



## Original Research

### Premarital screening and probabilities of genetic disease in premarital screening and probability of genetic diseases in couples preparing for marriage

Cut Muthiadin<sup>1</sup>

<sup>1</sup>Department of Biology, Universitas Islam Negeri Alauddin, Romangpolong Gowa, 92113

Corresponding author(s): [cutmuthiadin@uin-alauddin.ac.id](mailto:cutmuthiadin@uin-alauddin.ac.id); +62 81355279069

Article Info	Abstract
<p><i>Article history:</i> Received 5 September 2023 Accepted 17 November 2023</p> <p><i>Keywords:</i> premarital, genetic disease, screening, genetic counseling, hereditary</p> <p><i>How to cite:</i> Muthiadin.C. 2023. Premarital screening and probabilities of genetic disease in premarital screening and probability of genetic diseases in couples preparing for marriage. <i>Tropical Genetics</i>, 3(1): 20-25.</p>	<p>Genetic diseases are known to be difficult and could not be avoided. The disease is obtained from mutations and changes in genetic traits that are passed on from one or both genes from parent to child. Genes carrying traits or diseases can be dominant and recessive, their location can be on the autosomal chromosome or sex chromosome X or Y. This study was conducted on premarital female respondents with defined inclusion criteria. Data were collected through a google form questionnaire. A total of 110 respondents with several parameters of genetic counselling. The result was obtained that all respondents have known that certain diseases can be inherited. However, from all respondents, it was found that they had low knowledge of genetic counselling and premarital screening. From the respondent's data, it was also found that only 17% of respondents knew the history of the disease of the prospective partner and their family. In this case, it can be said that respondents might be carry the sick gene which means the probability that their child will carry the same gene is 50% normal: 50% sick. By screening, it might be predicted that the couple will be able to find out the potential diseases that will be passed on to their offspring later. Latest 48% decided to cancel and 26% said they did not agree and stated to continue the plan of marriage. Lack of knowledge about risk of genetic disorders need education about premarital screening to increase this information and premarital screening to the public.</p>

## Introduction

Genetic inheritance is the transmission of traits or genetic information from one generation (parents) to the next generation (children). The principles of modern genes and heredity were first put forward by Gregor Mendel, who is known for his crossover theory for independent genes. The theory states that the genes of the child are a combination (cross) of the genes of both parents. Genes are defined as intervals along DNA molecules. Most genes carry the information needed to make proteins ([McCarrey, 2018](#)).

The probability of the emergence of traits in several genes of one individual is determined by the dominance of the gene or allele that is inherited by one parent. The dominant trait beats the recessive gene and eventually a trait emerges in one individual, this is called an epistatic event. These traits that we can directly observe physical characteristics include, ear lobes, curly hair, dimples commonly known as phenotypic characteristics.

Humans have cells with 46 chromosomes, consisting of 2 sex chromosomes, and 22 pairs of autosomal chromosomes. Chromosome formulation in males is “46 AA, XY” and chromosomes in females is “46 AA, XX”. Chromosomes consist of very long combinations of proteins and DNA molecules. The failure to separate chromosomes during cell division events can cause genetic disorders such as: Down syndrome, Turner syndrome, Klinefelter syndrome (Khalifah, 2021).

If there is a defective gene in the chromosome, the chances of the emergence of a genetic disorder can be inherited, depending on the type of gene that is inherited is recessive or dominant. Diseases caused by a dominant gene, usually can appear even if only one copy of the gene with a DNA mutation from one of the parents. For example, if one parent has the disease, it means that each child has a 50 percent chance of developing the disease. Whereas diseases caused by recessive genes are less likely to cause serious harm to a person. This is because new recessive genes cause health problems if they are inherited from both parents or both copies of the gene must undergo DNA mutations. For example, if both parents have one copy of the mutated gene, then the child has a 25 percent chance of developing the disease. In this case, the parents are referred to as carriers ([Charlesworth, et al., 2021](#)).

Genetic disorders can also be caused by DNA mutations contained in a person's genes. This condition can affect how these genes work. Mutation events are initially obtained at the time of DNA replication where changes in the DNA sequence are far from normal ([Ronfort, 2021](#)). This implies that there is a common normal allele in the population and then a mutation turns it into a rare and abnormal variant. Genetic variations are produced continuously by this mutation process as natural and harmless. However, most of these mutations can be fatal to the organism.

Patients with classical chromosomal disorders have physical phenotypes that are different from normal people, are likely to get various health problems, lower intelligence than normal people and social problems with their environment and even with their families. Treatment for patients as early as possible in the womb, early birth, adequate family support, physical and vocational training will be able to improve the quality of life of sufferers of the syndrome. Families of sufferers in this case parents really need complete information about this disease and its inheritance, for that genetic counselling with professional genetic counsellors is needed. ([Genetic Alliance, 2009](#)).

To avoid the chance of the emergence of genetic disorders from marriage, premarital screening can be done. Premarital screening is a testing program to diagnose and treat previously unknown disorders or disorders, preventing the risk of disease transmission to partners or offspring. Through premarital screening, the individual's overall body condition can be identified so that it can prevent or minimize the occurrence of congenital abnormalities such as thalassemia. In addition, it is necessary to have premarital counseling for couples as the next stage of consultation so that they are able to know the risks during pregnancy and treatment when their child suffers from thalassemia, as well as alternative treatments. ([Utami & Kusumaningrum, 2020](#)). Based on this background, researchers are interested in conducting a survey in the form of a description of the knowledge of premarital couples about premarital screening as an initial study.

## Methods

### 1. Subjects

Respondents are 110 women, aged 20-40 years, currently in the process of planning a marriage. The exclusion criteria in this study were women from partners on H-7 marriage when selecting respondents.

### 2. Methods

This research is a type of quantitative descriptive survey research. The survey taking technique used was purposive sampling according to the inclusion criteria, namely women aged 20-40 years, currently in the process of planning a wedding. The exclusion criteria in this study were women from partners on H-7 marriage when selecting respondents. The

number of samples is 110 women. The questionnaire was uploaded to a google form, then distributed via Whatsapp messages to several communities.

### Results and Discussion

A total of 110 women who filled out the form were registering their marriage together who were willing to be respondents in this study.

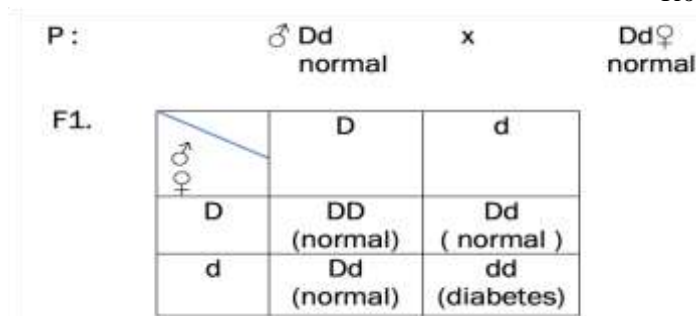
**Table 1. Demographics of Respondents.**

Characteristics of respondents	f	Percentage (%)
Age		
20 – 26 years	56	51
27 - 33 years old	45	41
34 – 60 years	9	8
Kinship relationship with potential partner		
Yes	42	38
Not	68	62
Spouse's family history of disease: DM, Thalassemia, etc		
Yes	19	
Not	91	17
History of hereditary screening		83
Yes	0	
Not	110	0

Table 1 shows that the majority of premarital couples (51%) are in the age range of 20-26 years. As many as 38% of respondents stated that they had a family relationship, while 62% did not have a family relationship. 17% had a family history of hereditary disease and none had a screening test (100%). Couples who still have two generations of kinship can increase the percentage of the risk of having thalassemia offspring (Hamamy, 2012). Based on a previous study by Saeed & Piracha (2016) which stated that kinship marriage should be prevented because it can increase the risk of thalassemia incidence in Pakistan. Furthermore, the study stated that culture and the role of parents in deciding the marriage of their children were still very strong,

Endogamy marriage in the medical dictionary, means as a process of reproduction through the marriage of people who are very close (relatives). Endogamy marriage or can be called inbreeding (inbreeding) involves the process of sexual reproduction in the form of fertilization of individual gametes with close family ties or Gametes of the same individual or genotype. This process tends to cause homozygosity and significant impairment of the expression of harmful alleles and can also reduce the level of genetic variation between offspring ([Khafizoh, 2017](#)).

Several disease disorders, including type II diabetes mellitus, in addition to the pattern of inheritance from a single autosomal recessive allele, can also be categorized into multifactorial effects or commonly known as environmental influences. DM is known to be inherited following Mendelian laws, but only partially contributes to a trait and is therefore more difficult to trace. It can be seen from the diagram (Figure 1) that heterozygous pairs as Normal (carriers) have the potential to have children with DM. So if a person is known to have diabetes, while both parents are normal, it can be ascertained that both parents are heterozygous. This inheritance pattern is called single allele inheritance by recessive disorders, the allele for DM is genotype d.



**Figure 1.** Marriage diagram of heterozygous couples (carriers of the DM gene).

Information obtained by the Indonesian people about premarital screening is still limited. There are not many institutions or public health service facilities that hold health education activities about premarital screening. Education to the public about premarital screening as a form of preventing genetic disorders can be started from the education bench. Education is a means of education that can improve the quality of a nation. Through education, it is hoped that community knowledge will develop and be followed by an increase in positive attitudes possessed by the community (Bener, Al-Mulla, & Clarke, 2019; Calzone et al., 2010 ; [Memish & Saeedi, 2011](#); [Mirza et al., 2013](#)).

**Table 2.** Description of respondents' knowledge regarding the inheritance of genetic disorders.

Knowledge of genetic disorders	f	Percentage (%)
Good Category Knowledge	18	16
Poor Category Knowledge	92	84

Table 2 describes about 84% of respondents have less knowledge about the description of inheritance of genetic disorders. In this case the types of diseases that are classified as hereditary diseases, risks and effects. More specific research conducted by ([Utami & Kusumaningrum, 2020](#)).

There are several factors that influence the knowledge of premarital couples about thalassemia, namely thalassemia disease is a disease that is still commonly heard among the public, besides that the disease is also rarely heard in the mass media. Not many people know about the disease and its prevention (Thirafi, 2017). The lack of information obtained by the public about thalassemia disease makes public knowledge related to thalassemia disease low. Information needed by premarital couples about thalassemia can be obtained through friends, mass media, educational institutions, family, health care facilities, and so on. The results of the study show that most couples get information from friends and news or media. Media has an important role in disseminating information. so that the role of the media as a provider of information for the community is the right means. The media can be a source of information for people in today's modernization era.

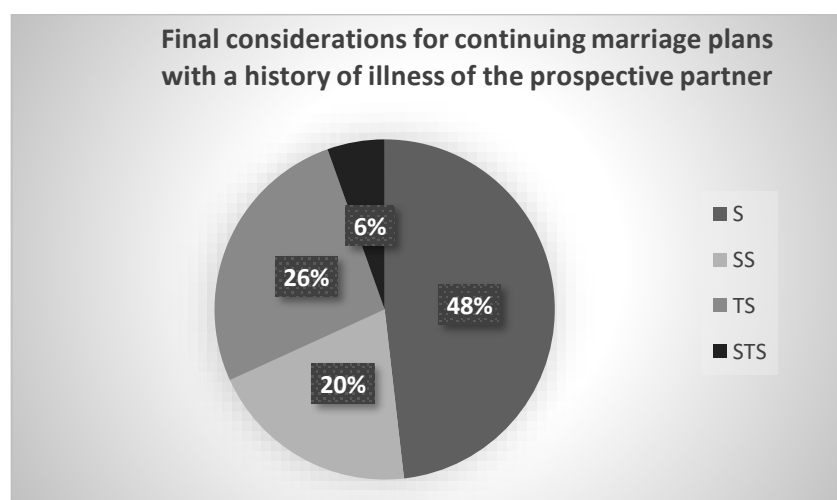
Research by ([Ibrahim, et al., 2011](#)) conducted to assess the knowledge and attitudes of unmarried female students at King Abdul-Aziz University (KAU) towards the premarital screening program, to determine predictors of high student knowledge scores and to increase their knowledge of PMS through the implementation of a national education campaign. Intervention strategies with mass communication campaigns were carried out. The educational program consists of 3 basic concepts; Pre-test, educational campaign and Post-test. As a result, the mean score of students' knowledge was  $9.85 \pm 5.36$  in the pre-test and increased to  $18.45 \pm 4.96$  in the post-test. This method can be a reference for future research.

Premarital counselling is used as an effort to provide knowledge and motivation for premarital couples towards married life. Through marriage counselling, the personality of each individual can be known so that the couple knows the condition of their partner, such as in terms of communicating ([Ibrahim, et al., 2011](#)). The purpose of premarital counselling is to assist couples in conducting health screening tests, providing knowledge regarding conditions and the risk of decreasing genetic diseases, one of which is thalassemia. In addition, counselling also facilitates couples to decide on

their health condition, the continuation of their marriage and reproduction ([Memish & Saeedi, 2011](#); [Mirza et al., 2013](#); [Mudiyanse & Senanayake, 2015](#)).

Marriage with other family relatives (to the extent of cousin II – the same great grandparents) is called a consanguineous marriage. Population studies show that children from inbreeding have a greater risk of developing certain genetic diseases. Especially those whose inheritance is autosomal recessive. In this type of decline, the carrier will not show any symptoms. Meanwhile, because people in the same family have the same proportion of genetic material, husbands and wives who are related to each other are also at risk of carrying the same genetic material. Children resulting from marriages (blood or not) where both parents are carriers ([Aziz et al., 2019](#)).

The amazing thing that was found in this study was obtained from additional questions in the form of consideration of continuing the relationship between marriage plans and a history of illness of the prospective partner, namely 48% decided to cancel and 26% said they did not agree and stated to continue (Figure 2).



**Figure 1.** Diagram of final considerations for continuing marriage plans with a history of illness of the prospective partner.

The limitation of this research is that the demographics of the respondents do not reach education and income, so they are unable to identify the condition of the respondents with the decisions mentioned above. This is a strong reason why research uses women as subjects in research as a strong bet and of course with the consideration that women can be used as educational media regarding potential marriages with a history of hereditary diseases and marriage of relatives.

Providing information and motivation through other people, mass media, schools and public health service facilities can increase public awareness and participation on the importance of premarital screening. Supported by technological sophistication and easy access, the community should be able to use it well to participate in disseminating positive information about the importance of premarital screening to others. The provision of supporting public information is expected to increase public awareness of the importance of premarital screening. People have the awareness to be able to share their knowledge with other communities (Sibbald et al., 2016).

## Conclusion

The conclusion in this study is that in general, the knowledge of premarital couples (women domiciled in Makassar City) regarding the risk of inheriting genetic disorders is still low. Thus, education about premarital screening to increase this information and premarital screening to the public is very necessary.

## Acknowledgments

Thanks to Directorate of Islamic Higher Education, funding to our study and presented in THE 20TH ANNUAL INTERNATIONAL CONFERENCE ON ISLAMIC STUDIES (AICIS) 2021.

## References

- Aziz, IR, Muthiadin, C., & Nurfitriah, AN (2019). Piebaldism in To Balo, South Sulawesi: Brief Report and Literature Review. *Biota*, 12(2).<https://doi.org/10.20414/JB.V12I2.213>
- Chial, H. (2008). Mendelian genetics: Patterns of inheritance and single-gene.
- Charlesworth, D., Morgan, MTΔ, & Charlesworth, B. (2021). The effect of linkage and population size on inbreeding depression due to mutational load. *genetics. Res., Camb*, 59, 49–61. <https://doi.org/10.1017/S0016672300030160>
- Genetic Alliance. (2009). Understanding Genetics: A New York, Mid-Atlantic Guide for Patients and Health Professionals Genetic Alliance, The New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services Genetic Alliance Posted in the Resource. <http://www.resourcepository.org/documents/1247/understandinggenetics:anewyork,mid-atlanticguideforpatientsandhealthprofessionals/>
- Hamamy, H. (2012). Consanguineous marriages. *Journal of Community Genetics*, 3(3), 185-192.
- Ibrahim, NKR., et al. (2011). An educational program about premarital screening for unmarried female students in King Abdul-Aziz University, Jeddah | Elsevier Enhanced Reader. <https://reader.elsevier.com/reader/sd/pii/S1876034110000833?token=>
- Irom BS. (2020) Genetic Disorders: A literature review. *Genet Mol Biol Res* Vol No. 4 Iss No. 2: 30.
- Khafizoh, A. (2017). Inbreeding in the Perspective of Islamic Law and Genetics.
- McCarrey, JR (2018). genetics vs. epigenetic inheritance overview. *Encyclopedia of tion*, 434–435.<https://doi.org/10.1016/B978-0-12-801238-3.64509-9>
- Memish, ZA, & Saeedi, MY (2011). Six-year outcome of the national premarital screening and genetic counseling program for sickle cell disease and -thalassemia in Saudi Arabia. *Annals of Saudi medicine*, 31(3), 229-235.
- Mirza, A., Ghani, A., Pal, A., Sami, A., Hannan, S., Ashraf, Z., . . . Fatmi, Z. (2013). Thalassemia and premarital screening: potential for implementation of a screening program among young people in Pakistan. *Hemoglobin*, 37(2), 160-170.
- Mudiyanse, R., & Senanayake, M. (2015). "Safe Marriages" for Thalassemia Prevention: A KAP Survey in Sri Lanka. *Translational Biomedicine*, 6(3), 26. doi:<https://doi.org/10.21767/2172-0479.100026>
- Reich DE, Schaffner SF, Daly MJ, et al. (2002). Human genome sequence variation and the influence of gene history, mutation and recombination. *Nature Genetics* 32: 135–142.
- Ronfort, J. (2021). The mutation load under tetrasomic inheritance and its consequences for the evolution of the selfing rate in autotetraploid species. *genetics. Res., Camb*, 74, 31–42. <https://doi.org/10.1017/S0016672399003845>
- Utami, ST, & Kusumaningrum, NS (2020). Knowledge of premarital couples about premarital screening for thalassemia. *Journal of Nursing*, 11(2).<https://doi.org/10.22219/jk.v11i2.10740>