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Case Series

Endometriosis Beyond the Usual: A Rare Case Series of Unusual Clinical Presentations Mimicking Malignancy, Mullerian Anomaly and Bowel Pathology

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ABSTRACT

Endometriosis is a benign, estrogen-dependent gynecological disorder defined by the presence of endometrial glands and stroma outside the uterine cavity. Although it most commonly involves pelvic organs such as the ovaries, pouch of Douglas, uterosacral ligaments, fallopian tubes and rectovaginal septum, it may occasionally present at extra-pelvic sites or mimic other surgical and gynecological conditions. Its clinical presentation may vary widely from chronic pelvic pain, dysmenorrhea and infertility to acute abdomen, bowel obstruction, pelvic mass, ascites or features suggestive of malignancy. Such atypical presentations may create diagnostic difficulty and may lead to suspicion of ovarian carcinoma, intestinal malignancy, bowel obstruction or congenital genital tract anomaly.

We present a rare case series of four reproductive-age women with endometriosis who presented with unusual clinical features mimicking different pathological entities. The first case presented as a pelvic mass with markedly elevated CA-125 and ascites, clinically resembling ovarian carcinoma. The second case presented as a Mullerian anomaly with rudimentary horn hematometra. The third case presented with features of small bowel obstruction, while the fourth case mimicked colonic malignancy with intestinal involvement. All patients underwent surgical management, and final diagnosis was established on histopathological examination. This case series highlights the deceptive nature of endometriosis and emphasizes the importance of maintaining a high index of suspicion in reproductive-age women presenting with pelvic mass, gastrointestinal symptoms, unexplained ascites or atypical abdominal pain.

Keywords: *Endometriosis; Ovarian carcinoma mimic; Mullerian anomaly; Bowel obstruction; Intestinal endometriosis.*

INTRODUCTION

Endometriosis is a chronic benign gynecological disorder characterized by the presence of functional endometrial glands and stroma outside the uterine cavity. It was first believed to have been described by Rokitansky in 1860. The disease predominantly affects women of reproductive age and is commonly associated with dysmenorrhea, dyspareunia, chronic pelvic pain and infertility. However, its clinical presentation is highly variable, and in some patients, endometriosis may remain asymptomatic or may present with unusual features that closely resemble malignant, inflammatory or congenital conditions.

The most commonly involved sites are the ovaries, followed by the pouch of Douglas, uterosacral ligaments, vesicouterine pouch, serosal surface of the uterus, fallopian tubes, round ligament and rectovaginal septum. Extra-pelvic or deeply infiltrating endometriosis may involve the bowel, urinary tract, abdominal wall, diaphragm or other rare locations. The reported prevalence of asymptomatic endometriosis is approximately 1–7% in women undergoing elective sterilization, 12–32% among women of reproductive age with pelvic pain, 9–50% in infertile women and nearly 50% in adolescents with chronic pelvic pain or dysmenorrhea.

In rare instances, advanced endometriosis may present with large pelvic masses, ascites, intestinal obstruction, abdominal distension, rectal symptoms or features suggestive of malignancy. Malignant transformation is rare and occurs in approximately 1% of cases, most commonly in the ovary, accounting for nearly 80% of endometriosis-associated malignancies. Because serum CA-125 may be markedly raised in endometriosis, especially in advanced disease, endometriotic lesions may clinically and radiologically mimic ovarian cancer. Similarly, bowel endometriosis may resemble intestinal malignancy or obstruction, while congenital Mullerian anomalies with obstructed outflow may mimic endometrioma or pelvic mass.

This case series describes four unusual presentations of endometriosis encountered in reproductive-age women. These cases highlight the diagnostic dilemma created by endometriosis and reinforce the need for careful clinical evaluation, appropriate imaging, surgical assessment and histopathological confirmation.

CASE SERIES

Case 1: Endometriotic Cyst Mimicking Ovarian Carcinoma

A 31-year-old woman, para 1 living 1, presented to the outpatient department with complaints of pain in the epigastrium and lower abdomen associated with constipation for three days. She also had vomiting and loose stools for one day. On examination, she was well built and her vital parameters were stable. General physical examination was unremarkable.

On abdominal examination, generalized tenderness was present over the whole abdomen. Per speculum examination showed a hypertrophied cervix with erosions present over the anterior lip of the cervix. Per vaginal examination revealed an anteverted uterus; however, the exact size of the uterus could not be clearly made out. Cervical motion tenderness was present, and bilateral fornices were extremely tender.

Routine hematological parameters were within normal limits. Serum CA-125 was markedly elevated at 2700 U/mL. X-ray abdomen erect was normal. Ultrasonography showed a pelvic mass lesion measuring approximately 8 × 6 × 7 cm with features of small bowel obstruction and moderate ascites. Contrast-enhanced computed tomography of the abdomen revealed an 8.7 × 6 × 5.5 cm cystic lesion in the pelvis anterior to the uterus, likely arising from the left adnexa. Endometrial thickness was 11 mm, with mild hemorrhagic ascites and omental thickening. Ultrasound-guided fine needle aspiration cytology was inconclusive and showed only degenerative cells with hemorrhagic material.

Considering the clinical presentation, high CA-125 level, pelvic mass, ascites and omental thickening, a differential diagnosis of ovarian malignancy was strongly considered along with endometriosis. After obtaining high-risk informed consent, exploratory laparotomy was performed.

Intraoperatively, two large masses measuring approximately 10 × 5 cm and 10 × 8 cm were seen in the left adnexa. The masses were cystic in consistency and were removed. The omentum was stained with chocolate-colored material. The bowel loops and omentum were inspected for injury. The right fallopian tube and ovary were normal, and the uterus appeared normal. The postoperative period was uneventful. The patient was started on injection leuprolide for six months.

Histopathological examination showed an external glistening surface with congested red blood vessels and attached fallopian tube. The final impression was consistent with an endometriotic cyst.

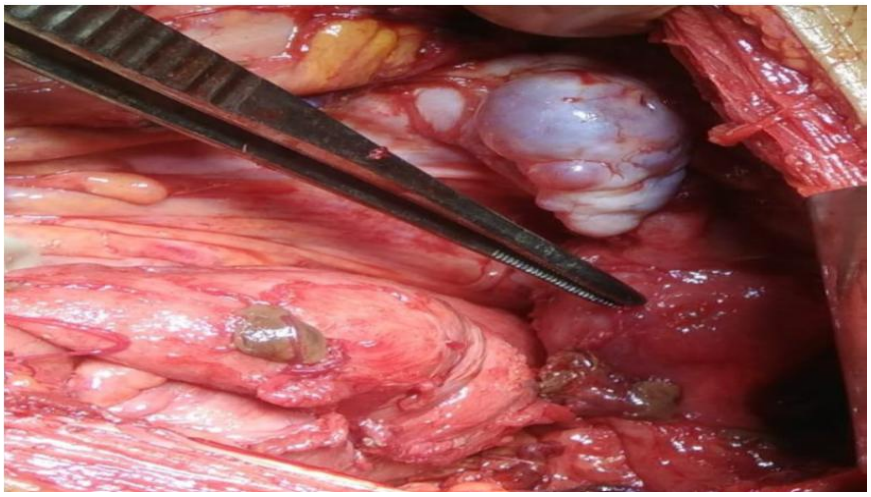

Figure 1: Intraoperative image showing left adnexal endometriotic cyst with chocolate-colored material and omental staining.

Case 2: Endometriosis Associated with Mullerian Anomaly and Rudimentary Horn Hematometra

A 25-year-old woman, para 1 living 1, with a known case of atrial septal defect, presented to the emergency department with severe lower abdominal pain for four days, vomiting for three days and fever for two days. On examination, she was thin built and her vital parameters were stable. General examination revealed slight pectus carinatum.

On abdominal examination, a vague, firm, non-tender mass of approximately 14 weeks' size was palpable in the suprapubic region. Per speculum examination showed a healthy cervix with no discharge. Per vaginal examination revealed a firm pelvic mass of approximately 14 weeks' size with restricted mobility, and fullness was present in the posterior fornix. Bilateral fornices were free and non-tender. Per rectal examination was normal.

Routine hematological investigations were normal. Echocardiography showed a large ostium secundum atrial septal defect measuring 24 mm, with ejection fraction of 58%. Serum CA-125 was 91.1 U/mL. Chest X-ray posteroanterior view showed normally visualized bilateral hilar shadows, normal cardiac size, clear bilateral costophrenic angles and scoliosis of the underlying thoracic spine.

Ultrasonography showed a well-defined multiloculated thick-walled cystic lesion measuring approximately 14.3 × 7.9 cm with homogeneous ground-glass echoes, likely hemorrhagic in nature, located on the right side of midline and suggestive of a right adnexal mass. MRI abdomen revealed a uterine anomaly in the form of two separate horns. One horn measured 8.6 × 2.5 cm, was located towards the left of midline and communicated with the cervix and vaginal cavity. The other bilobed rudimentary horn measured 19 × 6.5 cm, was located towards the right side extending up to the midline and showed intrauterine fluid collection.

Intraoperatively, an 18 × 18 cm rudimentary horn was identified, with the right ovary and tube embedded within it and adherent to adjacent gut and omentum. The uterus with left fallopian tube and ovary was impacted in the left lateral pelvic wall. The communicating uterus was connected to the vagina, whereas the rudimentary horn along with cervix had no communication with the uterus or vagina. It contained approximately 150 cc of dark brown hematometra. The gut was malrotated, caecum was present on the left side, Meckel's diverticulum was identified, and the right ureter was absent. The rudimentary horn was separated from the surrounding structures, removed and sent for histopathological examination.

Histopathology showed a specimen of uterus with cervix, with cut section of the cervix showing hemorrhagic areas. The final impression was Class IIb Mullerian duct anomaly with hematometra.

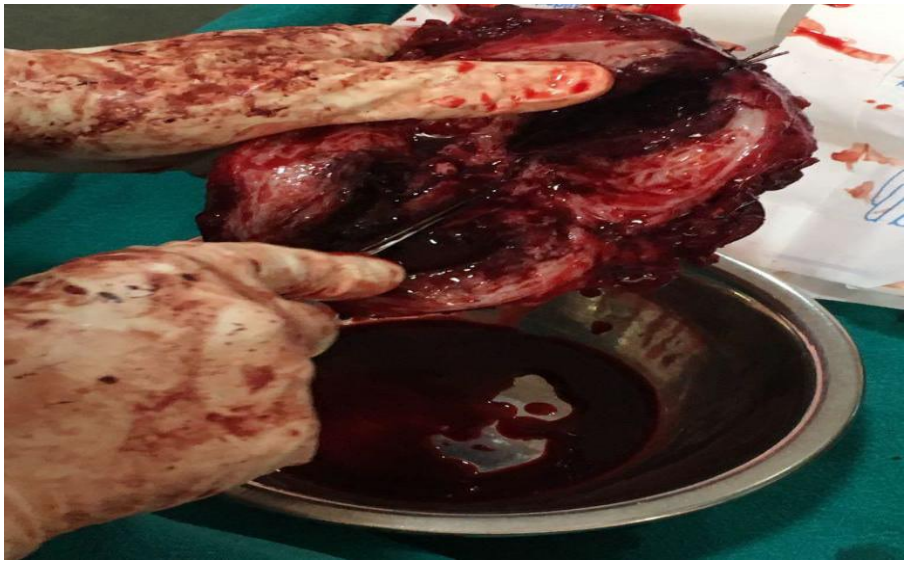

Figure 2: Intraoperative image showing large rudimentary horn with hematometra and adhesions to surrounding structures.

3: Endometriosis Presenting as Small Bowel Obstruction

A 35-year-old woman, para 1 living 1, presented with complaints of lower abdominal pain radiating to the back for one month. The pain was associated with nausea, vomiting and bloating. She also gave a history of weight loss for one month. On examination, she was moderately built. Abdominal distension was present, along with generalized tenderness over the whole abdomen.

Per speculum examination showed a hypertrophied cervix with multiple Nabothian follicles. Per vaginal examination revealed a retroverted uterus with bilateral fornicial fullness. Per rectal examination was normal. Routine hematological parameters were normal. Serum CA-125 was 109.45 U/mL.

Ultrasonography of the abdomen showed a normal-sized uterus with an ill-defined hypoechoic area measuring 40 × 30 × 26 mm in the posterior wall. Ascites was also present. Bilateral ovaries showed well-defined solid lesions measuring approximately 36 × 31 × 20 mm. A hypoechoic area measuring 12 × 9 × 5 mm was seen in the anterior aspect of the rectum.

Computed tomography of the whole abdomen showed small bowel obstruction with abrupt luminal narrowing, mesenteric and retroperitoneal lymphadenopathy, enlarged bilateral ovaries and multiple ovarian cysts. Considering the clinical and imaging features, a diagnosis of small bowel obstruction with tubo-ovarian pathology was considered, and surgical intervention was planned.

Intraoperatively, right and left tubo-ovarian masses were identified. Appendectomy was also performed. Histopathological examination of the right and left tubo-ovarian masses showed features of endometriosis.

Case 4: Intestinal Endometriosis Mimicking Colonic Malignancy

A 39-year-old woman, para 2 living 2, with a history of hysterectomy done two years earlier for abnormal uterine bleeding, presented with lower abdominal pain radiating to the back for one month. She also had vomiting, bloating and fullness of abdomen for 15 days.

On examination, she was moderately built. Per abdominal examination showed a soft abdomen with a midline scar. Per speculum examination revealed a healthy-looking vault. Per vaginal examination showed bilateral fornicial fullness and tenderness. Routine hematological parameters were normal. Serum CA-125 was elevated at 634.3 U/mL. Ultrasonography of the abdomen showed a minimal amount of free fluid with septations in the right iliac fossa.

Intraoperatively, bilateral ovaries showed simple cysts. There was involvement of the right colon with deposits of endometrial glands and stroma. Surgical management included bilateral oophorectomy and right hemicolectomy.

Histopathological examination showed bilateral simple ovarian cysts. Right hemicolectomy specimen showed endometrial glands and stroma involving the serosal adipose tissue and muscularis propria. The final impression was endometriosis involving the intestinal wall, omentum and skin.

HISTOPATHOLOGICAL SUMMARY OF ALL FOUR CASES

Case	Clinical Mimic	Operative Specimen	Histopathological Diagnosis
Case 1	Ovarian carcinoma	Left adnexal cystic masses with attached fallopian tube	Endometriotic cyst
Case 2	Mullerian anomaly with pelvic mass	Rudimentary horn with cervix and hematometra	Class IIb Mullerian duct anomaly with hematometra
Case 3	Small bowel obstruction	Right and left tubo-ovarian masses with appendix	Endometriosis involving tubo-ovarian masses
Case 4	Colonic malignancy	Bilateral ovaries and right hemicolectomy specimen	Endometriosis involving intestinal wall, omentum and skin

DISCUSSION

Endometriosis is a chronic gynecological disorder in which endometrial tissue is found outside the uterine cavity. Although it commonly remains confined to the pelvis, its presentation can be extremely variable. It may mimic malignant ovarian tumors, congenital genital tract anomalies, inflammatory pelvic disease, gastrointestinal obstruction or intestinal malignancy. This diagnostic complexity makes endometriosis one of the most deceptive benign conditions encountered in gynecological practice.

The exact pathogenesis of endometriosis remains unclear. Several theories have been proposed, including retrograde menstruation, coelomic metaplasia, lymphatic and vascular spread, endometrial stem cell implantation and Mullerian remnant abnormalities. The disease is considered multifactorial, involving genetic predisposition, altered immune response, hormonal influence and abnormal peritoneal environment. Increased inflammatory mediators such as interleukins, tumor necrosis factor and prostaglandins have been reported in endometriotic lesions, contributing to pain, adhesions and chronic inflammatory response.

The first case in this series presented with features mimicking ovarian carcinoma. The presence of pelvic mass, ascites, omental thickening and markedly elevated CA-125 created strong suspicion of malignancy. However, histopathology confirmed endometriotic cyst. This case highlights an important clinical issue: CA-125 is not a tumor-specific marker. It may be elevated in ovarian malignancy, but it may also rise significantly in benign gynecological conditions such as endometriosis, adenomyosis, leiomyoma and pelvic inflammatory disease. Therefore, CA-125 should be interpreted cautiously and in combination with clinical findings and imaging features.

The second case demonstrated the association of obstructive Mullerian anomaly with a clinical picture mimicking adnexal mass. MRI played a crucial role in identifying the uterine anomaly and differentiating the communicating uterine horn from the non-communicating rudimentary horn. Mullerian anomalies can present with cyclical abdominal pain, pelvic mass, hematometra and endometriosis-like symptoms. In such cases, MRI is the modality of choice because it accurately defines uterine anatomy, communication with the cervix and vagina, associated renal anomalies and relationship with adjacent pelvic structures.

The third case presented with features of small bowel obstruction. Gastrointestinal endometriosis can involve the rectosigmoid colon, small bowel, appendix and caecum. Patients may present with colicky abdominal pain, nausea, vomiting, constipation, diarrhea, abdominal distension, rectal bleeding, tenesmus or pain during defecation. Bowel involvement may be difficult to diagnose preoperatively because symptoms overlap with inflammatory bowel disease, bowel obstruction and malignancy. In reproductive-age women presenting with unexplained bowel symptoms and pelvic findings, endometriosis should be considered in the differential diagnosis.

The fourth case mimicked colonic malignancy. The patient had a prior hysterectomy and presented with abdominal pain, bloating and fullness. Intraoperative and histopathological findings revealed endometriosis involving the intestinal wall, omentum and skin. Intestinal endometriosis may involve the serosa, muscularis propria and rarely mucosa. Since mucosal involvement is uncommon, colonoscopy may be normal in many cases unless the lesion protrudes into the lumen. Deep infiltrating endometriosis involving bowel often requires a multidisciplinary surgical approach with gynecologist and colorectal surgeon.

Imaging is essential in preoperative assessment. Ultrasonography is often the first-line modality, especially for ovarian endometrioma, but MRI is superior in mapping deep infiltrating endometriosis, Mullerian anomalies and complex pelvic disease. Contrast-enhanced MRI helps differentiate benign and malignant ovarian masses by assessing solid components, papillary projections, mural nodules, hemorrhagic content and infiltration. Endometriosis-associated malignancy is rare but should be considered when a large hemorrhagic cystic mass contains an enhancing mural nodule.

Laparoscopy with histopathological confirmation remains the gold standard for diagnosis of endometriosis. However, in cases with extensive disease, suspected malignancy, bowel involvement or complex pelvic anatomy, laparotomy may be required. Surgical planning must be individualized according to site, extent of disease, fertility desire, severity of symptoms and involvement of adjacent organs. Medical management includes combined oral contraceptives, progestins, gonadotropin-releasing hormone agonists or antagonists, selective progesterone receptor modulators, non-steroidal anti-inflammatory drugs, aromatase inhibitors and danazol. In severe disease, surgical excision combined with long-term hormonal suppression may reduce recurrence.

This case series emphasizes that endometriosis is not always a straightforward diagnosis. It may present as ovarian cancer, obstructive Mullerian anomaly, bowel obstruction or colonic malignancy. A high index of clinical suspicion is required, particularly in women of reproductive age presenting with pelvic pain, adnexal mass, raised CA-125, unexplained bowel symptoms or complex pelvic lesions. Early recognition, appropriate imaging, surgical expertise and histopathological confirmation are essential for accurate diagnosis and effective management.

CONCLUSION

Endometriosis is a deceptive benign disease with a wide spectrum of clinical presentations, ranging from classical pelvic pain to rare presentations mimicking ovarian carcinoma, Mullerian anomaly, small bowel obstruction and colonic malignancy. Each patient may present differently, and diagnosis requires a high level of clinical suspicion, careful pelvic examination, appropriate imaging and histopathological confirmation. A multidisciplinary approach involving gynecologists, radiologists, urologists and colorectal surgeons is often required in extensive or atypical cases. Early recognition and individualized management can reduce morbidity, prevent delayed diagnosis and improve reproductive and surgical outcomes in women of reproductive age.

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