RENAL DISORDERS GENE PANEL DG 2.7/DG 2.8

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACTN4	151.8	99%	99%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	113.7	96%	92%	Thrombotic thrombocytopenic purpura, familial, 274150
ADCK4	106.9	100%	99%	Nephrotic syndrome, type 9, 615573
ADCY10	172.5	100%	99%	{Hypercalciuria, absorptive, susceptibility to}, 143870
AGTR1	159.8	100%	99%	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	149.5	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHI1	151.9	98%	94%	Joubert syndrome-3, 608629
ALG8	153.6	96%	93%	Congenital disorder of glycosylation, type Ih, 608104
ALMS1	197.7	99%	99%	Alstrom syndrome, 203800
ANKS6	99.5	93%	89%	Nephronophthisis 16, 615382
ANLN	156.4	96%	92%	Focal segmental glomerulosclerosis 8, 616032
AP2S1	154.3	90%	88%	Hypocalciuric hypercalcemia, familial, type III, 600740
APRT	68.4	99%	99%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	125.3	98%	93%	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	158.4	100%	99%	Nephrotic syndrome, type 8, 615244
ARL13B	100.4	99%	92%	Joubert syndrome 8, 612291
ARL6	99	97%	91%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ATP6V0A4	133.4	99%	99%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	196.6	100%	99%	Renal tubular acidosis with deafness, 267300
ATXN10	166.4	99%	96%	Spinocerebellar ataxia 10, 603516
AVPR2	154.9	98%	95%	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	119.3	92%	91%	?Meckel syndrome 9, 614209
B9D2	123.3	100%	100%	Meckel syndrome 10, 614175
BBS1	162.3	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	179.1	100%	99%	Bardet-Biedl syndrome 10, 615987

BBS12	225	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	210.1	100%	99%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	163.9	99%	97%	Bardet-Biedl syndrome 4, 615982
BBS7	135.5	97%	92%	Bardet-Biedl syndrome 7, 615984
BBS9	124.1	96%	93%	Bardet-Biedl syndrome 9, 615986
BCS1L	184.4	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BICC1	177.2	99%	99%	{Renal dysplasia, cystic, susceptibility to}, 601331
BSND	164.9	100%	100%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
C3	170.8	100%	99%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
CA2	166.3	98%	93%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CASR	181.3	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	137.5	98%	96%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC41	113.4	97%	89%	Nephronophthisis 18, 615862
CD2AP	98.1	98%	94%	Glomerulosclerosis, focal segmental, 3, 607832
CD46	140.5	97%	92%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CEP164	98.4	99%	96%	Nephronophthisis 15, 614845
CEP290	77.4	88%	77%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134

				Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	96.7	97%	90%	Joubert syndrome 15, 614464
CFB	21.3	81%	45%	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	193.2	98%	96%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	207	92%	90%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	107.1	85%	80%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFI	174.8	97%	96%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CLCN5	162.6	99%	97%	Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468 Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990
CLCNKB	106.8	98%	92%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN16	161.4	100%	99%	Hypomagnesemia 3, renal, 248250
CLDN19	143.1	98%	95%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	213.2	99%	99%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
COL4A1	101.8	98%	93%	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	96.3	97%	94%	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200

COL4A4	91.9	97%	93%	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign
COL4A5	59.6	91%	77%	Alport syndrome, 301050
COQ2	84.5	95%	92%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ6	154.8	98%	96%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	188.1	99%	98%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ9	105.7	99%	98%	Coenzyme Q10 deficiency, primary, 5, 614654
CRB2	111.2	99%	96%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CTNS	138.1	100%	100%	Cystinosis, atypical nephropathic, 219800 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750
CUBN	141.9	99%	98%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	128.6	98%	94%	Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	191.1	99%	99%	Hypercalcemia, infantile, 143880
DCDC2	160	99%	99%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DGKE	157.9	98%	93%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DMP1	168	99%	99%	Hypophosphatemic rickets, AR, 241520
DSTYK	152.4	99%	98%	{Congenital anomalies of kidney and urinary tract, susceptibility to}, 610805
DYNC2H1	102.9	95%	86%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
EGF	157.6	99%	99%	Hypomagnesemia 4, renal, 611718
EHHADH	184.4	100%	99%	?Fanconi renotubular syndrome 3, 615605
EMP2	113.6	100%	98%	Nephrotic syndrome, type 10, 615861
ENPP1	155.5	91%	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	160.1	99%	98%	Anterior segment anomalies with or without cataract, 113650 Branchiootic syndrome 1, 602588

				Branchiootorenal syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780
FAM20A	118.6	97%	92%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM58A	85.7	82%	78%	STAR syndrome, 300707
FAN1	167.4	100%	99%	Interstitial nephritis, karyomegalic, 614817
FAT1	234.3	100%	99%	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	129.9	99%	98%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FN1	158.8	99%	99%	Glomerulopathy with fibronectin deposits 2, 601894 Plasma fibronectin deficiency, 614101
FRAS1	165	100%	99%	Fraser syndrome, 219000
FREM1	156.4	99%	98%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FREM2	199.1	99%	99%	Fraser syndrome, 219000
FXYD2	105.5	100%	99%	Hypomagnesemia 2, renal, 154020
GALNT3	144.6	98%	94%	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GATA3	185.5	100%	100%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GLA	87.1	99%	97%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	93.9	99%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLIS2	106.8	99%	97%	Nephronophthisis 7, 611498
GLIS3	155	100%	99%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	173.1	100%	99%	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GRHPR	120.1	84%	79%	Hyperoxaluria, primary, type II, 260000

GSN	131.2	97%	93%	Amyloidosis, Finnish type, 105120
HNF1B	138.4	99%	99%	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	166.4	99%	99%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850 {Diabetes mellitus, noninsulin-dependent}, 125853
HOGA1	154.2	100%	97%	Hyperoxaluria, primary, type III, 613616
HPRT1	75.3	94%	84%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	166.6	87%	84%	Apparent mineralocorticoid excess, 218030
IFT122	164.4	100%	99%	Cranioectodermal dysplasia 1, 218330
IFT140	124.5	99%	98%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	128.2	99%	99%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	130.6	100%	100%	Cranioectodermal dysplasia 3, 614099
INF2	97.9	93%	90%	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
INPP5E	105.1	96%	91%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	176.4	100%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	105.9	86%	75%	Senior-Loken syndrome 5, 609254
ITGA8	141.5	99%	98%	Renal hypodysplasia/aplasia 1, 191830
JAG1	167	99%	98%	Alagille syndrome, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon
KAL1	106.4	89%	87%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KCNJ1	259.7	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	229	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KIF7	93.4	95%	88%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120

KL	186	97%	95%	Tumoral calcinosis, hyperphosphatemic, 211900 {Coronary artery disease, susceptibility to}
KLHL3	154.8	99%	99%	Pseudohypoaldosteronism, type IID, 614495
LAMB2	212.5	100%	99%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCAT	160.7	99%	96%	Fish-eye disease, 136120 Norum disease, 245900
LMX1B	130.3	99%	95%	Nail-patella syndrome, 161200
LRP2	199.9	100%	99%	Donnai-Barrow syndrome, 222448
LRP4	183.8	99%	98%	Cenani-Lenz syndactyly syndrome, 212780 Sclerosteosis 2, 614305 ?Myasthenic syndrome, congenital, 17, 616304
LYZ	192.2	100%	100%	Amyloidosis, renal, 105200
LZTFL1	136.5	98%	94%	Bardet-Biedl syndrome 17, 615994
MAFB	117.8	99%	98%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	86.6	99%	98%	Bartter syndrome, type 5,antenatal,transient,300971
MKKS	239.6	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	113.5	99%	98%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
MYH9	146.4	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	150.3	98%	96%	Glomerulosclerosis, focal segmental, 6, 614131
NEK1	124	97%	93%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	187.6	100%	99%	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NOTCH2	194.6	100%	99%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP1	141	99%	97%	Joubert syndrome 4, 609583

				Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	128.7	98%	94%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	148	99%	99%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	113.4	99%	97%	Nephrotic syndrome, type 1, 256300
NPHS2	118.5	98%	92%	Nephrotic syndrome, type 2, 600995
NR3C2	178.3	99%	97%	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 Pseudohypoaldosteronism type I, autosomal dominant, 177735
NUP107	133.5	98%	93%	Nephrotic syndrome, type 11, 616730
NUP205	152.8	99%	97%	?Nephrotic syndrome,type 13,616893
NUP93	158.1	97%	94%	Nephrotic syndrome, type 12,616892
OCRL	152.2	99%	97%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	56.1	84%	71%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
PAX2	184.8	99%	99%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PCBD1	124.7	99%	99%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDSS2	131.3	97%	93%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	145.5	98%	96%	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	28.2	42%	33%	Polycystic kidney disease, adult type I, 173900
PKD2	119.8	91%	87%	Polycystic kidney disease 2, 613095
PKHD1	173.7	99%	99%	Polycystic kidney and hepatic disease, 263200
PLCE1	171.1	99%	98%	Nephrotic syndrome, type 3, 610725
PTH1R	115.5	99%	99%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PTPRO	179.7	99%	98%	Nephrotic syndrome, type 6, 614196

REN	166.2	100%	100%	Hyperuricemic nephropathy, familial juvenile 2, 613092 Renal tubular dysgenesis, 267430 [Hyperproreninemia]
RET	163.1	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	142.9	99%	96%	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	158.8	98%	97%	Vesicoureteral reflux 2, 610878
RPGRIP1L	153.6	95%	93%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RRM2B	148.4	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	147.9	99%	98%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome, 107480
SALL4	154.6	98%	96%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SARS2	116.3	96%	95%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	138.9	99%	98%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	144.8	96%	94%	Bronchiectasis with or without elevated sweat chloride 2, 613021 Pseudohypoaldosteronism, type I, 264350
SCNN1B	167.2	100%	99%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SCNN1G	156.2	99%	97%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome, 177200 Pseudohypoaldosteronism, type I, 264350
SDCCAG8	136.3	99%	96%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEC61A1	156	99%	99%	Hyperuricemic nephropathy, familial juvenile, 4,617056
SIX1	121.1	99%	96%	Brachiootic syndrome 3, 608389

				Deafness, autosomal dominant 23, 605192
SIX5	51.6	90%	80%	Branchiootorenal syndrome 2, 610896
SLC12A1	194.5	100%	99%	Bartter syndrome, type 1, 601678
SLC12A3	157.4	100%	99%	Gitelman syndrome, 263800
SLC16A12	194.1	100%	100%	Cataract, juvenile, with microcornea and glucosuria, 612018
SLC22A12	119.8	100%	99%	Hypouricemia, renal, 220150
SLC26A3	183.9	99%	98%	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	189.5	100%	99%	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	139.5	99%	97%	Hypouricemia, renal, 2, 612076 {Uric acid concentration, serum, QTL 2}, 612076
SLC34A1	160.1	100%	99%	Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	115.2	98%	94%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC3A1	192	99%	98%	Cystinuria, 220100
SLC41A1	155	100%	99%	No OMIM phenotype
SLC4A1	154.4	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	150.3	99%	98%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	136.3	100%	99%	Renal glucosuria, 233100
SLC6A19	173.8	99%	99%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A20	190.5	99%	99%	Hyperglycinuria, 138500

				Iminoglycinuria, digenic, 242600
SLC7A7	127.2	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	152.6	100%	98%	Cystinuria, 220100
SLC9A3	178	98%	97%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	131.5	99%	98%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SMARCAL1	148.2	100%	99%	Schimke immunoosseous dysplasia, 242900
SOX17	79.2	97%	90%	Vesicoureteral reflux 3, 613674
STRA6	131	100%	100%	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STX16	168.8	99%	97%	Pseudohypoparathyroidism, type IB, 603233
TCTN1	117.4	95%	92%	Joubert syndrome 13, 614173
TCTN2	163.8	98%	95%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	135.8	99%	99%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
THBD	123	99%	97%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TMEM138	133.5	100%	99%	Joubert syndrome 16, 614465
TMEM216	159.2	100%	99%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	103.4	99%	98%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	115.6	99%	96%	Joubert syndrome 14, 614424
TMEM67	78.9	92%	83%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TNXB	17.8	59%	32%	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963
TRIM32	152.9	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRPC6	131.9	97%	94%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	172.4	99%	98%	Hypomagnesemia 1, intestinal, 602014

TSC1	149.6	99%	98%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	144.5	99%	98%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TTC21B	111.6	99%	96%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC8	109.1	98%	93%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
UMOD	130.6	97%	96%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UPK3A	129.2	99%	97%	No OMIM phenotype
VDR	134.4	99%	96%	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710
VIPAS39	163.3	100%	99%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	154	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	153.5	99%	97%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	167.2	98%	96%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	120.8	98%	95%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNK1	182.1	99%	98%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	135.7	99%	97%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	274.4	93%	92%	Mullerian aplasia and hyperandrogenism, 158330 SERKAL syndrome, 611812
WT1	100	95%	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	119.9	100%	99%	Xanthinuria, type I, 278300
XPNPEP3	148.8	98%	97%	Nephronophthisis-like nephropathy 1, 613159
ZNF423	279.5	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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 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 {} signify risk factors