blood work
images in hematology

BCOR/BCORL1 mutated hypergranular cells mimicking acute promyelocytic leukemia

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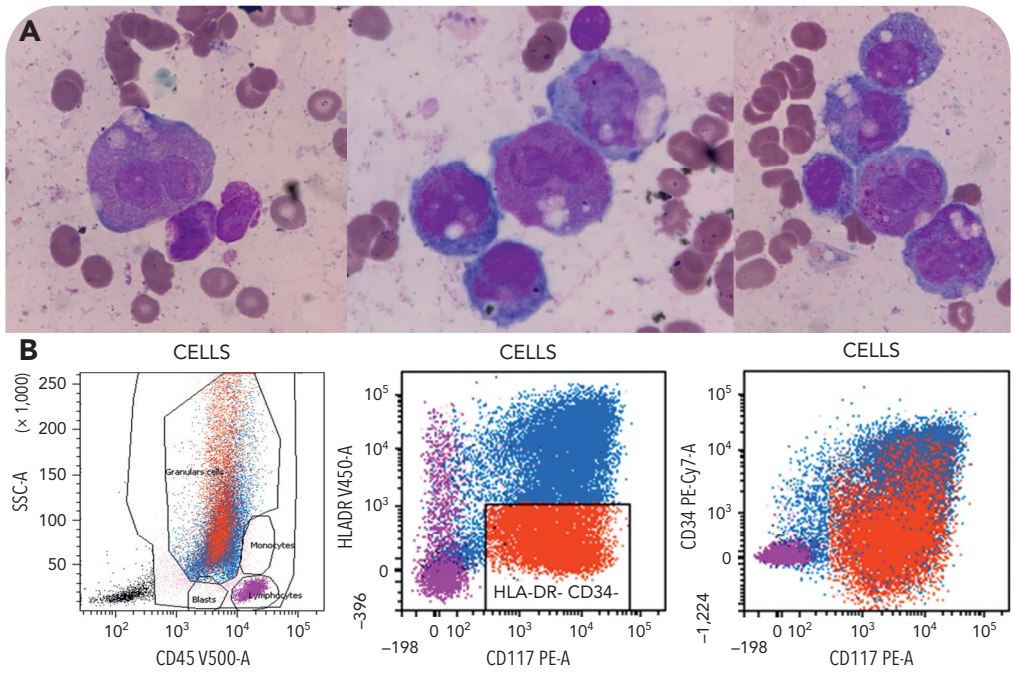

A 69-year-old woman was admitted for pancytopenia. Complete blood count showed the following: a leukocyte count of 1.3 × 10^9/L (reference values: 4-10 × 10^9/L); hemoglobin level of 46 g/L (reference values: 120-160 g/L); platelet count of 26 × 10^9/L (reference values: 150-350 × 10^9/L); and normal coagulation parameters. Bone marrow aspirate showed the presence of blast cells (90%), with multiple granulations associated with irregular nuclei, suggesting acute promyelocytic leukemia (APL) (panel A; May-Grunwald-Giemsa stain, objective 100×, original magnification ×100), although no Auer rod was detected. Flow cytometry analysis showed a side scatter pattern typical of APL diagnosis (blasts in granulocytes). Moreover, blast cells expressed CD13, CD33, and CD117, and 55% of them were negative for CD34 and HLA-DR (panel B), in agreement with the World Health Organization definition of APL. Full cytogenetic analyses based on 24 metaphases revealed 46,XX. Fluorescence in situ hybridization analysis revealed no *PML-RARA* gene fusion. Next-generation sequencing (46-gene panel) identified mutations in *BCOR* and *BCORL1* (variant allele fraction [VAF] 42%), *NRAS* and *STAG2* (VAF 39% for both), and 2 *TET2* variants (VAF 39% and 42%).

In this patient, the presence of atypical blast cells and flow cytometry features suggested APL, but without the classical *PML-RARA* gene fusion. Finally, the identification of *BCOR/BCORL1* mutations, which have been reported only once in acute myeloid leukemia, confirmed another mechanism for hypergranular acute myeloid leukemia.

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