Primary cutaneous alveolar rhabdomyosarcoma in a pediatric patient *

Rabdomiossarcoma alveolar cutâneo primário em paciente pediátrico

Livia Lima de Lima [1]
Priscilla Maria Rodrigues Pereira [3]
Walquíria Lima Tupinambá [1]
Carlos Alberto Chirano Rodrigues [2]
Antônio Pedro Mendes Schettini [4]

Abstract: Rhabdomyosarcoma is the most common soft tissue tumor in childhood; however, it rarely affects only the skin. This case report describes a child with a painful nodule on her face. Histopathology and immunohistochemistry confirmed the diagnosis of rhabdomyosarcoma, and a multidisciplinary team then followed up the patient. Soft tissue tumors are responsible for 6% of all childhood tumors, and 53% of these are rhabdomyosarcomas, which may affect any part of the body. Presentation in the form of skin nodules is rare and represents a diagnostic challenge, since there are no clinical characteristics that differentiate this condition from other pathologies.
Keywords: Head and neck neoplasms; Pediatrics; Rhabdomyosarcoma, Alveolar; Skin neoplasms

Resumo: O rabdomiossarcoma é o tumor de partes moles mais comum na infância, sendo raro o acometimento exclusivamente cutâneo. Apresenta-se caso de criança com nódulo doloroso na face, cuja análise histopatológica e imunoistoquímica confirmou tratar-se de rabdomiossarcoma, o qual foi conduzido por equipe multidisciplinar. Os tumores de partes moles são responsáveis por 6% de todos os tumores infantis; destes, 53% são rabdomiossarcomas, que podem acometer qualquer sítio. A manifestação como nódulo dérmico é incomum, representando um desafio diagnóstico, já que não possui características clínicas que o diferenciem de outras patologias.
Palavras-chave: Neoplasias cutâneas; Neoplasias de cabeça e pescoço; Pediatria; Rabdomiossarcoma alveolar

INTRODUCTION

Rhabdomyosarcoma is a malignant tumor that originates in the primitive mesenchymal cells, precursors of the striated skeletal muscle. The sites most affected by this tumor are the head and neck (35%), the genitourinary system and the extremities (40%) and, less commonly, the trunk, orbital cavity, intrathoracic and retroperitoneal regions. Only sixteen cases of the exclusively cutaneous form have been described in the literature. [1-3]

CASE REPORT

A 2-year old female patient presented with a painful lesion in the left paranasal region that had appeared at four months of age and was progressively increasing in size. No systemic symptoms such as fever or weight loss were reported. General physical examination was normal and there was no sign of lymphadenopathies. Dermatological examination revealed the presence of an erythematous, lobulated, firm, hard nodule measuring 3.2 x 2.4 cm, located in the left paranasal region and with telangiectasias on its surface (Figures 1 and 2).

Following biopsy, the histopathology report revealed a normal epidermis with dermal infiltrate consisting of small, round, blue cells with clear cytoplasm. These cells formed layers with fibrous septa

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[1] Physician currently participating in the residency program in Dermatology at the Alfredo da Matta Foundation of Tropical Dermatology and Venereology, Manaus, Amazonas, Brazil.
[2] Dermatologist certified by the Brazilian Society of Dermatology. Dermatologist, Alfredo da Matta Foundation of Tropical Dermatology and Venereology, Manaus, Amazonas, Brazil. Preceptor of the Medical Residency Program in Dermatology, Alfredo da Matta Foundation of Tropical Dermatology and Venereology, Manaus, Amazonas, Brazil.
[3] Physician currently participating in the residency program in Dermatology at the Getúlio Vargas Teaching Hospital, Manaus, Amazonas, Brazil.
[4] Master's degree in Tropical Pathology awarded by the Federal University of Amazonas. Dermatologist at the Alfredo da Matta Foundation of Tropical Dermatology and Venereology, Manaus, Amazonas, Brazil. Preceptor of the Medical Residency Program in Dermatology, Alfredo da Matta Foundation of Tropical Dermatology and Venereology, Manaus, Amazonas, Brazil.

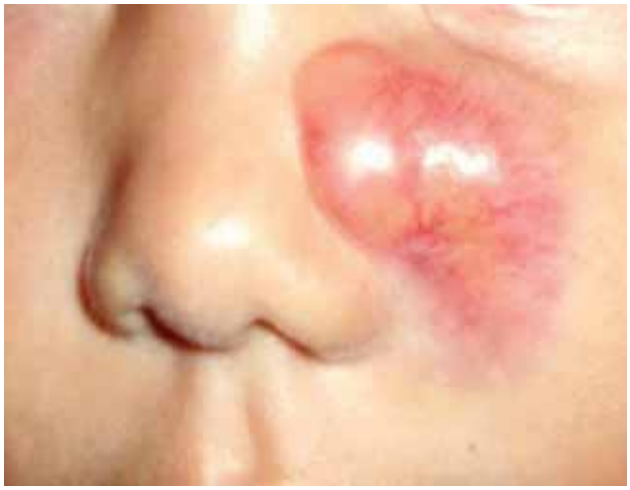

Figure 1: Erythematous, hardened, well-defined nodule measuring 3.2 x 2.4 cm on the left paranasal region

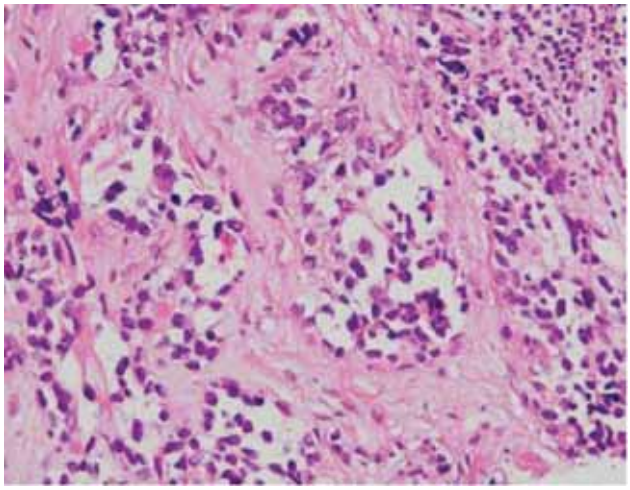

Figure 3: Dermal infiltrate composed of small, round, blue cells with clear cytoplasm, forming layers resembling pulmonary alveoli

resembling pulmonary alveoli (Figure 3). Immunohistochemistry was positive for desmin and myogenin, confirming a diagnosis of alveolar rhabdomyosarcoma (Figures 4 and 5).

Biochemical exams were performed in addition to bone scintigraphy and computed tomography of the chest, head and abdomen. All results were normal. Tomography of the paranasal sinuses showed a subcutaneous mass with well-defined contours and margins above the left maxillary sinus, close to the nasal fold. There was no bone invasion and the mass measured 2.3 x 1.5 cm.

Following investigation for metastases, the patient was staged as IA in accordance with the Intergroup Rhabdomyosarcoma Study Clinical Grouping System and was submitted to complete surgical resection of the lesion with reconstruction of the defect using a frontal flap. The patient is currently being submitted to adjuvant chemotherapy. Histopathology performed on the surgical specimen confirmed the same clinical staging.

DISCUSSION

Tumors originating in the soft tissues represent only 6% of all pediatric malignancies and 53% of these are rhabdomyosarcomas, which constitute 3.5% of all cancers in children under 14 years of age and 2% of cases in the 15-19 year age group. [1,4]

Primary cutaneous rhabdomyosarcoma is extremely rare and only 16 cases have been reported in the literature. It affects predominantly young males. Clinically, it manifests as an asymptomatic

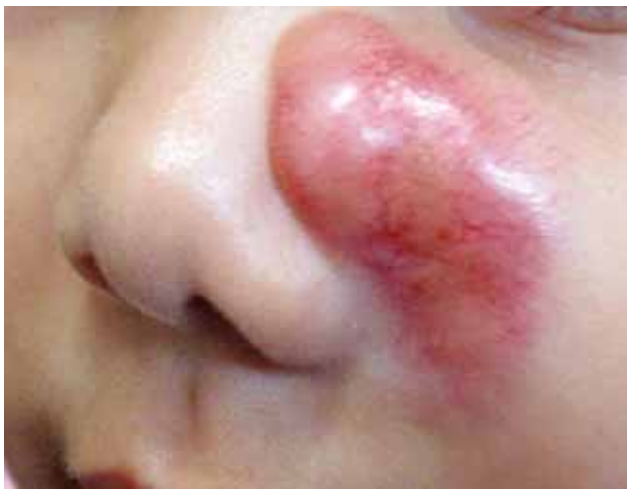

Figure 2: Erythematous, lobulated nodule with telangiectasias on its surface, measuring 3.2 x 2.4 cm, located on the left paranasal region

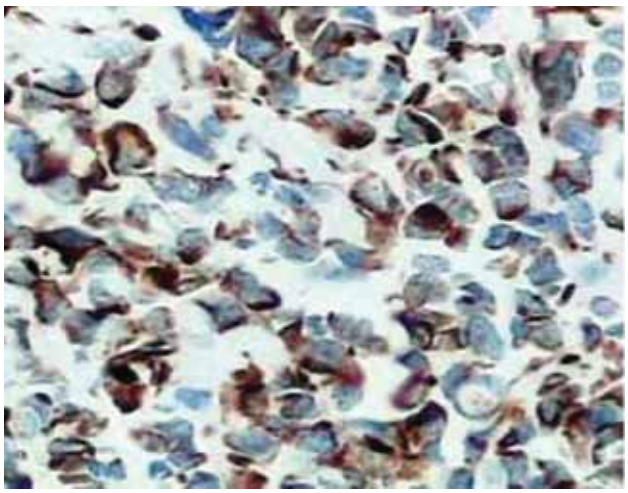

Figure 4: Desmin positive

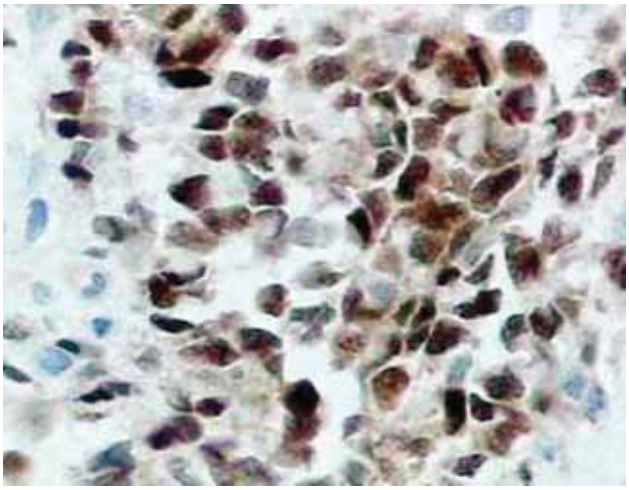

FIGURE 5: Myogenin positive

papule or nodule on the face, which grows progressively, altering anatomical structures and often causing obstruction. Metastases often occur by lymphatic dissemination to the regional lymph nodes and by hematogenic dissemination to the lungs, bones, bone marrow, central nervous system, heart, liver and breasts. [1-3,5]

Differential diagnosis of the skin lesions on the head and neck include: hemangioma, lymphoma, lymphangioma, cutaneous leukemia, angiofibroma, neuroblastoma, hematoma, cutaneous myofibromatosis, glioma, cellulitis, abscess, mastoiditis and other sarcomas. [2,5,6]

Histopathology is characterized by a dense dermal infiltrate composed of small, round, blue cells of clear cytoplasm with mitotic figures. These characteristics may be present in various pathologies such as Ewing's sarcoma, lymphoma and neuroblastoma; therefore, immunohistochemistry is mandatory in order to define diagnosis. Histopathological classification is divided into five groups: embryonic (58%), alveolar (31%), botryoid (6%), pleomorphic (4%) and undifferentiated (1%). The alveolar subtype is the most aggressive, progressing rapidly with early metastasis, resulting in high mortality rates compared to the embryonic type. [1-3,7,8]

Immunohistochemistry is an important tool in defining cell lineage. Anti-desmin, actin and myoglobin antibodies have been used as muscle markers, confirming the origin of the mesenchymal cells by differentiating skeletal muscle. Relatively new markers such as myogenin and MyoD1 identify nuclear proteins and are more sensitive and specific for rhabdomyosarcomas. The reactivity of the cells to desmin and myogenin is directly proportional to the degree of differentiation of the tumor, while the positivity of myogenin is strongly associated with the alveolar subtype. [2,3,8]

The treatment of choice is surgical, combined with adjuvant chemotherapy in all cases to control micrometastases. Radiotherapy is required when total resection is impossible. Combined therapy has increased survival by five years in recent decades from 25% in 1975 to 70% in 1991. [4,5,7]

Prognosis varies in accordance with the site of origin, the tumor size, clinical staging, the patient's age and the histological type. Factors indicative of good prognosis include early age at diagnosis, primary site in the genitourinary tract or orbital cavity, and embryonic or botryoid histological types. [1,5]

Despite the young age at which rhabdomyosarcomas affect patients, lesions that are completely resected are associated with a 5-year survival rate of 90%, which highlights the need for prompt diagnosis and treatment. ❑

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MAILING ADDRESS / ENDEREÇO PARA CORRESPONDÊNCIA:
Livia Lima de Lima
Rua A29, Conjunto Ajuricaba, 293, Planalto
69046-310 Manaus, AM, Brazil
Tel.: 92 9985-5252
E-mail: lilima_nb@hotmail.com

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