Atlas of Genetics and Cytogenetics in Oncology and Haematology

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# 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.

**Clinics**

Data on clinics and cytogenetics are missing.

## Cytogenetics

### Cytogenetics, morphological

May be undetectable (telomere-telomere translocation).

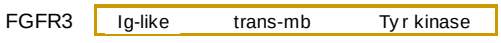
### Cytogenetics, molecular

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

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