

Leukaemia Section

Short Communication

t(3;5)(q25;q34)

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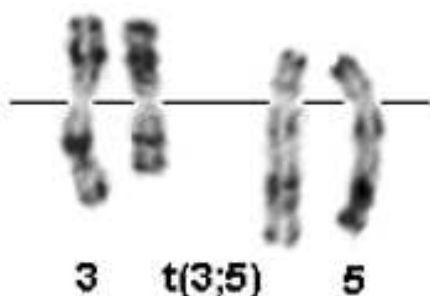
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Identity



t(3;5)(q25;q34) (R-banding) - Courtesy Jacques Boyer.

Clinics and pathology

Disease

myeloproliferative disorders (MPS), myelodysplastic syndromes (MDS), and acute non lymphocytic leukemia (ANLL).

Phenotype/cell stem origin

M2, M4, M6 ANLL; trilineage involvement.

Epidemiology

Median age: 35 yrs; balanced sex ratio.

Prognosis

CR: 8/12, but median survival is less than 1 yr.

Cytogenetics

Cytogenetics morphological

The breakpoints (3q25 and 5q34) have been controversial for a long time.

Additional anomalies

Most often none; +8.

Genes involved and proteins

MLF1

Location

3q25

Protein

31 KDa; widely expressed; cytoplasmic localisation; possible role in normal hematopoietic differentiation.

NPM1

Location

5q34

Protein

Nuclear localisation; binds to single and double strand nucleic acids; phosphoprotein that may transport ribonucleoproteins; may also have a role in DNA replication nuclear phosphoprotein; role in centrosome duplication and various relations with other proteins like p53, ARF, ..., (numerous interactions are under study).

Result of the chromosomal anomaly

Hybrid gene

Description

5' NPM-3' MLF1 on der(5).

Fusion protein

Description

54 kDa with the 175 N-term amino acids from NPM.

Expression / Localisation

Nucleus, mainly in the nucleolus.

References

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