t(9;11)(q34;q23) AF9q34/MLL

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Identity

Note: Not to be confused with the t(9;11)(q34;q23), involving MLL and FBP17.

Clinics and pathology

Disease
De novo AML M5.

Epidemiology
Only one case to date, a 62-year-old male patient.

Clinics
The patient presented with multi-organ failure due to leukostasis and infection. WBC was 248 x 10^9/L.

Prognosis
Patient died within a few months after diagnosis.

Cytogenetics

Additional anomalies
+8 and +13 in 40% of metaphases.

Genes involved and Proteins

MLL
Location: 11q23
DNA / RNA
13-15 kb mRNA.

Protein
431 kDa; contains two DNA binding motifs (a AT hook, and Zinc fingers), a DNA methyl transferase motif, a bromodomain; transcriptional regulatory factor; nuclear localisation.

AF9q34
Location: 9q34, centromeric of FBP17 and ABL.
DNA / RNA
14 exons stretched over an area of about 84 kb, 5192 bp mRNA.

Protein
967 amino acids; contains a GAP related domain (GRD), an ‘FLR’-motif, a Pleckstrin homology (PH) domain and a calcium/phospho-lipid-binding C2/CALB domain.

Results of the chromosomal anomaly

Hybrid gene
Transcript
5' MLL - 3' AF9q34.

Fusion protein
Description
MLL/AF9q34 contains the AT-hook, DNA-Methyltransferase, Zinc-Fingers domains of MLL and the entire GAP related domain (GRD) and the C2/CALB domain of AF9q34.
References

This article should be referenced as such:
von Bergh ARM. t(9;11)(q34;q23) AF9q34/MLL. Atlas Genet Cytogenet Oncol Haematol. 2006;10(1):22-23.