

<b>ID_and_epilepsy panel</b>		
<b>versie</b>	v6 (1689 genen)	Centrum voor Medische Genetica Gent
<b>Gene</b>	<b>OMIM gene ID</b>	<b>Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern</b>
AAAS	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
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
AASS	605113	Hyperlysinemia, 238700 (3), Autosomal recessive
ABAT	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
ABCA2	600047	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive
ABCC8	600509	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal dominant, Autosomal recessive; 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 dominant, Autosomal recessive
ABCC9	601439	Cardiomyopathy, dilated, 10, 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
ABCD1	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive
ABHD16A	142620	Spastic paraplegia 86, autosomal recessive, 619735 (3), Autosomal recessive
ABHD5	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ACAD9	611103	Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive
ACADM	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive
ACADS	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive
ACADSB	600301	2-methylbutyrylglycinuria, 610006 (3), Autosomal recessive
ACAT1	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive

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<i>ACO2</i>	100850	?Optic atrophy 9, 616289 (3), Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
<i>ACOX1</i>	609751	Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
<i>ACSF3</i>	614245	Combined malonic and methylmalonic aciduria, 614265 (3)
<i>ACSL4</i>	300157	Intellectual developmental disorder, X-linked 63, 300387 (3), X-linked dominant
<i>ACTB</i>	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
<i>ACTG1</i>	102560	Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant; Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant
<i>ACTL6A</i>	604958	No OMIM phenotype Intellectual disability (BAFopathy) (Pascolini (2020), Clin Genet 97(4):672-674 & Barom (2017), Hum Mutat 38(10):1365-1371), PMID: 31994175 - Autosomal dominant
<i>ACTL6B</i>	612458	Developmental and epileptic encephalopathy 76, 618468 (3), Autosomal recessive; Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant
<i>ACVR1</i>	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
<i>ACY1</i>	104620	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
<i>ADAM22</i>	603709	Developmental and epileptic encephalopathy 61, 617933 (3), Autosomal recessive
<i>ADAR</i>	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
<i>ADARB1</i>	601218	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 (3), Autosomal recessive
<i>ADAT3</i>	615302	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 (3), Autosomal recessive
<i>ADD3</i>	601568	Cerebral palsy, spastic quadriplegic, 3, 617008 (3), Autosomal recessive
<i>ADGRG1</i>	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
<i>ADGRL1</i>	616416	No OMIM phenotype ADGRL1 haploinsufficiency causes a variable spectrum of neurodevelopmental disorders in humans and alters synaptic activity and behavior in a mouse model (Vitobello (2022), Am J Hum Genet. 109(8):1436-1457), PMID: 35907405 - Autosomal dominant
<i>ADGRL2</i>	607018	No OMIM phenotype 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

<i>ADK</i>	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
<i>ADNP</i>	611386	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
<i>ADPRS</i>	610624	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive
<i>ADSL</i>	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
<i>AFF2</i>	300806	Intellectual developmental disorder, X-linked 109, 309548 (3), X-linked recessive
<i>AFF3</i>	601464	KINSSHIP syndrome, 619297 (3), Autosomal dominant
<i>AFF4</i>	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
<i>AFG3L2</i>	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
<i>AGA</i>	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
<i>AGO1</i>	606228	No OMIM phenotype De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. (Schalk (2021), J Med Genet. jmedgenet-2021-107751), PMID: 34930816 - Autosomal dominant
<i>AGO2</i>	606229	Lessel-Kreienkamp syndrome, 619149 (3), Autosomal dominant
<i>AGPAT2</i>	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
<i>AGTPBP1</i>	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive
<i>AHCY</i>	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
<i>AHDC1</i>	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
<i>AHI1</i>	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
<i>AIFM1</i>	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
<i>AIMP1</i>	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
<i>AIMP2</i>	600859	Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive
<i>AK1</i>	103000	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
<i>AKT3</i>	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant

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<i>ALDH18A1</i>	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
<i>ALDH3A2</i>	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
<i>ALDH4A1</i>	606811	Hyperprolinemia, type II, 239510 (3), Autosomal recessive
<i>ALDH5A1</i>	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
<i>ALDH7A1</i>	107323	Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive
<i>ALG1</i>	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
<i>ALG11</i>	613666	Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive
<i>ALG12</i>	607144	Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive
<i>ALG13</i>	300776	Developmental and epileptic encephalopathy 36, 300884 (3), X-linked
<i>ALG14</i>	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
<i>ALG2</i>	607905	Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
<i>ALG3</i>	608750	Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive
<i>ALG6</i>	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
<i>ALG8</i>	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
<i>ALG9</i>	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type II, 608776 (3), Autosomal recessive
<i>ALKBH8</i>	613306	Intellectual developmental disorder, autosomal recessive 71, 618504 (3), Autosomal recessive
<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>ALPL</i>	171760	Odontohypophosphatasia, 146300 (3), Autosomal dominant, Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, adult, 146300 (3), Autosomal dominant, Autosomal recessive
<i>ALX1</i>	601527	Frontonasal dysplasia 3, 613456 (3), Autosomal recessive
<i>ALX4</i>	605420	Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive
<i>AMACR</i>	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive

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<i>AMER1</i>	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
<i>AMMECR1</i>	300195	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive
<i>AMPD2</i>	102771	?Spastic paraplegia 63, 615686 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive
<i>AMT</i>	238310	Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>ANK2</i>	106410	Long QT syndrome 4, 600919 (3), Autosomal dominant; Cardiac arrhythmia, ankyrin-B-related, 600919 (3), Autosomal dominant
<i>ANK3</i>	600465	Intellectual developmental disorder, autosomal recessive 37, 615493 (3), Autosomal recessive
<i>ANKLE2</i>	616062	Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive
<i>ANKRD11</i>	611192	KBG syndrome, 148050 (3), Autosomal dominant
<i>ANKRD17</i>	615929	Chopra-Amiel-Gordon syndrome, 619504 (3), Autosomal dominant
<i>ANO10</i>	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
<i>ANTXR1</i>	606410	GAP0 syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
<i>AP1G1</i>	603533	Usmani-Riazuddin syndrome, autosomal recessive, 619548 (3); Usmani-Riazuddin syndrome, autosomal dominant, 619467 (3), Autosomal dominant
<i>AP1S1</i>	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
<i>AP1S2</i>	300629	Pettigrew syndrome, 304340 (3), X-linked recessive
<i>AP2M1</i>	601024	Intellectual developmental disorder 60 with seizures, 618587 (3), Autosomal dominant
<i>AP3B1</i>	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
<i>AP3B2</i>	602166	Developmental and epileptic encephalopathy 48, 617276 (3), Autosomal recessive
<i>AP3D1</i>	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
<i>AP4B1</i>	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
<i>AP4E1</i>	607244	Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive
<i>AP4M1</i>	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
<i>AP4S1</i>	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive
<i>AP5Z1</i>	613653	Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive
<i>APC2</i>	612034	Cortical dysplasia, complex, with other brain malformations 10, 618677 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 74, 617169 (3), Autosomal recessive
<i>APTX</i>	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
<i>ARCN1</i>	600820	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164 (3), Autosomal dominant

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<i>ARF1</i>	103180	Periventricular nodular heterotopia 8, 618185 (3), Autosomal dominant
<i>ARFGEF1</i>	604141	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964 (3), Autosomal dominant
<i>ARFGEF2</i>	605371	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
<i>ARG1</i>	608313	Argininemia, 207800 (3), Autosomal recessive
<i>ARHGAP31</i>	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
<i>ARHGEF9</i>	300429	Developmental and epileptic encephalopathy 8, 300607 (3), X-linked
<i>ARID1A</i>	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
<i>ARID1B</i>	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
<i>ARID2</i>	609539	Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant
<i>ARL13B</i>	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
<i>ARL6</i>	608845	Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Digenic recessive, Autosomal recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive
<i>ARMC9</i>	617612	Joubert syndrome 30, 617622 (3), Autosomal recessive
<i>ARSA</i>	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<i>ARSL</i>	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
<i>ARV1</i>	611647	Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive
<i>ARX</i>	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
<i>ASAH1</i>	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
<i>ASH1L</i>	607999	Intellectual developmental disorder, autosomal dominant 52, 617796 (3), Autosomal dominant
<i>ASL</i>	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
<i>ASNS</i>	108370	Asparagine synthetase deficiency, 615574 (3), Autosomal recessive
<i>ASPA</i>	608034	Canavan disease, 271900 (3), Autosomal recessive
<i>ASPM</i>	605481	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive
<i>ASS1</i>	603470	Citrullinemia, 215700 (3), Autosomal recessive
<i>ASXL1</i>	612990	Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant
<i>ASXL2</i>	612991	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
<i>ASXL3</i>	615115	Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant

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<i>ATAD1</i>	614452	Hyperekplexia 4, 618011 (3), Autosomal recessive
<i>ATAD3A</i>	612316	Harel-Yoon syndrome, 617183 (3), Autosomal dominant, Autosomal recessive; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
<i>ATCAY</i>	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
<i>ATG7</i>	608760	Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive
<i>ATIC</i>	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
<i>ATL1</i>	606439	Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant
<i>ATM</i>	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
<i>ATN1</i>	607462	Dentatorubral-pallidoluysian atrophy, 125370 (3), Autosomal dominant; Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 (3), Autosomal dominant
<i>ATP13A2</i>	610513	Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive
<i>ATP1A1</i>	182310	Hypomagnesemia, seizures, and mental retardation 2, 618314 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant
<i>ATP1A2</i>	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
<i>ATP1A3</i>	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
<i>ATP2A2</i>	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
<i>ATP2B1</i>	108731	Intellectual developmental disorder, autosomal dominant 66, 619910 (3), Autosomal dominant
<i>ATP6AP2</i>	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
<i>ATP6VOA1</i>	192130	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971 (3), Autosomal recessive; Developmental and epileptic encephalopathy 104, 619970 (3), Autosomal dominant

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<i>ATP6V0A2</i>	611716	Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive
<i>ATP6V1A</i>	607027	Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Developmental and epileptic encephalopathy 93, 618012 (3), Autosomal dominant
<i>ATP6V1B2</i>	606939	Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant; Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant
<i>ATP7A</i>	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
<i>ATP8A2</i>	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
<i>ATP9A</i>	609126	No OMIM phenotype Biallelic truncation variants in ATP9A are associated with a novel autosomal recessive neurodevelopmental disorder. (Mattioli (2021), NPJ Genom Med. 6(1):94), PMID: 34764295 - Autosomal recessive
<i>ATPAF2</i>	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
<i>ATR</i>	601215	Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant
<i>ATRX</i>	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
<i>ATXN2</i>	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
<i>AUH</i>	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
<i>AUTS2</i>	607270	Intellectual developmental disorder, autosomal dominant 26, 615834 (3), Autosomal dominant
<i>AVPR2</i>	300538	Diabetes insipidus, nephrogenic, 1, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive
<i>B3GALNT2</i>	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
<i>B3GALT6</i>	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
<i>B3GLCT</i>	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
<i>B4GALNT1</i>	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
<i>B4GALT1</i>	137060	Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive



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<i>B4GALT7</i>	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
<i>B4GAT1</i>	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
<i>B9D1</i>	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
<i>B9D2</i>	611951	?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive
<i>BAZ2B</i>	605683	No OMIM phenotype Developmental delay, intellectual disability, and autism spectrum disorder (Scott (2020), Hum Mutat), PMID: 31999386 - Autosomal dominant
<i>BBS1</i>	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
<i>BBS10</i>	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
<i>BBS12</i>	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
<i>BBS2</i>	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive
<i>BBS4</i>	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
<i>BBS5</i>	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
<i>BBS7</i>	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
<i>BBS9</i>	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
<i>BCAP31</i>	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
<i>BCAS3</i>	607470	Hengel-Marroofian-Schols syndrome, 619641 (3), Autosomal recessive
<i>BCKDHA</i>	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
<i>BCKDHB</i>	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
<i>BCKDK</i>	614901	Branched-chain keto acid dehydrogenase kinase deficiency, 614923 (3)
<i>BCL11A</i>	606557	Dias-Logan syndrome, 617101 (3), Autosomal dominant
<i>BCL11B</i>	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
<i>BCOR</i>	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
<i>BCORL1</i>	300688	Shukla-Vernon syndrome, 301029 (3), X-linked recessive
<i>BCS1L</i>	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive

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<i>BICD2</i>	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
<i>BICRA</i>	605690	Coffin-Siris syndrome 12, 619325 (3), Autosomal dominant
<i>BLM</i>	604610	Bloom syndrome, 210900 (3), Autosomal recessive
<i>BLOC1S1</i>	601444	No OMIM phenotype 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
<i>BLTP1</i> ( <i>KIAA1109</i> )	611565	Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive
<i>BMP4</i>	112262	Orofacial cleft 11, 600625 (3); Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant
<i>BOLA3</i>	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
<i>BPTF</i>	601819	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755 (3), Autosomal dominant
<i>BRAF</i>	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)
<i>BRAT1</i>	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
<i>BRD4</i>	608749	No OMIM phenotype 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
<i>BRF1</i>	604902	Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive
<i>BRPF1</i>	602410	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant
<i>BRSK2</i>	609236	No OMIM phenotype Neurodevelopmental disorder (Hiatt (2019), Am J Hum Genet 104(4):701-708), PMID: 30879638 - Autosomal dominant
<i>BRWD3</i>	300553	Intellectual developmental disorder, X-linked 93, 300659 (3), X-linked recessive
<i>BSCL2</i>	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
<i>BTB</i>	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
<i>BUB1B</i>	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
<i>C12orf4</i>	616082	Intellectual developmental disorder, autosomal recessive 66, 618221 (3), Autosomal recessive
<i>C12orf57</i>	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
<i>C2CD3</i>	615944	Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
<i>CA2</i>	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
<i>CA5A</i>	114761	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive
<i>CA8</i>	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3), Autosomal recessive
<i>CACNA1A</i>	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
<i>CACNA1B</i>	601012	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive
<i>CACNA1C</i>	114205	Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3), Autosomal dominant; Brugada syndrome 3, 611875 (3)
<i>CACNA1D</i>	114206	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive
<i>CACNA1E</i>	601013	Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant
<i>CACNA1G</i>	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant
<i>CACNA1H</i>	607904	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 (3); Hyperaldosteronism, familial, type IV, 617027 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 (3)
<i>CACNA1I</i>	608230	No OMIM phenotype CACNA1I gain-of-function mutations differentially affect channel gating and cause neurodevelopmental disorders. (El Ghaleb (2021), Brain. 144(7):2092-2106), PMID: 33704440 - Autosomal dominant

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		No OMIM phenotype
<i>CACNA2D1</i>	114204	Genomic aberrations of the CACNA2D1 gene in three patients with epilepsy and intellectual disability (Vergult (2015), Eur J Hum Genet 23, 628–632), PMID: 25074461
<i>CACNA2D2</i>	607082	Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive
<i>CACNB4</i>	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
<i>CACNG2</i>	602911	?Intellectual developmental disorder, autosomal dominant 10, 614256 (3), Autosomal dominant
<i>CAD</i>	114010	Developmental and epileptic encephalopathy 50, 616457 (3), Autosomal recessive
<i>CAMK2A</i>	114078	Intellectual developmental disorder, autosomal dominant 53, 617798 (3), Autosomal dominant; ?Intellectual developmental disorder, autosomal recessive 63, 618095 (3), Autosomal recessive
<i>CAMK2B</i>	607707	Intellectual developmental disorder, autosomal dominant 54, 617799 (3), Autosomal dominant
<i>CAMK2G</i>	602123	Intellectual developmental disorder, autosomal dominant 59, 618522 (3), Autosomal dominant
		No OMIM phenotype
<i>CAMK4</i>	114080	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
<i>CAMTA1</i>	611501	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 (3), Autosomal dominant
<i>CAPN10</i>	605286	{Diabetes mellitus, noninsulin-dependent 1}, 601283 (3)
<i>CAPN15</i>	603267	Oculogastrointestinal neurodevelopmental syndrome, 619318 (3), Autosomal recessive
<i>CARS1</i>	123859	Microcephaly, developmental delay, and brittle hair syndrome, 618891 (3), Autosomal recessive
<i>CARS2</i>	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
<i>CASK</i>	300172	Intellectual developmental disorder, with or without nystagmus, 300422 (3); Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked dominant; FG syndrome 4, 300422 (3)
<i>CBL</i>	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation
<i>CBS</i>	613381	Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive
<i>CC2D1A</i>	610055	Intellectual developmental disorder, autosomal recessive 3, 608443 (3), Autosomal recessive

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<i>CC2D2A</i>	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
<i>CCBE1</i>	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
<i>CCDC115</i>	613734	Congenital disorder of glycosylation, type Ilo, 616828 (3), Autosomal recessive
<i>CCDC174</i>	616735	Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive
<i>CCDC22</i>	300859	Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive
<i>CCDC47</i>	618260	Trichohepatoneurodevelopmental syndrome, 618268 (3), Autosomal recessive
<i>CCDC88A</i>	609736	?PEHO syndrome-like, 617507 (3), Autosomal recessive
<i>CCDC88C</i>	611204	?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive
<i>CCM2</i>	607929	Cerebral cavernous malformations-2, 603284 (3), Autosomal dominant
<i>CCND2</i>	123833	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant
<i>CCNK</i>	603544	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147 (3), Autosomal dominant
<i>CDC42</i>	116952	Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant
<i>CDC42BPB</i>	614062	Chilton-Okur-Chung neurodevelopmental syndrome, 619841 (3)
<i>CDH11</i>	600023	Teebi hypertelorism syndrome 2, 619736 (3), Autosomal dominant; Elshahy-Waters syndrome, 211380 (3), Autosomal recessive
<i>CDH15</i>	114019	Intellectual developmental disorder, autosomal dominant 3, 612580 (3)
<i>CDH2</i>	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
<i>CDK10</i>	603464	Al Kaissi syndrome, 617694 (3), Autosomal recessive
<i>CDK13</i>	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant
<i>CDK19</i>	614720	Developmental and epileptic encephalopathy 87, 618916 (3), Autosomal dominant
<i>CDK5RAP2</i>	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
<i>CDK6</i>	603368	?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive
<i>CDK8</i>	603184	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 (3), Autosomal dominant
<i>CDKL5</i>	300203	Developmental and epileptic encephalopathy 2, 300672 (3), X-linked dominant

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<i>CDKN1C</i>	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
<i>CDON</i>	608707	Holoprosencephaly 11, 614226 (3), Autosomal dominant
<i>CELF2</i>	602538	Developmental and epileptic encephalopathy 97, 619561 (3), Autosomal dominant
<i>CELSR3</i>	604264	No OMIM phenotype 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
<i>CENPF</i>	600236	Stromme syndrome, 243605 (3), Autosomal recessive
<i>CENPJ</i>	609279	Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive
<i>CEP104</i>	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 77, 619988 (3), Autosomal recessive
<i>CEP120</i>	613446	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive
<i>CEP135</i>	611423	Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive
<i>CEP152</i>	613529	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
<i>CEP290</i>	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
<i>CEP41</i>	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
<i>CEP55</i>	610000	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive
<i>CEP57</i>	607951	Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive
<i>CEP63</i>	614724	?Seckel syndrome 6, 614728 (3), Autosomal recessive
<i>CEP83</i>	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
<i>CEP85L</i>	618865	Lissencephaly 10, 618873 (3), Autosomal dominant
<i>CERS1</i>	606919	Epilepsy, progressive myoclonic, 8, 616230 (3), Autosomal recessive
<i>CERT1</i>	604677	Intellectual developmental disorder, autosomal dominant 34, 616351 (3), Autosomal dominant
<i>CHAMP1</i>	616327	Intellectual developmental disorder, autosomal dominant 40, 616579 (3), Autosomal dominant
<i>CHD1</i>	602118	Pilarowski-Bjornsson syndrome, 617682 (3), Autosomal dominant
<i>CHD2</i>	602119	Developmental and epileptic encephalopathy 94, 615369 (3), Autosomal dominant
<i>CHD3</i>	602120	Snijders Blok-Campeau syndrome, 618205 (3), Autosomal dominant

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<i>CHD4</i>	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
<i>CHD5</i>	610771	Parenti-Mignot neurodevelopmental syndrome, 619873 (3), Autosomal dominant
<i>CHD7</i>	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
<i>CHD8</i>	610528	Intellectual developmental disorder with autism and macrocephaly, 615032 (3), Autosomal dominant
<i>CHKB</i>	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
<i>CHMP1A</i>	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
<i>CHRNA2</i>	118502	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant
<i>CHRNA4</i>	118504	{Nicotine addiction, susceptibility to}, 188890 (3); Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant
<i>CHRN2</i>	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
<i>CIC</i>	612082	Intellectual developmental disorder, autosomal dominant 45, 617600 (3), Autosomal dominant
<i>CILK1</i>	612325	{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant; Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive
<i>CIT</i>	605629	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive
<i>CKAP2L</i>	616174	Filippi syndrome, 272440 (3), Autosomal recessive
<i>CLCN3</i>	600580	Neurodevelopmental disorder with seizures and brain abnormalities, 619517 (3), Autosomal recessive; Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512 (3), Autosomal dominant
<i>CLCN4</i>	302910	Raynaud-Claes syndrome, 300114 (3), X-linked dominant
<i>CLCNKB</i>	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
<i>CLDN11</i>	601326	Leukodystrophy, hypomyelinating, 22, 619328 (3), Autosomal dominant
<i>CLDN16</i>	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
<i>CLDN19</i>	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
<i>CLIC2</i>	300138	?Intellectual developmental disorder, X-linked syndromic 32, 300886 (3), X-linked recessive
<i>CLN3</i>	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
<i>CLN5</i>	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
<i>CLN6</i>	606725	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive
<i>CLN8</i>	607837	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive
<i>CLP1</i>	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive

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<i>CLPB</i>	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
<i>CLTC</i>	118955	Intellectual developmental disorder, autosomal dominant 56, 617854 (3), Autosomal dominant
<i>CNKS2</i>	300724	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008 (3), X-linked
<i>CNNM2</i>	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation, 616418 (3), Autosomal dominant, Autosomal recessive
<i>CNOT1</i>	604917	Vissers-Bodmer syndrome, 619033 (3), Autosomal dominant; Holoprosencephaly 12, with or without pancreatic agenesis, 618500 (3), Autosomal dominant
<i>CNOT2</i>	604909	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 (3), Autosomal dominant
<i>CNOT3</i>	604910	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 (3), Autosomal dominant
<i>CNPY3</i>	610774	Developmental and epileptic encephalopathy 60, 617929 (3), Autosomal recessive
<i>CNTN2</i>	190197	?Epilepsy, myoclonic, familial adult, 5, 615400 (3), Autosomal recessive
<i>CNTNAP1</i>	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
<i>CNTNAP2</i>	604569	Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; {Autism susceptibility 15}, 612100 (3)
<i>COA8</i>	616003	Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive
<i>COASY</i>	609855	Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
<i>COG1</i>	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
<i>COG4</i>	606976	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant
<i>COG5</i>	606821	Congenital disorder of glycosylation, type IIIi, 613612 (3), Autosomal recessive
<i>COG6</i>	606977	Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive
<i>COG7</i>	606978	Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive
<i>COG8</i>	606979	Congenital disorder of glycosylation, type IIh, 611182 (3)
<i>COL18A1</i>	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant



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<i>COL4A1</i>	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
<i>COL4A2</i>	120090	Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3)
<i>COLEC11</i>	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
<i>COPB1</i>	600959	Baralle-Macken syndrome, 619255 (3), Autosomal recessive
<i>COPB2</i>	606990	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884 (3), Autosomal dominant; ?Microcephaly 19, primary, autosomal recessive, 617800 (3), Autosomal recessive
<i>COQ2</i>	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
<i>COQ4</i>	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive
<i>COQ5</i>	616359	?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive
<i>COQ8A</i>	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
<i>COQ9</i>	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
<i>COX10</i>	602125	Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive
<i>COX15</i>	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
<i>COX6B1</i>	124089	Mitochondrial complex IV deficiency, nuclear type 7, 619051 (3), Autosomal recessive
<i>CPA6</i>	609562	Febrile seizures, familial, 11, 614418 (3), Autosomal recessive; Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal dominant, Autosomal recessive
<i>CPE</i>	114855	BDV syndrome, 619326 (3), Autosomal recessive
<i>CPLANE1</i>	614571	Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive
<i>CPLX1</i>	605032	Developmental and epileptic encephalopathy 63, 617976 (3), Autosomal recessive
<i>CPS1</i>	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
<i>CPSF3</i>	606029	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876 (3), Autosomal recessive
<i>CPT2</i>	600650	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal dominant, Autosomal recessive; 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 dominant, Autosomal recessive

<i>CRADD</i>	603454	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive
<i>CRB2</i>	609720	Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive
<i>CRBN</i>	609262	Intellectual developmental disorder, autosomal recessive 2, 607417 (3), Autosomal recessive
<i>CREBBP</i>	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
<i>CRH</i>	122560	No OMIM phenotype Corticotropin-releasing Hormone - Seizures.
<i>CRLF1</i>	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
<i>CRPPA</i>	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
<i>CSDE1</i>	191510	No OMIM phenotype Autism and related neurodevelopmental disabilities (Guo (2019), Sci Adv 25;5(9):eaax2166), PMID: 31579823 - Autosomal dominant
<i>CSNK1G1</i>	606274	No OMIM phenotype 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
<i>CSNK2A1</i>	115440	Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant
<i>CSNK2B</i>	115441	Poirier-Bienvenu neurodevelopmental syndrome, 618732 (3), Autosomal dominant
<i>CSPP1</i>	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
<i>CSTB</i>	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
<i>CTBP1</i>	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
<i>CTC1</i>	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
<i>CTCF</i>	604167	Intellectual developmental disorder, autosomal dominant 21, 615502 (3), Autosomal dominant
<i>CTDP1</i>	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
<i>CTNNA2</i>	114025	Cortical dysplasia, complex, with other brain malformations 9, 618174 (3), Autosomal recessive

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<i>CTNNB1</i>	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)
<i>CTNND1</i>	601045	Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant
<i>CTNND2</i>	604275	No OMIM phenotype Autism (Turner (2015) Nature 520,51), Autosomal dominant Intellectual disability (Hofmeister (2015) J Med Genet 52,111) - Autosomal dominant
<i>CTSA</i>	613111	Galactosialidosis, 256540 (3), Autosomal recessive
<i>CTSD</i>	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
<i>CTSF</i>	603539	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362 (3), Autosomal recessive
<i>CTU2</i>	617057	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 (3), Autosomal recessive
<i>CUBN</i>	602997	[Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imlerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive
<i>CUL3</i>	603136	Neurodevelopmental disorder with or without autism or seizures, 619239 (3), Autosomal dominant; Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant
<i>CUL4B</i>	300304	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354 (3), X-linked recessive
<i>CUX1</i>	116896	Global developmental delay with or without impaired intellectual development, 618330 (3), Autosomal dominant
<i>CUX2</i>	610648	Developmental and epileptic encephalopathy 67, 618141 (3), Autosomal dominant
<i>CWC27</i>	617170	Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive
<i>CWF19L1</i>	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
<i>CYB5R3</i>	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
<i>CYC1</i>	123980	Mitochondrial complex III deficiency, nuclear type 6, 615453 (3), Autosomal recessive
<i>CYFIP2</i>	606323	Developmental and epileptic encephalopathy 65, 618008 (3), Autosomal dominant
<i>CYP27A1</i>	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<i>CYP2U1</i>	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
<i>D2HGDH</i>	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
<i>DAG1</i>	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

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<i>DARS1</i>	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
<i>DARS2</i>	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
<i>DBT</i>	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
<i>DCAF17</i>	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
<i>DCC</i>	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
<i>DCHS1</i>	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
<i>DCPS</i>	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive
<i>DCX</i>	300121	Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked
<i>DDB1</i>	600045	White-Kernohan syndrome, 619426 (3), Autosomal dominant
<i>DDC</i>	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
<i>DDHD2</i>	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
<i>DDX11</i>	601150	Warsaw breakage syndrome, 613398 (3), Autosomal recessive
<i>DDX23</i>	612172	No OMIM phenotype 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
<i>DDX3X</i>	300160	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked recessive, X-linked dominant
<i>DDX59</i>	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
<i>DDX6</i>	600326	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653 (3), Autosomal dominant
<i>DEAF1</i>	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
<i>DEGS1</i>	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
<i>DENND5A</i>	617278	Developmental and epileptic encephalopathy 49, 617281 (3), Autosomal recessive
<i>DEPDC5</i>	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant
<i>DHCR24</i>	606418	Desmosterolosis, 602398 (3), Autosomal recessive
<i>DHCR7</i>	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive

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<i>DHDDS</i>	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
<i>DHFR</i>	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
<i>DHPS</i>	600944	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 (3), Autosomal recessive
<i>DHTKD1</i>	614984	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant; Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 (3), Autosomal recessive
<i>DHX16</i>	603405	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant
<i>DHX30</i>	616423	Neurodevelopmental disorder with severe motor impairment and absent language, 617804 (3), Autosomal dominant
<i>DHX37</i>	617362	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 (3), Autosomal recessive; 46, XY sex reversal 11, 273250 (3), Autosomal dominant
<i>DIAPH1</i>	602121	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
<i>DIS3L2</i>	614184	Perlman syndrome, 267000 (3), Autosomal recessive
<i>DKC1</i>	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
<i>DLAT</i>	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
<i>DLG</i>	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<i>DLG3</i>	300189	Intellectual developmental disorder, X-linked 90, 300850 (3), X-linked recessive
<i>DLG4</i>	602887	Intellectual developmental disorder, autosomal dominant 62, 618793 (3), Autosomal dominant
<i>DLL1</i>	606582	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 (3), Autosomal dominant
<i>DMD</i>	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
<i>DMPK</i>	605377	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
<i>DMXL2</i>	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
<i>DNAJC12</i>	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
<i>DNAJC19</i>	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
<i>DNAJC5</i>	611203	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350 (3), Autosomal dominant

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<i>DNM1</i>	602377	Developmental and epileptic encephalopathy 31, 616346 (3), Autosomal dominant
<i>DNM1L</i>	603850	Optic atrophy 5, 610708 (3), Autosomal dominant; Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal dominant, Autosomal recessive
<i>DNMT3A</i>	602769	Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant; Acute myeloid leukemia, somatic, 601626 (3); Heyn-Sproul-Jackson syndrome, 618724 (3), Autosomal dominant
<i>DNMT3B</i>	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant
<i>DOCK3</i>	603123	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive
<i>DOCK6</i>	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
<i>DOCK7</i>	615730	Developmental and epileptic encephalopathy 23, 615859 (3), Autosomal recessive
<i>DOCK8</i>	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
<i>DOLK</i>	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
<i>DONSON</i>	611428	Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive; Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive
<i>DPAGT1</i>	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive; Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive
<i>DPF2</i>	601671	Coffin-Siris syndrome 7, 618027 (3), Autosomal dominant
<i>DPH1</i>	603527	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive
<i>DPM1</i>	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
<i>DPM2</i>	603564	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive
<i>DPP6</i>	126141	Intellectual developmental disorder, autosomal dominant 33, 616311 (3); {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant
<i>DPYD</i>	612779	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
<i>DPYS</i>	613326	Dihydropyrimidinuria, 222748 (3), Autosomal recessive
<i>DPYSL5</i>	608383	Ritscher-Schinzel syndrome 4, 619435 (3), Autosomal dominant
		No OMIM phenotype
<i>DSCAM</i>	602523	The contribution of de novo coding mutations to autism spectrum disorder (Iossifov (2014) Nature 515(7526):216-21), PMID: 25363768 - Autosomal dominant

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<i>DYM</i>	607461	Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive
<i>DYNC1H1</i>	600112	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 13, 614563 (3), Autosomal dominant
<i>DYNC1I2</i>	603331	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492 (3), Autosomal recessive
<i>DYRK1A</i>	600855	Intellectual developmental disorder, autosomal dominant 7, 614104 (3), Autosomal dominant
<i>EARS2</i>	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
<i>EBF3</i>	607407	Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant
<i>EBP</i>	300205	MEND syndrome, 300960 (3), X-linked recessive; Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant
<i>ECHS1</i>	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
<i>EDC3</i>	609842	?Intellectual developmental disorder, autosomal recessive 50, 616460 (3), Autosomal recessive
<i>EDEM3</i>	610214	Congenital disorder of glycosylation, type 2V, 619493 (3), Autosomal recessive
<i>EED</i>	605984	Cohen-Gibson syndrome, 617561 (3), Autosomal dominant
<i>EEF1A2</i>	602959	Developmental and epileptic encephalopathy 33, 616409 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 38, 616393 (3), Autosomal dominant
<i>EFNB2</i>	600527	No OMIM phenotype EFNB2 haploinsufficiency causes a syndromic neurodevelopmental disorder. (Lévy (2018), Clin Genet. 93(6):1141-1147), PMID: 29508392 - Autosomal dominant
<i>EFTUD2</i>	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
<i>EHMT1</i>	607001	Kleefstra syndrome 1, 610253 (3), Autosomal dominant
<i>EIF2AK2</i>	176871	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 (3), Autosomal dominant; Dystonia 33, 619687 (3), Autosomal dominant, Autosomal recessive
<i>EIF2AK3</i>	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
<i>EIF2B1</i>	606686	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B2</i>	606454	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
<i>EIF2B3</i>	606273	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B4</i>	606687	Ovarioleukodystrophy, 603896 (3), Autosomal recessive; Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive

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<i>EIF2B5</i>	603945	Ovariëleukodystrofie, 603896 (3), Autosomaal recessief; Leukoencefalopathie met verdwijnend witte materie, 603896 (3), Autosomaal recessief
<i>EIF2S3</i>	300161	MEHMO syndroom, 300148 (3), X-geketend recessief
<i>EIF3F</i>	603914	Intellectuele ontwikkelingsstoornis, autosomaal recessief 67, 618295 (3), Autosomaal recessief
<i>EIF4A3</i>	608546	Robin sequentie met kloof in de kaak en ledemaatafwijkingen, 268305 (3), Autosomaal recessief
<i>EIF5A</i>	600187	Faundes-Banka syndroom, 619376 (3), Autosomaal dominant
<i>ELAC2</i>	605367	{Prostaatcarcinoom, erfelijk, 2, susceptibiliteit voor}, 614731 (3); Gecombineerde oxidatieve fosforyleringsdeficiëntie 17, 615440 (3), Autosomaal recessief
<i>ELOVL4</i>	605512	Spinocerebellaire ataxie 34, 133190 (3), Autosomaal dominant; Stargardt ziekte 3, 600110 (3), Autosomaal dominant; Ichthyosis, spastische quadriplegie, en mentale retardatie, 614457 (3), Autosomaal recessief
<i>ELP2</i>	616054	Intellectuele ontwikkelingsstoornis, autosomaal recessief 58, 617270 (3), Autosomaal recessief
<i>EMC1</i>	616846	Cerebellaire atrofie, visuele beperking, en psychomotorische retardatie, 616875 (3), Autosomaal recessief
<i>EMC10</i>	614545	Neurodevelopmentale stoornis met dysmorphe gezichten en variabele aanvallen, 619264 (3), Autosomaal recessief
<i>EML1</i>	602033	Band heterotopia, 600348 (3), Autosomaal recessief
<i>EMX2</i>	600035	Schizencefalie, 269160 (3)
<i>ENTPD1</i>	601752	Spastische paraplegie 64, autosomaal recessief, 615683 (3), Autosomaal recessief
<i>EP300</i>	602700	Menke-Hennekam syndroom 2, 618333 (3), Autosomaal dominant; Colorectale kanker, somatisch, 114500 (3); Rubinstein-Taybi syndroom 2, 613684 (3), Autosomaal dominant
<i>EPB41L1</i>	602879	?Intellectuele ontwikkelingsstoornis, autosomaal dominant 11, 614257 (3), Autosomaal dominant
<i>EPG5</i>	615068	Vici syndroom, 242840 (3), Autosomaal recessief
<i>EPHA7</i>	602190	No OMIM fenotype EPHA7 haploinsufficiëntie is geassocieerd met een neurodevelopmentale stoornis. (Lévy (2021), Clin Genet. 100(4):396-404), PMID: 34176129 - Autosomaal dominant
<i>EPM2A</i>	607566	Epilepsie, progressieve myoclonische 2A (Lafora), 254780 (3), Autosomaal recessief
<i>EPRS1</i>	138295	Leukodystrofie, hypomyeliniserend, 15, 617951 (3), Autosomaal recessief
<i>ERBB4</i>	600543	Amyotrofische laterale sclerose 19, 615515 (3), Autosomaal dominant
<i>ERCC1</i>	126380	Cerebrooculofaciale skeletale syndroom 4, 610758 (3), Autosomaal recessief
<i>ERCC2</i>	126340	Xeroderma pigmentosum, groep D, 278730 (3), Autosomaal recessief; Trichothiodystrofie 1, fotosensitief, 601675 (3), Autosomaal recessief; ?Cerebrooculofaciale skeletale syndroom 2, 610756 (3), Autosomaal recessief
<i>ERCC3</i>	133510	Trichothiodystrofie 2, fotosensitief, 616390 (3), Autosomaal recessief; Xeroderma pigmentosum, groep B, 610651 (3), Autosomaal recessief



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<i>ERCC5</i>	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
<i>ERCC6</i>	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), Autosomal dominant, Somatic mutation
<i>ERCC6L2</i>	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
<i>ERCC8</i>	609412	UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive
<i>ERLIN2</i>	611605	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
<i>ESCO2</i>	609353	Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive
<i>ETFA</i>	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
<i>ETFB</i>	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
<i>ETFDH</i>	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
<i>ETHE1</i>	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
<i>EXOC2</i>	615329	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306 (3), Autosomal recessive
<i>EXOC7</i>	608163	Neurodevelopmental disorder with seizures and brain atrophy, 619072 (3), Autosomal recessive
<i>EXOSC2</i>	602238	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive
<i>EXOSC3</i>	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
<i>EXOSC8</i>	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
<i>EXOSC9</i>	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
<i>EXT2</i>	608210	Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant
<i>EXTL3</i>	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
<i>EZH2</i>	601573	Weaver syndrome, 277590 (3), Autosomal dominant
<i>FA2H</i>	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
<i>FAM111A</i>	615292	Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant; Gracile bone dysplasia, 602361 (3), Autosomal dominant
<i>FAM149B1</i>	618413	Joubert syndrome 36, 618763 (3), Autosomal recessive

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<i>FAM20C</i>	611061	Raine syndrome, 259775 (3), Autosomal recessive
<i>FAM50A</i>	300453	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261 (3), X-linked recessive
<i>FANCD2</i>	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
<i>FAR1</i>	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant
<i>FARS2</i>	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
<i>FARSA</i>	602918	?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive
<i>FARSB</i>	609690	Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive
<i>FAT4</i>	612411	Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive
<i>FBRSL1</i>	No OMIM gene	No OMIM phenotype 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
<i>FBXL3</i>	605653	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 (3), Autosomal recessive
<i>FBXL4</i>	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
<i>FBXO11</i>	607871	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 (3), Autosomal dominant
<i>FBXO28</i>	609100	Developmental and epileptic encephalopathy 100, 619777 (3), Autosomal dominant
<i>FBXO31</i>	609102	?Intellectual developmental disorder, autosomal recessive 45, 615979 (3), Autosomal recessive
<i>FBXW11</i>	605651	Neurodevelopmental, jaw, eye, and digital syndrome, 618914 (3), Autosomal dominant
<i>FBXW7</i>	606278	No OMIM phenotype Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome (Stephenson (2022), Am J Hum Genet. 109(4):601-617), PMID: 35395208 - Autosomal dominant
<i>FDFT1</i>	184420	Squalene synthase deficiency, 618156 (3), Autosomal recessive
<i>FGD1</i>	300546	Intellectual developmental disorder, X-linked syndromic 16, 305400 (3), X-linked recessive; Aarskog-Scott syndrome, 305400 (3), X-linked recessive
<i>FGF12</i>	601513	Developmental and epileptic encephalopathy 47, 617166 (3), Autosomal dominant
<i>FGF13</i>	300070	Developmental and epileptic encephalopathy 90, 301058 (3), X-linked recessive, X-linked dominant
<i>FGF14</i>	601515	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant

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<i>FGFR1</i>	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; Trigenocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
<i>FGFR2</i>	176943	Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3); 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); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant
<i>FGFR3</i>	134934	Muenke syndrome, 602849 (3), Autosomal dominant; SADDAN, 616482 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); CATSHL syndrome, 610474 (3), Autosomal dominant, Autosomal recessive; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Bladder cancer, somatic, 109800 (3); Achondroplasia, 100800 (3), Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant
<i>FH</i>	136850	Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive
<i>FIBP</i>	608296	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive
<i>FIG4</i>	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
<i>FKRP</i>	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), 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

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<i>FKTN</i>	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; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive
<i>FLNA</i>	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
<i>FLVCR1</i>	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
<i>FLVCR2</i>	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive
<i>FMN2</i>	606373	Intellectual developmental disorder, autosomal recessive 47, 616193 (3), Autosomal recessive
<i>FMR1</i>	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
<i>FOLR1</i>	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
<i>FOXG1</i>	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
<i>FOXP1</i>	605515	Intellectual developmental disorder with language impairment with or without autistic features, 613670 (3), Autosomal dominant
<i>FOXP2</i>	605317	Speech-language disorder-1, 602081 (3), Autosomal dominant
<i>FOXRED1</i>	613622	Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive
<i>FRAS1</i>	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
<i>FREM2</i>	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive
<i>FRMD4A</i>	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
<i>FRMPD4</i>	300838	Intellectual developmental disorder, X-linked 104, 300983 (3), X-linked
<i>FRRS1L</i>	604574	Developmental and epileptic encephalopathy 37, 616981 (3), Autosomal recessive
<i>FTCD</i>	606806	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
<i>FTO</i>	610966	Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive
<i>FTSJ1</i>	300499	Intellectual developmental disorder, X-linked 9, 309549 (3), X-linked recessive
<i>FUCA1</i>	612280	Fucosidosis, 230000 (3), Autosomal recessive
<i>FUT8</i>	602589	Congenital disorder of glycosylation with defective fucosylation 1, 618005 (3), Autosomal recessive

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<i>FXVD2</i>	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
		No OMIM phenotype
<i>FZR1</i>	603619	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies (Manivannan (2022), Brain. 145(5):1684-1697), PMID: 34788397 - Autosomal dominant
<i>GABBR2</i>	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
<i>GABRA1</i>	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)
<i>GABRA2</i>	137140	Developmental and epileptic encephalopathy 78, 618557 (3), Autosomal dominant; {Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial
		No OMIM phenotype
<i>GABRA3</i>	305660	Epileptic Encephalopathy (Bamborschke (2019), Neuropediatrics 50(6)), PMID: 31319422 - Autosomal dominant
<i>GABRA5</i>	137142	Developmental and epileptic encephalopathy 79, 618559 (3), Autosomal dominant
<i>GABRB1</i>	137190	Developmental and epileptic encephalopathy 45, 617153 (3), Autosomal dominant
<i>GABRB2</i>	600232	Developmental and epileptic encephalopathy 92, 617829 (3), Autosomal dominant
<i>GABRB3</i>	137192	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Developmental and epileptic encephalopathy 43, 617113 (3), Autosomal dominant
<i>GABRD</i>	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
<i>GABRG2</i>	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
<i>GAD1</i>	605363	Developmental and epileptic encephalopathy 89, 619124 (3), Autosomal recessive
<i>GAL</i>	137035	?Epilepsy, familial temporal lobe, 8, 616461 (3), Autosomal dominant
<i>GALC</i>	606890	Krabbe disease, 245200 (3), Autosomal recessive
<i>GALE</i>	606953	Galactose epimerase deficiency, 230350 (3), Autosomal recessive
<i>GALNT2</i>	602274	Congenital disorder of glycosylation, type II, 618885 (3), Autosomal recessive
<i>GALT</i>	606999	Galactosemia, 230400 (3), Autosomal recessive
<i>GAMT</i>	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
<i>GATAD2B</i>	614998	GAND syndrome, 615074 (3), Autosomal dominant

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<i>GATM</i>	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant
<i>GBA1 (GBA)</i>	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
<i>GCDH</i>	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
<i>GCH1</i>	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal dominant, Autosomal recessive; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
<i>GCK</i>	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
<i>GCSH</i>	238330	?Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>GDI1</i>	300104	Intellectual developmental disorder, X-linked 41, 300849 (3), X-linked dominant
<i>GEMIN4</i>	606969	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913 (3), Autosomal recessive
<i>GEMIN5</i>	607005	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 (3), Autosomal recessive
<i>GFAP</i>	137780	Alexander disease, 203450 (3), Autosomal dominant
<i>GFER</i>	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive
<i>GFM1</i>	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
<i>GFM2</i>	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
<i>GJA1</i>	121014	Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Atrioventricular septal defect 3, 600309 (3), Autosomal dominant
<i>GJB1</i>	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
<i>GJC2</i>	608803	Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive

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<i>GK</i>	300474	Glycerol kinase deficiency, 307030 (3), X-linked recessive
<i>GLB1</i>	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
<i>GLDC</i>	238300	Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>GLI2</i>	165230	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant
<i>GLI3</i>	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
<i>GLIS3</i>	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
<i>GLRA1</i>	138491	Hyperekplexia 1, 149400 (3), Autosomal dominant, Autosomal recessive
<i>GLRB</i>	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
<i>GLS</i>	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
<i>GLUD1</i>	138130	Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant
<i>GLUL</i>	138290	Glutamine deficiency, congenital, 610015 (3), Autosomal recessive
<i>GLYCTK</i>	610516	D-glycemic aciduria, 220120 (3), Autosomal recessive
<i>GM2A</i>	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
<i>GMNN</i>	602842	Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant
<i>GMPPA</i>	615495	Alacrima, achalasia, and mental retardation syndrome, 615510 (3), Autosomal recessive
<i>GMPPB</i>	615320	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive
<i>GNAI1</i>	139310	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854 (3), Autosomal dominant
<i>GNAO1</i>	139311	Developmental and epileptic encephalopathy 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
<i>GNAQ</i>	600998	Capillary malformations, congenital, 1, somatic, mosaic, 163000 (3); Sturge-Weber syndrome, somatic, mosaic, 185300 (3)

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<i>GNAS</i>	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
<i>GNB1</i>	139380	Myelodysplastic syndrome, somatic, 614286 (3); Leukemia, acute lymphoblastic, somatic, 613065 (3); Intellectual developmental disorder, autosomal dominant 42, 616973 (3), Autosomal dominant
<i>GNB2</i>	139390	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 (3), Autosomal dominant; ?Sick sinus syndrome 4, 619464 (3), Autosomal dominant
<i>GNB5</i>	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
<i>GNPAT</i>	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
<i>GNPTAB</i>	607840	Mucopolipidosis III alpha/beta, 252600 (3), Autosomal recessive; Mucopolipidosis II alpha/beta, 252500 (3), Autosomal recessive
<i>GNPTG</i>	607838	Mucopolipidosis III gamma, 252605 (3), Autosomal recessive
<i>GNS</i>	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
<i>GOSR2</i>	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
<i>GOT2</i>	138150	Developmental and epileptic encephalopathy 82, 618721 (3), Autosomal recessive
<i>GPAA1</i>	603048	Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive
<i>GPC3</i>	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
<i>GPC4</i>	300168	Keipert syndrome, 301026 (3), X-linked recessive
<i>GPHN</i>	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
<i>GPT2</i>	138210	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive
<i>GRIA2</i>	138247	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917 (3), Autosomal dominant
<i>GRIA3</i>	305915	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699 (3), X-linked recessive
<i>GRIA4</i>	138246	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 (3), Autosomal dominant
<i>GRID2</i>	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
<i>GRIK2</i>	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



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<i>GRIN1</i>	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
<i>GRIN2A</i>	138253	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570 (3), Autosomal dominant
<i>GRIN2B</i>	138252	Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant
<i>GRIN2D</i>	602717	Developmental and epileptic encephalopathy 46, 617162 (3), Autosomal dominant
<i>GRIP1</i>	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
<i>GRM1</i>	604473	Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant
<i>GRM7</i>	604101	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 (3), Autosomal recessive
<i>GRN</i>	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
<i>GSS</i>	601002	Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive; Glutathione synthetase deficiency, 266130 (3), Autosomal recessive
<i>GTF2E2</i>	189964	Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive
<i>GTF2H5</i>	608780	Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive
<i>GTPBP2</i>	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
<i>GTPBP3</i>	608536	Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive
<i>GUSB</i>	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
<i>H1-4</i>	142220	Rahman syndrome, 617537 (3), Autosomal dominant
<i>H3-3A</i>	601128	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720 (3), Autosomal dominant
<i>H3-3B</i>	601058	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721 (3), Autosomal dominant
<i>H4C11</i>	602826	?Tessadori-van Haaften neurodevelopmental syndrome 2, 619759 (3), Autosomal dominant
<i>H4C3</i>	602827	Tessadori-van Haaften neurodevelopmental syndrome 1, 619758 (3), Autosomal dominant
<i>H4C5</i>	602830	Tessadori-van Haaften neurodevelopmental syndrome 3, 619950 (3), Autosomal dominant
<i>H4C9</i>	602833	Tessadori-van Haaften neurodevelopmental syndrome 4, 619951 (3), Autosomal dominant
<i>HACE1</i>	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive

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<i>HADH</i>	601609	Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive; 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive
<i>HADHA</i>	600890	HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency, 609015 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
<i>HAX1</i>	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
<i>HCCS</i>	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
<i>HCFC1</i>	300019	Methylmalonic aciduria and homocysteinemia, cblX type, 309541 (3), X-linked recessive
<i>HCN1</i>	602780	Developmental and epileptic encephalopathy 24, 615871 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 10, 618482 (3), Autosomal dominant
<i>HCN2</i>	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
<i>HDAC4</i>	605314	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797 (3), Autosomal dominant
<i>HDAC6</i>	300272	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant
<i>HDAC8</i>	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
<i>HECW2</i>	617245	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 (3), Autosomal dominant
<i>HEPACAM</i>	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926 (3), Autosomal dominant
<i>HERC1</i>	605109	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive
<i>HERC2</i>	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
<i>HESX1</i>	601802	Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal dominant, Autosomal recessive; Septooptic dysplasia, 182230 (3), Autosomal dominant, Autosomal recessive; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal dominant, Autosomal recessive
<i>HEXA</i>	606869	[Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
<i>HEXB</i>	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive

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<i>HGSNAT</i>	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
<i>HIBCH</i>	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
<i>HID1</i>	605752	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983 (3), Autosomal recessive
<i>HIVEP2</i>	143054	Intellectual developmental disorder, autosomal dominant 43, 616977 (3), Autosomal dominant
<i>HK1</i>	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
<i>HLCS</i>	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
<i>HMGB1</i>	163905	No OMIM phenotype 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
<i>HMGCL</i>	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
<i>HNMT</i>	605238	Intellectual developmental disorder, autosomal recessive 51, 616739 (3), Autosomal recessive; {Asthma, susceptibility to}, 600807 (3), Autosomal dominant
<i>HNRNPH1</i>	601035	No OMIM phenotype Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders (Gillentine (2021), Genome Med. 13(1):63), PMID: 33874999 - Autosomal dominant
<i>HNRNPH2</i>	300610	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986 (3), X-linked dominant
<i>HNRNPK</i>	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
<i>HNRNPR</i>	607201	No OMIM phenotype Multisystem developmental disorder (Duijkers (2019), Am J Hum Genet ;104(6):1040-1059), PMID: 31079900 - Autosomal dominant
<i>HNRNPU</i>	602869	Developmental and epileptic encephalopathy 54, 617391 (3), Autosomal dominant
<i>HOXA1</i>	142955	Bosley-Salih-Alorainy syndrome, 601536 (3), Autosomal recessive; Athabaskan brainstem dysgenesis syndrome, 601536 (3), Autosomal recessive
<i>HPD</i>	609695	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
<i>HPDL</i>	618994	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 (3), Autosomal recessive; Spastic paraplegia 83, autosomal recessive, 619027 (3), Autosomal recessive
<i>HPRT1</i>	308000	Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive

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<i>HRAS</i>	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
<i>HS2ST1</i>	604844	Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive
<i>HSD17B10</i>	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
<i>HSD17B4</i>	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
<i>HSPA9</i>	600548	Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant
<i>HSPD1</i>	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
<i>HTRA2</i>	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
<i>HUWE1</i>	300697	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 (3), X-linked
<i>HYCC1 (FAM126A)</i>	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
<i>IARS1</i>	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
<i>IARS2</i>	612801	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
<i>IBA57</i>	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
<i>IDH2</i>	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
<i>IDS</i>	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
<i>IDUA</i>	252800	Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive
<i>IER3IP1</i>	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
<i>IFIH1</i>	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
<i>IFT172</i>	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
<i>IFT27</i>	615870	Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive

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<i>IGF1</i>	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive
<i>IGF1R</i>	147370	Insulin-like growth factor I, resistance to, 270450 (3), Autosomal dominant, Autosomal recessive
<i>IKBK</i>	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
<i>IL1RAPL1</i>	300206	Intellectual developmental disorder, X-linked 21, 300143 (3), X-linked recessive
<i>IMPA1</i>	602064	Intellectual developmental disorder, autosomal recessive 59, 617323 (3), Autosomal recessive
<i>IMPDH2</i>	146691	[IMPDH2 enzyme activity, variation in], 617995 (3)
<i>INPP5E</i>	613037	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
<i>INPP5K</i>	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
<i>INTS1</i>	611345	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 (3), Autosomal recessive
<i>INTS8</i>	611351	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572 (3), Autosomal recessive
<i>IQSEC1</i>	610166	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 (3), Autosomal recessive
<i>IQSEC2</i>	300522	Intellectual developmental disorder, X-linked 1, 309530 (3), X-linked dominant
<i>IREB2</i>	147582	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive
<i>IRF2BPL</i>	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
<i>IRX5</i>	606195	Hamamy syndrome, 611174 (3), Autosomal recessive
<i>ISCA2</i>	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive
<i>ITGA7</i>	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
<i>ITPA</i>	147520	[Inosine triphosphatase deficiency], 613850 (3); Developmental and epileptic encephalopathy 35, 616647 (3), Autosomal recessive
<i>ITPR1</i>	147265	Gillespie syndrome, 206700 (3), Autosomal dominant, Autosomal recessive; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant
<i>IVD</i>	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive

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<i>JAG1</i>	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
<i>JAM2</i>	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive
<i>JAM3</i>	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
<i>JARID2</i>	601594	No OMIM phenotype JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome (Verberne (2021), Genet Med. 23(2):374-383), PMID: 33077894 - Autosomal dominant
<i>KANK1</i>	607704	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
<i>KANSL1</i>	612452	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant
<i>KARS1</i>	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
<i>KAT5</i>	601409	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103 (3), Autosomal dominant
<i>KAT6A</i>	601408	Arboleda-Tham syndrome, 616268 (3), Autosomal dominant
<i>KAT6B</i>	605880	SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant
<i>KAT8</i>	609912	Li-Ghorgani-Weisz-Hubshman syndrome, 618974 (3), Autosomal dominant
<i>KATNAL2</i>	614697	No OMIM phenotype Autisme (Sanders (2012) Nature 4;485(7397):237-41 & O'Roak (2012) Nature 485(7397)). PMID: 22495309, 22495306 - Autosomal dominant
<i>KATNB1</i>	602703	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
<i>KATNIP</i>	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
<i>KCNA1</i>	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
<i>KCNA2</i>	176262	Developmental and epileptic encephalopathy 32, 616366 (3), Autosomal dominant
<i>KCNA4</i>	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
<i>KCNB1</i>	600397	Developmental and epileptic encephalopathy 26, 616056 (3), Autosomal dominant
<i>KCNC1</i>	176258	Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant
<i>KCNC2</i>	176256	Developmental and epileptic encephalopathy 103, 619913 (3), Autosomal dominant

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<i>KCNC3</i>	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant No OMIM phenotype
<i>KCND2</i>	605410	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
<i>KCND3</i>	605411	Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant; Brugada syndrome 9, 616399 (3), Autosomal dominant
<i>KCNH1</i>	603305	Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant; Temple-Baraitser syndrome, 611816 (3), Autosomal dominant
<i>KCNJ10</i>	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
<i>KCNJ11</i>	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 dominant, Autosomal recessive
<i>KCNJ6</i>	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
<i>KCNK4</i>	605720	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381 (3), Autosomal dominant
<i>KCNK9</i>	605874	Birk-Barel syndrome, 612292 (3)
<i>KCNMA1</i>	600150	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Liang-Wang syndrome, 618729 (3), Autosomal dominant
<i>KCNN2</i>	605879	?Dystonia 34, myoclonic, 619724 (3), Autosomal dominant; Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 (3), Autosomal dominant
<i>KCNN3</i>	602983	Zimmermann-Laband syndrome 3, 618658 (3), Autosomal dominant
<i>KCNQ2</i>	602235	Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
<i>KCNQ3</i>	602232	Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant
<i>KCNQ5</i>	607357	Intellectual developmental disorder, autosomal dominant 46, 617601 (3), Autosomal dominant
<i>KCNT1</i>	608167	Developmental and epileptic encephalopathy 14, 614959 (3), Autosomal dominant; Epilepsy nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant
<i>KCNT2</i>	610044	Developmental and epileptic encephalopathy 57, 617771 (3), Autosomal dominant

No OMIM phenotype		
<i>KCTD3</i>	613272	Neurodevelopmental and neuropsychiatric disorders (Teng (2019), Review > CNS Neurosci Ther ;25(7):887-902), PMID: 31197948 - Autosomal dominant
<i>KCTD7</i>	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
<i>KDM1A</i>	609132	Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant
<i>KDM3B</i>	609373	Diets-Jongmans syndrome, 618846 (3), Autosomal dominant
<i>KDM4B</i>	609765	Intellectual developmental disorder, autosomal dominant 65, 619320 (3), Autosomal dominant
<i>KDM5B</i>	605393	Intellectual developmental disorder, autosomal recessive 65, 618109 (3), Autosomal recessive
<i>KDM5C</i>	314690	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
<i>KDM6A</i>	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
<i>KDM6B</i>	611577	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505 (3), Autosomal dominant
<i>KIAA0586</i>	610178	Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive
<i>KIDINS220</i>	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant; Ventriculomegaly and arthrogyrosis, 619501 (3), Autosomal recessive
<i>KIF11</i>	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
<i>KIF14</i>	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive
<i>KIF1A</i>	601255	NESCAV syndrome, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal dominant, Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal dominant, Autosomal recessive
No OMIM phenotype		
<i>KIF21B</i>	608322	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
<i>KIF2A</i>	602591	Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant
<i>KIF4A</i>	300521	?Intellectual developmental disorder, X-linked 100, 300923 (3), X-linked recessive
<i>KIF5A</i>	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
<i>KIF5C</i>	604593	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant



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<i>KIF7</i>	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
<i>KIFBP</i>	609367	Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive
<i>KLF7</i>	604865	No OMIM phenotype Developmental delay/intellectual disability, neuromuscular and psychiatric symptoms (Powis (2018), Clin Genet 93(5)), PMID: 29251763 - Autosomal dominant
<i>KLHL15</i>	300980	Intellectual developmental disorder, X-linked 103, 300982 (3), X-linked recessive
<i>KLHL7</i>	611119	Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive
<i>KMT2A</i>	159555	Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
<i>KMT2B</i>	606834	Intellectual developmental disorder, autosomal dominant 68, 619934 (3), Autosomal dominant; Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
<i>KMT2C</i>	606833	Kleefstra syndrome 2, 617768 (3), Autosomal dominant
<i>KMT2D</i>	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
<i>KMT2E</i>	608444	O'Donnell-Luria-Rodan syndrome, 618512 (3), Autosomal dominant
<i>KMT5B</i>	610881	Intellectual developmental disorder, autosomal dominant 51, 617788 (3), Autosomal dominant
<i>KNL1</i>	609173	Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive
<i>KPTN</i>	615620	Intellectual developmental disorder, autosomal recessive 41, 615637 (3), Autosomal recessive
<i>KRAS</i>	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)
<i>KRIT1</i>	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
<i>L1CAM</i>	308840	MASA syndrome, 303350 (3), X-linked recessive; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3), X-linked recessive; Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive; CRASH syndrome, 303350 (3), X-linked recessive; Hydrocephalus with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive

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<i>L2HGDH</i>	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
<i>LAMA1</i>	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
<i>LAMA2</i>	156225	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive
<i>LAMB1</i>	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
<i>LAMC3</i>	604349	Cortical malformations, occipital, 614115 (3), Autosomal recessive
<i>LAMP2</i>	309060	Danon disease, 300257 (3), X-linked dominant
<i>LARGE1</i>	603590	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive
<i>LARP7</i>	612026	Alazami syndrome, 615071 (3), Autosomal recessive
<i>LARS1</i>	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
<i>LAS1L</i>	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
<i>LGI1</i>	604619	Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant
<i>LGI4</i>	608303	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive
<i>LIAS</i>	607031	Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive
<i>LIG4</i>	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
<i>LINGO1</i>	609791	Intellectual developmental disorder, autosomal recessive 64, 618103 (3), Autosomal recessive
<i>LINS1</i>	610350	Intellectual developmental disorder, autosomal recessive 27, 614340 (3), Autosomal recessive
<i>LIPT1</i>	610284	Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive
<i>LMAN2L</i>	609552	?Intellectual developmental disorder, autosomal dominant 69, 617863 (3); ?Intellectual developmental disorder, autosomal recessive 52, 616887 (3), Autosomal recessive
<i>LMBRD2</i>	619490	Developmental delay with variable neurologic and brain abnormalities, 619694 (3), Autosomal dominant
<i>LMNB1</i>	150340	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant; Microcephaly 26, primary, autosomal dominant, 619179 (3), Autosomal dominant
<i>LMNB2</i>	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
<i>LONP1</i>	605490	CODAS syndrome, 600373 (3), Autosomal recessive
<i>LRP2</i>	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
<i>LRPPRC</i>	607544	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive

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<i>LSS</i>	600909	Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-intellectual disability syndrome 4, 618840 (3), Autosomal recessive
<i>LYRM7</i>	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
<i>LYST</i>	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
<i>LZTFL1</i>	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
<i>LZTR1</i>	600574	Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
<i>MAB21L1</i>	601280	Cerebellar, ocular, craniofacial, and genital syndrome, 618479 (3), Autosomal recessive
<i>MAB21L2</i>	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal dominant, Autosomal recessive
<i>MACF1</i>	608271	Lissencephaly 9 with complex brainstem malformation, 618325 (3), Autosomal dominant
<i>MADD</i>	603584	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 (3), Autosomal recessive; DEEAH syndrome, 619004 (3), Autosomal recessive
<i>MAF</i>	177075	Cataract 21, multiple types, 610202 (3), Autosomal dominant; Ayme-Gripp syndrome, 601088 (3), Autosomal dominant
<i>MAG</i>	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
<i>MAGEL2</i>	605283	Schaaf-Yang syndrome, 615547 (3), Autosomal dominant
<i>MAGT1</i>	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
<i>MAN1B1</i>	604346	Rafiq syndrome, 614202 (3), Autosomal recessive
<i>MAN2B1</i>	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<i>MANBA</i>	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
<i>MAOA</i>	309850	{Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive
<i>MAP1B</i>	157129	?Deafness, autosomal dominant 83, 619808 (3), Autosomal dominant; Periventricular nodular heterotopia 9, 618918 (3), Autosomal dominant
<i>MAP2K1</i>	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
<i>MAP2K2</i>	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
<i>MAPK1</i>	176948	Noonan syndrome 13, 619087 (3), Autosomal dominant
<i>MAPK8IP3</i>	605431	Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant
<i>MAPKAPK5</i>	606723	Neurocardiofaciodigital syndrome, 619869 (3), Autosomal recessive
<i>MAPRE2</i>	605789	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant

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<i>MASP1</i>	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
<i>MAST1</i>	612256	Mega-corporum-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 (3), Autosomal dominant
<i>MAT1A</i>	610550	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal dominant, Autosomal recessive; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal dominant, Autosomal recessive
<i>MBD5</i>	611472	Intellectual developmental disorder, autosomal dominant 1, 156200 (3), Autosomal dominant
<i>MBOAT7</i>	606048	Intellectual developmental disorder, autosomal recessive 57, 617188 (3), Autosomal recessive
<i>MBTPS2</i>	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
<i>MCCC1</i>	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive
<i>MCCC2</i>	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive
<i>MCM3AP</i>	603294	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive
<i>MCOLN1</i>	605248	Mucopolipidosis IV, 252650 (3), Autosomal recessive
<i>MCPH1</i>	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
<i>MDH2</i>	154100	Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive
<i>MECP2</i>	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
<i>MECR</i>	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive
<i>MED12</i>	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
<i>MED12L</i>	611318	Nizon-Isidor syndrome, 618872 (3), Autosomal dominant
<i>MED13</i>	603808	Intellectual developmental disorder, autosomal dominant 61, 618009 (3), Autosomal dominant
<i>MED13L</i>	608771	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant
<i>MED17</i>	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive

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<i>MED23</i>	605042	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249 (3), Autosomal recessive
<i>MED25</i>	610197	Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive
<i>MED27</i>	605044	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 (3), Autosomal recessive
<i>MEF2C</i>	600662	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443 (3), Autosomal dominant
<i>MEGF8</i>	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
<i>MEIS2</i>	601740	Cleft palate, cardiac defects, and mental retardation, 600987 (3), Autosomal dominant
<i>METTL23</i>	615262	Intellectual developmental disorder, autosomal recessive 44, 615942 (3), Autosomal recessive
<i>METTL5</i>	618628	Intellectual developmental disorder, autosomal recessive 72, 618665 (3), Autosomal recessive
<i>MFF</i>	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive
<i>MFSD2A</i>	614397	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 (3), Autosomal recessive
<i>MFSD8</i>	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
<i>MGAT2</i>	602616	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive
<i>MGP</i>	154870	Keutel syndrome, 245150 (3), Autosomal recessive
<i>MICAL1</i>	607129	No OMIM phenotype Mutations in MICAL-1 cause autosomal-dominant lateral temporal epilepsy (Dazzo (2018), Ann Neurol 83(3):483-493), PMID: 29394500 - Autosomal dominant
<i>MICU1</i>	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
<i>MID1</i>	300552	Opitz GBBB syndrome, 300000 (3), X-linked recessive
<i>MINPP1</i>	605391	{Thyroid carcinoma, follicular}, 188470 (3), Autosomal dominant, Somatic mutation; Pontocerebellar hypoplasia, type 16, 619527 (3), Autosomal recessive
<i>MKKS</i>	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
<i>MLC1</i>	605908	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive
<i>MLYCD</i>	606761	Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive
<i>MMAA</i>	607481	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 (3), Autosomal recessive
<i>MMAB</i>	607568	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 (3), Autosomal recessive

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<i>MMACHC</i>	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
<i>MMADHC</i>	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
<i>MMUT</i>	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
<i>MN1</i>	156100	CEBALID syndrome, 618774 (3), Autosomal dominant; Meningioma, 607174 (3), Autosomal dominant
<i>MOCS1</i>	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
<i>MOCS2</i>	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
<i>MOGS</i>	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
<i>MORC2</i>	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
<i>MPDU1</i>	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
<i>MPDZ</i>	603785	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive
<i>MPLKIP</i>	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
<i>MRAS</i>	608435	Noonan syndrome 11, 618499 (3), Autosomal dominant
<i>MRPL3</i>	607118	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
<i>MRPS22</i>	605810	Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
<i>MRPS34</i>	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
<i>MSL3</i>	300609	Basilicata-Akhtar syndrome, 301032 (3), X-linked dominant
<i>MSMO1</i>	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive
<i>MTFMT</i>	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
<i>MTHFR</i>	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
<i>MTHFS</i>	604197	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 (3), Autosomal recessive
<i>MTO1</i>	614667	Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive
<i>MTOR</i>	601231	Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant

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<i>MTR</i>	156570	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive
<i>MTRFR</i>	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
<i>MTRR</i>	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
<i>MVK</i>	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
<i>MYCN</i>	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant
<i>MYO5A</i>	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
<i>MYT1L</i>	613084	Intellectual developmental disorder, autosomal dominant 39, 616521 (3), Autosomal dominant
<i>NAA10</i>	300013	Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
<i>NAA15</i>	608000	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 (3), Autosomal dominant
<i>NAA20</i>	610833	Intellectual developmental disorder, autosomal recessive 73, 619717 (3), Autosomal recessive
<i>NACC1</i>	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant
<i>NAGA</i>	104170	Schindler disease, type I, 609241 (3), Autosomal recessive; Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
<i>NAGLU</i>	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
<i>NAGS</i>	608300	N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive
<i>NALCN</i>	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
<i>NANS</i>	605202	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive No OMIM phenotype
<i>NAPB</i>	611270	A novel NAPB splicing mutation identified by Trio-based exome sequencing is associated with early-onset epileptic encephalopathy (Zhao (2020), Eur J Med Genet. 64(1):104101), PMID: 33189936 - Autosomal dominant

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<i>NARS1</i>	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
<i>NARS2</i>	612803	Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive; ?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive
<i>NAXD</i>	615910	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive
<i>NAXE</i>	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
<i>NBEA</i>	604889	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157 (3), Autosomal dominant
<i>NBN</i>	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
<i>NCAPG2</i>	608532	Khan-Khan-Katsanis syndrome, 618460 (3), Autosomal recessive
<i>NCDN</i>	608458	Neurodevelopmental disorder with infantile epileptic spasms, 619373 (3), Autosomal dominant
<i>NCKAP1</i>	604891	No OMIM phenotype 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
<i>NCOR1</i>	600849	No OMIM phenotype Autism spectrum disorder, scoliosis, and abnormal palatogenesis (Sakaguchi (2018), case report > Am J Med Genet A 176(11):2466-2469) - Autosomal dominant
<i>NDE1</i>	609449	Microhydranencephaly, 605013 (3), Autosomal recessive; Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive
<i>NDP</i>	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
<i>NDST1</i>	600853	Intellectual developmental disorder, autosomal recessive 46, 616116 (3), Autosomal recessive
<i>NDUFA1</i>	300078	Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive
<i>NDUFA10</i>	603835	Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive
<i>NDUFA11</i>	612638	Mitochondrial complex I deficiency, nuclear type 14, 618236 (3), Autosomal recessive
<i>NDUFA12</i>	614530	Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
<i>NDUFA2</i>	602137	Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive
<i>NDUFA6</i>	602138	Mitochondrial complex I deficiency, nuclear type 33, 618253 (3), Autosomal recessive
<i>NDUFA8</i>	603359	Mitochondrial complex I deficiency, nuclear type 37, 619272 (3), Autosomal recessive



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<i>NDUFAF1</i>	606934	Mitochondrial complex I deficiency, nuclear type 11, 618234 (3), Autosomal recessive
<i>NDUFAF2</i>	609653	Mitochondrial complex I deficiency, nuclear type 10, 618233 (3), Autosomal recessive
<i>NDUFAF3</i>	612911	Mitochondrial complex I deficiency, nuclear type 18, 618240 (3), Autosomal recessive
<i>NDUFAF4</i>	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive
<i>NDUFAF5</i>	612360	Mitochondrial complex I deficiency, nuclear type 16, 618238 (3), Autosomal recessive
<i>NDUFAF8</i>	618461	Mitochondrial complex I deficiency, nuclear type 34, 618776 (3), Autosomal recessive
<i>NDUFB3</i>	603839	Mitochondrial complex I deficiency, nuclear type 25, 618246 (3), Autosomal recessive
<i>NDUFB9</i>	601445	?Mitochondrial complex I deficiency, nuclear type 24, 618245 (3), Autosomal recessive
<i>NDUFS1</i>	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
<i>NDUFS2</i>	602985	Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive
<i>NDUFS3</i>	603846	Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive
<i>NDUFS4</i>	602694	Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive
<i>NDUFS6</i>	603848	Mitochondrial complex I deficiency, nuclear type 9, 618232 (3), Autosomal recessive
<i>NDUFS7</i>	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
<i>NDUFS8</i>	602141	Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive
<i>NDUFV1</i>	161015	Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive
<i>NDUFV2</i>	600532	Mitochondrial complex I deficiency, nuclear type 7, 618229 (3), Autosomal recessive
<i>NECAP1</i>	611623	Developmental and epileptic encephalopathy 21, 615833 (3), Autosomal recessive
<i>NECTIN1</i>	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
<i>NEDD4L</i>	606384	Periventricular nodular heterotopia 7, 617201 (3), Autosomal dominant
<i>NEMF</i>	608378	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099 (3), Autosomal recessive
<i>NEU1</i>	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
<i>NEUROD2</i>	601725	Developmental and epileptic encephalopathy 72, 618374 (3), Autosomal dominant
<i>NEXMIF</i>	300524	Intellectual developmental disorder, X-linked 98, 300912 (3), X-linked dominant
<i>NF1</i>	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant
<i>NFASC</i>	609145	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 (3), Autosomal recessive
<i>NFE2L2</i>	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant

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<i>NFIA</i>	600727	Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant
<i>NFIB</i>	600728	Macrocephaly, acquired, with impaired intellectual development, 618286 (3), Autosomal dominant
<i>NFIX</i>	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Malan syndrome, 614753 (3), Autosomal dominant
<i>NFU1</i>	608100	Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive
<i>NGLY1</i>	610661	Congenital disorder of deglycosylation 1, 615273 (3), Autosomal recessive
<i>NHLRC1</i>	608072	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
<i>NHS</i>	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
<i>NIPBL</i>	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
<i>NKAP</i>	300766	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039 (3), X-linked recessive
<i>NKX2-1</i>	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
<i>NLGN3</i>	300336	{Asperger syndrome susceptibility, X-linked 1}, 300494 (3), Multifactorial, Isolated cases, X-linked; {Autism susceptibility, X-linked 1}, 300425 (3), X-linked
<i>NLGN4X</i>	300427	{Asperger syndrome susceptibility, X-linked 2}, 300497 (3), X-linked; Intellectual developmental disorder, X-linked, 300495 (3), Multifactorial, Isolated cases, X-linked; {Autism susceptibility, X-linked 2}, 300495 (3), Multifactorial, Isolated cases, X-linked
<i>NLRP3</i>	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
<i>NONO</i>	300084	Intellectual developmental disorder, X-linked syndromic 34, 300967 (3), X-linked
<i>NOVA2</i>	601991	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 (3), Autosomal dominant
<i>NPC1</i>	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
<i>NPC2</i>	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
<i>NPHP1</i>	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
<i>NPRL2</i>	607072	Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant
<i>NPRL3</i>	600928	Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant

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<i>NR2F1</i>	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
<i>NR4A2</i>	601828	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911 (3), Autosomal dominant
<i>NRAS</i>	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)
<i>NRROS</i>	615322	Seizures, early-onset, with neurodegeneration and brain calcification, 618875 (3), Autosomal recessive
<i>NRXN1</i>	600565	Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3)
<i>NSD1</i>	606681	Sotos syndrome, 117550 (3), Autosomal dominant
<i>NSD2</i>	602952	Rauch-Steindl syndrome, 619695 (3), Autosomal dominant
<i>NSDHL</i>	300275	CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant
<i>NSUN2</i>	610916	Intellectual developmental disorder, autosomal recessive 5, 611091 (3), Autosomal recessive
<i>NT5C2</i>	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
<i>NTNG2</i>	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
<i>NTRK1</i>	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive
<i>NTRK2</i>	600456	Developmental and epileptic encephalopathy 58, 617830 (3), Autosomal dominant; Obesity, hyperphagia, and developmental delay, 613886 (3), Autosomal dominant
<i>NUBPL</i>	613621	Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive
<i>NUDT2</i>	602852	Intellectual developmental disorder with or without peripheral neuropathy, 619844 (3), Autosomal recessive
<i>NUP107</i>	607617	?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
<i>NUP188</i>	615587	Sandestig-Stefanova syndrome, 618804 (3), Autosomal recessive
<i>NUP214</i>	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
<i>NUP62</i>	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
<i>NUS1</i>	610463	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive
<i>OAT</i>	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive

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<i>OCLN</i>	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
<i>OCRL</i>	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
<i>ODC1</i>	165640	Bachmann-Bupp syndrome, 619075 (3), Autosomal dominant
<i>OFD1</i>	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
<i>OGT</i>	300255	Intellectual developmental disorder, X-linked 106, 300997 (3), X-linked recessive
<i>OPHN1</i>	300127	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 (3), X-linked recessive
<i>ORC1</i>	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
<i>OSGEP</i>	610107	Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive
<i>OTC</i>	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked
<i>OTUD5</i>	300713	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056 (3), X-linked recessive
<i>OTUD6B</i>	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive
<i>OTX2</i>	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
<i>OXR1</i>	605609	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 (3), Autosomal recessive
<i>P4HTM</i>	614584	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 (3), Autosomal recessive
<i>PABPC1</i>	604679	No OMIM phenotype 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
<i>PACS1</i>	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
<i>PACS2</i>	610423	Developmental and epileptic encephalopathy 66, 618067 (3), Autosomal dominant
<i>PAFAH1B1</i>	601545	Subcortical laminar heterotopia, 607432 (3), Autosomal dominant; Lissencephaly 1, 607432 (3), Autosomal dominant
<i>PAH</i>	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
<i>PAK1</i>	602590	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158 (3), Autosomal dominant
<i>PAK3</i>	300142	Intellectual developmental disorder, X-linked 30, 300558 (3), X-linked recessive

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<i>PANK2</i>	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
<i>PARN</i>	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant
<i>PARP6</i>	619439	No OMIM phenotype Characterization of PARP6 Function in Knockout Mice and Patients with Developmental Delay (Vermehren-Schmaedick (2021), Cells. 10(6):1289), PMID: 34067418 - Autosomal dominant
<i>PARS2</i>	612036	Developmental and epileptic encephalopathy 75, 618437 (3), Autosomal recessive
<i>PAX1</i>	167411	Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
<i>PAX6</i>	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
<i>PAX8</i>	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
<i>PBX1</i>	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
<i>PC</i>	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
<i>PCCA</i>	232000	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCCB</i>	232050	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCDH12</i>	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
<i>PCDH19</i>	300460	Developmental and epileptic encephalopathy 9, 300088 (3), X-linked
<i>PCDHGC4</i>	606305	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880 (3), Autosomal recessive
<i>PCGF2</i>	600346	Turnpenny-Fry syndrome, 618371 (3), Autosomal dominant
<i>PCLO</i>	604918	?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive
<i>PCNT</i>	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
<i>PCYT2</i>	602679	Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive
<i>PDCD10</i>	609118	Cerebral cavernous malformations-3, 603285 (3), Autosomal dominant
<i>PDE2A</i>	602658	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 (3), Autosomal recessive
<i>PDE4D</i>	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant
<i>PDE6D</i>	602676	Joubert syndrome 22, 615665 (3), Autosomal recessive

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<i>PDGFRB</i>	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
<i>PDHA1</i>	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
<i>PDHB</i>	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive
<i>PDHX</i>	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
<i>PDP1</i>	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
<i>PDSS1</i>	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
<i>PDSS2</i>	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
<i>PDX1</i>	600733	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; Pancreatic agenesis 1, 260370 (3), Autosomal recessive; MODY, type IV, 606392 (3)
<i>PEPD</i>	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
<i>PET100</i>	614770	Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive
<i>PEX1</i>	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
<i>PEX10</i>	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
<i>PEX11B</i>	603867	Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
<i>PEX12</i>	601758	Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive
<i>PEX13</i>	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
<i>PEX14</i>	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
<i>PEX16</i>	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
<i>PEX19</i>	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
<i>PEX2</i>	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
<i>PEX26</i>	608666	Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive

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<i>PEX3</i>	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
<i>PEX5</i>	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
<i>PEX6</i>	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal dominant, Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive
<i>PEX7</i>	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
<i>PGAP1</i>	611655	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 (3), Autosomal recessive
<i>PGAP2</i>	615187	Hyperphosphatasia with mental retardation syndrome 3, 614207 (3), Autosomal recessive
<i>PGAP3</i>	611801	Hyperphosphatasia with mental retardation syndrome 4, 615716 (3), Autosomal recessive
<i>PGK1</i>	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
<i>PGM2L1</i>	611610	No OMIM phenotype Impaired glucose-1,6-biphosphate production due to bi-allelic PGM2L1 mutations is associated with a neurodevelopmental disorder (Morava (2021), Am J Hum Genet. 108(6):1151-1160), PMID: 33979636 - Autosomal recessive
<i>PGM3</i>	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
<i>PHACTR1</i>	608723	Developmental and epileptic encephalopathy 70, 618298 (3), Autosomal dominant
<i>PHF21A</i>	608325	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 (3), Autosomal dominant
<i>PHF6</i>	300414	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
<i>PHF8</i>	300560	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263 (3), X-linked recessive
<i>PHGDH</i>	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
<i>PHIP</i>	612870	Chung-Jansen syndrome, 617991 (3), Autosomal dominant
<i>PI4KA</i>	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
<i>PIBF1</i>	607532	Joubert syndrome 33, 617767 (3), Autosomal recessive

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<i>PIDD1</i>	605247	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827 (3), Autosomal recessive
<i>PIGA</i>	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)
<i>PIGB</i>	604122	Developmental and epileptic encephalopathy 80, 618580 (3), Autosomal recessive
<i>PIGC</i>	601730	Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive
<i>PIGF</i>	600153	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356 (3), Autosomal recessive
<i>PIGG</i>	616918	[Blood group, EMM system], 619812 (3), Autosomal recessive; Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917 (3), Autosomal recessive
<i>PIGH</i>	600154	Glycosylphosphatidylinositol biosynthesis defect 17, 618010 (3), Autosomal recessive
<i>PIGK</i>	605087	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 (3), Autosomal recessive
<i>PIGL</i>	605947	CHIME syndrome, 280000 (3), Autosomal recessive
<i>PIGN</i>	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive
<i>PIGO</i>	614730	Hyperphosphatasia with mental retardation syndrome 2, 614749 (3), Autosomal recessive
<i>PIGP</i>	605938	Developmental and epileptic encephalopathy 55, 617599 (3), Autosomal recessive
<i>PIGQ</i>	605754	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548 (3), Autosomal recessive
<i>PIGS</i>	610271	Developmental and epileptic encephalopathy 95, 618143 (3), Autosomal recessive
<i>PIGT</i>	610272	?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive
<i>PIGU</i>	608528	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 (3), Autosomal recessive
<i>PIGV</i>	610274	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive
<i>PIGW</i>	610275	Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive
<i>PIGY</i>	610662	Hyperphosphatasia with mental retardation syndrome 6, 616809 (3), Autosomal recessive
		No OMIM phenotype
<i>PIK3C2B</i>	602838	Defective lipid signalling caused by mutations in PIK3C2B underlies focal epilepsy (Gozzelino (2022), Brain. 145(7):2313-2331), PMID: 35786744 - Autosomal dominant



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<i>PIK3CA</i>	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)
<i>PIK3R2</i>	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant
<i>PISD</i>	612770	Liberfarb syndrome, 618889 (3), Autosomal recessive
<i>PITRM1</i>	618211	Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive
<i>PLA2G6</i>	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
<i>PLAA</i>	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive
<i>PLCB1</i>	607120	Developmental and epileptic encephalopathy 12, 613722 (3), Autosomal recessive
<i>PLCH1</i>	612835	Holoprosencephaly 14, 619895 (3), Autosomal recessive
<i>PLK4</i>	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
<i>PLP1</i>	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
<i>PLPBP</i>	604436	Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive
<i>PLXNA1</i>	601055	Dworschak-Punetha neurodevelopmental syndrome, 619955 (3), Autosomal recessive
<i>PLXND1</i>	604282	No OMIM phenotype Moebius syndrome (Tomas-Roca (2015) Nat Commun 6), Autosomal dominant Truncus arteriosus (Ta-Shma (2013) Am J Med Genet A 161,3115) {Diabetic nephropathy,association with} (McKnight (2009) Hugo J 3,77)
<i>PMM2</i>	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
<i>PMPCA</i>	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
<i>PMPCB</i>	603131	Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive
<i>PNKP</i>	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
<i>PNP</i>	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive

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<i>PNPLA6</i>	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
<i>PNPO</i>	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
<i>PNPT1</i>	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
<i>POC1A</i>	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
<i>POC1B</i>	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
<i>POGZ</i>	614787	White-Sutton syndrome, 616364 (3), Autosomal dominant
<i>POLA1</i>	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
<i>POLG</i>	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
<i>POLR1C</i>	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
<i>POLR2A</i>	180660	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 (3), Autosomal dominant
<i>POLR3A</i>	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
<i>POLR3B</i>	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
<i>POMGNT1</i>	606822	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (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
<i>POMGNT2</i>	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

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<i>POMK</i>	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
<i>POMT1</i>	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 mental retardation), type B, 1, 613155 (3), Autosomal recessive
<i>POMT2</i>	607439	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive
<i>PORCN</i>	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
<i>POU1F1</i>	173110	Pituitary hormone deficiency, combined or isolated, 1, 613038 (3), Autosomal dominant, Autosomal recessive
<i>POU3F3</i>	602480	Snijders Blok-Fisher syndrome, 618604 (3), Autosomal dominant
<i>PPFIBP1</i>	603141	No OMIM phenotype Bi-allelic loss-of-function variants in PPFIBP1 cause a neurodevelopmental disorder with microcephaly, epilepsy, and periventricular calcifications (Rosenhahn (2022), Am J Hum Genet. 109(8):1421-1435), PMID: 35830857 - Autosomal recessive
<i>PPIL1</i>	601301	Pontocerebellar hypoplasia, type 14, 619301 (3), Autosomal recessive
<i>PPM1D</i>	605100	Breast cancer, somatic, 114480 (3); Jansen de Vries syndrome, 617450 (3), Autosomal dominant
<i>PPP1CB</i>	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
<i>PPP1R12A</i>	602021	Genitourinary and/or/brain malformation syndrome, 618820 (3), Autosomal dominant
<i>PPP1R15B</i>	613257	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
<i>PPP1R21</i>	618159	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383 (3), Autosomal recessive
<i>PPP2CA</i>	176915	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354 (3), Autosomal dominant
<i>PPP2R1A</i>	605983	Intellectual developmental disorder, autosomal dominant 36, 616362 (3), Autosomal dominant
<i>PPP2R5D</i>	601646	Intellectual developmental disorder, autosomal dominant 35, 616355 (3), Autosomal dominant
<i>PPP3CA</i>	114105	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 (3), Autosomal dominant; Developmental and epileptic encephalopathy 91, 617711 (3), Autosomal dominant
<i>PPT1</i>	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive

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<i>PQBP1</i>	300463	Renpenning syndrome, 309500 (3), X-linked recessive
<i>PRDM13</i>	616741	Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive
<i>PRDM15</i>	617692	No OMIM phenotype 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
<i>PRF1</i>	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
<i>PRICKLE1</i>	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
<i>PRICKLE2</i>	608501	No OMIM phenotype PRICKLE2 revisited-further evidence implicating PRICKLE2 in neurodevelopmental disorders (Bayat (2021), Eur J Hum Genet. 29(8):1235-1244), PMID: 34092786 - Autosomal dominant
<i>PRIMA1</i>	613851	No OMIM phenotype PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy (Hildebrand (2015) Ann Clin Transl Neurol 2(8):821-30), PMID: 26339676 - Autosomal dominant
<i>PRKACB</i>	176892	Cardioacrofacial dysplasia 2, 619143 (3), Autosomal dominant, Somatic mosaicism
<i>PRKAR1A</i>	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
<i>PRKAR1B</i>	176911	Marbach-Schaaf neurodevelopmental syndrome, 619680 (3), Autosomal dominant
<i>PRMT7</i>	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
<i>PRODH</i>	606810	{Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant; Hyperprolinemia, type I, 239500 (3), Autosomal recessive
<i>PRPS1</i>	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
<i>PRR12</i>	616633	Neuroocular syndrome, 619539 (3), Autosomal dominant
<i>PRRT2</i>	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
<i>PRSS12</i>	606709	Intellectual developmental disorder, autosomal recessive 1, 249500 (3), Autosomal recessive

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<i>PRUNE1</i>	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive
<i>PSAP</i>	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); {Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 (3), Autosomal dominant
<i>PSAT1</i>	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
<i>PSMD12</i>	604450	Stankiewicz-Isidor syndrome, 617516 (3), Autosomal dominant
<i>PSPH</i>	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
<i>PTCH1</i>	601309	Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant; Basal cell nevus syndrome, 109400 (3), Autosomal dominant
<i>PTCHD1</i>	300828	{Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive
<i>PTDSS1</i>	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
<i>PTEN</i>	601728	{Glioma susceptibility 2}, 613028 (3); {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
<i>PTPN11</i>	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)
<i>PTPN23</i>	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive
<i>PTPN4</i>	176878	No OMIM phenotype PTPN4 germline variants result in aberrant neurodevelopment and growth (Chmielewska (2021), HGG Adv. 2021 2(3):100033), PMID: 34527963 - Autosomal dominant
<i>PTRH2</i>	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
<i>PTRHD1</i>	617342	No OMIM phenotype ?Neurodevelopmental disorder (Reuter (2017) JAMA Psychiatry), Autosomal recessive Intellectual disability and parkinsonism (Khodadadi (2017), Mov Disord and Elahi (2018), Mov Disord), Autosomal Recessive
<i>PTS</i>	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
<i>PUF60</i>	604819	Verheij syndrome, 615583 (3), Autosomal dominant
<i>PUM1</i>	607204	Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant

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<i>PURA</i>	600473	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158 (3), Autosomal dominant
<i>PUS1</i>	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
<i>PUS3</i>	616283	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 (3), Autosomal recessive
<i>PUS7</i>	616261	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 (3), Autosomal recessive
<i>PYCR1</i>	179035	Cutis laxa, autosomal recessive, type IIIB, 614438 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive
<i>PYCR2</i>	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
<i>QARS1</i>	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive
<i>QDPR</i>	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
<i>QRICH1</i>	617387	Ververi-Brady syndrome, 617982 (3), Autosomal dominant
<i>RAB11B</i>	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
<i>RAB18</i>	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
<i>RAB23</i>	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
<i>RAB27A</i>	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
<i>RAB39B</i>	300774	Intellectual developmental disorder, X-linked 72, 300271 (3), X-linked recessive; Waisman syndrome, 311510 (3), X-linked recessive
<i>RAB3GAP1</i>	602536	Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive
<i>RAB3GAP2</i>	609275	Martsolf syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
<i>RABGAP1</i>	615882	No OMIM phenotype 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
<i>RAC1</i>	602048	Intellectual developmental disorder, autosomal dominant 48, 617751 (3), Autosomal dominant
<i>RAC3</i>	602050	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 (3), Autosomal dominant
<i>RAD21</i>	606462	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant; ?Mungan syndrome, 611376 (3), Autosomal recessive
<i>RAF1</i>	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3)

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<i>RAI1</i>	607642	Smith-Magenis syndrome, 182290 (3), Autosomal dominant, Isolated cases
<i>RALA</i>	179550	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311 (3), Autosomal dominant
<i>RALGAPA1</i>	608884	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 (3), Autosomal recessive
<i>RARB</i>	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal dominant, Autosomal recessive
<i>RARS2</i>	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
<i>RBBP8</i>	604124	Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3)
<i>RBFOX1</i>	605104	No OMIM phenotype Epilepsy, rolandic (Lal (2013) PLoS One 8, e73323), Autosomal dominant Mental retardation (Bhalla (2004) J Hum Genet 49, 308) ?Autism spectrum disorder (Griswold (2015) Mol Autism 6, 43)
<i>RBM10</i>	300080	TARP syndrome, 311900 (3), X-linked recessive
<i>RBM28</i>	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
<i>RBPJ</i>	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
<i>RCBTB1</i>	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
<i>RELN</i>	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
<i>RERE</i>	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
<i>RFT1</i>	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
<i>RFX3</i>	601337	No OMIM phenotype 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
<i>RFX4</i>	603958	No OMIM phenotype 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
<i>RFX7</i>	612660	No OMIM phenotype 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

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		No OMIM phenotype
<i>RHEB</i>	601293	?Intellectual disability (Reijnders 2017 (Nat Commun)), Autosomal dominant
<i>RHOBTB2</i>	607352	Developmental and epileptic encephalopathy 64, 618004 (3), Autosomal dominant
<i>RIMS2</i>	606630	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive
<i>RIT1</i>	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
<i>RLIM</i>	300379	Tonne-Kalscheuer syndrome, 300978 (3), X-linked
<i>RMND1</i>	614917	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive
<i>RNASEH2A</i>	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
<i>RNASEH2B</i>	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
<i>RNASEH2C</i>	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
<i>RNASET2</i>	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
<i>RNF113A</i>	300951	Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked
<i>RNF125</i>	610432	Tenorio syndrome, 616260 (3), Autosomal dominant
<i>RNF13</i>	609247	Developmental and epileptic encephalopathy 73, 618379 (3), Autosomal dominant
<i>RNF220</i>	616136	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688 (3), Autosomal recessive
<i>RNPC3</i>	618016	Pituitary hormone deficiency, combined or isolated, 7, 618160 (3), Autosomal recessive
<i>RNU4ATAC</i>	601428	Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive
<i>RNU7-1</i>	617876	Aicardi-Goutieres syndrome 9, 619487 (3), Autosomal recessive
<i>ROGDI</i>	614574	Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive
<i>RORA</i>	600825	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 (3), Autosomal dominant
<i>RORB</i>	601972	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357 (3), Autosomal dominant
<i>RPGRIP1L</i>	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
<i>RPIA</i>	180430	Ribose 5-phosphate isomerase deficiency, 608611 (3), Autosomal recessive
<i>RPL10</i>	312173	{Autism, susceptibility to, X-linked 5}, 300847 (3); Intellectual developmental disorder, X-linked syndromic 35, 300998 (3), X-linked recessive
<i>RPS19</i>	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
<i>RPS6KA3</i>	300075	Intellectual developmental disorder, X-linked 19, 300844 (3), X-linked dominant; Coffin-Lowry syndrome, 303600 (3), X-linked dominant



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<i>RRAS2</i>	600098	Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant
<i>RRM2B</i>	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
<i>RSPRY1</i>	616585	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive
<i>RSRC1</i>	613352	Intellectual developmental disorder, autosomal recessive 70, 618402 (3), Autosomal recessive
<i>RTEL1</i>	608833	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal dominant, Autosomal recessive
<i>RTN4IP1</i>	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive
<i>RTTN</i>	610436	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
<i>RUBCN</i>	613516	Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
<i>RUSC2</i>	611053	Intellectual developmental disorder, autosomal recessive 61, 617773 (3), Autosomal recessive
<i>RXYLT1</i>	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
<i>SALL1</i>	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant
<i>SAMD9</i>	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
<i>SAMHD1</i>	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
<i>SARS1</i>	607529	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive
<i>SATB1</i>	602075	Kohlschutter-Tonz syndrome-like, 619229 (3), Autosomal dominant; Developmental delay with dysmorphic facies and dental anomalies, 619228 (3), Autosomal dominant
<i>SATB2</i>	608148	Glass syndrome, 612313 (3), Autosomal dominant
<i>SBDS</i>	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
<i>SBF1</i>	603560	Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive
<i>SC5D</i>	602286	Lathosterolosis, 607330 (3), Autosomal recessive

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<i>SCAF4</i>	616023	No OMIM phenotype 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
<i>SCAMP5</i>	613766	No OMIM phenotype 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
<i>SCAPER</i>	611611	Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive
<i>SCARB2</i>	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
<i>SCN1A</i>	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
<i>SCN1B</i>	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)
<i>SCN2A</i>	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
<i>SCN3A</i>	182391	Epilepsy, familial focal, with variable foci 4, 617935 (3), Autosomal dominant; Developmental and epileptic encephalopathy 62, 617938 (3), Autosomal dominant
<i>SCN8A</i>	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
<i>SCO1</i>	603644	Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive
<i>SCO2</i>	604272	Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive
<i>SCYL1</i>	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
<i>SDCCAG8</i>	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
<i>SDHA</i>	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

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<i>SDHAF1</i>	612848	Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive
<i>SEMA3E</i>	608166	?CHARGE syndrome, 214800 (3), Autosomal dominant
<i>SEMA6B</i>	608873	Epilepsy, progressive myoclonic, 11, 618876 (3), Autosomal dominant
<i>SEPSECS</i>	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
<i>SERAC1</i>	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
<i>SERPINI1</i>	602445	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3), Autosomal dominant
<i>SET</i>	600960	Intellectual developmental disorder, autosomal dominant 58, 618106 (3), Autosomal dominant
<i>SETBP1</i>	611060	Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 29, 616078 (3), Autosomal dominant
<i>SETD1A</i>	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
<i>SETD1B</i>	611055	Intellectual developmental disorder with seizures and language delay, 619000 (3), Autosomal dominant
<i>SETD2</i>	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant
<i>SETD5</i>	615743	Intellectual developmental disorder, autosomal dominant 23, 615761 (3), Autosomal dominant
<i>SFXN4</i>	615564	Combined oxidative phosphorylation deficiency 18, 615578 (3), Autosomal recessive
<i>SGPL1</i>	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
<i>SGSH</i>	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
<i>SHANK1</i>	604999	No OMIM phenotype Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders (May (2021), Genet Med. 23(10):1912-1921), PMID: 34113010 - Autosomal dominant
<i>SHANK2</i>	603290	{Autism susceptibility 17}, 613436 (3)
<i>SHANK3</i>	606230	Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3), Autosomal dominant
<i>SHH</i>	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
<i>SHMT2</i>	138450	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 (3), Autosomal recessive
<i>SHOC2</i>	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
<i>SHROOM4</i>	300579	Intellectual developmental disorder, X-linked syndromic, Stocco dos Santos type, 300434 (3), X-linked
<i>SIAH1</i>	602212	Buratti-Harel syndrome, 619314 (3), Autosomal dominant

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<i>SIK1</i>	605705	Developmental and epileptic encephalopathy 30, 616341 (3), Autosomal dominant
<i>SIL1</i>	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
<i>SIN3A</i>	607776	Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant
		No OMIM phenotype
<i>SIN3B</i>	607777	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
<i>SIX3</i>	603714	Schizencephaly, 269160 (3); Holoprosencephaly 2, 157170 (3), Autosomal dominant
<i>SKI</i>	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
<i>SKIC3</i> ( <i>TTC37</i> )	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
<i>SLC12A2</i>	600840	Kilquist syndrome, 619080 (3), Autosomal recessive; Delpire-McNeill syndrome, 619083 (3), Autosomal dominant; Deafness, autosomal dominant 78, 619081 (3), Autosomal dominant
<i>SLC12A5</i>	606726	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Developmental and epileptic encephalopathy 34, 616645 (3), Autosomal recessive
<i>SLC12A6</i>	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive
<i>SLC13A5</i>	608305	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive
<i>SLC16A1</i>	600682	Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal dominant, Autosomal recessive
<i>SLC16A2</i>	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
<i>SLC17A5</i>	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
<i>SLC19A3</i>	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
<i>SLC1A1</i>	133550	Dicarboxylic aminoaciduria, 222730 (3), Autosomal recessive; {?Schizophrenia susceptibility 18}, 615232 (3)
<i>SLC1A2</i>	600300	Developmental and epileptic encephalopathy 41, 617105 (3), Autosomal dominant
<i>SLC1A4</i>	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
<i>SLC25A1</i>	190315	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive; Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive
<i>SLC25A12</i>	603667	Developmental and epileptic encephalopathy 39, 612949 (3), Autosomal recessive
<i>SLC25A15</i>	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive

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<i>SLC25A19</i>	606521	Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive
<i>SLC25A20</i>	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
<i>SLC25A22</i>	609302	Developmental and epileptic encephalopathy 3, 609304 (3), Autosomal recessive
<i>SLC25A42</i>	610823	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive
<i>SLC2A1</i>	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal dominant, Autosomal recessive; 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
<i>SLC33A1</i>	603690	Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive
<i>SLC35A1</i>	605634	Congenital disorder of glycosylation, type II <sub>f</sub> , 603585 (3), Autosomal recessive
<i>SLC35A2</i>	314375	Congenital disorder of glycosylation, type II <sub>m</sub> , 300896 (3), Somatic mosaicism, X-linked dominant
<i>SLC35A3</i>	605632	Arthrogyriposis, impaired intellectual development, and seizures, 615553 (3), Autosomal recessive
<i>SLC35C1</i>	605881	Congenital disorder of glycosylation, type II <sub>c</sub> , 266265 (3), Autosomal recessive
<i>SLC39A14</i>	608736	?Hyperostosis cranialis interna, 144755 (3), Autosomal dominant; Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive
<i>SLC39A8</i>	608732	Congenital disorder of glycosylation, type II <sub>n</sub> , 616721 (3), Autosomal recessive
<i>SLC46A1</i>	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
<i>SLC4A4</i>	603345	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive
<i>SLC5A6</i>	604024	Sodium-dependent multivitamin transporter deficiency, 618973 (3), Autosomal recessive; Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903 (3), Autosomal recessive
<i>SLC6A1</i>	137165	Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant
<i>SLC6A17</i>	610299	Intellectual developmental disorder, autosomal recessive 48, 616269 (3), Autosomal recessive
<i>SLC6A19</i>	608893	Iminoglycinuria, digenic, 242600 (3), Digenic recessive, Autosomal recessive; Hartnup disorder, 234500 (3), Autosomal recessive; Hyperglycinuria, 138500 (3), Autosomal dominant
<i>SLC6A3</i>	126455	Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive; {Nicotine dependence, protection against}, 188890 (3)
<i>SLC6A8</i>	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
<i>SLC6A9</i>	601019	Glycine encephalopathy with normal serum glycine, 617301 (3), Autosomal recessive
<i>SLC7A7</i>	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
<i>SLC9A6</i>	300231	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 (3), X-linked

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<i>SLC9A7</i>	300368	Intellectual developmental disorder, X-linked 108, 301024 (3), X-linked recessive
<i>SMAD4</i>	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
<i>SMAD6</i>	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Radioulnar synostosis, nonsyndromic}, 179300 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
<i>SMARCA2</i>	600014	Nicolaidis-Baraitser syndrome, 601358 (3), Autosomal dominant; Blepharophimosis-impaired intellectual development syndrome, 619293 (3), Autosomal dominant
<i>SMARCA4</i>	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant
<i>SMARCA5</i>	603375	No OMIM phenotype Pathogenic variants in SMARCA5, a chromatin remodeler, cause a range of syndromic neurodevelopmental features (Li (2021), Sci Adv 7(20)), PMID: 33980485 - Autosomal dominant
<i>SMARCB1</i>	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
<i>SMARCC2</i>	601734	Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant
<i>SMARCD1</i>	601735	Coffin-Siris syndrome 11, 618779 (3), Autosomal dominant
<i>SMARCE1</i>	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
<i>SMC1A</i>	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
<i>SMC3</i>	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
<i>SMG8</i>	613175	Alzahrani-Kuwahara syndrome, 619268 (3), Autosomal recessive
<i>SMG9</i>	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
<i>SMOC1</i>	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
<i>SMPD1</i>	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
<i>SMPD4</i>	610457	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive

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<i>SMS</i>	300105	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 (3), X-linked recessive
<i>SNAP25</i>	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
<i>SNAP29</i>	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
<i>SNIP1</i>	608241	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501 (3), Autosomal recessive
<i>SNORD118</i>	616663	Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive
<i>SNRPN</i>	182279	Prader-Willi syndrome, 176270 (3), Autosomal dominant
<i>SNX14</i>	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
<i>SNX27</i>	611541	No OMIM phenotype Seizures, developmental delay, behavioral disturbance, and subcortical brain abnormalities (Parente (2019), Clin Genet 97(3):437-446), PMID: 31721175 - Autosomal recessive
<i>SOBP</i>	613667	Mental retardation, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive
<i>SON</i>	182465	ZTTK syndrome, 617140 (3), Autosomal dominant
<i>SOS1</i>	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
<i>SOS2</i>	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
<i>SOX10</i>	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
<i>SOX11</i>	600898	Coffin-Siris syndrome 9, 615866 (3), Autosomal dominant
<i>SOX2</i>	184429	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant; Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant
<i>SOX3</i>	313430	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
<i>SOX4</i>	184430	Coffin-Siris syndrome 10, 618506 (3), Autosomal dominant
<i>SOX5</i>	604975	Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant
<i>SOX6</i>	607257	Tolchin-Le Caignec syndrome, 618971 (3), Autosomal dominant
<i>SPART</i>	607111	Troyer syndrome, 275900 (3), Autosomal recessive
<i>SPAST</i>	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
<i>SPATA5</i>	613940	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 (3), Autosomal recessive

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<i>SPATA5L1</i>	619578	Deafness, autosomal recessive 119, 619615 (3), Autosomal recessive; Neurodevelopmental disorder with hearing loss and spasticity, 619616 (3), Autosomal recessive
<i>SPECC1L</i>	614140	Teebi hypertelorism syndrome 1, 145420 (3), Autosomal dominant; ?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant
<i>SPEN</i>	613484	Radio-Tartaglia syndrome, 619312 (3), Autosomal dominant
<i>SPG11</i>	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
<i>SPOP</i>	602650	Nabais Sa-de Vries syndrome, type 1, 618828 (3), Autosomal dominant; Nabais Sa-de Vries syndrome, type 2, 618829 (3), Autosomal dominant
<i>SPR</i>	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
<i>SPRED1</i>	609291	Legius syndrome, 611431 (3), Autosomal dominant
<i>SPTAN1</i>	182810	Developmental and epileptic encephalopathy 5, 613477 (3), Autosomal dominant
<i>SPTBN1</i>	182790	Developmental delay, impaired speech, and behavioral abnormalities, 619475 (3), Autosomal dominant
<i>SPTBN2</i>	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
<i>SPTBN4</i>	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive
<i>SRCAP</i>	611421	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 (3), Autosomal dominant; Floating-Harbor syndrome, 136140 (3), Autosomal dominant
<i>SRD5A3</i>	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
<i>SRPX2</i>	300642	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643 (3) No OMIM phenotype
<i>SRRM2</i>	606032	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder (Cuinat (2022), Genet Med. 24(8):1774-1780), PMID: 35567594 - Autosomal dominant
<i>SSR4</i>	300090	Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive
<i>ST3GAL3</i>	606494	Developmental and epileptic encephalopathy 15, 615006 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 12, 611090 (3), Autosomal recessive
<i>ST3GAL5</i>	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
<i>STAG1</i>	604358	Intellectual developmental disorder, autosomal dominant 47, 617635 (3), Autosomal dominant
<i>STAG2</i>	300826	Holoprosencephaly 13, X-linked, 301043 (3), X-linked recessive, X-linked dominant; Mullegama-Klein-Martinez syndrome, 301022 (3), X-linked



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<i>STAMBP</i>	606247	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
<i>STEEP1</i>	301012	?Intellectual developmental disorder, X-linked 107, 301013 (3), X-linked
<i>STIL</i>	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
<i>STRA6</i>	610745	Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive
<i>STRADA</i>	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive
<i>STT3A</i>	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
<i>STT3B</i>	608605	Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
<i>STX1B</i>	601485	Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant
<i>STXBP1</i>	602926	Developmental and epileptic encephalopathy 4, 612164 (3), Autosomal dominant
<i>SUCLA2</i>	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
<i>SUCLG1</i>	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
<i>SUFU</i>	607035	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Medulloblastoma, desmoplastic, 155255 (3), Autosomal dominant, Somatic mutation, Autosomal recessive; Basal cell nevus syndrome, 109400 (3), Autosomal dominant
<i>SUMF1</i>	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
<i>SUOX</i>	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
<i>SUPT16H</i>	605012	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480 (3), Autosomal dominant
<i>SURF1</i>	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive
<i>SUZ12</i>	606245	Imagawa-Matsumoto syndrome, 618786 (3), Autosomal dominant
<i>SVBP</i>	617853	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive
<i>SYN1</i>	313440	Intellectual developmental disorder, X-linked 50, 300115 (3), X-linked; Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3), X-linked recessive, X-linked dominant
<i>SYNCRIP</i>	616686	No OMIM phenotype 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

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<i>SYNE1</i>	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
<i>SYNGAP1</i>	603384	Intellectual developmental disorder, autosomal dominant 5, 612621 (3), Autosomal dominant
<i>SYNJ1</i>	604297	Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive; Developmental and epileptic encephalopathy 53, 617389 (3), Autosomal recessive
<i>SYP</i>	313475	Intellectual developmental disorder, X-linked 96, 300802 (3), X-linked recessive
<i>SYT1</i>	185605	Baker-Gordon syndrome, 618218 (3), Autosomal dominant
<i>SYT14</i>	610949	?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive
<i>SZT2</i>	615463	Developmental and epileptic encephalopathy 18, 615476 (3), Autosomal recessive
<i>TAF1</i>	313650	Intellectual developmental disorder, X-linked syndromic 33, 300966 (3), X-linked recessive; Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive
<i>TAF13</i>	600774	Intellectual developmental disorder, autosomal recessive 60, 617432 (3), Autosomal recessive
<i>TAF1C</i>	604905	No OMIM phenotype Homozygous TAF1C variants are associated with a novel childhood-onset neurological phenotype (Knuutinen (2020) Clin Genet 98(5):493-498), PMID: 32779182 - Autosomal dominant
<i>TAF2</i>	604912	Intellectual developmental disorder, autosomal recessive 40, 615599 (3), Autosomal recessive
<i>TAF6</i>	602955	Alazami-Yuan syndrome, 617126 (3), Autosomal recessive
<i>TFAZZIN</i>	300394	Barth syndrome, 302060 (3), X-linked recessive
<i>TANC2</i>	615047	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906 (3), Autosomal dominant
<i>TANGO2</i>	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
<i>TAOK1</i>	610266	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575 (3), Autosomal dominant
<i>TASP1</i>	608270	Suleiman-El-Hattab syndrome, 618950 (3), Autosomal recessive
<i>TAT</i>	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
<i>TBC1D20</i>	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
<i>TBC1D23</i>	617687	Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive
<i>TBC1D24</i>	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

<i>TBC1D2B</i>	619152	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323 (3), Autosomal recessive
<i>TBC1D7</i>	612655	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 (3), Autosomal recessive
<i>TBCD</i>	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
<i>TBCE</i>	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
<i>TBCK</i>	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive
<i>TBL1XR1</i>	608628	Intellectual developmental disorder, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant
<i>TBP</i>	600075	Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant
<i>TBR1</i>	604616	Intellectual developmental disorder with autism and speech delay, 606053 (3), Autosomal dominant
<i>TBX1</i>	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
<i>TCF20</i>	603107	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 (3), Autosomal dominant
<i>TCF4</i>	602272	Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant
<i>TCF7L2</i>	602228	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
<i>TCN2</i>	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
<i>TCTN1</i>	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
<i>TCTN2</i>	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
<i>TCTN3</i>	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
<i>TDP2</i>	605764	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive

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<i>TECPR2</i>	615000	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive
<i>TECR</i>	610057	Intellectual developmental disorder, autosomal recessive 14, 614020 (3), Autosomal recessive
<i>TELO2</i>	611140	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive
<i>TENM3</i>	610083	Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive
<i>TET3</i>	613555	Beck-Fahrner syndrome, 618798 (3), Autosomal dominant, Autosomal recessive
<i>TFAP2A</i>	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
<i>TFE3</i>	314310	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066 (3), X-linked; Renal cell carcinoma, papillary, 1, 300854 (3)
<i>TGDS</i>	616146	Catel-Manzke syndrome, 616145 (3), Autosomal recessive
<i>TGFBR1</i>	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
<i>TGFBR2</i>	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
<i>TGIF1</i>	602630	Holoprosencephaly 4, 142946 (3), Autosomal dominant
<i>TH</i>	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
<i>THOC2</i>	300395	Intellectual developmental disorder, X-linked 12, 300957 (3), X-linked recessive
<i>THOC6</i>	615403	Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive
<i>THRA</i>	190120	Hypothyroidism, congenital, nongoitrous, 6, 614450 (3), Autosomal dominant
<i>THRB</i>	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
<i>TIAM1</i>	600687	Neurodevelopmental disorder with language delay and seizures, 619908 (3), Autosomal recessive
<i>TIMM50</i>	607381	3-methylglutaconic aciduria, type IX, 617698 (3), Autosomal recessive
<i>TIMM8A</i>	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
<i>TINF2</i>	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
<i>TKT</i>	606781	Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive
<i>TLK2</i>	608439	Intellectual developmental disorder, autosomal dominant 57, 618050 (3), Autosomal dominant
<i>TMCO1</i>	614123	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980 (3), Autosomal recessive
<i>TMEM106B</i>	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant

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<i>TMEM107</i>	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
<i>TMEM138</i>	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
<i>TMEM165</i>	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
<i>TMEM216</i>	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
<i>TMEM222</i>	619469	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470 (3), Autosomal recessive
<i>TMEM231</i>	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
<i>TMEM237</i>	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
<i>TMEM240</i>	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
<i>TMEM63A</i>	618685	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 (3), Autosomal dominant
<i>TMEM67</i>	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
<i>TMEM70</i>	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
<i>TMEM94</i>	618163	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 (3), Autosomal recessive
<i>TMLHE</i>	300777	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
<i>TMTC3</i>	617218	Lissencephaly 8, 617255 (3), Autosomal recessive
<i>TMX2</i>	616715	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive
<i>TNIK</i>	610005	Intellectual developmental disorder, autosomal recessive 54, 617028 (3), Autosomal recessive
<i>TNK2</i>	606994	No OMIM phenotype Mutations in TNK2 in severe autosomal recessive infantile onset epilepsy (Hitomi (2013), Ann Neurol 74(3):496-501), PMID: 23686771 - Autosomal recessive
<i>TNPO2</i>	603002	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556 (3), Autosomal dominant
<i>TNR</i>	601995	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653 (3), Autosomal recessive
<i>TNRC6B</i>	610740	Global developmental delay with speech and behavioral abnormalities, 619243 (3), Autosomal dominant
<i>TOE1</i>	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive

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<i>TOGARAM1</i>	617618	Joubert syndrome 37, 619185 (3), Autosomal recessive
<i>TOR1A</i>	605204	{Dystonia-1, modifier of} (3); Arthrogryposis multiplex congenita 5, 618947 (3), Autosomal recessive; Dystonia-1, torsion, 128100 (3), Autosomal dominant
<i>TP53RK</i>	608679	Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive
<i>TP73</i>	601990	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466 (3), Autosomal recessive
<i>TPI1</i>	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
<i>TPK1</i>	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive
<i>TPO</i>	606765	Thyroid dysmorphogenesis 2A, 274500 (3), Autosomal recessive
<i>TPP1</i>	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
<i>TPRKB</i>	608680	Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive
<i>TRAF7</i>	606692	Cardiac, facial, and digital anomalies with developmental delay, 618164 (3), Autosomal dominant
<i>TRAIP</i>	605958	Seckel syndrome 9, 616777 (3), Autosomal recessive
<i>TRAK1</i>	608112	Developmental and epileptic encephalopathy 68, 618201 (3), Autosomal recessive
<i>TRAPPC10</i>	602103	No OMIM phenotype ESHG2021 - Biallelic TRAPPC10 variants are associated with a microcephalic TRAPPopathy disorder in humans and mice - Autosomal recessive
<i>TRAPPC11</i>	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
<i>TRAPPC12</i>	614139	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive
<i>TRAPPC2L</i>	610970	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 (3), Autosomal recessive
<i>TRAPPC4</i>	610971	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive
<i>TRAPPC6B</i>	610397	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 (3), Autosomal recessive
<i>TRAPPC9</i>	611966	Intellectual developmental disorder, autosomal recessive 13, 613192 (3), Autosomal recessive
<i>TREX1</i>	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal dominant, Autosomal recessive; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
<i>TRIM8</i>	606125	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428 (3), Autosomal dominant

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<i>TRIO</i>	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
<i>TRIP12</i>	604506	Intellectual developmental disorder, autosomal dominant 49, 617752 (3), Autosomal dominant
<i>TRIP4</i>	604501	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive
<i>TRIT1</i>	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
<i>TRMT1</i>	611669	Intellectual developmental disorder, autosomal recessive 68, 618302 (3), Autosomal recessive
<i>TRMT10A</i>	616013	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
<i>TRMT10C</i>	615423	Combined oxidative phosphorylation deficiency 30, 616974 (3), Autosomal recessive
<i>TRNT1</i>	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
<i>TRPM3</i>	608961	No OMIM phenotype Confirmation and Expansion of the Phenotype Associated with the Recurrent p.Val837Met Variant in TRPM3 (de Sainte Agathe (2020), Eur J Med Genet. 63(8):103942), PMID: 32439617 - Autosomal dominant
<i>TRPM6</i>	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
<i>TRRAP</i>	603015	?Deafness, autosomal dominant 75, 618778 (3), Autosomal dominant; Developmental delay with or without dysmorphic facies and autism, 618454 (3), Autosomal dominant
<i>TSC1</i>	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangi leiomyomatosis, 606690 (3)
<i>TSC2</i>	191092	Lymphangi leiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
<i>TSEN15</i>	608756	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
<i>TSEN2</i>	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive
<i>TSEN34</i>	608754	?Pontocerebellar hypoplasia type 2C, 612390 (3), Autosomal recessive
<i>TSEN54</i>	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
<i>TSMF</i>	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
<i>TSHB</i>	188540	Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
<i>TSPAN7</i>	300096	Intellectual developmental disorder, X-linked 58, 300210 (3), X-linked recessive
<i>TTC19</i>	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive

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<i>TTC21B</i>	612014	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal dominant, Autosomal recessive
<i>TTC5</i>	619014	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244 (3), Autosomal recessive
<i>TTC8</i>	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
<i>TTI1</i>	614425	No OMIM phenotype Novel Compound Heterozygous Mutations in TTI2 Cause Syndromic Intellectual Disability in a Chinese Family (Wang (2019), Front Genet 10:1060), PMID: 31737043 - Autosomal Recessive ESHG2021 - Biallelic TTI1 pathogenic variants cause a microcephalic neurodevelopmental disorder
<i>TTI2</i>	614426	Intellectual developmental disorder, autosomal recessive 39, 615541 (3), Autosomal recessive
<i>TUBA1A</i>	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
<i>TUBB</i>	191130	Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant
<i>TUBB2A</i>	615101	Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant
<i>TUBB2B</i>	612850	Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant
<i>TUBB3</i>	602661	Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant
<i>TUBB4A</i>	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
<i>TUBG1</i>	191135	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
<i>TUBGCP2</i>	617817	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 (3), Autosomal recessive
<i>TUBGCP4</i>	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
<i>TUBGCP6</i>	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
<i>TUFM</i>	602389	Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive
<i>TUSC3</i>	601385	Intellectual developmental disorder, autosomal recessive 7, 611093 (3), Autosomal recessive
<i>TWIST1</i>	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
<i>TWNK</i>	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



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No OMIM phenotype		
<i>U2AF2</i>	191318	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
<i>UBA5</i>	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive
<i>UBE2A</i>	312180	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 (3), X-linked recessive
<i>UBE3A</i>	601623	Angelman syndrome, 105830 (3), Autosomal dominant
<i>UBE3B</i>	608047	Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive
<i>UBE4A</i>	603753	Neurodevelopmental disorder with hypotonia and gross motor and seech delay, 619639 (3), Autosomal recessive
<i>UBR1</i>	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
<i>UBR7</i>	613816	Li-Campeau syndrome, 619189 (3), Autosomal recessive
<i>UBTF</i>	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
<i>UFC1</i>	610554	Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive
<i>UFM1</i>	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
<i>UFSP2</i>	611482	?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant; ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant
<i>UGDH</i>	603370	Developmental and epileptic encephalopathy 84, 618792 (3), Autosomal recessive
<i>UGP2</i>	191760	Developmental and epileptic encephalopathy 83, 618744 (3), Autosomal recessive
<i>UMPS</i>	613891	Orotic aciduria, 258900 (3), Autosomal recessive
<i>UNC80</i>	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive
<i>UPB1</i>	606673	Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive
<i>UPF3B</i>	300298	Intellectual developmental disorder, X-linked syndromic 14, 300676 (3), X-linked recessive
<i>UQCRQ</i>	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
<i>UROC1</i>	613012	?Urocanase deficiency, 276880 (3), Autosomal recessive
<i>USP18</i>	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
<i>USP27X</i>	300975	Intellectual developmental disorder, X-linked 105, 300984 (3), X-linked recessive
<i>USP7</i>	602519	Hao-Fountain syndrome, 616863 (3), Autosomal dominant
<i>USP9X</i>	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

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<i>VAMP1</i>	185880	Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
<i>VAMP2</i>	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant
<i>VAR51</i>	192150	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 (3), Autosomal recessive
<i>VLDLR</i>	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
<i>VPS11</i>	608549	?Dystonia 32, 619637 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
<i>VPS13B</i>	607817	Cohen syndrome, 216550 (3), Autosomal recessive
<i>VPS16</i>	608550	Dystonia 30, 619291 (3), Autosomal dominant
<i>VPS37A</i>	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
<i>VPS41</i>	605485	Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive
<i>VPS4A</i>	609982	CIMDAG syndrome, 619273 (3), Autosomal dominant
<i>VPS53</i>	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
<i>VRK1</i>	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
<i>WAC</i>	615049	Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant
<i>WARS2</i>	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
<i>WASF1</i>	605035	Neurodevelopmental disorder with absent language and variable seizures, 618707 (3), Autosomal dominant
<i>WASHC4</i>	615748	Intellectual developmental disorder, autosomal recessive 43, 615817 (3), Autosomal recessive
<i>WASHC5</i>	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
<i>WDFY3</i>	617485	?Microcephaly 18, primary, autosomal dominant, 617520 (3), Autosomal dominant
<i>WDR19</i>	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
<i>WDR26</i>	617424	Skraban-Deardorff syndrome, 617616 (3), Autosomal dominant
<i>WDR37</i>	618586	Neurooculocardio-genitourinary syndrome, 618652 (3), Autosomal dominant

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<i>WDR4</i>	605924	Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive
<i>WDR45</i>	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
<i>WDR45B</i>	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive
<i>WDR5</i>	609012	No OMIM phenotype ESHG2021 - A clustering of missense variants in the crucial chromatin modifier <i>WDR5</i> defines a new neurodevelopmental disorder - Snijders Blok (Nijmegen) - Autosomal dominant
<i>WDR62</i>	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
<i>WDR73</i>	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
<i>WDR81</i>	614218	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive
<i>WFS1</i>	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
<i>WNT1</i>	164820	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3); Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive
<i>WWOX</i>	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
<i>XK</i>	314850	McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked
<i>XPA</i>	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
<i>XPNPEP3</i>	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
<i>XRCC4</i>	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
<i>XYLT1</i>	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
<i>XYLT2</i>	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
<i>YAP1</i>	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 (3), Autosomal dominant
<i>YIF1B</i>	619109	Kaya-Barakat-Masson syndrome, 619125 (3), Autosomal recessive

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<i>YIPF5</i>	611483	Microcephaly, epilepsy, and diabetes syndrome 2, 619278 (3), Autosomal recessive
<i>YME1L1</i>	607472	?Optic atrophy 11, 617302 (3), Autosomal recessive
<i>YWHAG</i>	605356	Developmental and epileptic encephalopathy 56, 617665 (3), Autosomal dominant
<i>YY1</i>	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant
<i>ZBTB11</i>	618181	Intellectual developmental disorder, autosomal recessive 69, 618383 (3), Autosomal recessive
<i>ZBTB16</i>	176797	Leukemia, acute promyelocytic, PL2F/RARA type (3)
<i>ZBTB18</i>	608433	Intellectual developmental disorder, autosomal dominant 22, 612337 (3), Autosomal dominant
<i>ZBTB20</i>	606025	Primrose syndrome, 259050 (3), Autosomal dominant
<i>ZBTB24</i>	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
<i>ZC3H14</i>	613279	Intellectual developmental disorder, autosomal recessive 56, 617125 (3), Autosomal recessive
<i>ZC4H2</i>	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
<i>ZDHHHC9</i>	300646	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799 (3), X-linked
<i>ZEB2</i>	605802	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant
<i>ZFHX3</i>	104155	Prostate cancer, somatic, 176807 (3)
<i>ZFHX4</i>	606940	?Ptosis, congenital, 178300 (2), Autosomal dominant
<i>ZFYVE26</i>	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
<i>ZIC1</i>	600470	?Craniosynostosis 6, 616602 (3), Autosomal dominant; Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 (3), Autosomal dominant
<i>ZIC2</i>	603073	Holoprosencephaly 5, 609637 (3), Autosomal dominant
<i>ZMIZ1</i>	607159	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 (3), Autosomal dominant
<i>ZMYM2</i>	602221	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522 (3), Autosomal dominant
<i>ZMYND11</i>	608668	Intellectual developmental disorder, autosomal dominant 30, 616083 (3), Autosomal dominant
<i>ZNF142</i>	604083	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 (3), Autosomal recessive
<i>ZNF148</i>	601897	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 (3), Autosomal dominant
<i>ZNF292</i>	616213	Intellectual developmental disorder, autosomal dominant 64, 619188 (3), Autosomal dominant
<i>ZNF335</i>	610827	Microcephaly 10, primary, autosomal recessive, 615095 (3), Autosomal recessive
<i>ZNF407</i>	615894	SIMHA syndrome, 619557 (3), Autosomal recessive

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<i>ZNF423</i>	604557	Nephronophthisis 14, 614844 (3), Autosomal dominant, Autosomal recessive; Joubert syndrome 19, 614844 (3), Autosomal dominant, Autosomal recessive
<i>ZNF462</i>	617371	Weiss-Kruszka syndrome, 618619 (3), Autosomal dominant
<i>ZNF526</i>	614387	Dentici-Novelli neurodevelopmental syndrome, 619877 (3), Autosomal recessive
<i>ZNF699</i>	609571	DEGCAGS syndrome, 619488 (3), Autosomal recessive
<i>ZNF711</i>	314990	Intellectual developmental disorder, X-linked 97, 300803 (3), X-linked
<i>ZSWIM6</i>	615951	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant; Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant

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: August 24, 2022

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.