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SHORT COMMUNICATION

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Perinatal diagnosis of congenital urogenital sinus abnormality

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Abstract

Anomalies of the urogenital sinus, which is a transient feature of the early human embryological development, are rare birth defects. Urogenital sinus abnormalities commonly present as pelvic masses, hydrometrocolpos, or ambiguous genitalia and most commonly occur within the context of congenital adrenal hyperplasia. Anomalies of the urogenital sinus requires surgical repair. We experienced a case of a female newborn with congenital urogenital sinus abnormality in which the early diagnosis helped us to prevent complications by decompressing the vagina soon after birth. Antibiotic prophylaxis was sufficient to avoid infections and to decompress the genitourinary system, thus allowing a deferred elective surgery to correct the sinus.

KEYWORDS
cloaca, newborn, urogenital abnormalities

1 | INTRODUCTION

The urogenital sinus is an expected and transient feature of the early human embryological development. In the embryo, the genital and urinary tracts develop from common precursors and contains structures of endodermal (urogenital sinus) and mesodermal origin (mesonephric and paramesonephric duct).[1,2] The urogenital sinus, which is derived from the hindgut, is formed during the fourth to the seventh weeks of gestation, after the urorectal septum separates the cloaca into the ventral urogenital sinus and the dorsal anorectal canal. In female fetuses, the urogenital sinus leads to development of the urethra, vulva, and lower vagina.[3] The use of mouse models has allowed the understanding of normal and abnormal urogenital tract development: recent studies have revealed roles in urogenital development for several factors, such as transcription factors, homeobox genes, and hormonal factors. For example, the absence of androgens is related to the complete separation of the urogenital system from the cloaca, whereas the presence of androgens inhibits this process.[4,5]

Anomalies of the urogenital sinus are rare birth defects characterized by the confluence of the urethra and vagina that creates a common channel of varying length. Namely, depending on the length of the common channel (>3 cm or <3 cm), anomalies of the urogenital sinus can be classified into long common channel and short common channel, the latter being more common (60% approximately).[6] Contrary to cloacal malformations, the anorectal canal has a separate perineal opening[1]; however, several malformations, including gastrointestinal, sacral, cardiovascular and, obviously, genitourinary tract abnormalities may be associated with urogenital sinus anomalies.[7,8]

Urogenital sinus anomalies commonly present as pelvic masses, hydrometrocolpos, or ambiguous genitalia and most commonly occur within the context of disordered sexual differentiation (e.g., congenital adrenal hyperplasia).[9] Moreover, they may be associated with several syndromes, such as the Bardet–Biedl syndrome and the Mckusick–Kaufman syndrome[10] or, in rare cases, ascribed to an arrest of normal cloacal development.[11] Early diagnosis of urogenital sinus anomalies is critical since hydrometrocolpos may determine

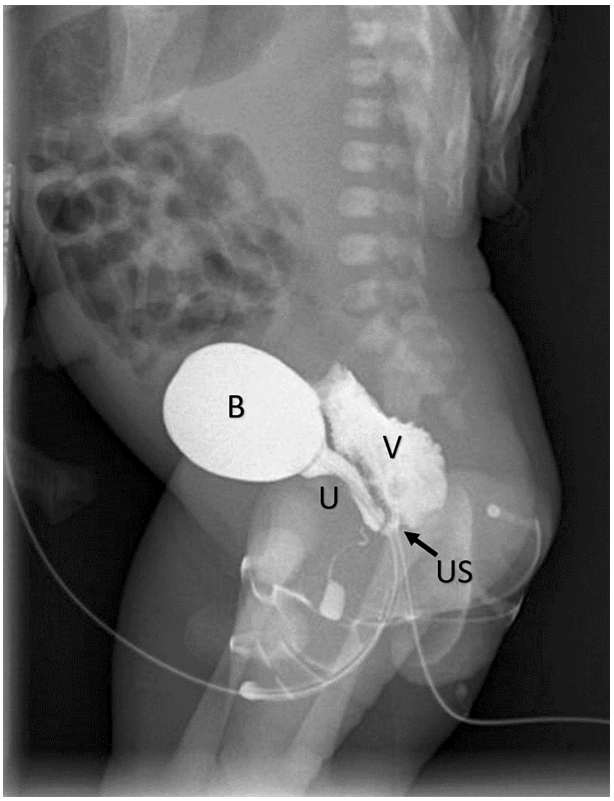

FIGURE 1 Cystovaginography of the female newborn showing the common channel bifurcating posteriorly into the vagina and anteriorly into the urinary bladder. B, urinary bladder; U, urethra; US, urogenital sinus; V, vagina.

urinary retention, dilatation of upper urinary tract, and compressive syndrome.

Here, we report the case of a female newborn showing at the prenatal ultrasound an abdominal mass of unknown origin, which was diagnosed as congenital urogenital sinus anomaly soon after birth.

2 | MATERIALS AND METHODS

This female newborn was delivered at 38 weeks' gestational age by vaginal delivery. The girl was the first offspring of unrelated healthy parents and family history was negative for congenital malformations and disorders. Pregnancy was unremarkable, with no history of drugs, ethanol, or radiation exposure; however, prenatal ultrasound revealed the presence of a pelvic mass of unknown origin. The cystic pelvic mass (30 × 40 × 70 mm), located behind the bladder, was identified at 30-week ultrasound scan.

At delivery, the Apgar score was 8 at 1 min and 9 at 5 min of life. Birth anthropometry: weight 2.890 g (10–25th percentile), length 47 cm (10–25th percentile), and head circumference 36 cm (97th percentile). Newborn's general appearance and clinical conditions were good, with normal vital signs. The initial full clinical examination performed soon after birth was normal: no abdominal/pelvic mass nor ambiguous genitalia were visible. Physical examination revealed a patent anus and a single anterior opening in the vestibulum; the urethral orifice was not visible.

Ultrasound of the abdomen on Day 1 showed a dilated vagina, filled with corpuscolated/echogenic fluid, and a hypertrophic uterus. The bladder was normal, but anteriorly displaced by the dilated vagina. The remaining parts of the renal and genitourinary systems were normal. Based on these findings, the presence of a urogenital sinus abnormality was hypothesized. A cystourethrogram was performed through the single anterior opening: interestingly, the vagina and the common channel were well outlined with contrast; however, we could not contrast the bladder. In the attempt to better delineate the anatomy and to confirm the diagnosis, a cystovaginography was performed by placing a second catheter anterior to the first one in the urethra. The exam demonstrated a common channel measuring 1.9 cm in length, a urethral opening ectopically located on the anterior vaginal wall, a dilated vagina, a normal bladder located anteriorly, and the urethra measuring 22 mm in length (Figure 1). These findings were consistent with a short common channel urogenital sinus. Nonetheless, the patient underwent a cystovaginoscopy to obtain a direct visualization of the common channel, vagina, and bladder in order to define diagnosis.

Therefore, the patient underwent an extensive screening for associated anomalies: laboratory examinations, such as hormonal investigations (ACTH, cortisol, renin, aldosterone, testosterone, and 17-OH progesterone) and karyotype were normal, ruling out disorders of sexual differentiation (e.g., congenital adrenal hyperplasia). Spine ultrasound and echocardiography were also negative, thus ruling out genetic syndromes and associated congenital malformations.

3 | RESULTS

Considering the clinical picture, the laboratory tests, and the imaging results, a diagnosis of persistent urogenital anomaly was made, and the patient was transferred to the Pediatric Surgery Unit. The patient attended regular outpatient follow-up at the Pediatric Surgery Unit; she underwent abdominal ultrasounds every 3 months, which resulted normal with no signs of hydronephrosis and megaureters. A cystovaginoscopy was repeated at 3 and 12 months of age and a mildly dilated vagina, a common channel measuring less than 2 cm and a normal bladder were observed. The baby is under continuous antibiotic prophylaxis to avoid infections.

4 | DISCUSSION

Anomalies of the urogenital sinus are rare birth defects whose incidence is estimated in 0.6/10 000 female live births.[10] The developmental mechanisms of these abnormalities are still under debate; however, it is likely that in females a persistent urogenital sinus may represent a developmental arrest, occurring soon after the urorectal septum separates the urogenital tract and the anorectal tract. In view of this, anomalies of the urogenital sinus are usually found as a single

common orifice of the vagina and urethra, while the rectum, anal canal, and anus are well developed.[11] Although anomalies of the urogenital sinus may be sporadic and isolated, sometimes they can be found in association with other anomalies and/or as a part of complex syndromes.[12,13] Most common associated anomalies are renal agenesis, hydronephrosis, polycystic kidneys, uterine anomalies (cervical atresia, bicornuate uterus), vaginal duplication or atresia, esophageal atresia, imperforate anus, and sacral hypoplasia.[14] Moreover, it should be remembered that anomalies of the urogenital sinus often occur within the context of congenital adrenal hyperplasia and other disorders of sex development.

Urogenital sinus anomalies commonly present as a pelvic mass, which is due to the distension of the vagina and uterus (hydrometrocolpos) and/or a dilated bladder; otherwise, they are sometimes suspected in the presence of ambiguous genitalia.[15] In our case, a pelvic mass was the only manifestation as no ambiguous genitalia nor other anomalies were detectable. Due to the rarity and complexity of this condition, the diagnosis of urogenital sinus anomaly is challenging. Indeed, when external genitalia look normal and the anus is normally placed, these anomalies can be easily overlooked. However, a high index of suspicion for urogenital sinus anomalies is mandatory among patients with pelvic cystic masses, hydrometrocolpos and a single anterior opening in the vestibulum (as in the present case), which is a pathognomonic feature.[7,16] Given that urogenital sinus anomalies may be associated to many entities, it is necessary a combined strategy of clinical examination, laboratory tests (karyotype and hormonal tests), and imaging tools to orient the diagnosis and the therapeutic approach. Although prenatal diagnosis of these anomalies may be performed, the prenatal findings need always to be confirmed postnatally.

Usually, postnatal ultrasound, voiding cystourethrogram, and cystovaginoscopy are sufficient to make diagnosis. Magnetic resonance imaging may be used to clarify complex anomalies.[17]

This report aims to stress the importance of the early diagnosis of urogenital sinus anomalies to prevent complications in the newborn. As a result of the urinary retention, abdominal distension, hydronephrosis, elevation of the diaphragm, or compression of the inferior vena cava may be possible. In the present case, the early diagnosis helped us to prevent complications by decompressing the vagina soon after birth. Antibiotic prophylaxis was sufficient to avoid infections and to decompress the genitourinary system, thus allowing a deferred elective surgery to correct the sinus.

5 | CONCLUSIONS

Although urogenital sinus anomalies are very rare, they should always be suspected in infants presenting with abdominal masses, hydrometrocolpos, or ambiguous genitalia. It is therefore imperative that neonatologists and pediatricians are aware of these abnormalities. The present case emphasizes the importance of an early diagnoses in order to stress the need of a combined strategy resulting from clinical examination, laboratory tests, and imaging tools to orient the diagnosis and the therapeutic approach and also to prevent detrimental consequences.

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CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

DATA AVAILABILITY STATEMENT

Data sharing not applicable—no new data generated.

ETHICS STATEMENT

Verbal and written consent was obtained from the parents regarding the publication of the case and images. This report does not contain any personal information that could lead to the identification of the patient.

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