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ALVEOLAR RHABDOMYOSARCOMA: PRESENTATION OF A CASE AND REVIEW OF THE LITERATURE

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ABSTRACT Rhabdomyosarcoma (RMS) is a rare type of soft tissue malignant neoplasm that currently represents about 7% of cancers in children, it is found in a much smaller proportion in adults, which represents about 1% of cancers, the global incidence of RMS is 4.5/million people under 20 years of age. Alveolar rhabdomyosarcoma (ARMS) is a widely studied subtype in minors and very rarely in adults. The most common presentation in extremities is of the alveolar subtype, most frequently affecting the lower extremities.

We present a narrative review based on a case report of a patient with alveolar rhabdomyosarcoma in a lower extremity.

KEYWORDS : Soft tissue tumors, Alveolar rhabdomyosarcoma, Embryonal rhabdomyosarcoma.

INTRODUCTION

Rhabdomyosarcoma (RMS) is a rare type of soft tissue malignant neoplasm that currently represents about 7% of cancers in children, it is found in a much smaller proportion in adults, which represents about 1% of cancers (1), the global incidence of RMS is 4.5/million people under 20 years of age. RMS can classically be found in three subtypes: alveolar rhabdomyosarcoma (RMSA), embryonal (RMSE) and pleomorphic rhabdomyosarcoma (RMSP). Histologic subtypes have differences in diagnosis, treatment, and prognosis. Different behaviors have been found depending on the age group, RMSA is much more frequent in adolescents between 15 and 19 years of age, as well as the RMSE variety is more frequent in those under 15 years of age (2), with a higher incidence in men with a 1.4:1 ratio (3).

Case report

Patient 35-year-old without prior personal history. He was admitted due to clinical symptoms of 2 months of evolution of nocturnal pain in the lower left limb, predominantly in the leg, with progressive increase in diameter with swelling and limitation for walking due to progressive and consistent pain. Physical examination shows an increase in the diameter of the left leg and thigh with respect to the contralateral. An X-ray of the leg is taken, showing soft tissue injuries that probably compromise the fibula (**Figure 1).** A magnetic resonance study was performed, finding alteration in soft tissues without bone involvement (**Figure 2**). A biopsy is performed, finding uniform round cells with poor differentiation, with immunoh istochemical studies confirming an alveolar rhabdom yosarcoma. The patient's treatment was surgical combined with chemotherapy with Vincristine, Actinomycin and Cyclophosphamide for 4 weeks. Patient without relapse at 12 weeks of follow-up.

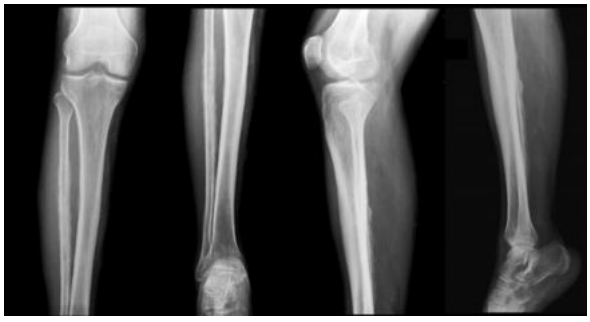

Figure 1. X-ray of the leg. Soft tissue lesions surrounding the fibula without osteolytic involvement.

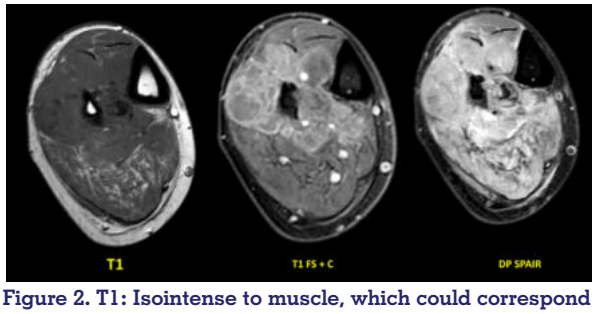

Figure 2. T1: Isointense to muscle, which could correspond to hemorrhage in Alveolar and Pleomorphic subtypes with flow voids with marked enhancement on T1C: Marked enhancement and restriction on DP SPAIR.

DISCUSSION

RMS is common in childhood, accounting for about 50% of soft

tissue neoplasms in this population, (3) about 250 cases are diagnosed each year in the United States, with an increase in recent years, with an incidence of up to 4.2% of the RMSA (1).

Alveolar rhabdomyosarcoma (ARMS) is a widely studied subtype in minors and very rarely in adults, mainly this subtype is described in different series in which a greater presentation was found at the time of diagnosis in extremities representing up to 50 % of the cases of ARMS (4), and the other sites of presentation occur in a smaller proportion in the trunk, pelvis, orbit, parameningeal sites. This is highly relevant to the study since the alveolar subtype is much more aggressive and prone to relapse, with results that imply a greater impact on mortality and treatment failure (5).

The most common presentation in extremities is of the alveolar subtype, most frequently affecting the lower extremities. RMSA classically presents as a palpable mass that could be associated with edema, and in some cases could cause disability and gait disturbances, as occurred in our patient. RMSA could also affect the trunk, chest wall, paravertebral muscles, and abdominal wall with a palpable mass sensation in most cases, in these locations a higher incidence is described in girls with an average age range of 12.5 years (3).

The approach is multidisciplinary for an early diagnosis and to improve the prognosis of the patient, based on the physical examination, on the presentation of a painless mass that should be taken to an early histopathological study (6).

In alveolar-type rhabdomyosarcoma pathology, uniform rounded cells with poor differentiation are observed in a growth pattern that has been classically described as alveolar when referring to diffuse sheets with multinucleated giant cells, sometimes with a crown-shaped pattern (7).

Characteristics in the sample that the pathologist will look for a diagnosis will be the presence of eosinophilic rhabdomyoblasts which are small and scarce in relation to multinucleated giant cells; however, these histological patterns often require immunohistochemistry for desmin and myogenin to differentiate both from the RMSE subtype and to distinguish RMSA from other malignant neoplasms such as Ewing's lymphoma or sarcoma (8). Positive nuclear staining for myogenin is described in up to 75% for the diagnosis of RMSA-type tumor cells. In the alveolar subtype, two translocations that are considered specific predominate, one is (2;8) that would work the PAX3 and FKHR genes and (1;9) for the PAX7 and FKHR genes, this is important to the extent that the detection of these transcripts by molecular methods such as RT PCR has become increasingly used based on the studies of Edwards (8).

Magnetic resonance imaging (MRI) is the ideal test for the evaluation of the primary site in most patients with rhabdomyosarcoma, it has a better diagnostic performance since it will help me characterize the borders and the degree of extension (7). Tomography is of great diagnostic utility due to its ability to evaluate head and neck tumors, given a superior evaluation of both bone and bone marrow by means of tomography and the ability to evaluate soft tissues much better by MRI. In adults, primary rhabdomyosarcoma is like other types of soft tissue neoplasms in its initial presentation, as a large one that, in relation to skeletal muscle, captures a lower signal on T1, on the contrary, a high intensity signal on T2 slices that would enhance contrast heterogeneously, an important difference in rhabdomyosarcomas is that their distant involvement could involve both regional lymph nodes and distant lymph nodes, a behavior similar to what is seen in lymphomas and epithelial neoplasms (9). RMSA present as large tumors with areas of necrosis at the lobulated edges with lymph vascular involvement in its extension, in T2 a hyperintense image with heterogeneous enhancement is seen that is better visualized in T1 slices.

In the last decade, different reviews and clinical trials have emerged for the management of RMSA, which has led to recommendations that have impacted survival, especially in pediatric patients. It is said that 5-year survival is greater than 70% in this group of age, the most important factors that have been shown to impact survival are the use of goal-guided multifocal therapies with multidisciplinary groups, including surgical resection and ionizing radiation of the primary tumor, as well as multi-agent chemotherapy (1).

Multiple tools have been developed to assess the staging of the disease to guide therapy depending on the risk, the different trials described by various research groups on malignant soft tissue neoplasms have focused their efforts on creating protocols, therapies and evaluating the impact on morbidity and mortality. The cure of primary ARMS includes surgical eradication as well as the use of ionizing radiation, and in groups where the disease is disseminated, systemic chemotherapy is recommended, because metastasis has been described in most children (10).

The treatment of these patients is governed by the stage of the disease to define a treatment, which can be treatment with chemotherapy based on Vincristine, Actinomycin D (VA) and additionally Cyclophosphamide known as VAC therapy. Another scheme is carried out with Cisplatin and Doxorubicin, which can receive local radiotherapy known as the 41.4 Gray (GY) protocol, administered in sessions of 1.8 GY for 4 to 5 weeks (11).

There are different analyzes that have been done and various reviews that seek to glimpse survival in ARMS, data have been found showing that survival is greater in children than in adults (12) where age is the strongest predictor at the time of diagnosis to predict both therapeutic outcomes and 5-year survival, and this is associated with the stage based on the extent of the disease. The 8-year survival rate reported for stage 1, 2, and 3 is 86%, 92%, and 70%, respectively (13).

It has been described those recurrences have a negative impact, significantly increasing mortality, with 5-year survival rates described between 17 and 49%, factors associated with a relapse have been demonstrated depending on the presence or absence of metastases, the use or absence of radiotherapy and the age (14).

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