t(3;5)(q25;q34)

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Published in Atlas Database: October 2004
Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0305.html
DOI: 10.4267/2042/38139
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Identity

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t(3;5)(q25;q34) \text{(R-banding)} - \text{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 interations 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


This article should be referenced as such: