DEVELOPMENT OF SINGLE-STRAND CONFORMATIONAL POLYMORPHISM TO SCREEN FOR MUTATIONS IN HOTSPOT REGIONS OF BETA-GLOBIN GENE OF BETA-THALASSEMIA PATIENTS OF SRI LANKA

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Abstract. Beta-thalassemia is prevalent in Sri Lanka and imposes a heavy economic and social burden in the country due to the patients’ life-long need for regular blood transfusion and treatment with iron chelation therapy. Thus, there is a need to develop a rapid, reliable and effective population-based presymptomatic and prenatal screening method for beta-thalassemia. Single-strand conformational polymorphism (SSCP) technique was developed as an adjunct for the previously developed allele-specific PCR (ASP) technique to screen the presence of mutations in beta-globin gene. A hotspot region of beta-globin gene containing 98% of known beta-thalassemia mutations was amplified from 24 clinically diagnosed beta-thalassemia patients and two normal individuals. Two overlapping amplicons of 238 bp and 268 bp were subjected to SSCP analysis. The SSCP banding patterns of these two fragments from beta-thalassemia patients were different from the corresponding regions of normal individuals. Sequence analysis of these regions revealed the presence of 4 mutations in the form of deletion and substitution that have not been reported previously from Sri Lanka. Therefore, the SSCP protocol developed in this study together with ASP should provide an appropriate screening approach for presymptomatic and parental diagnosis of beta-thalassemia in the Sri Lankan population.

Keywords: single-strand conformational polymorphism, beta-globin, beta-thalassemia, allele specific priming technique, mutations, Sri Lanka