ABSTRACT

Accurate localization of breakpoints and deleted regions on chromosome 7 in bone marrow cells of patients is an essential step in identifying genes involved in tumor transformation of a cell. In case of hematological malignancies usually oncogenes and tumor suppressor genes are activated or deleted by a change in the arrangement of genetic material. Aberration of chromosome 7, total or partial loss of chromosome, especially long arms 7q, are among the recurrent cytogenetic abnormalities in patients with myeloid diseases such as myelodysplastic syndromes (MDS) and acute myeloid leukemia (AML). Aberrations of chromosome 7 are an important prognostic marker occurring in 8-10% de novo MDS and AML and in 40-50% treated MDS / AML.

For a detailed analysis of chromosome 7 breakpoints and aberrations, samples of 51 adult patients diagnosed with AML / MDS were examined using conventional and molecular cytogenomic methods. In our testing group we demonstrated a separate 7q deletion in one patient (2%) and an isolated monosomy of chromosome 7 in six patients (12%). Aberration of chromosome 7 detected in combination with another change was found in 17 cases (33%) and 27 patients (53%) had complex karyotype changes including chromosome 7. The most frequent breakpoint was 7q22. In 26 patients we proved a deletion 7q, of which 25 patients had deleted the *EZH2* gene region (7q36.1). In 22 out of 26 patients we showed deletions of all regions examined by us - those being 7q22, 7q31 and 7q36. In all these cases, we confirmed the deletion of the tumor suppressor genes *KMT2E* and *EZH2*. Changes on the short arms were the most often located in the 7p11-7p12 area. The *IKZF1* gene, which was deleted in 11 patients, is located in this area. The incidence of both long arms deletions (7q) and short arms (7p) were detected in ten patients. Analysis of the prognostic effect of chromosome 7 alteration on overall survival did not reveal significant differences ($p = 0.815$).

Key words: Myelodysplastic syndromes, acute myeloid leukemia, chromosome 7 rearrangements, 7q deletion, complex rearrangements, FISH