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Infiltration of Acute Myeloid Leukemia in the Breast – A Case Report

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Abstract

The infiltration of Acute Myeloid Leukemia (AML) in the breast is a rare condition, posing both diagnostic and therapeutic challenges. Due to the absence of specific radiological descriptions for such infiltrations, differential diagnosis with primary breast tumors becomes crucial. The authors report a case of a 19-year-old patient diagnosed with AML (FAB M2 RUNX1-RUNX1T1/t(8;21) + CKIT exon 17 mutation KIT D816V) after the term delivery of her child, with no obstetric complications. Disease recurrence occurred after the second cycle of consolidation chemotherapy, accompanied by a palpable lesion in the right breast, exhibiting progressive growth. Morphological and immunohistochemical analysis was consistent with AML infiltration of the breast parenchyma. This article contributes to the literature by presenting a rare case of AML infiltration in the breast, along with its clinical, radiological, and anatomopathological characteristics.

Introduction

The occurrence of breast tumors arising from extramammary neoplasms is a rare event, accounting for only 0.5% to 6.6% of cases, particularly associated with lymphomas, melanomas, rhabdomyosarcomas, pulmonary tumors, and gynecological malignancies [1].

In this context, mammary infiltration of hematologic diseases, such as Acute Myeloid Leukemia (AML), is also rare and can clinically and radiologically mimic primary breast tumors. AML is a malignant disorder characterized by uncontrolled proliferation of immature myeloid cells in the bone marrow, leading to progressive replacement of normal hematopoietic tissue. Infiltration into organs and tissues outside the bone marrow, such as the breast, presents additional diagnostic and therapeutic challenges. The absence of specific radiological descriptions demands histopathological examination for accurate diagnosis [1].

The authors present a case of a patient diagnosed with recurrent AML presenting with a palpable breast tumor.

Case Presentation

A 19-year-old female patient diagnosed with AML (FAB M2 RUNX1-RUNX1T1/t(8;21) + CKIT exon 17 mutation KIT D816V) during the puerperium of her first child in September 2022. The pregnancy was uneventful, with routine prenatal care in the Brazilian public health system. After initial treatment following the International Consortium on Acute Myeloid Leukemia (ICALM) protocol, she was admitted to our service in April 2023 with disease recurrence, characterized by anomalous expression of CD56+++, after the second cycle of consolidation chemotherapy. She denied other comorbidities, using only continuous progesterone-only oral contraceptive, and had no family history of oncological disease.

During hospitalization, she developed a palpable mass in the right breast, progressively growing and associated with local pain. Clinical examination revealed a bulge in the upper quadrants of the breast at the retroareolar region, with a palpable 5 cm × 5 cm nodular lesion in the retroareolar region, non-adherent to deep planes, and yellowish multiductal nipple discharge. Breast ultrasound showed a predominantly hypoechoic, irregular, and microlobulated nodule in the retroareolar region of the right breast, measuring approximately 4.8 cm × 4.2 cm × 4.7 cm, just-areolar, ACR BI-RADS: 4 (Figure 1). A core needle biopsy confirmed morphological and immunohistochemical features consistent with AML infiltration of the breast parenchyma. The patient remained hospitalized with leukemia-focused treatment, exhibiting a reduction in clinical and radiological dimensions of the breast lesion to 2.7 cm × 2.6 cm (Figure 2).

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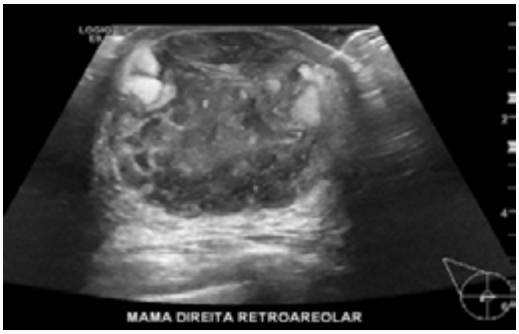

Figure 1: Right breast ultrasound.

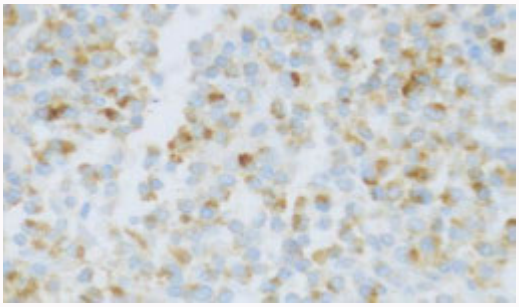

Figure 2: Biopsy slide with positive CKIT staining.

Despite improvement in the extramedullary lesion, the patient proved refractory to the MEC rescue protocol (mitoxantrone + etoposide + cytarabine). A new chemotherapy regimen, Mito-FLAG (mitoxantrone + fludarabine + cytarabine + filgrastim), was chosen, but the patient also showed refractoriness. During chemotherapy, the patient developed various opportunistic infections due to immunosuppression, including several episodes of febrile neutropenia, requiring hospitalization, and eventually succumbed to the disease eleven months after leukemia diagnosis.

Discussion

Palpable breast lesions of secondary etiology are rare, but awareness of this possibility is crucial for accurate diagnosis and treatment, especially when the disease initially presents as a breast lesion.

The pathophysiology of breast involvement in AML is still unknown, but it appears to be associated with the FAB M2 subtype and t(8;21), as in the presented case [2]. The FAB classification categorizes AML based on cell morphology and cytochemical study. M2 diseases lack mature cytoplasmic features and have few granules. Additionally, the evaluation of t(8;21) indicates translocation between chromosomes 8 and 21, resulting in pathological fusion of RUNX1-RUX1T1 proteins, blocking genes involved in hematopoiesis and affecting cellular differentiation and maturation. These types of leukemias occur in approximately 6% to 12% of cases, generally with a favorable prognosis but may more frequently exhibit mammary infiltrations [2]. However, in this specific case, the rapid disease relapse and the anomalous CD56+++ expression in the recurrence may indicate a worse hematological prognosis.

About 80% of AML infiltrations clinically manifest as palpable, painless, unilateral or bilateral masses [3] with accelerated growth, similar to the highlighted case. Imaging-specific presentations of these tumors are limited, with mammography typically showing hyperdense masses with microlobulated margins and architectural distortions. Microcalcifications are uncommon, as is axillary involvement. Ultrasound findings describe homogeneous, hypoechoic masses with indistinct or microlobulated margins, consistent with the reported case [4]. However, these features can be easily confused with primary breast tumors, necessitating histopathological confirmation.

The presented case adds to the literature by describing a young patient with recurrent and refractory AML and a breast tumor compatible with the underlying disease, showing improvement after a new chemotherapy regimen. The response to leukemia-focused treatment in this particular case underscores the interdependence between treating the underlying hematologic disease and controlling extramedullary manifestations. The differential diagnosis of breast tumors, especially in patients with other primary neoplasms, should be carefully established to define appropriate management. Emphasizing clinical surveillance, multidisciplinary evaluation, and in-depth investigation of patients with already defined oncologic diseases are crucial to optimize therapeutic and prognostic approaches in these unique cases.

Conclusion

The infiltration of AML in the breast represents a challenging clinical entity requiring careful assessment and vigilant investigation. Limited knowledge of the specific imaging characteristics of this condition highlights the need for advances in imaging diagnostic strategies and the importance of histopathological evaluation when facing differential diagnoses. The case report contributes to the literature by presenting an uncommon clinical scenario, emphasizing the importance of meticulous clinical surveillance, and emphasizing the need for further research to elucidate underlying pathophysiological mechanisms and optimize diagnostic and treatment strategies.

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