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

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Clinics
Age and sex: 69 yrs old female patient
Previous history: no 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: 1.7 x 10^9/l; Hb: 7.4 g/dl; platelets: 45 x 10^9/l;
Bone marrow: 85% blasts%

Survival
Date of diagnosis: 12-2000
Treatment: Daunorobucin and cytosine-arabinoside
Complete remission was obtained
Treatment related death: Died after several septicemic episodes during bone marrow suppression treatment
Relapse: -
Status: Dead 05-2001
Survival: 12 months

Karyotype
Sample: Bone marrow; culture time: 24H and 48 H; banding: GTG
Results: 46,XX, [4]/ 47, XX,+2 [18]
Other molecular cytogenetics technics: FISH using the BAC probe RP11-375H16 (2q23.1)

G-banding karyotype revealed isolated trisomy 2 of case 2

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 significance according to the IPSS [ref.2]. 5 of the 9 published cases evolved to acute leukaemia.
International scoring system for evaluating prognosis in myelodysplastic syndromes.
Blood 1997; 89: 2079-2088.
Medline 9058730

Cross-validation of prognostic scores in myelodysplastic syndromes on 386 patients from a single institution confirms importance of cytogenetics.
Medline 10460606

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