RENAL DISORDERS GENE PANEL DG 2.18 (291 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ACE	99,90%	98,40%	100%	100%	Renal tubular dysgenesis, 267430
ACTN4	100%	99,30%	100%	100%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	97,10%	93,80%	99,90%	99,50%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS9	99,50%	98,70%	100%	100%	No OMIM disease ID
ADCY10	100%	99,90%	100%	100%	No OMIM disease ID
AGT	100%	100%	100%	100%	Renal tubular dysgenesis, 267430
AGTR1	92,00%	91,80%	100%	100%	Renal tubular dysgenesis, 267430
AGXT	100%	100%	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHI1	99,70%	97,90%	100%	100%	Joubert syndrome 3, 608629
ALDOB	100%	99,10%	100%	100%	Fructose intolerance, hereditary, 229600
ALG8	97,20%	95,60%	96,60%	96,60%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	99,70%	100%	100%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	99,80%	99,50%	100%	100%	Alstrom syndrome, 203800
AMN	89,70%	80,00%	100%	100%	Megaloblastic anemia-1, Norwegian type, 261100
ANKS6	93,80%	89,50%	97,90%	95,80%	Nephronophthisis 16, 615382
ANLN	98,70%	97,50%	100%	100%	Focal segmental glomerulosclerosis 8, 616032
ANOS1	89,80%	88,90%	99,90%	99,40%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AP2S1	90,40%	90,30%	100%	100%	Hypocalciuric hypercalcemia, type III, 600740
APOL1	100%	100%	100%	100%	No OMIM disease ID
APRT	100%	99,50%	100%	100%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	100%	98,60%	100%	100%	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	100%	100%	100%	100%	Nephrotic syndrome, type 8, 615244
ARL13B	100%	99,20%	100%	100%	Joubert syndrome 8, 612291
ARL6	99,90%	98,60%	100%	100%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ATP1A1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314

ATP6V0A4	100%	99,90%	100%	100%	Renal tubular acidosis, distal, autosomal recessive, 602722
ATP6V1B1	100%	100%	100%	100%	Renal tubular acidosis with deafness, 267300
ATP7B	99,90%	99,20%	100%	100%	Wilson disease, 277900
AVP	84,90%	64,30%	100%	99,90%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	100%	99,40%	100%	100%	Nephrogenic syndrome of inappropriate antidiuresis, 300539 Diabetes insipidus, nephrogenic, 304800
B9D1	92,20%	92,00%	100%	100%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100%	100%	100%	100%	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BBIP1	98,60%	92,40%	100%	100%	?Bardet-Biedl syndrome 18, 615995
BBS1	100%	100%	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	99,80%	100%	100%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	100%	99,50%	100%	100%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,90%	99,30%	100%	100%	Bardet-Biedl syndrome 4, 615982
BBS5	99,00%	93,90%	100%	100%	Bardet-Biedl syndrome 5, 615983
BBS7	98,70%	95,50%	100%	100%	Bardet-Biedl syndrome 7, 615984
BBS9	99,70%	97,60%	100%	100%	Bardet-Biedl syndrome 9, 615986
BCS1L	100%	100%	100%	100%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BICC1	100%	100%	100%	100%	No OMIM disease ID
BSND	100%	100%	100%	100%	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522
C3	99,90%	99,20%	100%	100%	C3 deficiency, 613779
C5orf42	99,70%	98,40%	100%	100%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CA2	100%	100%	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CACNA1H	98,70%	96,40%	100%	99,90%	Hyperaldosteronism, familial, type IV, 617027
CASR	100%	99,90%	100%	100%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200
CBWD1	20,80%	19,40%	99,60%	99,30%	No OMIM disease ID

CC2D2A	99,70%	97,70%	98,20%	98,20%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CD2AP	99,90%	98,80%	100%	100%	Glomerulosclerosis, focal segmental, 3, 607832
CD46	99,90%	99,40%	100%	100%	No OMIM disease ID
CEP120	100%	99,50%	100%	100%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	99,90%	98,30%	100%	100%	Nephronophthisis 15, 614845
CEP290	96,10%	90,00%	100%	100%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	99,80%	97,40%	100%	100%	Joubert syndrome 15, 614464
CEP55	100%	99,80%	100%	100%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	99,80%	97,40%	100%	100%	Nephronophthisis 18, 615862
CFB	100%	100%	100%	100%	?Complement factor B deficiency, 615561
CFH	99,90%	99,00%	100%	99,90%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	96,40%	94,90%	95,40%	93,80%	No OMIM disease ID
CFHR3	94,00%	92,20%	96,00%	95,20%	No OMIM disease ID
CFI	99,20%	96,80%	100%	100%	Complement factor I deficiency, 610984
CHRM3	100%	100%	100%	100%	?Prune belly syndrome, 100100
CHRNA3	100%	99,40%	100%	100%	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CLCN5	99,90%	98,30%	100%	100%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Dent disease, 300009 Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468
CLCNKB	99,10%	95,90%	100%	100%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN10	100%	100%	100%	100%	HELIX syndrome, 617671
CLDN16	100%	100%	100%	100%	Hypomagnesemia 3, renal, 248250
CLDN19	98,50%	93,10%	100%	100%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	100%	100%	100%	100%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
COL4A1	98,70%	97,40%	100%	100%	?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	98,70%	98,00%	100%	100%	Hematuria, benign familial, 141200 Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200
COL4A4	99,90%	98,20%	100%	100%	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	97,80%	89,00%	100%	99,80%	Alport syndrome 1, X-linked, 301050
COQ2	98,00%	95,30%	97,20%	97,20%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	99,90%	98,40%	100%	100%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	99,80%	100%	100%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8B	100%	99,30%	100%	100%	Nephrotic syndrome, type 9, 615573
COQ9	100%	97,90%	100%	100%	Coenzyme Q10 deficiency, primary, 5, 614654
CRB2	98,50%	93,00%	100%	100%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CSPP1	99,80%	98,70%	100%	100%	Joubert syndrome 21, 615636
CTNS	100%	99,80%	100%	100%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CUBN	99,70%	98,30%	100%	100%	Megaloblastic anemia-1, Finnish type, 261100
CUL3	99,90%	98,80%	100%	100%	Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	100%	99,90%	100%	100%	Hypercalcemia, infantile, 1, 143880
DCDC2	100%	99,90%	100%	100%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DGKE	99,80%	98,10%	100%	100%	Nephrotic syndrome, type 7, 615008
DMP1	100%	99,90%	100%	100%	Hypophosphatemic rickets, AR, 241520
DNAJB11	100%	99,50%	100%	100%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DSTYK	99,90%	99,20%	100%	100%	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DYNC2H1	98,80%	95,50%	100%	100%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DZIP1L	99,90%	99,00%	100%	100%	Polycystic kidney disease 5, 617610
EGF	99,90%	99,70%	100%	100%	Hypomagnesemia 4, renal, 611718
EHHADH	100%	100%	100%	100%	?Fanconi renotubular syndrome 3, 615605
EMP2	99,90%	96,70%	100%	100%	Nephrotic syndrome, type 10, 615861

ENPP1	96,40%	91,20%	98,70%	97,80%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 Arterial calcification, generalized, of infancy, 1, 208000
EYA1	99,90%	99,70%	100%	100%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Branchiootic syndrome 1, 602588
FAH	100%	100%	100%	100%	Tyrosinemia, type I, 276700
FAM20A	99,60%	94,70%	100%	100%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM58A	83,10%	78,50%	98,90%	94,70%	STAR syndrome, 300707
FAN1	100%	99,80%	100%	100%	Interstitial nephritis, karyomegalic, 614817
FAT1	100%	100%	100%	100%	No OMIM disease ID
FGF23	99,60%	97,50%	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FN1	100%	99,30%	100%	100%	Glomerulopathy with fibronectin deposits 2, 601894 Spondylometaphyseal dysplasia, corner fracture type, 184255
FOXC2	100%	96,70%	100%	99,80%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXI1	100%	100%	100%	100%	Enlarged vestibular aqueduct, 600791
FRAS1	100%	99,40%	100%	100%	Fraser syndrome 1, 219000
FREM1	99,90%	99,10%	100%	100%	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	100%	99,30%	100%	100%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FXYD2	100%	100%	100%	100%	Hypomagnesemia 2, renal, 154020
G6PC	100%	100%	100%	100%	Glycogen storage disease Ia, 232200
GALNT3	99,80%	99,00%	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GANAB	99,90%	99,00%	100%	100%	Polycystic kidney disease 3, 600666
GATA3	100%	100%	100%	100%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GCM2	100%	100%	100%	100%	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated, 146200
GLA	99,80%	96,60%	100%	100%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLI3	100%	99,50%	100%	100%	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510

GLIS2	100%	99,80%	100%	100%	Nephronophthisis 7, 611498
GLIS3	100%	99,60%	100%	100%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	99,90%	95,00%	100%	100%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GREB1L	100%	99,90%	100%	100%	Renal hypodysplasia/aplasia 3, 617805
GRHPR	84,20%	81,30%	100%	99,30%	Hyperoxaluria, primary, type II, 260000
GRIP1	100%	99,70%	100%	100%	Fraser syndrome 3, 617667
GSN	95,80%	93,50%	99,90%	99,30%	Amyloidosis, Finnish type, 105120
HNF1B	99,30%	96,10%	100%	100%	Renal cysts and diabetes syndrome, 137920 Diabetes mellitus, noninsulin-dependent, 125853
HNF4A	99,90%	99,00%	100%	100%	MODY, type I, 125850 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026
HOGA1	100%	96,40%	100%	100%	Hyperoxaluria, primary, type III, 613616
HPRT1	99,30%	91,80%	100%	99,30%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	86,00%	82,70%	99,90%	98,10%	Apparent mineralocorticoid excess, 218030
IFNG	100%	100%	100%	100%	No OMIM disease ID
IFT122	100%	99,60%	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT140	99,80%	98,80%	100%	100%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,90%	99,10%	100%	100%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100%	100%	100%	100%	?Bardet-Biedl syndrome 19, 615996
IFT43	100%	100%	100%	100%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
INF2	86,70%	83,80%	100%	100%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
INPP5E	97,10%	92,70%	100%	100%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INTU	99,70%	98,10%	100%	100%	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	100%	100%	100%	100%	Nephronophthisis 2, infantile, 602088
IQCB1	93,90%	85,00%	100%	100%	Senior-Loken syndrome 5, 609254
ITGA3	99,50%	97,40%	100%	100%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA8	99,90%	99,70%	100%	100%	Renal hypodysplasia/aplasia 1, 191830

JAG1	97,70%	96,80%	100%	100%	Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500 ?Deafness, congenital heart defects, and posterior embryotoxon, 617992
KANK2	100%	100%	100%	100%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KCNJ1	100%	100%	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	89,30%	89,00%	100%	100%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ5	100%	100%	100%	100%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KIAA0556	100%	99,90%	100%	100%	Joubert syndrome 26, 616784
KIF14	99,60%	97,70%	100%	100%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF7	93,60%	90,60%	99,10%	97,80%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIRREL1	100%	99,90%	100%	100%	No OMIM disease ID
KL	98,20%	97,20%	98,50%	97,50%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLHL3	100%	99,30%	100%	100%	Pseudohypoaldosteronism, type IID, 614495
LAGE3	95,90%	85,10%	100%	100%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMB2	100%	99,70%	100%	100%	Pierson syndrome, 609049 Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199
LCAT	99,00%	93,80%	100%	100%	Norum disease, 245900 Fish-eye disease, 136120
LMX1B	99,60%	96,30%	100%	100%	Nail-patella syndrome, 161200
LRIG2	99,60%	98,80%	100%	100%	Urofacial syndrome 2, 615112
LRP2	100%	99,90%	100%	100%	Donnai-Barrow syndrome, 222448
LRP4	99,10%	98,80%	100%	100%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	98,50%	98,10%	100%	99,70%	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	100%	100%	100%	100%	Amyloidosis, renal, 105200
LZTFL1	99,90%	99,20%	100%	100%	Bardet-Biedl syndrome 17, 615994
MAFB	100%	99,40%	100%	100%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	99,80%	97,70%	100%	99,90%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGI2	94,50%	92,40%	94,70%	93,30%	Nephrotic syndrome, type 15, 617609
MAPKBP1	100%	100%	100%	100%	Nephronophthisis 20, 617271
MKKS	83,20%	83,20%	90,70%	90,70%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	99,80%	97,90%	100%	100%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MOCOS	99,80%	97,70%	100%	100%	Xanthinuria, type II, 603592
MYH9	100%	99,30%	100%	100%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO1E	99,90%	99,50%	100%	100%	Glomerulosclerosis, focal segmental, 6, 614131
NCAPG2	99,90%	99,20%	100%	100%	Khan-Khan-Katsanis syndrome, 618460
NEK1	99,80%	98,00%	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	100%	99,90%	100%	100%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NOTCH2	100%	99,50%	100%	100%	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPHP1	100%	99,00%	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	99,70%	98,40%	100%	100%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	100%	99,80%	100%	100%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	99,80%	99,10%	100%	100%	Nephrotic syndrome, type 1, 256300
NPHS2	100%	99,50%	100%	100%	Nephrotic syndrome, type 2, 600995
NR3C2	100%	99,70%	100%	100%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NUP107	99,80%	98,50%	100%	100%	Galloway-Mowat syndrome 7, 618348 ?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730

NUP133	99,70%	98,30%	100%	100%	Nephrotic syndrome, type 18, 618177 ?Galloway-Mowat syndrome 8, 618349
NUP160	100%	99,90%	100%	100%	?Nephrotic syndrome, type 19, 618178
NUP205	99,90%	99,40%	100%	100%	?Nephrotic syndrome, type 13, 616893
NUP85	100%	100%	100%	100%	Nephrotic syndrome, type 17, 618176
NUP93	98,00%	94,20%	95,50%	95,50%	Nephrotic syndrome, type 12, 616892
OCRL	99,90%	98,60%	100%	99,90%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	88,00%	73,70%	100%	99,90%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OSGEP	100%	99,40%	100%	100%	Galloway-Mowat syndrome 3, 617729
PAX2	100%	99,90%	100%	100%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PBX1	100%	99,40%	100%	100%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCBD1	100%	99,60%	100%	99,70%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDE6D	100%	100%	100%	100%	?Joubert syndrome 22, 615665
PDSS2	99,80%	97,10%	100%	100%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	100%	99,60%	99,90%	99,20%	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	39,20%	30,00%	99,20%	98,90%	Polycystic kidney disease 1, 173900
PKD2	95,50%	91,10%	99,30%	97,70%	Polycystic kidney disease 2, 613095
PKHD1	100%	99,60%	100%	100%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PLCE1	99,90%	99,30%	100%	100%	Nephrotic syndrome, type 3, 610725
PMM2	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PTH1R	100%	98,70%	100%	100%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTPRO	99,90%	99,40%	100%	100%	Nephrotic syndrome, type 6, 614196
RAD21	99,20%	96,60%	100%	100%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
REN	100%	100%	100%	100%	Renal tubular dysgenesis, 267430 Hyperuricemic nephropathy, familial juvenile 2, 613092
RMND1	100%	98,60%	100%	100%	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	99,40%	97,80%	100%	100%	Vesicoureteral reflux 2, 610878

RPGRIP1L	96,70%	95,70%	100%	99,50%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RRM2B	100%	99,70%	100%	100%	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	99,90%	99,00%	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	98,60%	96,70%	100%	100%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SARS2	95,80%	94,60%	100%	100%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	100%	99,80%	100%	100%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	99,70%	98,20%	100%	100%	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100%	99,70%	100%	100%	Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200 Bronchiectasis with or without elevated sweat chloride 1, 211400
SCNN1G	99,80%	98,30%	100%	100%	Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350 Bronchiectasis with or without elevated sweat chloride 3, 613071
SDCCAG8	100%	99,90%	100%	100%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SEC61A1	100%	100%	100%	100%	Hyperuricemic nephropathy, familial juvenile, 4, 617056
SGPL1	100%	100%	100%	100%	Nephrotic syndrome, type 14, 617575
SIX5	95,40%	88,20%	100%	100%	Branchiootorenal syndrome 2, 610896
SLC12A1	100%	99,90%	100%	100%	Bartter syndrome, type 1, 601678
SLC12A3	100%	99,90%	100%	100%	Gitelman syndrome, 263800
SLC16A12	100%	99,90%	100%	100%	Cataract 47, juvenile, with microcornea, 612018
SLC22A12	100%	99,80%	100%	100%	Hypouricemia, renal, 220150
SLC26A1	100%	99,60%	100%	100%	?Nephrolithiasis, calcium oxalate, 167030
SLC26A3	100%	99,50%	100%	100%	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	100%	100%	100%	100%	Fanconi-Bickel syndrome, 227810
SLC2A9	99,80%	96,10%	100%	100%	Hypouricemia, renal, 2, 612076
SLC34A1	99,90%	99,10%	100%	100%	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Hypercalcemia, infantile, 2, 616963 ?Fanconi renotubular syndrome 2, 613388
SLC34A3	100%	99,40%	100%	100%	Hypophosphatemic rickets with hypercalciuria, 241530

SLC36A2	100%	100%	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	100%	99,20%	100%	100%	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC3A1	100%	99,80%	96,60%	96,60%	Cystinuria, 220100
SLC41A1	100%	100%	100%	100%	No OMIM disease ID
SLC4A1	100%	99,80%	96,10%	96,10%	Spherocytosis, type 4, 612653 Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900 Renal tubular acidosis, distal, AD, 179800 Renal tubular acidosis, distal, AR, 611590
SLC4A4	99,80%	99,20%	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A2	100%	100%	100%	100%	Renal glucosuria, 233100
SLC6A19	100%	100%	100%	100%	Iminoglycinuria, digenic, 242600 HEARTnup disorder, 234500 Hyperglycinuria, 138500
SLC6A20	100%	99,90%	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC7A7	100%	99,90%	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	100%	99,90%	100%	100%	Cystinuria, 220100
SLC9A3	100%	99,70%	100%	99,90%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	100%	98,70%	100%	100%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLIT3	97,90%	95,30%	100%	100%	No OMIM disease ID
SMARCAL1	100%	99,90%	100%	100%	Schimke immunoosseous dysplasia, 242900
SOX17	100%	99,50%	100%	100%	Vesicoureteral reflux 3, 613674
STRA6	100%	99,80%	100%	100%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STX16	100%	98,60%	100%	100%	Pseudohypoparathyroidism, type IB, 603233
TBX18	99,50%	97,10%	100%	100%	Congenital anomalies of kidney and urinary tract 2, 143400
TCTN1	96,70%	93,00%	94,70%	94,70%	Joubert syndrome 13, 614173
TCTN2	100%	99,50%	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
THBD	100%	99,70%	100%	100%	Thrombophilia due to thrombomodulin defect, 614486
TMEM107	100%	100%	100%	100%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562

TMEM138	100%	99,10%	100%	100%	Joubert syndrome 16, 614465
TMEM216	99,90%	98,10%	100%	100%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	100%	99,60%	100%	100%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	100%	99,90%	100%	100%	Joubert syndrome 14, 614424
TMEM260	97,50%	93,40%	100%	100%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	99,50%	95,00%	100%	99,90%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TNXB	99,60%	95,10%	100%	100%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP53RK	92,50%	79,60%	100%	100%	Galloway-Mowat syndrome 4, 617730
TPRKB	81,10%	75,90%	81,90%	81,90%	Galloway-Mowat syndrome 5, 617731
TRAF3IP1	99,60%	97,60%	100%	100%	Senior-Loken syndrome 9, 616629
TRIM32	100%	100%	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRPC6	98,20%	96,10%	100%	100%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	99,90%	99,50%	100%	100%	Hypomagnesemia 1, intestinal, 602014
TSC1	99,80%	98,70%	100%	100%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100%	99,60%	100%	100%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TTC21B	99,90%	99,30%	100%	100%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC8	99,60%	98,10%	100%	100%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
UMOD	97,70%	96,20%	100%	100%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Medullary cystic kidney disease 2, 603860 Hyperuricemic nephropathy, familial juvenile 1, 162000
UPK3A	100%	99,50%	100%	100%	No OMIM disease ID
UQCC2	100%	99,70%	100%	100%	Mitochondrial complex III deficiency, nuclear type 7, 615824
VDR	99,90%	99,00%	100%	100%	Rickets, vitamin D-resistant, type IIA, 277440
VIPAS39	100%	100%	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404

VPS33B	100%	100%	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	100%	99,40%	100%	100%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	99,80%	98,90%	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR60	99,50%	97,00%	100%	100%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR73	100%	100%	100%	100%	Galloway-Mowat syndrome 1, 251300
WNK1	99,90%	99,60%	100%	100%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
WNK4	99,90%	99,30%	100%	100%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	99,10%	94,80%	98,90%	96,20%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	99,90%	98,30%	100%	100%	Mesothelioma, somatic, 156240 Wilms tumor, type 1, 194070 Frasier syndrome, 136680 Denys-Drash syndrome, 194080 Meacham syndrome, 608978 Nephrotic syndrome, type 4, 256370
XDH	100%	99,90%	100%	100%	Xanthinuria, type I, 278300
XPNPEP3	100%	100%	100%	100%	Nephronophthisis-like nephropathy 1, 613159
ZNF423	100%	100%	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors