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Leukaemia Section
Short Communication

t(2;21)(q11;q22)

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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Clinics and pathology

Disease

T-cell acute lymphoblastic leukemia.

Phenotype/cell stem origin

One case to date, a 6-year-old boy (Chinen et al., 2008).

Evolution

Complete remission was obtained. An allogenic bone marrow transplantation was performed, and the patient had remained in complete remission for 17 months at the time of the report.

Cytogenetics

Additional anomalies

A complex karyotype was found.

Genes involved and proteins

AFF3

Location
2q11.2
Protein
AFF3 belongs to a family of putative transcription factors also comprising AFF1 (AF4, FEL, MLLT2) in 4q21, AFF2 (FMR2, FRAXE) in Xq28 and AFF4 (AF5Q31) in 5q31. AFF3 has been found a susceptibility gene in autoimmune diseases, namely rheumatoid arthritis, psoriatic arthritis, and juvenile idiopathic arthritis (Barton et al., 2009; Castelino and Barton, 2010; Hinks et al., 2010). AFF3 is deleted in Nievergelt syndrome, an autosomal dominant mesomelic dysplasia (Steichen-Gersdorf et al., 2008). AFF3 was also found expressed in 20% of mammary tumor cells but not in normal acini in a study (To et al., 2005).

RUNX1

Location
21q22.3
Protein
Transcription factor (activator) for various hematopoietic-specific genes.

Result of the chromosomal anomaly

Hybrid gene

Description
Fusion of RUNX1 exon 7 to AFF3 exon 8.

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