IMAGES IN HEMATOLOGY
DOI: 10.4274/tjh.galenos.2025.2024.0391

Acute Myeloid Leukemia with NUP98::LNP1 Fusion Mimicking Chronic Myeloid Leukemia

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October 14, 2024
March 17, 2025

A 60-year-old female patient presented with a history of hepatitis, tuberculosis and gastric cancer, developed recurrent fever during the treatment for gastric cancer. Peripheral blood (PB) examination showed white blood cell count of 47.7×10^9/L, consisting 1% blasts, 1% promyelocytes, 2% neutrophilic myelocyte, 5% neutrophilic metamyelocytes, 13% neutrophils, 8% lymphocytes, 3% monocytes, 3% eosinophils, and 64% basophils. The hemoglobin concentration of 87 g/L, and platelet count of 75×10^9/L. The bone marrow (BM) smear displayed an active proliferation of granulocytes, with 10% myeloblasts (Figure A). The proportion of eosinophils and basophils in the BM was also notably increased, accounting for 9% and 24%, respectively (Figure A). Additionally, the percentage of basophils in the PB was significantly elevated, reaching 52% (Figure B). The morphologic features of BM and PB closely mimic the chronic myeloid leukemia (CML) in chronic high-risk phase, characterized by a significant elevation in basophils. Interestingly, both RT-qPCR and FISH indicated negativity for the *BCR::ABL1* fusion gene. The chromosomal karyotype was *46,XX,t(3;11)(q12.2;p15.4).* Meanwhile, RNA sequencing identified only the *NUP98::LNP1* fusion transcript in this case, with no detectable mutations in other genes (e.g., *ASXL1, SETBP1, NRAS, KRAS, SRSF2, TET2, CBL, CSF3R, JAK2, ETNK1, SETBP1*, etc.). Ultimately, a diagnosis of acute myeloid leukemia with *NUP98* rearrangement was established, as AML with *NUP98* rearrangements is exempt from the historical threshold of 20% blasts according to WHO 2022 guidelines[1,2].

AML with *NUP98::LNP1* fusion transcripts are extremely rare. To date, only one case has been reported in which the blasts count exceeded 20%[3]. This case represents the first reported instance of specific morphologic presentation in AML with *NUP98::LNP1* fusion transcripts. Additionally, numerous studies have demonstrated that *NUP98* rearrangements are associated with an unfavorable prognosis, highlighting the critical importance of their identification[4-6].

AUTHOR CONTRIBUTIONS

Z.Y and H.W wrote the manuscript, Y.P collect the data and information of patient. All authors reviewed and approved the final manuscript.

DATA AVAILABILITY STATEMENT

The data of this article are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

CONFLICT OF INTEREST DISCLOSURE

The authors declare no conflict of interest.

ETHICS APPROVAL STATEMENT

This study protocol was reviewed and approved by Chongqing University Cancer Hospital.

PERMISSION TO REPRODUCE MATERIAL FROM OTHER SOURCES

N/A.

CLINICAL TRIAL REGISTRATION

N/A.

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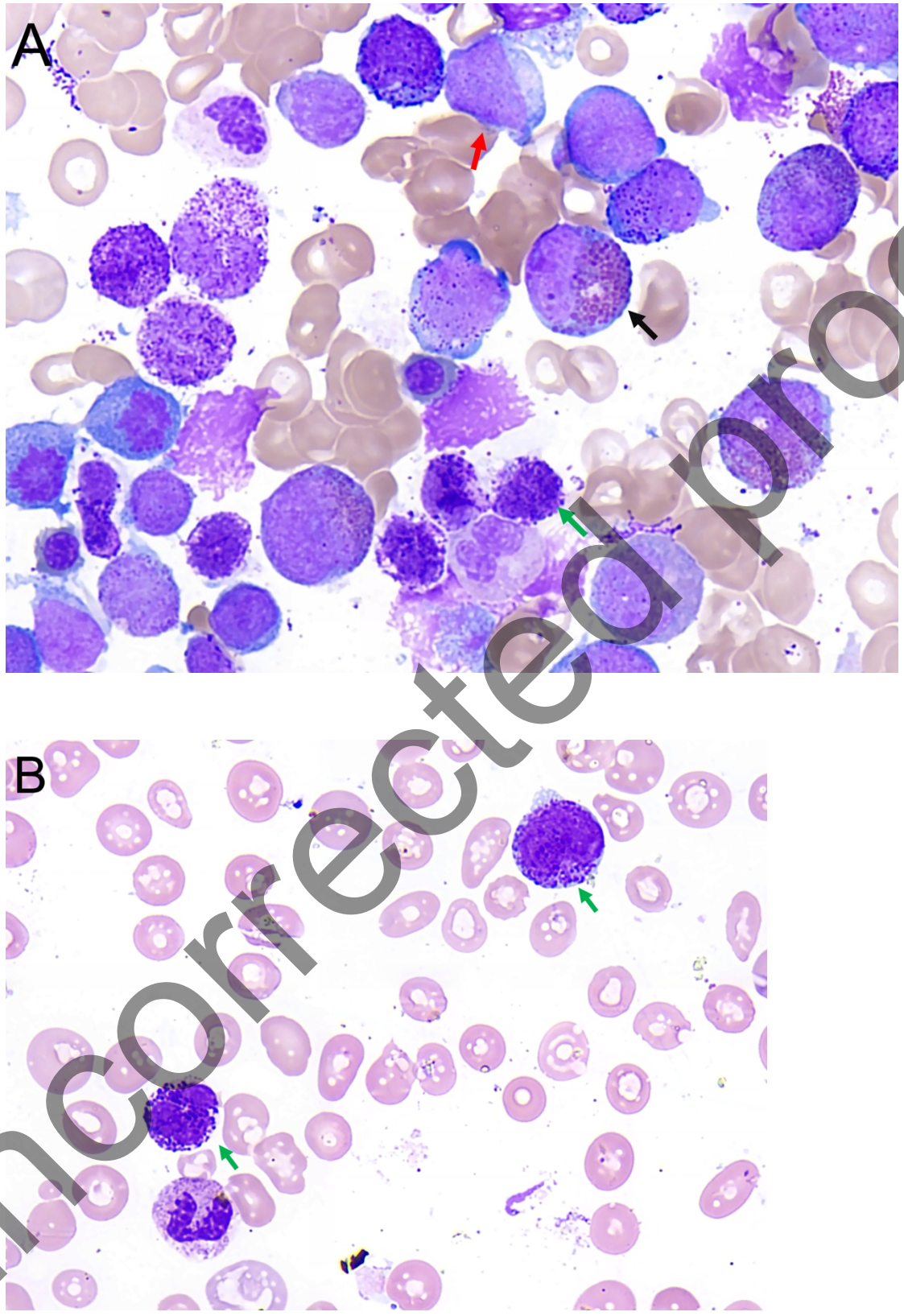

Figure A and B, ×1000 original magnification, Wright–Giemsa stain. The red arrow points to a myeloblast, the black arrow points to an eosinophil, and the green arrow points to a basophils.