A case of CML-like Disease with t(8;22)(q24;q11)

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Dear Editor

Chronic myelogenous leukemia (CML) is characterized in 85-90% of cases by the presence of the Philadelphia (Ph) chromosome and BCR-ABL fusion gene.1 A further 5-10% of cases have other translocations, most commonly complex variants that involve one or more chromosomal regions in addition to bands 9q34 and 22q11, but also simple variants that typically involve 22q11 and a chromosome other than 9. There are a few reports regarding observation of t(8;22) in patients with CML-like disease.2-6

We report a case of CML-like disease with t(8;22) who achieved hematological remission with hydroxyurea and Imatinib. A 27-year-old Iranian male presented with fatigue and malaise. Physical examination revealed bilateral axillary lymphadenopathy and huge splenomegaly. Peripheral blood smear showed hyperleukocytosis with shift to the left, basophilia, and eosinophilia. Bone marrow aspiration and biopsy was in accordance with CML in chronic phase. Cytogenetic study revealed t(8; 22)(q24; q11) in all 20 metaphases analyzed. The BCR-ABL fusion was positive which was proved to be falsely positive due to BCR gene disruption. (Figure 1A).

Bone marrow FISH study using D-FISH probes were negative for the BCR-ABL fusion in 200 interphase cells analyzed for this patient (Figure 1B). By D-FISH, the metaphases showed red (ABL) signals on both copies of chromosome 9; one large green (BCR) signal on the normal chromosome 22 with smaller green signals on the der (22) and on the der (8). These findings were consistent with the known karyotype and suggested

[Figure 1: A) 46XY, t(8;22)(q24;q11.2)[20], B) nuclear Fish(ABL×2),(BCR×3)]
that the chromosome 22 breakpoint must be close to, or within, the BCR. He initially received Imatinib mesylate and hydroxyurea which was followed by imatinib alone. He achieved complete hematological remission.

Although t(9;22) is diagnostic for CML, t(8;22) is another known cytogenetic abnormality in patients with CML-like disease. t (8;22) might have been classified cytogenetically as merely a simple variant of the t(9;22). A translocation between the long arms of chromosomes 8 and 22 described both in B-cell acute lymphoblastic leukemia (ALL) and non-Hodgkin lymphomas (NHL), especially in Burkitt lymphoma has been also reported with BCR breakpoint in 22q11.2 in CML-like disease. CML-like disease with t (8; 22) can benefit from TKI therapy.

Conflict of Interest: None declared.

References