RESEARCH NOTE

SURVEY OF COUPLES IN UPPER NORTHERN THAILAND (JANUARY - DECEMBER 2019) AT RISK OF HAVING NEWBORNS WITH THALASSEMIA DISEASE

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Abstract. Prevalence of thalassemia (thal) varies among different regions resulting in different proportions of couples at risk of carrying fetuses with thal disease. From January to December 2019, prevalence and genotypes were determined of couples (n = 370) in upper northern Thailand for risk of having newborns with thal diseases (homozygous α^0-thal (Hb Bart's hydrops fetalis), β-thal major and β-thal/Hb E). Based on Hb typing and DNA analysis, allele frequency of α^0-thal--^{SEA}, β-thal and Hb E was 0.034, 0.072 and 0.086, respectively, and the estimated number of newborns with Hb Bart's hydrops fetalis, β-thal major and β-thal/Hb E per 10,000 pregnancies was 12, 52, and 124, indicating for the identified at-risk couples (n = 28) of 5, 3 and 20 newborns, respectively. β0-thal codon 17 (A>T) and codon 41/42 (-TCTT) mutations were the most common alleles. Homozygous and compound heterozygous β-thal major individuals were also identified. Information from this study can be applied to prevention and control strategies of these severe thalassemias in Thailand and other parts of Southeast Asia.

Keywords: at-risk couple, β-thalassemia major, β-thalassemia/Hb E, Hb Barts's hydrops fetalis, northern Thailand, severe thalassemia disease

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