

RENAL DISORDERS PANEL DG-4.0.0 (326 GENES)

| <i>Gene</i> | <i>Twist X2 covered >10x</i> | <i>Twist X2 covered >20x</i> | <i>WGS covered >10x</i> | <i>WGS covered >20x</i> | <i>Associated Phenotype description and OMIM disease ID</i> |
|-------------|---------------------------------|---------------------------------|----------------------------|----------------------------|--|
| ACE | 100.0% | 100.0% | 99.9% | 96.5% | {Stroke, hemorrhagic}, 614519;Renal tubular dysgenesis, 267430;{Microvascular complications of diabetes 3}, 612624;{Myocardial infarction, susceptibility to}, ;[Angiotensin I-converting enzyme, benign serum increase], ;{SARS, progression of}, |
| ACTN4 | 98.9% | 98.9% | 100.0% | 99.0% | Glomerulosclerosis, focal segmental, 1, 603278 |
| ADAMTS13 | 100.0% | 100.0% | 100.0% | 98.4% | Thrombotic thrombocytopenic purpura, hereditary, 274150 |
| ADAMTS9 | 99.9% | 99.6% | 100.0% | 98.1% | |
| ADCY10 | 100.0% | 100.0% | 100.0% | 98.9% | {Hypercalciuria, absorptive, susceptibility to}, 143870 |
| AGT | 95.5% | 95.3% | 100.0% | 99.3% | Renal tubular dysgenesis, 267430;{Hypertension, essential, susceptibility to}, 145500;{Preeclampsia, susceptibility to}, |

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| AGTR1 | 100.0% | 100.0% | 100.0% | 99.1% | {Hypertension, essential}, 145500;Renal tubular dysgenesis, 267430 |
| AGXT | 100.0% | 100.0% | 100.0% | 99.6% | Hyperoxaluria, primary, type 1, 259900 |
| AHI1 | 98.7% | 98.7% | 100.0% | 98.2% | Joubert syndrome 3, 608629 |
| ALDOB | 100.0% | 100.0% | 100.0% | 99.4% | Fructose intolerance, hereditary, 229600 |
| ALG1 | 100.0% | 100.0% | 100.0% | 99.4% | Congenital disorder of glycosylation, type I _k , 608540 |
| ALG8 | 78.1% | 77.5% | 100.0% | 97.9% | Congenital disorder of glycosylation, type I _h , 608104;Polycystic liver disease 3 with or without kidney cysts, 617874 |
| ALG9 | 100.0% | 100.0% | 100.0% | 98.6% | Gillessen-Kaesbach- Nishimura syndrome, 263210;Congenital disorder of glycosylation, type II, 608776 |
| ALMS1 | 100.0% | 100.0% | 100.0% | 98.4% | Alstrom syndrome, 203800 |
| AMN | 100.0% | 100.0% | 100.0% | 97.9% | Imlerslund-Grasbeck syndrome 2, 618882 |
| ANKFY1 | 100.0% | 100.0% | 100.0% | 98.9% | |
| ANKS6 | 99.9% | 99.4% | 100.0% | 97.6% | Nephronophthisis 16, 615382 |

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| ANLN | 100.0% | 100.0% | 100.0% | 98.4% | Focal segmental glomerulosclerosis 8, 616032 |
| ANOS1 | 100.0% | 99.8% | 97.6% | 68.8% | Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 |
| AP2S1 | 100.0% | 100.0% | 100.0% | 94.3% | Hypocalciuric hypercalcemia, type III, 600740 |
| APOL1 | 100.0% | 100.0% | 100.0% | 98.7% | {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 |
| APRT | 100.0% | 100.0% | 100.0% | 99.0% | Adenine phosphoribosyltransferase deficiency, 614723 |
| AQP2 | 100.0% | 100.0% | 100.0% | 99.7% | Diabetes insipidus, nephrogenic, 2, 125800 |
| ARHGAP24 | 100.0% | 100.0% | 99.6% | 95.3% | |
| ARHGDI1 | 100.0% | 100.0% | 100.0% | 99.2% | Nephrotic syndrome, type 8, 615244 |
| ARHGEF6 | 100.0% | 100.0% | 97.4% | 69.1% | |
| ARL13B | 93.4% | 93.3% | 100.0% | 97.3% | Joubert syndrome 8, 612291 |
| ARL6 | 100.0% | 100.0% | 100.0% | 95.9% | Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151 |

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| ATP1A1 | 100.0% | 100.0% | 100.0% | 99.1% | Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 |
| ATP6V0A4 | 100.0% | 100.0% | 100.0% | 98.1% | Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722 |
| ATP6V1B1 | 100.0% | 100.0% | 100.0% | 99.0% | Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300 |
| ATP7B | 100.0% | 100.0% | 100.0% | 99.3% | Wilson disease, 277900 |
| AVIL | 100.0% | 100.0% | 100.0% | 99.1% | Nephrotic syndrome, type 21, 618594 |
| AVP | 100.0% | 100.0% | 100.0% | 97.5% | Diabetes insipidus, neurohypophyseal, 125700 |
| AVPR2 | 100.0% | 100.0% | 98.9% | 78.7% | Diabetes insipidus, nephrogenic, 1, 304800;Nephrogenic syndrome of inappropriate antidiuresis, 300539 |
| B9D1 | 100.0% | 100.0% | 100.0% | 99.5% | ?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120 |
| B9D2 | 100.0% | 100.0% | 100.0% | 99.9% | ?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175 |

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| BBIP1 | 100.0% | 100.0% | 100.0% | 98.9% | Bardet-Biedl syndrome 18, 615995 |
| BBS1 | 100.0% | 100.0% | 100.0% | 99.6% | Bardet-Biedl syndrome 1, 209900 |
| BBS10 | 100.0% | 100.0% | 100.0% | 98.7% | Bardet-Biedl syndrome 10, 615987 |
| BBS12 | 100.0% | 100.0% | 100.0% | 99.6% | Bardet-Biedl syndrome 12, 615989 |
| BBS2 | 98.0% | 98.0% | 100.0% | 98.8% | Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981 |
| BBS4 | 100.0% | 100.0% | 100.0% | 98.0% | Bardet-Biedl syndrome 4, 615982 |
| BBS5 | 100.0% | 100.0% | 100.0% | 98.9% | Bardet-Biedl syndrome 5, 615983 |
| BBS7 | 100.0% | 100.0% | 100.0% | 99.1% | Bardet-Biedl syndrome 7, 615984 |
| BBS9 | 95.8% | 95.8% | 100.0% | 97.9% | Bardet-Biedl syndrome 9, 615986 |
| BCS1L | 100.0% | 100.0% | 100.0% | 99.2% | GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000 |
| BICC1 | 100.0% | 99.4% | 100.0% | 99.0% | {Renal dysplasia, cystic, susceptibility to}, 601331 |

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| BSND | 100.0% | 100.0% | 100.0% | 99.1% | Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522 |
| C3 | 97.6% | 97.5% | 100.0% | 99.0% | C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378 |
| CA2 | 100.0% | 100.0% | 100.0% | 98.8% | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 |
| CACNA1H | 99.5% | 98.7% | 99.9% | 96.9% | {Epilepsy, childhood absence, susceptibility to, 6}, 611942;Hyperaldosteronism, familial, type IV, 617027;{Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 |
| CASR | 100.0% | 100.0% | 100.0% | 98.6% | 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 |

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| CC2D2A | 98.2% | 98.2% | 100.0% | 98.4% | COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285 |
| CCNQ | 100.0% | 99.9% | 96.5% | 73.7% | STAR syndrome, 300707 |
| CD151 | 100.0% | 100.0% | 100.0% | 99.8% | [Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057 |
| CD2AP | 100.0% | 100.0% | 100.0% | 96.7% | Glomerulosclerosis, focal segmental, 3, 607832 |
| CD46 | 100.0% | 100.0% | 100.0% | 98.1% | {Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 |
| CDC73 | 100.0% | 100.0% | 100.0% | 98.9% | Hyperparathyroidism, familial primary, 145000;Parathyroid adenoma with cystic changes, 145001;Parathyroid carcinoma, 608266;Hyperparathyroidism-jaw tumor syndrome, 145001 |
| CEP120 | 100.0% | 100.0% | 100.0% | 98.9% | Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761 |

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| CEP164 | 100.0% | 100.0% | 100.0% | 98.2% | Nephronophthisis 15, 614845 |
| CEP290 | 100.0% | 100.0% | 100.0% | 96.4% | Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134 |
| CEP41 | 100.0% | 100.0% | 100.0% | 98.0% | Joubert syndrome 15, 614464 |
| CEP55 | 100.0% | 100.0% | 100.0% | 98.3% | Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 |
| CEP83 | 100.0% | 100.0% | 100.0% | 95.9% | Nephronophthisis 18, 615862 |
| CFB | 100.0% | 100.0% | 100.0% | 99.1% | ?Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489 |

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| CFH | 97.5% | 97.4% | 100.0% | 99.3% | {Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 |
| CFHR1 | 99.2% | 97.7% | 95.4% | 81.7% | {Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 |
| CFHR3 | 99.8% | 99.4% | 96.9% | 84.2% | {Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 |
| CFHR5 | 100.0% | 100.0% | 100.0% | 98.4% | Nephropathy due to CFHR5 deficiency, 614809 |
| CFI | 100.0% | 100.0% | 100.0% | 98.3% | {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984 |
| CHRM3 | 100.0% | 100.0% | 100.0% | 99.0% | Prune belly syndrome, 100100 |

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| CHRNA3 | 100.0% | 100.0% | 100.0% | 97.1% | {Lung cancer susceptibility 2}, 612052;Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800 |
| CLCN2 | 100.0% | 100.0% | 100.0% | 98.7% | 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 | 100.0% | 99.9% | 97.7% | 71.8% | Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990;Hypophosphatemic rickets, 300554;Dent disease 1, 300009;Nephrolithiasis, type I, 310468 |
| CLCNKB | 100.0% | 100.0% | 100.0% | 98.6% | Bartter syndrome, type 3, 607364;Bartter syndrome, type 4b, digenic, 613090 |
| CLDN10 | 100.0% | 100.0% | 100.0% | 99.2% | HELIX syndrome, 617671 |
| CLDN16 | 100.0% | 100.0% | 100.0% | 98.8% | Hypomagnesemia 3, renal, 248250 |

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| CLDN19 | 100.0% | 100.0% | 100.0% | 99.9% | Hypomagnesemia 5, renal, with ocular involvement, 248190 |
| CNNM2 | 100.0% | 100.0% | 100.0% | 97.4% | Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418 |
| COL4A1 | 100.0% | 100.0% | 100.0% | 98.4% | ?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 | 100.0% | 100.0% | 100.0% | 98.1% | Alport syndrome 3A, autosomal dominant, 104200;Hematuria, benign familial, 2, 620320;Alport syndrome 3B, autosomal recessive, 620536 |
| COL4A4 | 99.5% | 98.6% | 100.0% | 98.5% | Hematuria, familial benign, 1, 141200;Alport syndrome 2, autosomal recessive, 203780 |

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| COL4A5 | 99.3% | 98.7% | 97.6% | 68.1% | Alport syndrome 1, X-linked, 301050 |
| COQ2 | 96.3% | 96.3% | 100.0% | 98.5% | {Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426 |
| COQ6 | 100.0% | 100.0% | 99.9% | 98.4% | Coenzyme Q10 deficiency, primary, 6, 614650 |
| COQ7 | 100.0% | 100.0% | 100.0% | 98.8% | Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402 |
| COQ8B | 100.0% | 100.0% | 100.0% | 99.0% | Nephrotic syndrome, type 9, 615573 |
| COQ9 | 100.0% | 100.0% | 100.0% | 98.8% | Coenzyme Q10 deficiency, primary, 5, 614654 |
| CPLANE1 | 100.0% | 100.0% | 100.0% | 98.0% | Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615 |
| CRB2 | 100.0% | 100.0% | 100.0% | 98.9% | Focal segmental glomerulosclerosis 9, 616220;Ventriculomegaly with cystic kidney disease, 219730 |
| CSPP1 | 96.9% | 96.9% | 100.0% | 97.9% | Joubert syndrome 21, 615636 |

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| CTNS | 100.0% | 100.0% | 99.8% | 97.9% | Cystinosis, nephropathic, 219800;Cystinosis, ocular nonnephropathic, 219750;Cystinosis, late-onset juvenile or adolescent nephropathic, 219900;Cystinosis, atypical nephropathic, 219800 |
| CUBN | 100.0% | 100.0% | 100.0% | 99.2% | [Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100 |
| CUL3 | 100.0% | 100.0% | 100.0% | 97.3% | Neurodevelopmental disorder with or without autism or seizures, 619239;Pseudohypoaldosteronism, type IIE, 614496 |
| CYP24A1 | 100.0% | 100.0% | 100.0% | 98.7% | Hypercalcemia, infantile, 1, 143880 |
| DAAM2 | 100.0% | 100.0% | 100.0% | 99.3% | Nephrotic syndrome, type 24, 619263 |
| DCDC2 | 100.0% | 100.0% | 100.0% | 97.6% | Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394 |
| DGKE | 100.0% | 100.0% | 100.0% | 98.7% | {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008 |

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| DLC1 | 100.0% | 100.0% | 100.0% | 98.6% | Colorectal cancer, somatic, 114500 |
| DMP1 | 100.0% | 100.0% | 100.0% | 99.1% | Hypophosphatemic rickets, AR, 241520 |
| DNAJB11 | 100.0% | 100.0% | 100.0% | 97.3% | Polycystic kidney disease 6 with or without polycystic liver disease, 618061 |
| DSTYK | 100.0% | 100.0% | 100.0% | 98.9% | Spastic paraplegia 23, autosomal recessive, 270750;Congenital anomalies of kidney and urinary tract 1, 610805 |
| DYNC2H1 | 99.8% | 99.4% | 100.0% | 97.9% | Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 |
| DYNC211 | 100.0% | 100.0% | 100.0% | 98.0% | Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 |
| DZIP1L | 100.0% | 100.0% | 100.0% | 99.0% | Polycystic kidney disease 5, 617610 |
| EGF | 100.0% | 100.0% | 100.0% | 98.8% | ?Hypomagnesemia 4, renal, 611718 |
| EHHADH | 100.0% | 100.0% | 100.0% | 99.2% | ?Fanconi renotubular syndrome 3, 615605 |
| EMP2 | 100.0% | 100.0% | 100.0% | 97.8% | Nephrotic syndrome, type 10, 615861 |

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|---------|--------|--------|--------|-------|---|
| ENPP1 | 100.0% | 99.7% | 100.0% | 97.7% | {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 | 100.0% | 100.0% | 100.0% | 99.1% | Branchiotoic syndrome 1, 602588;Branchiotoic renal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780 |
| FAH | 100.0% | 100.0% | 100.0% | 98.6% | Tyrosinemia, type I, 276700 |
| FAM111A | 100.0% | 100.0% | 100.0% | 98.8% | Kenny-Caffey syndrome, type 2, 127000;Gracile bone dysplasia, 602361 |
| FAM20A | 100.0% | 100.0% | 100.0% | 97.8% | Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 |
| FAN1 | 100.0% | 100.0% | 100.0% | 97.9% | Interstitial nephritis, karyomegalic, 614817 |
| FAT1 | 100.0% | 100.0% | 100.0% | 99.2% | |

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| FGF23 | 100.0% | 100.0% | 100.0% | 99.3% | Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100 |
| FN1 | 100.0% | 100.0% | 100.0% | 99.3% | Spondylometaphyseal dysplasia, corner fracture type, 184255;Glomerulopathy with fibronectin deposits 2, 601894 |
| FOXC2 | 100.0% | 100.0% | 99.9% | 92.9% | Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 |
| FOXI1 | 100.0% | 100.0% | 100.0% | 99.3% | Enlarged vestibular aqueduct, 600791 |
| FRAS1 | 100.0% | 99.9% | 100.0% | 99.1% | Fraser syndrome 1, 219000 |
| FREM1 | 100.0% | 100.0% | 100.0% | 98.9% | Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485 |
| FREM2 | 99.9% | 99.7% | 100.0% | 98.9% | Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570 |

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| FXD2 | 100.0% | 100.0% | 100.0% | 99.5% | Hypomagnesemia 2, renal, 154020 |
| G6PC1 | 100.0% | 100.0% | 100.0% | 99.4% | Glycogen storage disease Ia, 232200 |
| GALNT3 | 100.0% | 100.0% | 100.0% | 97.9% | Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 |
| GANAB | 100.0% | 100.0% | 100.0% | 99.3% | Polycystic kidney disease 3, 600666 |
| GAPVD1 | 100.0% | 100.0% | 100.0% | 98.5% | |
| GATA3 | 100.0% | 100.0% | 100.0% | 99.1% | Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 |
| GCM2 | 100.0% | 100.0% | 100.0% | 99.0% | Hypoparathyroidism, familial isolated 2, 618883;Hyperparathyroidism 4, 617343 |
| GFRA1 | 94.0% | 94.0% | 100.0% | 98.8% | Renal hypodysplasia/aplasia 4, 619887 |
| GLA | 91.4% | 91.4% | 98.4% | 73.6% | Fabry disease, cardiac variant, 301500;Fabry disease, 301500 |

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| GLI3 | 99.3% | 99.3% | 100.0% | 99.5% | Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700 |
| GLIS2 | 100.0% | 100.0% | 100.0% | 99.6% | Nephronophthisis 7, 611498 |
| GLIS3 | 100.0% | 100.0% | 100.0% | 99.1% | Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 |
| GNA11 | 100.0% | 100.0% | 100.0% | 97.4% | Hypocalciuric hypercalcemia, type II, 145981;Hypocalcemia, autosomal dominant 2, 615361 |
| GREB1L | 100.0% | 100.0% | 100.0% | 98.8% | Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805 |
| GRHPR | 100.0% | 100.0% | 100.0% | 98.9% | Hyperoxaluria, primary, type II, 260000 |
| GRIP1 | 100.0% | 99.8% | 100.0% | 99.2% | Fraser syndrome 3, 617667 |
| GSN | 100.0% | 100.0% | 100.0% | 98.0% | Amyloidosis, Finnish type, 105120 |

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| HNF1A | 100.0% | 100.0% | 100.0% | 99.6% | 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 | 100.0% | 100.0% | 100.0% | 98.7% | Type 2 diabetes mellitus, 125853;Renal cysts and diabetes syndrome, 137920;{Renal cell carcinoma}, 144700 |
| HNF4A | 100.0% | 100.0% | 100.0% | 99.3% | Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026;{Diabetes mellitus, noninsulin-dependent}, 125853;MODY, type I, 125850 |
| HOGA1 | 100.0% | 100.0% | 100.0% | 99.2% | Hyperoxaluria, primary, type III, 613616 |
| HPRT1 | 100.0% | 100.0% | 98.4% | 70.9% | Hyperuricemia, HRPT-related, 300323;Lesch-Nyhan syndrome, 300322 |
| HPSE2 | 100.0% | 100.0% | 100.0% | 98.3% | Urofacial syndrome 1, 236730 |
| HSD11B2 | 100.0% | 100.0% | 99.9% | 94.8% | Apparent mineralocorticoid excess, 218030 |

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| IFNG | 100.0% | 100.0% | 100.0% | 97.5% | {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 | 100.0% | 100.0% | 100.0% | 99.1% | Cranioectodermal dysplasia 1, 218330 |
| IFT140 | 100.0% | 100.0% | 100.0% | 99.1% | Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781 |
| IFT172 | 100.0% | 100.0% | 100.0% | 99.0% | Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 |
| IFT27 | 100.0% | 100.0% | 100.0% | 99.2% | Bardet-Biedl syndrome 19, 615996 |

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| IFT43 | 100.0% | 100.0% | 100.0% | 98.7% | ?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866 |
| IFT56 | 100.0% | 100.0% | 100.0% | 98.5% | Biliary, renal, neurologic, and skeletal syndrome, 619534 |
| INF2 | 100.0% | 99.9% | 99.8% | 95.8% | Glomerulosclerosis, focal segmental, 5, 613237;Charcot-Marie-Tooth disease, dominant intermediate E, 614455 |
| INPP5E | 100.0% | 100.0% | 100.0% | 97.0% | Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300 |
| INTU | 100.0% | 100.0% | 100.0% | 97.2% | ?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925 |
| INVS | 100.0% | 100.0% | 100.0% | 99.2% | Nephronophthisis 2, infantile, 602088 |
| IQCB1 | 100.0% | 100.0% | 100.0% | 97.9% | Senior-Loken syndrome 5, 609254 |

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|--------|--------|--------|--------|-------|--|
| ITGA3 | 100.0% | 100.0% | 100.0% | 99.4% | Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748 |
| ITGA8 | 100.0% | 100.0% | 100.0% | 98.6% | Renal hypodysplasia/aplasia 1, 191830 |
| ITSN1 | 100.0% | 100.0% | 100.0% | 97.9% | |
| ITSN2 | 100.0% | 100.0% | 99.9% | 97.0% | |
| JAG1 | 100.0% | 100.0% | 100.0% | 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% | 100.0% | 99.4% | Cerebral palsy, spastic quadriplegic, 2, 612900 |
| KANK2 | 100.0% | 100.0% | 100.0% | 99.6% | Nephrotic syndrome, type 16, 617783;Palmoplantar keratoderma and woolly hair, 616099 |
| KATNIP | 100.0% | 100.0% | 100.0% | 99.2% | Joubert syndrome 26, 616784 |
| KCNJ1 | 100.0% | 100.0% | 100.0% | 98.3% | Bartter syndrome, type 2, 241200 |

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| KCNJ10 | 100.0% | 100.0% | 100.0% | 99.5% | Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780 |
| KCNJ16 | 100.0% | 100.0% | 100.0% | 99.4% | Hypokalemic tubulopathy and deafness, 619406 |
| KCNJ5 | 100.0% | 100.0% | 100.0% | 99.0% | Long QT syndrome 13, 613485;Hyperaldosteronism , familial, type III, 613677 |
| KIF14 | 100.0% | 100.0% | 100.0% | 98.4% | Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258 |
| KIF7 | 100.0% | 99.9% | 100.0% | 98.4% | Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydroletharus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131 |
| KIRREL1 | 100.0% | 100.0% | 100.0% | 99.4% | Nephrotic syndrome, type 23, 619201 |
| KL | 99.8% | 99.2% | 99.6% | 96.1% | ?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994 |
| KLHL3 | 100.0% | 100.0% | 100.0% | 99.7% | Pseudohypoaldosteronism, type IID, 614495 |
| LAGE3 | 100.0% | 100.0% | 95.4% | 68.7% | Galloway-Mowat syndrome 2, X-linked, 301006 |

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| LAMA5 | 100.0% | 100.0% | 100.0% | 99.0% | Nephrotic syndrome, type 26, 620049;?Bent bone dysplasia syndrome 2, 620076 |
| LAMB2 | 100.0% | 100.0% | 100.0% | 99.8% | Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049 |
| LCAT | 100.0% | 100.0% | 100.0% | 98.9% | Fish-eye disease, 136120;Norum disease, 245900 |
| LMX1B | 100.0% | 100.0% | 99.9% | 94.8% | Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200 |
| LRIG2 | 100.0% | 100.0% | 100.0% | 98.4% | Urofacial syndrome 2, 615112 |
| LRP2 | 100.0% | 100.0% | 100.0% | 99.0% | Donnai-Barrow syndrome, 222448 |
| LRP4 | 100.0% | 100.0% | 100.0% | 99.3% | ?Myasthenic syndrome, congenital, 17, 616304;Sclerosteosis 2, 614305;Cenani-Lenz syndactyly syndrome, 212780 |

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| LRP5 | 100.0% | 100.0% | 99.8% | 98.2% | Osteopetrosis, autosomal dominant 1, 607634;[Bone mineral density variability 1], 601884;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;Exudative vitreoretinopathy 4, 601813 |
| LYZ | 100.0% | 100.0% | 100.0% | 99.2% | Amyloidosis, hereditary systemic 5, 620658 |
| LZTFL1 | 100.0% | 100.0% | 100.0% | 97.7% | Bardet-Biedl syndrome 17, 615994 |
| MAFB | 100.0% | 100.0% | 100.0% | 98.5% | Duane retraction syndrome 3, 617041;Multicentric carpotarsal osteolysis syndrome, 166300 |
| MAGED2 | 100.0% | 99.9% | 97.6% | 69.6% | Bartter syndrome, type 5, antenatal, transient, 300971 |
| MAGI2 | 98.9% | 97.3% | 99.1% | 91.2% | Nephrotic syndrome, type 15, 617609 |
| MAPKBP1 | 100.0% | 100.0% | 100.0% | 99.2% | Nephronophthisis 20, 617271 |
| MKKS | 100.0% | 100.0% | 100.0% | 99.3% | McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231 |

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| MKS1 | 99.0% | 99.0% | 100.0% | 98.9% | Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121 |
| MMACHC | 100.0% | 100.0% | 100.0% | 98.8% | Methylmalonic aciduria and homocystinuria, cb1C type, 277400 |
| MOCOS | 100.0% | 100.0% | 100.0% | 98.7% | Xanthinuria, type II, 603592 |
| MYH9 | 97.2% | 97.2% | 100.0% | 98.8% | Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622 |
| MYO1E | 100.0% | 100.0% | 100.0% | 98.9% | Glomerulosclerosis, focal segmental, 6, 614131 |
| NCAPG2 | 100.0% | 100.0% | 100.0% | 98.7% | Khan-Khan-Katsanis syndrome, 618460 |
| NEK1 | 100.0% | 100.0% | 100.0% | 98.2% | Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 |

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| NEK8 | 100.0% | 100.0% | 100.0% | 99.5% | Renal-hepatic-pancreatic dysplasia 2, 615415;Polycystic kidney disease 8, 620903;?Nephronophthisis 9, 613824 |
| NEU1 | 100.0% | 100.0% | 100.0% | 99.4% | Sialidosis, type II, 256550;Sialidosis, type I, 256550 |
| NHERF1 | 100.0% | 100.0% | 100.0% | 97.2% | Nephrolithiasis/osteoporosis , hypophosphatemic, 2, 612287 |
| NOS1AP | 100.0% | 100.0% | 100.0% | 98.7% | Nephrotic syndrome, type 22, 619155 |
| NOTCH2 | 100.0% | 100.0% | 100.0% | 99.5% | Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500 |
| NPHP1 | 100.0% | 100.0% | 100.0% | 98.8% | Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900 |
| NPHP3 | 100.0% | 100.0% | 100.0% | 98.3% | Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010 |
| NPHP4 | 100.0% | 100.0% | 100.0% | 99.5% | Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966 |
| NPHS1 | 100.0% | 100.0% | 100.0% | 98.5% | Nephrotic syndrome, type 1, 256300 |

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| NPHS2 | 100.0% | 100.0% | 100.0% | 98.3% | Nephrotic syndrome, type 2, 600995 |
| NR3C2 | 100.0% | 100.0% | 100.0% | 98.6% | Pseudohypoaldosteronism type I, autosomal dominant, 177735;Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 |
| NUP107 | 100.0% | 100.0% | 100.0% | 98.3% | ?Ovarian dysgenesis 6, 618078;Galloway-Mowat syndrome 7, 618348;Nephrotic syndrome, type 11, 616730 |
| NUP133 | 100.0% | 100.0% | 100.0% | 98.3% | ?Galloway-Mowat syndrome 8, 618349;Nephrotic syndrome, type 18, 618177 |
| NUP160 | 100.0% | 100.0% | 100.0% | 98.5% | ?Nephrotic syndrome, type 19, 618178 |
| NUP205 | 100.0% | 100.0% | 100.0% | 98.9% | ?Nephrotic syndrome, type 13, 616893 |
| NUP85 | 100.0% | 100.0% | 99.9% | 97.4% | Nephrotic syndrome, type 17, 618176 |
| NUP93 | 95.5% | 95.5% | 100.0% | 99.2% | Nephrotic syndrome, type 12, 616892 |
| NXF5 | | | | | |
| OCRL | 100.0% | 100.0% | 97.8% | 69.7% | Dent disease 2, 300555;Lowe syndrome, 309000 |

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| OFD1 | 100.0% | 100.0% | 96.1% | 66.2% | Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804 |
| OSGEP | 100.0% | 100.0% | 100.0% | 99.0% | Galloway-Mowat syndrome 3, 617729 |
| OXGR1 | 100.0% | 100.0% | 100.0% | 98.9% | Nephrolithiasis, calcium oxalate, 2, with nephrocalcinosis, 620374 |
| PAX2 | 100.0% | 100.0% | 100.0% | 97.5% | Glomerulosclerosis, focal segmental, 7, 616002;Papillorenal syndrome, 120330 |
| PBX1 | 100.0% | 99.9% | 100.0% | 98.4% | Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 |
| PCBD1 | 100.0% | 100.0% | 100.0% | 99.4% | Hyperphenylalaninemia, BH4-deficient, D, 264070 |
| PDE6D | 100.0% | 100.0% | 100.0% | 97.0% | Joubert syndrome 22, 615665 |
| PDSS2 | 100.0% | 100.0% | 100.0% | 98.5% | Coenzyme Q10 deficiency, primary, 3, 614652 |
| PHEX | 99.9% | 99.2% | 98.1% | 70.9% | Hypophosphatemic rickets, X-linked dominant, 307800 |

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| PKD1 | 99.9% | 99.7% | 100.0% | 98.3% | Polycystic kidney disease 1, 173900 |
| PKD2 | 100.0% | 100.0% | 99.8% | 93.4% | Polycystic kidney disease 2, 613095 |
| PKHD1 | 100.0% | 100.0% | 100.0% | 98.8% | Polycystic kidney disease 4, with or without hepatic disease, 263200 |
| PLCE1 | 100.0% | 99.8% | 100.0% | 98.5% | Nephrotic syndrome, type 3, 610725 |
| PMM2 | 100.0% | 100.0% | 100.0% | 98.2% | Congenital disorder of glycosylation, type Ia, 212065 |
| PODXL | 94.2% | 93.8% | 99.9% | 95.8% | |
| PTH1R | 100.0% | 100.0% | 100.0% | 99.3% | Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045 |
| PTPRO | 99.8% | 99.1% | 100.0% | 98.8% | Nephrotic syndrome, type 6, 614196 |
| RAD21 | 100.0% | 100.0% | 100.0% | 98.3% | Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376 |

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| REN | 100.0% | 100.0% | 100.0% | 98.6% | Renal tubular dysgenesis, 267430;Tubulointerstitial kidney disease, autosomal dominant, 4, 613092;[Hyperproreninemia], |
| RMND1 | 85.6% | 85.6% | 100.0% | 97.6% | Combined oxidative phosphorylation deficiency 11, 614922 |
| RNU4ATAC | | | | | Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710 |
| ROBO2 | 100.0% | 100.0% | 100.0% | 99.0% | Vesicoureteral reflux 2, 610878 |
| RPGRIP1L | 100.0% | 100.0% | 100.0% | 97.4% | Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113 |
| RRAGD | 100.0% | 100.0% | 100.0% | 97.7% | Hypomagnesemia 7, renal, with or without dilated cardiomyopathy, 620152 |

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| RRM2B | 100.0% | 100.0% | 100.0% | 97.7% | 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 | 100.0% | 100.0% | 100.0% | 98.4% | Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480 |
| SALL4 | 100.0% | 100.0% | 100.0% | 99.1% | ?IVIC syndrome, 147750;Duane-radial ray syndrome, 607323 |
| SARS2 | 100.0% | 100.0% | 100.0% | 98.6% | Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 |
| SCARB2 | 100.0% | 100.0% | 100.0% | 99.1% | Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 |

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| SCNN1A | 100.0% | 100.0% | 100.0% | 98.6% | Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350;?Liddle syndrome 3, 618126;Bronchiectasis with or without elevated sweat chloride 2, 613021 |
| SCNN1B | 100.0% | 100.0% | 100.0% | 99.3% | Bronchiectasis with or without elevated sweat chloride 1, 211400;Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125;Liddle syndrome 1, 177200 |
| SCNN1G | 100.0% | 100.0% | 100.0% | 99.4% | Bronchiectasis with or without elevated sweat chloride 3, 613071;Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126;Liddle syndrome 2, 618114 |
| SDCCAG8 | 100.0% | 100.0% | 100.0% | 97.9% | Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993 |
| SEC61A1 | 100.0% | 100.0% | 100.0% | 98.0% | Immunodeficiency, common variable, 15, 620670;?Neutropenia, severe congenital, 11, autosomal dominant, 620674;Tubulointerstitial kidney disease, autosomal dominant, 5, 617056 |
| SGPL1 | 96.6% | 96.6% | 100.0% | 99.1% | REN1 syndrome, 617575 |

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|----------|--------|--------|--------|-------|---|
| SIX5 | 100.0% | 100.0% | 99.8% | 95.0% | Branchiootorenal syndrome 2, 610896 |
| SLC12A1 | 96.4% | 96.3% | 100.0% | 98.4% | Bartter syndrome, type 1, 601678 |
| SLC12A3 | 100.0% | 100.0% | 100.0% | 99.0% | Gitelman syndrome, 263800 |
| SLC16A12 | 100.0% | 100.0% | 100.0% | 99.0% | Cataract 47, juvenile, with microcornea, 612018 |
| SLC22A12 | 100.0% | 99.8% | 99.9% | 97.2% | Hypouricemia, renal, 220150 |
| SLC26A1 | 100.0% | 100.0% | 100.0% | 99.8% | ?Hypersulfaturia, 620372;?Nephrolithiasis, calcium oxalate, 1, 167030 |
| SLC26A3 | 100.0% | 100.0% | 100.0% | 98.8% | Diarrhea 1, secretory chloride, congenital, 214700 |
| SLC2A2 | 100.0% | 100.0% | 100.0% | 99.4% | Fanconi-Bickel syndrome, 227810;{Diabetes mellitus, noninsulin-dependent}, 125853 |
| SLC2A9 | 100.0% | 100.0% | 100.0% | 98.9% | {Uric acid concentration, serum, QTL 2}, 612076;Hypouricemia, renal, 2, 612076 |
| SLC34A1 | 100.0% | 100.0% | 100.0% | 98.7% | ?Fanconi renal tubular syndrome 2, 613388;Hypercalcemia, infantile, 2, 616963;Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 |

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| SLC34A3 | 100.0% | 100.0% | 100.0% | 97.7% | Hypophosphatemic rickets with hypercalciuria, 241530 |
| SLC36A2 | 100.0% | 100.0% | 100.0% | 98.7% | [Iminoglycinuria], 242600;[Hyperglycinuria], 138500 |
| SLC37A4 | 100.0% | 100.0% | 100.0% | 99.7% | Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240 |
| SLC3A1 | 96.2% | 96.2% | 100.0% | 99.0% | Cystinuria, 220100 |
| SLC41A1 | 100.0% | 100.0% | 100.0% | 99.3% | ?Nephronophthisis-like nephropathy 2, 619468 |
| SLC4A1 | 100.0% | 100.0% | 100.0% | 99.2% | [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 | 97.3% | 97.0% | 100.0% | 98.2% | Proximal renal tubular acidosis-ocular anomaly syndrome, 604278 |

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| SLC5A2 | 100.0% | 100.0% | 100.0% | 99.3% | Renal glucosuria, 233100 |
| SLC6A19 | 100.0% | 100.0% | 100.0% | 99.4% | Hartnup disorder, 234500 |
| SLC6A20 | 100.0% | 100.0% | 100.0% | 99.5% | |
| SLC7A7 | 100.0% | 100.0% | 100.0% | 98.8% | Lysinuric protein intolerance, 222700 |
| SLC7A9 | 100.0% | 100.0% | 100.0% | 99.0% | Cystinuria, 220100 |
| SLC9A3 | 100.0% | 99.6% | 99.9% | 94.6% | Diarrhea 8, secretory sodium, congenital, 616868 |
| SLIT3 | 100.0% | 100.0% | 100.0% | 99.4% | |
| SMARCAL1 | 100.0% | 100.0% | 100.0% | 98.8% | Schimke immunoosseous dysplasia, 242900 |
| SOX17 | 100.0% | 100.0% | 100.0% | 99.7% | Vesicoureteral reflux 3, 613674 |
| STRA6 | 100.0% | 100.0% | 100.0% | 98.8% | Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186 |
| STX16 | 100.0% | 100.0% | 100.0% | 98.4% | Pseudohypoparathyroidism 1b, 603233 |
| TBC1D8B | 100.0% | 99.7% | 97.3% | 70.5% | Nephrotic syndrome, type 20, 301028 |
| TBX18 | 100.0% | 100.0% | 100.0% | 98.5% | Congenital anomalies of kidney and urinary tract 2, 143400 |
| TCTN1 | 97.8% | 96.4% | 100.0% | 97.5% | Joubert syndrome 13, 614173 |
| TCTN2 | 98.5% | 98.5% | 100.0% | 99.1% | Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885 |

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| TCTN3 | 100.0% | 100.0% | 100.0% | 98.9% | Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860 |
| THBD | 100.0% | 100.0% | 100.0% | 97.1% | Thrombophilia 12 due to thrombomodulin defect, 614486;{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 |
| TMEM107 | 100.0% | 100.0% | 100.0% | 98.7% | Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562 |
| TMEM138 | 100.0% | 96.8% | 100.0% | 98.7% | Joubert syndrome 16, 614465 |
| TMEM216 | 100.0% | 100.0% | 100.0% | 98.6% | Joubert syndrome 2, 608091;Meckel syndrome 2, 603194 |
| TMEM231 | 93.2% | 93.2% | 100.0% | 99.5% | Joubert syndrome 20, 614970;Meckel syndrome 11, 615397 |
| TMEM237 | 98.2% | 98.2% | 99.9% | 97.8% | Joubert syndrome 14, 614424 |
| TMEM260 | 100.0% | 100.0% | 100.0% | 97.9% | Structural heart defects and renal anomalies syndrome, 617478 |

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| TMEM67 | 96.1% | 96.1% | 100.0% | 95.5% | 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 | 100.0% | 100.0% | 100.0% | 99.3% | |
| TNXB | 100.0% | 100.0% | 100.0% | 98.9% | Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963 |
| TP53RK | 100.0% | 100.0% | 100.0% | 99.2% | Galloway-Mowat syndrome 4, 617730 |
| TPRKB | 82.0% | 81.2% | 100.0% | 98.0% | Galloway-Mowat syndrome 5, 617731 |
| TRAF3IP1 | 100.0% | 100.0% | 100.0% | 96.4% | Senior-Loken syndrome 9, 616629 |
| TRIM32 | 100.0% | 100.0% | 100.0% | 99.9% | ?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 |
| TRPC6 | 100.0% | 100.0% | 100.0% | 98.8% | Glomerulosclerosis, focal segmental, 2, 603965 |
| TRPM6 | 100.0% | 100.0% | 100.0% | 98.4% | Hypomagnesemia 1, intestinal, 602014 |

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| TSC1 | 100.0% | 100.0% | 100.0% | 98.8% | Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangi leiomyomatosis, 606690 |
| TSC2 | 100.0% | 100.0% | 100.0% | 99.5% | Lymphangi leiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254 |
| TTC21B | 98.2% | 97.6% | 100.0% | 98.6% | Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820 |
| TTC8 | 100.0% | 99.9% | 100.0% | 97.9% | Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464 |
| TULP3 | 100.0% | 100.0% | 100.0% | 99.2% | Hepatorenocardiac degenerative fibrosis, 619902 |
| UMOD | 100.0% | 100.0% | 100.0% | 99.1% | Tubulointerstitial kidney disease, autosomal dominant, 1, 162000 |
| UPK3A | 100.0% | 100.0% | 100.0% | 99.5% | |
| UQCC2 | 100.0% | 100.0% | 100.0% | 99.8% | Mitochondrial complex III deficiency, nuclear type 7, 615824 |
| VDR | 100.0% | 100.0% | 100.0% | 98.1% | Rickets, vitamin D-resistant, type IIA, 277440 |

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|---------|--------|--------|--------|-------|---|
| VIPAS39 | 100.0% | 100.0% | 100.0% | 99.2% | Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 |
| VPS33B | 100.0% | 100.0% | 100.0% | 98.5% | Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 |
| WDR19 | 100.0% | 100.0% | 99.9% | 97.7% | 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 | 100.0% | 100.0% | 100.0% | 98.9% | Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610 |
| WDR73 | 100.0% | 100.0% | 100.0% | 98.4% | Galloway-Mowat syndrome 1, 251300 |
| WNK1 | 100.0% | 100.0% | 100.0% | 98.7% | Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492 |

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|----------|--------|--------|--------|-------|---|
| WNK4 | 100.0% | 100.0% | 100.0% | 97.5% | Pseudohypoaldosteronism, type IIB, 614491 |
| WNT4 | 100.0% | 99.8% | 99.7% | 95.5% | ?SERKAL syndrome, 611812;Mullerian aplasia and hyperandrogenism, 158330 |
| WT1 | 100.0% | 100.0% | 99.9% | 96.2% | Mesothelioma, somatic, 156240;Meacham syndrome, 608978;Frasier syndrome, 136680;Nephrotic syndrome, type 4, 256370;Denys-Drash syndrome, 194080;Wilms tumor, type 1, 194070 |
| XDH | 100.0% | 100.0% | 100.0% | 99.2% | Xanthinuria, type I, 278300 |
| XPNPEP3 | 100.0% | 100.0% | 100.0% | 99.2% | Nephronophthisis-like nephropathy 1, 613159 |
| XPO5 | 100.0% | 100.0% | 100.0% | 99.0% | |
| ZMPSTE24 | 100.0% | 100.0% | 100.0% | 98.7% | Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210 |
| ZNF423 | 100.0% | 100.0% | 100.0% | 99.2% | Nephronophthisis 14, 614844;Joubert syndrome 19, 614844 |
| ZNG1A | 99.0% | 97.8% | 97.4% | 93.1% | |

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.

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.

srWGS GRCh38 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 GRCh38 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 : March 17th, 2023. This list is accurate for panel version DG 4.0.0

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors