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 granulomonocytic 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 \( t(8;16;\text{Var}) \) 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).

References


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