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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

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