(PB2633) SFMBT1-RELATED GENE FUSIONS IDENTIFIED IN PATIENTS WITH B-CELL ACUTE LYMPHOBLASTIC LEUKEMIA

Topic: 02. Acute lymphoblastic leukemia - Clinical

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Background

Gene fusions are major molecular abnormalities in B-cell acute lymphoblastic leukemia (B-ALL). They not only serve as markers for molecular subtyping and prognosis but also are valuable indicators for minimal residual disease (MRD) monitoring.

Aims

This study aimed to identify novel fusion genes in B-ALL patients.

Methods

Diagnostic bone marrow aspirates from 72 adult B-ALL patients were subjected to transcriptome sequencing. The expression levels of gene fusions mRNAs were evaluated using real-time quantitative PCR (RQ-PCR).

Results

Novel SFMBT1-related gene fusion mRNAs were detected in 2 (2.8%) patients, namely SFMBT1::LTF and SFMBT1::STIMATE. SFMBT1, LTF and STIMATE are all located on the short arm of chromosome 3 at band 21(3p21). Moreover, both fusions had the same two breakpoints in SFMBT1 gene within the 5'UTR (chr3: 53046460 and 53045816), one in the promoter region and the other in exon 1. Both patients were classified as standard-risk. They achieved complete remission after one course of induction therapy but both subsequently developed hematologic relapse following the second course of standard consolidation therapy. Furthermore, the fusion mRNA levels during treatment were correlated with the proportion of blast cells (Figure 1) which indicated that fusion mRNA is specifically expressed in the patients' leukemic cells and may serve as a molecular marker for MRD monitoring.

Summary/Conclusion

The recurrent SFMBT1-related gene fusions may be associated with poor prognosis in B-ALL patients, while their underlying mechanisms warrant further investigation. (Grant: The National Natural Science Foundation of China (82370160)).

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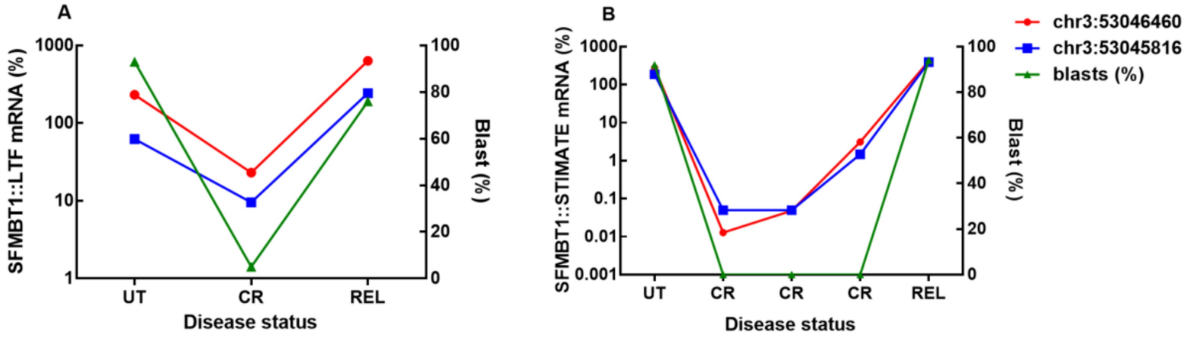


Figure 1. RQ-PCR analysis of SFMBT1 fusion mRNA levels at diagnosis and after treatment. (A) SFMBT1::LTF fusion and (B) SFMBT1::STIMATE fusion.

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