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Leukaemia Section
Short Communication

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.

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