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Type I congenital vaginal atresia with multiple organ malformations

Vrozená vaginální atrezie typu I s mnohočetnými orgánovými malformacemi

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Summary: **Background:** Vaginal atresia (VA) is a rare female genital tract malformation characterized by the absence or incomplete development of the vagina, often leading to cyclic abdominal pain and menstrual blood retention in adolescent patients. Vaginal atresia is often accompanied by multiple organ malformations. The condition poses significant challenges in diagnosis and management, requiring a multidisciplinary approach. **Case description:** Herein, we report a case of type I congenital VA in a 13-year-old female presenting with congenital biliary atresia and urinary system malformation. Upon initial evaluation, the patient exhibited VA incision and laparoscopic excision of a left ovarian cyst under general anesthesia. By the 2-month follow-up, the patient reported normal menstrual flow without accompanying abdominal pain. **Conclusion:** This case underscores the complexity of type I congenital VA concurrent with multiple organ malformations. Timely identification and proper management strategies are crucial for securing the best possible patient outcomes. Further research is warranted to elucidate the underlying mechanisms and improve treatment modalities for such cases.

Key word: vaginal atresia – biliary atresia – urinary system malformation – case report

Souhrn: **Východiska:** Vaginální atrezie (VA) je vzácná malformace ženského genitálního traktu charakterizovaná nepřítomností nebo neúplným vývojem pochvy, často vedoucí k cyklickým bolestem břicha a zadržování menstruační krve u dospívajících pacientek. Vaginální atrezie je často doprovázena mnohočetnými orgánovými malformacemi. Tento stav představuje významné problémy v diagnostice a léčbě a vyžaduje multidisciplinární přístup. **Popis případu:** Uvádíme případ vrozené VA typu I u 13leté ženy s vrozenou biliární atrezií a malformací močového systému. Po počátečním hodnocení pacientka podstoupila incizi VA a laparoskopickou excizi cysty levého ovaria v celkové anestezii. Během 2měsíčního sledování pacientka udávala normální menstruační tok bez doprovodných bolestí břicha. **Závěr:** Tento případ podtrhuje složitost vrozené VA typu I souběžné s mnohočetnými orgánovými malformacemi. Včasná identifikace a správná strategie léčby jsou zásadní pro zajištění nejlepších možných výsledků u pacientky. K objasnění základních mechanizmů a zlepšení léčebných modalit pro takové případy je zapotřebí další výzkum.

Klíčová slova: vaginální atrezie – biliární atrezie – malformace močového systému – kazuistika

Introduction

The theory of embryonic development posits that the upper two-thirds of the vagina, fallopian tubes, and uterus originate from the Müllerian ducts, although the lower part of the vagina depends on the urogenital sinus. Vaginal atresia (VA) arises due to abnormal development of the urogenital sinus, resulting in either the absence or incomplete formation of the vagina. This constitutes a rare type of female genital tract malformation, with reported incidences of VA ranging from 1 in 4,000 to 1 in 10,000, constituting 15% of female reproductive tract obstruction anomalies [1,2].

Literature reports that VA frequently accompanies genitourinary system anomalies, clinically presenting as unilateral renal agenesis, renal ectopia, renal malrotation, or renal dysplasia [3]. No cases of VA occurring simultaneously with biliary atresia and genitourinary system anomalies have been reported [4,5]. In this case, a or 13-year-old patient presented with abdominal pain, leading to the diagnosis of VA. The patient had a history of both biliary atresia and genitourinary system anomalies. The patient underwent VA incision surgery, followed by meticulous postoperative care and close monitoring of her recovery progress to ensure a quick recovery.

Case report

A 13-year-old female presented to West China Second University Hospital of Sichuan University on May 30, 2023, complaining of abdominal pain persisting for one month. She had experienced irregular lower abdominal distension one month prior to admission, worsening at night and unrelated to food intake or defecation, without fever, acid reflux, nausea, vomiting, or melena. Magnetic resonance imaging (MRI) findings included an anteverted uterus with normal morphology and size, endometrial thickness of approximately 0.85 cm, trumpet-shaped cervical o.s., significant vaginal dilation with an internally visualized mass showing short T1 and short T2 signals measuring approximately 7.0 × 4.1 × 5.0 cm, with clear borders and locally heterogeneous signal. A cystic mass with long T1 and long T2 signals, clear borders, and a diameter of approximately 1.9 cm, was noted in the left adnexal area. Pelvic fluid was observed, suggestive of possible hemoperitoneum. No special treatment was administered, and the abdominal pain symptoms disappeared. One day prior to admission, the patient experienced recurrent and worsening lower abdominal pain, suggesting a high likelihood of genital tract malformation.

The patient underwent liver transplantation at West China Hospital of Sichuan University at the age of 5 in 2014 due to congenital biliary atresia. Postoperatively, she has been receiving oral immunosuppressive therapy (tacrolimus combined with sirolimus) up until present. On August 9, 2020, the patient presented to West China Hospital of Sichuan University with a history of urinary incontinence persisting for over 7 years. Cystography revealed irregular bladder morphology and reduced capacity, while retrograde urethrography showed splenomegaly and indistinct visualization of the right ureterovesical junction. The left ureteral orifice was positioned below the bladder neck. Renal dynamic imaging indicated normal left kidney function and mild impairment of the right kidney, with obstruction of the left upper urinary tract and poor drainage of the right upper urinary tract. On August 18, 2020, the patient underwent laparoscopic-assisted left ureteral reimplantation and left ureteral reconstruction resulting in a diagnosis of ectopic opening of the left ureter. The patient's urinary control significantly improved compared to the preoperative state.

Admission Examination: Vital signs were within normal limits with a temperature of 36.5°C, pulse rate of 86/min, respiratory rate of 20/min, blood pressure of 115/64 mmHg, and heart rate 86/min. **Upon specialized examination:** An opening was observed in the vestibule of the external genitalia, but the urethral opening could not be identified. **Ultrasound indication (Fig. 1):** anteverted endometrium centrally located, measuring 0.25 cm (single layer) in thickness. Within the lower portion of the uterine cavity extending into the vagina, a fluid-filled dark area measuring 8.4 × 5.7 × 6.6 cm was observed, with multiple fine weak echoes and scattered slightly stronger echoes, without detectable blood flow signals. A fluid-filled dark area was observed in the pelvic cavity, approximately 1.7 cm deep. Renal ultrasound revealed: left kidney measuring 10.9 × 4.2 × 6.5 cm, and right kidney measuring 10.1 × 4.5 × 4.9 cm. Liver function and coagulation function showed no significant abnormalities.

The patient presented with congenital VA (Type I) along with multiple visceral malformations, currently experiencing menstrual retention and requiring surgical interventions. On June 1, 2023, at our institution, the patient underwent "VA incision and laparoscopic excision of a left ovarian cyst under general anesthesia." Intraoperatively, normal external genitalia development was observed. A 1.5 cm long opening was noted in the vestibule of the external genitalia with the urethral opening visible above it. A mucosal shallow depression was observed below, but the vaginal opening

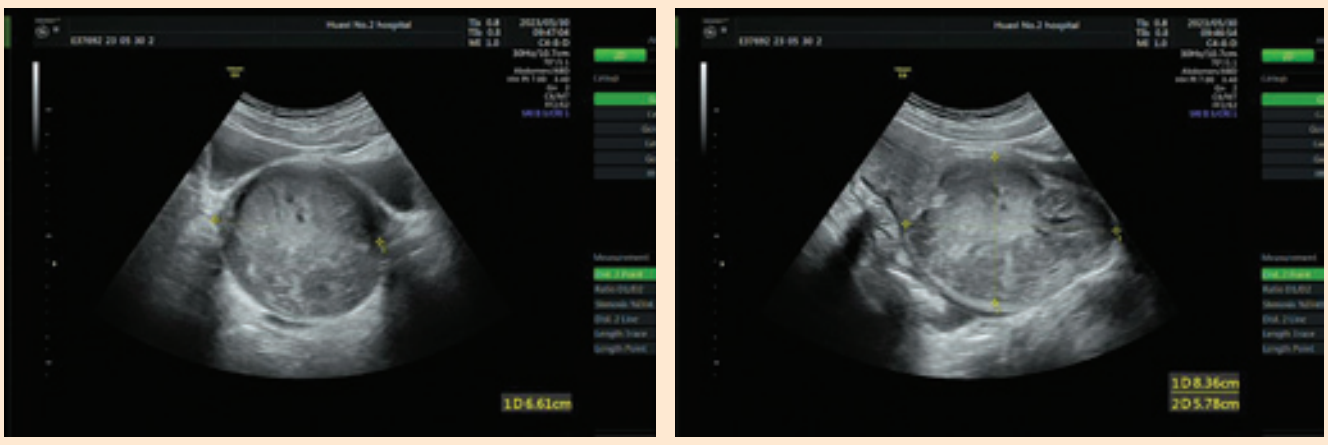

Fig. 1. Ultrasound indications.
Obr. 1. Ultrazvukové indikace.

was not reached. Cystoscopy revealed a smooth bladder wall with the right ureteral orifice located at the right bladder neck and the left ureteral orifice at the left bladder dome, both exhibiting normal urinary flow. The distance between the bladder neck and urethral meatus was approximately 1cm. A mucosal depression was incised below the urethral meatus, and connective tissue between the rectum and urethra was bluntly dissected, revealing the bulging end of the VA. Approximately 400 mL of chocolate-colored blood flowed out. Finally, an 8 × 3 cm vaginal cavity was formed, accommodating two fingers comfortably. The surgery proceeded successfully, with postoperative urinary catheterization and vaginal mold placement.

Postoperatively, vaginal molds were regularly replaced and vaginal dilation was performed to prevent scar contracture. The urinary catheter was removed on the second day post-surgery, and the patient experienced unobstructed urination. At the 1-month follow-up, the patient reported reduced menstrual flow. Gynecological examination showed no abnormalities in the external genitalia, a vaginal depth of 9cm, width sufficient for two fingers, well-healed incisions, and visible suture lines. By the two-month follow-up, the patient reported normal menstrual flow without accompanying abdominal pain.

Discussion

Currently, VA is classified into two types: Type 1, characterized by lower segment atresia with a normally developed upper vagina, cervix, and uterus along with a functional endometrium; Type 2, involving complete VA often associated with abnormal cervical development, while the uterus may be normally developed or exhibit malformations with a functional endometrium [6]. The size of the hematoma is inversely proportional to the length of the atresia.

In patients with VA, chromosomal and gonadal development are typically normal, and secondary sexual characteristics manifest as expected. Diagnosis is often challenging during infancy and adolescence. As patients reach puberty, clinical symptoms become evident due to menstrual blood obstruction, facilitating a definitive diagnosis. The most notable clinical feature of VA is the absence of menstrual periods during adolescence, despite normal secondary sexual characteristics accompanied by cyclic abdominal pain. The severity of abdominal pain is closely related to uterine development and the degree of VA. Generally, patients with complete VA but well-developed uteri may experience more severe abdominal pain at an earlier age.

In March 2020, the European Society of Urogenital Radiology (ESUR) released guidelines for congenital anomalies of the female reproductive tract, recommending sequences based on T2--weighted (T2WI) TSE/FSE sequences as the preferred MRI protocol for patients with congenital anomalies of the female reproductive tract. Requirements for T2WI sequences include slice thickness ≤ 4 mm, high matrix (512 × 512), and small field of view to achieve high--quality imaging [7].

Anticipating treatment exacerbates patients' suffering and may precipitate pelvic endometriosis and adenomyosis, compromising fertility and triggering severe complications such as pelvic infections and sepsis. Surgical intervention remains the sole effective approach for congenital vaginal atresia. To optimize surgical success rates, performing the procedure during the menstrual period is advisable when hematometra and symptoms are most pronounced, facilitating intraoperative identification of vaginal hematoma volume in Type I vaginal atresia patients. As per the 2019 ACOG committee's recommendations on managing acute obstructive uterovaginal developmental anomalies [8], vaginoplasty is the preferred treatment for distal vaginal atresia. Laparoscopic or combined abdominal and vaginal approaches may be considered for high vaginal atresia. Close postoperative follow-up is essential to monitor vaginal healing and dilation, ideally every 1–2 weeks until the patient adapts to vaginal pressure dilation and experiences pain relief [8]. In our case, the patient underwent vaginal atresia incision surgery under general anesthesia, with blunt and sharp dissection performed between the rectum and the urethra's connective tissue until liberation of the vaginal atresia end, draining menstrual blood and ultimately forming the vagina. Subsequently, the patient received regular mold pressure therapy showing satisfactory progress during postoperative follow-up.

The patient faced multiple challenges during the perioperative period. Firstly, due to her young age, the perineal surgical field was limited and the perineal tissue elasticity was poor, leading to increased surgical difficulty and a higher chance of postoperative vaginal adhesion and closure, possibly necessitating multiple surgical treatments. Secondly, the patient underwent liver transplant surgery for bile duct atresia and is currently on immunosuppressive therapy. Additionally, due to congenital anomalies of the ureters and bladder, the patient underwent left ureter cystoplasty and left urethroplasty. In recent years, she had experienced recurrent urinary tract infections many times. Preoperative comprehensive evaluations revealed no significant abnormalities in liver function, routine blood tests, renal function, and coagulation function, while urine analysis also did not indicate any urinary tract infections, which was crucial during the perioperative period. Considering the patient's oral immunosuppressive therapy, careful evaluation and balancing of infection risks during the perioperative period were paramount. Close monitoring of physiological parameters, timely implementation of infection prevention measures, maintain-

ing sterile procedures postoperatively, and comprehensive assessment of treatment efficacy and overall patient recovery during long-term follow-up were essential. Finally, attention was directed towards monitoring the patient's reproductive system function, wound healing in the surgical area, and potential signs of infection. Additionally, prompt adjustments and monitoring of immunosuppressive drug usage was carried out to maintain immune function balance and mitigate infection risks.

Conclusion

In conclusion, a comprehensive approach is crucial for diagnosing VA, involving thorough patient medical history assessment, detailed physical examination, and recommendation of comprehensive MRI imaging. Treatment methods should be tailored to the specific condition of the patient's atresia. Due to the complexity of postoperative scenarios, evaluating treatment outcomes and pregnancy rates requires a holistic assessment of the patient's overall condition. Active patient involvement in medical decision-making is advocated to ensure treatment plans meet individual needs and expectations. This holistic management approach aims to enhance patients' quality of life and optimize disease management throughout the treatment process.

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