Case Report

Desmoid Fibromatosis of the Breast; A Case Report

Memenin Desmoid Fibromatozisi; Olgu Sunumu

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ABSTRACT

Desmoid-type fibromatosis of the breast is a rare, locally aggressive stromal tumor. This lesion is observed in 0.2% of all breast tumors, often occurs between the ages of 25 and 45 and the sex ratio is 1:2 (male: female). In this study, we report a case of a 57-year-old woman with breast fibromatosis, which has been mimicking breast carcinoma. On physical examination, there was an irregularly shaped mass lesion of approximately 3x2 cm fixed to the chest wall in the upper outer quadrant of the left breast. On mammography, an increase in opacity covered by fibroglandular tissue was observed in the outer middle part of the left breast. On ultrasound examination, a heterogeneous hypoechoic solid lesion with microcalcifications in places and 32x10mm in size with irregular borders and internal echoes were observed in the upper outer quadrant of the left breast. (BIRADS 4). Since the tru-cut biopsy could not differentiate between Fibromatosis-like Metaplastic Breast Carcinoma and Desmoid Fibromatosis, an excisional biopsy was performed under general anaesthesia. Wide local excision was performed due to positive surgical margins. In conclusion, desmoid-type fibromatosis has nonspecific physical examination findings and radiological imaging and is confused with malignancy. The primary treatment of this lesion is excision with a negative surgical margin

Keywords: Desmoid-type fibromatosis (DF), breast tumor, fibromatosis

ÖZET

Memenin desmoid tip fibromatozisi nadir görülen, lokal agresif bir stromal tümördür. Bu lezyon tüm meme tümörlerinin %0,2'sinde görülür, sıklıkla 25-45 yaşları arasında ortaya çıkar ve erkek: kadın oranı 1:2'dir. Bu olgu sunumunda, meme kanserini taklit eden meme fibromatozisi olan 57 yaşında bir kadın hastayı sunuyoruz. Fizik muayenede sol meme üst dış kadranda göğüs duvarına fikse yaklaşık 3x2 cm boyutlarında düzensiz şekilli kitle lezyonu izlendi. Mamografide sol meme dış orta kısmında fibroglandüler doku ile örtülü opaklıkta artış izlendi. Ultrason muayenesinde sol meme üst dış kadranda 32x10mm boyutlarında, sınırları düzensiz, iç ekosu olan, yer yer mikrokalsifikasyonlu, heterojen hipoekoik solid lezyon izlendi. (BIRADS 4). Tru-cut biyopside fibromatozis benzeri metaplastik meme karsinomu ile desmoid fibromatozis ayırt edilemediğinden genel anestezi altında eksizyonel biyopsi gerçekleştirildi. Pozitif cerrahi sınırlar nedeniyle geniş lokal eksizyon uygulandı. Sonuç olarak, desmoid tip fibromatosis nonspesifik fizik muayene bulguları ve radyolojik görüntüleme bulgularına sahiptir ve malignite ile karıştırılmaktadır. Bu lezyonun primer tedavisi negatif cerrahi sınır ile eksizyondur.

Anahtar kelimeler: desmoid fibromatozis, meme tümörü, fibromatozis

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Introduction

Desmoid-type fibromatosis of the breast is a rare, locally aggressive stromal tumor that occurs from musculoaponeurotic structures. It is a clonal fibroblastic proliferation that occurs in the deep soft tissues. Although it tends to be locally aggressive, distant metastases are not observed.[1] Desmoid-type fibromatosis is observed in 0.2% of all breast tumors.[2] This lesion often occurs between the ages of 25 and 45 and the sex ratio is 1:2 (male: female)[3]. We report a case of a 57-year-old woman with breast fibromatosis, which has been mimicking breast carcinoma.

Case Report

A 57-year-old female patient who developed a palpable left breast mass in several weeks was referred to our centre. She did not have any additional disease. The patient did not have a history of trauma or surgery in the left breast or a family history of cancer. On physical examination, there was an irregularly shaped mass lesion of approximately 3x2 cm fixed to the chest wall in the upper outer quadrant of the left breast. On mammography, an increase in opacity covered by fibroglandular tissue was observed in the outer middle part of the left breast, and the patient was recommended to be evaluated with ultrasound. (Figure 1)

On ultrasound examination, a heterogeneous hypoechoic solid lesion with micro-calcifications in places and 32x10mm in size with irregular borders and internal echoes was observed in the upper outer quadrant of the left breast. (BIRADS 4). Since the tru-cut biopsy could not differentiate between fibromatosis-like metaplastic breast carcinoma and desmoid fibromatosis, an excisional biopsy was performed under general anaesthesia.

In the pathological examination, the tumor consists of myofibroblastic cells forming intersecting long fascicles in the collagenized stroma with bland spindle nuclei, small nucleoli, and eosinophilic nuclei cytoplasm with indistinct borders. The tumor shows a highly infiltrative growth pattern into the surrounding breast tissue. One mitosis was observed in 10 HPF. No necrosis or cytological atypia was observed. There are haemorrhage foci and hemosiderin-laden macrophages within the tumor. Lymphoid aggregates were observed at the periphery of the tumor. The tumor was continuous with the anterior, base, superior, inferior, and lateral surgical margins. The medial surgical margin is intact. By immunohistochemistry, diffuse nuclear β-Catenin expression was observed. Focal Desmin, SMA, and Calponin expression were seen. Sparse cell staining was observed with S-100. PanCK, CK7, CD34 and p63 were negative. (Figure 2) The Ki67 proliferation index was 10%. Wide local excision was performed due to positive surgical margins.

Discussion

Desmoid-type fibromatosis is observed in 0.2% of all breast tumors.[2] This lesion often occurs between the ages of 25 and 45 and the sex ratio is 1:2 (male: female).[3] Although the etiology of the disease is not known, publications report that it is related to familial adenomatous polyposis, Gardner syndrome, surgical trauma, and silicone implants.[4] Our patient did not have any risk factors. It has nonspecific physical examination findings and radiological imaging and is confused with malignancy.

On physical examination, it often presents as a solitary, hard, and painless nodule. Nipple and cutaneous skin retraction and pectoral muscle fascia invasion can be observed.[5] In the study of Jörn Lorenzen et al., which is one of the largest series in the literature with 15 patients, a fixed mass was observed in 10 patients and a relatively soft mass in 4 patients. In addition, 4 patients had skin retraction. [2] In our case, there was a fixed

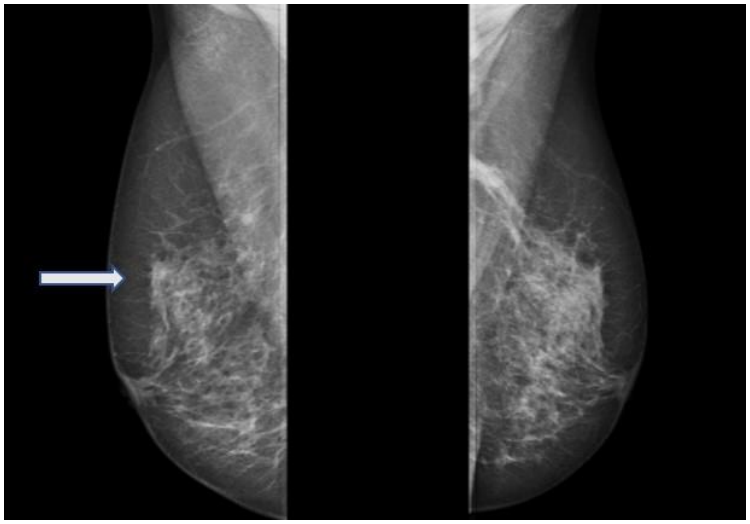

Figure-1. Left breast mammogram. An increase in opacity covered by fibroglandular tissue was observed in the outer middle part of the left breast.

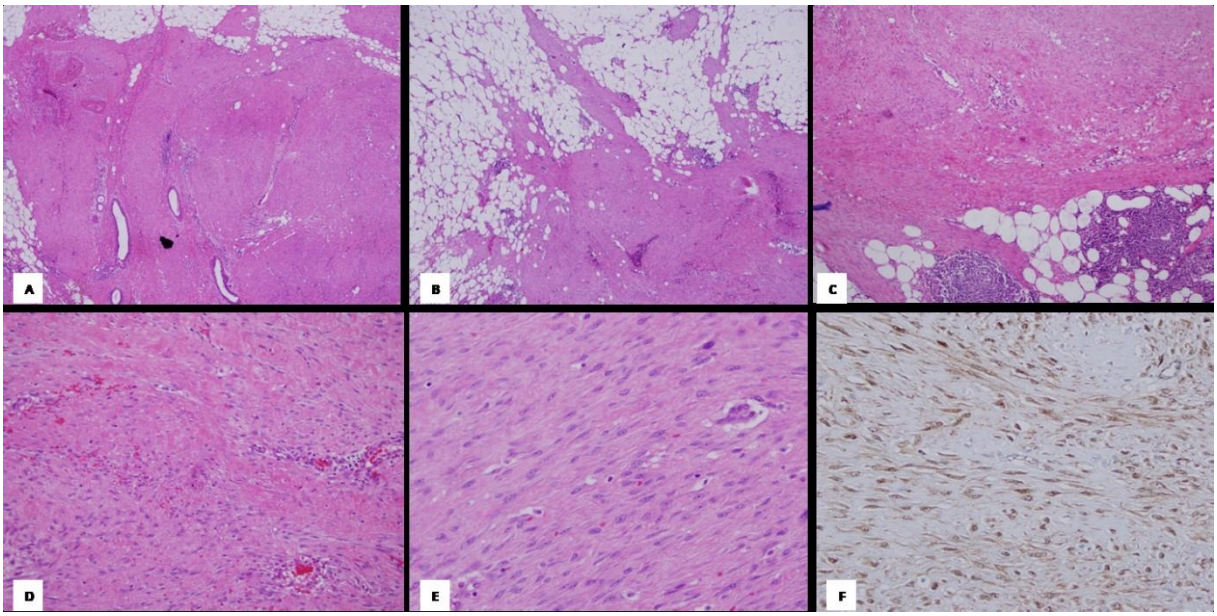

Figure-2. Tumor cells entrap normal breast ducts. B Spindle cells infiltrating the surrounding fat. C Lymphoid aggregates at the periphery of the tumor. D Collagenous stroma within the tumor. E Spindle cells appear bland with no mitotic activity. F Diffuse nuclear β-Catenin expression.

and hard palpable mass with irregular borders. In our case, there was an irregular, fixed, and hard palpable mass. However, there was no skin retraction as a deeply located mass invading the pectoral fascia was observed.

If we talk about radiological imaging, in the study of Michael R. Boland et al. (16 patients) and Jörn Lorenzen et al.; radial scars, architectural distortion, or a high-density mass appear as important mammographic findings. In our study, similar findings were observed in mammography. On ultrasound examination, the hypoechoic and irregular lesion was observed, which is consistent with the previous literature.[2, 6-10] Although there are studies suggesting MRI in the diagnosis and follow-up of desmoid fibromatosis, an excisional biopsy was performed without waiting for MRI because metaplastic carcinoma was suspected in our case.[11, 12]

Core biopsy is more valuable than fine-needle aspiration cytology. We also preferred the core biopsy first. Histopathological analyses play a critical role in the diagnosis. Diagnosis of low-grade spindle cell lesions in the breast is challenging because the differential diagnosis is broad and includes many rare lesions. The differential diagnosis includes fibromatosis-like metaplastic carcinoma, reactive spindle cell nodules, nodular fasciitis, inflammatory myofibroblastic tumor, myofibroblastoma, pseudoangiomatous stromal hyperplasia, phyllodes tumor, dermatofibrosarcoma protuberans, and spindle cell sarcomas. [13, 14] Desmoid fibromatosis is a locally aggressive mesenchymal neoplasm with infiltrative margins composed of a proliferation of uniform, cytologically bland fibroblasts, and myofibroblasts. Some histomorphologic clues to the diagnosis of desmoid fibromatosis include long fascicles of neoplastic spindle cells with entrapped benign breast glandular epithelial elements and peripheral lymphocytic aggregates. The spindle cells of desmoid fibromatosis express smooth muscle actin and show nuclear expression of Beta-Catenin, but lack cytokeratin expression. The most crucial lesion to distinguish from desmoid fibromatosis is fibromatosis-like metaplastic carcinoma. These tumors are composed of bland spindle cells resembling desmoid fibromatosis. A diagnosis of fibromatosis-like metaplastic carcinoma can be rendered based on any evidence of epithelial differentiation by histopathological or immunohistochemical analysis.[13, 15, 16] Due to tumor heterogeneity and focal cytokeratin expression, a definitive diagnosis can not be made in core biopsies. Broad macroscopic sampling and an immunohistochemical panel may be required to capture epithelial differentiation.

The primary treatment of desmoid-type fibromatosis is excision with a negative surgical margin. Wide (R0) microscopic margins resection is recommended. Positive (R1) microscopic margins can be accepted when cosmesis is problematic. There is no consensus about the indication of adjuvant radiotherapy after wide local excision. In addition, after R1 resection, there is no consensus between postoperative radiotherapy and reexcision, definitive radiotherapy can be given ın patients who cannot have surgery. It has been shown to provide sufficient local control. [17] On the other hand, a 'watch and wait' approach is also recommended in the current literature. This approach demonstrated a 60% 2-year progression-free survival similar to surgical treatment. This approach, at 3 to 6 months intervals for the first three years and then annually by MRI, consists of patient follow-up [11, 17]. Systemic treatment options for desmoid-type fibromatosis consist of non-steroidal anti-inflammatory drugs, anti-hormonal therapies, tyrosine kinase inhibitors, and conventional chemo-therapeutics.[18] Anti-hormonal agents (e.g.tamoxifen) have been frequently used alone or in combination with non-steroidal anti-inflammatory drugs. If the role of adjuvant chemotherapy is searched in the current literature, there are various regimens, including anthracycline-based regiments, sorafenib, imatinib, pazopanib, and vinblastine-methotrexate but no consensus. In addition, recent guidelines emphasize the importance of a multi-disciplinary approach. Re-excision was applied to the patient upon the decision of the multidisciplinary team council in our institution.

Conclusion

Desmoid-type fibromatosis of the breast is a rare stromal tumor. It has nonspecific physical examination findings and radiological imaging and is confused with malignancy. The primary treatment of desmoid-type fibromatosis is excision with a negative surgical margin.

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