

# Leukaemia Section

**Short Communication** 

# t(1;22)(p13;q13)

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Published in Atlas Database: June 2001

Online updated version : http://AtlasGeneticsOncology.org/Anomalies/t0122.html

DOI: 10.4267/2042/37759

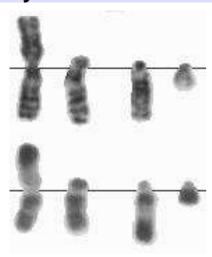
This article is an update of:

Huret JL. t(1;22)(p13;q13). Atlas Genet Cytogenet Oncol Haematol.1999;3(4):193-194.

Huret JL. t(1;22)(p13;q13). Atlas Genet Cytogenet Oncol Haematol.1997;1(1):17.

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# **Identity**



t(1;22)(p13;q13) G- and R- banding

# Clinics and pathology

## Disease

Only found so far in M7 ANLL (acute megakaryocytic leukaemia); not found in Down syndrome (DS), and yet, DS is a disease with highly elevated risk of M7 (see leukaemia and Down Syndrome); misdiagnoses of a solid tumour have been documented.

## Phenotype/cell stem origin

Megakaryocytic.

#### Etiology

No known toxic exposure.

### **Epidemiology**

About 40 known cases; 0% to 3% of paediatric ANLL; 70 to 100% of infants M7; age: infants: median age 4 mths; 20% are < 1 mth; 80% are < 1 yr; 95% are < 2 yrs; sex ratio: 15M/24F (non significant).

#### Clinics

No preceeding myelodysplasia, and no history of transient leukemoid reaction; prominent organomegaly; blood data: moderate WBC; thrombocytopenia; myelofibrosis and fibrosis of other organs.

#### Cytology

Platelet-specific markers: platelet-peroxidase by electron microscopy, or platelet glycoproteins IIb/IIIa (CD41) or IIIa (CD61).

#### **Treatment**

Bone marrow transplantation is indicated.

#### **Prognosis**

Complete remission in only 50% of cases; median survival: 8 months; a few long survivors; absence of a prognostic indicator.

# Cytogenetics

## Additional anomalies

60% of cases (mostly patients under 6 mths of age) have the t(1;22) as a single anomaly; the remaining third of cases (mainly patients above the age of 6 mths) exhibit complex and hyperploid clones, with a highly monomorph pattern: +2, +19, +der(1)t(1;22), +6, +21 were found in more than 50% of cases each, +10, +7, +15, +18, +8, +20, del(1p), +4, +9, +14, +17, add(21p)

t(1;22)(p13;q13) Huret JL

are also recurrent; survival was equivalent in cases with or without a complex karyotype; the frequent presence of an additional der(1) indicates that the crucial event is likely to lie on the der(1)t(1;22).

## **Variants**

2 cases of complex t(1;22) with a third chromosome have been described.

# **Genes involved and proteins**

# OTT (one twenty-two)

Location: 1p13

**Protein** 

Contains RNA recognition motif consensus.

## MAL (megakaryocytic acute leukemia)

Location: 22q13

**Protein** 

931 amino acids; could attach DNA to nuclear scaffold and be involved in chromatin organization

# Result of the chromosomal anomaly

# Hybrid gene

#### **Description**

5' OTT - 3' MAL, comprising most of OTT fused to most of MAL; the reciprocal 5' MAL - 3' OTT may or may not be present.

### Fusion protein

#### **Oncogenesis**

May modulate chromatin organization, HOX differentiation pathways, or extracellular signalling.

#### To be noted

#### Note

Individual data on the 39 published cases of t(1;22) and a complete bibiography can be found in our t(1;22) study group page

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This article should be referenced as such:

Huret JL. t(1;22)(p13;q13). Atlas Genet Cytogenet Oncol Haematol. 2001; 5(3):193-194.