Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

Leukaemia Section

Short Communication

del(20q) in myeloid malignancies

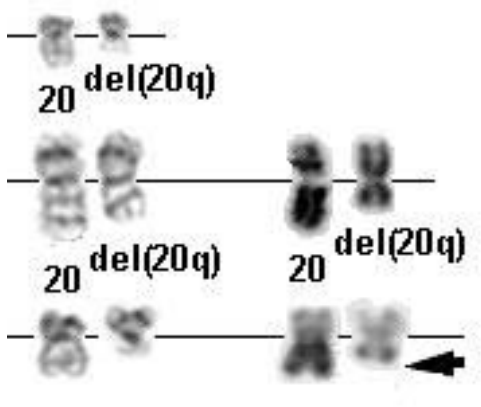
Chrystèle Bilhou-Nabera

Laboratoire d'Hématologie, Hôpital du Haut-Lévêque, CHU de Bordeaux, Ave de Magellan, 33 604 Pessac, France (CBN)

Published in Atlas Database: December 2000

Online updated version: http://AtlasGeneticsOncology.org/Anomalies/del20qID1040.html
DOI: 10.4267/2042/37701

Identity



del(20q) G- banding (left) - Courtesy Diane H. Norback, Eric B. Johnson, Sara Morrison-Delap Cytogenetics at theWaisman Center; R-banding (right) - top: Courtesy Jean-Luc Lai; bottom: Editor

Clinics and pathology

Disease

A very large spectrum of hematological malignancies as myelodysplastic syndromes (MDS), acute non lymphocytic leukemias (ANLL), polycythemia vera, chronic neutrophilic leukemia.

Phenotype/cell stem origin

As described in various types of hematological disorders, 20q- appears as a primary karyotypic abnormality occurring in a pluripotential hematopoietic stem cell; the pathogenic mechanism by which 20q- alters the hematopoietic stem cells in hematological disorders remains unknown; 20q- may confer a proliferative advantage to myeloid cells through deletion of a tumor suppressor gene.

Epidemiology

An interstitial or terminal deletion of the long arm of chromosome 20 (20q-) has been described as the second most frequent sole clonal structural abnormality (5 %) behind t(9.22).

Prognosis

In MDS, 20q- alone is associated with a good prognosis regarding survival and potential for AML evolution, as defined by the International Prognostic Scoring System (IPSS) for MDS prognosis.
In de novo acute leukemia, a poor response to treatment and a reduced survival is observed.
In myeloproliferative disorders, the presence of 20q does not appear to adversely affect survival.

Cytogenetics

Cytogenetics morphological

The breakpoint on chromosome 20 is not constant; 20q- is frequently associated with other cytogenetic abnormalities as del(5q), trisomy 8, trisomy 21, deletions or translocations involving the long arm of chromosome 13; a newly described translocation t(11;20)(p15;q11) resulting in a NUP98- TOP1 fusion gene was described in therapy-related myelodysplastic syndrome (RAEB); t(11;20)(p15;q11) is a rare recurrent translocation reported in patients with MDS, ANLL and polycythemia vera.

Cytogenetics molecular

A small fragment (around 8 Mb), proximally flanked by D20S206 and distally by D20S119 and UT 654 was identified using FISH.

Additional anomalies

del(5q), trisomy 8, deletions or translocations involving 13q and trisomy 21.

Genes involved and proteins

Note

Genes remaining within this deleted region are topoisomerase 1 (TPO1-OMIN 126420), phospholipase C (PLC1), hepatocyte factor nuclear 4 (HNF4) and adenosine desaminase (ADA); recently, a new gene KRML transcriptional regulator was mapped in the smallest commonly deleted region in malignant myeloid leukemias.

References

Mitelman F, Kaneko Y, Trent J. Report of the committee on chromosome changes in neoplasia. Cytogenetic Cell Genet 1990; 55(1-4):358-86.

Campbell LJ, Garson OM. The prognostic significance of deletion of the long arm of chromosome 20 in myeloid disorders. Leukemia. 1994 Jan;8(1):67-71

Kurtin PJ, Dewald GW, Shields DJ, Hanson CA. Hematologic disorders associated with deletions of chromosome 20q: a clinicopathologic study of 107 patients. Am J Clin Pathol. 1996 Nov;106(5):680-8

Greenberg P, Cox C, LeBeau MM, Fenaux P, Morel P, Sanz G, Sanz M, Vallespi T, Hamblin T, Oscier D, Ohyashiki K, Toyama K, Aul C, Mufti G, Bennett J. International scoring system for evaluating prognosis in myelodysplastic syndromes. Blood. 1997 Mar 15;89(6):2079-88

Wang PW, Iannantuoni K, Davis EM, Espinosa R 3rd, Stoffel M, Le Beau MM. Refinement of the commonly deleted segment in myeloid leukemias with a del(20q). Genes Chromosomes Cancer. 1998 Feb;21(2):75-81

Ahuja HG, Felix CA, Aplan PD. The t(11;20)(p15;q11) chromosomal translocation associated with therapy-related myelodysplastic syndrome results in an NUP98-TOP1 fusion. Blood. 1999 Nov 1;94(9):3258-61

Wang PW, Eisenbart JD, Cordes SP, Barsh GS, Stoffel M, Le Beau MM. Human KRML (MAFB): cDNA cloning, genomic structure, and evaluation as a candidate tumor suppressor gene in myeloid leukemias. Genomics. 1999 Aug 1;59(3):275-81

This article should be referenced as such:

Bilhou-Nabera C. del(20q) in myeloid malignancies. Atlas Genet Cytogenet Oncol Haematol. 2001; 5(1):33-34.