Association between Hemochromatosis *HFE*, *HAMP*, *TFR1* and *TFR2* Genes Variants and Iron Overload of Beta Thalassemia Patients

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Abstract

Thalassemia is an inherited blood disorder in which the body makes an abnormal form of hemoglobin. It is two types α and β according to hemoglobin chain defect. Treatment of β thalassemia depends on recurrent blood transfusion that leads to increased blood ferritin level. β thalassemia disease and hemochromatosis may be compatible in the same patients due to defects in the iron metabolism mechanism leading to iron overload. Many mutations may have an incidence of thalassemia and hemochromatosis. Much more iron is harmful to the body and can cause tissue damage over time. This study was aimed to determine the effect of some mutations (C282Y, H63D mutations in the *HFE* gene, G71D mutation in the *HAMP* gene, G142S mutation in the *TFR1*, and Y250X mutation in *TFR2* gene) on blood ferritin levels and iron overload in β thalassemia patients. Eighty-four samples (21 β thalassemia patients, 42 parents as carriers, and 21 healthy controls) were included in this study. The period of collected samples was from January to April 2020. The genotyping of genes mutations was conducted by the Polymerase Chain Reaction-Restriction Fragment Length Polymorphism method. The mean of serum ferritin levels in β thalassemia patients were 1810.5 ng /ml and the mean of hemoglobin in β thalassemia

patients was 8.2 g/dl. as well MCV, MCH was (81.5 fL, 25.9 pg) respectively. The C282Y mutation was not found in all 84 samples, for H63D mutation was nonsignificant between iron overload and β thalassemia patients and parents the P-value was (p=0.08, p=0.74) respectively. In addition, a nonsignificant association of G71D mutation between iron overload and β thalassemia patients but significant with parents, the P-value were (p=0.44, p<0.001) respectively. The G142S mutation was significant for patients and nonsignificant for parents, to the association between iron overload and β thalassemia, the P-value were (p=0.02, p=0.47) respectively and finally, the Y250X mutation was not found in all groups. The β thalassemia patient in Samawah city has a significant effect of iron overload with G142 but nonsignificant with (H63D, G71D) mutation. On the other hand (C282Y, Y250X) mutations were not found in all groups of this study.