Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Leukaemia Section

Mini Review

t(16;21)(q24;q22)

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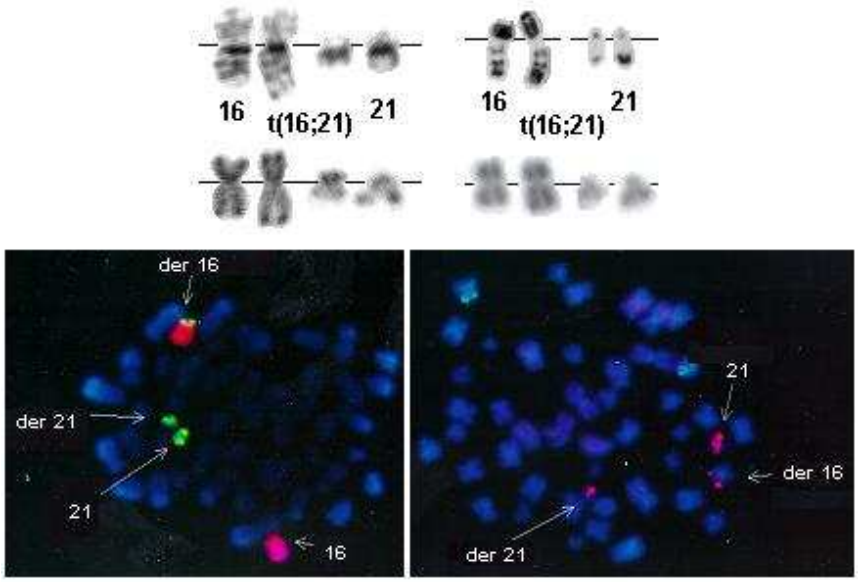
Identity



t(16;21)(q24;q22) G-banding (top left) - Courtesy Jean Luc LaÃ¯; R-banding (middle left - Courtesy Jean Luc LaÃ¯ and top and middle right - Courtesy Pascale Cornillet-Lefebvre and StÃ©phanie Struski). Fish studies: chromosome 16 and 21 paints; AML1 probe: the 2 chromosomes 21 and the der(16) are labelled (below) - Courtesy Pascale Cornillet-Lefebvre and StÃ©phanie Struski.

Clinics and pathology

Disease

Myelodysplastic syndromes (MDS) and acute non lymphocytic leukemias (ANLL) and therapy related ANLL.

Phenotype/cell stem origin

M1 or M2 ANLL.

Etiology

11 of 15 cases have had treatment for a previous malignancy (treatment related MDS or ANLL (t-MDS/ANLL)). Previous disease was a breast cancer in 5 cases, a hematologic malignancy in 4.

Epidemiology

15 available cases at least, sex ratio: 2M / 13F; mediane age around 50 yrs (range <15 - 62).

Clinics

Blood data: pancytopenia.

Prognosis

Poor.

Cytogenetics

Additional anomalies

+ 8 found in 7 of 15 cases.

Genes involved and proteins

CBFA2T3 (MTG16)

Location
16q24

Protein
Member of the ETO (MTG8) family.

AML1

Location
21q22

DNA/RNA
Transcription is from telomere to centromere.

Protein
Contains a Runt domain and, in the C-term, a transactivation domain; forms heterodimers; widely expressed; nuclear localisation; transcription factor (activator) for various hematopoietic-specific genes.

Result of the chromosomal anomaly

Hybrid gene

Description
5' AML1 - 3' CBFA2T3

To be noted

Case Report
A case of trisomy 8 and loss of the Y-chromosome as secondary aberrations in a ten year old boy with de novo AML FAB M2 and t(16;21)(q24;q22).

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