

Leukaemia Section

Short Communication

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 myelo-proliferative 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

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