Leukaemia Section
Mini Review

\[ t(9;11)(p22;q23) \]

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Identity

\[ \text{t}(9;11)(p22;q23) \text{ G-banding (left) - Courtesy Jean-Luc Lai and Alain Vanderhaegen; R-banding: center below: t}(9;11)+\text{der}(9)\text{t}(9;11) - \text{Courtesy Christiane Charrin; t}(9;22)(center above) and FISH (right) - Courtesy Pascale Cornillet-Lefebvre and Stéphanie Struski. The probe is } MLL; \text{ one signal is on the normal } 11, \text{ one signal on the der}(11), \text{ and one signal (arrow) on the der}(9). \]

Clinics and pathology

Disease

ANLL

Phenotype / cell stem origin

M5 most often (especially M5a), M4; de novo and; therapy related ANLL with antitopoisomerase II drugs (epipodophyllotoxins; anthracyclins, actinomycin D).

Epidemiology

2 to 5 % of ANLL; up to 25% of de novo M5a in children; all ages represented; sex ratio: 1M/1F.

Clinics

Organomegaly, frequent CNS involvement, especially in de novo cases; no preceding myelodysplastic phase, unlike classic therapy related ANLL with chromosome 5 and/or 7 involvement, short interval from initial drug therapy (may even be of 1-2 yrs).
Cytology
Absence of trilineage dysplasia, unlike classic therapy related ANLL.

Prognosis
CR in most de novo ANLL cases; the prognosis may not be as poor as in other 11q23 leukaemias, with a median survival around 4 yrs in de novo cases; very poor prognosis in secondary ANLL cases.

Cytogenetics

Cytogenetics, morphological
May easily be overlooked; better seen using R-banding.

Cytogenetics, molecular
FISH is indicated.

Additional anomalies
None in 70% of cases, +8 in 20%.

Variants
Complex 3 way translocations t(9;11;Var) involving a (variable) third chromosome have been described, and showed that der(11) is the crucial one.

Genes involved and Proteins

AF9
Location: 9p22
Protein
Contains a nuclear targeting sequence; transcriptional activator; nuclear localisation.

MLL
Location: 11q23
Protein
Contains two DNA binding motifs (a AT hook, and Zinc fingers), a DNA methyl transferase motif, a bromodomain; transcriptional regulatory factor; nuclear.

Results of the chromosomal anomaly

Hybrid gene
Description
5’ MLL - 3’ AF9; variable breakpoints.

Fusion protein
Description
N-term -- AT hook and DNA methyltransferase from MLL (1444 amino acids) fused to the 192 C-term amino acids from AF9 (as breakpoints are variable, this is only an exemple); 180 kDa.

Expression localisation
Nuclear localisation.

References

This article should be referenced as such: