Nefro panel

versie	v2 (343 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ACE	106180	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Myocardial infarction, susceptibility to} (3); {Microvascular complications of diabetes 3}, 612624 (3); [Angiotensin I-converting enzyme, benign serum increase] (3); {SARS, progression of} (3); {Stroke, hemorrhagic}, 614519 (3)
ACTG2	102545	Visceral myopathy, 155310 (3), Autosomal dominant
ACTN4	604638	Glomerulosclerosis, focal segmental, 1, 603278 (3), Autosomal dominant
ADAMTS13	604134	Thrombotic thrombocytopenic purpura, hereditary, 274150 (3), Autosomal recessive
ADAMTS9	605421	No OMIM phenotype
ADCY10	605205	{Hypercalciuria, absorptive, susceptibility to}, 143870 (3), Autosomal dominant
AGT	106150	{Preeclampsia, susceptibility to} (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
AGTR1	106165	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Hypertension, essential}, 145500 (3), Multifactorial
AGXT	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
AHI1	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
ALDOB	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive
ALG1	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
ALG8	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
ALG9	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive
ALMS1	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ALPL	171760	Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant; Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive
AMN	605799	Imerslund-Grasbeck syndrome 2, 618882 (3), Autosomal recessive
ANKS6	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
ANLN	616027	Focal segmental glomerulosclerosis 8, 616032 (3), Autosomal dominant

ANOS1	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
AP2S1	602242	Hypocalciuric hypercalcemia, type III, 600740 (3), Autosomal dominant
APOA1	107680	Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463 (3); Amyloidosis, 3 or more types, 105200 (3), Autosomal dominant; ApoA-I and apoC-III deficiency, combined, 618463 (3)
APOA2	107670	Apolipoprotein A-II deficiency (3); {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant
APOL1	603743	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 (3); {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3)
APRT	102600	Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive
AQP2	107777	Diabetes insipidus, nephrogenic, 125800 (3), Autosomal recessive, Autosomal dominant
ARHGAP24	610586	No OMIM phenotype
ARHGDIA	601925	Nephrotic syndrome, type 8, 615244 (3), Autosomal recessive
ARL13B	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL6	608845	?Retinitis pigmentosa 55, 613575 (3); Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive
ARSA	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ATP6V0A4	605239	Renal tubular acidosis, distal, autosomal recessive, 602722 (3), Autosomal recessive
ATP6V1B1	192132	Renal tubular acidosis with deafness, 267300 (3), Autosomal recessive
ATP7B	606882	Wilson disease, 277900 (3), Autosomal recessive
AVP	192340	Diabetes insipidus, neurohypophyseal, 125700 (3), Autosomal dominant
AVPR2	300538	Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive; Diabetes insipidus, nephrogenic, 304800 (3), X-linked recessive
B9D1	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
B9D2	611951	Joubert syndrome 34, 614175 (3), Autosomal recessive; ?Meckel syndrome 10, 614175 (3), Autosomal recessive
BBIP1	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
BBS1	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
BBS10	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
BBS4	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive

BBS5	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BICC1	614295	{Renal dysplasia, cystic, susceptibility to}, 601331 (3), Autosomal dominant
BMP4	112262	Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant; Orofacial cleft 11, 600625 (3)
BMP7	112267	No OMIM phenotype
BSND	606412	Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive; Bartter syndrome, type 4a, 602522 (3), Autosomal recessive
C3	120700	{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; C3 deficiency, 613779 (3), Autosomal recessive; {Macular degeneration, age-related, 9}, 611378 (3)
C5	120900	[Eculizumab, poor response to], 615749 (3), Autosomal dominant; C5 deficiency, 609536 (3)
C8orf37	614477	Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive
CA2	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CACNA1S	114208	{Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant; {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Autosomal dominant; {Malignant hyperthermia, susceptibility to, 5}, 601887 (3), Autosomal dominant; Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant
CASR	601199	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant; {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3); Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant
CC2D2A	612013	Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive
CCDC39	613798	Ciliary dyskinesia, primary, 14, 613807 (3)
CCNQ	300708	STAR syndrome, 300707 (3), X-linked dominant
CD151	602243	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (3); [Blood group, Raph], 179620 (3)
CD2AP	604241	Glomerulosclerosis, focal segmental, 3, 607832 (3)
CD46	120920	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal recessive, Autosomal dominant
CDC5L	602868	No OMIM phenotype
CDKN1C	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant

CEP104	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive
CEP120	613446	Joubert syndrome 31, 617761 (3), Autosomal recessive; Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive
CEP164	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
CEP290	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive
CEP41	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
CEP83	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
CFB	138470	?Complement factor B deficiency, 615561 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant
CFH	134370	Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant; Basal laminar drusen, 126700 (3), Autosomal dominant; {Macular degeneration, age-related, 4}, 610698 (3)
CFHR1	134371	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant
CFHR2	600889	No OMIM phenotype
CFHR3	605336	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant
CFHR4	605337	No OMIM phenotype
CFHR5	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
CFI	217030	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; Complement factor I deficiency, 610984 (3), Autosomal recessive; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant
CHD1L	613039	No OMIM phenotype
CHD7	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHRM3	118494	Prune belly syndrome, 100100 (3), Autosomal recessive
CHRNA3	118503	{Lung cancer susceptibility 2}, 612052 (3); Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800 (3), Autosomal recessive

CLCN5	300008	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive; Dent disease, 300009 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive
CLCN7	602727	Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant; Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant
CLCNKA	602024	Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLCNKB	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLDN10	617579	HELIX syndrome, 617671 (3), Autosomal recessive
CLDN16	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
CLDN19	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
CNNM2	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation, 616418 (3), Autosomal recessive, Autosomal dominant
COL4A1	120130	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant
COL4A3	120070	Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive; Alport syndrome 3, autosomal dominant, 104200 (3), Autosomal dominant; Hematuria, benign familial, 141200 (3), Autosomal dominant
COL4A4	120131	Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive; Hematuria, familial benign, 141200 (3), Autosomal dominant
COL4A5	303630	Alport syndrome 1, X-linked, 301050 (3), X-linked dominant
COQ2	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
COQ6	614647	Coenzyme Q10 deficiency, primary, 6, 614650 (3), Autosomal recessive
COQ7	601683	?Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive
COQ8A	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COQ8B	615567	Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive

COQ9	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
CPLANE1	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
CPT2	600650	CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
CRB2	609720	Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive; Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive
CSPP1	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CTNS	606272	Cystinosis, nephropathic, 219800 (3), Autosomal recessive; Cystinosis, ocular nonnephropathic, 219750 (3), Autosomal recessive; Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3), Autosomal recessive; Cystinosis, atypical nephropathic, 219800 (3), Autosomal recessive
CUBN	602997	[Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive
CUL3	603136	Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant
CYP11B2	124080	Aldosterone to renin ratio raised (3); Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive; {Low renin hypertension, susceptibility to} (3); Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive
CYP24A1	126065	Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive
DAAM2	606627	No OMIM phenotype
DCDC2	605755	Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive; Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive
DGKE	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive
DMP1	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
DNAJB11	611341	Polycystic kidney disease 6 with or without polycystic liver disease, 618061 (3), Autosomal dominant
DSTYK	612666	Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant; Spastic paraplegia 23, 270750 (3), Autosomal recessive
DYNC2H1	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive
DYNC2I1 (WDR60)	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
DZIP1L	617570	Polycystic kidney disease 5, 617610 (3), Autosomal recessive

EGF	131530	Hypomagnesemia 4, renal, 611718 (3)
EGFR	131550	?Inflammatory skin and bowel disease, neonatal, 2, 616069 (3), Autosomal recessive; Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal dominant, Somatic mutation; Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal dominant, Somatic mutation; {Nonsmall cell lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
EHHADH	607037	?Fanconi renotubular syndrome 3, 615605 (3), Autosomal dominant
EMP2	602334	Nephrotic syndrome, type 10, 615861 (3), Autosomal recessive
ENPP1	173335	Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
EVC	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	607261	Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant; Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive
EYA1	601653	?Otofaciocervical syndrome, 166780 (3), Autosomal dominant; Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; Branchiootic syndrome 1, 602588 (3), Autosomal dominant
FAH	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
FAHD2A	No OMIM gene	No OMIM phenotype
FAM186B	No OMIM gene	No OMIM phenotype
FAM20A	611062	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3), Autosomal recessive
FAN1	613534	Interstitial nephritis, karyomegalic, 614817 (3), Autosomal recessive
FAT1	600976	No OMIM phenotype
FGA	134820	Dysfibrinogenemia, congenital, 616004 (3); Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Hypodysfibrinogenemia, congenital, 616004 (3); Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
FGF20	605558	?Renal hypodysplasia/aplasia 2, 615721 (3), Autosomal recessive
FGF23	605380	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3), Autosomal recessive; Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant

FGFR1	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
FH	136850	Fumarase deficiency, 606812 (3), Autosomal recessive; Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant
FLCN	607273	Pneumothorax, primary spontaneous, 173600 (3), Autosomal dominant; Renal carcinoma, chromophobe, somatic, 144700 (3); Birt-Hogg-Dube syndrome, 135150 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3)
FN1	135600	Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant; Spondylometaphyseal dysplasia, corner fracture type, 184255 (3), Autosomal dominant
FRAS1	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
FREM1	608944	Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant; Bifid nose with or without anorectal and renal anomalies, 608980 (3)
FREM2	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive
FXYD2	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
G6PC	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
GALNT3	601756	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive
GALT	606999	Galactosemia, 230400 (3), Autosomal recessive
GANAB	104160	Polycystic kidney disease 3, 600666 (3), Autosomal dominant
GATA3	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3), Autosomal dominant
GATM	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant
GDNF	600837	{Pheochromocytoma, modifier of}, 171300 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 3}, 613711 (3), Autosomal dominant; Central hypoventilation syndrome, 209880 (3), Autosomal dominant
GLA	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked

GLI3	165240	Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; {Hypothalamic hamartomas, somatic}, 241800 (3); Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant
GLIS2	608539	Nephronophthisis 7, 611498 (3)
GNA11	139313	Hypocalciuric hypercalcemia, type II, 145981 (3), Autosomal dominant; Hypocalcemia, autosomal dominant 2, 615361 (3), Autosomal dominant
GON7	617436	No OMIM phenotype
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
GRHPR	604296	Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive
GRIP1	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
GSN	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
HNF1B	189907	Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3)
HNF4A	600281	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant; Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant
HOGA1	613597	Hyperoxaluria, primary, type III, 613616 (3), Autosomal recessive
HPRT1	308000	HPRT-related gout, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
HPSE2	613469	Urofacial syndrome 1, 236730 (3), Autosomal recessive
HSD11B2	614232	Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive
IFT122	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT140	614620	Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
IFT172	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT27	615870	?Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
IFT43	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive
IFT80	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
IFT81	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
INCENP	604411	No OMIM phenotype

INF2	610982	Glomerulosclerosis, focal segmental, 5, 613237 (3); Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3), Autosomal dominant
INPP5E	613037	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
INVS	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IQCB1	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
ITGA3	605025	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748 (3), Autosomal recessive
ITGA8	604063	Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive
ITGB4	147557	Epidermolysis bullosa of hands and feet, 131800 (3), Autosomal dominant; Epidermolysis bullosa, junctional, with pyloric atresia, 226730 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive
JAG1	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3); Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
KANK1	607704	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
KANK2	614610	Palmoplantar keratoderma and woolly hair, 616099 (3), Autosomal recessive; Nephrotic syndrome, type 16, 617783 (3), Autosomal recessive
KANK4	614612	No OMIM phenotype
KCNA1	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNJ1	600359	Bartter syndrome, type 2, 241200 (3), Autosomal recessive
KCNJ10	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
KIAA0586	610178	Joubert syndrome 23, 616490 (3), Autosomal recessive; Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive
KIF14	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive
KIF7	611254	?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; Joubert syndrome 12, 200990 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
KL	604824	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994 (3), Autosomal recessive
KLHL3	605775	Pseudohypoaldosteronism, type IID, 614495 (3), Autosomal recessive, Autosomal dominant
LAGE3	300060	Galloway-Mowat syndrome 2, X-linked, 301006 (3), X-linked recessive

LAMB2	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive
LMNA	150330	Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Malouf syndrome, 212112 (3), Autosomal dominant
LMX1B	602575	Nail-patella syndrome, 161200 (3), Autosomal dominant
LRIG2	608869	Urofacial syndrome 2, 615112 (3), Autosomal recessive
LRP2	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRP5	603506	van Buchem disease, type 2, 607636 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; {Osteoporosis}, 166710 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant
LRP6	603507	{Coronary artery disease, autosomal dominant, 2}, 610947 (3), Autosomal dominant; Tooth agenesis, selective, 7, 616724 (3), Autosomal dominant
LYZ	153450	Amyloidosis, renal, 105200 (3), Autosomal dominant
LZTFL1	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
MAGED2	300470	Bartter syndrome, type 5, antenatal, transient, 300971 (3), X-linked recessive
MAGI2	606382	Nephrotic syndrome, type 15, 617609 (3), Autosomal recessive
MAPKBP1	616786	Nephronophthisis 20, 617271 (3), Autosomal recessive
MET	164860	{Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; Hepatocellular carcinoma, childhood type, somatic, 114550 (3); ?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive; Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3)
MKKS	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
MKS1	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive

MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MUC1	158340	Medullary cystic kidney disease 1, 174000 (3), Autosomal dominant
MYH9	160775	Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant; Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 (3), Autosomal dominant
MYO1E	601479	Glomerulosclerosis, focal segmental, 6, 614131 (3), Autosomal recessive
NEIL1	608844	No OMIM phenotype
NEK1	604588	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant; Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive
NEK8	609799	?Nephronophthisis 9, 613824 (3); Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive
NOTCH2	600275	Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant; Alagille syndrome 2, 610205 (3), Autosomal dominant
NPHP1	607100	Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive; Joubert syndrome 4, 609583 (3), Autosomal recessive
NPHP3	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive
NPHP4	607215	Nephronophthisis 4, 606966 (3), Autosomal recessive; Senior-Loken syndrome 4, 606996 (3), Autosomal recessive
NPHS1	602716	Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive
NPHS2	604766	Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive
NR3C2	600983	Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3), Autosomal dominant; Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3)
NUP107	607617	Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; ?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
NUP133	607613	Nephrotic syndrome, type 18, 618177 (3), Autosomal recessive; ?Galloway-Mowat syndrome 8, 618349 (3), Autosomal recessive
NUP205	614352	?Nephrotic syndrome, type 13, 616893 (3)
NUP93	614351	Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive
NXF5	300319	No OMIM phenotype
OCRL	300535	Lowe syndrome, 309000 (3), X-linked recessive; Dent disease 2, 300555 (3), X-linked recessive
OFD1	300170	Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Joubert syndrome 10, 300804 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive

PAX2	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
PAX8	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
PBX1	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
PCBD1	126090	Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive
PDE6D	602676	?Joubert syndrome 22, 615665 (3), Autosomal recessive
PDSS1	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
PDSS2	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
PHEX	300550	Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant
PKD1	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
PKD2	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
PKHD1	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
PLCE1	608414	Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive
PLG	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PMM2	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PRKCSH	177060	Polycystic liver disease 1, 174050 (3), Autosomal dominant
PSAP	176801	Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Combined SAP deficiency, 611721 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive
PTEN	601728	Prostate cancer, somatic, 176807 (3); {Glioma susceptibility 2}, 613028 (3); Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant
PTPRO	600579	Nephrotic syndrome, type 6, 614196 (3), Autosomal recessive
PYGM	608455	McArdle disease, 232600 (3), Autosomal recessive
RBM48	No OMIM gene	No OMIM phenotype
RCOR1	607675	No OMIM phenotype
REN	179820	[Hyperproreninemia] (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; Hyperuricemic nephropathy, familial juvenile 2, 613092 (3), Autosomal dominant

RET	164761	Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant
ROBO2	602431	Vesicoureteral reflux 2, 610878 (3), Autosomal dominant
RPGRIP1	605446	Cone-rod dystrophy 13, 608194 (3); Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
RPGRIP1L	610937	COACH syndrome, 216360 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive
RRM2B	604712	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive
SALL1	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant
SALL4	607343	Duane-radial ray syndrome, 607323 (3), Autosomal dominant; IVIC syndrome, 147750 (3), Autosomal dominant
SARS2	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive
SCARB2	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
SCNN1A	600228	Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant
SCNN1B	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; Liddle syndrome 1, 177200 (3), Autosomal dominant
SCNN1G	600761	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Liddle syndrome 2, 618114 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
SDCCAG8	613524	Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive; Senior-Loken syndrome 7, 613615 (3)

SDHB	185470	Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Pheochromocytoma, 171300 (3), Autosomal dominant; Paragangliomas 4, 115310 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3)
SDHD	602690	Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paraganglioma and gastric stromal sarcoma, 606864 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
SEC61A1	609213	Hyperuricemic nephropathy, familial juvenile, 4, 617056 (3), Autosomal dominant
SEC63	608648	Polycystic liver disease 2, 617004 (3), Autosomal dominant
SGPL1	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
SIX1	601205	Deafness, autosomal dominant 23, 605192 (3), Autosomal dominant; Branchiootic syndrome 3, 608389 (3), Autosomal dominant
SIX2	604994	No OMIM phenotype
SIX5	600963	Branchiootorenal syndrome 2, 610896 (3)
SLC12A1	600839	Bartter syndrome, type 1, 601678 (3), Autosomal recessive
SLC12A3	600968	Gitelman syndrome, 263800 (3), Autosomal recessive
SLC22A12	607096	Hypouricemia, renal, 220150 (3), Autosomal recessive
SLC26A1	610130	?Nephrolithiasis, calcium oxalate, 167030 (3), Autosomal recessive
SLC26A3	126650	Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive
SLC2A2	138160	Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
SLC2A9	606142	{Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal recessive, Autosomal dominant; Hypouricemia, renal, 2, 612076 (3), Autosomal recessive, Autosomal dominant
SLC34A1	182309	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; ?Fanconi renotubular syndrome 2, 613388 (3), Autosomal recessive
SLC34A3	609826	Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive
SLC37A4	602671	Glycogen storage disease Ic, 232240 (3), Autosomal recessive; Glycogen storage disease Ib, 232220 (3), Autosomal recessive
SLC3A1	104614	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
SLC41A1	610801	No OMIM phenotype
SLC4A1	109270	Renal tubular acidosis, distal, AD, 179800 (3), Autosomal dominant; [Blood group, Swann], 601550 (3); [Blood group, Froese], 601551 (3); [Blood group, Waldner], 112010 (3); Renal tubular acidosis, distal, AR, 611590 (3), Autosomal recessive; Spherocytosis, type 4, 612653 (3), Autosomal dominant; Cryohydrocytosis, 185020 (3), Autosomal dominant; Ovalocytosis, SA type, 166900 (3), Autosomal dominant; [Malaria, resistance to], 611162 (3); [Blood group, Diego], 110500 (3); [Blood group, Wright], 112050 (3)

SLC4A4	603345	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive
SLC7A9	604144	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
SLC9A3R1	604990	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287 (3), Autosomal dominant
SMARCAL1	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
SOX17	610928	Vesicoureteral reflux 3, 613674 (3), Autosomal dominant
SRGAP1	606523	{Thyroid cancer, nonmedullary, 2}, 188470 (3), Autosomal dominant, Somatic mutation
STRADA	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive
STX16	603666	Pseudohypoparathyroidism, type IB, 603233 (3), Autosomal dominant
SYNPO	608155	No OMIM phenotype
TBC1D1	609850	No OMIM phenotype
TBC1D8B	301027	Nephrotic syndrome, type 20, 301028 (3), X-linked
TBX18	604613	Congenital anomalies of kidney and urinary tract 2, 143400 (3), Autosomal dominant
TCTN1	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
TCTN2	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	613847	Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive; Joubert syndrome 18, 614815 (3), Autosomal recessive
THBD	188040	Thrombophilia due to thrombomodulin defect, 614486 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant
TMEM107	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
TMEM138	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
TMEM216	613277	Meckel syndrome 2, 603194 (3), Autosomal recessive; Joubert syndrome 2, 608091 (3), Autosomal recessive
TMEM231	614949	Meckel syndrome 11, 615397 (3), Autosomal recessive; Joubert syndrome 20, 614970 (3), Autosomal recessive
TMEM237	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TMEM67	609884	Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive
TNFRSF25	603366	No OMIM phenotype
TNXB	600985	Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant
TP53RK	608679	Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive

TPRKB	608680	Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive
TRAP1	606219	No OMIM phenotype
TRIM32	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TRPC6	603652	Glomerulosclerosis, focal segmental, 2, 603965 (3), Autosomal dominant
TRPM6	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
TSC1	605284	Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, 606690 (3)
TSC2	191092	Tuberous sclerosis-2, 613254 (3), Autosomal dominant; ?Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, somatic, 606690 (3)
TTC21B	612014	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
TTC8	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
UMOD	191845	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 (3); Hyperuricemic nephropathy, familial juvenile 1, 162000 (3), Autosomal dominant; Medullary cystic kidney disease 2, 603860 (3)
UPK3A	611559	No OMIM phenotype
UPK3B	611887	No OMIM phenotype
VDR	601769	Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
VHL	608537	Pheochromocytoma, 171300 (3), Autosomal dominant; Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Hemangioblastoma, cerebellar, somatic (3)
VIPAS39	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
WDPCP	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
WDR19	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive

WDR35	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
WDR4	605924	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive; Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive
WDR73	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
WNK1	605232	Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive
WNK4	601844	Pseudohypoaldosteronism, type IIB, 614491 (3), Autosomal dominant
WNT4	603490	?SERKAL syndrome, 611812 (3), Autosomal recessive; Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant
WT1	607102	Mesothelioma, somatic, 156240 (3); Wilms tumor, type 1, 194070 (3), Autosomal dominant, Somatic mutation; Frasier syndrome, 136680 (3), Autosomal dominant, Somatic mutation; Denys-Drash syndrome, 194080 (3), Autosomal dominant, Somatic mutation; Meacham syndrome, 608978 (3); Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant
XDH	607633	Xanthinuria, type I, 278300 (3), Autosomal recessive
XPNPEP3	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
XPO5	607845	No OMIM phenotype
YRDC	612276	No OMIM phenotype
ZMPSTE24	606480	Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive
ZMYM2	602221	No OMIM phenotype
ZNF423	604557	Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant; Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: Sep 01, 2020

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

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.