International Surgery Journal
Alquthami GF et al. Int Surg J. 2026 Jan;13(1):101-104
http://www.ijsurgery.com
pISSN 2349-3305 | eISSN 2349-2902

DOI: https://dx.doi.org/10.18203/2349-2902.isj20254198

Case Report

Desmoid-type fibromatosis of the breast with positive margins: a case report and review of conservative management

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Received: 20 November 2025
Accepted: 15 December 2025

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ABSTRACT

Breast desmoid-type fibromatosis (BDF) is a rare, benign, but locally aggressive tumor that mimics breast carcinoma. Management has shifted toward conservative strategies, including active surveillance, especially for asymptomatic or indolent lesions. A 29-year-old woman presented with a 2 cm firm, immobile mass in the left inframammary fold. Ultrasound and magnetic resonance imaging (MRI) revealed an irregular hypoechoic lesion. Core needle biopsy showed spindle cell proliferation. Wide local excision confirmed desmoid-type fibromatosis, with nuclear β-catenin and SMA positivity, CD34 and S-100 negativity, and Ki-67 <5%. The superior margin was positive, while other margins were clear. Considering the low proliferative index and absence of aggressive features, the patient was managed with active surveillance and regular clinical and radiological follow-up. BDF can be safely monitored in selected patients, even with positive margins, avoiding unnecessary surgery while ensuring close follow-up for progression.

Keywords: Breast fibromatosis, Desmoid tumor, β-catenin, Active surveillance, Conservative management

INTRODUCTION

Desmoid-type fibromatosis of the breast (BDF) is an uncommon, benign but locally aggressive neoplasm of mesenchymal origin. It accounts for approximately 0.2-0.3% of all breast tumors and about 4% of extra-abdominal desmoid tumors, making it a true diagnostic rarity.[1,2] Although histologically benign and lacking metastatic potential, BDF demonstrates a unique biological behavior characterized by infiltrative growth, a propensity for local recurrence, and a clinical course that often mimics invasive breast carcinoma.[1,3]

Clinically, BDF usually presents as a painless, firm, irregular breast mass that may be tethered to skin, muscle, or chest wall, occasionally causing dimpling or nipple retraction.2,4 Radiologically, the lesion resembles malignancy: mammography typically reveals a spiculated, irregular mass, while ultrasound shows a hypoechoic lesion with posterior acoustic shadowing.[3,5] Magnetic resonance imaging (MRI) helps assess the extent of invasion, though no imaging modality is diagnostic. Thus, core needle biopsy or excisional biopsy is required for definitive diagnosis.[2,5]

At the molecular level, most BDF cases harbor CTNNB1 (β-catenin) mutations, leading to aberrant activation of the Wnt/β-catenin pathway.[3,6] A smaller subset is associated with APC mutations in familial adenomatous polyposis (FAP) or Gardner syndrome.[6] Risk factors include prior breast trauma, implants, or surgery, with some series reporting up to 40% of patients having a surgical history.[1,4]

Management of BDF has shifted significantly in recent years. Historically, wide local excision with negative margins was the standard. However, recurrence rates of 18–39% have been reported, even after apparently complete resections.[2,3] Current consensus guidelines increasingly favor active surveillance ("watch-and-wait"), particularly for asymptomatic or stable lesions, as

spontaneous regression occurs in a subset of patients.[3,6] When intervention is necessary, options include repeat surgery, radiotherapy, systemic agents (NSAIDs, antiestrogens, tyrosine kinase inhibitors), and newer targeted therapies such as nirogacestat, a γ-secretase inhibitor recently approved for desmoid tumors.[6]

Given its rarity, BDF remains diagnostically and therapeutically challenging. We present a case of a young woman with BDF excised with positive margins and managed conservatively, highlighting the role of active surveillance.

CASE REPORT

A 29-year-old woman with no family history of breast cancer or genetic syndromes presented with a palpable left breast mass at the inframammary fold. She had no prior breast surgery, trauma, or radiation exposure.

Examination

The lesion was firm, immobile, and adherent to overlying skin, which showed localized pigmentation but no ulceration or dimpling. No nipple discharge or axillary lymphadenopathy was present. Clinical size was ~2 cm.

Imaging

Ultrasound demonstrated an irregular, hypoechoic mass without calcifications or vascularity, MRI demonstrate left breast mass BIRAD 6 the mass abutting chest wall muscle but no clear invasion (Figures 1 and 2).

Pathology

Core needle biopsy revealed spindle cell proliferation, raising suspicion for a stromal lesion. The patient underwent wide local excision. Gross margins were assessed intraoperatively and appeared clear.

Final histopathology confirmed desmoid-type fibromatosis.

Immunohistochemistry

Nuclear βcatenin positive, SMA positive, CD34 and S-100 negative, Ki-67 <5% was found.

Margins

Superior, margin positive; tumor <2 mm from the skin; deep and lateral margins was clear.

Post-operative course

The case was discussed in a multidisciplinary meeting. Given the positive superior margin, proximity to skin, but otherwise indolent histology and negative deep margins, the options of re-excision versus surveillance were considered. Based on international consensus guidelines, the decision was made to pursue active surveillance with serial clinical and radiological follow-up.

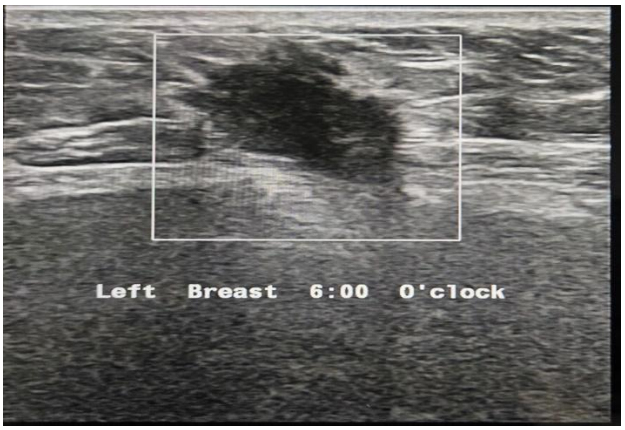

Figure 1: Ultrasound of left breast lesion.

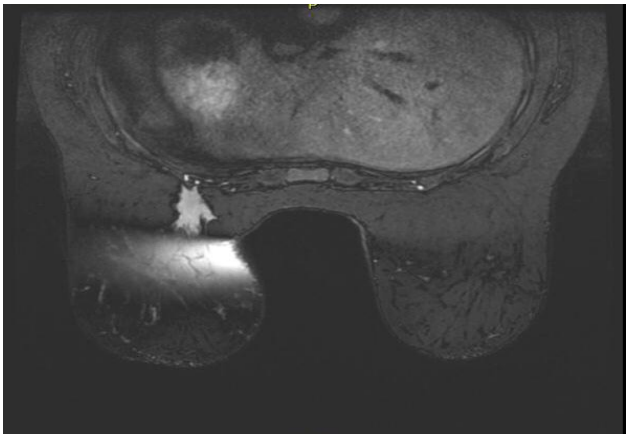

Figure 2: MRI of left breast lesion.

DISCUSSION

BDF represents a rare diagnostic and therapeutic challenge due to its paradoxical behavior: although histologically benign and lacking metastatic potential, it exhibits a locally infiltrative course with a high risk of recurrence.[1-3]

The condition accounts for only 0.2–0.3% of all breast tumors, yet its clinical and radiological presentation frequently mimics invasive carcinoma, often leading to initial diagnostic confusion.[2,4]

Etiology and risk factors

BDF is most commonly sporadic, but it may arise in the context of trauma, prior breast surgery, or implant placement, with reported associations in up to 40% of patients.[1,4,5] Genetic predispositions have also been implicated: CTNNB1 (β-catenin) mutations are found in 65–85% of cases, while APC mutations occur in the setting

of familial adenomatous polyposis (FAP) and Gardner syndrome.[3,6]

Hormonal influences and prior radiotherapy have also been described as potential contributors.[7] In the present case, no risk factors were identified, underscoring the sporadic nature of most presentations.

Clinical and radiological presentation

Clinically, BDF typically manifests as a firm, painless, irregular breast mass, which may be adherent to the skin or chest wall and can occasionally cause dimpling or nipple retraction.[2,4,7] Imaging modalities often fail to distinguish it from carcinoma.

Mammography may reveal a spiculated, irregular lesion, while ultrasound commonly shows a hypoechoic mass with posterior acoustic shadowing, often without vascularity or calcification.[5,8] MRI plays an important role when chest wall or muscular involvement is suspected, as it provides superior assessment of soft tissue infiltration.[8,9]

Pathology and immunohistochemistry

Histologically, BDF is characterized by bland spindle cells arranged in sweeping fascicles with finger-like extensions into adjacent parenchyma and adipose tissue.[9,10]

Immunohistochemically, nuclear β-catenin expression is considered a hallmark, seen in up to 80% of cases.[10,11] Other markers include positivity for smooth muscle actin (SMA) and variable desmin, while CD34 and S-100 are negative, helping exclude other spindle cell lesions such as phyllodes tumor or sarcoma.[9,11] The low proliferative index (Ki-67 <5%) observed in our patient further supports the indolent biological behavior.

Management

Traditionally, wide local excision with negative margins was considered the standard of care. However, recurrence rates remain significant, reported between 18% and 39%.[2,3,12]

Importantly, several studies have shown that margin status does not consistently predict recurrence, challenging the historical emphasis on radical resection.[12,13] Instead, there has been a global paradigm shift toward conservative management.

The 2020 joint global consensus guidelines recommend active surveillance as the first-line strategy for most newly diagnosed desmoid tumors, including BDF, particularly when asymptomatic and not threatening vital structures.[14]

Evidence supporting this approach comes from a large French study in which 35% of patients experienced spontaneous regression, 52% remained stable, and only 12% showed progression requiring treatment.[15]

When to intervene?

Active treatment is indicated when the tumor is progressive (rapid increase in size, pain, skin involvement, functional compromisation), symptomatic or chest wall invasion), vascular encasement) structures- in such scenarios, management options include.[14]

Surgery

Reserved for accessible, resectable tumors, particularly when cosmesis can be preserved.

Radiotherapy

Considered for unresectable, recurrent, or progressive disease, though evidence in breast fibromatosis remains limited.[13]

Systemic therapy

Options include NSAIDs, antiestrogen agents ((tamoxifen tyrosine kinase inhibitors (sorafenib, sunitinib), low-dose chemotherapy methotrexate + vinblastine), and the recently approved γ-secretase inhibitor nirogacestat, which demonstrated promising efficacy and tolerability in desmoid tumors.[6,14]

Our case in context

Our patient underwent excision with a positive superior margin (<2 mm from skin) but had a low Ki-67 index and no aggressive clinical features. In alignment with global consensus guidelines, we adopted active surveillance rather than re-excision, aiming to balance disease control with preservation of breast integrity. This case reinforces the evolving treatment philosophy that emphasizes function- and cosmesis-preserving strategies, reserving surgical or systemic intervention for progressive or symptomatic disease.

CONCLUSION

Breast desmoid-type fibromatosis is a rare and diagnostically challenging entity that is often prone to over treatment. Contemporary evidence and international guidelines now advocate active surveillance as an appropriate first-line strategy, even in cases with positive margins, provided that no clinical or radiological progression is observed. This approach allows for preservation of breast cosmesis, minimization of morbidity, and timely intervention only for patients demonstrating disease progression on follow-up.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Alquthami GF, Alsharif NF, Alqashqari KY. Desmoid-type fibromatosis of the breast with positive margins: a case report and review of conservative management. Int Surg J 2026;13:101-4.