Panel Dismorfología

Este panel está diseñado para ayudar a los médicos a diagnosticar a los pacientes que padecen un síndrome dismórfico. El panel incluye craneosinostosis, trastornos craneofaciales, paladar hendido/labio, holoprosencefalia, síndrome de Waardenburg, enfermedad de Hirschsprung, lisencefalia y trastornos de malformación cerebral, entre otros.

Nº de genes:	770
Entrega:	25 días
Cobertura:	≥99,5% ≥20x Cobertura media con profundidad ≥ 150 x
Detalles:	Análisis CNV incluido

Síndromes y trastornos comunes cubiertos

Síndrome de Bardet-Biedl

Síndrome cardiofaciocutáneo

Malformaciones cavernosas cerebrales

Ciliopatías

Labio leporino y paladar hendido

Síndrome de Coffin-Siris

Síndrome de Cornelia de Lange

Displasias esqueléticas ciliopáticas

Craneosinostosis y trastornos craneofaciales

Síndrome de heterotaxia

Enfermedad de Hirschsprung

Holoprosencefalia

Síndrome de Klippel-Feil

Lisencefalia y malformaciones cerebrales

Síndrome de Meckel

Displasia metafisaria

Microsíndrome

Espectro de microftalmía/anoftalmía/coloboma

Displasia epifisaria múltiple

Neurofibromatosis

Síndrome de Noonan

Síndrome de Seckel

Displasia esquelética extendida

Síndrome de Stickler

Esclerosis tuberosa

Síndrome de Waardenburg

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel:

Gene	OMIM (Gen)	Enfermedades asociadas (OMIM)	Herencia
A2ML1	610627	Otitis media, susceptibility to	AD
ABCA12	607800	Ichthyosis, congenital, AR 4A;Ichthyosis, congenital, AR 4B (harlequin)	AR
ABCB6	605452	Microphthalmia, isolated, with coloboma 7;[Blood group, Langereis system];Dyschromatosis universalis hereditaria 3;Pseudohyperkalemia, familial, 2, due to red cell leak	AD
ABCC6	603234	Pseudoxanthoma elasticum, forme fruste;Arterial calcification, generalized, of infancy, 2;Pseudoxanthoma elasticum	AD, AR
ABL1	189980	Congenital heart defects and skeletal malformations syndrome;Leukemia, Philadelphia chromosome-positive, resistant to imatinib	AD, SM
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation	AR
ACTA1	102610	Nemaline myopathy 3, AD or recessive;?Myopathy, scapulohumeroperoneal;Myopathy, congenital, with fiber-type disproportion 1;Myopathy, actin, congenital, with cores;Myopathy, actin, congenital, with excess of thin myofilaments	AD, AR, AD

ACTA2	102620	Aortic aneurysm, familial thoracic 6;Moyamoya disease 5;Multisystemic smooth muscle dysfunction syndrome	AD
ACTB	102630	Baraitser-Winter syndrome 1;?Dystonia, juvenile-onset	AD
ACTG1	102560	Baraitser-Winter syndrome 2;Deafness, AD 20/26	AD
ACVR2B	602730	Heterotaxy, visceral, 4, autosomal	
ADAMTS18	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus	AR
ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type	AR
ADAMTSL2	612277	Geleophysic dysplasia 1	AR
ADGRG1	604110	Polymicrogyria, bilateral perisylvian;Polymicrogyria, bilateral frontoparietal	AR
ADGRG6	612243	Lethal congenital contracture syndrome 9	AR
AEBP1	602981	Ehlers-Danlos syndrome, classic-like, 2	AR
AFF4	604417	CHOPS syndrome	AD
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3	AR
AGRN	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects	AR
AHDC1	615790	Xia-Gibbs syndrome	AD
AHI1	608894	Joubert syndrome 3	AR
AKR1C4	600451	46XY sex reversal 8, modifier of	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
ALDH1A3	600463	Microphthalmia, isolated 8	AR
ALG2	607905	?Congenital disorder of glycosylation, type Ii;Myasthenic syndrome, congenital, 14, with tubular aggregates	AR
ALMS1	606844	Alstrom syndrome	AR
ALPL	171760	Hypophosphatasia, infantile;Odontohypophosphatasia;Hypophosphatasia, childhood;Hypophosphatasia, adult	AR, AD, AR
ALX1	601527	Frontonasal dysplasia 3	AR
ALX4	605420	Craniosynostosis 5, susceptibility to;Parietal foramina 2;Frontonasal dysplasia 2	AD, AR
AMELX	300391	Amelogenesis imperfecta, type 1E	XLD
AMER1	300647	Osteopathia striata with cranial sclerosis	XLD
AMH	600957	Persistent Mullerian duct syndrome, type I	AR
AMHR2	600956	Persistent Mullerian duct syndrome, type II	AR
AMPD2	102771	?Spastic paraplegia 63;Pontocerebellar hypoplasia, type 9	AR
ANKH	605145	Chondrocalcinosis 2;Craniometaphyseal dysplasia	AD
ANKLE2	616062	Microcephaly 16, primary, AR	AR

ANKRD11	611192	KBG syndrome	AD
ANKS6	615370	Nephronophthisis 16	AR
ANO5	608662	Gnathodiaphyseal dysplasia;Muscular dystrophy, limb-girdle, AR 12;Miyoshi muscular dystrophy 3	AD, AR
ANOS1	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)	XLR
AP4M1	602296	Spastic paraplegia 50, AR	AR
ARFGEF2	605371	Periventricular heterotopia with microcephaly	AR
ARHGAP29	610496		
ARHGAP31	610911	Adams-Oliver syndrome 1	AD
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
ARL3	604695	Retinitis pigmentosa 83;Joubert syndrome 35	AD, AR
ARL6	608845	Retinitis pigmentosa 55;Bardet-Biedl syndrome 1, modifier of;Bardet-Biedl syndrome 3	AR, AR, DR
ARSL	300180	Chondrodysplasia punctata, XLR	XLR

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
ASPM	605481	Microcephaly 5, primary, AR	AR
ASXL1	612990	Bohring-Opitz syndrome;Myelodysplastic syndrome, somatic	AD
ASXL3	615115	Bainbridge-Ropers syndrome	AD
ATP6V0A2	611716	Cutis laxa, AR, type IIA;Wrinkly skin syndrome	AR
ATP6V0A4	605239	Distal renal tubular acidosis 3, with or without sensorineural hearing loss	AR
ATP6V1A	607027	Epileptic encephalopathy, infantile or early childhood, 3;Cutis laxa, AR, type IID	AD, AR
ATP6V1E1	108746	Cutis laxa, AR, type IIC	AR
ATP7A	300011	Menkes disease;Occipital horn syndrome;Spinal muscular atrophy, distal, XL 3	XLR

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
B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	AR
B3GAT3	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	AR
B3GLCT	610308	Peters-plus syndrome	AR
B4GALT7	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1	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
BBS1	209901	Bardet-Biedl syndrome 1	AR, DR
BBS10	610148	Bardet-Biedl syndrome 10	AR
BBS12	610683	Bardet-Biedl syndrome 12	AR
BBS2	606151	Bardet-Biedl syndrome 2;Retinitis pigmentosa 74	AR
BBS4	600374	Bardet-Biedl syndrome 4	AR
BBS5	603650	Bardet-Biedl syndrome 5	AR
BBS7	607590	Bardet-Biedl syndrome 7	AR
BBS9	607968	Bardet-Biedl syndrome 9	AR
BCL11A	606557	Dias-Logan syndrome	AD
BCOR	300485	Microphthalmia, syndromic 2	XLD
BGN	301870	Meester-Loeys syndrome;Spondyloepimetaphyseal dysplasia, XL	XL, XLR
BIN1	601248	Centronuclear myopathy 2	AR
BMP1	112264	Osteogenesis imperfecta, type XIII	AR
BMP2	112261	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies;HFE hemochromatosis, modifier of;Brachydactyly, type A2	AD, AR

BMP4	112262	Microphthalmia, syndromic 6;Orofacial cleft 11	AD
BMPR1B	603248	Brachydactyly, type A2;Acromesomelic dysplasia, Demirhan type;Brachydactyly, type A1, D	AD, AR
BNC2	608669	Lower urinary tract obstruction, congenital	AD
BRAF	164757	Cardiofaciocutaneous syndrome;Adenocarcinoma of lung, somatic;Noonan syndrome 7;Colorectal cancer, somatic;Melanoma, malignant, somatic,;LEOPARD syndrome 3	AD
C1R	613785	Ehlers-Danlos syndrome, periodontal type, 1	AD
C1S	120580	C1s deficiency;Ehlers-Danlos syndrome, periodontal type, 2	AD
CANT1	613165	Desbuquois dysplasia 1;Epiphyseal dysplasia, multiple, 7	AR
CASK	300172	Mental retardation, with or without nystagmus;Mental retardation and microcephaly with pontine and cerebellar hypoplasia;FG syndrome 4	XLD

CASR	601199	Epilepsy idiopathic generalized, susceptibility to, 8;Hypocalcemia, AD, with Bartter syndrome;Hypocalciuric hypercalcemia, type I;Hyperparathyroidism, neonatal;Hypocalcemia, AD	AD, AD, AR
CBL	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia;?Juvenile myelomonocytic leukemia	AD, AD, SM
CBS	613381	Thrombosis, hyperhomocysteinemic;Homocystinuria, B6-responsive and nonresponsive types	AR
CC2D2A	612013	Meckel syndrome 6;Joubert syndrome 9;COACH syndrome 2	AR
CCDC103	614677	Ciliary dyskinesia, primary, 17	AR
CCDC28B	610162	Bardet-Biedl syndrome 1, modifier of	AR, DR
CCDC39	613798	Ciliary dyskinesia, primary, 14	AR
CCDC40	613799	Ciliary dyskinesia, primary, 15	AR
CCDC65	611088	Ciliary dyskinesia, primary, 27	AR
CCM2	607929	Cerebral cavernous malformations-2	AD

CCN6	603400	Progressive pseudorheumatoid dysplasia	AR
CCNO	607752	Ciliary dyskinesia, primary, 29	AR
CDC45	603465	Meier-Gorlin syndrome 7	AR
CDH1	192090	Prostate cancer, susceptibility to;Breast cancer, lobular;Blepharocheilodontic syndrome 1;Ovarian cancer, somatic;Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate;Endometrial carcinoma, somatic	AD, SM, AD
CDK13	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder	AD
CDK5RAP2	608201	Microcephaly 3, primary, AR	AR
CDKN1C	600856	IMAGE syndrome;Beckwith-Wiedemann syndrome	AD
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

CEP164	614848	Nephronophthisis 15	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
CFAP298	615494	Ciliary dyskinesia, primary, 26	AR
CFAP418	614477	Retinitis pigmentosa 64;Cone-rod dystrophy 16;Bardet-Biedl syndrome 21	AR
CFAP53	614759	Heterotaxy, visceral, 6, AR	AR
CFL2	601443	Nemaline myopathy 7, AR	AR
CHAT	118490	Myasthenic syndrome, congenital, 6, presynaptic	AR
CHD4	603277	Sifrim-Hitz-Weiss syndrome	AD
CHD7	608892	CHARGE syndrome;Hypogonadotropic hypogonadism 5 with or without anosmia	AD
CHMP1A	164010	Pontocerebellar hypoplasia, type 8	AR

CHRNA 1	100690	Myasthenic syndrome, congenital, 1B, fast-channel;Myasthenic syndrome, congenital, 1A, slow-channel;Multiple pterygium syndrome, lethal type	AD, AR, AD, AR
CHRNB 1	100710	Myasthenic syndrome, congenital, 2A, slow-channel;?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	AD, AR
CHRND	100720	?Myasthenic syndrome, congenital, 3A, slow-channel;Myasthenic syndrome, congenital, 3B, fast-channel;Multiple pterygium syndrome, lethal type;?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency	AD, AR
CHRNE	100725	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency;Myasthenic syndrome, congenital, 4A, slow-channel;Myasthenic syndrome, congenital, 4B, fast-channel	AR, AD, AR

CHRNG	100730	Multiple pterygium syndrome, lethal type;Escobar syndrome	AR
CHST14	608429	Ehlers-Danlos syndrome, musculocontractural type 1	AR
CHST3	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations	AR
CHSY1	608183	Temtamy preaxial brachydactyly syndrome	AR
CILK1	612325	Endocrine-cerebroosteodysplasia;Epilepsy, juvenile myoclonic, susceptibility to, 10	AR, AD
CIT	605629	Microcephaly 17, primary, AR	AR
CLCN5	300008	Nephrolithiasis, type I;Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis;Dent disease;Hypophosphatemic rickets	XLR
CLP1	608757	Pontocerebellar hypoplasia, type 10	AR
CNOT1	604917	Holoprosencephaly 12, with or without pancreatic agenesis;Vissers-Bodmer syndrome	AD
CNTNAP1	602346	Lethal congenital contracture syndrome 7;Hypomyelinating neuropathy, congenital, 3	AR

COASY	609855	Neurodegeneration with brain iron accumulation 6;Pontocerebellar hypoplasia, type 12	AR
COG5	606821	Congenital disorder of glycosylation, type IIi	AR
COL10A1	120110	Metaphyseal chondrodysplasia, Schmid type	AD
COL11A1	120280	Marshall syndrome;Fibrochondrogenesis 1;Lumbar disc herniation, susceptibility to;?Deafness, AD 37;Stickler syndrome, type II	AD, AR
COL11A2	120290	Deafness, AR 53;Otospondylomegaepiphyseal dysplasia, AR;Fibrochondrogenesis 2;Otospondylomegaepiphyseal dysplasia, AD;Deafness, AD 13	AR, AD, AR, AD
COL12A1	120320	?Ullrich congenital muscular dystrophy 2;Bethlem myopathy 2	AR, AD
COL13A1	120350	Myasthenic syndrome, congenital, 19	AR

COL1A1	120150	Ehlers-Danlos syndrome, arthrochalasia type, 1;Bone mineral density variation QTL, osteoporosis;Osteogenesis imperfecta, type III;Osteogenesis imperfecta, type I;Caffey disease;Osteogenesis imperfecta, type IV;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1;Osteogenesis imperfecta, type II	AD
COL1A2	120160	Osteoporosis, postmenopausal;Ehlers-Danlos syndrome, cardiac valvular type;Ehlers-Danlos syndrome, arthrochalasia type, 2;Osteogenesis imperfecta, type III;Osteogenesis imperfecta, type IV;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2;Osteogenesis imperfecta, type II	AD, AR

COL2A1	120140	Legg-Calve-Perthes disease;Stickler syndrome, type I;Osteoarthritis with mild chondrodysplasia;Platyspondylic skeletal dysplasia, Torrance type;Spondyloepiphyseal dysplasia, Stanescu type;Kniest dysplasia;Czech dysplasia;Stickler sydrome, type I, nonsyndromic ocular;?Vitreoretinopathy with phalangeal epiphyseal dysplasia;?Epiphyseal dysplasia, multiple, with myopia and deafness;Avascular necrosis of the femoral head;Spondyloperipheral dysplasia;Achondrogenesis, type II or hypochondrogenesis;SMED Strudwick type;SED congenita	AD
COL3A1	120180	Ehlers-Danlos syndrome, vascular type;Polymicrogyria with or without vascular-type EDS	AD, 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
COL5A1	120215	Fibromuscular dysplasia, multifocal;Ehlers-Danlos syndrome, classic type, 1	AD
COL5A2	120190	Ehlers-Danlos syndrome, classic type, 2	AD
COL6A1	120220	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1	AD, AR
COL6A2	120240	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;?Myosclerosis, congenital	AD, AR, AR
COL6A3	120250	Dystonia 27;Ullrich congenital muscular dystrophy 1;Bethlem myopathy 1	AR, AD, AR
COL9A1	120210	?Epiphyseal dysplasia, multiple, 6;Stickler syndrome, type IV	AD

COL9A2	120260	?Stickler syndrome, type V;Epiphyseal dysplasia, multiple, 2	AR, AD
COL9A3	120270	Epiphyseal dysplasia, multiple, 3, with or without myopathy;Intervertebral disc disease, susceptibility to	AD
COLEC10	607620	3MC syndrome 3	AR
COLEC11	612502	3MC syndrome 2	AR
COLQ	603033	Myasthenic syndrome, congenital, 5	AR
COMP	600310	Carpal tunnel syndrome 2;Epiphyseal dysplasia, multiple, 1;Pseudoachondroplasia	AD
COX7B	300885	Linear skin defects with multiple congenital anomalies 2	XLD
CRB2	609720	Focal segmental glomerulosclerosis 9;Ventriculomegaly with cystic kidney disease	AR
CREB3L1	616215	Osteogenesis imperfecta, type XVI	AR
CREBBP	600140	Menke-Hennekam syndrome 1;Rubinstein-Taybi syndrome 1	AD
CRELD1	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome;Atrioventricular septal defect, susceptibility to, 2	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
CRTAP	605497	Osteogenesis imperfecta, type VII	AR
CSGALNACT1	616615	Skeletal dysplasia, mild, with joint laxity and advanced bone age	AR
CSPP1	611654	Joubert syndrome 21	AR
CTNNA2	114025	Cortical dysplasia, complex, with other brain malformations 9	AR
CTNND1	601045	Blepharocheilodontic syndrome 2	AD
CUL4B	300304	Mental retardation, XL, syndromic 15 (Cabezas type)	XLR
CUL7	609577	3-M syndrome 1	AR
CYB5A	613218	Methemoglobinemia and ambiguous genitalia	AR
CYP19A1	107910	Aromatase excess syndrome;Aromatase deficiency	AD
CYP1B1	601771	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset;Anterior segment dysgenesis 6, multiple subtypes	AR

CYP26B1	605207	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies	
DAG1	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9	AR
DCC	120470	Esophageal carcinoma, somatic;Colorectal cancer, somatic;Gaze palsy, familial horizontal, with progressive scoliosis, 2;Mirror movements 1 and/or agenesis of the corpus callosum	AR, AD
DCHS1	603057	Van Maldergem syndrome 1;Mitral valve prolapse 2	AR, AD
DCX	300121	Lissencephaly, XL;Subcortical laminal heterotopia, XL	XL
DDR2	191311	Spondylometaepiphyseal dysplasia, short limb-hand type;Warburg-Cinotti syndrome	AR, AD
DDX59	615464	Orofaciodigital syndrome V	AR

DEAF1	602635	Vulto-van Silfout-de Vries syndrome;Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures	AD, AR
DHCR24	606418	Desmosterolosis	AR
DHCR7	602858	Smith-Lemli-Opitz syndrome	AR
DHODH	126064	Miller syndrome	AR
DLL3	602768	Spondylocostal dysostosis 1, AR	AR
DLL4	605185	Adams-Oliver syndrome 6	AD
DMP1	600980	Hypophosphatemic rickets, AR	AR
DNAAF1	613190	Ciliary dyskinesia, primary, 13	AR
DNAAF2	612517	Ciliary dyskinesia, primary, 10	AR
DNAAF3	614566	Ciliary dyskinesia, primary, 2	AR
DNAAF4	608706	Dyslexia, susceptibility to, 1;Ciliary dyskinesia, primary, 25	AD, AR
DNAAF5	614864	Ciliary dyskinesia, primary, 18	AR
DNAH1	603332	Spermatogenic failure 18;?Ciliary dyskinesia, primary, 37	AR
DNAH11	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus	AR

DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus	AR
DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus	AR
DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus	AR
DNAJB13	610263	Ciliary dyskinesia, primary, 34	AR
DNAL1	610062	Ciliary dyskinesia, primary, 16	AR
DNM2	602378	Lethal congenital contracture syndrome 5;Charcot-Marie-Tooth disease, axonal type 2M;Charcot-Marie-Tooth disease, dominant intermediate B;Centronuclear myopathy 1	AR, AD
DOCK6	614194	Adams-Oliver syndrome 2	AR
DOK7	610285	Fetal akinesia deformation sequence 3;Myasthenic syndrome, congenital, 10	AR
DPAGT1	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates;Congenital disorder of glycosylation, type Ij	AR
DPF2	601671	Coffin-Siris syndrome 7	AD
DRC1	615288	Ciliary dyskinesia, primary, 21	AR

DSE	605942	Ehlers-Danlos syndrome, musculocontractural type 2	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
DYNC2H1	603297	Short-rib thoracic dysplasia 3 with or without polydactyly	AR, DR
DYNC2LI1	617083	Short-rib thoracic dysplasia 15 with polydactyly	AR
DYRK1A	600855	Mental retardation, AD 7	AD
EBP	300205	MEND syndrome;Chondrodysplasia punctata, XLD	XLR, XLD
ECEL1	605896	Arthrogryposis, distal, type 5D	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
EFEMP2	604633	Cutis laxa, AR, type IB	AR
EFNB1	300035	Craniofrontonasal dysplasia	XLD
EFTUD2	603892	Mandibulofacial dysostosis, Guion-Almeida type	AD
EGR2	129010	Dejerine-Sottas disease;Hypomyelinating neuropathy, congenital, 1;Charcot-Marie-Tooth disease, type 1D	AD, AR, AD
EIF2AK3	604032	Wolcott-Rallison syndrome	AR
EIF2S3	300161	MEHMO syndrome	XLR
ELN	130160	Cutis laxa, AD;Supravalvar aortic stenosis	AD
ENPP1	173335	Arterial calcification, generalized, of infancy, 1;Obesity, susceptibility to;Hypophosphatemic rickets, AR, 2;Cole disease;Diabetes mellitus, non-insulin-dependent, susceptibility to	AR, AD, AR, mi, AD
EOGT	614789	Adams-Oliver syndrome 4	AR
EPG5	615068	Vici syndrome	AR
ERCC1	126380	Cerebrooculofacioskeletal syndrome 4	AR

ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2;Xeroderma pigmentosum, group D;Trichothiodystrophy 1, photosensitive	AR
ERCC5	133530	Xeroderma pigmentosum, group G;Cerebrooculofacioskeletal syndrome 3;Xeroderma pigmentosum, group G/Cockayne syndrome	AR
ERCC6	609413	Lung cancer, susceptibility to;UV-sensitive syndrome 1;Premature ovarian failure 11;Macular degeneration, age-related, susceptibility to, 5;Cockayne syndrome, type B;De Sanctis-Cacchione syndrome;Cerebrooculofacioskeletal syndrome 1	AD, SM, AR, AD
ERF	611888	Chitayat syndrome;Craniosynostosis 4	AD
ESCO2	609353	Roberts syndrome;Juberg-Hayward syndrome;SC phocomelia syndrome	AR
EVC	604831	Ellis-van Creveld syndrome;?Weyers acrofacial dysostosis	AR, AD
EVC2	607261	Ellis-van Creveld syndrome;Weyers acrofacial dysostosis	AR, AD

EXOSC3	606489	Pontocerebellar hypoplasia, type 1B	AR
EXOSC9	606180	Pontocerebellar hypoplasia, type 1D	AR
EXT1	608177	Chondrosarcoma;Exostoses, multiple, type 1	SM, AD
EXT2	608210	Exostoses, multiple, type 2;Seizures, scoliosis, and macrocephaly syndrome	AD, AR
EYA1	601653	Anterior segment anomalies with or without cataract;Branchiootorenal syndrome 1, with or without cataracts;?Otofaciocervical syndrome;Branchiootic syndrome 1	AD
FAM20C	611061	Raine syndrome	AR
FAS	134637	Autoimmune lymphoproliferative syndrome, type IA;Autoimmune lymphoproliferative syndrome	AD
FAT4	612411	Hennekam lymphangiectasia-lymphedema syndrome 2;Van Maldergem syndrome 2	AR
FBLN1	135820	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses	AD

FBLN5	604580	Cutis laxa, AR, type IA;Neuropathy, hereditary, with or without age-related macular degeneration;Macular degeneration, age-related, 3;?Cutis laxa, AD 2	AR, AD
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
FBN2	612570	Contractural arachnodactyly, congenital;Macular degeneration, early-onset	AD
FBXL4	605654	Mi DNA depletion syndrome 13 (encephalomyopathic type)	AR
FBXW11	605651	Neurodevelopmental, jaw, eye, and digital syndrome	AD
FGD1	300546	Aarskog-Scott syndrome;Mental retardation, XL syndromic 16	XLR
FGF10	602115	Aplasia of lacrimal and salivary glands;LADD syndrome	AD

FGF23	605380	Tumoral calcinosis, hyperphosphatemic, familial, 2;Hypophosphatemic rickets, AD	AR, AD
FGF8	600483	Hypogonadotropic hypogonadism 6 with or without anosmia	AD
FGFR1	136350	Osteoglophonic dysplasia;Trigonocephaly 1;Pfeiffer syndrome;Encephalocraniocutaneous lipomatosis, somatic mosaic;Hypogonadotropic hypogonadism 2 with or without anosmia;Jackson-Weiss syndrome;Hartsfield syndrome	AD

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
FHL1	300163	Reducing body myopathy, XL 1b, with late childhood or adult onset;Scapuloperoneal myopathy, XLD;?Uruguay faciocardiomusculoskeletal syndrome;Myopathy, XL, with postural muscle atrophy;Reducing body myopathy, XL 1a, severe, infantile or early childhood onset;Emery-Dreifuss muscular dystrophy 6, XL	XL, XLD, XLR
FKBP10	607063	Osteogenesis imperfecta, type XI;Bruck syndrome 1	AR

FKBP14	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2	AR
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
FLCN	607273	Birt-Hogg-Dube syndrome;Pneumothorax, primary spontaneous;Renal carcinoma, chromophobe, somatic;Colorectal cancer, somatic	AD

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
FLNB	603381	Larsen syndrome;Boomerang dysplasia;Spondylocarpotarsal synostosis syndrome;Atelosteogenesis, type III;Atelosteogenesis, type I	AD, AR
FOXC1	601090	Anterior segment dysgenesis 3, multiple subtypes;Axenfeld-Rieger syndrome, type 3	AD
FOXE1	602617	Bamforth-Lazarus syndrome;Thyroid cancer, nonmedullary, 4	AR, AD
FOXL2	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 1;Blepharophimosis, epicanthus inversus, and ptosis, type 2;Premature ovarian failure 3	AD, AR, AD

FRAS1	607830	Fraser syndrome 1	AR
FREM1	608944	Manitoba oculotrichoanal syndrome;Trigonocephaly 2;Bifid nose with or without anorectal and renal anomalies	AR, AD
FREM2	608945	Fraser syndrome 2;Cryptophthalmos, unilateral or bilateral, isolated	AR
FZD6	603409	Nail disorder, nonsyndromic congenital, 1	AR
GAS8	605178	Ciliary dyskinesia, primary, 33	AR
GATA4	600576	Atrioventricular septal defect 4;?Testicular anomalies with or without congenital heart disease;Ventricular septal defect 1;Tetralogy of Fallot;Atrial septal defect 2	AD
GBA	606463	Lewy body dementia, susceptibility to;Gaucher disease, type IIIC;Parkinson disease, late-onset, susceptibility to;Gaucher disease, type II;Gaucher disease, type III;Gaucher disease, perinatal lethal;Gaucher disease, type I	AD, AR, AD, mi
GBE1	607839	Glycogen storage disease IV;Polyglucosan body disease, adult form	AR

GDF1	602880	Congenital heart defects, multiple types, 6;Right atrial isomerism (Ivemark)	AD, AR
GDF3	606522	Klippel-Feil syndrome 3, AD;Microphthalmia, isolated 7;Microphthalmia with coloboma 6	AD
GDF5	601146	Du Pan syndrome;Brachydactyly, type C;Osteoarthritis-5;Multiple synostoses syndrome 2;Chondrodysplasia, Grebe type;Brachydactyly, type A1, C;?Acromesomelic dysplasia, Hunter-Thompson type;Brachydactyly, type A2;Symphalangism, proximal, 1B	AR, AD, AD, AR
GDF6	601147	Leber congenital amaurosis 17;Multiple synostoses syndrome 4;Klippel-Feil syndrome 1, AD;Microphthalmia, isolated 4;Microphthalmia with coloboma 6, digenic	AR, AD
GDNF	600837	Pheochromocytoma, modifier of;Central hypoventilation syndrome;Hirschsprung disease, susceptibility to, 3	AD

GFPT1	138292	Myasthenia, congenital, 12, with tubular aggregates	AR
GGCX	137167	Vitamin K-dependent clotting factors, combined deficiency of, 1;Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	AR
GJA1	121014	Oculodentodigital dysplasia, AR;Atrioventricular septal defect 3;Syndactyly, type III;Craniometaphyseal dysplasia, AR;Palmoplantar keratoderma with congenital alopecia;Oculodentodigital dysplasia;Hypoplastic left heart syndrome 1;Erythrokeratodermia variabilis et progressiva 3	AR, AD
GLE1	603371	Congenital arthrogryposis with anterior horn cell disease;Lethal congenital contracture syndrome 1	AR
GLI2	165230	Holoprosencephaly 9;Culler-Jones syndrome	AD

GLI3	165240	Pallister-Hall syndrome;Polydactyly, preaxial, type IV;Polydactyly, postaxial, types A1 and B;Greig cephalopolysyndactyly syndrome	AD
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
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
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2	AR

GORAB	607983	Geroderma osteodysplasticum	AR
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1;Wilms tumor, somatic	XLR
GPC6	604404	Omodysplasia 1	AR
GRHL3	608317	Van der Woude syndrome 2	AD
GRIP1	604597	Fraser syndrome 3	AR
HBA1	141800	Methemoglobinemia, alpha type;Heinz body anemias, alpha-;Erythrocytosis 7;Thalassemias, alpha-;Hemoglobin H disease, nondeletional	AD
HCCS	300056	Linear skin defects with multiple congenital anomalies 1	XLD
HDAC8	300269	Cornelia de Lange syndrome 5	XLD
HES7	608059	Spondylocostal dysostosis 4, AR	AR
HESX1	601802	Septooptic dysplasia;Pituitary hormone deficiency, combined, 5;Growth hormone deficiency with pituitary anomalies	AD, AR
HEXA	606869	Tay-Sachs disease;[Hex A pseudodeficiency];GM2-gangliosidosis, several forms	AR
HMX1	142992	Oculoauricular syndrome	AR

HOXA13	142959	?Guttmacher syndrome;Hand-foot-uterus syndrome	AD
HOXD13	142989	Syndactyly, type V;?Brachydactyly-syndactyly syndrome;Brachydactyly, type D;Synpolydactyly 1;Brachydactyly, type E	AD
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
HSPG2	142461	Schwartz-Jampel syndrome, type 1;Dyssegmental dysplasia, Silverman-Handmaker type	AR
HUWE1	300697	Mental retardation, XL syndromic, Turner type	XL
HYDIN	610812	Ciliary dyskinesia, primary, 5	AR
HYLS1	610693	Hydrolethalus syndrome	AR
IER3IP1	609382	Microcephaly, epilepsy, and diabetes syndrome	AR
IFITM5	614757	Osteogenesis imperfecta, type V	AD

IFT122	606045	Cranioectodermal dysplasia 1	AR
IFT140	614620	Retinitis pigmentosa 80;Short-rib thoracic dysplasia 9 with or without polydactyly	AR
IFT172	607386	Retinitis pigmentosa 71;Bardet-Biedl syndrome 20;Short-rib thoracic dysplasia 10 with or without polydactyly	AR
IFT27	615870	?Bardet-Biedl syndrome 19	AR
IFT43	614068	?Cranioectodermal dysplasia 3;Short-rib thoracic dysplasia 18 with polydactyly;?Retinitis pigmentosa 81	AR
IFT80	611177	Short-rib thoracic dysplasia 2 with or without polydactyly	AR
IFT81	605489	Short-rib thoracic dysplasia 19 with or without polydactyly	AR
IGF1R	147370	Insulin-like growth factor I, resistance to	AD, AR
IHH	600726	Brachydactyly, type A1;Acrocapitofemoral dysplasia	AD, AR
IL11RA	600939	Craniosynostosis and dental anomalies	AR

INPP5E	613037	Mental retardation, truncal obesity, retinal dystrophy, and micropenis;Joubert syndrome 1	AR
INPPL1	600829	Opsismodysplasia	AR
INVS	243305	Nephronophthisis 2, infantile	AR
IPO8	605600	VISS syndrome	AR
IRF6	607199	Orofacial cleft 6;van der Woude syndrome;Popliteal pterygium syndrome 1	AD
ITGB4	147557	Epidermolysis bullosa, junctional, non-Herlitz type;Epidermolysis bullosa of hands and feet;Epidermolysis bullosa, junctional, with pyloric atresia	AR, AD
JAG1	601920	Alagille syndrome 1;Charcot-Marie-Tooth disease, axonal, type 2HH;?Deafness, congenital heart defects, and posterior embryotoxon;Tetralogy of Fallot	AD
KAT6B	605880	SBBYSS syndrome;Genitopatellar syndrome	AD
KATNB1	602703	Lissencephaly 6, with microcephaly	AR
KBTBD13	613727	Nemaline myopathy 6, AD	AD

KDM5C	314690	Mental retardation, XL, syndromic, Claes-Jensen type	XLR
KDM6A	300128	Kabuki syndrome 2	XLD
KIAA0586	610178	Joubert syndrome 23;Short-rib thoracic dysplasia 14 with polydactyly	AR
KIF11	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation	AD
KIF14	611279	?Meckel syndrome 12;Microcephaly 20, primary, AR	AR
KIF22	603213	Spondyloepimetaphyseal dysplasia with joint laxity, type 2	AD
KIF2A	602591	Cortical dysplasia, complex, with other brain malformations 3	AD
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

KIT	164920	Gastrointestinal stromal tumor, familial;Germ cell tumors, somatic;Piebaldism;Leukemia, acute myeloid, somatic;Mastocytosis, systemic, somatic;Mastocytosis, cutaneous	AD, IC, AD
KLHL40	615340	Nemaline myopathy 8, AR	AR
KLHL41	607701	Nemaline myopathy 9	AR
KMT2A	159555	Wiedemann-Steiner syndrome	AD
KMT2D	602113	Kabuki syndrome 1	AD
KNL1	609173	Microcephaly 4, primary, AR	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
KRIT1	604214	Cavernous malformations of CNS and retina;Cerebral cavernous malformations-1;Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations	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
LAMA2	156225	Muscular dystrophy, congenital, merosin deficient or partially deficient;Muscular dystrophy, limb-girdle, AR 23	AR
LAMA3	600805	Laryngoonychocutaneous syndrome;Epidermolysis bullosa, junctional, Herlitz type;Epidermolysis bullosa, generalized atrophic benign	AR
LAMB1	150240	Lissencephaly 5	AR
LAMB3	150310	Epidermolysis bullosa, junctional, non-Herlitz type;Amelogenesis imperfecta, type IA;Epidermolysis bullosa, junctional, Herlitz type	AR, AD

LAMC2	150292	Epidermolysis bullosa, junctional, non-Herlitz type;Epidermolysis bullosa, junctional, Herlitz type	AR
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
LBR	600024	?Reynolds syndrome;Greenberg skeletal dysplasia;Pelger-Huet anomaly;Pelger-Huet anomaly with mild skeletal anomalies	AD, AR
LFNG	602576	Spondylocostal dysostosis 3, AR	AR
LIFR	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	AR

LMNA	150330	Malouf syndrome;Emery-Dreifuss muscular dystrophy 3, AR;Emery-Dreifuss muscular dystrophy 2, AD;Hutchinson-Gilford progeria;Muscular dystrophy, congenital;Restrictive dermopathy, lethal;Lipodystrophy, familial partial, type 2;Charcot-Marie-Tooth disease, type 2B1;Mandibuloacral dysplasia;Cardiomyopathy, dilated, 1A;Heart-hand syndrome, Slovenian type	AD, AR
LMOD3	616112	Nemaline myopathy 10	AR
LMX1B	602575	Nail-patella syndrome;Focal segmental glomerulosclerosis 10	AD
LOX	153455	Aortic aneurysm, familial thoracic 10	AD
LRP2	600073	Donnai-Barrow syndrome	AR
LRP4	604270	?Myasthenic syndrome, congenital, 17;Cenani-Lenz syndactyly syndrome;Sclerosteosis 2	AR, AD, AR

LRP5	603506	van Buchem disease, type 2;Osteosclerosis;Osteoporosis;[Bone mineral density variability 1];Osteopetrosis, AD 1;Polycystic liver disease 4 with or without kidney cysts;Osteoporosis-pseudoglioma syndrome;Hyperostosis, endosteal;Exudative vitreoretinopathy 4	AD, AR, AD, AR
LTBP3	602090	Geleophysic dysplasia 3;Dental anomalies and short stature	AD, AR
LZTFL1	606568	Bardet-Biedl syndrome 17	AR
LZTR1	600574	Noonan syndrome 2;Schwannomatosis-2, susceptibility to;Noonan syndrome 10	AR, AD
MAB21L2	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome	AD, AR
MACF1	608271	Lissencephaly 9 with complex brainstem malformation	AD
MAFB	608968	Duane retraction syndrome 3;Multicentric carpotarsal osteolysis syndrome	AD
MAGEL2	605283	Schaaf-Yang syndrome	AD

MAP2K1	176872	Cardiofaciocutaneous syndrome 3;Melorheostosis, isolated, somatic mosaic	AD
MAP2K2	601263	Cardiofaciocutaneous syndrome 4	AD
MASP1	600521	3MC syndrome 1	AR
MATN3	602109	?Spondyloepimetaphyseal dysplasia, Borochowitz Cormier-Daire type;Epiphyseal dysplasia, multiple, 5;Osteoarthritis susceptibility 2	AR, AD
MBTPS2	300294	Osteogenesis imperfecta, type XIX;?Olmsted syndrome, XL;Keratosis follicularis spinulosa decalvans, XL;IFAP syndrome with or without BRESHECK syndrome	XLR
MCIDAS	614086	Ciliary dyskinesia, primary, 42	AR
MCPH1	607117	Microcephaly 1, primary, AR	AR
MED12	300188	Opitz-Kaveggia syndrome;Lujan-Fryns syndrome;Ohdo syndrome, XL;Hardikar syndrome	XLR, XLD

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
MEGF8	604267	Carpenter syndrome 2	AR
MEIS2	601740	Cleft palate, cardiac defects, and mental retardation	AD
MEOX1	600147	Klippel-Feil syndrome 2	AR
MESP2	605195	Spondylocostal dysostosis 2, AR	AR
MFAP5	601103	Aortic aneurysm, familial thoracic 9	AD
MFRP	606227	Microphthalmia, isolated 5;Nanophthalmos 2	AR
MFSD2A	614397	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities	AR
MGP	154870	Keutel syndrome	AR
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
MMP13	600108	Metaphyseal anadysplasia 1;Metaphyseal dysplasia, Spahr type;?Spondyloepimetaphyseal dysplasia, Missouri type	AD, AR
MMP15	602261		
MMP21	608416	Heterotaxy, visceral, 7, autosomal	AR
MMP9	120361	Metaphyseal anadysplasia 2	
MN1	156100	CEBALID syndrome;Meningioma	AD
MRAS	608435	Noonan syndrome 11	AD
MSMO1	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis	AR

MSX1	142983	Orofacial cleft 5;Tooth agenesis, selective, 1, with or without orofacial cleft;Ectodermal dysplasia 3, Witkop type	AD
MSX2	123101	Parietal foramina with cleidocranial dysplasia;Parietal foramina 1;Craniosynostosis 2	AD
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			
MTM1	300415	Myotubular myopathy, XL	XLR
MUSK	601296	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency;Fetal akinesia deformation sequence 1	AR
MYBPC1	160794	Arthrogryposis, distal, type 1B;Lethal congenital contracture syndrome 4;Myopathy, congenital, with tremor	AD, AR
MYCN	164840	Feingold syndrome 1	AD

MYH11	160745	Aortic aneurysm, familial thoracic 4;Visceral myopathy 2;Megacystis-microcolon-intestinal hypoperistalsis syndrome 2	AD, AR
MYH2	160740	Proximal myopathy and ophthalmoplegia	AD, AR
MYH3	160720	Arthrogryposis, distal, type 2B3 (Sheldon-Hall);Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B;Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A;Arthrogryposis, distal, type 2A (Freeman-Sheldon)	AD, AR
MYH8	160741	Trismus-pseudocamptodactyly syndrome;Carney complex variant	AD
MYLK	600922	Aortic aneurysm, familial thoracic 7;Megacystis-microcolon-intestinal hypoperistalsis syndrome	AD, AR
MYO18B	607295	Klippel-Feil syndrome 4, AR, with myopathy and facial dysmorphism	AR
MYO7A	276903	Deafness, AD 11;Usher syndrome, type 1B;Deafness, AR 2	AD, AR

MYO9A	604875	Myasthenic syndrome, congenital, 24, presynaptic	AR
NAA10	300013	Ogden syndrome;Microphthalmia, syndromic 1	XLD, XLR, XL
NALCN	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	AD, AR
NBAS	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly;Infantile liver failure syndrome 2	AR
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
NEB	161650	Nemaline myopathy 2, AR;Arthrogryposis multiplex congenita 6	AR
NECTIN 1	600644	Orofacial cleft 7;Cleft lip/palate-ectodermal dysplasia syndrome	AR

NEK1	604588	Short-rib thoracic dysplasia 6 with or without polydactyly;Amyotrophic lateral sclerosis, susceptibility to, 24	AR, DR, AD
NEK8	609799	Renal-hepatic-pancreatic dysplasia 2;?Nephronophthisis 9	AR
NEK9	609798	?Arthrogryposis, Perthes disease, and upward gaze palsy;Nevus comedonicus, somatic;Lethal congenital contracture syndrome 10	AR
NEPRO	617089	Anauxetic dysplasia 3	AR
NF1	613113	Watson syndrome;Leukemia, juvenile myelomonocytic;Neurofibromatosis, type 1;Neurofibromatosis, familial spinal;Neurofibromatosis-Noonan syndrome	AD, AD, SM
NF2	607379	Neurofibromatosis, type 2;Schwannomatosis, somatic;Meningioma, NF2-related, somatic	AD
NHEJ1	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	
NIPBL	608667	Cornelia de Lange syndrome 1	AD

NKX2-5	600584	Hypoplastic left heart syndrome 2;Hypothyroidism, congenital nongoitrous, 5;Atrial septal defect 7, with or without AV conduction defects;Conotruncal heart malformations, variable;Ventricular septal defect 3;Tetralogy of Fallot	AD
NKX3-2	602183	Spondylo-megaepiphyseal-metaphyseal dysplasia	AR
NME8	607421	Ciliary dyskinesia, primary, 6	AR
NODAL	601265	Heterotaxy, visceral, 5	AD
NOG	602991	Stapes ankylosis with broad thumbs and toes;Brachydactyly, type B2;Tarsal-carpal coalition syndrome;Multiple synostoses syndrome 1;Symphalangism, proximal, 1A	AD
NOTCH 1	190198	Adams-Oliver syndrome 5;Aortic valve disease 1	AD
NOTCH 2	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
NPR2	108961	Acromesomelic dysplasia, Maroteaux type;Short stature with nonspecific skeletal abnormalities;Epiphyseal chondrodysplasia, Miura type	AR, AD
NR2F2	107773	46,XX sex reversal 5;Congenital heart defects, multiple types, 4	AD
NRAS	164790	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Colorectal cancer, somatic;Neurocutaneous melanosis, somatic;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic;Noonan syndrome 6;Melanocytic nevus syndrome, congenital, somatic;Thyroid carcinoma, follicular, somatic;Epidermal nevus, somatic	AD
NRG1	142445	?Schizophrenia, susceptibility to	
NSD1	606681	Sotos syndrome 1	AD

NSDHL	300275	CK syndrome;CHILD syndrome	XLR, XLD
NSUN2	610916	Mental retardation, AR 5	AR
NT5E	129190	Calcification of joints and arteries	AR
OBSL1	610991	3-M syndrome 2	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
OTX2	600037	Retinal dystrophy, early-onset, with or without pituitary dysfunction;Microphthalmia, syndromic 5;Pituitary hormone deficiency, combined, 6	AD
P3H1	610339	Osteogenesis imperfecta, type VIII	AR
P4HB	176790	Cole-Carpenter syndrome 1	AD
PAFAH1B1	601545	Subcortical laminar heterotopia;Lissencephaly 1	AD

PAPSS2	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes	AR
PAX2	167409	Glomerulosclerosis, focal segmental, 7;Papillorenal syndrome	AD
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
PAX7	167410	Myopathy, congenital, progressive, with scoliosis;Rhabdomyosarcoma 2, alveolar	AR, SM
PAX9	167416	Tooth agenesis, selective, 3	AD
PCNT	605925	Microcephalic osteodysplastic primordial dwarfism, type II	AR
PDCD10	609118	Cerebral cavernous malformations 3	AD

PDE4D	600129	Acrodysostosis 2, with or without hormone resistance	AD
PDE6D	602676	Joubert syndrome 22	AR
PEX7	601757	Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata, type 1	AR
PGM1	171900	Congenital disorder of glycosylation, type It	AR
PHEX	300550	Hypophosphatemic rickets, XLD	XLD
PHF6	300414	Borjeson-Forssman-Lehmann syndrome	XLR
PHF8	300560	Mental retardation syndrome, XL, Siderius type	XLR
PHYH	602026	Refsum disease	AR
PIBF1	607532	Joubert syndrome 33	AR
PIEZO2	613629	Arthrogryposis, distal, type 3;?Marden-Walker syndrome;Arthrogryposis, distal, type 5;Arthrogryposis, distal, with impaired proprioception and touch	AD, AR
PIGA	311770	Paroxysmal nocturnal hemoglobinuria, somatic;Multiple congenital anomalies-hypotonia-seizures syndrome 2	XLR
PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1	AR
PIP5K1C	606102	Lethal congenital contractural syndrome 3	AR

PITX2	601542	Anterior segment dysgenesis 4;Ring dermoid of cornea;Axenfeld-Rieger syndrome, type 1	AD
PKD1L1	609721	Heterotaxy, visceral, 8, autosomal	AR
PKD2	173910	Polycystic kidney disease 2	AD
PKHD1	606702	Polycystic kidney disease 4, with or without hepatic disease	AR
PLEKHA7	612686		
PLK4	605031	Microcephaly and chorioretinopathy, AR, 2	AR
PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	AR
PLOD2	601865	Bruck syndrome 2	AR
PLP1	300401	Pelizaeus-Merzbacher disease;Spastic paraplegia 2, XL	XLR
PMM2	601785	Congenital disorder of glycosylation, type Ia	AR
PNKP	605610	Ataxia-oculomotor apraxia 4;Microcephaly, seizures, and developmental delay;?Charcot-Marie-Tooth disease, type 2B2	AR
PNPLA6	603197	Spastic paraplegia 39, AR;Boucher-Neuhauser syndrome;Oliver-McFarlane syndrome;?Laurence-Moon syndrome	AR

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
POMC	176830	Obesity, adrenal insufficiency, and red hair due to POMC deficiency;Obesity, early-onset, susceptibility to	AR, AD, AR, mi
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

POR	124015	Disordered steroidogenesis due to cytochrome P450 oxidoreductase;Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	AR
PORCN	300651	Focal dermal hypoplasia	XLD
PPARG	601487	Carotid intimal medial thickness 1;Obesity, severe;Lipodystrophy, familial partial, type 3;Diabetes, type 2;Insulin resistance, severe, digenic	AD, AR, mi, AD
PPIB	123841	Osteogenesis imperfecta, type IX	AR
PPP1CB	600590	Noonan syndrome-like disorder with loose anagen hair 2	AD
PQBP1	300463	Renpenning syndrome	XLR
PRDM5	614161	Brittle cornea syndrome 2	AR
PREPL	609557	Myasthenic syndrome, congenital, 22	AR
PRKAR1A	188830	Myxoma, intracardiac;Pigmented nodular adrenocortical disease, primary, 1;Carney complex, type 1;Acrodysostosis 1, with or without hormone resistance	AD
PRKG1	176894	Aortic aneurysm, familial thoracic 8	AD

PRPH2	179605	Retinitis pigmentosa 7 and digenic form;Retinitis punctata albescens;Choroidal dystrophy, central areolar 2;Macular dystrophy, patterned, 1;Macular dystrophy, vitelliform, 3;Leber congenital amaurosis 18	AD, AR, DD, AD, AR, AD
PRSS56	613858	Microphthalmia, isolated 6	AR
PTCH1	601309	Basal cell carcinoma, somatic;Holoprosencephaly 7;Basal cell nevus syndrome	AD
PTH1R	168468	Chondrodysplasia, Blomstrand type;Failure of tooth eruption, primary;Eiken syndrome;Metaphyseal chondrodysplasia, Murk Jansen type	AR, AD
PTHLH	168470	Brachydactyly, type E2	AD
PTPN11	176876	Leukemia, juvenile myelomonocytic, somatic;LEOPARD syndrome 1;Metachondromatosis;Noonan syndrome 1	AD
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
RAB18	602207	Warburg micro syndrome 3	AR
RAB23	606144	Carpenter syndrome	AR
RAB3GAP1	602536	Martsolf syndrome 2;Warburg micro syndrome 1	AR
RAB3GAP2	609275	Warburg micro syndrome 2;Martsolf syndrome	AR
RAD21	606462	Cornelia de Lange syndrome 4;?Mungan syndrome	AD, AR
RAF1	164760	Noonan syndrome 5;LEOPARD syndrome 2;Cardiomyopathy, dilated, 1NN	AD
RAPSN	601592	Fetal akinesia deformation sequence 2;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	AR
RARB	180220	Microphthalmia, syndromic 12	AD, AR
RARS2	611524	Pontocerebellar hypoplasia, type 6	AR

RASA1	139150	Basal cell carcinoma, somatic;Capillary malformation-arteriovenous malformation 1	AD
RASA2	601589		
RAX	601881	Microphthalmia, isolated 3	AR
RBBP8	604124	Seckel syndrome 2;Jawad syndrome	AR
RBP4	180250	Microphthalmia, isolated, with coloboma 10;Retinal dystrophy, iris coloboma, and comedogenic acne syndrome	AD, AR
RBPJ	147183	Adams-Oliver syndrome 3	AD
RELN	600514	Lissencephaly 2 (Norman-Roberts type);Epilepsy, familial temporal lobe, 7	AR, 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

RHO	180380	Retinitis punctata albescens;Retinitis pigmentosa 4, AD or recessive;Night blindness, congenital stationary, AD 1	AD, AR
RIN2	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis	AR
RIPK4	605706	Popliteal pterygium syndrome, Bartsocas-Papas type;CHAND syndrome	AR
RIPPLY 2	609891	?Spondylocostal dysostosis 6	AR
RIT1	609591	Noonan syndrome 8	AD
ROBO3	608630	Gaze palsy, familial horizontal, with progressive scoliosis, 1	AR
ROR2	602337	Brachydactyly, type B1;Robinow syndrome, AR	AD, AR
RPGRIP 1L	610937	Joubert syndrome 7;?COACH syndrome 3;Meckel syndrome 5	AR
RPL10	312173	Autism, susceptibility to, XL 5;Mental retardation, XL, syndromic, 35	XLR
RPS6KA 3	300075	Coffin-Lowry syndrome;Mental retardation, XL 19	XLD
RSPH1	609314	Ciliary dyskinesia, primary, 24	AR
RSPH3	615876	Ciliary dyskinesia, primary, 32	AR
RSPH4A	612647	Ciliary dyskinesia, primary, 11	

RSPH9	612648	Ciliary dyskinesia, primary, 12	
RTTN	610436	Microcephaly, short stature, and polymicrogyria with seizures	AR
RUNX2	600211	Cleidocranial dysplasia, forme fruste, with brachydactyly;Cleidocranial dysplasia;Cleidocranial dysplasia, forme fruste, dental anomalies only;Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly	AD
RXYLT1	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	AR
RYR1	180901	Malignant hyperthermia susceptibility 1;King-Denborough syndrome;Central core disease;Neuromuscular disease, congenital, with uniform type 1 fiber;Minicore myopathy with external ophthalmoplegia	AD, AD, AR, AR
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome;Townes-Brocks syndrome 1	AD
SALL4	607343	IVIC syndrome;Duane-radial ray syndrome	AD

SASS6	609321	?Microcephaly 14, primary, AR	AR
SATB2	608148	Glass syndrome	AD
SBDS	607444	Aplastic anemia, susceptibility to;Shwachman-Diamond syndrome	AR
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
SDCCAG8	613524	Bardet-Biedl syndrome 16;Senior-Loken syndrome 7	AR
SEC24D	607186	Cole-Carpenter syndrome 2	AR
SELENON	606210	Muscular dystrophy, rigid spine, 1;Myopathy, congenital, with fiber-type disproportion	AR, AD, AR
SEMA3E	608166	?CHARGE syndrome	AD
SEPSECS	613009	Pontocerebellar hypoplasia type 2D	AR
SERPINF1	172860	Osteogenesis imperfecta, type VI	AR

SERPINH1	600943	Preterm premature rupture of the membranes, susceptibility to;Osteogenesis imperfecta, type X	AR
SF3B4	605593	Acrofacial dysostosis 1, Nager type	AD
SH3PXD2B	613293	Frank-ter Haar syndrome	AR
SHH	600725	Schizencephaly;Microphthalmia with coloboma 5;Single median maxillary central incisor;Holoprosencephaly 3	AD
SHOC2	602775	Noonan syndrome-like with loose anagen hair 1	AD
SHROOM4	300579	Stocco dos Santos XL mental retardation syndrome	XL
SIX3	603714	Holoprosencephaly 2;Schizencephaly	AD
SIX6	606326	Optic disc anomalies with retinal and/or macular dystrophy	AR
SKI	164780	Shprintzen-Goldberg syndrome	AD
SLC18A3	600336	Myasthenic syndrome, congenital, 21, presynaptic	AR
SLC25A19	606521	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type);Microcephaly, Amish type	AR

SLC26A2	606718	Diastrophic dysplasia, broad bone-platyspondylic variant;Achondrogenesis Ib;Epiphyseal dysplasia, multiple, 4;De la Chapelle dysplasia;Diastrophic dysplasia;Atelosteogenesis, type II	AR
SLC2A10	606145	Arterial tortuosity syndrome	AR
SLC34A3	609826	Hypophosphatemic rickets with hypercalciuria	AR
SLC35D1	610804	Schneckenbecken dysplasia	AR
SLC38A8	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis	AR
SLC39A13	608735	Ehlers-Danlos syndrome, spondylodysplastic type, 3	AR
SLC5A7	608761	Neuronopathy, distal hereditary motor, type VIIA;Myasthenic syndrome, congenital, 20, presynaptic	AD, AR
SLC9A6	300231	Mental retardation, XL syndromic, Christianson type	XL

SMAD2	601366	Loeys-Dietz syndrome 6;Congenital heart defects, multiple types, 8, with or without heterotaxy	AD
SMAD3	603109	Loeys-Dietz syndrome 3	AD
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;Pancreatic cancer, somatic;Myhre syndrome;Polyposis, juvenile intestinal	AD
SMAD6	602931	Craniosynostosis 7, susceptibility to;Aortic valve disease 2;Radioulnar synostosis, nonsyndromic	AD
SMARCA2	600014	Blepharophimosis-impaired intellectual development syndrome;Nicolaides-Baraitser syndrome	AD
SMARCA4	603254	Coffin-Siris syndrome 4;Rhabdoid tumor predisposition syndrome 2	AD
SMARCAL1	606622	Schimke immunoosseous dysplasia	AR
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
SMCHD1	614982	Bosma arhinia microphthalmia syndrome;Fascioscapulohumeral muscular dystrophy 2, digenic	AD, DD
SMOC1	608488	Microphthalmia with limb anomalies	AR
SMS	300105	Mental retardation, XL, Snyder-Robinson type	XLR
SNAI2	602150	Waardenburg syndrome, type 2D;Piebaldism	AR, AD
SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	AR
SNTG1	608714		
SOS1	182530	Noonan syndrome 4;?Fibromatosis, gingival, 1	AD
SOS2	601247	Noonan syndrome 9	AD

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
SOX9	608160	Acampomelic campomelic dysplasia;Campomelic dysplasia;Campomelic dysplasia with autosomal sex reversal	AD
SP7	606633	Osteogenesis imperfecta, type XII	AR
SPAG1	603395	Ciliary dyskinesia, primary, 28	AR
SPARC	182120	Osteogenesis imperfecta, type XVII	AR
SPECC1L	614140	Opitz GBBB syndrome, type II;?Facial clefting, oblique, 1;Hypertelorism, Teebi type	AD
SPRED1	609291	Legius syndrome	AD
SPRY4	607984	Hypogonadotropic hypogonadism 17 with or without anosmia	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
SUFU	607035	Meningioma, familial, susceptibility to;Basal cell nevus syndrome;Medulloblastoma, desmoplastic;Joubert syndrome 32	AD, AD, AR, SM, AR
SYT2	600104	Myasthenic syndrome, congenital, 7B, presynaptic, AR;Myasthenic syndrome, congenital, 7, presynaptic	AR, AD
TAF6	602955	Alazami-Yuan syndrome	AR
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

TBX1	602054	Velocardiofacial syndrome;Tetralogy of Fallot;DiGeorge syndrome;Conotruncal anomaly face syndrome	AD
TBX15	604127	Cousin syndrome	AR
TBX2	600747	Vertebral anomalies and variable endocrine and T-cell dysfunction	AD
TBX22	300307	?Abruzzo-Erickson syndrome;Cleft palate with ankyloglossia	XL
TBX3	601621	Ulnar-mammary syndrome	AD
TBX5	601620	Holt-Oram syndrome	AD
TBX6	602427	Spondylocostal dysostosis 5	AD, AR
TCF12	600480	Craniosynostosis 3;Hypogonadotropic hypogonadism 26 with or without anosmia	AD, AD, AR
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

TENM3	610083	Microphthalmia, syndromic 15;?Microphthalmia, isolated, with coloboma 9	AR
TENT5A	611357	Osteogenesis imperfecta, type XVIII	AR
TFAP2A	107580	Branchiooculofacial syndrome	AD
TGDS	616146	Catel-Manzke syndrome	AR
TGFB1	190180	Camurati-Engelmann disease;Cystic fibrosis lung disease, modifier of;Inflammatory bowel disease, immunodeficiency, and encephalopathy	AD, AR
TGFB2	190220	Loeys-Dietz syndrome 4	AD
TGFB3	190230	Loeys-Dietz syndrome 5;Arrhythmogenic right ventricular dysplasia 1	AD
TGFBR1	190181	Loeys-Dietz syndrome 1;Multiple self-healing squamous epithelioma, susceptibility to	AD
TGFBR2	190182	Colorectal cancer, hereditary nonpolyposis, type 6;Loeys-Dietz syndrome 2;Esophageal cancer, somatic	AD
TGIF1	602630	Holoprosencephaly 4	AD
TK2	188250	Mi DNA depletion syndrome 2 (myopathic type);?Progressive external ophthalmoplegia with Mi DNA deletions, AR 3	AR

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
TMEM70	612418	Mi complex V (ATP synthase) deficiency, nuclear type 2	AR
TMTC3	617218	Lissencephaly 8	AR
TNFRSF11B	602643	Paget disease of bone 5, juvenile-onset	AR
TNNI2	191043	Arthrogryposis, distal, type 2B1	AD
TNNT1	191041	Nemaline myopathy 5, Amish type	AR
TNNT3	600692	Arthrogryposis, distal, type 2B2	AD
TNXB	600985	Vesicoureteral reflux 8;Ehlers-Danlos syndrome, classic-like, 1	AD, AR
TOE1	613931	Pontocerebellar hypoplasia, type 7	AR

TOR1A	605204	Dystonia-1, torsion;Arthrogryposis multiplex congenita 5	AD, AR
TP63	603273	Rapp-Hodgkin syndrome;Orofacial cleft 8;Limb-mammary syndrome;Split-hand/foot malformation 4;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3;Hay-Wells syndrome;ADULT syndrome	AD
TPM2	190990	Arthrogryposis, distal, type 2B4;Arthrogryposis, distal, type 1A;CAP myopathy 2;Nemaline myopathy 4, AD	AD
TPM3	191030	Myopathy, congenital, with fiber-type disproportion;Nemaline myopathy 1, AD or recessive;CAP myopathy 1	AD, AR
TRAF3IP1	607380	Senior-Loken syndrome 9	AR
TRIM32	602290	Muscular dystrophy, limb-girdle, AR 8;?Bardet-Biedl syndrome 11	AR
TRIP11	604505	Achondrogenesis, type IA;Osteochondrodysplasia	AR
TRMT10A	616013	Microcephaly, short stature, and impaired glucose metabolism 1	AR

TRPS1	604386	Trichorhinophalangeal syndrome, type I;Trichorhinophalangeal syndrome, type III	AD
TRPV4	605427	SED, Maroteaux type;Spondylometaphyseal dysplasia, Kozlowski type;Metatropic dysplasia;Brachyolmia type 3;Neuronopathy, distal hereditary motor, type VIII;[Sodium serum level QTL 1];?Avascular necrosis of femoral head, primary, 2;Scapuloperoneal spinal muscular atrophy;Parastremmatic dwarfism;Hereditary motor and sensory neuropathy, type IIc;Digital arthropathy-brachydactyly, familial	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

TSEN54	608755	Pontocerebellar hypoplasia type 2A;?Pontocerebellar hypoplasia type 5;Pontocerebellar hypoplasia type 4	AR
TTC21B	612014	Short-rib thoracic dysplasia 4 with or without polydactyly;Nephronophthisis 12	AR, AD, AR
TTC8	608132	Bardet-Biedl syndrome 8;?Retinitis pigmentosa 51	AR
TUBA1A	602529	Lissencephaly 3	AD
TUBA8	605742	Cortical dysplasia, complex, with other brain malformations 8	AR
TUBB	191130	Symmetric circumferential skin creases, congenital, 1;Cortical dysplasia, complex, with other brain malformations 6	AD
TUBB2A	615101	Cortical dysplasia, complex, with other brain malformations 5	AD
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

TUBG1	191135	Cortical dysplasia, complex, with other brain malformations 4	AD
TUBGCP4	609610	Microcephaly and chorioretinopathy, AR, 3	AR
TUBGCP6	610053	Microcephaly and chorioretinopathy, AR, 1	AR
TWIST1	601622	Craniosynostosis 1;Robinow-Sorauf syndrome;Saethre-Chotzen syndrome with or without eyelid anomalies;Sweeney-Cox syndrome	AD
TXNL4A	611595	Burn-McKeown syndrome	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
UBA1	314370	VEXAS syndrome, somatic;Spinal muscular atrophy, XL 2, infantile	XLR
VAMP1	185880	Myasthenic syndrome, congenital, 25;Spastic ataxia 1, AD	AR, AD

VCAN	118661	Wagner syndrome 1	AD
VIPAS39	613401	Arthrogryposis, renal dysfunction, and cholestasis 2	AR
VLDLR	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	AR
VPS13B	607817	Cohen syndrome	AR
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1	AR
VPS53	615850	Pontocerebellar hypoplasia, type 2E	AR
VRK1	602168	Pontocerebellar hypoplasia type 1A	AR
VSX2	142993	Microphthalmia, isolated 2;Microphthalmia with coloboma 3	
WDPCP	613580	Congenital heart defects, hamartomas of tongue, and polysyndactyly;?Bardet-Biedl syndrome 15	AR
WDR11	606417	Hypogonadotropic hypogonadism 14 with or without anosmia	AD
WDR19	608151	?Cranioectodermal dysplasia 4;?Short-rib thoracic dysplasia 5 with or without polydactyly;Nephronophthisis 13;Senior-Loken syndrome 8	AR

WDR35	613602	Cranioectodermal dysplasia 2;Short-rib thoracic dysplasia 7 with or without polydactyly	AR
WDR62	613583	Microcephaly 2, primary, AR, with or without cortical malformations	AR
WNT1	164820	Osteogenesis imperfecta, type XV;Osteoporosis, early-onset, susceptibility to, AD	AR
WNT5A	164975	Robinow syndrome, AD 1	AD
WNT7A	601570	Fuhrmann syndrome;Ulna and fibula, absence of, with severe limb deficiency	AR
XYLT1	608124	Desbuquois dysplasia 2;Pseudoxanthoma elasticum, modifier of severity of	AR
YWHAE	605066		
ZBTB24	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	AR
ZC4H2	300897	Wieacker-Wolff syndrome;Wieacker-Wolff syndrome, female-restricted	XLR, XLD
ZEB2	605802	Mowat-Wilson syndrome	AD
ZFPM2	603693	Tetralogy of Fallot;Diaphragmatic hernia 3;46XY sex reversal 9	AD

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
ZMYND 10	607070	Ciliary dyskinesia, primary, 22	AR
ZNF335	610827	Microcephaly 10, primary, AR	AR
ZNF423	604557	Joubert syndrome 19;Nephronophthisis 14	AD, AR
ZNF469	612078	Brittle cornea syndrome 1	AR
ZNF699	609571	DEGCAGS syndrome	AR
ZSWIM6	615951	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features;Acromelic frontonasal dysostosis	AD