Leukaemia Section

Mini Review

t(14;18)(q32;q21)
t(2;18)(p11;q21)
t(18;22)(q21;q11)

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Identity

Note: The 3 translocations are variants of each other, and they share the same clinical significance.

Left, from top to bottom: t(2;18)(p11;q21), t(14;18)(q32;q21), t(18;22)(q21;q11), G-banding - Courtesy Jean-Luc Lai and Alain Vanderhaegen; and, right: t(14;18)(q32;q21), R-banding - Editor.
Clinics and pathology

Disease
B-cell NHL mainly; may (rarely) be found in ALL or in chronic lymphoproliferative diseases.

Phenotype / cell stem origin
B lymphocyte; the translocation occurs at the pre B cell stage.

Epidemiology
Found in 80 to 90% of follicular lymphomas, 30% of diffuse large cell lymphomas (sometimes with prior follicular type), rarely in other lymphoproliferative disorders.

Prognosis
Small cleaved cell follicular lymphomas have a slow evolutivity and a median survival of 10 yrs or more; large cell lymphomas have a worse prognosis; the t(14;18) may have little or no prognostic significance, which would be in accordance with its 'oncogenic' role (see below).

Cytogenetics

Genes involved and Proteins

IgH
Location: 14q32
DNA / RNA
IGH is composed of IGHV genes, IGHD segments, IGHJ segments, and IGHC genes.

Protein
IGH encodes the immunoglobulin heavy chains. They result from the DNA rearrangement (with deletion of the intermediary DNA) of IGHV, IGHD and IGHJ; an additional DNA rearrangement (switch) with constant genes IGHM → IGHG, IGHA and IGHE can occur.

BCL2
Location: 18q21
DNA / RNA
3 exons; 2 alternative transcripts (a and b).

Protein
Member of the BCL2 family (with BAX, BCLX,...); contains dimerization domains (BH) and NH domains; BCL2a contains a hydrophobic tail for membrane anchorage; inhibits cell death process through heterodimerization.

Results of the chromosomal anomaly

Hybrid gene
Description
5' BCL2 translocated on chromosome 14 near JH (junctions genes of IgH) and C in 3'; the breakpoint in BCL2 is either in the 3' untranslated region of exon 3 (major breakpoint region (MBR) in 70% of cases) or more distal, in 3' of exon 3 (minor cluster region (MCR) in the remaining 30%); illegitimate recombination.

Fusion protein
Description
No fusion protein, but promoter exchange; the immunoglobulin gene enhancer stimulates the expression of BCL2.
Oncogenesis
As BCL2 is an apoptosis inhibitor, cell death is delayed, and there is cell accumulation more than real transformation (clones of long living B-cells with t(14;18) may even be seen in the normal population, and with frequency increasing with age).

References


This article should be referenced as such:
Huret JL. t(14;18)(q32;q21); t(2;18)(p11;q21); t(18;22)(q21;q11). Atlas Genet Cytogenet Oncol Haematol. 1998;2(4):134-136.