Leukaemia Section
Short Communication

t(4;14)(p16;q32)
Jean-Loup Huret, Jacky Bonaventure

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH); Unité INSERM 393, Hopital Necker-Enfants Malades, 149 rue de Sèvres 75743, Paris Cedex 15, France (JB)

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Clinics and pathology

**Disease**
Found in plasma cell leukaemia and multiple myeloma.

**Phenotype / cell stem origin**
Malignant plasma cells have the phenotype of mature terminally differentiated B-cells; there origin may be a pluripotent stem cell.

**Epidemiology**
Yet poorly described: found in 6 fresh tumours and 5 cell lines; might be frequent but karyotypically undetected.

**Clincs**
Data on clinics and cytogenetics are missing.

Cytogenetics

**Cytogenetics, morphological**
May be undetectable (telomere-telomere translocation).

**Cytogenetics, molecular**
Therefore molecular probes are indicated, and FISH is relevant.

Genes involved and Proteins

**FGFR3**
*Location:* 4p16.3

**Protein**
Contains an extracellular domain with Ig-like loops, a transmembrane domain, and intracellular tyrosine kinase domains; localisation: plasma membrane; tyrosine kinase receptor; role in signal transduction.

**IgH**
*Location:* 14q32

Results of the chromosomal anomaly

**Hybrid gene**
*Description*
FGFR3 is translocated on der(14) which contains the 3’ IgH enhancer.

**Fusion protein**
*Description*
No fusion protein, but promoter exchange between both partner genes; however, somatic mutations similar to what has been found in thanatophoric dwarfism have been identified in some cases; they may also contribute to abnormal FGFR3 activation.

**Oncogenesis**
Overexpression and activation of FGFR3 provides an oncogenic signal.

References


This article should be referenced as such: