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Clinical Manifestation and Molecular Diagnosis of Beta-Thalassemia Patients

A thesis submitted in partial fulfillment of requirements for the Master 's Degree of Science in Biology / Zoology

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Abstract

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Beta-thalassemias are a group of hereditary hemoglobin disorders in Iraq caused by different mutations in β-globin gene coding for β-globin chains of hemoglobin. The β-thalassemia mutations result to insufficient or absent β-globin synthesis. As a result, affected individuals have different degrees of severity of anemia and hemolysis, which in turn can cause bone changes, impaired growth, and iron overload. This study was aimed to investigate molecular, serological and hematological aspects of β-thalassemia patients in Al-Muthanna province. Ninety-eight blood samples collected from β-thalassemia patients in Women and Children Hospital, thalassemia unit in Al-Muthanna province, aged 1-29 years old and the samples used 61 males and 37 females. Blood samples were taken simultaneously from 32 control individuals to be used as control. Blood samples used for serum measurement of Ferritin, Vitamin D3 and 8-hydroxydeoxyguanosine using Enzyme Linked Immunosorbent Assay technique. Genomic DNA extraction from peripheral blood for detection of β-globin mutations frequency by using direct DNA Sanger sequencing. Result found mean of hemoglobin was significantly decreased in patients group in comparison to control subjects ($P<0.001$). The mean of ferritin concentration was significant increase in patients group in comparison to control subjects ($P<0.001$). The mean 25-hydroxy vitamin D was significant decrease in patients group in comparison to control subjects ($P<0.001$). The present study showed that the mean 8-OHdG was significant increase in patients group comparison to control subjects ($P<0.001$). Analysis revealed the presence of nine mutations in 98 β-thalassemia patients of which seven mutations not previously recorded in Iraq, including IVS-II-666 C> T, CD2 CAT>CAC , IVS-II-850 G>A, IVS-II-16 G>C , Hb kangsmill, Hb Saveh , and IVS-II-81 C>T, as well as two mutations previously recorded in Iraq include IVS II-1 G>A and IVS 1-5 G>C.