CASE PRESENTATION

DESmoid Tumor Masquerading as Fibroadenoma. Case Report and Review of Literature

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Abstract

We present a case of 19 years old women initially diagnosed with fibroadenoma of the breast. She was referred to us for further evaluation of the growing mass. Sonography revealed a non vascular, hypo echoic irregular lesion. Biopsy of the lesion revealed the typical histology of a desmoid tumor. She underwent wide local excision, with post-operative histopathology confirmation of negative surgical margins.

Keywords: Breast, Desmoid, fibromatosis, fibroadenoma

Introduction

Desmoid tumors of the breast are rare fibroblastic tumors that comprise 0.2% of all breast tumors. Although benign, they are often locally invasive and have a high rate of local recurrence. Clinical presentation, sonography and mammography often mimic carcinoma. Histologically, the lesion, which is composed of spindle cell proliferation and collagen deposition, can help in making the diagnosis. Management remains controversial and the mainstay of treatment is considered to be wide local excision with clear margins.

This case is unique for the initial misdiagnosis of the lesion as a benign tumor, which can be attributed to the patient’s young age and unremarkable medical history as well as lack of awareness to the existence of this rare lesion. Diagnosis is important due to the invasiveness of the tumor and wide local excision must be performed in order to try to prevent local recurrence.

Case

A previously healthy 19 years old woman presented to her local clinic with a newly found solitary, firm and mobile mass in her left breast. At first she was diagnosed as having a fibroadenoma. After complaining of the mass growing larger over the course of six months, she was referred to our hospital for further evaluation.

The patient had no prior history of trauma or surgery to the area. Family history was unremarkable. On clinical examination a non-tender, superficial, mobile and well defined 2 cm mass was palpated superiorly to the areola, on the upper outer quarter of the left breast. Axillary lymph nodes were not palpated. No signs of inflammation or nipple discharge were found. The patient was sent for a sonography which revealed a hypo echoic irregular
lesion measuring 2.1 cm, without any significant vascularity, Figure 1 and 2.

Based on these findings, it was decided to perform a Tru-Cut needle biopsy of the lesion. Biopsy revealed mild fibroblastic proliferation and increased collagenization with no atypia or mitoses, a finding consistent with fibromatosis. After consulting the oncologists and relaying the information to the patient, a wide excision of the lesion was performed.

During the operation a mass measuring about 2X2 cm was excised with additional 1 cm borders. The lesion was confined to the breast tissue and did not involve the underlying fascia. Post-operative histopathology revealed foci of spindle cell proliferation with fine collagen deposits and some myxoid areas, Figure 3.

No nuclear atypia or pleomorphism was seen, and mitotic index was low. Staining for Vimentin and Beta-Catenin was positive, Figure 4.

Removal of the lesion was complete with negative surgical margins. The diagnosis of fibromatosis was confirmed.

**Discussion**

Desmoid tumor, or aggressive fibromatosis, is a benign mesenchymal monoclonal proliferations arising from the connective tissue of the muscle and overlying the aponeurosis or fascia. Although it does not metastasize, it is characterized by an infiltrative growth pattern and carries a high rate of local recurrence [1,2]. Tumor-related mortality is rare, and morbidity is attributed mostly to disfigurement and loss of function from local progression or treatment [3].

Desmoid tumors represent 1.5-3.5% of all connective tissue tumors [4]. Pathogenesis is unknown, but hormonal and genetic factors, as well as local trauma or surgery have all been suggested. The majority of desmoid tumors occur sporadically, while the minority is associated Gardner’s syndrome [5]. A higher prevalence among females as well as increase in tumor size during pregnancy suggests estrogen may also play a role in the pathogenesis [1].
Desmoid tumor of the breast is even extremely rare and comprise 0.2% of all breast tumors [1]. These tumors can arise either primarily from the breast parenchyma, or by secondary infiltration from the musculo-aponeurotic layer of the pectoral muscles [6,7]. Most cases of breast desmoid tumors occur in women with only rare reports of affected males [8]. Desmoid tumor of the breast is thought to be more prevalent following surgical intervention to the affected area, but can also arise de novo [1,9]. Some recent case reports described cases following saline or silicone implants, implying a possible relationship to the prosthetic's capsule [7,8].

On physical examination the lesion is often palpated as a firm, movable mass. Skin retraction or dimpling may be present. Typical mammography shows irregularly shaped, uncalcified, high-density mass with spiculated margins. By sonography the tumor appears to be a poorly marginated, hypo echoic mass. Thus clinical presentation and imaging can often mimic carcinoma [4,7,10,11].

MRI can be useful in distinguishing desmoid tumor from other malignancies of the breast, as well as revealing the extent of the tumor and relationship to adjacent structures. The mass appears ovoid or irregular, crosses fascial compartments, is isointense on T1- and heterogeneously hyperintense on T2-weighted images and shows bands of low signal on all sequences. The lesions displayed moderate to strong heterogeneous enhancement after IV administration of contrast material [12,13].

Histologically the lesion is composed of spindle cell proliferation with varying amounts of collagen deposition, sometimes with focal myxoid areas. Fingerlike extensions into adjacent breast parenchyma and adipose tissue can be seen. The tumor cells are often organized in broad sheets, in storiform configuration or interlacing bundles with herringbone pattern. Overall cellularity is low to moderate, and mitotic figures are rare. Immunostaining shows variable reactivity for desmin and actin, and nuclear staining for Beta-Catenin is supportive but not pathognomonic [14,15].

Management remains controversial, since cases are scarce and in the literature there are mostly case reports with no larger study. Mainstay of treatment is considered to be wide local excision with clear margins. A high local recurrence rate after excision is reported by several series, ranging from 24% to 77% at 10 years [16]. Unfavorable risk factors related to recurrence after excision are young age, tumor > 3 cm, and arguably a microscopic positive surgical margin. Interestingly the impact of microscopic margin status in predicting recurrence is unclear, as some studies showed positive resection margin to be predictive of recurrence while others found no such correlation [1,16-19]. Recurrence rate for secondary breast lesions, arising from the musculo-aponeurotic tissue of the pectoral muscle, is much higher than that of primary mammary fibromatosis, which is reported to be 21-27% [20].

Radiation therapy can provide good local control of desmoid tumor as a supplement to surgery or even as the only treatment. The current trend is that when wide local excision will result with significant compromise of function or when surgical margins are positive, adjuvant radiotherapy should be considered [1,17,18].

Systemic treatment such as hormonal and anti-inflammatory agents, chemotherapy and biological treatment have all been used with mixed results and are generally reserved for cases in which local control is not feasible [3,19,21,22].

In conclusion, our case of desmoid tumor of the breast is unusual as it masqueraded as a different benign disease - fibroadenoma. It was different from most published cases, in which the lesion was described as mimicking carcinoma of the breast. The initial diagnosis in this case can be attributed to the patient's young age and lack of risk factors, as well as the rarity of desmoid tumors arising in the breast parenchyma.

References