Leukaemia Section
Short Communication

t(12;21)(q12;q22)
Jean-Loup Huret
Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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Clinics and pathology

Disease
Acute myeloid leukaemia (AML), M2 subtype.

Epidemiology
Only one case to date, a 76 year old male patient.

Prognosis
No data: the patient died, but no survival data was noted.

Cytogenetics

Additional anomalies
The t(12;21)(q12;q22) was the sole anomaly.

Genes involved and proteins

CPNE8
Location
12q12
Protein
CPNE8 is a member of the copines. Copines are highly conserved, widely expressed, calcium-dependent membrane binding proteins. They may have a role in membrane trafficking and mediate cellular processes by conferring calcium regulation to various signaling pathways. Copine 8 is strongly expressed in brain, heart, and prostate, and expressed at low level in most other tissues.

RUNX1
Location
21q22
Protein
Contains a RUNT binding domain and a trans-activation domain; forms heterodimers; nuclear localization; transcription factor; critical regulator of hematopoietic-cell development.

Result of the chromosomal anomaly

Hybrid gene
Description
5' RUNX1 (including the DNA binding domain) broken after exon 6 and fused to intron 2 of CPNE8. However, fusion is out of frame, and termination occurs after 2 amino acids from CPNE8, producing a truncated RUNX1.

Fusion protein
Description
The truncated RUNX1 includes the binding domain, but not the transactivation domain.

References


This article should be referenced as such: