PANEL DE DISCAPACIDAD INTELECTUAL

Nuestro panel incluye genes asociados con discapacidades intelectuales que cubren todos los mecanismos de herencia, así como autismo sindrómico y no sindrómico, microcefalia, trastornos de migración neuronal, regresión del desarrollo y Aicardi Goutierres. La detección del síndrome X frágil es posible ya que nuestro panel incluye la expansión repetida de FMR1.

Nº de genes:	817
Entrega:	25 días
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥150 x
Detalles:	Análisis CNV incluido Análisis de Expansión de repetición: FMR1

SÍNDROMES Y TRASTORNOS COMUNES CUBIERTOS

Síndrome de Aicardi-Goutieres

Síndrome de Bardet-Biedl

Discapacidad intelectual AD, AR, XL

Microsíndrome

Microcefalia

Trastornos de la migración neuronal

Autismo sindrómico

Genes	OMIM (Gen)	Enfermedades asociadas (OMIM)	Herencia
ABAT	137150	GABA-transaminase deficiency	AR
ABCA7	605414	Alzheimer disease 9, susceptibility to	AD
ABCD1	300371	Adrenoleukodystrophy; Adrenomyeloneuropathy, adult	XLR
ACE	106180	Microvascular complications of diabetes 3;Stroke, hemorrhagic;Renal tubular dysgenesis	AR
ACHE	100740	[Blood group, Yt system]	
ACSL4	300157	Mental retardation, XL 63	XLD
ACTB	102630	Baraitser-Winter syndrome 1;?Dystonia, juvenile-onset	AD
ACTG1	102560	Baraitser-Winter syndrome 2;Deafness, AD 20/26	AD
ACTN4	604638	Glomerulosclerosis, focal segmental, 1	AD
ADA	608958	Adenosine deaminase deficiency, partial;Severe combined immunodeficiency due to ADA deficiency	AR, SM
ADAR	146920	Aicardi-Goutieres syndrome 6;Dyschromatosis symmetrica hereditaria	AR, AD
ADAT3	615302	Mental retardation, AR 36	AR
ADCY5	600293	Dyskinesia with orofacial involvement, AR;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia;Dyskinesia, familial, with facial myokymia	AR, AD
ADGRG1	604110	Polymicrogyria, bilateral perisylvian;Polymicrogyria, bilateral frontoparietal	AR

ADK	102750	Hypermethioninemia due to adenosine kinase deficiency	AR
ADNP	611386	Helsmoortel-van der Aa syndrome	AD
ADSL	608222	Adenylosuccinase deficiency	AR
AFF2	300806	Mental retardation, XL, FRAXE type	XLR
AHDC1	615790	Xia-Gibbs syndrome	AD
AHI1	608894	Joubert syndrome 3	AR
AIMP1	603605	Leukodystrophy, hypomyelinating, 3	AR
AKT3	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2	AD
ALDH18A1	138250	Cutis laxa, AD 3;Cutis laxa, AR, type IIIA;Spastic paraplegia 9B, AR;Spastic paraplegia 9A, AD	AD, AR
ALDH5A1	610045	Succinic semialdehyde dehydrogenase deficiency	AR
ALDH7A1	107323	Epilepsy, pyridoxine-dependent	AR
ALG11	613666	Congenital disorder of glycosylation, type Ip	AR
ALG13	300776	?Congenital disorder of glycosylation, type Is;Developmental and epileptic encephalopathy 36	XL
ALX4	605420	Craniosynostosis 5, susceptibility to;Parietal foramina 2;Frontonasal dysplasia 2	AD, AR
AMMECR1	300195	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis	XLR
AMPD1	102770	Myopathy due to myoadenylate deaminase deficiency	AR
AMPD2	102771	Spastic paraplegia 63;Pontocerebellar hypoplasia, type 9	AR

AMT	238310	Glycine encephalopathy	AR
ANK2	106410	Long QT syndrome 4;Cardiac arrhythmia, ankyrin-B-related	AD
ANK3	600465	Mental retardation, AR, 37	AR
ANKLE2	616062	Microcephaly 16, primary, AR	AR
ANKRD11	611192	KBG syndrome	AD
AP1S1	603531	MEDNIK syndrome	AR
AP1S2	300629	Mental retardation, XL syndromic 5	XLR
AP2M1	601024	Intellectual developmental disorder 60 with seizures	AD
AP3B1	603401	Hermansky-Pudlak syndrome 2	AR
AP4B1	607245	Spastic paraplegia 47, AR	AR
AP4M1	602296	Spastic paraplegia 50, AR	AR
ARFGEF2	605371	Periventricular heterotopia with microcephaly	AR
ARHGEF10	608136	Slowed nerve conduction velocity, AD	AD
ARHGEF6	300267		
ARHGEF9	300429	Developmental and epileptic encephalopathy 8	XL
ARID1A	603024	Coffin-Siris syndrome 2	AD
ARID1B	614556	Coffin-Siris syndrome 1	AD
ARID2	609539	Coffin-Siris syndrome 6	AD
ARL13B	608922	Joubert syndrome 8	AR
ARX	300382	Developmental and epileptic encephalopathy 1;Mental retardation, XL 29 and others;Hydranencephaly with abnormal genitalia;Partington syndrome;Lissencephaly, XL 2;Proud syndrome	XLR, XL

ASAH1	613468	Spinal muscular atrophy with progressive myoclonic epilepsy;Farber lipogranulomatosis	AR
ASCL1	100790	Central hypoventilation syndrome, congenital;Haddad syndrome	AD
ASH1L	607999	Mental retardation, AD 52	AD
ASPM	605481	Microcephaly 5, primary, AR	AR
ASTN2	612856		
ASXL1	612990	Bohring-Opitz syndrome;Myelodysplastic syndrome, somatic	AD
ASXL3	615115	Bainbridge-Ropers syndrome	AD
ATP13A2	610513	Spastic paraplegia 78, AR;Kufor-Rakeb syndrome	AR
ATP1A1	182310	Hypomagnesemia, seizures, and mental retardation 2;Charcot-Marie-Tooth disease, axonal, type 2DD	AD
ATP1A3	182350	Alternating hemiplegia of childhood 2;Dystonia-12;Developmental and epileptic encephalopathy 99;CAPOS syndrome	AD
ATP6AP2	300556	Mental retardation, XL, syndromic, Hedera type;?Parkinsonism with spasticity, XL;Congenital disorder of glycosylation, type IIr	XLR
ATP6V0A2	611716	Cutis laxa, AR, type IIA;Wrinkly skin syndrome	AR
ATP7A	300011	Menkes disease;Occipital horn syndrome;Spinal muscular atrophy, distal, XL 3	XLR
ATP8A2	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4	AR

ATR	601215	?Cutaneous telangiectasia and cancer syndrome, familial;Seckel syndrome 1	AD, AR
ATRX	300032	Mental retardation-hypotonic facies syndrome, XL;Alpha-thalassemia/mental retardation syndrome;Alpha-thalassemia myelodysplasia syndrome, somatic	XLR, XLD
AUTS2	607270	Mental retardation, AD 26	AD
B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	AR
B4GAT1	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13	AR
B9D1	614144	?Meckel syndrome 9;Joubert syndrome 27	AR
B9D2	611951	?Meckel syndrome 10;Joubert syndrome 34	AR
BBS4	600374	Bardet-Biedl syndrome 4	AR
BCAP31	300398	Deafness, dystonia, and cerebral hypomyelination	XLR
BCKDK	614901	Branched-chain ketoacid dehydrogenase kinase deficiency	
BCL11A	606557	Dias-Logan syndrome	AD
BCOR	300485	Microphthalmia, syndromic 2	XLD
BCS1L	603647	GRACILE syndrome;Bjornstad syndrome;Mi complex III deficiency, nuclear type 1	AR
BDNF	113505		
BLOC1S1	601444		

BLOC1S3	609762	Hermansky-Pudlak syndrome 8	AR
BLOC1S6	604310	?Hermansky-pudlak syndrome 9	AR
BRAF	164757	Cardiofaciocutaneous syndrome;Adenocarcinoma of lung, somatic;Noonan syndrome 7;Colorectal cancer, somatic;Melanoma, malignant, somatic,;LEOPARD syndrome 3	AD
BRCA2	600185	Prostate cancer;Breast-ovarian cancer, familial, 2;Glioblastoma 3;Wilms tumor;Pancreatic cancer 2;Breast cancer, male, susceptibility to;Fanconi anemia, complementation group D1;Medulloblastoma	AD, SM, AD, AR, AD, AR, SM
BRWD3	300553	Mental retardation, XL 93	XLR
CA2	611492	Osteopetrosis, AR 3, with renal tubular acidosis	AR
CA8	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	AR
CACNA1A	601011	Episodic ataxia, type 2;Migraine, familial hemiplegic, 1;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia;Spinocerebellar ataxia 6;Developemental and epileptic encephalopathy 42	AD
CACNA1C	114205	Long QT syndrome 8;Brugada syndrome 3;Timothy syndrome	AD
CACNA1D	114206	Primary aldosteronism, seizures, and neurologic abnormalities;Sinoatrial node dysfunction and deafness	AD, AR

CACNA1E	601013	Developmental and epileptic encephalopathy 69	AD
CACNA1F	300110	Aland Island eye disease;Night blindness, congenital stationary (incomplete), 2A, XL;Cone-rod dystrophy, XL, 3	XL, XLR
CACNA1G	604065	Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits;Spinocerebellar ataxia 42	AD
CACNA1H	607904	Hyperaldosteronism, familial, type IV;Epilepsy, childhood absence, susceptibility to, 6;Epilepsy, idiopathic generalized, susceptibility to, 6	AD
CACNB2	600003	Brugada syndrome 4	
CAMK2A	114078	?Mental retardation, AR 63;Mental retardation, AD 53	AR, AD
CAMK2B	607707	Mental retardation, AD 54	AD
CAMK2G	602123	Mental retardation, AD 59	AD
CAMTA1	611501	Cerebellar ataxia, nonprogressive, with mental retardation	AD
CARD11	607210	Immunodeficiency 11A;Immunodeficiency 11B with atopic dermatitis;B-cell expansion with NFKB and T-cell anergy	AR, AD
CASK	300172	Mental retardation, with or without nystagmus;Mental retardation and microcephaly with pontine and cerebellar hypoplasia;FG syndrome 4	XLD

CBS	613381	Thrombosis, hyperhomocysteinemic;Homocystinuria, B6-responsive and nonresponsive types	AR
CC2D1A	610055	Mental retardation, AR 3	AR
CC2D2A	612013	Meckel syndrome 6;Joubert syndrome 9;COACH syndrome 2	AR
CCDC22	300859	Ritscher-Schinzel syndrome 2	XLR
CCDC40	613799	Ciliary dyskinesia, primary, 15	AR
CCDC88C	611204	Hydrocephalus, congenital, 1;?Spinocerebellar ataxia 40	AR, AD
CDH11	600023	Teebi hypertelorism syndrome 2;Elsahy-Waters syndrome	AD, AR
CDH15	114019	Mental retardation, AD 3	
CDK5RAP2	608201	Microcephaly 3, primary, AR	AR
CDKL5	300203	Developmental and epileptic encephalopathy 2	XLD
CDON	608707	Holoprosencephaly 11	AD
CENPF	600236	Stromme syndrome	AR
CENPJ	609279	?Seckel syndrome 4;Microcephaly 6, primary, AR	AR
CEP135	611423	Microcephaly 8, primary, AR	AR
CEP152	613529	Microcephaly 9, primary, AR;Seckel syndrome 5	AR
CEP290	610142	Leber congenital amaurosis 10;Meckel syndrome 4;?Bardet-Biedl syndrome 14;Senior-Loken syndrome 6;Joubert syndrome 5	AR
CEP41	610523	Joubert syndrome 15	AR
CEP63	614724	?Seckel syndrome 6	AR
CERT1	604677	Mental retardation, AD 34	AD

CHAMP1	616327	Mental retardation, AD 40	AD
CHD1	602118	Pilarowski-Bjornsson syndrome	AD
CHD2	602119	Epileptic encephalopathy, childhood-onset	AD
CHD3	602120	Snijders Blok-Campeau syndrome	AD
CHD7	608892	CHARGE syndrome;Hypogonadotropic hypogonadism 5 with or without anosmia	AD
CHD8	610528	Autism, susceptibility to, 18	AD
CHL1	607416		
CHMP1A	164010	Pontocerebellar hypoplasia, type 8	AR
CIB2	605564	Usher syndrome, type IJ;Deafness, AR 48	AR
CIC	612082	Mental retardation, AD 45	AD
CIT	605629	Microcephaly 17, primary, AR	AR
CLCN4	302910	Raynaud-Claes syndrome	XLD
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8;Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant	AR
CLP1	608757	Pontocerebellar hypoplasia, type 10	AR
CLTC	118955	Mental retardation, AD 56	AD
CNGB3	605080	Achromatopsia 3	AR
CNKSR2	300724	Mental retardation, XL, syndromic, Houge type	XL
CNTN4	607280		
CNTNAP2	604569	Cortical dysplasia-focal epilepsy syndrome;Pitt-Hopkins like syndrome 1;Autism susceptibility 15	AR
COASY	609855	Neurodegeneration with brain iron accumulation 6;Pontocerebellar hypoplasia, type 12	AR

COL4A1	120130	?Retinal arteries, tortuosity of;Hemorrhage, intracerebral, susceptibility to;Microangiopathy and leukoencephalopathy, pontine, AD;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps;Brain small vessel disease with or without ocular anomalies	AD
CPT2	600650	CPT II deficiency, myopathic, stress-induced;CPT II deficiency, infantile;Encephalopathy, acute, infection-induced, 4, susceptibility to;CPT II deficiency, lethal neonatal	AD, AR, AR
CRADD	603454	Mental retardation, AR 34, with variant lissencephaly	AR
CRBN	609262	Mental retardation, AR 2	AR
CREBBP	600140	Menke-Hennekam syndrome 1;Rubinstein-Taybi syndrome 1	AD
CRIPT	604594	Short stature with microcephaly and distinctive facies	AR
CRPPA	614631	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7	AR
CSMD1	608397		
CSPP1	611654	Joubert syndrome 21	AR
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts	AR
CTCF	604167	Mental retardation, AD 21	AD
CTNNA3	607667	Arrhythmogenic right ventricular dysplasia, familial, 13	AD

CTNNB1	116806	Ovarian cancer, somatic;Colorectal cancer, somatic;Pilomatricoma, somatic;Neurodevelopmental disorder with spastic diplegia and visual defects;Exudative vitreoretinopathy 7;Medulloblastoma, somatic;Hepatocellular carcinoma, somatic	AD
CUL3	603136	Pseudohypoaldosteronism, type IIE;Neurodevelopmental disorder with or without autism or seizures	AD
CUL4B	300304	Mental retardation, XL, syndromic 15 (Cabezas type)	XLR
CUL7	609577	3-M syndrome 1	AR
CUX1	116896	Global developmental delay with or without impaired intellectual development	AD
CUX2	610648	Developmental and epileptic encephalopathy 67	AD
CX3CR1	601470	Macular degeneration, age-related, 12;Coronary artery disease, resistance to;Rapid progression to AIDS from HIV1 infection	
CYB5R3	613213	Methemoglobinemia, type I;Methemoglobinemia, type II	AR
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency;Aldosteronism, glucocorticoid-remediable	AR, AD
CYP27A1	606530	Cerebrotendinous xanthomatosis	AR
DARS1	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	AR

DARS2	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	AR
DCAF17	612515	Woodhouse-Sakati syndrome	AR
DCX	300121	Lissencephaly, XL;Subcortical laminal heterotopia, XL	XL
DDC	107930	Aromatic L-amino acid decarboxylase deficiency	AR
DDX3X	300160	Intellectual developmental disorder, XL, syndrome, Snijders Blok type	XLD, XLR
DEAF1	602635	Vulto-van Silfout-de Vries syndrome;Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures	AD, AR
DHCR7	602858	Smith-Lemli-Opitz syndrome	AR
DIP2B	611379	Mental retardation, FRA12A type	AD
DKC1	300126	Dyskeratosis congenita, XL	XLR
DLG3	300189	Mental retardation, XL 90	XLR
DLG4	602887	Intellectual developmental disorder 62	AD
DLGAP2	605438		
DLX3	600525	Amelogenesis imperfecta, type IV;Trichodontoosseous syndrome	AD
DMXL2	612186	?Polyendocrine-polyneuropathy syndrome;Developmental and epileptic encephalopathy 81;?Deafness, AD 71	AR, AD
DNM1	602377	Developmental and epileptic encephalopathy 31	AD
DNMT3A	602769	Tatton-Brown-Rahman syndrome;Heyn-Sproul-Jackson syndrome;Acute myeloid leukemia, somatic	AD

DOCK8	611432	Hyper-IgE recurrent infection syndrome, AR	AR
DPF2	601671	Coffin-Siris syndrome 7	AD
DPP6	126141	Ventricular fibrillation, paroxysmal familial, 2;Mental retardation, AD 33	AD
DPYD	612779	5-fluorouracil toxicity;Dihydropyrimidine dehydrogenase deficiency	AR
DRD3	126451	Schizophrenia, susceptibility to;Essential tremor, hereditary, 1	AD
DST	113810	?Neuropathy, hereditary sensory and autonomic, type VI;Epidermolysis bullosa simplex, AR 2	AR
DTNBP1	607145	Hermansky-Pudlak syndrome 7	AR
DVL3	601368	Robinow syndrome, AD 3	AD
DYM	607461	Dyggve-Melchior-Clausen disease;Smith-McCort dysplasia	AR
DYNC1H1	600112	Spinal muscular atrophy, lower extremity-predominant 1, AD;Charcot-Marie-Tooth disease, axonal, type 20;Mental retardation, AD 13	AD
DYRK1A	600855	Mental retardation, AD 7	AD
EBF3	607407	Hypotonia, ataxia, and delayed development syndrome	AD
EDC3	609842	?Mental retardation, AR 50	AR
EDN3	131242	Waardenburg syndrome, type 4B;Hirschsprung disease, susceptibility to, 4;Central hypoventilation syndrome, congenital	AD, AR, AD

EDNRB	131244	Waardenburg syndrome, type 4A;Hirschsprung disease, susceptibility to, 2;ABCD syndrome	AD, AR, AD, AR
EEF1A2	602959	Mental retardation, AD 38;Developmental and epileptic encephalopathy 33	AD
EFTUD2	603892	Mandibulofacial dysostosis, Guion-Almeida type	AD
EHMT1	607001	Kleefstra syndrome 1	AD
EIF2B1	606686	Leukoencephalopathy with vanishing white matter	AR
EIF2B2	606454	Ovarioleukodystrophy;Leukoencephalopathy with vanishing white matter	AR
EIF2B3	606273	Leukoencephalopathy with vanishing white matter	AR
EIF2B4	606687	Ovarioleukodystrophy;Leukoencephalopathy with vanishing white matter	AR
EIF2B5	603945	Ovarioleukodystrophy;Leukoencephalopathy with vanishing white matter	AR
EIF2S3	300161	MEHMO syndrome	XLR
ELOVL4	605512	Ichthyosis, spastic quadriplegia, and mental retardation;Stargardt disease 3;Spinocerebellar ataxia 34	AR, AD
ELP2	616054	Mental retardation, AR 58	AR
EMC10	614545	Neurodevelopmental disorder with dysmorphic facies and variable seizures	AR
EMX2	600035	Schizencephaly	
EP300	602700	Rubinstein-Taybi syndrome 2;Colorectal cancer, somatic;Menke-Hennekam syndrome 2	AD

EPB41L1	602879	?Mental retardation, AD 11	AD
ETFB	130410	Glutaric acidemia IIB	AR
EXOC6B	607880	Spondyloepimetaphyseal dysplasia with joint laxity, type 3	AR
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B	AR
EXOSC9	606180	Pontocerebellar hypoplasia, type 1D	AR
EXT1	608177	Chondrosarcoma;Exostoses, multiple, type 1	SM, AD
EZH2	601573	Weaver syndrome	AD
FAM126A	610531	Leukodystrophy, hypomyelinating, 5	AR
FAN1	613534	Interstitial nephritis, karyomegalic	AR
FANCB	300515	Fanconi anemia, complementation group B	XLR
FBN1	134797	Marfan lipodystrophy syndrome;Geleophysic dysplasia 2;Acromicric dysplasia;Marfan syndrome;Weill-Marchesani syndrome 2, dominant;Stiff skin syndrome;MASS syndrome;Ectopia lentis, familial	AD
FBXO11	607871	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities	AD
FGA	134820	Hypodysfibrinogenemia, congenital;Afibrinogenemia, congenital;Amyloidosis, familial visceral;Dysfibrinogenemia, congenital	AR, AD
FGD1	300546	Aarskog-Scott syndrome;Mental retardation, XL syndromic 16	XLR

FGFR2	176943	Crouzon syndrome;Saethre-Chotzen syndrome;Craniofacial-skeletal-dermatologic dysplasia;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis;Gastric cancer, somatic;LADD syndrome;Beare-Stevenson cutis gyrata syndrome;Scaphocephaly, maxillary retrusion, and mental retardation;Apert syndrome;Bent bone dysplasia syndrome;Pfeiffer syndrome;Jackson-Weiss syndrome	AD
FGFR3	134934	Achondroplasia;CATSHL syndrome;Thanatophoric dysplasia, type I;Bladder cancer, somatic;Hypochondroplasia;SADDAN;Colorectal cancer, somatic;Crouzon syndrome with acanthosis nigricans;Cervical cancer, somatic;Nevus, epidermal, somatic;Thanatophoric dysplasia, type II;Spermatocytic seminoma, somatic;Muenke syndrome;LADD syndrome	AD, AD, AR
FH	136850	Fumarase deficiency;Leiomyomatosis and renal cell cancer	AR, AD

FKRP	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	AR
FKTN	607440	Cardiomyopathy, dilated, 1X;Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	AR
FLNA	300017	Frontometaphyseal dysplasia 1;Heterotopia, periventricular, 1;Terminal osseous dysplasia;Congenital short bowel syndrome;Otopalatodigital syndrome, type II;Melnick-Needles syndrome;Cardiac valvular dysplasia, XL;Intestinal pseudoobstruction, neuronal;?FG syndrome 2;Otopalatodigital syndrome, type I	XLR, XLD, XL
FLVCR1	609144	Ataxia, posterior column, with retinitis pigmentosa	AR
FMN2	606373	Mental retardation, AR 47	AR
FMR1	309550	Premature ovarian failure 1;Fragile X syndrome;Fragile X tremor/ataxia syndrome	XL, XLD

FOLR1	136430	Neurodegeneration due to cerebral folate transport deficiency	AR
FOXC1	601090	Anterior segment dysgenesis 3, multiple subtypes;Axenfeld-Rieger syndrome, type 3	AD
FOXG1	164874	Rett syndrome, congenital variant	AD
FOXL2	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 1;Blepharophimosis, epicanthus inversus, and ptosis, type 2;Premature ovarian failure 3	AD, AR, AD
FOXP1	605515	Mental retardation with language impairment and with or without autistic features	AD
FOXP2	605317	Speech-language disorder-1	AD
FRMPD4	300838	Mental retardation, XL 104	XL
FTL	134790	Neurodegeneration with brain iron accumulation 3;Hyperferritinemia-cataract syndrome;L-ferritin deficiency, dominant and recessive	AD, AD, AR
FTO	610966	Growth retardation, developmental delay, facial dysmorphism;Obesity, susceptibility to, BMIQ14	AR
FTSJ1	300499	Mental retardation, XL 9/44	XLR
G6PD	305900	Resistance to malaria due to G6PD deficiency;Hemolytic anemia, G6PD deficient (favism)	XLD

GABBR2	607340	Nicotine dependence, susceptibility to;Neurodevelopmental disorder with poor language and loss of hand skills;Developmental and epileptic encephalopathy 59;Nicotine dependence, protection against	AD
GABRA5	137142	Developmental and epileptic encephalopathy 79	AD
GABRB3	137192	Epilepsy, childhood absence, susceptibility to, 5;Developmental and epileptic encephalopathy 43	AD
GAD1	605363	?Cerebral palsy, spastic quadriplegic, 1;Developmental and epileptic encephalopathy 89	AR
GAMT	601240	Cerebral creatine deficiency syndrome 2	AR
GATAD2B	614998	GAND syndrome	AD
GATM	602360	Cerebral creatine deficiency syndrome 3;Fanconi renotubular syndrome 1	AR, AD
GCK	138079	Diabetes mellitus, permanent neonatal 1;Diabetes mellitus, noninsulin-dependent, late onset;Hyperinsulinemic hypoglycemia, familial, 3;MODY, type II	AR, AD
GDI1	300104	Mental retardation, XL 41	XLD
GDNF	600837	Pheochromocytoma, modifier of;Central hypoventilation syndrome;Hirschsprung disease, susceptibility to, 3	AD
GFAP	137780	Alexander disease	AD
GIGYF2	612003	Parkinson disease 11	

GJC2	608803	Leukodystrophy, hypomyelinating, 2;Spastic paraplegia 44, AR;Lymphatic malformation 3	AR, AD
GK	300474	Glycerol kinase deficiency	XLR
GLI2	165230	Holoprosencephaly 9;Culler-Jones syndrome	AD
GLRB	138492	Hyperekplexia 2	AR
GMPPA	615495	Alacrima, achalasia, and mental retardation syndrome	AR
GMPPB	615320	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14	AR
GNAO1	139311	Developmental and epileptic encephalopathy 17;Neurodevelopmental disorder with involuntary movements	AD
GNAS	139320	Pseudohypoparathyroidism Ic;Osseous heteroplasia, progressive;Pseudopseudohypoparathyroidism; Pseudohypoparathyroidism Ia;Pseudohypoparathyroidism Ib;Pituitary adenoma 3, multiple types, somatic;ACTH-independent macronodular adrenal hyperplasia;McCune-Albright syndrome, somatic, mosaic	AD, SM

GNB1	139380	Leukemia, acute lymphoblastic, somatic;Myelodysplastic syndrome, somatic;Mental retardation, AD 42	AD
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1;Wilms tumor, somatic	XLR
GPC4	300168	Keipert syndrome	XLR
GPC6	604404	Omodysplasia 1	AR
GPHN	603930	Molybdenum cofactor deficiency C	AR
GPT2	138210	Neurodevelopmental disorder with microcephaly and spastic paraplegia	AR
GPX1	138320	Hemolytic anemia due to glutathione peroxidase deficiency	AR
GRIA1	138248		
GRIA3	305915	Intellectual developmental disorder, XL, syndromic, Wu type	XLR
GRIA4	138246	Neurodevelopmental disorder with or without seizures and gait abnormalities	AD
GRID2	602368	Spinocerebellar ataxia, AR 18	AR
GRIK2	138244	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures;Mental retardation, AR, 6	AD, AR
GRIN1	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, AR;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, AD	AR, AD
GRIN2A	138253	Epilepsy, focal, with speech disorder and with or without mental retardation	AD

GRIN2B	138252	Mental retardation, AD 6;Developmental and epileptic encephalopathy 27	AD
GRIP1	604597	Fraser syndrome 3	AR
HBB	141900	Erythrocytosis 6;Methemoglobinemia, beta type;Delta-beta thalassemia;Thalassemia-beta, dominant inclusion-body;Sickle cell anemia;Hereditary persistence of fetal hemoglobin;Malaria, resistance to;Thalassemia, beta;Heinz body anemia	AD, AR
HCCS	300056	Linear skin defects with multiple congenital anomalies 1	XLD
HCFC1	300019	Mental retardation, XL 3 (methylmalonic acidemia and homocysteinemia, cblX type)	XLR
HCN1	602780	Generalized epilepsy with febrile seizures plus, type 10;Developmental and epileptic encephalopathy 24	AD
HDAC4	605314		
HDAC8	300269	Cornelia de Lange syndrome 5	XLD
HECW2	617245	Neurodevelopmental disorder with hypotonia, seizures, and absent language	AD
HEPACAM	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation;Megalencephalic leukoencephalopathy with subcortical cysts 2A	AD, AR

HERC2	605837	[Skin/hair/eye pigmentation 1, blond/brown hair];[Skin/hair/eye pigmentation 1, blue/nonblue eyes];Mental retardation, AR 38	AR
HIVEP2	143054	Mental retardation, AD 43	AD
HNMT	605238	Asthma, susceptibility to;Mental retardation, AR 51	AD, AR
HNRNPH2	300610	Mental retardation, XL, syndromic, Bain type	XLD
HNRNPU	602869	Developmental and epileptic encephalopathy 54	AD
HOXA1	142955	Bosley-Salih-Alorainy syndrome;Athabaskan brainstem dysgenesis syndrome	AR
HPRT1	308000	Hyperuricemia, HRPT-related;Lesch-Nyhan syndrome	XLR
HPS1	604982	Hermansky-Pudlak syndrome 1	AR
HPS4	606682	Hermansky-Pudlak syndrome 4	AR
HPS5	607521	Hermansky-Pudlak syndrome 5	AR
HPS6	607522	Hermansky-Pudlak syndrome 6	AR
HRAS	190020	Thyroid carcinoma, follicular, somatic;Spitz nevus or nevus spilus, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Bladder cancer, somatic;Costello syndrome;Nevus sebaceous or woolly hair nevus, somatic;Congenital myopathy with excess of muscle spindles	AD
HSD11B1	600713	Cortisone reductase deficiency 2	AD
HSD17B10	300256	HSD10 Mi disease	XLD
HSPD1	118190	Leukodystrophy, hypomyelinating, 4;Spastic paraplegia 13, AD	AR, AD
HUWE1	300697	Mental retardation, XL syndromic, Turner type	XL

HYDIN	610812	Ciliary dyskinesia, primary, 5	AR
IDS	300823	Mucopolysaccharidosis II	XLR
IER3IP1	609382	Microcephaly, epilepsy, and diabetes syndrome	AR
IFIH1	606951	Aicardi-Goutieres syndrome 7;Singleton-Merten syndrome 1	AD
IGBP1	300139	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia	XLR
IGF1	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency	AR
IGF1R	147370	Insulin-like growth factor I, resistance to	AD, AR
IL1RAPL1	300206	Mental retardation, XL 21/34	XLR
IMPA1	602064	Mental retardation, AR 59	AR
INPP5E	613037	Mental retardation, truncal obesity, retinal dystrophy, and micropenis;Joubert syndrome 1	AR
INVS	243305	Nephronophthisis 2, infantile	AR
IQSEC2	300522	Mental retardation, XL 1/78	XLD
IRF2BPL	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures	AD
IRX5	606195	Hamamy syndrome	AR
ITGA7	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency	AR
ITGB3	173470	Glanzmann thrombasthenia 2;Glanzmann thrombasthenia;Bleeding disorder, platelet-type, 16, AD;Myocardial infarction, susceptibility to;Bleeding disorder, platelet-type, 24, AD	AR, AD

ITPR1	147265	Spinocerebellar ataxia 15;Spinocerebellar ataxia 29, congenital nonprogressive;Gillespie syndrome	AD, AD, AR
JAG1	601920	Alagille syndrome 1;Charcot-Marie-Tooth disease, axonal, type 2HH;?Deafness, congenital heart defects, and posterior embryotoxon;Tetralogy of Fallot	AD
KANK1	607704	Cerebral palsy, spastic quadriplegic, 2	
KANSL1	612452	Koolen-De Vries syndrome	AD
KAT6A	601408	Arboleda-Tham syndrome	AD
KAT6B	605880	SBBYSS syndrome;Genitopatellar syndrome	AD
KATNB1	602703	Lissencephaly 6, with microcephaly	AR
KCNB1	600397	Developmental and epileptic encephalopathy 26	AD
KCNC1	176258	Epilepsy, progressive myoclonic 7	AD
KCNC3	176264	Spinocerebellar ataxia 13	AD
KCND3	605411	Brugada syndrome 9;Spinocerebellar ataxia 19	AD
KCNJ10	602208	Enlarged vestibular aqueduct, digenic;SESAME syndrome	AR
KCNK9	605874	Birk-Barel syndrome	
KCNMA1	600150	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy;Epilepsy, idiopathic generalized, susceptibility to, 16;Liang-Wang syndrome;Cerebellar atrophy, developmental delay, and seizures	AD, AR

KCNQ2	602235	Seizures, benign neonatal, 1;Developmental and epileptic encephalopathy 7;Myokymia	AD
KCNQ3	602232	Seizures, benign neonatal, 2	AD
KCNQ5	607357	Mental retardation, AD 46	AD
KDM4B	609765	Intellectual developmental disorder, AD 65	AD
KDM5B	605393	Mental retardation, AR 65	AR
KDM5C	314690	Mental retardation, XL, syndromic, Claes-Jensen type	XLR
KDM6A	300128	Kabuki syndrome 2	XLD
KIF11	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation	AD
KIF14	611279	?Meckel syndrome 12;Microcephaly 20, primary, AR	AR
KIF1A	601255	NESCAV syndrome;Spastic paraplegia 30, AR;Neuropathy, hereditary sensory, type IIC;Spastic paraplegia 30, AD	AD, AD, AR, AR
KIF5C	604593	Cortical dysplasia, complex, with other brain malformations 2	AD
KIF7	611254	Joubert syndrome 12;?Hydrolethalus syndrome 2;?Al-Gazali-Bakalinova syndrome;Acrocallosal syndrome	AR
KIFBP	609367	Goldberg-Shprintzen megacolon syndrome	AR
KIRREL3	607761		
KMT2A	159555	Wiedemann-Steiner syndrome	AD
KMT2C	606833	Kleefstra syndrome 2	AD
KMT2D	602113	Kabuki syndrome 1	AD
KMT5B	610881	Mental retardation, AD 51	AD
KNL1	609173	Microcephaly 4, primary, AR	AR
KPTN	615620	Mental retardation, AR 41	AR

KRAS	190070	Arteriovenous malformation of the brain, somatic;Gastric cancer, somatic;Oculoectodermal syndrome, somatic;RAS-associated autoimmune leukoproliferative disorder;Pancreatic carcinoma, somatic;Lung cancer, somatic;Cardiofaciocutaneous syndrome 2;Bladder cancer, somatic;Leukemia, acute myeloid, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Breast cancer, somatic;Noonan syndrome 3	AD
L1CAM	308840	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction;CRASH syndrome;Corpus callosum, partial agenesis of;Hydrocephalus with Hirschsprung disease;MASA syndrome;Hydrocephalus due to aqueductal stenosis	XLR
LAMA1	150320	Poretti-Boltshauser syndrome	AR
LAMA2	156225	Muscular dystrophy, congenital, merosin deficient or partially deficient;Muscular dystrophy, limb-girdle, AR 23	AR
LAMB1	150240	Lissencephaly 5	AR
LAMC3	604349	Cortical malformations, occipital	AR
LAMP2	309060	Danon disease	XLD
LARGE1	603590	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	AR

LEP	164160	Obesity, morbid, due to leptin deficiency	AR
LINS1	610350	Mental retardation, AR 27	AR
LMAN2L	609552	?Mental retardation, AR, 52	AR
LMX1B	602575	Nail-patella syndrome;Focal segmental glomerulosclerosis 10	AD
LRBA	606453	Immunodeficiency, common variable, 8, with autoimmunity	AR
LRP1	107770	?Keratosis pilaris atrophicans	AR
LRP2	600073	Donnai-Barrow syndrome	AR
LZTR1	600574	Noonan syndrome 2;Schwannomatosis-2, susceptibility to;Noonan syndrome 10	AR, AD
MACF1	608271	Lissencephaly 9 with complex brainstem malformation	AD
MAGEL2	605283	Schaaf-Yang syndrome	AD
MAGT1	300715	Congenital disorder of glycosylation, type Icc;Immunodeficiency, XL, with magnesium defect, Epstein-Barr virus infection and neoplasia	XLR
MAN1B1	604346	Rafiq syndrome	AR
MAOA	309850	Antisocial behavior;Brunner syndrome	XLR
MBD5	611472	Mental retardation, AD 1	AD
MBOAT7	606048	Mental retardation, AR 57	AR
MBTPS2	300294	Osteogenesis imperfecta, type XIX;?Olmsted syndrome, XL;Keratosis follicularis spinulosa decalvans, XL;IFAP syndrome with or without BRESHECK syndrome	XLR

MCCC2	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency	AR
MCM4	602638	Immunodeficiency 54	AR
MCM6	601806	Lactase persistence/nonpersistence	AD
MCPH1	607117	Microcephaly 1, primary, AR	AR
MECP2	300005	Rett syndrome, preserved speech variant;Encephalopathy, neonatal severe;Mental retardation, XL, syndromic 13;Rett syndrome;Mental retardation, XL syndromic, Lubs type;Rett syndrome, atypical;Autism susceptibility, XL 3	XLD, XLR, XL
MED12	300188	Opitz-Kaveggia syndrome;Lujan-Fryns syndrome;Ohdo syndrome, XL;Hardikar syndrome	XLR, XLD
MED13	603808	Intellectual developmental disorder 61	AD
MED13L	608771	Transposition of the great arteries, dextro-looped 1;Mental retardation and distinctive facial features with or without cardiac defects	AD
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR
MED23	605042	Mental retardation, AR 18	AR
MEF2C	600662	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations;Chromosome 5q14.3 deletion syndrome	AD

MEGF10	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset;Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant	AR
MEIS2	601740	Cleft palate, cardiac defects, and mental retardation	AD
METTL23	615262	Mental retardation, AR 44	AR
MFRP	606227	Microphthalmia, isolated 5;Nanophthalmos 2	AR
MFSD2A	614397	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities	AR
MGAT2	602616	Congenital disorder of glycosylation, type IIa	AR
MIB1	608677	Left ventricular noncompaction 7	AD
MID1	300552	Opitz GBBB syndrome, type I	XLR
MITF	156845	Melanoma, cutaneous malignant, susceptibility to, 8;Waardenburg syndrome, type 2A;Waardenburg syndrome/ocular albinism, digenic;Tietz albinism-deafness syndrome;COMMAD syndrome	AD, AR
MKKS	604896	McKusick-Kaufman syndrome;Bardet-Biedl syndrome 6	AR
MKS1	609883	Bardet-Biedl syndrome 13;Joubert syndrome 28;Meckel syndrome 1	AR
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts	AR
MPDZ	603785	Hydrocephalus, congenital, 2, with or without brain or eye anomalies	AR

MSMO1	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis	AR
MSR1	153622	Barrett esophagus/esophageal adenocarcinoma	
MT-ATP6			
MT-ATP8			
MT-CO1			
MT-CO2			
MT-CO3			
MT-CYB			
MT-ND1			
MT-ND2			
MT-ND3			
MT-ND4			
MT-ND4L			
MT-ND5			
MT-ND6			
MT-RNR1			
MT-RNR2			
MT-TA			
MT-TC			
MT-TD			
MT-TE			
MT-TF			
MT-TG			
MT-TH			
MT-TI			

MT-TK			
MT-TL1			
MT-TL2			
MT-TM			
MT-TN			
MT-TP			
MT-TQ			
MT-TR			
MT-TS1			
MT-TS2			
MT-TT			
MT-TV			
MT-TW			
MT-TY			
MTHFR	607093	Neural tube defects, susceptibility to;Homocystinuria due to MTHFR deficiency;Schizophrenia, susceptibility to;Thromboembolism, susceptibility to	AR, AD
MTM1	300415	Myotubular myopathy, XL	XLR
MTOR	601231	Focal cortical dysplasia, type II, somatic;Smith-Kingsmore syndrome	AD
MTR	156570	Neural tube defects, folate-sensitive, susceptibility to;Homocystinuria-megaloblastic anemia, cblG complementation type	AR
MYCN	164840	Feingold syndrome 1	AD
MYO1E	601479	Glomerulosclerosis, focal segmental, 6	AR
MYO5A	160777	Griscelli syndrome, type 1	AR
MYO9B	602129	Celiac disease, susceptibility to, 4	
MYT1L	613084	Mental retardation, AD 39	AD

NAA10	300013	Ogden syndrome;Microphthalmia, syndromic 1	XLD, XLR, XL
NAA15	608000	Mental retardation, AD 50	AD
NACC1	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination	AD
NAGA	104170	Schindler disease, type III;Kanzaki disease;Schindler disease, type I	AR
NALCN	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	AD, AR
NBEA	604889	Neurodevelopmental disorder with or without early-onset generalized epilepsy	AD
NCAPD3	609276	Microcephaly 22, primary, AR	AR
NDE1	609449	Lissencephaly 4 (with microcephaly);?Microhydranencephaly	AR
NDP	300658	Norrie disease;Exudative vitreoretinopathy 2, XL	XLR, XLD, XLR
NDST1	600853	Mental retardation, AR 46	AR
NDUFA1	300078	Mi complex I deficiency, nuclear type 12	XLR
NECTIN1	600644	Orofacial cleft 7;Cleft lip/palate-ectodermal dysplasia syndrome	AR
NEK10	618726	Ciliary dyskinesia, primary, 44	AR
NEXMIF	300524	Mental retardation, XL 98	XLD

NF1	613113	Watson syndrome;Leukemia, juvenile myelomonocytic;Neurofibromatosis, type 1;Neurofibromatosis, familial spinal;Neurofibromatosis-Noonan syndrome	AD, AD, SM
NFIA	600727	Brain malformations with or without urinary tract defects	AD
NFIB	600728	Macrocephaly, acquired, with impaired intellectual development	AD
NFIX	164005	Marshall-Smith syndrome;Sotos syndrome 2	AD
NHEJ1	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	
NHS	300457	Nance-Horan syndrome;Cataract 40, XL	XLD, XL
NIPA1	608145	Spastic paraplegia 6, AD	AD
NIPBL	608667	Cornelia de Lange syndrome 1	AD
NLGN3	300336	Autism susceptibility, XL 1;Asperger syndrome susceptibility, XL 1	XL, IC, mi, XL
NLGN4X	300427	Asperger syndrome susceptibility, XL 2;Autism susceptibility, XL 2;Mental retardation, XL	XL, IC, mi, XL
NOTCH2	600275	Alagille syndrome 2;Hajdu-Cheney syndrome	AD
NPHP1	607100	Joubert syndrome 4;Nephronophthisis 1, juvenile;Senior-Loken syndrome-1	AR
NPHP3	608002	Nephronophthisis 3;Meckel syndrome 7;Renal-hepatic-pancreatic dysplasia 1	AR
NR2F1	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome	AD

NR3C2	600983	Hypertension, early-onset, AD, with exacerbation in pregnancy;Pseudohypoaldosteronism type I, AD	AD
NRXN1	600565	Schizophrenia, susceptibility to, 17;Pitt-Hopkins-like syndrome 2	AR
NSD1	606681	Sotos syndrome 1	AD
NSDHL	300275	CK syndrome;CHILD syndrome	XLR, XLD
NSMCE3	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome	AR
NSUN2	610916	Mental retardation, AR 5	AR
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis	AR
NUP133	607613	Nephrotic syndrome, type 18;?Galloway-Mowat syndrome 8	AR
NUS1	610463	?Congenital disorder of glycosylation, type 1aa;Mental retardation, AD 55, with seizures	AR, AD
OCLN	602876	Pseudo-TORCH syndrome 1	AR
OCRL	300535	Lowe syndrome;Dent disease 2	XLR
ODAD4	617095	Ciliary dyskinesia, primary, 35	AR
OFD1	300170	Joubert syndrome 10;Simpson-Golabi-Behmel syndrome, type 2;?Retinitis pigmentosa 23;Orofaciodigital syndrome I	XLR, XLD
OPHN1	300127	Mental retardation, XL, with cerebellar hypoplasia and distinctive facial appearance	XLR
ORC1	601902	Meier-Gorlin syndrome 1	AR
OTC	300461	Ornithine transcarbamylase deficiency	XL
P4HA2	600608	Myopia 25, AD	AD
PACS1	607492	Schuurs-Hoeijmakers syndrome	AD

PAFAH1B1	601545	Subcortical laminar heterotopia;Lissencephaly 1	AD
PAH	612349	Phenylketonuria;[Hyperphenylalaninemia, non-PKU mild]	AR
PAK3	300142	Mental retardation, XL 30/47	XLR
PANK2	606157	HARP syndrome;Neurodegeneration with brain iron accumulation 1	AR
PAX3	606597	Waardenburg syndrome, type 3;Rhabdomyosarcoma 2, alveolar;Craniofacial-deafness-hand syndrome;Waardenburg syndrome, type 1	AD, AR, SM, AD
PAX6	607108	?Coloboma, ocular;Aniridia;?Morning glory disc anomaly;Keratitis;Optic nerve hypoplasia;?Coloboma of optic nerve;Anterior segment dysgenesis 5, multiple subtypes;Cataract with late-onset corneal dystrophy;Foveal hypoplasia 1	AD
PBX1	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay	AD
PCDH15	605514	Usher syndrome, type 1D/F digenic;Deafness, AR 23;Usher syndrome, type 1F	AR, DR, AR
PCDH19	300460	Developmental and epileptic encephalopathy 9	XL
PCNT	605925	Microcephalic osteodysplastic primordial dwarfism, type II	AR
PDCD1	600244	Systemic lupus erythematosus, susceptibility to, 2;Multiple sclerosis, disease progression, modifier of	mi

PDE6D	602676	Joubert syndrome 22	AR
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	XLD
PER2	603426	?Advanced sleep phase syndrome, familial, 1	AD
PEX1	602136	Peroxisome biogenesis disorder 1A (Zellweger);Peroxisome biogenesis disorder 1B (NALD/IRD);Heimler syndrome 1	AR
PEX10	602859	Peroxisome biogenesis disorder 6A (Zellweger);Peroxisome biogenesis disorder 6B	AR
PEX11B	603867	Peroxisome biogenesis disorder 14B	AR
PEX12	601758	Peroxisome biogenesis disorder 3B;Peroxisome biogenesis disorder 3A (Zellweger)	AR
PEX13	601789	Peroxisome biogenesis disorder 11B;Peroxisome biogenesis disorder 11A (Zellweger)	AR
PEX14	601791	Peroxisome biogenesis disorder 13A (Zellweger)	AR
PEX16	603360	Peroxisome biogenesis disorder 8A (Zellweger);Peroxisome biogenesis disorder 8B	AR
PEX19	600279	Peroxisome biogenesis disorder 12A (Zellweger)	AR
PEX2	170993	Peroxisome biogenesis disorder 5B;Peroxisome biogenesis disorder 5A (Zellweger)	AR
PEX26	608666	Peroxisome biogenesis disorder 7A (Zellweger);Peroxisome biogenesis disorder 7B	AR
PEX3	603164	Peroxisome biogenesis disorder 10A (Zellweger);?Peroxisome biogenesis disorder 10B	AR

PEX5	600414	Rhizomelic chondrodysplasia punctata, type 5;Peroxisome biogenesis disorder 2B;Peroxisome biogenesis disorder 2A (Zellweger)	AR
PEX6	601498	Peroxisome biogenesis disorder 4B;Peroxisome biogenesis disorder 4A (Zellweger);Heimler syndrome 2	AD, AR, AR
PEX7	601757	Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata, type 1	AR
PGAP1	611655	Mental retardation, AR 42	AR
PGAP2	615187	Hyperphosphatasia with mental retardation syndrome 3	AR
PGK1	311800	Phosphoglycerate kinase 1 deficiency	XLR
PHF6	300414	Borjeson-Forssman-Lehmann syndrome	XLR
PHF8	300560	Mental retardation syndrome, XL, Siderius type	XLR
PHIP	612870	Chung-Jansen syndrome	AD
PIGA	311770	Paroxysmal nocturnal hemoglobinuria, somatic;Multiple congenital anomalies-hypotonia-seizures syndrome 2	XLR
PIGC	601730	Glycosylphosphatidylinositol biosynthesis defect 16	AR
PIGG	616918	Mental retardation, AR 53	AR
PIGL	605947	CHIME syndrome	AR
PIGN	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1	AR
PIGO	614730	Hyperphosphatasia with mental retardation syndrome 2	AR
PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1	AR

PIK3R2	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1	AD
PITX1	602149	Liebenberg syndrome;Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly	AD
PITX2	601542	Anterior segment dysgenesis 4;Ring dermoid of cornea;Axenfeld-Rieger syndrome, type 1	AD
PLA2G6	603604	Neurodegeneration with brain iron accumulation 2B;Parkinson disease 14, AR;Infantile neuroaxonal dystrophy 1	AR
PLCB1	607120	Developmental and epileptic encephalopathy 12	AR
PLK1	602098		
PLK4	605031	Microcephaly and chorioretinopathy, AR, 2	AR
PLN	172405	Cardiomyopathy, dilated, 1P;Cardiomyopathy, hypertrophic, 18	AD
PLP1	300401	Pelizaeus-Merzbacher disease;Spastic paraplegia 2, XL	XLR
PNKP	605610	Ataxia-oculomotor apraxia 4;Microcephaly, seizures, and developmental delay;?Charcot-Marie-Tooth disease, type 2B2	AR
POGZ	614787	White-Sutton syndrome	AD
POLA1	312040	Van Esch-O'Driscoll syndrome;Pigmentary disorder, reticulate, with systemic manifestations, XL	XLR
POLR1C	610060	Leukodystrophy, hypomyelinating, 11;Treacher Collins syndrome 3	AR

POLR1D	613715	Treacher Collins syndrome 2	AD, AR
POLR3A	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism;Wiedemann-Rautenstrauch syndrome	AR
POLR3B	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	AR
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3;Retinitis pigmentosa 76	AR
POMGNT2	614828	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8	AR
POMK	615247	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12;?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12	AR

POMT1	607423	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	AR
POMT2	607439	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	AR
PON1	168820	Microvascular complications of diabetes 5	
PORCN	300651	Focal dermal hypoplasia	XLD
POT1	606478	Melanoma, cutaneous malignant, susceptibility to, 10;Glioma susceptibility 9	AD
PPM1D	605100	Jansen de Vries syndrome;Breast cancer, somatic	AD
PPOX	600923	Porphyria variegata	AD
PPP2R1A	605983	Mental retardation, AD 36	AD
PPP2R5D	601646	Mental retardation, AD 35	AD
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1	AR
PQBP1	300463	Renpenning syndrome	XLR
PRICKLE1	608500	Epilepsy, progressive myoclonic 1B	AR
PRKCA	176960		

PRKN	602544	Parkinson disease, juvenile, type 2;Ovarian cancer, somatic;Adenocarcinoma of lung, somatic	AR
PRODH	606810	Schizophrenia, susceptibility to, 4;Hyperprolinemia, type I	AD, AR
PRPS1	311850	Charcot-Marie-Tooth disease, XLR, 5;Deafness, XL 1;Phosphoribosylpyrophosphate synthetase superactivity;Arts syndrome;Gout, PRPS-related	XLR, XL
PRSS12	606709	Mental retardation, AR 1	AR
PSMD12	604450	Stankiewicz-Isidor syndrome	AD
PTCH1	601309	Basal cell carcinoma, somatic;Holoprosencephaly 7;Basal cell nevus syndrome	AD
PTCHD1	300828	Autism, susceptibility to, XL 4	XLR
PTEN	601728	Cowden syndrome 1;Lhermitte-Duclos syndrome;Macrocephaly/autism syndrome;Glioma susceptibility 2;Meningioma;Prostate cancer, somatic	AD
PTPN11	176876	Leukemia, juvenile myelomonocytic, somatic;LEOPARD syndrome 1;Metachondromatosis;Noonan syndrome 1	AD
PTPRC	151460	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive;Hepatitis C virus, susceptibility to	AR

PTS	612719	Hyperphenylalaninemia, BH4-deficient, A	AR
PURA	600473	Mental retardation, AD 31	AD
PUS3	616283	Neurodevelopmental disorder with microcephaly and gray sclerae	AR
PXDN	605158	Anterior segment dysgenesis 7, with sclerocornea	AR
PYCR1	179035	Cutis laxa, AR, type IIB;Cutis laxa, AR, type IIIB	AR
PYCR2	616406	Leukodystrophy, hypomyelinating, 10	AR
QARS1	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	AR
QRICH1	617387	Ververi-Brady syndrome	AD
RAB18	602207	Warburg micro syndrome 3	AR
RAB39B	300774	Mental retardation, XL 72;Waisman syndrome	XLR
RAB3GAP1	602536	Martsolf syndrome 2;Warburg micro syndrome 1	AR
RAB3GAP2	609275	Warburg micro syndrome 2;Martsolf syndrome	AR
RAC1	602048	Mental retardation, AD 48	AD
RAD21	606462	Cornelia de Lange syndrome 4;?Mungan syndrome	AD, AR
RAI1	607642	Smith-Magenis syndrome	AD, IC
RAP1GDS1	179502		
RARS2	611524	Pontocerebellar hypoplasia, type 6	AR
RBBP8	604124	Seckel syndrome 2;Jawad syndrome	AR
RBFOX1	605104		
RBM10	300080	TARP syndrome	XLR

RELN	600514	Lissencephaly 2 (Norman-Roberts type);Epilepsy, familial temporal lobe, 7	AR, AD
RERE	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart	AD
REST	600571	Wilms tumor 6, susceptibility to;Fibromatosis, gingival, 5;?Deafness, AD 27	AD
RET	164761	Hirschsprung disease, susceptibility to, 1;Multiple endocrine neoplasia IIA;Medullary thyroid carcinoma;Pheochromocytoma;Multiple endocrine neoplasia IIB;Hirschsprung disease, protection against;Central hypoventilation syndrome, congenital	AD
RIMS1	606629	Cone-rod dystrophy 7	AD
RNASEH2A	606034	Aicardi-Goutieres syndrome 4	AR
RNASEH2B	610326	Aicardi-Goutieres syndrome 2	AR
RNASEH2C	610330	Aicardi-Goutieres syndrome 3	AR
RNF168	612688	RIDDLE syndrome	AR
ROBO2	602431	Vesicoureteral reflux 2	AD
ROGDI	614574	Kohlschutter-Tonz syndrome	AR
ROR2	602337	Brachydactyly, type B1;Robinow syndrome, AR	AD, AR
RPGRIP1L	610937	Joubert syndrome 7;?COACH syndrome 3;Meckel syndrome 5	AR
RPL10	312173	Autism, susceptibility to, XL 5;Mental retardation, XL, syndromic, 35	XLR

RPS6KA3	300075	Coffin-Lowry syndrome;Mental retardation, XL 19	XLD
RTTN	610436	Microcephaly, short stature, and polymicrogyria with seizures	AR
RUSC2	611053	Mental retardation, AR 61	AR
RXYLT1	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	AR
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome;Townes-Brocks syndrome 1	AD
SAMHD1	606754	?Chilblain lupus 2;Aicardi-Goutieres syndrome 5	AD, AR
SASH1	607955	Dyschromatosis universalis hereditaria 1;?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma	AD, AR
SASS6	609321	?Microcephaly 14, primary, AR	AR
SATB2	608148	Glass syndrome	AD
SBF1	603560	Charcot-Marie-Tooth disease, type 4B3	AR
SC5D	602286	Lathosterolosis	AR
SCN1A	182389	Epilepsy, generalized, with febrile seizures plus, type 2;Migraine, familial hemiplegic, 3;Developmental and epileptic encephalopathy 6B, non-Dravet;Febrile seizures, familial, 3A;Dravet syndrome	AD
SCN2A	182390	Developmental and epileptic encephalopathy 11;Seizures, benign familial infantile, 3;Episodic ataxia, type 9	AD

SCN4A	603967	Paramyotonia congenita;Myotonia congenita, atypical, acetazolamide-responsive;Myasthenic syndrome, congenital, 16;Hyperkalemic periodic paralysis, type 2;Hypokalemic periodic paralysis, type 2	AD, AR
SCN8A	600702	?Myoclonus, familial, 2;Cognitive impairment with or without cerebellar ataxia;Developmental and epileptic encephalopathy 13;Seizures, benign familial infantile, 5	AD
SCN9A	603415	Erythermalgia, primary;Generalized epilepsy with febrile seizures plus, type 7;Febrile seizures, familial, 3B;Neuropathy, hereditary sensory and autonomic, type IID;Insensitivity to pain, congenital;Paroxysmal extreme pain disorder;Small fiber neuropathy	AD, AR
SCO2	604272	Myopia 6;Mi complex IV deficiency, nuclear type 2	AD, AR
SDCCAG8	613524	Bardet-Biedl syndrome 16;Senior-Loken syndrome 7	AR
SDHA	600857	Neurodegeneration with ataxia and late-onset optic atrophy;Cardiomyopathy, dilated, 1GG;Leigh syndrome;Mi respiratory chain complex II deficiency;Paragangliomas 5	AD, AR, AR, Mi
SEPSECS	613009	Pontocerebellar hypoplasia type 2D	AR
SET	600960	Mental retardation, AD 58	AD

SETBP1	611060	Schinzel-Giedion midface retraction syndrome;Mental retardation, AD 29	AD
SETD2	612778	Luscan-Lumish syndrome	AD
SETD5	615743	Mental retardation, AD 23	AD
SF3B1	605590	Myelodysplastic syndrome, somatic	
SGCA	600119	Muscular dystrophy, limb-girdle, AR 3	AR
SHANK2	603290	Autism susceptibility 17	
SHH	600725	Schizencephaly;Microphthalmia with coloboma 5;Single median maxillary central incisor;Holoprosencephaly 3	AD
SHROOM4	300579	Stocco dos Santos XL mental retardation syndrome	XL
SIL1	608005	Marinesco-Sjogren syndrome	AR
SIN3A	607776	Witteveen-Kolk syndrome	AD
SIX3	603714	Holoprosencephaly 2;Schizencephaly	AD
SLC12A5	606726	Epilepsy, idiopathic generalized, susceptibility to, 14;Developmental and epileptic encephalopathy 34	AD, AR
SLC16A2	300095	Allan-Herndon-Dudley syndrome	XL
SLC1A1	133550	?Schizophrenia susceptibility 18;Dicarboxylic aminoaciduria	AR
SLC25A12	603667	Developmental and epileptic encephalopathy 39	AR
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR

SLC25A19	606521	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type);Microcephaly, Amish type	AR
SLC27A4	604194	Ichthyosis prematurity syndrome	AR
SLC2A1	138140	GLUT1 deficiency syndrome 2, childhood onset;GLUT1 deficiency syndrome 1, infantile onset, severe;Epilepsy, idiopathic generalized, susceptibility to, 12;Stomatin-deficient cryohydrocytosis with neurologic defects;Dystonia 9	AD, AD, AR
SLC35A2	314375	Congenital disorder of glycosylation, type IIm	SM, XLD
SLC35A3	605632	?Arthrogryposis, mental retardation, and seizures	AR
SLC4A10	605556		
SLC4A4	603345	Renal tubular acidosis, proximal, with ocular abnormalities	AR
SLC6A1	137165	Myoclonic-atonic epilepsy	AD
SLC6A17	610299	Mental retardation, AR 48	AR
SLC6A3	126455	Nicotine dependence, protection against;Parkinsonism-dystonia, infantile, 1	AR
SLC6A4	182138	Obsessive-compulsive disorder;Anxiety-related personality traits	AD
SLC6A8	300036	Cerebral creatine deficiency syndrome 1	XLR
SLC7A7	603593	Lysinuric protein intolerance	AR

SLC9A6	300231	Mental retardation, XL syndromic, Christianson type	XL
SLC9A9	608396	?Autism susceptibility 16	
SLCO1B3	605495	Hyperbilirubinemia, Rotor type, digenic	DR
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;Pancreatic cancer, somatic;Myhre syndrome;Polyposis, juvenile intestinal	AD
SMARCA2	600014	Blepharophimosis-impaired intellectual development syndrome;Nicolaides-Baraitser syndrome	AD
SMARCA4	603254	Coffin-Siris syndrome 4;Rhabdoid tumor predisposition syndrome 2	AD
SMARCB1	601607	Coffin-Siris syndrome 3;Rhabdoid tumor predisposition syndrome 1;Schwannomatosis-1, susceptibility to;Rhabdoid tumors, somatic	AD
SMARCC2	601734	Coffin-Siris syndrome 8	AD
SMARCE1	603111	Coffin-Siris syndrome 5;Meningioma, familial, susceptibility to	AD
SMC1A	300040	Developmental and epileptic encephalopathy 85, with or without midline brain defects;Cornelia de Lange syndrome 2	XLD
SMC3	606062	Cornelia de Lange syndrome 3	AD
SMPD1	607608	Niemann-Pick disease, type A;Niemann-Pick disease, type B	AR
SMS	300105	Mental retardation, XL, Snyder-Robinson type	XLR

SNAI2	602150	Waardenburg syndrome, type 2D;Piebaldism	AR, AD
SNAP25	600322	?Myasthenic syndrome, congenital, 18	AD
SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	AR
SNIP1	608241	Psychomotor retardation, epilepsy, and craniofacial dysmorphism	AR
SOBP	613667	Mental retardation, anterior maxillary protrusion, and strabismus	AR
SOD1	147450	Spastic tetraplegia and axial hypotonia, progressive;Amyotrophic lateral sclerosis 1	AR, AD, AR
SOX10	602229	Waardenburg syndrome, type 4C;PCWH syndrome;Waardenburg syndrome, type 2E, with or without neurologic involvement	AD
SOX11	600898	Coffin-Siris syndrome 9	AD
SOX2	184429	Microphthalmia, syndromic 3;Optic nerve hypoplasia and abnormalities of the central nervous system	AD
SOX3	313430	Panhypopituitarism, XL;Mental retardation, XL, with isolated growth hormone deficiency	XL
SOX5	604975	Lamb-Shaffer syndrome	AD
SPAST	604277	Spastic paraplegia 4, AD	AD
SRCAP	611421	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities;Floating-Harbor syndrome	AD

SRD5A3	611715	Congenital disorder of glycosylation, type Iq;Kahrizi syndrome	AR
ST3GAL3	606494	?Developmental and epileptic encephalopathy 15;Mental retardation, AR 12	AR
STAG1	604358	Mental retardation, AD 47	AD
STAMBP	606247	Microcephaly-capillary malformation syndrome	AR
STIL	181590	Microcephaly 7, primary, AR	AR
STRA6	610745	Microphthalmia, isolated, with coloboma 8;Microphthalmia, syndromic 9	AR
STXBP1	602926	Developmental and epileptic encephalopathy 4	AD
SYN1	313440	?Intellectual developmental disorder, XL 50;Epilepsy, XL, with variable learning disabilities and behavior disorders	XL, XLD, XLR
SYN2	600755	Schizophrenia, susceptibility to	AD
SYNE1	608441	Arthrogryposis multiplex congenita 3, myogenic type;Emery-Dreifuss muscular dystrophy 4, AD;Spinocerebellar ataxia, AR 8	AR, AD
SYNGAP1	603384	Mental retardation, AD 5	AD
SYNJ1	604297	Developmental and epileptic encephalopathy 53;Parkinson disease 20, early-onset	AR
SYP	313475	Mental retardation, XL 96	XLR
TAF13	600774	Mental retardation, AR 60	AR
TAF2	604912	Mental retardation, AR 40	AR
TAF6	602955	Alazami-Yuan syndrome	AR
TAOK1	610266	Developmental delay with or without intellectual impairment or behavioral abnormalities	AD

TBC1D20	611663	Warburg micro syndrome 4	AR
TBC1D23	617687	Pontocerebellar hypoplasia, type 11	AR
TBC1D24	613577	Deafness, AD 65;Deafness , AR 86;Myoclonic epilepsy, infantile, familial;Developmental and epileptic encephalopathy 16;DOORS syndrome;Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp	AD, AR
TBCE	604934	Hypoparathyroidism-retardation-dysmorphism syndrome;Encephalopathy, progressive, with amyotrophy and optic atrophy;Kenny-Caffey syndrome, type 1	AR
TBL1XR1	608628	Pierpont syndrome;Mental retardation, AD 41	AD
TBR1	604616	Intellectual developmental disorder with autism and speech delay	AD
TBX1	602054	Velocardiofacial syndrome;Tetralogy of Fallot;DiGeorge syndrome;Conotruncal anomaly face syndrome	AD
TBX3	601621	Ulnar-mammary syndrome	AD
TCF20	603107	Developmental delay with variable intellectual impairment and behavioral abnormalities	AD
TCF4	602272	Pitt-Hopkins syndrome;Corneal dystrophy, Fuchs endothelial, 3	AD
TCOF1	606847	Treacher Collins syndrome 1	AD
TCTN1	609863	Joubert syndrome 13	AR
TCTN2	613846	?Meckel syndrome 8;Joubert syndrome 24	AR

TCTN3	613847	Joubert syndrome 18;Orofaciodigital syndrome IV	AR
TECR	610057	Mental retardation, AR 14	AR
TECTA	602574	Deafness, AD 8/12;Deafness, AR 21	AD, AR
TET2	612839	Myelodysplastic syndrome, somatic;Immunodeficiency 75	AR
TFAP2A	107580	Branchiooculofacial syndrome	AD
TGIF1	602630	Holoprosencephaly 4	AD
THRA	190120	Hypothyroidism, congenital, nongoitrous, 6	AD
THRB	190160	Thyroid hormone resistance, AR;Thyroid hormone resistance;Thyroid hormone resistance, selective pituitary	AR, AD
TIMM8A	300356	Mohr-Tranebjaerg syndrome	XLR
TINF2	604319	Revesz syndrome;Dyskeratosis congenita, AD 3	AD
TLK2	608439	Mental retardation, AD 57	AD
TMCO1	614123	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	AR
TMEM138	614459	Joubert syndrome 16	AR
TMEM216	613277	Meckel syndrome 2;Joubert syndrome 2	AR
TMEM231	614949	Joubert syndrome 20;Meckel syndrome 11	AR
TMEM237	614423	Joubert syndrome 14	AR
TMEM67	609884	COACH syndrome 1;?RHYNS syndrome;Meckel syndrome 3;Joubert syndrome 6;Bardet-Biedl syndrome 14, modifier of;Nephronophthisis 11	AR

TMLHE	300777	Autism, susceptibility to, XL 6	XLR
TMTC3	617218	Lissencephaly 8	AR
TNIK	610005	Mental retardation, AR 54	AR
TOE1	613931	Pontocerebellar hypoplasia, type 7	AR
TPO	606765	Thyroid dyshormonogenesis 2A	AR
TRAF7	606692	Cardiac, facial, and digital anomalies with developmental delay	AD
TRAPPC9	611966	Mental retardation, AR 13	AR
TREX1	606609	Aicardi-Goutieres syndrome 1, dominant and recessive;Chilblain lupus;Vasculopathy, retinal, with cerebral leukodystrophy;Systemic lupus erythematosus, susceptibility to	AD, AR, AD
TRIO	601893	Intellectual developmental disorder, AD 44, with microcephaly;Intellectual developmental disorder, AD 63, with macrocephaly	AD
TRIP12	604506	Mental retardation, AD 49	AD
TRMT10A	616013	Microcephaly, short stature, and impaired glucose metabolism 1	AR
TRPC6	603652	Glomerulosclerosis, focal segmental, 2	AD
TRPM1	603576	Night blindness, congenital stationary (complete), 1C, AR	
TRRAP	603015	Developmental delay with or without dysmorphic facies and autism;?Deafness, AD 75	AD
TSC1	605284	Lymphangioleiomyomatosis;Focal cortical dysplasia, type II, somatic;Tuberous sclerosis-1	AD

TSC2	191092	?Focal cortical dysplasia, type II, somatic;Lymphangioleiomyomatosis, somatic;Tuberous sclerosis-2	AD
TSEN15	608756	Pontocerebellar hypoplasia, type 2F	AR
TSEN2	608753	Pontocerebellar hypoplasia type 2B	AR
TSEN34	608754	?Pontocerebellar hypoplasia type 2C	AR
TSEN54	608755	Pontocerebellar hypoplasia type 2A;?Pontocerebellar hypoplasia type 5;Pontocerebellar hypoplasia type 4	AR
TSPAN7	300096	Mental retardation, XL 58	XLR
TTC21B	612014	Short-rib thoracic dysplasia 4 with or without polydactyly;Nephronophthisis 12	AR, AD, AR
TTI2	614426	Mental retardation, AR 39	AR
TTN	188840	Cardiomyopathy, dilated, 1G;Muscular dystrophy, limb-girdle, AR 10;Tibial muscular dystrophy, tardive;Myopathy, myofibrillar, 9, with early respiratory failure;Salih myopathy;Cardiomyopathy, familial hypertrophic, 9	AR, AD
TUBA1A	602529	Lissencephaly 3	AD
TUBA8	605742	Cortical dysplasia, complex, with other brain malformations 8	AR
TUBB2B	612850	Cortical dysplasia, complex, with other brain malformations 7	AD
TUBB3	602661	Cortical dysplasia, complex, with other brain malformations 1;Fibrosis of extraocular muscles, congenital, 3A	AD

TUBB4A	602662	Dystonia 4, torsion, AD;Leukodystrophy, hypomyelinating, 6	AD
TUBGCP4	609610	Microcephaly and chorioretinopathy, AR, 3	AR
TUBGCP6	610053	Microcephaly and chorioretinopathy, AR, 1	AR
TUSC3	601385	Mental retardation, AR 7	AR
TYR	606933	[Skin/hair/eye pigmentation 3, light/dark/freckling skin];Waardenburg syndrome/albinism, digenic;[Skin/hair/eye pigmentation 3, blue/green eyes];Melanoma, cutaneous malignant, susceptibility to, 8;Albinism, oculocutaneous, type IA;Albinism, oculocutaneous, type IB	AD, AR
UBE2A	312180	Mental retardation, XL syndromic, Nascimento-type	XLR
UBE3A	601623	Angelman syndrome	AD
UBE3B	608047	Kaufman oculocerebrofacial syndrome	AR
UBR1	605981	Johanson-Blizzard syndrome	AR
UNC80	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	AR
UPF3B	300298	Mental retardation, XL, syndromic 14	XLR
UROC1	613012	?Urocanase deficiency	AR
USH2A	608400	Usher syndrome, type 2A;Retinitis pigmentosa 39	AR
USP9X	300072	Mental retardation, XL 99;Mental retardation, XL 99, syndromic, female-restricted	XLR, XLD
USP9Y	400005	Spermatogenic failure, Y-linked, 2	Y-linked

VDR	601769	Rickets, vitamin D-resistant, type IIA	AR
VLDLR	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	AR
VPS13B	607817	Cohen syndrome	AR
VPS53	615850	Pontocerebellar hypoplasia, type 2E	AR
VRK1	602168	Pontocerebellar hypoplasia type 1A	AR
WAC	615049	Desanto-Shinawi syndrome	AD
WASHC4	615748	?Mental retardation, AR 43	AR
WDFY3	617485	?Microcephaly 18, primary, AD	AD
WDR26	617424	Skraban-Deardorff syndrome	AD
WDR45	300526	Neurodegeneration with brain iron accumulation 5	XLD
WDR62	613583	Microcephaly 2, primary, AR, with or without cortical malformations	AR
WDR81	614218	Hydrocephalus, congenital, 3, with brain anomalies;Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2	AR
WNT1	164820	Osteogenesis imperfecta, type XV;Osteoporosis, early-onset, susceptibility to, AD	AR
WNT5A	164975	Robinow syndrome, AD 1	AD
WWOX	605131	Esophageal squamous cell carcinoma, somatic;Developmental and epileptic encephalopathy 28;Spinocerebellar ataxia, AR 12	AR
YWHAE	605066		
YY1	600013	Gabriele-de Vries syndrome	AD
ZBTB18	608433	Mental retardation, AD 22	AD

ZBTB20	606025	Primrose syndrome	AD
ZBTB24	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	AR
ZC3H14	613279	Mental retardation, AR 56	AR
ZC4H2	300897	Wieacker-Wolff syndrome;Wieacker-Wolff syndrome, female-restricted	XLR, XLD
ZDHHC9	300646	Mental retardation, XL syndromic, Raymond type	XL
ZEB2	605802	Mowat-Wilson syndrome	AD
ZFYVE26	612012	Spastic paraplegia 15, AR	AR
ZIC1	600470	Structural brain anomalies with impaired intellectual development and craniosynostosis;?Craniosynostosis 6	AD
ZIC2	603073	Holoprosencephaly 5	AD
ZIC3	300265	VACTERL association, XL;Congenital heart defects, nonsyndromic, 1, XL;Heterotaxy, visceral, 1, XL	XLR
ZMYND11	608668	Mental retardation, AD 30	AD
ZNF292	616213	Intellectual developmental disorder, AD 64	AD
ZNF335	610827	Microcephaly 10, primary, AR	AR
ZNF41	314995		
ZNF423	604557	Joubert syndrome 19;Nephronophthisis 14	AD, AR
ZNF699	609571	DEGCAGS syndrome	AR
ZNF711	314990	Mental retardation, XL 97	XL
ZNF81	314998		

FMR1	309550	Premature ovarian failure 1;Fragile X syndrome;Fragile X tremor/ataxia syndrome	