

PREMARITAL SCREENING AND RELATED GENETIC DISORDERS PROBABILITIES IN COUPLES PLANNING FOR MARRIAGE

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Abstract

It is well recognized that genetic illnesses are challenging and unavoidable. Mutations and alterations in genetic characteristics that are inherited from one or both genes from one parent to the other cause the disease. Both dominant and recessive genes that transmit features or diseases can be found on the autosomal or sex chromosomes X or Y. Premarital female respondents who met specific inclusion criteria participated in this study. Data was gathered using a Google Form survey. 110 people in all with various genetic counseling characteristics. According to the results, every respondent was aware that some illnesses could run in families. Nonetheless, it was shown that none of the respondents knew much about premarital screening and genetic counseling. Only 17% of respondents were aware of the prospective partner's and their family's medical history, according to the respondent data. In this instance, it can be concluded that the respondents may have a sick gene, meaning that there is a 50% chance that their child would have the same gene. It may be anticipated that screening may enable the couple to identify any potential ailments that they may subsequently pass on to their children. As of right now, 26% said they disagreed and declared they will stick with the marriage plan, while 48% decided to cancel. The public has to be educated about premarital screening in order to enhance their awareness of the risk of genetic abnormalities.

Introduction

The transfer of characteristics or genetic data from one generation (parents) to the following generation (children) is known as genetic inheritance. Gregor Mendel, famous for his crossover theory for independent genes, was the first to propose the fundamentals of contemporary genes and heredity. According to the hypothesis, the child's genes are a cross between the genes of both parents. Intervals along DNA molecules are called genes. The majority of genes include the instructions required to produce proteins (McCarrey, 2018).

The dominance of the gene or allele inherited from one parent determines the likelihood that features would develop in many genes of a single individual. An epistatic event occurs when a trait eventually appears in one person as a result of the dominant trait outperforming the recessive gene. Commonly referred to as phenotypic characteristics, these physical attributes that humans may directly perceive include dimples, wavy hair, and ear lobes. Two sex chromosomes and twenty-two pairs of autosomal chromosomes make up the 46 chromosomes found in human cells. In men, the chromosomal formulation is "46 AA, XY," while in females, it is "46 AA, XX." Long sequences of DNA and protein molecules make up chromosomes. Genetic diseases such Turner syndrome, Klinefelter syndrome, and Down syndrome can result from the inability of chromosomes to split during cell division processes (Khalifah, 2021).

Depending on whether the gene is recessive or dominant, diseases caused by a dominant gene can typically manifest even if only one copy of the gene has a DNA mutation from one of the parents. If a gene is defective in the chromosome, the likelihood of a genetic disorder developing can be inherited. One parent having the illness, for instance, indicates that each child has a 50%

probability of getting it. On the other hand, illnesses brought on by recessive genes are less likely to seriously injure a person. This is due to the fact that new recessive genes that are inherited from both parents or that require DNA alterations in both copies result in health issues. For instance, a child has a 25% risk of getting the condition if both parents carry one copy of the mutant gene. The parents in this instance are known as carriers (Charlesworth et al., 2021). DNA mutations in an individual's genes can also result in genetic diseases. The way these genes function may be impacted by this disorder. When DNA replication occurs, mutation events are first produced when the DNA sequence varies significantly from normal (Ronfort, 2021). This suggests that a common normal allele exists in the population, and that a mutation transforms it into an uncommon and aberrant variation. This mutation process continuously produces genetic variants that are safe and natural. The majority of these mutations, nevertheless, have the potential to kill the creature.

In addition to having physical characteristics that differ from those of normal people, patients with classical chromosomal diseases are more likely to experience a variety of health issues, have lower IQs than average people, and experience social issues in their surroundings and even within their family. Early pregnancy treatment, early delivery, proper family support, and physical and vocational training can all help persons with the condition live better lives. Families of patients, in this case the parents, require comprehensive information about the condition and how it is inherited; hence, genetic counseling from qualified genetic counselors is required. (2009, Genetic Alliance).

Premarital screening can be done to reduce the likelihood that genetic abnormalities will develop after marriage. Premarital screening reduces the chance of illness transfer to partners or children by using testing to identify and treat conditions that have not yet been identified. Premarital screening can be used to determine a person's general health in order to prevent or reduce the likelihood of congenital conditions like thalassemia. Additionally, as the following step of consultation, couples must attend premarital counseling so that they can learn about the risks during pregnancy, how to treat their kid if they have thalassemia, and alternative treatments. Kusumaningrum and Umi (2020). Given this context, researchers are interested in performing an initial study that consists of a survey that describes premarital couples' knowledge about premarital screening.

Methods

1. Subjects

110 women between the ages of 20 and 40 who are presently engaged in marriage planning make up the respondents. The exclusion criteria in this study were women from partners on H-7 marriage when selecting respondents.

2. Methods

This study falls under the category of quantitative descriptive surveys. Purposive sampling was the method of survey taking employed in accordance with the inclusion criteria, which were women between the ages of 20 and 40 who were presently engaged in wedding preparation. When choosing respondents for this study, women from partners in H-7 marriages were excluded. There are 110 female samples. After being uploaded to a Google Form, the survey was shared with other communities via WhatsApp conversations.

Results and Discussion

110 women in all who completed the form were willing to participate in this study as respondents and were registering their marriage together.

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	19	
Yes		
Not	91	17
History of hereditary screening	0	83
Yes		
Not	110	0

The majority of premarital couples (51%) are between the ages of 20 and 26, according to Table 1. Up to 38% of those surveyed said they were in a familial relationship, whereas 62% said they were not. A 100% screening test was not performed, and 17% had a family history of inherited disease. According to Hamamy (2012), couples who still have two generations of kinfolk may be more likely to have children with thalassemia. According to a prior study by Saeed & Piracha (2016), familial marriage has to be avoided since it raises the possibility of thalassemia incidence in Pakistan. In addition, the study found that parents continue to play a significant role in determining their children's marriages. In the medical lexicon, endogamy marriage is defined as a process of reproduction through the union of close relatives. The process of sexual reproduction by fertilization of individual gametes with tight family links or gametes of the same individual or genotype is known as endogamy marriage, or inbreeding. This mechanism can lower the degree of genetic variation among offspring and tends to result in homozygosity and severe impairment of the expression of deleterious alleles (Khafizoh, 2017).

In addition to being inherited from a single autosomal recessive allele, a number of disease diseases, such as type II diabetes mellitus, can also be classified as multifactorial effects, or environmental impacts. Although DM is known to be inherited according to Mendelian rules, it is more challenging to identify because it only partially contributes to a trait. Heterozygous partners that are Normal (carriers) have the potential to bear offspring with DM, as shown in the schematic (Figure 1). Therefore, it can be determined that both parents are heterozygous if a person has diabetes and both parents are normal. The allele for DM is genotype d, and this form of heredity is known as single allele inheritance by recessive diseases.

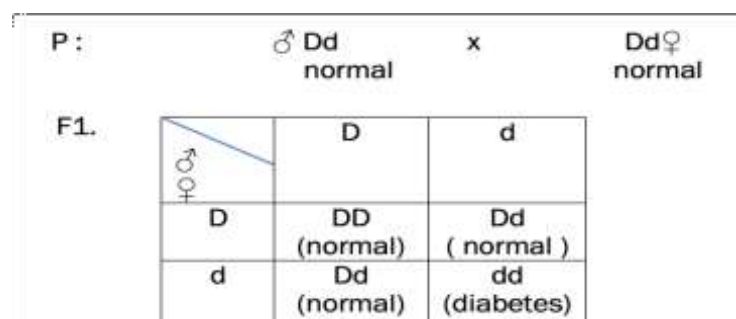


Figure 1. Marriage diagram of heterozygous couples (carriers of the DM gene).

There is relatively little information available to Indonesians on premarital screening. Few public health service agencies or organizations offer health education programs about premarital screening. The education bench is a good place to start educating the public about premarital screening as a way to prevent genetic problems. Education is a tool that can raise a country's

standard of living. The development of community knowledge and a rise in favorable attitudes within the community are anticipated outcomes of education (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

According to Table 2, 84% of respondents knew very little about how genetic illnesses are inherited. In this instance, the risks, consequences, and categories of diseases that fall within the category of inherited disorders. more detailed studies carried out by (Utami & Kusumaningrum, 2020).

Premarital couples' understanding of thalassemia is influenced by a number of factors, including the fact that the condition is still well known in the general public and that it is rarely discussed in the media. Few people are aware of the illness or how to avoid it (Thirafi, 2017). Public awareness of thalassemia disease is low due to the dearth of information available to the general public. Premarital couples can get the information they need about thalassemia from friends, the media, schools, family, medical facilities, and more. The study's findings indicate that the majority of couples obtain their information from friends, the media, and the news. The media plays a significant part in information dissemination. such that the media's function as a communal information source is the appropriate one. In this age of modernization, people can obtain information via the media.

In order to identify predictors of high student knowledge scores, evaluate the attitudes and knowledge of unmarried female students at King Abdul-Aziz University (KAU) regarding the premarital screening program, and increase their understanding of PMS through the implementation of a nationwide education campaign, Ibrahim et al. (2011) conducted research. Mass communication campaigns were used in conjunction with intervention measures. Three fundamental ideas make up the educational program: the pre-test, the instructional campaign, and the post-test. This resulted in a mean knowledge score of 9.85 ± 5.36 on the pre-test and 18.45 ± 4.96 on the post-test. This approach may serve as a model for further study. The purpose of premarital counseling is to educate and inspire premarital couples to pursue marriage. Each person's personality can be discovered through marriage counseling, allowing the couple to understand their partner's communication style, for example (Ibrahim et al., 2011). Premarital counseling is intended to help couples with health screenings, educating them about illnesses and the danger of reducing hereditary diseases, including thalassemia. Additionally, counseling helps couples make decisions about their health condition, Memish and Saeedi (2011), Mirza et al. (2013), and Mudiyanse and Senanayake (2015) discuss their health, marriage, and procreation.

Consanguineous marriage is defined as a union with other family members, including cousin II and the same great grandparents. According to population research, offspring of inbreeding are more likely to have specific hereditary illnesses. particularly those with autosomal recessive inheritance. The carrier will not exhibit any symptoms throughout this kind of decline. In the meantime, husbands and wives who are related to one another run the risk of sharing genetic material because members of the same family share the same percentage of genetic material. offspring from blood or non-blood unions in which both parents are carriers (Aziz et al., 2019). The study's most striking finding came from extra questions about whether or not to continue the relationship between marriage plans and the prospective partner's medical history; 48% of respondents chose to cancel, while 26% disagreed and said they would continue (Figure 2).

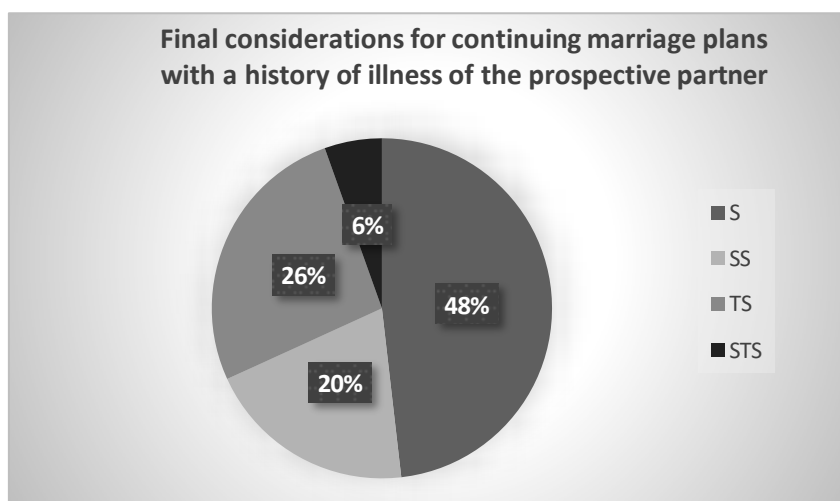


Figure 1. Diagram of final considerations for continuing marriage plans with a history of illness of the prospective partner.

The research's weakness is that the respondents' demographics do not include income and education, making it impossible to determine the respondents' status in relation to the aforementioned actions. In addition to the obvious fact that women can serve as instructional media about prospective marriages with a history of genetic diseases and weddings of relatives, this is a compelling argument for why research employs women as subjects.

Public awareness and involvement about the significance of premarital screening can be raised by disseminating information and encouragement via other individuals, the media, educational institutions, and public health facilities. With the use of advanced technology and convenient access, the community should be able to effectively engage in educating others about the value of premarital screening. It is anticipated that the dissemination of corroborating public information will raise awareness of the significance of premarital screening. Individuals are conscious enough to be able to impart their knowledge to other groups (Sibbald et al., 2016).

Conclusion

According to the study's findings, premarital couples—women who live in Makassar City—generally still don't know much about their risk of inheriting genetic illnesses. Therefore, it is imperative that the public be educated about premarital screening in order to promote awareness of this knowledge.

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