t(7;8)(q34;p11)
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Clinics and pathology

Disease
Acute myeloid leukemia (AML) and 8p11 myeloproliferative syndrome (EMS). Chromosome band 8p11 has been implicated in recurrent chromosome rearrangements associated either with acute myeloid leukemia (AML) or with a peculiar myeloproliferative disorder named 8p11 myeloproliferative syndrome (EMS). The latter is characterized by generalized lymphadenopathy and marked eosinophilia, rapid evolution to AML, and frequent association with non-Hodgkin lymphoma. The FGFR1 gene is constantly involved in EMS, often evolving to AML.

Phenotype / cell stem origin
AML-M4 (EMS).

Clinics
1 case to date, a female patient aged 49 yrs. The differential WBC count suggested a chronic MPD. Examination of a bone marrow smear showed 44% blasts, hypoplasia, and eosinophilia. The immunophenotypic characterization revealed the coexistence of two distinct components, a myelomonocytic part along with a lymphoid population. A diagnosis of AML-M4 was established.

Treatment
Induction therapy was started with daunorubicin, cytosine arabinoside, and etoposide.

Evolution
The patient died of sepsis during aplasia on day 20.

Cytogenetics

Probes
See figure below

Additional anomalies
None

Genes involved and Proteins

TRIM24
Location: 7q34
Note: Alias TIF1
DNA / RNA
See figure a. Transcript: 2 variants. For details see the specific NCBI page.

Protein
TIF1 encodes a nuclear protein, transcription intermediary factor 1a displaying an RBCC motif (RING finger, B-BOX, and coiled-coil domains, also called tripartite motif, TRIM) in its N-terminus and PHD and bromo domains at the C-terminus (figure b).

FGFR1
Location: 8p11
DNA / RNA
See figure c. Transcript: various isoforms. For details see the specific NCBI page.

Protein
The FGFR1 gene encodes the fibroblast growth factor receptor 1, a transmembrane receptor with a tyrosine kinase (TK) domain in the intracellular C-terminus, a transmembrane (TM) domain, and 3 immunoglobulin-like C-2 type extracellular domains.
Genomic clones and genes in the FGFR1 (8p11) and TIF1 (7q34) regions. To the left are shown the positions of the RPCI11-350N15 at 8p11 and of the 2 adjacent genomic clones, RPCI11-513D5 and 675F6 (vertical white bars, genes in the genomic region of interest). On the right are the positions of the 4 clones that span the TIF1 locus (indicated by a vertical white bar) at 7q34 (RPCI11-18L16, 256C24, 199L18, and 269N18).

Genomic structure (not drawn to scale) of the TIF1 loci (numbered black boxes, exons) in figure a. The corresponding protein are represented in figure b.

Genomic structure (not drawn to scale) of the FGFR1 loci (numbered black boxes, exons) in figure c. The corresponding proteins are represented in figure d.
Results of the chromosomal anomaly

**Hybrid gene**

Note: TIF1-FGFR1 and FGFR1-TIF1: see figure e.

**Fusion protein**

Note: TIF1-FGFR1 and FGFR1-TIF1: see figure f.

**References**


The t(7;8)(q34;p11) translocation leading to fusion of the FGFR1 and TIF1 genes. Genes Chromosomes Cancer 2005;42(3):320-325.


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