Isolated trisomy 2 is non-random and may be found in myelodysplastic syndrome and in acute myeloblastic leukaemia. Case 1

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Clinics
Age and sex: 58 years old male patient
Previous History: preleukemia; no previous malignant disease; no inborn condition of note.
Organomegaly: no hepatomegaly; no splenomegaly; no enlarged lymph nodes; no central nervous system involvement.

Blood
WBC: 1.9 x 10^9/l; Hb: 9.1 g/dl; platelets: 282x 10^9/l.
Bone marrow: 1.4% blasts

Cytopathology classification
Precise diagnosis: MDS: refractory cytopenia with multilineage dysplasia.

Survival
Date of diagnosis: 1979.
Treatment: Red cell transfusion monthly.
Complete remission was obtained.
Treatment related death: -
Relapse: -
Status: Alive 08-2005
Survival: 26 years +

Karyotype
Sample: Bone marrow; Culture time: 24/48h;
Bandaging: GTG.
Results: 46,XY, [6]/47, XY, +2 [14].

Other molecular cytogenetic technics: FISH using the BAC probe RP11-375H16 (2q23.1).
Other molecular cytogenetics results: 59% normal metaphases and 41% of metaphases with 3 chromosomes 2.

G-banding karyotype revealed isolated trisomy 2 of case 1.

Comments
Trisomy 2 as single chromosomal abnormality appears to be associated with MDS on the contrary to AML where it is frequently encountered in association to other unbalanced chromosomal abnormalities [ref.1]. This observation therefore suggests that trisomy 2 could be an early genetic abnormality in MDS. Indeed, from the 9 MDS/AML described cases with isolated trisomy 2 (including our 2 cases), 7 cases revealed isolated trisomy 2 at MDS presentation. MDS in transformation was diagnosed among the 4 oldest patients, though age does not carry prognostic
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significance according to the IPSS [ref.2]. 5 of the 9 published cases evolved to acute leukaemia.

References


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