Leukaemia Section

Mini Review

t(7;9)(q34;q32)

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Clinics and pathology

**Disease**
Specifically associated with T-cell acute lymphoblastic leukemia (T-ALL).

**Epidemiology**
The t(7;9)(q34;q32) is present in two cases of a series of 5 patients with 7q34 involvement.

**Clinics**
Children with large mediastinal mass.

**Cytology**
The hematological feature of the T-ALL with rearrangement of 7q34 shows high WBC (range 95 to 400 x 10⁹/L).

Cytogenetics

**Cytogenetics morphological**
9q32 is a partner of 7q34. The other partners are 1p34, 1p32, 9q34, 10q24, 11p13, 15q22 and 19p13.

**Additional anomalies**
del(6)(q21), del(4)(q31q35).

Genes involved and proteins

**TCRB (T-cell receptor beta-chain)**

**Location**
7q35

**DNA/RNA**
The TRB locus at 7q35 spans 685 kb. The locus contains 2 types of coding elements: TCR elements (64-67 variable genes TRBV, 2 clusters of diversity, joining and constant segments) and 8 trypsinogen genes. A portion of the TCRB locus has been duplicated and translocated to the chromosome 9 at 9p21.

**Protein**
T cell receptor beta chains.

**TAL2**

**Location**
9q32

**DNA/RNA**
The TAL2 gene is located at q32.

**Protein**
TAL2 potentially encodes a basic helix-loop-helix (bHLH) phosphoprotein (size 108 amino acids) that is highly related to those specified by TAL1 and LYL1 also implicated in T-ALL. The bHLH protein interacts with the product of RBTN1 and RBTN2 (cysteine-rich LIM motifs).

Result of the chromosomal anomaly

**Hybrid gene**

**Description**
TAL2 is transcriptionally activated by t(7;9)(q34;q32) in T-ALL. The chromosome 9 breakpoints of the t(7;9)(q34;q32) occur 33 kbp downstream of sequences that encode the TAL2 HLH domain. Translocated TAL2 are juxtaposed with transcriptional regulatory elements within the T-cell receptor beta-chain locus.

**Fusion protein**

**Note**
No fusion protein.

**Oncogenesis**
The TAL2 transcription is activated ectopically in lymphoid cells and the inappropriate expression of...
TAL2 in these cells promotes development of T-ALL. Normally, the TAL genes are not expressed in the thymus. The TAL genes become activated and expressed in the thymus upon chromosomal translocation which ultimately leads to the development of T-ALL.

The (7;9) translocation express a TAL2 gene product of 108 amino acids. In leukemic cells this product exists in both a phosphorylated and an unphosphorylated form. Serine residue 100 is the major site of TAL2 phosphorylation in vivo. And it serves as an effective in vitro substrate for MAP kinases such as ERK1. TAL2 polypeptides interact in vivo with the E2A gene products to form HLH heterodimers that bind DNA, the result is the E2A inactivation. The E2A products are transcriptional factors implicated in the B and T cell development.

TAL2 product was also shown to bind with a GTP binding protein (DRG). The properties of TAL2 broadly resemble those described previously for TAL1 and therefore support the idea that both encoded proteins promote T-ALL by a common mechanism and the malignant potential of these proteins is likely to reside within their HLH domains though the inactivation of E2A.

To be noted

Case Report
Translocation t(7;9)(q34;q32) found in pediatric T-cell acute lymphoblastic leukemia.

References

This article should be referenced as such: