

Case Report Section

Paper co-edited with the European LeukemiaNet

A Case of Myelodysplastic Syndrome with a Translocation t(1;12)(p36;p13)

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Published in Atlas Database: August 2008

Online updated version : <http://AtlasGeneticsOncology.org/Reports/0112BacherID100040.html>
DOI: 10.4267/2042/44549

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Clinics

Age and sex

46 years old male patient.

Previous history

No preleukemia. No previous malignancy. No inborn condition of note.

Organomegaly

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

Blood

WBC: $1.9 \times 10^9/l$

HB: 10.4g/dl

Platelets: $42 \times 10^9/l$

Bone marrow: Hypercellular, trilineage dysplasia, blasts <5%

Cyto-Pathology Classification

Diagnosis

Myelodysplastic syndrome - subtype refractory anemia cytopenia with multilineage dysplasia (MDS - RCMD) according to the WHO classification.

Survival

Date of diagnosis: 03-2008

Treatment: Allogeneic stem cell transplantation from an HLA-mismatched unrelated donor as first-line

strategy (peripheral stem cell transplantation; PBSCT) after a dose reduced conditioning regimen (fludarabine, amsacrine, cytarabine, busulfane) in combination with thymoglobuline.

Complete remission: no

Treatment related death: yes (transplant associated mortality, TRM) (day +15 after allogeneic transplantation). (Cause of death: severe hemolysis after ABO minor mismatched allo-transplantation; suspicion of thrombotic thrombocytopenic purpura (TTP) in association to cyclosporine A for immunosuppression; intestinal bleeding of unclear origin).

Relapse: not applicable due to early death after transplantation.

Status: Dead.

Survival: 3 months from diagnosis of MDS.

Karyotype

Sample: Bone marrow

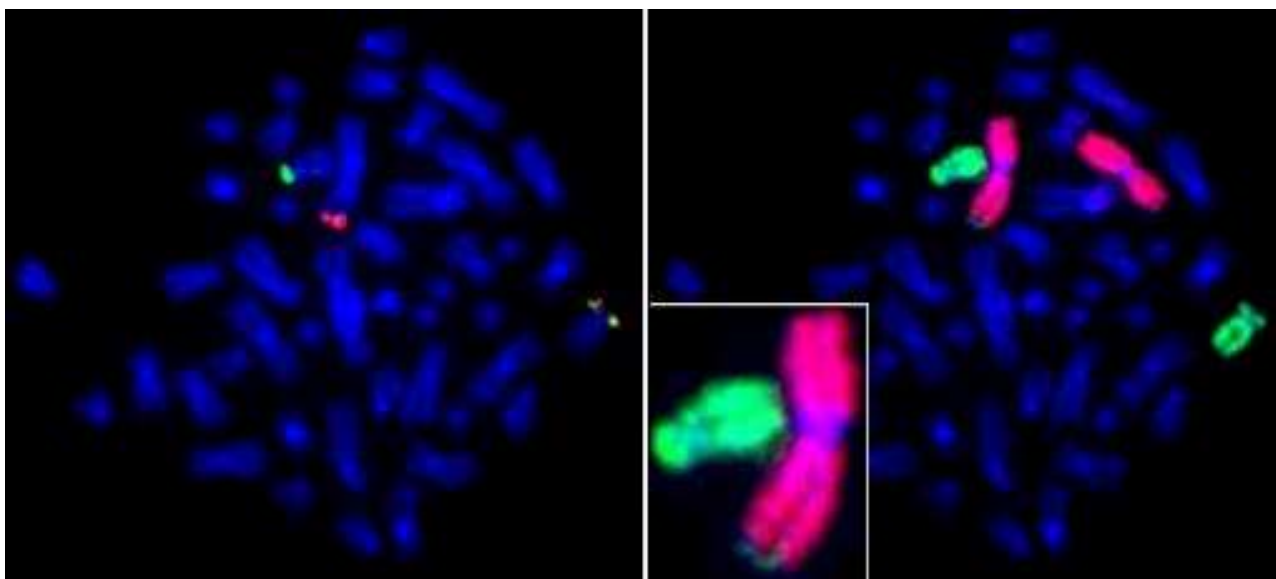
Culture time: 24, 48 hours

Banding: Giemsa

Results: 46,XY,t(1;12)(p36;p13) [9]; 46,XY [11]

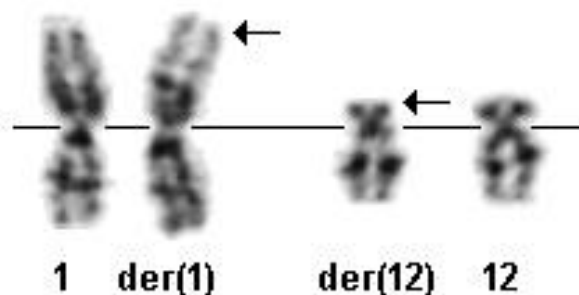
Other molecular cytogenetics technics:

See figure below: Fluorescence in situ hybridization with probes flanking the breakpoints within the ETV6 gene demonstrating an ETV6 rearrangement (left) and in a second hybridization with whole chromosome painting probes for chromosome 1 (red) and chromosome 12 (green) on the same metaphase.



Other molecular cytogenetics results:

No evidence of the FLT3-ITD/LM (internal tandem duplication/length mutation), NRAS-mutation, or MLL-PTD (partial tandem duplication) by polymerase chain reaction (PCR) analyses.



Comments

Two our knowledge so far two cases with a t(1;12)(p36;p13) were described in the literature. The first reported case suffered from chronic myeloid leukemia (CML) (Vassallo et al., 1993). The second patient, a 66-old female, showed MDS in transformation (RAEB-T) and rapidly proceeded to secondary acute myeloid leukemia (AML) (Oedro et al., 2002). The here reported case - a 46 year old male - had MDS in an initial stage (RCMD). Treatment was performed by upfront allogeneic stem cell

transplantation unfortunately followed by severe complications with hemolysis and intestinal bleeding resulting in early transplant-associated mortality.

So far, the prognostic impact of the t(1;12)(p36;p13)/ETV6-TEL cannot be determined. As the respective translocation is difficult to detect in chromosome banding analyses, the true frequency might be higher than actually thought.

Call for Collaborations

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This article should be referenced as such:

Bacher U, Haferlach T, Haferlach C. A Case of Myelodysplastic Syndrome with a Translocation t(1;12)(p36;p13). *Atlas Genet Cytogenet Oncol Haematol*. 2009; 13(8):618-619.