

## Leukaemia Section

### Mini Review

## t(9;11)(p22;p15)

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Published in Atlas Database: May 2007

Online updated version: <http://AtlasGeneticsOncology.org/Anomalies/t0911p22p15ID1232.html>

DOI: 10.4267/2042/16966

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### Identity

**Note:** rare abnormality.

### Clinics and pathology

#### Disease

Acute non lymphoblastic leukemia (ANLL), one case of transformed chronic myeloid leukemia (CML-BC).

#### Phenotype / cell stem origin

ANLL FAB TYPE M1, M2, M2/M3.

### Epidemiology

Five cases reported to date: four adults and one 5-year-old girl.

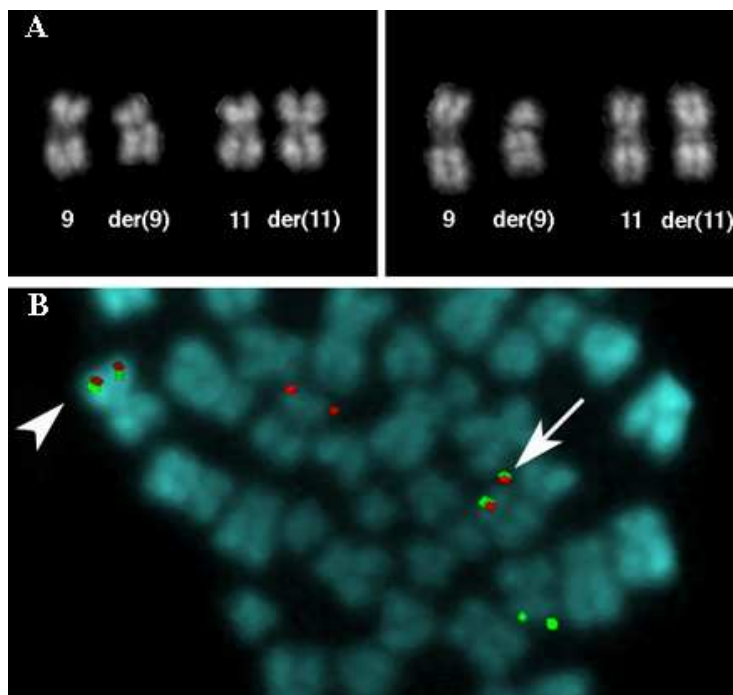
### Prognosis

Unfavorable outcome.

### Cytogenetics

#### Additional anomalies

Sole anomaly in the four ANLL cases, t(9;11) in addition to the t(9;22) in the CML-BC case.



A) Partial Q-banded karyotype showing the t(9;11)(p22;p15); derivative chromosomes are on the right of each pair. B) FISH analysis using PAC 1173K1 (NUP98) and RP11-356J15 (PSIP1) probes (green and red signals, respectively). Arrow and arrowhead indicate the fusion signals on the der(9) and the der(11), respectively.

## Genes involved and Proteins

### **NUP98**

**Location:** 11p15.5

#### **Protein**

Nucleoporin 98, a 98 kDa component of the nuclear pore complex involved in nucleo-cytoplasmic transport.

### **PSIP1 (PC4 and SFRS1 interacting protein 1)**

Aliases LEDGF (lens epithelium-derived growth factor), p75, p52

**Location:** 9p22.3

**Note:** The gene contains at least 15 exons and 14 introns.

#### **DNA / RNA**

Two alternative splice variants: p75 and p52.

#### **Protein**

Chromatin-associated protein involved in transcriptional regulation, mRNA splicing and cell survival in vitro. Contains a PWWP domain and AT hook-like motifs.

## Results of the chromosomal anomaly

### **Hybrid gene**

#### **Description**

5'NUP98 - 3'PSIP1; The breakpoint in the NUP98 gene is the same in three out of four cases studied (nucleotide 1230), while the breakpoints in PSIP1 are variable.

### **Fusion protein**

#### **Description**

It fuses the GLFG repeat domains of NUP98 to the COOH-terminal of the PSIP1.

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*This article should be referenced as such:*

Morerio C, Panarello C. t(9;11)(p22;p15). *Atlas Genet Cytogenet Oncol Haematol.* 2007;11(4):322-323.

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