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Case report

Anterior urethral hamartoma in a female infant with anorectal malformation: A rare case report and literature review

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ABSTRACT

Introduction and importance: Urethral hamartoma is highly unusual, as documented in the literature with only one case that ever reported. Here, we report a case of female infant with urethral hamartoma and anorectal malformation.

Case presentation: An 8 months old female infant was previously diagnosed with anorectal malformation (ARM) with rectovestibular fistula that had underwent colostomy. She presented with a vestibular mass protruding from her vestibule 4 months prior. Preoperative voiding cystourethrography (VCUG) and intraoperative cystoscopy supported that the mass did not originate from the bladder mucosa but rather from anterior urethra. Subsequently, the mass excised, and histopathology result confirmed urethral hamartoma. The procedure was successful without any observed complication and no evidence of recurrence.

Clinical discussion: Urethral hamartoma is often misdiagnosed as other diseases such as polyps, ureterocele, condyloma acuminatum, and malignant tumors. VCUG and urethral cystoscopy before a complete mass excision are helpful to clarify the location, texture, and size of the tumor, as well as its relationship with the surrounding tissues. Combined with clinical manifestations, the diagnosis of benign tumor can be initially determined, and the final diagnosis depends on the histopathological examination.

Conclusion: Our case is the first case reported in Indonesia. Due to its rarity and nonspecific symptoms, urethral hamartoma may be misdiagnosis as other urethral conditions. This report emphasizes the importance of accurate clinical and histological assessment in pediatric urogenital tumors to avoid misdiagnosis and excessive management. This case also highlights the potential association of urethral hamartoma with other congenital malformations particularly anorectal malformation.

1. Introduction

Urogenital tumors are generally rare in female infants, with specific types and frequencies varying by region and study population [1]. The incidence of these tumors in this demographic is not widely documented due to its rarity, however, when the mass occur, it typically include benign and low-grade malignancies, such as rhabdomyosarcoma, nephroblastoma (Wilms tumor), and occasionally hamartomas [2,3]. The most common histopathological findings of urogenital tumors in children include rhabdomyosarcoma which often presents with spindle cells and is typically diagnosed in the genitourinary tract, and the other is nephroblastoma (Wilms tumor), characterized by a triphasic pattern with blastemal, stromal, and epithelial components [2,4].

The term "hamartoma" refers to a benign, although "tumor-like" exaggerated proliferation of elements that recapitulates the normal components of the tissue in which the hamartoma arises [5]. Urothelial hamartoma is extremely rare, as far as we know with only one case ever reported in the literature, meaning an accurate incidence rate is difficult to determine. Hamartoma caused by a mistake in combination and arrangement of normal tissues in an organ during development. It is often a kind of abnormal development and growth of tissues after birth. This overgrown tissue still has the morphology of mature tissue and does not show the characteristics of tumor tissue, but sometimes can develop into a tumor. The most common site of hamartoma is the kidney, followed by the lung, hypothalamus, and liver. It rarely occurs in the perineum, and it is even rarer in the urethra [3].

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Anorectal malformations (ARM) are congenital anomalies of the anorectum which cover a wide spectrum of anatomical anomalies, characterized by an absence of a normally formed anus at its normal position within the perineum. It is well known that children with ARM show a high incidence of associated anomalies in other organ systems. The overall incidence of these associated anomalies is more than 60 %. Urological anomalies are frequently seen in patients with ARM and can result in upper urinary tract deterioration. Study conducted by Rowe reported that incidence of urogenital anomalies associated with high or intermediate forms of ARM presented in 50–60 % of cases. Previous studies recommended that all children with ARM should undergo an ultrasonography of the urinary tract in the neonatal period [6,7].

Here, we reported a case of a female infant with urethral hamartoma and anorectal malformation that was recently treated in our department and shared our experiences in the diagnosis and treatment process. This manuscript was prepared following the SCARE criteria [8].

2. Case presentation

An 8 months old female infant was previously diagnosed with anorectal malformation (ARM) with rectovestibular fistula that had underwent colostomy. Patient also had other congenital anomalies such as hemivertebrae and thumb hypoplasia. She presented with a vestibular mass protruded from her vestibule 4 months prior. The mass was painless and did not bleed easily, the surface was uneven, with ruddy mucosa without skin coverage. The serum creatinine level was 0.54 mg/dL and ultrasound of the urinary system findings were within normal limits, there was no sign of hydronephrosis from right and left kidney and the bladder image displayed there was no cystic dilatation from ureterovesical junction. VCUG was performed, the results showed the contrast filled the urinary bladder completely, there was no mass, no vesicoureteral reflux (VUR) and no bladder prolapse (Fig. 1). The patient underwent cystoscopy, vaginoscopy and mass excision under general anesthesia. The procedures were performed in Dr. Hasan Sadikin Hospital, Bandung, Indonesia, by pediatric surgery consultant. Intraoperative cystoscopy finding revealed that the bladder neck mucosa and vaginal mucosa was smooth without abnormal mass and there was no mass in the inner wall of the urethra (Fig. 2 A, B, C, D). The silicone catheter No. 8 Fr was inserted to the urethra after cystoscopy. We stretched the reddish, polypoid vestibular mass measuring 2 cm × 1.5 cm × 1 cm, with a stalk measuring 0.5 cm, with the base attached to the outer wall of the urethra, located 1.5 cm from urethral opening (Fig. 3 B). Subsequently, the mass was excised completely, and a careful hemostasis was performed. Post operative period was uneventful and patient was discharged home with the catheter maintained for 14 days. The child was reexamined in the outpatient department of our hospital. The first follow-up conducted on day 14 for catheter removal and the wound condition assessment which was appeared to be good.

The mass sent for thorough pathological examination. The histopathology result (Fig. 4) showed a layer of transitional epithelium forming polypoid structures with edematous connective tissue stroma containing scattered infiltrate of lymphocytes and histiocytes, among haphazardly arranged hyperplastic muscle cells and foci of von Brunn's nests, given this findings, an urethral hamarthoma diagnosis was made. The patient underwent routine follow-ups for up to one year, there was no any observed complication and no evidence of recurrence.

3. Discussion

Urethral hamartoma manifests as a painless, slow-growing, and polypoid tumor-like lesion located in the distal urethra. The clinical manifestations are mainly urinary retention, dysuria, and hematuria. It is often misdiagnosed as other diseases such as polyps, ureterocele, condyloma acuminatum, and malignant tumors. X-ray imaging, ultrasonography, CT scan, MRI, VCUG can help diagnose urethral hamartoma. A surgical excision is the effective method for the treatment of urethral hamartoma. Urethral cystoscopy before an excision is helpful to clarify the location, texture, and size of the tumor, as well as its relationship with the surrounding tissues. Combined with clinical manifestations, the diagnosis of benign tumor can be initially determined, and the final diagnosis depends on the histopathological examination [3,9,10].

Consistent with the literature review, in this study, an 8-month-old female infant presented with a vestibular, painless, polypoid mass, measuring 2 × 2,5 × 1 cm, protruding from her vestibule since 4 months

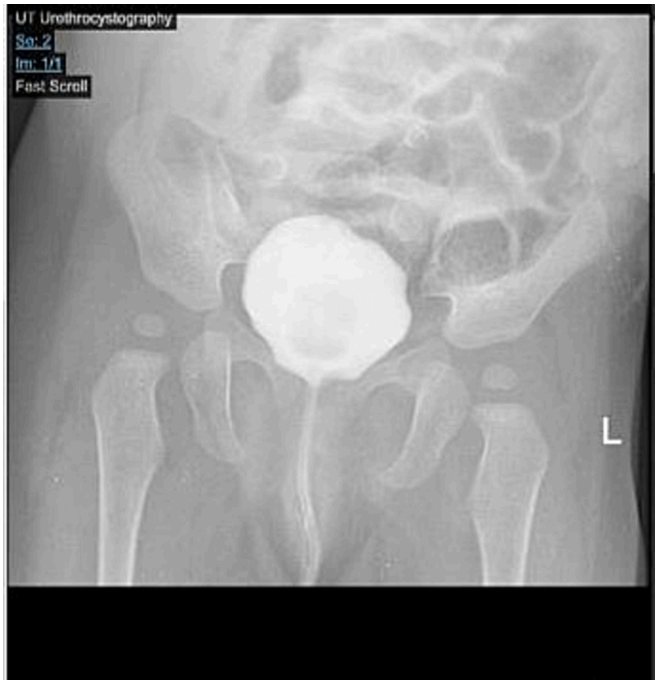

Fig. 1. Voiding cystourethrography (VCUG) revealed the contrast filled the bladder completely and there was no bladder prolapse.

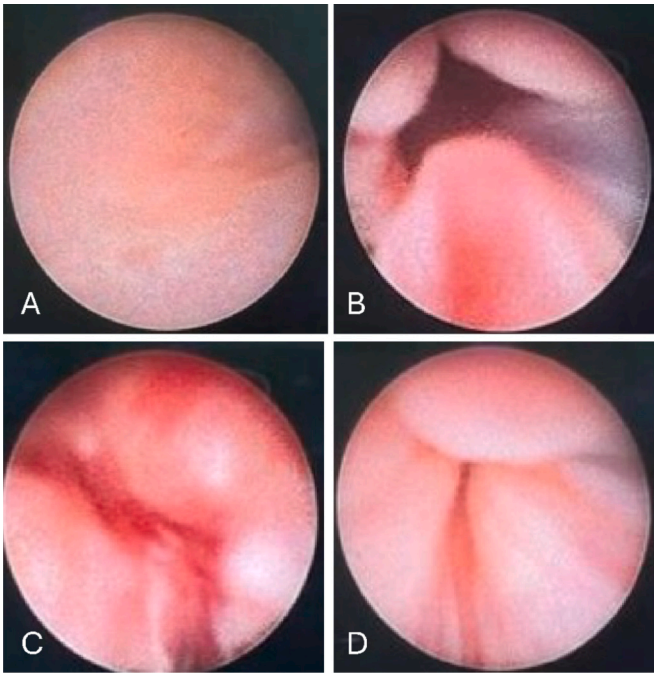

Fig. 2. (A & B) There was no mass from bladder and bladder neck; (C) There was no mass from vagina; (D) The urethra.

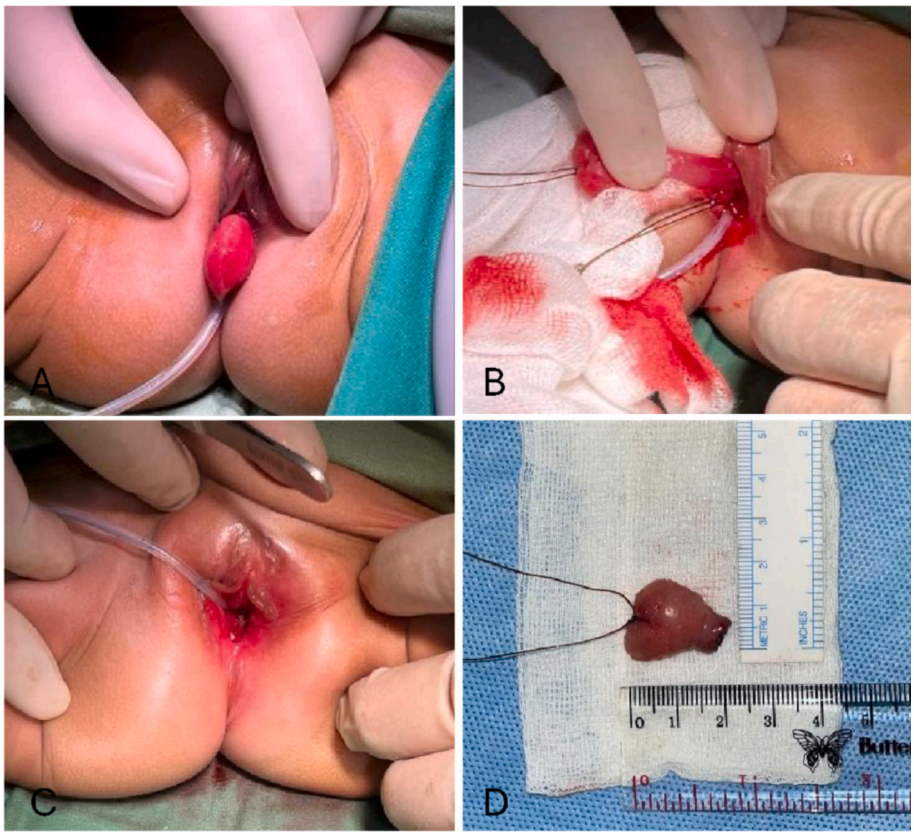

Fig. 3. Direct evaluation of the mass under general anesthesia. (A) Reddish polypoid mass protruded from vestibule; (B) The stretched mass appears to have a stalk, with the base attached to the outer wall of the urethra, located 1.5 cm from urethral opening; (C) Post mass excision; (D) The mass measuring 2 cm × 2,5 cm × 1 cm with 0.5 cm stalk, was successfully excised completely.

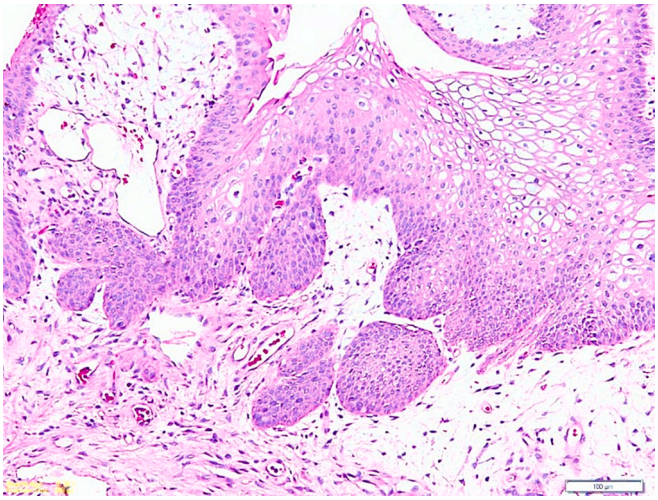

Fig. 4. Microscopic feature of urethral hamartoma (Hematoxylin and eosin stain; 100× magnification). The lession showed a layer of transitional epithelium forming polypoid structures, edematous connective tissue stroma containing scattered infiltrate of lymphocytes and histiocytes, hyperplastic muscle cells and foci of von Brunn's nests. No malignant signs are observed.

prior. The solid mass extended from the anterior urethra, it did not bleed easily. Several differential diagnoses, such as polyps, caruncle, ureterocele and neoplasia, were considered based on the findings of the mass. Normal serum creatinine level and ultrasound of the urinary system findings indicated that the kidney function was within the normal range and there were no structural abnormalities in the urinary system such as hydronephrosis, double collecting system or cystic dilatation from ureterovesical junction. The VCUG result showed contrast fully filling the bladder without evidence of mass, vesicoureteral reflux (VUR) and bladder prolapse. The ureterovesical junction appearance showed no abnormalities on both examinations, ruling out the possibility of ureterocele [11]. Subsequently, we conducted cystoscopy and vaginoscopy, revealing the bladder neck and bladder mucosa was smooth, the vaginal mucosa was normal and no mass detected. However, we identified vaginal septation, a rectovestibular fistula, and a reddish mass measuring 2 cm × 1.5 cm × 1 cm originating from the anterior urethra. Macroscopically, the mass appears to adhere to the outer wall of the urethra, making it invisible on urethrocystoscopy examination. Consequently, we decided to perform a complete excision of the urethral mass and sent it as a specimen for anatomical pathology examination. After surgery, the patient healed uneventfully and had routine follow-ups up to one year with no sign of reccurence.

The histopathological features of urethral hamartoma are as follows: the tumor is covered by normal transitional epithelium, and the stroma is composed of smooth muscle, blood vessels, and glands. Hamartoma has no atypical hyperplasia or malignant changes in the epithelium and has a low risk of recurrence after complete resection [3].

In this study, the specimen shows a layer of transitional epithelium that is partially hyperplastic, forming polypoid structures, with nuclei within normal limits. The subepithelial area consists of edematous connective tissue stroma with an infiltrate of lymphocytes and histiocytes. Among this, there are hyperplastic muscle cells arranged haphazardly, with nuclei within normal limits, and von Brunn's nests are found in several areas. No malignant signs were observed. This finding was consistent with urethral hamartoma, mesenchymal tissue was found, nests of urethral epithelium (von Brunn nest) and muscle component that showed no malignancy.

Hamartoma is not a true tumor, and it grows slowly. It can grow with the development and growth of the body, but stops when it reaches a certain size. It is more coordinated with the body and has a low malignant transformation rate [3]. Urethral hamartoma generally tends to be benign. The underlying cause is unknown but may involve an abnormal growth of cells during embryonic development, potentially due to an error in the complex signaling pathways that control tissue development and growth [12]. In the case of a urethral hamartoma, it is thought that an error in the development of cells that normally line the urethra may lead to the overgrowth of these cells, resulting in the formation of the hamartoma. As in this case, the polypoid mass covered by transitional epithelium with non-atypical nuclei, within a stroma containing haphazardly arranged bundles of muscle cells accompanied by urothelial proliferation forming nests known as von Brunn's nests, indicating an error in the signaling of urethral cells during embryonic development, leading to the abnormal growth of the local structures.

Hamartoma in the urethra has been reported to be associated with other malformations. For example, a case of a posterior urethral hamartoma with hypospadias in a child has been documented, including those affecting the anorectal region. This association may be seen more commonly in syndromic cases, such as in children with conditions like Beckwith-Wiedemann syndrome or other congenital syndromes that involve malformations of multiple organ systems [3,7]. In current case, the patient had multiple congenital abnormalities, including anorectal malformation with rectovestibular fistula that had underwent colostomy, hemivertebrae, and thumb hypoplasia, which suggests a possible underlying developmental disorder.

Our case is the first case reported in Indonesia. This case highlighted the importance of careful clinical examination and proper imaging to establish the correct diagnosis, as urethral hamartoma can mimic other urethral lesions. In the described case, the urethral hamartoma was successfully removed via complete mass excision, with the patient achieving a good postoperative outcome.

4. Conclusion

Urethral hamartoma in infant is an exceedingly uncommon condition, to the best of our knowledge with only one case ever reported. It presents as a painless, slow-growing, polypoid mass typically located in the distal urethra and it can be misdiagnosed as other conditions such as polyps, ureterocele, condyloma, or malignant tumors. VCUG and cystoscopy are essential for determining the tumor's location and assessing the urethra's condition, while surgical excision remains the primary treatment, showing no signs of recurrence. Histopathological examination is essential for diagnosis confirmation. More cases are needed to be observed as well as longer-term effective follow-ups to confidently verify this conclusion. This case report highlights the importance of accurate clinical assessment, imaging, and pathological evaluation in managing rare urethral lesions like hamartomas. In particular, within the context of underlying developmental disorders. This will further deepen the understanding of tumors in children, provide a certain theoretical basis for clinical diagnosis, and reduce the misdiagnosis rate.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in Chief of this journal on request.

Ethical approval

Our study is exempt from ethical approval in our institution, Faculty of medicine, University of Padjajaran/Hasan Sadikin Hospital.

Guarantor

Vita Indriasari
Karina

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Author contribution

Vita Indriasari: Study concept, writing the paper, data analysis
Karina: Writing the paper and data collection
Hermin Aminah Usman: Data analysis

Declaration of competing interest

There is no conflict of interest throughout the process of preparing this case report.

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