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Leiomyosarcoma of the colon: A challenging diagnosis in young women - A case report

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Abstract:

Leiomyosarcoma (LMS) of the colon is an extremely rare colorectal malignancy, accounting for less than 0.1% of all colorectal cancers. The clinical symptoms of colonic LMS are often vague and nonspecific, including abdominal pain, changes in bowel habits, rectal bleeding, or an abdominal mass, which makes early diagnosis difficult. The majority of cases occur in middle-aged adults, with a slight male predominance. We report a rare case of colonic LMS in a young woman, whose clinical symptoms closely resembled those of a ruptured bleeding ovarian cyst or ectopic pregnancy, leading to an initial misdiagnosis. The definitive diagnosis was made through laparoscopic surgery and histopathological biopsy. Histopathological examination using histopathological examination (H&E) staining and immunohistochemistry (IHC) confirmed colonic LMS. Metastatic involvement was identified in 7 out of 11 regional lymph nodes. Our report highlights a very rare case of LMS involving the rectum and sigmoid colon, underscoring the diagnostic challenge in young female patients and emphasising the importance of surgical exploration and pathological confirmation in atypical pelvic presentations.

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1. Introduction

Leiomyosarcoma of the colon is a rare and aggressive malignant tumour arising from the smooth muscle cells of the muscularis propria. Unlike adenocarcinomas, which are the most common colorectal malignancies, LMS belongs to the group of soft tissue sarcomas and accounts for only a small percentage of colorectal cancers [1]. The clinical characteristics of smooth muscle tumours depend on their size, location, and associated bleeding complications. Symptoms are often non-specific and may include abdominal pain, changes in bowel habits, rectal bleeding, weight loss, or the presence of an abdominal mass. In some cases, the tumour may cause obstruction or perforation, leading to acute complications [2-5]. The majority of cases occur in adults between the ages of 36 and 89 years [3]. Imaging studies such as CT scan and MRI help identify the tumour's location, size, and extent of tumour invasion. Colonic leiomyomas originate from the muscle layer of the colon and are characterised by spindle cells with eosinophilic cytoplasm. The result of IHC staining of leiomyoma is typically positive for desmin and smooth muscle actin. Biopsy and IHC staining (e.g., positive for smooth muscle markers such as desmin and actin) confirm the diagnosis [6, 7]. Surgical resection with negative margins remains the primary treatment approach. Unlike adenocarcinomas, LMS is less responsive to chemotherapy and radiotherapy, though these may be considered in advanced cases. Prognosis is generally poor due to the aggressive nature of the tumour and its tendency to recur and metastasise, particularly to the liver and lungs [8].

2. Case presentation

A 21-year-old woman with no significant medical history presented with severe right pelvic pain and mild blood loss. No significant findings were noted in her family history. On abdominal examination, there was marked tenderness over the right iliac fossa and a positive guarding response. Ultrasonography revealed a large amount of blood in the abdomen and a mixed-echo mass (~3 cm) near the right ovary (Fig. 1).

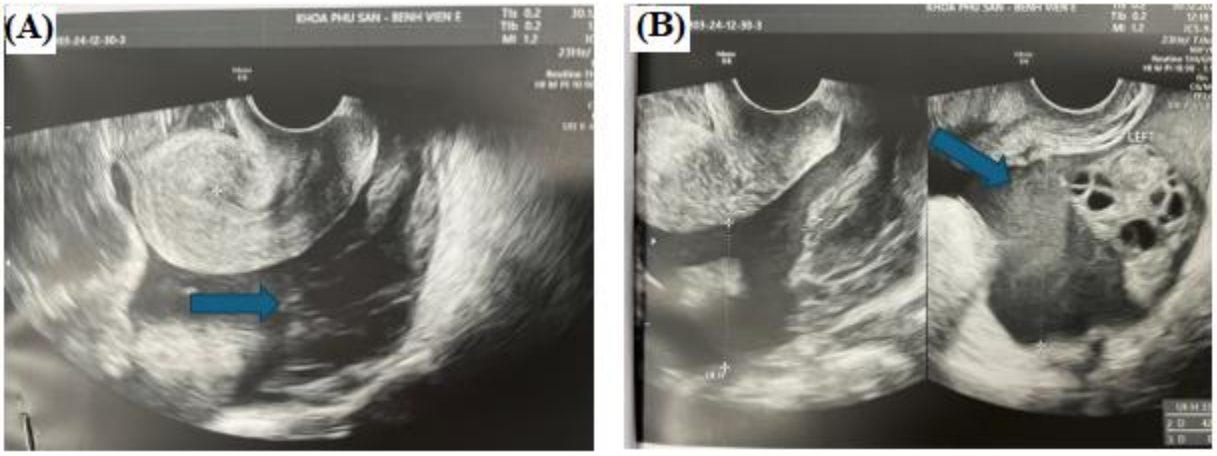

Fig. 1. Ultrasonography showing (A) a large amount of blood in the abdominal cavity and (B) a solid structure in the right pelvis (indicated by the arrows).

Blood test results were as follows: RBC 3.33 T/L, Hb 102 g/l, Hct 0.296 l/l, and β-hCG <2 mIU/ml. The patient underwent emergency surgery with a preliminary diagnosis of intra-abdominal bleeding due to a ruptured ovarian follicular cyst. During laparoscopy, a large volume of fresh blood was found in the abdominal cavity, and both ovaries appeared normal. An actively bleeding tumour was detected at the caecal base, extending into the pelvis near the right ovary (Fig. 2). Immediate frozen section histopathology revealed that the tumour tissue consisted of spindle cells, with an increased nuclear-to-cytoplasmic ratio and rare mitoses. During surgery, a friable, scabrous tumour mass accompanied by multiple enlarged mesenteric lymph nodes was identified. Given the intraoperative suspicion of malignant LMS, wide excision and lymph node dissection were performed to evaluate the presence of nodal metastases. An immediate biopsy confirmed a leiomyoma, excluding gastrointestinal stromal tumours (GIST). The surgical procedure involved the complete resection of the ascending colon, caecum (which contained the tumour), appendix, and a segment of the ileum, along with mesenteric lymph node dissection. Metastasis was identified in 7 of the 11 lymph nodes (Fig. 3). An anastomosis between the ileum and transverse colon was then performed. The initial procedure was performed laparoscopically; however, upon intraoperative identification of a suspected malignant LMS, the operation was converted to an open approach for wide excision and lymph node dissection.

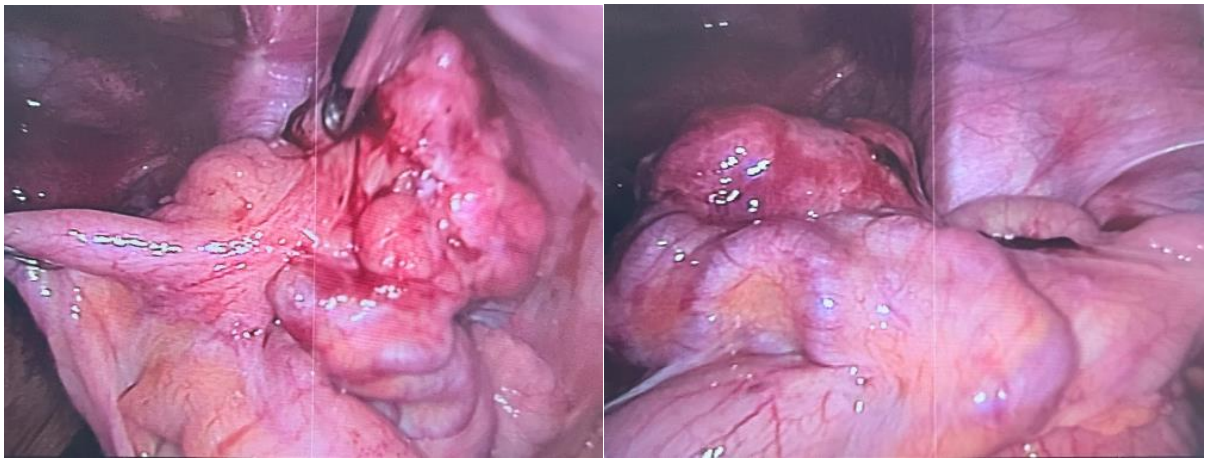

Fig. 2. Image of bleeding tumour at the caecal base.

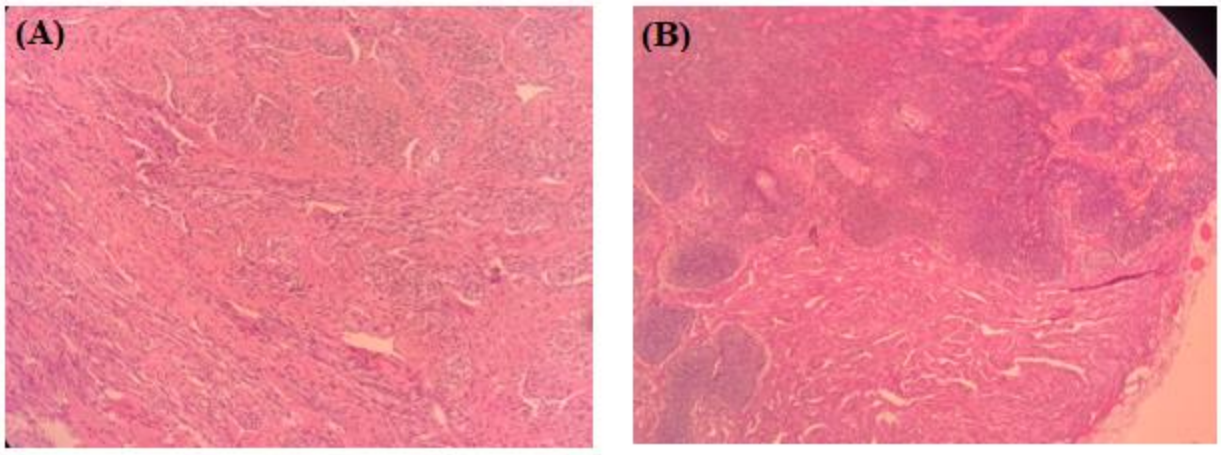

Fig. 3. Histological images showing (A) numerous spindle cells of the tumour and (B) the spindle cells metastasising to lymph nodes (Haematoxylin and Eosin staining; 10x).

Histopathological examination (H&E) and IHC staining confirmed the diagnosis of colonic LMS (Desmin and SMA positive; CD34, CD117, and DOG1 negative) (Figs. 3 and 4). The sample also tested negative for CD117, DOG1, and CD34, further confirming its smooth muscle origin and consistency with colonic LMS. Post-operatively, the patient was clinically monitored without additional specific treatment and was subsequently referred to the Oncology Department for further management and follow-up.

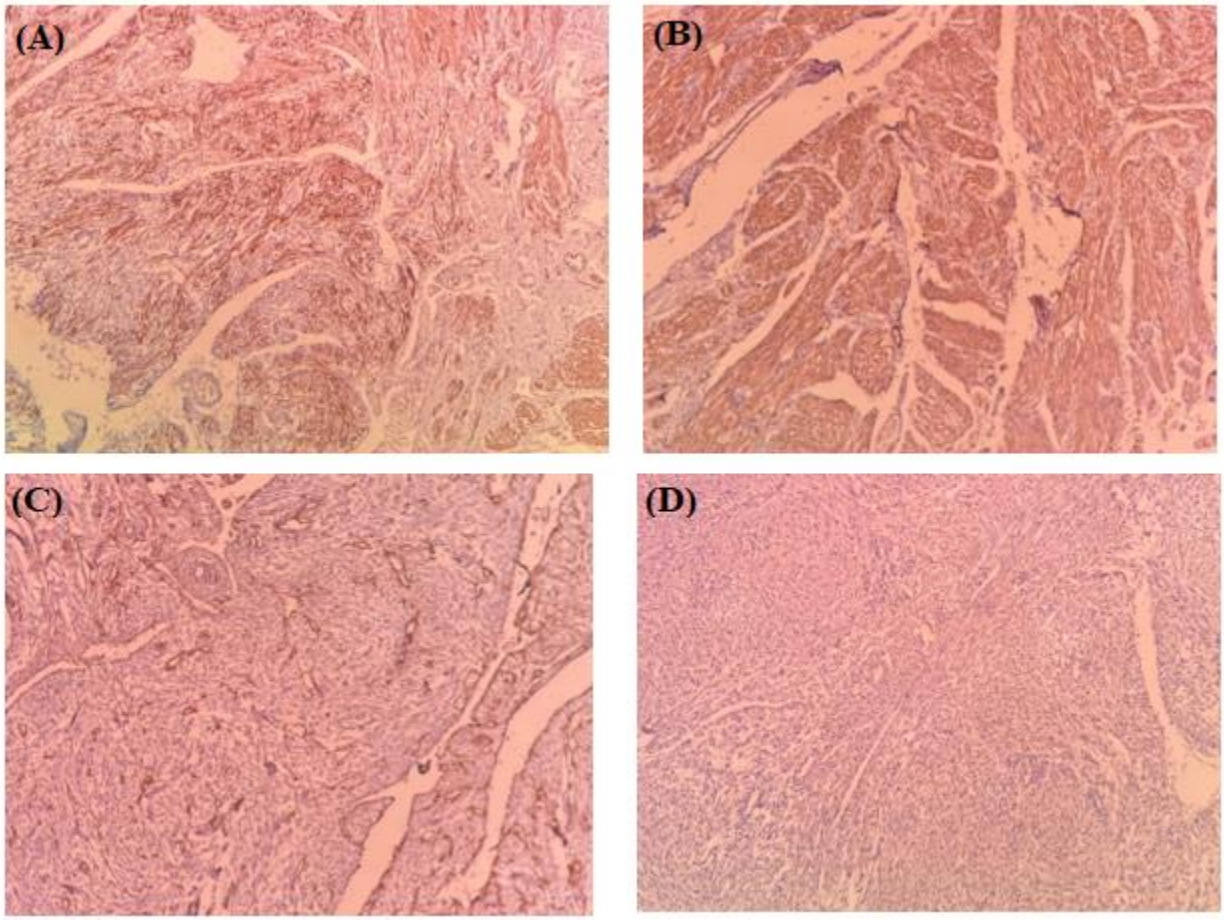

Fig. 4. Immunohistochemistry showing spindle cells positive for Desmin (A) and SMA (B), and spindle cells negative for CD34 (C), CD117, DOG1 (D) (Immunohistochemistry staining; 10x).

3. Discussion

Colonic LMS are rare malignant tumours, accounting for approximately 0.1% of all malignant tumours of the colon [2]. The clinical manifestations of colonic LMS are non-specific and are often discovered incidentally during colonoscopy. Some tumours appear polypoid, while others develop within the colonic wall and may protrude into the lumen as submucosal masses. Symptoms depend on the tumour's size and location. Tumours larger than 2 cm that protrude into the colon may cause symptoms of subacute intestinal obstruction, such as abdominal pain, bloating, constipation, and bleeding [8, 9].

In young women, rectosigmoid LMS may present with non-specific clinical manifestations such as acute abdominal pain, pelvic fullness, or haemoperitoneum, which can closely resemble gynaecological emergencies. This overlap frequently directs initial diagnostic

consideration toward reproductive tract pathology. Among the most relevant differentials are ectopic pregnancy, ruptured ovarian cyst, ovarian neoplasms, tubo-ovarian abscess, and uterine fibroids with degeneration or sarcomatous transformation. Ectopic pregnancy is often the foremost consideration in reproductive-aged women presenting with acute abdominal pain and haemoperitoneum, particularly when accompanied by vaginal bleeding and adnexal tenderness. A positive serum β-hCG and ultrasonographic evidence of an adnexal gestational sac are key distinguishing features. Similarly, ruptured ovarian cysts, particularly haemorrhagic or functional types, may cause sudden pelvic pain and intra-abdominal bleeding, but are usually characterised by a collapsed cyst on ultrasonography and a negative β-hCG. Ovarian neoplasms, whether benign or malignant, can also mimic gastrointestinal tumours through mass effect, abdominal distension, or torsion, yet they typically demonstrate adnexal rather than bowel wall origin on imaging and may be associated with tumour marker abnormalities. Tubo-ovarian abscesses represent another important differential, especially in patients with fever, leucocytosis, or a history of pelvic infection, and they usually respond to antibiotic therapy. Finally, uterine fibroids - particularly when undergoing degeneration or sarcomatous transformation - may present as pelvic masses with pain and abnormal bleeding, but their uterine wall origin is typically demonstrable on MRI [10, 11]. In our case, the LMS of the rectum and sigmoid colon initially mimicked an ectopic pregnancy or ruptured ovarian cyst due to the acute pelvic pain, haemoperitoneum, and mass effect. Only through endoscopic surgery and subsequent histopathological evaluation was the correct diagnosis established. This highlights the importance of considering gastrointestinal mesenchymal tumours in the differential diagnosis of young women presenting with acute pelvic symptoms, particularly when initial gynaecological evaluations are inconclusive.

In laparoscopic imaging, colonic leiomyomas may resemble mucosal adenomas, making diagnosis challenging. H.H. Choi, et al. (2016) [8] reported that only 46% of colonic leiomyomas are correctly identified based on endoscopic appearance. For this type of lesion, en bloc resection and histopathological examination are required to confirm the diagnosis. Leiomyomas of the colon wall can be visualised via laparoscopy when they develop into the colorectal lumen. Some grow externally in the colonic lumen and, as in this case, are only visible on ultrasonography and CT scan. Unfortunately, our patient did not undergo a CT scan because she was misdiagnosed

with a ruptured ovarian follicular cyst and taken for emergency surgery. Colonic leiomyomas in the sigmoid or rectum can cause symptoms similar to gynaecological tumours, such as rectal bleeding and lower abdominal pain. The limited literature on the clinical characteristics of colonic LMS makes diagnosis challenging. Due to its malignant nature and high recurrence rate, it is essential to distinguish it from GIST, the most common mesenchymal neoplasms of the gastrointestinal tract. IHC staining results showed positivity for smooth muscle markers actin and desmin, but negativity for CD34, CD117, and S100 proteins. These findings supported the diagnosis of colonic LMS. By contrast, GISTs are typically positive for CD117 (C-Kit), CD34, and/or DOG-1 [2, 12, 13]. These features help distinguish leiomyomas from GISTs, which may appear similar under a microscope.

Intraoperative exploration, particularly via endoscopic surgery, serves not only as a therapeutic approach but also as a crucial diagnostic tool. Literature reports confirm that several rare intestinal pathologies - including LMS, gastrointestinal stromal tumours, and atypical inflammatory lesions - have been identified unexpectedly during surgical procedures initially undertaken for suspected gynecologic conditions [14]. This highlights the importance of maintaining a broad differential diagnosis when evaluating young female patients presenting with acute pelvic symptoms. Moreover, the intraoperative discovery of intestinal neoplasms underscores the limitations of imaging modalities in distinguishing between gynaecological and gastrointestinal pathologies in the pelvis. Surgical exploration, therefore, remains indispensable in cases where imaging results are equivocal or inconsistent with clinical findings. Such an approach ensures timely and accurate diagnosis, enabling appropriate surgical management and reducing the risk of misdiagnosis or delayed treatment.

Surgery is the most effective treatment for LMS, with complete resection offering the highest chance of cure. Any residual malignant cells can significantly increase the risk of local recurrence or metastasis. Therefore, an accurate diagnosis and adequate resection margins are essential. However, achieving a correct diagnosis before surgery is extremely difficult, and very few cases are confirmed through histology. Our patient underwent emergency surgery without sufficient preoperative information, such as CT imaging, laparoscopy, or histological evaluation. Moreover, because she was a young woman, doctors initially suspected a gynaecological cause rather than LMS, which is extremely rare. Fortunately, the surgeon performing the operation,

who specialised in oncology, requested an immediate biopsy and histological testing, which confirmed a leiomyoma. The surgical team then proceeded with a wide resection and mesenteric lymph node dissection. Postoperative histopathology, including H&E and IHC staining, further confirmed the diagnosis of colonic LMS. Ten days after the operation, a contrast-enhanced CT scan of the chest and abdomen revealed normal organs and no free fluid. The treatment strategies and the extent of resection depend on the tumour's nature, size, location, and mitotic count. G. Fujimoto, et al. (2019) [7] suggest that wide resection should be performed due to the difficulty of distinguishing benign from malignant tumours intraoperatively. Tumour size is an important prognostic factor, with tumours larger than 5 cm being suggestive of malignancy [8]. Although colonic leiomyomas generally have a good prognosis after complete resection, LMS is aggressive due to its malignant nature and high recurrence rate. For this reason, patients require close follow-up and adjuvant chemotherapy after surgery.

4. Conclusions

This rare case highlights the difficulty of diagnosing colonic LMS preoperatively, especially in young women. Most cases are identified incidentally or upon presentation with bleeding complications, often leading to misdiagnosis as a ruptured ovarian cyst or an ectopic pregnancy. Current guidelines recommend surgery as the primary treatment for colonic LMS, with intraoperative radiation and adjuvant chemotherapy following surgery or in cases of metastasis. In our case, a caecal LMS was accurately diagnosed through an immediate intraoperative biopsy. This report emphasises the critical role of intraoperative and postoperative histopathology in diagnosing and distinguishing LMS.

COMPETING INTERESTS

The authors declare that there is no conflict of interest regarding the publication of this article.

CRediT author statement

Anh Vu Hong: Conceptualisation, Writing original draft; Ha Vu Thi: Writing - Reviewing and Editing; Trung Nguyen Cong, Nam Trinh Thi Hoai, Thuy Nguyen Thi: Investigation, Data curation; Trung Le Thanh: Visualisation, Methodology.

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