

Leukaemia Section

Short Communication

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

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Published in Atlas Database: August 2010

Online updated version : <http://AtlasGeneticsOncology.org/Anomalies/dic0309p14p13ID1553.html>

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 GNCANTGAAGCGTGAC, 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 repress genes involved in other lineage commitments. Activates the surface cell receptor CD19 and repress 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 naive T cells (Shi et al., 2008; Feng et al., 2010; Rao et al., 2010; Zhang et al., 2010; Hisaoka et al., 2010).

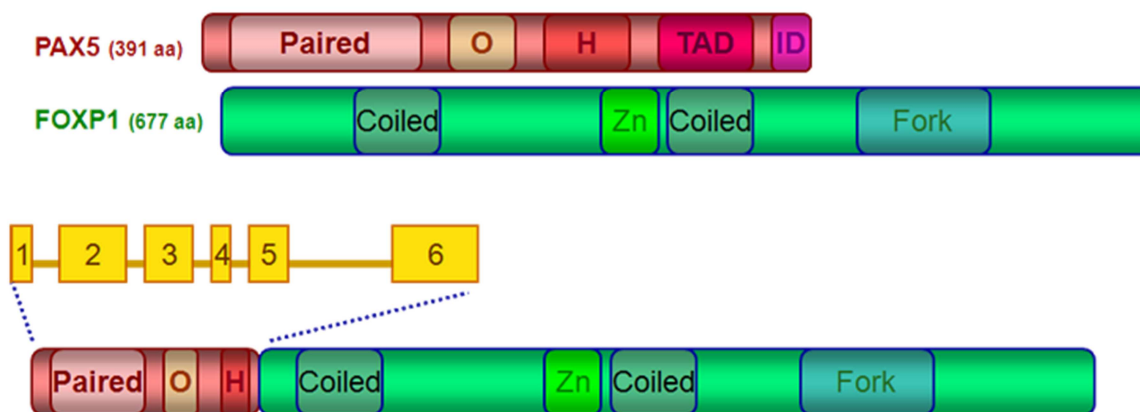
Result of the chromosomal anomaly

Hybrid gene

Description

Fusion of PAX5 exon 6 to FOXP1 exon 7.

Fusion protein



Paired: paired domain; O: octapeptide; H: homeodomain; TAD: transactivation domain; ID: inhibitory domain;
Coiled: coiled-coil domain; Zn: zinc finger; Fork: fork-head domain

dic(3;9)(p14;p13) PAX5/FOXP1 (877 aa) Editor, adapted from Mullighan et al., Nature 2007.

Description

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.

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This article should be referenced as such:

Huret JL. dic(3;9)(p14;p13) PAX5/FOXP1. *Atlas Genet Cytogenet Oncol Haematol.* 2011; 15(5):463-464.