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Baseline Characteristics of the Korean Genetic Cohort of Inherited Cystic Kidney Disease

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Objectives: Identification of inherited cystic kidney disease genes in each case is necessary for precise treatment.

Methods: We performed a 3-year prospective, multicenter cohort study at eight hospitals from May 2019 to May 2022. Patients with more than three renal cysts were enrolled and classified into two categories: typical autosomal dominant polycystic kidney disease (ADPKD) and atypical polycystic kidney disease (PKD) (Figure 1). Clinical and genetic characteristics were evaluated.

Results: A total of 725 adult patients were enrolled. Mean age was 46.2 ± 14.0 years, and 48.6% were male. Patients were categorized into typical ADPKD (560, 77.2%) and atypical PKD (165, 22.8%). Typical ADPKD based on Mayo imaging classification (MIC) I were classified as follows: (1) 1A 55 (9.9%), (2) 1B 149 (26.9%), (3) 1C 198 (35.8%), (4) 1D 90 (16.3%), and (5) 1E 61 (11.0%). Atypical PKD by MIC II included bilateral cystic with bilateral atrophic (31, 37.3%), lopsided (27, 32.5%), unilateral (9, 10.8%), segmental (8, 9.6%), bilateral cystic with unilateral atrophic (7, 8.4%), and asymmetric (1, 1.2%). The mutation detection rate of a gene panel was 64.3% (466 out of 725). In 99 cases (13.7%), no variants were found while 85 (11.7%) were variants of unknown significance, and 75 (10.3%) cases were double variants (Figure 2).

Conclusions: We report the baseline characteristics and genetic findings for the first nationwide genetic cohort for Korean hereditary cystic kidney disease prior to detailed molecular analysis.

Figure 1. Study flowchart