

Leukaemia Section

Short Communication

t(16;21)(p11;q22)

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Clinics and pathology

Disease

De novo ANLL; (one case of blast crisis CML).

Phenotype / cell stem origin

Mainly M2 or M4, but also M1, M5a, M5b, or M7 ANLL; may be preceded by MDS.

Epidemiology

About 30 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 monocytic cell count at times.

Cytology

Myelocytic and monocytoid features are often present; eosinophils 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).

Cytogenetics

Additional anomalies

Rarely found solely; most often associated with various numerical or structural abnormalities; trisomy 10 was found in 4 of 17 cases.

Genes involved and Proteins

FUS

Location: 16p11

ERG

Location: 21q22

Results of the chromosomal anomaly

Hybrid gene

Description

5' FUS - 3' ERG

Fusion protein

Description

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

Oncogenesis

Seems to act as a transcriptional activator.

References

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