t(3;7)(q26;q21)
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Identity

Partial karyotype showing an unbalanced t(3;7)(q26;q21).

Clinics and pathology

**Note:** This translocation has been observed in myeloid leukemia [one case of acute myeloid leukemia (AML), subtype M4, and two cases of chronic myeloid leukemia in blast crisis (CML-BC)].

**Disease**
Blast crisis chronic myelogenous leukemia (myeloid-myeloid/NK phenotype).

**Phenotype / cell stem origin**
Myeloid leukemia.

**Prognosis**
Poor.

**Disease**
AML M4.

**Phenotype / cell stem origin**
Acute myeloid leukemia.

**Prognosis**
Poor.

Cytogenetics

**Cytogenetics morphological**
t(3;7)(q26;q21) in BC-CML; -7, der(7)t(3;7)(q26;q21) in AML M4.

Probes
RP11-33A1 (EVI1) RP11-332M5 (CDK6).

Additional anomalies
Sole anomaly in AML; Ph chromosome in BC-CML patients.

Variants
No variants described.

Genes involved and Proteins

**EVI1 (ecotropic viral integration site 1) (alias PRDM3)**

**Location:** 3q26.2

**Note:** EVI1 is expressed at very low levels in normal peripheral blood and bone marrow. The gene is overexpressed in myeloid leukemias and myelodysplastic syndromes as a result of chromosomal rearrangements at either the 5' region of the gene in t(3;3)(q21;q26) or at the 3' region in inv(3)(q21q26) by juxtaposition of the gene to putative enhancer elements of the Ribophorin I gene in 3q21. High expression of EVI1 can also occur in the t(3;21)(q26;q22) as part of the fusion gene AML1 / MDS1/EVI1 in CML-BC, or MDS or as part of the fusion gene ETV6 /MDS1/EVI1 in AML with t(3;12) translocation. EVI1 is also involved in other translocations such as t(2;3)(p13;q26), t(2;3)(q23;q26), t(3;17)(q26;q22) and t(3;13)(q26q13-14). Other studies have reported abnormal expression of EVI1 in MDS and AML without 3q26 structural abnormalities, suggesting that inappropriate activation of this gene occurs through various mechanisms.

**DNA / RNA**
16 exons spanning 64.2 kb. Transcriptional orientation is from telomere to centromere. 6 splicing variants.

**Protein**
1051 amino acids; 118335 Da. Nuclear location, contains 10 C2H2-type zinc fingers.
FISH cohybridization between clones identifying breakpoints on chromosome 3 (RP11-33A1) and 7 (RP11-332M5) in a case of BC-CML with a t(3:7)(q21;q26).

**CDK6 (cyclin-dependent kinase 6) (alias PLSTIRE)**

**Location:** 7q21.2  
**DNA / RNA**  
7 exons spanning 229 kb. Transcriptional orientation is from telomere to centromere.  
**Protein**  
326 amino acids; 36938 Da. It belongs to the Ser/Thr protein kinase family, CDC2/CDKX subfamily. It is probably involved in the control of the cell cycle. Interacts with D-type G1 cyclins.

**Results of the chromosomal anomaly**

**Hybrid gene**  
**Note:** overexpression of EVI1 in bone marrow; no detected CDK6/EVI1 fusion gene in any of the myeloid leukemia cases analyzed.

**References**


*This article should be referenced as such:*  