Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Leukaemia Section

Mini Review

t(10;11)(p11.2;q23)

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This article is an update of: Huret JL. t(10;11)(p11.2;q23). Atlas Genet Cytogenet Oncol Haematol.2000;4(2):71.

Identity

Note: must not be confused with the t(10;11)(p12;q23) involving AF10 in 10p12 and MLL, or the t(10;11)(p13;q14-21), also involving AF10, but with CALM on chromosome 11.

Clinics and pathology

Disease

Acute non lymphoblastic leukemia (ANLL).

Phenotype / cell stem origin

M4/M5.

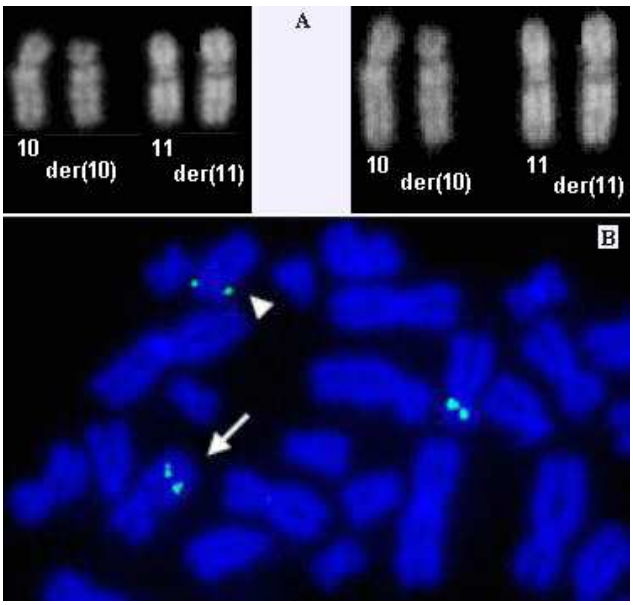
Epidemiology

Only three cases reported to date: all infants (2M/1F).

Clinics

Two boys aged 2 and 8 months respectively, achieved complete remission (1 years+, 5 years+), the newborn girl died soon for infection during induction.

Cytogenetics



A. Partial Q-banded karyotype showing the t(10;11)(p11.2;q23), derivative chromosomes are on the right.
B. FISH using RP13-31H8 (ABI1) shows one signal on the normal chromosome 10 and the another one split between the p arm of der(10) (arrowheads) and the q arm of der(11) (arrow). The BAC clone was provided by Prof. M.Rocchi.

Genes involved and Proteins

ABI-1

Location: 10p11.2

DNA / RNA

Different splicings.

Protein

Possesses a SH3 domain; cell growth inhibitor.

MLL

Location: in 11q23

DNA / RNA

13-15 kb mRNA.

Protein

431 kDa; contains two DNA binding motifs (a AT hook, and Zinc fingers), a DNA methyl transferase motif, a bromodomain; transcriptional regulatory factor; nuclear localisation.

Results of the chromosomal anomaly

Hybrid gene

Description

5' MLL - 3' ABI1; fusion at MLL exon 6-7.
The breakpoint of ABI1 gene is the same in the two cases studied (nucleotide 433), while the breakpoint of MLL can be located either in exon 6 or 7.

Fusion protein

Description

1727 amino acids (1406 from MLL and 321 from ABI-1); NH2-AT-hook, DNA methyltransferase, and transcriptional repression domain of MLL, fused to the homeodomain homologous region and the SH3 domain of ABI-1 in COOH.

References

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Shibuya N, Taki T, Mugishima H, Chin M, Tsuchida M, Sako M, Kawa K, Ishii E, Miura I, Yanagisawa M, Hayashi Y. t(10;11)-acute leukemias with MLL-AF10 and MLL-ABI1 chimeric transcripts: specific expression patterns of ABI1 gene in leukemia and solid tumor cell lines. Genes Chromosomes Cancer 2001;32:1-10.

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