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GENOMIC INSTABILITY IN AML: ASSOCIATION BETWEEN FLT3-ITD AND CHROMOSOMAL TRANSLOCATION

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Introduction: Myelodysplastic Neoplasms (MDS) and Acute Myeloid Leukemia (AML) represent spectra of a common clonal hematologic process, whose progression is often associated with genomic instability and the acquisition of somatic mutations in genes that regulate hematopoiesis. Among these alterations, the FLT3-ITD mutation stands out as a significant molecular marker of poor prognosis, whose early detection may aid in risk stratification and monitoring of clonal evolution. **Objectives:** This study aimed to investigate the presence of the FLT3-ITD mutation in patients with MDS and AML, with emphasis on karyotype analysis and mutation confirmation via Sanger sequencing. **Material and methods:** A total of 45 bone marrow samples were analyzed from patients diagnosed with MDS (subtypes EB1 and EB2) and AML (de novo and secondary), collected in EDTA tubes. Genomic DNA was extracted using the PureLink® kit (Invitrogen) and subjected to amplification of the ITD region of the FLT3 gene by end-point PCR using primers described in the literature. PCR products were visualized by 2% agarose gel electrophoresis, with preliminary detection of the mutation based on band patterns suggestive of insertions. Samples with a suspected ITD pattern were purified and subjected to Sanger sequencing, with results analyzed via electropherograms and alignment to the reference sequence from NCBI using BLAST. In parallel, conventional cytogenetic analysis was performed on bone marrow using G-banding, with at least 20 metaphases evaluated per sample. **Results:** Among the 45 patients analyzed, a 33-year-old female patient with de novo AML presented with an abnormal karyotype, showing a chromosomal translocation between chromosomes 2 and 11 [t(2;11)], a non-recurrent alteration associated with genomic instability. The FLT3-ITD mutation was initially suggested by gel electrophoresis, with a band pattern indicative of a mutated fragment. Sanger sequencing confirmed the mutation, revealing a 27-base pair insertion in the region between exons 14 and 15 of the FLT3 gene, within the juxtamembrane domain, with 94% identity to the BLAST reference sequence. **Discussion and conclusion:** The simultaneous presence of a structural chromosomal abnormality and an activating FLT3 mutation reinforces the role of genomic instability in the progression of myeloid neoplasms. Moreover, the study demonstrated the effectiveness of Sanger sequencing as the gold standard method for confirming ITD mutations, particularly in scenarios where the size and exact location of the duplication directly impact clinical interpretation. The association between altered karyotype and point mutations highlights the importance of integrating classical cytogenetics and molecular analysis in the diagnosis and prognosis of hematologic malignancies.

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GEOSPATIAL ANALYSIS OF GENE FUSIONS IN PEDIATRIC ALL AND THEIR POSSIBLE ASSOCIATION WITH ENVIRONMENTAL FACTORS IN THE AMAZON REGION (PARÁ, 2023–2025)

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Introduction: Acute Lymphoblastic Leukemia (ALL) is the most common childhood cancer, accounting for 25% to 30% of pediatric neoplasms. The disease is characterized by chromosomal translocations and aneuploidies, genetic alterations that determine prognosis and treatment response. In the state of Pará, an estimated 360 new cases of leukemia are diagnosed annually, an incidence that may be influenced by regional genetic and environmental factors. **Objectives:** To investigate the potential influence of regional environmental factors on the distribution of gene fusions in patients diagnosed with ALL in the state of Pará between 2023 and 2025, considering the geographic origin of each case. **Material and methods:** This study (Ethics Committee approval No. 4.040.805) analyzed patients with ALL diagnosed at the Octávio Lobo Children's Cancer Hospital (HOIOL) between 2023 and 2025. From blood samples, RNA was extracted, converted to cDNA, and used to detect five specific gene fusions (*BCR::ABL*, *TCF3::PBX1*, *KMT2A::AFF1*, *ETV6::RUNX1*, and *SIL::*