

Case Report Section

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Translocation t(7;9)(q34;q32) found in pediatric T-cell acute lymphoblastic leukemia

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Clinics

Age and sex: 4 years old female 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: 148 x 10⁹/l; Hb: 9.8 g/dl; platelets: 13x 10⁹/l; blasts: peripheral 60%. Bone marrow: 100%

Cytopathology classification

Cytology: L2.
Immunophenotype: NK+, CD2, CD3, CD4, CD5, CD7, CD8, and TdT+.
Precise diagnosis: acute lymphoblastic leukemia, L2.

Survival

Date of diagnosis: 02-2006.
Treatment: chemotherapy (intrathecal and systemic), cranial X-ray therapy, and allogenic stem cell transplant.
Relapse: + central nervous system (8/06 with cytogenetics) and bone marrow relapse.
Status: Alive 08-2007.
Survival: 31 months to date.

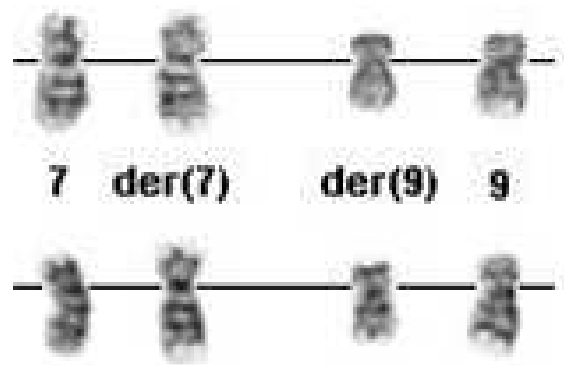
Karyotype

Sample: diagnostic sample was of bone marrow.
Culture time: analysis of diagnostic sample was performed on overnight colcemid, 18-hour and 72-hour cultures.

Banding: 350 band level.

Results: 46,XX,t(7;9)(q34;q31~q32)[16]/46,XX[4]

Karyotype at relapse: Date: 08-2006; Result: 46,XX,del(6)(q21), t(7;9)(q34;q31~q32)[cp2]/46,XX[19]



Partial karyotypes of metaphases from diagnostic sample overnight colcemid culture.

Comments

The patient presented in this case report has features consistent with other reported cases of T cell acute lymphoblastic leukemia (T-ALL) with t(7;9)(q34;q32) involving the TCRbeta locus on 7q34 and the TAL2 gene on 9q34. In particular, review of the literature revealed a case in a 3 year old male with similar clinical and hematological findings and the same additional cytogenetic anomaly, del(6)(q21). Approximately 60% of clinically normal individuals have t(7;9)(q34;q32) created by V(D)J recombination which uses a fortuitous recombination site (RSS) located 3' of the TAL2 oncogene. In T-ALL affected individuals a rare second rearrangement occurs between the junction point of the original t(7;9) and Jbeta2. This brings the TAL2 gene under the control of TCRbeta enhancer, and leads to its' inappropriate expression.

References

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