

Deep Infiltrating Endometriosis With Rectal Wall Involvement Diagnosed By Rectal Endoscopic Ultrasound: A Case Report

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Abstract

Background: Deep infiltrating endometriosis (DIE) involving the rectal wall is an uncommon manifestation that can mimic colorectal malignancy or inflammatory bowel disease. Early and accurate diagnosis is critical for guiding optimal management.

Case Presentation: A 37-year-old female presented with a six-month history of pelvic pain and dysmenorrhea. Pelvic MRI revealed a hypointense lesion on T2-weighted images involving the anterior rectal wall, suspicious for deep endometriosis. Rectal endoscopic ultrasound (EUS) confirmed a hypoechoic heterogeneous lesion infiltrating the muscularis propria. Fine-needle biopsy under EUS guidance demonstrated endometrial glands and stroma within the rectal wall, confirming the diagnosis of endometriosis. The patient was managed medically with hormonal therapy, resulting in significant improvement of symptoms.

Conclusion: This case highlights the diagnostic value of EUS in confirming rectal wall endometriosis, complementing MRI findings and facilitating appropriate non-surgical management.

Keywords: Deep infiltrating endometriosis, Rectal endometriosis, Endoscopic ultrasound, Pelvic pain, Dysmenorrhea

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I. Introduction:

Endometriosis is characterized by the presence of functional endometrial tissue outside the uterine cavity and affects approximately 10% of women of reproductive age [1]. Deep infiltrating endometriosis (DIE) represents the most severe form, defined by lesions extending more than 5 mm beneath the peritoneal surface [2]. Bowel involvement occurs in about 5–12% of cases, most frequently affecting the rectosigmoid colon [3,4].

Clinical presentation varies, and rectal endometriosis can mimic irritable bowel syndrome, inflammatory bowel disease, or even colorectal malignancy. Imaging modalities such as magnetic resonance imaging (MRI) and endoscopic ultrasound (EUS) have greatly improved the preoperative diagnosis of DIE.

We present a case of rectal wall endometriosis confirmed by EUS-guided biopsy, emphasizing the complementary diagnostic role of MRI and EUS in the evaluation of bowel endometriosis.

II. Case Presentation:

A 37-year-old female presented with a six-month history of pelvic pain and progressive dysmenorrhea. There was no rectal bleeding, change in bowel habits, or weight loss. Her menstrual cycles were regular. She had no prior surgeries or significant family history.

On physical examination, abdominal findings were unremarkable. Digital rectal examination elicited mild tenderness in the anterior rectal wall without a palpable mass.

Laboratory investigations, including complete blood count and inflammatory markers, were within normal limits. Serum CA-125 was mildly elevated at 48 U/mL (reference <35).

MRI of the pelvis demonstrated a T2-hypointense, T1-iso- to hyperintense lesion measuring approximately 2.2×1.8 cm along the anterior rectal wall, 12 cm from the anal verge. The lesion caused focal thickening and mild retraction of the adjacent uterine posterior serosa. These findings were suggestive of deep infiltrating endometriosis with possible rectal muscularis involvement. Colonoscopy was performed and doesn't show any abnormal findings.

Rectal EUS was performed using a high-frequency linear probe. It revealed a heterogeneous, hypoechoic, poorly circumscribed lesion involving the muscularis propria layer of the rectal wall, with preservation of the mucosa and submucosa. No lymphadenopathy was seen (figure 1).

EUS-guided fine-needle biopsy was performed using a 22-gauge needle, obtaining core samples.

Microscopic examination demonstrated endometrial glands and stroma embedded within fibromuscular tissue, confirming endometriosis of the rectal wall. There was no evidence of malignancy.

Given the absence of obstructive symptoms and the moderate size of the lesion, the patient was managed medically with progestin-based hormonal therapy. Over the subsequent three months, her pelvic pain and dysmenorrhea improved substantially, and follow-up imaging showed stable lesion size. She continues under gynecologic and gastroenterologic follow-up.

III. Discussion:

Endometriosis affects around 10% of women of reproductive age and represents a major cause of chronic pelvic pain and infertility [1]. Deep infiltrating endometriosis (DIE), defined by lesions extending more than 5 mm beneath the peritoneum, is the most aggressive form [2]. Bowel involvement, particularly of the rectosigmoid colon, occurs in approximately 5–12% of cases [3,4].

The exact mechanism remains multifactorial. Sampson's theory of retrograde menstruation remains the most widely accepted, complemented by coelomic metaplasia and lymphatic dissemination [5]. Recurrent hemorrhage and inflammation within ectopic endometrial foci lead to fibrosis and muscular infiltration.

Patients typically present with cyclical pelvic pain, dysmenorrhea, dyspareunia, and dyschezia, but bowel symptoms can be absent, as in this case, making diagnosis challenging [6].

Magnetic Resonance Imaging (MRI) plays a central role in mapping pelvic disease. DIE nodules usually appear hypointense on T2-weighted images due to their fibrotic composition, and may show small hyperintense foci on T1-weighted images indicating hemorrhagic content [7,8]. MRI provides excellent delineation of lesion extent and its relationship to pelvic organs.

However, MRI can sometimes underestimate mucosal invasion or fail to define precise rectal wall layer involvement. In such cases, Endoscopic Ultrasound (EUS) offers superior resolution for evaluating rectal wall stratification [9].

EUS is particularly useful in determining whether lesions involve the muscularis propria alone or extend to the submucosa or mucosa, influencing surgical strategy [10]. In our case, EUS-guided biopsy confirmed rectal wall invasion, allowing definitive diagnosis without surgery.

While EUS is traditionally used for staging rectal tumors, its role in diagnosing rectal endometriosis has gained recognition. The ability to obtain real-time, layer-specific imaging and perform fine-needle biopsy makes it invaluable when MRI findings are inconclusive [9,10].

Histological confirmation remains the diagnostic gold standard, and in this case, EUS-guided biopsy provided a minimally invasive route for confirmation, avoiding surgical morbidity.

Management of rectal endometriosis should be individualized based on symptom severity, depth of invasion, and fertility goals.

Medical treatment—such as progestins, combined oral contraceptives, or GnRH analogues—is effective for pain control and may reduce lesion activity [11]. Surgical intervention (e.g., rectal shaving, discoid excision, or segmental resection) is reserved for patients with obstruction, severe pain, or infertility [12]. In our patient, hormonal therapy provided substantial symptomatic relief, supporting a conservative approach.

Medical management often alleviates pain but does not eradicate lesions, necessitating follow-up imaging and symptom monitoring.

MRI or EUS can be repeated periodically to assess stability. Early detection and multidisciplinary collaboration among gastroenterologists, gynecologists, and radiologists are essential to optimize outcomes [12,13].

Imaging findings of rectal wall thickening may mimic colorectal carcinoma or Crohn's disease. However, endometriosis typically spares the mucosa and has characteristic cyclical symptomatology. MRI's high soft-tissue contrast and EUS's wall-layer analysis help distinguish these entities [13].

IV. Summary

This case underscores the complementary roles of MRI and EUS in diagnosing rectal wall endometriosis. EUS not only delineates the depth of wall invasion but also enables tissue confirmation through biopsy, which is particularly valuable when surgery is not immediately indicated.

V. Conclusion

Deep infiltrating endometriosis of the rectal wall is a challenging diagnosis due to nonspecific symptoms and overlap with other gastrointestinal disorders. This case highlights the importance of a multimodal imaging approach, with EUS-guided biopsy providing definitive diagnosis and guiding appropriate medical management. Early identification and coordinated multidisciplinary care can significantly improve patient quality of life and avoid unnecessary surgery.

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Figure 1: Rectal EUS using linear probe. Showing a heterogeneous, hypoechoic, poorly circumscribed lesion involving the muscularis propria layer of the rectal wall, with preservation of the mucosa and submucosa.