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Pericardial Cyst: a Novel Extrarenal Manifestation of Autosomal Dominant Polycystic Kidney Disease

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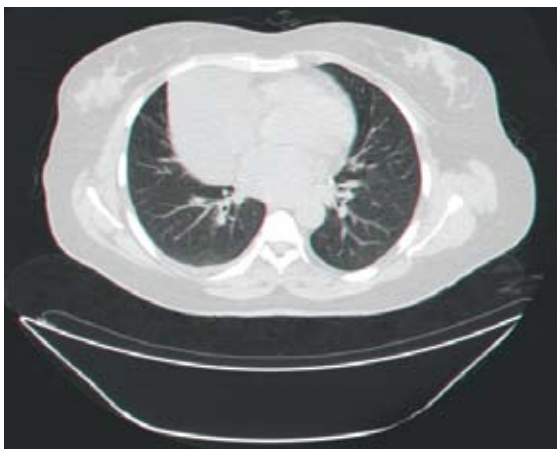
Autosomal dominant polycystic kidney disease is associated with cysts in the kidneys and, in many cases, asymptomatic cysts in the liver and pancreas that can help in confirming the diagnosis. In addition, patients may have a variety of other abnormalities, many of which are consistent with a generalized defect in epithelial cell differentiation and/or extracellular matrix function as a primary expression of the genetic abnormality in this disorder [1]. To date, the reported cardiac complications of ADPKD have included valvular abnormalities of uncertain significance (mainly mild mitral valve prolapse and aortic valve regurgitation), coronary artery aneurysms and asymptomatic pericardial effusions [2,3]. However, to the best of our knowledge, there have been only two reports of pericardial cysts in patients with ADPKD [4,5]. We report such an association in a woman who was diagnosed with ADPKD 13 years earlier and presented to the emergency department in our center with cough and dyspnea.

ADPKD = autosomal dominant polycystic kidney disease

Chest tomography of the patient showing large right paramediastinal pericardial cyst



PATIENT DESCRIPTION

A 48 year old woman presented to the emergency room with complaints of dry cough and dyspnea of a few days duration. She suffered neither fever nor chest pain and had no other symptoms suggestive of heart failure, including abdominal bloating and leg edema. On physical examination vital signs were normal. Jugular vein pressure was not elevated. Chest percussion, breath sounds, and palpation of the heart were all normal. Heart sounds were normal without murmurs or a friction rub. The abdomen was non-tender, and there was no evidence of ascitis. Both kidneys were easily palpable with numerous non-tender masses.

Blood chemistry revealed normal blood sugar, normal urea, elevated creatinine 2.3 mg/dl (normal 0.5–0.9 mg/dl), normal troponin and normal electrolytes. Complete blood count revealed a decreased hemoglobin concentration of 10 g/dl (normal 12–15 g/dl) without a significant deviation from the patient's usual hemoglobin concentration. Electrocardiogram was normal, but a chest X-ray revealed an enlarged cardiac shadow.

The patient was admitted to our department for further investigation. Her previous medical history was notable for ADPKD, which was diagnosed in 1996 via renal ultrasonography performed for the investigation of flank pain. Since then she has been under regular nephrology follow-up, during which her blood creatinine rose from 0.9 mg/dl in 1996 to 2.3 mg/dl in 2009. A cranial, thoracic and abdominal computed tomography scan performed in 1996 as part of a search for extrarenal manifestations of ADPKD revealed no cerebral aneurysms or hepatic cysts but did show minimal pericardial effusion. Since then the patient had not been under echocardiographic or cardiologic follow-up.

During her hospitalization she underwent a transthoracic echocardiography that revealed normal heart chamber size and normal ventricular function and no evidence of pericardial fluid. Due to the suspicion of a mediastinal mass a thoracic CT was performed, which revealed a right para-mediastinal lesion measuring 5 x 9 x 11 cm extending from the aortic arch until the right cardiophrenic angle, and was judged by density measurements to be a pericardial cyst [Figure]. During her hospitalization the patient's symptoms disappeared spontaneously. No further investigation was deemed necessary and she was discharged 2 days later with a presumptive diagnosis of upper respiratory infection and an incidental finding of an asymptomatic pericardial cyst.

COMMENT

The first description of the association between ADPKD and pericardial cysts was made by Paemelaère et al. in 1995 [4]. They reported a case of pericardial

cyst discovered incidentally in a 71 year old man who suffered from ADPKD and presented with prolonged chest pain. The diagnosis was made on thoracic CT. The second description of such an association was given by Hooda and Narula in 2005 [5]. They reported the case of a 32 year old man who presented with chest pain 3 years earlier, was diagnosed with pericardial cyst via transthoracic echocardiography, and underwent excision of the cyst. Histological examination confirmed the diagnosis of pericardial cyst. Later the patient was diagnosed with ADPKD. The authors argued that two-dimensional echocardiography remains a reliable method for diagnosing pericardial cysts, and when it is inconclusive transesophageal echocardiography is helpful.

It is noteworthy that in both of the two previous cases and the present case the pericardial cyst was an incidental finding and that in the present case transthoracic echocardiography failed to make the diagnosis.

There is a well-documented association between pericardial effusion and APKD that may share a common pathophysiology with pericardial cysts in ADPKD. This association was best shown in a retrospective analysis from the Mayo Clinic in which the presence and severity of pericardial effusions were analyzed by CT in 60 patients with ADPKD (mean serum creatinine concentration 1.8 mg/dl), 100 patients with chronic kidney disease not due to ADPKD (mean serum creatinine concentration 1.8 mg/dl), and 100 healthy kidney donors. A pericardial effusion was found in 35%, 9% and 4% of ADPKD patients, chronic kidney disease patients, and healthy donors respectively. A moderate to high effusion severity score was noted in nearly 50% of patients with ADPKD and an effusion, but in none of the others. Despite the size of the effusions they were generally well tolerated and clinically inconsequential. Based on these results the authors concluded that pericardial effusion is not merely due to uremia but is a previously unrecognized extrarenal manifestation of ADPKD. They also hypothesized that the pathogenesis of pericardial effusion in ADPKD may reflect a defect in the structure and function of connective tissue and extracellular matrix that may also underlie other extrarenal manifestations of ADPKD. These abnormalities are reproduced by targeted mutations of the *Pkd1* gene in mice [3].

Given the frequency and diversity of extrarenal manifestations of ADPKD, it seems plausible that the finding of pericardial cysts in patients with ADPKD is not merely coincidental but reflects an as yet under-reported complication of ADPKD that shares the same pathogenesis with pericardial effusion in ADPKD. Further studies need to be undertaken to determine the frequency and clinical significance of pericardial cysts in ADPKD.

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