Abstract

Leukemias are the most common cancer diseases in childhood. The majority of cases represent acute leukemias, the most common of which is acute lymphoblastic leukemia (ALL), the second most frequent subtype is acute myeloid leukemia (AML). One of the basic laboratory examinations at the time of diagnosis is the cytogenetic analysis of the karyotype of leukemic cells in which we are looking for the recurrent chromosomal aberrations. These changes in the structure or number of the chromosomes can be found in up to 90 % of patients and the exact prognostic significance is known for most of them. In ALL, the findings of high hyperdiploidy (>50 chromosomes) and translocation t(12;21)(p13;q22) are considered the most significant prognostic factor associated with good prognosis and translocations involving the *KMT2A* gene in the 11q23 region are associated with poor prognosis. In AML, the most frequent aberrations are t(8;21)(q22;q22), t(15;17)(q24;q21) and inv(16)(p13;q22) which are considered indicator of good prognosis. An important unfavourable prognostic finding in AML are the *KMT2A* gene rearrangements, the most common of which is the translocation t(9;11)(q23;p13.1). Nowadays, there are many ways to detect chromosomal aberrations in leukemic cells. G-banding is the most common method of classical cytogenetics, which has the great advantage because it allows simultaneous observation of all chromosomes and chromosomal aberrations present in one cell. Molecular cytogenetic methods (FISH, mFISH, mBAND, aCGH/SNP, etc.) allow detection of specific genetic aberrations, precise identification of chromosomal breakpoints and/or determination of the origin of marker chromosomes, and therefore significantly complement the results of conventional cytogenetic analysis.