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Genetic Study Findings in the Korean ADPKD Registry

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Inherited cystic kidney disease is a constellation of heterogenous genetic diseases that commonly share renal cystic phenotype. Including well-known PKD1 or PKD2, there have been enormous discovery of novel genes that can cause renal cysts. However, renal outcome according to different genotypes have not been fully discovered. Therefore, we have analyzed renal outcome (annual change of estimated glomerular filtration rate and total kidney volume) among Korean genetic cohort of inherited cystic kidney disease. Primary genetic analysis was conducted using a targeted gene panel including 89 genes associated with ciliopathy. Family segregation study and targeted exome sequencing of PKD1 was additionally performed to finalize pathogenic variants. Whole exome sequencing was performed among the patients with no pathogenic variants or variants of unknown significance. Copy number variation (CNV) was also examined. Genotypes were classified into 6 groups as follows: PKD1, PKD2, minor genotypes, double variants, VUS, and no mutation detected. PKD1 genotype was subclassified into PKD1 protein truncating mutation (PKD1-PT), PKD1 non-truncating mutation (PKD1-NT), and PKD1 in-frame insertion/deletion (PKD1-IF). A total of 824 patients were included in the study from 2019 to 2021. PPKD1 genotype demonstrated poorer renal outcome compared to other genotypes. Minor genotypes showed favorable renal outcome while the patients with double variants demonstrated poorer renal outcome with shrinkage of kidneys. CNV analysis additionally found PKD2L2 CNV deletion as the pathogenic variants causing cystic kidney disease. Our study demonstrated comprehensive genetic analysis using gene panel, whole exome sequencing, and CNV analysis can effectively reveal pathogenic variants of cystic kidney disease.

Keywords: Kidney Diseases, Cystic, High-Throughput Nucleotide Sequencing, Exome Sequencing, Glomerular Filtration Rate