INFLUENCE OF IRON DEFICIENCY AND ALPHA THALASSEMIA-2 ON THE DIAGNOSIS OF BETA THALASSEMIA CARRIER DETERMINED BY HEMOGLOBIN A2 LEVEL

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Background: A number of genetic or acquired conditions are capable of reducing hemoglobin (Hb) A_2 which is critical for the diagnosis of heterozygous beta thalassemia. This work conducted to study the influence of iron deficiency and alpha thalassemia-2 gene upon the diagnosis of heterozygous beta thalassemia determined by Hb A_2 quantitation in the thalassemia prevention and control program.

Methods: One thousand one hundred and forty seven pregnant women and their spouses who attended the antenatal clinic at Buddhachinaraj hospital, Phitsanulok from November 2004 to July 2005 were prospectively recruited for thalassemia screening using kinetic osmotic fragility test. Positive results would be underwent further thalassemic investigation with high performance liquid chromatography for Hb A_2 quantitation, measured serum ferritin level for the diagnosis of iron deficiency and performed alpha thalassemia-2 polymerase chain reaction. Direct DNA sequencing was performed in all participants who had normal Hb A_2 level with iron deficient state and / or had concomitant alpha thalassemia-2 gene. Outcome of interest was the diagnosis of heterozygous beta thalassemia with normal Hb A_2 level in these 2 conditions.

Results: From 1,129 evaluable participants, there were 22 (1.9%) with high Hb A_2 level (> 4%) which made the diagnosis of heterozygous beta thalassemia. One hundred and thirty one participants with normal Hb A_2 level who had iron deficiency (ferritin < 15 microgram / liter) and / or alpha thalassemia-2 gene (40 iron deficiencies, 80 alpha thalassemia-2 carriers, 11 with both conditions) were underwent DNA sequencing for beta thalassemia mutation with no positive results. On the other hand, there were 5 heterozygous beta thalassemia (Hb $A_2 > 4\%$) who had concomitant iron deficient state or alpha thalassemia-2 gene (2 iron deficiencies, 3 alpha thalassemia-2 carriers) detected. No homozygous alpha thalassemia-2 was detected in this study.

Conclusions: It seems no needs looking for iron deficiency and alpha thalassemia-2 gene for the diagnosis of heterozygous beta thalassemia determined by Hb A_2 quantitation in Phitsanulok and lower northern area of Thailand. However, it is too early to conclude that homozygous alpha thalassemia-2 will not be the problem in the diagnosis of heterozygous beta thalassemia in region where both alpha and beta thalassemia are in high prevalence such as Chiang Mai and upper northern Thailand.