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The e8a2 fusion transcript in B lymphoblastic leukemia with *BCR-ABL1* rearrangement

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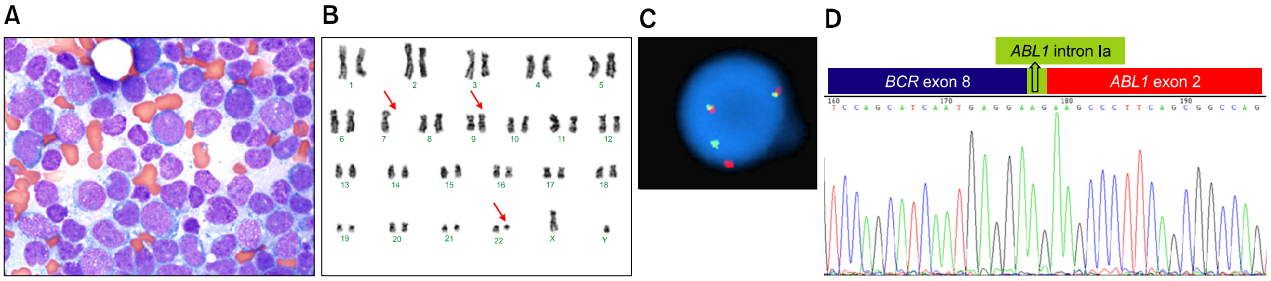

A 73-year-old man presented with fever. The peripheral blood findings were as follows: hemoglobin, 11.7 g/dL; platelets, 55×10^9/L; and WBC count, 95.8×10^9/L (blasts, 94%). Bone marrow aspiration revealed the increasing replacement of hypercellular marrow by blasts showing dispersed chromatin and prominent nucleoli; they accounted for 93% of all nucleated cells **(A)**. Flow cytometric analysis revealed that the blasts were positive for CD10, CD19, CD34, CD45, CD 13, TdT, cytoplasmic CD22 and CD33, and negative for CD117, CD65, CD15, CD14, CD41, CD20, CD2, CD3, CD5, CD7, cytoplasmic CD3 and CD56, and Sm IgM. Chromosomal study showed 45,XY,-7,t(9;22)(q34;q11.2) in 19 of the 20 metaphase cells analyzed **(B)**. Fluorescence *in situ* hybridization using *BCR-ABL1* dual-color, dual-fusion probe showed abnormal signal patterns in 95.5% of the examined nuclei **(C)**. The patient was diagnosed with B lymphoblastic leukemia with *BCR-ABL1* rearrangement. A multiplex reverse transcriptase-PCR analysis for the detection of *BCR-ABL1* rearrangement and subsequent cloning and sequencing analyses confirmed a breakpoint within exon 8 of *BCR* (ENSE00001755753) and 2 nucleotides upstream of exon 2 of *ABL1* (ENST00000984287), resulting in a chimeric exon "e8*-AG-e2" **(D)**. Thus, the breakpoint on the mRNA is identical to that on the genomic DNA. The open reading frame is kept intact by this chimeric fusion exon. The e8a2 *BCR-ABL1* transcript has been reported in CML cases, but not in B lymphoblastic leukemia cases. Because the biological and clinical significance of e8a2 fusion transcript in B lymphoblastic leukemia and CML is still unclear, further studies elucidating the molecular mechanisms involved are necessary.

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