

Mixed phenotype acute leukemia mimicking adult-onset Still's disease in a pregnant female: A rare case report

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Introduction

Adult-onset Still's disease (AOSD) is a rare systemic autoinflammatory disorder characterized by quotidian fever, salmon-pink rash, arthritis or arthralgia, leukocytosis, and markedly elevated ferritin levels.[1] Diagnosis relies on the Yamaguchi and Fautrel criteria after excluding infectious, autoimmune, and malignant conditions.[2,3] AOSD can closely mimic hematologic malignancies, particularly in patients with persistent fever, leukocytosis, and hyperferritinemia.[1,4] Mixed-phenotype acute leukemia (MPAL) is a rare leukemia characterized by blasts expressing both myeloid and lymphoid lineage markers.[6,7] We report a rare case of a pregnant woman who initially fulfilled criteria for AOSD but was subsequently diagnosed with MPAL. This case highlights the importance of considering hematologic malignancy in patients with atypical features, persistent blood abnormalities, or poor treatment response, and emphasizes the role of early bone marrow evaluation.

Case Report

1. Examination Findings
Joint swelling and tenderness: wrists, elbows, knees, ankles.
Bilateral cervical, preauricular, and postauricular lymphadenopathy.
Mild abdominal distension consistent with pregnancy.
Hepatosplenomegaly on ultrasound.

2. Investigations
Hb: 9.5 g/dL
TLC: $38,200 \times 10^9/L$
Platelets: $44,000 \times 10^9/L$
Ferritin: 980 ng/mL
Inflammatory markers: markedly elevated.
Lymph-node FNAC: no malignancy.

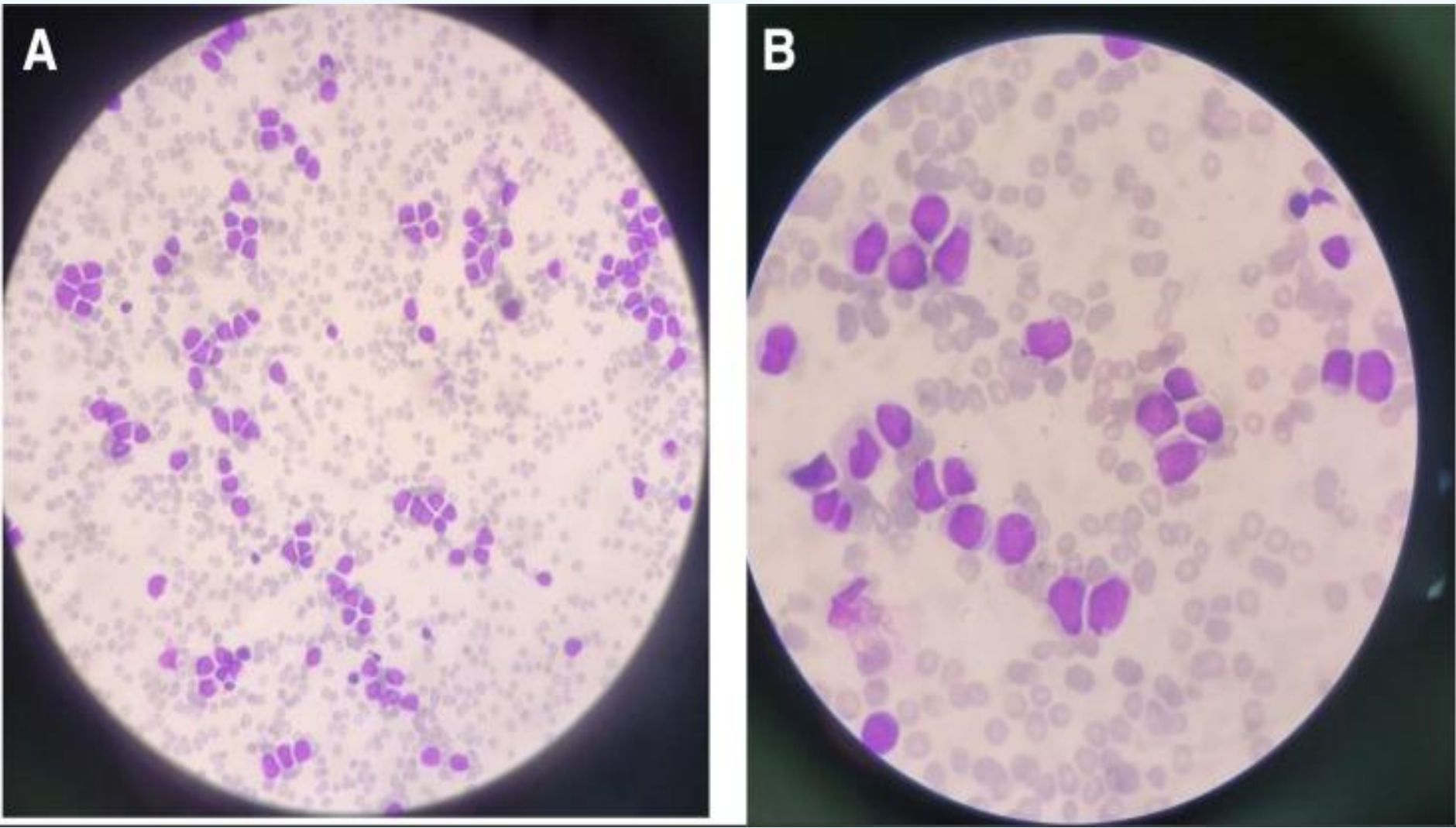
3. Initial Diagnosis AOSD
Fulfilled Yamaguchi criteria (2 major + 3 minor).

4. Clinical Course
Treated with methylprednisolone and hydroxychloroquine showed no significant clinical improvement.
Repeat smear showed circulating blasts.

5. Final Diagnosis
Flow cytometry showed B-lineage and myeloid/monocytic blasts.
Diagnosis: Mixed-phenotype acute leukemia (MPAL).
Bone marrow biopsy was not performed due to patient refusal.
BCR-ABL PCR: negative.

6. Treatment
AOSD therapy discontinued.
Modified hyper-CVAD chemotherapy initiated.
Methotrexate withheld due to pregnancy.
Prophylactic intrathecal therapy given for CNS protection.

Radiography & Echocardiography



Microscopic pictures of the peripheral blood smear show hypercellularity, with blasts comprising >80% of total nucleated cells. The leukemic blasts are large, high nuclear-to-cytoplasmic (N:C) ratio, finely dispersed chromatin, and 1 to 3 prominent nucleoli. The cytoplasm is moderately abundant and basophilic without definitive Auer rods.

Clinical Timeline

Time frame	Events
10 weeks ago	Progressive polyarthralgia involving multiple joints
2 weeks ago	Cervical, preauricular, and postauricular lymphadenopathy developed
Day 1	Presented at 20 wk POG with arthralgia, fatigue, and lymphadenopathy; labs showed leukocytosis, anemia, thrombocytopenia, and elevated ferritin.
Initial diagnosis	AOSD diagnosed according to Yamaguchi criteria.
Day 7	Poor response to treatment; repeat smear revealed circulating blasts
Day 21	Flow cytometry confirmed MPAL with myeloid and lymphoid blasts.
Day 23	Modified hyper-CVAD chemotherapy initiated.
Day 42	At 26 wk POG, viable fetus and improved clinical/laboratory parameters.

Laboratory Profile

Test	Patient value
Hemoglobin	9.5 g/dL
Platelet count	$44 \times 10^9/L$
TLC	$38.2 \times 10^9/L$
Neutrophils	81%
Ferritin	985 ng/mL
ANA	Negative
Anti-dsDNA	0.5 IU/mL
Anti-CCP	0.5 U/mL
Anti-HCV	Negative

Conclusion

This case highlights the atypical presentation of MPAL during pregnancy with concurrent AOSD-like features, leading to delays in appropriate hematological workup, diagnostic ambiguity, and therapeutic challenges. Chemotherapy-induced teratogenicity and the competing priorities of maternal and fetal welfare posed distinct therapeutic dilemmas. A timely diagnosis, multidisciplinary approach, and individualized chemotherapy are essential to optimize maternal and fetal outcomes in such complex cases.

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