RENAL DISORDERS
GENE PANEL DG 3.6.0 (319 GENES)

Releasedate: 05-04-2023

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACE	100%	100%	Renal tubular dysgenesis, 267430
ACTN4	100%	100%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	100%	100%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS9	99%	99%	No OMIM disease ID
ADCY10	100%	100%	No OMIM disease ID
AGT	100%	100%	Renal tubular dysgenesis, 267430
AGTR1	100%	100%	Renal tubular dysgenesis, 267430
AGXT	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHI1	100%	100%	Joubert syndrome 3, 608629
ALDOB	100%	100%	Fructose intolerance, hereditary, 229600
ALG1	100%	100%	Congenital disorder of glycosylation, type Ik, 608540
ALG8	96%	96%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	100%	100%	Alstrom syndrome, 203800
AMN	100%	100%	Imerslund-Grasbeck syndrome 2, 618882
ANKFY1	100%	100%	No OMIM disease ID
ANKS6	99%	99%	Nephronophthisis 16, 615382
ANLN	100%	100%	Focal segmental glomerulosclerosis 8, 616032
ANOS1	100%	99%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AP2S1	100%	100%	Hypocalciuric hypercalcemia, type III, 600740
APOL1	100%	100%	No OMIM disease ID
APRT	100%	100%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	100%	100%	Diabetes insipidus, nephrogenic, 2, 125800
ARHGAP24	100%	100%	No OMIM disease ID

ARHGDIA	100%	100%	Nephrotic syndrome, type 8, 615244
ARL13B	100%	100%	Joubert syndrome 8, 612291
ARL6	100%	100%	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ATP1A1	100%	100%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP6V0A4	100%	100%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1B1	100%	100%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP7B	100%	100%	Wilson disease, 277900
AVIL	100%	100%	Nephrotic syndrome, type 21, 618594
AVP	100%	100%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	100%	100%	Diabetes insipidus, nephrogenic, 1, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	100%	100%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100%	100%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BBIP1	100%	100%	?Bardet-Biedl syndrome 18, 615995
BBS1	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	100%	100%	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	Bardet-Biedl syndrome 7, 615984
BBS9	95%	95%	Bardet-Biedl syndrome 9, 615986
BCS1L	100%	100%	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BICC1	100%	99%	No OMIM disease ID
BSND	100%	100%	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
C3	100%	100%	C3 deficiency, 613779
CA2	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CACNA1H	100%	100%	Hyperaldosteronism, familial, type IV, 617027

CASR	100%	100%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalciuric hypercalcemia, type I, 145980
CBWD1	99%	97%	No OMIM disease ID
CC2D2A	98%	98%	COACH syndrome 2, 619111 Retinitis pigmentosa 93, 619845 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCNQ	100%	99%	STAR syndrome, 300707
CD151	100%	100%	Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD2AP	100%	100%	Glomerulosclerosis, focal segmental, 3, 607832
CD46	100%	100%	No OMIM disease ID
CEP120	100%	100%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP164	100%	100%	Nephronophthisis 15, 614845
CEP290	100%	100%	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134
CEP41	100%	100%	Joubert syndrome 15, 614464
CEP55	100%	100%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	100%	100%	Nephronophthisis 18, 615862
CFB	100%	100%	?Complement factor B deficiency, 615561
CFH	100%	100%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	99%	97%	No OMIM disease ID
CFHR3	99%	99%	No OMIM disease ID
CFHR5	100%	100%	Nephropathy due to CFHR5 deficiency, 614809
CFI	100%	100%	Complement factor I deficiency, 610984
CHRM3	100%	100%	Prune belly syndrome, 100100
CHRNA3	100%	100%	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CLCN2	100%	100%	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN5	100%	99%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Hypophosphatemic rickets, 300554

			Dent disease 1, 300009 Nephrolithiasis, type I, 310468
CLCNKB	100%	100%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN10	100%	100%	HELIX syndrome, 617671
CLDN16	100%	100%	Hypomagnesemia 3, renal, 248250
CLDN19	100%	100%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	100%	100%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and impaired intellectual development 1, 616418
COL4A1	100%	100%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 Brain small vessel disease with or without ocular anomalies, 175780
COL4A3	100%	100%	Hematuria, benign familial, 141200 Alport syndrome 3, autosomal dominant, 104200 Alport syndrome 2, autosomal recessive, 203780
COL4A4	100%	100%	Hematuria, familial benign, 141200 Alport syndrome 2, autosomal recessive, 203780
COL4A5	99%	98%	Alport syndrome 1, X-linked, 301050
COQ2	96%	96%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	100%	100%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	100%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8B	100%	100%	Nephrotic syndrome, type 9, 615573
COQ9	100%	100%	Coenzyme Q10 deficiency, primary, 5, 614654
CPLANE1	100%	100%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CRB2	100%	100%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CSPP1	100%	100%	Joubert syndrome 21, 615636
CTNS	100%	100%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CUBN	100%	100%	Imerslund-Grasbeck syndrome 1, 261100
CUL3	100%	100%	Neurodevelopmental disorder with or without autism or seizures, 619239 Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	100%	100%	Hypercalcemia, infantile, 1, 143880

DAAM2	100%	100%	Nephrotic syndrome, type 24, 619263
DCDC2	100%	100%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394
DGKE	100%	100%	Nephrotic syndrome, type 7, 615008
DLC1	100%	100%	Colorectal cancer, somatic, 114500
DMP1	100%	100%	Hypophosphatemic rickets, AR, 241520
DNAJB11	100%	100%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DSTYK	100%	100%	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DYNC2H1	99%	99%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DZIP1L	100%	100%	Polycystic kidney disease 5, 617610
EGF	100%	100%	?Hypomagnesemia 4, renal, 611718
EHHADH	100%	100%	?Fanconi renotubular syndrome 3, 615605
EMP2	100%	100%	Nephrotic syndrome, type 10, 615861
ENPP1	100%	99%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522
EYA1	100%	100%	Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 602588 ?Otofaciocervical syndrome, 166780
FAH	100%	100%	Tyrosinemia, type I, 276700
FAM111A	100%	100%	Kenny-Caffey syndrome, type 2, 127000 Gracile bone dysplasia, 602361
FAM20A	100%	100%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAN1	100%	100%	Interstitial nephritis, karyomegalic, 614817
FAT1	100%	100%	No OMIM disease ID
FGF23	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FN1	100%	100%	Spondylometaphyseal dysplasia, corner fracture type, 184255 Glomerulopathy with fibronectin deposits 2, 601894
FOXC2	100%	100%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXI1	100%	100%	Enlarged vestibular aqueduct, 600791
FRAS1	100%	99%	Fraser syndrome 1, 219000

FREM1	100%	100%	Manitoba oculotrichoanal syndrome, 248450 Bifid nose with or without anorectal and renal anomalies, 608980 Trigonocephaly 2, 614485
FREM2	99%	99%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FXYD2	100%	100%	Hypomagnesemia 2, renal, 154020
G6PC	100%	100%	Glycogen storage disease Ia, 232200
GALNT3	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GANAB	100%	100%	Polycystic kidney disease 3, 600666
GAPVD1	100%	100%	No OMIM disease ID
GATA3	100%	100%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GCM2	100%	100%	Hypoparathyroidism, familial isolated 2, 618883 Hyperparathyroidism 4, 617343
GFRA1	100%	100%	Renal hypodysplasia/aplasia 4, 619887
GLA	90%	90%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLI3	100%	100%	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GLIS2	100%	100%	Nephronophthisis 7, 611498
GLIS3	100%	100%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	100%	100%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GREB1L	100%	100%	Deafness, autosomal dominant 80, 619274 Renal hypodysplasia/aplasia 3, 617805
GRHPR	100%	100%	Hyperoxaluria, primary, type II, 260000
GRIP1	100%	100%	Fraser syndrome 3, 617667
GSN	100%	100%	Amyloidosis, Finnish type, 105120
HNF1A	100%	100%	Hepatic adenoma, somatic, 142330 Diabetes mellitus, insulin-dependent, 20, 612520 MODY, type III, 600496 Renal cell carcinoma, 144700
HNF1B	100%	100%	Type 2 diabetes mellitus, 125853 Renal cysts and diabetes syndrome, 137920
HNF4A	100%	100%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850

HOGA1	100%	100%	Hyperoxaluria, primary, type III, 613616
HPRT1	100%	100%	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	100%	100%	Apparent mineralocorticoid excess, 218030
IFNG	100%	100%	?Immunodeficiency 69, mycobacteriosis, 618963
IFT122	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT140	100%	100%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	100%	100%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100%	100%	Bardet-Biedl syndrome 19, 615996
IFT43	100%	100%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
INF2	100%	99%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
INPP5E	100%	100%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INTU	100%	100%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100%	100%	Nephronophthisis 2, infantile, 602088
IQCB1	100%	100%	Senior-Loken syndrome 5, 609254
ITGA3	100%	100%	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748
ITGA8	100%	100%	Renal hypodysplasia/aplasia 1, 191830
ITSN1	100%	100%	No OMIM disease ID
ITSN2	100%	100%	No OMIM disease ID
JAG1	100%	100%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KANK1	100%	100%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	100%	100%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KCNJ1	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	100%	100%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780

KCNJ16	100%	100%	Hypokalemic tubulopathy and deafness, 619406
KCNJ5	100%	100%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KIAA0556	100%	100%	Joubert syndrome 26, 616784
KIF14	100%	100%	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF7	100%	99%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KIRREL1	100%	100%	Nephrotic syndrome, type 23, 619201
KL	99%	99%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLHL3	100%	100%	Pseudohypoaldosteronism, type IID, 614495
LAGE3	100%	100%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA5	100%	100%	Nephrotic syndrome, type 26, 620049 ?Bent bone dysplasia syndrome 2, 620076
LAMB2	100%	100%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCAT	100%	100%	Fish-eye disease, 136120 Norum disease, 245900
LMX1B	100%	100%	Focal segmental glomerulosclerosis 10, 256020 Nail-patella syndrome, 161200
LRIG2	100%	100%	Urofacial syndrome 2, 615112
LRP2	100%	100%	Donnai-Barrow syndrome, 222448
LRP4	100%	100%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	100%	100%	Osteopetrosis, autosomal dominant 1, 607634 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Exudative vitreoretinopathy 4, 601813 van Buchem disease, type 2, 607636
LYZ	100%	100%	Amyloidosis, renal, 105200
LZTFL1	100%	100%	Bardet-Biedl syndrome 17, 615994
MAFB	100%	100%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300

MAGED2	100%	99%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGI2	98%	97%	Nephrotic syndrome, type 15, 617609
MAPKBP1	100%	100%	Nephronophthisis 20, 617271
MKKS	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKS1	100%	100%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MMACHC	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MOCOS	100%	100%	Xanthinuria, type II, 603592
MYH9	100%	100%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYO1E	100%	100%	Glomerulosclerosis, focal segmental, 6, 614131
NCAPG2	100%	100%	Khan-Khan-Katsanis syndrome, 618460
NEK1	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	100%	100%	Renal-hepatic-pancreatic dysplasia 2, 615415 ?Nephronophthisis 9, 613824
NEU1	100%	100%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NOS1AP	100%	100%	Nephrotic syndrome, type 22, 619155
NOTCH2	100%	100%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP1	100%	100%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	100%	100%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	100%	100%	Senior-Loken syndrome 4, 606996 Nephronophthisis 4, 606966
NPHS1	100%	100%	Nephrotic syndrome, type 1, 256300
NPHS2	100%	100%	Nephrotic syndrome, type 2, 600995
NR3C2	100%	100%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NUP107	100%	100%	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348 Nephrotic syndrome, type 11, 616730

NUP133	100%	100%	?Galloway-Mowat syndrome 8, 618349 Nephrotic syndrome, type 18, 618177
NUP160	100%	100%	?Nephrotic syndrome, type 19, 618178
NUP205	100%	100%	?Nephrotic syndrome, type 13, 616893
NUP85	100%	100%	Nephrotic syndrome, type 17, 618176
NUP93	95%	95%	Nephrotic syndrome, type 12, 616892
NXF5	100%	99%	No OMIM disease ID
OCRL	100%	100%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	100%	100%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
OSGEP	100%	100%	Galloway-Mowat syndrome 3, 617729
OXGR1	100%	100%	No OMIM disease ID
PAX2	100%	100%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PBX1	100%	99%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCBD1	100%	100%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDE6D	100%	100%	Joubert syndrome 22, 615665
PDSS2	100%	100%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	99%	99%	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	99%	99%	Polycystic kidney disease 1, 173900
PKD2	100%	100%	Polycystic kidney disease 2, 613095
PKHD1	100%	100%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PLCE1	100%	99%	Nephrotic syndrome, type 3, 610725
PMM2	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PODXL	94%	93%	No OMIM disease ID
PTH1R	100%	100%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Chondrodysplasia, Blomstrand type, 215045
PTPRO	99%	99%	Nephrotic syndrome, type 6, 614196
RAD21	100%	100%	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376

REN	100%	100%	Renal tubular dysgenesis, 267430 Tubulointerstitial kidney disease, autosomal dominant, 4, 613092
RMND1	100%	100%	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	100%	100%	Vesicoureteral reflux 2, 610878
RPGRIP1L	100%	100%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RRM2B	100%	100%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
SALL1	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	100%	100%	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323
SARS2	100%	100%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	100%	100%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	100%	100%	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100%	100%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125 Liddle syndrome 1, 177200
SCNN1G	100%	100%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126 Liddle syndrome 2, 618114
SDCCAG8	100%	100%	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993
SEC61A1	100%	100%	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SGPL1	100%	100%	Nephrotic syndrome, type 14, 617575
SIX5	100%	100%	Branchiootorenal syndrome 2, 610896
SLC12A1	96%	96%	Bartter syndrome, type 1, 601678
SLC12A3	100%	100%	Gitelman syndrome, 263800
SLC16A12	100%	100%	Cataract 47, juvenile, with microcornea, 612018
SLC22A12	100%	99%	Hypouricemia, renal, 220150
SLC26A1	100%	100%	?Nephrolithiasis, calcium oxalate, 167030
SLC26A3	100%	100%	Diarrhea 1, secretory chloride, congenital, 214700

SLC2A2	100%	100%	Fanconi-Bickel syndrome, 227810
SLC2A9	100%	100%	Hypouricemia, renal, 2, 612076
SLC34A1	100%	100%	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	100%	100%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC36A2	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	100%	100%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC3A1	96%	96%	Cystinuria, 220100
SLC41A1	100%	100%	?Nephronophthisis-like nephropathy 2, 619468
SLC4A1	100%	100%	Distal renal tubular acidosis 1, 179800 Spherocytosis, type 4, 612653 Distal renal tubular acidosis 4 with hemolytic anemia, 611590 Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900
SLC4A4	100%	99%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	100%	100%	Renal glucosuria, 233100
SLC6A19	100%	100%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A20	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC7A7	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	100%	100%	Cystinuria, 220100
SLC9A3	100%	99%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	100%	100%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLIT3	100%	100%	No OMIM disease ID
SMARCAL1	100%	100%	Schimke immunoosseous dysplasia, 242900
SOX17	100%	100%	Vesicoureteral reflux 3, 613674
STRA6	100%	100%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STX16	100%	100%	Pseudohypoparathyroidism, type IB, 603233
TBC1D8B	100%	99%	Nephrotic syndrome, type 20, 301028
TBX18	100%	100%	Congenital anomalies of kidney and urinary tract 2, 143400

TCTN1	95%	94%	Joubert syndrome 13, 614173
TCTN2	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100%	100%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
THBD	100%	100%	Thrombophilia 12 due to thrombomodulin defect, 614486
TMEM107	100%	100%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM138	100%	100%	Joubert syndrome 16, 614465
TMEM216	100%	100%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	100%	100%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100%	100%	Joubert syndrome 14, 614424
TMEM260	100%	100%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	99%	97%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TNS2	100%	100%	No OMIM disease ID
TNXB	100%	100%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP53RK	100%	100%	Galloway-Mowat syndrome 4, 617730
TPRKB	82%	81%	Galloway-Mowat syndrome 5, 617731
TRAF3IP1	100%	100%	Senior-Loken syndrome 9, 616629
TRIM32	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRPC6	100%	100%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	100%	100%	Hypomagnesemia 1, intestinal, 602014
TSC1	100%	100%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
TSC2	100%	100%	Lymphangioleiomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254

TTC21B	100%	99%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 Nephronophthisis 12, 613820
TTC8	100%	99%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
UMOD	100%	100%	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000
UPK3A	100%	100%	No OMIM disease ID
UQCC2	100%	100%	Mitochondrial complex III deficiency, nuclear type 7, 615824
VDR	100%	100%	Rickets, vitamin D-resistant, type IIA, 277440
VIPAS39	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100%	100%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 Cholestasis, progressive familial intrahepatic, 12, 620010 Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	100%	100%	Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378 Senior-Loken syndrome 8, 616307 Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Spermatogenic failure 72, 619867
WDR35	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR60	100%	100%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR73	100%	100%	Galloway-Mowat syndrome 1, 251300
WNK1	100%	100%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	100%	100%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	100%	99%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	100%	100%	Mesothelioma, somatic, 156240 Meacham syndrome, 608978 Frasier syndrome, 136680 Nephrotic syndrome, type 4, 256370 Denys-Drash syndrome, 194080 Wilms tumor, type 1, 194070
XDH	100%	100%	Xanthinuria, type I, 278300
XPNPEP3	100%	100%	Nephronophthisis-like nephropathy 1, 613159
XPO5	100%	100%	No OMIM disease ID
ZMPSTE24	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy 1, 275210

ZNF423	100%	100%	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.
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.
TWIST X2 is the chemistry used for WES analysis.
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 coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : March 17th, 2023
This list is accurate for panel version DG 3.6.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors