

SIGNIFICANCE STATEMENT

Congenital anomalies of the kidney and urinary tract (CAKUT) are the most common cause of CKD in the first three decades of life. Several lines of evidence support a monogenic disease hypothesis for CAKUT. This manuscript describes the utility of whole-exome sequencing for the identification of likely disease-causing mutations in a large pediatric cohort of 232 families with CAKUT. We find that, in 14% of families, a monogenic disease-causing CAKUT gene can be identified. Furthermore, WES provides the opportunity for identifying novel candidate genes for CAKUT, which will provide insights into the underlying pathogenesis of CAKUT.