ORIGINAL REPORT

Investigating knowledge, attitude, and practice regarding the health risks of consanguineous marriages in Saudi Arabia: a cross-sectional study

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ABSTRACT

Objective: This study aimed to study knowledge, attitude, and practice regarding the health risks of consanguineous marriage (CM) among the population in the Western region of Saudi Arabia.

Methods: A cross-sectional study was conducted among a total of 1,789 adults aged 18 years and above, residing in the Western region of Saudi Arabia, from June to July 2025, using a snowball sampling technique. An online questionnaire was designed based on a literature review.

Results: The participants were predominantly females (70.3%), aged 21-30 years (42.3%), and university educated (58.0%), with 20.4% working in medical professions. Of the 762 who responded to the consanguinity question, 33.2% reported being in a CM, mostly with the first cousin (25.2%). Overall mean knowledge score was 73.4% ± 26.4%, with 80.8% scoring above 50%; mean attitude score was 70.1% ± 9.1, with 98.4% scoring above 50%. Knowledge gaps were identified in awareness of premarital screening components (26.4% correct) and availability of premarital whole exome sequencing (51.6% correct). Female gender, younger age (18-20 years), university or high school education, and healthcare employment were independently associated with higher knowledge scores ($p < 0.05$). Attitudes were uniformly positive toward genetic screening but varied toward CM. In practice, 60% reported avoiding CM if screening results were incompatible.

Conclusion: While general knowledge and attitudes toward genetic screening were favorable, substantial educational gaps persisted, particularly regarding available testing and residual genetic risk. This highlighted the need for targeted public health education and campaigns for older, less-educated, and non-healthcare populations.

Keywords: Consanguineous marriage, KAP, premarital screening, health risks, Saudi Arabia.

Introduction

Consanguineous marriages (CM) are unions between a man and a woman who share a common ancestor or blood relation. This term is typically used to describe marriages between individuals related as second cousins or closer (1). CM is considered a respected practice in many communities worldwide, particularly in Saudi Arabia and many Arab and Middle Eastern countries, with varying rates depending on religion, culture, and geography (2).

Saudi Arabia has the highest incidence of CM, which is reported to be approaching about 60%. Such practice has its own risk, specifically when it comes to inherited disorders (3). The possibility of parents sharing the same recessive genes in CM is considerably greater

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than in non-consanguineous unions. Consequently, populations with a high prevalence of consanguinity experience an increased occurrence of autosomal recessive disorders (4). The chance of passing on these illnesses between parents with the same mutated gene is a 25% chance of the child developing the disease and a 50% chance of them being carriers of the mutated gene (5).

In Saudi Arabia, one of the major autosomal recessive disorders with high prevalence is the recessively inherited blood disorder, namely sickle cell anemia and Thalassemia (6). To decrease their incidence, Saudi Arabia launched a mandatory premarital screening program in 2004, which includes blood tests to screen for Sickle cell anemia and Thalassemia as well as infectious diseases (HIV, Hepatitis B, and Hepatitis C). Following a marriage proposal, each couple must undergo these tests before being able to proceed with the marriage contract. In terms of inherited blood disorders, if both couples are identified as carriers, they would be labeled as incompatible and offered genetic counseling, but the decision to proceed with this marriage is left up to them (7).

Around the world, there have been different strategies for autosomal recessive disease prevention. For example, the Jewish community tests for the most prevalent and severe diseases in their communities, such as cystic fibrosis, spinal muscular atrophy, and some of the neurodegenerative disorders, before marriage or even before conception (8,9). A broader approach became more popular in the year 2021, where premarital carrier screening uses whole exome sequencing (WES) for couples entering a consanguineous union to identify disease-causing variants in more than 500 genes that are responsible for more than 800 prevalent autosomal recessive disorders (10,11). If both couples are found to be carriers for disease-causing variants in the same gene, they would be identified as having a high risk for their future children to develop this genetic disorder. In the United Arab Emirates, premarital WES became an essential part of the premarital screen in the year 2025 (12,13). However, this remained optional for high-risk couples in Saudi Arabia.

In Saudi Arabia, these initiatives have effectively targeted the intended population and identified couples at high risk (14). There is still a significant number of couples who decided to enter CM or proceed with marriage despite being identified as high risk (15). This behavior could be explained by cultural beliefs about the importance of CM, as well as a lack of knowledge about its associated risks and a poor understanding of the premarital screen, its available options, and limitations.

While previous research had examined cousin marriages in some regions, there is a lack of studies specifically focusing on the Western region of Saudi Arabia. Furthermore, many existing studies do not address perception toward recent public health interventions, specifically premarital WES. This study aimed to address these gaps by investigating the knowledge, attitude, and practice (KAP) regarding cousin marriages, their association with genetic disorders, and the premarital screening program in the Western region of Saudi Arabia. The findings would provide updated insights that can inform culturally sensitive public health strategies and effectively address the health risks associated with cousin marriages.

Materials and Methods

Study design and setting

This study employed a descriptive cross-sectional design using an online questionnaire to investigate KAP regarding CM among adults in the Western region of Saudi Arabia, from June to July 2025.

Study population and eligibility criteria

The target population comprised adults aged 18 years and above residing in the Western region of Saudi Arabia. Inclusion criteria were: (1) adults aged 18 years and above, (2) individuals residing in the Western region of Saudi Arabia, (3) both genders, and (4) individuals who consented to participate and could adequately respond to the questionnaire. Exclusion criteria included: (1) participants who did not complete the survey, (2) those who refused to participate, and (3) visitors to the region who were not permanent residents.

Sample size determination and sampling technique

The sample size was calculated using the Raosoft sample size calculator. Based on the Western region population of 8,021,463 according to the Saudi census 2022 (16), with a 95% confidence interval, 50% anticipated frequency, and accounting for design effect, the minimum required sample size was determined to be 385 participants. Data collection was conducted electronically through social media platforms using a snowball sampling technique to reach the target population.

Data collection instrument

A structured questionnaire was developed based on extensive literature review and comprised four main sections: (1) demographic information including age, gender, education level, employment status, marital status, and residence details; (2) knowledge assessment containing eight items evaluating understanding of genetic diseases, their prevalence, preventability, and the relationship between CMs and genetic disorders; (3) attitude evaluation using 7 Likert-scale items (ranging from "strongly disagree" to "strongly agree") measuring participants' attitudes toward CM and premarital genetic screen; and (4) practice assessment examining actual behaviors and decision-making processes related to CM and premarital genetic screen. Three experts in the field reviewed the questionnaire, and a pilot study of 30 participants (10% of the sample) was conducted.

Knowledge domain scoring

Knowledge was assessed through eight binary (Yes/No) questions and one multiple-choice question regarding premarital screening components. Each correct response to binary questions received one point, while incorrect responses received zero points. For the multiple-choice question on premarital screening components, scoring was based on the selection of evidence-based correct options: "Infectious Diseases," "Hereditary blood disorders," and "All genetic disorders." The knowledge domain score was calculated as the percentage of correct responses across all knowledge items, with a maximum possible score of 100%.

Attitude domain scoring

Attitudes were measured using 7 Likert-scale items, each scored from 1 to 5 points based on response favourability toward genetic health awareness and responsible marriage practices. Items were scored as follows: "Strongly Disagree" = 1, "Disagree" = 2, "Neutral" = 3, "Agree" = 4, "Strongly Agree" = 5. For items where disagreement indicated a positive attitude (e.g., "CM is acceptable even without family genetic history"), reverse scoring was applied (1→5, 2→4, 3→3, 4→2, 5→1). The attitude domain score was calculated as the sum of individual item scores, with a maximum possible score of 35 points, which was also converted to a percentage for comparative analysis.

Data collection procedures

The Arabic-language questionnaire was distributed electronically via Google Forms through various social media platforms such as WhatsApp, Telegram, and Twitter. The survey link was accompanied by clear explanations of the study objectives, target population criteria, and voluntary participation information.

Statistical analysis

Data analysis was conducted using R v 4.3. Descriptive statistics were calculated for all variables, with continuous data presented as means ± standard deviations or medians with interquartile ranges depending on distribution normality. Categorical variables were presented as frequencies and percentages. For univariate analysis, group comparisons were performed using Student's t-test or Mann-Whitney U test for continuous variables, and chi-square tests for categorical variables. Post-hoc analyses using Tukey's Tukey's Honestly Significance difference were conducted for multiple group comparisons, with results presented using superscript letters to indicate significant differences between groups.

Multivariate linear regression models were constructed to identify predictors of knowledge, attitude, and total scores. Independent variables included gender, age group, education level, employment status, and marital status, with appropriate reference categories established for meaningful interpretation. Model fit was assessed using R-squared values, and regression coefficients were presented with 95% confidence intervals. Statistical significance was set at $p < 0.05$ for all analyses. Score categorization was performed using predetermined cut-off points: low (<50%) and high (≥75%) for knowledge and attitude scores.

Ethical considerations and data management

Ethical approval was obtained from the Umm Al-Qura University Institutional Research Board prior to data collection (Approval number (HAPO-02-K-012-2025-04-2630) on 10/05/2024. Participant confidentiality was maintained through a combined system of codes, numbers, and pseudonyms, with no identifiable personal information collected. Access to data was restricted to the research team members only. All survey responses were collected anonymously, and participation was entirely voluntary with informed consent obtained electronically before questionnaire completion.

Results

Among the 1,789 participants, the sample was predominantly females (70.3%) and young adults aged 21-30 years (42.3%). Most held a university degree (58.0%) and were single (57.4%). Employment was largely outside the healthcare field (60.2%). Of the 762 who responded to the consanguinity question, 33.2% reported being in a CM. Among those, 25.2% were directly related as children of an aunt or uncle, while 66.3% reported no biological relation despite possibly sharing a family name (Table 1).

The highest correct response rate was for the item linking CM to genetic disorders (85.5%), followed closely by recognition of the seriousness of genetic diseases (83.5%) and awareness that a child might inherit a genetic disease from a parent (82.2%). In contrast, only 51.6% correctly identified the availability of premarital carrier screening (premarital WES), indicating a significant knowledge gap. Other areas with moderate correct response rates included the preventability of genetic diseases (73.3%), understanding child risk when both parents are carriers (70.0%), and awareness of genetic disease prevalence (67.6%). The sibling's risk of inheriting a genetic disease was correctly identified by 63.2% of participants. Notably, the lowest correct rate was for identifying screening components included in the premarital test, with only 26.4% responding correctly (Figure 1).

Participants showed strong support for genetic screening initiatives. Agreement was highest for comprehensive testing being beneficial (4.28 ± 0.97) and for mandating such testing (4.16 ± 0.99), with the majority expressing either agreement or strong agreement. Conversely, concerns about screening violating privacy were low, as most participants strongly disagreed with this notion (1.74 ± 1.12). Attitudes toward CM were more variable. While participants agreed that CM increases genetic risk even in the absence of family history (3.56 ± 1.11), many also expressed openness to such unions. Specifically, mean scores were moderate for willingness to consider CM (3.43 ± 1.19) or to marry a relative with a known genetic history (3.39 ± 1.23). The statement asking if the current premarital screen showed a compatible result

Table 1. Sociodemographic characteristics of participants (n = 1,789).

Variable	Frequency (Percentage) n (%)
Gender	
Female	1,257 (70.3)
Male	532 (29.7)
Age group	
18-20 years	333 (18.6)
21-30 years	757 (42.3)
31-40 years	176 (9.84)
41-50 years	264 (14.8)
>51 years	259 (14.5)
Education level	
Less than high school	43 (2.40)
High school	435 (24.3)
Diploma	113 (6.32)
University	1,038 (58.0)
MSc	98 (5.48)
PhD	62 (3.47)
Marital status	
Single	1,027 (57.4)
Married	671 (37.5)
Divorced	65 (3.63)
Widowed	26 (1.45)
Employment	
Medical (Doctor, Resident, Med Student, Genetic Counselor)	365 (20.4)
Other HCP	347 (19.4)
Other	1,077 (60.2)
CM (n = 762)	
Yes	253 (33.2)
No	509 (66.8)
Type of consanguineous relationship (n = 762)	
No relation, different family name	294 (38.6)
No relation, same family name	211 (27.7)
Son/Daughter of aunt/uncle	192 (25.2)
Other	65 (8.53)

Table 2. Mean (SD) scores of knowledge, attitude, and total knowledge by demographic variables.

Domain	Mean	SD	Median	Min	Max	Above 50%	Below 50%
Knowledge % (9 items)	73.40	26.40	77.80	0.00	111.10	1,446 (80.8%)	343 (19.2%)
Attitude % (7 items)	70.10	9.10	71.40	37.10	100.00	1,761 (98.4%)	28 (1.6%)

means there would be no risk for genetic statement received mixed (3.12 ± 1.20), and about 43% of the participants agreed or strongly agreed with this statement Figures 2-5.

In response to receiving incompatible results following the current premarital screen, the majority of participants indicated they would not proceed with marriage (n = 1074, 60.0%). Around one-quarter would seek genetic counseling (n = 446, 24.9%), while smaller proportions were unsure (n = 203, 11.3%) or stated they would proceed regardless (n = 66, 3.7%). When asked under what conditions they would consider doing the premarital WES, the most frequently endorsed reason was commitment to marriage regardless of circumstances (n = 758, 42.4%). Others indicated needing more information
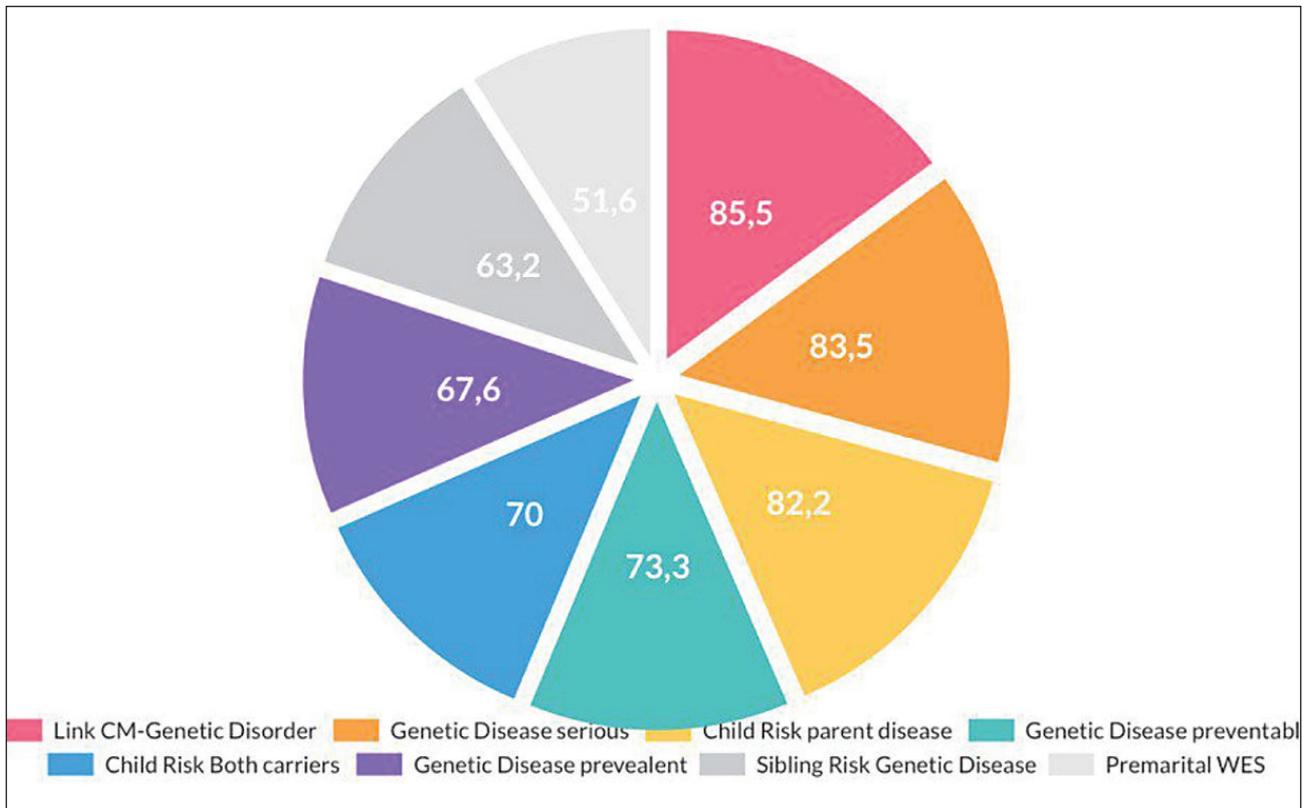
Figure 1. Percentage of correct response to knowledge assessment questions.

	Mean (SD)	Strongly disagree	Disagree	Neutral	Agree	Strongly agree
Would Marry Relative with Genetic History	3.39 (1.23)	11.9%	10.0%	23.8%	36.1%	18.3%
Mandatory Screening Violates Privacy	1.74 (1.12)	59.9%	20.2%	9.5%	6.4%	4.0%
Considering Consanguineous Marriage	3.43 (1.19)	10.0%	10.3%	24.8%	36.4%	18.4%
Consanguineous Marriage Increases Risk Without Family History	3.56 (1.11)	6.8%	9.3%	25.1%	39.2%	19.6%
Premarital WES Should be Mandatory	4.16 (0.99)	2.2%	4.6%	15.2%	30.6%	47.4%
Premarital WES Beneficial	4.28 (0.97)	3.0%	3.1%	9.2%	32.3%	52.3%
Compatible Results Allow Marriage Without Risk	3.12 (1.20)	13.1%	16.1%	27.8%	32.1%	10.9%

Figure 2. Attitude assessment toward CM and genetic testing.

($n = 417$, 23.3%) or the presence of a family history of genetic disorders ($n = 362$, 20.2%) as potential motivators. A few participants reported that they would do the test if entering a CM ($n = 151$, 8.4%). About 23.3% ($n = 417$) indicated that they would need more information to decide whether to do the test, and about 5.6% ($n = 101$) indicated that they would not consider doing it.

Participants demonstrated a mean knowledge percentage score of 73.4% ± 26.4% (median = 77.8%). A total of 1,446 individuals (80.8%) scored above 50%. Attitude scores (based on 7 Likert-scale items) were high overall,
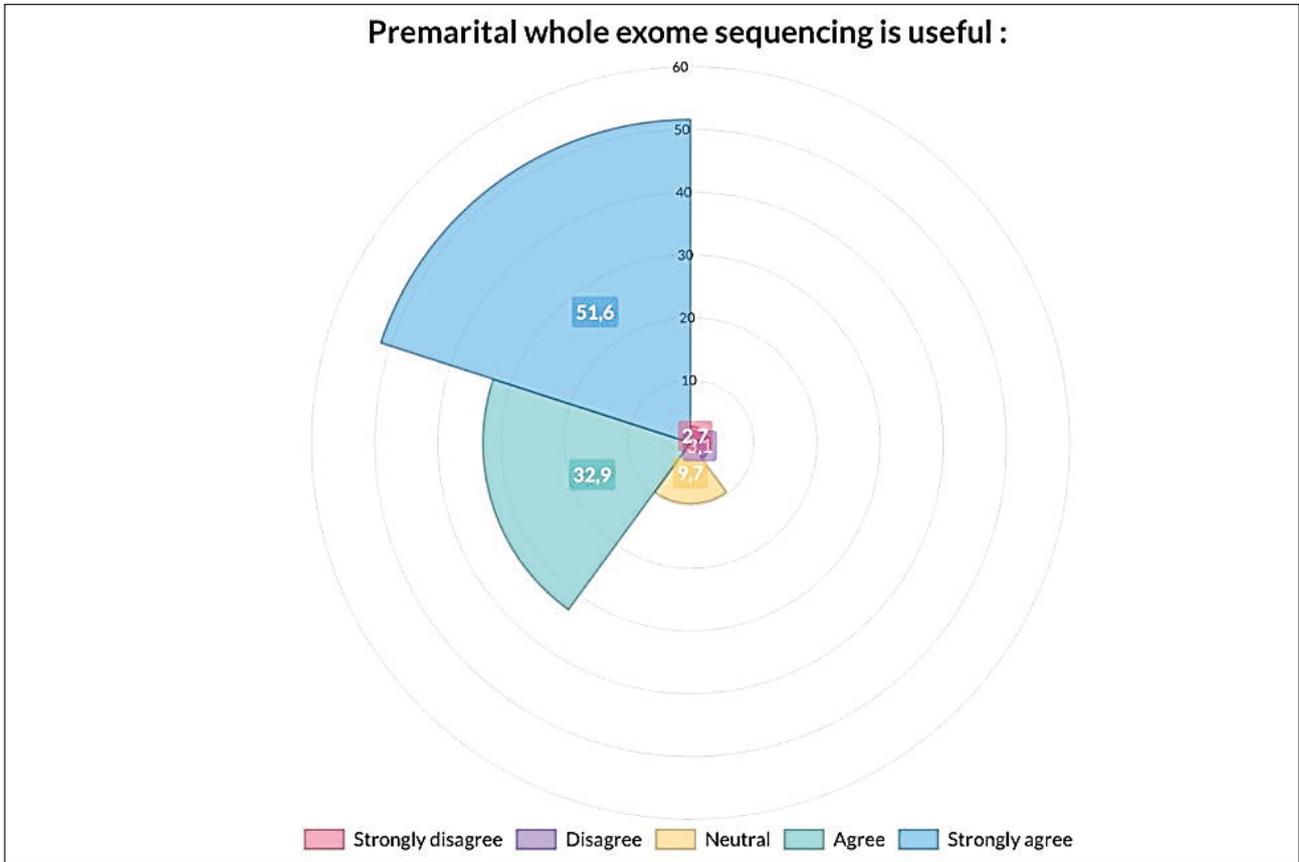
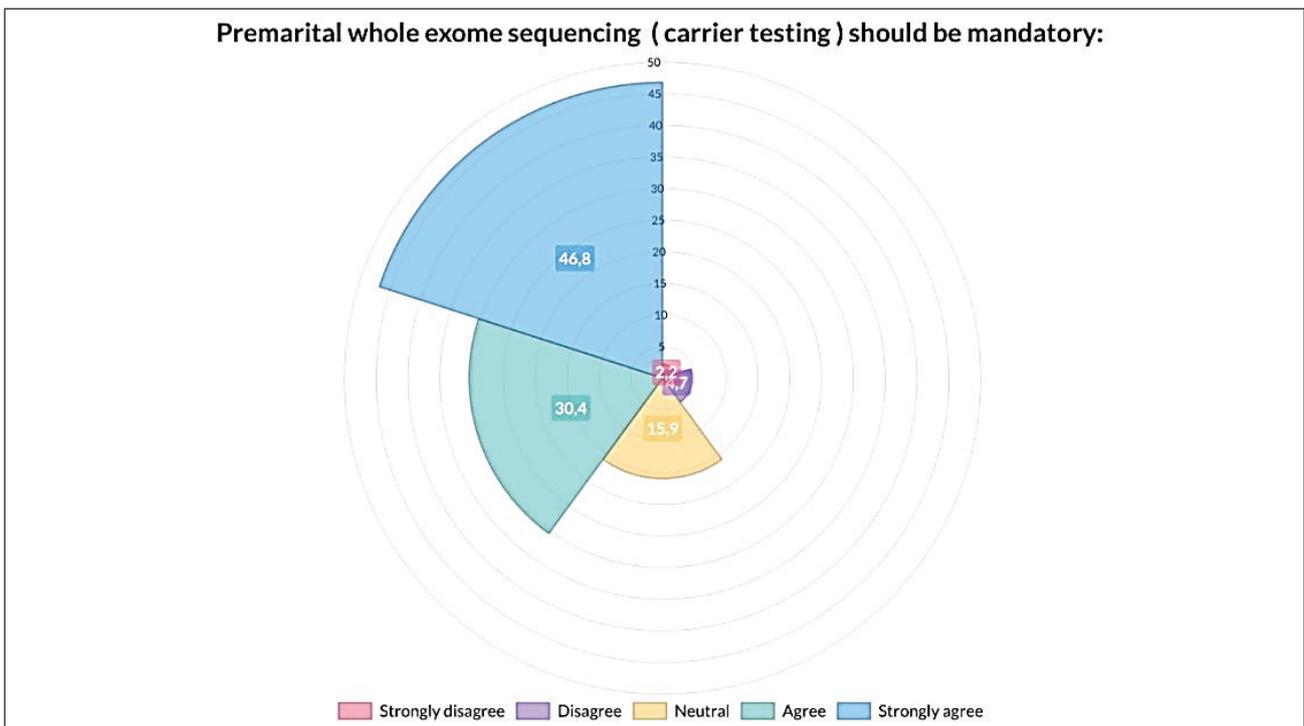**Figure 3.** *Participants' perceptions of the usefulness of premarital WES.*

Figure 4. *Participants' attitudes toward mandating premarital WES.*

with a mean of 70.1 ± 9.1 (median = 71.4), and 98.4% (n = 1,761) of participants scored above the 50% threshold (Table 2).

Female participants had higher knowledge scores than males (74.9 ± 26.1 *vs.* 70.1 ± 26.9; $p < 0.001$), as well as higher attitude scores (24.9 ± 3.1 *vs.* 23.7 ± 3.2; $p <$
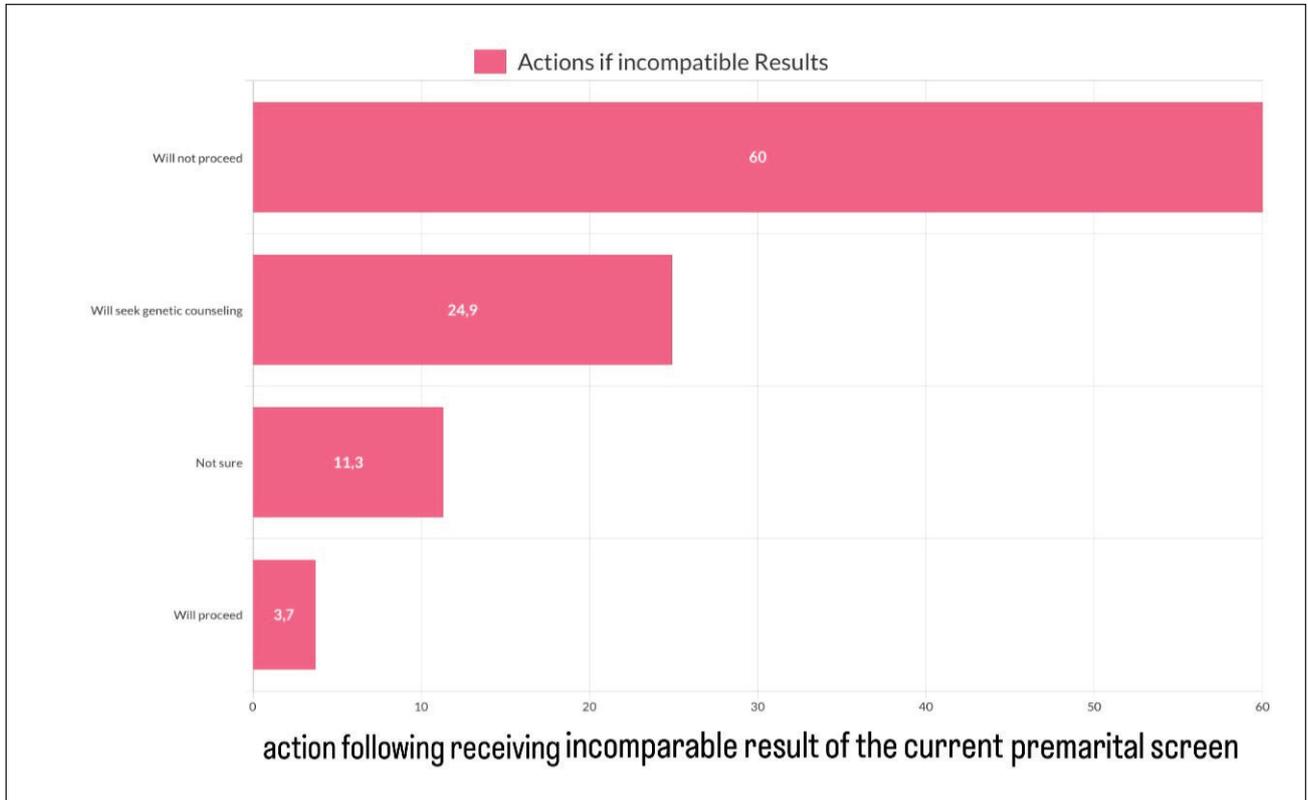
Figure 5. *Participants actions toward incompatible results and testing considerations.*

Table 3. Differences in knowledge and attitude scores across demographic groups.

Variable	Group	Knowledge	Attitude
Gender	Female	74.9 (26.1)[b]	24.9 (3.1)[b]
	Male	70.1 (26.9)[a]	23.7 (3.2)[a]
	p-value	*p* < 0.001	*p* < 0.001
Age group	18-20	80.8 (22.6)[a]	25.0 (3.0)[a]
	21-30	76.2 (27.3)[a]	24.4 (3.4)[a]
	31-40	72.3 (24.8)[a]	24.5 (3.2)[a]
	41-50	71.3 (21.9)[a]	24.8 (3.1)[a]
	>51	58.9 (28.0)[a]	24.1 (2.9)[a]
	p-value	*p* < 0.001	*p*-value = 0.001
Education level	Less than high school	55.0 (29.9)[b]	22.7 (3.2)[b]
	High school	75.5 (25.6)[a]	24.5 (2.9)[a]
	Diploma	59.3 (24.9)[b]	24.4 (3.0)[a]
	University	76.1 (25.1)[a]	24.7 (3.2)[a]
	MSc	63.9 (28.0)[b]	23.8 (3.8)[ab]
	PhD	68.3 (34.4)[ab]	24.2 (3.9)[ab]
	p-value	*p* < 0.001	*p* < 0.001
Employment	Other	68.3 (25.7)[b]	24.4 (3.0)[a]
	HCP	74.3 (28.0)[a]	24.8 (3.4)[a]
	Doctor/Resident/Medical Student/Genetic Counselor	87.9 (21.1)[c]	24.6 (3.5)[a]
	p-value	*p* < 0.001	*p*-value = 0.209
Marital status	Single	77.9 (25.7)[b]	24.6 (3.2)[a]
	Married	67.1 (26.3)[a]	24.5 (3.1)[a]
	Divorced	72.1 (24.3)[ab]	24.1 (3.4)[a]
	Widowed	64.1 (25.8)[a]	24.2 (3.2)[a]
	p-value	*p* < 0.001	*p*-value = 0.41

Table 4. *Multiple linear regression results for knowledge and attitude scores.*

Variable (Reference Category)	Knowledge score	Attitude score
Gender (Male)		
Female	3.61 (0.99-6.23)**	1.13 (0.81-1.46)***
Age group (21-30 years)		
18-20 years	5.43 (2.00-8.87)**	0.59 (0.16-1.01)**
31-40 years	2.42 (-2.65-7.48)	0.53 (-0.10-1.16)
41+ years	-2.54 (-7.08-2.01)	0.72 (0.16-1.29)*
Education (High school)		
Diploma	-10.89 (-16.14-5.64)***	0.26 (-0.40-0.91)
University	2.80 (-0.05-5.65)	0.41 (0.06-0.77)*
Post-graduate	-5.50 (-10.21-0.79)*	-0.18 (-0.76-0.41)
Employment (Non-healthcare)		
Healthcare	10.78 (8.04-13.52)***	0.41 (0.07-0.75)*
Marital status (Single)		
Divorced	3.25 (-3.98-10.49)	-0.79 (-1.69-0.11)
Married	-2.28 (-6.48-1.92)	-0.32 (-0.84-0.20)
Widowed	-0.56 (-11.28-10.16)	-0.82 (-2.16-0.51)
Model statistics		
R^2 / Adjusted R^2	0.105 / 0.100	0.042 / 0.036

*$p < 0.05$; **$p < 0.01$; ***$p < 0.001$.

Estimates are unstandardized coefficients from multiple linear regression. Confidence intervals are in parentheses.

0.001). The highest knowledge was observed among participants aged 18-20 years (80.8 ± 22.6), while those older than 51 years had the lowest (58.9 ± 28.0; $p < 0.001$). Regarding education, university graduates (76.1 ± 25.1) and high school graduates (75.5 ± 25.6) scored significantly higher than those with less than high school education (55.0 ± 29.9; $p < 0.001$). In terms of employment, doctors, residents, and medical students had the highest knowledge scores (87.9 ± 21.1), significantly surpassing other groups (e.g., 68.3 ± 25.7 among non-health workers; $p < 0.001$). Single participants also had higher knowledge scores (77.9 ± 25.7) compared to married (67.1 ± 26.3) and widowed individuals (64.1 ± 25.8; $p < 0.001$). In contrast, attitude scores showed minimal variability and did not differ significantly by employment (p-value = 0.209) or marital status (p-value = 0.41) (Table 3).

Means are presented as mean (SD). Differences between group levels were assessed using one-way ANOVA followed by Tukey post-hoc comparisons. Superscript letters indicate statistically significant differences between levels within each variable ($p < 0.05$). Pairwise missing values were excluded.

Female participants had significantly higher knowledge scores (+3.61; 95% CI: 0.99-6.23; $p < 0.01$) and attitude scores (+1.13; 95% CI: 0.81-1.46; $p < 0.001$) compared to males. Those aged 18-20 years reported higher knowledge (+5.43; 95% CI: 2.00-8.87; $p < 0.01$) and attitude (+0.59; 95% CI: 0.16-1.01; $p < 0.01$) than the 21-30 years age group, while participants aged 41+ years had higher attitude only (+0.72; 95% CI: 0.16-1.29; p

< 0.05). Compared to high school graduates, diploma holders and post-graduates had lower knowledge scores (-10.89 and -5.50; $p < 0.001$ and $p < 0.05$, respectively), while university education was not significantly different. Healthcare workers scored higher in both knowledge (+10.78; $p < 0.001$) and attitude (+0.41; $p < 0.05$) (Table 4).

Discussion

Consanguinity or marriage between relatives is a deeply rooted practice in the culture and familial customs of the Saudi Arabian population. This might also open advantages from the economic or social point of view; however, it carries an important risk of transferring autosomal recessive genetic disorders (2,4,17). In the current study cohort, the prevalence of CM among study participants was about 53% (between cousin marriage or marriage from the same tribe). In terms of knowledge, the current study cohort showed a moderately good performance in knowing the risk of genetic disease associated with CM, the risk of transmission from carrier parents and siblings, and how they can be prevented. These results seem to be overall similar in the general awareness of genetic risks to those found in Riyadh and the Eastern Province (14,15).

Nevertheless, in every region, there seems to be a discrepancy between awareness of the risk being there and behavior toward it. As most of the current study cohort were open to consanguineous union, and even 78% of them were neutral or showed agreement to marry a relative with a known family history of genetic disorder.

This means that cultural beliefs and family norms could overcome knowledge. Recent systematic reviews also indicated that, although awareness was relatively high in Saudi Arabia, cultural and family traditions often continue to override health-based decision-making (17,18).

Another systematic review in the Middle East found that although many countries had mandated premarital screening, this did not decrease the incidence of CM effectively in any of them (19). Another study from Jazan targeted couples who proceeded with their marriage decision despite incompatibility in premarital screen found out that more than half of them rejected the counselling advice as their marriage was unavoidable, 30% thought that no clear explanation provided to them, 18% thought the risk of transmitting the disease to their children was low and 5% thought that their children life would not be affected by the disease (20).

This poor adherence to premarital screen recommendations could be attributed to their poor knowledge about premarital screens. In the current study cohort, a fair number of participants believed that genetic disease could be preventable, but when it comes to preventive strategies, most importantly, premarital screening, they showed poor knowledge. This aligned with Al Eissa et al. (21), who reported about the population in the central region of Saudi Arabia, where 30% believed that the current premarital screening covers all genetic disorders. Another national study reported that only 6% of people in Saudi Arabia understood the rationale behind premarital screening, and 50% knew that sickle cell anemia and thalassemia are genetic disorders, while they are the two most common genetic diseases in Saudi Arabia (18).

Both poor understanding of the premarital screening measures and strong value of consanguinity could be a possible reason why the prevalence of sickle cell anemia and Thalassemia in Saudi Arabia remained high (22). However, such programs are effective in some communities, such as the targeted premarital screen in the Jewish community, which resulted in a decrease in the incidence of spinal muscle atrophy by 57%, cystic fibrosis, and severe neurodegenerative disorder to a very low prevalence (8).

An interesting point to mention about the Jewish community is that they have a high incidence of consanguinity, similar to the Saudi community. Still, as it is a strong cultural practice, their preventive program is mainly periconceptional (for couples who are willing to have children). Carrier screen done for both couples, and if high risk is identified, further counseling is conducted for further preventive strategies, such as in vitro fertilization and preimplantation genetic diagnosis, or early pregnancy testing and termination if the fetus is affected. This strategy reserved some autonomy for partner decision-making while continuing to offer risk detection and prevention of genetic disorders. They offer these tests as a premarital test if the couple chooses to do this (8,21).

The current study community might not be willing to avoid CM due to cultural pressure, but they were willing to seek help in terms of possible prevention. This was observed in their attitude and action toward premarital testing. Most of the participants in the current study did not believe that mandatory premarital screening violates their privacy; rather, about 80% thought that premarital WES should be Mandatory. Further, 40% thought that they would do premarital WES if they got married. Similarly, a study in the Northern region determined that when people were asked, "would you add a test to make the premarital screen broader", > 90% answered yes (23). This meant that the current study community was not against the testing and prevention, but the marriage choice was possibly a limiting factor.

It would be hard for such programs to be effective if people do not understand them well, or do not know they exist, or the meaning of the result. Public education concentrating on the area of genetic disease prevention, specifically premarital screening, components, and limitations, would be of huge significance. If premarital screening were expanded to include prevalent and severe disorders in Saudi Arabia or even WES, it would be accepted by the public based on their response in these surveys. In addition to premarital screening effectiveness in decreasing the disease prevalence, it is also cost-effective. Rabea et al. (24) compared the cost of premarital screening for spinal muscle atrophy to the cost of interventions required for diagnosed patients and found it to be less by 14 to 28 fold. Such studies do not exist for premarital WES, as it is a relatively new practice.

Several recommendations could be addressed, such as government-based intervention to consider expanding the current premarital screen to involve prevalent and severe disorders in Saudi Arabia, or even WES with frequent evaluation to assess its effectiveness. Also, public education about the available preventive measures, specifically the premarital screening program and the availability of the optional premarital WES. Social media, a heavily used platform, should be used in this education. Last but not least, counseling following premarital testing compatibility should not only include proceeding or not proceeding with the marriage, but also be widened to discuss further intervention, such as *in vitro* fertilization and preimplantation genetic diagnosis, or early pregnancy detection and termination, if it remains within the scope of religion, if the couple is planning to proceed with a high-risk marriage. Longitudinal studies following the introduction of these educational programs to evaluate their effectiveness are also required.

Despite the huge and respected efforts, limitations could exist. The cross-sectional design cannot establish causality. The mostly young sample limited generalizability, and online snowball sampling might introduce selection bias. Self-reported data could not reflect real behavior.

Conclusion

While general knowledge and attitudes toward genetic screening were favorable, substantial educational gaps persisted, particularly regarding available testing and residual genetic risk. Although people thought that consanguinity was a cultural norm, they were willing to proceed with preventive actions. Hence, improving public

education about this area would be greatly beneficial. Further, female gender, younger age (18-20 years), university or high school education, and healthcare employment were independently associated with higher knowledge scores ($p < 0.05$). This highlighted the need for targeted public health education and campaigns for older, less educated, and non-healthcare populations.

List of Abbreviations

CM   Consanguineous Marriage
HIV   Human Immunodeficiency virus
WES   Whole Exome Sequence
KAP   Knowledge, Attitude and practice
HCP   Health Care Provider

Disclosure

The authors have no conflict of interests, and the work was not supported or funded by any drug company. The authors also declare that all authors contributed equally in interpreting the results, editing, and approving the manuscript before submission.

Conflict of interests

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Ethical approval

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References

1. Islam MM. The practice of consanguineous marriage in Oman: prevalence, trends and determinants. J Biosoc Sci. 2012;44(5):571–94. https://doi.org/10.1017/S0021932012000016

2. 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. https://doi.org/10.7759/cureus.53888

3. Al-Owain M, Al-Zaidan H, Al-Hassnan Z. Map of autosomal recessive genetic disorders in Saudi Arabia: concepts and future directions. Am J Med Genet Part A. 2012;158A(10):2629–40. https://doi.org/10.1002/ajmg.a.35551

4. Temaj G, Nuhii N, Sayer JA. The impact of consanguinity on human health and disease with an emphasis on rare diseases. J Rare Dis. 2022;1(1):1–7. https://doi.org/10.1007/s44162-022-00004-5

5. Taglia A, Picillo E, D'Ambrosio P, Cecio MR, Viggiano E, Politano L. Genetic counseling in Pompe disease. Acta Myol. 2011;30(3):179

6. AlHamdan NAR, AlMazrou YY, AlSwaidi FM, Choudhry AJ. Premarital screening for thalassemia and sickle cell disease in Saudi Arabia. Genet Med. 2007;9(6):372–7. https://doi.org/10.1097/GIM.0b013e318065a9e8

7. Ministry of Health, Kingdom of Saudi Arabia. Premarital screening Riyadh, Saudi Arabia: Ministry of Health [cited 2025 Oct 18]. Available from: https://www.moh.gov.sa/en/HealthAwareness/Beforemarriage/Pages/default.aspx

8. Nov-Klaiman T, Horn R, Raz A. More of the same? Israel's expanded carrier screening for cystic fibrosis. Eur J Hum Genet. 2025;33(12):1555–7. https://doi.org/10.1038/s41431-025-01851-8

9. Singer A, Sagi-Dain L. Impact of a national genetic carrier-screening program for reproductive purposes. Acta Obstet Gynecol Scand. 2020;99(6):802–8. https://doi.org/10.1111/aogs.13858

10. Dos Santos CMA, Heller AH, Pena HB, Pena SDJ. A protocol for preconceptional screening of consanguineous couples using whole exome sequencing. Front Genet. 2021;12:685123. https://doi.org/10.3389/fgene.2021.685123

11. Huang Q, Wang Z, Teng Y, Zhang W, Wen J, Zhu H, et al. Application of whole exome sequencing in carrier screening for high-risk families without probands. Front Genet. 2024;15:1415811. https://doi.org/10.3389/fgene.2024.1415811

12. Abu Dhabi Public Health Centre. Premarital screening Abu Dhabi, UAE: Abu Dhabi Public Health Centre [cited 2025 Oct 18]. Available from: https://www.adphc.gov.ae/en/Public-Health-Programs/Premarital-Screening

13. Ministry of Health and Prevention - UAE. Genetic testing as part of premarital screening for Emiratis [cited 2025 Oct 18]. Available from: https://mohap.gov.ae/en/w/genetic-testing-as-part-of-premarital-screening-for-emiratis

14. Bakry H, Alaiban RA, Alkhyyat AA, Alshamrani BH, Naitah RN, Almoayad F. Predictors of consanguinity marriage decision in Saudi Arabia: a pilot study. Healthcare. 2023;11(13):1925. https://doi.org/10.3390/healthcare11131925

15. Gosadi IM. National screening programs in Saudi Arabia: overview, outcomes, and effectiveness. J Infect Public Health. 2019;12(5):608–14. https://doi.org/10.1016/j.jiph.2019.06.001

16. General Authority for Statistics, Kingdom of Saudi Arabia Riyadh, Saudi Arabia: General Authority of statistics [cited 2025 Oct 7]. Available from: https://www.stats.gov.sa/web/guest/statistics?index=119025&subindex=127380

17. Albanghali MA. Prevalence of consanguineous marriage among Saudi Citizens of Albaha, a cross-sectional study. Int J Environ Res Public Health. 2023;20(4):3767. https://doi.org/10.3390/ijerph20043767

18. 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. https://doi.org/10.1186/s40246-024-00700-x

19. Saffi M, Howard N. Exploring the effectiveness of mandatory premarital screening and genetic counselling programmes for β-thalassaemia in the Middle East: a scoping review. Public Health Genomics. 2015;18(4):193–203. https://doi.org/10.1159/000430837

20. Gosadi IM, Gohal GA, Dalak AE, Alnami AA, Aljabri NA, Zurayyir AJ. Assessment of factors associated with the effectiveness of premarital screening for hemoglobinopathies in the South of Saudi Arabia. Int J Gen Med. 2021;14:3079–86. https://doi.org/10.2147/IJGM.S321046

21. 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. https://doi.org/10.1186/s12889-024-19029-0

22. Makkawi M, Alasmari S, Hawan AA, Al Shahrani MM, Dera AA. Hemoglobinopathies. Saudi Med J. 2021;42(7):784–9. https://doi.org/10.15537/smj.2021.42.7.20210273

23. Hafiz MN, Suhail N, Mohammed ZMS, Elzein HO, Almasmoum HA, Abass AE, et al. Awareness and attitude of the general population towards inherited hemoglobinopathies in the premarital screening program in the Northern Region of Saudi Arabia. Hematol Rep. 2025;17(1):9. https://doi.org/10.3390/hematolrep17010009

24. Rabea F, El Naofal M, Chekroun I, Khalaf M, Zaabi N Al, AlZaabi K, et al. Spinal muscular atrophy genetic epidemiology and the case for premarital genomic screening in Arab populations. Commun Med. 2024;4(1):1–6. https://doi.org/10.1038/s43856-024-00548-1