | *Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481* |
| *PSAP* | *100%* | *100%* | *Combined SAP deficiency, 611721*<br>*Krabbe disease, atypical, 611722*<br>*Metachromatic leukodystrophy due to SAP-b deficiency, 249900*<br>*Gaucher disease, atypical, 610539* |
| *PSAT1* | *100%* | *100%* | *Neu-Laxova syndrome 2, 616038*<br>*?Phosphoserine aminotransferase deficiency, 610992* |
| *PSMC5* | *100%* | *100%* | *No OMIM disease ID* |
| *PSMD12* | *100%* | *100%* | *Stankiewicz-Isidor syndrome, 617516* |
| *PSPH* | *100%* | *100%* | *Phosphoserine phosphatase deficiency, 614023* |
| *PTCH1* | *100%* | *100%* | *Basal cell carcinoma, somatic, 605462*<br>*Holoprosencephaly 7, 610828*<br>*Basal cell nevus syndrome, 109400* |
| *PTCHD1* | *100%* | *100%* | *No OMIM disease ID* |
| *PTDSS1* | *100%* | *100%* | *Lenz-Majewski hyperostotic dwarfism, 151050* |
| *PTEN* | *100%* | *100%* | *Cowden syndrome 1, 158350*<br>*Lhermitte-Duclos disease, 158350*<br>*Prostate cancer, somatic, 176807*<br>*Macrocephaly/autism syndrome, 605309* |
| *PTF1A* | *100%* | *100%* | *Pancreatic and cerebellar agenesis, 609069*<br>*Pancreatic agenesis 2, 615935* |
| *PTPN11* | *100%* | *100%* | *Noonan syndrome 1, 163950*<br>*LEOPARD syndrome 1, 151100*<br>*Metachondromatosis, 156250*<br>*Leukemia, juvenile myelomonocytic, somatic, 607785* |
| *PTPN23* | *100%* | *100%* | *Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890* |
| *PTRH2* | *100%* | *100%* | *Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263* |
| *PTRHD1* | *100%* | *100%* | *No OMIM disease ID* |
| *PTS* | *100%* | *100%* | *Hyperphenylalaninemia, BH4-deficient, A, 261640* |
| *PUF60* | *100%* | *100%* | *Verheij syndrome, 615583* |
| *PUM1* | *100%* | *100%* | *Spinocerebellar ataxia 47, 617931* |
| *PURA* | *100%* | *100%* | *Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158* |
| *PUS1* | *100%* | *100%* | *Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462* |
| *PUS3* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly and gray sclerae, 617051* |
| *PUS7* | *100%* | *100%* | *Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342* |

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| *PYCR1* | *100%* | *100%* | *Cutis laxa, autosomal recessive, type IIIB, 614438*<br>*Cutis laxa, autosomal recessive, type IIB, 612940* |
| *PYCR2* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 10, 616420* |
| *QARS1* | *100%* | *100%* | *Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760* |
| *QDPR* | *100%* | *100%* | *Hyperphenylalaninemia, BH4-deficient, C, 261630* |
| *QRICH1* | *100%* | *100%* | *Ververi-Brady syndrome, 617982* |
| *RAB11B* | *100%* | *100%* | *Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807* |
| *RAB14* | *100%* | *100%* | *No OMIM disease ID* |
| *RAB18* | *100%* | *100%* | *Warburg micro syndrome 3, 614222* |
| *RAB23* | *100%* | *100%* | *Carpenter syndrome, 201000* |
| *RAB27A* | *100%* | *100%* | *Griscelli syndrome, type 2, 607624* |
| *RAB39B* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 72, 300271*<br>*Waisman syndrome, 311510* |
| *RAB3GAP1* | *99%* | *99%* | *Martsolf syndrome 2, 619420*<br>*Warburg micro syndrome 1, 600118* |
| *RAB3GAP2* | *100%* | *100%* | *Martsolf syndrome 1, 212720*<br>*Warburg micro syndrome 2, 614225* |
| *RAC1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 48, 617751* |
| *RAC3* | *100%* | *100%* | *Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577* |
| *RAD21* | *100%* | *100%* | *Cornelia de Lange syndrome 4, 614701*<br>*?Mungan syndrome, 611376* |
| *RAF1* | *100%* | *100%* | *Cardiomyopathy, dilated, 1NN, 615916*<br>*Noonan syndrome 5, 611553*<br>*LEOPARD syndrome 2, 611554* |
| *RAI1* | *100%* | *100%* | *Smith-Magenis syndrome, 182290* |
| *RALA* | *100%* | *100%* | *Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311* |
| *RALGAPA1* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797* |
| *RARB* | *100%* | *100%* | *Microphthalmia, syndromic 12, 615524* |
| *RARS1* | *94%* | *94%* | *Leukodystrophy, hypomyelinating, 9, 616140* |
| *RARS2* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 6, 611523* |
| *RBBP8* | *100%* | *100%* | *Seckel syndrome 2, 606744*<br>*Jawad syndrome, 251255*<br>*Pancreatic carcinoma, somatic,* |
| *RBFOX1* | *100%* | *100%* | *No OMIM disease ID* |
| *RBM10* | *100%* | *100%* | *TARP syndrome, 311900* |
| *RBM28* | *100%* | *100%* | *?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079* |

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| *RBPJ* | *100%* | *100%* | *Adams-Oliver syndrome 3, 614814* |
| *RCBTB1* | *100%* | *100%* | *Retinal dystrophy with or without extraocular anomalies, 617175* |
| *RECQL4* | *100%* | *100%* | *Baller-Gerold syndrome, 218600*<br>*Rothmund-Thomson syndrome, type 2, 268400*<br>*RAPADILINO syndrome, 266280* |
| *RELN* | *100%* | *100%* | *Lissencephaly 2 (Norman-Roberts type), 257320* |
| *RERE* | *100%* | *100%* | *Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975* |
| *REV3L* | *98%* | *98%* | *No OMIM disease ID* |
| *RFT1* | *100%* | *100%* | *Congenital disorder of glycosylation, type In, 612015* |
| *RFX3* | *100%* | *99%* | *No OMIM disease ID* |
| *RFX4* | *100%* | *100%* | *No OMIM disease ID* |
| *RFX7* | *100%* | *100%* | *No OMIM disease ID* |
| *RHEB* | *100%* | *100%* | *No OMIM disease ID* |
| *RHOBTB2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 64, 618004* |
| *RIC1* | *100%* | *100%* | *CATIFA syndrome, 618761* |
| *RIMS2* | *100%* | *100%* | *Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970* |
| *RIT1* | *100%* | *100%* | *Noonan syndrome 8, 615355* |
| *RLIM* | *100%* | *100%* | *Tonne-Kalscheuer syndrome, 300978* |
| *RMND1* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 11, 614922* |
| *RMRP* | *NC* | *NC* | *Anauxetic dysplasia 1, 607095*<br>*Metaphyseal dysplasia without hypotrichosis, 250460*<br>*Cartilage-hair hypoplasia, 250250* |
| *RNASEH2A* | *100%* | *100%* | *Aicardi-Goutieres syndrome 4, 610333* |
| *RNASEH2B* | *91%* | *91%* | *Aicardi-Goutieres syndrome 2, 610181* |
| *RNASEH2C* | *100%* | *100%* | *Aicardi-Goutieres syndrome 3, 610329* |
| *RNASET2* | *100%* | *100%* | *Leukoencephalopathy, cystic, without megalencephaly, 612951* |
| *RNF113A* | *100%* | *100%* | *Trichothiodystrophy 5, nonphotosensitive, 300953* |
| *RNF125* | *100%* | *100%* | *Tenorio syndrome, 616260* |
| *RNF13* | *100%* | *100%* | *Developmental and epileptic encephalopathy 73, 618379* |
| *RNF2* | *100%* | *100%* | *Luo-Schoch-Yamamoto syndrome, 619460* |
| *RNF220* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688* |
| *RNPC3* | *100%* | *100%* | *Pituitary hormone deficiency, combined or isolated, 7, 618160* |
| *RNU4ATAC* | *NC* | *NC* | *Roifman syndrome, 616651*<br>*Lowry-Wood syndrome, 226960*<br>*Microcephalic osteodysplastic primordial dwarfism, type I, 210710* |

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| *ROGDI* | *100%* | *100%* | *Kohlschutter-Tonz syndrome, 226750* |
| *ROR2* | *100%* | *100%* | *Brachydactyly, type B1, 113000*<br>*Robinow syndrome, autosomal recessive, 268310* |
| *RORA* | *100%* | *100%* | *Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060* |
| *RORB* | *100%* | *100%* | *No OMIM disease ID* |
| *RPGRIP1L* | *100%* | *100%* | *Joubert syndrome 7, 611560*<br>*Meckel syndrome 5, 611561*<br>*?COACH syndrome 3, 619113* |
| *RPIA* | *100%* | *100%* | *Ribose 5-phosphate isomerase deficiency, 608611* |
| *RPL10* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic 35, 300998* |
| *RPS19* | *100%* | *100%* | *Diamond-Blackfan anemia 1, 105650* |
| *RPS6KA3* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 19, 300844*<br>*Coffin-Lowry syndrome, 303600* |
| *RRM2B* | *100%* | *100%* | *Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075*<br>*Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075*<br>*Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315*<br>*Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077* |
| *RRP7A* | *100%* | *100%* | *?Microcephaly 28, primary, autosomal recessive, 619453* |
| *RSPRY1* | *100%* | *100%* | *Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723* |
| *RSRC1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 70, 618402* |
| *RTEL1* | *100%* | *100%* | *Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373*<br>*Dyskeratosis congenita, autosomal dominant 4, 615190*<br>*Dyskeratosis congenita, autosomal recessive 5, 615190* |
| *RTN4IP1* | *100%* | *100%* | *Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732* |
| *RTTN* | *100%* | *100%* | *Microcephaly, short stature, and polymicrogyria with seizures, 614833* |
| *RUBCN* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 15, 615705* |
| *RUSC2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 61, 617773* |
| *RXYLT1* | *100%* | *100%* | *Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041* |
| *SALL1* | *100%* | *100%* | *Townes-Brocks syndrome 1, 107480*<br>*Townes-Brocks branchiootorenal-like syndrome, 107480* |
| *SAMD9* | *100%* | *100%* | *Tumoral calcinosis, familial, normophosphatemic, 610455*<br>*Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041*<br>*MIRAGE syndrome, 617053* |
| *SAMHD1* | *100%* | *100%* | *?Chilblain lupus 2, 614415*<br>*Aicardi-Goutieres syndrome 5, 612952* |
| *SARS1* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709* |

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| *SATB1* | *100%* | *100%* | *Kohlschutter-Tonz syndrome-like, 619229*<br>*Developmental delay with dysmorphic facies and dental anomalies, 619228* |
| *SATB2* | *100%* | *100%* | *Glass syndrome, 612313* |
| *SBDS* | *100%* | *100%* | *Shwachman-Diamond syndrome 1, 260400* |
| *SC5D* | *100%* | *100%* | *Lathosterolosis, 607330* |
| *SCAF4* | *100%* | *100%* | *No OMIM disease ID* |
| *SCAMP5* | *100%* | *100%* | *No OMIM disease ID* |
| *SCAPER* | *100%* | *100%* | *Intellectual developmental disorder and retinitis pigmentosa, 618195* |
| *SCN1A* | *100%* | *100%* | *Developmental and epileptic encephalopathy 6B, non-Dravet, 619317*<br>*Migraine, familial hemiplegic, 3, 609634*<br>*Dravet syndrome, 607208*<br>*Febrile seizures, familial, 3A, 604403*<br>*Generalized epilepsy with febrile seizures plus, type 2, 604403* |
| *SCN1B* | *100%* | *100%* | *Generalized epilepsy with febrile seizures plus, type 1, 604233*<br>*Developmental and epileptic encephalopathy 52, 617350*<br>*Cardiac conduction defect, nonspecific, 612838*<br>*Atrial fibrillation, familial, 13, 615377*<br>*Brugada syndrome 5, 612838* |
| *SCN2A* | *100%* | *100%* | *Seizures, benign familial infantile, 3, 607745*<br>*Developmental and epileptic encephalopathy 11, 613721*<br>*Episodic ataxia, type 9, 618924* |
| *SCN3A* | *100%* | *100%* | *Epilepsy, familial focal, with variable foci 4, 617935*<br>*Developmental and epileptic encephalopathy 62, 617938* |
| *SCN8A* | *100%* | *100%* | *?Myoclonus, familial, 2, 618364*<br>*Seizures, benign familial infantile, 5, 617080*<br>*Cognitive impairment with or without cerebellar ataxia, 614306*<br>*Developmental and epileptic encephalopathy 13, 614558* |
| *SCO1* | *100%* | *100%* | *Mitochondrial complex IV deficiency, nuclear type 4, 619048* |
| *SCO2* | *100%* | *100%* | *Myopia 6, 608908*<br>*Mitochondrial complex IV deficiency, nuclear type 2, 604377* |
| *SCUBE3* | *100%* | *100%* | *Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184* |
| *SCYL1* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 21, 616719* |
| *SDCCAG8* | *100%* | *100%* | *Senior-Loken syndrome 7, 613615*<br>*Bardet-Biedl syndrome 16, 615993* |
| *SDHA* | *100%* | *100%* | *Cardiomyopathy, dilated, 1GG, 613642*<br>*Mitochondrial complex II deficiency, nuclear type 1, 252011* |

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| | | | *Neurodegeneration with ataxia and late-onset optic atrophy, 619259*<br>*Paragangliomas 5, 614165* |
| *SEC31A* | *100%* | *100%* | *?Halperin-Birk syndrome, 618651* |
| *SEMA3E* | *100%* | *100%* | *No OMIM disease ID* |
| *SEPSECS* | *100%* | *100%* | *Pontocerebellar hypoplasia type 2D, 613811* |
| *SERAC1* | *100%* | *100%* | *3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739* |
| *SET* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 58, 618106* |
| *SETBP1* | *100%* | *100%* | *Schinzel-Giedion midface retraction syndrome, 269150*<br>*Intellectual developmental disorder, autosomal dominant 29, 616078* |
| *SETD1A* | *100%* | *100%* | *Epilepsy, early-onset, with or without developmental delay, 618832*<br>*Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056* |
| *SETD1B* | *100%* | *100%* | *Intellectual developmental disorder with seizures and language delay, 619000* |
| *SETD2* | *100%* | *100%* | *Luscan-Lumish syndrome, 616831* |
| *SETD5* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 23, 615761* |
| *SFXN4* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 18, 615578* |
| *SGPL1* | *100%* | *100%* | *Nephrotic syndrome, type 14, 617575* |
| *SGSH* | *100%* | *100%* | *Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900* |
| *SHANK1* | *100%* | *100%* | *No OMIM disease ID* |
| *SHANK2* | *100%* | *100%* | *No OMIM disease ID* |
| *SHANK3* | *100%* | *99%* | *Phelan-McDermid syndrome, 606232* |
| *SHH* | *100%* | *100%* | *Microphthalmia with coloboma 5, 611638*<br>*Schizencephaly, 269160*<br>*Single median maxillary central incisor, 147250*<br>*Holoprosencephaly 3, 142945* |
| *SHMT2* | *100%* | *100%* | *Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121* |
| *SHOC2* | *100%* | *100%* | *Noonan syndrome-like with loose anagen hair 1, 607721* |
| *SHROOM4* | *100%* | *100%* | *No OMIM disease ID* |
| *SIAH1* | *100%* | *100%* | *Buratti-Harel syndrome, 619314* |
| *SIK1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 30, 616341* |
| *SIL1* | *100%* | *100%* | *Marinesco-Sjogren syndrome, 248800* |
| *SIN3A* | *100%* | *100%* | *Witteveen-Kolk syndrome, 613406* |
| *SIN3B* | *100%* | *100%* | *No OMIM disease ID* |
| *SIX3* | *100%* | *100%* | *Schizencephaly, 269160*<br>*Holoprosencephaly 2, 157170* |
| *SKI* | *100%* | *100%* | *Shprintzen-Goldberg syndrome, 182212* |

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|---|---|---|---|
| *TTC37* | *100%* | *100%* | *Trichohepatoenteric syndrome 1, 222470* |
| *SLC12A2* | *100%* | *100%* | *Kilquist syndrome, 619080*<br>*Delpire-McNeill syndrome, 619083*<br>*Deafness, autosomal dominant 78, 619081* |
| *SLC12A5* | *100%* | *100%* | *Developmental and epileptic encephalopathy 34, 616645* |
| *SLC12A6* | *100%* | *100%* | *Agenesis of the corpus callosum with peripheral neuropathy, 218000*<br>*Charcot-Marie-Tooth disease, axonal, type 2II, 620068* |
| *SLC13A5* | *100%* | *100%* | *Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905* |
| *SLC16A2* | *100%* | *100%* | *Allan-Herndon-Dudley syndrome, 300523* |
| *SLC17A5* | *100%* | *100%* | *Salla disease, 604369*<br>*Sialic acid storage disorder, infantile, 269920* |
| *SLC19A3* | *100%* | *98%* | *Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483* |
| *SLC1A1* | *100%* | *100%* | *Dicarboxylic aminoaciduria, 222730* |
| *SLC1A2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 41, 617105* |
| *SLC1A4* | *100%* | *100%* | *Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657* |
| *SLC25A1* | *100%* | *100%* | *Combined D-2- and L-2-hydroxyglutaric aciduria, 615182*<br>*Myasthenic syndrome, congenital, 23, presynaptic, 618197* |
| *SLC25A12* | *100%* | *100%* | *Developmental and epileptic encephalopathy 39, 612949* |
| *SLC25A15* | *100%* | *100%* | *Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970* |
| *SLC25A22* | *100%* | *100%* | *Developmental and epileptic encephalopathy 3, 609304* |
| *SLC25A24* | *100%* | *100%* | *Fontaine progeroid syndrome, 612289* |
| *SLC25A42* | *100%* | *100%* | *Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416* |
| *SLC2A1* | *100%* | *100%* | *Dystonia 9, 601042*<br>*GLUT1 deficiency syndrome 1, infantile onset, severe, 606777*<br>*Stomatin-deficient cryohydrocytosis with neurologic defects, 608885*<br>*GLUT1 deficiency syndrome 2, childhood onset, 612126* |
| *SLC33A1* | *100%* | *100%* | *Spastic paraplegia 42, autosomal dominant, 612539*<br>*Congenital cataracts, hearing loss, and neurodegeneration, 614482* |
| *SLC35A1* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIf, 603585* |
| *SLC35A2* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIm, 300896* |
| *SLC35A3* | *98%* | *93%* | *Arthrogryposis, impaired intellectual development, and seizures, 615553* |
| *SLC35B2* | *100%* | *100%* | *No OMIM disease ID* |
| *SLC35C1* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIc, 266265* |
| *SLC38A3* | *100%* | *100%* | *Developmental and epileptic encephalopathy 102, 619881* |
| *SLC39A14* | *94%* | *94%* | *?Hyperostosis cranalis interna, 144755*<br>*Hypermanganesemia with dystonia 2, 617013* |

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| *SLC39A8* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIn, 616721* |
| *SLC45A1* | *100%* | *100%* | *Intellectual developmental disorder with neuropsychiatric features, 617532* |
| *SLC46A1* | *100%* | *100%* | *Folate malabsorption, hereditary, 229050* |
| *SLC4A4* | *100%* | *100%* | *Renal tubular acidosis, proximal, with ocular abnormalities, 604278* |
| *SLC5A6* | *100%* | *100%* | *Sodium-dependent multivitamin transporter deficiency, 618973*<br>*Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903* |
| *SLC6A1* | *100%* | *100%* | *Myoclonic-atonic epilepsy, 616421* |
| *SLC6A17* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 48, 616269* |
| *SLC6A19* | *100%* | *100%* | *Iminoglycinuria, digenic, 242600*<br>*Hartnup disorder, 234500*<br>*Hyperglycinuria, 138500* |
| *SLC6A3* | *100%* | *100%* | *Parkinsonism-dystonia, infantile, 1, 613135* |
| *SLC6A8* | *100%* | *100%* | *Cerebral creatine deficiency syndrome 1, 300352* |
| *SLC6A9* | *100%* | *100%* | *Glycine encephalopathy with normal serum glycine, 617301* |
| *SLC7A7* | *100%* | *100%* | *Lysinuric protein intolerance, 222700* |
| *SLC9A6* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243* |
| *SLC9A7* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 108, 301024* |
| *SMAD4* | *100%* | *100%* | *Pancreatic cancer, somatic, 260350*<br>*Myhre syndrome, 139210*<br>*Polyposis, juvenile intestinal, 174900*<br>*Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050* |
| *SMARCA1* | *100%* | *100%* | *No OMIM disease ID* |
| *SMARCA2* | *100%* | *100%* | *Nicolaides-Baraitser syndrome, 601358*<br>*Blepharophimosis-impaired intellectual development syndrome, 619293* |
| *SMARCA4* | *100%* | *100%* | *Coffin-Siris syndrome 4, 614609* |
| *SMARCA5* | *100%* | *100%* | *No OMIM disease ID* |
| *SMARCB1* | *100%* | *100%* | *Rhabdoid tumors, somatic, 609322*<br>*Coffin-Siris syndrome 3, 614608* |
| *SMARCC2* | *100%* | *100%* | *Coffin-Siris syndrome 8, 618362* |
| *SMARCD1* | *100%* | *100%* | *Coffin-Siris syndrome 11, 618779* |
| *SMARCE1* | *100%* | *100%* | *Coffin-Siris syndrome 5, 616938* |
| *SMC1A* | *100%* | *100%* | *Cornelia de Lange syndrome 2, 300590*<br>*Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044* |
| *SMC3* | *100%* | *100%* | *Cornelia de Lange syndrome 3, 610759* |
| *SMG8* | *100%* | *100%* | *Alzahrani-Kuwahara syndrome, 619268* |

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| *SMG9* | *100%* | *100%* | *Heart and brain malformation syndrome, 616920*<br>*Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995* |
| *SMOC1* | *100%* | *100%* | *Microphthalmia with limb anomalies, 206920* |
| *SMPD1* | *100%* | *100%* | *Niemann-Pick disease, type B, 607616*<br>*Niemann-Pick disease, type A, 257200* |
| *SMPD4* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622* |
| *SMS* | *100%* | *99%* | *Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583* |
| *SNAP25* | *100%* | *100%* | *?Myasthenic syndrome, congenital, 18, 616330* |
| *SNAP29* | *100%* | *100%* | *Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528* |
| *SNIP1* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501* |
| *SNORD118* | *NC* | *NC* | *Leukoencephalopathy, brain calcifications, and cysts, 614561* |
| *SNRPB* | *100%* | *100%* | *Cerebrocostomandibular syndrome, 117650* |
| *SNRPN* | *100%* | *100%* | *No OMIM disease ID* |
| *SNX14* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 20, 616354* |
| *SNX27* | *100%* | *100%* | *No OMIM disease ID* |
| *SOBP* | *100%* | *100%* | *Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671* |
| *SON* | *100%* | *100%* | *ZTTK syndrome, 617140* |
| *SOS1* | *100%* | *100%* | *Noonan syndrome 4, 610733*<br>*?Fibromatosis, gingival, 1, 135300* |
| *SOS2* | *100%* | *100%* | *Noonan syndrome 9, 616559* |
| *SOX10* | *100%* | *100%* | *Waardenburg syndrome, type 4C, 613266*<br>*PCWH syndrome, 609136*<br>*Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584* |
| *SOX11* | *100%* | *100%* | *Coffin-Siris syndrome 9, 615866* |
| *SOX2* | *100%* | *100%* | *Optic nerve hypoplasia and abnormalities of the central nervous system, 206900*<br>*Microphthalmia, syndromic 3, 206900* |
| *SOX3* | *100%* | *100%* | *Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123*<br>*Panhypopituitarism, X-linked, 312000* |
| *SOX4* | *100%* | *100%* | *Coffin-Siris syndrome 10, 618506* |
| *SOX5* | *100%* | *100%* | *Lamb-Shaffer syndrome, 616803* |
| *SOX6* | *100%* | *99%* | *Tolchin-Le Caignec syndrome, 618971* |
| *SPART* | *100%* | *100%* | *Troyer syndrome, 275900* |
| *SPAST* | *100%* | *100%* | *Spastic paraplegia 4, autosomal dominant, 182601* |
| *SPATA5* | *100%* | *100%* | *Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577* |

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| *SPATA5L1* | *100%* | *100%* | *Deafness, autosomal recessive 119, 619615*<br>*Neurodevelopmental disorder with hearing loss and spasticity, 619616* |
| *SPECC1L* | *100%* | *100%* | *Teebi hypertelorism syndrome 1, 145420*<br>*?Facial clefting, oblique, 1, 600251* |
| *SPEN* | *100%* | *100%* | *Radio-Tartaglia syndrome, 619312* |
| *SPG11* | *100%* | *100%* | *Amyotrophic lateral sclerosis 5, juvenile, 602099*<br>*Charcot-Marie-Tooth disease, axonal, type 2X, 616668*<br>*Spastic paraplegia 11, autosomal recessive, 604360* |
| *SPOCK1* | *100%* | *100%* | *No OMIM disease ID* |
| *SPOP* | *100%* | *100%* | *Nabais Sa-de Vries syndrome, type 1, 618828*<br>*Nabais Sa-de Vries syndrome, type 2, 618829* |
| *SPR* | *100%* | *100%* | *Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716* |
| *SPRED1* | *100%* | *100%* | *Legius syndrome, 611431* |
| *SPRED2* | *100%* | *100%* | *Noonan syndrome 14, 619745* |
| *SPTAN1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 5, 613477* |
| *SPTBN1* | *100%* | *100%* | *Developmental delay, impaired speech, and behavioral abnormalities, 619475* |
| *SPTBN2* | *100%* | *100%* | *Spinocerebellar ataxia 5, 600224*<br>*Spinocerebellar ataxia, autosomal recessive 14, 615386* |
| *SPTBN4* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519* |
| *SRCAP* | *100%* | *100%* | *Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595*<br>*Floating-Harbor syndrome, 136140* |
| *SRD5A3* | *100%* | *100%* | *Kahrizi syndrome, 612713*<br>*Congenital disorder of glycosylation, type Iq, 612379* |
| *SRP54* | *100%* | *100%* | *Neutropenia, severe congenital, 8, autosomal dominant, 618752* |
| *SRPX2* | *100%* | *100%* | *?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643* |
| *SRRM2* | *100%* | *100%* | *No OMIM disease ID* |
| *SSR4* | *100%* | *100%* | *Congenital disorder of glycosylation, type Iy, 300934* |
| *ST3GAL3* | *97%* | *95%* | *Developmental and epileptic encephalopathy 15, 615006*<br>*Intellectual developmental disorder, autosomal recessive 12, 611090* |
| *ST3GAL5* | *98%* | *98%* | *Salt and pepper developmental regression syndrome, 609056* |
| *STAG1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 47, 617635* |
| *STAG2* | *100%* | *100%* | *Holoprosencephaly 13, X-linked, 301043*<br>*Mullegama-Klein-Martinez syndrome, 301022* |
| *STAMBP* | *100%* | *100%* | *Microcephaly-capillary malformation syndrome, 614261* |
| *CXorf56* | *100%* | *100%* | *?Intellectual developmental disorder, X-linked 107, 301013* |
| *STIL* | *100%* | *100%* | *Microcephaly 7, primary, autosomal recessive, 612703* |

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| *STRA6* | *100%* | *100%* | *Microphthalmia, syndromic 9, 601186*<br>*Microphthalmia, isolated, with coloboma 8, 601186* |
| *STRADA* | *100%* | *100%* | *Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087* |
| *STT3A* | *100%* | *100%* | *Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714*<br>*Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596* |
| *STT3B* | *100%* | *100%* | *Congenital disorder of glycosylation, type Ix, 615597* |
| *STX1B* | *100%* | *100%* | *Generalized epilepsy with febrile seizures plus, type 9, 616172* |
| *STXBP1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 4, 612164* |
| *SUCLA2* | *100%* | *100%* | *Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073* |
| *SUCLG1* | *100%* | *100%* | *Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400* |
| *SUFU* | *100%* | *100%* | *Joubert syndrome 32, 617757*<br>*Basal cell nevus syndrome, 109400* |
| *SUMF1* | *100%* | *100%* | *Multiple sulfatase deficiency, 272200* |
| *SUOX* | *100%* | *100%* | *Sulfite oxidase deficiency, 272300* |
| *SUPT16H* | *100%* | *100%* | *Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480* |
| *SURF1* | *100%* | *100%* | *Charcot-Marie-Tooth disease, type 4K, 616684*<br>*Mitochondrial complex IV deficiency, nuclear type 1, 220110* |
| *SUZ12* | *100%* | *100%* | *Imagawa-Matsumoto syndrome, 618786* |
| *SVBP* | *100%* | *100%* | *Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569* |
| *SYN1* | *100%* | *100%* | *Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491*<br>*Intellectual developmental disorder, X-linked 50, 300115* |
| *SYNCRIP* | *100%* | *100%* | *No OMIM disease ID* |
| *SYNGAP1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 5, 612621* |
| *SYNJ1* | *100%* | *100%* | *Parkinson disease 20, early-onset, 615530*<br>*Developmental and epileptic encephalopathy 53, 617389* |
| *SYP* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 96, 300802* |
| *SYT1* | *100%* | *100%* | *Baker-Gordon syndrome, 618218* |
| *SZT2* | *100%* | *100%* | *Developmental and epileptic encephalopathy 18, 615476* |
| *TACO1* | *100%* | *100%* | *Mitochondrial complex IV deficiency, nuclear type 8, 619052* |
| *TAF1* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic 33, 300966*<br>*Dystonia-Parkinsonism, X-linked, 314250* |
| *TAF13* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 60, 617432* |
| *TAF1C* | *100%* | *100%* | *No OMIM disease ID* |
| *TAF2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 40, 615599* |
| *TAF4* | *90%* | *85%* | *No OMIM disease ID* |

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| *TAF6* | *100%* | *100%* | *Alazami-Yuan syndrome, 617126* |
| *TAF8* | *89%* | *89%* | *Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972* |
| *TANC2* | *100%* | *100%* | *Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906* |
| *TANGO2* | *100%* | *100%* | *Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878* |
| *TAOK1* | *100%* | *100%* | *Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575* |
| *TASP1* | *100%* | *100%* | *Suleiman-El-Hattab syndrome, 618950* |
| *TAT* | *100%* | *100%* | *Tyrosinemia, type II, 276600* |
| *TBC1D20* | *100%* | *100%* | *Warburg micro syndrome 4, 615663* |
| *TBC1D23* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 11, 617695* |
| *TBC1D24* | *100%* | *100%* | *Deafness, autosomal recessive 86, 614617*<br>*Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105*<br>*Myoclonic epilepsy, infantile, familial, 605021*<br>*Deafness, autosomal dominant 65, 616044*<br>*Developmental and epileptic encephalopathy 16, 615338*<br>*DOORS syndrome, 220500* |
| *TBC1D2B* | *100%* | *100%* | *Neurodevelopmental disorder with seizures and gingival overgrowth, 619323* |
| *TBC1D7* | *100%* | *100%* | *Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000* |
| *TBCD* | *100%* | *100%* | *Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193* |
| *TBCE* | *100%* | *100%* | *Kenny-Caffey syndrome, type 1, 244460*<br>*Hypoparathyroidism-retardation-dysmorphism syndrome, 241410*<br>*Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207* |
| *TBCK* | *100%* | *100%* | *Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900* |
| *TBL1XR1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 41, 616944*<br>*Pierpont syndrome, 602342* |
| *TBP* | *100%* | *100%* | *Spinocerebellar ataxia 17, 607136* |
| *TBR1* | *100%* | *100%* | *Intellectual developmental disorder with autism and speech delay, 606053* |
| *TBX1* | *98%* | *96%* | *Tetralogy of Fallot, 187500*<br>*DiGeorge syndrome, 188400*<br>*Conotruncal anomaly face syndrome, 217095*<br>*Velocardiofacial syndrome, 192430* |
| *TCF20* | *100%* | *100%* | *Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430* |
| *TCF4* | *100%* | *100%* | *Pitt-Hopkins syndrome, 610954*<br>*Corneal dystrophy, Fuchs endothelial, 3, 613267* |
| *TCF7L2* | *100%* | *100%* | *No OMIM disease ID* |
| *TCN2* | *100%* | *100%* | *Transcobalamin II deficiency, 275350* |

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| *TCTN2* | *100%* | *100%* | *Joubert syndrome 24, 616654*<br>*?Meckel syndrome 8, 613885* |
| *TCTN3* | *100%* | *100%* | *Joubert syndrome 18, 614815*<br>*Orofaciodigital syndrome IV, 258860* |
| *TDP2* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 23, 616949* |
| *TECPR2* | *100%* | *100%* | *Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031* |
| *TECR* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 14, 614020* |
| *TELO2* | *100%* | *100%* | *You-Hoover-Fong syndrome, 616954* |
| *TENM3* | *100%* | *100%* | *Microphthalmia, syndromic 15, 615145*<br>*?Microphthalmia, isolated, with coloboma 9, 615145* |
| *TET3* | *100%* | *100%* | *Beck-Fahrner syndrome, 618798* |
| *TFAP2A* | *100%* | *100%* | *Branchiooculofacial syndrome, 113620* |
| *TFE3* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066*<br>*Renal cell carcinoma, papillary, 1, 300854* |
| *TGDS* | *100%* | *100%* | *Catel-Manzke syndrome, 616145* |
| *TGFBR1* | *100%* | *100%* | *Loeys-Dietz syndrome 1, 609192* |
| *TGIF1* | *100%* | *100%* | *Holoprosencephaly 4, 142946* |
| *TH* | *100%* | *100%* | *Segawa syndrome, recessive, 605407* |
| *THOC2* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 12, 300957* |
| *THOC6* | *100%* | *100%* | *Beaulieu-Boycott-Innes syndrome, 613680* |
| *THRB* | *100%* | *100%* | *Thyroid hormone resistance, autosomal recessive, 274300*<br>*Thyroid hormone resistance, 188570*<br>*Thyroid hormone resistance, selective pituitary, 145650* |
| *THUMPD1* | *100%* | *100%* | *Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989* |
| *TIAM1* | *100%* | *100%* | *Neurodevelopmental disorder with language delay and seizures, 619908* |
| *TIMM50* | *100%* | *100%* | *3-methylglutaconic aciduria, type IX, 617698* |
| *TIMM8A* | *100%* | *100%* | *Mohr-Tranebjaerg syndrome, 304700* |
| *TINF2* | *100%* | *100%* | *Dyskeratosis congenita, autosomal dominant 3, 613990*<br>*Revesz syndrome, 268130* |
| *TKFC* | *100%* | *100%* | *Triokinase and FMN cyclase deficiency syndrome, 618805* |
| *TKT* | *98%* | *98%* | *Short stature, developmental delay, and congenital heart defects, 617044* |
| *TLK2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 57, 618050* |
| *TMCO1* | *88%* | *88%* | *Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980* |
| *TMEM106B* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 16, 617964* |
| *TMEM165* | *100%* | *100%* | *Congenital disorder of glycosylation, type IIk, 614727* |

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|---|---|---|---|
| *TMEM216* | *100%* | *100%* | *Joubert syndrome 2, 608091*<br>*Meckel syndrome 2, 603194* |
| *TMEM222* | *100%* | *100%* | *Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470* |
| *TMEM231* | *100%* | *100%* | *Joubert syndrome 20, 614970*<br>*Meckel syndrome 11, 615397* |
| *TMEM237* | *100%* | *100%* | *Joubert syndrome 14, 614424* |
| *TMEM240* | *100%* | *100%* | *Spinocerebellar ataxia 21, 607454* |
| *TMEM63A* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 19, transient infantile, 618688* |
| *TMEM63C* | *100%* | *100%* | *Spastic paraplegia 87, autosomal recessive, 619966* |
| *TMEM67* | *100%* | *98%* | *Nephronophthisis 11, 613550*<br>*Joubert syndrome 6, 610688*<br>*Meckel syndrome 3, 607361*<br>*?RHYNS syndrome, 602152*<br>*COACH syndrome 1, 216360* |
| *TMEM70* | *100%* | *100%* | *Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052* |
| *TMEM94* | *100%* | *100%* | *Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316* |
| *TMLHE* | *100%* | *99%* | *No OMIM disease ID* |
| *TMTC3* | *100%* | *100%* | *Lissencephaly 8, 617255* |
| *TMX2* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730* |
| *TNIK* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 54, 617028* |
| *TNPO2* | *100%* | *100%* | *Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556* |
| *TNR* | *100%* | *100%* | *Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653* |
| *TNRC6B* | *100%* | *100%* | *Global developmental delay with speech and behavioral abnormalities, 619243* |
| *TOE1* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 7, 614969* |
| *TOGARAM1* | *100%* | *100%* | *Joubert syndrome 37, 619185* |
| *TOMM70* | *100%* | *100%* | *No OMIM disease ID* |
| *TOR1A* | *91%* | *91%* | *Arthrogryposis multiplex congenita 5, 618947*<br>*Dystonia-1, torsion, 128100* |
| *TP53RK* | *100%* | *100%* | *Galloway-Mowat syndrome 4, 617730* |
| *TP73* | *100%* | *100%* | *Ciliary dyskinesia, primary, 47, and lissencephaly, 619466* |
| *TPI1* | *100%* | *100%* | *Hemolytic anemia due to triosephosphate isomerase deficiency, 615512* |
| *TPO* | *100%* | *100%* | *Thyroid dyshormonogenesis 2A, 274500* |
| *TPP1* | *100%* | *100%* | *Ceroid lipofuscinosis, neuronal, 2, 204500*<br>*Spinocerebellar ataxia, autosomal recessive 7, 609270* |
| *TPP2* | *100%* | *100%* | *Immunodeficiency 78 with autoimmunity and developmental delay, 619220* |

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|---|---|---|---|
| *TPRKB* | *82%* | *81%* | *Galloway-Mowat syndrome 5, 617731* |
| *TRAF7* | *100%* | *100%* | *Cardiac, facial, and digital anomalies with developmental delay, 618164* |
| *TRAIP* | *100%* | *100%* | *Seckel syndrome 9, 616777* |
| *TRAK1* | *100%* | *100%* | *Developmental and epileptic encephalopathy 68, 618201* |
| *TRAPPC11* | *100%* | *100%* | *Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356* |
| *TRAPPC12* | *100%* | *100%* | *Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669* |
| *TRAPPC2L* | *100%* | *100%* | *Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331* |
| *TRAPPC4* | *100%* | *100%* | *Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741* |
| *TRAPPC6B* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862* |
| *TRAPPC9* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 13, 613192* |
| *TREX1* | *100%* | *100%* | *Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315*<br>*Aicardi-Goutieres syndrome 1, dominant and recessive, 225750*<br>*Chilblain lupus, 610448* |
| *TRIM32* | *100%* | *100%* | *?Bardet-Biedl syndrome 11, 615988*<br>*Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110* |
| *TRIM8* | *100%* | *100%* | *Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428* |
| *TRIO* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061*<br>*Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825* |
| *TRIP12* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 49, 617752* |
| *TRIT1* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 35, 617873* |
| *TRMT1* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 68, 618302* |
| *TRMT10A* | *100%* | *100%* | *Microcephaly, short stature, and impaired glucose metabolism 1, 616033* |
| *TRNT1* | *100%* | *100%* | *Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084*<br>*Retinitis pigmentosa and erythrocytic microcytosis, 616959* |
| *TRPM3* | *100%* | *100%* | *No OMIM disease ID* |
| *TRRAP* | *100%* | *100%* | *?Deafness, autosomal dominant 75, 618778*<br>*Developmental delay with or without dysmorphic facies and autism, 618454* |
| *TSC1* | *100%* | *100%* | *Focal cortical dysplasia, type II, somatic, 607341*<br>*Tuberous sclerosis-1, 191100*<br>*Lymphangioleiomyomatosis, 606690* |
| *TSC2* | *100%* | *100%* | *Lymphangioleiomyomatosis, somatic, 606690*<br>*?Focal cortical dysplasia, type II, somatic, 607341*<br>*Tuberous sclerosis-2, 613254* |
| *TSEN15* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 2F, 617026* |

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|---|---|---|---|
| *TSEN2* | *100%* | *100%* | *Pontocerebellar hypoplasia type 2B, 612389* |
| *TSEN54* | *100%* | *100%* | *Pontocerebellar hypoplasia type 2A, 277470*<br>*Pontocerebellar hypoplasia type 4, 225753*<br>*?Pontocerebellar hypoplasia type 5, 610204* |
| *TSFM* | *94%* | *94%* | *Combined oxidative phosphorylation deficiency 3, 610505* |
| *TSHB* | *100%* | *100%* | *Hypothyroidism, congenital, nongoitrous 4, 275100* |
| *TSPAN7* | *99%* | *99%* | *Intellectual developmental disorder, X-linked 58, 300210* |
| *TTC19* | *100%* | *100%* | *Mitochondrial complex III deficiency, nuclear type 2, 615157* |
| *TTC5* | *100%* | *100%* | *Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244* |
| *TTC8* | *100%* | *100%* | *Bardet-Biedl syndrome 8, 615985*<br>*?Retinitis pigmentosa 51, 613464* |
| *TTI2* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 39, 615541* |
| *TUBA1A* | *100%* | *100%* | *Lissencephaly 3, 611603* |
| *TUBA8* | *100%* | *100%* | *Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840* |
| *TUBB* | *100%* | *99%* | *Symmetric circumferential skin creases, congenital, 1, 156610*<br>*Cortical dysplasia, complex, with other brain malformations 6, 615771* |
| *TUBB2A* | *100%* | *100%* | *Cortical dysplasia, complex, with other brain malformations 5, 615763* |
| *TUBB2B* | *100%* | *100%* | *Cortical dysplasia, complex, with other brain malformations 7, 610031* |
| *TUBB3* | *100%* | *100%* | *Fibrosis of extraocular muscles, congenital, 3A, 600638*<br>*Cortical dysplasia, complex, with other brain malformations 1, 614039* |
| *TUBB4A* | *99%* | *96%* | *Dystonia 4, torsion, autosomal dominant, 128101*<br>*Leukodystrophy, hypomyelinating, 6, 612438* |
| *TUBG1* | *100%* | *100%* | *Cortical dysplasia, complex, with other brain malformations 4, 615412* |
| *TUBGCP2* | *97%* | *97%* | *Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737* |
| *TUBGCP4* | *100%* | *100%* | *Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335* |
| *TUBGCP6* | *100%* | *100%* | *Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270* |
| *TUSC3* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 7, 611093* |
| *TWIST1* | *100%* | *100%* | *Craniosynostosis 1, 123100*<br>*Robinow-Sorauf syndrome, 180750*<br>*Sweeney-Cox syndrome, 617746*<br>*Saethre-Chotzen syndrome with or without eyelid anomalies, 101400* |
| *TWNK* | *100%* | *100%* | *Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245*<br>*Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286*<br>*Perrault syndrome 5, 616138* |

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| *U2AF2* | *100%* | *100%* | *No OMIM disease ID* |
| *UBA5* | *100%* | *100%* | *?Spinocerebellar ataxia, autosomal recessive 24, 617133*<br>*Developmental and epileptic encephalopathy 44, 617132* |
| *UBE2A* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860* |
| *UBE3A* | *100%* | *100%* | *Angelman syndrome, 105830* |
| *UBE3B* | *100%* | *100%* | *Kaufman oculocerebrofacial syndrome, 244450* |
| *UBE4A* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia and gross motor and seech delay, 619639* |
| *UBR1* | *98%* | *98%* | *Johanson-Blizzard syndrome, 243800* |
| *UBR7* | *100%* | *100%* | *Li-Campeau syndrome, 619189* |
| *UBTF* | *100%* | *100%* | *Neurodegeneration, childhood-onset, with brain atrophy, 617672* |
| *UFC1* | *100%* | *100%* | *Neurodevelopmental disorder with spasticity and poor growth, 618076* |
| *UFM1* | *100%* | *100%* | *Leukodystrophy, hypomyelinating, 14, 617899* |
| *UFSP2* | *100%* | *100%* | *?Hip dysplasia, Beukes type, 142669*<br>*Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974*<br>*Developmental and epileptic encephalopathy 106, 620028* |
| *UGDH* | *100%* | *100%* | *Developmental and epileptic encephalopathy 84, 618792* |
| *UGP2* | *96%* | *94%* | *Developmental and epileptic encephalopathy 83, 618744* |
| *UNC13A* | *100%* | *100%* | *No OMIM disease ID* |
| *UNC45A* | *100%* | *100%* | *Osteootohepatoenteric syndrome, 619377* |
| *UNC80* | *100%* | *100%* | *Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801* |
| *UPB1* | *100%* | *100%* | *Beta-ureidopropionase deficiency, 613161* |
| *UPF1* | *100%* | *99%* | *No OMIM disease ID* |
| *UPF3B* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic 14, 300676* |
| *UROC1* | *100%* | *100%* | *?Urocanase deficiency, 276880* |
| *USP27X* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 105, 300984* |
| *USP7* | *100%* | *100%* | *Hao-Fountain syndrome, 616863* |
| *USP9X* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 99, 300919*<br>*Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968* |
| *VAMP1* | *100%* | *100%* | *Myasthenic syndrome, congenital, 25, 618323*<br>*Spastic ataxia 1, autosomal dominant, 108600* |
| *VAMP2* | *100%* | *100%* | *Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760* |
| *VARS1* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802* |
| *VARS2* | *100%* | *100%* | *Combined oxidative phosphorylation deficiency 20, 615917* |
| *VLDLR* | *100%* | *100%* | *Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050* |

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| *VPS11* | *100%* | *100%* | *?Dystonia 32, 619637*<br>*Leukodystrophy, hypomyelinating, 12, 616683* |
| *VPS13B* | *100%* | *99%* | *Cohen syndrome, 216550* |
| *VPS16* | *100%* | *100%* | *Dystonia 30, 619291* |
| *VPS35L* | *100%* | *100%* | *Ritscher-Schinzel syndrome 3, 619135* |
| *VPS37A* | *100%* | *100%* | *Spastic paraplegia 53, autosomal recessive, 614898* |
| *VPS41* | *100%* | *100%* | *Spinocerebellar ataxia, autosomal recessive 29, 619389* |
| *VPS4A* | *100%* | *100%* | *CIMDAG syndrome, 619273* |
| *VPS50* | *100%* | *100%* | *Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685* |
| *VPS53* | *100%* | *100%* | *Pontocerebellar hypoplasia, type 2E, 615851* |
| *VRK1* | *100%* | *100%* | *Pontocerebellar hypoplasia type 1A, 607596* |
| *VWA3B* | *100%* | *100%* | *?Spinocerebellar ataxia, autosomal recessive 22, 616948* |
| *WAC* | *100%* | *100%* | *Desanto-Shinawi syndrome, 616708* |
| *WARS2* | *100%* | *100%* | *Parkinsonism-dystonia 3, childhood-onset, 619738*<br>*Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710* |
| *WASF1* | *100%* | *100%* | *Neurodevelopmental disorder with absent language and variable seizures, 618707* |
| *WASHC4* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 43, 615817* |
| *WDFY3* | *100%* | *100%* | *?Microcephaly 18, primary, autosomal dominant, 617520* |
| *WDPCP* | *98%* | *97%* | *?Bardet-Biedl syndrome 15, 615992*<br>*Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085* |
| *WDR11* | *100%* | *100%* | *Hypogonadotropic hypogonadism 14 with or without anosmia, 614858* |
| *WDR13* | *100%* | *100%* | *No OMIM disease ID* |
| *WDR26* | *100%* | *100%* | *Skraban-Deardorff syndrome, 617616* |
| *WDR37* | *100%* | *100%* | *Neurooculocardiogenitourinary syndrome, 618652* |
| *WDR4* | *100%* | *100%* | *Galloway-Mowat syndrome 6, 618347*<br>*Microcephaly, growth deficiency, seizures, and brain malformations, 618346* |
| *WDR45* | *100%* | *100%* | *Neurodegeneration with brain iron accumulation 5, 300894* |
| *WDR45B* | *100%* | *100%* | *Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977* |
| *WDR62* | *100%* | *100%* | *Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317* |
| *WDR73* | *100%* | *100%* | *Galloway-Mowat syndrome 1, 251300* |
| *WDR81* | *100%* | *100%* | *Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185*<br>*Hydrocephalus, congenital, 3, with brain anomalies, 617967* |

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| *WFS1* | *100%* | *100%* | *Deafness, autosomal dominant 6/14/38, 600965*<br>*?Cataract 41, 116400*<br>*Wolfram-like syndrome, autosomal dominant, 614296*<br>*Wolfram syndrome 1, 222300* |
| *WNK3* | *100%* | *100%* | *No OMIM disease ID* |
| *WWOX* | *100%* | *100%* | *Esophageal squamous cell carcinoma, somatic, 133239*<br>*Developmental and epileptic encephalopathy 28, 616211*<br>*Spinocerebellar ataxia, autosomal recessive 12, 614322* |
| *XPA* | *100%* | *100%* | *Xeroderma pigmentosum, group A, 278700* |
| *XRCC4* | *100%* | *100%* | *Short stature, microcephaly, and endocrine dysfunction, 616541* |
| *XYLT1* | *100%* | *100%* | *Desbuquois dysplasia 2, 615777* |
| *YIF1B* | *90%* | *90%* | *Kaya-Barakat-Masson syndrome, 619125* |
| *YIPF5* | *100%* | *100%* | *Microcephaly, epilepsy, and diabetes syndrome 2, 619278* |
| *YME1L1* | *100%* | *100%* | *?Optic atrophy 11, 617302* |
| *YWHAE* | *100%* | *100%* | *No OMIM disease ID* |
| *YWHAG* | *100%* | *100%* | *Developmental and epileptic encephalopathy 56, 617665* |
| *YY1* | *100%* | *100%* | *Gabriele-de Vries syndrome, 617557* |
| *ZBTB11* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 69, 618383* |
| *ZBTB16* | *100%* | *100%* | *Leukemia, acute promyelocytic, PL2F/RARA type,* |
| *ZBTB18* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 22, 612337* |
| *ZBTB20* | *100%* | *100%* | *Primrose syndrome, 259050* |
| *ZBTB24* | *100%* | *100%* | *Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069* |
| *ZBTB7A* | *100%* | *100%* | *Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769* |
| *ZC3H14* | *100%* | *100%* | *Intellectual developmental disorder, autosomal recessive 56, 617125* |
| *ZC4H2* | *100%* | *100%* | *Wieacker-Wolff syndrome, 314580*<br>*Wieacker-Wolff syndrome, female-restricted, 301041* |
| *ZDHHC9* | *100%* | *100%* | *Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799* |
| *ZEB2* | *97%* | *97%* | *Mowat-Wilson syndrome, 235730* |
| *ZFHX4* | *100%* | *99%* | *No OMIM disease ID* |
| *ZFYVE26* | *100%* | *100%* | *Spastic paraplegia 15, autosomal recessive, 270700* |
| *ZIC1* | *100%* | *100%* | *?Craniosynostosis 6, 616602*<br>*Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736* |
| *ZIC2* | *100%* | *100%* | *Holoprosencephaly 5, 609637* |
| *ZMIZ1* | *100%* | *100%* | *Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659* |
| *ZMYM2* | *100%* | *100%* | *Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522* |

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|---|---|---|---|
| *ZMYND11* | *100%* | *100%* | *Intellectual developmental disorder, autosomal dominant 30, 616083* |
| *ZNF142* | *100%* | *100%* | *Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425* |
| *ZNF148* | *100%* | *100%* | *Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260* |
| *ZNF292* | *99%* | *99%* | *Intellectual developmental disorder, autosomal dominant 64, 619188* |
| *ZNF335* | *100%* | *100%* | *Microcephaly 10, primary, autosomal recessive, 615095* |
| *ZNF407* | *100%* | *100%* | *SIMHA syndrome, 619557* |
| *ZNF41* | *100%* | *100%* | *No OMIM disease ID* |
| *ZNF462* | *100%* | *100%* | *Weiss-Kruszka syndrome, 618619* |
| *ZNF526* | *100%* | *100%* | *Dentici-Novelli neurodevelopmental syndrome, 619877* |
| *ZNF699* | *100%* | *100%* | *DEGCAGS syndrome, 619488* |
| *ZNF711* | *100%* | *100%* | *Intellectual developmental disorder, X-linked 97, 300803* |
| *ZSWIM6* | *98%* | *96%* | *Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865*<br>*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.*
*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 is the chemistry used for WES analysis.*
*Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.*
*Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.*
*Genes with coverage denoting NC are non-protein coding genes.*
*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 28th , 2022.*
*This list is accurate for panel version DG 3.5.0*
*Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*