t(9;21)(q34;q22)

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Clinics and pathology

Disease
Acute myeloid leukaemia (AML)

Epidemiology
Only one case to date.

Clinics
The patient was a 75 year old male patient with a myeloproliferative syndrome (MPS) in transformation to AML. The MPS was a 8p11 myeloproliferative syndrome (EMS).

Cytogenetics

Cytogenetics morphological
The karyotype also comprised a t(8;22)(p11;q11) with BCR/FGFR fusion, responsible for the EMS.

Genes involved and proteins

Note
RUNX1 was involved in the translocation. RUNX1, also called AML1 or CBFA2, is a transcription factor, critical regulator of hematopoietic-cell development, involved in many de novo and treatment related leukaemias. The exons 1-4 of RUNX1 were fused to repetitive sequences from chromosome 9, adding 70 amino acids to RUNX1 exon 4 encoding sequences, resulting in a truncated RUNX1. The t(9;21)/RUNX1 involvement may be responsible for the transformation of the EMS.

References

Fioretos T, Panagopoulos I, Lassen C, Swedin A, Billström R, Isaksson M, Strömbeck B, Olofsson T, Mitelman F, Johansson B. Fusion of the BCR and the fibroblast growth factor receptor-1 (FGFR1) genes as a result of t(8;22)(p11;q11) in a myeloproliferative disorder: the first fusion gene involving BCR but not ABL. Genes Chromosomes Cancer. 2001 Dec;32(4):302-10


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