ABSTRACT

The mutations in β-globin gene from 6 Thai patients with previously uncharacterized β-thalassemia/HbE disease were studied using a RNase protection assay. The results showed that one of the patient had a mutation in the region of codons 41-42. Direct DNA sequencing of the amplified β-globin gene identified a cytidine deletion in codon 41 (TTC -> TT) causing a frameshift mutation and resulting a premature termination at codon 60/61. This is the first report of this type of rare β-globin gene mutation in conjuction with HbE.