Abstract

Chromosome 11 abnormalities are found in many hematological malignancies. In acute myeloid leukemia (AML), a proto-oncogene *MLL* (11q23.3) is frequently altered. However, rearrangements to other regions of chromosome 11 have been reported. Therefore, we have identified and characterized the chromosome 11 breakpoints and common deleted and amplified areas in the bone marrow or peripheral blood cells of newly diagnosed patients with AML.

Many recurrent and random chromosome 11 breakpoints were identified (recurrent in bands 11p15.4 (in *NUP98* gene), 11q23.3 (in the *MLL* gene), 11p13, 11p12 and 11q13.2) and deleted or duplicated/amplified regions were determined. We notified new possibly significant genes in the development of AML. Contrary to the *MLL* rearrangements, patients with other chromosome 11 changes were older, with complex karyotype, unbalanced aberrations and short survival. FISH screening was proved very helpful in case of deviding cells lack and cryptic *MLL* gene rearrangement.

In conclusion, molecular analyses of chromosomal breakpoints and amplified or deleted areas are very important not only for the patient stratification into specific prognostic and clinical subgroups but also for the identification of genes involved in tumour pathogenesis. Further investigation of the affected genes and their protein products will improve our understanding of the oncogenesis of AML and could be clinically applied for the designation of more effective therapeutic approach.

Key words: acute myeloid leukemia (AML), *MLL*, complex karyotype, FISH mapping.