E-ISSN 2250-0944
ISSN 2250-1150
doi: 10.31838/ijprt/15.02.421

Research Article

Primary Vulvar Leiomyosarcoma Presenting As A Cyst-Like Mass In A 46-Year-Old Woman: A Case Report And Review Of The Literature

Dr. Swagata Karan[1], Dr. Aditi Bhattacharyya[2]
[1]Consultant Pathologist, Rabindranath Tagore International Institute of Cardiac Sciences, Kolkata.
[2]Professor and HOD, Tamralipto Government Medical College and Hospital.
Received: 08.09.25, Revised: 07.10.25, Accepted: 17.11.25

ABSTRACT
Background: Vulvar leiomyosarcoma (VLMS) is the commonest vulvar sarcoma yet represents <1 % of all vulvar malignancies, making evidence-based management challenging. Its ability to masquerade as benign Bartholin-gland cysts or leiomyomas frequently delays definitive treatment.
Case: We describe a 46-year-old multiparous woman who presented with a slowly enlarging, painful swelling of the left vulvo-vaginal vestibule. Baseline laboratory evaluation revealed mild normocytic anaemia (Hb 11.5 g/dL) with an ESR of 62 mm 1st h but otherwise normal liver, renal and coagulation profiles. Pelvic ultrasonography demonstrated a well-circumscribed hypoechoic mass measuring 5.8 × 4.2 × 3.0 cm separate from Bartholin's gland. The lesion was excised with 1-cm macroscopic margins. Grossly the tumour was tan-white, whorled and partly cystic. Histology disclosed intersecting fascicles of spindle cells with moderate to marked nuclear pleomorphism, brisk mitotic activity (12/10 HPF) and focal necrosis. Immunohistochemistry was strongly positive for SMA and desmin and negative for S-100, confirming grade 2 leiomyosarcoma. Surgical margins were histologically free. Adjuvant radiotherapy was deferred after multidisciplinary review because of clear margins and low-intermediate grade. The patient remains disease-free at 9 months.
Conclusion: Early complete excision with histological confirmation is paramount in VLMS because clinical and radiological findings may mimic benign disease. Close surveillance is essential in view of documented late recurrences. This case adds to the limited pool of VLMS reports and highlights contemporary diagnostic and therapeutic considerations.

Keywords: Vulvar Leiomyosarcoma, Soft-Tissue Sarcoma, Vulvar Mass, Immunohistochemistry, Case Report.

INTRODUCTION

Leiomyosarcoma is the most common histological subtype of vulvar sarcomas, yet fewer than 150 well-documented cases have appeared in the world literature, accounting for < 1 % of all primary malignancies of the vulva [1]. The median age at presentation is 44 – 56 years, with reported extremes from adolescence to the eighth decade, and clinical manifestations range from asymptomatic sub-centimetre nodules to rapidly enlarging, ulcerated or fungating masses that may bleed or cause dyspareunia [2]. Because smooth-muscle elements are sparse in the vulvar tissues, proposed cells of origin include the dartos muscle of the labia majora, the erectile tissue of the vestibular bulb, the muscular coats of small- and medium-sized blood vessels, and pluripotent mesenchymal cells within Bartholin-gland stroma [3]. Epidemiologically, VLMS shows no clear association with race or with the typical hormonal and reproductive risk factors implicated in uterine leiomyosarcoma. Rare case series have noted a slightly higher incidence in multiparous women and in those with long-term lichen sclerosus, suggesting chronic inflammatory micro-environments might facilitate malignant transformation. Molecular data remain sparse, but limited next-generation sequencing points to complex karyotypes with copy-number gains on 1q and 13q and homozygous loss of RB1, paralleling soft-tissue leiomyosarcoma at other sites [4].
Over-expression of p16 and aberrant p53 staining patterns—hallmarks of genomic instability—have been documented in up to two-thirds of cases and correlate with higher histological grade and poorer disease-free survival. Diagnostic delay is frequent; up to 60 % of VLMS are initially mislabelled as Bartholin cysts, recurrent abscesses, epidermal inclusion cysts or benign leiomyomas owing to overlapping clinical and radiological characteristics [5]. Ultrasonography typically shows a well-circumscribed, hypoechoic solid lesion but adds little specificity. Magnetic resonance imaging may demonstrate

intermediate-to-high T_2 signal intensity and heterogeneous enhancement; however, benign vulvar leiomyoma can exhibit an almost identical appearance [6]. Distinguishing features such as ill-defined margins, infiltrative growth, necrotic foci > 25 % and diffusion restriction (low ADC values) favour a malignant diagnosis, yet even those criteria lack absolute predictive value. Consequently, histopathological confirmation is mandatory, and the 2020 WHO classification defines VLMS by ≥ 10 mitoses per 10 high-power fields, moderate-to-severe cytological atypia and tumour-cell necrosis [7]. Immunoreactivity for smooth-muscle actin (SMA), desmin and h-caldesmon substantiates smooth-muscle differentiation, whereas negativity for S-100, SOX-10 and cytokeratin excludes neural and epithelial mimics [8]. Current ESGO and NCCN guidelines for vulvar cancer do not provide sarcoma-specific algorithms, compelling clinicians to extrapolate from management principles established for extremity and uterine leiomyosarcoma [9]. Wide local excision with histologically clear margins ("no ink on tumour") is universally advocated, with margin widths of ≥ 1 cm preferred when anatomically feasible.

Sentinel-node biopsy or inguino-femoral lymphadenectomy is controversial because VLMS is thought to metastasise haematogenously rather than via lymphatics; nodal involvement has been documented in < 10 % of reported cases and is usually associated with large, high-grade tumours [10]. The role of adjuvant radiotherapy remains unsettled: retrospective series suggest a reduction in local-recurrence risk for tumours > 5 cm or grade 3 lesions, yet no randomised data exist. Similarly, the benefit of systemic chemotherapy (anthracycline- or gemcitabine-based) is largely extrapolated from non-gynaecologic soft-tissue sarcoma trials and is generally reserved for unresectable, metastatic or recurrent disease. Herein we present a new case of VLMS managed at a tertiary centre in eastern India, detailing its clinico-radiologic presentation, surgical management and pathologic features, and we synthesise contemporary evidence on diagnosis, prognostication and multidisciplinary treatment strategies, thereby contributing to the limited but growing body of knowledge on this rare malignancy.

Case Presentation

A 46-year-old multiparous Bengali woman with no significant past medical history presented with a five-month history of a gradually enlarging, mildly tender swelling in the left vulvo-vaginal vestibule, unaccompanied by pruritus, discharge, constitutional "B" symptoms or menstrual irregularity. Physical examination revealed "a", firm, mobile submucosal mass measuring approximately 6 cm; there was no ulceration or palpable inguino-femoral lymphadenopathy. Baseline investigations demonstrated mild normocytic anaemia (haemoglobin 11.5 g/dL), an elevated erythrocyte-sedimentation rate of 62 mm in the first hour, normal white-cell and platelet counts, and essentially unremarkable hepatic, renal, coagulation and fasting–post-prandial glucose profiles, save for a modestly raised alkaline-phosphatase of 121 U/L; VDRL and HIV serology were non-reactive. High-frequency trans-perineal ultrasonography showed a well-circumscribed, predominantly solid, hypoechoic lesion measuring 5.8 × 4.2 × 3.0 cm, containing focal cystic areas and minimal internal vascularity, lying separate from Bartholin's gland and with no sonographic evidence of pelvic nodal disease or uterine/adnexal abnormality (endometrial thickness 7.6 mm).

The mass was excised via a longitudinal vestibular incision under spinal anaesthesia with a macroscopic 1-cm margin; the encapsulated ovoid specimen (5.5 × 3.9 × 3.2 cm) had a tan-white, whorled cut surface interspersed with myxoid change and haemorrhagic streaks. Histology revealed a partly infiltrated malignant tumour, composed of long intersecting and fascicles of spindle cells. Cells show moderate to marked nuclear pleomorphism having plumped vesicular to hyper chromatic nuclei and pale eosinophilic cytoplasm.Mitotic count 12 /10 HPF. Atypical mitosis present.

Focal tumor necrosis identified; immunohistochemistry was diffusely positive for smooth-muscle actin and desmin, focally positive for h-caldesmon, negative for S-100 and cytokeratin, and showed a Ki-67 proliferation index of ~25 %, establishing a Fédération Nationale des Centres de Lutte Contre le Cancer (FNCLCC) grade 2 vulvar leiomyosarcoma with all circumferential margins free by at least 4 mm. Post-operative recovery was uneventful, and a multidisciplinary sarcoma board recommended close surveillance without adjuvant therapy given the intermediate grade and negative

margins; the patient remains asymptomatic and disease-free nine months after surgery on a regimen of three-monthly clinical examinations and six-monthly imaging.

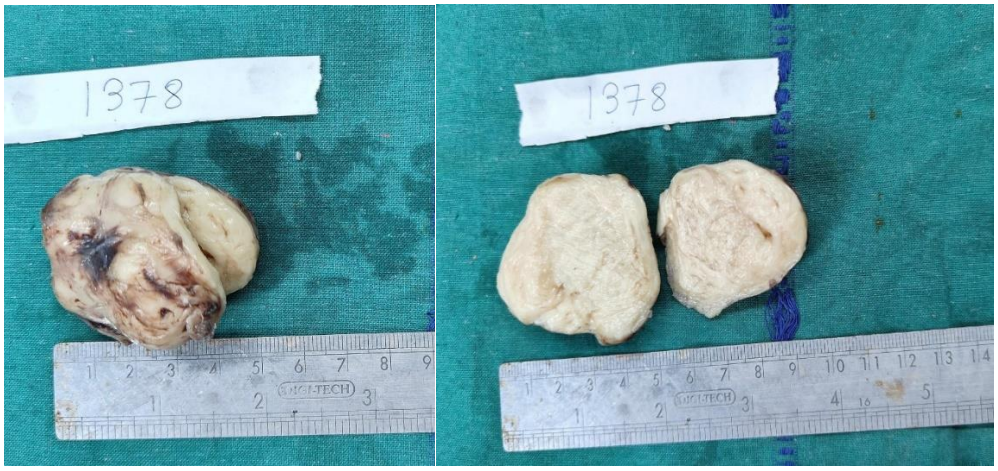
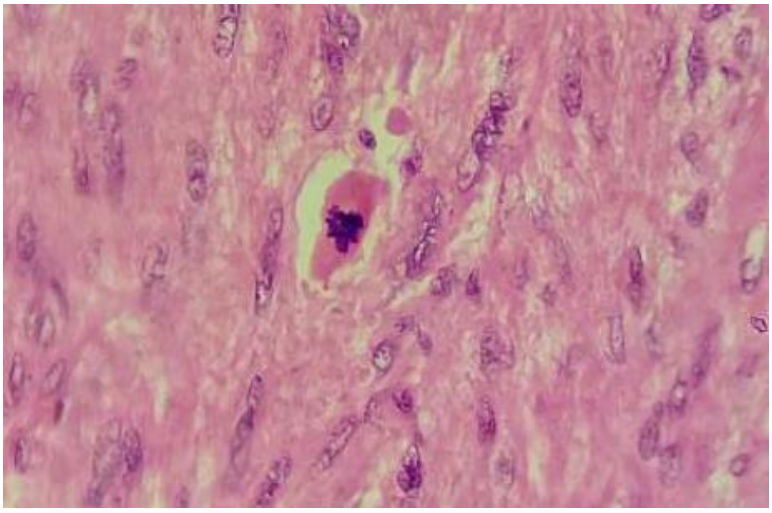


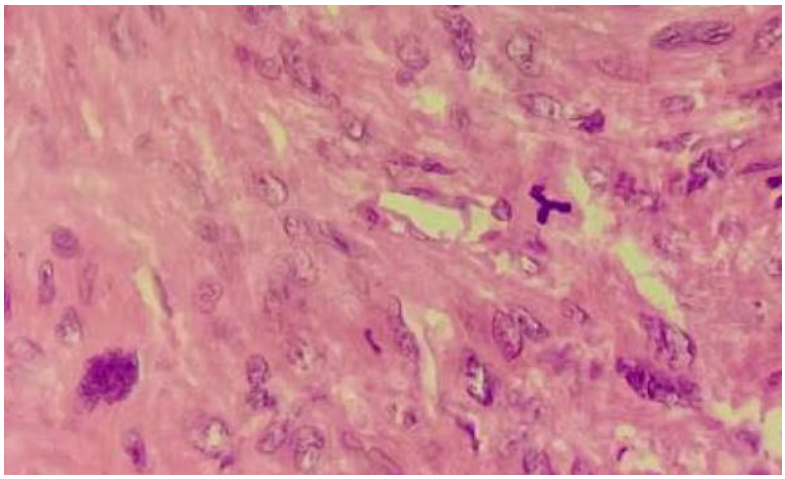
Figure 1: Gross Morphology of Excised Vulvar Leiomyosarcoma Intact and Bisected Views

Picture Procedure



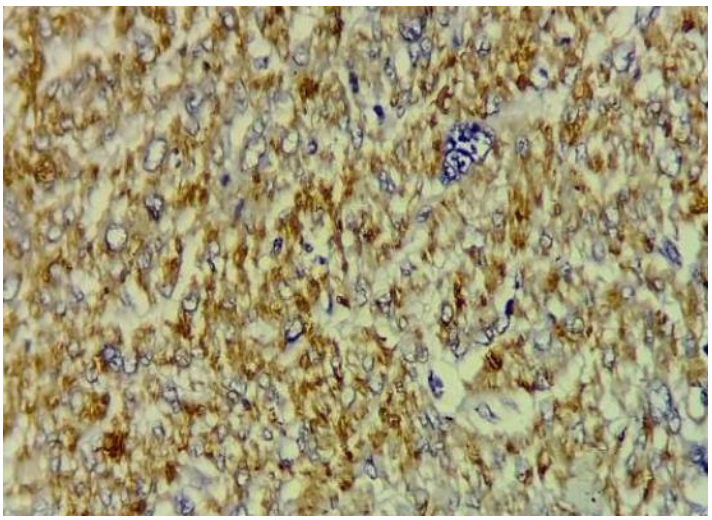
Picture 1: Histopathology of Atypical Mitotic Figure in High-Grade Spindle Cell Neoplasm

High-power H&E section demonstrates spindle cell pleomorphism with atypical mitotic figure, suggesting high-grade malignant potential.



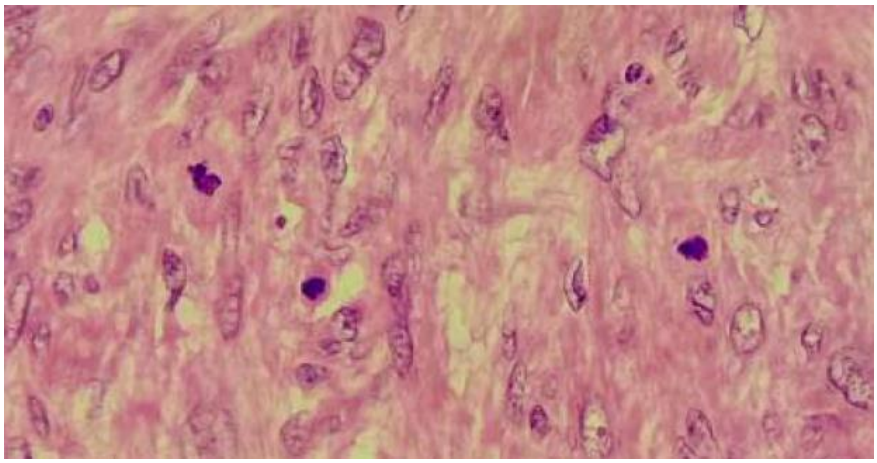
Picture 2: Histological Field Demonstrating Tripolar Atypical Mitosis in High-Grade Spindle Cell Sarcoma

H&E micrograph highlights bizarre tripolar mitosis amid pleomorphic spindle cells, consistent with aggressive sarcomatous proliferation.



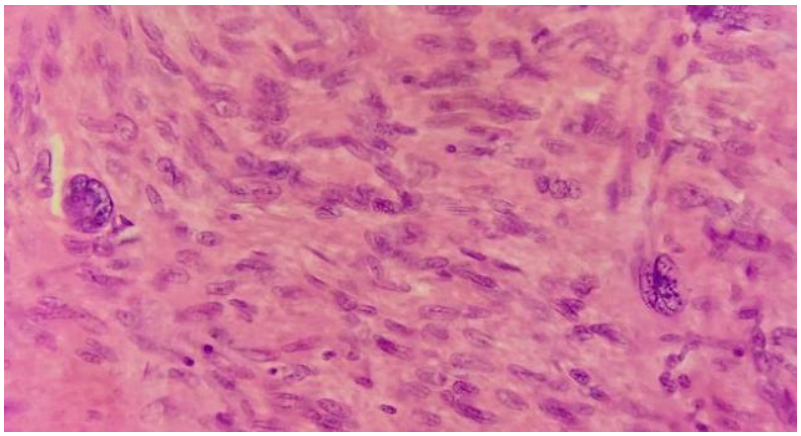
Picture 3: Diffuse Desmin Immunoreactivity in Spindle-Cell Sarcoma Suggesting Myogenic Lineage

Widespread cytoplasmic desmin immunoreactivity indicates robust myogenic differentiation, favouring leiomyosarcoma among spindle-cell malignancies clinically.



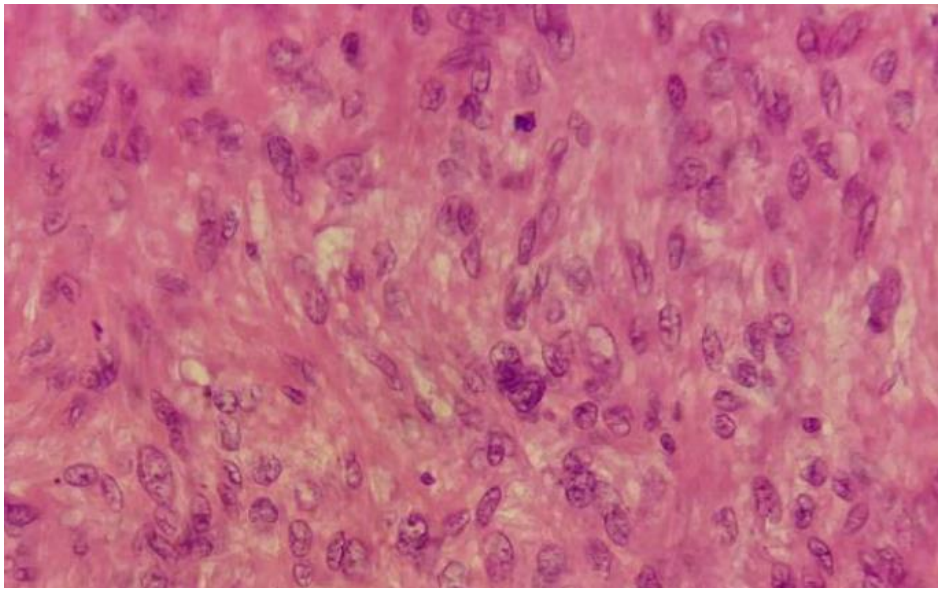
Picture 4: High Mitotic Index within Pleomorphic Spindle Cell Sarcoma

Numerous dark mitotic figures among pleomorphic spindle cells denote elevated proliferation, supporting high-grade malignant sarcoma diagnosis.



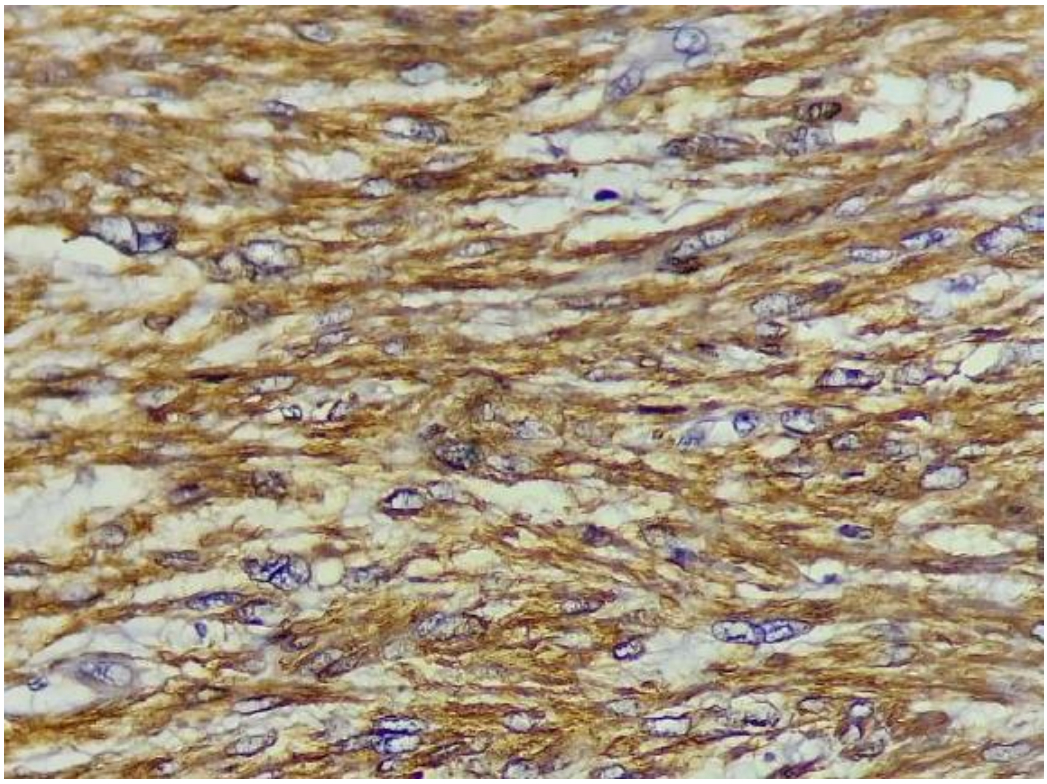
Picture 5: Marked Nuclear Atypia in High-Grade Spindle Cell Neoplasm

Pleomorphic hyperchromatic spindle nuclei with irregular contours signify pronounced atypia, supporting aggressive malignant behavior potential.



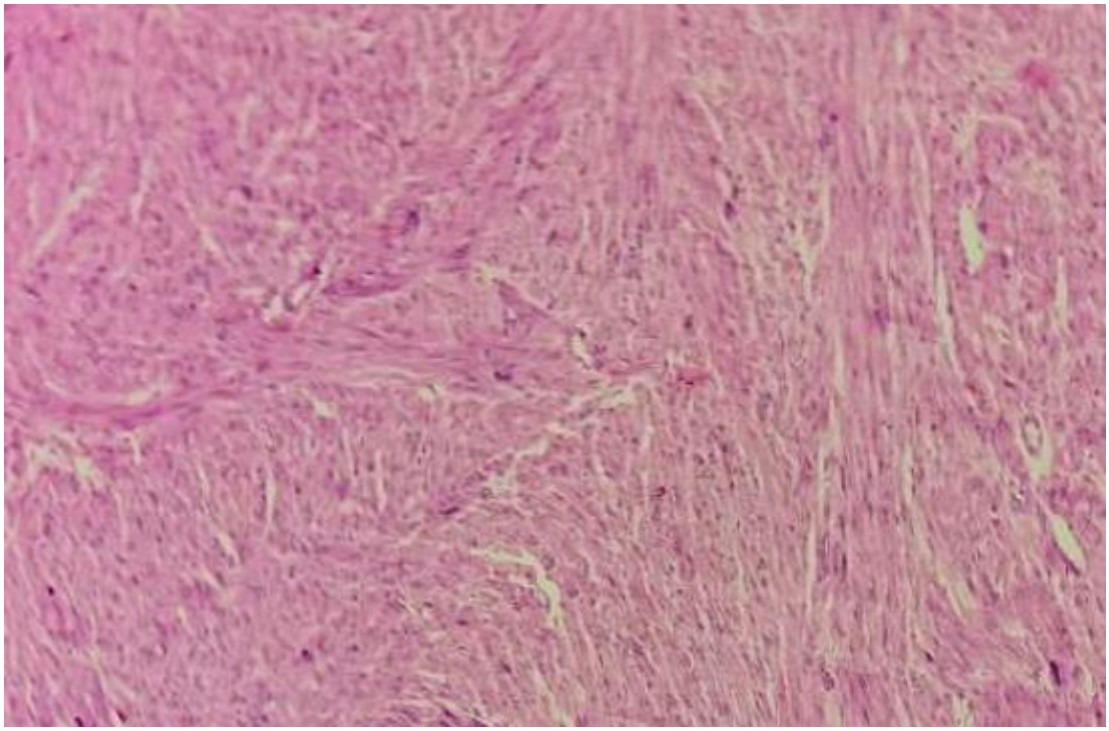
Picture 6: Pronounced Nuclear Pleomorphism In Spindle-Cell Sarcoma Section

Hyperchromatic pleomorphic spindle nuclei populate stroma, indicating severe atypia and high malignant proliferation rate potential.

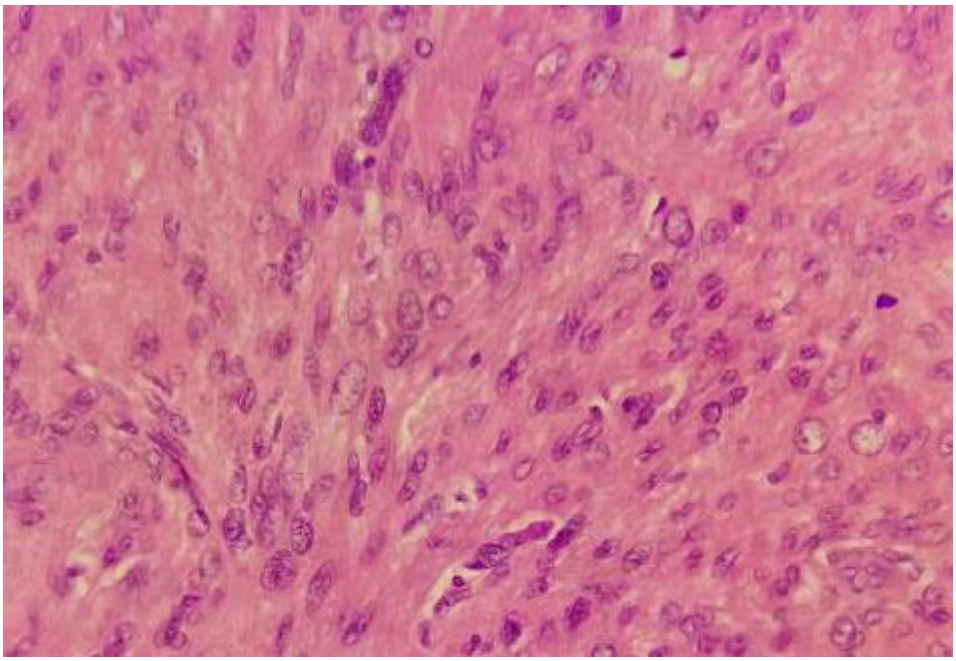


Picture 7: Diffuse Smooth Muscle Actin Immunopositivity in Spindle-Cell Sarcoma

Diffuse cytoplasmic SMA staining highlights interlacing spindle cells, supporting smooth muscle phenotype and tumor origin.



Picture 8: Intersecting Fascicular Pattern of Spindle Cells in Leiomyosarcoma Low-Power View



Picture 9: High-Power Fascicular Spindle Cell Pattern in Leiomyosarcoma Section

High-power H&E shows intersecting spindle fascicles with moderate nuclear atypia, favoring malignant smooth-muscle neoplasm diagnosis.

DISCUSSION

VLMS is an exceptionally rare neoplasm arising from vulvar smooth-muscle derivatives, accounting for 0.2–0.4 % of all malignancies of the female genital tract. Because fewer than 30 single-patient reports have been published in the past decade, each new case contributes valuable clinical experience. The index patient's lesion was clinically indistinguishable from a Bartholin cyst or leiomyoma. Up to 50 % of VLMS are initially misdiagnosed, especially when the tumour is <5 cm and overlying skin remains intact. Ultrasound is widely available and may reveal a solid, vascular mass, but MRI remains superior for delineating deep extension and relation to urethra or clitoris. Definitive diagnosis mandates surgical excision and histology because fine-needle cytology is often

inconclusive in spindle-cell lesions. The 2020 WHO Classification of Female Genital Tumours defines VLMS by the presence of ≥10 mitoses/10 HPF, moderate-severe atypia and tumour necrosis [11]. Ancillary markers (SMA, desmin, h-caldesmon) confirm smooth-muscle differentiation, whereas S-100 negativity excludes peripheral nerve-sheath tumours. Our case fulfilled all criteria and exhibited a Ki-67 index comparable with prior series (20–30 %). Wide local excision with ≥1 cm histological margin is the cornerstone of treatment and yields 5-year disease-specific survival of ~75 %. Radical vulvectomy does not improve survival and increases morbidity; sentinel node biopsy is reserved for sarcomas >10 cm or high-grade tumours with dermal invasion [12]. In the present case, free margins of 4 mm satisfied NCCN "no ink on tumour" guidance for intermediate-grade soft-tissue sarcoma, and nodal evaluation was not pursued in the absence of clinically evident disease [10]. Adjuvant radiation reduces local-recurrence risk in high-grade extremity sarcoma, but data in VLMS are limited to small retrospective series [13-14]. Chemotherapy (doxorubicin ± ifosfamide) is considered for unresectable, metastatic or recurrent disease; response rates rarely exceed 25 %. Given the patient's clear margins and grade 2 status, surveillance alone was deemed appropriate, consistent with ESGO recommendations. Tumour size >10 cm, grade 3 histology and positive margins are independent predictors of recurrence [15]. Local relapse may occur as late as 15 years post-treatment; hence lifelong follow-up with clinical examination every 3–6 months for 2 years, then bi-annually is advised. Our patient's elevated ESR and ALP normalised post-operatively, suggesting that routine laboratory tests may offer inexpensive surveillance adjuncts, although this notion requires validation. This report documents comprehensive clinical, radiological and pathological correlates—including high-resolution gross and microscopic images—of a grade 2 VLMS managed with organ-preserving surgery. Limitations include short follow-up and the absence of MRI. Nonetheless, the case underscores the importance of considering malignancy in any "cystic" vulvar mass persisting beyond six weeks.

CONCLUSION

VLMS, albeit rare, should remain on the differential for any solid vulvar lesion in middle-aged women. Prompt excision with clear margins enables accurate diagnosis and achieves durable local control in most grade 1–2 tumours. Cross-disciplinary collaboration and meticulous long-term surveillance are indispensable to optimise outcomes in this enigmatic disease entity.

REFERENCE

1. International Agency for Research on Cancer (IARC). WHO Classification of Tumours: Female Genital Tumours. 5th ed. Lyon: IARC; 2020.
2. Nielsen GP, Rosenberg AE. Soft-tissue tumours of the vulva. Clin Lab Med. 2013;33(2):371-92.
3. Barroso J, Mendes-Felix A, Simões-Silva L, et al. Misdiagnosed vulvar leiomyosarcoma: case report and literature review. Case Rep Obstet Gynecol. 2012;2012:601854.
4. Laskin WB, Fetsch JF, Tavassoli FA. Vulvar and vaginal smooth-muscle tumors: clinicopathologic analysis of 35 cases. Am J Surg Pathol. 1997;21(8):905-18.
5. Mbatani N, Olawaiye AB, Prat J. Vulvar leiomyosarcoma: a review. Int J Gynecol Cancer. 2022;32(1):124-32.
6. Devereaux KA, McCluggage WG, Walsh CJ, et al. Primary leiomyosarcoma of the vulva. Gynecol Oncol. 1989;33(1):111-4.
7. Hensley ML, Sohaib SA, Uzan C, et al. Imaging of gynecologic sarcomas. Radiographics. 2018;38(4):1119-43.
8. Anderson-Montoya B, Gonzalez-Moran F, Alvarado-Cabrero I, et al. Cytologic challenges in vulvar spindle-cell lesions. Pathol Res Pract. 2015;211(11):835-40.
9. European Society of Gynaecological Oncology (ESGO). ESGO Guidelines for the Management of Vulvar Cancer. Brussels: ESGO; 2024.
10. National Comprehensive Cancer Network (NCCN). NCCN Clinical Practice Guidelines in Oncology: Vulvar Cancer Including Sarcomas. Version 3.2024. Plymouth Meeting (PA): NCCN; 2024.
11. Leitao MM Jr, Zivanovic O, Chi DS, et al. Management of vulvar sarcomas. Int J Gynecol Cancer. 2003;13(4):528-32.
12. Hensley ML, Enserro D, Hatcher H, et al. Uterine and vulvar sarcomas: chemotherapeutic considerations. Oncologist. 2017;22(5):511-20.
13. Jenner BD, Chong S, Duffy S, et al. Long-term survival after wide excision of

vulvar leiomyosarcoma. J Surg Case Rep. 2021;2021(3):rjab058.

14. Ghanim B, Hanzal E, Witt A, et al. Adjuvant radiotherapy in vulvar sarcoma: a single-institution series. Oncol Lett. 2014;8(2):509-14.

15. Caldwell L, Desai P, Wilson G, et al. Patterns of recurrence in vulvar leiomyosarcoma. Gynecol Oncol Rep. 2020;34:100636.