# ID and epilepsy panel

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| **versie** | v7 (1825 genen) | Centrum voor Medische Genetica Gent |

| Gene | OMIM gene ID | Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern | % coding region covered >=20x |
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| *AAAS* | 605378 | Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive | 99.88% |
| *AARS1* | 601065 | Developmental and epileptic encephalopathy 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 (3), Autosomal dominant; Trichothiodystrophy 8, nonphotosensitive, 619691 (3), Autosomal recessive | 99.99% |
| *AARS2* | 612035 | Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive | 99.98% |
| *AASS* | 605113 | Hyperlysinemia, 238700 (3), Autosomal recessive | 99.61% |
| *ABAT* | 137150 | GABA-transaminase deficiency, 613163 (3), Autosomal recessive | 99.98% |
| *ABCA2* | 600047 | Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive | 100% |
| *ABCC8* | 600509 | Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal recessive, Autosomal dominant; Diabetes mellitus, transient neonatal 2, 610374 (3); Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal recessive, Autosomal dominant | 99.98% |
| *ABCC9* | 601439 | Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Intellectual disability and myopathy syndrome, 619719 (3), Autosomal recessive | 99.92% |
| *ABCD1* | 300371 | Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive | 99.98% |

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| *ABCD4* | 603214 | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive | 100% |
| *ABHD16A* | 142620 | Spastic paraplegia 86, autosomal recessive, 619735 (3), Autosomal recessive | 100% |
| *ABHD5* | 604780 | Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive | 99.98% |
| *ACAD9* | 611103 | Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive | 100% |
| *ACADM* | 607008 | Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive | 96.14% |
| *ACADS* | 606885 | Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive | 99.99% |
| *ACADSB* | 600301 | 2-methylbutyrylglycinuria, 610006 (3), Autosomal recessive | 99.93% |
| *ACAT1* | 607809 | Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive | 99.81% |
| *ACER3* | 617036 | ?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive | 99.76% |
| *ACO2* | 100850 | Optic atrophy 9, 616289 (3), Autosomal recessive, Autosomal dominant; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive | 99.99% |
| *ACOX1* | 609751 | Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive | 99.98% |
| *ACSF3* | 614245 | Combined malonic and methylmalonic aciduria, 614265 (3), Autosomal recessive | 99.99% |
| *ACSL4* | 300157 | Intellectual developmental disorder, X-linked 63, 300387 (3), X-linked dominant | 99.59% |
| *ACTB* | 102630 | Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant | 100% |
| *ACTG1* | 102560 | Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant; Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant | 100% |
| ACTL6A | 604958 | No OMIM phenotype<br>Intellectual disability (BAFopathy) (Pascolini (2020), Clin Genet 97(4):672-674 & Barom (2017), Hum Mutat 38(10):1365-1371), PMID: 31994175 - Autosomal dominant | 99.74% |
| *ACTL6B* | 612458 | Developmental and epileptic encephalopathy 76, 618468 (3), Autosomal recessive; Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant | 99.9% |
| *ACVR1* | 102576 | Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant | 99.94% |
| *ACY1* | 104620 | Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive | 100% |
| *ADAM22* | 603709 | Developmental and epileptic encephalopathy 61, 617933 (3), Autosomal recessive | 99.15% |
| *ADAR* | 146920 | Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive | 99.84% |
| *ADARB1* | 601218 | Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 (3), Autosomal recessive | 94.29% |

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| *ADAT3* | 615302 | Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 (3), Autosomal recessive | 100% |
| *ADCY5* | 600293 | Dyskinesia with orofacial involvement, autosomal dominant, 606703 (3), Autosomal dominant; Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 (3), Autosomal recessive; Dyskinesia with orofacial involvement, autosomal recessive, 619647 (3), Autosomal recessive | 99.98% |
| *ADD1* | 102680 | {Hypertension, essential, salt-sensitive}, 145500 (3), Multifactorial | 100% |
| *ADD3* | 601568 | Cerebral palsy, spastic quadriplegic, 3, 617008 (3), Autosomal recessive | 99.95% |
| *ADGRG1* | 604110 | Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3) | 99.9% |
| *ADGRL1* | 616416 | Developmental delay, behavioral abnormalities, and neuropsychiatric disorders, 620065 (3), Autosomal dominant | 99.98% |
| ADGRL2 | 607018 | No OMIM phenotype<br>A de novo variant in ADGRL2 suggests a novel mechanism underlying the previously undescribed association of extreme microcephaly with severely reduced sulcation and rhombencephalosynapsis. (Vezain (2018), Acta Neuropathol Commun. 6(1):109), PMID: 30340542 - Autosomal dominant | 98.18% |
| *ADK* | 102750 | Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive | 99.78% |
| *ADNP* | 611386 | Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant | 100% |
| *ADPRS* | 610624 | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive | 99.94% |
| *ADSL* | 608222 | Adenylosuccinase deficiency, 103050 (3), Autosomal recessive | 99.93% |
| *AFF2* | 300806 | Intellectual developmental disorder, X-linked 109, 309548 (3), X-linked recessive | 99.89% |
| *AFF3* | 601464 | KINSSHIP syndrome, 619297 (3), Autosomal dominant | 99.63% |
| *AFF4* | 604417 | CHOPS syndrome, 616368 (3), Autosomal dominant | 99.94% |
| *AFG2A (SPATA5)* | 613940 | Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 (3), Autosomal recessive | 99.82% |
| *AFG2B (SPATA5L1)* | 619578 | Deafness, autosomal recessive 119, 619615 (3), Autosomal recessive; Neurodevelopmental disorder with hearing loss and spasticity, 619616 (3), Autosomal recessive | 99.91% |
| *AFG3L2* | 604581 | Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant | 99.97% |
| *AGA* | 613228 | Aspartylglucosaminuria, 208400 (3), Autosomal recessive | 99.92% |

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| AGMO | 613738 | No OMIM phenotype<br>Biallelic variants in AGMO with diminished enzyme activity are associated with a neurodevelopmental disorder (Okur (2019) Hum Genet. 2019 Dec;138(11-12):1259-1266), PMID: 31555905 - Autosomal recessive | 99.77% |
| *AGO1* | 606228 | Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292 (3), Autosomal dominant | 99.73% |
| *AGO2* | 606229 | Lessel-Kreienkamp syndrome, 619149 (3), Autosomal dominant | 99.97% |
| *AGPAT2* | 603100 | Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive | 100% |
| *AGTPBP1* | 606830 | Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive | 99.68% |
| *AHCY* | 180960 | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive | 100% |
| *AHDC1* | 615790 | Xia-Gibbs syndrome, 615829 (3), Autosomal dominant | 100% |
| *AHI1* | 608894 | Joubert syndrome 3, 608629 (3), Autosomal recessive | 99.86% |
| *AIFM1* | 300169 | Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive | 99.92% |
| *AIMP1* | 603605 | Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive | 99.97% |
| *AIMP2* | 600859 | Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive | 99.99% |
| *AK1* | 103000 | Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive | 100% |
| *AKT3* | 611223 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant | 99.81% |
| *ALDH18A1* | 138250 | Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant | 99.96% |
| *ALDH3A2* | 609523 | Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive | 99.95% |
| *ALDH4A1* | 606811 | Hyperprolinemia, type II, 239510 (3), Autosomal recessive | 98.97% |
| *ALDH5A1* | 610045 | Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive | 96.19% |
| *ALDH7A1* | 107323 | Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive | 99.49% |
| *ALG1* | 605907 | Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive | 86.66% |
| *ALG11* | 613666 | Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive | 99.99% |
| *ALG12* | 607144 | Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive | 100% |

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| *ALG13* | 300776 | Developmental and epileptic encephalopathy 36, 300884 (3), X-linked | 99.44% |
| *ALG14* | 612866 | Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 (3), Autosomal recessive; Myopathy, epilepsy, and progressive cerebral atrophy, 619036 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3), Autosomal recessive | 99.34% |
| *ALG2* | 607905 | Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive | 100% |
| *ALG3* | 608750 | Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive | 99.96% |
| *ALG6* | 604566 | Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive | 93.37% |
| *ALG8* | 608103 | Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant | 95.49% |
| *ALG9* | 606941 | Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive | 99.73% |
| *ALKBH8* | 613306 | Intellectual developmental disorder, autosomal recessive 71, 618504 (3), Autosomal recessive | 99.98% |
| *ALMS1* | 606844 | Alstrom syndrome, 203800 (3), Autosomal recessive | 99.9% |
| *ALPL* | 171760 | Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant | 99.88% |
| *ALX1* | 601527 | Frontonasal dysplasia 3, 613456 (3), Autosomal recessive | 97.99% |
| *ALX4* | 605420 | Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive | 100% |
| *AMACR* | 604489 | Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive | 100% |
| *AMER1* | 300647 | Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant | 100% |
| *AMMECR1* | 300195 | Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive | 99.87% |
| *AMPD2* | 102771 | ?Spastic paraplegia 63, 615686 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive | 99.91% |
| *AMT* | 238310 | Glycine encephalopathy 2, 620398 (3) | 100% |
| *ANK3* | 600465 | Intellectual developmental disorder, autosomal recessive 37, 615493 (3), Autosomal recessive | 99.79% |
| *ANKLE2* | 616062 | Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive | 99.99% |
| *ANKRD11* | 611192 | KBG syndrome, 148050 (3), Autosomal dominant | 99.85% |

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| *ANKRD17* | 615929 | Chopra-Amiel-Gordon syndrome, 619504 (3), Autosomal dominant | 99.92% |
| ANKS1B | 607815 | No OMIM phenotype<br>Haploinsufficiency in the ANKS1B gene encoding AIDA-1 leads to a neurodevelopmental syndrome (Carbonell (2019) Nat Commun. 2019 Aug 6;10(1):3529), PMID: 31388001 - Autosomal dominant | 99.74% |
| *ANO10* | 613726 | Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive | 99.93% |
| *ANTXR1* | 606410 | GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant | 99.96% |
| *AP1B1* | 600157 | Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150 (3), Autosomal recessive | 99.99% |
| *AP1G1* | 603533 | Usmani-Riazuddin syndrome, autosomal recessive, 619548 (3); Usmani-Riazuddin syndrome, autosomal dominant, 619467 (3), Autosomal dominant | 99.95% |
| *AP1S1* | 603531 | MEDNIK syndrome, 609313 (3), Autosomal recessive | 99.49% |
| *AP1S2* | 300629 | Pettigrew syndrome, 304340 (3), X-linked recessive | 99.56% |
| *AP2M1* | 601024 | Intellectual developmental disorder 60 with seizures, 618587 (3), Autosomal dominant | 99.69% |
| *AP3B1* | 603401 | Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive | 99.89% |
| *AP3B2* | 602166 | Developmental and epileptic encephalopathy 48, 617276 (3), Autosomal recessive | 100% |
| *AP3D1* | 607246 | ?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive | 100% |
| *AP4B1* | 607245 | Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive | 96.92% |
| *AP4E1* | 607244 | Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive | 99.94% |
| *AP4M1* | 602296 | Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive | 99.98% |
| *AP4S1* | 607243 | Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive | 87.89% |
| *AP5Z1* | 613653 | Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive | 100% |
| *APC2* | 612034 | Cortical dysplasia, complex, with other brain malformations 10, 618677 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 74, 617169 (3), Autosomal recessive | 99.98% |
| *APTX* | 606350 | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive | 99.92% |
| *ARCN1* | 600820 | Short stature-micrognathia syndrome, 617164 (3), Autosomal dominant | 99.92% |
| *ARF1* | 103180 | Periventricular nodular heterotopia 8, 618185 (3), Autosomal dominant | 99.99% |
| ARF3 | 103190 | No OMIM phenotype<br>Dominant ARF3 variants disrupt Golgi integrity and cause a neurodevelopmental disorder recapitulated in zebrafish (Fasano (2022) Nat Commun. 2022 Nov 11;13(1):6841), PMID: 36369169 - Autosomal dominant | 99.99% |

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| *ARFGEF1* | 604141 | Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964 (3), Autosomal dominant | 99.86% |
| *ARFGEF2* | 605371 | Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive | 99.99% |
| *ARG1* | 608313 | Argininemia, 207800 (3), Autosomal recessive | 99.95% |
| *ARHGAP31* | 610911 | Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant | 100% |
| *ARHGEF9* | 300429 | Developmental and epileptic encephalopathy 8, 300607 (3), X-linked | 99.89% |
| *ARID1A* | 603024 | Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant | 99.83% |
| *ARID1B* | 614556 | Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant | 99.69% |
| *ARID2* | 609539 | Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant | 99.48% |
| *ARL13B* | 608922 | Joubert syndrome 8, 612291 (3), Autosomal recessive | 99.53% |
| *ARL6* | 608845 | Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive | 99.9% |
| *ARMC9* | 617612 | Joubert syndrome 30, 617622 (3), Autosomal recessive | 99.77% |
| *ARPC4* | 604226 | Developmental delay, language impairment, and ocular abnormalities, 620141 (3), Autosomal dominant | 100% |
| *ARSA* | 607574 | Metachromatic leukodystrophy, 250100 (3), Autosomal recessive | 99.99% |
| *ARSL* | 300180 | Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive | 99.93% |
| *ARV1* | 611647 | Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive | 99.85% |
| *ARX* | 300382 | Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Intellectual developmental disorder, X-linked 29, 300419 (3), X-linked recessive | 95.36% |
| *ASAH1* | 613468 | Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive | 99.9% |
| *ASH1L* | 607999 | Intellectual developmental disorder, autosomal dominant 52, 617796 (3), Autosomal dominant | 99.79% |
| *ASL* | 608310 | Argininosuccinic aciduria, 207900 (3), Autosomal recessive | 99.98% |
| *ASNS* | 108370 | Asparagine synthetase deficiency, 615574 (3), Autosomal recessive | 99.63% |
| *ASPA* | 608034 | Canavan disease, 271900 (3), Autosomal recessive | 99.98% |
| *ASPM* | 605481 | Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive | 99.57% |
| *ASS1* | 603470 | Citrullinemia, 215700 (3), Autosomal recessive | 77.52% |
| *ASXL1* | 612990 | Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant | 100% |

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| *ASXL2* | 612991 | Shashi-Pena syndrome, 617190 (3), Autosomal dominant | 99.82% |
| *ASXL3* | 615115 | Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant | 99.99% |
| *ATAD1* | 614452 | Hyperekplexia 4, 618011 (3), Autosomal recessive | 99.85% |
| *ATAD3A* | 612316 | Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive | 99.62% |
| *ATCAY* | 608179 | Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive | 100% |
| ATG4D | 611340 | No OMIM phenotype<br>Bi-allelic ATG4D variants are associated with a neurodevelopmental disorder characterized by speech and motor impairment (Morimoto (2023) NPJ Genom Med. 2023 Feb 10;8(1):4), PMID: 36765070, Autosomal recessive | 99.97% |
| *ATG7* | 608760 | Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive | 99.9% |
| *ATIC* | 601731 | AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive | 99.86% |
| *ATL1* | 606439 | Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant | 99.95% |
| *ATM* | 607585 | Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3) | 99.83% |
| *ATN1* | 607462 | Dentatorubral-pallidoluysian atrophy, 125370 (3), Autosomal dominant; Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 (3), Autosomal dominant | 99.9% |
| *ATP13A2* | 610513 | Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive | 99.96% |
| *ATP1A1* | 182310 | Hypomagnesemia, seizures, and impaired intellectual development 2, 618314 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant | 98.41% |
| *ATP1A2* | 182340 | Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant | 99.85% |
| *ATP1A3* | 182350 | Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Developmental and epileptic encephalopathy 99, 619606 (3), Autosomal dominant | 99.98% |

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| *ATP2A2* | 108740 | Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant | 99.98% |
| *ATP2B1* | 108731 | Intellectual developmental disorder, autosomal dominant 66, 619910 (3), Autosomal dominant | 99.39% |
| *ATP5F1A* | 164360 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358 (3), Autosomal dominant; ?Combined oxidative phosphorylation deficiency 22, 616045 (3), Autosomal recessive; ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228 (3), Autosomal recessive | 99.99% |
| *ATP5F1E* | 606153 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3), Autosomal recessive | 100% |
| *ATP5PO* | 600828 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359 (3), Autosomal recessive | 99.94% |
| *ATP6AP2* | 300556 | Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive; Congenital disorder of glycosylation, type IIr, 301045 (3), X-linked recessive | 99.55% |
| *ATP6V0A1* | 192130 | Neurodevelopmental disorder with epilepsy and brain atrophy, 619971 (3), Autosomal recessive; Developmental and epileptic encephalopathy 104, 619970 (3), Autosomal dominant | 99.85% |
| *ATP6V0A2* | 611716 | Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive | 99.92% |
| ATP6V0C | 108745 | No OMIM phenotype<br>ATP6V0C gene variants were identified in individuals with epilepsy, with or without developmental delay (Zhao (2023) J Hum Genet. 2023 Sep;68(9):589-597), PMID: 37161035 - Autosomal dominant | 100% |
| *ATP6V1A* | 607027 | Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Developmental and epileptic encephalopathy 93, 618012 (3), Autosomal dominant | 99.73% |
| *ATP6V1B2* | 606939 | Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant; Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant | 99.99% |
| *ATP7A* | 300011 | Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive | 99.87% |
| *ATP8A2* | 605870 | ?Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive | 100% |
| *ATP9A* | 609126 | Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242 (3), Autosomal recessive | 99.99% |
| *ATPAF2* | 608918 | ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive | 99.96% |

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| *ATR* | 601215 | Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant | 99.83% |
| *ATRX* | 300032 | Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Intellectual disability-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive | 99.44% |
| *ATXN2* | 601517 | {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant | 99.88% |
| *AUH* | 600529 | 3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive | 99.95% |
| *AUTS2* | 607270 | Intellectual developmental disorder, autosomal dominant 26, 615834 (3), Autosomal dominant | 99.87% |
| *AVPR2* | 300538 | Diabetes insipidus, nephrogenic, 1, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive | 100% |
| *B3GALNT2* | 610194 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 (3), Autosomal recessive | 92.79% |
| *B3GALT6* | 615291 | Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive; Al-Gazali syndrome, 609465 (3), Autosomal recessive | 100% |
| *B3GLCT* | 610308 | Peters-plus syndrome, 261540 (3), Autosomal recessive | 99.9% |
| *B4GALNT1* | 601873 | Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive | 99.97% |
| *B4GALT1* | 137060 | Combined low LDL and fibrinogen, 620364 (3), Autosomal recessive; Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive | 99.97% |
| *B4GALT7* | 604327 | Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive | 99.99% |
| *B4GAT1* | 605517 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive | 100% |
| *B9D1* | 614144 | ?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive | 99.8% |
| *B9D2* | 611951 | ?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive | 99.88% |
| *BAP1* | 603089 | Kury-Isidor syndrome, 619762 (3), Autosomal dominant; Tumor predisposition syndrome 1, 614327 (3), Autosomal dominant; {Uveal melanoma, susceptibility to, 2}, 606661 (3), Autosomal dominant | 99.99% |
| BAZ2B | 605683 | No OMIM phenotype<br>Developmental delay, intellectual disability, and autism spectrum disorder (Scott (2020), Hum Mutat), PMID: 31999386 - Autosomal dominant | 99.63% |

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| *BBS1* | 209901 | Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive | 100% |
| *BBS10* | 610148 | Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive | 99.98% |
| *BBS12* | 610683 | Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive | 100% |
| *BBS2* | 606151 | Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive | 99.9% |
| *BBS4* | 600374 | Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive | 99.88% |
| *BBS5* | 603650 | Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive | 99% |
| *BBS7* | 607590 | Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive | 99.42% |
| *BBS9* | 607968 | Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive | 99.75% |
| *BCAP31* | 300398 | Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive | 99.95% |
| *BCAS3* | 607470 | Hengel-Maroofian-Schols syndrome, 619641 (3), Autosomal recessive | 99.31% |
| *BCKDHA* | 608348 | Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive | 99.97% |
| *BCKDHB* | 248611 | Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive | 99.73% |
| *BCKDK* | 614901 | Branched-chain keto acid dehydrogenase kinase deficiency, 614923 (3) | 99.99% |
| *BCL11A* | 606557 | Dias-Logan syndrome, 617101 (3), Autosomal dominant | 99.93% |
| *BCL11B* | 606558 | Immunodeficiency 49, severe combined, 617237 (3), Autosomal dominant; Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant | 100% |
| *BCOR* | 300485 | Microphthalmia, syndromic 2, 300166 (3), X-linked dominant | 99.97% |
| *BCORL1* | 300688 | Shukla-Vernon syndrome, 301029 (3), X-linked recessive | 99.99% |
| *BCS1L* | 603647 | GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive | 99.99% |
| *BICD2* | 609797 | Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 (3), Autosomal dominant | 99.99% |
| *BICRA* | 605690 | Coffin-Siris syndrome 12, 619325 (3), Autosomal dominant | 99.99% |
| *BLM* | 604610 | Bloom syndrome, 210900 (3), Autosomal recessive | 99.8% |
| BLOC1S1 | 601444 | No OMIM phenotype<br>Combining exome/genome sequencing with data repository analysis reveals novel gene-disease associations for a wide range of genetic disorders. (Bertoli-Avella (2021), Genet Med. 23(8):1551-1568), PMID: 33875846 - Autosomal dominant | 99.95% |
| *BLTP1 (KIAA1109)* | 611565 | Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive | 99.79% |

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| *BMP4* | 112262 | Orofacial cleft 11, 600625 (3); Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant | 100% |
| *BOLA3* | 613183 | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive | 99.22% |
| *BPTF* | 601819 | Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755 (3), Autosomal dominant | 99.84% |
| *BRAF* | 164757 | Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3) | 99.78% |
| *BRAT1* | 614506 | Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 (3), Autosomal recessive; Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive | 100% |
| BRD4 | 608749 | No OMIM phenotype<br>Cornelia de Lange-like syndrome (Alesi (2019), Ann Hum Genet 83(2):100-109 & Olley (2018) Case Reports> Nat Genet 50(3):329-332), PMID: 30302754 - Autosomal dominant | 99.98% |
| *BRF1* | 604902 | Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive | 100% |
| *BRPF1* | 602410 | Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant | 100% |
| BRSK2 | 609236 | No OMIM phenotype<br>Neurodevelopmental disorder (Hiatt (2019), Am J Hum Genet 104(4):701-708), PMID: 30879638 - Autosomal dominant | 99.98% |
| *BRWD3* | 300553 | Intellectual developmental disorder, X-linked 93, 300659 (3), X-linked recessive | 99.4% |
| *BSCL2* | 606158 | Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VC, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive | 99.99% |
| *BTD* | 609019 | Biotinidase deficiency, 253260 (3), Autosomal recessive | 100% |
| *BUB1* | 602452 | Colorectal cancer with chromosomal instability, somatic, 114500 (3); Microcephaly 30, primary, autosomal recessive, 620183 (3), Autosomal recessive | 99.64% |
| *BUB1B* | 602860 | Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive | 100% |
| *C12orf4* | 616082 | Intellectual developmental disorder, autosomal recessive 66, 618221 (3), Autosomal recessive | 99.93% |
| *C12orf57* | 615140 | Temtamy syndrome, 218340 (3), Autosomal recessive | 100% |

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| *C2CD3* | 615944 | Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive | 99.88% |
| *C2orf69* | 619219 | Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive | 99.97% |
| *CA2* | 611492 | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive | 99.62% |
| *CA5A* | 114761 | Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive | 99.99% |
| *CA8* | 114815 | Cerebellar ataxia, impaired intellectual development and dysequilibrium syndrome 3, 613227 (3), Autosomal recessive | 99.71% |
| *CACNA1A* | 601011 | Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant | 98.16% |
| *CACNA1B* | 601012 | Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive | 100% |
| *CACNA1C* | 114205 | Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029 (3), Autosomal dominant; Brugada syndrome 3, 611875 (3), Autosomal dominant | 100% |
| *CACNA1D* | 114206 | Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive | 99.98% |
| *CACNA1E* | 601013 | Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant | 99.82% |
| *CACNA1G* | 604065 | Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant | 99.95% |
| *CACNA1I* | 608230 | Neurodevelopmental disorder with speech impairment and with or without seizures, 620114 (3), Autosomal dominant | 99.98% |
| *CACNA2D1* | 114204 | Developmental and epileptic encephalopathy 110, 620149 (3), Autosomal recessive | 97.12% |
| *CACNA2D2* | 607082 | Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive | 99.99% |
| *CACNB4* | 601949 | {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; Episodic ataxia, type 5, 613855 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant | 99.2% |
| *CACNG2* | 602911 | ?Intellectual developmental disorder, autosomal dominant 10, 614256 (3), Autosomal dominant | 99.99% |
| *CAD* | 114010 | Developmental and epileptic encephalopathy 50, 616457 (3), Autosomal recessive | 99.86% |
| *CAMK2A* | 114078 | Intellectual developmental disorder, autosomal dominant 53, 617798 (3), Autosomal dominant; ?Intellectual developmental disorder, autosomal recessive 63, 618095 (3), Autosomal recessive | 99.99% |

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| *CAMK2B* | 607707 | Intellectual developmental disorder, autosomal dominant 54, 617799 (3), Autosomal dominant | 99.92% |
| *CAMK2G* | 602123 | Intellectual developmental disorder, autosomal dominant 59, 618522 (3), Autosomal dominant | 99.95% |
| CAMK4 | 114080 | No OMIM phenotype<br>A unique de novo gain-of-function variant in CAMK4 associated with intellectual disability and hyperkinetic movement disorder. (Zech (2018), old Spring Harb Mol Case Stud. 4(6):a003293), PMID: 30262571 - Autosomal dominant | 99.78% |
| *CAMSAP1* | 613774 | Cortical dysplasia, complex, with other brain malformations 12, 620316 (3), Autosomal recessive | 99.99% |
| *CAMTA1* | 611501 | Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 (3), Autosomal dominant | 99.97% |
| *CAPN10* | 605286 | {Diabetes mellitus, noninsulin-dependent 1}, 601283 (3) | 99.99% |
| *CAPN15* | 603267 | Oculogastrointestinal neurodevelopmental syndrome, 619318 (3), Autosomal recessive | 99.98% |
| CAPRIN1 | 601178 | No OMIM phenotype<br>CAPRIN1 haploinsufficiency causes a neurodevelopmental disorder with language impairment, ADHD and ASD (Pavinato (2023) Brain. 2023 Feb 13;146(2):534-548), PMID: 35979925 - Autosomal dominant | 99.56% |
| *CARS1* | 123859 | Microcephaly, developmental delay, and brittle hair syndrome, 618891 (3), Autosomal recessive | 99.99% |
| *CARS2* | 612800 | Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive | 99.99% |
| *CASK* | 300172 | Intellectual developmental disorder, with or without nystagmus, 300422 (3), X-linked recessive; Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked dominant; FG syndrome 4, 300422 (3), X-linked recessive | 98.95% |
| *CBL* | 165360 | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant | 99.95% |
| *CBS* | 613381 | Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive | 17.79% |
| *CC2D1A* | 610055 | Intellectual developmental disorder, autosomal recessive 3, 608443 (3), Autosomal recessive | 99.98% |
| *CC2D2A* | 612013 | COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive | 99.95% |
| *CCBE1* | 612753 | Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive | 99.52% |
| *CCDC115* | 613734 | Congenital disorder of glycosylation, type IIo, 616828 (3), Autosomal recessive | 99.9% |
| *CCDC174* | 616735 | Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive | 99.98% |

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| CCDC186 | 619249 | No OMIM phenotype<br>Functional Evidence of CCDC186 as a New Disease-Associated Gene with Endocrine and Central Nervous System Alterations (Arrabal (2023) Int J Mol Sci. 2023 Aug 1;24(15):12319), PMID: 37569695 - Autosomal recessive | 99.88% |
| *CCDC22* | 300859 | Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive | 99.91% |
| *CCDC32* | 618941 | Cardiofacioneurodevelopmental syndrome, 619123 (3), Autosomal recessive | 99.97% |
| *CCDC47* | 618260 | Trichohepatoneurodevelopmental syndrome, 618268 (3), Autosomal recessive | 99.97% |
| *CCDC88A* | 609736 | ?PEHO syndrome-like, 617507 (3), Autosomal recessive | 99.48% |
| *CCDC88C* | 611204 | ?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive | 100% |
| *CCM2* | 607929 | Cerebral cavernous malformations-2, 603284 (3), Autosomal dominant | 99.93% |
| *CCND2* | 123833 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant | 100% |
| *CCNK* | 603544 | ?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147 (3), Autosomal dominant | 99.93% |
| *CDC42* | 116952 | Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant | 98.05% |
| *CDC42BPB* | 614062 | Chilton-Okur-Chung neurodevelopmental syndrome, 619841 (3), Autosomal dominant | 99.99% |
| *CDH11* | 600023 | Teebi hypertelorism syndrome 2, 619736 (3), Autosomal dominant; Elsahy-Waters syndrome, 211380 (3), Autosomal recessive | 99.94% |
| *CDH15* | 114019 | Intellectual developmental disorder, autosomal dominant 3, 612580 (3), Autosomal dominant | 99.98% |
| *CDH2* | 114020 | Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 (3), Autosomal dominant; ?Attention deficit-hyperactivity disorder 8, 619957 (3), Autosomal recessive; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant | 99.84% |
| *CDK10* | 603464 | Al Kaissi syndrome, 617694 (3), Autosomal recessive | 99.98% |
| *CDK13* | 603309 | Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant | 99.83% |
| *CDK19* | 614720 | Developmental and epileptic encephalopathy 87, 618916 (3), Autosomal dominant | 99.26% |
| *CDK5RAP2* | 608201 | Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive | 99.97% |
| *CDK6* | 603368 | ?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive | 99.47% |
| *CDK8* | 603184 | Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 (3), Autosomal dominant | 99.81% |
| *CDKL5* | 300203 | Developmental and epileptic encephalopathy 2, 300672 (3), X-linked dominant | 99.88% |

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| *CDKN1C* | 600856 | IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant | 100% |
| *CELF2* | 602538 | Developmental and epileptic encephalopathy 97, 619561 (3), Autosomal dominant | 99.99% |
| CELSR3 | 604264 | No OMIM phenotype<br>CELSR3 variants are associated with febrile seizures and epilepsy with antecedent febrile seizures. (Li (2022), CNS Neurosci Ther. 28(3):382-389), PMID: 34951123 - Autosomal dominant | 99.99% |
| *CENPF* | 600236 | Stromme syndrome, 243605 (3), Autosomal recessive | 99.97% |
| *CENPJ* | 609279 | Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive | 99.92% |
| *CEP104* | 616690 | Joubert syndrome 25, 616781 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 77, 619988 (3), Autosomal recessive | 99.99% |
| *CEP120* | 613446 | Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive | 99.9% |
| *CEP135* | 611423 | Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive | 99.82% |
| *CEP152* | 613529 | Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive | 99.93% |
| *CEP290* | 610142 | Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive | 98.1% |
| *CEP41* | 610523 | Joubert syndrome 15, 614464 (3), Autosomal recessive | 99.99% |
| *CEP55* | 610000 | Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive | 99.92% |
| *CEP57* | 607951 | Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive | 99.92% |
| *CEP63* | 614724 | ?Seckel syndrome 6, 614728 (3), Autosomal recessive | 94.73% |
| *CEP83* | 615847 | Nephronophthisis 18, 615862 (3), Autosomal recessive | 98.68% |
| *CEP85L* | 618865 | Lissencephaly 10, 618873 (3), Autosomal dominant | 100% |
| *CERS1* | 606919 | Epilepsy, progressive myoclonic, 8, 616230 (3), Autosomal recessive | 100% |
| *CERT1* | 604677 | Intellectual developmental disorder, autosomal dominant 34, 616351 (3), Autosomal dominant | 99.66% |
| *CHAMP1* | 616327 | Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579 (3), Autosomal dominant | 99.99% |
| *CHD1* | 602118 | Pilarowski-Bjornsson syndrome, 617682 (3), Autosomal dominant | 99.46% |
| *CHD2* | 602119 | Developmental and epileptic encephalopathy 94, 615369 (3), Autosomal dominant | 99.97% |

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| *CHD3* | 602120 | Snijders Blok-Campeau syndrome, 618205 (3), Autosomal dominant | 99.06% |
| *CHD4* | 603277 | Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant | 99.99% |
| *CHD5* | 610771 | Parenti-Mignot neurodevelopmental syndrome, 619873 (3), Autosomal dominant | 99.97% |
| *CHD7* | 608892 | Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant | 99.99% |
| *CHD8* | 610528 | Intellectual developmental disorder with autism and macrocephaly, 615032 (3), Autosomal dominant | 99.96% |
| *CHKA* | 118491 | Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023 (3), Autosomal recessive | 99.95% |
| *CHKB* | 612395 | Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive | 100% |
| *CHMP1A* | 164010 | Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive | 100% |
| *CHRNA2* | 118502 | Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant | 99.98% |
| *CHRNA4* | 118504 | {Nicotine addiction, susceptibility to}, 188890 (3); Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant | 100% |
| *CHRNB2* | 118507 | Epilepsy, nocturnal frontal lobe, 3, 605375 (3) | 99.99% |
| *CIC* | 612082 | Intellectual developmental disorder, autosomal dominant 45, 617600 (3), Autosomal dominant | 98.32% |
| *CIT* | 605629 | Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive | 99.99% |
| *CKAP2L* | 616174 | Filippi syndrome, 272440 (3), Autosomal recessive | 99.54% |
| *CLCN3* | 600580 | Neurodevelopmental disorder with seizures and brain abnormalities, 619517 (3), Autosomal recessive; Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512 (3), Autosomal dominant | 99.95% |
| *CLCN4* | 302910 | Raynaud-Claes syndrome, 300114 (3), X-linked dominant | 99.98% |
| *CLCN6* | 602726 | Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities, 619173 (3), Autosomal dominant | 100% |
| *CLCNKB* | 602023 | Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive | 99.98% |
| *CLDN11* | 601326 | Leukodystrophy, hypomyelinating, 22, 619328 (3), Autosomal dominant | 100% |
| *CLDN16* | 603959 | Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive | 99.98% |
| *CLDN19* | 610036 | Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive | 99.02% |
| *CLIC2* | 300138 | ?Intellectual developmental disorder, X-linked syndromic 32, 300886 (3), X-linked recessive | 99.82% |
| *CLN3* | 607042 | Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive | 99.92% |
| *CLN5* | 608102 | Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive | 100% |

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| *CLN6* | 606725 | Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive | 100% |
| *CLN8* | 607837 | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive | 100% |
| *CLP1* | 608757 | Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive | 99.98% |
| *CLPB* | 616254 | Neutropenia, severe congenital, 9, autosomal dominant, 619813 (3), Autosomal dominant; 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 (3), Autosomal recessive; 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 (3), Autosomal dominant | 99.97% |
| *CLTC* | 118955 | Intellectual developmental disorder, autosomal dominant 56, 617854 (3), Autosomal dominant | 99.56% |
| *CNKSR2* | 300724 | Intellectual developmental disorder, X-linked syndromic, Houge type, 301008 (3), X-linked | 99.54% |
| *CNNM2* | 607803 | Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and impaired intellectual development 1, 616418 (3), Autosomal recessive, Autosomal dominant | 99.94% |
| *CNOT1* | 604917 | Vissers-Bodmer syndrome, 619033 (3), Autosomal dominant; Holoprosencephaly 12, with or without pancreatic agenesis, 618500 (3), Autosomal dominant | 99.83% |
| *CNOT2* | 604909 | Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 (3), Autosomal dominant | 99% |
| *CNOT3* | 604910 | Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 (3), Autosomal dominant | 99.99% |
| *CNPY3* | 610774 | Developmental and epileptic encephalopathy 60, 617929 (3), Autosomal recessive | 99.98% |
| *CNTN2* | 190197 | ?Epilepsy, myoclonic, familial adult, 5, 615400 (3), Autosomal recessive | 99.95% |
| *CNTNAP1* | 602346 | Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive | 99.98% |
| *CNTNAP2* | 604569 | Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; {Autism susceptibility 15}, 612100 (3) | 99.99% |
| *COA8* | 616003 | Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive | 99.94% |
| *COASY* | 609855 | Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive | 99.98% |
| *COG1* | 606973 | Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive | 100% |
| *COG4* | 606976 | Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant | 99.96% |
| *COG5* | 606821 | Congenital disorder of glycosylation, type IIi, 613612 (3), Autosomal recessive | 99.92% |

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| *COG6* | 606977 | Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive | 99.86% |
| *COG7* | 606978 | Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive | 99.74% |
| *COG8* | 606979 | Congenital disorder of glycosylation, type IIh, 611182 (3) | 100% |
| *COL18A1* | 120328 | Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant | 99.99% |
| *COL4A1* | 120130 | ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant | 99.99% |
| *COL4A2* | 120090 | Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3) | 99.98% |
| *COLEC11* | 612502 | 3MC syndrome 2, 265050 (3), Autosomal recessive | 100% |
| *COLGALT1* | 617531 | Brain small vessel disease 3, 618360 (3), Autosomal recessive | 99.82% |
| *COPB1* | 600959 | Baralle-Macken syndrome, 619255 (3), Autosomal recessive | 99.87% |
| *COPB2* | 606990 | Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884 (3), Autosomal dominant; ?Microcephaly 19, primary, autosomal recessive, 617800 (3), Autosomal recessive | 99.83% |
| *COQ2* | 609825 | {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive | 99.9% |
| *COQ4* | 612898 | Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive | 100% |
| *COQ5* | 616359 | ?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive | 99.95% |
| *COQ7* | 601683 | Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive | 100% |
| *COQ8A* | 606980 | Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive | 100% |
| *COQ9* | 612837 | Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive | 99.62% |
| *COX10* | 602125 | Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive | 99.99% |
| *COX15* | 603646 | Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive | 100% |
| *COX6B1* | 124089 | Mitochondrial complex IV deficiency, nuclear type 7, 619051 (3), Autosomal recessive | 100% |
| *CPA6* | 609562 | Febrile seizures, familial, 11, 614418 (3), Autosomal recessive; Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal recessive, Autosomal dominant | 99.99% |
| *CPE* | 114855 | BDV syndrome, 619326 (3), Autosomal recessive | 99.93% |
| *CPLANE1* | 614571 | Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive | 99.81% |

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| *CPLX1* | 605032 | Developmental and epileptic encephalopathy 63, 617976 (3), Autosomal recessive | 100% |
| *CPS1* | 608307 | Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3) | 99.91% |
| *CPSF3* | 606029 | Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876 (3), Autosomal recessive | 99.93% |
| *CPT2* | 600650 | {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant | 99.65% |
| *CRADD* | 603454 | Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive | 99.9% |
| *CRB2* | 609720 | Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive | 99.95% |
| *CRBN* | 609262 | Intellectual developmental disorder, autosomal recessive 2, 607417 (3), Autosomal recessive | 99.97% |
| *CREBBP* | 600140 | Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant | 99.97% |
| *CRLF1* | 604237 | Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive | 99.99% |
| *CRPPA* | 614631 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive | 99.98% |
| CSDE1 | 191510 | No OMIM phenotype<br>Autism and related neurodevelopmental disabilities (Guo (2019), Sci Adv 25;5(9):eaax2166), PMID: 31579823 - Autosomal dominant | 97.17% |
| *CSF1R* | 164770 | Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 (3), Autosomal dominant | 99.92% |
| CSNK1G1 | 606274 | No OMIM phenotype<br>Heterozygous de novo variants in CSNK1G1 are associated with syndromic developmental delay and autism spectrum disorder. (Gold (2020), Clin Genet. 98(6):571-576), PMID: 33009664 - Autosomal dominant | 99.95% |
| *CSNK2A1* | 115440 | Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant | 99.96% |
| *CSNK2B* | 115441 | Poirier-Bienvenu neurodevelopmental syndrome, 618732 (3), Autosomal dominant | 99.65% |
| *CSPP1* | 611654 | Joubert syndrome 21, 615636 (3), Autosomal recessive | 98.31% |
| *CSTB* | 601145 | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive | 100% |

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| *CTBP1* | 602618 | Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant | 99.98% |
| *CTC1* | 613129 | Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive | 100% |
| *CTCF* | 604167 | Intellectual developmental disorder, autosomal dominant 21, 615502 (3), Autosomal dominant | 99.87% |
| *CTDP1* | 604927 | Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive | 99.97% |
| *CTNNA2* | 114025 | Cortical dysplasia, complex, with other brain malformations 9, 618174 (3), Autosomal recessive | 99.84% |
| *CTNNB1* | 116806 | Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatricoma, somatic, 132600 (3); Colorectal cancer, somatic, 114500 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3); Ovarian cancer, somatic, 167000 (3); Hepatocellular carcinoma, somatic, 114550 (3) | 99.95% |
| *CTNND1* | 601045 | Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant | 99.92% |
| CTNND2 | 604275 | No OMIM phenotype<br>Autism (Turner (2015) Nature 520,51), Autosomal dominant Intellectual disability (Hofmeister (2015) J Med Genet 52,111) - Autosomal dominant | 99.97% |
| CTR9 | 609366 | No OMIM phenotype<br>Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder (Meuwissen (2022) Genet Med. 2022 Jul;24(7):1583-1591), PMID: 35499524 - Autosomal dominant | 99.98% |
| *CTSA* | 613111 | Galactosialidosis, 256540 (3), Autosomal recessive | 99.98% |
| *CTSD* | 116840 | Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive | 100% |
| *CTSF* | 603539 | Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362 (3), Autosomal recessive | 99.96% |
| CTTNBP2 | 609772 | No OMIM phenotype<br>Autism-linked mutations of CTTNBP2 reduce social interaction and impair dendritic spine formation via diverse mechanisms (Shih (2020) Acta Neuropathol Commun. 2020 Nov 9;8(1):185), PMID: 33168105 - Autosomal dominant | 99.97% |
| *CTU2* | 617057 | Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 (3), Autosomal recessive | 99.91% |
| *CUBN* | 602997 | [Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive | 99.99% |
| *CUL3* | 603136 | Neurodevelopmental disorder with or without autism or seizures, 619239 (3), Autosomal dominant; Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant | 99.76% |
| *CUL4B* | 300304 | Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354 (3), X-linked recessive | 99.67% |

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| *CUX1* | 116896 | Global developmental delay with or without impaired intellectual development, 618330 (3), Autosomal dominant | 99.37% |
| *CUX2* | 610648 | Developmental and epileptic encephalopathy 67, 618141 (3), Autosomal dominant | 99.96% |
| *CWC27* | 617170 | Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive | 99.67% |
| *CWF19L1* | 616120 | Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive | 99.91% |
| *CYB5R3* | 613213 | Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive | 99.93% |
| *CYC1* | 123980 | Mitochondrial complex III deficiency, nuclear type 6, 615453 (3), Autosomal recessive | 99.86% |
| *CYFIP2* | 606323 | Developmental and epileptic encephalopathy 65, 618008 (3), Autosomal dominant | 100% |
| *CYP27A1* | 606530 | Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive | 100% |
| *CYP2U1* | 610670 | Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive | 99.99% |
| *D2HGDH* | 609186 | D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive | 100% |
| *DAG1* | 128239 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive | 100% |
| *DARS1* | 603084 | Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive | 98.85% |
| *DARS2* | 610956 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive | 98.31% |
| *DBT* | 248610 | Maple syrup urine disease, type II, 248600 (3), Autosomal recessive | 94.51% |
| *DCAF17* | 612515 | Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive | 99.84% |
| *DCC* | 120470 | Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant; Esophageal carcinoma, somatic, 133239 (3); Colorectal cancer, somatic, 114500 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive | 99.96% |
| *DCHS1* | 603057 | Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive | 100% |
| *DCPS* | 610534 | Al-Raqad syndrome, 616459 (3), Autosomal recessive | 99.98% |
| *DCX* | 300121 | Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked | 99.99% |
| *DDB1* | 600045 | White-Kernohan syndrome, 619426 (3), Autosomal dominant | 99.96% |
| *DDC* | 107930 | Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive | 99.67% |
| *DDHD2* | 615003 | Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive | 99.97% |
| *DDOST* | 602202 | Congenital disorder of glycosylation, type Ir, 614507 (3), Autosomal recessive | 99.93% |

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| *DDX11* | 601150 | Warsaw breakage syndrome, 613398 (3), Autosomal recessive | 99.74% |
| DDX23 | 612172 | No OMIM phenotype<br>Syndromic neurodevelopmental disorder associated with de novo variants in DDX23. (Burns (2021), Am J Med Genet A. 185(10):2863-2872), PMID: 34050707 - Autosomal dominant | 99.94% |
| *DDX3X* | 300160 | Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked recessive, X-linked dominant | 99.01% |
| *DDX59* | 615464 | Orofaciodigital syndrome V, 174300 (3), Autosomal recessive | 99.67% |
| *DDX6* | 600326 | Intellectual developmental disorder with impaired language and dysmorphic facies, 618653 (3), Autosomal dominant | 99.88% |
| *DEAF1* | 602635 | Vulto-van Silfout-de Vries syndrome, 615828 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171 (3), Autosomal recessive | 99.9% |
| *DEGS1* | 615843 | Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive | 99.99% |
| *DENND5A* | 617278 | Developmental and epileptic encephalopathy 49, 617281 (3), Autosomal recessive | 99.99% |
| *DEPDC5* | 614191 | Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant | 99.18% |
| *DHCR24* | 606418 | Desmosterolosis, 602398 (3), Autosomal recessive | 99.93% |
| *DHCR7* | 602858 | Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive | 99.97% |
| *DHDDS* | 608172 | Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive | 98.65% |
| *DHFR* | 126060 | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive | 98.89% |
| *DHPS* | 600944 | Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 (3), Autosomal recessive | 93.17% |
| *DHTKD1* | 614984 | ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant; Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 (3), Autosomal recessive | 99.95% |
| *DHX16* | 603405 | Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant | 99.98% |
| *DHX30* | 616423 | Neurodevelopmental disorder with variable motor and speech impairment, 617804 (3), Autosomal dominant | 99.96% |
| *DHX37* | 617362 | Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 (3), Autosomal recessive; 46XY sex reversal 11, 273250 (3), Autosomal dominant | 99.98% |

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| *DIAPH1* | 602121 | Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive | 99.95% |
| *DIP2B* | 611379 | Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630 (3), Autosomal dominant | 99.65% |
| *DIS3L2* | 614184 | Perlman syndrome, 267000 (3), Autosomal recessive | 99.9% |
| *DKC1* | 300126 | ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive | 99.59% |
| *DLAT* | 608770 | Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive | 99.65% |
| *DLD* | 238331 | Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive | 99.89% |
| *DLG3* | 300189 | Intellectual developmental disorder, X-linked 90, 300850 (3), X-linked recessive | 99.96% |
| *DLG4* | 602887 | Intellectual developmental disorder, autosomal dominant 62, 618793 (3), Autosomal dominant | 99.99% |
| *DLL1* | 606582 | Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 (3), Autosomal dominant | 99.99% |
| *DMD* | 300377 | Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive | 99.76% |
| *DMPK* | 605377 | Myotonic dystrophy 1, 160900 (3), Autosomal dominant | 99.93% |
| *DMXL2* | 612186 | Developmental and epileptic encephalopathy 81, 618663 (3), Autosomal recessive; ?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive | 99.86% |
| DNAH14 | 603341 | No OMIM phenotype<br>DNAH14 variants are associated with neurodevelopmental disorders (Li (2022) Hum Mutat. 2022 Jul;43(7):940-949), PMID: 35438214 - Autosomal recessive | 99.64% |
| *DNAJC12* | 606060 | Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive | 99.72% |
| *DNAJC19* | 608977 | 3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive | 99.76% |
| *DNAJC5* | 611203 | Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350 (3), Autosomal dominant | 99.99% |
| *DNAJC6* | 608375 | Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive; Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive | 99.48% |
| *DNM1* | 602377 | Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352 (3), Autosomal recessive; Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346 (3), Autosomal dominant | 92.28% |
| *DNM1L* | 603850 | Optic atrophy 5, 610708 (3), Autosomal dominant; Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal recessive, Autosomal dominant | 99.4% |

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| *DNMT3A* | 602769 | Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant; Acute myeloid leukemia, somatic, 601626 (3); Heyn-Sproul-Jackson syndrome, 618724 (3), Autosomal dominant | 100% |
| *DNMT3B* | 602900 | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant | 99.98% |
| *DOCK3* | 603123 | Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive | 99.96% |
| *DOCK6* | 614194 | Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive | 100% |
| *DOCK7* | 615730 | Developmental and epileptic encephalopathy 23, 615859 (3), Autosomal recessive | 94.41% |
| *DOCK8* | 611432 | Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive | 99.86% |
| *DOHH* | 611262 | Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066 (3), Autosomal recessive | 100% |
| *DOLK* | 610746 | Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive | 100% |
| *DONSON* | 611428 | Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive; Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive | 99.99% |
| *DPAGT1* | 191350 | Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive; Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive | 100% |
| *DPF2* | 601671 | Coffin-Siris syndrome 7, 618027 (3), Autosomal dominant | 99.99% |
| *DPH1* | 603527 | Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive | 100% |
| *DPH2* | 603456 | Developmental delay with short stature, dysmorphic facial features, and sparse hair 2, 620062 (3), Autosomal recessive | 99.97% |
| *DPH5* | 611075 | Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070 (3), Autosomal recessive | 96.83% |
| *DPM1* | 603503 | Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive | 90.68% |
| *DPM2* | 603564 | Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive | 100% |
| *DPP6* | 126141 | Intellectual developmental disorder, autosomal dominant 33, 616311 (3), Autosomal dominant; {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant | 99.99% |
| *DPYD* | 612779 | Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive | 94.53% |
| *DPYS* | 613326 | Dihydropyrimidinuria, 222748 (3), Autosomal recessive | 99.99% |
| *DPYSL5* | 608383 | Ritscher-Schinzel syndrome 4, 619435 (3), Autosomal dominant | 99.93% |

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| DSCAM | 602523 | No OMIM phenotype<br>The contribution of de novo coding mutations to autism spectrum disorder (Iossifov (2014) Nature 515(7526):216-21), PMID: 25363768 - Autosomal dominant | 99.99% |
| *DTYMK* | 188345 | Neurodegeneration, childhood-onset, with progressive microcephaly, 619847 (3), Autosomal recessive | 99.99% |
| *DYM* | 607461 | Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive | 99.96% |
| *DYNC1H1* | 600112 | Charcot-Marie-Tooth disease, axonal, type 2O, 614228 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 13, 614563 (3), Autosomal dominant | 99.99% |
| *DYNC1I2* | 603331 | Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492 (3), Autosomal recessive | 98.97% |
| *DYRK1A* | 600855 | Intellectual developmental disorder, autosomal dominant 7, 614104 (3), Autosomal dominant | 99.98% |
| *EARS2* | 612799 | Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive | 99.96% |
| *EBF3* | 607407 | Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant | 99.99% |
| *EBP* | 300205 | MEND syndrome, 300960 (3), X-linked recessive; Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant | 99.92% |
| *ECHS1* | 602292 | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive | 100% |
| *ECM1* | 602201 | Urbach-Wiethe disease, 247100 (3), Autosomal recessive | 99.99% |
| *EDC3* | 609842 | ?Intellectual developmental disorder, autosomal recessive 50, 616460 (3), Autosomal recessive | 99.97% |
| *EDEM3* | 610214 | Congenital disorder of glycosylation, type IIv, 619493 (3), Autosomal recessive | 98.38% |
| *EED* | 605984 | Cohen-Gibson syndrome, 617561 (3), Autosomal dominant | 93.81% |
| *EEF1A2* | 602959 | Developmental and epileptic encephalopathy 33, 616409 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 38, 616393 (3), Autosomal dominant | 100% |
| EEF1B2 | 600655 | No OMIM phenotype<br>New evidence that biallelic loss of function in EEF1B2 gene leads to intellectual disability (Larcher (2020) Clin Genet. 2020 Apr;97(4):639-643), PMID: 31845318 - Autosomal recessive | 99.59% |
| EEF1D | 130592 | No OMIM phenotype<br>Expanding the spectrum of EEF1D neurodevelopmental disorders: Biallelic variants in the guanine exchange domain (Averdunk (2023) Clin Genet. 2023 Apr;103(4):484-491), PMID: 36576126 - Autosomal recessive | 99.99% |
| *EEF2* | 130610 | ?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant | 99.95% |

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| EFNB2 | 600527 | No OMIM phenotype<br>EFNB2 haploinsufficiency causes a syndromic neurodevelopmental disorder. (Lévy (2018), Clin Genet. 93(6):1141-1147), PMID: 29508392 - Autosomal dominant | 99.92% |
| *EFTUD2* | 603892 | Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant | 99.93% |
| *EHMT1* | 607001 | Kleefstra syndrome 1, 610253 (3), Autosomal dominant | 98.38% |
| *EIF2AK2* | 176871 | Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 (3), Autosomal dominant; Dystonia 33, 619687 (3), Autosomal recessive, Autosomal dominant | 99.7% |
| *EIF2AK3* | 604032 | Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive | 97.43% |
| *EIF2B1* | 606686 | Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896 (3), Autosomal recessive | 99.98% |
| *EIF2B2* | 606454 | Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312 (3) | 99.9% |
| *EIF2B3* | 606273 | Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313 (3) | 97.26% |
| *EIF2B4* | 606687 | Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314 (3) | 99.96% |
| *EIF2B5* | 603945 | Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 (3) | 99.98% |
| *EIF2S3* | 300161 | MEHMO syndrome, 300148 (3), X-linked recessive | 99.34% |
| *EIF3F* | 603914 | Intellectual developmental disorder, autosomal recessive 67, 618295 (3), Autosomal recessive | 99.96% |
| EIF4A2 | 601102 | No OMIM phenotype<br>De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation (Mao (2020) Am J Hum Genet. 2020 Apr 2;106(4):570-583), PMID: 32197074 - Autosomal dominant | 99.99% |
| *EIF4A3* | 608546 | Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive | 99.99% |
| *EIF5A* | 600187 | Faundes-Banka syndrome, 619376 (3), Autosomal dominant | 100% |
| *ELAC2* | 605367 | {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3); Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive | 99.9% |
| *ELOVL4* | 605512 | Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 (3), Autosomal recessive | 99.91% |
| *ELP2* | 616054 | Intellectual developmental disorder, autosomal recessive 58, 617270 (3), Autosomal recessive | 99.82% |
| *EMC1* | 616846 | Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive | 99.85% |
| *EMC10* | 614545 | Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264 (3), Autosomal recessive | 99.96% |
| *EML1* | 602033 | Band heterotopia, 600348 (3), Autosomal recessive | 99.99% |

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| *EMX2* | 600035 | Schizencephaly, 269160 (3) | 100% |
| *ENTPD1* | 601752 | Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive | 99.98% |
| *EP300* | 602700 | Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant | 99.97% |
| *EPB41L1* | 602879 | ?Intellectual developmental disorder, autosomal dominant 11, 614257 (3), Autosomal dominant | 100% |
| *EPG5* | 615068 | Vici syndrome, 242840 (3), Autosomal recessive | 99.95% |
| *EPM2A* | 607566 | Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive | 99.99% |
| *EPRS1* | 138295 | Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive | 99.53% |
| *ERBB4* | 600543 | Amyotrophic lateral sclerosis 19, 615515 (3), Autosomal dominant | 99.92% |
| *ERCC1* | 126380 | Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive | 99.96% |
| *ERCC2* | 126340 | Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive | 99.98% |
| *ERCC3* | 133510 | Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive | 99.9% |
| *ERCC5* | 133530 | Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive | 99.99% |
| *ERCC6* | 609413 | UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant | 99.6% |
| *ERCC6L2* | 615667 | Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive | 99.94% |
| *ERCC8* | 609412 | UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive | 99.79% |
| *ERLIN2* | 611605 | Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive | 99.94% |
| *ESAM* | 614281 | Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371 (3), Autosomal recessive | 99.99% |
| *ESCO2* | 609353 | Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive | 99.92% |
| *ETFA* | 608053 | Glutaric acidemia IIA, 231680 (3), Autosomal recessive | 99.88% |
| *ETFB* | 130410 | Glutaric acidemia IIB, 231680 (3), Autosomal recessive | 100% |

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| *ETFDH* | 231675 | Glutaric acidemia IIC, 231680 (3), Autosomal recessive | 99.82% |
| *ETHE1* | 608451 | Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive | 84.97% |
| *EXOC2* | 615329 | Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306 (3), Autosomal recessive | 99.97% |
| *EXOC7* | 608163 | Neurodevelopmental disorder with seizures and brain atrophy, 619072 (3), Autosomal recessive | 100% |
| *EXOSC2* | 602238 | Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive | 100% |
| *EXOSC3* | 606489 | Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive | 100% |
| *EXOSC5* | 606492 | Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576 (3), Autosomal recessive | 99.98% |
| *EXOSC8* | 606019 | Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive | 99.91% |
| *EXOSC9* | 606180 | Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive | 94.91% |
| *EXT2* | 608210 | Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant | 99.98% |
| *EXTL3* | 605744 | Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive | 99.99% |
| *EZH2* | 601573 | Weaver syndrome, 277590 (3), Autosomal dominant | 99.89% |
| *FA2H* | 611026 | Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive | 99.98% |
| *FAM111A* | 615292 | Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant; Gracile bone dysplasia, 602361 (3), Autosomal dominant | 100% |
| *FAM149B1* | 618413 | Joubert syndrome 36, 618763 (3), Autosomal recessive | 99.69% |
| *FAM20C* | 611061 | Raine syndrome, 259775 (3), Autosomal recessive | 100% |
| *FAM50A* | 300453 | Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261 (3), X-linked recessive | 99.99% |
| *FANCD2* | 613984 | Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive | 99.86% |
| *FAR1* | 616107 | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant | 99.82% |
| *FARS2* | 611592 | Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive | 100% |
| *FARSA* | 602918 | ?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive | 100% |
| *FARSB* | 609690 | Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive | 99.64% |
| *FASTKD2* | 612322 | Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive | 99.93% |

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| *FAT4* | 612411 | Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive | 99.98% |
| FBRSL1 | 620123 | No OMIM phenotype<br>De novo mutations in FBRSL1 cause a novel recognizable malformation and intellectual disability syndrome (Ufartes (2020), Hum Genet. 139(11):1363-1379), PMID: 32424618 - Autosomal dominant | 95.66% |
| *FBXL3* | 605653 | Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 (3), Autosomal recessive | 99.48% |
| *FBXL4* | 605654 | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive | 100% |
| *FBXO11* | 607871 | Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 (3), Autosomal dominant | 99.53% |
| *FBXO28* | 609100 | Developmental and epileptic encephalopathy 100, 619777 (3), Autosomal dominant | 99.67% |
| *FBXO31* | 609102 | ?Intellectual developmental disorder, autosomal recessive 45, 615979 (3), Autosomal recessive | 99.99% |
| *FBXW11* | 605651 | Neurodevelopmental, jaw, eye, and digital syndrome, 618914 (3), Autosomal dominant | 99.96% |
| *FBXW7* | 606278 | Developmental delay, hypotonia, and impaired language, 620012 (3), Autosomal dominant | 99.9% |
| *FCSK* | 608675 | Congenital disorder of glycosylation with defective fucosylation 2, 618324 (3), Autosomal recessive | 99.97% |
| *FDFT1* | 184420 | Squalene synthase deficiency, 618156 (3), Autosomal recessive | 99.99% |
| *FGD1* | 300546 | Intellectual developmental disorder, X-linked syndromic 16, 305400 (3), X-linked recessive; Aarskog-Scott syndrome, 305400 (3), X-linked recessive | 99.96% |
| *FGF12* | 601513 | Developmental and epileptic encephalopathy 47, 617166 (3), Autosomal dominant | 99.94% |
| *FGF13* | 300070 | Developmental and epileptic encephalopathy 90, 301058 (3), X-linked recessive, X-linked dominant; Intellectual developmental disorder, X-linked 110, 301095 (3), X-linked recessive | 99.76% |
| *FGF14* | 601515 | Spinocerebellar ataxia 27A, 193003 (3), Autosomal dominant; Spinocerebellar ataxia 27B, late-onset, 620174 (3), Autosomal dominant | 99.99% |
| *FGFR1* | 136350 | Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3) | 100% |

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| *FGFR2* | 176943 | Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome 1, 149730 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant | 99.99% |
| *FGFR3* | 134934 | Muenke syndrome, 602849 (3), Autosomal dominant; SADDAN, 616482 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Bladder cancer, somatic, 109800 (3); LADD syndrome 2, 620192 (3), Autosomal dominant; Achondroplasia, 100800 (3), Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant | 100% |
| *FH* | 136850 | Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive | 99.95% |
| *FIBP* | 608296 | Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive | 99.83% |
| *FIG4* | 609390 | Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive | 99.83% |
| *FITM2* | 612029 | Siddiqi syndrome, 618635 (3), Autosomal recessive | 99.99% |
| *FKRP* | 606596 | Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive | 100% |
| *FKTN* | 607440 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152 (3), Autosomal recessive | 99.94% |

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| *FLNA* | 300017 | Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive | 99.99% |
| *FLVCR1* | 609144 | Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive | 99.91% |
| *FLVCR2* | 610865 | Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive | 100% |
| *FMN2* | 606373 | Intellectual developmental disorder, autosomal recessive 47, 616193 (3), Autosomal recessive | 99.99% |
| *FMR1* | 309550 | Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Fragile X syndrome, 300624 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked | 99.56% |
| *FOLR1* | 136430 | Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive | 100% |
| *FOXG1* | 164874 | Rett syndrome, congenital variant, 613454 (3), Autosomal dominant | 99.91% |
| *FOXP1* | 605515 | Intellectual developmental disorder with language impairment with or without autistic features, 613670 (3), Autosomal dominant | 99.98% |
| *FOXP2* | 605317 | Speech-language disorder-1, 602081 (3), Autosomal dominant | 99.98% |
| FOXP4 | 608924 | No OMIM phenotype<br>Heterozygous variants that disturb the transcriptional repressor activity of FOXP4 cause a developmental disorder with speech/language delays and multiple congenital abnormalities (Snijders Blok (2021) Genet Med. 2021 Mar;23(3):534-542), PMID: 33110267 - Autosomal dominant | 99.98% |
| *FOXRED1* | 613622 | Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive | 100% |
| *FRA10AC1* | 608866 | Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113 (3), Autosomal recessive | 99.76% |
| *FRAS1* | 607830 | Fraser syndrome 1, 219000 (3), Autosomal recessive | 99.97% |
| *FREM2* | 608945 | Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive | 99.97% |
| *FRMD4A* | 616305 | ?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive | 100% |
| *FRMD5* | 616309 | Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094 (3), Autosomal dominant | 100% |

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| *FRMPD4* | 300838 | Intellectual developmental disorder, X-linked 104, 300983 (3), X-linked | 99.92% |
| *FRRS1L* | 604574 | Developmental and epileptic encephalopathy 37, 616981 (3), Autosomal recessive | 99.91% |
| *FTCD* | 606806 | Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive | 99.99% |
| *FTO* | 610966 | Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive | 99.66% |
| *FTSJ1* | 300499 | Intellectual developmental disorder, X-linked 9, 309549 (3), X-linked recessive | 99.95% |
| *FUCA1* | 612280 | Fucosidosis, 230000 (3), Autosomal recessive | 98.72% |
| *FUT8* | 602589 | Congenital disorder of glycosylation with defective fucosylation 1, 618005 (3), Autosomal recessive | 99.98% |
| *FXYD2* | 601814 | Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant | 100% |
| *FZR1* | 603619 | Developmental and epileptic encephalopathy 109, 620145 (3), Autosomal dominant | 99.99% |
| GABBR1 | 603540 | No OMIM phenotype<br>GABBR1 monoallelic de novo variants linked to neurodevelopmental delay and epilepsy (Cediel (2022) Am J Hum Genet. 2022 Oct 6;109(10):1885-1893), PMID: 36103875 - Autosomal dominant | 99.91% |
| *GABBR2* | 607340 | {Nicotine dependence, protection against}, 188890 (3); {Nicotine dependence, susceptibility to}, 188890 (3); Developmental and epileptic encephalopathy 59, 617904 (3), Autosomal dominant; Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant | 99.96% |
| *GABRA1* | 137160 | {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Developmental and epileptic encephalopathy 19, 615744 (3), Autosomal dominant; {Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3) | 100% |
| *GABRA2* | 137140 | Developmental and epileptic encephalopathy 78, 618557 (3), Autosomal dominant; {Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial | 99.9% |
| *GABRA3* | 305660 | Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091 (3), X-linked | 99.96% |
| *GABRA5* | 137142 | Developmental and epileptic encephalopathy 79, 618559 (3), Autosomal dominant | 99.96% |
| *GABRB1* | 137190 | Developmental and epileptic encephalopathy 45, 617153 (3), Autosomal dominant | 99.97% |
| *GABRB2* | 600232 | Developmental and epileptic encephalopathy 92, 617829 (3), Autosomal dominant | 99.92% |
| *GABRB3* | 137192 | {Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Developmental and epileptic encephalopathy 43, 617113 (3), Autosomal dominant | 99.66% |
| *GABRD* | 137163 | {Epilepsy, idiopathic generalized, 10}, 613060 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to}, 613060 (3), Autosomal dominant; {Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060 (3), Autosomal dominant | 99.99% |

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| *GABRG2* | 137164 | Developmental and epileptic encephalopathy 74, 618396 (3), Autosomal dominant; Febrile seizures, familial, 8, 607681 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 3, 607681 (3), Autosomal dominant | 91.88% |
| *GAD1* | 605363 | Developmental and epileptic encephalopathy 89, 619124 (3), Autosomal recessive | 99.92% |
| *GAL* | 137035 | ?Epilepsy, familial temporal lobe, 8, 616461 (3), Autosomal dominant | 99.9% |
| *GALC* | 606890 | Krabbe disease, 245200 (3), Autosomal recessive | 99.92% |
| *GALE* | 606953 | Galactose epimerase deficiency, 230350 (3), Autosomal recessive | 99.9% |
| *GALNT2* | 602274 | Congenital disorder of glycosylation, type IIt, 618885 (3), Autosomal recessive | 99.44% |
| *GALT* | 606999 | Galactosemia, 230400 (3), Autosomal recessive | 100% |
| *GAMT* | 601240 | Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive | 100% |
| *GATAD2B* | 614998 | GAND syndrome, 615074 (3), Autosomal dominant | 99.44% |
| *GATM* | 602360 | Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant | 99.92% |
| *GBA1 (GBA)* | 606463 | {Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant | 96.92% |
| *GCDH* | 608801 | Glutaricaciduria, type I, 231670 (3), Autosomal recessive | 100% |
| *GCH1* | 600225 | Dystonia, DOPA-responsive, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive | 99.94% |
| *GCK* | 138079 | MODY, type II, 125851 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 1, 606176 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant | 99.99% |
| *GCSH* | 238330 | Multiple mitochondrial dysfunctions syndrome 7, 620423 (3), Autosomal recessive | 98.75% |
| *GDI1* | 300104 | Intellectual developmental disorder, X-linked 41, 300849 (3), X-linked dominant | 99.99% |
| *GEMIN4* | 606969 | Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913 (3), Autosomal recessive | 100% |
| *GEMIN5* | 607005 | Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 (3), Autosomal recessive | 99.96% |
| *GFAP* | 137780 | Alexander disease, 203450 (3), Autosomal dominant | 99.99% |
| *GFER* | 600924 | Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive | 100% |

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| *GFM1* | 606639 | Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive | 99.95% |
| *GFM2* | 606544 | Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive | 99.87% |
| *GJB1* | 304040 | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant | 100% |
| *GJC2* | 608803 | Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive | 100% |
| *GK* | 300474 | Glycerol kinase deficiency, 307030 (3), X-linked recessive | 99.27% |
| *GLB1* | 611458 | GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive | 100% |
| *GLDC* | 238300 | Glycine encephalopathy1, 605899 (3), Autosomal recessive | 99.99% |
| *GLI2* | 165230 | Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant | 99.93% |
| *GLI3* | 165240 | Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant | 100% |
| *GLIS3* | 610192 | Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive | 99.99% |
| *GLRA1* | 138491 | Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant | 100% |
| *GLRA2* | 305990 | Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076 (3), X-linked | 99.87% |
| *GLRB* | 138492 | Hyperekplexia 2, 614619 (3), Autosomal recessive | 99.79% |
| *GLS* | 138280 | Global developmental delay, progressive ataxia, and elevated glutamine, 618412 (3), Autosomal recessive; ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 (3), Autosomal dominant; Developmental and epileptic encephalopathy 71, 618328 (3), Autosomal recessive | 99.78% |
| *GLUD1* | 138130 | Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant | 99.8% |
| *GLUL* | 138290 | Glutamine deficiency, congenital, 610015 (3), Autosomal recessive | 99.88% |
| *GLYCTK* | 610516 | D-glyceric aciduria, 220120 (3), Autosomal recessive | 100% |
| *GM2A* | 613109 | GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive | 100% |
| *GMNN* | 602842 | Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant | 99.73% |
| *GMPPA* | 615495 | Alacrima, achalasia, and impaired intellectual development syndrome, 615510 (3), Autosomal recessive | 99.97% |

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| *GMPPB* | 615320 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive | 100% |
| *GNAI1* | 139310 | Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854 (3), Autosomal dominant | 99.7% |
| *GNAO1* | 139311 | Developmental and epileptic encephalopathy 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant | 99.86% |
| *GNAQ* | 600998 | Capillary malformations, congenital, 1, somatic, mosaic, 163000 (3); Sturge-Weber syndrome, somatic, mosaic, 185300 (3) | 99.94% |
| *GNAS* | 139320 | ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant | 100% |
| *GNB1* | 139380 | Myelodysplastic syndrome, somatic, 614286 (3); Leukemia, acute lymphoblastic, somatic, 613065 (3); Intellectual developmental disorder, autosomal dominant 42, 616973 (3), Autosomal dominant | 100% |
| *GNB2* | 139390 | Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 (3), Autosomal dominant; ?Sick sinus syndrome 4, 619464 (3), Autosomal dominant | 99.99% |
| *GNB5* | 604447 | Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 (3), Autosomal recessive; Intellectual developmental disorder with cardiac arrhythmia, 617173 (3), Autosomal recessive | 99.98% |
| *GNPAT* | 602744 | Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive | 99.78% |
| *GNPTAB* | 607840 | Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive; Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive | 99.76% |
| *GNPTG* | 607838 | Mucolipidosis III gamma, 252605 (3), Autosomal recessive | 100% |
| *GNS* | 607664 | Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive | 99.59% |
| *GOLGA2* | 602580 | Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240 (3), Autosomal recessive | 100% |
| *GOSR2* | 604027 | Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive; Muscular dystrophy, congenital, with or without seizures, 620166 (3), Autosomal recessive | 98.92% |
| *GOT2* | 138150 | Developmental and epileptic encephalopathy 82, 618721 (3), Autosomal recessive | 99.78% |

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| *GPAA1* | 603048 | Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive | 100% |
| *GPC3* | 300037 | Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive | 99.6% |
| *GPC4* | 300168 | Keipert syndrome, 301026 (3), X-linked recessive | 99.89% |
| *GPHN* | 603930 | Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive | 99.94% |
| *GPSM2* | 609245 | Chudley-McCullough syndrome, 604213 (3), Autosomal recessive | 96.11% |
| *GPT2* | 138210 | Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive | 99.97% |
| *GRIA1* | 138248 | ?Intellectual developmental disorder, autosomal recessive 76, 619931 (3), Autosomal recessive; Intellectual developmental disorder, autosomal dominant 67, 619927 (3), Autosomal dominant | 99.97% |
| *GRIA2* | 138247 | Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917 (3), Autosomal dominant | 99.99% |
| *GRIA3* | 305915 | Intellectual developmental disorder, X-linked syndromic, Wu type, 300699 (3), X-linked recessive | 99.93% |
| *GRIA4* | 138246 | Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 (3), Autosomal dominant | 99.93% |
| *GRID2* | 602368 | Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive | 99.97% |
| *GRIK2* | 138244 | Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580 (3), Autosomal dominant; Intellectual developmental disorder, autosomal recessive 6, 611092 (3), Autosomal recessive | 99.9% |
| *GRIN1* | 138249 | Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Developmental and epileptic encephalopathy 101, 619814 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant | 100% |
| *GRIN2A* | 138253 | Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570 (3), Autosomal dominant | 100% |
| *GRIN2B* | 138252 | Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant | 99.99% |
| *GRIN2D* | 602717 | Developmental and epileptic encephalopathy 46, 617162 (3), Autosomal dominant | 99.97% |
| *GRIP1* | 604597 | Fraser syndrome 3, 617667 (3), Autosomal recessive | 99.83% |
| *GRM1* | 604473 | Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant | 100% |

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| *GRM7* | 604101 | Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 (3), Autosomal recessive | 99.99% |
| *GRN* | 138945 | Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive | 100% |
| *GSS* | 601002 | Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive; Glutathione synthetase deficiency, 266130 (3), Autosomal recessive | 99.99% |
| *GTF2E2* | 189964 | Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive | 100% |
| *GTF2H5* | 608780 | Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive | 100% |
| *GTPBP2* | 607434 | Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive | 99.98% |
| *GTPBP3* | 608536 | Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive | 99.99% |
| *GUSB* | 611499 | Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive | 95.07% |
| *H1-4* | 142220 | Rahman syndrome, 617537 (3), Autosomal dominant | 100% |
| *H3-3A* | 601128 | Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720 (3), Autosomal dominant | 35.67% |
| *H3-3B* | 601058 | Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721 (3), Autosomal dominant | 100% |
| *H4C11* | 602826 | ?Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 2, 619759 (3), Autosomal dominant | 99.08% |
| *H4C3* | 602827 | Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758 (3), Autosomal dominant | 99.99% |
| *H4C5* | 602830 | Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950 (3), Autosomal dominant | 100% |
| *H4C9* | 602833 | Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951 (3), Autosomal dominant | 100% |
| *HAAO* | 604521 | Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 (3), Autosomal recessive | 99.96% |
| *HACE1* | 610876 | Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive | 99.76% |
| *HADH* | 601609 | Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive; 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive | 99.86% |
| *HADHA* | 600890 | HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency 1, 609015 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive | 99.98% |
| *HAX1* | 605998 | Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive | 100% |
| *HCCS* | 300056 | Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant | 99.9% |
| *HCFC1* | 300019 | Methylmalonic aciduria and homocysteinemia, cblX type, 309541 (3), X-linked recessive | 99.99% |
| *HCN1* | 602780 | Developmental and epileptic encephalopathy 24, 615871 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 10, 618482 (3), Autosomal dominant | 99.99% |

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| *HCN2* | 602781 | Febrile seizures, familial, 2, 602477 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 11, 602477 (3), Autosomal dominant | 97.35% |
| *HDAC4* | 605314 | Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797 (3), Autosomal dominant | 99.98% |
| *HDAC6* | 300272 | ?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant | 99.96% |
| *HDAC8* | 300269 | Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant | 99.74% |
| *HEATR3* | 614951 | Diamond-Blackfan anemia 21, 620072 (3), Autosomal recessive | 99.72% |
| *HECTD4* | 620209 | Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250 (3), Autosomal recessive | 99.96% |
| *HECW2* | 617245 | Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 (3), Autosomal dominant | 99.91% |
| *HEPACAM* | 611642 | Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926 (3), Autosomal dominant | 100% |
| *HERC1* | 605109 | Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive | 99.92% |
| *HERC2* | 605837 | Intellectual developmental disorder, autosomal recessive 38, 615516 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive | 95.66% |
| *HESX1* | 601802 | Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant | 99.77% |
| *HEXA* | 606869 | [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive | 99.99% |
| *HEXB* | 606873 | Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive | 99.91% |
| *HGSNAT* | 610453 | Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive | 99.93% |
| *HIBCH* | 610690 | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive | 99.7% |
| *HID1* | 605752 | Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983 (3), Autosomal recessive | 99.99% |
| *HIVEP2* | 143054 | Intellectual developmental disorder, autosomal dominant 43, 616977 (3), Autosomal dominant | 99.99% |

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| *HK1* | 142600 | Retinitis pigmentosa 79, 617460 (3), Autosomal dominant; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive | 99.97% |
| *HLCS* | 609018 | Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive | 99.97% |
| HMGB1 | 163905 | No OMIM phenotype<br>Heterozygous HMGB1 loss-of-function variants are associated with developmental delay and microcephaly (Uguen (2021), Clin Genet. 100(4):386-395), PMID: 34164801 - Autosomal dominant | 54.18% |
| *HMGCL* | 613898 | HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive | 99.31% |
| *HNMT* | 605238 | Intellectual developmental disorder, autosomal recessive 51, 616739 (3), Autosomal recessive; {Asthma, susceptibility to}, 600807 (3), Autosomal dominant | 99.53% |
| *HNRNPH1* | 601035 | Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083 (3), Autosomal dominant | 99.98% |
| *HNRNPH2* | 300610 | Intellectual developmental disorder, X-linked syndromic, Bain type, 300986 (3), X-linked dominant | 99.96% |
| *HNRNPK* | 600712 | Au-Kline syndrome, 616580 (3), Autosomal dominant | 99.93% |
| *HNRNPR* | 607201 | Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073 (3), Autosomal dominant | 98.05% |
| *HNRNPU* | 602869 | Developmental and epileptic encephalopathy 54, 617391 (3), Autosomal dominant | 99.95% |
| *HOXA1* | 142955 | Bosley-Salih-Alorainy syndrome, 601536 (3), Autosomal recessive; Athabaskan brainstem dysgenesis syndrome, 601536 (3), Autosomal recessive | 100% |
| *HPD* | 609695 | Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive | 99.99% |
| *HPDL* | 618994 | Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 (3), Autosomal recessive; Spastic paraplegia 83, autosomal recessive, 619027 (3), Autosomal recessive | 99.99% |
| *HPRT1* | 308000 | Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive | 97.8% |
| *HRAS* | 190020 | Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant | 100% |
| *HS2ST1* | 604844 | Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive | 95.79% |

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| *HSD17B10* | 300256 | HSD10 mitochondrial disease, 300438 (3), X-linked dominant | 99.98% |
| *HSD17B4* | 601860 | D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive | 99.71% |
| *HSPA9* | 600548 | Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant | 99.96% |
| *HSPD1* | 118190 | Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive | 83.42% |
| *HTRA2* | 606441 | {Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive | 99.99% |
| *HUWE1* | 300697 | Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 (3), X-linked | 99.88% |
| *HYCC1 (FAM126A)* | 610531 | Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive | 99.81% |
| *IARS1* | 600709 | Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive | 99.89% |
| *IARS2* | 612801 | Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive | 99.77% |
| *IBA57* | 615316 | Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive | 100% |
| *IDH2* | 147650 | D-2-hydroxyglutaric aciduria 2, 613657 (3) | 100% |
| *IDS* | 300823 | Mucopolysaccharidosis II, 309900 (3), X-linked recessive | 99.82% |
| *IDUA* | 252800 | Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive | 99.99% |
| *IER3IP1* | 609382 | Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive | 99.92% |
| *IFIH1* | 606951 | Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant | 99.84% |
| *IFT172* | 607386 | Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive | 99.98% |
| *IFT27* | 615870 | Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive | 100% |
| *IFT74* | 608040 | Bardet-Biedl syndrome 22, 617119 (3), Autosomal recessive; Spermatogenic failure 58, 619585 (3), Autosomal recessive; Joubert syndrome 40, 619582 (3), Autosomal recessive | 99.71% |
| *IGBP1* | 300139 | ?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472 (3), X-linked recessive | 99.9% |

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| *IGF1* | 147440 | Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive | 100% |
| *IGF1R* | 147370 | Insulin-like growth factor I, resistance to, 270450 (3), Autosomal recessive, Autosomal dominant | 100% |
| *IKBKG* | 300248 | Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked | 57.34% |
| *IL1RAPL1* | 300206 | Intellectual developmental disorder, X-linked 21, 300143 (3), X-linked recessive | 99.77% |
| *IMPA1* | 602064 | Intellectual developmental disorder, autosomal recessive 59, 617323 (3), Autosomal recessive | 99.95% |
| *IMPDH2* | 146691 | [IMPDH2 enzyme activity, variation in], 617995 (3) | 100% |
| *INPP5E* | 613037 | Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive | 99.85% |
| *INPP5K* | 607875 | Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive | 99.94% |
| *INTS1* | 611345 | Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 (3), Autosomal recessive | 100% |
| *INTS11* | 611354 | Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428 (3), Autosomal recessive | 100% |
| *INTS8* | 611351 | ?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572 (3), Autosomal recessive | 99.89% |
| *IQSEC1* | 610166 | Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 (3), Autosomal recessive | 99.99% |
| *IQSEC2* | 300522 | Intellectual developmental disorder, X-linked 1, 309530 (3), X-linked dominant | 99.97% |
| *IREB2* | 147582 | Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive | 99.9% |
| *IRF2BPL* | 611720 | Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant | 99.21% |
| *IRX5* | 606195 | Hamamy syndrome, 611174 (3), Autosomal recessive | 100% |
| *ISCA1* | 611006 | Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive | 99.79% |
| *ISCA2* | 615317 | Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive | 100% |
| *ITGA7* | 600536 | Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive | 99.87% |
| *ITPA* | 147520 | [Inosine triphosphatase deficiency], 613850 (3); Developmental and epileptic encephalopathy 35, 616647 (3), Autosomal recessive | 99.99% |

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| *ITPR1* | 147265 | Gillespie syndrome, 206700 (3), Autosomal recessive, Autosomal dominant; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant | 99.98% |
| ITSN1 | 602442 | No OMIM phenotype<br>ITSN1: a novel candidate gene involved in Autosomal dominant neurodevelopmental disorder spectrum (Bruel (2022) Eur J Hum Genet. 2022 Jan;30(1):111-116), PMID: 34707297 - Autosomal dominant | 99.89% |
| *IVD* | 607036 | Isovaleric acidemia, 243500 (3), Autosomal recessive | 100% |
| *JAG1* | 601920 | ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant | 100% |
| *JAM2* | 606870 | Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive | 91.82% |
| *JAM3* | 606871 | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive | 100% |
| *JARID2* | 601594 | Developmental delay with variable intellectual disability and dysmorphic facies, 620098 (3), Autosomal dominant | 99.97% |
| *KANK1* | 607704 | Cerebral palsy, spastic quadriplegic, 2, 612900 (3) | 99.99% |
| *KANSL1* | 612452 | Koolen-De Vries syndrome, 610443 (3), Autosomal dominant | 99.85% |
| *KARS1* | 601421 | Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive; Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 (3), Autosomal recessive | 99.98% |
| *KAT5* | 601409 | Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103 (3), Autosomal dominant | 100% |
| *KAT6A* | 601408 | Arboleda-Tham syndrome, 616268 (3), Autosomal dominant | 99.93% |
| *KAT6B* | 605880 | SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant | 99.79% |
| *KAT8* | 609912 | Li-Ghorgani-Weisz-Hubshman syndrome, 618974 (3), Autosomal dominant | 99.98% |
| KATNAL2 | 614697 | No OMIM phenotype<br>Autisme (Sanders (2012) Nature 4;485(7397):237-41 & O'Roak (2012) Nature 485(7397)). PMID: 22495309, 22495306 - Autosomal dominant | 99.98% |
| *KATNB1* | 602703 | Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive | 99.99% |
| *KATNIP* | 616650 | Joubert syndrome 26, 616784 (3), Autosomal recessive | 99.13% |

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| *KCNA1* | 176260 | Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant | 100% |
| *KCNA2* | 176262 | Developmental and epileptic encephalopathy 32, 616366 (3), Autosomal dominant | 99.99% |
| *KCNA4* | 176266 | Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive | 100% |
| *KCNB1* | 600397 | Developmental and epileptic encephalopathy 26, 616056 (3), Autosomal dominant | 100% |
| *KCNC1* | 176258 | Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant | 100% |
| *KCNC2* | 176256 | Developmental and epileptic encephalopathy 103, 619913 (3), Autosomal dominant | 99.88% |
| *KCNC3* | 176264 | Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant | 99.98% |
| KCND2 | 605410 | No OMIM phenotype<br>KCND2 variants associated with global developmental delay differentially impair Kv4.2 channel gating (Zhang (2021), Hum Mol Genet. 30(23):2300-2314), PMID: 34245260 - Autosomal dominant | 99.9% |
| *KCND3* | 605411 | Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant; Brugada syndrome 9, 616399 (3), Autosomal dominant | 99.98% |
| *KCNH1* | 603305 | Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant; Temple-Baraitser syndrome, 611816 (3), Autosomal dominant | 99.92% |
| KCNH5 | 605716 | No OMIM phenotype<br>Neurodevelopmental and Epilepsy Phenotypes in Individuals With Missense Variants in the Voltage-Sensing and Pore Domains of KCNH5 (Happ (2023) Neurology. 2023 Feb 7;100(6):e603-e615), PMID: 36307226 - Autosomal dominant | 99.89% |
| *KCNJ10* | 602208 | Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive | 99.98% |
| *KCNJ11* | 600937 | Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive, Autosomal dominant | 100% |
| *KCNJ6* | 600877 | Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant | 100% |
| *KCNK4* | 605720 | Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381 (3), Autosomal dominant | 99.99% |
| *KCNK9* | 605874 | Birk-Barel syndrome, 612292 (3) | 100% |
| *KCNMA1* | 600150 | {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), | 99.89% |

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| | | Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Liang-Wang syndrome, 618729 (3), Autosomal dominant | |
| *KCNN2* | 605879 | ?Dystonia 34, myoclonic, 619724 (3), Autosomal dominant; Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 (3), Autosomal dominant | 91.25% |
| *KCNN3* | 602983 | Zimmermann-Laband syndrome 3, 618658 (3), Autosomal dominant | 99.97% |
| *KCNQ2* | 602235 | Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant | 100% |
| *KCNQ3* | 602232 | Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant | 99.98% |
| *KCNQ5* | 607357 | Intellectual developmental disorder, autosomal dominant 46, 617601 (3), Autosomal dominant | 99.89% |
| *KCNT1* | 608167 | Developmental and epileptic encephalopathy 14, 614959 (3), Autosomal dominant; Epilepsy nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant | 99.98% |
| *KCNT2* | 610044 | Developmental and epileptic encephalopathy 57, 617771 (3), Autosomal dominant | 98.77% |
| KCTD3 | 613272 | No OMIM phenotype<br>Neurodevelopmental and neuropsychiatric disorders (Teng (2019), Review > CNS Neurosci Ther ;25(7):887-902), PMID: 31197948 - Autosomal recessive | 99.75% |
| *KCTD7* | 611725 | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive | 99.98% |
| *KDM1A* | 609132 | Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant | 89.42% |
| *KDM3B* | 609373 | Diets-Jongmans syndrome, 618846 (3), Autosomal dominant | 99.98% |
| *KDM4B* | 609765 | Intellectual developmental disorder, autosomal dominant 65, 619320 (3), Autosomal dominant | 99.99% |
| KDM5A | 180202 | No OMIM phenotype<br>KDM5A mutations identified in autism spectrum disorder using forward genetics (Hayek (2020) Elife. 2020 Dec 22;9:e56883), PMID: 33350388 - Autosomal dominant | 99.9% |
| *KDM5B* | 605393 | Intellectual developmental disorder, autosomal recessive 65, 618109 (3), Autosomal recessive | 97.15% |
| *KDM5C* | 314690 | Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 (3), X-linked recessive | 99.98% |
| *KDM6A* | 300128 | Kabuki syndrome 2, 300867 (3), X-linked dominant | 99.74% |
| *KDM6B* | 611577 | Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505 (3), Autosomal dominant | 99.99% |
| *KIAA0586* | 610178 | Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive | 95.75% |

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| *KIDINS220* | 615759 | Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant; Ventriculomegaly and arthrogryposis, 619501 (3), Autosomal recessive | 99.94% |
| *KIF11* | 148760 | Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant | 99.84% |
| *KIF14* | 611279 | Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive | 97.8% |
| *KIF1A* | 601255 | NESCAV syndrome, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal recessive, Autosomal dominant; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal recessive, Autosomal dominant | 99.96% |
| KIF21B | 608322 | No OMIM phenotype<br>Mutations in the KIF21B kinesin gene cause neurodevelopmental disorders through imbalanced canonical motor activity (Asselin (2020), Nat Commun. 11(1):2441), PMID: 32415109 - Autosomal dominant | 99.83% |
| *KIF26A* | 613231 | Cortical dysplasia, complex, with other brain malformations 11, 620156 (3), Autosomal recessive | 100% |
| *KIF2A* | 602591 | Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant | 99.79% |
| *KIF4A* | 300521 | ?Intellectual developmental disorder, X-linked 100, 300923 (3), X-linked recessive | 99.76% |
| *KIF5A* | 602821 | Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant | 99.91% |
| *KIF5C* | 604593 | Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant | 99.93% |
| *KIF7* | 611254 | Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive | 100% |
| *KIFBP* | 609367 | Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive | 99.91% |
| KLF7 | 604865 | No OMIM phenotype<br>Developmental delay/intellectual disability, neuromuscular and psychiatric symptoms (Powis (2018), Clin Genet 93(5)), PMID: 29251763 - Autosomal dominant | 99.99% |
| *KLHL15* | 300980 | Intellectual developmental disorder, X-linked 103, 300982 (3), X-linked recessive | 100% |
| KLHL20 | 617679 | No OMIM phenotype<br>De novo missense variants in the E3 ubiquitin ligase adaptor KLHL20 cause a developmental disorder with intellectual disability, epilepsy, and autism spectrum disorder (Sleyp (2022) Genet Med. 2022 Dec;24(12):2464-2474), PMID: 36214804 - Autosomal dominant | 99.51% |

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| *KLHL7* | 611119 | Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive | 99.95% |
| *KMT2A* | 159555 | Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant | 99.97% |
| *KMT2B* | 606834 | Intellectual developmental disorder, autosomal dominant 68, 619934 (3), Autosomal dominant; Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant | 99.99% |
| *KMT2C* | 606833 | Kleefstra syndrome 2, 617768 (3), Autosomal dominant | 98.98% |
| *KMT2D* | 602113 | Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 (3), Autosomal dominant; Kabuki syndrome 1, 147920 (3), Autosomal dominant | 99.98% |
| *KMT2E* | 608444 | O'Donnell-Luria-Rodan syndrome, 618512 (3), Autosomal dominant | 99.92% |
| *KMT5B* | 610881 | Intellectual developmental disorder, autosomal dominant 51, 617788 (3), Autosomal dominant | 99.91% |
| *KNL1* | 609173 | Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive | 98.43% |
| *KPTN* | 615620 | Intellectual developmental disorder, autosomal recessive 41, 615637 (3), Autosomal recessive | 99.98% |
| *KRAS* | 190070 | Gastric cancer, somatic, 613659 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3) | 99.13% |
| *KRIT1* | 604214 | Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 (3), Autosomal dominant; Cerebral cavernous malformations-1, 116860 (3), Autosomal dominant; Cavernous malformations of CNS and retina, 116860 (3), Autosomal dominant | 99.33% |
| *L1CAM* | 308840 | MASA syndrome, 303350 (3), X-linked recessive; Hydrocephalus, congenital, X-linked, 307000 (3), X-linked recessive; ?Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive | 99.98% |
| *L2HGDH* | 609584 | L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive | 99.92% |
| *LAMA1* | 150320 | Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive | 99.98% |
| *LAMA2* | 156225 | Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive | 99.95% |
| *LAMB1* | 150240 | Lissencephaly 5, 615191 (3), Autosomal recessive | 99.87% |
| *LAMC3* | 604349 | Cortical malformations, occipital, 614115 (3), Autosomal recessive | 99.97% |
| *LAMP2* | 309060 | Danon disease, 300257 (3), X-linked dominant | 98.95% |

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| *LARGE1* | 603590 | Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive | 100% |
| *LARP7* | 612026 | Alazami syndrome, 615071 (3), Autosomal recessive | 99.63% |
| *LARS1* | 151350 | ?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive | 99.87% |
| *LAS1L* | 300964 | Wilson-Turner syndrome, 309585 (3), X-linked recessive | 99.97% |
| *LETM1* | 604407 | Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 (3), Autosomal recessive | 99.97% |
| *LGI1* | 604619 | Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant | 99.99% |
| *LGI4* | 608303 | Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive | 99.98% |
| LHX2 | 603759 | No OMIM phenotype<br>LHX2 haploinsufficiency causes a variable neurodevelopmental disorder (Schmid (2023) Genet Med. 2023 Jul;25(7):100839), PMID: 37057675 - Autosomal dominant | 100% |
| *LIAS* | 607031 | Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive | 99.98% |
| *LIG3* | 600940 | Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive | 99.99% |
| *LIG4* | 601837 | LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation | 100% |
| *LINGO1* | 609791 | Intellectual developmental disorder, autosomal recessive 64, 618103 (3), Autosomal recessive | 100% |
| *LINS1* | 610350 | Intellectual developmental disorder, autosomal recessive 27, 614340 (3), Autosomal recessive | 99.96% |
| *LIPT1* | 610284 | Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive | 99.89% |
| *LMAN2L* | 609552 | ?Intellectual developmental disorder, autosomal dominant 69, 617863 (3); ?Intellectual developmental disorder, autosomal recessive 52, 616887 (3), Autosomal recessive | 99.91% |
| *LMBRD1* | 612625 | Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive | 99.67% |
| *LMBRD2* | 619490 | Developmental delay with variable neurologic and brain abnormalities, 619694 (3), Autosomal dominant | 99.66% |
| *LMNB1* | 150340 | Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant; Microcephaly 26, primary, autosomal dominant, 619179 (3), Autosomal dominant | 99.73% |
| *LMNB2* | 150341 | Microcephaly 27, primary, autosomal dominant, 619180 (3), Autosomal dominant; ?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant | 99.99% |
| *LNPK* | 610236 | Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090 (3), Autosomal recessive | 92.81% |

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| *LONP1* | 605490 | CODAS syndrome, 600373 (3), Autosomal recessive | 99.99% |
| *LRP2* | 600073 | Donnai-Barrow syndrome, 222448 (3), Autosomal recessive | 99.86% |
| *LRPPRC* | 607544 | Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive | 99.8% |
| *LSS* | 600909 | Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-intellectual disability syndrome 4, 618840 (3), Autosomal recessive | 99.98% |
| *LYRM7* | 615831 | Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive | 99.98% |
| *LYST* | 606897 | Chediak-Higashi syndrome, 214500 (3), Autosomal recessive | 99.87% |
| *LZTFL1* | 606568 | Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive | 100% |
| *LZTR1* | 600574 | Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant | 99.46% |
| *MAB21L1* | 601280 | Cerebellar, ocular, craniofacial, and genital syndrome, 618479 (3), Autosomal recessive | 100% |
| *MAB21L2* | 604357 | Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant | 100% |
| *MACF1* | 608271 | Lissencephaly 9 with complex brainstem malformation, 618325 (3), Autosomal dominant | 99.33% |
| *MADD* | 603584 | Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 (3), Autosomal recessive; DEEAH syndrome, 619004 (3), Autosomal recessive | 99.95% |
| *MAF* | 177075 | Cataract 21, multiple types, 610202 (3), Autosomal dominant; Ayme-Gripp syndrome, 601088 (3), Autosomal dominant | 99.73% |
| *MAG* | 159460 | Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive | 99.99% |
| *MAGEL2* | 605283 | Schaaf-Yang syndrome, 615547 (3), Autosomal dominant | 99.99% |
| *MAGT1* | 300715 | Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3), X-linked recessive; Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive | 99.54% |
| *MAN1B1* | 604346 | Rafiq syndrome, 614202 (3), Autosomal recessive | 99.94% |
| *MAN2B1* | 609458 | Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive | 99.99% |
| *MAN2C1* | 154580 | Congenital disorder of deglycosylation 2, 619775 (3), Autosomal recessive | 99.96% |
| *MANBA* | 609489 | Mannosidosis, beta, 248510 (3), Autosomal recessive | 99.81% |
| *MAOA* | 309850 | {Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive | 99.83% |
| *MAP1B* | 157129 | ?Deafness, autosomal dominant 83, 619808 (3), Autosomal dominant; Periventricular nodular heterotopia 9, 618918 (3), Autosomal dominant | 100% |

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| *MAP2K1* | 176872 | Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3) | 99.98% |
| *MAP2K2* | 601263 | Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant | 99.99% |
| *MAPK1* | 176948 | Noonan syndrome 13, 619087 (3), Autosomal dominant | 99.84% |
| *MAPK8IP3* | 605431 | Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant | 100% |
| *MAPKAPK5* | 606723 | Neurocardiofaciodigital syndrome, 619869 (3), Autosomal recessive | 99.95% |
| *MAPRE2* | 605789 | Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant | 99.96% |
| *MARCHF6* | 613297 | Epilepsy, familial adult myoclonic, 3, 613608 (3), Autosomal dominant | 99.97% |
| *MASP1* | 600521 | 3MC syndrome 1, 257920 (3), Autosomal recessive | 99.99% |
| *MAST1* | 612256 | Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 (3), Autosomal dominant | 100% |
| *MAST3* | 612258 | Developmental and epileptic encephalopathy 108, 620115 (3), Autosomal dominant | 97.04% |
| MAST4 | 618002 | No OMIM phenotype<br>De novo variants in MAST4 related to neurodevelopmental disorders with developmental delay and infantile spasms: Genotype-phenotype association (Zhang (2023) Front Mol Neurosci. 2023 Feb 22;16:1097553), PMID: 36910266 - Autosomal dominant | 99.98% |
| *MAT1A* | 610550 | Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal recessive, Autosomal dominant; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal recessive, Autosomal dominant | 99.7% |
| *MBD5* | 611472 | Intellectual developmental disorder, autosomal dominant 1, 156200 (3), Autosomal dominant | 99.79% |
| *MBOAT7* | 606048 | Intellectual developmental disorder, autosomal recessive 57, 617188 (3), Autosomal recessive | 100% |
| *MBTPS2* | 300294 | Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive | 99.81% |
| *MCCC1* | 609010 | 3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive | 99.86% |
| *MCCC2* | 609014 | 3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive | 99.97% |
| *MCM3AP* | 603294 | Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive | 99.98% |
| *MCOLN1* | 605248 | Mucolipidosis IV, 252650 (3), Autosomal recessive | 100% |
| *MCPH1* | 607117 | Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive | 100% |

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| *MDH1* | 154200 | ?Developmental and epileptic encephalopathy 88, 618959 (3), Autosomal recessive | 99.96% |
| *MDH2* | 154100 | Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive | 99.54% |
| *MECP2* | 300005 | Rett syndrome, atypical, 312750 (3), X-linked dominant; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Intellectual developmental disorder, X-linked syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant | 99.95% |
| *MECR* | 608205 | Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive | 99.63% |
| *MED11* | 612383 | Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327 (3), Autosomal recessive | 99.99% |
| *MED12* | 300188 | Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Hardikar syndrome, 301068 (3), X-linked dominant; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive | 99.94% |
| *MED12L* | 611318 | Nizon-Isidor syndrome, 618872 (3), Autosomal dominant | 99.95% |
| *MED13* | 603808 | Intellectual developmental disorder, autosomal dominant 61, 618009 (3), Autosomal dominant | 99.55% |
| *MED13L* | 608771 | Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant | 99.99% |
| *MED17* | 603810 | Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive | 99.82% |
| *MED23* | 605042 | Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249 (3), Autosomal recessive | 99.82% |
| *MED25* | 610197 | Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive | 99.95% |
| *MED27* | 605044 | Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 (3), Autosomal recessive | 99.99% |
| *MEF2C* | 600662 | Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443 (3), Autosomal dominant | 99.57% |
| *MEGF8* | 604267 | Carpenter syndrome 2, 614976 (3), Autosomal recessive | 99.9% |
| *MEIS2* | 601740 | Cleft palate, cardiac defects, and mental retardation, 600987 (3), Autosomal dominant | 99.97% |
| *METTL23* | 615262 | Intellectual developmental disorder, autosomal recessive 44, 615942 (3), Autosomal recessive | 100% |
| *METTL5* | 618628 | Intellectual developmental disorder, autosomal recessive 72, 618665 (3), Autosomal recessive | 99.75% |

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| *MFF* | 614785 | Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive | 99.97% |
| *MFSD2A* | 614397 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 (3), Autosomal recessive | 99.78% |
| *MFSD8* | 611124 | Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive | 99.7% |
| *MGAT2* | 602616 | Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive | 100% |
| *MGP* | 154870 | Keutel syndrome, 245150 (3), Autosomal recessive | 99.95% |
| MICAL1 | 607129 | No OMIM phenotype<br>Mutations in MICAL-1cause Autosomal-dominant lateral temporal epilepsy (Dazzo (2018), Ann Neurol 83(3):483-493), PMID: 29394500 - Autosomal dominant | 99.99% |
| *MICU1* | 605084 | Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive | 99.56% |
| *MID1* | 300552 | Opitz GBBB syndrome, 300000 (3), X-linked recessive | 99.9% |
| *MINPP1* | 605391 | {Thyroid carcinoma, follicular}, 188470 (3), Somatic mutation, Autosomal dominant; Pontocerebellar hypoplasia, type 16, 619527 (3), Autosomal recessive | 99.73% |
| *MKKS* | 604896 | McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive | 100% |
| *MKS1* | 609883 | Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive | 99.92% |
| *MLC1* | 605908 | Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004 (3), Autosomal recessive | 99.99% |
| *MLYCD* | 606761 | Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive | 99.95% |
| *MMAA* | 607481 | Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 (3), Autosomal recessive | 99.95% |
| *MMAB* | 607568 | Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 (3), Autosomal recessive | 99.99% |
| *MMACHC* | 609831 | Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive | 99.98% |
| *MMADHC* | 611935 | Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive | 99.76% |
| *MMUT* | 609058 | Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive | 99.68% |
| *MN1* | 156100 | CEBALID syndrome, 618774 (3), Autosomal dominant; Meningioma, 607174 (3), Autosomal dominant | 99.98% |
| *MOCS1* | 603707 | Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive | 99.95% |
| *MOCS2* | 603708 | Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive | 99.96% |
| *MOGS* | 601336 | Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive | 100% |

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| *MORC2* | 616661 | Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant; Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 (3), Autosomal dominant | 100% |
| *MPDU1* | 604041 | Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive | 99.97% |
| *MPDZ* | 603785 | Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive | 99.86% |
| *MPLKIP* | 609188 | Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive | 99.99% |
| *MRAS* | 608435 | Noonan syndrome 11, 618499 (3), Autosomal dominant | 99.97% |
| *MRPL3* | 607118 | Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive | 99.94% |
| *MRPS22* | 605810 | Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive | 99.87% |
| *MRPS34* | 611994 | Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive | 100% |
| *MSL3* | 300609 | Basilicata-Akhtar syndrome, 301032 (3), X-linked dominant | 99.83% |
| *MSMO1* | 607545 | Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive | 99.88% |
| *MTFMT* | 611766 | Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive | 99.98% |
| *MTHFR* | 607093 | {Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive | 99.97% |
| *MTHFS* | 604197 | Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 (3), Autosomal recessive | 100% |
| *MTO1* | 614667 | Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive | 90.25% |
| *MTOR* | 601231 | Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant | 99.98% |
| *MTR* | 156570 | {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive | 99.95% |
| *MTRFR* | 613541 | Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive | 99.87% |
| *MTRR* | 602568 | Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive | 99.98% |

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| *MTSS2* | 616951 | Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086 (3), Autosomal dominant | 100% |
| *MVK* | 251170 | Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive | 99.97% |
| MYCBP2 | 610392 | No OMIM phenotype<br>Loss-of-function variants in MYCBP2 cause neurobehavioural phenotypes and corpus callosum defects (AlAbdi (2023) Brain. 2023 Apr 19;146(4):1373-1387), PMID: 36200388 - Autosomal dominant | 99.88% |
| *MYCN* | 164840 | Feingold syndrome 1, 164280 (3), Autosomal dominant | 100% |
| *MYH10* | 160776 | Neurodevelopmental disorders and variable congenital anomalies & fetal abnormalities (PMID:Â 35980381, 30712878) | 99.91% |
| *MYO5A* | 160777 | Griscelli syndrome, type 1, 214450 (3), Autosomal recessive | 99.94% |
| *MYT1L* | 613084 | Intellectual developmental disorder, autosomal dominant 39, 616521 (3), Autosomal dominant | 99.99% |
| *NAA10* | 300013 | Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant | 99.99% |
| *NAA15* | 608000 | Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 (3), Autosomal dominant | 99.71% |
| *NAA20* | 610833 | Intellectual developmental disorder, autosomal recessive 73, 619717 (3), Autosomal recessive | 99.93% |
| *NACC1* | 610672 | Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant | 99.99% |
| *NAE1* | 603385 | Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210 (3), Autosomal recessive | 99.81% |
| *NAGA* | 104170 | Schindler disease, type I, 609241 (3), Autosomal recessive; Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive | 100% |
| *NAGLU* | 609701 | ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive | 100% |
| *NAGS* | 608300 | N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive | 99.99% |
| *NALCN* | 611549 | Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant; Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive | 99.97% |
| *NANS* | 605202 | Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive | 100% |
| *NAPB* | 611270 | Developmental and epileptic encephalopathy 107, 620033 (3), Autosomal recessive | 100% |

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| *NARS1* | 108410 | Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 (3), Autosomal dominant; Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091 (3), Autosomal recessive | 99.94% |
| *NARS2* | 612803 | Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive; ?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive | 99.59% |
| *NAXD* | 615910 | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive | 99.99% |
| *NAXE* | 608862 | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive | 99.99% |
| *NBEA* | 604889 | Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157 (3), Autosomal dominant | 99.98% |
| *NBN* | 602667 | Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive | 99.93% |
| *NCAPG2* | 608532 | Khan-Khan-Katsanis syndrome, 618460 (3), Autosomal recessive | 99.97% |
| *NCDN* | 608458 | Neurodevelopmental disorder with infantile epileptic spasms, 619373 (3), Autosomal dominant | 99.99% |
| NCKAP1 | 604891 | No OMIM phenotype<br>NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism (Guo (2020), Am J Hum Genet. 107(5):963-976), PMID: 33157009 - Autosomal dominant | 99.62% |
| NCOR1 | 600849 | No OMIM phenotype<br>Autism spectrum disorder, scoliosis, and abnormal palatogenesis (Sakaguchi (2018), case report > Am J Med Genet A 176(11):2466-2469) - Autosomal dominant | 99.88% |
| *NDE1* | 609449 | Microhydranencephaly, 605013 (3), Autosomal recessive; Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive | 100% |
| *NDP* | 300658 | Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive | 99.98% |
| *NDST1* | 600853 | Intellectual developmental disorder, autosomal recessive 46, 616116 (3), Autosomal recessive | 100% |
| *NDUFA1* | 300078 | Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive | 99.93% |
| *NDUFA10* | 603835 | Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive | 99.98% |
| *NDUFA11* | 612638 | Mitochondrial complex I deficiency, nuclear type 14, 618236 (3), Autosomal recessive | 98.22% |
| *NDUFA12* | 614530 | Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive | 99.21% |
| *NDUFA2* | 602137 | Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive | 99.95% |
| *NDUFA6* | 602138 | Mitochondrial complex I deficiency, nuclear type 33, 618253 (3), Autosomal recessive | 99.96% |

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| *NDUFA8* | 603359 | Mitochondrial complex I deficiency, nuclear type 37, 619272 (3), Autosomal recessive | 100% |
| *NDUFAF1* | 606934 | Mitochondrial complex I deficiency, nuclear type 11, 618234 (3), Autosomal recessive | 100% |
| *NDUFAF2* | 609653 | Mitochondrial complex I deficiency, nuclear type 10, 618233 (3), Autosomal recessive | 99.88% |
| *NDUFAF3* | 612911 | Mitochondrial complex I deficiency, nuclear type 18, 618240 (3), Autosomal recessive | 100% |
| *NDUFAF4* | 611776 | Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive | 99.95% |
| *NDUFAF5* | 612360 | Mitochondrial complex I deficiency, nuclear type 16, 618238 (3), Autosomal recessive | 99.89% |
| *NDUFAF8* | 618461 | Mitochondrial complex I deficiency, nuclear type 34, 618776 (3), Autosomal recessive | 99.98% |
| *NDUFB3* | 603839 | Mitochondrial complex I deficiency, nuclear type 25, 618246 (3), Autosomal recessive | 99.6% |
| *NDUFB9* | 601445 | ?Mitochondrial complex I deficiency, nuclear type 24, 618245 (3), Autosomal recessive | 100% |
| *NDUFS1* | 157655 | Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive | 99.79% |
| *NDUFS2* | 602985 | Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive | 99.66% |
| *NDUFS3* | 603846 | Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive | 100% |
| *NDUFS4* | 602694 | Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive | 99.99% |
| *NDUFS6* | 603848 | Mitochondrial complex I deficiency, nuclear type 9, 618232 (3), Autosomal recessive | 100% |
| *NDUFS7* | 601825 | Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive | 99.99% |
| *NDUFS8* | 602141 | Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive | 100% |
| *NDUFV1* | 161015 | Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive | 99.99% |
| *NDUFV2* | 600532 | Mitochondrial complex I deficiency, nuclear type 7, 618229 (3), Autosomal recessive | 99.98% |
| *NECAP1* | 611623 | Developmental and epileptic encephalopathy 21, 615833 (3), Autosomal recessive | 100% |
| *NECTIN1* | 600644 | Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive | 99.99% |
| *NEDD4L* | 606384 | Periventricular nodular heterotopia 7, 617201 (3), Autosomal dominant | 99.97% |
| *NEMF* | 608378 | Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099 (3), Autosomal recessive | 99.94% |
| *NEU1* | 608272 | Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive | 99.98% |
| *NEUROD2* | 601725 | Developmental and epileptic encephalopathy 72, 618374 (3), Autosomal dominant | 100% |
| *NEXMIF* | 300524 | Intellectual developmental disorder, X-linked 98, 300912 (3), X-linked dominant | 99.99% |
| *NF1* | 613113 | Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant | 99.88% |

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| *NFASC* | 609145 | Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 (3), Autosomal recessive | 99.94% |
| *NFE2L2* | 600492 | Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant | 99.97% |
| *NFIA* | 600727 | Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant | 97.55% |
| *NFIB* | 600728 | Macrocephaly, acquired, with impaired intellectual development, 618286 (3), Autosomal dominant | 99.9% |
| *NFIX* | 164005 | Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Malan syndrome, 614753 (3), Autosomal dominant | 99.99% |
| *NFU1* | 608100 | Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive | 99.48% |
| *NGLY1* | 610661 | Congenital disorder of deglycosylation 1, 615273 (3), Autosomal recessive | 99.93% |
| *NHLRC1* | 608072 | Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive | 100% |
| *NHS* | 300457 | Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant | 99.96% |
| *NIPBL* | 608667 | Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant | 99.34% |
| *NKAP* | 300766 | Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039 (3), X-linked recessive | 99.1% |
| *NKX2-1* | 600635 | Chorea, hereditary benign, 118700 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant | 100% |
| *NLGN3* | 300336 | {Autism susceptibility, X-linked 1}, 300425 (3), X-linked | 99.98% |
| *NLGN4X* | 300427 | Intellectual developmental disorder, X-linked, 300495 (3), X-linked; {Autism susceptibility, X-linked 2}, 300495 (3), X-linked | 99.98% |
| *NLRP3* | 606416 | CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant | 100% |
| *NONO* | 300084 | Intellectual developmental disorder, X-linked syndromic 34, 300967 (3), X-linked | 99.94% |
| *NOVA2* | 601991 | Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 (3), Autosomal dominant | 99.95% |
| *NPC1* | 607623 | Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive | 99.99% |
| *NPC2* | 601015 | Niemann-pick disease, type C2, 607625 (3), Autosomal recessive | 100% |
| *NPHP1* | 607100 | Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive | 99.05% |

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| *NPRL2* | 607072 | Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant | 100% |
| *NPRL3* | 600928 | Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant | 99.99% |
| *NR2F1* | 132890 | Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant | 99.99% |
| *NR4A2* | 601828 | Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911 (3), Autosomal dominant | 99.97% |
| *NRAS* | 164790 | Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3) | 99.66% |
| *NRCAM* | 601581 | Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833 (3), Autosomal recessive | 99.82% |
| *NRROS* | 615322 | Seizures, early-onset, with neurodegeneration and brain calcification, 618875 (3), Autosomal recessive | 100% |
| *NRXN1* | 600565 | Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3) | 99.98% |
| *NSD1* | 606681 | Sotos syndrome, 117550 (3), Autosomal dominant | 99.98% |
| *NSD2* | 602952 | Rauch-Steindl syndrome, 619695 (3), Autosomal dominant | 99.87% |
| *NSDHL* | 300275 | CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant | 99.87% |
| *NSF* | 601633 | Developmental and epileptic encephalopathy 96, 619340 (3), Autosomal dominant | 52.85% |
| *NSRP1* | 616173 | Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001 (3), Autosomal recessive | 99.96% |
| *NSUN2* | 610916 | Intellectual developmental disorder, autosomal recessive 5, 611091 (3), Autosomal recessive | 99.96% |
| NSUN6 | 617199 | No OMIM phenotype<br>Biallelic variants in NSUN6 cause an Autosomal recessive neurodevelopmental disorder (Mattioli (2023) Genet Med. 2023 Sep;25(9):100900), PMID: 37226891 - Autosomal recessive | 99.95% |
| *NT5C2* | 600417 | Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive | 99.96% |
| *NTNG2* | 618689 | Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive | 99.98% |
| *NTRK1* | 191315 | Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive | 99.86% |
| *NTRK2* | 600456 | Developmental and epileptic encephalopathy 58, 617830 (3), Autosomal dominant; Obesity, hyperphagia, and developmental delay, 613886 (3), Autosomal dominant | 99.9% |

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| *NUBPL* | 613621 | Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive | 99.62% |
| *NUDT2* | 602852 | Intellectual developmental disorder with or without peripheral neuropathy, 619844 (3), Autosomal recessive | 100% |
| *NUP107* | 607617 | ?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive | 97.46% |
| *NUP188* | 615587 | Sandestig-Stefanova syndrome, 618804 (3), Autosomal recessive | 99.88% |
| *NUP214* | 114350 | Leukemia, T-cell acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, somatic, 601626 (3); {Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 (3), Autosomal recessive | 99.98% |
| *NUP62* | 605815 | Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive | 100% |
| *NUS1* | 610463 | Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive | 99.9% |
| *OAT* | 613349 | Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive | 90.17% |
| *OCLN* | 602876 | Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive | 82.91% |
| *OCRL* | 300535 | Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive | 99.89% |
| *ODC1* | 165640 | Bachmann-Bupp syndrome, 619075 (3), Autosomal dominant | 99.99% |
| *OFD1* | 300170 | Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive | 99.68% |
| *OGDH* | 613022 | Oxoglutarate dehydrogenase deficiency, 203740 (3), Autosomal recessive | 99.93% |
| *OGDHL* | 617513 | Yoon-Bellen neurodevelopmental syndrome, 619701 (3), Autosomal recessive | 99.95% |
| *OGT* | 300255 | Intellectual developmental disorder, X-linked 106, 300997 (3), X-linked recessive | 99.79% |
| *OPHN1* | 300127 | Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 (3), X-linked recessive | 99.92% |
| *ORC1* | 601902 | Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive | 99.7% |
| *OSGEP* | 610107 | Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive | 100% |
| *OTC* | 300461 | Ornithine transcarbamylase deficiency, 311250 (3), X-linked | 99.42% |
| *OTUD5* | 300713 | Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056 (3), X-linked recessive | 99.93% |
| *OTUD6B* | 612021 | Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive | 99.92% |

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| OTUD7A | 612024 | No OMIM phenotype<br>Biallelic loss of OTUD7A causes severe muscular hypotonia, intellectual disability, and seizures (Suzuki (2020) Am J Med Genet A. 2021 Apr;185(4):1182-1186), PMID: 33381903 - Autosomal recessive | 99.04% |
| *OTX2* | 600037 | Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant | 100% |
| *OXR1* | 605609 | Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 (3), Autosomal recessive | 99.96% |
| *P4HTM* | 614584 | Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 (3), Autosomal recessive | 100% |
| PABPC1 | 604679 | No OMIM phenotype<br>De novo variants in the PABP domain of PABPC1 lead to developmental delay (Wegler (2022), Genet Med. 24(8):1761-1773), PMID: 35511136 - Autosomal dominant | 99.84% |
| *PACS1* | 607492 | Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant | 99.96% |
| *PACS2* | 610423 | Developmental and epileptic encephalopathy 66, 618067 (3), Autosomal dominant | 99.99% |
| *PAFAH1B1* | 601545 | Subcortical laminar heterotopia, 607432 (3), Autosomal dominant; Lissencephaly 1, 607432 (3), Autosomal dominant | 99.96% |
| *PAH* | 612349 | [Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive | 99.96% |
| *PAK1* | 602590 | Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158 (3), Autosomal dominant | 99.99% |
| *PAK3* | 300142 | Intellectual developmental disorder, X-linked 30, 300558 (3), X-linked recessive | 92.53% |
| PALS1 | 606958 | No OMIM phenotype<br>De novo variants in MPP5 cause global developmental delay and behavioral changes (Sterling (2020) Hum Mol Genet. 2020 Dec 18;29(20):3388-3401), PMID: 33073849 - Autosomal dominant | 99.82% |
| PAN2 | 617447 | No OMIM phenotype<br>Biallelic PAN2 variants in individuals with a syndromic neurodevelopmental disorder and multiple congenital anomalies (Reuter (2022) Eur J Hum Genet. 2022 May;30(5):611-618), PMID: 35304602 - Autosomal recessive | 99.95% |
| *PANK2* | 606157 | HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive | 99.99% |

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| *PARN* | 604212 | Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371 (3), Autosomal dominant | 99.75% |
| PARP6 | 619439 | No OMIM phenotype<br>Characterization of PARP6 Function in Knockout Mice and Patients with Developmental Delay (Vermehren-Schmaedick (2021), Cells. 10(6):1289), PMID: 34067418 - Autosomal dominant | 99.97% |
| *PARS2* | 612036 | Developmental and epileptic encephalopathy 75, 618437 (3), Autosomal recessive | 99.99% |
| *PAX1* | 167411 | Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive | 100% |
| *PAX5* | 167414 | {Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3) | 99.82% |
| *PAX6* | 607108 | Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant | 99.95% |
| *PAX8* | 167415 | Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant | 99.99% |
| *PBX1* | 176310 | Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant | 99.88% |
| *PC* | 608786 | Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive | 99.99% |
| *PCCA* | 232000 | Propionicacidemia, 606054 (3), Autosomal recessive | 99.9% |
| *PCCB* | 232050 | Propionicacidemia, 606054 (3), Autosomal recessive | 99.97% |
| *PCDH12* | 605622 | Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive | 100% |
| *PCDH19* | 300460 | Developmental and epileptic encephalopathy 9, 300088 (3), X-linked | 99.98% |
| *PCDHGC4* | 606305 | Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880 (3), Autosomal recessive | 100% |
| *PCGF2* | 600346 | Turnpenny-Fry syndrome, 618371 (3), Autosomal dominant | 99.78% |
| *PCLO* | 604918 | ?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive | 99.41% |
| *PCNT* | 605925 | Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive | 99.97% |
| *PCYT2* | 602679 | Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive | 100% |
| *PDCD10* | 609118 | Cerebral cavernous malformations-3, 603285 (3), Autosomal dominant | 99.94% |
| *PDE10A* | 610652 | Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant; Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive | 87.37% |

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| *PDE2A* | 602658 | Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 (3), Autosomal recessive | 99.95% |
| *PDE4D* | 600129 | Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant | 99.89% |
| *PDE6D* | 602676 | Joubert syndrome 22, 615665 (3), Autosomal recessive | 99.94% |
| *PDGFRB* | 173410 | Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant | 99.99% |
| *PDHA1* | 300502 | Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant | 99.04% |
| *PDHB* | 179060 | Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive | 99.94% |
| *PDHX* | 608769 | Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive | 99.64% |
| *PDP1* | 605993 | Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive | 100% |
| *PDSS1* | 607429 | Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive | 95.7% |
| *PDSS2* | 610564 | Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive | 99.87% |
| *PDX1* | 600733 | {Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; Pancreatic agenesis 1, 260370 (3), Autosomal recessive; MODY, type IV, 606392 (3) | 100% |
| *PDZD8* | 614235 | Intellectual developmental disorder with autism and dysmorphic facies, 620021 (3), Autosomal recessive | 99.99% |
| *PEPD* | 613230 | Prolidase deficiency, 170100 (3), Autosomal recessive | 99.98% |
| *PET100* | 614770 | Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive | 99.98% |
| *PEX1* | 602136 | Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive | 98.8% |
| *PEX10* | 602859 | Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive | 100% |
| *PEX11B* | 603867 | Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive | 99.62% |
| *PEX12* | 601758 | Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive | 100% |
| *PEX13* | 601789 | Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive | 99.36% |
| *PEX14* | 601791 | Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive | 100% |
| *PEX16* | 603360 | Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive | 99.94% |

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| *PEX19* | 600279 | Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive | 99.25% |
| *PEX2* | 170993 | Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive | 100% |
| *PEX26* | 608666 | Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive | 100% |
| *PEX3* | 603164 | Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive | 99.85% |
| *PEX5* | 600414 | Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive | 99.89% |
| *PEX6* | 601498 | Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive | 99.99% |
| *PEX7* | 601757 | Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive | 99.72% |
| *PGAP1* | 611655 | Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 (3), Autosomal recessive | 99.56% |
| *PGAP2* | 615187 | Hyperphosphatasia with impaired intellectual development syndrome 3, 614207 (3), Autosomal recessive | 99.99% |
| *PGAP3* | 611801 | Hyperphosphatasia with impaired intellectual development syndrome 4, 615716 (3), Autosomal recessive | 99.97% |
| *PGK1* | 311800 | Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive | 99.93% |
| *PGM2L1* | 611610 | Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191 (3), Autosomal recessive | 99.82% |
| *PGM3* | 172100 | Immunodeficiency 23, 615816 (3), Autosomal recessive | 99.94% |
| *PHACTR1* | 608723 | Developmental and epileptic encephalopathy 70, 618298 (3), Autosomal dominant | 100% |
| *PHF21A* | 608325 | Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 (3), Autosomal dominant | 99.91% |
| *PHF6* | 300414 | Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive | 99.16% |
| *PHF8* | 300560 | Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263 (3), X-linked recessive | 99.87% |
| *PHGDH* | 606879 | Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive | 99.79% |

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| *PHIP* | 612870 | Chung-Jansen syndrome, 617991 (3), Autosomal dominant | 99.63% |
| PI4K2A | 609763 | No OMIM phenotype<br>PI4K2A deficiency causes innate error in intracellular trafficking with developmental and epileptic-dyskinetic encephalopathy (Dafsari (2022) Ann Clin Transl Neurol. 2022 Sep;9(9):1345-1358), PMID: 35880319 - Autosomal recessive | 99.85% |
| *PI4KA* | 600286 | Spastic paraplegia 84, autosomal recessive, 619621 (3), Autosomal recessive; Gastrointestinal defects and immunodeficiency syndrome 2, 619708 (3), Autosomal recessive; Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive | 99.76% |
| *PIBF1* | 607532 | Joubert syndrome 33, 617767 (3), Autosomal recessive | 99.9% |
| *PIDD1* | 605247 | Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827 (3), Autosomal recessive | 100% |
| *PIGA* | 311770 | Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3); Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 (3) | 99.81% |
| *PIGB* | 604122 | Developmental and epileptic encephalopathy 80, 618580 (3), Autosomal recessive | 99.92% |
| *PIGC* | 601730 | Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive | 100% |
| *PIGF* | 600153 | Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356 (3), Autosomal recessive | 98.59% |
| *PIGG* | 616918 | [Blood group, EMM system], 619812 (3), Autosomal recessive; Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917 (3), Autosomal recessive | 99.99% |
| *PIGH* | 600154 | Glycosylphosphatidylinositol biosynthesis defect 17, 618010 (3), Autosomal recessive | 100% |
| *PIGK* | 605087 | Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 (3), Autosomal recessive | 92.56% |
| *PIGL* | 605947 | CHIME syndrome, 280000 (3), Autosomal recessive | 99.98% |
| *PIGM* | 610273 | Glycosylphosphatidylinositol deficiency, 610293 (3), Autosomal recessive | 99.97% |
| *PIGN* | 606097 | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive | 99.91% |
| *PIGO* | 614730 | Hyperphosphatasia with impaired intellectual development syndrome 2, 614749 (3), Autosomal recessive | 99.99% |
| *PIGP* | 605938 | Developmental and epileptic encephalopathy 55, 617599 (3), Autosomal recessive | 99.87% |
| *PIGQ* | 605754 | Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548 (3), Autosomal recessive | 99.99% |
| *PIGS* | 610271 | Developmental and epileptic encephalopathy 95, 618143 (3), Autosomal recessive | 100% |
| *PIGT* | 610272 | ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Somatic mutation, Autosomal dominant; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive | 99.95% |

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| *PIGU* | 608528 | Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 (3), Autosomal recessive | 99.98% |
| *PIGV* | 610274 | Hyperphosphatasia with impaired intellectual development syndrome 1, 239300 (3), Autosomal recessive | 100% |
| *PIGW* | 610275 | Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive | 99.87% |
| *PIGY* | 610662 | Hyperphosphatasia with impaired intellectual development syndrome 6, 616809 (3), Autosomal recessive | 99.99% |
| PIK3C2B | 602838 | No OMIM phenotype<br>Defective lipid signalling caused by mutations in PIK3C2B underlies focal epilepsy (Gozzelino (2022), Brain. 145(7):2313-2331), PMID: 35786744 - Autosomal dominant | 99.79% |
| *PIK3CA* | 171834 | CLOVE syndrome, somatic, 612918 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Cerebral cavernous malformations 4, somatic, 619538 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); Macrodactyly, somatic, 155500 (3); CLAPO syndrome, somatic, 613089 (3); Keratosis, seborrheic, somatic, 182000 (3); Nevus, epidermal, somatic, 162900 (3); Gastric cancer, somatic, 613659 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3) | 99.74% |
| *PIK3R2* | 603157 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant | 99.95% |
| *PIP5K1C* | 606102 | Lethal congenital contractural syndrome 3, 611369 (3), Autosomal recessive | 99.94% |
| *PISD* | 612770 | Liberfarb syndrome, 618889 (3), Autosomal recessive | 100% |
| *PITRM1* | 618211 | Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive | 99.89% |
| *PLA2G6* | 603604 | Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive | 99.98% |
| *PLAA* | 603873 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive | 99.79% |
| *PLCB1* | 607120 | Developmental and epileptic encephalopathy 12, 613722 (3), Autosomal recessive | 99.98% |
| *PLCH1* | 612835 | Holoprosencephaly 14, 619895 (3), Autosomal recessive | 99.99% |
| *PLK4* | 605031 | Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive | 99.89% |
| *PLP1* | 300401 | Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive | 99.98% |
| *PLPBP* | 604436 | Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive | 99.99% |

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| *PLXNA1* | 601055 | Dworschak-Punetha neurodevelopmental syndrome, 619955 (3), Autosomal recessive | 100% |
| *PLXND1* | 604282 | Congenital heart defects, multiple types, 9, 620294 (3), Autosomal recessive | 99.98% |
| *PMM2* | 601785 | Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive | 99.93% |
| *PMPCA* | 613036 | Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive | 99.99% |
| *PMPCB* | 603131 | Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive | 99.94% |
| *PNKP* | 605610 | ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive | 100% |
| *PNP* | 164050 | Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive | 100% |
| *PNPLA6* | 603197 | Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive | 99.99% |
| *PNPO* | 603287 | Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive | 99.9% |
| *PNPT1* | 610316 | Spinocerebellar ataxia 25, 608703 (3), Autosomal dominant; Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive | 99.56% |
| *POC1A* | 614783 | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive | 99.98% |
| *POC1B* | 614784 | Cone-rod dystrophy 20, 615973 (3), Autosomal recessive | 100% |
| *POGZ* | 614787 | White-Sutton syndrome, 616364 (3), Autosomal dominant | 99.49% |
| *POLA1* | 312040 | Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive | 99.57% |
| *POLG* | 174763 | Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive | 100% |
| *POLG2* | 604983 | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425 (3), Autosomal recessive | 99.51% |

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| *POLR1C* | 610060 | Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive | 100% |
| *POLR2A* | 180660 | Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 (3), Autosomal dominant | 99.95% |
| *POLR3A* | 614258 | Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive | 99.97% |
| *POLR3B* | 614366 | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 (3), Autosomal dominant | 99.94% |
| *POLRMT* | 601778 | Combined oxidative phosphorylation deficiency 55, 619743 (3), Autosomal recessive, Autosomal dominant | 99.99% |
| *POMGNT1* | 606822 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive | 99.69% |
| *POMGNT2* | 614828 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive | 100% |
| *POMK* | 615247 | ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive | 100% |
| *POMT1* | 607423 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155 (3), Autosomal recessive | 99.96% |
| *POMT2* | 607439 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156 (3), Autosomal recessive | 99.98% |
| *PORCN* | 300651 | Focal dermal hypoplasia, 305600 (3), X-linked dominant | 99.93% |
| *POU1F1* | 173110 | Pituitary hormone deficiency, combined or isolated, 1, 613038 (3), Autosomal recessive, Autosomal dominant | 99.98% |

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| POU3F2 | 600494 | No OMIM phenotype<br>Monoallelic intragenic POU3F2 variants lead to neurodevelopmental delay and hyperphagic obesity, confirming the gene's candidacy in 6q16.1 deletions (Schönauer (2023) Am J Hum Genet. 2023 Jun 1;110(6):998-1007), PMID: 37207645 - Autosomal dominant | 99.81% |
| *POU3F3* | 602480 | Snijders Blok-Fisher syndrome, 618604 (3), Autosomal dominant | 99.96% |
| *PPFIBP1* | 603141 | Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024 (3), Autosomal recessive | 99.11% |
| *PPIL1* | 601301 | Pontocerebellar hypoplasia, type 14, 619301 (3), Autosomal recessive | 100% |
| *PPM1D* | 605100 | Breast cancer, somatic, 114480 (3); Jansen-de Vries syndrome, 617450 (3), Autosomal dominant | 99.81% |
| *PPP1CB* | 600590 | Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant | 99.89% |
| *PPP1R12A* | 602021 | Genitourinary and/or/brain malformation syndrome, 618820 (3), Autosomal dominant | 99.08% |
| *PPP1R15B* | 613257 | Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive | 99.92% |
| *PPP1R21* | 618159 | Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383 (3), Autosomal recessive | 99.92% |
| *PPP2CA* | 176915 | Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354 (3), Autosomal dominant | 99.99% |
| *PPP2R1A* | 605983 | Intellectual developmental disorder, autosomal dominant 36, 616362 (3), Autosomal dominant | 100% |
| *PPP2R5D* | 601646 | Intellectual developmental disorder, autosomal dominant 35, 616355 (3), Autosomal dominant | 99.99% |
| *PPP3CA* | 114105 | Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 (3), Autosomal dominant; Developmental and epileptic encephalopathy 91, 617711 (3), Autosomal dominant | 99.84% |
| *PPT1* | 600722 | Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive | 97.48% |
| *PQBP1* | 300463 | Renpenning syndrome, 309500 (3), X-linked recessive | 99.99% |
| *PRDM13* | 616741 | Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive | 99.99% |
| PRDM15 | 617692 | No OMIM phenotype<br>Mutations in PRDM15 Are a Novel Cause of Galloway-Mowat Syndrome (Mann (2021), J Am Soc Nephrol. 32(3):580-596), PMID: 33593823 - Autosomal recessive | 99.99% |
| *PRDM8* | 616639 | ?Epilepsy, progressive myoclonic, 10, 616640 (3), Autosomal recessive | 99.99% |
| *PREPL* | 609557 | Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive | 99.63% |
| *PRF1* | 170280 | Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3) | 100% |

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| *PRICKLE1* | 608500 | Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive | 99.87% |
| PRICKLE2 | 608501 | No OMIM phenotype<br>PRICKLE2 revisited-further evidence implicating PRICKLE2 in neurodevelopmental disorders (Bayat (2021), Eur J Hum Genet. 29(8):1235-1244), PMID: 34092786 - Autosomal dominant | 99.98% |
| PRIMA1 | 613851 | No OMIM phenotype<br>PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy (Hildebrand (2015) Ann Clin Transl Neurol 2(8):821-30), PMID: 26339676 - Autosomal dominant | 99.98% |
| *PRKACB* | 176892 | Cardioacrofacial dysplasia 2, 619143 (3), Somatic mosaicism, Autosomal dominant | 87.33% |
| *PRKAR1A* | 188830 | Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant | 100% |
| *PRKAR1B* | 176911 | Marbach-Schaaf neurodevelopmental syndrome, 619680 (3), Autosomal dominant | 100% |
| *PRMT7* | 610087 | Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive | 99.95% |
| *PRODH* | 606810 | {Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant; Hyperprolinemia, type I, 239500 (3), Autosomal recessive | 4.29% |
| *PRORP* | 609947 | Combined oxidative phosphorylation deficiency 54, 619737 (3), Autosomal recessive | 99.8% |
| *PRPF8* | 607300 | Retinitis pigmentosa 13, 600059 (3), Autosomal dominant | 99.98% |
| *PRPS1* | 311850 | Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive | 99.95% |
| *PRR12* | 616633 | Neuroocular syndrome, 619539 (3), Autosomal dominant | 100% |
| *PRRT2* | 614386 | Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant | 99.97% |
| *PRSS12* | 606709 | Intellectual developmental disorder, autosomal recessive 1, 249500 (3), Autosomal recessive | 99.98% |
| *PRUNE1* | 617413 | Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive | 99.85% |
| *PSAP* | 176801 | Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), | 99.94% |

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| | | Autosomal recessive; Gaucher disease, atypical, 610539 (3); {Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 (3), Autosomal dominant | |
| *PSAT1* | 610936 | Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive | 99.98% |
| *PSMD12* | 604450 | Stankiewicz-Isidor syndrome, 617516 (3), Autosomal dominant | 99.95% |
| *PSPH* | 172480 | Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive | 99.09% |
| *PTCH1* | 601309 | Basal cell nevus syndrome 1, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant | 99.99% |
| *PTCHD1* | 300828 | {Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive | 99.97% |
| *PTDSS1* | 612792 | Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant | 99.95% |
| *PTEN* | 601728 | {Glioma susceptibility 2}, 613028 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos disease, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant | 99.89% |
| *PTF1A* | 607194 | Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive; Pancreatic agenesis 2, 615935 (3), Autosomal recessive | 100% |
| *PTPN11* | 176876 | Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3) | 99.98% |
| *PTPN23* | 606584 | Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive | 100% |
| PTPN4 | 176878 | No OMIM phenotype<br>PTPN4 germline variants result in aberrant neurodevelopment and growth (Chmielewska (2021), HGG Adv. 2021 2(3):100033), PMID: 34527963 - Autosomal dominant | 98.2% |
| *PTRH2* | 608625 | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive | 99.99% |
| PTRHD1 | 617342 | No OMIM phenotype<br>?Neurodevelopmental disorder (Reuter (2017) JAMA Psychiatry), Autosomal recessive<br>Intellectual disability and parkinsonism (Khodadadi (2017), Mov Disord and Elahi (2018), Mov Disord), Autosomal Recessive | 100% |
| *PTS* | 612719 | Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive | 99.93% |
| *PUF60* | 604819 | Verheij syndrome, 615583 (3), Autosomal dominant | 100% |

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| *PUM1* | 607204 | Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant | 98.14% |
| *PURA* | 600473 | Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158 (3), Autosomal dominant | 100% |
| *PUS1* | 608109 | Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive | 100% |
| *PUS3* | 616283 | Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 (3), Autosomal recessive | 100% |
| *PUS7* | 616261 | Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 (3), Autosomal recessive | 99.89% |
| *PYCR1* | 179035 | Cutis laxa, autosomal recessive, type IIIB, 614438 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive | 99.99% |
| *PYCR2* | 616406 | Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive | 99.95% |
| *QARS1* | 603727 | Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive | 100% |
| *QDPR* | 612676 | Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive | 99.92% |
| *QRICH1* | 617387 | Ververi-Brady syndrome, 617982 (3), Autosomal dominant | 100% |
| *RAB11B* | 604198 | Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant | 100% |
| *RAB18* | 602207 | Warburg micro syndrome 3, 614222 (3), Autosomal recessive | 99.76% |
| *RAB23* | 606144 | Carpenter syndrome, 201000 (3), Autosomal recessive | 99.97% |
| *RAB27A* | 603868 | Griscelli syndrome, type 2, 607624 (3), Autosomal recessive | 99.94% |
| *RAB39B* | 300774 | Intellectual developmental disorder, X-linked 72, 300271 (3), X-linked recessive; Waisman syndrome, 311510 (3), X-linked recessive | 99.99% |
| *RAB3GAP1* | 602536 | Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive | 99.73% |
| *RAB3GAP2* | 609275 | Martsolf syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive | 99.69% |
| RABGAP1 | 615882 | No OMIM phenotype<br>Biallelic loss-of-function variants in RABGAP1 cause a novel neurodevelopmental syndrome (Youjin Oh (2022), Genet Med. S1098-3600(22)00877-2), PMID: 36083289 - Autosomal recessive | 99.99% |
| *RAC1* | 602048 | Intellectual developmental disorder, autosomal dominant 48, 617751 (3), Autosomal dominant | 99.75% |
| *RAC3* | 602050 | Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 (3), Autosomal dominant | 100% |

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| *RAD21* | 606462 | Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant; ?Mungan syndrome, 611376 (3), Autosomal recessive | 99.91% |
| *RAF1* | 164760 | Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3), Autosomal dominant | 99.97% |
| *RAI1* | 607642 | Smith-Magenis syndrome, 182290 (3), Isolated cases, Autosomal dominant | 99.22% |
| *RALA* | 179550 | Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311 (3), Autosomal dominant | 99.69% |
| *RALGAPA1* | 608884 | Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 (3), Autosomal recessive | 99.73% |
| *RAP1GDS1* | 179502 | Lymphocytic leukemia, acute T-cell (3) | 99.8% |
| *RAPGEF2* | 609530 | ?Epilepsy, familial adult myoclonic, 7, 618075 (3), Autosomal dominant | 99.84% |
| *RARB* | 180220 | Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant | 99.99% |
| *RARS2* | 611524 | Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive | 99.88% |
| *RBBP8* | 604124 | Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3) | 99.91% |
| RBFOX1 | 605104 | No OMIM phenotype<br>Epilepsy, rolandic (Lal (2013) PLoS One 8, e73323), Autosomal dominant<br>Mental retardation (Bhalla (2004) J Hum Genet 49, 308)<br>?Autism spectrum disorder (Griswold (2015) Mol Autism 6, 43) | 99.95% |
| *RBL2* | 180203 | Brunet-Wagner neurodevelopmental syndrome, 619690 (3), Autosomal recessive | 99.44% |
| *RBM10* | 300080 | TARP syndrome, 311900 (3), X-linked recessive | 99.98% |
| *RBM28* | 612074 | ?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive | 99.99% |
| *RBPJ* | 147183 | Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant | 99.96% |
| *RCBTB1* | 607867 | Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive | 99.99% |
| *RELN* | 600514 | {Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive | 99.98% |
| *RERE* | 605226 | Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant | 99.94% |
| *RFT1* | 611908 | Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive | 99.79% |
| RFX3 | 601337 | No OMIM phenotype<br>Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior (Harris (2021), Genet Med. 23(6):1028-1040), PMID: 33658631 - Autosomal dominant | 99.95% |

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| RFX4 | 603958 | No OMIM phenotype<br>Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior (Harris (2021), Genet Med. 23(6):1028-1040), PMID: 33658631 - Autosomal dominant | 99.95% |
| *RFX7* | 612660 | Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities, 620330 (3), Autosomal dominant | 99.99% |
| RHEB | 601293 | No OMIM phenotype<br>?Intellectual dsiability (Reijnders 2017 (Nat Commun)), Autosomal dominant | 99.97% |
| *RHOBTB2* | 607352 | Developmental and epileptic encephalopathy 64, 618004 (3), Autosomal dominant | 100% |
| *RIMS2* | 606630 | Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive | 99.97% |
| *RIT1* | 609591 | Noonan syndrome 8, 615355 (3), Autosomal dominant | 99.78% |
| *RLIM* | 300379 | Tonne-Kalscheuer syndrome, 300978 (3), X-linked | 99.93% |
| *RMND1* | 614917 | Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive | 99.92% |
| *RNASEH2A* | 606034 | Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive | 99.95% |
| *RNASEH2B* | 610326 | Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive | 99.94% |
| *RNASEH2C* | 610330 | Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive | 99.99% |
| *RNASET2* | 612944 | Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive | 99.99% |
| *RNF113A* | 300951 | Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked | 99.99% |
| *RNF125* | 610432 | Tenorio syndrome, 616260 (3), Autosomal dominant | 99.98% |
| *RNF13* | 609247 | Developmental and epileptic encephalopathy 73, 618379 (3), Autosomal dominant | 99.81% |
| *RNF220* | 616136 | Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688 (3), Autosomal recessive | 99.68% |
| *RNPC3* | 618016 | Pituitary hormone deficiency, combined or isolated, 7, 618160 (3), Autosomal recessive | 88.9% |
| *RNU4ATAC* | 601428 | Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive | 99.95% |
| *RNU7-1* | 617876 | Aicardi-Goutieres syndrome 9, 619487 (3), Autosomal recessive | 33.9% |
| *ROBO1* | 602430 | Pituitary hormone deficiency, combined or isolated, 8, 620303 (3), Autosomal dominant; Neurooculorenal syndrome, 620305 (3), Autosomal recessive; ?Nystagmus 8, congenital, autosomal recessive, 257400 (3), Autosomal recessive | 99.83% |
| *ROGDI* | 614574 | Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive | 99.98% |
| *RORA* | 600825 | Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 (3), Autosomal dominant | 99.97% |

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| *RORB* | 601972 | {Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357 (3), Autosomal dominant | 99.86% |
| *RPGRIP1L* | 610937 | Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive | 96.35% |
| *RPIA* | 180430 | Ribose 5-phosphate isomerase deficiency, 608611 (3), Autosomal recessive | 99.84% |
| *RPL10* | 312173 | {Autism, susceptibility to, X-linked 5}, 300847 (3); Intellectual developmental disorder, X-linked syndromic 35, 300998 (3), X-linked recessive | 84.91% |
| *RPS19* | 603474 | Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant | 100% |
| *RPS6KA3* | 300075 | Intellectual developmental disorder, X-linked 19, 300844 (3), X-linked dominant; Coffin-Lowry syndrome, 303600 (3), X-linked dominant | 98.93% |
| RRAS | 165090 | No OMIM phenotype<br>The Role of R-Ras Proteins in Normal and Pathologic Migration and Morphologic Change (Weber (2021) Am J Pathol. 2021 Sep;191(9):1499-1510), PMID: 34111428 - Autosomal dominant | 99.98% |
| *RRAS2* | 600098 | Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant | 99.94% |
| *RRM2B* | 604712 | Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant | 99.97% |
| *RSPRY1* | 616585 | Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive | 99.73% |
| *RSRC1* | 613352 | Intellectual developmental disorder, autosomal recessive 70, 618402 (3), Autosomal recessive | 99.95% |
| *RTEL1* | 608833 | Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 (3), Autosomal dominant | 100% |
| *RTN4IP1* | 610502 | Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732 (3), Autosomal recessive | 99.98% |
| *RTTN* | 610436 | Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive | 99.93% |
| *RUBCN* | 613516 | Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive | 100% |
| *RUSC2* | 611053 | Intellectual developmental disorder, autosomal recessive 61, 617773 (3), Autosomal recessive | 99.99% |
| *RXYLT1* | 605862 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive | 99.48% |
| *SALL1* | 602218 | Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant | 100% |

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| *SAMD12* | 618073 | Epilepsy, familial adult myoclonic, 1, 601068 (3), Autosomal dominant | 100% |
| *SAMD9* | 610456 | Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant | 99.93% |
| *SAMHD1* | 606754 | ?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive | 99.98% |
| *SARS1* | 607529 | Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive | 98.53% |
| SART3 | 611684 | No OMIM phenotype<br>Variants in SART3 cause a spliceosomopathy characterised by failure of testis development and neuronal defects (Ayers (2023) Nat Commun. 2023 Jun 9;14(1):3403), PMID: 37296101 - Autosomal dominant | 99.99% |
| *SATB1* | 602075 | den Hoed-de Boer-Voisin syndrome, 619229 (3), Autosomal dominant; Developmental delay with dysmorphic facies and dental anomalies, 619228 (3), Autosomal dominant | 100% |
| *SATB2* | 608148 | Glass syndrome, 612313 (3), Autosomal dominant | 99.96% |
| *SBDS* | 607444 | {Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive | 99.93% |
| *SBF1* | 603560 | Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive | 99.99% |
| *SC5D* | 602286 | Lathosterolosis, 607330 (3), Autosomal recessive | 99.97% |
| SCAF4 | 616023 | No OMIM phenotype<br>Neurodevelopmental disorder characterized by mild intellectual disability, seizures, behavioral abnormalities, and various skeletal and structural anomalies (Fliedner (2020), Am J Hum Genet 107(3):544-554), PMID: 32730804 - Autosomal dominant | 99.91% |
| SCAMP5 | 613766 | No OMIM phenotype<br>Identification of an Identical de Novo SCAMP5 Missense Variant in Four Unrelated Patients With Seizures and Severe Neurodevelopmental Delay (Jiao (2020), Front Pharmacol.11:599191), PMID: 33390987 - Autosomal dominant | 99.99% |
| *SCAPER* | 611611 | Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive | 99.75% |
| *SCARB2* | 602257 | Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive | 99.99% |
| *SCN1A* | 182389 | Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Dravet syndrome, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 2, 604403 (3), Autosomal dominant | 99.94% |

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| *SCN1B* | 600235 | Generalized epilepsy with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Developmental and epileptic encephalopathy 52, 617350 (3), Autosomal recessive; Cardiac conduction defect, nonspecific, 612838 (3); Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3) | 99.98% |
| *SCN2A* | 182390 | Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant; Developmental and epileptic encephalopathy 11, 613721 (3), Autosomal dominant; Episodic ataxia, type 9, 618924 (3), Autosomal dominant | 99.86% |
| *SCN3A* | 182391 | Epilepsy, familial focal, with variable foci 4, 617935 (3), Autosomal dominant; Developmental and epileptic encephalopathy 62, 617938 (3), Autosomal dominant | 99.76% |
| *SCN8A* | 600702 | ?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Developmental and epileptic encephalopathy 13, 614558 (3), Autosomal dominant | 99.77% |
| *SCO1* | 603644 | Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive | 99.98% |
| *SCO2* | 604272 | Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive | 100% |
| *SCYL1* | 607982 | Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive | 100% |
| *SDCCAG8* | 613524 | Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive | 100% |
| *SDHA* | 600857 | Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Paragangliomas 5, 614165 (3), Autosomal dominant | 99.98% |
| *SDHAF1* | 612848 | Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive | 99.99% |
| *SEMA3E* | 608166 | CHARGE like (PMID: 31691538, 31691538) | 99.13% |
| *SEMA6B* | 608873 | Epilepsy, progressive myoclonic, 11, 618876 (3), Autosomal dominant | 99.98% |
| *SEPSECS* | 613009 | Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive | 99.78% |
| *SERAC1* | 614725 | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive | 99.9% |
| *SERPINI1* | 602445 | Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3), Autosomal dominant | 99.98% |
| *SET* | 600960 | Intellectual developmental disorder, autosomal dominant 58, 618106 (3), Autosomal dominant | 78.28% |
| *SETBP1* | 611060 | Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 29, 616078 (3), Autosomal dominant | 100% |

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| *SETD1A* | 611052 | Epilepsy, early-onset, with or without developmental delay, 618832 (3), Autosomal dominant; Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056 (3), Autosomal dominant | 99.99% |
| *SETD1B* | 611055 | Intellectual developmental disorder with seizures and language delay, 619000 (3), Autosomal dominant | 99.99% |
| *SETD2* | 612778 | Luscan-Lumish syndrome, 616831 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 70, 620157 (3), Autosomal dominant; Rabin-Pappas syndrome, 620155 (3), Autosomal dominant | 99.91% |
| *SETD5* | 615743 | Intellectual developmental disorder, autosomal dominant 23, 615761 (3), Autosomal dominant | 99.99% |
| *SFXN4* | 615564 | Combined oxidative phosphorylation deficiency 18, 615578 (3), Autosomal recessive | 99.97% |
| *SGPL1* | 603729 | Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive | 99.95% |
| *SGSH* | 605270 | Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive | 100% |
| *SHANK1* | 604999 | NDD & autism (PMID: 34113010, 22503632 | 99.99% |
| *SHANK2* | 603290 | {Autism susceptibility 17}, 613436 (3) | 99.97% |
| *SHANK3* | 606230 | Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3), Autosomal dominant | 98.45% |
| *SHH* | 600725 | Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant | 100% |
| *SHMT2* | 138450 | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 (3), Autosomal recessive | 99.92% |
| *SHOC2* | 602775 | Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant | 99.96% |
| *SHQ1* | 613663 | Neurodevelopmental disorder with dystonia and seizures, 619922 (3), Autosomal recessive; ?Dystonia 35, childhood-onset, 619921 (3), Autosomal recessive | 99.81% |
| *SIAH1* | 602212 | Buratti-Harel syndrome, 619314 (3), Autosomal dominant | 100% |
| *SIK1* | 605705 | Developmental and epileptic encephalopathy 30, 616341 (3), Autosomal dominant | 3.83% |
| *SIL1* | 608005 | Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive | 99.95% |
| *SIN3A* | 607776 | Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant | 99.97% |
| SIN3B | 607777 | No OMIM phenotype<br>Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder (Latypova (2021), Am J Hum Genet. 108(5):929-941), PMID: 33811806 - Autosomal dominant | 99.98% |
| *SIX3* | 603714 | Schizencephaly, 269160 (3); Holoprosencephaly 2, 157170 (3), Autosomal dominant | 100% |

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| *SKI* | 164780 | Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant | 99.98% |
| *SKIC3 (TTC37)* | 614589 | Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive | 99.82% |
| *SLC12A2* | 600840 | Kilquist syndrome, 619080 (3), Autosomal recessive; Delpire-McNeill syndrome, 619083 (3), Autosomal dominant; Deafness, autosomal dominant 78, 619081 (3), Autosomal dominant | 99.55% |
| *SLC12A5* | 606726 | {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Developmental and epileptic encephalopathy 34, 616645 (3), Autosomal recessive | 99.99% |
| *SLC12A6* | 604878 | Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2II, 620068 (3), Autosomal dominant | 99.98% |
| *SLC13A5* | 608305 | Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive | 99.99% |
| *SLC16A1* | 600682 | Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant | 99.26% |
| *SLC16A2* | 300095 | Allan-Herndon-Dudley syndrome, 300523 (3), X-linked | 99.97% |
| *SLC17A5* | 604322 | Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive | 99.71% |
| *SLC19A3* | 606152 | Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive | 99.95% |
| *SLC1A1* | 133550 | Dicarboxylic aminoaciduria, 222730 (3), Autosomal recessive; {?Schizophrenia susceptibility 18}, 615232 (3) | 99.98% |
| *SLC1A2* | 600300 | Developmental and epileptic encephalopathy 41, 617105 (3), Autosomal dominant | 99.95% |
| *SLC1A4* | 600229 | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive | 99.97% |
| *SLC25A1* | 190315 | Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive; Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive | 99.93% |
| *SLC25A12* | 603667 | Developmental and epileptic encephalopathy 39, 612949 (3), Autosomal recessive | 99.72% |
| *SLC25A15* | 603861 | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive | 100% |
| *SLC25A19* | 606521 | Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive | 99.99% |
| *SLC25A20* | 613698 | Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive | 100% |
| *SLC25A22* | 609302 | Developmental and epileptic encephalopathy 3, 609304 (3), Autosomal recessive | 100% |

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| *SLC25A26* | 611037 | Combined oxidative phosphorylation deficiency 28, 616794 (3), Autosomal recessive | 99.76% |
| *SLC25A42* | 610823 | Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive | 99.99% |
| *SLC2A1* | 138140 | Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant | 99.93% |
| SLC32A1 | 616440 | No OMIM phenotype<br>Biallelic variants in SLC35B2 cause a novel chondrodysplasia with hypomyelinating leukodystrophy (Guasto (2022) Brain. 2022 Oct 21;145(10):3711-3722), PMID: 35325049 - Autosomal recessive | 100% |
| *SLC33A1* | 603690 | Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive | 99.67% |
| *SLC35A1* | 605634 | Congenital disorder of glycosylation, type IIf, 603585 (3), Autosomal recessive | 99.81% |
| *SLC35A2* | 314375 | Congenital disorder of glycosylation, type IIm, 300896 (3), X-linked dominant, Somatic mosaicism | 99.97% |
| *SLC35A3* | 605632 | Arthrogryposis, impaired intellectual development, and seizures, 615553 (3), Autosomal recessive | 94.67% |
| *SLC35B2* | 610788 | Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269 (3), Autosomal recessive | 100% |
| *SLC35C1* | 605881 | Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive | 100% |
| *SLC38A3* | 604437 | Developmental and epileptic encephalopathy 102, 619881 (3), Autosomal recessive | 100% |
| *SLC39A14* | 608736 | ?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant; Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive | 92.87% |
| *SLC39A8* | 608732 | Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive | 99.95% |
| *SLC45A1* | 605763 | Intellectual developmental disorder with neuropsychiatric features, 617532 (3), Autosomal recessive | 99.99% |
| *SLC46A1* | 611672 | Folate malabsorption, hereditary, 229050 (3), Autosomal recessive | 100% |
| *SLC4A4* | 603345 | Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive | 99.97% |
| *SLC5A6* | 604024 | Sodium-dependent multivitamin transporter deficiency, 618973 (3), Autosomal recessive; Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903 (3), Autosomal recessive | 100% |
| *SLC6A1* | 137165 | Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant | 99.96% |
| *SLC6A17* | 610299 | Intellectual developmental disorder, autosomal recessive 48, 616269 (3), Autosomal recessive | 99.79% |
| *SLC6A19* | 608893 | Hartnup disorder, 234500 (3), Autosomal recessive | 99.99% |
| *SLC6A3* | 126455 | Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive; {Nicotine dependence, protection against}, 188890 (3) | 99.96% |

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| *SLC6A8* | 300036 | Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive | 99.99% |
| *SLC6A9* | 601019 | Glycine encephalopathy with normal serum glycine, 617301 (3), Autosomal recessive | 99.93% |
| *SLC7A6OS* | 619192 | Epilepsy, progressive myoclonic, 12, 619191 (3), Autosomal recessive | 99.96% |
| *SLC7A7* | 603593 | Lysinuric protein intolerance, 222700 (3), Autosomal recessive | 99.99% |
| *SLC9A6* | 300231 | Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 (3), X-linked | 99.42% |
| *SLC9A7* | 300368 | Intellectual developmental disorder, X-linked 108, 301024 (3), X-linked recessive | 99.91% |
| *SLITRK2* | 300561 | Intellectual developmental disorder, X-linked 111, 301107 (3), X-linked | 100% |
| *SMAD4* | 600993 | Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant | 99.97% |
| *SMARCA2* | 600014 | Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant; Blepharophimosis-impaired intellectual development syndrome, 619293 (3), Autosomal dominant | 99.95% |
| *SMARCA4* | 603254 | Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant | 99.99% |
| SMARCA5 | 603375 | No OMIM phenotype<br>Pathogenic variants in SMARCA5, a chromatin remodeler, cause a range of syndromic neurodevelopmental features (Li (2021), Sci Adv 7(20)), PMID: 33980485 - Autosomal dominant | 99.81% |
| *SMARCB1* | 601607 | Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant | 99.99% |
| *SMARCC2* | 601734 | Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant | 99.73% |
| *SMARCD1* | 601735 | Coffin-Siris syndrome 11, 618779 (3), Autosomal dominant | 99.81% |
| *SMARCE1* | 603111 | {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant | 99.87% |
| *SMC1A* | 300040 | Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 (3), X-linked dominant | 99.98% |
| *SMC3* | 606062 | Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant | 99.91% |
| *SMG8* | 613175 | Alzahrani-Kuwahara syndrome, 619268 (3), Autosomal recessive | 99.92% |
| *SMG9* | 613176 | Heart and brain malformation syndrome, 616920 (3), Autosomal recessive; Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995 (3), Autosomal recessive | 99.99% |
| *SMOC1* | 608488 | Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive | 100% |

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| *SMPD1* | 607608 | Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive | 100% |
| *SMPD4* | 610457 | Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive | 99.9% |
| *SMS* | 300105 | Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 (3), X-linked recessive | 98.56% |
| *SNAP25* | 600322 | ?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant | 99.89% |
| *SNAP29* | 604202 | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive | 99.85% |
| *SNIP1* | 608241 | Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501 (3), Autosomal recessive | 99.96% |
| *SNORD118* | 616663 | Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive | 100% |
| *SNRPN* | 182279 | Prader-Willi syndrome, autosomal dominant (PMID: 29437285, 34200226, 34099539) | 99.97% |
| *SNX14* | 616105 | Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive | 99.73% |
| SNX27 | 611541 | No OMIM phenotype<br>Seizures, developmental delay, behavioral disturbance, and subcortical brain abnormalities (Parente (2019), Clin Genet 97(3):437-446), PMID: 31721175 - Autosomal recessive | 99.59% |
| *SOBP* | 613667 | ?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive | 99.99% |
| *SON* | 182465 | ZTTK syndrome, 617140 (3), Autosomal dominant | 99.95% |
| *SOS1* | 182530 | Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant | 99.68% |
| *SOS2* | 601247 | Noonan syndrome 9, 616559 (3), Autosomal dominant | 99.39% |
| *SOX10* | 602229 | Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant | 100% |
| *SOX11* | 600898 | Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866 (3), Autosomal dominant | 100% |
| *SOX2* | 184429 | Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant; Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant | 100% |
| *SOX3* | 313430 | Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked | 100% |
| *SOX4* | 184430 | Coffin-Siris syndrome 10, 618506 (3), Autosomal dominant | 99.36% |

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| *SOX5* | 604975 | Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant | 99.96% |
| *SOX6* | 607257 | Tolchin-Le Caignec syndrome, 618971 (3), Autosomal dominant | 99.89% |
| *SPART* | 607111 | Troyer syndrome, 275900 (3), Autosomal recessive | 99.98% |
| *SPAST* | 604277 | Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant | 99.77% |
| *SPECC1L* | 614140 | Teebi hypertelorism syndrome 1, 145420 (3), Autosomal dominant; ?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant | 99.98% |
| *SPEN* | 613484 | Radio-Tartaglia syndrome, 619312 (3), Autosomal dominant | 99.98% |
| *SPG11* | 610844 | Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive | 99.89% |
| *SPOP* | 602650 | Nabais Sa-de Vries syndrome, type 1, 618828 (3), Autosomal dominant; Nabais Sa-de Vries syndrome, type 2, 618829 (3), Autosomal dominant | 99.85% |
| *SPR* | 182125 | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), Autosomal recessive, ?Autosomal dominant | 99.99% |
| *SPRED1* | 609291 | Legius syndrome, 611431 (3), Autosomal dominant | 99.99% |
| *SPRED2* | 609292 | Noonan syndrome 14, 619745 (3), Autosomal recessive | 99.99% |
| *SPTAN1* | 182810 | Developmental and epileptic encephalopathy 5, 613477 (3), Autosomal dominant | 99.96% |
| *SPTBN1* | 182790 | Developmental delay, impaired speech, and behavioral abnormalities, 619475 (3), Autosomal dominant | 99.98% |
| *SPTBN2* | 604985 | Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive | 99.98% |
| *SPTBN4* | 606214 | Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive | 99.91% |
| *SRCAP* | 611421 | Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 (3), Autosomal dominant; Floating-Harbor syndrome, 136140 (3), Autosomal dominant | 99.99% |
| *SRD5A3* | 611715 | Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive | 99.94% |
| *SRRM2* | 606032 | Intellectual developmental disorder, autosomal dominant 72, 620439 (3), Autosomal dominant | 99.98% |
| *SSR4* | 300090 | Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive | 99.99% |
| *ST3GAL3* | 606494 | Developmental and epileptic encephalopathy 15, 615006 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 12, 611090 (3), Autosomal recessive | 99.98% |
| *ST3GAL5* | 604402 | Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive | 99.96% |
| *STAG1* | 604358 | Intellectual developmental disorder, autosomal dominant 47, 617635 (3), Autosomal dominant | 99.88% |

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| *STAG2* | 300826 | Holoprosencephaly 13, X-linked, 301043 (3), X-linked recessive, X-linked dominant; Mullegama-Klein-Martinez syndrome, 301022 (3), X-linked | 99.08% |
| *STAMBP* | 606247 | Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive | 99.95% |
| *STARD7* | 616712 | Epilepsy, familial adult myoclonic, 2, 607876 (3), Autosomal dominant | 99.6% |
| *STEEP1* | 301012 | ?Intellectual developmental disorder, X-linked 107, 301013 (3), X-linked | 99.89% |
| *STIL* | 181590 | Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive | 99.11% |
| *STRA6* | 610745 | Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive | 99.95% |
| *STRADA* | 608626 | Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive | 99.98% |
| *STT3A* | 601134 | Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 (3), Autosomal dominant; Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596 (3), Autosomal recessive | 99.99% |
| *STT3B* | 608605 | Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive | 99.96% |
| STX1A | 186590 | No OMIM phenotype<br>Heterozygous and homozygous variants in STX1A cause a neurodevelopmental disorder with or without epilepsy (Luppe (2023) Eur J Hum Genet. 2023 Mar;31(3):345-352), PMID: 36564538 - Autosomal dominant and Autosomal recessive | 99.98% |
| *STX1B* | 601485 | Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant | 99.97% |
| *STXBP1* | 602926 | Developmental and epileptic encephalopathy 4, 612164 (3), Autosomal recessive, Autosomal dominant | 99.99% |
| *SUCLA2* | 603921 | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive | 99.96% |
| *SUCLG1* | 611224 | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive | 99.64% |
| *SUFU* | 607035 | {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Basal cell nevus syndrome 2, 620343 (3); {Medulloblastoma}, 155255 (3), Autosomal recessive, Somatic mutation, Autosomal dominant | 100% |
| *SUMF1* | 607939 | Multiple sulfatase deficiency, 272200 (3), Autosomal recessive | 99.95% |
| *SUOX* | 606887 | Sulfite oxidase deficiency, 272300 (3), Autosomal recessive | 100% |
| *SUPT16H* | 605012 | Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480 (3), Autosomal dominant | 99.97% |
| *SURF1* | 185620 | Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive | 100% |

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| *SUZ12* | 606245 | Imagawa-Matsumoto syndrome, 618786 (3), Autosomal dominant | 98.58% |
| *SVBP* | 617853 | Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive | 99.04% |
| *SYN1* | 313440 | Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491 (3), X-linked; Intellectual developmental disorder, X-linked 50, 300115 (3), X-linked | 99.98% |
| SYNCRIP | 616686 | No OMIM phenotype<br>Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder (Semino (2021) Hum Mutat 42(9):1094-1100), PMID: 34157790 - Autosomal dominant | 99.93% |
| *SYNE1* | 608441 | Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive | 99.95% |
| *SYNGAP1* | 603384 | Intellectual developmental disorder, autosomal dominant 5, 612621 (3), Autosomal dominant | 99.96% |
| *SYNJ1* | 604297 | Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive; Developmental and epileptic encephalopathy 53, 617389 (3), Autosomal recessive | 99.91% |
| *SYP* | 313475 | Intellectual developmental disorder, X-linked 96, 300802 (3), X-linked recessive | 99.99% |
| *SYT1* | 185605 | Baker-Gordon syndrome, 618218 (3), Autosomal dominant | 99.7% |
| *SYT14* | 610949 | ?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive | 99.94% |
| *SZT2* | 615463 | Developmental and epileptic encephalopathy 18, 615476 (3), Autosomal recessive | 99.88% |
| *TACO1* | 612958 | Mitochondrial complex IV deficiency, nuclear type 8, 619052 (3), Autosomal recessive | 100% |
| *TAF1* | 313650 | Intellectual developmental disorder, X-linked syndromic 33, 300966 (3), X-linked recessive; Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive | 99.84% |
| *TAF13* | 600774 | Intellectual developmental disorder, autosomal recessive 60, 617432 (3), Autosomal recessive | 95.71% |
| TAF1C | 604905 | No OMIM phenotype<br>Homozygous TAF1C variants are associated with a novel childhood-onset neurological phenotype (Knuutinen (2020) Clin Genet 98(5):493-498), PMID: 32779182 - Autosomal dominant | 100% |
| *TAF2* | 604912 | Intellectual developmental disorder, autosomal recessive 40, 615599 (3), Autosomal recessive | 99.72% |
| *TAF4* | 601796 | Intellectual developmental disorder, autosomal dominant 73, 620450 (3), Autosomal dominant | 99.39% |
| *TAF6* | 602955 | Alazami-Yuan syndrome, 617126 (3), Autosomal recessive | 99.96% |
| *TAF8* | 609514 | Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972 (3), Autosomal recessive | 99.91% |
| *TANC2* | 615047 | Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906 (3), Autosomal dominant | 99.85% |

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| *TANGO2* | 616830 | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive | 99.85% |
| *TAOK1* | 610266 | Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575 (3), Autosomal dominant | 99.77% |
| *TARS2* | 612805 | Combined oxidative phosphorylation deficiency 21, 615918 (3), Autosomal recessive | 99.77% |
| *TASP1* | 608270 | Suleiman-El-Hattab syndrome, 618950 (3), Autosomal recessive | 99.86% |
| *TAT* | 613018 | Tyrosinemia, type II, 276600 (3), Autosomal recessive | 99.99% |
| *TBC1D20* | 611663 | Warburg micro syndrome 4, 615663 (3), Autosomal recessive | 100% |
| *TBC1D23* | 617687 | Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive | 98.7% |
| *TBC1D24* | 613577 | Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive | 100% |
| *TBC1D2B* | 619152 | Neurodevelopmental disorder with seizures and gingival overgrowth, 619323 (3), Autosomal recessive | 99.94% |
| *TBC1D32* | 615867 | Ciliopathy (PMID: 32573025, 31130284, 32060556) | 99.75% |
| *TBC1D7* | 612655 | Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 (3), Autosomal recessive | 99.9% |
| *TBCD* | 604649 | Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive | 100% |
| *TBCE* | 604934 | Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive | 99.91% |
| *TBCK* | 616899 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive | 99.75% |
| *TBL1XR1* | 608628 | Intellectual developmental disorder, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant | 99.92% |
| *TBP* | 600075 | Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant | 99.98% |
| *TBR1* | 604616 | Intellectual developmental disorder with autism and speech delay, 606053 (3), Autosomal dominant | 99.99% |

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| *TBX1* | 602054 | Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant | 99.95% |
| *TCEAL1* | 300237 | Neurodevelopmental disorder with gait disturbance, dysmorphic facies and behavioral abnormalities, X-linked, 301094 (3), X-linked dominant | 100% |
| *TCF20* | 603107 | Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 (3), Autosomal dominant | 100% |
| *TCF4* | 602272 | Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant | 98.56% |
| *TCF7L2* | 602228 | {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant | 99.69% |
| *TCN2* | 613441 | Transcobalamin II deficiency, 275350 (3), Autosomal recessive | 100% |
| *TCTN1* | 609863 | Joubert syndrome 13, 614173 (3), Autosomal recessive | 99.92% |
| *TCTN2* | 613846 | Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive | 99.99% |
| *TCTN3* | 613847 | Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive | 99.92% |
| *TDP2* | 605764 | Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive | 99.97% |
| *TECPR2* | 615000 | Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive | 99.96% |
| *TECR* | 610057 | Intellectual developmental disorder, autosomal recessive 14, 614020 (3), Autosomal recessive | 99.99% |
| *TELO2* | 611140 | You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive | 99.99% |
| *TENM3* | 610083 | Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive | 99.99% |
| *TET3* | 613555 | Beck-Fahrner syndrome, 618798 (3), Autosomal recessive, Autosomal dominant | 99.95% |
| *TFAP2A* | 107580 | Branchiooculofacial syndrome, 113620 (3), Autosomal dominant | 100% |
| *TFE3* | 314310 | Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066 (3), X-linked; Renal cell carcinoma, papillary, 1, 300854 (3) | 99.87% |
| *TGDS* | 616146 | Catel-Manzke syndrome, 616145 (3), Autosomal recessive | 99.86% |
| *TGIF1* | 602630 | Holoprosencephaly 4, 142946 (3), Autosomal dominant | 100% |
| *TH* | 191290 | Segawa syndrome, recessive, 605407 (3), Autosomal recessive | 99.99% |
| *THG1L* | 618802 | Spinocerebellar ataxia, autosomal recessive 28, 618800 (3), Autosomal recessive | 99.99% |
| *THOC2* | 300395 | Intellectual developmental disorder, X-linked 12, 300957 (3), X-linked recessive | 99.11% |
| *THOC6* | 615403 | Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive | 99.94% |

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| *THRA* | 190120 | Hypothyroidism, congenital, nongoitrous, 6, 614450 (3), Autosomal dominant | 100% |
| *THRB* | 190160 | Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant | 99.82% |
| *THUMPD1* | 616662 | Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989 (3), Autosomal recessive | 99.79% |
| *TIAM1* | 600687 | Neurodevelopmental disorder with language delay and seizures, 619908 (3), Autosomal recessive | 99.99% |
| *TIMM50* | 607381 | 3-methylglutaconic aciduria, type IX, 617698 (3), Autosomal recessive | 99.99% |
| *TIMM8A* | 300356 | Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive | 100% |
| *TINF2* | 604319 | Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant | 100% |
| *TKT* | 606781 | Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive | 97.39% |
| *TLK2* | 608439 | Intellectual developmental disorder, autosomal dominant 57, 618050 (3), Autosomal dominant | 98.83% |
| *TMCO1* | 614123 | Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980 (3), Autosomal recessive | 99.48% |
| *TMEM106B* | 613413 | Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant | 99.92% |
| *TMEM107* | 616183 | Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive | 100% |
| *TMEM138* | 614459 | Joubert syndrome 16, 614465 (3), Autosomal recessive | 100% |
| *TMEM147* | 613585 | Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075 (3), Autosomal recessive | 100% |
| *TMEM163* | 618978 | Leukodystrophy, hypomyelinating, 25, 620243 (3), Autosomal dominant | 100% |
| *TMEM165* | 614726 | Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive | 99.97% |
| *TMEM216* | 613277 | Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive | 99.98% |
| *TMEM222* | 619469 | Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470 (3), Autosomal recessive | 99.93% |
| *TMEM231* | 614949 | Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive | 88.88% |
| *TMEM237* | 614423 | Joubert syndrome 14, 614424 (3), Autosomal recessive | 99.3% |
| *TMEM240* | 616101 | Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant | 99.99% |
| *TMEM63A* | 618685 | Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 (3), Autosomal dominant | 99.91% |

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| *TMEM67* | 609884 | Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive | 99.69% |
| *TMEM70* | 612418 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive | 99.99% |
| *TMEM94* | 618163 | Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 (3), Autosomal recessive | 99.96% |
| *TMLHE* | 300777 | {Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive | 77.23% |
| *TMTC3* | 617218 | Lissencephaly 8, 617255 (3), Autosomal recessive | 97.8% |
| *TMX2* | 616715 | Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive | 99.99% |
| *TNIK* | 610005 | Intellectual developmental disorder, autosomal recessive 54, 617028 (3), Autosomal recessive | 99.93% |
| *TNPO2* | 603002 | Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556 (3), Autosomal dominant | 99.99% |
| *TNR* | 601995 | Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653 (3), Autosomal recessive | 99.8% |
| *TNRC6A* | 610739 | ?Epilepsy, familial adult myoclonic, 6, 618074 (3), Autosomal dominant | 99.48% |
| *TNRC6B* | 610740 | Global developmental delay with speech and behavioral abnormalities, 619243 (3), Autosomal dominant | 99.99% |
| *TOE1* | 613931 | Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive | 99.96% |
| *TOGARAM1* | 617618 | Joubert syndrome 37, 619185 (3), Autosomal recessive | 99.93% |
| *TOP3A* | 601243 | ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 (3), Autosomal recessive; Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 (3), Autosomal recessive | 99.91% |
| *TOR1A* | 605204 | {Dystonia-1, modifier of} (3); Arthrogryposis multiplex congenita 5, 618947 (3), Autosomal recessive; Dystonia-1, torsion, 128100 (3), Autosomal dominant | 100% |
| *TP53RK* | 608679 | Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive | 100% |
| *TP73* | 601990 | Ciliary dyskinesia, primary, 47, and lissencephaly, 619466 (3), Autosomal recessive | 100% |
| *TPI1* | 190450 | Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive | 99.95% |
| *TPK1* | 606370 | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive | 99.96% |
| *TPO* | 606765 | Thyroid dyshormonogenesis 2A, 274500 (3), Autosomal recessive | 100% |

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| *TPP1* | 607998 | Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive | 99.99% |
| *TPRKB* | 608680 | Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive | 81.09% |
| TRA2B | 602719 | No OMIM phenotype<br>Clustered variants in the 5' coding region of TRA2B cause a distinctive neurodevelopmental syndrome (Ramond (2023) Genet Med. 2023 Apr;25(4):100003), PMID: 36549593 - Autosomal dominant | 99.98% |
| *TRAF7* | 606692 | Cardiac, facial, and digital anomalies with developmental delay, 618164 (3), Autosomal dominant | 99.97% |
| *TRAIP* | 605958 | Seckel syndrome 9, 616777 (3), Autosomal recessive | 99.97% |
| *TRAK1* | 608112 | Developmental and epileptic encephalopathy 68, 618201 (3), Autosomal recessive | 99.98% |
| *TRAPPC10* | 602103 | Neurodevelopmental disorder with microcephaly, short stature, and speech delay, 620027 (3), Autosomal recessive | 84.42% |
| *TRAPPC11* | 614138 | Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive | 99.93% |
| *TRAPPC12* | 614139 | Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive | 99.96% |
| *TRAPPC2L* | 610970 | Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 (3), Autosomal recessive | 100% |
| *TRAPPC4* | 610971 | Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive | 100% |
| *TRAPPC6B* | 610397 | Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 (3), Autosomal recessive | 99.74% |
| *TRAPPC9* | 611966 | Intellectual developmental disorder, autosomal recessive 13, 613192 (3), Autosomal recessive | 99.98% |
| *TREX1* | 606609 | Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant | 100% |
| *TRIM32* | 602290 | ?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive | 100% |
| *TRIM8* | 606125 | Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428 (3), Autosomal dominant | 99.82% |
| *TRIO* | 601893 | Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825 (3), Autosomal dominant | 99.98% |
| *TRIP12* | 604506 | Intellectual developmental disorder, autosomal dominant 49, 617752 (3), Autosomal dominant | 99.62% |

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| *TRIP4* | 604501 | ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive | 99.97% |
| *TRIT1* | 617840 | Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive | 99.25% |
| *TRMT1* | 611669 | Intellectual developmental disorder, autosomal recessive 68, 618302 (3), Autosomal recessive | 100% |
| *TRMT10A* | 616013 | Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive | 99.9% |
| *TRMT10C* | 615423 | Combined oxidative phosphorylation deficiency 30, 616974 (3), Autosomal recessive | 99.55% |
| *TRNT1* | 612907 | Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive | 99.97% |
| *TRPM3* | 608961 | ?Cataract 50 with or without glaucoma, 620253 (3); Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224 (3), Autosomal dominant | 99.89% |
| *TRPM6* | 607009 | Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive | 99.93% |
| *TRRAP* | 603015 | ?Deafness, autosomal dominant 75, 618778 (3), Autosomal dominant; Developmental delay with or without dysmorphic facies and autism, 618454 (3), Autosomal dominant | 99.79% |
| *TSC1* | 605284 | Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3) | 99.99% |
| *TSC2* | 191092 | Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant | 99.98% |
| *TSEN15* | 608756 | Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive | 99.57% |
| *TSEN2* | 608753 | Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive | 99.98% |
| *TSEN34* | 608754 | ?Pontocerebellar hypoplasia type 2C, 612390 (3), Autosomal recessive | 100% |
| *TSEN54* | 608755 | Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive | 100% |
| *TSFM* | 604723 | Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive | 100% |
| *TSHB* | 188540 | Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive | 99.95% |
| *TSPAN7* | 300096 | Intellectual developmental disorder, X-linked 58, 300210 (3), X-linked recessive | 99.97% |
| *TSPOAP1* | 610764 | Dystonia 22, 620453 (3), Autosomal recessive | 99.95% |
| *TTC19* | 613814 | Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive | 99.99% |
| *TTC21B* | 612014 | Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant | 99.5% |

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| *TTC5* | 619014 | Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244 (3), Autosomal recessive | 100% |
| *TTC8* | 608132 | Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive | 99.67% |
| *TTI1* | 614425 | Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445 (3), Autosomal recessive | 100% |
| *TTI2* | 614426 | Intellectual developmental disorder, autosomal recessive 39, 615541 (3), Autosomal recessive | 99.94% |
| *TUBA1A* | 602529 | Lissencephaly 3, 611603 (3), Autosomal dominant | 99.97% |
| *TUBB* | 191130 | Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant | 100% |
| *TUBB2A* | 615101 | Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant | 87.26% |
| *TUBB2B* | 612850 | Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant | 87.69% |
| *TUBB3* | 602661 | Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant | 100% |
| *TUBB4A* | 602662 | Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant | 100% |
| *TUBG1* | 191135 | Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant | 99.95% |
| *TUBGCP2* | 617817 | Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 (3), Autosomal recessive | 96.54% |
| *TUBGCP4* | 609610 | Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive | 99.8% |
| *TUBGCP6* | 610053 | Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive | 100% |
| *TUFM* | 602389 | Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive | 100% |
| *TUSC3* | 601385 | Intellectual developmental disorder, autosomal recessive 7, 611093 (3), Autosomal recessive | 99.96% |
| *TWIST1* | 601622 | Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant | 100% |
| *TWNK* | 606075 | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive | 100% |
| U2AF2 | 191318 | No OMIM phenotype<br>Global developmental delay, systemic dysmorphism and epilepsy in a patient with a de novo U2AF2 variant (Hiraide (2021), J Hum Genet. 66(12):1185-1187), PMID: 34112922 - Autosomal dominant | 99.98% |

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| *UBA5* | 610552 | ?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive | 99.95% |
| UBAP2L | 616472 | No OMIM phenotype<br>De novo variants in genes regulating stress granule assembly associate with neurodevelopmental disorders (Jia (2022) Sci Adv. 2022 Aug 19;8(33):eabo7112), PMID: 35977029 - Autosomal dominant | 99.45% |
| *UBE2A* | 312180 | Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 (3), X-linked recessive | 99.77% |
| *UBE3A* | 601623 | Angelman syndrome, 105830 (3), Autosomal dominant | 99.23% |
| *UBE3B* | 608047 | Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive | 99.99% |
| *UBE4A* | 603753 | Neurodevelopmental disorder with hypotonia and gross motor and seech delay, 619639 (3), Autosomal recessive | 99.99% |
| *UBR1* | 605981 | Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive | 99.93% |
| *UBR7* | 613816 | Li-Campeau syndrome, 619189 (3), Autosomal recessive | 99.96% |
| *UBTF* | 600673 | Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant | 99.99% |
| *UFC1* | 610554 | Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive | 99.35% |
| *UFM1* | 610553 | Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive | 99.25% |
| *UFSP2* | 611482 | ?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant; Developmental and epileptic encephalopathy 106, 620028 (3), Autosomal recessive | 99.74% |
| *UGDH* | 603370 | Developmental and epileptic encephalopathy 84, 618792 (3), Autosomal recessive | 99.77% |
| *UGP2* | 191760 | Developmental and epileptic encephalopathy 83, 618744 (3), Autosomal recessive | 99.85% |
| *UMPS* | 613891 | Orotic aciduria, 258900 (3), Autosomal recessive | 99.9% |
| *UNC80* | 612636 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive | 98.04% |
| *UPB1* | 606673 | Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive | 100% |
| *UPF3B* | 300298 | Intellectual developmental disorder, X-linked syndromic 14, 300676 (3), X-linked recessive | 99.26% |
| *UQCRQ* | 612080 | Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive | 99.96% |
| *UROC1* | 613012 | ?Urocanase deficiency, 276880 (3), Autosomal recessive | 99.99% |
| *USP18* | 607057 | Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive | 93.05% |
| *USP27X* | 300975 | Intellectual developmental disorder, X-linked 105, 300984 (3), X-linked recessive | 99.99% |
| *USP7* | 602519 | Hao-Fountain syndrome, 616863 (3), Autosomal dominant | 99.9% |

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| *USP9X* | 300072 | Intellectual developmental disorder, X-linked 99, 300919 (3), X-linked recessive; Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant | 99.84% |
| *VAMP1* | 185880 | Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant | 100% |
| *VAMP2* | 185881 | Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant | 100% |
| *VARS1* | 192150 | Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 (3), Autosomal recessive | 99.99% |
| *VARS2* | 612802 | Combined oxidative phosphorylation deficiency 20, 615917 (3), Autosomal recessive | 99.98% |
| *VLDLR* | 192977 | Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050 (3), Autosomal recessive | 99.99% |
| *VPS11* | 608549 | ?Dystonia 32, 619637 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive | 99.99% |
| *VPS13B* | 607817 | Cohen syndrome, 216550 (3), Autosomal recessive | 99.9% |
| *VPS16* | 608550 | Dystonia 30, 619291 (3), Autosomal dominant | 100% |
| *VPS37A* | 609927 | Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive | 99.96% |
| *VPS41* | 605485 | Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive | 99.92% |
| *VPS4A* | 609982 | CIMDAG syndrome, 619273 (3), Autosomal dominant | 99.98% |
| *VPS51* | 615738 | Pontocerebellar hypoplasia, type 13, 618606 (3), Autosomal recessive | 93% |
| *VPS53* | 615850 | Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive | 100% |
| *VRK1* | 602168 | Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive | 99.98% |
| *WAC* | 615049 | Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant | 99.76% |
| *WARS1* | 191050 | Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, type IX, 617721 (3), Autosomal dominant | 99.97% |
| *WARS2* | 604733 | Parkinsonism-dystonia 3, childhood-onset, 619738 (3), Autosomal recessive; Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive | 97.72% |
| *WASF1* | 605035 | Neurodevelopmental disorder with absent language and variable seizures, 618707 (3), Autosomal dominant | 99.86% |
| *WASHC4* | 615748 | Intellectual developmental disorder, autosomal recessive 43, 615817 (3), Autosomal recessive | 99.84% |
| *WASHC5* | 610657 | Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant | 99.98% |

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| *WDFY3* | 617485 | ?Microcephaly 18, primary, autosomal dominant, 617520 (3), Autosomal dominant | 99.93% |
| *WDR19* | 608151 | Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive | 99.8% |
| *WDR26* | 617424 | Skraban-Deardorff syndrome, 617616 (3), Autosomal dominant | 96.29% |
| *WDR37* | 618586 | Neurooculocardiogenitourinary syndrome, 618652 (3), Autosomal dominant | 99.94% |
| *WDR4* | 605924 | Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive | 99.95% |
| *WDR45* | 300526 | Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant | 99.99% |
| *WDR45B* | 609226 | Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive | 100% |
| WDR5 | 609012 | No OMIM phenotype<br>ESHG2021 - A clustering of missense variants in the crucial chromatin modifier WDR5 defines a new neurodevelopmental disorder - Snijders Blok (Nijmegen) - Autosomal dominant | 100% |
| *WDR62* | 613583 | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive | 99.96% |
| *WDR73* | 616144 | Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive | 99.92% |
| *WDR81* | 614218 | Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive | 100% |
| *WFS1* | 606201 | Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive | 99.99% |
| *WIPI2* | 609225 | ?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453 (3), Autosomal recessive | 99.99% |
| *WLS* | 611514 | Zaki syndrome, 619648 (3), Autosomal recessive | 99.6% |
| WNK3 | 300358 | No OMIM phenotype<br>Rare pathogenic variants in WNK3 cause X-linked intellectual disability (Küry (2022) Genet Med. 2022 Sep;24(9):1941-1951), PMID: 35678782 - X-linked recessive | 99.82% |
| *WNT1* | 164820 | {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3), Autosomal dominant; Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive | 99.97% |

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| *WWOX* | 605131 | Esophageal squamous cell carcinoma, somatic, 133239 (3); Developmental and epileptic encephalopathy 28, 616211 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive | 100% |
| *XK* | 314850 | McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked | 99.98% |
| *XPA* | 611153 | Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive | 99.68% |
| *XPNPEP3* | 613553 | Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive | 99.99% |
| *XRCC4* | 194363 | Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive | 99.89% |
| *XYLT1* | 608124 | Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive | 99.98% |
| *XYLT2* | 608125 | {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive | 99.97% |
| *YAP1* | 606608 | Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433 (3), Autosomal dominant | 99.87% |
| *YEATS2* | 613373 | ?Epilepsy, myoclonic, familial adult, 4, 615127 (3), Autosomal dominant | 99.96% |
| *YIF1B* | 619109 | Kaya-Barakat-Masson syndrome, 619125 (3), Autosomal recessive | 99.8% |
| *YIPF5* | 611483 | Microcephaly, epilepsy, and diabetes syndrome 2, 619278 (3), Autosomal recessive | 99.99% |
| *YME1L1* | 607472 | ?Optic atrophy 11, 617302 (3), Autosomal recessive | 99.87% |
| YWHAE | 605066 | No OMIM phenotype<br>YWHAE loss of function causes a rare neurodevelopmental disease with brain abnormalities in human and mouse (Denommé-Pichon (2023) Genet Med. 2023 Jul;25(7):100835), PMID: 36999555 - Autosomal dominant | 84.93% |
| *YWHAG* | 605356 | Developmental and epileptic encephalopathy 56, 617665 (3), Autosomal dominant | 99.99% |
| *YY1* | 600013 | Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant | 100% |
| *ZBTB11* | 618181 | Intellectual developmental disorder, autosomal recessive 69, 618383 (3), Autosomal recessive | 99.97% |
| *ZBTB16* | 176797 | Leukemia, acute promyelocytic, PL2F/RARA type (3) | 100% |
| *ZBTB18* | 608433 | Intellectual developmental disorder, autosomal dominant 22, 612337 (3), Autosomal dominant | 99.41% |
| *ZBTB20* | 606025 | Primrose syndrome, 259050 (3), Autosomal dominant | 100% |
| *ZBTB24* | 614064 | Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive | 99.99% |
| *ZBTB7A* | 605878 | Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769 (3), Autosomal dominant | 100% |
| *ZC3H14* | 613279 | Intellectual developmental disorder, autosomal recessive 56, 617125 (3), Autosomal recessive | 99.96% |

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| *ZC4H2* | 300897 | Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant | 99.98% |
| *ZDHHC9* | 300646 | Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799 (3), X-linked | 99.61% |
| *ZEB2* | 605802 | Mowat-Wilson syndrome, 235730 (3), Autosomal dominant | 99.97% |
| *ZFHX3* | 104155 | Prostate cancer, somatic, 176807 (3) | 99.96% |
| *ZFHX4* | 606940 | ?Ptosis, congenital, 178300 (2), Autosomal dominant | 99.99% |
| *ZFYVE26* | 612012 | Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive | 100% |
| *ZIC1* | 600470 | ?Craniosynostosis 6, 616602 (3), Autosomal dominant; Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 (3), Autosomal dominant | 99.99% |
| *ZIC2* | 603073 | Holoprosencephaly 5, 609637 (3), Autosomal dominant | 100% |
| *ZMIZ1* | 607159 | Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 (3), Autosomal dominant | 99.92% |
| *ZMYM2* | 602221 | Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522 (3), Autosomal dominant | 99.88% |
| *ZMYM3* | 300061 | Intellectual developmental disorder, X-linked 112, 301111 (3), X-linked recessive | 99.98% |
| *ZMYND11* | 608668 | Intellectual developmental disorder, autosomal dominant 30, 616083 (3), Autosomal dominant | 99.99% |
| ZMYND8 | 615713 | No OMIM phenotype<br>De Novo ZMYND8 variants result in an Autosomal dominant neurodevelopmental disorder with cardiac malformations (Dias (2022) enet Med. 2022 Sep;24(9):1952-1966), PMID: 35916866 - Autosomal dominant | 99.99% |
| *ZNF142* | 604083 | Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 (3), Autosomal recessive | 100% |
| *ZNF148* | 601897 | Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 (3), Autosomal dominant | 99.85% |
| *ZNF292* | 616213 | Intellectual developmental disorder, autosomal dominant 64, 619188 (3), Autosomal dominant | 99.91% |
| *ZNF335* | 610827 | Microcephaly 10, primary, autosomal recessive, 615095 (3), Autosomal recessive | 100% |
| *ZNF407* | 615894 | SIMHA syndrome, 619557 (3), Autosomal recessive | 99.98% |
| *ZNF423* | 604557 | Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant; Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant | 98.94% |
| *ZNF462* | 617371 | Weiss-Kruszka syndrome, 618619 (3), Autosomal dominant | 99.99% |
| *ZNF526* | 614387 | Dentici-Novelli neurodevelopmental syndrome, 619877 (3), Autosomal recessive | 100% |
| *ZNF668* | 617103 | Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194 (3), Autosomal recessive | 99.99% |

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| *ZNF699* | 609571 | DEGCAGS syndrome, 619488 (3), Autosomal recessive | 100% |
| *ZNF711* | 314990 | Intellectual developmental disorder, X-linked 97, 300803 (3), X-linked | 99.43% |
| *ZNHIT3* | 604500 | PEHO syndrome, 260565 (3), Autosomal recessive | 62.89% |
| *ZSWIM6* | 615951 | Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant; Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant | 98.94% |

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: July 31, 2023

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.