Molecular and hematologic features of hemoglobin E heterozygotes with different forms of α-thalassemia in Thailand

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Abstract
We describe the hematological and DNA characterization of hemoglobin (Hb) E heterozygote with various forms of α-thalassemia in Thai individuals. Altogether, 202 unrelated adult subjects with Hb E heterozygotes either with or without α-thalassemia determinant were studied. The most prevalent interaction was found to be a double heterozygote for Hb E/a-thalassemia 2, followed by a double Hb E/α-thalassemia 1 and a Hb E/ Hb Constant Spring (CS), even though the Hb CS was not detected. Double heterozygotes for Hb E and homozygous α-thalassemia 2 and Hb E with a compound α-thalassemia 2/Hb CS were also encountered with lower frequencies. Unexpectedly, as many as 18 cases previously diagnosed as Hb E carriers at routine Hb analysis were indeed Hb E heterozygotes with compound α-thalassemia 1/α-thalassemia 2, indicating a need for globin genotyping for accurate diagnosis. A change in Hb E level was observed which was related to a concomitant inheritance of athalassemia. The hematological expression of these Hb E heterozygotes with various forms of α-thalassemia, including a hitherto undescribed condition of double heterozygosity for Hb E/Hb Paks_ identified in two subjects, is presented comparatively with those of the 80 cases of pure Hb E carriers. A multiplex allele-specific polymerase chain reaction (PCR) assay for simultaneous detection of Hb E and Hb CS genes is also described.

Keywords: Hemoglobin E, Hemoglobin Constant Spring, Hemoglobin Paks, α-Thalassemia, EABart’s disease

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