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An unbalanced whole-arm translocation t(1;13)(q10;q10) in Burkitt lymphoma

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Introduction

Structural rearrangements involving the long arm of chromosome 1 are common secondary aberrations in Burkitt lymphoma associated with L3 morphology of lymphoblasts. Among unbalanced translocations of 1q, whole-arm translocation involving breakage and reunion of nonhomologous chromosomes at their centromeres is relatively rare and the clinical importance of whole-arm translocations remains to be completely clarified. We describe here an unbalanced whole-arm translocation t(1;13)(q10;q10) in a case of Burkitt lymphoma.

Case Report

A 15-year-old male sought care for epigastric pain. At admission, peripheral blood tests revealed a white blood cell count of 12.33x10^9/L, hemoglobin of 15.5 g/dL, and platelet count of 161x10^9/L. Computed tomography images showed hepatosplenomegaly with innumerable tiny nodules and mesenteric lymphadenopathy. Core biopsy of the nodules confirmed Burkitt lymphoma. Bone marrow aspiration and biopsy analysis from both sides revealed normocellular marrow with significant infiltration of atypical lymphocytes, the majority of which were small to medium sized, with small amount of vacuolated basophilic cytoplasm and distinct nucleoli (Fig.1A). Immunophenotyping of the atypical lymphocytes by flow cytometry identified B-cell phenotype with expression of CD10, CD19, CD20, CD79a, and lambda-restricted surface immunoglobulin. In addition to the primary aberration t(8;14)(q24;q32), der(1;13)(q10;q10) was detected by cytogenetic study of his bone marrow sample (Fig.2A, B). By acquisition of the long arm of chromosome 1, the der(1;13)(q10;q10) resulted in trisomy 1q. Fluorescent in situ hybridization (FISH) using dual-color dual-fusion MYC (8q24) orange/IGH (14q32) green probes (Kreatech Diagnostics, Amsterdam, Netherlands) demonstrated t(8;14)(q24;q32) (Fig.2C)

Based on the morphology and the immunophenotype along with cytogenetic data, a diagnosis of Burkitt leukemia was made. The patient successfully completed induction chemotherapy with cyclophosphamide, vincristine, and prednisolone (COP).

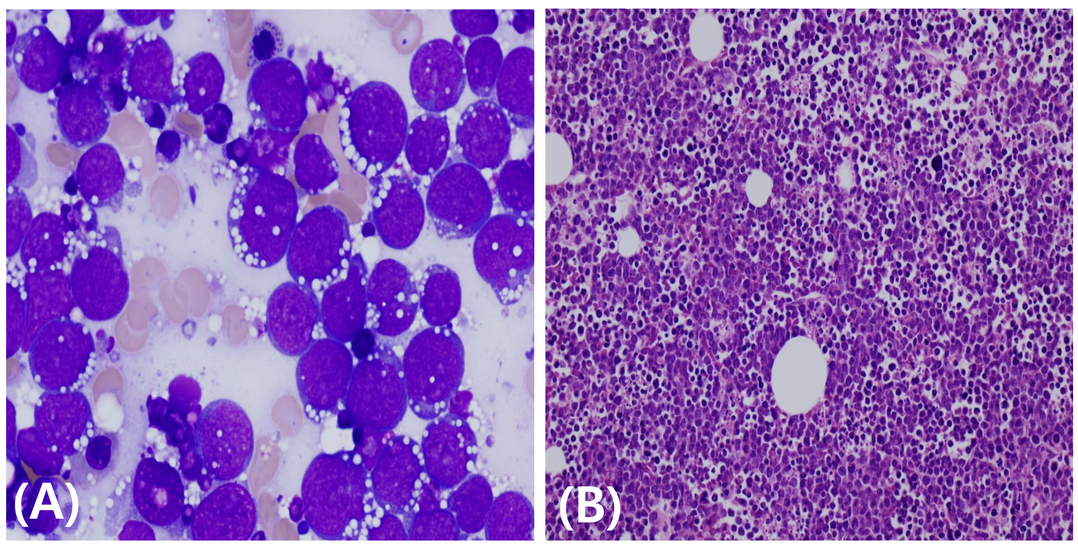

Fig. 1 (A) Bone marrow aspiration smear at the diagnosis of Burkitt lymphoma (Wright-Giemsa stain, x 1,000) (B) H&E section showing sheets of Burkitt cells in the marrow space (x 400)

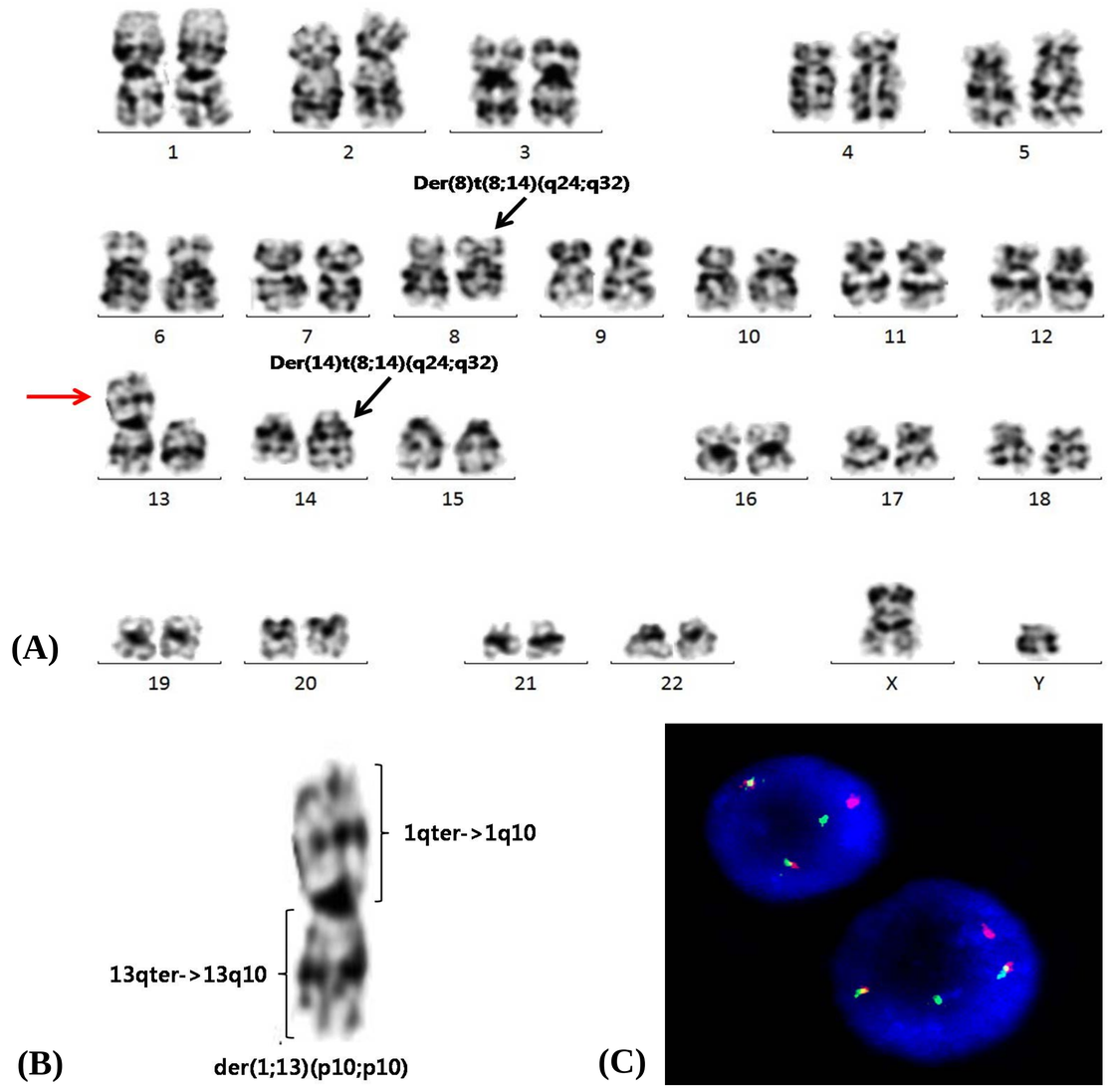

Fig. 2. (A) Giemsa-banding karyogram of bone marrow cells: 46,XY,+1,der(1;13)(q10;q10),t(8;14)(q24;q32). The red arrow indicates the derivative chromosome 13. (B) Karyotype showing der(13)t(1;13)(p10;p10). (C) Interphase FISH using IGH/MYC probes shows one normal IGH (green) signal, one normal MYC (orange) signal, and two fusion signals indicating a t(8;14)(q24;q32).

Discussion

Most 1q abnormalities consist of partial duplications of 1q and have usually been reported to be associated with a poor clinical outcome in Burkitt lymphoma. However, a whole-arm translocations of chromosome 1q is unusual and so far poorly understood finding. We hypothesize that the number and the type of partner chromosomes, the breakpoints involved in the rearrangements, and/or the presence of additional aberrations may have a prognostic impact, but confirmation of this assumption will require further cases to be studied. We believe that this case represents an acquired cytogenetic abnormality which has not yet been described in Burkitt lymphoma.

Reference

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