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Session Name : Genetic Disease

Session Topic : Genetic Disease

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PKD Cohort Study in Korea: Clinical and Genetic Characteristics

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Autosomal dominant polycystic kidney disease (ADPKD) the most common inherited cystic kidney disease that affects 1 in every 4,000~10,000 people. From May 2019 to June 2024, we recruited a total of 1,628 probands in the Korean genetic cohort study of inherited cystic kidney diseases. Among them, 1,495 patients were clinically ADPKD. The study results showed a mutation detection rate of 57.5% in the Phase 1 cohort of 725 patients, 62.3% in typical ADPKD patients, and 41.8% in atypical cases. Of the 100 patients who had no mutations identified in the gene panel, additional PKD1 mutations were detected by long-range PCR in 79, increasing the overall detection rate to 62%. WES was performed on 125 patients, including those who had no mutations or VUS detected in the initial test, and IFT140 was identified as a new candidate gene. In Phase 2, 770 patients were enrolled, of whom 761 underwent targeted gene panel testing. The mutation detection rate was 69.5%, and the additional long-range PCR or WES increased the detection rate to 80%. Among 1,186 adult patients with typical ADPKD, the mutation detection rate of ciliopathy gene panel was 73.7%, but it improved to 88.4% as we performed secondary genetic testing in a stepwise manner: first a ciliopathy gene panel as a screening test followed by a few other genetic testing methods as secondary options including targeted exome sequencing for PKD1 after long-range polymerase chain reaction, familial segregation study, multiplex ligation-dependent probe amplification, whole exome sequencing and/or minigene assay as needed. This study systematically elucidated the genetic characteristics of Korean ADPKD patients and significantly increased the diagnostic rate through stage-specific genetic testing.

Keywords: ADPKD, Genetic test