

Acute lymphoblastic leukemia is the most common type of cancer in children. It is a very heterogeneous disease in which many recurrent chromosomal abnormalities have been described. The most important chromosomal abnormalities associated with a good prognosis are t(12;21)(p13;q22) which result in ETV6/RUNX1 fusion and hyperdiploidy. On the contrary findings suggesting a poor prognosis are t(9;22)(q34;q11) leading to fusion gene BCR/ABL1, MLL rearrangements or hypodiploidy.

Heteroploidy is one of the most frequent findings in childhood ALL. It is characterised by nonrandom gain or loss of chromosomes from diploid cells. One of the most important findings in childhood ALL is hyperdiploidy where a non-random gain of chromosomes is present. Hyperdiploidy has a favorable prognosis and the impact of additional structural aberrations requires further research. Another prognostically important group of heteroploidy is hypodiploidy. It is a quite rare finding and has a very poor outcome. There are non-random acquired chromosome losses observed in hypodiploid cells. Hypodiploid cell line may be masked with a doubled hyperdiploid clone which makes it difficult to identify.

Proper and early cytogenetical analysis of heteroploid cells is very important as it contributes assigning correct diagnosis and risk stratification, making it easier to choose adequate therapy.