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Late Retroperitoneal Metastasis from Invasive Ductal Carcinoma of the Breast: Diagnostic Challenges and the Role of Biopsy and Immunohistochemical Profiling — A Case Report

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ABSTRACT

Abstract:

Invasive ductal carcinoma (IDC) is the most prevalent invasive breast cancer. Breast cancer metastasis displays varied patterns and occurs in several organs, with rare occurrence in some regions, including the gastrointestinal system and retroperitoneum. The symptoms of breast cancer metastasis are always nonspecific. Here, we present a female case of retroperitoneal metastasis from IDC involving the cecum and terminal ileum with a late diagnosis. The patient is a 48-year-old female diagnosed with right breast IDC who received neoadjuvant chemotherapy. She subsequently underwent a right modified radical mastectomy with right axillary dissection, followed by adjuvant endocrine therapy and adjuvant radiation therapy. She maintained regular clinical follow-up. Surveillance imaging then detected a possible local recurrence and a retroperitoneal mass invading the cecum that could be another primary. The biopsy from the right chest wall confirmed a recurrence of luminal B breast cancer, but the patient lost follow-up. In February 2025, the patient presented to the ER with a picture of small bowel obstruction (SBO) with other non-specific symptoms. The CT revealed that SBO was secondary to the locally invasive retroperitoneal mass that invaded the cecum and terminal ileum. The peritoneal biopsy was performed, and the histopathology report revealed infiltrative malignant cells with an immune profile consistent with metastatic carcinoma of mammary origin. Therefore, retroperitoneal metastasis from IDC can occur, although this is an unusual location for it. Furthermore, diagnosis of this metastasis after a long time of breast cancer surgery and therapy is a rare condition. Cross-sectional imaging, particularly CT, plays a crucial role in identifying unusual metastatic sites, while histopathological confirmation is necessary to determine the metastatic origin..

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1. INTRODUCTION

Breast cancer (BC) is the most prevalent cancer in women with varied morphology and behavior, and it is the leading reason for global female malignancy mortality [1]. IDC is the most frequent histopathological type of invasive BC, followed by invasive lobular carcinoma (ILC) [1]. IDC represents 80%, whereas ILC represents only 15% of cases [2]. BC metastasis is responsible for the majority of deaths related to BC [3]. The patterns of BC metastasis have been changed, especially among patients with advanced BC, due to the improvement of systemic and local therapy in recent decades [4, 5]. Metastasis of BC can occur in several organs, and the most frequent locations include the skin, liver, bone, and lung [6]. The gastrointestinal tract is more prone to metastasizing by lobular carcinoma, but the gastrointestinal system is a rare location.

for metastasis in IDC **[7]**, and BC retroperitoneal metastasis is rare **[1]**. The clinical manifestations of this disorder are always non-specific, and laboratory tests aren't always advantageous in diagnosis and assessment **[1]**. Literature regarding retroperitoneal metastasis is rare, especially metastasis related to IDC and late presentation and diagnosis. Therefore, here, we present a female case of retroperitoneal metastasis involving the cecum and terminal ileum from IDC with a late diagnosis.

Case presentation:

A 48-year-old premenopausal female with a family history of breast cancer presented to our hospital in August 2020 due to a right breast mass that she had noticed three years prior, which began to increase in size one year ago and was associated with pain, ulceration, and bleeding; however, she initially neglected it. Eventually, she sought medical help. She was diagnosed with right breast IDC, clinically staged as T4dN3bM0, grade 2, with ER 85%, PR 90%, HER2-negative status, and a Ki-67 index of 50%, corresponding to Stage IIIC disease. Genetic analysis revealed no pathogenic variants, although a PTCH variant of uncertain significance (VUS) was identified.

She completed neoadjuvant chemotherapy consisting of four cycles of dose-dense doxorubicin and cyclophosphamide, followed by weekly paclitaxel with carboplatin administered every three weeks for four cycles, with the final cycle given in January 2021.

In Feb 2021, she underwent a right modified radical mastectomy (MRM) with right axillary dissection. The final surgical pathology demonstrated residual IDC, grade 2, with multiple microscopic foci measuring up to 6 mm (ypT1b), showing therapy-related changes. The tumor was fibrotic and adherent to the latissimus dorsi and pectoralis minor muscles. A single focus of intermediate-grade ductal carcinoma in situ (DCIS) was identified. All surgical margins were free of malignancy, with no evidence of lymphovascular invasion (LVI) or perineural invasion (PNI). The skin showed fibrosis and treatment-related changes. Immunohistochemistry revealed ER positivity (90%), PR positivity (10%), and HER2-negative status. Axillary dissection yielded eight lymph nodes, all negative for metastasis with a treatment effect (ypN0).

In April 2021, she initiated adjuvant endocrine therapy with tamoxifen in combination with ovarian function suppression (OFS). This regimen was planned for two years, after which she would transition to letrozole with calcium supplementation, without OFS, for an additional five years. In July 2021, she completed adjuvant radiation therapy. She discontinued tamoxifen/OFS in September 2023 and transitioned to letrozole with calcium supplementation.

In September 2023, she had a CT CAP that revealed possible local recurrence and a retroperitoneal mass invading the cecum (Figure 1). In November 2023, a right chest wall biopsy confirmed recurrent luminal B breast cancer. Despite these findings, the patient subsequently lost follow-up and declined the recommended colonoscopy for tissue biopsy of the mass invading the cecum and terminal ileum. Although the procedure had been scheduled multiple times, she did not attend any of the appointments.

In February 2025, the patient presented to the ER with clinical features of SBO, including multiple episodes of whitish vomiting and associated nausea. For the past two months, she had been experiencing recurrent abdominal pain associated with diarrhea occurring 1 to 4 times per day. She did not report severe pain, fever, night sweats, unintentional weight loss, per rectal bleeding, melena, hematemesis, headache, visual disturbances, or projectile vomiting.

A CT scan was performed (Figure 2), which revealed that the small bowel obstruction (SBO) was secondary to a locally invasive retroperitoneal mass invading the cecum, appendix, terminal ileum, right psoas muscle, and distal ureter. This invasion resulted in upstream dilatation of the small bowel loop reaching up to 4.7 cm, indicating high-grade SBO; however, there were no signs of bowel ischemia or perforation.

She underwent a peritoneal biopsy under local anesthesia in Feb 2025. The histopathology report revealed two white cores measuring 0.3 cm and 1.7 cm in length. The core biopsy displayed infiltrative malignant cells arranged in cords, surrounded by a sclerotic background. The tumor cells were positive for the following immunohistochemical stains: pan-cytokeratin, cytokeratin 7, GATA3, and ER, whereas they were negative for PR, Her 2, CK20, CDX2, S100, and Desmin. The immunoprofile was consistent with metastatic carcinoma of mammary origin.

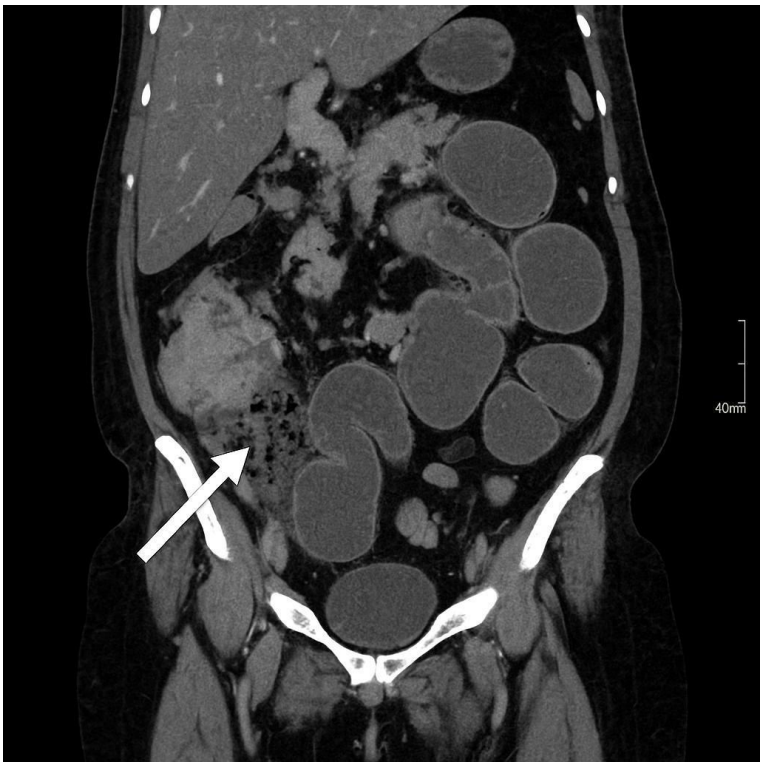

Fig. 1: Coronal contrast-enhanced CT of the abdomen and pelvis

The image demonstrates a right retroperitoneal soft-tissue mass (white arrow) invading the cecum and terminal ileum, with complete loss of the intervening fat planes. The mass causes upstream small-bowel dilatation, consistent with high-grade obstruction.

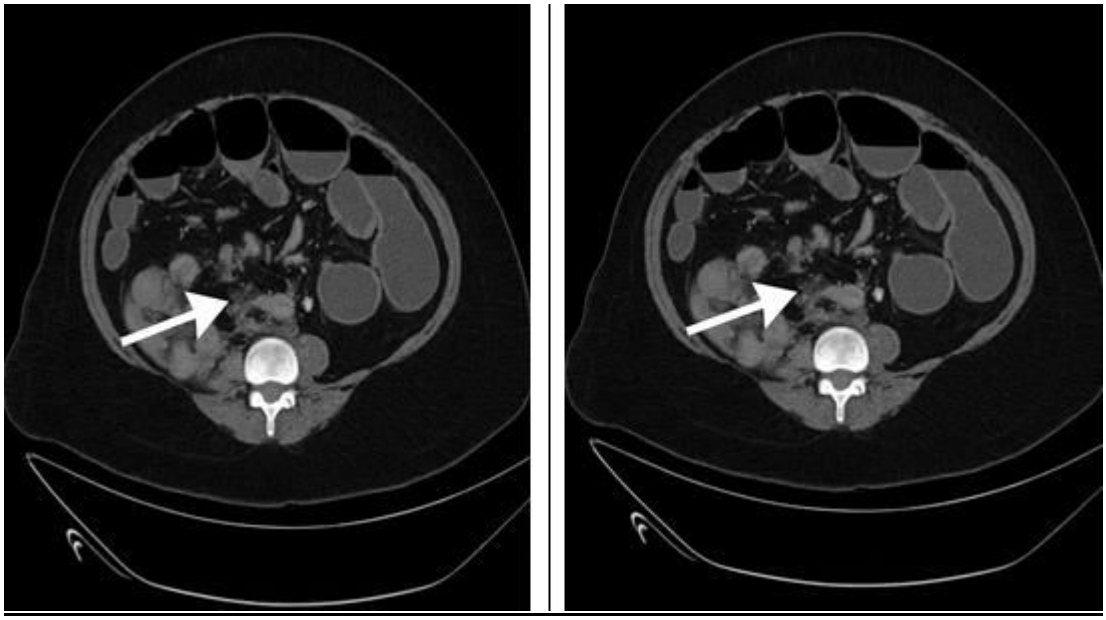

Fig 2: Axial contrast-enhanced CT of the abdomen

The axial image shows a right retroperitoneal soft-tissue mass (white arrow) measuring approximately 6.3 × 10.3 cm. The lesion invades the right psoas muscle, cecum, appendix, and terminal ileum, and encases the distal right ureter, resulting in upstream small-bowel dilatation (up to 4.7 cm) and moderate right hydroureteronephrosis. No CT evidence of bowel ischemia or free perforation is present.

Discussion:

Metastatic involvement of the gastrointestinal tract from IDC is exceptionally uncommon [7], and retroperitoneal metastasis from breast cancer is even rarer [1]. The reported prevalence of gastrointestinal metastases is approximately 6% in living patients and 31% in autopsy series [8], underscoring the diagnostic challenges associated with these atypical patterns of dissemination. This case represents a rare presentation of retroperitoneal metastasis from IDC, highlighting important diagnostic and clinical considerations.

The nonspecific clinical manifestations of retroperitoneal metastasis often delay its diagnosis [1]. Symptoms related to metastatic involvement of the colon may include abdominal pain, melena, weight loss, vomiting, and fatigue [9]. In our case, the patient presented with nonspecific abdominal symptoms, including vomiting, nausea, and abdominal pain, without red-flag signs such as melena or weight loss. Previous reports describe retroperitoneal metastases manifesting as retroperitoneal fibrosis with secondary complications including SBO, hydronephrosis, ureteral obstruction, or back pain [10, 11]. Our patient developed SBO, which is a known but nonspecific manifestation. CT imaging clarified the etiology, demonstrating a locally invasive retroperitoneal mass infiltrating the cecum and terminal ileum.

Endoscopy is the preferred diagnostic modality for evaluating gastrointestinal tract involvement in patients with a history of breast cancer and new gastrointestinal symptoms [12]. However, in this case, the patient repeatedly declined colonoscopy. Consequently, she underwent a peritoneal biopsy after CT imaging delineated the precise size and location of the retroperitoneal mass. Histopathological examination revealed infiltrative malignant cells with an immunohistochemical profile consistent with metastatic carcinoma of mammary origin, confirming that the retroperitoneal lesion represented metastatic breast cancer.

The diagnostic approach used in this case aligns with strategies described in previous reports, where CT imaging was first employed to identify the extent and position of the retroperitoneal mass, followed by image-guided core needle biopsy to establish a definitive diagnosis [1]. This underscores the value of cross-sectional imaging and tissue sampling, particularly when endoscopic evaluation is not feasible.

This case represents a rare presentation of retroperitoneal metastasis from IDC, involving the cecum and terminal ileum. While breast cancer commonly metastasizes to the lungs, brain, bones, and lymph nodes, retroperitoneal involvement is unusual [1]. Furthermore, IDC is less likely to metastasize to the gastrointestinal tract and retroperitoneum compared with ILC, which more frequently demonstrates diffuse infiltration of these sites [13]. Thus, the retroperitoneal spread observed in this patient highlights an atypical metastatic pattern for IDC.

The interval between primary breast cancer diagnosis and the development of gastrointestinal metastases is highly variable. Although rare, delayed presentations have been documented. One study reported a mean interval of seven years [9], while several cases described latency periods exceeding 20 years [7, 14, 15]. In our case, retroperitoneal metastasis was diagnosed five years after the initial breast cancer diagnosis, indicating a relatively delayed and clinically silent progression.

Conclusion:

Retroperitoneal metastasis from IDC is exceedingly rare, as the retroperitoneum is an unusual site of breast cancer dissemination. Moreover, delayed diagnosis of such metastasis is uncommon. This case illustrates the value of maintaining a high index of suspicion for atypical metastatic patterns in patients with a history of breast cancer who present with unexplained abdominal or retroperitoneal symptoms. Cross-sectional imaging, especially CT, is very important for finding unusual places where cancer has spread. Once a mass is found, a tissue biopsy is still very important for making a final diagnosis. Although rare, retroperitoneal involvement should be considered in the differential diagnosis of new abdominal or pelvic masses in breast cancer survivors.

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