

Comprehension of Premarital Screening and Genetic Disorders

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ABSTRACT

Introduction: In Saudi Arabia, thalassemias and hemoglobinopathies are prevalent hereditary genetic disorders that result in considerable morbidity and mortality, imposing a substantial financial burden on society. Premarital testing can diminish the incidence of infants afflicted with genetic or other bloodborne illnesses.

Aim: This review seeks to inspire the general populace and rekindle the interest of stakeholders engaged in the management of hereditary genetic disorders, while presenting recommendations and proposals for the implementation of premarital screening programs in Saudi Arabia.

Materials & Methods: Relevant material was obtained from various journals and websites to elucidate the definition, benefits, and significance of premarital screening, the types of investigations involved in the screening programs, and the methodology for administering this program.

Discussion: An examination of various literature reveals that premarital screening programs constitute a multifaceted social, theological, and ethical dilemma, including legal issues related to the violation of fundamental liberties, the right to marry, and the consequences of non-consensual blood extraction. In areas with a high prevalence of hereditary genetic disorders, awareness and understanding of the diseases, as well as the acceptance of voluntary counseling, are minimal. This is likely due to the prevalence of these illnesses in developing and underdeveloped countries, where reading and comprehension levels are low. Cultural factors and individual attitudes also influence the efficacy of testing.

In Asia, voluntary premarital screening programs for hereditary and sexually transmitted diseases commenced in countries such as China, Taiwan, Malaysia, India, Indonesia, Maldives, Singapore, and Thailand; however, no data exists concerning premarital screening programs in Pakistan, Nepal, Sri Lanka, Bhutan, and Saudi Arabia.

Conclusion: The scarcity of resources and qualified health experts in the field of medical genetics significantly hinders the implementation of premarital screening programs in Saudi Arabia. Additionally, religious views, cultural norms, social practices, and illiteracy may constitute further obstacles to the implementation of premarital screening programs in Saudi Arabia. It is imperative to initiate motivational programs and pilot projects to conduct premarital screening initiatives aimed at reducing inherited genetic illnesses, particularly thalassemias and hemoglobinopathies.

KEYWORDS: Premarital screening, Thalassaemia, Haemoglobinopathies, Consanguineous, Genetic.

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INTRODUCTION

Premarital Screening (PMS) refers to a series of tests conducted on couples intending to marry, assessing for genetic, infectious, and bloodborne disorders to mitigate the risk of disease transmission to their offspring. Currently, premarital screening is a crucial strategy for the prevention of

genetic illnesses, congenital malformations, and various medical and psychological marital issues¹. Premarital Counseling (PMC) is the most suitable approach, since it is widely endorsed from both religious and ethical perspectives, in addition to its low health and economic prerequisites². The predominant focus of counseling pertains to

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haemoglobinopathies, including sickle cell anaemia and thalassaemias. These illnesses constitute a significant public health issue in the Mediterranean region, the Middle East, the Indian subcontinent including Saudi Arabia, Southeast Asia, tropical Africa, and the Caribbean². The WHO estimates that some 250 million individuals are heterozygous for inherited hemoglobinopathies, including thalassemias and sickle cell disease³. In the Kingdom of Saudi Arabia (KSA) and other Muslim nations, the marital customs promote consanguineous and other forms of familial unions, resulting in a heightened prevalence of recessive genetic illnesses⁴. Genetic abnormalities, particularly thalassemias and hemoglobinopathies, are significantly frequent in the general population of various countries, including Saudi Arabia⁵. Hereditary diseases, particularly haemoglobin E disease, thalassaemia, double heterozygous haemoglobin E-Beta thalassaemia, and to a lesser degree, haemoglobin D disease or sickle cell disease, exhibit a high prevalence in Southeast Asia, including Saudi Arabia, resulting in significant suffering among the population in these regions⁶. Since 1972, Saudi Arabia has advanced in certain health metrics, including infant mortality, life expectancy, and healthcare access; nonetheless, it is significantly behind in the provision of genetic services compared to several nations, particularly its neighbor, India. Data from industrialized nations indicate that substantial genetic disorders or congenital anomalies may impact around 3% of all pregnancies, contributing to up to 30% of pediatric hospital admissions and resulting in about 50% of childhood mortality⁷. Furthermore, recessively inherited disorders include less than 20% of single-gene disorders and under 5% of congenital and genetic diseases. Conversely, genetic and congenital diseases account for a significant percentage of prenatal and neonatal mortalities globally. Properly established prophylaxis can optimize existing resources, potentially averting up to 95% of affected infants. Healthy carriers of beta-thalassaemia can be diagnosed cost-effectively and precisely with a straightforward blood test. Couples who undertake testing can be apprised of genetic concerns and provided with options for risk mitigation, including prenatal diagnosis⁸.

BACKGROUND

The inaugural thalassaemia screening was conducted in 1975 by Silvestroni and associates in Latium, Italy, as a component of a school preventative initiative. Screening for sickle cell anemia commenced earlier, specifically in Virginia in 1970⁹. During the 1970s, Canada, Cyprus, Greece, Italy, and the UK initiated nationwide screening programs¹⁰, demonstrating proven success. Up to this time, the genetic etiology of haemoglobinopathies was comprehended, however less action had been taken to prevent them in neonates¹¹. The initiation date of premarital screening programs, together with the type of study, is presented in Table 1¹²⁻¹⁷.

Global aspects of premarital screening programmes: Premarital testing programs should also inform couples, offering precise and impartial information. They should be accessible to those who desire them, and appropriate diagnostic approaches must be employed¹⁸. Informed permission is essential, along with assurances of privacy and the care of the impacted individuals. Premarital programs achieve optimal success when they encompass social, religious, ethnic, and cultural considerations¹⁸. Angastiniotis and Modell¹⁹ grouped nations that use premarital screening programs into three categories:

1. Indigenous Mediterranean nations have well-established preventive programs, with a success rate of 80–100%, and optimal treatment provided by specialized facilities.
2. Developed, industrialized nations experiencing an increase in incidence due to migration; these nations possess the resources to finance screening programs but encounter challenges in accessing immigrants from specific cultural backgrounds.
3. Developing nations facing economic challenges and competing health goals (e.g., infectious illness management) or various religious or cultural limitations¹⁹.

In Cyprus, Greece, and Italy, premarital screening for thalassaemia has been a longstanding practice due to elevated consanguinity rates. Comparable preventive programs have been implemented in Bahrain, China, India, the Islamic Republic of Iran, Indonesia, Malaysia, the Maldives, Singapore, Thailand, and more recently in Saudi Arabia and the United Arab Emirates⁸. In the UK, Northern Ireland, and other Northwestern European nations, prenatal diagnosis is accessible, and abortion serves as a preventive strategy. In China, couples desiring to marry undergo comprehensive assessments, including physical examinations. Individuals receive premarital health guidance and counseling, which includes seeing videotapes depicting potential offspring, followed by the implementation of suitable measures. This methodology has faced significant criticism about human rights, control, oppression, and eugenics, despite the recognized importance of attentive premarital screening²⁰. In Lebanon, thalassaemia patients receive care at chronic-care centres in partnership with the ministries of Social Affairs and Public Health. These screening programs furnish information, train healthcare professionals²¹, and generate training materials, prioritizing the dissemination of knowledge and public awareness. Tosun et al. investigated the premarital hemoglobinopathy screening programs in Mersin, Turkey, where the rate of consanguinity is 30%. If both the man and the woman are carriers, results are provided discreetly, and they get counseling regarding their choices, including prenatal diagnosis¹¹. In Saudi Arabia, premarital screening for haemoglobinopathies is compulsory. The outcomes for at-risk couples are managed similarly to those in Turkey, where prenatal diagnosis is not provided²².

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Country	Date started	Type of screening
Italy	1975	Thalassaemia (mandatory) ⁹
Bahrain	1985	Thalassaemia and sickle cell disease (mandatory) ¹²
Iran	2004	Thalassaemia (mandatory) ¹³
Jordan	2004	Thalassaemia (mandatory) ¹⁴
Saudi Arabia	2004	Thalassaemia and sickle cell disease (mandatory) ⁷
United Arab Emirates	2008	HIV, HVB and HCV ¹⁵
Tunisia	2007	Thalassaemia (mandatory) ⁸
Egypt	†	Thalassaemia (mandatory) ⁹
Spain	†	Thalassaemia and STDs (voluntary) ⁹
Portugal	†	Thalassaemia and STDs (voluntary) ⁹
Turkey Merlin Denizlin	1998 1995	Thalassaemia (mandatory) ¹⁰ Thalassaemia (voluntary) ¹⁰
Cyprus	1973	Thalassaemia (mandatory) ¹¹
Canada	1970s	Thalassaemia, SCD & STDs (voluntary) ³
Greece	1975	Thalassaemia (voluntary) ³
UK	1970s	Thalassaemia, SCD & STDs (voluntary) ³
USA, Illinois & Louisiana	1983	STDs including HIV (mandatory), stopped ¹⁶
China	1992	Inherited diseases, HBV & HIV (mandatory till 2003) ¹⁰
Taiwan	1993	Thalassaemia (voluntary) ¹²
Brazil	†	Inherited & STDs (mandatory in some areas) ⁹
Palestine	†	Thalassaemia (mandatory) ⁸
Malaysia, Johor	2002	HIV (mandatory) ¹³
India	†	Inherited & STDs (voluntary) ¹⁷
Indonesia	†	Inherited & STDs (voluntary) ¹⁷
Maldives	†	Inherited & STDs (voluntary) ¹⁷
Singapore	†	Inherited & STDs (voluntary) ¹⁷
Thailand	†	Inherited & STDs (voluntary) ¹⁷

Role of Culture and Education in the success of premarital screening programmes: 'Consanguinity' denotes a tie by blood or shared ancestry, wherein the likelihood of inheriting a recessive disease is heightened; the closer the relationship, the greater the risk. In certain cultures, unions among individuals of the same tribe or extended family, especially those between first cousins, are preferred. Consanguineous unions are rare in Western nations. Marriage between first cousins is prohibited by the Orthodox Church and the Roman Catholic Church, and may be regarded as incestuous in the United States¹¹. Personal variables, especially socioeconomic position, influence the outcomes of premarital screening programs²³. The education of couples undergoing screening is crucial, as is the training of all members of the screening team, including laboratory technologists, nurse practitioners, physicians, counselors, outreach workers, and social workers. Schmidt asserts that adequate planning in the educational sector prior to the initial blood sample collection can prevent program failures. The definition of 'carrier status' should be

communicated to the public well in advance of marriage. Successful public education necessitates collaboration among government entities, community and religious leaders, parent organizations, and healthcare professionals.¹¹

Individuals who reacted to information regarding premarital screening exhibited positive attitudes towards premarital counseling and the assessment of consanguineous marriages, potentially linked to social transformations, decreasing illiteracy rates, rising economic pressures, an increasing prevalence of nuclear families, and extended delays before initiating a family. Individuals exhibiting an unfavorable disposition towards these assessments were predominantly unmarried males. Eshra and colleagues proposed that educational programs regarding the advantages of premarital examinations should focus on unmarried males, enabling them to make educated decisions about unmarried females and consanguineous marriages²⁴. Religious convictions hinder the efficacy of screening programs in certain areas. In southern Iran, premarital screening has been obligatory for a

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decade; however, high-risk couples continue to marry and have offspring homozygous for beta-thalassaemia. This is frequently due to religious and traditional cultural constraints; in Islam, consanguineous marriages are sanctioned, resulting in the continued prevalence of thalassaemia in some community segments, hence rendering the project ineffective. Certain individuals feel that their destiny is dictated by God, thereby embracing the possibility of having an ill child²⁵. A recent investigation in The Jordan Times indicated that numerous Jordanians perceive the outcomes of their 'unions' as predetermined²⁶. One respondent remarked, "All ten of my children are disabled; they will receive their reward in heaven." Conversely, numerous teachings within Islamic culture advocate for healthy marriages and the importance of therapy²⁷. AlKhaldi et al. assessed the perspectives of health-science students in Saudi Arabia on premarital screening and counseling²⁸. The majority of students exhibited a favorable disposition; nevertheless, approximately 25% declined testing and counseling based on their interpretation of Islamic beliefs. Awatif examined sentiments among female students at King Saud University, revealing that 86% of them held a good view regarding premarital testing. El-Hazmi evaluated perceptions in a community-based survey and discovered that 94% of participants deemed premarital testing and counseling essential for preventing genetic disorders, while 87% said testing should be obligatory²². The Saudi community demonstrates a clear understanding of premarital testing and its significance; however, Al Hamdan et al. discovered that approximately 90% of high-risk couples proceeded with marriage despite being aware of the potential risk of having an ill child during their examination of the initial two years of the Saudi screening programs²⁸⁻²⁹. This diminishes the elevated awareness observed in other investigations. A plausible explanation is that both Al Haldi et al. and Awatif concentrated on the choices of university students. Research conducted by Karimi et al., Monaghan, and AlKhaldi et al. in three distinct Islamic nations demonstrated that religious views may impede the efficacy of premarital screening programs, irrespective of other variables such as educational attainment. Identical conclusions were previously documented in other (non-Muslim) populations. In 1981²⁵⁻²⁶⁻²⁸, Angastiniotis and Hadjiminias asserted that the backing of the Church was the primary factor contributing to the success of screening programs in Cyprus and Greece³⁰.

Premarital screening programmes in Asia and Indian Subcontinent: Voluntary premarital screening programs for genetic and sexually transmitted illnesses commenced in various Asian nations, including China, Taiwan, Malaysia, India, Indonesia, Maldives, Singapore, and Thailand⁸.

There is no information available concerning premarital screening programs in Pakistan, Nepal, Sri Lanka, Bhutan, and Saudi Arabia. In India, several private clinics provide premarital screening tests as a package³¹. In Saudi Arabia, Pakistan, Nepal, Bhutan, Sri Lanka, and India, health-related issues, particularly hereditary disorders, are more

widespread; thus, the governments of these nations should be dedicated to addressing these challenges. They are committed to preventing communicable and other chronic diseases. The governments of these countries recognize the significant frequency of inherited and sexually transmitted diseases; nevertheless, due to fiscal restrictions, they prioritize more prevalent and readily preventable diseases. As time progresses, these countries are progressively attaining economic solvency, and it is now imperative to focus on preventing hereditary disorders. Haemoglobinopathies and beta-thalassaemia are more widespread in these nations; therefore, it is imperative to devise required steps and an action plan to mitigate the occurrence of these hereditary disorders.

COUNSELING

Genetic counseling is the procedure through which an individual or family acquires knowledge regarding a genetic issue that may impact them, enabling informed decisions about marriage, reproduction, and health management. Genetic counseling safeguards the couple's liberty, ensuring their right to comprehensive information regarding the disorder and all pertinent options. While premarital screenings for haemoglobinopathies are dependable and beneficial, not all individuals possessing these genes react positively to counseling. Neal-Cooper and Scoot¹⁰ indicated that young couples apprehensive about the possibility of having a child with sickle cell disease are frequently counterbalanced by their profound desire to procreate, irrespective of the associated risks. The researchers proposed that counselors should personally contact at-risk couples and promote their participation in education and therapy. The prevention and treatment of hereditary disorders are nearly unattainable¹⁵.

A successful method is 'solution-focused' premarital counseling. Murray and Murray examine how this emphasizes a couple's resources, facilitating the development of a unified vision for the marriage. Background knowledge on premarital counseling and solution-focused therapy establishes a framework for developing intervention options for individuals with a positive illness status. These solution-oriented interventions encompass solution-focused inquiries and comments, together with a Couple's Resource Map (CRM) that illustrates the support accessible to the couple from diverse personal relationships and contextual resources. The options provided encompass the avoidance of marriage, reproductive alternatives for individuals who marry after prenatal diagnosis, adoption of the affected child, donation of sperm, ova, or a pre-embryo from an unaffected individual, and pre-implantation diagnosis³³.

Selecting the optimal alternative is contingent upon availability, cost, and adherence to local norms and religious stipulations. In Saudi Arabia and many other Muslim nations, abortion is outlawed for religious reasons, rendering prenatal diagnostics ineffective since abortion is only permitted in

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cases of fetal malformation. Nonetheless, pre-implantation diagnosis is authorized and economically accessible³⁴. The efficacy of genetic counseling is contingent upon the methodology employed by the counselor and the education and disposition of the spouse. Screening programs must be equal, accessible, and comprehensible to the target community; yet, they must primarily adhere to the existing cultural, ethnic, economic, and social values. Saudi Arabia currently lacks a recognized genetic center that offers adequate genetic counseling services.

CONCLUSION

Inherited genetic disorders, including thalassemias and hemoglobinopathies, are prevalent, incurable, autosomal recessive hemoglobin illnesses that result in considerable morbidity and mortality, imposing a substantial financial burden on society. A straightforward blood test, such as the assessment of Mean Red Cell Corpuscular Volume (MCV), before to marriage can effectively identify carriers of certain disorders. Premarital health screening is conducted to educate couples about the likelihood of having impacted offspring and to provide them with suitable guidance. Premarital screening programs have gained widespread acceptance and significant benefit in preventive healthcare, leading many governments to mandate them. Numerous potent social factors affect the acceptability of preventive programs, including religious beliefs, cultural norms, literacy and education levels, government regulations, and the attitudes of individual couples. Scientists, medical professionals, politicians, intellectuals, religious leaders, educators, socio-cultural organizations, activists, and policymakers should collaborate to promote awareness of premarital screening testing and assist in the formulation and implementation of legislation for a premarital screening program in the country. Journalists from electronic and print media, along with physicians, midwives, genetic counselors, and clinical laboratory scientists engaged in pre-conceptional or prenatal care, can significantly contribute to raising awareness among individuals, particularly prospective life partners, about premarital screening programs.

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