RENAL DISORDERS GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ACTN4	95	99%	96%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	57,7	94%	84%	Thrombotic thrombocytopenic purpura, familial, 274150
AGTR1	147,4	97%	97%	Hypertension, essential, 145500 Renal tubular dysgenesis, 267430
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	99%	95%	Joubert syndrome 8, 612291
ARL6	161,4	100%	100%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ATP6V0A4	77,6	99%	93%	Renal tubular acidosis, distal, autosomal recessive, 602722

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	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	100%	99%	{Renal dysplasia, cystic, susceptibility to}, 601331
BSND	116,3	100%	98%	Bartter syndrome, type 4a, 602522 Sensorineural 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 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 Hypercalciuric hypercalcemia {Calcium, serum level of}
CC2D2A	92,9	98%	97%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
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 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
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 Nephrolithiasis, type I, 310468 Hypophosphatemic rickets, 300554 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCNKB	74,1	86%	80%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
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 Brain small vessel disease with hemorrhage, 607595 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle, 611773 Brain small vessel disease with Axenfeld-Rieger anomaly, 607595 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	74,2	97%	94%	Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200 Alport syndrome, autosomal dominant, 104200
COL4A4	92	100%	97%	Alport syndrome, autosomal recessive, 203780 Hematuria,familial benign
COL4A5	38,2	94%	78%	Alport syndrome, 301050
COQ2	75,5	99%	96%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ6	109,2	99%	95%	Coenzyme Q10 deficiency, primary, 6, 614650
CTNS	114,9	97%	94%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800 -3
CUBN	88,4	99%	96%	Megaloblastic anemia-1, Finnish type, 261100
DGKE	111,9	100%	99%	Nephrotic syndrome, type 7, 615008
DMP1	110	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
DMP1	110	100%	100%	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 Short rib-polydactyly syndrome, type III, 263510 Short rib-polydactyly syndrome, type IIB, 615087
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 Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Otofaciocervical syndrome, 166780
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 Osteomalacia, tumor-induced (1) Tumoral calcinosis, hyperphosphatemic, familial, 211900
FN1	88,7	99%	96%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101 (1)
FRAS1	99	98%	95%	Fraser syndrome, 219000
FREM1	108,6	100%	98%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
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 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLIS2	88,9	100%	98%	Nephronophthisis 7, 611498
GLIS3	95,7	100%	98%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 -3
GNA11	95,5	100%	99%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GSN	71,1	93%	86%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
GSN	71,1	93%	86%	Amyloidosis, Finnish type, 105120
HNF1B	78,7	98%	96%	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853 {Renal cell carcinoma}, 144700
HPRT1	50,8	100%	78%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
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 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
INPP5E	74,2	100%	99%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INVS	124,2	100%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	95,1	99%	93%	Senior-Loken syndrome 5, 609254

JAG1	110,2	99%	96%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 Deafness, congenital heart defects, and posterior embryotoxon
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 Enlarged vestibular aqueduct, digenic, 600791
KIF7	67	93%	86%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
LAMB2	111,3	100%	99%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCAT	107,1	93%	88%	Norum disease, 245900 Fish-eye disease, 136120
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 Bardet-Biedl syndrome 6, 209900
MKS1	110,6	100%	99%	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 209900

MYH9	96,5	100%	98%	May-Hegglin anomaly, 155100 Fechtner syndrome, 153640 Sebastian syndrome, 605249 Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Macrothrombocytopenia and progressive sensorineural deafness, 600208
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 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NOTCH2	89,5	91%	89%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP1	167,1	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	110,4	100%	99%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	89,5	98%	93%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
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 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
OCRL	58,4	97%	94%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	37	88%	78%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
PAX2	105,5	96%	94%	Ataxia, spastic 2, autosomal recessive (2)

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
PTPRO	106,4	98%	97%	Nephrotic syndrome, type 6, 614196
REN	104,6	100%	100%	Hyperproreninemia] Renal tubular dysgenesis, 267430 Hyperuricemic nephropathy, familial juvenile 2, 613092
RET	90,8	98%	94%	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIB, 162300 Central hypoventilation syndrome, congenital, 209880 Pheochromocytoma, 171300 Renal agenesis, 191830 {Hirschsprung disease, susceptibility to, 1}, 142623
ROBO2	108,8	100%	99%	Vesicoureteral reflux 2, 610878
RPGRIP1L	101,3	98%	96%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RRM2B	108	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, AD, 5, 613077 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
SALL1	145	100%	98%	Townes-Brocks syndrome, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	97,7	97%	95%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750

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 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	89,4	98%	96%	Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 1, 211400
SCNN1G	129,4	100%	99%	Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 3, 613071
SDCCAG8	105	100%	100%	Senior-Loken syndrome 7, 613615
SIX1	80,7	100%	100%	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX5	44,7	94%	81%	Branchiootorenal syndrome 2, 610896
SLC12A1	142	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	96%	91%	Hypouricemia, renal, 220150
SLC26A3	113,4	100%	98%	?Colon cancer (1) Chloride diarrhea, congenital, Finnish type, 214700
SLC2A2	125,4	100%	100%	{Diabetes mellitus, noninsulin-dependent} Fanconi-Bickel syndrome, 227810
SLC2A9	71	100%	97%	{Uric acid concentration, serum, QTL 2}, 612076 Hypouricemia, renal, 2, 612076
SLC34A1	84,3	99%	96%	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Fanconi renotubular syndrome 2, 613388
SLC34A3	74	98%	90%	Hypophosphatemic rickets with hypercalciuria, 241530

SLC3A1	134	100%	100%	Cystinuria, 220100
SLC41A1	92,1	100%	98%	Parkinson disease (Yan (2011) Int J Neurosci 121,632) Nephrolithiasis-like phenotype (Hurd (2013) J Am Soc Nephrol 24, 967)
SLC4A1	91,9	99%	96%	Ovalocytosis Spherocytosis, type 4, 612653 [Malaria, resistance to], 611162 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590 [Blood group, Diego], 110500 [Blood group, Waldner], 112010 [Blood group, Wright], 112050 [Blood group, Froese], 601551 [Blood group, Swann], 601550
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 Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC6A20	86,5	91%	89%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
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 Microphthalmia, isolated, with coloboma 8, 601186
STX16	113,8	100%	100%	Pseudohypoparathyroidism, type IB, 603233

TCTN2	91,8	100%	98%	Meckel syndrome 8, 613885
TCTN3	103,8	100%	99%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TMEM138	97,1	100%	100%	Joubert syndrome 16, 614465
TMEM216	70,2	97%	77%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	73,6	97%	90%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397 -3
TMEM237	101,8	100%	94%	Joubert syndrome 14, 614424
TMEM67	115,6	100%	99%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TRIM32	106,2	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TRPC6	79,3	94%	89%	Glomerulosclerosis, focal segmental, 2, 603965
TSC1	95,7	99%	97%	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341
TSC2	85,9	99%	95%	Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690
TTC21B	112,5	99%	98%	Nephronophthisis 12, 613820 Asphyxiating thoracic dystrophy 4, 613819
TTC8	111,8	100%	100%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
UMOD	82,4	99%	96%	Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860 Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886
UPK3A	60,3	96%	85%	Renal adysplasia, 191830 Urogenital 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 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378
WDR35	121,3	99%	97%	Cranioectodermal dysplasia 2, 613610 Short rib-polydactyly syndrome, type V, 614091
WDR60	101,9	99%	98%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 Jeune syndrome (McInerney-Leo (2013) Am J Hum Genet 93, 515)
WNK1	144,1	100%	99%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
WNK4	114,7	100%	99%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	141,9	92%	92%	SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	66,3	100%	98%	Wilms tumor, type 1, 194070 Denys-Drash syndrome, 194080 Nephrotic syndrome, type 4, 256370 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240
XPNPEP3	137,5	100%	99%	Nephronophthisis-like nephropathy 1, 613159
ZNF423	127,9	100%	99%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
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 : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors