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CASE IMAGE

Congenital abnormalities of heart and kidney

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Abstract

Patients with congenital anomalies of the kidney and urinary tract (CAKUT) may be at risk for congenital cardiac defects or cardiomyopathies as comorbidities. It is crucial to recognize the coexistence of cardiac abnormalities and CAKUT and recommend screening for cardiac involvement in CAKUT patients using echocardiography.

KEYWORDS

atrial septal defect, autosomal dominant polycystic kidney disease, hemodialysis, left ventricular hypertrophy

1 | CASE DESCRIPTION

Patients with congenital anomalies of the kidney and urinary tract (CAKUT) may be at risk for congenital cardiac defects or cardiomyopathies as comorbidities. It is crucial to recognize the coexistence of cardiac abnormalities and CAKUT and recommend screening for cardiac involvement in CAKUT patients using echocardiography.

A 53-year-old woman with an atrial septal defect (ASD) was referred to our hospital for defect closure. She was diagnosed with autosomal dominant polycystic kidney disease (ADPKD) (Figure 1) in her 20's. Mutation to PKD1 encoding polycystin-1 was detected.

She had no history of hypertension. Ten years prior to consult, hemodialysis was initiated due to worsening kidney function. Four years prior to consult, ASD was incidentally noted in routine examination for the first time. However, no intervention was done at that time because she was asymptomatic, and no signs of right heart overload were noted. In the interim, her symptoms (shortness of breath and general fatigue) gradually worsened.

Transthoracic echocardiography revealed left ventricular hypertrophy and left ventricular diastolic dysfunction; E/e′ 17.9, e′ (septal) 4.7 cm/s, TRV 2.0 m/s, LAVI 39 mL/m^2 (Figure 2) with left ventricular ejection fraction 67%, and right-sided dilatation with a pulmonary to systemic flow ratio of 1.6. Moreover, transesophageal echocardiography revealed a 11 × 5-mm single secundum ASD, with aortic rim deficiency and malalignment (Figure 3). A percutaneous ASD closure was performed. A 13-mm Amplatzer septal occluder (Abbott) was successfully deployed without procedural complications (Figure 4).

Congenital anomalies of the kidney and urinary tract (CAKUT) are a collection of abnormalities affecting the urinary and cardiovascular systems. These systems share a common embryologic origin, the mesoderm. Thus, an insult to the mesoderm during embryogenesis may cause defects in both organs. Patients with CAKUT may be at risk for congenital cardiac defects or cardiomyopathies as comorbidities.[1,2] The coexistence of cardiac abnormalities and CAKUT (here, ADPKD) is rarely diagnosed in adulthood. It is well known that common cardiovascular complications of ADPKD are valvular abnormalities (mitral valve prolapse, mitral regurgitation, aortic regurgitation, and tricuspid regurgitation), coronary aneurysm, and intracranial aneurysmal dilatation. This shows a case where a percutaneous ASD closure was an effective and less invasive strategy in an adult patient with concomitant

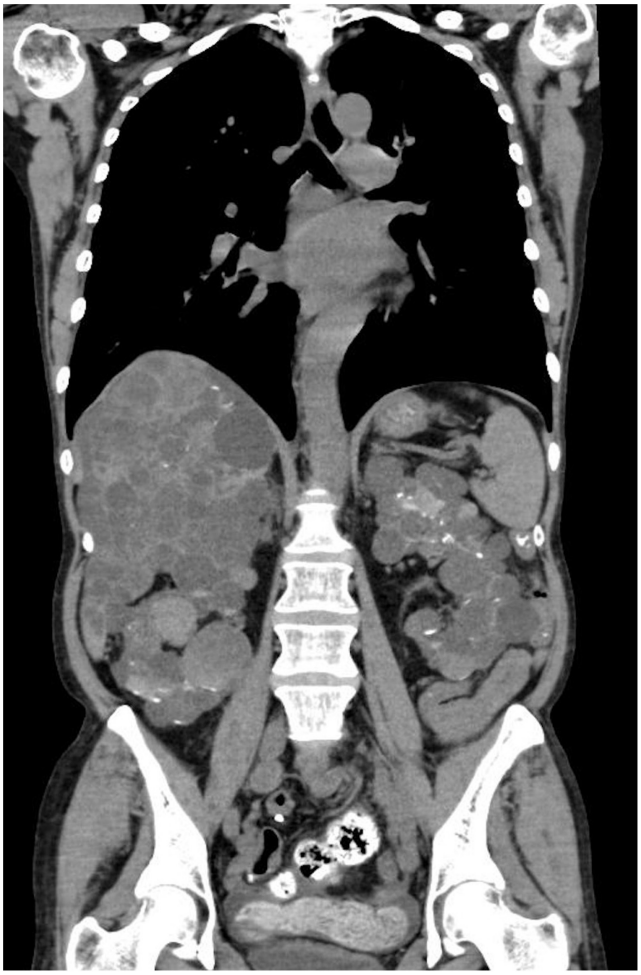

FIGURE 1 Computed tomography on admission reveals bilateral polycystic kidneys with liver involvement.

ADPKD after hemodialysis was initiated. The indications for ASD closure with left ventricular hypertrophy should be seriously considered because it may lead to heart failure after closure; however, this case could be well-volume controlled due to hemodialysis without pulmonary hypertension before closure. It is crucial to recognize the coexistence of cardiac abnormalities and CAKUT and recommend screening for cardiac involvement in CAKUT patients using echocardiography.

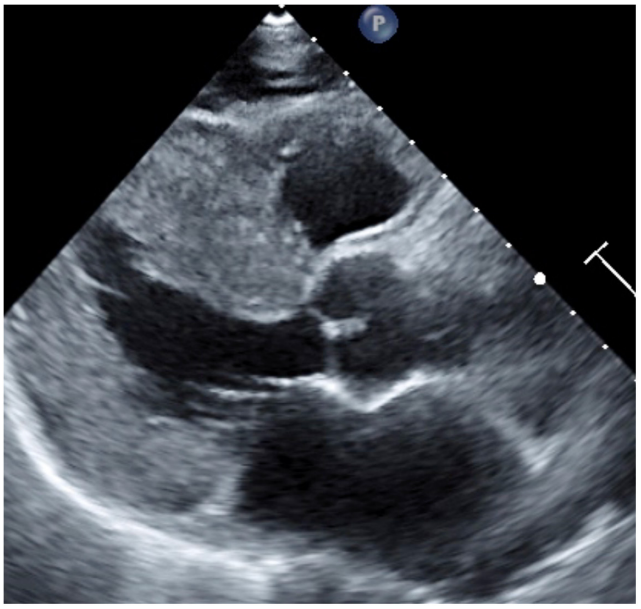

FIGURE 2 Transthoracic echocardiography reveals left ventricular hypertrophy.

AUTHOR CONTRIBUTIONS

Hiroya Takafuji: Writing – original draft. **Nahoko Kato:** Writing – review and editing. **Yuta Azumi:** Writing – review and editing. **Kotaro Obunai:** Writing – review and editing.

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CONFLICT OF INTEREST STATEMENT

All authors have reported that they have no relationships relevant to the contents of this paper to disclose.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

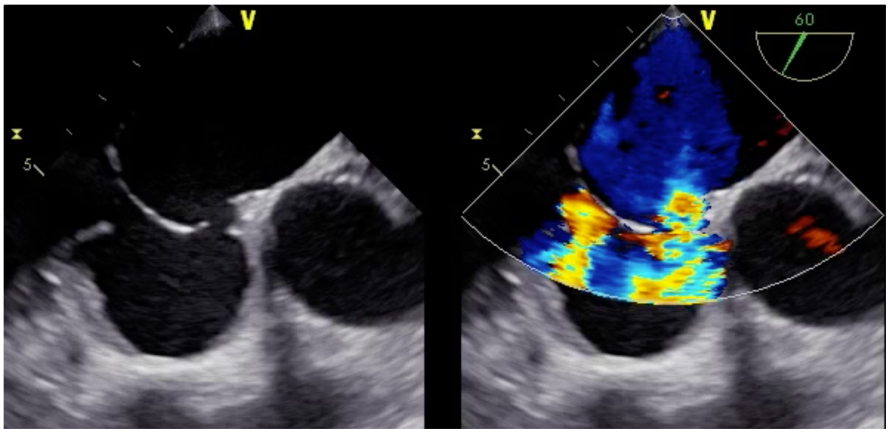

FIGURE 3 Transesophageal echocardiography reveals an atrial septal defect, with aortic rim deficiency and malalignment.

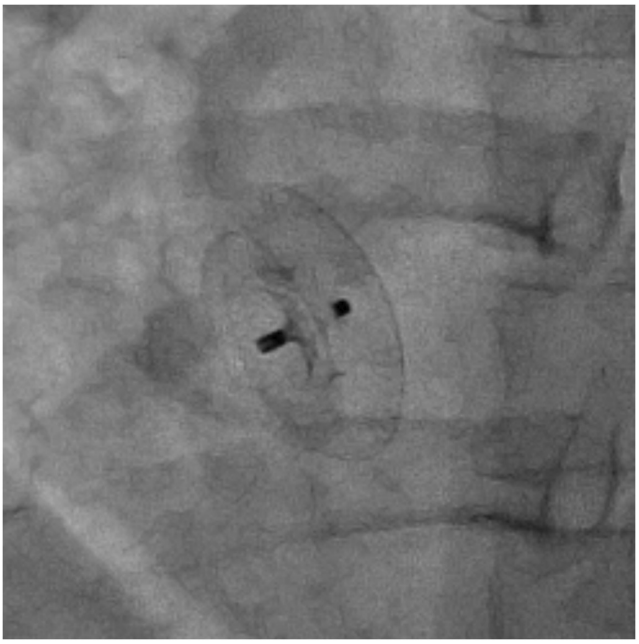

FIGURE 4 An Amplatzer septal occluder was successfully deployed.

CONSENT

We obtained written informed consent for publication from the patient.

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