Leukaemia Section
Short Communication

dic(9;18)(p13;q11) PAX5/ZNF521

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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 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).

ZNF521
Location
18q11
Protein
Transcription factor. Involved in B-cell lineage commitment, in the differentiation of neural progenitors; Inhibits EBF1. Binds Runx2 and represses its transcriptional activity. Regulates osteoblast differentiation and bone formation (Bond et al., 2008; Lobo et al., 2008; Wu et al., 2009; Hesse et al., 2010).

Result of the chromosomal anomaly

Hybrid gene
Description
Fusion of PAX5 exon 7 to ZNF521 exon 4.
**Fusion protein**

\[
\text{PAX5 (391 aa)} \quad \text{Paired} \quad \text{O} \quad \text{H} \quad \text{T} \quad \text{ID} \\
\text{ZNF521 (1311 aa)} \quad \text{DNA} \quad \text{BRE} \quad \text{SMAD} \quad \text{EBF}
\]

1. Paired: paired domain; O: octapeptide; H: homeodomain; T: transactivation domain; ID: inhibitory domain
2. DNA: DNA binding domain; BRE: BMP response element domains; SMAD: SMAD interacting domain; EBF: EBF interaction domains

**Description**

The predicted fusion protein contains the DNA binding paired domain of PAX5 and the DNA-binding and transcriptional regulator domain of ZNF521. 1541 amino acids.

**References**


This article should be referenced as such: