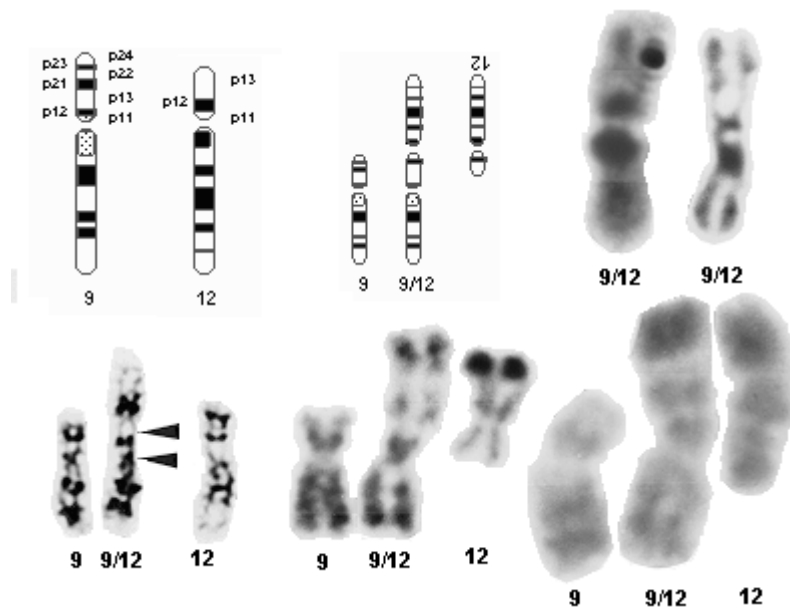


dic(9;12)(p13;p13) (updated: old version not available)

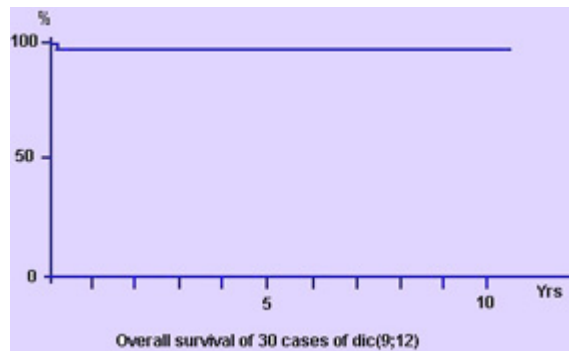
Identity



dic(9;12)(p13;p13) diagram and breakpoints and C-banding (above) - Editor; G-banding (left) - Courtesy Jean-Luc Lai; R- banding with chr 12 up side down (right)- Editor

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 Clg+ 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 dicentric with loss of parts of 9p and 12p --> ploidy: 45 chromosomes
Morphological

Additional anomalies +8, +21, ...

Genes involved and Proteins

Gene Name [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 promoter 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.

Gene Name [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.

External links

Other database [dic\(9;12\)\(p13;p13\)](#) [Mitelman database \(CGAP - NCBI\)](#)

To be noted

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

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