

Desmoid type Fibromatosis in Breast: A rare case report

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Abstract: Fibromatosis (desmoid type) is a locally infiltrative lesion without metastatic potential that arises from fibroblasts or myofibroblasts. It can occur within the breast parenchyma, but frequently arises from the pectoral fascia and extends into the breast.

Case History: A 59-year-old female patient presented with complaints of painless, hard lump in the upper outer quadrant of the left breast. On examination, the swelling was single, firm to hard, non-mobile measuring 6x4cm with skin dimpling. Radiological findings were suspicious of BIRADS 4B.

Conclusion: We reported this case as Spindle cell tumour of benign aetiology on histopathology, which was reported as Desmoid type fibromatosis after confirmation with IHC.

Index Terms: Breast fibromatosis, Desmoid tumor, Spindle cell tumors, Immunohistochemistry

INTRODUCTION

Desmoid type Fibromatosis is rare benign, infiltrative and locally aggressive lesion, which is also referred to as desmoid tumor, low-grade fibrosarcoma, or aggressive fibromatosis. This tumor doesn't metastasize and arises from fibroblasts or myofibroblasts. [1,2]

Desmoid fibromatosis mostly occurs within the mesentery, the abdominal wall, and the extremities, extra-abdominal fibromatosis occurs in the breast parenchyma, but frequently arises from the pectoral fascia and extends into the breast.[3] The differential diagnosis on histopathology are scar formation, fibrosarcoma, Spindle cell carcinoma fibromatosis-like variant, Lipomatous myofibroblastoma, and Nodular fasciitis.[4]

CASE REPORT

A 59-year-old female patient presented with complaints of rapidly growing, painless, hard lump in the upper outer quadrant of the left breast for 15 days. On examination, the swelling was single, firm to hard, mobile measuring 6x4cm with skin dimpling. On breast ultrasound, a well-circumscribed hypoechoic area with a homogenous echotexture with irregular and lobulated margin in the upper outer quadrant of left breast was noted. Micro-calcifications noted within the lesion. BIRADS 4B (moderate suspicion of malignancy) was reported on USG. A core needle biopsy was performed on the palpable left breast mass, which was inconclusive of any opinion, hence lumpectomy was done and the specimen was received in histopathology section.

Gross examination: The specimen was 7x6.5x3cm in size with an elliptical flap of skin measuring 4.1x1.5cm, well circumscribed firm tumour with off-white, whorled pattern, without necrosis (Fig. 1,2). The specimen was fixed in 10% neutral buffered formalin for 24 hr, appropriate sections were taken and processed in a tissue-processing machine. The processing included dehydration, clearing and paraffin embedding. The paraffin blocks were cut with rotary microtome to produce 3-5 µm sections. The sections were then stained with hematoxylin and eosin.



Figure 1: Lumpectomy specimen with elliptical skin flap and sutures for orientation received for histopathology.



Figure 2: Cut section showing a well circumscribed, grey white tumour with whorled appearance.

Microscopic examination: Histological evaluation of the surgical specimen revealed well circumscribed and partially encapsulated tumor composed of predominantly spindle cells arranged in short intersecting fascicles and storiform pattern (Figure 3a). Few stellate cells were also seen. Stroma was variable showing fibrocollagenous tissue, thick and hyalinised collagen bundles along with abundant myxoid matrix (Figure 3b). Tumor cells were noted to be infiltrating the breast parenchyma and fat. The periphery of tumor showed dense lymphocytic infiltration along with few plasma cells. No necrosis or mitosis identified (Figure 4a-c).

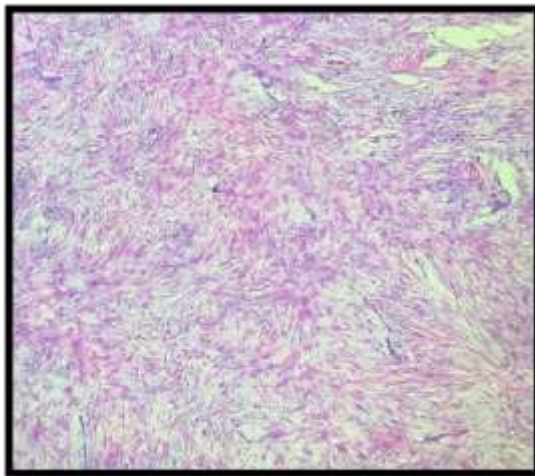


Figure 3a: H and E section showing spindle cells in fascicles and storiform pattern along with myxoid stroma (10x)

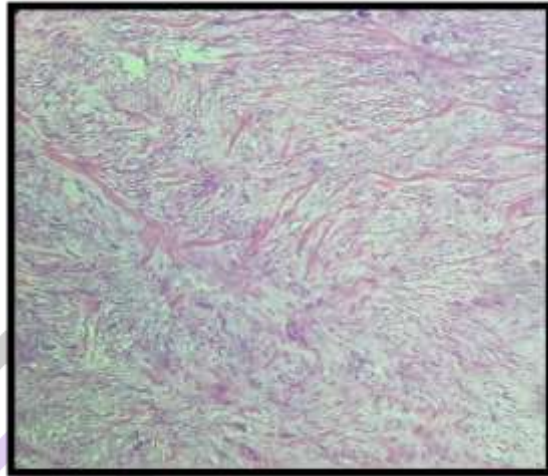


Figure 3b: H and E section showing (B) hyalinised 'ropy' collagen. (10x)

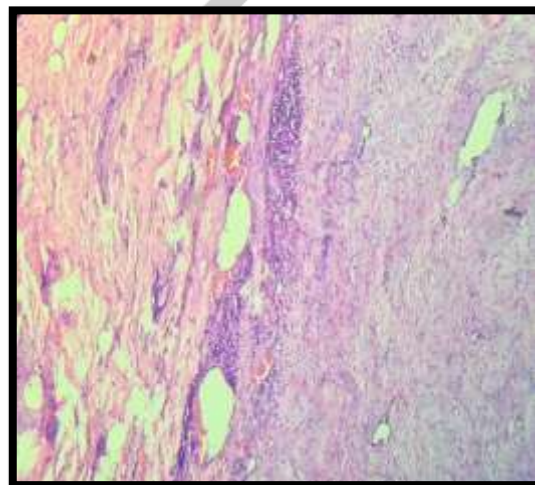


Figure 4a: H & E section from tumour showing a peripheral collar of lymphocytes. (10x)

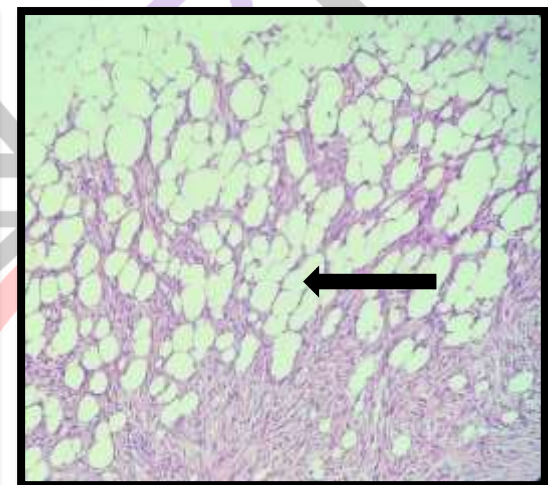


Figure 4b: H & E section showing infiltration of tumour cells into the surrounding fat. (Arrow Head) (10X)

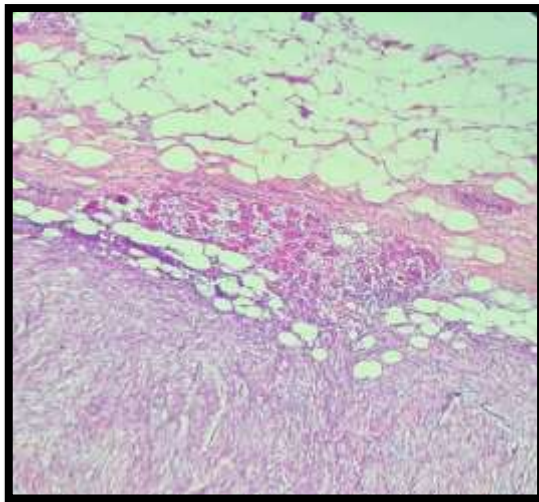


Figure 4c: H & E section showing compressed ducts of Breast at one focus. (10x)

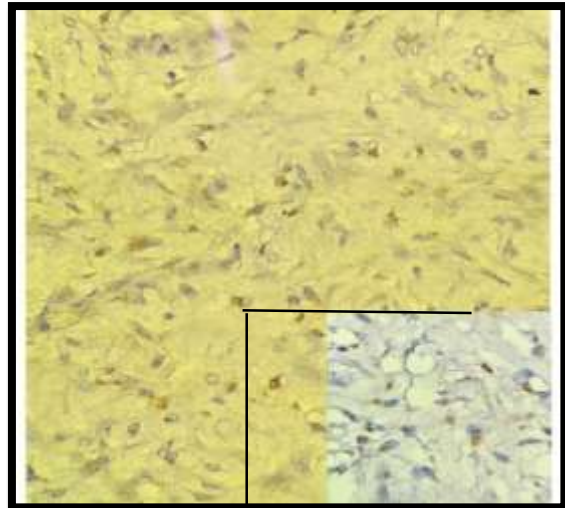


Figure 5a: IHC slide showing Focal granular nuclear positivity with Beta- catenin. (40x)

On Immunohistochemical studies, the proto-oncoprotein marker **β -catenin** showed focal granular nuclear positivity. **Vimentin** and **SMA** showed cytoplasmic positivity (Figure 5a-c). Tumour cells were negative for **S-100**, **Desmin** and **Cytokeratin**.

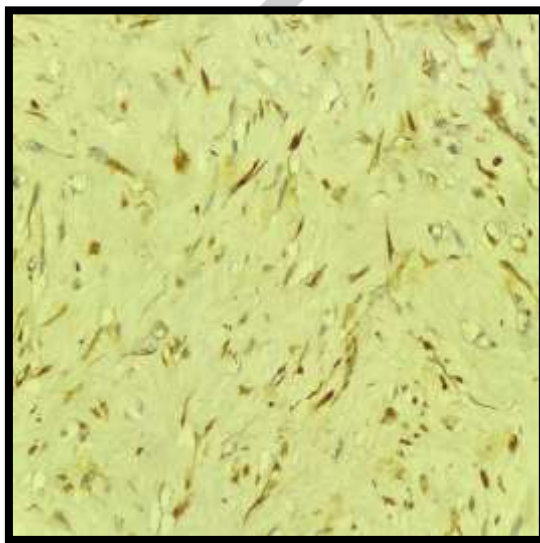


Figure 5b: IHC slides showing Cytoplasmic positivity for Vimentin. (40x)

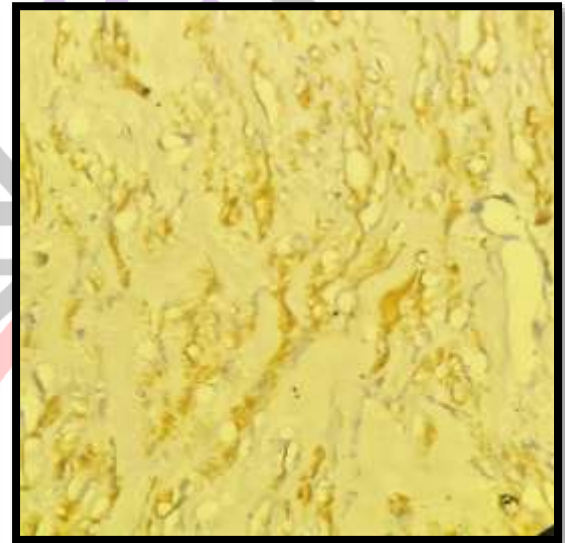


Figure 5c: IHC slides showing Cytoplasmic positivity for SMA. (40x)

DISCUSSION

Desmoid tumors, also known as deep fibromatoses, of the breast are comprising fewer than 0.2% of all breast tumors.[5] The World Health Organization defined desmoid-type fibromatosis as an intermediate soft-tissue tumor and is characterized by clonal fibroblastic proliferation arising in the deep soft tissues. The first case of breast fibromatosis was reported in 1923 and is the rare site for extra-abdominal fibromatosis. It occurs at a wide range of ages and is much more common in females. The etiology is not well understood although few cases of breast fibromatosis have been associated with Gardner's syndrome.[6] There is an association with previous trauma, particularly surgery, including implants.

Desmoid tumors can be divided as abdominal, extra abdominal, and intra-abdominal depending upon their anatomic location. Desmoid tumors in the breast may present with a several months history of a palpable nontender breast mass with breast carcinoma-like symptoms, such as skin infiltration and dimpling, as well as nipple retraction.

Breast imaging are not specific for fibromatosis and mimic breast carcinoma. Breast desmoid tumors are a diagnostic and therapeutic challenges as they clinically and radiologically mimic carcinoma.[7]

In Desmoid type fibromatosis of the breast, an active surveillance approach showed progression-free courses or remissions in up to 88% of patients. [8,9] Definitive diagnosis is made by diagnostic surgical biopsy.

In diagnosing tumors of mesenchymal origin, large-core needle biopsies are not always successful.[10] Hence for therapeutic planning and follow-up results, a histopathological diagnosis is of great importance.[11] On gross examination, the appearance of specimen varies from being well-circumscribed nodular lesions to irregular infiltrative lesions. On microscopic examination, the diagnostic feature of a desmoid type fibromatosis is a non-encapsulated bland-looking spindle cells that are organized into long sweeping and intersecting fascicles. Also noted are finger-like extensions at the periphery of the lesion into adjacent breast parenchyma and adipose tissue.[12] The overall cellularity is low to moderate with no cytologic pleomorphism or increase in mitotic activity, which is important to distinguish it from metaplastic fibrosarcoma. Immunohistochemistry studies show positivity for actin and vimentin hence is helpful for the diagnosis of desmoid type fibromatosis. Rare positivity of Desmin, whereas S100 and CD34 usually negative is seen. β -catenin nuclear staining is also considered for diagnosis, but it may be only focally positive. However, Cytokeratin staining is done for ruling out a carcinoma. The histological evaluation along with immunohistochemistry, with evaluations of imaging findings and the clinical course can help in accurate diagnosis of such cases. [4,13]

Surgical therapy with wide local excision with clear margins is a valuable therapeutic option, especially in symptomatic and progressive desmoid type fibromatosis. Recurrence is less likely if a wide excision is performed and resection margins are disease-free. Positive excision margins and intralesional excisions are associated with a greater rate of recurrence. Younger age and larger tumor size are also associated with an increased risk of recurrence [14]

CONCLUSION

Desmoid like fibromatosis is a rare breast neoplasm. Breast fibromatosis does possess the potential for aggressive local behaviour, although being classified as an intermediate soft tissue tumor. Breast imaging examinations aren't specific for fibromatosis and sometimes imitate carcinoma. The tumor is best differentiated histologically. With the arrival of more accurate imaging methods, alongside proper histopathologic interpretation and therefore the judicious use of ancillary methods like immunohistochemistry, most of the entities making up this spindle cell lesion are often identified with certainty, facilitating treatment planning.

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