RENAL DISORDERS GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACTN4	161.1	100%	99%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	119.3	96%	93%	Thrombotic thrombocytopenic purpura, familial, 274150
ADCK4	104.6	100%	99%	Nephrotic syndrome, type 9, 615573
ADCY10	181	100%	99%	{Hypercalciuria, absorptive, susceptibility to}, 143870
AGTR1	187.9	100%	100%	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	168.5	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHI1	176.9	99%	98%	Joubert syndrome-3, 608629
ALG8	147.2	96%	95%	Congenital disorder of glycosylation, type Ih, 608104
ALMS1	208.7	99%	99%	Alstrom syndrome, 203800
ANKS6	95.2	93%	90%	Nephronophthisis 16, 615382
ANLN	165.7	97%	95%	Focal segmental glomerulosclerosis 8, 616032
AP2S1	145.7	90%	89%	Hypocalciuric hypercalcemia, familial, type III, 600740
APRT	73.3	100%	99%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	132.9	99%	97%	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	174.4	100%	100%	Nephrotic syndrome, type 8, 615244
ARL13B	117.5	99%	97%	Joubert syndrome 8, 612291
ARL6	121.4	100%	97%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ATP6V0A4	139.7	100%	99%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	197.9	100%	100%	Renal tubular acidosis with deafness, 267300
ATXN10	158.1	99%	97%	Spinocerebellar ataxia 10, 603516
AVPR2	149.9	99%	96%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	127.2	92%	91%	?Meckel syndrome 9, 614209
B9D2	126.9	100%	100%	Meckel syndrome 10, 614175
BBS1	178.6	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	195.4	100%	99%	Bardet-Biedl syndrome 10, 615987

BBS12	237	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	201.1	100%	99%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	153.5	99%	98%	Bardet-Biedl syndrome 4, 615982
BBS7	156.1	99%	95%	Bardet-Biedl syndrome 7, 615984
BBS9	139.9	97%	95%	Bardet-Biedl syndrome 9, 615986
BCS1L	199	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BICC1	175.4	99%	99%	{Renal dysplasia, cystic, susceptibility to}, 601331
BSND	180.6	100%	100%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
C3	167.5	100%	99%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
CA2	178.8	99%	97%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CASR	192.1	99%	99%	Hypercalciuric hypercalcemia Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Calcium, serum level of} {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CC2D2A	144.7	99%	97%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC41	126.7	99%	96%	Nephronophthisis 18, 615862
CD2AP	115.6	99%	97%	Glomerulosclerosis, focal segmental, 3, 607832
CD46	157.5	99%	95%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CEP164	100.3	99%	97%	Nephronophthisis 15, 614845
CEP290	92.8	95%	87%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189

				?Bardet-Biedl syndrome 14, 615991
CEP41	98.2	98%	93%	Joubert syndrome 15, 614464
CFB	23	84%	51%	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	216.6	99%	97%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	202.7	92%	90%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	101.2	88%	83%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFI	198	98%	97%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CLCN5	149.8	99%	98%	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCNKB	113.9	99%	94%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN16	171.5	100%	100%	Hypomagnesemia 3, renal, 248250
CLDN19	144.2	99%	96%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	228.4	100%	99%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
COL4A1	103.5	98%	94%	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	104.5	98%	95%	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200
COL4A4	97.9	98%	95%	Alport syndrome, autosomal recessive, 203780

				Hematuria, familial benign
COL4A5	61	93%	81%	Alport syndrome, 301050
COQ2	92.8	96%	93%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ6	155.4	99%	97%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	183.6	99%	99%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ9	94.2	99%	96%	Coenzyme Q10 deficiency, primary, 5, 614654
CRB2	116.5	99%	97%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CTNS	142.7	100%	99%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CUBN	134	99%	98%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	149.7	99%	97%	Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	194.4	100%	99%	Hypercalcemia, infantile, 143880
DCDC2	180	100%	99%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DGKE	160	99%	97%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DMP1	151.2	99%	99%	Hypophosphatemic rickets, AR, 241520
DSTYK	145.6	99%	99%	{Congenital anomalies of kidney and urinary tract, susceptibility to}, 610805
DYNC2H1	110.6	98%	91%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
EGF	165.2	100%	99%	Hypomagnesemia 4, renal, 611718
EHHADH	188.7	100%	99%	?Fanconi renotubular syndrome 3, 615605
EMP2	101.9	100%	99%	Nephrotic syndrome, type 10, 615861
ENPP1	174.9	94%	88%	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	167.2	100%	99%	Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650

				?Otofaciocervical syndrome, 166780
FAM20A	115.2	98%	94%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM58A	88.6	82%	78%	STAR syndrome, 300707
FAN1	168.9	100%	99%	Interstitial nephritis, karyomegalic, 614817
FAT1	236.8	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 (2015) Kidney Int 89,476) ?Autism (Neale (2012) Nature 485,242)
FGF23	122.5	99%	98%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FN1	156.3	100%	99%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101
FRAS1	168.5	100%	99%	Fraser syndrome, 219000
FREM1	149.2	99%	99%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	194.3	99%	99%	Fraser syndrome, 219000
FXYD2	104.4	100%	99%	Hypomagnesemia 2, renal, 154020
GALNT3	166.7	99%	98%	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GATA3	205.9	100%	100%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GLA	83.5	99%	97%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	97.3	99%	97%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLIS2	108.7	99%	97%	Nephronophthisis 7, 611498
GLIS3	147.4	100%	99%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	185.8	100%	99%	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GRHPR	118	86%	81%	Hyperoxaluria, primary, type II, 260000
GSN	141.9	97%	92%	Amyloidosis, Finnish type, 105120

HNF1B	137.9	99%	97%	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	159.4	100%	99%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HOGA1	156.5	99%	98%	Hyperoxaluria, primary, type III, 613616
HPRT1	82.8	96%	89%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	179.5	88%	84%	Apparent mineralocorticoid excess, 218030
IFT122	165.7	100%	99%	Cranioectodermal dysplasia 1, 218330
IFT140	124.5	99%	98%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	125.7	100%	99%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	128	100%	100%	Cranioectodermal dysplasia 3, 614099
INF2	110.1	93%	91%	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
INPP5E	109	97%	92%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	166	100%	100%	Nephronophthisis 2, infantile, 602088
IQCB1	122	92%	81%	Senior-Loken syndrome 5, 609254
ITGA8	142.8	99%	99%	Renal hypodysplasia/aplasia 1, 191830
JAG1	160.7	99%	98%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
KAL1	102.2	89%	87%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KCNJ1	235	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	219.9	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KIF7	95.3	95%	89%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KL	185.9	97%	96%	Tumoral calcinosis, hyperphosphatemic, 211900 {Coronary artery disease, susceptibility to}

KLHL3	156.5	99%	99%	Pseudohypoaldosteronism, type IID, 614495
LAMB2	220.8	100%	99%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCAT	166.1	99%	96%	Fish-eye disease, 136120 Norum disease, 245900
LMX1B	131.5	99%	96%	Nail-patella syndrome, 161200
LRP2	205.6	100%	99%	Donnai-Barrow syndrome, 222448
LRP4	184.3	99%	98%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305 ?Myasthenic syndrome, congenital, 17, 616304
LYZ	187	100%	100%	Amyloidosis, renal, 105200
LZTFL1	142	99%	98%	Bardet-Biedl syndrome 17, 615994
MAFB	125.4	99%	99%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	84.1	99%	96%	Bartter syndrome, type 5,antenatal,transient,300971
MKKS	216.9	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	114.6	99%	99%	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	146.8	99%	97%	Glomerulosclerosis, focal segmental, 6, 614131
NEK1	142.4	99%	96%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	192.1	100%	100%	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NOTCH2	180.3	100%	99%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP1	154.6	99%	98%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	146	99%	97%	Meckel syndrome 7, 267010

				Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	152.6	100%	99%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	114.9	99%	98%	Nephrotic syndrome, type 1, 256300
NPHS2	111	99%	96%	Nephrotic syndrome, type 2, 600995
NR3C2	176.6	99%	97%	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NUP107	147	99%	97%	Nephrotic syndrome, type 11, 616730
NUP205	170.2	99%	98%	?Nephrotic syndrome,type 13,616893
NUP93	154.5	97%	95%	Nephrotic syndrome, type 12,616892
OCRL	140.5	99%	98%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	59.2	87%	75%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
PAX2	200.3	100%	99%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PCBD1	129.9	99%	99%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDSS2	146.3	98%	95%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	138.1	98%	97%	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	30.1	41%	33%	Polycystic kidney disease, adult type I, 173900
PKD2	129.6	92%	88%	Polycystic kidney disease 2, 613095
PKHD1	176.8	100%	99%	Polycystic kidney and hepatic disease, 263200
PLCE1	166.9	99%	98%	Nephrotic syndrome, type 3, 610725
PTH1R	118.4	99%	98%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPRO	175	100%	99%	Nephrotic syndrome, type 6, 614196
REN	163.2	100%	99%	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia]

RET	169	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, 1}, 142623
RMND1	168.4	99%	98%	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	167.1	99%	97%	Vesicoureteral reflux 2, 610878
RPGRIP1L	160.8	96%	95%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RRM2B	163	99%	98%	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	144	99%	98%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome, 107480
SALL4	161.4	98%	96%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SARS2	120.4	96%	95%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	141.5	100%	99%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	147.9	97%	95%	Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	172.3	100%	99%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	160.7	99%	97%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SDCCAG8	155	99%	98%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEC61A1	165.8	100%	100%	Hyperuricemic nephropathy, familial juvenile, 4,617056
SIX1	125.4	99%	96%	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX5	57.7	93%	85%	Branchiootorenal syndrome 2, 610896
SLC12A1	185.9	100%	99%	Bartter syndrome, type 1, 601678

SLC12A3	164.5	100%	100%	Gitelman syndrome, 263800
SLC16A12	179.4	100%	100%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC22A12	127.3	100%	99%	Hypouricemia, renal, 220150
SLC26A3	198.1	100%	99%	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	205.9	100%	99%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	145.8	99%	98%	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC34A1	163.1	100%	99%	Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	120.6	99%	96%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC3A1	205.3	100%	99%	Cystinuria, 220100
SLC41A1	157.2	100%	99%	No OMIM phenotype Nephrolithiasis-like phenotype (Hurd (2013) J Am Soc Nephrol 24,967) ?Parkinson disease (Yan (2011) Int J Neurosci 121,632)
SLC4A1	154.1	100%	99%	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	159.1	99%	99%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	139.8	100%	99%	Renal glucosuria, 233100
SLC6A19	177.6	99%	99%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A20	189.1	100%	99%	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC7A7	114.5	100%	99%	Lysinuric protein intolerance, 222700

SLC7A9	144.1	99%	99%	Cystinuria, 220100
SLC9A3	173.3	99%	97%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	134.2	100%	99%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SMARCAL1	153.8	100%	99%	Schimke immunoosseous dysplasia, 242900
SOX17	83.7	99%	93%	Vesicoureteral reflux 3, 613674
STRA6	120.5	100%	99%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STX16	153.7	99%	98%	Pseudohypoparathyroidism, type IB, 603233
TCTN1	116.8	96%	93%	Joubert syndrome 13, 614173
TCTN2	166.2	99%	97%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	133.6	100%	99%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
THBD	129.5	99%	98%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TMEM138	128	100%	99%	Joubert syndrome 16, 614465
TMEM216	147.1	99%	99%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	105.5	99%	98%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	126	99%	98%	Joubert syndrome 14, 614424
TMEM67	93.3	95%	89%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TNXB	17.8	58%	32%	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963
TRIM32	147.3	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRPC6	127.5	98%	96%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	166.1	100%	99%	Hypomagnesemia 1, intestinal, 602014
TSC1	140.4	99%	97%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100

TSC2	150.1	99%	99%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TTC21B	132.4	99%	98%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC8	118.3	99%	98%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
UMOD	131.5	97%	96%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UPK3A	147.3	99%	97%	No OMIM phenotype Renal hypodysplasia (Schonfelder (2006) Am J Kidney Dis 47, 1004) Renal aysplasia (Jenkins (2005) J Am Soc Nephrol 16, 2141)
VDR	124.6	99%	96%	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710
VIPAS39	156.9	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	142.9	100%	99%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	170	100%	99%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	186.3	99%	98%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	131.2	99%	97%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNK1	178.6	99%	99%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	142.3	99%	98%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	284.8	93%	92%	Mullerian aplasia and hyperandrogenism, 158330 SERKAL syndrome, 611812
WT1	94.2	96%	89%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
XDH	118.7	100%	99%	Xanthinuria, type I, 278300

XPNPEP3	142.9	98%	97%	Nephronophthisis-like nephropathy 1, 613159
ZNF423	268.6	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 14th 2017
This list is accurate for panel version DG 2.9

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