RENAL DISORDERS GENE PANEL DG 2.3.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
ACTN4	91,2	98%	94%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	58,2	92%	78%	Thrombotic thrombocytopenic purpura, familial, 274150
ADCK4	65,6	99%	96%	Nephrotic syndrome type 9, 615573
AGTR1	152,3	100%	100%	Hypertension, essential, 145500
AGXT	89,7	99%	93%	Hyperoxaluria, primary, type 1, 259900
AHI1	105,6	100%	98%	Joubert syndrome-3, 608629
ALG8	87,7	96%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALMS1	189	98%	98%	Alstrom syndrome, 203800
ANKS6	56,3	94%	80%	Nephronophthisis 16, 615382
AP2S1	82,7	90%	88%	Hypocalciuric hypercalcemia, familial, type III, 600740
APRT	54,4	99%	88%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	82,7	99%	86%	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	102,1	100%	100%	?Nephrotic syndrome type 8, 615244
ARL13B	121,8	100%	95%	Joubert syndrome 8, 612291
ARL6	155,1	100%	100%	Bardet-Biedl syndrome 3, 209900 Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ATP6V0A4	77,5	96%	92%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	104,1	100%	98%	Renal tubular acidosis with deafness, 267300
ATXN10	110	100%	99%	Spinocerebellar ataxia 10, 603516
AVPR2	47,3	97%	94%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	72,5	92%	86%	Meckel syndrome 9, 614209
B9D2	49,5	100%	100%	Meckel syndrome 10, 614175
BBS1	116,1	99%	98%	Bardet-Biedl syndrome 1, 209900
BBS10	115,8	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	132,3	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	114,4	100%	100%	Bardet-Biedl syndrome 2, 209900

BBS4	98,5	96%	93%	Bardet-Biedl syndrome 4, 209900
BBS7	119,1	100%	98%	Bardet-Biedl syndrome 7, 209900
BBS9	122,1	100%	98%	Bardet-Biedl syndrome 9, 209900
BCS1L	133,7	100%	100%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358
BICC1	97,7	100%	97%	{Renal dysplasia, cystic, susceptibility to}, 601331
BSND	93,5	100%	99%	Bartter syndrome, type 4a, 602522 Sen sorineural deafness with mild renal dysfunction, 602522
CA2	147,9	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CASR	118	100%	99%	Hyperparathyroidism,neonatal,239200 Hypocalcemia,autosomal dominant,601198 Hypocalciuric hypercalcemia,type I,145980 {Epilepsy idiopathic generalized,susceptibility to,8},612899
CC2D2A	94,8	98%	97%	COACH syndrome,216360 Joubert syndrome 9,612285 Meckel syndrome 6,612284
CCDC41	123,6	100%	100%	Nephronophthisis 18,615862
CD2AP	112,5	100%	99%	Glomerulosclerosis, focal segmental, 3, 607832
CEP164	72,4	97%	90%	Nephronophthisis 15, 614845
CEP290	95,3	100%	98%	?Bardet-Biedl syndrome 14,615991 Joubert syndrome 5,610188 Leber congenital amaurosis 10,611775 Meckel syndrome 4,611134 Senior-Loken syndrome 6,610189
CEP41	89,6	100%	100%	Joubert syndrome 15, 614464
CLCN5	82,1	100%	98%	Dent disease, 300009 Hypophosphatemic rickets,300554 Nephrolithiasis,type I,310468 Proteinuria,low molecular weight,with hypercalciuric nephrocalcinosis,308990
CLCNKB	77,3	88%	84%	Bartter syndrome, type 3, 607364 Bartter syndrome,type 4b,digenic,613090
CLDN16	129,1	95%	92%	Hypomagnesemia 3, renal, 248250

CLDN19	77,2	100%	99%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	128,7	100%	99%	Hypomagnesemia 6, renal, 613882
COL4A1	79,8	98%	94%	Porencephaly 1, 175780
COL4A3	72	97%	93%	Alport syndrome, autosomal recessive, 203780 Alport syndrome, autosomal dominant, 104200 Hematuria,benign familial, 141200
COL4A4	83,4	99%	96%	Alport syndrome, autosomal recessive, 203780
COL4A5	35,5	94%	74%	Alport syndrome, 301050
COQ2	69,8	94%	81%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	108,9	100%	95%	Coenzyme Q10 deficiency, primary, 6, 614650
CRB2	76,6	98%	87%	Focal segmental glomerulosclerosis 9
CTNS	110,7	96%	90%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic,219900 Cystinosis,ocular nonnephropathic,219750
CUBN	83,8	98%	96%	Megaloblastic anemia-1, Finnish type, 261100
DCDC2	159,6	100%	99%	Nephronophthisis 19
DGKE	102,9	100%	98%	Nephrotic syndrome, type 7, 615008
DMP1	106,4	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	101,2	99%	97%	{Congenital anomalies of kidney and urinary tract, susceptibility to}, 610805
DYNC2H1	113,2	99%	98%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
EGF	106,7	99%	98%	Hypomagnesemia 4, renal, 611718
EHHADH	141,5	100%	100%	?Fanconi renotubular syndrome 3, 615605
EMP2	70,2	100%	97%	Nephrotic syndrome, type 10,615861
EYA1	112,2	100%	99%	Branchiootorenal syndrome 1, with or without cataracts, 113650
FAM58A	17,5	50%	34%	STAR syndrome, 300707

FAN1	106,1	99%	98%	Interstitial nephritis, karyomegalic, 614817
FGF23	78	99%	94%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia,tumor-induced Tumoral calcinosis,hyperphosphatemic,familial,211900
FN1	87,6	98%	94%	Glomerulopathy with fibronectin deposits 2, 601894
FRAS1	96,4	98%	95%	Fraser syndrome, 219000
FREM1	103,7	100%	98%	Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	124,7	100%	99%	Fraser syndrome, 219000
FXYD2	69,2	81%	80%	Hypomagnesemia-2, renal, 154020
GALNT3	111,2	100%	99%	Tumoral calcinosis, hyperphosphatemic, familial,211900
GATA3	129,9	100%	99%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GLA	46,1	98%	93%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	75,6	100%	95%	GM1-gangliosidosis, type I, 230500
GLIS2	88,3	100%	97%	Nephronophthisis 7, 611498
GLIS3	95,9	100%	98%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	82,1	100%	98%	Hypocalcemia,autosomal dominant 2,615361 Hypocalciuric hypercalcemia, type II, 145981
GSN	80,5	95%	90%	Amyloidosis, Finnish type, 105120 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HNF1B	78,3	100%	94%	Diabetes mellitus,noninsulin-dependent,125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma},144700
HPRT1	50	97%	81%	HPRT-related gout,300323 Lesch-Nyhan syndrome, 300322
HSD11B2	115,7	78%	76%	Apparent mineralocorticoid excess, 218030
IFT122	77,6	96%	92%	Cranioectodermal dysplasia 1, 218330
IFT140	80,2	99%	94%	Mainzer-Saldino syndrome, 266920
IFT172	92,7	99%	96%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	88,9	100%	100%	Cranioectodermal dysplasia 3, 614099
INF2	63,8	93%	87%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease,dominant intermediate E,614455
INPP5E	75,8	99%	96%	Joubert syndrome 1,213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156

INVS	114,4	99%	97%	Nephronophthisis 2, infantile, 602088
IQCB1	93	100%	93%	Senior-Loken syndrome 5, 609254
ITGA8	93,3	99%	97%	Renal hypodysplasia/aplasia 1, 191830
JAG1	101,6	97%	95%	Alagille syndrome, 118450
KAL1	44,3	88%	78%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KCNJ1	139,3	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	158,3	100%	100%	SESAME syndrome, 612780
KIF7	68,8	89%	84%	Hydrolethalus syndrome 2, 614120
KL	139,5	97%	96%	Tumoral calcinosis, hyperphosphatemic,211900
LAMB2	107,8	100%	100%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome,609049
LCAT	101,4	97%	88%	Norum disease, 245900
LMX1B	96,8	100%	97%	Nail-patella syndrome, 161200
LRP2	104,2	99%	98%	Donnai-Barrow syndrome, 222448
LRP4	94,5	99%	97%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LYZ	102,4	100%	100%	Amyloidosis, renal, 105200
LZTFL1	85	100%	98%	Bardet-Biedl syndrome 17, 615994
MAFB	91,3	100%	100%	Multicentric carpotarsal osteolysis syndrome, 166300
MKKS	128,9	89%	89%	Bardet-Biedl syndrome 6,605231 McKusick-Kaufman syndrome, 236700
MKS1	102,4	98%	95%	Bardet-Biedl syndrome 13,615990 Meckel syndrome 1, 249000
MYH9	85,7	99%	97%	Deafness,autosomal dominant 17,603622 Epstein syndrome,153650 Fechtner syndrome,153640 Macrothrombocytopenia and progressive sensorineural deafness,600208 May-Hegglin anomaly, 155100 Sebastian syndrome,605249
MYO1E	88,7	99%	98%	Glomerulosclerosis, focal segmental, 6, 614131
NEK1	119,4	99%	99%	Short rib-polydactyly syndrome, type IIA, 263520
NEK8	104,7	100%	100%	?Nephronophthisis 9, 613824
NOTCH2	88	90%	87%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome,102500

NPHP1	118,9	100%	99%	Joubert syndrome 4,609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1,266900
NPHP3	105	100%	99%	Meckel syndrome 7,267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1,208540
NPHP4	87,7	98%	93%	Nephronophthisis 4, 606966
NPHS1	80,8	99%	95%	Nephrotic syndrome, type 1, 256300
NPHS2	124,3	100%	100%	Nephrotic syndrome, type 2, 600995
NR3C2	133	99%	93%	Pseudohypoaldosteronism type I, autosomal dominant, 177735
OCRL	59,7	98%	96%	Dent disease 2,300555 Lowe syndrome, 309000
OFD1	36,6	89%	76%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome type 2,300209
PAX2	108,9	94%	94%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830
PCBD1	58,6	100%	86%	Hyperphenylalaninemia, BH4-deficient, D,264070
PDSS2	92,8	100%	100%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	62,5	98%	96%	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	13,7	20%	18%	Polycystic kidney disease, adult type I, 173900
PKD2	97,1	95%	89%	Polycystic kidney disease 2, 613095
PKHD1	97,1	98%	96%	Polycystic kidney and hepatic disease, 263200
PLCE1	119,1	98%	96%	Nephrotic syndrome, type 3, 610725
PTH1R	72,6	100%	90%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk-Jansen type, 156400
PTPRO	98	98%	96%	Nephrotic syndrome, type 6, 614196
REN	94,8	100%	100%	Hyperuricemic nephropathy,familial juvenile 2,613092 Renal tubular dysgenesis,267430 [Hyperproreninemia]

RET	87,3	96%	92%	Multiple endocrine neoplasia IIA, 171400
ROBO2	104	100%	98%	Vesicoureteral reflux 2, 610878
RPGRIP1L	99,8	96%	96%	COACH syndrome,216360 Joubert syndrome 7, 611560 Meckel syndrome 5,611561
RRM2B	111,5	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075
SALL1	133,7	99%	98%	Townes-Brocks syndrome, 107480
SALL4	96,3	96%	95%	Duane-radial ray syndrome, 607323
SARS2	69,7	95%	88%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	91,1	99%	90%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	90,1	93%	89%	Bronchiectasis with or without elevated sweat chloride 2,613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	79,3	100%	98%	Bronchiectasis with or without elevated sweat chloride 1,211400 Liddle syndrome, 177200 Pseudohypoaldosteronism,type I,264350
SCNN1G	128,1	100%	99%	Bronchiectasis with or without elevated sweat chloride 3,613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I,264350
SDCCAG8	100,8	100%	99%	Senior-Loken syndrome 7, 613615
SIX1	84,2	95%	95%	Brachiootic syndrome 3, 608389 Deafness,autosomal dominant 23,605192
SIX5	42,6	94%	78%	Branchiootorenal syndrome 2, 610896
SLC12A1	136,8	99%	98%	Bartter syndrome, type 1, 601678
SLC12A3	84,9	100%	98%	Gitelman syndrome, 263800
SLC16A12	108,2	96%	92%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC22A12	84,8	100%	92%	Hypouricemia, renal, 220150
SLC26A3	111,2	100%	96%	Diarrhea 1,secretory chloride,congenital,214700
SLC2A2	115,6	100%	99%	Fanconi-Bickel syndrome,227810 {Diabetes mellitus, noninsulin-dependent},125853
SLC2A9	60,2	97%	86%	Hypouricemia,renal,2,612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC34A1	83,7	100%	97%	Fanconi renotubular syndrome 2,613388 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	70,8	96%	86%	Hypophosphatemic rickets with hypercalciuria, 241530

SLC3A1	120,1	96%	96%	Cystinuria, 220100
SLC41A1	82,8	98%	96%	No OMIM phenotype
SLC4A1	86,7	99%	95%	Ovalocytosis Renal tubular acidosis,distal,AD,179800 Renal tubular acidosis,distal,AR,611590 Spherocytosis,type 4,612653
SLC4A4	107,3	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	72,2	100%	98%	Renal glucosuria, 233100
SLC6A19	81,7	98%	94%	Hartnup disorder, 234500 Hyperglycinuria,138500 Iminoglycinuria,digenic,242600
SLC6A20	85,9	91%	87%	Hyperglycinuria, 138500
SLC7A7	88	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	73,5	100%	99%	Cystinuria, 220100
SLC9A3R1	107,4	100%	97%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SMARCAL1	116,6	99%	94%	Schimke immunoosseous dysplasia, 242900
SOX17	73,4	100%	100%	Vesicoureteral reflux 3, 613674
STRA6	71,8	100%	96%	Microphthalmia, syndromic 9, 601186
STX16	113,8	99%	98%	Pseudohypoparathyroidism, type IB, 603233
TCTN1	100	95%	94%	Joubert syndrome 13, 614173
TCTN2	85,8	100%	97%	?Meckel syndrome 8, 613885
TCTN3	98,3	100%	99%	Joubert syndrome 18,614815 Orofaciodigital syndrome IV, 258860
TMEM138	92,6	100%	100%	Joubert syndrome 16, 614465
TMEM216	65,6	100%	86%	Joubert syndrome 2, 608091 Meckel syndrome 2,603194
TMEM231	59,6	97%	87%	Joubert syndrome 20, 614970 Meckel syndrome 11,615397
TMEM237	87,8	100%	92%	Joubert syndrome 14, 614424
TMEM67	115,7	100%	99%	COACH syndrome,216360 Joubert syndrome 6,610688 Meckel syndrome 3,607361 Nephronophthisis 11,613550 {Bardet-Biedl syndrome 14,modifier of},209900

TNXB	10,5	45%	16%	Ehlers-Danlos syndrome due to tenascin X deficiency,606408 Vesicoureteral reflux 8,615963
TRIM32	106,2	100%	100%	?Bardet-Biedl syndrome 11,615988 Muscular dystrophy, limb-girdle, type 2H, 254110
TRPC6	74,8	91%	88%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	113,1	99%	97%	Hypomagnesemia 1, intestinal,602014
TSC1	86,3	99%	96%	Focal cortical dysplasia,Taylor balloon cell type,607341 Lymphangioleiomyomatosis,606690 Tuberous sclerosis-1, 191100
TSC2	80,3	99%	95%	Lymphangioleiomyomatosis,somatic,606690 Tuberous sclerosis-2, 613254
TTC21B	108,1	98%	98%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly,613819
TTC8	103,9	100%	100%	?Retinitis pigmentosa 51,613464 Bardet-Biedl syndrome 8, 615985
UMOD	73,2	97%	96%	Glomerulocystic kidney disease with hyperuricemia and isothenuria,609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2,603860
UPK3A	62	99%	92%	No OMIM phenotype
VIPAS39	109,8	100%	99%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	96,5	100%	99%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	122,7	100%	100%	?Cranioectodermal dysplasia 4,614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly,614376 Nephronophthisis 13,614377 Senior-Loken syndrome 8,616307
WDR35	112,4	100%	97%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly,614091
WDR60	106,8	99%	98%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNK1	130,9	100%	99%	Neuropathy,hereditary sensory and autonomic type II,201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	109,1	100%	97%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	122,7	92%	92%	Mullerian aplasia and hyperandrogenism,158330 SERKAL syndrome, 611812

WT1	57,7	100%	96%	Denys-Drash syndrome,194080 Frasier syndrome,136680 Meacham syndrome,136680 Mesothelioma,somatic,156240 Nephrotic syndrome, type 4,256370 Wilms tumor, type 1, 194070
XPNPEP3	115,8	97%	94%	Nephronophthisis-like nephropathy 1, 613159
ZNF423	123,1	100%	99%	Joubert syndrome 19,614844 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 : June 30th, 2015
This list is accurate for all panel versions starting with DG 2.3. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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