Rare Malignant Transformation of Endometriosis into Endometrioid Adenocarcinoma of the Rectosigmoid: Case Report

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Background

- Malignant transformation of endometriosis occurs in 0.7–1% of endometriosis patients.
- The colon and rectum are involved in only 5% of endometriosis associated malignancies.
- These tumors can mimic primary colonic adenocarcinoma and can be misdiagnosed as colorectal malignancy.
- This is rare case of postmenopausal woman without predisposing factors who presented with rectosigmoid tumor of endometrioid origin.

Case Presentation

- Asymptomatic 81 year-old female presented with positive Cologuard test.
- She had prior remote history of right sided complex adnexal mass and underwent laparoscopic salpingoophorectomy.
- Colonoscopy done(Figure 1), it showed infiltrative mass at the rectosigmoid junction, involving one-third of the lumen circumference.
- Biopsy revealed at least high-grade dysplasia.
- Endorectal ultrasound was done during attempted endoscopic mucosal resection (Figure 2), and suggested invasion into the muscularis propria.
- CT chest, abdomen, and pelvis revealed rectosigmoid thickening without metastatic disease (Figure 3).
- The patient underwent robotic low anterior resection.
- Surgical pathology revealed moderately differentiated invasive endometrioid adenocarcinoma with multifocal residual benign ectopic endometriosis with negative margins and no nodal involvement.
- By immunohistochemistry, the tumor cells were diffusely and strongly positive to CK7, patchy positive to PAX8, but were negative to CK20, CD2, positive to estrogen receptors (ER) and progesterone receptors (PR).

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Case Presentation

- Tumor markers including CEA, CA 19-9, and CA 125 were all within normal limit.
- Transvaginal ultrasound was previously normal.
- Genetic molecular testing showed proficient Mismatch Repair status (MMR-p), Microsatellite Instability low (MSI-stable), ER+, PR+.
- The patient declined adjuvant chemotherapy but agreed to Letrozole therapy. Patient agreed to continue surveillance.

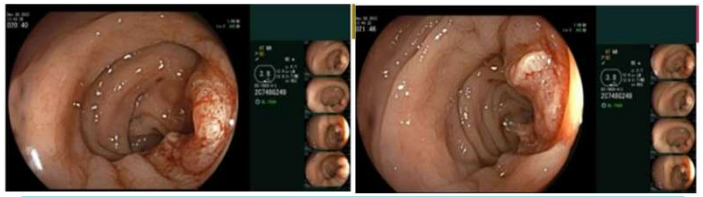


Figure 1

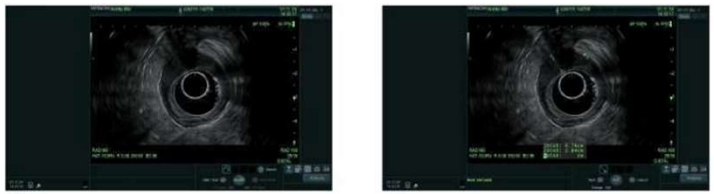


Figure 2

Conclusion

- We present a rare case of endometriosis-associated intestinal tumor in a postmenopausal patient without prior history of endometriosis nor hormone replacement therapy.
- Endometriosis-associated neoplasm in extragonadal locations is a rare entity.
- It requires a high index of suspicion in addition to careful pathologic and immunohistochemical analysis.
- The diagnosis is often made in postoperative specimen with a specific immunohistochemical pattern (CK7+/CK20−).
- Literature is lacking and further research is needed to standardize targeted approach to this disease entity.

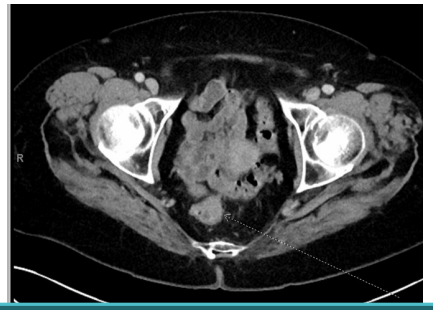


Figure 3

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