
Exploring the Willingness for Carrier Screening among Extended Family Members of a Thalassaemia Carrier Individual: A Lesson Learned

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Thalassaemia carrier screening is commonly conducted among direct-related or immediate family members of thalassaemia patients followed by a counselling about the thalassaemia. In countries where prevalence of thalassaemia carrier is high, carrier screening in general population is mandatory for example in pregnant women, in high school students, young adults or even before marriage [1, 2]. This screening may play a major role for carrier identification. A study conducted in Pakistan showed that siblings identified as β -thalassaemia carriers is higher as opposed to carriers in the general population i.e. 62.2% vs 5 to 8 %, respectively [3]. Therefore, it is practical to focus on siblings of identified thalassaemia patients when both resources and budgets are limited. In Indonesia, thalassaemia major is placed as number 6th in the catastrophic list. Although Indonesia harbours 6 to 10% thalassaemia carriers, carrier screening has not been put as mandatory yet. Furthermore, the willingness for carrier screening is still lacking. The purpose of this report was to explore the willingness for carrier screening in an extended family members of thalassaemia carrier individuals. This was an observational and descriptive study, conducted during a cross sectional event for thalassaemia carrier screening during a family gathering for 3 generations. A thalassaemia carrier individual, designated as Index Carrier (IC) aged 20 years old, was identified during a regular medical check-up prior student admission to the university. After counselling, the IC intended to screen the extended family from both father's and mother's side. The family was approached and during their family gathering, thalassaemia case story was introduced and shared. A family tree was drawn to identify whether thalassaemia patients presence among the extended family. The carrier screening was initially offered for the unmarried children older than 15 years old who came in the gathering, however, their parents were also encouraged to screen for the carrier status. After

verbal informed consent and permission of the parents, blood was drawn in an EDTA tube. Complete blood count was measured and Shine and Lal index ($MCV^2 \times MCH/100$) was calculated. Value of <1530 was designated as β -thalassaemia carriers as described earlier [4]. MCV and MCH value of <75 fl and <25 pg, respectively, were further subjected for HbA2 measurement. Those who were suspected β -thalassaemia carriers were counselled individually with their parents, especially for those who were younger than 18 years old.

None of the direct-related and extended family members of IC knew about thalassaemia as a disease. The family tree had identified a suspected thalassaemia major patient, i.e. the son of grandfather's brother of IC, from the mother side, who died 30 years ago at aged 5 years old after receiving several times of blood transfusions. This thalassaemia patient had 2 sisters; the first sister later migrated to Australia, and admitted during the gathering that she was a carrier, detected during her first pregnancy with all of her three children were also carriers, detected later in high school after knowing that the mother was a carrier. The second sister in Indonesia didn't know much about the disease, and both sisters never discussed about this disease until the gathering event. She was not willing to be screened, nor her '*healthy appearance*' children. Stigma and anxiety may play a major role; therefore, when an individual is detected to have thalassaemia carrier status, screening to extended family needs to be taken seriously into account. This is especially when no thalassaemia cases in the family members. Thalassaemia information need to be introduced and the knowledge need to be enhanced towards a positive attitude and perception. Individual with higher thalassaemia knowledge would consider and have better willingness to examine their thalassaemia carrier status earlier [5]. Haematology results showed that 6 (3 boys and 3 girls) of 16 examined unmarried children (37.5%) from 6 families had MCV <65 fL and MCH <20 pg, with Shine and Lal index <1530 , indicating β -thalassaemia carriers, confirming by HbA2 value of $>3.5\%$. Interestingly, all boys had normal Hb and all girls had moderate anemia (8-10 g/dL) and they all felt healthy and active in sports. One of those carriers was a sibling of IC while the other IC's sibling was normal. Further examination showed that IC's mother was a carrier but not the father. The most common β -thalassaemia mutation in West Java is IVS1nt5, followed by IVS1nt1, and HbE (unpublished data), therefore, DNA analysis is important to explore mutation types, needed for the global data.

The limitation of this study was that there was no iron measurement and DNA analysis due to limited budget. Simple haematology parameters may help identifying the carriers. Furthermore, the low knowledge and the anxiety of the participants hindered the willingness to screen their thalassaemia carrier status. As Indonesia is a multi-ethnic population country, a holistic approach to education programs in thalassaemia need to be considered for an optimal thalassaemia carrier screening. Campaign and education for community carrier screening, especially among extended family members of identified carrier individuals, need to be increased, towards zero thalassaemia. Further qualitative study needs to be explored to know the reasons for unwillingness for thalassaemia carrier screening.

Keywords: Thalassaemia carrier screening, extended family, Indonesia

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Acknowledgement

This study was funded by Academic Leadership Grant (ALG) 2017 Universitas Padjadjaran Bandung, Indonesia.

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