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When Nature Heals: A Case of Spontaneous Remission in Breast Desmoid Tumor

Soufiane Benaazza[1*], Asaad Elbakkari[1], Sanae Amalik[1], Fatimazahra Laamrani[1], Youssef Omor[1], Rachida Latib[1]

[1]Radiology Department, National Institute of Oncology INO, Rabat, Morocco

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***Corresponding author:** Soufiane Benaazza
Radiology Department, National Institute of Oncology INO, Rabat, Morocco

Abstract **Case Report**

Desmoid tumors are rare mesenchymal neoplasms arising from musculoaponeurotic structures, which affects predominantly women. They are characterized by aggressive local invasion, with no metastatic potential but a high propensity for local recurrence. Imaging modalities such as ultrasound, CT, and MRI are essential for assessing local extension, guiding surgical planning, and monitoring for recurrence. Histological analysis remains the gold standard to confirm the diagnosis. Treatment options range from surgical resection to non-surgical approaches, including chemotherapy, radiotherapy, and hormonal therapy such as tamoxifen. Recent guidelines increasingly advocate for active surveillance as an initial strategy, particularly to minimize treatment-related morbidity, given the potential for spontaneous regression in some cases. We report the case of a 73-year-old woman who presented with a palpable mass in the left breast, exhibiting some suspicious features on Ultrasound. Histological analysis confirmed the diagnosis of mammary fibromatosis. Surgical resection was initially performed, and the Follow-up imaging with CT and MRI later demonstrated a local recurrence with infiltration of adjacent structures. The case was reviewed in a multidisciplinary tumor board [MDT], where a decision was made to pursue active surveillance. Remarkably, MRI follow-up after three months revealed spontaneous regression of the recurrent soft tissue mass.

Keywords: Desmoid tumor, mammary fibromatosis, recurrence, spontaneous regression.

INTRODUCTION

Desmoid tumors are rare neoplasms characterized by their high local aggressiveness and tendency to recur, despite lacking metastatic potential [1]. These tumors can arise in various anatomical locations, including the abdominal cavity, shoulder, and breast [2]. their management represent a real challenge for the clinician, highlighting the need for discussion within a multidisciplinary team meeting. treatment may include surgery, medical therapy, or in some cases, active surveillance, as spontaneous regression has been observed [2,3]. We report the case of a patient who underwent two surgeries at our institution for a breast desmoid tumor, then experienced a recurrence of the disease, and later showed spontaneous regression.

CASE PRESENTATION

We report the case of a 73-year-old postmenopausal woman who presented with a newly appeared left breast mass, first noticed one month ago. Clinical examination revealed a subcutaneous, mobile mass measuring approximately 3 cm, located at the junction of the internal quadrants of the left breast, with no skin changes, nipple discharge, or palpable lymphadenopathy. A complementary breast ultrasound showed a suspicious hypoechoic mass in the same location [9 o'clock position], characterized by irregular contours, with no detectable axillary or supraclavicular lymphadenopathy.

A biopsy was requested to confirm the nature of the mass, which was found to be a spindle cell tumor with an immunohistochemical profile suggestive of mammary fibromatosis. A wide local excision with margins was performed, and the histopathological analysis confirmed the diagnosis. However, tumor involvement at the margins was identified, necessitating a re-excision of the tumor bed to achieve negative margins.

A follow-up CT scan performed one month later revealed tissue infiltration at the lumpectomy site, with involvement of the thoracic wall and the sterno-costal cartilage [figure 1].

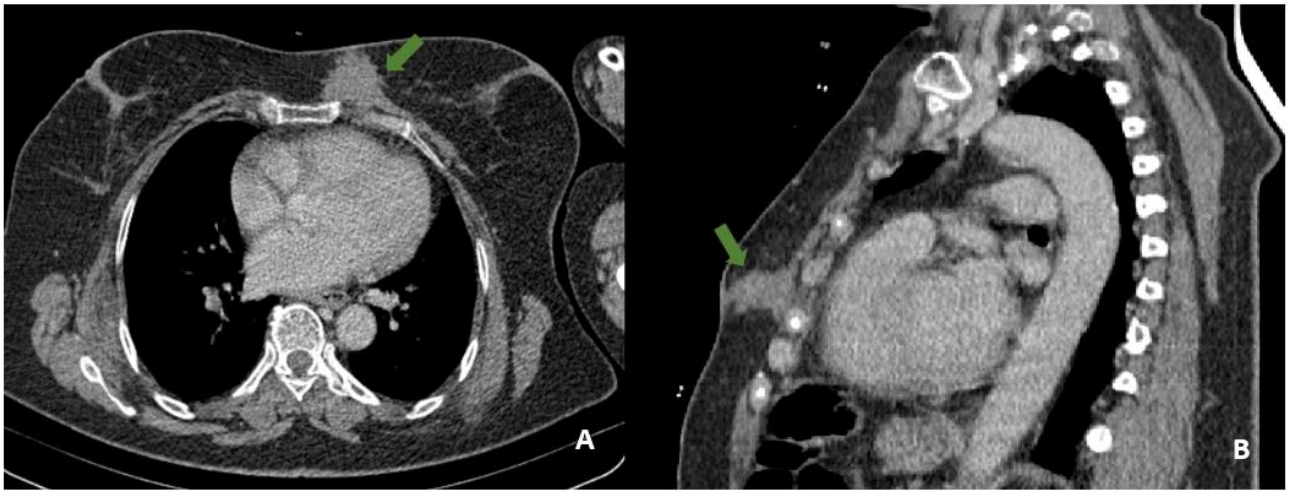

Figure 1 : Axial [A] and sagittal [B] images of a chest CT, revealing tissue infiltration in the inner quadrant of the left breast, with involvement of the thoracic wall [green arrow]

An additional MRI was conducted to better assess the mass's relationships and its extension to adjacent structures. It confirmed the presence of cutaneous and subcutaneous tissue infiltration at the lumpectomy scar, extending to the ipsilateral pectoral muscle and chondrocostal cartilage, strongly suggestive of tumor recurrence [Figure 2].

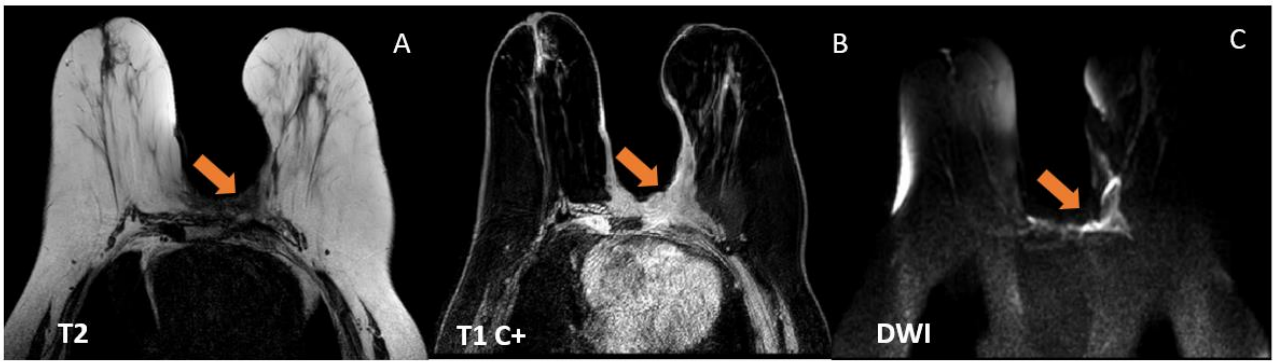

Figure 2 : Axial slices from a breast MRI revealing cutaneous and subcutaneous infiltration adjacent to the lumpectomy scar, with intermediate T2 signal intensity, diffusion hyperintensity, and contrast enhancement, extending to the ipsilateral pectoral muscle and chondrocostal cartilage

Given the surgical difficulty, the patient was discussed in a multidisciplinary tumor board [MDT], with no indication to initiate chemotherapy or radiotherapy. Active surveillance was recommended. A follow-up MRI after three months showed regression of the tissular infiltration at the lumpectomy site in the inner quadrant of the left breast, with no signs of tumor activity, suggesting spontaneous regression of the lesion [Figure 3].

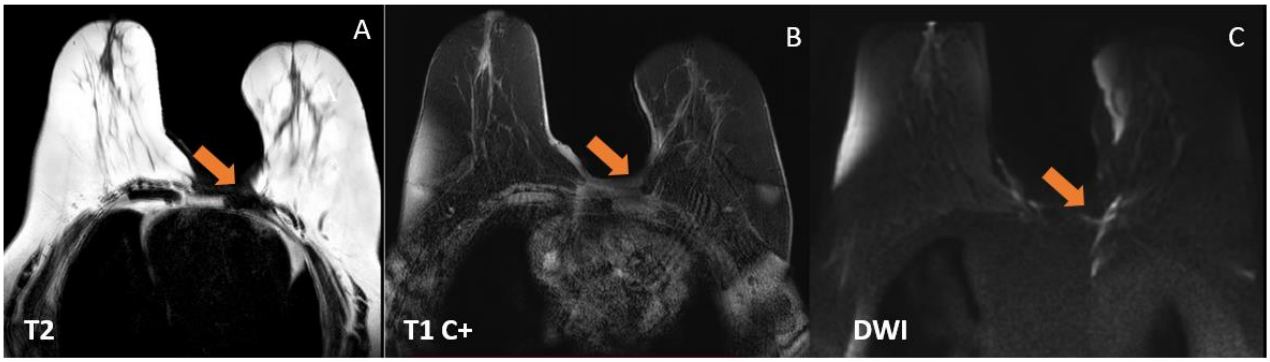

Figure 3 : axial slices of a breast MRI revealing a regression of the tissular infiltration at the lumpectomy site

DISCUSSION

Desmoid tumors [DT], also referred to as aggressive or musculoaponeurotic fibromatosis, are mesenchymal neoplasms that histopathologically present as monoclonal fibroblastic proliferations originating from musculoaponeurotic structures. This type of tumors does not metastasize but are locally invasive, and have a high recurrence rate, leading generally to significant morbidity and mortality [1,2].

Desmoid tumors are rare, with an estimated incidence of 2 to 4 cases per million people, accounting for approximately 0.03% of all neoplasms. They often occur between the ages of 30 and 40 years and affect predominantly women with a ratio of 3:1 [1,3]. These tumors can develop in various anatomical sites, involving most commonly the hip and shoulder girdles, abdominal wall, intra-abdominal cavity, and breast [2]. Patients with a history of prior surgery, trauma, or pregnancy are more prone to developing these lesions [1,3], without forgetting that augmentation mammoplasty can increase the risk of breast desmoid tumors, as it was described in multiple case reports [2,4,5].

These tumors, including breast localizations, are generally linked to beta-catenin-activating mutations, primarily involving the Wnt/beta-catenin/APC pathway [6,7]. Mutations in *CTNNB1* and *APC* genes contribute to beta-catenin accumulation, as these genes play a key role in its degradation, resulting sometimes in tumor formation. A strong association is observed between the development of desmoid tumors and patients with familial adenomatous polyposis [FAP], who have a higher incidence of APC genes mutations [6,8].

Concerning desmoid tumors of the breast, they appear as a painless, firm lump that may be either mobile or attached to the skin or chest wall. Some other local signs may be present such as nipple retraction or skin dimplings [orange skin appearance]. Generally, no axillary lymphadenopathies are found. Upon presentation, the clinician must first rule out malignancy due to the frequently suspicious clinical and radiological findings [2].

Imaging modalities are very helpful for diagnosis, on mammography these lesions can present as a stellate lesion and may contain microcalcifications, features usually linked to primary breast carcinoma [9]. Its sensitivity in detecting abnormalities ranges between 62% and 78.5% [10,11]. On ultrasound, Desmoid tumors appear as an irregular hypoechoic mass with indistinct margins and posterior acoustic shadowing, this imaging modality offers greater accuracy in detecting the lesion [2,12].

CT and MRI are employed to evaluate local extension and determine surgical planes, with MRI being more advantageous [1,2]. On CT, desmoid tumors usually appear as a well-circumscribed mass, enhancing after contrast administration, even though some cases exhibit a more aggressive appearance with ill-defined margins [13]. On MRI, they typically appear hypo- to isointense on T1-weighted images and heterogeneously hyperintense on T2-weighted images, dynamic MRI generally reveals gradual enhancement, likely due to the tumor's high collagen content and myxoid changes. However, enhancement patterns may vary as some cases of breast fibromatosis show rapid enhancement followed by washout on dynamic MRI, resembling invasive carcinoma. In such cases, the absence of peripheral ring enhancement, a characteristic feature of breast cancer, can help distinguish between the two [14].

Histological confirmation is essential for the diagnosis of breast desmoid tumors before initiating treatment [1,2]. Studies have found that core needle biopsy has a greater accuracy than fine needle biopsy, with an accuracy that can reach up to 97% when performed by an experienced operator [3]. Desmoid tumors are marked histologically by the proliferation of uniform spindle-shaped cells resembling myofibroblasts, embedded in a collagen-rich stroma with a prominent vascular network [1]. Immunohistochemistry is also crucial to confirm the diagnosis, as these tumors are typically positive for beta-catenin, smooth muscle actin, and vimentin, while they are negative for cytokeratin as well as estrogen, progesterone, and androgen receptors [2].

Surgery has traditionally been the primary treatment for breast fibromatosis [3,15]. Due to the significant risk of local recurrence, a wide excision with margins of at least 1 cm is indicated, followed by close surveillance. When surgical margins are positive, a second surgery is usually recommended. Recurrence rates reported in the literature range between 5% and 25%, with most cases occurring within three to six years after the initial surgery. In cases of multiple recurrences, mastectomy with reconstruction may be considered [15,16].

Non-surgical modalities such as radiotherapy, NSAIDs, tamoxifen, and chemotherapy are generally considered second-line treatments given the limited evidence supporting their effectiveness. They may be appropriate when surgery poses significant morbidity due to tumor location or patient-related factors, or as adjuvant therapy in cases of positive [R1] margins [1,2].

Active surveillance may be the most appropriate approach in certain cases. Recently, the European Society for Medical Oncology [ESMO] recommended a "watch-and-wait" strategy for managing desmoid tumors at various sites, particularly in the breast

[17]. This is supported by findings that approximately 35% of patients monitored without surgery experienced spontaneous regression [18]. Therefore, in cases of minimally symptomatic tumors, observation should be considered as a first-line option, with a clinical evaluation and imaging examinations conducted every three months [3].

CONCLUSION

Breast desmoid tumor [DT] is a rare entity that should be considered as a differential diagnosis of lesions suspicious for breast carcinoma. Given the complexity of both diagnosis and management, a multidisciplinary approach involving oncologists, pathologists, and radiologists is essential to optimize treatment and minimize morbidity. While surgery remains a valid therapeutic option, preserving function and quality of life is a priority. As such, medical therapies may represent an alternative, and in some cases, active surveillance alone is justified, since many patients with desmoid tumors may experience spontaneous regression or stabilization of the disease.

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