

Not your usual breast tumor: An unexpected embryonal rhabdomyosarcoma

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Abstract

Primary breast sarcomas are exceptionally uncommon, and rhabdomyosarcoma represents an even rarer subset that is predominantly encountered in younger populations. The embryonal form arising in the breast is particularly unusual and is recognized for its highly aggressive behavior. We describe a patient who presented with a rapidly progressive breast mass associated with distant metastatic involvement.

Histopathological and immunohistochemical evaluation revealed features consistent with embryonal rhabdomyosarcoma, showing evidence of skeletal muscle differentiation. The clinical evolution was marked by swift deterioration, and the patient succumbed to the disease shortly after diagnosis, before any therapeutic strategy could be implemented. This case underscores the exceptional nature of breast embryonal rhabdomyosarcoma in adults and illustrates the diagnostic and therapeutic challenges posed by its fulminant progression, emphasizing the need for heightened clinical suspicion and prompt pathological assessment despite the generally unfavorable prognosis.

Keywords: Adult rhabdomyosarcoma; Aggressive malignancy; Breast sarcoma, Embryonal rhabdomyosarcoma; Primary breast tumor; Poor prognosis

1. Introduction

Primary rhabdomyosarcoma of the breast is an exceptionally uncommon malignancy. Overall, breast sarcomas account for only a very small fraction of malignant breast tumors [1]. These neoplasms arise from the breast's mesenchymal components and encompass a highly diverse group of connective-tissue tumors. Reported histologic subtypes include malignant fibrous histiocytoma, liposarcoma, leiomyosarcoma, fibrosarcoma, angiosarcoma, rhabdomyosarcoma, clear cell sarcoma, neurogenic sarcoma, and other soft-tissue sarcomas [2].

Breast sarcomas may develop de novo or, less frequently, in association with prior radiotherapy or chronic lymphedema of the upper limb. Clinically, these tumors often resemble more common breast carcinomas, which may lead to diagnostic uncertainty [3]. However, their biological behavior, therapeutic approach, and prognostic implications differ significantly from epithelial breast cancers. The aim of this article is to outline the clinical and pathological characteristics of this rare malignancy and to highlight its key histological features.

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2. Case Presentation

We report the case of a 50-year-old woman with no known personal or family history of cancer, who presented to the university hospital with a large left breast mass that had rapidly increased in size over the past two months. Clinical examination revealed an enlarged, inflamed breast that was completely ulcerated (figure 1).



Figure 1 Clinical appearance of a large ulcerated and necrotic breast mass revealing an embryonal rhabdomyosarcoma

An ultrasonography study showed a 4.5 cm × 4 cm mass with evidence of central necrosis, heterogeneous echogenicity, posterior enhancement and internal vascularity. No skin invasion, dilated ducts or signs of inflammation noted in the study. The other breast and the axilla were unremarkable. A TAP CT scan showed a Voluminous breast tissue mass occupying the entire right breast, with irregular contours, poorly limited, heterogeneously enhanced after injection of PDCI.measuring 190x150x200 mm in diameter, and containing air bubbles, with secondary pulmonary localizations, Whole body bone scintigraphy was normal.

Histopathology results showed malignant tumor forming irregular fascicles and sheets. It showed a densely cellular proliferation composed of small round to oval tumor cells with hyperchromatic nuclei and scant eosinophilic cytoplasm, consistent with an embryonal-type morphology (Figures 2), Special staining (immunohistochemistry) was positive for Desmin (figure 3) , Vimentin (figure 4), CD99, MyoD1, Myoglobin, Muscle Specific Actin (focal).

Despite being identified as a candidate for chemotherapy her overall health rapidly declined, marked by the onset of asthenia and anorexia, resulting in a significant and concerning deterioration in her physical well- being.

Tragically, this decline culminated in a fatal outcome, leading to her death within days. This unfortunate event occurred before the planned chemotherapy treatment had even begun, underlining the aggressive and rapid nature of the disease.

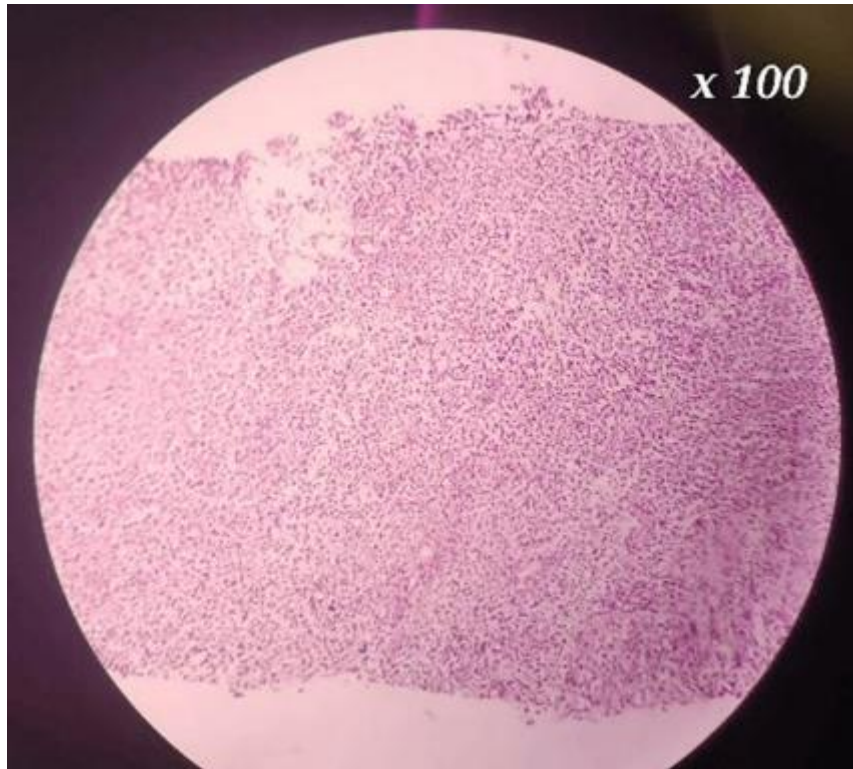


Figure 2 Hematoxylin-Eosin Stain ($\times 100$): Embryonal Rhabdomyosarcoma Morphology

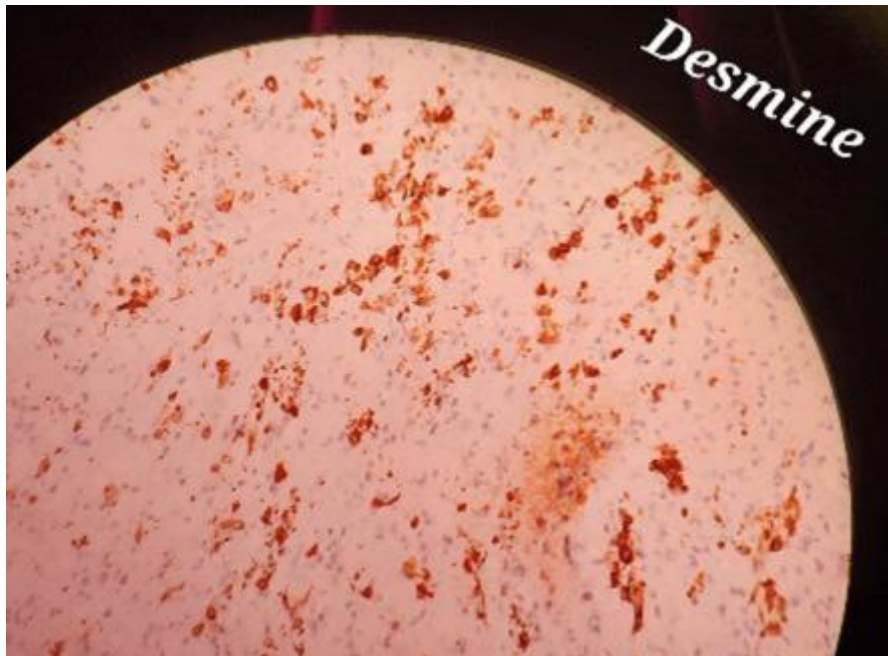


Figure 3 Desmin Immunohistochemistry: Focal Cytoplasmic Positivity Supporting Myogenic Differentiation

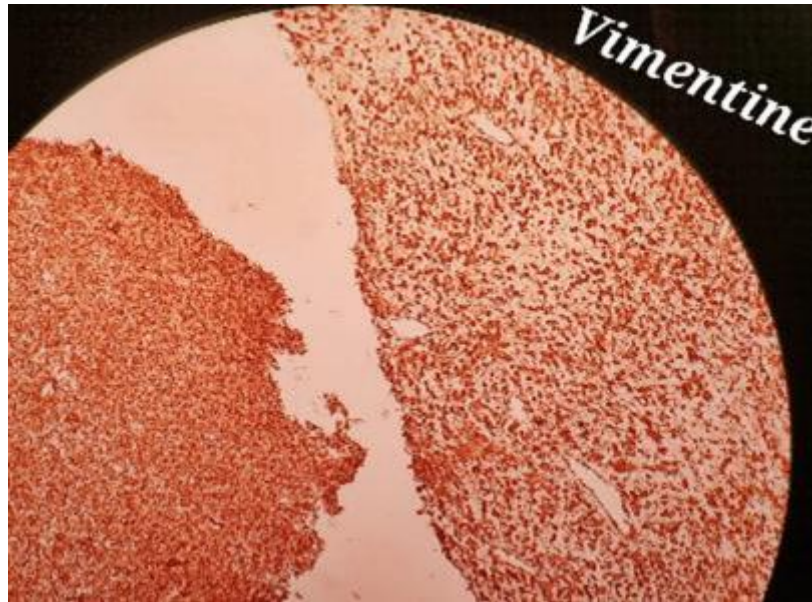


Figure 4 Vimentin Immunostaining: Diffuse Cytoplasmic Positivity Highlighting Mesenchymal Origin

3. Discussion

Most breast sarcomas arise without an identifiable cause, although prior radiotherapy is the most clearly established risk factor. Several hereditary cancer predisposition syndromes, including Li-Fraumeni syndrome, Gardner syndrome, and type 1 neurofibromatosis, have also been associated with an increased likelihood of developing mesenchymal breast tumors. Other environmental or iatrogenic factors—such as exposure to certain chemotherapeutic agents, arsenic compounds, vinyl chloride, or prolonged immunosuppression—as well as viral infections, particularly HIV and human herpesvirus, have also been implicated in the pathogenesis of soft-tissue sarcomas [4].

Pure non-epithelial malignant tumors of the breast are extremely rare, accounting for well under 1% of all breast cancers [4]. Among them, primary breast rhabdomyosarcoma in adults is exceptionally uncommon. Adult rhabdomyosarcoma represents only a small proportion of all soft-tissue sarcomas, making its occurrence in the breast particularly unusual [5]. Although these tumors may clinically and radiologically mimic more common breast neoplasms, their biological behavior and therapeutic implications differ significantly, underscoring the importance of an accurate diagnosis [4].

The histogenesis of primary breast rhabdomyosarcoma remains poorly understood. Several hypotheses have been proposed, including malignant transformation of displaced embryonic mesenchymal remnants, rhabdomyoblastic differentiation within mesenchymal-origin tumors, or neoplastic transformation of pluripotent periductal or acinar mesenchymal cells capable of differentiating into skeletal muscle cells. Regardless of the mechanism, primary breast rhabdomyosarcoma remains a rare entity from both diagnostic and biological perspectives.

Clinically, breast sarcomas often present as large, painless masses without associated nipple changes, skin involvement, or axillary lymphadenopathy. The typical patient described in the literature is a middle-aged woman, although cases have been reported across a wide age range and in both sexes [4]. Because lymphatic spread is uncommon in primary breast sarcomas, lymph node involvement at diagnosis is exceptional.

Management requires a multidisciplinary approach. Surgery remains the mainstay of treatment, with the primary objective being wide local excision with clear margins. Mastectomy may be considered for large tumors or local recurrence. Routine axillary lymph node dissection is generally not indicated due to the low incidence of nodal metastases, although it may be justified in cases of clinical lymphadenopathy or histologic variants with epithelial components, such as carcinosarcoma or liposarcoma. Sentinel lymph node biopsy has been proposed as a potential staging tool in selected cases, although its role has not yet been clearly established [5,6].

The benefits of adjuvant radiotherapy remain uncertain. Some authors suggest that it may be considered in patients with high-grade tumors, positive or close surgical margins, or large lesions, although its impact on disease-free survival

is variable [4]. Similarly, chemotherapy has no clearly defined role in adult breast sarcoma, and evidence supporting its routine use is limited.

Tumor size appears to be one of the most important prognostic factors. In a series of 25 patients with primary breast sarcoma, both overall and disease-specific five-year survival exceeded 90% for tumors smaller than 5 cm but declined markedly for lesions larger than 5 cm. Recurrent or metastatic tumors had significantly larger mean diameters than non-recurrent lesions [7]. These findings highlight the aggressive potential of large, rapidly growing sarcomas and reinforce the importance of early detection and complete surgical excision.

4. Conclusions

Although primary or metastatic breast rhabdomyosarcoma (RMS) is generally associated with young adolescents, our observations suggest that it may also occur in middle-aged women. Embryonal RMS carries a degree of metastatic potential. Myogenin is a useful marker for differential diagnosis. Axillary lymph node involvement and age may influence the prognosis of patients with primary breast RMS.

Compliance with ethical standards

Acknowledgments

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Disclosure of conflict of interest

The authors declare no competing interests

Author contributions

- All authors contributed to the conception, literature search, analysis, and manuscript drafting.
- All authors approved the final manuscript.

Data availability

All data are contained within the article and its references.

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