Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Leukaemia Section

Mini Review

t(3;7)(q26;q21)

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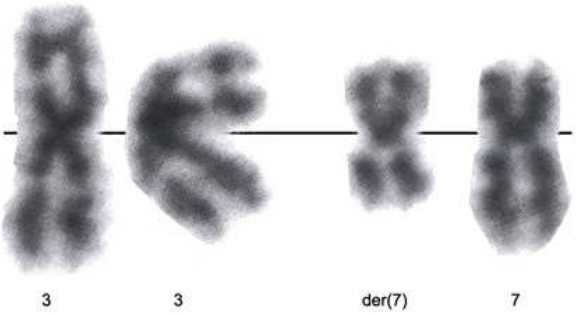
Identity



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

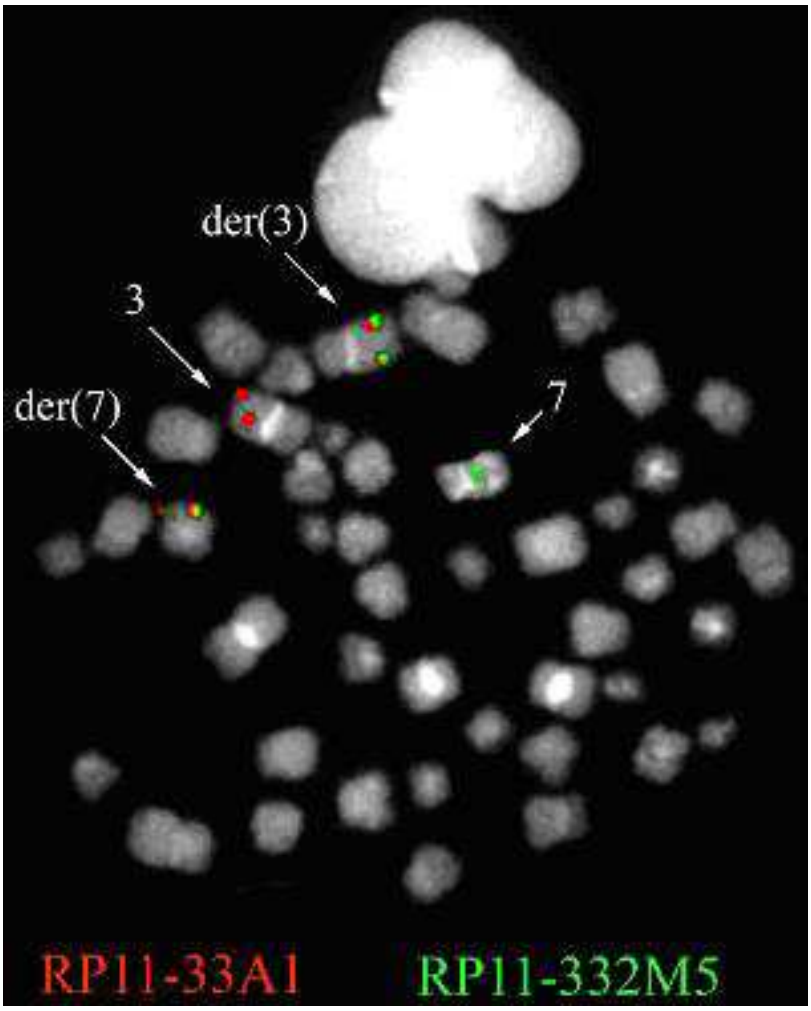
Note: EVI1 is expressed at very low levels in normal peripheral blood and bone marrow. The gene is overexpressed in myeloid leukemias and myelodysplastic syndromes as a result of chromosomal rearrangements at either the 5' region of the gene in t(3;3)(q21;q26) or at the 3' region in inv(3)(q21q26) by 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)(q26;q13-14). 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

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