t(3;21)(q26;q22)

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Identity

Cytology
Presence of micromegakaryocytes, both in BC-CML and MDS/ANLL cases; low platelet count and dysmyelopoiesis in MDS/ANLL cases.

Clinics and pathology

Disease
CML-BC of myeloid type (as far as 1% of cases); ANLL and MDS, often therapy related.

Phenotype / cell stem origin
No FAB specificity.

Epidemiology
>1% of ANLL; all ages represented.

Clinics
May be secondary to toxic exposure, as to antitopoisomerase II.

Cytology
Presence of micromegakaryocytes, both in BC-CML and MDS/ANLL cases; low platelet count and dysmyelopoiesis in MDS/ANLL cases.
Prognosis
Poor survival.

Genes involved and Proteins

**EVI1**

**Location:** 3q26
**Note:** or EAP (129 amino acids; putative nuclear localization signal) and/or MDS1 (rich in: proline, serine, and acidic residues), both also in 3q26.

**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.

Results of the chromosomal anomaly

**Hybrid gene**

**Description**
Fusion gene: on the der(3); 5' AML1 - 3' EVI1 (or 5' AML1 - 3' EAP/MDS1).

**Fusion protein**

**Description**
AML1-EVI1: 180 kDa; breakpoint after exon 5 or 6 in AML1, at the very 5' end of EVI1→translocation protein includes N-term AML1 with the Runt domain and most of the gene EVI1, from the second untranslated exon to C-term, which includes the 2 zinc fingers.

**Oncogenesis**
Chimeric transcription factor with the dual functions of AML1 and EVI1: differentiation block (due to Runt) and stimulation of proliferation (from the zn fingers).

References


This article should be referenced as such: