



DOI: 10.4274/ejbh.galenos.2026.2025-10-6

Eur J Breast Health

Primary Synovial Sarcoma of the Breast Initially Mimicking Dermatofibrosarcoma Protuberans: A Case Report

Ines Mkhinini¹, Hammadi Jawaher¹, Samar Knaz¹, Nour Rouis¹, Chayma Selmi¹, Amira Ounissi¹,
 Rania Boukadida¹, Yosra Fejji², Salwa Mejri², Ridha Fatnassi¹

¹Department of Gynecology and Obstetrics, Ibn El Jazzar Hospital, Kairouan; University of Sousse Faculty of Medicine of Sousse, Sousse, Tunisia

²Department of Anatomopathology, Ibn El Jazzar Hospital, Kairouan; University of Sousse Faculty of Medicine of Sousse, Sousse, Tunisia

ABSTRACT

Primary synovial sarcoma of the breast is an extremely rare malignancy, representing less than 1% of breast tumors. Its clinical presentation can mimic other spindle cell neoplasms, including dermatofibrosarcoma protuberans (DFSP), making diagnosis challenging. We report the case of a 60-year-old postmenopausal woman presenting with a firm, mobile 10 cm mass in the upper outer quadrant of the left breast, classified on imaging as breast imaging reporting and data System 4. A core needle biopsy initially suggested DFSP because of strong CD34 positivity. A simple mastectomy revealed a highly cellular, spindle-cell tumor arranged in fascicles, forming a characteristic whorled pattern. Immunohistochemistry showed CD34 negativity, transducin-like enhancer of split 1 positivity, B-cell lymphoma 2 positivity, and focal epithelial membrane antigen positivity. Molecular analysis confirmed SYT-SSX1 fusion, establishing the diagnosis of monophasic synovial sarcoma. Surgical margins were negative, and one-year follow-up showed that the patient remained disease-free. This case highlights the importance of immunohistochemistry and molecular testing for accurate diagnosis and appropriate management.

Keywords: Synovial sarcoma; breast; dermatofibrosarcoma protuberans; immunohistochemistry; rare tumor; mastectomy

KEY POINTS

- Synovial sarcoma of the breast is extremely rare and can initially mimic dermatofibrosarcoma protuberans.
- Core needle biopsy may be misleading; a full immunohistochemical and molecular evaluation is essential.
- Wide local excision with negative margins is the cornerstone of treatment.
- Axillary dissection is rarely indicated.
- Awareness of this diagnostic pitfall improves early recognition and management.

Corresponding Author: Ines Mkhinini, MD;

E-mail: mkhinini.ines2020@gmail.com **ORCID:** orcid.org/0009-0009-2949-1759

Received: 22.10.2025 **Accepted:** 01.01.2026 **Epub:** 06.04.2026

Cite this article as: Mkhinini I, Jawaher H, Knaz S, Rouis N, Selmi C, Ounissi A, et al. Primary synovial sarcoma of the breast initially mimicking dermatofibrosarcoma protuberans: a case report. Eur J Breast Health. [Epub Ahead of Print]



Introduction

Primary breast sarcomas are rare, accounting for less than 1% of all breast malignancies, and synovial sarcoma (SS) of the breast is exceptionally uncommon (1-3). SS is a high-grade spindle-cell malignancy that most frequently arises in the extremities of young adults, but it can rarely occur in the breast. Its monophasic spindle-cell variant may mimic other spindle-cell neoplasms, including dermatofibrosarcoma protuberans (DFSP), fibromatosis, and solitary fibrous tumors, creating diagnostic challenges.

DFSP is a low-grade cutaneous sarcoma characterized by locally aggressive behavior and low metastatic potential. In rare instances, DFSP can involve the breast, usually presenting as a slowly enlarging, firm, nodular mass. Small biopsies can make it difficult to differentiate DFSP from other spindle cell tumors, especially SS, since DFSP typically exhibits strong CD34 positivity (1, 2, 4).

This report describes a case that was initially diagnosed as DFSP on core biopsy but was ultimately confirmed as a primary monophasic SS after surgical excision, illustrating the importance of detailed immunohistochemical and molecular testing in rare breast tumors.

Case Presentation

Patient Information

A 60-year-old postmenopausal woman with no personal or family history of breast malignancy presented with a left breast mass that had been slowly enlarging over several decades. The patient reported a recent aesthetic concern due to the size of the mass, which measured approximately 10 cm. There was no history of trauma, previous breast surgery, or radiation exposure.

Clinical Findings

Physical examination revealed a firm, mobile mass in the upper outer quadrant of the left breast. The overlying skin was intact; no nipple retraction or discharge was present. No palpable axillary lymphadenopathy was noted (Figure 1).

Diagnostic Assessment

Imaging

- **Mammography:** Large oval opacity with lobulated contours and coarse macrocalcifications Breast Imaging Reporting and Data System (BI-RADS) 4 (Figure 2).
- **Ultrasound:** Hypoechoic, hypervascular mass measuring 10×5 cm with lobulated margins (Figure 3).

Biopsy

Core needle biopsy suggested DFSP, based on spindle-cell morphology and strong CD34 positivity.

Further Evaluation

A thoraco-abdominopelvic computed tomography scan revealed no regional or distant metastases.



Figure 1. Breast examination reveals a prominent, firm, mobile mass measuring 10 cm located in the upper outer quadrant of the left breast

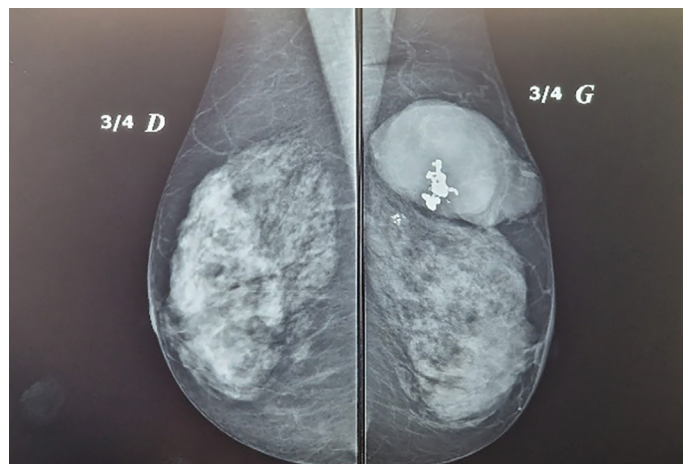


Figure 2. The external oblique mammographic view reveals a large oval opacity with lobulated contours in the upper outer quadrant, measuring approximately 10 cm in its longest axis, containing coarse granular macrocalcifications clustered in groups. Lesion classified as Breast Imaging Reporting and Data System (BI-RADS) 4

Diagnosis

- **Initial:** DFSP on core biopsy.
- **Final:** Monophasic SS confirmed after surgical excision based on histopathology, immunohistochemistry, and SYT-SSX1 fusion.

Therapeutic Intervention

A simple mastectomy without axillary dissection was performed because of the large tumor size and the difficulty in achieving clear margins with breast-conserving surgery.

Histopathological Findings

- **Tumor:** Highly cellular, composed of monomorphic spindle-cell cells arranged in fascicles.
- The cells had eosinophilic cytoplasm and oval nuclei with fine chromatin; mitotic figures were rare (Figures 4-9).

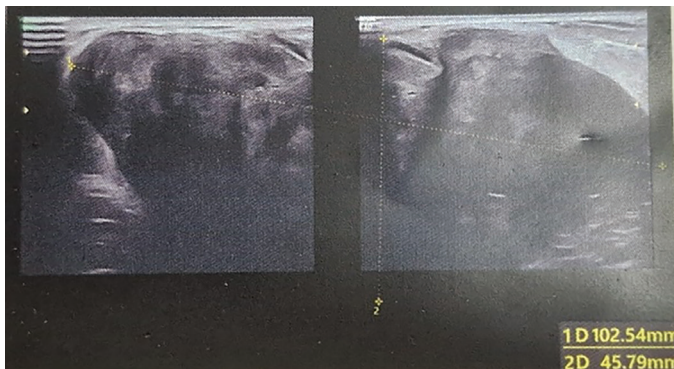


Figure 3. Ultrasound imaging shows a large tissue mass in the left upper outer quadrant measuring 10×5 cm along its major axes; the mass is hypoechoic with lobulated contours

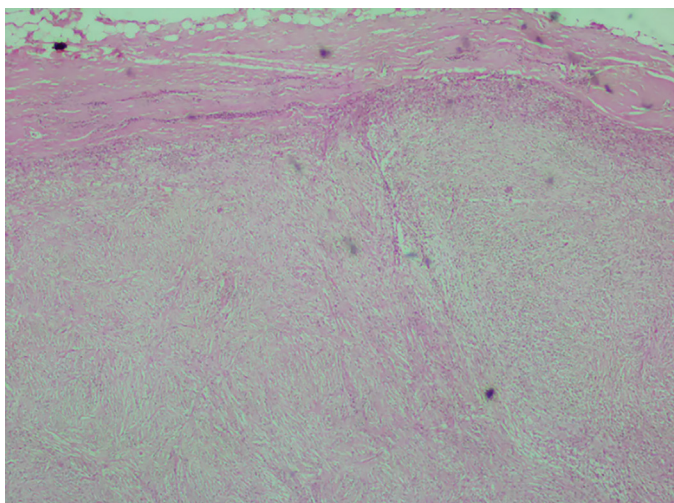


Figure 4. Low-power hematoxylin and eosin view showing a spindle-cell tumor located in the breast parenchyma beneath the dermis, without the honeycomb infiltration typical of dermatofibrosarcoma protuberans

- **Immunohistochemistry:** CD34- (Figure 7), TLE1+ (Figure 8), STAT6- (Figure 9), B-cell lymphoma 2 (BCL2)+, focal epithelial membrane antigen (EMA)+.
- **Molecular analysis:** SYT-SSX1 fusion confirmed.
- **Surgical margins:** Negative.
- **Postoperative course:** Uneventful; no adjuvant therapy administered.

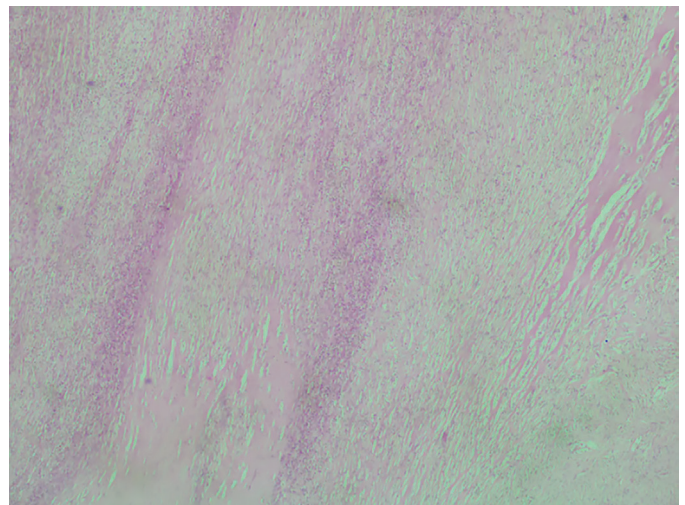


Figure 5. Intermediate-power hematoxylin and eosin view showing uniform spindle cells arranged in long fascicles, lacking the storiform pattern expected in dermatofibrosarcoma protuberans

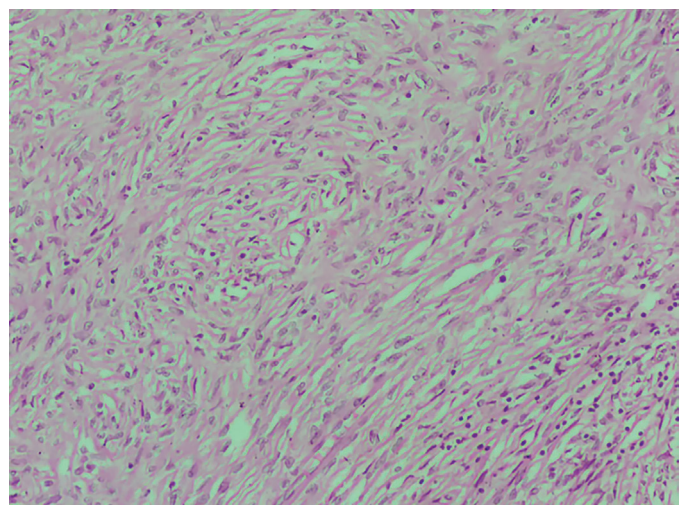


Figure 6. High-power hematoxylin and eosin view demonstrating intersecting fascicles and whorled arrangements characteristic of monophasic synovial sarcoma

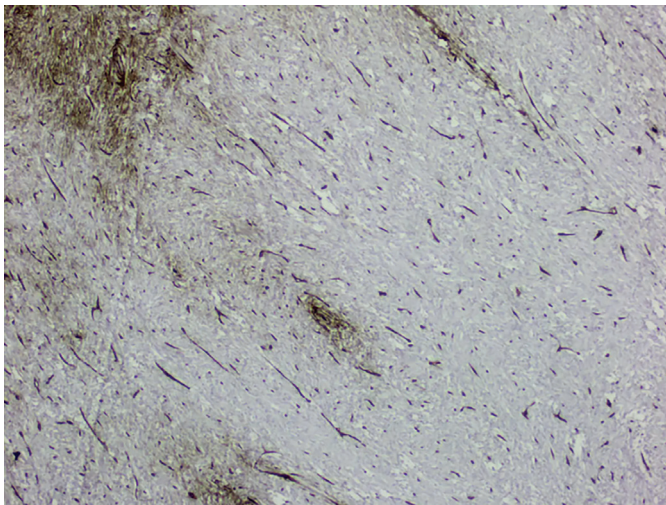


Figure 7. CD34 immunostaining showing complete negativity of tumor cells. This finding argues strongly against dermatofibrosarcoma protuberans, which typically shows diffuse CD34 positivity

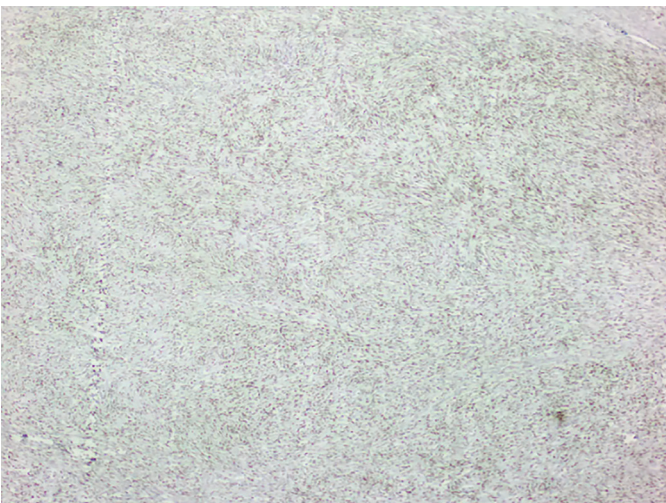


Figure 8. Immunohistochemistry showing diffuse nuclear positivity for transducin-like enhancer of split 1, supporting the diagnosis of monophasic synovial sarcoma of the breast

Follow-up and Outcomes

The patient remains disease-free at one-year follow-up. Clinical and imaging surveillance every six months has shown no recurrence. The patient expressed satisfaction with the outcome and relief after complete excision.

Discussion and Conclusion

Primary SS of the breast is an exceptionally rare malignancy. The first reports, which appeared in the late 1990s, described small case series and emphasized diagnostic challenges arising from overlapping histological features with other spindle cell tumors

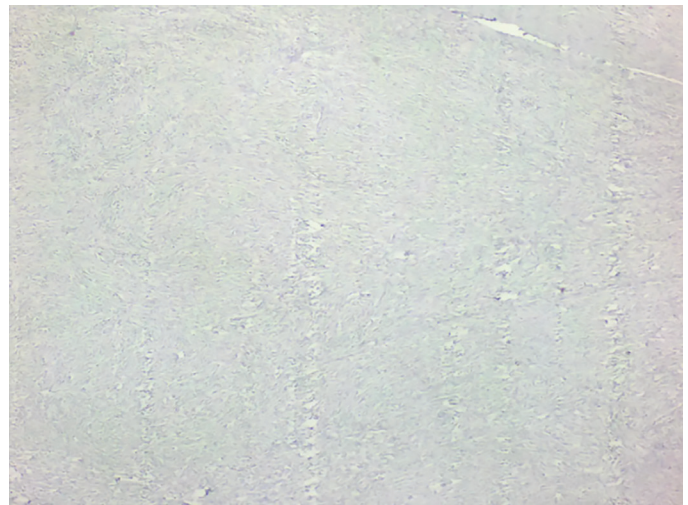


Figure 9. Signal transducer and activator of transcription 6 immunostaining negative, excluding solitary fibrous tumor

(5, 6). Its rarity, combined with the predominance of spindle cell morphology, makes it a significant diagnostic challenge, particularly on core needle biopsies. In the present case, the lesion was initially diagnosed as DFSP due to strong CD34 positivity on biopsy, illustrating a common pitfall recognized in earlier reports (1, 2). Misdiagnosis may result in inappropriate surgical planning, underscoring the necessity of thorough histopathologic, immunohistochemical, and molecular evaluation.

SS of the breast typically presents as a painless, firm, and slow-growing mass. Early descriptions highlighted the tendency for these tumors to mimic benign breast lesions or low-grade sarcomas, complicating early detection (5, 6). In our patient, the lesion had been present for decades and enlarged slowly, consistent with prior observations of delayed presentation in elderly patients. Most reported cases localize to the upper outer quadrant and lack overlying skin changes or nipple involvement, contributing to diagnostic delay (4, 6).

Radiologically, SS of the breast is non-specific. Ultrasound generally reveals a hypochoic, lobulated, and hypervascular mass, while mammography may show a well-circumscribed, dense lesion, sometimes with coarse calcifications (6, 7). Magnetic resonance imaging can provide additional soft tissue characterization, but it cannot definitively distinguish SS from DFSP or other spindle cell neoplasms (6). These imaging limitations were confirmed in our patient, emphasizing that histopathological evaluation remains the cornerstone of diagnosis.

Histologically, monophasic SS consists of uniform spindle cells arranged in fascicles, often with elongated nuclei and eosinophilic cytoplasm. Mitotic activity may be low or

moderate, and necrosis is usually absent in low-grade lesions (3, 5). Immunohistochemistry is crucial: TLE1 demonstrates high sensitivity and specificity for SS; CD34 negativity excludes DFSP; STAT6 negativity rules out solitary fibrous tumor. Positivity for BCL2 and EMA further supports the diagnosis (3, 7, 8). Molecular confirmation via detection of *SYT-SSX1* or *SYT-SSX2* fusion genes is considered the gold standard and was decisive in our case (9, 10).

The differential diagnosis includes DFSP, fibromatosis, myofibroblastic tumors, malignant peripheral nerve sheath tumors, and solitary fibrous tumors. DFSP typically exhibits diffuse CD34 positivity and a storiform pattern, occasionally leading to misdiagnosis on limited biopsy samples, as noted in earlier case reports (1, 2, 4). Our case illustrates the importance of combining morphology, immunohistochemistry, and molecular studies for accurate tumor classification.

Treatment of primary breast SS centers on complete surgical excision with negative margins, which remains the mainstay of therapy (3-6). Wide local excision or mastectomy can be selected depending on tumor size, location, and cosmetic considerations. Axillary lymph node dissection is rarely necessary, given the low incidence of nodal metastasis (3, 5). Hematogenous spread, particularly to the lungs, is more common in advanced or metastatic disease (3, 5). Adjuvant radiotherapy or chemotherapy may be considered for high-risk cases, including those with positive margins or high-grade tumors, although the data remain limited (6, 7, 10).

Prognosis is influenced by tumor size, margin status, mitotic index, and necrosis. Achieving negative surgical margins significantly reduces the risk of local recurrence. Long-term follow-up is recommended, with clinical and imaging surveillance every 6–12 months during the first 2–3 years and annually thereafter (3, 5, 7).

This case emphasizes several key points. Long-standing breast masses with spindle cell morphology, even in elderly patients, require thorough evaluation. Misdiagnosis on core biopsy is possible, underscoring the need for immunohistochemistry and molecular studies. Finally, appropriate surgical management with negative margins can provide excellent local control and a favorable prognosis, even for large tumors (3-7, 10).

This case underscores the diagnostic challenge posed by primary SS of the breast, which was initially misdiagnosed as DFSP. Comprehensive immunohistochemistry and molecular testing

are essential to avoid misdiagnosis. Complete surgical excision with negative margins remains the mainstay of treatment, and long-term follow-up is necessary due to potential local recurrence.

Ethics

Informed Consent: Written informed consent was obtained from the patient for publication of this case report.

Footnotes

Authorship Contributions

Surgical and Medical Practices: I.M., S.K., R.F.; Concept: I.M., H.J., C.S., A.O., R.B., R.F.; Design: H.J., N.R.; Data Collection or Processing: I.M., H.J., N.R.; Analysis or Interpretation: H.J., S.K., R.B., Y.F., S.M., R.F.; Literature Search: H.J., S.K., N.R., C.S., A.O.; Writing: I.M., H.J., S.K., N.R., C.S., A.O.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

References

- Dickson BC, Hornick JL, Fletcher CDM, Demicco EG, Howarth DJ, Swanson D, et al. Dermatofibrosarcoma protuberans with a novel COL6A3-PDGFD fusion gene and apparent predilection for breast. *Genes Chromosomes Cancer*. 2018; 57: 437-445. (PMID: 30014607) [\[Crossref\]](#)
- Al-Farsi A, Al-Brashdi A, Al-Salhi S, Al-Rahbi N, Al-Rahbi S, Al-Masqari M, et al. Dermatofibrosarcoma protuberans mimicking primary breast neoplasm: a case report and literature review. *Sultan Qaboos Univ Med J*. 2020; 20: e368-e371. (PMID: 33110655) [\[Crossref\]](#)
- Do Q, Katiyar V, Breaux A, Singh V. Primary monophasic breast synovial sarcoma in a female patient. *BMJ Case Rep*. 2021; 14: e242313. (PMID: 34373249) [\[Crossref\]](#)
- Juliana Lopes de Aguiar JLA, Sousa UW, Batista de Aquino FM, Gurgel de Lira L, de Andrade Almeida IMR. Monophasic breast synovial sarcoma – case report. *Mastology*. 2020; 30(Suppl 1): 10.
- Essary LR, Vargas SO, Fletcher CD. Primary synovial sarcoma of the breast: a clinicopathologic study of 15 cases. *Am J Surg Pathol*. 1999; 23: 26-35. [\[Crossref\]](#)
- Jeong YJ, Kim SH, Kim YH, et al. Primary synovial sarcoma of the breast: imaging findings. *AJR Am J Roentgenol*. 2006; 186: 285-289. [\[Crossref\]](#)
- Yoshida A, Tsuta K, et al. Primary synovial sarcoma of the breast: a systematic analysis of reported cases. *Pathol Int*. 2010; 60: 415-422. [\[Crossref\]](#)
- Shidham VB, et al. Synovial sarcoma of the breast: cytologic diagnosis and differential considerations. *Diagn Cytopathol*. 2000; 22: 377-381. [\[Crossref\]](#)
- Miettinen M, et al. TLE1 expression in synovial sarcoma: a diagnostic marker. *Am J Surg Pathol*. 2008; 32: 1436-1443. [\[Crossref\]](#)
- Ladanyi M. Fusions of the SYT and SSX genes in synovial sarcoma. *Oncogene*. 2001; 20: 5755-5762. (PMID: 11607825) [\[Crossref\]](#)