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RENAL DISORDERS PANEL DG-5.1.0 (328 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
ACE	100%	100%	100%	99.8%	98.5%	Renal tubular dysgenesis, 267430;{Microvascular complications of diabetes 3}, 612624;[Angiotensin I-converting enzyme, benign serum increase], 621420
ACTN4	93%	92.9%	100%	99.9%	99.1%	Glomerulosclerosis, focal segmental, 1, 603278
ADAMTS13	100%	100%	100%	100%	99.4%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS9	100%	100%	100%	99.8%	98.8%	
ADCY10	100%	100%	100%	100%	99.9%	{Hypercalciuria, absorptive, susceptibility to}, 143870
AGT	95.7%	95.3%	100%	100%	99.5%	Renal tubular dysgenesis, 267430
AGTR1	100%	100%	100%	100%	99.9%	{Hypertension, essential}, 145500;Renal tubular dysgenesis, 267430
AGXT	97.4%	96.4%	100%	100%	99.5%	Hyperoxaluria, primary, type 1, 259900
AHI1	98.7%	98.7%	100%	99.9%	99.6%	Joubert syndrome 3, 608629
AIRE	100%	100%	100%	99.8%	99.1%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300

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ALDOB	100%	100%	100%	99.9%	99.8%	Fructose intolerance, hereditary, 229600
ALG1	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ik, 608540
ALG5	91.6%	91.6%	100%	100%	99.9%	Polycystic kidney disease 7, 620056
ALG8	78.4%	77.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	100%	99.9%	99.5%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776
ALMS1	100%	100%	100%	100%	99.8%	Alstrom syndrome, 203800
AMN	100%	100%	100%	99.9%	99.1%	Imerslund-Grasbeck syndrome 2, 618882
ANKFY1	100%	100%	100%	100%	99.5%	
ANKS6	100%	99.9%	100%	100%	99.5%	Nephronophthisis 16, 615382
ANLN	98.2%	98.2%	100%	100%	99.7%	Focal segmental glomerulosclerosis 8, 616032
ANOS1	100%	100%	98.7%	89.8%	71.3%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AP2S1	82.5%	82.5%	100%	99.9%	98.8%	Hypocalciuric hypercalcemia, type III, 600740
APOL1	100%	100%	100%	100%	99.5%	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551

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APRT	100%	100%	100%	100%	99.1%	Adenine phosphoribosyltransferase deficiency, 614723
AQP2	100%	100%	100%	100%	99.8%	Diabetes insipidus, nephrogenic, 2, 125800
ARHGAP24	100%	100%	100%	99.9%	99.2%	
ARHGDIA	100%	100%	100%	100%	99%	Nephrotic syndrome, type 8, 615244
ARHGEF6	93.6%	93.6%	99.2%	90.8%	73.8%	
ARL13B	93.4%	93.3%	100%	100%	99.7%	Joubert syndrome 8, 612291
ARL6	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
ATP1A1	97.2%	95.4%	100%	100%	99.6%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP6V0A4	100%	100%	100%	99.9%	99%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1B1	100%	100%	100%	100%	99.5%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP7B	100%	100%	100%	100%	99.6%	Wilson disease, 277900
AVIL	100%	100%	100%	100%	99.7%	Nephrotic syndrome, type 21, 618594
AVP	100%	100%	100%	100%	99.8%	Diabetes insipidus, neurohypophyseal, 125700

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AVPR2	100%	100%	98.8%	89%	71.9%	Diabetes insipidus, nephrogenic, 1, 304800;Nephrogenic syndrome of inappropriate antidiuresis, 300539
B9D1	100%	100%	100%	100%	99.4%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120
B9D2	100%	100%	100%	100%	99.7%	?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175
BBIP1	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 18, 615995
BBS1	93.7%	93.7%	100%	100%	99.3%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	100%	100%	99.8%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	98%	98%	100%	100%	99.8%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 7, 615984
BBS9	91.9%	91.9%	100%	100%	99.7%	Bardet-Biedl syndrome 9, 615986
BCS1L	100%	100%	100%	100%	99.6%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BICC1	100%	100%	100%	100%	99.8%	{Renal dysplasia, cystic, susceptibility to}, 601331

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BSND	100%	100%	100%	100%	99.8%	Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522
C3	97%	97%	100%	99.9%	99.2%	C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378
CA2	100%	100%	100%	100%	99.8%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CACNA1H	99.6%	99%	100%	99.9%	99%	{Epilepsy, childhood absence, susceptibility to, 6}, 611942;Hyperaldosteronism, familial, type IV, 617027;{Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CASR	100%	100%	100%	100%	99.5%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;{?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CC2D2A	98.2%	98.2%	100%	100%	99.8%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CCNQ	96.7%	96.7%	99.5%	90.1%	70.1%	STAR syndrome, 300707

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CD151	100%	100%	100%	100%	99.7%	[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD2AP	100%	100%	100%	100%	99.8%	Glomerulosclerosis, focal segmental, 3, 607832
CD46	100%	100%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CDC73	100%	100%	100%	99.9%	99.4%	Hyperparathyroidism, familial primary, 145000;Parathyroid adenoma with cystic changes, 145001;Parathyroid carcinoma, 608266;Hyperparathyroidism-jaw tumor syndrome, 145001
CEP120	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761
CEP164	100%	100%	100%	100%	99.5%	Nephronophthisis 15, 614845
CEP290	100%	100%	100%	100%	99.8%	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%	100%	100%	99.8%	Joubert syndrome 15, 614464

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CEP55	100%	100%	100%	100%	99.9%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	100%	100%	100%	100%	99.8%	Nephronophthisis 18, 615862
CFB	100%	100%	100%	100%	99.4%	Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	97.5%	97.5%	100%	100%	99.9%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFHR1	98.3%	96.4%	91.7%	87.3%	76.3%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR3	98%	96.9%	93.2%	89.3%	78.5%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR5	100%	100%	100%	100%	99.8%	Nephropathy due to CFHR5 deficiency, 614809

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CFI	98.6%	95.9%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984
CHRM3	100%	100%	100%	100%	99.8%	Prune belly syndrome, 100100
CHRNA3	92%	92%	100%	99.9%	99.4%	{Lung cancer susceptibility 2}, 612052;Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CLCN2	100%	99.1%	100%	100%	99.6%	Leukoencephalopathy with ataxia, 615651;Hyperaldosteronism, familial, type II, 605635;{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628;{Epilepsy, juvenile absence, susceptibility to, 2}, 607628;{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628
CLCN5	100%	100%	99.4%	92.1%	73.4%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990;Hypophosphatemic rickets, 300554;Dent disease 1, 300009;Nephrolithiasis, type I, 310468
CLCNKB	98.6%	97.4%	100%	100%	99.5%	Bartter syndrome, type 3, 607364;Bartter syndrome, type 4b, digenic, 613090
CLDN10	100%	100%	100%	100%	99.8%	HELIX syndrome, 617671

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CLDN16	95%	95%	100%	100%	99.8%	Hypomagnesemia 3, renal, 248250
CLDN19	100%	100%	100%	100%	99.5%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CNNM2	100%	100%	100%	100%	99.4%	Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418
COL4A1	100%	100%	100%	100%	99.7%	?Retinal arteries, tortuosity of, 180000;{Hemorrhage, intracerebral, susceptibility to}, 614519;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%	100%	100%	99.6%	Alport syndrome 3A, autosomal dominant, 104200;Hematuria, benign familial, 2, 620320;Alport syndrome 3B, autosomal recessive, 620536
COL4A4	99.7%	98.9%	100%	100%	99.7%	Hematuria, familial benign, 1, 141200;Alport syndrome 2, autosomal recessive, 203780
COL4A5	100%	100%	99.2%	91.7%	73%	Alport syndrome 1, X-linked, 301050

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COQ2	96.3%	96.3%	100%	100%	99.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ6	100%	100%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	100%	100%	100%	99.5%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
COQ8B	92.1%	92.1%	100%	100%	99.6%	Nephrotic syndrome, type 9, 615573
COQ9	91.9%	91.9%	100%	99.9%	99.2%	Coenzyme Q10 deficiency, primary, 5, 614654
CPLANE1	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CRB2	100%	100%	100%	100%	99.6%	Focal segmental glomerulosclerosis 9, 616220;Ventriculomegaly with cystic kidney disease, 219730
CSPP1	96.9%	96.9%	100%	100%	99.6%	Joubert syndrome 21, 615636
CTNS	100%	100%	100%	100%	99.4%	Cystinosis, nephropathic, 219800;Cystinosis, ocular nonnephropathic, 219750;Cystinosis, late-onset juvenile or adolescent nephropathic, 219900;Cystinosis, atypical nephropathic, 219800

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CUBN	100%	100%	100%	100%	99.8%	[Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100
CUL3	97.6%	97.6%	100%	100%	99.8%	Neurodevelopmental disorder with or without autism or seizures, 619239;Pseudohypoaldosteronism, type IIE, 614496
CYP24A1	100%	100%	100%	100%	99.6%	Hypercalcemia, infantile, 1, 143880
DAAM2	100%	100%	100%	100%	99.5%	Nephrotic syndrome, type 24, 619263
DCDC2	100%	100%	100%	100%	99.8%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394
DGKE	100%	100%	100%	100%	99.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008
DLC1	100%	100%	100%	100%	99.6%	Colorectal cancer, somatic, 114500
DMP1	100%	100%	100%	100%	99.1%	Hypophosphatemic rickets, AR, 241520
DNAJB11	100%	100%	100%	100%	99.8%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DSTYK	100%	100%	100%	100%	99.5%	Spastic paraplegia 23, autosomal recessive, 270750;Congenital anomalies of kidney and urinary tract 1, 610805
DYNC2H1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091

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DYNC2I1	95.7%	95.7%	100%	100%	99.5%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DZIP1L	100%	100%	100%	100%	99.7%	Polycystic kidney disease 5, 617610
EGF	100%	100%	100%	100%	99.8%	?Hypomagnesemia 4, renal, 611718
EHHADH	100%	100%	100%	100%	99.8%	?Fanconi renotubular syndrome 3, 615605
EMP2	100%	100%	100%	100%	98.7%	Nephrotic syndrome, type 10, 615861
ENPP1	100%	100%	100%	100%	99.8%	{Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522
EYA1	100%	100%	100%	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	95.2%	93.3%	100%	100%	99.5%	Tyrosinemia, type I, 276700
FAM111A	100%	100%	100%	100%	99.9%	Kenny-Caffey syndrome, type 2, 127000;Gracile bone dysplasia, 602361
FAM20A	100%	99.9%	100%	99.8%	99%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690

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FAN1	95.9%	94%	100%	100%	99.6%	Interstitial nephritis, karyomegalic, 614817
FAT1	100%	100%	100%	100%	99.7%	
FGF23	100%	100%	100%	100%	99.2%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100
FN1	100%	100%	100%	100%	99.7%	Spondylometaphyseal dysplasia, corner fracture type, 184255;Glomerulopathy with fibronectin deposits 2, 601894
FOXC2	100%	100%	100%	99.8%	98.2%	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXI1	100%	100%	100%	100%	99.8%	Enlarged vestibular aqueduct, 600791
FRAS1	100%	100%	100%	100%	99.7%	Fraser syndrome 1, 219000
FREM1	100%	100%	100%	100%	99.6%	Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485
FREM2	100%	100%	100%	100%	99.7%	Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570
FXYD2	80.6%	80.4%	100%	99.7%	99%	Hypomagnesemia 2, renal, 154020
G6PC1	100%	100%	100%	100%	99.7%	Glycogen storage disease Ia, 232200

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GALNT3	100%	100%	100%	100%	99.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GANAB	100%	100%	100%	100%	99.6%	Polycystic kidney disease 3, 600666
GAPVD1	100%	100%	100%	100%	99.8%	
GATA3	100%	100%	100%	100%	98.6%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GCM2	100%	100%	100%	100%	99.7%	Hypoparathyroidism, familial isolated 2, 618883;Hyperparathyroidism 4, 617343
GFRA1	94%	94%	100%	100%	99.5%	Renal hypodysplasia/aplasia 4, 619887
GLA	91.4%	91.4%	99.3%	91.7%	72.9%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
GLI3	99.3%	99.3%	100%	99.9%	99.2%	Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700
GLIS2	100%	100%	100%	99.8%	99.1%	Nephronophthisis 7, 611498
GLIS3	100%	100%	100%	100%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GNA11	100%	100%	100%	100%	99.5%	Hypocalciuric hypercalcemia, type II, 145981;Hypocalcemia, autosomal dominant 2, 615361

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GREB1L	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805
GRHPR	86.7%	86.6%	100%	100%	99.7%	Hyperoxaluria, primary, type II, 260000
GRIP1	100%	100%	100%	100%	99.9%	Fraser syndrome 3, 617667
GSN	92.1%	90%	100%	99.8%	98.9%	Amyloidosis, Finnish type, 105120
HNF1A	100%	100%	100%	100%	99.5%	Hepatic adenoma, somatic, 142330;Diabetes mellitus, insulin-dependent, 20, 612520;{Diabetes mellitus, noninsulin-dependent, 2}, 125853;MODY, type III, 600496;{Diabetes mellitus, insulin-dependent}, 222100;Renal cell carcinoma, 144700
HNF1B	100%	100%	100%	100%	99.6%	Type 2 diabetes mellitus, 125853;Renal cysts and diabetes syndrome, 137920;{Renal cell carcinoma}, 144700
HNF4A	100%	100%	100%	100%	99.3%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026;{Diabetes mellitus, noninsulin-dependent}, 125853;MODY, type I, 125850
HOGA1	100%	100%	100%	100%	99.5%	Hyperoxaluria, primary, type III, 613616
HPRT1	87.6%	87.4%	99%	91.6%	74%	Hyperuricemia, HPRT-related, 300323;Lesch-Nyhan syndrome, 300322

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HPSE2	100%	100%	100%	100%	99.5%	Urofacial syndrome 1, 236730
HSD11B2	100%	100%	100%	100%	99.4%	Apparent mineralocorticoid excess, 218030
IFNG	100%	100%	100%	100%	100%	{Hepatitis C virus, response to therapy of}, 609532;{TSC2 angiomyolipomas, renal, modifier of}, 613254;{Aplastic anemia}, 609135;?Immunodeficiency 69, mycobacteriosis, 618963;{Tuberculosis, protection against}, 607948;{AIDS, rapid progression to}, 609423
IFT122	100%	100%	100%	100%	99.6%	Cranioectodermal dysplasia 1, 218330
IFT140	100%	100%	100%	100%	99.6%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180
IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	99.8%	96.5%	100%	100%	99.9%	Bardet-Biedl syndrome 19, 615996

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IFT43	93.5%	93.5%	100%	100%	99.7%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT56	100%	100%	100%	100%	99.8%	Biliary, renal, neurologic, and skeletal syndrome, 619534
INF2	100%	100%	99.9%	99.5%	98.1%	Glomerulosclerosis, focal segmental, 5, 613237;Charcot-Marie-Tooth disease, dominant intermediate E, 614455
INPP5E	100%	100%	100%	99.8%	98.1%	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300
INTU	100%	100%	100%	100%	99.7%	?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100%	100%	100%	100%	99.9%	Nephronophthisis 2, infantile, 602088
IQCB1	95.8%	95.8%	100%	100%	99.7%	Senior-Loken syndrome 5, 609254
ITGA3	100%	100%	100%	100%	99.5%	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748
ITGA8	100%	100%	100%	100%	99.7%	Renal hypodysplasia/aplasia 1, 191830
ITSN1	100%	100%	100%	100%	99.7%	
ITSN2	100%	100%	100%	100%	99.7%	

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JAG1	100%	100%	100%	100%	99.6%	?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	98.1%	98.1%	100%	100%	99.7%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	100%	100%	100%	100%	99.6%	Nephrotic syndrome, type 16, 617783;Palmoplantar keratoderma and woolly hair, 616099
KATNIP	100%	100%	100%	100%	99.4%	Joubert syndrome 26, 616784
KCNJ1	100%	100%	100%	100%	99.9%	Bartter syndrome, type 2, 241200
KCNJ10	100%	100%	100%	100%	99.7%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780
KCNJ16	100%	100%	100%	100%	99.9%	Hypokalemic tubulopathy and deafness, 619406
KCNJ5	100%	100%	100%	100%	99.2%	Long QT syndrome 13, 613485;Hyperaldosteronism, familial, type III, 613677
KIF14	97.6%	97.6%	100%	100%	99.8%	Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258
KIF7	100%	100%	100%	100%	99.2%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131

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KIRREL1	100%	100%	100%	100%	99.2%	Nephrotic syndrome, type 23, 619201
KL	100%	99.8%	99.8%	99.3%	98.4%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLHL3	100%	100%	100%	100%	99.6%	Pseudohypoaldosteronism, type IID, 614495
LAGE3	100%	100%	98.7%	89%	67.1%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA5	100%	100%	100%	99.9%	99.2%	Nephrotic syndrome, type 26, 620049;?Bent bone dysplasia syndrome 2, 620076
LAMB2	100%	100%	100%	100%	99.7%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049
LCAT	100%	100%	100%	99.9%	99%	Fish-eye disease, 136120;Norum disease, 245900
LMX1B	100%	100%	100%	100%	99%	Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LRIG2	98%	98%	100%	100%	99.8%	Urofacial syndrome 2, 615112
LRP2	100%	100%	100%	100%	99.8%	Donnai-Barrow syndrome, 222448
LRP4	100%	100%	100%	99.9%	99.4%	?Myasthenic syndrome, congenital, 17, 616304;Sclerosteosis 2, 614305;Cenani-Lenz syndactyly syndrome, 212780

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LRP5	100%	100%	100%	100%	99%	Osteopetrosis, autosomal dominant 1, 607634;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;[Bone mineral density variability 1, high bone mass], 601884;Exudative vitreoretinopathy 4, 601813
LYZ	100%	100%	100%	100%	99.8%	Amyloidosis, hereditary systemic 5, 620658
LZTFL1	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 17, 615994
MAFB	100%	100%	100%	99.9%	98.8%	Duane retraction syndrome 3, 617041;Multicentric carpotarsal osteolysis syndrome, 166300
MAGED2	99.4%	97.2%	98.6%	89.3%	70.4%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGI2	99.6%	97.9%	100%	99.8%	99%	Nephrotic syndrome, type 15, 617609
MAPKBP1	100%	100%	100%	100%	99.8%	Nephronophthisis 20, 617271
MKKS	100%	100%	100%	100%	100%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKS1	99.1%	99%	100%	100%	99.7%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MMACHC	100%	100%	100%	100%	99.5%	Methylmalonic aciduria and homocystinuria, cblC type, 277400

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MOCOS	97.6%	97.6%	100%	100%	99.6%	Xanthinuria, type II, 603592
MYH9	97.3%	97%	100%	100%	99.4%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYO1E	100%	100%	100%	100%	99.6%	Glomerulosclerosis, focal segmental, 6, 614131
NCAPG2	100%	100%	100%	100%	99.8%	Khan-Khan-Katsanis syndrome, 618460
NEK1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK8	100%	100%	100%	100%	99.8%	Renal-hepatic-pancreatic dysplasia 2, 615415;Polycystic kidney disease 8, 620903;?Nephronophthisis 9, 613824
NEU1	100%	100%	100%	99.9%	99.5%	Sialidosis, type II, 256550;Sialidosis, type I, 256550
NHERF1	85.8%	85.8%	100%	100%	99.2%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
NOS1AP	100%	100%	100%	100%	99.6%	Nephrotic syndrome, type 22, 619155
NOTCH2	100%	100%	100%	100%	99.8%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500

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NPHP1	100%	100%	100%	100%	99.8%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900
NPHP3	100%	100%	100%	100%	99.6%	Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010
NPHP4	100%	100%	100%	100%	99.5%	Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966
NPHS1	100%	100%	100%	100%	99.3%	Nephrotic syndrome, type 1, 256300
NPHS2	100%	100%	100%	100%	99.3%	Nephrotic syndrome, type 2, 600995
NR3C2	100%	100%	100%	99.8%	99.1%	Pseudohypoaldosteronism type I, autosomal dominant, 177735;Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NUP107	97.8%	97.8%	100%	100%	99.9%	?Ovarian dysgenesis 6, 618078;Galloway-Mowat syndrome 7, 618348;Nephrotic syndrome, type 11, 616730
NUP133	97.9%	97.9%	100%	100%	99.8%	?Galloway-Mowat syndrome 8, 618349;Nephrotic syndrome, type 18, 618177
NUP160	94.6%	94.6%	100%	100%	99.8%	?Nephrotic syndrome, type 19, 618178
NUP205	97%	97%	100%	100%	99.6%	?Nephrotic syndrome, type 13, 616893
NUP85	100%	99.8%	100%	100%	99.4%	Nephrotic syndrome, type 17, 618176

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NUP93	95.5%	95.5%	100%	100%	99.7%	Nephrotic syndrome, type 12, 616892
NXF5						
OCRL	100%	100%	99.3%	92.4%	74.8%	Dent disease 2, 300555;Lowe syndrome, 309000
OFD1	100%	100%	99.1%	91%	73.2%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
OSGEP	100%	100%	100%	100%	99.8%	Galloway-Mowat syndrome 3, 617729
OXGR1	100%	100%	100%	100%	99.8%	Nephrolithiasis, calcium oxalate, 2, with nephrocalcinosis, 620374
PAX2	100%	100%	100%	100%	99.2%	Glomerulosclerosis, focal segmental, 7, 616002;Papillorenal syndrome, 120330
PBX1	100%	100%	100%	100%	99.7%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCBD1	100%	100%	100%	100%	99.5%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PDE6D	100%	100%	100%	100%	99.1%	Joubert syndrome 22, 615665
PDSS2	100%	100%	100%	100%	99.4%	Coenzyme Q10 deficiency, primary, 3, 614652
PHEX	100%	100%	99%	92.2%	75%	Hypophosphatemic rickets, X-linked dominant, 307800

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PKD1	100%	99.9%	100%	99.9%	99.1%	Polycystic kidney disease 1, 173900
PKD2	100%	100%	100%	100%	99.1%	Polycystic kidney disease 2, 613095
PKHD1	100%	100%	100%	100%	99.7%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PLCE1	100%	100%	100%	100%	99.8%	Nephrotic syndrome, type 3, 610725
PMM2	94.6%	94.6%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PODXL	91.7%	91.4%	100%	100%	99.3%	
PTH1R	100%	100%	100%	100%	99%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045
PTPRO	100%	100%	100%	100%	99.8%	Nephrotic syndrome, type 6, 614196
RAD21	100%	100%	100%	99.9%	99.6%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
REN	100%	100%	100%	100%	99.4%	Renal tubular dysgenesis, 267430;[Hyperprorenin emia];Tubulointerstitial kidney disease, autosomal dominant, 4, 613092
RMND1	85.8%	85.8%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 11, 614922

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RNU4ATAC						Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
ROBO2	100%	100%	100%	100%	99.8%	Vesicoureteral reflux 2, 610878
RPGRIP1L	100%	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RRAGD	100%	100%	100%	100%	99.8%	Hypomagnesemia 7, renal, with or without dilated cardiomyopathy, 620152
RRM2B	100%	100%	100%	100%	99.9%	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%	100%	100%	99.2%	Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	100%	100%	100%	100%	99.6%	?IVIC syndrome, 147750;Duane-radial ray syndrome, 607323

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SARS2	100%	100%	100%	100%	99.7%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCARB2	100%	100%	100%	100%	99.7%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCNN1A	100%	100%	100%	99.9%	99.5%	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350;?Liddle syndrome 3, 618126;Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100%	100%	100%	100%	99.6%	Bronchiectasis with or without elevated sweat chloride 1, 211400;Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125;Liddle syndrome 1, 177200
SCNN1G	100%	100%	100%	100%	99.2%	Bronchiectasis with or without elevated sweat chloride 3, 613071;Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126;Liddle syndrome 2, 618114
SDCCAG8	100%	100%	100%	100%	99.9%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993
SEC61A1	92.3%	92.3%	100%	99.9%	99.4%	Immunodeficiency, common variable, 15, 620670;?Neutropenia, severe congenital, 11, autosomal dominant, 620674;Tubulointerstitial kidney disease, autosomal dominant, 5, 617056

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SGPL1	95.5%	95.5%	100%	100%	99.7%	RENI syndrome, 617575
SIX5	100%	100%	100%	100%	99.4%	Branchiootorenal syndrome 2, 610896
SLC12A1	96.3%	96.3%	100%	100%	99.8%	Bartter syndrome, type 1, 601678
SLC12A3	94.7%	94.7%	100%	100%	99.7%	Gitelman syndrome, 263800
SLC16A12	100%	100%	100%	99.9%	99.8%	Cataract 47, juvenile, with microcornea, 612018
SLC22A12	100%	100%	100%	99.9%	98.7%	Hypouricemia, renal, 220150
SLC26A1	100%	100%	100%	100%	99.5%	?Hypersulfaturia, 620372;?Nephrolithiasis, calcium oxalate, 1, 167030
SLC26A3	100%	100%	100%	100%	99.9%	Diarrhea 1, secretory chloride, congenital, 214700
SLC2A2	100%	100%	100%	100%	99.8%	Fanconi-Bickel syndrome, 227810;{Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	100%	100%	100%	100%	99.7%	{Uric acid concentration, serum, QTL 2}, 612076;Hypouricemia, renal, 2, 612076
SLC34A1	100%	100%	100%	99.9%	99.4%	?Fanconi renotubular syndrome 2, 613388;Hypercalcemia, infantile, 2, 616963;Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A3	100%	100%	100%	99.8%	98.6%	Hypophosphatemic rickets with hypercalciuria, 241530

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SLC36A2	100%	100%	100%	100%	99.8%	[Iminoglycinuria], 242600;[Hyperglycinuria], 138500
SLC37A4	100%	100%	100%	100%	99.8%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240
SLC3A1	96.2%	96.2%	100%	100%	99.8%	Cystinuria, 220100
SLC41A1	100%	100%	100%	100%	99.8%	?Nephronophthisis-like nephropathy 2, 619468
SLC4A1	100%	100%	100%	100%	99.3%	[Blood group, Swann], 601550;[Blood group, Wright], 112050;Distal renal tubular acidosis 1, 179800;[Blood group, Waldner], 112010;Spherocytosis, type 4, 612653;[Blood group, Froese], 601551;Distal renal tubular acidosis 4 with hemolytic anemia, 611590;{Malaria, resistance to}, 611162;Cryohydrocytosis, 185020;Ovalocytosis, SA type, 166900;[Blood group, Diego], 110500
SLC4A4	95%	95%	100%	100%	99.9%	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278
SLC5A2	100%	100%	100%	99.9%	99.4%	Renal glucosuria, 233100
SLC6A19	100%	100%	100%	99.9%	99%	Hartnup disorder, 234500
SLC6A20	99.9%	98.6%	100%	99.9%	99.5%	
SLC7A7	100%	100%	100%	100%	98.8%	Lysinuric protein intolerance, 222700
SLC7A9	100%	100%	100%	100%	99.5%	Cystinuria, 220100

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SLC9A3	100%	100%	100%	99.7%	98.5%	Diarrhea 8, secretory sodium, congenital, 616868
SLIT3	100%	100%	100%	100%	99.5%	
SMARCAL1	95.6%	95.6%	100%	100%	99.7%	Schimke immunoosseous dysplasia, 242900
SOX17	100%	100%	100%	100%	98.6%	Vesicoureteral reflux 3, 613674;Pulmonary hypertension, primary, 7, 621248
STRA6	100%	100%	100%	100%	99.3%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186
STX16	100%	100%	100%	100%	99.6%	Pseudohypoparathyroidism Ib, 603233
TBC1D8B	100%	100%	99.3%	92.3%	75.5%	Nephrotic syndrome, type 20, 301028
TBX18	100%	100%	100%	100%	99.2%	Congenital anomalies of kidney and urinary tract 2, 143400
TCTN1	92.6%	90.8%	100%	100%	99.5%	Joubert syndrome 13, 614173
TCTN2	98.5%	98.5%	100%	100%	99.5%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	99.5%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
THBD	100%	100%	100%	99.8%	98.7%	Thrombophilia 12 due to thrombomodulin defect, 614486;{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926

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TMEM107	100%	100%	100%	100%	99.2%	Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562
TMEM138	100%	98.8%	100%	99.9%	99.7%	Joubert syndrome 16, 614465
TMEM216	100%	100%	100%	100%	99.8%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194
TMEM231	93.5%	93.5%	100%	100%	99.5%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397
TMEM237	98.2%	98.2%	100%	100%	99.7%	Joubert syndrome 14, 614424
TMEM260	94.8%	94.8%	100%	100%	99.7%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	96.1%	96.1%	100%	100%	99.7%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TNS2	100%	100%	100%	99.9%	98.5%	
TNXB	100%	100%	100%	99.9%	98.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963
TP53RK	100%	100%	100%	100%	99.8%	Galloway-Mowat syndrome 4, 617730
TPRKB	83.2%	81.2%	100%	100%	99.8%	Galloway-Mowat syndrome 5, 617731
TRAF3IP1	100%	100%	100%	100%	99.6%	

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TRIM32	100%	100%	100%	100%	99.6%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRPC6	100%	100%	100%	100%	99.7%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM6	100%	100%	100%	100%	99.8%	Hypomagnesemia 1, intestinal, 602014
TSC1	100%	100%	100%	100%	99.6%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
TSC2	100%	100%	100%	100%	99.3%	Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254
TTC21B	98.4%	98.1%	100%	100%	99.8%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820
TTC8	91.4%	91.4%	100%	100%	99.9%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464
TULP3	93.4%	93.4%	100%	100%	99.5%	Hepatorenocardiac degenerative fibrosis, 619902
UMOD	90.8%	89.6%	100%	100%	99.8%	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000
UPK3A	100%	100%	100%	100%	99.7%	
UQCC2	100%	100%	100%	100%	99.9%	Mitochondrial complex III deficiency, nuclear type 7, 615824

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VDR	100%	100%	100%	100%	99.1%	Rickets, vitamin D-resistant, type IIA, 277440
VIPAS39	99.8%	97.9%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100%	100%	100%	100%	99.9%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR19	100%	100%	100%	100%	99.8%	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%	100%	100%	99.7%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WDR73	100%	100%	100%	100%	99.3%	Galloway-Mowat syndrome 1, 251300
WNK1	100%	100%	100%	99.9%	99%	Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492
WNK4	100%	100%	100%	99.9%	98.7%	Pseudohypoaldosteronism, type IIB, 614491
WNT4	100%	100%	100%	100%	99.1%	Mullerian aplasia and hyperandrogenism, 158330;SERKAL syndrome, 611812

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WT1	100%	100%	100%	99.9%	99.3%	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%	100%	100%	99.6%	Xanthinuria, type I, 278300
XPNPEP3	100%	100%	100%	99.9%	99.6%	Nephronophthisis-like nephropathy 1, 613159
XPO5	100%	100%	100%	100%	99.6%	
ZMPSTE24	100%	100%	100%	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210
ZNF423	100%	100%	100%	100%	99.4%	Nephronophthisis 14, 614844;Joubert syndrome 19, 614844
ZNG1A	99.2%	97.9%	96.4%	95.8%	94%	

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 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors