Breast Fibromatosis: A Rare Benign Tumor that Mimics Sarcoma

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ABSTRACT

Breast fibromatosis represents a significant diagnostic challenge due to its rarity and clinical and anatomical-radiological similarity to aggressive forms of cancer such as sarcoma. This case report describes the case of a 65-year-old woman, with a history of a previous subcentimeter malignant breast neoplasm. The patient complains about worsening pain in the area of the surgical scar that radiates to the chest wall with radiological features suspicious for liponecrosis or recurrence of the disease.

Despite the intrinsic benignity of breast fibromatosis, its clinical and radiological presentation characterized by locally advanced growth patterns, it can often mimic that of a malignancy, leading to diagnostic difficulties and potential inappropriate treatments. Through the study of this case, we intend to explore the distinctive features of breast fibromatosis and the various therapeutic options, emphasizing the importance of an accurate diagnosis in order to carry out a proper management.

CASE REPORT

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A 65-year-old multiparous woman, with no risk factors and family history of breast cancer. Denies taking oral contraceptives, smoking habits, taking medications and significantly relevant pathologies.

The clinical history of this patient begins in September 2021 when during preventive check-ups on the mammographic examination in the inner lower quadrant of the left breast, an opacity with morphology and irregular margins of 8mm is found with some microcalcifications in the context, suspected for malignancy. US characteristics confirm the presence of a hypoechoic neoformation with irregular margins classified as BIRADS 5-U5 and is therefore subjected to TRUCUT needle biopsy with an 18G needle (Figure1).

Pathological examination diagnoses infiltrating ductal carcinoma with a desmoplastic reaction and mild lymphocytic infiltrate. Therefore, the patient undergoes quadrantectomy with removal of the sentinel lymph node. Intraoperative examination confirms the presence of the mass with disease-free margins and the absence of lymph node metastases. In conclusion, the patient undergoes locoregional radiation therapy.

According to follow-up protocol, in October 2022, the patient performs a mammographic examination that shows along the left submammary sulcus, at the site of the previous surgery, an opacity of 22 mm with spiculated margins associated with rare microcalcifications, with radiolucency in the context as for adipose component likely compatible with a picture of liponecrosis (Figure 2a,2b) . Ultrasound examination showed a pseudonodular hypo-anechoic finding, with corpuscular content that confirms the diagnostic hypothesis of surgical outcomes in the absence of suspicious findings of recurrence (Figure 2c).

In June 2023, the patient returned to an early check-up due to the onset of worsening pain in the paracicatricial site that radiated to the rib cage. He underwent a breast examination and ultrasound imaging again. The sonographic examination shows an increase in the size of the known formation previously reported at this location, which at the current control was about 3 cm (vs about 2 cm in October 2022) (Figure 3). The finding also showed irregular margins and presented itself with greater ecostructural inhomogeneity due to the appearance of hyperechoic areas in the context. Therefore, if a recurrence is suspected, the patient undergoes a TRU CUT biopsy with an 18G needle. Histological examination with needle biopsy excludes the presence of malignancy and makes a diagnosis

of breast fibromatosis (Figure 4). The case is discussed within the multidisciplinary team of the breast unit, and it is decided, despite the clinical and imaging characteristics, to monitor the situation with short-distance checks.

In August 2023, he also underwent an MRI examination that confirmed the presence of a unifocal lesion with irregular morphology and margins of 5cm, therefore further increased in size. It is characterized by early and intense enhancement, with an apparent absence of a cleavage plane with the underlying chest wall structures, in the absence of suspicious lymphadenomegaly (Figure 5).

In the following months, with the worsening of the pain and the persistent increase in size of the lesion, it is decided to undergo surgery on the patient (Figure 6).

Definitive histopathological examination confirmed the diagnosis of mammary fibromatosis with disease-free margins of the operative piece. Therefore, the patient was discharged with an indication to perform only follow-up checks.

DISCUSSION

Etiology & Demographics

Breast fibromatosis is a rare benign condition characterized by the growth of fibroblasts and myofibroblasts. It accounts for 0.2% of all breast cancers. It is characterized by a locally aggressive growth pattern with a high frequency of local recurrence, with no metastatic potential [9].

It usually develops from musculoskeletal aponeurotic structures. Commonly in the breast it is located deeply, adhered to the pectoralis muscle, however localizations within the mammary gland can be found.

It usually presents as a unilateral lesion, with incidence predominantly in women of reproductive age.

However, rare cases of multifocal fibromatosis [15], bilateral fibromatosis [11,14,20], male fibromatosis [7,8], and fibromatosis in the elderly and adolescents have been described.

From an etiopathogenetic point of view, there are two forms of breast fibromatosis. The first is related to genetic mutations such as the APC and CTNNB1 genes. Therefore, in patients with FAP (Familial Adenomatous Polyposis) this condition should be considered [12,14]. These genetic mutations lead to an alteration in catenin synthesis □. In these people, there is an accumulation of beta catenins, which is a stimulus for cell proliferation [13]. However, this condition is less common than the sporadic form. In particular, it has been seen that the sporadic form is related to previous traumas such as breast surgery, breast implants [17] and lipofillers [14].

Clinical & Imaging Findigs

Clinically, it appears as a hard, fixed, palpable mass adhered to the chest wall. Cases of skin and nipple retraction have been described in the literature. Pain may be associated [11].

Mammographically it appears as an irregular opacity, with spiculate margins more frequently adhered to the pectoralis muscle than intramammary localization. Intralesional calcifications are usually not present. Absence of lymphadenomegaly.

The use of tomosynthesis also allows the identification of small desmoid tumors that appear as distortions or occult spiculate masses on conventional mammography [23].

US features include hypoechoic masses with irregular morphology and blurred margins. Perilesional echogenic rims and intralesional ECD signals may be present.

According to a prospective study, CEUS can be used as an additional tool to evaluate uncertain lesions detected to convetional US and mammography [21].

The US-guided CNB is an accurate method for histological typing [19].

The MRI study shows an irregular formation with enhancement characteristics ranging from slow and persistent to rapid with plateaus with or without wash out kinetics.

Even in the T2-weighted sequence (STIR) the signal characteristics are variable, so if the myxoid component prevails we will have a hyperintense signal, if instead the fibrous component prevails it will be hypointense. The MRI study allows us to study the extent of the injury and the possible involvement of the deep muscle planes. In this case, the examination could be supplemented by the use of specific sequences suitable for the study of musculoskeletal structures [16]. CT can assess bone and periosteum destruction in patients with DF [22].

Breast fibromatosis belongs to a group of lesions called spindle cell lesions. The latter constitute a heterogeneous group of tumor entities ranging from malignant neoplasms with metastatic potential to indolent forms. However, this distinction is often challenging in daily practice, especially in FNAB biopsies due to the morphological and immunochemical overlap shown by the different lesions. Fibroblastic lesions are mainly or entirely composed of elongated spindle cells with sparse, pale to slightly eosinophilic cytoplasm, elongated nuclei with absent or only inconspicuous nucleoli. Fibroblasts are usually stained with vimentin and CD34, generic mesenchymal markers. Focal and faint spots can be seen with α-smooth muscle actin. In contrast, myofibroblasts — modified fibroblasts with the ability to contract — are plumper than fibroblasts, showing more abundant slightly or deeply eosinophilic cytoplasmic and ovoid nuclei with obvious small nucleoli [6,16].

Treatment & Prognosis

As far as treatment is concerned, surgery is the therapy of first choice, especially in symptomatic cases, characterized by rapid growth, with borderline histological and radiological

features. The surgery consists of excision of the formation with wide margins [2].

Locoregional therapies such as ablative therapy, radiotherapy and antiestrogen therapy are currently considered as adjuvant therapies to surgery in case of non-lesion-free margins, in relapses and in inoperable patients [2,3].

However, a common complication of the surgery itself is the recurrence rate, which today ranges from 15 to 77% [4].

According to studies in the literature, imaging-guided thermal ablative therapies contribute to symptom relief with a duration of more than 6 months and have a low rate of major complications [5].

The role of medical therapies based on the use of antiestrogenic drugs is still unclear [9].

Differential diagnosis

Differential diagnoses include invasive carcinoma, postoperative scar, liponecrosis, diabetic mastopathy, spindle cell carcinomas, sarcomas, phyllodes tumors, nodular fasciitis, PASH (Table 1) [6]

Immunohistochemistry and microscopic features therefore play a fundamental role in differential diagnosis [18].

CONCLUSION

Breast fibromatosis is a rare benign disease, however locally aggressive, characterized by a high frequency of recurrence and mimicking malignant pathologies.

It is important, therefore, to consider a multidisciplinary approach in the personalized choice of the patient's diagnostic-therapeutic pathway. Nowadays, surgical excision is considered the therapy of choice. Locoregional therapies such as ablative therapy, radiotherapy and antiestrogen therapy are currently considered as adjuvant therapies to surgery in case of non-lesion-free margins, in relapses and in inoperable patients [2,3].

In relation to the high frequency of recurrence after surgery and the absence of metastatic potential, a minimally invasive approach could be considered as a first-line therapy. In particular, the less aggressive forms of breast fibromatosis and in young patients innovative minimally invasive therapies such as cryoablation could be considered in order to obtain a good compromise between therapeutic and aesthetic results, reducing the risk of recurrence which today ranges from 15 to 77 % [4].

In addition, new medical therapies such as tyrosine kinase inhibitors, tamoxifen and radiation may be considered [9].

TEACHING POINT

Breast fibromatosis is a rare benign disease, however locally aggressive, characterized by a high frequency of recurrence and mimicking malignant pathologies.

QUESTION

Which of the following statements is correct regarding breast fibromatosis?

A) It is a malignant neoplasm with high metastatic potential.
B) It is a benign but locally invasive condition, without metastatic potential.
C) It always requires a radical mastectomy as a primary treatment.
D) It occurs exclusively in post-menopausal women.
E) The diagnosis does not include histological diagnosis by means of ultrasound-guided biopsy.

Correct answer:

B) It is a benign but locally invasive condition, without metastatic potential. It develops mainly in women of reproductive age, but cases of fibromatosis in adolescents and elderly people have been described. Histological diagnosis is mandatory to exclude malignancy. Usually US-guided biopsy is performed. Surgery is the therapy of first choice. The surgery consists of excision of the formation with wide margins.

However other therapeutic options are available, the choice is personalized and agreed through a multidisciplinary team.

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FIGURES

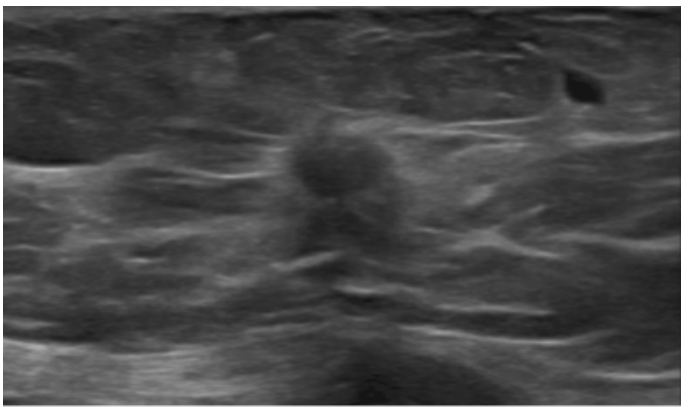

Figure 1: Left breast US. A disomogeneous ipoechoic lesion with irregular margins sospicious for malignancy. Biopsy confirms the malignant hypothesis and reveals the presence of an infiltrating ductal carcinoma.

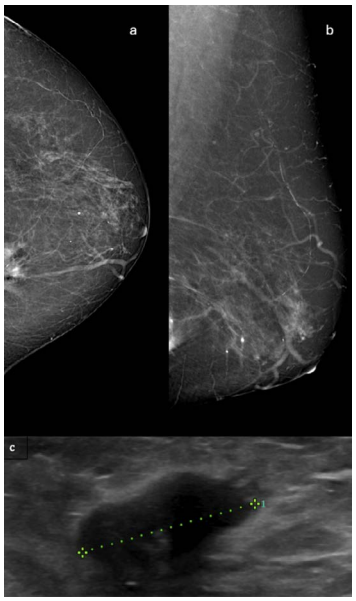

Figure 2: (a,b) Mammography of left breast (CC and MLO view). Close to the site of previous surgery opacity with spiculated margins associated with rare microcalcifications and radiolucency areas are described. US shows a pseudonodular hypo-anechoic finding, with corpuscular content that confirms diagnostic hypothesis of surgical outcomes in the absence of suspicious findings of recurrence (c) .

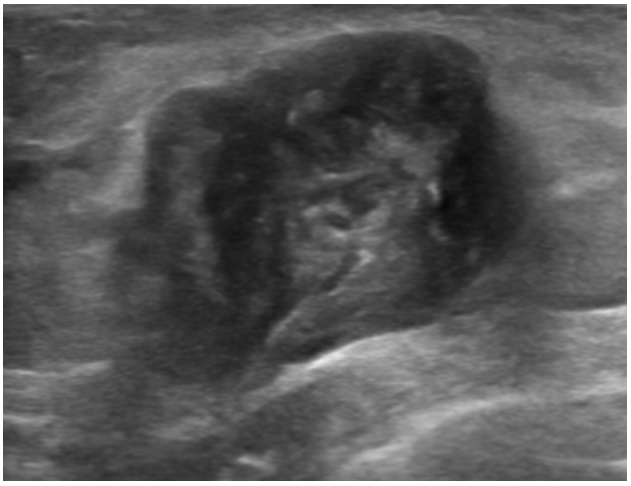

Figure 3: US shows a dimensional increase **of previous** mass with with greater ecostructural inhomogeneity due to the **appearance** of hyperechoic areas in the context.

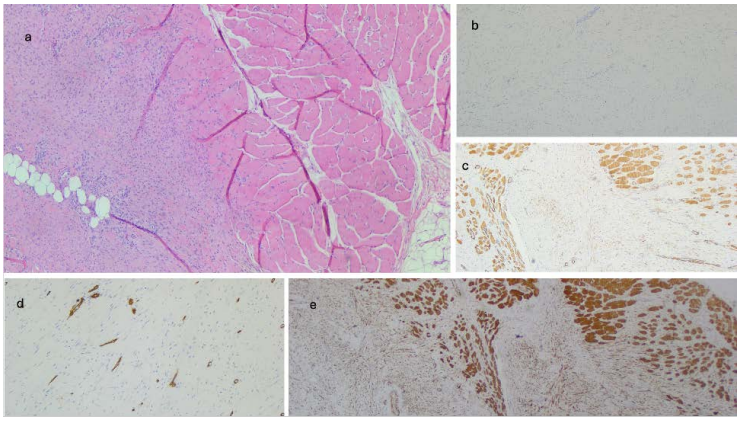

Figure 4 (a): E/E. Cytologically soft fused cells infiltrate striated muscle tissue. (b) CK5N Negativity of the entire neoplastic population. (c) Actin smooth muscle. Neoplastic cells show weak positivity. Lower staining intensity than the striated muscle present. (d) CD34 Negativity of neoplastic cells. Positive internal control (CD34 + vases). (e) Desmina. Neoplastic cells show diffuse positivity with coloration intensities comparable to that of the striated muscle present.

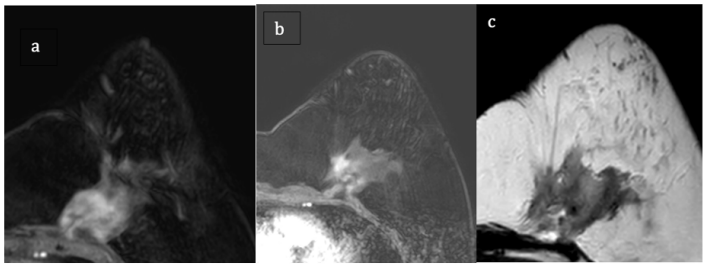

Figure 5: Axial MRI examination show a deep irregular mass with irregular margins hypointense in T2 [c] characterized by heterogenous enhancing on postcontrast T1 with fat saturation [a,b]

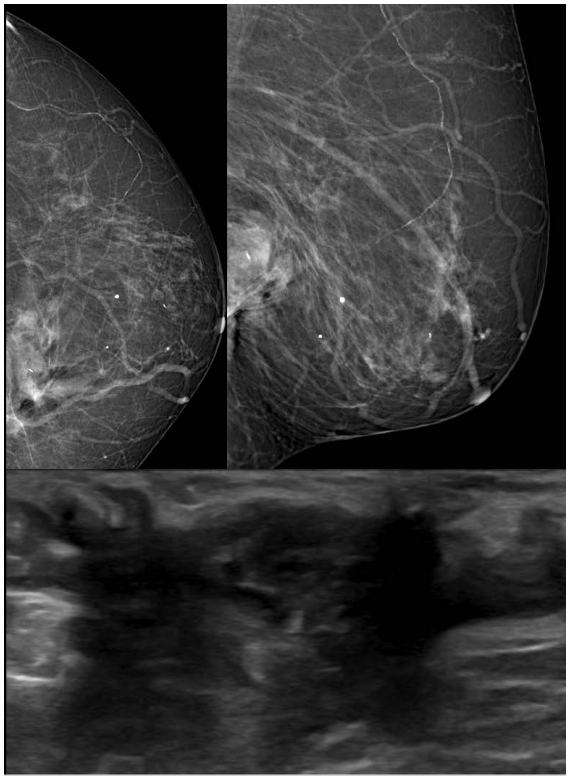

Figure 6: Mammography of left breast (MLO and CC view) shows an increase in size and radiographic density of the mass (a,b). US confirms an increased irregular mass with non-homogeneous ecostructure (c)

Table 1: Histopathological mimicry table

	Marker	Mitosis	Pleomorphism	CD34	B-catenin	Other findings
Spindle cell carcinoma (especially fibromatosis-like variant)	Epithelial (high-weight cytokeratins)	+	Nuclear	-	-	Cohesive epitheloid foci (often squamoid) and foci of CDIS
Fibrosarcomas	Stromal	+	Nuclear And Cellular	+/-	-	Rare in the breast
Phyllodes tumors	Stromal	+	Nuclear	+	-	Associated benign epithelial component
Miofibroblastoma Lipomatous	Vimentin	rare	-/+	+	-	Circumscribed solid mass that does not surround normal ducts and lobules
Nodular fasciitis	Stromal	+	+/-	-	-	Circumscribed mass that grows rapidly, painful in the subcutaneous area
PASH	CD34+	-	-	+	-	Spazi simil vascolari CD34+,CD31-, panCK-
Scar	Stromal	-	-	-	-	Foamy or haemosiderin macrophages, foreign body granulomas with possible fat necrosis

KEYWORDS

Breast fibromatosis, breast benign disease, Spindle Cell Lesions, Desmoid fibromatosis, fibroblastic proliferation