

KidneySeq™ CAKUT

Cost: \$2000 | CPT Code: 81455

Disease	Inheritance Pattern	Gene(s)
Alagille syndrome	AD	<i>JAG1, NOTCH2</i>
Axenfeld-Rieger Syndrome*	AD	<i>FOXC1*</i>
Bilateral renal agenesis*	AR	<i>GFRA1*</i>
Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT*	AR	<i>CHRNA3*</i>
Branchiooculofacial syndrome	AD	<i>TFAP2A</i>
Branchio-oto-renal syndrome	AD	<i>EYA1, SIX1, SIX5</i>
CAKUT with VACTERL	AR	<i>TRAP1</i>
CHARGE syndrome	AD	<i>CHD7, SEMA3E</i>
Cogan oculomotor apraxia	AR	<i>NPHP1, NPHP4</i>
Common CAKUT	AD	<i>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*</i>
	AR	<i>CTU2, FAT4, HPSE2, TRAP1, TRPS1</i>
Congenital lipoid adrenal hyperplasia (lipoid CAH)*	AR	<i>STAR*</i>
Donnai-Barrow syndrome*	AR	<i>LRP2*</i>
Fraser syndrome	AR	<i>FRAS1, FREM1, FREM2, GREM1, GRIP1</i>
Genitopatellar syndrome	AD	<i>KAT6B</i>
Hypoparathyroidism, sensorineural deafness, and renal dysplasia	AD	<i>GATA3</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>
Hypogonadotropic hypogonadism with or without anosmia (Kallmann syndrome)	AD	<i>CHD7, FGFR1</i>
	XL	<i>ANOS1</i>
Lower urinary tract obstruction, congenital*	AD	<i>BNC2*</i>
Mayer-Rokitansky-Küster-Hauser syndrome	AD	<i>WNT4</i>
Multicystic dysplastic kidney	AD	<i>CHD1L, DLG5*, HNF1B, ROBO2, SALL1</i>
Primary Macronodular Adrenal Hyperplasia*		<i>GNAS*</i>
Posterior urethral valves	AD	<i>CHD1L, HNF1B, ROBO2, SALL1, SIX2</i>
Renal-Coloboma syndrome	AR	<i>PAX2</i>
Renal cysts and diabetes syndrome	AD	<i>HNF1B</i>
Renal fibrosis	AD	<i>PARN</i>
Renal tubular dysgenesis	AR	<i>ACE, AGT, AGTR1, REN</i>
Townes-Brocks syndrome	AD	<i>SALL1</i>
Unilateral renal agenesis	AD	<i>DSTYK, HNF1B, RET, SALL1</i>
UPJ obstruction	AD	<i>DSTYK, EYA1, HNF1B, RET, ROBO2, SALL1</i>
UVJ obstruction	AD	<i>CHD1L, DLG5*, PAX2, SIX5</i>
Vesicoureteral reflux	AD	<i>DSTYK, EYA1, GATA3, HNF1B, RET, ROBO2, SALL1, SOX17, TNXB, UPK3A, COL4A1</i>

\*New to KidneySeq™ Version 5