Abstract

Myelodysplastic syndrome (MDS) is a clonal disease of hematopoesis resulting from damage to hematopoietic stem cells. The most common chromosomal aberration in patients with MDS is deletion of the long arms of chromosome 5, del(5q). The aim of this study is to analyse unbalanced aberrations of chromosome 5 in MDS patients, to compare the extent of 5q deletion in groups of patients with isolated del(5q) and with del(5q) in complex karyotypes, and to study the effect of the extent of del(5q) on overall survival and prognosis of the disease. We combined cytogenomic methods to examine 88 bone marrow samples from patients with MDS and del(5q) confirmed by conventional banding methods. Del(5q) was present in the karyotype as an isolated aberration in 31 patients (35,2 %), in combination with one other clonal aberration in 9 patients (10,2 %), and as part of complex karyotypes in 48 patients (54,6 %). Patients with complex karyotypes had a lower overall survival than patients with isolated del(5q). The occurrence of complex karyotypes was associated with a large extent of 5q deletion. When both the occurrence of complex karyotypes and the extent of 5q deletion were considered, only karyotype complexity had a significant effect on patients' overall survival. The extent of the deletion does not affect patients' survival. We cannot exclude the possibility that isolated large 5q deletions put patients at increased risk of clonal evolution and complex karyotypes.