

Systematic Review

Attitude and awareness of the public toward genetic testing in Saudi Arabia

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ABSTRACT

Genetic disorders are prevalent in Saudi Arabia due to high rates of consanguinity, making public awareness and attitudes toward genetic testing critical for national health strategies. This systematic review synthesized studies published between 2020 and 2025 from PubMed, Google Scholar, ScienceDirect, and the Saudi digital library, focusing on awareness, perception, and sociocultural influences on genetic testing in Saudi Arabia. The findings reveal generally positive public attitudes toward premarital screening (PMS) and genetic testing, with stronger acceptance among individuals with higher education or a family history of genetic disorders. However, barriers remain, including cultural resistance to altering marriage plans, limited knowledge, and low trust in genetic services. Sociocultural and religious beliefs strongly influence decision-making, with many couples proceeding with high-risk marriages despite unfavorable screening results. The review underscores the importance of integrating culturally sensitive education, expanding genetic counseling services, and leveraging public health initiatives to bridge the gap between awareness and behavior.

Keywords: Genetic testing, Premarital screening, Consanguinity, Public awareness, Attitude toward genetic counselling, Saudi Arabia

INTRODUCTION

Genetic testing is one of the premarital counselling services, and it is a therapeutic intervention for couples who plan to marry and have a susceptibility to transmit the disordered genes to their offspring, and are used for tumour molecular characterization, preconception carrier screening, prenatal and postnatal atypical condition detection, and infectious illness diagnosis especially in consanguineous and tribal marriages, where spouses are related by ancestry.^{1,2} This is prevalent in Saudi Arabia for 60% and 70% and other Arab countries.³ The high rate of this kind of marriage is directly proportional to the elevated risk of genetic disorders and health repercussions due to consanguineous union gene mutations “(inherited genetic diseases)” when genetic diseases can be passed on

through generations and inherited from parents through the germ line.⁴ In some cases, this occurs during meiosis. In other cases, it is present in the parents’ cells that are passed onto the offspring.⁵ Resulting in disorders in their children such as cystic fibrosis, thalassemia, and Tay-Sachs disease. The expression of deleterious genes potentially leads to reduced immunity, and the accumulation of harmful mutations over generations can result in a higher prevalence of multifactorial diseases, such as heart disease, diabetes, and immunodeficiency disorders.⁴ And hemoglobinopathies and liver diseases (Wilson), polycystic kidney disease, other diseases can arise spontaneously during conception, and other genetic diseases involve genes that arise during an individual’s lifetime (mostly cancer).^{3,5} Familial and inherited cancers can present early; thus, screening for these cancers should be performed earlier than in the general population. Early

screening decreases cancer mortality and morbidity. Saudi women, especially older adults, have suboptimal knowledge about inherited and familial cancers and poor attitudes toward genetic screening.⁶

The public's attitude towards genetic testing worldwide for assessing disease risk is generally encouraging and positive. In one of the studies conducted in the US, it was reported that 97 percentages of total participants showed interest in the area of genetic testing, and most of the participants showed a positive attitude towards the use of genetic testing as a tool for the detection of diseases. A Dutch survey conducted by also reported a positive attitude of the population towards genetic testing, as per their study, 64 percentages of participants agreed that genetic testing can help people live longer. Another study conducted in African Americans revealed a positive view about genetic testing for preventive care and presymptomatic detection, with some concerns concerning privacy.⁷ On the other hand, in Palestine, there is insufficient knowledge about genetic counselling among Palestinians due to the high rates of consanguineous marriages.⁸

Several studies have shown that individual attitudes toward genetic testing are connected to their genomic literacy.⁸ Many statistics have been recorded from recent studies to determine the attitude toward genetic testing among the Saudi population. Many statistics indicate a positive attitude towards genetic testing from various regions in Saudi Arabia.² Other studies recorded that married people were more in favor of genetic testing than their single counterparts. Secondly, single-status people supported the idea of having genetic tests before their marriage. They choose abortion knowing that the embryo has a severe genetic disorder, neither marital status or gender. They were supportive of the idea of the government taking a leading role in enforcing genetic testing and the creation of the genetic disease database and family maps, and genetic banks.⁷ Particularly the Saudi university students. For instance, at King Abdulaziz university, Jeddah, Saudi Arabia, most of the students prefer genetic testing and premarital counselling in consanguineous marriages.⁹ And a high level of awareness about consanguineous marriage and its association with genetic disorders among female university students in Riyadh.³ In Jazan, premarital and prenatal testing are generally positive. Otherwise, only 29.6% agreed with the decision to divorce due to the higher probability of the genetic disease in their children.¹⁰

The concept of genetic counseling and genetic screening poses many challenges in the Islamic world, especially in the Kingdom of Saudi Arabia, as it must be implemented within the framework of religion and culture through the Islamic moral and cultural context of the people and society.¹⁰ Tribalism has long been a fundamental aspect of the social, cultural, and economic fabric in the Arabian Peninsula. Saudi tribes have historically prioritised the

preservation of community cohesion, identity, and patrimony by encouraging marriage alliances within the tribe, lineage, or sub-tribe.¹¹ In 2012, the knowledge and awareness in the general population about the PMSGC program was low. Including genetic testing.¹² Thus, the government of Saudi Arabia has made a considerable effort to reduce the risk of marriages between high-risk couples. However, despite this information, 90% of the couples proceed with marriage despite the potential risk of having affected offspring.¹¹ The present study, one of the largest national surveys in a highly consanguineous society, highlights that even the young and college-educated participants have low awareness of the genetic disease burden, which is strikingly high in all corners of the country. Low awareness among men was observed, and women are more aware of the carrier screening tests than men.¹³ In a specific society, the university students, the current study revealed a notable deficiency in the understanding of medical genetics among medical students and interns in Saudi Arabia, particularly regarding genetic inheritance and testing.¹⁴

Despite the high level of awareness, many people are reaching up to 90%, and still choose to get married despite the incompatible results. The reasoning behind their decision was due to their inability to cancel their plans for the wedding, emotions toward their partner, lack of awareness, religious reasons, and social stigma or, traditional beliefs, some new ideas are sometimes rejected by a community even if it is beneficial for the larger good of the community.¹⁵ Many factors affect the decision of the traditional communities, like the idea being way ahead of its time, the idea not being introduced properly, bad past experiences with similar ideas, the normal fear of everything new, and the tendency to resist change.⁷ Moreover; a previous study conducted on Arab Middle Eastern primary care practitioners revealed that the lack of knowledge and expertise about genetic testing was considered a major barrier against requesting such tests, thus education about genetic testing and counseling could be used to encourage the use of genetic services in the medical field, or even if genetic services are not in the place, genetic counseling could not be implemented as well.^{5,8}

Other factors influencing the recognition of the benefits of PMS tests, individuals with higher educational levels were more likely to recognize the benefits compared to others. A positive family history was positively associated with the recognition of PMS benefits.¹¹

Genetic testing is acceptable from the religious and ethical point of view as well as the economic requirements.¹ Current developmental trends in genomics have continuously led to the enhancement of the quality of life for several disorders by enabling the availability of cheap, easy, and fast genetic testing either through the primary health care system as the newborn screening panels or via direct-to-consumer (DTC) testing on the internet.⁷

Saudi Arabia introduced the first national PMS program (NPMS) in 2014 to reduce the risk of inherited disorders. This program helps limit the danger of genetic diseases passing down through the generations. In addition to genetic counseling, Saudi Arabia now offers clinical diagnostic, therapeutic, and preventative programs such as neonatal, premarital, and preimplantation genetic diagnosis (PGD).²

Since the official launch of the Saudi PMS program, few studies have examined its efficiency in reducing the rate of haemoglobinopathies, altering the attitudes of high-risk couples towards marriage.¹¹

The PMS and genetic counseling (PMSGC) program (currently called the healthy marriage program) tests couples planning to marry for common genetic disorders such as sickle cell disease (SCD) and thalassemia, which are the most common inherited diseases in humans. Hemoglobin disorders such as SCD and thalassemia affect more than 300,000 newborns each year, and unfortunately, this number is projected to increase in the upcoming years. The prevalence of SCD in KSA is estimated to be higher than 45,100 cases per 1,000,000 adults. The highest rates are observed in the Eastern region, followed by the Asir and Jazan regions. It detects infectious diseases as well, such as hepatitis B virus (HBV), hepatitis C virus (HCV), and human immunodeficiency virus (HIV). Around 2.58 million children were within the age group of 0e19.7 Daily in 2022, approximately 740 children were newly infected with HIV. The primary goal of the PMSGC program is to prevent or reduce the number of at-risk marriages, as well as to assess the size and distribution of those who are carriers or suffer from sickle cell anemia or the thalassemia. It also aims to make incompatible couples aware of their chances of having children with the diseases, as well as to offer them alternatives to the canceling their wedding.¹⁶

Added to the different genetic services within the Kingdom is the Saudi human genome program (SHGP), the largest genome initiative in the Middle East.² In 2013, the SHGP was launched. Although the project initially cost over USD 3 billion.¹⁷ Which aims to reduce and prevent genetic diseases from being required for the application of a marriage certificate.² Due to the high rate of consanguinity in Saudi Arabia (SA), which exceeds 60% of total marriages, and large family sizes, SA is an ideal country in which to discover novel variants. With the growth in genomic technology and understanding of the value of genetics, multiple countries-initiated genome projects such as the encyclopaedia of DNA elements (ENCODE). Many respondents revealed moderate awareness and attitude towards the SHGP and minimal knowledge regarding its benefits and applications. Respondents with new advances in genetic testing, differential decisions for the predisposition to diseases are becoming applicable for numerous diseases, e. g., dementia, diabetes, and cancer etc.^{7,17}

Therefore, the objective of this systematic review is to synthesize existing literature from 2020 to 2025 regarding public awareness and attitudes toward genetic testing in Saudi Arabia. This review aims to identify prevailing perceptions, sociocultural and religious influences, and the barriers and facilitators affecting public engagement with genetic services. By consolidating findings across multiple studies, the review seeks to provide comprehensive insights that can inform policy development, enhance educational efforts, and guide culturally appropriate genetic counseling practices in the Kingdom.

METHODS

This review utilized a narrative approach to synthesize available literature on public awareness and attitudes toward genetic testing in Saudi Arabia, with particular attention to consanguinity, PMS, sociocultural influences, and trust in genetic services. A comprehensive literature search was conducted across multiple electronic databases, including PubMed, Google Scholar, ScienceDirect, and the Saudi Digital Library, covering publications from January 2020 to May 2025. Search terms used in various combinations included: “genetic testing,” “public awareness,” “attitude,” “premarital screening,” “consanguinity,” “genetic counseling,” and “Saudi Arabia.”

Studies were included if they met the following criteria: peer-reviewed articles published in English or Arabic, studies focused on the Saudi population or involved comparative analyses within Arab or Islamic cultural contexts; and papers that explored aspects of public knowledge, perception, attitudes, or engagement with genetic testing or counseling. Exclusion criteria included non-human studies, non-peer-reviewed sources (e.g., editorials, opinion pieces), abstracts without full-text availability, and studies lacking relevance to the research objective.

A total of 124 records were initially retrieved through database searches, and 10 additional sources were identified through reference tracking and manual searches. After the removal of duplicates, 120 unique articles remained. These were screened based on titles and abstracts, leading to the exclusion of 60 studies. The remaining 60 full-text articles were assessed for eligibility. After excluding 23 studies due to reasons such as irrelevance, lack of original data, or poor methodological clarity, 37 articles were selected for inclusion in the final analysis.

The selected studies were grouped thematically and analyzed under the following domains: public attitudes, knowledge and awareness, socio-cultural and religious influences, trust in genetic services, and the impact of national screening programs. A PRISMA-style flowchart illustrating the selection process is provided below to ensure transparency.

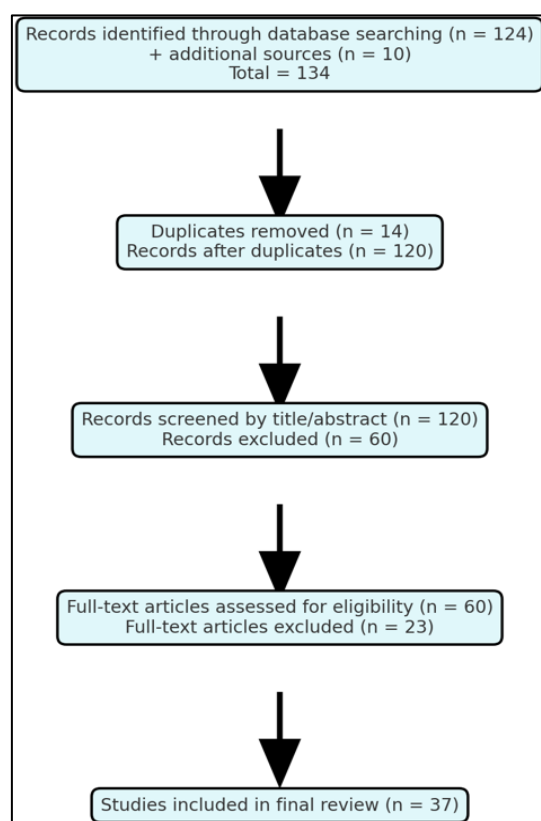


Figure 1: PRISMA flow chart.

RESULTS

Attitudes toward genetic testing in Saudi Arabia

The majority of studies reported generally positive attitudes toward genetic testing, especially for the prevention of inherited diseases. National data indicate that more than 83% of participants considered PMS important for protecting future generations. In Jazan, nearly 80% supported mandatory premarital testing, and 67% expressed willingness to undergo testing in future. Central region data showed similarly strong support, with 91% of respondents emphasizing the need for greater awareness and 87% supporting enhanced governmental involvement in reducing genetic health risks.

Demographic factors such as higher education, income, and family history of genetic disorders were positively associated with favorable attitudes. Among university students, clinical majors expressed more positive perceptions compared to non-clinical peers. However, despite widespread approval of PMS, emotional and cultural influences—such as family expectations and social stigma—often prevented individuals from altering marriage plans based on high-risk results.

Awareness of genetic testing in Saudi Arabia

Awareness of genetic testing and PMS programs was high, but in-depth knowledge remained limited. While

nearly all participants reported familiarity with PMS, only about 9% demonstrated satisfactory knowledge of its scope and implications. Many respondents were unclear about which conditions genetic testing could detect. For example, although female university students in Riyadh were aware of the link between consanguinity and genetic disorders, they lacked detailed knowledge of inheritance patterns or disease specifics.

Educational attainment was the most consistent predictor of awareness. Respondents with university/ postgraduate education were more than twice as likely to show adequate knowledge compared to those with only secondary education. Similarly, medical and science students consistently demonstrated stronger understanding than peers in non-scientific fields. Despite this, misconceptions persisted, including limited recognition of prevalent genetic disorders such as sickle cell anemia and thalassemia.

Trust and barriers

Trust in genetic services emerged as a major concern. Although many respondents supported PMS and genetic testing, significant proportions expressed doubts about the reliability and privacy of these services. For e. g. in Jazan, 79.1% supported mandatory PMS, yet only 2/3rd willing to undergo tests themselves. Couples often proceeded with high-risk marriages due to cultural expectations, emotional factors/desire to preserve tribal ties.

The lack of trained genetic counselors further contributed to hesitancy. In many cases, counseling was provided by general practitioners rather than specialized professionals, undermining confidence in the quality of advice. In addition, cultural and religious beliefs strongly influenced responses to ethically sensitive issues such as pregnancy termination and PGD, which many participants considered inconsistent with religious values.

Impact of national programs

Saudi Arabia's initiatives, including the healthy marriage program and the SHGP, were widely recognized but not fully understood by the public. The PMS program contributed to a decrease in the prevalence of β -thalassemia, though sickle cell anemia rates remained high. Despite these advances, many couples with incompatible results chose to proceed with marriage, highlighting the enduring impact of cultural norms.

Awareness of the SHGP was moderate, but detailed knowledge of its objectives and applications was limited. While participants often expressed willingness to participate in genetic research, concerns about ethical and cultural implications constrained full acceptance.

Attitude

Many studies in Saudi Arabia indicate that the public generally holds a positive attitude toward genetic testing, particularly for the prevention of inherited diseases. For instance, a national survey revealed that over 83% of participants considered PMS important, despite a limited understanding of the subject.¹⁸ In the Jazan region, nearly 80% supported mandatory premarital testing and expressed willingness to undergo testing in the future. However, fewer participants felt comfortable with more

sensitive implications, such as ending a marriage or terminating a pregnancy due to genetic risks.¹⁰ Another study demonstrated that individuals with higher education, income levels, and access to information were significantly more likely to view genetic testing favorably.² In central Saudi Arabia, approximately 91% of respondents agreed that public awareness of genetic diseases should be enhanced, and 87% supported increased government involvement in reducing genetic health risks—even though traditional practices, such as consanguineous marriage, remained common.¹¹ Among college students, attitudes were generally positive, though uncertainty remained regarding ethically sensitive issues like abortion in the case of severe genetic disorders, reflecting the influence of personal beliefs on perceptions of genetic testing.¹⁹

Overall, many individuals in Saudi Arabia express positive attitudes toward genetic testing, particularly when its health benefits are communicated. A national study involving over 6,000 participants found that more than 83% supported PMS and regarded it as important for protecting future generations from genetic diseases.¹⁸ In the Jazan region, approximately 67% of respondents were willing to undergo genetic testing in the future, and nearly 80% favored making such testing mandatory before marriage.¹⁰ These favorable attitudes were significantly associated with higher education levels, greater income, and increased awareness.² Among university students, those enrolled in clinical majors tended to have more positive perceptions than their peers in non-clinical fields, likely due to greater exposure to genetic concepts in their curricula.¹ Additionally, students with a family history of inherited disorders or consanguineous marriages showed increased interest in and acceptance of genetic testing, suggesting that personal experience can strongly influence public attitudes.²⁰

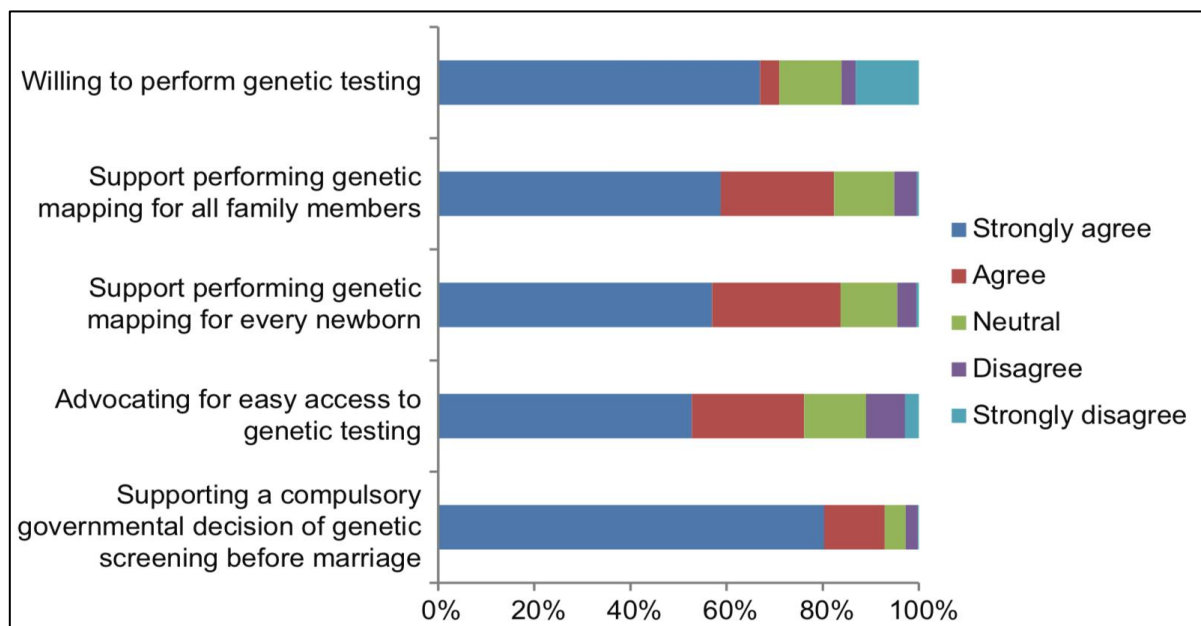


Figure 2: Participant's attitude toward genetic testing.¹⁰

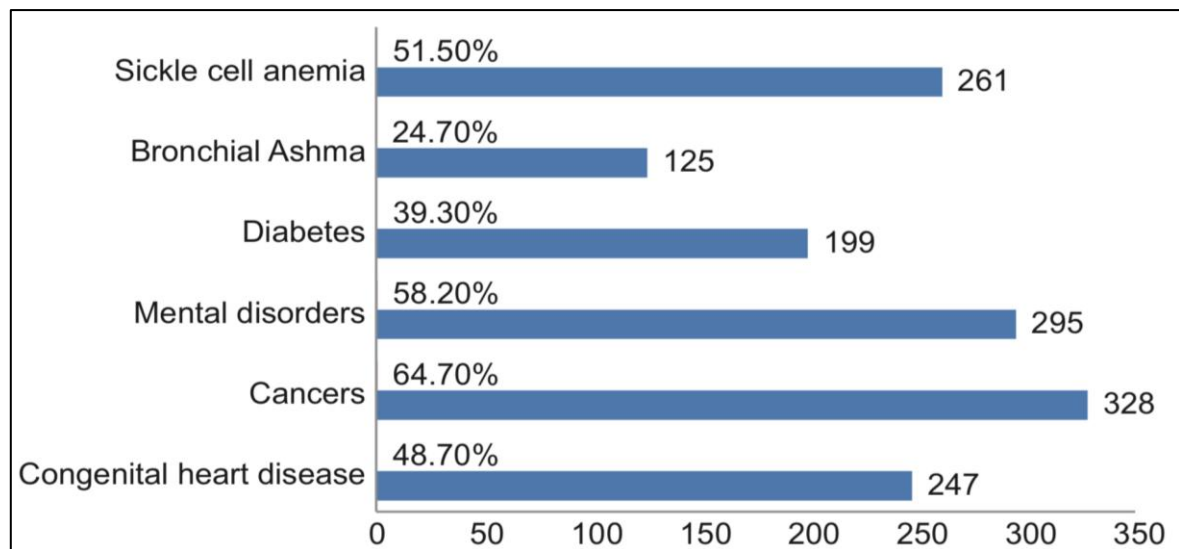


Figure 3: The common diseases that participant's most need to detect early through genetic testing, (n=507).

Despite these encouraging findings, several studies have identified persistent fears and misconceptions that shape public perceptions. For example, while many individuals supported genetic screening, they remained uncomfortable with certain implications, such as canceling a marriage based on incompatible test results.¹¹ Emotional factors, family expectations, and fear of social stigma often influence such decisions. A cross-sectional study of college students revealed that although most believed in the value of testing, many were uncertain about how to interpret the results or act upon them.¹⁹ Moreover, while 91% of participants in a national study endorsed expanding awareness campaigns, misconceptions remained—such as the belief that genetic testing could eliminate inherited diseases.¹¹ These findings highlight a gap between general support for genetic testing and a nuanced understanding of its scope and limitations. Strengthening education, particularly in schools and healthcare settings, may further enhance public confidence and informed decision-making regarding genetic services.^{2,10}

In Saudi Arabia, cultural and religious beliefs significantly influence public attitudes toward genetic testing. Even when individuals are informed about the risks of inherited diseases, many continue to favor consanguineous marriages due to deeply rooted cultural traditions and societal expectations.¹¹ One study reported that nearly 90% of couples proceeded with marriage despite receiving high-risk genetic results, primarily due to familial pressure and a strong desire to preserve tribal ties.²¹ This suggests that cultural allegiance often takes precedence over medical recommendations.

Religious beliefs also play a critical role in shaping attitudes toward certain genetic interventions. Options such as pregnancy termination and PGD are often viewed as conflicting with divine will, even when they offer the potential to prevent severe hereditary conditions.¹⁹ In a

large national survey, although the majority of participants supported PMS, many were hesitant to alter marriage plans based on unfavorable results.¹⁸ Similarly, a study from the Jazan region found that while there was general support for genetic testing, decisions involving divorce or abortion were less accepted, even in the presence of significant genetic risk.¹⁰ These findings highlight the ongoing tension between growing awareness and the persistent influence of cultural and religious norms on responses to genetic information.

In the Eastern Province, public perception of PGD remains limited, despite the high prevalence of chromosomal abnormalities in the region, such as SCD. Increasing public understanding and acceptance of such technologies could contribute to reducing the burden of inherited disorders. Given that Saudi public opinion is often shaped by cultural and religious values, educational and healthcare initiatives must be sensitive to these perspectives.²²

The present study indicated that respondents held generally positive attitudes toward PMS. A majority reported that they would not proceed with marriage if found to be carriers or affected by a hereditary condition. Additionally, most participants agreed that it would be inappropriate to marry in cases of incompatible PMS results. These findings are consistent with previous studies conducted in the Saudi population.²³

A key finding of this study is the overall positive attitude among participants toward the use of PGD for various medical conditions, as reflected by relatively high attitude scores. Statistical analysis revealed that certain demographic factors—such as region of origin or knowing someone with prior experience of PGD—were significantly associated with more favorable views. In contrast, variables such as gender, age, and educational level were not significantly correlated with attitudes toward PGD.²¹

Trust remains a critical factor shaping public engagement with genetic services in Saudi Arabia. Although awareness of genetic testing is increasing, concerns persist regarding the reliability, privacy, and consequences of these services. For instance, a national study involving healthcare professionals highlighted challenges such as unclear clinical guidelines and insufficient patient education, which may hinder broader acceptance of personalized genetic testing.²⁴ In Jazan, while 79.1 percentages of participants supported mandatory PMS, only about two-thirds indicated willingness to undergo testing themselves, suggesting lingering hesitation and distrust.¹⁰ Similarly, in Jeddah, many individuals proceeded with high-risk marriages despite unfavourable screening outcomes. This gap between awareness and behavior likely reflects skepticism about the utility and impact of genetic test results.¹²

Concerns regarding privacy and the management of genetic information significantly impact public trust in genetic services. A study conducted in Riyadh found that over 87% of respondents supported PMS, and more than 80% expressed willingness to participate in genetic research studies. However, when faced with sensitive decisions-such as abortion in cases of severe genetic disorders-many participants expressed reluctance.^{25,19} This indicates that although general support exists, trust may falter when individuals are uncertain about how genetic information might be used or interpreted.

Another factor affecting trust is the limited availability of trained genetic counsellors in Saudi Arabia. In most cases, patients receive genetic advice from general medical practitioners rather than specialized professionals, which may compromise the perceived accuracy and quality of information.²⁶ To enhance trust, genetic services must be delivered by well-qualified professionals who can offer clear, compassionate, and culturally appropriate counselling.

Cultural and familial dynamics also influence the acceptance of genetic guidance. In Saudi society, decisions regarding genetic testing-particularly those involving marriage-often involve extended family members rather than the individual alone. Despite the well-documented risks, consanguineous marriages remain common, reflecting a cultural emphasis on family cohesion and tradition over medical advice.^{27,28}

In some instances, couples proceed with marriage even after receiving high-risk genetic results. This behaviour is not necessarily a rejection of science but is often rooted in social norms, emotional bonds, or concerns about stigma. Building lasting trust in genetic services, therefore, requires more than offering access to testing. It necessitates active community engagement, respect for cultural values, as well as the delivery of information that is not only accurate but also empathetic and empowering.¹²

Awareness

Genetic testing and PMS programs have become increasingly common in Saudi Arabia; however, public understanding of these services remains limited. While many individuals report having heard of genetic testing-primarily through national campaigns or social media-they often lack a clear understanding of its purpose or clinical implications. For example, a national study found that although nearly all respondents were aware of PMS, only about 9% demonstrated a satisfactory level of knowledge about it.¹⁸ Similarly, a study in Jazan revealed broad support for premarital genetic testing, yet many participants were unsure about which conditions it could detect or how the process works.¹⁰ Among female university students in Riyadh, awareness of the relationship between consanguinity and genetic disorders was high, but few could identify specific diseases or explain modes of inheritance.³ Across various demographics, there is a general awareness that consanguineous marriage increases the risk of genetic disorders, yet this understanding is rarely connected to the purpose of genetic testing.^{2,29} Even students in health-related academic programs often had only a basic grasp of genetic principles, and many were unfamiliar with the clinical applications of genetic tests.^{19,20} Awareness appears to be influenced by sociodemographic factors, with individuals who have higher education levels or a family history of genetic disorders tending to be more informed.^{2,15,18} Nonetheless, misconceptions persist; for instance, many do not recognize conditions such as sickle cell anemia or thalassemia as genetic, despite their high prevalence in Saudi Arabia.^{29,30} This gap between visibility and comprehension suggests that while awareness campaigns have been effective in raising public interest, more targeted education is necessary to promote informed decision-making.

Educational attainment has consistently been identified as a key determinant of public knowledge and attitudes toward genetic testing in Saudi Arabia. Several studies have shown that individuals with university or postgraduate education are significantly more likely to understand the role of genetic screening and counselling in disease prevention. A nationwide survey found that respondents with university degrees were more than twice as likely to exhibit adequate knowledge of PMS and genetic disorders compared to those with only secondary education.¹⁸ Likewise, a study among university students revealed that those in medical or science faculties had a stronger grasp of genetic risks and inheritance patterns than students in non-scientific disciplines, highlighting the importance of curricular content in shaping awareness.¹⁹ Education has also been linked to greater acceptance of preventive actions; for instance, more educated individuals-particularly women-were more willing to avoid consanguineous marriages if genetic risk was present.^{3,15} Moreover, higher education levels were associated not only with awareness but also with proactive behaviours, including a greater willingness to

undergo genetic testing, seek counselling, and recommend these services to family members.^{10,20} In the Jazan region, support for mandatory premarital testing was significantly higher among college-educated participants than those with lower educational backgrounds.¹⁰ A cross-regional study similarly found that individuals with advanced education were more likely to consult official health sources for genetic information, rather than relying solely on informal channels like social media.² This pattern is consistent with findings from female university populations in Riyadh, where higher education levels were associated with reduced acceptance of cultural misconceptions surrounding consanguinity.³ Collectively, these findings underscore the critical role of education in fostering informed attitudes and supporting public health strategies aimed at reducing the burden of genetic disorders.

There is a need for more active awareness programs to inform the public about the advantages and disadvantages of consanguineous mating, to reduce the prevalence of consanguinity in a population where great preference is given to family traditions and values.³⁵ Further research is needed to study the level of knowledge and attitude towards consanguineous marriage and its related factors among educated Saudi adults, to understand how to decrease its rate to prevent the possible negative consequences for offspring.³⁶ Consanguinity, particularly among first cousins, is an added risk factor for these families, particularly in societies considered a common cultural practice, as confirmed in previous studies conducted in Saudi Arabia and other countries. Through comprehensive genetic testing of affected families, we have been better able to understand the genetic basis of the various cardiac lesions and to delineate the molecular mechanisms involved in cardiac morphogenesis.³⁷ Earlier studies highlighted the significantly limited awareness of the higher prevalence of genetic disease due to consanguinity, even among educated Arabs. In Saudi Arabia, more than 50% of marriages are between first cousins. This national study aims to gauge the level of public awareness regarding consanguinity and its impact on the prevalence of genetic diseases across Saudi Arabia.²⁹

The most common genetic testing in SA compared to worldwide

Saudi Arabia introduced mandatory PMS (The healthy marriage program) for selected diseases, including sickle-cell anaemia, thalassemia, and a few infectious diseases. A five-year study to assess the effect of PMS on the prevalence of thalassemia and SCD reveals that the rate of B-thalassemia decreased while sickle cell anaemia remained high.

The catalogue of transmission genetics in Arabs (CTGA) of the centre for Arab genome studies (CAGS) recorded the highest proportion (50-60%) of recessive disorders from Saudi Arabia among all Arab nations.¹³

Genetic testing in Saudi Arabia

Multigene panels

The first instances of recessive inheritance of previously assumed strictly dominant disorders (involving ITPR1, VAMP1, MCTP2, and TBP). Multigene panels accounted for 672 tests.³¹

Whole-exome sequencing

Whole-exome sequencing represented the remaining 347 tests. Pathogenic or likely pathogenic variants that explain the clinical indications were identified in 34% (27% in panels and 43% in exomes).³¹

A total of 867 molecular genetics tests were conducted on 738 probands. These tests included 610 exome sequencing (ES) tests, four genome sequencing (GS) tests, 82 molecular panels, 106 single-nucleotide polymorphism (SNP) arrays, four methylation studies, 58 single-gene studies, and three mitochondrial genome sequencing tests. The diagnostic yield among molecular genetics studies was 41.8% in ES, 24% in panels, 12% in SNP array, and 24% in single-gene studies. We found a significant correlation between the ES detection rate and positive consanguinity.³²

NIPT

We developed and validated a highly sensitive and specific whole-genome amplification approach for cfDNA purification and enrichment to screen pregnant women at high risk of having a child with family-specific AR disease. Our approach could overcome limitations such as maternal genomic interference and low fetal fractions. Furthermore, we demonstrated the feasibility of applying this strategy to pregnant women in the first trimester. The developed method could facilitate substantial advances in detecting and preventing several family-oriented AR diseases, especially in highly consanguineous marriage populations.³³

PMS test

PMS is an amazingly effective tool to prevent various genetic disorders in societies with a high incidence of consanguinity. It is a form of advice offered to expected couple regarding chance of transmission of profoundly serious and debilitating disorders like thalassemia, SCD, and other hemoglobinopathies in the coming generations.⁹

Table 1: PMS.

Diseases	N	Percentage (%)
Sexually transmitted	1092	64
Genetic disorders	510	29.9
Inherited metabolic disorder	55	3.2
Infective viral disorder	50	2.9

Due to the high prevalence of hereditary disorders, the Saudi PMS program was established in 2004 as a mandatory screening initiative for couples planning to marry. The screening protocol targets diseases that pose a significant health burden on both the government and individuals, aiding in the assessment of future family planning decisions. Priority diseases include hemoglobinopathies, such as thalassaemia and sickle cell anaemia and infectious diseases, such as hepatitis B and C and HIV. The objective of this screening test is to provide the carrier or affected couples with the option to proceed with the marriage while fully understanding the likelihood of their children being affected by the inherited condition and to refer them to a genetic counsellor (GC) for further guidance.¹¹

The country introduced the first NPMS in 2014 to reduce the risk of inherited disorders similar to hereditary haemoglobin abnormalities. This program helps limit the danger of genetic diseases passing down through the generations. Despite being required for the application of a marriage certificate, all couples with marriage proposals have the freedom not to complete their marriage, whatever the results of the NPMS. Saudi Arabia now offers clinical diagnostic, therapeutic, and preventative programs such as neonatal, premarital, and PGD. Added to the different genetic services within the Kingdom is the SHGP, the largest genome initiative in the Middle East, which aims to reduce and prevent genetic diseases.²

Mandatory PMS for conditions like sickle-cell anaemia and β -thalassaemia in Saudi Arabia represents a shift toward informed decision-making in marriages, rather than outright prevention of unions at genetic risk. This approach, particularly for hemoglobinopathies and thalassaemia, has gained acceptance due to the simplicity and affordability of tests like haemoglobin electrophoresis. However, extending PMS to include other prevalent recessive disorders hinges on the development of molecular genetic screening techniques.⁴

Next-generation sequencing (NGS)

Based on preimplantation genetic testing for aneuploidies, techniques came into use. This technique is essential for successful embryo transfer and accomplishing pregnancy, thus reducing the time and cost of additional cycles. NGS-based PGT-A is an applicable and reliable technique for routine embryo screening, especially for couples suffering from recurrent miscarriages or multiple embryo transfer failures.³⁹

Genetic testing worldwide

Prenatal whole-exome sequencing

It has the potential to increase the ability to provide more diagnostic capabilities in fetuses with sonographic abnormalities, which will then improve the ability to counsel families.³⁴

Genomic testing

It is now being used for tumour molecular characterization, preconception carrier screening, prenatal and postnatal atypical condition detection, and infectious illness diagnosis.²

Next-generation sequencing

It is a new technology used for DNA and RNA sequencing and variant/mutation detection. NGS can sequence hundreds or thousands of genes or a whole genome in a short period. NGS testing, “liquid biopsy,” can also be used in non-invasive prenatal testing. The NGS assay has important applications in hereditary cancer diagnosis or risk population assessment. Since the variant allelic frequencies of hereditary cancers are usually around 50% or 100%, the detection of such genetic changes needs less depth of sequencing when compared to those in cancers.

DISCUSSION

This review highlights a paradox within the Saudi population: while attitudes toward genetic testing are generally positive, actionable understanding and behavioural follow-through remain limited. Most participants across studies expressed strong support for PMS programs and acknowledged their importance in preventing inherited diseases.^{10,11,18} However, this support was not consistently translated into practice, as many couples proceeded with marriage despite high-risk results, largely due to cultural traditions, family expectations, and social stigma.^{11,21}

Our findings are consistent with national surveys reporting that although awareness of PMS exceeds 90%, only a minority of respondents possess in-depth knowledge of its scope and implications.¹⁸ Al-Shroby et al found that only 9% of participants had adequate knowledge of PMS and counselling despite widespread awareness.¹⁸ Similarly, Alahdal et al observed that even among university students, awareness of genetic disorders was high, yet detailed understanding of inheritance and risk interpretation remained limited.³

International comparisons reveal similar patterns. A US study reported that 97% of respondents were interested in genetic testing for preventive purposes.⁷ Likewise, a Dutch survey indicated that 64% believed genetic testing could help people live longer.⁷ However, concerns about privacy and result interpretation were common, paralleling Saudi respondents’ hesitations.²⁵ In Palestine, despite high rates of consanguinity, public knowledge of genetic counseling was insufficient, resembling Saudi trends.⁸

Cultural and religious beliefs emerged as strong determinants of behavior. Even when participants were aware of genetic risks, consanguineous marriages often

continued, reflecting deeply rooted traditions.^{11,27,28} Options such as PGD and pregnancy termination were frequently perceived as conflicting with religious values, limiting their acceptance.^{19,21} Alawad et al reported similar findings, showing that while PGD was generally supported, abortion remained widely rejected.²¹ These findings align with global evidence that ethical and cultural contexts shape the acceptance of genetic interventions.²²

Trust in genetic services was another major barrier. Respondents expressed concerns about reliability, privacy, and the consequences of genetic results.^{12,24} The shortage of trained genetic counsellors further exacerbated this issue, as many individuals relied on general practitioners for guidance.²⁶ Shenbagam et al emphasized that in the Middle East, limited counselling infrastructure and cultural sensitivities restrict effective delivery of genetic services.²⁶ This lack of trust undermines uptake, even when public interest exists.

National initiatives such as the healthy marriage program demonstrated measurable impact, reducing the prevalence of β -thalassemia though with limited effect on sickle cell anaemia.¹⁶ The SHGP, while widely recognized, was poorly understood by the public, underscoring the need for sustained community education and transparent communication regarding its objectives and benefits.¹⁷

Overall, these results suggest that while Saudi Arabia shares many challenges with global populations, its unique context of high consanguinity and strong cultural-religious influences necessitates tailored interventions. Education, culturally sensitive counselling, and improved professional infrastructure will be essential to bridge the gap between awareness and informed decision-making.

CONCLUSION

This review highlights that while public attitudes toward genetic testing and PMS in Saudi Arabia are generally favourable, significant gaps remain in knowledge, interpretation, and behavioural follow-through. These discrepancies are shaped by complex sociocultural dynamics, including consanguineous marriage traditions, religious considerations, and varying levels of trust in healthcare services. Although national initiatives such as the healthy marriage program and the Saudi human genome project demonstrate commendable efforts, their long-term success depends on integrating genetic literacy into public education and ensuring accessible, culturally competent counselling services. Moving forward, targeted health communication strategies, enhanced training of primary care providers, and community-engaged policymaking will be critical to bridge the divide between awareness and informed action in genetic health.

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REFERENCES

- Osman HT, Baraia ZAA, Abdelati IH. Awareness and attitude of university students regarding premarital counseling and examination. *Trends Nurs Health Care J*. 2021;3(2):18-43.
- Alotaibi A, Alkhalidi NK, Al Nassir AM, Al Ayoubi LA, Al Malki NA, Almughyiri RA, et al. Exploring People's Knowledge of Genetics and Attitude towards Genetic Testing: A Cross-Sectional Study in a Population with a High Prevalence of Consanguinity. *Healthcare (Basel)*. 2022;10(11):2227.
- Al Ahdal H, Al Shanbari HM, Al Mazroa H, Al Ayesh S, Al Rhaili A, Al Qubi N, et al. Consanguinity, awareness, and genetic disorders among female university students in Riyadh, Saudi Arabia. *J Biochem Clin Genetics*. 2021;4(1):27-34.
- Khayat AM, Alshareef BG, Alharbi SF, AlZahrani MM, Alshangity BA, Tashkandi NF. Consanguineous Marriage and Its Association with Genetic Disorders in Saudi Arabia: A Review. *Cureus*. 2024;16(2):e53888.
- Niyibizi JB, Rutayisire E, Mochama M, Michael H, Zephania N, Daniel S. Awareness, attitudes towards genetic diseases and acceptability of genetic interventions among pregnant women in Burera district, Rwanda. *BMC Public Health*. 2023;23(1):1961.
- Alghuson L, Alturki NI, Alsulayhim AS, Alsughayer LY, Akkour KM. Awareness, Knowledge, Perceptions, and Attitudes towards Familial and Inherited Cancer. *Medicina (Kaunas)*. 2022;58(10):1400.
- Arafah A, AlJawadi MH, Aldheefi M, Rehman MU. Attitude and awareness of the public towards genetic testing in Riyadh, Saudi Arabia. *Saudi J Biol Sci*. 2021;28(1):255-61.
- Ghanim M, Rabayaa M, Alqub M, Ahmad H, Mohammad A, Belal R, et al. Investigating knowledge and attitudes toward genetic testing and counseling among Palestinians. *Sci Rep*. 2025;15:4446.
- Jameel T, Baig M, Murad MA, Gazzaz ZJ, Mal Y, Alyoubi WE, et al. Consanguineous marriages, premarital screening, and genetic testing: a survey among Saudi university students. *Front Public Health*. 2024;12:1328300.
- Eltyeb EE, Alhazmi SA, Maafa SHI, Mobarki SJ, Sobaikhi NH, Sumayli RA, et al. Public's perception and attitude toward genetic testing in the Jazan region. *J Family Med Prim Care*. 2024;13(10):4715-20.
- Al Eissa MM, Almsned F, Alkharji RR, Aldossary YM, AlQurashi R, Hawsa EA, et al. The perception of genetic diseases and premarital screening tests in the central region of Saudi Arabia. *BMC Public Health*. 2024;24(1):1556.
- Ibrahim NK, Bashawri J, Al Bar H, Al Ahmadi J, Al Bar A, Qadi M, et al. Premarital Screening and

- Genetic Counseling program: knowledge, attitude, and satisfaction of attendees of governmental outpatient clinics in Jeddah. *J Infect Public Health*. 2013;6(1):41-54.
13. Yousef NA, ElHarouni AA, Shaik NA, Banaganapalli B, Al Ghamdi AF, Galal AH, et al. Nationwide survey on awareness of consanguinity and genetic diseases in Saudi Arabia: challenges and potential solutions to reduce the national healthcare burden. *Hum Genomics*. 2024;18(1):138.
14. Zakariyah AF, Alamri SA, Alzahrani MM, Alamri AA, Khan MA, Hanbazazh MA. Identifying knowledge deficiencies in genetics education among medical students and interns in Saudi Arabia-A cross-sectional study. *BMC Med Educ*. 2024;24(1):778.
15. Alwhaibi RM, Almuwais AK, Alotaibi M, Al Taleb HM, Alsamiri SM, Khan R. Health beliefs of unmarried adult Saudi individuals toward safe marriage and the role of premarital screening in avoiding consanguinity: a nationwide cross-sectional study. *Front Public Health*. 2024;12:1379326.
16. Al Zuayr SN, Sulimani SM, Abd-Ellatif EE. Outcomes of the premarital screening program in Riyadh Region, KSA in 2021-2022: A cross-sectional study. *J Taibah Univ Med Sci*. 2024;19(5):1067-74.
17. Alrefaei AF, Hawsawi YM, Almaleki D, Alafif T, Alzahrani FA, Bakhrebah MA. Genetic data sharing and artificial intelligence in the era of personalized medicine based on a cross-sectional analysis of the Saudi human genome program. *Sci Rep*. 2022;12(1):1405.
18. Al-Shroby WA, Sulimani SM, Alhurishi SA, Bin Dayel ME, Alsanie NA, Alhraiwil NJ. Awareness of Premarital Screening and Genetic Counseling among Saudis and its Association with Sociodemographic Factors: a National Study. *J Multidiscip Healthc*. 2021;14:389-99.
19. Olwi D, Merdad L, Ramadan E. Knowledge of Genetics and Attitudes toward Genetic Testing among College Students in Saudi Arabia. *Public Health Genomics*. 2016;19(5):260-8.
20. Jairoun AA, Al-Hemyari SS, Shahwan M, Karuniawati H, Zyoud SH, Abu-Gharbieh E, et al. Assessing Students' Knowledge and Attitudes Regarding the Risks and Prevention of Consanguineous Marriage: A Cross-Sectional Online Survey. *J Multidiscip Healthc*. 2024;17:1251-63.
21. Alawad N, Alangari A, Allhybi A, Masud N, Almuzaini F, Alshamari M, et al. Assessment of attitudes towards the use of preimplantation genetic diagnosis in a single center in Riyadh, Saudi Arabia. *J Genet Couns*. 2023;32(5):1032-9.
22. Tehsin F, Almutawah FH, Almutawah HK, Alwabari ME, AlSultan ZM, Buawadh HS. Preimplantation Genetic Testing: A Perceptual Study from the Eastern Province, Saudi Arabia. *Cureus*. 2021;13(12):e20421.
23. Al Otaiby S, Alqahtani A, Saleh R, Mazyad A, Albohigan A, Kutbi E. Comprehension of premarital screening and genetic disorders among the population of Riyadh. *J Taibah Univ Med Sci*. 2023;18(4):822-30.
24. Uddin M, Deeb A, Ardah H, Nashbat M, Ayoub MY, Alfadhel M. A nationwide cross-sectional study in Saudi Arabia for the assessment of understanding and practices of clinicians towards personalized genetic testing. *Sci Rep*. 2024;14(1):31748.
25. Arafah A, Al Jawadi MH, Al Dheefi M, Rehman MU. Attitude and awareness of the public towards genetic testing in Riyadh, Saudi Arabia. *Saudi J Biol Sci*. 2021;28(1):255-61.
26. Shenbagam S, Taylor A, Jain R, Fakhro K, Alkuraya F, Abou Tayoun A. Genetic counseling in the Middle East: provider perspectives of patient attitudes and cultural challenges. *Hum Genomics*. 2025;19(1):59.
27. Modell B, Darr A. Science and society: genetic counselling and customary consanguineous marriage. *Nat Rev Genet*. 2002;3(3):225-9.
28. Bakry H, Alaiban RA, Alkhyat AA, Alshamrani BH, Naitah RN, Almoayad F. Predictors of Consanguinity Marriage Decision in Saudi Arabia: A Pilot Study. *Healthcare (Basel)*. 2023;11(13):1925.
29. Yousef NA, ElHarouni AA, Shaik NA, Banaganapalli B, Al Ghamdi AF, Galal AH, et al. Nationwide survey on awareness of consanguinity and genetic diseases in Saudi Arabia: challenges and potential solutions to reduce the national healthcare burden. *Hum Genomics*. 2024;18(1):138.
30. Al Sulaiman A, Suliman A, Al Mishari M, Al Sawadi A, Owaidah TM. Knowledge and attitude toward the hemoglobinopathies premarital screening program in Saudi Arabia: population-based survey. *Hemoglobin*. 2008;32(6):531-8.
31. Monies D, Abouelhoda M, Al Sayed M, Al Hassnan Z, Alotaibi M, Kayyali H, et al. The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. *Hum Genet*. 2017;136(8):921-39.
32. Alqahtani AS, Alotibi RS, Aloraini T, Almsned F, Alassali Y, Alfares A, et al. Prospect of genetic disorders in Saudi Arabia. *Front Genet*. 2023;14:1243518.
33. Alyafee Y, Al Tuwajiri A, Umair M, Alharbi M, Haddad S, Ballow M, et al. Non-invasive prenatal testing for autosomal recessive disorders: A new promising approach. *Front Genet*. 2022;13:1047474.
34. Jelin AC, Vora N. Whole Exome Sequencing: Applications in Prenatal Genetics. *Obstet Gynecol Clin North Am*. 2018;45(1):69-81.
35. Warsy AS, Al-Jaser MH, Albdass A, Al-Daihan S, Alanazi M. Is consanguinity prevalence decreasing in Saudis? A study in two generations. *Afr Health Sci*. 2014;14(2):314-21.
36. Mahboub SM, Alsaqabi AA, Allwimi NA, Aleissa DN, Al-Mubarak BA. Prevalence and pattern of consanguineous marriage among educated married

- individuals in Riyadh. *J Biosoc Sci.* 2020;52(5):768-75.
37. Albeshar N, Massadeh S, Hassan SM, Alaamery M. Consanguinity and Congenital Heart Disease Susceptibility: Insights into Rare Genetic Variations in Saudi Arabia. *Genes (Basel).* 2022;13(2):354.
38. Alyafee Y, Alam Q, Tuwajri AA, Umair M, Haddad S, Alharbi M, et al. Next-Generation Sequencing-Based Pre-Implantation Genetic Testing for Aneuploidy (PGT-A): First Report from Saudi Arabia. *Genes (Basel).* 2021;12(4):461.
39. Qin D. Next-generation sequencing and its clinical application. *Cancer Biol Med.* 2019;16(1):4-10.

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