Leukaemia Section
Short Communication

dic(9;12)(p13;p12) PAX5/SLCO1B3

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Identity

Note
While dic(9;12)(p13;p12) is usually associated with PAX5/ETV6 involvement, one case has been described with SLCO1B3 involvement instead of ETV6. 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, a 1-year-old girl (An et al., 2008).

Prognosis
No data.

Cytogenetics

Additional anomalies
The dic(9;12) was the sole anomaly.

Genes involved and proteins

PAX5

Location
9p13.2

Protein
Lineage-specific transcription factor; recognizes the consensus recognition sequence

SLCO1B3

Location
12p12.2

Protein
Multi-pass membrane protein. Organic anion transporting polypeptide. Mediates the transport for various molecules such as bile acids, steroids, thyroid hormones, and exogenous drugs. Normally expressed in the basolateral membrane of hepatocytes around the central vein (Hagenbuch and Meier, 2003; Briz et al., 2006).

Result of the chromosomal anomaly

Hybrid gene

Description
Break in PAX5 intron 4. Out of frame fusion of 5' PAX5 - 3' SLCO1B3

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).
**Fusion protein**

The predicted fusion protein contains the DNA binding paired domain of PAX5.

**Oncogenesis**

Loss of function of PAX5 is likely to be the oncogenic event.

**References**


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This article should be referenced as such: