t(8;13)(p12;q12)

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Published in Atlas Database: December 2000
Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t813ID1094.html
DOI: 10.4267/2042/37704

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Identity

![Identity Diagram]

t(8;13)(p12;q12) G-banding - Top: Courtesy Melanie Zenger and Claudia Haferlach; Middle and bottom: Courtesy Charles Bangs and Patty Jones.
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 T-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 cosmids 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

FGFR1
Location: 8p12

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-(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).

Expression / Localisation
Cytoplasmic.

Oncogenesis
Through constitutive activation of FGFR1 signal transduction pathways, possibly via dimerization capability mediated by the FIM N-term sequences of the fusion protein.
**To be noted**

**Case Report**

t(8;13)(p12;q12) in an atypical chronic myeloid leukaemia case.

**References**


Kulkarni S, Reiter A, Smedley D, Goldman JM, Cross NC. The genomic structure of ZNF198 and location of breakpoints in the t(8;13) myeloproliferative syndrome. Genomics. 1999 Jan 1;55(1):118-21


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