Autosomal Dominant Polycystic Kidney Disease with Extensive Cystic Degeneration and Renal Abscess but a Functioning Kidney - Management Dilemma

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Abstract

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is a genetic disorder that leads to the growth of numerous cysts in the kidneys, often resulting in kidney enlargement, progressive kidney function decline, and associated complications such as renal abscesses. When a patient presents with extensive cystic degeneration and renal abscess but still has a functioning kidney, it presents a complex management dilemma. We report the case of a middle-aged man with ADPKD having extensive cystic degeneration with renal abscess which was managed successfully with conservative management alone. We have also provided key points and approaches to be considered for managing such cases**.**

Keywords: Polycystic kidney disease, autosomal dominant, antibiotics, conservative.

Introduction

One of the most frequent causes of end-stage renal illness is autosomal dominant polycystic kidney disease (ADPKD). It is marked by the growth and development of cysts that increase the total kidney volume (TKV) and lead to a decline in renal function. It is also linked to pain, nephrolithiasis, cyst infections, hypertension, gross haematuria, and episodes of cyst hemorrhage. One in 1000 people have ADPKD [1]. While autosomal recessive polycystic kidney disease (ARPKD), a rarer illness, develops considerably earlier during the perinatal or neonatal period, most hereditary polycystic kidney disease (ADPKD) is inherited in an autosomal dominant form and usually manifests in adult life.

Case Report

A 52-year-old male patient, presented with complaints of fever with chills and rigor (two to three episodes) and dull aching discomfort in his left flank for one week. There were no aggravating or alleviating factors. Both kidneys appeared enlarged during the computed tomography (CT) assessment. Many cysts of various sizes replaced both kidneys' entire parenchyma. Only a few

of the cysts had a hyperdense appearance that suggested bleeding. The middle and lower poles of the left kidney were shown to have a poorly defined collection with air-fluid level within, suggesting the likelihood of a renal abscess. There was also an enlargement of the liver. Numerous cysts of various sizes were observed encompassing both liver lobes. Not a trace of extrahepatic or intrahepatic biliary radicle dilatation was seen. No evidence of cysts was observed in the pancreatic or seminal vesicles. Tests for liver and renal function were normal and total counts were grossly elevated (21,300). Given the normal renal function status, despite having highly elevated counts we initiated conservative treatment with analgesics, broad-spectrum antibiotics (later changed based on urine culture and sensitivity results), and intravenous fluids. The patient responded well to conservative management alone (without percutaneous/surgical drainage of collection), his total counts returned to normal limits, improved symptomatically, and was discharged.

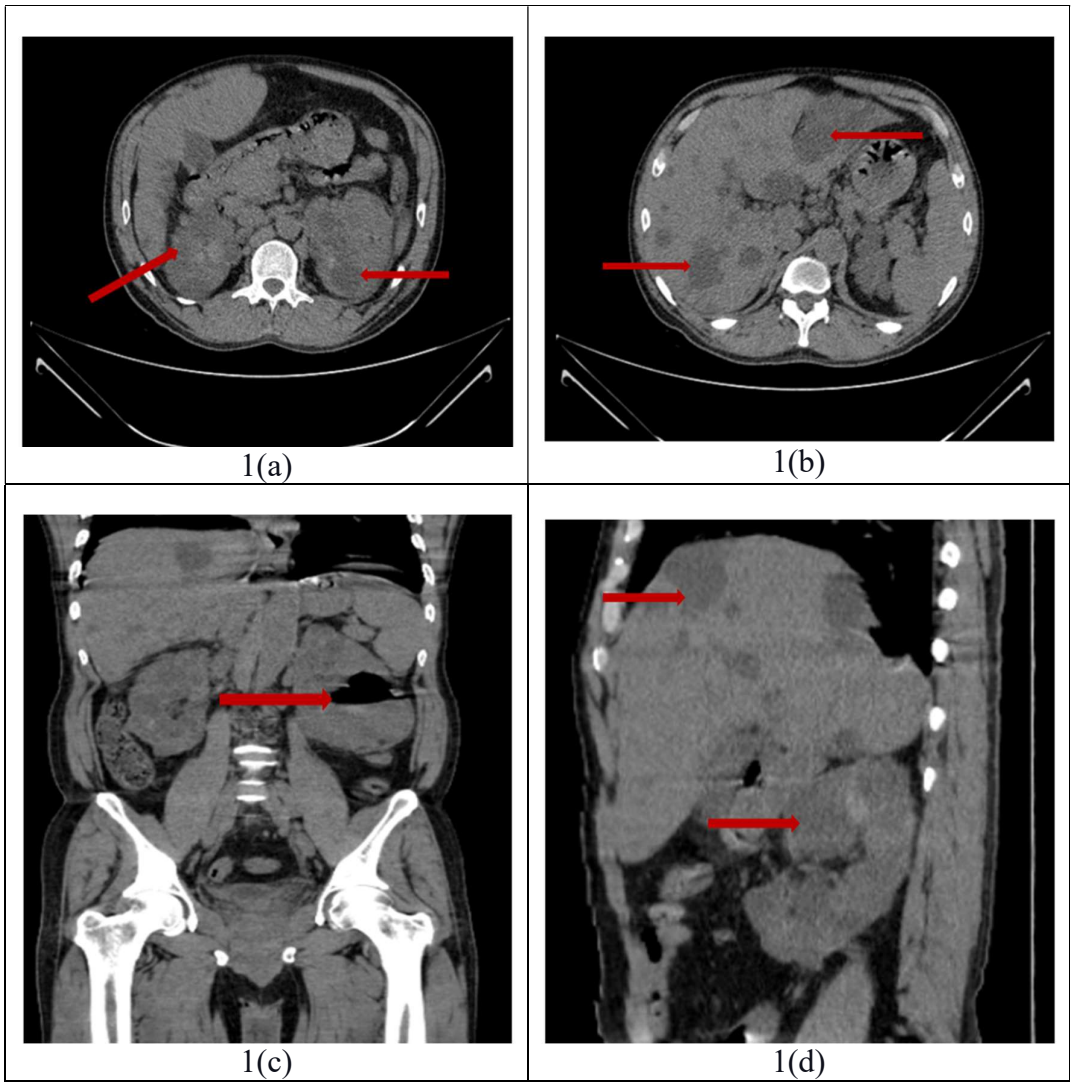

Figure 1 – Computed tomography (CT) images of the abdomen and pelvis

Figure 1(a) - Axial images show enlarged kidneys studded with multiple cysts of varying sizes *(red arrows)*, Figure 1(b) - Axial images also show multiple cysts in both lobes of the liver *(red arrows)*, Figure 1(c) - Coronal images showing air-fluid level in the mid and lower poles of the left kidney *(red arrow)*, Figure 1(d) - Sagittal images showing both hepatic and renal cysts *(red arrows)*.

Discussion

The majority of ADPKD cases (85%) are caused by mutations in the PKD1 gene, which is found on chromosome 16 and encodes polycystin-1 (PC1) [2]. A gene on chromosome 4 (4q21) called PKD2 encodes polycystin-2 (PC2), which accounts for 15% of cases [3]. PKD1 mutation carriers experience a more severe form of the disease than PKD2 carriers do, with an earlier onset of hypertension, larger total kidney volume, and an earlier onset of end-stage renal disease [4]. Most people with enlarged kidneys or low glomerular filtration rate also have hypertension. 1-2 percent of all nephrons are thought to contain cysts. As they get bigger and more numerous, they cause the proximal tubule cells to undergo apoptosis, impede urine flow, and squeeze the tubular vasculature next to them. This causes the renin-angiotensin system to be activated, which damages the renal parenchyma and causes UTIs, hypertension, and renal injury, respectively [5].

There are various ways to diagnose ADPKD, such as ultrasonography assessment and gene testing. The diagnosis of ADPKD does not frequently include genetic testing. This approach is presently utilized in patients when ADPKD needs to be ruled out completely, such as in the instance of a living kidney donor. The recommended initial method for the diagnosis of ADPKD is ultrasonography. Ravine and Demetriou developed specific diagnostic recommendations for type 1 and type 2 ADPKD, respectively. The presence of at least two (unilateral or bilateral) renal cysts, two cysts in each kidney, or at least four cysts in each kidney is diagnostic in patients aged 15 to 29, 30 to 59 years, and more than 60 years respectively according to age-dependent ultrasonography diagnostic criteria for type 1 ADPKD [6].

Less than two cysts per kidney are thought to be adequate to rule out ADPKD. Ultrasonography is not advised as a standard diagnostic method for type 2 ADPKD in patients younger than 14 years of age. Nonetheless, in those over 30 who are at 50% risk, it is 100% dependable in ruling out type 2 ADPKD [7]. Pei et al. developed an integrated strategy based on the MR criteria and positive family history [8]. The rate of total kidney volume (TKV) increase is determined by adding the volume of the freshly generated cyst to the growth of the existing cyst. If nothing is done, the rate of increase in TKV will be steady and will rise exponentially. An effective FDA-approved biomarker for predicting the rate of renal function decline and the likelihood of end-stage

renal disease is the TKV estimate [9]. When it comes to measuring volume, computed tomography is just as accurate as magnetic resonance imaging (MRI).

In patients with ADPKD older than 35 years, liver cysts are found in over 90% of cases [10]. Abdominal pain, early satiety, gastro-oesophageal reflux, and in extreme cases, portal hypertension with ascites are symptoms associated with the burden of liver cysts. Deterioration of liver synthetic function is uncommon. In females, the frequency of hepatic cysts is higher, and their size is more significant [11]. In female patients with severe polycystic liver disease, the total liver volume tends to decline after the age of 48 [12]. Liver transplantation or partial hepatectomy is necessary for certain patients. In patients with ADPKD, intracranial aneurysms are more common. Hepatic cysts and symptomatic berry aneurysms are more common in patients with diverticulosis [13]. Patients with high-risk jobs, a family history of cerebral aneurysms, or aneurysmal rupture are screened for intracranial aneurysms using magnetic resonance angiography [14]. Additional extra-renal symptoms include the formation of pancreatic, ovarian, and seminal vesicle cysts, as well as heart conditions that are rarely symptomatic and do not warrant screening. The lack of renal hypertrophy, extra-renal features, and familial history of cystic illness typically differentiate acquired cystic kidney disease from ADPKD.

The consumption of large amounts of water has been suggested as a therapy for ADPKD due to its ability to inhibit vasopressin activity. However, water therapy trials have yielded conflicting results [15]. Tolvaptan, a highly specific vasopressin antagonist, is a novel medication that the FDA has approved to "slow down the kidney function decline in adults at risk of rapidly progressing ADPKD" [16]. Urine retention, cyst cell proliferation, and fluid production are all present in ADPKD patients. This is due to the synthesis of cyclic-AMP in the distal nephrons and renal collecting ducts by arginine vasopressin. Tolvaptan causes a decrease in cyst cell proliferation by increasing water secretion and decreasing urine osmolality [17].

Severe bleeding, persistent and severe cyst infection, unbearable pain, suspicion of kidney cancer, and space constraints before transplantation are among the indications for a nephrectomy. Other techniques that have been employed include embolization, cyst fenestration, and cyst aspiration followed by foam sclerotherapy.

Decision-making Dilemma

Managing autosomal dominant polycystic kidney disease (ADPKD) with a renal abscess presents a unique clinical dilemma due to the combination of

cystic kidney disease and the risk of infection, which can complicate both the management of ADPKD and the treatment of the abscess.

Key Issues in Management:

1. Complexity of ADPKD:
- Cystic formation in ADPKD leads to progressive kidney enlargement and can distort normal kidney anatomy. This increases the risk of infection within cysts or in the parenchyma of the kidney.
- Renal failure in ADPKD patients is common, especially as the disease progresses, making it harder to manage infections with reduced renal function.

2. Renal Abscess in ADPKD:
- A renal abscess occurs when bacteria infect the renal parenchyma, and in ADPKD, the presence of cysts can complicate the infection's pathophysiology.
- The abscess can be either in the cysts or in the kidney parenchyma.
- The risk of developing a renal abscess in ADPKD increases due to urinary stasis, cyst rupture, and compromised immune function.

3. Differential Diagnosis:
- It is important to distinguish between a renal abscess and other complications of ADPKD, such as cyst hemorrhage or renal infarction.
- Imaging studies (CT, ultrasound, or MRI) are critical in diagnosing a renal abscess and determining the extent of the infection.

Management Approach:

1. Initial Assessment:
- Clinical Evaluation: Assess the patient for fever, flank pain, tenderness, dysuria, and other signs of infection.
- Laboratory Tests: Blood cultures and urine cultures should be obtained. The urine may show pyuria and bacteriuria if the infection is in the urinary tract.
- Imaging: A CT scan or ultrasound is crucial for identifying the location and size of the abscess and differentiating it from other complications (e.g., ruptured cysts). In cases of complex cysts, MRI may be used for a detailed assessment.

2. Antibiotic Therapy (First line):
- Empirical Antibiotics: Start broad-spectrum intravenous antibiotics immediately. Common pathogens in renal abscesses include *E. coli*, *Klebsiella*, *Proteus*, and other gram-negative organisms. Coverage for

methicillin-resistant *Staphylococcus aureus* (MRSA) should also be considered, especially in immunocompromised or hospitalized patients.

- Tailored Antibiotics: Once culture and sensitivity results are available, adjust the antibiotic therapy accordingly.
- Duration: The typical antibiotic course for renal abscesses is 4-6 weeks, depending on the size of the abscess and clinical response.

3. Drainage of the Abscess:
   - Percutaneous Drainage: In cases where conservative management fails, percutaneous drainage under imaging guidance (ultrasound or CT) is the preferred option in ADPKD patients.
   - Surgical Drainage: In cases where percutaneous drainage is unsuccessful or if the abscess is large and not accessible, surgical drainage (nephrectomy or nephrostomy) may be necessary.

4. Supportive Care:
   - Renal Function Monitoring: Regularly monitor kidney function (serum creatinine, electrolytes) as ADPKD patients are at high risk for renal failure.
   - Hydration and Electrolyte Management: Ensure the patient is adequately hydrated and maintains fluid and electrolyte balance, especially if they are undergoing dialysis or have impaired renal function.

5. Consideration for ADPKD-Specific Complications:
   - Preventing Cyst Rupture: In ADPKD, any intervention in the kidney, especially drainage, should be done carefully to prevent rupture of cysts, which can worsen infection or lead to bleeding.
   - Dialysis and Renal Replacement Therapy: In severe cases of renal failure, dialysis or even a kidney transplant may be needed, depending on the stage of ADPKD.

Special Considerations:

- Immunocompromised State: If the patient is immunocompromised (due to dialysis, immunosuppressive therapy for transplantation, etc.), the infection may be more aggressive, and longer courses of antibiotics or more intensive management may be required.
- Recurrent Abscesses: If abscesses recur or are difficult to treat, this may signal advanced ADPKD or an underlying problem with immune function, necessitating further investigation and possible changes to the treatment plan.

Conclusion

The management of a renal abscess in a patient with ADPKD involves prompt diagnosis with imaging, initial management with broad-spectrum antibiotics if the patient is clinically stable along with a normal renal function, considering percutaneous or surgical drainage if conservative therapy fails or in case of a large multiloculated abscess and nephrectomy as the last resort when all other modalities fail. It is important to preserve as much as nephrons as possible, especially in a functioning kidney. The decision-making process depends on the patient's clinical status and abscess characteristics. Close coordination between urologists, nephrologists, and radiologists is essential to optimize outcomes.

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