RENAL DISORDERS GENE PANEL DG 3.2.0 (317 genes)

Releasedate: 16-09-2021

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	99,9	Renal tubular dysgenesis, 267430
ACTN4	99,9	98	100	100	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	97	94,3	99,8	98,9	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS9	99,4	98,4	100	100	No OMIM disease ID
ADCY10	100	99,6	100	100	No OMIM disease ID
AGT	100	99,9	100	100	Renal tubular dysgenesis, 267430
AGTR1	91,9	91,8	100	100	Renal tubular dysgenesis, 267430
AGXT	100	100	100	100	Hyperoxaluria, primary, type 1, 259900
AHI1	99,4	97,4	100	100	Joubert syndrome 3, 608629
ALDOB	98,8	95,7	100	100	Fructose intolerance, hereditary, 229600
ALG1	53,6	46,9	100	100	Congenital disorder of glycosylation, type Ik, 608540
ALG8	96,6	95,9	96,6	96,6	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	99,9	99,3	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALMS1	99,7	99,5	100	100	Alstrom syndrome, 203800
AMN	92,5	82,9	100	100	Imerslund-Grasbeck syndrome 2, 618882
ANKFY1	100	98,7	100	100	No OMIM disease ID
ANKS6	94,2	89,7	97	95	Nephronophthisis 16, 615382
ANLN	98,4	97	100	100	Focal segmental glomerulosclerosis 8, 616032
ANOS1	89,8	88,3	99,9	99,4	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AP2S1	90,4	90	100	100	Hypocalciuric hypercalcemia, type III, 600740
APOL1	100	100	100	100	No OMIM disease ID
APRT	100	100	100	100	Adenine phosphoribosyltransferase deficiency, 614723

AQP2	100	99,2	100	100	Diabetes insipidus, nephrogenic, 2, 125800
ARHGAP24	100	100	100	100	No OMIM disease ID
ARHGDIA	100	100	100	100	Nephrotic syndrome, type 8, 615244
ARL13B	100	99,3	100	100	Joubert syndrome 8, 612291
ARL6	99,1	98,4	100	100	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ATP1A1	100	99,8	100	100	Hypomagnesemia, seizures, and mental retardation 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP6V0A4	100	99,3	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	99,8	100	100	Nephrotic syndrome, type 21, 618594
AVP	90,2	66,6	100	100	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	100	99,8	100	100	Diabetes insipidus, nephrogenic, 1, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	85,2	85,2	95,8	94	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100	100	100	100	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BBIP1	95,7	87,4	100	100	?Bardet-Biedl syndrome 18, 615995
BBS1	100	100	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	100	99,9	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	100	100	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	99,4	98	100	100	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981
BBS4	99,9	98,9	100	99,9	Bardet-Biedl syndrome 4, 615982
BBS5	98,4	94,7	100	100	Bardet-Biedl syndrome 5, 615983
BBS7	99	96,5	100	99,9	Bardet-Biedl syndrome 7, 615984
BBS9	92	89	95,8	95,8	Bardet-Biedl syndrome 9, 615986
BCS1L	100	100	100	100	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000

BICC1	100	100	100	100	No OMIM disease ID
BSND	100	99,9	100	100	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
C3	99,9	98,5	100	100	C3 deficiency, 613779
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CACNA1H	98,9	96,3	100	99,9	Hyperaldosteronism, familial, type IV, 617027
CASR	100	99,5	100	100	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalciuric hypercalcemia, type I, 145980
CBWD1	20,5	19,1	99,4	98,5	No OMIM disease ID
CC2D2A	98,3	96,6	97,1	97	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCNQ	82,9	78,3	99,8	98,2	STAR syndrome, 300707
CD151	100	100	100	100	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057
CD2AP	99,6	98,8	100	99,9	Glomerulosclerosis, focal segmental, 3, 607832
CD46	99,7	98,9	100	99,9	No OMIM disease ID
CEP120	99,9	99,6	100	100	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP164	99,8	98,2	100	100	Nephronophthisis 15, 614845
CEP290	96,2	90,8	100	99,9	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134
CEP41	98,8	93,4	100	100	Joubert syndrome 15, 614464
CEP55	100	99,8	100	100	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	99	96,6	100	99,9	Nephronophthisis 18, 615862
CFB	100	99,6	100	100	?Complement factor B deficiency, 615561
CFH	99,8	98,5	100	99,9	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	91,7	89,6	96,3	94,1	No OMIM disease ID

CFHR3	89	87,8	97,7	96	No OMIM disease ID
CFHR5	99,8	97,6	100	100	Nephropathy due to CFHR5 deficiency, 614809
CFI	99,3	96	100	99,9	Complement factor I deficiency, 610984
CHRM3	100	100	100	100	Prune belly syndrome, 100100
CHRNA3	100	99,2	100	100	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CLCN2	100	99,3	100	100	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN5	99,7	97,1	100	99,9	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Hypophosphatemic rickets, 300554 Dent disease 1, 300009 Nephrolithiasis, type I, 310468
CLCNKB	98,7	95,3	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	98,3	92,9	100	100	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	100	99,9	100	100	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
COL4A1	99	97	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	98,9	97,4	100	100	Hematuria, benign familial, 141200 Alport syndrome 3, autosomal dominant, 104200 Alport syndrome 2, autosomal recessive, 203780
COL4A4	99,6	97,4	100	100	Hematuria, familial benign, 141200 Alport syndrome 2, autosomal recessive, 203780
COL4A5	97,6	86,6	100	99,8	Alport syndrome 1, X-linked, 301050
COQ2	97,6	96,7	97,2	97,2	Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	99,9	98,5	100	100	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100	99,6	100	100	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8B	100	99,2	100	100	Nephrotic syndrome, type 9, 615573
COQ9	100	98,7	100	100	Coenzyme Q10 deficiency, primary, 5, 614654

CPLANE1	99,4	98,2	100	100	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CRB2	98,9	94,2	100	100	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CSPP1	99,7	98,1	100	100	Joubert syndrome 21, 615636
CTNS	100	99,3	100	100	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CUBN	99,2	97,1	100	100	Imerslund-Grasbeck syndrome 1, 261100
CUL3	99,4	97,4	100	100	Neurodevelopmental disorder with or without autism or seizures, 619239 Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	100	100	100	100	Hypercalcemia, infantile, 1, 143880
DAAM2	99,1	98,2	100	100	Nephrotic syndrome, type 24, 619263
DCDC2	100	99,9	100	100	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394
DGKE	99,7	98,5	100	100	Nephrotic syndrome, type 7, 615008
DLC1	100	99,9	100	100	Colorectal cancer, somatic, 114500
DMP1	99,9	99,9	100	100	Hypophosphatemic rickets, AR, 241520
DNAJB11	99,9	99,6	100	100	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DSTYK	99,9	98,8	100	100	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DYNC2H1	98,6	95,2	100	99,8	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	99,3	95,8	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DZIP1L	99,8	98	100	100	Polycystic kidney disease 5, 617610
EGF	99,9	99,8	100	100	?Hypomagnesemia 4, renal, 611718
EHHADH	100	100	100	100	?Fanconi renotubular syndrome 3, 615605
EMP2	98,9	93	100	100	Nephrotic syndrome, type 10, 615861
ENPP1	96,5	90,6	98,8	97,8	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522

EYA1	99,9	99,5	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	99,5	100	99,9	Tyrosinemia, type I, 276700
FAM20A	99,6	94,4	100	100	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAN1	100	99,8	100	100	Interstitial nephritis, karyomegalic, 614817
FAT1	100	99,9	100	100	No OMIM disease ID
FGF23	99,4	96,7	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FN1	99,9	98,7	100	100	Spondylometaphyseal dysplasia, corner fracture type, 184255 Glomerulopathy with fibronectin deposits 2, 601894
FOXC2	100	98,1	100	99,6	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	99,2	100	100	Fraser syndrome 1, 219000
FREM1	99,8	98,4	100	100	Manitoba oculotrichoanal syndrome, 248450 Bifid nose with or without anorectal and renal anomalies, 608980 Trigonocephaly 2, 614485
FREM2	99,8	98,7	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	98,7	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GANAB	99,8	97,8	100	100	Polycystic kidney disease 3, 600666
GAPVD1	99,9	98,9	100	100	No OMIM disease ID
GATA3	100	100	100	100	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GCM2	100	100	100	100	Hypoparathyroidism, familial isolated 2, 618883 Hyperparathyroidism 4, 617343
GFRA1	100	99,9	100	100	No OMIM disease ID
GLA	91	85,9	91,3	91,3	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLI3	98,5	97,7	100	100	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200

					Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GLIS2	100	99,9	100	100	Nephronophthisis 7, 611498
GLIS3	98,5	97,4	100	100	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	98,4	93,1	100	100	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GREB1L	100	99,9	100	100	Deafness, autosomal dominant 80, 619274 Renal hypodysplasia/aplasia 3, 617805
GRHPR	83,3	79,2	100	99,3	Hyperoxaluria, primary, type II, 260000
GRIP1	100	99,3	100	100	Fraser syndrome 3, 617667
GSN	95,7	93,5	100	99,7	Amyloidosis, Finnish type, 105120
HNF1A	100	99,8	100	100	Hepatic adenoma, somatic, 142330 Diabetes mellitus, insulin-dependent, 20, 612520 MODY, type III, 600496 Renal cell carcinoma, 144700
HNF1B	99	95,7	100	100	Type 2 diabetes mellitus, 125853 Renal cysts and diabetes syndrome, 137920
HNF4A	99,9	98,6	100	100	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850
HOGA1	99,5	95,5	100	100	Hyperoxaluria, primary, type III, 613616
HPRT1	98,6	90,6	99,5	98,4	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HSD11B2	87,6	83,8	99,9	97,6	Apparent mineralocorticoid excess, 218030
IFNG	100	99,9	100	100	?Immunodeficiency 69, mycobacteriosis, 618963
IFT122	99,9	99,2	100	100	Cranioectodermal dysplasia 1, 218330
IFT140	99,9	99,2	100	100	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	99,6	98,6	100	100	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 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 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866

INF2	87,2	84,8	100	100	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
INPP5E	96,9	93,2	100	100	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INTU	99,9	98,6	100	100	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100	99,8	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92,8	82,8	100	100	Senior-Loken syndrome 5, 609254
ITGA3	99,6	97,9	100	100	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA8	99,9	99,3	100	100	Renal hypodysplasia/aplasia 1, 191830
ITSN1	98,7	96	100	100	No OMIM disease ID
ITSN2	97,8	95,4	100	100	No OMIM disease ID
JAG1	97,8	96,7	100	100	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
KANK1	100	99,9	100	100	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	100	100	100	100	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KIAA0556	100	99,6	100	100	Joubert syndrome 26, 616784
KCNJ1	100	100	100	100	Bartter syndrome, type 2, 241200
KCNJ10	89,2	88,5	100	100	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ16	98,3	98,3	100	100	Hypokalemic tubulopathy and deafness, 619406
KCNJ5	100	100	100	100	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KIF14	99,2	96,8	100	99,9	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF7	93,6	91,9	99,7	98,6	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KIRREL1	100	99,9	100	100	Nephrotic syndrome, type 23, 619201
KL	98,5	97,5	98,7	97,9	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLHL3	100	98,9	100	100	Pseudohypoaldosteronism, type IID, 614495

LAGE3	96,1	84,3	100	100	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA5	98,5	96,3	100	99,9	No OMIM disease ID
LAMB2	99,9	99,3	100	100	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LCAT	98,8	93,3	100	100	Fish-eye disease, 136120 Norum disease, 245900
LMX1B	99,3	96,8	100	100	Focal segmental glomerulosclerosis 10, 256020 Nail-patella syndrome, 161200
LRIG2	99,8	99,2	100	100	Urofacial syndrome 2, 615112
LRP2	100	99,8	100	100	Donnai-Barrow syndrome, 222448
LRP4	99,1	98,4	100	100	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	99,2	98,2	99,8	99,2	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	99,9	100	100	Amyloidosis, renal, 105200
LZTFL1	99,7	99,4	100	99,9	Bardet-Biedl syndrome 17, 615994
MAFB	100	99,8	100	100	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	99,5	97,6	100	99,8	Bartter syndrome, type 5, antenatal, transient, 300971
MAGI2	94,2	91	94,7	93,3	Nephrotic syndrome, type 15, 617609
MAPKBP1	100	100	100	100	Nephronophthisis 20, 617271
MKKS	100	100	100	100	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKS1	99,4	96,3	100	100	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MMACHC	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MOCOS	99,9	97,8	100	100	Xanthinuria, type II, 603592

MYH9	99,9	98,9	100	100	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYO1E	99,9	98,6	100	100	Glomerulosclerosis, focal segmental, 6, 614131
NCAPG2	99,8	99	100	100	Khan-Khan-Katsanis syndrome, 618460
NEK1	99,5	98,2	100	99,9	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	100	99,8	100	100	Renal-hepatic-pancreatic dysplasia 2, 615415 ?Nephronophthisis 9, 613824
NEU1	99,3	96,1	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NOS1AP	100	100	100	100	Nephrotic syndrome, type 22, 619155
NOTCH2	100	99,2	100	100	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NPHP1	99,8	99,1	100	100	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	99,6	98,5	100	99,9	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	100	99,8	100	100	Senior-Loken syndrome 4, 606996 Nephronophthisis 4, 606966
NPHS1	99,7	99	100	100	Nephrotic syndrome, type 1, 256300
NPHS2	100	99,6	100	99,9	Nephrotic syndrome, type 2, 600995
NR3C2	99,9	99,8	100	100	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NUP107	99,7	98,4	100	99,9	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348 Nephrotic syndrome, type 11, 616730
NUP133	99,4	97,3	100	100	?Galloway-Mowat syndrome 8, 618349 Nephrotic syndrome, type 18, 618177
NUP160	100	99,8	100	100	?Nephrotic syndrome, type 19, 618178
NUP205	99,9	99,3	100	99,9	?Nephrotic syndrome, type 13, 616893
NUP85	100	100	100	100	Nephrotic syndrome, type 17, 618176
NUP93	96,7	92,7	95,5	95,5	Nephrotic syndrome, type 12, 616892

NXF5	58,3	57	99,9	99,9	No OMIM disease ID
OCRL	99,4	97,6	100	99,9	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	87,1	71,3	100	99,8	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
OSGEP	99,8	95,7	100	100	Galloway-Mowat syndrome 3, 617729
PAX2	100	100	100	100	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330
PBX1	100	99,1	100	100	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCBD1	100	99,8	100	100	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDE6D	100	99,9	100	100	Joubert syndrome 22, 615665
PDSS2	98,4	94,3	100	100	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	99,9	98,9	100	99,2	Hypophosphatemic rickets, X-linked dominant, 307800
PKD1	40,6	32,8	99,3	99	Polycystic kidney disease 1, 173900
PKD2	96	93,3	99,6	97,9	Polycystic kidney disease 2, 613095
PKHD1	100	99,6	100	100	Polycystic kidney disease 4, with or without hepatic disease, 263200
PLCE1	99,8	98,9	100	100	Nephrotic syndrome, type 3, 610725
PMM2	99,8	99,8	100	100	Congenital disorder of glycosylation, type Ia, 212065
PODXL	94,3	94	94,4	94,3	No OMIM disease ID
PTH1R	99,6	95,9	100	100	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Chondrodysplasia, Blomstrand type, 215045
PTPRO	99,7	98,9	100	100	Nephrotic syndrome, type 6, 614196
RAD21	99,2	95,9	100	100	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
REN	100	100	100	100	Renal tubular dysgenesis, 267430 Tubulointerstitial kidney disease, autosomal dominant, 4, 613092
RMND1	99,7	97,2	100	99,9	Combined oxidative phosphorylation deficiency 11, 614922
ROBO2	99,1	97,3	100	100	Vesicoureteral reflux 2, 610878

RPGRIP1L	96,5	95,3	100	99,4	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RRM2B	100	99,8	100	99,9	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
SALL1	99,7	97,5	100	100	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	99,1	96,4	100	100	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323
SARS2	95,7	94,5	100	100	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	99,9	99,4	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	99,7	97,5	100	100	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100	99,8	100	100	Bronchiectasis with or without elevated sweat chloride 1, 211400 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200
SCNN1G	99,8	98,4	100	100	Bronchiectasis with or without elevated sweat chloride 3, 613071 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 2, 618114
SDCCAG8	99,8	99,8	100	100	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993
SEC61A1	100	100	100	100	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SGPL1	100	100	100	100	Nephrotic syndrome, type 14, 617575
SIX5	96,9	90,1	100	100	Branchiootorenal syndrome 2, 610896
SLC12A1	96,2	96	96,2	96,2	Bartter syndrome, type 1, 601678
SLC12A3	100	100	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	99,7	100	100	?Nephrolithiasis, calcium oxalate, 167030
SLC26A3	100	99,5	100	100	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	100	99,8	100	100	Fanconi-Bickel syndrome, 227810
SLC2A9	99,3	95	100	100	Hypouricemia, renal, 2, 612076

SLC34A1	100	99,7	100	100	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	100	99,3	100	100	Hypophosphatemic rickets with hypercalciuria, 241530
SLC36A2	100	99,8	99,9	99,8	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC37A4	99,8	97,6	100	100	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC3A1	100	99,7	96,6	96,6	Cystinuria, 220100
SLC41A1	100	99,9	100	100	?Nephronophthisis-like nephropathy 2, 619468
SLC4A1	100	99,9	96,1	96,1	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	99,9	99,4	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	99,8	100	100	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700
SLC7A9	100	99,4	100	100	Cystinuria, 220100
SLC9A3	90,5	86	96	93,6	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	99,9	98,2	100	100	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLIT3	97,9	95,6	100	100	No OMIM disease ID
SMARCAL1	100	99,8	100	100	Schimke immunoosseous dysplasia, 242900
SOX17	100	99,9	100	100	Vesicoureteral reflux 3, 613674
STRA6	100	99,9	100	100	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STX16	100	99	100	100	Pseudohypoparathyroidism, type IB, 603233
TBC1D8B	98	92,1	100	99,7	Nephrotic syndrome, type 20, 301028

TBX18	99,5	97,1	100	100	Congenital anomalies of kidney and urinary tract 2, 143400
TCTN1	96,8	92,8	94,7	94,7	Joubert syndrome 13, 614173
TCTN2	99,9	99,1	100	100	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100	100	100	100	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
THBD	100	99,9	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	99,8	93,1	100	100	Joubert syndrome 16, 614465
TMEM216	98,5	92,8	100	100	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	100	99,3	100	100	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	99,8	99,3	100	100	Joubert syndrome 14, 614424
TMEM260	98,7	95,4	100	100	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	98,6	93,5	100	99,6	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TNS2	100	100	100	100	No OMIM disease ID
TNXB	98,7	93,9	100	100	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP53RK	95,5	84,9	100	100	Galloway-Mowat syndrome 4, 617730
TPRKB	80,2	75,2	81,9	81,7	Galloway-Mowat syndrome 5, 617731
TRAF3IP1	98,7	95,4	100	100	Senior-Loken syndrome 9, 616629
TRIM32	100	99,9	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRPC6	97,1	94,5	100	100	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	99,9	99,1	100	100	Hypomagnesemia 1, intestinal, 602014

TSC1	99,5	98,2	100	100	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
TSC2	100	99,8	100	100	Lymphangioleiomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254
TTC21B	99,7	99,1	100	99,9	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 Nephronophthisis 12, 613820
TTC8	99,5	98	100	100	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
UMOD	97,5	95,9	100	100	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000
UPK3A	100	99,7	100	100	No OMIM disease ID
UQCC2	99,9	98,5	100	100	Mitochondrial complex III deficiency, nuclear type 7, 615824
VDR	96,7	94,4	99,5	97,7	Rickets, vitamin D-resistant, type IIA, 277440
VIPAS39	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100	99,9	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	99,8	98,6	100	99,9	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Cranioectodermal dysplasia 4, 614378
WDR35	99,6	98,4	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	99,8	99,3	100	100	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	99,9	99	100	100	Pseudohypoaldosteronism, type IIB, 614491
WNT4	97,8	93,6	99,3	96,5	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	97,6	96,1	97,7	97,7	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	99,8	100	100	Xanthinuria, type I, 278300

XPNPEP3	100	100	100	100	Nephronophthisis-like nephropathy 1, 613159
XPO5	100	99,5	99,9	99,7	No OMIM disease ID
ZMPSTE24	99,6	99,4	100	99,9	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
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.
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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-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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors