Panel Nefrología Plus

Si se sospecha de poliquistosis renal recomendamos Panel Nephro Plus, ya que incluye todos los genes analizados en Panel Nephro pero además incluye el análisis del gen PKD1.

Nº de genes:	496
Entrega:	25 días
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥150 x
Detalles:	El análisis de CNV incluye el análisis de PDKD1

Subpaneles incluidos

Panel para el síndrome de Alport

Panel para Bardet-Biedl

Panel para el síndrome de Bartter

Panel para discinesia ciliar (primaria)

Panel para la deficiencia combinada de hormonas hipofisarias

Panel para glomerulonefrosis focal Panel para heterotaxia

Panel para colestasis intrahepática

Panel para el síndrome de Joubert

Panel para el síndrome de Kallmann y Hipogonadismo hipogonadotrópico

Panel para la amaurosis congénita de Leber

Panel para el síndrome de Meckel Panel para nefronoptisis

Panel para el síndrome nefrótico

Panel para hepatopatías mitocondriales neonatales

Panel para la enfermedad poliquística renal

Panel para pseudohipoaldosteronismo

Panel para acidosis tubular renal

Panel para ciliopatías con afectación esquelética extensa

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

Genes	OMIM (Gen)	Enfermedades asociadas (OMIM)	Herencia
ABCB11	603201	Cholestasis, benign recurrent intrahepatic, 2;Cholestasis, progressive familial intrahepatic 2	AR
ABCB4	171060	Gallbladder disease 1;Cholestasis, progressive familial intrahepatic 3;Cholestasis, intrahepatic, of pregnancy, 3	AD, AR, AR
ABCC2	601107	Dubin-Johnson syndrome	AR
ACE	106180	Microvascular complications of diabetes 3;Stroke, hemorrhagic;Renal tubular dysgenesis	AR
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation	AR
ACTG2	102545	Visceral myopathy;Megacystis-microcolon-intestinal hypoperistalsis syndrome 5	AD
ACTN4	604638	Glomerulosclerosis, focal segmental, 1	AD
ACVR2B	602730	Heterotaxy, visceral, 4, autosomal	
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3	AR
AGT	106150	Hypertension, essential, susceptibility to;Renal tubular dysgenesis	MF, AR
AGTR1	106165	Hypertension, essential;Renal tubular dysgenesis	MF, AR
AHI1	608894	Joubert syndrome 3	AR
AIPL1	604392	Cone-rod dystrophy;Leber congenital amaurosis 4;Retinitis pigmentosa, juvenile	AD, AR
AKR1D1	604741	Bile acid synthesis defect, congenital, 2	AR
ALDOB	612724	Fructose intolerance, hereditary	AR

ALG8	608103	Polycystic liver disease 3 with or without kidney cysts;Congenital disorder of glycosylation, type Ih	AD, AR
ALG9	606941	Congenital disorder of glycosylation, type Il;Gillessen-Kaesbach-Nishimura syndrome	AR
ALPL	171760	Hypophosphatasia, infantile;Odontohypophosphatasia;Hypophosphatasia, childhood;Hypophosphatasia, adult	AR, AD, AR
AMER1	300647	Osteopathia striata with cranial sclerosis	XLD
ANKH	605145	Chondrocalcinosis 2;Craniometaphyseal dysplasia	AD
ANKS6	615370	Nephronophthisis 16	AR
ANLN	616027	Focal segmental glomerulosclerosis 8	AD
ANO5	608662	Gnathodiaphyseal dysplasia;Muscular dystrophy, limb-girdle, autosomal recessive 12;Miyoshi muscular dystrophy 3	AD, AR
ANOS1	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)	XLR
AP2S1	602242	Hypocalciuric hypercalcemia, type III	AD
ARHGAP31	610911	Adams-Oliver syndrome 1	AD
ARHGDIA	601925	Nephrotic syndrome, type 8	AR
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
ARMC5	615549	ACTH-independent macronodular adrenal hyperplasia 2	AD, SM
ARMC9	617612	Joubert syndrome 30	AR
ARSL	300180	Chondrodysplasia punctata, XLR	XLR
ATP6V0A4	605239	Distal renal tubular acidosis 3, with or without sensorineural hearing loss	AR
ATP6V1B1	192132	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	AR
ATP8B1	602397	Cholestasis, progressive familial intrahepatic 1;Cholestasis, intrahepatic, of pregnancy, 1;Cholestasis, benign recurrent intrahepatic	AR, AD
ATR	601215	?Cutaneous telangiectasia and cancer syndrome, familial;Seckel syndrome 1	AD, AR
AVPR2	300538	Nephrogenic syndrome of inappropriate antidiuresis;Diabetes insipidus, nephrogenic	XLR
B9D1	614144	?Meckel syndrome 9;Joubert syndrome 27	AR
B9D2	611951	?Meckel syndrome 10;Joubert syndrome 34	AR

BAAT	602938	Hypercholanemia, familial;Bile acid conjugation defect 1	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
BCS1L	603647	GRACILE syndrome;Bjornstad syndrome;MT complex III deficiency, nuclear type 1	AR
BICC1	614295	Renal dysplasia, cystic, susceptibility to	AD
BMP1	112264	Osteogenesis imperfecta, type XIII	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
BSND	606412	Bartter syndrome, type 4a;Sensorineural deafness with mild renal dysfunction	AR
C2CD3	615944	Orofaciodigital syndrome XIV	AR
CA2	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	AR
CABP4	608965	Cone-rod synaptic disorder, congenital nonprogressive	AR
CANT1	613165	Desbuquois dysplasia 1;Epiphyseal dysplasia, multiple, 7	AR
CASP10	601762	Autoimmune lymphoproliferative syndrome, type II;Gastric cancer, somatic;Lymphoma, non-Hodgkin, somatic	AD
CASR	601199	Epilepsy idiopathic generalized, susceptibility to, 8;Hypocalcemia, AD, with Bartter syndrome;Hypocalciuric hypercalcemia, type I;Hyperparathyroidism, neonatal;Hypocalcemia, AD	AD, AD, 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
CCN6	603400	Progressive pseudorheumatoid dysplasia	AR
CCNO	607752	Ciliary dyskinesia, primary, 29	AR
CD2AP	604241	Glomerulosclerosis, focal segmental, 3	
CDKN1C	600856	IMAGE syndrome;Beckwith-Wiedemann syndrome	AD
CENPF	600236	Stromme syndrome	AR
CENPJ	609279	?Seckel syndrome 4;Microcephaly 6, primary, AR	AR
CEP120	613446	Joubert syndrome 31;Short-rib thoracic dysplasia 13 with or without polydactyly	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
CEP55	610000	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly	AR
CEP83	615847	Nephronophthisis 18	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
CFTR	602421	Congenital bilateral absence of vas deferens;Pancreatitis, hereditary;Bronchiectasis with or without elevated sweat chloride 1, modifier of;Cystic fibrosis	AR, AD
CHD1L	613039		
CHD7	608892	CHARGE syndrome;Hypogonadotropic hypogonadism 5 with or without anosmia	AD
CHRNA3	118503	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT;Lung cancer susceptibility 2	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
CLCN5	300008	Nephrolithiasis, type I;Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis;Dent disease;Hypophosphatemic rickets	XLR
CLCNKA	602024	Bartter syndrome, type 4b, digenic	DR

CLCNKB	602023	Bartter syndrome, type 3;Bartter syndrome, type 4b, digenic	AR, DR
CLDN16	603959	Hypomagnesemia 3, renal	AR
CLDN19	610036	Hypomagnesemia 5, renal, with ocular involvement	AR
COL10A1	120110	Metaphyseal chondrodysplasia, Schmid type	AD
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
COL4A3	120070	Alport syndrome 3, AD;Hematuria, benign familial;Alport syndrome 2, AR	AD, AR
COL4A4	120131	Alport syndrome 2, AR;Hematuria, familial benign	AR, AD
COL4A5	303630	Alport syndrome 1, XL	XLD
COL9A3	120270	Epiphyseal dysplasia, multiple, 3, with or without myopathy;Intervertebral disc disease, susceptibility to	AD
COMP	600310	Carpal tunnel syndrome 2;Epiphyseal dysplasia, multiple, 1;Pseudoachondroplasia	AD
COQ2	609825	Coenzyme Q10 deficiency, primary, 1;Multiple system atrophy, susceptibility to	AR, AD, AR
COQ6	614647	Coenzyme Q10 deficiency, primary, 6	AR
COQ8B	615567	Nephrotic syndrome, type 9	AR
COQ9	612837	Coenzyme Q10 deficiency, primary, 5	AR
CPLANE1	614571	Orofaciodigital syndrome VI;Joubert syndrome 17	AR
CRB1	604210	Leber congenital amaurosis 8;Retinitis pigmentosa-12;Pigmented paravenous chorioretinal atrophy	AR, AD
CRB2	609720	Focal segmental glomerulosclerosis 9;Ventriculomegaly with cystic kidney disease	AR
CRELD1	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome;Atrioventricular septal defect, susceptibility to, 2	AD
CRTAP	605497	Osteogenesis imperfecta, type VII	AR
CRX	602225	Leber congenital amaurosis 7;Cone-rod retinal dystrophy-2	AD
CSPP1	611654	Joubert syndrome 21	AR

CTNS	606272	Cystinosis, late-onset juvenile or adolescent nephropathic;Cystinosis, ocular nonnephropathic;Cystinosis, nephropathic;Cystinosis, atypical nephropathic	AR
CTU2	617057	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome	AR
CUBN	602997	[Proteinuria, chronic benign];Imerslund-Grasbeck syndrome 1	AR
CUL3	603136	Pseudohypoaldosteronism, type IIE;Neurodevelopmental disorder with or without autism or seizures	AD
CWC27	617170	Retinitis pigmentosa with or without skeletal anomalies	AR
CYP7B1	603711	Spastic paraplegia 5A, AR;Bile acid synthesis defect, congenital, 3	AR
DCDC2	605755	Nephronophthisis 19;?Deafness, autosomal recessive 66;Sclerosing cholangitis, neonatal	AR
DDR2	191311	Spondylometaepiphyseal dysplasia, short limb-hand type;Warburg-Cinotti syndrome	AR, AD
DDX59	615464	Orofaciodigital syndrome V	AR
DGKE	601440	Nephrotic syndrome, type 7;Hemolytic uremic syndrome, atypical, susceptibility to, 7	AR
DGUOK	601465	Portal hypertension, noncirrhotic;Progressive external ophthalmoplegia with MT DNA deletions, autosomal recessive 4;MT DNA depletion syndrome 3 (hepatocerebral type)	AR
DHCR7	602858	Smith-Lemli-Opitz syndrome	AR
DICER1	606241	Pleuropulmonary blastoma;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors;GLOW syndrome, somatic mosaic;Rhabdomyosarcoma, embryonal, 2	AD
DLL3	602768	Spondylocostal dysostosis 1, AR	AR
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
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
DNAJB11	611341	Polycystic kidney disease 6 with or without polycystic liver disease	AD
DNAL1	610062	Ciliary dyskinesia, primary, 16	AR
DNAAF11	614930	Ciliary dyskinesia, primary, 19	AR
DRC1	615288	Ciliary dyskinesia, primary, 21	AR
DSTYK	612666	Spastic paraplegia 23;Congenital anomalies of kidney and urinary tract 1	AR, AD
DUSP6	602748	Hypogonadotropic hypogonadism 19 with or without anosmia	AD
DYM	607461	Dyggve-Melchior-Clausen disease;Smith-McCort dysplasia	AR
DYNC2H1	603297	Short-rib thoracic dysplasia 3 with or without polydactyly	AR, DR
DYNC2LI1	617083	Short-rib thoracic dysplasia 15 with polydactyly	AR
DYNC2I1	615462	Short-rib thoracic dysplasia 8 with or without polydactyly	AR
DYNC2I2	613363	Short-rib thoracic dysplasia 11 with or without polydactyly	AR
DZIP1L	617570	Polycystic kidney disease 5	AR
EBP	300205	MEND syndrome;Chondrodysplasia punctata, XLD	XLR, XLD
EIF2AK3	604032	Wolcott-Rallison syndrome	AR
EMP2	602334	Nephrotic syndrome, type 10	AR
ENPP1	173335	Arterial calcification, generalized, of infancy, 1;Obesity, susceptibility to;Hypophosphatemic rickets, autosomal recessive, 2;Cole disease;Diabetes mellitus, non-insulin-dependent, susceptibility to	AR, AD, AR, MF, 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
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
FAH	613871	Tyrosinemia, type I	AR
FAM20C	611061	Raine syndrome	AR
FAN1	613534	Interstitial nephritis, karyomegalic	AR
FAS	134637	Autoimmune lymphoproliferative syndrome, type IA;Autoimmune lymphoproliferative syndrome	AD
FASLG	134638	Autoimmune lymphoproliferative syndrome, type IB;Lung cancer, susceptibility to	AD, AD, SM
FAT4	612411	Hennekam lymphangiectasia-lymphedema syndrome 2;Van Maldergem syndrome 2	AR
FEZF1	613301	Hypogonadotropic hypogonadism 22, with or without anosmia	AR
FGF17	603725	Hypogonadotropic hypogonadism 20 with or without anosmia	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
FKBP10	607063	Osteogenesis imperfecta, type XI;Bruck syndrome 1	AR
FLNB	603381	Larsen syndrome;Boomerang dysplasia;Spondylocarpotarsal synostosis syndrome;Atelosteogenesis, type III;Atelosteogenesis, type I	AD, AR
FLRT3	604808	Hypogonadotropic hypogonadism 21 with anosmia	AD

FN1	135600	Glomerulopathy with fibronectin deposits 2;Spondylometaphyseal dysplasia, corner fracture type	AD
FOXP1	605515	Mental retardation with language impairment and with or without autistic features	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
FSHB	136530	Hypogonadotropic hypogonadism 24 without anosmia	AR
FXYD2	601814	Hypomagnesemia 2, renal	AD
GANAB	104160	Polycystic kidney disease 3	AD
GATA3	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	AD
GDF1	602880	Congenital heart defects, multiple types, 6;Right atrial isomerism (Ivemark)	AD, AR
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
GFM1	606639	Combined oxidative phosphorylation deficiency 1	AR
GHR	600946	Hypercholesterolemia, familial, modifier of;Laron dwarfism;Growth hormone insensitivity, partial;Increased responsiveness to growth hormone	AD, AR, AR, AD
GLA	300644	Fabry disease;Fabry disease, cardiac variant	XL
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
GLIS2	608539	Nephronophthisis 7	
GLIS3	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism	AR
GNA11	139313	Hypocalciuric hypercalcemia, type II;Hypocalcemia, AD 2	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
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2	AR
GNRH1	152760	?Hypogonadotropic hypogonadism 12 with or without anosmia	AR
GNRHR	138850	Hypogonadotropic hypogonadism 7 without anosmia	AR
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1;Wilms tumor, somatic	XLR
GPC6	604404	Omodysplasia 1	AR
GREB1L	617782	Deafness, AD 80;Renal hypodysplasia/aplasia 3	AD
GRIP1	604597	Fraser syndrome 3	AR
GUCY2D	600179	Cone-rod dystrophy 6;Leber congenital amaurosis 1;Night blindness, congenital stationary, type 1I;?Choroidal dystrophy, central areolar 1	AD, AR, AR, AD
HAAO	604521	Vertebral, cardiac, renal, and limb defects syndrome 1	AR
HAMP	606464	Hemochromatosis, type 2B	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
HFE	613609	Porphyria variegata, susceptibility to;Alzheimer disease, susceptibility to;Hemochromatosis;[Transferrin serum level QTL2];Porphyria cutanea tarda, susceptibility to;Microvascular complications of diabetes 7	AD, AR, AD, AR
HNF1B	189907	Renal cysts and diabetes syndrome;Diabetes mellitus, noninsulin-dependent;Renal cell carcinoma	AD
HNF4A	600281	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young;Diabetes mellitus, noninsulin-dependent;MODY, type I	AD
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
HPSE2	613469	Urofacial syndrome 1	AR
HS6ST1	604846	Hypogonadotropic hypogonadism 15 with or without anosmia	AD
HSD11B2	614232	Apparent mineralocorticoid excess	AR
HSD3B7	607764	Bile acid synthesis defect, congenital, 1	AR
HSPG2	142461	Schwartz-Jampel syndrome, type 1;Dyssegmental dysplasia, Silverman-Handmaker type	AR
HYDIN	610812	Ciliary dyskinesia, primary, 5	AR
HYLS1	610693	Hydrolethalus 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
IHH	600726	Brachydactyly, type A1;Acrocapitofemoral dysplasia	AD, AR
IL17RD	606807	Hypogonadotropic hypogonadism 18 with or without anosmia	AD, AR, DD
IMPDH1	146690	Leber congenital amaurosis 11;Retinitis pigmentosa 10	AD
INF2	610982	Glomerulosclerosis, focal segmental, 5;Charcot-Marie-Tooth disease, dominant intermediate E	AD
INPP5E	613037	Mental retardation, truncal obesity, retinal dystrophy, and micropenis;Joubert syndrome 1	AR
INPPL1	600829	Opsismodysplasia	AR
INVS	243305	Nephronophthisis 2, infantile	AR
IQCB1	609237	Senior-Loken syndrome 5	AR
ITGA3	605025	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	AR

ITGA8	604063	Renal hypodysplasia/aplasia 1	AR
JAG1	601920	Alagille syndrome 1;Charcot-Marie-Tooth disease, axonal, type 2HH;?Deafness, congenital heart defects, and posterior embryotoxon;Tetralogy of Fallot	AD
KANK2	614610	Nephrotic syndrome, type 16;Palmoplantar keratoderma and woolly hair	AR
KCNJ1	600359	Bartter syndrome, type 2	AR
KCNJ10	602208	Enlarged vestibular aqueduct, digenic;SESAME syndrome	AR
KCNJ13	603208	Snowflake vitreoretinal degeneration;Leber congenital amaurosis 16	AD, AR
KCNJ5	600734	Hyperaldosteronism, familial, type III;Long QT syndrome 13	AD
KDM6A	300128	Kabuki syndrome 2	XLD
KIAA0586	610178	Joubert syndrome 23;Short-rib thoracic dysplasia 14 with polydactyly	AR
KIF14	611279	?Meckel syndrome 12;Microcephaly 20, primary, AR	AR
KIF22	603213	Spondyloepimetaphyseal dysplasia with joint laxity, type 2	AD
KIF7	611254	Joubert syndrome 12;?Hydrolethalus syndrome 2;?Al-Gazali-Bakalinova syndrome;Acrocallosal syndrome	AR
KISS1	603286	?Hypogonadotropic hypogonadism 13 with or without anosmia	AR
KISS1R	604161	?Precocious puberty, central, 1;Hypogonadotropic hypogonadism 8 with or without anosmia	AD, AR
KLHL3	605775	Pseudohypoaldosteronism, type IID	AD, AR
KMT2D	602113	Kabuki syndrome 1	AD
KYNU	605197	?Hydroxykynureninuria;Vertebral, cardiac, renal, and limb defects syndrome 2	AR
LAGE3	300060	Galloway-Mowat syndrome 2, XL	XLR
LAMB2	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities;Pierson syndrome	AR
LBR	600024	?Reynolds syndrome;Greenberg skeletal dysplasia;Pelger-Huet anomaly;Pelger-Huet anomaly with mild skeletal anomalies	AD, AR
LCA5	611408	Leber congenital amaurosis 5	AR
LCAT	606967	Norum disease;Fish-eye disease	AR
LCT	603202	Lactase deficiency, congenital	AR
LEP	164160	Obesity, morbid, due to leptin deficiency	AR
LEPR	601007	Obesity, morbid, due to leptin receptor deficiency	AR

LHB	152780	Hypogonadotropic hypogonadism 23 with or without anosmia	AR
LHX3	600577	Pituitary hormone deficiency, combined, 3	AR
LHX4	602146	Pituitary hormone deficiency, combined, 4	AD
LIFR	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	AR
LMF1	611761	Lipase deficiency, combined	AR
LMX1B	602575	Nail-patella syndrome;Focal segmental glomerulosclerosis 10	AD
LRAT	604863	Leber congenital amaurosis 14;Retinal dystrophy, early-onset severe;Retinitis pigmentosa, juvenile	AR
LRIG2	608869	Urofacial syndrome 2	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
LZTFL1	606568	Bardet-Biedl syndrome 17	AR
MAFB	608968	Duane retraction syndrome 3;Multicentric carpotarsal osteolysis syndrome	AD
MAGI2	606382	Nephrotic syndrome, type 15	AR
MAPKBP1	616786	Nephronophthisis 20	AR
MATN3	602109	?Spondyloepimetaphyseal dysplasia, Borochowitz Cormier-Daire type;Epiphyseal dysplasia, multiple, 5;Osteoarthritis susceptibility 2	AR, AD
MCEE	608419	Methylmalonyl-CoA epimerase deficiency	AR
MERTK	604705	Retinitis pigmentosa 38	AR
MESP2	605195	Spondylocostal dysostosis 2, AR	AR
MGP	154870	Keutel syndrome	AR
MKKS	604896	McKusick-Kaufman syndrome;Bardet-Biedl syndrome 6	AR
MKS1	609883	Bardet-Biedl syndrome 13;Joubert syndrome 28;Meckel syndrome 1	AR
MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	AR
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	AR

MMADHC	611935	Methylmalonic aciduria and homocystinuria, cblD type;Methylmalonic aciduria, cblD type, variant 2;Homocystinuria, cblD type, variant 1	AR
MMP13	600108	Metaphyseal anadysplasia 1;Metaphyseal dysplasia, Spahr type;?Spondyloepimetaphyseal dysplasia, Missouri type	AD, AR
MMP21	608416	Heterotaxy, visceral, 7, autosomal	AR
MMP9	120361	Metaphyseal anadysplasia 2	
MMUT	609058	Methylmalonic aciduria, mut(0) type	AR
MPV17	137960	Charcot-Marie-Tooth disease, axonal, type 2EE;MT DNA depletion syndrome 6 (hepatocerebral type)	AR
MUC1	158340	Medullary cystic kidney disease 1	AD
MYH9	160775	Deafness, AD 17;Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	AD
MYO1E	601479	Glomerulosclerosis, focal segmental, 6	AR
MYO5B	606540	Microvillus inclusion disease	AR
MYO7A	276903	Deafness, AD 11;Usher syndrome, type 1B;Deafness, autosomal recessive 2	AD, AR
MYOCD	606127	Megabladder, congenital	AD
NADSYN1	608285	Vertebral, cardiac, renal, and limb defects syndrome 3	AR
NBAS	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly;Infantile liver failure syndrome 2	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
NEUROG3	604882	Diarrhea 4, malabsorptive, congenital	AR
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

NMNAT1	608700	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis;Leber congenital amaurosis 9	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
NOTCH2	600275	Alagille syndrome 2;Hajdu-Cheney syndrome	AD
NPC1	607623	Niemann-Pick disease, type C1;Niemann-Pick disease, type D	AR
NPC2	601015	Niemann-pick disease, type C2	AR
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
NPHP4	607215	Senior-Loken syndrome 4;Nephronophthisis 4	AR
NPHS1	602716	Nephrotic syndrome, type 1	AR
NPHS2	604766	Nephrotic syndrome, type 2	AR
NPR2	108961	Acromesomelic dysplasia, Maroteaux type;Short stature with nonspecific skeletal abnormalities;Epiphyseal chondrodysplasia, Miura type	AR, AD
NR0B1	300473	Adrenal hypoplasia, congenital;46XY sex reversal 2, dosage-sensitive	XLR, XL
NR0B2	604630	Obesity, mild, early-onset	AD, AR, MF
NR1H4	603826	Cholestasis, progressive familial intrahepatic, 5	AR
NR3C2	600983	Hypertension, early-onset, AD, with exacerbation in pregnancy;Pseudohypoaldosteronism type I, AD	AD
NSDHL	300275	CK syndrome;CHILD syndrome	XLR, XLD
NSMF	608137	Hypogonadotropic hypogonadism 9 with or without anosmia	AD
NUP107	607617	Galloway-Mowat syndrome 7;Nephrotic syndrome, type 11;?Ovarian dysgenesis 6	AR
NUP93	614351	Nephrotic syndrome, type 12	AR
OBSL1	610991	3-M syndrome 2	AR
OCRL	300535	Lowe syndrome;Dent disease 2	XLR
ODAD1	615038	Ciliary dyskinesia, primary, 20	AR
ODAD2	615408	Ciliary dyskinesia, primary, 23	AR
ODAD3	615956	Ciliary dyskinesia, primary, 30	AR

OFD1	300170	Joubert syndrome 10;Simpson-Golabi-Behmel syndrome, type 2;?Retinitis pigmentosa 23;Orofaciodigital syndrome I	XLR, XLD
OSGEP	610107	Galloway-Mowat syndrome 3	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
PAPSS2	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes	AR
PAX2	167409	Glomerulosclerosis, focal segmental, 7;Papillorenal syndrome	AD
PBX1	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay	AD
PCSK1	162150	Obesity with impaired prohormone processing;Obesity, susceptibility to, BMIQ12	AR
PDE4D	600129	Acrodysostosis 2, with or without hormone resistance	AD
PDE6D	602676	Joubert syndrome 22	AR
PDSS2	610564	Coenzyme Q10 deficiency, primary, 3	AR
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
PEX12	601758	Peroxisome biogenesis disorder 3B;Peroxisome biogenesis disorder 3A (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
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
PHEX	300550	Hypophosphatemic rickets, XLD	XLD
PHF6	300414	Borjeson-Forssman-Lehmann syndrome	XLR
PIBF1	607532	Joubert syndrome 33	AR

PKD1	601313	Polycystic kidney disease 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
PLCE1	608414	Nephrotic syndrome, type 3	AR
PLOD2	601865	Bruck syndrome 2	AR
PMM2	601785	Congenital disorder of glycosylation, type Ia	AR
PNPLA6	603197	Spastic paraplegia 39, AR;Boucher-Neuhauser syndrome;Oliver-McFarlane syndrome;?Laurence-Moon syndrome	AR
POLG	174763	Progressive external ophthalmoplegia, autosomal recessive 1;Progressive external ophthalmoplegia, AD 1;MT recessive ataxia syndrome (includes SANDO and SCAE);MT DNA depletion syndrome 4B (MNGIE type);MT DNA depletion syndrome 4A (Alpers type)	AR, AD
POLR3B	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism;Charcot-Marie-Tooth disease, demyelinating, type 1I	AR, AD
POMC	176830	Obesity, adrenal insufficiency, and red hair due to POMC deficiency;Obesity, early-onset, susceptibility to	AR, AD, AR, MF
POU1F1	173110	Pituitary hormone deficiency, combined, 1	AD, AR
PPARG	601487	Carotid intimal medial thickness 1;Obesity, severe;Lipodystrophy, familial partial, type 3;Diabetes, type 2;Insulin resistance, severe, digenic	AD, AR, MF, AD
PPIB	123841	Osteogenesis imperfecta, type IX	AR
PRKAR1A	188830	Myxoma, intracardiac;Pigmented nodular adrenocortical disease, primary, 1;Carney complex, type 1;Acrodysostosis 1, with or without hormone resistance	AD
PRKCSH	177060	Polycystic liver disease 1	AD
PROK2	607002	Hypogonadotropic hypogonadism 4 with or without anosmia	AD
PROKR2	607123	Hypogonadotropic hypogonadism 3 with or without anosmia	AD
PROM1	604365	Cone-rod dystrophy 12;Macular dystrophy, retinal, 2;Retinitis pigmentosa 41;Stargardt disease 4	AD, AR, AD, AR
PROP1	601538	Pituitary hormone deficiency, combined, 2	AR

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
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
PTPRO	600579	Nephrotic syndrome, type 6	AR
RBBP8	604124	Seckel syndrome 2;Jawad syndrome	AR
RD3	180040	Leber congenital amaurosis 12	AR
RDH12	608830	Leber congenital amaurosis 13	AD, AR
RDH5	601617	Fundus albipunctatus	AD, AR
REN	179820	Hyperuricemic nephropathy, familial juvenile 2;Renal tubular dysgenesis	AD, AR
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
RLBP1	180090	Newfoundland rod-cone dystrophy;Fundus albipunctatus;Retinitis punctata albescens;Bothnia retinal dystrophy	AD, AR, AR
RMND1	614917	Combined oxidative phosphorylation deficiency 11	AR
RNF216	609948	Cerebellar ataxia and hypogonadotropic hypogonadism	AR
ROBO1	602430		
ROBO2	602431	Vesicoureteral reflux 2	AD
ROR2	602337	Brachydactyly, type B1;Robinow syndrome, AR	AD, AR
RPE65	180069	Leber congenital amaurosis 2;Retinitis pigmentosa 20;Retinitis pigmentosa 87 with choroidal involvement	AR, AD
RPGRIP1	605446	Cone-rod dystrophy 13;Leber congenital amaurosis 6	AR
RPGRIP1L	610937	Joubert syndrome 7;?COACH syndrome 3;Meckel syndrome 5	AR

RRM2B	604712	MT DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy);Progressive external ophthalmoplegia with MT DNA deletions, AD 5;MT DNA depletion syndrome 8B (MNGIE type)	AR, AD
RSPH1	609314	Ciliary dyskinesia, primary, 24	AR
RSPH4A	612647	Ciliary dyskinesia, primary, 11	
RSPH9	612648	Ciliary dyskinesia, primary, 12	
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
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome;Townes-Brocks syndrome 1	AD
SALL4	607343	IVIC syndrome;Duane-radial ray syndrome	AD
SBDS	607444	Aplastic anemia, susceptibility to;Shwachman-Diamond syndrome	AR
SCARB2	602257	Epilepsy, progressive myoclonic 4, with or without renal failure	AR
SCNN1A	600228	Pseudohypoaldosteronism, type I;?Liddle syndrome 3;Bronchiectasis with or without elevated sweat chloride 2	AR, AD
SCNN1B	600760	Bronchiectasis with or without elevated sweat chloride 1;Pseudohypoaldosteronism, type I;Liddle syndrome 1	AD, AR
SCNN1G	600761	Pseudohypoaldosteronism, type I;Liddle syndrome 2;Bronchiectasis with or without elevated sweat chloride 3	AR, AD
SDCCAG8	613524	Bardet-Biedl syndrome 16;Senior-Loken syndrome 7	AR
SEC61A1	609213	Hyperuricemic nephropathy, familial juvenile, 4	AD
SEC63	608648	Polycystic liver disease 2	AD
SEMA3A	603961	Hypogonadotropic hypogonadism 16 with or without anosmia	AD
SERPINA1	107400	Hemorrhagic diathesis due to antithrombin Pittsburgh;Emphysema-cirrhosis, due to AAT deficiency;Emphysema due to AAT deficiency	AR
SERPINF1	172860	Osteogenesis imperfecta, type VI	AR
SERPINH1	600943	Preterm premature rupture of the membranes, susceptibility to;Osteogenesis imperfecta, type X	AR
SGPL1	603729	Nephrotic syndrome, type 14	AR

SH3PXD2B	613293	Frank-ter Haar syndrome	AR
SIX1	601205	Deafness, AD 23;Branchiootic syndrome 3	AD
SIX2	604994		
SIX5	600963	Branchiootorenal syndrome 2	
SLC12A1	600839	Bartter syndrome, type 1	AR
SLC12A3	600968	Gitelman syndrome	AR
SLC25A13	603859	Citrullinemia, type II, neonatal-onset;Citrullinemia, adult-onset type II	AR
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	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
SLC26A3	126650	Diarrhea 1, secretory chloride, congenital	AR
SLC2A2	138160	Fanconi-Bickel syndrome;Diabetes mellitus, noninsulin-dependent	AR, AD
SLC34A1	182309	Nephrolithiasis/osteoporosis, hypophosphatemic, 1;?Fanconi renotubular syndrome 2;Hypercalcemia, infantile, 2	AD, AR
SLC34A3	609826	Hypophosphatemic rickets with hypercalciuria	AR
SLC35D1	610804	Schneckenbecken dysplasia	AR
SLC4A1	109270	[Blood group, Wright];Distal renal tubular acidosis 1;Ovalocytosis, SA type;[Malaria, resistance to];[Blood group, Swann];[Blood group, Froese];[Blood group, Waldner];Cryohydrocytosis;[Blood group, Diego];Distal renal tubular acidosis 4 with hemolytic anemia;Spherocytosis, type 4	AD, AR
SLC4A4	603345	Renal tubular acidosis, proximal, with ocular abnormalities	AR
SLCO1B1	604843	Hyperbilirubinemia, Rotor type, digenic	DR
SLCO1B3	605495	Hyperbilirubinemia, Rotor type, digenic	DR
SLIT2	603746		
SMARCAL1	606622	Schimke immunoosseous dysplasia	AR
SMPD1	607608	Niemann-Pick disease, type A;Niemann-Pick disease, type B	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
SOX17	610928	Vesicoureteral reflux 3	AD
SOX2	184429	Microphthalmia, syndromic 3;Optic nerve hypoplasia and abnormalities of the central nervous system	AD
SOX3	313430	Panhypopituitarism, X-linked;Mental retardation, X-linked, with isolated growth hormone deficiency	XL
SOX9	608160	Acampomelic campomelic dysplasia;Campomelic dysplasia;Campomelic dysplasia with autosomal sex reversal	AD
SPAG1	603395	Ciliary dyskinesia, primary, 28	AR
SPATA7	609868	Retinitis pigmentosa, juvenile, AR;Leber congenital amaurosis 3	
SPINT2	605124	Diarrhea 3, secretory sodium, congenital, syndromic	AR
SPRY4	607984	Hypogonadotropic hypogonadism 17 with or without anosmia	AD
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
TAC3	162330	Hypogonadotropic hypogonadism 10 with or without anosmia	AR
TACR3	162332	Hypogonadotropic hypogonadism 11 with or without anosmia	AR
TBC1D1	609850		
TBX15	604127	Cousin syndrome	AR
TBX18	604613	Congenital anomalies of kidney and urinary tract 2	AD
TBX3	601621	Ulnar-mammary syndrome	AD
TBX5	601620	Holt-Oram syndrome	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
TFR2	604720	Hemochromatosis, type 3	AR
TJP2	607709	Hypercholanemia, familial;Cholestasis, progressive familial intrahepatic 4	AR
TMEM107	616183	Meckel syndrome 13;?Joubert syndrome 29;Orofaciodigital syndrome XVI	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
TNFRSF11B	602643	Paget disease of bone 5, juvenile-onset	AR
TP53RK	608679	Galloway-Mowat syndrome 4	AR
TPRKB	608680	Galloway-Mowat syndrome 5	AR
TRAF3IP1	607380	Senior-Loken syndrome 9	AR
TRAP1	606219		
TRIM32	602290	Muscular dystrophy, limb-girdle, autosomal recessive 8;?Bardet-Biedl syndrome 11	AR
TRIP11	604505	Achondrogenesis, type IA;Osteochondrodysplasia	AR
TRMU	610230	Liver failure, transient infantile;Deafness, MT, modifier of	AR, MT
TRPC6	603652	Glomerulosclerosis, focal segmental, 2	AD
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
TTC21B	612014	Short-rib thoracic dysplasia 4 with or without polydactyly;Nephronophthisis 12	AR, AD, AR
TTC37	614589	Trichohepatoenteric syndrome 1	AR
TTC8	608132	Bardet-Biedl syndrome 8;?Retinitis pigmentosa 51	AR
TULP1	602280	Retinitis pigmentosa 14;Leber congenital amaurosis 15	AR
TXNDC15	617778		

UGT1A1	191740	[Gilbert syndrome];Crigler-Najjar syndrome, type II;Crigler-Najjar syndrome, type I;Hyperbilirubinemia, familial transient neonatal;[Bilirubin, serum level of, QTL1]	AR
UMOD	191845	Glomerulocystic kidney disease with hyperuricemia and isosthenuria;Hyperuricemic nephropathy, familial juvenile 1;Medullary cystic kidney disease 2	AD
UPK3A	611559		
VHL	608537	Pheochromocytoma;von Hippel-Lindau syndrome;Renal cell carcinoma, somatic;Erythrocytosis, familial, 2	AD, AR
VIPAS39	613401	Arthrogryposis, renal dysfunction, and cholestasis 2	AR
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1	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
WDR4	605924	Microcephaly, growth deficiency, seizures, and brain malformations;Galloway-Mowat syndrome 6	AR
WDR73	616144	Galloway-Mowat syndrome 1	AR
WNK1	605232	Neuropathy, hereditary sensory and autonomic, type II;Pseudohypoaldosteronism, type IIC	AR, AD
WNK4	601844	Pseudohypoaldosteronism, type IIB	AD
WNT4	603490	Mullerian aplasia and hyperandrogenism;?SERKAL syndrome	AD, AR
WNT5A	164975	Robinow syndrome, AD 1	AD
WNT7A	601570	Fuhrmann syndrome;Ulna and fibula, absence of, with severe limb deficiency	AR
WT1	607102	Denys-Drash syndrome;Mesothelioma, somatic;Frasier syndrome;Meacham syndrome;Wilms tumor, type 1;Nephrotic syndrome, type 4	AD, SM, AD
XPNPEP3	613553	Nephronophthisis-like nephropathy 1	AR
XYLT1	608124	Desbuquois dysplasia 2;Pseudoxanthoma elasticum, modifier of severity of	AR

ZIC3	300265	VACTERL association, X-linked;Congenital heart defects, nonsyndromic, 1, X-linked;Heterotaxy, visceral, 1, XL	XLR
ZMYND10	607070	Ciliary dyskinesia, primary, 22	AR
ZNF423	604557	Joubert syndrome 19;Nephronophthisis 14	AD, AR
PKD1	601313	Polycystic kidney disease 1	AD