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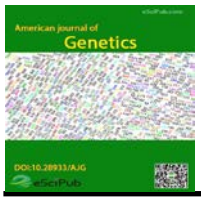

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Validation of the Conventional PCR technique for diagnosis of AML with NPM1 gene mutation

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ABSTRACT

Introduction: NPM1 gene mutations are the most frequent mutations in adults with acute myeloid leukemia (AML) they are found in 30% of adult de novo AML and in 60% of AML with normal karyotype. Because its clinical relevancy the European Leukemianet group recommends the research of NPM1 mutations at diagnosis to better stratify the risk of patients with AML. The current methodologies used to detect these mutations – Sanger sequence and fragment analysis- are expensive, therefore, there is the necessity of the development of new and cheapest methodologies for implementation in a routine diagnosis for patients with AML, mainly thinking of development countries. Objectives: To validate the technique of PCR followed by electrophoresis on agarose gel to detect mutations in NPM1 gene in patients with AML. Methodology: 196 patients with de novo AML were analyzed. Conventional PCR was made and the products of its amplification were seen by electrophoresis on agarose gel with 4%. The gold standard used for comparisons was the fragment analysis by the MEGABACE 1000 equipment (GE Healthcare-Amersham). Results and Discussion: 19,4% of the patients were mutated (38 of 196 patients) and 80,6% were normal (158 of 196 patients). All mutations were insertions of 4 base pair. The conventional PCR technique revealed a sensibility and specificity of 100% showing to be a sensitive, simple and economic method which can be used as an alternative to others more expansive methodologies that also need a more specialized professional to the diagnostic routine of patients with AML. Conclusions: This study proposes a faster, economic and sensitive technique to detect mutations on NPM1 gene in a routine diagnostic of patients with AML.

Keywords: Diagnosis; AML; NPM1; PCR

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