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Preventing Genetic Diseases Through Genetic Counseling to Build a Harmonious Family: a Narrative Review

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Abstract
Indonesia is a country that has complex genetic diversity which can be seen from the diversity of ethnicity, language, and culture that extends from various islands. Seeing this, it can be estimated that Indonesia also has a high number of genetic diseases. The aim of this study is to examine the use of genetic counseling as a preventive measure for genetic diseases in order to form a harmonious family. This study uses library research methods with content analysis techniques. The findings of this study are that genetic counseling is a process of communicating genetic disease problems so that they are conveyed properly to a family. The aim is to provide an understanding of the causes of genetic disease, how to reduce risk in the family, and the selection of optimal actions to overcome the disease. Genetic counseling is very important before marriage and can be part of premarital counseling. This is done so that the offspring produced later, are born in good and quality conditions in order to achieve a harmonious family.

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INTRODUCTION

Indonesia is a country with complex genetic diversity. These facts are reflected in the ethnic, linguistic, and cultural diversity that stretches from island to other island. Migration processes and mixed marriages are two causes of diversity. From the research literature, we can estimate that Indonesia has the potential to have a high rate of genetic diseases. Carriers of thalassemia, a recessive disorder, reach 10% of the population (Wahidiyat et al., 2021; Costrie Ganes Widayanti et al., 2011; Costrie Gones Widayanti & Dewi, 2012). Using the Hardy-Weinberg law (in Rujito, 2018), it can be estimated that the current number of thalassemia sufferers in Indonesia is two million patients, either registered or unregistered.

Other diseases such as down syndrome are also mentioned in one in 700 people (Rujito, 2018). In Indonesia, extrapolated data found about 300,000 people are living with down syndrome. Hemophilia is a blood clotting disorder that has a ratio of one of 10,000 in developing countries (Abdulqader et al., 2020; Laksono et al., 2011; Shah et al., 2020). Cases of Fragile X Syndrome with mental retardation as a feature occur for generations in the Semin, Gunung Kidul (Novianti, 2017; Rujito, 2018). At first glance, the prevalence of rare diseases is very small when viewed case by case. However, when these prevalences are combined, it represents an unusually large number. If all people with rare diseases lived in one country, it would be the 3rd most populous country in the world. If converted to the total population of Indonesia, 12.5 million people live with various rare diseases.

Genetic counseling is one of the interventions to reduce genetic diseases as described above. Genetic counseling is a more comprehensive intervention compared to routine interventions or standard care such as health education or psychoeducation (Mwangi & Mbwayo, 2020). In genetic counseling, there is a recurrence calculation process, carrier screening test, and decision making that can affect the patient's psychosocial status, while other interventions only focus on general information about the disease (Setiawan et al., 2018). Various studies have shown that genetic counseling is used because it can help a person to understand and adapt to medical, psychological, family implications, and genetic contributions to a disease that is not found in other interventions (Setiawan et al., 2018). This can be seen in **Figure 1**. In building a harmonious family, it is very important to know the disease history of the husband and wife so that later if they have a child, it will be a quality child and have a healthy condition, and free from genetic diseases.

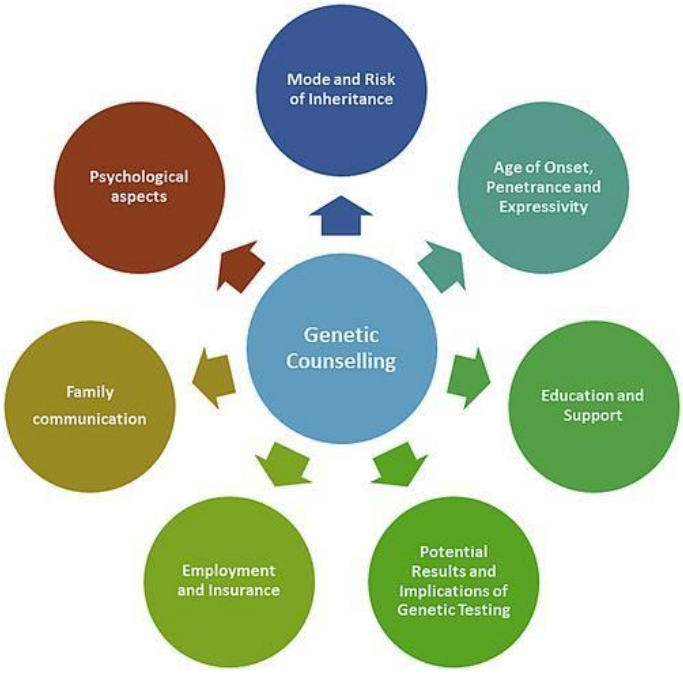

Figure 1. The Characteristic of Genetic Counseling

Genetic counseling started initially in the United States in 1969, while in Indonesia it was only initiated by the Faculty of Medicine, Diponegoro University in 2007 and until 2010 it had not yet reached the national policy stage (Rujito, 2018; Rujito & Ghozali, 2010). Then, in 2017, Gadjah Mada University began to initiate the development of genetic counseling services in Indonesia. Furthermore, the Faculty of Medicine, University of Padjadjaran through Dr. Hospital. Hasan Sadikin Bandung also provides genetic counseling services. In this case, genetic counseling is carried out related to hereditary diseases, such as sickle cell anemia, thalassemia, chromosomal abnormalities, adult onset, and so on.

Based on the description above, this study aims to examine the use of genetic counseling as a preventive measure for genetic diseases in order to form a harmonious family.

RESEARCH METHOD

This research uses the library method. This library research is carried out by collecting various reading references that are relevant to the problem under study, then understanding is carried out in a careful and careful way, so as to obtain research findings (Alawiyah et al., 2020; Rahmat, 2019; Rahmat, Kasmi, et al., 2020; Rahmat, Nurmalasari, et al., 2020; Rahmat & Alawiyah, 2020). In analyzing the data, the author uses the method of content analysis, namely the data obtained and then compiled so as to facilitate the discussion of existing problems (Marufah et al., 2020; Priambodo et al., 2020; Putri et al., 2020; Rahmat, Madjid, et al., 2020; Rahmat, Ramadhani, et al., 2020; Widha et al., 2021).

RESULT AND DISCUSSION

1. Genetic Counseling: a Short Review

Genetic counseling according to the National Society of Genetic Counselors (Rujito, 2018; in Rujito & Ghozali, 2010) is a communication process related to human health problems, namely the incidence of genetic diseases in a family. Joao (in Setiawan et al., 2020) states that genetic counseling is a process of communicating genetic disease problems so that they are conveyed properly to a family. This process involves various parties such as one or more trained people who assist the family or individual in understanding the medical facts related to the diagnosis, prognosis, and management of genetic diseases. Evans & Biesecker (in Mirani, 2009) also explain genetic counseling as a communication process for an individual or family with a medical condition consisting of diagnosis of the cause of the disease, disease management, pattern of disease decline, risk of recurrence in the family, and making the best possible way to do it. therapy, as well as knowing the risk of recurrence of the disorder. The purpose of genetic counseling is to provide understanding about the causes of the disease, how to reduce risk in the family, and choosing the optimal action to overcome the disease.

In addition, Evans & Biesecker (2006) mention the objectives of genetic counseling are as follows.

a. Understanding genetic disorders in families.
b. Understand patterns of inheritance and risk of recurrence in families.
c. Understand options related to illness.

d. Using this information can reduce psychological effects and increase personal control.
e. Make choices that are appropriate to the risk of disease and family goals, and act according to the options that have been selected.
f. Make the most appropriate choice on the sick family and on the offspring of the person.

According to Ariyanto et al. (2021), the stages of genetic counseling are presented in Table 1.

Table 1. The Stages of Genetic Counseling

Stages	Explanation
Pre-Counseling	- Preparing the necessary information. - Reviewing health records relevant to the client's genetic problems. - Discussing again the client's health condition with the relevant health personnel. - Preparing information to be provided with educational aids in the form of flipcharts and leaflets.
Inter-Counseling - Opening/ Information Greetings	- Saying greetings. - Please sitting in a place that has been prepared. - Confirming the identity of the respondent. - Asking about his health or condition. - Thank you for coming.
- Introduction	- Introducing myself. - Asking family members who suffer from genetic diseases. - Explaining the purpose and objectives of genetic counseling.
- Diagnosis	- Asking for information on the course of the disease so that the doctor diagnoses the genetic disease.
- Information Gathering	- Explaining the definition, epidemiology, etiology, pathophysiology, classification, clinical manifestations, therapy, prognosis, and prevention of this genetic disease. - Identifying family pedigree. - Calculating the risk of recurrence in the next generation.
Psychological Assessment	- Observing the psychological response of participants during genetic counseling or answering the questions given.
Discussion	- Confirming the participant's ability to capture the information provided. - Providing opportunities to ask questions about unclear information.
Decision Making	- Asking the next lineage plan. - Asking family members about screening test options. - Asking about the commitment to maintain a lifestyle.

Stages	Explanation
Closing	- On-going support with a contact number or email address. - Saying thanks. - Saying greetings.
Post-Counseling	- Evaluating the implementation of genetic counseling. - Record documentation of the implementation of genetic counseling.

2. Overview of Harmonious Family

A harmonious family is a happy family which is characterized by reduced tension, disappointment, and satisfaction with all circumstances and their existence which includes physical, mental, emotional, and social aspects (Sainul, 2018). Basri (in Abdurrahman et al., 2020) also provides the concept of a harmonious family, namely a harmonious, happy, orderly family, respecting each other, forgiving, helping in virtue, having a good work ethic, being neighbors with mutual respect, being obedient to worship, and uses free time with positive things and is able to fulfill the family basis.

Ali (in Sainul, 2018)states that a harmonious family is a family filled with tranquility, peace, love, descent and continuity of generations of people, compassion and sacrifice, complementing and perfecting each other, as well as helping and working together. According to Sarwono (in Siahaan, 2016), family harmony will be created if all family members feel happy and help each other. Psychologically, it can mean two things, namely the creation of desires, ideals, and hopes from all family members, and a little bit of personal conflict may occur between individuals. Dlori (in Widayani & Mardyawati, 2021) explains that a harmonious family is a form of relationship that is filled with love and affection, because these two things are the bonds of harmony. This loving family life in Islam is called *mawaddah-warahmah*, namely a family that maintains feelings of love, namely love for husband/ wife, love for children, and also love for work.

Based on the description above, a harmonious family is a situation and condition in the family in which a strong life is created, a warm atmosphere, mutual respect, mutual understanding, mutual openness, mutual care, and colored by love and mutual trust, thus enabling children to to grow and develop in a balanced way and create a spirit of learning in children. According to Dlori (in Aziz & Mangestuti, 2021), the keys to the formation of a harmonious family are as follows.

a. Love. Without them, the household will not run harmoniously. Because both are power to run household life.
b. Adaptation in all types of interactions, both differences in ideas, goals, likes, desires, and all the things behind the problem. It must be based on one goal, namely household harmony.
c. Fulfillment of inner and outer living in the family. With a living, the expectations of families and children can be realized so as to create continuity in the household.

Sainul (2018) explained in Islam, the characteristics of a harmonious family are as follows.

a. The formation of a family based on the hope of *Allah*'s pleasure without others. Both parties complement and perfect each other, fulfill the call of *fitrah* and *sunnah*, establish friendship and affection, and achieve physical peace and tranquility.
b. The purpose of forming a family. Household harmony will be realized if both partners are consistent with each other in the agreement they set together. Their main goal is to go to the path that has been outlined by *Allah* and seek His pleasure, in all their actions their focus is on *Allah* alone.
c. Environment. In a harmonious family, the effort that is always maintained is a loving atmosphere and each member carries out his role perfectly. The family environment is a place for shelter and shelter, a place where development and hardship are shared.
d. The relationship between the two partners. In a harmonious and balanced household relationship, husband and wife try to complement and perfect each other. They blend in and share what other family members feel. They treat each other, make each other happy and unite steps and goals, both of which prepare the means to draw closer to *Allah.*
e. Relationship with children. Harmonious families consider children as part of themselves. They build relationships on the basis of respect, protection of rights, education, proper guidance, purification of affection, and moral supervision of children's behavior.
f. Sit together. Harmonious families are always ready to sit together and talk with their family members, they try to understand each other and create an intimate relationship. Islam teaches that the old love the young, and the young respect the old.
g. Cooperation helps each other. In a harmonious domestic life, each member of the household has a certain task, they unite to carry the burden together.
h. Efforts for the common good. In a harmonious family life they make each other happy. They try to fulfill each other's desires and maintain their partner's tastes. For the common interest they always consult and communicate to ask for opinions, when the child is able to understand the problem, he is included in the deliberation process.

3. Reducing Genetic Diseases with Premarital Genetic Counseling

Genetic diseases are diseases that are passed from one generation to the next. Genetic diseases in a family are often overlooked by most people. As a result, abnormal offspring will be born but as carriers or patients with genetic diseases and even cause death. The process of spreading can also occur through marriage (Rujito & Ghozali, 2010). The disease is inherited directly by one or both parents who suffer or the occurrence of genetic crosses between two individuals. Marriage causes genetic variation both at the level of chromosomes, genes and DNA. These variations will cause a variety of clinical manifestations.

Manifestations of genetic disease cannot be avoided if there is birth and only need clinical management through treatment such as surgery and therapy. However, its manifestation can be prevented in the offspring to be born. Preventive action that can be taken is premarital genetic counseling. Premarital genetic counseling provides education about family history of genetic diseases, inheritance patterns in children, diagnosis and prognosis, supporting examinations, and decision making.

Premarital genetic counseling helps couples, especially in families with a prevalence of genetic diseases (prevalence > 5%) to conduct an examination of their respective health conditions (Bener et al., 2019). This needs to be done before planning a marriage to produce offspring. The use of genetic counseling is also expected as an effort to mitigate the prevalence of people with genetic diseases.

In a few decades, such premarital checks need to be done. This takes into account the increasing prevalence of genetic diseases in the world that even cause death. This examination aims to obtain an overview of family medical history and genetic diseases that have a probability of being passed on to the next generation. After that, management and handling if there are problems can be done as a prevention effort.

Genetic diseases are genetic problems or disorders that are passed down from one generation to the next with complex variations or follow Mendel's Laws. Buzzle (in Tjahjani & Zuhaida, 2017) states that variations in the genetic scope that will cause genetic diseases are classified into three, namely single, multifactorial and polygene genes, and mitochondrial DNA. This will be explained as follows.

a. Single gene variation means a genetic disorder that occurs in a single gene. The disorder is caused by a mutation in a single gene. Mutations will cause disease directly in the offspring and can also be passed down without causing clinical manifestations. The variations that occur consist of 5 types, namely autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, and Y-linked. Diseases due to this variation include Hintington's, galactosemia, X-linked hypophosphatemia, ocular albinism, infertility, albino, thalassemia, and hemophilia.
b. Polygenic and multifactorial occur when genetic factors and non-genetic factors interact, such as environment, lifestyle, and habits. Manifestations of this disease include autism, diabetes mellitus, cancer, hyperthyroidism, Alzheimer's disease, schizophrenia, cerebral palsy, bipolar disorder, cleft lip and palate, osteoporosis, cardiovascular disorders, anemia, and Parkinson's disease.
c. Mitochondrial DNA-associated abnormalities can be caused by functionally inherited maternally inherited mitochondrial DNA (mtDNA) damage. Manifestations of mtDNA genetic dysfunction are found in several diseases such as Pearson syndrome, Kearns-Sayre syndrome (KSS), leber hereditary optic neuropathy (LHON), myoclonic epilepsy and ragged-red fibers (MERRF), maternally-inherited diabetes and deafness (MIDD), neuropathy ataxia and retinitis pigmentosa (NARP), chronic progressive external ophthalmoplegia (CPEO), and Mitochondral myopathy, encephalopathy, lactic acidosis and stroke-like episodes (MELAS).

Based on the description above, it is illustrated that genetic diseases include dangerous diseases and have a very high risk of death for sufferers. Therefore, an effort is needed to prevent or control the manifestation of genetic diseases through genetic counseling. Genetic counseling is done by tracing family medical history and pedigree analysis before marriage. Premarital genetic counseling is targeted so that the resulting offspring will be born in good and quality conditions. This activity is carried out thoroughly for each partner and their family with prevalence. By considering ethical, moral, religious, and cultural values, genetic counseling can be carried out optimally.

CONCLUSION

Genetic counseling is a process of communicating genetic disease problems so that they are conveyed properly to a family. The aim is to provide an understanding of the causes of genetic disease, how to reduce risk in the family, and the selection of optimal actions in overcoming the disease. Genetic counseling is very important before marriage and can be part of premarital counseling. This is done so that the resulting offspring will be born in good and quality conditions in order to achieve a harmonious family.

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