t(12;22)(p13;q11-12)
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Identity

G- banding - Courtesy Melanie Zenger and Claudia Haferlach.

Clinics and pathology

Disease
Myeloid lineage (ANLL, MDS)

Phenotype / cell stem origin
M4 ANLL often (6/11); M7 ANLL, other ANLL; RAEB evolving towards ANLL; ‘atypical CML’; may occur secondary to genotoxic exposure.

Epidemiology
Yet poorly known; median age: 22 yrs (range 8-60; n=11 cases herein reviewed); male predominance so far (8/3).

Prognosis
Yet uncertain: survival range 0 mths - 6 yrs, median 2 yrs, n=9.

Cytogenetics

GENES INVOLVED

ETV6
Location: 12p13
DNA / RNA
9 exons; alternate splicing.

Protein
Contains a Helix-Loop-Helix and ETS DNA binding domains; wide expression; nuclear localisation; ETS-related transcription factor.

MN1
Location: 22q11
DNA / RNA
Alternate splicing.

Protein
Glutamine and prolin rich protein; widely expressed; transcriptional regulator.

Results of the chromosomal anomaly

Hybrid gene
Description
Variable breakpoints in ETV6.
Fusion protein

Description
N term- MN1-ETV6-C term, with most of MN1, including the glutamine/proline rich domain, fused to the DNA binding of ETV6; the reciprocal ETV6/MN1 may or may not be expressed.

Expression localisation
Nuclear protein.

Oncogenesis
May act as an altered transcription factor.

References


This article should be referenced as such: