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Acute myeloid leukemia with erythroid and megakaryocytic differentiation associated with Down syndrome

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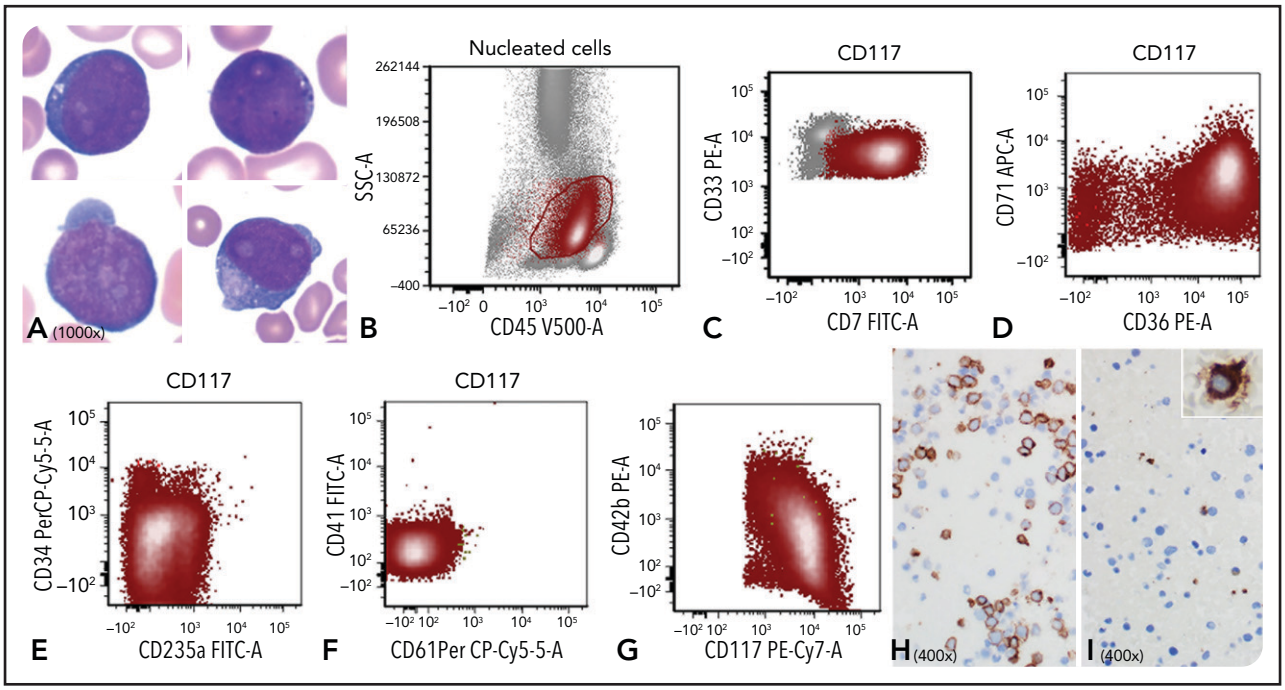

An infant with Down syndrome (DS) was diagnosed with transient abnormal myelopoiesis (TAM) at 1 week of life when complete blood count showed moderate thrombocytopenia (platelets 53K/μL) and 4% blasts; however, next-generation sequencing (NGS) identified no GATA1/2 or other mutations. TAM resolved spontaneously. The infant presented with pancytopenia (white blood cell count 3.9 K/μL; hemoglobin 11.6 g/dL; platelets 15 × 10^3/μL) and 5% circulating blasts 11 months later. Bone marrow aspirate showed 27% blasts with basophilic cytoplasm, few with cytoplasmic blebs (panel A; May-Grünwald-Giemsa stain, original magnification ×1000). Flow cytometry identified a single $CD117^+CD34^-$ blast population (highlighted in brown) with erythroid and megakaryocytic immunophenotype: $CD7^+CD13CD33^+CD36^{++}CD42partial^+CD45dim^+CD56partial^+$ $CD71^+CD235adim^+$ and HLA-DR-cytoCD3$^-$CD19$^-$CD41$^-$CD61$^-$MPO− (panels B-G). Immunostaining highlights numerous $CD117^+CD71^+$ blasts (panel H; original magnification ×400) with a subset expressing E-cadherin. CD61 highlights micromegakaryocytes only (inset, panel I; original magnification ×400). Cytogenetics revealed 48,XX,+8, +21c[18]/47,XX,+21c[2], and NGS showed mutations in GATA1, RAD21, and SUZ12.

Approximately 20% to 30% of infants with DS with prior TAM develop acute myeloid leukemia (AML) in 1 to 4 years with many (>50%) showing megakaryoblastic differentiation. Erythroid or erythroid/megakaryoblastic differentiation is less frequent. The morphologic and immunophenotypic findings in this case support AML with mixed erythroid and megakaryocytic differentiation. Acquisition of additional cytogenetic abnormality (trisomy 8) and mutations in GATA1 along with cohesins (RAD21) indicates progression to AML in DS.

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