t(8;13)(p12;q12) (updated: old version not available)

Clinics and Pathology

Disease	a myeloproliferative disorder that is frequently associated with T cell, or less commonly, B-cell non Hodgkin lymphoma
Phenotype / cell stem origin	may involve a stem cell involving both myeloid, T lineage, and B-cell lineage
Epidemiology	14 cases are described; median age 43 yrs (range 18-68); sex ratio: 6M/8F
Clinics	aggressive disease; complex picture of myeloid hyperplasia progressing to myelodysplasia and Tor -B- cell lymphoma; enlarged lymph node; blood data: high WBC (median 40 X 10^9/l); myelemia; monocytosis and eosinophilia
Evolution	the disease transforms to ANLL, or occasionally ALL, in a median of 6 months
Prognosis	median survival: 12 months

Cytogenetics

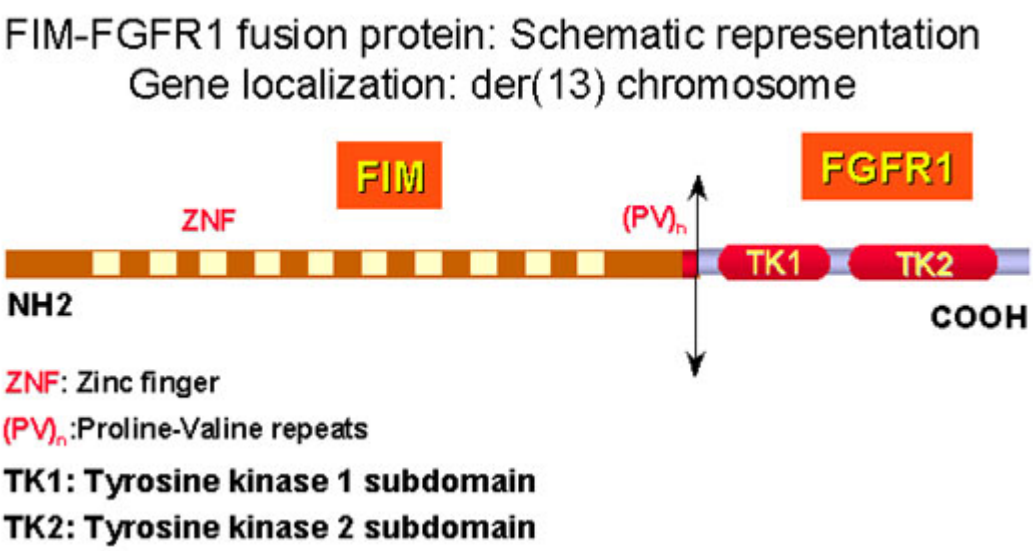
Cytogenetics Morphological	the same t(8;13) is found both in the bone marrow and in the lymph node, ruling out the hypothesis of a leukemoid reaction caused by a lymphoma; the multilineage involvement suggests the malignant transformation of a primitive hematopoietic stem cell.
Probes	megac Yacs 770-c-2 (1390 kb) and 959-a-4 (1260kb), 856-b-6, 967; 899e2 - (CEPH); BAC 7M15; PAC RPCI 20-G12; FGFR1-specific cosmid 134.8;
Additional anomalies	usually occurs as a single anomaly; duplication of the der(13) was found during disease progression, suggesting that the crucial event might lie on this derivative chromosome; +8, +21 are also recurrently found

Genes involved and Proteins

Gene Name	FGFR1
Location	8p12
Gene Name	ZNF198 (also called FIM or ID_P)
Location	13q12
Protein	zinc finger protein (ten repeats in the N-terminal region with the consensus sequence C-X2-C-X18-24-(F/Y)-C-X3-C that corresponds to a novel type of zing finger motifs), a hydrophobic repeat (proline-rich), and potentially two putative nuclear localisation signals

Result of the chromosomal anomaly

Hybrid gene



Description	breakpoint in FGFR1 intron 8
Fusion Protein Description	Aberrant tyrosine kinase composed of the N-term two-thirds of FIM (retaining the 10 putative zinc finger motifs), and the FGFR1 intracellular region minus the major part of the juxtamembrane domain (and deleting the N-term immunoglobulin-like and central transmembrane domains of FGFR1)
Oncogenesis	through constitutive activation of FGFR1 signal transduction pathways, possibly via dimerization capability mediated by the FIM N-term sequences of the fusion protein

External links

Other database	t(8;13)(p12;q12)	Mitelman database (CGAP - NCBI)
Other database	t(8;13)(p12;q12)	CancerChromosomes (NCBI)

To be noted

Additional cases are needed to delineate the epidemiology of this rare entity:
you are welcome to submit a paper to our new Case Report section.

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Contributor(s)

Written 03-1998 Jean-Loup Huret, Dominique Leroux and Alain Bernheim
Updated 12-2000 Marie-Josèphe Pébusque, and Nicholas CP Cross

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