ID & Epilepsy panel		
versie	v1 (juli 2017)	Centrum voor Medische Genetica Gent

Gene	Associated phenotype, OMIM ID
A2ML1	NO OMIM
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 29, 616339 (3), Autosomal recessive
AASS	Hyperlysinemia, 238700 (3), Autosomal recessive; Saccharopinuria, 268700 (1), Autosomal recessive
ABAT	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
ABCC8	Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 606176 (3), 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	Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Cardiomyopathy, dilated, 1O, 608569 (3); Hypertrichotic osteochondrodysplasia, 239850 (3), Autosomal dominant
ABCD1	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive
ABHD5	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ACAD9	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126 (3), Autosomal recessive
ACO2	Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive; ?Optic atrophy 9, 616289 (3), Autosomal recessive
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
ACSF3	Combined malonic and methylmalonic aciduria, 614265 (3)
ACSL4	Mental retardation, X-linked 63, 300387 (3), X-linked dominant
ACTB	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
ACTG1	Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant; Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant
ACVR1	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
ACY1	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
ADAM22	NO OMIM
ADAR	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant
ADAT3	Mental retardation, autosomal recessive 36, 615286 (3), Autosomal recessive
ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
ADNP	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
ADSL	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
AFF2	Mental retardation, X-linked, FRAXE type, 309548 (3), X-linked recessive
AFF4	CHOPS syndrome, 616368 (3), Autosomal dominant
AFG3L2	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
AGA	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AGO2	NO OMIM
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
AGTR2	NO OMIM
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
AHDC1	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
AHI1	Joubert syndrome 3, 608629 (3), Autosomal recessive

AIFM1	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	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant
ALDH18A1	Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive, Isolated cases; Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive
ALDH3A2	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALDH4A1	Hyperprolinemia, type II, 239510 (3), Autosomal recessive
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive
ALG1	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
ALG11	Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive
ALG12	Congenital disorder of glycosylation, type Ig, 607143 (3)
ALG13	?Congenital disorder of glycosylation, type Is, 300884 (3), X-linked dominant; Epileptic encephalopathy, early infantile, 36, 300884 (3), X-linked dominant
ALG2	?Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
ALG3	Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive
ALG6	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
ALG8	Congenital disorder of glycosylation, type Ih, 608104 (3)
ALG9	Congenital disorder of glycosylation, type Il, 608776 (3); Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive
ALMS1	Alstrom syndrome, 203800 (3), Autosomal recessive
ALX1	?Frontonasal dysplasia 3, 613456 (3)
ALX4	{Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive; Parietal foramina 2, 609597 (3), Autosomal dominant
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
AMMECR1	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive
AMPD2	Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, 615686 (3), Autosomal recessive
AMT	Glycine encephalopathy, 605899 (3), Autosomal recessive
ANK3	?Mental retardation, autosomal recessive, 37, 615493 (3), Autosomal recessive
ANKH	Chondrocalcinosis 2, 118600 (3), Autosomal dominant; Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant
ANKLE2	?Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive
ANKRD11	KBG syndrome, 148050 (3), Autosomal dominant
ANO10	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
ANTXR1	GAPO syndrome, 230740 (3), Autosomal recessive; {Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
AP1S1	MEDNIK syndrome, 609313 (3), Autosomal recessive
AP1S2	Mental retardation, X-linked syndromic 5, 304340 (3), X-linked recessive
AP3B1	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3B2	Epileptic encephalopathy, early infantile, 48, 617276 (3), Autosomal recessive
AP4B1	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
AP4E1	Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive; Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant
AP4M1	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive

APC2	?Sotos syndrome 3, 617169 (3), Autosomal recessive
APOPT1	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
ARFGEF2	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
ARG1	Argininemia, 207800 (3), Autosomal recessive
ARHGAP31	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARHGEF6	Mental retardation, X-linked 46, 300436 (3), X-linked recessive
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607 (3), X-linked recessive
ARID1A	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
ARID1B	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
ARID2	NO OMIM
ARL13B	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL6	{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)
ARSA	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARSE	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ARX	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	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive
ASCL1	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; Haddad syndrome, 209880 (3), Autosomal dominant
ASL	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
ASNS	Asparagine synthetase deficiency, 615574 (3), Autosomal recessive
ASPA	Canavan disease, 271900 (3), Autosomal recessive
ASPM	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive
ASS1	Citrullinemia, 215700 (3), Autosomal recessive
ASXL1	Bohring-Opitz syndrome, 605039 (3), Autosomal dominant; Myelodysplastic syndrome, somatic, 614286 (3)
ASXL2	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
ASXL3	Bainbridge-Ropers syndrome, 615485 (3)
ATAD3A	Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant
ATCAY	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
ATN1	Dentatorubro-pallidoluysian atrophy, 125370 (3), Autosomal dominant
ATP1A2	Alternating hemiplegia of childhood, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP1A3	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant
ATP2A2	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
ATP6AP2	Mental retardation, X-linked, syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive; Wrinkly skin syndrome, 278250 (3), Autosomal recessive
ATP6V1B2	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant; Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant
ATP7A	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	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive

ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
ATR	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant; Seckel syndrome 1, 210600 (3), Autosomal recessive
ATRX	Mental retardation, X-linked 52, 300504 (2), X-linked recessive
ATRX	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	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
AUTS2	Mental retardation, autosomal dominant 26, 615834 (3), Autosomal dominant
AVPR2	Diabetes insipidus, nephrogenic, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
B3GALT6	Ehlers-Danlos syndrome, progeroid type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive
B3GLCT	Peters-plus syndrome, 261540 (3), Autosomal recessive
B4GALNT1	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
B4GALT1	Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive
B4GALT7	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070 (3), Autosomal recessive
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
BBS1	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
BBS10	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
BBS4	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
BCKDHA	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
BCL11A	Dias-Logan syndrome, 617101 (3), Autosomal dominant
BCOR	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BCORL1	NO OMIM
BCS1L	Bjornstad syndrome, 262000 (3), Autosomal recessive; GRACILE syndrome, 603358 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive
BLM	Bloom syndrome, 210900 (3), Autosomal recessive
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
BRAF	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	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive
BRF1	Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive
BRWD3	Mental retardation, X-linked 93, 300659 (3), X-linked recessive
BSCL2	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	Biotinidase deficiency, 253260 (3), Autosomal recessive
BUB1B	Colorectal cancer, somatic, 114500 (3); Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive; [Premature chromatid separation trait], 176430 (3), Autosomal dominant
C12orf4	NO OMIM
C12orf57	Temtamy syndrome, 218340 (3), Autosomal recessive
C12orf65	Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive; Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive
C2CD3	?Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
C5orf42	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3), Autosomal recessive
CACNA1A	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	Brugada syndrome 3, 611875 (3); Timothy syndrome, 601005 (3), Autosomal dominant
CACNA1E	NO OMIM
CACNA2D1	NO OMIM
CACNG2	?Mental retardation, autosomal dominant 10, 614256 (3)
CAD	Epileptic encephalopathy, early infantile, 50, 616457 (3), Autosomal recessive
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756 (3), Autosomal dominant
CAPN10	{Diabetes mellitus, noninsulin-dependent 1}, 601283 (3)
CASK	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	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive; Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive
CC2D1A	Mental retardation, autosomal recessive 3, 608443 (3), Autosomal recessive
CC2D2A	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
CCDC174	Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive
CCDC22	Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive
CCDC78	Myopathy, centronuclear, 4, 614807 (3), Autosomal dominant
CCDC88C	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 (3), Autosomal recessive; ?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant
CCND2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant
CDH15	Mental retardation, autosomal dominant 3, 612580 (3)
CDK5RAP2	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
CDK6	?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672 (3), X-linked dominant
CDKN1C	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; IMAGE syndrome, 614732 (3), Autosomal dominant
CDON	Holoprosencephaly 11, 614226 (3), Autosomal dominant, Isolated cases
CENPJ	Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive

CEP135	Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive
CEP152	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
CEP290	?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	Joubert syndrome 15, 614464 (3), Autosomal recessive
CEP63	?Seckel syndrome 6, 614728 (3), Autosomal recessive
CEP83	Nephronophthisis 18, 615862 (3), Autosomal recessive
CEP89	NO OMIM
CHAMP1	Mental retardation, autosomal dominant 40, 616579 (3), Autosomal dominant
CHD2	Epileptic encephalopathy, childhood-onset, 615369 (3), Autosomal dominant
CHD3	NO OMIM
CHD4	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
CHD7	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3)
CHD8	{Autism, susceptibility to, 18}, 615032 (3), Autosomal dominant
CHKB	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant; {Nicotine addiction, susceptibility to}, 188890 (3)
CHRNB2	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CIC	NO OMIM
CIT	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive
CKAP2L	Filippi syndrome, 272440 (3), Autosomal recessive
CLCN4	Mental retardation, X-linked 49/15, 300114 (3), X-linked recessive
CLCNKB	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLDN16	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
CLIC2	?Mental retardation, X-linked, syndromic 32, 300886 (3), X-linked recessive
CLIP1	NO OMIM
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive
CLP1	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive
CNKSR2	NO OMIM
CNNM2	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation, 616418 (3), Autosomal recessive, Autosomal dominant
CNTN2	?Epilepsy, myoclonic, familial adult, 5, 615400 (3), Autosomal recessive
CNTNAP2	{Autism susceptibility 15}, 612100 (3); Cortical dysplasia-focal epilepsy syndrome, 610042 (3); Pitt-Hopkins like syndrome 1, 610042 (3)
COASY	Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
COG1	Congenital disorder of glycosylation, type IIg, 611209 (3)
COG4	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive
COG5	Congenital disorder of glycosylation, type IIi, 613612 (3)
COG6	Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive; Shaheen syndrome, 615328 (3), Autosomal recessive
COG7	Congenital disorder of glycosylation, type IIe, 608779 (3)
COG8	Congenital disorder of glycosylation, type IIh, 611182 (3)

COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 607595 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 1, 175780 (3), Autosomal dominant; ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant
COL4A2	{Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 2, 614483 (3), Autosomal dominant
COL4A3BP	Mental retardation, autosomal dominant 34, 616351 (3), Autosomal dominant
COLEC11	3MC syndrome 2, 265050 (3), Autosomal recessive
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive; {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant
COQ4	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
COX15	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	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
CPA6	Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal recessive, Autosomal dominant; Febrile seizures, familial, 11, 614418 (3), Autosomal recessive
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3); {Venoocclusive disease after bone marrow transplantation} (3)
CPT2	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	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive
CRBN	Mental retardation, autosomal recessive 2, 607417 (3), Autosomal recessive
CREBBP	Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CRLF1	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
CSNK2A1	Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant
CSPP1	Joubert syndrome 21, 615636 (3), Autosomal recessive
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
CTBP1	NO OMIM
CTCF	Mental retardation, autosomal dominant 21, 615502 (3), Autosomal dominant
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
CTNNB1	Colorectal cancer, somatic, 114500 (3); Hepatocellular carcinoma, somatic, 114550 (3); Medulloblastoma, somatic, 155255 (3); Mental retardation, autosomal dominant 19, 615075 (3), Autosomal dominant; Ovarian cancer, somatic, 167000 (3); Pilomatricoma, somatic, 132600 (3)
CTNND1	NO OMIM
CTNND2	NO OMIM
CTSA	Galactosialidosis, 256540 (3), Autosomal recessive
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362 (3), Autosomal recessive
CTTNBP2	NO OMIM
CUBN	Megaloblastic anemia-1, Finnish type, 261100 (3), Autosomal recessive
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 (3), X-linked recessive
CWF19L1	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive

CYB5R3	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
CYP27A1	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
CYP2U1	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
D2HGDH	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
DAG1	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	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
DBT	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
DCAF17	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DCC	Colorectal cancer, somatic, 114500 (3); Esophageal carcinoma, somatic, 133239 (3); Mirror movements 1, 157600 (3), Autosomal dominant
DCHS1	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
DCPS	Al-Raqad syndrome, 616459 (3), Autosomal recessive
DCX	Lissencephaly, X-linked, 300067 (3), X-linked; Subcortical laminal heteropia, X-linked, 300067 (3), X-linked
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
DDHD2	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
DDX11	Warsaw breakage syndrome, 613398 (3), Autosomal recessive
DDX3X	Mental retardation, X-linked 102, 300958 (3), X-linked recessive, X-linked dominant
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 (3), Autosomal recessive; Mental retardation, autosomal dominant 24, 615828 (3), Autosomal dominant
DENND5A	Epileptic encephalopathy, early infantile, 49, 617281 (3)
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant
DHCR24	Desmosterolosis, 602398 (3), Autosomal recessive
DHCR7	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant
DIAPH1	Deafness, autosomal dominant 1, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
DIP2B	Mental retardation, FRA12A type, 136630 (3), Autosomal dominant
DKC1	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DLG3	Mental retardation, X-linked 90, 300850 (3), X-linked recessive
DLG4	NO OMIM
DMD	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
DMPK	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
DNAJC19	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3), Autosomal dominant
DNM1	Epileptic encephalopathy, early infantile, 31, 616346 (3), Autosomal dominant
DNMT3A	Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive
DOCK6	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DOCK7	Epileptic encephalopathy, early infantile, 23, 615859 (3), Autosomal recessive
DOLK	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
DONSON	NO OMIM

DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive
DPH1	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901 (3), Autosomal recessive
DPM1	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
DPM2	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive
DPP6	Mental retardation, autosomal dominant 33, 616311 (3); {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
DPYS	Dihydropyrimidinuria, 222748 (3), Autosomal recessive
DYM	Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive; Smith-McCort dysplasia, 607326 (3), Autosomal recessive
DYNC1H1	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	Mental retardation, autosomal dominant 7, 614104 (3), Autosomal dominant
EBP	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
EDC3	?Mental retardation, autosomal recessive 50, 616460 (3), Autosomal recessive
EEF1A2	Epileptic encephalopathy, early infantile, 33, 616409 (3), Autosomal dominant; Mental retardation, autosomal dominant 38, 616393 (3), Autosomal dominant
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
EGF	Hypomagnesemia 4, renal, 611718 (3)
EHMT1	Kleefstra syndrome, 610253 (3), Autosomal dominant
EIF2AK3	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
EIF4A3	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
EIF4G1	{Parkinson disease 18}, 614251 (3), Autosomal dominant
ELAC2	Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive; {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3)
ELOVL4	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	Mental retardation, autosomal recessive 58, 617270 (3), Autosomal recessive
EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
EMX2	Schizencephaly, 269160 (3)
ENTPD1	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
EP300	Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
EPB41L1	?Mental retardation, autosomal dominant 11, 614257 (3)
EPG5	Vici syndrome, 242840 (3), Autosomal recessive
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
ERCC2	Cerebrooculofacioskeletal syndrome 2, 610756 (3); Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
ERCC3	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC5	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	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 recessive; {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	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
ERLIN2	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
ESCO2	Roberts syndrome, 268300 (3), Autosomal recessive; SC phocomelia syndrome, 269000 (3), Autosomal recessive
ETFB	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
ETHE1	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
EXOSC2	NO OMIM
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
EZH2	Weaver syndrome, 277590 (3), Autosomal dominant
FA2H	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
FANCD2	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive
FARS2	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; ?Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive; Van Maldergem syndrome 2, 615546 (3), Autosomal recessive
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
FBXO31	?Mental retardation, autosomal recessive 45, 615979 (3), Autosomal recessive
FGD1	Aarskog-Scott syndrome, 305400 (3), X-linked recessive; Mental retardation, X-linked syndromic 16, 305400 (3), X-linked recessive
FGF12	Epileptic encephalopathy, early infantile, 47, 617166 (3), Autosomal dominant
FGF14	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant
FGFR1	Encephalocraniocutaneous lipomatosis, 613001 (3), Somatic mosaicism; 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	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal recessive; 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	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	Fumarase deficiency, 606812 (3), Autosomal recessive; Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant
FIBP	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive
FIGN	NO OMIM
FKRP	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	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	Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked recessive; Congenital short bowel syndrome, 300048 (3), X-linked recessive; FG syndrome 2, 300321 (3); Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive; Heterotopia, periventricular, 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)
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FMN2	Mental retardation, autosomal recessive 47, 616193 (3), Autosomal recessive
FMR1	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	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
FOXG1	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670 (3), Autosomal dominant
FOXP2	Speech-language disorder-1, 602081 (3), Autosomal dominant
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
FRAS1	Fraser syndrome, 219000 (3), Autosomal recessive
FREM2	Fraser syndrome, 219000 (3), Autosomal recessive
FRMD4A	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
FRMPD4	Mental retardation, X-linked 104, 300983 (3), X-linked recessive
FRRS1L	Epileptic encephalopathy, early infantile, 37, 616981 (3), Autosomal recessive
FTCD	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
FTO	Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive
FTSJ1	Mental retardation, X-linked 9/44, 309549 (3), X-linked recessive
FUCA1	Fucosidosis, 230000 (3), Autosomal recessive
FXYD2	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
GABRA1	{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
GABRB1	Epileptic encephalopathy, early infantile, 45, 617153 (3), Autosomal dominant
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Epileptic encephalopathy, early infantile, 43, 617113 (3), Autosomal dominant
GABRG2	{Epilepsy, childhood absence, susceptibility to, 2}, 607681 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 3, 611277 (3), Autosomal dominant; Febrile seizures, familial, 8, 611277 (3), Autosomal dominant
GAD1	?Cerebral palsy, spastic quadriplegic, 1, 603513 (3), Autosomal recessive
GALE	Galactose epimerase deficiency, 230350 (3), Autosomal recessive

GALT	Galactosemia, 230400 (3), Autosomal recessive
GAMT	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
GATAD2B	Mental retardation, autosomal dominant 18, 615074 (3), Autosomal dominant
GATM	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive
GCDH	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GCK	Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 606176 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; MODY, type II, 125851 (3), Autosomal dominant
GCSH	Glycine encephalopathy, 605899 (3), Autosomal recessive
GDI1	Mental retardation, X-linked 41, 300849 (3), X-linked dominant
GFAP	Alexander disease, 203450 (3), Autosomal dominant
GFM2	NO OMIM
GJA1	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratodermia variabilis et progressiva, 133200 (3), Autosomal recessive, 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
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive; Lymphedema, hereditary, IC, 613480 (3), Autosomal dominant; Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive
GK	Glycerol kinase deficiency, 307030 (3), X-linked recessive
GLB1	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	Glycine encephalopathy, 605899 (3), Autosomal recessive
GLI2	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant
GLI3	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	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400 (3), Autosomal recessive, Autosomal dominant
GLRB	Hyperekplexia 2, autosomal recessive, 614619 (3)
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant
GLYCTK	D-glyceric aciduria, 220120 (3), Autosomal recessive
GM2A	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
GMPPA	Alacrima, achalasia, and mental retardation syndrome, 615510 (3), Autosomal recessive
GMPPB	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	Epileptic encephalopathy, early infantile, 17, 615473 (3), Autosomal dominant
GNAS	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Isolated cases; Acromegaly, somatic, 102200 (3); McCune-Albright syndrome, somatic, mosaic, 174800 (3); Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant

GNB1	Leukemia, acute lymphoblastic, somatic, 613065 (3); Mental retardation, autosomal dominant 42, 616973 (3), Autosomal dominant
GNB5	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	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
GNPTAB	Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive; Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive
GNS	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
GOSR2	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
GPHN	Molybdenum cofactor deficiency C, 615501 (3)
GPT2	Mental retardation, autosomal recessive 49, 616281 (3), Autosomal recessive
GRIA3	Mental retardation, X-linked 94, 300699 (3), X-linked recessive
GRID2	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
GRIK2	Mental retardation, autosomal recessive, 6, 611092 (3), Autosomal recessive
GRIN1	Mental retardation, autosomal dominant 8, 614254 (3)
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570 (3), Autosomal dominant
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 (3), Autosomal dominant; Mental retardation, autosomal dominant 6, 613970 (3)
GRIN3B	NO OMIM
GRIP1	Fraser syndrome, 219000 (3), Autosomal recessive
GRM1	Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive
GRN	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
GSE1	NO OMIM
GSS	Glutathione synthetase deficiency, 266130 (3), Autosomal recessive; Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive
GTF2H5	Trichothiodystrophy 3, photosensitive, 616395 (3)
GTPBP3	Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive
GUSB	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive
HAX1	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
HCCS	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541 (3), X-linked recessive
HCN1	Epileptic encephalopathy, early infantile, 24, 615871 (3), Autosomal dominant
HDAC4	NO OMIM
HDAC6	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant
HDAC8	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
HECTD1	NO OMIM
HECW2	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 (3), Autosomal dominant
HEPACAM	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	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive

HERC2	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	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	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
HIVEP2	Mental retardation, autosomal dominant 43, 616977 (3), Autosomal dominant
HLCS	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
HMGCL	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
HNMT	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; Mental retardation, autosomal recessive 51, 616739 (3), Autosomal recessive
HNRNPH2	Mental retardation, X-linked, syndromic, Bain type, 300986 (3), X-linked dominant
HNRNPK	Au-Kline syndrome, 616580 (3), Autosomal dominant
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391 (3), Autosomal dominant
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 (3); Bosley-Salih-Alorainy syndrome, 601536 (3)
HPD	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
HPRT1	HPRT-related gout, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
HRAS	{Bladder cancer, somatic}, 109800 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant, Isolated cases; Costello syndrome, 218040 (3), Autosomal dominant, Isolated cases; {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	HDS10 mitochondrial disease, 300438 (3), X-linked dominant
HSD17B4	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
HSPA9	Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant; Even-plus syndrome, 616854 (3), Autosomal recessive
HSPD1	Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive; Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant
HUWE1	Mental retardation, X-linked syndromic, Turner type, 300706 (3)
IARS	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
IDH2	D-2-hydroxyglutaric aciduria 2, 613657 (3)
IDS	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
IDUA	Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
IFIH1	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IFT172	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT81	NO OMIM
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 (3), X-linked recessive
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive

IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 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	Mental retardation, X-linked 21/34, 300143 (3), X-linked recessive
IMPA1	Mental retardation, autosomal recessive 59, 617323 (3), Autosomal recessive
INPP5E	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
IQSEC2	Mental retardation, X-linked 1/78, 309530 (3), X-linked dominant
ISPD	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
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
ITPR1	Gillespie syndrome, 206700 (3); Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant
IVD	Isovaleric acidemia, 243500 (3), Autosomal recessive
JAG1	Alagille syndrome 1, 118450 (3), Autosomal dominant; ?Deafness, congenital heart defects, and posterior embryotoxon (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
JMJD1C	NO OMIM
KALRN	{Coronary heart disease, susceptibility to, 5}, 608901 (3)
KANK1	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
KANSL1	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant
KAT6A	Mental retardation, autosomal dominant 32, 616268 (3), Autosomal dominant
KAT6B	Genitopatellar syndrome, 606170 (3), Autosomal dominant; SBBYSS syndrome, 603736 (3)
KATNB1	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
KCNA1	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNA2	Epileptic encephalopathy, early infantile, 32, 616366 (3), Autosomal dominant
KCNA4	NO OMIM
KCNB1	Epileptic encephalopathy, early infantile, 26, 616056 (3), Autosomal dominant
KCNC1	Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant
KCNC3	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
KCNH1	Temple-Baraitser syndrome, 611816 (3), Autosomal dominant; Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
KCNJ11	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 dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant
KCNJ6	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
KCNK9	Birk-Barel mental retardation dysmorphism syndrome, 612292 (3)
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446 (3), Autosomal dominant
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant
KCNQ3	Seizures, benign neonatal, type 2, 121201 (3), Autosomal dominant
KCNQ5	NO OMIM
KCNT1	Epilepsy, nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 14, 614959 (3), Autosomal dominant

KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
KDM1A	Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
KDM6A	Kabuki syndrome 2, 300867 (3), X-linked dominant
KIAA0586	Joubert syndrome 23, 616490 (3), Autosomal recessive; Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive
KIAA1109	NO OMIM
KIDINS220	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
KIF1A	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	Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive
KIF2A	Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant
KIF4A	?Mental retardation, X-linked 100, 300923 (3), X-linked recessive
KIF5C	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant
KIF7	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	Mental retardation, autosomal dominant 4, 612581 (3)
KLHL15	Mental retardation, X-linked 103, 300982 (3), X-linked recessive
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 (2), Autosomal dominant; Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
KMT2B	Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
KMT2C	NO OMIM
KMT2D	Kabuki syndrome 1, 147920 (3), Autosomal dominant
KNL1	Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive
KPTN	Mental retardation, autosomal recessive 41, 615637 (3), Autosomal recessive
KRAS	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; Lung cancer, somatic, 211980 (3); Noonan syndrome 3, 609942 (3); Pancreatic carcinoma, somatic, 260350 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)
L1CAM	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	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LAMA1	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
LAMA2	Muscular dystrophy, congenital merosin-deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3), Autosomal recessive
LAMC3	Cortical malformations, occipital, 614115 (3), Autosomal recessive
LAMP2	Danon disease, 300257 (3), X-linked dominant
LARGE1	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	Alazami syndrome, 615071 (3), Autosomal recessive

LAS1L	Wilson-Turner syndrome, 309585 (3), X-linked recessive
LGI1	Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive
LIG4	LIG4 syndrome, 606593 (3); {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
LINS1	Mental retardation, autosomal recessive 27, 614340 (3), Autosomal recessive
LMAN2L	?Mental retardation, autosomal recessive, 52, 616887 (3), Autosomal recessive
LONP1	CODAS syndrome, 600373 (3), Autosomal recessive
LRP2	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRPPRC	Leigh syndrome, French-Canadian type, 220111 (3), Autosomal recessive
LZTFL1	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
MAF	Ayme-Gripp syndrome, 601088 (3), Autosomal dominant; Cataract 21, multiple types, 610202 (3), Autosomal dominant
MAGEL2	Schaaf-Yang syndrome, 615547 (3), Autosomal dominant
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3)
MAN1B1	Mental retardation, autosomal recessive 15, 614202 (3), Autosomal recessive
MAN2B1	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MANBA	Mannosidosis, beta, 248510 (3), Autosomal recessive
MAOA	{Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279 (3)
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280 (3)
MAPRE2	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant
MASP1	3MC syndrome 1, 257920 (3), Autosomal recessive
MAT1A	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	Mental retardation, autosomal dominant 1, 156200 (3), Autosomal dominant
MBOAT7	Mental retardation, autosomal recessive 57, 617188 (3), Autosomal recessive
MBTPS2	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
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive
MCOLN1	Mucolipidosis IV, 252650 (3), Autosomal recessive
MCPH1	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
MDH2	Epileptic encephalopathy, early infantile, 51, 617339 (3), Autosomal recessive
MECP2	{Autism susceptibility, X-linked 3}, 300496 (3), Isolated cases, X-linked, Multifactorial; 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	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive
MED12	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
MED13L	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	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive
MED23	Mental retardation, autosomal recessive 18, 614249 (3), Autosomal recessive
MED25	Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive

MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 (3), Autosomal dominant
MEGF8	Carpenter syndrome 2, 614976 (3), Autosomal recessive
MEIS2	Cleft palate, cardiac defects, and mental retardation, 600987 (3), Autosomal dominant
METTL23	Mental retardation, autosomal recessive 44, 615942 (3), Autosomal recessive
MFSD2A	Microcephaly 15, primary, autosomal recessive, 616486 (3), Autosomal recessive
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive; Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive
MGAT2	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive
MICU1	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
MID1	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
MID2	?Mental retardation, X-linked 101, 300928 (3), X-linked recessive
MKKS	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
MKS1	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100 (3), Autosomal recessive
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MMADHC	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
MOCS1	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
MOCS2	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
MOGS	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MPDU1	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MPDZ	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219 (3), Autosomal recessive
MPLKIP	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
MRPL3	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
MSL3	NO OMIM
MTFMT	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive
MTHFR	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	Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MUT	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
MVK	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Mevalonic aciduria, 610377 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant
MYCN	Feingold syndrome 1, 164280 (3), Autosomal dominant
MYH9	Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant; Epstein syndrome, 153650 (3), Autosomal dominant; Fechtner syndrome, 153640 (3), Autosomal dominant; Macrothrombocytopenia and progressive sensorineural deafness, 600208 (3), Autosomal dominant; May-Hegglin anomaly, 155100 (3), Autosomal dominant; Sebastian syndrome, 605249 (3), Autosomal dominant
MYO5A	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive

MYT1L	Mental retardation, autosomal dominant 39, 616521 (3), Autosomal dominant
NAA10	?Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
NACC1	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant
NAGA	Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type I, 609241 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
NALCN	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	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive
NARS2	Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive
NBN	Aplastic anemia, 609135 (3); Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NDE1	Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive; ?Microhydranencephaly, 605013 (3), Autosomal recessive
NDP	Exudative vitreoretinopathy 2, X-linked, 305390 (3); Norrie disease, 310600 (3), X-linked recessive
NDST1	Mental retardation, autosomal recessive 46, 616116 (3), Autosomal recessive
NDUFA1	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFA11	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFA12	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000 (3), Autosomal recessive, Mitochondrial
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial
NDUFAF1	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFAF2	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFAF3	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFAF4	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFAF5	Mitochondrial complex 1 deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFB3	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFB9	?Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS1	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS2	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS4	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS6	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS7	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial

NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial
NDUFV1	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFV2	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NECAP1	?Epileptic encephalopathy, early infantile, 21, 615833 (3), Autosomal recessive
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
NEDD4L	Periventricular nodular heterotopia 7, 617201 (3), Autosomal dominant
NEU1	Sialidosis, type I, 256550 (3), Autosomal recessive; Sialidosis, type II, 256550 (3), Autosomal recessive
NEXMIF	Mental retardation, X-linked 98, 300912 (3), X-linked recessive
NF1	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
NFATC1	NO OMIM
NFIA	Brain malformations and urinary tract defects, 613735 (3), Isolated cases
NFIX	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Sotos syndrome 2, 614753 (3), Autosomal dominant
NGLY1	Congenital disorder of deglycosylation, 615273 (3), Autosomal recessive
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
NHS	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
NIPBL	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NKX2-1	Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant; {Thyroid cancer, monmedullary, 1}, 188550 (3), Autosomal dominant
NLGN3	{Asperger syndrome susceptibility, X-linked 1}, 300494 (3), Isolated cases, X-linked, Multifactorial; {Autism susceptibility, X-linked 1}, 300425 (3), Isolated cases, X-linked, Multifactorial
NLGN4X	{Asperger syndrome susceptibility, X-linked 2}, 300497 (3), Isolated cases, X-linked, Multifactorial; {Autism susceptibility, X-linked 2}, 300495 (3), Isolated cases, X-linked, Multifactorial; Mental retardation, X-linked, 300495 (3), Isolated cases, X-linked, Multifactorial
NLRP3	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold-induced inflammatory syndrome 1, 120100 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NONO	Mental retardation, X-linked, syndromic 34, 300967 (3), X-linked
NPC1	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NPHP1	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPRL2	Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant
NPRL3	Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
NRAS	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	Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3)

NSD1	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (1), Autosomal dominant; Sotos syndrome 1, 117550 (3), Autosomal dominant
NSDHL	CHILD syndrome, 308050 (3), X-linked dominant; CK syndrome, 300831 (3), X-linked recessive
NSUN2	Mental retardation, autosomal recessive 5, 611091 (3), Autosomal recessive
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive; Medullary thyroid carcinoma, familial, 155240 (3), Autosomal dominant
NUBPL	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NUP62	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
OCLN	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
OCRL	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
ODC1	{Colonic adenoma recurrence, reduced risk of}, 114500 (3), Autosomal dominant
OFD1	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
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 (3), X-linked recessive
ORC1	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
OTC	Ornithine transcarbamylase deficiency, 311250 (3), X-linked recessive
PACS1	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
PAFAH1B1	Lissencephaly 1, 607432 (3), Isolated cases; Subcortical laminar heterotopia, 607432 (3), Isolated cases
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
PAK3	Mental retardation, X-linked 30/47, 300558 (3), X-linked recessive
PANK2	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PANX1	NO OMIM
PAX1	?Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
PAX6	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	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
PC	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
PCCA	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	Propionicacidemia, 606054 (3), Autosomal recessive
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088 (3), X-linked
PCGF2	NO OMIM
PCLO	?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant; {Stroke, susceptibility to, 1}, 606799 (3)
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3)
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive

PDX1	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; MODY, type IV, 606392 (3); Pancreatic agenesis 1, 260370 (3), Autosomal recessive
PEPD	Prolidase deficiency, 170100 (3), Autosomal recessive
PET100	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
PEX1	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	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
PEX11B	?Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive; Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive; Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
PEX5	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	Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive
PEX7	Peroxisome biogenesis disorder 9B, 614879 (3); Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
PGAP1	Mental retardation, autosomal recessive 42, 615802 (3), Autosomal recessive
PGAP2	Hyperphosphatasia with mental retardation syndrome 3, 614207 (3), Autosomal recessive
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716 (3), Autosomal recessive
PGK1	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PGM3	Immunodeficiency 23, 615816 (3), Autosomal recessive
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
PHF8	Mental retardation syndrome, X-linked, Siderius type, 300263 (3), X-linked recessive
PHGDH	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PHIP	NO OMIM
PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
PIGC	NO OMIM
PIGG	Mental retardation, autosomal recessive 53, 616917 (3), Autosomal recessive
PIGL	CHIME syndrome, 280000 (3), Autosomal recessive
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749 (3), Autosomal recessive
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive; ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive

PIGW	?Hyperphosphatasia with mental retardation syndrome 5, 616025 (3), Autosomal recessive
PIGY	Hyperphosphatasia with mental retardation syndrome 6, 616809 (3), Autosomal recessive
PIK3CA	Breast cancer, somatic, 114480 (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); 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	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant
PLA2G6	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
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722 (3), Autosomal recessive
PLP1	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PLXND1	NO OMIM
PMM2	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PMPCA	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
PNKP	Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
PNPO	Pyridoxamine 5-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
POGZ	White-Sutton syndrome, 616364 (3), Autosomal dominant
POLG	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	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive
POMGNT1	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	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 (3), Autosomal recessive
POMK	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	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	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	Focal dermal hypoplasia, 305600 (3), X-linked dominant

POU1F1	Pituitary hormone deficiency, combined, 1, 613038 (3), Autosomal recessive, Autosomal dominant
POU3F3	NO OMIM
PPOX	Porphyria variegata, 176200 (3), Autosomal dominant
PPP1CB	NO OMIM
PPP1R15B	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
PPP2R1A	Mental retardation, autosomal dominant 36, 616362 (3), Autosomal dominant
PPP2R5B	NO OMIM
PPP2R5C	NO OMIM
PPP2R5D	Mental retardation, autosomal dominant 35, 616355 (3), Autosomal dominant
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
PQBP1	Renpenning syndrome, 309500 (3), X-linked recessive
PRICKLE1	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
PRICKLE2	NO OMIM
PRKAR1A	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	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
PRODH	Hyperprolinemia, type I, 239500 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant
PRPS1	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
PRRT2	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	Mental retardation, autosomal recessive 1, 249500 (3), Autosomal recessive
PSAP	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	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, Isolated cases
PTCH1	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Holoprosencephaly 7, 610828 (3), Autosomal dominant
PTCHD1	{Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive
PTDSS1	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
PTEN	Bannayan-Riley-Ruvalcaba syndrome, 153480 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; Malignant melanoma, somatic, 155600 (3); {Meningioma}, 607174 (3), Autosomal dominant; PTEN hamartoma tumor syndrome (3); {Prostate cancer, somatic}, 176807 (3); Squamous cell carcinoma, head and neck, somatic, 275355 (3); VATER association with macrocephaly and ventriculomegaly, 276950 (3), Autosomal recessive
PTPN11	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	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
PTRHD1	NO OMIM
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
PUF60	Verheij syndrome, 615583 (3), Autosomal dominant
PURA	Mental retardation, autosomal dominant 31, 616158 (3), Autosomal dominant
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
PUS3	?Mental retardation, autosomal recessive 55, 617051 (3), Autosomal recessive
PUS7	NO OMIM
PYCR1	Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIIB, 614438 (3)
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
RAB18	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
RAB27A	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
RAB39B	Mental retardation, X-linked 72, 300271 (3), X-linked recessive; ?Waisman syndrome, 311510 (3), X-linked recessive
RAB3GAP1	Warburg micro syndrome 1, 600118 (3), Autosomal recessive
RAB3GAP2	Martsolf syndrome, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
RAB40AL	NO OMIM
RAC1	NO OMIM
RAD21	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant
RAF1	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3)
RAI1	Smith-Magenis syndrome, 182290 (3), Autosomal dominant, Isolated cases
RARB	Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant
RARS2	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
RBBP8	Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3); Seckel syndrome 2, 606744 (3), Autosomal recessive
RBFOX1	NO OMIM
RBM10	TARP syndrome, 311900 (3), X-linked recessive
RBM28	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
RBPJ	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
RCBTB1	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
RELN	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
REV3L	NO OMIM
RFT1	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
RHEB	NO OMIM
RIT1	Noonan syndrome 8, 615355 (3), Autosomal dominant
RLIM	Mental retardation, X-linked 61, 300978 (3), X-linked recessive
RMND1	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive
RMRP	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
RNF113A	?Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked dominant

RNF125	Tenorio syndrome, 616260 (3), Autosomal dominant
ROGDI	Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive
RPGRIP1L	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
RPL10	{Autism, susceptibility to, X-linked 5}, 300847 (3)
RPS6KA3	Coffin-Lowry syndrome, 303600 (3), X-linked dominant, Isolated cases; Mental retardation, X-linked 19, 300844 (3), X-linked dominant
RRM2B	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	Spondylopeimetaphyseal dsyplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive
RTEL1	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
RTTN	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
RUBCN	?Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
RUSC2	NO OMIM
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant; Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
SATB2	Glass syndrome, 612313 (3), Autosomal dominant
SBDS	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
SC5D	Lathosterolosis, 607330 (3), Autosomal recessive
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 6, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant
SCN1B	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	Epileptic encephalopathy, early infantile, 11, 613721 (3), Autosomal dominant; Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant
SCO1	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 (3), Autosomal recessive; Myopia 6, 608908 (3), Autosomal dominant
SDHA	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	?CHARGE syndrome, 214800 (3), Autosomal dominant
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SETBP1	Mental retardation, autosomal dominant 29, 616078 (3), Autosomal dominant; Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant
SETD1A	NO OMIM

SETD2	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant
SETD5	Mental retardation, autosomal dominant 23, 615761 (3), Autosomal dominant
SF1	NO OMIM
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
SHANK2	{Autism susceptibility 17}, 613436 (3)
SHANK3	Phelan-McDermid syndrome, 606232 (3), Isolated cases; {Schizophrenia 15}, 613950 (3)
SHH	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	Noonan-like syndrome with loose anagen hair, 607721 (3), Autosomal dominant
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome, 300434 (3), X-linked
SIK1	Epileptic encephalopathy, early infantile, 30, 616341 (3), Autosomal dominant
SIL1	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SIN3A	Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant
SIX3	Holoprosencephaly 2, 157170 (3), Autosomal dominant, Isolated cases; Schizencephaly, 269160 (3)
SKI	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive
SLC13A5	Epileptic encephalopathy, early infantile, 25, 615905 (3), Autosomal recessive
SLC16A1	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	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked dominant
SLC17A5	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
SLC1A1	Dicarboxylic aminoaciduria, 222730 (3), Autosomal recessive; {?Schizophrenia susceptibility 18}, 615232 (3)
SLC1A2	Epileptic encephalopathy, early infantile, 41, 617105 (3), Autosomal dominant
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive
SLC25A12	Epileptic encephalopathy, early infantile, 39, 612949 (3), Autosomal recessive
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304 (3), Autosomal recessive
SLC2A1	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	Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive; Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585 (3), Autosomal recessive
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896 (3), X-linked dominant, Somatic mosaicism
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC39A12	NO OMIM
SLC39A8	Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive
SLC6A1	Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant
SLC6A17	Mental retardation, autosomal recessive 48, 616269 (3), Autosomal recessive
SLC6A3	{Nicotine dependence, protection against}, 188890 (3); Parkinsonism-dystonia, infantile, 613135 (3), Autosomal recessive

SLC6A8	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
SLC7A7	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243 (3), X-linked dominant
SMAD4	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
SMARCA2	Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant
SMARCA4	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant
SMARCB1	Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid predisposition syndrome 1}, 609322 (3), Autosomal dominant; Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant
SMARCC2	NO OMIM
SMARCE1	Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant; {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant
SMC1A	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant
SMC3	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
SMOC1	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SMPD1	Niemann-Pick disease, type A, 257200 (3), Autosomal recessive; Niemann-Pick disease, type B, 607616 (3), Autosomal recessive
SMS	Mental retardation, X-linked, Snyder-Robinson type, 309583 (3), X-linked recessive
SNAP25	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501 (3), Autosomal recessive
SNRPN	Prader-Willi syndrome, 176270 (3), Isolated cases
SNX14	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
SOBP	Mental retardation, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive
SON	ZTTK syndrome, 617140 (3), Autosomal dominant
SOS1	?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant; Noonan syndrome 4, 610733 (3), Autosomal dominant
SOS2	Noonan syndrome 9, 616559 (3), Autosomal dominant
SOX10	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	Mental retardation, autosomal dominant, 27, 615866 (3), Autosomal dominant
SOX2	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
SOX5	Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant
SPAST	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome, 616577 (3), Autosomal recessive
SPG11	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
SPOCK1	NO OMIM
SPRED1	Legius syndrome, 611431 (3), Autosomal dominant
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477 (3), Autosomal dominant
SPTBN2	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
SRCAP	Floating-Harbor syndrome, 136140 (3), Autosomal dominant
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive; Kahrizi syndrome, 612713 (3), Autosomal recessive

SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 (3)
SSR4	Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive
ST3GAL3	?Epileptic encephalopathy, early infantile, 15, 615006 (3), Autosomal recessive; Mental retardation, autosomal recessive 12, 611090 (3), Autosomal recessive
ST3GAL5	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
STAG1	NO OMIM
STAMBP	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
STIL	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive; Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive
STT3A	?Congenital disorder of glycosylation, type Iw, 615596 (3), Autosomal recessive
STT3B	?Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
STX1B	Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164 (3), Autosomal dominant
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
SUMF1	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
SUOX	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3), X-linked recessive, X-linked dominant
SYNCRIP	NO OMIM
SYNE1	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
SYNGAP1	Mental retardation, autosomal dominant 5, 612621 (3), Autosomal dominant
SYP	Mental retardation, X-linked 96, 300802 (3), X-linked dominant
SYT14	Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive
SZT2	Epileptic encephalopathy, early infantile, 18, 615476 (3), Autosomal recessive
TAF1	Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive; Mental retardation, X-linked, syndromic 33, 300966 (3), X-linked recessive
TAF2	Mental retardation, autosomal recessive 40, 615599 (3), Autosomal recessive
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TAT	Tyrosinemia, type II, 276600 (3), Autosomal recessive
TBC1D20	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
TBC1D24	DOOR syndrome, 220500 (3), Autosomal recessive; Deafness , autosomal recessive 86, 614617 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 16, 615338 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive
TBC1D7	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 (3), Autosomal recessive
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
TBCE	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	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive
TBL1XR1	Mental retardation, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant
TBP	{Parkinson disease, susceptibility to}, 168600 (3), Isolated cases, Multifactorial; Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant
TBR1	NO OMIM

TCF20	NO OMIM
TCF4	Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant; Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant
TCF7L2	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
TCN2	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
TCTN3	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
TDP2	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive
TECPR2	Spastic paraplegia 49, autosomal recessive, 615031 (3), Autosomal recessive
TECR	Mental retardation, autosomal recessive 14, 614020 (3), Autosomal recessive
TELO2	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive
TFAP2A	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
TGFBR1	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
TGIF1	Holoprosencephaly 4, 142946 (3), Autosomal dominant
TH	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
THOC2	Mental retardation, X-linked 12/35, 300957 (3), X-linked recessive
THOC6	Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive
THRB	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
TIMM8A	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TINF2	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TLK2	NO OMIM
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980 (3), Autosomal recessive
TMEM165	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
TMEM231	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	Joubert syndrome 14, 614424 (3), Autosomal recessive
TMEM240	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
TMEM67	{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
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
TMLHE	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
TNIK	Mental retardation, autosomal recessive 54, 617028 (3), Autosomal recessive
TOE1	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
TPO	Thyroid dyshormonogenesis 2A, 274500 (3), Autosomal recessive
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TRAPPC11	Muscular dystrophy, limb-girdle, type 2S, 615356 (3), Autosomal recessive
TRAPPC9	Mental retardation, autosomal recessive 13, 613192 (3), Autosomal recessive
TREX1	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	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2H, 254110 (3), Autosomal recessive

TRIO	Mental retardation, autosomal dominant 44, 617061 (3), Autosomal dominant
TRIP12	NO OMIM
TRMT1	NO OMIM
TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
TRPM6	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
TSC1	Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, 606690 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, somatic, 606690 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
TSEN15	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
TSHB	Hypothryoidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
TSPAN7	Mental retardation, X-linked 58, 300210 (3), X-linked recessive
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
TTC37	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
TTC8	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TTI2	Mental retardation, autosomal recessive 39, 615541 (3), Autosomal recessive
TUBA1A	Lissencephaly 3, 611603 (3), Autosomal dominant
TUBA8	Polymicrogyria with optic nerve hypoplasia, 613180 (3), Autosomal recessive
TUBB	Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant; Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant
TUBB2B	Polymicrogyria, symmetric or asymmetric, 610031 (3), Autosomal dominant
TUBB3	Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant
TUBB4A	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
TUSC3	Mental retardation, autosomal recessive 7, 611093 (3), Autosomal recessive
TWIST1	Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Saethre-Chotzen syndrome with eyelid anomalies, 101400 (3), Autosomal dominant
TWNK	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	Epileptic encephalopathy, early infantile, 44, 617132 (3), Autosomal recessive; ?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type, 300860 (3), X-linked recessive
UBE3A	Angelman syndrome, 105830 (3), Isolated cases
UBE3B	Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive
UBR1	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive
UPB1	Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive

UPF3B	Mental retardation, X-linked, syndromic 14, 300676 (3), X-linked recessive
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
UROC1	?Urocanase deficiency, 276880 (3), Autosomal recessive
USP27X	Mental retardation 105, 300984 (3), X-linked recessive
USP7	NO OMIM
USP9X	Mental retardation, X-linked 99, 300919 (3), X-linked recessive; Mental retardation, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
VPS11	Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
VPS13B	Cohen syndrome, 216550 (3), Autosomal recessive
VPS37A	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
VPS53	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
VRK1	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
VWA3B	?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive
WAC	Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant
WASHC4	?Mental retardation, autosomal recessive 43, 615817 (3), Autosomal recessive
WDR13	NO OMIM
WDR19	?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
WDR45	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
WDR73	Galloway-Mowat syndrome, 251300 (3), Autosomal recessive
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive
WFS1	?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, 222300 (3), Autosomal recessive; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 (3), Autosomal recessive; Esophageal squamous cell carcinoma, somatic, 133239 (3); Spinocrebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
XK	McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked
XPA	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
XYLT1	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
YAP1	Coloboma, ocular, 120433 (3), Autosomal dominant; Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 (3), Autosomal dominant
YME1L1	?Optic atrophy 11, 617302 (3), Autosomal recessive
YWHAE	NO OMIM
YY1	NO OMIM
ZBTB16	Leukemia, acute promyelocytic, PL2F/RARA type (3); Skeletal defects, genital hypoplasia, and mental retardation, 612447 (3), Autosomal recessive
ZBTB18	Mental retardation, autosomal dominant 22, 612337 (3), Autosomal dominant
ZBTB20	Primrose syndrome, 259050 (3), Autosomal dominant
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069 (3), Autosomal recessive
ZC3H14	Mental retardation, autosomal recessive 56, 617125 (3), Autosomal recessive
ZC4H2	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive
ZDHHC15	?Mental retardation, X-linked 91, 300577 (3), X-linked dominant
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type, 300799 (3)
ZEB2	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant

ZFYVE26	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
ZIC1	Craniosynostosis 6, 616602 (3), Autosomal dominant
ZIC2	Holoprosencephaly 5, 609637 (3), Autosomal dominant
ZMYND11	Mental retardation, autosomal dominant 30, 616083 (3), Autosomal dominant
ZNF292	NO OMIM
ZNF407	NO OMIM
ZNF41	Mental retardation, X-linked 89, 300848 (2), X-linked dominant
ZNF592	NO OMIM
ZNF674	Mental retardation, X-linked 92, 300851 (2), X-linked recessive
ZNF711	Mental retardation, X-linked 97, 300803 (3), X-linked
ZNF81	Mental retardation, X-linked 45, 300498 (2), X-linked
ZSWIM6	Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant

Gene symbols used are according to the HGNC guidelines.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: June 06, 2017

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