RENAL DISORDERS GENE PANEL DG 2.16 (282 genes)

Releasedate: 07-06-2019

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM disease ID
ACE	120,1	99.9%	99.5%	Renal tubular dysgenesis, 267430 [Angiotensin I-converting enzyme, benign serum increase], 0 {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to}, 0 {SARS, progression of}, 0 {Stroke, hemorrhagic}, 614519
ACTN4	130,5	100.0%	99.9%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	103,8	98.1%	95.2%	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS9	115,7	99.5%	97.4%	No OMIM phenotype
ADCY10	126,3	100.0%	99.8%	{Hypercalciuria, absorptive, susceptibility to}, 143870
AGT	192,1	100.0%	100.0%	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}, 0
AGTR1	143,6	91.9%	91.6%	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	160,8	100.0%	100.0%	Hyperoxaluria, primary, type 1, 259900
AHI1	129,8	99.7%	98.3%	Joubert syndrome 3, 608629
ALDOB	135,3	100.0%	99.3%	Fructose intolerance, hereditary, 229600
ALG8	118,5	96.6%	96.2%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALMS1	172,8	100.0%	99.7%	Alstrom syndrome, 203800
AMN	101,5	98.1%	90.6%	Megaloblastic anemia-1, Norwegian type, 261100
ANKS6	94,2	98.3%	94.4%	Nephronophthisis 16, 615382
ANLN	140,7	98.7%	97.7%	Focal segmental glomerulosclerosis 8, 616032
ANOS1	76,7	91.7%	88.0%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AP2S1	110,7	90.4%	89.8%	Hypocalciuric hypercalcemia, type III, 600740
APOL1	146,1	100.0%	100.0%	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551

APRT	93,2	100.0%	100.0%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	126,1	100.0%	99.9%	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	202,4	100.0%	100.0%	Nephrotic syndrome, type 8, 615244
ARL13B	102,2	100.0%	99.4%	Joubert syndrome 8, 612291
ARL6	100,3	99.8%	98.2%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ATP6V0A4	105,4	100.0%	99.2%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	172,2	100.0%	100.0%	Renal tubular acidosis with deafness, 267300
ATP7B	128,7	99.9%	99.1%	Wilson disease, 277900
AVP	65,2	98.2%	83.8%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	133,1	100.0%	99.8%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	103,7	92.2%	92.1%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	105,8	100.0%	100.0%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BBIP1	119,7	97.3%	90.3%	?Bardet-Biedl syndrome 18, 615995
BBS1	146,4	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	158,1	100.0%	99.9%	Bardet-Biedl syndrome 10, 615987
BBS12	187,1	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	150,7	99.9%	99.6%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	110,2	99.9%	99.2%	Bardet-Biedl syndrome 4, 615982
BBS5	98,5	98.0%	93.3%	Bardet-Biedl syndrome 5, 615983
BBS7	142,9	99.1%	96.5%	Bardet-Biedl syndrome 7, 615984
BBS9	113,6	98.6%	94.4%	Bardet-Biedl syndrome 9, 615986
BCS1L	147,9	100.0%	100.0%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BICC1	139,2	100.0%	99.9%	{Renal dysplasia, cystic, susceptibility to}, 601331
BSND	139,4	100.0%	99.9%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
C3	141,6	100.0%	99.4%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925

				{Macular degeneration, age-related, 9}, 611378
C5orf42	NC	NC	NC	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	137,4	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CACNA1H	139,3	99.4%	98.1%	Hyperaldosteronism, familial, type IV, 617027 {Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CASR	154,3	100.0%	99.7%	Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CC2D2A	111,7	99.0%	97.1%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CD2AP	121,7	99.7%	98.3%	Glomerulosclerosis, focal segmental, 3, 607832
CD46	125,5	99.7%	98.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CEP120	131,7	100.0%	99.4%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	89,3	99.8%	98.0%	Nephronophthisis 15, 614845
CEP290	82,6	97.3%	91.7%	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	78,2	98.9%	94.4%	Joubert syndrome 15, 614464
CEP55	124,5	100.0%	100.0%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	108,8	99.4%	96.6%	Nephronophthisis 18, 615862
CFB	119,4	100.0%	99.9%	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	155,4	99.4%	97.9%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	158,4	93.6%	90.8%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075

CFHR3	98,4	93.8%	91.6%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFI	139	99.5%	97.0%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CLCN5	104,3	99.7%	96.5%	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCNKB	99,4	99.7%	97.1%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN10	138,2	100.0%	100.0%	HELIX syndrome, 617671
CLDN16	126,6	100.0%	100.0%	Hypomagnesemia 3, renal, 248250
CLDN19	125,4	99.1%	95.1%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	199,8	100.0%	100.0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
COL4A3	90,5	99.6%	97.7%	Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200 Hematuria, benign familial, 141200
COL4A4	92,4	99.8%	97.4%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	56,8	96.8%	85.7%	Alport syndrome 1, X-linked, 301050
COQ2	103,5	97.6%	97.1%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ6	127,5	99.9%	98.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	138,3	99.9%	99.6%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8B	99,5	100.0%	99.8%	Nephrotic syndrome, type 9, 615573
COQ9	73,8	100.0%	98.1%	Coenzyme Q10 deficiency, primary, 5, 614654
CRB2	116	99.8%	98.5%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CSPP1	119	100.0%	99.1%	Joubert syndrome 21, 615636
CTNS	112,6	100.0%	99.5%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CUBN	103,2	99.6%	97.6%	Megaloblastic anemia-1, Finnish type, 261100

CUL3	119	99.9%	98.9%	Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	169,1	100.0%	100.0%	Hypercalcemia, infantile, 1, 143880
DCDC2	150	99.9%	99.8%	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DGKE	127,8	99.8%	98.3%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DMP1	133	100.0%	99.9%	Hypophosphatemic rickets, AR, 241520
DNAJB11	107,4	99.8%	99.5%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DSTYK	121,7	99.6%	97.7%	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DYNC2H1	102,2	98.8%	95.5%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DZIP1L	96,7	99.7%	98.0%	Polycystic kidney disease 5, 617610
EGF	110,5	100.0%	99.7%	Hypomagnesemia 4, renal, 611718
EHHADH	133,4	100.0%	99.7%	?Fanconi renotubular syndrome 3, 615605
EMP2	78,4	99.5%	96.1%	Nephrotic syndrome, type 10, 615861
ENPP1	129,2	97.5%	93.3%	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665
EYA1	120,2	99.9%	99.8%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
FAH	128,4	100.0%	99.8%	Tyrosinemia, type I, 276700
FAM20A	111,1	100.0%	99.4%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM58A	NC	NC	NC	STAR syndrome, 300707
FAN1	132,2	100.0%	99.9%	Interstitial nephritis, karyomegalic, 614817
FAT1	157,1	100.0%	99.9%	No OMIM phenotype Nephrotic syndrome, tubular ectasia and haematuria (Gee (2016) Nat Commun 7,10822) Facioscapulohumeral dystrophy-like phenotype (Puppo (2015) Hum Mutat 36,443) ?Congenital anomalies of the kidney and urinary tract (Nicolaou (201
FGF23	122,3	99.7%	97.7%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993

FN1	106	99.9%	98.9%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 Spondylometaphyseal dysplasia, corner fracture type, 184255
FOXC2	122,3	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXI1	194,5	100.0%	100.0%	Enlarged vestibular aqueduct, 600791
FRAS1	119,2	99.9%	99.2%	Fraser syndrome 1, 219000
FREM1	110,7	99.8%	98.4%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	152,4	100.0%	99.5%	Cryptophthalmos, unilateral or bilateral, isolated, 123570 Fraser syndrome 2, 617666
FXYD2	108,1	100.0%	100.0%	Hypomagnesemia 2, renal, 154020
G6PC	146,8	100.0%	99.9%	Glycogen storage disease Ia, 232200
GALNT3	125,8	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GANAB	107,1	99.9%	98.3%	Polycystic kidney disease 3, 600666
GATA3	220,5	100.0%	100.0%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GCM2	135,1	100.0%	100.0%	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
GLA	73,6	99.5%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLIS2	129,6	100.0%	100.0%	Nephronophthisis 7, 611498
GLIS3	123,7	100.0%	99.5%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	162,4	100.0%	99.5%	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GREB1L	128	99.9%	99.0%	Renal hypodysplasia/aplasia 3, 617805
GRHPR	99,6	84.2%	81.7%	Hyperoxaluria, primary, type II, 260000
GRIP1	111,1	100.0%	99.3%	Fraser syndrome 3, 617667
GSN	115,5	95.6%	93.5%	Amyloidosis, Finnish type, 105120
HNF1B	118,8	99.6%	96.8%	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	129,8	99.9%	99.0%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HOGA1	149,6	100.0%	99.1%	Hyperoxaluria, primary, type III, 613616

HPRT1	59,8	98.3%	88.2%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	165,6	94.3%	87.3%	Apparent mineralocorticoid excess, 218030
IFNG	146,9	100.0%	100.0%	{AIDS, rapid progression to}, 609423 {Aplastic anemia}, 609135 {Hepatitis C virus, response to therapy of}, 609532 {TSC2 angiomyolipomas, renal, modifier of}, 613254 {Tuberculosis, protection against}, 607948
IFT122	120,5	99.9%	99.0%	Cranioectodermal dysplasia 1, 218330
IFT140	117,6	99.9%	99.2%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	94,5	100.0%	99.4%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	115,8	100.0%	100.0%	?Bardet-Biedl syndrome 19, 615996
IFT43	112,4	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
INF2	99,5	85.6%	83.5%	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
INPP5E	116,8	100.0%	98.6%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INTU	115,4	99.8%	98.7%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	143,7	100.0%	99.9%	Nephronophthisis 2, infantile, 602088
IQCB1	93,3	91.6%	80.0%	Senior-Loken syndrome 5, 609254
ITGA3	150,1	99.7%	98.0%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA8	115,5	100.0%	99.5%	Renal hypodysplasia/aplasia 1, 191830
JAG1	133,7	99.2%	97.1%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KANK2	163,4	100.0%	99.9%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KCNJ1	157,5	100.0%	100.0%	Bartter syndrome, type 2, 241200
KCNJ10	148,6	89.2%	88.1%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ5	160,1	100.0%	99.8%	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485

KIAA0556	126,6	100.0%	99.6%	Joubert syndrome 26, 616784
KIF14	116,6	99.6%	97.9%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF7	105,2	98.2%	93.5%	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KL	171,8	99.2%	98.1%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLHL3	105,6	99.9%	97.7%	Pseudohypoaldosteronism, type IID, 614495
LAGE3	68,3	98.1%	90.5%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMB2	166,5	100.0%	99.6%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCAT	140,7	99.4%	95.1%	Fish-eye disease, 136120 Norum disease, 245900
LMX1B	146,6	99.9%	98.5%	Nail-patella syndrome, 161200
LRIG2	126,8	99.8%	99.1%	Urofacial syndrome 2, 615112
LRP2	139,2	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRP4	128	99.7%	99.0%	?Myasthenic syndrome, congenital, 17, 616304 Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	168,1	99.8%	98.7%	Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteopetrosis, autosomal dominant 1, 607634 Osteoporosis-pseudoglioma syndrome, 259770 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 van Buchem disease, type 2, 607636 [Bone mineral density variability 1], 601884 {Osteoporosis}, 166710
LYZ	143	100.0%	100.0%	Amyloidosis, renal, 105200
LZTFL1	117	99.8%	99.2%	Bardet-Biedl syndrome 17, 615994
MAFB	124	100.0%	99.9%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	86,7	99.5%	97.6%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGI2	90,4	94.6%	91.7%	Nephrotic syndrome, type 15, 617609
MAPKBP1	132,5	100.0%	100.0%	Nephronophthisis 20, 617271

MKKS	155,7	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	92,4	99.6%	97.8%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MOCOS	147,2	99.9%	99.1%	Xanthinuria, type II, 603592
MYH9	128,5	99.6%	98.5%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO1E	115,4	99.8%	98.7%	Glomerulosclerosis, focal segmental, 6, 614131
NCAPG2	121,5	99.8%	98.2%	No OMIM phenotype
NEK1	115,9	99.7%	98.1%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK8	141,3	100.0%	99.9%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NOTCH2	123,7	100.0%	99.6%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP1	121,2	99.8%	98.5%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	121,4	99.8%	98.5%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	125,6	100.0%	99.7%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	106,3	100.0%	99.5%	Nephrotic syndrome, type 1, 256300
NPHS2	114,5	100.0%	99.5%	Nephrotic syndrome, type 2, 600995
NR3C2	123,4	100.0%	98.1%	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NUP107	126,6	99.8%	98.6%	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348 Nephrotic syndrome, type 11, 616730
NUP133	121,1	99.7%	97.8%	?Galloway-Mowat syndrome 8, 618349 Nephrotic syndrome, type 18, 618177
NUP160	137,7	100.0%	99.9%	?Nephrotic syndrome, type 19, 618178
NUP205	133,5	99.6%	98.7%	?Nephrotic syndrome, type 13, 616893
NUP85	125,7	100.0%	100.0%	Nephrotic syndrome, type 17, 618176
NUP93	117,8	96.9%	93.8%	Nephrotic syndrome, type 12, 616892

OCRL	106,2	99.8%	98.3%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	51,9	85.8%	70.8%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OSGEP	98,1	100.0%	97.3%	Galloway-Mowat syndrome 3, 617729
PAX2	184,1	100.0%	100.0%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PBX1	111,7	99.9%	98.2%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCBD1	103,9	100.0%	99.7%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDE6D	114,7	100.0%	99.9%	?Joubert syndrome 22, 615665
PDSS2	112,9	99.6%	96.1%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	107,9	99.8%	98.6%	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	35,9	43.0%	35.0%	Polycystic kidney disease 1, 173900
PKD2	102,3	98.7%	95.8%	Polycystic kidney disease 2, 613095
PKHD1	130,4	99.9%	99.4%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PLCE1	125	99.8%	99.0%	Nephrotic syndrome, type 3, 610725
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PTH1R	106,6	100.0%	99.1%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPRO	128,1	99.9%	99.4%	Nephrotic syndrome, type 6, 614196
REN	127,5	100.0%	100.0%	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia], 0
RMND1	132,6	100.0%	99.0%	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	127,4	98.8%	97.5%	Vesicoureteral reflux 2, 610878
RPGRIP1L	123,4	96.7%	95.4%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RRM2B	143,9	99.9%	99.4%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077

SALL1	113,3	99.9%	98.9%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480
SALL4	135	99.9%	98.1%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SARS2	117,9	95.1%	93.2%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	105,8	99.8%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	127	99.8%	98.3%	?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	130,6	100.0%	100.0%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome 1, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	142,2	99.5%	97.2%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350
SDCCAG8	124,1	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEC61A1	121	100.0%	99.9%	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SGPL1	132,3	100.0%	100.0%	Nephrotic syndrome, type 14, 617575
SIX5	76,3	99.9%	97.3%	Branchiootorenal syndrome 2, 610896
SLC12A1	144,2	100.0%	99.8%	Bartter syndrome, type 1, 601678
SLC12A3	140	100.0%	100.0%	Gitelman syndrome, 263800
SLC16A12	128,5	100.0%	99.9%	Cataract 47, juvenile, with microcornea, 612018
SLC22A12	117,7	100.0%	99.8%	Hypouricemia, renal, 220150
SLC26A1	149,3	100.0%	100.0%	?Nephrolithiasis, calcium oxalate, 167030
SLC26A3	132,5	99.9%	99.2%	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	158,1	100.0%	99.9%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	104,8	100.0%	98.7%	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC34A1	149,8	100.0%	99.9%	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	141,1	99.9%	99.0%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC36A2	100,3	100.0%	99.9%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600

SLC37A4	114,3	100.0%	99.6%	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC3A1	144,5	100.0%	99.4%	Cystinuria, 220100
SLC41A1	140,2	100.0%	99.9%	No OMIM phenotype Nephrolithiasis-like phenotype (Hurd (2013) J Am Soc Nephrol 24,967) ?Parkinson disease (Yan (2011) Int J Neurosci 121,632)
SLC4A1	139,2	100.0%	99.8%	Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 Spherocytosis, type 4, 612653 [Blood group, Diego], 110500 [Blood group, Froese], 601551 [Blood group, Swann], 601550 [Blood group, Waldner], 112010 [Blood group, Wright], 112050 [Malaria, resistance to], 611162
SLC4A4	113,9	99.8%	98.3%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	135	100.0%	100.0%	Renal glucosuria, 233100
SLC6A19	129,3	100.0%	100.0%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A20	151,9	100.0%	99.8%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC7A7	105,5	100.0%	99.6%	Lysinuric protein intolerance, 222700
SLC7A9	119,8	100.0%	98.8%	Cystinuria, 220100
SLC9A3	161,7	100.0%	99.8%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	142,8	100.0%	100.0%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLIT3	122,7	99.2%	96.7%	No OMIM phenotype Major depressive disorder (Glessner (2010) PLoS One 5, e15463) ?Autism spectrum disorder (Bi (2012) Hum Mutat 33, 1635) ?Glioma and Hodgkin lymphoma (Ritter (2015) Genet Med 17, 831) ?Schizophrenia (Gulsuner (2013) Cell 154,
SMARCAL1	113,2	100.0%	99.6%	Schimke immunoosseous dysplasia, 242900
SOX17	124,8	100.0%	100.0%	Vesicoureteral reflux 3, 613674
STRA6	117,6	100.0%	99.8%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186

STX16	111	99.9%	98.5%	Pseudohypoparathyroidism, type IB, 603233
TBX18	103,3	99.6%	97.6%	Congenital anomalies of kidney and urinary tract 2, 143400
TCTN1	94,8	95.6%	92.3%	Joubert syndrome 13, 614173
TCTN2	122,4	99.9%	99.0%	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	116,3	100.0%	99.9%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
THBD	181,1	100.0%	100.0%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TMEM107	148,7	100.0%	100.0%	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM138	82,7	100.0%	99.2%	Joubert syndrome 16, 614465
TMEM216	88	99.7%	95.7%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	101,1	100.0%	99.3%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	117,7	100.0%	99.2%	Joubert syndrome 14, 614424
TMEM260	117,5	99.6%	97.6%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	83,1	99.1%	94.6%	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TNXB	105,6	99.5%	95.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP53RK	81,3	99.7%	96.1%	Galloway-Mowat syndrome 4, 617730
TPRKB	59,4	80.3%	73.9%	Galloway-Mowat syndrome 5, 617731
TRAF3IP1	84,2	99.4%	97.1%	Senior-Loken syndrome 9, 616629
TRIM32	123	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRPC6	91,4	98.0%	96.0%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	126,6	99.9%	99.1%	Hypomagnesemia 1, intestinal, 602014
TSC1	112,5	99.6%	98.2%	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690

				Tuberous sclerosis-1, 191100
TSC2	140,5	100.0%	99.9%	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TTC21B	119,5	99.7%	98.8%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC8	115,2	99.8%	98.8%	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
UMOD	110,4	97.6%	94.8%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UPK3A	106,3	100.0%	99.9%	No OMIM phenotype Renal hypodysplasia (Schonfelder (2006) Am J Kidney Dis 47, 1004) Renal aysplasia (Jenkins (2005) J Am Soc Nephrol 16, 2141)
UQCC2	132,2	100.0%	98.1%	Mitochondrial complex III deficiency, nuclear type 7, 615824
VDR	108,8	99.1%	96.0%	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440
VIPAS39	114,7	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	107,2	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	126,8	100.0%	99.2%	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR35	141,8	99.7%	98.4%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	108,1	99.7%	98.1%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR73	153,2	100.0%	99.9%	Galloway-Mowat syndrome 1, 251300
WNK1	134,4	100.0%	99.5%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	143,3	99.9%	99.5%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	226,5	99.5%	97.3%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	90,1	100.0%	99.3%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370

				Wilms tumor, type 1, 194070
XDH	93,8	100.0%	99.7%	Xanthinuria, type I, 278300
XPNPEP3	99,9	100.0%	99.4%	Nephronophthisis-like nephropathy 1, 613159
ZNF423	192,9	100.0%	100.0%	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019
This list is accurate for panel version DG 2.16

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors