**t(6;8)(q27;p12)**

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Clinics and pathology

**Disease**  
Multilineage disorder with combined occurrence of myeloid malignancy and T-cell NHL, or myeloid metaplasia.

**Phenotype/cell stem origin**  
The same t(6;8)(q27;p12) is found both in the bone marrow and in the lymph node: the multilineage involvement suggests the malignant transformation of a primitive hematopoietic stem cell.

**Epidemiology**  
4 cases are described; median age 29 years (range 23-48); sex ratio: 2M/2F.

**Clinics**  
Aggressive disease; complex picture of myeloid hyperplasia progressing to myelodysplasia and T-lymphoma, and acute non lymphocytic leukemia; enlarged lymph node infiltrated by myeloid blast cells; blood data: high WBC (median 40 X 10^9/l); myelemia; monocytosis and eosinophilia.

**Evolution**  
CR is obtained, but is promptly followed by relapse progressing rapidly to acute non lymphocytic leukemia.

**Prognosis**  
Median survival: 6 months.

Cytogenetics

**Cytogenetics morphological**  
Occurs as a single anomaly.

**Cytogenetics molecular**  
Mega YAC 959-A -4 (1260 kb) from CEPH; FGFR1-specific cosmid 134.8.

Genes involved and proteins

**FGFR1**  
**Location:** 8p12  
**Protein**  
FGF receptor with tyrosine kinase activity.

**FOP (FGFR1 Oncogene Partner)**  
**Location:** 6q27  
**Protein**  
Hydrophobic protein containing alpha-helices in the N- and C-termini with leucine-rich repeats.

Result of the chromosomal anomaly

**Hybrid gene**  
**Description**  
Breakpoint in FGFR1 intron 8 which encodes the juxtamembrane domain, breakpoint in FOP intron 6.

**Fusion protein**  
**Description**  
Aberrant tyrosine kinase composed of the putative leucine-rich N-terminal region of FOP, and the FGFR1 intracellular region minus the major part of the juxtamembrane domain.

**Oncogenesis**  
Through constitutive activation of FGFR1 signal transduction pathways, via putative dimerization of the fusion protein via the FOP leucine-rich repeats.

References


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