Perineal leiomyosarcoma: Avoid consequences or prevent recurrences?

Debra Nielsen, Cristian Nicolás Lucas, Pablo Catalano

Sanatorio Franchin, Ciudad de Buenos Aires. Argentina

ABSTRACT

Soft tissue leiomyosarcomas (LMS) are malignant tumors that, when located in the perineum, pose additional difficulties because in this region multiple relevant anatomical elements coexist in extreme proximity. It is known that local recurrence and distant metastasis rates reach 37 and 62%, respectively, so in selected cases aggressive surgical approaches with wide margins and adjuvant radiochemotherapy are usually recommended to reduce these percentages. The case of a 42-year-old woman with a right perineal tumor of 9 years of evolution, painful due to increase in size, is presented. On physical examination, she had a mobile lesion not attached to deep planes. Computed tomography with intravenous contrast shows tumor with clear edges, in contact with the lateral wall of the rectum. A marginal resection of the tumor was performed, respecting the integrity of its capsule. The pathological study reported low-grade LMS. After 18 months of follow-up, she has no recurrence. Conservative resections may be appropriate in selected low-risk patients with localized disease. More evidence is needed to confirm these findings.

Keywords: *perianal leiomyosarcoma, perineal sarcoma, perineal tumor*

INTRODUCTION

Soft tissue leiomyosarcomas (LMS) are malignant tumors of muscular origin, which represent 3% of sarcomas and 0.04% of all neoplasms. Due to their low incidence, for their management and staging they are grouped together with a set of other malignant entities of mesenchymal origin. Its prognosis worsens as its location deepens, reaching local and distant recurrence rates of up to 37 and 62%, respectively.[1]
The age of presentation is between the 4th and 5th decades of life. They are usually single, voluminous, painless, slow-growing tumors reminiscent of lipomas but with a harder consistency. Occasionally, they may be associated with itching or sweating.
Given its low incidence, there is no specific TNM for LMS, so the AJCC classification of sarcomas is used.

CASE

A 42-year-old woman consults for a 9-year-old soft tissue tumor in the right ischiorectal fossa. She reports slow and sustained growth, and recent appearance of pain and tenesmus due to increased volume. She has no history of perineal surgery or trauma. The physical examination revealed a hard, mobile tumor, not adhered to deep planes, measuring 7 cm, which displaces the anal orifice and the sphincter apparatus without compromising it macroscopically, and which causes a marked asymmetry in the perineum. (Fig. 1).

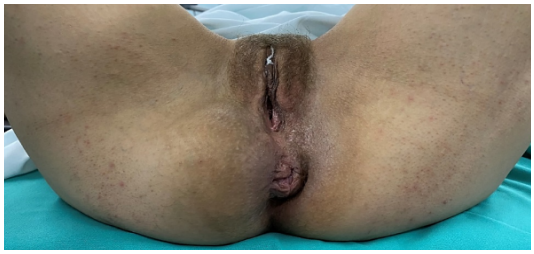

Figure 1. *In the lithotomy position, the tumor is observed in the right ischiorectal fossa.*

Computed tomography with intravenous contrast revealed a solid, homogeneous, hyperdense mass, measuring 8 cm, in the right ischiorectal fossa, in contact with the lateral rectal wall. It has defined edges, enhanced with intravenous contrast. No enlarged lymph nodes are evident and both the liver and lungs are free of pathological images (Fig. 2).
The patient is evaluated by the multi-disciplinary tumor board, considering two alternatives: radical surgery, including abdominoperineal resection, or marginal resection and re-evaluation with the result of the biopsy.
Given that the patient categorically rejects a resection that could cause serious local consequences or compromise her continence, it was jointly decided to perform a marginal resection.
In the lithotomy position, an incision is made over the tumor, which contacts the left lateral wall of the rectum without invading it (Fig. 3). Enucleation is performed respecting the pseudocapsule.
The patient progressed favorably and was discharged after 24 hours. The pathological anatomy reported a low-grade LMS measuring 9 x 8 x 8 cm. Mitotic count up to 5 figures/mm^2, with a cell differentiation score of 1 and Ki67 7%.
As it was a Grade I tumor, it was decided to reserve adjuvant treatment only in case of recurrence. The patient is currently under strict follow-up with MRI of the abdomen and pelvis and thoracic CT quarterly for the first 3 years, in accordance with current recommendations for sarcomas.[2]
There is no evidence of local or distant recurrence after 18 months of follow-up.

The authors declare no conflict of interest.
Received: June 9, 2023. Accepted: May 2, 2024

Debra Nielsen https://orcid.org/0000-00001-8697-5690 Cristian Nicolás Lucas https://orcid.org/0000-0002-4874-3386 Pablo Catalano https://orcid.org/0000-0001-7582-2162

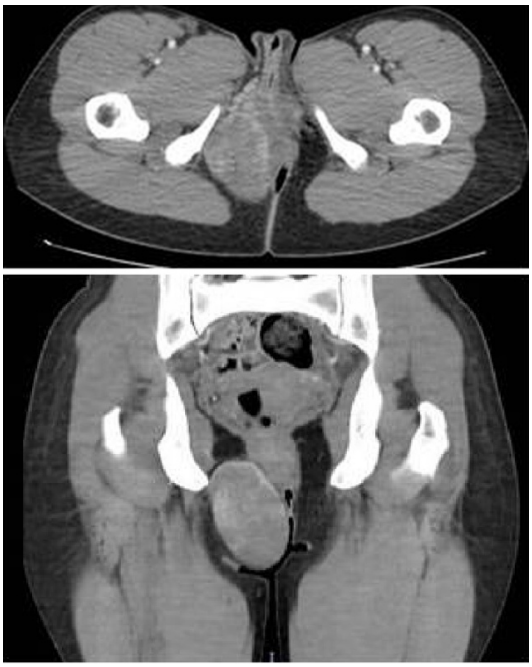

Figure 2. *Computed tomography with intravenous contrast showing the location of the tumor in close contact with the rectal wall and the sphincter complex.*

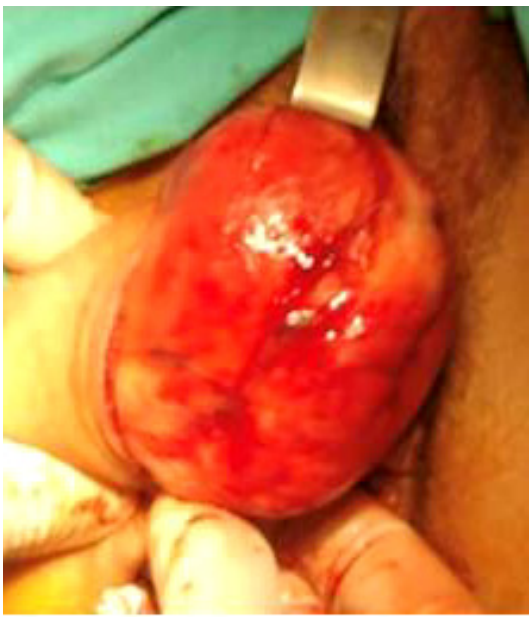

Figure 3*. Enucleation of the tumor with its pseudocapsule intact.*

DISCUSSION

Given the low incidence of perineal LMS, diagnostic suspicion is usually low, often presenting as an unsuspected result in the late pathological study of the tumor. This results in a worse prognosis due to delayed treatment.[3]

According to the initial publications on LMS of the extremities,[4,5] treatment was based on an aggressive surgical approach, with wide resections and minimum free margins of 4 cm.

However, even in patients with R0 resections, local recurrence is evident in up to 37%,[6] which encouraged the study of poor prognostic factors independent of the treatment offered.

According to the ESMO-EURACAN-GENTURIS Clinical Practice Guidelines, updated in 2021,[2] prognostic factors in soft tissue and visceral sarcomas include the size of the lesion, its location, the mitotic index and the presence of necrosis and vascular invasion. On the other hand, the Federation Nationale des Centers de Lutte Contre le Cancer (FNCLCC) classifies soft tissue LMS into three degrees of malignancies according to a score based on cellular differentiation (good, moderate or poor), necrosis (absent, minor or major: >50%) and mitotic count (<10, 10 to 20, >20 figures/mm^2).[7] With a maximum score of 8 points, Grade I adds up to 3 and has 5-year survival rates >95%. Grade II and III add 4 or more points and have a formal indication for multimodal treatment, because local relapse rates rise up to 60%.[8]

In the current treatment of this pathology, wide surgical resections continue to prevail as the standard treatment, guaranteeing lesion-free margins ideally greater than 10 mm.[9]

The role of radiotherapy, although still debated, may be especially relevant when surgical margins are involved or threatened after surgery and re-resection is not possible, particularly in high-grade lesions. Likewise, radiotherapy is useful in the palliative local control of cases with advanced disease and presence of metastases.[6]

Chemotherapy constitutes the fundamental basis of the treatment of metastatic disease. Although it is not curative, it can delay its progression.[10] The most commonly used drugs include doxorubicin, ifosfamide, gemcitabine, taxotere, dacarbazine and trabectedin, in combination regimens.

In both cited guidelines,[2,7] marginal resection as the only treatment, is offered only in carefully selected cases. This is an individualized decision in patients with Grade I tumors with complete pseudocapsule and without distant disease, who can adapt to a strict follow-up in high complexity centers for at least 5 years.

CONCLUSION

Perineal soft tissue LMS are a rare entity, so there is little scientific evidence to indicate the best approach in the treatment of this disease. Although wide resections are the most widely accepted surgery for the treatment of all sarcomas, the perineal location could imply the need for resection of the sphincter, lower rectum and urinary tract, with the need for permanent ostomy, multiple functional impairments and severe impact on the quality of life. The correct selection of patients allows opting for more conservative surgery, with less morbidity but similar oncological safety.

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;96(3):278-83.
2. Gronchi A, Miah AB, Dei Tos AP, Abecassis N, Bajpai J, Bauer S, et al. Soft tissue and visceral sarcomas: ESMO-EURACAN-GENTURIS Clinical Practice Guidelines for diagnosis, treatment and follow-up. *Ann Oncol.* 2021;32(11):1348-65.
3. Rice JP, MacGillivray DC, Sharpe RW, Weiser EB, Ghosh BC. Perineal leiomyosarcoma. *Gynecol Oncol.* 1990;37(1):132-37.
4. Rosenberg SA, Suit HD, Baker LH. Sarcomas of soft tissues. In: DeVita VT Jr,, Hellman S, Rosenberg SA. *DeVita, Hellman, and Rosenberg's Cancer: Principles and Practice of Oncology, 10th ed.* Lippincott: Philadelphia, 1985, pp. 1243-91.
5. Enterline, HT. Histopathology of sarcomas. *Semin Oncol.* 1981;8(2):133-55.
6. Gebhardt MC, Baldini EH, Ryan CW. Overview of multimodality treatment for primary soft tissue sarcoma of the extremities and superficial trunk. *UpToDate.* Version June, 2024.

7. Lin X, Davion S, Bertsch EC, Omar I, Nayar R, Laskin WB. FNCLCC grading of soft tissue sarcomas on needle core biopsies using surrogate markers *Human Pathol.* 2016;56:147-54.
8. Chouliaras K, Patel N, Senehi R, Ethun CG, Poultsides G, Grignol V, et al. Impact of resection margin on outcomes in high-grade soft tissue sarcomas of the extremity-A USSC analysis. *J Surg Oncol.* 2021;123(2):479-88.
9. Starling J 3rd, Coldiron BM. Mohs micrographic surgery for the treatment of cutaneous leiomyosarcoma. *J Am Acad Dermatol.* 2011;64(6):1119-22.
10. Garcia del Muro X, de Alava E, Artigas V, Bague S, Braña A, Cubedo R. Clinical practice guidelines for the diagnosis and treatment of patients with soft tissue sarcoma by the Spanish group for research in sarcomas (GEIS). *Cancer Chemother Pharmacol.* 2016;77(1):133-46.