

Panel Nefrología

Aproximadamente el 10% de la población mundial padecerá de alguna afección renal crónica. Los avances en las técnicas genéticas están proporcionando nuevos conocimientos sobre el diagnóstico, la patogénesis y la terapia de la enfermedad renal.

Panel Nephro es una herramienta integral para detectar los trastornos renales hereditarios más importantes como la poliquistosis renal, el síndrome de Alport, la acidosis tubular renal, la glomerulonefrosis focal y la hiperoxaluria primaria, entre otros.

El análisis de PKD1 no se incluye en este panel.

Genes:	495
Entrega:	25 days
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥150 x
Detalles:	Análisis incluido de CNV

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	60463 8	Glomerulosclerosis, focal segmental, 1	AD
ACVR2B	60273 0	Heterotaxy, visceral, 4, autosomal	
AGPS	60305 1	Rhizomelic chondrodysplasia punctata, type 3	AR
AGT	10615 0	Hypertension, essential, susceptibility to;Renal tubular dysgenesis	MF, AR
AGTR1	10616 5	Hypertension, essential;Renal tubular dysgenesis	MF, AR
AHI1	60889 4	Joubert syndrome 3	AR
AIPL1	60439 2	Cone-rod dystrophy;Leber congenital amaurosis 4;Retinitis pigmentosa, juvenile	AD, AR
AKR1D1	60474 1	Bile acid synthesis defect, congenital, 2	AR
ALDOB	61272 4	Fructose intolerance, hereditary	AR
ALG8	60810 3	Polycystic liver disease 3 with or without kidney cysts;Congenital disorder of glycosylation, type 1h	AD, AR
ALG9	60694 1	Congenital disorder of glycosylation, type II;Gillessen-Kaesbach-Nishimura syndrome	AR
ALPL	17176 0	Hypophosphatasia, infantile;Odontohypophosphatasia;Hypophosphatasia, childhood;Hypophosphatasia, adult	AR, AD, AR
AMER1	30064 7	Osteopathia striata with cranial sclerosis	XLD
ANKH	60514 5	Chondrocalcinosis 2;Craniometaphyseal dysplasia	AD
ANKS6	61537 0	Nephronophthisis 16	AR
ANLN	61602 7	Focal segmental glomerulosclerosis 8	AD
ANO5	60866 2	Gnathodiaphyseal dysplasia;Muscular dystrophy, limb-girdle, autosomal recessive 12;Miyoshi muscular dystrophy 3	AD, AR
ANOS1	30083 6	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)	XLR
AP2S1	60224 2	Hypocalciuric hypercalcemia, type III	AD
ARHGAP3 1	61091 1	Adams-Oliver syndrome 1	AD
ARHGDI A	60192 5	Nephrotic syndrome, type 8	AR
ARL13B	60892 2	Joubert syndrome 8	AR
ARL3	60469 5	Retinitis pigmentosa 83;Joubert syndrome 35	AD, AR

ARL6	60884 5	Retinitis pigmentosa 55;Bardet-Biedl syndrome 1, modifier of;Bardet-Biedl syndrome 3	AR, AR, DR
ARMC5	61554 9	ACTH-independent macronodular adrenal hyperplasia 2	AD, SM
ARMC9	61761 2	Joubert syndrome 30	AR
ARSL	30018 0	Chondrodysplasia punctata, XLR	XLR
ATP6V0A4	60523 9	Distal renal tubular acidosis 3, with or without sensorineural hearing loss	AR
ATP6V1B1	19213 2	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	AR
ATP8B1	60239 7	Cholestasis, progressive familial intrahepatic 1;Cholestasis, intrahepatic, of pregnancy, 1;Cholestasis, benign recurrent intrahepatic	AR, AD
ATR	60121 5	?Cutaneous telangiectasia and cancer syndrome, familial;Seckel syndrome 1	AD, AR
AVPR2	30053 8	Nephrogenic syndrome of inappropriate antidiuresis;Diabetes insipidus, nephrogenic	XLR
B9D1	61414 4	?Meckel syndrome 9;Joubert syndrome 27	AR
B9D2	61195 1	?Meckel syndrome 10;Joubert syndrome 34	AR
BAAT	60293 8	Hypercholanemia, familial;Bile acid conjugation defect 1	AR
BBS1	20990 1	Bardet-Biedl syndrome 1	AR, DR
BBS10	61014 8	Bardet-Biedl syndrome 10	AR
BBS12	61068 3	Bardet-Biedl syndrome 12	AR
BBS2	60615 1	Bardet-Biedl syndrome 2;Retinitis pigmentosa 74	AR
BBS4	60037 4	Bardet-Biedl syndrome 4	AR
BBS5	60365 0	Bardet-Biedl syndrome 5	AR
BBS7	60759 0	Bardet-Biedl syndrome 7	AR
BBS9	60796 8	Bardet-Biedl syndrome 9	AR
BCS1L	60364 7	GRACILE syndrome;Bjornstad syndrome;MT complex III deficiency, nuclear type 1	AR
BICC1	61429 5	Renal dysplasia, cystic, susceptibility to	AD
BMP1	11226 4	Osteogenesis imperfecta, type XIII	AR

BMP4	11226 2	Microphthalmia, syndromic 6;Orofacial cleft 11	AD
BMPR1B	60324 8	Brachydactyly, type A2;Acromesomelic dysplasia, Demirhan type;Brachydactyly, type A1, D	AD, AR
BNC2	60866 9	Lower urinary tract obstruction, congenital	AD
BSND	60641 2	Bartter syndrome, type 4a;Sensorineural deafness with mild renal dysfunction	AR
C2CD3	61594 4	Orofaciodigital syndrome XIV	AR
CA2	61149 2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	AR
CABP4	60896 5	Cone-rod synaptic disorder, congenital nonprogressive	AR
CANT1	61316 5	Desbuquois dysplasia 1;Epiphyseal dysplasia, multiple, 7	AR
CASP10	60176 2	Autoimmune lymphoproliferative syndrome, type II;Gastric cancer, somatic;Lymphoma, non-Hodgkin, somatic	AD
CASR	60119 9	Epilepsy idiopathic generalized, susceptibility to, 8;Hypocalcemia, AD, with Bartter syndrome;Hypocalciuric hypercalcemia, type I;Hyperparathyroidism, neonatal;Hypocalcemia, AD	AD, AD, AR
CC2D2A	61201 3	Meckel syndrome 6;Joubert syndrome 9;COACH syndrome 2	AR
CCDC103	61467 7	Ciliary dyskinesia, primary, 17	AR
CCDC28B	61016 2	Bardet-Biedl syndrome 1, modifier of	AR, DR
CCDC39	61379 8	Ciliary dyskinesia, primary, 14	AR
CCDC40	61379 9	Ciliary dyskinesia, primary, 15	AR
CCDC65	61108 8	Ciliary dyskinesia, primary, 27	AR
CCN6	60340 0	Progressive pseudorheumatoid dysplasia	AR
CCNO	60775 2	Ciliary dyskinesia, primary, 29	AR
CD2AP	60424 1	Glomerulosclerosis, focal segmental, 3	
CDKN1C	60085 6	IMAGE syndrome;Beckwith-Wiedemann syndrome	AD
CENPF	60023 6	Stromme syndrome	AR
CENPJ	60927 9	?Seckel syndrome 4;Microcephaly 6, primary, AR	AR

CEP120	61344 6	Joubert syndrome 31;Short-rib thoracic dysplasia 13 with or without polydactyly	AR
CEP152	61352 9	Microcephaly 9, primary, AR;Seckel syndrome 5	AR
CEP164	61484 8	Nephronophthisis 15	AR
CEP290	61014 2	Leber congenital amaurosis 10;Meckel syndrome 4;?Bardet-Biedl syndrome 14;Senior-Loken syndrome 6;Joubert syndrome 5	AR
CEP41	61052 3	Joubert syndrome 15	AR
CEP55	61000 0	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly	AR
CEP83	61584 7	Nephronophthisis 18	AR
CFAP298	61549 4	Ciliary dyskinesia, primary, 26	AR
CFAP418	61447 7	Retinitis pigmentosa 64;Cone-rod dystrophy 16;Bardet-Biedl syndrome 21	AR
CFAP53	61475 9	Heterotaxy, visceral, 6, AR	AR
CFTR	60242 1	Congenital bilateral absence of vas deferens;Pancreatitis, hereditary;Bronchiectasis with or without elevated sweat chloride 1, modifier of;Cystic fibrosis	AR, AD
CHD1L	61303 9		
CHD7	60889 2	CHARGE syndrome;Hypogonadotropic hypogonadism 5 with or without anosmia	AD
CHRNA3	11850 3	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT;Lung cancer susceptibility 2	AR
CHST3	60379 9	Spondyloepiphyseal dysplasia with congenital joint dislocations	AR
CHSY1	60818 3	Temtamy preaxial brachydactyly syndrome	AR
CILK1	61232 5	Endocrine-cerebroosteodysplasia;Epilepsy, juvenile myoclonic, susceptibility to, 10	AR, AD
CLCN5	30000 8	Nephrolithiasis, type I;Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis;Dent disease;Hypophosphatemic rickets	XLR
CLCNKA	60202 4	Bartter syndrome, type 4b, digenic	DR
CLCNKB	60202 3	Bartter syndrome, type 3;Bartter syndrome, type 4b, digenic	AR, DR

CLDN16	60395 9	Hypomagnesemia 3, renal	AR
CLDN19	61003 6	Hypomagnesemia 5, renal, with ocular involvement	AR
COL10A1	12011 0	Metaphyseal chondrodysplasia, Schmid type	AD
COL4A1	12013 0	?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	12007 0	Alport syndrome 3, AD;Hematuria, benign familial;Alport syndrome 2, AR	AD, AR
COL4A4	12013 1	Alport syndrome 2, AR;Hematuria, familial benign	AR, AD
COL4A5	30363 0	Alport syndrome 1, XL	XLD
COL9A3	12027 0	Epiphyseal dysplasia, multiple, 3, with or without myopathy;Intervertebral disc disease, susceptibility to	AD
COMP	60031 0	Carpal tunnel syndrome 2;Epiphyseal dysplasia, multiple, 1;Pseudoachondroplasia	AD
COQ2	60982 5	Coenzyme Q10 deficiency, primary, 1;Multiple system atrophy, susceptibility to	AR, AD, AR
COQ6	61464 7	Coenzyme Q10 deficiency, primary, 6	AR
COQ8B	61556 7	Nephrotic syndrome, type 9	AR
COQ9	61283 7	Coenzyme Q10 deficiency, primary, 5	AR
CPLANE1	61457 1	Orofaciodigital syndrome VI;Joubert syndrome 17	AR
CRB1	60421 0	Leber congenital amaurosis 8;Retinitis pigmentosa-12;Pigmented paravenous chorioretinal atrophy	AR, AD
CRB2	60972 0	Focal segmental glomerulosclerosis 9;Ventriculomegaly with cystic kidney disease	AR
CRELD1	60717 0	Atrioventricular septal defect, partial, with heterotaxy syndrome;Atrioventricular septal defect, susceptibility to, 2	AD
CRTAP	60549 7	Osteogenesis imperfecta, type VII	AR
CRX	60222 5	Leber congenital amaurosis 7;Cone-rod retinal dystrophy-2	AD
CSPP1	61165 4	Joubert syndrome 21	AR

CTNS	60627 2	Cystinosis, late-onset juvenile or adolescent nephropathic;Cystinosis, ocular nonnephropathic;Cystinosis, nephropathic;Cystinosis, atypical nephropathic	AR
CTU2	61705 7	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome	AR
CUBN	60299 7	[Proteinuria, chronic benign];Imerslund-Grasbeck syndrome 1	AR
CUL3	60313 6	Pseudohypoaldosteronism, type IIE;Neurodevelopmental disorder with or without autism or seizures	AD
CWC27	61717 0	Retinitis pigmentosa with or without skeletal anomalies	AR
CYP7B1	60371 1	Spastic paraplegia 5A, AR;Bile acid synthesis defect, congenital, 3	AR
DCDC2	60575 5	Nephronophthisis 19;?Deafness, autosomal recessive 66;Sclerosing cholangitis, neonatal	AR
DDR2	19131 1	Spondylometaphyseal dysplasia, short limb-hand type;Warburg-Cinotti syndrome	AR, AD
DDX59	61546 4	Orofaciodigital syndrome V	AR
DGKE	60144 0	Nephrotic syndrome, type 7;Hemolytic uremic syndrome, atypical, susceptibility to, 7	AR
DGUOK	60146 5	Portal hypertension, noncirrhotic;Progressive external ophthalmoplegia with MT DNA deletions, autosomal recessive 4;MT DNA depletion syndrome 3 (hepatocerebral type)	AR
DHCR7	60285 8	Smith-Lemli-Opitz syndrome	AR
DICER1	60624 1	Pleuropulmonary blastoma;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors;GLOW syndrome, somatic mosaic;Rhabdomyosarcoma, embryonal, 2	AD
DLL3	60276 8	Spondylocostal dysostosis 1, AR	AR
DMP1	60098 0	Hypophosphatemic rickets, AR	AR
DNAAF1	61319 0	Ciliary dyskinesia, primary, 13	AR
DNAAF2	61251 7	Ciliary dyskinesia, primary, 10	AR
DNAAF3	61456 6	Ciliary dyskinesia, primary, 2	AR
DNAAF4	60870 6	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
DNAAF11	614930	Ciliary dyskinesia, primary, 19	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
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	60935 3	Roberts syndrome;Juberg-Hayward syndrome;SC phocomelia syndrome	AR
EVC	60483 1	Ellis-van Creveld syndrome;?Weyers acrofacial dysostosis	AR, AD
EVC2	60726 1	Ellis-van Creveld syndrome;Weyers acrofacial dysostosis	AR, AD
EXT1	60817 7	Chondrosarcoma;Exostoses, multiple, type 1	SM, AD
EXT2	60821 0	Exostoses, multiple, type 2;Seizures, scoliosis, and macrocephaly syndrome	AD, AR
EYA1	60165 3	Anterior segment anomalies with or without cataract;Branchiootorenal syndrome 1, with or without cataracts;?Otofaciocervical syndrome;Branchiootic syndrome 1	AD
FAH	61387 1	Tyrosinemia, type I	AR
FAM20C	61106 1	Raine syndrome	AR
FAN1	61353 4	Interstitial nephritis, karyomegalic	AR
FAS	13463 7	Autoimmune lymphoproliferative syndrome, type IA;Autoimmune lymphoproliferative syndrome	AD
FASLG	13463 8	Autoimmune lymphoproliferative syndrome, type IB;Lung cancer, susceptibility to	AD, AD, SM
FAT4	61241 1	Hennekam lymphangiectasia-lymphedema syndrome 2;Van Maldergem syndrome 2	AR
FEZF1	61330 1	Hypogonadotropic hypogonadism 22, with or without anosmia	AR
FGF17	60372 5	Hypogonadotropic hypogonadism 20 with or without anosmia	AD
FGF23	60538 0	Tumoral calcinosis, hyperphosphatemic, familial, 2;Hypophosphatemic rickets, AD	AR, AD
FGF8	60048 3	Hypogonadotropic hypogonadism 6 with or without anosmia	AD
FGFR1	13635 0	Osteoglophonic dysplasia;Trigonocephaly 1;Pfeiffer syndrome;Encephalocraniocutaneous lipomatosis, somatic mosaic;Hypogonadotropic hypogonadism 2 with or without anosmia;Jackson-Weiss syndrome;Hartsfield syndrome	AD

FGFR2	17694 3	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	60706 3	Osteogenesis imperfecta, type XI;Bruck syndrome 1	AR
FLNB	60338 1	Larsen syndrome;Boomerang dysplasia;Spondylocarpotarsal synostosis syndrome;Atelosteogenesis, type III;Atelosteogenesis, type I	AD, AR
FLRT3	60480 8	Hypogonadotropic hypogonadism 21 with anosmia	AD
FN1	13560 0	Glomerulopathy with fibronectin deposits 2;Spondylometaphyseal dysplasia, corner fracture type	AD
FOXP1	60551 5	Mental retardation with language impairment and with or without autistic features	AD
FRAS1	60783 0	Fraser syndrome 1	AR
FREM1	60894 4	Manitoba oculotrichoanal syndrome;Trigonocephaly 2;Bifid nose with or without anorectal and renal anomalies	AR, AD
FREM2	60894 5	Fraser syndrome 2;Cryptophthalmos, unilateral or bilateral, isolated	AR
FSHB	13653 0	Hypogonadotropic hypogonadism 24 without anosmia	AR
FXVD2	60181 4	Hypomagnesemia 2, renal	AD
GANAB	10416 0	Polycystic kidney disease 3	AD
GATA3	13132 0	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	AD
GDF1	60288 0	Congenital heart defects, multiple types, 6;Right atrial isomerism (Ivemark)	AD, AR
GDF5	60114 6	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	60663 9	Combined oxidative phosphorylation deficiency 1	AR
GHR	60094 6	Hypercholesterolemia, familial, modifier of;Laron dwarfism;Growth hormone insensitivity, partial;Increased responsiveness to growth hormone	AD, AR, AR, AD
GLA	30064 4	Fabry disease;Fabry disease, cardiac variant	XL
GLI2	16523 0	Holoprosencephaly 9;Culler-Jones syndrome	AD
GLI3	16524 0	Pallister-Hall syndrome;Polydactyly, preaxial, type IV;Polydactyly, postaxial, types A1 and B;Greig cephalopolysyndactyly syndrome	AD
GLIS2	60853 9	Nephronophthisis 7	
GLIS3	61019 2	Diabetes mellitus, neonatal, with congenital hypothyroidism	AR
GNA11	13931 3	Hypocalciuric hypercalcemia, type II;Hypocalcemia, AD 2	AD
GNAS	13932 0	Pseudohypoparathyroidism 1c;Osseous heteroplasia, progressive;Pseudopseudohypoparathyroidism;Pseudohypoparathyroidism 1a;Pseudohypoparathyroidism 1b;Pituitary adenoma 3, multiple types, somatic;ACTH-independent macronodular adrenal hyperplasia;McCune-Albright syndrome, somatic, mosaic	AD, SM
GNPAT	60274 4	Rhizomelic chondrodysplasia punctata, type 2	AR
GNRH1	15276 0	?Hypogonadotropic hypogonadism 12 with or without anosmia	AR
GNRHR	13885 0	Hypogonadotropic hypogonadism 7 without anosmia	AR
GPC3	30003 7	Simpson-Golabi-Behmel syndrome, type 1;Wilms tumor, somatic	XLR
GPC6	60440 4	Omodysplasia 1	AR
GREB1L	61778 2	Deafness, AD 80;Renal hypodysplasia/aplasia 3	AD
GRIP1	60459 7	Fraser syndrome 3	AR
GUCY2D	60017 9	Cone-rod dystrophy 6;Leber congenital amaurosis 1;Night blindness, congenital stationary, type 11;?Choroidal dystrophy, central areolar 1	AD, AR, AR, AD
HAAO	60452 1	Vertebral, cardiac, renal, and limb defects syndrome 1	AR
HAMP	60646 4	Hemochromatosis, type 2B	AR

HESX1	60180 2	Septo-optic dysplasia; Pituitary hormone deficiency, combined, 5; Growth hormone deficiency with pituitary anomalies	AD, AR
HEXA	60686 9	Tay-Sachs disease; [Hex A pseudodeficiency]; GM2-gangliosidosis, several forms	AR
HFE	61360 9	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	18990 7	Renal cysts and diabetes syndrome; Diabetes mellitus, noninsulin-dependent; Renal cell carcinoma	AD
HNF4A	60028 1	Fanconi renal tubular syndrome 4, with maturity-onset diabetes of the young; Diabetes mellitus, noninsulin-dependent; MODY, type I	AD
HOXA13	14295 9	?Guttmacher syndrome; Hand-foot-uterus syndrome	AD
HOXD13	14298 9	Syndactyly, type V; ?Brachydactyly-syndactyly syndrome; Brachydactyly, type D; Synpolydactyly 1; Brachydactyly, type E	AD
HPSE2	61346 9	Urofacial syndrome 1	AR
HS6ST1	60484 6	Hypogonadotropic hypogonadism 15 with or without anosmia	AD
HSD11B2	61423 2	Apparent mineralocorticoid excess	AR
HSD3B7	60776 4	Bile acid synthesis defect, congenital, 1	AR
HSPG2	14246 1	Schwartz-Jampel syndrome, type 1; Dyssegmental dysplasia, Silverman-Handmaker type	AR
HYDIN	61081 2	Ciliary dyskinesia, primary, 5	AR
HYLS1	61069 3	Hydroletharus syndrome	AR
IFITM5	61475 7	Osteogenesis imperfecta, type V	AD
IFT122	60604 5	Cranioectodermal dysplasia 1	AR
IFT140	61462 0	Retinitis pigmentosa 80; Short-rib thoracic dysplasia 9 with or without polydactyly	AR
IFT172	60738 6	Retinitis pigmentosa 71; Bardet-Biedl syndrome 20; Short-rib thoracic dysplasia 10 with or without polydactyly	AR
IFT27	61587 0	?Bardet-Biedl syndrome 19	AR

IFT43	61406 8	?Cranioectodermal dysplasia 3;Short-rib thoracic dysplasia 18 with polydactyly;?Retinitis pigmentosa 81	AR
IFT80	61117 7	Short-rib thoracic dysplasia 2 with or without polydactyly	AR
IFT81	60548 9	Short-rib thoracic dysplasia 19 with or without polydactyly	AR
IHH	60072 6	Brachydactyly, type A1;Acrocapitofemoral dysplasia	AD, AR
IL17RD	60680 7	Hypogonadotropic hypogonadism 18 with or without anosmia	AD, AR, DD
IMPDH1	14669 0	Leber congenital amaurosis 11;Retinitis pigmentosa 10	AD
INF2	61098 2	Glomerulosclerosis, focal segmental, 5;Charcot-Marie-Tooth disease, dominant intermediate E	AD
INPP5E	61303 7	Mental retardation, truncal obesity, retinal dystrophy, and micropenis;Joubert syndrome 1	AR
INPPL1	60082 9	Opsismodysplasia	AR
INVS	24330 5	Nephronophthisis 2, infantile	AR
IQCB1	60923 7	Senior-Loken syndrome 5	AR
ITGA3	60502 5	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	AR
ITGA8	60406 3	Renal hypodysplasia/aplasia 1	AR
JAG1	60192 0	Alagille syndrome 1;Charcot-Marie-Tooth disease, axonal, type 2HH;?Deafness, congenital heart defects, and posterior embryotoxon;Tetralogy of Fallot	AD
KANK2	61461 0	Nephrotic syndrome, type 16;Palmoplantar keratoderma and woolly hair	AR
KCNJ1	60035 9	Bartter syndrome, type 2	AR
KCNJ10	60220 8	Enlarged vestibular aqueduct, digenic;SESAME syndrome	AR
KCNJ13	60320 8	Snowflake vitreoretinal degeneration;Leber congenital amaurosis 16	AD, AR
KCNJ5	60073 4	Hyperaldosteronism, familial, type III;Long QT syndrome 13	AD
KDM6A	30012 8	Kabuki syndrome 2	XLD
KIAA0586	61017 8	Joubert syndrome 23;Short-rib thoracic dysplasia 14 with polydactyly	AR

KIF14	61127 9	?Meckel syndrome 12;Microcephaly 20, primary, AR	AR
KIF22	60321 3	Spondyloepimetaphyseal dysplasia with joint laxity, type 2	AD
KIF7	61125 4	Joubert syndrome 12;?Hydroletharus syndrome 2;?Al-Gazali-Bakalinova syndrome;Acrocallosal syndrome	AR
KISS1	60328 6	?Hypogonadotropic hypogonadism 13 with or without anosmia	AR
KISS1R	60416 1	?Precocious puberty, central, 1;Hypogonadotropic hypogonadism 8 with or without anosmia	AD, AR
KLHL3	60577 5	Pseudohypoaldosteronism, type IID	AD, AR
KMT2D	60211 3	Kabuki syndrome 1	AD
KYNU	60519 7	?Hydroxykynureninuria;Vertebral, cardiac, renal, and limb defects syndrome 2	AR
LAGE3	30006 0	Galloway-Mowat syndrome 2, XL	XLR
LAMB2	15032 5	Nephrotic syndrome, type 5, with or without ocular abnormalities;Pierson syndrome	AR
LBR	60002 4	?Reynolds syndrome;Greenberg skeletal dysplasia;Pelger-Huet anomaly;Pelger-Huet anomaly with mild skeletal anomalies	AD, AR
LCA5	61140 8	Leber congenital amaurosis 5	AR
LCAT	60696 7	Norum disease;Fish-eye disease	AR
LCT	60320 2	Lactase deficiency, congenital	AR
LEP	16416 0	Obesity, morbid, due to leptin deficiency	AR
LEPR	60100 7	Obesity, morbid, due to leptin receptor deficiency	AR
LHB	15278 0	Hypogonadotropic hypogonadism 23 with or without anosmia	AR
LHX3	60057 7	Pituitary hormone deficiency, combined, 3	AR
LHX4	60214 6	Pituitary hormone deficiency, combined, 4	AD
LIFR	15144 3	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	AR
LMF1	61176 1	Lipase deficiency, combined	AR
LMX1B	60257 5	Nail-patella syndrome;Focal segmental glomerulosclerosis 10	AD
LRAT	60486 3	Leber congenital amaurosis 14;Retinal dystrophy, early-onset severe;Retinitis pigmentosa, juvenile	AR

LRIG2	60886 9	Urofacial syndrome 2	AR
LRP4	60427 0	?Myasthenic syndrome, congenital, 17;Cenani-Lenz syndactyly syndrome;Sclerosteosis 2	AR, AD, AR
LRP5	60350 6	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	60656 8	Bardet-Biedl syndrome 17	AR
MAFB	60896 8	Duane retraction syndrome 3;Multicentric carpotarsal osteolysis syndrome	AD
MAGI2	60638 2	Nephrotic syndrome, type 15	AR
MAPKBP1	61678 6	Nephronophthisis 20	AR
MATN3	60210 9	?Spondyloepimetaphyseal dysplasia, Borochowitz Cormier-Daire type;Epiphyseal dysplasia, multiple, 5;Osteoarthritis susceptibility 2	AR, AD
MCEE	60841 9	Methylmalonyl-CoA epimerase deficiency	AR
MERTK	60470 5	Retinitis pigmentosa 38	AR
MESP2	60519 5	Spondylocostal dysostosis 2, AR	AR
MGP	15487 0	Keutel syndrome	AR
MKKS	60489 6	McKusick-Kaufman syndrome;Bardet-Biedl syndrome 6	AR
MKS1	60988 3	Bardet-Biedl syndrome 13;Joubert syndrome 28;Meckel syndrome 1	AR
MMAA	60748 1	Methylmalonic aciduria, vitamin B12-responsive	AR
MMAB	60756 8	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	AR
MMADHC	61193 5	Methylmalonic aciduria and homocystinuria, cblD type;Methylmalonic aciduria, cblD type, variant 2;Homocystinuria, cblD type, variant 1	AR
MMP13	60010 8	Metaphyseal anadysplasia 1;Metaphyseal dysplasia, Spahr type;?Spondyloepimetaphyseal dysplasia, Missouri type	AD, AR
MMP21	60841 6	Heterotaxy, visceral, 7, autosomal	AR

MMP9	12036 1	Metaphyseal anadysplasia 2	
MMUT	60905 8	Methylmalonic aciduria, mut(0) type	AR
MPV17	13796 0	Charcot-Marie-Tooth disease, axonal, type 2EE;MT DNA depletion syndrome 6 (hepatocerebral type)	AR
MUC1	15834 0	Medullary cystic kidney disease 1	AD
MYH9	16077 5	Deafness, AD 17;Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	AD
MYO1E	60147 9	Glomerulosclerosis, focal segmental, 6	AR
MYO5B	60654 0	Microvillus inclusion disease	AR
MYO7A	27690 3	Deafness, AD 11;Usher syndrome, type 1B;Deafness, autosomal recessive 2	AD, AR
MYOCD	60612 7	Megabladder, congenital	AD
NADSYN1	60828 5	Vertebral, cardiac, renal, and limb defects syndrome 3	AR
NBAS	60802 5	Short stature, optic nerve atrophy, and Pelger-Huet anomaly;Infantile liver failure syndrome 2	AR
NEK1	60458 8	Short-rib thoracic dysplasia 6 with or without polydactyly;Amyotrophic lateral sclerosis, susceptibility to, 24	AR, DR, AD
NEK8	60979 9	Renal-hepatic-pancreatic dysplasia 2;?Nephronophthisis 9	AR
NEUROG3	60488 2	Diarrhea 4, malabsorptive, congenital	AR
NIPBL	60866 7	Cornelia de Lange syndrome 1	AD
NKX2-5	60058 4	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	60218 3	Spondylo-megaepiphyseal-metaphyseal dysplasia	AR
NME8	60742 1	Ciliary dyskinesia, primary, 6	AR
NMNAT1	60870 0	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis;Leber congenital amaurosis 9	AR
NODAL	60126 5	Heterotaxy, visceral, 5	AD

NOG	60299 1	Stapes ankylosis with broad thumbs and toes;Brachydactyly, type B2;Tarsal-carpal coalition syndrome;Multiple synostoses syndrome 1;Symphalangism, proximal, 1A	AD
NOTCH2	60027 5	Alagille syndrome 2;Hajdu-Cheney syndrome	AD
NPC1	60762 3	Niemann-Pick disease, type C1;Niemann-Pick disease, type D	AR
NPC2	60101 5	Niemann-pick disease, type C2	AR
NPHP1	60710 0	Joubert syndrome 4;Nephronophthisis 1, juvenile;Senior-Loken syndrome-1	AR
NPHP3	60800 2	Nephronophthisis 3;Meckel syndrome 7;Renal-hepatic-pancreatic dysplasia 1	AR
NPHP4	60721 5	Senior-Loken syndrome 4;Nephronophthisis 4	AR
NPHS1	60271 6	Nephrotic syndrome, type 1	AR
NPHS2	60476 6	Nephrotic syndrome, type 2	AR
NPR2	10896 1	Acromesomelic dysplasia, Maroteaux type;Short stature with nonspecific skeletal abnormalities;Epiphyseal chondrodysplasia, Miura type	AR, AD
NR0B1	30047 3	Adrenal hypoplasia, congenital;46XY sex reversal 2, dosage-sensitive	XLR, XL
NR0B2	60463 0	Obesity, mild, early-onset	AD, AR, MF
NR1H4	60382 6	Cholestasis, progressive familial intrahepatic, 5	AR
NR3C2	60098 3	Hypertension, early-onset, AD, with exacerbation in pregnancy;Pseudohypoaldosteronism type I, AD	AD
NSDHL	30027 5	CK syndrome;CHILD syndrome	XLR, XLD
NSMF	60813 7	Hypogonadotropic hypogonadism 9 with or without anosmia	AD
NUP107	60761 7	Galloway-Mowat syndrome 7;Nephrotic syndrome, type 11;?Ovarian dysgenesis 6	AR
NUP93	61435 1	Nephrotic syndrome, type 12	AR
OBSL1	61099 1	3-M syndrome 2	AR
OCRL	30053 5	Lowe syndrome;Dent disease 2	XLR
OFD1	30017 0	Joubert syndrome 10;Simpson-Golabi-Behmel syndrome, type 2;?Retinitis pigmentosa 23;Orofaciodigital syndrome I	XLR, XLD

OSGEP	61010 7	Galloway-Mowat syndrome 3	AR
OTX2	60003 7	Retinal dystrophy, early-onset, with or without pituitary dysfunction;Microphthalmia, syndromic 5;Pituitary hormone deficiency, combined, 6	AD
ODAD1	61503 8	Ciliary dyskinesia, primary, 20	AR
ODAD2	61540 8	Ciliary dyskinesia, primary, 23	AR
ODAD3	61595 6	Ciliary dyskinesia, primary, 30	AR
P3H1	61033 9	Osteogenesis imperfecta, type VIII	AR
PAPSS2	60300 5	Brachyolmia 4 with mild epiphyseal and metaphyseal changes	AR
PAX2	16740 9	Glomerulosclerosis, focal segmental, 7;Papillorenal syndrome	AD
PBX1	17631 0	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay	AD
PCSK1	16215 0	Obesity with impaired prohormone processing;Obesity, susceptibility to, BMIQ12	AR
PDE4D	60012 9	Acrodysostosis 2, with or without hormone resistance	AD
PDE6D	60267 6	Joubert syndrome 22	AR
PDSS2	61056 4	Coenzyme Q10 deficiency, primary, 3	AR
PEX1	60213 6	Peroxisome biogenesis disorder 1A (Zellweger);Peroxisome biogenesis disorder 1B (NALD/IRD);Heimler syndrome 1	AR
PEX10	60285 9	Peroxisome biogenesis disorder 6A (Zellweger);Peroxisome biogenesis disorder 6B	AR
PEX12	60175 8	Peroxisome biogenesis disorder 3B;Peroxisome biogenesis disorder 3A (Zellweger)	AR
PEX2	17099 3	Peroxisome biogenesis disorder 5B;Peroxisome biogenesis disorder 5A (Zellweger)	AR
PEX26	60866 6	Peroxisome biogenesis disorder 7A (Zellweger);Peroxisome biogenesis disorder 7B	AR
PEX5	60041 4	Rhizomelic chondrodysplasia punctata, type 5;Peroxisome biogenesis disorder 2B;Peroxisome biogenesis disorder 2A (Zellweger)	AR
PEX6	60149 8	Peroxisome biogenesis disorder 4B;Peroxisome biogenesis disorder 4A (Zellweger);Heimler syndrome 2	AD, AR, AR

PEX7	60175 7	Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata, type 1	AR
PHEX	30055 0	Hypophosphatemic rickets, XLD	XLD
PHF6	30041 4	Borjeson-Forssman-Lehmann syndrome	XLR
PIBF1	60753 2	Joubert syndrome 33	AR
PKD1L1	60972 1	Heterotaxy, visceral, 8, autosomal	AR
PKD2	17391 0	Polycystic kidney disease 2	AD
PKHD1	60670 2	Polycystic kidney disease 4, with or without hepatic disease	AR
PLCE1	60841 4	Nephrotic syndrome, type 3	AR
PLOD2	60186 5	Bruck syndrome 2	AR
PMM2	60178 5	Congenital disorder of glycosylation, type Ia	AR
PNPLA6	60319 7	Spastic paraplegia 39, AR;Boucher-Neuhauser syndrome;Oliver-McFarlane syndrome;?Laurence-Moon syndrome	AR
POLG	17476 3	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	61436 6	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism;Charcot-Marie-Tooth disease, demyelinating, type 1I	AR, AD
POMC	17683 0	Obesity, adrenal insufficiency, and red hair due to POMC deficiency;Obesity, early-onset, susceptibility to	AR, AD, AR, MF
POU1F1	17311 0	Pituitary hormone deficiency, combined, 1	AD, AR
PPARG	60148 7	Carotid intimal medial thickness 1;Obesity, severe;Lipodystrophy, familial partial, type 3;Diabetes, type 2;Insulin resistance, severe, digenic	AD, AR, MF, AD
PPIB	12384 1	Osteogenesis imperfecta, type IX	AR
PRKAR1A	18883 0	Myxoma, intracardiac;Pigmented nodular adrenocortical disease, primary, 1;Carney complex, type 1;Acrodysostosis 1, with or without hormone resistance	AD

PRKCSH	17706 0	Polycystic liver disease 1	AD
PROK2	60700 2	Hypogonadotropic hypogonadism 4 with or without anosmia	AD
PROKR2	60712 3	Hypogonadotropic hypogonadism 3 with or without anosmia	AD
PROM1	60436 5	Cone-rod dystrophy 12;Macular dystrophy, retinal, 2;Retinitis pigmentosa 41;Stargardt disease 4	AD, AR, AD, AR
PROP1	60153 8	Pituitary hormone deficiency, combined, 2	AR
PRPH2	17960 5	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	16846 8	Chondrodysplasia, Blomstrand type;Failure of tooth eruption, primary;Eiken syndrome;Metaphyseal chondrodysplasia, Murk Jansen type	AR, AD
PTHLH	16847 0	Brachydactyly, type E2	AD
PTPRO	60057 9	Nephrotic syndrome, type 6	AR
RBBP8	60412 4	Seckel syndrome 2;Jawad syndrome	AR
RD3	18004 0	Leber congenital amaurosis 12	AR
RDH12	60883 0	Leber congenital amaurosis 13	AD, AR
RDH5	60161 7	Fundus albipunctatus	AD, AR
REN	17982 0	Hyperuricemic nephropathy, familial juvenile 2;Renal tubular dysgenesis	AD, AR
RET	16476 1	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	18038 0	Retinitis punctata albescens;Retinitis pigmentosa 4, AD or recessive;Night blindness, congenital stationary, AD 1	AD, AR
RLBP1	18009 0	Newfoundland rod-cone dystrophy;Fundus albipunctatus;Retinitis punctata albescens;Bothnia retinal dystrophy	AD, AR, AR
RMND1	61491 7	Combined oxidative phosphorylation deficiency 11	AR

RNF216	60994 8	Cerebellar ataxia and hypogonadotropic hypogonadism	AR
ROBO1	60243 0		
ROBO2	60243 1	Vesicoureteral reflux 2	AD
ROR2	60233 7	Brachydactyly, type B1;Robinow syndrome, AR	AD, AR
RPE65	18006 9	Leber congenital amaurosis 2;Retinitis pigmentosa 20;Retinitis pigmentosa 87 with choroidal involvement	AR, AD
RPGRIP1	60544 6	Cone-rod dystrophy 13;Leber congenital amaurosis 6	AR
RPGRIP1L	61093 7	Joubert syndrome 7;?COACH syndrome 3;Meckel syndrome 5	AR
RRM2B	60471 2	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	60931 4	Ciliary dyskinesia, primary, 24	AR
RSPH4A	61264 7	Ciliary dyskinesia, primary, 11	
RSPH9	61264 8	Ciliary dyskinesia, primary, 12	
RUNX2	60021 1	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	60221 8	Townes-Brocks branchiootorenal-like syndrome;Townes-Brocks syndrome 1	AD
SALL4	60734 3	IVIC syndrome;Duane-radial ray syndrome	AD
SBDS	60744 4	Aplastic anemia, susceptibility to;Shwachman-Diamond syndrome	AR
SCARB2	60225 7	Epilepsy, progressive myoclonic 4, with or without renal failure	AR
SCNN1A	60022 8	Pseudohypoaldosteronism, type I;?Liddle syndrome 3;Bronchiectasis with or without elevated sweat chloride 2	AR, AD
SCNN1B	60076 0	Bronchiectasis with or without elevated sweat chloride 1;Pseudohypoaldosteronism, type I;Liddle syndrome 1	AD, AR

SCNN1G	60076 1	Pseudohypoaldosteronism, type I;Liddle syndrome 2;Bronchiectasis with or without elevated sweat chloride 3	AR, AD
SDCCAG8	61352 4	Bardet-Biedl syndrome 16;Senior-Loken syndrome 7	AR
SEC61A1	60921 3	Hyperuricemic nephropathy, familial juvenile, 4	AD
SEC63	60864 8	Polycystic liver disease 2	AD
SEMA3A	60396 1	Hypogonadotropic hypogonadism 16 with or without anosmia	AD
SERPINA1	10740 0	Hemorrhagic diathesis due to antithrombin Pittsburgh;Emphysemacirrhosis, due to AAT deficiency;Emphysema due to AAT deficiency	AR
SERPINF1	17286 0	Osteogenesis imperfecta, type VI	AR
SERPINH1	60094 3	Preterm premature rupture of the membranes, susceptibility to;Osteogenesis imperfecta, type X	AR
SGPL1	60372 9	Nephrotic syndrome, type 14	AR
SH3PXD2 B	61329 3	Frank-ter Haar syndrome	AR
SIX1	60120 5	Deafness, AD 23;Branchiootic syndrome 3	AD
SIX2	60499 4		
SIX5	60096 3	Branchiootorenal syndrome 2	
SLC12A1	60083 9	Bartter syndrome, type 1	AR
SLC12A3	60096 8	Gitelman syndrome	AR
SLC25A13	60385 9	Citrullinemia, type II, neonatal-onset;Citrullinemia, adult-onset type II	AR
SLC25A15	60386 1	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR
SLC26A2	60671 8	Diastrophic dysplasia, broad bone-platyspondylic variant;Achondrogenesis Ib;Epiphyseal dysplasia, multiple, 4;De la Chapelle dysplasia;Diastrophic dysplasia;Atelosteogenesis, type II	AR
SLC26A3	12665 0	Diarrhea 1, secretory chloride, congenital	AR
SLC2A2	13816 0	Fanconi-Bickel syndrome;Diabetes mellitus, noninsulin-dependent	AR, AD
SLC34A1	18230 9	Nephrolithiasis/osteoporosis, hypophosphatemic, 1;?Fanconi renotubular syndrome 2;Hypercalcemia, infantile, 2	AD, AR

SLC34A3	60982 6	Hypophosphatemic rickets with hypercalciuria	AR
SLC35D1	61080 4	Schneckenbecken dysplasia	AR
SLC4A1	10927 0	[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	60334 5	Renal tubular acidosis, proximal, with ocular abnormalities	AR
SLCO1B1	60484 3	Hyperbilirubinemia, Rotor type, digenic	DR
SLCO1B3	60549 5	Hyperbilirubinemia, Rotor type, digenic	DR
SLIT2	60374 6		
SMARCAL1	60662 2	Schimke immunoosseous dysplasia	AR
SMPD1	60760 8	Niemann-Pick disease, type A;Niemann-Pick disease, type B	AR
SOX10	60222 9	Waardenburg syndrome, type 4C;PCWH syndrome;Waardenburg syndrome, type 2E, with or without neurologic involvement	AD
SOX11	60089 8	Coffin-Siris syndrome 9	AD
SOX17	61092 8	Vesicoureteral reflux 3	AD
SOX2	18442 9	Microphthalmia, syndromic 3;Optic nerve hypoplasia and abnormalities of the central nervous system	AD
SOX3	31343 0	Panhypopituitarism, X-linked;Mental retardation, X-linked, with isolated growth hormone deficiency	XL
SOX9	60816 0	Acampomelic campomelic dysplasia;Campomelic dysplasia;Campomelic dysplasia with autosomal sex reversal	AD
SPAG1	60339 5	Ciliary dyskinesia, primary, 28	AR
SPATA7	60986 8	Retinitis pigmentosa, juvenile, AR;Leber congenital amaurosis 3	
SPINT2	60512 4	Diarrhea 3, secretory sodium, congenital, syndromic	AR
SPRY4	60798 4	Hypogonadotropic hypogonadism 17 with or without anosmia	AD
STRA6	61074 5	Microphthalmia, isolated, with coloboma 8;Microphthalmia, syndromic 9	AR

SUFU	60703 5	Meningioma, familial, susceptibility to;Basal cell nevus syndrome;Medulloblastoma, desmoplastic;Joubert syndrome 32	AD, AD, AR, SM, AR
TAC3	16233 0	Hypogonadotropic hypogonadism 10 with or without anosmia	AR
TACR3	16233 2	Hypogonadotropic hypogonadism 11 with or without anosmia	AR
TBC1D1	60985 0		
TBX15	60412 7	Cousin syndrome	AR
TBX18	60461 3	Congenital anomalies of kidney and urinary tract 2	AD
TBX3	60162 1	Ulnar-mammary syndrome	AD
TBX5	60162 0	Holt-Oram syndrome	AD
TCTN1	60986 3	Joubert syndrome 13	AR
TCTN2	61384 6	?Meckel syndrome 8;Joubert syndrome 24	AR
TCTN3	61384 7	Joubert syndrome 18;Orofaciodigital syndrome IV	AR
TFR2	60472 0	Hemochromatosis, type 3	AR
TJP2	60770 9	Hypercholanemia, familial;Cholestasis, progressive familial intrahepatic 4	AR
TMEM107	61618 3	Meckel syndrome 13;?Joubert syndrome 29;Orofaciodigital syndrome XVI	AR
TMEM138	61445 9	Joubert syndrome 16	AR
TMEM216	61327 7	Meckel syndrome 2;Joubert syndrome 2	AR
TMEM231	61494 9	Joubert syndrome 20;Meckel syndrome 11	AR
TMEM237	61442 3	Joubert syndrome 14	AR
TMEM67	60988 4	COACH syndrome 1;?RHYNS syndrome;Meckel syndrome 3;Joubert syndrome 6;Bardet-Biedl syndrome 14, modifier of;Nephronophthisis 11	AR
TNFRSF11B	60264 3	Paget disease of bone 5, juvenile-onset	AR
TP53RK	60867 9	Galloway-Mowat syndrome 4	AR
TPRKB	60868 0	Galloway-Mowat syndrome 5	AR
TRAF3IP1	60738 0	Senior-Loken syndrome 9	AR

TRAP1	60621 9		
TRIM32	60229 0	Muscular dystrophy, limb-girdle, autosomal recessive 8;?Bardet-Biedl syndrome 11	AR
TRIP11	60450 5	Achondrogenesis, type IA;Osteochondrodysplasia	AR
TRMU	61023 0	Liver failure, transient infantile;Deafness, MT, modifier of	AR, MT
TRPC6	60365 2	Glomerulosclerosis, focal segmental, 2	AD
TRPS1	60438 6	Trichorhinophalangeal syndrome, type I;Trichorhinophalangeal syndrome, type III	AD
TRPV4	60542 7	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;Scapulooperoneal spinal muscular atrophy;Parastremmatic dwarfism;Hereditary motor and sensory neuropathy, type IIc;Digital arthropathy-brachydactyly, familial	AD
TSC1	60528 4	Lymphangioliomyomatosis;Focal cortical dysplasia, type II, somatic;Tuberous sclerosis-1	AD
TSC2	19109 2	?Focal cortical dysplasia, type II, somatic;Lymphangioliomyomatosis, somatic;Tuberous sclerosis-2	AD
TTC21B	61201 4	Short-rib thoracic dysplasia 4 with or without polydactyly;Nephronophthisis 12	AR, AD, AR
TTC37	61458 9	Trichohepatoenteric syndrome 1	AR
TTC8	60813 2	Bardet-Biedl syndrome 8;?Retinitis pigmentosa 51	AR
TULP1	60228 0	Retinitis pigmentosa 14;Leber congenital amaurosis 15	AR
TXNDC15	61777 8		
UGT1A1	19174 0	[Gilbert syndrome];Crigler-Najjar syndrome, type II;Crigler-Najjar syndrome, type I;Hyperbilirubinemia, familial transient neonatal;[Bilirubin, serum level of, QTL1]	AR
UMOD	19184 5	Glomerulocystic kidney disease with hyperuricemia and isosthenuria;Hyperuricemic nephropathy, familial juvenile 1;Medullary cystic kidney disease 2	AD
UPK3A	61155 9		

VHL	60853 7	Pheochromocytoma;von Hippel-Lindau syndrome;Renal cell carcinoma, somatic;Erythrocytosis, familial, 2	AD, AR
VIPAS39	61340 1	Arthrogyrosis, renal dysfunction, and cholestasis 2	AR
VPS33B	60855 2	Arthrogyrosis, renal dysfunction, and cholestasis 1	AR
WDR11	60641 7	Hypogonadotropic hypogonadism 14 with or without anosmia	AD
WDR19	60815 1	?Cranioectodermal dysplasia 4;?Short-rib thoracic dysplasia 5 with or without polydactyly;Nephronophthisis 13;Senior-Loken syndrome 8	AR
WDR35	61360 2	Cranioectodermal dysplasia 2;Short-rib thoracic dysplasia 7 with or without polydactyly	AR
WDR4	60592 4	Microcephaly, growth deficiency, seizures, and brain malformations;Galloway-Mowat syndrome 6	AR
WDR73	61614 4	Galloway-Mowat syndrome 1	AR
WNK1	60523 2	Neuropathy, hereditary sensory and autonomic, type II;Pseudohypoaldosteronism, type IIC	AR, AD
WNK4	60184 4	Pseudohypoaldosteronism, type IIB	AD
WNT4	60349 0	Mullerian aplasia and hyperandrogenism;?SERKAL syndrome	AD, AR
WNT5A	16497 5	Robinow syndrome, AD 1	AD
WNT7A	60157 0	Fuhrmann syndrome;Ulna and fibula, absence of, with severe limb deficiency	AR
WT1	60710 2	Denys-Drash syndrome;Mesothelioma, somatic;Frasier syndrome;Meacham syndrome;Wilms tumor, type 1;Nephrotic syndrome, type 4	AD, SM, AD
XPNPEP3	61355 3	Nephronophthisis-like nephropathy 1	AR
XYLT1	60812 4	Desbuquois dysplasia 2;Pseudoxanthoma elasticum, modifier of severity of	AR
ZIC3	30026 5	VACTERL association, X-linked;Congenital heart defects, nonsyndromic, 1, X-linked;Heterotaxy, visceral, 1, XL	XLR
ZMYND10	60707 0	Ciliary dyskinesia, primary, 22	AR
ZNF423	60455 7	Joubert syndrome 19;Nephronophthisis 14	