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Prevalence of genetic kidney diseases in the Korean cohort study

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Objectives: Recent studies suggest that hereditary causes of chronic kidney disease (CKD) appears to be more frequent than expected, accounting for up to 25% of kidney failure patients. The present study aimed to identify the clinical and genetic features of Korean CKD patients.

Methods: This multicenter registry enrolled 1,054 patients who was genetically confirmed kidney diseases from 10 pediatric and adult nephrology centers. Clinical diagnoses was divided into 6 categories-glomerulopathy, tubulopathy, congenital anomalies of kidney and urinary tract (CAKUT), unknown origin of CKD and others. Genetic diagnoses were assessed by sanger sequencing or whole exome sequencing.

Results: Clinical diagnosis of 457 (43.3%) of the total 1,054 patients belonged to tubulopathies, 344 patients (32.6%) glomerulopathies, 151 patients (14.3%) CAKUT, 41 patients (3.9%) unknown origin of CKD and 30 patients (2.8%) others. *COL4A5* was the most commonly found causative gene of glomerulopathies, accounting for 44.8%, followed by *COL4A4* (23 patients, 6.7%) and *WT1* (19 patients, 5.5%). *PHEX* was the most common causative gene of tubulopathies found in 53 patients (11.6%), followed by *CLCN5* in 43 patients (9.4%), and *AVPR2* in 38 patients (8.3%).

Conclusions: This is the first nationwide registry on patients with genetic kidney diseases in Korea. It is expected that this study will be helpful in describing the clinical and genetic characteristics of Korean hereditary kidney disease.