

# Leukaemia Section

## Short Communication

### t(12;17)(p13;p13)

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## Clinics and pathology

### Disease

Chronic myelomonocytic leukaemia (CMML) in transformation into an acute myeloid leukaemia (AML) (Penas et al., 2003).

### Epidemiology

Only 1 case to date, a 74-year-old male patient.

### Prognosis

No data.

## Cytogenetics

### Cytogenetics morphological

Cryptic translocation: the karyotype suggested a del(12p), and FISH analyses uncovered the translocation.

### Additional anomalies

The translocation was the sole anomaly.

## Genes involved and proteins

### ETV6

#### Location

12p13.2

#### Protein

ETV6 is composed of a HLH domain (pointed or sterile alpha motif (SAM) domain), responsible for dimerization and an ETS domain, responsible for

sequence specific DNA-binding. Transcriptional regulator.

### PER1

#### Location

17p13.1

#### Protein

PER1 contains a bHLH and PAS region involved in dimerization. Transcriptional regulator.

## Result of the chromosomal anomaly

### Hybrid gene

#### Description

An ETV6-PER1 transcript was detected, joining exon 1 of ETV6 to Exon 22 of PER1; however, PER1 has an antisense orientation, and the sequence was ETV6 exon 1, PER1 exon 22, and part of PER1 exon 21. No reciprocal transcript.

## References

Murga Penas EM, Cools J, Algenstaedt P, Hinz K, Seeger D, Schafhausen P, Schilling G, Marynen P, Hossfeld DK, Dierlamm J. A novel cryptic translocation t(12;17)(p13;p12-p13) in a secondary acute myeloid leukemia results in a fusion of the ETV6 gene and the antisense strand of the PER1 gene. *Genes Chromosomes Cancer*. 2003 May;37(1):79-83

*This article should be referenced as such:*

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