Leukaemia Section

Short Communication

dic(3;9)(p14;p13) PAX5/FOXP1

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Published in Atlas Database: August 2010


DOI: 10.4267/2042/45026

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Identity

Note
See also the paper on dic(9;20)(p11-13;q11).

Clinics and pathology

Disease
Acute lymphoblastic leukaemia (ALL).

Phenotype/cell stem origin
B-cell precursor ALL.

Epidemiology
One case to date (Mullighan et al., 2007).

Prognosis
No data.

Genes involved and proteins

**PAX5**

Location
9p13.2

Protein
Lineage-specific transcription factor; recognizes the consensus recognition sequence GNCCANTGAAGCGTGAC, where N is any nucleotide. Involved in B-cell differentiation.

Entry of common lymphoid progenitors into the B cell lineage depends on E2A, EBF1, and PAX5; activates B-cell specific genes and represses genes involved in other lineage commitments. Activates the surface cell receptor CD19 and represses FLT3. Pax5 physically interacts with the RAG1/RAG2 complex, and removes the inhibitory signal of the lysine-9-methylated histone H3, and induces V-to-DJ rearrangements. Genes repressed by PAX5 expression in early B cells are restored in their function in mature B cells and plasma cells, and PAX5 repressed (Fuxa et al., 2004; Johnson et al., 2004; Zhang et al., 2006; Cobaleda et al., 2007).

**FOXP1**

Location
3p14

Protein
Transcriptional repressor. Involved in cardiomyocyte proliferation, motor neuron migration, B-lymphocyte development, and the generation of quiescent naïve T cells (Shi et al., 2008; Feng et al., 2010; Rao et al., 2010; Zhang et al., 2006; Hisaoka et al., 2010).

Result of the chromosomal anomaly

**Hybrid gene**

Description
Fusion of PAX5 exon 6 to FOXP1 exon 7.
**Fusion protein**

The predicted fusion protein contains the DNA binding paired domain of PAX5 and the DNA-binding and transcriptional regulator domain of FOXP1. 877 amino acids.

**References**


**This article should be referenced as such:**