Leukaemia Section
Short Communication

t(14;21)(q11;q22)
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Clinics and pathology

Disease
T-cell Acute lymphoblastic leukemia (T-ALL)

Epidemiology
Rare.

Clinics
One case reported: The patient was a 7-years-old female with high white blood count with lymphoblasts positive for T-cell antigens; cerebrospinal fluid negative for malignant cells; superior mediastinal mass.

Prognosis
The patient attained a complete remission with standard chemotherapy but relapsed and died after 4 months of therapy.

Cytogenetics

Cytogenetics morphological
The t(14;21)(q11.2;q22) was accompanied with del(6)(q21). Cryptic t(14;21) translocation cases may exist.

Genes involved and proteins

TCRA
Location
14q11.2
Protein
T cell receptor.

BHLHB1
Location
21q22

Note
Alias OLIG2.

DNA/RNA
This gene maps within a 9 to 12 Mb region of chromosome 21q22.

Protein
This gene was shown to possess a helix-loop-helix (bHLH) motif which inhibits the E2A function in transfection assays. E2A is required for normal T-cell differentiation.

Result of the chromosomal anomaly

Hybrid gene
Note
Translocation of the BHLHB1 gene 130kb upstream of the TCRA enhancer. This translocation activates the BHLHB1 gene and produces high levels of BHLHB1 mRNA. Expression of HLHB1 inhibits E2A-mediated transcription activation in vitro.

Fusion protein
Note
No fusion protein.

Oncogenesis
Several helix-loop-helix (HLH) proteins are proposed to function as transcriptional regulatory factors based on their ability to bind in vitro the E-box motif of transcriptional enhancers.

The observation that ectopic BHLHB1 expression can inhibit E2A activity suggests that BHLHB1 exerts its leukemogenic effects through a functional inhibition of E2A.
References


This article should be referenced as such: