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No Evidence for Role of Common Anion Exchanger 1 Mutations on the Severity Difference in HB E-β-Thalassemia Disease in Northeast Thailand

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ABSTRACT In a distal renal tubular acidosis, a patient with coexistence of anion exchanger 1 (AE1) mutation and α-thalassemia had more severe clinical phenotype than usually observed. AE1 mutation might therefore modify the hematological and clinical presentations of the hemoglobin (Hb) E-β-thalassemia disease. To examine the role of AE1 mutation on severity difference in Hb E-β-thalassemia disease in northeast Thailand, three common AE1 mutations were investigated in Hb E-β-thalassemia patients with thalassemia major (TM) and thalassemia intermedia (TI) phenotypes. One hundred forty-eight patients including 103 TM and 45 TI were studied. Three common AE1 mutations including Southeast Asian Ovalocytosis (SAO), G701D and A858D were screened for using allele specific polymerase chain reaction (PCR) and PCR-restriction fragment length polymorphism (RFLP) assays. No any AE1 mutation was detected in all these Thai patients with Hb E-β-thalassemia. This result indicates that AE1 mutation is rare and should not be an important phenotypic modifier in the Hb E-β-thalassemia disease. Therefore, screening of common AE1 mutation in thalassemia patient is not necessary.