

## **Detection of common beta globin gene mutations on heterozygous beta thalassaemia including haemoglobin E among high school students**

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Beta thalassaemia is one of the most common genetic disorders worldwide with a defect in haemoglobin synthesis including Myanmar. This study was conducted to determine the proportion of high school students who have heterozygous  $\beta$ -thalassaemia including Hb E and different types of common beta globin gene mutations by multiplex polymerase chain reaction. This community-based cross-sectional descriptive study was done in Myanmar adolescents in 2018. A total 290 apparently healthy high school students in which male 140 (48.3%) and female 150 (51.7%) studying in Basic Education High School, Anisakhan, Pyin Oo Lwin Township, Mandalay District, Myanmar were involved. Thalassaemia screening tests were done by combined NESTROFT/DCIP precipitation and confirmed test was done by Isoelectric Focusing (IEF). PCR- based genetic confirmatory by using mutant genes IVS 1-1, CD 41/42, IVS 1-5, CD 17, and nt-28. Total 131 cases (131/290, 45.2%) were positive with either of the two screening tests. These cases were considered as thalassaemia trait (carrier) and/or haemoglobin E carrier. Isoelectric focusing was done to find Hb E by which 45 cases (45/131, 34.3%) were found to have Hb E carrier. Remaining 86 cases were further run multiplex- PCR to detect beta thalassaemia carrier. Total 44 out of 86 cases were  $\beta$ -thalassaemia traits (25 CD 41/42 (-TCTT), 13 IVS 1-5 (G-C), 1 IVS 1-1 (G-T); 5 compound heterozygous (IVS 1-5(G-C) /CD 41/42 (-TCTT)). CD 17 (A-T) and nt 28 (A-G) mutations were not detected in this study. NESTROFT screening test has 87% sensitivity and 82% sensitivity for the detection of Hb E carrier and  $\beta$ -thalassaemia carrier respectively. DCIP screening test has 67.1% sensitivity and 68% sensitivity for the Hb E carrier and  $\beta$ -thalassaemia carriers respectively. Combined both screening tests have 100% sensitivity for the detection of Hb E and  $\beta$ -thalassaemia carriers. This study has highlighted that both screening tests should be used for population screening of thalassaemia and haemoglobinopathies in Myanmar.

**Key Words:** beta thalassaemia, beta globin gene, haemoglobin E, high school students