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Posterior Urethral Valves Disorder in Non-Twin Siblings: Case Report and Literature Review

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Abstract

Although a rare condition, Posterior Urethral Valves (PUVs) are the most common obstructive cause of chronic renal disease in children. In order to provide an early diagnosis, it is crucial to identify families with a high risk of developing this disorder. Here, we report a rare case of non-twin siblings with PUV. The first sibling was diagnosed at 2 months of age, presenting with renal insufficiency and managed properly. Despite the brother's history and general prenatal care, the second sibling presented with the same disorder, at 3 months of age, suffering from acute renal insufficiency, urinary tract infection and severe anemia, revealing even more important complications than the first sibling. We have reviewed the 12 cases of non-twin siblings previously reported in the literature, and noticed that in the majority of these families, late diagnosis was observed. Herein, we discuss aspects of management of the disorder, and highlight the importance of specific antenatal evaluation in boys with a positive family history.

Keywords: Posterior urethral valves; Siblings; Genetic predisposition to disease; Screening

Introduction

The incidence of Posterior Urethral Valve (PUV) disorder, a life-threatening obstructive anomaly that affects males, is approximately 1 in 4000-25000 births [1-3]. Although a few familial cases have been reported, including in siblings, there is no established genetic predisposition to the disorder [4]. It has been postulated an autosomal recessive pattern of inheritance, from genetic investigation from families with affected offsprings, but further investigation need to be performed confirm this hypothesis [5]. The scarse data for this kind of research reassures the importance of reporting such cases.

In developed countries, the diagnosis is made antenatally based on the prenatal ultrasonographic findings of bilateral hydronephrosis with a dilated bladder and dilated posterior urethra (keyhole sign). In addition, the bladder wall, which normally does not exceed 3 mm, may be thickened [6].

PUVs are the most common cause of Chronic Kidney Disease (CKD) due to urinary tract obstruction in children, demonstrating the importance of an early diagnosis and implementation of an appropriate management paradigm [7].

Twelve cases of non-twin siblings with PUVs have been previously described in the literature. Even in these cases, most children did not receive an early diagnosis, leading to irreversible urinary tract damage.

Here, we present the case of non-twin siblings with PUVs that were diagnosed separately. In a combination with a review of previously reported cases, we discuss the importance of early identification of this disease for improved patient outcome.

Case History

The first sibling presented with urinary retention at 2 months of age and his serum creatinine level was 2.5 mg/dL. A diagnosis of PUV was made based on the findings of an ultrasonography examination and voiding cystourethrogram (Figure 1). Initially, a bilateral proximal loop ureterostomy was performed, and 6 months later, PUV endoscopic electrosurgical ablation was performed. At 2 years of age, the right ureterostomy was closed, and a cystoscopy showed residual valves that were ablated using the same technique. A year later, the left ureterostomy was closed. During follow-up, Renal Bladder Ultrasound (RBUS) and urodynamic testing revealed normal findings. At present, the 5-year-old boy has no voiding complaints and demonstrates stable renal function confirmed by Dimercaptosuccinic Acid (DMSA) scintigraphy.

The second sibling was brought to the clinic at the age of 3 months, because of the need for frequent diaper changes, the presence of a distended abdomen, and prostration behavior. On physical examination, a distended bladder was evident. An RBUS showed a normal right kidney, severe left hydroureteronephrosis, and a distended bladder. A voiding cystourethrogram showed typical findings of PUVs (images not available). Laboratory tests revealed severe anemia (hemoglobin level, 5.9 g/dL), a positive urinary culture (*Escherichia coli*), and a serum

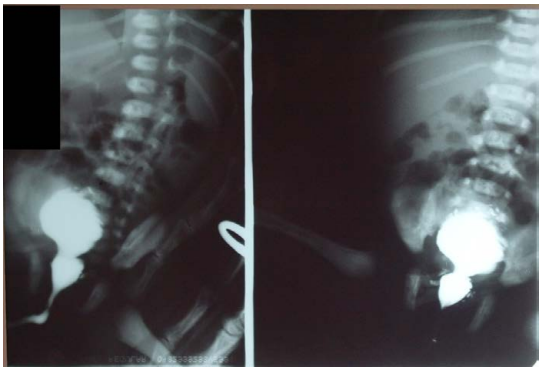

Figure 1: Voiding cystourethrogram of the first child showing posterior urethra dilation and irregular bladder wall.

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creatinine level of 2.1 mg/dL. After antibiotic treatment, the child underwent valve ablation as an initial treatment. The boy recovered, but due to persistent high detrusor pressures and postvoid residual urine, clean intermittent catheterization was implemented. At the current age of 3 years, RBUS showed persistent left hydroureteronephrosis and bladder wall thickening. DMSA scintigraphy showed impairment of relative left renal function, and his serum creatinine level is 1.8 mg/dL. Urodynamic testing showed normal sensitivity and bladder capacity, and no involuntary contractions. Currently, the child has spontaneous voiding four times a day and is catheterized three to four times a day.

Prenatal ultrasound records were unavailable, but the mother reported that no problems arose during either pregnancy.

Discussion

Early diagnosis of PUV is essential for decreasing associated comorbidities, because renal impairment and other complications begin developing as early as the antenatal period [8]. It is important to clarify that the siblings underwent different initial treatment approaches, but current evidence shows no long-term advantage of urinary diversion prior to valve ablation compared with direct ablation when urinary tract instrumentation is feasible.

In an extensive review of literature, only 12 cases of families with non-twin siblings with PUV were found (Table 1) [4,5]. The age of diagnosis varied from antenatal to 3 years, with 6 cases presenting after 6 months.

Author	Age of Diagnosis	Presentation
Counseller and Menville (1935)	7 months 6 months	Urinary tract infection Urinary tract infection
Hasen and Song (1955)	7 months 22 months	Palpable abdominal mass Frequency, poor urinary stream
Kjellberg et al. (1957)	Unknown Unknown	Unknown Unknown
Farkas and Skinner (1976)	7 days 2 years	Urinary retention with overflow dribbling No signs, evaluated because of brother
Thomalla et al. (1989)	1 day 1 day	Unknown Abdominal mass
Borzi et al. (1992)	1 day 7 weeks	Potter`s syndrome Poor urinary stream
Borzi et al. (1992)	3 days 3 weeks	Potter`s syndrome Poor urinary stream
Crankson and Ahmed (1993)	Antenatal Antenatal	Bilateral hydronephrosis Bilateral hydronephrosis with dilated bladder
Trembath and Rijhsinghani (2002)	Antenatal Antenatal	Bilateral hydronephrosis with dilated bladder Bilateral hydronephrosis with dilated bladder
Sawant et al. (2005)	2 days 3 days	Anuria, palpable abdominal mass Abdominal distention, urinary dribbling
Weber et al. (2005)	Unkonwn Unkonwn Unkonwn Unkonwn Unkonwn	Urosepsis Unkonwn Unkonwn Unkonwn Unkonwn
Schreuder et al. (2008)	Antenatal 3 years Neonatal	Bilateral hydronephrosis Urinary tract infection Strain, dribbling stream, and vesicourethral reflux

Table 1: Current data on non-twin Siblings with PUV [4,5].

At our institution, most cases are detected by morphological prenatal ultrasonography and further confirmed by voiding cystourethrogram after the first week, but unfortunately, not all pregnant women have full access to prenatal programs, and some children may have not been fully screened for this manageable condition.

The present report found that the family history must be considered at the initial evaluation of children with PUV because diagnosis in the second sibling was delayed despite a history of PUV in the elder sibling. Although an incomplete prenatal ultrasonography screening program may explain late diagnosis in these cases, according to the current literature, the identification of this anomaly during the antenatal period was made in only a small proportion of cases. This suggests that the fetal ultrasonographer should take particular care while evaluating the urinary system or the screening should include more frequent examination in those families with a history of urinary tract anomalies.

Conclusion

In order to minimize associated comorbidities, early diagnosis should be the mainstay of the management of PUV. This case adds on the limited information about this disease and supports the notion that antenatal ultrasonography in male siblings of patients with PUV focus on the identifying early signs of the disorder in order to allow adequate and timely intervention.

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