PREMARITAL HEALTH SCREENING – A REVIEW AND UPDATE

Malik Arman[1], Sana Farooq[2], Ayesha Khan[3], Omar Rahim[4], Nadia Siddiqui[5]

[1]*Department of Psychology, Faculty of Social Sciences, Oxfordshire University, Oxford, UK*
[2]*Department of International Relations, Faculty of Law, Cambridge Metropolitan University, Cambridge, UK*
[3]*Department of Software Engineering, Faculty of Computer Science, Westminster Tech University, London, UK*
[4]*Department of Educational Studies, Faculty of Education, Yorkshire University, Leeds, UK*

**** Correspondence:***
Dr. Emma Richardson
Department of Educational Technology
Faculty of Education, Manchester University
M13 9PL, Manchester, UK
Email: emma.richardson@manchester.ac.uk
Phone: +44 (161) 555 9876
Fax: +44 (161) 555 1234

Abstract

Introduction:
In Bangladesh, inherited genetic disorders such as thalassemia and hemoglobinopathies are prevalent, contributing to substantial morbidity and mortality. These conditions also place a significant economic burden on the healthcare system. Premarital screening presents a viable solution to reduce the number of children born with these genetic disorders or other blood-transmitted diseases.
Aim:
This review aims to raise awareness among the general public and rekindle the interest of healthcare stakeholders in the management of inherited genetic disorders. It also seeks to propose recommendations for the establishment and implementation of premarital screening programs across Bangladesh.
Materials & Methods:
Relevant literature from academic journals and reputable online sources was collected to discuss the definition, benefits, and significance of premarital screening. The review also explores various types of investigations conducted during screening programs and outlines strategies for their effective implementation in Bangladesh.

Keywords: *Premarital screening, Thalassaemia, Haemoglobinopathies, Consanguineous, Genetic.*

Introduction

Premarital Health Screening (PMS) is a preventive measure in which couples intending to marry undergo tests to screen for genetic disorders, infectious diseases, and blood-transmitted conditions. The goal is to minimize the risk of passing any hereditary or infectious diseases to offspring. In contemporary society, PMS has become an essential component in preventing genetic disorders, congenital abnormalities, and various medical and psychological challenges that may arise in marital relationships (Rahman, 2014). In addition to PMS, Premarital Counseling (PMC) plays a crucial role in promoting healthy marriages. It is widely accepted across different cultural and religious settings due to its low health and financial costs. It provides couples with essential information to make informed decisions about their health and marital future (Naznin, 2014). A significant focus of premarital counseling is the prevention of hemoglobinopathies, such as sickle cell anemia and thalassemias. These inherited diseases are major public health concerns in regions such as the Mediterranean, the Middle East, the Indian subcontinent (including Bangladesh), Southeast Asia, tropical Africa, and the Caribbean. According to the World Health Organization (WHO), approximately 250 million people globally are carriers of inherited hemoglobinopathies, including thalassemia and sickle cell disease (Giti, 2014). In countries like Saudi Arabia and other parts of the Muslim world, the high rate of consanguineous and familial marriages has resulted in a higher incidence of recessive genetic disorders, further exacerbating these health challenges (Islam, 2014). Genetic disorders, particularly thalassemia and hemoglobinopathies, are widespread in regions like South Asia, including Bangladesh, where they cause significant health issues. These hereditary diseases, such as hemoglobin E disease, thalassemia, and sickle cell disease, are prevalent in Southeast Asia and are a cause of considerable suffering (Khatun, 2014). The burden of these conditions remains a significant public health challenge in Bangladesh and other South Asian countries, highlighting the need for effective preventive strategies.

Despite advancements in healthcare in Bangladesh, including improvements in infant mortality and life expectancy, the country lags behind its neighbors, such as India, in providing genetic services. Studies from industrialized nations show that genetic diseases or birth defects affect approximately 3% of all pregnancies, contributing to up to 30% of pediatric hospital admissions and nearly 50% of childhood deaths. Furthermore, recessively inherited disorders contribute to less than 20% of single-gene disorders and less than 5% of congenital and genetic diseases (Rahman, 2014). In contrast, genetic and congenital disorders contribute significantly to perinatal and neonatal mortality worldwide. With proper intervention, effective prevention measures can reduce up to 95% of affected births. One of the most effective methods of preventing genetic disorders, such as beta-thalassemia, is through carrier screening, which can be easily and inexpensively performed with a simple blood test. This allows couples to understand the genetic risks they face and make informed decisions about their reproductive health, including options for prenatal diagnosis (Naznin, 2014).

Background

Initial screening for thalassaemia was first introduced in 1975 by Silvestroni and colleagues in Latium, Italy, as part of a school-based prevention program. Similarly, sickle cell anaemia screening began earlier, in Virginia, USA, in 1970. By the 1970s, several countries including Canada, Cyprus, Greece, Italy, and the UK had initiated national screening programs, which proved to be successful. At that time, although the genetic causes of haemoglobinopathies were understood, little was being done to prevent these conditions in newborns. The commencement dates of premarital screening programs, along with the types of investigations conducted, are summarized in Table-I.

Global Aspects of Premarital Screening Programs

Premarital screening programs are designed to educate couples, providing them with accurate and unbiased information. These programs should be universally accessible to those who wish to participate, using proper diagnostic techniques. It is crucial that informed consent is obtained, along with assurances of privacy and the fair treatment of affected individuals. The success of premarital screening is greatest when social, religious, ethnic, and cultural factors are all taken into consideration.

Endemic Mediterranean countries: These countries have well-established and highly successful preventive

programs (with a success rate of 80–100%) and provide optimal treatment through specialized clinics. Developed, industrialized countries: In these countries, the prevalence of haemoglobinopathies is rising due to immigration. While they can fund screening programs, reaching immigrant populations with specific cultural backgrounds remains a challenge. Developing countries: These nations face economic challenges and prioritize other health concerns, such as infectious disease control, or may be hindered by cultural or religious restrictions. In Mediterranean countries such as Cyprus, Greece, and Italy, premarital screening for thalassaemia has been a long-standing practice due to high levels of consanguinity. Similar preventive programs have been introduced in Bahrain, China, India, Iran, Indonesia, Malaysia, the Maldives, Singapore, Thailand, and more recently in Saudi Arabia and the United Arab Emirates. In the UK, Northern Ireland, and other countries in Northwest Europe, prenatal screening is available, with abortion being one of the preventive measures. In China, couples planning to marry undergo thorough screening, including physical examinations. They are provided with premarital health education and counseling, which includes watching videos about the potential outcomes of childbearing. However, this practice has faced criticism regarding human rights, control, oppression, and eugenics, despite the acknowledged value of comprehensive premarital screening.

In Lebanon, thalassaemia patients receive care in chronic-care centers in collaboration with the ministries of Social Affairs and Public Health. These programs focus on raising public awareness, training healthcare professionals, and developing educational materials. In Mersin, Turkey, where consanguinity rates are high, premarital screening for haemoglobinopathies is offered. Couples who are both carriers of haemoglobinopathies are counseled confidentially about their options, including the possibility of prenatal diagnosis. In Saudi Arabia, premarital testing for haemoglobinopathies is mandatory, and couples at risk receive counseling and guidance, though prenatal diagnosis is not offered.

Country	Date Started	Type of Screening
Italy	1975	Thalassaemia (mandatory)
Bahrain	1985	Thalassaemia and sickle cell disease (mandatory)
Iran	2004	Thalassaemia (mandatory)
Jordan	2004	Thalassaemia (mandatory)
Saudi Arabia	2004	Thalassaemia and sickle cell disease (mandatory)
United Arab Emirates	2007	Thalassaemia (mandatory)
Tunisia	†	Thalassaemia (mandatory)
Egypt	†	Thalassaemia and STDs (voluntary)
Spain	†	Thalassaemia and STDs (voluntary)
Portugal	†	Thalassaemia and STDs (voluntary)
Turkey (Merlin)	1998	Thalassaemia (mandatory)
Turkey (Denizlin)	1995	Thalassaemia (voluntary)
Cyprus	1973	Thalassaemia (mandatory)
Canada	1970s	Thalassaemia, SCD & STDs (voluntary)
Greece	1975	Thalassaemia (voluntary)
UK	1970s	Thalassaemia, SCD & STDs (voluntary)
USA, Illinois & Louisiana	1983	STDs including HIV (mandatory), stopped
China	1992	Inherited diseases, HBV & HIV (mandatory till 2003)
Taiwan	1993	Thalassaemia (voluntary)
Brazil	†	Inherited & STDs (mandatory in some areas)
Palestine	†	Thalassaemia (mandatory)
Malaysia, Johor	2002	HIV (mandatory)
India	†	Inherited & STDs (voluntary)
Indonesia	†	Inherited & STDs (voluntary)
Maldives	†	Inherited & STDs (voluntary)

Singapore	†	Inherited & STDs (voluntary)
Thailand	†	Inherited & STDs (voluntary)

Role of Culture and Education in the Success of Premarital Screening Programs:

'Consanguinity,' which refers to relationships by blood or shared ancestry, increases the likelihood of inheriting recessive diseases. The risk grows with the closeness of the relationship, particularly in marriages between first cousins, which is common in certain cultures. In contrast, consanguineous marriages are rare in Western countries, where unions between first cousins are prohibited by the Orthodox Church and Roman Catholic Church and may be considered incestuous in the United States. Personal factors such as socioeconomic status can significantly affect the outcome of premarital screening programs. Educating couples and the entire screening team (including laboratory technicians, nurses, physicians, counselors, outreach workers, and social workers) is vital for the success of such programs. Schmidt emphasized that thorough educational planning before collecting the first blood sample can help avoid failures in the program. It is essential that the concept of 'carrier status' is explained to the public long before marriage, and that public education efforts involve collaboration between the government, religious and community leaders, health professionals, and school parent organizations.

People who are exposed to information about premarital screening generally show positive attitudes toward premarital counseling and screening for consanguineous marriages. This positive shift is linked to social changes, decreasing illiteracy, growing economic pressures, the rise of nuclear families, and delayed childbearing. Those with negative attitudes towards such screenings tend to be unmarried men. Eshra and colleagues proposed that educational campaigns about the benefits of premarital testing should focus on unmarried men, as they play a significant role in decisions regarding consanguineous marriages. In some communities, religious beliefs pose barriers to the effectiveness of screening programs. Despite the mandatory nature of premarital screening in southern Iran for over ten years, high-risk couples still choose to marry and have children affected by beta-thalassaemia. This is often due to cultural and religious traditions, as consanguineous marriages are allowed in Islam, contributing to the persistence of thalassaemia in certain areas. Many people also believe that their fate is determined by divine will, and thus accept the risks associated with having a child with genetic disorders. A recent report in The Jordan Times highlighted that some Jordanians view their union outcomes as destiny, with one interviewee commenting that despite having ten disabled children, they believe they will be rewarded in heaven. However, Islamic teachings also encourage healthy marriage practices and counseling to prevent genetic diseases.

AlKhaldi et al. assessed the attitudes of health science students in Saudi Arabia towards premarital screening and counseling. While most students held positive views, about 25% refused testing and counseling based on their interpretation of Islamic teachings. Awatif's study of female students at King Saud University revealed that 86% of them had a positive opinion on premarital testing. El-Hazmi's community-based study found that 94% of participants considered premarital counseling and testing crucial for preventing genetic diseases, with 87% believing such testing should be mandatory. Despite high levels of awareness, studies like Al Hamdan et al.'s on the first two years of Saudi screening programs revealed that approximately 90% of high-risk couples still proceeded with marriage, even after being informed of their genetic risks. This may be due to the study's focus on university students. Similar findings have been reported in various Islamic countries, where religious beliefs often undermine the success of premarital screening programs, despite the influence of education. Reports by Karimi et al., Monaghan, and AlKhaldi et al. in different Islamic countries suggest that religious factors can act as significant obstacles to screening success, regardless of educational levels. A study by Angastiniotis and Hadjiminas in 1981 revealed that religious support played a central role in the success of screening programs in Cyprus and Greece.

Premarital Screening Programs in Asia and the Indian Subcontinent:

In Asia, voluntary premarital screening programs for inherited and sexually transmitted diseases began in countries like China, Taiwan, Malaysia, India, Indonesia, Maldives, Singapore, and Thailand. However, there is no available data regarding premarital screening programs in countries such as Pakistan, Nepal, Sri Lanka, Bhutan, and Bangladesh. In India, a few private clinics offer premarital screening packages.

In countries like Bangladesh, Pakistan, Nepal, Bhutan, Sri Lanka, and India, inherited diseases, including hemoglobinopathies, are prevalent. Although these governments are focused on preventing more common communicable and chronic diseases due to economic constraints, as their economies improve, there is growing recognition that inherited diseases should also receive attention. Hemoglobinopathies and beta-thalassaemia are more common in these regions, and it is crucial for governments to formulate strategies and action plans to reduce the incidence of these inherited diseases.

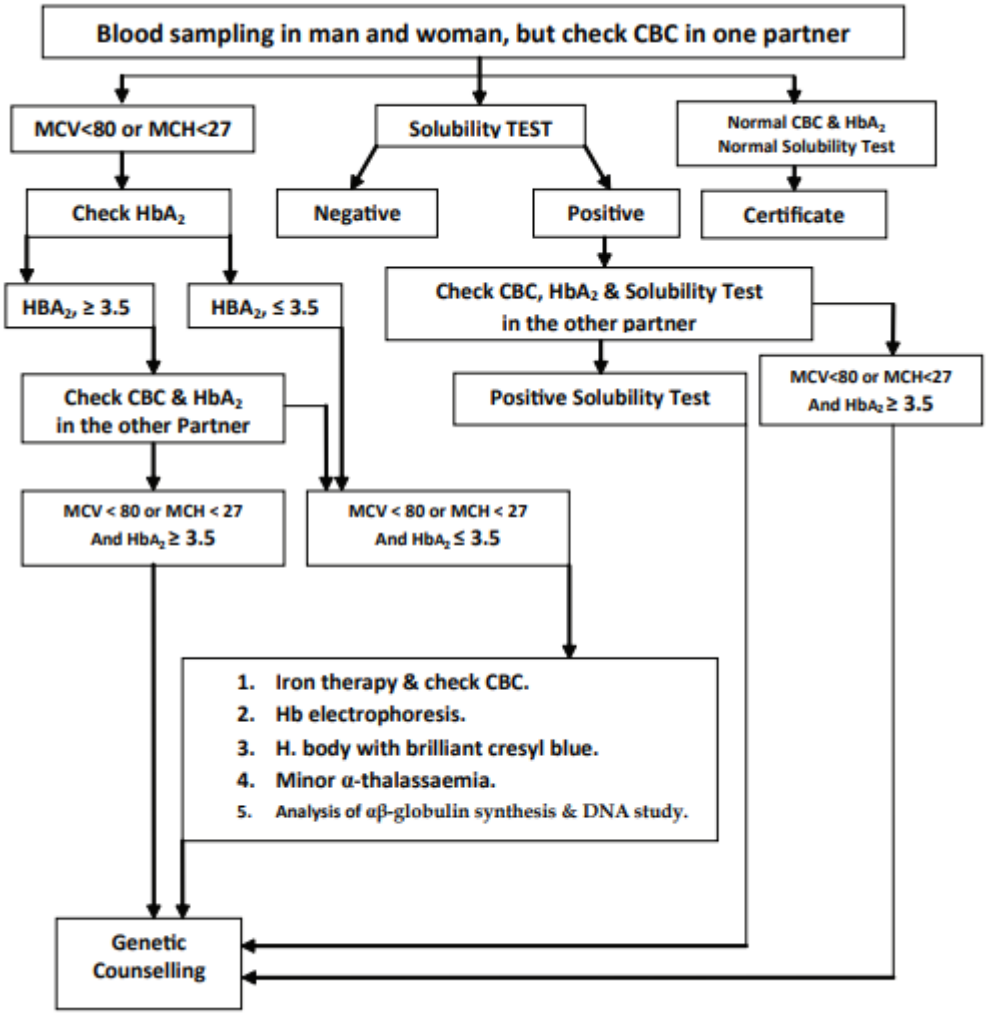

Fig -1: Recommended premarital screening programme.

Findings

Genetic counseling is an essential process in modern healthcare, aimed at providing individuals or families with information about potential genetic conditions that may affect them or their offspring. It helps them make informed decisions about marriage, reproduction, and healthcare management. The primary goal of genetic counseling is to protect the autonomy of individuals and couples, ensuring that they are fully informed about the genetic conditions that may impact their lives. This process includes not only offering information on the nature of genetic conditions but also providing valuable options that allow couples to navigate their choices regarding genetic risks, reproductive options, and health management. For individuals undergoing premarital screening for genetic disorders such as haemoglobinopathies, genetic counseling offers the opportunity to understand the risks and options available to them in a supportive, non-judgmental environment.

The Importance of Genetic Counseling in Premarital Screening

Premarital screening programs have become widely recognized for their effectiveness in detecting genetic disorders, such as thalassaemia and sickle cell disease, among others. However, despite the success of these screening programs, the effectiveness of genetic counseling in improving outcomes depends significantly on how well it is conducted and the willingness of individuals to accept and act on the advice given. According to Neal-Cooper and Scoot (2005), young couples, particularly those concerned about the possibility of producing a child with a genetic condition like sickle cell disease, are often driven by a deep desire to have children regardless of the associated risks. These individuals may overlook or resist counseling recommendations, which can complicate the decision-making process and delay or limit the potential benefits of genetic counseling.

Neal-Cooper and Scoot's study suggests that couples who are at risk of passing on genetic disorders should be directly contacted by counselors and actively encouraged to engage in education and counseling. In such cases, preventive measures and treatment options may not be as effective in preventing the disease, but counseling serves as a crucial step in guiding the couple through their decision-making process. Genetic diseases, especially those caused by inherited genetic mutations, are notoriously difficult to prevent or treat. Thus, the importance of counseling lies in its ability to offer couples the knowledge and support they need to make the most informed choices possible about their health, marriage, and future children.

Solution-Focused Premarital Counseling

One effective model for genetic counseling in the context of premarital screening is "solution-focused" premarital counseling. This approach, as discussed by Murray and Murray, centers around the couple's existing resources and strengths, helping them build a shared vision for their life together despite the potential challenges presented by genetic conditions. The focus is not solely on the medical or genetic risk, but rather on how the couple can move forward together, using their available resources to cope with any potential challenges. This approach is holistic and considers the emotional, psychological, and social aspects of the couple's relationship, rather than focusing solely on the genetic risks.

Solution-focused counseling typically includes intervention strategies tailored to the specific needs of the couple. These strategies may involve solution-oriented questions, feedback, and collaborative decision-making. A critical tool used in solution-focused counseling is the Couple's Resource Map (CRM), a visual representation of the couple's support network, including personal relationships, social connections, and contextual resources. This map helps the couple understand what support is available to them and how they can access it when needed. Additionally, solution-focused counseling provides a framework for discussing various options available to couples who test positive for a genetic disorder, including avoiding marriage altogether, exploring reproductive options such as prenatal diagnosis, or considering adoption. Other options include sperm or ova donation, pre-embryo donation from an unaffected individual, and pre-implantation genetic diagnosis (PGD), which involves testing embryos for genetic disorders before implantation during in vitro fertilization (IVF).

Ethical, Religious, and Cultural Considerations in Genetic Counseling

The success of genetic counseling in premarital screening programs also depends on several factors outside of medical expertise, particularly the cultural, religious, and ethical values of the couple and the society in which they live. These values can significantly influence the couple's willingness to accept genetic counseling and act on the information provided. For instance, in many cultures, marriage and reproduction are viewed as personal and sacred decisions. Religious or cultural beliefs may influence how couples perceive and respond to genetic counseling and prenatal diagnosis.

In many Muslim-majority countries, for example, abortion is prohibited on religious grounds, meaning that prenatal diagnosis is not as useful in preventing the birth of a child with a genetic condition. In these countries, the possibility of abortion is not an option unless the fetus is severely malformed. However, pre-implantation genetic diagnosis (PGD) is often permitted and may be more acceptable from a religious perspective. PGD allows couples to test embryos for genetic conditions before implantation and select those embryos that are not affected by the genetic disorder. This technique provides couples with the opportunity to have children free from genetic diseases without violating religious principles.

Religious beliefs aside, the availability and cost of various reproductive options play a significant role in decision-making. For instance, PGD, although a powerful tool for preventing genetic disorders, can be prohibitively expensive for many couples. The decision to pursue PGD may thus depend on the financial resources available to the couple and the healthcare system in their country. In some countries, public healthcare systems may cover the costs of these procedures, while in others, individuals may have to pay out of pocket, limiting accessibility for those without sufficient financial resources.

Global Disparities in Access to Genetic Counseling

While genetic counseling and premarital screening programs have proven effective in many countries, access to these services remains a major issue in low-resource settings. For example, in Bangladesh, there are still no recognized genetic centers that provide appropriate counseling services, leaving many couples without the necessary information to make informed decisions. Similarly, in many developing countries, economic challenges, limited healthcare infrastructure, and competing public health priorities often prevent the widespread implementation of genetic counseling and screening programs. In these settings, couples may not have access to the resources needed to make informed decisions about genetic risks, and as a result, the benefits of genetic counseling may be severely limited.

This lack of access to genetic counseling is particularly problematic in countries where consanguinity (marriage between close relatives) is common, as the risk of inherited genetic disorders, such as thalassaemia, is significantly higher. In countries like Saudi Arabia, where consanguinity rates are high, premarital screening and genetic counseling programs are crucial in helping couples understand their genetic risks and make decisions about marriage and reproduction. Without proper counseling, couples may unknowingly marry close relatives, increasing the risk of passing on genetic disorders to their offspring.

Conclusion

Genetic counseling plays a critical role in premarital screening programs by providing individuals and couples with the information and support they need to make informed decisions about their reproductive health. While screening for genetic disorders such as thalassaemia and sickle cell disease can identify at-risk individuals, counseling is essential to help couples understand their options and the implications of those choices. The success of genetic counseling depends on several factors, including the counselor's approach, the couple's willingness to accept counseling, and the cultural, ethical, and religious context in which the counseling takes place. Moreover, for counseling to be effective, it must be accessible, equitable, and culturally sensitive, addressing the needs of the target population. In countries where access to genetic counseling is limited, efforts should be made to expand these services, ensuring that all individuals have the opportunity to make informed choices about their reproductive health.

Conclusions

Inherited genetic disorders, particularly thalassaemias and haemoglobinopathies, are prevalent, incurable, autosomal recessive conditions that lead to significant health complications and pose a substantial financial burden on society. These disorders can be easily detected through simple tests, such as the measurement of Mean Red Cell Corpuscular Volume (MCV), which can identify carriers of these conditions before marriage. Premarital health screening serves as an essential tool to inform couples about the genetic risks they may face in having children and to provide them with the necessary counseling to make informed reproductive decisions.

Over time, premarital screening programs have gained widespread acceptance and are now considered a key component of preventive healthcare. In many countries, these programs have been made mandatory, reflecting their importance in reducing the incidence of genetic disorders and improving public health outcomes. However, the success and implementation of these programs are significantly influenced by various social factors, including religious beliefs, cultural norms, education levels, government policies, and the individual attitudes of couples.

For premarital screening programs to be effective, a collaborative approach involving a wide range of stakeholders is necessary. Scientists, healthcare professionals, politicians, educators, religious leaders, and social activists all have crucial roles to play in motivating the public and supporting the development of policies and legislation aimed at promoting and implementing premarital screening. Additionally, media outlets, including journalists and mass communication platforms, can play an influential role in raising awareness and informing the general population about the importance of genetic testing and screening. Physicians, midwives, genetic counselors, and clinical laboratory scientists, particularly those involved in prenatal and pre-conceptional care, can also be instrumental in educating prospective couples and ensuring they are well-informed about their reproductive options.

Ultimately, the success of premarital screening programs depends not only on the availability of testing but also on the commitment of society as a whole to create an environment in which individuals are empowered to make informed and responsible decisions about their health and future families. By fostering awareness and understanding, we can help prevent the transmission of genetic disorders and reduce the associated healthcare burdens, thereby improving public health outcomes and enhancing the quality of life for affected families.

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