Abstract

Thalassemia is a genetic disorder most commonly found in Indonesia. Iron absorption in β thalassemia carrier has doubled compared to individuals with normal hemoglobin levels, causing an increase in iron accumulation and increased susceptibility to iron overload. HFE gene is one of the alleged tertiary modifier on the severity of iron overload complication in thalassemia. HFE gene plays a role in hereditary hemochromatosis disorders characterized by iron overload. H63D HFE gene polymorphisms alter the ability of HFE to reduce the affinity of the transferrin receptor (TFR), causing an increase in the iron status of individuals with mutant-type genotype. To identify the presence of HFE gene polymorphism and association of HFE gene polymorphism with iron status of Javanese ethnic β thalassemia carriers. H63D gene polymorphism and iron status were assessed in Javanese β thalassemia carriers. Thirty-six subjects (23 females and 13 males) were involved in this study. Isolated genomic DNA was used to detect polymorphism in H63D gene by using PCR and RFLP based methods. The mean of serum iron levels, transferrin saturation and serum ferritin levels in female β thalassemia carrier were 69.6 ± 24 mg / dl, 28 ± 15.3% and 95.6 ± 69.7 ng /ml, respectively. The mean of serum iron levels, transferrin saturation and serum ferritin levels in male β thalassemia carrier were 85.8 ± 31 mg / dl, 34.6 ± 16.5% and 150.5 ± 90.9 ng / ml, respectively. There was one female proband with heterozygous mutant of H63D gene however her iron status was within normal reference values. Polymorphism of H63D HFE gene is rarely found in Javanese β thalassemia carrier. There is no increase in iron status of Javanese β thalassemia carrier with H63D heterozygote mutant.

Keywords: β thalassemia carrier, HFE H63D, iron status, Javanese ethnic

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