

KidneySeq™ Comprehensive

Cost: \$2250 CPT Code: 81455

Disease	Inheritance Pattern	Gene(s)
17 α -hydroxylase/17,20-lyase deficiency*	AR	CYP17A1*
Adrenal hyperplasia, congenital (11b-OH deficiency)	AR	CYP11B1
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency*	AR	CYP21A1P, 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
Arthrogyrosis, 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
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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
Congenital 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	ARHGDI1, PLCE1, WT1
Donnai-Barrow syndrome*	AR	LRP2*
Duane-radial ray syndrome (Okhiro syndrome)	AD	SALL4

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

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Hypomagnesemia, renal	AD	<i>CNNM2, FXRD2</i>
	AR	<i>CLDN16, CLDN19, EGF, HNF1B, KCNJ10, TRPM6</i>
Hypoparathyroidism, sensorineural deafness, and renal dysplasia	AD	<i>GATA3</i>
Hypophosphatasia, adult*	AD, AR	<i>ALPL*</i>
Hypophosphatasia, child*	AR	<i>ALPL*</i>
Hypophosphatasia, infantile*	AR	<i>ALPL*</i>
Hypophosphatemic rickets	AD	<i>FGF23</i>
	AR	<i>DMP1, ENPP1, SLC34A3, VDR</i>
	XLR	<i>CLCN5, PHEX</i>
	XLD	<i>PHEX</i>
Hypouricemia, renal	AD	<i>SLC2A9</i>
	AR	<i>SLC22A12</i>
Infantile sialic acid storage disease	AR	<i>SLC17A5</i>
Interstitial nephritis, karyomegalic	AR	<i>FAN1</i>
Isolated renal hypo-dysplasia	AD	<i>BMP4, DSTYK, HNF1B, PAX2, RET, SALL1, SIX2</i>
	AR	<i>DACH1, FGF20, ITGA8</i>
Isolated renal hypoplasia: renal adysplasia	AR	<i>RET, UPK3</i>
Jeune syndrome (JN+)	AR	<i>DYNC2H1, IFT80, IFT121*, IFT122, IFT139*, IFT140, IFT144*, IFT172, NEK1, TTC21B, WDR19</i>
Joubert syndrome (JN+)	AR	<i>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</i>
Joubert syndrome (JN+)	AD, AR	<i>ZNF423</i>
Juvenile nephronophthisis (JN)	AR	<i>AHI1, ANKS6, ATXN10, IQCB1, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, MAGI2, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3</i>
Juvenile nephronophthisis (JN)	AD, AR	<i>ZNF423</i>
Lesch-Nyhan syndrome	XLR	<i>HPRT1</i>
Liddle syndrome (AD) (pseudoprimary hyperaldosteronism)	AD	<i>SCNN1B, SCNN1G</i>
Lipodystrophy, familial, partial	AD	<i>LMNA</i>
Lipoprotein glomerulopathy		<i>APOE</i>
Lower urinary tract obstruction, congenital*	AD	<i>BNC2*</i>
Mayer-Rokitansky-Küster-Hauser syndrome	AD	<i>WNT4</i>
Meckel syndrome (MKS)/Meckel-Gruber syndrome (JN+)	AR	<i>B9D1, B9D2, CC2DA, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM107, TMEM216, TMEM237, TMEM67</i>
Medullary cystic kidney disease 2	AD	<i>UMOD</i>
Mitochondrial cytopathies	AR	<i>COQ2</i>
Muckle-Wells syndrome	AD	<i>NLRP3</i>
Multicystic dysplastic kidney	AD	<i>CHD1L, DLG5*, HNF1B, ROBO2, SALL1</i>
Nail patella syndrome	AD	<i>LMX1B</i>
Nephrogenic diabetes insipidus	AD, AR	<i>AQP2</i>
	XLR	<i>AVPR2</i>
Nephrogenic syndrome of inappropriate antidiuresis	XLR	<i>AVPR2</i>
Nephrolithiasis/osteoporosis, hypophosphatemic	AD	<i>SLC9A3R1</i>

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	AD	<i>SLC9A3R1</i>
Nephropathy with pretibial epidermolysis bullosa and deafness	AR	<i>CD151</i>
Nephrotic syndrome – steroid sensitive		<i>CDK20*</i> , <i>DLG5*</i> , <i>PLCG2</i>
Nephrotic syndrome*	AR	<i>COQ8B*</i> , <i>ITSN1*</i> , <i>ITSN2*</i> , <i>KIRREL1*</i>
Neurohypophyseal diabetes insipidus*	AR	<i>AVP*</i>
Orofaciodigital syndrome 1	XLD	<i>OFD1</i>
Pallister-Hall syndrome	AD	<i>GLI3</i>
Pierson syndrome – nephrotic syndrome with microcoria	AR	<i>LAMB2</i>
Polycystic kidney disease, autosomal dominant (ADPKD)	AD	<i>GANAB</i> , <i>HNF1B</i> , <i>IFT140</i> , <i>NOTCH2</i> , <i>OFD1</i> , <i>PKD1</i> , <i>PKD2</i>
Polycystic kidney disease, autosomal recessive (ARPKD)	AR	<i>DZIP1L</i> , <i>PKHD1</i>
Polycystic liver disease 3 with or without kidney cysts*	AD	<i>ALG8*</i> , <i>LRP5*</i> , <i>PRKCSH*</i> , <i>SEC63*</i>
Posterior urethral valves	AD	<i>CHD1L</i> , <i>HNF1B</i> , <i>ROBO2</i> , <i>SALL1</i> , <i>SIX2</i>
Primary Macronodular Adrenal Hyperplasia*		<i>GNAS*</i>
Pseudohypoaldosteronism I	AD	<i>NR3C2</i>
	AR	<i>SCNN1A</i> , <i>SCNN1B</i> , <i>SCNN1G</i>
Renal cysts and diabetes syndrome	AD	<i>HNF1B</i>
Renal fibrosis	AD	<i>PARN</i>
Renal tubular acidosis, distal	AD, AR	<i>ATP6V0A4</i> , <i>ATP6V1B1</i> , <i>ATP6V1C2*</i> , <i>FOXI1</i> , <i>SLC4A1</i> , <i>WDR72*</i>
Renal tubular acidosis, proximal	AD	<i>EHHADH</i> , <i>HNF4A</i>
	AR	<i>ATP7B</i> , <i>CTNS</i> , <i>FAH</i> , <i>SLC34A1</i> , <i>SLC4A4</i>
	XLR	<i>ATP7B</i> , <i>CLCN5</i>
Renal tubular acidosis, type IV	AD	<i>CUL3</i> , <i>KLHL3</i> , <i>NR3C2</i> , <i>WNK1</i> , <i>WNK4</i>
	AR	<i>SCNN1A</i> , <i>SCNN1B</i> , <i>SCNN1G</i>
Renal tubular acidosis, with osteopetrosis	AR	<i>CA2</i>
Renal tubular disease, hypertension related		<i>NEDD4L</i>
Renal tubular dysgenesis	AR	<i>ACE</i> , <i>AGT</i> , <i>AGTR1</i> , <i>REN</i>
Renal-Coloboma syndrome	AR	<i>PAX2</i>
Rubinstein-Taybi syndrome	AD	<i>CREBBP</i>
Schimke immuno-osseus dysplasia	AR	<i>SMARCAL1</i>
Senior-Loken syndrome- (JN with retinitis pigmentosa)	AR	<i>CEP290</i> , <i>INVS</i> , <i>IQCB1</i> , <i>NPHP1</i> , <i>NPHP3</i> , <i>NPHP4</i> , <i>SDCCAG8</i> , <i>WDR19</i>
Sensenbrenner syndrome/Cranioectodermal dysplasia (CED)	AR	<i>IFT122</i> , <i>IFT43</i> , <i>WDR19</i> , <i>WDR35</i>
SERKAL syndrome – 46XX sex reversal with dysgenesis of kidneys, adrenal and lungs	AR	<i>WNT4</i>
Serpentine fibula with polycystic kidney disease (SFPKS)/ Hajdu-Cheney syndrome (HJCYS)	AD	<i>NOTCH2</i>
SESAME syndrome / East syndrome	AR	<i>KCNJ10</i>
Short-rib thoracic dysplasia with or without polydactyly*	AR	<i>CEP120*</i> , <i>KIAA0586*</i>
Simpson-Golabi-Behmel syndrome	XLR	<i>GPC3</i>
Smith-Lemli-Optiz syndrome	AR	<i>DHCR7</i>
Thin basement membrane disease (benign familial hematuria)	AD	<i>COL4A3</i> , <i>COL4A4</i>
Townes-Brocks syndrome	AD	<i>SALL1</i>
Tuberous sclerosis	AD	<i>TSC1</i> , <i>TSC2</i>
Unilateral renal agenesis	AD	<i>DSTYK</i> , <i>HNF1B</i> , <i>RET</i> , <i>SALL1</i>
UPJ obstruction	AD	<i>DSTYK</i> , <i>EYA1</i> , <i>HNF1B</i> , <i>RET</i> , <i>ROBO2</i> , <i>SALL1</i>
UVJ obstruction	AD	<i>CHD1L</i> , <i>DLG5*</i> , <i>PAX2</i> , <i>SIX5</i>
Vesicoureteral reflux	AD	<i>DSTYK</i> , <i>EYA1</i> , <i>GATA3</i> , <i>HNF1B</i> , <i>RET</i> , <i>ROBO2</i> , <i>SALL1</i> , <i>SOX17</i> , <i>TNXB</i> , <i>UPK3A</i> , <i>COL4A1</i>

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Disease	Inheritance Pattern	Gene(s)
von Hippel-Landau syndrome – familial cancer syndrome	AD	<i>VHL</i>
Williams-Beuren syndrome	AD	7q 11.23
Xanthine oxidase deficiency	AR	<i>XDH</i>

*New to KidneySeq™