Leukaemia Section
Short Communication

\(t(8;12)(q24;q22)\)

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France

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Clinics and pathology

Disease
Chronic lymphocytic leukemia (CLL) in blastic transformation.

Note: CLL was diagnosed after bone marrow recovery following intensive chemotherapy for acute lymphoblastic leukemia.

Epidemiology
Only one case to date, 69-year-old female patient.

Cytogenetic
The patient had a primary \(t(11;14)(q13;q32)\) in all lymphocytic/lymphoblastic cells. A subpopulation of cells carried in addition a \(t(8;12)(q24;q22)\) together with the loss of the normal chromosome 8 and homozygosity for the abnormal der(8). It is likely that the \(t(11;14)\) corresponded to the CLL, and the subclone with \(t(8;12)\) to the blast population.

Prognosis
The patient died 5 months after initial blastic phase.

Genes involved and Proteins

\textbf{MYC}

Location: 8q24

Protein
Transcription factor binding to specific DNA sequences, upon dimerization to Max (Max can also form heterodimers with Mad as well as homodimers with itself). Myc/Max complexes activate transcription and promote cell proliferation and transformation. Mad/Max complexes, repress transcription.

\textbf{BTG1}

Location: 12q22

Protein
B-cell translocation gene 1 (BTG1) is an anti-proliferative gene; it regulates cell growth and differentiation. BTG1 is strongly expressed in the G0/G1 phases of the cell cycle, and then down-regulated during the G1 phase. Overexpression of BTG1 results in a retardation of cell proliferation. Potentially high-affinity self-reactive B cells are eliminated through the interaction of membrane Ig (mIg) with self-antigens. Anti-IgM upregulates BTG1 and BTG2, resulting in growth inhibition. Engagement of mIg on the cells results in G1 arrest and eventual apoptosis.

References


This article should be referenced as such: