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Secondary AML with *MLL* gene amplification

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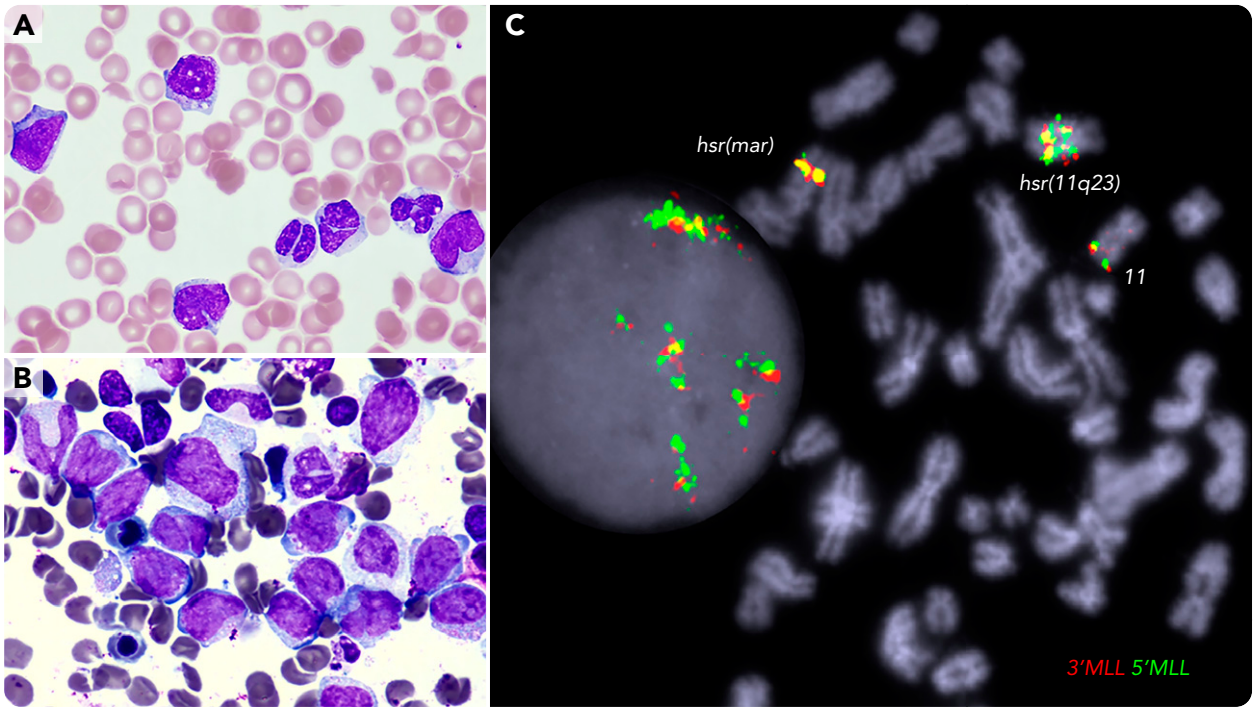

A 72-year-old female with a history of *JAK2*-mutated essential thrombocythemia presented with easy bruising and hematuria; white blood cell count of 33 × 10^9/L; monocyte count of 4.95 × 10^9/L; platelet count of 35 × 10^9/L; prothrombin time of 17.1 seconds; APTT, 71 seconds; fibrinogen level of 1.74 g/L; D-dimer of 80 000 µg/L FEU; indicative of disseminative intravascular coagulation (DIC). The blood film (panel A; May-Grünwald-Giemsa stain, 100× objective, original magnification ×1000) showed vacuolated blasts, monocytosis, dysplastic neutrophils, red cell fragments, and thrombocytopenia. Bone marrow smear (panel B; May-Grünwald-Giemsa stain, 100× objective, original magnification ×1000) showed large blasts and hypogranulated granulocytes; by flow cytometry, blasts (41%) were positive for CD34, CD33, CD15, CD38, and cMPO. Molecular karyotyping identified a complex genome with chromothrypsis within the 11q23/qter segment, resulting in amplification of many genes, including the *MLL* gene. Fluorescence in situ hybridization analysis detected *MLL* amplification and mapped it at the hrs (homogenously stained) regions (panel C; 100× objective, original magnification ×1000; DAPI staining). Next-generation sequencing identified *TP53* p.Arg248Gln (VAF 98%), *JAK2* p.Val617Phe (VAF 64%), and *BCORL1* p.Pro482GlnfsTer16 (VAF 18%) variants. The patient was treated with daily blood products, venetoclax and azacytidine. She died 2 weeks later with refractory leukemia and persistent DIC.

Acute myeloid leukemia with *MLL* amplification is associated with elderly patients, *TP53* mutation, complex karyotype, frequent DIC, an aggressive clinical course, poor response to chemotherapy, and extremely short survival. Clinical trial approach is warranted.

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