Leukaemia Section
Short Communication

dic(7;9)(p11-12;p12-13) PAX5/LOC392027
Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

Published in Atlas Database: February 2010
DOI: 10.4267/2042/44899

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.
© 2010 Atlas of Genetics and Cytogenetics in Oncology and Haematology

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
13 cases to date; sex ratio was 7M/6F, median age was 17 years (range 2-51 years) (An et al., 2008).

Prognosis
No data.

Cytogenetics

Additional anomalies
The dic(7;9) was the sole anomaly in 8 cases, accompanied a t(9;22) with BCR-ABL1 involvement in 3 cases, and was accompanied with other anomaly in 2 other cases, including a del(6q).

Genes involved and proteins

LOC392027

Location
7p12

Protein
Ribosome-binding protein 1 pseudogene.

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).

Result of the chromosomal anomaly

Hybrid gene

Description
Break in PAX5 intron 4. Out of frame fusion of 5' PAX5 - 3' LOC392027.
Fusion protein

Description
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


This article should be referenced as such: