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# Rectal endometriosis without typical gynecological symptoms mimics gastrointestinal stromal tumors: a case report and literature review

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Rectal endometriosis may be similar to gastrointestinal stromal tumors (GIST), especially in women without typical gynecological or cyclical gastrointestinal symptoms. A 45-year-old female patient had an eight-year history of rectal defecation difficulty and increased defecation frequency, but no dysmenorrhea or increased menstrual flow. Colonoscopy revealed a smooth protruding lesion on the rectal surface, and endoscopic ultrasound showed a heterogeneous lesion within the muscular layer, with intact surface mucosa, suggesting the possibility of GIST. A laparoscopic anterior resection was performed. Postoperative histopathology showed ectopic endometrial glands and stroma, combined with positive estrogen and progesterone receptors and expression of CD10 in the stroma, confirming rectal endometriosis. The patient's symptoms improved significantly and recovered well. Subsequently, this study reviewed 12 reported cases, and the results showed that intestinal endometriosis and gastrointestinal tumors have overlapping diagnoses in terms of diagnosis. Even without periodic symptoms, the possibility of rectal endometriosis should be considered for women with rectal submucosal lesions. A comprehensive assessment of clinical history, pelvic anatomical relationship, imaging examinations, and histological evaluation should be conducted to establish the diagnosis and guide treatment.

KEYWORDS
**case report, differential diagnosis, endoscopic ultrasonography, gastrointestinal stromal tumor, rectal endometriosis**

## 1 Introduction

Endometriosis (EM) is a common benign gynecological disease characterized by the ectopic growth of functional endometrium-like tissues outside the uterine cavity, which can lead to chronic pelvic pain, infertility and related organ dysfunction (1). Although endometriosis mainly involves the pelvic peritoneum, ovaries, uterosacral ligaments and other sites, intestinal involvement may occur in some patients (2).

Intestinal endometriosis typically involves the serosal and muscular layers of the intestinal wall. Its clinical manifestations may include cyclic abdominal pain, dyschezia, hematochezia during menstruation, and gastrointestinal symptoms correlated with the menstrual cycle. As the lesions usually infiltrate inward from the outer side of the intestinal wall, the mucosa may remain intact. Conventional colonoscopy and mucosal biopsy have limited detection capacity for deep lesions, and a negative result cannot rule out this disease (3). Meanwhile, some patients may present no typical dysmenorrhea or cyclic symptoms, leading to more insidious disease manifestations that are easily confused with gastrointestinal tumors, inflammatory diseases and other submucosal lesions (4).

Rectal endometriosis can be manifested in imaging and endoscopic ultrasonography (EUS) as localized thickening of the intestinal wall or a space-occupying lesion originating from the muscular layer. It shares certain similarities with gastrointestinal stromal tumors (GIST) (5). Therefore, in cases where there are no typical gynecological symptoms, accurate preoperative diagnosis remains challenging. Some patients may receive treatment targeting tumor-like lesions due to their imaging manifestations (6).

This article reports a case of rectal endometriosis presenting without cyclic symptoms. The patient's primary manifestations were chronic tenesmus and abnormal defecation. EUS of the rectum revealed a mass originating from the muscularis propria, raising strong suspicion of GIST. The diagnosis of rectal endometriosis was ultimately confirmed through surgical resection combined with histopathological and immunohistochemical examinations. This case highlights the importance of considering the possibility of intra-intestinal endometriosis in the differential diagnosis of lesions within the female rectal wall, even in the absence of typical gynecological or cyclical symptoms. Recognizing this possibility can assist in guiding preoperative assessment, but the best diagnostic method still requires further research.

## 2 Case presentation

A 45-year-old female was admitted with a chief complaint of anal tenesmus for 8 years, aggravated for 1 month. Eight years ago, she developed anal tenesmus without obvious predisposing factors, accompanied by increased bowel movement frequency and tenesmus, without abdominal pain, diarrhea, melena, weight loss, fatigue, nausea, or vomiting. During the past eight years, no relevant examinations or treatments were carried out at medical institutions. The patient had regular menstruation without dysmenorrhea. No definite exacerbation of anal tenesmus or bowel abnormalities during menstruation or relief after menstruation was observed, and the relationship between gastrointestinal symptoms and the menstrual cycle was not significant. Past history, personal history, and family history were unremarkable. Married, gravida 1 para 1, spontaneous vaginal delivery of 1 female infant, normal menstruation.

The physical examination findings of the patient are as follows: the abdomen is flat, no gastrointestinal pattern or peristaltic wave is observed, no abdominal wall varicose veins are seen, the abdomen is soft, with no tenderness, rebound tenderness or muscular tension, the liver and spleen are not palpable below the costal margin, no mass is palpated, tympanic sound is detected on percussion, and shifting dullness is negative. No edema is noted in both lower extremities. A mass is palpable via digital rectal examination, which is located at the 12 o'clock position in the lithotomy position, 10 cm from the anal verge, and no blood stain is observed on the withdrawn finger cot.

Colonoscopy revealed a 0.4 cm polyp in the ascending colon approximately 70 cm from the anal verge (Figure 1A), which was subsequently removed by forceps polypectomy. The pathological diagnosis was tubular adenoma. The folds of the transverse colon were regular with smooth mucosa, while the mucosa of the descending colon and sigmoid colon presented with hyperemia and edema. Segmental hyperemia and edema were observed in the rectal mucosa, and a protruding lesion measuring approximately 3.0 cm × 2.0 cm (longitudinal length × transverse width) with smooth surface mucosa was identified at the 12 o'clock position in the lithotomy position 10 cm from the anal verge (Figures 1B, C). Endorectal ultrasonography was then performed, and 20 MHz mini-probe exploration showed that the 1st to 3rd layers (mucosal layer and submucosal layer) at the rectal lesion were continuous and intact, while an oval mixed echo area with a cross-section of approximately 10.2 mm × 19.3 mm (thickness × width) was visualized in the 4th layer (muscularis propria). No definite signs of involvement were detected in the 5th layer (serosa/adventitia layer), and the lesion was highly suspected to be a gastrointestinal stromal tumor (Figure 1D).

Preoperative abdominal and pelvic CT examinations conducted in our hospital reveal that the uterus/cervix in the central pelvis is in close proximity to the anterior wall of the rectum (Figure 2A), with local space narrowing, yet no obvious compression or invasion of the rectum is observed. Preoperative CT revealed a high-density lesion in the distal rectum, anatomically corresponding to the lesion identified on colonoscopy, while no definite abnormalities were detected in the remaining segments of the rectum (Figure 2B).

Preoperative tumor marker test results: The serum CA-125 level was borderline elevated at 35.08 U/mL, slightly exceeding the upper reference limit of 35.00 U/mL (reference range: 0.00–35.00 U/mL), while all other tumor markers were within their respective reference ranges.

Combined with the patient's clinical symptoms and preoperative imaging manifestations, the preoperative diagnosis of this case was primarily focused on gastrointestinal stromal tumor. EUS revealed that the lesion was located in the muscularis propria of the rectum. Therefore, clinical evaluation was conducted mainly following the diagnostic pathway for suspected gastrointestinal neoplasms, while intestinal endometriosis was not included in the preoperative differential diagnosis, and no gynecological consultation was arranged. As this potential diagnosis was not considered at that time, the possibility of obtaining histological evidence via endoscopic ultrasound-guided fine needle aspiration (FNA) or fine needle biopsy (FNB) was not further assessed preoperatively. Retrospectively, there exist limitations in this diagnostic approach.

After admission, the patient completed relevant examinations, and no significant surgical contraindications were identified. Given

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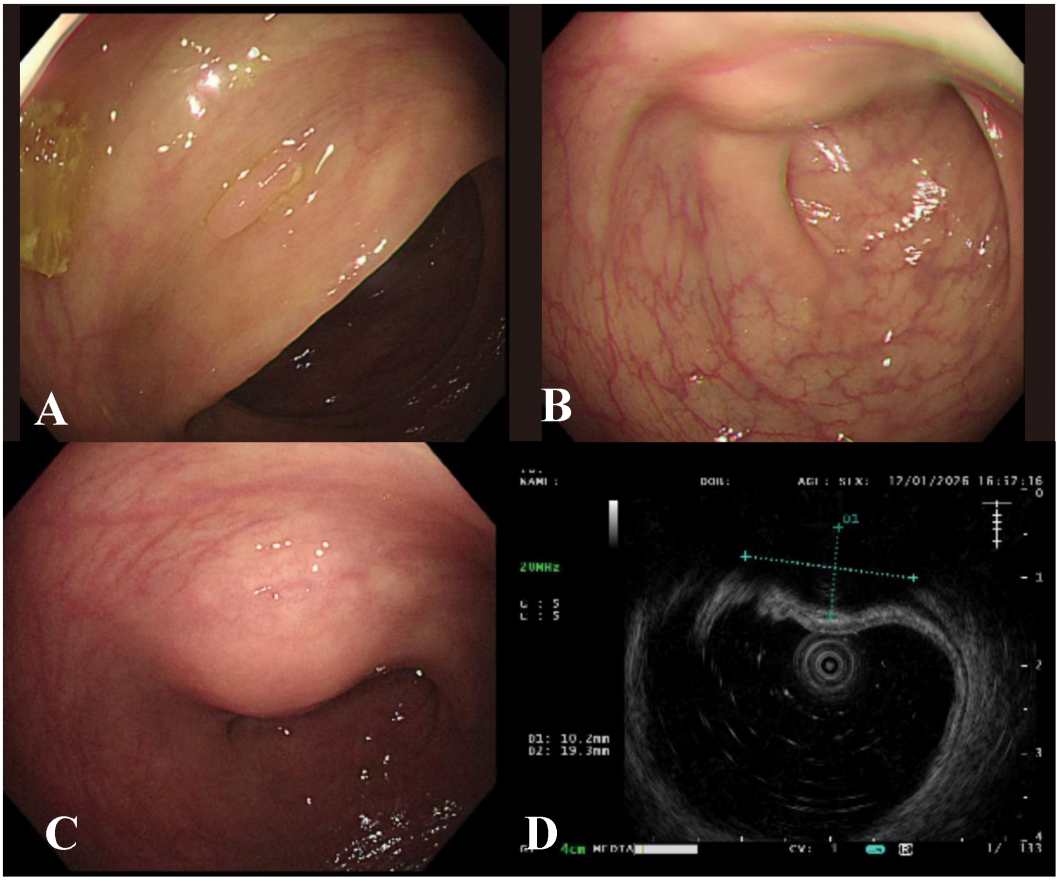



FIGURE 1
The colonoscopy and endoscopic ultrasound examinations of the patient. **(A)** Ascending colon polyp; **(B, C)** Endoscopic rectal protruding lesion with smooth surface mucosa; **(D)** Under EUS, an oval-shaped heterogeneous echo area can be observed in the fourth layer (the muscularis propria) of the rectal wall.

that the long-standing defecation symptoms have recently worsened, and colonoscopy and EUS strongly suggest GIST, a laparoscopic anterior rectal resection was performed to remove the lesion and obtain the final histopathological diagnosis. Laparoscopic anterior resection of the rectum was performed under general anesthesia, and the surgical procedure proceeded uneventfully. Intraoperatively, the uterine wall was found to be in close proximity to the serosal layer of the rectum, which was completely separated via blunt dissection. The diseased intestinal segment was resected, and no other endometriotic lesions or space-occupying lesions were observed in the uterus, ovaries and pelvic cavity. The postoperative specimen showed that the size of the tumor was 3 × 2 × 1 cm. The histopathological examination revealed the presence of ectopic endometrial glands and stroma within the rectal wall. No cellular

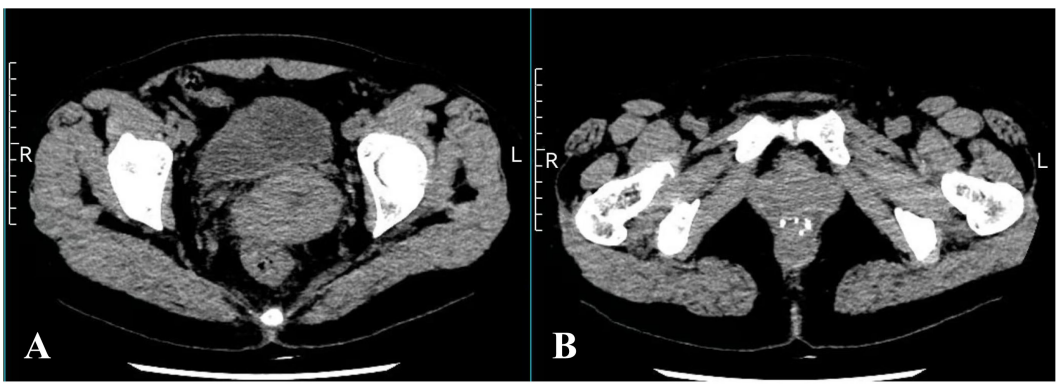



FIGURE 2
Pelvic CT examination. **(A)** The adjacent relationships of the bladder, the uterus, and the rectum; **(B)** High-density lesions can be observed in the distal part of the rectum.

atypia or malignant transformation was observed, and there was no infiltration of endometriosis lesions at the resection margins. Postoperative hematoxylin-eosin (HE) staining of the pathological specimen revealed ectopic endometrial tissue in the rectal wall, with focal hemorrhage and chronic inflammation of the intestinal mucosa. Under low-power microscopy, irregular endometrial glands were observed infiltrating the muscularis propria and submucosa of the rectum, without continuity with the rectal mucosal layer (Figure 3A). At medium magnification, the ectopic endometrial glands were lined by columnar epithelial cells and surrounded by endometrial-type stromal cells, which were also present between adjacent glands (Figure 3B). Immunohistochemical staining showed that ER and PR were expressed in the ectopic glandular epithelium and the surrounding stromal cells (Figures 4A, B), while CD10 was positive in the stromal components (Figure 4C). CK7 was positive in the ectopic glands, but negative in the primary intestinal mucosal epithelium, while CK20 exhibited the opposite staining pattern (Figures 4D, E). These findings provided supplementary evidence for differentiating ectopic endometrial glands from primary intestinal mucosa and supported the histological diagnosis combined with the hematoxylin-eosin staining results. This patient did not undergo PAX8 immunohistochemical examination. The Ki-67 labeling index in the ectopic glands was approximately 5% (Figure 4F). This result is only for descriptive reporting and is not an independent basis for excluding malignancy.

The patient experienced postoperative symptom relief, uneventful recovery, and was discharged smoothly. Given that the patient is 45 years old, approaching perimenopause, and presented primarily with intestinal symptoms this time without typical dysmenorrhea or chronic pelvic pain, joint gynecological follow-up was recommended after discharge, and no hormone-related treatment was administered. At the 1-month, 3-month, and 6-month postoperative follow-up visits, key inquiries were made regarding anal distension, defecation habits, pelvic pain, menstrual status, and the presence of cyclic intestinal symptoms, and no significant abnormalities were observed. The patient reported symptomatic improvement and enhanced quality of life.

## 3 Discussion

The clinical significance of this case lies in the fact that rectal endometriosis can present with long-term gastrointestinal symptoms and intestinal wall occupying lesions, while lacking typical gynecological symptoms and menstrual cycle-related manifestations. This patient sought medical attention due to long-term anal distension, increased frequency of defecation, and difficulty in defecation. Colonoscopy revealed a smooth, elevated lesion on the surface of the rectal mucosa, and EUS suggested that the lesion originated from the muscular layer. Preoperative highly suspected GIST was suspected, and ultimately, the diagnosis of rectal endometriosis was confirmed by postoperative pathology. Although EUS-guided FNA or FNB is technically feasible and may help clarify the nature of the lesion, due to the preoperative diagnosis mainly focusing on GIST and not including intestinal endometriosis in the differential diagnosis, no further tissue sampling was implemented. The limitations of this diagnostic approach may restrict the opportunity to clearly determine the nature of the lesion and further evaluate alternative treatment options before the operation. Eventually, a rectal anterior resection was performed in the context of persistent and recently aggravated defecation symptoms, the presence of rectal lesions, and the inability to rule out malignancy. However, the existing data are insufficient to determine that this surgical approach is the only appropriate choice. If the preoperative consideration included intestinal endometriosis and further examinations and gynecological or multidisciplinary evaluations were conducted, or alternative surgical options with a smaller scope could be explored, their applicability and whether they could change the final treatment decision remain undetermined. This case highlights that for rectal lesions of unknown nature in female patients, even without typical gynecological symptoms, intestinal endometriosis should be considered, and the necessity of preoperative tissue sampling and multidisciplinary diagnosis should be carefully evaluated based on clinical manifestations and imaging features to improve diagnostic accuracy and optimize individualized treatment decisions.

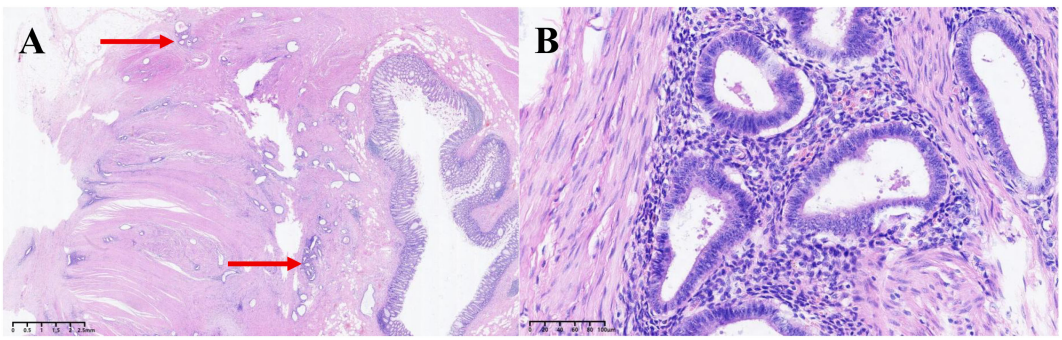



FIGURE 3
Postoperative microscopic pathological HE staining. **(A)** The irregular glands grow interdigitatingly within the rectal muscular layer and submucosa (indicated by the red arrow), and are not continuous with the rectal mucosal layer. 10×; **(B)** The glandular epithelium is columnar, arranged densely, with deeply stained chromatin in the cell nuclei. The endometrial glands are surrounded by endometrial stromal cells. 200×.

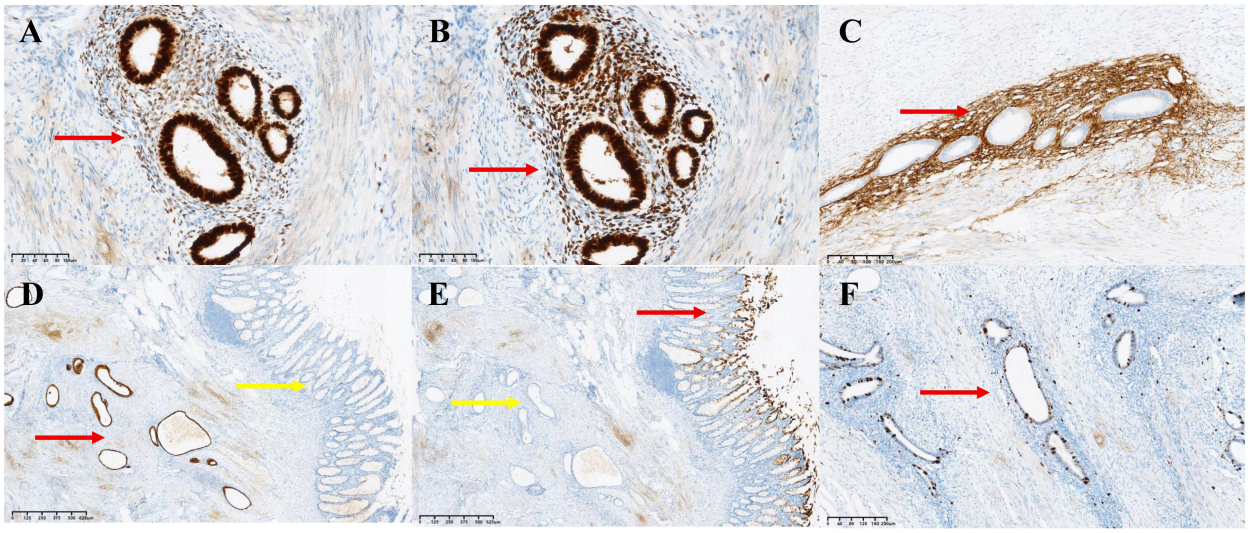



FIGURE 4
Immunohistochemical staining. **(A)** ER expression in the ectopic glandular epithelium and surrounding stromal cells, 200×; **(B)** PR expression in the ectopic glandular epithelium and surrounding stromal cells, 200×; **(C)** CD10 positivity in the stromal component, 100×; **(D)** CK7 positivity in the ectopic glands and negative staining in the native intestinal mucosal epithelium, 40×; **(E)** CK20 negativity in the ectopic glands and positive staining in the native intestinal mucosal epithelium, 40×; **(F)** Ki-67 labeling index of approximately 5% in the ectopic glands, 100×. The arrows indicate the corresponding positive or negative staining patterns.

Bowel endometriosis is a pathological condition defined by the involvement of the intestinal wall by endometrioid glands and stroma, which predominantly affects women of reproductive age and frequently coexists with other pelvic endometriotic lesions. Approximately 5% to 12% of patients with endometriosis present with intestinal involvement, among which lesions in the rectum and sigmoid colon account for around 90% of all bowel lesions, indicating a distinct predilection for the pelvic region and distal colon in its distribution (2). The pathogenesis of bowel endometriosis has not been fully elucidated, and it is currently recognized to involve the synergistic effects of ectopic cell origins, pelvic anatomical environment, local hormones, and immune inflammatory responses. The classic retrograde menstruation and implantation theory posits that viable endometrial cells, after flowing back through the fallopian tubes into the peritoneal cavity, can adhere to the intestinal surface and form ectopic lesions. The rectouterine pouch facilitates the deposition of retrograde tissues, the anatomical location of the sigmoid colon may restrict their diffusion to the upper abdomen, and the adjacent relationship between the rectosigmoid colon and the ectopic lesions in the posterior pelvic cavity may also promote local involvement, thus accounting for its predilection sites (7). Under the combined action of estrogen-dependent growth, local immune abnormalities and chronic inflammation, the ectopic tissues can survive continuously, and promote angiogenesis, smooth muscle hyperplasia and fibrosis through repeated tissue injury and repair (8, 9). The lesions usually infiltrate from the outer side of the intestinal wall to the muscularis propria, while mucosal involvement is relatively rare. As a result, intramural masses with intact mucosa can be formed, and persistent gastrointestinal symptoms can be caused by intestinal wall thickening, fibrosis and adhesions, which provides a pathological basis for its atypical clinical manifestations and the difficulty in differentiating it from intestinal neoplasms (2, 7).

The diagnostic value of colonoscopy for intestinal endometriosis mainly lies in observing the changes in the intestinal lumen and mucosa, obtaining tissues and differentiating it from other colorectal diseases. However, its detection ability is limited by the infiltration depth of the lesion (10). Since the lesions usually infiltrate from the outer side of the intestinal wall to the inside, they mainly affect the serosa and muscular layer, and the mucosa can remain intact. Therefore, a normal colonoscopy or negative routine mucosal biopsy cannot rule out this disease. Abnormal endoscopic manifestations may include eccentric intestinal wall thickening, submucosal protrusions, polypoid lesions and surface nodules, but these manifestations lack specificity. A prospective study conducted by Milone et al. in 174 patients with deep pelvic endometriosis showed that the sensitivity of colonoscopy for identifying intestinal involvement was only 7%, and the specificity was 98%; among 76 patients with intestinal involvement confirmed by surgery and pathology, only 2 had mucosal invasion, and mucosal invasion was relatively rare, which was an important reason for the missed diagnosis by colonoscopy (3). However, when there are surface abnormalities, targeted biopsy may still provide a diagnostic basis. Kim et al.'s retrospective study of 17 patients with colorectal endometriosis found that the histological diagnosis rate of biopsy tissue in patients with surface nodules was 67%, while in those without surface nodules it was 0%, suggesting that the value of biopsy is closely related to the surface characteristics of the lesion (11).

EUS can observe the surface of the intestinal lumen while showing the layering of the intestinal wall and adjacent tissues, thus having supplementary diagnostic value for patients with intact mucosa, negative routine biopsy, and still having intramural or extramural lesions. Rectal endometriosis on EUS can present as low-echo or heterogeneous lesions connected to the muscular layer of the rectal wall, accompanied by local intestinal wall thickening, irregular contour or disturbed normal layering (12, 13). By judging

the infiltration layer, the size of the lesion and its relationship with adjacent pelvic tissues, EUS can help identify intestinal wall infiltration and provide a basis for preoperative assessment and combined diagnosis by gynecology and colorectal surgery. Its value is particularly evident in cases where conventional colonoscopy can only detect protrusions or external pressure but is difficult to clearly determine the nature and depth of the lesion layer (13). It is worth noting that the hypoechoic mass seen when intestinal endometriosis involves the muscular layer is similar to the imaging manifestations of gastrointestinal stromal tumors, so it is difficult to determine its nature based solely on the layer where the lesion is located and its echo characteristics (5, 14). For cases with atypical manifestations, unclear lesion nature and uncertain pathological results that may affect treatment decisions, EUS-guided sampling can be considered after evaluating feasibility and risks to obtain histological evidence. However, when sampling is insufficient, negative results still need to be combined with clinical and other imaging data for comprehensive judgment (15).

Differential diagnosis between intestinal endometriosis and GIST represents a critical clinical issue in the diagnostic workup of intestinal submucosal masses. When endometriotic lesions involve the muscularis propria and are accompanied by fibromuscular hyperplasia, they may present as a submucosal protrusion with intact mucosa under colonoscopy, and appear as a hypoechoic mass contiguous with the muscularis propria on EUS. These imaging features overlap with those of GIST, rendering the originating layer and echogenic characteristics of the lesion insufficient for definitive standalone diagnosis of its nature (5, 14). A case reported by Wadhwa et al. was initially suspected to be GIST or leiomyoma based on EUS manifestations, but was ultimately confirmed as endometriosis via surgical pathology, indicating that this condition can mimic mesenchymal neoplasms (14). Particularly in female patients without dysmenorrhea, cyclic gastrointestinal symptoms, or a prior history of endometriosis, the lack of typical clinical clues may increase the difficulty of differential diagnosis. Accordingly, intestinal endometriosis should be included in the differential diagnosis list for submucosal lesions of the rectum and sigmoid colon (4). Histopathology serves as the cornerstone for differentiating intestinal endometriosis from GIST. Endometriosis is primarily diagnosed based on the morphological features of ectopic endometrial-like glands and stroma, with immunohistochemistry providing supplementary diagnostic evidence: CD10 can help identify endometrial-type stroma, while ER and PR expression can support the identification of ectopic endometrial tissue. PAX8 may be useful for highlighting ectopic glandular epithelium; however, PAX8 immunostaining was not performed in the present case. In contrast, GIST is typically characterized by proliferating spindle or epithelioid tumor cells, and its diagnosis requires correlation with immunohistochemical markers such as CD117 and DOG1. Molecular testing can be further performed for diagnostically challenging cases (16–18).

This lesion is located in the anterior wall of the rectum. The CT scan indicates that the uterus or cervix is closely related to the anterior wall of the rectum, and during the operation, it was also observed that the two were closely related locally. These findings alone cannot confirm endometriosis of the uterus, but they can serve as important anatomical clues for re-examining the tumor diagnosis. For female patients with such clues, further gynecological assessment and targeted pelvic ultrasound or MRI can help supplement information on the extent of the lesion and the relationship with adjacent organs. The ESHRE guidelines recommend using ultrasound or MRI in the diagnostic evaluation of suspected endometriosis, and emphasize that negative imaging cannot completely rule out the disease (19). Due to the lack of clinical experience with gastrointestinal endometriosis, no pelvic MRI or gynecological ultrasound examination was performed on the patient. This is something that needs to be noted in future clinical diagnosis and treatment, but it cannot be asserted that these examinations will definitely avoid misdiagnosis or change the surgical plan.

In order to identify the previously published cases related to the current diagnostic challenges, we conducted a targeted literature search in PubMed up to August 2026. The search terms used included “case reports”, “endometriosis”, “intestinal endometriosis”, “rectal endometriosis”, “colon endometriosis”, “gastrointestinal stromal tumor” and “GIST”. The inclusion criteria were as follows: (i) patients with a confirmed diagnosis of intestinal endometriosis; (ii) cases in which the preoperative diagnosis or differential diagnosis included gastrointestinal neoplasms, such as GIST or colorectal cancer; and (iii) published reports providing sufficient information on clinical manifestations, endoscopic or imaging findings, preoperative diagnosis, or diagnostic procedures to allow comparison with the index case. Finally, 12 cases that are highly relevant to the aforementioned standards and the patient in this case were determined. Table 1 is designed to provide a selective narrative comparison of clinically pertinent cases, rather than constituting a comprehensive or systematic review of all published cases of intestinal endometriosis.

Among the 12 selected cases, patients’ ages ranged from 27 to 49 years, with a median age of 40 years. The lesions were located in the rectum in 4 cases, the sigmoid colon in 7 cases, and the rectosigmoid colon in 1 case. A total of 6 cases had clear records of menstruation-related symptoms. Among them, 5 cases had no clear correlation, and 1 case had cyclic pelvic pain. The 7 cases were considered to have GIST before surgery, 1 case was considered to have GIST or endometriosis, 2 cases were considered to have rectal cancer, and 2 cases were considered to have sigmoid colon cancer before surgery. The reported endoscopic findings were heterogeneous, including mucosal protrusions, submucosal or extramural masses, polypoid or ulcerofungating lesions, and luminal narrowing. Normal mucosa was also reported in one case, further illustrating the variability of endoscopic presentations of intestinal endometriosis. Endoscopic ultrasonography (EUS) was reported in 3 cases, with lesions involving the muscularis propria in 2 cases and the muscularis mucosa in 1 case. These cases and this case jointly suggest that the clinical and endoscopic manifestations of intestinal endometriosis are diverse, and it is not uncommon to lack clear menstrual-related symptoms. Some lesions, even after assessment by endoscopic ultrasound, may still be judged as GIST or other tumors. This indicates that in the diagnosis and treatment of female rectal and sigmoid colon masses, symptoms, the relationship between the lesion and pelvic organs, and histological evidence

TABLE 1 Summary of previous related cases.

| Case | Author | Time | Age | Cardinal symptom | Related to menstruation | Childbearing history | Colonoscopy findings | Biopsy | CT | EUS | Bowel wall layer involved | Involving the intestines | Preoperative diagnosis |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | Kim (20) | 2009 | 41 | Hematochezia | No | G3P2 | Ulcero-fungating mass | Chronic inflammation | Colonic wall thickening | No | Not assessed | Sigmoid | SC |
| 2 | Kim (20) | 2009 | 27 | Hematochezia | No | G1P0 | Ulcero-fungating mass | Normal | Rectal wall thickening | No | Not assessed | Rectum | RC |
| 3 | Kim (20) | 2009 | 46 | None | No | G2P1 | Mucosal protrusion | Not performed | Submucosal tumor | No | Not assessed | Rectum | GIST |
| 4 | Kim (20) | 2009 | 47 | Abdominal pain and obstipation | No | G2P2 | Luminal narrowing | Not performed | No evidence of disease | No | Not assessed | Sigmoid | SC |
| 5 | Kim (20) | 2009 | 38 | None | No | G0P0 | Polypoid mass | Chronic inflammation | No evidence of disease | No | Not assessed | Rectum | RC |
| 6 | Lee (21) | 2012 | 48 | Pelvic pain | Cyclic pelvic pain | Not reported | Mucosal protrusion | Not performed | No evidence of disease | Yes | Muscularis propria | Sigmoid | GIST |
| 7 | Wadhwa (14) | 2015 | 35 | Diarrhea and abdominal pain | Not reported | Not reported | Submucosal mass | Nondiagnostic (EUS-FNA) | No report | Yes | Muscularis propria | Sigmoid | GIST |
| 8 | Kafadar (6) | 2020 | 37 | Pelvic pain and abdominal bloating | Not reported | G0P0 | Submucosal mass | Not performed | A mass with an unclear boundary from the intestinal wall | No | Not assessed | Rectosigmoid colon | GIST or EM |
| 9 | Kondo (22) | 2020 | 49 | Positive FOBT | Not reported | Not reported | Extramural mass | Not performed | Sigmoid colon wall thickening | No | Not assessed | Sigmoid | GIST |
| 10 | Kondo (22) | 2020 | 48 | Positive FOBT and constipation | Not reported | Not reported | Normal mucosa | Not performed | Rectal anterior wall mass | No | Not assessed | Rectum | GIST |
| 11 | Kondo (22) | 2020 | 37 | Positive FOBT | Not reported | Not reported | Submucosal mass | Not performed | Sigmoid colon mass | No | Not assessed | Sigmoid | GIST |
| 12 | Ebrahimi (23) | 2022 | 39 | Diarrhea with blood, abdominal pain during defecation | Not reported | G0P0 | Aphthous lesion with a large mass | Active focal colitis | No evidence of disease | Yes | Muscularis mucosa | Sigmoid | GIST |

CT, Computed tomography; EUS, Endoscopic ultrasonography; EUS-FNA, Endoscopic ultrasound-guided fine-needle aspiration; EM, Endometriosis; GIST, Gastrointestinal stromal tumor; RC, Rectal cancer; SC, Sigmoid colon cancer; FOBT, Fecal occult blood test; G, gravidity (total number of pregnancies); P, parity (number of pregnancies resulting in delivery at or beyond the gestational age threshold for viability).





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should be comprehensively considered for differentiation, and the uncertainty of preoperative diagnosis should be fully taken into account in the treatment decision-making.

This report has the following limitations: Firstly, the single-case design cannot determine the frequency of this manifestation or compare the effectiveness of different treatment strategies. Secondly, the selective narrative literature review may have selection and publication biases, and the 12 cases cannot be used to estimate the prevalence or misdiagnosis rate. Thirdly, although abdominal and pelvic CT examinations were performed before the operation, targeted pelvic MRI, gynecological ultrasound, or gynecological consultation were not conducted. Therefore, it is impossible to determine whether further evaluation can change the diagnosis or treatment plan. Finally, the follow-up period was only 6 months and was mainly based on symptom assessment. The long-term recurrence and subsequent treatment needs are still unclear. Therefore, this case only provides clinical reference and cannot prove that a specific diagnostic pathway or a smaller-scale surgery can improve prognosis.

In conclusion, for female patients with rectal space-occupying lesions who present no typical gynecological symptoms or definite menstruation-related manifestations, endometriosis should still be included in the differential diagnosis, especially when the lesion is located in the anterior rectal wall and has a close anatomical relationship with the uterus or cervix. The intact mucosa observed under enteroscopy and the intramuscular lesion shown by endoscopic ultrasonography are insufficient to determine the nature of the lesion independently. A comprehensive diagnosis shall be made by combining the medical history, pelvic anatomical relationship, targeted imaging examinations and necessary histological evaluations. The CARE checklist is described in Supplementary File 1.

## Data availability statement

The raw data supporting the conclusions of this article will be made available by the authors, without undue reservation.

## Ethics statement

Written informed consent was obtained from the individual(s) for the publication of any potentially identifiable images or data included in this article. Written informed consent was obtained from the participant/patient(s) for the publication of this case report.

## Author contributions

ZH: Writing – review & editing, Data curation. XM: Writing – original draft, Writing – review & editing, Data curation. JC: Writing – review & editing. YZ: Writing – review & editing, Conceptualization, Writing – original draft. GZ: Writing – review & editing. ZT: Writing – review & editing, Writing – original draft, Funding acquisition.

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## Conflict of interest

The author(s) declared that this work was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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## Supplementary material

The Supplementary Material for this article can be found online at: https://www.frontiersin.org/articles/10.3389/fonc.2026.2023871/full#supplementary-material

SUPPLEMENTARY FILE 1
The CARE checklist.

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