Abstract

Complex chromosomal aberrations occurs are described in approximately 20 % of patients with myelodysplastic syndrome (MDS) and are associated with a poor prognosis. Nevertheless, the mechanism and possible causes responsible for the emergence of these aberrations are not fully understood. There are two models describing the emergence of these aberrations, namely shattering of single chromosomes or their parts during the so-called cellular crisis (chromothripsis) and/or progressive accumulation of chromosomal aberrations during the course of the disease (clonal evolution). Using combination of cytogenomic methods we examined 61 samples of bone marrow from adult patients with MDS and a complex karyotype. Unbalanced aberrations with loss of genetical material were found in most cases. Chromosomes 5, 7 and 12 were most frequently involved in rearrangements. Clonal development, chromothripsis and both mechanism was detected in 26, 12 and 14 patients, respectively. Patients with deleted chromosome 5 included in complex karyotype had the shortest overall survival. The cause of emergence of complex aberrations did not affect survival. Cytogenomic analysis of complex aberrations allows detection of balanced and unbalanced changes and identification of important processes of tumorigenesis such as clonal evolution and chromothripsis. Thus, the study of chromosomal aberration and identification of mechanism that play significant role in the course of the disease contributes to the correct diagnosis and prognosis of patients and might contribute to the development of new therapeutic interventions.