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SPECTRUM OF RARE AND COMBINED BCR::ABL1 TRANSCRIPT VARIANTS IN ACUTE MYELOID LEUKEMIA: RESULTS OF MOLECULAR GENETIC SCREENING

Background. According to the WHO 5th Edition and ICC, BCR::ABL1 AML is a distinct high-risk entity characterized by an aggressive clinical course. Diagnosis requires $\geq 20\%$ myeloid blasts and the strict exclusion of CML in blast crisis. This differentiation relies on a thorough clinical and anamnestic evaluation to rule out prior splenomegaly or a myeloproliferative history, thereby confirming the de novo status of the leukemia. Aim: To determine the prevalence and molecular spectrum of BCR::ABL1 transcripts in Ukrainian AML patients during the 2021–2025 period. **Materials and Methods.** Bone marrow samples from 223 patients (125 children and 98 adults) with suspected de novo AML were analyzed at the National Children's Specialized Hospital "Ohmatdyt." Primary screening for genetic aberrations was performed using targeted next-generation sequencing (NGS) with the Oncomine Myeloid Assay on the Ion Torrent platform. This panel enables the simultaneous detection of DNA mutations and RNA fusion transcripts across multiple myeloid-associated genes. Positive results were verified via real-time PCR. **Results.** The overall frequency of BCR::ABL1-positive AML was 2.7% (6/223). A significant difference in prevalence was established depending on the age group: the frequency was 5.1% (5/98) in adult patients, while it was 0.8% (1/125) in the pediatric group. Analysis of the isoform structure showed that the e14a2 transcript was most characteristic in adults, identified in 60% of positive cases. The e1a2 transcript was encountered in both age groups. Of particular scientific interest was a unique case of co-expression of rare e18a2/e19a2 variants in an adult patient – an extremely rare event ($<1\%$ globally) indicating a complex clonal architecture within the tumor. **Conclusions.** Our findings confirm the low frequency of BCR::ABL1 in AML but underscore its critical role in risk stratification. The detection of rare variants (e1a2 and e18a2/e19a2 transcripts) justifies incorporating NGS-based screening into primary diagnostics. While these molecular findings provide a rationale for using tyrosine kinase inhibitors (TKIs), this study only partially confirms the clinical necessity of such therapy, as treatment efficacy was not directly analyzed. However, given international evidence on the benefits of TKIs in high-risk groups, comprehensive molecular monitoring remains a cornerstone of personalized precision medicine.

Keywords: acute myeloid leukemia, fusion, transcripts.

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