ID en epilepsie panel

versie	V3 (1292 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 (AARS)	601065	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 29, 616339 (3), Autosomal recessive
AASS	605113	Hyperlysinemia, 238700 (3), Autosomal recessive; Saccharopinuria, 268700 (1), Autosomal recessive
ABAT	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
ABCC8	600509	Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 606176 (3), Autosomal recessive, Autosomal dominant; Diabetes mellitus, transient neonatal 2, 610374 (3); Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal recessive, Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant
ABCC9	601439	Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Cardiomyopathy, dilated, 1O, 608569 (3); Hypertrichotic osteochondrodysplasia, 239850 (3), Autosomal dominant
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
ABHD5	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ACAD9	611103	Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive
ACADS	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive
ACAT1	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
ACO2	100850	Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive; ?Optic atrophy 9, 616289 (3), Autosomal recessive
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
ACSF3	614245	Combined malonic and methylmalonic aciduria, 614265 (3)

ACSL4	300157	Mental retardation, 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	Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant; Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant
ACVR1	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ACY1	104620	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
ADAR	146920	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant
ADAT3	615302	Mental retardation, autosomal recessive 36, 615286 (3), Autosomal recessive
ADGRG1	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
ADK	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
ADNP	611386	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
ADSL	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
AFF2	300806	Mental retardation, X-linked, FRAXE type, 309548 (3), X-linked recessive
AFF4	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
AFG3L2	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
AGA	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AGPAT2	603100	Lipodystrophy, congenital generalized, type 1, 608594 (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; Deafness, X-linked 5, 300614 (3), X-linked recessive
AIMP1	603605	Leukodystrophy, hypomyelinating, 3, 260600 (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	Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive
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	?Congenital disorder of glycosylation, type Is, 300884 (3), X-linked dominant; Epileptic encephalopathy, early infantile, 36, 300884 (3), X-linked dominant
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	Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive; Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive
ALMS1	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ALX1	601527	?Frontonasal dysplasia 3, 613456 (3)
ALX4	605420	{Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive; Parietal foramina 2, 609597 (3), Autosomal dominant
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	Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, 615686 (3), Autosomal recessive

AMT	238310	Glycine encephalopathy, 605899 (3), Autosomal recessive
ANK3	600465	?Mental retardation, 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
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
AP1S1	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
AP1S2	300629	Mental retardation, X-linked syndromic 5, 304340 (3), X-linked recessive
AP3B1	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3B2	602166	Epileptic encephalopathy, early infantile, 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	Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive; Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant
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	?Sotos syndrome 3, 617169 (3), Autosomal recessive
APTX	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
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	Epileptic encephalopathy, early infantile, 8, 300607 (3), X-linked recessive
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	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive; ?Retinitis pigmentosa 55, 613575 (3)

ARMC9	617612	Joubert syndrome 30, 617622 (3), Autosomal recessive
ARSA	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARSL (ARSE)	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ARV1	611647	Epileptic encephalopathy, early infantile, 38, 617020 (3), Autosomal recessive
ARX	300382	Epileptic encephalopathy, early infantile, 1, 308350 (3), X-linked recessive; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Lissencephaly, X-linked 2, 300215 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive; Partington syndrome, 309510 (3), X-linked recessive; Proud syndrome, 300004 (3), X-linked
ASAH1	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive
ASCL1	100790	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; Haddad syndrome, 209880 (3), Autosomal dominant
ASH1L	607999	Mental retardation, 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	Bohring-Opitz syndrome, 605039 (3), Autosomal dominant; Myelodysplastic syndrome, somatic, 614286 (3)
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 recessive, Autosomal dominant
ATCAY	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
ATIC	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
ATL1	606439	Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant; Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant
ATN1	607462	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 (3), Autosomal dominant; Dentatorubral-pallidoluysian atrophy, 125370 (3), Autosomal dominant

ATP1A2	182340	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; CAPOS syndrome, 601338 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant
ATP2A2	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
ATP6AP2	300556	Mental retardation, X-linked, syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive
ATP6V0A2	611716	Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive; Wrinkly skin syndrome, 278250 (3), Autosomal recessive
ATP6V1A	607027	Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Epileptic encephalopathy, infantile or early childhood, 3, 618012 (3), Autosomal dominant
ATP6V1B2	606939	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant; Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant
ATP7A	300011	Menkes disease, 309400 (3), X-linked recessive; Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
ATP8A2	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
ATPAF2	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
ATR	601215	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant; Seckel syndrome 1, 210600 (3), Autosomal recessive
ATRX	300032	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Mental retardation-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
AUH	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
AUTS2	607270	Mental retardation, autosomal dominant 26, 615834 (3), Autosomal dominant
AVPR2	300538	Diabetes insipidus, nephrogenic, 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
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	Joubert syndrome 27, 617120 (3), Autosomal recessive; ?Meckel syndrome 9, 614209 (3), Autosomal recessive
B9D2	611951	Joubert syndrome 34, 614175 (3), Autosomal recessive; ?Meckel syndrome 10, 614175 (3), Autosomal recessive
BAZ2B	605683	No OMIM phenotype Developmental delay, intellectual disability, and autism spectrum disorder (Scott (2020), Hum Mutat), Autosomal dominant
BBS1	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
BBS10	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (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
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 ketoacid dehydrogenase kinase deficiency, 614923 (3)
BCL11A	606557	Dias-Logan syndrome, 617101 (3), Autosomal dominant
BCL11B	606558	Immunodeficiency 49, 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	Bjornstad syndrome, 262000 (3), Autosomal recessive; GRACILE syndrome, 603358 (3), Autosomal recessive; Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive
BLM	604610	Bloom syndrome, 210900 (3), Autosomal recessive
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	Adenocarcinoma of lung, somatic, 211980 (3); Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Colorectal cancer, somatic (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Melanoma, malignant, somatic (3); Nonsmall cell lung cancer, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant
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
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), Autosomal dominant
BRWD3	300553	Mental retardation, X-linked 93, 300659 (3), X-linked recessive
BSCL2	606158	Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant
BTD	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
BUB1B	602860	Colorectal cancer, somatic, 114500 (3); Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive; [Premature chromatid separation trait], 176430 (3), Autosomal dominant
C12orf4	616082	Mental retardation, autosomal recessive 66, 618221 (3), Autosomal recessive
C12orf57	615140	Temtamy syndrome, 218340 (3), Autosomal recessive

C12orf65	613541	Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive; Spastic paraplegia 55, autosomal recessive, 615035 (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	Epileptic encephalopathy, early infantile, 42, 617106 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant
CACNA1C	114205	Brugada syndrome 3, 611875 (3); Long QT syndrome 8, 618447 (3); Timothy syndrome, 601005 (3), Autosomal dominant
CACNA1E	601013	Epileptic encephalopathy, early infantile, 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
CACNA2D1	114204	No OMIM phenotype Brugada syndrome (Burashnikov (2010) Heart Rhythm 7,1872) Short QT syndrome (Templin (2011) Eur Heart J 32,1077), Autosomal dominant Histiocytoid cardiomyopathy (Cataldo (2014) Cardiol Young epub), Autosomal recessive West syndrome (Hino-Fukuyo (2015) Hum Genet 134,649)
CACNA2D2	607082	Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive
CACNG2	602911	?Mental retardation, autosomal dominant 10, 614256 (3), Autosomal dominant
CAD	114010	Epileptic encephalopathy, early infantile, 50, 616457 (3), Autosomal recessive
CAMK2A	114078	Mental retardation, autosomal dominant 53, 617798 (3), Autosomal dominant; ?Mental retardation, autosomal recessive 63, 618095 (3), Autosomal recessive
CAMK2B	607707	Mental retardation, autosomal dominant 54, 617799 (3), Autosomal dominant
CAMTA1	611501	Cerebellar ataxia, nonprogressive, with mental retardation, 614756 (3), Autosomal dominant
CAPN10	605286	{Diabetes mellitus, noninsulin-dependent 1}, 601283 (3) PMID: 25773692 - Autosomal recessive intellectual disability

CARS2	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
CASK	300172	FG syndrome 4, 300422 (3); Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked dominant; Mental retardation, with or without nystagmus, 300422 (3)
CBL	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
CBS	613381	Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive; Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive
CC2D1A	610055	Mental retardation, autosomal recessive 3, 608443 (3), Autosomal recessive
CC2D2A	612013	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (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
CCDC88A	609736	?PEHO syndrome-like, 617507 (3), Autosomal recessive
CCDC88C	611204	Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive; ?Spinocerebellar ataxia 40, 616053 (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
CDH11	600023	Elsahy-Waters syndrome, 211380 (3), Autosomal recessive
CDH15	114019	Mental retardation, autosomal dominant 3, 612580 (3)
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
CDK5RAP2	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
CDK6	603368	?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive

CDK8	603184	{Autism susceptibility 15}, 612100 (3), Autosomal dominant Syndromic Intellectual Disability (Calpena (2019), Am J Hum Genet 104(4)), Autosomal dominant
CDKL5	300203	Epileptic encephalopathy, early infantile, 2, 300672 (3), X-linked dominant
CDKN1C	600856	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; IMAGE syndrome, 614732 (3), Autosomal dominant
CDON	608707	Holoprosencephaly 11, 614226 (3), 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
CEP120	613446	Joubert syndrome 31, 617761 (3), Autosomal recessive; Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (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	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Meckel syndrome 4, 611134 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive
CEP41	610523	Joubert syndrome 15, 614464 (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
CERT1 (COL4A3BP)	604677	Mental retardation, autosomal dominant 34, 616351 (3), Autosomal dominant
CHAMP1	616327	Mental retardation, autosomal dominant 40, 616579 (3), Autosomal dominant
CHD1	602118	Pilarowski-Bjornsson syndrome, 617682 (3), Autosomal dominant
CHD2	602119	Epileptic encephalopathy, childhood-onset, 615369 (3), Autosomal dominant
CHD3	602120	Snijders Blok-Campeau syndrome, 618205 (3), Autosomal dominant
CHD4	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
CHD7	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant

CHD8	610528	{Autism, susceptibility to, 18}, 615032 (3), Autosomal dominant
CHKB	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
CHRNA2	118502	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant
CHRNA4	118504	Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant; {Nicotine addiction, susceptibility to}, 188890 (3)
CHRNB2	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CIC	612082	Mental retardation, autosomal dominant 45, 617600 (3), Autosomal dominant
CIT	605629	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive
CKAP2L	616174	Filippi syndrome, 272440 (3), Autosomal recessive
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
CLDN16	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
CLDN19	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
CLIC2	300138	?Mental retardation, 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, 6, 601780 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive
CLP1	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
CLPB	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive
CLTC	118955	Mental retardation, autosomal dominant 56, 617854 (3), Autosomal dominant
CNKSR2	300724	Mental retardation, 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 recessive, Autosomal dominant
CNOT1	604917	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	Epileptic encephalopathy, early infantile, 60, 617929 (3), Autosomal recessive
CNTN2	190197	?Epilepsy, myoclonic, familial adult, 5, 615400 (3), Autosomal recessive
CNTNAP2	604569	{Autism susceptibility 15}, 612100 (3); Cortical dysplasia-focal epilepsy syndrome, 610042 (3), Autosomal recessive; Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive
COA8 (APOPT1)	616003	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
COASY	609855	Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 12, 618266 (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)
COG6	606977	Congenital disorder of glycosylation, type IIl, 614576 (3), Autosomal recessive; Shaheen syndrome, 615328 (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)
COL4A1	120130	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; ?Retinal arteries, tortuosity of, 180000 (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
COQ2	609825	Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive; {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant
COQ4	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (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	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
COX15	603646	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 (3), Autosomal recessive; Leigh syndrome due to cytochrome c oxidase deficiency, 256000 (3), Autosomal recessive, Mitochondrial
COX6B1	124089	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
CPA6	609562	Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal recessive, Autosomal dominant; Febrile seizures, familial, 11, 614418 (3), Autosomal recessive
CPLANE1 (C5orf42)	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
CPLX1	605032	Epileptic encephalopathy, early infantile, 63, 617976 (3), Autosomal recessive
CPS1	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
CPT2	600650	CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
CRADD	603454	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive
CRBN	609262	Mental retardation, autosomal recessive 2, 607417 (3), Autosomal recessive
CREBBP	600140	Menke-Hennekam syndrome 1, 618332 (3); Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CRLF1	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
CRPPA (ISPD)	614631	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive
CSNK2A1	115440	Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant
CSNK2B	115441	No OMIM phenotype Epilepsy, Intellectual Disability and Developmental Delay (Li (2019), Sci Rep. 9(1)), 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
CTCF	604167	Mental retardation, 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	Colorectal cancer, somatic, 114500 (3); Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); Medulloblastoma, somatic, 155255 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Ovarian cancer, somatic, 167000 (3); Pilomatricoma, somatic, 132600 (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
CUBN	602997	Megaloblastic anemia-1, Finnish type, 261100 (3), Autosomal recessive
CUL4B	300304	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 (3), X-linked recessive
CUX1	116896	Global developmental delay with or without impaired intellectual development, 618330 (3), Autosomal dominant
CUX2	610648	Epileptic encephalopathy, early infantile, 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
CXorf56	301012	?Mental retardation, X-linked 107, 301013 (3), X-linked
CYB5R3	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
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

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	Colorectal cancer, somatic, 114500 (3); Esophageal carcinoma, somatic, 133239 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive; Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant
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	Lissencephaly, X-linked, 300067 (3), X-linked; Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked
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
DDX3X	300160	Mental retardation, X-linked 102, 300958 (3), X-linked recessive, X-linked dominant
DDX59	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DEAF1	602635	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 (3), Autosomal recessive; Mental retardation, autosomal dominant 24, 615828 (3), Autosomal dominant
DENND5A	617278	Epileptic encephalopathy, early infantile, 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
DHFR	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
DHTKD1	614984	2-aminoadipic 2-oxoadipic aciduria, 204750 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant
DHX30	616423	Neurodevelopmental disorder with severe motor impairment and absent language, 617804 (3), Autosomal dominant
DIAPH1	602121	Deafness, autosomal dominant 1, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
DIP2B	611379	Mental retardation, FRA12A type, 136630 (3), Autosomal dominant

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	Mental retardation, X-linked 90, 300850 (3), X-linked recessive
DLG4	602887	No OMIM phenotype Autism spectrum disorder (An (2014) Transl Psychiatry 4,e394), Autosomal dominant Intellectual disability with marfanoid features (Moutton (2018), Clin Genet. 93(6)), 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
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, Parry type, 162350 (3), Autosomal dominant
DNM1	602377	Epileptic encephalopathy, early infantile, 31, 616346 (3), Autosomal dominant
DNMT3A	602769	Acute myeloid leukemia, somatic, 601626 (3); Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant
DNMT3B	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive
DOCK6	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DOCK7	615730	Epileptic encephalopathy, early infantile, 23, 615859 (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	Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (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	Mental retardation, 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
DYM	607461	Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive; Smith-McCort dysplasia, 607326 (3), Autosomal recessive
DYNC1H1	600112	Charcot-Marie-Tooth disease, axonal, type 20, 614228 (3), Autosomal dominant; Mental retardation, autosomal dominant 13, 614563 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant
DYRK1A	600855	Mental retardation, autosomal dominant 7, 614104 (3), Autosomal dominant
EBF3	607407	Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant
EBP	300205	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
ECHS1	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
EDC3	609842	?Mental retardation, autosomal recessive 50, 616460 (3), Autosomal recessive
EED	605984	Cohen-Gibson syndrome, 617561 (3), Autosomal dominant
EEF1A2	602959	Epileptic encephalopathy, early infantile, 33, 616409 (3), Autosomal dominant; Mental retardation, autosomal dominant 38, 616393 (3), Autosomal dominant
EFTUD2	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
EGF	131530	Hypomagnesemia 4, renal, 611718 (3)
EHMT1	607001	Kleefstra syndrome 1, 610253 (3), Autosomal dominant
EIF2AK3	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
EIF2S3	300161	MEHMO syndrome, 300148 (3), X-linked recessive
EIF4A3	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
EIF4G1	600495	{Parkinson disease 18}, 614251 (3), Autosomal dominant
ELAC2	605367	Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive; {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3)
ELOVL4	605512	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive; Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant
ELP2	616054	Mental retardation, autosomal recessive 58, 617270 (3), Autosomal recessive

EMC1	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (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	Colorectal cancer, somatic, 114500 (3); Menke-Hennekam syndrome 2, 618333 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
EPB41L1	602879	?Mental retardation, autosomal dominant 11, 614257 (3), Autosomal dominant
EPG5	615068	Vici syndrome, 242840 (3), Autosomal recessive
EPM2A	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
EPRS1 (EPRS)	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
ERCC3	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC5	133530	Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
ERCC6	609413	Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive
ERCC8	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
ERLIN2	611605	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
ESCO2	609353	Roberts syndrome, 268300 (3), Autosomal recessive; SC phocomelia syndrome, 269000 (3), Autosomal recessive
ETFB	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
ETHE1	608451	Ethylmalonic encephalopathy, 602473 (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
EXOSC9	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
EZH2	601573	Weaver syndrome, 277590 (3), Autosomal dominant
FA2H	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FAM126A	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal 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
FARS2	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
FARSB	609690	Rajab interstitial lung disease with brain calcifications, 613658 (3), Autosomal recessive
FAT4	612411	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive; Van Maldergem syndrome 2, 615546 (3), Autosomal recessive
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
FBXO31	609102	?Mental retardation, autosomal recessive 45, 615979 (3), Autosomal recessive
FGD1	300546	Aarskog-Scott syndrome, 305400 (3), X-linked recessive; Mental retardation, X-linked syndromic 16, 305400 (3), X-linked recessive
FGF12	601513	Epileptic encephalopathy, early infantile, 47, 617166 (3), Autosomal dominant
FGF14	601515	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant
FGFR1	136350	Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3); Hartsfield syndrome, 615465 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant

FGFR2	176943	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Crouzon syndrome, 123500 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3)
FGFR3	134934	Achondroplasia, 100800 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Muenke syndrome, 602849 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); SADDAN, 616482 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant
FH	136850	Fumarase deficiency, 606812 (3), Autosomal recessive; Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant
FIBP	608296	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive
FKRP	606596	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive; 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
FKTN	607440	Cardiomyopathy, dilated, 1X, 611615 (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; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive

FLNA	300017	Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; Congenital short bowel syndrome, 300048 (3), X-linked recessive; ?FG syndrome 2, 300321 (3), X-linked; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant
FLVCR1	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FMN2	606373	Mental retardation, autosomal recessive 47, 616193 (3), Autosomal recessive
FMR1	309550	Fragile X syndrome, 300624 (3), X-linked dominant; Fragile X tremor/ataxia syndrome, 300623 (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	Mental retardation with language impairment and 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	Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive; Fraser syndrome 2, 617666 (3), Autosomal recessive
FRMD4A	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
FRMPD4	300838	Mental retardation, X-linked 104, 300983 (3), X-linked
FRRS1L	604574	Epileptic encephalopathy, early infantile, 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	Mental retardation, X-linked 9/44, 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

GABBR2	607340	Epileptic encephalopathy, early infantile, 59, 617904 (3), Autosomal dominant; Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant; {Nicotine dependence, protection against}, 188890 (3); {Nicotine dependence, susceptibility to}, 188890 (3)
GABRA1	137160	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3); {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Epileptic encephalopathy, early infantile, 19, 615744 (3), Autosomal dominant
GABRA2	137140	{Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial; Epileptic encephalopathy, early infantile, 78, 618557 (3), Autosomal dominant
GABRA3	305660	No OMIM phenotype Epileptic Encephalopathy (Bamborschke (2019), Neuropediatrics 50(6)), Autosomal dominant
GABRB1	137190	Epileptic encephalopathy, early infantile, 45, 617153 (3), Autosomal dominant
GABRB3	137192	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Epileptic encephalopathy, early infantile, 43, 617113 (3), Autosomal dominant
GABRG2	137164	Epilepsy, generalized, with febrile seizures plus, type 3, 607681 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 74, 618396 (3), Autosomal dominant; Febrile seizures, familial, 8, 607681 (3), Autosomal dominant
GAD1	605363	?Cerebral palsy, spastic quadriplegic, 1, 603513 (3), Autosomal recessive
GALE	606953	Galactose epimerase deficiency, 230350 (3), Autosomal recessive
GALT	606999	Galactosemia, 230400 (3), Autosomal recessive
GAMT	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
GATAD2B	614998	Mental retardation, autosomal dominant 18, 615074 (3), Autosomal dominant
GATM	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive
GCDH	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GCH1	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GCK	138079	Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 606176 (3), Autosomal recessive, Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; MODY, type II, 125851 (3), Autosomal dominant
GCSH	238330	?Glycine encephalopathy, 605899 (3), Autosomal recessive
GDI1	300104	Mental retardation, X-linked 41, 300849 (3), X-linked dominant

GFAP	137780	Alexander disease, 203450 (3), Autosomal dominant
GFM1	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
GFM2	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
GJA1	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
GJC2	608803	Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive; Lymphatic malformation 3, 613480 (3), Autosomal dominant; Spastic paraplegia 44, autosomal recessive, 613206 (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 II, 230600 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (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; {Hypothalamic hamartomas, somatic}, 241800 (3); Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GLRA1	138491	Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant
GLRB	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
GLUD1	138130	Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant
GLYCTK	610516	D-glyceric aciduria, 220120 (3), Autosomal recessive
GM2A	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
GMPPA	615495	Alacrima, achalasia, and mental retardation syndrome, 615510 (3), Autosomal recessive

GMPPB	615320	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive
GNAO1	139311	Epileptic encephalopathy, early infantile, 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
GNAS	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
GNB1	139380	Leukemia, acute lymphoblastic, somatic, 613065 (3); Mental retardation, autosomal dominant 42, 616973 (3), Autosomal dominant
GNB5	604447	Intellectual developmental disorder with cardiac arrhythmia, 617173 (3), Autosomal recessive; Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 (3), Autosomal recessive
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
GNPTAB	607840	Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive; Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive
GNS	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
GOSR2	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
GPAA1	603048	Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
GPHN	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
GPT2	138210	Mental retardation, autosomal recessive 49, 616281 (3), Autosomal recessive
GRIA2	138247	No OMIM phenotype Intellectual disability and neurodevelopmental abnormalities including autism spectrum disorder, Rett syndrome-like features, and seizures or developmental epileptic encephalopathy (Salpietro (2019), Nat Commun 10(1)), Autosomal dominant
GRIA3	305915	Mental retardation, X-linked 94, 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	Mental retardation, autosomal recessive, 6, 611092 (3), Autosomal recessive
GRIN1	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive
GRIN2A	138253	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570 (3), Autosomal dominant
GRIN2B	138252	Epileptic encephalopathy, early infantile, 27, 616139 (3), Autosomal dominant; Mental retardation, autosomal dominant 6, 613970 (3), Autosomal dominant
GRIN2D	602717	Epileptic encephalopathy, early infantile, 46, 617162 (3), Autosomal dominant
GRIP1	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
GRM1	604473	Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive
GRN	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
GSS	601002	Glutathione synthetase deficiency, 266130 (3), Autosomal recessive; Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive
GTF2H5	608780	Trichothiodystrophy 3, photosensitive, 616395 (3)
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 (HIST1H1E)	142220	Rahman syndrome, 617537 (3), Autosomal dominant
H4C3 (HIST1H4C)	602827	No OMIM phenotype Growth delay, microcephaly and intellectual disability (Tessadori (2019), Eur J Hum Genet), Autosomal dominant
HACE1	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 4, 609975 (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	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541 (3), X-linked recessive
HCN1	602780	Epileptic encephalopathy, early infantile, 24, 615871 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 10, 618482 (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	Mental retardation, 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	Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant; Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant
HEXA	606869	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 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
HIVEP2	143054	Mental retardation, autosomal dominant 43, 616977 (3), Autosomal dominant
HLCS	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
HMGCL	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive

HNMT	605238	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; Mental retardation, autosomal recessive 51, 616739 (3), Autosomal recessive
HNRNPH2	300610	Mental retardation, X-linked, syndromic, Bain type, 300986 (3), X-linked dominant
HNRNPK	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
HNRNPU	602869	Epileptic encephalopathy, early infantile, 54, 617391 (3), Autosomal dominant
HOXA1	142955	Athabaskan brainstem dysgenesis syndrome, 601536 (3); Bosley-Salih-Alorainy syndrome, 601536 (3)
HPD	609695	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
HPRT1	308000	HPRT-related gout, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
HRAS	190020	Bladder cancer, somatic, 109800 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Costello syndrome, 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); Thyroid carcinoma, follicular, somatic, 188470 (3)
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	Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant; Even-plus syndrome, 616854 (3), Autosomal recessive
HSPD1	118190	Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive; Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant
HTRA2	606441	3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive; {Parkinson disease 13}, 610297 (3)
HUWE1	300697	Mental retardation, X-linked syndromic, Turner type, 309590 (3), X-linked
IARS1 (IARS)	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
IDH2	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
IDS	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive

IDUA	252800	Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive
IER3IP1	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
IFIH1	606951	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; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (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 recessive, Autosomal dominant
IKBKG	300248	Ectodermal dysplasia and immunodeficiency 1, 300291 (3); Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3); Immunodeficiency 33, 300636 (3), X-linked recessive; Immunodeficiency, isolated, 300584 (3); Incontinentia pigmenti, 308300 (3), X-linked dominant; Invasive pneumococcal disease, recurrent isolated, 2, 300640 (3)
IL1RAPL1	300206	Mental retardation, X-linked 21/34, 300143 (3), X-linked recessive
IMPA1	602064	Mental retardation, autosomal recessive 59, 617323 (3), Autosomal recessive
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
IQSEC2	300522	Mental retardation, X-linked 1/78, 309530 (3), X-linked dominant
IRF2BPL	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
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	Epileptic encephalopathy, early infantile, 35, 616647 (3), Autosomal recessive; [Inosine triphosphatase deficiency], 613850 (3)
ITPR1	147265	Gillespie syndrome, 206700 (3), Autosomal recessive, Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant
IVD	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive

JAG1	601920	Alagille syndrome 1, 118450 (3), Autosomal dominant; ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
JMJD1C	604503	No OMIM phenotype ?Rett syndrome (Saez (2016) Genet Med 18,378), Autosomal dominant ?Congenital heart disease in 22q11.2 deletion syndrome patients (Guo (2015) Am J Hum Genet 97,869), Autosomal dominant ?Autism spectrum disorder (Saez (2016) Genet Med 18,378), Autosomal dominant ?Intellectual disability (Saez (2016) Genet Med 18,378), Autosomal dominant
KANK1	607704	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
KANSL1	612452	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant
KAT6A	601408	Mental retardation, autosomal dominant 32, 616268 (3), Autosomal dominant
KAT6B	605880	Genitopatellar syndrome, 606170 (3), Autosomal dominant; SBBYSS syndrome, 603736 (3), Autosomal dominant
KATNB1	602703	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
KCNA1	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNA2	176262	Epileptic encephalopathy, early infantile, 32, 616366 (3), Autosomal dominant
KCNA4	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
KCNB1	600397	Epileptic encephalopathy, early infantile, 26, 616056 (3), Autosomal dominant
KCNC1	176258	Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant
KCNC3	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
KCNH1	603305	Temple-Baraitser syndrome, 611816 (3), Autosomal dominant; Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant
KCNJ10	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
KCNJ11	600937	Diabetes mellitus, transient neonatal, 3, 610582 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Diabetes, permanent neonatal, with or without neurologic features, 606176 (3), Autosomal recessive, Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant

KCNJ6	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
KCNK9	605874	Birk-Barel mental retardation dysmorphism syndrome, 612292 (3)
KCNMA1	600150	Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3); Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant
KCNQ2	602235	Epileptic encephalopathy, early infantile, 7, 613720 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant
KCNQ3	602232	Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant
KCNQ5	607357	Mental retardation, autosomal dominant 46, 617601 (3), Autosomal dominant
KCNT1	608167	Epilepsy, nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 14, 614959 (3), 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
KDM5B	605393	Mental retardation, autosomal recessive 65, 618109 (3), Autosomal recessive
KDM5C	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
KDM6A	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KIAA0556	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
KIAA0586	610178	Joubert syndrome 23, 616490 (3), Autosomal recessive; Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive
KIAA1109	611565	Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive
KIDINS220	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant
KIF11	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
KIF14	611279	?Meckel syndrome 12, 616258 (3), Autosomal recessive; Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive
KIF1A	601255	Mental retardation, autosomal dominant 9, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal recessive
KIF1BP	609367	Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive

KIF2A	602591	Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant
KIF4A	300521	?Mental retardation, X-linked 100, 300923 (3), X-linked recessive
KIF5C	604593	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant
KIF7	611254	Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; Joubert syndrome 12, 200990 (3), Autosomal recessive
KIRREL3	607761	Mental retardation, autosomal dominant 4, 612581 (3)
KLF7	604865	No OMIM phenotype ? Developmental delay/intellectual disability, neuromuscular and psychiatric symptoms (Powis (2018), Clin Genet 93(5)), Autosomal dominant
KLHL15	300980	Mental retardation, X-linked 103, 300982 (3), X-linked recessive
KMT2A	159555	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 (2), Autosomal dominant; Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
KMT2B	606834	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 recessive
KMT5B	610881	Mental retardation, autosomal dominant 51, 617788 (3), Autosomal dominant
KNL1	609173	Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive
KPTN	615620	Mental retardation, autosomal recessive 41, 615637 (3), Autosomal recessive
KRAS	190070	Arteriovenous malformation of the brain, somatic, 108010 (3); Bladder cancer, somatic, 109800 (3); Breast cancer, somatic, 114480 (3); Cardiofaciocutaneous syndrome 2, 615278 (3); Gastric cancer, somatic, 137215 (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation; Lung cancer, somatic, 211980 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; Oculoectodermal syndrome, somatic, 600268 (3); Pancreatic carcinoma, somatic, 260350 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)
L1CAM	308840	CRASH syndrome, 303350 (3), X-linked recessive; Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive; Hydrocephalus with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3), X-linked recessive; MASA syndrome, 303350 (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, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (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 brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive
LARP7	612026	Alazami syndrome, 615071 (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, 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	Mental retardation, autosomal recessive 64, 618103 (3), Autosomal recessive
LINS1	610350	Mental retardation, autosomal recessive 27, 614340 (3), Autosomal recessive
LMAN2L	609552	?Mental retardation, autosomal recessive, 52, 616887 (3), Autosomal recessive
LONP1	605490	CODAS syndrome, 600373 (3), Autosomal recessive
LRP2	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRPPRC	607544	Leigh syndrome, French-Canadian type, 220111 (3), Autosomal recessive
LZTFL1	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
LZTR1	600574	Noonan syndrome 10, 616564 (3), Autosomal dominant; Noonan syndrome 2, 605275 (3), Autosomal recessive; {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 recessive, Autosomal dominant
MACF1	608271	Lissencephaly 9 with complex brainstem malformation, 618325 (3), Autosomal dominant

MAF	177075	Ayme-Gripp syndrome, 601088 (3), Autosomal dominant; Cataract 21, multiple types, 610202 (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	Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive; Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3)
MAN1B1	604346	Mental retardation, autosomal recessive 15, 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	No OMIM phenotype Intellectual disability and extensive white matter deficit (Walters (2018), Nat Commun 9(1)), Autosomal dominant
MAP2K1	176872	Cardiofaciocutaneous syndrome 3, 615279 (3)
MAP2K2	601263	Cardiofaciocutaneous syndrome 4, 615280 (3)
MAPK8IP3	605431	Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant
MAPRE2	605789	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant
MASP1	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
MAT1A	610550	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal recessive, Autosomal dominant; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal recessive, Autosomal dominant
MBD5	611472	Mental retardation, autosomal dominant 1, 156200 (3), Autosomal dominant
MBOAT7	606048	Mental retardation, autosomal recessive 57, 617188 (3), Autosomal recessive
MBTPS2	300294	IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (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

MCOLN1	605248	Mucolipidosis IV, 252650 (3), Autosomal recessive
MCPH1	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
MDH2	154100	Epileptic encephalopathy, early infantile, 51, 617339 (3), Autosomal recessive
MECP2	300005	{Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Mental retardation, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; Mental retardation, X-linked, syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, atypical, 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; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive
MED13	603808	Intellectual developmental disorder 61, 618009 (3), Autosomal dominant
MED13L	608771	Mental retardation and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant; Transposition of the great arteries, dextro-looped 1, 608808 (3), Autosomal dominant
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive
MED23	605042	Mental retardation, autosomal recessive 18, 614249 (3), Autosomal recessive
MED25	610197	Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive
MEF2C	600662	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 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	Mental retardation, autosomal recessive 44, 615942 (3), Autosomal recessive
MFF	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive
MFSD2A	614397	Microcephaly 15, primary, autosomal recessive, 616486 (3), Autosomal recessive
MFSD8	611124	Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive; Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive
MGAT2	602616	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive

MGP	154870	Keutel syndrome, 245150 (3), Autosomal recessive
MICU1	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
MID1	300552	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
MID2	300204	?Mental retardation, X-linked 101, 300928 (3), X-linked recessive
MKKS	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
MKS1	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (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, 251100 (3), Autosomal recessive
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110 (3), Autosomal recessive
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MMADHC	611935	Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive
MMUT (MUT)	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
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
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
MRPL3	607118	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
MRPS22	605810	Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive; Ovarian dysgenesis 7, 618117 (3), Autosomal recessive
MSL3	300609	Mental retardation, X-linked, syndromic, 36, 301032 (3)
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	Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Vascular disease, susceptibility to} (3)
MTOR	601231	Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant
MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (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; Mevalonic aciduria, 610377 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant
MYCN	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant
MYH9	160775	Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant; Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 (3), Autosomal dominant
MYO5A	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
MYT1L	613084	Mental retardation, 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	Mental retardation, autosomal dominant 50, 617787 (3), Autosomal dominant
NACC1	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant
NAGA	104170	Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type I, 609241 (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
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
NARS2	612803	Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive; ?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive
NBEA	604889	No OMIM phenotype Neurodevelopmental disorder (Mulhern (2018), Ann Neurol 84(5)), Autosomal dominant
NBN	602667	Aplastic anemia, 609135 (3); Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NDE1	609449	Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive; ?Microhydranencephaly, 605013 (3), Autosomal recessive
NDP	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3); Norrie disease, 310600 (3), X-linked recessive
NDST1	600853	Mental retardation, autosomal recessive 46, 616116 (3), Autosomal recessive
NDUFA1	300078	Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked 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
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
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	?Epileptic encephalopathy, early infantile, 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
NEU1	608272	Sialidosis, type I, 256550 (3), Autosomal recessive; Sialidosis, type II, 256550 (3), Autosomal recessive
NEXMIF	300524	Mental retardation, X-linked 98, 300912 (3), X-linked dominant
NF1	613113	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; Watson syndrome, 193520 (3), Autosomal dominant
NFIA	600727	Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant
NFIX	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Sotos syndrome 2, 614753 (3), Autosomal dominant
NGLY1	610661	Congenital disorder of deglycosylation, 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
NKX2-1	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant
NLGN3	300336	{Asperger syndrome susceptibility, X-linked 1}, 300494 (3), Isolated cases, X-linked, Multifactorial; {Autism susceptibility, X-linked 1}, 300425 (3), X-linked
NLGN4X	300427	{Asperger syndrome susceptibility, X-linked 2}, 300497 (3), X-linked; {Autism susceptibility, X-linked 2}, 300495 (3), Isolated cases, X-linked, Multifactorial; Mental retardation, X-linked, 300495 (3), Isolated cases, X-linked, Multifactorial
NLRP3	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NONO	300084	Mental retardation, X-linked, syndromic 34, 300967 (3), X-linked

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	No OMIM phenotype Intellectual disability and autism spectrum disorder (Lévy (2018), Clin Genet 94(2)), Autosomal dominant
NRAS	164790	Colorectal cancer, somatic, 114500 (3); Epidermal nevus, somatic, 162900 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Neurocutaneous melanosis, somatic, 249400 (3); Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3)
NRXN1	600565	Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3)
NSD1	606681	Leukemia, acute myeloid, 601626 (1), Autosomal dominant, Somatic mutation; Sotos syndrome 1, 117550 (3), Autosomal dominant
NSD2	602952	No OMIM phenotype Intellectual disability and dysmorphic features (Lozier (2018), J Hum genet), Autosoma dominant
NSDHL	300275	CHILD syndrome, 308050 (3), X-linked dominant; CK syndrome, 300831 (3), X-linked recessive
NSUN2	610916	Mental retardation, autosomal recessive 5, 611091 (3), Autosomal recessive
NT5C2	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive; Medullary thyroid carcinoma, familial, 155240 (3), Autosomal dominant
NTRK2	600456	Epileptic encephalopathy, early infantile, 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
NUP62	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive

NUS1	610463	?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive; Mental retardation, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant
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	{Colonic adenoma recurrence, reduced risk of}, 114500 (3), Autosomal dominant, Somatic mutation
OFD1	300170	Joubert syndrome 10, 300804 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
OGT	300255	Mental retardation, X-linked 106, 300997 (3), X-linked recessive
OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 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 recessive
OTUD6B	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive
OTX2	600037	Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant
P4HTM	614584	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 (3), Autosomal recessive
PACS1	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
PACS2	610423	Epileptic encephalopathy, early infantile, 66, 618067 (3), Autosomal dominant
PAFAH1B1	601545	Lissencephaly 1, 607432 (3), Autosomal dominant; Subcortical laminar heterotopia, 607432 (3), Autosomal dominant
PAH	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
PAK3	300142	Mental retardation, X-linked 30/47, 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
PAX1	167411	?Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
PAX6	607108	Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3); Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Optic nerve hypoplasia, 165550 (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
PCDH19	300460	Epileptic encephalopathy, early infantile, 9, 300088 (3), X-linked
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
PDE4D	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant
PDE6D	602676	?Joubert syndrome 22, 615665 (3), Autosomal recessive
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3)
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; MODY, type IV, 606392 (3); Pancreatic agenesis 1, 260370 (3), Autosomal recessive

PEPD	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PET100	614770	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
PEX1	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (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 3A (Zellweger), 614859 (3), Autosomal recessive; Peroxisome biogenesis disorder 3B, 266510 (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 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (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 7A (Zellweger), 614872 (3), Autosomal recessive; Peroxisome biogenesis disorder 7B, 614873 (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 2A (Zellweger), 214110 (3), Autosomal recessive; Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
PEX6	601498	Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant
PEX7	601757	Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
PGAP1	611655	Mental retardation, autosomal recessive 42, 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
PGM3	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
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	Mental retardation syndrome, X-linked, 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	Developmental delay, intellectual disability, obesity, and dysmorphism, 617991 (3), Autosomal dominant
PI4KA	600286	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
PIGA	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
PIGC	601730	Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive
PIGG	616918	Mental retardation, autosomal recessive 53, 616917 (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	?Epileptic encephalopathy, early infantile, 55, 617599 (3), Autosomal recessive
PIGT	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive; ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation
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

PIK3CA	171834	Breast cancer, somatic, 114480 (3); CLAPO syndrome, somatic, 613089 (3); CLOVE syndrome, somatic, 612918 (3); Colorectal cancer, somatic, 114500 (3); Cowden syndrome 5, 615108 (3); Gastric cancer, somatic, 613659 (3); Hepatocellular carcinoma, somatic, 114550 (3); Keratosis, seborrheic, somatic, 182000 (3); Macrodactyly, somatic, 155500 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Nevus, epidermal, somatic, 162900 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Ovarian cancer, somatic, 167000 (3)
PIK3R2	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant
PLA2G6	603604	Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive
PLAA	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive
PLCB1	607120	Epileptic encephalopathy, early infantile, 12, 613722 (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
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	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	Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (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
POLG	174763	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POLR3A	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive; Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive
POLR3B	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive; 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
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 (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive; ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive
POMT1	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive
POMT2	607439	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive

PORCN	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
POU1F1	173110	Pituitary hormone deficiency, combined, 1, 613038 (3), Autosomal recessive, Autosomal dominant
POU3F3	602480	Snijders Blok-Fisher syndrome, 618604 (3), Autosomal dominant
PPM1D	605100	Breast cancer, somatic, 114480 (3); Jansen de Vries syndrome, 617450 (3), Autosomal dominant
PPOX	600923	Porphyria variegata, 176200 (3), Autosomal dominant
PPP1CB	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
PPP1R15B	613257	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
PPP2CA	176915	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354 (3), Autosomal dominant
PPP2R1A	605983	Mental retardation, autosomal dominant 36, 616362 (3), Autosomal dominant
PPP2R5D	601646	Mental retardation, autosomal dominant 35, 616355 (3), Autosomal dominant
PPP3CA	114105	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 (3), Autosomal dominant; Epileptic encephalopathy, infantile or early childhood, 1, 617711 (3), Autosomal dominant
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
PQBP1	300463	Renpenning syndrome, 309500 (3), X-linked recessive
PRF1	170280	Aplastic anemia, 609135 (3); Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Lymphoma, non-Hodgkin, 605027 (3)
PRICKLE1	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
PRKAR1A	188830	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; Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant
PRMT7	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
PRODH	606810	Hyperprolinemia, type I, 239500 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant
PRPS1	311850	Arts syndrome, 301835 (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; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive
PRR12	616633	No OMIM phenotype Global developmental delay, intellectual disability, eye and vision abnormalities, dysmorphic features, and neuropsychiatric problems (Leduc (2018), Hum Genet), Autosomal dominant
PRRT2	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant
PRSS12	606709	Mental retardation, 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; Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive
PSEN1	104311	?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant
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); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Holoprosencephaly 7, 610828 (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	Cowden syndrome 1, 158350 (3), Autosomal dominant; {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3)

PTPN11	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3); Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), 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	Mental retardation, autosomal dominant 31, 616158 (3), Autosomal dominant
PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
PUS3	616283	Mental retardation, autosomal recessive 55, 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 IIB, 612940 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIIB, 614438 (3)
PYCR2	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
QARS1 (QARS)	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
RAB27A	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
RAB39B	300774	Mental retardation, X-linked 72, 300271 (3), X-linked recessive; Waisman syndrome, 311510 (3), X-linked recessive
RAB3GAP1	602536	Warburg micro syndrome 1, 600118 (3), Autosomal recessive
RAB3GAP2	609275	Martsolf syndrome, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive

RAC1	602048	Mental retardation, 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; LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3), Autosomal dominant
RAI1	607642	Smith-Magenis syndrome, 182290 (3), Autosomal dominant, Isolated cases
RALA	179550	No OMIM phenotype Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities (Wagner (2020), Am J Hum Genet 106(2)), Autosomal recessive
RARB	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant
RARS2	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
RBBP8	604124	Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3); Seckel syndrome 2, 606744 (3), Autosomal recessive
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
REV3L	602776	No OMIM phenotype Moebius syndrome (Tomas-Roca (2015) Nat Commun 6), Autosomal dominant Psoriasis,association with} (Strange (2010) Nat Genet 42,985) {Colorectal cancer,increased risk,association with} (Webb (2006) Hum Mol Genet 15,3263), Autosomal dominant

RFT1	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
RHEB	601293	No OMIM phenotype ?Intellectual dsiability (Reijnders 2017 (Nat Commun)), Autosomal dominant
RHOBTB2	607352	Epileptic encephalopathy, early infantile, 64, 618004 (3), Autosomal dominant
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
RMRP	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (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 dominant
RNF125	610432	Tenorio syndrome, 616260 (3), Autosomal dominant
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
RPGRIP1L	610937	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
RPL10	312173	{Autism, susceptibility to, X-linked 5}, 300847 (3); Mental retardation, X-linked, syndromic, 35, 300998 (3), X-linked recessive
RPS19	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
RPS6KA3	300075	Coffin-Lowry syndrome, 303600 (3), X-linked dominant; Mental retardation, X-linked 19, 300844 (3), X-linked dominant
RRM2B	604712	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (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

RTEL1	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant
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	Mental retardation, autosomal recessive 61, 617773 (3), Autosomal recessive
RXYLT1 (TMEM5)	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant; Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant
SAMD9	610456	MIRAGE syndrome, 617053 (3), Autosomal dominant; Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive
SAMHD1	606754	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
SATB2	608148	Glass syndrome, 612313 (3), Autosomal dominant
SBDS	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
SC5D	602286	Lathosterolosis, 607330 (3), Autosomal recessive
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	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant
SCN1B	600235	Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3); Cardiac conduction defect, nonspecific, 612838 (3); Epilepsy, generalized, with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 52, 617350 (3), Autosomal recessive

SCN2A	182390	Epileptic encephalopathy, early infantile, 11, 613721 (3), Autosomal dominant; Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant
SCN3A	182391	Epilepsy, familial focal, with variable foci 4, 617935 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 62, 617938 (3), Autosomal dominant
SCN8A	600702	Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant; ?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant
SCO1	603644	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
SCO2	604272	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 (3), Autosomal recessive; Myopia 6, 608908 (3), Autosomal dominant
SCYL1	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
SDCCAG8	613524	Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive; Senior-Loken syndrome 7, 613615 (3)
SDHA	600857	Cardiomyopathy, dilated, 1GG, 613642 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive; Paragangliomas 5, 614165 (3), Autosomal dominant
SEMA3E	608166	?CHARGE syndrome, 214800 (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
SETBP1	611060	Mental retardation, autosomal dominant 29, 616078 (3), Autosomal dominant; Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant
SETD1A	611052	No OMIM phenotype Schizophrenia (Takata (2014) Neuron 82, 723), Autosomal dominant Schizophrenia and develpmental disorders (Singh (2016) Nat Neurosci), Autosomal dominant
SETD1B	611055	No OMIM phenotype Intellectual disability, autism spectrum disorder and epilepsy with myoclonic absences (Hiraide (2019), Epilepsia Open 4(3)), Autosomal dominant
SETD2	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant
SETD5	615743	Mental retardation, autosomal dominant 23, 615761 (3), Autosomal dominant
SGPL1	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive

SGSH	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
SHANK2	603290	{Autism susceptibility 17}, 613436 (3)
SHANK3	606230	Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3), Autosomal dominant
SHH	600725	Holoprosencephaly 3, 142945 (3), Autosomal dominant; Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant
SHOC2	602775	Noonan-like syndrome with loose anagen hair, 607721 (3), Autosomal dominant
SHROOM4	300579	Stocco dos Santos X-linked mental retardation syndrome, 300434 (3), X-linked
SIK1	605705	Epileptic encephalopathy, early infantile, 30, 616341 (3), Autosomal dominant
SIL1	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SIN3A	607776	Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant
SIX3	603714	Holoprosencephaly 2, 157170 (3), Autosomal dominant; Schizencephaly, 269160 (3)
SKI	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
SLC12A5	606726	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 34, 616645 (3), Autosomal recessive
SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive
SLC13A5	608305	Epileptic encephalopathy, early infantile, 25, 615905 (3), Autosomal recessive
SLC16A1	600682	Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant
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	Epileptic encephalopathy, early infantile, 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	Epileptic encephalopathy, early infantile, 39, 612949 (3), Autosomal recessive
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
SLC25A22	609302	Epileptic encephalopathy, early infantile, 3, 609304 (3), Autosomal recessive
SLC2A1	138140	Dystonia 9, 601042 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant
SLC33A1	603690	Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive; Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant
SLC35A1	605634	Congenital disorder of glycosylation, type IIf, 603585 (3), Autosomal recessive
SLC35A2	314375	Congenital disorder of glycosylation, type IIm, 300896 (3), X-linked dominant, Somatic mosaicism
SLC35A3	605632	?Arthrogryposis, mental retardation, and seizures, 615553 (3), Autosomal recessive
SLC35C1	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC39A14	608736	Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive; ?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant
SLC39A8	608732	Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive
SLC4A4	603345	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive
SLC6A1	137165	Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant
SLC6A17	610299	Mental retardation, autosomal recessive 48, 616269 (3), Autosomal recessive
SLC6A19	608893	Hartnup disorder, 234500 (3), Autosomal recessive; Hyperglycinuria, 138500 (3), Autosomal dominant; Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive
SLC6A3	126455	{Nicotine dependence, protection against}, 188890 (3); Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive
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	Mental retardation, X-linked syndromic, Christianson type, 300243 (3), X-linked dominant
SLC9A7	300368	Intellectual developmental disorder, X-linked 108, 301024 (3), X-linked recessive

SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant; Myhre syndrome, 139210 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant
SMAD6	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
SMARCA2	600014	Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant
SMARCA4	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant
SMARCB1	601607	Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant; Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant
SMARCC2	601734	Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant
SMARCE1	603111	Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant; {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant
SMC1A	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant
SMC3	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
SMG9	613176	Heart and brain malformation syndrome, 616920 (3), Autosomal recessive
SMOC1	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SMPD1	607608	Niemann-Pick disease, type A, 257200 (3), Autosomal recessive; Niemann-Pick disease, type B, 607616 (3), Autosomal recessive
SMS	300105	Mental retardation, X-linked, 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	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501 (3), Autosomal recessive
SNRPN	182279	Prader-Willi syndrome, 176270 (3), Autosomal dominant
SNX14	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), 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	?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant; Noonan syndrome 4, 610733 (3), Autosomal dominant
SOS2	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SOX10	602229	PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
SOX11	600898	Coffin-Siris syndrome 9, 615866 (3), Autosomal dominant
SOX2	184429	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
SOX3	313430	Mental retardation, 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
SPART	607111	Troyer syndrome, 275900 (3), Autosomal recessive
SPAST	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
SPATA5	613940	Epilepsy, hearing loss, and mental retardation syndrome, 616577 (3), Autosomal recessive
SPECC1L	614140	?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant; Hypertelorism, Teebi type, 145420 (3), Autosomal dominant; Opitz GBBB syndrome, type II, 145410 (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
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	Epileptic encephalopathy, early infantile, 5, 613477 (3), Autosomal dominant
SPTBN2	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
SRCAP	611421	Floating-Harbor syndrome, 136140 (3), Autosomal dominant
SRD5A3	611715	Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive; Kahrizi syndrome, 612713 (3), Autosomal recessive
SRPX2	300642	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 (3)
SSR4	300090	Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive

ST3GAL3	606494	?Epileptic encephalopathy, early infantile, 15, 615006 (3), Autosomal recessive; Mental retardation, autosomal recessive 12, 611090 (3), Autosomal recessive
ST3GAL5	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
STAG1	604358	Mental retardation, autosomal dominant 47, 617635 (3), Autosomal dominant
STAMBP	606247	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
STIL	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
STRA6	610745	Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive; Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive
STRADA	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive
STT3A	601134	?Congenital disorder of glycosylation, type Iw, 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	Epileptic encephalopathy, early infantile, 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
SUMF1	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
SUOX	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
SURF1	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial
SUZ12	606245	No OMIM phenotype Weaver-like syndrome (Cyrus (2019), Am J Med Genet C Semin Med Genet 181(4)), Autosomal dominant
SVBP	617853	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive
SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3), X-linked recessive, X-linked dominant
SYNE1	608441	Arthrogryposis multiplex congenita, 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	Mental retardation, autosomal dominant 5, 612621 (3), Autosomal dominant

SYNJ1	604297	Epileptic encephalopathy, early infantile, 53, 617389 (3), Autosomal recessive; Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive
SYP	313475	Mental retardation, 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	Epileptic encephalopathy, early infantile, 18, 615476 (3), Autosomal recessive
TAF1	313650	Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive; Mental retardation, X-linked, syndromic 33, 300966 (3), X-linked recessive
TAF13	600774	Mental retardation, autosomal recessive 60, 617432 (3), Autosomal recessive
TAF2	604912	Mental retardation, autosomal recessive 40, 615599 (3), Autosomal recessive
TAF6	602955	Alazami-Yuan syndrome, 617126 (3), Autosomal recessive
TANC2	615047	No OMIM phenotype Neurodevelopmental syndrome associated with psychiatric disorders (Guo (2019), Nat Commun 10(1)), Autosomal dominant
TANGO2	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (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	DOORS syndrome, 220500 (3), Autosomal recessive; Deafness , autosomal recessive 86, 614617 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 16, 615338 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (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	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive

TBCK	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive
TBL1XR1	608628	Mental retardation, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant
TBP	600075	{Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant
TBR1	604616	Intellectual developmental disorder with autism and speech delay, 606053 (3), Autosomal dominant
TBX1	602054	Conotruncal anomaly face syndrome, 217095 (3); DiGeorge syndrome, 188400 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TCF20	603107	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 (3), Autosomal dominant
TCF4	602272	Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant; Pitt-Hopkins syndrome, 610954 (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	Spastic paraplegia 49, autosomal recessive, 615031 (3), Autosomal recessive
TECR	610057	Mental retardation, autosomal recessive 14, 614020 (3), Autosomal recessive
TELO2	611140	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive
TFAP2A	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
TGDS	616146	Catel-Manzke syndrome, 616145 (3), Autosomal recessive
TGFBR1	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
TGFBR2	190182	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
TGIF1	602630	Holoprosencephaly 4, 142946 (3), Autosomal dominant
TH	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive

THOC2	300395	Mental retardation, X-linked 12/35, 300957 (3), X-linked recessive
THOC6	615403	Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive
THRB	190160	Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant
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	Mental retardation, autosomal dominant 57, 618050 (3), Autosomal dominant
TMCO1	614123	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980 (3), Autosomal recessive
TMEM107	616183	?Joubert syndrome 29, 617562 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; Orofaciodigital syndrome XVI, 617563 (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
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
TMEM67	609884	{Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive
TMEM70	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
TMLHE	300777	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
TMTC3	617218	Lissencephaly 8, 617255 (3), Autosomal recessive
TNIK	610005	Mental retardation, autosomal recessive 54, 617028 (3), Autosomal recessive

TOE1	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive
TP53RK	608679	Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive
TPI1	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (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
TRAPPC11	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
TRAPPC6B	610397	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 (3), Autosomal recessive
TRAPPC9	611966	Mental retardation, autosomal recessive 13, 613192 (3), Autosomal recessive
TREX1	606609	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant
TRIM32	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TRIO	601893	Mental retardation, autosomal dominant 44, 617061 (3), Autosomal dominant
TRIP12	604506	Mental retardation, 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	Mental retardation, autosomal recessive 68, 618302 (3), Autosomal recessive
TRMT10A	616013	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
TRNT1	612907	Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive; Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive
TRPM6	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive

TRRAP	603015	Developmental delay with or without dysmorphic facies and autism, 618454 (3), Autosomal dominant
TSC1	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, 606690 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant
TSC2	191092	?Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, somatic, 606690 (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
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	Mental retardation, X-linked 58, 300210 (3), X-linked recessive
TTC19	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
TTC21B	612014	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
TTC37	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
TTC8	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TTI2	614426	Mental retardation, autosomal recessive 39, 615541 (3), Autosomal recessive
TUBA1A	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
TUBA8	605742	Cortical dysplasia, complex, with other brain malformations 8, 613180 (3), Autosomal recessive
TUBB	191130	Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant; Symmetric circumferential skin creases, congenital, 1, 156610 (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	Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 3A, 600638 (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

TUBGCP4	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
TUBGCP6	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
TUSC3	601385	Mental retardation, autosomal recessive 7, 611093 (3), Autosomal recessive
TWIST1	601622	Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant
TWNK	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Perrault syndrome 5, 616138 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant
UBA5	610552	Epileptic encephalopathy, early infantile, 44, 617132 (3), Autosomal recessive; ?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive
UBE2A	312180	Mental retardation, 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
UBR1	605981	Johanson-Blizzard syndrome, 243800 (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
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	Mental retardation, 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
USP27X	300975	Mental retardation, X-linked 105, 300984 (3), X-linked recessive
USP7	602519	No OMIM phenotype Neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies (Fountain (2019) Genet Med (21(8)), Autosomal domionant
USP9X	300072	Mental retardation, X-linked 99, 300919 (3), X-linked recessive; Mental retardation, 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
VLDLR	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
VPS11	608549	Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
VPS13B	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS37A	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
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	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	?Mental retardation, autosomal recessive 43, 615817 (3), Autosomal recessive
WDR19	608151	?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
WDR26	617424	Skraban-Deardorff syndrome, 617616 (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
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	?Cataract 41, 116400 (3), Autosomal dominant; Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant
WWOX	605131	Epileptic encephalopathy, early infantile, 28, 616211 (3), Autosomal recessive; Esophageal squamous cell carcinoma, somatic, 133239 (3); 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
YME1L1	607472	?Optic atrophy 11, 617302 (3), Autosomal recessive
YWHAG	605356	Epileptic encephalopathy, early infantile, 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); Skeletal defects, genital hypoplasia, and mental retardation, 612447 (3), Autosomal recessive
ZBTB18	608433	Mental retardation, 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	Mental retardation, autosomal recessive 56, 617125 (3), Autosomal recessive
ZC4H2	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive
ZDHHC15	300576	?Mental retardation, X-linked 91, 300577 (3), X-linked dominant
ZDHHC9	300646	Mental retardation, X-linked syndromic, Raymond type, 300799 (3)
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
ZIC2	603073	Holoprosencephaly 5, 609637 (3), Autosomal dominant
ZMIZ1	607159	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 (3), Autosomal dominant
ZMYND11	608668	Mental retardation, autosomal dominant 30, 616083 (3), Autosomal dominant
ZNF148	601897	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 (3), Autosomal dominant
ZNF292	616213	No OMIM phenotype ? Neurodevelopmental disorder with features of autism spectrum disorder (Mirzaa (2019), Genet Med), Autosomal dominant
ZNF423	604557	Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant; Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant
ZNF462	617371	Weiss-Kruszka syndrome, 618619 (3), Autosomal dominant
ZNF592	613624	Spinocerebellar ataxia, autosomal recessive 5, 251300
ZNF674	300573	Mental retardation, X-linked 92, 300851
ZNF711	314990	Mental retardation, X-linked 97, 300803 (3), X-linked
ZSWIM6	615951	Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant; Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (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: Sept 30, 2019

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.