RENAL DISORDERS GENE PANEL DG 2.17 (286 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACE	129.3	100.0%	99.8%	Renal tubular dysgenesis, 267430
ACTN4	143.2	100.0%	99.9%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	115.6	98.0%	96.0%	Thrombotic thrombocytopenic purpura, familial, 274150
ADAMTS9	119.6	99.5%	97.8%	No OMIM Disease ID
ADCY10	127.7	100.0%	99.8%	No OMIM disease ID
AGT	211.6	100.0%	100.0%	Renal tubular dysgenesis, 267430
AGTR1	147.7	92.0%	91.9%	Renal tubular dysgenesis, 267430
AGXT	176.8	100.0%	100.0%	Hyperoxaluria, primary, type 1, 259900
AHI1	125.5	99.9%	97.6%	Joubert syndrome 3, 608629
ALDOB	140.0	100.0%	99.1%	Fructose intolerance, hereditary, 229600
ALG8	118.5	96.8%	95.7%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	114.8	100.0%	99.8%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	178.0	100.0%	99.8%	Alstrom syndrome, 203800
AMN	118.2	99.1%	93.0%	Megaloblastic anemia-1, Norwegian type, 261100
ANKS6	101.2	99.3%	96.5%	Nephronophthisis 16, 615382
ANLN	139.3	98.6%	97.0%	Focal segmental glomerulosclerosis 8, 616032
ANOS1	78.3	91.4%	87.3%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AP2S1	120.3	90.4%	89.6%	Hypocalciuric hypercalcemia, type III, 600740
APOL1	157.3	100.0%	100.0%	No OMIM disease ID
APRT	105.5	100.0%	100.0%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	142.2	100.0%	100.0%	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	226.3	100.0%	99.9%	Nephrotic syndrome, type 8, 615244
ARL13B	98.7	100.0%	99.7%	Joubert syndrome 8, 612291
ARL6	91.8	99.9%	97.7%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ATP1A1	114.1	100.0%	99.7%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP6V0A4	108.1	100.0%	99.2%	Renal tubular acidosis, distal, autosomal recessive, 602722

ATP6V1B1	184.8	100.0%	100.0%	Renal tubular acidosis with deafness, 267300
ATP7B	137.1	99.9%	99.3%	Wilson disease, 277900
AVP	73.9	99.9%	90.5%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	150.2	100.0%	99.8%	Nephrogenic syndrome of inappropriate antidiuresis, 300539 Diabetes insipidus, nephrogenic, 304800
B9D1	111.4	92.2%	92.2%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	115.7	100.0%	100.0%	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BBIP1	116.1	98.5%	91.4%	?Bardet-Biedl syndrome 18, 615995
BBS1	156.1	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	156.7	100.0%	100.0%	Bardet-Biedl syndrome 10, 615987
BBS12	193.6	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	153.3	100.0%	99.7%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	113.2	100.0%	98.4%	Bardet-Biedl syndrome 4, 615982
BBS5	94.9	98.4%	92.3%	Bardet-Biedl syndrome 5, 615983
BBS7	136.8	99.0%	95.3%	Bardet-Biedl syndrome 7, 615984
BBS9	113.2	98.8%	94.8%	Bardet-Biedl syndrome 9, 615986
BCS1L	160.0	100.0%	100.0%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BICC1	140.2	100.0%	100.0%	No OMIM disease ID
BSND	150.8	100.0%	100.0%	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
C3	153.6	100.0%	99.7%	C3 deficiency, 613779
C5orf42	122.3	99.7%	97.4%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	141.8	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CACNA1H	157.1	99.5%	98.5%	Hyperaldosteronism, familial, type IV, 617027
CASR	167.3	100.0%	99.8%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200

CC2D2A	112.6	99.0%	97.0%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CD2AP	118.7	99.9%	98.7%	Glomerulosclerosis, focal segmental, 3, 607832
CD46	126.4	100.0%	99.1%	No OMIM disease ID
CEP120	131.3	100.0%	99.6%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	96.7	99.9%	98.4%	Nephronophthisis 15, 614845
CEP290	77.6	96.9%	88.7%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	79.1	98.7%	94.4%	Joubert syndrome 15, 614464
CEP55	123.2	100.0%	100.0%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	103.3	99.8%	96.2%	Nephronophthisis 18, 615862
CFB	125.5	100.0%	100.0%	?Complement factor B deficiency, 615561
CFH	148.8	99.4%	97.4%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	156.0	94.1%	92.0%	No OMIM disease ID
CFHR3	89.5	93.2%	91.0%	No OMIM disease ID
CFI	137.6	99.2%	96.8%	Complement factor I deficiency, 610984
CLCN5	105.8	99.7%	96.8%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468
CLCNKB	109.1	100.0%	98.3%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN10	144.8	100.0%	99.9%	HELIX syndrome, 617671
CLDN16	132.1	100.0%	100.0%	Hypomagnesemia 3, renal, 248250
CLDN19	141.2	99.8%	97.2%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	222.5	100.0%	100.0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
COL4A1	100.3	99.8%	98.0%	?Retinal arteries, tortuosity of, 180000 Brain small vessel disease with or without ocular anomalies, 175780 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564

COL4A3	93.8	99.6%	97.9%	Hematuria, benign familial, 141200 Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200
COL4A4	96.5	99.7%	98.0%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	57.3	97.4%	87.3%	Alport syndrome 1, X-linked, 301050
COQ2	107.7	97.7%	97.0%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	136.6	99.6%	97.4%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	142.8	99.9%	99.6%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8B	109.0	100.0%	99.9%	Nephrotic syndrome, type 9, 615573
COQ9	78.6	99.9%	98.5%	Coenzyme Q10 deficiency, primary, 5, 614654
CRB2	132.1	99.9%	98.7%	Ventriculomegaly with cystic kidney disease, 219730 Focal segmental glomerulosclerosis 9, 616220
CSPP1	117.4	100.0%	99.4%	Joubert syndrome 21, 615636
CTNS	118.7	100.0%	99.6%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CUBN	103.2	99.6%	98.0%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	114.0	99.9%	98.7%	Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	178.7	100.0%	100.0%	Hypercalcemia, infantile, 1, 143880
DCDC2	158.0	100.0%	99.9%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DGKE	132.2	100.0%	98.3%	Nephrotic syndrome, type 7, 615008
DMP1	135.8	100.0%	99.9%	Hypophosphatemic rickets, AR, 241520
DNAJB11	107.4	99.9%	99.4%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DSTYK	127.7	99.8%	98.0%	Spastic paraplegia 23, 270750 Congenital anomalies of kidney and urinary tract 1, 610805
DYNC2H1	98.0	98.9%	94.3%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DZIP1L	103.1	100.0%	98.4%	Polycystic kidney disease 5, 617610
EGF	111.5	100.0%	99.8%	Hypomagnesemia 4, renal, 611718
EHHADH	138.8	100.0%	99.9%	?Fanconi renotubular syndrome 3, 615605
EMP2	79.3	99.8%	96.9%	Nephrotic syndrome, type 10, 615861
ENPP1	128.6	97.9%	92.4%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 Arterial calcification, generalized, of infancy, 1, 208000

EYA1	121.6	100.0%	99.9%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Branchiootic syndrome 1, 602588
FAH	136.7	100.0%	99.8%	Tyrosinemia, type I, 276700
FAM20A	122.6	99.9%	99.2%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM58A	57.8	83.7%	78.2%	STAR syndrome, 300707
FAN1	136.8	100.0%	99.9%	Interstitial nephritis, karyomegalic, 614817
FAT1	162.9	100.0%	100.0%	No OMIM Disease ID
FGF23	130.1	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FN1	109.6	100.0%	99.3%	Glomerulopathy with fibronectin deposits 2, 601894 Spondylometaphyseal dysplasia, corner fracture type, 184255
FOXC2	144.2	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXI1	209.3	100.0%	100.0%	Enlarged vestibular aqueduct, 600791
FRAS1	123.1	99.9%	99.3%	Fraser syndrome 1, 219000
FREM1	112.5	99.8%	98.8%	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	160.8	100.0%	99.6%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FXYD2	118.4	100.0%	100.0%	Hypomagnesemia 2, renal, 154020
G6PC	149.8	100.0%	100.0%	Glycogen storage disease Ia, 232200
GALNT3	126.0	99.9%	98.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GANAB	113.2	100.0%	98.9%	Polycystic kidney disease 3, 600666
GATA3	247.4	100.0%	100.0%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GCM2	142.3	100.0%	100.0%	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
GLA	74.4	99.4%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLIS2	148.5	100.0%	100.0%	Nephronophthisis 7, 611498
GLIS3	133.1	100.0%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	176.2	100.0%	99.8%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GREB1L	133.2	100.0%	99.4%	Renal hypodysplasia/aplasia 3, 617805
GRHPR	106.8	85.1%	82.3%	Hyperoxaluria, primary, type II, 260000

GRIP1	114.2	100.0%	99.4%	Fraser syndrome 3, 617667
GSN	123.5	95.6%	93.8%	Amyloidosis, Finnish type, 105120
HNF1B	130.8	99.8%	97.9%	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853
HNF4A	140.7	100.0%	99.3%	MODY, type I, 125850 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026
HOGA1	163.0	100.0%	99.5%	Hyperoxaluria, primary, type III, 613616
HPRT1	56.9	97.8%	87.8%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	183.2	95.8%	89.4%	Apparent mineralocorticoid excess, 218030
IFNG	145.5	100.0%	100.0%	No OMIM Disease ID
IFT122	126.6	100.0%	99.6%	Cranioectodermal dysplasia 1, 218330
IFT140	127.6	100.0%	99.6%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	98.4	100.0%	99.5%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	122.9	100.0%	99.9%	?Bardet-Biedl syndrome 19, 615996
IFT43	119.5	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
INF2	112.4	86.1%	84.3%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
INPP5E	131.1	100.0%	99.3%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INTU	112.9	99.8%	98.0%	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	147.7	100.0%	100.0%	Nephronophthisis 2, infantile, 602088
IQCB1	90.6	91.0%	79.0%	Senior-Loken syndrome 5, 609254
ITGA3	162.5	99.4%	98.1%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA8	117.1	100.0%	99.8%	Renal hypodysplasia/aplasia 1, 191830
JAG1	143.4	99.4%	97.6%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
KANK2	180.1	100.0%	100.0%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KCNJ1	159.7	100.0%	100.0%	Bartter syndrome, type 2, 241200

KCNJ10	157.5	89.3%	88.6%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ5	171.5	100.0%	100.0%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KIAA0556	135.2	100.0%	99.8%	Joubert syndrome 26, 616784
KIF14	112.4	99.8%	97.7%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF7	120.4	99.3%	96.6%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIRREL1	175.9	100.0%	100.0%	No OMIM Disease ID
KL	185.9	99.8%	98.9%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLHL3	110.5	99.9%	98.3%	Pseudohypoaldosteronism, type IID, 614495
LAGE3	73.3	99.2%	94.3%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMB2	182.1	100.0%	99.7%	Pierson syndrome, 609049 Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199
LCAT	156.5	99.6%	96.1%	Norum disease, 245900 Fish-eye disease, 136120
LMX1B	163.9	100.0%	99.3%	Nail-patella syndrome, 161200
LRIG2	131.7	99.9%	99.0%	Urofacial syndrome 2, 615112
LRP2	140.5	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRP4	136.5	99.8%	99.1%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	183.1	99.9%	99.4%	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Osteopetrosis, autosomal dominant 1, 607634
LYZ	142.4	100.0%	100.0%	Amyloidosis, renal, 105200
LZTFL1	116.5	99.9%	99.2%	Bardet-Biedl syndrome 17, 615994
MAFB	140.5	100.0%	100.0%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	95.1	99.8%	98.5%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGI2	93.7	95.1%	92.1%	Nephrotic syndrome, type 15, 617609

MAPKBP1	144.1	100.0%	100.0%	Nephronophthisis 20, 617271
MKKS	161.5	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	98.8	99.9%	98.5%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MOCOS	156.5	100.0%	99.2%	Xanthinuria, type II, 603592
MYH9	140.9	99.7%	99.0%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO1E	119.1	99.9%	98.5%	Glomerulosclerosis, focal segmental, 6, 614131
NCAPG2	121.6	99.6%	97.6%	Khan-Khan-Katsanis syndrome, 618460
NEK1	111.2	99.9%	98.0%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	153.7	100.0%	99.9%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NOTCH2	130.5	100.0%	99.8%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPHP1	119.7	99.8%	97.8%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	121.6	99.7%	98.3%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	134.2	100.0%	99.8%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	116.5	100.0%	99.8%	Nephrotic syndrome, type 1, 256300
NPHS2	121.7	100.0%	99.5%	Nephrotic syndrome, type 2, 600995
NR3C2	129.7	99.8%	98.5%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NUP107	126.3	99.9%	98.0%	Galloway-Mowat syndrome 7, 618348 ?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP133	119.3	99.9%	98.3%	Nephrotic syndrome, type 18, 618177 ?Galloway-Mowat syndrome 8, 618349
NUP160	139.2	100.0%	99.8%	?Nephrotic syndrome, type 19, 618178
NUP205	134.3	99.5%	98.7%	?Nephrotic syndrome, type 13, 616893
NUP85	132.5	100.0%	100.0%	Nephrotic syndrome, type 17, 618176
NUP93	122.3	97.1%	94.3%	Nephrotic syndrome, type 12, 616892

OCRL	106.2	99.9%	98.6%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	52.3	85.5%	70.0%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OSGEP	104.4	100.0%	97.8%	Galloway-Mowat syndrome 3, 617729
PAX2	198.0	100.0%	100.0%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PBX1	115.4	100.0%	98.3%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCBD1	109.5	100.0%	99.8%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDE6D	121.7	100.0%	100.0%	?Joubert syndrome 22, 615665
PDSS2	115.4	99.3%	95.2%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	107.9	99.7%	98.2%	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	41.1	44.3%	36.9%	Polycystic kidney disease 1, 173900
PKD2	105.1	99.2%	96.8%	Polycystic kidney disease 2, 613095
PKHD1	132.5	100.0%	99.5%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PLCE1	129.1	99.9%	99.2%	Nephrotic syndrome, type 3, 610725
PMM2	130.3	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PTH1R	121.2	100.0%	99.5%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTPRO	127.1	100.0%	99.3%	Nephrotic syndrome, type 6, 614196
REN	135.2	100.0%	99.9%	Renal tubular dysgenesis, 267430 Hyperuricemic nephropathy, familial juvenile 2, 613092
RMND1	130.7	100.0%	99.0%	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	129.6	98.8%	97.4%	Vesicoureteral reflux 2, 610878
RPGRIP1L	124.2	96.8%	95.8%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RRM2B	142.6	100.0%	99.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
SALL1	127.8	99.9%	99.3%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480

SALL4	147.5	100.0%	98.7%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SARS2	129.0	95.4%	93.8%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	106.4	99.9%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	140.2	99.9%	98.9%	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	138.7	100.0%	100.0%	Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200 Bronchiectasis with or without elevated sweat chloride 1, 211400
SCNN1G	152.3	99.7%	97.5%	Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 3, 613071
SDCCAG8	123.5	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEC61A1	125.6	100.0%	99.9%	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SGPL1	133.4	100.0%	100.0%	Nephrotic syndrome, type 14, 617575
SIX5	87.2	100.0%	98.9%	Branchiootorenal syndrome 2, 610896
SLC12A1	144.9	100.0%	99.7%	Bartter syndrome, type 1, 601678
SLC12A3	151.1	100.0%	100.0%	Gitelman syndrome, 263800
SLC16A12	134.4	100.0%	99.9%	Cataract 47, juvenile, with microcornea, 612018
SLC22A12	131.0	100.0%	99.9%	Hypouricemia, renal, 220150
SLC26A1	171.5	100.0%	100.0%	?Nephrolithiasis, calcium oxalate, 167030
SLC26A3	133.1	100.0%	99.1%	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	159.5	100.0%	99.9%	Fanconi-Bickel syndrome, 227810
SLC2A9	108.3	100.0%	98.9%	Hypouricemia, renal, 2, 612076
SLC34A1	166.0	100.0%	99.9%	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Hypercalcemia, infantile, 2, 616963 ?Fanconi renotubular syndrome 2, 613388
SLC34A3	165.6	100.0%	99.5%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC36A2	104.3	100.0%	99.9%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	122.0	100.0%	99.7%	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC3A1	147.7	100.0%	99.7%	Cystinuria, 220100
SLC41A1	153.1	100.0%	100.0%	No OMIM Disease ID

SLC4A1	151.7	100.0%	100.0%	Cryohydrocytosis, 185020 Spherocytosis, type 4, 612653 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590
SLC4A4	114.4	99.8%	97.9%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	150.5	100.0%	100.0%	Renal glucosuria, 233100
SLC6A19	139.1	100.0%	100.0%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A20	164.1	100.0%	99.9%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC7A7	110.7	100.0%	99.8%	Lysinuric protein intolerance, 222700
SLC7A9	126.6	100.0%	99.3%	Cystinuria, 220100
SLC9A3	181.6	100.0%	99.8%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	159.7	100.0%	100.0%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLIT3	133.0	99.6%	97.9%	No OMIM Disease ID
SMARCAL1	119.6	100.0%	99.8%	Schimke immunoosseous dysplasia, 242900
SOX17	147.6	100.0%	100.0%	Vesicoureteral reflux 3, 613674
STRA6	125.5	100.0%	99.9%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STX16	116.6	99.9%	99.0%	Pseudohypoparathyroidism, type IB, 603233
TBX18	111.5	99.9%	98.4%	Congenital anomalies of kidney and urinary tract 2, 143400
TCTN1	96.0	95.7%	92.6%	Joubert syndrome 13, 614173
TCTN2	127.0	100.0%	99.0%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	121.0	100.0%	100.0%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
THBD	208.2	100.0%	100.0%	Thrombophilia due to thrombomodulin defect, 614486
TMEM107	161.8	100.0%	100.0%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM138	87.8	100.0%	99.0%	Joubert syndrome 16, 614465
TMEM216	92.0	99.9%	96.9%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	112.1	100.0%	99.7%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970

TMEM237	114.5	99.9%	98.8%	Joubert syndrome 14, 614424
TMEM260	117.1	99.9%	97.8%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	80.6	99.3%	93.5%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TNXB	119.2	99.8%	97.6%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP53RK	93.0	99.8%	97.7%	Galloway-Mowat syndrome 4, 617730
TPRKB	57.9	81.5%	74.8%	Galloway-Mowat syndrome 5, 617731
TRAF3IP1	87.5	99.1%	96.7%	Senior-Loken syndrome 9, 616629
TRIM32	132.8	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRPC6	94.2	98.0%	95.8%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	128.5	99.9%	99.0%	Hypomagnesemia 1, intestinal, 602014
TSC1	117.4	99.6%	98.4%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	155.5	100.0%	100.0%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TTC21B	115.1	99.9%	98.8%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC8	116.8	99.7%	97.8%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
UMOD	123.1	97.7%	95.8%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Medullary cystic kidney disease 2, 603860 Hyperuricemic nephropathy, familial juvenile 1, 162000
UPK3A	114.2	100.0%	100.0%	No OMIM Disease ID
UQCC2	146.3	99.8%	98.8%	Mitochondrial complex III deficiency, nuclear type 7, 615824
VDR	116.5	99.0%	96.4%	Rickets, vitamin D-resistant, type IIA, 277440
VIPAS39	114.4	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	111.7	100.0%	100.0%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	125.3	100.0%	99.4%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376

WDR35	137.8	99.5%	98.3%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR60	111.7	99.8%	97.8%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR73	164.4	100.0%	100.0%	Galloway-Mowat syndrome 1, 251300
WNK1	138.8	100.0%	99.6%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
WNK4	157.3	100.0%	99.6%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	254.1	99.9%	99.1%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	96.5	100.0%	99.6%	Mesothelioma, somatic, 156240 Wilms tumor, type 1, 194070 Frasier syndrome, 136680 Denys-Drash syndrome, 194080 Meacham syndrome, 608978 Nephrotic syndrome, type 4, 256370
XDH	98.4	100.0%	99.8%	Xanthinuria, type I, 278300
XPNPEP3	103.7	100.0%	99.9%	Nephronophthisis-like nephropathy 1, 613159
ZNF423	215.2	100.0%	100.0%	Nephronophthisis 14, 614844 Joubert syndrome 19, 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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors