CASE REPORT

Interstitial deletion of 9q in a case of acute myeloid leukemia with t(8;21)

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Introduction

The Acute Myeloid Leukemias (AMLs) are a heterogeneous group of diseases with diverse morphologic, cytochemical, immunophenotypic and clinical characteristics. However, specific chromosomal abnormalities correlate with certain morphologic features [1]. Myeloid blasts with the t(8;21)(q22;q22) exhibit unique morphologic features characterized by FAB M2 with abnormal cellular maturation and was the first specific chromosomal rearrangement identified in AML in 1973 [2]. Our report describes a case of AML-M2 with t(8;21) in association with interstitial 9q deletion. Association of interstitial 9q deletion is rare among AML-M2.

Case report

A 16-year-old female was hospitalized in November 2006 with complaints of intermittent fever on and off for two

months associated with fatigue and poor appetite. She had a history of cough of 1-week duration with effort intolerance. On physical examination she was markedly pale. There was no significant lymph node. Chest was clinically clear and the abdomen was soft with no organomegaly. A CT scans chest showed nodular opacities in left upper lobe, middle lobe, lingula and both lower lobes suggestive of fungal etiology.

Cytology

Bone marrow aspirates showed intensely hypercellular particles and cell trails. There was relative erythroid hypoplasia. Myeloid lineage revealed 34% blasts with Auer rods and eosinophilia, which was consistent with AML-M2.

Immunophenotype

The bone marrow cells were tested by flow cytometry using a panel of monoclonal antibodies and an indirect immunofluorescence technique. Immunophenotyping results showed blast expression of myeloid markers CD13, CD33, and CD117 along with other markers such as CD34, CD45 and HLA-DR. The antigen expression profile by flow cytometry in correlation with morphology is consistent with diagnosis of AML M2 of FAB classification.

Cytogenetics

Chromosome preparations were obtained by standard method on bone marrow cells after short-term culture. Karyotype analyses were performed using GTG banding technique [3]. Chromosome analyses of the patient revealed

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Fig. 1 Karyotype showing 46,XX,t(8;21)(q22;q22),del(9)(q21q22) by GTG banding

46 chromosomes, with the presence of reciprocal translocation between chromosomes 8 and 21 at bands q22 and q22 respectively and an interstitial deletion of chromosome 9 from the band q21 to q22 and hence, the karyotype is interpreted as 46,XX,t(8;21)(q22;q22),del(9)(q21q22) according to the guideline for cancer cytogenetics (Fig. 1) [4].

Discussion

Cytogenetics plays a crucial role in risk stratifying children with AML. Children with t(8;21), inv(16) and t(15;17) chromosome abnormalities do not require stem cell transplantation up front as they can be potentially cured with chemotherapy [5,6]. AML with t(8;21) represents a unique clinical and cytogenetics association. Cytogenetically some patients have additional chromosomal abnormalities like the loss of a sex chromosome, trisomy 8, deletion of 9q or a structural aberration of 7q [7]. Our patient had del(9q) in addition to t(8;21). This constantly deleted region has not yet been more precisely defined. It is not known whether deletions of one or more critical gene(s) are involved [8]. del(9q) as a secondary anomaly in t(8;21) has no prognostic

consequence for some workers and is a factor of worse prognosis for others [8,9]. Our patient has been treated with the UK MRC AML 15 protocol [10] with four cycles of chemotherapy and remains in remission since December 2006. t(8;21) with additional chromosomal abnormalities has no adverse prognostic features. However, del(9q) as an isolated abnormality has in general been associated with an adverse outcome.

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