Pentasomy 21 as a sole abnormality in an atypical CML patient in chronic phase.


Clinics
Age and sex: 65 yrs old female patient
Previous history: no preleukemia; no previous malignant disease; no inborn condition of note;
Organomegaly: no hepatomegaly; splenomegaly; no enlarged lymph nodes; no central nervous system involvement

Blood
WBC: 61.8 x 10^9/l; Hb: 11.5 g/dl; platelets: 348 x 10^9/l; blasts: 2%; (Myelocyte 13%, Meta Myelocyte 7%, Band cells 7%, P49/E4/B6/L12)%
Bone marrow: Increased cellularity/ M:E ratio, Megakaryocytes present, Erythropoiesis normoblastic. Blasts-8%, Promyelocytes-5%, Myelocytes-41%, Metamyelocytes-10%, Band cells-9%, Polymorphs-14%, Eosinophils-0%, Basophils-1%, Lymphocytes-05%, Monocytes-0%, Pronormoblasts-0%, Early normoblasts-0%, Internormoblasts-2%, Late normoblasts-5%.

Survival
Date of diagnosis: March 1999
Treatment: Hydrea
Complete remission: None
Treatment related death: -
Relapse: -
Status: Dead
Survival: 6 months

Karyotype
Sample: Bone marrow and Blood; culture time: Overnight; banding: G-banding
Results: 49XX,+21, +21, +21. (Pentasomy 21) in all 20 karyotypes (Fig 1).

Other molecular studies
Technics: Whole chromosome painting probe for chromosome 21, and BCR-abl gene
rearrangement (Vysis, USA).
results: Pentasomy confirmed (Fig 2), BCR-abl gene rearrangement was not present (Fig 3).

A G-banded Metaphase showing five copies of chromosome 21 (arrows) as a sole abnormality and the partial karyotype of the metaphase

A DAPI-counterstained metaphase after fluorescence in situ hybridization using FITC-labeled whole chromosome painting probe for chromosome 21 from Vysis, USA
A DAPI stained metaphase after fluorescence in situ hybridization using probe for detection of BCR-abl rearrangement from Vysis, USA

**Comments**

This is the first report of pentasomy 21 as a sole abnormality in a Philadelphia negative, bcr-abl negative i.e. atypical CML patient. Earlier this was reported in very young patients with; a congenital acute leukemia, a Diamond-Blackfan anemia, a neonatal AML, and acute leukemia patients with Down syndrome. One patient (72-year-old male) with AML without maturation has been reported recently. In majority of the cases pentasomy was due to isochromosome 21. To the best of our knowledge, this is the first case of atypical CML with pentasomy 21.

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