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KNOWLEDGE AND AWARENESS LEVELS OF GENETIC DISEASES RESULTING FROM CONSANGUINEOUS MARRIAGES AMONG SAUDI POPULATION

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Abstract

Background: Consanguineous marriages, prevalent in Saudi Arabia, significantly increase the risk of genetic disorders as unions often involve close relatives. Public awareness of the genetic implications of such marriages is critical for reducing the incidence of hereditary diseases. This study aimed to evaluate the knowledge and awareness levels of genetic diseases associated with consanguineous marriages among the Saudi population.

Methods: A randomized cross-sectional study was conducted from July to December 2024, involving 402 Saudi adults aged 18 and above. Participants were recruited via a structured questionnaire assessing their awareness of genetic diseases related to consanguinity. Data analysis was performed using the Statistical Package for Social Sciences (SPSS) version 25, focusing on demographic factors, levels of knowledge, and attitudes towards consanguineous marriages and genetic screening.

Results: The study revealed that 58% of respondents opposed consanguineous marriages, while 75.9% acknowledged health risks associated with these unions. A significant knowledge gap was noted, with only 39.1% demonstrating high awareness of genetic diseases, although 99.3% were aware of premarital screening's importance. Despite recognizing the life-threatening nature of genetic diseases (91%), only 25.4% believed effective treatments were available. Statistical analysis indicated significant correlations between knowledge levels and age ($p=0.024$) and educational attainment ($p=0.048$), with a concerning proportion of participants exhibiting low awareness levels (37.6%).

Conclusion: The findings highlight a crucial need for enhanced education and awareness initiatives regarding the genetic consequences of consanguinity in Saudi Arabia. While recognition of health risks associated with such marriages is relatively high, misconceptions about premarital screening and treatment options persist. These gaps in knowledge underscore the necessity for targeted public health campaigns to mitigate the hereditary impact of consanguineous marriages and improve community health outcomes. Improved education could promote more informed decisions and practices regarding

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marriage and genetic health within the Saudi population.

Keywords: Genetic Diseases, Consanguineous Marriages, Inherited Diseases, Awareness and Knowledge, Saudi Arabia.

Introduction:
Consanguineous marriage describes a union that involves two closely related adults. In clinical genetics, this concept is more particular to unions between second-degree relatives or closer in link [1]. Consanguinity increases the possibility of pairing between two individual heterozygotes for the same recessive mutant allele, leading hidden disorders in the family to manifest in subsequent generations [2]. People in Saudi society are keen to marry consanguineously, which has caused the transfer of numerous hereditary disorders [3]. Saudi Arabia has worked to lower the incidence of genetic illnesses in previous decades by implementing national, community, awareness, and preventive initiatives [4]**.** Among all marriages, consanguineous marriages account for 20–50% of records, particularly in areas where Islam is the dominant religion. Consanguinity is viewed negatively in the Arab world regarding family trees [5]. According to recent reports, Saudi Arabia is among the countries with the highest percentages of consanguinity marriages, with rates as high as 60% and 70% [6]. Approximately 5% of all diseases worldwide are caused by genetic abnormalities. Thirty percent (30%) of children and 10% of adult admissions in developed nations are caused by hereditary factors [7].
According to 2020 research, the Saudi community is increasing its general knowledge and awareness of the risk of consanguineous marriage and its influence on the spreading of genetic disorders among the younger population [8]. Another study found that respondents' understanding of consanguinity was below average among adults [9]. A low rate of attitudes towards consanguineous marriage was found and correlated with education level [10]. In addition, female health and non-health students were found to have a higher level of awareness [11]. The main motivation behind conducting this research was the lack of previous studies on the general Saudi population. Therefore, the main objective of this study is to assess the level of knowledge and awareness levels among residents of Saudi Arabia regarding genetic diseases resulting from consanguineous marriages.

Objective:
the study aimed to assess the community's knowledge and awareness of genetic diseases resulting from consanguinity.

Methodology:
Study Design and Setting:
A randomized cross-sectional study was conducted among the Saudi population from July to December 2024. The study’s population consisted of Saudi adults over 18 years old. Participants were recruited in August 2024 from people receiving the questionnaire.

Sample size:
The sample size was estimated using the Qualtrics calculator tool with a confidence level of 95%, and the minimum number of participants was 385.

Inclusion and Exclusion criteria:
Including the requirements of Saudi residents, males/females over 18 years old from all provinces of Saudi Arabia.

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Data collection tools:
A survey instrument derived from questions used in a prior study of a similar nature was used to collect data [12]. It was given out anonymously; the questionnaire was available in Arabic and English and had three sections. The first section asked about demographic characteristics, including age and gender. The second section questioned their understanding of consanguinity marriage with seven questions. Finally, the third section has Thirteen questions about the subject's opinions on consanguineous marriages and the likelihood of contracting a particular illness.

Scoring system:
Knowledge score: The responses provided in the knowledge section were considered to calculate the knowledge score for consanguineous marriage. The participants were asked seven common questions, and their overall knowledge was calculated based on the combined scores of the questions. 1 point was given for correct answers and 0 points for wrong answers. The original Bloom's cut-off points, 80.0% -100.0, 60%-79%, and <59%, were used from the original study. Knowledge scores ranging from 1 to 7 points were categorized into three levels: high level (7-5.6 points), moderate level (5.5-4.2 points), and low level (4.1-0 points).
Awareness score: Each participant received 13 questions regarding their views on consanguineous marriages and the risk of developing specific diseases. The responses observed were **agree**, **disagree**, or **I don't know**. 1 to 2 points were given based on the level of awareness present, and "I don't know" received 0 points, making the highest possible score 26. Bloom's cut-off points were used to determine awareness levels. Awareness scores could range from 1 to 26, with classifications as follows: high awareness (26-20.8), moderate awareness (20.5-15.6), and low awareness (15.3-0).

Pilot test:
A pilot study was conducted by distributing the questionnaire to 20 individuals to assess the questionnaire's simplicity and the research's feasibility. Data collected during this pilot phase was not included in the final data set of the main study.

Analyzes and entry method:
The computer's "Microsoft Office Excel Software" Windows (2021) was used to enter data. The Statistical Package of Social Science Software (SPSS) application, version 25 (IBM SPSS Statistics for Microsoft Windows, Version 25.0.), was then used to receive the gathered data and perform statistical analysis.

Results:

Table (1) displays various demographic parameters of the participants with a total number of (402). The age distribution shows a peak between 32 and 49 years at the age of 35.3 years with a standard deviation of 14.1 years; 27.1% of the participants were aged between 32 and 49 years. First, note the skew towards females in the sample of 69.7% of participants. Marital status reveals that a huge majority of the participants are either married or single, the former being 51.0 % and the later 44.0 %, which can be interpreted as a relatively young and stable set of people. This high educational attainment seems to correlate with the income distribution of nearly half of our participants (47.5%), who reported monthly income of 1,000 to 5,000 SAR. Additionally, the overwhelming majority of participants are Saudi nationals (98.5%), and the data strongly suggests that there is a strong regional concentration to the south (55.5%) which suggests further possibility for regional disparities in socio-economic research.

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Table (1): Sociodemographic characteristics of participants (n=402)

Parameter		***No.***	***Percent (%)***
Age (Mean: 35.3, STD: 14.1)	Less than 23	100	24.9
	23 to 31	100	24.9
	32 to 49	109	27.1
	50 or more	93	23.1
Gender	Female	280	69.7
	Male	122	30.3
Marital status	Single	177	44.0
	Married	205	51.0
	Divorced	13	3.2
	Widowed	7	1.7
Education level	Middle school	7	1.7
	High school	94	23.4
	Bachelor's degree	274	68.2
	Postgraduate degree	26	6.5
	Uneducated	1	.2
Nationality	Saudi	396	98.5
	Non-Saudi	6	1.5
Monthly income in SAR	1000 to 5000	191	47.5
	5001 to 10000	92	22.9
	10001 to 20000	85	21.1
	More than 20000	34	8.5
Region	Northern region	5	1.2
	Southern region	17	4.2
	Central region	223	55.5
	Eastern region	70	17.4
	Western region	87	21.6

As shown in figure 1, From a total sample size of 402 respondents, we have 125, or 31.1 % of respondents who believed that such unions lower the risk of genetic diseases. On the contrary, a vast majority — 210 respondents, or 52.2 percent — disagreed with this notion, expressing their overall scepticism around the genetic aspects of intermarriage within a larger network of family. Additionally, 67 (16.7%) participants were uncertain about the risk studied.

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Figure (1): Illustrates whether marrying a second or third-degree relative reduces the risk of genetic diseases among participants.

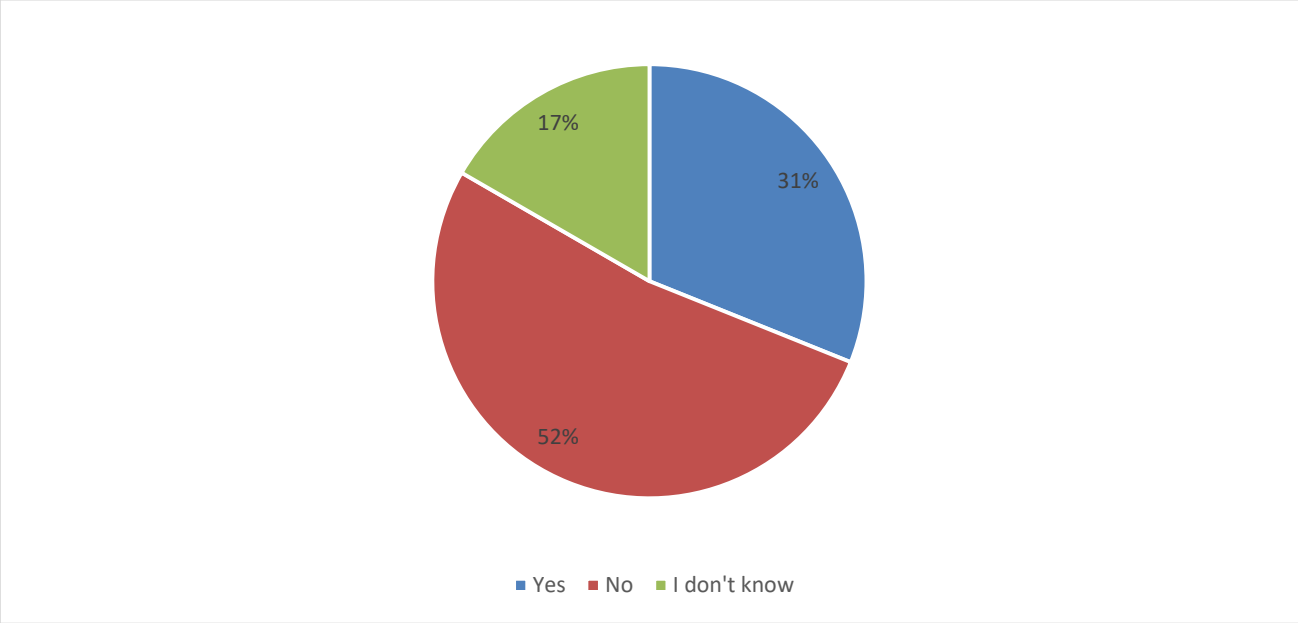

Table 2 shows perspectives amongst a sample population on consanguineous marriages and their genetic implications. A clear 58.0% of respondents against such marriages, a notable proportion (22.6%) uncertain and which may indicate a real confusion and ignorance of the matter. It is interesting that only 31.1% agree that marrying the second or third degree relative can mitigate the risk of genetic disease while more than half (52.2%) contradict this claim. Our big skepticism is also supported by the overwhelming agreement on the life-threatening nature of genetic diseases (91.0 %) but only 25.4 % admit that there is any treatments available, suggesting a chasm of lack of optimism in medical advancement. Premarital screening is highly aware (99.3%), as opposed to only infectious diseases (64.2%), since they believe that screening only for infectious diseases.

Table (2): Parameters related to knowledge of genetic diseases resulting from consanguineous marriages (n=402).

Parameter		***No.***	***Percent (%)***
Are you for or against consanguineous marriage?	With	78	19.4
	Against	233	58.0
	I don't know	91	22.6
Do you think that marrying a second or third-degree relative reduces the risk of genetic diseases?	Yes	125	31.1
	No	210	52.2
	I don't know	67	16.7
Do you think genetic diseases are life-threatening?	Yes	366	91.0
	No	15	3.7
	I don't know	21	5.2

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Do you think genetic diseases can be treated?	Yes	102	25.4
	No	173	43.0
	I don't know	127	31.6
Have you heard about premarital screening?	Yes	399	99.3
	I don't know	3	.7
Do you think premarital screening is limited to infectious diseases (such as hepatitis and AIDS)?	Yes	94	23.4
	No	258	64.2
	I don't know	50	12.4
Do you know what the consequences of insisting on marriage are after learning the test results are positive?	Yes	329	81.8
	No	73	18.2
Do you think it is a realistic test and you must make a serious decision about it?	Yes	393	97.8
	No	2	.5
	I don't know	7	1.7

As shown in figure (2), In particular, 393, or 97.8 percent, of respondents — all of whom supported the test's realism — expressed high levels of confidence in it. In comparison, 2 respondents out of 441 (0.5%) said No, and 7 (1.7%) were uncertain and said I don't know.

Figure (2): Illustrates the confidence of premarital testing among participants.

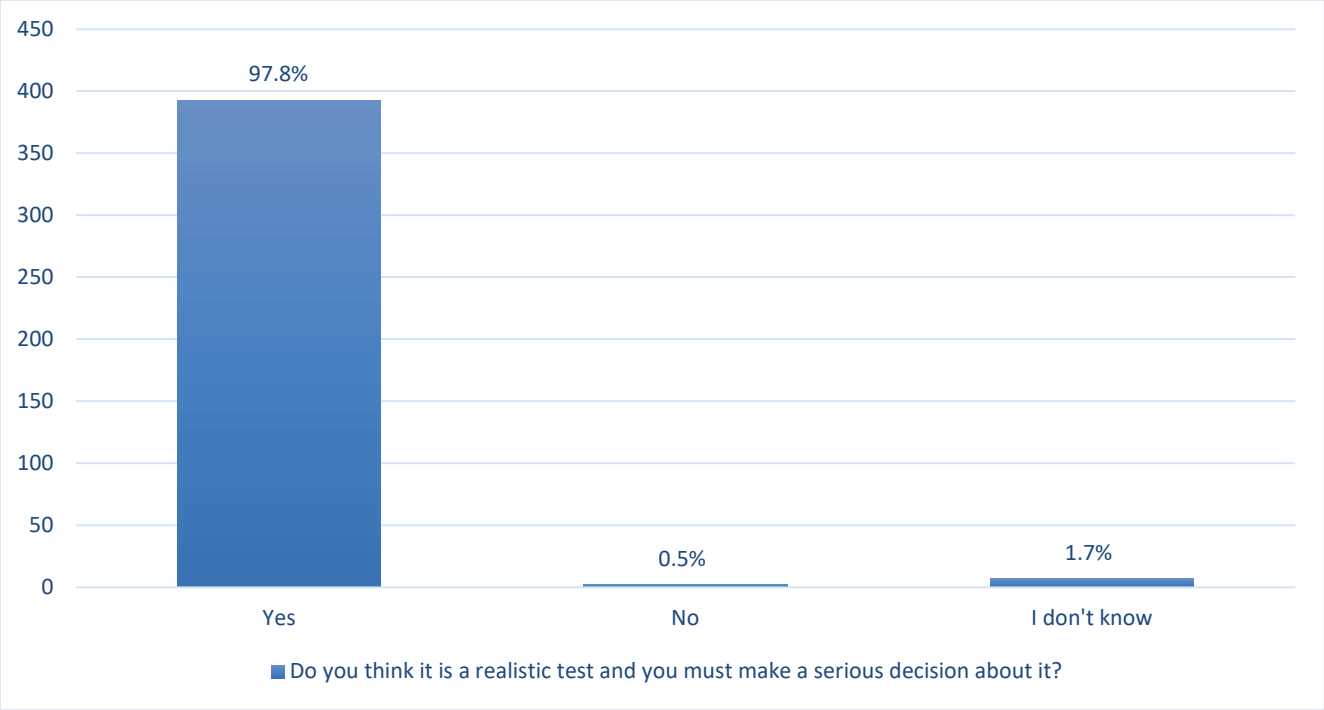

Table 3 summarizes in depth information on participants' level of awareness of genetic diseases associated to consanguineous marriages. Respondents were therefore strikingly aware of the connection between consanguinity and the health problems in offspring with 75.9% confessing the connection. Similarly, 79.6 per cent noted a likelihood of hereditary blood disorders indicating an advanced grasp

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of certain genetic issues. But while awareness of things like Down's syndrome and mental retardation, though not entirely accurate, seem better focused, with just 43.5 percent and 55.2 percent of people agreeing that these are hereditary, there is some doubt about other things. Significantly, there was a large percentage of respondents in this group that were uncertain about the relationship between deafness and consanguinity, with 32.8% of respondents saying that they did not know. Conditions such as blood cancer and asthma are also viewed this way.

Table (3): participants' awareness levels of genetic diseases resulting from consanguineous marriages (n=402).

Parameter		No.	Percent (%)
Health problems in offspring	I agree	305	75.9
	I disagree	35	8.7
	I don't know	62	15.4
Congenital disability and deformity	I agree	287	71.4
	I disagree	46	11.4
	I don't know	69	17.2
Hereditary blood disorder	I agree	320	79.6
	I disagree	21	5.2
	I don't know	61	15.2
Deafness	I agree	212	52.7
	I disagree	58	14.4
	I don't know	132	32.8
Congenital heart disease	I agree	235	58.5
	I disagree	60	14.9
	I don't know	107	26.6
Down's syndrome	I agree	175	43.5
	I disagree	109	27.1
	I don't know	118	29.4
Mental retardation (mental retardation, failure of the brain to fully develop)	I agree	222	55.2
	I disagree	65	16.2
	I don't know	115	28.6
Epilepsy	I agree	190	47.3
	I disagree	83	20.6
	I don't know	129	32.1
Asthma	I agree	207	51.5
	I disagree	100	24.9
	I don't know	95	23.6
Diabetic	I agree	283	70.4
	I disagree	60	14.9
	I don't know	59	14.7
Blood cancer	I agree	166	41.3
	I disagree	92	22.9

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	I don't know	144	35.8
Flu	I agree	60	14.9
	I disagree	253	62.9
	I don't know	89	22.1
Pneumonia	I agree	95	23.6
	I disagree	170	42.3
	I don't know	137	34.1

Table 4 presents data on the knowledge level of genetic diseases among consanguineous marriages among a population of 402 respondents. This gave a very noteworthy 39.1% of respondents a high knowledge level which was very noticeable of the effects that were involved with such unions. By contrast, 35.3% of the participants were classified as belonging to the moderate knowledge category, which indicates that many have some good understanding but that perhaps in key areas it might be lacking depth. On the other hand, 25.6 percent (respondents) comprised the low knowledge level.

Table (4): Shows knowledge of genetic diseases resulting from consanguineous marriages score results.

	Frequency	**Percent**
High knowledge Level	157	39.1
Moderate knowledge	142	35.3
Low knowledge level	103	25.6
Total	402	100.0

Table 5 describes the level of awareness of different levels of consanguineous marriage in relation to genetic disease among the surveyed populations. Significantly, 37.6% of respondents were low aware and had a critical gap in knowledge which can severely affect public health. For example, they represent 33.8% of the sample, and moderate awareness contains 28.6%.

Table (5): Shows awareness of genetic diseases resulting from consanguineous marriages score results.

	Frequency	**Percent**
High awareness level	136	33.8
Moderate awareness	115	28.6
Low awareness level	151	37.6
Total	402	100.0

Table (6) shows that knowledge of genetic diseases resulting from consanguineous marriages has statistically significant relation to age (P value=0.024), and educational level (P value=0.048). It also shows statistically insignificant relation to gender, marital status, nationality, monthly income in SAR, region, and whether the participants are for or against consanguineous marriage.

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Table (6): Relation between knowledge of genetic diseases resulting from consanguineous marriages and sociodemographic characteristics.

Parameters		***Knowledge Level***		***Total (N=402)***	***P value****
		High knowledge Level	**Moderate or low**		
Gender	Female	108	172	280	0.763
		68.8%	70.2%	69.7%	
	Male	49	73	122	
		31.2%	29.8%	30.3%	
Age	Less than 23	44	56	100	0.024
		28.0%	22.9%	24.9%	
	23 to 31	45	55	100	
		28.7%	22.4%	24.9%	
	32 to 49	44	65	109	
		28.0%	26.5%	27.1%	
	50 or more	24	69	93	
		15.3%	28.2%	23.1%	
Marital status	Single	79	98	177	0.229
		50.3%	40.0%	44.0%	
	Married	71	134	205	
		45.2%	54.7%	51.0%	
	Divorced	5	8	13	
		3.2%	3.3%	3.2%	
	Widowed	2	5	7	
		1.3%	2.0%	1.7%	
Educational level	Middle school	0	7	7	0.048
		0.0%	2.9%	1.7%	
	High school	33	61	94	
		21.0%	24.9%	23.4%	
	Bachelor's degree	109	165	274	
		69.4%	67.3%	68.2%	
	Postgraduate degree	15	11	26	
		9.6%	4.5%	6.5%	
	Uneducated	0	1	1	
		0.0%	0.4%	0.2%	
Nationality	Saudi	154	242	396	0.580
		98.1%	98.8%	98.5%	
	Non-Saudi	3	3	6	
		1.9%	1.2%	1.5%	
Monthly income in SAR	1000 to 5000	77	114	191	0.051
		49.0%	46.5%	47.5%	

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	5001 to 10000	32	60	92	
		20.4%	24.5%	22.9%	
	10001 to 20000	28	57	85	
		17.8%	23.3%	21.1%	
	More than 20000	20	14	34	
		12.7%	5.7%	8.5%	
Region	Northern region	1	4	5	0.067
		0.6%	1.6%	1.2%	
	Southern region	4	13	17	
		2.5%	5.3%	4.2%	
	Central region	81	142	223	
		51.6%	58.0%	55.5%	
	Eastern region	37	33	70	
		23.6%	13.5%	17.4%	
	Western region	34	53	87	
		21.7%	21.6%	21.6%	
Are you for or against consanguineous marriage?	With	28	50	78	0.053
		17.8%	20.4%	19.4%	
	Against	102	131	233	
		65.0%	53.5%	58.0%	
	I don't know	27	64	91	
		17.2%	26.1%	22.6%	

****P value was considered significant if ≤ 0.05.***

Table (7) shows that awareness of genetic diseases resulting from consanguineous marriages has statistically significant relation to whether the participants are for or against consanguineous marriage (P value=0.0001). It also shows statistically insignificant relation to age, gender, marital status, educational level, nationality, monthly income in SAR, and region.

Table (7): Awareness level of genetic diseases resulting from consanguineous marriages in association with sociodemographic characteristics.

Parameters		***Awareness Level***		***Total (N=402)***	***P value****
		High or moderate awareness	**Low awareness level**		
Gender	Female	176	104	280	0.793
		70.1%	68.9%	69.7%	
	Male	75	47	122	
		29.9%	31.1%	30.3%	
Age	Less than 23	66	34	100	0.053
		26.3%	22.5%	24.9%	
	23 to 31	70	30	100	
		27.9%	19.9%	24.9%	
	32 to 49	67	42	109	
		26.7%	27.8%	27.1%	

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	50 or more	48	45	93	
		19.1%	29.8%	23.1%	
Marital status	Single	120	57	177	0.268
		47.8%	37.7%	44.0%	
	Married	119	86	205	
		47.4%	57.0%	51.0%	
	Divorced	8	5	13	
		3.2%	3.3%	3.2%	
	Widowed	4	3	7	
		1.6%	2.0%	1.7%	
Educational level	Middle school	4	3	7	0.490
		1.6%	2.0%	1.7%	
	High school	64	30	94	
		25.5%	19.9%	23.4%	
	Bachelor's degree	168	106	274	
		66.9%	70.2%	68.2%	
	Postgraduate degree	15	11	26	
		6.0%	7.3%	6.5%	
	Uneducated	0	1	1	
		0.0%	0.7%	0.2%	
Nationality	Saudi	246	150	396	0.287
		98.0%	99.3%	98.5%	
	Non-Saudi	5	1	6	
		2.0%	0.7%	1.5%	
Monthly income in SAR	1000 to 5000	124	67	191	0.077
		49.4%	44.4%	47.5%	
	5001 to 10000	47	45	92	
		18.7%	29.8%	22.9%	
	10001 to 20000	58	27	85	
		23.1%	17.9%	21.1%	
	More than 20000	22	12	34	
		8.8%	7.9%	8.5%	
Region	Northern region	4	1	5	0.189
		1.6%	0.7%	1.2%	
	Southern region	10	7	17	
		4.0%	4.6%	4.2%	
	Central region	135	88	223	
		53.8%	58.3%	55.5%	
	Eastern region	39	31	70	
		15.5%	20.5%	17.4%	
	Western region	63	24	87	
		25.1%	15.9%	21.6%	
Are you for or against consanguineous	With	54	24	78	0.0001
		21.5%	15.9%	19.4%	

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marriage?	Against	162	71	233	
		64.5%	47.0%	58.0%	
	I don't know	35	56	91	
		13.9%	37.1%	22.6%	

P value was considered significant if ≤ 0.05.

Discussion:

Consanguineous marriages have been a part of human societies since the onset of modern humanity. Currently, approximately 20% of the global population resides in communities that favor such marriages [13]. The frequency of consanguinity varies among different populations, influenced by factors like religion, culture, and geographical location. Notably, many Arab nations report some of the highest levels of consanguineous marriages worldwide, with rates between 20% and 50% of all unions, particularly emphasizing first cousin marriages which have average rates of around 20% to 30%. Socio-cultural influences such as the preservation of family integrity and property, simplified matrimonial arrangements, enhanced relationships with in-laws, and financial benefits concerning dowries appear to significantly contribute to the preference for consanguinity within Arab communities [14]. It is commonly believed that consanguineous unions tend to be more stable than those between unrelated individuals, although there is a lack of studies comparing divorce rates between consanguineous and non-consanguineous marriages among Arabs. Investigations into the link between consanguinity and various reproductive health indicators have shown that about 3% to 5% of live births possess a medically significant birth defect. A recent report from March of Dimes estimated that the rate of birth defects exceeds 69.9 per 1,000 live births in many Arab nations, compared to less than 52.1 per 1,000 in Europe, North America, and Australia [15]. The risk of birth defects in first-cousin unions may be estimated to be 2 to 2.5 times higher than the general population's risk, primarily due to the manifestation of autosomal recessive disorders [16]. Thus, we aimed in this study to assess the community's knowledge and awareness of genetic diseases resulting from consanguinity.

Our findings revealed a notable opposition to consanguineous marriages, with 58.0% of respondents expressing concerns about such unions, while 75.9% acknowledge the associated health risks. This aligns with existing literature, including a study by Fatima M Elmugadam et al., [17] which found that 73.7% of respondents were aware of the potential adverse effects of intra-familial marriages on offspring health. This awareness is significantly higher than the levels reported in India and the Netherlands [18,19]. Further emphasizing the link between knowledge and attitudes, the research conducted in Riyadh [20] revealed that nearly half of the participants opposed consanguineous marriage, echoing our findings wherein 52.2% disagree with the notion that marrying a second or third-degree relative mitigates genetic disease risk. This correlation suggests an overarching trend where knowledge level directly influences attitudes towards consanguinity. Additionally, findings by Huny Bakry et al., [21] illustrate that almost half of the participants showed poor knowledge of genetic blood disorders. Additionally, most of the participants who had good knowledge were from medical and science colleges. This highlights a potential gap in public education regarding genetic health, which resonates with our discovery that only 39.1% of the surveyed population exhibits high knowledge levels about genetics. The importance of premarital screening emerges prominently across studies, with a Saudi Arabian study showing that 94.3% of participants viewed premarital carrier screening (PMCS) as a vital preventive measure against genetic disorders [22]. This finding complements our high awareness level of screening (99.3%), although we identified a misalignment, as many respondents conflated screening with infectious diseases rather than genetic risks. Furthermore, similar findings by Al-Aama et al., [23]

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and Al Sulaiman et al., [24] resonate with our results: 91% and 94% respectively recognized that blood testing is a component of PMCS aimed directly at detecting genetic disorders, illuminating a critical public health tool that is under-utilized in practical discourse. Despite the high awareness of potential genetic issues identified in our study, a stark contrast exists when considering the attitudes towards treatments for genetic diseases; only 25.4% believe in the effectiveness of available treatments, while an overwhelming 91.0% acknowledge the severe implications of genetic diseases. This skepticism towards medical advancements also mirrors findings from a study in Riyadh indicating a significant lack of understanding of genetic diseases, specifically sickle cell disease, where over two-thirds of respondents were poorly informed [25]. Similarly, knowledge deficits noted in thalassemia context in a Jeddah study [26] further illustrate the prevalent gaps in genetic disease awareness. Adding to these public health considerations, El-Hazmi et al. [27] noted that 86.9% of respondents agreed on the necessity of compulsory pre-marital examination, emphasizing a common desire for regulatory measures to mitigate genetic risks. This is particularly poignant in light of documented trends where higher frequencies of consanguineous marriages have been correlated with congenital malformations, an issue that persists across various Arab populations including those in Kuwait [28], Oman [29], Iraq [30], and Egypt [31].

Conclusion:

In conclusion, this study highlights a concerning yet informative picture of the awareness and knowledge related to genetic diseases in the context of consanguineous marriages among the Saudi population. While a significant percentage of respondents (58.0%) expressed opposition to consanguinity, indicating a shift in societal attitudes, only 39.1% demonstrated a high level of genetic knowledge. The strong awareness regarding the association between consanguinity and hereditary health risks (75.9%) contrasts with the inadequate understanding of genetic diseases and the perceived efficacy of available treatments, where only 25.4% expressed belief in effective interventions. Additionally, the confusion surrounding premarital screening—while highly recognized (99.3%)—often conflated with infectious disease testing, underscores the critical need for enhanced public education on genetic health. The findings suggest that targeted educational campaigns and stronger implementation of premarital genetic screening could significantly improve the understanding and management of genetic risks associated with consanguineous marriages, ultimately benefiting public health outcomes in Saudi Arabia.

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Ethical approval:

After fully explaining the study and emphasizing that participation is optional, each participant gave their informed consent. The information gathered was safely stored and utilized exclusively for study.

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Conflict of interests:

The authors declare no conflict of interest.

Informed consent:
Written informed consent was acquired from each individual study participant.

Data and materials availability:
All data associated with this study are present in the paper.

Reference:

1. Ricca J, Brandt JS, Jacob N, Ashkinadze E. Uptake rate of carrier screening among consanguineous couples. Prenat Diagn. 2024;(February).
2. Elmugadam FM, Ahmed H, KARAMELGHANI M, Ali A, Ali I, Ahmed A, et al. Awareness of consanguineous marriage burden and willingness towards premarital genetic testing in Sudan: a national cross-sectional study. Ann Med Surg. 2024;3959–71.
3. Albitar SM, Alokly MA, Osman A, Saad S. The Demographic , Social and Economic Dimensions of Consanguineous Marriage and its Relationship to Genetic Diseases : Sickle Cell Anemia as a Model. 8984:155–68.
4. Alrefaei A, Ashoor A, Alzahrani I, Tarbiyyah I, Alrehily M, Alharbi R, et al. Knowledge and awareness of genetic diseases among residents of the western region of Saudi Arabia. J Biochem Clin Genet. 2021;4(April):85–92.
5. Niaz M, Ali SH. Relationship of Consanguineous Marriages and Genetic Disabilities Among Children. Glob Anthropol Stud Rev. 2021 Dec 30;IV(II):33–41.
6. Alahdal H, Alshanbari H, Almazroa H, Alayesh S, Alrhaili A, Alqubi N, et al. Consanguinity, awareness, and genetic disorders among female university students in Riyadh, Saudi Arabia. J Biochem Clin Genet. 2021;4(September 2020):27–34.
7. El-Hosany AE, Khaton SE. Tanta Scientific Nursing Journal Knowledge and Attitudes among Tanta University Students Regarding to Genetic Disorders and Genetic Counseling. Vol. 21. 2021.
8. Ahmad Basuliman O, Faris Filimban G, Sami Alsabbah O, Ahmad Alzhrani K, Talal Fadhl M, Abdulhafeez Mahbub I, et al. Public Awareness of the Consanguinity and Genetic Disease Prevalence Connection in Saudi Arabia. Asian J Pharm Res Heal Care. 2020;12(4):213–22.
9. Ahmed AE, Alharbi OA, Al-hamam AA, Al-shaia WA, Hala M, Bagha M. Awareness of Health Consequences of Consanguineous Marriages among Saudi Adults. J Public Heal Dev Ctries [Internet]. 2016;2(1):121–9. Available from: https://www.jphdc.org/index.php/jphdc/article/download/42/30%0Ahttps://www.semanticscholar.org/paper/Awareness-of-Health-Consequences-of-Consanguineous-Ahmed-Alharbi/856fc9958eed844a379d67af2c1fb525100d122b#citing-papers
10. Karbouji M, AlGain R, Althaher A, Alhejaili S, AlAyoubi S. Studying the relationship between the educational level and the society awareness toward the risk of the consanguinity marriage in Al-Madinah. Int J Med Dev Ctries. 2017;3(August):107–16.
11. Alotaibi MS, Irfan UM. The knowledge, attitude and perception of saudi adult population on consanguinity and genetic disorders. In: Proceedings of the International Conference on Advances in Nursing, Pharmaceutical and Medical Sciences: ANPMS-2017, Kuala Lumpur, Malaysia. 2017. p. 20–1.
12. El-Beshbishy HA, Alsabilah TZ, Fayadh BK, Alghamdi AA, Alharbi MM, Barasheed SM, et al. Community Awareness and KnowledEl-Beshbishy, H. A., Alsabilah, T. Z., Fayadh, B. K., Alghamdi, A. A., Alharbi, M. M., Barasheed, S. M., Nawawi, E. F., Allehyani, A., Almutairi,

H. J., & Alzahrani, K. (2023). Community Awareness and Knowledge Assessment To. Arch Pharm Pract. 2023;14(1–2023):1–9.
13. Modell B, Darr A. Science and society: genetic counselling and customary consanguineous marriage. Nat Rev Genet. 2002;3:225–229. doi: 10.1038/nrg754. [DOI] [PubMed] [Google Scholar]
14. Bittles AH. A community genetics perspective on consanguineous marriage. Community Genet. 2008;11:324–330. doi: 10.1159/000133304. [DOI] [PubMed] [Google Scholar]
15. Christianson A, Howson C, Modell B. Global Report on Birth Defects The Hidden Toll of Dying and Disabled Children March of Dimes Birtn Defects Foundation. White Plains, New York; 2006. [Google Scholar]
16. Jaber L, Halpern GJ, Shohat M. The impact of consanguinity worldwide. Community Genet. 1998;1:12–17. doi: 10.1159/000016130. [DOI] [PubMed] [Google Scholar]
17. Elmugadam FM, Ahmed H, Karamelghani M, Ali A, Ali I, Ahmed A, Salman M, Mohamed W, Ahmed EA, Abbasher Hussien Mohamed Ahmed K, Mustafa Ahmed GE, Elsayed L, Musa A. Awareness of consanguineous marriage burden and willingness towards premarital genetic testing in Sudan: a national cross-sectional study. Ann Med Surg (Lond). 2024 May 28;86(7):3959-3971. doi: 10.1097/MS9.0000000000002210. PMID: 38989216; PMCID: PMC11230780.
18. Joseph N, Pavan KK, Ganapathi K, et al. 'Health awareness and consequences of consanguineous marriages: a community-based study'. J Prim Care Commun Health 2015;6:121–127. [DOI] [PubMed] [Google Scholar]
19. Morren M, Rijken M, Baanders AN, et al. 'Perceived genetic knowledge, attitudes towards genetic testing, and the relationship between these among patients with a chronic disease'. Patient Educ Couns 2007;65:197–204. [DOI] [PubMed] [Google Scholar]
20. Mahboub S.M., Alsaqabi A.A., Allwimi N.A., Aleissa D.N., Al-Mubarak B.A. Knowledge and attitude towards consanguineous marriage among educated adults in Riyadh. Int. J. Community Med. Public Health. 2018;6:30–37. doi: 10.18203/2394-6040.ijcmph20185223. [DOI] [Google Scholar]
21. Bakry H, Alaiban RA, Alkhyyat AA, Alshamrani BH, Naitah RN, Almoayad F. Predictors of Consanguinity Marriage Decision in Saudi Arabia: A Pilot Study. Healthcare (Basel). 2023 Jul 3;11(13):1925. doi: 10.3390/healthcare11131925. PMID: 37444759; PMCID: PMC10340424.
22. El-Hazmi MA. Pre-marital examination as a method of prevention from genetic disorders. Commun View Saudi Med J. 2006;27:1291–1295. [PubMed] [Google Scholar]
23. Al-Aama JY. Attitudes towards mandatory national premarital screening for hereditary hemolytic disorders. Health Policy. 2010;97:32–37. doi: 10.1016/j.healthpol.2010.02.009. [DOI] [PubMed] [Google Scholar]
24. 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;32:531–538. doi: 10.1080/03630260802508384. [DOI] [PubMed] [Google Scholar]
25. Al-Qattan H.M., Amlih D.F., Sirajuddin F.S., Alhuzaimi D.I., Alageel M.S., Bin Tuwaim R.M., Al Qahtani F.H. Quantifying the Levels of Knowledge, Attitude, and Practice Associated with Sickle Cell Disease and Premarital Genetic Counseling in 350 Saudi Adults. Adv. Hematol. 2019;2019:3961201. doi: 10.1155/2019/3961201. [DOI] [PMC free article] [PubMed] [Google Scholar]
26. .Olwi D.I., Merdad L.A., Ramadan E.K. Thalassemia: A prevalent disease yet unknown term

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among college students in Saudi Arabia. J. Community Genet. 2018;9:277–282. doi: 10.1007/s12687-017-0351-3. [DOI] [PMC free article] [PubMed] [Google Scholar]
27. El-Hazmi MAF, Al-Swailem AR, Warsy AS. Consanguinity among the Saudi Arabian population. J Med Genet. 1995;32:623–626. doi: 10.1136/jmg.32.8.623. [DOI] [PMC free article] [PubMed] [Google Scholar]
28. Madi SA, Al-Naggar RL, Al-Awadi SA, Bastaki LA. Profile of major congenital malformations in neonates in Al-Jahra region of Kuwait. East Mediterr Health J. 2005;11:700–706. [PubMed] [Google Scholar]
29. Sawardekar KP. Profile of major congenital malformations at Nizwa Hospital, Oman: 10-year review. J Paediatr Child Health. 2005;41:323–330. doi: 10.1111/j.1440-1754.2005.00625.x. [DOI] [PubMed] [Google Scholar]
30. Hamamy HA, al-Hakkak ZS. Consanguinity and reproductive health in Iraq. Hum Hered. 1989;39:271–275. doi: 10.1159/000153871. [DOI] [PubMed] [Google Scholar]
31. Temtamy SA, Abdel Meguid N, Mazen I, Ismail SR, Kassem NS, Bassiouni R. A genetic epidemiological study of malformations at birth in Egypt. East Mediterr Health J. 1998;4:252–259. [Google Scholar]