Unbalanced rearrangement, der(9;18)(p10;q10) in a patient with myeloproliferative neoplasm: Case 0001M

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Clinics

Age and sex
71 years old male patient.

Previous history
Preleukemia. No previous malignancy. No inborn condition of note.

Organomegaly
No hepatomegaly, splenomegaly (Spleen appears enlarged measures 15.8 cm in length), no enlarged lymph nodes, no central nervous system involvement.

Blood

WBC: 112.7X 10^9/l
HB: 13.3g/dl
Platelets: 42X 10^9/l

Cyto-Pathology Classification

Cytology
MPN (near 100% cellular marrow with granulocytic and megakaryocytic hyperplasia consistent with chronic myeloproliferative neoplasm).

Immunophenotype: NA
Rearranged Ig Tcr: NA
Diagnosis: CMPN

Survival

Date of diagnosis: 10-2004
Treatment: Could tolerate Interferon or Hydrea and is on regulated dose of Busulfan.
Complete remission: None
Treatment related death: NA
Relapse: no
Phenotype at relapse: NA
Status: Alive. Last follow up: 12-2010.
Survival: 74 months

Karyotype

Culture time: 24 and 72 hours with overnight Colcemid
Banding: GTW at 400 bands

Other molecular cytogenetics technics: None

Other Molecular Studies

Technics: PCR
Results: JAK2V617F mutation
Unbalanced rearrangement, der(9;18)(p10;q10) in a patient with myeloproliferative neoplasm. Case 0001M.

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.

References


This article should be referenced as such: