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Leukaemia Section
Short Communication

t(5;14)(q35;q32)

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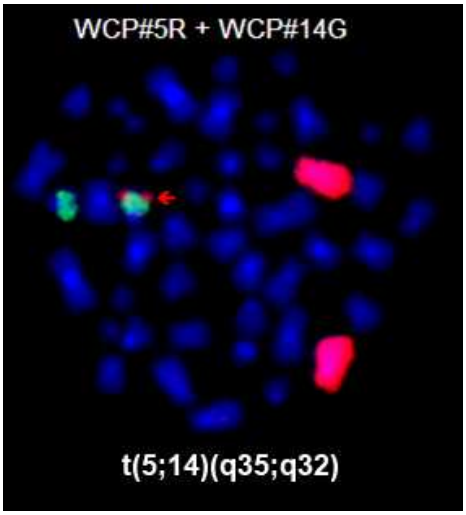

t(5;14)(q35;q32) FISH - Courtesy Melanie Zenger and Claudia Haferlach.

Clinics and pathology

Disease

T cell acute lymphoblastic leukemia (ALL).

Phenotype/cell stem origin

Cortical T cell leukemia (CD1a+, CD10+).

Epidemiology

Frequent in T-cell ALL in children (in about 20% of childhood T-cell ALLs); less frequent in adult T-ALL. Not seen in B-cell ALL.

Cytology

FAB nomenclature: L1 or L2 ALL.

Prognosis

Present data suggest that t(5;14)(q35;q32) is associated with poor outcome, but confirmatory data is necessary prior to conclude.

Cytogenetics

Cytogenetics morphological

Cryptic translocation (banded karyotype). Often apparently normal karyotype with banding techniques.

Cytogenetics molecular

t(5;14)(q35;q32) can be detected with FISH techniques. Several probes may be used: chromosome painting, combination of painting probes and YAC, multicolor-

FISH with adequate probes. The localization of the chromosomal breakpoint with BACs/PACs will be performed in a second step.

Additional anomalies

Variable.

Genes involved and proteins

Note

The consequence of the translocation is the ectopic expression of the HOX11L2, gene normally located to 5q35, and normally not expressed in ALL without 5q rearrangement. The "deregulation" of HOX11L2 expression is thought to result from abnormal control of the gene by CTPI2, located to 14q32, as a consequence of the chromosomal rearrangement. The chromosome 5 breakpoint is usually located within the locus of another gene, RanBP17, often disrupted by the chromosomal rearrangement. The breakpoint on chromosome 5 is consequently distant from the gene abnormally expressed (HOX11L2).

HOX11L2

Location

5q35

Protein

Homeobox domain; belongs to HOX 11 family.

Result of the chromosomal anomaly

Fusion protein

Description

No fusion protein, but abnormal expression of HOX11L2.

Oncogenesis

HOX11L2 is transcriptionally activated, due to control by CITP2 regulatory sequences.

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