Leukaemia Section
Short Communication

dic(9;16)(p13;q11) PAX5/?

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Abstract

Short communication on dic(9;16)(p13;q11) PAX5/?, with data on clinics, and the genes implicated.

Clinics and pathology

Disease

Acute lymphoblastic leukemia (ALL)

Epidemiology

Only one case to date, a 15-year old boy with a pre-B-ALL (Coyaud et al., 2010).

Cytogenetics

Cytogenetics morphological

The dic(9;16) was the sole abnormality within a subclone.

Genes involved and proteins

PAX5

Location
9p13.2

Protein
391 amino acids; from N-term to C-term, PAX5 contains: a paired domain (aa: 16-142); an octapeptide (aa: 179-186); a partial homeodomain (aa: 228-254); a transactivation domain (aa: 304-359); and an inhibitory domain (aa: 359-391).

Result of the chromosomal anomaly

Hybrid gene

Description
Truncation of PAX5 after exon 5. The region in 16q11 does not contain any gene.

Fusion protein

Description
256 amino acids. The truncated protein contains the DNA binding paired domain and octapeptide of PAX5 (201 aa) and 55 aa from contiguous introns.
Truncated PAX5.

**References**


*This article should be referenced as such:*