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Acute Myeloid Leukemia with NUP98::HOXA9: Case Report and Review of Literature

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Abstract

Background

A 60-year-old woman was admitted to our hospital complaining of moderate anemia and thrombocytopenia in March 2025.

Methods

A peripheral blood examination showed the following counts: white blood cell count(WBC) $9.02 \times 10^9/L$, red blood cell count (RBC) $3.59 \times 10^{12}/L$, hemoglobin 80g/L, platelet count $48 \times 10^9/L$, and 16% of leukemic blasts were stained gray-blue. Leukemic blasts accounted for 20% of the bone-marrow cells. The patient demonstrated a acute myeloid leukemia(AML) phenotype, and showed expression of HLA-DR, CD13, CD33, CD34, CD38, CD117, CD123 and MPO. A biopsy analysis revealed scattered granulocytic immature cells with myelofibrosis, and megakaryocytes with obvious pathological

changes. A traditional cytogenetic analysis revealed 46,XX,t(7;11)(p15;p15). A 43 fusion-genes screening analysis showed a NUP98::HOXA9 fusion. In conclusion, the woman definitive diagnosed AML with NUP98::HOXA9 which belonged to high risk group according to the chinese guidelines for diagnosis and treatment of adult acute myeloid leukemia(not APL)(2023). Two months later, first complete remission(CR) was achieved after induction chemotherapy with VA(venetoclax and azacitidine) regimen. To september 2025, the patient was followed up well.

Conclusion

NUP98::HOXA9 defines a subgroup of AML, which most frequently appearance of t(7;11)(p15;p15) karyotype. It has since been estimated that AML with NUP98::HOXA9 accounts for 0.66% of all AMLs. Patient with NUP98::HOXA9 can achieve a good remission rate with first induction, but be prone to relapse.

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