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Case Report

Primary Umbilical Endometriosis in a 17-Year-Old Adolescent: Expanding the Clinical Spectrum of an Uncommon Entity

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ABSTRACT

Introduction: Primary umbilical endometriosis is a very uncommon condition characterized by the ectopic presence of endometrial structures in the umbilicus without any other endometriosis localization. Through this observation, we aim to highlight the clinical and paraclinical imaging of the condition and the modalities of its management.

Case presentation: We hereby report the case of a 17-year-old woman, who reported cyclic umbilical pain and bleeding. She exhibited no other symptoms and no additional endometriotic lesions were identified clinically or on MRI. The condition was successfully treated through excision of the endometriotic tissue, followed by plastic reconstruction of the umbilicus. Follow-up was uneventful.

Clinical discussion: Primary umbilical endometriosis is possibly caused by retrograde menstruation or lymphatic dissemination of endometrial cells. It presents as a painful, discolored umbilical mass with cyclical bleeding linked to menstruation. Diagnosis relies on imaging, such as MRI and histopathological confirmation, while surgical excision with wide margins is the standard treatment to minimize recurrence.

Conclusion: Raising awareness of primary umbilical endometriosis is crucial for its timely recognition and management. Incorporating cases into ambulatory surgery programs improves recovery, reduces costs and enhances patient satisfaction, highlighting the value of this approach.

Keywords: Primary umbilical endometriosis, Plastic reconstruction, Ambulatory surgery

1. Introduction

Endometriosis (EM) is defined by the ectopic presence of endometrial tissue outside the uterine cavity[1]. Umbilical endometriosis is a rare extra pelvic manifestation[2]. While endometriosis affects approximately 10% of the female population, this particular presentation accounts for only about 1% of all reported cases[1,2]. Primary umbilical endometriosis (PUE), first described by Villar in 1886, is even less common[2]. The exact pathogenesis remains unclear, with three main theories proposed to explain its development[3].

Therefore, we hereby present a very uncommon case of primary umbilical endometriosis. The patient, a 17-year-old woman, reported cyclic umbilical pain and bleeding. She exhibited no other symptoms and no additional endometriotic

lesions were identified clinically or on MRI. The condition was successfully treated through excision of the endometriotic tissue, followed by plastic reconstruction of the umbilicus.

2. Case Presentation

A 17-year-old woman, gravida 0, para 0, presented with a progressively enlarging umbilical mass over the past 11 months, associated with cyclic umbilical pain and bleeding. The bleeding was purplish and tender at the onset of menstruation, with the discharge being thick and brownish at the end of the menstrual period.

Clinical examination revealed a dark-colored, firm-to-firm nodule measuring 3 × 2 cm, involving the entire umbilicus **(Figure 1)**. It was not reducible with gentle bidigital pressure. An ultrasound examination showed a complex echogenic soft tissue lesion of 32 mm in vertical length, predominantly hypoechoic, located approximately 3 mm beneath the skin surface at the umbilicus. Abdominopelvic MRI revealed a 31 mm umbilical mass, hyperintense on T1-weighted sequences and hypointense on T2-weighted sequences, with no intraperitoneal communication or other endometriotic lesions detected **(Figure 2)**. The key clinical feature leading to the correct diagnosis of primary umbilical endometriosis was the cyclical association of umbilical nodule bleeding with her menstrual periods.

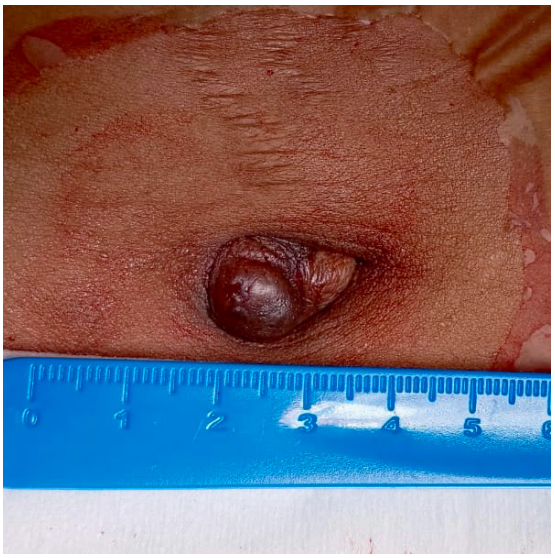

Figure 1: Clinical examination of the umbilical nodule.

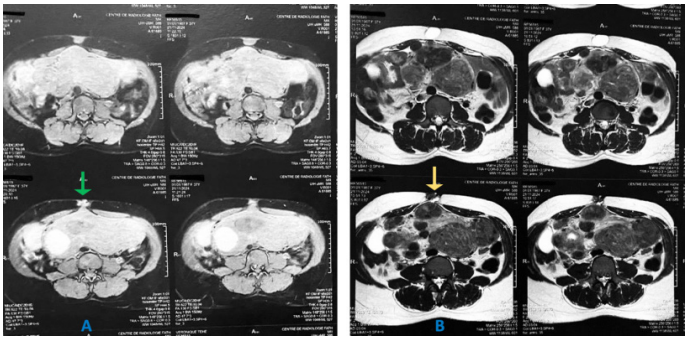

Figure 2: MRI imaging of the umbilical mass.

- **A:** T1-weighted sequence showing a 31 mm hyperintense umbilical mass (green arrow) without evidence of intraperitoneal communication.
- **B:** T2-weighted sequence showing the same umbilical mass as hypointense (yellow arrow), also without intraperitoneal communication.

The patient was offered surgical management, with an explanation of potential risks of recurrence and scar endometriosis. She successfully underwent excision of the nodule with umbilical reconstruction **(Figure 3)**. Histological examination of the surgical specimen revealed hyperplastic epidermal lining, mildly inflamed dermis and dermo hypodermal junction with fibrous scarring, a focus of cells suggestive of cytogenic stroma and glandular structures of endometrial type, consistent with an umbilical endometrioma. No epithelial atypia was observed and resection margins were clear.

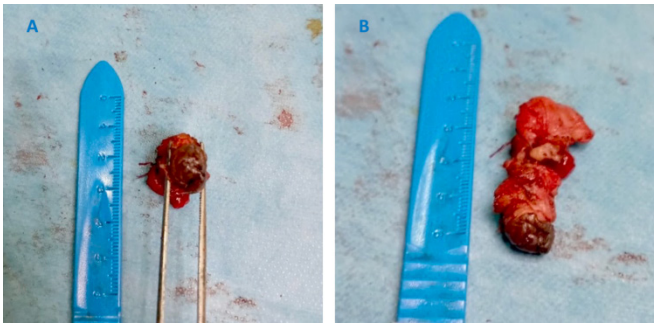

Figure 3: Postoperative photographs of the excised umbilical mass:

- **A:** Superior view showing a width of 20 mm.
- **B:** Lateral view showing a length of 52 mm.

The postoperative course was uneventful. A progestin-based contraceptive was initiated postoperatively. The patient was reviewed six weeks after surgery and was asymptomatic, with a normal-appearing umbilicus.

3. Discussion

Endometriosis is defined by the abnormal presence of functional endometrial tissue outside the uterine cavity[1]. Umbilical endometriosis, also known as Villar's nodule, is a rare extra-pelvic manifestation of endometriosis[2]. Its prevalence is estimated to be around 1% of all endometriosis cases[2]. It predominantly affects women during their reproductive years and is rare before menarche, tending to decrease after menopause. This is to our knowledge the first reported case of primary umbilical endometriosis in a 17-year-old adolescent, expanding the clinical spectrum of this rare entity.

Endometriosis is a chronic disease with a multifactorial etiology and its pathophysiological mechanisms remain partially elucidated[3]. Several theories have been proposed to explain the genesis of endometriosis. The Sampson theory, currently the most widely accepted, suggests that endometriosis arises from viable endometrial cells refluxing through the fallopian tubes during menstruation and implanting on the peritoneal surface and pelvic organs[4]. The Meyer theory proposes that endometriosis originates from a metaplastic process, where cells derived from the coelomic epithelium undergo metaplasia into endometrial-like cells under the influence of various infectious, toxic or hormonal factors[5].

The lymphatic and vascular metastasis theory proposes that endometriosis may result from the dissemination of endometrial cells via lymphatic and hematogenous routes[6]. Lymphatic metastasis to distant sites such as the umbilicus, retroperitoneal space and lower limbs is anatomically possible due to lymphatic communication between these structures and the endometrium[6]. In our case, both the Sampson theory and lymphatic metastasis

could explain the umbilical localization. A systematic literature review on umbilical endometriosis, released in February 2022, suggested that primary umbilical endometriosis could develop from the implantation of regurgitated endometrial cells[7]. These cells are carried by the clockwise movement of peritoneal circulation to the right hemidiaphragm and then funneled toward the umbilicus by the falciform and round ligaments of the liver[7].

Umbilical endometriosis presents as a painful umbilical mass with cyclical bleeding corresponding to the menstrual cycle. The cyclical nature coinciding with menstruation is fundamental and sometimes sufficient to suggest the diagnosis[2]. Ultrasound, computed tomography (CT) and magnetic resonance imaging (MRI) are utilized to assist in diagnosing umbilical endometriosis[8]. The imaging characteristics are not specific and depend on several factors, including the phase of the menstrual cycle, the degree of inflammatory response and the distribution between stromal and glandular components. MRI is particularly useful for confirming the presence of other locations, especially pelvic ones[9].

The diagnosis of primary umbilical endometriosis is confirmed through histopathological examination of the surgical specimen, which reveals ectopic endometrial glands, stroma and muscle fibers[10]. Surgical excision with wide margins is the standard treatment to reduce recurrence risk[11]. We prefer utilizing a monopolar electrosurgical unit during excision to help cauterize any clinically undetected endometriotic implants. The abdominal wall is closed in anatomical layers and the umbilical depression is reconstructed using a resorbable suture attaching the dermis to the aponeurosis of the rectus muscles.

Differential diagnoses for umbilical endometriosis include pyogenic granuloma, hernia and pemphigus vegetans. Due to its variable macroscopic appearance, these lesions may initially be mistaken for malignant tumors, such as melanoma[12]. In a case series, a patient presented with an umbilical hernia associated with the nodule, which was only diagnosed during surgery[12]. Another reported case of primary umbilical endometriosis was associated with a large irreducible umbilical hernia that was treated in the same surgery[13].

Since the introduction of ambulatory surgery in our facility, we have integrated our patient into our day hospital program, allowing for same-day discharge. The use of simple regional anesthesia techniques, such as spinal or epidural blocks, enables effective pain management during and after the procedure, facilitating early mobilization and reducing the need for postoperative analgesics[14]. This approach not only improves patient satisfaction but also contributes to cost savings by decreasing recovery room time and minimizing hospital admissions[14]. However, it is regrettable that the adoption of ambulatory surgery has not progressed as rapidly as it should worldwide[14,15]. It should be encouraged and highlighted whenever utilized, as in our current case, to promote its numerous benefits for patients and healthcare systems.

4. Conclusions

PUE is an exceptionally rare form of endometriosis. By increasing awareness of this uncommon presentation as a potential diagnosis of a painful, discolored umbilical tumefaction, we hope that this condition will be optimally recognized and managed.

This case report has been reported in line with the SCARE Criteria[16].

5. Abbreviations

EM: Endometriosis

PUE: Primary Umbilical Endometriosis

6. Declarations

6.1. Conflicts of interest

The authors declare that they have no competing interests.

6.2. Authors' contribution

AS: study concept and design, data collection, data analysis and interpretation, writing the paper. YE: study concept, data collection, data analysis, writing the paper. KE: study concept, data collection, data analysis, writing the paper. HL: study concept, data collection, data analysis, writing the paper. OEH: study concept, data collection, data analysis, writing the paper. SM: study design, data collection, data interpretation, writing the paper. AB: study design, data collection, data interpretation, writing the paper.

6.3. Sources of funding

There are no funding sources to be declared.

6.4. Ethical approval

Ethical clearance was not required for this case report as it involved only a single patient and did not involve any experimental or invasive procedures.

6.5. Consent

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

6.6. Guarantor of submission

The corresponding author is the guarantor of submission.

7. Acknowledgements

None.

7.1. Availability of data and materials

Supporting material is available if further analysis is needed.

7.2. Provenance and peer review

Not commissioned, externally peer-reviewed.

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