

## 소아청소년의 간신 섬유낭종성 질환

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### Hepatorenal Fibrocystic Diseases in Children

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**Background:** Hepatorenal fibrocystic diseases (HRFCDs) are a group of monogenic disorders characterized by developmental abnormalities commonly involving the liver and kidney. HRFCDs belong to a larger group of disorders, so-called ciliopathies, and a number of diseases have been listed under the category of HRFCDs, including autosomal dominant and recessive polycystic kidney diseases (ADPKD and ARPKD), Joubert, Bardet-Biedl, Meckel-Gruber, oral-facial-digital syndromes, and so on. In this study, we performed genotype and phenotype analyses of childhood patients with HRFCD.

**Methods:** A total of 36 children with HRFCD were recruited, and patients with ADPKD were excluded. Genetic test, targeted exome sequencing or Sanger sequencing, was performed for 23 patients, and the diagnosis of ARPKD in the remaining 13 patients was solely based on their clinical features.

**Results:** Genetic test revealed pathogenic mutations in 16 of 23 patients. Including 13 patients with clinical diagnosis of ARPKD, ARPKD was the most common HRFCD (44.4%) followed by nephronophthisis 13 (NPHP13, 30.6%) and Meckel-Gruber syndrome type 3 (MKS3, 11.1%). Severe renal dysfunction was more common in non-ARPKD patients than in ARPKD patients, and the progression rate of the renal dysfunction was relatively faster in NPHP13 patients than in patients with ARPKD or MKS3. The main hepatic pathology was Caroli disease in the NPHP13 group, while most patients in other groups had Caroli syndrome or congenital hepatic fibrosis. Of note, 3 of 4 MKS3 patients had accompanying choledochal cysts. None of the ARPKD patients had other organ involvement than liver and kidney. About a half of the NPHP13 patients had ocular involvement, and only 1 patient had neurodevelopmental involvement. In contrast, all MKS3 patients had severe ocular and neurodevelopmental involvement.

**Conclusions:** NPHP13 is one of the major disease entities belong to HRFCDs, and thorough evaluation of the clinical features, including kidney, liver and other organ involvement, may provide clinical clues for the differential diagnosis in patients with HRFCD.

**Key Words:** 섬유낭종성 질환, 간신 섬유낭종성 질환

Ciliopathy, Hepatorenal fibrocystic diseases