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Title

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Permalink

<https://escholarship.org/uc/item/3r57837b>

Journal

ACG Case Reports Journal, 8(6)

ISSN

2326-3253

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Publication Date

2021

DOI

10.14309/crj.0000000000000557

Peer reviewed

A Masked Diagnosis: Sigmoid Endometriosis

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ABSTRACT

Serosal involvement in intestinal endometriosis is relatively common, and patients often present with nonspecific gastrointestinal symptoms; however, presentation with deeper mucosal infiltration and rectal bleeding is rare. We report a case of a 40-year-old woman with a history of breast cancer in remission who presented with periodic rectal bleeding and abdominal pain. Computed tomography scan showed sigmoid lesions concerning for metastatic disease. Colonoscopy showed hypervascular sigmoid lesions which were confirmed to be endometriosis on histopathology. This case highlights endometriosis as a rare differential to be considered in young women with abnormal bowel imaging or catamenial rectal bleeding.

INTRODUCTION

Endometriosis frequently involves the bowels with rectosigmoid involvement being the most common.¹ Although serosal infiltration is not uncommon and typically leads to alterations in bowel habits, mucosal involvement is rare and, hence, the presentation with rectal bleeding is rare. Diagnosis without pathology is difficult, given symptoms frequently mimic irritable bowel syndrome, inflammatory bowel disease, or malignancy. On imaging and endoscopy, these lesions have been reported to masquerade as malignancy, further supporting the importance of pathology for diagnosis.

CASE REPORT

A 40-year-old G1P0 woman with a history of ER+/PR-/HER2+ breast cancer presented with worsening rectal bleeding for 1 year.² She was diagnosed with breast cancer 5 years earlier and treated with neoadjuvant chemotherapy, lumpectomy, radiotherapy, and 1 year of trastuzumab. Since being on maintenance tamoxifen, she has been in remission with no evidence of recurrence. She reported red blood in her stools which mostly coincided with menses, but was also associated with dyschezia, abdominal pain, occasional loose stools, and dysmenorrhea. The onset of menarche was at age 15 with regular length menstrual cycles. The digital rectal examination was normal, and no mass was palpated. Basic laboratory test results included normal hemoglobin. Because of the history of breast cancer, a computed tomography scan was obtained which demonstrated a focal area of sigmoid wall thickening and a 3.2 × 4-cm luminal filling defect with narrowing (Figure 1). Lack of other remarkable findings on computed tomography and pelvic ultrasound lowered suspicion for gynecologic malignancy. Primary colonic malignancy and metastasis from her previous breast cancer were considered in the differential. Colonoscopy revealed an area of extreme tortuosity in the sigmoid colon that could not be traversed. Several 5- to 15-mm, hypervascular, friable, oval lesions embedded in the colonic wall were noted in the proximal sigmoid colon at the site of narrowing (Figure 2).

Cold forceps biopsy of the lesions showed benign colonic mucosa with well-circumscribed foci of small glands dispersed in a variably cellular stroma. The glands consisted of bland pseudostratified columnar epithelial cells and were associated with a stromal condensation of small bland fusiform cells, capillaries, and microscopic hemorrhage. There were no decidual changes of the glands to suggest progestin therapy or pregnancy, and there was no cytologic or architectural atypia to suggest a neoplastic process (Figure 3). An immunohistochemical stain for CD10 highlighted the stromal cells as endometrial-type stroma, which supported endometriosis. The stain for estrogen receptor (ER) highlighted the nuclei of the glands and stroma, further supporting endometriosis. The stains for CD10 and ER delineated a clear demarcation between the endometriosis and the colonic lamina propria (Figure 4).



Figure 1. Computed tomography showing wall thickening without surrounding inflammatory changes and a 3.2 × 4.0-cm luminal filling defect in the sigmoid colon.

After a discussion of multiple therapeutic modalities, the patient was treated with goserelin acetate with complete resolution of her rectal bleeding. After a year, goserelin therapy was discontinued because of side effects of fatigue and joint pain, and she has since been on danazol suppositories. Surgery consultation was obtained, but the surgical intervention was deferred because she did not have colonic obstructive symptoms and had an excellent response to medical therapies.

DISCUSSION

Endometriosis is defined as the presence of endometrial glands and stroma in extrauterine sites. Bowel endometriosis deeply infiltrates more than 5 mm below the peritoneum.³ Most bowel endometriotic lesions do not penetrate the bowel wall. In 1 review of patients who underwent surgical resection for bowel lesions, 95% of lesions invaded the serosa and muscularis propria, 38% entered the submucosa, and only 6% invaded the mucosa.⁴ Of women with endometriosis, the prevalence of

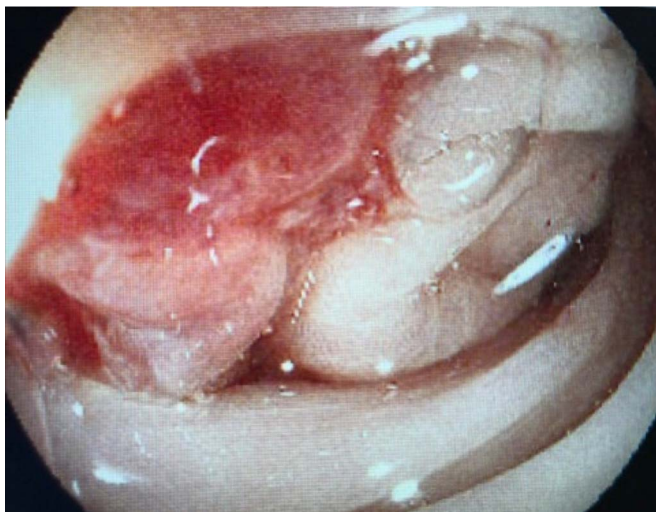


Figure 2. Colonoscopic view of a hypervascular, oval lesion found embedded at the sigmoid colon.

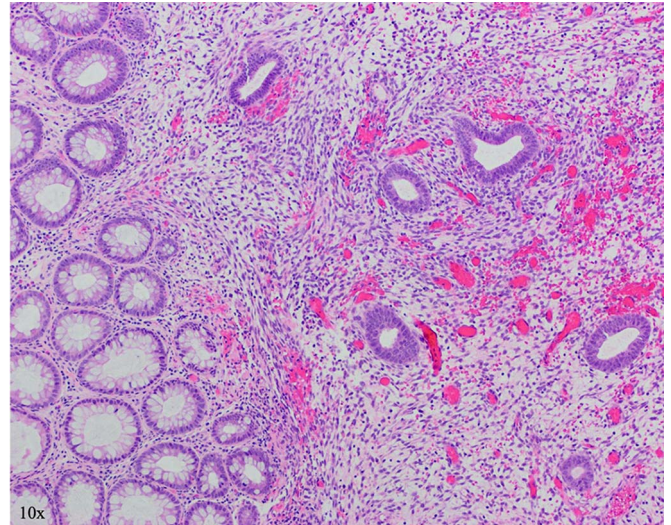


Figure 3. Hematoxylin and eosin images showing well-circumscribed foci of small glands dispersed in a variably cellular stroma within benign colonic mucosa. The glands consist of bland pseudostratified columnar epithelial cells and are associated with stromal condensation of small bland fusiform cells, capillaries, and microscopic hemorrhage. There is no decidual change of the glands to suggest progestin therapy or pregnancy.

bowel endometriosis has a wide range of 5%–25%, which has been hypothesized to be due to variable time of diagnosis and referral patterns.^{5,6}

Depending on the site of endometriosis, symptoms vary from nonspecific pain, diarrhea, and bloating to the classic symptoms of dyschezia, rectal bleeding, and constipation.⁷ Notably, the severity of symptoms does not predict the size of lesions or the full extent of the disease.⁸ The nonspecific gastrointestinal symptoms seen with intestinal endometriosis coupled with the rarity of the disease presents a diagnostic challenge to gastroenterologists, particularly if a thorough menstrual history is not taken. Other mimickers of this disease include malignancy, inflammatory bowel disease, irritable bowel syndrome, diverticulitis, and hemorrhoids.

Untreated deep endometriotic nodules have the potential to progress which becomes particularly important for young patients with rectosigmoid endometriosis who wish to conceive. Currently, universally accepted surveillance criteria are lacking, but some type of surveillance should be considered to assess the growth of the nodules to prevent significant consequences, especially if amenorrhea has not been achieved.⁹ Although symptoms of colonic endometriosis are typically chronic; late diagnoses can lead to unfortunate sequelae, including severe bleeding or misdiagnosis with colonic malignancy and acute bowel obstruction. Nonsteroidal anti-inflammatory drugs, hormonal contraceptives, gonadotropin-releasing hormone agonists, and aromatase inhibitors are medical options used in the treatment of endometriosis. Early diagnosis is clinically important to attempt medical therapies before severe bleeding

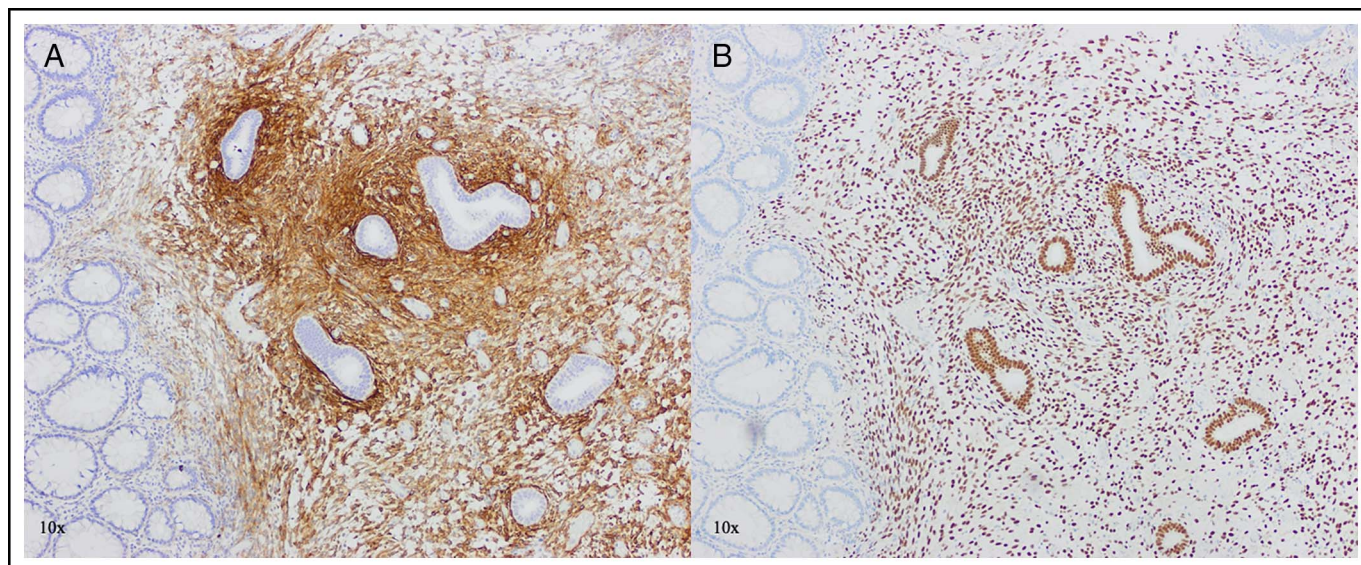


Figure 4. (A) Immunohistochemical stain (brown chromogen) for CD10 expression highlights the cytoplasm of stromal cells as endometrial-type stroma. (B) The stain for estrogen receptor highlights the nuclei of the glands and stroma, further supporting endometriosis. The stains for CD10 and estrogen receptor delineate a clear demarcation between the endometriosis and the colonic lamina propria.

or obstructive complications necessitate surgical intervention. Studies have shown more than 20% of patients undergoing surgery for lesion excision required additional surgeries, further highlighting the importance of early diagnosis.¹⁰

In conclusion, this case highlights that gastroenterologists should be aware of mucosal endometriosis in the differential of rectal bleeding in women in their reproductive years and be familiar with its unusual endoscopic appearance which can mimic malignancy. Despite the friable nature of the lesion, adequate sampling should be performed for confirmation on histopathology. We also emphasize the importance of good history taking, where the cyclical nature of rectal bleeding coinciding with menses is an important clue for prompt diagnosis.

DISCLOSURES

Author contributions: L. Subbaraj and A. Singh wrote the manuscript, revised the manuscript for intellectual content, reviewed the literature, and approved the final manuscript. CJ Bowman edited the manuscript, revised the manuscript for intellectual content, reviewed the literature, and approved the final manuscript. L. Subbaraj is the article guarantor.

Financial disclosure: None to report.

Previous presentation: This case was presented at the American College of Gastroenterology Annual Scientific Meeting; October 25–30, 2019; San Antonio, Texas.

Informed consent could not be obtained from the patient despite several attempts. All identifying information has

been removed from this case report to protect patient privacy.

Received June 18, 2020; Accepted October 4, 2020

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