Leukaemia Section
Short Communication

\(t(8;21)(q24;q22)\)

Jean-Loup Huret

Genetics, Dept Medical Information, UMR 8125 CNRS, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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Clinics and pathology

**Disease**
T-cell acute lymphoblastic leukemia (ALL) and acute non lymphocytic leukemia (ANLL).

**Phenotype/cell stem origin**
1 case of T-cell ALL and 2 cases of ANLL, one of which was a M4.

**Epidemiology**
2 documented cases, male patients aged 5 yrs (ALL case) and 42 yrs (ANLL case).

Cytogenetics

**Cytogenetics morphological**
+21 (ALL case); complex karyotype (ANLL case).

Genes involved and proteins

**Note**
This translocation may be heterogeneous at the molecular level, as it is concerning the phenotype.

**TRPS1**

**Location**
8q24

**Protein**
Transcriptional repressor.

**Germinal mutations**
Involved in tricho-rhino-phalangeal syndrome.

**Somatic mutations**
Involved with AML1 in the M4-ANLL case.

**AML1**

**Location**
21q22

**DNA/RNA**
Transcription is from telomere to centromere

**Protein**
Contains a Runt domain and, in the C-term, a transactivation domain; forms heterodimers; widely expressed; nuclear localisation; transcription factor (activator) for various hematopoietic-specific genes.

References


Asou N, Matsuno N, Mitsuya H. AML1-TRPS1 chimeric protein is generated by \(t(8;21)(q24;q22)\) in relapsing acute myeloblastic leukemia. ASH 43 Annual meeting, Blood 2001; 98(11):564a.


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