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Case Report

Navigating advanced polycystic kidney disease: a case study of bilateral nephrectomy and life-saving transplant

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ABSTRACT

This case report discusses a 60-year-old female with polycystic kidney disease (PKD) who developed end-stage renal disease (ESRD). Diagnosed with polycystic kidney and liver disease in 2004, her condition worsened over the years, ultimately requiring hemodialysis and, later, kidney transplantation. Pre-transplant preparations included bilateral nephrectomy due to extensive cystic involvement of both kidneys. Post-transplant follow-up showed marked improvement in kidney function, with careful management using immunosuppressive, antifungal, and antihypertensive medications to prevent rejection and infection. This case underscores the critical role of multidisciplinary care in PKD patients undergoing transplantation for ESRD.

Keywords: Polycystic kidney disease, End stage renal disease, Autosomal dominant polycystic kidney disease, Renal replacement therapy

INTRODUCTION

Polycystic kidney disease (PKD) is a serious genetic condition characterized by development of fluid filled cysts mainly in kidneys, which can compromise the function and lead to severe complications.[1,2]

There are two main types of PKD: autosomal recessive PKD (ARPKD) and autosomal dominant PKD (ADPKD).[2,3] ADPKD is more common form and is a highly prevalent genetic disorder that affects the kidneys and other organs.[1,4] Approximately 12.5 million individuals worldwide are affected by ADPKD, with a prevalence of around 3.5 per 10,000 people in European Union.[1,4] When compared to combined prevalence of conditions such as sickle cell disease, Down syndrome, hemophilia, cystic fibrosis, and Huntington's disease, ADPKD is more common. In contrast, ARPKD is much rarer, with occurrence of about 1 in 20,000 individuals.[1,2]

It is caused by mutations in either PKD1 or PKD2 genes on chromosomes 16 and 4, respectively. PKD1 mutations account for around 85% of cases, while PKD2 mutations account for 15%.[5]

ADPKD is a fully penetrant disease, meaning that any person who inherits the mutation for ADPKD will manifest the disease symptoms. In most cases, patients with ADPKD have a positive family history of the condition.[2,6]

The clinical manifestations of ADPKD are mainly related to renal and extra-renal complications.[6] Renal manifestations include enlargement of both kidneys due to multiple cysts, discomfort in the lower abdomen or loin, renal colic or acute loin pain, pain in flanks, and hypertension which is the most common clinical feature of the disease, affecting 80% of cases.[2,6] The disease is expected to cause hypertension due to the secretion of renin when the renal tubules bearing cysts are stretched.[6]

Hematuria appeared first at about 30 years of age in about 19% to 35% of patients due to cystic hemorrhage.[5,6] Other symptoms may include urinary tract infection, cyst infection, polyuria, lower back pain, shortness of breath, early satiety, and renal stones. Kidney stones are found in

20% to 30% of patients, along with manifestations related to declining glomerular filtration rate (GFR).[5,6]

Extra-renal manifestations include intracranial aneurysms, cardiac and valvular diseases, and cystic involvement of other organs such as the liver and pancreas.[5,6] The prognosis for patients with renal insufficiency is typically poor, with the need for dialysis and renal transplant being a common outcome.[5,6]

Purpose of the study was to illustrate the clinical course and management of a patient with PKD, focusing on the complications associated with disease progression, the need for transplantation, and post-transplant care.

CASE REPORT

A 60-year-old female was hospitalized in nephrological department in 2019 with complaints of pain in lumbar region, increased blood pressure fatigue, polyuria, and polydipsia. She was diagnosed with polycystic kidney and liver disease in 2004. Her disease progressed significantly over time, leading to ESRD. In 2016, her kidney function declined to the point where conservative treatment was no longer enough, the patient was assessed for kidney transplantation due to the severity of her illness and the poor prognosis associated with dialysis alone.

The transplantologist along with her internal medicine physician briefed her about the advantages and disadvantages of having a kidney transplant as well as the process she would be going through and by June 2022, she started her hemodialysis.

To aid the hemodialysis a native distal arteriovenous fistula with auto-transplantation of a segment of the saphenous vein was carried out, preparing her for long-term hemodialysis.

Table 1: Laboratory results of patient the with PKD before and after kidney transplant.

Category	Laboratory tests	Reference	Results			
			2019	2020	2021	2022
Complete blood count	Red blood cells	4-9×109/l	4.19	3.63	3.75	3.85
	Hemoglobin	130-170 g/l	117	105	107	99
Biochemical analysis	Total protein	65-85 g/l	79	71	71	66
	Urea	1.7-8.3 mmol/l	23.1	25.3	32.1	50
	Creatinine	53-115 micromol/l	412	266	416	869
	Uric acid	0.2-0.42 mmol/l	0.41	0.33	0.35	0.37
	Total bilirubin	5-20.5 micromol/l	12.1	11.3	13.9	16.4
	Aspartate aminotransferase	5-37 U/l	23	12	13	16
	Alanine aminotransferase	5-42 U/l	15	11	10	14
	Iron	9-30.4 micromol/l	7.6	10.2	8.6	3.9
Urinalysis	Relative density	1018-1030	1012	1004	1015	1000
	Protein	0-0.15 g/l	0.11	0.715	0.25	0.407

The patient, diagnosed with PKD, experienced a progressive decline in kidney function between 2019 and 2022. Urine relative density consistently stayed between 1000 and 1015, indicating the kidneys' reduced ability to concentrate urine, a common issue in PKD due to the cysts impairing normal kidney function. Proteinuria increased significantly over time, peaking at 0.407 g/L in 2022, reflecting kidney damage as cysts further hampered the filtration process. Creatinine levels rose sharply, reaching 869 µmol/L, and urea levels peaked at 50 mmol/L in 2022, indicating severe kidney impairment as cysts obstructed normal waste elimination.

Patient's total protein levels also dropped, from 79-66 g/L, likely reflecting protein loss through urine and possible malnutrition, often associated with advanced kidney disease. Furthermore, anemia worsened, with red blood cell counts (3.85×10^9/L), hemoglobin (99 g/L), and iron (3.9 µmol/L) levels declining, which is typical in PKD due to reduced erythropoietin production by damaged kidneys.

MRI findings

Magnetic resonance imaging (MRI) of the patient's abdominal cavity revealed multiple cystic formations characteristic of advanced ADPKD. The liver appeared enlarged, with a measurement of up to 191 mm along the midclavicular line. Within the liver parenchyma, numerous areas of increased signal intensity on T2-weighted images were observed. These formations, varying in size from 3 mm to 17×26.5 mm, were well-defined, round to irregular in shape, and likely represent cysts. The gallbladder exhibited normal wall thickness and was devoid of visible stones, with the common bile duct measuring up to 8 mm in diameter.

The spleen, measuring 83×35.5 mm, displayed non-uniform parenchyma with areas of increased signal on T2-weighted images, suggesting possible cysts, the largest being 6.5 mm in diameter. The pancreas maintained a

normal configuration, with no signs of cystic involvement, and the main pancreatic duct was non-dilated.

In both kidneys, the renal parenchyma was almost entirely replaced by large, fluid-filled cysts. Some of these cysts showed homogeneous contents, while others demonstrated high signal intensity on T1-weighted images, indicating proteinaceous or hemorrhagic content. The complete absence of functional renal parenchyma confirmed the diagnosis of severe polycystic kidney disease, with associated renal replacement by cystic structures.

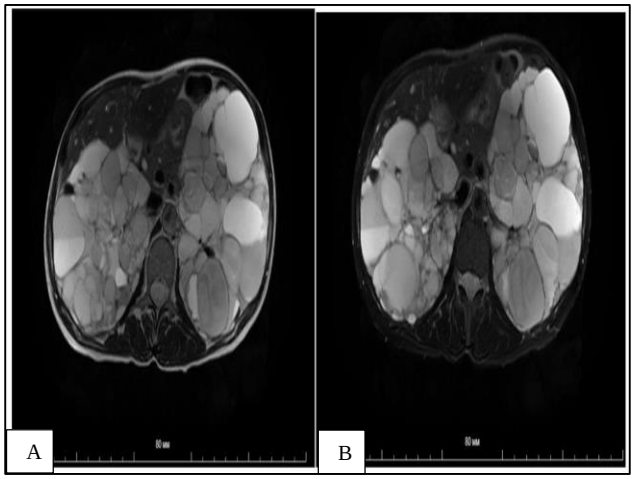

Figure 1 (A and B): Axial MRI images of the abdomen displaying the extensive cystic replacement in both kidneys.

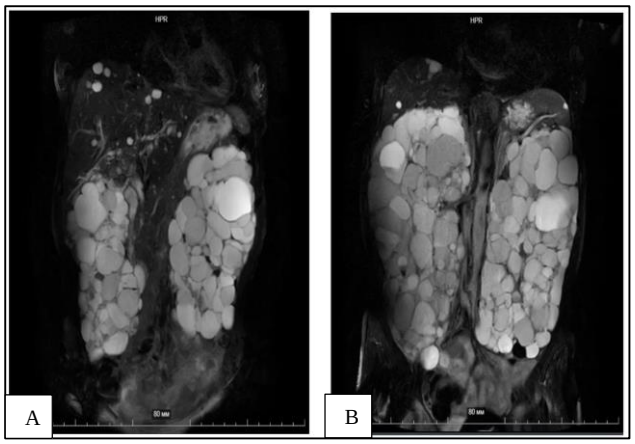

Figure 2 (A and B): Coronal MRI images illustrating the extensive bilateral cystic involvement in the kidneys.

Due to the severe cystic involvement of both kidneys, she underwent a right nephrectomy in June 2023 and a left nephrectomy in September 2023 as part of the pre-transplant preparation.

In October 2023, the patient was admitted to the department of surgical and transplantation in preparation for a scheduled kidney transplant, as a suitable donor organ became available. Laboratory tests showed despite patient being on dialysis three times a week her blood creatinine level was 617 µmol/l and urea level was 27.3 mmol/l.

The patient received a heterotopic cadaveric kidney transplant in October 2023. Ultrasound imaging performed after surgery showed that the graft, which was 116×60×67 mm with a parenchymal thickness of 18-19 mm, had been successfully placed in the right iliac area. Blood flow was intact after the transplant, and there was no evidence of acute rejection.

Later, at the end of October 2023, serum urea had dropped to 8.5 mmol/l and creatinine to 83 µmol/l, showing a notable improvement in kidney function.

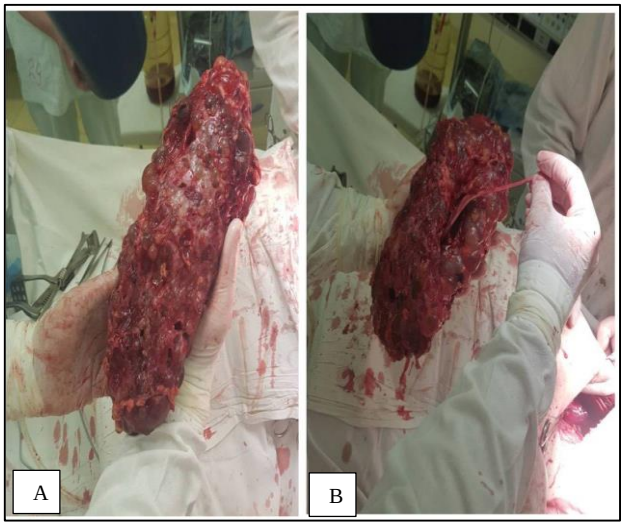

Figure 3 (A and B): Excised kidney showing extensive cystic changes characteristic of advanced ADPKD. The kidney is massively enlarged and distorted, with numerous fluid-and blood-filled cysts, almost completely replacing normal tissue.

The patient had thorough in-hospital care to ensure the kidney transplant would go well throughout their stay. To prevent organ rejection, immunosuppressive medications were started, which included the use of mycophenolate mofetil (MMF) and cyclosporine. Antibiotics and antifungals such nystatin, co-trimaxazole, and valganciclovir were administered to prevent infections. Subcutaneous dalteparin (LMW heparin) was administered to prevent thromboembolic challenges, considering the potential of clot formation during the post-operative period.

To preserve graft viability, the patient was prescribed an ongoing course of immunosuppressive medication that included cyclosporine and MMF after being discharged. Moreover, nystatin was used as an antifungal medication and valganciclovir and co-trimaxozole were kept up for the purpose of preventing infections. Antihypertensive treatment and other supportive drugs were also prescribed to support general health and control blood pressure.

After the transplant, the patient returned home in good health with no post-transplant concerns. To support patient health and long-term graft success, certain post-discharge directions were given. These included changing one's way of life to minimize the risk of infections, such as staying away from busy areas, and eating a low-cholesterol, hypo purine diet to boost general health. Also, the patient received advice to have a consistent drinking schedule and to be well hydrated. It was essential that patient be regularly monitored by a nephrologist transplantologist to guarantee the transplant's ongoing success and early identification of any foreseeable problems. It was specified that long-term immunosuppressive treatment is an essential part of continuing care to avoid organ rejection. Additionally, annual ultrasound exams of the internal organs and regular check-ups were recommended to check the condition of the transplant and overall organ health.

Following the kidney transplant in 2023, the patient's condition markedly improved. Post-transplant, creatinine levels dropped to 83 µmol/L and urea decreased to 8.5 mmol/L, indicating restored kidney filtration. Uric acid levels normalized, and proteinuria reduced significantly. The improvement in anemia was evident as red blood cell count, hemoglobin, and iron levels stabilized, reflecting better overall kidney function and the body's ability to maintain healthier blood and protein levels. The transplant significantly reversed many of the complications caused by CKD, greatly improving the patient's lab values and overall health.

DISCUSSION

ADPKD is a genetic disorder primarily affecting the kidneys and other organs, such as the liver, characterized by the formation of cysts that eventually impair organ function.[7] It is considered the leading genetic cause of ESRD worldwide.[8]

ADPKD involves mutations in several genes, with PKD1 (chromosome 16p13.3) and PKD2 (4q21) being the most commonly identified, accounting for 85% and 15% of cases, respectively.[8] PKD1 encodes polycystin-1, which plays a critical role in cell-cell and cell-matrix interactions, while PKD2 encodes polycystin-2, which is involved in intracellular calcium regulation. Dysfunction in these genes leads to cyst formation.[8]

Genetic testing for ADPKD can be conducted using direct DNA sequencing or gene linkage analysis. PKD1 mutations are associated with an earlier onset of ESRD, approximately 20 years earlier than PKD2 mutations.[7] Furthermore, PKD1-related disease is characterized by a greater number of cysts and a more rapid disease progression, emphasizing the importance of genetic testing as a prognostic tool, especially in patients with no prior family history of ADPKD.[1]

Screening for ADPKD is often performed using ultrasound based on diagnostic criteria developed for PKD1 mutation carriers.[6,9] However, as PKD2 mutations typically present with a milder disease course, alternative diagnostic approaches, such as CT or MRI, may be more suitable, particularly for younger individuals.[6,9]

Ultrasound criteria for ADPKD (Original ravine PKD1 diagnostic criteria):[9] Ages 15 to 29 years: ≥2 cysts, unilateral or bilateral, ages 30 to 59 years: ≥2 cysts in each kidney and ages ≥60 years: ≥4 cysts in each kidney.

Our patient, a 60-year-old diagnosed in 2004, experienced disease progression to ESRD, necessitating complex management strategies, including bilateral nephrectomy, hemodialysis, and kidney transplantation. This case underscores the limitations in ADPKD treatment, as there are currently no pharmacological interventions capable of halting cyst growth or reversing kidney damage.

Renal replacement therapies (RRTs), while life-saving, are temporary measures until transplantation can be performed. For patients with significant cyst expansion, bilateral nephrectomy before transplantation is critical for symptom relief (e.g., flank pain, recurrent infections) and to accommodate the transplanted kidney.[10] In our patient, nephrectomy was instrumental in avoiding post-transplant complications.

Although kidney transplantation remains the definitive treatment for ESRD in ADPKD, therapeutic options to prevent cyst growth are limited.[10] Experimental therapies, such as tolvaptan, have shown potential in slowing cyst progression but are associated with significant side effects.[11,12] Consequently, ADPKD management predominantly focuses on symptomatic treatment and RRT.

Post-transplant management in our patient demonstrated marked improvement in kidney function, facilitated by a multidisciplinary approach.[10,13] Immunosuppressive agents, antifungals, and antihypertensives were crucial in preventing rejection and infection.[10] Continuous monitoring remains vital to addressing potential complications, including rejection and infection.[5,10,12,14]

In conclusion, the lack of curative therapies for ADPKD highlights the need for further research to develop effective treatment strategies. While RRT and transplantation remain the most effective interventions, ADPKD continues to pose significant challenges in clinical management.[6,10,11,14]

CONCLUSION

This clinical report signifies the importance of kidney transplantation for patients with ESRD providing attention to the significant and progressive complications that arises from PKD. ESRD is seen as the final stage of CKD, at which the patient has to rely on renal replacing therapy options such as dialysis or kidney transplantation.

Effective post-transplantation care is of topmost importance. The usage of immunosuppressive medications is crucial to prevent kidney rejection and reduce the risk of infections. This report also enunciates the need for continuous patient monitoring and lifestyle modifications to prevent any post-transplant complications.

To sum up, this clinical report showcases the complexity of managing PKD and the importance of a multidisciplinary approach to ensure that PKD patients who have undergone kidney transplantation are provided with effective long-term care.

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