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Cardiac complications and genotype-phenotype correlations in patients with autosomal dominant polycystic kidney disease in the Korean Cohort Study for Outcomes in Patients with KNOW-CKD cohort: an analysis using next-generation sequencing data

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Objectives : As an extrarenal manifestation, the cardiovascular system is also affected by autosomal dominant polycystic kidney disease (ADPKD), and such complications are associated with mortality and morbidity. However, the discrete prevalence of cardiac complications and corresponding genotype-phenotype correlations are not well understood. The purpose of this study was to elucidate the prevalence of cardiovascular complications and to identify genotype-phenotype correlations in ADPKD subjects in the Korean Cohort Study for Outcomes in Patients with Chronic Kidney Disease (KNOW-CKD) using PKD1 and PKD2 gene information.

Methods : The subjects were enrolled in the KNOW-CKD study at Seoul National University Hospital. Baseline echocardiography was performed for both ADPKD and non-ADPKD CKD subjects. The prevalence of valvular heart disease, pericardial effusion, and aortic aneurysm was investigated and compared with other non-ADPKD subjects. Next, the correlations between cardiovascular complications and the PKD1 and PKD2 genes were analyzed.

Results : A total of 643 subjects (ADPKD vs. non-ADPKD, 227 vs. 416) were included in the analysis. The ADPKD group was younger (ADPKD vs. non-ADPKD, 46.1 ± 10.9 years vs. 55.5 ± 13.2 years, p<.001), had fewer males (ADPKD vs. non-ADPKD, 31.9% vs. 39.9%, p=.036) and had higher estimated glomerular filtration rates (eGFR) (ADPKD vs. non-ADPKD, 73.2 ± 28.8 mL/min/1.73 m2 vs. 50.2 ± 27.7 mL/min/1.73 m2, p<.001). Except for mitral regurgitation (MR), the prevalence of valvular heart disease adjusted for age, sex, and eGFR was not significantly different between the 2 groups (MR, 11.0%

vs. 9.4%, p=0.026; tricuspid regurgitation, 32.6% vs. 29.3%, p=.377; and aortic regurgitation, 6.6% vs. 8.9%, p=0.442 in the ADPKD and non-ADPKD groups, respectively), and no valvular stenosis was observed in the ADPKD group. Aortic dilatation or aneurysm (ADPKD vs. non-ADPKD, 2.6% vs. 5.3%, p=.002) and pericardial effusion (ADPKD vs. non-ADPKD, 7.0% vs. 2.9%; p<.001) were more common in ADPKD patients. Adjusting for age, sex, and eGFR, no significant associations were found among PKD1 protein truncating, PKD2 non-truncating, and the PKD-no mutation genotypes with valvular heart disease, pericardial effusion, or aortic aneurysm.

Conclusions : The prevalence of MR was higher in ADPKD patients, but lower than has been reported in other studies (25%-30%). Moreover, the prevalence of aortic aneurysm and pericardial effusion was higher in ADPKD patients than in non-ADPKD patients. The effect of the PKD1 and PKD2 genes on cardiovascular complications was not significant, meaning that an unknown third factor must affect the cardiac phenotype.

Keywords : Autosomal dominant polycystic kidney disease, cardiovascular complication, genetic variation, phenotype