Hb Loei [β31(B13)Leu⟶Met] : a novel mutation in exon 2 of beta globin gene found in association with HbE [β26(B8)Glu⟶Lys] and a deletional α^0-thalassemia ($--^{SEA}$)

Yupin Jopang[1], Kobkorn Pongjantharasatien[2], Phatareeya Laochinchat[2], Sumalee Jindadamrongwech[2], Panida Jantharachit[3], Rawiwan Puangplruk[1], and Paripat Netnee[1]

[1] Regional Health Promotion Center 9, Nakhon Ratchasima, Thailand

[2]Department of pathology, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Bangkok, Thailand

[3] Nongbaurawae hospital, Chaiyaphum, Thailand

A 34-year-old Thai male visited the antenatal care unit with his wife, in order to identify carrier couple in the public health strategy for the prevention and control of thalassemia. Initial hemoglobin (Hb) analysis showed Hb E heterozygote pattern with 32.5% by a high performance liquid chromatography (HPLC) system. It is generally accepted that Hb E heterozygote with Hb E level of 25-35 % does not co-existing with a deletional α^0-thalassemia, however, all Hb E heterozygote who having α^0-thalassemia carrying partners are prospectively screened for the presence of α^0-thalassemia. Of course, mutation was further identified by PCR and related techniques while his wife was a carrier of α^0-thalassemia ($--^{SEA}$). DNA sequencing and PCR analysis demonstrated that he was a carrier of a complex interaction of a novel mutation in exon 2 of beta globin gene [β31(B13)Leu⟶Met] which we named Hb Loei, Hb E [β26(B8)Glu⟶Lys], and a deletional α^0-thalassemia ($--^{SEA}$). This combination of three molecular defects and atypical heterozygous Hb E expressed is the first case reported in the literature. Thus, it would be necessary to exclude co-existing α^0-thalassemia in all Hb E heterozygote and homozygote whose partners were α^0-thalassemia carriers.

Keywords: hemoglobin Loei; hemoglobin E; α^0-thalassemia; hemoglobin variant