CLINICAL AND HEMATOLOGICAL PHENOTYPE OF HOMOZYGOUS HEMOGLOBIN E: REVISIT OF A BENIGN CONDITION WITH HIDDEN REPRODUCTIVE RISK

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Abstract. Hemoglobin E (HbE) is one of the most prevalent β-globin variant, which is widely distributed in Southeast Asia especially in Thailand. Homozygosity for this variant is common and may occur with iron deficiency. In order to study clinical and hematological phenotypes without the confounding effect of iron deficiency, investigations were carried out before and after iron supplementation for 2 months. The effect of G6PD deficiency and coinheritance of α-thalassemia in homozygous HbE were also studied. HbE homozygotes were clinically benign, never had been transfused and had no hepatosplenomegaly. Out of 76 HbE homozygotes, hematological parameters of 7 individuals with iron deficiency improved after iron supplementation. Hemoglobin analysis revealed that HbE was the main hemoglobin detected, but 12 subjects were found to have a substantial percentage of HbF, which might lead to misdiagnosis as HbE/β-thalassemia. Both clinical and hematological phenotypes of simple homozygous HbE did not differ from those who also inherited α-thalassemia and/or G6PD deficiency. It is necessary to perform a comprehensive DNA analysis for α-thalassemia in cases of homozygous HbE when their partner is suspected of having α-thalassemia 1 gene.

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