Leukemia Section
Short Communication

t(5;21)(q13;q22)
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Published in Atlas Database: February 2003
Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0521q13q22ID1174.html
DOI: 10.4267/2042/37964

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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.

References


This article should be referenced as such: