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Ciliopathy and PKD in pediatrics

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Cilia are located in almost every cell type in the body and play an important role in various cellular functions during development, morphogenesis and homeostasis. Ciliary dysfunction affects many organs, including brain, eyes, ears, nose, respiratory tract, liver, pancreas and kidney.

Ciliopathy of the kidney results from uncontrolled epithelial cell proliferation, growth, and polarity due to dysregulation of ciliary-dependent signaling. The phenotype observed in renal ciliopathy include cystic kidney disease such as polycystic kidney disease, nephronophthisis, Joubert syndrome, Meckel-Gruber syndrome and Bardet-Biedl syndrome. To date, nearly 100 genes that cause renal cystic ciliopathy have been identified, and a genetic diagnostic yield is reported to be approximately 50%. Similar findings were observed in a cohort of Korean pediatric patients with inherited cystic kidney disease. In this cohort, the causative variant detection rate was 50% by targeted exome sequencing. Autosomal dominant polycystic kidney disease was the most common diagnosis, but other renal ciliopathies were also diagnosed through genetic analysis.

With the recent advance in genomic sequencing technology, diverse genetic diagnoses have been made for children with renal cystic ciliopathy, and through molecular genetic identification, the components involved in cilia-specific functions and the molecular mechanisms underlying various ciliopathies are increasingly being elucidated. Hopefully, we expect these findings to be used in the development of novel and effective therapies for renal cystic ciliopathy, which is still a big challenge in the pediatric population.