Case Report

Autosomal dominant polycystic kidney disease: A case report

Jean V. Varghese[1], Antriya Annie Tom[2]

From [1]PharmD Intern, Department of PharmD, [2]Associate Professor, Department of Pharmacy Practice, Nirmala College of Pharmacy, Muvattupuzha, Kerala, India

ABSTRACT

Autosomal dominant polycystic kidney disease (ADPKD) is an inherited condition that causes small fluid-filled sacs called cysts to develop in the kidneys. ADPKD affects 1 in 500–1000 people. We report the case of a 70-year-old male patient with complaints of difficulty in passing urine, lower abdominal pain, fever with chills, and hematuria. The ultrasonography report of the abdomen and pelvis showed bilaterally mildly enlarged kidneys with multiple variable simple cysts slightly distorting the normal renal architecture and mildly increased residual visualized cortical echoes concerning adult polycystic kidney changes. The therapeutic regimen was targeted to treat the symptoms of the patient. In India, there is a need for epidemiological studies on ADPKD since insignificant data are present that could rationally reflect its prevalence and incidence, for which this case report could be utilized.

Key words: Autosomal dominant polycystic kidney disease, Tolvaptan, Urinary tract infection

Autosomal dominant polycystic kidney disease (ADPKD) is an inherited condition that causes small fluid-filled sacs called cysts to develop in the kidneys [1]. They are classified as rare diseases affecting fewer than 200,000 people [2]. ADPKD has two subdivisions: ADPKD1 and ADPKD2. ADPKD affects 1 in 500–1000 people, with the autosomal recessive type in an estimated 1 in 20,000–40,000 people [3]. According to the European Renal Association - European Dialysis and Transplant Association, the application of a uniform definition of prevalence to population-based studies consistently indicate that the ADPKD point prevalence is <5/10,000, the threshold for rare disease in the emergency unit [4]. A single study from India recently reported ADPKD to be the etiology of chronic kidney disease (CKD) in 2.6% of cases [4]. Symptoms caused by cyst formation in the kidneys include high blood pressure (hypertension), pain on the sides of the body between the last rib and the hip (flank pain), blood in the urine (hematuria), and progressively poor functioning of the kidneys (renal insufficiency) [1]. ADPKD can be diagnosed using ultrasound, computed tomography scan, or magnetic resonance imaging studies. A genetic test can detect mutations in the PKD1 and PKD2 genes, the genes that, when altered, cause ADPKD.

Here, we present the case of a 70-year-old male patient with ADPKD. In India, there is a need for epidemiological studies for ADPKD as insignificant data are present that could rationally reflect its prevalence and incidence [5]. This case report can be useful for the epidemiological studies of ADPKD in India.

Access this article online	
Received - 19 June 2021 Initial Review - 06 July 2021 Accepted - 17 July 2021	Quick Response code
DOI: 10.32677/IJCR.2021.v07.i08.2965	

CASE REPORT

A 70-year-old male patient was brought to the hospital with complaints of difficulty in passing urine, lower abdominal pain, fever with chills, and hematuria for 2 days. The patient was not reported to have any medical history. This case report is presented using CARE guidelines [6].

On admission, the temperature of the patient was found to be 100° F. His blood pressure, pulse, and respiratory rate were found to be normal. Significant changes in the laboratory investigations were found in hemoglobin (9 g%), erythrocyte sedimentation rate (51 mm/h), and lymphocyte (17%). Serum calcium was found to be 7.2 mg/dl. On urine analysis, trace amounts of albumin, pus cells (4–6), red blood cells (2–4), and yeast cells (few) were found. Scanty growth of a non-pathogenic organism was found in the urine culture. Renal function test was done which showed urea - 45 mg/dl, creatinine - 1.3 mg/dl, sodium - 136 meq/l, potassium - 4.7 mmol/l, uric acid - 4.4 mg/dl, and creatinine clearance- 31 ml/min.

The ultrasound sonography test report of the abdomen and pelvis done on the day of admission showed bilaterally mildly enlarged kidneys with multiple variable simple cysts slightly distorting the normal renal architecture and mildly increased residual visualized cortical echoes concerning adult polycystic

Correspondence to: Mrs. Antriya Annie Tom, Highest Academic Degree: M. Pharm, Nirmala College of Pharmacy, Muvattupuzha, Ernakulam, Kerala, India. E-mail: annietomantriya@gmail.com

kidney changes (renal function test correlation). The urinary bladder wall was thickened (13 mm) and enlarged prostate volume was found (32cc). Using the subjective and objective assessment, the patient was diagnosed with ADPKD along with urinary tract infection and benign prostatic hypertrophy.

The medications administered were Inj. Paracetamol 650 mg IV, inj. tranexamic acid 1amp (100 mg) q8h, tab. Rabeprazole 20 mg bd, tab. Ciprofloxacin 500 mg bd, and tab. Tamsulosin 0.4 mg h of sleep (HS). The patient was discharged with tab. Silofast 8 mg bd, tab. Etizolam 0.25 mg HS, tab. Cifran 500 mg bd for 5 days, tab. Rabium 20 mg bd, and tab. Maxical 500 mg hs. The patient became symptomatically better and was discharged on the 4th day.

DISCUSSION

Approximately 30–50% of patients with ADPKD have a urinary tract infection (UTI) during their lifetime [7]. The infections are typically caused by Gram-negative enteric organisms [8]. In this case, the patient was prescribed medications to relieve the symptoms.

ADPKD is a progressive disorder and if left untreated, may lead to renal failure and worsening of symptoms. Patients with ADPKD are at increased risk for hypertension, cardiovascular events, and cardiovascular mortality when compared to the general population [9]. The diagnostic criteria for individuals have a 50% risk of developing ADPKD include: At least two unilateral or bilateral cysts in individuals who are younger than age 30, at least two cysts in each kidney in individuals who are between 30 and 59 years, and at least four cysts in each kidney in individuals who are 60 years old or older [3].

In patients with ADPKD, pharmacologic therapy is necessary to control blood pressure, treat urinary tract infections, treat hematuria, and reduce abdominal pain produced by enlarged kidneys, and slow the decline of kidney function [10]. In the past, treatment of ADPKD was limited to the management of symptoms and their complications. But recently, Food and Drug Administration (FDA) has approved tolvaptan as the first drug to slow kidney function decline in adults who are at risk of rapidly progressing ADPKD [11]. As per the National Institute for Health and Clinical Excellence guidelines, tolvaptan is recommended as an option for treating ADPKD in adults to slow the progression of cyst development and renal insufficiency only if (i) they have CKD stages 2 or 3 at the start of treatment, and (ii) there is evidence of rapidly progressing [12].

Tolvaptan is a selective vasopressin antagonist. By inhibiting the binding of vasopressin to the V2 receptors, tolvaptan reduces cell proliferation, cyst formation, and fluid excretion which reduces kidney growth and protects kidney function [12]. A recent study showed that initiating or maintaining tolvaptan therapy significantly delayed glomerular filtration rate (GFR) decline in subjects with baseline eGFR of 15–24 and in subjects with baseline eGFR of 25–29 ml/min/1.73 m^2 [13]. Tolvaptan could be prescribed to this patient in order to slow kidney function decline. Tolvaptan should be given at 45 mg postoperative initially on waking and 15 mg taken 8 h later (i.e. 60 mg.day) [14]. A study also showed that long-term administration of tolvaptan at a high dose is both safe and effective to reserve kidney function, though a gradual increase in total kidney volume was seen, particularly during the later phase [15].

CONCLUSION

ADPKD is a multisystemic and progressive disorder characterized by cyst formation and enlargement in the kidneys. As a result of this, patients experience urinary hesitancy, lower abdominal pain, fever with chills, and hematuria. ADPKD therapy is mainly targeted at managing symptoms and reducing complications. However, ADPKD being a progressive disease, and if left untreated, can lead to renal failure. FDA has approved tolvaptan as the first drug to slow kidney function decline. In India, there is a need for epidemiological studies for ADPKD as insignificant data are present that could rationally reflect its prevalence and incidence. This case report can be useful for the epidemiological studies of ADPKD in India.

REFERENCES

1. Autosomal Dominant Polycystic Kidney Disease; 2021. Available from: https://www.nhs.uk/conditions/autosomal-dominant-polycystic-kidney-disease-adpkd. [Last accessed on 2021 Jun 15].
2. Willey CJ, Blais JD, Hall AK, Krasa HB, Makin AJ, Czerwiec FS. Prevalence of autosomal dominant polycystic kidney disease in the European Union. Nephrol Dial Transplant 2017;32:1356-63.
3. Polycystic Kidney Disease: From Genetics Home Reference; 2020. Available from: https://www.medlineplus.gov/genetics/condition/polycystic-kidney-disease/#causes. [Last accessed on 2021 Jun 06].
4. Rajapurkar MM, John GT, Kirpalani AL, Abraham G, Agarwal SK, Almeida AF, *et al*. What do we know about chronic kidney disease in India: First report of the Indian CKD registry. BMC Nephrol 2012;13:10.
5. CARE. What Are the CARE Guidelines? 2021. Available from: https://www.care-statement.org. [Last accessed on 2021 May 15].
6. Autosomal Dominant Polycystic Kidney Disease (ADPKD): Evaluation and Management of Urinary Tract Infections; 2020. Available from: https://www.uptodate.com/contents/autosomal-dominant-polycystic-kidney-disease-adpkd-evaluation-and-management-of-urinary-tract-infections#h1. [Last accessed on 2021 Jun 23].
7. Idrizi A, Barbullushi M, Koroshi A, Dibra M, Bolleku E, Bajrami V, *et al*. Urinary tract infections in polycystic kidney disease. Med Arh 2011;65:213-5.
8. Schrier RW. Hypertension and autosomal dominant polycystic kidney disease. Am J Kidney Dis 2011;57:811-3.
9. Polycystic Kidney Disease Treatment and Management; 2020. Available from: https://www.emedicine.medscape.com/article/244907-treatment#d1. [Last accessed on 2021 May 26].
10. Chebib FT, Perrone RD, Chapman AB, Dahl NK, Harris PC, Mrug M, *et al*. A practical guide for treatment of rapidly progressive ADPKD with Tolvaptan. J Am Soc Nephrol 2018;29:2458-70.
11. Torres VE, Gansevoort RT, Perrone RD, Chapman AB, Ouyang J, Lee J, *et al*. Tolvaptan in ADPKD patients with very low kidney function. Kidney Int Rep 2021:1-8.
12. NICE Guidance. Tolvaptan for Treating Autosomal Dominant Polycystic Kidney Disease: Evidence-Based Recommendations on Tolvaptan for Treating Autosomal Dominant Polycystic Kidney Disease in Adults; 2021. Available from: https://www.nice.org.uk/guidance/ta358. [Last accessed on 2021 Jun 25].
13. Tolvaptan (Rx); 2021. Available from: https://www.reference.medscape.com/drug/samsca-jynarque-tolvaptan-999103. [Last accessed on 2021 May 02].

14. Nakatani S, Ishimura E, Machiba Y, Fujimoto K, Uedono H, Tsuda A, *et al*. Long-term effects of high-dose tolvaptan for autosomal dominant polycystic kidney disease patients. Case Rep Nephrol Dial 2020;10:9-17.
15. Saini AK, Saini R, Singh S. Autosomal dominant polycystic kidney disease and pioglitazone for its therapy: A comprehensive review with an emphasis on the molecular pathogenesis and pharmacological aspects. Mol Med 2020;26:128.

Funding: None; Conflicts of Interest: None Stated.

How to cite this article: Varghese JV, Tom AA. Autosomal dominant polycystic kidney disease: A case report. Indian J Case Reports. 2021;7(8):339-341.