



Genetic Disorders and Consanguinity in the Saudi Population

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Abstract

Saudi Arabia has one of the highest rates of consanguineous marriages in the world, which is strongly embedded in culture and has significant implications on the health of people. The practice has a great impact on increasing the prevalence of autosomal recessive genetic diseases, which place a significant burden on the national healthcare system and the economy. This study carries out a synthesis of recent evidence to understand the complex association between consanguinity and the genetic disease situation in Saudi Arabia. It focuses on which particular genetic illnesses this practice enhances, studies the current state of popular knowledge and feelings, and determines the efficiency of the existing preventive strategies, including the national premarital screening program. In addition, the socio-cultural mechanisms that promote consanguinity and the multi-faceted measures that can be taken to reduce the related health risks are critically discussed and proposed in the paper. The results highlight the need to have a national initiative where cultural sensitivity is taken into consideration and encourages genetic literacy and promotes knowledge about reproductive choices.

Introduction

The issue of the genetic disorders and consanguinity among the Saudi population is a multifaceted problem of interaction between genes, culture, and social health. Consanguinity marriage, or the marriage between people whose blood relationship is defined by second cousins or even closer and it is considered as a common cultural practice in most Arab and Muslim-majority states, with the highest levels reported in Saudi Arabia (Abdulhadi, 2018). The recent research continues to confirm the fact that more than half of all marriages in the Kingdom are consanguineous, with the most frequent ones being first-cousin marriages (Yousef et al., 2024; Alshamlani et al., 2024). Genetically, consanguinity predisposes the possibility of children inheriting the same recessive gene mutations from both parents and, therefore, significantly elevates the likelihood of manifesting autosomal recessive diseases (Khayat et al., 2024). This biological process is the primary cause of the high morbidity associated with rare and monogenic diseases in the Saudi population, transforming a cultural preference into a significant health issue.



The low rate of intermarriage has also directly led to a distinctive and intricate distribution of genetic diseases in Saudi Arabia, which places a future burden on the healthcare system because of the expensive burden of treating chronic inherited diseases (Yousef et al., 2024). This paper will attempt to present an in-depth examination of this problem by initially defining the narrow range of genetic disorders that are linked to consanguinity as described by studies in the field of genomics. It will then critically evaluate the status of the present perspective on the social-cultural awareness that informs the choice of marriage. It will also try to compare the available public health interventions, which mostly include the premarital screening program, and offer evidence-based interventions to decrease the occurrence of such disorders in the future and relieve the consequent burden on healthcare in the country.

The Spectrum of Genetic Disorders Enhanced through Consanguinity.

The clinical implications of consanguinity are deep-seated in Saudi Arabia, as the prevalence of autosomal recessive and monogenic diseases among the pediatric and adult population in Saudi Arabia is disproportionately high. This is the direct consequence of greater homozygosity of the deleterious alleles, and more, importantly, the genetic principle that, in clinical terms, is staring back at Saudi healthcare information which include greater concentrations of genetic diseases in families. Recent cross-sectional research of two years at one of the largest tertiary care centers in Riyadh found that 64 percent of all patients who were referred to genetic testing were children of consanguinity unions, showing a high relationship between the intermarriage of relatives and the development of an inherited disease (Alshamlani et al., 2024). It is remarkable to note that use of whole exome sequencing (WES) importantly, (WES), which is a complex molecular diagnostic method (WES), method, gave conclusive diagnosis in 45 percent of cases compared with the world average. This diagnostic yield indicates the enrichment of rare, harmful variants of the Saudi gene pool, which were mostly preserved during generations of marriage within the endogamous tradition (Alshamlani et al., 2024). The phenotypic consequences of these genetic patterns are broad and include neurodevelopmental, hematologic, metabolic, and multisystem disorders. Out of these, neurodevelopmental disorders come out as exceptional. They represented 18 percent of the total number of referrals in the Riyadh cohort and 35.7 percent in a larger national cohort (Alqahtani et al., 2023). These data highlight the physical fact that consanguinity does not simply augment genetic hazard in hypothetical perspective, but it is a tangible fact that it has a concrete manifestation in the form of a higher rate of severe and early-onset and frequently life-limiting morbidity in clinical practice. The integration of clinical genetics and population data, therefore, is a strong argument that the genetic effects of consanguinity are not just quantifiable but also have significant impact on the health sector of Saudi Arabia.

In addition to general correlations, national studies have been paying more attention to the determination of the most amplified genetic variants and disorders in the context of



consanguinity. A comprehensive study that reviewed clinical exome sequencing data found 37 shared the common autosomal recessive variants of 35 genes that are common in a variety of centers (Aleissa et al., 2022). The related disorders majorly impact the hematologic (32%), endocrine (21%), metabolic (11%), and immunologic (10%) systems which indicate the extent of genetic disease impact on the Saudi population. Among the most common inherited diseases in the Kingdom are the hematologic group, especially sickle cell anemia and thalassemia. This fact is in sharp contrast with the level of public awareness because, according to Yousef et al. (2024), a considerable percentage of citizens do not perceive these typical disorders as genetic. Genetic effects of consanguinity do not just end with blood disorders; as Albeshar et al. (2022) showed since consanguineous marriages have a significant impact on the risk of congenital heart disease (CHD) because the expression of rare recessive variants is encouraged. Equally, Aljabri et al. (2024) verified via systematic review that the rate of congenital sensorineural hearing loss is significantly higher in children of consanguineous couples. Notably, there is not even distribution of the genetic burden in Saudi Arabia. Local studies of regions show some variation that incorporates the historical gene pool as well as local cultural practices. Regarding the Albaha area, Albanghali (2023) reported that the consanguinity rate was 40 percent, and the level of cardiovascular, hematologic, ophthalmic, and cancer diseases among the children of close parents was significantly higher than that of the general population. This geographic variation highlights the importance of the existence of localized marriage practices, tribal endogamy, and geographic isolation in creating the unique genetic disease patterns within the Kingdom. These results collectively paint a complex and multidimensional picture where cultural maintenance, lesser geographical genes, and biomedical frailty overlap—and this presents complicated issues to national health policy on public health and genetic counseling.

Public Awareness, Knowledge and Attitudes

The paradox in the understanding of consanguinity is a common finding that national studies have recorded throughout Saudi Arabia despite the extensive awareness of consanguinity. In an unprecedented national survey conducted by Yousef et al. (2024) on over 9,000 individuals, it was found that despite the fact that about 85.45% of the study participants were aware that consanguinity marriages predispose individuals to genetic disorders, in many cases such awareness was superficial. The majority of the participants were unable to describe properly the principles of genetic heredity or name certain disorders that are associated with such marriages. It is important to note that almost sixty percent of men and forty-eight percent of women were not aware of sickle cell anemia and thalassemia as inherited conditions, although both are common in Saudi Arabia. Another significant finding in the study was the wide gender difference between the men (76.61%) and women (88.53%). This loophole poses critical health concerns to the people, because men are usually the decision-makers in marriage relations in the traditional patriarchal society. Therefore, lack of male awareness can become



the bane of adopting premarital screening and other preventive measures, and thus the continuation of inherited diseases in families and communities.

Education levels have continued to be among the best predictors of genetic literacy in Saudi society. As Basuliman et al. (2020) discovered, those respondents with medical or scientific experience had much more knowledge about the genetic risks of consanguinity than those with less education did. In a similar pattern, Jameel et al. (2024) established that learners in health-related sciences (especially women) expressed a more favorable response to premarital genetic testing and premarital counseling as prevention methods. These results highlight the transformational nature of education in increasing knowledge and influencing the masses on health behaviors. Nonetheless, the cultural and social forces are strong and limit the translation of awareness to behavior change. Bakry et al. (2023) also found that the social constructs and family customs still have a powerful impact on the marriage decisions that, in many cases, override the personal knowledge of the risks associated with genetics. Consanguinity marriage is still viewed in most societies as a way of keeping families together, enhancing tribalism, and ensuring economic sustainability. The continuation of the beliefs leads to a cultural inertia that is not affected by biomedical advice.

Interventions in Public Health: Genetic Counseling and Premarital Screening

In 2004 the consanguinity-related disorders had taken toll of health sector of Saudi Arabia and for this reason the government introduced National Premarital Screening (PMS) Program, which over the years has been expanded to include some of the infectious diseases and certain genetic disorders. This was done in an effort to lower the number of hereditary diseases due to early diagnosis of at-risk couples prior to getting married. The program has already made significant advances in increasing awareness and making people interested in genetic health problems. An example is Al Eissa et al.'s (2024) ruling that more than 91.3 percent of participants surveyed supported the need to raise awareness about genetic disorders among the population, which shows that genetic screening as a preventive intervention is widely accepted. In spite of such advances, there are still serious challenges. The biggest constraint of the PMS program is that the population does not understand the scope and objectives of the program. Yousef et al. (2024) conducted a survey among a huge national group to discover that just 6.87% of them are aware of the purpose of PMS and the diseases that it is designed to identify. This indicates that people are very aware of the program, but they do not have a good grasp of the preventive role of the program. In addition, the screening panel per se is limited in its area of focus since it is mainly focused on sickle cell anemia and thalassemia, which are not only prevalent but also only a fraction of the recessive diseases associated with Saudi families. Scientists like Albanghali (2023) and Aleissa et al. (2022) have suggested the addition of multiple testing panel items to cover other conditions common to consanguinity groups, like G6PD deficiency, cystic fibrosis, and some metabolic disorders. The PMS program



can easily face the threat of being a mere procedural obligation without such expansion and genetic counseling, which can change the face of public health by lowering the prevalence of inheritable diseases.

There are also interventions of public health through prenatal diagnosis (PND) and, in more complicated ethical situations, abortion of the fetus (TOP). A study by AbdulAzeez et al. (2019) has shown that the level of acceptance of PND by Saudi couples is 68% and indicates an increase in confidence in genetic technologies as diagnostic methods. Nevertheless, the readiness to use TOP in case of severe genetic abnormalities is significantly lower at 33%, where religious restrictions and ethical objections are the major issues. Termination is not allowed in general in the Islamic jurisprudence with certain medical conditions, which is the possible reason why the population does not want to follow this practice. However, shifting demographics are determining the gradual changes in attitude. AbdulAzeez et al. (2019) reported that non-consanguinity couples, couples with already affected children, and more educated couples had increased acceptance of PND and TOP. Generally, the results show that the government of Saudi Arabia has started to make progressive changes to ensure people can make sound decisions related to their reproductive health and creating knowledge on the dangers of consanguineous marriages when it comes to their health.

Discussion and Proposed Solutions

The overall evidence develops the complex picture of the genetic health situation in Saudi Arabia, that is, the presence of world-level scientific potential and a disproportionate transfer of this knowledge to societal consciousness and behavioral modification. In Saudi Arabia, the level of genetic research and diagnostic facilities has improved significantly, and hospitals and laboratories are technically literate. This knowledge, however, has not as yet trickled into the community level of understanding or decision-making, as recent studies point out. Although the rate of consanguinity marriages is declining with the national awareness campaigns and mandatory screening programs, it is too high, implying that the knowledge dissemination efforts are not enough to change the well-rooted and embedded cultural practices. Their longevity is partly attributed to the extent of cultural and social logic that consanguinity is based on. To most Saudi families, cousin marriage still represents family cohesiveness, societal confidence, and economic stability. Inter marriages between relatives are also thought to reinforce kinship ties and maintain the continuity of the lineage—values potentially more significant than the abstract of genetic risk (Jameel et al., 2024; Bakry et al., 2023). Within such a socio-cultural context, genetic disorders can be perceived as a tragic yet controllable consequence as opposed to a discouraging effect on consanguineous marriages. Moreover, as Kari et al. (2014) found, such a context of research or genetic intervention needs profound cultural and religious sensitivity. The ethical issues related to prenatal diagnosis (PND) and pregnancy termination (TOP) are especially complicated because Islamic



jurisprudence most often limits the termination to the instances of serious fetal malformation prior to ensoulment (120 days of gestation). This theological and ethical environment restricts the extent of some preventive alternatives that can be adopted in the Western context of biomedical spheres. Digital platforms have not been utilized to access the population, and with more than 99% of the Saudi population using social media, although half of them are using it to seek health information, it is an untapped resource (Yousef et al., 2024). These avenues can be used by health authorities and genetic professionals to create interactive, easy-to-understand material that not only includes the risks of consanguinity but also the process of inheritance, prevalent disorders, and how genetic counseling can lead to making informed decisions related to reproduction. Second, the Premarital Screening (PMS) Program must be increased and enhanced. Recessive conditions remain unidentified with current limitations that only screen sickle cell anemia and thalassemia. Scientists such as Aleissa et al. (2022) recommend the introduction of other genetic issues in the area and the free delivery of post-screening genetic counseling so that the couples fully understand their test outcomes and choices (Al Eissa et al., 2024). Third, inclusion of genetics education in the national curricular programs is essential. Saudi Arabia can raise a more genetically literate generation by providing modules on heredity, disease prevention, and genetic ethics in high schools and universities, especially for non-health students (Bakry et al., 2023). The result of such education is critical thinking and a decrease in genetic disorder stigma. Lastly, the increased genetic counseling workforce and the improvement of access to counseling services, in particular in rural areas, is also a critical issue. Mediators between biomedical information and cultural realities are trained counselors who assist the family to make painful decisions about reproductive choices. Integration of education, easy access to counseling, policy change, and culture-based communication is the most prospective path to sustainable genetic risk reduction in Saudi Arabia.

Conclusion

The interaction of consanguinity and genetic disorders in Saudi Arabia is the problem of critical social health in which culture, biology, and medicine intersect. The consanguinity of marriages has been shown to be high to an extent that it has resulted in an increased burden of autosomal recessive and monogenic diseases, which has affected thousands of families, which has brought a heavy cost burden on the healthcare system in the long term. Although there is a limited general knowledge about the subject where it is usually shallow and does not involve a thorough discourse of particular diseases or the mechanism of prevention programs such as premarital screening. Saudi Arabia can empower the citizens by using the advantages of modern communication tools, increasing and improving the screening and counseling services, and integrating the knowledge of genetics into the mass media which allow them to make well-informed decisions. These endeavors are hopeful in the mitigation of the occurrences of curable



genetic disorders and hence contribute to the health of children and lessen the healthcare burden on the national health system for the succeeding generations.

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