RENAL DISORDERS GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ACTN4	163.7	100%	99%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	123.3	96%	93%	Thrombotic thrombocytopenic purpura, familial, 274150
ADCK4	107.2	100%	100%	Nephrotic syndrome type 9, 615573
AGTR1	137.5	100%	100%	Hypertension, essential, 145500
AGXT	164.3	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHI1	145.2	97%	90%	Joubert syndrome-3, 608629
ALG8	135.4	93%	87%	Congenital disorder of glycosylation, type Ih, 608104
ALMS1	187.4	99%	99%	Alstrom syndrome, 203800
ANKS6	101.7	92%	89%	Nephronophthisis 16, 615382
ANLN	148.8	94%	89%	Focal segmental glomerulosclerosis 8,616032
AP2S1	160.8	90%	88%	Hypocalciuric hypercalcemia, familial, type III, 600740
APRT	64.2	100%	99%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	151	100%	96%	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	141.9	100%	100%	Nephrotic syndrome,type 8,615244
ARL13B	86.2	98%	91%	Joubert syndrome 8, 612291
ARL6	103.6	99%	99%	Bardet-Biedl syndrome 3, 209900 Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ATP6V0A4	122.8	100%	99%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	210	100%	100%	Renal tubular acidosis with deafness, 267300
ATXN10	158.7	100%	97%	Spinocerebellar ataxia 10, 603516
AVPR2	112.1	96%	88%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	120.9	92%	92%	Meckel syndrome 9, 614209

B9D2	133.6	100%	100%	Meckel syndrome 10, 614175
BBS1	151.9	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	178.4	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	229	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	204.3	100%	99%	Bardet-Biedl syndrome 2, 209900
BBS4	164.7	100%	96%	Bardet-Biedl syndrome 4, 209900
BBS7	109.7	95%	89%	Bardet-Biedl syndrome 7, 209900
BBS9	111	95%	94%	Bardet-Biedl syndrome 9, 209900
BCS1L	197.5	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BICC1	172.4	100%	97%	{Renal dysplasia,cystic,susceptibility to},601331
BSND	173.1	100%	100%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
CA2	167.4	92%	85%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CASR	198.8	100%	99%	Hyperparathyroidism,neonatal,239200 Hypocalcemia,autosomal dominant,601198 Hypocalciuric hypercalcemia,type I,145980 {Epilepsy idiopathic generalized,susceptibility to,8},612899
CC2D2A	133.6	99%	96%	COACH syndrome,216360 Joubert syndrome 9,612285 Meckel syndrome 6,612284
CD2AP	71.9	98%	85%	Glomerulosclerosis, focal segmental, 3, 607832
CEP164	102.5	100%	97%	Nephronophthisis 15, 614845
CEP290	65.9	81%	68%	?Bardet-Biedl syndrome 14,615991 Joubert syndrome 5,610188 Leber congenital amaurosis 10,611775 Meckel syndrome 4,611134 Senior-Loken syndrome 6,610189
CEP41	92.8	100%	96%	Joubert syndrome 15, 614464
CEP83				Nephronophthisis 18,615862

CLCN5	118.6	99%	95%	Dent disease, 300009 Hypophosphatemic rickets,300554 Nephrolithiasis,type I,310468 Proteinuria,low molecular weight,with hypercalciuric nephrocalcinosis,308990
CLCNKB	115.3	100%	95%	Bartter syndrome, type 3, 607364 Bartter syndrome,type 4b,digenic,613090
CLDN16	167.3	100%	100%	Hypomagnesemia 3, renal, 248250
CLDN19	168.5	100%	99%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	210.6	100%	100%	Hypomagnesemia 6, renal, 613882
COL4A1	100.2	98%	94%	?Retinal arteries,tortuosity of,180000 Angiopathy,hereditary,with nephropathy,aneurysms and muscle cramps,611773 Brain small vessel disease with or without ocular anomalies,607595 Porencephaly 1,175780 {Hemorrhage,intracerebral,susceptibility to},614519
COL4A3	89.5	98%	92%	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria,benign familial, 141200
COL4A4	85.5	96%	93%	Alport syndrome, autosomal recessive, 203780
COL4A5	35.5	84%	65%	Alport syndrome, 301050
COQ2	74.1	95%	93%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	161.2	99%	92%	Coenzyme Q10 deficiency, primary, 6, 614650
CRB2	126.7	99%	98%	Focal segmental glomerulosclerosis 9,616220 Ventriculomegaly with cystic kidney disease,219730
CTNS	138.3	100%	100%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic,219900 Cystinosis,ocular nonnephropathic,219750
CUBN	138.8	99%	98%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	116.5	94%	93%	Pseudohypoaldosteronism,type IIE,614496
DCDC2	157	99%	96%	?Deafness,autosomal recessive 66,610212

				Nephronophthisis 19,616217
DGKE	173.1	94%	85%	Nephrotic syndrome, type 7, 615008
DMP1	183.4	99%	96%	Hypophosphatemic rickets,AR,241520
DSTYK	153.5	100%	99%	{Congenital anomalies of kidney and urinary tract,susceptibility to},610805
DYNC2H1	90.3	91%	80%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
EGF	162.1	100%	99%	Hypomagnesemia 4, renal, 611718
EHHADH	176.4	100%	99%	?Fanconi renotubular syndrome 3,615605
EMP2	110.6	100%	99%	Nephrotic syndrome,type 10,615861
ENPP1	146.2	84%	79%	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	162.5	100%	99%	Branchiootorenal syndrome 1, with or without cataracts, 113650
FAM58A	65.4	79%	75%	STAR syndrome, 300707
FAN1	167.1	100%	100%	Interstitial nephritis,karyomegalic,614817
FGF23	128.4	100%	97%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia,tumor-induced Tumoral calcinosis,hyperphosphatemic,familial,211900
FN1	168.7	100%	99%	Glomerulopathy with fibronectin deposits 2, 601894
FRAS1	173.4	100%	100%	Fraser syndrome, 219000
FREM1	157.5	100%	99%	Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	217.1	99%	98%	Fraser syndrome, 219000
FXYD2	109.5	100%	100%	Hypomagnesemia-2, renal, 154020
GALNT3	135	93%	86%	Tumoral calcinosis, hyperphosphatemic, familial,211900

GATA3	187.9	100%	100%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GLA	50.5	98%	92%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	97.5	100%	97%	GM1-gangliosidosis, type I, 230500
GLIS2	119.5	100%	97%	Nephronophthisis 7, 611498
GLIS3	163.4	100%	100%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	171.5	100%	100%	Hypocalcemia,autosomal dominant 2,615361 Hypocalciuric hypercalcemia, type II, 145981
GSN	139.9	99%	90%	Amyloidosis, Finnish type, 105120 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HNF1B	135.4	100%	100%	Diabetes mellitus,noninsulin-dependent,125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma},144700
HPRT1	40.9	86%	61%	HPRT-related gout,300323 Lesch-Nyhan syndrome, 300322
HSD11B2	189.4	88%	85%	Apparent mineralocorticoid excess, 218030
IFT122	169.2	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT140	125.2	100%	99%	Mainzer-Saldino syndrome, 266920
IFT172	133.5	99%	98%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	124.7	100%	100%	Cranioectodermal dysplasia 3, 614099
INF2	106.9	94%	92%	Charcot-Marie-Tooth disease,dominant intermediate E,614455 Glomerulosclerosis, focal segmental, 5, 613237
INPP5E	108.4	99%	92%	Joubert syndrome 1,213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	163.3	100%	100%	Nephronophthisis 2, infantile, 602088
IQCB1	94.7	82%	69%	Senior-Loken syndrome 5, 609254
ITGA8	128.3	99%	97%	Renal hypodysplasia/aplasia 1, 191830
JAG1	180	99%	97%	Alagille syndrome, 118450
KAL1	74.8	87%	85%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700

KCNJ1	279.2	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	265.2	100%	100%	SESAME syndrome, 612780
KIF7	89.4	96%	87%	?Al-Gazali-Bakalinova syndrome,607131 ?Hydrolethalus syndrome 2,614120 Acrocallosal syndrome,200990 Joubert syndrome 12,200990
KL	192	98%	96%	Tumoral calcinosis, hyperphosphatemic,211900
KLHL3	169.1	99%	99%	Pseudohypoaldosteronism,type IID,614495
LAMB2	241.7	100%	100%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome,609049
LCAT	175.1	100%	98%	Norum disease, 245900
LMX1B	133.8	100%	99%	Nail-patella syndrome, 161200
LRP2	212.6	100%	99%	Donnai-Barrow syndrome, 222448
LRP4	201.9	99%	98%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305
LYZ	182.9	100%	100%	Amyloidosis, renal, 105200
LZTFL1	124.9	92%	88%	Bardet-Biedl syndrome 17, 615994
MAFB	119	100%	99%	Multicentric carpotarsal osteolysis syndrome, 166300
MKKS	239.5	89%	89%	Bardet-Biedl syndrome 6,605231 McKusick-Kaufman syndrome, 236700
MKS1	113.1	100%	100%	Bardet-Biedl syndrome 13,615990 Meckel syndrome 1, 249000
MYH9	152.2	99%	98%	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	148.1	97%	97%	Glomerulosclerosis, focal segmental, 6, 614131
NEK1	108.1	95%	91%	Short rib-polydactyly syndrome, type IIA, 263520
NEK8	194.8	100%	100%	?Nephronophthisis 9,613824 ?Renal-hepatic-pancreatic dysplasia 2,615415

NOTCH2	234.3	100%	100%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome,102500
NPHP1	125.8	96%	93%	Joubert syndrome 4,609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1,266900
NPHP3	116.4	97%	90%	Meckel syndrome 7,267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1,208540
NPHP4	162.2	100%	99%	Nephronophthisis 4, 606966
NPHS1	122.3	100%	98%	Nephrotic syndrome, type 1, 256300
NPHS2	127.1	99%	95%	Nephrotic syndrome, type 2, 600995
NR3C2	175	96%	95%	Pseudohypoaldosteronism type I, autosomal dominant, 177735
OCRL	91.1	98%	96%	Dent disease 2,300555 Lowe syndrome, 309000
OFD1	30.9	72%	55%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome type 2,300209
PAX2	191.4	100%	100%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330 Renal hypoplasia, isolated, 191830
PCBD1	128.9	100%	99%	Hyperphenylalaninemia, BH4-deficient, D,264070
PDSS2	119.9	94%	93%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	93.6	96%	93%	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	29.2	42%	35%	Polycystic kidney disease, adult type I, 173900
PKD2	115.9	88%	86%	Polycystic kidney disease 2, 613095
PKHD1	176	99%	99%	Polycystic kidney and hepatic disease, 263200
PLCE1	173	98%	98%	Nephrotic syndrome, type 3, 610725
PTH1R	133.1	100%	100%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk-Jansen type, 156400
PTPRO	178.5	99%	98%	Nephrotic syndrome, type 6, 614196

REN	170.5	100%	99%	Hyperuricemic nephropathy,familial juvenile 2,613092 Renal tubular dysgenesis,267430 [Hyperproreninemia]
RET	183.9	99%	98%	Central hypoventilation syndrome, congenital, 209880 Medullary thyroid carcinoma, 155240 Multiple endocrine neoplasia IIA, 171400 Multiple endocrine neoplasia IIB, 162300 Pheochromocytoma, 171300 {Hirschsprung disease, susceptibility to},142623
RMND1	131.3	99%	94%	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	166.1	97%	97%	Vesicoureteral reflux 2, 610878
RPGRIP1L	141.9	95%	90%	COACH syndrome,216360 Joubert syndrome 7, 611560 Meckel syndrome 5,611561
RRM2B	116.5	100%	85%	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,613077
SALL1	157.2	100%	95%	Townes-Brocks syndrome, 107480
SALL4	160.2	97%	95%	Duane-radial ray syndrome, 607323
SARS2	115.2	96%	95%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	132.5	100%	100%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	152.5	96%	95%	Bronchiectasis with or without elevated sweat chloride 2,613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	181.7	100%	100%	Bronchiectasis with or without elevated sweat chloride 1,211400 Liddle syndrome, 177200 Pseudohypoaldosteronism,type I,264350

SCNN1G	153.4	99%	96%	Bronchiectasis with or without elevated sweat chloride 3,613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I,264350
SDCCAG8	129.7	99%	93%	Senior-Loken syndrome 7, 613615
SIX1	124.9	100%	99%	Brachiootic syndrome 3, 608389 Deafness,autosomal dominant 23,605192
SIX5	44.5	90%	76%	Branchiootorenal syndrome 2, 610896
SLC12A1	191.9	100%	98%	Bartter syndrome, type 1, 601678
SLC12A3	161.7	100%	100%	Gitelman syndrome, 263800
SLC16A12	190.4	100%	99%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC22A12	128.7	100%	100%	Hypouricemia, renal, 220150
SLC26A3	189.1	99%	96%	Diarrhea 1,secretory chloride,congenital,214700
SLC2A2	193.5	100%	100%	Fanconi-Bickel syndrome,227810 {Diabetes mellitus, noninsulin-dependent},125853
SLC2A9	145.8	99%	96%	Hypouricemia,renal,2,612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC34A1	173.6	100%	100%	Fanconi renotubular syndrome 2,613388 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	121.2	100%	94%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC3A1	191	99%	98%	Cystinuria, 220100
SLC41A1	161.3	100%	100%	No OMIM disease
SLC4A1	161.3	100%	100%	Ovalocytosis Renal tubular acidosis,distal,AD,179800 Renal tubular acidosis,distal,AR,611590 Spherocytosis,type 4,612653
SLC4A4	148.4	99%	96%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	138	100%	100%	Renal glucosuria, 233100
SLC6A19	206.2	100%	99%	Hartnup disorder, 234500 Hyperglycinuria,138500 Iminoglycinuria,digenic,242600
SLC6A20	214.4	100%	99%	Hyperglycinuria, 138500

SLC7A7	124.3	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	160	100%	98%	Cystinuria, 220100
SLC9A3R1	131.6	100%	98%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SMARCAL1	154.2	100%	100%	Schimke immunoosseous dysplasia, 242900
SOX17	68.5	99%	92%	Vesicoureteral reflux 3, 613674
STRA6	134.2	100%	100%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STX16	164.9	100%	95%	Pseudohypoparathyroidism, type IB, 603233
TCTN1	113	97%	91%	Joubert syndrome 13, 614173
TCTN2	163.9	95%	92%	?Meckel syndrome 8, 613885
TCTN3	121.6	98%	98%	Joubert syndrome 18,614815 Orofaciodigital syndrome IV, 258860
TMEM138	150.4	100%	100%	Joubert syndrome 16, 614465
TMEM216	159.4	100%	98%	Joubert syndrome 2, 608091 Meckel syndrome 2,603194
TMEM231	119.8	100%	100%	Joubert syndrome 20, 614970 Meckel syndrome 11,615397
TMEM237	89.7	97%	96%	Joubert syndrome 14, 614424
TMEM67	69.2	90%	78%	COACH syndrome,216360 Joubert syndrome 6,610688 Meckel syndrome 3,607361 Nephronophthisis 11,613550 {Bardet-Biedl syndrome 14,modifier of},209900
TNXB	17.7	59%	29%	Ehlers-Danlos syndrome due to tenascin X deficiency,606408 Vesicoureteral reflux 8,615963
TRIM32	161.9	100%	100%	Bardet-Biedl syndrome 11, 209900 Muscular dystrophy, limb-girdle, type 2H, 254110
TRPC6	131.8	95%	90%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	169.5	98%	98%	Hypomagnesemia 1, intestinal,602014
TSC1	151.9	99%	98%	Focal cortical dysplasia, Taylor balloon cell type, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100

TSC2	151.8	99%	97%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TTC21B	94.2	99%	95%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly,613819
TTC8	87.4	97%	91%	?Retinitis pigmentosa 51,613464 Bardet-Biedl syndrome 8, 615985
UMOD	157.1	97%	97%	Glomerulocystic kidney disease with hyperuricemia and isothenuria,609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2,603860
UPK3A	142.1	100%	94%	No OMIM disease
VIPAS39	158.9	100%	99%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	158.1	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	147	99%	95%	?Cranioectodermal dysplasia 4,614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly,614376 Nephronophthisis 13,614377 Senior-Loken syndrome 8,616307
WDR35	152.7	97%	94%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly,614091
WDR60	121.5	97%	96%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNK1	202.1	99%	99%	Neuropathy,hereditary sensory and autonomic type II,201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	135.4	100%	99%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	352.5	92%	92%	Mullerian aplasia and hyperandrogenism,158330 SERKAL syndrome, 611812
WT1	110.2	96%	86%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370

				Wilms tumor, type 1, 194070
XPNPEP3	143.9	97%	97%	Nephronophthisis-like nephropathy 1, 613159
ZNF423	338.6	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 : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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