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Effect of genotype on renal outcome among the patients with inherited cystic kidney disease

Hayne Cho Park[1], Dha Woon Im[2], Yong Chul Kim[3], Curie Ahn[4], Yaerim Kim[7], Hee Gyung Kang, Jungmin Choi, Yun Kyu Oh
[1]Department of Internal Medicine-Nephrology, Kangnam Sacred Heart Hospital, Korea, Republic of
[2]Department of Internal Medicine-Nephrology, Uijeongbu Eulji Medical Center, Eulji University, Korea, Republic of
[3]Department of Internal Medicine-Nephrology, Seoul National University Hospital, Korea, Republic of
[4]Department of Internal Medicine-Nephrology, National Medical Center, Korea, Republic of
[5]Department of Internal Medicine-Nephrology, Keimyung University Dongsan Medical Center, Korea, Republic of
[6]Department of Pediatrics-Nephrology, Seoul National University Hospital, Korea, Republic of
[7]Department of Biomedical Sciences, Korea University College of Medicine, Korea, Republic of
[8]Department of Internal Medicine-Nephrology, Seoul National University Boramae Medical Center, Korea, Republic of

Objectives : Inherited cystic kidney disease is a constellation of heterogenous genetic diseases that commonly share renal cystic phenotype. We have analyzed renal outcome (annual change of estimated glomerular filtration rate (eGFR) and total kidney volume (TKV)) among Korean genetic cohort of inherited cystic kidney disease.
Methods : Primary genetic analysis was conducted using a targeted gene panel including 89 ciliopathy-associated genes. A total of 705 patients were included in the cross-sectional analysis. A total of 375 patients with longitudinal data without history of Tolvaptan treatment were included in the renal outcome analysis. Genotypes were classified into PKD1, PKD2, minor genotypes, or double variants (DV).
Results : PKD1 group showed younger age at diagnosis of cystic kidney disease and hypertension, and higher proportion of rapid progressor. Interestingly, the patients with minor genotypes showed better renal function but smaller kidneys than PKD1 group. The DV group demonstrated similar profiles with PKD1 group. PKD1 genotype showed faster eGFR decline (-2.69± 9.2 mL/min/1.73m^2/yr) compared to PKD2 (-0.15 ± 8.1 mL/min/1.73m^2/yr) and minor genotypes (0.68 ± 9.5 mL/min/1.73m^2/yr, p=0.03). The DV group demonstrated faster eGFR decline rate than PKD1 genotype (-3.53 ± 7.4 mL/min/1.73m^2/yr). PKD1 group (59.2 ± 93.0 mL per year) and PKD2 group (47.5 ± 79.5 mL per year) demonstrated TKV growth during study period while minor genotypes showed shrinkage of kidneys (-8.6 ± 55.8 mL per year, p=0.052).
Conclusions : PKD1 genotype demonstrated poorer renal outcome compared to other genotypes. Minor genotypes showed favorable renal outcome with shrinkage of kidneys.

Table 1.png

Table 1. Annual change of estimated glomerular filtration rate according to genotype

	PKD1-PT (n=141)	PKD1-NT (n=78)	PKD1-IF (n=10)	PKD2 (n=67)	Other (n=33)	DV (n=46)	p-value
Annual change of eGFR (mL/min/1.73m^2/year)	-2.78 ± 9.7	-2.36 ± 8.57	-3.89 ± 5.9	-0.15 ±8.09	0.68 ± 9.53	-3.53 ± 7.43	0.04
Duration of follow-up (yr)	1.33 ± 0.47	1.24 ± 0.48	1.37 ± 0.56	1.37 ± 0.47	1.31 ± 0.53	1.23 ± 0.45	0.485

Table 1.png

Table 2. Annual change of total kidney volume according to genotype

	PKD1-PT (n=43)	PKD1-NT (n=19)	PKD1-IF (n=3)	PKD2 (n=19)	Other (n=10)	DV (n=8)	p-value
Annual change of TKV (mL/year)	63.0 ± 107.1	60.3 ± 55.8	-1.4 ± 49.0	47.5 ± 79.5	-8.6 ± 55.8	54.0 ± 130.2	0.056
Duration of follow-up (yr)	2.12 ± 0.44	1.96 ± 0.31	2.24 ± 0.12	1.98 ± 0.27	2.06 ± 0.38	2.0 ± 0.14	0.474