

Knowledge, Attitude and Perception of Medical Students about Consanguineous Marriages and Associated Patterns of Genetic Diseases in Arar, Saudi Arabia: A Descriptive Cross-sectional Survey

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ABSTRACT

Introduction: Consanguineous Marriage (CONM) is a well-recognised contributor to Genetic Diseases (GD) and adverse reproductive outcomes, particularly in Saudi Arabia where CONM prevalence is high. Understanding medical students' knowledge, attitudes, and perceptions toward CONM and GD is critical for shaping future clinical practice in this context.

Aim: To evaluate the knowledge, attitudes, and perceptions regarding CONM and GD among medical students at Northern Border University (NBU), Arar, Saudi Arabia.

Materials and Methods: The present descriptive cross-sectional survey was conducted between January and July 2024 among medical students at the College of Medicine, Northern Border University (NBU). A structured, self-administered questionnaire comprising 23 items across four domains (sociodemographic data, consanguinity and family history, knowledge of CONM-related consequences, and knowledge of GD risk factors) was used.

Results: A total of 233 students participated {mean age 21.6±1.8 years; 132/233 (56.7%) males}. Two-thirds reported CONM as prevalent in Saudi Arabia 153/233 (65.7%). Nearly, all recognised its association with GD 224/233, (96.1%) and were aware of Premarital Screening (PMS) services 222/233 (95.3%). Fewer than half had received formal information about CONM 112/233 (48.1%), while more than half expressed a need for further training in genetic counselling 128/233 (54.9%).

Conclusion: Medical students at NBU demonstrated strong awareness of CONM prevalence and its link to GD, alongside widespread recognition of Premarital Screening (PMS) services. Nonetheless, critical gaps remain in quantitative genetic risk assessment, particularly in recurrence-risk estimation and distinguishing chromosomal from monogenic disorders. The findings highlight an urgent need for targeted curricular revision integrating Mendelian inheritance, recurrence-risk calculation, and genetic counselling skills to better prepare future clinicians for practice in high-consanguinity populations.

Keywords: Awareness, Disease Alleles, Genetic abnormalities, Genetic defects, Genetic mutations

INTRODUCTION

The CONM refers to a marital union between individuals who are genetically closely related. This practice has gained considerable attention in the medical and genetic sciences due to its implications for future generations. Observed in various cultures especially in regions such as the Middle East CONM raises significant concerns regarding the risk of genetic abnormalities in the offspring born from these unions. Children from CONMs are at a heightened risk for congenital disorders and inherited diseases that can adversely affect their health and development [1]. The prevalence of CONM varies by race and religion, despite its global practice [2].

The primary concern associated with CONM is the increased risk of GD. When closely related individuals reproduce, the likelihood that both parents carry the same genetic mutation increases. As a result, there is a higher probability that recessive GDs will manifest in their descendants. Because related couples are more likely to share the same recessive disease alleles, the children of CONMs are more susceptible to inheriting two copies of a harmful recessive variant, predisposing them to genetic defects and hereditary illnesses [2,3].

Globally, consanguinity accounts for up to 10% of the population, with rates varying significantly, from approximately 80.6% in certain Middle Eastern regions to less than 1% in Western countries [4]. In Saudi Arabia, the rate of CONM is around 50%, with first-

cousin marriages being particularly prevalent and enduring across generations [5]. These unions may perpetuate both familial and socioeconomic disparities, as they are often influenced by cultural traditions or economic conditions rather than individual choice [6,7].

The high incidence of genetic illnesses among consanguineous families contributes to increased mortality and morbidity rates at a young age. Despite efforts to raise awareness about the potential risks associated with CONM, the practice continues to be common within families. One study indicated that while two-thirds of educated couples engaged in CONM were aware of the inherited genetic conditions linked to it, only a small proportion knew about the available diagnostic facilities [8].

Moreover, there is a lack of data concerning the knowledge, attitudes, and perceptions of medical students, particularly at institutions in the Northern Border Region of Saudi Arabia, which has some of the highest rates of consanguinity in the Kingdom. The existing literature does not sufficiently address whether future clinicians in this high-risk region are equipped to provide genetic counselling or assess recurrence risks [9]. Thus the present study aimed to fill that gap. Assessing university students' understanding, beliefs, and attitudes towards CONM and GDs is crucial in a culture where CONM is prevalent, as these students will become future parents and decision-makers. This study seeks to evaluate the

knowledge, attitudes, and perceptions regarding CONM and GD among medical students at NBU, Saudi Arabia.

MATERIALS AND METHODS

The present descriptive cross-sectional survey was conducted among medical students at the College of Medicine, Northern Border University (NBU), between January and July 2024. The study received ethical clearance from the local bioethical committee of NBU (HAP-09-A-043) on January 31, 2024, with decision number 10/24/H.

Inclusion criteria: Medical students of both genders from all academic years at NBU who provided informed consent were included in the study.

Exclusion criteria: Students who were unwilling to participate or who were from outside NBU were excluded.

Sample size calculation: The sample size was calculated using Epi Info version 7.2.4.0, with an expected knowledge level of 50%, a margin of error of 0.05, and a 95% confidence level. Based on these inputs, the estimated sample size was 248. Fifteen questionnaires were excluded due to missing data, resulting in a final sample size of 233 and a response rate of 94%.

- Population size = 700
- Confidence level = 95%
- Expected frequency (P) = 50%
- Margin of error (d) = 0.05
- Design effect = 1
- Cluster = 1

Thus, the required sample size was rounded to 248.

Study Procedure

A self-administered, structured questionnaire in English was developed by the first three researchers (one professor, one associate professor, and one assistant professor) after reviewing relevant literature [10,11]. A team of experts in epidemiology, public health, and family medicine reviewed and approved the instrument for content and face validity. A pilot study involving 10 students was conducted to assess the simplicity of the questionnaire and the time required to complete it. Feedback from this pilot study resulted in minor adjustments to the text.

The internal consistency of the questionnaire was assessed using Cronbach's alpha, which yielded a value of 0.896, indicating that the instrument meets the study's objectives.

The questionnaire comprised four components with a total of 23 items:

- A. The first part collected sociodemographic information, including age, gender, academic grade, family income, father's education, mother's education, and paternal consanguinity (9 items).
- B. The second section included questions regarding consanguinity and the family history of hereditary and congenital diseases (6 items), assessing knowledge and attitudes about these topics.
- C. The third part contained questions about the participants' knowledge of the potential consequences of consanguinity (4 items).
- D. The fourth section had questions concerning participants' understanding of the risk factors for GDs, including recurrence-risk estimation for autosomal recessive conditions (4 items).

Therefore, the instrument assessed knowledge in detail across sections two to four, while sections one and two also captured relevant attitudinal items (e.g., parental preferences for consanguinity and perceived prevalence of CONM).

After obtaining lists of all sections, one section from each academic grade was randomly selected using a simple random sampling

method. From the selected sections, students who consented to participate were sequentially recruited by the researchers via a secure online tool (Google forms), ensuring broad access and maintaining participant anonymity until the desired sample size was reached.

STATISTICAL ANALYSIS

Data were entered, cleaned, and analysed using Statistical Package for Social Sciences (SPSS) V24 (IBM SPSS Statistics for Windows, Version 24.0, Armonk, NY: IBM Corp). Categorical data were presented as frequencies and percentages.

RESULTS

Of the 233 students studied, the mean age was 21.6±1.8 years with a range of 18 to 27 years. Male participants made up over half the sample 132 (56.7%). Additionally, 160 (68.7%) of the participants had university-educated fathers, and 151 (64.8%) had university-educated mothers.

Parental consanguinity included over 40% of participants 107 (45.9%); first cousins constituted 41.1%; and somewhat more than sixty percent 143 (61.4%) favoured marrying from unrelated families [Table/Fig-1].

Items		n (%)
Age (Mean±SD) (years)	21.6±1.8	
Gender	Male	132 (56.7)
	Female	101 (43.3)
Academic grade	1 st	22 (9.4)
	2 nd	25 (10.7)
	3 rd	44 (18.9)
	4 th	38 (16.3)
	5 th	71 (30.5)
	6 th	19 (8.2)
	Intern	14 (6.0)
Father education	High or less	73 (31.3)
	University	160 (68.7)
Mother education	High or less	82 (35.2)
	University	151 (64.8)
Family income per month	5000 SR or less	24 (10.3)
	5000-10000 SR	60 (25.8)
	More than 10000 SR	149 (63.9)
Parents' consanguinity [#]	Yes	107 (45.9)
	No	126 (54.1)
Degree of consanguinity	First cousin	44 (41.1)
	Second cousin	29 (27.1)
	More distant	34 (31.8)
Who prefer to marry	First cousin	12 (5.2)
	Second cousin	8 (3.4)
	Not related	143 (61.4)
	Don't know	70 (30)

[Table/Fig-1]: Sociodemographic distribution of participants (N=233).
[#]Of the 233 participants, 91 (39.1%) were preclinical students (1st-3rd academic year)

Two-thirds of participants acknowledged that CONM is widespread in Saudi Arabia, with the majority associating CONM with GD 224 (96.1%) and recognising the PMS 222 (95.3%).

Twenty-five per cent of respondents reported a familial history of GDs 59 (25.3%), while a small fraction indicated the existence of CONMs 31 (13.3), mostly Down syndrome 10 (32.3%) [Table/Fig-2].

Sixty percent of participants said that CONM is associated with a heightened probability of having children with intellectual disability, whereas fifty percent affirmed that CONM increases the chance of

Items	n (%)
How common is CONM in your country?	
Common	153 (65.7)
Not common	23 (9.9)
#I do not know	57 (24.5)
Is CONM associated with GD in the offspring?	
Yes	224 (96.1)
No	9 (3.9)
Do you know about the PMS service provided by the Ministry of Health, KSA that identifies Genetic Disorders (GD)?	
Yes	222 (95.3)
No	11 (4.7)
Family history of inherited disease	
Yes	59 (25.3)
No	174 (74.7)
Family history of children with Congenital Malformation (CONM)	
Yes	31 (13.3)
No	202 (86.7)
If yes, what type?*	
Down syndrome	10 (32.3)
Others, such as Patau syndrome, Edward syndrome, Turner syndrome, Klinefelter syndrome, and Congenital hip dysplasia	21 (67.7)
[Table/Fig-2]: Distribution of the respondents' knowledge of CONM, family history of inherited and congenital disorders.	
*31	

Down syndrome. Only 27 (11.6%) believed that non-consanguineous couples with no family history of congenital abnormalities have a 2.5% risk of having a child with one. Furthermore, a tiny minority 28 (12%) correctly recognised that for consanguineous couples with no family history of congenital deformity, the likelihood of birthing a child with a congenital defect is 5%.

For couples with autosomal recessive conditions, such as beta thalassemia, around one-quarter 56 (24%) correctly identified that a carrier couple for an autosomal recessive condition, such as beta-thalassaemia, has a 25% probability of having an affected child with each pregnancy, consistent with Mendelian recessive inheritance. This figure (24%) is the proportion of students who selected the correct response; the theoretical recurrence risk itself is 25%.

Slightly less than half 112 (48.1%) thought that first-cousin marriage increased the risk of having Down syndrome, and slightly less than 40% 90 (38.6%) claimed it is associated with a higher abortion rate [Table/Fig-3].

Items	n (%)
The type of marriage is linked to an increased likelihood of having children with intellectual disabilities.	
Consanguineous	140 (60.2)
Non-consanguineous	8 (3.4)
None	29 (12.4)
Don't know	56 (24)
Is CONM associated with an increased chance of having a kid with Down syndrome?	
Yes	116 (49.8)
No	36 (15.5)
I don't know	81 (34.8)
In non-consanguineous couples with no family history of congenital abnormalities, the risk of having a child with a congenital anomaly is	
0 %	24 (10.3)
0.5%	30 (12.9)
2.5%	27 (11.6)
5%	18 (7.7)

10%	11 (4.7)
Don't know	123 (52.8)
For consanguineous couples with no family history of congenital deformity, the chance of having a kid with a congenital anomaly is	
0 %	15 (6.4)
0.5%	42 (18.0)
5%	28 (12)
10%	20 (8.6)
Don't know	128 (54.9)
Couples who are carriers of an autosomal recessive condition, such as beta thalassemia, have a higher probability of having an afflicted kid.	
0%	13 (5.6)
0.5%	10 (4.3)
2.5%	10 (4.3)
5%	15 (6.4)
10%	11 (4.7)
25%	56 (24)
Don't know	118 (50.6)
First-cousin marriages increase the likelihood of having a child with a congenital condition.	
Yes	130 (55.8)
No	21 (9.0)
I don't know	82 (35.2)
First-cousin marriages increase the likelihood of getting a child with Down syndrome.	
Yes	112 (48.1)
No	28 (12.0)
I don't know	93 (39.9)
Do first-cousin marriages have a higher abortion rate?	
Yes	90 (38.6)
No	38 (16.3)
I don't know	105 (45.1)
[Table/Fig-3]: Participants' knowledge of the potential consequences of consanguinity.	

DISCUSSION

In the present study, 107/233 (45%) of participants reported parental consanguinity, while 143/233 (61.4%) expressed a preference to marry outside their family. These findings align with regional variability: parental relatedness has been reported at 48% in Jeddah, Saudi Arabia [12], 43.8% in the UAE [13], 64% [14], and 50.7% [15]. Comparable figures include over 40% in Qatar [16], 36% in Palestine [17], 28.9% among Palestinian university students [18], 30.2% in Sudan [19], and 27% in Turkey [20]. Thus, the prevalence observed in the present study cohort falls within the established range for populations with high consanguinity.

Despite this, more than 60% of participants preferred not to marry relatives, consistent with regional trends among younger, educated populations. For example, 57.2% of Saudis disagreed with CONM [21], 72.4% of single Omani men rejected cousin marriage [22], 54.2% of Egyptians expressed negative attitudes [23], 69.6% of Yemenis opposed cousin marriage [24], 73.9% of Palestinians reported similar views [18], and 63.9% of Sudanese respondents indicated the same [19]. These findings suggest a generational shift toward caution regarding CONM, even though the practice remains widespread.

Knowledge about CONMs and their relationship with GDs was generally high. In the present study cohort, 153/233 (65.7%) recognised the prevalence of CONMs in Saudi Arabia, 224/233 (96.1%) acknowledged the link between CONMs and GDs, and 222/233 (95.3%) were aware of the national PMS program. These results are consistent with prior studies in Saudi Arabia and the region, where awareness ranged from 75.6 to 97.5% [11,15,19,21,24-28]. Similarly, 59/233 (25.3%) reported a family history of inherited

disease, comparable to findings among university students in Qatar, Palestine (21.6-38.8%) [16-18] and Jordan [27].

However, significant gaps were evident in genomic risk knowledge. Only 56/233 (24%) correctly identified the 25% recurrence risk for autosomal recessive disorders such as beta-thalassemia, lower than the 35.5% reported by Al Naqeb D et al., among healthcare providers in Saudi Arabia [10]. This deficit is concerning, as future medical professionals will be expected to deliver genetic counselling in populations with extensive familial relations. Zakariyah AF et al., have similarly noted that genetics education is marginalised across Saudi medical universities [29].

Another issue was the high frequency of “don’t know” responses, ranging from 34.8% to 54.9% across genetics knowledge items [Table/Fig-3]. Unlike incorrect answers, these responses reflect a lack of recall or understanding, even among senior students, suggesting poor retention and application of clinical genetics concepts [23].

A further misconception was that nearly half of participants 116/233 (49.8%) incorrectly associated CONM with increased risk of Down syndrome. Since trisomy 21 results from chromosomal non-disjunction rather than autosomal recessive inheritance, consanguinity does not increase recurrence risk. This misunderstanding, also reported by Al Naqeb D et al., highlights a widespread gap in distinguishing between autosomal recessive disorders and chromosomal aneuploidies [10]. Educational programs should emphasise this distinction to prevent misinformed counselling.

Taken together, these findings reveal that medical students-who should possess greater genetics knowledge than non-medical peers-demonstrate substantial deficiencies in core concepts such as recurrence risk and disorder classification. This reflects systemic shortcomings in medical curricula and underscores the need for targeted reforms.

Limitation(s)

The descriptive design precludes causal inference, and the self-administered questionnaire is subject to recall bias. The tool was non-standardised, applied in a single Institution, and lacked a validated scoring rubric, limiting generalisability and preventing categorical judgments of “adequate” knowledge despite acceptable internal consistency (Cronbach’s $\alpha=0.896$). No inferential analyses (e.g., chi-square, logistic regression) were performed to explore associations between sociodemographic variables and knowledge outcomes. Future studies should employ standardised scoring systems, Likert-based attitudinal constructs, and robust statistical analyses to identify predictors of genetic knowledge, such as academic year, parental consanguinity, or family history of GDs.

CONCLUSION(S)

The present study shows that medical students at NBU are generally aware of the existence and consequences of CONM and its link to GDs, as well as the availability of PMS programs. However, important gaps remain in their understanding of quantitative genetic risk, particularly recurrence risk for autosomal recessive conditions and the distinction between chromosomal and monogenic disorders. Misconceptions such as linking consanguinity with Down syndrome highlight the need for clearer teaching of genetic principles. Students themselves expressed a need for more training in genetic counselling, underscoring the urgency of curricular revision. Overall, the findings point to the importance of integrating targeted genetics and counselling content into medical education, with future multi-institutional and longitudinal studies needed to evaluate the impact of such reforms.

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