Alveolar Subtype Rhabdomyosarcoma of the Thenar Region with Axillary Lymph Node Invasion in an Adult: Case Report

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Rabdomiossarcoma do Subtipo Alveolar em Região Tenar com Invasão Linfonodal Axilar em Adulto: Relato de Caso

Rabdomiosarcoma del Subtipo Alveolar en la Región Tenar con Invasión de los Ganglios Linfáticos Axilares en Adulto: Informe de Caso

Aline Graciele Henriques Campos[1]; Estela Cesar Oyama[2]; Maria Eduarda Pardal Simonato[3]; Maikely Bruna Leite[4]; Rafael Gomes Paz[5]; Victor Pereira da Silva[6]; Liara Camila Tusset[7]; Matheus Vieira da Costa[8]

ABSTRACT

Introduction: Rhabdomyosarcoma is a rare subtype of soft tissue malignancy whose cells are similar to myoblasts. It represents 1% of neoplasms in adults, with a poor prognosis and high recurrence rate. In this work, the objective is to report an ultra-rare case of alveolar rhabdomyosarcoma that affected the hand of an adult patient, in addition to conducting an integrative review of similar cases, given that primary cancers in the hand have an incidence of less than 1%. **Case report:** A 33-year-old woman sought clinical care due to the presence of a purplish, non-ulcerated lesion in the thenar region of her left hand, with lymphadenomegaly in the left axilla. Initially, the MRI revealed an expansive formation in the thenar region, in which the incisional biopsy and histochemical examination suggested high-grade alveolar rhabdomyosarcoma. Amputation of the first metacarpal of the left hand and axillary dissection of the affected lymph nodes were performed, in addition to SAIME chemotherapy interspersed with SAVAC every three weeks, and interval radiotherapy for six months. Treatment without complications and currently without recurrences. **Conclusion:** The reported case has an unusual anatomical presentation and age range for rhabdomyosarcoma, and the main aspects for favorable clinical follow-up are early diagnosis and multimodal therapy.
Key words: Rhabdomyosarcoma; Soft Tissue Neoplasms; Adult; Surgical Amputation; Case Report.

RESUMO

Introdução: O rabdomiossarcoma é um subtipo raro de neoplasia maligna de tecidos moles cujas células são semelhantes a mioblastos. Representa 1% das neoplasias em adultos, com mau prognóstico e alta taxa de recidiva. Neste trabalho, objetiva-se relatar um caso ultrarraro de rabdomiossarcoma alveolar que acometeu a mão de uma paciente adulta, além de realizar uma revisão integrativa de casos semelhantes, haja vista que cânceres primários na mão têm incidência inferior a 1%. **Relato do caso:** Mulher, 33 anos, procurou atendimento clínico por presença de lesão arroxeada e não ulcerada em região de tenar da mão esquerda, com linfadenomegalia em axila esquerda. Inicialmente, a ressonância revelou uma formação expansiva na região tenar, na qual a biópsia incisional e o exame-histoquímico sugeriram rabdomiossarcoma alveolar de alto grau. Realizou-se amputação do primeiro metacarpo de mão esquerda e dissecção axilar dos linfonodos comprometidos, além de quimioterapia SAIME intercalada com SAVAC a cada três semanas, e radioterapia de intervalo, por seis meses. Tratamento sem complicações e atualmente sem recidivas. **Conclusão:** O caso relatado é de apresentação anatômica e faixa etária pouco comum para o rabdomiossarcoma, e os principais aspectos para bom seguimento clínico são o diagnóstico precoce e a terapia multimodal.
Palavras-chave: Rabdomiossarcoma; Neoplasias de Tecidos Moles; Adulto; Amputação Cirúrgica; Relato de Caso.

RESUMEN

Introducción: El rabdomiosarcoma es un subtipo raro de neoplasia maligna de tejidos blandos cuyas células son similares a los mioblastos. Representa el 1% de las neoplasias en adultos, con mal pronóstico y alta tasa de recurrencia. En este trabajo el objetivo es reportar un caso extremadamente raro de rabdomiosarcoma alveolar que afectó la mano de un paciente adulto, además de realizar una revisión integradora de casos similares, dado que los cánceres primarios en la mano tienen una incidencia menor del 1%. **Informe del caso:** Mujer de 33 años acudió a atención clínica por la presencia de una lesión violácea no ulcerada en la región tenar de la mano izquierda, con linfadenomegalia en la axila izquierda. Inicialmente, la resonancia magnética reveló una formación expansiva en la región tenar, en el cual la biopsia incisional y el examen histoquímico sugirieron rabdomiosarcoma alveolar de alto grado. Se realizó amputación del primer metacarpo de la mano izquierda y disección axilar de los ganglios afectados, además de quimioterapia SAIME intercalada con SAVAC cada tres semanas y radioterapia interválica durante seis meses. Tratamiento sin complicaciones y actualmente sin recidivas. **Conclusión:** El caso reportado tiene una presentación anatómica y rango de edad inusual para rabdomiosarcoma, y los principales aspectos para un buen seguimiento clínico son el diagnóstico temprano y la terapia multimodal.
Palabras clave: Rabdomiosarcoma; Neoplasias de los Tejidos Blandos; Adulto; Amputación Quirúrgica; Informe de Caso.

[1-6,8]Universidade Estadual do Oeste do Paraná (Unioeste-PR). Francisco Beltrão (PR), Brasil. E-mails: alineghcampos3@gmail.com; estela.oyama19@gmail.com; mariaeduarda.psh@gmail.com; maikelyleite20@gmail.com; rafaelgomespaz@hotmail.com; silvapereiravictor@hotmail.com; matpatologia@yahoo.com.br. Orcid iD: https://orcid.org/0000-0002-3784-7569; Orcid iD: https://orcid.org/0009-0001-8387-1850; Orcid iD: https://orcid.org/0009-0003-7244-0750; Orcid iD: https://orcid.org/0000-0003-0023-5144; Orcid iD: https://orcid.org/0009-0003-3666-2399; Orcid iD: https://orcid.org/0009-0005-9907-584X; Orcid iD: https://orcid.org/0009-0009-4055-0705

[7]Centro Universitário de Pato Branco. Pato Branco (PR), Brasil. E-mail: liara.oncologia@gmail.com. Orcid iD: https://orcid.org/0009-0002-7904-6046

Corresponding author: Aline Graciele Henriques Campos. Rua Bahia, 835, apto. 14 – Presidente Kennedy. Francisco Beltrão (PR), Brasil. CEP 85605-270. E-mail: alineghcampos3@gmail.com

INTRODUCTION

Rhabdomyosarcoma (RMS) is a rare malignant tumor that evolves from mesenchymal cells that differentiate into skeletal muscle[1]. The disease is locally invasive, requiring a multimodal therapeutic approach. The prognosis varies according to the origin site, tumor size, clinical staging, patient age, and histological type[2,3]. Based on clinical pathological characteristics and genetic abnormalities, it can be divided into subtypes: embryonal (variation from primitive rounded to densely eosinophilic cells), alveolar (layers of medium-sized cells, scattered giant cells, and alveolar pattern formation), spindle or sclerosing (spindle cells in fascicles, with pseudovascular sclerosing pattern) and pleomorphic (atypical rhabdomyoblasts)[1,2,4]. Rhabdomyosarcoma represents 5% of all childhood cancers, with the most common subtypes being embryonal and alveolar[5], and 1% of adult neoplasms, for which pleomorphic is the most common variant[5]. The incidence of alveolar rhabdomyosarcoma (RMSa) is constant from 0 to 19 years old, with approximately one case per million children and adolescents, and presents lower incidence after that age range. In addition, primary malignant tumors that affect the hand are extremely rare, with an incidence rate lower than 1%[6].

Thus, given the exposed data, the objective of this study is to report an ultra-rare case of RMSa that afflicted the hand of an adult patient, discussing treatment, factors for a worse prognosis, and outpatient follow-up to establish a parallel between how the patient was approached and what is described in the literature. This study will also perform an integrative review of case reports of primary RMS in adult extremities. This study has been approved by the Research Ethics Committee of the *Universidade Estadual do Oeste do Paraná*, report number 7050791 (CAAE (submission for ethical review): 82359624.7.0000.0107), according to Resolution no. 466/12[7] of the National Health Council (CNS).

CASE REPORT

Female patient, 33 years old, from the town of Pato Branco in the State of Paraná (PR), married, two children, history of hypothyroidism and gestational diabetes mellitus, making continuous use of levothyroxine sodium 75 mcg. Previous surgeries: C-section and conjunctival tumor excision. No smoking or alcohol drinking, pathological family history of leukemia (grandmother).

Sought outpatient clinical care in May 2021 due to a purplish, non-ulcerated lesion in the thenar region of her left hand, measuring around 3.5 cm, associated with lymphadenomegaly in the left axilla. A magnetic resonance imaging test (MRI) was performed on the left hand, showing an expansive formation on the thenar region measuring 3.3 x 3.0 x 2.2 cm. An incisional biopsy showed a poorly differentiated neoplasm, and the immunohistochemical test suggested high-grade alveolar RMS. Staging was conducted with chest and total abdomen tomographies, which showed only the lymphadenomegaly on the left axilla identified on the previous physical exam, measuring 3.3 x 2.0 cm, thus resulting in clinical staging III cT1aN1M0 of intermediate risk.

An up-front surgery was performed in May 2023 with amputation of the first left-hand metacarpus and left axillary lymphadenectomy, whose anatomopathological test, in Figure 1, revealed a poorly differentiated RMSa, measuring 3.7 cm, non-infiltrative to the bone tissue and with free margins, with 18 positive lymph nodes out of a total of 43. The postoperative period had no complications, and the total period of hospitalization was two days. The patient went through the SAIME (etoposide + ifosfamide + mesna) chemotherapy protocol, alternated with SAVAC (vincristine + doxorubicin + cyclophosphamide) every three weeks, with interval radiotherapy, for six months. The patient shows no recurrence at this point.

DISCUSSION

RMS of extremities constitutes a minority of reported cases in the literature – less than one-third of sarcomas – and usually presents as a disseminated disease upon diagnosis, with a worse prognosis when compared to RMS in other sites[8,9]. The risk factors for the disease are genetic, like the presence of Li-Fraumeni syndrome, neurofibromatosis type 1, Costello syndrome, and

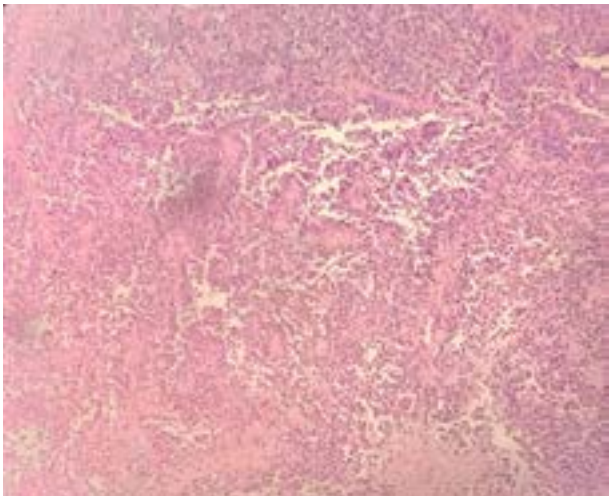

Figure 1. Anatomopathological test showing poorly differentiated alveolar rhabdomyosarcoma

Noonan syndrome; and environmental, like prenatal exposure to X-rays, parental drug use, childhood allergies, use of fertility medication, and premature birth[8]. The clinical presentation is associated with the emergence of soft tissue masses, which are frequently painless; compressive symptoms may also arise. The diagnosis is done through incisional, excisional, or core needle biopsy, followed by histological and molecular analysis. RMSa is composed by densely compacted round cells that coat septations similar to pulmonary alveoli[8]. These characteristics are present in the biopsy of the reported patient, which, according to Figure 1, presents big cells ranging from round to polygonal with defined borders, and slight variation in the individual size of tumoral cells, with abundant eosinophilic granular cytoplasm, round and vesicular nucleus with prominent nucleolus growing in nests or organoids.

Treatment for adults is similar to pediatric therapy, with multimodal approach through surgical resection, chemotherapy for primary cytoreduction and eradication of metastatic disease, and ionizing radiotherapy to control microscopic residual disease according to staging, pathological and molecular variety. This multimodal therapy presents more clinical efficacy, with lower treatment toxicity[10]. This patient did not undergo tumor molecular analysis, but the use of two adjuvant chemotherapy protocols and radiotherapy potentially overcomes the tumor's treatment resistance mechanisms since the therapies act in different cell signaling routes to increase clinical efficacy and decrease side effects[11]. This aggressive and coordinated treatment proved effective since there are no signs of recurrence after over six months of treatment.

RMSa in adults presents a worse prognosis for the alveolar histology or when the anatomical location is disadvantageous, like extremities, such as in the reported case. Other poor prognoses factors are advanced age, female sex, presence of distant metastases at diagnosis, size

Table 1. Case reports of primary rhabdomyosarcoma in body extremities in patients over 18 years old, published up to January 2025

Author	Sex/ Age (years)	Number of cases	Location of primary rhabdomyosarcoma	Size of sarcoma (cm)	Histological subtype	Metastases	Treatment	Follow-up (months)	Clinical Result
12	M/60	1	Right thigh	4.5	Pleomorphic	Lung	S + CT + RT	30	Remission
13	M/30	1	Left foot	11	Sclerosing	-	S	28	Remission
14	F/33*	2	Right thigh/Right triceps	6 x 4 x 3 / 6 x 6.5 x 4	Alveolar	-	S + CT + RT / CT + RT	-	Remission
15	M/37	1	Right elbow	9 x 6	Sclerosing	Testicle, mediastinal, lung and thigh	S + CT	19	Death
16	F/44	1	Right ankle	11	Sclerosing	-	CT + S + RT	24	Remission
17	M/36	1	Left leg	4 × 3 × 2.8	Sclerosing	-	S + CT + RT	9	Remission
18	F/33*	2	Right thigh/Right triceps	6 × 4 × 3 / 6 × 6.5 × 4	Alveolar	-	S + CT + RT	9*	Remission
19	M/31	1	Right calf	-	Pleomorphic	-	CT + S	-	-
20	M/33	1	Anterior compartment of leg	12 x 10 x 25	Embryonal	-	CT + RT + S	48	Remission
21	M/18	1	Left forearm	6.7 x 6.9 x 5.9	-	-	CT + S	-	-
22	M/30	1	Left foot	11	Sclerosing	-	-	-	-
23	F/18	1	Foot	-	Alveolar	Iris, lower limb, external inguinal chain, mediastinal, lung	S + CT	26	Death
24	M/36	1	Left leg	4 x 3 x 2.8	Sclerosing	-	CT + S + RT	-	Remission

Captions: * = mean; F = female sex; M = male sex; RT = radiotherapy; S = surgery; CT = chemotherapy.

of primary tumor larger than 5 cm at localized disease; all associated with adults[10]. In the reported case, the 3.7 cm primary tumor and its location on the left hand, although less common, were associated with a significant lymph node involvement, which reinforces the importance of aggressive multimodal therapy.

Table 1[12-24] presents reports and a series of cases of primary RMS in extremities, in patients over 18 years old, retrieved from PubMed.gov, fully available for free. The search strategy used the following descriptors and Boolean markers: *rhabdomyosarcoma AND hand OR foot OR extremity*.

CONCLUSION

The reported case has an unusual anatomical presentation and age range for RMSa, and the main aspects for favorable clinical follow-up are early diagnosis and multimodal therapy.

CONTRIBUTIONS

All the authors have substantially contributed to the study design, acquisition, analysis and interpretation of the data, wording, and critical review. They approved the final version for publication.

DECLARATION OF CONFLICT OF INTERESTS

There is no conflict of interest to declare.

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