

Leukaemia Section

Short Communication

t(5;14)(q35;q32)

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Published in Atlas Database: February 2002

Online updated version: <http://AtlasGeneticsOncology.org/Anomalies/t0514q35q32ID1227.html>

DOI: 10.4267/2042/37849

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Clinics and pathology

Disease

T cell acute lymphoblastic leukemia (ALL).

Phenotype/cell stem origin

Immature cortical T cell leukemia.

Epidemiology

8 cases so far described; this cryptic anomaly (see below) may nonetheless be frequent in T-cell ALL in children (representing 15-20% of T-cell ALL cases in children); not found in adults, nor in B-cell ALL; male predominance (7M: 1F); med age: 9-13 yrs (range 5-19).

Clinics

High WBC in 3 of 8 patients; organomegaly.

Cytology

L2 or L1 ALL; CD1a+.

Cytogenetics

Cytogenetics morphological

Cryptic translocation, and no additional anomaly in most cases; therefore, most patients present with a normal karyotype.

Cytogenetics molecular

Chromosome painting is needed.

Additional anomalies

+8 in one case.

Genes involved and proteins

Note

HOX11L2, in 5q35, is likely to be involved, but RanBP17 also lies in the proximity of the breakpoint. The gene involved in 14q32 may be CTPI2.

HOX11L2

Location

5q35

Protein

DNA binding (homeobox) domain; role during embryonic development; and also in adult tissues.

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

HOX11L2 is transcriptionally activated; the translocation brings CTIP2 close to HOX11L2; CTIP2 is highly expressed in T-cells.

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This article should be referenced as such:

Huret JL. t(5;14)(q35;q32). *Atlas Genet Cytogenet Oncol Haematol*. 2002; 6(2):130-131.
