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# ACUTE ERYTHROBLASTIC LEUKEMIA, A RARE ENTITY WITH POOR PROGNOSIS: A CASE FROM THE NATIONAL CANCER INSTITUTE OF MEXICO


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## INTRODUCTION

Acute erythroid leukemia (AEL) also known as primary erythroid leukemia (PEL) is a unique, aggressive, entity characterized by the neoplastic proliferation of erythroid cells, usually ≥ 80% bone marrow elements of which ≥ 30% are proerythroblasts and represents < 1% of all acute myeloid leukemias. It carries a dismal prognosis with a median overall survival of < 6 months.

## CLINICAL CASE

A sixty-six-year-old woman with no medical history was referred to our hospital with a six-month history of weight loss, asthenia, adynamia, and dyspnea and a recent episode of lower gastrointestinal bleeding that was managed with transfusions. Due to pancytopenia, a bone marrow aspirate was performed, revealing blasts prior to referral. Upon admission, physical examination was remarkable for general pallor and fever; there were no enlarged lymph nodes. Complete blood count showed pancytopenia. A bone marrow aspirate and biopsy with flow cytometry, cytogenomics and molecular studies was performed**.**

## FOLLOW-UP

She was managed with antibiotics for sepsis of urinary and pulmonary origin, with isolation of a carbapenemase-producing organism. The bone marrow aspirate showed 84% erythroid predominance and flow cytometry demonstrated 54% proerythroblasts with an immunophenotype CD34-, HLADR -, cyMPO-, CD16-, CD7+h, CD117+, CD36+h, CD105+, CD71+, suggestive of AEL. Cytogenetic studies revealed a complex karyotype with aneuploidies and presence of five marker chromosomes. Significant material losses were observed in autosomes. Despite treatment, she developed respiratory failure which required intensive care management and passed away.

## DIAGNOSTIC STUDIES

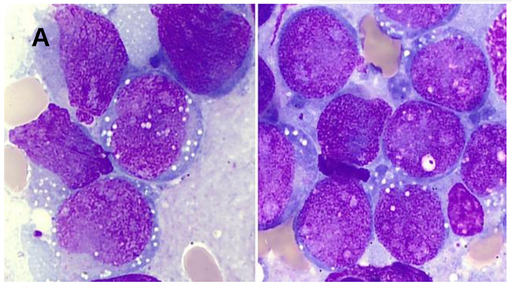



Figure A. Proerythroblasts predominance on aspirate

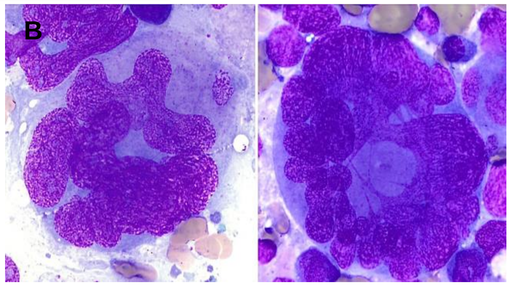



Figure B. Multinucleated proerythroblasts

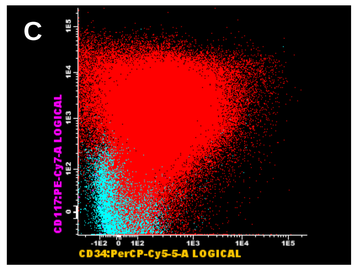



Figure C. CD 34 negativity with CD 117 positiviity

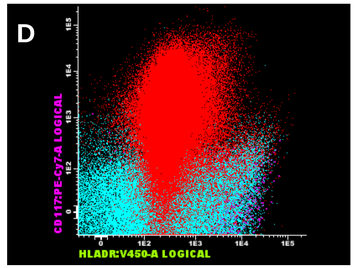



Figure D. HLA DR negativity with CD 117 positivity

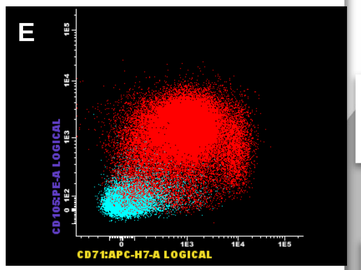



Figure E. CD71 and CD 105 positivity

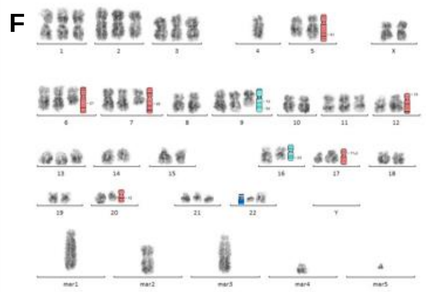



Figure F. Complex Karyotype

| Parameters | Value |
|---|---|
| Hemoglobin | 6.8 g/dL |
| Hematocrit | 18.9 % |
| Median Corpuscular Volume | 89.4 fL |
| Leukocytes | 1.66 x $10^9$/L |
| Blasts | 3% |
| Neutrophils | 0.4 x $10^9$/L |
| Platelets | 12 000 x $10^9$/L |
| Fibrinogen | 385 mg/dL |

## CONCLUSIONS

AEL is a rare and aggressive form of leukemia whose diagnosis poses a challenge and requires urgent attention. It has a unique morphology, immunophenotype characterized by the presence of erythroid specific antigens and complex cytogenetics often associated with *ETV*, *PDGFRB*, *KMT2A* and *TP53* mutations, which carries a dismal prognosis. There is no established therapy nowadays and the identification of new cases as the one described contributes to the understanding of this entity and the potential development of novel treatment options.

## ACKNOWLEDGEMENTS


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## CONTACT INFORMATION


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## REFERENCES


1. Reichard, K.K., Tefferi, A., Abdelmagid, M. *et al.* Pure (acute) erythroid leukemia: morphology, immunophenotype, cytogenetics, mutations, treatment details, and survival data among 41 Mayo Clinic cases. *Blood Cancer J.* 12, 147 (2022).
2. Wang W, Wang SA, Medeiros LJ, Khoury JD. Pure erythroid leukemia. Am J Hematol. 2017 Mar;92(3):292-29.
3.Bacher U, Haferlach C, Alpermann T, et al. Comparison of genetic and clinical aspects in patients with acute myeloid leukemia and myelodysplastic syndromes all with more than 50% of bone marrow erythropoietic cells. Haematologica. 2011;96:1284–1292.