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Desmoid-Type Fibromatosis of the Breast Mimicking Carcinoma in a 26-Year-Old Female: A Case Report and Discussion of B-Catenin Negative Diagnostic Challenges

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Abstract

Desmoid-Type Fibromatosis (DTF) is a rare, non-metastasizing, but locally aggressive soft tissue tumor developing from monoclonal myofibroblast growth. It is generally connected to mutations in the CTNNB1 gene (encoding β-catenin) or, less commonly, the APC gene in Familial Adenomatous Polyposis (FAP) patients. While lacking metastatic potential, DTF produces severe morbidity due to infiltrative development, high recurrence rates, and an unpredictable clinical outcome that might include spontaneous remission or chronic progression. It most usually affects young adults (peak age 30-40), with a slight female predominance, and can arise in practically any anatomical place, albeit it is unusual in the breast. We report a case of a 26-year-old Saudi female who came with a solid, painless lump in the left inframammary fold. Initial imaging and core biopsy were inconclusive. Although immunohistochemistry was negative for nuclear β-catenin, a subsequent excisional biopsy confirmed the diagnosis based on conventional morphological characteristics and a low proliferative index. We aimed to highlight the diagnostic challenges of β-catenin-negative breast fibromatosis through a case analysis supported by immunohistochemical and morphological evidence. This example underscores the diagnostic difficulties of breast DTF, particularly in β-catenin negative cases. It underlines the requirement of a multidisciplinary approach and complete histological examination to avoid misdiagnosis, overtreatment, and to minimize unnecessary invasive operations.

Keywords: Desmoid-type fibromatosis; Breast; β-catenin negative; Diagnostic; Case report

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Introduction

Desmoid-type Fibromatosis (DF) is an uncommon mesenchymal tumor, accounting for only roughly 0.2% of all breast tumors. While histologically benign and devoid of metastatic potential, it is marked by aggressive local infiltration and a high rate of recurrence. Breast DF often closely mimics breast cancer both on physical exam and imaging, resulting to diagnostic and treatment ambiguity. The illness is categorized into three primary types: spontaneous, linked with Familial Adenomatous Polyposis (FAP), and extra-abdominal. Herein, we offer a diagnostically problematic case of a left breast desmoid tumor in a young female, confirmed by excisional biopsy despite unusual immunohistochemical features [1-3].

Materials and Methods

Study Design

This is a case report of a 26-year-old Saudi female patient diagnosed with desmoid-type fibromatosis (DF) of the breast at King Salman bin Abdulaziz Medical City. The study was conducted in accordance with ethical standards, and informed consent was obtained from the patient.

Clinical evaluation

The patient arrived with a palpable breast nodule. A detailed clinical history was acquired, including personal and family history. A physical examination of the breast was performed to define the nodule, which was found in the left inframammary fold.

Diagnostic imaging

An initial breast ultrasound was conducted to analyze the characteristics of the palpable lesion.

Pathological Assessment

Core needle biopsy

An ultrasound-guided core needle biopsy of the breast lesion was conducted. The collected tissue samples were processed for histological evaluation using standard Hematoxylin and Eosin (H&E) staining.

Immunohistochemistry (IHC)

The core biopsy specimen was submitted to immunohistochemistry analysis utilizing a panel of antibodies to help diagnosis. The indicators tested included: Pan-Cytokeratin (Pan-CK), Cytokeratin 7 (CK7), High Molecular Weight Cytokeratin (HMWCK), Smooth Muscle Actin (SMA), P63, Beta-catenin (β-catenin), and Calponin. The Ki-67 proliferation index was also calculated.

Excisional biopsy

Due to unclear findings from the core biopsy, a definitive excisional lumpectomy with re-excision of the tumor boundaries was performed. The entire surgical specimen was sent for histological investigation.

Post-excision IHC

The lumpectomy specimen received further immunohistochemistry examination with an enlarged antibody panel, including SMA, S100, β-catenin, Pan-CK, Desmin, and CD34. β-catenin IHC was repeated, and the Ki-67 index was re-evaluated to confirm the initial findings and ensure diagnostic accuracy.

Diagnostic criteria

The final diagnosis of desmoid-type fibromatosis was established based on a combination of characteristic morphological features on H&E staining (Figure 1a, 1b), the immunohistochemical profile (positive for SMA and S100, negative for epithelial and other specific markers), and a low Ki-67 proliferation index, despite the absence of nuclear β-catenin staining. The diagnostic reasoning involved systematically excluding key differential diagnoses. The absence of Pan-CK and P63 staining helped rule out fibromatosis-like metaplastic carcinoma. The tumor's morphology and IHC profile (e.g., positive SMA, negative CD34 and Desmin) distinguished it from myofibroblastoma, nodular fasciitis, and low-grade fibrosarcoma.

Data analysis

This report is based on a descriptive study of the patient's clinical, radiological, and histological data.

Results and Discussion

Breast Desmoid Fibromatosis (BDF) is an uncommon condition that often radiologically resembles breast cancer. It mainly affects women in the 20-40 age bracket and can be connected with variables such as trauma or hormonal impacts. Our patient fulfills this demographic profile.

The principal diagnostic issue in this instance was the absence of nuclear β-catenin staining, which is a critical immunohistochemistry marker seen in around 80% of DF patients. This marker is a critical distinction from metaplastic carcinoma. However, it is known that up to 20% of DF patients might be β-catenin negative. In such instances, diagnosis must rely on a combination of classic morphology (as illustrated in Figure 1a and 1b), a low proliferative index, and the absence of markers for other entities in the differential diagnosis, which in this case included fibromatosis-like metaplastic carcinoma, myofibroblastoma, nodular fasciitis, and low-grade fibrosarcoma. This instance indicates that early and accurate diagnosis can avert more invasive surgical treatments.

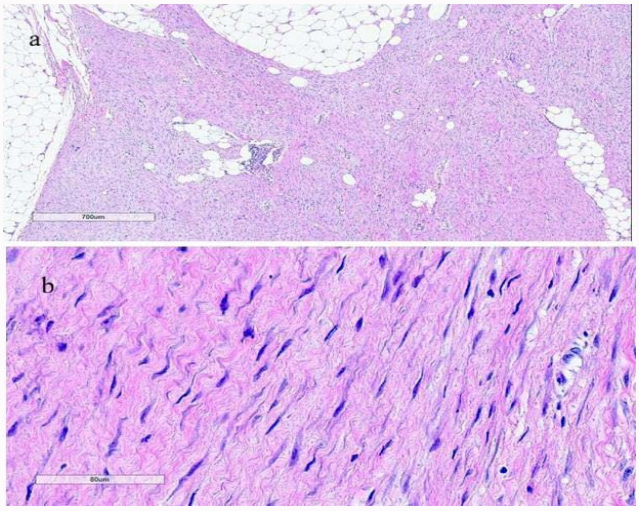

Figure 1: Histopathological examination of the excised breast lesion: **(a)** H&E section from breast involved by desmoid fibromatosis shows poorly demarcated spindle cell tumor with infiltrative borders, **(b)** Tumor composed of long intersecting fascicles showing bland spindle cells with indistinct borders, hyperchromatic nuclei, eosinophilic cytoplasm, and no mitotic figures.

The management of DF is developing. While active surveillance is now the recommended first-line method for stable, asymptomatic disease, excision remains a conventional technique when the diagnosis is questionable or when vital structures are endangered by tumor progression, as was a consideration here. The presence of implicated margins in our case prompted crucial considerations regarding the probability of local recurrence. The patient was referred to the oncology team for discussion of adjuvant radiotherapy; however, in a multidisciplinary meeting, it was found that she did not require radio- or chemotherapy, a decision consistent with the 2020 global consensus recommendations which favor surveillance for many cases, even with positive margins [6]. This underscores the significance of tailored management planning in line with the 2020 global consensus recommendations. The patient was placed on a regimen of active surveillance with clinical and radiological follow-up every 6 months [4-9].

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