



Presentation and Diagnosis of Desmoid Tumours (Fibromatosis) in the Male Breast: A Case Report

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Abstract

Desmoid tumours, also referred to as fibromatosis, are benign masses found in the abdominal wall. It is extremely rare to find them within extra-abdominal sites and desmoid tumours account for less than 0.2% of all breast tumours. Further, it is unusual to find these locally aggressive tumours within the male population. This case report presents the case of a man who was diagnosed with a desmoid tumour involving the left breast.

Keywords

Male breast, Desmoid tumour, Fibromatosis

Introduction

Desmoid tumours, also known as fibromatosis, account for less than 0.2% of all breast tumours [1]. These tumours typically occur in the abdominal wall and are rarely found within extra-abdominal sites such as the breast. They are characterized by the proliferation of monoclonal myofibroblastic neoplasms [2]. Further, they are more prevalent in females than males and thus, it is extremely uncommon to encounter these tumours within the male population [3]. Desmoid tumours are known to lack metastatic potential; however, they are often locally aggressive with a tendency to recur [3]. Breast desmoid tumours generally mimic breast carcinomas in clinical and radiologic presentation and diagnosis is confirmed based on histological findings [1,3-5]. Primary therapy for desmoid tumours consists of surgical resection, and the role of adjuvant therapies including radiation remains unclear [3].

The purpose of this case report is to describe and discuss the clinical, imaging and histological features of this rare type of locally aggressive and recurrent benign tumour in a male patient.

Case Presentation

A 52-year-old male with no family history of breast cancer noticed a left breast mass. He continued to mon-

itor the lesion, which remained unchanged, and had an ultrasound followed by a biopsy four months later. The biopsy was suggestive of a desmoid tumour.

The patient underwent a partial resection of his left breast. Pathology results indicated vimentin-positive spindle cell proliferation primarily of fibroblasts with minimal mitotic activity, with no breast tissue in the sample, compatible with fibromatosis. Morphology and immunohistochemical staining profile were also compatible with fibromatosis. Despite no significant atypia or malignancy, there was a focally deep margin. Consequently, the patient underwent a second wider excision a month later, to remove the residual disease and prevent recurrence. The pathology after the second surgery showed positive anterior and medial margins. His

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subsequent follow-up appointment 5 months later suggested that the tumour had recurred in the area of the residual disease. MRI imaging found a low-signal mass measuring 7.5×7.0 cm in the left chest wall infiltrating the pectoralis major muscle, skin, and left nipple with ill-defined margins.

As a result of a large amount of disease, it was recommended that the patient have radiotherapy followed by a wide excision of the area to attain widely clear margins. Six months after the second surgery, the patient had pre-operative radiotherapy to assist with local control before the scheduled third surgery. Post radiotherapy MRI of the left chest wall 2 months later revealed an ill-defined, heterogeneous, low T1/low T2 superficial soft tissue mass infiltrating the pectoralis major muscle - the mass was reduced compared to the previous MRI, measuring $6.9 \times 4.9 \times 1.6$ cm. He then had a third surgery one month after this MRI, which included a wide surgical resection and a flap reconstruction of the area. Pathology revealed multiple small foci of residual fibromatosis with all resection margins free of tumor.

Five years after his diagnosis of desmoid tumour, the patient was diagnosed with low-risk prostate cancer. A biopsy revealed Gleason 6 adenocarcinoma of the prostate. The patient was scheduled to undergo a robotic-assisted laparoscopic radical prostatectomy five years after this diagnosis. At his last follow up appointment, approximately 10 years after his initial breast desmoid tumour diagnosis the patient remained free of disease from this perspective.

Discussion

Desmoid tumours are rare lesions when found in extra-abdominal sites such as the breast [6]. These tumours are known to be locally aggressive and infiltrative without any potential for distant metastasis [3]. Desmoid tumours in the breast are found to be more common in females than in males, and some studies have suggested a hormonal or genetic influence [3,6]. The etiology of these masses is still widely debated and remains largely unknown.

We have identified nine published case studies or case series of desmoid tumours or fibromatosis of the breast [1,3-10]. Like other cases in the literature, our patient presented with a palpable mass in the left breast [7]. The patient underwent imaging with ultrasound, as well as a biopsy of the mass in order to rule out breast carcinoma. Diagnosis of desmoid tumours can be a challenge. Histopathology often reveals mature fibroblast without nuclear atypia or increased mitotic activity [11]. Diagnosis by fine needle aspiration or core needle biopsy is difficult, as histological findings can be bland and may be interpreted as inconclusive or benign [3,8]. Further, desmoid

tumours can infiltrate the pectoralis muscle and cause nipple retraction, mimicking the presentation of breast cancer [11]. In our patient, pathological reporting after the biopsy and surgical resection confirmed fibromatosis with no breast tissue in the sample.

Desmoid tumours are known to recur locally if not removed entirely. If the desmoid tumours originate from the breast tissue, the risk of recurrence when the resection margins are positive is 21% [9]. On the other hand, if they arise from the musculoaponeurotic structures of the underlying pectoralis major muscle, the risk of recurrence is 57% [9]. These masses generally have poorly defined margins, posing challenges for surgical resection; however, the primary treatment of these tumours remains surgery with negative margins [3,8]. Radiation therapy has been employed to improve local control rates and has been shown to improve outcome [12]. Our patient initially underwent two resections, and then pre-operative radiotherapy followed by a final resection with flap reconstruction in order to treat his locally recurring ill-defined desmoid tumour.

Conclusions

Desmoid tumours of the breast are a rare finding within the male population [3]. As a result, very little is known about their etiology. Unfortunately, the presentation of these tumours in terms of both radiological findings from mammograms and histological findings from biopsies mimics breast carcinomas and can lead to misdiagnoses and inappropriate treatment [3,10]. As a result, it is important for health care providers to be aware of this condition and include it within the differential diagnoses of breast masses in male patients. Accurate differential diagnosis can help avoid unnecessary treatment, since pharmacological therapies have not been found to be effective for this tumour type [3]. Further, the rarity of desmoid tumours has resulted in a lack of definitive evaluation of diagnostic and treatment options. Moving forward, collaborative multi-centre studies should be performed in order to obtain large enough patient numbers to understand the etiology and treatment options other than surgical resection for male patients presenting with desmoid tumours.

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