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Case Report

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Autosomal dominant polycystic kidney disease: are we underestimating the burden in Africa?

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Abstract

Autosomal dominant polycystic kidney (ADPKD) is a multisystemic disease previously referred to as adult polycystic kidney disease. It is inherited in an autosomal dominant fashion and has 100% penetrance. ADPKD is rare among Africans. In this report, a middle-aged woman that met the diagnostic criteria for ADPKD is presented. She was evaluated and managed, but none of her first-degree relatives was available for screening despite adequate education and counselling.This report suggests that the poor health seeking disposition of some Africans may be leading to under-reporting of ADPKD among Africans.

Keywords: Africans, Autosomal dominant polycystic kidney, adult polycystic kidney disease

Introduction

Autosomal dominant polycystic kidney disease (ADPKD) is a multisystemic disease characterized by multiple, bilateral renal cysts, associated with cysts in other organs such as the liver, pancreas, arachnoid membrane and seminar vesicle. Non cystic extra renal manifestations include intracranial aneurisms, cervico-cephalic arterial dissection, thoracic aortic dissection, mitral valve prolapse and valvular insufficiency [1]. It was suggested to be exceptionally rare among black Africans [2-4]. However, the availability and increased utilization of imaging techniques have led to increase in the diagnosis of ADPKD in our environment [5]. In a previous report, a prevalence of 8% among patients with renal disease was recorded in Ilorin [5]. A retrospective study in Dakar found a prevalence of 1 in 250 of black population [3].

The report of rarity of the disease in Africa may also not be unconnected with poor health seeking disposition of some Africans, and reluctance to present for screening when they are asymptomatic.

Case

A 55 yr old female fish seller was referred by the family physician to the Nephrology Clinic of University of Ilorin Teaching Hospital, Ilorin, Kwara State, Nigeria, on account of recurrent abdominal pains and progressive abdominal swelling of 4 years duration. She was in her usual state of health until about 4years before presentation when she first noticed left sided abdominal pains, dull in nature, radiating to the right side of the abdomen. There was no known relieving or aggravating factors, but was associated with recurrent low-grade fever, chills and rigor. Pain did not disturb her daily activities. Anorexia, nausea, vomiting, diarrhoea or progressive weight loss were absent, but there was occasional constipation. There was no yellowness of the eyes at any time. She also observed progressive abdominal swelling without associated swelling of any other part of the body. There was nocturia, recurrent dysuria and urgency, but no frothy urine, haematuria or reduction in urine output. The patient experienced occasional throbbing headache, but there was no convulsion, neck pains or weakness of any part of the body. She was diagnosed to have systemic hypertension 4years before presentation at a general hospital, and placed on four drugs which could not be specified. The patient reported that she stopped the antihypertensive drugs a year before presentation. She was not a known diabetic, epileptic or sickle cell disease patient. She was married with 5 children and history of similar illness in her first-degree relatives could not be ascertained. There was no family history of diabetes mellitus, systemic hypertension, stroke or

kidney disease. She neither smoked cigarette nor took alcoholic drinks. All her pregnancies, labour and deliveries were uneventful.

Clinical examination revealed that she was febrile (temperature of 38.2˚C), not pale, anicteric or cyanosed. There was no finger clubbing, significant peripheral lymph node enlargement or pedal oedema. Her abdomen was distended, moved with respiration, and there was tenderness in the right hypochondrium, right and left lumber region, liver was span was 16cm. Spleen was not palpably enlarged. Both kidneys were ballotably enlarged, firm and tender. No ascites, bowel sounds were normoactive and digital rectal examination was essentially normal. She had a pulse rate of 84/min, which was regular and of normal volume, and the arterial wall was not thickened. Blood pressure was 170/100mmHg taken at the right arm in supine position. The jugular venous pressure and heart sounds were normal. Respiratory and nervous system examinations were essentially normal.

Abdominopelvic ultrasound scan revealed hepatomegaly due to multiple cysts involving both lobes of the liver (Figure 1). The intrahepatic biliary tree was poorly defined due to the cysts. The spleen was within normal limits; the pancreas and paraaortic areas were also ill-defined due to extensive cystic involvement. Both kidneys were grossly enlarged with the right measuring 18.3 x 9.2cm and the left 18.5 x 8.1cm due to multiple huge renal cysts. The bowel loops were displaced downwards and centrally. The cysts are still seen in the right and left iliac fossa. The urinary bladder and the uterus were within normal limits.

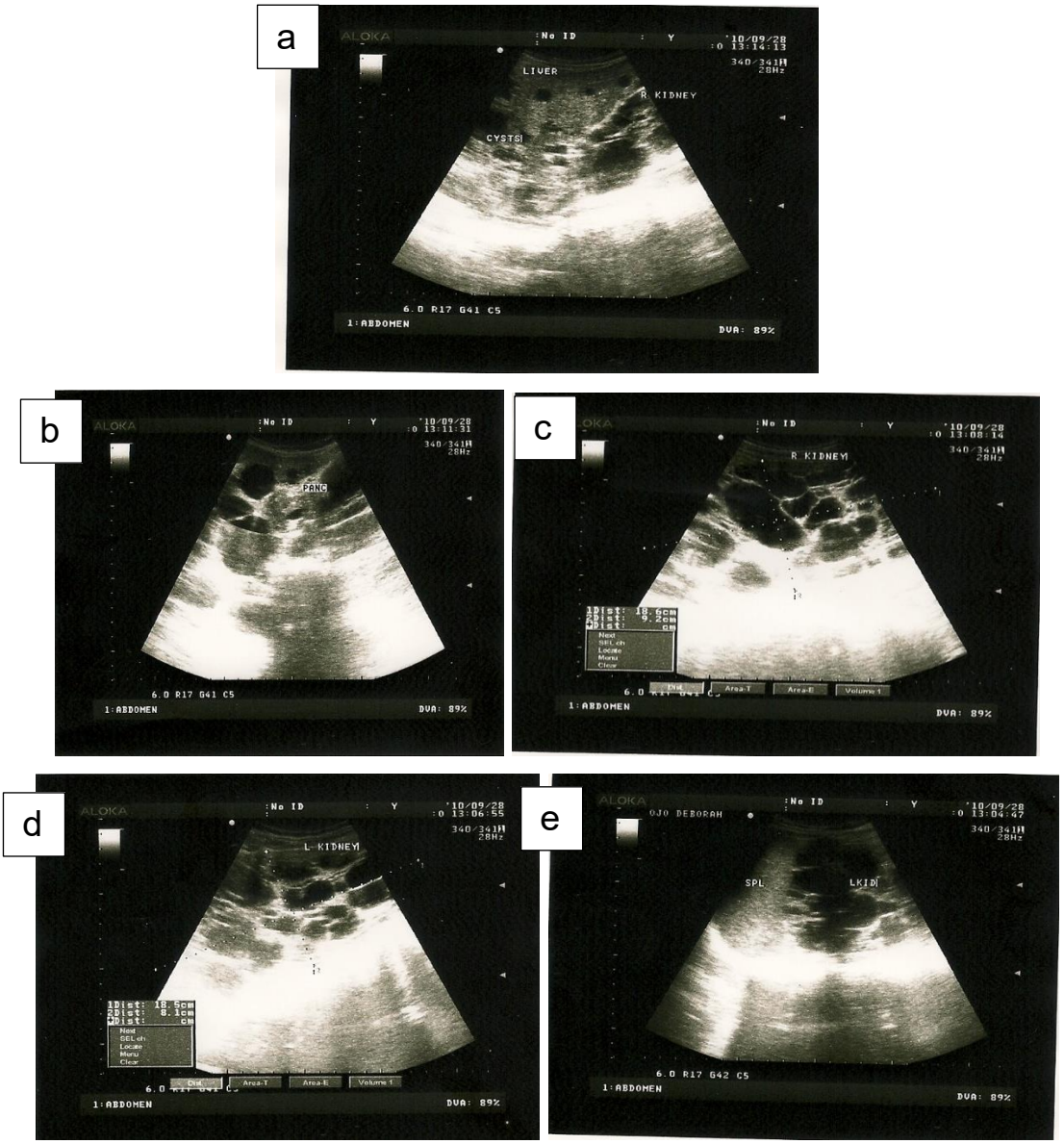

Figure 1: Ultrasound scan showing (a) multiple cysts in the liver and hepatomegaly, (b) ill-defined pancreas due to cystic involvement, (c) multiple cysts and renomegally in the right kideney, (d) multiple cysts and renomegally in left kidney, and (e) spleen that was within normal limits

Electrocardiogram revealed left atrial enlargement and left ventricular enlargement with mean QRS axis of +30°. M- mode/2-D echocardiography revealed mild pericardial effusion. The cardiac valves were normal morphologically. Left ventricle was hypertrophied with ventricular ejection fraction of 65%. There was no evidence of septal defect, and no wall motion abnormality or mural thrombus was seen. Doppler echocardiography showed that the peak aortic and pulmonary artery velocities were normal. The mitral and tricuspid E/A ratios were reversed, and there was no regurgitation across any of the valves.

The data for baseline urinalysis and blood chemistry are provided in Tables 1 and 2.

Table 1: Baseline urinalysis data for the patient

Variable	Value
Urinalysis	
Urine colour	Amber
Specific gravity	1.015
pH	6
Leucocyte	++
Protein	+
Glucose, ketone, blood, and nitrite	-
Ketone	-
Blood	-
Nitrite	-
Urobilinogen	Normal
24 hr urine protein estimation	0.32 g
Creatinine clearance	48 ml/min/1.73m^2
Estimated glomerular filtration rate (eGFR)	52 ml/min/1.73m^2

Table 2: Baseline blood chemistry data

Variable	Value
Minerals	
Sodium	142 mmol/L
Potassium	5.4 mmol/L
Urea	8.8 mmol/L
Creatinine	120 µmol/L
Urate	0.4 mmol/L
Calcium	1.95 mmol/L
Phosphate	0.5 mmol/L
Full blood count	
PCV	30%
WBC	10.6 x10^9/L
Neutrophil	84%,
Lymphocyte	16%.
Fasting serum lipid profile	
Total cholesterol	5.4 mmol/L
Triglyceride	0.8 mmol/L
Low density lipoprotein cholesterol (LDL-C)	2.8 mmol/L
High-density lipoprotein cholesterol (HDL-C)	1.0 mmol/L

The urine microscopy was unremarkable and culture yielded no growth.

A diagnosis of autosomal dominant polycycstic kidney (ADPKD) was made based on the clinical presentation and ultrasound findings using the unified criteria for ultrasonographic diagnosis of ADPKD [6]. The patient was educated about the nature of her illness and advised to encourage her first-degree relatives and children to go for sonographic screening.

She was placed on dietary phosphate, potassium and sodium restriction as well as on lisinopril (5 mg daily), bendrofluazide (5mg daily), diclofenac (50 mg twice daily for 5 days), ciprofloxacin (500 mg twice daily for 14 days), calcium gluconate (600 mg daily) and alpha-calcitriol (0.5 µg daily). She was regular on clinic visit. Her blood pressure became controlled at 110/70 mmHg by the next clinic visit and abdominal pains also resolved. However, the serum creatinine and urea concentration were still raised at 3 months follow up visit as shown in the blood chemistry (Table 3).

Table 3: Blood chemistry data at three months of treatment

Variable	Value
Sodium	134 mmol/L
Potassium	4.4 mmol/L
Urea	8.1 mmol/L
Creatinine	133 µmol/L
Urate	0.52 mmol/L
Calcium	2.29 mmol/L
Phosphate	0.77 mmol/L

None of her siblings or children showed up for screening till date despite thorough medical education and repeated emphasis at each visit.

Discussion

The age and clinical presentation of the above patient made the diagnosis of autosomal dominant polycystic kidney disease (ADPKD) most likely based on the unified criteria for ultrasonographic diagnosis of ADPKD that stipulates the presence of two or more cysts in each kidney for individuals aged 40 to 59 years [6]. There are other cystic kidney diseases with extra renal manifestations. Example include Autosomal recessive polycystic kidney disease (ARPKD), which is most often detected in-utero by fetal scan showing bilaterally enlarged echogenic kidney, oligohydramnios with or without potter phenotype [7]. Most of patients with ARPKD succumb to critical pulmonary hypoplasia at birth or develop severe morbidity and mortality from systemic hypertension, portal hypertension and renal insufficiency [7]. Beyond the neonatal period, the prognosis of patients with ARPKD is not as bleak as popularly thought [8,9].

Autosomal dominant medullary cystic kidney disease and medullary sponge kidney (MSK) are closer differential based on the age of presentation of the patient (though adolescent presentations of MSK have been reported) [10]. The diagnosis of the case illustrated was not likely to be either of these two. This is because Autosomal dominant medullary cystic kidney disease is not association with extra renal manifestations, while MSK frequently occur in association with other congenital abnormalities like; congenital hemihypertrophy, Beckwitt- wiedermann syndrome, Ehler-Danlos and Marfan syndrome [11]. In addition, cystic lesion in the liver is not characteristic

of MSK, and blood pressure is not usually elevated [7,11].

Screening of her first-degree relatives and children was crucial because ADPKD is inherited in autosomal dominant fashion with 100% penetrance though with variable expressivity [2]. These people could receive appropriate genetic counselling, risk factors for progression of chronic kidney disease can be identified and intervention instituted early if diagnosed to have ADPKD. Pre-symptomatic screening of first-degree relative is however controversial due to problems of insurability and employability. Individuals at risk should therefore be counselled on the advantages and disadvantages of screening. Gene linkage analysis is a more sensitive screening tool [7], however, this is not readily available in this environment.

Some renal manifestations of ADPKD seen in this patient include impaired renal concentrating ability leading to hyposthenuria, abdominal pains and mass, systemic hypertension, recurrent urinary tract infection and renal insufficiency. Extrarenal manifestations seen in this case were hepatomegaly due to cystic involvement of the liver, cystic lesions in the pancreas and paraaortic area. Headache may be manifestation of unruptured intracranial aneurism; however, brain magnetic resonance imaging (MRI) or computed tomography (CT) angiography was not requested for mainly because there were no clear-cut indications. These indications include family history of intracranial aneurism or subarachnoid haemorrhage, previous aneurismal rupture, preparation for elective surgery with potential hemodynamic instability, high risk occupation and anxiety on the part of the patient despite adequate education on risk. The renal and extrarenal features seen in this case were also consistent with the findings in the Darka study which reported the mean age of patients with ADPKD to be 47 ± 5 years, systemic hypertension was the most common sign (68%), and liver cyst was found in 45.5% of the patients [3]. Chijioke et al reported an overall mean age of 49.8 ± 3.6 years, and mean age for male and female of 52 ± 4.5 years and 45 ± 5.97 years respectively, with a male to female ratio of 2:1 in Ilorin, Nigeria [5]. The commonest mode of presentation in the later study was chronic renal failure, followed by hypertension and abdominal pains [5].

Management is determined by peculiarity of presentations and complications. Symptoms should be managed specifically and general measures taken to delay progression of chronic kidney disease like controlling systemic hypertension and other risk factors for progression. An extended follow up on the modification of diet in renal disease study in which ADPKD patients constituted one fourth of the study subjects showed a delay in onset of renal failure, all cost mortality and composite outcome of kidney failure in the low blood pressure group compared to usual blood pressure group [12].

The natural history of renal insufficiency is varied, but in about 50% of patients, end stage renal disease (ESRD) will occur by the age of 57 to 73 years [2]. Risk factors for progressive renal failure include, PKD1 genotype, male sex, black race, diagnosis before age 30 years, onset of haematuria before the age 30 years, onset of hypertension before age 35 years, hyperlipidaemia, low HDL cholesterol, sickle cell trait and double deletion polymorphism in angiotensin converting enzyme (ACE) gene [13]. Renal transplantation is the treatment of choice for ESRD in ADPKD [7].

Conclusion

Autosomal dominant polycystic kidney disease (ADPKD) may not be as rare as previously suggested. It is possibly under diagnosed in Africa. possibly due to refusal of at-risk individuals to present for screening among other reasons. This disposition is worth looking into. Detail evaluation of all patients with systemic hypertension and/or impaired renal function is very crucial for diagnosis and early intervention to preserve renal function.

List of abbreviations

ADPKD – Autosomal dominant polycystic kidney disease.
GFR – Glomerular filtration rate.
PCV – Packed cell volume.
WBC – White blood cell count.
LDL-C – Low density lipoprotein cholesterol.
HDL-C – High density lipoprotein cholesterol.
ARPKD – Autosomal recessive polycystic kidney disease.
MSK – Medullary sponge kidney.
ESRD – End stage renal disease.
PKD – Polycystic kidney disease.
ACE – Angiotensin-converting-enzyme.

Declarations

Ethical approval

None provided.

Availability of data and materials

The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

Competing interests

No conflict of interest associated with this work.

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Contribution of Authors

We declare that this work was done by the authors named in this article and all liabilities pertaining to claims relating to the content of this article will be borne by the authors.

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