

Acute Leukemia of Mixed B/Myeloid Lineage with Dual *BCR::ABL1* and *CBFβ::MYH11* Fusion Genes

Lu X. et al.: Acute Leukemia of Mixed B/Myeloid Lineage with Dual *BCR::ABL1* and *CBFβ::MYH11* Fusion Genes

Xulin Lu¹, Jigang Xiao¹, Xiaojin Cai¹, Chengwen Li¹, Zhijian Xiao^{1,2}

¹State Key Laboratory of Experimental Hematology, National Clinical Research Center for Blood Diseases, Haihe Laboratory of Cell Ecosystem, Chinese Academy of Medical Sciences & Peking Union Medical College, Tianjin, P.R. China

²Tianjin Institutes of Health Science, Tianjin, P.R. China

Zhijian Xiao, M.D., State Key Laboratory of Experimental Hematology, National Clinical Research Center for Blood Diseases, Haihe Laboratory of Cell Ecosystem, Chinese Academy of Medical Sciences & Peking Union Medical College; Tianjin Institutes of Health Science, Tianjin, P.R. China
zxiao@ihcams.ac.cn

June 12, 2025

July 17, 2025

Keywords: Acute leukemia, mixed B/myeloid lineage, *BCR::ABL1*, *CBFβ::MYH11*

A 14-year-old male presented with dizziness, nausea, and fever. Blood tests revealed: WBC $89.71 \times 10^9/L$, HB 98g/L, and PLT $392 \times 10^9/L$. Bone marrow aspirate and peripheral blood revealed 57% and 34% blasts separately (Figure1 A, B). Flow cytometry (FCM) revealed two abnormal cell populations: one expressed CD34, CD123, TDT, CD38, HLA-DR, CD33, CD13, CD117 (strong), CD7, CD36 (partial); the other expressed CD19, cCD79a, CD22, TDT, CD34, CD38, HLA-DR, CD13, CD33, CD123, and CD117 (partial) (Figure1 C). Cytogenetic analysis showed: 46,XY,t(9;22)(q34.1;q11.2),inv(16)(p13.1q22)[20] (Figure1 D). Fluorescence in situ hybridization (FISH) revealed positive of *BCR::ABL1* and *CBFB* rearrangement (Figure1 E, F). Whole-transcriptome RNA sequencing of fusion genes revealed existence of *CBFβ::MYH11*, *BCR::ABL1* (p210), and *ABL1::BCR*. After one cycle of chemotherapy including vincristine (VCR), daunorubicin (DNR), PEG-asparaginase (PEG-asp), prednisone (Pred) concurrently with dasatinib, morphologic complete response was achieved and MRD negative by FCM, but both fusion signals remained positive by FISH.

Considering the splenomegaly and the *BCR::ABL1* fusion signal detected in lobulated granulocytes, this case was deemed to have originated from the blast crisis phase of chronic myeloid leukemia (CML-BC)[1,2]. Cases of acute leukemia of mixed B/myeloid lineage, along with

BCR::ABL1 and *CBFβ::MYH11*, have rarely been reported previously. Early use of tyrosine kinase inhibitors (TKIs) in conjunction with hematopoietic stem cell transplantation may improve outcomes [3].

Ethics

Informed Consent: No personally identifiable patient information is included in this publication and informed consent was not required.

Footnotes

Authorship Contributions: Data Collection or Processing: X.L.; Writing: X.L., J.X.; Literature Search: X.C., C.L.; Analysis or Interpretation: Z.X.

Conflict-of-Interest Statement: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

Running Head: Acute leukemia with *BCR::ABL1* and *CBFβ::MYH11*

References

1. Valentina R. Minciocchi, Rahul Kumar, Daniela S. Krause. Chronic Myeloid Leukemia: A Model Disease of the Past, Present and Future. *Cells*. 2021; 10:117-139.
2. Cedric Pastoret, Roch Houot. "Chronic myelogenous leukemia in primary blast crisis" rather than "de novo BCR-ABL1-positive acute myeloid leukemia". *Clinical Case Reports*. 2017; 5(6): 757-760.
3. Stephanie Sembill, Maria Ampatzidou, Sonali Chaudhury, Michael Dworzak, Krzysztof Kalwak, Axel Karow, Alexander Kiani, Manuela Krumbholz, Maaike Luesink, Nora Naumann-Bartsch, Barbara De Moerloose, Michael Osborn, Kirk R. Schultz, Petr Sedlacek, Fiorina Giona, Christian Michel Zwaan, Hiroyuki Shimada, Birgitta Versluijs, Frederic Millot, Nobuko Hijiya, Meinolf Suttorp, Markus Metzler. Management of children and adolescents with chronic myeloid leukemia in blast phase: International pediatric CML expert panel recommendations. *Leukemia*. 2023; 37:505-517.

