

**KidneySeq™ Ciliopathies/Tubulointerstitial Diseases**

Cost: \$2000 | CPT Code: 81455

Disease	Inheritance Pattern	Gene(s)
Autosomal dominant tubulointerstitial disease	AD	<i>HNF1B, REN, UMOD</i>
Bardet-Biedl syndrome (BBS)	AR	<i>ARL6, BBIP1, BBS2, BBS4, BBS5, BBS7, BBS10, BBS12, C8orf37, CEP290, IFT27, IFT74, LZTFL1, MKKS, PTHB1, SDCCAG8, TRIM32, TTC8, WDPCP</i>
	AR, DR	<i>BBS1</i>
COACH syndrome (JN+)	AR	<i>CC2D2A, RPGRIP1L, TMEM67</i>
HANAC syndrome	AD	<i>COL4A1</i>
Familial juvenile hyperuricemic nephropathy	AD	<i>SEC61A1, HNF1B</i>
Hypokalemic-alkalotic salt-losing nephropathy		<i>CLDN10</i>
Interstitial nephritis, karyomegalic	AR	<i>FAN1</i>
Jeune syndrome (JN+)	AR	<i>DYNC2H1, IFT80, IFT121*, IFT122, IFT139*, IFT140, IFT144*, IFT172, NEK1, 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>
	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>
	AD, AR	<i>ZNF423</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>
Orofaciodigital syndrome 1	XLD	<i>OFD1</i>
Polycystic kidney disease, autosomal recessive (ARPKD)	AR	<i>DZIP1L, PKHD1</i>
Polycystic kidney disease, autosomal dominant (ADPKD)	AD	<i>GANAB, HNF1B, IFT140, NOTCH2, OFD1, PKD1, PKD2, TSC2,</i>
Polycystic liver disease 3 with or without kidney cysts*	AD	<i>ALG8*, LRP5*, PRKCSH*, SEC63*</i>
Renal cysts and diabetes syndrome	AD	<i>HNF1B</i>
Serpentine fibula with polycystic kidney disease (SFPKS)/ Hajdu-Cheney syndrome (HJCYS)	AD	<i>NOTCH2</i>
Sensenbrenner syndrome/Cranioectodermal dysplasia (CED)	AR	<i>IFT122, IFT43, WDR19, WDR35</i>
Senior-Loken syndrome- (JN with retinitis pigmentosa)	AR	<i>CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, WDR19</i>
Short-rib thoracic dysplasia with or without polydactyly*	AR	<i>CEP120*, KIAA0586*</i>

\*New to KidneySeq™