**Case Report Section**

**t(6;17)(p21;p13) and acquisition of the Philadelphia chromosome translocation with p190 BCR-ABL1 transcript during the course of myelodysplastic syndrome**

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**Abstract**

Case report on t(6;17)(p21;p13) and acquisition of the Philadelphia chromosome translocation with p190 BCR-ABL1 transcript during the course of myelodysplastic syndrome.

**Clinics**

**Age and sex**
47 years old male patient.

**Previous history**
Preleukemia The patient had thrombocytopenia for 10 years (platelet counts ranged between 70x10^6/L to 130x10^6/L, for what he had never received any treatment; no previous malignancy; no inborn condition of note

**Organomegaly**
no hepatomegaly , no splenomegaly , no enlarged lymph nodes , no central nervous system involvement

**Blood**

**WBC:** 2.5X 10^9/l  
**HB:** 8.6g/dl  
**Platelets:** 6X 10^9/l  
**Blasts:** 43%  
**Bone marrow:** The bone marrow was hypocellular with 6.5% blasts at presentation. 7 months later, the patient MDS transformed to AML and his bone marrow biopsy showed markedly hypercellular marrow with 69% blasts (0.4% promyelocytes, 2.7% myelocytes, 1.1% neutrophils, 1.8% lymphocytes and 25% erythroblasts).

**Cyto-Pathology Classification**

**Phenotype**
Acute myeloid leukemia without maturation

**Immunophenotype**
Myeloid immunophenotype, (positive for CD13, CD33, CD34, HLDR, CS45 and CD117) with aberrant expression of CD7.

**Diagnosis**
AML with myelodysplasia-related changes.  
Provisional entity: AML with BCR/ABL1

**Survival**

**Date of diagnosis**
08-2011

**Treatment**
After the patient was diagnosed with MDS, therapy with decitabine was administrated but the patient remained cytopenic. At progression chemotherapy combined with imatinib (400 mg/day) was administrated that was escalated to 800 mg/day two weeks later.  
**Complete remission:** no
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Status
Alive

Last follow up: 04-2012

Survival
7+ The patient had traveled to his native country for further treatment. months

Karyotype

Sample bone marrow

Culture time 24h

Banding G-banding

Results
47,XY,t(6;17)(p21;p13),+8 [10]/46,XY [10] at presentation;
47,XY,t(6;17)(p21;p13),+8,t(9;22)(q34;q11)[18]/48
,XY,t(6;17)(p21;p13),+8,t(9;22)(q34;q11),+10
[2]/46,XY [20 ] at transition

Other Molecular Studies

Technics:
Molecular studies were performed from total RNA extracted from the bone marrow sample using the TriZol LS reagent (Invitrogen, US) according to the manufacturer’s recommendations. cDNA was prepared from 2μg of total RNA with the Superscript II cDNA Synthesis Kit (Invitrogen, US) according to manufacturer’s instructions.

Results:
Quantitative molecular studies performed with GeneXpert (Cepheid, US) were negative for the major BCR/ABL1 transcript. The absence of p210 BCR-ABL1 transcript was confirmed by reverse transcription-polymerase chain reaction analysis; however it showed breakpoint cluster region rearrangement between exons e1 and a2, compatible with the minor p190 BCR/ABL1 transcript. Developing a Philadelphia chromosome, considered a primary anomaly, during the course of MDS is extremely rare phenomenon, described only in few patients (Melo et al., 1994; Onozawa et al., 2003; Advani et al., 2004; Keung et al., 2004). Our study illustrates that the Ph translocation may develop from an existing MDS clone, accompanying or perhaps inducing disease transformation. The availability of preleukemic MDS phase in our patient offers an unique opportunity to analyze the role of BCR-ABL1 in leukemogenesis and the evolution of leukemia clones in hematological malignancies.

Comments
We described a rare t(6;17)(p21;p13) (La Starza et al., 26) in a patient diagnosed with myelodysplastic syndrome that terminated in AML associated with acquisition of the Philadelphia chromosome translocation and the p190 BCR/ABL1 transcript. The patient initially had thrombocytopenia for 10 years, but developed MDS and at that time karyotyping revealed the chromosomal translocation t(6;17)(p21;p13) associated with an extra chromosome 8. The initial myelodysplastic syndrome terminated to acute myeloid leukemia, accompanied by an appearance of a new clone, characterized by a Philadelphia chromosome. Reverse transcription-polymerase chain reaction analysis further revealed the presence of the minor p190 BCR-ABL1 transcript. Developing a Philadelphia chromosome, considered a primary anomaly, during the course of MDS is extremely rare phenomenon, described only in few patients (Melo et al., 1994; Onozawa et al., 2003; Advani et al., 2004; Keung et al., 2004). Our study illustrates that the Ph translocation may develop from an existing MDS clone, accompanying or perhaps inducing disease transformation. The availability of preleukemic MDS phase in our patient offers an unique opportunity to analyze the role of BCR-ABL1 in leukemogenesis and the evolution of leukemia clones in hematological malignancies.
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Figure 2. (A) Representative metaphase of the patient from the time of transition showing the 47,XY,t(6;17)(p21;p13),+8,t(9;22)(q34;q11) karyotype. (B) Fluorescence in situ hybridization with LSI BCR-ABL1 probe (Vysis, IL, US) showing the fusion BCR-ABL1 signal on der(9) and der(22) chromosomes. (C) Ethidium-bromide-stained agarose gel showing RT-PCR-amplified BCR-ABL1 chimaeric transcripts.1 Lanes M, molecular weight marker; lane 1, e1a2 fusion in a patient; lane 2, ABL internal control; lane 3, e1a2-positive cell control; lane 4, negative control; lane 5, negative reaction with p210BCR/ABL primers in a patient; lane 6, p210 CML positive control.

References


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