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Case Report

Primary Umbilical Endometriosis (Villar's Nodule) Coexisting with Deep Infiltrating Endometriosis: A Case Report and Diagnostic Challenge

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ABSTRACT

Introduction: Endometriosis is a chronic gynecological condition affecting over 10% of women of reproductive age, characterized by endometrial-like tissue outside the uterus, causing pelvic pain, dysmenorrhea, and infertility. While typically pelvic, rare extrapelvic manifestations such as primary umbilical endometriosis present as painful umbilical nodules, often mistaken for abscesses or hernias. This report emphasizes that the presence of an umbilical nodule should raise high suspicion for concomitant deep infiltrating endometriosis (DIE).

Case Presentation: A 41-year-old woman presented with cyclical umbilical bleeding and a tender 2 cm umbilical mass, alongside longstanding dysmenorrhea. She had no prior surgeries. Ultrasound revealed a 3.75 cm ovarian endometrioma. Laparoscopy confirmed umbilical endometriosis (Villar's nodule) with more extensive pelvic disease than anticipated, including uterosacral ligament lesions and suspected adenomyosis, consistent with DIE. The umbilical nodule and ovarian endometrioma were resected, and histopathology confirmed ectopic endometrial tissue. She received six months of Leuprolide Acetate and remained asymptomatic with no recurrence at one year.

Conclusion: This case highlights that an umbilical nodule may indicate more extensive pelvic disease and should prompt strong suspicion for concomitant DIE, as demonstrated by laparoscopy. Early recognition supports comprehensive evaluation and appropriate surgical management.

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INTRODUCTION

Endometriosis is a chronic gynecological condition where tissue resembling the uterine lining grows outside the uterus, affecting millions of women of reproductive age worldwide [1]. Its global prevalence is estimated at 10%, though rates vary significantly. In Indonesia, hospital-based data show a wide prevalence range of 13.6% to 69.5% among infertile women. The condition commonly presents with symptoms like chronic pelvic pain, painful periods (dysmenorrhea), and infertility, which tend to worsen in advanced stages [2-5].

The pathogenesis of endometriosis is complex and not fully understood, though several theories exist, including retrograde menstruation, hormonal imbalance, and immune dysfunction. This leads to a chronic inflammatory state within the peritoneal cavity, driven by factors

like iron overload and oxidative stress. This inflammation is a key contributor to the severe symptoms, such as abdominal cramping, and underscores the significant clinical and socioeconomic burden of the disease [6].

Endometriotic lesions most frequently occur on the ovaries and pelvic ligaments but can also be found in extrapelvic sites. A rare form of this is primary umbilical endometriosis, where endometrial tissue develops in the umbilicus without prior surgical scarring. First described in 1886, it accounts for a very small fraction of all endometriosis cases, clinically presenting as a painful, discolored nodule that often worsens during menstruation [7,8].

Due to its rarity and nonspecific appearance, umbilical endometriosis is frequently misdiagnosed as other conditions like abscesses or hernias, highlighting the need for a high index of suspicion. Accurate diagnosis is crucial to rule out malignancy and ensure proper management. This report aims to enhance awareness and understanding of this rare manifestation to improve its recognition and treatment in clinical practice.

CASE PRESENTATION

This study presents a descriptive, retrospective analysis based on a comprehensive review of a single patient's medical records. The data collected and examined encompass a wide array of clinical information, including detailed documentation of clinical findings, results from various laboratory examinations, and a complete set of radiological imaging studies. The primary objective of this case review was to systematically analyze the clinical presentation, diagnostic journey, and therapeutic outcomes. This study was approved by the Institutional Review Board of Brawijaya University, and written informed consent was obtained from the patient, permitting the use of their data for research and publication while maintaining anonymity.

A 41-year-old married female, Mrs.R presented to the Gynecology outpatient clinic at Saiful Anwar General Hospital in November 2024. Her initial complaint dated back to January 2022, when she began experiencing persistent abdominal pain during menstruation, a symptom she had managed at home without seeking medical consultation. The situation changed in October 2024, when her cyclical abdominal pain was accompanied by a new and distressing symptom: bleeding from her umbilicus. This prompted her to visit Dr. M. Saleh Regional General Hospital in Probolinggo, where a clinical examination revealed a two-centimeter blackish mass at the umbilicus. A suspicion of umbilical endometriosis was raised, leading to her referral for specialized care.

A review of systems was unremarkable, with no reported weight loss, leukorrhea, or changes in bowel or bladder habits. Her past medical history was negative for chronic conditions such as diabetes, hypertension, or asthma. Notably, she had no prior surgical history, making a scar-related etiology for the umbilical lesion unlikely. Her family history was also negative for tumors or gynecologic malignancies. Her menstrual history began at menarche at age 14, with regular cycles every 28–30 days lasting 5–7 days. She reported a long-standing history of dysmenorrhea since menarche, suggestive of underlying endometriosis. She married at age 22, and her last menstrual period began on November 2, 2024.

Investigation

Upon physical examination, the patient appeared moderately ill but was fully conscious and cooperative. Vital signs were stable: blood pressure 128/76 mmHg, heart rate 83 bpm, respiratory rate 20 bpm, oxygen saturation 98% on room air, and she was afebrile at 36.5°C. Her BMI was 23.1 kg/m², within normal limits. The general physical examination was otherwise unremarkable, with no signs of anemia or jaundice, and respiratory and cardiovascular exams were normal. Abdominal examination revealed a well-circumscribed, firm, and exquisitely tender nodule measuring approximately 2x2 cm located within the umbilical ring. The mass displays a characteristic multilobulated appearance with a variegated deep violaceous to brownish-black pigmentation, suggestive of trapped hemorrhagic content. There is no evidence of active bleeding, purulent discharge, or surrounding erythema at the time of

examination. A speculum and bimanual vaginal examination showed no abnormal discharge, smooth vaginal walls, and a closed, smooth cervical os. The uterine size was estimated to be approximately eight to ten weeks of gestation, with no adnexal masses or tenderness appreciated (Figure 1).



Figure 1. Clinical Picture of Patient with Umbilical Nodule (Red Arrow)

A transabdominal ultrasound was performed as an initial imaging study. While this examination is a useful screening tool, it is not the optimal modality for a comprehensive evaluation of endometriosis, particularly for deep-infiltrating disease. The ultrasound showed a uterus in anteflexed position, measuring approximately 5.09 by 3.84 centimeters, which is within normal limits. A key finding was a cystic mass with internal echoes, measuring 3.75 by 3.60 centimeters, located in the region of the ovary. This finding was suggestive of an endometrioma, commonly referred to as a chocolate cyst. Importantly, the examination did not document or assess for involvement of the bowel, rectovaginal septum, or uterosacral ligaments, and the sliding sign was not documented. A more sensitive preoperative imaging study, such as a targeted transvaginal ultrasound for endometriosis mapping or a pelvic magnetic resonance imaging, was not performed prior to the surgical intervention.

Based on the comprehensive clinical picture, which integrated the patient's history of long-standing dysmenorrhea, the pathognomonic symptom of cyclical umbilical bleeding, and a visible umbilical mass, along with the ultrasound finding of an ovarian endometrioma, a clinical diagnosis was established. The patient was diagnosed with umbilical endometriosis (Villar's nodule), ovarian endometrioma, and suspected pelvic endometriosis.

It is important to note that while the ovarian and umbilical involvement were clinically evident, the full extent of her pelvic disease was not fully mapped preoperatively. The presence of an umbilical lesion, a form of extragenital endometriosis, strongly suggests the possibility of more extensive pelvic disease.

Treatment

The patient was scheduled for elective surgical management under general anesthesia. A written informed consent was obtained prior to the procedure. An exploratory laparoscopy was performed on November 26, 2024, with the dual purpose of confirming the diagnosis and providing definitive treatment. Intraoperatively, the surgical findings were more extensive than what was suggested by the preoperative ultrasound. The uterus appeared enlarged, consistent with an eight-to ten-week gestation size, and showed signs suggestive of adenomyosis. Endometriotic lesions were visualized and confirmed on both the right and left uterosacral ligaments, a finding that had not been documented on the routine transabdominal ultrasound. The previously identified ovarian endometrioma was confirmed, and a cystectomy was successfully performed. Notably, both fallopian tubes appeared within normal limits, with no evidence of hydrosalpinx or significant adhesions.

Following the pelvic component of the surgery, the umbilical mass was resected, and the umbilicus was surgically repaired. Tissue specimens from both the pelvis and the umbilicus were sent for histopathological analysis. The pathology report, dated November 28, 2024, confirmed the clinical diagnosis. The pelvic

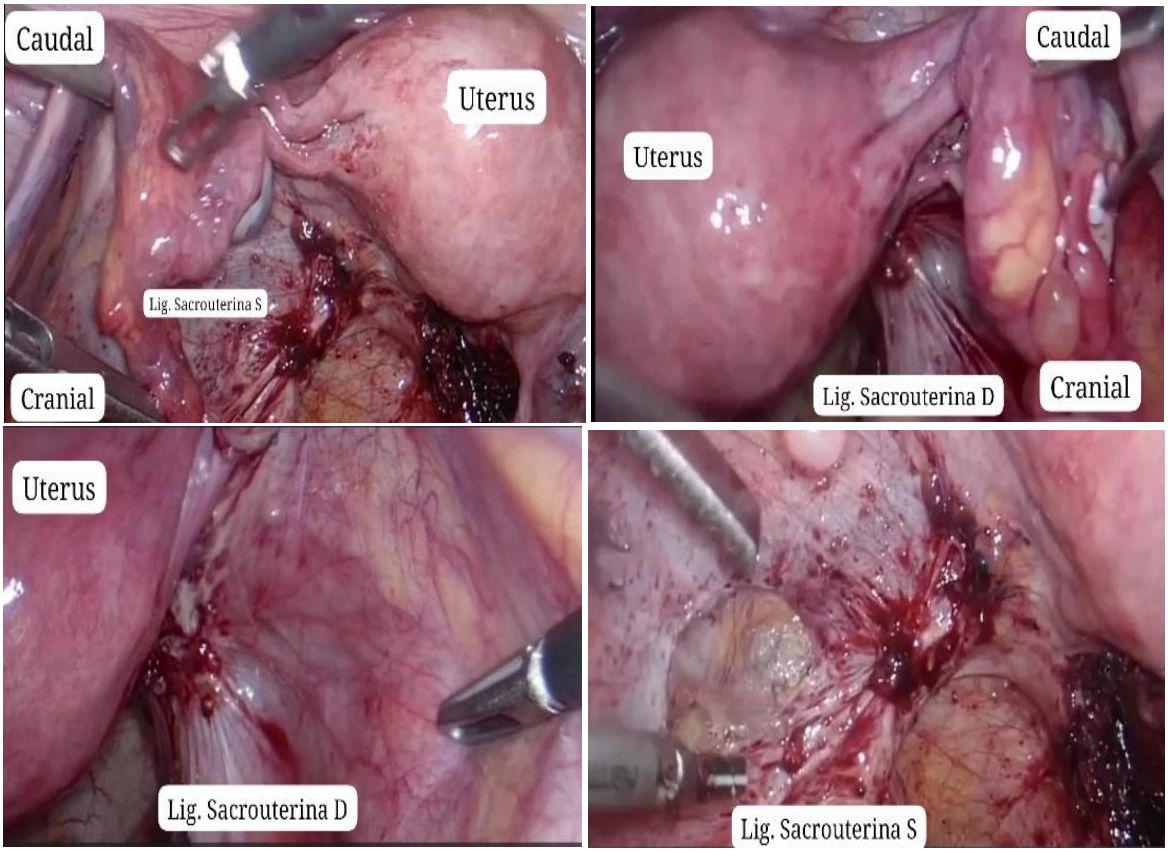


Figure 2. Intraoperative Finding Documentations

nodule was identified as ectopic endometrial tissue, characterized by the presence of endometrial glands and stroma outside the uterine cavity. The diagnosis of umbilical endometriosis was confirmed by histopathological examination of the umbilical specimen, which revealed endometrial glands and stroma with hemosiderin-laden macrophages and surrounding fibrosis, characteristic of an ectopic endometriotic lesion. No evidence of atypia or malignancy was found. These findings confirm Villar's nodule, providing definitive confirmation of this rare form of extragenital endometriosis, and support the presence of concurrent pelvic endometriosis. The surgical intervention was successful in both removing the visible disease and providing a histopathological diagnosis, addressing the patient's primary complaints, and confirming the full extent of her condition (Figure 2).

Outcomes and Follow Up

The patient underwent a laparoscopic resection of sacrouterine and umbilical endometriosis. Postoperatively, she received six cycles of Leuprolide Acetate (3.75 mg injections) from December 2024 to May 2025. At each monthly follow-up visit, she remained asymptomatic, with a stable general appearance and normal vital signs. Physical examinations consistently revealed a well-healed umbilical scar with no tenderness, a soft abdomen, and no abnormal genital findings.

A final evaluation in June 2025, including an abdominal ultrasound, was within normal limits, with no evidence of pathological lesions in the uterus or adnexa. The patient reported no complaints, indicating a successful postoperative outcome with no signs of recurrence at that time.

DISCUSSION

Primary umbilical endometriosis, also known as Villar's nodule, is a rare form of extrapelvic endometriosis, representing approximately 0.5–1% of all endometriosis cases. Clinically, it is characterized by a reddish-purple or bluish-black umbilical nodule accompanied by cyclic pain and bleeding that coincides with menstruation. These catamenial symptoms are a key diagnostic clue, and their presence should raise strong clinical suspicion for Villar's nodule. In this patient, a 2 × 2 cm umbilical mass was observed, with a clear history of bleeding that occurred predominantly during menstruation. The combination of a visible umbilical lesion and cyclic symptoms strongly supports the diagnosis of umbilical endometriosis. Furthermore, the literature indicates that umbilical endometriosis may coexist with pelvic endometriosis in a significant proportion of cases. Therefore, the identification of an umbilical nodule should prompt a thorough evaluation of the pelvis to rule out concurrent disease, as early detection of pelvic involvement can guide more comprehensive management [8].

A thorough history taking and physical examination are essential in establishing the diagnosis of endometriosis. A case–control study from the United Kingdom involving more than 5,000 patients with endometriosis found that affected individuals were more likely to experience dysmenorrhea, dyspareunia or postcoital bleeding, abdominopelvic pain, menorrhagia, and a history of subfertility [9].

The differential diagnosis for an umbilical nodule includes umbilical hernia, granuloma/pyogenic granuloma, and Sister Mary Joseph nodule. An umbilical hernia was ruled out in this case as the nodule was non-reducible and exhibited cyclic symptoms, unlike a typical hernia. Granuloma was also unlikely given the absence of trauma or local inflammation and the presence of cyclic bleeding, which is not characteristic of such lesions. Sister Mary Joseph nodule, an umbilical metastasis from an intra-abdominal malignancy, was considered but excluded due to the absence of systemic symptoms like weight loss and, definitively, by histopathological examination, which confirmed endometriosis without atypia. This case underscores that while clinical presentation can guide the diagnosis, histopathological analysis following lesion excision remains the gold standard for confirmation [8,10].

In this case, the patient has no history of abdominal surgery or umbilical trauma, thus falling into the category of primary (spontaneous) umbilical endometriosis. Two main theories have been proposed to explain the mechanism behind this spontaneous form. First, the metastatic theory states that endometrial cells can spread through the vascular (haematogenous) or lymphatic pathways from the uterus to ectopic locations. This theory is supported by the discovery of endometrial cells within lymphatic and venous blood vessels in histopathological specimens, as well as reports of endometriosis cases in very distant locations such as the lungs, pleura, and brain without any direct anatomical connection to the pelvis. Second, the celomic metaplasia theory states that primitive mesothelial cells or multipotent stem cells remaining in the umbilical region can differentiate into ectopic endometrial tissue due to chronic hormonal stimulation (especially estrogen) or local inflammatory mediators. This theory better explains why endometriosis can be found in men undergoing estrogen therapy, in foetuses, and in patients with uterine agenesis.

In our patient, given the absence of a surgical history and the precise location of the lesion at the fundus umbilicus (not in the ligamentum tract), the theory of coelomic metaplasia seems more likely, although the contribution of haematogenous migration cannot be completely ruled out. The combination of these two mechanisms, known as the dual origin theory may play a role in this case [11-13].

The European Society of Human Reproduction and Embryology (ESHRE) recommends transvaginal ultrasonography as the first-line imaging for endometriosis, with targeted evaluation of the posterior compartment essential for diagnosing deep infiltrating endometriosis (DIE) [14]. In this case, preoperative ultrasound identified an ovarian endometrioma and umbilical nodule, but specific DIE mapping was not performed. In this patient, diagnostic evaluation for suspected deep infiltrating endometriosis (DIE) of the posterior

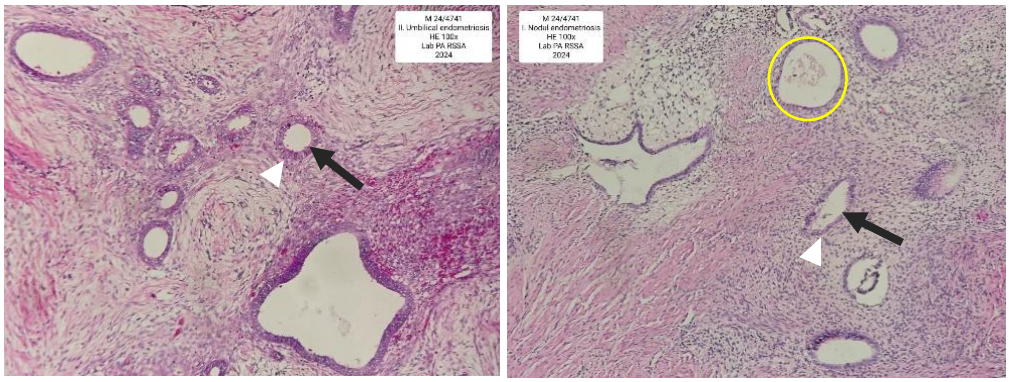


Figure 3. Histopathological Findings of Endometriosis**.** (a) Histopathological confirmation of umbilical endometriosis. Photomicrograph of the umbilical nodule specimen showing endometrial glands (black arrows) surrounded by cellular endometrial stroma (white arrowheads) infiltrating the subcutaneous adipose tissue. Hematoxylin and eosin (HE) staining. Original magnification: ×100. Scale bar = 200 µm. No evidence of malignancy or atypia was observed. (b) Histopathological confirmation of ovarian endometrioma. Photomicrograph of the endometrioma cyst wall demonstrating an inner lining of endometrial-type columnar epithelium (black arrows) with underlying endometrial stroma (white arrowheads). Note the presence of hemosiderin-laden macrophages (yellow circles) within the stroma, indicating chronic cyclic hemorrhage. Hematoxylin and eosin (HE) staining. Original magnification: ×100. Scale bar = 200 µm.

compartment was performed using transabdominal ultrasound (TAUS). However, based on a systematic review by Capraş et al. (2025), it is stated that "TVUS is consistently reported as a reliable and cost-effective imaging modality and, in line with international guidelines, should be considered the first-line option for posterior compartment DIE." This statement implicitly asserts that the first-line modality according to international guidelines is TVUS, while TAUS is not mentioned at all in those recommendations. Thus, the use of TAUS in this case is not recommended for the diagnosis of DIE because this modality does not have diagnostic performance equivalent to TVUS and is not recognised as a primary option in international guidelines [15]. Consequently, uterosacral ligament involvement was only confirmed intraoperatively, highlighting the need for comprehensive preoperative DIE assessment in patients with extrapelvic endometriosis, given the frequent coexistence of pelvic and extrapelvic disease.

The diagnosis of umbilical endometriosis was confirmed by histopathological examination. The specimen revealed endometrial glands and stroma with hemosiderin-laden macrophages and surrounding fibrosis, characteristic of an ectopic endometriotic lesion. No evidence of atypia or malignancy was found. These findings confirm Villar's nodule and support the presence of concurrent pelvic endometriosis, underscoring the essential role of histopathology in distinguishing this condition from other umbilical lesions such as granuloma, hernia, or Sister Mary Joseph nodule [8] (Figure 3).

Medical therapy using GnRH agonists (e.g., Leuprolide Acetate) works by suppressing the hypothalamic-pituitary-ovarian axis, creating a temporary state of hypoestrogenism. However, in umbilical endometriosis, this approach is only temporarily palliative. It induces atrophy of ectopic endometrial tissue during treatment but fails to eliminate the embedded endometrial stem cells within the subcutaneous tissue. Consequently, recurrence almost always occurs within 6–12 months after stopping therapy. Medical therapy also has two other significant drawbacks: 1) it does not provide histopathological confirmation (the gold standard for diagnosis), meaning the rare potential for malignant transformation (incidence <1%) can be

missed especially in extrapelvic disease; and 2) it causes systemic side effects such as hot flashes, osteoporosis, and mood disturbances, which can lower the patient's quality of life. Given these limitations, total surgical excision (via laparoscopy or open local excision) with healthy tissue margins is the treatment of choice for symptomatic umbilical nodules. The surgical approach offers definitive advantages: complete removal allows for permanent healing, histopathological specimens provide absolute diagnostic confirmation, and the procedure is minimally invasive with a short hospital stay. In the case described (a 2 cm nodule, cyclic pain disrupting daily activities, and the patient's desire for a definitive solution), surgical excision was therefore chosen as first-line therapy [16,17].

The patient successfully underwent resection of the umbilical mass with clear margins followed by primary repair, achieving macroscopic clearance of the lesion. In addition to local excision, a simultaneous diagnostic laparoscopy was performed to evaluate the pelvis for concomitant disease. This combined approach is clinically important, as umbilical endometriosis frequently coexists with pelvic endometriosis and may indicate more extensive involvement of structures such as the ovaries, peritoneum, or uterosacral ligaments. Performing both procedures in a single operative setting allows for comprehensive staging and management of disease at all sites, reduces the need for future interventions, and ensures optimal patient outcomes [8].

This case demonstrates the coexistence of umbilical endometriosis with pelvic disease, including an ovarian endometrioma and bilateral uterosacral ligament lesions (r-ASRM Stage III, #Enzian B2). Several theories explain this association. The lymphovascular theory proposes benign metastasis of endometrial cells to distant sites, similar to the mechanism of Sister Mary Joseph nodules. Alternatively, the coelomic metaplasia theory suggests that hormonal stimulation transforms embryonic mesothelial remnants into endometrial tissue, which may explain lesions developing without a direct anatomical connection [8,18].

The reported coexistence of umbilical and pelvic endometriosis ranges from 13% to 80%, suggesting that an umbilical nodule may serve as a clinical marker for posterior compartment involvement rather than a definitive indicator of disease stage. In this patient, the presence of an ovarian endometrioma and uterosacral lesions confirms moderate pelvic disease. However, evidence does not consistently link umbilical endometriosis to advanced severity, as it occurs across all stages. Therefore, every patient with umbilical endometriosis should undergo comprehensive pelvic evaluation, including targeted imaging or laparoscopy, to assess for clinically significant posterior compartment disease that may not be apparent on initial examination [8,19,20].

CONCLUSION

Umbilical endometriosis is a rare but clinically significant entity that should be regarded as a sentinel lesion, a surface marker for potentially hidden deep infiltrating endometriosis within the pelvis. The literature indicates that 30 to 50 percent of patients with spontaneous umbilical endometriosis have coexisting pelvic deep infiltrating endometriosis, most commonly involving the uterosacral ligament, rectovaginal septum, and ovaries. As illustrated by the present case of a 41-year-old woman with concurrent uterosacral and umbilical endometriosis, this association supports theories of lymphovascular dissemination or metaplasia and highlights that an umbilical nodule may signal more extensive pelvic disease.

Given these implications, we recommend a specific clinical approach for every reproductive-age woman presenting with a cyclic, painful umbilical nodule. First, clinicians should perform a targeted history taking for pelvic deep infiltrating endometriosis symptoms, including deep dyspareunia, severe dysmenorrhea, cyclic defecation pain, and infertility. Second, a pelvic magnetic resonance imaging with an endometriosis protocol should be obtained to detect occult pelvic lesions. Third, referral for diagnostic and therapeutic laparoscopy is advised rather than isolated umbilical nodule excision, as surgical management aims to alleviate symptoms and reduce recurrence.

The key practical message is that an umbilical nodule should never be considered an isolated entity. Management focused solely on nodule excision without pelvic evaluation risks missing clinically significant deep infiltrating endometriosis lesions, which can progress to hydronephrosis, infertility, or severe chronic pelvic pain. Adopting this sentinel lesion algorithm enables clinicians to provide comprehensive, definitive care. This case underscores the complexity of endometriosis and reinforces the need for clinical vigilance and thorough evaluation in affected patients.

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CONFLICT OF INTEREST

The authors confirm that there are no conflicts of interest related to this study. Additionally, the patient has provided informed consent for the use and publication of her medical information.

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