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Comprehensive Genetic Diagnosis of Pediatric Patients with Cystic Kidney Disease

Jeesu Min[1], Hyeonju Lee[1], Peong Gang Park[3], Jungmin Choi[5], Hayne Cho Park[4], Yo Han Ahn[1], Hee Gyung Kang[1], Ji Hyun Kim[2], Eujin Park[4]
[1]Department of Pediatrics, Seoul National University Hospital, Korea, Republic of
[2]Department of Pediatrics-Nephrology, Seoul National University Bundang Hospital, Korea, Republic of
[3]Department of Pediatrics-Nephrology, Ministry of Health and Welfare, Korea, Republic of
[4]Department of Pediatrics-Nephrology, Kangnam Sacred Heart Hospital, Korea, Republic of
[5]Department of Biomedical Sciences, Korea University College of Medicine, Korea, Republic of

Objectives: In children with chronic kidney disease (CKD), a monogenic cause accounts for almost 30%, of which inherited cystic kidney disease (iCKD) is the most common. Nearly 100 causes of renal cystic ciliopathies have been identified and the genetic diagnostic yield is reported to be approximately 50%. Here, we report results of comprehensive genetic testing in a cohort of Korean pediatric patients with iCKD.

Methods: From July 2019 to February 2021, children under the age of 18 with three or more cysts in both kidneys on imaging studies were recruited from three centers in Korea. Genetic analysis was performed using targeted exome sequencing (TES) including 89 genes known as cystogenesis-related or causative-ciliopathy.

Results: A total of 46 pediatric patients (28 boys, 60.9%) with iCKD were recruited. The median age was 9.2 years (IQR, 5.49-14.53). The clinical diagnoses of the patients were autosomal dominant polycystic kidney disease (ADPKD) in 10 patients, autosomal recessive polycystic kidney disease in 5, multicystic dysplastic kidney in 2, nephronophthisis in 1, and CKD of unknown etiology in the others. The mutation detection rate was 52.2% (24 of 46); *PKD1* was the most common causative gene (16 patients, 34.8%), followed by *HNF1B* (3 patients). *PAX2* (2 patients), *PKD2* (1 patient), *PKHD1* (1 patient), and *NPHP3* (1 patient). Twelve patients (27.9%) had a family history of iCKD, all of which pathogenic or likely pathogenic variants were identified. In patients without a family history, causative variants were found in 35.2% (12 of 34).

Conclusions: The causative variant detection rate in this cohort of Korean children with iCKD was 52.2% by TES. ADPKD was the most common diagnosis, but other diseases showing renal cysts were diagnosed through genetic analysis. Since genetic analysis yields a diverse genetic diagnosis in half of children with iCKD, molecular genetic testing is worthwhile in this population for better management.