t(1;21)(p32;q22) as a non-random abnormality in AML M4

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Clinics
Age and sex: 63 year(s) 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: 3.980 x 10^9/l; Hb: 7.9 g/dl; platelets: 64.000 x 10^9/l; blasts: 48,5%

Cytology classification
Cytology: (FAB) AML M4.
Immunophenotype: Hypercellular bone marrow showed a myelomonocytic blast population. 49.5% blasts were detected in total bone marrow. 30% of the cells were clearly EST positive. Furthermore POX was positive, no ringsideroblasts were found and erythropoiesis showed dysplasia. Myelomonocytic cells with MPO+ (48%), CD13+ (17%), CD33+ (63%), CD14 (19%) and CD64 (37%).
Precise diagnosis: AML M4

Survival
Date of diagnosis: 06-2008
Treatment: None
Complete remission: None
Treatment related death: -
Relapse: -
Status: Lost

Karyotype
Sample: bone marrow; culture time: 24 - 48 h; banding: GAG.
Results: 46,XX,t(1;21)(p32;q22)[15/15].
Other molecular cytogenetics technics: FISH with commercial AML1 probe (Abbott) and whole chromosome painting with WCP#1 and WCP#21 (MetaSystems).
Other molecular cytogenetics results: 40% of cells with AML1-split.

Other molecular studies
Technics: PCR
Results: Tandem duplication of MLL gene (MLL-PTD positive).

Partial GTG-banding karyotype showing t(1;21)(p32;q22).
FISH and whole chromosome painting of the same metaphase with t(1;21)(p32;q22): Left picture: AML1 probe on metaphase; Right picture: whole chromosome painting, WCP#1 green, WCP#21 red.

Comments
Only two cases with t(1;21)(p32;q22) were described so far in literature. The first reported case is a 25-year-old male with an acute myelomonoblastic leukemia (M4 by FAB subtype) (Cherry et al., 2001). The second patient, a 29-year-old Japanese male, showed a acute myelogenous leukemia M4 with NUP98-HOXA9 fusion detected by PCR at the initial diagnosis. In relapse he acquired additional to the NUP98-HOXA9 fusion a t(1;22)(p32;q22) (Aoki et al., 2008). The here reported case is a 63-year-old female with an acute myeloid leukemia (M4 by FAB subtype). So far the cases have the same morphology in common. Correlations to age or sex cannot be determined yet.

Call for collaboration
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