Radio-Diagnostic Evaluation of Adult Polycystic Disease of the Kidneys and Liver in a 53-Year-Old Homemaker – A Case Report and Review of the Literature

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Abstract

Autosomal dominant polycystic kidney disease (ADKPD) is the most common hereditary kidney disease and one of the most common causes of end-stage kidney diseases, with approximately 30% of patients with ADKPD having polycystic liver in addition. A case of a 53-year-old homemaker who presented with marked abdominal swelling, distension, and respiratory distress, in whom ultrasonographic and computed tomographic features of multiple noncommunicating cystic lesions in the kidneys and the liver were demonstrated, was presented to emphasize the key role of medical imaging in the diagnosis of ADKPD. A review of the literature was also done.

Keywords: Adult polycystic kidney disease, autosomal dominant, genes, hereditary disease, polycystic kidney disease 1, polycystic kidney disease 2

Introduction

Autosomal dominant polycystic kidney disease (ADPKD) is the most common hereditary disease affecting the kidneys. It usually presents in adults and affects 1 in 400–1 in 1000 live births and accounts for 5% of end-stage renal disease in the United States and Europe. It is caused by a defect in the polycystic kidney disease (PKD1) or PKD2 genes.[1,2]

It is characterized by the development of multiple renal cysts variably associated with extrarenal (mainly hepatic and cardiovascular) abnormalities. Approximately 30% of patients with ADKPD have polycystic liver in addition, with a minority of them presenting with massive hepatomegaly.[1-3]

Case Report

OM is a 53-year-old homemaker. She presented to the Medical Outpatient Clinic of the University of Benin Teaching Hospital (UBTH). Her complaints were gradual abdominal swelling associated with heaviness and discomfort for about 11 years before presentation at the UBTH. She had sought medical attention from both orthodox doctors and herbal practitioners with no improvement before she decided to come to the UBTH.

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Clinical examination revealed a chronically ill-looking middle-aged woman. She was afebrile, but mildly pale, and in mild respiratory distress. The abdomen was markedly distended with a dull mass of about 20 cm below the right costal margin suggestive of a liver mass. Masses were also palpated in both flanks. Her blood pressure was 160/100 mmHg. The patient was admitted for investigations.

An abdominopelvic ultrasound scan done with a Sonoace machine fitted with a 3.5 MHz frequency probe showed a significantly enlarged liver measuring about 20 and 17 cm below the right and left costal margins, respectively. There were multiple noncommunicating anechoic cystic lesions within the liver parenchyma [Figure 1].

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Both kidneys were also enlarged with multiple noncommunicating cystic lesions measuring 22.4 cm × 10.4 cm and 23.4 cm × 10.2 cm for the right and left kidneys, respectively [Figures 2 and 3]. Massive ascites gravitating into the pouch of Douglas with bowel loops of normal caliber floating within was noted.

The gallbladder and pancreas were not demonstrated due to the massive cystic changes in the liver and kidneys. The spleen was of normal size and parenchyma echo pattern.

The uterus, adnexa, and urinary bladder were normal in anatomic configuration and echo pattern.

An abdominal computed tomography (CT) scan done showed multiple nonenhancing hypodense round lesions in the liver and both kidneys [Figure 4]. However, the pancreas and spleen could not be delineated because the liver and kidneys were markedly enlarged, thereby compressing the other organs.

Based on ultrasound and CT features, a diagnosis of adult PKD with liver involvement was made. The patient was scheduled for further investigation and conservative management but was lost to follow-up.

Discussion

There are approximately 600,000 PKD patients in the USA and over 10,000,000 worldwide, and it accounts for approximately 5% of nondiabetic dialysis and renal transplant.[3] ADPKD is caused by a defect in PKD1 or PKD2 genes, which are located on chromosomes 16 and 4, respectively. PKD1 accounts for about 85% of cases, while PKD2 is responsible for the remaining 25%.[1] Liver involvement is the most frequent extrarenal manifestation in adult PKD,[4] as was seen in this patient.

The ADKPD genotypes are characterized by bilateral kidney cysts, hypertension, hematuria, renal infection, stones, and renal insufficiency. ADPKD is a systemic disorder; cysts appear with decreasing frequency in the kidneys, liver pancreas, brain, spleen, ovaries, and testis.[5] This patient, a female, had cysts in both kidneys and liver and an elevated blood pressure of 160/100 mmHg. The other organs were spared.

DNA analysis is the gold standard for the diagnosis of ADPKD-2, especially in young people; however, ultrasound and CT examinations have become more reliable in excluding ADPKD-2 in family members over the age of 30 years, who are at 50% risk. The current patient, who is a 53-year-old homemaker, was diagnosed using B-mode real-time ultrasonography and CT. Parfrey *et al*.[6] observed in a study that in most persons with a 50% risk of ADPKD, imaging techniques are the only mode of reaching a diagnosis before symptoms appear.

Radiology plays an important role in the diagnosis and treatment of patients with ADPKD. Ultrasound using B-mode, CT, and magnetic resonance imaging (MRI) are the main radiological modalities for evaluating ADPKD.[7] The imaging appearances vary with the severity of the disease. Ultrasonography or CT is more sensitive in adolescent or young adult who is not yet clinically symptomatic. The full-blown disease presents with bilaterally enlarged kidneys with numerous cysts of varying size,[8] as was observed with this patient. The sensitivity of ultrasonography in patients aged 30 years and above is 100%. Ultrasonography is limited in the differentiation of hemorrhage into cysts or the presence of cyst infection as both conditions show internal echoes, fluid debris levels, and thickened walls.[8,9]

CT scanning is as sensitive as ultrasound in the detection of cystic disease and more specific than sonography in differentiating an obstructed renal pelvis from a parapelvic cyst. It is also superior to ultrasonography in assessing retroperitoneal rupture of a cyst and perinephric extension

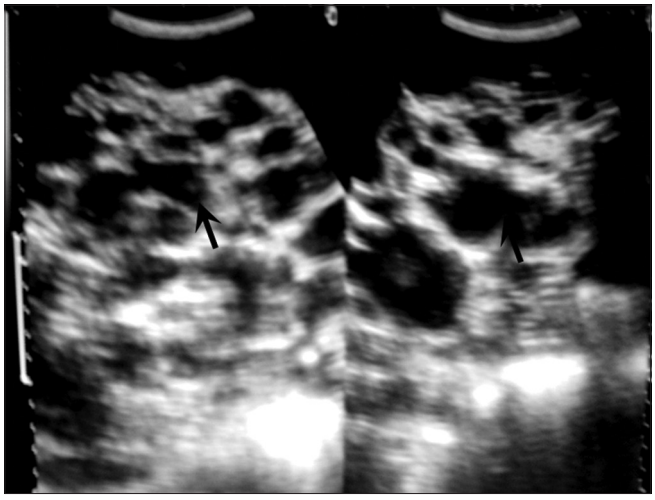

Figure 1: Abdominal sonogram showing multiple anechoic cystic lesions within the liver causing destructive compression of the liver parenchyma (arrows)

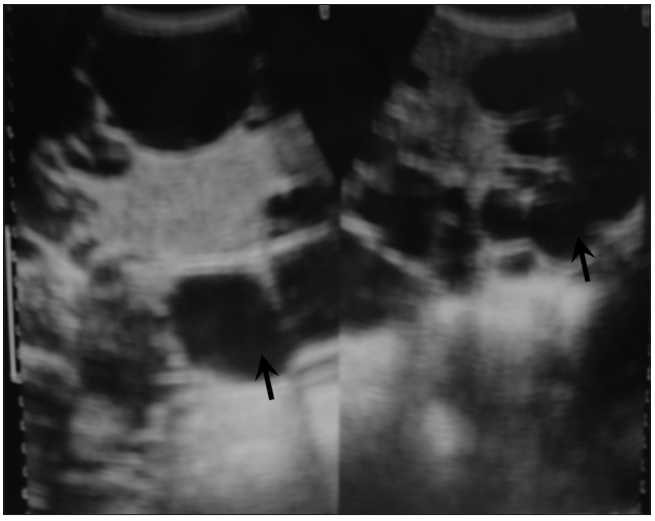

Figure 2: Abdominal sonogram showing multiple noncommunicating anechoic cystic lesions within the right kidney (upper and lower poles) causing destructive compression of the kidney parenchyma (arrows)

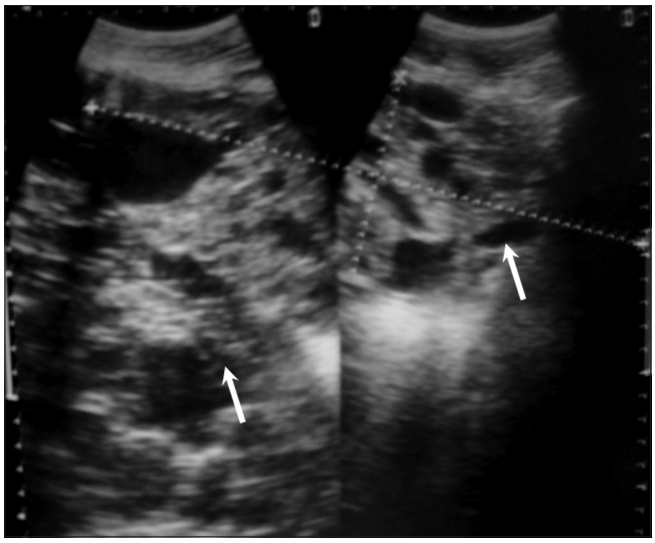

Figure 3: Abdominal sonogram showing multiple noncommunicating anechoic cystic lesions within the left kidney (upper and lower poles) causing destructive compression of the kidney parenchyma (white arrows)

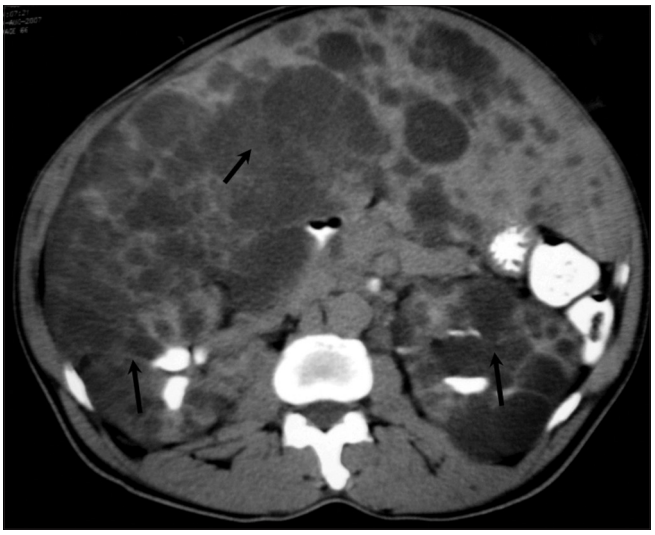

Figure 4: Abdominal contrast-enhanced axial computed tomography image showing multiple nonenhancing hypodense lesions in the liver and both kidneys (black arrows)

of blood or pus from an infected cyst.[10] This patient had the benefit of being evaluated with CT.

MRI does not use ionizing radiation or iodinated contrast, and it allows for multiplanar imaging. It is, therefore, useful for examining patients who are allergic to iodinated contrast media and those with compromised renal function, who are at risk of iodinated contrast-induced renal failure. MRI is a useful modality in differentiating between simple cysts and neoplasms when the findings on CT and ultrasonography are equivocal. There was no need for the evaluation of this patient with MRI as the diagnosis was conclusive on ultrasonographic and CT assessment.[8,10] Ultrasonography, CT, and MRI have been used for many years to quantify the increase in renal volume in patients with ADPKD. Imaging with these techniques has also been used to accurately quantify the rate of increased kidney and total cyst volume in patients. In a review, Bae and Grantham,[11] discussed the overwhelming evidence in support of the view that imaging is an invaluable tool to monitor the onset and progression of ADPKD and is well-suited to gauge the response of this disease to targeted therapy before renal function begins to decline.

Angiography is invasive and of limited role in the diagnosis of ADPKD, while radionuclide studies play a complementary role in the assessment of renal function; however, our patient was not evaluated with this modality due to its nonavailability.[10]

Treatment of ADPKD is supportive. It includes dietary regulation of salt and protein intake, control of hypertension and renal stones, and dialysis and transplantation at the end stage.[12] However, there are new insights into the genetic basis and therapeutic options of ADPKD. Identification of the PKD1 and PKD2 gene mutations has spurred recent insights into the molecular pathophysiology of PKD, with the suggestion that the PKD1-encoded protein, polycystin-1, is a renal epithelial cell membrane mechanoreceptor.[13] This could be the subject of research into new therapeutic agents targeted at the prevention of PKD or to slow down the progression of the disease.

In some reviews of new insights into the treatment of ADPKD, Murcia *et al*.[14] and Helal[15] identified promising treatments, which include dual inhibition of the renin-angiotensin-aldosterone system (RAAS), vasopressin receptor antagonists, increased fluid intake, and oral antiglycemic agents that block certain receptors of cyclic adenosine monophosphate, which in turn reduce the rate of growth of the cysts and ADPKD progression. However, the main and most effective therapy remains the control of hypertension by RAAS blockade, which leads to improvement in ADPKD outcomes.[15]

The patient was lost to follow-up after the initial set of radiological evaluations.

Summary

A case of adult PKD with liver involvement in a 53-year-old homemaker has been presented to underscore the important role of radiological modalities in the diagnosis and management of ADPKD. Some new insights into molecular pathophysiology and new potential therapeutic options were also discussed.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published, and due efforts will be made to conceal her identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

References
1. Baker LR. Renal disease. In: Kumar P, Clark M, editors. Clinical Medicine. 5th ed. Toronto: WB Saunders; 2002. p. 658-60.
2. Tahvanainen E, Tahvanainen P, Kääriäinen H, Höckerstedt K. Polycystic liver and kidney diseases. Ann Med 2005;37:546-55.
3. Grantham JJ. The Jeremiah Metzger lecture. Polycystic kidney disease: Old disease in a new context. Trans Am Clin Climatol Assoc 2002;113:211-24.
4. Chauveau D, Fakhouri F, Grünfeld JP. Liver involvement in autosomal-dominant polycystic kidney disease: Therapeutic dilemma. J Am Soc Nephrol 2000;11:1767-75.
5. Demetriou K, Tziakouri C, Anninou K, Eleftheriou A, Koptides M, Nicolaou A, *et al.* Autosomal dominant polycystic kidney disease-type 2. Ultrasound, genetic and clinical correlations. Nephrol Dial Transplant 2000;15:205-11.
6. Parfrey PS, Bear JC, Morgan J, Cramer BC, McManamon PJ, Gault MH, *et al.* The diagnosis and prognosis of autosomal dominant polycystic kidney disease. N Engl J Med 1990;323:1085-90.
7. Saxena AK, Karnatakam A. Autosomal dominant polycystic kidney disease and pain: Radiologist's perspective. J Postgrad Med 2004;50:308.
8. Rottenberg G, Rankin S. Renal masses In: Grainger RG, Allison DJ, editors. Diagnostic Radiology. A Textbook of Medical Imaging. 5th ed. Philadelphia: Churchill Livingstone; 2008. p. 857-61.
9. Nicolau C, Torra R, Badenas C, Vilana R, Bianchi L, Gilabert R, *et al.* Autosomal dominant polycystic kidney disease types 1 and 2: Assessment of US sensitivity for diagnosis. Radiology 1999;213:273-6.
10. Khan AN. Autosomal Dominant Polycystic Kidney Disease; 2007. Available from: https://www.emedicine.
11. Bae KT, Grantham JJ. Imaging for the prognosis of autosomal dominant polycystic kidney disease. Nat Rev Nephrol 2010;6:96-106.
12. Martinez JR, Grantham JJ. Polycystic kidney disease: Etiology, pathogenesis, and treatment. Dis Mon 1995;41:693-765.
13. Wilson PD. Polycystic kidney disease: New understanding in the pathogenesis. Int J Biochem Cell Biol 2004;36:1868-73.
14. Murcia NS, Sweeney WE Jr., Avner ED. New insights into the molecular pathophysiology of polycystic kidney disease. Kidney Int 1999;55:1187-97.
15. Helal I. Autosomal dominant polycystic kidney disease: New insights into treatment. Saudi J Kidney Dis Transpl 2013;24:230-4.