GOIANA JOURNAL OF MEDICINE

DOI: 10.29327/2396527.65.65-7

ISSN 0034-9585

SCIENTIFIC ARTICLE - CASE REPORT

# AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE: CASE REPORT

NATHALIE BORGES COSTA[1]; LUÍS OTÁVIO TORRES[1]; SUMAYA VIEIRA CANEDO PRUDENTE[1]; CAMILLA CHAGAS MOREIRA ALVES[1]; LAYS RABELO RATES[1]; CIBELLE CAMILO BARBOSA JARDIM[1]; AFONSO LUCAS OLIVEIRA NASCIMENTO

1 – Centro Universitário Alfredo Nasser (UNIFAN) - Goiânia GO

**ABSTRACT**

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is characterized by the progressive development of numerous bilateral renal cysts that encroach upon the functional parenchyma, culminating in renal function loss and Chronic Kidney Disease. Additionally, there are extrarenal involvements, meaning cysts arise in different bodily systems. The objective is to investigate ADPKD in a patient, aiming, through a case report, to explore updates on diagnosis, treatment, and the epidemiological situation of the disease in the country. This case report described a 65-year-old patient complaining of lower back pain associated with hypertension and subsequent diagnosis of Autosomal Dominant Polycystic Kidney Disease. The diagnosis of ADPKD is based on imaging exams and the patient's family history. Treatment for ADPKD, in turn, is related to renal and extrarenal complications of the disease, aimed at limiting morbidity and mortality. The search for family history indicating the manifestation of characteristic symptoms and signs of the disease is of utmost importance as it is a genetically inherited condition, and the earlier the diagnosis, the better the quality of life associated with its carriers.

**Keywords:** Autosomal dominant polycystic kidney disease; bilateral renal cysts; chronic kidney disease.

## INTRODUCTION

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is defined by the progressive development of numerous bilateral renal cysts that encroach upon the functional parenchyma, culminating in kidney function loss and Chronic Kidney Disease (CKD)[1]. Additionally, there are extrarenal involvements, meaning cysts arise in different body systems. In this context, it is important to emphasize that ADPKD is the most common hereditary renal pathology in humans, presenting genetic heterogeneity, i.e., they are diseases that manifest with similar phenotypic traits but can be caused by mutations in different genes[2].

Understanding the disease involves understanding, according to the literature, that ADPKD is present from intrauterine life, clinically manifesting in adulthood. Furthermore, in the majority of cases, this disease is asymptomatic, which explains the fact that only 50% of carriers are diagnosed. In this analysis, the confirmation of the clinical condition occurs only between the third and fourth decades of life through routine exams or genetic analysis[3].

Among the clinical manifestations related to ADPKD, Systemic Arterial Hypertension (SAH) is present in 66.7% of patients, being the most prevalent associated disease, which is related to the increased renal

debility that precedes kidney function loss and establishes a factor in CKD progression[1,3]. In this context, other manifestations are noted, such as nephrolithiasis, renal failure, portal hypertension, gastrointestinal bleeding, esophageal varices rupture, thrombocytopenia, splenomegaly, cholangitis, and jaundice[4].

The present study aims to investigate ADPKD in a patient, seeking, through a case report, to explore updates on diagnosis, treatment, and the epidemiological situation of the disease in the country.

## CASE REPORT

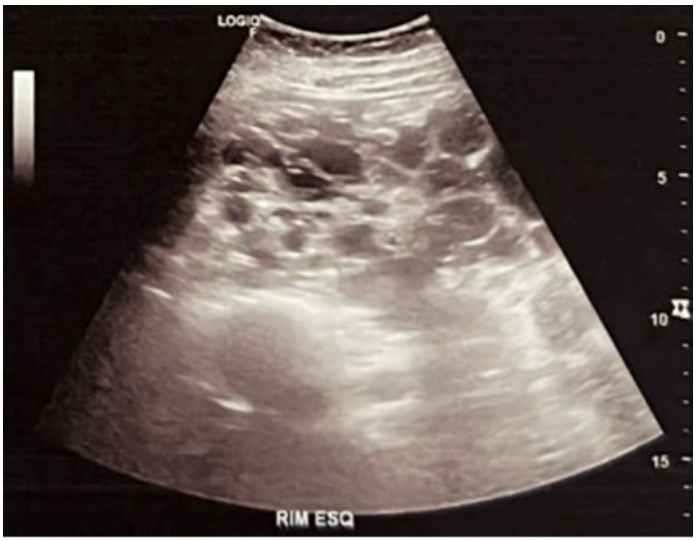
Patient DPCM, female, 65 years old, attended the nephrologist's office with complaints of low back pain associated with hypertension. With a family history of kidney disease, her mother died due to an unspecified nephropathy, and her siblings are carriers of ADPKD.

The patient denied complaints such as recurrent cystitis, hematuria, and urinary changes, which are commonly found in this disease. On physical examination, there were no changes on palpation of the abdomen. She presented Giordano's sign, which indicates inflammation of the renal parenchyma upon percussion.

Laboratory tests revealed creatinine 4.7 mg/dL, urea 129 mg/dL, hemogram showing hemoglobin = 10.8 g/dL, hematocrit = 32.5%, 7,000 leukocytes, 263,000 platelets /mm3, aspartate aminotransferase 15 U/L, and alanine aminotransferase 17 U/L.

She underwent an abdominal ultrasound which revealed both kidneys to be enlarged, consisting of several medium-sized cysts. The image of the left kidney is presented below (the appearance is similar to that of the right kidney) (Figure 1).

**Figure 1: Ultrasound image of the left kidney**




Treated clinically with atenolol, chlorthalidone, furosemide, losartan, amlodipine. Currently maintains renal function and partially controlled hypertension. The nephrology service evaluation opted for the creation of an Arteriovenous Fistula (AVF) in the right arm, anticipating a future need for hemodialysis.

## DEVELOPMENT

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is a hereditary disease, classified as a multisystem monogenic disorder, and presents genetic heterogeneity, being the most common among humans. It originates from the growth and development of multiple bilateral renal cysts that destroy the functional parenchyma, as well as from extrarenal manifestations, evidencing cysts in other organs, cardiac valvular abnormalities, cerebral aneurysms, abdominal hernias, body pains, calculous cholecystopathy, and diverticular disease[1].

The prevalence of ADPKD is reported to be 1 in 400 to 1 in 1000 live births, in studies in Denmark and the United States. Based on this prevalence, it is presumed that more than 10 million people worldwide, considering all ethnic groups, have ADPKD, thus constituting the greatest public health problem[5].

In patients undergoing dialysis treatment, ADPKD affects between 13.4% and 5% of patients in the United States and Europe, respectively. In Brazil, on the other hand, the frequency in dialysis patients varies from 3% to 10.3%. Furthermore, regarding prevalence, there is only one study conducted in the national territory that analyzed the existence of 9.1 cases per 100,000 inhabitants in the northwest region of the state of Paraná[6]. Additionally, it is important to note that prevalence studies in autopsies suggest much higher numbers of individuals identified with the disease. This information suggests the existence of ineffective diagnoses[5].

When it comes to factors that entail a high risk for ADPKD patients, the male gender is a determining factor in the disease's worse progression. As a result, men require kidney transplantation at an earlier stage due to this and other gender-related risk factors, such as diagnosis before the age of 30, increased kidney size, and hormonal influence[4].

Regarding the pathology of the disease, it is related to the dysfunction of two genes: PKD1 and PKD27. These genes are responsible for encoding, respectively, the proteins Polycystin 1 (PC-1) and Polycystin 2 (PC-2), which are proteins related to cell multiplication and differentiation and substance transport. Patients with mutations in PKD1 progress more rapidly to Chronic Kidney Disease (CKD) stage five and have a larger kidney size compared to individuals with mutations in PKD2, who form fewer cysts[6].

In the context of genetic mutations, gene dysfunctions lead to erroneous production of proteins related to the genes. In this sense, the regulatory function of PC-1 on the activity of PC-2 does not occur, which causes problems in intracellular Ca++ concentrations, and the product of PC-1 cleavage, important for maintaining the integrity of distal nephrons, is not formed. Furthermore, genetic dysfunction influences the movement of chloride through Na+-K+-2Cl- cotransporters positioned in the basolateral membrane of the cystic epithelium, contributing to cystic expansion[8].

As a multisystemic disease, ADPKD presents with both renal and extrarenal manifestations in clinical cases. Firstly, renal manifestations include defects in urine concentration, reduced renal blood flow, hypertension -and consequent target organ damage-, hemorrhagic cyst, among other conditions. On the other hand, extrarenal manifestations include polycystic liver disease, intracranial aneurysm, vascular abnormalities such as thoracic aortic dissection, valvular heart disease, and cyst formation in various organs such as the pancreas and arachnoid membrane[7].

Regarding the clinical aspect, it is also important to highlight that hypertension is the main sign that deserves attention for the suspicion of ADPKD, and treatment is indicated in 70% of cases. Furthermore, gastrointestinal bleeding, esophageal varices rupture, thrombocytopenia, splenomegaly, jaundice, and cholangitis are highlighted as indicators of the disease[4].

The diagnosis of ADPKD is based on imaging exams and the patient's family history. For imaging exams, an Ultrasonography (USG), Computed Tomography (CT), or Magnetic Resonance Imaging (MRI) of the kidneys can be performed, in which multiple cysts may be observed, increasing in number with age. It is important to highlight that USG is the first choice due to its low cost, while MRI is preferred for quantifying renal volume[7].

Molecular diagnosis is also of great value in some cases where there is suspicion of ADPKD and cannot be evaluated by conventional methods. For this, gene linkage analysis, direct gene testing, and DNA sequencing are performed[7].

Treatment for ADPKD is related to renal and extrarenal complications of the disease, aimed at limiting morbidity and mortality. The main situations are related to flank pain, with tricyclic antidepressants, analgesics, and opioids being used for proper management. Additionally, in cases where there is a decrease in hematocrit and hemodynamic instability related to severe complications of hemorrhagic cysts, hospitalization and blood transfusion are opted for, while in mild cases caused by these cysts, only rest, analgesics, and fluid intake are recommended. On the other hand, kidney transplantation is a definitive option for ADPKD in patients who meet the criteria for this treatment[9].

Hypertension, the main sign associated with ADPKD, deserves attention regarding therapeutic management related to the disease. In this situation, healthcare professionals use antihypertensive drugs such as angiotensin-converting enzyme inhibitors (ACE inhibitors) or angiotensin receptor blockers (ARBs) to prevent the progression of renal damage, which is already so prominent due to the progression of ADPKD. It is also important to note the existence of infectious cystic conditions related to urinary tract infection, which are treated with antibiotics[7].

In the analysis of therapeutic approaches related to ADPKD, the progression of understanding the genetic and molecular bases involved in the pathogenesis of the disease, such as the mechanisms of cyst development and growth, has enabled the use of substances in clinical trials. In this context, molecules such as the mammalian target of rapamycin inhibitors (mTOR) stand out. This molecule has been shown to decrease the number of cysts in animals that use it compared to the control group. Furthermore, V2 receptor antagonists related to vasopressin have been shown to be effective in reducing the frequency of situations related to ADPKD, such as renal pain, urinary tract infection, and hematuria[9].

## CONCLUSION

ADPKD is a condition with different nuances, as it goes beyond renal dysfunction as a consequence. In this context, the complexity of the mechanisms that generate the disease at the renal level stands out, which are related to cyst formation and, subsequently, organ dysfunction.

In this scenario, the search for family history indicating the manifestation of characteristic symptoms and signs of the disease is of paramount importance because it is a genetic disorder. On the other hand, early screening, mainly through ultrasound, is essential to improve the quality of life of ADPKD carriers early on.

## REFERENCES

1. Alves EF, Tsuneto LT, Borelli SD, Cadidé RC, de Freitas RA, Gravena AAF, et al. Características sociodemográficas e aspectos clínicos de pacientes com doença renal policística do adulto submetidos à hemodiálise. Sci Med. 2013 Out 19;23(3):156-62.

2.Alves EF, Tsuneto LT, Pelloso SM, Torres PRA, Otto GLG, Silva AA, et al. Doença renal policística autossômica dominante em pacientes em hemodiálise no sul do Brasil. J Bras Nefrol. 2014 Jan-Mar;36(1):18-25.

3.Bittencourt ZZLC. Epidemiologia da doença renal policística em pacientes renais crônicos no município de Campinas. Serv Soc Saúde. 2015 May 4;1(1):121-52.

4.Saraiva LM, Monteiro MO, Oliveira SMA, Chaves KRM. Doença renal policística: relato de caso e revisão de literatura. Residência Pediátrica. 2016;6(3):152-4.

5.Bergmann C, Guay-Woodford LM, Harris PC, Horie S, Peters DJM, Torres VE. Polycystic kidney disease. Nat Rev Dis Primers. 2018 Dec 6;4:50.

6.Alves EF, Borelli SD, Tsuneto LT. Doença renal policística autossômica dominante: uma atualização sobre aspectos moleculares e edpidemiológicos. Rev USP. 2015 Dec 11;48(4):380-5.

7.Jhonson RJ, Feehally J, Floege J. Nefrologia Clínica. 5th ed.: Elsevier Editora Ltda, 2015.

8.Riella MC. Princípios da nefrologia e distúrbios hidreletrolíticos. 6th ed. Rio de Janeiro: Guanabara Koogan Ltda, 2018.

9.Bissler JJ. Therapies for polyystic kidney disease. Curr Opin Pediatr. 2015 Apr;27(2):227-32

NATHALIE BORGES COSTA - http://lattes.cnpq.br/4301588567236901 - https://orcid.org/0000-0003-4450-8650

LUÍS OTÁVIO TORRES - http://lattes.cnpq.br/6820759397258447 - https://orcid.org/0009-0007-7397-5131

SUMAYA VIEIRA CANEDO PRUDENTE - http://lattes.cnpq.br/9399977117244518 - https://orcid.org/0009-0007-3376-0515

CAMILLA CHAGAS MOREIRA ALVES - http://lattes.cnpq.br/4588693706708632 - https://orcid.org/0009-0004-2748-4612

LAYS RABELO RATES - http://lattes.cnpq.br/2967162455921658 - https://orcid.org/0009-0007-8645-2575

CIBELLE CAMILO BARBOSA JARDIM - http://lattes.cnpq.br/0891642228157520 - https://orcid.org/0009-0003-3020-6683

AFONSO LUCAS OLIVEIRA NASCIMENTO - http://lattes.cnpq.br/1024773861852268 - https://orcid.org/0009-0007-1420-9656

## ADDRESS

NATHALIE BORGES COSTA
Av. Bela Vista, 26 - Jardim Esmeraldas, Goiânia-GO, 74905020
E-mail: nath_bc3@hotmail.com

Library Review - Romulo Arantes
Spell Check: Dario Alvares
Received: 24/01/24. Accepted: 02/04/24. Published in: 26/04/24.