| KidneySeq™ Comprehensive | | |
|---|---|---|
| Cost: $2250 \| CPT Code: 81455 | | |
| Disease | Inheritance Pattern | Gene(s) |
| 17α-hydroxylase/17,20-lyase deficiency* | AR | *CYP17A1** |
| Adrenal hyperplasia, congenital (11b-OH deficiency) | AR | *CYP11B1* |
| Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency* | AR | *CYP21AP, CYP21A2** |
| Alagille syndrome | AD | *JAG1, NOTCH2* |
| Alport syndrome | AD, AR | *COL4A3, COL4A4* |
| | XLD | *COL4A5* |
| | XLR | *COL4A6* |
| Alstrom syndrome | AR | *ALMS1* |
| Amyloidosis, hereditary | AD | *APOA1, B2M, FGA, GSN, LYZ, NLRP3, TNFRSF1A, TTR* |
| Apparent mineralocorticoid excess | AR | *HSD11B2* |
| APRT deficiency (stones and ESRD) | AR | *APRT* |
| Arthrogryposis, renal dysfunction | AR | *VIPAS39, VPS33B* |
| Autosomal dominant tubulointerstitial disease | AD | *HNF1B, REN, UMOD* |
| Axenfeld-Rieger Syndrome* | | *FOXC1** |
| Bardet-Biedl syndrome (BBS) | AR | *ARL6, BBIP1, BBS2, BBS4, BBS5, BBS7, BBS10, BBS12, C8orf37, CEP290, IFT27, IFT74, LZTFL1, MKKS, PTHB1, SDCCAG8, TRIM32, TTC8, WDPCP* |
| Bardet-Biedl syndrome (BBS) | AR, DR | *BBS1* |
| Bartter syndrome | AD | *CaSR* |
| | AR | *BSND, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1* |
| Bilateral renal agenesis* | AR | *GFRA1** |
| Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT* | AR | *CHRNA3** |
| Branchiooculofacial syndrome | AD | *TFAP2A* |
| Branchio-oto-renal syndrome | AD | *EYA1, SIX1, SIX5* |
| CAKUT with VACTERL | AR | *TRAP1* |
| Charcot-Marie Tooth disease (CMTDIE) | AD | *INF2* |
| CHARGE syndrome | AD | *CHD7, SEMA3E* |
| CHARGE syndrome | AD | *CHD7, SEMA3E* |
| COACH syndrome (JN+) | AR | *CC2D2A, RPGRIP1L, TMEM67* |
| Cogan oculomotor apraxia | AR | *NPHP1, NPHP4* |
| Common CAKUT | AD | *ACTG1, AGTR1, CHD1L, DLG1, DLG5*, DSTYK, ETV4, EYA1, FOXP1, GATA3, GDNF, GFRA1*, GREB1L, HNF1B, KAT6B, KIF12, KMT2D, NRIP1, PAX2, PBX1, RET, ROBO2, SALL1, SIX2, SIX5, SLIT2, SRGAP1, TBX18, ZMYM2** |
| | AR | *CTU2, FAT4, HPSE2, TRAP1, TRPS1* |
| Congenial lipoid adrenal hyperplasia (lipoid CAH) * | AR | *STAR** |
| Congenital disorder of glycosylation, type 1h* | AR | *ALG8** |
| Congenital lung disease, nephrotic syndrome, and mild epidermolysis bullosa | AR | *ITGA3* |
| Congenital nephrotic syndrome | AR | *LAMB2, MAGI2, NPHS1, NPHS2, PLCE1, WT1* |
| COQ2 nephropathy | AR | *COQ2* |
| Cystinosis | AR | *CTNS* |
| Cystinuria | AD, AR | *SLC3A1, SLC34A1, SLC7A9* |
| Dent disease | XLR | *CLCN5, OCRL* |
| Denys-Drash syndrome; Frasier syndrome | AD | *WT1* |
| Diffuse mesangial sclerosis | AR | *ARHGDIA, PLCE1, WT1* |
| Donnai-Barrow syndrome* | AR | *LRP2** |
| Duane-radial ray syndrome (Okihiro syndrome) | AD | *SALL4* |

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| Epstein/Fechtner - renal disease with macrothrombocytopenia | AD | *MYH9* |
| Fabry disease | XL | *GLA* |
| Familial hypocalciuric hypercalcemia | AD | *CaSR* |
| Familial juvenile hyperuricemic nephropathy | AD | *SEC61A1, HNF1B* |
| Familial lecithin cholesterol acyltransferase (LCAT) deficiency | AR | *LCAT* |
| Familial Mediterranean Fever | AD, AR | *MEFV* |
| Fanconi syndrome, generalized proximal defect | AD | *EHHADH, HNF4A* |
| | AR | *ATP7B, CTNS, FAH, SLC34A1* |
| | XLR | *CLCN5* |
| Fanconi-Bickel syndrome | AR | *SLC2A2* |
| Finlay-Marks syndrome | AD | *KCTD1* |
| Focal segmental glomerulosclerosis (AR)/SRNS | AR | *ADCK4, ALG1, ANKFY1, ARHGDIA, CUBN, DGKE, DHTKD1, DLC1, EMP2, FAT1, GAPVD1, LAMB2, NPHS1, NPHS2, NUP107, NUP205, NUP93, PLCE1, PDSS2, PMM2, PTPRO, SCARB2, SGPL1, XPO5, ZMPSTE24* |
| Focal segmental glomerulosclerosis (FSGS) AD/XL | AD | *ACTN4, ANLN, ARHGAP24, CD2AP, CFI, COL4A3, COL4A4, E2F3, INF2, LMX1B, PAX2, TRPC6, WT1* |
| | XL | *COL4A5* |
| Focal segmental glomerulosclerosis (FSGS) AR | AR | *APOL1, COQ6, COQ8B, CRB2, ITGB4, LAMA5, MYO1E, NPHP4, NUP133, NUP160, NUP85, TTC21B* |
| Focal segmental glomerulosclerosis (FSGS) with Duane retraction syndrome | AD | *MAFB* |
| Fraser syndrome | AR | *FRAS1, FREM1, FREM2, GREM1, GRIP1* |
| Galloway-Mowat syndrome | AR | *GON7*, NUP133, OSGEP, TP53RK, TPRKB, WDR73, YRDC** |
| | XLR | *LAGE3* |
| Genitopatellar syndrome | AD | *KAT6B* |
| Gillessen-Kaesbach-Nishimura syndrome* | AR | *ALG9** |
| Gitelman syndrome | AR | *CLCNKB, SLC12A3* |
| Glomerulopathy with fibronectin deposits | AD | *FN1* |
| Glucosuria, renal | AD, AR | *SLC5A1, SLC5A2* |
| HANAC syndrome | AD | *COL4A1* |
| Hyperaldosteronism, familial | AD | *CACNA1D, CACNA1H, CLCN2, CYP11B1, KCNJ5* |
| Hyperaldosteronism, glucocorticoid remediable | AD | *CYP11B1-CYP11B2 fusion* |
| Hypercalcemia, infantile | AR | *CYP24A1* |
| Hypercalciuria | AD | *ADCY10* |
| Hyperoxaluria, primary | AR | *AGXT, GRHPR, HOGA1* |
| Hyperparathyroidism, neonatal | AR | CASR |
| Hypertension with hyperkalemia (Gordon's syndrome), Pseudohypoaldosteronism II | AD | *CUL3, KLHL3, WNK1, WNK4* |
| | AR | *KLHL3* |
| Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome (HUPRAS) | AR | *SARS2* |
| Hypoaldosteronism, congenital | AR | *CYP11B2* |
| Hypocalcemia, autosomal dominant | AD | *CASR* |
| Hypocalciuric hypercalcemia | AD | CASR |
| Hypogonadotropic hypogonadism with or without anosmia (Kallmann syndrome) | AD | *CHD7, FGFR1* |
| | XL | *ANOS1* |
| Hypokalemic- salt-losing nephropathy | AR | *CLDN10, KCNJ10* |
| Hypokalemic-alkalotic salt-losing nephropathy | | *CLDN10* |
| Hypomagnesemia with hypercalciuria | AR | *CLDN16, CLDN19* |

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| Hypomagnesemia, renal | AD | *CNNM2, FXYD2* |
| | AR | *CLDN16, CLDN19, EGF, HNF1B, KCNJ10, TRPM6* |
| Hypoparathyroidism, sensorineural deafness, and renal dysplasia | AD | *GATA3* |
| Hypophosphatasia, adult* | AD, AR | *ALPL** |
| Hypophosphatasia, child* | AR | *ALPL** |
| Hypophosphatasia, infantile* | AR | *ALPL** |
| Hypophosphatemic rickets | AD | *FGF23* |
| | AR | *DMP1, ENPP1, SLC34A3, VDR* |
| | XLR | *CLCN5, PHEX* |
| | XLD | *PHEX* |
| Hypouricemia, renal | AD | *SLC2A9* |
| | AR | *SLC22A12* |
| Infantile sialic acid storage disease | AR | *SLC17A5* |
| Interstitial nephritis, karyomegalic | AR | *FAN1* |
| Isolated renal hypo-dysplasia | AD | *BMP4, DSTYK, HNF1B, PAX2, RET, SALL1, SIX2* |
| | AR | *DACH1, FGF20, ITGA8* |
| Isolated renal hypoplasia: renal adysplasia | AR | *RET, UPK3* |
| Jeune syndrome (JN+) | AR | *DYNC2H1, IFT80, IFT121*, IFT122, IFT139*, IFT140, IFT144*, IFT172, NEK1, TTC21B, WDR19* |
| Joubert syndrome (JN+) | AR | *AHI1, ARL13B, ARMC9, ATXN10, B9D1, B9D2, C2CD3, C5orf42, CC2D2A, CEP104, CEP120*, CEP290, CEP41, CFAP410, CSPP1, IFT172, INPP5E, KATNIP, KIF7, MKS1, NPHP1, OFD1, PDE6D*, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231*, TMEM237, TMEM67, TTC21B* |
| Joubert syndrome (JN+) | AD, AR | *ZNF423* |
| Juvenile nephronophthisis (JN) | AR | *AHI1, ANKS6, ATXN10, IQCB1, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, MAGI2, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3* |
| Juvenile nephronophthisis (JN) | AD, AR | *ZNF423* |
| Lesch-Nyhan syndrome | XLR | *HPRT1* |
| Liddle syndrome (AD) (pseudoprimary hyperaldosteronism) | AD | *SCNN1B, SCNN1G* |
| Lipodystrophy, familial, partial | AD | *LMNA* |
| Lipoprotein glomerulopathy | | *APOE* |
| Lower urinary tract obstruction, congenital* | AD | *BNC2** |
| Mayer-Rokitansky-Küster-Hauser syndrome | AD | *WNT4* |
| Meckel syndrome (MKS)/Meckel-Gruber syndrome (JN+) | AR | *B9D1, B9D2, CC2DA, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM107, TMEM216, TMEM237, TMEM67* |
| Medullary cystic kidney disease 2 | AD | *UMOD* |
| Mitochondrial cytopathies | AR | *COQ2* |
| Muckle-Wells syndrome | AD | *NLRP3* |
| Multicystic dysplastic kidney | AD | *CHD1L, DLG5*, HNF1B, ROBO2, SALL1* |
| Nail patella syndrome | AD | *LMX1B* |
| Nephrogenic diabetes insipidus | AD, AR | *AQP2* |
| | XLR | *AVPR2* |
| Nephrogenic syndrome of inappropriate antidiuresis | XLR | *AVPR2* |
| Nephrolithiasis/osteoporosis, hypophosphatemic | AD | *SLC9A3R1* |

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| | AD | *SLC9A3R1* |
| Nephropathy with pretibial epidermolysis bullosa and deafness | AR | *CD151* |
| Nephrotic syndrome – steroid sensitive | | *CDK20*, DLG5*, PLCG2* |
| Nephrotic syndrome* | AR | *COQ8B*, ITSN1*, ITSN2*, KIRREL1** |
| Neurohypophyseal diabetes insipidus* | AR | *AVP** |
| Orofaciodigital syndrome 1 | XLD | *OFD1* |
| Pallister-Hall syndrome | AD | *GLI3* |
| Pierson syndrome – nephrotic syndrome with microcoria | AR | *LAMB2* |
| Polycystic kidney disease, autosomal dominant (ADPKD) | AD | *GANAB, HNF1B, NOTCH2, OFD1, PKD1, PKD2* |
| Polycystic kidney disease, autosomal recessive (ARPKD) | AR | *DZIP1L, PKHD1* |
| Polycystic liver disease 3 with or without kidney cysts* | AD | *ALG8*, LRP5*, PRKCSH*, SEC63** |
| Posterior urethral valves | AD | *CHD1L, HNF1B, ROBO2, SALL1, SIX2* |
| Primary Macronodular Adrenal Hyperplasia* | | *GNAS** |
| Pseudohypoaldosteronism I | AD | *NR3C2* |
| | AR | *SCNN1A, SCNN1B, SCNN1G* |
| Renal cysts and diabetes syndrome | AD | *HNF1B* |
| Renal fibrosis | AD | *PARN* |
| Renal tubular acidosis, distal | AD, AR | *ATP6V0A4, ATP6V1B1, ATP6V1C2*, FOXI1, SLC4A1, WDR72** |
| Renal tubular acidosis, proximal | AD | *EHHADH, HNF4A* |
| | AR | *ATP7B, CTNS, FAH, SLC34A1, SLC4A4* |
| | XLR | *ATP7B, CLCN5* |
| Renal tubular acidosis, type IV | AD | *CUL3, KLHL3, NR3C2, WNK1, WNK4* |
| | AR | *SCNN1A, SCNN1B, SCNN1G* |
| Renal tubular acidosis, with osteopetrosis | AR | *CA2* |
| Renal tubular disease, hypertension related | | *NEDD4L* |
| Renal tubular dysgenesis | AR | *ACE, AGT, AGTR1, REN* |
| Renal-Coloboma syndrome | AR | *PAX2* |
| Rubinstein-Taybi syndrome | AD | *CREBBP* |
| Schimke immuno-osseus dysplasia | AR | *SMARCAL1* |
| Senior-Loken syndrome- (JN with retinitis pigmentosa) | AR | *CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, WDR19* |
| Sensenbrenner syndrome/Cranioectodermal dysplasia (CED) | AR | *IFT122, IFT43, WDR19, WDR35* |
| SERKAL syndrome – 46XX sex reversal with dysgenesis of kidneys, adrenal and lungs | AR | *WNT4* |
| Serpentine fibula with polycystic kidney disease (SFPKS)/ Hajdu-Cheney syndrome (HJCYS) | AD | *NOTCH2* |
| SESAME syndrome / East syndrome | AR | *KCNJ10* |
| Short-rib thoracic dysplasia with or without polydactyly* | AR | *CEP120*, KIAA0586** |
| Simpson-Golabi-Behmel syndrome | XLR | *GPC3* |
| Smith-Lemli-Optiz syndrome | AR | *DHCR7* |
| Thin basement membrane disease (benign familial hematuria) | AD | *COL4A3, COL4A4* |
| Townes-Brocks syndrome | AD | *SALL1* |
| Tuberous sclerosis | AD | *TSC1, TSC2* |
| Unilateral renal agenesis | AD | *DSTYK, HNF1B, RET, SALL1* |
| UPJ obstruction | AD | *DSTYK, EYA1, HNF1B, RET, ROBO2, SALL1* |
| UVJ obstruction | AD | *CHD1L, DLG5*, PAX2, SIX5* |
| Vesicoureteral reflux | AD | *DSTYK, EYA1, GATA3, HNF1B, RET, ROBO2, SALL1, SOX17, TNXB, UPK3A, COL4A1* |

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| von Hippel-Landau syndrome – familial cancer syndrome | AD | *VHL* |
| Williams-Beuren syndrome | AD | 7q 11.23 |
| Xanthine oxidase deficiency | AR | *XDH* |

*New to KidneySeq™