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GENOMIC CHARACTERIZATION OF ACUTE LYMPHOBLASTIC LEUKEMIA IN CHILDREN. THE RESULTS OF GENETIC DIAGNOSTICS IN UKRAINE DURING 2021 – 2025

Background. Acute lymphoblastic leukemia (ALL) is the most common pediatric cancer. The latest WHO classification of ALL includes more than 13 subtypes defined by different genetic alterations (GA), each of which has its own prognostic characteristics. Our study **aimed** to decipher the genomic characteristics of patients with ALL. **Methods.** Bone marrow (BM) samples were collected from **1212** pediatric patients with ALL during last 5 years. The gender distribution was as follows: 56.20% boys and 43.80% girls. Samples of nucleic acids (DNA and RNA) were isolated from BM. Accurate detection of GA was performed using qPCR and MLPA. Cytogenetic analysis was performed on BM cells using GTG banding, FISH, and a DNA probe panel, following ISCN 2020 guidelines. **Results.** In our group we detected recurrent molecular abnormalities in 361 cases (29.78%). Prognostically favorable GA such as ETV6::RUNX1 and TCF3::PBX have been identified with frequency 57.34% (207 cases) and 9.97% (36 cases), respectively. Patients who had prognostically unfavorable GA such as BCR::ABL and KMT2A-rearrangement were found in 35 (9.7%) and 43 (11.91%) cases respectively. Other GA (del IKZF1, IGH::MYC, PML::RARα) were detected in 10 cases (2.77%). IKZF1plus a complex biomarker associated with a poor prognosis was detected in 30 cases (8.31%). Karyotyping was performed in 427 patients. Patients were classified according to the presence of the following chromosomal abnormalities (CA): normal karyotype (25.76%), hyperdiploidy (>50 chromosomes), encompassing almost 27% cases and associated with good survival and favorable outcome. Controversially, hypodiploidy (<44 chromosomes) that was observed in 5 cases (1.17%) has been linked to unfavorable prognosis. Complex karyotypes which are rare events in children ALL and typically involve from three to five CA were found in 17 cases (3.98%) and are also associated with an adverse outcome. The remaining 180 cases (42.15%) had other CA. **Conclusion.** Our findings demonstrate that Ukrainian pediatric patients with ALL have a high prevalence of GA associated with poor outcomes.

Keywords: acute leukemia, genetic alterations, karyotyping, prognosis.

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