extramedullary involvement, a particular blast morphology, and a worse outcome as compared to fusion-negative AML pediatric patients, including a high frequency of disease refractoriness and relapse. NGS studies did not show CBFA2T3-GLIS2 in our patient. Thus, our results indicate that RAM+ associated with the driver p210-BCR-ABL1, even with specific targeted therapy, may lead to treatment resistance regardless of CBFA2T3-GLIS2+. **Conclusion:** Here we highlight for the first time the presence of p210-BCR-ABL1 associated with RAM phenotype in the absence of CBFA2T3-GLIS2 and disease refractoriness in a non-DS-AMKL infant.

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MOLECULAR CYTOGENETIC AND NEXT-GENERATION SEQUENCING STUDIES OF AN ADOLESCENT WITH ACUTE MYELOID LEUKEMIA AND INV(16) ASSOCIATED WITH A KIT PATHOGENIC MUTATION

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Objectives: Inv(16)(p13q22)/CBFb-MYH11 in Acute Myeloid Leukemia (AML) is hard to characterize and comprises about 5%–7% of all cases and 5%–9% in children. Inv(16) is associated with a good prognosis both in adults and children. However, approximately 30%–40% of affected patients experience relapse. In this context, additional type I and II mutations have been related to inv(16). KIT mutations associated with inv(16) have been related to a worse prognosis. Here we describe the clinical and molecular data of a pediatric patient with AML, presenting inv(16) associated with a KIT pathogenic mutation. **Material and methods:** A 12-year-old boy with bleeding was referred to Pedro Ernesto University Hospital (HUPE), Rio de Janeiro, Brazil. The patient presented with suspected hyperleukocyte acute leukemia. Laboratory tests revealed a WBC count of 80×10^3, a platelet count of 9×10^3, and a hemoglobin level of 5.1 g/dL. Flow cytometry results demonstrated an immunophenotypic profile characteristic of LMA-M4. We conducted G-banding, FISH, and NGS studies. **Results:** Cytogenetic analysis was non-informative. Based on the clinical and immunophenotypic data, we conducted an experimental design using PML-RARA and CBFB-MYH11 FISH probes. The patient was negative for the PML-RARA fusion, but CBFb-MYH11 was confirmed. The NGS Assay revealed a pathogenic missense mutation in KIT. The results were negative for FLT3/ITD mutations. **Discussion:** AML patients presenting inv(16)/CBFb-MYH11 usually have a good prognosis with better remission and overall survival rates compared to those with normal karyotypes. However, due to the high rates of disease relapse, the prognostic value of alterations associated with inv(16)/CBFb-MYH11 has been widely discussed in the literature. Our patient presented with excessive bleeding and heavy hemorrhage, which initially raised the suspicion of acute promyelocytic leukemia. Due to the heterogeneity of the CBFb-MYH11 fusion, featuring distinct clinical and genetic profiles depending on its molecular size molecular and the association of these profiles with specific genomic alterations, a precise genomic characterization is necessary. KIT mutations, such as the one observed in our patient, are present as alterations associated with inv(16) in approximately 9%–53% of AML cases. Previous studies have demonstrated the potential prognostic impact of KIT mutations in the affected patients. In this context, missense mutations may lead KIT to an overactive state. This type of mutation has been shown to play a pivotal role in mediating clinical response and disease refractoriness in patients treated with targeted therapeutics. Therefore, the information provided through the NGS can be crucial for treatment adaptation. **Conclusion:** In this work, we contribute with the literature showing the clinical, molecular, and genomic data of an adolescent with AML and inv(16) associated with a KIT pathogenic mutation. Besides, we highlight the importance of NGS studies in characterizing the genomic profile of patients affected by this neoplasm. Although, the prognostic implications of these findings remain to be investigated, especially in the context of new generation tyrosine kinase inhibitors targeting KIT in future clinical trials for inv(16)/CBFb-MYH11 pediatric patients.

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NEXT-GENERATION GENOMIC MONITORING REVEAL A NOVEL RUNX1 PATHOGENIC MUTATION ASSOCIATED WITH TRISOMY 4 AS A SOLE ABNORMALITY IN A YOUNG ADULT WITH ACUTE MYELOID LEUKEMIA

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