blood work
images in hematology

NUP98 rearrangement in B lymphoblastic leukemia with hyperdiploidy

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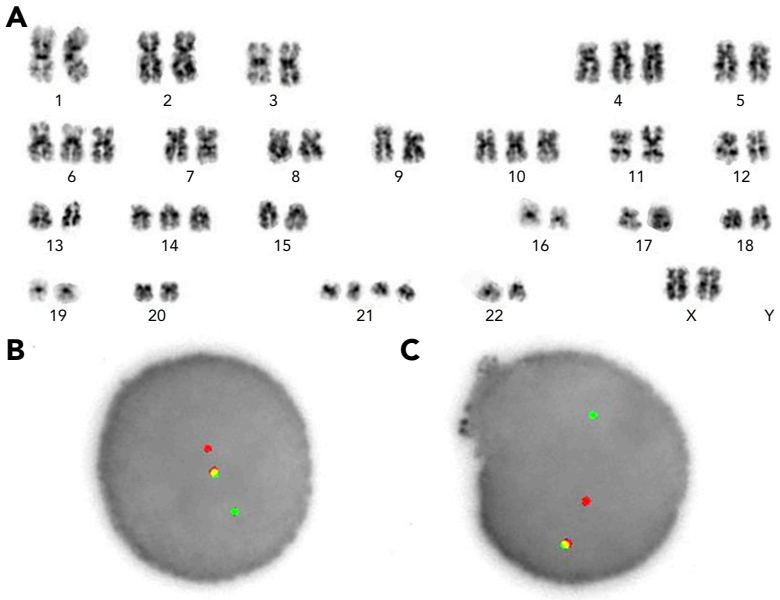

A 2-year-old girl presented with 1 month of fatigue, intermittent fever, and severe bicytopenia (white blood cell count, 4.7 × 10^9/L; hemoglobin, 3.1 g/dL; platelet count, 9 × 10^9/L). Peripheral blood (PB) and bone marrow (BM) showed increased B lymphoblasts (positive, CD10, CD19, CD22, CD34, CD38, dim CD45, CD58, CD123, and HLA-DR; negative, CD20, κ/λ light chains, and T/myeloid markers). Karyotype was 52,XX,+4,+6,+10,add11(p15),+14,+21,+21[16]/46;XX[4] (panel A; objective, ×100, total magnification, ×1000). The structural alteration add11(p15) prompted fluorescence in situ hybridization analysis with the breakapart probe for the nucleoporin 98 gene (*NUP98*) (11p15.4) (5′*NUP98*, red; 3′*NUP98*, green; Cytocell). *NUP98* rearrangement was detected in 13.6% of the interphase cells (panels B-C, split red and green signals; objective, ×100, total magnification, ×1000). An attempt to identify the partner gene failed. She was diagnosed with B lymphoblastic leukemia (B-ALL) with hyperdiploidy.

Hyperdiploid B-ALL is characterized by a numerical increase in chromosomes (>50) and typically does not involve structural alterations, including translocations. *NUP98* rearrangement has been reported in myeloid neoplasms and T-ALL, with poor prognosis. The detection of a *NUP98* translocation in B-ALL is a novel finding, which may have contributed to the delayed blast clearance after induction therapy. Minimal residual disease detection by flow cytometry was positive on day 8 (PB, 0.31%) and day 29 (BM, 0.05%). The patient's status was changed from standard- to high-risk ALL. She has been in remission since consolidation (AALL1131).

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