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Case Report Section

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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.

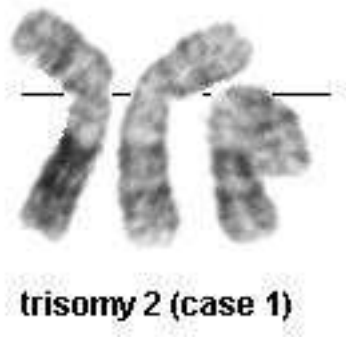
Survival

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;
Banding: 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

significance according to the IPSS [ref.2]. 5 of the 9 published cases evolved to acute leukaemia.

References

Greenberg P, Cox C, LeBeau MM, Fenaux P, Morel P, Sanz G, Sanz M, Vallespi T, Hamblin T, Oscier D, Ohyashiki K, Toyama K, Aul C, Mufti G, Bennett J. International scoring system for evaluating prognosis in myelodysplastic syndromes. Blood 1997;89:2079-2088.

Pfeilstöcker M, Reisner R, Nösslinger T, Grüner H, Nowotny H, Tüchler H, Schlögl E, Pittermann E, Heinz R. Cross-validation of prognostic scores in myelodysplastic syndromes on 386 patients from a single institution confirms importance of cytogenetics. Br J Haematol 1999;106:455-463.

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