

<b>ID en epilepsie panel (v2)</b>
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**versie** 29-11-2018  
(1174 genen)

Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>AARS</i>	601065	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 29, 616339 (3), Autosomal recessive
<i>AASS</i>	605113	Hyperlysinemia, 238700 (3), Autosomal recessive; Saccharopinuria, 268700 (1), Autosomal recessive
<i>ABAT</i>	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
<i>ABCC8</i>	600509	Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 606176 (3), Autosomal recessive, Autosomal dominant; Diabetes mellitus, transient neonatal 2, 610374 (3); Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal recessive, Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant
<i>ABCC9</i>	601439	Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Cardiomyopathy, dilated, 10, 608569 (3); Hypertrichotic osteochondrodysplasia, 239850 (3), Autosomal dominant
<i>ABCD1</i>	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
<i>ABCD4</i>	603214	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive
<i>ABHD5</i>	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
<i>ACAD9</i>	611103	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126 (3), Autosomal recessive
<i>ACO2</i>	100850	Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive; ?Optic atrophy 9, 616289 (3), Autosomal recessive
<i>ACOX1</i>	609751	Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
<i>ACSF3</i>	614245	Combined malonic and methylmalonic aciduria, 614265 (3)
<i>ACSL4</i>	300157	Mental retardation, X-linked 63, 300387 (3), X-linked dominant
<i>ACTB</i>	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
<i>ACTG1</i>	102560	Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant; Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant
<i>ACVR1</i>	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
<i>ACY1</i>	104620	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
<i>ADAR</i>	146920	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant

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<i>ADAT3</i>	615302	Mental retardation, autosomal recessive 36, 615286 (3), Autosomal recessive
<i>ADGRG1</i>	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
<i>ADK</i>	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
<i>ADNP</i>	611386	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
<i>ADSL</i>	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
<i>AFF2</i>	300806	Mental retardation, X-linked, FRAXE type, 309548 (3), X-linked recessive
<i>AFF4</i>	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
<i>AFG3L2</i>	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
<i>AGA</i>	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
<i>AGPAT2</i>	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
<i>AHCY</i>	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
<i>AHDC1</i>	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
<i>AHI1</i>	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
<i>AIFM1</i>	300169	Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
<i>AIMP1</i>	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
<i>AK1</i>	103000	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
<i>AKT3</i>	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant
<i>ALDH18A1</i>	138250	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
<i>ALDH3A2</i>	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
<i>ALDH4A1</i>	606811	Hyperprolinemia, type II, 239510 (3), Autosomal recessive
<i>ALDH5A1</i>	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
<i>ALDH7A1</i>	107323	Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive
<i>ALG1</i>	605907	Congenital disorder of glycosylation, type I <sub>k</sub> , 608540 (3), Autosomal recessive
<i>ALG11</i>	613666	Congenital disorder of glycosylation, type I <sub>p</sub> , 613661 (3), Autosomal recessive
<i>ALG12</i>	607144	Congenital disorder of glycosylation, type I <sub>g</sub> , 607143 (3), Autosomal recessive
<i>ALG13</i>	300776	?Congenital disorder of glycosylation, type I <sub>s</sub> , 300884 (3), X-linked dominant; Epileptic encephalopathy, early infantile, 36, 300884 (3), X-linked dominant
<i>ALG2</i>	607905	?Congenital disorder of glycosylation, type I <sub>i</sub> , 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
<i>ALG3</i>	608750	Congenital disorder of glycosylation, type I <sub>d</sub> , 601110 (3), Autosomal recessive
<i>ALG6</i>	604566	Congenital disorder of glycosylation, type I <sub>c</sub> , 603147 (3), Autosomal recessive
<i>ALG8</i>	608103	Congenital disorder of glycosylation, type I <sub>h</sub> , 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant

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<i>ALG9</i>	606941	Congenital disorder of glycosylation, type II, 608776 (3), Autosomal recessive; Gillespie-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive
<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>ALX1</i>	601527	?Frontonasal dysplasia 3, 613456 (3)
<i>ALX4</i>	605420	{Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive; Parietal foramina 2, 609597 (3), Autosomal dominant
<i>AMACR</i>	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
<i>AMER1</i>	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
<i>AMMECR1</i>	300195	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive
<i>AMPD2</i>	102771	Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, 615686 (3), Autosomal recessive
<i>AMT</i>	238310	Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>ANK3</i>	600465	?Mental retardation, autosomal recessive, 37, 615493 (3), Autosomal recessive
<i>ANKH</i>	605145	Chondrocalcinosis 2, 118600 (3), Autosomal dominant; Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant
<i>ANKLE2</i>	616062	?Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive
<i>ANKRD11</i>	611192	KBG syndrome, 148050 (3), Autosomal dominant
<i>ANO10</i>	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
<i>ANTXR1</i>	606410	GAPPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
<i>AP1S1</i>	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
<i>AP1S2</i>	300629	Mental retardation, X-linked syndromic 5, 304340 (3), X-linked recessive
<i>AP3B1</i>	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
<i>AP3B2</i>	602166	Epileptic encephalopathy, early infantile, 48, 617276 (3), Autosomal recessive
<i>AP3D1</i>	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
<i>AP4B1</i>	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
<i>AP4E1</i>	607244	Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive; Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant
<i>AP4M1</i>	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
<i>AP4S1</i>	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive
<i>AP5Z1</i>	613653	Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive
<i>APC2</i>	612034	?Sotos syndrome 3, 617169 (3), Autosomal recessive
<i>APOPT1</i>	616003	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>APTX</i>	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
<i>ARFGEF2</i>	605371	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
<i>ARG1</i>	608313	Argininemia, 207800 (3), Autosomal recessive
<i>ARHGAP31</i>	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
<i>ARHGEF6</i>	300267	?Mental retardation, X-linked 46, 300436 (3), X-linked recessive
<i>ARHGEF9</i>	300429	Epileptic encephalopathy, early infantile, 8, 300607 (3), X-linked recessive

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<i>ARID1A</i>	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
<i>ARID1B</i>	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
<i>ARID2</i>	609539	Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant
<i>ARL13B</i>	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
<i>ARL6</i>	608845	{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)
<i>ARSA</i>	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<i>ARSE</i>	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
<i>ARX</i>	300382	Epileptic encephalopathy, early infantile, 1, 308350 (3), X-linked recessive; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Lissencephaly, X-linked 2, 300215 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive; Partington syndrome, 309510 (3), X-linked recessive; Proud syndrome, 300004 (3), X-linked
<i>ASAH1</i>	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive
<i>ASCL1</i>	100790	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; Haddad syndrome, 209880 (3), Autosomal dominant
<i>ASH1L</i>	607999	Mental retardation, autosomal dominant 52, 617796 (3), Autosomal dominant
<i>ASL</i>	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
<i>ASNS</i>	108370	Asparagine synthetase deficiency, 615574 (3), Autosomal recessive
<i>ASPA</i>	608034	Canavan disease, 271900 (3), Autosomal recessive
<i>ASPM</i>	605481	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive
<i>ASS1</i>	603470	Citrullinemia, 215700 (3), Autosomal recessive
<i>ASXL1</i>	612990	Bohring-Opitz syndrome, 605039 (3), Autosomal dominant; Myelodysplastic syndrome, somatic, 614286 (3)
<i>ASXL2</i>	612991	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
<i>ASXL3</i>	615115	Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant
<i>ATAD3A</i>	612316	Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant
<i>ATCAY</i>	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
<i>ATIC</i>	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
<i>ATN1</i>	607462	Dentatorubro-pallidoluysian atrophy, 125370 (3), Autosomal dominant
<i>ATP1A2</i>	182340	Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
<i>ATP1A3</i>	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant
<i>ATP2A2</i>	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
<i>ATP6AP2</i>	300556	Mental retardation, X-linked, syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive
<i>ATP6VOA2</i>	611716	Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive; Wrinkly skin syndrome, 278250 (3), Autosomal recessive
<i>ATP6V1B2</i>	606939	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant; Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant

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<i>ATP7A</i>	300011	Menkes disease, 309400 (3), X-linked recessive; Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
<i>ATP8A2</i>	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
<i>ATPAF2</i>	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
<i>ATR</i>	601215	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant; Seckel syndrome 1, 210600 (3), Autosomal recessive
<i>ATRX</i>	300032	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Mental retardation-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
<i>AUH</i>	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
<i>AUTS2</i>	607270	Mental retardation, autosomal dominant 26, 615834 (3), Autosomal dominant
<i>AVPR2</i>	300538	Diabetes insipidus, nephrogenic, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive
<i>B3GALNT2</i>	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
<i>B3GALT6</i>	615291	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive
<i>B3GLCT</i>	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
<i>B4GALNT1</i>	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
<i>B4GALT1</i>	137060	Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive
<i>B4GALT7</i>	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
<i>B4GAT1</i>	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
<i>B9D1</i>	614144	Joubert syndrome 27, 617120 (3), Autosomal recessive; ?Meckel syndrome 9, 614209 (3), Autosomal recessive
<i>B9D2</i>	611951	Joubert syndrome 34, 614175 (3), Autosomal recessive; ?Meckel syndrome 10, 614175 (3), Autosomal recessive
<i>BBS1</i>	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
<i>BBS10</i>	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
<i>BBS12</i>	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
<i>BBS2</i>	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
<i>BBS4</i>	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
<i>BBS5</i>	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
<i>BBS7</i>	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
<i>BBS9</i>	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
<i>BCAP31</i>	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
<i>BCKDHA</i>	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
<i>BCKDHB</i>	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
<i>BCL11A</i>	606557	Dias-Logan syndrome, 617101 (3), Autosomal dominant
<i>BCOR</i>	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant

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		No OMIM phenotype
<i>BCORL1</i>	300688	?Autism (Sanders (2012) Nature 485, 237), X-linked Autism spectrum disorder (Jiang (2013) Am J Hum Genet 93, 249), X-linked Intellectual disability, coarse face & hypotonia (Schuurs-Hoeijmakers (2013) J Med Genet 50, 802), X-linked
<i>BCS1L</i>	603647	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
<i>BLM</i>	604610	Bloom syndrome, 210900 (3), Autosomal recessive
<i>BOLA3</i>	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
<i>BRAF</i>	164757	Adenocarcinoma of lung, somatic, 211980 (3); Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Colorectal cancer, somatic (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Melanoma, malignant, somatic (3); Non-small cell lung cancer, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant
<i>BRAT1</i>	614506	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive
<i>BRF1</i>	604902	Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive
<i>BRPF1</i>	602410	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant
<i>BRWD3</i>	300553	Mental retardation, X-linked 93, 300659 (3), X-linked recessive
<i>BSCL2</i>	606158	Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant
<i>BTD</i>	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
<i>BUB1B</i>	602860	Colorectal cancer, somatic, 114500 (3); Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive; [Premature chromatid separation trait], 176430 (3), Autosomal dominant
		No OMIM phenotype
<i>C12orf4</i>	616082	Intellectual disability, autosomal recessive (Philips (2017) Clin Genet 91,100), Autosomal recessive Intellectual disability, ADHD and hypotonia (Alazami (2015) Cell Rep 10. 148), Autosomal recessive
<i>C12orf57</i>	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
<i>C12orf65</i>	613541	Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive; Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive
<i>C2CD3</i>	615944	?Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
<i>CA2</i>	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
<i>CA5A</i>	114761	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive
<i>CA8</i>	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3), Autosomal recessive
<i>CACNA1A</i>	601011	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
<i>CACNA1C</i>	114205	Brugada syndrome 3, 611875 (3); Timothy syndrome, 601005 (3), Autosomal dominant

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<i>CACNA1E</i>	601013	No OMIM phenotype ?Epileptic encephalopathy with infantile spasms (Helbig (2016) Genet Med Epub,Epub), Autosomal recessive/Autosomal dominant ?Autism (O'Roak (2012) Nature 485,246), Autosomal dominant
<i>CACNA2D1</i>	114204	No OMIM phenotype Brugada syndrome (Burashnikov (2010) Heart Rhythm 7,1872) Short QT syndrome (Templin (2011) Eur Heart J 32,1077), Autosomal dominant Histiocytoid cardiomyopathy (Cataldo (2014) Cardiol Young epub), Autosomal recessive West syndrome (Hino-Fukuyo (2015) Hum Genet 134,649)
<i>CACNA2D2</i>	607082	No OMIM phenotype Epileptic encephalopathy (Pippucci (2013) PLoS One 8,e82154), Autosomal recessive ?Schizophrenia (Purcell (2014) Nature 506, 185) Edvardson (2013) J Med Genet 50, 118 (EIEE), AR
<i>CACNG2</i>	602911	?Mental retardation, autosomal dominant 10, 614256 (3)
<i>CAD</i>	114010	Epileptic encephalopathy, early infantile, 50, 616457 (3), Autosomal recessive
<i>CAMK2A</i>	114078	Mental retardation, autosomal dominant 53, 617798 (3), Autosomal dominant
<i>CAMK2B</i>	607707	Mental retardation, autosomal dominant 54, 617799 (3), Autosomal dominant
<i>CAMTA1</i>	611501	Cerebellar ataxia, nonprogressive, with mental retardation, 614756 (3), Autosomal dominant
<i>CAPN10</i>	605286	{Diabetes mellitus, noninsulin-dependent 1}, 601283 (3)
<i>CARS2</i>	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
<i>CASK</i>	300172	FG syndrome 4, 300422 (3); Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked dominant; Mental retardation, with or without nystagmus, 300422 (3)
<i>CBL</i>	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
<i>CBS</i>	613381	Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive; Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive
<i>CC2D1A</i>	610055	Mental retardation, autosomal recessive 3, 608443 (3), Autosomal recessive
<i>CC2D2A</i>	612013	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive
<i>CCBE1</i>	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
<i>CCDC115</i>	613734	Congenital disorder of glycosylation, type Ilo, 616828 (3), Autosomal recessive
<i>CCDC174</i>	616735	Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive
<i>CCDC22</i>	300859	Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive
<i>CCDC78</i>	614666	?Centronuclear myopathy 4, 614807 (3), Autosomal dominant
<i>CCDC88A</i>	609736	?PEHO syndrome-like, 617507 (3), Autosomal recessive
<i>CCDC88C</i>	611204	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 (3), Autosomal recessive; ?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant
<i>CCND2</i>	123833	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant
<i>CDH15</i>	114019	Mental retardation, autosomal dominant 3, 612580 (3)

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<i>CDK10</i>	603464	Al Kaissi syndrome, 617694 (3), Autosomal recessive
<i>CDK13</i>	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant
<i>CDK5RAP2</i>	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
<i>CDK6</i>	603368	?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive
<i>CDKL5</i>	300203	Epileptic encephalopathy, early infantile, 2, 300672 (3), X-linked dominant
<i>CDKN1C</i>	600856	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; IMAGE syndrome, 614732 (3), Autosomal dominant
<i>CDON</i>	608707	Holoprosencephaly 11, 614226 (3), Autosomal dominant, Isolated cases
<i>CENPF</i>	600236	Stromme syndrome, 243605 (3), Autosomal recessive
<i>CENPJ</i>	609279	Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive
<i>CEP104</i>	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive
<i>CEP120</i>	613446	Joubert syndrome 31, 617761 (3), Autosomal recessive; Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive
<i>CEP135</i>	611423	Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive
<i>CEP152</i>	613529	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
<i>CEP290</i>	610142	?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
<i>CEP41</i>	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
<i>CEP63</i>	614724	?Seckel syndrome 6, 614728 (3), Autosomal recessive
<i>CEP83</i>	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
<i>CHAMP1</i>	616327	Mental retardation, autosomal dominant 40, 616579 (3), Autosomal dominant
<i>CHD1</i>	602118	Pilarowski-Bjornsson syndrome, 617682 (3), Autosomal dominant
<i>CHD2</i>	602119	Epileptic encephalopathy, childhood-onset, 615369 (3), Autosomal dominant
<i>CHD4</i>	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
<i>CHD7</i>	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
<i>CHD8</i>	610528	{Autism, susceptibility to, 18}, 615032 (3), Autosomal dominant
<i>CHKB</i>	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
<i>CHRNA2</i>	118502	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant
<i>CHRNA4</i>	118504	Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant; {Nicotine addiction, susceptibility to}, 188890 (3)
<i>CHRN2</i>	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
<i>CIC</i>	612082	Mental retardation, autosomal dominant 45, 617600 (3), Autosomal dominant
<i>CIT</i>	605629	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive
<i>CKAP2L</i>	616174	Filippi syndrome, 272440 (3), Autosomal recessive
<i>CLCN4</i>	302910	Mental retardation, X-linked 49/15, 300114 (3), X-linked recessive



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<i>CLCNKB</i>	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
<i>CLDN16</i>	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
<i>CLDN19</i>	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
<i>CLIC2</i>	300138	?Mental retardation, X-linked, syndromic 32, 300886 (3), X-linked recessive
<i>CLN3</i>	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
<i>CLN5</i>	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
<i>CLN6</i>	606725	Ceroid lipofuscinosis, neuronal, 6, 601780 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive
<i>CLN8</i>	607837	Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive
<i>CLP1</i>	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
<i>CLPB</i>	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive
<i>CLTC</i>	118955	Mental retardation, autosomal dominant 56, 617854 (3)
<i>CNKSRR2</i>	300724	Mental retardation, X-linked, syndromic, Houge type, 301008 (3), X-linked
<i>CNNM2</i>	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation, 616418 (3), Autosomal recessive, Autosomal dominant
<i>CNTN2</i>	190197	?Epilepsy, myoclonic, familial adult, 5, 615400 (3), Autosomal recessive
<i>CNTNAP2</i>	604569	{Autism susceptibility 15}, 612100 (3); Cortical dysplasia-focal epilepsy syndrome, 610042 (3), Autosomal recessive; Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive
<i>COASY</i>	609855	Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
<i>COG1</i>	606973	Congenital disorder of glycosylation, type IIg, 611209 (3)
<i>COG4</i>	606976	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive
<i>COG5</i>	606821	Congenital disorder of glycosylation, type IIi, 613612 (3)
<i>COG6</i>	606977	Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive; Shaheen syndrome, 615328 (3), Autosomal recessive
<i>COG7</i>	606978	Congenital disorder of glycosylation, type IIe, 608779 (3)
<i>COG8</i>	606979	Congenital disorder of glycosylation, type IIh, 611182 (3)
<i>COL4A1</i>	120130	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; Schizencephaly, 269160 (3)
<i>COL4A2</i>	120090	{Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 2, 614483 (3), Autosomal dominant
<i>COL4A3BP</i>	604677	Mental retardation, autosomal dominant 34, 616351 (3), Autosomal dominant
<i>COLEC11</i>	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
<i>COQ2</i>	609825	Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive; {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant
<i>COQ4</i>	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive

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<i>COQ8A</i>	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
<i>COQ9</i>	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
<i>COX10</i>	602125	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>COX15</i>	603646	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 (3), Autosomal recessive; Leigh syndrome due to cytochrome c oxidase deficiency, 256000 (3), Autosomal recessive, Mitochondrial
<i>COX6B1</i>	124089	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>CPA6</i>	609562	Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal recessive, Autosomal dominant; Febrile seizures, familial, 11, 614418 (3), Autosomal recessive
<i>CPLANE1</i> ( <i>C5orf42</i> )	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
<i>CPS1</i>	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3); {Venoocclusive disease after bone marrow transplantation} (3)
<i>CPT2</i>	600650	CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
<i>CRADD</i>	603454	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive
<i>CRBN</i>	609262	Mental retardation, autosomal recessive 2, 607417 (3), Autosomal recessive
<i>CREBBP</i>	600140	Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
<i>CRLF1</i>	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
<i>CSNK2A1</i>	115440	Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant
<i>CSPP1</i>	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
<i>CSTB</i>	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
<i>CTBP1</i>	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
<i>CTCF</i>	604167	Mental retardation, autosomal dominant 21, 615502 (3), Autosomal dominant
<i>CTDP1</i>	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
<i>CTNNB1</i>	116806	Colorectal cancer, somatic, 114500 (3); Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; 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)
<i>CTNND1</i>	601045	Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant No OMIM phenotype
<i>CTNND2</i>	604275	Autism (Turner (2015) Nature 520,51), Autosomal dominant Intellectual disability (Hofmeister (2015) J Med Genet 52,111), Autosomal dominant
<i>CTSA</i>	613111	Galactosialidosis, 256540 (3), Autosomal recessive
<i>CTSD</i>	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
<i>CTSF</i>	603539	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362 (3), Autosomal recessive
<i>CUBN</i>	602997	Megaloblastic anemia-1, Finnish type, 261100 (3), Autosomal recessive
<i>CUL4B</i>	300304	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 (3), X-linked recessive
<i>CUX2</i>	610648	Epileptic encephalopathy, early infantile, 67, 618141 (3), Autosomal dominant

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<i>CWF19L1</i>	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
<i>CYB5R3</i>	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
<i>CYP27A1</i>	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<i>CYP2U1</i>	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
<i>D2HGDH</i>	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
<i>DAG1</i>	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive
<i>DARS2</i>	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
<i>DBT</i>	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
<i>DCAF17</i>	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
<i>DCC</i>	120470	Colorectal cancer, somatic, 114500 (3); Esophageal carcinoma, somatic, 133239 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive; Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant
<i>DCHS1</i>	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
<i>DCPS</i>	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive
<i>DCX</i>	300121	Lissencephaly, X-linked, 300067 (3), X-linked; Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked
<i>DDC</i>	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
<i>DDHD2</i>	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
<i>DDX11</i>	601150	Warsaw breakage syndrome, 613398 (3), Autosomal recessive
<i>DDX3X</i>	300160	Mental retardation, X-linked 102, 300958 (3), X-linked recessive, X-linked dominant
<i>DEAF1</i>	602635	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 (3), Autosomal recessive; Mental retardation, autosomal dominant 24, 615828 (3), Autosomal dominant
<i>DENND5A</i>	617278	Epileptic encephalopathy, early infantile, 49, 617281 (3), Autosomal recessive
<i>DEPDC5</i>	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant
<i>DHCR24</i>	606418	Desmosterolosis, 602398 (3), Autosomal recessive
<i>DHCR7</i>	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
<i>DHFR</i>	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
<i>DHTKD1</i>	614984	2-aminoadipic 2-oxoadipic aciduria, 204750 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant
<i>DHX30</i>	616423	Neurodevelopmental disorder with severe motor impairment and absent language, 617804 (3), Autosomal dominant
<i>DIAPH1</i>	602121	Deafness, autosomal dominant 1, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
<i>DIP2B</i>	611379	Mental retardation, FRA12A type, 136630 (3), Autosomal dominant
<i>DKC1</i>	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
<i>DLAT</i>	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
<i>DLD</i>	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<i>DLG3</i>	300189	Mental retardation, X-linked 90, 300850 (3), X-linked recessive

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No OMIM phenotype

<i>DLG4</i>	602887	Autism spectrum disorder (An (2014) Transl Psychiatry 4,e394), Autosomal dominant Intellectual disability with marfanoid features (Moutton (2018), Clin Genet. 93(6)), Autosomal dominant
<i>DMD</i>	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
<i>DMPK</i>	605377	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
<i>DNAJC12</i>	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
<i>DNAJC19</i>	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
<i>DNAJC5</i>	611203	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3), Autosomal dominant
<i>DNM1</i>	602377	Epileptic encephalopathy, early infantile, 31, 616346 (3), Autosomal dominant
<i>DNMT3A</i>	602769	Acute myeloid leukemia, somatic, 601626 (3); Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant
<i>DNMT3B</i>	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive
<i>DOCK6</i>	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
<i>DOCK7</i>	615730	Epileptic encephalopathy, early infantile, 23, 615859 (3), Autosomal recessive
<i>DOLK</i>	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
<i>DONSON</i>	611428	Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive; Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive
<i>DPAGT1</i>	191350	Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive
<i>DPH1</i>	603527	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901 (3), Autosomal recessive
<i>DPM1</i>	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
<i>DPM2</i>	603564	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive
<i>DPP6</i>	126141	Mental retardation, autosomal dominant 33, 616311 (3); {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant
<i>DPYD</i>	612779	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
<i>DPYS</i>	613326	Dihydropyrimidinuria, 222748 (3), Autosomal recessive
<i>DYM</i>	607461	Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive; Smith-McCort dysplasia, 607326 (3), Autosomal recessive
<i>DYNC1H1</i>	600112	Charcot-Marie-Tooth disease, axonal, type 20, 614228 (3), Autosomal dominant; Mental retardation, autosomal dominant 13, 614563 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant
<i>DYRK1A</i>	600855	Mental retardation, autosomal dominant 7, 614104 (3), Autosomal dominant
<i>EBF3</i>	607407	Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant
<i>EBP</i>	300205	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
<i>ECHS1</i>	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
<i>EDC3</i>	609842	?Mental retardation, autosomal recessive 50, 616460 (3), Autosomal recessive
<i>EED</i>	605984	Cohen-Gibson syndrome, 617561 (3), Autosomal dominant

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<i>EEF1A2</i>	602959	Epileptic encephalopathy, early infantile, 33, 616409 (3), Autosomal dominant; Mental retardation, autosomal dominant 38, 616393 (3), Autosomal dominant
<i>EFTUD2</i>	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
<i>EGF</i>	131530	Hypomagnesemia 4, renal, 611718 (3)
<i>EHMT1</i>	607001	Kleefstra syndrome 1, 610253 (3), Autosomal dominant
<i>EIF2AK3</i>	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
<i>EIF2S3</i>	300161	MEHMO syndrome, 300148 (3), X-linked recessive
<i>EIF4A3</i>	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
<i>EIF4G1</i>	600495	{Parkinson disease 18}, 614251 (3), Autosomal dominant
<i>ELAC2</i>	605367	Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive; {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3)
<i>ELOVL4</i>	605512	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive; Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant
<i>ELP2</i>	616054	Mental retardation, autosomal recessive 58, 617270 (3), Autosomal recessive
<i>EMC1</i>	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
<i>EMX2</i>	600035	Schizencephaly, 269160 (3)
<i>ENTPD1</i>	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
<i>EP300</i>	602700	Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
<i>EPB41L1</i>	602879	?Mental retardation, autosomal dominant 11, 614257 (3)
<i>EPG5</i>	615068	Vici syndrome, 242840 (3), Autosomal recessive
<i>EPM2A</i>	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
<i>EPRS</i>	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
<i>ERCC2</i>	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
<i>ERCC3</i>	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
<i>ERCC5</i>	133530	Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
<i>ERCC6</i>	609413	Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; {Lung cancer, susceptibility to}, 211980 (3), Autosomal 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
<i>ERCC8</i>	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
<i>ERLIN2</i>	611605	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
<i>ESCO2</i>	609353	Roberts syndrome, 268300 (3), Autosomal recessive; SC phocomelia syndrome, 269000 (3), Autosomal recessive
<i>ETFB</i>	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
<i>ETHE1</i>	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
<i>EXOSC2</i>	602238	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive
<i>EXOSC3</i>	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive

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<i>EZH2</i>	601573	Weaver syndrome, 277590 (3), Autosomal dominant
<i>FA2H</i>	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
<i>FAM126A</i>	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
<i>FANCD2</i>	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
<i>FAR1</i>	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive
<i>FARS2</i>	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
<i>FAT4</i>	612411	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive; Van Maldergem syndrome 2, 615546 (3), Autosomal recessive
<i>FBXL4</i>	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
<i>FBXO11</i>	607871	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 (3), Autosomal Dominant
<i>FBXO31</i>	609102	?Mental retardation, autosomal recessive 45, 615979 (3), Autosomal recessive
<i>FGD1</i>	300546	Aarskog-Scott syndrome, 305400 (3), X-linked recessive; Mental retardation, X-linked syndromic 16, 305400 (3), X-linked recessive
<i>FGF12</i>	601513	Epileptic encephalopathy, early infantile, 47, 617166 (3), Autosomal dominant
<i>FGF14</i>	601515	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant
<i>FGFR1</i>	136350	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; Osteoglyphonic dysplasia, 166250 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant
<i>FGFR2</i>	176943	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Crouzon syndrome, 123500 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3)
<i>FGFR3</i>	134934	Achondroplasia, 100800 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Muenke syndrome, 602849 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); SADDAN, 616482 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant
<i>FH</i>	136850	Fumarase deficiency, 606812 (3), Autosomal recessive; Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant
<i>FIBP</i>	608296	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive

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<i>FKRP</i>	606596	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive
<i>FKTN</i>	607440	Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive
<i>FLNA</i>	300017	Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked recessive; Congenital short bowel syndrome, 300048 (3), X-linked recessive; ?FG syndrome 2, 300321 (3), X-linked; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive; Heterotopia, periventricular, 300049 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant
<i>FLVCR1</i>	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
<i>FMN2</i>	606373	Mental retardation, autosomal recessive 47, 616193 (3), Autosomal recessive
<i>FMR1</i>	309550	Fragile X syndrome, 300624 (3), X-linked dominant; Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked
<i>FOLR1</i>	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
<i>FOXG1</i>	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
<i>FOXP1</i>	605515	Mental retardation with language impairment and with or without autistic features, 613670 (3), Autosomal dominant
<i>FOXP2</i>	605317	Speech-language disorder-1, 602081 (3), Autosomal dominant
<i>FOXRED1</i>	613622	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
<i>FRAS1</i>	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
<i>FREM2</i>	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive
<i>FRMD4A</i>	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
<i>FRMPD4</i>	300838	Mental retardation, X-linked 104, 300983 (3), X-linked recessive
<i>FRRS1L</i>	604574	Epileptic encephalopathy, early infantile, 37, 616981 (3), Autosomal recessive
<i>FTCD</i>	606806	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
<i>FTO</i>	610966	Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive
<i>FTSJ1</i>	300499	Mental retardation, X-linked 9/44, 309549 (3), X-linked recessive
<i>FUCA1</i>	612280	Fucosidosis, 230000 (3), Autosomal recessive
<i>FXYP2</i>	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
<i>GABBR2</i>	607340	Epileptic encephalopathy, early infantile, 59, 617904 (3), Autosomal dominant; Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant; {Nicotine dependence, protection against}, 188890 (3); {Nicotine dependence, susceptibility to}, 188890 (3)
<i>GABRA1</i>	137160	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3); {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Epileptic encephalopathy, early infantile, 19, 615744 (3), Autosomal dominant

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<i>GABRB1</i>	137190	Epileptic encephalopathy, early infantile, 45, 617153 (3), Autosomal dominant
<i>GABRB3</i>	137192	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Epileptic encephalopathy, early infantile, 43, 617113 (3), Autosomal dominant
<i>GABRG2</i>	137164	{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
<i>GAD1</i>	605363	?Cerebral palsy, spastic quadriplegic, 1, 603513 (3), Autosomal recessive
<i>GALE</i>	606953	Galactose epimerase deficiency, 230350 (3), Autosomal recessive
<i>GALT</i>	606999	Galactosemia, 230400 (3), Autosomal recessive
<i>GAMT</i>	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
<i>GATAD2B</i>	614998	Mental retardation, autosomal dominant 18, 615074 (3), Autosomal dominant
<i>GATM</i>	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive
<i>GCDH</i>	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
<i>GCH1</i>	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
<i>GCK</i>	138079	Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 606176 (3), Autosomal recessive, Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; MODY, type II, 125851 (3), Autosomal dominant
<i>GCSH</i>	238330	?Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>GDI1</i>	300104	Mental retardation, X-linked 41, 300849 (3), X-linked dominant
<i>GFAP</i>	137780	Alexander disease, 203450 (3), Autosomal dominant No OMIM phenotype
<i>GFM2</i>	606544	Leigh syndrome with arthrogryposis multiplex congenita (Fukumura (2015) J Hum Genet 60,509), Autosomal recessive Wolcott-Rallison syndrome (Dixon-Salazar (2012) Sci Transl Med 4,138ra78) {Atorvastatin sensitivity} (Callegari (2012) PLoS Genet 8,e1002755)
<i>GJA1</i>	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant
<i>GJB1</i>	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
<i>GJC2</i>	608803	Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive; Lymphedema, hereditary, IC, 613480 (3), Autosomal dominant; Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive
<i>GK</i>	300474	Glycerol kinase deficiency, 307030 (3), X-linked recessive
<i>GLB1</i>	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive
<i>GLDC</i>	238300	Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>GLI2</i>	165230	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant



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<i>GLI3</i>	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; {Hypothalamic hamartomas, somatic}, 241800 (3); Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
<i>GLRA1</i>	138491	Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant
<i>GLRB</i>	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
<i>GLUD1</i>	138130	Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant
<i>GLYCTK</i>	610516	D-glyceric aciduria, 220120 (3), Autosomal recessive
<i>GM2A</i>	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
<i>GMPPA</i>	615495	Alacrima, achalasia, and mental retardation syndrome, 615510 (3), Autosomal recessive
<i>GMPPB</i>	615320	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive
<i>GNAO1</i>	139311	Epileptic encephalopathy, early infantile, 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
<i>GNAS</i>	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Isolated cases; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
<i>GNB1</i>	139380	Leukemia, acute lymphoblastic, somatic, 613065 (3); Mental retardation, autosomal dominant 42, 616973 (3), Autosomal dominant
<i>GNB5</i>	604447	Intellectual developmental disorder with cardiac arrhythmia, 617173 (3), Autosomal recessive; Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 (3), Autosomal recessive
<i>GNPAT</i>	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
<i>GNPTAB</i>	607840	Mucopolidosis II alpha/beta, 252500 (3), Autosomal recessive; Mucopolidosis III alpha/beta, 252600 (3), Autosomal recessive
<i>GNS</i>	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
<i>GOSR2</i>	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
<i>GPC3</i>	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
<i>GPHN</i>	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
<i>GPT2</i>	138210	Mental retardation, autosomal recessive 49, 616281 (3), Autosomal recessive
<i>GRIA3</i>	305915	Mental retardation, X-linked 94, 300699 (3), X-linked recessive
<i>GRID2</i>	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
<i>GRIK2</i>	138244	Mental retardation, autosomal recessive, 6, 611092 (3), Autosomal recessive
<i>GRIN1</i>	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive
<i>GRIN2A</i>	138253	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570 (3), Autosomal dominant

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<i>GRIN2B</i>	138252	Epileptic encephalopathy, early infantile, 27, 616139 (3), Autosomal dominant; Mental retardation, autosomal dominant 6, 613970 (3), Autosomal dominant
<i>GRIN2D</i>	602717	Epileptic encephalopathy, early infantile, 46, 617162 (3), Autosomal dominant
<i>GRIP1</i>	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
<i>GRM1</i>	604473	Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive
<i>GRN</i>	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
<i>GSS</i>	601002	Glutathione synthetase deficiency, 266130 (3), Autosomal recessive; Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive
<i>GTF2H5</i>	608780	Trichothiodystrophy 3, photosensitive, 616395 (3)
<i>GTPBP3</i>	608536	Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive
<i>GUSB</i>	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
<i>HACE1</i>	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
<i>HADH</i>	601609	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive
<i>HAX1</i>	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
<i>HCCS</i>	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
<i>HCFC1</i>	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cbIX type ), 309541 (3), X-linked recessive
<i>HCN1</i>	602780	Epileptic encephalopathy, early infantile, 24, 615871 (3), Autosomal dominant
<i>HDAC6</i>	300272	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant
<i>HDAC8</i>	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
<i>HECW2</i>	617245	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 (3), Autosomal dominant
<i>HEPACAM</i>	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926 (3), Autosomal dominant
<i>HERC1</i>	605109	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive
<i>HERC2</i>	605837	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
<i>HESX1</i>	601802	Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant; Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant
<i>HEXA</i>	606869	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
<i>HEXB</i>	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive

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<i>HGSNAT</i>	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
<i>HIVEP2</i>	143054	Mental retardation, autosomal dominant 43, 616977 (3), Autosomal dominant
<i>HLCS</i>	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
<i>HMGCL</i>	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
<i>HNMT</i>	605238	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; Mental retardation, autosomal recessive 51, 616739 (3), Autosomal recessive
<i>HNRNPH2</i>	300610	Mental retardation, X-linked, syndromic, Bain type, 300986 (3), X-linked dominant
<i>HNRNPK</i>	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
<i>HNRNPU</i>	602869	Epileptic encephalopathy, early infantile, 54, 617391 (3), Autosomal dominant
<i>HOXA1</i>	142955	Athabaskan brainstem dysgenesis syndrome, 601536 (3); Bosley-Salih-Alorainy syndrome, 601536 (3)
<i>HPD</i>	609695	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
<i>HPRT1</i>	308000	HPRT-related gout, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
<i>HRAS</i>	190020	{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)
<i>HSD17B10</i>	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
<i>HSD17B4</i>	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
<i>HSPA9</i>	600548	Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant; Even-plus syndrome, 616854 (3), Autosomal recessive
<i>HSPD1</i>	118190	Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive; Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant
<i>HUWE1</i>	300697	Mental retardation, X-linked syndromic, Turner type, 300706 (3)
<i>IARS</i>	600709	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
<i>IARS2</i>	612801	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
<i>IDH2</i>	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
<i>IDS</i>	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
<i>IDUA</i>	252800	Mucopolysaccharidosis I <sub>h</sub> , 607014 (3), Autosomal recessive; Mucopolysaccharidosis I <sub>h/s</sub> , 607015 (3), Autosomal recessive; Mucopolysaccharidosis I <sub>s</sub> , 607016 (3), Autosomal recessive
<i>IER3IP1</i>	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
<i>IFIH1</i>	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
<i>IFT172</i>	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
<i>IGBP1</i>	300139	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 (3), X-linked recessive
<i>IGF1</i>	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive

## H9.1-OP2-B2: Genpanel Intellectual Disability Epilepsy v2, in voege op 01/01/2019

<i>IKBKG</i>	300248	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)
<i>IL1RAPL1</i>	300206	Mental retardation, X-linked 21/34, 300143 (3), X-linked recessive
<i>IMPA1</i>	602064	Mental retardation, autosomal recessive 59, 617323 (3), Autosomal recessive
<i>INPP5E</i>	613037	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
<i>INPP5K</i>	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
<i>IQSEC2</i>	300522	Mental retardation, X-linked 1/78, 309530 (3), X-linked dominant
<i>ISCA2</i>	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive
<i>ISPD</i>	614631	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive
<i>ITGA7</i>	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
<i>ITPA</i>	147520	Epileptic encephalopathy, early infantile, 35, 616647 (3), Autosomal recessive; [Inosine triphosphatase deficiency], 613850 (3)
<i>ITPR1</i>	147265	Gillespie syndrome, 206700 (3), Autosomal recessive, Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant
<i>IVD</i>	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
<i>JAG1</i>	601920	Alagille syndrome 1, 118450 (3), Autosomal dominant; ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>JAM3</i>	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
<i>JMJD1C</i>	604503	No OMIM phenotype ?Rett syndrome (Saez (2016) Genet Med 18,378), Autosomal dominant ?Congenital heart disease in 22q11.2 deletion syndrome patients (Guo (2015) Am J Hum Genet 97,869), Autosomal dominant ?Autism spectrum disorder (Saez (2016) Genet Med 18,378), Autosomal dominant ?Intellectual disability (Saez (2016) Genet Med 18,378), Autosomal dominant
<i>KALRN</i>	604605	{Coronary heart disease, susceptibility to, 5}, 608901 (3)
<i>KANK1</i>	607704	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
<i>KANSL1</i>	612452	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant
<i>KAT6A</i>	601408	Mental retardation, autosomal dominant 32, 616268 (3), Autosomal dominant
<i>KAT6B</i>	605880	Genitopatellar syndrome, 606170 (3), Autosomal dominant; SBBYSS syndrome, 603736 (3), Autosomal dominant
<i>KATNB1</i>	602703	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
<i>KCNA1</i>	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
<i>KCNA2</i>	176262	Epileptic encephalopathy, early infantile, 32, 616366 (3), Autosomal dominant
<i>KCNA4</i>	176266	No OMIM phenotype
<i>KCNB1</i>	600397	Abnormal striatum, congenital cataract and intellectual disability (Kaya (2016) J Med Genet 53,786), Autosomal recessive Epileptic encephalopathy, early infantile, 26, 616056 (3), Autosomal dominant

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<i>KCNC1</i>	176258	Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant
<i>KCNC3</i>	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
<i>KCNH1</i>	603305	Temple-Baraitser syndrome, 611816 (3), Autosomal dominant; Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant
<i>KCNJ10</i>	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
<i>KCNJ11</i>	600937	Diabetes mellitus, transient neonatal, 3, 610582 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Diabetes, permanent neonatal, with or without neurologic features, 606176 (3), Autosomal recessive, Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant
<i>KCNJ6</i>	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
<i>KCNK9</i>	605874	Birk-Barel mental retardation dysmorphism syndrome, 612292 (3)
<i>KCNMA1</i>	600150	?Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant
<i>KCNQ2</i>	602235	Epileptic encephalopathy, early infantile, 7, 613720 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant
<i>KCNQ3</i>	602232	Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant
<i>KCNQ5</i>	607357	Mental retardation, autosomal dominant 46, 617601 (3), Autosomal dominant
<i>KCNT1</i>	608167	Epilepsy, nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 14, 614959 (3), Autosomal dominant
<i>KCTD7</i>	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
<i>KDM1A</i>	609132	Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant
<i>KDM5C</i>	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
<i>KDM6A</i>	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
<i>KIAA0556</i>	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
<i>KIAA0586</i>	610178	Joubert syndrome 23, 616490 (3), Autosomal recessive; Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive
<i>KIAA1109</i>	611565	Alkuraya-Kucinkas syndrome, 617822 (3), Autosomal recessive
<i>KIDINS220</i>	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant
<i>KIF11</i>	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
<i>KIF1A</i>	601255	Mental retardation, autosomal dominant 9, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal recessive
<i>KIF1BP</i>	609367	Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive
<i>KIF2A</i>	602591	Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant
<i>KIF4A</i>	300521	?Mental retardation, X-linked 100, 300923 (3), X-linked recessive
<i>KIF5C</i>	604593	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant
<i>KIF7</i>	611254	Acrocaldosal syndrome, 200990 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive; ?Hydroletharus syndrome 2, 614120 (3), Autosomal recessive; Joubert syndrome 12, 200990 (3), Autosomal recessive

## H9.1-OP2-B2: Genpanel Intellectual Disability Epilepsy v2, in voege op 01/01/2019

<i>KIRREL3</i>	607761	Mental retardation, autosomal dominant 4, 612581 (3)
<i>KLHL15</i>	300980	Mental retardation, X-linked 103, 300982 (3), X-linked recessive
<i>KMT2A</i>	159555	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 (2), Autosomal dominant; Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
<i>KMT2B</i>	606834	Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
<i>KMT2C</i>	606833	Kleefstra syndrome 2, 617768 (3), Autosomal dominant
<i>KMT2D</i>	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
<i>KMT5B</i>	610881	Mental retardation, autosomal dominant 51, 617788 (3), Autosomal dominant
<i>KNL1</i>	609173	Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive
<i>KPTN</i>	615620	Mental retardation, autosomal recessive 41, 615637 (3), Autosomal recessive
<i>KRAS</i>	190070	Arteriovenous malformation of the brain, somatic, 108010 (3); Bladder cancer, somatic, 109800 (3); Breast cancer, somatic, 114480 (3); Cardiofaciocutaneous syndrome 2, 615278 (3); Gastric cancer, somatic, 137215 (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant; Lung cancer, somatic, 211980 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; Pancreatic carcinoma, somatic, 260350 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)
<i>L1CAM</i>	308840	CRASH syndrome, 303350 (3), X-linked recessive; Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive; Hydrocephalus with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3), X-linked recessive; MASA syndrome, 303350 (3), X-linked recessive
<i>L2HGDH</i>	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
<i>LAMA1</i>	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
<i>LAMA2</i>	156225	Muscular dystrophy, congenital merosin-deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3), Autosomal recessive
<i>LAMB1</i>	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
<i>LAMC3</i>	604349	Cortical malformations, occipital, 614115 (3), Autosomal recessive
<i>LAMP2</i>	309060	Danon disease, 300257 (3), X-linked dominant
<i>LARGE1</i>	603590	Muscular dystrophy-dystroglycanopathy (congenital with 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
<i>LARP7</i>	612026	Alazami syndrome, 615071 (3), Autosomal recessive
<i>LAS1L</i>	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
<i>LGI1</i>	604619	Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant
<i>LIAS</i>	607031	Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive
<i>LIG4</i>	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
<i>LINGO1</i>	609791	No OMIM phenotype
<i>LINS1</i>	610350	Intellectual disability, microcephaly, speech and motor delay (Ansar (2018), Genet Med 20(7)), Autosomal recessive
<i>LINS1</i>	610350	Mental retardation, autosomal recessive 27, 614340 (3), Autosomal recessive
<i>LMAN2L</i>	609552	?Mental retardation, autosomal recessive, 52, 616887 (3), Autosomal recessive
<i>LONP1</i>	605490	CODAS syndrome, 600373 (3), Autosomal recessive

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<i>LRP2</i>	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
<i>LRPPRC</i>	607544	Leigh syndrome, French-Canadian type, 220111 (3), Autosomal recessive
<i>LZTFL1</i>	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
<i>LZTR1</i>	600574	Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
<i>MAB21L2</i>	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant
<i>MAF</i>	177075	Ayme-Gripp syndrome, 601088 (3), Autosomal dominant; Cataract 21, multiple types, 610202 (3), Autosomal dominant
<i>MAG</i>	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
<i>MAGEL2</i>	605283	Schaaf-Yang syndrome, 615547 (3), Autosomal dominant
<i>MAGT1</i>	300715	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3)
<i>MAN1B1</i>	604346	Mental retardation, autosomal recessive 15, 614202 (3), Autosomal recessive
<i>MAN2B1</i>	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<i>MANBA</i>	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
<i>MAOA</i>	309850	{Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive
<i>MAP2K1</i>	176872	Cardiofaciocutaneous syndrome 3, 615279 (3)
<i>MAP2K2</i>	601263	Cardiofaciocutaneous syndrome 4, 615280 (3)
<i>MAPRE2</i>	605789	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant
<i>MASP1</i>	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
<i>MAT1A</i>	610550	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal recessive, Autosomal dominant; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal recessive, Autosomal dominant
<i>MBD5</i>	611472	Mental retardation, autosomal dominant 1, 156200 (3), Autosomal dominant
<i>MBOAT7</i>	606048	Mental retardation, autosomal recessive 57, 617188 (3), Autosomal recessive
<i>MBTPS2</i>	300294	IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
<i>MCCC1</i>	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive
<i>MCCC2</i>	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive
<i>MCOLN1</i>	605248	Mucopolipidosis IV, 252650 (3), Autosomal recessive
<i>MCPH1</i>	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
<i>MDH2</i>	154100	Epileptic encephalopathy, early infantile, 51, 617339 (3), Autosomal recessive
<i>MECP2</i>	300005	{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
<i>MECR</i>	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive
<i>MED12</i>	300188	Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive

H9.1-OP2-B2: Genpanel Intellectual Disability Epilepsy v2, in voege op 01/01/2019

<i>MED13L</i>	608771	Mental retardation and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant; Transposition of the great arteries, dextro-looped 1, 608808 (3), Autosomal dominant
<i>MED17</i>	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive
<i>MED23</i>	605042	Mental retardation, autosomal recessive 18, 614249 (3), Autosomal recessive
<i>MED25</i>	610197	Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive
<i>MEF2C</i>	600662	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 (3), Autosomal dominant
<i>MEGF8</i>	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
<i>MEIS2</i>	601740	Cleft palate, cardiac defects, and mental retardation, 600987 (3), Autosomal dominant
<i>METTL23</i>	615262	Mental retardation, autosomal recessive 44, 615942 (3), Autosomal recessive
<i>MFF</i>	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive
<i>MFSD2A</i>	614397	Microcephaly 15, primary, autosomal recessive, 616486 (3), Autosomal recessive
<i>MFSD8</i>	611124	Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive; Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive
<i>MGAT2</i>	602616	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive
<i>MICU1</i>	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
<i>MID1</i>	300552	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
<i>MID2</i>	300204	?Mental retardation, X-linked 101, 300928 (3), X-linked recessive
<i>MKKS</i>	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
<i>MLC1</i>	605908	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive
<i>MLYCD</i>	606761	Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive
<i>MMAA</i>	607481	Methylmalonic aciduria, vitamin B12-responsive, 251100 (3), Autosomal recessive
<i>MMAB</i>	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110 (3), Autosomal recessive
<i>MMACHC</i>	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
<i>MMADHC</i>	611935	Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive
<i>MOCS1</i>	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
<i>MOCS2</i>	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
<i>MOGS</i>	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
<i>MPDU1</i>	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
<i>MPDZ</i>	603785	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219 (3), Autosomal recessive
<i>MPLKIP</i>	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
<i>MRPL3</i>	607118	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
<i>MRPS22</i>	605810	Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive



H9.1-OP2-B2: Genpanel Intellectual Disability Epilepsy v2, in voege op 01/01/2019

<i>MSL3</i>	300609	X-linked syndrome characterized by global developmental delay, progressive gait disturbance and facial dysmorphism (Basilicata 2018 Oct, Nat Genet 50(10)), X-linked dominant and recessive
<i>MTFMT</i>	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive
<i>MTHFR</i>	607093	Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Vascular disease, susceptibility to} (3)
<i>MTOR</i>	601231	Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant
<i>MTR</i>	156570	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
<i>MTRR</i>	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
<i>MUT</i>	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
<i>MVK</i>	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Mevalonic aciduria, 610377 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant
<i>MYCN</i>	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant
<i>MYH9</i>	160775	Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant; Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 (3), Autosomal dominant
<i>MYO5A</i>	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
<i>MYT1L</i>	613084	Mental retardation, autosomal dominant 39, 616521 (3), Autosomal dominant
<i>NAA10</i>	300013	?Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
<i>NAA15</i>	608000	Mental retardation, autosomal dominant 50, 617787 (3), Autosomal dominant
<i>NACC1</i>	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant
<i>NAGA</i>	104170	Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type I, 609241 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
<i>NAGLU</i>	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
<i>NALCN</i>	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant; Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive
<i>NANS</i>	605202	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive
<i>NARS2</i>	612803	Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive
<i>NBN</i>	602667	Aplastic anemia, 609135 (3); Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
<i>NDE1</i>	609449	Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive; ?Microhydranencephaly, 605013 (3), Autosomal recessive
<i>NDP</i>	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3); Norrie disease, 310600 (3), X-linked recessive
<i>NDST1</i>	600853	Mental retardation, autosomal recessive 46, 616116 (3), Autosomal recessive
<i>NDUFA1</i>	300078	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFA11</i>	612638	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial

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<i>NDUFA12</i>	614530	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000 (3), Autosomal recessive, Mitochondrial
<i>NDUFA2</i>	602137	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial
<i>NDUFAF1</i>	606934	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFAF2</i>	609653	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFAF3</i>	612911	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFAF4</i>	611776	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFAF5</i>	612360	Mitochondrial complex 1 deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFB3</i>	603839	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFB9</i>	601445	?Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFS1</i>	157655	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFS2</i>	602985	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFS3</i>	603846	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
<i>NDUFS4</i>	602694	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFS6</i>	603848	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFS7</i>	601825	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial
<i>NDUFS8</i>	602141	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial
<i>NDUFV1</i>	161015	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NDUFV2</i>	600532	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NECAP1</i>	611623	?Epileptic encephalopathy, early infantile, 21, 615833 (3), Autosomal recessive
<i>NECTIN1</i>	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
<i>NEDD4L</i>	606384	Periventricular nodular heterotopia 7, 617201 (3), Autosomal dominant
<i>NEU1</i>	608272	Sialidosis, type I, 256550 (3), Autosomal recessive; Sialidosis, type II, 256550 (3), Autosomal recessive
<i>NEXMIF</i>	300524	Mental retardation, X-linked 98, 300912 (3), X-linked dominant
<i>NF1</i>	613113	Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant; Watson syndrome, 193520 (3), Autosomal dominant
<i>NFIA</i>	600727	Brain malformations with or without urinary tract defects, 613735 (3), Isolated cases
<i>NFIX</i>	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Sotos syndrome 2, 614753 (3), Autosomal dominant
<i>NGLY1</i>	610661	Congenital disorder of deglycosylation, 615273 (3), Autosomal recessive
<i>NHLRC1</i>	608072	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
<i>NHS</i>	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
<i>NIPBL</i>	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
<i>NKX2-1</i>	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant
<i>NLGN3</i>	300336	{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

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<i>NLGN4X</i>	300427	{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
<i>NLRP3</i>	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
<i>NONO</i>	300084	Mental retardation, X-linked, syndromic 34, 300967 (3), X-linked
<i>NPC1</i>	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
<i>NPC2</i>	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
<i>NPHP1</i>	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
<i>NPRL2</i>	607072	Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant
<i>NPRL3</i>	600928	Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant
<i>NR2F1</i>	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
<i>NRAS</i>	164790	Colorectal cancer, somatic, 114500 (3); Epidermal nevus, somatic, 162900 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Neurocutaneous melanosis, somatic, 249400 (3); Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3)
<i>NRXN1</i>	600565	Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3)
<i>NSD1</i>	606681	Leukemia, acute myeloid, 601626 (1), Autosomal dominant; Sotos syndrome 1, 117550 (3), Autosomal dominant
<i>NSD2</i>	602952	No OMIM phenotype
<i>NSDHL</i>	300275	Intellectual disability and dysmorphic features (Lozier (2018), J Hum genet), Autosoma dominant
<i>NSUN2</i>	610916	CHILD syndrome, 308050 (3), X-linked dominant; CK syndrome, 300831 (3), X-linked recessive
<i>NTRK1</i>	191315	Mental retardation, autosomal recessive 5, 611091 (3), Autosomal recessive
<i>NUBPL</i>	613621	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive; Medullary thyroid carcinoma, familial, 155240 (3), Autosomal dominant
<i>NUP62</i>	605815	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
<i>NUS1</i>	610463	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
<i>OAT</i>	613349	?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive; Mental retardation, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant
<i>OCLN</i>	602876	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
<i>OCRL</i>	300535	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
<i>ODC1</i>	165640	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
<i>OFD1</i>	300170	{Colonic adenoma recurrence, reduced risk of}, 114500 (3), Autosomal dominant
<i>OGT</i>	300255	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
		Mental retardation, X-linked 106, 300997 (3), X-linked recessive

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<i>OPHN1</i>	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 (3), X-linked recessive
<i>ORC1</i>	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
<i>OSGEP</i>	610107	Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive
<i>OTC</i>	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked recessive
<i>OTUD6B</i>	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive
<i>PACS1</i>	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
<i>PAFAH1B1</i>	601545	Lissencephaly 1, 607432 (3), Isolated cases; Subcortical laminar heterotopia, 607432 (3), Isolated cases
<i>PAH</i>	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
<i>PAK3</i>	300142	Mental retardation, X-linked 30/47, 300558 (3), X-linked recessive
<i>PANK2</i>	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
<i>PAX1</i>	167411	?Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
<i>PAX6</i>	607108	Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3); Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Optic nerve hypoplasia, 165550 (3), Autosomal dominant
<i>PAX8</i>	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
<i>PBX1</i>	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
<i>PC</i>	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
<i>PCCA</i>	232000	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCCB</i>	232050	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCDH19</i>	300460	Epileptic encephalopathy, early infantile, 9, 300088 (3), X-linked
<i>PCGF2</i>	600346	No OMIM phenotype ?Developmental disorder (Fitzgerald (2015) Nature 519,223), Autosomal dominant
<i>PCLO</i>	604918	?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive
<i>PCNT</i>	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
<i>PDE4D</i>	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant; {Stroke, susceptibility to, 1}, 606799 (3)
<i>PDE6D</i>	602676	?Joubert syndrome 22, 615665 (3), Autosomal recessive
<i>PDHA1</i>	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
<i>PDHB</i>	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3)
<i>PDP1</i>	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
<i>PDSS1</i>	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
<i>PDSS2</i>	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive

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<i>PDX1</i>	600733	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; MODY, type IV, 606392 (3); Pancreatic agenesis 1, 260370 (3), Autosomal recessive
<i>PEPD</i>	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
<i>PET100</i>	614770	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>PEX1</i>	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive
<i>PEX10</i>	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
<i>PEX11B</i>	603867	?Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
<i>PEX12</i>	601758	Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive; Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive
<i>PEX13</i>	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
<i>PEX14</i>	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
<i>PEX16</i>	603360	Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive
<i>PEX19</i>	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
<i>PEX2</i>	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
<i>PEX26</i>	608666	Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive; Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive
<i>PEX3</i>	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
<i>PEX5</i>	600414	Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
<i>PEX6</i>	601498	Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant
<i>PEX7</i>	601757	Peroxisome biogenesis disorder 9B, 614879 (3); Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
<i>PGAP1</i>	611655	Mental retardation, autosomal recessive 42, 615802 (3), Autosomal recessive
<i>PGAP2</i>	615187	Hyperphosphatasia with mental retardation syndrome 3, 614207 (3), Autosomal recessive
<i>PGAP3</i>	611801	Hyperphosphatasia with mental retardation syndrome 4, 615716 (3), Autosomal recessive
<i>PGK1</i>	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
<i>PGM3</i>	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
<i>PHF6</i>	300414	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
<i>PHF8</i>	300560	Mental retardation syndrome, X-linked, Siderius type, 300263 (3), X-linked recessive
<i>PHGDH</i>	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
<i>PHIP</i>	612870	Developmental delay, intellectual disability, obesity, and dysmorphic features, 617991 (3), Autosomal dominant

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<i>PI4KA</i>	600286	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogyriposis, 616531 (3), Autosomal recessive
<i>PIGA</i>	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
<i>PIGC</i>	601730	Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive
<i>PIGG</i>	616918	Mental retardation, autosomal recessive 53, 616917 (3), Autosomal recessive
<i>PIGL</i>	605947	CHIME syndrome, 280000 (3), Autosomal recessive
<i>PIGN</i>	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive
<i>PIGO</i>	614730	Hyperphosphatasia with mental retardation syndrome 2, 614749 (3), Autosomal recessive
<i>PIGP</i>	605938	?Epileptic encephalopathy, early infantile, 55, 617599 (3), Autosomal recessive
<i>PIGT</i>	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive; ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation
<i>PIGV</i>	610274	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive
<i>PIGW</i>	610275	Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive
<i>PIGY</i>	610662	Hyperphosphatasia with mental retardation syndrome 6, 616809 (3), Autosomal recessive
<i>PIK3CA</i>	171834	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); Non-small cell lung cancer, somatic, 211980 (3); Ovarian cancer, somatic, 167000 (3)
<i>PIK3R2</i>	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant
<i>PLA2G6</i>	603604	Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive
<i>PLCB1</i>	607120	Epileptic encephalopathy, early infantile, 12, 613722 (3), Autosomal recessive
<i>PLK4</i>	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
<i>PLP1</i>	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
<i>PLPBP</i>	604436	Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive No OMIM phenotype
<i>PLXND1</i>	604282	Moebius syndrome (Tomas-Roca (2015) Nat Commun 6), Autosomal dominant Truncus arteriosus (Ta-Shma (2013) Am J Med Genet A 161,3115) {Diabetic nephropathy, association with} (McKnight (2009) Hugo J 3,77)
<i>PMM2</i>	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
<i>PMPCA</i>	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
<i>PNKP</i>	605610	Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
<i>PNP</i>	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
<i>PNPLA6</i>	603197	Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive

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<i>PNPO</i>	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
<i>POC1A</i>	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
<i>POC1B</i>	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
<i>POGZ</i>	614787	White-Sutton syndrome, 616364 (3), Autosomal dominant
<i>POLG</i>	174763	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
<i>POLR3A</i>	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
<i>POLR3B</i>	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive
<i>POMGNT1</i>	606822	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive
<i>POMGNT2</i>	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive
<i>POMK</i>	615247	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive; ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive
<i>POMT1</i>	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive
<i>POMT2</i>	607439	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive
<i>PORCN</i>	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
<i>POU1F1</i>	173110	Pituitary hormone deficiency, combined, 1, 613038 (3), Autosomal recessive, Autosomal dominant
<i>POU3F3</i>	602480	No OMIM phenotype ?Intellectual disability (Dheedene (2014) Mol Syndromol 5,32), Autosomal dominant
<i>PPM1D</i>	605100	Breast cancer, somatic, 114480 (3); Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450 (3), Autosomal dominant
<i>PPOX</i>	600923	Porphyria variegata, 176200 (3), Autosomal dominant
<i>PPP1CB</i>	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
<i>PPP1R15B</i>	613257	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
<i>PPP2R1A</i>	605983	Mental retardation, autosomal dominant 36, 616362 (3), Autosomal dominant
<i>PPP2R5D</i>	601646	Mental retardation, autosomal dominant 35, 616355 (3), Autosomal dominant
<i>PPP3CA</i>	114105	Epileptic encephalopathy, infantile or early childhood, 1, 617711 (3), Autosomal dominant
<i>PPT1</i>	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive

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<i>PQBP1</i>	300463	Renpenning syndrome, 309500 (3), X-linked recessive
<i>PRF1</i>	170280	Aplastic anemia, 609135 (3); Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Lymphoma, non-Hodgkin, 605027 (3)
<i>PRICKLE1</i>	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
<i>PRKAR1A</i>	188830	Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant; Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant
<i>PRMT7</i>	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
<i>PRODH</i>	606810	Hyperprolinemia, type I, 239500 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant
<i>PRPS1</i>	311850	Arts syndrome, 301835 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive No OMIM phenotype
<i>PRR12</i>	616633	Global developmental delay, intellectual disability, eye and vision abnormalities, dysmorphic features, and neuropsychiatric problems (Leduc (2018), Hum Genet), Autosomal dominant
<i>PRRT2</i>	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant
<i>PRSS12</i>	606709	Mental retardation, autosomal recessive 1, 249500 (3), Autosomal recessive
<i>PSAP</i>	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive
<i>PSEN1</i>	104311	?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant, Isolated cases
<i>PSPH</i>	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
<i>PTCH1</i>	601309	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Holoprosencephaly 7, 610828 (3), Autosomal dominant
<i>PTCHD1</i>	300828	{Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive
<i>PTDSS1</i>	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
<i>PTEN</i>	601728	Bannayan-Riley-Ruvalcaba syndrome, 153480 (3); 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



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<i>PTPN11</i>	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3); Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant
<i>PTRH2</i>	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
<i>PTRHD1</i>	617342	No OMIM phenotype ?Neurodevelopmental disorder (Reuter (2017) JAMA Psychiatry), Autosomal recessive Intellectual disability and parkinsonism (Khodadadi (2017), Mov Disord and Elahi (2018), Mov Disord), Autosomal Recessive
<i>PTS</i>	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
<i>PUF60</i>	604819	Verheij syndrome, 615583 (3), Autosomal dominant
<i>PUM1</i>	607204	Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant
<i>PURA</i>	600473	Mental retardation, autosomal dominant 31, 616158 (3), Autosomal dominant
<i>PUS1</i>	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
<i>PUS3</i>	616283	?Mental retardation, autosomal recessive 55, 617051 (3), Autosomal recessive
<i>PYCR1</i>	179035	Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIIB, 614438 (3)
<i>PYCR2</i>	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
<i>QARS</i>	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive
<i>QDPR</i>	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
<i>RAB11B</i>	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
<i>RAB18</i>	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
<i>RAB27A</i>	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
<i>RAB39B</i>	300774	Mental retardation, X-linked 72, 300271 (3), X-linked recessive; ?Waisman syndrome, 311510 (3), X-linked recessive
<i>RAB3GAP1</i>	602536	Warburg micro syndrome 1, 600118 (3), Autosomal recessive
<i>RAB3GAP2</i>	609275	Martsolf syndrome, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
<i>RAC1</i>	602048	Mental retardation, autosomal dominant 48, 617751 (3), Autosomal dominant
<i>RAC3</i>	602050	No OMIM phenotype Severe intellectual disability and brain malformations (Costain 2018 Genet Med), Autosomal dominant
<i>RAD21</i>	606462	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant
<i>RAF1</i>	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3), Autosomal dominant
<i>RAI1</i>	607642	Smith-Magenis syndrome, 182290 (3), Autosomal dominant, Isolated cases
<i>RARB</i>	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant
<i>RARS2</i>	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
<i>RBBP8</i>	604124	Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3); Seckel syndrome 2, 606744 (3), Autosomal recessive

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		No OMIM phenotype
<i>RBFOX1</i>	605104	Epilepsy, rolandic (Lal (2013) PLoS One 8, e73323), Autosomal dominant Mental retardation (Bhalla (2004) J Hum Genet 49, 308 ?Autism spectrum disorder (Griswold (2015) Mol Autism 6, 43) ?Developmental coordination disorder (Mosca (2016) J Med Genet 53,812) ?Developmental delay (Kamien (2014) Am J Med Genet A 164, 1411)
<i>RBM10</i>	300080	TARP syndrome, 311900 (3), X-linked recessive
<i>RBM28</i>	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
<i>RBPJ</i>	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
<i>RCBTB1</i>	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
<i>RELN</i>	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
<i>RERE</i>	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
		No OMIM phenotype
<i>REV3L</i>	602776	Moebius syndrome (Tomas-Roca (2015) Nat Commun 6), Autosomal dominant Psoriasis,association with} (Strange (2010) Nat Genet 42,985) {Colorectal cancer,increased risk,association with} (Webb (2006) Hum Mol Genet 15,3263), Autosomal dominant
<i>RFT1</i>	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
		No OMIM phenotype
<i>RHEB</i>	601293	?Intellectual disability (Reijnders 2017 (Nat Commun)), Autosomal dominant
<i>RIT1</i>	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
<i>RLIM</i>	300379	Mental retardation, X-linked 61, 300978 (3), X-linked recessive
<i>RMND1</i>	614917	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive
<i>RMRP</i>	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive
<i>RNASEH2A</i>	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
<i>RNASEH2B</i>	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
<i>RNASEH2C</i>	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
<i>RNASET2</i>	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
<i>RNF113A</i>	300951	?Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked dominant
<i>RNF125</i>	610432	Tenorio syndrome, 616260 (3), Autosomal dominant
<i>ROGDI</i>	614574	Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive
<i>RPGRIP1L</i>	610937	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
<i>RPL10</i>	312173	{Autism, susceptibility to, X-linked 5}, 300847 (3); Mental retardation, X-linked, syndromic, 35, 300998 (3), X-linked recessive
<i>RPS6KA3</i>	300075	Coffin-Lowry syndrome, 303600 (3), X-linked dominant, Isolated cases; Mental retardation, X-linked 19, 300844 (3), X-linked dominant

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<i>RRM2B</i>	604712	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
<i>RSPRY1</i>	616585	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive
<i>RTEL1</i>	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant
<i>RTTN</i>	610436	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
<i>RUBCN</i>	613516	?Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
<i>RUSC2</i>	611053	Mental retardation, autosomal recessive 61, 617773 (3), Autosomal recessive
<i>SALL1</i>	602218	Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant; Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant
<i>SAMHD1</i>	606754	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
<i>SATB2</i>	608148	Glass syndrome, 612313 (3), Autosomal dominant
<i>SBDS</i>	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
<i>SC5D</i>	602286	Lathosterolosis, 607330 (3), Autosomal recessive
<i>SCARB2</i>	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
<i>SCN1A</i>	182389	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant
<i>SCN1B</i>	600235	Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3); Cardiac conduction defect, nonspecific, 612838 (3); Epilepsy, generalized, with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 52, 617350 (3), Autosomal recessive
<i>SCN2A</i>	182390	Epileptic encephalopathy, early infantile, 11, 613721 (3), Autosomal dominant; Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant
<i>SCN8A</i>	600702	?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
<i>SCO1</i>	603644	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>SCO2</i>	604272	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 (3), Autosomal recessive; Myopia 6, 608908 (3), Autosomal dominant
<i>SCYL1</i>	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
<i>SDHA</i>	600857	Cardiomyopathy, dilated, 1GG, 613642 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive; Paragangliomas 5, 614165 (3), Autosomal dominant
<i>SEMA3E</i>	608166	?CHARGE syndrome, 214800 (3), Autosomal dominant
<i>SEPSECS</i>	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
<i>SERAC1</i>	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
<i>SETBP1</i>	611060	Mental retardation, autosomal dominant 29, 616078 (3), Autosomal dominant; Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant

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<i>SETD1A</i>	611052	No OMIM phenotype Schizophrenia (Takata (2014) Neuron 82, 723), Autosomal dominant
<i>SETD2</i>	612778	Schizophrenia and developmental disorders (Singh (2016) Nat Neurosci), Autosomal dominant
<i>SETD5</i>	615743	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant
<i>SF1</i>	601516	Mental retardation, autosomal dominant 23, 615761 (3), Autosomal dominant
<i>SGSH</i>	605270	No OMIM phenotype
<i>SHANK2</i>	603290	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
<i>SHANK3</i>	606230	{Autism susceptibility 17}, 613436 (3)
<i>SHH</i>	600725	Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3) 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
<i>SHOC2</i>	602775	Noonan-like syndrome with loose anagen hair, 607721 (3), Autosomal dominant
<i>SHROOM4</i>	300579	Stocco dos Santos X-linked mental retardation syndrome, 300434 (3), X-linked
<i>SIK1</i>	605705	Epileptic encephalopathy, early infantile, 30, 616341 (3), Autosomal dominant
<i>SIL1</i>	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
<i>SIN3A</i>	607776	Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant
<i>SIX3</i>	603714	Holoprosencephaly 2, 157170 (3), Autosomal dominant; Schizencephaly, 269160 (3)
<i>SKI</i>	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
<i>SLC12A5</i>	606726	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 34, 616645 (3), Autosomal recessive
<i>SLC12A6</i>	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive
<i>SLC13A5</i>	608305	Epileptic encephalopathy, early infantile, 25, 615905 (3), Autosomal recessive
<i>SLC16A1</i>	600682	Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant
<i>SLC16A2</i>	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
<i>SLC17A5</i>	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
<i>SLC19A3</i>	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
<i>SLC1A1</i>	133550	Dicarboxylic aminoaciduria, 222730 (3), Autosomal recessive; {?Schizophrenia susceptibility 18}, 615232 (3)
<i>SLC1A2</i>	600300	Epileptic encephalopathy, early infantile, 41, 617105 (3), Autosomal dominant
<i>SLC1A4</i>	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
<i>SLC25A1</i>	190315	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive
<i>SLC25A12</i>	603667	Epileptic encephalopathy, early infantile, 39, 612949 (3), Autosomal recessive
<i>SLC25A15</i>	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
<i>SLC25A22</i>	609302	Epileptic encephalopathy, early infantile, 3, 609304 (3), Autosomal recessive

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<i>SLC2A1</i>	138140	Dystonia 9, 601042 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant
<i>SLC33A1</i>	603690	Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive; Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant
<i>SLC35A1</i>	605634	Congenital disorder of glycosylation, type II <sub>f</sub> , 603585 (3), Autosomal recessive
<i>SLC35A2</i>	314375	Congenital disorder of glycosylation, type II <sub>m</sub> , 300896 (3), X-linked dominant, Somatic mosaicism
<i>SLC35C1</i>	605881	Congenital disorder of glycosylation, type II <sub>c</sub> , 266265 (3), Autosomal recessive
<i>SLC39A8</i>	608732	Congenital disorder of glycosylation, type II <sub>n</sub> , 616721 (3), Autosomal recessive
<i>SLC4A4</i>	603345	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive
<i>SLC6A1</i>	137165	Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant
<i>SLC6A17</i>	610299	Mental retardation, autosomal recessive 48, 616269 (3), Autosomal recessive
<i>SLC6A3</i>	126455	{Nicotine dependence, protection against}, 188890 (3); Parkinsonism-dystonia, infantile, 613135 (3), Autosomal recessive
<i>SLC6A8</i>	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
<i>SLC7A7</i>	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
<i>SLC9A6</i>	300231	Mental retardation, X-linked syndromic, Christianson type, 300243 (3), X-linked dominant
<i>SMAD4</i>	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant; Myhre syndrome, 139210 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant
<i>SMAD6</i>	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
<i>SMARCA2</i>	600014	Nicolaiides-Baraitser syndrome, 601358 (3), Autosomal dominant
<i>SMARCA4</i>	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant
<i>SMARCB1</i>	601607	Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant; Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant
<i>SMARCE1</i>	603111	Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant; {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant
<i>SMC1A</i>	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant
<i>SMC3</i>	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
<i>SMG9</i>	613176	Heart and brain malformation syndrome, 616920 (3), Autosomal recessive
<i>SMOC1</i>	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
<i>SMPD1</i>	607608	Niemann-Pick disease, type A, 257200 (3), Autosomal recessive; Niemann-Pick disease, type B, 607616 (3), Autosomal recessive
<i>SMS</i>	300105	Mental retardation, X-linked, Snyder-Robinson type, 309583 (3), X-linked recessive
<i>SNAP25</i>	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
<i>SNAP29</i>	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive

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<i>SNIP1</i>	608241	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501 (3), Autosomal recessive
<i>SNRPN</i>	182279	Prader-Willi syndrome, 176270 (3), Isolated cases
<i>SNX14</i>	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
<i>SOBP</i>	613667	Mental retardation, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive
<i>SON</i>	182465	ZTTK syndrome, 617140 (3), Autosomal dominant
<i>SOS1</i>	182530	?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant; Noonan syndrome 4, 610733 (3), Autosomal dominant
<i>SOS2</i>	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
<i>SOX10</i>	602229	PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
<i>SOX11</i>	600898	Mental retardation, autosomal dominant 27, 615866 (3), Autosomal dominant
<i>SOX2</i>	184429	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
<i>SOX3</i>	313430	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
<i>SOX5</i>	604975	Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant
<i>SPART</i>	607111	Troyer syndrome, 275900 (3), Autosomal recessive
<i>SPAST</i>	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
<i>SPATA5</i>	613940	Epilepsy, hearing loss, and mental retardation syndrome, 616577 (3), Autosomal recessive
<i>SPECC1L</i>	614140	?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant; Opitz GBBB syndrome, type II, 145410 (3), Autosomal dominant
<i>SPG11</i>	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
<i>SPRED1</i>	609291	Legius syndrome, 611431 (3), Autosomal dominant
<i>SPTAN1</i>	182810	Epileptic encephalopathy, early infantile, 5, 613477 (3), Autosomal dominant
<i>SPTBN2</i>	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
<i>SRCAP</i>	611421	Floating-Harbor syndrome, 136140 (3), Autosomal dominant
<i>SRD5A3</i>	611715	Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive; Kahrizi syndrome, 612713 (3), Autosomal recessive
<i>SRPX2</i>	300642	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 (3)
<i>SSR4</i>	300090	Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive
<i>ST3GAL3</i>	606494	?Epileptic encephalopathy, early infantile, 15, 615006 (3), Autosomal recessive; Mental retardation, autosomal recessive 12, 611090 (3), Autosomal recessive
<i>ST3GAL5</i>	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
<i>STAG1</i>	604358	Mental retardation, autosomal dominant 47, 617635 (3), Autosomal dominant
<i>STAMBP</i>	606247	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
<i>STIL</i>	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
<i>STRA6</i>	610745	Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive; Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive

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<i>STT3A</i>	601134	?Congenital disorder of glycosylation, type Iw, 615596 (3), Autosomal recessive
<i>STT3B</i>	608605	?Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
<i>STX1B</i>	601485	Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant
<i>STXBP1</i>	602926	Epileptic encephalopathy, early infantile, 4, 612164 (3), Autosomal dominant
<i>SUCLA2</i>	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
<i>SUCLG1</i>	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
<i>SUMF1</i>	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
<i>SUOX</i>	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
<i>SURF1</i>	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial
<i>SYN1</i>	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3), X-linked recessive, X-linked dominant
<i>SYNE1</i>	608441	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
<i>SYNGAP1</i>	603384	Mental retardation, autosomal dominant 5, 612621 (3), Autosomal dominant
<i>SYNJ1</i>	604297	Epileptic encephalopathy, early infantile, 53, 617389 (3), Autosomal recessive; Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive
<i>SYP</i>	313475	Mental retardation, X-linked 96, 300802 (3), X-linked dominant
<i>SYT14</i>	610949	Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive
<i>SZT2</i>	615463	Epileptic encephalopathy, early infantile, 18, 615476 (3), Autosomal recessive
<i>TAF1</i>	313650	Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive; Mental retardation, X-linked, syndromic 33, 300966 (3), X-linked recessive
<i>TAF13</i>	600774	Mental retardation, autosomal recessive 60, 617432 (3), Autosomal recessive
<i>TAF2</i>	604912	Mental retardation, autosomal recessive 40, 615599 (3), Autosomal recessive
<i>TAF6</i>	602955	Alazami-Yuan syndrome, 617126 (3), Autosomal recessive
<i>TANGO2</i>	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
<i>TAT</i>	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
<i>TBC1D20</i>	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
<i>TBC1D24</i>	613577	DOORS 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
<i>TBC1D7</i>	612655	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 (3), Autosomal recessive
<i>TBCD</i>	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
<i>TBCE</i>	604934	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

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<i>TBCK</i>	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive
<i>TBL1XR1</i>	608628	Mental retardation, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant
<i>TBP</i>	600075	{Parkinson disease, susceptibility to}, 168600 (3), Isolated cases, Multifactorial; Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant
<i>TBR1</i>	604616	No OMIM phenotype Intellectual disability (Hamdan (2014) PLoS Genet 10), Autosomal dominant ?Autism (O'Roak (2012) Science 338,1619), Autosomal dominant ?Ventriculomegaly (Traylor (2012) Mol Syndromol 3,102), Autosomal dominant (deletions) Cortical malformations and intellectual disability (Vegas (2018) Eur J Med Genet), Autosomal dominant
<i>TCF20</i>	603107	No OMIM phenotype Autism spectrum disorder (Babbs (2014) J Med Genet 51,737), Autosomal dominant Intellectual disability and postnatal overgrowth (Schäffgen (2016) Eur J Hum Genet), Autosomal dominant
<i>TCF4</i>	602272	Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant; Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant
<i>TCF7L2</i>	602228	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
<i>TCN2</i>	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
<i>TCTN1</i>	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
<i>TCTN2</i>	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
<i>TCTN3</i>	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
<i>TDP2</i>	605764	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive
<i>TECPR2</i>	615000	Spastic paraplegia 49, autosomal recessive, 615031 (3), Autosomal recessive
<i>TECR</i>	610057	Mental retardation, autosomal recessive 14, 614020 (3), Autosomal recessive
<i>TELO2</i>	611140	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive
<i>TFAP2A</i>	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
<i>TGDS</i>	616146	Catel-Manzke syndrome, 616145 (3), Autosomal recessive
<i>TGFBR1</i>	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
<i>TGFBR2</i>	190182	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
<i>TGIF1</i>	602630	Holoprosencephaly 4, 142946 (3), Autosomal dominant
<i>TH</i>	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
<i>THOC2</i>	300395	Mental retardation, X-linked 12/35, 300957 (3), X-linked recessive
<i>THOC6</i>	615403	Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive
<i>THRB</i>	190160	Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant
<i>TIMM8A</i>	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
<i>TINF2</i>	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant



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<i>TKT</i>	606781	Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive No OMIM phenotype
<i>TLK2</i>	608439	?Schizophrenia (Gulsuner (2013) Cell 154,518), Autosomal dominant ?Autism spectrum disorder (Li (2016) Mol Psychiatry 21,290), Autosomal dominant Neurodevelopmental disorder (Reijnders (2018) Am J Hum Genet), Autosomal dominant
<i>TMCO1</i>	614123	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980 (3), Autosomal recessive
<i>TMEM107</i>	616183	?Joubert syndrome 29, 617562 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive
<i>TMEM138</i>	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
<i>TMEM165</i>	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
<i>TMEM216</i>	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
<i>TMEM231</i>	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
<i>TMEM237</i>	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
<i>TMEM240</i>	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
<i>TMEM67</i>	609884	{Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive
<i>TMEM70</i>	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
<i>TMLHE</i>	300777	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
<i>TNIK</i>	610005	Mental retardation, autosomal recessive 54, 617028 (3), Autosomal recessive
<i>TOE1</i>	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive
<i>TP53RK</i>	608679	Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive
<i>TPI1</i>	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
<i>TPO</i>	606765	Thyroid dysmorphogenesis 2A, 274500 (3), Autosomal recessive
<i>TPP1</i>	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
<i>TPRKB</i>	608680	Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive
<i>TRAIP</i>	605958	Seckel syndrome 9, 616777 (3), Autosomal recessive
<i>TRAPPC11</i>	614138	Muscular dystrophy, limb-girdle, type 2S, 615356 (3), Autosomal recessive
<i>TRAPPC9</i>	611966	Mental retardation, autosomal recessive 13, 613192 (3), Autosomal recessive
<i>TREX1</i>	606609	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2H, 254110 (3), Autosomal recessive
<i>TRIO</i>	601893	Mental retardation, autosomal dominant 44, 617061 (3), Autosomal dominant
<i>TRIP12</i>	604506	Mental retardation, autosomal dominant 49, 617752 (3), Autosomal dominant
<i>TRIP4</i>	604501	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive

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No OMIM phenotype

<i>TRMT1</i>	611669	Intellectual disability (Davarniya (2015) PLoS One 10,e0129631), Autosomal recessive
<i>TRMT10A</i>	616013	Intellectual disability and addition aberrations (Blaesius (2018) Am J Med Genet), Autosomal Recessive
<i>TRNT1</i>	612907	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
<i>TRPM6</i>	607009	Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive; Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive
<i>TSC1</i>	605284	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
<i>TSC2</i>	191092	Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioliomyomatosis, 606690 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant
<i>TSEN15</i>	608756	?Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioliomyomatosis, somatic, 606690 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
<i>TSEN54</i>	608755	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
<i>TSHB</i>	188540	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
<i>TSPAN7</i>	300096	Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
<i>TTC19</i>	613814	Mental retardation, X-linked 58, 300210 (3), X-linked recessive
<i>TTC21B</i>	612014	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
<i>TTC37</i>	614589	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
<i>TTC8</i>	608132	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
<i>TTI2</i>	614426	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
<i>TUBA1A</i>	602529	Mental retardation, autosomal recessive 39, 615541 (3), Autosomal recessive
<i>TUBA8</i>	605742	Lissencephaly 3, 611603 (3), Autosomal dominant
<i>TUBB</i>	191130	Cortical dysplasia, complex, with other brain malformations 8, 613180 (3), Autosomal recessive
<i>TUBB2A</i>	615101	Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant; Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant
<i>TUBB2B</i>	612850	Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant
<i>TUBB3</i>	602661	Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant
<i>TUBB4A</i>	602662	Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant
<i>TUBG1</i>	191135	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
<i>TUBGCP4</i>	609610	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
<i>TUBGCP6</i>	610053	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
<i>TUSC3</i>	601385	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
<i>TWIST1</i>	601622	Mental retardation, autosomal recessive 7, 611093 (3), Autosomal recessive
		Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant

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<i>TWNK</i>	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Perrault syndrome 5, 616138 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant
<i>UBA5</i>	610552	Epileptic encephalopathy, early infantile, 44, 617132 (3), Autosomal recessive; ?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive
<i>UBE2A</i>	312180	Mental retardation, X-linked syndromic, Nascimento-type, 300860 (3), X-linked recessive
<i>UBE3A</i>	601623	Angelman syndrome, 105830 (3), Isolated cases
<i>UBE3B</i>	608047	Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive
<i>UBR1</i>	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
<i>UBTF</i>	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
<i>UNC80</i>	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive
<i>UPB1</i>	606673	Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive
<i>UPF3B</i>	300298	Mental retardation, X-linked, syndromic 14, 300676 (3), X-linked recessive
<i>UQCRQ</i>	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
<i>UROC1</i>	613012	?Urocanase deficiency, 276880 (3), Autosomal recessive
<i>USP27X</i>	300975	Mental retardation, X-linked 105, 300984 (3), X-linked recessive
<i>USP9X</i>	300072	Mental retardation, X-linked 99, 300919 (3), X-linked recessive; Mental retardation, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant
<i>VLDLR</i>	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
<i>VPS11</i>	608549	Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
<i>VPS13B</i>	607817	Cohen syndrome, 216550 (3), Autosomal recessive
<i>VPS37A</i>	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
<i>VPS53</i>	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
<i>VRK1</i>	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
<i>VWA3B</i>	614884	?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive
<i>WAC</i>	615049	Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant
<i>WASHC4</i>	615748	?Mental retardation, autosomal recessive 43, 615817 (3), Autosomal recessive
<i>WDR19</i>	608151	?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
<i>WDR26</i>	617424	Skraban-Deardorff syndrome, 617616 (3), Autosomal dominant
<i>WDR45</i>	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
<i>WDR62</i>	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
<i>WDR73</i>	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
<i>WDR81</i>	614218	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, nonsyndromic, autosomal recessive 3, 617967 (3)
<i>WFS1</i>	606201	?Cataract 41, 116400 (3), Autosomal dominant; Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant

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<i>WVOX</i>	605131	Epileptic encephalopathy, early infantile, 28, 616211 (3), Autosomal recessive; Esophageal squamous cell carcinoma, somatic, 133239 (3); Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
<i>XK</i>	314850	McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked
<i>XPA</i>	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
<i>XPNPEP3</i>	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
<i>XRCC4</i>	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
<i>XYLT1</i>	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
<i>XYLT2</i>	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
<i>YAP1</i>	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 (3), Autosomal dominant
<i>YME1L1</i>	607472	?Optic atrophy 11, 617302 (3), Autosomal recessive
<i>YWHAG</i>	605356	Epileptic encephalopathy, early infantile, 56, 617665 (3), Autosomal dominant
<i>YY1</i>	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant
<i>ZBTB16</i>	176797	Leukemia, acute promyelocytic, PL2F/RARA type (3); Skeletal defects, genital hypoplasia, and mental retardation, 612447 (3), Autosomal recessive
<i>ZBTB18</i>	608433	Mental retardation, autosomal dominant 22, 612337 (3), Autosomal dominant
<i>ZBTB20</i>	606025	Primrose syndrome, 259050 (3), Autosomal dominant
<i>ZBTB24</i>	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
<i>ZC3H14</i>	613279	Mental retardation, autosomal recessive 56, 617125 (3), Autosomal recessive
<i>ZC4H2</i>	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive
<i>ZDHC15</i>	300576	?Mental retardation, X-linked 91, 300577 (3), X-linked dominant
<i>ZDHC9</i>	300646	Mental retardation, X-linked syndromic, Raymond type, 300799 (3)
<i>ZEB2</i>	605802	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant
<i>ZFHX3</i>	104155	{Prostate cancer, susceptibility to, somatic}, 176807 (3)
<i>ZFHX4</i>	606940	?Ptosis, congenital, 178300 (2), Autosomal dominant
<i>ZFYVE26</i>	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
<i>ZIC1</i>	600470	Craniosynostosis 6, 616602 (3), Autosomal dominant
<i>ZIC2</i>	603073	Holoprosencephaly 5, 609637 (3), Autosomal dominant
<i>ZMYND11</i>	608668	Mental retardation, autosomal dominant 30, 616083 (3), Autosomal dominant
<i>ZNF148</i>	601897	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 (3), Autosomal dominant
<i>ZNF41</i>	314995	Mental retardation, X-linked 89, 300848
<i>ZNF423</i>	604557	Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant; Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant
<i>ZNF592</i>	613624	Spinocerebellar ataxia, autosomal recessive 5, 251300
<i>ZNF674</i>	300573	Mental retardation, X-linked 92, 300851
<i>ZNF711</i>	314990	Mental retardation, X-linked 97, 300803 (3), X-linked

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<i>ZNF81</i>	314998	Mental retardation, X-linked 45, 300498
<i>ZSWIM6</i>	615951	Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant; Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant

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Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets. Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern. OMIM release used for OMIM disease identifiers and descriptions: July 04, 2018

### Possible phenotype mapping keys

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

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional.

More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.