Leukaemia Section
Short Communication

t(1;11)(q21;q23)
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Published in Atlas Database: April 1998
Online updated version: http://Atlas GeneticsOncology.org/Anomalies/t111ID1004.html
DOI: 10.4267/2042/37442

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Clinics and pathology

Disease
ANLL.

Phenotype / cell stem origin
Mostly M4; M5, biphenotypic leukaemia.

Epidemiology
6 known cases, some with few or no data; infants and children; 1 young adult.

Prognosis
A poor response to chemotherapy has been reported in 2 cases; only one case with survival data: the patient died 70 mths after diagnosis.

Cytogenetics

Additional anomalies
Sole anomaly, or with +19 (recurrent) or other numerical anomalies; duplicated der(11).

Genes involved and Proteins

AF1q
Location: 1q21
DNA / RNA
1.8 kb mRNA.
Protein
9 kDa.

MLL
Location: 11q23
DNA / RNA
21 exons, spanning over 100 kb; 13-15 kb mRNA.

Protein
431 kDa; contains two DNA binding motifs (a AT hook, and Zinc fingers), a DNA methyl transferase motif, a bromodomain; transcriptional regulatory factor; nuclear localisation.

Results of the chromosomal anomaly

Hybrid gene
Description
5' MLL - 3' AF1q; breakpoints: between exons 6 and 7 in MLL and within the 5' untranslated region in AF1q.

Fusion protein
Description
N-term -- AT hook (DNA binding) and DNA methyltransferase motif from MLL fused to the entire AF1q on the der(11); the reciprocal on der(1) is out of frame.

References


Tse W, Zhu W, Chen HS, Cohen A. A novel gene, AF1q, fused to MLL in t(1;11)(q21;q23), is specifically expressed in leukemic and immature hematopoietic cells. Blood 1995 Feb 1;85(3):650-6.

This article should be referenced as such:
Huret JL. t(1;11)(q21;q23). Atlas Genet Cytogenet Oncol Haematol.1998;2(3):100