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Case Report Section

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t(1;16)(q11-12;q11) presented as a der(16)t(1;16) in a patient with acute lymphoblastic leukemia

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Clinics

Age and sex: 56 years old male patient.
Previous history:
- no preleukemia;
- no previous malignant disease;
- no inborn condition of note.

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

Blood

WBC: 213 x 10^9/l; Hb: 9.6 g/dl; platelets: 23 x 10^9/l; blasts: 93%
Bone marrow: Markedly hypercellular, normal granulopoiesis depressed, near total replacement by blasts with high N/C ratio, agranular lightly basophilic cytoplasm, Poly 2, Lymp 4, Eos 1. PAS positive, SBB negative.

Cytopathology classification

Cytology: Acute lymphoblastic leukemia
Immunophenotype: Positive for CD45, CD10, CD19, CD34, HLADR, TdT.
Rearranged Ig or Tcr: -
Pathology: -
Electron microscopy: -
Precise diagnosis: Acute lymphoblastic leukemia, L1 (pre-B).

Survival

Date of diagnosis: 08-2006.
Treatment: Methotrexate, Ara-C, Hyper-CVAD protocol.
Complete remission was obtained.
Treatment related death: -;
Relapse: -;
Phenotype at relapse: -;
Status: Alive (04-2007);
Survival: 9 months.

Karyotype

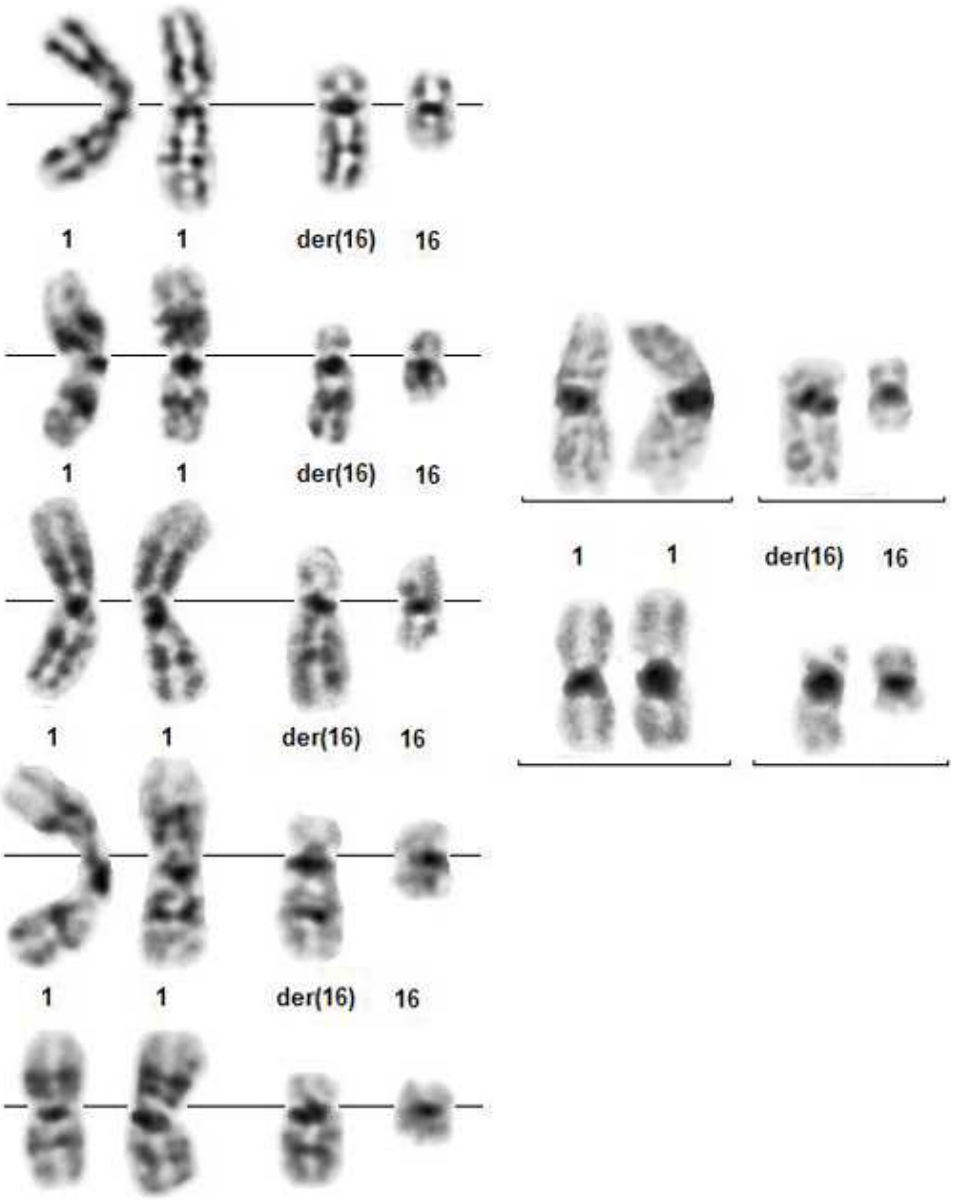
Sample: BM; Culture time: 24h; Banding: G-band.
Results: 46,XY,der(16)t(1;16)(q11-12;q11) [20]
Other molecular cytogenetic techniques: Fluorescence in situ hybridisation (FISH), with LSI CBFB DC and WCP probes for chromosome 1 and 16 (WPC DNA Probe 1, SpectrumOrange; WPC DNA Probe 16, SpectrumGreen) obtained from Vysis (Downers Grove IL, USA).
Other molecular cytogenetics results : The analysis with LSI CBFB DC probe revealed one normal signal on the CBFB allele in the normal chromosome 16, while on the der(16) no red/green signal was detected, confirming the rearrangement of 16q. Hybridization with WCP 1 SpectrumOrange and WCP 16 SpectrumGreen probes revealed 2 normal chromosomes 1, one normal chromosome 16 and confirmed the der(16)t(1;16).

Other molecular studies

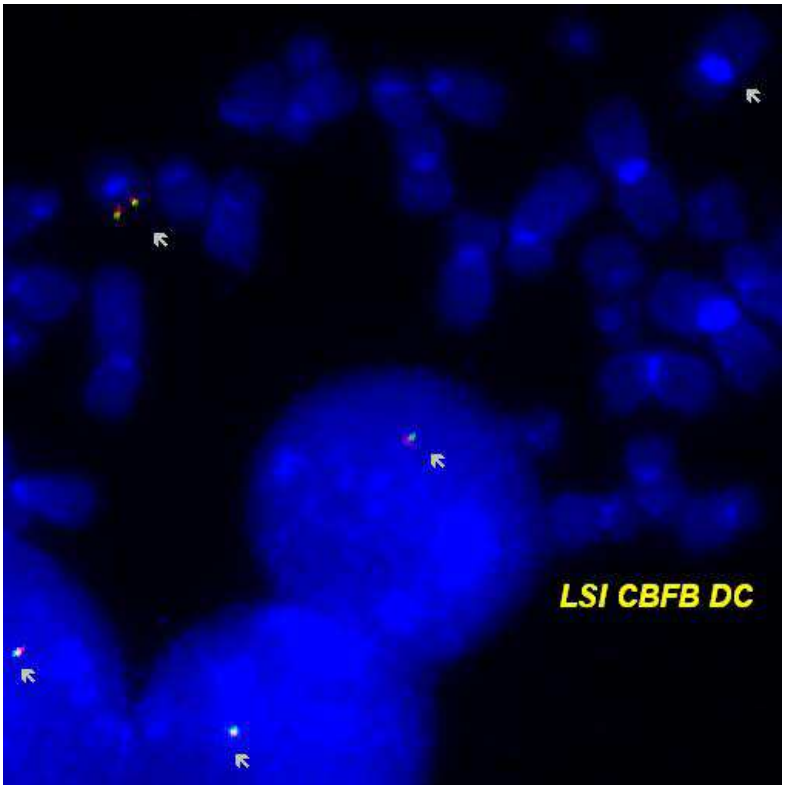
Technics: RT-PCR for BCR-ABL
Results: The BCR-ABL transcript was negative by the conventional method of molecular analysis.

Comments

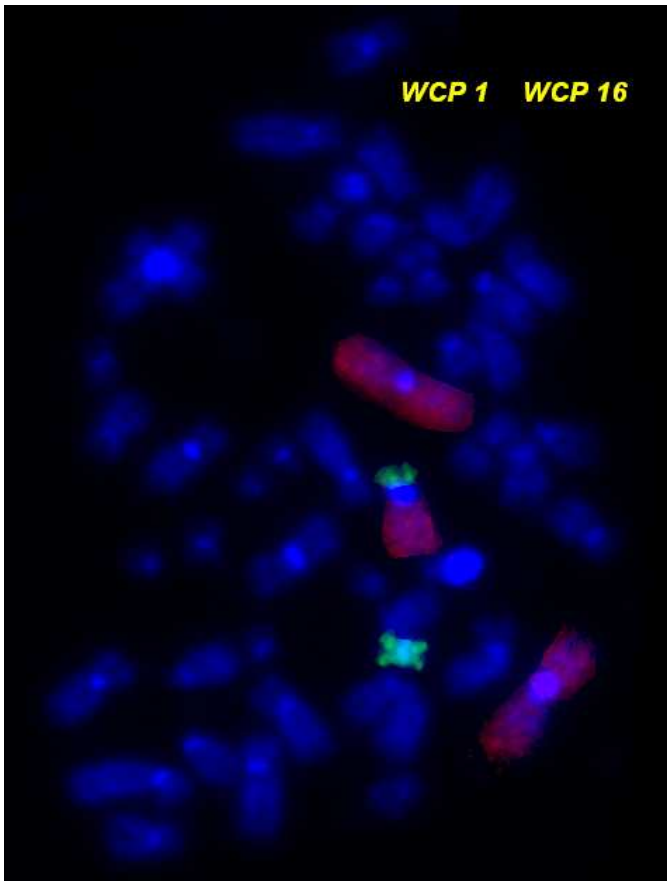
A 47-years old Filipino male was diagnosed with ALL in August 2006. Cytogenetic analysis of the bone marrow sample revealed a clearly abnormal chromosome 16 and the karyotype 46,XY,-16,+der(16)t(1;16)(q11-12;q11) was identified in all the 30 examined metaphases. Recurrent whole-arm translocation of 1q to the centromeric region of chromosome 16 has been detected in a number of malignancies, but only occasionally described in hematological malignancies. The previously described 3 MDS, 4 AML and 3 ALL cases with t(1;16)(q11-q12;q11-12) were always unbalanced, suggesting either trisomy of 1q or monosomy of 16q may potentially contribute to leukemogenesis.



Partial karyotypes demonstrating 2 normal chromosomes 1, one normal chromosome 16 and the der(16)t(1;16). C-banded chromosomes on the right side.



LSI CBFB DC, Break Apart Rearrangement Probe exhibiting one normal signal on the CBFB allele on normal chromosome 16.



Whole chromosome painting showing 2 normal chromosomes 1 and the rearrangened chromosomes 1 and 16.

References

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Mugneret F, Dastugue N, Favre B, Sidaner I, Salles B, Huguet-Rigal F, Solary E. Der(16)t(1;16)(q11;q11) in myelodysplastic syndromes: a new non-random abnormality characterized by cytogenetic and fluorescence in situ hybridization studies. Br J Haematol 1995;90:119-124.

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