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


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## Clinics

<table>
<thead>
<tr>
<th>Age and sex</th>
<th>65 years old female patient.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Previous history</td>
<td>No preleukemia. No previous malignancy. No inborn condition of note.</td>
</tr>
<tr>
<td>Organomegaly</td>
<td>No hepatomegaly, splenomegaly, no enlarged lymph nodes, no central nervous system involvement.</td>
</tr>
</tbody>
</table>

## Blood

- **WBC**: 61.8X 10^9/l
- **HB**: 11.5g/dl
- **Platelets**: 348X 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%.

## Cyto-Pathology Classification

**Diagnosis**
Atypical CML chronic phase.

## 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).

## Survival

- **Date of diagnosis**: 03-1999
- **Treatment**: Hydrea
- **Complete remission**: no
- **Treatment related death**: no
- **Relapse**: no
- **Status**: Death. Last follow up: 07-1999 (expired in 08-1999).
- **Survival**: 6 months
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Roy SK et al.

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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.
References


This article should be referenced as such: