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Caught on Second Look: A Rare Case of Cutaneous Leiomyosarcoma of the Breast at the Crossroads of Rarity and Mimicry

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Abstract

Cutaneous leiomyosarcoma (CLMS) is a rare smooth muscle malignancy of the skin, constituting 2–3% of cutaneous soft tissue sarcomas and representing an exceedingly small fraction, approximately 0.04%, of all reported neoplasms. Among cutaneous sarcomas, CLMS ranks third in frequency, following dermatofibrosarcoma protuberans and Kaposi's sarcoma. We present a rare and instructive case involving a 54-year-old female who developed a slowly enlarging, painful lesion on the left breast skin, just above the nipple-areola complex. Initial clinical evaluation and investigations, including mammography, fine-needle aspiration cytology (FNAC), and excisional biopsy, misidentified the lesion as a benign spindle cell neoplasm, likely myofibroblastoma. However, the lesion recurred within four months, prompting a wide local excision. Histopathological analysis of the excised specimen revealed an infiltrative tumor consisting of atypical spindle cells with pleomorphic nuclei and an increased mitotic rate. Immunohistochemical staining confirmed the diagnosis of cutaneous leiomyosarcoma through positivity for smooth muscle actin, desmin, and h-caldesmon. Ki-67 labelling index was 40%, indicating a high proliferative index. However, the wide local excision also further required a revision surgery for margin clearance. The case highlights the diagnostic challenges posed by CLMS, especially when clinical and histopathological features mimic benign entities. Accurate diagnosis relies on comprehensive histopathological and immunohistochemical analysis. Wide local excision with negative margins remains the cornerstone of treatment, as radiotherapy and chemotherapy are generally considered ineffective. Given the high recurrence rate and metastatic potential of CLMS, vigilant long-term follow-up is essential.

Introduction:

Cutaneous leiomyosarcoma is a rare malignant neoplasm that originates from the smooth muscle elements of the skin, including arrector pili muscles and vascular smooth muscle. The estimated incidence rate is approximately 0.2 per 100,000 people annually, which adds to the challenges of its diagnosis and therapeutic management.[1,2] CLMS predominantly affects males, with a male-to-female ratio ranging from 2:1 to 3:1. Most CLMS occur in the fifth to seventh decades of life, with subcutaneous LMS usually occurring in patients aged 50- to 80-years[1,3].[4] The most commonly affected anatomical sites are the lower extremities (50%–75%), followed by upper extremities (20%–30%), and the trunk (10%–15%)[5,6]. Primary leiomyosarcoma of the breast is an extraordinarily rare entity, representing less than 1% of breast tumors and fewer than 5% of soft tissue sarcomas, with an estimated prevalence of under 0.0006% among breast malignancies. [7,8] The precise etiology remains obscure, although potential associations have been suggested with ionizing irradiation, solar exposure, antecedent trauma, chemical exposure, and chronic inflammatory conditions including lupus vulgaris.

Clinically, CLMS often presents as a solitary, firm, painful, pruritic or paresthetic

dermal nodule. Pain is an early feature and may be due to involvement of cutaneous nerves. Overlying skin is normal or has a red color. Rarely, due to skin trauma it may ulcerate. When located in the subcutaneous tissue, cutaneous leiomyosarcoma may present as an indurated plaque.[9] Nevertheless, clinical recognition is difficult, and a definitive diagnosis can be rendered only on the basis of histopathological analysis..Differential diagnosis can be made with any solitary nodule developed on the skin or subcutaneous: lipoma, dermatofibroma, dermatofibrosarcoma, neurofibroma.[10] Histopathologically, cutaneous leiomyosarcoma is defined by a dermal proliferation of elongated spindle cells disposed in intersecting fascicles. The neoplastic cells possess blunt-ended, cigar-shaped nuclei and abundant eosinophilic cytoplasm, with mitotic figures frequently identifiable. Because of its morphologic overlap with other spindle cell tumors, immunohistochemistry is indispensable for accurate diagnosis. Characteristically, CLMS demonstrates immunoreactivity for smooth muscle actin (SMA), desmin, and h-caldesmon, while showing negativity for CD34.[3]

Case Presentation:

A 54-year-old woman presented with an eight-month history of a painful nodule over the left breast, located just above the nipple-areola complex. The lesion was firm and well-defined, measuring about 1.0 × 0.5 cm, and exhibited slow growth. The overlying skin was thickened but not ulcerated, and there was no discharge or axillary lymphadenopathy. The patient had no significant past medical history, and routine systemic investigations, including complete blood count, fasting blood sugar, liver and renal function tests, hepatitis B virus surface antigen, and HIV testing were all within normal limits or non-reactive.

Mammography revealed a small, well-defined, oval hypoechoic lesion at the 11 o'clock position in the left breast, interpreted as fibroadenoma (BIRADS Category 3). FNAC suggested a benign spindle cell lesion, and subsequent excisional biopsy diagnosed it as myofibroblastoma. Four months after surgery, the patient experienced recurrence of the lesion, which had enlarged to approximately 3 cm in diameter. A wide local excision was performed with an aim of 2 cm margin clearance to remove the recurrent tumor.

Histopathological evaluation of the recurrent lesion demonstrated a dermal tumor composed of intersecting fascicles of spindle-shaped cells exhibiting elongated, blunt-ended nuclei and eosinophilic cytoplasm. Notable features included nuclear pleomorphism, the presence of multinucleated tumor giant cells, and a mitotic rate of 2–4 per 10 high-power fields. The tumor demonstrated an infiltrative growth pattern and extending into the subcutaneous with involved margins.

Immunohistochemical (IHC) analysis was critical in reaching a definitive diagnosis. The tumor cells exhibited strong positivity for SMA, desmin, and h-caldesmon, and were negative for CD34. Ki-67 proliferation index was 40%, indicative of a high mitotic activity. Based on these findings, a final diagnosis of cutaneous leiomyosarcoma was established. Revision surgery was performed to obtain margin clearance.Follow-up imaging, including CT scans of the chest and abdomen, and a PET scan, showed no evidence of metastatic disease.

Discussion:

CLMS and myofibroblastoma are rare spindle cell neoplasms, necessitating careful differentiation for appropriate management. In the current case both the CLMS and myofibroblastoma entered the differential diagnosis, posing diagnostic and therapeutic challenges, particularly

when either of them presents in uncommon anatomical locations

The malignant cutaneous leiomyosarcoma originates from smooth muscles of dermis and hence is commonly located in lower extremities being the most frequently involved site (50–75%), followed by upper limbs and trunk.[3] In contrast, the benign MFB arises from myofibroblasts and is most frequently found in the breast, though extramammary sites such as the vulva, groin, abdominal wall, and paratesticular region have been reported.[11–15] The present case was unusual because CLMS occurred in the breast—a site far more typical for MFB but representing <1% of CLMS cases.[8] CLMS has a male predilection with a ratio ranging from 2:1 to 3:1, but the current case obscured the clinical impression and occurred in a female.[16] Age could not be used as a distinct clinical differentiator as myofibroblast occurs in a wide age range, though CLMS is restricted to 50-80 years.[14]

Though the etiology of CLMS remains largely unclear, the commonly implicated ones include ionizing radiation, chronic sun or chemical exposure, trauma, chronic infections such as lupus vulgaris and sporadic genetic aberrations such as deletions in chromosome 13q14–21.[10,17] Notably, the current case did not have any h/o long time sun exposure, recallable trauma/ long standing infection at the lesion site. However, the genetic cause of sporadic case can still hold true. Myofibroblastoma, on the other hand, is also considered part of the so-called "13q/Rb family" of tumors, which includes entities such as spindle cell lipoma and cellular angiofibroma, detectable by immunohistochemistry.[18] Hormonal receptor positivity of estrogen receptor (ER), progesterone receptor (PR), and androgen receptor (AR) in tumor cells in MFB further distinguishes its biology.[19,20]

In terms of clinical features, CLMS typically manifests as a painful, firm, reddish dermal nodule, whereas MFB presents incidentally without any pain.[10,21] Though our patient presented with a mildly on and off painful nodule, slightly favoring CLMS, the absence of other features like pruritis, and paraesthesia made MFB a plausible consideration. Mobility could not be assessed to differentiate the condition, due to the lesion's cutaneous location.[10,21] Although MFB usually demonstrates slow growth and CLMS tends to progress rapidly, our case initially appeared indolent but showed accelerated growth following the first biopsy. This atypical course contributed to the initial misdiagnosis of MFB before the final confirmation of CLMS.[10,21] The poor clinical differentiating features between the two usually warrant histopathological differentiation.

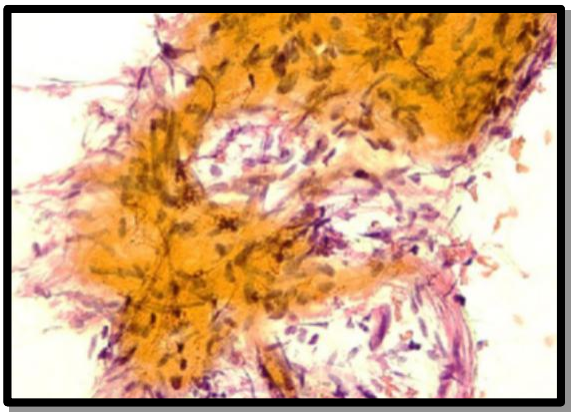
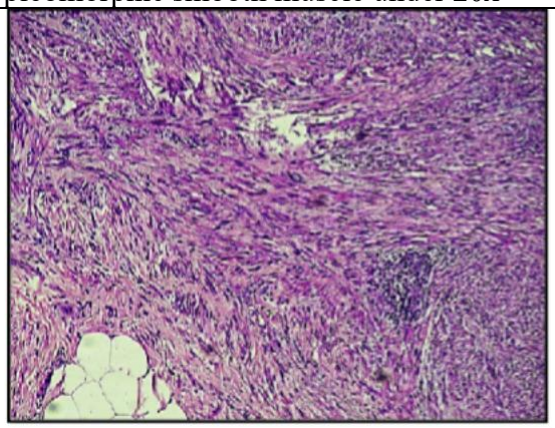
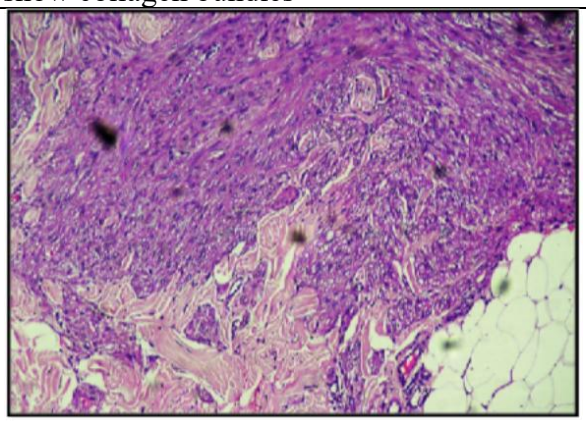
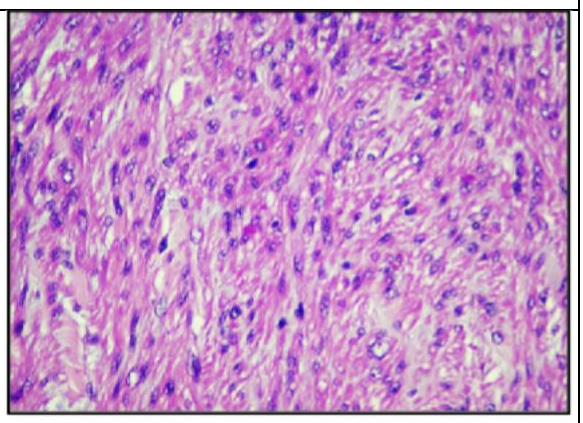
CLMS exhibits a variable histologic spectrum, ranging from nodular lesions with high cellularity, nuclear atypia, and frequent mitoses to more diffuse patterns with reduced mitotic activity. Characteristically, it is composed of spindle cells with eosinophilic cytoplasm, nuclear pleomorphism, mitotic activity, infiltrative margins and necrosis.[22] However, rare variants such as epithelioid, myxoid, granular cell, giant cell, and sclerotic types may not present the same. In contrast, MFB is well circumscribed, composed of bland spindle cells with hyalinized collagen, minimal atypia, and rare mitoses.[23] In our case, the initial biopsy revealed spindle cells with low mitotic activity, mild atypia, lack of necrosis and collagenized stroma—features aligning with MFB. However, recurrence within four months and re-excision showing nuclear pleomorphism, multinucleated tumor giant cells, mitoses(2–4/HPF), and infiltrative growth were more consistent with CLMS.

IHC ultimately confirmed the diagnosis, as the findings were consistent with CLMS, which typically shows diffuse positivity for SMA, desmin, and h-caldesmon, and negativity for CD34 and S-100, thereby

distinguishing it from MFB and other spindle cell tumors.[1,23] Radiology cannot reliably distinguish CLMS from MFB in the skin and is mainly useful for assessing extent and origin of lesion.

In our case, although the tumor measured less than 5 cm, its high proliferative activity necessitated re-excision and close surveillance, underscoring the prognostic relevance of both biological behavior and tumor spread.By contrast, myofibroblastoma is a benign lesion curable by simple excision, with recurrence being uncommon and typically related to incomplete removal.

Thus, the initial misdiagnosis arose due to shared clinical and histological features between CLMS and MFB—particularly the unusual breast location, female sex, and relatively bland initial morphology. On recurrence and detailed IHC evaluation, the diagnosis of CLMS was established, highlighting the importance of vigilance in differentiating these rare spindle cell tumors.

1a. FNAC -Benign appearing smooth muscles under 40x	1b. Interlacing facsciles and bundles of pleomorphic smooth muscle under 20x
	
1c. pleomorphic tumor cells extend into subcutaneous fat and periphery of the tumor show collagen bundles	1d. pleomorphic tumor cells under 40x
	
1e. positive smooth muscle actin under 20x	1d. Ki 67-30-40% (20x)

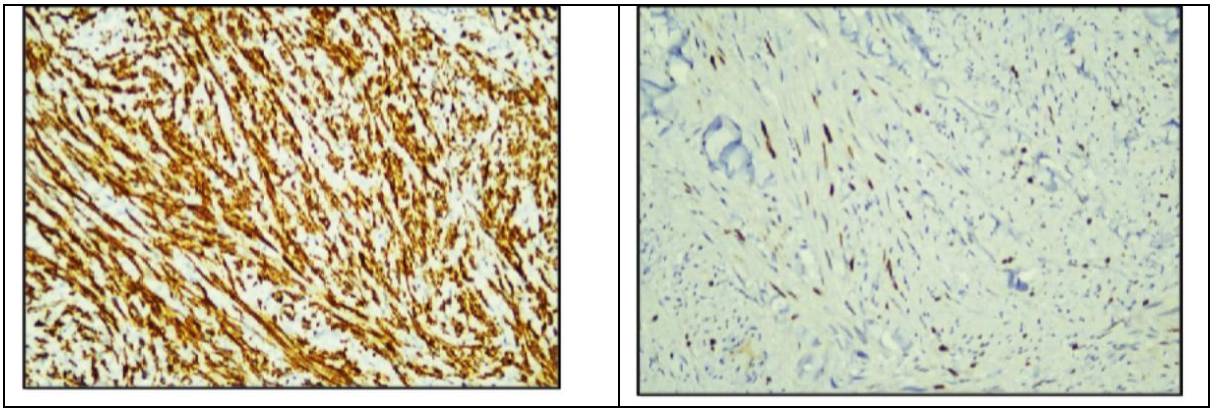


Figure: 1

Pathological findings in CLMS from a real case

Conclusion:

This case illustrates the diagnostic complexity of cutaneous leiomyosarcoma of the breast, particularly when the initial clinical and histological impressions mimic benign conditions. Immunohistochemical analysis is essential for accurate diagnosis. Wide local excision with clear margins is the cornerstone of effective management. Given the potential for recurrence and metastasis, a structured long-term follow-up protocol is crucial. This case reinforces the importance of a high index of suspicion for malignancy in recurrent dermal lesions, even when initially classified as benign.

References:

1. Soares Queirós C, Filipe P, Soares de Almeida L. Cutaneous leiomyosarcoma: a 20-year retrospective study and review of the literature. An Bras Dermatol. 2021 May 1;96(3):278–83.
2. Kohlmeyer J, Steimle-Grauer SA, Hein R. Cutaneous sarcomas. JDDG: Journal der Deutschen Dermatologischen Gesellschaft . 2017 Jun 1;15(6):630–48.
3. Rodríguez-Lomba E, Molina-López I, Parra-Blanco V, Suárez-Fernández R, Pulido-Pérez A. Clinical and Histopathologic Findings of Cutaneous Leiomyosarcoma: Correlation with Prognosis in 12 Patients. Actas Dermosifiliogr. 2018 Mar 1;109(2):140–7.
4. Winchester DS, Hocker TL, Brewer JD, Baum CL, Hochwalt PC, Arpey CJ, et al. Leiomyosarcoma of the skin: Clinical, histopathologic, and prognostic factors that influence outcomes. J Am Acad Dermatol. 2014;71(5):919–25.
5. Walid B, Farah BH, Sbei F, Samar BM, Mohamed Ali S. Frontal cutaneous leiomyosarcoma: A case report and review of the literature. SAGE Open Med Case Rep. 2023 Jan 1; 11:2050313X231197325.
6. Gingrich A, Wangsiricharoen S, Torres MB, Ravi V, Ratan R, Keung EZ, et al. Clinical Variables Influencing Outcomes in Patients with Atypical Intradermal Smooth Muscle Neoplasms (Formerly Cutaneous Leiomyosarcomas): Single-Institution Study of 95 Surgical Patients. Ann Surg Oncol. 2024 Nov 1;31(12):7950–6.
7. Gervais MK, Gladdy RA, Gupta A, Kandel R, Catton C. Breast Sarcoma. Textbook

of Uncommon Cancer: Fifth Edition. 2017 Jan 1;76(2):345–56.

8. Horton L, Wohlfeil M, Al-Kourainy N, Choi L. Rare case of primary leiomyosarcoma of the breast treated with wide local excision and planned cosmetic breast reduction surgery. BMJ Case Rep. 2020 Nov 30;13(11):e236013.

9. Ciurea ME, Georgescu C V., Radu CC, Georgescu CC, Stoica LE. Cutaneous leiomyosarcoma - Case report. J Med Life. 2014 Jun 15;7(2):270–3.

10. Goldsmith LA, Fitzpatrick TB. Fitzpatrick's dermatology in general medicine. 8th ed. New York: McGraw-Hill Medical; 2012. 1 p.

11. Howitt BE, Fletcher CDM. Mammary-type myofibroblastoma: Clinicopathologic characterization in a series of 143 cases. American Journal of Surgical Pathology.2016;40(3):361–7.

12. McMenamin ME, Fletcher CDM. Mammary-type myofibroblastoma of soft tissue: A tumor closely related to spindle cell lipoma. American Journal of Surgical Pathology. 2001;25(8):1022–9.

13. Yoo CC, Pui JC, Torosian MH. Myofibroblastoma associated with bilateral gynecomastia: A case report and literature review. Oncol Rep. 1998;5(3):731–3.

14. Magro G. Mammary myofibroblastoma: a tumor with a wide morphologic spectrum. Arch Pathol Lab Med. 2008 Nov 1;132(11):1813–20

15. Zhang Y, Jorda M, Goldblum JR. Perianal mammary-type myofibroblastoma. Ann Diagn Pathol. 2010 Oct;14(5):358–60.

16. Yajima K, Shirai Y, Fujita N, Sato D, Umezu H, Hatakeyama K. A giant subcutaneous leiomyosarcoma arising in the inguinal region. World J Surg Oncol. 2005 Feb 24 ;3:14.

17. Yang J, Du X, Chen K, Ylipää A, Lazar AJF, Trent J, et al. Genetic Aberrations in Soft Tissue Leiomyosarcoma. Cancer Lett. 2008 Mar 8;275(1):1.

18. Pauwels P, Sciot R, Croiset F, Rutten H, Van Den Berghe H, Dal Cin P. Myofibroblastoma of the breast: Genetic link with spindle cell lipoma. Journal of Pathology. 2000 Jul;191(3):282–5.

19. Magro G, Bisceglia M, Michal M. Expression of steroid hormone receptors, their regulated proteins, and bcl-2 protein in myofibroblastoma of the breast. Histopathology. 2000;36(6):515–21.

20. Morgan MB, Pitha J V. Myofibroblastoma of the breast revisited: An etiologic association with androgens? Hum Pathol. 1998;29(4):347–51.

21. Kraft S, Fletcher CDM. Atypical intradermal smooth muscle neoplasms: Clinicopathologic analysis of 84 cases and a reappraisal of cutaneous "leiomyosarcoma." American Journal of Surgical Pathology. 2011 Apr;35(4):599–607.

22. Hall BJ, Grossmann AH, Webber NP, Ward RA, Tripp SR, Rosenthal HG, et al. Atypical intradermal smooth muscle neoplasms (formerly cutaneous leiomyosarcomas): Case series, immunohistochemical profile and review of the literature. Applied Immunohistochemistry and Molecular Morphology. 2013 Mar;21(2):132–8.

23. Koufopoulos N, Gouloumis AR, Dimas DT, Kontogeorgi A, Sitara K, Boutas I, et al. Mammary Myofibroblastoma: Report of a Case and Review of the Literature. Cureus. 2022 Dec 9 ;14(12).