

# Leukaemia Section

## Short Communication

### ins(5;11)(q31;q13q23)

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Published in Atlas Database: April 2000

Online updated version : <http://AtlasGeneticsOncology.org/Anomalies/ins511ID1167.html>

DOI: 10.4267/2042/37619

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## Clinics and pathology

### Disease

Acute lymphoblastic leukemia (ALL).

### Phenotype/cell stem origin

CD19+

### Epidemiology

Poorly defined: only 1 case to date.

### Clinics

A girl aged 4 months, who entered complete remission, relapsed and died 20 months after diagnosis.

### Prognosis

Yet unknown, likely to be poor.

## Cytogenetics

### Additional anomalies

i(17q)

### 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

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; fusion at MLL exon 10.

## References

Taki T, Kano H, Taniwaki M, Sako M, Yanagisawa M, Hayashi Y. AF5q31, a newly identified AF4-related gene, is fused to MLL in infant acute lymphoblastic leukemia with ins(5;11)(q31;q13q23). Proc Natl Acad Sci U S A. 1999 Dec 7;96(25):14535-40

*This article should be referenced as such:*

Huret JL. ins(5;11)(q31;q13q23). Atlas Genet Cytogenet Oncol Haematol. 2000; 4(2):74.