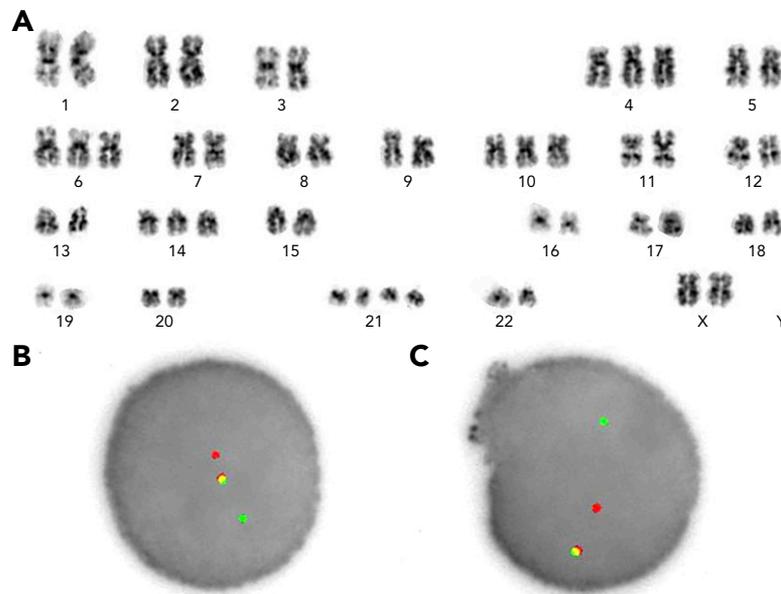


## NUP98 rearrangement in B lymphoblastic leukemia with hyperdiploidy

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A 2-year-old girl presented with 1 month of fatigue, intermittent fever, and severe bicytopenia (white blood cell count,  $4.7 \times 10^9/L$ ; hemoglobin, 3.1 g/dL; platelet count,  $9 \times 10^9/L$ ). 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,  $\kappa/\lambda$  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,  $\times 100$ , total magnification,  $\times 1000$ ). 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,  $\times 100$ , total magnification,  $\times 1000$ ). 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).