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Case Report

Urogenital Sinus Malformations: A Case Report

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Abstract

Persistent urogenital sinus (PUGS) is a rare congenital anomaly where the urogenital sinus fails to separate into distinct urinary and genital passages, resulting in a single common passage for both the urethra and vagina in female neonates. PUGS is most commonly associated with disorders of sex development. We report the case of a three-year-old female, with normal external genitalia, who presented with recurrent urinary tract infections and abdominal distension. She was ultimately diagnosed with PUGS complicated by hydroureteronephrosis. A cutaneous vesicostomy was done to decompress the hydroureteronephrosis followed by total urogenital sinus mobilization (TUM) to correct the congenital anomaly. The report discusses the diagnostic challenges, emphasizing the importance of timely intervention. It explores the embryological basis of PUGS, its diverse clinical presentations and the surgical approaches for correcting this anomaly and restoring normal urinary and genital anatomy. The rarity of isolated PUGS underscores the need for heightened clinical awareness in pediatric patients with unusual urinary symptoms. Early diagnosis and intervention are crucial to prevent complications associated with this congenital anomaly.

Keywords: *Persistent Urogenital Sinus (PUGS); Total Urogenital Sinus Mobilization (TUM); Vesicostomy; Urogenital Sinus Malformations (USM)*

Introduction

Persistent urogenital sinus (PUGS) is a rare, congenital anomaly in which the urogenital sinus fails to separate into distinct urinary and genital passages during fetal development. This condition results in a single common passage for both the urethra and vagina in female neonates. The overall incidence of urogenital sinus malformations (USM) is 6 in 100,000 female births [1,2]. In 80% of the cases, PUGS is associated with congenital adrenal hyperplasia [3]. PUGS is also a characteristic of syndromes like McKusick-Kaufman syndrome, Bardet-Biedl syndrome and hand-foot-genital syndrome [4,5]. The mere persistence of the embryological structure, i.e. the urogenital sinus, in the absence of any disorder of sexual differentiation or other syndromic features, is extremely rare [3,5]. In our index case the external genitalia were completely normal and no other syndromic features were present. This rarity underscores the need for heightened clinical awareness and thorough evaluation when faced with recurrent UTIs and unusual urinary symptoms in pediatric patients.

Herein, we report the case of a three-year-old female with a history of recurrent urinary tract infections (UTIs) and abdominal distension who was ultimately diagnosed with PUGS. This case report explores the diagnostic and management challenges associated with PUGS, emphasizing the importance of timely intervention to prevent complications. The discussion delves into the embryological basis of

PUGS, the diverse clinical presentations it can manifest, and the significance of radiological investigations in making an accurate diagnosis. Additionally, we elucidate the surgical approaches employed to correct this congenital anomaly and restore normal urinary and genital anatomy.

Case Presentation

A 3 years old female, with a past medical history of recurrent episodes of urinary tract infection for which her mother took the patient to different clinics, was admitted at our hospital on 27/06/2022 with complaints of crying while micturition since 3 - 4 months after birth, and increased frequency of small amounts of urine and distended urinary bladder. The physical examination reveals a pulse of 110 bpm, temperature of 100F, RR of 23 br/min, a distended lower abdomen, and middle tenderness. The patient does not present organomegaly. A plan of urinary catheterization was done, although urethral stricture was suspected due to the failure of catheterization. On further inquiry on the urination problem to the mother, she reveals the patient used to urinate from the vaginal orifice since birth, and clinically urogenital sinus abnormality was suspected and had no other systemic illness. The mother also referred to having had an uneventful pregnancy and that the other sibling is healthy.

Urine examination revealed pus cells 10 - 12/HPF and creatinine with a value of 2.75 mg. dL. USG of the whole abdomen was done and it showed bilateral hydroureteronephrosis. IVU couldn't be performed at that time on account of raised creatinine. She was evaluated under general anesthesia, and there was an opening of the urethra at the right wall of the lower vaginal canal. A cutaneous vesicostomy was done and the catheter was placed on the vesicostomy site. On 2/7/22 creatinine was rechecked and it was 1.76 mg/dL Later she was discharged with a catheter in situ on 6/7/22 and advised to readmission to the hospital.

She was readmitted on 25/7/22. Creatinine was rechecked and it was 1.76 mg/dL. Then IVU and a urogram with contrast were performed which showed bilateral hydroureteronephrosis mild on the right, moderate to gross on the left due to congenital short segment stricture in the terminal ureter, about 4 mm of posterior urethra well delineated and a thick irregular wall with a towering urinary bladder.

A surgery was done on 2/8/22 to repair the urogenital sinus, and the ectopic urethral opening was freed from the surrounding vaginal wall. Then it was placed to the normal urethral opening. In doing so the vaginal canal was decreased in size, so vaginoplasty was also done to make vaginal opening appropriate. A catheter was placed on a new urethral opening. Postoperatively there were no complications and the patient recovered well. After some days, the catheter on the vesicostomy site was clamped and urine started to come from the catheter placed on the urethral opening. On 13/8/22, the catheter from the vesicostomy site was removed but urine started to leak from the vesicostomy site, so a catheter was placed back at vesicostomy site and vesicostomy site was repaired. The patient was discharged on 15/8/22 and told to follow up after 15 days. Due to the fact of having recovered completely after 15 days, the patient maintained a follow up of 12 months.

Discussion

PUGS is a rare congenital anomaly in females, where the urogenital sinus fails to properly separate during fetal development, resulting in a single, shared passage for both the urinary and genital systems. The classical presentation of PUGS consists of a female neonate with a pelvic mass or abdominal distension along with ambiguous genitalia [6,7]. The pelvic mass/abdominal distention is secondary to the buildup of urine in the vagina and uterus called hydrometrocolpos [2,8,9]. PUGS obstructs the outflow of urine from the common passage. On micturition, the vaginal orifice acts as a valve, which causes retrograde flow of urine into the vagina, resulting in its distention. Severe hydrometrocolpos can also cause urinary retention and dilation of the upper urinary tract, i.e. hydronephrosis.

Our patient had lower abdominal distention on presentation; however, on abdominal ultrasound and IVU, there was isolated hydroureteronephrosis without any sign of hydrometrocolpos. We can speculate two reasons for this isolated presentation: a congenital stricture in the left ureter (as shown on IVU) and a stenotic urethral opening in the vaginal canal. In rare cases, neonates may present with urinary ascites due to leakage of the retained urine into the peritoneal cavity [8-10]. Massive urinary ascites can also cause upward displacement of the diaphragm and resultant pulmonary hypoplasia in a neonate [10]. Distal-confluence PUGS may present with complaints of recurrent urinary tract infections. This was the main complaint of our patient. PUGS may also present in adult women with cyclical hematuria and superficial dyspareunia, as reported by Amer., *et al*. and Kumar., *et al*. respectively [1,11]. The early diagnosis and surgical intervention of PUGS are thus essential to prevent the development of the aforementioned complications [7,10].

A comprehensive understanding of the intricate embryological basis of PUGS is crucial for diagnosis and surgical management. In the fourth week of gestation, the digestive, urinary, and genital systems empty into a common cavity known as the cloaca [5,12]. By the sixth week, a caudally growing sheet of mesenchyme called the urorectal septum separates the cloaca into a ventral urogenital sinus and dorsal rectum. By this time, the genitourinary systems in male and female fetuses are undifferentiated and identical [4]. In female fetuses, the lack of the SrY gene and its downstream products, namely testosterone and anti-Mullerian hormone (AMH), lead to the development and caudal migration of the Mullerian ducts [4,5]. The Mullerian ducts fuse laterally to form the uterovaginal primordium and vertically with the urogenital sinus to form the Muller tubercle [7]. The Muller tubercle induces the formation of endodermal outgrowths called sino-vaginal bulbs. These bulbs initially grow into a solid epithelial vaginal plate, which later recanalizes to form the lower thirds of the vagina [1].

Any abnormality in the caudal migration of the Mullerian ducts and their fusion with the urogenital sinus results in PUGS. If the caudal migration is arrested at an early stage of development, it results in the formation of a long urogenital sinus (> 3 cm) with a high urethral opening and a shorter vagina [1,5]. On the other hand, if the migration is arrested at a later stage, it results in the formation of a short urogenital sinus (< 3 cm) with a normal vagina and low urethral opening [5]. In the current case a low urethral opening was noted which suggests a late-stage developmental arrest. Singh., *et al*. reported a similar case; however, they speculated that the anomaly may be due to the failure of exstrophy or eversion of the distal urogenital sinus, due to which the vagina and urethra did not acquire separate openings in the perineum [2]. The molecular basis of PUGS has been studied in great detail in chick embryos. An endoderm-derived signalling molecule called sonic hedgehog (shh) has been implicated in the coordinated development of the external genitalia, bladder and urethra [2,13].

A diagnosis of PUGS is made with the help of radiological investigations. Prenatal diagnosis is possible with the help of an ultrasound scan. Hydrometrocolpos can be identified on ultrasound as anechoic, septate mass lying posterior to the fetal bladder [6]. Fetal MRI can also be used to better understand the anatomy of the PUGS [10]. Postnatally, a high index of suspicion must be kept in patients presenting with recurrent urinary tract infections, a pelvic/abdominal mass, cyclical hematuria or dyspareunia. An abdominal ultrasound is the first line of investigation which reveals dilated vagina, uterus and urinary tract. A retrograde genitogram can indicate the point of confluence and the distended structures. Micturating urethrograms (MCUG), cystovaginoscopy and MRI are accessory investigations to aid the diagnosis [5,6].

The first step in managing our patient was to decompress the urinary tract. The obstruction of the outward flow of urine was causing an increase in the intraglomerular pressure. This high pressure impaired filtration of excretory products like creatinine [14]. Due to this reason creatinine levels were elevated. A cutaneous vesicostomy was done to decompress the hydroureteronephrosis and subsequently lower creatinine levels. Timely surgical correction was done which prevented any permanent scarring and loss of kidney function.

The definitive management of a PUGS involves surgical correction of the anomaly to return the urethra and vagina to their normal location in the perineum without jeopardizing the function of either of them. When the point of confluence is low-lying, the urethra is of an adequate length; the correction can be made by relocating the ectopic urethral opening from the vaginal orifice to its normal position in the perineum. This procedure is called total urogenital sinus mobilization (TUM). TUM requires extensive dissection to free the urogenital sinus from all its attachments; hence there is a risk of vaginal stenosis. For this reason, a perennial flap vaginoplasty is done to preserve the function of the vagina [10]. This was the procedure done in our case with no significant post-operative complications. However, when the confluence point is higher and urethral length is short, correction via anterior sagittal trans anorectal approach (ASTRA) has been recommended [7,10].

This case report highlights the importance of timely diagnosis and prompt surgical management to reduce the risk of complications due to a seemingly innocuous congenital anomaly. Correction of a urogenital abnormality during childhood is also essential to prevent the psychological repercussions on self-esteem and sexuality that are likely to arise during puberty or adulthood [7].

Conclusion

In conclusion, this case underscores the importance of early diagnosis and intervention in managing PUGS, not only to prevent physical complications but also to address potential psychological consequences in the patient's later life. It reinforces the significance of clinical awareness and thorough evaluation in the face of unusual urinary symptoms in pediatric patients and contributes to our understanding of this rare congenital anomaly.

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