Recurrent Abdominal Pain in Female Adolescence: An Overlooked Diagnosis

Dor Abdominal Recorrente em Rapariga Adolescente: Um Diagnóstico Pouco Reconhecido

Keywords: 46, XX Disorders of Sex Development; Abdominal Pain; Adolescent; Congenital Abnormalities; Mullerian Ducts
Palavras-chave: Adolescente; Anomalias Congenitas; Dor Abdominal; Distúrbios 46, XX do Desenvolvimento Sexual; Ductos Paramesonéfricos

Dear Editor,

We report a clinical case of recurrent abdominal pain in an adolescent girl ultimately diagnosed with type I Mayer-Rokitansky-Küster-Hauser syndrome. This rare congenital condition results from Müllerian duct developmental failure, leading to vaginal and/or uterine agenesis with intact ovarian function. Patients typically present with normal secondary sexual characteristics and external genitalia, and diagnosis is usually established during adolescence because of primary amenorrhea or cyclic pelvic pain, or rarely, later in life due to dyspareunia and/or infertility. Imaging is crucial for diagnosis. Management requires screening for associated malformations and focuses on restoring sexual function and quality of life through surgical or nonsurgical neovagina creation.[1-5]

A 13-year-old girl presented to the emergency department (ED) with progressively worsening left lower abdominal cramp-like pain for 24 hours and vomiting. She reported a one-year history of cyclic abdominal pain lasting three to four days, sometimes with nausea and vomiting, leading to several ED visits. Her medical history included surgically corrected tetralogy of Fallot. Growth and pubertal development were normal, with no menarche. Physical examination revealed sexual maturity rating/Tanner Stage 5 secondary sexual characteristics, normal external genitalia, with mild tenderness in the left lower quadrant.

Abdominopelvic ultrasound and laboratory studies were unremarkable. Gynecologic evaluation revealed normal external genitalia but inability to introduce a swab into the vestibule, suggesting vaginal anomaly. Rectal ultrasound demonstrated an absence of the vaginal canal and moderate peritoneal free fluid. Pelvic magnetic resonance imaging (MRI) demonstrated complete vaginal agenesis associated with a hypoplastic cervix and a normal uterine body (U0C4V4, ESHRE/ESGE classification system[6] – Fig. 1). The ovaries and urinary tract were normal. Hormonal profile and karyotype were normal. Genetic testing (TBX1, SNAP29) revealed no mutations.

Combined oral contraceptives improved symptoms. An exploratory laparoscopy was performed to confirm the anatomical findings. At 19, she underwent a mini-laparoscopic modified Vecchietti procedure, in which a traction device was laparoscopically attached to the vaginal dimple and gradually advanced to create a neovagina, with intraoperative indocyanine green guiding safe dissection. Millimetric endometriotic lesions and a small right ovarian endometrioma were noted. Postoperative vaginal dilators maintained and progressively increased neovaginal length. She is currently asymptomatic and able to lead a normal sexual life. In the event of future patient desire, a subsequent procedure to anastomose the neovagina to the uterus may be considered.

This case highlights that complex Müllerian anomalies may be diagnostically challenging in adolescents with cyclic abdominal pain, even after recurrent ED visits. Early recognition and multidisciplinary management are essential to achieving optimal outcomes.

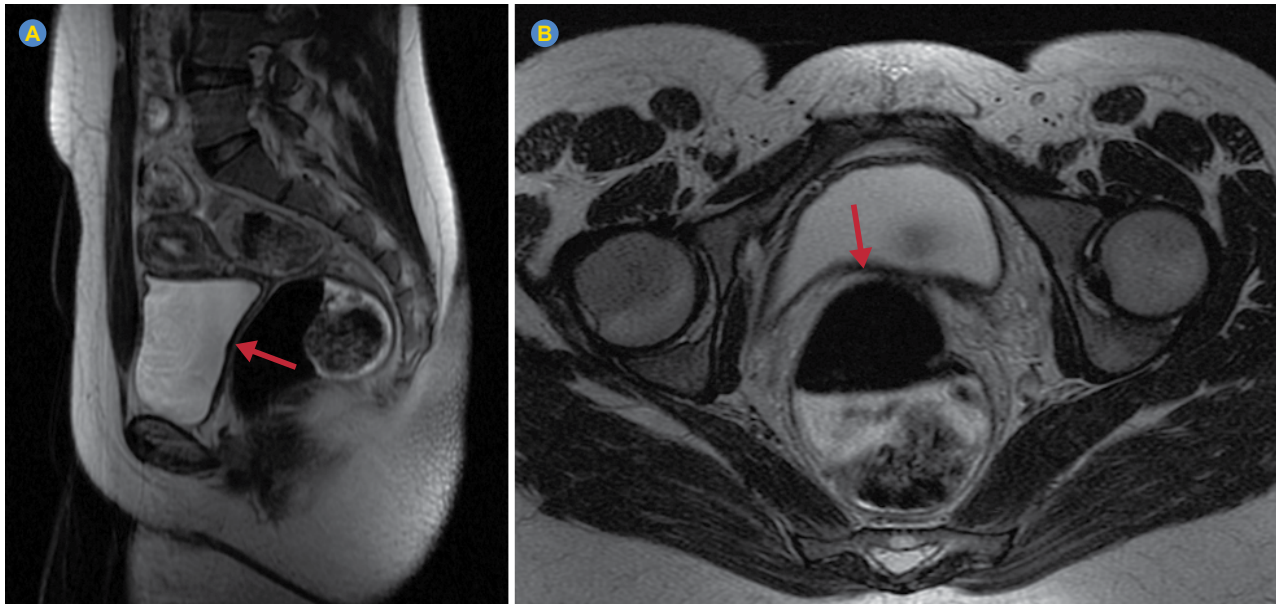

Figure 1 – Abdominal and pelvic MRI. The red arrows show the absence of normal vaginal musculature between the bladder and rectum. (A) Sagittal plane T2 image. Normal uterus is seen with a hypoplastic cervix. (B) Axial plane T2.

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AUTHOR CONTRIBUTIONS

ABS: Writing of the manuscript.

ASG, CR, HF: Writing and critical review of the manuscript.

PF: Study design, writing and critical review of the manuscript.

All authors approved the final version to be published.

PROTECTION OF HUMANS AND ANIMALS

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the Helsinki Declaration of the World Medical Association updated in October 2024.

DATA CONFIDENTIALITY

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

PATIENT CONSENT

Obtained.

CONFLICTS OF INTEREST

The authors have no conflicts of interest to declare.

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