

# Leukaemia Section

## Short Communication

### t(1;12)(q25;p13)

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## Clinics and pathology

### Disease

Acute non lymphocytic leukemia (ANLL).

### Phenotype/cell stem origin

M4Eo ANLL.

### Epidemiology

Only one case available.

## Cytogenetics

### Cytogenetics morphological

A cryptic inv(16) was present, ascertained by a CBFb/MYH11 rearrangement; there fore, the t(1;12) may be a secondary anomaly.

## Genes involved and proteins

### ABL2

#### Location

1q25

#### Protein

Tyrosine kinase; closely related to ABL1.

### ETV6

#### Location

12p13

#### DNA/RNA

Alternative transcripts.

### Protein

Belong to the ETS transcription factors family characterized by the ETS domain, domain which is responsible for the sequence specific DNA-binding activity.

## Result of the chromosomal anomaly

### Hybrid gene

#### Transcript

Both reciprocal transcripts are detected.

### Fusion protein

#### Description

The fusion protein is composed of the HLH oligomerization domain of ETV6 and the SH2, SH3, and protein tyrosine kinase domains of ABL2.

## References

Cazzaniga G, Tosi S, Aloisi A, Giudici G, Daniotti M, Pioltelli P, Kearney L, Biondi A. The tyrosine kinase abl-related gene ARG is fused to ETV6 in an AML-M4Eo patient with a t(1;12)(q25;p13): molecular cloning of both reciprocal transcripts. *Blood*. 1999 Dec 15;94(12):4370-3

Cazzaniga G, Tosi S, Aloisi A, Giudici G, Pioltelli P, Kearney L, Biondi A. The tyrosine kinase ABL-related gene 'ARG' is fused to ETV6 in an AML-M4Eo patient with a t(1;12)(q25;p13): molecular cloning of both reciprocal transcripts. *Blood* 1999;94 Suppl 1:Abst 233

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