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

This article should be referenced as such: