**Leukaemia Section**  
**Mini Review**

**dic(9;12)(p13;p13)**

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**Identity**

**Clinics and pathology**

**Disease**

Acute lymphocytic leukemia (ALL) most often; rarely: chronic myelogenous leukemia (CML) in blast crisis, T-cell leukaemia or non Hodgkin lymphoma (NHL).

**Phenotype/cell stem origin**

ALLs with dic(9;12) are most often L1/L2 and CD10+, at times CIg+ ALL.

**Epidemiology**

1% of paediatric ALL; sex ratio: 2M/1F; children and young adults. (>1yr, <25yrs); no infant case.

**Clinics**

Moderate organomegaly; blood data: moderate WBC.
Treatment
Bone marrow transplantation is not indicated; no high risk protocol.

Prognosis
Complete remission is obtained in all cases; 5 yrs survival > 95%.

Cytogenetics

Cytogenetics morphological
Dicentric with loss of parts of 9p and 12p --> ploidy: 45 chromosomes.

Additional anomalies
+8, +21.

Genes involved and proteins

PAX5
Location
9p13
DNA/RNA
The PAX5 coding region extends over a genomic interval of approximately 200kb and comprises 10 exons. Two alternative transcripts have been identified, originating from alternative promotor usage, containing exon 1A or 1B. Full length mRNA is 3650bp.

Protein
PAX5 belongs to the paired box family of transcription factors, involved in a multitude of developmental processes. PAX5 was originally identified as a B-cell specific transcription factor (B-cell-specific activator protein, BSAP). Recently, it has been shown that PAX5 expression is not only continuously required for B cell lineage commitment during early B cell development but also for B lineage maintenance. Contains a paired box (DNA binding) domain, a truncated homeo domain homology region, and a transactivation domain.

ETV6
Location
12p13

DNA/RNA
Alternative transcripts.

Protein
Contains a HLH domain and a ETS-DNA binding domain; ETS-related transcription factor, transcriptional repressor that binds to DNA sequence 5'-CCGGAAGT-3', can form homodimers or heterodimers with TEL2 or FLI1.

Result of the chromosomal anomaly

Hybrid gene
Description
Fusion of PAX5 to ETV6; constant breakpoints in PAX5 intron 4 and ETV6 intron 2, in the three cases described so far.

Transcript
5'PAX5-3'ETV6 transcript, no reciprocal transcript due to deletion.

Detection
RT-PCR, FISH.

Fusion protein
Description
The PAX5/ETV6 chimeric transcript results in fusion of the paired box domain (PRD) of PAX5 to the helix-loop-helix and ETS-binding domains of ETV6. Of note: the putative chimeric protein contains the DNA-binding domains of both fusion partners, namely the PRD and the ETS-domain.

To be noted
Note
Bone marrow transplantation should not be performed, as the prognosis of the dic(9;12)/ALL is excellent. Cases with dic(9;12) involvement are collected and analyzed for the presence of the PAX5/ETV6 fusion. If you are interested in participating in this study, please contact: sabine.strehl@ccri.at

References


This article should be referenced as such: