

Monocytosis the great masquerade; ETP-ALL hiding behind JMML in a 5 year old boy- A Case Report

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Background:

Persistent monocytosis in pediatric patients can be associated with reactive conditions as well as hematological malignancies.¹ Important differentials include Juvenile Myelomonocytic Leukemia (JMML), a myeloproliferative/myelodysplastic neoplasm, associated with hepatosplenomegaly, leucocytosis and absolute monocytosis, while there may also exist a clinical and morphological overlap between JMML and certain other malignancies, including Early T-cell Precursor Acute Lymphoblastic Leukemia (ETP-ALL)²

Aim: To highlight the diagnostic pit falls in resource-limited countries and how to navigate them accordingly

Case Discussion: A 5 year old boy presented to our clinic with:

- Cytopenias
- Absolute monocytosis (1,150/ μ L)
- Hepatosplenomegaly

Based on bone marrow morphology and immunohistochemistry, he was again diagnosed with JMML. He couldn't afford treatment so after a few months, he presented again with deteriorating health.

Another bone marrow and flowcytometry:

A repeat bone marrow biopsy now revealed Acute Leukemia. A flowcytometry was advised by clinician for accurate diagnosis, which revealed pattern most consistent with Early T- Precursor Lymphoblastic Leukemia/Lymphoma (ETP-ALL).

Cytogenetics in play:

A concurrent cytogenetic studies revealed Monosomy 7, indicating a high risk disease in setting of ETP-ALL.

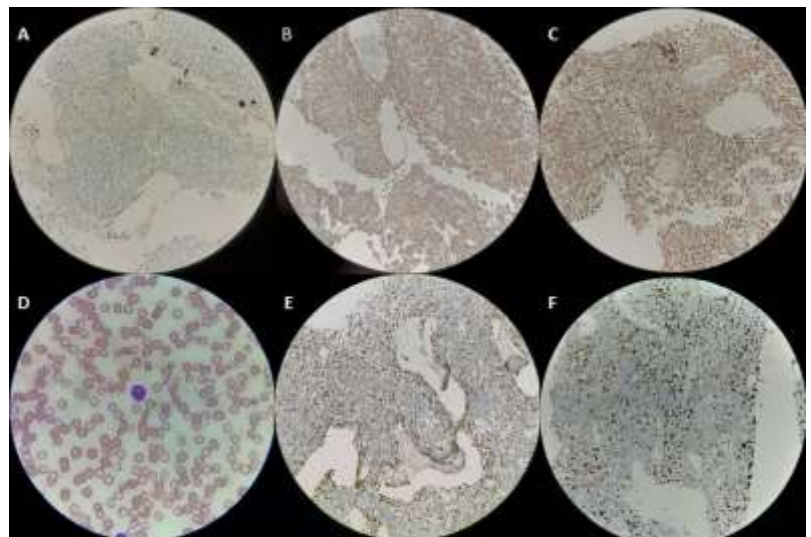


Figure 1: Section A, B and C show CD34, MPO and CD68 immunostains respectively which aided in the diagnosis of JMML. Section D shows a blast in peripheral smear which was seen on subsequent marrow. Section E and F show CD34 and TdT immunostains respectively in ETP-ALL.



Figure 2: Illustrates Monosomy 7 on conventional cytogenetics

Flow cytometry revealed blasts

Positive for CD33, CD34, CD38, cytoCD3, CD7, CD117 (partial), CD5 (weak and partial) and TdT

Negative for CD13, CD4, CD8 and MPO

Results: Patient received UK-ALL regimen protocol but responded poorly as MRD remained positive but cytogenetic remission was achieved. CNS involvement was also encountered. However, patient tragically passed away due to a brain aneurysm rupture.

Conclusion: In setting of resource-limited countries, timely diagnosis of overlapping malignancies can be delayed. Reliance on morphology alone is insufficient; therefore, integrating flowcytometry, cytogenetics and NGS is essential for risk stratification and personalized patient care.

References: 1. Di Giacomo D, et al. Diagnostic challenges in acute monoblastic/monocytic leukemia in children. Front Pediatr. 2022;10:911093. doi: [10.3389/fped.2022.911093](https://doi.org/10.3389/fped.2022.911093)

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