Quest Journals
Journal of Medical and Dental Science Research
Volume 13~ Issue 6 (June 2026) pp: 22-26
ISSN(Online): 2394-076X ISSN (Print):2394-0751
www.questjournals.org

Research Paper

Myeloid Sarcoma Associated with Pericardial Effusion Revealing Acute Myeloid Leukemia in a Child: A Case Report and Literature Review

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Abstract
Introduction
Myeloid sarcoma is a rare extramedullary proliferation of myeloid blasts, with pericardial involvement in children being an exceedingly uncommon presentation.
Case Report
We report the case of an 11-year-old girl admitted for a right breast mass, cervical lymphadenopathy, and abdominal pain evolving over two months. Laboratory workup revealed hyperleukocytosis at 200,000/mm³ with 48% blasts on peripheral blood smear. Echocardiography demonstrated a large circumferential pericardial effusion of 38 mm, and cytological analysis of the pericardial fluid identified 52% blasts. The diagnosis of acute myeloid leukemia complicated by pericardial myeloid sarcoma was established. Despite initiation of chemotherapy, the clinical course was rapidly unfavorable and the child died.
Conclusion
This case underscores the importance of considering leukemic cardiac infiltration in any unexplained pericardial effusion occurring in the context of a hematological malignancy, and highlights the key role of echocardiography and pericardial fluid cytology in the diagnostic workup.

Keywords
Myeloid sarcoma; Acute myeloid leukemia; Pericardial involvement; Pericardial effusion; Child.

*Received 06 June., 2026; **R**evised 15 June, 2026; **A**ccepted 17 June., 2026 © The author(s) 2026.*
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I. INTRODUCTION

The term chloroma is an anatomopathological diagnosis referring to an extramedullary proliferation of blasts belonging to one or more myeloid lineages. It represents an uncommon manifestation of acute myeloblastic leukemia (AML), although the diagnosis may be established before or after that of AML. Cardiac infiltration by myeloid sarcoma is exceedingly rare, and among the few published cases in the literature, a diagnosis of leukemia had almost invariably already been established.

II. CASE REPORT

1. Demographic Data

- An 11-year-old girl, the youngest of five siblings, with no significant past medical history, was admitted for a right breast mass.

2. History of PresentIllness

- Symptoms began two months prior to admission with the appearance of a right breast mass, abdominal pain, cervical swellings, and episodes of mild epistaxis, evolving in an afebrile context with deterioration of general condition.

3. Physical Examination

General Signs

- Afebrile child with impaired general condition characterized by asthenia, weight loss, and anorexia.

BreastExamination

Right breast:

- Large, firm mass (9×6 cm) with regular borders located in the upper quadrants, non-tender, with no signs of inflammation (no erythema, warmth, or edema).

Leftbreast:

- No abnormalitydetected.

CardiovascularExamination

- Tachycardia; S1 and S2 heart sounds clearly audible; no jugular venous distension, no lower limb edema, no cardiac murmur on auscultation.

PulmonaryExamination

- SpO2: 98%; respiratory rate: 21 breaths/min; vesicular breath sounds bilaterally preserved; no adventitious sounds on auscultation.

Abdominal Examination

- Soft abdomen; splenomegaly present; no collateral venous circulation; no hepatomegaly.

LymphNodeExamination

- Cervical lymphadenopathy: bilateral submental, left subdigastric, right spinal accessory, middle jugulo-carotid, and right retroauricular nodes.

4. Laboratory Investigations

Complete Blood Count (CBC)

- Hyperleukocytosis at 200,000/mm³ associated with bicytopenia.

Peripheral Blood Smear

- Blasts at 48%;myelemia at 13%.

PericardialFluidAnalysis

- Cytological examination of the pericardial fluid identified 52% blasts.

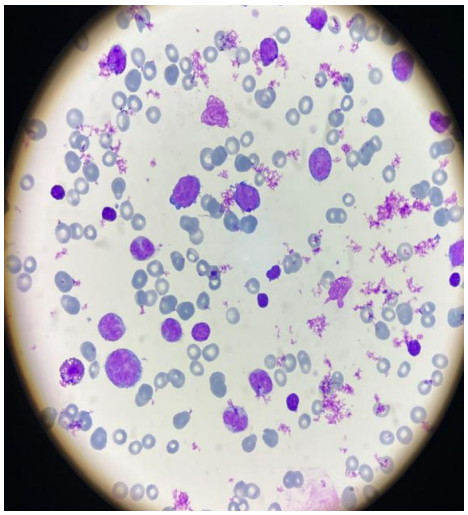

Figure 2. Cytological examination of the pericardial fluid (May-Grünwald-Giemsa stain). The preparation reveals a predominance of large immature myeloid cells (blasts) with high nucleus-to-cytoplasm ratio, fine chromatin, and visible nucleoli, representing 52% of the cellularity. This finding is consistent with extramedullary leukemic infiltration (pericardial myeloid sarcoma).

5. Imaging

Chest X-ray

- Cardiomegalywiththoracic distension.

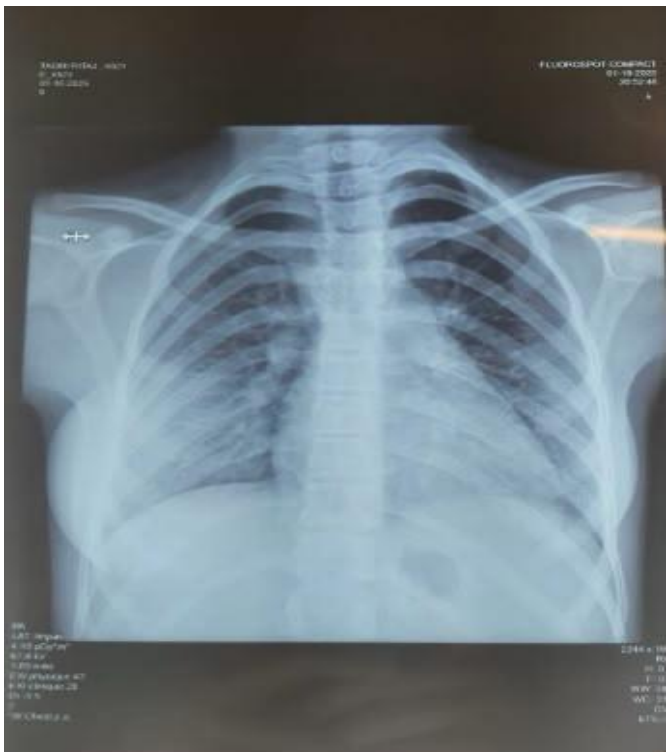

Figure 1. Chest X-ray showing cardiomegaly and thoracic distension in an 11-year-old girl with acute myeloid leukemia complicated by pericardial myeloid sarcoma. Note the enlarged cardiac silhouette consistent with large circumferential pericardial effusion (38 mm on echocardiography).

Echocardiography

- Large circumferential pericardial effusion of 38 mm.

6. Final Diagnosis

- Acute MyeloidLeukemia (AML)
- Pericardialmyeloidsarcoma

7. Treatment

Initial SupportiveMeasures

- Supplementaloxygentherapy
- Fluidresuscitation: 20 mL/kg
- Transfusion of one packed red blood cell unit and four platelet concentrates

Chemotherapy

- Two courses of chemotherapyadministered

8. Clinical Course

- The clinical course was marked by rapid deterioration and death of the patient despite initial resuscitation measures and initiation of chemotherapy.

III. DISCUSSION

Myeloid sarcoma (MS), formerly known as chloroma or granulocytic sarcoma, is defined as an extramedullary tumoral proliferation of immature myeloid precursor cells. (1)

A case series reviewing all reports of acute myeloid leukemia over a 12-year period revealed that primary myeloid sarcoma accounted for only 1.4% of all cases, with the most common extramedullary manifestations involving the skin, lymph nodes, and testes. Consequently, extramedullary myeloid sarcoma within the myocardium is exceedingly rare. (2)

Cardiac infiltration is an uncommon finding in any hematological malignancy. (3) Among cases with infiltration, clinical signs are rare and observed in fewer than 1% of cases. (4)

A post-mortem study of patients with known leukemia revealed cardiac infiltration in only 4% of cases, the vast majority of which were asymptomatic and undiagnosed prior to autopsy. (3)

A literature review published in the Journal of Clinical and Diagnostic Research, which analyzed all case reports from 1960 to 2016 referencing cardiac myeloid sarcoma, identified fewer than thirty reported cases, underscoring the exceptionally rare nature of this location. (5)

The epidemiology of myeloid sarcoma (MS) is difficult to establish due to imprecise definitions, studies including cases without histological confirmation, and the fact that imaging is not a standard investigation for characterizing acute myeloid leukemia (AML). Adult patients with MS are generally between 40 and 70 years of age, with a male predominance. (6,7,8)

Predisposing factors for the development of myeloid sarcoma are similar to those favoring AML, including occupational, environmental, lifestyle-related, and genetic factors. [9] However, the mechanisms by which leukemic stem cells (LSCs) engraft in tissues to form solid tumors remain poorly understood. Some authors suggest that the mobilization and migration of LSCs from the bone marrow to the peripheral blood, spleen, and extramedullary sites may be influenced by several factors, including alterations in LSC adhesion to the medullary niche due to modified expression and dysregulation of adhesion molecules, chemokine receptors/ligands, and integrins. Additional mechanisms such as activation of the RAS-MAPK/ERK pathway, dysregulation of epithelial-mesenchymal transition (EMT) pathways, and pathogenic mutations of nuclear factor erythroid 2 (NF-E2) also contribute to this process. [10]

The most frequently implicated genetic abnormalities in the pathogenesis of de novo MS are t(8;21), inv(16), KMT2Ar, JAK2 V617F, and in secondary cases, KMT2Ar and BCR::ABL1. (11)

The diagnosis of myeloid sarcoma is typically established through anatomopathological examination combined with immunohistochemistry. (12) Cellular morphology is assessed on hematoxylin and eosin-stained slides. [13]

Conventionally, the diagnosis of acute leukemia relies on a combination of evidence including morphological analysis of the bone marrow aspirate (myelogram), immunophenotyping by flow cytometry, and cytogenetic and molecular data. Nevertheless, the combination of marked hyperleukocytosis, significant peripheral blast count, the presence of blasts in the pericardial fluid, and a strongly suggestive clinical context allowed for a high-probability diagnosis of AML complicated by pericardial myeloid sarcoma.

However, in certain situations, as in our case, these investigations could not be completed due to the rapidly unfavorable clinical course and early death of the patient prior to completion of the full hematological workup.

Nevertheless, the diagnosis was strongly supported by several concordant elements. The presence of blastic cells in the pericardial fluid constitutes a major argument in favor of extramedullary leukemic infiltration. Cytological examination of effusion fluids is recognized as a reliable diagnostic method when blasts are identified, particularly in a suggestive hematological context. Furthermore, the clinical and biological findings, including the suspicion of acute leukemia at initial workup, reinforce the diagnostic hypothesis.

With regard to treatment, management is primarily based on the chemotherapy protocols used in AML. (14) However, in the absence of randomized prospective studies, optimal therapeutic recommendations for patients with MS have yet to be established.

Despite the initiation of appropriate treatment, the prognosis remains frequently poor in forms associated with cardiac involvement, particularly when the diagnosis is delayed or when the clinical presentation is complicated by a large pericardial effusion.

In our case, the clinical course was unfortunately rapidly unfavorable despite initial resuscitative measures and administration of chemotherapy.

IV. CONCLUSION

This case report highlights the importance of considering leukemic cardiac infiltration in the presence of any unexplained pericardial effusion occurring in a context suggestive of hematological malignancy. It also underscores the essential role of echocardiography and cytological analysis of the pericardial fluid in the diagnostic workup. Early recognition of this rare entity may allow for more rapid management and potentially improve prognosis.

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