

## Case Report Section

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# Unbalanced rearrangement, der(9;18)(p10;q10) in a patient with myelodysplastic syndrome: Case 0002M

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### Clinics

#### Age and sex

85 years old male patient.

#### Previous history

Preleukemia. Previous malignancy Bladder cancer, status post removal and BCG treatment. 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 : 10.9g/dl

Platelets :  $57 \times 10^9/l$

### Cyto-Pathology Classification

#### Cytology

MDS (normocellular marrow with dysmegakaryopoiesis and dysgranulopoiesis; consistent with myelodysplastic syndrome)

**Immunophenotype:** NA

**Rearranged Ig Tcr:** NA

**Diagnosis:** MDS

### Survival

**Date of diagnosis:** 03-2005

**Treatment:** not on any treatment

**Complete remission :** None

**Treatment related death :** NA

**Relapse :** no

**Phenotype at relapse:** NA

**Status:** Alive. Last follow up: 12-2010

**Survival:** 66 months.

### Karyotype

**Sample:** 3/2005 BM, 6/2007 BM and 12/2010 BM

**Culture time :** 24 and 72 hours with overnight Colcemid

**Banding:** GTW at 400 bands

#### Results

3/2005 BM 45,X,-Y[5]/46,XY,+9,

der(9;18)(p10;q10)[11]/46,XY[4];

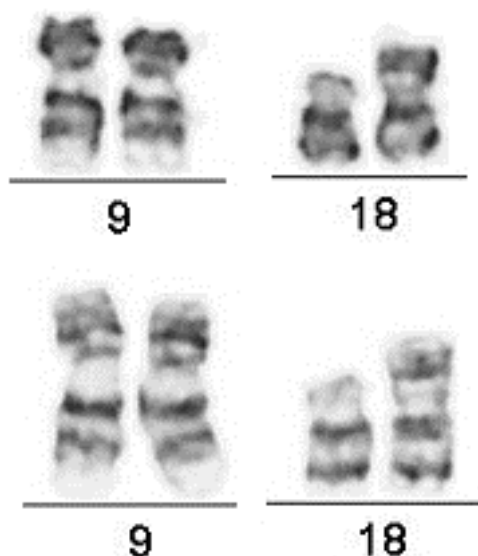
6/2007 BM 45,X,-Y[5][4]/46,XY[16];

12/2010 BM 46,XY,+9,

der(9;18)(p10;q10)[15]/46,XY[5]

**Karyotype at Relapse:** NA

**Other molecular cytogenetics technics:** None



Case 0002M : two partial karyotypes with a normal chromosome 9 pair, a *der(9;18)* and a normal chromosome 18 and arrow)

## Comments

Both the cases described in this study were followed for >5 years. Case 0001M, had thrombocytosis and could not tolerate Interferon or Hydrea treatment and hence was treated with Busulfan. The patient was positive for JAK2 mutation (on chromosome 9). A recent study was to rule out transformation of MPN as there was myelofibrosis, splenomegaly and apparent progression of the disease. The *der(9;18)* was first identified in the stem line and a sideline had partial deletion of chromosome 13q. Case 0002M was a MDS case with a *der(9;18)* detected in the initial study and again when the patient was suspected to be transforming >5 years later. This patient had very little symptoms and was not treated.

In this report for the first time a long standing MDS case was found to have the *der(9;18)* at initial diagnosis and after over 5 years. Others reported with *der(9;18)*(n 7) had PV (n 3) or post PV myelofibrosis (n

4) and one had sAML after ET. The JAK2V617F is a gain in function mutation on chromosome 9. Hence, the extra copy of 9p may exacerbate the MPN as observed in 0001M case. The patient had splenomegaly and also myelofibrosis when the patient was found with the *der(9;18)*. *Der(9;18)* is the sole abnormality in most reported cases, balanced translocations or complex aberrant karyotypes were reported as additional abnormalities. Our patient had *del(13)* in a sideline and this abnormality is observed in MPN. Among the 9 patients with *der(9;18)* two arose post treatment (present case 0001M and Andrieux et al 2003), and the other were at diagnosis.

The *der(9;18)* supports progression of the disease in case 0001M but in case 0002M with MDS it reappears when there is suspicion of transformation and its role is less uncertain.

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