

Leukaemia Section

Short Communication

dic(17;20)(p11.2;q11.2)

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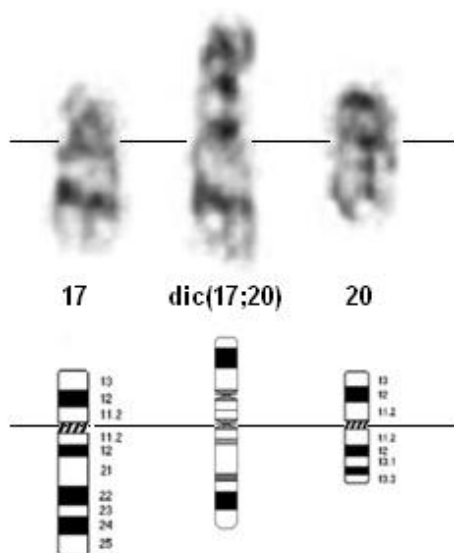
Published in Atlas Database: September 2008

Online updated version : <http://AtlasGeneticsOncology.org/Anomalies/dic1720p11q11ID1485.html>

DOI: 10.4267/2042/44563

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Identity



Dicentric (17;20)(p11.2;q11.2) partial karyotype and ideogram.

Clinics and pathology

Disease

De novo acute myeloid leukemia (AML) and myelodysplastic syndrome (MDS); treatment-related AML (t-AML) and MDS (t-MDS).

Phenotype/cell stem origin

8 cases reported: 4 de novo AML cases (including one AML-M2 and one erythroleukemia), 3 de novo MDS cases (including one refractory anemia), one t-MDS, and one t-MDS in transformation to AMMoL.

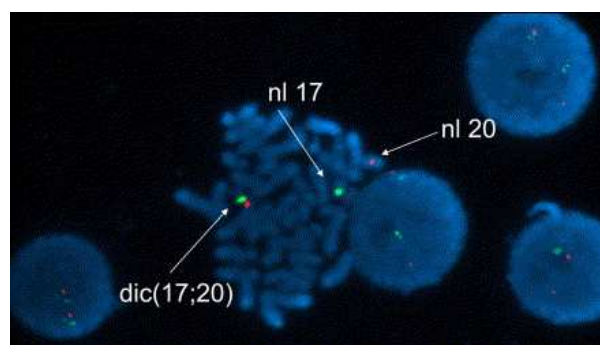
Epidemiology

Epidemiology of the 8 patients reported to date, 7 were male and one was female, aged 47 to 87 years.

Prognosis

Poor; majority of patients died between 2 and 8 months post diagnosis.

Cytogenetics



FISH image of a metaphase showing a normal copy of chromosomes 17 and 20 as well as the dic(17;20). The metaphase appears stained in blue (DAPI counterstain). Red signal, chromosome 20 centromere; green signal: chromosome 17 centromere. The dic(17;20) shows both centromeres.

Additional anomalies

Sole anomaly in one case; remaining cases with additional abnormalities; association with -5/del(5q), -7/del(7q), and/or +8 is frequent.

Genes involved and proteins

Note

dic(17;20) leads to loss of 17p (TP53 gene). Because of

this, patients with this abnormality may have a prognostic outcome similar to the patients with "17p-syndrome". Dicentric (17;20) also leads to loss of 20q [various genes involved: topoisomerase 1 (TOP1), phospholipase C (PLC1), hepatocyte factor nuclear 4 (HNF4), adenosine deaminase (ADA); KRML transcriptional regulator].

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This article should be referenced as such:

Meloni-Ehrig AM. dic(17;20)(p11.2;q11.2). *Atlas Genet Cytogenet Oncol Haematol.* 2009; 13(9):668-669.
