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Session Topic : Genetic Disease

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Genetic Etiology in Adult CKD in Japan

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Recent advances in genetics have significantly transformed our understanding of inherited kidney diseases and the way they are diagnosed. Our group has long been engaged in comprehensive genetic analysis of inherited kidney diseases in Japanese patients in the field of nephrology. We conducted comprehensive genetic analysis of Japanese adult patients who had initiated dialysis therapy due to CKD of unknown etiology. This study revealed that inherited kidney diseases accounted for more than 10% of these cases, and that most of these conditions had not been accurately diagnosed in routine clinical practice. The diseases identified through genetic analysis included conditions such as Alport syndrome and Fabry disease, in which early diagnosis and intervention before dialysis initiation may help prevent or delay progression to kidney failure. To our knowledge, this was the first comprehensive genetic analysis targeting CKD in Japan. Our study demonstrated the presence of undiagnosed inherited kidney diseases among Japanese adult CKD patients and highlighted the need to incorporate genetic testing into the diagnostic approach for selected CKD patients. In addition to this broader CKD-based approach, disease-specific investigation of inherited kidney diseases is also essential. We are also advancing research on the genetics, pathogenesis, and therapeutic development of specific disorders, including polycystic kidney disease and nephronophthisis, which I will introduce in this lecture. As research progresses, it has become increasingly clear that inherited kidney diseases contribute to CKD more frequently than previously expected, and that the causative genes are relatively limited. In this lecture, I will outline a practical clinical strategy for identifying hidden inherited kidney diseases among CKD patients and linking genetic diagnosis to appropriate treatment.

Keywords: Genetic Kidney Disease, CKD, Genetic Diagnosis, Chronic Kidney Disease of Unknown Etiology