Abstract

Hematological neoplasias are characterized by a wide spectrum of genetic alterations. We analyzed 15 specimens from patients with various types of hematological malignancies by means of the FISH technique in order to detect aneuploidy of chromosome 17 and deletion of TP53 gene. In 11 of them chromosomal analyses were also carried out using conventional cytogenetic techniques; in 6 of these 11 specimens (54.5%) abnormal karyotypes were detected, namely: 3 translocations and 3 mosaicisms. FISH results revealed that in 26.7% of the 15 specimens there was chromosome 17 aneuploidy, and that 33.3% had TP53 deletion. Out of the 6 cases with abnormal karyotypes, further alterations were detected in two by FISH. In 5 cases chromosomal abnormalities were detected by FISH but not by the conventional cytogenetic procedures. Only in 3 (20%) out of the 15 specimens the results of chromosomal analyses were normal by both the conventional cytogenetics and FISH. These results corroborate the low frequency of chromosome 17 aneuploidy and of TP53 gene deletion in hematological neoplasias. However, the prognostic value of these genetic alterations is still not well defined.

Keywords
Aneuploidy, Deletion, FISH, Genetic instability, Hematological neoplasias, TP53 gene.