

Leukaemia Section

Short Communication

t(5;21)(q13;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

Myelodysplastic syndrome (MDS) and acute non lymphocytic leukemia (ANLL).

Phenotype/cell stem origin

1 case of refractory anemia with excess of blasts in transformation (RAEB-t), 1 MDS evolving towards a M4-ANLL, 2 M2-ANLL, and 1 ANLL not otherwise specified.

Epidemiology

5 cases to date; 3M/2F, aged 58 yr (median, range: 31-75).

Cytogenetics

Cytogenetics morphological

Sole anomaly in 2 cases, complex karyotypes in 2 other cases.

Genes involved and proteins

Note

The gene in 5q13 is yet unknown, and, because cryptic t(12;21) ETV6 /AML1 are not rare, it is therefore uncertain whether this translocation involve a new AML1 partner.

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.

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