glândula adrenal em pacientes com talassemia, 80% delas do lado direito e 73,3% tratados com cirurgia.1 HEM em sítios atípicos deve ser considerada em pacientes com TDT que não aderem ou não tem acesso a terapia transfusional adequada, levando a um diagnóstico diferencial importante com doenças neoplásicas ou metabólicas.

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HEMOGLOBIN A_2 AND FETAL HEMOGLOBIN PROFILES IN CD-39, IVSI-6, AND IVSI-110 BETA-THALASSEMIA MUTATIONS IN ASSOCIATION WITH HB S

LF De Lima, L Ramos Pereira, CR Bonini-Domingos

Universidade Estadual Paulista "Júlio de Mesquita Filho" (IBILCE/UNESP), São José do Rio Preto, SP, Brazil

Introduction: Beta-thalassemia is a hereditary hemoglobinopathy caused by mutations in the HBB gene on chromosome 11, which encodes β-globin chains. These mutations, often substitutions or deletions in coding regions or splicing sites, reduce or prevent β-globin synthesis. The imbalance between α- and β-chains leads to excess free α-chains that precipitate in developing erythrocytes, causing oxidative membrane damage, apoptosis of erythroid precursors (ineffective erythropoiesis), and extravascular hemolysis. Clinical severity ranges from asymptomatic (thalassemia trait) to severe (thalassemia major), with microcytic hypochromic anemia, jaundice, splenomegaly, skeletal deformities from marrow hyperplasia, and, in chronic cases, iron overload from transfusions and increased absorption. Laboratory findings include elevated HbA_2 (> 3.5%) in heterozygotes and, sometimes, increased HbF, aiding diagnosis. Common mutations are CD-39 (c.118C>T, β^0), causing absence of β-chains; IVSI-6 (c.92 +6T>C, β^+), with partial production; and IVSI-110 (c.93-21G>A, severe β^+), markedly impairing mRNA processing. These variants influence HbA_2, HbF, and clinical expression. **Objectives:** The objective of this study is to evaluate HbA_2 and HbF variations in these mutations when associated with Hb S. **Material and methods:** 23 individuals with an HPLC profile consistent with Hb S/beta-thalassemia were evaluated, divided according to the mutations identified: 6 with the CD-39 mutation in heterozygosis, 12 with the IVSI-6 mutation, and 5 with the IVSI-110 mutation. **Results:** For hemoglobin A_2 levels, median values were 4.25 ± 0.62% for the CD-39 mutation, 3.9 ± 0.31% for IVSI-6, and 4.1 ± 0.72% for IVSI-110. For fetal hemoglobin, median values were 5.80 ± 3.49% for CD-39, 12.1 ± 2.85% for IVSI-6, and 14.3 ± 3.25% for IVSI-110. **Discussion and conclusion:** The results obtained demonstrate that, regardless of the mutation, all individuals presented HbA_2 levels above the diagnostic cutoff for beta-thalassemia, reinforcing its role as a sensitive marker, as expected. The CD-39 (β^0) mutation exhibited the highest mean HbA_2 level (4.25 ± 0.62%), consistent with the complete absence of β-chain synthesis, which redirects compensatory production toward δ- and γ-chains. In contrast, IVSI-6 (β^+) showed the lowest mean (3.9 ± 0.31%), compatible with partial preservation of beta synthesis, thereby reducing the drive for delta-globin upregulation. IVSI-110 (severe β^+) presented an intermediate value (4.1 ± 0.72%), consistent with marked splicing impairment. Regarding fetal hemoglobin (HbF), a progressive gradient was observed — CD-39 (5.80 ± 3.49%), IVSI-6 (12.1 ± 2.85%), and IVSI-110 (14.3 ± 3.25%) — suggesting that β^+ mutations may favor greater compensatory activation of γ-globin synthesis, possibly due to prolonged ineffective erythropoiesis. The notably high HbF levels in IVSI-6 and IVSI-110 are clinically relevant, as increased HbF can mitigate clinical manifestations by improving overall oxygen transport capacity, particularly in association with Hb S. Thus, each mutation evaluated in β-thalassemia presents a distinct profile of HbA_2 and HbF, reflecting its specific impact on globin chain synthesis. While CD-39 (β^0) was associated with the greatest increase in HbA_2, the IVSI-6 and IVSI-110 (β^+) mutations showed markedly elevated HbF levels, possibly as a compensatory mechanism. These findings reinforce the usefulness of the combined assessment of hematological markers and molecular analysis in the diagnosis, prognosis, and genetic counseling of the disease.

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HEMOGLOBINOPATIAS NO ESTADO DE SÃO PAULO

LT Miura, S Soriano, L Figueiredo, MV Proença

Secretaria de Estado da Saúde de São Paulo, São Paulo, SP, Brasil

Introdução: O Ministério da Saúde, órgão federal responsável pelo Programa Nacional de Pessoas com Doença Falciforme e outras Hemoglobinopatias, desenvolveu um sistema informatizado chamado de Web - Hemoglobinopatias com a finalidade de sistematizar informações para monitoramento das doenças através da manutenção/cadastro de pacientes portadores de hemoglobinopatias, onde sua utilização adequada nos serviços assistenciais poderá favorecer no manejo do tratamento do paciente. As hemoglobinopatias são doenças genéticas e hereditárias, nos quais ocorrem alterações estruturais na hemoglobina, redução ou ausência na síntese da hemoglobina, causando diversas complicações clínicas ao indivíduo. O diagnóstico precoce associado ao avanço de tecnologias para tratamento resultaram em um aumento na estimativa de vida dos pacientes, diminuindo o número de mortes. A transfusão sanguínea é uma das principais terapias não medicamentosa utilizada no tratamento das hemoglobinopatias (em especial a anemia falciforme e a talassemia),