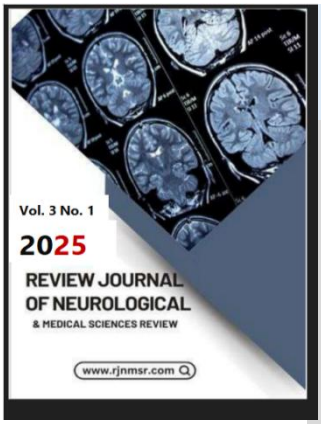

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Genetic Awareness and Genetic Testing in Pakistani Population

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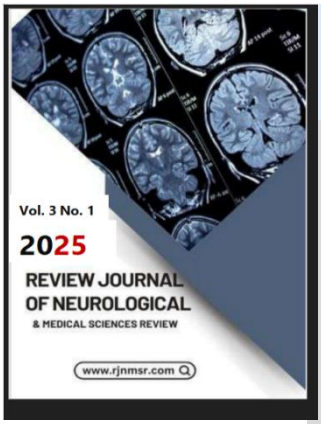
Abstract

Genetic disorders are an emerging public health issue in Pakistan, further aggravated by one of the highest rates of consanguineous unions in the world (~70%), resulting in a higher incidence of autosomal recessive disorders like beta-thalassemia, metabolic disorders, and mental retardation. However, the nation is seriously deficient in genetic healthcare providers, with just two qualified geneticists catering to a population of over 220 million. The current research seeks to assess public knowledge and attitudes toward genetic testing and inherited disorders in Pakistan using a descriptive cross-sectional study. 109 participants were surveyed, finding that 78.89% knew about genetic testing, while 87.15% reported willingness to participate in genetic testing if it is available. Additionally, 87.15% agreed on implementing pre-marital genetic testing for the prevention of inherited diseases. These results highlight a strong public interest in genetic services, even in the face of an underdeveloped genetic healthcare infrastructure in the country. The research emphasizes the imperative for policy formulation and the development of integrated genetic counseling and testing services, as well as investment in the education of genetic professionals. This paper offers practical recommendations on the gaps in genetic healthcare and proposes ways to address these gaps to reduce the burden of genetic disorders in Pakistan.

Keywords: Genetic diseases, consanguineous marriages, beta-thalassemia, metabolic disorders, mental retardation, genetic testing, genetic counseling, premarital genetic screening, public education, inherited illnesses, Pakistan, clinical genetics.



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Introduction

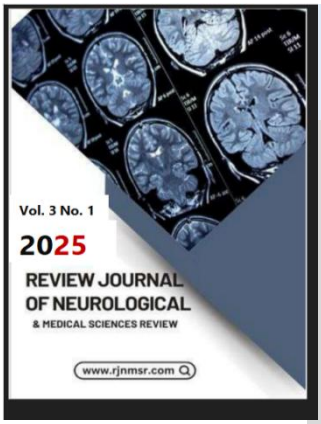
Genetic diseases pose a growing health issue in Pakistan, a country that has one of the highest rates of consanguinity in the world (~70%).(Bener and Mohammad 2017) This high rate is the main reason for the high frequency of autosomal recessive disorders, such as beta-thalassemia, metabolic diseases, congenital abnormalities, and mental retardation.(Samir 2024) In contrast to the documented genetic disease burden, Pakistan's health infrastructure is lamentably underdeveloped for clinical genetics facilities, trained staff, and awareness. (SHARIF 2020)With about 5-7% of the population being carriers of beta-thalassemia, equating to roughly 9 million individuals, and given recent reports revealing a diagnostic yield as high as 61% in consanguineous pedigrees, it is apparent that genetic testing and counseling are an urgent need.(Fareed and Afzal 2017)

But Pakistan's ability to meet this demand is grossly constrained. There are presently only two formally trained geneticists to cater to a population of more than 220 million, creating a critical shortage of genetic professionals.(Krutish 2019) The Royal College of Physicians (UK) estimates that Pakistan would need at least 660 geneticists and 1320-2460 genetic counselors to cover the needs of the country. This gross imbalance aggravates the health inequalities and puts additional pressure on the already strained healthcare system.(Sanders 2023)

In spite of these difficulties, there is a developing interest in meeting the country's genetic health needs.(Chung, Project et al. 2022) The Pakistani Society of Medical Genetics and Genomics (PSMG), in response to the rising number of genetic disorders, has launched efforts to enhance public access to genetic services.(Christianson and Modell 2004) An effort of this nature was a nationwide webinar and needs assessment that had strong support expressed by both health professionals and the general public to introduce formal training in genetic counseling education (91.6%) and increased accessibility of genetic testing (100%). These findings speak to a dramatic gap in accessing genetic services but also show general public interest across the board for building these fundamental services.(Mehlman and Botkin 1998)

Furthermore, while India and Iran in neighboring countries have taken positive steps towards enacting national genetic policies, Pakistan has not yet formulated a unified national approach to deal with its genetic health requirements.(Jiwani 2023) Consequently, there is still an urgent necessity to not only enhance the public's knowledge about genetic disorders and the need for genetic testing but also to institute a complete, affordable, and ethically based system for genetic counseling and clinical services.(Khan 2024)

The purpose of this paper is to study the present scenario of genetic services in Pakistan, assess awareness and attitudes of the general public as well as healthcare professionals, and identify prime areas for enhancing genetic healthcare provisions.(Uzair, Fatima et al. 2024) Through the identification of gaps in education, policy, and service delivery, this paper will offer insights that can be used to shape the development of a strong genetic services framework within Pakistan, leading to



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genetic healthcare becoming an important part of the country's public health system.(Hosen, Anwar et al. 2021)

Materials and Methods

Study Design and Setting

The descriptive cross-sectional study was carried out between the months of February to May 2025 for a period of four months to evaluate public perceptions and awareness about genetic testing and inherited diseases in Pakistan.

Study Population and Sampling Technique

Recruitment employed a non-probability convenience sampling method. Eligible participants were Pakistani residents aged 18 years or more, regardless of gender, education, or socioeconomic status. Based on informed consent, individuals who refused or could not complete the questionnaire were excluded. The sample size of 109 participants was based on a feasibility estimate and previous studies with comparable aims that had yielded high awareness levels (>70%)—facilitating meaningful descriptive analysis within available resources.

Instrument Design and Validation

Data were gathered through a structured, self-completed questionnaire with close-ended questions across the following areas: (1) Demographics, (2) Knowledge of genetic testing, and (3) Attitudes and perceptions towards genetic testing. The questionnaire was derived from previously validated instruments employed in comparable regional research and checked for content and face validity by three subject-matter experts (clinical geneticist, bioethicist, and epidemiologist). Internal consistency was determined by piloting with 20 participants and achieving a Cronbach's alpha of 0.82, which is very good reliability.

Data Collection Procedure

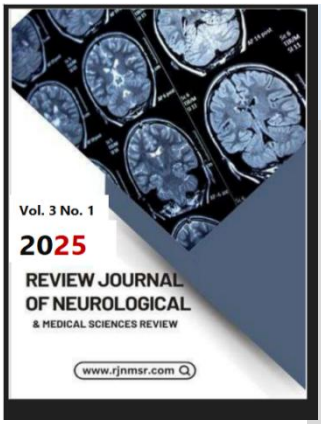
The survey was given both online using Google Forms and in paper form to reach more participants, particularly from regions with poor internet connectivity. All the respondents were supplied with an information sheet for the purpose of the research, confidentiality guarantees, and voluntary participation before they participated. Electronic or written informed consent was given.

Ethical Considerations

Ethical guidelines like voluntary participation, anonymity, and confidentiality of data were strictly observed. Participants were never requested to give names or identifiers. As the study was minimal risk and for research purposes alone, formal ethical permission was not sought but all procedures still conformed to ethical principles set out in the Declaration of Helsinki (2013).

Data Analysis

Data were entered and processed using Microsoft Excel 2019. Descriptive statistics were used to present the frequency and percentage of each response item for awareness and perception. Tables were presented to show the distribution of the responses of the participants.



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Results

Descriptive statistical analysis was carried out to evaluate the extent of public awareness and perception about genetic testing and inherited conditions among the research participants (n = 109).

Among the total subjects, 30.29% (n = 33) said they had a family history of inherited disorders and 69.72% (n = 76) said they did not.

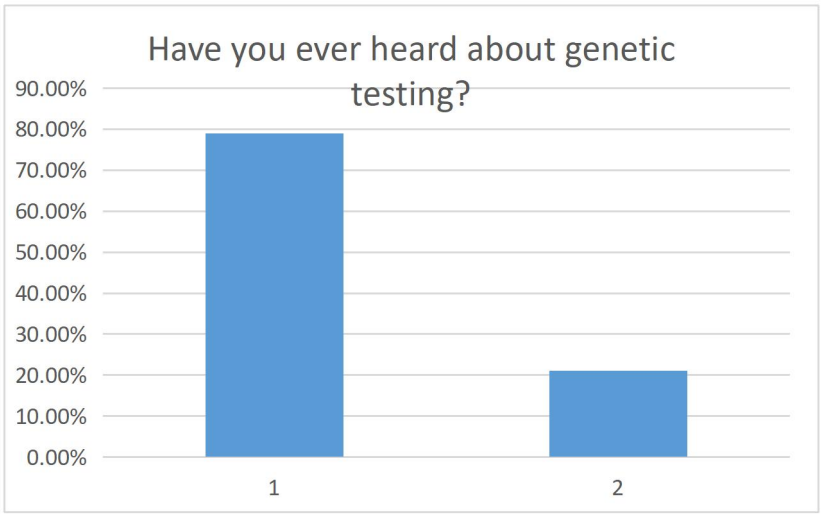


Fig. 4.1. Family History of Inherited Diseases Among Participants

Knowledge regarding genetic testing was comparatively good, with 78.89% (n = 86) reporting having heard of it, and 21.10% (n = 23) saying they had not.

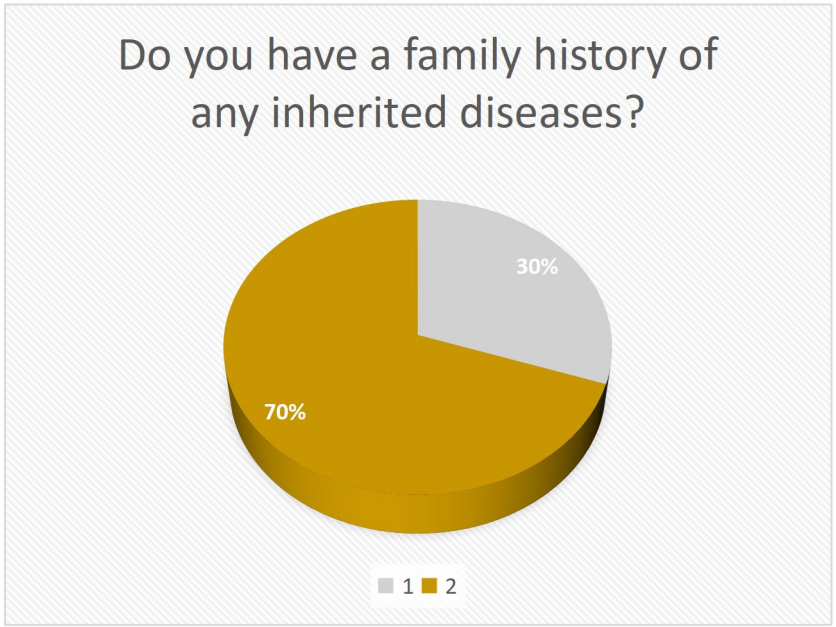
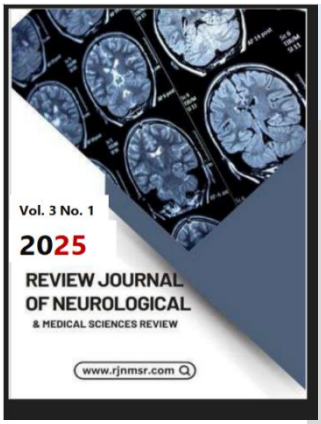


Fig. 4.2. Awareness of Genetic Testing Among Participants



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When questioned if parents can transmit genetic diseases to offspring, a large majority of 87.15% (n = 95) said "Yes." However, 12.84% were uncertain, and no one answered "No" to this question, suggesting a general understanding of heredity.

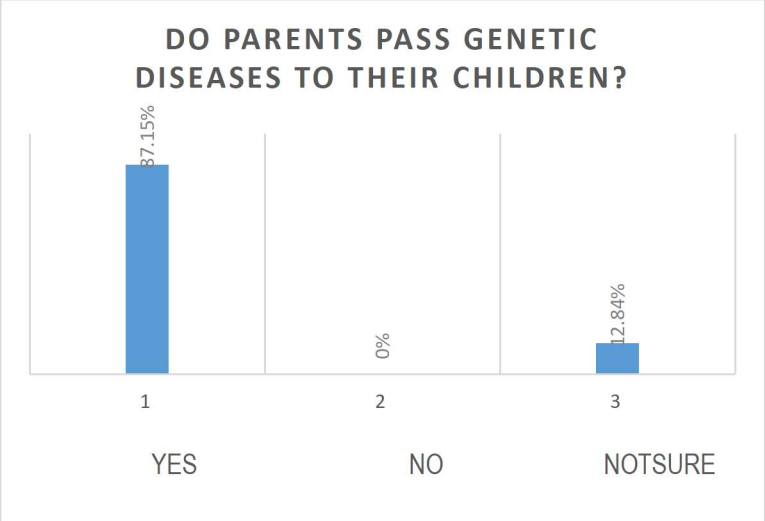

Fig. 4.3. Parental Transmission of Genetic Disease

As for the preventive value of genetic testing, 87.15% (n = 91) thought that it can prevent disease. But again, 16.51% were uncertain, and none disagreed in so many words.

On the willingness to undergo genetic testing if provided in Pakistan, 87.15% (n = 77) out of the 108 participants replied in the affirmative, 8.33% (n = 9) replied in the negative, and 20.37% remained uncertain. It shows a very high interest in genetic services with some degree of uncertainty among the population.

A large majority of respondents favored the concept of pre-marital genetic testing, with 98 of 109 (87.15%) holding the view that couples should undergo testing to prevent inherited diseases. 3.66% (n = 4) of respondents disagreed, and 6.42% were uncertain.

Lastly, 96 participants (87.15%) were interested in knowing more about genetic testing, whereas 10.28% (n = 11) were not interested and 6.42% were uncertain.

These findings indicate that even though a significant number of people have no family history of genetic disorders, there is high awareness and interest in genetic testing, especially for its preventive and educational significance. The consistently high proportion of positive answers to all questions indicates a favorable attitude towards the incorporation of genetic testing into public health services in Pakistan.

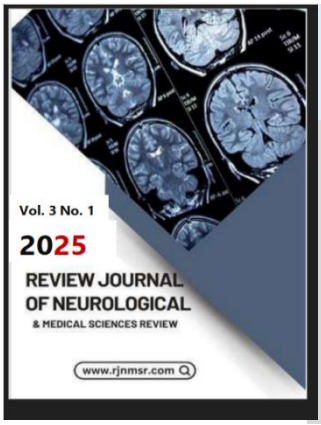

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Table 1: Descriptive Statistics for Awareness and Perception of Genetic Testing

Question	Yes	No	Total	%Yes	%No / %Not sure
Do you have a family history of any inherited diseases?	33	76	109	30.29%	69.72%
Have you ever heard about genetic testing?	86	23	109	78.89%	21.10%
Do parents pass genetic diseases to their children?	95	0	109	87.15%	12.84% (Not sure)
Can genetic testing help in preventing diseases?	91	0	109	87.15%	16.51% (Not sure)
Would you get a genetic test if it was available in Pakistan?	77	9	108	87.15%	8.33% / 20.37% (Not sure)
Should couples get genetic testing before marriage to avoid inherited diseases?	98	4	109	87.15%	3.66% / 6.42% (Not sure)
Would you like to learn more about genetic testing?	96	11	107	87.15%	10.28% / 6.42% (Not sure)

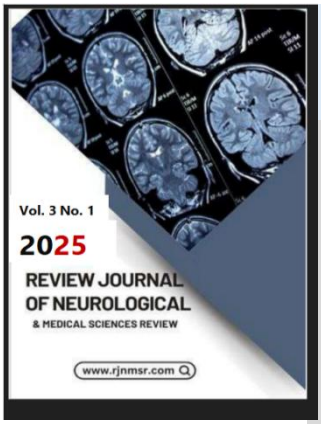

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Table 1: Descriptive statistics of responses to questions pertaining to awareness and perception of genetic testing among the survey population. The data suggests high awareness about genetic testing and good support for premarital genetic testing.

Discussion

The research sought to analyze public perception and awareness of genetic testing and hereditary diseases in the Pakistani populace.(Chin and Tham 2020) The research indicated that the majority of respondents (78.89%) had knowledge of genetic testing, while a still higher percentage (87.15%) indicated willingness to be tested should it be introduced in Pakistan. These findings concur with the research of Siddiqui et al. (2021), where there was reported high public interest in genetic counseling and testing services as per a national needs assessment by the Pakistani Society of Medical Genetics and Genomics (PSMG), in which 91.6% endorsed formal education in genetic counseling and 100% supported enhanced access to genetic testing.(Ashfaq, Ahmed et al. 2023)

In addition, 87.15% of our study's participants acknowledged the inherited character of genetic disorders, and the same percentage held genetic testing to have preventative potential.(Al Eissa, Almsned et al. 2024) This supports increased public awareness about genetics and health implications, as concluded by Ullah et al. (2021), and which pointed to the burden caused by genetic disorders in Pakistan by virtue of consanguinity being high and pointed out the paramount need for better genetic services and trained personnel.

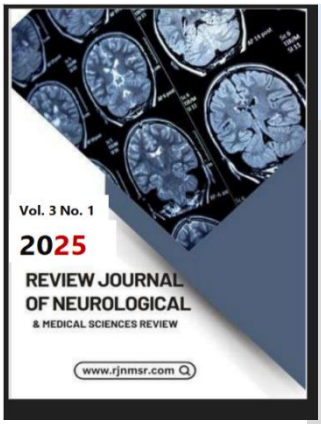
Notably, even with the nation's woefully under-equipped clinical genetics facility with only two formally trained geneticists to serve more than 220 million citizens public interest in learning more about genetic testing continues to be strong (87.15%). This is an essential opportunity for policymakers and healthcare planners to tap this public interest and work towards developing a sustainable infrastructure for genetics education, counseling, and testing.(Parthasarathy 2012)

The high level of support for premarital genetic testing (87.15%) seen in our sample indicates a positive approach among the population to prevent inherited disorders.(Al-Shroby, Sulimani et al. 2021) As Siddiqui et al. (2021) also pointed out, there is growing advocacy among healthcare professionals for the inclusion of genetic testing in national health policies, particularly in high-risk consanguineous populations. The lack of a national policy in Pakistan, in contrast to India and Iran, restricts the enforcement of mass preventive genetic measures.(Riaz, Tiller et al. 2019)

While our research utilized convenience sampling and was constrained by a relatively small sample size, the consistent patterns in responses indicate an underlying trend of positive attitude towards genetic services.(Birmingham, Agarwal et al. 2013) The results therefore support the contention of Ullah et al. (2021) that public health interventions for genetic awareness and capacity-building can be extremely effective in nations such as Pakistan.(Sarkar and Jha 2025)

Conclusion

The findings of this research demonstrate a high degree of awareness and a positive sentiment towards genetic testing among the population of Pakistan, even though



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there is a critical shortage of genetic healthcare facilities and trained personnel within the country. With almost 80% of the respondents having heard about genetic testing and well over 87% willing to undergo it and advocate premarital screening, it is clear that there exists a demand and a need for affordable and quality genetic services.

These findings highlight the importance of incorporating genetic testing and counseling into Pakistan's national health agenda. Policymakers need to capitalize on increased public interest and create formal genetic services, fund training clinical geneticists and counselors, and institute targeted public information campaigns. Consultation with professional organizations such as the PSMG and adoption of regional experience in India and Iran can reinforce this initiative even further.

Finally, by filling these gaps, Pakistan can progress toward a more preventive and equitable model of healthcare that includes genetic knowledge to minimize the load of hereditary diseases.

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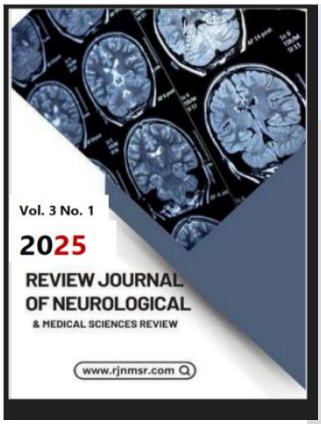

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