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CASE REPORT

Adult Diagnosis of Solitary Kidney and Renal Dysplasia in a Male Born Prematurely as a Twin: A Case Report

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Background: Congenital anomalies of the kidney and urinary tract (CAKUT) are a common cause of chronic renal disease and usually manifest either antenatally or in the early years of life. Solitary Kidney with Renal Dysplasia presents early in life due to regular antenatal sonographic scans. The rarity of diagnosis being deferred until adulthood has important implications when there is prematurity and other associated congenital anomalies, such as Congenital Heart Disease.

Case Presentation: We present a 23-year-old male experiencing sequential generalized edematous changes, decreased urine output, early feelings of fullness during meals, and vomiting following the intake of a meal for the last month. The patient was born prematurely at approximately seven months of gestation, and he was a twin, with the loss of the co-twin soon after birth. There was neither a previous chronic disease nor hospitalization. Physical evaluation showed the patient to be hypertensive with generalized edematous changes of the face and both lower limbs, along with systolic murmurs at the lower edge of the left sternal region. Laboratory findings indicated the presence of severe renal impairment with high serum creatinine and potassium, massive proteinuria, and severe anemia. Renal ultrasound revealed a small, echogenic right kidney compatible with chronic renal disease and an absent left kidney, confirming the suspicion of a solitary right dysplastic kidney. The cardiac study showed dilated cardiac chambers with an atrial septal defect, mild pericardial effusion, and the patient was placed on antihypertensive drugs and diuretics and started on hemodialysis.

Conclusion: This is a rare adult presentation of solitary kidney and renal dysplasia in a prematurely born male twin, further compounded by atrial septal defect and end-stage renal disease. This report underscores the significant implications of these complications as a direct sequel to prematurity regarding the associated severe consequences for the subsequent renal and cardiovascular systems of these patients. Antenatal ultrasound scans play a crucial role in the early detection and management of such abnormalities.

Keywords: solitary kidney, renal dysplasia, prematurity, atrial septal defect, antenatal ultrasonography

Introduction

Congenital anomalies of the kidney and the urinary tract (CAKUT) rank among the most frequent causes of chronic kidney disease (CKD) in the pediatric population, making a significant contribution to the renal morbidity that persists throughout life. With an estimated prevalence of 3–7 per 1000 live births worldwide, CAKUT encompasses a broad range of developmental abnormalities, such as renal agenesis, hypoplasia, and dysplasia.[1,2] These conditions affect about 2% of pregnancies and are frequently linked to genetic syndromes or other developmental abnormalities.[3]

One of the most common types of CAKUT is solitary kidney, which can be caused by either unilateral renal agenesis or a severely dysplastic non-functioning kidney.

The majority of the time, a single kidney is found during an early childhood evaluation for urinary tract abnormalities or during routine obstetric ultrasonography. However, delayed or accidental diagnosis still happens in adulthood, especially in those who do not have regular medical follow-up or early-life imaging. Even in the absence of symptoms during childhood, adult presentation is clinically significant because people with a single functioning kidney are more likely to experience hypertension, proteinuria, and a gradual decline in renal function over time.[2]

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Atypical nephrogenesis, characterized by an irregular renal pattern and reduced numbers of renal units, is known to be characteristic of renal dysplasia. The susceptibility to chronic renal disease and hypertension in adults has been well noted to be related to reduced numbers of renal units. As it has been clarified that nephrogenesis persists until gestational week 34–36 and can be disturbed in preterm births, susceptibility to chronic renal disease and hypertension in adulthood is additionally noted in adults who have been preterm.[4] Experimentation and epidemiological studies have shown that susceptibility to changes in renal function in early life exposes people to reduced renal function and damage in adulthood.[5]

The incidence of prematurity and low birth weight is significantly high in twin births, both being established risk factors for abnormally developed kidneys. However, long-term observation for kidney conditions in preterm-born twins is hardly practiced, more so in developing countries. Consequently, congenital kidney conditions can remain asymptomatic till a critical condition like hypertension or kidney damage surfaces in their adult life. CAKUT can coexist with congenital heart problems because both use common developmental and genetic factors during embryogenesis.[5,6]

In this case, we have an adult male who was born prematurely as a twin, being diagnosed later in life with a single kidney, renal dysplasia, and an atrial septal defect. This case clearly illustrates that preterm twins are highly predisposed to multisystem congenital anomalies, which, in essence, is very important for prevention by utilizing prenatal follow-up by ultrasonography.

Case Presentation

A 23-year-old male patient presented to the hospital for the evaluation of progressive swelling of the body for a month. Initially, the swelling was restricted to the lower limbs and then progressed to the face. Additionally, the patient presented with early satiety and vomiting of swallowed food, yet the patient had maintained his appetite. Moreover, the patient complained of decreased urination. There were no predisposing factors of prolonged medication administration, existing chronic illnesses, or hospitalization.

As a result of the twin pregnancy, he was born prematurely, approximately seven months of gestation; his twin brother died shortly after birth. According to his mother, as a baby, he was suffering from a dysmorphic notch in his chest (may be pectus excavatum), which resolved spontaneously. The patient does not have any relatives with chronic kidney or heart problems. His alcohol use and other significant social risk factors are denied by him.

Upon presentation, the vital signs included a blood pressure of 158/108 mmHg, pulse rate of 68 per minute, respiratory rate of 20 per minute, and a body temperature of 36°C. In addition, he complained of decreased urination in the genitourinary system, easy satiation and vomiting in the gastrointestinal system, and palpitations with shortness of breath with exertion, specifically walking uphill, and thus NYHA Class I symptoms.

Physical examination revealed a patient not in distress and not appearing ill, with facial edema. Pitting edema in both lower limbs is evident and classified as stage II. Cardiovascular examination showed normal heart sounds with a systolic murmur that is best appreciated over the lower left sternal edge. Pulmonary examination showed clear lung fields.

There was mild pericardial effusion and mild ascites on the ultrasonography of the abdomen and heart. The heart chambers were enlarged, with an atrial septal defect visible and hypertrophy of the interventricular septum. Renal ultrasonography revealed a small echogenic kidney on the right side, indicating chronic renal disease, with the left kidney absent.

Laboratory investigations showed hyperkalemia with a potassium level of 5.69 mmol/L (reference range 3.5–5.1 mmol/L), elevated urea at 31.6 mmol/L (reference range 1.7–6.0 mmol/L), and markedly elevated serum creatinine of 1095 μmol/L (reference value approximately 62 μmol/L). Hematological analysis revealed severe anemia, with a hemoglobin level of 6.8 g/dL (reference range 12–18 g/dL) and a red blood cell count of 2.35 $\times 10^6/\mu L$. A 24-hour urine collection demonstrated significant proteinuria of 2.351 g (reference range 0–0.14 g).

The treatment given to the patient consisted of antihypertensive medication with oral nifedipine 40 mg twice a day, intravenous furosemide 40 mg three times a day, and oral spironolactone 25 mg once a day. Due to the degree of renal impairment and biochemical abnormalities, hemodialysis was started. The diagnosis was made of congenital malformation of the heart and kidney involving a solitary kidney with renal dysplasia, atrial septal defect.

Patient Perspective

The patient reported never knowing about a possible congenital problem with his kidneys or heart until he was an adult, and was upset with his diagnosis and hemodialysis needs. After his clinical condition improved, his motivations for long-term care, such as evaluation for a transplant, and improving his personal and future goals, which include a return to his educational pursuits, became evident.

Discussion

Single kidney and renal dysplasia are one of the most frequent congenital anomalies of the kidney and urinary tract (CAKUT), but their adult presentation is relatively rare. In fact, the majority of them are usually found antenatally or within the first years of childhood as a routine finding on imaging studies, making it possible to have an early follow-up and treatment.[7,8] The delayed diagnosis that occurred in this particular case highlights the need to consider congenital anomalies even in patients who are being evaluated for an adult-onset renal failure, proteinuria, or hypertension for the first time in their lives. In patients who have only one functioning kidney, compensatory hypertrophy and hyperfiltration can preserve apparent renal function during childhood and young adulthood. Later on, proteinuria, hypertension, and progressive renal dysfunction due to hyperfiltration can cause the delayed presentation that is often observed in patients who present as adults with CAKUT.[9]

The main reliance for the early identification of CAKUT is on antenatal ultrasonography and targeted renal evaluation in the neonate, particularly high-risk groups such as premature infants, twins, and those presenting with associated congenital anomalies. Although there is no documented need for screening for the responsible gene or condition on a neonate-wide scale, evaluation for the gene can be proposed for individuals presenting with a syndrome.[10]

There has always been an association with premature birth and impaired nephrogenesis. Nephron development goes on until 34–36 weeks of gestation, and preterm delivery, when it occurs, may lead to an underdeveloped number of nephrons, causing the person to be prone to hypertension, proteinuria, and chronic renal diseases later in life.[4] The twin pregnancies and premature and LBW babies increase the risk of complications and anomalies associated with the development of renal anomalies.[11] In this case, the patient's preterm twin birth likely contributed to solitary kidney development and renal dysplasia, highlighting the long-term sequelae of early-life perinatal factors. Late adult presentations of solitary kidney and renal dysplasia have been reported, though few. This case is unique in the context of prematurity, twin birth, and the association of atrial septal defect, underlining the importance of multisystem evaluation even in adults.[12]

This phenomenon is the result of shared embryological pathways in organogenesis: Congenitally anomalous kidneys most often occur in conjunction with cardiac malformations. Atrial septal defect (ASD) is one of the most common cardiac anomalies found together with renal anomalies, sharing common genetic and developmental mechanisms that involve abnormalities in mesodermal derivatives.[13] Such findings emphasize that when either renal or cardiac anomalies are diagnosed, there needs to be consideration of multisystem diseases, even into adulthood.

Antenatal ultrasonography detects renal and cardiac anomalies relatively early in pregnancy. Routine prenatal imaging should aid in the detection of solitary kidney, renal dysplasia, and associated congenital heart defects, thus enabling timely postnatal monitoring and intervention.[8,14] In settings where resources are limited or antenatal care is irregular, these anomalies may go undetected until later in adult life, as illustrated in this case.

Without longitudinal follow-up, a delayed presentation of CAKUT can be no different from acquired CKD in that both would present similarly as hypertension, proteinuria, anemia, and end-stage renal failure. It is significant to mention that a suggestive birth or postnatal history of congenital abnormalities and/ or absence of one kidney would direct the clinician to a developmentally related cause of CKD in a patient evaluated in adulthood.

Early detection allows for organized follow-up and reduces the risk of progression towards renal dysfunction, while also providing important information on cardiovascular risk assessment.

Patients with single kidneys and renal dysplasias should be managed by managing their blood pressure, the amount of proteins in the urine, and preventing further nephrotoxicity. When the stages are advanced, these individuals will need renal replacement therapies, which include hemodialysis. Follow-up should not be restricted to the renal aspects but

should also involve the cardiovascular issues that are commonly associated with these stages. This particular case underlines the necessity for personalized care in individuals with congenital multisystem anomalies.

The limitations of the case are that Prenatal ultrasound records and genetic investigations were unavailable. In addition, the original ultrasound images were not archived; therefore, assessment was based on official radiology reports confirming a solitary kidney with renal dysplasia and associated congenital cardiac abnormalities.

Conclusion

This case illustrates a rare adult presentation of a solitary dysplastic kidney in a male who was born prematurely as a twin. It emphasizes prematurity and twin pregnancy as significant risk factors for multisystem anomalies that can present clinically late into adulthood. It reiterates the role of antenatal ultrasonography and organized lifelong follow-up in such high-risk patients, which permits timely detection, delayed progression of CKD, and successful long-term renal and cardiovascular outcomes.

Data Sharing Statement

All data generated or analysed during this case report are included in the published article.

Ethics Approval and Consent to Participate

Ethical approval was not required for this case report in accordance with local institutional policies. Written informed consent was obtained from the patient for participation and publication of this case.

Consent for Publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying data. A copy of the written consent is available for review by the Editor upon request.

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Disclosure

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