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Leukaemia Section
Short Communication

t(8;16)(p11;p13)

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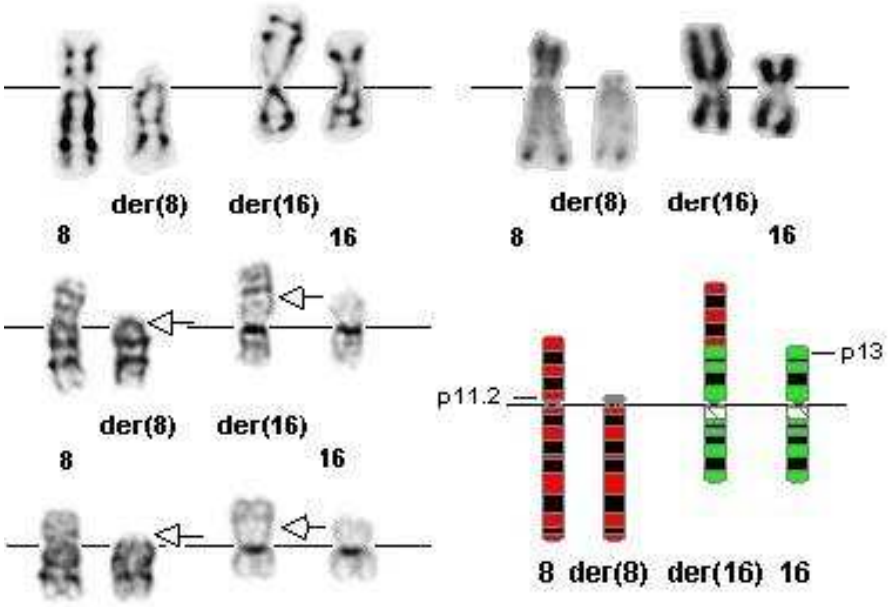
Identity



t(8;16)(p11;p13) G- banding (left) - Courtesy Jean-Luc Lai (top left) and Charles D. Bangs (middle and bottom left), R- banding (top right) - Courtesy Jean-Luc Lai, and ideogram (bottom right) - Courtesy Charles D. Bangs.

Clinics and pathology

Disease

ANLL; t-ANLL

Phenotype/cell stem origin

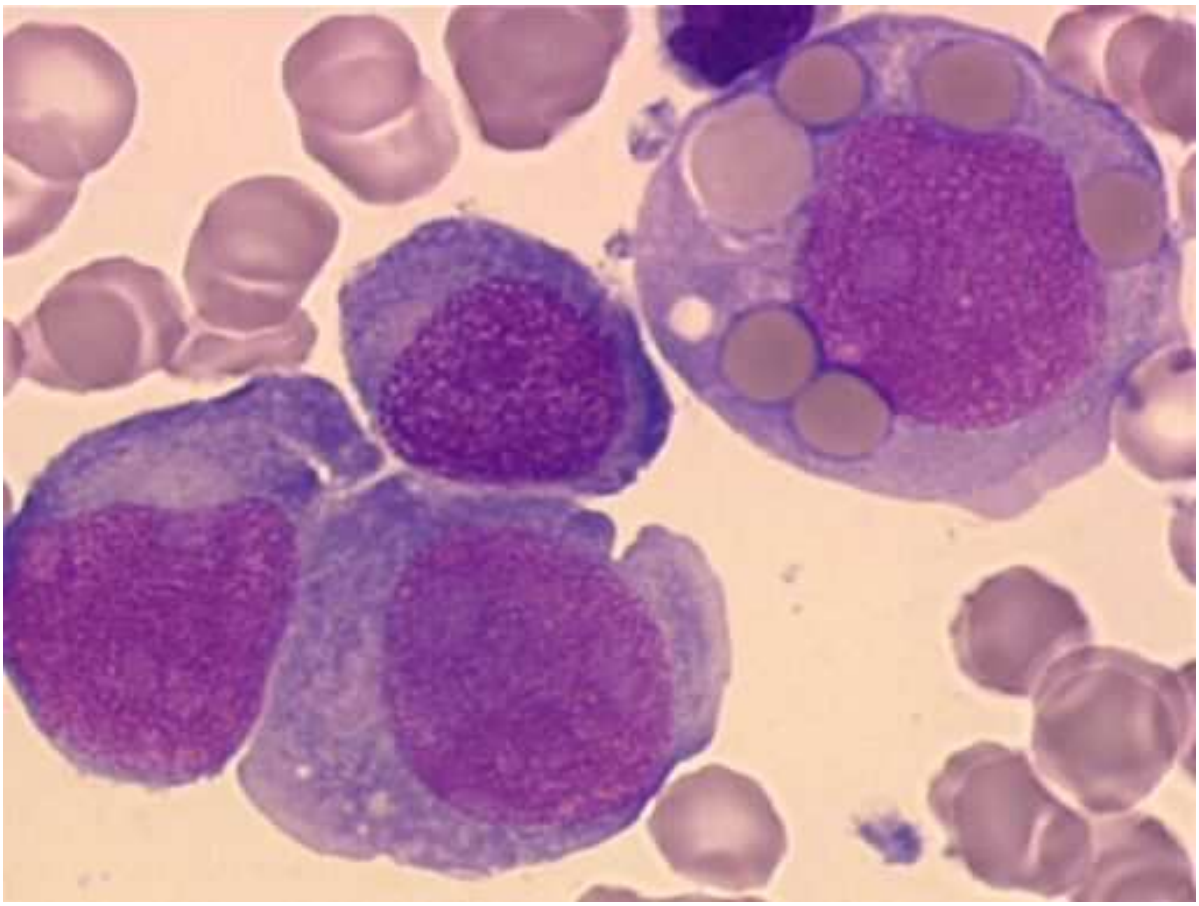
M4, M5a, M5b; possible involvement of a granulo-monocytic precursor; no preceeding MDS.

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.



The t(8;16) has been cloned and shown to fuse the MOZ (monocytic leukemia zinc finger) gene at 8p11.2 to the CBP (CREB binding protein) gene at 16p13.3. The MOZ gene has also been found to be involved in variant translocations t(8;19)(p11;q13) and t(8;22)(p11;q13) and inv(8)(p11q13) translocations associated with M5/M4 AML.This translocation is associated with AML M5/M4. In the majority of cases it is associated with features of hemophagocytosis by leukemic cells, particularly erythrophagocytosis - Courtesy Georges Flandrin, CD-ROM AML/MDS G.Flandrin/ICG. TRIBVN.

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 but spontaneous remission has occurred (at least) once.

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

Result 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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