



Desmoid fibromatosis of the breast, a case report of an uncommon breast lesion

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Abstract

Introduction: Desmoid-type fibromatosis is a rare soft tissue tumor. The incidence is approximately 0.2% of all neoplasms of the breast. Surgical excision with wide margins remains a valuable therapeutic option.

Case report: A 33-years-old female with left breast mass, mammogram and ultrasound showed a 0.9 cm palpable lesion with architectural distortion and a vacuum assisted biopsy showed infiltrating bland looking spindle cells proliferation in long sweeping fascicles that are partially positive for beta-catenin immunostaining, suggestive of desmoid fibromatosis.

Wide local excision was performed. Histopathological examination of the specimen confirmed the diagnosis of desmoid type fibromatosis of the breast.

Discussion: Fibromatosis is a rare tumor with locally aggressive behavior and high incidence of local recurrence. The reported risk factors include surgical trauma, silicone implants in addition to its association with Gardener's syndrome.

Conclusion: Fibromatosis represents a diagnostic and therapeutic challenge due to its infiltrative nature and propensity for recurrence. A multidisciplinary approach involving oncologists, surgeons, and radiologists is crucial for optimal management.

Keywords: Breast surgery, oncology.

Introduction

Desmoid-type fibromatosis, is a rare soft tissue tumor with the typical clinical behavior of frequent local recurrence without distant spread [2]. The main locations include the mesentery, the abdominal wall, and the extremities [3]. The incidence is approximately 0.2% of all neoplasms of the breast [2,4]. The main risk factors are trauma and surgery, as well as being an association with Gardener's syndrome [5,6]. Some have suggested that this entity arises from within the breast parenchyma itself, while others have suggested that it arises from the aponeurosis overlying the pectoralis major muscle [4,9]. According to the

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Keywords: Breast; Desmoid, Spindle cell; Tumor; Extra abdominal.

current consensus from existing research, e.g., of the Desmoid Tumor Working Group and ESMO, active primary surgical therapy is no longer the method of choice for asymptomatic patients [4,7,8], however surgical excision with wide margins remains a valuable therapeutic option, especially in symptomatic and progressive desmoid type fibromatosis or impaired quality of life to avoid recurrence of this locally aggressive tumor [3].

The current report aims to report a rare case of desmoid type fibromatosis of the breast. The report has been arranged in line with SCARE guidelines with a brief literature review [1].

Patient case presentation

Patient information

A 33-year-old Saudi female, married, not known to have chronic medical illness, was referred in January 2024 to breast oncoplastic clinic in King Saud Medical City (NGHA) in Jeddah, KSA, for a painless left breast lump that is increasing in size over 2 months, without skin or nipple changes or discharge.

Patient denied any history of trauma, family history of breast cancer, previous breast surgery.

Clinical findings

Bilateral asymmetrical breasts, the right breast was bigger, Size D cup, grade 2 ptosis, Left breast: unifocal around 1 x 1 cm palpable non tender mass at 3 o'clock, firm, not attached to the skin or chest wall.

Diagnostic assessment and interpretation

Left breast and axilla ultrasound and Mammogram showed:

Left breast palpable lesion with architectural distortion in the upper lateral mid/anterior third at 3:00, It measures 0.9 cm and seen with no significant vascularity. A clip was placed.

Left breast, vacuum assisted biopsy of the indexed lesion showed:

Infiltrating bland looking spindle cells proliferation in long sweeping fascicles that are partially positive for beta-catenin immunostaining, suggestive of desmoid fibromatosis.

Immunohistochemistry study: The spindle cells are partially positive for Beta-catenin (nuclear staining), focal positive for SMA, while negative for PAN CK, CK5/6, P63, CD34.

Intervention

Once a diagnosis was established, surgery was scheduled. The patient was taken to the operating room for Left breast conserving surgery.

Which performed under general anesthesia with selective intubation intraoperative mammogram was done after the resection confirming the clip at the center of the specimen.

The left wide local excision site was then closed in a standard manner. No attempts at cosmetic breast reconstruction with autologous tissue transfer or expander/implant placement were considered at that time.

The surgically removed specimen was weighing 44.4 grams and measuring 5.5 x 4.9 x 2.8 cm, with grossly tan-white, firm mass with irregular border, The specimen was sent to histopathology department in formalin.

Follow up and outcome

Histological evaluation of the surgical specimens revealed desmoid type fibromatosis with tumor size 1.5 cm in greatest dimension, tumor extent, infiltration into the surrounding adipose tissue with entrapped benign mammary ducts, margin status.

All negative for tumor and distance from closest margin: 4 mm from inferior margin.

The post-operative course was uneventful, and the patient was discharged home at the same day, seen in the clinic week after, wound as it was well healed.

Case was discussed in the tumor board; decision was toward no further treatment is required

CT CAP done as a part of the work up, findings on the intra-thoracic and intra-abdominal organs were physiological.

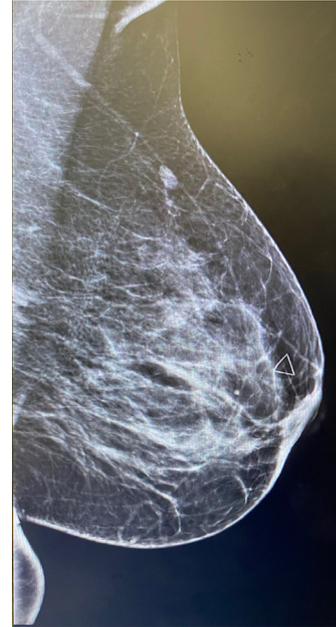


Figure 1: Left mammographic views ML of the left breast showing an irregular, spiculated, high-density mass in the upper outer quadrant. The lesion causes architectural distortion without associated calcifications. Findings are consistent with desmoid-type fibromatosis confirmed on histology.

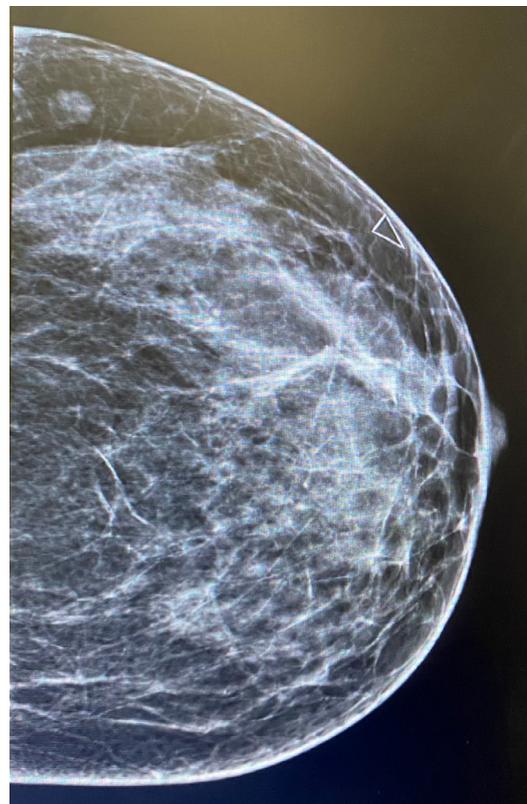


Figure 2: Left mammographic views CC of the left breast showing an irregular, spiculated, high-density mass in the upper outer quadrant. The lesion causes architectural distortion without associated calcifications. Findings are consistent with desmoid-type fibromatosis confirmed on histology.

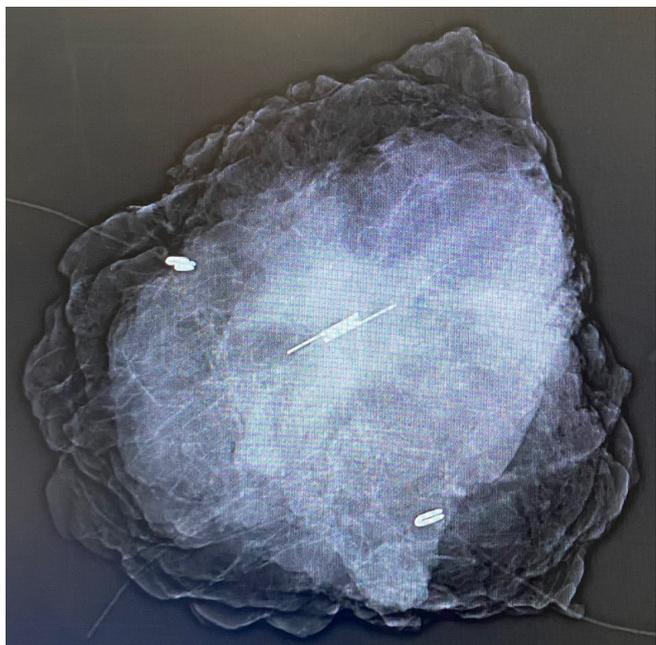


Figure 3: Intra operative mammogram for the left breast excision Specimen showing the SAVI reflector in the middle of the tissue.

Discussion

World Health Organization (WHO) defined desmoid type fibromatosis as an intermediate soft tissue tumor with the characteristic of clonal fibroblastic proliferation derived in the deep soft tissue with the ability of local infiltration [2]. Fibromatosis is a rare tumor with locally aggressive behavior and high incidence of local recurrence. The reported risk factors include surgical trauma, silicone implants in addition to its association with Gardner's syndrome [6]. Clinically, this tumor manifests as a broad spectrum and in most cases, desmoid type fibromatosis of the breast is described as a suspicious, mobile, firm, and painless nodule [5].

Mammography presents irregular walled and highly dense lesion with no calcifications mimicking sometimes breast carcinoma [6].

Radiologic evaluation of our case revealed soft tissue mass inducing architectural distortion.

Histopathological examination is crucial for diagnosis, and often revealing spindle cells with minimal cytological atypia and abundant collagenous matrix [4]. A diagnosis can be made from microscopic findings on routine hematoxylin and eosin-stained sections. Furthermore, immunohistochemical staining for β -catenin with nuclear positivity is also useful in establishing a diagnosis, but there are no specific immunomarkers for breast fibromatosis [2].

In conclusion, fibromatosis represents a diagnostic and therapeutic challenge due to its infiltrative nature and propensity for recurrence. A multidisciplinary approach involving oncologists, surgeons, and radiologists is crucial for optimal management. Further research is needed to elucidate the molecular pathways driving fibromatosis and to develop targeted therapies that can improve outcomes while minimizing morbidity.

Conclusion

Due to the rare involvement of the breast in patients with desmoid-like fibromatosis, the present study reports this case with its clinical feature and histological finding to improve and add to our knowledge of the disease.

Declarations

Human subjects: Patient informed consent was obtained.

Conflicts of interest: In compliance with the ICMJE uniform disclosure form, all authors declare the following: Payment/services info: All authors have declared that no financial support was received from any organization for the submitted work.

Financial relationships: All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work.

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