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Autosomal Dominant Polycystic Kidney Disease in Children

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Case Study : Autosomal dominant polycystic kidney disease (ADPKD) is the most common monogenic disorder of end-stage kidney disease, with an estimated prevalence between 1:1,000 and 1:2,500. ADPKD, previously called adult polycystic kidney disease, is a dominant inherited disorder characterized by cystic dilatations in all parts of the nephron. Variants in one of two genes, PKD1 or PKD2, account for most cases of ADPKD. A variant in the PKD1 gene, which is located on chromosome 16 and encodes polycystin 1, is present in 78 percent of patients with ADPKD. Most of the remaining patients (approximately 15 percent) have a variant in the PKD2 gene, which encodes polycystin 2 and is located on chromosome 4. These genes encode proteins localized to the primary cilia of renal epithelial cell, which are involved with intracellular calcium signaling and activation of cyclic adenosine monophosphate. Although a third ADPKD gene locus remains unclear, mutations in SEC63, SEC61B, GANAB, PRKCSH, DNAJB11, ALG8, and ALG9 in the endoplasmic reticulum protein biosynthetic pathway have been associated with atypical forms of ADPKD. Most children with ADPKD are asymptomatic and are identified by ultrasound screening because of a positive family history or incidentally when ultrasound is performed for an unrelated clinical condition. However, approximately 2 to 5 percent of affected children will present with early onset of disease with similar renal findings as those of affected adults. In contrast, the extrarenal manifestations of ADPKD commonly seen in adults (eg, cysts in the liver and pancreas) are infrequently or rarely observed in pediatric patients. Although most affected children do not become symptomatic until adulthood, pediatric patients may have any of the following manifestations: hypertension, renal concentrating defect, proteinuria, gross hematuria, flank and abdominal pain, and rarely, impaired kidney function. Herein, we describe children diagnosed with ADPKD using ultrasonography and confirmed by genetic testing.