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Autosomal Dominant Polycystic Kidney Patients May Be Predisposed to Various Cardiomyopathies

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Abstract : Background: Mutations in PKD1 and PKD2, the genes encoding the proteins polycystin-1 (PC1) and polycystin-2 (PC2) cause autosomal dominant polycystic kidney disease (ADPKD). ADPKD is a systemic disease associated with several extrarenal manifestations. Animal models have suggested an important role for the polycystins in cardiovascular function. The aim of the current study is to evaluate the association of various cardiomyopathies in a large cohort of patients with ADPKD. Methods: Clinical data was retrieved from medical records for all patients with ADPKD and cardiomyopathies (n=159). Genetic analysis was performed on available DNA by direct sequencing. Results: Among the 58 patients included in this case series, 39 patients had idiopathic dilated cardiomyopathy (IDCM), 17 had hypertrophic obstructive cardiomyopathy (HOCM), and 2 had left ventricular noncompaction (LVNC). The mean age at cardiomyopathy diagnosis was 53.3, 59.9 and 53.5 years in IDCM, HOCM and LVNC patients respectively. The median left ventricular ejection fraction at initial diagnosis of IDCM was 25%. Average basal septal thickness was 19.9 mm in patients with HOCM. Genetic data was available in 19, 8 and 2 cases of IDCM, HOCM, and LVNC respectively. PKD1 mutations were detected in 47.4%, 62.5% and 100% of IDCM, HOCM and LVNC cases. PKD2 mutations were detected only in IDCM cases and were overrepresented (36.8%) relative to the expected frequency in ADPKD (~15%). The prevalence of IDCM, HOCM, and LVNC in our ADPKD clinical cohort was 1:17, 1:39 and 1:333 respectively. When compared to the general population, IDCM and HOCM was approximately 10-fold more prevalent in patients with ADPKD. Conclusions: In summary, we suggest that PKD1 or PKD2 mutations may predispose to idiopathic dilated or hypertrophic cardiomyopathy. There is a trend for patients with PKD2 mutations to develop the former and for patients with PKD1 mutations to develop the latter. Predisposition to various cardiomyopathies may be another extrarenal manifestation of ADPKD.

Keywords : autosomal dominant polycystic kidney (ADPKD), polycystic kidney disease, cardiovascular, cardiomyopathy, idiopathic dilated cardiomyopathy, hypertrophic cardiomyopathy, left ventricular noncompaction
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