

## Case Report Section

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# inv(8)(p11.2q13) found in a patient with chronic myelomonocytic leukemia that progressed to acute myeloid leukemia

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### Clinics

Age and sex: 67 years old male patient.  
Organomegaly : no hepatomegaly ; splenomegaly ; no enlarged lymph nodes ; no central nervous system involvement.

Previous history: 67 year old man with normal CBC until 02.23.1993, when first noted to have monocytosis (wbc 6900; mono 17%; Absolute Mono Count (AMC) 1173/mm<sup>3</sup>), mild anemia (Hgb 13.4/ nl 13.6-16.7) and thrombocytopenia (121,000/nl 150-400,000). Otherwise clinically well. No significant other medical history. CMML by marrow biopsy 12/2001 (WBC 15,300; Hg 13.0; plt 65,000; AMC 3060/mm<sup>3</sup>). Initial cytogenetic analysis was normal, 46,XY. No treatment for 5 years because of stable counts and no symptoms. He presented with fatigue and purpura December 2006 (see description below)

### Blood

WBC: 30.5 x 10<sup>9</sup>/l; Hb: 13.5 g/dl; platelets: 17,000 x 10<sup>9</sup>/l; blasts: 1%.  
Bone marrow: AML, M4; 80% monocytoid blasts.

### Cytopathology classification

Cytology: M4.

Immunophenotype: Positive for: CD33, CD13, HLA-DR, CD64, CD16, CD14, CD56 (partial), MPO, CD45  
Negative for: CD34, CD2, CD3, CD19, CD20, TdT, cyto CD3, CD7.

Rearranged Ig or Tcr: not done.

Pathology: AML, M4.

Electron microscopy: not done.

Precise diagnosis: AML, M4.

### Survival

Date of diagnosis: 12-2006.

Treatment: Ara-C/Daunorubicin.

Complete remission: remission was obtained

Comments: marrow still recovering as of 02/09/07 w/ WBC 1900; ANC 670; Hg 10.0; plt 63,000; Abs Mono Count 228.

Relapse: +

Phenotype at relapse: normal karyotype 46,XY. The patient died shortly after the last cytogenetic analysis due to disease relapse.

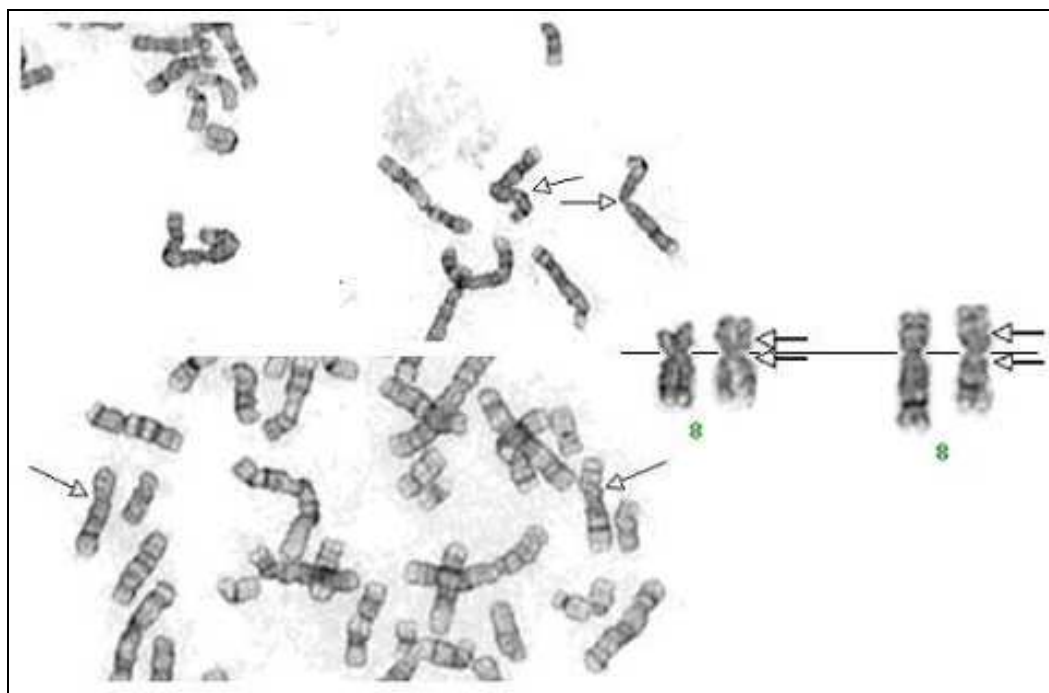
Status: Dead 02-2007

Survival: 2 months

### Karyotype

Sample: Bone Marrow; Culture time: Overnight and 24h; Banding: G-banding; 450 band level.

Results: 46,XY,inv(8)(p11.2q13)[6]/46,XY[14]



Partial metaphases and partial karyotypes showing the *inv(8)(p11q13)*

## Comments

A review of the literature revealed six reports of *inv(8)(p11q13)* associated with AML M4 or M5 all involving female patients ages 10 months, 15, 19, 21, 29, and 56 years old (References 1-6, 8-9). This is the first report of *inv(8)(p11.2q13)* associated with AML M4 in a male patient and older than 56 years. The inversion causes the fusion of the MOZ gene at 8p11.2 with TIF2 at 8q13. Deguchi et al described the requirement for C2HC nucleosome recognition of MOZ and the CBP recruitment activity of TIF2 for transformation leading to leukemogenesis. The propensity of affected females requires further investigation, but may just be a bias in the literature (Reference 7).

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