Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Leukaemia Section

Mini Review

t(6;14)(p21;q32)

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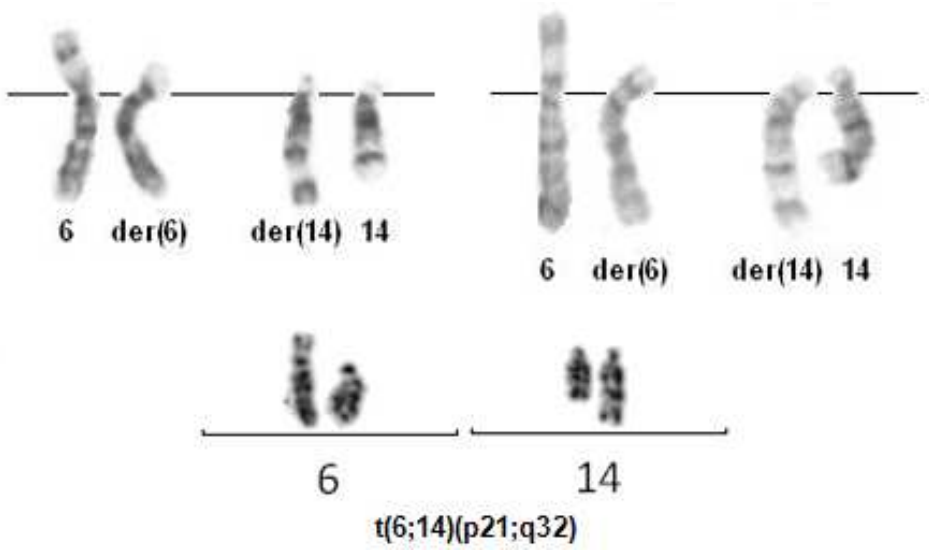

G-banding - Top: Courtesy Jean Luc Lai; Bottom: Courtesy Melanie Zenger and Claudia Haferlach.

Clinics and pathology

Disease

t(6;14)(p21.1;q32.3) is mainly multiple myeloma / plasma cell leukaemia, but also observed in diffuse large B cell non Hodgkin lymphoma (DLBCL) and marginal zone B cell lymphoma.

Phenotype/cell stem origin

Mature B-lymphocyte.

Epidemiology

Approximately 4% of primary MM cases. MGUS? Should be rather frequent in lymphoma but not yet systematically estimated.

Cytology

CD138+ dystrophic or not dystrophic plasma cells in MM; CD5+ diffuse large B cell lymphoma and splenic lymphoma with villous lymphocytes in NHL.

Prognosis

No prognosis value clearly established.

Cytogenetics

Cytogenetics morphological

May be not easy to detect.

Cytogenetics molecular

Translocation detected by FISH with CCND3 and IgH probes; der(14) sometimes in multiple copies; der(6) sometimes lost.

Additional anomalies

Complex karyotype in most cases.

Genes involved and proteins

CCND3 (cyclin D3)

Location
6p21.1

Note
Synonym CGD3

DNA/RNA
6,88kb, 5 exons, 5^1 - 3^1 telomeric orientation.

Protein
32,5kDA, 292 amino acids; regulates Rb1 phosphorylation and, hence, cell cycle G1/S transition, as a result of interaction with CDK4 and CDK6 protein kinases.

IGH

Location
14q32.3

Result of the chromosomal anomaly

Hybrid gene

Description
6p21.1 breakpoint centromeric to CCND3; 14q32.3 breakpoint in the switch region; leads to proximity between IGHS sequences and CCND3 on der(14).

Fusion protein

Note
No

Oncogenesis
Dysregulation and overexpression of CCND3.

To be noted

Case Report
A case of Chronic Lymphocytic Leukemia (CLL) with a rare chromosome abnormality: t(1;14;6)(q21;q32;p21), a variant of t(6;14)(p21;q32).
A case of Chronic Lymphocytic Leukemia (CLL) with a rare chromosome abnormality: t(1;14;6)(q21;q32;p21), a variant of t(6;14)(p21;q32).

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