

Nefro panel		
versie	V1 (328 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ACE</i>	106180	[Angiotensin I-converting enzyme, benign serum increase] (3); {Microvascular complications of diabetes 3}, 612624 (3); {Myocardial infarction, susceptibility to} (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {SARS, progression of} (3); {Stroke, hemorrhagic}, 614519 (3)
<i>ACTN4</i>	604638	Glomerulosclerosis, focal segmental, 1, 603278 (3), Autosomal dominant
<i>ADAMTS13</i>	604134	Thrombotic thrombocytopenic purpura, familial, 274150 (3), Autosomal recessive
<i>ADCY10</i>	605205	{Hypercalciuria, absorptive, susceptibility to}, 143870 (3), Autosomal dominant
<i>AGT</i>	106150	{Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; {Preeclampsia, susceptibility to} (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive
<i>AGTR1</i>	106165	{Hypertension, essential}, 145500 (3), Multifactorial; Renal tubular dysgenesis, 267430 (3), Autosomal recessive
<i>AGXT</i>	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
<i>AHI1</i>	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
<i>ALDOB</i>	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive
<i>ALG1</i>	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
<i>ALG8</i>	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>ALPL</i>	171760	Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant
<i>AMN</i>	605799	Megaloblastic anemia-1, Norwegian type, 261100 (3), Autosomal recessive
<i>ANKS6</i>	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
<i>ANLN</i>	616027	Focal segmental glomerulosclerosis 8, 616032 (3), Autosomal dominant
<i>ANOS1</i>	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive

<i>AP2S1</i>	602242	Hypocalciuric hypercalcemia, type III, 600740 (3), Autosomal dominant
<i>APOA1</i>	107680	Amyloidosis, 3 or more types, 105200 (3), Autosomal dominant; ApoA-I and apoC-III deficiency, combined, 618463 (3); Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463 (3)
<i>APOA2</i>	107670	Apolipoprotein A-II deficiency (3); {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant
<i>APOL1</i>	603743	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 (3); {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3)
<i>APRT</i>	102600	Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive
<i>AQP2</i>	107777	Diabetes insipidus, nephrogenic, 125800 (3), Autosomal recessive, Autosomal dominant
<i>ARHGAP24</i>	610586	No OMIM phenotype
<i>ARHGDI1</i>	601925	Nephrotic syndrome, type 8, 615244 (3), Autosomal recessive
<i>ARL13B</i>	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
<i>ARL6</i>	608845	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive; ?Retinitis pigmentosa 55, 613575 (3)
<i>ARSA</i>	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<i>ATP6VOA4</i>	605239	Renal tubular acidosis, distal, autosomal recessive, 602722 (3)
<i>ATP6V1B1</i>	192132	Renal tubular acidosis with deafness, 267300 (3), Autosomal recessive
<i>ATP7B</i>	606882	Wilson disease, 277900 (3), Autosomal recessive
<i>AVP</i>	192340	Diabetes insipidus, neurohypophyseal, 125700 (3), Autosomal dominant
<i>AVPR2</i>	300538	Diabetes insipidus, nephrogenic, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive
<i>B9D1</i>	614144	Joubert syndrome 27, 617120 (3), Autosomal recessive; ?Meckel syndrome 9, 614209 (3), Autosomal recessive
<i>B9D2</i>	611951	Joubert syndrome 34, 614175 (3), Autosomal recessive; ?Meckel syndrome 10, 614175 (3), Autosomal recessive
<i>BBIP1</i>	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
<i>BBS1</i>	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
<i>BBS10</i>	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
<i>BBS12</i>	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
<i>BBS2</i>	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
<i>BBS4</i>	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
<i>BBS5</i>	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
<i>BBS7</i>	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
<i>BBS9</i>	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive

<i>BICC1</i>	614295	{Renal dysplasia, cystic, susceptibility to}, 601331 (3), Autosomal dominant
<i>BMP4</i>	112262	Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant; Orofacial cleft 11, 600625 (3)
<i>BMP7</i>	112267	No OMIM phenotype
<i>BSND</i>	606412	Bartter syndrome, type 4a, 602522 (3), Autosomal recessive; Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive
<i>C3</i>	120700	C3 deficiency, 613779 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; {Macular degeneration, age-related, 9}, 611378 (3)
<i>C5</i>	120900	C5 deficiency, 609536 (3); [Eculizumab, poor response to], 615749 (3), Autosomal dominant
<i>C8orf37</i>	614477	Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Retinitis pigmentosa 64, 614500 (3), Autosomal recessive
<i>CA2</i>	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
<i>CACNA1S</i>	114208	Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant; {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Autosomal dominant
<i>CASR</i>	601199	{Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3); Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant
<i>CC2D2A</i>	612013	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive
<i>CCDC39</i>	613798	Ciliary dyskinesia, primary, 14, 613807 (3)
<i>CCNQ (FAM58A)</i>	300708	STAR syndrome, 300707 (3), X-linked dominant
<i>CD151</i>	602243	[Blood group, Raph], 179620 (3); Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (3)
<i>CD2AP</i>	604241	Glomerulosclerosis, focal segmental, 3, 607832 (3)
<i>CD46</i>	120920	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal recessive, Autosomal dominant
<i>CDC5L</i>	602868	No OMIM phenotype
<i>CDKN1C</i>	600856	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; IMAGE syndrome, 614732 (3), Autosomal dominant
<i>CEP104</i>	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive

<i>CEP120</i>	613446	Joubert syndrome 31, 617761 (3), Autosomal recessive; Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive
<i>CEP164</i>	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
<i>CEP290</i>	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Meckel syndrome 4, 611134 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive
<i>CEP41</i>	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
<i>CEP83</i>	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
<i>CFB</i>	138470	?Complement factor B deficiency, 615561 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3)
<i>CFH</i>	134370	Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, 4}, 610698 (3)
<i>CFHR1</i>	134371	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant
<i>CFHR2</i>	600889	No OMIM phenotype
<i>CFHR3</i>	605336	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant
<i>CFHR4</i>	605337	No OMIM phenotype
<i>CFHR5</i>	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
<i>CFI</i>	217030	Complement factor I deficiency, 610984 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant
<i>CHD1L</i>	613039	No OMIM phenotype
<i>CHD7</i>	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
<i>CHRM3</i>	118494	?Prune belly syndrome, 100100 (3), Autosomal recessive
<i>CLCN5</i>	300008	Dent disease, 300009 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive; Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive

<i>CLCN7</i>	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive
<i>CLCNKA</i>	602024	Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
<i>CLCNKB</i>	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
<i>CLDN10</i>	617579	HELIX syndrome, 617671 (3), Autosomal recessive
<i>CLDN16</i>	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
<i>CLDN19</i>	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
<i>CNNM2</i>	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation, 616418 (3), Autosomal recessive, Autosomal dominant
<i>COL4A1</i>	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); Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant
<i>COL4A3</i>	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
<i>COL4A4</i>	120131	Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive; Hematuria, familial benign, 141200 (3), Autosomal dominant
<i>COL4A5</i>	303630	Alport syndrome 1, X-linked, 301050 (3), X-linked dominant
<i>COQ2</i>	609825	Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive; {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant
<i>COQ6</i>	614647	Coenzyme Q10 deficiency, primary, 6, 614650 (3), Autosomal recessive
<i>COQ7</i>	601683	?Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive
<i>COQ8A</i>	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
<i>COQ8B</i>	615567	Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive
<i>COQ9</i>	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
<i>CPLANE1 (C5orf42)</i>	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive

<i>CPT2</i>	600650	CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
<i>CRB2</i>	609720	Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive
<i>CSPP1</i>	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
<i>CTNS</i>	606272	Cystinosis, atypical nephropathic, 219800 (3), Autosomal recessive; Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3), Autosomal recessive; Cystinosis, nephropathic, 219800 (3), Autosomal recessive; Cystinosis, ocular nonnephropathic, 219750 (3), Autosomal recessive
<i>CUBN</i>	602997	Megaloblastic anemia-1, Finnish type, 261100 (3), Autosomal recessive
<i>CUL3</i>	603136	Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant
<i>CYP11B2</i>	124080	Aldosterone to renin ratio raised (3); Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive; Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive; {Low renin hypertension, susceptibility to} (3)
<i>CYP24A1</i>	126065	Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive
<i>DCDC2</i>	605755	?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Nephronophthisis 19, 616217 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive
<i>DGKE</i>	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive
<i>DMP1</i>	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
<i>DNAJB11</i>	611341	Polycystic kidney disease 6 with or without polycystic liver disease, 618061 (3), Autosomal dominant
<i>DSTYK</i>	612666	Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant; Spastic paraplegia 23, 270750 (3), Autosomal recessive
<i>DYNC2H1</i>	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive
<i>DZIP1L</i>	617570	Polycystic kidney disease 5, 617610 (3), Autosomal recessive
<i>EGF</i>	131530	Hypomagnesemia 4, renal, 611718 (3)

<i>EGFR</i>	131550	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal dominant, Somatic mutation; ?Inflammatory skin and bowel disease, neonatal, 2, 616069 (3), Autosomal recessive; Non-small cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal dominant, Somatic mutation; {Non-small cell lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
<i>EHHADH</i>	607037	?Fanconi renal tubular syndrome 3, 615605 (3), Autosomal dominant
<i>EMP2</i>	602334	Nephrotic syndrome, type 10, 615861 (3), Autosomal recessive
<i>ENPP1</i>	173335	Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>EVC</i>	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EVC2</i>	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EYA1</i>	601653	Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant
<i>FAH</i>	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
<i>FAHD2A</i>	No OMIM gene	No OMIM phenotype
<i>FAM186B</i>	No OMIM gene	No OMIM phenotype
<i>FAM20A</i>	611062	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3), Autosomal recessive
<i>FAN1</i>	613534	Interstitial nephritis, karyomegalic, 614817 (3), Autosomal recessive
<i>FAT1</i>	600976	No OMIM phenotype
<i>FGA</i>	134820	Afibrinogenemia, congenital, 202400 (3), Autosomal recessive; Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Dysfibrinogenemia, congenital, 616004 (3); Hypodysfibrinogenemia, congenital, 616004 (3)
<i>FGF20</i>	605558	?Renal hypodysplasia/aplasia 2, 615721 (3), Autosomal recessive
<i>FGF23</i>	605380	Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant; Osteomalacia, tumor-induced (1); Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3)
<i>FGFR1</i>	136350	Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3); Hartsfield syndrome, 615465 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3),

		Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Trigenocephaly 1, 190440 (3), Autosomal dominant
<i>FH</i>	136850	Fumarase deficiency, 606812 (3), Autosomal recessive; Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant
<i>FLCN</i>	607273	Birt-Hogg-Dube syndrome, 135150 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Pneumothorax, primary spontaneous, 173600 (3), Autosomal dominant; Renal carcinoma, chromophobe, somatic, 144700 (3)
<i>FN1</i>	135600	Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant; Plasma fibronectin deficiency, 614101 (1), Autosomal dominant; Spondylometaphyseal dysplasia, corner fracture type, 184255 (3), Autosomal dominant
<i>FRAS1</i>	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
<i>FREM1</i>	608944	Bifid nose with or without anorectal and renal anomalies, 608980 (3); Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Trigenocephaly 2, 614485 (3), Autosomal dominant
<i>FREM2</i>	608945	Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive; Fraser syndrome 2, 617666 (3), Autosomal recessive
<i>FXD2</i>	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
<i>G6PC</i>	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
<i>GALNT3</i>	601756	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive
<i>GALT</i>	606999	Galactosemia, 230400 (3), Autosomal recessive
<i>GANAB</i>	104160	Polycystic kidney disease 3, 600666 (3), Autosomal dominant
<i>GATA3</i>	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3), Autosomal dominant
<i>GDNF</i>	600837	Central hypoventilation syndrome, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 3}, 613711 (3), Autosomal dominant; {Pheochromocytoma, modifier of}, 171300 (3), Autosomal dominant
<i>GLA</i>	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked
<i>GLIS2</i>	608539	Nephronophthisis 7, 611498 (3)
<i>GNA11</i>	139313	Hypocalcemia, autosomal dominant 2, 615361 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type II, 145981 (3), Autosomal dominant
<i>GON7</i>	617436	No OMIM phenotype
<i>GPC3</i>	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)

<i>GRHPR</i>	604296	Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive
<i>GRIP1</i>	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
<i>GSN</i>	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
<i>HNF1B</i>	189907	Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3); Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant
<i>HNF4A</i>	600281	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant
<i>HOGA1</i>	613597	Hyperoxaluria, primary, type III, 613616 (3)
<i>HPRT1</i>	308000	HPRT-related gout, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
<i>HPSE2</i>	613469	Urofacial syndrome 1, 236730 (3), Autosomal recessive
<i>HSD11B2</i>	614232	Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive
<i>IFT122</i>	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
<i>IFT140</i>	614620	Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
<i>IFT172</i>	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
<i>IFT27</i>	615870	?Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
<i>IFT43</i>	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive
<i>IFT80</i>	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
<i>IFT81</i>	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
<i>INCENP</i>	604411	No OMIM phenotype
<i>INF2</i>	610982	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3), Autosomal dominant; Glomerulosclerosis, focal segmental, 5, 613237 (3)
<i>INPP5E</i>	613037	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
<i>INVS</i>	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
<i>IQCB1</i>	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
<i>ITGA3</i>	605025	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748 (3), Autosomal recessive
<i>ITGA8</i>	604063	Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive

<i>ITGB4</i>	147557	Epidermolysis bullosa of hands and feet, 131800 (3), Autosomal dominant; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, with pyloric atresia, 226730 (3), Autosomal recessive
<i>JAG1</i>	601920	Alagille syndrome 1, 118450 (3), Autosomal dominant; ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>KANK1</i>	607704	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
<i>KANK2</i>	614610	Nephrotic syndrome, type 16, 617783 (3), Autosomal recessive; Palmoplantar keratoderma and woolly hair, 616099 (3), Autosomal recessive
<i>KANK4</i>	614612	No OMIM phenotype
<i>KCNA1</i>	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
<i>KCNJ1</i>	600359	Bartter syndrome, type 2, 241200 (3), Autosomal recessive
<i>KCNJ10</i>	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
<i>KIAA0586</i>	610178	Joubert syndrome 23, 616490 (3), Autosomal recessive; Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive
<i>KIF14</i>	611279	?Meckel syndrome 12, 616258 (3), Autosomal recessive; Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive
<i>KIF7</i>	611254	Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Al-Gazali-Bakalnova syndrome, 607131 (3), Autosomal recessive; ?Hydroletharus syndrome 2, 614120 (3), Autosomal recessive; Joubert syndrome 12, 200990 (3), Autosomal recessive
<i>KL</i>	604824	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994 (3)
<i>KLHL3</i>	605775	Pseudohypoaldosteronism, type IID, 614495 (3), Autosomal recessive, Autosomal dominant
<i>LAGE3</i>	300060	Galloway-Mowat syndrome 2, X-linked, 301006 (3), X-linked recessive
<i>LAMB2</i>	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive
<i>LMNA</i>	150330	Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (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; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Muscular dystrophy, congenital, 613205

(3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3),
Autosomal recessive

<i>LMX1B</i>	602575	Nail-patella syndrome, 161200 (3), Autosomal dominant
<i>LRIG2</i>	608869	Urofacial syndrome 2, 615112 (3), Autosomal recessive
<i>LRP2</i>	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
<i>LRP5</i>	603506	[Bone mineral density variability 1], 601884 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; {Osteoporosis}, 166710 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; van Buchem disease, type 2, 607636 (3), Autosomal dominant
<i>LRP6</i>	603507	{Coronary artery disease, autosomal dominant, 2}, 610947 (3), Autosomal dominant; Tooth agenesis, selective, 7, 616724 (3), Autosomal dominant
<i>LYZ</i>	153450	Amyloidosis, renal, 105200 (3), Autosomal dominant
<i>LZTFL1</i>	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
<i>MAGED2</i>	300470	Bartter syndrome, type 5, antenatal, transient, 300971 (3), X-linked recessive
<i>MAGI2</i>	606382	Nephrotic syndrome, type 15, 617609 (3), Autosomal recessive
<i>MET</i>	164860	?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive; Hepatocellular carcinoma, childhood type, somatic, 114550 (3); {Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3)
<i>MKKS</i>	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
<i>MMACHC</i>	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive

<i>MYH9</i>	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
<i>MYO1E</i>	601479	Glomerulosclerosis, focal segmental, 6, 614131 (3), Autosomal recessive
<i>NEIL1</i>	608844	No OMIM phenotype
<i>NEK1</i>	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
<i>NEK8</i>	609799	?Nephronophthisis 9, 613824 (3); Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive
<i>NOTCH2</i>	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
<i>NPHP1</i>	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
<i>NPHP3</i>	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive
<i>NPHP4</i>	607215	Nephronophthisis 4, 606966 (3), Autosomal recessive; Senior-Loken syndrome 4, 606996 (3), Autosomal recessive
<i>NPHS1</i>	602716	Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive
<i>NPHS2</i>	604766	Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive
<i>NR3C2</i>	600983	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3); Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3), Autosomal dominant
<i>NUP107</i>	607617	Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive; ?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive
<i>NUP133</i>	607613	?Galloway-Mowat syndrome 8, 618349 (3), Autosomal recessive; Nephrotic syndrome, type 18, 618177 (3), Autosomal recessive
<i>NUP205</i>	614352	?Nephrotic syndrome, type 13, 616893 (3)
<i>NUP93</i>	614351	Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive
<i>NXF5</i>	300319	No OMIM phenotype
<i>OCRL</i>	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
<i>OFD1</i>	300170	Joubert syndrome 10, 300804 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
<i>PAX2</i>	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
<i>PAX8</i>	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant

<i>PBX1</i>	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
<i>PCBD1</i>	126090	Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive
<i>PDE6D</i>	602676	?Joubert syndrome 22, 615665 (3), Autosomal recessive
<i>PDSS1</i>	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
<i>PDSS2</i>	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
<i>PHEX</i>	300550	Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant
<i>PKD1</i>	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
<i>PKD2</i>	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
<i>PKHD1</i>	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
<i>PLCE1</i>	608414	Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive
<i>PLG</i>	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
<i>PMM2</i>	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
<i>PRKCSH</i>	177060	Polycystic liver disease 1, 174050 (3), Autosomal dominant
<i>PSAP</i>	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive
<i>PTEN</i>	601728	Cowden syndrome 1, 158350 (3), Autosomal dominant; {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3)
<i>PTPRO</i>	600579	Nephrotic syndrome, type 6, 614196 (3), Autosomal recessive
<i>PYGM</i>	608455	McArdle disease, 232600 (3), Autosomal recessive
<i>RBM48</i>	No OMIM gene	No OMIM phenotype
<i>RCOR1</i>	607675	No OMIM phenotype
<i>REN</i>	179820	[Hyperproreninemia] (3); Hyperuricemic nephropathy, familial juvenile 2, 613092 (3), Autosomal dominant; Renal tubular dysgenesis, 267430 (3), Autosomal recessive
<i>RET</i>	164761	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; Multiple

		endocrine neoplasia IIB, 162300 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
<i>ROBO2</i>	602431	Vesicoureteral reflux 2, 610878 (3), Autosomal dominant
<i>RPGRIP1</i>	605446	Cone-rod dystrophy 13, 608194 (3); Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
<i>RPGRIP1L</i>	610937	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
<i>SALL1</i>	602218	Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant; Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant
<i>SALL4</i>	607343	Duane-radial ray syndrome, 607323 (3), Autosomal dominant; IVIC syndrome, 147750 (3), Autosomal dominant
<i>SARS2</i>	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive
<i>SCARB2</i>	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
<i>SCNN1A</i>	600228	Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
<i>SCNN1B</i>	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Liddle syndrome 1, 177200 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
<i>SCNN1G</i>	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
<i>SDCCAG8</i>	613524	Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive; Senior-Loken syndrome 7, 613615 (3)
<i>SDHB</i>	185470	Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Paraganglioma and gastric stromal sarcoma, 606864 (3); Paragangliomas 4, 115310 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
<i>SDHD</i>	602690	Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paraganglioma and gastric stromal sarcoma, 606864 (3); Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant

<i>SEC61A1</i>	609213	Hyperuricemic nephropathy, familial juvenile, 4, 617056 (3), Autosomal dominant
<i>SEC63</i>	608648	Polycystic liver disease 2, 617004 (3), Autosomal dominant
<i>SIX1</i>	601205	Branchiootic syndrome 3, 608389 (3), Autosomal dominant; Deafness, autosomal dominant 23, 605192 (3), Autosomal dominant
<i>SIX2</i>	604994	No OMIM phenotype
<i>SIX5</i>	600963	Branchiootorenal syndrome 2, 610896 (3)
<i>SLC12A1</i>	600839	Bartter syndrome, type 1, 601678 (3), Autosomal recessive
<i>SLC12A3</i>	600968	Gitelman syndrome, 263800 (3), Autosomal recessive
<i>SLC22A12</i>	607096	Hypouricemia, renal, 220150 (3), Autosomal recessive
<i>SLC26A3</i>	126650	Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive
<i>SLC2A2</i>	138160	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive
<i>SLC2A9</i>	606142	Hypouricemia, renal, 2, 612076 (3), Autosomal recessive, Autosomal dominant; {Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal recessive, Autosomal dominant
<i>SLC34A1</i>	182309	?Fanconi renotubular syndrome 2, 613388 (3), Autosomal recessive; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant
<i>SLC34A3</i>	609826	Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive
<i>SLC37A4</i>	602671	Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Glycogen storage disease Ic, 232240 (3), Autosomal recessive
<i>SLC3A1</i>	104614	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
<i>SLC41A1</i>	610801	No OMIM phenotype
<i>SLC4A1</i>	109270	[Blood group, Diego], 110500 (3); [Blood group, Froese], 601551 (3); [Blood group, Swann], 601550 (3); [Blood group, Waldner], 112010 (3); [Blood group, Wright], 112050 (3); Cryohydrocytosis, 185020 (3), Autosomal dominant; [Malaria, resistance to], 611162 (3); Ovalocytosis, SA type, 166900 (3), Autosomal dominant; Renal tubular acidosis, distal, AD, 179800 (3), Autosomal dominant; Renal tubular acidosis, distal, AR, 611590 (3), Autosomal recessive; Spherocytosis, type 4, 612653 (3), Autosomal dominant
<i>SLC4A4</i>	603345	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive
<i>SLC7A9</i>	604144	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
<i>SLC9A3R1</i>	604990	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287 (3), Autosomal dominant
<i>SMARCAL1</i>	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
<i>SOX17</i>	610928	Vesicoureteral reflux 3, 613674 (3), Autosomal dominant
<i>SRGAP1</i>	606523	{Thyroid cancer, nonmedullary, 2}, 188470 (3), Autosomal dominant, Somatic mutation

<i>STX16</i>	603666	Pseudohypoparathyroidism, type IB, 603233 (3), Autosomal dominant
<i>SYNPO</i>	608155	No OMIM phenotype
<i>TBC1D1</i>	609850	No OMIM phenotype
<i>TBX18</i>	604613	Congenital anomalies of kidney and urinary tract 2, 143400 (3), Autosomal dominant
<i>TCTN1</i>	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
<i>TCTN2</i>	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
<i>TCTN3</i>	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
<i>THBD</i>	188040	{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant; Thrombophilia due to thrombomodulin defect, 614486 (3)
<i>TMEM107</i>	616183	?Joubert syndrome 29, 617562 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive
<i>TMEM138</i>	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
<i>TMEM216</i>	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
<i>TMEM231</i>	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
<i>TMEM237</i>	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
<i>TMEM67</i>	609884	{Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive; ?RHYS syndrome, 602152 (3), Autosomal recessive
<i>TNFRSF25</i>	603366	No OMIM phenotype
<i>TNXB</i>	600985	Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant
<i>TP53RK</i>	608679	Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive
<i>TPRKB</i>	608680	Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive
<i>TRAP1</i>	606219	No OMIM phenotype
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
<i>TRPC6</i>	603652	Glomerulosclerosis, focal segmental, 2, 603965 (3), Autosomal dominant
<i>TRPM6</i>	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
<i>TSC1</i>	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioliomyomatosis, 606690 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant

<i>TSC2</i>	191092	?Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangiomyomatosis, somatic, 606690 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
<i>TTC21B</i>	612014	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
<i>TTC8</i>	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
<i>UMOD</i>	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)
<i>UPK3A</i>	611559	No OMIM phenotype
<i>UPK3B</i>	611887	No OMIM phenotype
<i>VDR</i>	601769	?Osteoporosis, involutional, 166710 (1), Autosomal dominant; Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
<i>VHL</i>	608537	Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; Hemangioblastoma, cerebellar, somatic (3); Pheochromocytoma, 171300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant
<i>VIPAS39</i>	613401	Arthrogyrosis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
<i>VPS33B</i>	608552	Arthrogyrosis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
<i>WDPCP</i>	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
<i>WDR19</i>	608151	?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
<i>WDR35</i>	613602	Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive; Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive
<i>WDR4</i>	605924	Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive
<i>WDR60</i>	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
<i>WDR73</i>	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive

<i>WNK1</i>	605232	Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive; Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant
<i>WNK4</i>	601844	Pseudohypoaldosteronism, type IIB, 614491 (3), Autosomal dominant
<i>WNT4</i>	603490	Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant; ?SERKAL syndrome, 611812 (3), Autosomal recessive
<i>WT1</i>	607102	Denys-Drash syndrome, 194080 (3), Autosomal dominant, Somatic mutation; Frasier syndrome, 136680 (3), Autosomal dominant, Somatic mutation; Meacham syndrome, 608978 (3); Mesothelioma, somatic, 156240 (3); Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Wilms tumor, type 1, 194070 (3), Autosomal dominant, Somatic mutation
<i>XDH</i>	607633	Xanthinuria, type I, 278300 (3), Autosomal recessive
<i>XPNPEP3</i>	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
<i>XPO5</i>	607845	No OMIM phenotype
<i>YRDC</i>	612276	No OMIM phenotype
<i>ZMPSTE24</i>	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
<i>ZNF423</i>	604557	Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant; Nephronophthisis 14, 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: Sept 30, 2019

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.