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Leukaemia Section

Short Communication

t(8;16)(p11;p13)

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Clinics and pathology

Disease

ANLL; t-ANLL

Phenotype / cell stem origin

M4, M5a, M5b; possible involvement of a granulo-monocytic precursor; no preceeding MDS.

Epidemiology

Rare disease (<1% of ANLL); found in children (including infants) and young adults of both sexes.

Clinics

Disseminated intra vascular coagulation may be present; extramedullary infiltration; 20% of the cases could be therapy-related.

Cytology

Erythrophagocytosis, strong peroxidase and esterase activities.

Prognosis

Poor: remission may be obtained in half cases; infections, bleeding; survival is often less than 1 year.

Cytogenetics

Additional anomalies

In half cases; +8, various; complex karyotype may be found.

Variants

Complex t(8;16;Var) involving a (variable) third chromosome have been described; 8p11 breakpoint with another partner as well, of which is the recurrent t(8;22)(p11;q13), which may involve P300 on 22q13 in the place of CBP: this translocation would therefore be an equivalent (not identical), and not a simple variant with hidden 16p13 involvement.

Genes involved and Proteins

MOZ

Location: 8p11

CBP

Location: 16p13

Results of the chromosomal anomaly

Hybrid gene

Description

5' MOZ - 3' CBP

Fusion protein

Description

N-term MOZ fused to most of CBP; 3722 amino acids; 415 kDa; combines the MOZ finger motifs (DNA binding) and acetyl transferase with the transcriptional coactivator from CBP; the reciprocal CBP-MOZ has no role (as it is out of frame).

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