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Concurrent Inheritance of Deletional α-thalassaemia in Malays with HbE Trait

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ABSTRACT

Introduction: HbE is the commonest beta haemoglobin (Hb) variant in Southeast Asia. It causes a reduction in synthesis of the beta-globin E (β^E) chain. Studies indicate HbE coinherited with α-thalassaemia leads to a milder clinical phenotype. This study investigates the concomitant inheritance of α-thalassaemia in Malays with HbE. **Methods:** Four hundred and fourteen (414) blood samples were screened for haemoglobinopathy using primarily the first 3 steps of the BHES [(B) blood counts, blood film; (H), HPLC; (E), electrophoresis; (S), stability)] protocol. Complete blood counts were generated on an automated blood cell analyser, Hb typing with cation exchange high-performance liquid chromatography (HPLC) and Hb electrophoresis at an alkaline pH (pH 8.5). Forty-five (10.9%) were identified as HbE trait and DNA analysis was done for deletional α-thalassaemia using a single-tube multiplex-PCR assay. **Results:** Among the 45 subjects with HbE trait, 4 (8.9%) were found to have alpha-thalassaemia-2 (α^+) ($\alpha^{-3.7}$ kb deletion) and 1 (2.2%) the alpha-thalassaemia-1 (α^0) ($—^{SEA}$ 20.5 kb deletion) defects respectively. **Discussion:** These findings show that 11.1% of Malays with HbE inherit alpha-thalassaemia concurrently. The most prevalent interaction found was a double heterozygote for HbE/α-thalassaemia 2, followed by HbE/α-thalassaemia 1. **Conclusion:** Molecular screening of deletional α-thalassemia identified its concurrent inheritance in 11.1% of Malays who were HbE carriers. This information will guide genetic counseling and the planning of treatment modalities in patients with HbE alpha-thalassaemia.

Keywords: α-thalassaemia, concurrent inheritance, HbE trait, Malays

INTRODUCTION

Genetic defects of haemoglobin synthesis (haemoglobinopathies) are the most common genetic disorders worldwide. It occurs mainly in tropical and subtropical areas.[1] HbE is the commonest beta-globin chain variant in Southeast Asia. The frequency of HbE approaches 60% in many regions of Thailand, Laos and Cambodia.[2,3,4,5] It is also found in Sri Lanka, North Eastern India, Bangladesh, Pakistan, Nepal and Vietnam. In Malaysia, micromapping studies indicate 4% of Malays are carriers of HbE.[6]

In HbE, an alternate splicing site found within exon 1 of the beta-globin gene in the primary mRNA transcript causes reduced production of the β^E-globin chain.[2,5]

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