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Case Report Section

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t(3;4)(p21;q34) as a sole anomaly in acute myeloid leukemia patient

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Clinics

Age and sex: 32 years old male patient.
Previous History:
- no preleukemia;
- no previous malignant disease;
- no inborn condition of note.

Organomegaly:
- hepatomegaly;
- splenomegaly;
- enlarged lymph nodes;
- no central nervous system involvement.

Blood

WBC: 68.4 x 10^9/l; Hb: 11.7 g/dl; platelets: 120 x 10^9/l; blasts: 93%
Bone marrow: Markedly hypercellular, erythropoiesis depressed, leucopoiesis hyperplastic, 83.5% blasts (PM 1%, MY 2%, MM 3.5%, Po 0.5%, Ly 6.5%, Mono 2%, Eb 1%), near total replacement by blasts, megakaryocytes depressed, occasionally Auer rods seen.

Cytopathology classification

Cytology: Acute myeloid leukemia
Immunophenotype: Positive for CD 34, HLDR, CD33, CD34, CD68, myeloperoxidase.
Rearranged Ig or Tcr: -
Pathology: -
Electron microscopy: -
Precise diagnosis: Acute myeloid leukemia, M1.

Survival

Date of diagnosis: 05-2006.
Treatment: Allopurinol, Hydroxyurea, Tazocin, Amikacin (ADE 10, ADE 8).
Complete remission: None;
Treatment related death: -;
Relapse: -;
Phenotype at relapse: -;
Status: Alive (05-2007 traveled to receive BMT, allogenic BMT on 29-08-06);
Survival: 12 months.

Karyotype

Sample: BM; Culture time: 24h; Banding: G-band.
Results: 46,XY,t(3;4)(p21;q34)
Other molecular cytogenetic techniques: Fluorescence in situ hybridisation (FISH), with WCP 3 and 4 probes to confirm the t(3;4). To confirm the translocation of 3p and to exclude the translocation t(3;5)(q25;q34-35) FISH studies with LSI BCL6 and EGR1 SO/D5S23 probes were performed (Vysis, Downers Grove IL, USA).
Other molecular cytogenetics results: Using WCP 3 and 4 probes we confirmed the rearranged chromosomes 3 and 4. Analysis with LSI BCL6 probe revealed one red/green fusion signal on the 3q27 allele in the normal chromosome 3, and a fusion signal on the long arm of the der(3). Hybridization with LSI 5q SpectrumOrange/5p SpectrumGreen probe revealed 2 normal chromosomes 5, excluding the rearrangement of chromosome 5.

Other findings

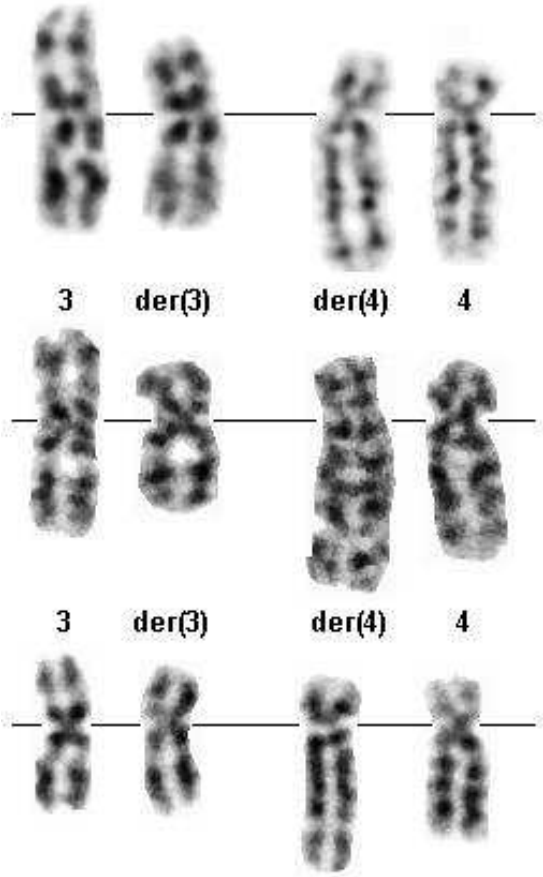
Results: LDH almost 3 folds upper normal limit.

Comments

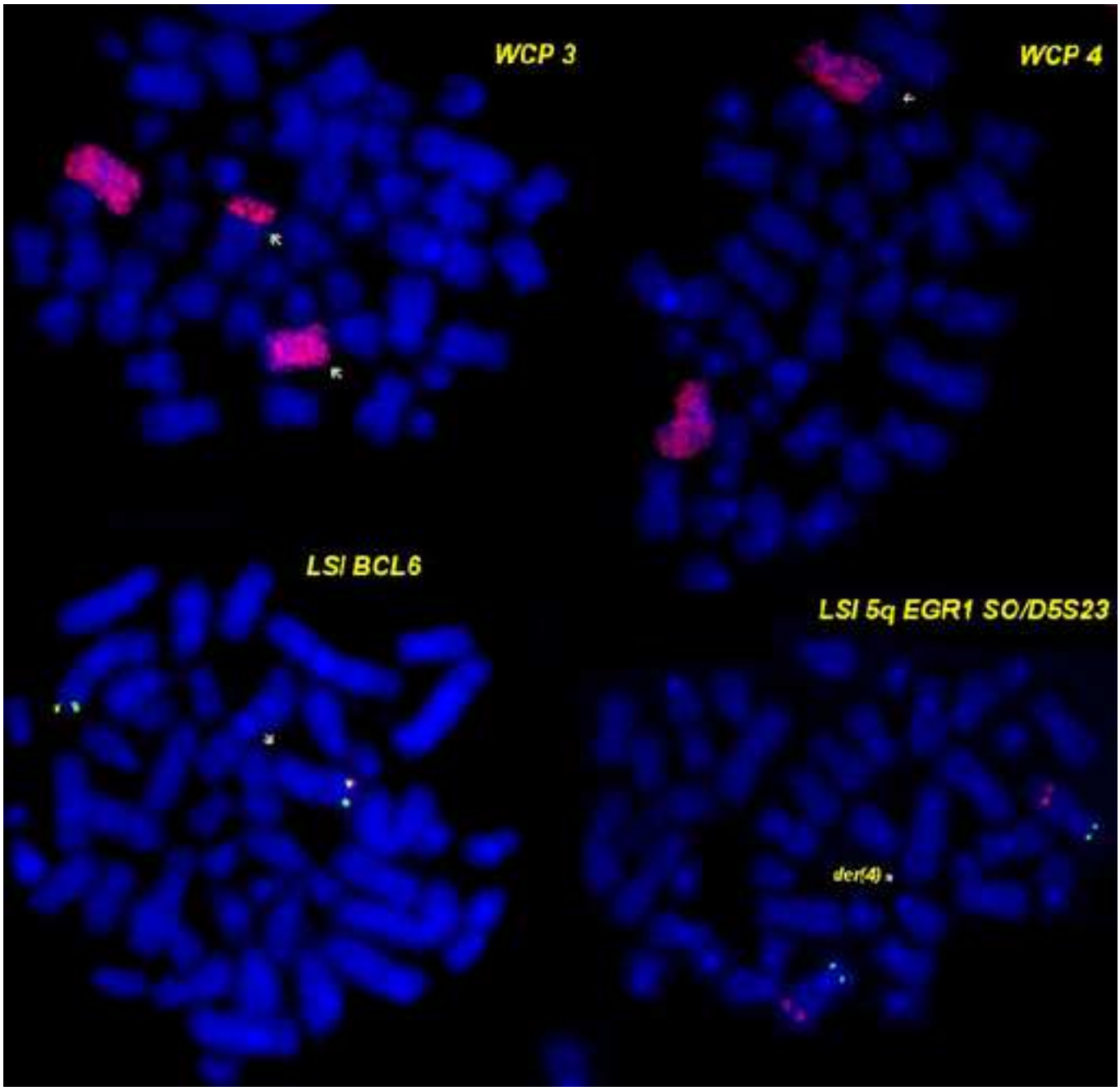
A 31-year-old Kuwaiti male presented with 3-months history of fatigue, recurrent sore throat attacks and a 2-day history of recurrent vomiting and loose motions. Physical examinations revealed left cervical lymphadenopaty and hepatosplenomegaly. Laboratory investigations showed Hb 11.7g/dl, platelets $120x10^9$l and white blood cells $68.4x10^9$l. Bone marrow smears were markedly hypercellular with 93% large blast cells. Cytochemical studies showed myeloperoxydase positive (60%), Sudan Black B positive (74.6%), PAS and non-specific esterase negative blast cells.

On the basis of these morphological findings, a diagnosis of acute myeloid leukemia (FAB-M1 type) was made.

3p21 is a recurrent breakpoint in MDS/AML and t-MDS/t-AML suggesting, 3p21 site is likely to contain a gene (genes) involved in the pathogenesis of t(3;4)(p21;q34). One previous case of t(3;4)(p21;q34) was found in a refractory anemia, making this anomaly recurrent. The similar cytogenetic appearance of a rare t(3;4)(p21;q34) and the more frequent t(3;5)(q25;q34) in suboptimal preparations reinforces the utility of FISH technique for assessing chromosomal abnormalities in AML.



Partial karyotypes (G-banding) demonstrating rearrangened chromosomes 3 and 4.



Whole chromosome paintings with rearrangened chromosomes 3 and 4 and hybridization with LSI BCL6 and LSI 5qSO/5pSG probes showing the fusion signal on normal chromosome 3 and on der(3) chromosome and two normal chromosomes 5.

References

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