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Fibromatosis of the Breast: Report of a Case with Cytohistological Correlation

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ABSTRACT

Background: Breast fibromatosis is a rare, locally infiltrative lesion without metastatic potential that arises from either stromal fibroblasts or myofibroblasts of the breast or the pectoral fascia, extending into the breast, with its cytological and histological features only rarely been described.

Case Presentation: A 58-year-old woman, with no past medical/surgical or family history, was diagnosed on regular mammographic and ultrasound examination with a nodular tumor density, in the upper inner part of her right breast. There were no calcifications or apparent lymph nodes in the right axilla. The woman underwent FNA and US-guided biopsy and final resection biopsy under hook marking. We reviewed the cytological findings of fibromatosis of the breast, as they presented in FNAC aspirates of a non-palpable mammographic finding and the histological findings in both preoperative core-needle biopsy and excision specimen. The final diagnosis was of fibromatosis of the breast. No further actions were taken. The woman is well, without recurrence, more than four years afterwards.

Conclusion: The diagnosis in the presented patient can make the pathologists more acquainted with the cytological and pathologic features of a rare tumor entity and the clinicians with a rare breast lesion, which can mimic malignancy both clinically and radiologically. The diagnosis of fibromatosis of the breast is more reliable in excision specimens. Nevertheless, cytology can be an invaluable adjunct to histology, pre-operatively, as it can exclude cancer and help in the preoperative planning.

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INTRODUCTION

The term fibromatosis was introduced for the first time by Arthur Purdy Stout.¹ Desmoid fibromatosis is a locally aggressive, but non-metastasizing, deep-seated (myo) fibroblastic neoplasm with infiltrative growth and propensity for local recurrence.²

Fibromatosis usually occurs in the abdominal wall and the superficial musculo-aponeurotic tissues of the limbs. Fibromatosis can occur in other sites, including the breast, where it is usually the result of the extension of a lesion arising from the pectoral fascia or the shoulder girdle. Primary fibromatosis (desmoid type) of the breast has been described,^{1,3,4} and is defined as a locally aggressive, non-encapsulated and well-differentiated (myo)fibroblastic growth, which arises within the mammary gland.^{5,6} It has no metastatic potential but can recur locally after resection. It is

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important to recognize mammary fibromatosis as it can mimic cancer clinically and radiologically.⁴ Here, we report a case of fibromatosis of the breast in a 58-year-old female with cytological and pathologic correlation.

CASE PRESENTATION

A 58-year-old postmenopausal woman, with no past medical or family history of breast cancer, was diagnosed on regular mammographic, magnetic resonance imaging (MRI) (Figure 1) and ultrasound examination (Figure 2), with a nodular tumor density, of 2.2x1.6cm, in the upper inner middle part of her right breast (R, UI, 1 o'clock, middle) (Figure 1), with largely smooth and focally stellate borders, in a distance of 12.9cm from the areola, 3.97cm from the skin and 3.5cm from the front thoracic wall. There were no microcalcifications or apparent lymph nodes in the right axilla. As FNA is widely accepted and used as an initial first-line investigation tool of nodular breast lesions, the lesion was aspirated under ultrasound guidance (US-FNA). Air-dried smears stained with May-Grünwald-Giemsa (MGG), as well as alcohol-fixed smears stained with Papanicolaou stain (PAP) (Figure 3), were prepared.

The smears were hypocellular with occasional bland spindle cells embedded in hypocellular dense

stromal fragments. As it was not clear if those findings were representative of the lesion, or scanty stromal fibrous fragments (non-diagnostic), a complementary to FNA, US-guided core-needle biopsy was undertaken. The histological findings in CNB were of a bland-looking mesenchymal lesion, with long sweeping fascicles of bland spindle cells with slightly wavy nuclei in a densely collagenous matrix, with no cytologic atypia (Figure 4).

Although the histomorphological and immunohistochemical findings (positive expression for VIM, KP1, SMA, desmin (focal) and negative expression for CKAE1/AE3, CD34, S-100, ER, β -catenin (in the biopsy specimen) were more in favor of a myofibroblastic lesion of fibromatosis type, the establishment of the final diagnosis would be more reliable after the examination of the whole lesion in the excision specimen. The main concern in these cases is also to exclude fibrous scar tissue. The exclusion of breast carcinoma and the planning of wide excision of the lesion was possible. A final excision biopsy under hook marking was, therefore, undertaken. The lumpectomy specimen X-Ray confirmed the excision of the lesion, demonstrating the presence of both the mass and penetrating hook-wire (Figure 5).

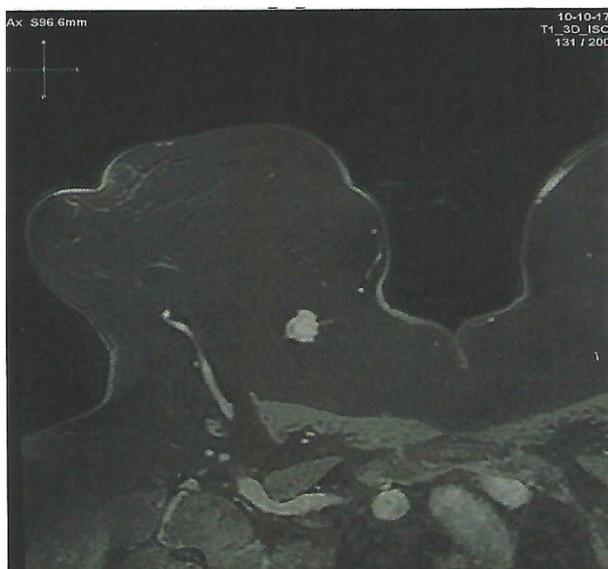


Figure 1. Magnetic resonance imaging findings of a circumscribed density deep in the breast

The surgical specimen measured 6.5x6.5x2cm, which included a circumscribed, whitish, solid tumor, of 2cm maximum dimension, of elastic consistency, close to the nearest surgical margin (Figure 6). The

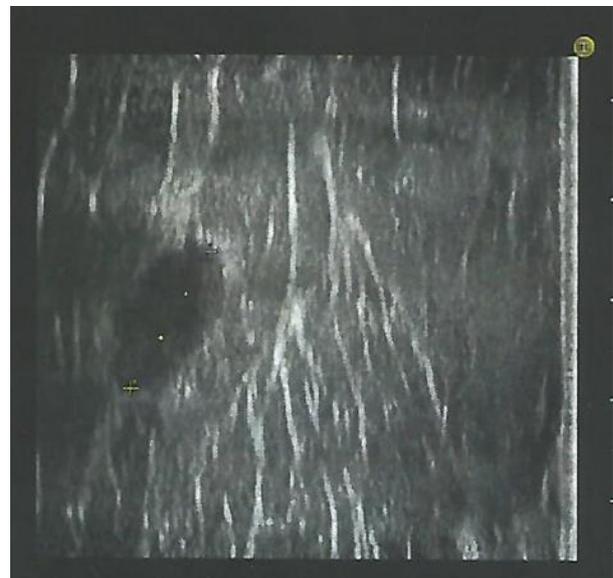


Figure 2. Ultrasound findings of a circumscribed hypoechoic lesion with posterior acoustic shadowing

lesion was fully included and examined. In the excision specimen, the lesion was mainly circumscribed (Figure 7), with focal finger-like protrusions (Figures 7) in the neighboring fat, incorporating breast lobules and ductules in rare areas (Figure 7).

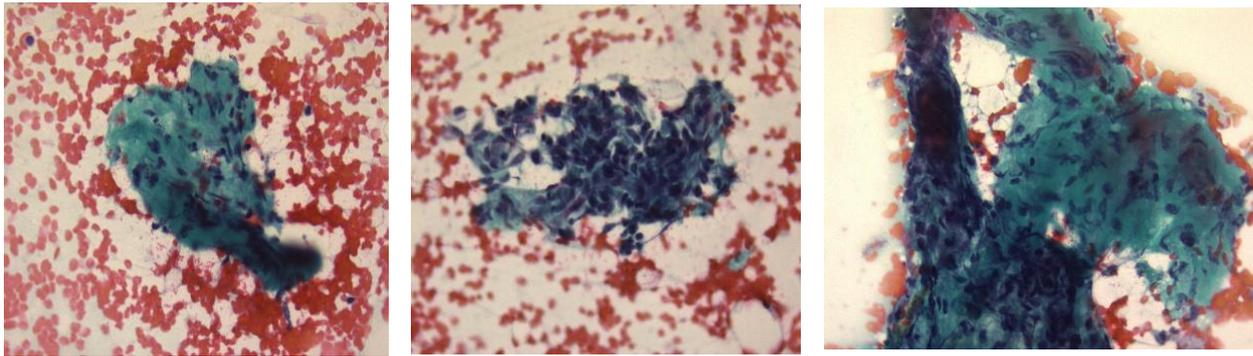


Figure 3. (a-c). Cytological findings from FNAC aspirates (PAP stain). Scanty hypocellular stromal fragments with bland spindle cells embedded in them

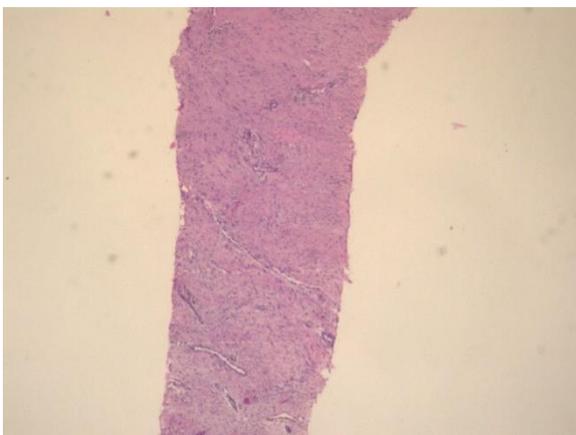


Figure 4. Histological appearance in CNB (H&E stain). Mesenchymal lesion with long sweeping fascicles of bland spindle cells



Figure 5. Specimen mammography with a peripheral circumscribed lesion



Figure 6. Macroscopic appearance of the lesion

It was composed of intersecting long fascicles of bland spindle cells with narrow, elongated, slightly wavy nuclei, without cellular atypia or appreciable mitotic activity (Figure 7), merging focally with more collagenized areas. There were focal prominent mast cells (Figure 8), as well as rare small

lymphocytic aggregates at the periphery of the lesion (Figure 8). Immunohistochemically, the spindle cells were diffusely positive for smooth muscle actin (SMA) and negative for a broad panel of antibodies to keratins, such as CKAE1/AE3, CK14, CK903, CK5/6, CD34, ER and p63. Immunostaining for β -catenin was repeated in the excision specimen in all sections of the lesion examined. In two (out of five sections), there was positive nuclear expression for β -catenin (Figure 8). Although there was no significant cytological atypia, the exclusion of fibromatosis-like metaplastic carcinoma is mandatory. Negative expression for high- and low molecular weight cytokeratins and p63, was exclusive of fibromatosis-like metaplastic carcinoma. Mast cells were positive for bcl-2. MIB-1 expression was <1%. There were no inflammatory cells, haemosiderin-laden macrophages, or foreign body giant cell reaction (indicative of trauma), in or at the periphery of the lesion (apart for rare small lymphocytic aggregates at the periphery of the lesion, as they are described in cases of fibromatosis of the breast).

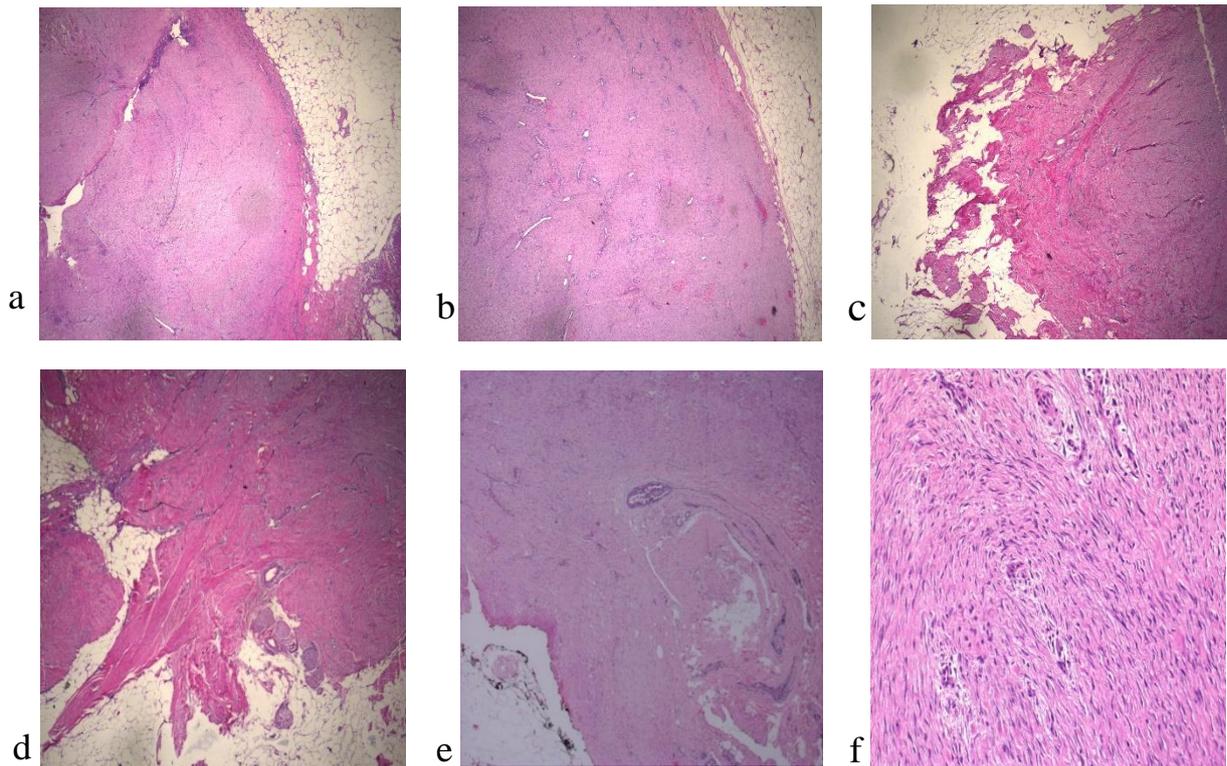


Figure 7(a-f). Histologic whole-mount sections from excision specimen (H&E): a. Circumscribed margins of the lesion, b. Obvious gaping blood vessels, throughout the lesion c, d. Focal peripheral finger-like protrusions, e. Enclosed breast parenchyma at the periphery of the lesion, f. Mildly cellular areas of fibromatosis, with intervening blood vessels. Nuclear spacing is obvious

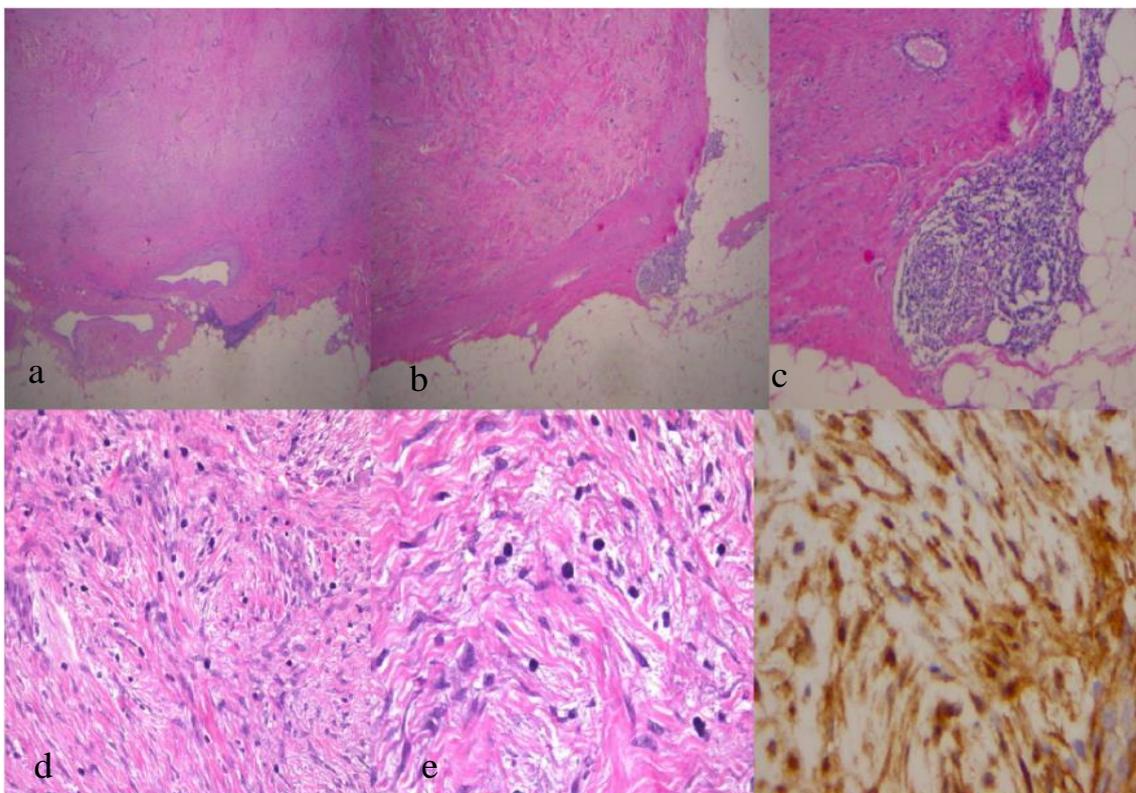


Figure 8(a-f). Histologic whole-mount sections from excision specimen (H&E): a, b, c. Small lymphocytic aggregates at the periphery of the lesion (different magnifications) d, e. Focal prominence of mast cells (different magnifications) f. Immunohistochemical (IHC) positive nuclear staining for β -catenin



The histological and immunohistochemical features were fully consistent with desmoid fibromatosis of the breast. Although the lesion focally involved the surgical margins, no further actions were taken, as the predictive value of the surgical margin status is uncertain⁴ and positive margins do not recur in all patients. The woman is well and without recurrence after more than four years and yearly mammographic follow up.

DISCUSSION

Desmoid-type fibromatosis is a locally aggressive, infiltrative spindle cell lesion composed of (myo)fibroblasts and a propensity for local recurrence, but with no metastatic potential, which usually occurs in deep soft tissues.² Typically, it occurs in one of three main clinical settings: extra-abdominal, abdominal and intra-abdominal fibromatosis. Extra-abdominal fibromatosis may occur in almost any anatomic location, including the breast.

Primary breast fibromatosis accounts for approximately 0.2% of primary breast tumors and less than 10% of desmoid fibromatosis.^{3,4,5} Extension to breast tissues from the pectoralis fascia can also occur. Fibromatosis of the breast affects women with a mean age of 43, although the reported age range is broad (17-83 years).^{3,4,5} Although the exact pathogenesis of mammary fibromatosis is not clear, a history of surgery, trauma to the involved area or saline-filled breast implants has been described.^{4,7,8,9,10} Some cases are associated with familial adenomatous polyposis (including Gardner's and Turcot syndromes) and germline mutations in APC/ β -catenin pathway. Sporadic cases are also associated with mutations in APC gene or β -catenin gene.¹¹ Estrogen is thought to influence growth.¹² In our patient, there was a history of an endometrial polyp excision. There was no FAP history in her family or past surgical manipulation. It is of note that although there was a suggestion of a scar in the upper outer quadrant of the right breast in radiologic images, the woman firmly denied having previous surgical manipulations in her breast.

Clinically, desmoid fibromatosis presents as a painless, palpable firm tumor, sometimes with retraction and dimpling of the overlying skin, or the nipple,^{5,12} or as an incidental mammographic finding, causing concern for carcinoma. On imaging, fibromatosis often presents as a mammographic density with spiculated margins, which may mimic carcinoma,¹⁰ and less commonly as a mass with circumscribed margins. Calcifications are unusual in fibromatosis. Rarely, the tumor is non-palpable and detected initially by mammography,³ as in our case. They present as nodular soft tissue masses and thus are amenable to fine needle aspiration cytology (FNAC). The mammographic findings in our case were of a

circumscribed density in the breast, distant from the thoracic wall. Ultrasound was also in favor of a circumscribed, solid hypoechoic lesion.

Pre-operative diagnosis of fibromatosis by FNAC is challenging and frequently inconclusive, owing to the indistinct cytomorphological features and low yield of dense fibrous tissue aspirates. Although FNAC is a well-established initial front-line diagnostic procedure for tumoral lesions, CNB is more accurate than FNAC in diagnosing and sub-typing soft tissue lesions.¹³ FNAC is difficult and often yields fibrous, non-diagnostic material. When successful, it shows a few small or larger mildly cellular fragments with spindle cells embedded in them. There are a few case series, as well as case reports, in the English literature, of FNAC of fibromatoses, which include fibromatosis of the breast,¹³⁻¹⁷ concerning the cytological features of this rare tumor entity. FNAC from fibromatoses may yield a spectrum of cytological findings based on the site aspirated and the phase and the age of the lesion, ranging from the cellular, mildly atypical aspirate mimicking low-grade sarcoma to acellular or hypocellular benign-appearing aspirate.^{14,15} Initial diagnosis of fibromatosis may be challenging, but recurrent lesions can be easily recognized on FNAC. Nevertheless, fine needle aspiration cytology, although not entirely specific, may be a source of important information in patients with breast fibromatosis. In particular, it confidentially allows the exclusion of breast cancer and other more common diseases and is useful in planning a surgical approach to the lesion.⁹

Mammary fibromatoses are histologically similar to those arising elsewhere in the body. Microscopically, they are characterized by interlacing, proliferating fibroblasts of long, sweeping and intersecting fascicles of bland spindle cells, a varying amount of collagenization, thin-walled blood vessels with a gaping or staghorn appearance, perivascular lymphocytic infiltrate at the advancing edge of the lesion and finger-like extensions into the surrounding fat. The cellularity of the lesion varies with the patient's age.¹⁸ Lesions in younger patients are significantly more cellular than those of the peri- and postmenopausal age. In the peri- and postmenopausal patients, the lesions are significantly more fibrous with more prominent inflammatory cells, including those that mimic keloid. Lymphocytic aggregates are typically seen at the periphery of the lesion, with lymphoid follicles only occasionally seen. A mild degree of pleomorphism may be present, but mitotic figures are usually absent. The focal prominence of mast cells has been described.

Breast spindle cell lesions are generally classified into two main categories: bland-appearing and malignant-appearing.¹⁹ Pathologically, fibromatosis must be differentiated from scar/reactive changes due



to prior surgery or trauma, myofibroblastoma, nodular fasciitis, phyllodes tumor and spindle-cell (fibromatosis-like metaplastic) carcinoma.

Core needle biopsy can be an accurate method for diagnosing fibromatosis, although it is challenging. Ancillary techniques, including immunohistochemistry (IHC) can be helpful. Fibromatosis cells typically display β -catenin nuclear staining, smooth muscle actin (SMA) cytoplasmic staining, and lack of immunoreactivity for CKs, p63 and CD34. CD34 is expressed in the stroma-rich phyllodes tumors myofibroblastoma and solitary fibrous tumor. In cases of scar, diagnosis can be facilitated by a history of trauma or previous surgery and recognition of other features like fat necrosis, haemosiderin deposition, foamy macrophages and foreign body giant cells. The distinction from fibromatosis-like metaplastic breast carcinoma, particularly on CNB, is of crucial importance to ensure appropriate management. The presence of nuclear spacing in the long fascicles of fibromatosis is something that is typically missing in fibromatosis-like MBC.¹⁹

The UK NHSBSP (National Health Service Breast Cancer Screening Programmes) and RCPATH (Royal College of Pathologists) guidelines recommend categorizing benign-appearing breast spindle cell lesions (BSCLs) that lack defining features on morphology or IHC on CNB as B3 (lesions of uncertain malignant potential).^{20,21} This is generally accompanied by a recommendation for further tissue sampling or excision for a definite diagnosis and to exclude the possibility of fibromatosis-like MBC. However, if the diagnosis of fibromatosis is confirmed on CNB, a conservative approach or therapeutic approach (excision with free margins) is recommended.²²

Desmoid fibromatosis has a propensity for local recurrence within the first three years, with reported recurrence rates in the breast ranging from 20 to 30%. Size, atypia, cellularity and number of mitoses cannot predict the likelihood of recurrence.³ Recurrences do occur despite negative surgical margins. There is no capacity for metastatic spread. Our patient is well, with no sign of recurrence, more than four years after surgery.

Treatment should be based on an initial excision biopsy followed by a wider complete excision, with

attempts aimed at uninvolved margins if the residual disease is suspected because of positive margins detected on paraffin sections. Frozen sections are unreliable in making the correct diagnosis and in assessing the margins and should be avoided.^{4,6} Not all patients with positive margins will experience recurrence.²³

The management of fibromatosis has traditionally involved wide excision with negative surgical margins, as a mainstay of therapy, sometimes necessitating mastectomy and/or chest wall resections. Some centers are shifting towards a more conservative watchful waiting approach, particularly for asymptomatic tumors.²⁴ There is no well-established role for adjuvant therapies, although radiotherapy and anti-estrogenic therapy have been used in some centres,²⁵ for multiple recurrences.¹²

A non-operative approach to recurrences with regular mammographic follow-up has been adopted for the first time by Yiangou et al.,⁶ as spontaneous regression of extra-abdominal fibromatosis has been described.^{5,26} Trey Thomas et al.,²⁷ suggests that patients should undergo quarterly clinical examination for a minimum of three years, as the majority of local recurrences manifest within this time frame.

CONCLUSION

Although fibromatosis of the breast is a very rare occurrence and cytological and pathologic features have only rarely been described in the literature, our case can make pathologists more acquainted with the cytological and pathological features of this rare tumor entity, especially in association with a characteristic site of occurrence.

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None.

CONFLICT OF INTEREST

The authors have no conflicts of interest to report in relation to this work.

ETHICAL CONSIDERATIONS

Informed consent obtained from the patient to present her clinical history and medical document to publish in the Archives of Breast Cancer.

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