Prevalence of Thalassemia in Southeast Asia

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Introduction

Thalassemia is a hereditary red blood cell disorder. It is due to globin gene mutations in either alpha and/or beta globin genes resulting imbalance in numbers of alpha (α) and beta (β) chains in red blood cells. There are two major types of thalassemia which are α- and β-thalassemia, in which the former is the most common form of thalassemia worldwide especially in Southeast Asia populations[1,2]. In 2020, it was estimated that 6.8% of Malaysians are thalassemia carriers[3]. This study aimed to determine the prevalence of both α- and β-thalassemia among the KadazanDusun populations in Sabah and pooled the data (from 2010 to 2020) to estimate the prevalence rate of α-thalassemia in the Southeast Asia region through a comprehensive meta-analysis.

Materials & Methods

DNA was extracted from 125 blood samples of unrelated KadazanDusun individuals in Sabah, East Malaysia with written consent

Detection of α- and β-thalassemia in the KadazanDusun populations using a polymerase chain reaction approach

Data of α-thalassemia was pooled with other populations in the Southeast Asia countries for a comprehensive meta-analysis

Results & Discussion

Prevalence of α- and β-thalassemia in the KadazanDusun populations in Sabah

Single α-globin gene deletion (-$\alpha^{3.7}$) was detected in 42/125 KadazanDusun individuals whereas 16/125 of the studied population possessed the Filipino β-thalassemia deletion in the β-globin gene (Figure 1).

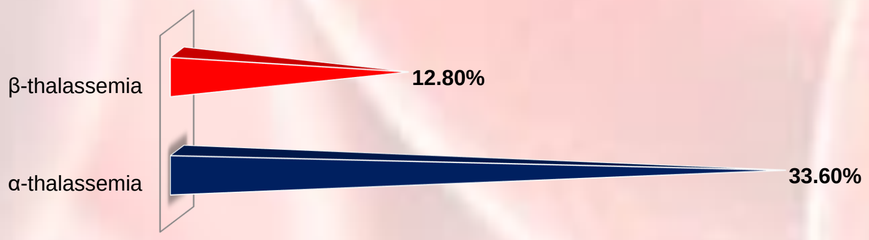

Figure 1: Prevalence of α- and β-thalassemia among the KadazanDusuns.

Prevalence of α-thalassemia in Southeast Asia

Overall, the prevalence rate of α-thalassemia occurrence in the Southeast Asia region was 22.6%. There is a reduction of ~50% of the prevalence in the Southeast Asia region since 2010. The World Health Organization had reported the α-thalassemia prevalence as 44.6% in 2008[4].

In the subgroup analysis based on country, Vietnam had the highest prevalence rate (51.5%) of α-thalassemia followed by Cambodia (39.5%), Laos (26.8%), Thailand (20.1%), and Malaysia (17.3%) (Table 1 and Figure 2).

Table 1: Prevalence rate and heterogeneity of a-thalassemia in this study

Heterogeneity		Prevalence rate (95% CI)	Sample size (*N*)	No. of Studies (*N*)	Subgroups
I^2 (%)	p-value				
99.53	<0.001	0.226 (0.168-0.296)	83,674	32	Overall
0	0.560	0.395 (0.372-0.419)	1,652	2	Cambodia
97.26	<0.001	0.268 (0.096-0.559)	812	3	Laos
98.20	<0.001	0.173 (0.060-0.407)	2,526	5	Malaysia
99.47	<0.001	0.201 (0.143-0.273)	76,955	20	Thailand
99.22	<0.001	0.515 (0.190-0.828)	1,729	2	Vietnam

No α-thalassemia-related studies that fulfilled our inclusion and exclusion criteria from other Southeast Asia countries, including Brunei, Indonesia, Myanmar, Philippines, Singapore, and Timor-Leste.

Most Southeast Asia countries have thalassemia screening programs. In Malaysia, the National Thalassemia Screening Program started in late 2004.

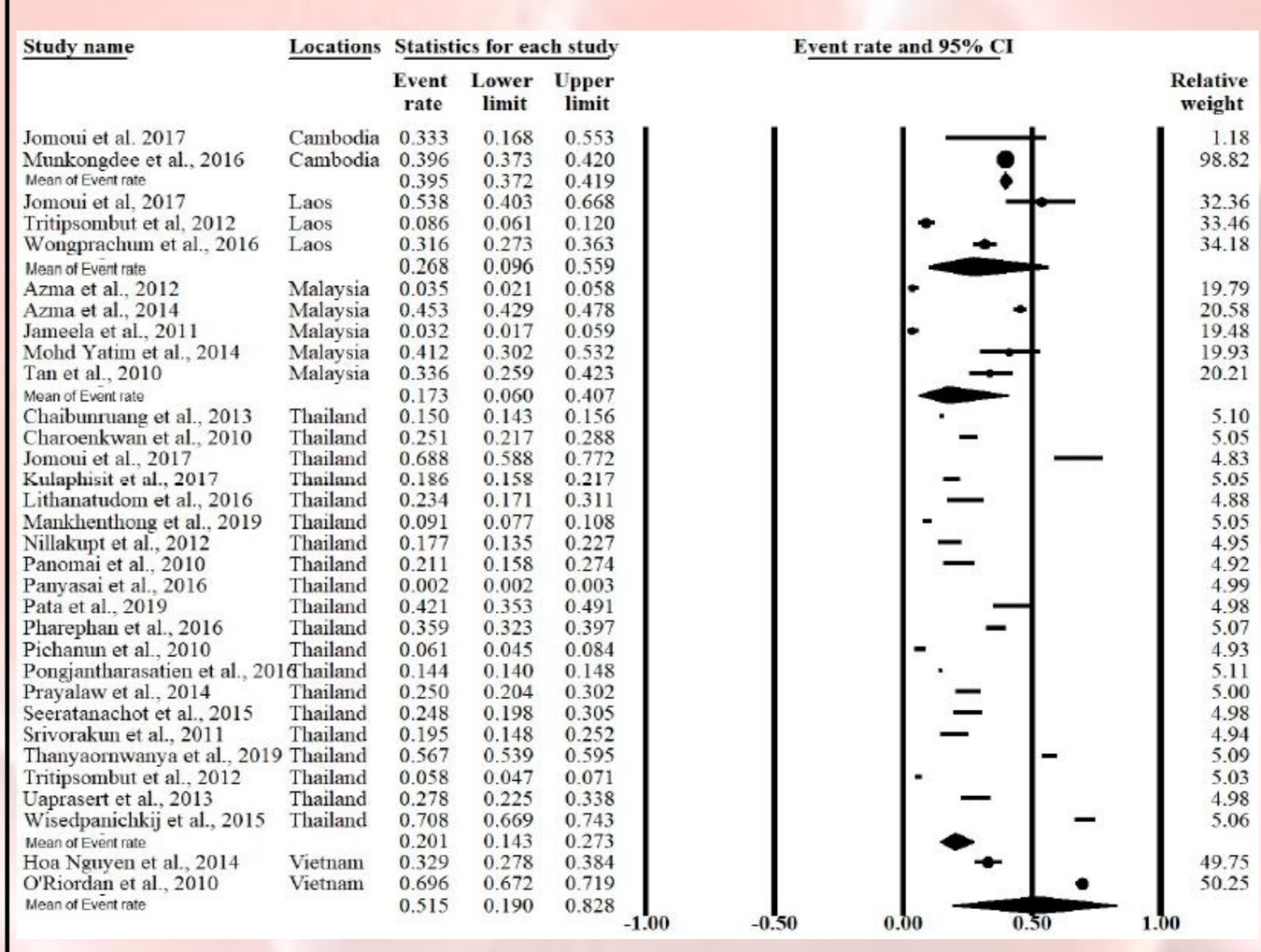

Figure 2: Forest plot of the α-thalassemia prevalence grouped according to country

Conclusions

This study suggests a high prevalence of α- and β-thalassemia occurred in the KadazanDusun population. Meta-analysis also showed a high prevalence of α-thalassemia in Southeast Asia that provides a perspective to design healthcare policies with better genetic counselling programs for thalassemia.

References

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