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Case Report

Incidental finding of autosomal dominant polycystic kidney disease in a 52-year-old man: a case report.

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ABSTRACT

Background: Autosomal dominant polycystic kidney disease (ADPKD) also known as Adult Polycystic Kidney Disease (APKD) is one of the most common systemic hereditary diseases. The disease usually presents between 20 to 39 years of age, although milder forms may not present until over 60 years and absence of renal failure has been rarely observed in some patients up to 80 years of age. Three distinct gene defects have been implicated in the pathogenesis of APKD designated as *PKD1*, *PKD2*, and *PKD3*. Patients with APKD may be asymptomatic, or may usually present with hypertension (in 50-70% of cases), renal insufficiency, and the complications of multiple cysts (haematuria, pain and infection) or as an abdominal mass discovered on incidental clinical or imaging examination. APKD is said to be rare in Africans. **Case report:** We report a case of an incidental finding of autosomal dominant polycystic kidney disease (ADPKD) in a 52-year-old normotensive man with a normal renal function test. Haematuria, dysuria and low-grade fever were the presenting complaints. The role of radiology in the diagnosis and management of ADPKD was highlighted. **Conclusion:** Although APKD is rare in Africans, a high index of suspicion for the disease is essential for the diagnosis of the disease especially in elderly patients with a family history of APKD who present with haematuria associated with multiple renal cysts.

Keywords: *Adult Polycystic Kidney Disease, Autosomal Dominant Polycystic Kidney Disease, Incidental finding, Normotensive.*

Introduction

Autosomal dominant polycystic kidney disease (ADPKD) also known as Adult Polycystic Kidney Disease (APKD) is one of the most common systemic hereditary diseases.[1] Its prevalence is estimated to be between 1 in 400 and 1 in 1000.[1,2] APKD accounts for 10% to 15% of end-stage renal disease cases requiring dialysis.[1,3] It is characterized by renal cyst growth and enlargement leading to renal failure[4] and affects both sexes in equal proportion without racial predilection.[5] However, APKD is said to be rare in Africans.[6]

Three distinct gene defects have been implicated in the pathogenesis of APKD designated as *PKD1*, *PKD2*, and *PKD3*.[1-4,7] In 1985, Reeders *et al*[8] identified a defect on chromosome 16p13 (*PKD1*) that is responsible for 85% of the cases of APKD. The *PKD2* is located on chromosome 4q21 and accounts for 10% to 15% of patients with APKD.[7]

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This high phenotypic variability is contributed by genic, allelic and gene-modifier factors. The *PKD1* gene is associated with more severe disease than *PKD2*. The greater severity of *PKD1* is caused by the development of more cysts at an early age resulting in end-stage renal disease (ESRD) at an earlier age.[10]

As the disease progresses it may be associated with the presence of cysts in other organs including the liver (50-80%), pancreas (9%) and rarely in the lungs, spleen, thyroid, ovaries, uterus, and testis.[11] Other associations include intracranial aneurysms (3-13%), mitral valve prolapse, colonic diverticulosis and renal stones formation.[7]

Pathologically, numerous cysts of varying sizes, often becoming extremely large, develop within the kidneys, gradually replacing normal renal parenchyma and ultimately producing renal failure.[9] Patients with APKD may be asymptomatic,[1] or may usually present with hypertension (in 50-70% of cases), renal insufficiency, and complications of multiple cysts (haematuria, pain and infection) or as an abdominal mass discovered on incidental clinical or imaging examination.[9] The disease causes renal failure in 25% of patients by the age of 50 and in 50% of patients by the age of 70 depending on the type of gene defect involved; hence, patients with *PKD1* have renal symptoms earlier and progress to kidney failure at a mean age of 54.3-56.7 years while patients with *PKD2* manifest with renal failure at a mean age of 69.4-74.0 years.[7] Imaging studies not only help in the diagnosis but also play an important role in the assessment of complications, prognosis, the efficacy of treatment, and long-term follow-up.[3,5] This report presents a case of an incidental finding of APKD in a 52-year-old normotensive man with normal renal functions.

Case presentation: M.A.A. is a 52-year-old farmer who presented to the general outpatient department (GOPD) of the University of Maiduguri Teaching Hospital with a two-month history of total haematuria and three weeks history of bilateral loin pains, dysuria, and low-grade fever. The patient thought he was having bilharziasis (urinary schistosomiasis) when he started passing bloody urine. There was no history of headache, vomiting, easy satiety, weight loss, cough or drenching night sweats. The patient said he has never been hospitalized in the past. His immediate elder brother died from kidney problems five years previously. He does not smoke or consume alcohol. He was not a known hypertensive or diabetic. The patient was married to three wives and had sixteen children, all alive and well.

On physical examination, he was found to be a middle-aged man, conscious, afebrile, with conjunctival pallor. No pitting pedal oedema was noted. Abdominal examination revealed mild fullness of the flanks bilaterally, with mild tenderness. The liver and spleen were not enlarged. Cardiovascular examination revealed a pulse rate of 76 beats per minute, regular and of normal volume. His blood pressure was 130/80 mmHg. Other system examinations were essentially normal.

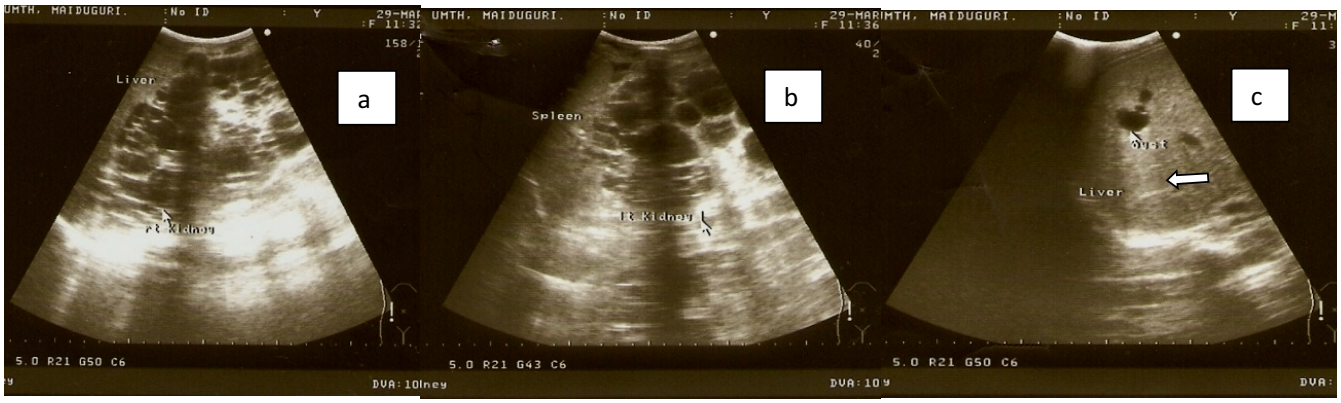
Abdominal ultrasonographic scans (Figs. 1a, b, and c) showed gross enlargement of both kidneys with each measuring >20cm in its bipolar length. Associated distortion of the renal architecture was also noted. Both kidneys contained multiple cysts of varying sizes, with the largest measuring 3.0cm x 3.5cm in dimension. There was no evidence of calcification seen. Cysts of varying sizes were also noted in the liver with the largest in the right lobe measuring 2.5cm x 3.0cm in dimension. The pancreas, spleen, urinary bladder, and prostate were normal.

Magnetic resonance imaging (MRI) scans of the abdomen (Figs. 3 and 4) revealed multiple fluid-filled cysts (hypointense on T1W and hyperintense on T2W sequences) involving both kidneys which appeared grossly enlarged. The liver also showed two intrahepatic cysts involving the right lobe and a peripheral cyst in the left lobe, hyperintense on T2W sequence (Fig. 5). Computed tomography (CT) scan was not done at the time the patient presented.

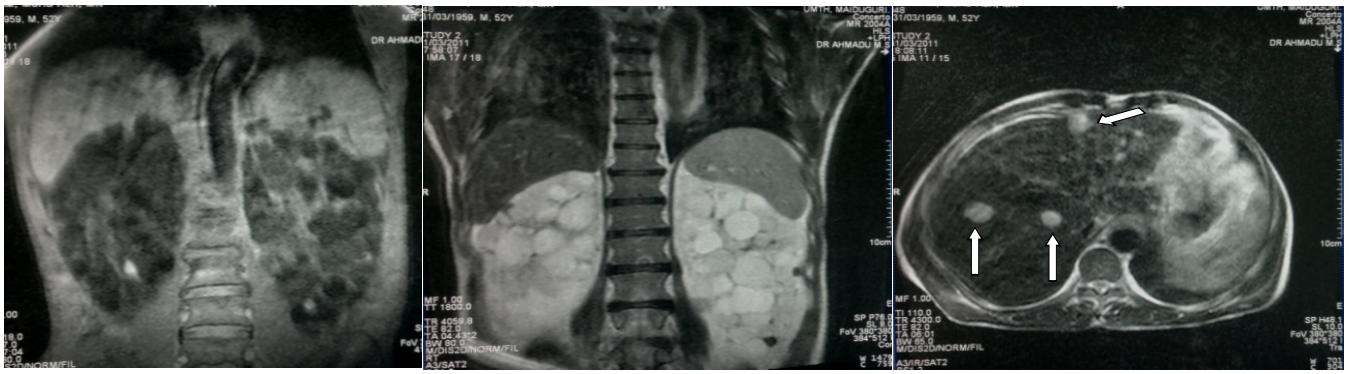
Packed cell volume (PCV) was 26%; serum electrolytes were within normal limits; Blood urea was 5.1mmol/L; serum creatinine was 130µmol/L. The liver function test was normal. However, urinalysis showed blood 2++, and urine microscopy, culture and sensitivity yielded significant bacterial growth. Urine cytology was normal.

Based on the ultrasonographic and MRI scan findings a diagnosis of ADPKD was made and the patient was

admitted due to the haematuria and low PCV of 26% and was transfused 3 pints of blood. He was also treated for urinary tract infection (UTI) to which he responded well and the haematuria subsided markedly. His post-transfusion PCV was 34%. He was discharged home after one week of admission and was placed on two weekly follow-ups to monitor his blood pressure and function for signs of affectation and associated complications of the ADPKD. However, the patient was lost to follow-up after two clinic visits.



Figures 1(a), (b), and (c): Abdominal ultrasonography (longitudinal views) of the right and left kidneys, and the liver showing gross enlargement of both kidneys with each measuring >20cm in its bipolar dimension and containing multiple asymmetrical cysts of varying sizes. Associated distortion of the renal architecture was also noted. A cyst in the right lobe of the liver is noted. Note the posterior acoustic enhancement (white arrow).



Figures 2 (a), (b), and (c): Coronal T1W and T2W images of the abdomen showing grossly enlarged kidneys containing hypointense on T1W sequence and hyperintense on T2W sequence lesions representing multiple renal cysts. Image (c) is an axial T2W image of the abdomen showing hyperintense liver cysts; two intrahepatic in the right lobe and one located peripherally in the left lobe (white arrows).

Discussion

The diagnosis of APKD may first be suspected based on an imaging test, such as ultrasonography, performed for some other reason.[2] The APKD in the case presented became apparent after he had an ultrasound scan done, hence making the diagnosis of APKD incidental. Initially, APKD was not suspected as the patient did not present with the classical features of hypertension and renal function derangement.

Adult polycystic kidney disease has also been reported to be rare in Africans.[6] This fact had also made the diagnosis of APKD in the case presented unlikely initially. Although no direct family history of APKD could be ascertained in the case presented, however, it was reported that the patient's immediate elder brother died from kidney problems.

Gross haematuria is often the first presenting sign of APKD and the reason patients first seek medical attention,[7] but APKD was not suspected in the index case because of his age at presentation. Gross

haematuria is usually secondary to renal cyst rupture into the renal pelvis. Infection, segmental renal infarction, and passage of renal calculi also cause gross haematuria in APKD patients.[3] Haematuria was the reason the patient in this case report presented to the hospital. He also had UTI. The haematuria in this case presented may have been caused by renal cysts rupture. Renal malignancy could be a differential diagnosis in a patient with haematuria.[7] However, the urine cytology of the patient in the case presented was normal.

The most common extrarenal manifestation of APKD is hepatic cysts and their incidence increases in the second through fifth decades of life and by 50 years of age 80% of patients with APKD have hepatic cysts.[7] The case reported also presented with hepatic cysts at 52 years of age, which is in agreement with the existing literature.[7-9,11] Females are more likely to form hepatic cysts and their development is believed to be related to female sex hormones and pregnancy.[7,8] However, the case presented was a male patient. The presence of cysts in other organs apart from the liver has also been reported.[9] However, the patient in this case report only presented with cysts in the liver.

It has been reported that 50-70% of patients with APKD present with hypertension and that patients with *PDK1* are four times more likely to suffer from hypertension than patients with *PKD2*.[7,9] Patients with *PKD2* have also been reported to have a less severe course of the disease.[2] Absence of renal failure has also been observed in some patients up to 80 years of age by Julian and Carl.[9] Although no gene mapping was done in this patient to ascertain the genetic cause of his APKD, it may be inferred from his normotensive status that the gene defect could be due to *PKD2*. The presence of hypertension has also been predicted to serve as an indicator for poor kidney outcomes in APKD.[2] The case presented neither had hypertension nor renal function derangement (an indicator of poor kidney outcome).
Ultrasonography, CT scan, and MRI are the main radiological imaging modalities for the evaluation of patients with APKD. T2W MRI is more sensitive and identifies renal cysts as small as 3 mm in diameter. Contrast-enhanced CT is equally sensitive but involves the use of ionizing radiation and iodinated rather than gadolinium-containing contrast medium.[5] Computed tomography and MRI may also be employed to detect associations and complications of APKD including cysts and vascular complications. The case presented had ultrasonography and MRI done and features of APKD were noted.

Intravenous Urography (IVU) of patients with APKD demonstrates characteristic stretching of calyces by the cysts.[9] Technetium-99m DMSA scan shows the reduced function of the affected kidney if unilateral, or poor visualisation of the kidneys if bilateral.[1] Although plain films have no role in the surveillance of patients with established APKD, the diagnosis may be suspected when the renal outlines are enlarged, multilobulated or difficult to discern, with an associated displacement of loops of bowel which are non-specific findings.[7]

Management of patients with APKD is aimed at treating complications of the disease.[7] Reduction of the kidney size by ultrasound-guided needle aspiration and sclerosing of the renal cysts is also a treatment option. Although percutaneous sclerotherapy may be difficult in APKD due to the presence of multiple cysts, selective ablation (under sonographic guidance) is, however, effective.[4] In the report presented the patient was managed conservatively. He was also treated for urinary tract infection and placed on the monthly follow-up to monitor signs of complications of the APKD. However, the patient was lost to follow-up after two clinic visits.

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