Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case report

Case report: Atypical polycystic kidney disease ☆

Christopher Shin, MD, Leonard Berliner, MD*

Department of Radiology, Northwell at Staten Island University Hospital, Staten Island, NY 11794, USA

ARTICLE INFO

Article history:
Received 28 February 2021
Revised 28 March 2021
Accepted 29 March 2021

Keywords:
Autosomal dominant polycystic kidney disease
End-Stage Renal Disease
Atypical polycystic kidney disease
Unilateral renal cystic disease

ABSTRACT

Atypical or unilateral polycystic kidney disease is a rare entity that is found incidentally and is characterized on imaging as asymmetric or unilateral distribution of cysts confined to the kidneys. We present a case of an incidental finding of atypical polycystic kidney disease in a 72-year-old male. Computed tomography imaging showed asymmetric distribution of cysts only in the kidneys and the patient had no genitourinary symptoms, had normal renal function, and did not have a family history of renal disease. Although considered to be benign, rare cases of progression of atypical polycystic kidney disease to bilateral polycystic kidney disease has been documented in the literature, which portends a worse prognosis. It is important for clinicians to be aware of this entity so that patients can be monitored periodically for progression of disease.

Introduction

Renal cysts are common entities and can range from being clinically insignificant to progressing to renal failure. They may be isolated incidental findings, have a genetic component, or be part of a disease syndrome with extra-renal manifestations such as cerebral aneurysms, cardiac valvular disease, colonic diverticula, and extra-renal cysts. Autosomal dominant polycystic kidney disease (ADPKD) is the most common cystic kidney disease in adults and tends to present as innumerable cysts with symmetric involvement of both kidneys. ADPKD, classified as a ciliopathy, progresses to renal insufficiency and end-stage renal failure in 50% of cases [1]. In rare instances, ADPKD can be unilateral or be classified as a completely separate entity called atypical polycystic kidney disease or unilateral renal cystic disease. Atypical polycystic kidney disease cannot be differentiated radiographically or histologically from the typical bilateral polycystic kidney disease (PKD). It is considered to be benign, not known to have extra-renal manifestations, does not progress to renal failure, and there is no known genetic predisposition.

Case report

A 72-year-old male patient presented with rectal pain and rectal abscess that had gradually developed over a span of 4 days. The patient denied having any fevers, chills, abdominal pain, flank pain, or any other pertinent symptoms. On physical exam, the patient was found to have a draining rectal abscess, but the remainder of the examination was unremarkable. The patient's vital signs were within normal limits and on laboratory testing, he was found to have mild leukocytosis of 14K/uL

☆ Competing Interests: No conflict of interest

* Corresponding author.
E-mail address: chris.shin2019eras@gmail.com (C. Shin).

https://doi.org/10.1016/j.radcr.2021.03.068

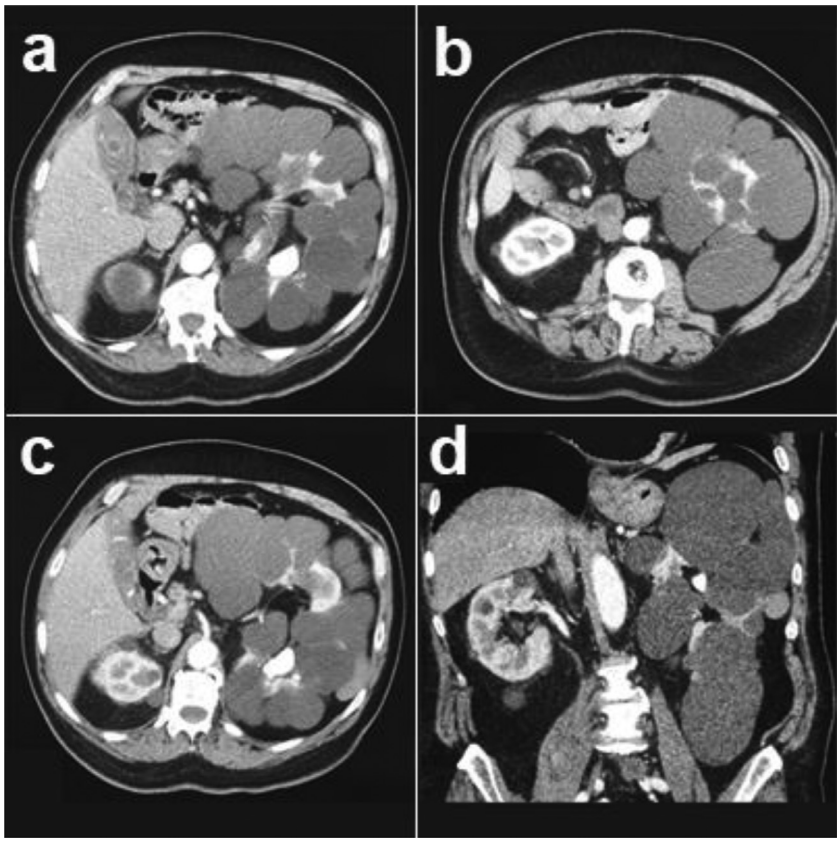

Figure 1 – Axial (A-C) and Coronal (D) CT scan with intravenous contrast demonstrates a polycystic left kidney and several simple cysts in the right kidney. Cysts in other organs were not present. Cholelithiasis is incidentally noted.

but was otherwise normal. Computed tomography (CT) of the abdomen and pelvis incidentally showed innumerable cysts of varying size in the left kidney and a few simple cysts in the right kidney (Figure 1). The right kidney measures 7.3 × 4.7 × 8 cm and the left kidney measures 15.3 × 7.7 × 12 cm. Cysts were not present in any of the other organs. Upon chart review and interview with the patient, he described having kidney stones in the past, but no other kidney related issues. He also stated there was no history of polycystic kidney disease (PKD) in his family. Furthermore, the patient had normal renal function with a creatinine of 1.2 and glomerular filtration rate (GFR) of 60. The patient was discharged and scheduled for routine outpatient follow-up.

Discussion

Atypical polycystic kidney disease (APKD) is a rare, benign condition that is indistinguishable from autosomal dominant polycystic kidney disease (ADPKD) both radiographically and histologically, but is currently recognized as a completely separate entity [2,3]. It can be differentiated from ADPKD in several ways. APKD is benign, non–progressive, does not have extra-renal manifestations, and is thought to be a non–genetic disorder whereas ADPKD is symmetric, bilateral, and monogenic [4,5]. There has been documentation of unilateral ADPKD, but is extremely rare [4,5]. Unlike ADPKD and some other polycystic kidney diseases, extra-renal cysts are not seen in APKD [6].

APKD is described as having multiple cysts involving the entire kidney or a specific region of the kidney with intervening normal parenchyma [3]. The pathogenesis is still unclear, but it has been hypothesized that it may arise from a somatic mutation involving the renal parenchyma [7,8,9].

ADPKD is now understood to be classified as a ciliopathy. In these disorders, mutations or absence of genes that alter the structure and/or function of the primary (immotile) cilia of renal (and in some cases, biliary) tubular cells result in cystic dilatation of these structures [1,10]. Renal cysts associated with ADPKD are both cortical and medullary in location, are generally simple, and will typically increase in size and number with time [10]. Specifically, in ADPKD, mutations in 2 genes, PKD1 and PKD2, are responsible for the loss of the membrane proteins, polycystin-1, and polycystin-2 respectively. The absence of these proteins, which are normally responsible for fluid regulating activity within primary renal cilia, is primarily responsible for cyst formation, along with additional local or somatic factors. Mutations in PKD1 and PKD2 are responsible for up to 85% and 15% of cases of ADPKD respectively [11], with approximately 5%-15% of cases resulting from a sporadic de novo mutation, with no identifiable family history [10].

It is postulated that these sporadic cases of PKD may result in “atypical” cases, such as the case of APKD presented

herein. Some of these cases are due to mutations in the protein biosynthetic pathway in the endoplasmic reticulum [12]. Other cases of PKD, in patients with no mutation detected, are felt to be due to somatic mosaicism. Mosaicism refers to the presence of 2 genetically distinct cell populations within 1 individual resulting from a somatic mutation during embryogenesis. These mutations may involve the germline and/or somatic (body) cells. Somatic mosaicism is felt to be responsible for cases of PKD with atypical kidney imaging patterns, including asymmetric, unilateral, and lopsided patterns [12].

Patients with APKD can present with symptoms such as hematuria or flank pain, but patients are often asymptomatic and the cysts are found incidentally as in our case. On imaging, multiple simple cysts are noted with intervening normal enhancing renal parenchyma and absence of a capsule, which help to distinguish APKD from other renal cystic disease such as cystic nephroma [13]. Typically, the cysts tend to show regional involvement with preference for the poles, and involvement of the entire kidney is rare [13]. APKD predominantly involves only 1 kidney, but there have been cases reported where the contralateral kidney also has a few cysts as in our case. Rarely, these atypical cases can evolve into bilateral disease [14].

We presented a case of APKD which is a rare entity. Despite the benign disease course based on current literature, it is an important diagnosis for clinicians to recognize. As above, since it has been reported that there have been cases of progression of atypical PKD to bilateral PKD, periodic monitoring may be of benefit to patients.

Authors' contributions

Christopher Shin – Writing of entire manuscript; Leonard Berliner – Writing of discussion of manuscript. Review and editing of entire manuscript.

Patient consent

No; formal consent was waived and review by our Institutional Review Board was deemed not necessary for a case report using de-identified medical imaging.

Human and animal rights

No experiments were performed on human or animal subjects. All treatments within the manuscript are reported retrospectively.

REFERENCES

[1] Ma M. Cilia and polycystic kidney disease. Semin Cell Dev Biol 2021;110:139–48. doi:10.1016/j.semcdb.2020.05.003.
[2] Neyaz Z, Kumar S, Lal H, Kapoor R. Localized cystic disease of the kidney: A rare entity. J Radiol Case Rep 2012;6:29–35. doi:10.3941/jrcr.v6i7.1026.
[3] Darmadi D, Ruslie RH, Siregar NQ, Theo D, Anas S. Unilateral renal cystic disease: A case report and literature review. Maced J Med Sci 2020;8(C):160–3. doi:10.3889/oamjms.2020.5031.
[4] Jeong GH, Park BS, Jeong TK, Ma SK, Yeum CH, Kim SW, et al. Unilateral autosomal dominant polycystic kidney disease with contralateral renal agenesis: a case report. J Korean Med Sci 2003;18:284–6. doi:10.3346/jkms.2003.18.2.284.
[5] Tandon A, Qureshi MS, Ahmad I, Singh UR, Bhatt S. Unilateral autosomal dominant polycystic kidney disease with co-existent renal cell carcinoma: A rare entity. Egypt J Radiol Nucl Med 2018;49(1):245–8. doi:10.1016/j.ejrnm.2017.11.009.
[6] Choh NA, Rashid M. Unilateral renal cystic disease. Indian J Nephrol 2010;20(2):116–17. doi:10.4103/0971-4065.65310.
[7] Aldan JT, Kagan I. Unilateral renal cystic disease. Proc UCLA Health 2019;23.
[8] Smyth B, Coleman P. Localized cystic disease of the kidney: case report and review of the literature. CEN Case Rep 2014;3(2):198–201. doi:10.1007/s13730-014-0117-2.
[9] Baradhi KM, Abuelo GJ. Unilateral renal cystic disease. Kidney Int 2012;81(2):220. doi:10.1038/ki.2011.343.
[10] Dillman JR, Trout AT, Smith EA, Alexander J, Towbin AJ. Hereditary renal cystic disorders: imaging of the kidneys and beyond. RadioGraphics 2017;37:924–46.
[11] Torres VE, Harris PC, Pirson Y. Autosomal dominant polycystic kidney disease. Lancet 2007;369:1287–301.
[12] Lanktree MB, Haghighi A, di Bari I, Song X, Pei Y. Insights into autosomal dominant polycystic kidney disease from genetic studies. CJASN 2021;16. doi:10.2215/CJN.02320220.
[13] Slywotzky CM, Bosniak MA. Localized cystic disease of the kidney. AJR Am J Roentgenol 2001;176(4):843–9. doi:10.2214/ajr.176.4.1760843.
[14] Hwang DY, Ahn C, Lee JG, Kim SH, Oh HY, Kim YY, et al . Unilateral renal cystic disease in adults. Nephrol Dial Transplant 1999;14(8):1999–2003. doi:10.1093/ndt/14.8.1999.