blood work
images in hematology

Acute myeloid leukemia with *NUP98*::*HOXA9* mimicking acute promyelocytic leukemia

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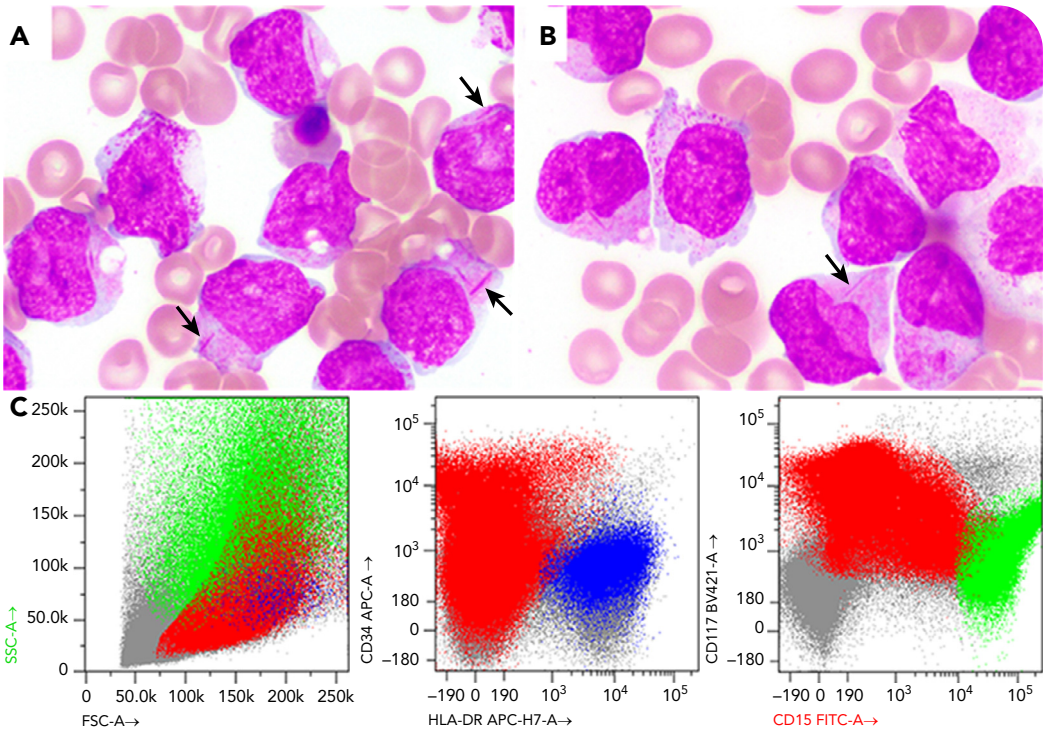

Flow cytometric analysis (red: neoplastic myeloid blasts; green: granulocytes; blue: monocytes)

A 56-year-old woman was admitted for anemia (hemoglobin, 7.8 g/dL) and thrombocytopenia (platelets, 25 000/μL), which was concerning for acute myeloid leukemia (AML). The peripheral blood (PB) and bone marrow (BM) aspirate smears showed numerous hypergranular promyelocytes with Auer rods (panels A and B, black arrows; Wright-Giemsa stain on BM aspirate; 100× objective). Flow cytometric study on BM aspirate (panel C) demonstrated abundant promyelocytes (62%) with high side scatter (corroborating hypergranularity) and $CD34^{partial+}$/human leukocyte antigen (HLA)-$DR^{predominantly-}$/$CD15^{partial+}$/$CD117^{+}$. There was a mild increase in D-dimer (2.31 μg/L; reference value, ≤0.59 μg/L). These findings were suspicious for acute promyelocytic leukemia (APL). The patient was initially treated with all-*trans* retinoic acid, but treatment was discontinued once fluorescence in situ hybridization studies indicated an *NUP98* rearrangement but no evidence of *PML*::*RARA* or *RARA* rearrangement. Cytogenetics revealed an isolated 46,XX,t(7;11)(p15;p15), resulting in the *NUP98*::*HOXA9* fusion that was confirmed by next-generation sequencing (NGS) (additional *TET2* [p.Leu1229Arg and p.Gln635*] detected). The patient was started on standard induction chemotherapy with the 7 + 3 regimen.

To our knowledge, this is the first case of *NUP98*::*HOXA9* rearranged AML that morphologically and immunophenotypically resembles APL; notably, *NUP98*::*RARA* fusion has been recently reported in APL. This case highlights the importance of combining different techniques (PB/BM examination, flow cytometry, cytogenetics, and NGS) to identify driver lesions and select appropriate therapies.

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