Leukaemia Section
Short Communication

dic(9;12)(p13;p13)

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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 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.

**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
- **Protein**: Transcription factor B-cell specific activator.

**ETV6**
- **Location**: 12p13
- **DNA/RNA**: Alternative transcripts.

**Protein**
Belongs to the ETS transcription factors family characterized by the ETS domain, domain which is responsible for the sequence specific DNA-binding activity.

**Result of the chromosomal anomaly**

**Hybrid gene**
*Description*
Fusion of PAX5 exon 4 to ETV6 exon 3.

**Fusion protein**
*Description*
Fusion of the paired box domain of PAX5 to the HLH and ETS binding domains of ETV6.

**To be noted**

**Note**
Bone marrow transplantation should not be performed, as the prognosis of the dic(9;12)/ALL is excellent.

**References**

*This article should be referenced as such:*