

Knowledge About Premarital Screening Thalassemia Genetic Counseling Among Students In the Medical Education Program at the Faculty Of Medicine, Sriwijaya University

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Abstract :Thalassemia is a genetic disorder with a high prevalence in Indonesia. Prevention through premarital screening and genetic counseling is essential to reduce the prevalence of this disease. As future medical professionals, medical students are expected to possess adequate knowledge about premarital screening and genetic counseling to support thalassemia prevention. This study aims to identify the knowledge of medical students at the Faculty of Medicine, Universitas Sriwijaya, regarding Premarital Screening and Genetic Counseling for Thalassemia. This research is a descriptive observational study with a cross-sectional design. The sample consisted of medical students from the Faculty of Medicine, Universitas Sriwijaya, who met the inclusion criteria and were selected using stratified random sampling. The research instrument was a Knowledge Questionnaire on Premarital Screening and Genetic Counseling for Thalassemia. The findings showed that 74.5% of students had adequate knowledge. Adequate knowledge was found among 2021 (80.2%), 2022 (70.9%), and 2023 (72.4%) cohorts. Female students showed a higher percentage (79.9%) compared to males (61.3%). Students with a family history had 100% adequate knowledge, while those without a family history had 74.3%. Those with interaction history scored 85.7%, compared to 73.9% for those without interaction history. Medical students at the Faculty of Medicine, Universitas Sriwijaya, possess adequate knowledge about Premarital Screening and Genetic Counseling for Thalassemia

1. INTRODUCTION

Thalassemia is a chronic hemolytic disease inherited in an autosomal recessive manner, characterized by a decrease or absence of the formation of one of the globin chains (such as the α or β globin chain) that makes up the human hemoglobin molecule. (Wahidiyat & Permono, 2018) Based on the disturbed globin chain, thalassemia is divided into two main types, namely α thalassemia and β thalassemia. α thalassemia occurs due to reduced synthesis of the α -globin chain, while β thalassemia is caused by reduced synthesis of the β -globin chain (Thalassaemia International Federation, 2023).

According to the World Health Organization (WHO), approximately 80–90 million people, or approximately 7% of the world's population, are affected by thalassemia, with most of them living in developing countries. Indonesia is one of the countries in the Thalassemia Belt, with a relatively high prevalence of cases. According to data from the Indonesian Ministry of Health in 2019, there were 10,531 cases of thalassemia in Indonesia, with an estimated 2,500 babies born each year with this

condition. The highest number of thalassemia carriers is found in Palembang (9%), followed by Makassar (8%), and Java (6–8%). According to national and regional epidemiological data, the carrier prevalence of α -thalassemia in Indonesia is estimated to range from approximately 2–11%, while the prevalence of β -thalassemia carriers is reported to be around 3–10%. Higher carrier rates have been observed in several regions, including Sumatra and Sulawesi, reflecting the geographic distribution of hemoglobinopathies in the country (Reinoso, 2018), (Thalassaemia International Federation, 2023).

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To date, thalassemia treatment has not been able to provide a complete cure and is very expensive because patients require lifelong blood transfusions and iron chelation therapy. Therefore, prevention efforts are the most effective step to reduce the incidence of this disease (Kementerian Kesehatan Republik Indonesia, 2018). One proven effective prevention method is through premarital screening and genetic counseling (Aziz & Abdulla, 2022; Keandre et al., 2021) which aims to help individuals learn their genetic status, establish an early diagnosis, and prevent the transmission or inheritance of genetic diseases such as thalassemia to their offspring (Balci, 2014; Shen et al., 2018).

Several international studies have shown that public awareness of premarital screening and genetic counseling remains low. Studies in Qatar (Bener et al., 2019) and Jeddah indicate that although the public has shown acceptance of these programs, knowledge and awareness remain limited (Ibrahim et al., 2013). In Indonesia, research on medical students in Banyumas Regency, Padjadjaran University, and Muhammadiyah University of Semarang showed that students generally have good knowledge and attitudes toward genetic screening and premarital counseling, but their in-depth understanding of genetic counseling and post-test decision-making still needs to be improved (Keandre et al., 2021).

Given these conditions, increasing knowledge and awareness regarding premarital screening and genetic counseling is crucial, especially for medical students as future healthcare professionals. Medical students are expected to become educational agents, providing information to the public about the importance of early detection and prevention of genetic diseases such as thalassemia.

Palembang City itself is one of the areas with a high frequency of thalassemia genes in Indonesia. (Kementerian Kesehatan Republik Indonesia, 2018). Therefore, research is needed to assess the extent of medical students' knowledge regarding premarital screening and genetic counseling for thalassemia, as a basis for developing future education and prevention programs. Based on this, this study aims to determine the level of knowledge of students in the Medical Education Study Program, Faculty of Medicine, Sriwijaya University regarding premarital screening and genetic counseling for thalassemia.

2. METHODS

This study is a descriptive observational study with a cross-sectional design that aims to determine the level of knowledge of students in the Medical Education Study Program, Faculty of Medicine,

Sriwijaya University regarding premarital screening and genetic counseling for thalassemia. The study was conducted online by filling out a Google form in the Medical Education Study Program, Faculty of Medicine, Sriwijaya University. The sample size was determined using the Slovin formula with a 5% error rate from a total population of 721 students, resulting in a minimum of 258 respondents. The sampling technique used stratified random sampling based on class strata, namely 86 students from the class of 2021, 86 students from the class of 2022, and 87 students from the class of 2023. The inclusion criteria for this study were students from the class of 2021-2023 who were willing to participate and completed the questionnaire completely. Data were collected using a knowledge questionnaire about premarital screening and genetic counseling for thalassemia consisting of 8 questions. The questionnaire has been tested for validity and with a calculated r value of > 0.361 and reliability with a Cronbach's Alpha of 0.777. The level of students' knowledge about premarital screening and genetic counseling for thalassemia was categorized as adequate (≥ 7 correct answers) and inadequate (<7 correct answers). Data were analyzed using narrative description.

3. RESULTS

3.1. Sociodemographic characteristics

Table 1. Distribution of Sociodemographic Characteristics of Medical Students FK Unsri.

Sociodemographic Characteristics of Responden		(n)	(%)
Year of Enrollment	2021	86	33.2
	2022	86	33.2
	2023	87	33.6
Gender	Male	75	29.0
	Female	184	71.0
Family history of Thalassemia	Yes	2	0.8
	No	257	99.2
History of interaction with patients	Ever interacted	14	5.4
	Never interacted	245	94.6

Table 1 shows data from 259 respondents from the Sriwijaya University PSPD program, with a nearly even distribution of students from the 2021, 2022, and 2023 intakes, at 33.2%, 33.2%, and 33.6%, respectively. Female students were more dominant than male students (71%). Only two students (0.8%) had a family history of thalassemia, and 14 (5.4%) had interacted with thalassemia patients.

3.2 Distribution Knowledge of Medical Students

Table 2. Distribution Knowledge of Medical Students of the Faculty of Medicine, Unsri.

Knowledge of Medical Students Universitas Sriwijaya regarding Premarital Screening and Genetic Counseling for Thalassemia.	(n)	(%)
Adequate	193	74.5
Inadequate	64	25.5
Total	257	100

Table 2 above shows that there are 193 students (74.5%) who have adequate knowledge of Premarital Screening and Thalassemia Genetic Counseling.

3.3. Distribution of scores per item of the Knowledge Questionnaire on Premarital Screening and Genetic Counseling for Thalassemia

Table 3. Results of the distribution of scores per item of the Knowledge Questionnaire on Premarital Screening and Genetic Counseling for Thalassemia.

Based on table 3. it was found that 258 students (99.6%) knew that premarital thalassemia screening aims to detect thalassemia in individuals before marriage. In the second point, 241 students (93.1%) knew that premarital thalassemia screening is carried out using HB electrophoresis. In the third point, 152 students (58.7%) knew that a positive result obtained from premarital thalassemia screening does not necessarily indicate that a person has thalassemia. In the fourth point, 255 students (98.5%) knew that someone who gets a positive result on premarital thalassemia screening is recommended to consult a genetic counselor.

In the fifth item, 229 students (88.4%) knew that premarital screening for thalassemia can be performed on all families, whether they have a history of thalassemia or not. In the sixth item, 257 students (99.2%) knew that thalassemia genetic counseling is the process of providing information and education about thalassemia. Furthermore, in the seventh item, 255 students (98.5%) knew that thalassemia genetic counseling helps couples make decisions regarding pregnancy and reproduction. Then, in the eighth item, 186 students (71.8%) knew that thalassemia genetic

counseling cannot change the genetic risk of thalassemia.

4. DISCUSSION

Based on the results of this study, of the 259 respondents, PSPD students from the Faculty of Medicine, Unsri, it was found that the majority of

Knowledge Statement	Response	n*	%
Premarital screening aims to detect the presence of thalassemia before marriage	Correct	258	99.6
	Incorrect	1	0.4
Premarital screening for thalassemia can be performed using hemoglobin electrophoresis	Correct	241	93.1
	Incorrect	18	6.9
Premarital screening with a positive result indicates that a person is definitely thalassemia	Correct	152	58.7
	Incorrect	107	41.3
Individual with a positive screening result be referred genetic counseling	Correct	255	98.5
	Incorrect	4	1.5
Premarital screening can only be performed in couples with a family history of thalassemia	Correct	229	88.4
	Incorrect	30	11.6
Genetic counseling provides information about thalassemia and its genetic risks	Correct	257	99.2
	Incorrect	2	0.8
Genetic counseling helps couples make decisions about pregnancy and reproduction	Correct	255	98.5
	Incorrect	4	1.5
Genetic counseling cannot change the genetic risk of thalassemia	Correct	186	71.8
	Incorrect	73	28.2
		*Total n = 259	

students, 74.5%, had good knowledge. This is because PSPD students from the Faculty of Medicine, Unsri, have received training on genetic screening and counseling, as well as thalassemia, since the first semester, specifically in the basic block, namely biology. This is in line with research conducted by Keandre et al. in 2021, conducted on medical and health students in Banyumas, which showed that of the 379 respondents, 75.46% had good knowledge of premarital screening (Keandre et al., 2021). A strong foundation of knowledge is essential for delivering optimal medical services, and this foundation is established early during medical education. Medical students who understand thalassemia well will be able to provide optimal education, treatment, and prevention in the future when they become doctors. Extensive knowledge will support doctors to be more

empathetic, both in establishing a diagnosis and implementing thalassemia prevention efforts (Flores et al., 2022; Wolyniak et al., 2015).

The results of this study indicate that more than half of the respondents (74.5%) have adequate knowledge of premarital screening and genetic counseling for thalassemia. This indicates that most students in the Medical Education Program, Faculty of Medicine, Sriwijaya University, have adequate knowledge of premarital screening and genetic counseling for thalassemia. The high proportion of respondents (99.6%) who understood the purpose of premarital screening indicates a strong awareness of thalassemia prevention strategies among students. This finding may reflect early exposure to genetic education in the medical curriculum. Adequate knowledge of screening is essential because it supports preventive efforts and informed reproductive decision-making. Through this premarital screening, couples can learn about the risk of genetic diseases or disorders, such as thalassemia, so they can take preventive measures to help reduce the risk of congenital abnormalities in their offspring (Balci, 2014; Shen et al., 2018).

The majority of respondents (93.1%) correctly understood the screening method for thalassemia, indicating a good level of knowledge regarding diagnostic procedures. This finding suggests that students are familiar with the basic principles of laboratory screening, likely due to their exposure to clinical pathology and hematology during medical training. (Kementerian Kesehatan Republik Indonesia, 2018; Saxena, 2019).

More than half of respondents (58.7%) correctly recognized that a positive screening result does not necessarily indicate a diagnosis of thalassemia, while a small proportion (41.3%) still misunderstood this concept. This finding suggests that some students have difficulty distinguishing between thalassemia disease and carrier status. Strengthening educational components related to screening interpretation and genetic counseling may help reduce this misconception. Even when all these tests show positive results in one individual, carrier status can only be confirmed after testing the partner. Given that thalassemia is a genetic disorder with an autosomal recessive inheritance pattern, which is why genetic counseling is a crucial component in this process, especially if both partners are identified as carriers. This complex and systematic diagnostic process indicates that a positive screening result in thalassemia screening is only the first step in the thalassemia diagnostic algorithm. This comprehensive and stepwise diagnostic approach is important to avoid misdiagnosis and ensure appropriate treatment for individuals with thalassemia and carriers (Kementerian Kesehatan Republik Indonesia, 2018; Langlois et al.,

2008).

In contrast, in question three, many students (41.3%) still answered incorrectly regarding the positive results obtained by a person and the indications of thalassemia in that person. This is because some students still do not understand the difference between thalassemia as a disease (thalassemia major or intermedia) and as a carrier (thalassemia minor). Screening methods, including HbA₂ measurement using high-performance liquid chromatography (HPLC), are widely used to identify individuals who may be carriers of thalassemia. However, these tests are intended for screening purposes and do not establish a definitive diagnosis. Confirmatory testing, particularly molecular or DNA analysis, is required to accurately determine the presence of thalassemia mutations (Mensah & Sheth, 2021). According to the Indonesian National Standards for Medical Professional Education (SNPPDI), medical curricula are expected to include comprehensive training in genetic disease prevention and diagnosis. Curricula designed in accordance with SNPPDI must integrate promotive and preventive approaches, including the ability of physicians to understand and explain genetic screening results to patients and the public. However, the educational focus, which tends to be more clinical, often results in a less emphasis on understanding the interpretation of genetic screening results (Konsil Kedokteran Indonesia, 2019).

Most respondents (88.4%) correctly understood that premarital screening is recommended for the general population, not only for individuals with a family history of thalassemia. However, a small proportion of students (11.6%) still believed that screening is limited to high-risk groups. This misconception may result from educational emphasis on family history as a risk factor (Alswaidi & O'Brien, 2009; Kementerian Kesehatan Republik Indonesia, 2018). Population-based screening programs have been shown to effectively reduce the incidence of thalassemia in several countries, highlighting the importance of promoting universal screening awareness. This population-based premarital thalassemia screening program is very relevant to be implemented in various regions, especially in areas with a high prevalence of thalassemia, such as the city of Palembang. Considering that Palembang is one of the areas included in the Thalassemia belt, with a fairly high frequency of thalassemia genes, this program is very important to prevent the spread of the disease (Alswaidi & O'Brien, 2009; Kementerian Kesehatan Republik Indonesia, 2018). However, there are some students (11.6%) who are not aware that premarital screening for thalassemia is recommended for everyone, not just those with a family history of thalassemia, this is because the educational approach

is still focused on groups with high risk factors. This emphasis on individuals with a family history creates the false perception that only certain groups need to be aware of thalassemia. In fact, thalassemia is an autosomal recessive genetic disorder that can be passed down by carrier partners without a clear family history (Utami & Kusumaningrum, 2020).

The Indonesian National Standards for Medical Professional Education (SNPPDI) states that physicians are expected to possess competency in promotive and preventive efforts as part of community medicine. Physicians are not only responsible for handling clinical cases but also for providing public health education based on the prevention of genetic diseases, including thalassemia. In this context, medical students need to understand the importance of premarital screening for the general population, as its primary goal is to detect carriers of the genetic trait and prevent the birth of children with thalassemia major, rather than simply diagnosing a clinical disease. SNPPDI-based education also emphasizes that medical competency encompasses a community-based approach. Students must understand that preventing genetic diseases requires equitable education, regardless of family history. The lack of implementation of relevant case-based learning in the curriculum may contribute to students' weak understanding of the importance of premarital screening for everyone (Konsil Kedokteran Indonesia, 2019).

Most respondents demonstrated good knowledge of genetic counseling, including its role in providing information about genetic risks (99.2%) and supporting reproductive decision-making (98.5%). However, a notable proportion of students (28.2%) still believed that genetic counseling could change genetic risk. This finding indicates a misunderstanding of the fundamental purpose of genetic counseling, which focuses on risk communication rather than risk modification. Genetic counseling is a communication process that provides individuals or couples with information about genetic risks and inheritance patterns. In the context of thalassemia, it helps individuals understand the risk of passing the disease to their children, particularly when both partners are carriers. Genetic counseling also supports informed decision-making regarding reproductive options through a nondirective approach that respects patients' values and preferences. (Uhlmann et al., 2011).

However, in the eighth point, many students (28.2%) still do not understand that genetic counseling cannot change the genetic risk of thalassemia. This could be due to several factors, such as a lack of emphasis on basic concepts in genetic education during medical studies, limited clinical exposure to genetic cases, or misperceptions about the purpose of genetic counseling. Genetic counseling is actually

intended to provide accurate information about genetic risk, reproductive options, and emotional support, not to modify or eliminate the genetic risk itself (Keandre et al., 2021).

5. CONCLUSION

Most students in the Medical Education Study Program, Faculty of Medicine, Sriwijaya University demonstrated an adequate level of knowledge regarding thalassemia. Adequate knowledge was observed across different cohorts, genders, and backgrounds, including students with and without a family history of thalassemia or prior interaction with thalassemia patients. These findings indicate that students generally possess sufficient knowledge; however, continuous educational efforts remain important to further strengthen understanding of thalassemia and its prevention.

REFERENCES

- Alswaidi, F. M., & O'Brien, S. J. (2009). Premarital screening programmes for haemoglobinopathies, HIV and hepatitis viruses: review and factors affecting their success. *Journal of Medical Screening*, *16*(1), 22–28. <https://doi.org/10.1258/jms.2008.008029>
- Aziz, N., & Abdulla, S. (2022). Attitudes of Couples attending Family Counselling Clinic in Sulaimani City towards the Premarital Screening and Genetic Counseling Programme. *Erbil Journal of Nursing and Midwifery*, *4*(2), 96–104. <https://doi.org/10.15218/ejnm.2021.11>
- Balci, Y. I. (2014). Thalassemia Premarital Screening Program: Public View, What has been Done, What Needs to be Done? *International Journal of Hematology and Oncology*, 247–252. <https://doi.org/10.4999/uhod.14569>
- Bener, A., Al-Mulla, M., & Clarke, A. (2019). Premarital screening and genetic counseling program: Studies from an endogamous population. *International Journal of Applied and Basic Medical Research*, *9*(1), 20. https://doi.org/10.4103/ijabmr.IJABMR_42_18
- Flores, A., Burgos, S., & Abarca-Barriga, H. (2022). Knowledge level of medical students and physicians about rare diseases in Lima, Peru.

- Intractable & Rare Diseases Research*, 11(4), 180–188. <https://doi.org/10.5582/irdr.2022.01079>
- Hokahay, D. C. (2021). *Skrining Pembawa Gen Thalassemia Beta pada Mahasiswa Fakultas Kedokteran Sebuah Kedokteran di Bandung*. Universitas Kristen Maranatha.
- Ibrahim, N. K., Bashawri, J., Al Bar, H., Al Ahmadi, J., Al Bar, A., Qadi, M., Milaat, W., & Fedaa, H. (2013). Premarital Screening and Genetic Counseling program: Knowledge, attitude, and satisfaction of attendees of governmental outpatient clinics in Jeddah. *Journal of Infection and Public Health*, 6(1), 41–54. <https://doi.org/10.1016/j.jiph.2012.05.001>
- Keandre, N., Rujito, L., & Munfiah, S. (2021). Pengetahuan, Sikap dan Perilaku Mahasiswa Kedokteran dan Kesehatan terhadap Skrining Genetik dan Premarital di Kabupaten Banyumas. *Medical and Health Journal*, 1(1), 22. <https://doi.org/10.20884/1.mhj.2021.1.1.4670>
- Kementerian Kesehatan Republik Indonesia. (2022). *Keputusan Menteri Kesehatan Republik Indonesia tentang pelayanan skrining kesehatan*. Jakarta: Kementerian Kesehatan Republik Indonesia.
- Konsil Kedokteran Indonesia. (2019). *Standar Nasional Pendidikan Profesi Dokter Indonesia*.
- Langlois, S., Ford, J. C., Chitayat, D., CCMG PRENATAL DIAGNOSIS COMMITTEE, & SOGC GENETICS COMMITTEE. (2008). Carrier screening for thalassemia and hemoglobinopathies in Canada. *Journal of Obstetrics and Gynaecology Canada : JOGC = Journal d'obstetrique et Gynecologie Du Canada : JOGC*, 30(10), 950–959. [https://doi.org/10.1016/S1701-2163\(16\)32975-9](https://doi.org/10.1016/S1701-2163(16)32975-9)
- Mensah, C., & Sheth, S. (2021). Optimal strategies for carrier screening and prenatal diagnosis of α - and β -thalassemia. *Hematology. American Society of Hematology. Education Program*, 2021(1), 607–613. <https://doi.org/10.1182/hematology.2021000296>
- Reinoso, J. (2018). KEPUTUSAN MENTERI KESEHATAN REPUBLIK INDONESIA NOMOR HK.01.07/MENKES/1/2018 TENTANG PEDOMAN NASIONAL PELAYANAN KEDOKTERAN TATA LAKSANA THALASEMIA. *Journal of Materials Processing Technology*, 1(1), 1–8. <http://dx.doi.org/10.1016/j.cirp.2016.06.001><http://dx.doi.org/10.1016/j.powtec.2016.12.055><https://doi.org/10.1016/j.ijfatigue.2019.02.006><https://doi.org/10.1016/j.matlet.2019.04.024><https://doi.org/10.1016/j.matlet.2019.12.7252>
- Saxena, S. (2019). *Screening of thalassemia and other hemoglobinopathies in blood donors by capillary hemoglobin electrophoresis system*. *International Journal of Research in Medical Sciences*, 7(11), 4210–4215. <https://doi.org/10.18203/2320-6012.ijrms20194747>
- Shen, Y., Qiu, X., Gui, B., He, S., Huang, H., Xue, J., Xu, X., Zhang, X., & He, L. (2018). Implementing comprehensive genetic carrier screening in China—Harnessing the power of genomic medicine for the effective prevention/management of birth defects and rare genetic diseases in China. *Pediatric Investigation*, 2(1), 30–36. <https://doi.org/10.1002/ped4.12021>
- Thalassaemia International Federation. (2023). *Guidelines for the Management of Transfusion Dependent Thalassaemia (TDT)* (Alan Cohen, Ali Taher, John Porter, Maria-Domenica Cappellini, & Vip Viprakasit, Eds.; 4th ed.). Thalassaemia International Federation.
- Uhlmann, W. R. ., Schuette, J. L. ., & Yashar, Beverly. (2011). *A Guide to Genetic Counseling (2nd ed.)*. Hoboken, NJ: Wiley-Blackwell.
- Utami, S. T., & Kusumaningrum, N. S. D. (2020). Knowledge of Premarital Couples Regarding Premarital Screening Thalassemia. *Jurnal Keperawatan*, 11(2), 180–187. <https://doi.org/10.22219/jk.v11i2.10740>
- Wahidiyat, P., & Permono, B. (2018). *BUKU AJAR Hematologi Onkologi Anak*, (W. Y. N. E, M. S, S. B, & U. I, Eds.). Jakarta: Ikatan Dokter Anak Indonesia.
- Wolyniak, M. J., Bemis, L. T., & Prunuske, A. J. (2015). Improving medical students' knowledge of genetic disease: a review of current and emerging pedagogical practices. *Advances in Medical Education and Practice*, 6, 597–607. <https://doi.org/10.2147/AMEP.S73644>