

Interaction of Hb South Florida (codon 1; GTG→ATG) and HbE, with β -thalassemia (IVS1-1; G→A): expression of different clinical phenotypes

Abstract

Introduction Interactions of different hemoglobin variants with thalassemia alleles can result in various clinical phenotypes. HbE- β -thalassemia generally manifests with severe anemia where individuals exhibit β -thalassemia major with regular blood transfusions or β -thalassemia intermedia with periodic blood transfusions. This study presents a unique Malay family with three β -globin gene defects—HbE, Hb South Florida, and IVS1-1 (G→A). **Materials and methods** HbE activates a cryptic splice site that produces non-functional mRNAs. Hb South Florida is a rare β -hemoglobin variant, and its interactions with other β -thalassemia alleles have not been reported. IVS1-1 is a Mediterranean mutation that affects mRNA processing giving rise to β o-thalassemia. **Results and discussion** Fifteen mutations along the β -globin gene complex were analyzed using the amplification refractory mutation system. Hb South Florida was identified by direct sequencing using genomic DNA. **Conclusion** The affected child with HbE/IVS1-1 produced a β -thalassemia major phenotype. Compound heterozygosity for Hb South Florida/IVS1-1 produced a β -thalassemia carrier phenotype in the mother.

Keyword: Amplification refractory mutation system; Direct sequencing; Ethnic groups; Structural hemoglobin variants; Thalassemia