Case Report

An Atypical Presentation of Vaginal Agenesis

Patricia S. Huguelet MD [1,*], Lauren T. May MD [2], Stephen M. Scott MD [1]

[1] Department of Obstetrics and Gynecology, Pediatric and Adolescent Gynecology, Children's Hospital Colorado, University of Colorado School of Medicine, Aurora, Colorado
[2] Department of Obstetrics and Gynecology, University of Colorado School of Medicine, Aurora, Colorado

A B S T R A C T

Background: Vaginal agenesis is rare and generally presents with primary amenorrhea and cyclic abdominal pain. We describe a case in which the diagnosis was delayed due to lack of initial pelvic examination and atypical findings on imaging.
Case: A 13-year-old girl with a known renal anomaly presented to the emergency department with primary amenorrhea and cyclic abdominal pain. She declined a pelvic examination and had normal laboratory testing and pelvic magnetic resonance imaging results. At 16 months later, she presented again and was diagnosed with vaginal agenesis and a large endometrioma.
Summary and Conclusion: This case illustrates the importance of the physical examination in the evaluation of primary amenorrhea. Further, it demonstrates that hematometra may not be present on imaging. Here, an endometrioma was the only abnormality noted on magnetic resonance imaging after 18 months of retrograde menstruation.
Key Words: Vaginal agenesis, Endometriosis, Clinical presentation

Introduction

The incidence of vaginal agenesis is approximately 1:5000 females, with a range of 1:4000 to 1:10,000.[1] It is usually accompanied by cervical and uterine agenesis. However, approximately 7% to 10% of these women have a functional endometrium within either a uterus that is obstructed but otherwise structurally normal or a rudimentary horn.[2]

Distal vaginal agenesis may occur as an isolated defect secondary to failure of the urogenital sinus to develop the caudal aspect of the vagina,[1] or it may occur with associated degrees of Müllerian aplasia. The very rare instance of total vaginal atresia with normal upper genital tract is thought to occur from either complete failure of vaginal plate formation or total failure of canalization of the vaginal plate.[1] Regardless of the etiology, patients with vaginal agenesis and a functional endometrium often present with primary amenorrhea and cyclic abdominal pain related to hematometra.[2–6] Coexisting urologic and skeletal abnormalities are also common.[1,6]

Here we review the case of a patient with vaginal agenesis presenting with a symptomatic ovarian endometrioma in the absence of hematometra. While endometriosis is frequently associated with outflow tract obstruction, endometriomas in the adolescent are rare,[7] and to our knowledge, this is the first case report to describe an endometrioma as the initial diagnostic indicator of vaginal agenesis.

Our objectives are to highlight this unusual presentation and to discuss the importance of a complete physical examination, especially when clinical suspicion of an anomaly is high.

Case

A 13-year-old girl was referred to the pediatric gynecology subspecialty clinic with the complaint of cyclic abdominal pain for the past 2 months. Her pain was located in the periumbilical region, was sharp and aching, and was exacerbated by physical activity. The pain was not associated with eating, urination, or bowel movements. She had been previously evaluated by a gastroenterologist and was diagnosed with constipation. However, daily polyethylene glycol (Miralax) provided only mild relief.

Her medical history was notable for a duplicate ureter, status post uncomplicated ureteroureterostomy in 2005. On review of systems, she reported breast development for the past year but had not yet menstruated.

On examination, she was noted to have normal vital signs and was well appearing. Tanner 3 breast development was noted. Her abdomen was soft and nondistended with normal bowel sounds. She had generalized periumbilical tenderness, as well as mild increase in tenderness with contraction of her abdominal wall muscles. She strongly declined a genital and rectal examination.

Based on the location of her pain and known constipation, a genitourinary source was deemed less likely. However, given the patient's history of a known renal anomaly, a pelvic magnetic resonance imaging (MRI) study was ordered to rule out the possibility of a noncommunicating uterine horn.

Imaging of the pelvis demonstrated a retroflexed uterus with slight vascular engorgement but normal contour

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* *Address correspondence to:* Patricia S. Huguelet, MD, Department of Obstetrics and Gynecology, University of Colorado and Children's Hospital Colorado, Anschutz Medical Campus, Academic Office 1, Box B-198-2, 12631 E 17th Ave, Aurora, CO 80045; Phone: (303) 724-2038; fax (303) 724-2056
E-mail address: Patricia.Huguelet@ucdenver.edu (P.S. Huguelet).

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(Fig. 1). The left and right ovaries were present with multiple small follicles. A 1.8-cm hemorrhagic cyst was noted in the left ovary. No uterine anomaly was identified. The left kidney demonstrated significant renal scarring but otherwise appeared normal.

The patient was then lost to follow-up until she presented to our emergency department approximately 16 months later. She reported 3 days of new-onset, severe left lower quadrant abdominal pain, radiating to her back. Pain was associated with nausea and vomiting but no diarrhea. She denied any fevers, chills, or burning with urination. She reported a normal bowel movement the previous day. The patient again reiterated that she had never menstruated and had been experiencing similar pain every month for the past 18 months, lasting for about 3 days with each episode.

On physical examination, she was afebrile, with blood pressure 122/69, heart rate 94/min, respiratory rate 20/min, and normal oxygen saturation on room air. The patient was in mild acute distress with significant tenderness to palpation and rebound throughout her abdomen. Limited genital examination revealed a normal urethral meatus and external female genitalia, but only a vaginal dimple with inability to insert a cotton-tipped swab (Q-Tip) past the labia.

Laboratory assessment was significant for an elevated white blood cell count to 25 with a left shift. All other laboratory results were unremarkable, including normal urinalysis. Ultrasound revealed a normal appendix and a large pelvic mass containing echogenic fluid with dependent debris, measuring 13.7 cm in greatest dimension. No identifiable left ovary was visualized. The uterus was displaced by the mass, with thickened echogenic endometrium but no other abnormalities. Overall impression was consistent with a hemorrhagic left ovarian cyst versus endometrioma. Tubo-ovarian abscess was deemed much less likely based on the homogeneous appearance on ultrasound.

Given the patient's abnormal pelvic examination and known renal anomaly, repeat pelvic MRI was ordered to evaluate for a noncommunicating horn as the etiology of the mass. MRI revealed a uterus displaced to the right, measuring $13.4 \times 7.9 \times 10.7$ cm. Echogenic debris within the endometrium measured 15 mm at the greatest diameter (Fig. 2). A 13-cm left adnexal mass was noted consistent with the ultrasound findings. The vaginal canal was poorly visualized (Fig. 3). A 2.4-cm hyperintense lesion was noted in the area of the cervix containing heterogeneous debris, consistent with blood within the cervical canal. The leading differential diagnosis included an ovarian endometrioma versus a noncommunicating left uterine horn and vaginal obstruction.

By the time imaging was completed, the patient had complete resolution of her pain and was resting comfortably. She was therefore discharged to home with a plan for outpatient pediatric gynecology follow-up.

The patient was subsequently taken to the operating room 1 month later for an examination under anesthesia and laparoscopic exploration. Her genital examination revealed hymenal tissue at the introitus but complete vaginal agenesis. On rectal examination, an area was palpated consistent with the cervix and lower uterine segment.

On laparoscopic exploration, a 13-cm left ovarian mass was noted. The cyst was drained, yielding 1000 mL of chocolate-brown fluid. Diffuse endometriotic implants were also noted throughout the upper abdomen and pelvis, consistent with stage IV endometriosis. Given the gross appearance of the cyst and diffuse implants, histologic confirmation was deemed unnecessary. A normal uterus was identified with bilateral round ligament attachments. A normal appendix was noted. Upper abdominal anatomy

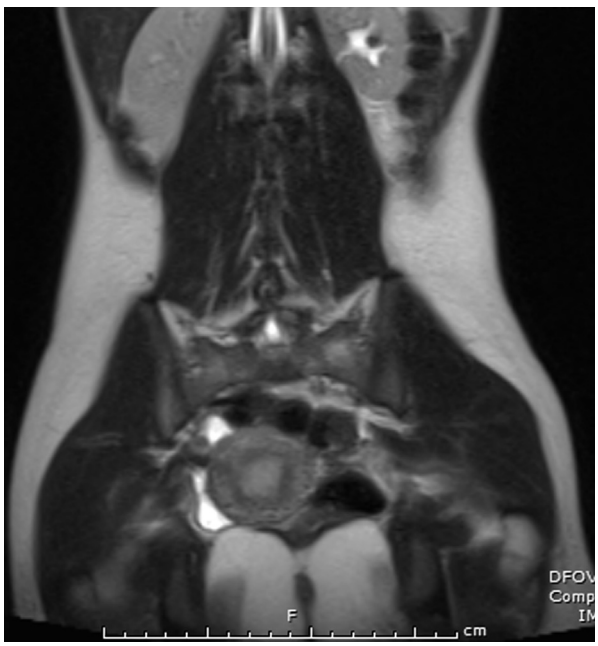

Fig. 1. Initial magnetic resonance imaging demonstrating uterus with slight vascular engorgement, but normal contour.

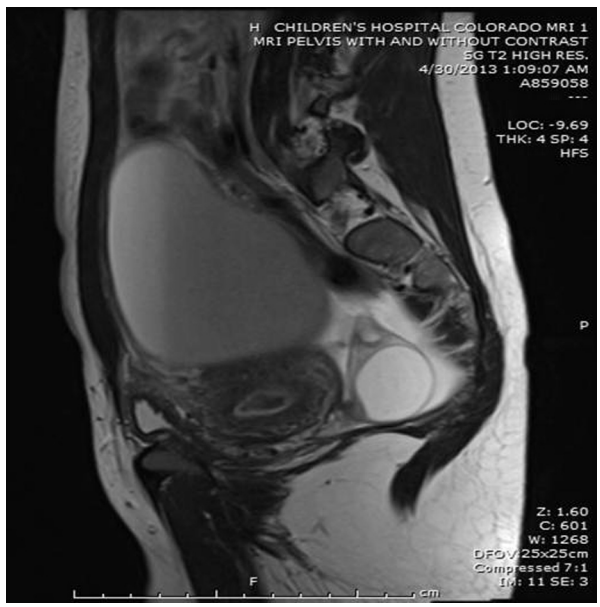

Fig. 2. Follow-up magnetic resonance imaging demonstrating large endometrioma and uterus with thickened endometrium but no hematometra.