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Mixed-phenotype acute leukemia, T/megakaryoblastic

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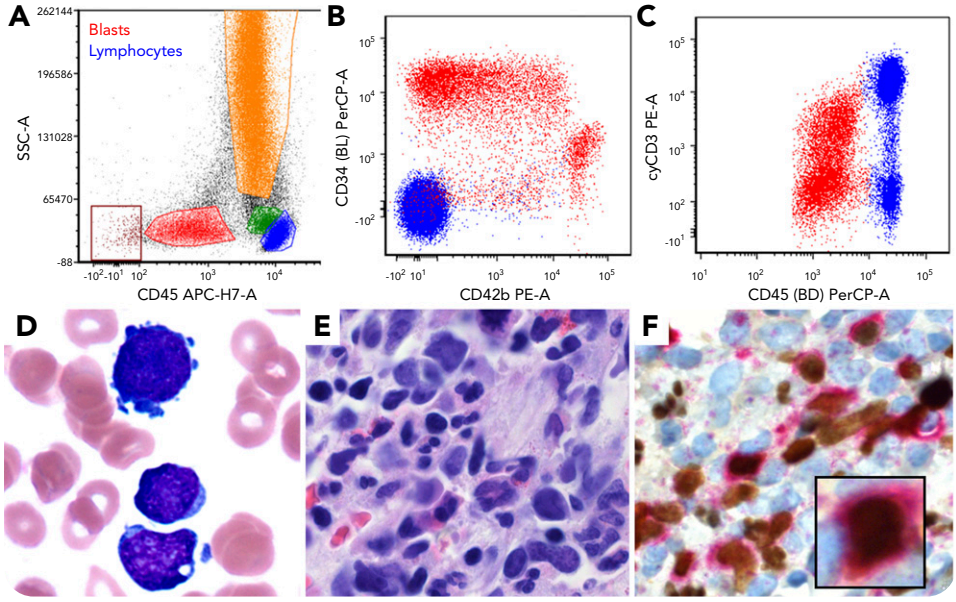

A newborn boy presented with leukocytosis (45 × 10^9/L), anemia (10 g/dL), and thrombocytopenia (12 × 10^9/L). Peripheral blood flow cytometry showed CD45 dim-to-negative blasts (panel A) positive for CD13, CD33 (subset), CD117, CD34, CD42b (panel B; >50% CD34-positive blasts express CD42b), CD41, and cyCD3 (panel C), while negative for CD1a, CD2, sCD3, CD4, CD5, CD7, CD8, CD19, and cyMPO. Bone marrow examination identified a dimorphic blast population (28%) with megakaryoblast and lymphoblast features (panel D; aspirate smear, Wright-Giemsa stain, original magnification ×1000) and fibrosis (panel E; core biopsy, hematoxylin and eosin stain, original magnification ×600). An immunohistochemical double-stain for GATA1 (brown) and CD3 (magenta) showed numerous biphenotypic blasts with megakaryocytic/T-lymphoid differentiation (panel F; original magnification ×600, inset ×1000). The conventional cytogenetic karyotype was 46,XY. Next-generation sequencing identified a novel *CBFA2T3-GLIS3* fusion. He underwent acute lymphoblastic leukemia–directed induction therapy and allogeneic stem cell transplantation but relapsed with pure acute megakaryoblastic leukemia (cyCD3 negative), which was refractory to subsequent treatments.

Mixed-phenotype acute leukemia, T/megakaryoblastic, is exceptionally rare. The case presented here satisfies the criteria for both T-lineage assignment (cyCD3 intensity reaches that of normal T-lymphocytes) and acute megakaryoblastic leukemia (≥1 megakaryocytic marker in >50% blasts) and thus represents one of the first documented examples of this unusual entity.

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