Leukaemia Section
Short Communication

ins(5;11)(q31;q13q23)

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Clinics and pathology

Disease
Acute lymphoblastic leukemia (ALL) and ALL evolving towards a M4- acute non lymphocytic leukemia (ANLL).

Phenotype/cell stem origin
CD19+

Epidemiology
Poorly defined: only 2 cases to date.

Clinics
A girl aged 4 mths, who entered complete remission, relapsed and died 20 mths after diagnosis, and a 3 mth old boy who had a 18 mths remission, developed relapses and died.

Prognosis
Yet unknown, likely to be poor.

Cytogenetics

Cytogenetics morphological
The chromosomal rearrangement may be hidden if only a small portion of chromosome 11 (with MLL) is transferred into chromosome 5 (in the vicinity of AF5Q31).

Cytogenetics molecular
The use of MLL probes can uncover the anomaly.

Additional anomalies
i(17q) in one case, and a complex karyotype in the other case.

Variants
A few cases of t(5;11)(q31;q23) have been described, but it is unknown if they involve the same genes.

Genes involved and proteins

AF5q31
Location
5q31.1
Protein
Present homologies with AF4.

MLL
Location
in 11q23
DNA/RNA
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.

Result of the chromosomal anomaly

Hybrid gene
Description
5’ MLL- 3’ AF5q31

References
Taki T, Kano H, Taniwaki M, Sako M, Yanagisawa M, Hayashi Y. AF5q31, a newly identified AF4-related gene, is fused to
Deveney R, Chervinsky DS, Jani-Sait SN, Grossi M, Aplan PD. Insertion of MLL sequences into chromosome band 5q31 results in an MLL-AF5Q31 fusion and is a rare but recurrent abnormality associated with infant leukemia. Genes Chromosomes Cancer. 2003 Jul;37(3):326-31

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