Original Article :

Genetic Counseling for Hereditary Blood Disorders: Knowledge, Attitudes, and Practices of Medical Professionals and Students in Saudi Arabia

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Abstract

Background and Aims:
Hereditary blood disorders are blood diseases passed down from parents to their children. Genetic counseling can give information on how hereditary blood disorders can affect a person or their family. This study aims to evaluate the knowledge, attitudes, and practices of medical professionals and students in Saudi Arabia toward genetic counseling for hereditary blood disorders. This study aims to evaluate the knowledge, attitudes, and practices of medical professionals and students in Saudi Arabia toward genetic counseling for hereditary blood disorders.

Methods:
A cross-sectional study was conducted using a self-developed questionnaire to evaluate the knowledge, attitudes, and practices of medical professionals and students in Saudi Arabia toward genetic counseling for hereditary blood disorders. The data were analyzed statistically.

Results:
Among 676 participants, nearly 70% had poor knowledge of genetic counseling. While 81.4% considered it important, 62.1% were unsure how to refer patients. Lack of awareness (85.4%) was the main barrier. Enhancing clinical experience and incorporating genetic counseling into medical curricula have been recognized as vital strategies for improving training.

Conclusion:
The study highlights gaps in knowledge about genetic counseling for hereditary blood disorders among medical professionals and students. Despite positive attitudes, there are still uncertainties in patient referrals and significant barriers, such as lack of awareness. Enhancing education, training, and health campaigns could improve knowledge and awareness of genetic counseling.

Keywords:
Genetic Counseling, Hereditary, Blood Disorders, Knowledge, Attitudes, Practices

Introduction:

Hereditary or genetic blood disorders are a group of diseases that are passed down from parents to their children. Genetic defects can cause certain blood disorders because they lead to abnormalities in the composition and components of red blood cells, resulting in malfunction of red blood cells. The main genetic blood disorders are thalassemia and sickle cell anemia. These disorders are passed down from parent to child through the genes carried on the chromosomes. When both parents have a genetic defect, there's a 25% chance that each child will be born with the disease. However, if one parent is healthy and the other parent is carrying the trait, then the disease can pass down to some children, making them carriers of the disease. The prevalence of genetic blood disorders (sickle cell anemia and thalassemia) varies among the different regions in Saudi Arabia. The highest rates are recorded in the eastern and southern regions, while the rates are lower in the central and northern regions of the Kingdom. Genetic counseling is very important to understand how genetic blood disorders can affect a person or their family. Consulting a genetic counselor can help in preventing or reducing the chances of genetic blood disorders [1].

Genetic counselors are trained healthcare professionals who use their expertise in genetics, epidemiology, and counseling to assess genetic risks and provide education and support to patients and families for genetic conditions. Genetic counseling is an evolving field in Saudi Arabia. In 2015, genetic counseling was recognized as a master's program by the Saudi Commission for Health Specialties [2].

This study aims to evaluate the knowledge, attitudes, and practices of medical professionals and students in Saudi Arabia toward genetic counseling for hereditary blood disorders.

Methods:

Study Designs:

This cross-sectional study was conducted in Saudi Arabia with a sample of 385 participants to evaluate the knowledge, attitudes, and practices of medical professionals and students toward genetic counseling for hereditary blood disorders. The study employed an online survey to gather data from medical professionals and students aged 18 years and older. The findings will provide insights into how genetic counseling for hereditary blood disorders is perceived in the region.

Identification of deleterious nsSNPs:

To predict the functional effects of nsSNPs collected from the dbSNP and Ensembl databases, the PredictSNP tool (https://loschmidt.chemi.muni.cz/predictsnp/ , accessed on 21 June

2023) was used. This approach provided accurate and stringent results, with SNPs predicted as damaging by all six programs being classified as high-risk. PredictSNP leverages six prediction programs (MAPP, PhD-SNP, PolyPhen-1, PolyPhen-2, SIFT, and SNAP) to identify disease-related mutations. Additionally, PANTHER was employed to robustly and accurately relate protein sequence relationships to functional relationships. This algorithm classifies proteins based on their evolutionary relationships, molecular functions, and interactions with other proteins, analyzing substitutions using position-specific evolutionary conservation scores derived from alignments of evolutionarily related proteins.

Participant Recruitment:

Participants in the medical field were selected from the general adult population of Saudi Arabia. Inclusion criteria included male and female Saudi adults aged 18 years and older who were in the medical field (medical students and medical professionals) and were able to provide informed consent. Exclusion criteria included participants who were not in the medical field and participants who refused to participate or did not provide consent to participate. The survey link was distributed to eligible participants via email, WhatsApp, and other social media platforms.

Data Collection:

Data collection for this study was conducted using an online survey distributed to 385 participants. The questionnaire was self-developed and was reviewed by a panel of experts to ensure strength and reliability. It consisted of four sections: demographics, knowledge, attitudes, and practices toward genetic counseling for hereditary blood disorders. Participants were recruited through social media platforms to ensure a diverse sample. The survey was anonymous, and participants were informed about the study's objectives and were required to provide informed consent before participating in the study. To ensure participant confidentiality, all data were anonymized, securely stored, and accessed only by authorized members of the research team. To minimize bias, we used standardized data collection tools, ensured anonymous responses to limit bias, applied consistent inclusion criteria, and trained data collectors uniformly. Data were collected over nearly 2 months, from January 2025 to March 2025, to ensure sufficient participation and representation of the target population.

Sample Size Calculation:

The sample size was calculated using the Raosoft sample size calculator, which applies a standard formula for estimating sample size, based on data from the

2022 Saudi census. A sample size of 385 participants was determined to achieve a 95% confidence level and a 5% margin of error.

Data Analysis:

The data collected for this study were analyzed using SPSS version 27 (IBM Corporation, 2019). The overall knowledge score for each participant was calculated by assigning 1 point for each correct answer. The scores were then converted into percentages to assess the participants' level of knowledge about genetic counseling for hereditary blood disorders. Participants who scored below 60% were categorized as having poor knowledge, while those who scored 60% or higher were categorized as having good knowledge. Descriptive statistics, including frequencies and percentages, were computed to summarize the demographic characteristics of the study participants and their responses to the knowledge-related questions. To examine the relationship between various factors (such as region, age, gender, educational level, and specific practices) and the participants' overall knowledge levels, the chi-square test was employed. The chi-square test assessed whether significant differences existed between participants with poor knowledge and those with good knowledge based on the various factors. A p-value of less than 0.05 was considered statistically significant for all tests.

Ethical Approval:

The IMSIU Institutional Review Board (IRB) Committee approved the study (project number 745/2024; approval date, December 22, 2024). All writing was collected in accordance with the ethical principles of the Declaration of Helsinki. The survey link included a brief study description and a more detailed explanation on the front page.

Results:

Table 1 presents the personal characteristics of the study's medical professionals and students in Saudi Arabia (N=676). Among the participants, 31.8% (215) are from the Central Region, 15.4% (104) from the Northern Region, 20.3% (137) from the Eastern Region, 12.0% (81) from the Western Region, and 20.6% (139) from the Southern Region. Regarding age distribution, the majority (65.8%, 445) are between 21-29 years old, while 16.7% (113) are aged 18-20, 12.4% (84) are between 30-39 years, 3.7% (25) are in the 40-49 age range, and just 1.3% (9) are aged 50 or above. The gender shows that 46.3% (313) are male, and 53.7% (363) are female. As for educational level, most participants had a bachelor's degree (60.7%, 410), followed by 26.9% (182) with secondary education or below, 8.6% (58) with a post-graduate degree, and 3.8% (26) with a diploma.

Finally, when asked about formal training or lectures on genetic counseling, 37.7% (255) reported having attended.

Table 1: Personal characteristics of the study medical Professionals and Students in Saudi Arabia (N=676).

Personal data	No	%
Region		
Central Region	215	31.8%
Northern Region	104	15.4%
Eastern Region	137	20.3%
Western Region	81	12.0%
Southern Region	139	20.6%
Age in years		
18-20	113	16.7%
21-29	445	65.8%
30-39	84	12.4%
40-49	25	3.7%
50+	9	1.3%
Gender		
Male	313	46.3%
Female	363	53.7%
Educational level		
Secondary education / below	182	26.9%
Diploma	26	3.8%
Bachelor degree	410	60.7%
Post-graduate degree	58	8.6%
Have you ever attended any formal training or lectures on genetic counseling?		
Yes	255	37.7%
No	421	62.3%

Table 2 shows the knowledge of genetic counseling for hereditary blood disorders among medical professionals and students, and the relationship between the level of knowledge and demographic characteristics of the participants in Saudi Arabia (N=676). Regarding which disorders require genetic counseling, the majority recognized sickle cell anemia (78.4%, 530), followed by hemophilia (67.6%, 457) and G6PD deficiency (favism) (65.4%, 442). However, leukemia was only identified by 43.3%

(293), while 8.1% (55) of respondents were unsure. When asked about the primary goal of genetic counseling, 70.3% (475) indicated educating individuals, and 65.2% (441) selected the diagnosis of the disease. As for awareness of genetic testing methods for hereditary blood disorders, 34.3% (232) were aware. Additionally, many respondents were unsure about the genetic testing methods that may not work, with 49.9% (337) stating, "I don't know." Among those who provided answers, Western Blotting (16.4%, 111), Southern Blotting (16.1%, 109), and NGS (15.2%, 103) were most frequently recognized as methods that may not work, with PCR (18.3%, 124) and Karyotyping (18.5%, 125) also identified as potentially ineffective by some respondents. Finally, when asked about the importance of premarital screening in preventing hereditary blood diseases, a large majority (86.4%, 584) agreed that it reduces the burden of disease, while only 12.1% (82) felt it played no important role, and 5.8% (39) were unsure. The relationship between the level of knowledge and demographic characteristics was also addressed. The region of residence, observing or participating in genetic counseling sessions, frequency of discussing genetic risks with patients or families, and knowing how to refer patients to a genetic counselor were significantly associated with better knowledge. Participants from the Western (42.0%) and Central (37.7%) regions had higher proportions of good knowledge. Those who observed or participated in genetic counseling sessions had 38.3% good knowledge compared to 25.3% among those who did not. Participants who always discussed genetic risks had 43.2% good knowledge, while those who never did had 24.6%. Among those who knew how to refer patients to a genetic counselor, 37.5% had good knowledge compared to 26.2% who did not. Other factors such as age, gender, education level, prior training, and qualification level were not significantly associated, although participants with postgraduate degrees (39.7%) and prior training (34.5%) had higher knowledge levels.

Table 2: Knowledge of Genetic Counseling for Hereditary Blood Disorders among Medical Professionals and Students and the Relationship between Knowledge and Demographic Data of the Participants in Saudi Arabia (N=676).

Knowledge	No	%
Which of the following disorders requires genetic counseling?		
Sickle cell anemia	530	78.4%
G6PD deficiency (favism)	442	65.4%
Hemophilia	457	67.6%
Leukemia	293	43.3%
I don't know	55	8.1%
The main goal of genetic counseling		
Diagnosis of the disease	441	65.2%
Educating individuals	475	70.3%
Ensuring adherence to treatment	296	43.8%
Providing emotional support	219	32.4%
I don't know	50	7.4%
Are you aware of the genetic testing methods available for hereditary blood disorders?		
Yes	232	34.3%
No	277	41.0%
I don't know	167	24.7%
Are you aware of the genetic testing methods available for hereditary blood disorders?		
Yes	232	34.3%
No	277	41.0%
I don't know	167	24.7%
Genetic testing methods that may not work		
Western Blotting	111	16.4%
Hemoglobin Electrophoresis	137	20.3%
Southern Blotting	109	16.1%
NGS	103	15.2%
MLPA	104	15.4%
Amniocentesis	84	12.4%
Karyotyping	125	18.5%
PCR	124	18.3%
CVS	84	12.4%
I don't know	337	49.9%
The importance of premarital screening in preventing hereditary blood diseases		
Reduces the burden of disease	584	86.4%
Academic purposes only	44	6.5%
No important role	82	12.1%
I don't know	39	5.8%

The Relationship between Knowledge and Demographic Data	Overall knowledge level				p-value
	Poor		Good		
	No	%	No	%	
Region					.001*
Central Region	134	62.3%	81	37.7%	
Northern Region	85	81.7%	19	18.3%	
Eastern Region	109	79.6%	28	20.4%	
Western Region	47	58.0%	34	42.0%	
Southern Region	95	68.3%	44	31.7%	
Age in years					.153^
18-20	85	75.2%	28	24.8%	
21-29	300	67.4%	145	32.6%	
30-39	58	69.0%	26	31.0%	
40-49	18	72.0%	7	28.0%	
50+	9	100.0%	0	0.0%	
Gender					.267
Male	211	67.4%	102	32.6%	
Female	259	71.3%	104	28.7%	
Educational level					.161
Secondary education / below	128	70.3%	54	29.7%	
Diploma	22	84.6%	4	15.4%	
Bachelor degree	285	69.5%	125	30.5%	
Post-graduate degree	35	60.3%	23	39.7%	
Have you ever attended any formal training or lectures on genetic counseling?					.076
Yes	167	65.5%	88	34.5%	
No	303	72.0%	118	28.0%	
Have you observed or participated in a genetic counseling session during your clinical training, lecture, or discussion?					.001*
Yes	166	61.7%	103	38.3%	
No	304	74.7%	103	25.3%	
Do you discuss genetic risks with patients or their families?					.001*
Never	86	75.4%	28	24.6%	
Rarely	117	78.0%	33	22.0%	
Often	154	72.3%	59	27.7%	
Always	113	56.8%	86	43.2%	
Do you know how to refer a patient to a genetic counselor?					.002*
Yes	160	62.5%	96	37.5%	
No	310	73.8%	110	26.2%	

Figure 1 illustrates the overall knowledge level of genetic counseling for hereditary blood disorders among medical professionals and students in Saudi Arabia. The data shows that most participants, 69.5% (470), have a poor level of knowledge, while 30.5% (206) possess a good level of knowledge.

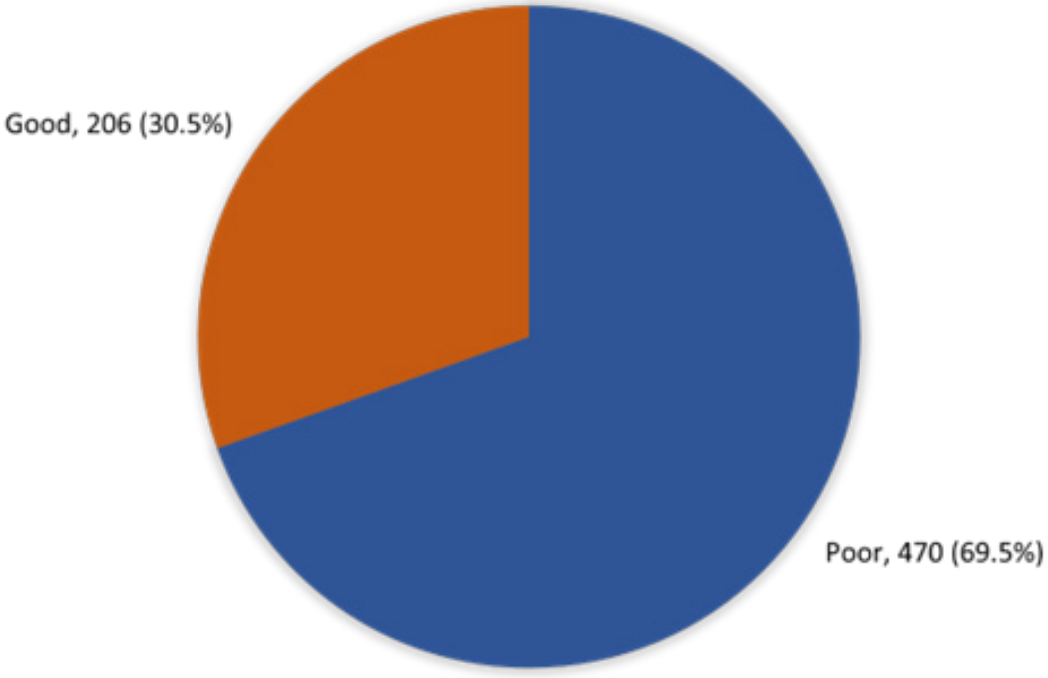


Figure1: T The overall Knowledge of Genetic Counseling for Hereditary Blood Disorders among Medical Professionals and Students in Saudi Arabia (N=676).

Table 3 illustrates the attitudes and practices of medical professionals and students in Saudi Arabia regarding genetic counseling for hereditary blood disorders. A significant majority, 81.4% (550), consider genetic counseling to be very important in managing hereditary blood disorders, with only 1.2% (8) deeming it not important. Additionally, 96.9% (655) of participants encourage patients with a family history of hereditary blood disorders to seek genetic counseling, showing a high level of advocacy for this service. When it comes to cultural and religious influences, 70.1% (474) believe that cultural or religious beliefs can influence acceptance of genetic counseling. Lastly, the majority of respondents (62.3%, 421) strongly agree that genetic counseling should be included as a routine part of medical care, with only 1.2% (8) disagreeing.

Table 3: Attitudes Toward Genetic Counseling for Hereditary Blood Disorders Among Medical Professionals and Students in Saudi Arabia (N=676).

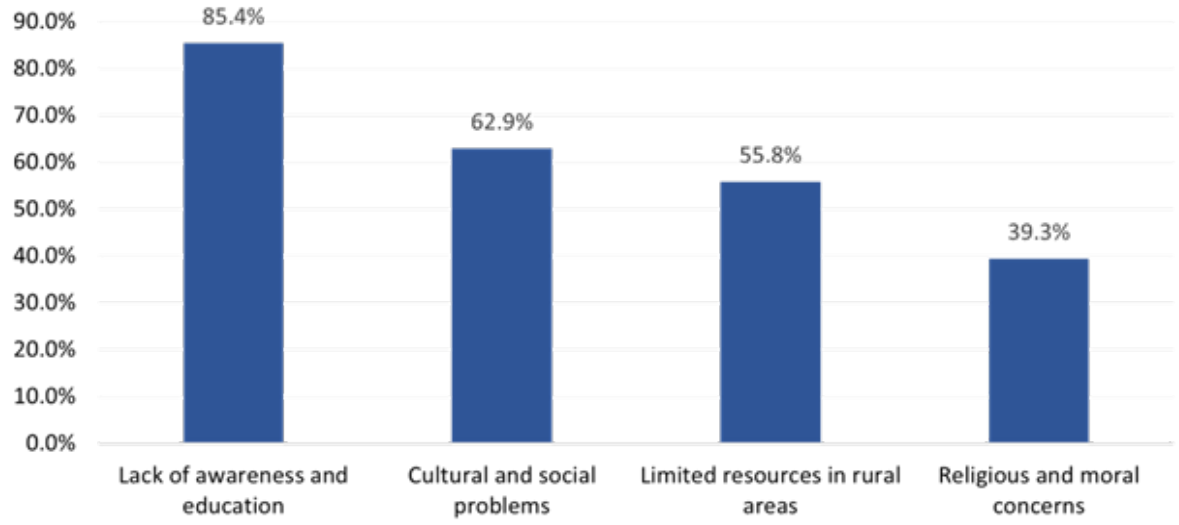
Attitude	No	%
In your opinion, how important is genetic counseling in managing hereditary blood disorders?		
Very important	550	81.4%
Somewhat important	118	17.5%
Not important	8	1.2%
Do you encourage patients with a family history of hereditary blood disorders to seek genetic counseling?		
Yes	655	96.9%
No	21	3.1%
Do you think cultural or religious beliefs can influence acceptance of genetic counseling?		
Yes	474	70.1%
No	202	29.9%
Genetic counseling should be included as a routine part of medical care		
Strongly agree	421	62.3%
Agree	201	29.7%
Neutral	46	6.8%
Disagree	8	1.2%

Table 4 provides the participation and practices related to genetic counseling among medical professionals and students in Saudi Arabia. When asked if they had observed or participated in a genetic counseling session during their clinical training, lecture, or discussion, only 39.8% (269) reported having done so, while 60.2% (407) had no such experience. About discussing genetic risks with patients or their families, 31.5% (213) of respondents reported discussing genetic risks often, and 29.4% (199) always engage in such discussions. When asked about referring patients to genetic counselors, a significant majority, 62.1% (420), were unsure of how to make such referrals, while 37.9% (256) knew how to refer a patient.

Table 4: Participation and Practices in Genetic Counseling Among Medical Professionals and Students in Saudi Arabia (N=676).

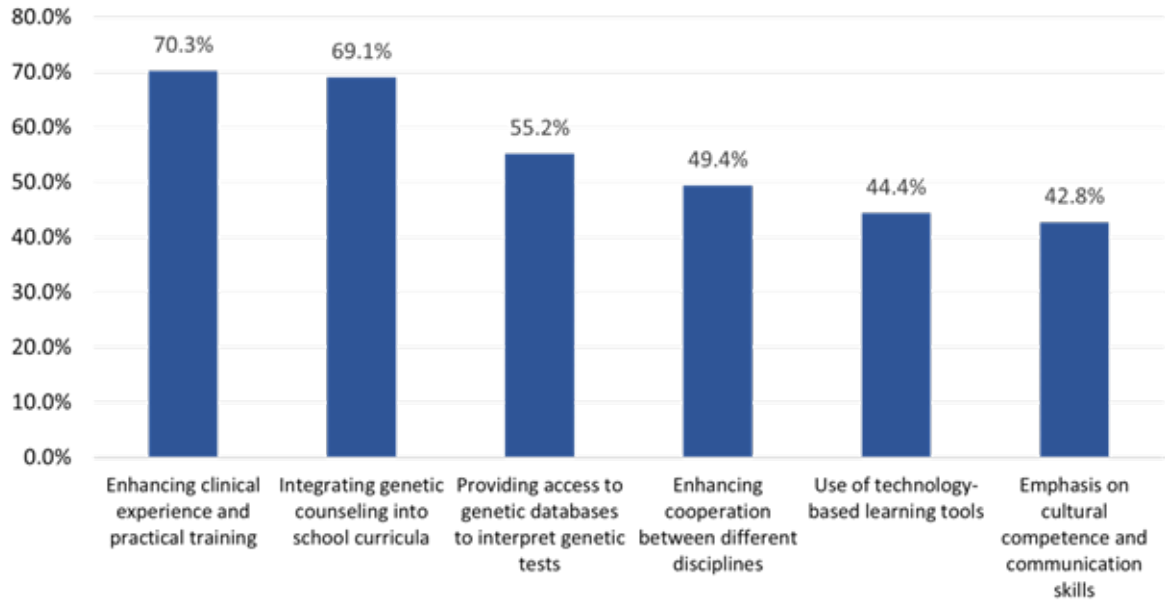
Practice	No	%
Have you observed or participated in a genetic counseling session during your clinical training, lecture, or discussion?		
Yes	269	39.8%
No	407	60.2%
Do you discuss genetic risks with patients or their families?		
Never	114	16.9%
Rarely	150	22.2%
Often	213	31.5%
Always	199	29.4%
Do you know how to refer a patient to a genetic counselor?		
Yes	256	37.9%
No	420	62.1%

Figure 2 highlights the reported barriers to implementing genetic counseling for hereditary blood disorders among medical professionals and students in Saudi Arabia. The most significant barrier identified by 85.4% (577) of participants is the lack of awareness and education. Cultural and social problems were reported by 62.9% (425) of respondents, and limited resources in rural areas were reported by 55.8% (377), pointing to a disparity in access to genetic counseling, particularly outside urban centers. Lastly, religious and moral concerns were identified by 39.3% (266).



Figer2: The Reported Barriers to implementing genetic counseling for hereditary blood disorders.

Figure 3 outlines several methods for improving genetic counseling training for medical students and healthcare workers. The most favored approach, cited by 70.3% (475) of participants, is enhancing clinical experience and practical training. Integrating genetic counseling into school curricula also received strong support, with 69.1% (467) of respondents advocating for its inclusion, 55.2% (373) of participants recommending providing access to genetic databases to interpret genetic tests. The use of technology-based learning tools (44.4%, 300) and an emphasis on cultural competence and communication skills (42.8%, 289) were also seen as important strategies to enhance training. Exactly 49.4% (334) of participants supported enhancing cooperation between different disciplines.



Figer3: Methods for improving genetic counseling training for medical students and healthcare workers.

Discussion:

The current study aimed to evaluate the knowledge, attitudes, and practices of medical professionals and students in Saudi Arabia toward genetic counseling for hereditary blood disorders. Study participants included both medical professionals and students from across Saudi Arabia, representing all major regions, with the Central Region having the most participants. Most participants were in their twenties. Also, there was nearly equal gender distribution. The majority had a bachelor's degree, while some had postgraduate qualifications. Remarkably, about one-third of the group had formal training or had attended lectures on genetic counseling.

As for knowledge, our study findings showed varying levels of awareness

regarding genetic counseling for hereditary blood disorders. While sickle cell anemia, hemophilia, and G6PD deficiency were mostly recognized as conditions requiring genetic counseling, leukemia was less frequently identified. This inconsistency may reflect differences in exposure to these conditions in clinical practice or medical education, as sickle cell disease and thalassemia are more prevalent in the region compared to hereditary forms of leukemia[3,4]. As for education and disease diagnosis, the primary goals of genetic counseling match well-known guidelines, which focus on the importance of patient empowerment and accurate risk assessment[5]. However, the limited awareness of genetic testing methods, together with the high proportion of respondents who were unsure about ineffective techniques, indicates a need for further training in molecular diagnostics. The strong agreement on the value of premarital screening corresponds with Saudi Arabia's national screening program, which has been instrumental in reducing the incidence of hemoglobinopathies [6]. Nevertheless, the persistence of some uncertainty highlights the importance of continuous public and professional education to reinforce the preventive role of genetic counseling in hereditary blood disorders.

Generally, our study revealed that medical professionals and students in Saudi Arabia had limited knowledge regarding genetic counseling for hereditary blood disorders, a finding consistent with literature findings. This knowledge deficit is not exceptional to Saudi Arabia, as similar defects have been observed in different populations in the U.S. and Australia, where awareness of genetic counseling for hereditary conditions is markedly low [7]. Furthermore, socioeconomic barriers, including financial constraints, geographical challenges, and lack of awareness, significantly limit access to genetic counseling services[8]. While the theoretical importance of genetic counseling, particularly for risk assessment and patient education, is generally acknowledged[9], a disconnect exists between this recognition and its practical application, especially concerning genetic testing methods. This gap is also evident in South Africa, where healthcare professionals displayed limited knowledge of referral processes and the availability of genetic counseling services[10]. However, some regions, like Nigeria, show higher awareness, particularly regarding premarital genetic counseling, suggesting that targeted public health initiatives can be effective [11]. Eventually, the challenge lies in translating the recognized value of genetic counseling into practical knowledge and effective implementation, necessitating

targeted interventions such as improved medical education, clearer referral systems, and the integration of genetic counselors into multidisciplinary healthcare teams.

Locally, Similar to our results, Al-Shroby et al.[12] showed that 69.5% of participants had poor knowledge of genetic counseling. A large-scale study found that only 9.2% of the general Saudi population had "satisfactory" knowledge of premarital screening and genetic counseling (PMSGC. However, our study focused specifically on medical professionals and students, suggesting that even those with medical training may lack sufficient expertise in this specialized area. This contrasts with another Saudi study by Arafah et al.[13], where 87% of respondents (mostly students and young adults) expressed willingness to undergo genetic testing before marriage. Our study found that leukemia was less frequently recognized as requiring genetic counseling compared to sickle cell anemia or hemophilia. This is consistent with findings from a study by Olwi et al.[14], where students demonstrated good general genetics knowledge but lacked depth in specific areas.

The attitudes toward genetic counseling among Saudi medical professionals and students were highly positive, with a large majority recognizing its importance and actively encouraging patient referrals, which is a significant confirmation aligning with much Saudi support for premarital genetic screening [12,13]. This strong advocacy contrasts favorably with international studies that report referral barriers among non-genetics specialists[11], suggesting a potentially more receptive environment in Saudi Arabia for integrating genetic services. However, a significant portion approved of the potential influence of cultural and religious beliefs on acceptance. This high level of professional support, coupled with existing public approval, creates a promising basis for implementing comprehensive genetic counseling frameworks in Saudi Arabia.

The implementation of genetic counseling for hereditary blood disorders in Saudi Arabia faces several significant barriers, as identified by medical professionals and students. The most prominent challenge is the lack of awareness and education. Similar findings were reported by previous studies that even healthcare providers often have insufficient training in clinical genetics, limiting their ability to deliver or refer patients for genetic counseling [15-17]. Cultural and social barriers were noted by about two-thirds of participants, reflecting the influence of familial and societal factors on healthcare decisions. Studies have shown that stigma, misconceptions about genetic diseases, and family dynamics can deter individuals from seeking counseling[3].

Additionally, limited resources in rural areas indicate gaps in healthcare access, with genetic services primarily concentrated in urban centers. Finally, religious and moral concerns are still a consideration, particularly regarding sensitive topics such as prenatal testing and reproductive choices. While Islamic teachings generally support preventive medicine, ethical dilemmas may arise in clinical practice [18].

Regarding suggestions to improve, participants strongly favored enhancing clinical experience and integrating genetic counseling into school curricula to improve training. Access to genetic databases, technology-based learning, and cultural competence training were also seen as valuable. Additionally, interdisciplinary cooperation was considered important.

Study Limitations:

This study has several limitations that must be considered when interpreting the results. Firstly, since this is a cross-sectional study, it shows the results at a single point in time. Second, the study relied on self-reported data, so there is a risk of response bias. In addition, there could be a non-response bias, as individuals who chose not to participate might differ systematically from those who participated. Lastly, some participants may misinterpret certain questions, which may lead to inaccuracies in the data collected. Despite these limitations, the research provides valuable insights into the knowledge, attitudes, and practices of medical professionals and students in Saudi Arabia toward genetic counseling for hereditary blood disorders, which can improve future research and public health campaigns.

Conclusions:

In conclusion, the findings highlight critical deficiencies in the awareness and accessibility of genetic counseling services for hereditary blood disorders within Saudi Arabia's healthcare system. Despite strong support from medical professionals for genetic counseling, significant barriers such as inadequate training, cultural and religious factors, regional disparities in resources, and ethical considerations impede its widespread implementation. To overcome these challenges, a comprehensive approach is essential. Integrating genetics education into medical school curricula and offering continuous professional development opportunities for healthcare providers would enhance their knowledge and competence in genetic counseling. Additionally, national public health campaigns aimed at reducing stigma and increasing genetic literacy could play a pivotal role in improving public understanding of hereditary disorders. Expanding access to genetic counseling services, particularly in rural and

underserved areas, through telemedicine or mobile health clinics could help bridge the gap in service availability.

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