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CLINICAL IMAGE

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An unusual case of chronic lymphocytic leukemia with trisomy 12 presenting with prolymphocytic transformation and t(8;21)(q22;q22)

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Abstract

First report of t(8;21)(q22;q22) in a patient with CLL. RUNX1-RUNX1T1 fusion gene resulting from the translocation may have played a role in the prolymphocytic transformation.

KEYWORDS

hematology, oncology

An 81-year-old man with 20-year history of chronic lymphoid leukemia/small lymphocytic lymphoma (CLL/SLL) with trisomy 12 presented with weakness. CBC showed WBC 60.2 × 10^9 cells/L, hemoglobin 13.2 g/dL, and platelet count of 95 × 10^9 cells/L. Imaging revealed lymphadenopathy and splenomegaly. Peripheral blood and marrow aspirate showed prolymphocytes (Figure 1A-B). Cytogenetics showed a t(8;21)(q22;q22.1) and trisomy 12 in six out of twenty metaphases (Figure 2A).

Chronic lymphoid leukemia/small lymphocytic lymphoma is an indolent lymphoproliferative disorder that can transform into an aggressive malignancy with unfavorable prognosis in about 5%-10% of patients.[1,2] Transformation can be prolymphocytic or into diffuse large cell lymphoma,

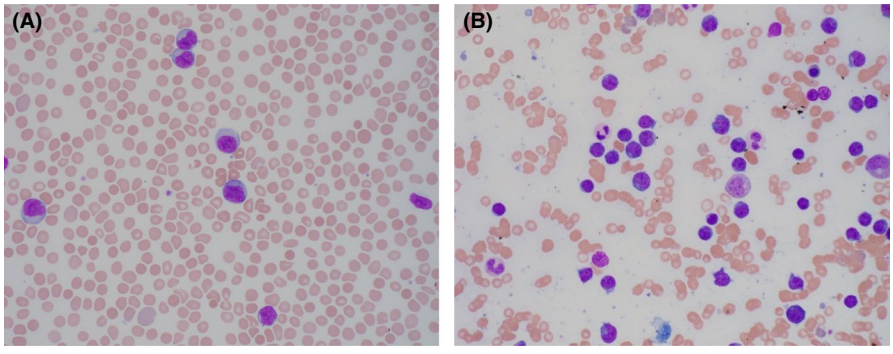

FIGURE 1 Peripheral blood showing prolymphocytic transformation to CLL cells (1A) and bone marrow biopsy with transformation of CLL to prolymphocytic leukemia (1B)

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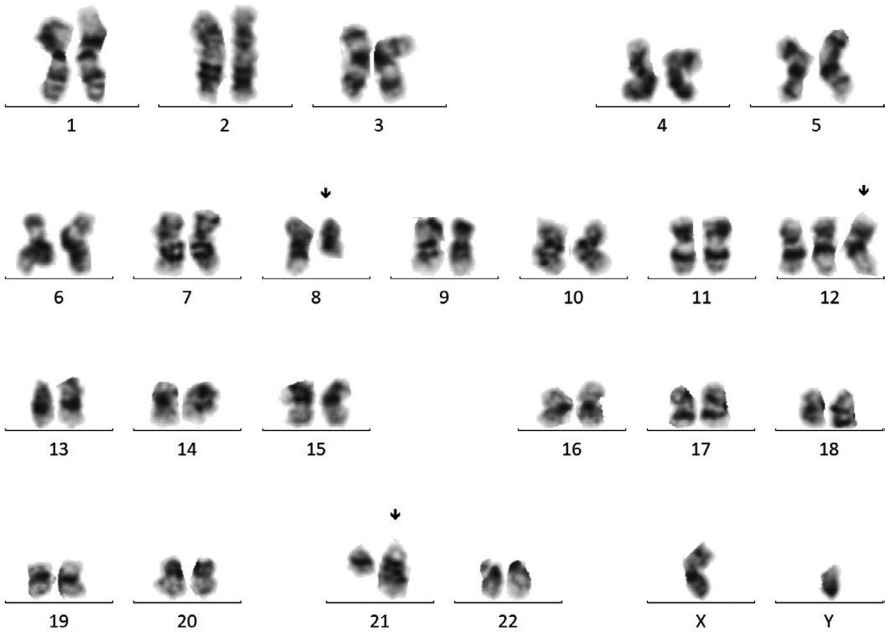

FIGURE 2 Cytogenetics showed an abnormal male karyotype with six out of twenty metaphases showing a translocation between chromosomes 8 and 21 in addition to trisomy 12 (2A)

also known as Richter transformation.[3-7] There are no known cases to date of transformation to prolymphocytic leukemia with t(8;21)(q22;q22.1), which is typically observed in acute myeloid leukemia.[8,9] The t(8;21) results in a fusion oncoprotein between RUNX1 and ETO.[10] RUNX1-RUNX1T1 fusion leads to disruption of normal function of the core-binding factor in the regulation of hematopoietic differentiation and maturation. Given that RUNX1 is critical for maturation of a wide range of hematopoietic stem cells and has been implicated in pathogenesis of several myeloid and lymphoid malignancies, we postulate that the t(8;21)(q22;q22.1) played a role in prolymphocytic transformation in this patient.[11-13]

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CONFLICT OF INTEREST

None.

AUTHOR CONTRIBUTIONS

I, MB: am the first author of this publication, and KRK: is the advising author of this publication.

DATA AVAILABILITY STATEMENT

No datasets were generated or analyzed during the current study.

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