Leukaemia Section
Mini Review

t(2;3)(p15-23;q26-27)
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Identity

Note: There are 2 subtypes of the t(2p;3q); in one type the breakpoint on chromosome 2 is assigned to bands 2p21-23, whereas the breakpoint for the other type of breakpoint is localized at 2p15-21.

Clinics and pathology

Disease

Myeloid malignancies: myelodysplastic syndrome (MDS) in 1/4 of cases, with various FAB diagnoses (refractory anaemia (RA), RA with ringed sideroblasts (RARS), RA with excess of blasts (RAEB), RAEB in transformation (RAEB-t), and chronic myelomonocytic leukemia (CMML)), acute non lymphocytic leukaemia (ANLL) in 60% of cases (M2-ANLL in 1/4 of all cases), blast crisis of a chronic myelogenous leukaemia (BC-CML) in about 10% of cases.

Partial GTG (Marian Stevens-Kroef, left) and RFA (Anne Hagemeijer, right) banded karyotypes of t(2;3)(p15-23;q26-27) with the distal (A) and proximal (B) breakpoint on chromosome 2.
Etiology
1/4 of cases were therapy related leukemias, and 10% were BC-CML cases.

Epidemiology
At least 50 cases described; sex ratio: 1.33 M/F; median age around 50 years, most patients being between 30 and 70 year old (range 3-80 years).

Cytology
High platelet count, dysmegakaryopoiesis, and multilineage dysplasia in 80 to 90% of cases.

Prognosis
Median survival 12 months (range 1-53 months), with a few patients surviving with bone marrow transplantation.

Cytogenetics
Note: Heterogeneous breakpoints by cytogenetic and FISH analysis; FISH mapping of 2p breakpoints was very heterogeneous ranging from p14 or p15 to p23; FISH mapping of the 3q breakpoint was within the EVII-MDS region (between RP11-694D5 (centromeric) and RP11-362K14 (telomeric) in the great majority of cases.

Additional anomalies
Sole anomaly in 40%, associated with -7 in 30%, with del(5q) in 15%, with del(7q) in 10%, with t(9;22)(q34;q11) in 10%, and with a complex karyotype in 20% of cases.

Genes involved and Proteins
Note: Molecular analysis has been performed in only a very few cases. In most of these, ectopic expression of EVII was demonstrated, but rare cases seem not to involve this gene. Therefore, characteristics of EVII involvement (high platelet count, multilineage dysplasia, monosomy 7, prior history of carcinogen exposure and a poor prognosis) may not be present in further cases with apparently the same breakpoints. The gene(s) involved in chromosome 2 is/are unknown.

EVII
Location: 3q26
Note: There is a direct correlation between mapping of the 3q breakpoint in the above given EVII-MDS region and EVII ectopic expression by RT-PCR. Rare case with 3q break outside this interval failed to show ectopic expression of EVII.

DNA / RNA
EVII contains 12 exons.

Protein
EVII may play an important role in organogenesis, cell migration, cell growth, and differentiation.

References
Yunis JJ. Recurrent chromosomal defects are found in most patients with acute nonlymphocytic leukemia. Cancer Genet Cytogenet 1984;1:125-137.


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