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Desmoid Fibromatosis Of Breast, Clinically And Radiologically Mimicking Carcinoma Breast

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Abstract

Aim: To describe the case of desmoid fibromatosis clinically and radiologically mimicking carcinoma breast.

Background: Desmoid fibromatosis is a locally aggressive but non metastasizing neoplasm that rarely occurs in the breast.Histopathology shows uniform-looking, slender, spindle shaped cells form interwoven, arcuate fascicles that characteristically infiltrate adjacent tissues,such as mammaryducts/lobules,adipose tissue, and skeletal muscle. Morphological features do not always distinguish desmoid fibromatosis from phyllodes tumour or metaplastic carcinoma, which must be excluded with molecular and/or immunohistochemical studies.

Case Vignette: A 61-year-old female patient presented with right breast mass for 1 month. On examination, a hard, irregular mass measuring 2 X2 cm seen in right breast. Mammogram revealed :BIRADS 5lesion.

Treatment: Wide local excision of right breast along with sentinel lymph node biopsy was done.

Conclusion:

Desmoid fibromatosis of the breast is an uncommon entity that can closely mimic carcinoma clinically and radiologically. Histopathological and immunohistochemical evaluation is essential for accurate diagnosis. Awareness of this condition can help prevent misdiagnosis and unnecessary aggressive treatment. A multidisciplinary approach and close follow-up are recommended due to the risk of local recurrence.

In our case, Histopathology showed uniform bland spindle cells in woven fascicles, the neoplasm was infiltrating skeletal muscle bundles and adipose tissue. Our first differential was desmoid fibromatosis, but we have to rule out fibromatosis like metaplastic carcinoma, phyllodes tumour, myofibroblastoma. Special stain with mason trichrome :the tumour cell cytoplasm stained red in colour indicating myofibroblastic origin. IHC for cytokeratin was negative, ruling out metaplastic carcinoma. IHC for cd34 was negative ruling out phyllodes and myofibroblastoma and thus diagnosis of desmoid fibromatosis was made.

Keywords: Desmoid fibromatosis, breast , locally aggressive, IHC

Introduction

Desmoid fibromatosis is an infiltrative spindle cell neoplasm characterized by fibroblastic/myofibroblastic differentiation and activation of the WNT/B-catenin pathway.1 It is a locally aggressive but non metastasizing neoplasm commonly encountered in the abdominal wall and extra-abdominal sites but it rarely occurs in the breast2.

Desmoid fibromatosis accounts for < 0.2% of all breast tumours.1. The median patient age at presentation is in the third to fifth decades of life, but tumours can present at any age.1

Pathogenesis:CTNNB1 activating mutations have been detected in 83-95% of sporadic desmoid fibromatosis cases . These mutations have a strong predilection for exon3 of beta catenin.1

Breast imaging techniques aren't specific for desmoid type fibromatosis, it is difficult to distinguish fibromatosis with breast malignancy through imaging.3Histopathology shows uniform-looking, slender, spindle shaped cells form interwoven, arcuate fascicles of varying cellularity that characteristically infiltrate adjacent tissues, such as mammary ducts/lobules, adipose tissue, and skeletal muscle.1

Desmoid has significant risk for local recurrence rate even with adequate surgical resection but no metastatic potential .4Standard treatment involves a wide surgical resection with safe margins . The reasons for such an aggressive surgical approach include the potential for the fibromatosis to undergo aggressive, local growth and for invasion into the surrounding structures, plus a high local recurrence rate when incompletely excised with positive surgical margins.5

Case Report

A 61-year-old female patient presented with right breast painless mass for 1 month. She is a known case of hypertension, hypercholesterolemia and diabetes mellitus. On examination The patient had right breast hard, irregular mass measuring 2 X2 cm. Mammogram report: well hypoechoic lesion with irregular margins in lower inner quadrant of right breast measuring 21x11x15mm.impression given was irregular focal lesion in lower inner quadrant of right breast: BIRADS 5.

Treatment: Wide local excision of right breast along with sentinel lymph node biopsy was done.

Gross Specimen: Received WLE specimen measuring 6x5.5x2 cm. cut section showed an irregular grey white to grey brown lesion measuring 2.2x2.2x1.5cm which was close to the anterior skin margin.All other margins were free of neoplasm.

FIG1: GROSS IMAGE(irregular grey white to grey brown lesion)

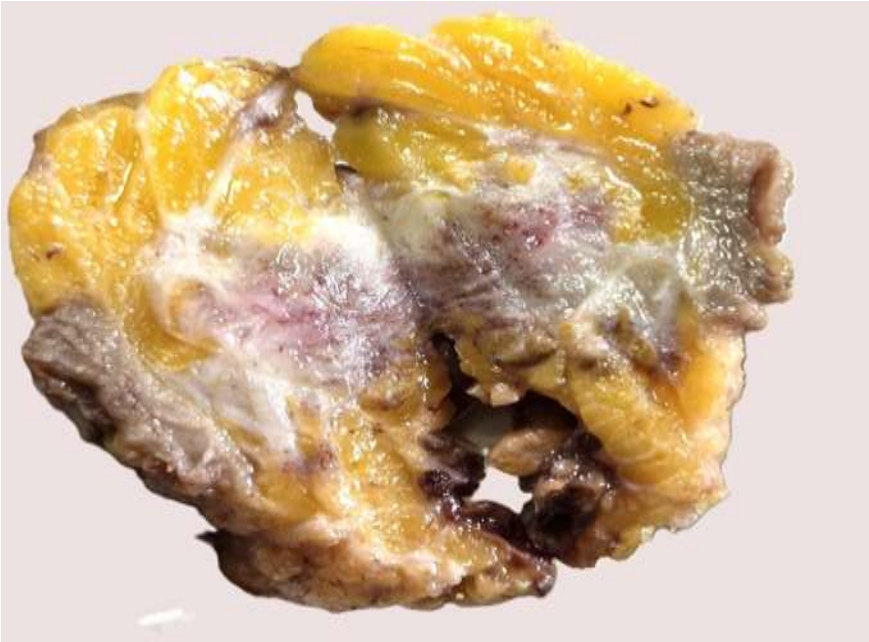

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FIG 2:Microscopy- Uniform bland spindle cells arranged in woven fascicles and bundles.

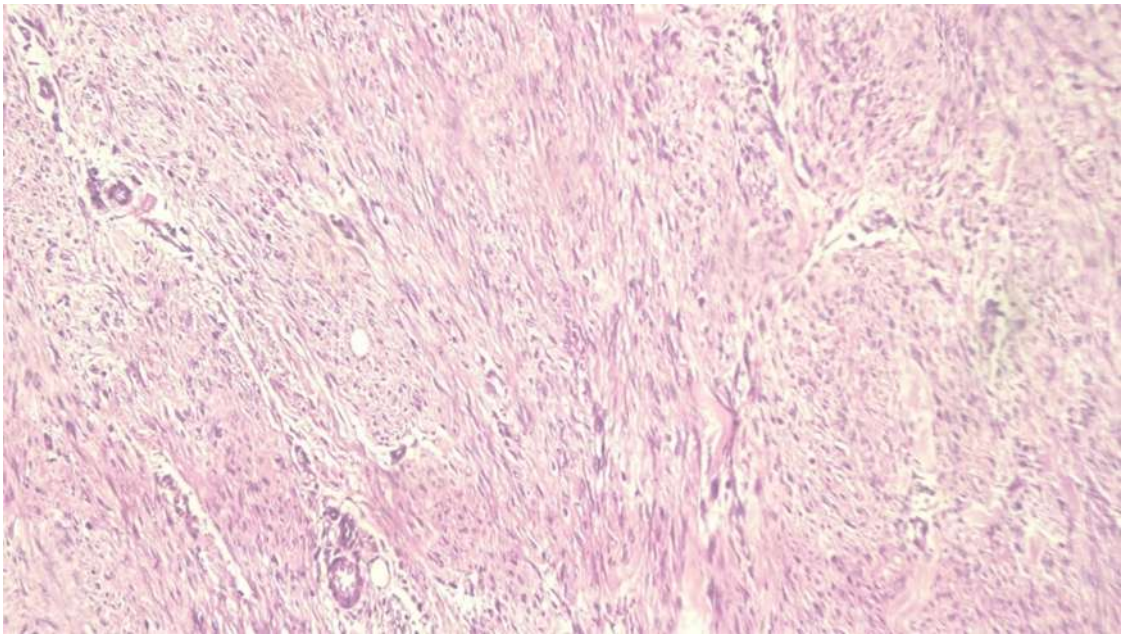

FIG 3:Higher power view showing cells with indistinct cell membrane spindle shaped bland nuclei.

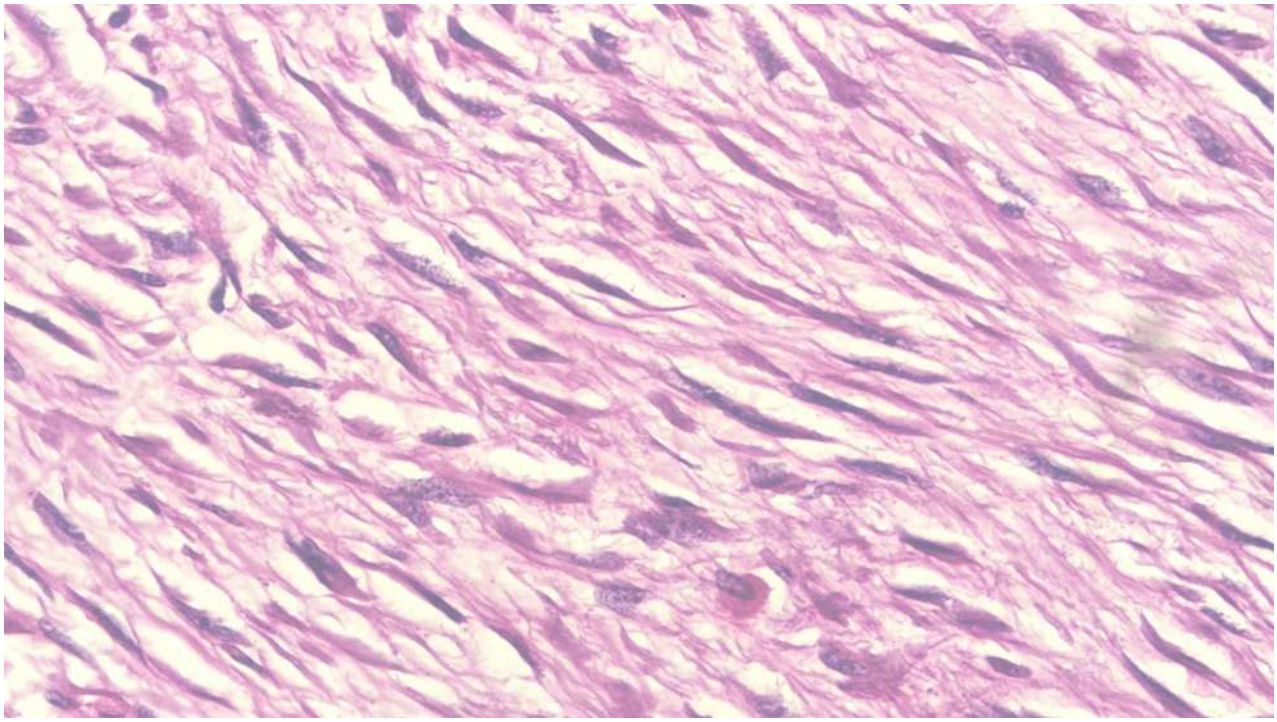

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FIG 4: The neoplasm is seen infiltrating the skeletal muscle bundles and adipose tissue.

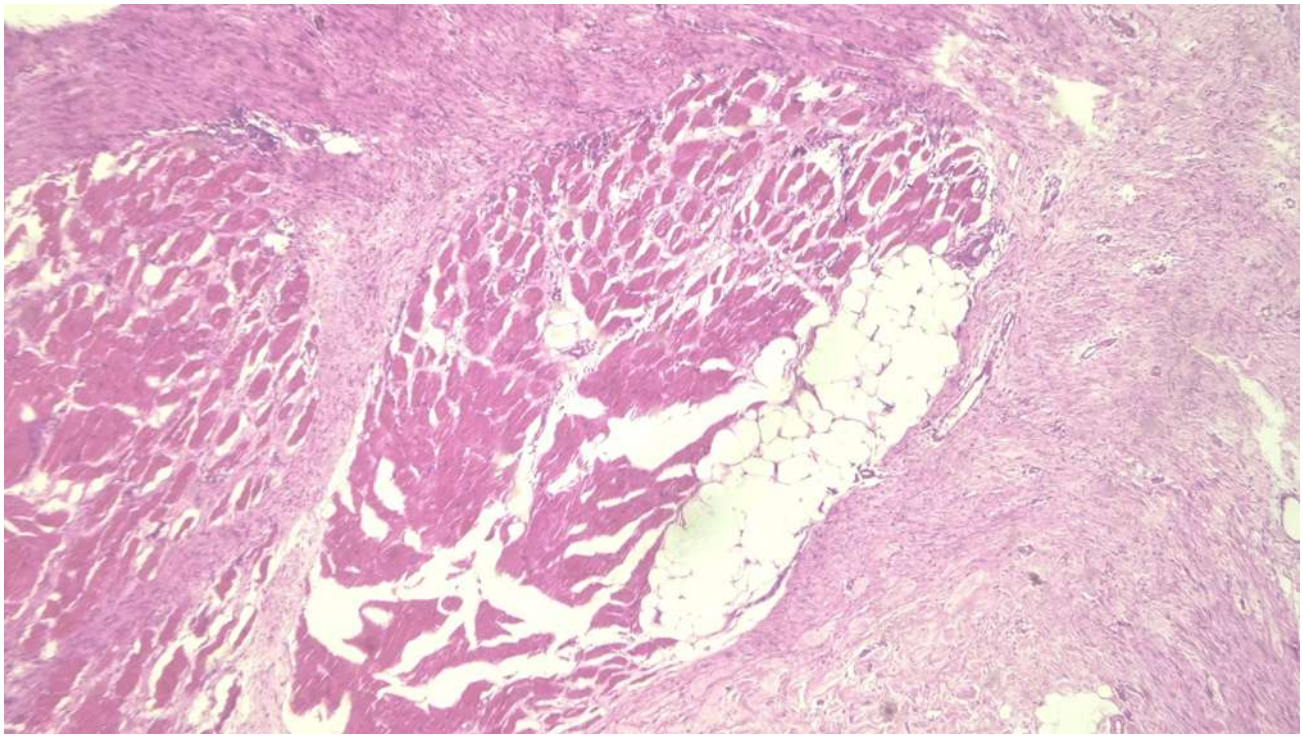

FIG 5:CD34 -negative (tumour cells negative for CD34,blood vessel acting as internal control.)

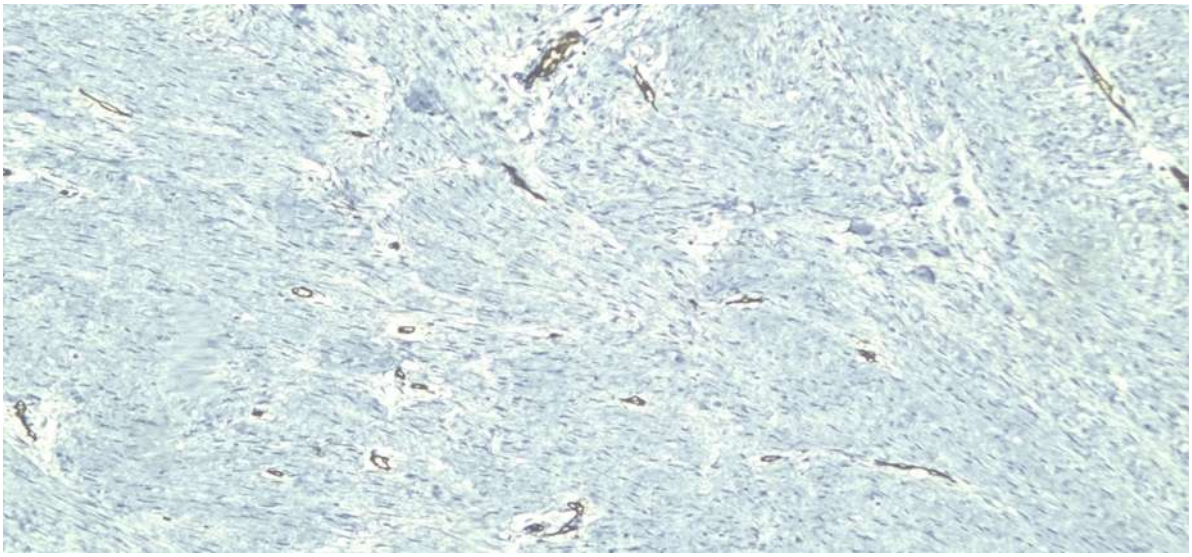

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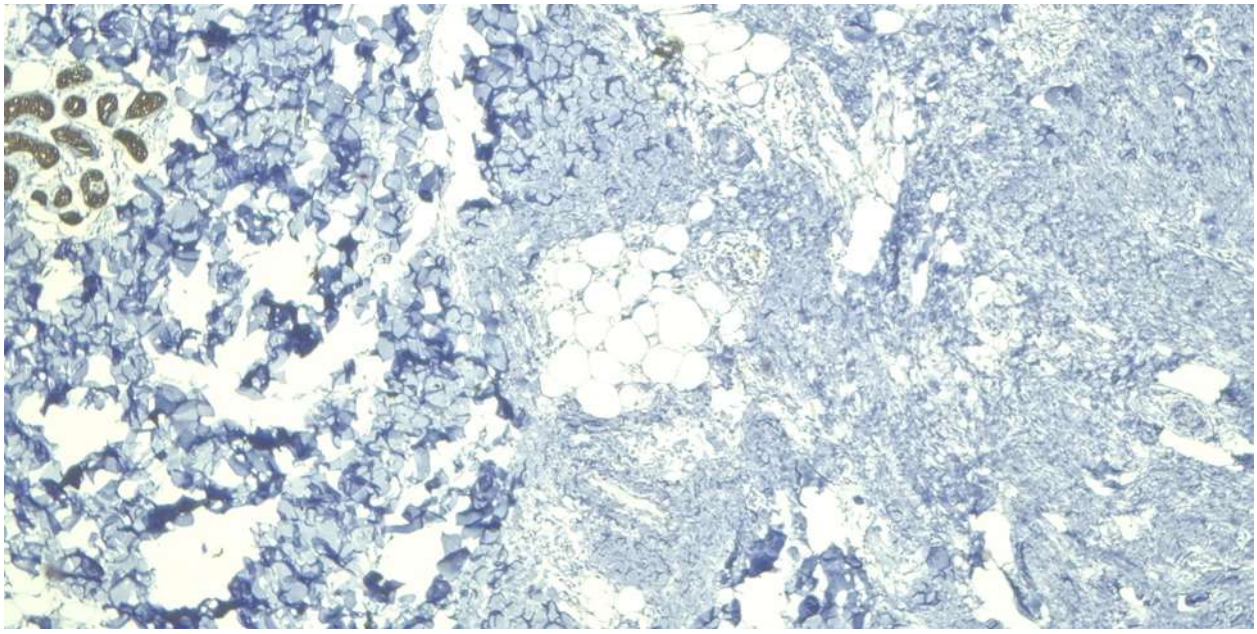
FIG 6:Cytokeratin negative (adjacent breast ducts acting as internal control)



Discussion

Desmoid fibromatosis of the breast is a rare benign mesenchymal tumor characterized by locally aggressive behavior and a tendency for recurrence, without metastatic potential. Its clinical and radiological resemblance to breast carcinoma makes it a significant diagnostic pitfall.

Patients commonly present with a firm, irregular, painless mass, often associated with skin tethering or dimpling, closely simulating malignancy.. Mammographically, these lesions typically appear as spiculated, high-density masses, while ultrasonography often reveals an ill-defined hypoechoic lesion with posterior acoustic shadowing. Such imaging characteristics frequently lead to misclassification as Breast Imaging Reporting and Data System (BIRADS) category 4 or 5 lesions. Although MRI can provide additional detail, imaging findings remain non-specific, necessitating histopathological confirmation.

Microscopically, desmoid fibromatosis demonstrates uniform spindle cells arranged in long sweeping fascicles within a collagen-rich stroma. The cells exhibit minimal cytological atypia, low mitotic activity, and absence of necrosis. Immunohistochemistry plays a crucial role in diagnosis, with nuclear β-catenin positivity serving as a key diagnostic marker. Tumor cells typically express vimentin and smooth muscle actin, while lacking expression of epithelial markers such as cytokeratin, thereby helping to exclude metaplastic carcinoma and other spindle cell malignancies.

The principal differential diagnosis is fibromatosis-like metaplastic carcinoma, which may share overlapping histological features but shows cytokeratin positivity. Other differentials include phyllodes tumor, nodular fasciitis, and scar tissue. Accurate differentiation is critical to avoid overtreatment.

In our case, the clinical diagnosis was carcinoma breast and mammogram showed BIRADS 5 lesion. Histopathology showed uniform bland spindle cells in woven fascicles, the neoplasm was infiltrating skeletal muscle bundles and adipose tissue. Our first differential was desmoid fibromatosis, but we have to rule out fibromatosis like metaplastic carcinoma, phyllodes tumour, myofibroblastoma. Morphological features do not always distinguish desmoid fibromatosis from phyllodes tumour or metaplastic carcinoma, which must be excluded with molecular and/or immunohistochemical studies.1

Special stain mason trichrome stained cytoplasm red in colour indicating myofibroblastic origin.

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IHC for Cytokeratin was negative,ruling out metaplastic carcinoma.IHC for CD34 was negative ruling out phyllodes and myofibroblastoma

Desmoid fibromatosis exhibits infiltrative local growth with no potential for metastasis or evolution to high-grade malignancy. Local recurrence has been reported in 25-29% of breast lesions after local excision , but the predictive value of surgical margin status is uncertain.1

Conclusion

Desmoid fibromatosis of the breast is an uncommon entity that can closely mimic carcinoma clinically and radiologically. Histopathological and immunohistochemical evaluation is essential for accurate diagnosis. Awareness of this condition can help prevent misdiagnosis and unnecessary aggressive treatment. A multidisciplinary approach and close follow-up are recommended due to the risk of local recurrence.

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