

4 ORIGINAL REPORT

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

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12 ABSTRACT

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

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

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

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

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

32 Introduction

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

Saudi Arabia has the highest incidence of CM, which 42  
is reported to be approaching about 60%. Such practice 43  
has its own risk, specifically when it comes to inherited 44

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**Received:** 16 November 2025 | **Accepted:** 26 December 2025



45	disorders (3). The possibility of parents sharing the same	and practice (KAP) regarding cousin marriages, their	106
46	recessive genes in CM is considerably greater than in	association with genetic disorders, and the premarital	107
47	non-consanguineous unions. Consequently, populations	screening program in the Western region of Saudi Arabia.	108
48	with a high prevalence of consanguinity experience an	The findings would provide updated insights that can	109
49	increased occurrence of autosomal recessive disorders	inform culturally sensitive public health strategies and	110
50	(4). The chance of passing on these illnesses between	effectively address the health risks associated with cousin	111
51	parents with the same mutated gene is a 25% chance of	marriages.	112
52	the child developing the disease and a 50% chance of		
53	them being carriers of the mutated gene (5).	<b>Materials and Methods</b>	113
54	In Saudi Arabia, one of the major autosomal recessive	<i>Study design and setting</i>	114
55	disorders with high prevalence is the recessively	This study employed a descriptive cross-sectional	115
56	inherited blood disorder, namely sickle cell anemia and	design using an online questionnaire to investigate KAP	116
57	Thalassemia (6). To decrease their incidence, Saudi	regarding CM among adults in the Western region of	117
58	Arabia launched a mandatory premarital screening	Saudi Arabia, from June to July 2025.	118
59	program in 2004, which includes blood tests to screen for		
60	Sickle cell anemia and Thalassemia as well as infectious	<i>Study population and eligibility criteria</i>	119
61	diseases (HIV, Hepatitis B, and Hepatitis C). Following	The target population comprised adults aged 18 years and	120
62	a marriage proposal, each couple must undergo these	above residing in the Western region of Saudi Arabia.	121
63	tests before being able to proceed with the marriage	Inclusion criteria were: (1) adults aged 18 years and	122
64	contract. In terms of inherited blood disorders, if both	above, (2) individuals residing in the Western region of	123
65	couples are identified as carriers, they would be labeled	Saudi Arabia, (3) both genders, and (4) individuals who	124
66	as incompatible and offered genetic counseling, but the	consented to participate and could adequately respond	125
67	decision to proceed with this marriage is left up to them	to the questionnaire. Exclusion criteria included: (1)	126
68	(7).	participants who did not complete the survey, (2) those	127
69	Around the world, there have been different strategies	who refused to participate, and (3) visitors to the region	128
70	for autosomal recessive disease prevention. For example,	who were not permanent residents.	129
71	the Jewish community tests for the most prevalent and		
72	severe diseases in their communities, such as cystic	<i>Sample size determination and sampling</i>	130
73	fibrosis, spinal muscular atrophy, and some of the	<i>technique</i>	131
74	neurodegenerative disorders, before marriage or even	The sample size was calculated using the Raosoft sample	132
75	before conception (8,9). A broader approach became	size calculator. Based on the Western region population of	133
76	more popular in the year 2021, where premarital carrier	8,021,463 according to the Saudi census 2022 (16), with	134
77	screening uses whole exome sequencing (WES) for	a 95% confidence interval, 50% anticipated frequency,	135
78	couples entering a consanguineous union to identify	and accounting for design effect, the minimum required	136
79	disease-causing variants in more than 500 genes that	sample size was determined to be 385 participants. Data	137
80	are responsible for more than 800 prevalent autosomal	collection was conducted electronically through social	138
81	recessive disorders (10,11). If both couples are found	media platforms using a snowball sampling technique to	139
82	to be carriers for disease-causing variants in the same	reach the target population.	140
83	gene, they would be identified as having a high risk for		
84	their future children to develop this genetic disorder. In	<i>Data collection instrument</i>	141
85	the United Arab Emirates, premarital WES became an	A structured questionnaire was developed based	142
86	essential part of the premarital screen in the year 2025	on extensive literature review and comprised	143
87	(12,13). However, this remained optional for high-risk	four main sections: (1) demographic information	144
88	couples in Saudi Arabia.	including age, gender, education level, employment	145
89	In Saudi Arabia, these initiatives have effectively	status, marital status, and residence details; (2)	146
90	targeted the intended population and identified couples	knowledge assessment containing eight items	147
91	at high risk (14). There is still a significant number	evaluating understanding of genetic diseases, their	148
92	of couples who decided to enter CM or proceed with	prevalence, preventability, and the relationship	149
93	marriage despite being identified as high risk (15). This	between CMs and genetic disorders; (3) attitude	150
94	behavior could be explained by cultural beliefs about	evaluation using 7 Likert-scale items (ranging from	151
95	the importance of CM, as well as a lack of knowledge	“strongly disagree” to “strongly agree”) measuring	152
96	about its associated risks and a poor understanding	participants’ attitudes toward CM and premarital	153
97	of the premarital screen, its available options, and	genetic screen; and (4) practice assessment	154
98	limitations.	examining actual behaviors and decision-making	155
99	While previous research had examined cousin marriages	processes related to CM and premarital genetic	156
100	in some regions, there is a lack of studies specifically	screen. Three experts in the field reviewed the	157
101	focusing on the Western region of Saudi Arabia.	questionnaire, and a pilot study of 30 participants	158
102	Furthermore, many existing studies do not address	(10% of the sample) was conducted.	159
103	perception toward recent public health interventions,		
104	specifically premarital WES. This study aimed to address		
105	these gaps by investigating the knowledge, attitude,		

160	<b>Knowledge domain scoring</b>	216
161	Knowledge was assessed through eight binary (Yes/No)	217
162	questions and one multiple-choice question regarding	218
163	premarital screening components. Each correct response	219
164	to binary questions received one point, while incorrect	
165	responses received zero points. For the multiple-choice	
166	question on premarital screening components, scoring	
167	was based on the selection of evidence-based correct	
168	options: “Infectious Diseases,” “Hereditary blood	
169	disorders,” and “All genetic disorders.” The knowledge	
170	domain score was calculated as the percentage of correct	
171	responses across all knowledge items, with a maximum	
172	possible score of 100%.	
173	<b>Attitude domain scoring</b>	220
174	Attitudes were measured using 7 Likert-scale items, each	221
175	scored from 1 to 5 points based on response favourability	222
176	toward genetic health awareness and responsible	223
177	marriage practices. Items were scored as follows:	224
178	“Strongly Disagree” = 1, “Disagree” = 2, “Neutral” =	225
179	3, “Agree” = 4, “Strongly Agree” = 5. For items where	226
180	disagreement indicated a positive attitude (e.g., “CM is	227
181	acceptable even without family genetic history”), reverse	228
182	scoring was applied (1→5, 2→4, 3→3, 4→2, 5→1).	229
183	The attitude domain score was calculated as the sum of	230
184	individual item scores, with a maximum possible score	231
185	of 35 points, which was also converted to a percentage	232
186	for comparative analysis.	
187	<b>Data collection procedures</b>	233
188	The Arabic-language questionnaire was distributed	234
189	electronically via Google Forms through various social	235
190	media platforms such as WhatsApp, Telegram, and	236
191	Twitter. The survey link was accompanied by clear	237
192	explanations of the study objectives, target population	238
193	criteria, and voluntary participation information.	239
194	<b>Statistical analysis</b>	240
195	Data analysis was conducted using R v 4.3. Descriptive	241
196	statistics were calculated for all variables, with continuous	242
197	data presented as means ± standard deviations or medians	243
198	with interquartile ranges depending on distribution	244
199	normality. Categorical variables were presented as	245
200	frequencies and percentages. For univariate analysis,	246
201	group comparisons were performed using Student’s <i>t</i> -test	247
202	or Mann-Whitney <i>U</i> test for continuous variables, and chi-	248
203	square tests for categorical variables. Post-hoc analyses	249
204	using Tukey’s Tukey’s Honestly Significance difference	250
205	were conducted for multiple group comparisons, with	251
206	results presented using superscript letters to indicate	252
207	significant differences between groups.	253
208	Multivariate linear regression models were constructed	254
209	to identify predictors of knowledge, attitude, and total	255
210	scores. Independent variables included gender, age	256
211	group, education level, employment status, and marital	257
212	status, with appropriate reference categories established	258
213	for meaningful interpretation. Model fit was assessed	259
214	using <i>R</i> -squared values, and regression coefficients were	
215	presented with 95% confidence intervals. Statistical	
	significance was set at $p < 0.05$ for all analyses. Score	216
	categorization was performed using predetermined cut-	217
	off points: low ( $<50\%$ ) and high ( $\geq 75\%$ ) for knowledge	218
	and attitude scores.	219
	<b>Ethical considerations and data management</b>	220
	Ethical approval was obtained from the Umm Al-	221
	Qura University Institutional Research Board prior	222
	to data collection (Approval number (HAPO-	223
	02-K-012-2025-04-2630) on 10/05/2024. Participant	224
	confidentiality was maintained through a combined	225
	system of codes, numbers, and pseudonyms, with no	226
	identifiable personal information collected. Access to	227
	data was restricted to the research team members only.	228
	All survey responses were collected anonymously,	229
	and participation was entirely voluntary with informed	230
	consent obtained electronically before questionnaire	231
	completion.	232
	<b>Results</b>	233
	Among the 1,789 participants, the sample was	234
	predominantly females (70.3%) and young adults aged	235
	21-30 years (42.3%). Most held a university degree	236
	(58.0%) and were single (57.4%). Employment was	237
	largely outside the healthcare field (60.2%). Of the 762	238
	who responded to the consanguinity question, 33.2%	239
	reported being in a CM. Among those, 25.2% were	240
	directly related as children of an aunt or uncle, while	241
	66.3% reported no biological relation despite possibly	242
	sharing a family name (Table 1).	243
	The highest correct response rate was for the item linking	244
	CM to genetic disorders (85.5%), followed closely by	245
	recognition of the seriousness of genetic diseases (83.5%)	246
	and awareness that a child might inherit a genetic disease	247
	from a parent (82.2%). In contrast, only 51.6% correctly	248
	identified the availability of premarital carrier screening	249
	(premarital WES), indicating a significant knowledge	250
	gap. Other areas with moderate correct response rates	251
	included the preventability of genetic diseases (73.3%),	252
	understanding child risk when both parents are carriers	253
	(70.0%), and awareness of genetic disease prevalence	254
	(67.6%). The sibling’s risk of inheriting a genetic	255
	disease was correctly identified by 63.2% of participants.	256
	Notably, the lowest correct rate was for identifying	257
	screening components included in the premarital test,	258
	with only 26.4% responding correctly (Figure 1).	259
	Participants showed strong support for genetic screening	260
	initiatives. Agreement was highest for comprehensive	261
	testing being beneficial ( $4.28 \pm 0.97$ ) and for mandating	262
	such testing ( $4.16 \pm 0.99$ ), with the majority expressing	263
	either agreement or strong agreement. Conversely,	264
	concerns about screening violating privacy were low,	265
	as most participants strongly disagreed with this notion	266
	( $1.74 \pm 1.12$ ). Attitudes toward CM were more variable.	267
	While participants agreed that CM increases genetic risk	268
	even in the absence of family history ( $3.56 \pm 1.11$ ), many	269
	also expressed openness to such unions. Specifically,	270
	mean scores were moderate for willingness to consider	271
	CM ( $3.43 \pm 1.19$ ) or to marry a relative with a known	272
	genetic history ( $3.39 \pm 1.23$ ). The statement asking if	273
	the current premarital screen showed a compatible result	274

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

Variable	Frequency (Percentage) <i>n</i> (%)
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 ( <i>n</i> = 762)	
Yes	253 (33.2)
No	509 (66.8)
Type of consanguineous relationship ( <i>n</i> = 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.

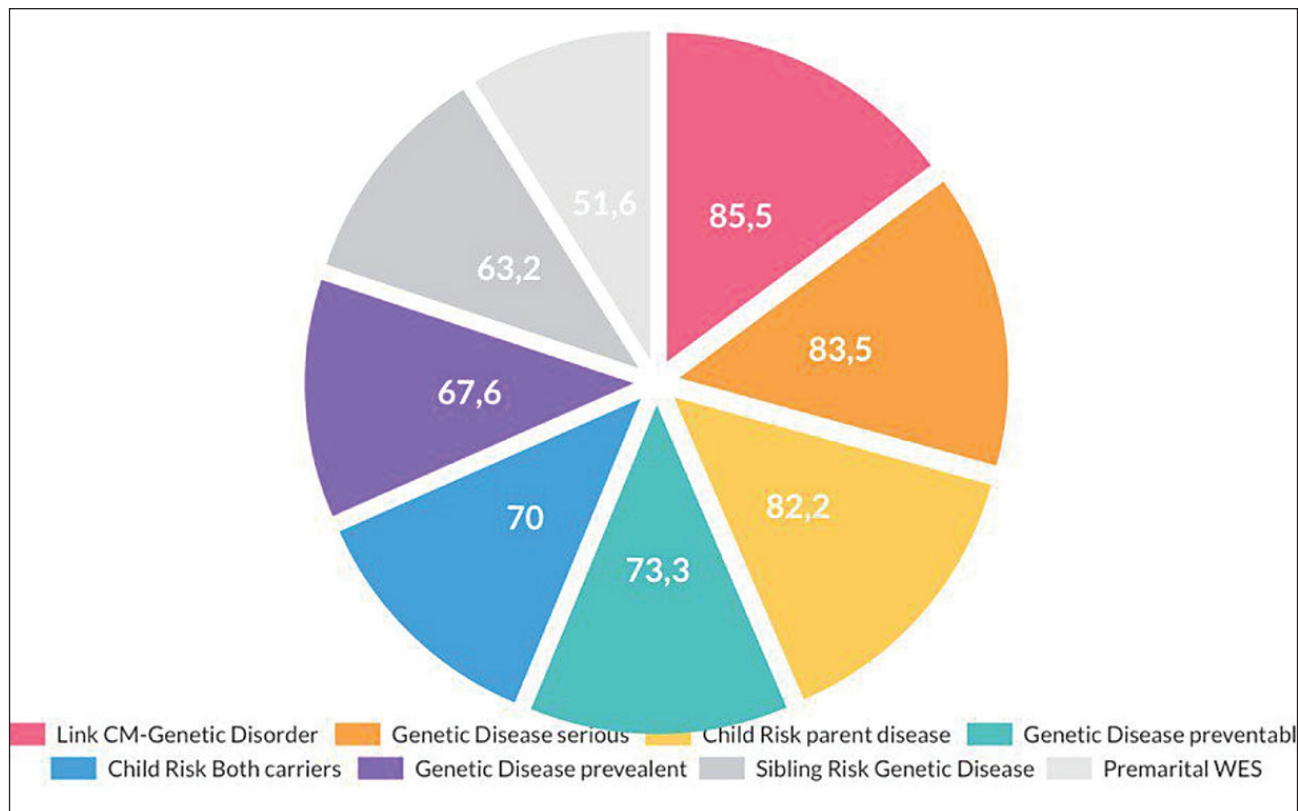
Domain	Mean	SD	Median	Min	Max	Above 50%	Below 50%
Knowledge % (9 items)	73.40	26.40	77.80	0.00	111.10	1,446 (80.8%)	343 (19.2%)
Attitude % (7 items)	70.10	9.10	71.40	37.10	100.00	1,761 (98.4%)	28 (1.6%)

means there would be no risk for genetic statement received mixed ( $3.12 \pm 1.20$ ), 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





291 **Figure 1.** Percentage of correct response to knowledge assessment questions.

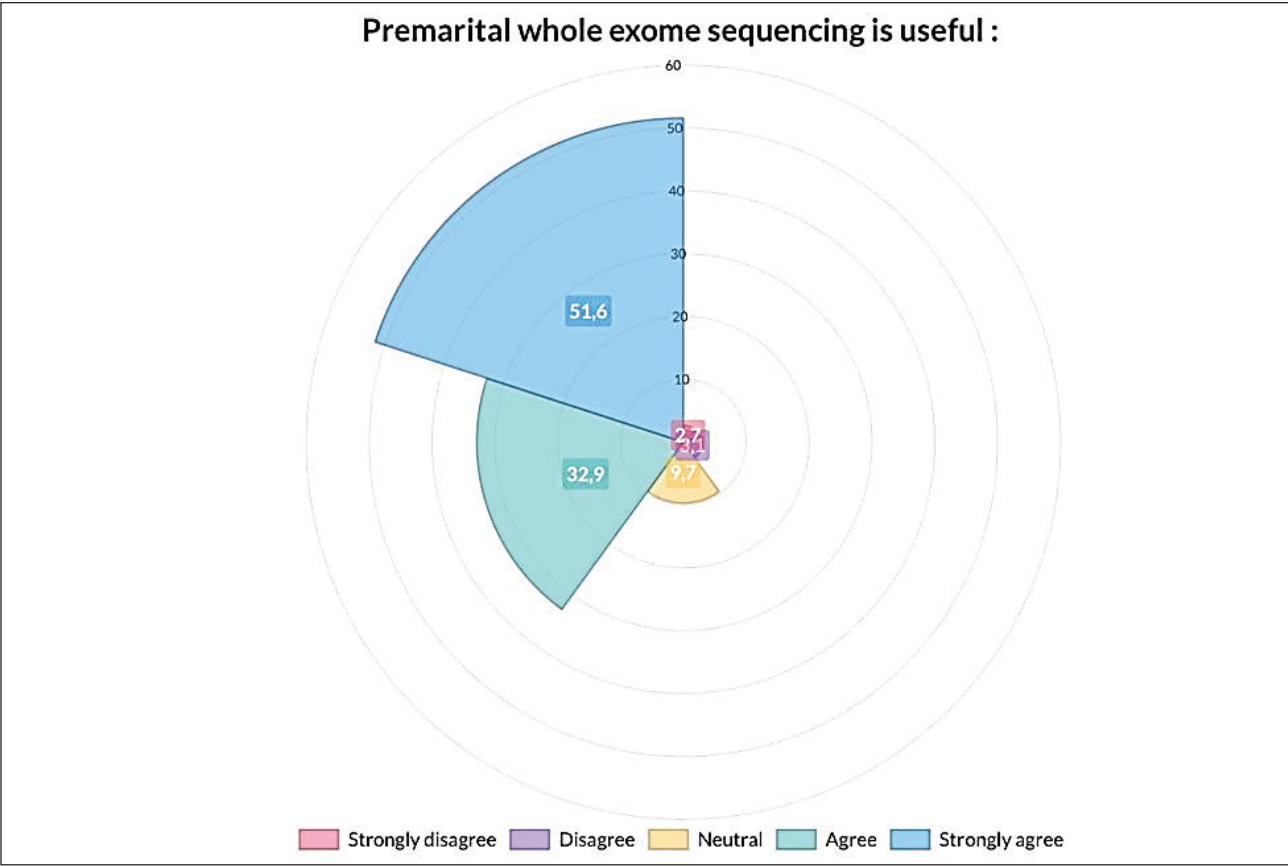
Would Marry Relative with Genetic History	3.39 (1.23)	11.9%	10.0%	23.8%	36.1%	18.3%
Mandatory Screening Violates Privacy	1.74 (1.12)	59.9%	20.2%	9.5%	6.4%	4.0%
Considering Consanguineous Marriage	3.43 (1.19)	10.0%	10.3%	24.8%	36.4%	18.4%
Consanguineous Marriage Increases Risk Without Family History	3.56 (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%
Mean (SD)		Strongly disagree	Disagree	Neutral	Agree	Strongly agree

292 **Figure 2.** Attitude assessment toward CM and genetic testing.

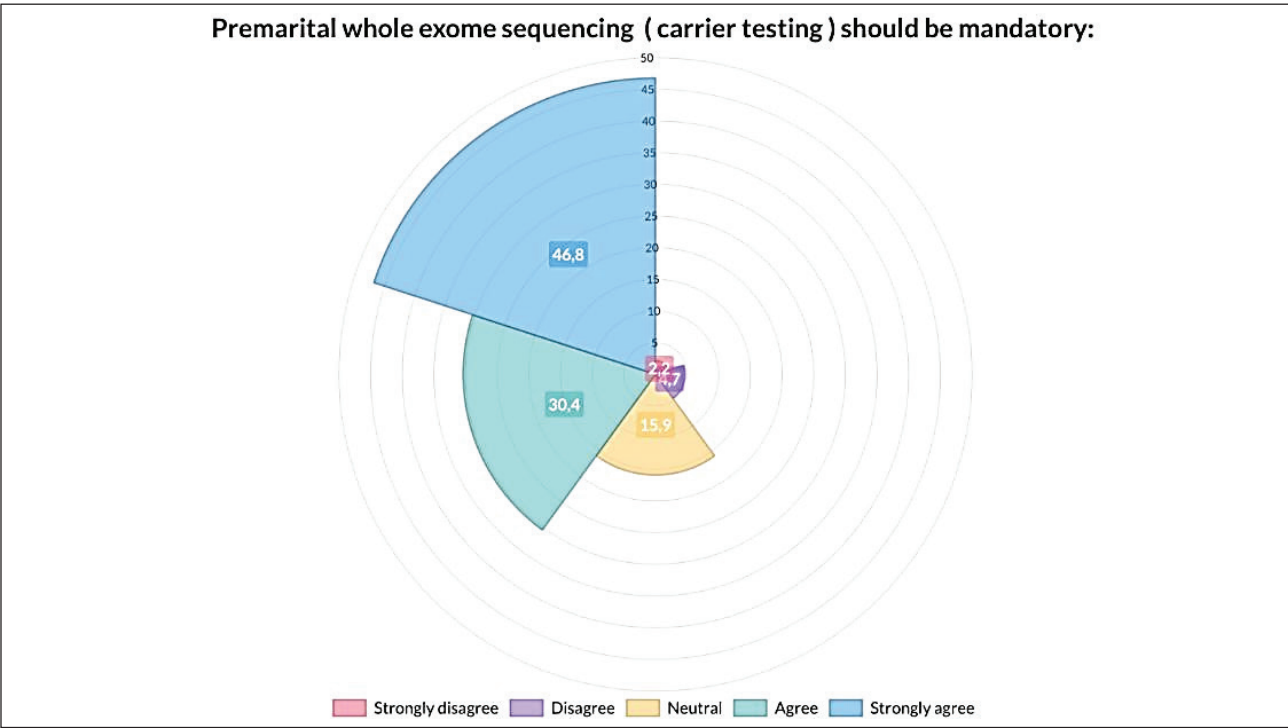
293 ( $n = 417$ , 23.3%) or the presence of a family history  
 294 of genetic disorders ( $n = 362$ , 20.2%) as potential  
 295 motivators. A few participants reported that they  
 296 would do the test if entering a CM ( $n = 151$ , 8.4%).  
 297 About 23.3% ( $n = 417$ ) indicated that they would need  
 298 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  $73.4\% \pm 26.4\%$  (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,



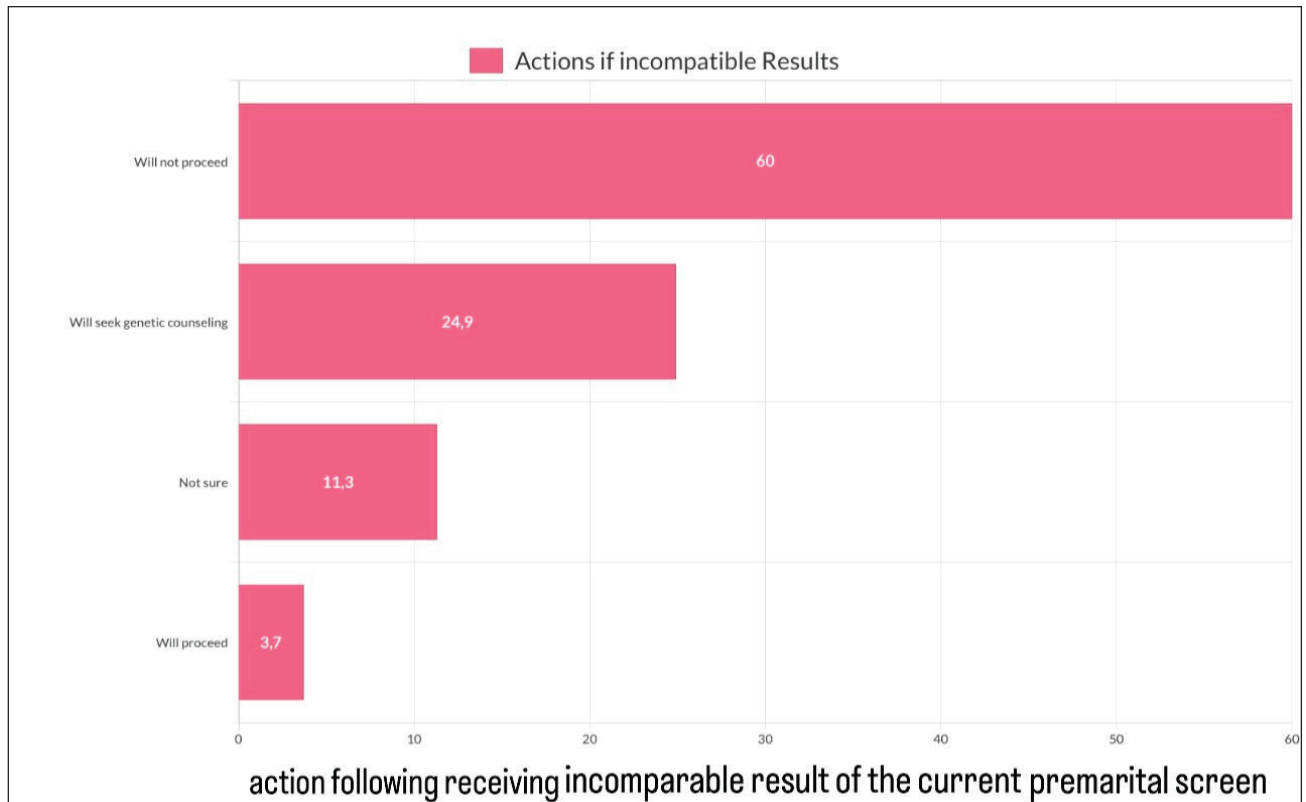
305 **Figure 3.** Participants' perceptions of the usefulness of premarital WES.



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

307 with a mean of  $70.1 \pm 9.1$  (median = 71.4), and 98.4% ( $n$   
308 = 1,761) of participants scored above the 50% threshold  
309 (Table 2).

Female participants had higher knowledge scores than 310  
males ( $74.9 \pm 26.1$  vs.  $70.1 \pm 26.9$ ;  $p < 0.001$ ), as well 311  
as higher attitude scores ( $24.9 \pm 3.1$  vs.  $23.7 \pm 3.2$ ;  $p <$  312



**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) <sup>b</sup>	24.9 (3.1) <sup>b</sup>
	Male	70.1 (26.9) <sup>a</sup>	23.7 (3.2) <sup>a</sup>
	<i>p</i> -value	<i>p</i> < 0.001	<i>p</i> < 0.001
Age group	18-20	80.8 (22.6) <sup>a</sup>	25.0 (3.0) <sup>a</sup>
	21-30	76.2 (27.3) <sup>a</sup>	24.4 (3.4) <sup>a</sup>
	31-40	72.3 (24.8) <sup>a</sup>	24.5 (3.2) <sup>a</sup>
	41-50	71.3 (21.9) <sup>a</sup>	24.8 (3.1) <sup>a</sup>
	>51	58.9 (28.0) <sup>a</sup>	24.1 (2.9) <sup>a</sup>
	<i>p</i> -value	<i>p</i> < 0.001	<i>p</i> -value = 0.001
Education level	Less than high school	55.0 (29.9) <sup>b</sup>	22.7 (3.2) <sup>b</sup>
	High school	75.5 (25.6) <sup>a</sup>	24.5 (2.9) <sup>a</sup>
	Diploma	59.3 (24.9) <sup>b</sup>	24.4 (3.0) <sup>a</sup>
	University	76.1 (25.1) <sup>a</sup>	24.7 (3.2) <sup>a</sup>
	MSc	63.9 (28.0) <sup>b</sup>	23.8 (3.8) <sup>ab</sup>
	PhD	68.3 (34.4) <sup>ab</sup>	24.2 (3.9) <sup>ab</sup>
	<i>p</i> -value	<i>p</i> < 0.001	<i>p</i> < 0.001
Employment	Other	68.3 (25.7) <sup>b</sup>	24.4 (3.0) <sup>a</sup>
	HCP	74.3 (28.0) <sup>a</sup>	24.8 (3.4) <sup>a</sup>
	Doctor/Resident/Medical Student/Genetic Counselor	87.9 (21.1) <sup>c</sup>	24.6 (3.5) <sup>a</sup>
	<i>p</i> -value	<i>p</i> < 0.001	<i>p</i> -value = 0.209
Marital status	Single	77.9 (25.7) <sup>b</sup>	24.6 (3.2) <sup>a</sup>
	Married	67.1 (26.3) <sup>a</sup>	24.5 (3.1) <sup>a</sup>
	Divorced	72.1 (24.3) <sup>ab</sup>	24.1 (3.4) <sup>a</sup>
	Widowed	64.1 (25.8) <sup>a</sup>	24.2 (3.2) <sup>a</sup>
	<i>p</i> -value	<i>p</i> < 0.001	<i>p</i> -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 ( $80.8 \pm 22.6$ ), while those older than 51 years had the lowest ( $58.9 \pm 28.0$ ;  $p < 0.001$ ). Regarding education, university graduates ( $76.1 \pm 25.1$ ) and high school graduates ( $75.5 \pm 25.6$ ) scored significantly higher than those with less than high school education ( $55.0 \pm 29.9$ ;  $p < 0.001$ ). In terms of employment, doctors, residents, and medical students had the highest knowledge scores ( $87.9 \pm 21.1$ ), significantly surpassing other groups (e.g.,  $68.3 \pm 25.7$  among non-health workers;  $p < 0.001$ ). Single participants also had higher knowledge scores ( $77.9 \pm 25.7$ ) compared to married ( $67.1 \pm 26.3$ ) and widowed individuals ( $64.1 \pm 25.8$ ;  $p < 0.001$ ). 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.



374 This means that cultural beliefs and family norms could  
375 overcome knowledge. Recent systematic reviews also  
376 indicated that, although awareness was relatively high  
377 in Saudi Arabia, cultural and family traditions often  
378 continue to override health-based decision-making  
379 (17,18).

380 Another systematic review in the Middle East found  
381 that although many countries had mandated premarital  
382 screening, this did not decrease the incidence of CM  
383 effectively in any of them (19). Another study from Jazan  
384 targeted couples who proceeded with their marriage  
385 decision despite incompatibility in premarital screen  
386 found out that more than half of them rejected the  
387 counselling advice as their marriage was unavoidable,  
388 30% thought that no clear explanation provided to them,  
389 18% thought the risk of transmitting the disease to their  
390 children was low and 5% thought that their children life  
391 would not be affected by the disease (20).

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

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

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

432 The current study community might not be willing to  
433 avoid CM due to cultural pressure, but they were willing  
434 to seek help in terms of possible prevention. This was

observed in their attitude and action toward premarital 435  
testing. Most of the participants in the current study did 436  
not believe that mandatory premarital screening violates 437  
their privacy; rather, about 80% thought that premarital 438  
WES should be Mandatory. Further, 40% thought that 439  
they would do premarital WES if they got married. 440  
Similarly, a study in the Northern region determined that 441  
when people were asked, “would you add a test to make 442  
the premarital screen broader”, > 90% answered yes 443  
(23). This meant that the current study community was 444  
not against the testing and prevention, but the marriage 445  
choice was possibly a limiting factor. 446

It would be hard for such programs to be effective if 447  
people do not understand them well, or do not know 448  
they exist, or the meaning of the result. Public education 449  
concentrating on the area of genetic disease prevention, 450  
specifically premarital screening, components, and 451  
limitations, would be of huge significance. If premarital 452  
screening were expanded to include prevalent and 453  
severe disorders in Saudi Arabia or even WES, it would 454  
be accepted by the public based on their response 455  
in these surveys. In addition to premarital screening 456  
effectiveness in decreasing the disease prevalence, it is 457  
also cost-effective. Rabea et al. (24) compared the cost 458  
of premarital screening for spinal muscle atrophy to the 459  
cost of interventions required for diagnosed patients and 460  
found it to be less by 14 to 28 fold. Such studies do not 461  
exist for premarital WES, as it is a relatively new practice. 462

Several recommendations could be addressed, such as 463  
government-based intervention to consider expanding 464  
the current premarital screen to involve prevalent 465  
and severe disorders in Saudi Arabia, or even WES 466  
with frequent evaluation to assess its effectiveness. 467  
Also, public education about the available preventive 468  
measures, specifically the premarital screening program 469  
and the availability of the optional premarital WES. 470  
Social media, a heavily used platform, should be used in 471  
this education. Last but not least, counseling following 472  
premarital testing compatibility should not only include 473  
proceeding or not proceeding with the marriage, but also 474  
be widened to discuss further intervention, such as *in vitro* 475  
fertilization and preimplantation genetic diagnosis, or 476  
early pregnancy detection and termination, if it remains 477  
within the scope of religion, if the couple is planning to 478  
proceed with a high-risk marriage. Longitudinal studies 479  
following the introduction of these educational programs 480  
to evaluate their effectiveness are also required. 481

Despite the huge and respected efforts, limitations 482  
could exist. The cross-sectional design cannot 483  
establish causality. The mostly young sample limited 484  
generalizability, and online snowball sampling might 485  
introduce selection bias. Self-reported data could not 486  
reflect real behavior. 487

## 488 Conclusion

While general knowledge and attitudes toward genetic 489  
screening were favorable, substantial educational gaps 490  
persisted, particularly regarding available testing and 491  
residual genetic risk. Although people thought that 492  
consanguinity was a cultural norm, they were willing to 493  
proceed with preventive actions. Hence, improving public 494

- 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**
- The authors of this article have no affiliations with or involvement in any organization or entity with any financial interest or non-financial interest in the subject matter or materials discussed in this manuscript.
- Funding**
- None.
- Consent to participate**
- Informed consent was obtained from all the participants.
- Ethical approval**
- 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).
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- References**
- 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>
  - 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>
  - 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>
  - 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>
  - Taglia A, Picillo E, D'Ambrosio P, Cecio MR, Viggiano E, Politano L. Genetic counseling in Pompe disease. *Acta Myol.* 2011;30(3):179
  - 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>
  - 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>
  - 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>
  - 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>
  - 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>
  - 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>
  - 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>
  - 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>
  - Bakry H, Alaiban RA, Alkhyat 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>
  - 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>
  - 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>
  - 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>
  - 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.*

- 615 2024;18(1):138. [https://doi.org/10.1186/s40246-024-](https://doi.org/10.1186/s40246-024-00700-x) 632  
616 00700-x 633
- 617 19. Saffi M, Howard N. Exploring the effectiveness of 634  
618 mandatory premarital screening and genetic counselling 635  
619 programmes for  $\beta$ -thalassaemia in the Middle East: a 636  
620 scoping review. *Public Health Genomics*. 2015;18(4):193– 637  
621 203. <https://doi.org/10.1159/000430837> 638
- 622 20. Gosadi IM, Gohal GA, Dalak AE, Alnami AA, Aljabri 639  
623 NA, Zurayyir AJ. Assessment of factors associated 640  
624 with the effectiveness of premarital screening for 641  
625 hemoglobinopathies in the South of Saudi Arabia. *Int J* 642  
626 *Gen Med*. 2021;14:3079–86. [https://doi.org/10.2147/](https://doi.org/10.2147/IJGM.S321046) 643  
627 *IJGM.S321046* 644
- 628 21. Al Eissa MM, Almsned F, Alkharji RR, Aldossary YM, 645  
629 AlQurashi R, Hawsa EA, et al. The perception of 646  
630 genetic diseases and premarital screening tests in the 647  
631 central region of Saudi Arabia. *BMC Public Health*. 648  
2024;24(1):1556. [https://doi.org/10.1186/s12889-024-](https://doi.org/10.1186/s12889-024-19029-0) 649  
19029-0
22. Makkawi M, Alasmari S, Hawan AA, Al Shahrani MM, Dera 634  
AA. Hemoglobinopathies. *Saudi Med J*. 2021;42(7):784– 635  
9. <https://doi.org/10.15537/smj.2021.42.7.20210273> 636
23. Hafiz MN, Suhail N, Mohammed ZMS, Elzein HO, 637  
Alasmoum HA, Abass AE, et al. Awareness and 638  
attitude of the general population towards inherited 639  
hemoglobinopathies in the premarital screening 640  
program in the Northern Region of Saudi Arabia. 641  
*Hematol Rep*. 2025;17(1):9. [https://doi.org/10.3390/](https://doi.org/10.3390/hematolrep17010009) 642  
*hematolrep17010009* 643
24. Rabea F, El Naofal M, Chekroun I, Khalaf M, Zaabi N 644  
Al, AlZaabi K, et al. Spinal muscular atrophy genetic 645  
epidemiology and the case for premarital genomic 646  
screening in Arab populations. *Commun Med*. 647  
2024;4(1):1–6. [https://doi.org/10.1038/s43856-024-](https://doi.org/10.1038/s43856-024-00548-1) 648  
00548-1 649