## Supplement Table 3. Priority Nephropathy Genes

| NCBI GENE ID | GENE SYMBOL | SYNDROME NAME | OMIM ID | INHERITANCE | N Exons Not Targeted by Agilent Kit |
|---|---|---|---|---|---|
| **1636** | *ACE* | Renal tubular dysgenesis | (+)106180 | recessive | 0 |
| **81** | *ACTN4* | Nephrotic syndrome (FSGS 1) | *604638 | dominant | 0 |
| **11093** | *ADAMTS13* | Thrombotic thrombocytopenic purpura, familial | #274150 | dominant | 0 |
| **79934** | *ADCK4* | Coq10 deficiency | *615567 | recessive | 0 |
| **183** | *AGT* | Renal tubular dysgenesis | (+)106150 | recessive | 0 |
| **185** | *AGTR1* | Renal tubular dysgenesis | *106165 | recessive | 0 |
| **189** | *AGXT* | Hyperoxaluria, primary, type I | 604285 | recessive | 0 |
| **54806** | *AHI1* | Nephronophtisis (Joubert syndrome 3) | *608894 | recessive | 0 |
| **79053** | *ALG8* | Congenital disorder of glycosylation type Ih, CDG1H | 608103 | recessive | 0 |
| **7840** | *ALMS1* | Alstrom syndrome | 606844 | recessive | 1 |
| **81693** | *AMN* | Megaloblastic anemia 1-Norwegian Type | 605799 | recessive | 0 |
| **362515** | *ANKS6* | Nephronophthisis 16 | 615370 | recessive | 0 |
| **1175** | *AP2S1* | Hypocalciuric hypercalcemia, familial type III | *602242 | dominant | 0 |
| **335** | *APOA1* | Amyloidosis, familial visceral, 3 or more types | 107680 | dominant | 0 |
| **348** | *APOE* | Lipoprotein glomerulopathy, LPG | 107741 | dominant | 0 |
| **8542** | *APOL1* | Nephrotic syndrome (FSGS 4) | *603743 | dominant | 0 |
| **353** | *APRT* | Adenine phosphoribosyltransferase deficiency | 102600 | recessive | 0 |
| **359** | *AQP2* | Diabetes insipidus, nephrogenic | *107777 | recessive | 0 |
| **200894** | *ARL13B* | Nephronophtisis (Joubert syndrome 8) | *608922 | recessive | 0 |
| **84100** | *ARL6* | Bardet-Biedl syndrome 3 | 608845 | recessive | 0 |
| **488** | *ATP2A2* | Darier-White disease, DAR | 108740 | dominant | 0 |
| **525** | *ATP6B1* | Renal tubular acidosis with deafness | *192132 | recessive | 0 |
| **50617** | *ATP6V0A4* | Renal tubular acidosis, distal, autosomal recessive | *605239 | recessive | 0 |
| **551** | *AVP* | Diabetes insipidus, neurohypophyseal | *192340 | dominant | 0 |
| **554** | *AVPR2* | Diabetes insipidus, nephrogenic | *300538 | X-linked | 1 |
| **582** | *BBS1* | Bardet-Biedl syndrome 1 | 209901 | recessive | 0 |

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| **79738** | *BBS10* | Bardet-Biedl syndrome 10 | 610148 | recessive | 0 |
| **166379** | *BBS12* | Bardet-Biedl syndrome 12 | 610683 | recessive | 0 |
| **583** | *BBS2* | Bardet-Biedl syndrome 2 | 606151 | recessive | 0 |
| **585** | *BBS4* | Bardet-Biedl syndrome 4 | 600374 | recessive | 0 |
| **129880** | *BBS5* | Bardet-Biedl syndrome 5 | 603650 | recessive | 0 |
| **55212** | *BBS7* | Bardet-Biedl syndrome 7 | 607590 | recessive | 0 |
| **607968** | *BBS9* | Bardet-Biedl syndrome 9 | 607968 | recessive | 0 |
| **617** | *BCS1L* | Mitochondrial complex III deficiency, nuclear type 1 (Bjornstad syndrome, GRACILE syndrome, Leigh syndrome) | 603647 | recessive | 0 |
| **652** | *BMP4* | Renal hypodysplasia/agenesis | *112262 | dominant | 0 |
| **673** | *BRAF* | Cardiofaciocutaneous syndrome | 164757 | dominant | 0 |
| **83990** | *BRIP1* | Fanconi anemia, complementation group J | 605882 | recessive | 0 |
| **7809** | *BSND* | Bartter syndrome, type 4a | *606412 | recessive | 0 |
| **718** | *C3* | Hemolytic uremic syndrome, atypical, (susceptibility to, 5) | *120700 | dominant | 0 |
| **760** | *CA2* | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis | *611492 | recessive | 0 |
| **779** | *CACNA1S* | Hypokalemic periodic paralysis, hokpp | 114208 | dominant | 0 |
| **846** | *CASR* | Hypocalcemia, autosomal dominant, with Bartter syndrome | (+)601199 | dominant | 0 |
| **57545** | *CC2D2A* | Nephronophthisis (Joubert syndrome 9) | *612013 | recessive | 2 |
| **79140** | *CCDC28B* | {Bardet-Biedl syndrome, modifier of} | 610162 | recessive | 0 |
| **977** | *CD151* | Nephropathy with pretibial epidermolysis bullosa and deafness | 602243 | recessive | 0 |
| **23607** | *CD2AP* | Nephrotic syndrome (FSGS 3) | *604241 | dominant | 1 |
| **988** | *CDC5L* | Congenital anomalies of kidney and urinary tract 2, CAKUT 2 | 602868 | | 0 |
| **1028** | *CDKN1C* | Beckwith-Wiedemann syndrome, BWS | 600856 | dominant | 0 |
| **22897** | *CEP164* | Nephronopthisis 15 | *614848 | recessive | 0 |
| **80184** | *CEP290* | Nephronophtisis (Joubert syndrome 5, Senior-Loken syndrome 6, Bardet-Biedl) | *610142 | recessive | 1 |
| **629** | *CFB* | Hemolytic uremic syndrome, atypical, (susceptibility to, 4) | (+)138470 | dominant | 0 |

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| **3075** | *CFH* | MPGN (Complement factor H deficiency) | *134370 | dominant | 0 |
| **3078** | *CFHR1* | Hemolytic uremic syndrome, atypical, (susceptibility to} | *134371 | dominant | 1 |
| **10878** | *CFHR3* | MPGN (atypical HUS) | *605336 | dominant | 3 |
| **81494** | *CFHR5* | MPGN (CFHR5 deficiency ) | *608593 | dominant | 0 |
| **3426** | *CFI* | Hemolytic uremic syndrome, atypical, (susceptibility to, 3) | *217030 | dominant | 0 |
| **55636** | *CHD7* | CHARGE syndrome | 608892 | dominant | 0 |
| **1131** | *CHRM3* | Eagle-Barrett syndrome (Prune-Belly syndrome) | *118494 | recessive | 0 |
| **51057** | *C2orf86* | Bardet-Biedl syndrome 15 | 613580 | recessive | 0 |
| **1188** | *CLCKNB* | Bartter syndrome, type 3 and type 4b | *602023 | recessive | 0 |
| **1184** | *CLCN5* | Dent disease | *300008 | X-linked | 0 |
| **10686** | *CLDN16* | Hypomagnesemia 3, renal | *603959 | recessive | 0 |
| **149461** | *CLDN19* | Hypomagnesemia 5, renal, with ocular involvement | *610036 | recessive | 0 |
| **1285** | *COL4A3* | Alport syndrome | *120070 | Dominant or recessive | 0 |
| **1286** | *COL4A4* | Alport syndrome | *120131 | Dominant or recessive | 0 |
| **1287** | *COL4A5* | Alport syndrome | *303630 | X-linked | 2 |
| **1288** | *COL4A6* | Alport syndrome with leiomyomatosis | *303631 | X-linked | 2 |
| **27235** | *COQ2* | Coenzyme Q10 deficiency | 609825 | recessive | 0 |
| **51004** | *COQ6* | Nephrotic syndrome | *614647 | recessive | 0 |
| **1376** | *CPT2* | Lethal neonatal carnitine palmitoyltransferase ii deficiency (CPT deficiency hepatic type II, Myopathy due to CPT II deficiency, Encephalopathy acute infection-induced 4 susceptibility to | 600650 | recessive | 0 |
| **1387** | *CREBBP* | Rubinstein Taybi syndrome type 1 | 600140 | dominant | 0 |
| **79848** | *CSPP1* | Joubert Syndrome | *611654 | recessive | 0 |
| **1479** | *CTNS* | Cystinosis | *606272 | recessive | 0 |
| **8029** | *CUBN* | Megaloblastic anemia 1-Finnish Type | 602997 | recessive | 0 |
| **8452** | *CUL3* | Pseudohypoaldosteronism, type IIE | *603136 | dominant | 0 |
| **1584** | *CYP11B1* | Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency | *610613 | recessive | 2 |
| **1586** | *CYP17A1* | 17-alpha-hydroxylase/17,20-lyase deficiency | *609300 | recessive | 0 |
| **8526** | *DGKE* | Nephrotic syndrome type 7 | #615008 | recessive | 0 |

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| **1717** | *DHCR7* | Smith Lemli Opitz syndrome | 602858 | recessive | 0 |
| **1741** | *DLG3* | Mental retardation, X-linked 90 | 300189 | X-linked | 0 |
| **10683** | *DLL3* | Spondylocostal dysostosis 1 | 602768 | recessive | 0 |
| **1789** | *DNMT3B* | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, ICF1 | 602900 | recessive | 0 |
| **25778** | *DSTYK* | Congenital anomalies of kidney and urinary tract 1, susceptibility to; CAKUT 1 | *612666 | dominant | 0 |
| **1950** | *EGF* | Hypomagnesemia 4, renal | *131530 | dominant | 0 |
| **13666** | *EIF2AK3* | Multiple epiphyseal dysplasia with early-onset diabetes mellitus | 604032 | recessive | 0 |
| **2072** | *ERCC4* | XFE progeroid syndrome, Fanconi anemia, complementation group Q, Fanconi anemia, complementation group Q, Xeroderma pigmentosum, type F/Cockayne syndrome | 133520 | Recessive | 0 |
| **1161** | *ERCC8* | Cockayne syndrome, type a; CSA | 609412 | recessive | 0 |
| **157570** | *ESCO2* | Roberts syndrome, SC phocomelia syndrome | 609353 | recessive | 0 |
| **2108** | *ETFA* | Glutaric aciduria IIA | 608053 | recessive | 0 |
| **2109** | *ETFB* | Glutaric acidemia IIB | 130410 | recessive | 0 |
| **2110** | *ETFDH* | Glutaric acidemia IIC | 231675 | recessive | 0 |
| **2138** | *EYA1* | CAKUT (Branchio-oto-renal syndrome 1) | *601653 | dominant | 0 |
| **22909** | *FAN1* | Interstitial nephritis, karyomegalic | *613534 | recessive | 0 |
| **2175** | *FANCA* | Fanconi anemia, complementation group A | 607139 | recessive | 1 |
| **2187** | *FANCB* | VACTERL syndrome with hydrocephalus | 300515 | recessive | 0 |
| **2176** | *FANCC* | Fanconi anemia, complementation group C | 613899 | recessive | 0 |
| **2177** | *FANCD2* | Fanconi anemia, complementation group D2 | 613984 | recessive | 5 |
| **2178** | *FANCE* | Fanconi anemia | 613976 | recessive | 0 |
| **2188** | *FANCF* | Fanconi anemia | 613897 | recessive | 0 |
| **2189** | *FANCG (XRCC9)* | Fanconi anemia | 602956 | recessive | 0 |
| **55215** | *FANCI* | Fanconi anemia | 609053 | recessive | 0 |
| **55120** | *FANCL* | Fanconi anemia | 614083 | recessive | 0 |
| **57697** | *FANCM* | Fanconi anemia | 609644 | recessive | 0 |

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| **2243** | *FGA* | Amyloidosis, familial visceral, hereditary renal | 134820 | dominant | 0 |
| **2255** | *FGF10* | Lacrimoauriculodentodigital syndrome, LADD | 602115 | dominant | 0 |
| **26281** | *FGF20* | Renal hypoplasia/adysplasia 2 | *605558 | recessive | 0 |
| **8074** | *FGF23* | Hypophosphatemic rickets, autosomal dominant | *605380 | dominant | 0 |
| **2260** | *FGFR1* | Kallmann syndrome 2, KAL2 | 136350 | recessive | 0 |
| **2263** | *FGFR2* | Antley-Bixler syndrome, ABS | 176943 | recessive | 0 |
| **2261** | *FGFR3* | Crouzon syndrome with acanthosis nigricans, CAN | 134934 | dominant | 0 |
| **2335** | *FN1* | MPGN (Glomerulopathy with fibronectin deposits 2) | *135600 | dominant | 0 |
| **2296** | *FOXC1* | Iridogoniodysgenesis, type 1; IRID1 | 601090 | dominant | 0 |
| **2303** | *FOXC2* | Lymphedema-distichiasis syndrome | 602402 | dominant | 0 |
| **2294** | *FOXF1* | Alveolar capillary dysplasia with misalignment of pulmonary veins, ACDMPV | 601089 | dominant | 0 |
| **80144** | *FRAS1* | CAKUT (Fraser Syndrome) | *607830 | recessive | 0 |
| **158326** | *FREM1* | CAKUT (Bifid nose with or without anorectal and renal anomalies) | *608944 | recessive | 0 |
| **65211** | *FRTS* | Fanconi syndrome | 134600% | dominant | 0 |
| **486** | *FXYD2* | Hypomagnesemia-2, renal | *601814 | dominant | 0 |
| **2591** | *GALNT3* | Tumoral calcinosis, hyperphosphatemic, familial, HFTC | 601756 | recessive | 0 |
| **2668** | *GDNF* | CAKUT | *600837 | dominant | 0 |
| **2717** | *GLA* | Fabry disease | *300644 | X-linked | 0 |
| **84662** | *GLIS2* | Nephronophthisis 7 | *608539 | recessive | 0 |
| **2719** | *GPC3* | Simpson Golabi Behmel syndrome type 1 | 300037 | X-linked | 0 |
| **9380** | *GRHPR* | Hyperoxaluria, primary, type II | 604296 | recessive | 0 |
| **2934** | *GSN* | Amyloidosis, Finnish type cerebral amyloid angiopathy, GSN-related | 137350 | dominant | 1 |
| **283120** | *H19* | Beckwith-Wiedemann syndrome, BWS | 103280 | dominant | 0 |
| **6928** | *HNF1B* | CAKUT (Renal cysts and diabetes, isolated hypoplasia) | *189907 | dominant | 1 |
| **3198** | *HOXA1* | Bosley Salih Alorainy syndrome | 142955 | recessive | 0 |
| **3209** | *HOXA13* | Hand-foot-uterus syndrome | 142959 | dominant | 0 |

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| **3239** | *HOXD13* | VACTERL association with hydrocephalus | 142989 | dominant | 0 |
| **60495** | *HPSE2* | CAKUT (urofacial syndrome) | *613469 | recessive | 0 |
| **3265** | *HRAS* | Costello syndrome | 190020 | recessive | 0 |
| **3291** | *HSD11B2* | Apparent mineralocorticoid excess | *614232 | recessive | 0 |
| **3295** | *HSD17B4* | D-bifunctional protein deficiency | 601860 | X-linked | 1 |
| **22858** | *ICK* | Endocrine-cerebroosteodysplasia, eco | 612325 | recessive | 0 |
| **8518** | *IKBKAP* | Hereditary sensory and autonomic neuropathy type iii, hsan3 | 603722 | recessive | 0 |
| **64423** | *INF2* | Nephrotic syndrome (FSGS 5) | *610982 | dominant | 2 |
| **56623** | *INPP5E* | Nephronophtisis (Joubert syndrome 1) | *613037 | recessive | 0 |
| **27130** | *INVS* | Nephronophthisis 2, infantile | *243305 | recessive | 0 |
| **9657** | *IQCB1* | Senior-Loken syndrome 5 | *609237 | recessive | 0 |
| **182** | *JAG1* | Alagille syndrome type 1 | 601920 | dominant | 0 |
| **95681** | *JBTS15* | Joubert syndrome 15 | #614464 | recessive | 0 |
| **3766** | *KCNJ10* | Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SESAME syndrome) | | recessive | 0 |
| **3762** | *KCNJ5* | Hyperaldosteronism, familial, type III | *600734 | dominant | 0 |
| **10984** | *KCNQ1OT1* | Beckwith-Wiedemann syndrome, BWS | 604115 | dominant | 0 |
| **26249** | *KLHL3* | Pseudohypoaldosteronism, type IID | *605775 | dominant | 0 |
| **3845** | *KRAS* | Cardiofaciocutaneous syndrome 2 | 190070 | dominant | 0 |
| **3913** | *LAMB2* | Nephrotic syndrome, type 5, with or without ocular abnormalities | *150325 | recessive | 0 |
| **3931** | *LCAT* | Norum disease | 606967 | recessive | 0 |
| **16917** | *LMX1B* | Nephrotic syndrome (Nail-Patella syndrome) | *602575 | dominant | 0 |
| **9860** | *LRIG2* | CAKUT (urofacial syndrome) | *608869 | recessive | 0 |
| **4036** | *LRP2* | Donnai-Barrow syndrome | 600073 | recessive | 0 |
| **4038** | *LRP4* | Cenani-Lenz syndactyly syndrome; CSS | 604270 | recessive | 0 |
| **4069** | *LYZ* | Amyloidosis, familial visceral, renal | 153450 | dominant | 0 |
| **5604** | *MAP2K1* | Cardiofaciocutaneous syndrome 3 | 176872 | dominant | 0 |
| **5605** | *MAP2K2* | Cardiofaciocutaneous syndrome 4 | 601263 | dominant | 0 |

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| **4149** | *MAX* | Pheochromocytoma, (susceptibility to) | *154950 | dominant | 1 |
| **4179** | *MCP (CD46)* | Hemolytic uremic syndrome, atypical (susceptibility to, 2) | *120920 | dominant | 0 |
| **4210** | *MEFV* | Familial Mediterranean fever | 608107 | Dominant or recessive | 0 |
| **8195** | *MKKS* | Bardet-Biedl syndrome 6 | 604896 | recessive | 0 |
| **54903** | *MKS1* | Meckel syndrome 1 | *609883 | recessive | 0 |
| **8085** | *MLL2* | Kabuki syndrome 1 | *602113 | dominant | 0 |
| **25974** | *MMACHC* | Methylmalonic aciduria and homocystinuria, cblC type | 609831 | recessive | 0 |
| **4312** | *MMP1* | Epidermolysis bullosa dystrophica, autosomal recessive, RDEB | 120353 | recessive | 0 |
| **3110** | *MNX1* | VACTERL/Caudal regression syndrome/ Currarino syn | *142994 | dominant | 0 |
| **4361** | *MRE11A* | Ataxia-telangiectasia-like disorder | *600814 | recessive | 1 |
| **4582** | *MUC1* | Medullary cystic kidney disease | *158340 | dominant | 0 |
| **4594** | *MUT* | Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency | 609058 | recessive | 0 |
| **4598** | *MVK* | Hyper-IgD syndrome | 251170 | recessive | 0 |
| **4613** | *MYCN* | Feingold syndrome | 164840 | dominant | 0 |
| **4627** | *MYH9* | Nephrotic syndrome | *160775 | dominant | 0 |
| **4643** | *MYO1E* | Nephrotic syndrome (FSGS 6) | *601479 | recessive | 0 |
| **284086** | *NEK8* | Nephronophthisis 9 | *609799 | recessive | 0 |
| **25836** | *NIPBL* | Cornelia de Lange syndrome type 1 | 608667 | dominant | 0 |
| **6557** | *NKCC2* | Bartter syndrome, type 1 | *600839 | recessive | 0 |
| **114548** | *NLRP3* | Muckle-Wells syndrome | 606416 | dominant | 0 |
| **4853** | *NOTCH2* | Alagille syndrome 2 | 600275 | dominant | 4 |
| **4867** | *NPHP1* | Nephronophthisis 1 (juvenile) | *607100 | recessive | 1 |
| **27031** | *NPHP3* | Nephronophthisis 3 (adolescent) | *608002 | recessive | 0 |
| **261734** | *NPHP4* | Nephronophthisis 4 | *607215 | recessive | 0 |
| **4868** | *NPHS1* | Nephrotic syndrome type 1 (Finnish type) | *602716 | recessive | 0 |
| **7827** | *NPHS2* | Nephrotic syndrome type 2 | *604766 | recessive | 0 |
| **4306** | *NR3C2* | Pseudohypoaldosteronism type I, autosomal dominant | *600983 | dominant | 0 |
| **64324** | *NSD1* | Beckwith-Wiedemann syndrome, BWS | 606681 | dominant | 0 |
| **4952** | *OCRL* | Dent disease 2 | *300353 | X-linked | 0 |
| **8481** | *OFD1* | Joubert syndrome type 10 | 300170 | recessive | 2 |
| **54623** | *PAF1* | Zellweger syndrome 3, ZWS3 | 170993 | | 1 |

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| **79728** | *PALB2* | Fanconi anemia, complementation group N | 610355 | recessive | 0 |
| **5076** | *PAX2* | CAKUT (Papillorenal syndrome, isolated hypoplasia) | *167409 | dominant | 0 |
| **57107** | *PDSS2* | Coenzyme Q10 deficiency, primary, 3 | 610564 | recessive | 0 |
| **5189** | *PEX1* | Zellweger syndrome, ZS | 602136 | recessive | 0 |
| **5193** | *PEX12* | Zellweger syndrome, ZS (Peroxisome biogenesis disorder 3A) | 601758 | recessive | 0 |
| **5195** | *PEX14* | Zellweger syndrome, ZS (Peroxisome biogenesis disorder 13A) | 601791 | recessive | 0 |
| **55670** | *PEX26* | Zellweger syndrome, ZS (Peroxisome biogenesis disorder 7A) | 608666 | recessive | 0 |
| **5830** | *PEX5* | Zellweger syndrome, ZS | *611058 | recessive | 0 |
| **5190** | *PEX6* | Zellweger syndrome, ZS (Peroxisome biogenesis disorder 4A) | 610498 | recessive | 0 |
| **18675** | *PHEX* | Hypophosphatemic rickets, X-linked dominant | *300550 | X-linked | 0 |
| **5310** | *PKD1* | Polycystic kidney disease 1 | *601313 | dominant | 33 |
| **5311** | *PKD2* | Polycystic kidney disease 2 | *173910 | dominant | 0 |
| **5314** | *PKHD1* | Polycystic and hepatic disease | *606702 | recessive | 1 |
| **51196** | *PLCE1* | Nephrotic syndrome type 3 | *608414 | recessive | 0 |
| **60675** | *PROK2* | Kallmann syndrome 4, KAL4 (Hypogonadotropic hypogonadism 4 with or without anosmia) | 607002 | dominant | 0 |
| **128674** | *PROKR2* | Kallmann syndrome 3, KAL3 (Hypogonadotropic hypogonadism 3 with or without anosmia) | 607123 | dominant | 0 |
| **5728** | *PTEN* | Vacterl association with hydrocephalus | 601728 | recessive | 1 |
| **5781** | *PTPN11* | Leopard syndrome 1 | 176876 | dominant | 3 |
| **5800** | *PTPRO* | Nephrotic syndrome | *600579 | recessive | 0 |
| **5818** | *PVRL1* | Cleft lip/palate-ectodermal dysplasia syndrome, clped1 orofacial cleft 7, OFC7 | 600644 | recessive | 0 |
| **5828** | *PXMP3 (PEX2)* | Zellweger syndrome, ZS (Peroxisome biogenesis disorder 5A) | 170993 | recessive | 0 |
| **9401** | *RECQL4* | Baller-Gerold syndrome, BGS | 603780 | recessive | 0 |

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| **5972** | *REN* | Renal tubular dysgenesis / Hyperuricemic nephropathy, familial juvenile 2 | *179820 | dominant | 0 |
| **5979** | *RET* | CAKUT (renal agenesis) | (+)164761 | dominant | 0 |
| **6092** | *ROBO2* | CAKUT | *602431 | dominant | |
| **56379** | *ROMK* | Bartter syndrome, type 2 | *600359 | recessive | 0 |
| **4920** | *ROR2* | Robinow syndrome | 602337 | recessive | 0 |
| **23322** | *RPGRIP1L* | Nephronophtisis (Joubert Syndrome 6, Meckel syndrome 5) | *610937 | recessive | 0 |
| **55811** | *SAC (ADCY10)* | Hypercalciuria, absorptive, 2, HCA2 | 605205 | dominant | 0 |
| **6299** | *SALL1* | Townes-Brocks branchiootorenal-like syndrome | *602218 | dominant | 0 |
| **57167** | *SALL4* | CAKUT | *607343 | dominant | 0 |
| **950** | *SCARB2* | Action myoclonus-renal failure syndrome, AMRF | 602257 | recessive | 0 |
| **6329** | *SCN4A* | Hypokalemic periodic paralysis, HOKPP Type 2 | 603967 | dominant | 0 |
| **6337** | *SCNN1A* | Pseudohypoaldosteronism, type I | *600228 | recessive | 0 |
| **6338** | *SCNN1B* | sodium channel, non-voltage-gated 1, beta subunit | *600760 | dominant | 0 |
| **6340** | *SCNN1G* | Pseudohypoaldosteronism, type I (Liddle syndrome) | *600761 | dominant | 0 |
| **10806** | *SDCCAG8* | Senior Loken syndrome type 7 | 613524 | recessive | 0 |
| **6390** | *SDHB* | Pheochromocytoma | *185470 | dominant | 0 |
| **6392** | *SDHD* | Pheochromocytoma | *602690 | dominant | 4 |
| **9723** | *SEMA3E* | CHARGE syndrome | 608166 | dominant | 0 |
| **6469** | *SHH* | CAKUT | 600725 | dominant | 0 |
| **6495** | *SIX1* | CAKUT (Branchio-oto-renal syndrome 3) | *601205 | dominant | 0 |
| **10736** | *SIX2* | Renal hypodysplasia | *604994 | dominant | 0 |
| **147912** | *SIX5* | CAKUT (Branchio-oto-renal syndrome 2) | *600963 | dominant | 0 |
| **6559** | *SLC12A3* | Gitelman syndrome | *600968 | recessive | 0 |
| **116085** | *SLC22A12* | Hypouricemia, renal, 1, RHUC1 | 607096 | recessive | 0 |
| **5172** | *SLC26A4* | Pendred syndrome | 605646 | recessive | 0 |
| **81031** | *SLC2A10* | Arterial tortuosity syndrome, ATS | 606145 | recessive | 0 |
| **20505** | *SLC34A1* | Nephrolithiasis/osteoporosis, hypophosphatemic, 1 | *182309 | recessive | 0 |
| **142680** | *SLC34A3* | Hypophosphatemic rickets with hypercalciuria, hereditary, HHRH | 609826 | recessive | 0 |
| **6519** | *SLC3A1* | Cystinuria | *104614 | dominant | 0 |

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| **6521** | *SLC4A1* | Renal tubular acidosis, distal, AD or AR | (+)109270 | Dominant/ Recessive | 0 |
| **8671** | *SLC4A4* | Renal tubular acidosis, proximal, with ocular abnormalities | *603345 | recessive | 0 |
| **6524** | *SLC5A2* | Renal glucosuria | 182381 | dominant | 0 |
| **11136** | *SLC7A9* | Cystinuria | *604144 | dominant | 0 |
| **9368** | *SLC9A3R1* | Nephrolithiasis/osteoporosis, hypophosphatemic, 2 | *604990 | recessive | 0 |
| **9353** | *SLIT2* | CAKUT | *603746 | dominant | 0 |
| **338072** | *SMARCAL1* | Schimke's immunoosseous dystrophy | 242 900 | recessive | 0 |
| **8243** | *SMC1A* | Cornelia de Lange syndrome type 2 | 300040 | dominant | 0 |
| **6658** | *SOX3* | Intellectual disability X linked with isolated growth hormone deficiency | 313430 | X-linked | 0 |
| **64220** | *STRA6* | Microphthalmia, syndromic 9, MCOPS9 | 610745 | dominant | 0 |
| **8803** | *SUCLA2* | Mitochondrial DNA depletion syndrome, encephalomyopathic form, with methylmalonic aciduria, autosomal recessive | 603921 | recessive | 0 |
| **7020** | *TFAP2A* | Branchiooculofacial syndrome, BOFS | 107580 | dominant | 1 |
| **7056** | *THBD* | Hemolytic uremic syndrome, atypical, (susceptibility to, 6) | *188040 | dominant | 0 |
| **55654** | *TMEM127* | Pheochromocytoma (susceptibility to) | *613403 | dominant | 0 |
| **51259** | *TMEM216* | Nephronophtisis (Joubert syndrome 2) | *613277 | recessive | 0 |
| **65062** | *TMEM237* | Joubert syndrome type 14 | 614423 | recessive | 0 |
| **609884** | *TMEM67* | {Bardet-Biedl syndrome 14, modifier of} | 609884 | recessive | 0 |
| **91147** | *TMEM67* | Nephronophtisis 11 | *609884 | recessive | 0 |
| **7132** | *TNFRSF1A* | Autosomal Dominant Periodic Fever Syndrome | 191190 | dominant | 0 |
| **8626** | *TP63* | Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3 | 603273 | dominant | 0 |
| **10131** | *TRAP1* | CAKUT; VACTERL | *606219 | recessive | 0 |
| **22954** | *TRIM32* | Bardet-Biedl syndrome 11 | 602290 | recessive | 0 |
| **7225** | *TRPC6* | Nephrotic syndrome (FSGS 2) | 603652 | dominant | 0 |
| **140803** | *TRPM6* | Hypomagnesemia 1, intestinal | *607009 | dominant | 0 |
| **7248** | *TSC1* | Tuberous sclerosis-1 | *605284 | dominant | 0 |
| **7249** | *TSC2* | Tuberous sclerosis-2 | *191092 | dominant | 0 |
| **79809** | *TTC21B* | Nephronophthisis 12 | *612014 | recessive | 0 |

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| **123016** | *TTC8* | Bardet-Biedl syndrome 8 | 608132 | recessive | 0 |
| **7276** | *TTR* | Amyloidosis, hereditary, transthyretin-related | *176300 | dominant | 0 |
| **7337** | *UBE3A* | Angelman syndrome | 601623 | Complex | 0 |
| **07369** | *UMOD* | Medullary cystic kidney disease 2; Hyperuricemic nephropathy, familial juvenile 1 | *191845 | dominant | 0 |
| **7380** | *UPK3A* | Renal adysplasia, urogenital adysplasia | *611559 | dominant | 0 |
| **7381** | *UQCRB* | Mitochondrial complex III deficiency, nuclear type 3 | 191330 | recessive | 0 |
| **27089** | *UQCRQ* | Mitochondrial complex III deficiency, nuclear type 4 | 612080 | recessive | 0 |
| **7399** | *USH2A* | Retinitis pigmentosa type 39 | 608400 | recessive | 0 |
| **81839** | *VANGL1* | VACTERL / Caudal regression syndrome | *610132 | dominant | 0 |
| **7428** | *VHL* | Pheochromocytoma (Von Hippel Lindau syndrome) | *608537 | dominant | 0 |
| **63894** | *VIPAR (VIPAS39)* | Arthrogryposis, renal dysfunction, and cholestasis 2; ARCS2 | 613401 | dominant | 0 |
| **26276** | *VPS33B* | Arthrogryposis, renal dysfunction, and cholestasis 1; ARCS1 | 608552 | dominant | 0 |
| **57728** | *WDR19* | Nephronophthisis type 13 | 608151 | recessive | 0 |
| **7466** | *WFS1* | Wolfram syndrome type 1 | 606201 | recessive | 0 |
| **65125** | *WNK1* | Pseudohypoaldosteronism, type IIC | *605232 | dominant | 0 |
| **65266** | *WNK4* | Pseudohypoaldosteronism, type IIB | *601844 | dominant | 0 |
| **7473** | *WNT3* | Tetraamelia | 165330 | recessive | 0 |
| **7490** | *WT1* | CAKUT/nephrotic syndrome (Danys-Drash and Frasier syndrome) | *607102 | Dominant | 0 |
| **7498** | *XDH* | Xanthinuria, type I | 607633 | recessive | 0 |
| **63929** | *XPNPEP3* | Nephronophthisis-like nephropathy 1 | *613553 | recessive | 0 |
| **23090** | *ZNF423* | Joubert syndrome 16 | *604557 | recessive | 0 |