blood work
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Pure erythroid leukemia evolved from *JAK2*^{V617F}-positive polycythemia vera: blast phase transformation along erythroid line

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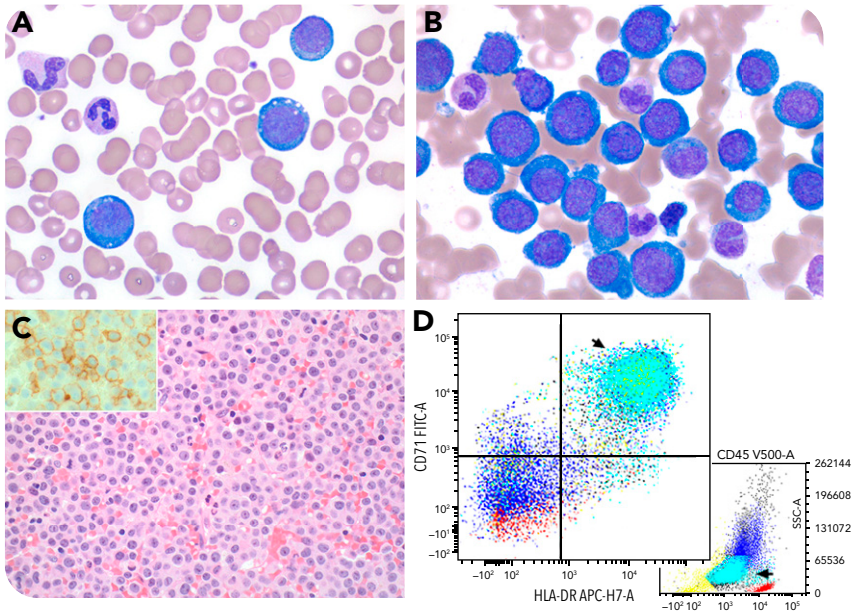

A 76-year-old man diagnosed with *JAK2*^{V617F}-positive polycythemia vera (PV) had his blood counts maintained well with phlebotomy and hydroxyurea. Two years later, he complained of increased fatigue and was found to have moderately decreased platelets, which soon progressed to severe thrombocytopenia and mild anemia (leukocytes, 4.9 × 10^9/L; hemoglobin, 119 g/L; platelets, 29 × 10^9/L). Blood smear showed circulating "blasts" with deeply basophilic cytoplasm with vacuoles (panel A: ×1000). Bone marrow examination exhibited markedly increased "blasts" with morphology suggestive of proerythroblasts (panel B: aspirate smear, ×1000; panel C: core biopsy, ×400). Immunohistochemistry demonstrated membrane staining for E-cadherin (panel C, inset: ×400). Flow cytometry detected 66% "blasts" that were negative-dim for CD45, expressed bright CD71, HLA-DR (panel D), and dim CD33, and was negative for CD34 and CD117. Cytogenetics demonstrates complex karyotype: 41,XY,-3,-4,add (7)(q22),+del(11)(p11.2),-13,-14,-16,17[2]/40,idem,der(18;21)(q10;q10)[5]. Genomic sequencing analysis revealed pathogenic mutations on *TP53*: (NM_000546)c.517G>A(p.Val173Met) (62.3%), *TET2* (NM_001127208)c.3861delT(p.Phe1287LeufsTer76) (19.5%), and *TET2* (NM_001127208)c.5422A>T(p.Arg1808Ter) (26.9%), besides *JAK2*^{V617F} (61.2%). The diagnosis of pure erythroid leukemia (PEL) evolved clonally from PV was established, and chemotherapy was started. The patient died 2 weeks after diagnosis of transformation to PEL resulting from multiorgan failure.

PEL transformed from PV is extremely rare, and pathogenesis is unclear. In our case, the acquisition of a *TP53* mutation likely produced a "double-hit" effect with the *JAK2*^{V617F}-positive clone to drive this leukemic transformation in the erythroid lineage.

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