ANALYSIS OF C282Y AND H63D AND ALLELE DISTRIBUTION OF HFE PROTEIN GENE IN PATIENTS WITH HYPERFERRITINEMIA

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INTRODUCTION: Hereditary Hemochromatosis is a disorder that belongs to a group of conditions known as iron overload diseases. It is a genetic disease where a mutation in the gene for a regulatory protein (HFE), involves excessive high iron absorption and deposition this mineral in various organs. OBJECTIVES: This study aims to identify the C282Y and H63D genetic mutations in the gene for hemochromatosis (HFE protein gene) and evaluating the frequency of mutations in this gene in patients with hyperferritinemia assisted in the Centro de Oncologia e Hematologia de Mossoró-RN.

METHODOLOGY: Mutation identification was performed by Polymerase Chain Reaction (PCR) with the help of a restricted enzyme SnaBI called for C282Y and H63D for Bcl I. The fragments were subjected to enzymatic digestion and transilluminator was used for visualization. RESULTS AND DISCUSSION: Of the 58 patients studied to both mutations, the following features were found: C282Y mutation (WT/WT in 93,1%, WT/C282Y in 5,2% and C282Y/C282Y in 1,8%). The allelic frequency of the C282Y mutation was 4,3%. H63D mutation (WT/WT in 65,5%, WT/H63D in 27,6% and H63D/H63D in 6,9%). The allelic frequency of the H63D mutation was 20,7%. Milder form of HH is related to the presence of H63D variation, while its heterozygous state together with the C282Y enhances the risk of developing HH. The minority, about 1 to 2% of the compounds heterozygous C282Y/H63D develop clinical symptoms of hemochromatosis.

CONCLUSION: It was identified that the distribution of the C282Y and H63D mutation were similar to countries as Spain and France, suggesting an important role of people of these countries in our miscegenation. And is important to note that the meeting of a certain genotype determines a and not the genetic susceptibility to a clinical diagnosis HH, which in turn requires further analysis, such as liver function, transferrin saturation and ferritin.

Keywords: Hereditary Hemochromatosis, Mutations C282Y and H63D, Molecular Diagnostics.

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