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# DETECTION OF EXON 1 MUTATIONS IN THE β-GLOBIN GENE ON β-THALASSEMIA CARRIER IN YOGYAKARTA

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## Abstract

Thalassemia is a hereditary dissorder caused by point mutation on the globin gene. β-thalassemia is the most common type in Indonesia. This study aimed to performed the molecular detection of exon 1 mutation in the β-globin gene by using polymerase chain reaction-single strand conformation polymorphism (PCR-SSCP) method. DNA samples was collected during 2012 to 2013 in Yogyakarta. DNA of each individual suspected β-thalassemia carriers was isolated from blood and amplified with specific DNA primer dan PCR conditions. Individual suspected β-thalassemia carriers with mutant alleles showed more than two DNA bands on polyacrilamide electrophoresis analysis. PCR-SSCP results showed that 17 of 21 individual suspected β-thalassemia carriers have mutation site in exon 1 β-globin gene.

Keywords: mutation, exon 1, β-globin gene, β-thalassemia carrier