【Symposium 8-1】

Genotype and Phenotype in Atypical Polycystic Kidney Disease Genes

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Autosomal dominant polycystic kidney disease (ADPKD) is the most common inherited kidney disorder, affecting at least 1 in 1000 individuals, and is a significant cause of end-stage kidney disease. The Consortium for Radiologic Imaging Studies of Polycystic Kidney Disease demonstrated that total kidney volume increases by approximately 5% annually in adulthood and is a reliable predictor of chronic kidney disease progression. The Mayo Clinic Imaging Classification, based on age- and height-adjusted TKV measured via MRI, offers a validated method for CKD risk stratification, patient selection for clinical trials, and routine clinical use. However, it excludes patients with atypical imaging patterns (class 2), found in 8.8% of the Mayo Clinic derivation cohort, which are characterized by features such as unilateral, asymmetric, or segmental cyst distribution, or kidney atrophy.

In this presentation, we will delve into the atypical imaging patterns, genetic profiles, and clinical features of patients diagnosed with atypical PKD, with a particular focus on the Taiwanese population affected by ADPKD. Our discussion will begin with an overview of atypical imaging patterns, which include unique presentations such as unilateral, asymmetric, or segmental cyst distribution, as well as cases accompanied by kidney atrophy. These patterns distinguish atypical PKD from the more common diffuse, bilateral cystic kidney disease and pose challenges for diagnosis and classification. Next, we will explore the genetic landscape of these patients, emphasizing the lower prevalence of detectable PKD1 and PKD2 mutations compared to typical ADPKD cases. We will discuss how this genetic heterogeneity impacts disease inheritance, progression, and therapeutic strategies. Additionally, we will examine the clinical features that set this group apart, such as older age at diagnosis, reduced family history, and a notably lower likelihood of progression to chronic kidney disease (CKD) stages 3–5. By analyzing these characteristics, we aim to clarify how atypical PKD differs in its disease trajectory and prognosis. Through this targeted analysis of atypical APKD within the Taiwanese context, we aim to provide insights into the unique needs of this distinct subgroup, enhancing diagnostic accuracy and paving the way for tailored clinical management and research approaches.