Sonographic analysis of adult polycystic kidney disease: Retrospective data from South-East Nigeria

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Abstract

Background: Autosomal dominant polycystic kidney disease (APKD), an inheritable multisystem disease characterized by intrarenal and at times extrarenal disease, has been studied extensively among Caucasian populations. Despite the fact that being black is a risk factor for progressive disease, there is paucity of local published data. As a result, true local incidence and peculiarities in clinical and sonographic characteristics are unknown.

Aim: To present data from 19 patients diagnosed with APKD in a medium-sized facility over a 16-year period.

Materials and Methods: A retrospective search was done on the ultrasound registers for patients who had undergone abdominal ultrasound in 16 years (1997-2013). Of the 29 sonographic diagnoses of bilateral PKD made, only 19 had complete records and were included in the study. Data extracted were: age, sex, working diagnosis, renal size, diameter of renal cysts, presence or absence of extrarenal cysts, family history of renal cystic disease, blood pressure at diagnosis, and patient outcome.

Results: A total of 19 diagnoses of APKD were made- 12 males and seven females with a sex ratio of 1:0.6. Total mean age was 54.8 years (range 31–79 years)- 40.1 years for females and 57.2 years for males. In 89.5% of cases, no family history of APKD was obtained. Only six (31.6%) patients were hypertensive at presentation and three patients (16%) were already in renal failure. Ultrasound showed a mean renal size of 88.92 cm² for the right kidney and 98.97 cm² for the left. Mean cyst diameter was 3.46 cm (range 2.08-4.85 cm). Only one patient had documented extrarenal cystic disease. Two patients were lost to renal failure and congestive cardiac failure.

Conclusion: APKD appears to be uncommon in our environment; however, more studies may be elucidatory. Standard sonographic protocol for collecting data from patients with APKD is needed.

Key words: Polycystic kidneys, screening, ultrasound

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Introduction

Autosomal dominant polycystic kidney disease (APKD) is a multisystem hereditary disease typified by renal parenchymal cysts and extrarenal cystic and non-cystic changes (See Figure 1a and b). It is a monogenic disorder and has been sonologically diagnosed in utero and literature from Caucasian population studies suggests that it is a common condition. While available local literature asserts that APKD is rare in blacks, a being a black male has been a recognized risk factor for progressive disease. However, data that may contribute to the determination of its local prevalence along with possible peculiarities in its clinical and sonographic features remain sparse. Yet, the incidence of renal impairing disease worldwide (Nigeria inclusive) is high. In addition, APKD, a progressive renal disease with a relatively long course is indicted as the fourth leading cause of end stage renal disease in adults. In the course of this disease, there is morbidity from destruction of renal parenchyma in more than half of affected patients. In addition there are other extrarenal non-cystic manifestations such as intracranial

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vascular aneurysms whose rupture may lead to death or disability. Hypertension is a frequent finding in patients with APKD. Renal ultrasonic has been used in the detection of cysts APKD. These cysts appearing as thin-walled structures containing echo-clear fluid with distinct posterior enhancement progressively increase in size at variable rates. Renal insufficiency ultimately supervenes.

### Materials and Methods

A detailed search of ultrasound registers of patients who had undergone abdominal scans for various indications in the past 16 years at the ultrasound clinic of a specialist hospital in south-east Nigeria was done. Of the 33,280 patients referred for abdominal scans within the period, 29 patients with a sonographic diagnosis of bilateral polycystic kidney disease (PKD) were identified. Of these only 19 patients had their case notes traceable and were included in the study. The rest of the patients whose clinical records remained untraceable were excluded. From these records, demographic, clinical, and ultrasound scan data were extracted. The indices captured included the following: Age, sex, family history of renal cystic disease, history of hypertension and high blood pressure at diagnosis,

<table>
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<th>Table 1: The demographic, clinical and sonographic indices captured from patients’ records</th>
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APKD = Autosomal dominant polycystic kidney disease, M = Male, F = Female, BP = Blood pressure, Rt = Right, Lt = Left, CCF = Congestive Cardiac Failure, BPH = Benign Prostatic Hypertrophy, HTN = Hypertension, FHx = Family History
comorbidity/clinical presentation, the latest ultrasound measurement of renal size (length and width), range of diameter of renal parenchymal cysts, presence or absence of extrarenal cysts, and patient outcome (See Table 1).

Results

A total of 19 patients with APKD were seen out of a total of 33,280 abdominal sonographic scans. Out of 19 patients, 12 were male and seven were female with a male to female ratio of 1:0.6. Overall age range was (31-79 years). The mean age of females was 50.9 ± 6.2 years (n = 7) and that of males was 57.2 ± 4.3 years (n = 12). The difference between the mean ages was 6.3 ± 7.4, which was found to be insignificant (P = 0.4, t = 0.85, Degree of freedom = 17). The clinical indications for ultrasonography included the following: Uremia, renal failure, benign prostatic hypertrophy, seizures, male infertility, allergy, inguinal hernia, anemia, and congestive cardiac failure. A few other patients presented for routine annual abdominal scans. At diagnosis, no family history of APKD was found in the records in 17 patients (89.5% of cases).

Hypertensive heart disease was a presenting feature in six (31.6%) of the total number of patients. The average systolic blood pressure among male and female hypertensive patients was 160 mmHg. Majority of those discovered to be hypertensive (66.7%) were males with an average systolic pressure of 165 mmHg, whereas among the females, 150 mmHg was the average systolic pressure. At presentation, three patients (15.7%) had already clinical and laboratory indices of renal failure. Sonographic assessment of patients’ kidneys showed total mean renal size (length × renal width) to be 88.92 cm² for the right kidney and 98.97 cm² for the left kidney. Renal sizes for males and females were 92.2 and 83.3 cm², respectively, for the right kidney and 105.5 and 91.8 cm², respectively, for the left kidney. Mean renal cyst diameter in both sexes was 3.46 cm (range 2.08-4.85 cm). In males, cyst diameter ranged from 2.09-4.58 cm. In females, the cyst diameter ranged from 2.04-5.31 cm (See Table 2). Only one patient (5.2%) had extrarenal cystic disease. Two patients (10.5%) were lost to renal and congestive cardiac failure.

Discussion

Out of 33,280 patients referred for abdominal ultrasound scans, 29 adults were diagnosed with multiple bilateral renal parenchymal thin-walled cystic lesions consistent with polycystic kidney disease and 19 with traceable case notes eventually included in this study. In all the patients, none was clinically diagnosed, considering the known symptoms of APKD. Furthermore, nothing in the patients’ records suggested any prior patient knowledge of the renal condition. A total of 11 patients (58%) either came for routine abdominal scan with no specific complaints or had other symptoms unrelated to APKD [see Table 1]. Milutinovic et al.,[35] in their series found 36% of patients with PKD to be asymptomatic, normotensive, and without previous problems. We observed that all the patients in our study presented late. While there was no significant difference between the ages of sexes at presentation, all had sonographically detectable mature renal cysts. This is not surprising as it is known that by the age of 30 years, 68% of patients with APKD will have visible cysts by ultrasound.[36] Apart from presenting late, none of the patients in our series presented any relatives for screening. This includes two patients who admitted to a positive family history of renal cystic disease. Of these two, one patient admitted that a first degree relative had died of complications unrelated to APKD [see Table 1]. Milutinovic et al.,[35] in their series found 36% of patients with PKD to be asymptomatic, normotensive, and without previous problems. We observed that all the patients in our study presented late. While there was no significant difference between the ages of sexes at presentation, all had sonographically detectable mature renal cysts. This is not surprising as it is known that by the age of 30 years, 68% of patients with APKD will have visible cysts by ultrasound.[36] Apart from presenting late, none of the patients in our series presented any relatives for screening. This includes two patients who admitted to a positive family history of renal cystic disease. Of these two, one patient admitted that a first degree relative had died of complications from stroke. Across a spectrum of many disease conditions and social strata in the authors’ locality, there exists a pervading trend of poor health-seeking behavior fuelled by economic constraints, fear, and local taboos, leading to late presentation.[37] In 1991, however, Onuigbo et al.,[38] in one of the few family studies done in our locality found five siblings and the mother of an APKD patient to have sonologic features consistent with APKD, thus demonstrating the utility of premorbid ultrasound diagnosis.

Hypertension undoubtedly has an impact on the morbidity and mortality of patients with APKD.[22,39] The incidence of hypertension was found to differ in various series. Differences appear to be due to the benchmark used by the various workers to define hypertension.[13,35,40-42] In spite of the relatively high mean age of 54.8 years in our series, hypertension was found only in about 32%. Other workers have detected hypertension in about 60% of patients with APKD long before diagnosis of frank renal impairment.[25,26] Other APKD-related complications found in our series included congestive cardiac failure (5%), anemia (10%), and renal failure (16%). Two patient mortalities or 10.5% were recorded in our series. The causes of death were congestive cardiac failure and...
renal failure and the median age at demise was 65 years. It has long been established that the most common cause of death in patients with APKD is cardiovascular disease. In a study by Rahman et al., the most common cause of death stemmed from cardiovascular causes (46.6%) and median age at demise was 60.5 years. Intracranial aneurysms when they rupture result to subarachnoid bleed and have been described as the most devastating extrarenal complication of APKD, often resulting in premature death or disability. Meanwhile, the prevalence of unruptured intracranial aneurysms is about 8% of the APKD population and is five times higher than the general population. No record of a patient with intracranial aneurysm was found in our series. This is ostensibly because no Computed Tomography (CT) or Magnetic Resonance Imaging (MRI) of the brain was done. Again, no records requesting either of these investigations as screening were found. For this reason, there were no data of the incidence of unruptured aneurysms among our patients.

While the use of ultrasonography without doubt played a crucial role in the detection of the renal cysts in our series, thus calling attention to the presence of an erstwhile clinically occult disease, no anteroposterior (AP) renal dimensions were available from records. For this reason, renal volume could not be calculated. The importance of renal volume lies in the fact that larger kidneys, linked to progressive increase in cyst volume heralds worse prognosis in age-matched patients with APKD. The increase in renal volume has thus been found to be a good prognostic index of renal impairment, even better than estimated glomerular filtration rate (eGFR) assays.

Renal volume has been sonographically evaluated in health and disease. Studies suggest that renal volume determination using the ellipsoid method: Length (L) × width (W) × AP dimension × 0.523 is better than renal length alone as potential surrogate marker of depressed renal function. However, there exists considerable disagreement on the accuracy of two dimensional sonographically-determined renal volumes ostensibly due to claims of significant volume underestimation (as much as 25%). Still, it is agreed that sonographic estimation of renal volume has a role to play where high exactitudes are not paramount as offered by volumetric methods available with CT and MRI. Without doubt, in a resource-challenged setting such as ours that suffers from a dearth of the more profound modalities, sonographic estimation of renal volumes may continue to be useful.

Bilateral renal cysts found in all our patients exhibited marked size heterogeneity both within and between individual kidneys. While we offer no explanation for this observation, we note that other workers have made the same observations. While males had larger renal sizes for either kidneys compared with females, females appeared to have larger average cyst sizes. Therefore, cyst size alone does not explain the disparity in renal sizes between male and female patients with APKD. Gender differences may have also contributed. Sex matching is therefore important in the evaluation and interpretation of renal size and renal cyst size.

**Conclusion**

From our study it may appear that APKD is uncommon in our environment (prevalence, 0.7%); however, this may well be artifactual due to the observed local poor health-seeking behavior. It is therefore imperative that increased population and physician awareness be encouraged. Nonetheless, due to the importance of the kidneys to the body, the growing prevalence of renal-impairing disease worldwide as well as in the authors’ country must be a source of concern. Further studies are required to showcase the contribution of APKD in renal and extrarenal morbidity.

Because ultrasonography in a resource-challenged environment like ours will likely continue to play a role in the evaluation of the kidneys of APKD patients including renal volume determinations, AP renal dimensions should be routinely included to allow renal volumes to be calculated. Attending radiologists must be aware of the increased risk of intracranial aneurysms in patients with APKD. The potential devastation to patients, should rupture of these aneurysms occur, ought to galvanize more efforts toward ensuring that this extrarenal complication is screened.

Gender differences and not just cyst diameters may play a role in determining renal size in patients with APKD.

**References**


29. King BF, Reed JE, Bergstralh, EJ, Sheedy PF 2


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