

**Leukaemia Section**

Short Communication


**t(8;12)(q12;p13)**

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**Clinics and pathology**

**Disease**
Myelodysplastic syndrome (MDS) and acute myeloid leukemia (AML).

**Phenotype/cell stem origin**
Refractory anemia (RA), refractory anemia with an excess of blasts (RAEB) and AML, FAB subtypes M2. A primitive myeloid progenitor is likely to be involved.

**Epidemiology**
Only three cases reported to date: one case with RA, one with RAEB and the other with AML M2. All the patients were adults (more than 65-year old); sex ratio: 2M/1F.

**Clinics**
Splenomegaly, anemia, thrombocytopenia and leukopenia.

**Cytology**
Positive for CD34, CD33, CD13, CD15, HLA-DR.

**Prognosis**
The three patients died. The survival in AML was 4 weeks, in RA 16 months and in RAEB 5 years.

**Genetics**
Deletion of ETV6 and CDKN1B sequences were observed in the case of RA.

**Cytogenetics**
Balanced translocation in the three cases.

**Cytogenetics molecular**
Metaphase FISH analysis performed on the RA patient, using specific probes (cosmid and YACs) for the 12p region, revealed hemizygous loss of the ETV6 and CDKN1B regions. On the other hand, FISH studies on the RAEB patient showed that the breakpoints in 12p13 mapped immediately distal to the breakpoint cluster region frequently involved in hematological neoplasms.

**Additional anomalies**
The t(8;12)(q12;p13) was observed as a sole anomaly in the RAEB case, and accompanied by an extra der(8) and del(5)(q13q31) in the AML and RA cases respectively.

**Genes involved and proteins**

**Note**
Not yet defined.

**References**

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