t(16;21)(p11;q22)

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Identity

t(16;21)(p11;q22) G- banding - Courtesy Melanie Zenger and Claudia Haferlach.

Clinics and pathology

Disease
de novo acute non lymphocytic leukemia (ANLL); to be noted is one case of chronic myelogenous leukemia (CML) -blast crisis.

Phenotype/cell stem origin
ANLL cases: mainly M1, M2, M4, M5a, M5b, or M7 ANLL; may be preceded by a myelodysplastic syndrome (MDS).

Epidemiology
About 40 reported cases, mainly found in young adults; children cases are described; median age is about 30 yrs; balanced sex ratio.

Clinics
Blood data: anemia, thrombocytopenia, mild hyperleucocytosis; with high monocytc cell count at times.

Cytology
Myelocytic and monocytoid features are often present; eosinphils in the bone marrow are sometimes abnormal and/or elevated; erythrophagocytosis may be found.

Prognosis
Seems poor: complete remission may not be achieved; there is high incidence of relapse within a year and a median of survival is about 22 months (cases herein reviewed).

Ewing tumours

Note

Ewing tumours, a paediatric neoplasm with small round-cells derived from neural crests cells usually associated with translocations involving EWSR1.
Cytogenetics

Ewing tumours are usually associated with a t(11;22)(q24;q12) with 5' EWSR1 - 3' FLI1 involvement, less often associated with t(21;22)(q22;q12) with 5' EWSR1 - 3' ERG involvement, rarely associated with t(2;22)(q36; q12) (5' EWSR1-3' FEV) or with t(17;22)(q21;q12) (5' EWSR1-3' ETV4).

Prognosis

Recent treatments have improved the prognosis of Ewing's tumours. The prognosis is mainly determined by the presence of metastases at the time of diagnosis.

Cytogenetics

Additional anomalies

ANLL cases: found solely in about 60% of cases in at least a subclone; associated with +10, +8, or de(9q)/-9 in about 10% of cases each.

Genes involved and proteins

FUS

Location 16p11
Protein RNA binding protein; member of the TET family, like EWSR1.

ERG

Location 21q22
Protein ETS transcription factor.

Result of the chromosomal anomaly

Hybrid gene

Description 5' FUS including exons 1 to 6, 7 or 8 - 3' ERG from exon 7, 8 or 9 to C-term.

Fusion protein

Description N-term FUS transactivation domain fused to the C-term DNA binding ETS domain of ERG.

Oncogenesis

Seems to act as a transcriptional activator.

References


Harigae H, Kobayashi M, Mihara A, Watanabe N. Detection of minimal residual disease in cerebro-spinal fluid of a patient...


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