RENAL DISORDERS GENE PANEL DG 2.15 (282 genes)

Releasedate: 31-01-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACE	120.7	99.5	97.4	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	143.7	99.5	97.7	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	99.9	96.3	91.7	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS9	141.4	97.9	96.7	Nephronophthisis
ADCY10	149.4	100	99.9	{Hypercalciuria, absorptive, susceptibility to}, 143870
AGT	214.2	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}, 0
AGTR1	134.6	92	91.9	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	139.5	99.9	99.2	Hyperoxaluria, primary, type 1, 259900
AHI1	139.3	99.2	95.1	Joubert syndrome 3, 608629
ALDOB	165.7	100	99.4	Fructose intolerance, hereditary, 229600
ALG8	126	96.5	95.1	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALMS1	179.8	99.9	99.7	Alstrom syndrome, 203800
AMN	66.8	83.5	71.6	Megaloblastic anemia-1, Norwegian type, 261100
ANKS6	91.8	92.8	88.6	Nephronophthisis 16, 615382
ANLN	146.2	97.2	93.3	Focal segmental glomerulosclerosis 8, 616032
ANOS1	90.3	89.4	87.6	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AP2S1	115.7	90.4	89.7	Hypocalciuric hypercalcemia, type III, 600740
APOL1	192.2	100	100	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551

APRT	68.2	100	98.7	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	117.4	99.5	95.6	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	143.9	100	99.9	Nephrotic syndrome, type 8, 615244
ARL13B	97.3	98.9	92.8	Joubert syndrome 8, 612291
ARL6	85.2	99.8	95.3	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ATP6V0A4	116.4	99.9	98.6	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	176.6	100	100	Renal tubular acidosis with deafness, 267300
ATP7B	168.9	100	99.8	Wilson disease, 277900
AVP	53.3	76.3	57.3	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	127.5	99.3	97.1	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	115.3	92.1	91.4	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	110.9	100	100	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BBIP1	132	99.4	94.8	?Bardet-Biedl syndrome 18, 615995
BBS1	148.9	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	172.6	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	208.6	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	181.8	100	99.8	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	135.9	99.7	97.3	Bardet-Biedl syndrome 4, 615982
BBS5	81.1	95.8	84.1	Bardet-Biedl syndrome 5, 615983
BBS7	120.7	98.1	91.7	Bardet-Biedl syndrome 7, 615984
BBS9	112.9	96	93.8	Bardet-Biedl syndrome 9, 615986
BCS1L	182.3	100	100	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BICC1	152.3	100	99.6	{Renal dysplasia, cystic, susceptibility to}, 601331
BSND	137.1	100	100	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522

C3	145.5	100	99.7	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
C5orf42	122.8	98.6	95.5	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	140.7	100	99.3	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CACNA1H	103.4	97.6	94.3	Hyperaldosteronism, familial, type IV, 617027 {Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CASR	178	100	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	127.4	99.5	97.1	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CD2AP	98.2	99.6	96	Glomerulosclerosis, focal segmental, 3, 607832
CD46	115.1	97.8	93.2	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CEP120	129.7	99.8	98.1	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	94.2	99.9	98	Nephronophthisis 15, 614845
CEP290	66.1	88.4	76.7	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	83.5	97.7	89.6	Joubert syndrome 15, 614464
CEP55	129.5	100	99.9	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	96.7	98.3	89.1	Nephronophthisis 18, 615862
CFB	147.1	100	100	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	183.2	98.7	95.3	Basal laminar drusen, 126700

				Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	236.5	95.8	94.2	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	101	90.6	85.6	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFI	145.5	96.6	92.8	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CLCN5	134.6	99.6	98	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCNKB	100.7	98.5	90.5	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN10	148.9	100	100	HELIX syndrome, 617671
CLDN16	136.3	100	99.9	Hypomagnesemia 3, renal, 248250
CLDN19	123.7	98.2	93.7	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	188.4	100	99.2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
COL4A3	89.6	97.8	95.5	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200
COL4A4	85	97.6	93.5	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign, 0
COL4A5	52.3	92.1	77.5	Alport syndrome, 301050
COQ2	89.3	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ6	143.9	99.3	96	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	158.5	99.7	98.9	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8B	90.5	100	99.1	Nephrotic syndrome, type 9, 615573
COQ9	91.4	99.9	96.6	Coenzyme Q10 deficiency, primary, 5, 614654
CRB2	112.4	99.4	94.7	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730

CSPP1	112	99.8	97.8	Joubert syndrome 21, 615636
CTNS	120.1	100	99.9	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CUBN	127.8	99.8	98.4	Megaloblastic anemia-1, Finnish type, 261100
CUL3	110.3	98.6	94.7	Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	165.3	100	100	Hypercalcemia, infantile, 1, 143880
DCDC2	150.5	99.9	99.6	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DGKE	142.3	99.5	95.2	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DMP1	159.5	99.9	99.1	Hypophosphatemic rickets, AR, 241520
DNAJB11	111.3	100	99.6	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DSTYK	144.7	100	99.4	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DYNC2H1	90.5	96.6	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DZIP1L	98.3	99.4	95.5	Polycystic kidney disease 5, 617610
EGF	135.2	100	99.8	Hypomagnesemia 4, renal, 611718
EHHADH	163.2	100	99.7	?Fanconi renotubular syndrome 3, 615605
EMP2	101.1	99.7	96.9	Nephrotic syndrome, type 10, 615861
ENPP1	134.8	92.4	83.2	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	144.2	100	99.7	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
FAH	151.3	100	99.9	Tyrosinemia, type I, 276700
FAM20A	105.4	98.4	92.1	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM58A	73.2	82.8	78.8	STAR syndrome, 300707

FAN1	143.9	100	99.9	Interstitial nephritis, karyomegalic, 614817 Colorectal cancer
FAT1	204.6	100	100	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	106	99.9	97.8	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993
FN1	145.4	100	99.5	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 Spondylometaphyseal dysplasia, corner fracture type, 184255
FOXC2	44.3	95.2	78.8	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXI1	152.5	100	100	Enlarged vestibular aqueduct, 600791
FRAS1	147.8	100	99.7	Fraser syndrome 1, 219000
FREM1	138.4	99.9	99.1	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	182.4	100	99.5	Fraser syndrome 2, 617666
FXYD2	96.7	99.8	99.2	Hypomagnesemia 2, renal, 154020
G6PC	180.7	100	100	Glycogen storage disease Ia, 232200
GALNT3	128.2	99.2	96	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GANAB	120.1	99.9	98.9	Polycystic kidney disease 3, 600666
GATA3	186.8	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GCM2	161.8	100	100	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
GLA	81.3	99.7	97.6	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLIS2	109	99.9	98.2	Nephronophthisis 7, 611498
GLIS3	133.4	99.9	99.3	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	149.5	99.5	96.4	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GREB1L	148.6	100	99.5	Renal hypodysplasia/aplasia 3, 617805

GRHPR	112.5	85.1	78.2	Hyperoxaluria, primary, type II, 260000
GRIP1	130.8	100	99.9	Fraser syndrome 3, 617667
GSN	119.2	94.2	89	Amyloidosis, Finnish type, 105120
HNF1B	123.7	99.9	98.9	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	140.2	99.9	99.1	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HOGA1	147.5	99.8	98.1	Hyperoxaluria, primary, type III, 613616
HPRT1	58.2	96	84.8	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	165.2	85.7	82.5	Apparent mineralocorticoid excess, 218030
IFNG	131	100	99.8	{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	152	100	99.9	Cranioectodermal dysplasia 1, 218330
IFT140	114.7	99.9	99	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99.6	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	131.7	100	99.6	?Bardet-Biedl syndrome 19, 615996
IFT43	114.8	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
INF2	79.2	84.1	81.1	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
INPP5E	89.1	95.8	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INTU	122	99.7	96.6	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	160.5	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92.2	89.3	75.4	Senior-Loken syndrome 5, 609254

ITGA3	141.5	99.8	98.3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA8	122.4	99.6	98.1	Renal hypodysplasia/aplasia 1, 191830
JAG1	148.4	98.1	97.5	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KANK2	151.4	99.9	99.5	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KCNJ1	233.9	100	100	Bartter syndrome, type 2, 241200
KCNJ10	213.4	89.3	89.1	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ5	193.9	100	99.8	Hyperaldosteronism, familial, type III, 613677 Long QT syndrome 13, 613485
KIAA0556	134.4	99.9	99.4	Joubert syndrome 26, 616784
KIF14	111.3	98.1	89.9	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF7	85.7	93.5	88.9	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KL	179.9	97.2	96	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLHL3	141.1	100	99.8	Pseudohypoaldosteronism, type IID, 614495
LAGE3	50.3	95	81.2	Galloway-Mowat syndrome 2, X-linked, 301006
LAMB2	201.7	100	99.8	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCAT	145.6	97.8	91.7	Fish-eye disease, 136120 Norum disease, 245900
LMX1B	111.4	97	92.3	Nail-patella syndrome, 161200
LRIG2	148.5	99.4	97.5	Urofacial syndrome 2, 615112
LRP2	176.3	100	99.8	Donnai-Barrow syndrome, 222448
LRP4	166.6	99.1	98.9	?Myasthenic syndrome, congenital, 17, 616304 Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LRP5	189.8	98.2	97.9	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	165.3	100	100	Amyloidosis, renal, 105200
LZTFL1	109.1	99.1	95.3	Bardet-Biedl syndrome 17, 615994
MAFB	93.8	99.7	97.8	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	80	99.7	97.4	Bartter syndrome, type 5, antenatal, transient, 300971
MAGI2	98	91.8	88.3	Nephrotic syndrome, type 15, 617609
MAPKBP1	144.1	100	100	Nephronophthisis 20, 617271
MKKS	208.5	83.2	83.1	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	114.5	99.9	98.5	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MOCOS	169	99.1	96.6	Xanthinuria, type II, 603592
MYH9	130.5	99.4	98.1	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO1E	131.9	98.6	97	Glomerulosclerosis, focal segmental, 6, 614131
NCAPG2	127.3	99.3	96.8	nephronophthisis syndrome
NEK1	103.2	98.1	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK8	171.4	100	99.9	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NOTCH2	172.4	100	99.9	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP1	117.6	98.8	96.4	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	115.6	99.4	96.1	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387

				Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	136.7	99.9	99.3	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	102	99.9	98.9	Nephrotic syndrome, type 1, 256300
NPHS2	116.9	99.8	95.9	Nephrotic syndrome, type 2, 600995
NR3C2	159.5	99.4	95.9	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NUP107	122.9	99.2	94.1	?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP133	127.1	96.9	94.3	Nephrotic syndrome, type 18, 618177
NUP160	161.1	100	99.8	?Nephrotic syndrome, type 19, 618178
NUP205	133.7	98.9	98	?Nephrotic syndrome, type 13, 616893
NUP85	135.3	100	100	Nephrotic syndrome, type 17, 618176
NUP93	140.7	97.9	94.9	Nephrotic syndrome, type 12, 616892
OCRL	122.2	98.8	96.3	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	51.5	84	67.8	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OSGEP	120.5	100	99.6	Galloway-Mowat syndrome 3, 617729
PAX2	168.5	99.9	99.3	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PBX1	111.8	99.3	95.2	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCBD1	113.3	99.5	99.1	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDE6D	106.1	100	99.9	?Joubert syndrome 22, 615665
PDSS2	126.8	96.5	93.5	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	125	99.9	98	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	28.1	42.6	34.5	Polycystic kidney disease 1, 173900
PKD2	110.6	89.3	84.2	Polycystic kidney disease 2, 613095
PKHD1	154.9	100	99.7	Polycystic kidney disease 4, with or without hepatic disease, 263200
PLCE1	155.3	99.5	98.9	Nephrotic syndrome, type 3, 610725
PMM2	141.1	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065

PTH1R	108.5	99.9	98.8	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPRO	140.8	99.9	99	Nephrotic syndrome, type 6, 614196
REN	148.9	100	100	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia], 0
RMND1	137.2	99.8	97.3	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	155.2	97.9	96.7	Vesicoureteral reflux 2, 610878
RPGRIP1L	126.2	96.4	93.9	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RRM2B	128.6	99.7	97.5	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	138.5	99.3	98.4	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480
SALL4	147.5	97.6	96.3	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SARS2	104.8	94.8	92.7	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	121.2	100	99.9	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	134.3	99	96.3	?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	148.9	100	99.8	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome 1, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	139.4	99.7	97.1	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 618114 Pseudohypoaldosteronism, type I, 264350
SDCCAG8	123.9	99.8	97.4	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615

SEC61A1	132.9	100	100	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SGPL1	164.1	100	100	Nephrotic syndrome, type 14, 617575
SIX5	43.8	88.3	76.1	Branchiootorenal syndrome 2, 610896
SLC12A1	172.7	99.8	99.1	Bartter syndrome, type 1, 601678
SLC12A3	139.3	100	100	Gitelman syndrome, 263800
SLC16A12	164.4	100	99.9	Cataract 47, juvenile, with microcornea, 612018
SLC22A12	105.7	100	99.7	Hypouricemia, renal, 220150
SLC26A1	139.8	100	99.9	?Nephrolithiasis, calcium oxalate, 167030
SLC26A3	156.1	99.9	98.9	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	178.4	100	99.9	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	119	99.2	96.2	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC34A1	153.2	100	99.5	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	105.6	98.9	94.5	Hypophosphatemic rickets with hypercalciuria, 241530
SLC36A2	123.9	100	100	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC37A4	140.2	100	99.9	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC3A1	162.8	100	99.5	Cystinuria, 220100
SLC41A1	149.8	100	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	140	100	99.9	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	122.3	99	97.1	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	118.7	100	100	Renal glucosuria, 233100
SLC6A19	149.1	100	99.3	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A20	178.6	100	99.9	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC7A7	123.9	100	99.9	Lysinuric protein intolerance, 222700
SLC7A9	125.5	99.9	99	Cystinuria, 220100
SLC9A3	147.1	98.7	96.9	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	111.5	99.5	96	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLIT3	146.8	97.3	95.2	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	134.6	100	99.9	Schimke immunoosseous dysplasia, 242900
SOX17	70.8	99.6	94.6	Vesicoureteral reflux 3, 613674
STRA6	116.5	100	99.9	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STX16	140.6	99.8	97.8	Pseudohypoparathyroidism, type IB, 603233
TBX18	94.4	98	95.2	Congenital anomalies of kidney and urinary tract 2, 143400
TCTN1	98.8	95.7	92.8	Joubert syndrome 13, 614173
TCTN2	144.2	99.5	97	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	127.6	100	99.8	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
THBD	108.2	99.8	97.8	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TMEM107	163.8	100	100	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM138	100.2	100	99.5	Joubert syndrome 16, 614465

TMEM216	111.9	100	98.7	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	111.5	100	99.9	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100.7	99.8	98.3	Joubert syndrome 14, 614424
TMEM260	116.7	96.8	90	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	72.9	93.3	83.4	?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	96.4	98.4	91.4	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP53RK	37.4	91.3	76.5	Galloway-Mowat syndrome 4, 617730
TPRKB	56.7	79.3	67.1	Galloway-Mowat syndrome 5, 617731
TRAF3IP1	90.3	96.3	92.8	Senior-Loken syndrome 9, 616629
TRIM32	141.2	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRPC6	103.8	99	96.1	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	151.1	99.8	98.7	Hypomagnesemia 1, intestinal, 602014
TSC1	128.8	99.8	98.8	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	131.2	100	99	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TTC21B	100.7	99.7	97.6	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC8	106.9	97.9	92	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
UMOD	127	97.8	97.2	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UPK3A	110.5	99.2	96.7	No OMIM phenotype

				Renal hypodysplasia (Schonfelder (2006) Am J Kidney Dis 47, 1004) Renal aysplasia (Jenkins (2005) J Am Soc Nephrol 16, 2141)
UQCC2	96.6	100	99.1	Mitochondrial complex III deficiency, nuclear type 7, 615824
VDR	123.3	98	95.2	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440
VIPAS39	144.6	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	138.3	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	132.1	99.8	98.1	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR35	145.1	99.3	97.7	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	114.2	99.1	96.3	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR73	138.9	100	100	Galloway-Mowat syndrome 1, 251300
WNK1	167.7	99.9	99.5	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	123.1	99.7	98.7	Pseudohypoaldosteronism, type IIB, 614491
WNT4	263.1	93.4	92.7	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	76.5	91.8	81.4	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
XDH	109.1	100	99.9	Xanthinuria, type I, 278300
XPNPEP3	134	100	99.2	Nephronophthisis-like nephropathy 1, 613159
ZNF423	250.8	100	100	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 : December 31st , 2018.
This list is accurate for panel version DG 2.15

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