RENAL DISORDERS GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ACTN4	95.0	99%	96%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	57.7	94%	84%	Thrombotic thrombocytopenic purpura, familial, 274150
ADCK4	64.0	100%	91%	Nephrotic syndrome type 9, 615573
AGTR1	147.4	97%	97%	Hypertension, essential, 145500
AGXT	88.5	97%	90%	Hyperoxaluria, primary, type 1, 259900
AHI1	114.6	100%	99%	Joubert syndrome-3, 608629
ALG8	85.2	96%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALMS1	195.4	98%	98%	Alstrom syndrome, 203800
ANKS6	62.7	96%	85%	Nephronophthisis 16, 615382
AP2S1	84.7	100%	100%	Hypocalciuric hypercalcemia, familial, type III, 600740
APRT	47.5	100%	91%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	87.6	91%	87%	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	115.8	100%	100%	?Nephrotic syndrome type 8, 615244
ARL13B	126.0	99%	95%	Joubert syndrome 8, 612291
ARL6	161.4	100%	100%	Bardet-Biedl syndrome 3, 209900 Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ATP6V0A4	77.6	99%	93%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	98.0	100%	98%	Renal tubular acidosis with deafness, 267300
ATXN10	118.5	100%	100%	Spinocerebellar ataxia 10, 603516
AVPR2	45.6	94%	86%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	83.9	100%	93%	Meckel syndrome 9, 614209
B9D2	49.9	100%	98%	Meckel syndrome 10, 614175
BBS1	114.8	100%	98%	Bardet-Biedl syndrome 1, 209900
BBS10	126.9	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	144.1	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	115.0	100%	99%	Bardet-Biedl syndrome 2, 209900
BBS4	91.7	95%	91%	Bardet-Biedl syndrome 4, 209900
BBS7	129.6	100%	99%	Bardet-Biedl syndrome 7, 209900

BBS9	124.3	100%	100%	Bardet-Biedl syndrome 9, 209900
BCS1L	148.6	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358
BICC1	96.0	100%	99%	{Renal dysplasia, cystic, susceptibility to}, 601331
BSND	116.3	100%	98%	Bartter syndrome, type 4a, 602522 Sen sorineural deafness with mild renal dysfunction, 602522
CA2	143.7	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CASR	118.8	100%	99%	Hypocalciuric hypercalcemia, type I, 145980
CC2D2A	92.9	98%	97%	Joubert syndrome 9, 612285
CD2AP	116.4	100%	99%	Glomerulosclerosis, focal segmental, 3, 607832
CEP164	73.8	98%	91%	Nephronophthisis 15, 614845
CEP290	101.5	100%	98%	Joubert syndrome 5, 610188
CEP41	88.3	100%	99%	Joubert syndrome 15, 614464
CFHR5	107.7	94%	92%	Nephropathy due to CFHR5 deficiency, 614809
CLCN5	84.2	99%	94%	Dent disease, 300009
CLCNKB	74.1	86%	80%	Bartter syndrome, type 3, 607364
CLDN16	126.8	98%	95%	Hypomagnesemia 3, renal, 248250
CLDN19	69.3	100%	93%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	136.5	100%	99%	Hypomagnesemia 6, renal, 613882
COL4A1	87.4	99%	96%	Porencephaly 1, 175780
COL4A3	74.2	97%	94%	Alport syndrome, autosomal recessive, 203780
COL4A4	92.0	100%	97%	Alport syndrome, autosomal recessive, 203780
COL4A5	38.2	94%	78%	Alport syndrome, 301050
COQ2	75.5	99%	96%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	109.2	99%	95%	Coenzyme Q10 deficiency, primary, 6, 614650
CTNS	114.9	97%	94%	Cystinosis, nephropathic, 219800
CUBN	88.4	99%	96%	Megaloblastic anemia-1, Finnish type, 261100
DGKE	111.9	100%	99%	Nephrotic syndrome, type 7, 615008

DMP1	110.0	100%	100%	Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Brachydactyly, type A2, 112600 Symphalangism, proximal, 1B, 615298 Multiple synostoses syndrome 2, 610017 {Osteoarthritis-5}, 612400 Brachydactyly, type A1, C, 615072 Hypophosphatemic rickets, AR, 241520
DSTYK	100.2	99%	97%	{Congenital anomalies of kidney and urinary tract, susceptibility to}, 610805
DYNC2H1	116.3	100%	99%	Asphyxiating thoracic dystrophy 3, 613091
EGF	113.7	99%	98%	Hypomagnesemia 4, renal, 611718
EHHADH	153.4	100%	100%	?Fanconi renotubular syndrome 3, 615605
EYA1	113.1	100%	99%	Branchiootorenal syndrome 1, with or without cataracts, 113650
FAM58A	20.3	48%	45%	STAR syndrome, 300707
FAN1	111.8	100%	99%	Interstitial nephritis, karyomegalic, 614817
FGF23	75.3	96%	92%	Hypophosphatemic rickets, autosomal dominant, 193100
FN1	88.7	99%	96%	Glomerulopathy with fibronectin deposits 2, 601894
FRAS1	99.0	98%	95%	Fraser syndrome, 219000
FREM1	108.6	100%	98%	Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	127.7	100%	99%	Fraser syndrome, 219000
FXYD2	64.3	99%	80%	Hypomagnesemia-2, renal, 154020
GATA3	126.7	100%	98%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GLA	47.2	95%	86%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	73.6	99%	94%	GM1-gangliosidosis, type I, 230500
GLIS2	88.9	100%	98%	Nephronophthisis 7, 611498
GLIS3	95.7	100%	98%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	95.5	100%	99%	Hypocalciuric hypercalcemia, type II, 145981
HNF1B	78.7	98%	96%	Renal cysts and diabetes syndrome, 137920
HPRT1	50.8	100%	78%	Lesch-Nyhan syndrome, 300322
HSD11B2	113.2	78%	78%	Apparent mineralocorticoid excess, 218030
IFT122	79.1	96%	95%	Cranioectodermal dysplasia 1, 218330
IFT140	83.3	99%	95%	Mainzer-Saldino syndrome, 266920
IFT172	97.6	100%	97%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630

IFT43	84.7	100%	100%	Cranioectodermal dysplasia 3, 614099
INF2	67.9	93%	85%	Glomerulosclerosis, focal segmental, 5, 613237
INPP5E	74.2	100%	99%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	124.2	100%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	95.1	99%	93%	Senior-Loken syndrome 5, 609254
ITGA8	95.9	99%	98%	Renal hypodysplasia/aplasia 1, 191830
JAG1	110.2	99%	96%	Alagille syndrome, 118450
KAL1	47.6	93%	81%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KCNJ1	146.1	97%	97%	Bartter syndrome, type 2, 241200
KCNJ10	151.4	100%	100%	SESAME syndrome, 612780
KIF7	67.0	93%	86%	Hydrolethalus syndrome 2, 614120
LAMB2	111.3	100%	99%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199
LCAT	107.1	93%	88%	Norum disease, 245900
LMX1B	86.7	100%	98%	Nail-patella syndrome, 161200
LRP2	107.4	100%	99%	Donnai-Barrow syndrome, 222448
LRP4	93.9	99%	96%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LYZ	108.4	100%	100%	Amyloidosis, renal, 105200
LZTFL1	90.2	100%	100%	Bardet-Biedl syndrome 17, 615994
MAFB	87.4	100%	100%	Multicentric carpotarsal osteolysis syndrome, 166300
MKKS	148.8	100%	100%	McKusick-Kaufman syndrome, 236700
MKS1	110.6	100%	99%	Meckel syndrome 1, 249000
MYH9	96.5	100%	98%	May-Hegglin anomaly, 155100
MYO1E	92.5	100%	98%	Glomerulosclerosis, focal segmental, 6, 614131
NEK1	129.9	100%	99%	Short rib-polydactyly syndrome, type IIA, 263520
NEK8	111.4	100%	100%	?Nephronophthisis 9, 613824
NOTCH2	89.5	91%	89%	Alagille syndrome 2, 610205
NPHP1	167.1	100%	100%	Nephronophthisis 1, juvenile, 256100
NPHP3	110.4	100%	99%	Nephronophthisis 3, 604387
NPHP4	89.5	98%	93%	Nephronophthisis 4, 606966
NPHS1	87.3	100%	96%	Nephrotic syndrome, type 1, 256300
NPHS2	128.4	100%	100%	Nephrotic syndrome, type 2, 600995
NR3C2	128.3	100%	96%	Pseudohypoaldosteronism type I, autosomal dominant, 177735
OCRL	58.4	97%	94%	Lowe syndrome, 309000
OFD1	37.0	88%	78%	Oral-facial-digital syndrome 1, 311200

PAX2	105.5	96%	94%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830
PDSS2	90.7	100%	99%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	63.1	100%	98%	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	13.9	19%	18%	Polycystic kidney disease, adult type I, 173900
PKD2	101.4	98%	92%	Polycystic kidney disease 2, 613095
PKHD1	103.8	98%	96%	Polycystic kidney and hepatic disease, 263200
PLCE1	123.8	99%	97%	Nephrotic syndrome, type 3, 610725
PTH1R	77.8	97%	93%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk-Jansen type, 156400
PTPRO	106.4	98%	97%	Nephrotic syndrome, type 6, 614196
REN	104.6	100%	100%	Hyperproreninemia]
RET	90.8	98%	94%	Multiple endocrine neoplasia IIA, 171400
ROBO2	108.8	100%	99%	Vesicoureteral reflux 2, 610878
RPGRIP1L	101.3	98%	96%	Joubert syndrome 7, 611560
RRM2B	108.0	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075
SALL1	145.0	100%	98%	Townes-Brocks syndrome, 107480
SALL4	97.7	97%	95%	Duane-radial ray syndrome, 607323
SARS2	66.5	98%	91%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	88.5	100%	95%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	94.5	97%	93%	Pseudohypoaldosteronism, type I, 264350
SCNN1B	89.4	98%	96%	Liddle syndrome, 177200
SCNN1G	129.4	100%	99%	Liddle syndrome, 177200
SDCCAG8	105.0	100%	100%	Senior-Loken syndrome 7, 613615
SIX1	80.7	100%	100%	Brachiootic syndrome 3, 608389
SIX5	44.7	94%	81%	Branchiootorenal syndrome 2, 610896
SLC12A1	142.0	100%	100%	Bartter syndrome, type 1, 601678
SLC12A3	85.6	99%	97%	Gitelman syndrome, 263800
SLC16A12	110.4	99%	95%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC22A12	85.0	96%	91%	Hypouricemia, renal, 220150
SLC26A3	113.4	100%	98%	?Colon cancer (1)
SLC2A2	125.4	100%	100%	{Diabetes mellitus, noninsulin-dependent}

SLC2A9	71.0	100%	97%	{Uric acid concentration, serum, QTL 2}, 612076
SLC34A1	84.3	99%	96%	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	74.0	98%	90%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC3A1	134.0	100%	100%	Cystinuria, 220100
SLC41A1	92.1	100%	98%	Parkinson disease (Yan (2011) Int J Neurosci 121,632)
SLC4A1	91.9	99%	96%	Ovalocytosis
SLC4A4	108.4	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	81.4	98%	95%	Renal glucosuria, 233100
SLC6A19	93.4	98%	95%	Hartnup disorder, 234500
SLC6A20	86.5	91%	89%	Hyperglycinuria, 138500
SLC7A7	90.3	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	69.4	100%	96%	Cystinuria, 220100
SLC9A3R1	101.2	100%	95%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SMARCAL1	121.3	99%	96%	Schimke immunoosseous dysplasia, 242900
SOX17	74.3	100%	100%	Vesicoureteral reflux 3, 613674
STRA6	71.7	100%	96%	Microphthalmia, syndromic 9, 601186
STX16	113.8	100%	100%	Pseudohypoparathyroidism, type IB, 603233
TCTN1	103.9	96%	94%	Joubert syndrome 13, 614173
TCTN2	91.8	100%	98%	Meckel syndrome 8, 613885
TCTN3	103.8	100%	99%	Orofaciodigital syndrome IV, 258860
TMEM138	97.1	100%	100%	Joubert syndrome 16, 614465
TMEM216	70.2	97%	77%	Joubert syndrome 2, 608091
TMEM231	73.6	97%	90%	Joubert syndrome 20, 614970
TMEM237	101.8	100%	94%	Joubert syndrome 14, 614424
TMEM67	115.6	100%	99%	Meckel syndrome 3, 607361
TRIM32	106.2	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110
TRPC6	79.3	94%	89%	Glomerulosclerosis, focal segmental, 2, 603965
TSC1	95.7	99%	97%	Tuberous sclerosis-1, 191100
TSC2	85.9	99%	95%	Tuberous sclerosis-2, 613254
TTC21B	112.5	99%	98%	Nephronophthisis 12, 613820
TTC8	111.8	100%	100%	Bardet-Biedl syndrome 8, 209900
UMOD	82.4	99%	96%	Hyperuricemic nephropathy, familial juvenile 1, 162000
UPK3A	60.3	96%	85%	Renal adysplasia, 191830
VIPAS39	115.3	100%	97%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	102.6	100%	97%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	123.9	100%	100%	Asphyxiating thoracic dystrophy 5, 614376

WDR35	121.3	99%	97%	Cranioectodermal dysplasia 2, 613610
WDR60	101.9	99%	98%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNK1	144.1	100%	99%	Pseudohypoaldosteronism, type IIC, 614492
WNK4	114.7	100%	99%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	141.9	92%	92%	SERKAL syndrome, 611812
WT1	66.3	100%	98%	Wilms tumor, type 1, 194070
XPNPEP3	137.5	100%	99%	Nephronophthisis-like nephropathy 1, 613159
ZNF423	127.9	100%	99%	Nephronophthisis 14, 614844

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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
OMIM release used for OMIM disease identifiers and descriptions : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors