

## CASE REPORT

# Retrospective Study of Three Cases of Congenital Leukemia with Clinical Presentations and Particular Cytogenetic Abnormality

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## SUMMARY

**Background:** The goal is to assess the prognosis of cytogenetic abnormality, because cytogenetic abnormality is rarely encountered in clinical practice.

**Methods:** We retrospectively report three cytogenetic abnormality cases with clinical, cytogenetic, and genetic characteristic.

**Results:** All cases occurred within one month of birth and had prominent hepatosplenomegaly, including acute myeloid leukemia (case 1, case 2) and acute leukemia (case 3). Moreover, case 1 appeared as leukemia cutis at birth, case 2 was born with respiratory distress, and both showed hyperleukocytosis. The R-banded karyotype detected cytogenetic abnormality in three cases, case 1 with 46,XY,t(8;12)(q21;p13), case 2 with 47,XX,+21 and case 3 with 46,XY,t(6;X)(q22;p12), respectively. Especially in case 1, reverse transcription–polymerase chain reaction analysis showed *MLL-AF10* rearranged.

**Conclusions:** In our studies, all cases had not received chemotherapy and survived about 1 - 2 months. It suggests that cytogenetic disorders are closely related to disease development and likely result in fatal outcome if untreated. Thus, we proposed that a proper treatment decision is urgently needed in congenital leukemia.

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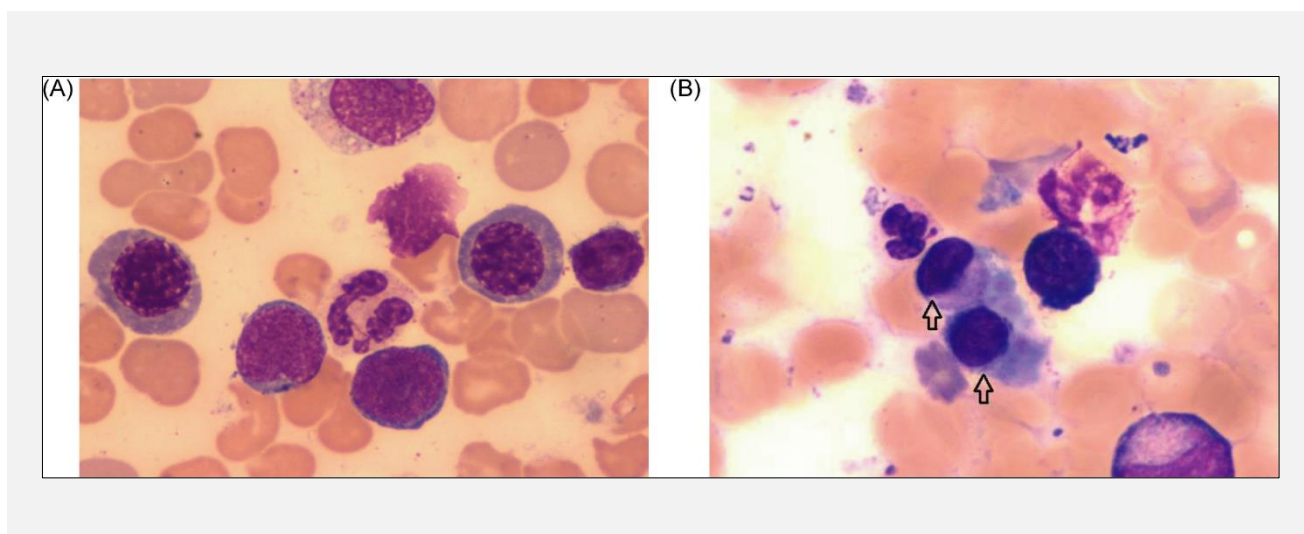
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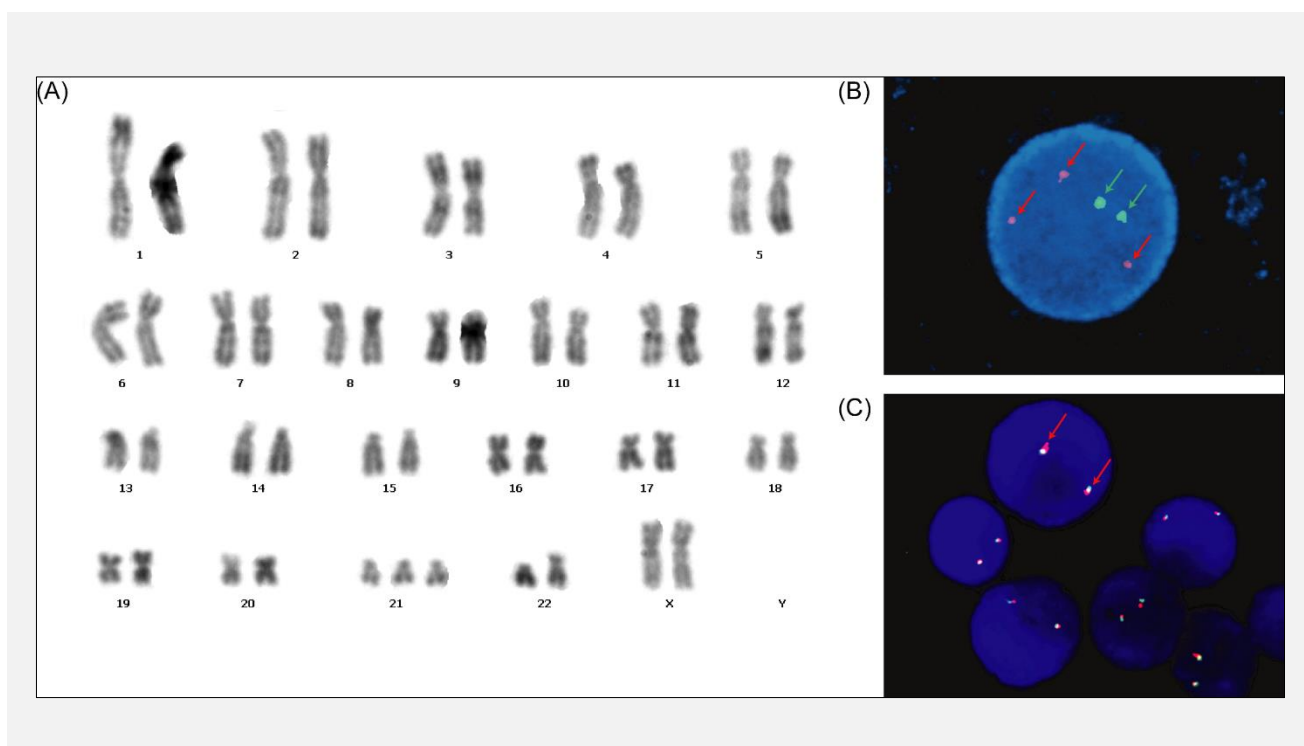
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### Supplementary Figures



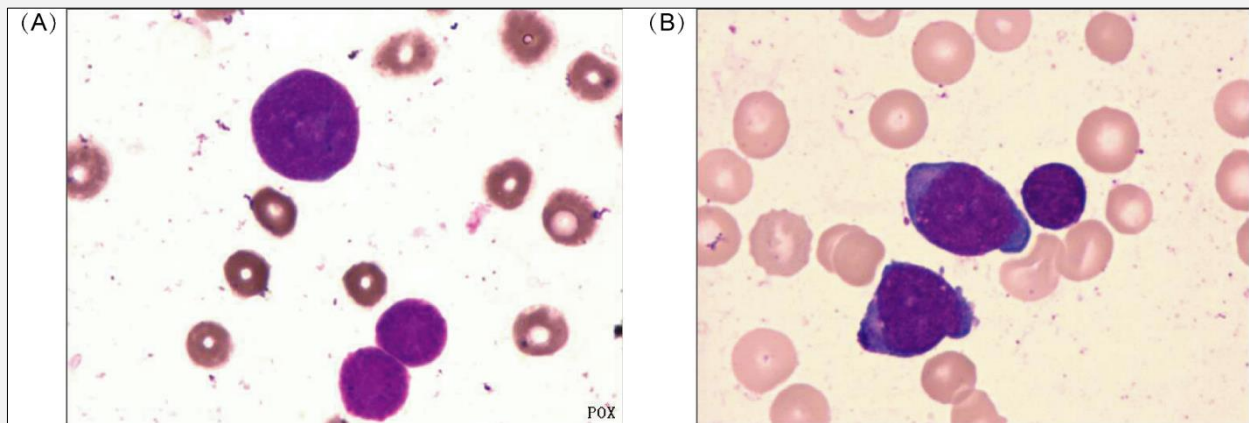
**Figure S1. Bone marrow morphologic staining features (case 2).**

(A): BM blasts had obvious irregular nuclei, the cytoplasm is grey-blue, and filled with granules. (B): BM smear shows small megakaryocyte (arrow).



**Figure S2. Karyotype and fluorescence *in situ* hybridization (FISH) analysis (case 2).**

(A): karyotype (R-banding): 47,XX,+21. (B): FISH analysis with the GLP 13/21 dual color probe (located at 13q14/21q22). There were two green signals (green arrow) on native chromosome 13×2, three red signals on native chromosome 21×2 and extra chromosome 21 (red arrow), respectively. (C): FISH analysis with GLP *MLL* probe (located at 11q23). The picture shows two yellow signals (red arrow).



**Figure S3. Bone marrow morphologic staining features (case 3).**

**(A):** The bone marrow cells showed negative for peroxidase staining. **(B):** BM blasts were obvious irregular, the cytoplasm is grey-blue, and had granules.