

# Leukaemia Section

## Short Communication

### del(13q) in multiple myeloma

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Published in Atlas Database: March 2001

Online updated version : <http://AtlasGeneticsOncology.org/Anomalies/del13qMMyeloid2094.html>

DOI: 10.4267/2042/37733

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#### Identity



del(13q) G- banding - Courtesy Melanie Zenger and Claudia Haferlach.

#### Clinics and pathology

##### Disease

Multiple myeloma (MM) is a monoclonal B-cell malignancy, which originates theoretically in lymph node germinal centers but locates and expands in bone marrow. It represents 10% of all the hematopoietic cancers, with a great variability in clinical presentation, response to therapy and survival duration. In more than 1/3 of cases, MM can be preceded by a phase of monoclonal gammopathy of uncertain significance (MGUS). At the extreme it can evolve in plasma blast acute leukemia.

##### Phenotype/cell stem origin

Malignant myeloma cells are long-lived cells with morphological features varying from normal to dystrophic considering size of the cells, presence of nucleolar structures and aspect of the chromatin. Immunophenotype includes inconstant expression of CD56, CD38, CD40 and CD138.

##### Epidemiology

del(13q) is detected in 15-20% of MM patients by conventional karyotype and in 33-52% of cases by FISH analysis.

##### Prognosis

-13/del(13q) appears as one of the main prognostic factors with  $\gamma$ 2-microglobulin serum level and the percentage of bone marrow plasma cells. Patients with del(13q) have a significantly lower event-free survival, overall survival and complete remission duration, either in standard-dose or in high dose therapy protocols.

#### Cytogenetics

##### Cytogenetics morphological

del(13q) is a frequent occurrence in chronic lymphoproliferative diseases and in non Hodgkin lymphoma.

del(13q) in MM is rarely observed as a sole anomaly; detected both in hyperdiploid and hypodiploid karyotypes, but with a higher incidence in hypodiploid forms; consequently, according to some authors, the prognostic value of del(13q) should have to be related to the ploidy.

It is considered as a secondary event, however occurring early in the evolution of MM because it is observed in patients with MGUS.

The minimal common region of deletion is in band 13q14.3, the same as in chronic lymphocytic leukemia. Del(13q) is clearly underscored by karyotyping because a number of deletions are submicroscopic or only detected in interphase nuclei. It involves rb-1, and loci D13S319 and D13S272 which are approximately 100kb distal from rb-1.

rb-1 deletion / mutation would be a key event in MM evolution; however other gene(s) would be involved at

13q14.3 because rb-1 and D13S319 deletions are dissociated in some cases.

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*This article should be referenced as such:*

Viguié F. del(13q) in multiple myeloma. *Atlas Genet Cytogenet Oncol Haematol.* 2001; 5(2):123-124.

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