RENAL DISORDERS GENE PANEL DG 3.00 (305 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
ACE	99,9	98,4	100	100	{Stroke, hemorrhagic}, 614519 Renal tubular dysgenesis, 267430 {SARS, progression of}, 0 {Microvascular complications of diabetes 3}, 612624 {Myocardial infarction, susceptibility to}, 0 [Angiotensin I-converting enzyme, benign serum increase], 0
ACTN4	100	99,3	100	100	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	97,1	93,8	99,9	99,5	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS9	99,5	98,7	100	100	No OMIM disease ID
ADCY10	100	99,9	100	100	{Hypercalciuria, absorptive, susceptibility to}, 143870
AGT	100	100	100	100	Renal tubular dysgenesis, 267430 {Preeclampsia, susceptibility to}, 0 {Hypertension, essential, susceptibility to}, 145500
AGTR1	92	91,8	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	100	100	100	100	Hyperoxaluria, primary, type 1, 259900
AHI1	99,7	97,9	100	100	Joubert syndrome 3, 608629
ALDOB	99,4	96,6	100	100	Fructose intolerance, hereditary, 229600
ALG1	53	45,8	100	100	Congenital disorder of glycosylation, type Ik, 608540
ALG8	97,2	95,6	96,6	96,6	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100	99,7	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	99,8	99,5	100	100	Alstrom syndrome, 203800
AMN	89,7	80	100	100	Imerslund-Grasbeck syndrome 2, 618882

ANKS6	93,8	89,5	97,9	95,8	Nephronophthisis 16, 615382
ANLN	98,7	97,5	100	100	Focal segmental glomerulosclerosis 8, 616032
ANOS1	89,8	88,9	99,9	99,4	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AP2S1	90,4	90,3	100	100	Hypocalciuric hypercalcemia, type III, 600740
APOL1	100	100	100	100	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
APRT	100	99,5	100	100	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	100	98,6	100	100	Diabetes insipidus, nephrogenic, 125800
ARHGDIA	100	100	100	100	Nephrotic syndrome, type 8, 615244
ARL13B	100	99,2	100	100	Joubert syndrome 8, 612291
ARL6	99,9	98,6	100	100	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ATP1A1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP6V0A4	100	99,9	100	100	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1B1	100	100	100	100	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP7B	99,9	99,2	100	100	Wilson disease, 277900
AVIL	100	99,9	100	100	Nephrotic syndrome, type 21, 618594
AVP	84,9	64,3	100	99,9	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	100	99,4	100	100	Diabetes insipidus, nephrogenic, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	85,2	85,1	94,2	93,9	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100	100	100	100	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BBIP1	98,6	92,4	100	100	?Bardet-Biedl syndrome 18, 615995
BBS1	100	100	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	100	99,8	100	100	Bardet-Biedl syndrome 10, 615987

BBS12	100	100	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	100	99,5	100	100	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,9	99,3	100	100	Bardet-Biedl syndrome 4, 615982
BBS5	99	93,9	100	100	Bardet-Biedl syndrome 5, 615983
BBS7	98,7	95,5	100	100	Bardet-Biedl syndrome 7, 615984
BBS9	92,3	90,4	95,8	95,8	Bardet-Biedl syndrome 9, 615986
BCS1L	100	100	100	100	GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BICC1	100	100	100	100	{Renal dysplasia, cystic, susceptibility to}, 601331
BSND	100	100	100	100	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
C3	99,9	99,2	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 C3 deficiency, 613779 {Macular degeneration, age-related, 9}, 611378
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CACNA1H	98,7	96,4	100	99,9	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 Hyperaldosteronism, familial, type IV, 617027
CASR	100	99,9	100	100	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200
CBWD1	20,8	19,4	99,6	99,3	No OMIM disease ID
CC2D2A	98,5	96,5	97,1	97,1	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome 2, 619111
CCNQ	83,1	78,5	98,9	94,6	STAR syndrome, 300707
CD2AP	99,9	98,8	100	100	Glomerulosclerosis, focal segmental, 3, 607832
CD46	99,9	99,4	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922

CEP120	100	99,5	100	100	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	99,9	98,3	100	100	Nephronophthisis 15, 614845
CEP290	96,1	90	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	99,8	97,4	100	100	Joubert syndrome 15, 614464
CEP55	100	99,8	100	100	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	99,8	97,4	100	100	Nephronophthisis 18, 615862
CFB	100	100	100	100	?Complement factor B deficiency, 615561 {Macular degeneration, age-related, 14, reduced risk of}, 615489 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924
CFH	99,9	99	100	99,9	Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 Basal laminar drusen, 126700 {Macular degeneration, age-related, 4}, 610698
CFHR1	96,4	94,9	95,4	93,8	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	94	92,2	96	95,2	{Macular degeneration, age-related, reduced risk of}, 603075 {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFI	99,2	96,8	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 Complement factor I deficiency, 610984 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CHRM3	100	100	100	100	Prune belly syndrome, 100100
CHRNA3	100	99,4	100	100	{Lung cancer susceptibility 2}, 612052 Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CLCN2	100	99,5	100	100	{Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN5	99,9	98,3	100	100	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Dent disease, 300009

					Hypophosphatemic rickets, 300554 Nephrolithiasis, type I, 310468
CLCNKB	99,1	95,9	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	98,5	93,1	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	98,7	97,4	100	100	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 {Hemorrhage, intracerebral, susceptibility to}, 614519 ?Retinal arteries, tortuosity of, 180000 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COL4A3	98,7	98	100	100	Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200 Hematuria, benign familial, 141200
COL4A4	99,9	98,2	100	100	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL4A5	97,8	89,1	100	99,8	Alport syndrome 1, X-linked, 301050
COQ2	98	95,3	97,2	97,2	{Multiple system atrophy, susceptibility to}, 146500 Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	99,9	98,4	100	100	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100	99,8	100	100	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8B	100	99,3	100	100	Nephrotic syndrome, type 9, 615573
COQ9	100	97,9	100	100	Coenzyme Q10 deficiency, primary, 5, 614654
CPLANE1	99,7	98,4	100	100	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CRB2	98,5	93	100	100	Ventriculomegaly with cystic kidney disease, 219730 Focal segmental glomerulosclerosis 9, 616220
CSPP1	99,8	98,7	100	100	Joubert syndrome 21, 615636
CTNS	100	99,8	100	100	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750

					Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CUBN	99,7	98,3	100	100	[Proteinuria, chronic benign], 618884 Imerslund-Grasbeck syndrome 1, 261100
CUL3	99,9	98,8	100	100	Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	100	99,9	100	100	Hypercalcemia, infantile, 1, 143880
DCDC2	100	99,9	100	100	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DGKE	99,8	98,1	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 Nephrotic syndrome, type 7, 615008
DMP1	100	99,9	100	100	Hypophosphatemic rickets, AR, 241520
DNAJB11	100	99,5	100	100	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DSTYK	99,9	99,2	100	100	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DYNC2H1	98,8	95,5	100	100	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	99,5	97	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DZIP1L	99,9	99	100	100	Polycystic kidney disease 5, 617610
EGF	99,9	99,7	100	100	Hypomagnesemia 4, renal, 611718
EHHADH	100	100	100	100	?Fanconi renotubular syndrome 3, 615605
EMP2	99,9	96,7	100	100	Nephrotic syndrome, type 10, 615861
ENPP1	96,4	91,2	98,7	97,8	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 {Obesity, susceptibility to}, 601665 Arterial calcification, generalized, of infancy, 1, 208000 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853
EYA1	99,9	99,7	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	99,6	94,7	100	100	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690

FAN1	100	99,8	100	100	Interstitial nephritis, karyomegalic, 614817
FAT1	100	100	100	100	No OMIM disease ID
FGF23	99,6	97,5	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FN1	100	99,3	100	100	Glomerulopathy with fibronectin deposits 2, 601894 Spondylometaphyseal dysplasia, corner fracture type, 184255
FOXC2	100	96,7	100	99,8	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	99,4	100	100	Fraser syndrome 1, 219000
FREM1	99,9	99,1	100	100	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	100	99,3	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	99,8	99	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GANAB	99,9	99	100	100	Polycystic kidney disease 3, 600666
GAPVD1	100	99,3	100	100	No OMIM disease ID
GATA3	100	100	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GCM2	100	100	100	100	Hyperparathyroidism 4, 617343 Hypoparathyroidism, familial isolated 2, 618883
GFRA1	100	100	100	100	No OMIM disease ID
GLA	91,1	88,2	91,3	91,3	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLI3	98,5	98	100	100	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GLIS2	100	99,8	100	100	Nephronophthisis 7, 611498

GLIS3	98,6	98,2	100	100	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	99,9	95	100	100	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GREB1L	100	99,9	100	100	Renal hypodysplasia/aplasia 3, 617805
GRHPR	84,2	81,3	100	99,3	Hyperoxaluria, primary, type II, 260000
GRIP1	100	99,7	100	100	Fraser syndrome 3, 617667
GSN	95,8	93,5	99,9	99,3	Amyloidosis, Finnish type, 105120
HNF1B	99,3	96,1	100	100	Diabetes mellitus, noninsulin-dependent, 125853 Renal cysts and diabetes syndrome, 137920 {Renal cell carcinoma}, 144700
HNF4A	99,9	99	100	100	{Diabetes mellitus, noninsulin-dependent}, 125853 MODY, type I, 125850 Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026
HOGA1	100	96,4	100	100	Hyperoxaluria, primary, type III, 613616
HPRT1	99,3	91,8	100	99,3	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	86	82,7	99,9	98,1	Apparent mineralocorticoid excess, 218030
IFNG	100	100	100	100	?Immunodeficiency 69, mycobacteriosis, 618963 {AIDS, rapid progression to}, 609423 {TSC2 angiomyolipomas, renal, modifier of}, 613254 {Hepatitis C virus, response to therapy of}, 609532 {Aplastic anemia}, 609135 {Tuberculosis, protection against}, 607948
IFT122	100	99,6	100	100	Cranioectodermal dysplasia 1, 218330
IFT140	99,8	98,8	100	100	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,9	99,1	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	86,7	83,8	100	100	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
INPP5E	97,1	92,7	100	100	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INTU	99,7	98,1	100	100	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	100	100	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	93,9	85	100	100	Senior-Loken syndrome 5, 609254
ITGA3	99,5	97,4	100	100	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA8	99,9	99,7	100	100	Renal hypodysplasia/aplasia 1, 191830
ITSN1	99,4	97,4	100	100	No OMIM disease ID
ITSN2	98,8	96,5	100	100	No OMIM disease ID
JAG1	97,7	96,8	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KANK1	100	100	100	100	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	100	100	100	100	Palmoplantar keratoderma and woolly hair, 616099 Nephrotic syndrome, type 16, 617783
KIAA0556	100	99,9	100	100	Joubert syndrome 26, 616784
KCNJ1	100	100	100	100	Bartter syndrome, type 2, 241200
KCNJ10	89,3	89	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
KIF14	99,6	97,7	100	100	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF7	93,6	90,6	99,1	97,8	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIRREL1	100	99,9	100	100	No OMIM disease ID
KL	98,2	97,2	98,5	97,5	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994

KLHL3	100	99,3	100	100	Pseudohypoaldosteronism, type IID, 614495
LAGE3	95,9	85,1	100	100	Galloway-Mowat syndrome 2, X-linked, 301006
LAMB2	100	99,7	100	100	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCAT	99	93,8	100	100	Norum disease, 245900 Fish-eye disease, 136120
LMX1B	99,6	96,3	100	100	Nail-patella syndrome, 161200 Focal segmental glomerulosclerosis 10, 256020
LRIG2	99,6	98,8	100	100	Urofacial syndrome 2, 615112
LRP2	100	99,9	100	100	Donnai-Barrow syndrome, 222448
LRP4	99,1	98,8	100	100	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	98,5	98,1	100	99,7	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 {Osteoporosis}, 166710 [Bone mineral density variability 1], 601884
LYZ	100	100	100	100	Amyloidosis, renal, 105200
LZTFL1	99,9	99,2	100	100	Bardet-Biedl syndrome 17, 615994
MAFB	100	99,4	100	100	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	99,8	97,7	100	99,9	Bartter syndrome, type 5, antenatal, transient, 300971
MAGI2	94,5	92,4	94,7	93,3	Nephrotic syndrome, type 15, 617609
MAPKBP1	100	100	100	100	Nephronophthisis 20, 617271
MKKS	100	100	100	100	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700

MKS1	99,8	97,9	100	100	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MMACHC	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MOCOS	99,8	97,7	100	100	Xanthinuria, type II, 603592
MYH9	100	99,3	100	100	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO1E	99,9	99,5	100	100	Glomerulosclerosis, focal segmental, 6, 614131
NCAPG2	99,9	99,2	100	100	Khan-Khan-Katsanis syndrome, 618460
NEK1	99,8	98	100	100	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	100	99,9	100	100	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEU1	99,7	97,7	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NOTCH2	100	99,5	100	100	Hajdu-Cheney syndrome, 102500 Alagille syndrome 2, 610205
NPHP1	100	99	100	100	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	99,7	98,4	100	100	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	100	99,8	100	100	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	99,8	99,1	100	100	Nephrotic syndrome, type 1, 256300
NPHS2	100	99,5	100	100	Nephrotic syndrome, type 2, 600995
NR3C2	100	99,7	100	100	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NUP107	99,8	98,5	100	100	Galloway-Mowat syndrome 7, 618348 ?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP133	99,7	98,3	100	100	Nephrotic syndrome, type 18, 618177 ?Galloway-Mowat syndrome 8, 618349

NUP160	100	99,9	100	100	?Nephrotic syndrome, type 19, 618178
NUP205	99,9	99,4	100	100	?Nephrotic syndrome, type 13, 616893
NUP85	100	100	100	100	Nephrotic syndrome, type 17, 618176
NUP93	98	94,2	95,5	95,5	Nephrotic syndrome, type 12, 616892
OCRL	99,9	98,6	100	99,9	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	88	73,7	100	99,9	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OSGEP	100	99,4	100	100	Galloway-Mowat syndrome 3, 617729
PAX2	100	99,9	100	100	Papillorenal syndrome, 120330 Glomerulosclerosis, focal segmental, 7, 616002
PBX1	100	99,4	100	100	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCBD1	100	99,6	100	99,7	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDE6D	100	100	100	100	Joubert syndrome 22, 615665
PDSS2	99,8	97,1	100	100	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	100	99,6	99,9	99,2	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	39,2	30	99,2	98,9	Polycystic kidney disease 1, 173900
PKD2	95,5	91,1	99,3	97,7	Polycystic kidney disease 2, 613095
PKHD1	100	99,6	100	100	Polycystic kidney disease 4, with or without hepatic disease, 263200
PLCE1	99,9	99,3	100	100	Nephrotic syndrome, type 3, 610725
PMM2	100	100	100	100	Congenital disorder of glycosylation, type Ia, 212065
PTH1R	100	98,7	100	100	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTPRO	99,9	99,4	100	100	Nephrotic syndrome, type 6, 614196

RAD21	99,2	96,6	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 [Hyperproreninemia], 0
RMND1	100	98,6	100	100	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	99,4	97,8	100	100	Vesicoureteral reflux 2, 610878
RPGRIP1L	96,7	95,7	100	99,5	?COACH syndrome 3, 619113 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RRM2B	100	99,7	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	99,9	99	100	100	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	98,6	96,7	100	100	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SARS2	95,8	94,6	100	100	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	100	99,8	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	99,7	98,2	100	100	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100	99,7	100	100	Bronchiectasis with or without elevated sweat chloride 1, 211400 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200
SCNN1G	99,8	98,3	100	100	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350
SDCCAG8	100	99,9	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	95,4	88,2	100	100	Branchiootorenal syndrome 2, 610896
SLC12A1	96,2	96,1	96,2	96,2	Bartter syndrome, type 1, 601678

SLC12A3	100	99,9	100	100	Gitelman syndrome, 263800
SLC16A12	100	99,9	100	100	Cataract 47, juvenile, with microcornea, 612018
SLC22A12	100	99,8	100	100	Hypouricemia, renal, 220150
SLC26A1	100	99,6	100	100	?Nephrolithiasis, calcium oxalate, 167030
SLC26A3	100	99,5	100	100	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	100	100	100	100	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	99,8	96,1	100	100	{Uric acid concentration, serum, QTL 2}, 612076 Hypouricemia, renal, 2, 612076
SLC34A1	99,9	99,1	100	100	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Hypercalcemia, infantile, 2, 616963 ?Fanconi renotubular syndrome 2, 613388
SLC34A3	100	99,4	100	100	Hypophosphatemic rickets with hypercalciuria, 241530
SLC36A2	100	100	100	100	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	100	99,2	100	100	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC3A1	100	99,8	96,6	96,6	Cystinuria, 220100
SLC41A1	100	100	100	100	No OMIM disease ID
SLC4A1	100	99,8	96,1	96,1	[Blood group, Swann], 601550 [Blood group, Froese], 601551 [Blood group, Waldner], 112010 Spherocytosis, type 4, 612653 Cryohydrocytosis, 185020 Distal renal tubular acidosis 4 with hemolytic anemia, 611590 Ovalocytosis, SA type, 166900 [Malaria, resistance to], 611162 [Blood group, Diego], 110500 Distal renal tubular acidosis 1, 179800 [Blood group, Wright], 112050
SLC4A4	99,8	99,2	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 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A20	100	99,9	100	100	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700
SLC7A9	100	99,9	100	100	Cystinuria, 220100
SLC9A3	90,6	86	96,4	94,1	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	100	98,7	100	100	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLIT3	97,9	95,3	100	100	No OMIM disease ID
SMARCAL1	100	99,9	100	100	Schimke immunoosseous dysplasia, 242900
SOX17	100	99,5	100	100	Vesicoureteral reflux 3, 613674
STRA6	100	99,8	100	100	Microphthalmia, isolated, with coloboma 8, 601186 Microphthalmia, syndromic 9, 601186
STX16	100	98,6	100	100	Pseudohypoparathyroidism, type IB, 603233
TBC1D8B	98,5	93,2	100	100	Nephrotic syndrome, type 20, 301028
TBX18	99,5	97,1	100	100	Congenital anomalies of kidney and urinary tract 2, 143400
TCTN1	96,7	93	94,7	94,7	Joubert syndrome 13, 614173
TCTN2	100	99,5	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	99,7	100	100	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
TMEM107	100	100	100	100	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM138	100	99,1	100	100	Joubert syndrome 16, 614465
TMEM216	99,9	98,1	100	100	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	100	99,6	100	100	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970

TMEM237	100	99,9	100	100	Joubert syndrome 14, 614424
TMEM260	97,5	93,4	100	100	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	99,5	95	100	99,9	Meckel syndrome 3, 607361 COACH syndrome 1, 216360 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991 Joubert syndrome 6, 610688
TNS2	100	99,9	100	100	No OMIM disease ID
TNXB	99,1	93,7	100	99,9	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP53RK	92,5	79,6	100	100	Galloway-Mowat syndrome 4, 617730
TPRKB	81,1	75,9	81,9	81,9	Galloway-Mowat syndrome 5, 617731
TRAF3IP1	99,6	97,6	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	98,2	96,1	100	100	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	99,9	99,5	100	100	Hypomagnesemia 1, intestinal, 602014
TSC1	99,8	98,8	100	100	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100	99,6	100	100	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TTC21B	99,9	99,3	100	100	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC8	99,6	98,1	100	100	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
UMOD	97,7	96,2	100	100	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 Hyperuricemic nephropathy, familial juvenile 1, 162000 Medullary cystic kidney disease 2, 603860
UPK3A	100	99,5	100	100	No OMIM disease ID
UQCC2	100	99,7	100	100	Mitochondrial complex III deficiency, nuclear type 7, 615824

VDR	97,2	94,9	98,2	95,2	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	99,4	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	99,8	98,9	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR73	100	100	100	100	Galloway-Mowat syndrome 1, 251300
WNK1	99,9	99,6	100	100	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
WNK4	99,9	99,3	100	100	Pseudohypoaldosteronism, type IIB, 614491
WNT4	99,1	94,8	98,9	96,2	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	97,3	95,4	97,7	97,7	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	99,9	100	100	Xanthinuria, type I, 278300
XPNPEP3	100	100	100	100	Nephronophthisis-like nephropathy 1, 613159
XPO5	100	99,9	100	99,7	No OMIM disease ID
ZMPSTE24	100	99,9	100	100	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
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 : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors