ID_and_epilepsy panel

versie v6 (1689 genen) Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
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, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Intellectual disability and myopathy syndrome, 619719 (3), Autosomal recessive
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

ACO2	100850	?Optic atrophy 9, 616289 (3), Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
ACOX1	609751	Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
ACSF3	614245	Combined malonic and methylmalonic aciduria, 614265 (3)
ACSL4	300157	Intellectual developmental disorder, X-linked 63, 300387 (3), X-linked dominant
ACTB	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
ACTG1	102560	Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant; Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant
ACTL6A	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
ACTL6B	612458	Developmental and epileptic encephalopathy 76, 618468 (3), Autosomal recessive; Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant
ACVR1	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ACY1	104620	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
ADAM22	603709	Developmental and epileptic encephalopathy 61, 617933 (3), Autosomal recessive
ADAR	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
ADARB1	601218	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 (3), Autosomal recessive
ADAT3	615302	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 (3), Autosomal recessive
ADD3	601568	Cerebral palsy, spastic quadriplegic, 3, 617008 (3), Autosomal recessive
ADGRG1	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
ADGRL1	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
ADGRL2	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
ADK	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
ADNP	611386	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
ADPRS	610624	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive
ADSL	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
AFF2	300806	Intellectual developmental disorder, X-linked 109, 309548 (3), X-linked recessive
AFF3	601464	KINSSHIP syndrome, 619297 (3), Autosomal dominant
AFF4	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
AFG3L2	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
AGA	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AGO1	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
AGO2	606229	Lessel-Kreienkamp syndrome, 619149 (3), Autosomal dominant
AGPAT2	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
AGTPBP1	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive
AHCY	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
AHDC1	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
AHI1	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
AIFM1	300169	Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
AIMP1	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
AIMP2	600859	Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive
AK1	103000	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
AKT3	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant

ALDH18A1	138250	Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant
ALDH3A2	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALDH4A1	606811	Hyperprolinemia, type II, 239510 (3), Autosomal recessive
ALDH5A1	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
ALDH7A1	107323	Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive
ALG1	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
ALG11	613666	Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive
ALG12	607144	Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive
ALG13	300776	Developmental and epileptic encephalopathy 36, 300884 (3), X-linked
ALG14	612866	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 (3), Autosomal recessive; Myopathy, epilepsy, and progressive cerebral atrophy, 619036 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3), Autosomal recessive
ALG2	607905	Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
ALG3	608750	Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive
ALG6	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
ALG8	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
ALG9	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive
ALKBH8	613306	Intellectual developmental disorder, autosomal recessive 71, 618504 (3), Autosomal recessive
ALMS1	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ALPL	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
ALX1	601527	Frontonasal dysplasia 3, 613456 (3), Autosomal recessive
ALX4	605420	Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive
AMACR	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive

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

ARF1	103180	Periventricular nodular heterotopia 8, 618185 (3), Autosomal dominant
ARFGEF1	604141	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964 (3), Autosomal dominant
ARFGEF2	605371	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
ARG1	608313	Argininemia, 207800 (3), Autosomal recessive
ARHGAP31	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARHGEF9	300429	Developmental and epileptic encephalopathy 8, 300607 (3), X-linked
ARID1A	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
ARID1B	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
ARID2	609539	Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant
ARL13B	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL6	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
ARMC9	617612	Joubert syndrome 30, 617622 (3), Autosomal recessive
ARSA	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARSL	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ARV1	611647	Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive
ARX	300382	Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Intellectual developmental disorder, X-linked 29, 300419 (3), X-linked recessive
ASAH1	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
ASH1L	607999	Intellectual developmental disorder, autosomal dominant 52, 617796 (3), Autosomal dominant
ASL	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
ASNS	108370	Asparagine synthetase deficiency, 615574 (3), Autosomal recessive
ASPA	608034	Canavan disease, 271900 (3), Autosomal recessive
ASPM	605481	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive
ASS1	603470	Citrullinemia, 215700 (3), Autosomal recessive
ASXL1	612990	Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant
ASXL2	612991	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
ASXL3	615115	Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant

ATAD1	614452	Hyperekplexia 4, 618011 (3), Autosomal recessive
ATAD3A	612316	Harel-Yoon syndrome, 617183 (3), Autosomal dominant, Autosomal recessive; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
ATCAY	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
ATG7	608760	Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive
ATIC	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
ATL1	606439	Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant
ATM	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)
ATN1	607462	Dentatorubral-pallidoluysian atrophy, 125370 (3), Autosomal dominant; Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 (3), Autosomal dominant
ATP13A2	610513	Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive
ATP1A1	182310	Hypomagnesemia, seizures, and mental retardation 2, 618314 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant
ATP1A2	182340	Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP1A3	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Developmental and epileptic encephalopathy 99, 619606 (3), Autosomal dominant
ATP2A2	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
ATP2B1	108731	Intellectual developmental disorder, autosomal dominant 66, 619910 (3), Autosomal dominant
ATP6AP2	300556	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive; Congenital disorder of glycosylation, type IIr, 301045 (3), X-linked recessive
ATP6V0A1	192130	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971 (3), Autosomal recessive; Developmental and epileptic encephalopathy 104, 619970 (3), Autosomal dominant

ATP6V0A2	611716	Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive
ATP6V1A	607027	Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Developmental and epileptic encephalopathy 93, 618012 (3), Autosomal dominant
ATP6V1B2	606939	Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant; Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant
ATP7A	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
ATP8A2	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
ATP9A	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
ATPAF2	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
ATR	601215	Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant
ATRX	300032	Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Intellectual disability-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
ATXN2	601517	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant
AUH	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
AUTS2	607270	Intellectual developmental disorder, autosomal dominant 26, 615834 (3), Autosomal dominant
AVPR2	300538	Diabetes insipidus, nephrogenic, 1, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive
B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
B3GALT6	615291	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive; Al-Gazali syndrome, 609465 (3), Autosomal recessive
B3GLCT	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
B4GALNT1	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
B4GALT1	137060	Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive

B4GALT7	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
B4GAT1	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
B9D1	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
B9D2	611951	?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive
BAZ2B	605683	No OMIM phenotype Developmental delay, intellectual disability, and autism spectrum disorder (Scott (2020), Hum Mutat), PMID: 31999386 - Autosomal dominant
BBS1	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
BBS10	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive
BBS4	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BCAP31	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
BCAS3	607470	Hengel-Maroofian-Schols syndrome, 619641 (3), Autosomal recessive
BCKDHA	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
BCKDK	614901	Branched-chain keto acid dehydrogenase kinase deficiency, 614923 (3)
BCL11A	606557	Dias-Logan syndrome, 617101 (3), Autosomal dominant
BCL11B	606558	Immunodeficiency 49, severe combined, 617237 (3), Autosomal dominant; Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant
BCOR	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BCORL1	300688	Shukla-Vernon syndrome, 301029 (3), X-linked recessive
BCS1L	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive

BICD2	609797	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 (3), Autosomal dominant
BICRA	605690	Coffin-Siris syndrome 12, 619325 (3), Autosomal dominant
BLM	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BLOC1S1	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
BLTP1 (KIAA1109)	611565	Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive
BMP4	112262	Orofacial cleft 11, 600625 (3); Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant
BOLA3	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
BPTF	601819	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755 (3), Autosomal dominant
BRAF	164757	Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3)
BRAT1	614506	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 (3), Autosomal recessive; Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive
BRD4	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
BRF1	604902	Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive
BRPF1	602410	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant
BRSK2	609236	No OMIM phenotype Neurodevelopmental disorder (Hiatt (2019), Am J Hum Genet 104(4):701-708), PMID: 30879638 - Autosomal dominant
BRWD3	300553	Intellectual developmental disorder, X-linked 93, 300659 (3), X-linked recessive
BSCL2	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VC, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3),

		Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
BTD	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
BUB1B	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
C12orf4	616082	Intellectual developmental disorder, autosomal recessive 66, 618221 (3), Autosomal recessive
C12orf57	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
C2CD3	615944	Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
CA2	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CA5A	114761	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive
CA8	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3), Autosomal recessive
CACNA1A	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CACNA1B	601012	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive
CACNA1C	114205	Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3), Autosomal dominant; Brugada syndrome 3, 611875 (3)
CACNA1D	114206	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive
CACNA1E	601013	Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant
CACNA1G	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant
CACNA1H	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)
CACNA1I	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

CACNA2D1	114204	No OMIM phenotype 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
CACNA2D2	607082	Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive
CACNB4	601949	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; Episodic ataxia, type 5, 613855 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant
CACNG2	602911	?Intellectual developmental disorder, autosomal dominant 10, 614256 (3), Autosomal dominant
CAD	114010	Developmental and epileptic encephalopathy 50, 616457 (3), Autosomal recessive
CAMK2A	114078	Intellectual developmental disorder, autosomal dominant 53, 617798 (3), Autosomal dominant; ?Intellectual developmental disorder, autosomal recessive 63, 618095 (3), Autosomal recessive
CAMK2B	607707	Intellectual developmental disorder, autosomal dominant 54, 617799 (3), Autosomal dominant
CAMK2G	602123	Intellectual developmental disorder, autosomal dominant 59, 618522 (3), Autosomal dominant
CAMK4	114080	No OMIM phenotype 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
CAMTA1	611501	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 (3), Autosomal dominant
CAPN10	605286	{Diabetes mellitus, noninsulin-dependent 1}, 601283 (3)
CAPN15	603267	Oculogastrointestinal neurodevelopmental syndrome, 619318 (3), Autosomal recessive
CARS1	123859	Microcephaly, developmental delay, and brittle hair syndrome, 618891 (3), Autosomal recessive
CARS2	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
CASK	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)
CBL	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation
CBS	613381	Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive
CC2D1A	610055	Intellectual developmental disorder, autosomal recessive 3, 608443 (3), Autosomal recessive

CC2D2A	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CCBE1	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
CCDC115	613734	Congenital disorder of glycosylation, type IIo, 616828 (3), Autosomal recessive
CCDC174	616735	Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive
CCDC22	300859	Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive
CCDC47	618260	Trichohepatoneurodevelopmental syndrome, 618268 (3), Autosomal recessive
CCDC88A	609736	?PEHO syndrome-like, 617507 (3), Autosomal recessive
CCDC88C	611204	?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive
CCM2	607929	Cerebral cavernous malformations-2, 603284 (3), Autosomal dominant
CCND2	123833	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant
CCNK	603544	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147 (3), Autosomal dominant
CDC42	116952	Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant
CDC42BPB	614062	Chilton-Okur-Chung neurodevelopmental syndrome, 619841 (3)
CDH11	600023	Teebi hypertelorism syndrome 2, 619736 (3), Autosomal dominant; Elsahy-Waters syndrome, 211380 (3), Autosomal recessive
CDH15	114019	Intellectual developmental disorder, autosomal dominant 3, 612580 (3)
CDH2	114020	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 (3), Autosomal dominant; ?Attention deficit-hyperactivity disorder 8, 619957 (3), Autosomal recessive; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant
CDK10	603464	Al Kaissi syndrome, 617694 (3), Autosomal recessive
CDK13	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant
CDK19	614720	Developmental and epileptic encephalopathy 87, 618916 (3), Autosomal dominant
CDK5RAP2	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
CDK6	603368	?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive
CDK8	603184	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 (3), Autosomal dominant
CDKL5	300203	Developmental and epileptic encephalopathy 2, 300672 (3), X-linked dominant

CDKN1C	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
CDON	608707	Holoprosencephaly 11, 614226 (3), Autosomal dominant
CELF2	602538	Developmental and epileptic encephalopathy 97, 619561 (3), Autosomal dominant
CELSR3	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
CENPF	600236	Stromme syndrome, 243605 (3), Autosomal recessive
CENPJ	609279	Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive
CEP104	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 77, 619988 (3), Autosomal recessive
CEP120	613446	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive
CEP135	611423	Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive
CEP152	613529	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
CEP290	610142	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive
CEP41	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
CEP55	610000	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive
CEP57	607951	Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive
CEP63	614724	?Seckel syndrome 6, 614728 (3), Autosomal recessive
CEP83	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
CEP85L	618865	Lissencephaly 10, 618873 (3), Autosomal dominant
CERS1	606919	Epilepsy, progressive myoclonic, 8, 616230 (3), Autosomal recessive
CERT1	604677	Intellectual developmental disorder, autosomal dominant 34, 616351 (3), Autosomal dominant
CHAMP1	616327	Intellectual developmental disorder, autosomal dominant 40, 616579 (3), Autosomal dominant
CHD1	602118	Pilarowski-Bjornsson syndrome, 617682 (3), Autosomal dominant
CHD2	602119	Developmental and epileptic encephalopathy 94, 615369 (3), Autosomal dominant
CHD3	602120	Snijders Blok-Campeau syndrome, 618205 (3), Autosomal dominant

CHD4	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
CHD5	610771	Parenti-Mignot neurodevelopmental syndrome, 619873 (3), Autosomal dominant
CHD7	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHD8	610528	Intellectual developmental disorder with autism and macrocephaly, 615032 (3), Autosomal dominant
CHKB	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
CHMP1A	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
CHRNA2	118502	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant
CHRNA4	118504	{Nicotine addiction, susceptibility to}, 188890 (3); Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant
CHRNB2	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CIC	612082	Intellectual developmental disorder, autosomal dominant 45, 617600 (3), Autosomal dominant
CILK1	612325	{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant; Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive
CIT	605629	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive
CKAP2L	616174	Filippi syndrome, 272440 (3), Autosomal recessive
CLCN3	600580	Neurodevelopmental disorder with seizures and brain abnormalities, 619517 (3), Autosomal recessive; Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512 (3), Autosomal dominant
CLCN4	302910	Raynaud-Claes syndrome, 300114 (3), X-linked dominant
CLCNKB	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLDN11	601326	Leukodystrophy, hypomyelinating, 22, 619328 (3), Autosomal dominant
CLDN16	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
CLDN19	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
CLIC2	300138	?Intellectual developmental disorder, X-linked syndromic 32, 300886 (3), X-linked recessive
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
CLN6	606725	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive
CLP1	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive

CLPB	616254	Neutropenia, severe congenital, 9, autosomal dominant, 619813 (3), Autosomal dominant; 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 (3), Autosomal recessive; 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 (3), Autosomal dominant
CLTC	118955	Intellectual developmental disorder, autosomal dominant 56, 617854 (3), Autosomal dominant
CNKSR2	300724	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008 (3), X-linked
CNNM2	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation, 616418 (3), Autosomal dominant, Autosomal recessive
CNOT1	604917	Vissers-Bodmer syndrome, 619033 (3), Autosomal dominant; Holoprosencephaly 12, with or without pancreatic agenesis, 618500 (3), Autosomal dominant
CNOT2	604909	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 (3), Autosomal dominant
CNOT3	604910	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 (3), Autosomal dominant
CNPY3	610774	Developmental and epileptic encephalopathy 60, 617929 (3), Autosomal recessive
CNTN2	190197	?Epilepsy, myoclonic, familial adult, 5, 615400 (3), Autosomal recessive
CNTNAP1	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
CNTNAP2	604569	Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; {Autism susceptibility 15}, 612100 (3)
COA8	616003	Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive
COASY	609855	Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
COG1	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
COG4	606976	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant
COG5	606821	Congenital disorder of glycosylation, type IIi, 613612 (3), Autosomal recessive
COG6	606977	Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive
COG7	606978	Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive
COG8	606979	Congenital disorder of glycosylation, type IIh, 611182 (3)
COL18A1	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant

COL4A1	120130	?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant
COL4A2	120090	Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3)
COLEC11	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
COPB1	600959	Baralle-Macken syndrome, 619255 (3), Autosomal recessive
COPB2	606990	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884 (3), Autosomal dominant; ?Microcephaly 19, primary, autosomal recessive, 617800 (3), Autosomal recessive
COQ2	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
COQ4	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive
COQ5	616359	?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive
COQ8A	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COQ9	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
COX10	602125	Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive
COX15	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
COX6B1	124089	Mitochondrial complex IV deficiency, nuclear type 7, 619051 (3), Autosomal recessive
CPA6	609562	Febrile seizures, familial, 11, 614418 (3), Autosomal recessive; Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal dominant, Autosomal recessive
CPE	114855	BDV syndrome, 619326 (3), Autosomal recessive
CPLANE1	614571	Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive
CPLX1	605032	Developmental and epileptic encephalopathy 63, 617976 (3), Autosomal recessive
CPS1	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
CPSF3	606029	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876 (3), Autosomal recessive
CPT2	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
CRADD	603454	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive
CRB2	609720	Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive
CRBN	609262	Intellectual developmental disorder, autosomal recessive 2, 607417 (3), Autosomal recessive
CREBBP	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CRH	122560	No OMIM phenotype Corticotropin-releasing Hormone - Seizures.
CRLF1	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
CRPPA	614631	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive
CSDE1	191510	No OMIM phenotype Autism and related neurodevelopmental disabilities (Guo (2019), Sci Adv 25;5(9):eaax2166), PMID: 31579823 - Autosomal dominant
CSNK1G1	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
CSNK2A1	115440	Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant
CSNK2B	115441	Poirier-Bienvenu neurodevelopmental syndrome, 618732 (3), Autosomal dominant
CSPP1	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CSTB	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
CTBP1	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTCF	604167	Intellectual developmental disorder, autosomal dominant 21, 615502 (3), Autosomal dominant
CTDP1	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
CTNNA2	114025	Cortical dysplasia, complex, with other brain malformations 9, 618174 (3), Autosomal recessive

CTNNB1	116806	Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatricoma, somatic, 132600 (3); Colorectal cancer, somatic, 114500 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3); Ovarian cancer, somatic, 167000 (3); Hepatocellular carcinoma, somatic, 114550 (3)
CTNND1	601045	Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant
CTNND2	604275	No OMIM phenotype Autism (Turner (2015) Nature 520,51), Autosomal dominant Intellectual disability (Hofmeister (2015) J Med Genet 52,111) - Autosomal dominant
CTSA	613111	Galactosialidosis, 256540 (3), Autosomal recessive
CTSD	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
CTSF	603539	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362 (3), Autosomal recessive
CTU2	617057	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 (3), Autosomal recessive
CUBN	602997	[Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive
CUL3	603136	Neurodevelopmental disorder with or without autism or seizures, 619239 (3), Autosomal dominant; Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant
CUL4B	300304	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354 (3), X-linked recessive
CUX1	116896	Global developmental delay with or without impaired intellectual development, 618330 (3), Autosomal dominant
CUX2	610648	Developmental and epileptic encephalopathy 67, 618141 (3), Autosomal dominant
CWC27	617170	Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive
CWF19L1	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
CYB5R3	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
CYC1	123980	Mitochondrial complex III deficiency, nuclear type 6, 615453 (3), Autosomal recessive
CYFIP2	606323	Developmental and epileptic encephalopathy 65, 618008 (3), Autosomal dominant
CYP27A1	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
CYP2U1	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
D2HGDH	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
DAG1	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive

DARS1	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
DARS2	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
DBT	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
DCAF17	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DCC	120470	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant; Esophageal carcinoma, somatic, 133239 (3); Colorectal cancer, somatic, 114500 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive
DCHS1	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
DCPS	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive
DCX	300121	Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked
DDB1	600045	White-Kernohan syndrome, 619426 (3), Autosomal dominant
DDC	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
DDHD2	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
DDX11	601150	Warsaw breakage syndrome, 613398 (3), Autosomal recessive
DDX23	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
DDX3X	300160	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked recessive, X-linked dominant
DDX59	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DDX6	600326	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653 (3), Autosomal dominant
DEAF1	602635	Vulto-van Silfout-de Vries syndrome, 615828 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171 (3), Autosomal recessive
DEGS1	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
DENND5A	617278	Developmental and epileptic encephalopathy 49, 617281 (3), Autosomal recessive
DEPDC5	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant
DHCR24	606418	Desmosterolosis, 602398 (3), Autosomal recessive
DHCR7	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive

DHDDS	608172	Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
DHFR	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
DHPS	600944	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 (3), Autosomal recessive
DHTKD1	614984	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant; Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 (3), Autosomal recessive
DHX16	603405	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant
DHX30	616423	Neurodevelopmental disorder with severe motor impairment and absent language, 617804 (3), Autosomal dominant
DHX37	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
DIAPH1	602121	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
DIS3L2	614184	Perlman syndrome, 267000 (3), Autosomal recessive
DKC1	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DLAT	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DLG3	300189	Intellectual developmental disorder, X-linked 90, 300850 (3), X-linked recessive
DLG4	602887	Intellectual developmental disorder, autosomal dominant 62, 618793 (3), Autosomal dominant
DLL1	606582	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 (3), Autosomal dominant
DMD	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
DMPK	605377	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
DMXL2	612186	Developmental and epileptic encephalopathy 81, 618663 (3), Autosomal recessive; ?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive
DNAJC12	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
DNAJC19	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
DNAJC5	611203	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350 (3), Autosomal dominant

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

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

EIF2B5	603945	Ovarioleukodystrophy, 603896 (3), Autosomal recessive; Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
EIF2S3	300161	MEHMO syndrome, 300148 (3), X-linked recessive
EIF3F	603914	Intellectual developmental disorder, autosomal recessive 67, 618295 (3), Autosomal recessive
EIF4A3	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
EIF5A	600187	Faundes-Banka syndrome, 619376 (3), Autosomal dominant
ELAC2	605367	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3); Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive
ELOVL4	605512	Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive
ELP2	616054	Intellectual developmental disorder, autosomal recessive 58, 617270 (3), Autosomal recessive
EMC1	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
EMC10	614545	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264 (3), Autosomal recessive
EML1	602033	Band heterotopia, 600348 (3), Autosomal recessive
EMX2	600035	Schizencephaly, 269160 (3)
ENTPD1	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
EP300	602700	Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
EPB41L1	602879	?Intellectual developmental disorder, autosomal dominant 11, 614257 (3), Autosomal dominant
EPG5	615068	Vici syndrome, 242840 (3), Autosomal recessive
EPHA7	602190	No OMIM phenotype EPHA7 haploinsufficiency is associated with a neurodevelopmental disorder. (Lévy (2021), Clin Genet. 100(4):396-404), PMID: 34176129 - Autosomal dominant
EPM2A	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
EPRS1	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
ERBB4	600543	Amyotrophic lateral sclerosis 19, 615515 (3), Autosomal dominant
ERCC1	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
ERCC2	126340	Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive
ERCC3	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive

ERCC5	133530	Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
ERCC6	609413	UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
ERCC6L2	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
ERCC8	609412	UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive
ERLIN2	611605	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
ESCO2	609353	Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive
ETFA	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
ETFB	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
ETFDH	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
ETHE1	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
EXOC2	615329	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306 (3), Autosomal recessive
EXOC7	608163	Neurodevelopmental disorder with seizures and brain atrophy, 619072 (3), Autosomal recessive
EXOSC2	602238	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
EXOSC8	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
EXOSC9	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
EXT2	608210	Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant
EXTL3	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
EZH2	601573	Weaver syndrome, 277590 (3), Autosomal dominant
FA2H	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FAM111A	615292	Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant; Gracile bone dysplasia, 602361 (3), Autosomal dominant
FAM149B1	618413	Joubert syndrome 36, 618763 (3), Autosomal recessive

FAM20C	611061	Raine syndrome, 259775 (3), Autosomal recessive
FAM50A	300453	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261 (3), X-linked recessive
FANCD2	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
FAR1	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant
FARS2	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
FARSA	602918	?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive
FARSB	609690	Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive
FAT4	612411	Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive
FBRSL1	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
FBXL3	605653	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 (3), Autosomal recessive
FBXL4	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
FBXO11	607871	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 (3), Autosomal dominant
FBXO28	609100	Developmental and epileptic encephalopathy 100, 619777 (3), Autosomal dominant
FBXO31	609102	?Intellectual developmental disorder, autosomal recessive 45, 615979 (3), Autosomal recessive
FBXW11	605651	Neurodevelopmental, jaw, eye, and digital syndrome, 618914 (3), Autosomal dominant
FBXW7	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
FDFT1	184420	Squalene synthase deficiency, 618156 (3), Autosomal recessive
FGD1	300546	Intellectual developmental disorder, X-linked syndromic 16, 305400 (3), X-linked recessive; Aarskog-Scott syndrome, 305400 (3), X-linked recessive
FGF12	601513	Developmental and epileptic encephalopathy 47, 617166 (3), Autosomal dominant
FGF13	300070	Developmental and epileptic encephalopathy 90, 301058 (3), X-linked recessive, X-linked dominant
FGF14	601515	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant

FGFR1	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
FGFR2	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
FGFR3	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
FH	136850	Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive
FIBP	608296	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive
FIG4	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive
FKRP	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

FKTN	607440	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive
FLNA	300017	Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive
FLVCR1	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FLVCR2	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive
FMN2	606373	Intellectual developmental disorder, autosomal recessive 47, 616193 (3), Autosomal recessive
FMR1	309550	Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Fragile X syndrome, 300624 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked
FOLR1	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
FOXG1	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
FOXP1	605515	Intellectual developmental disorder with language impairment with or without autistic features, 613670 (3), Autosomal dominant
FOXP2	605317	Speech-language disorder-1, 602081 (3), Autosomal dominant
FOXRED1	613622	Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive
FRAS1	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
FREM2	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive
FRMD4A	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
FRMPD4	300838	Intellectual developmental disorder, X-linked 104, 300983 (3), X-linked
FRRS1L	604574	Developmental and epileptic encephalopathy 37, 616981 (3), Autosomal recessive
FTCD	606806	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
FTO	610966	Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive
FTSJ1	300499	Intellectual developmental disorder, X-linked 9, 309549 (3), X-linked recessive
FUCA1	612280	Fucosidosis, 230000 (3), Autosomal recessive
FUT8	602589	Congenital disorder of glycosylation with defective fucosylation 1, 618005 (3), Autosomal recessive

FXYD2	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
FZR1	603619	No OMIM phenotype De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies (Manivannan (2022), Brain. 145(5):1684-1697), PMID: 34788397 - Autosomal dominant
GABBR2	607340	{Nicotine dependence, protection against}, 188890 (3); {Nicotine dependence, susceptibility to}, 188890 (3); Developmental and epileptic encephalopathy 59, 617904 (3), Autosomal dominant; Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant
GABRA1	137160	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Developmental and epileptic encephalopathy 19, 615744 (3), Autosomal dominant; {Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3)
GABRA2	137140	Developmental and epileptic encephalopathy 78, 618557 (3), Autosomal dominant; {Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial
GABRA3	305660	No OMIM phenotype Epileptic Encephalopathy (Bamborschke (2019), Neuropediatrics 50(6)), PMID: 31319422 - Autosomal dominant
GABRA5	137142	Developmental and epileptic encephalopathy 79, 618559 (3), Autosomal dominant
GABRB1	137190	Developmental and epileptic encephalopathy 45, 617153 (3), Autosomal dominant
GABRB2	600232	Developmental and epileptic encephalopathy 92, 617829 (3), Autosomal dominant
GABRB3	137192	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Developmental and epileptic encephalopathy 43, 617113 (3), Autosomal dominant
GABRD	137163	{Epilepsy, idiopathic generalized, 10}, 613060 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to}, 613060 (3), Autosomal dominant; {Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060 (3), Autosomal dominant
GABRG2	137164	Developmental and epileptic encephalopathy 74, 618396 (3), Autosomal dominant; Febrile seizures, familial, 8, 607681 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 3, 607681 (3), Autosomal dominant
GAD1	605363	Developmental and epileptic encephalopathy 89, 619124 (3), Autosomal recessive
GAL	137035	?Epilepsy, familial temporal lobe, 8, 616461 (3), Autosomal dominant
GALC	606890	Krabbe disease, 245200 (3), Autosomal recessive
GALE	606953	Galactose epimerase deficiency, 230350 (3), Autosomal recessive
GALNT2	602274	Congenital disorder of glycosylation, type IIt, 618885 (3), Autosomal recessive
GALT	606999	Galactosemia, 230400 (3), Autosomal recessive
GAMT	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
GATAD2B	614998	GAND syndrome, 615074 (3), Autosomal dominant

GATM	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant
GBA1 (GBA)	606463	{Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant
GCDH	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GCH1	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal dominant, Autosomal recessive; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GCK	138079	MODY, type II, 125851 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 1, 606176 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant
GCSH	238330	?Glycine encephalopathy, 605899 (3), Autosomal recessive
GDI1	300104	Intellectual developmental disorder, X-linked 41, 300849 (3), X-linked dominant
GEMIN4	606969	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913 (3), Autosomal recessive
GEMIN5	607005	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 (3), Autosomal recessive
GFAP	137780	Alexander disease, 203450 (3), Autosomal dominant
GFER	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive
GFM1	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
GFM2	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
GJA1	121014	Erythrokeratodermia 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
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
GJC2	608803	Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive

GK	300474	Glycerol kinase deficiency, 307030 (3), X-linked recessive
GLB1	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
GLDC	238300	Glycine encephalopathy, 605899 (3), Autosomal recessive
GLI2	165230	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant
GLI3	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GLIS3	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
GLRA1	138491	Hyperekplexia 1, 149400 (3), Autosomal dominant, Autosomal recessive
GLRB	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
GLS	138280	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 (3), Autosomal recessive; ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 (3), Autosomal dominant; Developmental and epileptic encephalopathy 71, 618328 (3), Autosomal recessive
GLUD1	138130	Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant
GLUL	138290	Glutamine deficiency, congenital, 610015 (3), Autosomal recessive
GLYCTK	610516	D-glyceric aciduria, 220120 (3), Autosomal recessive
GM2A	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
GMNN	602842	Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant
GMPPA	615495	Alacrima, achalasia, and mental retardation syndrome, 615510 (3), Autosomal recessive
GMPPB	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
GNAI1	139310	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854 (3), Autosomal dominant
GNAO1	139311	Developmental and epileptic encephalopathy 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
GNAQ	600998	Capillary malformations, congenital, 1, somatic, mosaic, 163000 (3); Sturge-Weber syndrome, somatic, mosaic, 185300 (3)

GNAS	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
GNB1	139380	Myelodysplastic syndrome, somatic, 614286 (3); Leukemia, acute lymphoblastic, somatic, 613065 (3); Intellectual developmental disorder, autosomal dominant 42, 616973 (3), Autosomal dominant
GNB2	139390	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 (3), Autosomal dominant; ?Sick sinus syndrome 4, 619464 (3), Autosomal dominant
GNB5	604447	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 (3), Autosomal recessive; Intellectual developmental disorder with cardiac arrhythmia, 617173 (3), Autosomal recessive
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
GNPTAB	607840	Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive; Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive
GNPTG	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GNS	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
GOSR2	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
GOT2	138150	Developmental and epileptic encephalopathy 82, 618721 (3), Autosomal recessive
GPAA1	603048	Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive
GPC3	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
GPC4	300168	Keipert syndrome, 301026 (3), X-linked recessive
GPHN	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
GPT2	138210	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive
GRIA2	138247	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917 (3), Autosomal dominant
GRIA3	305915	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699 (3), X-linked recessive
GRIA4	138246	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 (3), Autosomal dominant
GRID2	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
GRIK2	138244	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580 (3), Autosomal dominant; Intellectual developmental disorder, autosomal recessive 6, 611092 (3), Autosomal recessive

GRIN1	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Developmental and epileptic encephalopathy 101, 619814 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant
GRIN2A	138253	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570 (3), Autosomal dominant
GRIN2B	138252	Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant
GRIN2D	602717	Developmental and epileptic encephalopathy 46, 617162 (3), Autosomal dominant
GRIP1	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
GRM1	604473	Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant
GRM7	604101	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 (3), Autosomal recessive
GRN	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive
GSS	601002	Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive; Glutathione synthetase deficiency, 266130 (3), Autosomal recessive
GTF2E2	189964	Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive
GTF2H5	608780	Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive
GTPBP2	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
GTPBP3	608536	Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive
GUSB	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
H1-4	142220	Rahman syndrome, 617537 (3), Autosomal dominant
H3-3A	601128	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720 (3), Autosomal dominant
H3-3B	601058	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721 (3), Autosomal dominant
H4C11	602826	?Tessadori-van Haaften neurodevelopmental syndrome 2, 619759 (3), Autosomal dominant
H4C3	602827	Tessadori-van Haaften neurodevelopmental syndrome 1, 619758 (3), Autosomal dominant
H4C5	602830	Tessadori-van Haaften neurodevelopmental syndrome 3, 619950 (3), Autosomal dominant
H4C9	602833	Tessadori-van Haaften neurodevelopmental syndrome 4, 619951 (3), Autosomal dominant
HACE1	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive

HADH	601609	Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive; 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive
HADHA	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
HAX1	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
HCCS	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
HCFC1	300019	Methylmalonic aciduria and homocysteinemia, cblX type, 309541 (3), X-linked recessive
HCN1	602780	Developmental and epileptic encephalopathy 24, 615871 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 10, 618482 (3), Autosomal dominant
HCN2	602781	Febrile seizures, familial, 2, 602477 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 11, 602477 (3), Autosomal dominant
HDAC4	605314	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797 (3), Autosomal dominant
HDAC6	300272	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant
HDAC8	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
HECW2	617245	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 (3), Autosomal dominant
HEPACAM	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
HERC1	605109	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive
HERC2	605837	Intellectual developmental disorder, autosomal recessive 38, 615516 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive
HESX1	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
HEXA	606869	[Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive

HGSNAT	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
HIBCH	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
HID1	605752	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983 (3), Autosomal recessive
HIVEP2	143054	Intellectual developmental disorder, autosomal dominant 43, 616977 (3), Autosomal dominant
HK1	142600	Retinitis pigmentosa 79, 617460 (3), Autosomal dominant; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive
HLCS	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
HMGB1	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
HMGCL	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
HNMT	605238	Intellectual developmental disorder, autosomal recessive 51, 616739 (3), Autosomal recessive; {Asthma, susceptibility to}, 600807 (3), Autosomal dominant
HNRNPH1	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
HNRNPH2	300610	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986 (3), X-linked dominant
HNRNPK	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
HNRNPR	607201	No OMIM phenotype Multisystem developmental disorder (Duijkers (2019), Am J Hum Genet ;104(6):1040-1059), PMID: 31079900 - Autosomal dominant
HNRNPU	602869	Developmental and epileptic encephalopathy 54, 617391 (3), Autosomal dominant
HOXA1	142955	Bosley-Salih-Alorainy syndrome, 601536 (3), Autosomal recessive; Athabaskan brainstem dysgenesis syndrome, 601536 (3), Autosomal recessive
HPD	609695	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
HPDL	618994	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 (3), Autosomal recessive; Spastic paraplegia 83, autosomal recessive, 619027 (3), Autosomal recessive
HPRT1	308000	Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive

HRAS	190020	Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant
HS2ST1	604844	Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive
HSD17B10	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
HSD17B4	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
HSPA9	600548	Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant
HSPD1	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
HTRA2	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
HUWE1	300697	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 (3), X-linked
HYCC1 (FAM126A)	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
IARS1	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
IARS2	612801	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
IBA57	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
IDH2	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
IDS	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
IDUA	252800	Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive
IER3IP1	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
IFIH1	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IFT172	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT27	615870	Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive

IGF1	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive
IGF1R	147370	Insulin-like growth factor I, resistance to, 270450 (3), Autosomal dominant, Autosomal recessive
IKBKG	300248	Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked
IL1RAPL1	300206	Intellectual developmental disorder, X-linked 21, 300143 (3), X-linked recessive
IMPA1	602064	Intellectual developmental disorder, autosomal recessive 59, 617323 (3), Autosomal recessive
IMPDH2	146691	[IMPDH2 enzyme activity, variation in], 617995 (3)
INPP5E	613037	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
INPP5K	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
INTS1	611345	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 (3), Autosomal recessive
INTS8	611351	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572 (3), Autosomal recessive
IQSEC1	610166	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 (3), Autosomal recessive
IQSEC2	300522	Intellectual developmental disorder, X-linked 1, 309530 (3), X-linked dominant
IREB2	147582	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive
IRF2BPL	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
IRX5	606195	Hamamy syndrome, 611174 (3), Autosomal recessive
ISCA2	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive
ITGA7	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
ITPA	147520	[Inosine triphosphatase deficiency], 613850 (3); Developmental and epileptic encephalopathy 35, 616647 (3), Autosomal recessive
ITPR1	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
IVD	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive

JAG1	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
JAM2	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive
JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
JARID2	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
KANK1	607704	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
KANSL1	612452	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant
KARS1	601421	Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive; Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 (3), Autosomal recessive
KAT5	601409	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103 (3), Autosomal dominant
KAT6A	601408	Arboleda-Tham syndrome, 616268 (3), Autosomal dominant
KAT6B	605880	SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant
KAT8	609912	Li-Ghorgani-Weisz-Hubshman syndrome, 618974 (3), Autosomal dominant
KATNAL2	614697	No OMIM phenotype Autisme (Sanders (2012) Nature 4;485(7397):237-41 & O'Roak (2012) Nature 485(7397)). PMID: 22495309, 22495306 - Autosomal dominant
KATNB1	602703	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
KATNIP	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
KCNA1	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNA2	176262	Developmental and epileptic encephalopathy 32, 616366 (3), Autosomal dominant
KCNA4	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
KCNB1	600397	Developmental and epileptic encephalopathy 26, 616056 (3), Autosomal dominant
KCNC1	176258	Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant
KCNC2	176256	Developmental and epileptic encephalopathy 103, 619913 (3), Autosomal dominant

KCNC3	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
KCND2	605410	No OMIM phenotype 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
KCND3	605411	Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant; Brugada syndrome 9, 616399 (3), Autosomal dominant
KCNH1	603305	Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant; Temple-Baraitser syndrome, 611816 (3), Autosomal dominant
KCNJ10	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
KCNJ11	600937	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal dominant, Autosomal recessive
KCNJ6	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
KCNK4	605720	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381 (3), Autosomal dominant
KCNK9	605874	Birk-Barel syndrome, 612292 (3)
KCNMA1	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
KCNN2	605879	?Dystonia 34, myoclonic, 619724 (3), Autosomal dominant; Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 (3), Autosomal dominant
KCNN3	602983	Zimmermann-Laband syndrome 3, 618658 (3), Autosomal dominant
KCNQ2	602235	Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
KCNQ3	602232	Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant
KCNQ5	607357	Intellectual developmental disorder, autosomal dominant 46, 617601 (3), Autosomal dominant
KCNT1	608167	Developmental and epileptic encephalopathy 14, 614959 (3), Autosomal dominant; Epilepsy nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant
KCNT2	610044	Developmental and epileptic encephalopathy 57, 617771 (3), Autosomal dominant

KCTD3	613272	No OMIM phenotype Neurodevelopmental and neuropsychiatric disorders (Teng (2019), Review > CNS Neurosci Ther ;25(7):887-902), PMID: 31197948 - Autosomal dominant
KCTD7	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
KDM1A	609132	Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant
KDM3B	609373	Diets-Jongmans syndrome, 618846 (3), Autosomal dominant
KDM4B	609765	Intellectual developmental disorder, autosomal dominant 65, 619320 (3), Autosomal dominant
KDM5B	605393	Intellectual developmental disorder, autosomal recessive 65, 618109 (3), Autosomal recessive
KDM5C	314690	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
KDM6A	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KDM6B	611577	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505 (3), Autosomal dominant
KIAA0586	610178	Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive
KIDINS220	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant; Ventriculomegaly and arthrogryposis, 619501 (3), Autosomal recessive
KIF11	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
KIF14	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive
KIF1A	601255	NESCAV syndrome, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal dominant, Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal dominant, Autosomal recessive
KIF21B	608322	No OMIM phenotype 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
KIF2A	602591	Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant
KIF4A	300521	?Intellectual developmental disorder, X-linked 100, 300923 (3), X-linked recessive
KIF5A	602821	Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant
KIF5C	604593	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant

KIF7	611254	Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
KIFBP	609367	Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive
KLF7	604865	No OMIM phenotype Developmental delay/intellectual disability, neuromuscular and psychiatric symptoms (Powis (2018), Clin Genet 93(5)), PMID: 29251763 - Autosomal dominant
KLHL15	300980	Intellectual developmental disorder, X-linked 103, 300982 (3), X-linked recessive
KLHL7	611119	Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive
KMT2A	159555	Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
KMT2B	606834	Intellectual developmental disorder, autosomal dominant 68, 619934 (3), Autosomal dominant; Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
KMT2C	606833	Kleefstra syndrome 2, 617768 (3), Autosomal dominant
KMT2D	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
KMT2E	608444	O'Donnell-Luria-Rodan syndrome, 618512 (3), Autosomal dominant
KMT5B	610881	Intellectual developmental disorder, autosomal dominant 51, 617788 (3), Autosomal dominant
KNL1	609173	Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive
KPTN	615620	Intellectual developmental disorder, autosomal recessive 41, 615637 (3), Autosomal recessive
KRAS	190070	Gastric cancer, somatic, 613659 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3)
KRIT1	604214	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 (3), Autosomal dominant; Cerebral cavernous malformations-1, 116860 (3), Autosomal dominant; Cavernous malformations of CNS and retina, 116860 (3), Autosomal dominant
L1CAM	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

L2HGDH	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LAMA1	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
LAMA2	156225	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive
LAMB1	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
LAMC3	604349	Cortical malformations, occipital, 614115 (3), Autosomal recessive
LAMP2	309060	Danon disease, 300257 (3), X-linked dominant
LARGE1	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
LARP7	612026	Alazami syndrome, 615071 (3), Autosomal recessive
LARS1	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
LAS1L	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
LGI1	604619	Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant
LGI4	608303	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive
LIAS	607031	Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive
LIG4	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
LINGO1	609791	Intellectual developmental disorder, autosomal recessive 64, 618103 (3), Autosomal recessive
LINS1	610350	Intellectual developmental disorder, autosomal recessive 27, 614340 (3), Autosomal recessive
LIPT1	610284	Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive
LMAN2L	609552	?Intellectual developmental disorder, autosomal dominant 69, 617863 (3); ?Intellectual developmental disorder, autosomal recessive 52, 616887 (3), Autosomal recessive
LMBRD2	619490	Developmental delay with variable neurologic and brain abnormalities, 619694 (3), Autosomal dominant
LMNB1	150340	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant; Microcephaly 26, primary, autosomal dominant, 619179 (3), Autosomal dominant
LMNB2	150341	Microcephaly 27, primary, autosomal dominant, 619180 (3), Autosomal dominant; ?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant
LONP1	605490	CODAS syndrome, 600373 (3), Autosomal recessive
LRP2	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRPPRC	607544	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive

LSS	600909	Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-intellectual disability syndrome 4, 618840 (3), Autosomal recessive
LYRM7	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
LYST	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
LZTFL1	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
LZTR1	600574	Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
MAB21L1	601280	Cerebellar, ocular, craniofacial, and genital syndrome, 618479 (3), Autosomal recessive
MAB21L2	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal dominant, Autosomal recessive
MACF1	608271	Lissencephaly 9 with complex brainstem malformation, 618325 (3), Autosomal dominant
MADD	603584	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 (3), Autosomal recessive; DEEAH syndrome, 619004 (3), Autosomal recessive
MAF	177075	Cataract 21, multiple types, 610202 (3), Autosomal dominant; Ayme-Gripp syndrome, 601088 (3), Autosomal dominant
MAG	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
MAGEL2	605283	Schaaf-Yang syndrome, 615547 (3), Autosomal dominant
MAGT1	300715	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3), X-linked recessive; Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive
MAN1B1	604346	Rafiq syndrome, 614202 (3), Autosomal recessive
MAN2B1	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MANBA	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
MAOA	309850	{Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive
MAP1B	157129	?Deafness, autosomal dominant 83, 619808 (3), Autosomal dominant; Periventricular nodular heterotopia 9, 618918 (3), Autosomal dominant
MAP2K1	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
MAP2K2	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
MAPK1	176948	Noonan syndrome 13, 619087 (3), Autosomal dominant
MAPK8IP3	605431	Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant
MAPKAPK5	606723	Neurocardiofaciodigital syndrome, 619869 (3), Autosomal recessive
MAPRE2	605789	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant

MASP1	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
MAST1	612256	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 (3), Autosomal dominant
MAT1A	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
MBD5	611472	Intellectual developmental disorder, autosomal dominant 1, 156200 (3), Autosomal dominant
MBOAT7	606048	Intellectual developmental disorder, autosomal recessive 57, 617188 (3), Autosomal recessive
MBTPS2	300294	Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
MCCC1	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive
MCM3AP	603294	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive
MCOLN1	605248	Mucolipidosis IV, 252650 (3), Autosomal recessive
MCPH1	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
MDH2	154100	Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive
MECP2	300005	Rett syndrome, atypical, 312750 (3), X-linked dominant; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Intellectual developmental disorder, X-linked syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
MECR	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive
MED12	300188	Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Hardikar syndrome, 301068 (3), X-linked dominant; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive
MED12L	611318	Nizon-Isidor syndrome, 618872 (3), Autosomal dominant
MED13	603808	Intellectual developmental disorder, autosomal dominant 61, 618009 (3), Autosomal dominant
MED13L	608771	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive

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

MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MMADHC	611935	Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive
MMUT	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
MN1	156100	CEBALID syndrome, 618774 (3), Autosomal dominant; Meningioma, 607174 (3), Autosomal dominant
MOCS1	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
MOCS2	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
MOGS	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MORC2	616661	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant; Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 (3), Autosomal dominant
MPDU1	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MPDZ	603785	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive
MPLKIP	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
MRAS	608435	Noonan syndrome 11, 618499 (3), Autosomal dominant
MRPL3	607118	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
MRPS22	605810	Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
MRPS34	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
MSL3	300609	Basilicata-Akhtar syndrome, 301032 (3), X-linked dominant
MSMO1	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive
MTFMT	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
MTHFR	607093	{Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive
MTHFS	604197	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 (3), Autosomal recessive
MTO1	614667	Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive
MTOR	601231	Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant

MTR	156570	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive
MTRFR	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MVK	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MYCN	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant
MYO5A	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
MYT1L	613084	Intellectual developmental disorder, autosomal dominant 39, 616521 (3), Autosomal dominant
NAA10	300013	Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
NAA15	608000	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 (3), Autosomal dominant
NAA20	610833	Intellectual developmental disorder, autosomal recessive 73, 619717 (3), Autosomal recessive
NACC1	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant
NAGA	104170	Schindler disease, type I, 609241 (3), Autosomal recessive; Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
NAGLU	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
NAGS	608300	N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive
NALCN	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant; Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive
NANS	605202	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive
NAPB	611270	No OMIM phenotype 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

NARS1	108410	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 (3), Autosomal dominant; Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091 (3), Autosomal recessive
NARS2	612803	Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive; ?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive
NAXD	615910	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive
NAXE	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
NBEA	604889	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157 (3), Autosomal dominant
NBN	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NCAPG2	608532	Khan-Khan-Katsanis syndrome, 618460 (3), Autosomal recessive
NCDN	608458	Neurodevelopmental disorder with infantile epileptic spasms, 619373 (3), Autosomal dominant
NCKAP1	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
NCOR1	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
NDE1	609449	Microhydranencephaly, 605013 (3), Autosomal recessive; Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive
NDP	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
NDST1	600853	Intellectual developmental disorder, autosomal recessive 46, 616116 (3), Autosomal recessive
NDUFA1	300078	Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive
NDUFA10	603835	Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive
NDUFA11	612638	Mitochondrial complex I deficiency, nuclear type 14, 618236 (3), Autosomal recessive
NDUFA12	614530	Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
NDUFA2	602137	Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive
NDUFA6	602138	Mitochondrial complex I deficiency, nuclear type 33, 618253 (3), Autosomal recessive
NDUFA8	603359	Mitochondrial complex I deficiency, nuclear type 37, 619272 (3), Autosomal recessive

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

NFIA	600727	Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant
NFIB	600728	Macrocephaly, acquired, with impaired intellectual development, 618286 (3), Autosomal dominant
NFIX	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Malan syndrome, 614753 (3), Autosomal dominant
NFU1	608100	Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive
NGLY1	610661	Congenital disorder of deglycosylation 1, 615273 (3), Autosomal recessive
NHLRC1	608072	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
NHS	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
NIPBL	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NKAP	300766	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039 (3), X-linked recessive
NKX2-1	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant
NLGN3	300336	{Asperger syndrome susceptibility, X-linked 1}, 300494 (3), Multifactorial, Isolated cases, X-linked; {Autism susceptibility, X-linked 1}, 300425 (3), X-linked
NLGN4X	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
NLRP3	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NONO	300084	Intellectual developmental disorder, X-linked syndromic 34, 300967 (3), X-linked
NOVA2	601991	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 (3), Autosomal dominant
NPC1	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NPHP1	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPRL2	607072	Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant
NPRL3	600928	Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant

NR2F1	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
NR4A2	601828	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911 (3), Autosomal dominant
NRAS	164790	Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3)
NRROS	615322	Seizures, early-onset, with neurodegeneration and brain calcification, 618875 (3), Autosomal recessive
NRXN1	600565	Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3)
NSD1	606681	Sotos syndrome, 117550 (3), Autosomal dominant
NSD2	602952	Rauch-Steindl syndrome, 619695 (3), Autosomal dominant
NSDHL	300275	CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant
NSUN2	610916	Intellectual developmental disorder, autosomal recessive 5, 611091 (3), Autosomal recessive
NT5C2	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
NTNG2	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive
NTRK2	600456	Developmental and epileptic encephalopathy 58, 617830 (3), Autosomal dominant; Obesity, hyperphagia, and developmental delay, 613886 (3), Autosomal dominant
NUBPL	613621	Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive
NUDT2	602852	Intellectual developmental disorder with or without peripheral neuropathy, 619844 (3), Autosomal recessive
NUP107	607617	?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
NUP188	615587	Sandestig-Stefanova syndrome, 618804 (3), Autosomal recessive
NUP214	114350	Leukemia, T-cell acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, somatic, 601626 (3); {Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 (3), Autosomal recessive
NUP62	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
NUS1	610463	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive
OAT	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive

OCLN	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
OCRL	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
ODC1	165640	Bachmann-Bupp syndrome, 619075 (3), Autosomal dominant
OFD1	300170	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive
OGT	300255	Intellectual developmental disorder, X-linked 106, 300997 (3), X-linked recessive
OPHN1	300127	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 (3), X-linked recessive
ORC1	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
OSGEP	610107	Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive
OTC	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked
OTUD5	300713	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056 (3), X-linked recessive
OTUD6B	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive
OTX2	600037	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant
OXR1	605609	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 (3), Autosomal recessive
P4HTM	614584	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 (3), Autosomal recessive
PABPC1	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
PACS1	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
PACS2	610423	Developmental and epileptic encephalopathy 66, 618067 (3), Autosomal dominant
PAFAH1B1	601545	Subcortical laminar heterotopia, 607432 (3), Autosomal dominant; Lissencephaly 1, 607432 (3), Autosomal dominant
PAH	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
PAK1	602590	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158 (3), Autosomal dominant
PAK3	300142	Intellectual developmental disorder, X-linked 30, 300558 (3), X-linked recessive

PANK2	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PARN	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant
PARP6	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
PARS2	612036	Developmental and epileptic encephalopathy 75, 618437 (3), Autosomal recessive
PAX1	167411	Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
PAX6	607108	Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant
PAX8	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
PBX1	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
PC	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
PCCA	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PCDH12	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
PCDH19	300460	Developmental and epileptic encephalopathy 9, 300088 (3), X-linked
PCDHGC4	606305	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880 (3), Autosomal recessive
PCGF2	600346	Turnpenny-Fry syndrome, 618371 (3), Autosomal dominant
PCLO	604918	?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive
PCNT	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
PCYT2	602679	Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive
PDCD10	609118	Cerebral cavernous malformations-3, 603285 (3), Autosomal dominant
PDE2A	602658	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 (3), Autosomal recessive
PDE4D	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant
PDE6D	602676	Joubert syndrome 22, 615665 (3), Autosomal recessive

PDGFRB	173410	Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive
PDHX	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PDP1	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
PDSS1	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
PDSS2	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
PDX1	600733	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; Pancreatic agenesis 1, 260370 (3), Autosomal recessive; MODY, type IV, 606392 (3)
PEPD	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PET100	614770	Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive
PEX1	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive
PEX10	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
PEX11B	603867	Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
PEX12	601758	Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive
PEX13	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
PEX14	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
PEX16	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
PEX19	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
PEX2	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
PEX26	608666	Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive

PEX3	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
PEX5	600414	Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
PEX6	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
PEX7	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
PGAP1	611655	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 (3), Autosomal recessive
PGAP2	615187	Hyperphosphatasia with mental retardation syndrome 3, 614207 (3), Autosomal recessive
PGAP3	611801	Hyperphosphatasia with mental retardation syndrome 4, 615716 (3), Autosomal recessive
PGK1	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PGM2L1	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
PGM3	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PHACTR1	608723	Developmental and epileptic encephalopathy 70, 618298 (3), Autosomal dominant
PHF21A	608325	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 (3), Autosomal dominant
PHF6	300414	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
PHF8	300560	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263 (3), X-linked recessive
PHGDH	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PHIP	612870	Chung-Jansen syndrome, 617991 (3), Autosomal dominant
PI4KA	600286	Spastic paraplegia 84, autosomal recessive, 619621 (3), Autosomal recessive; Gastrointestinal defects and immunodeficiency syndrome 2, 619708 (3), Autosomal recessive; Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
PIBF1	607532	Joubert syndrome 33, 617767 (3), Autosomal recessive

PIDD1	605247	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827 (3), Autosomal recessive
PIGA	311770	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3); Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 (3)
PIGB	604122	Developmental and epileptic encephalopathy 80, 618580 (3), Autosomal recessive
PIGC	601730	Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive
PIGF	600153	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356 (3), Autosomal recessive
PIGG	616918	[Blood group, EMM system], 619812 (3), Autosomal recessive; Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917 (3), Autosomal recessive
PIGH	600154	Glycosylphosphatidylinositol biosynthesis defect 17, 618010 (3), Autosomal recessive
PIGK	605087	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 (3), Autosomal recessive
PIGL	605947	CHIME syndrome, 280000 (3), Autosomal recessive
PIGN	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive
PIGO	614730	Hyperphosphatasia with mental retardation syndrome 2, 614749 (3), Autosomal recessive
PIGP	605938	Developmental and epileptic encephalopathy 55, 617599 (3), Autosomal recessive
PIGQ	605754	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548 (3), Autosomal recessive
PIGS	610271	Developmental and epileptic encephalopathy 95, 618143 (3), Autosomal recessive
PIGT	610272	?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive
PIGU	608528	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 (3), Autosomal recessive
PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive
PIGW	610275	Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive
PIGY	610662	Hyperphosphatasia with mental retardation syndrome 6, 616809 (3), Autosomal recessive
PIK3C2B	602838	No OMIM phenotype Defective lipid signalling caused by mutations in PIK3C2B underlies focal epilepsy (Gozzelino (2022), Brain. 145(7):2313-2331), PMID: 35786744 - Autosomal dominant

PIK3CA	171834	CLOVE syndrome, somatic, 612918 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Cerebral cavernous malformations 4, somatic, 619538 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); Macrodactyly, somatic, 155500 (3); CLAPO syndrome, somatic, 613089 (3); Keratosis, seborrheic, somatic, 182000 (3); Nevus, epidermal, somatic, 162900 (3); Gastric cancer, somatic, 613659 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3)
PIK3R2	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant
PISD	612770	Liberfarb syndrome, 618889 (3), Autosomal recessive
PITRM1	618211	Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive
PLA2G6	603604	Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive
PLAA	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive
PLCB1	607120	Developmental and epileptic encephalopathy 12, 613722 (3), Autosomal recessive
PLCH1	612835	Holoprosencephaly 14, 619895 (3), Autosomal recessive
PLK4	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
PLP1	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PLPBP	604436	Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive
PLXNA1	601055	Dworschak-Punetha neurodevelopmental syndrome, 619955 (3), Autosomal recessive
PLXND1	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)
PMM2	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PMPCA	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
PMPCB	603131	Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive
PNKP	605610	?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
PNP	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive

PNPLA6	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
PNPT1	610316	Spinocerebellar ataxia 25, 608703 (3), Autosomal dominant; Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive
POC1A	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
POC1B	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
POGZ	614787	White-Sutton syndrome, 616364 (3), Autosomal dominant
POLA1	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
POLG	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POLR1C	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
POLR2A	180660	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 (3), Autosomal dominant
POLR3A	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3B	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 (3), Autosomal dominant
POMGNT1	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
POMGNT2	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive

POMK	615247	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive
POMT1	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive
POMT2	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
PORCN	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
POU1F1	173110	Pituitary hormone deficiency, combined or isolated, 1, 613038 (3), Autosomal dominant, Autosomal recessive
POU3F3	602480	Snijders Blok-Fisher syndrome, 618604 (3), Autosomal dominant
PPFIBP1	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
PPIL1	601301	Pontocerebellar hypoplasia, type 14, 619301 (3), Autosomal recessive
PPM1D	605100	Breast cancer, somatic, 114480 (3); Jansen de Vries syndrome, 617450 (3), Autosomal dominant
PPP1CB	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
PPP1R12A	602021	Genitourinary and/or/brain malformation syndrome, 618820 (3), Autosomal dominant
PPP1R15B	613257	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
PPP1R21	618159	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383 (3), Autosomal recessive
PPP2CA	176915	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354 (3), Autosomal dominant
PPP2R1A	605983	Intellectual developmental disorder, autosomal dominant 36, 616362 (3), Autosomal dominant
PPP2R5D	601646	Intellectual developmental disorder, autosomal dominant 35, 616355 (3), Autosomal dominant
PPP3CA	114105	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 (3), Autosomal dominant; Developmental and epileptic encephalopathy 91, 617711 (3), Autosomal dominant
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive

PQBP1	300463	Renpenning syndrome, 309500 (3), X-linked recessive
PRDM13	616741	Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive
PRDM15	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
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
PRICKLE1	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
PRICKLE2	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
PRIMA1	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
PRKACB	176892	Cardioacrofacial dysplasia 2, 619143 (3), Autosomal dominant, Somatic mosaicism
PRKAR1A	188830	Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant
PRKAR1B	176911	Marbach-Schaaf neurodevelopmental syndrome, 619680 (3), Autosomal dominant
PRMT7	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
PRODH	606810	{Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant; Hyperprolinemia, type I, 239500 (3), Autosomal recessive
PRPS1	311850	Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive
PRR12	616633	Neuroocular syndrome, 619539 (3), Autosomal dominant
PRRT2	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant
PRSS12	606709	Intellectual developmental disorder, autosomal recessive 1, 249500 (3), Autosomal recessive

PRUNE1	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive
PSAP	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); {Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 (3), Autosomal dominant
PSAT1	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PSMD12	604450	Stankiewicz-Isidor syndrome, 617516 (3), Autosomal dominant
PSPH	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
PTCH1	601309	Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant; Basal cell nevus syndrome, 109400 (3), Autosomal dominant
PTCHD1	300828	{Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive
PTDSS1	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
PTEN	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
PTPN11	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
PTPN23	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive
PTPN4	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
PTRH2	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
PTRHD1	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
PTS	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
PUF60	604819	Verheij syndrome, 615583 (3), Autosomal dominant
PUM1	607204	Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant

PURA	600473	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158 (3), Autosomal dominant
PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
PUS3	616283	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 (3), Autosomal recessive
PUS7	616261	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 (3), Autosomal recessive
PYCR1	179035	Cutis laxa, autosomal recessive, type IIIB, 614438 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive
PYCR2	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
QARS1	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
QRICH1	617387	Ververi-Brady syndrome, 617982 (3), Autosomal dominant
RAB11B	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
RAB18	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
RAB23	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
RAB27A	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
RAB39B	300774	Intellectual developmental disorder, X-linked 72, 300271 (3), X-linked recessive; Waisman syndrome, 311510 (3), X-linked recessive
RAB3GAP1	602536	Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive
RAB3GAP2	609275	Martsolf syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
RABGAP1	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
RAC1	602048	Intellectual developmental disorder, autosomal dominant 48, 617751 (3), Autosomal dominant
RAC3	602050	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 (3), Autosomal dominant
RAD21	606462	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant; ?Mungan syndrome, 611376 (3), Autosomal recessive
RAF1	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3)

RAI1	607642	Smith-Magenis syndrome, 182290 (3), Autosomal dominant, Isolated cases
RALA	179550	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311 (3), Autosomal dominant
RALGAPA1	608884	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 (3), Autosomal recessive
RARB	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal dominant, Autosomal recessive
RARS2	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
RBBP8	604124	Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3)
RBFOX1	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)
RBM10	300080	TARP syndrome, 311900 (3), X-linked recessive
RBM28	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
RBPJ	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
RCBTB1	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
RELN	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
RERE	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
RFT1	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
RFX3	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
RFX4	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
RFX7	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

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

RRAS2	600098	Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant
RRM2B	604712	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
RSPRY1	616585	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive
RSRC1	613352	Intellectual developmental disorder, autosomal recessive 70, 618402 (3), Autosomal recessive
RTEL1	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
RTN4IP1	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive
RTTN	610436	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
RUBCN	613516	Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
RUSC2	611053	Intellectual developmental disorder, autosomal recessive 61, 617773 (3), Autosomal recessive
RXYLT1	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
SALL1	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant
SAMD9	610456	Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant
SAMHD1	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
SARS1	607529	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive
SATB1	602075	Kohlschutter-Tonz syndrome-like, 619229 (3), Autosomal dominant; Developmental delay with dysmorphic facies and dental anomalies, 619228 (3), Autosomal dominant
SATB2	608148	Glass syndrome, 612313 (3), Autosomal dominant
SBDS	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
SBF1	603560	Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive
SC5D	602286	Lathosterolosis, 607330 (3), Autosomal recessive

SCAF4	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
SCAMP5	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
SCAPER	611611	Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive
SCARB2	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
SCN1A	182389	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Dravet syndrome, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 2, 604403 (3), Autosomal dominant
SCN1B	600235	Generalized epilepsy with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Developmental and epileptic encephalopathy 52, 617350 (3), Autosomal recessive; Cardiac conduction defect, nonspecific, 612838 (3); Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3)
SCN2A	182390	Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant; Developmental and epileptic encephalopathy 11, 613721 (3), Autosomal dominant; Episodic ataxia, type 9, 618924 (3), Autosomal dominant
SCN3A	182391	Epilepsy, familial focal, with variable foci 4, 617935 (3), Autosomal dominant; Developmental and epileptic encephalopathy 62, 617938 (3), Autosomal dominant
SCN8A	600702	?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Developmental and epileptic encephalopathy 13, 614558 (3), Autosomal dominant
SCO1	603644	Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive
SCO2	604272	Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive
SCYL1	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
SDCCAG8	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
SDHA	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Paragangliomas 5, 614165 (3), Autosomal dominant

SDHAF1	612848	Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive
SEMA3E	608166	?CHARGE syndrome, 214800 (3), Autosomal dominant
SEMA6B	608873	Epilepsy, progressive myoclonic, 11, 618876 (3), Autosomal dominant
SEPSECS	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
SERAC1	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SERPINI1	602445	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3), Autosomal dominant
SET	600960	Intellectual developmental disorder, autosomal dominant 58, 618106 (3), Autosomal dominant
SETBP1	611060	Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 29, 616078 (3), Autosomal dominant
SETD1A	611052	Epilepsy, early-onset, with or without developmental delay, 618832 (3), Autosomal dominant; Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056 (3), Autosomal dominant
SETD1B	611055	Intellectual developmental disorder with seizures and language delay, 619000 (3), Autosomal dominant
SETD2	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant
SETD5	615743	Intellectual developmental disorder, autosomal dominant 23, 615761 (3), Autosomal dominant
SFXN4	615564	Combined oxidative phosphorylation deficiency 18, 615578 (3), Autosomal recessive
SGPL1	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
SGSH	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
SHANK1	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
SHANK2	603290	{Autism susceptibility 17}, 613436 (3)
SHANK3	606230	Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3), Autosomal dominant
SHH	600725	Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant
SHMT2	138450	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 (3), Autosomal recessive
SHOC2	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
SHROOM4	300579	Intellectual developmental disorder, X-linked syndromic, Stocco dos Santos type, 300434 (3), X-linked
SIAH1	602212	Buratti-Harel syndrome, 619314 (3), Autosomal dominant

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

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

SLC9A7	300368	Intellectual developmental disorder, X-linked 108, 301024 (3), X-linked recessive
SMAD4	600993	Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant
SMAD6	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Radioulnar synostosis, nonsyndromic}, 179300 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
SMARCA2	600014	Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant; Blepharophimosis-impaired intellectual development syndrome, 619293 (3), Autosomal dominant
SMARCA4	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant
SMARCA5	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
SMARCB1	601607	Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant
SMARCC2	601734	Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant
SMARCD1	601735	Coffin-Siris syndrome 11, 618779 (3), Autosomal dominant
SMARCE1	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
SMC1A	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 (3), X-linked dominant
SMC3	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
SMG8	613175	Alzahrani-Kuwahara syndrome, 619268 (3), Autosomal recessive
SMG9	613176	Heart and brain malformation syndrome, 616920 (3), Autosomal recessive; Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995 (3), Autosomal recessive
SMOC1	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SMPD1	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
SMPD4	610457	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive

SMS	300105	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 (3), X-linked recessive
SNAP25	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
SNIP1	608241	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501 (3), Autosomal recessive
SNORD118	616663	Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive
SNRPN	182279	Prader-Willi syndrome, 176270 (3), Autosomal dominant
SNX14	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
SNX27	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
SOBP	613667	Mental retardation, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive
SON	182465	ZTTK syndrome, 617140 (3), Autosomal dominant
SOS1	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
SOS2	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SOX10	602229	Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant
SOX11	600898	Coffin-Siris syndrome 9, 615866 (3), Autosomal dominant
SOX2	184429	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant; Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant
SOX3	313430	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
SOX4	184430	Coffin-Siris syndrome 10, 618506 (3), Autosomal dominant
SOX5	604975	Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant
SOX6	607257	Tolchin-Le Caignec syndrome, 618971 (3), Autosomal dominant
SPART	607111	Troyer syndrome, 275900 (3), Autosomal recessive
SPAST	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
SPATA5	613940	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 (3), Autosomal recessive

SPATA5L1	619578	Deafness, autosomal recessive 119, 619615 (3), Autosomal recessive; Neurodevelopmental disorder with hearing loss and spasticity, 619616 (3), Autosomal recessive
SPECC1L	614140	Teebi hypertelorism syndrome 1, 145420 (3), Autosomal dominant; ?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant
SPEN	613484	Radio-Tartaglia syndrome, 619312 (3), Autosomal dominant
SPG11	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
SPOP	602650	Nabais Sa-de Vries syndrome, type 1, 618828 (3), Autosomal dominant; Nabais Sa-de Vries syndrome, type 2, 618829 (3), Autosomal dominant
SPR	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
SPRED1	609291	Legius syndrome, 611431 (3), Autosomal dominant
SPTAN1	182810	Developmental and epileptic encephalopathy 5, 613477 (3), Autosomal dominant
SPTBN1	182790	Developmental delay, impaired speech, and behavioral abnormalities, 619475 (3), Autosomal dominant
SPTBN2	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
SPTBN4	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive
SRCAP	611421	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 (3), Autosomal dominant; Floating-Harbor syndrome, 136140 (3), Autosomal dominant
SRD5A3	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
SRPX2	300642	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643 (3)
SRRM2	606032	No OMIM phenotype Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder (Cuinat (2022), Genet Med. 24(8):1774-1780), PMID: 35567594 - Autosomal dominant
SSR4	300090	Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive
ST3GAL3	606494	Developmental and epileptic encephalopathy 15, 615006 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 12, 611090 (3), Autosomal recessive
ST3GAL5	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
STAG1	604358	Intellectual developmental disorder, autosomal dominant 47, 617635 (3), Autosomal dominant
STAG2	300826	Holoprosencephaly 13, X-linked, 301043 (3), X-linked recessive, X-linked dominant; Mullegama-Klein-Martinez syndrome, 301022 (3), X-linked

STAMBP	606247	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
STEEP1	301012	?Intellectual developmental disorder, X-linked 107, 301013 (3), X-linked
STIL	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
STRA6	610745	Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive
STRADA	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive
STT3A	601134	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714 (3), Autosomal dominant; Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596 (3), Autosomal recessive
STT3B	608605	Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
STX1B	601485	Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant
STXBP1	602926	Developmental and epileptic encephalopathy 4, 612164 (3), Autosomal dominant
SUCLA2	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SUCLG1	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
SUFU	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
SUMF1	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
SUOX	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
SUPT16H	605012	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480 (3), Autosomal dominant
SURF1	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive
SUZ12	606245	Imagawa-Matsumoto syndrome, 618786 (3), Autosomal dominant
SVBP	617853	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive
SYN1	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
SYNCRIP	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

SYNE1	608441	Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
SYNGAP1	603384	Intellectual developmental disorder, autosomal dominant 5, 612621 (3), Autosomal dominant
SYNJ1	604297	Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive; Developmental and epileptic encephalopathy 53, 617389 (3), Autosomal recessive
SYP	313475	Intellectual developmental disorder, X-linked 96, 300802 (3), X-linked recessive
SYT1	185605	Baker-Gordon syndrome, 618218 (3), Autosomal dominant
SYT14	610949	?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive
SZT2	615463	Developmental and epileptic encephalopathy 18, 615476 (3), Autosomal recessive
TAF1	313650	Intellectual developmental disorder, X-linked syndromic 33, 300966 (3), X-linked recessive; Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive
TAF13	600774	Intellectual developmental disorder, autosomal recessive 60, 617432 (3), Autosomal recessive
TAF1C	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
TAF2	604912	Intellectual developmental disorder, autosomal recessive 40, 615599 (3), Autosomal recessive
TAF6	602955	Alazami-Yuan syndrome, 617126 (3), Autosomal recessive
TAFAZZIN	300394	Barth syndrome, 302060 (3), X-linked recessive
TANC2	615047	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906 (3), Autosomal dominant
TANGO2	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TAOK1	610266	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575 (3), Autosomal dominant
TASP1	608270	Suleiman-El-Hattab syndrome, 618950 (3), Autosomal recessive
TAT	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
TBC1D20	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
TBC1D23	617687	Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive
TBC1D24	613577	Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal

		dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive
TBC1D2B	619152	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323 (3), Autosomal recessive
TBC1D7	612655	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 (3), Autosomal recessive
TBCD	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
TBCE	604934	Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive
TBCK	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive
TBL1XR1	608628	Intellectual developmental disorder, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant
TBP	600075	Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant
TBR1	604616	Intellectual developmental disorder with autism and speech delay, 606053 (3), Autosomal dominant
TBX1	602054	Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TCF20	603107	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 (3), Autosomal dominant
TCF4	602272	Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant
TCF7L2	602228	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
TCN2	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
TCTN1	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
TCTN2	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
TDP2	605764	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive

TECPR2	615000	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive
TECR	610057	Intellectual developmental disorder, autosomal recessive 14, 614020 (3), Autosomal recessive
TELO2	611140	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive
TENM3	610083	Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive
TET3	613555	Beck-Fahrner syndrome, 618798 (3), Autosomal dominant, Autosomal recessive
TFAP2A	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
TFE3	314310	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066 (3), X-linked; Renal cell carcinoma, papillary, 1, 300854 (3)
TGDS	616146	Catel-Manzke syndrome, 616145 (3), Autosomal recessive
TGFBR1	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
TGFBR2	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
TGIF1	602630	Holoprosencephaly 4, 142946 (3), Autosomal dominant
TH	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
THOC2	300395	Intellectual developmental disorder, X-linked 12, 300957 (3), X-linked recessive
THOC6	615403	Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive
THRA	190120	Hypothyroidism, congenital, nongoitrous, 6, 614450 (3), Autosomal dominant
THRB	190160	Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant
TIAM1	600687	Neurodevelopmental disorder with language delay and seizures, 619908 (3), Autosomal recessive
TIMM50	607381	3-methylglutaconic aciduria, type IX, 617698 (3), Autosomal recessive
TIMM8A	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TINF2	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TKT	606781	Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive
TLK2	608439	Intellectual developmental disorder, autosomal dominant 57, 618050 (3), Autosomal dominant
TMCO1	614123	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980 (3), Autosomal recessive
TMEM106B	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant

TMEM107	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
TMEM138	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
TMEM165	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
TMEM216	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM222	619469	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470 (3), Autosomal recessive
TMEM231	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TMEM240	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
TMEM63A	618685	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 (3), Autosomal dominant
TMEM67	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
TMEM70	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
TMEM94	618163	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 (3), Autosomal recessive
TMLHE	300777	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
TMTC3	617218	Lissencephaly 8, 617255 (3), Autosomal recessive
TMX2	616715	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive
TNIK	610005	Intellectual developmental disorder, autosomal recessive 54, 617028 (3), Autosomal recessive
TNK2	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
TNPO2	603002	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556 (3), Autosomal dominant
TNR	601995	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653 (3), Autosomal recessive
TNRC6B	610740	Global developmental delay with speech and behavioral abnormalities, 619243 (3), Autosomal dominant
TOE1	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive

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

TRIO	601893	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825 (3), Autosomal dominant
TRIP12	604506	Intellectual developmental disorder, autosomal dominant 49, 617752 (3), Autosomal dominant
TRIP4	604501	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive
TRIT1	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
TRMT1	611669	Intellectual developmental disorder, autosomal recessive 68, 618302 (3), Autosomal recessive
TRMT10A	616013	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
TRMT10C	615423	Combined oxidative phosphorylation deficiency 30, 616974 (3), Autosomal recessive
TRNT1	612907	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive
TRPM3	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
TRPM6	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
TRRAP	603015	?Deafness, autosomal dominant 75, 618778 (3), Autosomal dominant; Developmental delay with or without dysmorphic facies and autism, 618454 (3), Autosomal dominant
TSC1	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3)
TSC2	191092	Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
TSEN15	608756	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
TSEN2	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive
TSEN34	608754	?Pontocerebellar hypoplasia type 2C, 612390 (3), Autosomal recessive
TSEN54	608755	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
TSFM	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
TSHB	188540	Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
TSPAN7	300096	Intellectual developmental disorder, X-linked 58, 300210 (3), X-linked recessive
TTC19	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive

TTC21B	612014	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal dominant, Autosomal recessive
TTC5	619014	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244 (3), Autosomal recessive
TTC8	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TTI1	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
TTI2	614426	Intellectual developmental disorder, autosomal recessive 39, 615541 (3), Autosomal recessive
TUBA1A	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
TUBB	191130	Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant
TUBB2A	615101	Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant
TUBB2B	612850	Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant
TUBB3	602661	Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant
TUBB4A	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
TUBG1	191135	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
TUBGCP2	617817	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 (3), Autosomal recessive
TUBGCP4	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
TUBGCP6	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
TUFM	602389	Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive
TUSC3	601385	Intellectual developmental disorder, autosomal recessive 7, 611093 (3), Autosomal recessive
TWIST1	601622	Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant
TWNK	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive

U2AF2	191318	No OMIM phenotype 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
UBA5	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive
UBE2A	312180	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 (3), X-linked recessive
UBE3A	601623	Angelman syndrome, 105830 (3), Autosomal dominant
UBE3B	608047	Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive
UBE4A	603753	Neurodevelopmental disorder with hypotonia and gross motor and seech delay, 619639 (3), Autosomal recessive
UBR1	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
UBR7	613816	Li-Campeau syndrome, 619189 (3), Autosomal recessive
UBTF	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
UFC1	610554	Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive
UFM1	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
UFSP2	611482	?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant; ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant
UGDH	603370	Developmental and epileptic encephalopathy 84, 618792 (3), Autosomal recessive
UGP2	191760	Developmental and epileptic encephalopathy 83, 618744 (3), Autosomal recessive
UMPS	613891	Orotic aciduria, 258900 (3), Autosomal recessive
UNC80	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive
UPB1	606673	Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive
UPF3B	300298	Intellectual developmental disorder, X-linked syndromic 14, 300676 (3), X-linked recessive
UQCRQ	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
UROC1	613012	?Urocanase deficiency, 276880 (3), Autosomal recessive
USP18	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
USP27X	300975	Intellectual developmental disorder, X-linked 105, 300984 (3), X-linked recessive
USP7	602519	Hao-Fountain syndrome, 616863 (3), Autosomal dominant
USP9X	300072	Intellectual developmental disorder, X-linked 99, 300919 (3), X-linked recessive; Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant

VAMP1	185880	Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
VAMP2	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant
VARS1	192150	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 (3), Autosomal recessive
VLDLR	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
VPS11	608549	?Dystonia 32, 619637 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
VPS13B	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS16	608550	Dystonia 30, 619291 (3), Autosomal dominant
VPS37A	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
VPS41	605485	Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive
VPS4A	609982	CIMDAG syndrome, 619273 (3), Autosomal dominant
VPS53	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
VRK1	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
WAC	615049	Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant
WARS2	604733	Parkinsonism-dystonia 3, childhood-onset, 619738 (3), Autosomal recessive; Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive
WASF1	605035	Neurodevelopmental disorder with absent language and variable seizures, 618707 (3), Autosomal dominant
WASHC4	615748	Intellectual developmental disorder, autosomal recessive 43, 615817 (3), Autosomal recessive
WASHC5	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
WDFY3	617485	?Microcephaly 18, primary, autosomal dominant, 617520 (3), Autosomal dominant
WDR19	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive
WDR26	617424	Skraban-Deardorff syndrome, 617616 (3), Autosomal dominant
WDR37	618586	Neurooculocardiogenitourinary syndrome, 618652 (3), Autosomal dominant

WDR4	605924	Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive
WDR45	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
WDR45B	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive
WDR5	609012	No OMIM phenotype ESHG2021 - A clustering of missense variants in the crucial chromatin modifier WDR5 defines a new neurodevelopmental disorder - Snijders Blok (Nijmegen) - Autosomal dominant
WDR62	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
WDR73	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
WDR81	614218	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive
WFS1	606201	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive
WNT1	164820	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3); Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive
WWOX	605131	Esophageal squamous cell carcinoma, somatic, 133239 (3); Developmental and epileptic encephalopathy 28, 616211 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
XK	314850	McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked
XPA	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XPNPEP3	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
XRCC4	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
XYLT1	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
XYLT2	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
YAP1	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 (3), Autosomal dominant
YIF1B	619109	Kaya-Barakat-Masson syndrome, 619125 (3), Autosomal recessive

YIPF5	611483	Microcephaly, epilepsy, and diabetes syndrome 2, 619278 (3), Autosomal recessive
YME1L1	607472	?Optic atrophy 11, 617302 (3), Autosomal recessive
YWHAG	605356	Developmental and epileptic encephalopathy 56, 617665 (3), Autosomal dominant
YY1	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant
ZBTB11	618181	Intellectual developmental disorder, autosomal recessive 69, 618383 (3), Autosomal recessive
ZBTB16	176797	Leukemia, acute promyelocytic, PL2F/RARA type (3)
ZBTB18	608433	Intellectual developmental disorder, autosomal dominant 22, 612337 (3), Autosomal dominant
ZBTB20	606025	Primrose syndrome, 259050 (3), Autosomal dominant
ZBTB24	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
ZC3H14	613279	Intellectual developmental disorder, autosomal recessive 56, 617125 (3), Autosomal recessive
ZC4H2	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
ZDHHC9	300646	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799 (3), X-linked
ZEB2	605802	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant
ZFHX3	104155	Prostate cancer, somatic, 176807 (3)
ZFHX4	606940	?Ptosis, congenital, 178300 (2), Autosomal dominant
ZFYVE26	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
ZIC1	600470	?Craniosynostosis 6, 616602 (3), Autosomal dominant; Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 (3), Autosomal dominant
ZIC2	603073	Holoprosencephaly 5, 609637 (3), Autosomal dominant
ZMIZ1	607159	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 (3), Autosomal dominant
ZMYM2	602221	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522 (3), Autosomal dominant
ZMYND11	608668	Intellectual developmental disorder, autosomal dominant 30, 616083 (3), Autosomal dominant
ZNF142	604083	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 (3), Autosomal recessive
ZNF148	601897	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 (3), Autosomal dominant
ZNF292	616213	Intellectual developmental disorder, autosomal dominant 64, 619188 (3), Autosomal dominant
ZNF335	610827	Microcephaly 10, primary, autosomal recessive, 615095 (3), Autosomal recessive
ZNF407	615894	SIMHA syndrome, 619557 (3), Autosomal recessive

ZNF423	604557	Nephronophthisis 14, 614844 (3), Autosomal dominant, Autosomal recessive; Joubert syndrome 19, 614844 (3), Autosomal dominant, Autosomal recessive
ZNF462	617371	Weiss-Kruszka syndrome, 618619 (3), Autosomal dominant
ZNF526	614387	Dentici-Novelli neurodevelopmental syndrome, 619877 (3), Autosomal recessive
ZNF699	609571	DEGCAGS syndrome, 619488 (3), Autosomal recessive
ZNF711	314990	Intellectual developmental disorder, X-linked 97, 300803 (3), X-linked
ZSWIM6	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.