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Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL INIST-CNRS

Case Report Section

Paper co-edited with the European LeukemiaNet

Translocation t(1;3)(p36;p21) and other aberrations in a case of AML secondary to MDS

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Published in Atlas Database: August 2013

Online updated version : http://AtlasGeneticsOncology.org/Reports/t0103p36p21BrazID100074.html
DOI: 10.4267/2042/53493

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Abstract

Case report on a case of t(1;3)(p36;p21) and other aberrations in a case of AML secondary to MDS.

Clinics

Age and sex
45 years old male patient.

Previous history
Preleukemia MDS-RAEB1 (6,6% blasts BM, normal karyotype) (03/2008), no previous malignancy, no inborn condition of note.

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

Blood

WBC: 101 X 10^9/l
HB: 9,3g/dl
Platelets: 18 X 10^9/l
Blasts: 92% (PB)
Bone marrow: blasts: 87%, red cells: 1% and lymphocytes: 12%

Cyto-Pathology Classification

Immunophenotype
CD15-/+ (35%),CD33-/+ (17%), CD34-/+ (64%), CD45+/++, CD64-/+ weak, CD117+/++, CD123+, HLA-DR+/+++ heterogeneous. Negativity: CD3, CD4,CD8, CD36.

Pathology
Acute Myeloid Leukemia, M2

Survival

Date of diagnosis: 03-2008

Treatment
Allogenic Bone Marrow Transplantation: 1st 04-2008 and 2nd 08-2009. The patient was treated with alkylating agents (cyclophosphamide and Busulfan) during conditioning regimen for the first bone marrow transplantation. For the second one, it was used cyclophosphamide and TBI (total body irradiation).
Complete remission: complete remission was obtained
Treatment related death: no
Relapse: +

Status: Dead

Last follow up: 04-2013

Survival: 49 months

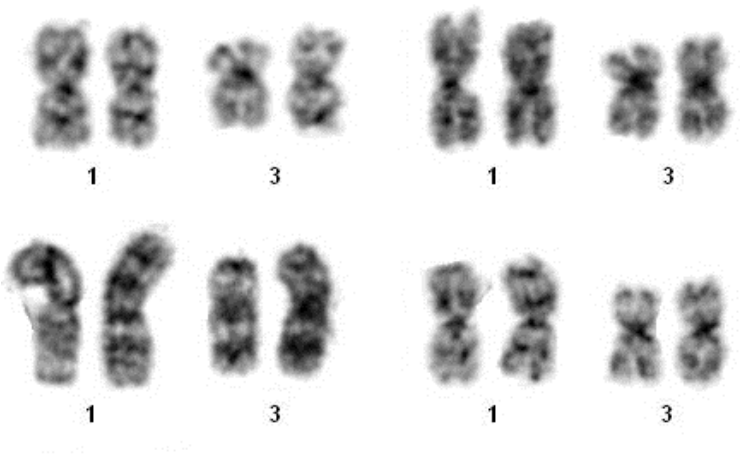
Karyotype

Sample: Bone marrow

Culture time: 24h

Banding: GTG

Results
46,XY,t(1;3)(p36.2;p21),t(4;5)(q31;q35),t(5;16)(q13;p13.3),del(7)(p15),t(13;22)(q13;q13)[20]



Partial karyotypes with G-banding showing the t(1;3)(p36.2;p21)

Comments

The patient was diagnosed as MDS-RAEB1 in 2008. In the same year he underwent a bone marrow transplantation. One year after (2009) he relapsed as sAML. He was submitted to a second HSCT. He was in remission until 2011, when the disease relapsed as AML. In 2012 a third remission was achieved.

In 2013 he relapsed as AML M2 after the last therapy. At this point he presented this complex karyotype: 46,XY,t(1;3)(p36.2;p21),t(4;5)(q31;q35),t(5;16)(q13;p13.3),del(7)(p15),t(13;22)(q13;q13). Data reported in literature suggest that 3p21 is a recurrent treatment-related breakpoint in MDS and AML (Shi et al., 1996). The t(1;3)(p36;p21) is a recurring chromosomal translocation and it was described after treatment with alkylating agent in several cases (Sato et al., 2002). Our patient was also treated with alkylating agents (cyclophosphamide and Busulfan) during conditioning regimen for the first bone marrow transplantation. For the second one, it was used cyclophosphamide and TBI (total body irradiation). Sato et al., 2002 and other reports have described a high frequency of abnormalities of chromosomes 5 and 7 (Smith et al., 2003) as part of a complex karyotype in patients with t(1;3). In our report, we could observe both chromosome 5 involved in different aberrations. It was not possible to prove all the aberrations with FISH (not available in our service) and also we couldn't find t(13;22) in other reports.

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This article should be referenced as such:

Schmid Braz AT, Merfort LW, Jamur VR, Borgonovo T, Azambuja AP, Malvezzi M. Translocation t(1;3)(p36;p21) and other aberrations in a case of AML secondary to MDS. Atlas Genet Cytogenet Oncol Haematol. 2014; 18(3):222-223.