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LEIOMYOSARCOMA OF THE EPIDIDYMIS: A CASE REPORT

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Abstract: Urological leiomyosarcomas have a poor prognosis due to their aggressiveness and high probability of metastasis, especially via the hematological route. Although it is a rare disease, studies show that the best treatment results are achieved with surgery, radiotherapy and chemotherapy, and recently molecular target therapy has been used. The following case report deals with a 56-year-old patient, with no previous history of disease, who by chance discovered such a neoplasm of the epididymis, confirmed as leiomyosarcoma in immunohistochemistry, and was approached surgically, with total removal of the neoplastic cells, and with no evidence of metastases in the complementary exams.
Keywords: Leiomyosarcoma; Epididymis; Orchiectomy; Urological neoplasms.

INTRODUCTION

Sarcomas of the genitourinary tract are uncommon and account for only 1-2% of all urological malignancies [1,2]. With regard to primary soft tissue tumors in the scrotal region, they are divided into groups according to the structure affected, with the epididymis being found in the paratesticular category [2]. The location of these tumors in the epididymis is rare, accounting for 4% of all paratesticular sarcomas [1]. Epididymal leiomyosarcoma is a malignant tumor originating from the smooth muscle cells of the epididymis [3]. The incidence of this tumor is extremely low, with around 0.3-1.4 cases per million inhabitants per year[4]. It has a higher incidence in men aged 40-60([])([5])].

Paratesticular leiomyosarcoma is known to arise in smooth muscle structures such as the epididymal wall, ductus deferens, cremaster muscle and contractile tissues of the tunica albuginea[6]. The pathophysiology involves genetic mutations that lead to uncontrolled proliferation of smooth muscle cells ([])([7])]. Risk factors include: Exposure to chemical substances [8]. Family history of cancer[9]. Genetic diseases (e.g. Li-Fraumeni syndrome)([])([10])]. Patients present with the following symptoms: scrotal pain, a palpable mass in the epididymis, urethral bleeding and even sexual dysfunction. Anamnesis, physical examination and imaging tests with ultrasound, CT and MRI are important for diagnosis, followed by pathological examination[11,12,13([])]. Treatment can be radical surgery (orchiectomy and epididymectomy), radiotherapy and chemotherapy and recently molecular target therapy in selected cases[14,15].

As paratesticular sarcomas are uncommon, there is limited material on the natural history of the disease or the results of long-term treatment[1]. The aim of this paper is to report a rare case of leiomyosarcoma of the right epididymis in a middle-aged man.

CASE REPORT

S.D.R., a 56-year-old male with no comorbidities and no previous surgeries, came to the Urology outpatient clinic at Santa Casa Clínicas in São Carlos/SP in December 2023 for outpatient screening due to a paternal family history of prostate cancer, as well as a complaint of inguinal and testicular discomfort on the right. Physical examination revealed a cyst in the cranial region of the epididymis on the right, with no other alterations.

An ultrasound of the scrotum showed a nodular area measuring 1.9 x 1.2 cm, which may correspond to a foreign body granuloma. Serum markers (human chorionic gonadotrophin (BHCG), alpha fetoprotein, lactate dehydrogenase (DHL) and prostate-specific antigen (PSA)) were then measured, which showed no alterations. An excisional biopsy was then performed on the nodule, with a result suggestive of malignant fusocellular neoplasia, with compromised margins, on two revisions of the slide, requiring an immunohistochemical study to define the diagnosis.

Antibodies	Clone	Results	Observation
- SF-1	EP434	Negative	
- Cytokeratins of 10.48, 50 and 50.6 kDa	AEI/AE3	Negative	
- Alpha-inhibin	R1	Negative	
- Desmin (muscle cell intermediate filament)	D33	Positive	
- Protein S-100	Polyclonal	Negative	
- Caldesmon	H-CALD	Positive	
- Myogenin, skeletal muscle cell antigen	F5D	Negative	
- MDM2	SMP14	Negative	
- CDK34, cyclin-dependent kinase 4, clone DCS-31	EP180	Negative	
- Hist. H3,3K27me	K27-C36B11	Preserved Expression	
- CD34 - hematopoietic cell and pericyte antigen	QBEnd10	Negative	

Figure 1: Immunohistochemistry showing positive antibodies suggesting smooth muscle differentiation.

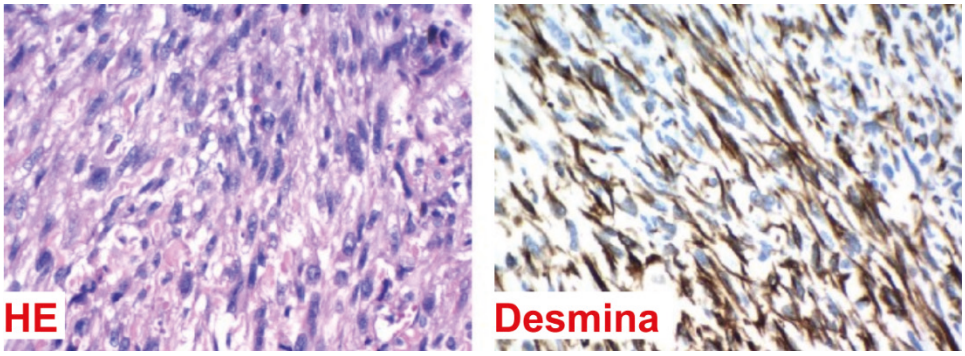

Figure 2: Immunohistochemistry slide positive for desmin and caldesmon.

Subsequently, the immunohistochemistry study (figure 1) indicated a definitive diagnosis of high-grade spindle cell sarcoma (leiomyosarcoma), with positive results for desmin and caldesmon antibodies (figure 2). Desmin is a marker antibody for smooth muscle and striated muscle tumors, while caldesmon is related to endometrial stromal tumors and smooth muscle tumors. Both are filament proteins that indicate smooth muscle differentiation, which, when correlated with the clinical picture, suggest leiomyosarcoma.

In February 2024, a radical right orchiectomy was performed, with no complications. The patient is recovering well after surgery and is being followed up, with no indication for chemotherapy or radiotherapy at the moment. The anatomical specimen of the right testicle showed the margin of the spermatic cord free of neoplasia and the staging tomography showed no alterations.

High-grade sarcomas are characterized by more aggressive tumor biology and a high risk of metastatic spread[16]. In relation to our case, specifically, epididymal sarcoma is rare and the most common is primary soft tissue sarcoma of the epididymis, called leiomyosarcoma[17]. The age group most affected by epididymal leiomyosarcoma (EL) is men, between the sixth and seventh decades of life, but there are differences in the literature[2].

As far as diagnosis is concerned, it is difficult to differentiate (LE) from other scrotal tumors on the basis of physical examination and clinical symptoms alone [(])(18)]. It usually presents as a growing mass which, as it increases in size, presses on nearby structures, causing symptoms such as discomfort or pain in the scrotum and groin[19]. As a result, the rate of misdiagnosis is high[(])(18)]. LE is prone to early hematogenous spread, and the prognosis varies greatly according to the degree of differentiation[18].

Even in the absence of management protocols, ultrasound is the test indicated as an aid to diagnosis, since it represents factors relevant to prognosis and investigation, such as the location, density, vascularity and size of the tumor[20]. These tumours are almost solid masses, with heterogeneous internal structures, echogenicity and hypervascularization[18]. When an ultrasound of the scrotum was carried out on the patient, a 1.9x1.2cm nodular area was detected on the right, with the hypothesis of a foreign body granuloma, leading to the lesion being approached and subsequently biopsied.

Currently, high-grade sarcomas represent a challenge to multimodal oncological treatment, both in terms of preserving organs and limbs and reducing the risk of metastasis and death[17]. According to Helm (1986), pelvic lymph node dissection does not seem justifiable and, as with leiomyosarcomas in general, neither radiotherapy nor chemotherapy is likely to be of any further help[21]. As with the patient above, after radical orchiectomy on the right, there was no indication for post-operative ionizing radiation.

CONCLUSION

As seen in this report, this neoplasm has a rare prevalence, with few cases described in the literature. There is no consensus on the most appropriate course of treatment, which should be individualized and preferably involve a multi-professional team. The patient in question received an early diagnosis, which enabled appropriate treatment and a good prognosis.

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