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Exploring Pakistani Doctors' Perspectives on Genetic Counseling: Challenges and Opportunities

Sidra Sajid[1], Fiza Laraib[2], Areesha Rashid*[3], Asma Arshad[4], Naeem Ahmad[5]

Areeba Rashid[6]

[1]M Phil, Department of Zoology (Human Genetics), Quaid-i-Azam University, Islamabad, 45320, Pakistan, Email: sidrasajid@bs.qau.edu.pk

[2]M Phil, Department of Zoology (Molecular Biology), Quaid-i-Azam University, Islamabad, 45320, Pakistan, Email: fizalaraib94@gmail.com

[3]M Phil, Department of Zoology (Human Genetics), Quaid-i-Azam University, Islamabad, 45320, Pakistan, ORCID: https://orcid.org/0009-0006-0401-2174,
E-Mail: areesha.rashid@bs.qau.edu.pk

[4]M Phil, Department of Zoology (Molecular Biology), Quaid-i-Azam University, Islamabad, 45320, Pakistan, Email: asmaashraf@gcuf.edu.pk

[5]PhD Scholars, Human Molecular Genetics Laboratory, Health Biotechnology Division, National Institute for Biotechnology and Genetic Engineering College (NIBGE-C), Pakistan Institute of Engineering and Applied Sciences (PIEAS), Islamabad 44000, Pakistan,
Email: naemahmd1@gmail.com

[6]MBBS Scholar (Final year) Health Department, D.G. Khan Medical College, Dera Ghazi Khan, 32200, Punjab, Pakistan, E-Mail: areebar@dgkmc.edu.pk

Corresponding Author: Areesha Rashid, M Phil, Department of Zoology (Human Genetics), Quaid-i-Azam University, Islamabad, 45320, Pakistan, ORCID: https://orcid.org/0009-0006-0401-2174, Email: areesha.rashid@bs.qau.edu.pk

ABSTRACT

Background: Genetic disorders pose a significant health burden globally due to high consanguinity rates and limited access to Genetic Counseling (GC) services.

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Objective: This study evaluates the acquaintance and perceived constraints of GC in Pakistani doctors. Moreover, it explores Pakistani doctors' familiarity, referral practices, perceived barriers and their possible solutions to enhance GC practice in Pakistan.

Methods: A cross-sectional study was conducted using an online questionnaire to collect quantitative data, which was analyzed through descriptive statistics and thematic analysis.

Results: Using a cross-sectional design, data from 52 doctors revealed that 41% of females were "very familiar" with GC, compared to 23.1% of males. The results indicate that all male doctors (100%) were familiar with GC, whereas 12.8% of female doctors reported a lack of familiarity with GC concept. Referral rates remained low, however, only 20.5% of female doctors and 7.7% of male doctors' frequently referring patients for GC were reported. The key barriers cited were limited patient understanding and superstitious beliefs.

Conclusion: Over 90% of doctors expressed interest in attending GC training and were in support of making it an integral part of healthcare services. In future, this implementation can strengthen patient access to GC, and doctor's approach to informed decision-making about patient's healthcare.

KEYWORDS: Consanguinity, Doctors, Genetic, Genetic Counseling (GC), Pakistan

INTRODUCTION

Genetic disorders result from variations in genes, DNA, or chromosomal material, leading to a range of symptoms associated with specific diseases [1, 2] and come with a wide range of physical, mental, and behavioral phenotypes. An estimated 3.5% to 5.9% of individuals worldwide have one of approximately 7,000 rare or genetic conditions [3]. Pakistan, a Muslim-majority nation in South Asia, is the world's 5th most populous country, with a population exceeding 241.49 million [4] faces a significant burden of genetic and hereditary disorders due to sociocultural, economic, and religious factors. Poor economic conditions in the developing countries prohibits the facility of health and medical needs to control the disease [5]. There is a need to improve healthcare system in developing countries.

A major contributing factor is the high prevalence of consanguinity. Globally, consanguinity is practiced by about 10% of the population, with rates as high as 80.6% in some Middle Eastern provinces and less than 1% in Western societies [6]. Consanguinity is described as the marriage

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or union of individuals with mutual biological ancestors, generally two second cousins, and occurs in more than 73% of Pakistan marriages [7] consanguinity has social advantages such as economic stability and reinforcement of familial bonds, it markedly increases the risk of inherited disorders due to a higher probability of homozygosity for deleterious alleles [8]. This has resulted in a high prevalence of β-thalassemia affecting 5%-7% (9 million carriers in the entire population [9, 10]. Down syndrome is approximately one in every 300 births [11] and microcephaly is 1 in 10,000 newborns [12, 13].

Though unfortunately, many rare genetic disorders are currently untreatable, appropriate Genetic Counseling (GC) can help direct affected individuals toward available therapeutic interventions and empower them to make informed reproductive decisions [1]. GC is the process of helping people to understand and adapt the medical, psychological, and familial implications of the genetic contributions to disease. These services are often utilized and include at-risk family identification, inheritance pattern analysis, and patient direction for disease prevention or treatment [14]. GC has emerged as an important healthcare service worldwide, with programs established in places including the U.S. and countries across Europe since the 1990s [15]. This profession is still nascent in many parts of Asia, such as in Pakistan. GC has been integrated into health systems [15] in some countries, like; Saudi Arabia and India, whereas, Pakistan remains behind the curve with no formal postgraduate training programs for GC [16]. Indeed, World Health Organization (WHO) has acknowledged the scarcity of genetic services in developing countries, such as Pakistan, and claims that 70% of worldwide birth defects could be avoided or treated if clinical genetic services were enhanced in developing nations [10].

GC services are rarely provided in Pakistan by ultrasonologists, obstetricians, or community workers [17]. Two of the main obstacles to widespread capture of these vital data have been the high cost of prenatal testing and a lack of awareness among healthcare providers and patients themselves, even though such testing has been available since 1994. In addition, the limited availability of trained genetic professionals (with only two trained geneticists in Karachi with a population of 15 million) leads to a staggering ratio of 1 geneticist for every 7.5 million people [10]. For every 1 million people, the Royal College of London recommends 3 geneticists and 6–12 genetic counselors [10]. These organizations help to address this gap by offering affordable

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medical genetics services at the tertiary level, both in academic centers and by outreach initiatives [10]. GC services enjoy strong support among Pakistani doctors and professionals as per surveys. For example, 90% of physicians indicated that they would refer patients to GC [18], and 77% of elites agreed that premarital screening for recessive disorders be practiced [13]. GC services are still unavailable on a large scale, and public awareness of repeated consanguinity in generations are absent from rural areas [19].

This paper attempts to fill this gap in knowledge by examining the current understanding of GC and its practice in Pakistan. These findings provide understanding into physician awareness of GC. The results of this study will provide insights into the determinants of access to GC in Pakistan which can be utilized to enhance genetic counselling services.

METHODOLOGY

This study employed a descriptive cross-sectional design approach to explore Pakistani doctors' perspectives on GC. An online questionnaire was used to collect the data to make it convenient for participants and maximize geographic coverage across Pakistan. A snowball sampling method was used. A survey-link was created using Google Forms which was initially shared with family and professional connections who work as doctors. Participants were encouraged to share the link to people in their network and thus the sample grew organically. A total of 52 doctors participated in this study, all of whom were recruited anonymously.

The purpose of study was given in Google form to the participants to aid in the informed consent process. Data were collected through an online questionnaire with a range of closed and open-ended questions that enabled the extraction of quantitative and qualitative statistics. Some demographic factors like; gender, age and specialties were collected. No identifiable data about the participants were asked to preserve anonymity. It also focused on doctors' understanding of GC and their views on how important it is. Moreover, doctors' knowledge of genetic concepts and the barriers they faced in delivering genetic information were assessed. Common misconceptions among patients were established alongside the doctors' willingness to work with genetic counselors. Also, there were questions on the need for training and the integration of GC into Pakistan's healthcare system. Descriptive statistics were used to analyze the data alongside

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thematic analysis. The quantitative data, relating to demographic characteristics and levels of awareness, were summarized through frequency distributions and percentage distributions.

RESULTS

Data reveals that in gender comparison 75% of the responses were from the female doctors and 25% were from male doctors. Table 1 shows the detailed analysis of GC gender-wise.

Table 1 Gender-wise analysis of GC

Category	Female; Frequency (%)	Male; Frequency (%)
Familiarity with the concept of GC		
Not familiar	5 (12.8%)	0 (0%)
Somewhat familiar	18 (46.2%)	10 (76.9%)
Very Familiar	16 (41.0%)	3 (23.1%)
P value	0.19	
Refer patients for GC		
Frequently	8 (20.5%)	1 (7.7%)
Never	4 (10.3%)	2 (15.4%)
Occasionally	18 (46.2%)	7 (53.8%)
Rarely	8 (20.5%)	3 (23.1%)
No response	1 (2.6%)	0 (0%)
P value	0.80	
Level of comfort discussing genetic concepts		
Somewhat comfortable	18 (46.2%)	6 (46.2%)
Very comfortable	20 (51.3%)	7 (53.8%)
No response	1 (2.6%)	0 (0%)
P value	0.49	
Information about common genetic conditions		
Not informed	16 (41.0%)	3 (23.1%)

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Informed	22 (56.4%)	10 (76.9%)
No response	1 (2.6%)	0 (0%)
P value	1	
Collaboration with a genetic counselor		
No collaborations	32 (82.1%)	12 (92.3%)
Have collaborations	7 (17.9%)	1 (7.7%)
P value	0.66	
Need to have collaborations with genetic counselors		
No need to have collaborations	2 (5.1%)	0 (0%)
Need to have collaborations	37 (94.9%)	13 (100%)
P value	1	
Interest in attending workshops or training sessions on GC		
No interest	2 (5.1%)	1 (7.7%)
Interested	37 (94.9%)	12 (92.3%)
P value	1	
Need of integration of GC services into routine healthcare		
Agree	14 (35.9%)	8 (61.5%)
Strongly agree	25 (64.1%)	5 (38.5%)
P value	0.12	

The study highlighted important trends in healthcare providers' familiarity, practices, and attitudes toward GC. A higher percentage of female respondents (41.0%) reported being very familiar with GC compared to male respondents (23.1%), while more males (76.9%) indicated being somewhat familiar. Notably, 12.8% of females reported no familiarity with GC, whereas none of the male respondents reported this lack of familiarity.

In terms of referral practices, females were more likely to frequently refer patients for GC (20.5%) compared to males (7.7%), while males were more likely to refer patients occasionally (53.8% vs.

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46.2% for females). Both genders expressed high levels of comfort in discussing genetic concepts, with over 50% of females (51.3%) and males (53.8%) reporting being very comfortable.

Awareness of common genetic conditions was higher among males (76.9%) than females (56.4%), though a significant proportion of females (41.0%) reported being uninformed. Despite the majority of respondents reporting no collaborations with genetic counselors (82.1% of females and 92.3% of males), nearly all expressed a strong need for such collaborations (94.9% of females and 100% of males). Additionally, the majority of respondents showed strong interest in attending GC workshops or training sessions (94.9% of females and 92.3% of males).

Finally, a significant proportion of females (64.1%) strongly agreed on the need to integrate GC services into routine healthcare, compared to 38.5% of males, who were more likely to simply agree (61.5%). These findings underscore a strong interest in GC education and integration, particularly among female healthcare providers. As all p-values in our study were statistically non-significant ($p > 0.05$ threshold value), it suggests that there is no statistically meaningful differences in doctors' perspectives on GC.

Table 2, shows the participants that were allowed to choose more than one answer, so the percentages reflect how many people selected each option out of the total number of respondents. Hence each question reflects the selection out of 100 percent.

Table 2: Various factors related to Genetic counseling along selected percentages (N = 51)

Factors	**Percentage**
Role of genetic counselling in healthcare	
Providing information about genetic conditions	35.30%
Assessing risk factors and family history	58.80%
Assisting patients in making informed decisions about genetics	60.80%
Providing emotional support and counseling	19.60%
Factors that influence decisions to refer patients for GC	
Family history of genetic conditions	84.30%
Abnormal test results indicating genetic risk	51%

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Patient's request for genetic information	35.30%
Presence of complex medical or family history	43.10%
Family history and abnormal results	2%
Challenges doctors face during the discussion of genetic information	
Limited patient understanding of genetic concepts	88.20%
Emotional reactions from patients	52.90%
Time constraints during consultations	23.50%
Superstitious beliefs about genetic disorders	60.80%
Misconceptions or myths related to Genetics	
Genetic testing can predict all diseases and health outcomes accurately	38.50%
Only individuals with a family history of genetic disorders need genetic counselling	55.80%
Genetic counselling is only for couples planning to have children	42.30%
Genetic conditions are always severe and untreatable.	40.40%
Genetic counselling can't change someone's fate	51.90%
Steps that can be taken to enhance the availability and accessibility of GC	
Spreading awareness about the genetic concepts	88.50%
Introduce virtual counselling for patients in remote areas	44.20%
Increase the number of trained genetic counselors in healthcare facilities	67.30%
Incorporate genetic counselling services into routine healthcare appointments	61.50%

The study highlights several key aspects of GC from the perspective of healthcare providers. This research reveals that primary roles of GC were identified as assisting patients in making informed decisions (60.8%) and assessing risk factors and family history (58.8%), while providing emotional support and counselling (19.6%) was considered less emphasized. When it comes to

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referring patients for GC, family history of genetic conditions (84.3%) was the most influential factor, followed by abnormal test results indicating genetic risk (51%) and the presence of complex medical or family history (43.1%). In contrast, patient requests for genetic information (35.3%) and the combination of family history and abnormal results (2%) played a smaller role in referral decisions.

Data reflects that Doctors faced significant challenges in discussing genetic information, with limited patient understanding of genetic concepts (88.2%) being the most prominent barrier. Other challenges included superstitious beliefs about genetic disorders (60.8%), emotional reactions from patients (52.9%), and time constraints during consultations (23.5%). Misconceptions about genetics were also prevalent, with the most common being that only individuals with a family history of genetic disorders need GC (55.8%). Other myths included the belief that GC can't change someone's fate (51.9%), genetic testing can predict all diseases and health outcomes accurately (38.5%), and GC is only for couples planning to have children (42.3%). Additionally, some believed that genetic conditions are always severe and untreatable (40.4%).

As for the concern to enhance the availability and accessibility of GC, spreading awareness about genetic concepts (88.5%) was identified as the most critical step. Other important measures included increasing the number of trained genetic counselors in healthcare facilities (67.3%), incorporating GC into routine healthcare appointments (61.5%), and introducing virtual counselling for patients in remote areas (44.2%). These findings underscore the need for greater education, awareness, and integration of GC into healthcare systems to address existing challenges and misconceptions.

DISCUSSION

The study provides important views and findings regarding the opinions and practices of Pakistani doctors related to GC with several implications for research and practical purposes. The study revealed that while a significant proportion of doctors, particularly female respondents, reported being "very familiar" with GC, a notable percentage of females reported no familiarity. Such findings reveal a gap of knowledge, especially given the high frequency of genetic disorders due to consanguinity in the country. The higher familiarity among female doctors could be explained by the fact that they are concerned with subjects like obstetrics wherein GC is dealt with more

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regularly. Still, the lack of overall familiarity points toward the specific need for further training. This finding is consistent with findings from previous studies in Pakistan, which have pointed out the fact that there has been no formal GC training for healthcare providers [16]. The gender discrepancy regarding exposure seems rather extraordinary and needs further reflection.

A greater proportion of female doctors indicated they would "often" refer patients for GC as opposed to males. However, most made referrals "from time to time," indicating that referral in Pakistan is not yet a routine procedure for patients. This shows that the provision of genetic services is in a rather undeveloped state, with very few trained counselors available. The low referral frequency has been reported consistently across the world in other low-resource settings [10, 15].

According to the physicians, barriers such as low patients' knowledge and religious beliefs were frequently reported obstacles. These obstacles slow the progressive GC, and they show the cultural and educational gaps that have to be bridged. These challenges are congruent with the findings of other studies from Pakistan and similar cultural settings, all of which strongly highlight the need for cultural sensitivity in GC [17, 19]. For instance, Ullah found cultural beliefs and knowledge deficiency to be the two main barriers to the acceptance of GC in Pakistan, and they recommended awareness campaigns and culturally sensitive educational programs to overcome the specific barriers [19].

A strong interest among physicians in GC workshops highlights the need to incorporate GC training into standard healthcare programs, promoting greater awareness and practical implementation in clinical settings. This suggests the potential high interest in GC knowledge and skills that could help to overcome the high burden of genetic disorders in Pakistan which is consistent with previous studies advocating for widespread improvement in GC education and training [16, 18].

LIMITATIONS

The snowballing technique utilized in this study invites selection bias, with participants employed through professional networks, thus potentially limiting the generalizability of the findings. This small sample size makes it even harder to generalize. Furthermore, since the data were obtained using self-report methods, response bias may have occurred, meaning the respondents might have

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overestimated their familiarity or comfort with GC. The study was further restricted by the absence of a centralized data repository of doctors practicing in Pakistan, thereby making it impossible to sample doctors in a representative manner.

CONCLUSION

In conclusion, importance of GC integration in healthcare can't be denied. It is a process of helping patients and their family to understand the genetic basis of the conditions they are suffering from, the potential risk factors associated with the disease and any available preventive measures. This study shows how doctors view GC and according to them, what measures should be taken to incorporate GC in healthcare system. It's been noted that the majority of the participants highlighted that currently there is no opportunity provided by the healthcare system to collaborate with genetic counselors, but at the same time the majority of participants showed their willingness to collaborate with the genetic counselors if they are given the opportunity. Moreover, the majority of the doctors were eager to increase their knowledge regarding genetic disorders and GC to better help their patients. Despite their willingness to work for the improvement of GC services, the availability of correct and timely opportunities is largely limited. In future this concern of medical professionals should be addressed.

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Data Availability: The data regarding this study is available from the corresponding author, Areesha Rashid, upon rational request.

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Conflicts of Interest: The authors declare that there is no conflict of interest.

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