KidneySeq™ Ciliopathies/Tubulointerstitial Diseases		
Cost: $2000 | CPT Code: 81455		
Disease	**Inheritance Pattern**	**Gene(s)**
Autosomal dominant tubulointerstitial disease	AD	*DNAJB11, HNF1B, REN, UMOD*
Bardet-Biedl syndrome (BBS)	AR	*ARL6, BBIP1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, C8orf37, CEP290, IFT27, IFT74, LZTFL1, MKKS, SDCCAG8, TRIM32, TTC8, WDPCP*
	AR, DR	*BBS1*
COACH syndrome (JN+)	AR	*CC2D2A, RPGRIP1L, TMEM67*
Cogan oculomotor apraxia	AR	*NPHP1, NPHP4*
HANAC syndrome	AD	*COL4A1*
Familial juvenile hyperuricemic nephropathy	AD	*SEC61A1, HNF1B*
Hypokalemic-alkalotic salt-losing nephropathy		*CLDN10*
Interstitial nephritis, karyomegalic	AR	*FAN1*
Jeune syndrome (JN+)	AR	*DYNC2H1, IFT80, IFT121*, IFT122, IFT140, IFT172, NEK1, TTC21B, WDR19*
Joubert syndrome (JN+)	AR	*AHI1, ARL13B, ARMC9, ATXN10, B9D1, B9D2, C2CD3, CC2D2A, CEP104, CEP120*, CEP290, CEP41, CFAP410, CPLANE1, CSPP1, IFT172, INPP5E, KATNIP (KIAA0056), KIF7, MKS1, NPHP1, OFD1, PDE6D*, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231*, TMEM237, TMEM67, TTC21B*
	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*
	AD, AR	*ZNF423*
Meckel syndrome (MKS)/Meckel-Gruber syndrome (JN+)	AR	*B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM107, TMEM216, TMEM237, TMEM67*
Medullary cystic kidney disease 2	AD	*UMOD*
Nephronophthisis related ciliopathy		*ADAMTS9*
Orofaciodigital syndrome 1	XLD	*OFD1*
Polycystic kidney disease, autosomal recessive (ARPKD)	AR	*DZIP1L, PKHD1*
Polycystic kidney disease, autosomal dominant (ADPKD)	AD	*DNAJB11, GANAB, HNF1B, IFT140, NOTCH2, OFD1, PKD1, PKD2, TSC2,*
Polycystic liver disease 3 with or without kidney cysts*	AD	*ALG8*, LRP5*, PRKCSH*, SEC63**
Renal cysts and diabetes syndrome	AD	*HNF1B*
Serpentine fibula with polycystic kidney disease (SFPKS)/ Hajdu-Cheney syndrome (HJCYS)	AD	*NOTCH2*
Sensenbrenner syndrome/Cranioectodermal dysplasia (CED)	AR	*IFT122, IFT43, WDR19, WDR35*
Senior-Loken syndrome- (JN with retinitis pigmentosa)	AR	*CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, WDR19*
Short-rib thoracic dysplasia with or without polydactyly*	AR	*CEP120*, KIAA0586**

*New to KidneySeq™