

First Evidence of Impaired Cardiac Geometry and Function in Children with Autosomic Recessive Polycystic Kidney Disease.

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Background: Autosomic recessive polycystic kidney disease (ARPKD) is a rare clinical condition associated with renal failure and neonatal death. In patients surviving the neonatal age, progressive renal dysfunction and liver fibrosis develop. No previous study has yet defined the prevalence of cardiac geometric and mechanical function abnormalities in children with ARPKD.

Methods: Standard echocardiograms were obtained in 22 children with ARPKD aged 0-18 yr (50% boys) and in 88 healthy children of similar age, gender distribution, and body build. Left ventricular (LV) mass was calculated by the Devereux formula and LV hypertrophy was defined according to our recently proposed criteria (J Ped 2016). Cardiac geometry was defined by age-adjusted relative wall thickness (RWT) while systolic function was assessed by both measurement of ejection fraction and fractional shortening at the midwall (MFS), representing cardiac mechanical function.

Results: Patients with ARPKD exhibited higher LV mass index as compared to normal individuals (40.4 ± 8.2 versus $28.9 \pm 4.3 \text{g}/(\text{m}^{2.16} + 0.09)$; $P < 0.001$), mainly due to a more concentric LV geometry ($\text{RWT} = 0.33 \pm 0.07$ versus 0.27 ± 0.03 ; $\% ; P < 0.001$). Accordingly, prevalence of LV hypertrophy was significantly higher in ARPKD (18.2 vs 0% ; $P < 0.005$). No differences could be observed among patients and controls in ejection fraction (66.2 ± 4.6 versus $68.4 \pm 5.9\%$; $P = \text{NS}$), while a significantly lower MFS (19.3 ± 3.6 versus $21.6 \pm 2.1\%$; $P < 0.05$) could be observed. In analysis of covariance, adjusting for differences in age, systolic blood pressure and gender, both LV mass index and RWT remained significantly higher in the ARPKD group, while MFS remained significantly reduced (all $p < 0.05$). The prevalence of subclinical systolic dysfunction (defined by $\text{MFS} < 16\%$), was significantly higher in patients with ARPKD as compared with control subjects (22.7 versus 0% ; $P < 0.001$), demonstrating significantly impaired cardiac function despite normal ejection fraction (figure).

Conclusions: This is the first study reporting on cardiac geometry and mechanical function in children with ARPKD. Children with ARPKD show significantly impaired cardiac phenotype, characterized by high rates of LV hypertrophy paired with significant systolic mechanical dysfunction, possibly contributing to the high cardiovascular risk of children affected by ARPKD.

Figure: filled circles (and bold line) represent normal controls; crosses (and dashed line) represent ARPKD patients (see text for explanations).

