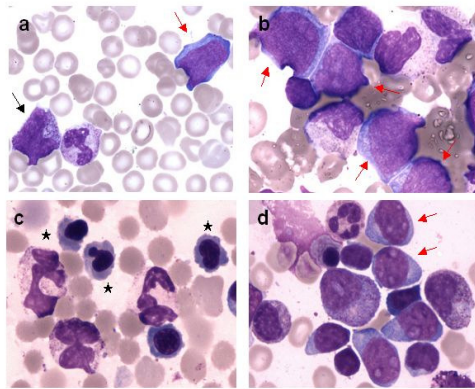


Supplementary Figures

Supplementary figure 1: Morphological evidence of JMML on May-Grunwald-Giemsastained hematopoietic smears from infants with NS-JMML. (a) Blood from patient LMMJ-4 showing hyperleukocytosis with the presence of undifferentiated myeloid blasts and myeloid precursors in peripheral blood. (b) Bone marrow from patient LMMJ-6 showing 12% undifferentiated myeloid blasts. (c) Bone marrow from patient LMMJ-3 showing erythroid hyperplasia (G/E<1) followed three months later (d) by blast crisis with 19% undifferentiated myeloid blasts in the marrow. Multilineage dysplasia together with an excess of undifferentiated myeloid blasts was observed in all patients. Red arrows show undifferentiated myeloid blasts, while black arrows and black stars indicate myeloid precursors and erythroblasts, respectively.



Supplementary figure 2: Analysis of clonality using X-chromosome inactivation analysis. Fluorescent PCR products were separated by capillary electrophoresis. The unmethylated allele (indicated by a black arrow) almost completely disappears after digestion with the methyl-specific restriction enzyme HHA1. Allelic ratios were calculated as follows: $R = [(A1/A2)_{\text{HHA1 digested}}] / [(A1/A2)_{\text{undigested}}]$. The ratio was 39 as compared to 1.2 in a normal control displaying the same allelic pattern as Patient LMMJ-13 (AR locus), and 6 as compared to 1.1 in a normal control for Patient MPD-6 (ZDHHC15 locus), demonstrating skewed X chromosome inactivation in these two patients.

