Submucosal Lesions Presenting With Rectal Bleeding-Endometriosis in the GI Tract

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Summary

Endometriosis is typically marked by the implantation and proliferation of foci of endometrial stroma and glands outside the endometrial cavity and uterine musculature. Extra-gonadal seeding has been observed in sites like the small and large bowel, peritoneum, appendix, pleura, umbilicus, old wound scars, subarachnoid space, inguinal canal, heart and lungs.^{14,7} The gastrointestinal tract is the third most common localization of endometriosis after the ovaries and the peritoneum, with intestinal involvement being reported in about 12 to 37% of individuals.^{1,2,5-7} Establishing a diagnosis of intestinal endometriosis can often be a diagnostic challenge. We present an interesting case of sigmoidal endometriosis in a 46-year-old female, whose initial presentation was irregular bowel movements with rectal bleeding.

Résumé

L'endométriose se caractérise habituellement par l'implantation et la prolifération de foyers du stroma et des glandes de l'endomètre à l'extérieur de la cavité endométriale et de la musculature utérine. Un ensemencement extra-gonadique a déjà été observé dans des sites comme le petit et le gros intestin, le péritoine, l'appendice, la plèvre, l'ombilic, de vieilles cicatrices de blessures, l'espace sous-arachnoïdien, le canal inguinal, le cœur et les poumons^{14,7}. Le tube digestif vient au troisième rang en termes de localisation de l'endométriose, après les ovaires et le péritoine, avec des atteintes intestinales relevées dans 12 à 37 % des cas^{1,2,5-7}. Il peut souvent être difficile d'établir un diagnostic d'endométriose intestinale. Nous présentons ici un cas intéressant d'endométriose sigmoïdienne chez une femme de 46 ans et dont les premières manifestations consistaient en selles irrégulières accompagnées de saignements au niveau du rectum.

Case

A 46-year-old female presented with a 4-week history of irregular bowel movements (increased frequency and looser than baseline), and rectal bleeding (both outside and mixed within stool). There was no associated abdominal pain, nausea/vomiting, hematemesis, coffee ground emesis, or melena. There were no fevers, chills, or weight loss (some weight gain). No history of travel or sick contacts. Review of symptoms was unremarkable. Family history was negative for colon cancer or any GI conditions including celiac disease or inflammatory bowel disease (IBD). She had never received a colonoscopy in the past.

Her past medical history was unremarkable with no prior surgeries. She was on no medications including no over the counter medications. She had been a non-smoker and non-drinker all her life and had no drug allergies.

Examination was unremarkable with no palpable masses or areas of tenderness in the abdomen. DRE was unremarkable.

Laboratory investigations including CBC, electrolytes, creatinine, urea, metabolic panel, and inflammatory markers were all within normal limits.

Given the rectal bleeding and history of irregular bowel movements, with no biochemical abnormalities or physical exam findings, an elective colonoscopy was performed. Two

Fig 1. First submucosal lesion seen at ~30 cm from the rectum.

Fig 2. Second submucosal lesion seen at 35-40 cm from the rectum.

large submucosal lesions were found (30 cm and 40 cm from the rectum respectively) that were firmer than a typical lipoma and appearances were most in keeping with GIST tumors (Figure 1 and 2). Deep biopsies were taken. The transverse colon appeared mildly edematous, with appearances suggestive of melanosis coli. Some internal hemorrhoids (not bleeding) were noted upon retroflexion.

Pathologies from the attempted deep biopsies revealed normal colonic mucosa, muscularis mucosa and superficial submucosa, with no significant abnormalities and no neoplastic spindle cell lesions present.

A CT scan of the abdomen/pelvis showed unremarkable bowel loops, and the submucosal masses visualized during colonoscopy could not be appreciated on imaging. The spleen, pancreas, adrenals, kidneys, ovaries, bone, and soft tissue were all unremarkable.

Given the persistent GI symptoms with focally identified lesions on colonoscopy that looked characteristic of GIST tumors, and no evidence of metastatic disease on imaging, our patient underwent a repeat colonoscopy where the submucosal lesions were tattooed; and subsequently a laparotomy for resection of the lesions, with left hemi-colectomy and end anastomosis. Intraoperatively, there was no evidence of metastatic disease in the abdomen; however she did have a large uterine fibroid, enlarged appendix (resected), and evidence of endometriosis on both ovaries.

Pathologies from the resected recto-sigmoid specimen interestingly revealed extensive endometriosis characterized by variably sized columnar-lined glands cuffed by endometrial-type stroma, extending from the sub-mucosa to the mesentery. The diagnosis was supported by immunohistochemistry (positive ER immunostain with CD10 reactivity). The appendix showed evidence of endometriosis as well, and the anastomotic margins were histologically normal.

The patient did very well post-operatively. Her bowel movements became regular and formed, with no evidence of lower GI bleeds.

Discussion

Endometriosis occurs in 6 to 10% of the general female population.¹² Its presence in the gastrointestinal tract has been reported in several case reports with the sigmoid colon and rectum being the two most common sites, followed by the ileum, ileocecal area, appendix and anterior rectal wall.³

Establishing a diagnosis of intestinal endometriosis is often a diagnostic challenge as none of its symptoms are pathognomonic, and even though the symptoms should physiologically be worse cyclically with menses, this is

not always the case.⁸ The disease is primarily diagnosed in premenopausal women, but can also cause symptoms in postmenopausal women as endometrial implants may continue to induce a fibrotic reaction in the muscle of the affected bowel wall even after cessation of ovarian function.¹³ Presenting symptoms are often non-specific and can include unexplained digestive problems, tenesmus, per-rectal bleeding, bloating, constipation, diarrhea, and pain. Pain can be epigastric, abdominal, pelvic or rectal with the symptoms varying depending on the location, depth of invasion, adnexal adhesions,⁹ and the presence of a consequential partial or complete bowel obstruction.^{3,15}

Endoscopic diagnosis of intestinal endometriosis is challenging, as most lesions do not infiltrate the gastrointestinal mucosa; and therefore radiological modalities such as CT scans, MRI or trans-vaginal ultrasonography may help with making the diagnosis. Based on case reports and observational studies, luminal findings suggestive of endometriosis range from eccentric wall thickenings, polypoid lesions, endometrioid heterotopias of the mucosa, and gross surface nodularities/ulcerations.^{16,17} Rates of histological confirmation of the diagnosis seem to be highest in cases with surface nodularities, with inconclusive results seen in most other cases.¹⁷

Management involves symptom control and maintaining fertility. This is achieved primarily from surgical resection of all visible endometriosis. Hormonal therapies may improve symptoms in the short term but they do not improve fertility.^{10,11} Patients with intestinal endometriosis achieve disease control with either laparoscopic bowel resection, or segmental bowel resection (via laparotomy and primary end anastomosis) depending on the extent of disease.⁴

In our patient, recto-sigmoidal endometriosis presented endoscopically with what looked like classic GIST lesions. Radiological imaging and biopsies did not help us make the diagnosis. Even though the ultimate management of our patient in either case would have been identical, the take-home learning point from the case was that the differential diagnoses for sub-mucosal GI lesions in the recto-sigmoidal region, amongst other known benign and malignant etiologies, must also include Endometriosis.

In summary, the diagnosis of intestinal endometriosis can be challenging. However, it is prudent to consider this in the differential diagnosis, especially in a premenopausal patient presenting with vague abdominal symptoms including unexplained digestive problems, pain, changes in bowel movements or bleeding from the GI tract. Cyclical bowel symptoms with menstruation along with concurrent findings on history including menometrorrhagia, infertility and

dyspareunia should point one in the direction of intestinal seeding; however, the absence of these should not deter one from considering the diagnosis.

Including Endometriosis in the differential diagnoses of undiagnosed submucosal lesions in the GI tract could thereby help guide patient management and allow for timely referrals as needed.

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