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Case Report

Desmoid Tumor and Implant-Based Breast Reconstruction

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Desmoid tumor · Breast reconstruction · Breast implant · Breast imaging

Abstract
Desmoid tumors are rare locally aggressive myelodysplastic tumors that are usually abdominally based. They account for 0.2% of breast tumors. Certain factors like prior surgery, familial adenomatous polyposis, pregnancy, and high estrogen states are associated with chest wall desmoid tumor occurrence. We present a patient with a history of intraductal carcinoma of the left breast who underwent mastectomy with implant-based reconstruction who had a desmoid tumor of the breast detected during workup for cardiac transplantation for chemotherapy-induced heart failure. The tumor was originally thought to be recurrent breast cancer during workup with imaging obscured by the implant. Excisional biopsy demonstrated a desmoid tumor with a positive deep margin requiring rib resection, synthetic mesh, and pectoralis major flap reconstruction. Breast desmoid tumors are reactive malignancies that have been diagnosed after prior breast implant surgery but without an established risk associated with breast implants. Excision with microscopically negative margins and chest wall reconstruction when indicated is the current established treatment protocol; however, recent paradigm shifts include "watchful waiting" and medical management among treatment strategies.

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Introduction

Desmoid tumors are locally aggressive tumors that portend high rates of recurrence even with complete resection without known metastatic spread. They account for roughly 0.3% of all neoplasms and less than 3% of all soft tissue neoplasms [1]. Most desmoid tumors are noted to be abdominal or intrabdominal and have a highest rate of occurrence among patients with a history of familial adenomatous polyposis [2], although pregnancy associated with high estrogen states and antecedent trauma have also been shown to be predisposing factors for desmoid tumor occurrence [3].

Evidence suggests that dysplastic wound healing may play a role in the pathogenesis of desmoid tumors. Adenomatous polyposis coli (APC) gene and beta-catenin, which are both components of the Wnt signaling pathway, are thought to be involved in the molecular pathogenesis that leads to desmoid tumor formation [4]. APC is a regulatory inhibitor in the activation of beta-catenin. Mutation of either the APC protein or the beta-catenin protein can cause abnormalities in the Wnt signaling pathway, leading to accumulation of beta-catenin in the cytosol and translocation into the nucleus, promoting proliferation and enhanced cell survival [5]. The Wnt signaling pathway is important for homeostasis of multiple organ systems and has been shown to have an important role in wound healing, with elevated beta-catenin levels noted during the hyperplastic stage of wound healing and dysplastic beta-catenin activity pathogenic in hyperplastic scars [4, 5].

There is no established predictor for the occurrence of breast desmoid tumors, but there is thought that antecedent trauma or delayed wound healing portends a role in the pathogenesis. Breast desmoid tumors have been mistaken for primary or recurrent breast cancer both symptomatically and radiographically [6]. Diagnosis can be preempted by a mass that may or may not be tender to palpation, capsular contracture if the patient has an implant or skin dimpling, or it can be diagnosed upon routine imaging [6, 7]. While there are few cases of desmoid tumors arising in patients with breast implants, a recent review of the literature showed that persons with breast implants had a lower incidence of breast desmoid tumors than the general population [7]. As it stands, current FDA recommendations for silicone implant surveillance outside of reconstructive intervention includes imaging such as ultrasound or MRI at 5–6 years and subsequent imaging every 2–3 years after that to monitor for implant rupture. If there is any concerning pathology, imaging is recommended sooner [8].

We present a case of a young woman with a history of invasive ductal carcinoma who underwent mastectomy with silicone implant-based reconstruction and subsequently developed a desmoid tumor that was camouflaged by the implant and identified on preoperative imaging obtained for cardiac transplantation workup. Written informed consent was obtained from the patient for publication of this case report and all accompanying images.

Case Presentation

The patient is a 42-year-old female with a history of left breast invasive ductal carcinoma at age 37 that was estrogen receptor negative, progesterone receptive negative, and Her 2 nonamplified. There was no associated lobular carcinoma in situ and no lymphovascular invasion on initial excisional biopsy. She had neoadjuvant chemotherapy with four cycles of dose dense Adriamycin and Cytoxan followed by four cycles of dose dense Taxol. She had a left mastectomy with sentinel lymph node biopsy and subsequent left smooth silicone implant-based reconstruction and right mastopexy augmentation for symmetry. Her pathology showed one negative sentinel lymph node and stromal fibrosis with reactive changes consistent with neoadjuvant therapy effect on the mastectomy tissue.

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The patient developed orthopnea and dyspnea on exertion less than 1 year after completion of chemotherapy. She was found to have decompensated heart failure with an ejection fraction of 25% and diagnosed with NYHA functional class 2. She had an ICD placed 4 years after completion of chemotherapy with subsequent worsening heart function. Given her worsening heart failure, the patient was being evaluated for cardiac transplant and obtained a CT of the chest which demonstrated a 5.7 × 4.3 cm hypodense mass of the left chest wall and left axillary adenopathy (Fig. 1). On exam, she did not have any palpable masses.

A mammogram was subsequently obtained, confirming a mass in the upper inner quadrant at the 10 o'clock position 10 cm from the nipple. The image was classified as Bi-RADS 4. She then had a positron emission tomography scan which showed FDG avid anterior left chest wall mass extending into the breast implant without evidence of FDG avid nodal or distant metastatic disease. She was referred to plastic and reconstructive surgery for implant removal and excisional biopsy of the mass. In surgery, it was noted that the mass was firmly adherent to the chest wall, extended into the ribs, and was approximately 6 cm in diameter. It was excised at the base for biopsy (Fig. 2). Histologic examination demonstrated a proliferation of bland spindle cells with tapered nuclei and a single nucleolus set in a fibrous background. There was no cytologic atypia and mitotic activity was low. Immunohistochemical staining for beta-catenin demonstrated diffuse nuclear localization of the protein in keeping with a diagnosis of desmoid type fibromatosis (Fig. 3).

She had subsequent left anterior chest wall resection of the residual desmoid tumor including ribs 3 and 4 with final size of resected tissue measuring 8.2 × 6.1 cm. The defect was reconstructed with Gore-Tex and pectoralis major advancement flap. Pathology again demonstrated desmoid tumor (aggressive fibromatosis) and fat necrosis; margins were negative. Patient was doing well in follow-up and will be undergoing imaging surveillance to monitor for recurrence.

Conclusion

Breast desmoid tumors account for less than 0.2% of breast neoplasms [9]. The myelodysplastic tumor is locally destructive with high recurrence rates but without known metastatic spread. The pathogenesis of breast desmoid tumors remains unproven and speculated to arise from surgical trauma and genetic predisposition with somatic mutations in either beta-catenin or APC genes [10, 11]. According to the reported literature, breast desmoid tumors are associated with implants in 10% of the cases and tend to occur within 3 years after placement [7, 11, 12]. In a recent review of the literature by Tzur et al. [7], it was demonstrated that the incidence of breast desmoid tumors in patients with implant placement was not higher than the occurrence of breast desmoid tumors in the general population.

When breast desmoid tumors do occur, they can resemble new breast cancer or recurrence both symptomatically and radiographically. Mammography commonly demonstrates a spiculated, high-density mass without calcifications and an irregularly shaped, hypoechoic mass on ultrasound [6]. According to Neuman et al. [6], breast desmoid tumors were only visible with mammography in one-third of the cases whereas they were visible in all cases when sonography was used. MRI can be both diagnostic and useful for preoperative planning. Breast desmoid tumors are isointense masses on T1-weighted images with either lower or higher intensity lesions seen on T2-weighted images and can be distinguished from chest wall desmoid tumors [6, 13].

Treatment for desmoid tumors has largely been surgical excision with microscopically negative margins; however, there has been a recent shift in paradigm to watchful waiting with or without medical treatment and intra-arterial chemoembolization [13, 14]. Preservation of

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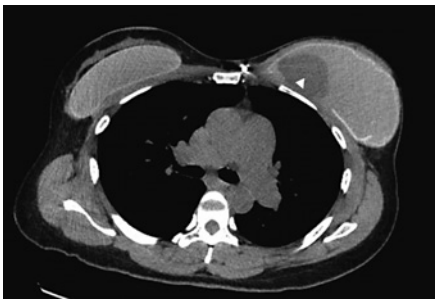

Fig. 1. CT scan of the chest revealing desmoid tumor originating from intercostal musculature and invading the central aspect of the left breast implant. The desmoid tumor is marked with a white arrowhead.

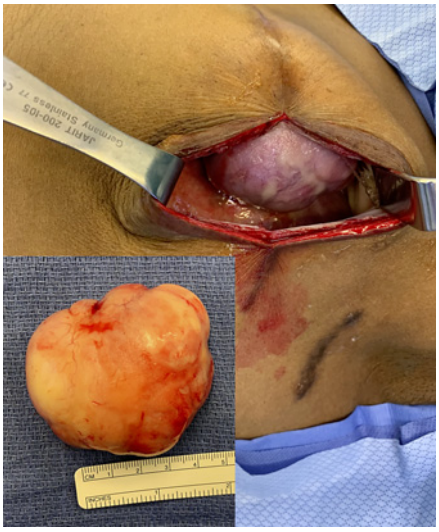

Fig. 2. Intraoperative photos of the desmoid tumor of the left chest. The tumor was smooth, fungating from the intercostal space and extending into the substance of the left breast implant. Excised mass is featured in the inset image next to a ruler to show its dimensions.

function has become a hallmark of desmoid tumor treatment, favoring careful surveillance in cases of desmoid tumors where functional loss after surgical excision may be a major contributor to morbidity [14]. Spontaneous regression has also been noted in patients with current studies evaluating how the immunologic environment of the host may contribute to regression [13, 14]. Radiotherapy (with or without surgery) and medical immunotherapy have also been advocated as both treatment or adjuvant therapies to help control the progression of desmoid tumors, particularly when surgery may cause significant morbidity or when microscopic or gross surgical margins are positive [14]. Our patient had positive margins after first excisional biopsy, but given she was pending cardiac transplantation secondary to chemotherapy-induced cardiac failure, a consensus was made to proceed with re-excision to obtain disease-free microscopic margins to prevent further delay. Histopathologic confirmation of desmoid tumors is mandatory before initiating therapy and should be established by core needle biopsies [6, 13, 14]. In our clinical case, the desmoid tumor invaded the breast implant preventing needle biopsy in favor of open surgical excisional biopsy. Desmoid tumors can be misdiagnosed in 30–40% of initial workups and presence of *CTNNB1* mutations was shown to be diagnostic on histology [14]. There have been multiple retrospective studies that have demonstrated progression-free survival rates of 50% at 5 years for asymptomatic patients and regression rates of 20–30%; however, this is limited by the initial size of the tumor and the site of occurrence, with chest wall portending higher risks of recurrence [14].

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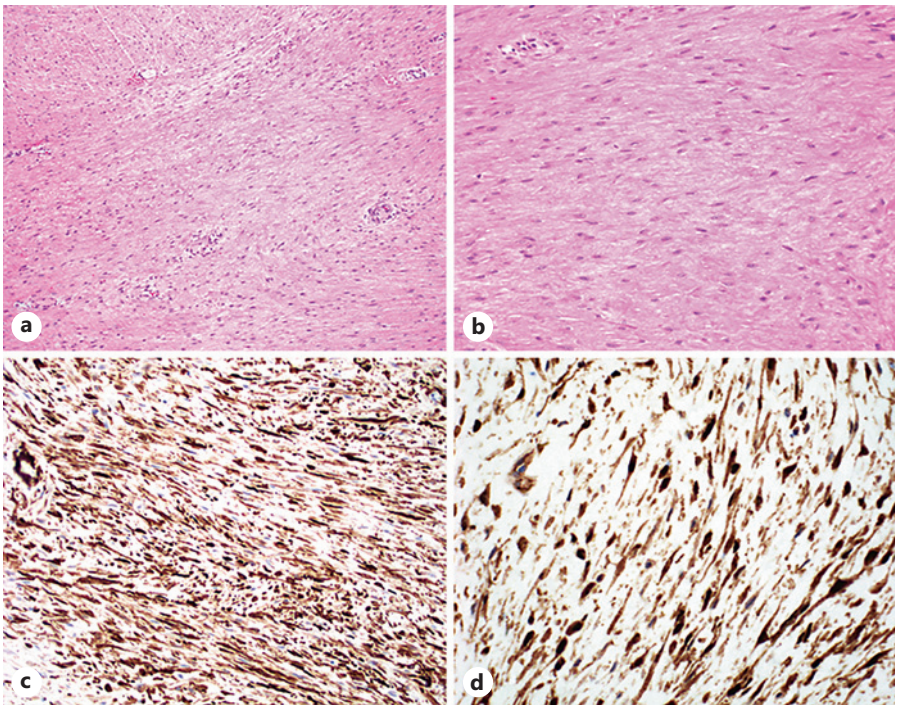

Fig. 3. Desmoid-type fibromatosis. H&E. ×100 (**a**) and H&E. ×200 (**b**): bland spindle cell proliferation composed of elongated nuclei with prominent nucleoli arranged in long sweeping fascicles. **c** Smooth muscle actin immunohistochemistry, ×200: shows diffuse cytoplasmic and membranous expression. **d** Beta-catenin immunohistochemistry, ×200: shows nuclear localization, characteristic of desmoid type fibromatosis.

Current FDA recommendations for surveillance after breast implant placement are to obtain ultrasound or MRI at 5-6 years and periodic imaging every 2-3 years afterward to monitor for implant rupture [8]. Women in the USA have a risk of 1 in 7 lifetime risk of breast cancer, with different agencies recommending initiation of screening mammography between the ages of 40–50 with yearly to biannual screening thereafter [15]. There has been no established etiologic risk outside of BIA-ALCL association with textured implants between cancer and breast implants, with some studies demonstrating that implants increase the palpability of breast tumors without worsening prognosis [15]. The Eklund technique has been used to displace the implant to visualize breast tissue; however, even with vigilant technique mammographic screening is impaired to some extent with implants [15].

Breast desmoid tumors remain a rare occurrence that are postulated to be associated with surgical trauma without an established predilection with the presence of implants. However, the question remains whether breast implants can mask desmoid tumors until they become large enough to be detected symptomatically or seen on routine imaging. Matrai et al. [11] demonstrated that breast desmoid tumors tend to occur 3 years after operation, with current FDA guidelines to begin screening for implant rupture at 5-6 years [8]. Regardless of screening recommendations, the presence of an implant may limit access for tissue diagnosis, requiring incisional or excisional biopsy. Our patient presented with a breast mass only diagnosed on workup for cardiac transplantation that was not palpable on physical exam and thought to represent recurrent breast cancer after tumor board analysis. She underwent excisional biopsy and removal of the implant with wide local excision after tissue confirmation. While rare, breast desmoid tumors remain a source of morbidity on the operated

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breast patient, and clear protocols are needed for screening to make sure the implants do not camouflage tumor occurrence and tissue diagnosis.

Statement of Ethics

This manuscript reflects the original work of the authors and this work has not been published or presented elsewhere. No off-label uses of any products were discussed in this manuscript. The current study qualified as quality improvement by the University of Virginia Institutional Review Board for Health Sciences Research and as a result was exempt from full review board approval. Written informed consent was obtained from the patient for publication of this case report and all accompanying images. This article does not contain any experimentation involving human or animal subjects.

Conflict of Interest Statement

While unrelated to this case report, the corresponding author, Chris A. Campbell, MD, has received grant support from Lifenet Health for basic science research, served on a medical advisory board for Integra Life Sciences, and received compensation for professional education services from Mentor, a subsidiary of Johnson & Johnson. None of the other authors have any conflicts of interest to declare.

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Author Contributions

Catherine Kilmartin, MD: performed the reconstruction and made substantial contributions to the conception and design of the work as well as acquisition and interpretation of data for the work. She drafted the work and gave final approval of the version to be published and agrees to be accountable for all aspects of the work ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. Clinton Westover, MD, and Shyam Raghavan, MD: performed the pathologic analysis and made substantial contributions to the conception and design of the work as well as acquisition and interpretation of data for the work. He drafted the work and gave final approval of the version to be published and agrees to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. Patrick M. Dillon, MD: made substantial contributions to the conception and design of the work as well as acquisition and interpretation of data for the work. He drafted the work and gave final approval of the version to be published and agrees to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. Chris A. Campbell, MD: performed the reconstruction reported here and made substantial contributions to the conception and design of the work as well as acquisition and interpretation of

data for the work. He drafted the work and gave final approval of the version to be published and agrees to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Data Availability Statement

This manuscript is a case report describing the clinical care of a single patient. As such, there is no dataset other than the patient's clinical data. All data that support the findings of this study are included in this article. The corresponding author may be contacted for any additional de-identified clinical information that the editors might require.

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