RENAL DISORDERS
GENE PANEL DG 2.11 (242 genes)

Gene	*Median*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACTN4	143.9	99	97	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	100	96	91	Thrombotic thrombocytopenic purpura, familial, 274150
ADCK4	90.6	100	99	Nephrotic syndrome, type 9, 615573
ADCY10	149.5	100	99	{Hypercalciuria, absorptive, susceptibility to}, 143870
AGTR1	149.9	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	139.7	99	99	Hyperoxaluria, primary, type 1, 259900
AHI1	139.1	99	95	Joubert syndrome-3, 608629
ALG8	126	96	95	Congenital disorder of glycosylation, type Ih, 608104
ALMS1	179.4	99	99	Alstrom syndrome, 203800
ANKS6	91.8	92	88	Nephronophthisis 16, 615382
ANLN	146.1	97	93	Focal segmental glomerulosclerosis 8, 616032
AP2S1	116.1	90	89	Hypocalciuric hypercalcemia, familial, type III, 600740
APOL1	188	100	100	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
APRT	68.4	100	98	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	117.5	99	95	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	143.9	100	99	Nephrotic syndrome, type 8, 615244
ARL13B	97.3	98	92	Joubert syndrome 8, 612291
ARL6	85.2	99	95	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ATP6V0A4	116.6	99	98	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	176.8	100	100	Renal tubular acidosis with deafness, 267300
AVPR2	128.6	99	97	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	115.4	92	91	?Meckel syndrome 9, 614209

B9D2	111	100	100	Meckel syndrome 10, 614175
BBS1	149.1	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	172.3	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	208.7	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	181.9	100	99	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	135.2	99	97	Bardet-Biedl syndrome 4, 615982
BBS7	120.5	98	91	Bardet-Biedl syndrome 7, 615984
BBS9	112.5	96	93	Bardet-Biedl syndrome 9, 615986
BCS1L	182.6	100	100	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BICC1	152.4	100	99	{Renal dysplasia, cystic, susceptibility to}, 601331
BSND	137.5	100	100	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
C3	145.8	100	99	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
CA2	140.5	100	99	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CASR	178	100	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	127.4	99	97	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC41	96.9	98	89	Nephronophthisis 18, 615862
CD2AP	98.1	99	96	Glomerulosclerosis, focal segmental, 3, 607832
CD46	115.2	97	93	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CEP164	94.4	99	97	Nephronophthisis 15, 614845

CEP290	66	88	76	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	83.6	97	89	Joubert syndrome 15, 614464
CEP55	129.5	100	99	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly ,236500
CFB	33.4	23	23	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	183.1	98	95	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	234.8	95	94	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	103	92	87	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFI	152.5	98	96	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CLCN5	135.4	99	98	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCNKB	100.1	98	90	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN10	149.1	100	100	No OMIM phenotype {Ulcerative colitis, susceptibility to} (Saadati (2016) BMC Med Genet 17,26)
CLDN16	136.3	100	99	Hypomagnesemia 3, renal, 248250
CLDN19	123.8	98	93	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	188.5	100	99	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418

COL4A3	89.5	97	95	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200
COL4A4	85	97	93	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign
COL4A5	52.6	92	77	Alport syndrome, 301050
COQ2	89.2	96	93	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ6	144.1	99	95	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	158.7	99	98	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ9	91.6	99	96	Coenzyme Q10 deficiency, primary, 5, 614654
CRB2	111.4	99	94	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CTNS	120.3	100	99	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CUBN	127.9	99	98	Megaloblastic anemia-1, Finnish type, 261100
CUL3	110.1	98	94	Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	165.3	100	100	Hypercalcemia, infantile, 143880
DCDC2	150.6	99	99	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DGKE	142.3	99	95	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DMP1	159.9	99	99	Hypophosphatemic rickets, AR, 241520
DSTYK	146.4	100	99	{Congenital anomalies of kidney and urinary tract, susceptibility to}, 610805
DYNC2H1	90.4	96	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DZIP1L	98.3	99	95	Polycystic kidney disease 5, 617610
EGF	135.2	100	99	Hypomagnesemia 4, renal, 611718
EHHADH	163.2	100	99	?Fanconi renotubular syndrome 3, 615605
EMP2	101.3	99	96	Nephrotic syndrome, type 10, 615861

ENPP1	134.9	92	83	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	144.4	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	104.8	98	91	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM58A	74.2	83	79	STAR syndrome, 300707
FAN1	144.1	100	99	Interstitial nephritis, karyomegalic, 614817
FAT1	204.9	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	106.4	99	97	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FN1	145.5	100	99	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101
FRAS1	147.7	100	99	Fraser syndrome, 219000
FREM1	138.4	99	99	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	182.5	100	99	Fraser syndrome, 219000
FXYD2	96.9	99	99	Hypomagnesemia 2, renal, 154020
GALNT3	128	99	96	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GANAB	120.3	99	98	Polycystic kidney disease 3, 600666
GATA3	187.1	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GLA	82	99	97	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLIS2	109.2	100	98	Nephronophthisis 7, 611498
GLIS3	133.5	99	99	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199

GNA11	150	99	96	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GRHPR	112.8	85	78	Hyperoxaluria, primary, type II, 260000
GSN	120.7	97	91	Amyloidosis, Finnish type, 105120
HNF1B	123.8	99	99	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	142.1	99	99	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HOGA1	147.7	99	98	Hyperoxaluria, primary, type III, 613616
HPRT1	58.6	96	84	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	165.4	85	82	Apparent mineralocorticoid excess, 218030
IFNG	130.5	100	99	{AIDS, rapid progression to}, 609423 {Aplastic anemia}, 609135 {Hepatitis C virus, response to therapy of}, 609532 {TSC2 angiomyolipomas, renal, modifier of}, 613254 {Tuberculosis, protection against}, 607948
IFT122	152.3	100	99	Cranioectodermal dysplasia 1, 218330
IFT140	115	100	99	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	114.8	100	100	Cranioectodermal dysplasia 3, 614099
INF2	88.2	93	90	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
INPP5E	89.3	95	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INTU	123.8	99	96	No OMIM phenotype
INVS	159.6	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92.2	89	75	Senior-Loken syndrome 5, 609254
ITGA3	142.8	99	98	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA8	122.6	99	98	Renal hypodysplasia/aplasia 1, 191830

JAG1	148.6	98	97	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
KAL1	90.9	89	87	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KCNJ1	233.8	100	100	Bartter syndrome, type 2, 241200
KCNJ10	238.8	100	99	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KIAA0556	134.6	99	99	?Joubert syndrome 26, 616784
KIF7	85.8	93	88	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
KL	181.3	97	96	Tumoral calcinosis, hyperphosphatemic, 211900 {Coronary artery disease, susceptibility to}
KLHL3	141.1	99	99	Pseudohypoaldosteronism, type IID, 614495
LAGE3	50.6	95	81	Galloway-Mowat syndrome 2, X-linked, 301006
LAMB2	202	100	99	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCAT	145.9	97	91	Fish-eye disease, 136120 Norum disease, 245900
LMX1B	111.5	96	92	Nail-patella syndrome, 161200
LRP2	176.4	100	99	Donnai-Barrow syndrome, 222448
LRP4	166.8	99	98	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305 ?Myasthenic syndrome, congenital, 17, 616304
LYZ	165.5	100	100	Amyloidosis, renal, 105200
LZTFL1	108.7	99	95	Bardet-Biedl syndrome 17, 615994
MAFB	93.8	99	97	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	80.6	99	97	Bartter syndrome, type 5,antenatal,transient,300971
MAGI2	98.5	91	88	Nephrotic syndrome 15, 617609
MAPKBP1	144.3	100	100	No OMIM phenotype
MKKS	225.1	89	89	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700

MKS1	115.6	99	98	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
MOCOS	169.1	99	96	Xanthinuria, type II, 603592
MYH9	130.8	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	132.1	98	97	Glomerulosclerosis, focal segmental, 6, 614131
NEK1	103	98	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	171.6	100	99	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NOTCH2	172.7	100	99	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP1	117.6	98	96	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	114.3	99	95	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	137.3	99	99	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	102.2	99	98	Nephrotic syndrome, type 1, 256300
NPHS2	117	99	95	Nephrotic syndrome, type 2, 600995
NR3C2	159.7	99	95	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NUP107	122.9	99	94	Nephrotic syndrome, type 11, 616730
NUP205	133.8	98	98	?Nephrotic syndrome,type 13,616893
NUP93	140.8	97	94	Nephrotic syndrome, type 12,616892
OCRL	123	98	96	Dent disease 2, 300555 Lowe syndrome, 309000

OFD1	53	84	68	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OSGEP	120.7	100	99	Galloway-Mowat syndrome 3, 617729
PAX2	168.6	99	99	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PCBD1	113.7	99	99	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDSS2	126.9	96	93	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	123.8	98	96	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	28.1	42	34	Polycystic kidney disease, adult type I, 173900
PKD2	110.7	89	84	Polycystic kidney disease 2, 613095
PKHD1	155	100	99	Polycystic kidney and hepatic disease, 263200
PLCE1	155.4	99	98	Nephrotic syndrome, type 3, 610725
PMM2	141.3	99	99	Congenital disorder of glycosylation, type Ia, 212065
PTH1R	108.6	99	98	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPRO	140.8	99	99	Nephrotic syndrome, type 6, 614196
REN	149.1	100	100	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia]
RMND1	137	99	97	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	155.3	98	96	Vesicoureteral reflux 2, 610878
RPGRIP1L	126.2	96	93	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RRM2B	128.8	99	97	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	138.8	99	98	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome, 107480

SALL4	147.6	97	96	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SARS2	106.6	95	93	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	120.6	100	99	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	130.9	96	93	Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	149	100	99	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	139.6	99	97	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SDCCAG8	124.4	99	97	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEC61A1	133.2	100	100	Hyperuricemic nephropathy, familial juvenile, 4,617056
SIX1	117.3	99	97	Brachiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX5	43.8	88	76	Branchiootorenal syndrome 2, 610896
SLC12A1	172.7	99	99	Bartter syndrome, type 1, 601678
SLC12A3	139.5	100	100	Gitelman syndrome, 263800
SLC16A12	164.4	100	99	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC22A12	105.9	100	99	Hypouricemia, renal, 220150
SLC26A1	140	100	99	?Nephrolithiasis,calcium oxalate,167030
SLC26A3	156.2	99	98	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	178.3	100	99	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	119	99	96	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC34A1	153.5	100	99	Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	105.8	98	94	Hypophosphatemic rickets with hypercalciuria, 241530
SLC3A1	162.9	100	99	Cystinuria, 220100

SLC41A1	150.2	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	142.3	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	122.1	99	97	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	118.9	100	100	Renal glucosuria, 233100
SLC6A19	149.5	100	99	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A20	178.9	100	99	Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC7A7	124	100	99	Lysinuric protein intolerance, 222700
SLC7A9	125.5	99	99	Cystinuria, 220100
SLC9A3	147.5	98	96	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	111.6	99	96	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SMARCAL1	134.9	100	99	Schimke immunoosseous dysplasia, 242900
SOX17	70.9	99	94	Vesicoureteral reflux 3, 613674
STRA6	116.6	100	99	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STX16	138.2	99	97	Pseudohypoparathyroidism, type IB, 603233
TCTN1	100.5	96	94	Joubert syndrome 13, 614173
TCTN2	144.2	99	97	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	127.5	100	99	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860

THBD	108.4	99	97	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TMEM107	164.2	100	100	?Joubert syndrome 29 , 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM138	100.3	100	99	Joubert syndrome 16, 614465
TMEM216	112	100	98	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	111.8	100	99	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100.6	99	98	Joubert syndrome 14, 614424
TMEM67	72.9	93	83	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TNXB	19.7	19	18	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963
TP53RK	37.2	91	76	Galloway-Mowat syndrome 4, 617730
TPRKB	56.6	79	66	Galloway-Mowat syndrome 5, 617731
TRAF3IP1	90.2	96	92	Senior-Loken syndrome 9, 616629
TRIM32	141.3	100	100	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRPC6	103.7	99	96	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	150.6	99	98	Hypomagnesemia 1, intestinal, 602014
TSC1	128.9	99	98	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	131.3	100	99	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TTC21B	100.6	99	97	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC8	106.8	97	92	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464

UMOD	127.4	97	97	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UPK3A	110.7	99	96	No OMIM phenotype Renal hypodysplasia (Schonfelder (2006) Am J Kidney Dis 47, 1004) Renal aysplasia (Jenkins (2005) J Am Soc Nephrol 16, 2141)
VDR	121.1	98	95	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710
VIPAS39	144.7	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	138.4	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	132	99	98	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	145.1	99	97	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	114.2	99	96	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNK1	167.2	99	99	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	123.2	99	98	Pseudohypoaldosteronism, type IIB, 614491
WNT4	263.6	93	92	Mullerian aplasia and hyperandrogenism, 158330 SERKAL syndrome, 611812
WT1	85.3	93	86	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
XDH	109.2	100	99	Xanthinuria, type I, 278300
XPNPEP3	134.8	98	97	Nephronophthisis-like nephropathy 1, 613159
ZNF423	251.1	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 2.11

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