Right Ventricular Blood Cyst in an Adult Autosomal Dominant Polycystic Kidney Disease Patient: An Unusual Association

Ersin Cagri Simsek[1], Dilek Oncel[2] and Erdem Ozel[1]

[1]*Department of Cardiology, University of Health Science, Tepecik Training and Research Hospital, Izmir, Turkey*
[2]*Department of Radiology, University of Health Science, Tepecik Training and Research Hospital, Izmir, Turkey*

ABSTRACT

Blood cysts of the heart are unusual primary cardiac tumor-like masses, which are usually located on the cardiac valves. These are very rare in adults, especially in a non-valvular location. Autosomal-dominant polycystic kidney disease (ADPKD) typically presents with multiple bilateral renal cysts, resulting in chronic kidney disease. Whilst many of the extra-renal manifestations of ADPKD are well-documented, associated cardiac masses are extremely rare: and cardiac blood cyst has not been reported in a patient with APKD to date. We present a 57-year man with a history of ADPKD and end-stage renal disease with a 2-cm-diameter right ventricular blood cyst, which was detected on multimodality imaging.

Key words: *Autosomal dominant polycystic kidney disease, Cardiac blood cyst, Extra-renal manifestations.*

How to cite this article: Simsek EC, Oncel D, Ozel E. Right Ventricular Blood Cyst in an Adult Autosomal Dominant Polycystic Kidney Disease Patient:
An Unusual Association. *J Coll Physicians Surg Pak* 2021; **31(12)**:1500-1502.

INTRODUCTION

Autosomal-dominant polycystic kidney disease (ADPKD) is an inherited disease, which is characterized by multiple, bilateral renal cysts; and extra-renal manifestations such as liver cysts as the most common manifestation. The estimated prevalence of ADPKD is 5–10% in end-stage renal disease (ESRD) patients.[1] Besides polycystic liver disease, ADPKD is associated with an increased risk of hypertension, cardiac disease, intracranial aneurysm, colonic diverticula, and cysts in other organs such as seminal vesicles, pancreas, and arachnoid membrane.[2] Herein, we report a rare case of ADPKD coexisting with a right ventricular blood cyst.

CASE REPORT

A 57-year man with a history of hypertension, ADPKD and ESRD presented at the Cardiology Outpatient Clinic with dyspnea. He had been on routine hemodialysis programme for the last two years. On admission, his blood pressure was 180/90 mmHg despite taking amlodipine (10 mg/per day) as antihypertensive treatment.

Correspondence to: Dr. Ersin Cagri Simsek, Department of Cardiology, University of Health Science, Tepecik Training and Research Hospital, Izmir, Turkey
E-mail: ercagsim@hotmail.com

Received: December 10, 2019; Revised: June 21, 2020; Accepted: August 14, 2020
DOI: https://doi.org/10.29271/jcpsp.2021.12.1500

Aside from abdomen fullness in the left upper quadrant, his physical examination was normal. He had no family history of cardiac disease or ADPKD. Among his routine laboratory data, hemoglobin was 15.2 g/dL, blood urea was 140 mg/dL, and serum creatinine was 5.2 mg/dL. Alkaline phosphatase (ALP) was 122 U/L (normal range: 30-120 U/L), and gamma-glutamyl transferase (GGT) 83 U/L (normal range: 0-55 U/L). Serological tests of hydatid disease were negative. Trans-thoracic echocardiography (TTE) detected a cyst of 20×18 mm size originating from right ventricular mid-septum without causing right ventricular outflow obstruction or tricuspid valve insufficiency (Figure 1). The cyst was well-circumscribed, with a thick wall, and hypo-echogenic core. The right-ventricular size and function were found to be normal. Cardiac and full abdominal magnetic resonance imaging (MRI) were performed to determine the type of cyst in the right ventricle and to look for cysts in other localisations. Cardiac MRI showed an immobile, thick walled cyst, approximately 2 cm in size, which was attached to the interventricular septum in the right ventricle (Figure 2). Abdominal MRI demonstrated numerous cysts of varying sizes throughout the liver and kidneys, which were different from the cardiac cysts in terms of MRI density (Figure 2F). Computed tomography (CT) imaging of the abdomen also confirmed the presence of multiple cysts in liver and kidneys (Figure 3). According to MRI and CT findings, cardiac cyst in right ventricle was diagnosed as 'cardiac blood cyst'. Since right ventricular blood cyst did not lead to obstruction or mechanical dysfunction, surgical resection was not deemed necessary. So, histopathological diagnosis could not be confirmed.

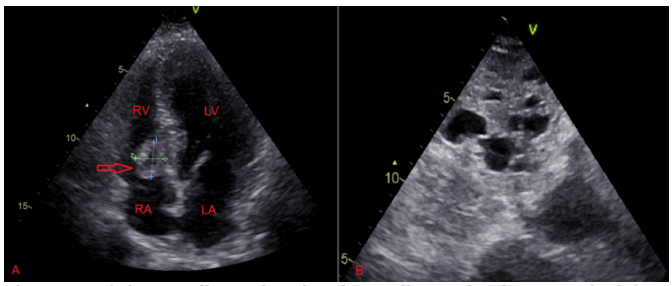

Figure 1: (A) Two-dimensional echocardiography in an apical-four chamber and (B) subcostal echocardiography views showing the blood cyst (arrow); attached to the interventricular septum in A and multiple hepatic cysts in B.

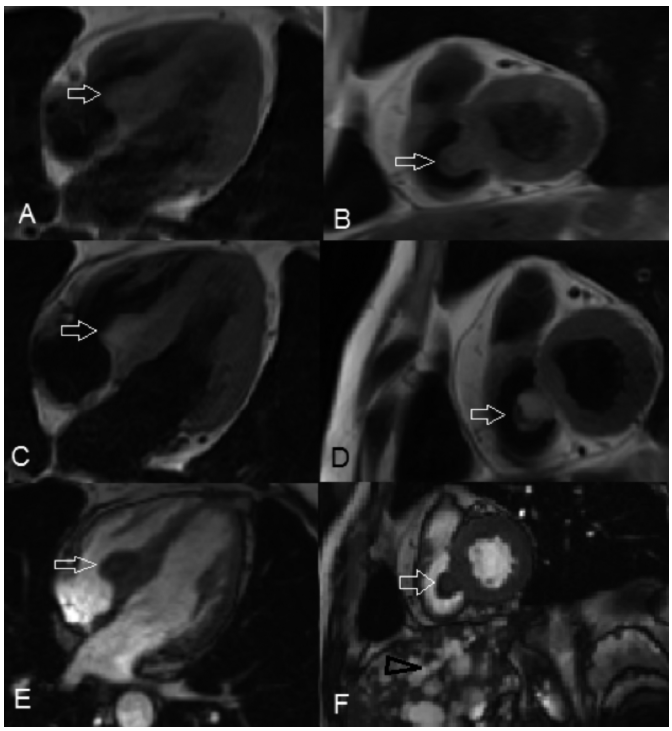

Figure 2: Cardiac magnetic resonance showing a fixed-cystic mass (arrows) with thick walls and attached to the interventricular septum without evidence of tricuspid valve dysfunction. A-B: T1-weighted image in four-chamber and short-axis views, C-D: T2-weighted image in four-chamber and short-axis views, E-F: Cine image in four-chamber and short-axis views (arrowhead; multiple hepatic cysts).

DISCUSSION

The reported cardiac complications of ADPKD in the literature are cardiac valvular abnormalities (mostly mitral valve prolapse), pericardial effusion, left ventricular hypertrophy, and coronary artery aneurysms.[2] The co-existence of ADPKD with a cardiac mass is extremely rare. In the literature, there are few cases which report the coexistence of ADPKD with atrial myxoma and pericardial cyst.[3-5] However, the co-existence of ADPKD and cardiac cyst in a heart chamber has not been reported before.

Cardiac blood cysts are rare benign lesions and usually located on the cardiac valves.[6] They are most commonly seen in pediatric age and are extremely rare in adults especially in a non--valvular location.[7,8] There is no consensus for the optimal management of patients with cardiac blood cysts in literature.

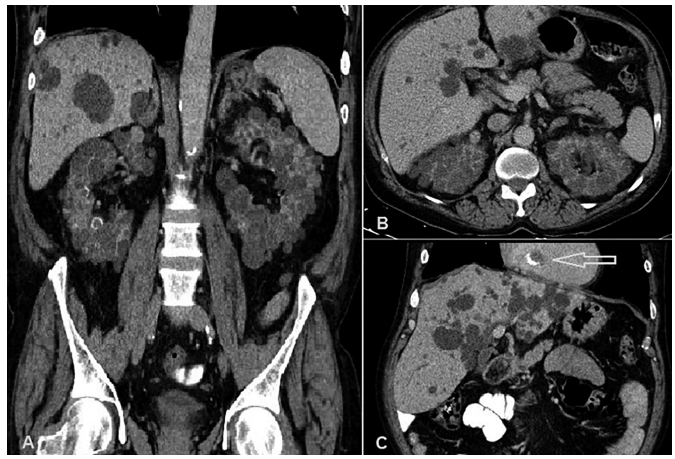

Figure 3: Non-contrast enhanced computed tomography demonstrating multiple cysts in the liver and markedly enlarged bilateral kidneys by innumerable cysts (A-C). Calcific-walled cardiac mass is also noted (arrow).

ADPKD is caused by a mutation in PKD1 or PKD2 genes that both cause morphological changes in renal epithelial cells.[1] Cyst formation is stimulated *via* these genes, which lead to connective tissue abnormalities and formation of extracellular matrix.[2] Extra-renal endothelial localisation of proteins encoded by PKD1 or PKD2 genes could explain the extra-renal manifestations of ADPKD. The co-existence of cardiac blood cyst and ADPKD in our patient could be co-incidental, but cardiac blood cyst formation may also be the result of a generalised defect in epithelial and/or extracellular matrix function as an expression of the genetic abnormality ADPKD.

In conclusion, this is a rare case of right ventricular blood cyst in adult age; but also to the best of our knowledge, the first case demonstrating the co-existence of cardiac blood cyst and ADPKD in the literature.

PATIENT'S CONSENT:

Informed consent was taken from the patient to use the descriptive information and radiological images.

CONFLICT OF INTEREST:

The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

ECS: Literature search and follow up.
DO: Data collection, literature search.
EO: Final draft of the case report.

REFERENCES

1. Torres VE, Harris PC, Pirson Y. Autosomal dominant polycystic kidney disease. *Lancet* 2007; **369(9569)**: 1287-301. doi: 10.1016/S0140-6736(07)60601-1.
2. Luciano RL, Dahl NK. Extra-renal manifestations of autosomal dominant polycystic kidney disease (ADPKD): Considerations for routine screening and management. *Nephrol Dial Transplant* 2014; **29(2)**:247-54. doi: 10.1093/ndt/gft437.
3. Iglesias D, Fraga AR, Arrizurieta E, Martin RS, Herrera M, Bernath V, *et al.* Atrial myxoma in a woman with autosomal dominant polycystic kidney disease type-2. *Am J Kidney Dis*

1997; **29(1)**:164-5. doi: 10.1016/s0272-6386(97)90024-x.

4. Alfishawy M, Ali H, Daoud A. A patient with polycystic kidney disease, hepatic cysts and atrial myxoma. *Saudi J Kidney Dis Transpl* 2018; **29(3)**:723-7. doi: 10.4103/1319-2442.235204.
5. Rock W, Mazen E, Saliba W. Pericardial cyst: A novel extrarenal manifestation of autosomal dominant polycystic kidney disease. *Isr Med Assoc J* 2011; **13(10)**:630-1.
6. Basso C, Valente M, Poletti A, Casarotto D, Thiene G. Surgical pathology of primary cardiac and pericardial tumors. *Eur J Cardiothorac Surg* 1997; **12(5)**:730-7; discussion 737-8. doi: 10.1016/s1010-7940(97)00246-7.
7. Roubelakis A, Moorjani N, Modi A, Harden S, Ohri SK. Right ventricular blood cyst causing outflow tract obstruction in an adult. *J Card Surg* 2012; **27(4)**:452-4. doi: 10.1111/j.1540-8191.2012.01455.x.
8. Akutsu H, Muraoka A, Misawa Y. Left ventricular blood cyst in an adult case. *Clin Case Rep* 2017; **5(9)**:1441-3. doi: 10.1002/ccr3.1083.