Introduction

- Beta thalassemia is caused by variations encoding the human beta-globin gene (HBB).
- The most common HBB gene mutation reported in Indian population is IVS I-5 (G/C) followed by IVS I-1 (G/T), 619-bp deletion, Codon 41/42(-TCTT) and Codon 8/9 (+G).
- No study has been conducted in the densely populated state of Kerala to understand the prevalence of mutation.

Objectives

- To evaluate the prevalence of HBB gene variations in beta thalassemia patients in Kerala state.
Materials and Method

• Forty-three patients from the Department of Pediatric genetics, AIMS, Cochin during the period of 2018-2019 with symptoms of beta-thalassemia were included in this study.

• DNA was extracted and sequenced for the HBB gene using Sanger sequencing method. The sequences were aligned to the HBB reference sequence (NG_000007.3) and compared using variant analysis programs codon code aligner Ver.7.2.

Results

• We have identified pathogenic variations in HBB gene for 31/43 (72%) patients analysed in our laboratory.

• The common variation identified in our study was NM_000518.5(HBB):c.92+5G>C variation (28%), followed by NM_000518:c. 20A>T; p.Glu7Val sickle cell variant and NM_000518.5(HBB):c.79G>A (p.Glu27Lys) in compound heterozygous patients.

Conclusions

• We are hereby reporting for the first time the HBB mutation pattern from the state of Kerala.

• The most common variation identified in our study was NM_000518.5(HBB):c.92+5G>C variation seen in 28% of patients.