Nefro

Gene panel

Gene panel information

Gene panel	**Nefro**
Version	3
Total genes	392
Activation date	Friday 11 october 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ACE	99.98 %	106180	{Stroke, hemorrhagic}, 614519 (3); 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)
ACTG2	99.99 %	102545	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431 (3), Autosomal dominant; Visceral myopathy 1, 155310 (3), Autosomal dominant
ACTN4	100 %	604638	Glomerulosclerosis, focal segmental, 1, 603278 (3), Autosomal dominant
ADAMTS13	100 %	604134	Thrombotic thrombocytopenic purpura, hereditary, 274150 (3), Autosomal recessive
ADAMTS9	99.94 %	605421	*No OMIM phenotypes*
AGT	100 %	106150	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Preeclampsia, susceptibility to} (3); {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
AGTR1	99.97 %	106165	{Hypertension, essential}, 145500 (3), Multifactorial; Renal tubular dysgenesis, 267430 (3), Autosomal recessive
AGXT	100 %	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
AHI1	99.86 %	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
ALDOB	100 %	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive
ALG1	86.66 %	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
ALG5	99.94 %	604565	Polycystic kidney disease 7, 620056 (3), Autosomal dominant
ALG8	95.49 %	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	99.73 %	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive
ALMS1	99.9 %	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ALPL	99.88 %	171760	Odontohypophosphatasia, 146300 (3), Autosomal dominant, Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, adult, 146300 (3), Autosomal dominant, Autosomal recessive
AMN	100 %	605799	Imerslund-Grasbeck syndrome 2, 618882 (3), Autosomal recessive
ANKS6	100 %	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
ANLN	99.85 %	616027	Focal segmental glomerulosclerosis 8, 616032 (3), Autosomal dominant
ANOS1	99.96 %	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
AP2S1	99.98 %	602242	Hypocalciuric hypercalcemia, type III, 600740 (3), Autosomal dominant

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APOA1	100 %	107680	Hypoalphalipoproteinemia, primary, 2, 618463 (3), Autosomal recessive; Amyloidosis, hereditary systemic 3, 620657 (3); Hypoalphalipoproteinemia, primary, 2, intermediate, 619836 (3), Autosomal dominant
APOA2	99.84 %	107670	Apolipoprotein A-II deficiency (3); {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant, Autosomal recessive
APOC2	99.97 %	608083	Hyperlipoproteinemia, type Ib, 207750 (3), Autosomal recessive
APOC3	100 %	107720	Apolipoprotein C-III deficiency, 614028 (3)
APOE	99.98 %	107741	Alzheimer disease 2, 104310 (3), Autosomal dominant; Sea-blue histiocyte disease, 269600 (3), Autosomal recessive; {?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822 (3), Autosomal dominant; {Coronary artery disease, severe, susceptibility to}, 617347 (3); Lipoprotein glomerulopathy, 611771 (3); {?Macular degeneration, age-related}, 603075 (3), Autosomal dominant; Hyperlipoproteinemia, type III, 617347 (3)
APOL1	99.99 %	603743	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3), Autosomal dominant
APRT	100 %	102600	Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive
AQP2	100 %	107777	Diabetes insipidus, nephrogenic, 2, 125800 (3), Autosomal dominant, Autosomal recessive
ARHGDIA	100 %	601925	Nephrotic syndrome, type 8, 615244 (3), Autosomal recessive
ARL13B	99.53 %	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL6	99.9 %	608845	Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Digenic recessive, Autosomal recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive
ARMC9	99.77 %	617612	Joubert syndrome 30, 617622 (3), Autosomal recessive
ARSA	99.99 %	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ATP1A1	98.41 %	182310	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant
ATP6V0A4	99.93 %	605239	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722 (3), Autosomal recessive
ATP6V1B1	99.98 %	192132	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300 (3), Autosomal recessive
ATP7B	100 %	606882	Wilson disease, 277900 (3), Autosomal recessive
AVP	100 %	192340	Diabetes insipidus, neurohypophyseal, 125700 (3), Autosomal dominant
AVPR2	100 %	300538	Diabetes insipidus, nephrogenic, 1, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive
B2M	100 %	109700	Amyloidosis, hereditary systemic 6, 620659 (3); Immunodeficiency 43, 241600 (3), Autosomal recessive
B9D1	99.8 %	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
B9D2	99.88 %	611951	?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive
BBIP1	99.99 %	613605	Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
BBS1	100 %	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
BBS10	99.98 %	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	100 %	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	99.9 %	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive

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BBS4	99.88 %	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	99 %	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	99.42 %	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	99.75 %	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BMP4	100 %	112262	Orofacial cleft 11, 600625 (3); Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant
BNC2	99.97 %	608669	Lower urinary tract obstruction, congenital, 618612 (3), Autosomal dominant
BSND	99.92 %	606412	Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive; Bartter syndrome, type 4a, 602522 (3), Autosomal recessive
C3	100 %	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)
C5	99.92 %	120900	C5 deficiency, 609536 (3), Autosomal recessive; [Eculizumab, poor response to], 615749 (3), Autosomal dominant
CA2	99.62 %	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CACNA1H	100 %	607904	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 (3); Hyperaldosteronism, familial, type IV, 617027 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 (3)
CACNA1S	99.96 %	114208	{Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Autosomal dominant; Congenital myopathy 18 due to dihydropyridine receptor defect, 620246 (3), Autosomal dominant, Autosomal recessive; Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant
CASR	99.99 %	601199	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hyperparathyroidism, neonatal, 239200 (3), Autosomal dominant, Autosomal recessive; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant; {?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3), Autosomal dominant
CC2D2A	99.95 %	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CCNQ	99.98 %	300708	STAR syndrome, 300707 (3), X-linked dominant
CD151	100 %	602243	[Blood group, Raph], 179620 (3); Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057 (3), Autosomal recessive
CD2AP	99.69 %	604241	Glomerulosclerosis, focal segmental, 3, 607832 (3)
CD46	99.86 %	120920	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal dominant, Autosomal recessive
CDC5L	99.83 %	602868	*No OMIM phenotypes*
CDKN1C	100 %	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
CENPF	99.97 %	600236	Stromme syndrome, 243605 (3), Autosomal recessive
CEP104	99.99 %	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 77, 619988 (3), Autosomal recessive
CEP120	99.9 %	613446	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive
CEP164	99.99 %	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive

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CEP290	98.1 %	610142	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive
CEP41	99.99 %	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
CEP55	99.92 %	610000	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive
CEP83	98.68 %	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
CFAP47	99.21 %	301057	Spermatogenic failure, X-linked 3, 301059 (3), X-linked recessive
CFB	99.97 %	138470	?Complement factor B deficiency, 615561 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant
CFH	99.12 %	134370	{Macular degeneration, age-related, 4}, 610698 (3), Autosomal dominant; Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal dominant, Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal dominant, Autosomal recessive
CFHR1	84.44 %	134371	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal dominant, Autosomal recessive
CFHR2	90.26 %	600889	*No OMIM phenotypes*
CFHR3	91.62 %	605336	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal dominant, Autosomal recessive
CFHR4	99.86 %	605337	*No OMIM phenotypes*
CFHR5	99.68 %	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
CFI	99.87 %	217030	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant; Complement factor I deficiency, 610984 (3), Autosomal recessive
CHD1L	98.53 %	613039	*No OMIM phenotypes*
CHD7	99.99 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHRM3	100 %	118494	Prune belly syndrome, 100100 (3), Autosomal recessive
CHRNA3	99.95 %	118503	{Lung cancer susceptibility 2}, 612052 (3); Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800 (3), Autosomal recessive
CILK1	99.69 %	612325	{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant; Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive
CLCN5	99.67 %	300008	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Dent disease 1, 300009 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive
CLCN7	99.99 %	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant
CLCNKA	99.98 %	602024	Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive

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CLCNKB	99.98 %	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLDN10	99.97 %	617579	HELIX syndrome, 617671 (3), Autosomal recessive
CLDN16	99.98 %	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
CLDN19	99.02 %	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
CNNM2	99.94 %	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and impaired intellectual development 1, 616418 (3), Autosomal dominant, Autosomal recessive
COL4A1	99.99 %	120130	?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant
COL4A3	99.94 %	120070	Alport syndrome 3A, autosomal dominant, 104200 (3), Autosomal dominant; Hematuria, benign familial, 2, 620320 (3), Autosomal dominant; Alport syndrome 3B, autosomal recessive, 620536 (3)
COL4A4	99.95 %	120131	Hematuria, familial benign, 1, 141200 (3), Autosomal dominant; Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive
COL4A5	99.64 %	303630	Alport syndrome 1, X-linked, 301050 (3), X-linked dominant
COQ2	99.9 %	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
COQ6	99.94 %	614647	Coenzyme Q10 deficiency, primary, 6, 614650 (3), Autosomal recessive
COQ7	100 %	601683	Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 (3), Autosomal recessive
COQ8B	99.94 %	615567	Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive
COQ9	99.62 %	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
CPLANE1	99.81 %	614571	Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive
CRB2	99.95 %	609720	Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive
CSPP1	98.31 %	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CST3	100 %	604312	{Macular degeneration, age-related, 11}, 611953 (3); Cerebral amyloid angiopathy, 105150 (3), Autosomal dominant
CTNS	100 %	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
CTU2	99.91 %	617057	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 (3), Autosomal recessive
CUBN	99.99 %	602997	[Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive
CUL3	99.76 %	603136	Neurodevelopmental disorder with or without autism or seizures, 619239 (3), Autosomal dominant; Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant

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CYP11B1	100 %	610613	Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant; Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive
CYP11B2	100 %	124080	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive; Aldosterone to renin ratio raised (3); {Low renin hypertension, susceptibility to} (3); Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive
CYP17A1	100 %	609300	17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive; 17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive
CYP24A1	100 %	126065	Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive
DAAM2	99.99 %	606627	Nephrotic syndrome, type 24, 619263 (3), Autosomal recessive
DCDC2	99.96 %	605755	Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive
DDX59	99.67 %	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DGKE	99.1 %	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive
DHCR7	99.97 %	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DLC1	99.98 %	604258	Colorectal cancer, somatic, 114500 (3)
DLG5	99.91 %	604090	Yuksel-Vogel-Bauser syndrome, 620703 (3), Autosomal recessive
DMP1	99.99 %	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
DNAJB11	99.97 %	611341	Polycystic kidney disease 6 with or without polycystic liver disease, 618061 (3), Autosomal dominant
DSTYK	99.83 %	612666	Spastic paraplegia 23, autosomal recessive, 270750 (3), Autosomal recessive; Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant
DYNC2H1	99.66 %	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Digenic recessive, Autosomal recessive
DYNC2I1	99.99 %	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
DZIP1L	98.46 %	617570	Polycystic kidney disease 5, 617610 (3), Autosomal recessive
EGF	99.96 %	131530	?Hypomagnesemia 4, renal, 611718 (3), Autosomal recessive
EHHADH	99.99 %	607037	?Fanconi renotubular syndrome 3, 615605 (3), Autosomal dominant
EMP2	100 %	602334	Nephrotic syndrome, type 10, 615861 (3), Autosomal recessive
ENPP1	99.88 %	173335	{Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant
EVC	99.95 %	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	99.97 %	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EXOC3L2	99.88 %	616927	*No OMIM phenotypes*
EYA1	99.81 %	601653	Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant
FAH	99.98 %	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive

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FAM20A	100 %	611062	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3), Autosomal recessive
FAN1	99.73 %	613534	Interstitial nephritis, karyomegalic, 614817 (3), Autosomal recessive
FAT1	99.99 %	600976	*No OMIM phenotypes*
FGA	99.98 %	134820	Amyloidosis, hereditary systemic 2, 105200 (3), Autosomal dominant; Hypodysfibrinogenemia, congenital, 616004 (3); Dysfibrinogenemia, congenital, 616004 (3); Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
FGF20	99.71 %	605558	?Renal hypodysplasia/aplasia 2, 615721 (3), Autosomal recessive
FGF23	100 %	605380	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3), Autosomal recessive; Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant
FGFR1	100 %	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
FLCN	99.51 %	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)
FN1	99.95 %	135600	Spondylometaphyseal dysplasia, corner fracture type, 184255 (3), Autosomal dominant; Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant
FOXF1	99.99 %	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
FOXI1	100 %	601093	Enlarged vestibular aqueduct, 600791 (3), Autosomal recessive
FRAS1	99.97 %	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
FREM1	99.98 %	608944	Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Bifid nose with or without anorectal and renal anomalies, 608980 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant
FREM2	99.97 %	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive
FXYD2	100 %	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
G6PC1	99.93 %	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
GALNT3	99.52 %	601756	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive
GANAB	99.97 %	104160	Polycystic kidney disease 3, 600666 (3), Autosomal dominant
GATA3	99.96 %	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3), Autosomal dominant
GATM	99.92 %	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant
GDNF	99.99 %	600837	{Hirschsprung disease, susceptibility to, 3}, 613711 (3), Autosomal dominant
GFRA1	100 %	601496	Renal hypodysplasia/aplasia 4, 619887 (3), Autosomal recessive
GLA	99.9 %	300644	Fabry disease, cardiac variant, 301500 (3), X-linked; Fabry disease, 301500 (3), X-linked
GLI3	100 %	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GLIS2	100 %	608539	Nephronophthisis 7, 611498 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GNA11	99.99 %	139313	Hypocalciuric hypercalcemia, type II, 145981 (3), Autosomal dominant; Hypocalcemia, autosomal dominant 2, 615361 (3), Autosomal dominant
GNAS	100 %	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
GON7	99.92 %	617436	Galloway-Mowat syndrome 9, 619603 (3), Autosomal recessive
GPC3	99.6 %	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
GREB1L	99.99 %	617782	Deafness, autosomal dominant 80, 619274 (3), Autosomal dominant; Renal hypodysplasia/aplasia 3, 617805 (3), Autosomal dominant
GRHPR	99.93 %	604296	Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive
GRIP1	99.83 %	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
GSN	99.93 %	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
HAAO	99.96 %	604521	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 (3), Autosomal recessive
HNF1B	100 %	189907	Type 2 diabetes mellitus, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3)
HNF4A	100 %	600281	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant
HOGA1	100 %	613597	Hyperoxaluria, primary, type III, 613616 (3), Autosomal recessive
HOXA13	99.94 %	142959	Hand-foot-genital syndrome, 140000 (3), Autosomal dominant; ?Guttmacher syndrome, 176305 (3), Autosomal dominant
HPRT1	97.8 %	308000	Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
HPSE2	100 %	613469	Urofacial syndrome 1, 236730 (3), Autosomal recessive
HS2ST1	95.79 %	604844	Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive
HSD11B2	99.99 %	614232	Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive
HYLS1	100 %	610693	Hydrolethalus syndrome, 236680 (3), Autosomal recessive
IFT122	99.98 %	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT140	100 %	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive
IFT172	99.98 %	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT27	100 %	615870	Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
IFT43	99.97 %	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
IFT80	99.69 %	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
IFT81	94.64 %	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
INF2	99.99 %	610982	Glomerulosclerosis, focal segmental, 5, 613237 (3); Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3), Autosomal dominant
INPP5E	99.85 %	613037	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
INVS	99.94 %	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IQCB1	99.72 %	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
ITGA3	99.86 %	605025	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748 (3), Autosomal recessive
ITGA8	99.95 %	604063	Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive
ITGB4	99.99 %	147557	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 (3), Autosomal recessive; Epidermolysis bullosa, junctional 5A, intermediate, 619816 (3), Autosomal recessive
ITSN1	99.89 %	602442	*No OMIM phenotypes*
JAG1	100 %	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
KANK1	99.99 %	607704	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
KANK2	99.99 %	614610	Nephrotic syndrome, type 16, 617783 (3), Autosomal recessive; Palmoplantar keratoderma and woolly hair, 616099 (3), Autosomal recessive
KANK4	99.7 %	614612	*No OMIM phenotypes*
KCNJ1	100 %	600359	Bartter syndrome, type 2, 241200 (3), Autosomal recessive
KCNJ10	99.98 %	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
KCNJ16	100 %	605722	Hypokalemic tubulopathy and deafness, 619406 (3), Autosomal recessive
KCNJ5	99.99 %	600734	Long QT syndrome 13, 613485 (3), Autosomal dominant; Hyperaldosteronism, familial, type III, 613677 (3), Autosomal dominant
KDM6A	99.74 %	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KIAA0586	95.75 %	610178	Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive
KIAA0753	100 %	617112	?Orofaciodigital syndrome XV, 617127 (3), Autosomal recessive; ?Joubert syndrome 38, 619476 (3), Autosomal recessive; Short-rib thoracic dysplasia 21 without polydactyly, 619479 (3), Autosomal recessive
KIF14	97.8 %	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive
KIF7	100 %	611254	Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
KIRREL1	99.82 %	607428	Nephrotic syndrome, type 23, 619201 (3), Autosomal recessive
KL	99.98 %	604824	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994 (3), Autosomal recessive
KLHL3	99.97 %	605775	Pseudohypoaldosteronism, type IID, 614495 (3), Autosomal dominant, Autosomal recessive
KMT2D	99.98 %	602113	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 (3), Autosomal dominant; Kabuki syndrome 1, 147920 (3), Autosomal dominant
KYNU	99.19 %	605197	?Hydroxykynureninuria, 236800 (3), Autosomal recessive; Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LAGE3	99.99 %	300060	Galloway-Mowat syndrome 2, X-linked, 301006 (3), X-linked recessive
LAMA5	99.99 %	601033	Nephrotic syndrome, type 26, 620049 (3), Autosomal recessive; ?Bent bone dysplasia syndrome 2, 620076 (3), Autosomal recessive
LAMB2	99.99 %	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3), Autosomal recessive; Pierson syndrome, 609049 (3), Autosomal recessive
LCAT	99.97 %	606967	Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive
LIFR	99.69 %	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive
LMNA	99.96 %	150330	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant
LMX1B	100 %	602575	Focal segmental glomerulosclerosis 10, 256020 (3), Autosomal dominant; Nail-patella syndrome, 161200 (3), Autosomal dominant
LRIG2	97.97 %	608869	Urofacial syndrome 2, 615112 (3), Autosomal recessive
LRP2	99.86 %	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRP4	99.89 %	604270	?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal dominant, Autosomal recessive; Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive
LRP5	99.95 %	603506	Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Endosteal hyperostosis, 144750 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Exudative vitreoretinopathy 4, 601813 (3), Autosomal dominant, Autosomal recessive
LYZ	99.86 %	153450	Amyloidosis, hereditary systemic 5, 620658 (3)
LZTFL1	100 %	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
MAGED2	99.98 %	300470	Bartter syndrome, type 5, antenatal, transient, 300971 (3), X-linked recessive
MAGI2	99.89 %	606382	Nephrotic syndrome, type 15, 617609 (3), Autosomal recessive
MAPKBP1	99.98 %	616786	Nephronophthisis 20, 617271 (3), Autosomal recessive
MKKS	100 %	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
MKS1	99.92 %	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
MMACHC	99.98 %	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MNX1	99.83 %	142994	Currarino syndrome, 176450 (3), Autosomal dominant
MOCOS	99.98 %	613274	Xanthinuria, type II, 603592 (3), Autosomal recessive
MTX2	99.2 %	608555	Mandibuloacral dysplasia progeroid syndrome, 619127 (3), Autosomal recessive
MUC1	99.98 %	158340	Tubulointerstitial kidney disease, autosomal dominant, 2, 174000 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MYH9	99.95 %	160775	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 (3), Autosomal dominant; Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant
MYO1E	99.94 %	601479	Glomerulosclerosis, focal segmental, 6, 614131 (3), Autosomal recessive
MYOCD	99.99 %	606127	Megabladder, congenital, 618719 (3), Autosomal dominant
NADSYN1	99.79 %	608285	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845 (3), Autosomal recessive
NEK1	99.83 %	604588	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Digenic recessive, Autosomal recessive; ?Orofaciodigital syndrome II, 252100 (3), Autosomal recessive; {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant
NEK8	99.99 %	609799	Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; Polycystic kidney disease 8, 620903 (3); ?Nephronophthisis 9, 613824 (3)
NIPBL	99.34 %	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NLRP3	100 %	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NOS1AP	99.91 %	605551	Nephrotic syndrome, type 22, 619155 (3), Autosomal recessive
NOTCH2	99.03 %	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
NPHP1	99.05 %	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPHP3	99.89 %	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
NPHP4	99.98 %	607215	Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive
NPHS1	99.97 %	602716	Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive
NPHS2	99.87 %	604766	Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive
NR3C1	99.93 %	138040	Glucocorticoid resistance, 615962 (3), Autosomal dominant
NR3C2	100 %	600983	Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3), Autosomal dominant; Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3)
NUP107	97.46 %	607617	?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
NUP133	99.45 %	607613	?Galloway-Mowat syndrome 8, 618349 (3), Autosomal recessive; Nephrotic syndrome, type 18, 618177 (3), Autosomal recessive
NUP205	99.92 %	614352	?Nephrotic syndrome, type 13, 616893 (3), Autosomal recessive
NUP85	99.98 %	170285	Nephrotic syndrome, type 17, 618176 (3), Autosomal recessive
NUP93	99.87 %	614351	Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive
OCRL	99.89 %	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
OFD1	99.68 %	300170	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
OSGEP	100 %	610107	Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive
PAX2	99.99 %	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
PBX1	99.88 %	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
PCBD1	99.84 %	126090	Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive
PDE3A	99.97 %	123805	Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant
PDE6D	99.94 %	602676	Joubert syndrome 22, 615665 (3), Autosomal recessive
PDIA6	99.75 %	611099	*No OMIM phenotypes*
PDSS1	95.7 %	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
PDSS2	99.87 %	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
PHEX	99.83 %	300550	Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant
PKD1	99.98 %	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
PKD2	99.91 %	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
PKHD1	99.95 %	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
PLCE1	99.98 %	608414	Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive
PLG	99.89 %	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PLVAP	99.97 %	607647	Diarrhea 10, protein-losing enteropathy type, 618183 (3), Autosomal recessive
PMM2	99.93 %	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PODXL	93.29 %	602632	*No OMIM phenotypes*
PRDM15	99.99 %	617692	*No OMIM phenotypes*
PRKCSH	99.99 %	177060	Polycystic liver disease 1, 174050 (3), Autosomal dominant
PTPRO	99.91 %	600579	Nephrotic syndrome, type 6, 614196 (3), Autosomal recessive
RCAN1	99.99 %	602917	*No OMIM phenotypes*
REN	99.85 %	179820	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; [Hyperproreninemia] (3); Tubulointerstitial kidney disease, autosomal dominant, 4, 613092 (3), Autosomal dominant
RET	99.97 %	164761	{Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant
RMND1	99.92 %	614917	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive
ROBO1	99.83 %	602430	Pituitary hormone deficiency, combined or isolated, 8, 620303 (3), Autosomal dominant; Neurooculorenal syndrome, 620305 (3), Autosomal recessive; ?Nystagmus 8, congenital, autosomal recessive, 257400 (3), Autosomal recessive
ROBO2	99.8 %	602431	Vesicoureteral reflux 2, 610878 (3), Autosomal dominant
ROR2	99.99 %	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
RPGRIP1	99.95 %	605446	Cone-rod dystrophy 13, 608194 (3), Autosomal recessive; Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
RPGRIP1L	96.35 %	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RRAGD	99.93 %	608268	Hypomagnesemia 7, renal, with or without dilated cardiomyopathy, 620152 (3), Autosomal dominant
RRM2B	99.97 %	604712	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
SALL1	100 %	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant
SALL4	100 %	607343	?IVIC syndrome, 147750 (3), Autosomal dominant; Duane-radial ray syndrome, 607323 (3), Autosomal dominant
SARS2	99.99 %	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive
SCARB2	99.99 %	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
SCLT1	95.17 %	611399	*No OMIM phenotypes*
SCN4A	99.98 %	603967	Paramyotonia congenita, 168300 (3), Autosomal dominant; Hyperkalemic periodic paralysis, 170500 (3), Autosomal dominant; Congenital myopathy 22B, severe fetal, 620369 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Congenital myopathy 22A, classic, 620351 (3), Autosomal recessive
SCNN1A	100 %	600228	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350 (3), Autosomal recessive; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant
SCNN1B	99.38 %	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125 (3), Autosomal recessive; Liddle syndrome 1, 177200 (3), Autosomal dominant
SCNN1G	99.94 %	600761	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126 (3), Autosomal recessive; Liddle syndrome 2, 618114 (3), Autosomal dominant
SDCCAG8	100 %	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
SEC61A1	99.99 %	609213	Immunodeficiency, common variable, 15, 620670 (3), Autosomal dominant; ?Neutropenia, severe congenital, 11, autosomal dominant, 620674 (3), Autosomal dominant; Tubulointerstitial kidney disease, autosomal dominant, 5, 617056 (3), Autosomal dominant
SEC63	99.84 %	608648	Polycystic liver disease 2, 617004 (3), Autosomal dominant
SGPL1	99.95 %	603729	RENI syndrome, 617575 (3), Autosomal recessive
SIX1	100 %	601205	Deafness, autosomal dominant 23, 605192 (3), Autosomal dominant; Branchiootic syndrome 3, 608389 (3), Autosomal dominant
SIX5	100 %	600963	Branchiootorenal syndrome 2, 610896 (3)
SLC12A1	99.9 %	600839	Bartter syndrome, type 1, 601678 (3), Autosomal recessive
SLC12A3	99.84 %	600968	Gitelman syndrome, 263800 (3), Autosomal recessive
SLC22A12	99.99 %	607096	Hypouricemia, renal, 220150 (3), Autosomal recessive
SLC26A1	100 %	610130	?Hypersulfaturia, 620372 (3), Autosomal recessive; ?Nephrolithiasis, calcium oxalate, 1, 167030 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC26A3	99.97 %	126650	Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive
SLC2A2	99.96 %	138160	Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
SLC2A9	99.98 %	606142	{Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal dominant, Autosomal recessive; Hypouricemia, renal, 2, 612076 (3), Autosomal dominant, Autosomal recessive
SLC34A1	99.99 %	182309	?Fanconi renotubular syndrome 2, 613388 (3), Autosomal recessive; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant
SLC34A3	100 %	609826	Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive
SLC37A4	99.9 %	602671	Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Congenital disorder of glycosylation, type IIw, 619525 (3), Autosomal dominant; Glycogen storage disease Ic, 232240 (3), Autosomal recessive
SLC3A1	99.98 %	104614	Cystinuria, 220100 (3), Autosomal dominant, Autosomal recessive
SLC41A1	99.98 %	610801	?Nephronophthisis-like nephropathy 2, 619468 (3), Autosomal recessive
SLC4A1	99.94 %	109270	[Blood group, Swann], 601550 (3); [Blood group, Wright], 112050 (3); Distal renal tubular acidosis 1, 179800 (3), Autosomal dominant; [Blood group, Waldner], 112010 (3); Spherocytosis, type 4, 612653 (3), Autosomal dominant; [Blood group, Froese], 601551 (3); Distal renal tubular acidosis 4 with hemolytic anemia, 611590 (3), Autosomal recessive; {Malaria, resistance to}, 611162 (3); Cryohydrocytosis, 185020 (3), Autosomal dominant; Ovalocytosis, SA type, 166900 (3), Autosomal dominant; [Blood group, Diego], 110500 (3)
SLC4A4	99.97 %	603345	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278 (3), Autosomal recessive
SLC5A2	99.99 %	182381	Renal glucosuria, 233100 (3), Autosomal dominant, Autosomal recessive
SLC7A9	99.97 %	604144	Cystinuria, 220100 (3), Autosomal dominant, Autosomal recessive
SLC9A3R1	100 %	604990	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287 (3), Autosomal dominant
SLIT2	99.9 %	603746	*No OMIM phenotypes*
SMARCAL1	99.97 %	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
SOX17	100 %	610928	Vesicoureteral reflux 3, 613674 (3), Autosomal dominant
SRGAP1	99.68 %	606523	{Thyroid cancer, nonmedullary, 2}, 188470 (3), Somatic mutation, Autosomal dominant
STRA6	99.95 %	610745	Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive
STRADA	99.98 %	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive
STX16	100 %	603666	Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant
TBC1D1	99.96 %	609850	*No OMIM phenotypes*
TBC1D8B	99.31 %	301027	Nephrotic syndrome, type 20, 301028 (3), X-linked
TBX18	99.5 %	604613	Congenital anomalies of kidney and urinary tract 2, 143400 (3), Autosomal dominant
TCTN1	99.92 %	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
TCTN2	99.99 %	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	99.92 %	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
TFAP2A	100 %	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
THBD	100 %	188040	Thrombophilia 12 due to thrombomodulin defect, 614486 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant
TMEM107	100 %	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
TMEM138	100 %	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
TMEM216	99.98 %	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM231	88.88 %	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	99.3 %	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TMEM260	99.9 %	617449	Structural heart defects and renal anomalies syndrome, 617478 (3), Autosomal recessive
TMEM67	99.69 %	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
TNS2	99.98 %	607717	*No OMIM phenotypes*
TP53RK	100 %	608679	Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive
TPRKB	81.09 %	608680	Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive
TRAF3IP1	99.96 %	607380	Senior-Loken syndrome 9, 616629 (3), Autosomal recessive
TRAP1	100 %	606219	*No OMIM phenotypes*
TRIM8	99.82 %	606125	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428 (3), Autosomal dominant
TRPC6	99.99 %	603652	Glomerulosclerosis, focal segmental, 2, 603965 (3), Autosomal dominant
TRPM6	99.93 %	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
TSC1	99.99 %	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3)
TSC2	99.98 %	191092	Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
TTC21B	99.5 %	612014	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal dominant, Autosomal recessive
TTC8	99.67 %	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TTR	100 %	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
TUBB4B	100 %	602660	Leber congenital amaurosis with early-onset deafness, 617879 (3), Autosomal dominant
TULP3	99.91 %	604730	Hepatorenocardiac degenerative fibrosis, 619902 (3), Autosomal recessive
TXNDC15	99.78 %	617778	Meckel syndrome 14, 619879 (3), Autosomal recessive
UMOD	99.98 %	191845	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000 (3), Autosomal dominant
UPK3A	100 %	611559	*No OMIM phenotypes*
VDR	99.86 %	601769	Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
VHL	100 %	608537	Hemangioblastoma, cerebellar, somatic (3); Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
VIPAS39	99.87 %	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
VPS33B	99.95 %	608552	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 12, 620010 (3), Autosomal recessive; Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
WBP11	99.88 %	618083	Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227 (3), Autosomal dominant
WDPCP	99.87 %	613580	Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
WDR19	99.8 %	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive
WDR35	99.92 %	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
WDR4	99.95 %	605924	Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive
WDR72	96.51 %	613214	Amelogenesis imperfecta, type IIA3, 613211 (3), Autosomal recessive
WDR73	99.92 %	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
WNK1	99.98 %	605232	Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive; Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant
WNK4	99.98 %	601844	Pseudohypoaldosteronism, type IIB, 614491 (3), Autosomal dominant
WNT4	99.94 %	603490	?SERKAL syndrome, 611812 (3), Autosomal recessive; Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant
WNT5A	100 %	164975	Robinow syndrome, autosomal dominant 1, 180700 (3), Autosomal dominant
WT1	99.99 %	607102	Mesothelioma, somatic, 156240 (3); Meacham syndrome, 608978 (3), Autosomal dominant; Frasier syndrome, 136680 (3), Somatic mutation, Autosomal dominant; Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Denys-Drash syndrome, 194080 (3), Somatic mutation, Autosomal dominant; Wilms tumor, type 1, 194070 (3), Somatic mutation, Autosomal dominant
XDH	99.97 %	607633	Xanthinuria, type I, 278300 (3), Autosomal recessive
XPNPEP3	99.99 %	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
XPO5	99.89 %	607845	*No OMIM phenotypes*
YRDC	99.85 %	612276	Galloway-Mowat syndrome 10, 619609 (3), Autosomal recessive
ZIC3	99.9 %	300265	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive
ZMPSTE24	98.7 %	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy 1, 275210 (3), Autosomal recessive
ZMYM2	99.88 %	602221	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522 (3), Autosomal dominant
ZNF423	98.94 %	604557	Nephronophthisis 14, 614844 (3), Autosomal dominant, Autosomal recessive; Joubert syndrome 19, 614844 (3), Autosomal dominant, Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.