

Knowledge About Genetics in Congenital Hearing Loss: A Survey Among Audiologists

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J Health Allied Sci^{NU}

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Abstract

Introduction According to the World Health Organization, there are more than 63 million people in India who have disabling hearing loss. Hearing loss at birth can be caused due to genetic factors. Referral for genetic testing and counseling can be directly influenced by the knowledge among the audiologists. Specific guidelines and recommendations have been provided by experts in the field for competency in genetics among professionals to ensure early and appropriate management in young children with hearing loss. In India, there is limited research done on assessing audiologists' knowledge concerning the importance of genetic testing and counseling. Thus, the present study aimed to explore the knowledge about the role of genetics in congenital hearing loss among audiologists.

Method The study was conducted in two phases. Phase I included the development and validation of the questionnaire. Phase II included the administration of the questionnaire to the professionals from the field of audiology and speech-language pathology through Google Forms.

Results The study findings revealed that the knowledge level about general genetics was adequate; however, aspects related to genetic testing and syndromic and non-syndromic congenital hearing loss were moderate. There was a statistically significant difference observed in the scores obtained by professionals who had completed Master of Science in Audiology as compared to those who had completed Bachelors in Audiology and Speech Language Pathology and Masters in Audiology and Speech Language Pathology.

Conclusion The study has highlighted the current knowledge of audiologists with respect to domains of genetic testing and syndromic and nonsyndromic congenital hearing loss. There is a need to educate audiologists working in the area of early identification and intervention about the genetic basis of hearing loss.

Keywords

- ▶ audiologists
- ▶ genetics
- ▶ syndromic and nonsyndromic hearing loss

Introduction

Globally, hearing loss is the second most commonly occurring sensory disability.¹ According to the World Health Organization, by the year 2050 one in four persons will have some degree of hearing loss.¹ Prelingual hearing loss

is a serious concern because it affects the development of spoken language and may interfere with the development of social and cognitive skills. Around 50 to 60% of all hearing losses are now recognized as having a genetic component.² Congenital hearing loss can be hereditary or caused by

EPub Ahead of Print: 29 November 2024 **DOI** <https://doi.org/10.1055/s-0044-1793837>.
Published: 23 April 2025 **ISSN** 2582-4287.

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Thieme Medical and Scientific Publishers Pvt. Ltd., A-12, 2nd Floor, Sector 2, Noida-201301 UP, India

This article was initially published as Online First by Thieme Medical & Scientific Publishers Pvt. Ltd., and later included in the Issue compiled by Scientific Scholar.

diseases like rubella or the cytomegalovirus that are received during pregnancy. It is more common in infants who are in the neonatal intensive care unit and it can be a standalone disorder or a sign of a syndrome with other symptoms. Genetic factors alone account for at least 40% of all the cases of congenital hearing loss.³ Also, 20 to 30% of congenital hearing loss cases are syndromic that can occur together with structural or functional anomalies of other organs and 70 to 80% are nonsyndromic.⁴ The extraordinary discoveries in the field of molecular genetics during the last three decades have contributed substantially to the current knowledge about genetic hearing loss and have increased the diagnostic rate, enabling the detection of novel variants in deafness-related genes.³ There are over 120 nonsyndromic hearing loss genes identified to date, and many additional loci have been identified that are associated with hearing loss.⁵ Genetic evaluation and testing are becoming increasingly accepted as a critical process in understanding the diagnosis and treatment of hearing loss. One role of audiologist is to identify those patients who would benefit from referral to a genetic counselor.⁶ The objectives of the early hearing detection and intervention programs are to use audiological procedures to screen all infants for hearing before 1 month of age, to identify hearing loss by 3 months of age, and to coordinate the necessary intervention services by 6 months of age.⁷

Professional societies, advocacy organizations, and government agencies have published recommendations for competency in genetics for audiologists and other health care professionals to help individuals with communication disorders with regards to the management.^{7,8} As genetics is becoming a more clinically relevant topic, audiologists must obtain an in-depth understanding of the process of genetic counseling and evaluation, its benefits and limitations, and the number of genetic resources available to deaf children, their families, and professionals.⁹ If the audiologist is unaware of hearing genetics, a chance for referral to a genetic counselor at the time when patients can benefit from this crucial information may be lost. They must also go over the advantages of undergoing a genetic test, as some parents may need extra encouragement and information. Therefore, a solid grasp of hearing genetics is required for audiologists working in the area of early identification and intervention of hearing loss. The Joint Committee on Infant Hearing¹⁰ has suggested initiatives to improve audiologists' genetics expertise and knowledge and recommended that all individuals with congenital hearing loss should have a thorough genetic evaluation, with an audiologist functioning as a member of the interdisciplinary team. Furthermore, this information enables the identification of the comorbidities that may need a referral for specialty care, thus allowing early treatment and helping with the identification of potential candidates for cochlear implants.⁴ The Rehabilitation Council of India, which is a statutory body under the Government of India, regulates and standardizes the minimum education and training courses necessary for professionals working with people with disabilities. From the 2018 to 2019 academic year, the Council had initiated the Master of

Science (Audiology) program, which includes genetics of hearing as a subject in the course curriculum.¹¹

Lapham et al¹² surveyed members from a number of health care organizations regarding their provision of genetic services. Out of these, American Speech and Hearing Association members claimed to have discussed the genetic component of disorders with approximately 70.5% of their patients, but they only referred 26.1% of them for genetic counseling. They reported that educational training in genetics is regularly required. In India, few research studies have attempted to explore the knowledge level and application of genetics in hearing loss among practicing audiologists. They concluded that there is a need to update the understanding of genetics related to hearing for these professionals.^{13,14} However, there is limited research done on the audiologists' knowledge concerning the importance of genetics and genetic testing in congenital hearing loss. Thus, the study aimed to explore the knowledge of audiologists about the role of genetics in congenital hearing loss.

Materials and Method

The study design used was cross-sectional and exploratory. Ethical approval was obtained from the Ethical Committee of Bharati Vidyapeeth (Deemed to be University) Medical College (BVDUMC/IEC/188 dated on August 17, 2022). A questionnaire-based survey comprising of 30 questions was developed and content validated by five audiologists and one geneticist. The questions formulated were divided into two domains with 18 questions on general genetics and genetic testing and 12 questions on syndromic and nonsyndromic congenital hearing loss. Each question had two response options "yes" and "no." A score of 1 was given to every correct answer. The maximum obtainable score after filling the questionnaire was 30 and the domain-wise scores were 18 and 12 for domains A and B, respectively. The scores were then self-categorized based on the total score. A score obtained between 0 and 8 was considered as low level of knowledge, score which was greater than 8 but less than 15 was categorized as moderate level of knowledge, score which was greater than 15 but less than 23 was categorized as good level of knowledge, and score greater than 23 and equal to 30 was categorized as excellent level of knowledge.

The finalized questionnaire was created using Google Forms and circulated through email and social media platform.

Participants and Procedure

Participants who had completed postgraduation either in Master of Science–Audiology (M.Sc. Aud) or Masters in Audiology and Speech Language Pathology (M.ASLP) or Bachelors in Audiology and Speech Language Pathology (B.ASLP) were included in this study. Participants who had family members who were geneticist or genetic counselors and participants who had family members with congenital hearing loss were excluded from this study.

The study was conducted in two phases. The first phase of the study consisted of the development and validation of the questionnaire in English language. The second phase of the study included administration of the developed questionnaire on the selected population. The questionnaire was sent to approximately 290 professionals who had B.ASLP or M.ASLP or M.Sc. (Aud) qualification.

Statistical Analysis

The data collected was entered into IBM SPSS software (Statistical Package for Social Sciences) version 23 and further subjected to normality check. Descriptive statistics was applied to explain the scores obtained by the participants. One-way analysis of variance (ANOVA) was used to compare the scores obtained by the participants with different educational qualification.

Results

The questionnaire was circulated across 290 professionals out of which 210 responded (return rate 72.42 %). The test-retest reliability of the finalized questionnaire on 15 participants was determined by calculating the intraclass correlation coefficient (ICC). The ICC value of the finalized questionnaire consisting of 30 questions was 0.919.

As shown in ► **Fig. 1**, the number of females who filled out the questionnaire was more as compared to male participants. Majority of the participants were in the age range of 21 to 30 years with work experience of less than 5 years. With regards to the work place, most of them

had private clinics followed by hospital set-up and academic institutions.

Shapiro–Wilk test for normality was used to check the domain-wise scores and total scores obtained by the three professionals. Scores obtained on domain A for professionals who completed B.ASLP and domain B for professionals who completed M.Sc. (Aud) and M.ASLP were normally distributed ($p > 0.05$), whereas the scores obtained for domain A of M.Sc. (Aud) and M.ASLP and domain B scores of B.ASLP were not normally distributed ($p < 0.05$). The total scores obtained by all the three groups of professionals were normally distributed.

For domain A, which included questions on general genetics and genetic testing, the mean score obtained by the participants who had B.ASLP qualification was 11.78. Eighty-eight percent of the participants scored in the range of more than 50% of the total domain score. The median score obtained by participants with M.ASLP and M.Sc. (Aud) qualification was 12 and 13, respectively. Ninety-two percent of the participants with M.ASLP qualification and 96% of the participants with M.Sc. (Aud) qualification had scored more than 50% of the total score. However, none of the participants achieved a full score in this domain. For domain B that included questions on syndromic and nonsyndromic hearing loss, the median score obtained by the participants who had B.ASLP qualification was 7. Seventy-two percent of the participants scored in the range of more than 50% of the total domain score. The mean score obtained by participants with M.ASLP and M.Sc. (Aud) qualification was 7.06 and 7.88, respectively. Only 62% of the participants with M.ASLP

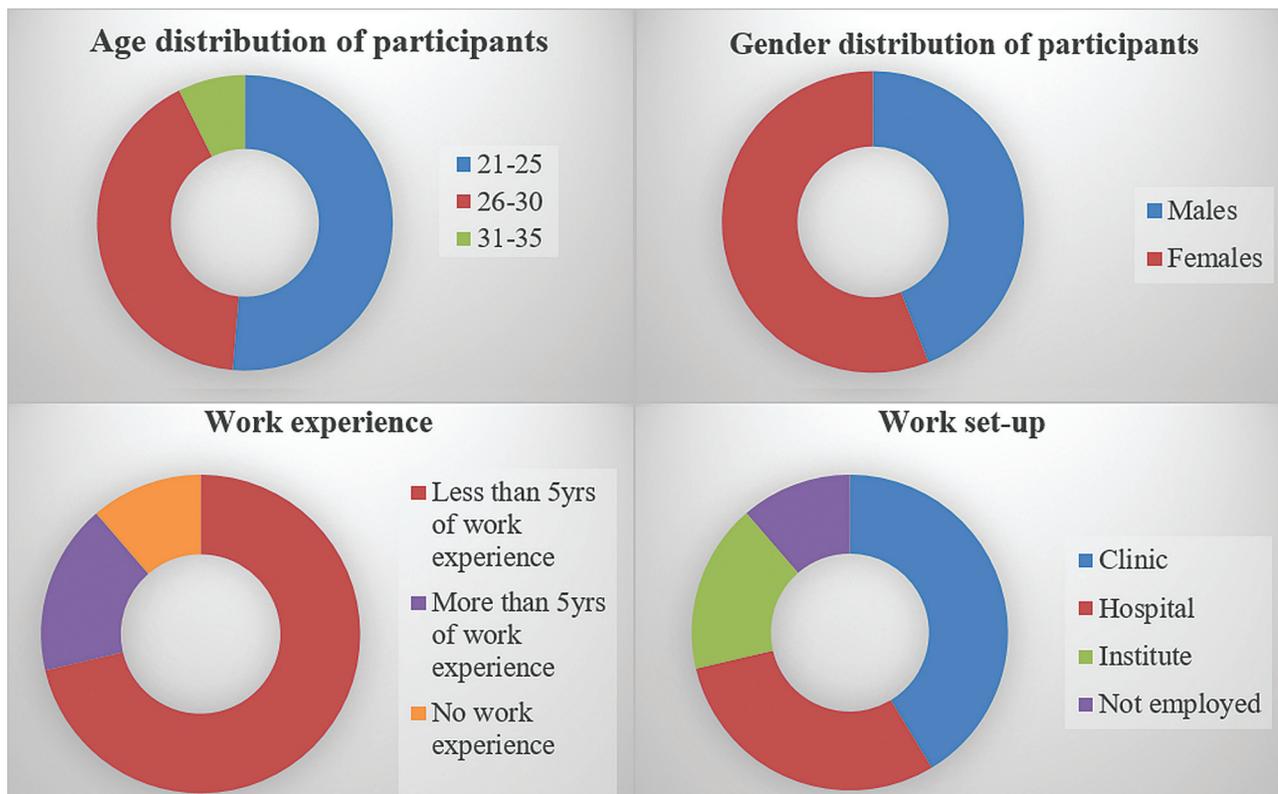


Fig. 1 Graph showing the demographic details of participants ($n = 150$).

qualification and 80% of the participants with M.Sc. (Aud) qualification had scored more than 50% in this domain.

With regards to the answers obtained for each question in domain A, majority of participants with B.ASLP and M.Sc. (Aud) qualification had correct responses to questions related to general genetics, but they scored less on questions related to genetic testing. However, participants with M. ASLP qualification scored less on questions related to genetic testing and counseling.

In domain B, majority of the participants had correctly responded to the questions related to TORCH infection, Usher syndrome, Down syndrome, Pierre Robin syndrome, Treacher–Collins syndrome, associated ear anomalies, and hearing loss. However, questions related to genes responsible for nonsyndromic hearing loss, Jervell and Lange–Nielsen syndrome, and Hunter’s syndrome were answered incorrectly. The detailed results for domains A and B are provided in **–Supplementary Tables S1 and S2**, respectively.

The mean total scores obtained by the participants with B. ASLP, M.Sc. (Aud), and M.ASLP qualification were 19.1, 21.2, and 18.96, respectively. The professionals with M.Sc. (Aud) qualification scored highest among the three groups. Further, a one-way ANOVA was performed to compare the effect of educational qualification on the total scores obtained by the participants. The results revealed that there was a statistically significant difference in the mean total scores between at least two groups of participants ($F(2,147) = [8.46], p = 0.001$). Thus, post hoc Bonferroni analysis done for between-group comparison revealed that the mean scores were significantly different between the M.Sc. (Aud) and B.ASLP group ($p = 0.001$) and M.Sc. (Aud) and M. ASLP group ($p = 0.001$).

Discussion

The current investigation aimed to explore the level of knowledge of audiologists regarding the role of genetics in congenital hearing loss. Appropriate referral for genetic testing and counseling can be directly influenced by the knowledge among the audiologists. The knowledge level with regards to general genetics was good for all the audiologists with different educational qualifications. However, the number of audiologists with M.Sc. (Aud) qualification who provided correct responses was consistently higher than the audiologists with B.ASLP and M.ASLP qualification. A possible explanation for this could be attributed to the inclusion of a separate subject “Genetics of Hearing and Pediatric Audiology” in the third semester of M.Sc. (Aud) coursework by the Rehabilitation Council of India and made effective from the academic year 2018. Whereas the B.ASLP and M.ASLP coursework included general genetics and genetics of hearing loss as a short topic in the course content. Also, as compared to other audiologists, those with M.Sc. (Aud) qualification had recently completed their post-graduate education and had less than 5 years of work experience, which contributed to higher scores. With regards to questions related to the definition of genotype, testing methods used to provide genetic diagnosis like karyotype

analysis, sanger sequencing, and type of samples used in genetic testing, very few audiologists who had B.ASLP, M.Sc. (Aud), or M.ASLP qualification answered correctly. These study findings were in coherence with previous research findings reported by Ravi et al,¹⁵ wherein 70.3% of the responses were correct for the genetic knowledge-based questions and the confidence levels and attitudes of professionals were in the medium categories. However, the present findings do contradict the results observed by Vishnuram et al,¹⁴ where they have reported that audiologists had insufficient knowledge of genetics and practical genetic skills.

Most of the audiologists in the present study agreed with the fact that genetic counseling is desirable for families who had a history of congenital deafness. However, for the question related to genetic counseling, whether it should be done by an audiologist, majority of them who had B.ASLP and M.ASLP qualification responded incorrectly. The present results were in agreement with earlier research studies,^{13,14} where they reported that the audiologists strongly agreed that they could provide genetic counseling to patients. There is dearth of literature exploring the knowledge level regarding these specifications among professionals involved in health care services. Furthermore, majority of the audiologists with different educational qualification scored less than 80% on the aspects of syndromic and nonsyndromic congenital hearing loss, like the previous domain. It was interesting to note that very few audiologists could provide correct answers for questions related to genes responsible for hearing loss and major contributors to the etiology of genetic hearing loss. These results align with the research conducted by Burton et al,¹⁶ reporting that a significant number of professionals had insufficient understanding of the genetics of hearing loss. Similarly, the outcomes are in harmony with the observations of an earlier study where the researchers reported lack of knowledge on medical genetics among nursing students.¹⁷ Thus, the present findings highlight the importance of regular continuing education programs for updating these practicing audiologists regarding advances in genetic testing, significance of referral to geneticists for counseling, and the common genotypes and phenotypes observed in children with congenital hearing loss.

Conclusion

The present study revealed that the overall level of knowledge was good for practicing audiologists with different educational qualification on the aspects of general genetics, whereas there seems to be a need for updating the knowledge in the domain of syndromic and nonsyndromic congenital hearing loss. Practicing audiologists working in the field of early intervention of hearing loss should be provided with continuing education programs focusing on the genetics basis of hearing loss. Future studies with larger sample size focusing on the effect of regular education programs on the knowledge of the practicing professionals can be incorporated.

Authors' Contributions

N.K. contributed to the conceptualization, data curation, formal analysis, investigation, methodology, resources, software, validation, as well as the writing of the original draft and the review and editing of the manuscript. S.D. was involved in the conceptualization, data curation, formal analysis, investigation, methodology, project administration, resources, software, supervision, validation, and also contributed to the writing of the original draft and the review and editing of the manuscript.

Conflict of Interest

None declared.

Acknowledgment

We acknowledge our respected Principal for this opportunity, valuable knowledge, and experience.

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