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Egyptian Journal of Medical Human Genetics (2023) 24:39
https://doi.org/10.1186/s43042-023-00417-x

Egyptian Journal of Medical Human Genetics

RESEARCH

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Knowledge of genetics of hearing and genetic counseling among practicing audiologists

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Abstract

Introduction Hearing loss occurs during various periods of life. Around half of the world's hearing loss is considered to be inherited or genetic. Audiologists play a key role in educating the patient/family about the origin of hearing loss and counseling on genetic aspects of hearing. Genetics has been part of the curriculum in India for the specialised master's degree in audiology since 2009. There is a need to investigate the impact of this course on the field of knowledge and practice of hearing genetics among audiologists. In this study, we examined the knowledge and practice of genetics of hearing among audiologists practicing in India. In addition, we compared graduates who learnt genetics in their curriculum with graduates who did not.

Method A thirty-six-point questionnaire was developed based on clinical and research studies conducted *hitherto* and inputs from practicing geneticists. The survey consists of five domains. 133 audiologists participated in the study.

Results Based on percentage analysis, overall knowledge and practice of the genetics of hearing among the audiologists were not adequate in certain aspects of the genetics of hearing (inheritance pattern and genetic counseling). There were no significant differences in knowledge and practice between audiologists based on educational qualifications and work experience.

Conclusion This novel study compared knowledge of audiologists and practice based on their work experience and educational qualifications. The present study reported that the majority of audiologists were interested in updating their knowledge about the genetics of hearing.

Keywords Audiologist, Genetics of hearing, Knowledge, Practice, Questionnaire

Introduction

Hearing loss is the loss of sensory ability and is one of the most common disabilities worldwide (WHO, 2021). Hearing loss occurs when a person's hearing threshold is ≥ 20 dB. According to the World Health Organisation, by 2050, one in every four people will have some degree of hearing loss. Hearing loss can occur during various stages of life. Nearly 50% of hearing loss is due to environmental factors such as prenatal infections, hyperbilirubinemia, hypoxia, and the use of ototoxic medications. The other 50% of cases are caused by genetic factors involving syndromic and non-syndromic conditions [1]. Non-syndromic conditions account for 70% of overall hearing loss, whereas syndromic conditions contribute to the remaining 30% [1]. Some syndromes have characteristics that make the diagnosis easier. In these cases, it is more probable that the dysmorphic characteristics are

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identified by a doctor before the patient's first session with an audiologist. In less extreme cases, an audiologist may be the first medical professional to identify the presence of a genetic disorder. This is particularly important for disorders without dysmorphic facial characteristics [2, 15]. When a genetic form of hearing loss occurs in isolation, it is referred to as non-syndromic hearing loss. More than 400 genes responsible for hearing loss have been identified [3]. The *GJB2* gene encodes connexin 26 and *GJB6* gene encodes for connexin 30 protein, the mutation of *GJB2* gene is responsible for causing autosomal recessive non-syndromic hearing loss [4]. Audiologists and speech-language pathologists (SLPs) routinely serve patients and clients with genetic etiologies [5]. They are crucial for explaining the reasons for hearing loss and providing family members with counseling [16, 17]. The possibility of recurrence and other potential health effects may be revealed through the diagnosis of hearing loss with a genetic cause. Hence, an audiologist must be able to determine whether a genetic referral is required and be ready to discuss the advantages of genetic testing for patients and their families [2, 16]. Audiologists will be acknowledged by physicians as experts in hearing loss, and they will be entrusted with making important decisions. A missed opportunity for genetic consultation at a time when patients may benefit from important information could occur if the audiologist lacks knowledge of hearing genetics. Explaining the need for genetic testing to patients and their families, as well as a thorough investigation of familial history of hearing loss, are some of the additional roles that audiologists must take on in order to provide holistic care to patients [2]. Once a genetic referral is established, the audiologist must also discuss the benefits of undertaking a genetic test, as some parents may require additional encouragement and information due to their lack of knowledge about the potential consequences of a genetic diagnosis on their child and other family members.

Therefore, a strong understanding of genetics in hearing is necessary for audiologists to perform these roles [2]. The Joint Committee on Infant Hearing (JCIH) has proposed efforts to increase knowledge in genetics and competencies among audiologists. According to JCIH position statement 2000, all individuals with congenital hearing loss should undergo comprehensive genetic evaluation with an audiologist serving as a member of the interdisciplinary team (JCIH, 2000).

In 1964, the first Audiology and Speech-Language Pathology program was started at the twin institutes: BYL Nair Charitable Hospital and T.N. Medical College, Mumbai. They offered a dual degree in audiology and speech-language pathology at master's level. Master's in audiology was approved from the academic session 2018–2019 by the Rehabilitation Council of India. Genetics was included in the course curriculum of the specialised master's degree in audiology (M.Sc. Audiology), in India. There has been no evidence-based research on the influence of this curriculum on knowledge and practice till date or the comparison between the two groups of graduates. An audiologist's expertise in clinical perspectives is just as important for managing hearing aids as their comprehension of the genetic perspective behind the hearing loss. No evidence-based study has been conducted on whether audiologists recognise the importance of genetic counseling. According to a study conducted *hitherto*, out of 31 audiologists in Telangana, most of the audiologists were unaware of genetics of hearing and did not refer patients or family members to a geneticist. [6] Thus, we need to evaluate the understanding of hearing genetics in a broader population in India to drive a national perspective. Overall, research on knowledge of audiologist on genetic disorders/syndromes requires more definitive data before any strong conclusion can be drawn. Therefore, the goal of this study was to go beyond unreliable data and fill research gaps concerning audiologists' pre-service and professional development needs.

Objectives

- To assess knowledge of genetics of hearing among practicing audiologists.
- To assess and compare the knowledge of genetics of hearing between graduates who have studied genetics as a part of their curriculum and those who have not.
- To assess the knowledge of the genetics of hearing based on years of working experience

Method

The Alumni of Sri Ramachandra Institute of Higher Education and Research (DU) & the Indian Speech and Hearing Association (with dual and specialised masters) were contacted. Additionally, some audiologists were contacted via Linked In. Based on quantitative methodology, a cross-sectional, descriptive survey study was conducted. Survey method was selected because data can be collected over a short period. The survey was conducted between February 15th and April 15th 2022. Consent forms were obtained from participants before the commencement of the survey. Only audiologists who agreed to participate in the survey had their responses collected. Individuals had to meet the inclusion criteria given below in order to participate in the survey. Participants could respond only once per email address. The responses were immediately saved in a database that was

accessible only to the investigator. The database replies were reviewed for missing information and responses that did not meet the inclusion criteria.

Inclusion criteria

- Audiologist with at least one year of working experience.
- Audiologists who completed postgraduate studies (M.Sc. Audiology or M.ASLP).
- Audiologists employed in various settings, including private clinics, multi-specialty or teaching hospitals, ENT, and pediatric clinics.
- Must be literate in English.

Exclusion criteria

- Audiologists not practicing in India at the time of the study.
- Audiologists with an undergraduate degree (B.ASLP).

The study was carried out in two phases.

Phase I: Development and validation of the questionnaire

A thirty-six-point questionnaire was developed based on clinical and research studies conducted *hitherto*, and inputs from practicing geneticists. Demographic details included questions about educational qualifications, employment settings, work experience and a self-rating scale of genetics-related hearing impairment knowledge. The survey comprises five domains. The first domain contains questions about non-syndromic hearing loss. It included questions about the exact gene mutation responsible for non-syndromic hearing loss, age at the commencement of non-syndromic hearing loss, auditory neuropathy mutations and the overall percentage of non-syndromic hearing loss. Seven questions about hearing deficits caused by syndromic conditions constitute the second domain. Most inquiries focus on hearing impairment caused by *Down syndrome*, *Waardenburg syndrome*, *Alport syndrome*, and *Usher syndrome*. The third domain addressed inheritance patterns. Eight questions in this area dealt with the inheritance of various types of hearing loss. All three domains required closed-ended answers. The fourth domain contained questions related to genetic counseling. Responses were collected using a 5-point Likert scale for agreement (strongly disagree, disagree, neutral, agree, strongly agree) and a 4-point Likert scale for frequency (always, often, occasion, never). The last domain included questions about genetic testing procedures, their applications, and their significance in diagnosing hearing loss.

Table 1 Characteristics of the participants

Characteristics	*N* (%)
Gender	
Male	66 (50%)
Female	67 (50%)
Age in years	
< 28 Yrs	70 (52.3%)
> 28 Yrs	63 (46.7%)
Experience (in years)	
Less than 5	74 (55.6%)
Equal to or more than 5	59 (44.4%)
Education	
M.ASLP	69 (51.5%)
M.Sc. Audiology	64 (47.8%)
Employment setup	
Teaching Hospital	35 (26.1%)
Hearing Aid Dispensing Centre	51 (38.1%)
Private Hospitals	26 (19.4%)
Multi-specialty Hospitals	21 (15.6%)

The content and format of the questionnaire were evaluated by two audiologists and two geneticists with more than five years of experience in their respective areas of specialisation. Experts were asked to comment on the relevance and clarity of the questions. The questions were revised and reorganised in response to expert critiques.

Phase II: Administration of the survey instrument

The developed questionnaire was converted into an electronic survey and sent online to the respondents using Google Forms. All licensed audiologists who met the criteria for participation received a link to the Google form. Emails were used to disseminate the survey data. This included a four-page questionnaire as an attachment and a cover letter, with a brief summary of the research. Three weeks after the initial email, two additional emails were sent to increase the number of survey responses. The information was retrieved from Google forms once data collection was finished and analysed using an Microsoft excel spreadsheet.

Results

1100 questionnaires were sent via email. Fifteen participants did not provide their consent to participate in the study. Ninety-three responses were returned. One hundred and fifty questionnaires were circulated through social media (Linked In), of which forty were returned. One hundred and thirty-four responses were received. One response was excluded as it did not meet the

inclusion criteria. The characteristics of the participants are listed in Table 1, and the self-rating scale is shown in Fig. 1.

Statistical analysis

The test data were analysed using a Microsoft Excel spreadsheet. Audiologists' responses to each question were profiled using percentage and frequency analysis. An independent *t* test was used to compare the audiologists' knowledge. Chi-square analysis was used to compare the practice items on a 5-point Likert scale. The collected data were analysed using IBM SPSS Statistics for Windows version 16.0. Armonk, NY: IBM Corp.

Knowledge-based questions

Tables 2, 3 & 4 comprise questions in domain 1, 2 & 3. Only (23.1%) were aware that 50% of hearing loss was non-syndromic (Table 2). Most of the audiologists answered incorrectly. Three-fourths (70.1%) of the audiologists correctly identified that in autosomal recessive inheritance, two copies of a gene are necessary for a trait to develop (Table 2). Only 38.8% of the participants correctly identified almost 400 syndromes with hearing loss (Table 3). More than three-fourths (79.1%) of the participants correctly identified that *Alport syndrome* caused progressive sensorineural hearing loss (Table 3). A greater number of audiologists (95.5%) correctly reported that *Down syndrome* is caused by a mutation in the 21st chromosome (Table 3).

Almost 91% of the audiologists correctly identified all the categories of genetic disorders (Table 4). The first pedigree represented autosomal recessive inheritance, and only 56.7% of participants were able to identify the inheritance pattern (Table 4). In the second pedigree,

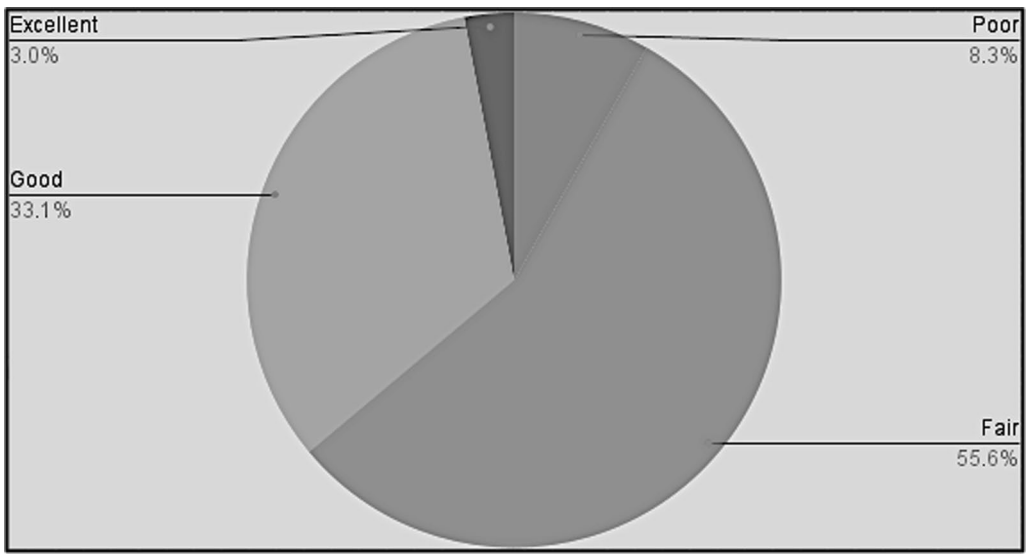

Fig. 1 Self-rating scale

Table 2 Overall knowledge of non-syndromic hearing loss (Domain 1)

Non-syndromic hearing loss	Correct response N (%)	Incorrect response N (%)
(1) What percentage of hearing loss is non-syndromic? (50%)	31 (23.3%)	102 6.7%)
(2) In autosomal recessive inheritance, how many copies of an abnormal gene must be present for a trait to develop? (Two copies)	93 (69.9%)	40 (31.1%)
(3) Select the most common gene mutation causing non-syndromic hearing loss. (*GJB2*)	113 (85%)	21 (15%)
(4) Sporadic mutation can occur in children with? (Healthy parents)	74 (55.6%)	59 (44.4%)
(5) Mutation in which gene causes auditory neuropathy? (*OTOF*)	90 (67.7%)	43 (32.3%)
(6) The age of onset of non-syndromic hearing loss can be? (Both pre-lingual and post-lingual)	111 (83.5%)	22 (16.5%)
(7) *DFNA9* gene mutation causes? (Late-onset progressive sensorineural hearing loss	89 (66.9%)	44 (33.1%)

only 41.8% of the audiologists correctly identified X-linked dominant inheritance (Table 4).

Practice based questions

Table 5 shows the responses to the practice questions. In the practice domain, when the audiologists were asked about their familiarity with the genetic aspects of the patient's condition, 42.5% reported being only slightly familiar, and 38.1% of the participants reported moderate familiarity. Only 0.7% of participants reported being extremely familiar with the genetic aspects of hearing.

55.2% of the participants reported that they would not refer the patient for genetic counseling when they did not see much benefit from genetic testing for this multiple-answer question, whereas 41.8% of the participants reported that not knowing how the counseling might help the patient as the reason for not referring patients for genetic counseling.

Table 3 Overall knowledge of syndromic hearing loss (Domain 2)

Syndromic hearing loss	Correct response N (%)	Incorrect response N (%)
(1) Approximately how many syndromes are identified with hearing loss? (400)	52 (39.1%)	81 (60.9%)
(2) *Waardenburg Syndrome* is an autosomal recessive inheritance causing? (Sensorineural hearing loss)	98 (73.7%)	35 (26.3%)
(3) *Alport Syndrome* which follows X-linked inheritance causes? (Progressive sensorineural hearing loss)	105 (78.9%)	28 (21%)
(4) *Usher Syndrome* type II causes? (Pre-lingual hearing loss with progressive vision loss)	85 (63.9%)	48 (36.1%)
(5) *Down syndrome* is caused by which chromosomal aberration? (Trisomy 21)	127 (95.5%)	6 (4.5%)
(6) *Down syndrome* causes which type of hearing loss? (Conductive hearing loss)	99 (74.4%)	34 (25.6%)
(7) *Neurofibromatosis* type II causes? (Bilateral sensorineural hearing loss)	121 (91%)	11 (9%)

Table 4 Overall knowledge of inheritance patterns (Domain 3)

Inheritance pattern	Correct response N (%)	Incorrect response N (%)
(1) Genetic diseases can be categorised into? (Single-gene, multifactorial, and chromosomal)	121 (91%)	12 (9%)
(2) For a patient with hereditary hearing loss, how many generations in a pedigree chart are required for finding the pattern of inheritance? (Three generations)	86 (64.7%)	47 (35.3%)
(3) How many copies of a gene are mutated to express the phenotype of a condition in autosomal dominant disorder? (One copy)	66 (49.6%)	67 (50.4%)
(4) The below pedigree chart shows which pattern/mode of inheritance? (Autosomal recessive inheritance)	76 (57.1%)	58 (42.9%)
(5) Mitochondrial associated mutation is? (Maternal inheritance)	51 (38.3%)	82 (61.7%)
(6) The below pedigree chart shows which pattern/mode of inheritance? (X-Linked dominant inheritance)	56 (42.1%)	77 (57.9%)
(7) *Usher's syndrome* is a type of? (Autosomal recessive inheritance)	79 (59.4%)	54 (40.6%)
(8) A newborn child has a bilateral profound sensorineural hearing impairment. This type of hearing impairment can be inherited in one of the following patterns? (Recessive or Dominant- Both)	88 (66.2%)	45 (33.8%)

Table 5 The overall practice of genetics in the field of audiology

Questions	Always n (%)	Frequently n (%)	Sometimes n (%)	Seldom n (%)	Never n (%)
(1) In your practice, how often do you take a family history/ pedigree chart?	51 (38.3%)	38 (28.6%)	23 (17.3%)	9 (6.8%)	12 (9%)
(2) How often do you refer a patient with concern for genetic evaluation?	10 (7.5%)	11 (8.3%)	55 (41.4%)	40 (30.1%)	17 (12.8%)
(3) How often do you discuss the genetic basis of the disorder/ condition with the client?	16 (12%)	36 (27.1%)	56 (42.1%)	21 (15.8%)	4 (3%)
(4) How often do you provide counseling or guidance to the clients in deciding whether to have genetic testing or not?	12 (9%)	20 (15%)	57 (42.9%)	20 (15%)	24 (18%)

In the final domain, when the participants were asked if they can practice genetic counseling, more than half (64.9%) reported that they could practice genetic counseling. Almost 67.9% of participants correctly identified karyotyping as the test used to identify structural and numerical abnormalities in the chromosome.

Comparison of knowledge and practice between groups of audiologists (M.ASLP and M.Sc. Audiology)

Tables 6, 7, and 8 show the responses to the knowledge questions between both groups of graduates. An independent *t* test was performed to compare the significance of the knowledge questions across the two groups. The findings revealed no significant differences (*P* value- 0.361). 75% of the respondents with a dual degree correctly identified the percentage of non-syndromic hearing loss, as compared to only 25% of audiologists with a specialised degree (Table 6). The difference in knowledge between the two groups was < 10% for the remaining questions. A very small percentage of respondents from both groups (42%, M.ASLP; 35.9%, M.Sc. Aud) correctly identified the total number of syndromes that caused the hearing loss (Table 7). The majority of the participants from both groups (88.4%, M. ASLP; 93%, M.Sc. Aud) correctly identified that genetic disorders can be categorised into single-gene, multifactorial, and chromosomal disorders. The difference in knowledge between the two groups was < 10% in the knowledge domain.

In the practice domain (Table 9), only 37.6% of the participants with a dual degree and 39% of the participants a with specialised degree reported always having a family history during their practice. Sixty percent of the audiologists with a dual degree and specialised master's degree have reported 'sometimes' 'seldom,' and 'never' for three out of four practice questions.

Sixty-five percent of the practitioners with a specialised master's degree and 66% of the practitioners with a dual master's degree have correctly identified that MLPA is a kind of genetic testing.

Comparison of knowledge and practice based on years of experience (< 5 years, ≥ 5 years)

Tables 10, 11, and 12 show the responses to the knowledge questions between both groups based on working experience. Respondents were divided based on their years of experience (< 5 and ≥ 5 years). The percentage of non-syndromic hearing loss reported correctly by audiologists with a 5-year experience was 68.92%, while only

Table 6 Comparison of knowledge between both groups of graduates (Domain 1)

Non-syndromic hearing loss	Correct response N (%)	Incorrect response N (%)
(1) What percentage of the hearing losses are non-syndromic? (50%)		
M.ASLP	52 (75.36%)	17 (24.64%)
M.Sc. Audiology	16 (25%)	48 (75%)
(2) In autosomal recessive inheritance, how many copies of an abnormal gene must be present for the trait to develop? (Two copies)		
M.ASLP	54 (78.26%)	15 (21.74%)
M.Sc. Audiology	39 (60.94%)	25 (39.06%)
(3) Select the most common gene mutation causing non-syndromic hearing loss. (*GJB2*)		
M.ASLP	60 (86.96%)	9 (13.04%)
M.Sc. Audiology	53 (82.81%)	11 (17.19%)
(4) Sporadic mutation can occur in children with? (Healthy parents)		
M.ASLP	29 (42.03%)	40 (57.97%)
M.Sc. Audiology	45 (70.31%)	19 (29.69%)
(5) Mutation in which gene causes auditory neuropathy? (*OTOF*)		
M.ASLP	45 (65.22%)	24 (34.78%)
M.Sc. Audiology	45 (70.31%)	19 (29.69%)
(7) The age of onset of non-syndromic hearing loss can be? (Both—pre-lingual and post-lingual)		
M.ASLP	55 (79.71%)	14 (20.29%)
M.Sc. Audiology	56 (87.5%)	8 (12.5%)
(8) *DFNA9* gene mutation causes? (Late-onset progressive sensorineural hearing loss		
M.ASLP	44 (63.77%)	25 (36.23%)
M.Sc. Audiology	45 (70.31%)	19 (29.69%)

(*P* value = .361, > .05)

Table 7 Comparison of knowledge between both groups of graduates (Domain 2)

Syndromic hearing loss	Correct response N (%)	Incorrect response N (%)
(1) Approximately how many syndromes are identified with hearing loss? (400)		
M.ASLP	29 (42.03%)	40 (57.97%)
M.Sc. Audiology	23 (35.94%)	41 (64.06%)
(2) *Waardenburg Sy*ndrome is an autosomal recessive inheritance causing? (Sensorineural hearing loss)		
M.ASLP	51 (73.91%)	18 (26.09%)
M.Sc. Audiology	47 (64.06%)	17 (26.56%)
(3) *Alport Syndrome* which follows X-linked inheritance causes? (Progressive sensorineural hearing loss)		
M.ASLP	54 (78.26%)	15 (21.74%)
M.Sc. Audiology	51 (79.69%)	13 (20.31%)
(4) *Usher Syndrome* type II causes? (Pre-lingual hearing loss with progressive vision loss)		
M.ASLP	44 (63.77%)	25 (36.23%)
M.Sc. Audiology	41 (64.06%)	23 (35.4%)
(5) *Down syndrome* is caused by which chromosomal aberration? (Trisomy 21)		
M.ASLP	65 (94.20%)	4 (5.8%)
M.Sc. Audiology	62 (96.88%)	2 (3.13%)
(6) *Down syndrome* causes which type of hearing loss? (Conductive hearing loss)		
M.ASLP	53 (76.81%)	16 (23.19%)
M.Sc. Audiology	47 (64.06%)	17 (26.56%)
(7) *Neurofibromatosis* type II causes? (Bilateral sensorineural hearing loss)		
M.ASLP	61 (88.41%)	8 (11.59%)
M.Sc. Audiology	61 (95.31%)	3 (4.69%)

(P value = 0.361, > 0.05)

20.3% of audiologists with a 5-year experience reported the same (Table 10). Three-fourths of the population from both groups correctly identified the type of hearing loss in *Down syndrome, Alport syndrome* and *Waardenburg syndrome* (Table 11). In the pedigree chart analysis, only 41.8% of audiologists with < 5 years of experience and 40.68% of audiologists with ≥ 5 years of experience responded correctly (Table 12). The difference in responses between the two groups was less than 10%.

The practice domain was also analysed based on years of work experience. Larger number of participants with < 5 years of experience (40.5%) reported having always inquired about their family history of hearing loss in their practice, whereas only 35.5% of participants with ≥ 5 years of experience reported that they inquired about their family history of hearing loss (Table 13).

Discussion

The present study proposes a questionnaire-based survey to measure the aspects of knowledge and practice in the genetics of hearing among audiologists. The results of this study revealed evidence of awareness and practice of hearing genetics among 133 audiologists across different states in India.

Knowledge

The questions for the present study were directed toward a fundamental genetic understanding of hearing loss. The knowledge domain elicited responses regarding awareness of non-syndromic hearing loss (Table 2), awareness of a syndromic condition that causes hearing loss (Table 3), and awareness of inheritance patterns responsible for different types of hearing loss (Table 4). The World Health Organisation (WHO) 2021 reported that the prevalence of genetic hearing loss in children is increasing. The present study reported that more than half of the practitioners were unaware of the frequency of non-syndromic hearing loss. It is important that with the increasing prevalence of non-syndromic hearing loss, knowledge and awareness of the genetics of hearing loss are significant to the professional profile of the audiologist.

The connection between genetic abnormalities and auditory neuropathy has been reported in many studies over the past 20 years [7]. In the present study, most participants were aware of the most common genes (OTOF and GJB2) responsible for the etiology of auditory neuropathy and non-syndromic hearing loss. This is in accordance with the results of a study conducted by Vishnuram et al. [6], where approximately half of the

Table 8 Comparison of knowledge between both groups of graduates (Domain 3)

Inheritance pattern	Correct response N (%)	Incorrect response N (%)
1.Genetic diseases can be categorised into? (Single-gene, multifactorial and chromosomal)		
M.ASLP	61 (88.41%)	8 (11.59%)
M.Sc. Audiology	60 (93.75%)	4 (6.25%)
2.For a patient with hereditary hearing loss, how many generations in a pedigree chart are required for finding the pattern of inheritance? (Three generations)		
M.ASLP	42 (60.87%)	27 (39.13%)
M.Sc. Audiology	44 (68.75%)	20(31.25%)
3.How many copies of a gene are mutated to express the phenotype of a condition in autosomal dominant disorder? (One copy)		
M.ASLP	31 (44.93%)	38 (55.07%)
M.Sc. Audiology	36 (56.25%)	28 (43.75%)
4. The below pedigree chart shows which pattern/mode of inheritance? (Autosomal recessive inheritance)		
M.ASLP	41 (59.42%)	28 (40.58%)
M.Sc. Audiology	35 (54.69%)	29 (45.31%)
5. Mitochondrial associated mutation is? (Maternal inheritance)		
M.ASLP	26 (37.68%)	43 (62.32%)
M.Sc. Audiology	25 (39.06%)	39 (60.94%)
6. The below pedigree chart shows which pattern/mode of inheritance? (X-Linked dominant inheritance)		
M.ASLP	33 (47.83%)	36 (52.17%)
M.Sc. Audiology	24 (37.5%)	40 (62.5%)
7. *Usher's syndrome* is a type of? (Autosomal recessive inheritance)		
M.ASLP	39 (56.52%)	30 (43.48%)
M.Sc. Audiology	40 (62.5%)	24 (37.5%)
8. A newborn child has a bilateral profound sensorineural hearing impairment. This type of hearing impairment can be inherited in one of the following patterns? (Recessive or Dominant- Both)		
M.ASLP	45(65.22%)	24 (34.78%)
M.Sc. Audiology	43 (67.19%)	21(32.81%)

(P value = 0.361, > 0.05)

participants correctly identified GJB2 as the most common gene associated with non-syndromic hearing loss.

Approximately 400 syndromes have been associated with HL. In the present study, only few participants reported correct answers. A previous dichotomous survey identified awareness of syndromes in almost half of the respondents [6]. This reduction in the correct response rate is probably due to the multiple-choice questions used in the present study and the dichotomous survey (yes/no) used an earlier study. In the present study regard to the syndromes, the audiologists were able to provide adequate answers. Nearly all audiologists correctly identified chromosomal aberrations that cause *Down syndrome* and the type of hearing loss that affects a person with *Down syndrome*. The increased prevalence and awareness of *Down syndrome* could have contributed to exceptional knowledge and responses. The prevalence of *Down syndrome* in India is one in every 692 births [8]. Camps and awareness initiatives for *Down syndrome* have increased in recent years. This could have contributed to better knowledge compared with other syndromes.

Understanding the underlying causes of any condition critically depends on identifying the inheritance patterns [2]. Therefore, knowing the inheritance pattern helps the audiologist advise the members of the family for future evaluation and management. In this study, only half of the participants were able to correctly identify the type of inheritance based on the given example in the questionnaire (Table 4). Most practitioners demonstrated a lack of knowledge of inheritance patterns (Table 4).

Overall, there is a limited understanding of inheritance patterns in the current investigation. Our present study correlated with the results of a survey conducted on 162 healthcare professionals [9], who reported poor knowledge of medical genetics. Vishnuram et al. [6] also reported that audiologists had insufficient knowledge of genetics and practical genetics skills.

Table 9 Comparison of practice between graduates

Questions	M.ASLP	M.Sc. Aud	*P* value
1. In your practice, how often do you take a family history/ pedigree chart?			
Always	26 (37.6%)	25 (39%)	0.073
Frequently	26 (37.6%)	12 (18.7%)	
Sometimes	8 (11.5%)	15 (23.4%)	
Seldom	5 (7.2%)	4 (6.2%)	
Never	4 (5.8%)	8 (12.5%)	
2. How often do you refer a patient with concern for genetic evaluation?			
Always	7 (10.1%)	3 (4.6%)	0.153
Frequently	5 (7.2%)	6 (9.3%)	
Sometimes	33 (47.8%)	22 (34.3%)	
Seldom	19 (27.5%)	21 (32.8%)	
Never	5 (7.2%)	12 (18.7%)	
3. How often do you discuss the genetic basis of the disorder/condition with the client?			
Always	8 (11.5%)	8 (12.5%)	0.396
Frequently	22 (31.8%)	14 (21.8%)	
Sometimes	30 (43.4%)	26 (40.6%)	
Seldom	8 (11.5%)	13 (20.3%)	
Never	1 (1.4%)	3 (4.6%)	
4. How often do you provide counseling or guidance to the clients in deciding whether to have genetic testing or not?			
Always	5 (7.2%)	7 (10.9%)	0.059
Frequently	16 (23.1%)	4 (6.2%)	
Sometimes	28 (40.5%)	29 (45.3%)	
Seldom	11 (15.9%)	9 (14%)	
Never	9 (13%)	15 (23.4%)	

Practice and attitude of the audiologists regarding the genetics of hearing

The present study also explored audiologists' attitudes and practices toward the genetics of hearing. Questions were constructed for different scenarios during the audiologist's practice (Table 5). Family history or pedigree charts should be considered as a part of the case history. Approximately half of the participants reported taking family history/pedigree charts during their practice, which contrasted with the results of the study conducted by [6], who reported that all audiologists practiced pedigree charts mandatorily during case history.

Audiologists must appropriately refer patients' concerns to other healthcare providers in their capacity as healthcare experts. Only a few audiologists in the present study (Table 5) reported referring patients with genetic concerns for testing or counseling. The results of the present study is similar to a previous study [10], that reported that very few professionals had high confidence in referring patients to genetic professionals. *E Sarlier* research stated that many audiologists were not confident in making decisions regarding referrals to genetic professionals [2]. This may be a result of their lack of expertise in genetics and inadequate genetic background. However, another study reported that the majority of participants indicated that they did not refer to genetic counselors [11]. This is because of the discrepancy between reported awareness and utilisation of services. One of the main reasons for fewer referrals was the lack of awareness of genetic services among professionals.

This assertion is corroborated by the present study because when the audiologists were asked the reason for fewer referrals, the majority reported not being aware of any professional genetic services as the major reason for their perception of lower referral advantages or outcomes. This was validated by a previous study that stated that Ohio speech-language pathologists did not know where and how to find genetic counselors to make appropriate referrals which was yet another reported barrier to referring patients for genetic counseling [12]. More than half of the participants were unaware of how to refer patients, the next half were unaware of the genetic services around them, and few felt that it was not their responsibility to refer patients for genetic counseling [13].

Informing the family of the hereditary origin of the condition is a necessary step in therapy and management planning. As soon as the need for a referral is established, an audiologist must discuss the hereditary cause of the disorder. Once parents understand the reason for hearing loss, they are more inclined to undergo genetic testing [2]. More than half of the audiologists in the present study reported no significant benefits from genetic counseling. More training is needed for audiologists to understand the benefits of genetic counseling.

Audiologists have to be equipped with knowledge of genetic testing to provide family guidance when deciding on testing. In the present study, very few participants reported having provided counseling or guidance for patients to decide on genetic testing (Table 5). [10] reported that only a few participants in their study were confident in guiding clients regarding testing. This finding was consistent with the results of the present study. Due to their lack of expertise in certain areas of genetics, clinicians are less likely to feel confident informing parents about genetic testing or counseling. There is a gap between audiologists' knowledge of genetics of hearing and their practice of genetics in their field.

Genetic counseling in management

In the present study, approximately half of the participants reported that they could provide genetic counseling during their practice. [6] Similar results reported that the majority of audiologists agreed that they could provide genetic counseling to patients during their

Table 10 Comparison based on working experience (Domain 1)

Non-syndromic hearing loss	Correct response N (%)	Incorrect response N (%)
(1) What percentage of the hearing losses are non-syndromic?		
Less than 5 years	51 (68.92%)	23 (31.08%)
5 or more than 5 years	12 (20.3%)	47 (79.6%)
(2) In autosomal recessive inheritance, how many copies of an abnormal gene must be present for the trait to develop?		
Less than 5 years	62 (83.78%)	12 (16.22%)
5 or more than 5 years	43 (72.8%)	16 (27.12%)
(3) Select the most common gene mutation causing non-syndromic hearing loss		
Less than 5 years	39 (52.7%)	35 (47.3%)
5 or more than 5 years	51 (86.44%)	8 (13.56%)
(4) Sporadic mutation can occur in children with?		
Less than 5 years	49 (66.22%)	25 (33.78%)
5 or more than 5 years	35 (59.32%)	24 (40.68%)
(5) Mutation in which gene causes auditory neuropathy?		
Less than 5 years	45 (60.81%)	29 (39.19%)
5 or more than 5 years	42 (71.19%)	17 (28.81%)
(6) The age of onset of non-syndromic hearing loss can be?		
Less than 5 years	62 (83.78%)	12 (16.22%)
5 or more than 5 years	49 (83.05%)	10 (16.95%)
(7) *DFNA9* gene mutation causes?		
Less than 5 years	48 (64.86%)	26 (35.14%)
5 or more than 5 years	41 (69.49%)	18 (30.51%)

P value- 0.712, > 0.05

practice. Some audiologists in the present study reported that they were not confident about providing counseling. The reason for this might be that the participants misunderstood that audiologists should only refer to and not provide genetic counseling. Another study reported that knowledge of the role of genetic counselors among AHS professionals is limited [13].

Multiplex ligation-dependent probe amplification is a technique used to detect gene deletions/duplications. The present study reported that the majority of audiologists appropriately conveyed that MLPA was a type of genetic testing. An audiologist must be aware of basic genetic testing and its process to explain it to patients and their families. As they carefully counsel patients and their families, they should be informed of the potential benefits and drawbacks of genetic testing.

Comparison of knowledge and practice between graduates

The present research is a novel study that compares the knowledge and practice of audiologists based on their educational qualifications. In the current study, there was no statistical difference in the overall knowledge between the graduates. Both groups of audiologists lacked knowledge of a few aspects of the genetics of hearing, such as finding the inheritance pattern, different types of inheritance and mutations. The overall knowledge of both groups is insufficient and more emphasis should be placed on the theory of genetics in hearing to understand the basics and practice them in the field of audiology.

Even though audiologists reported better performance on a few knowledge questions, there was a dearth in the practice of genetics of hearing in their field, which is important. Although the genetics course was a part of the curriculum in the specialised master's degree, this did not influence the knowledge and practice of audiologists.

Comparison of knowledge and practice based on experience

In this present study, a comparison was made based on work experience. The present study is novel in that it compares work experience of audiologists. Audiologists were divided into two groups (less than 5 years and five or more years of working experience). Both knowledge and practice components did not differ statistically between the two groups. Small differences in knowledge were noted for certain questions between the two groups.

Table 11 Comparison based on working experience (Domain 2)

Non-syndromic hearing loss	Correct response N (%)	Incorrect response N (%)
(1) Approximately how many syndromes are identified with hearing loss?		
Less than 5 years	28 (37.84%)	46 (62.16%)
5 or more than 5 years	24 (40.68%)	35 (59.32%)
(2) *Waardenburg syndrome* is an autosomal recessive inheritance causing?		
Less than 5 years	52 (70.27%)	22 (29.73%)
5 or more than 5 years	46 (77.97%)	13 (22.03%)
(3) *Alport syndrome* which follows X-linked inheritance causes?		
Less than 5 years	58 (78.38%)	16 (21.62%)
5 or more than 5 years	47 (79.66%)	12 (20.34%)
(4) *Usher syndrome* type II causes?		
Less than 5 years	46 (62.16%)	28 (37.84%)
5 or more than 5 years	38 (64.41%)	21 (35.59%)
(5) *Down syndrome* is caused by which chromosomal aberration?		
Less than 5 years	72 (97.3%)	2 (2.7%)
5 or more than 5 years	55 (93.22%)	4 (6.78%)
(6) *Down syndrome* is caused by which type of hearing loss?		
Less than 5 years	53 (71.62%)	21 (28.38%)
5 or more than 5 years	46 (77.97%)	13 (22.03%)
(7) *Neurofibromatosis* type II causes?		
Less than 5 years	68 (91.89%)	6 (10.81%)
5 or more than 5 years	53 (89.83%)	6 (10.17%)

P value- 0.712, > 0.05

A gap was reported between knowledge of genetics and practice in audiologists based on the results of the current study. Although differences in the years of experience were noted between the audiologists, the practice of genetics remained the same in both groups.

The last question of the present study was regarding additional training requirements for audiologists to help them integrate genetics into their clinical practice. Nearly 90% of audiologists expressed interest in and willingness to learn more about hearing-related genetics. This was supported by a previous study, in which nearly two-thirds of the respondents reported that they would be interested in attending educational workshops in genetics to increase their professional confidence [10]. Yet another study also authenticated the present study, with similar findings where almost 93% of the allied-healthcare professionals were interested in improving their knowledge in genetics [13]. Due to the availability of additional technologies and the under preparedness of healthcare workers in this area, there has been a discernible increase in interest in promoting genetic-related courses during the past few years.

Many websites provide information on genetics, such as the NCHPEG (National Coalition for Health Professional Education in Genetics) website (www.nchpeg.org), is available exclusively for speech-language pathologists and audiologists. They have accurately addressed genetic concepts from the perspectives of all professions and collaborated with organisations that include specialists such as SLPs, audiologists, and genetic counselors. To provide patients with the most relevant information, there should be greater emphasis on genetics in the curriculum and practice of audiology.

Conclusion

Understanding the genetic factors is vital for the appropriate diagnosis, management, and counseling of people with hearing loss. The present research has contributed to the development of evidence-based techniques for increasing knowledge in genetics and practice in audiology, ultimately improving patient outcomes and the discipline of audiology. More emphasis on practical knowledge of inheritance patterns, genetic counseling and genetic testing and their benefits must

Table 12 Comparison based on working experience (Domain 3)

Non-syndromic hearing loss	Correct response N (%)	Incorrect response N (%)
(1) Genetic diseases can be categorised into?		
Less than 5 years	65 (87.84%)	9 (12.16%)
5 or more than 5 years	55 (93.22%)	4 (6.78%)
(2) For a patient with hereditary hearing loss, how many generations in a pedigree chart are required for finding the pattern of inheritance?		
Less than 5 years	50 (67.57%)	24 (32.43%)
5 or more than 5 years	37 (62.71%)	22 (37.29%)
(3) How many copies of a gene are mutated to express the phenotype of a condition in an autosomal dominant disorder?		
Less than 5 years	41 (55.41%)	33 (44.59%)
5 or more than 5 years	26 (44.07%)	33 (55.93%)
(4) The below pedigree chart shows which patterns/mode of inheritance? (Autosomal recessive)		
Less than 5 years	45 (60.81%)	29 (39.19%)
5 or more than 5 years	31 (52.54)	28 (47.46%)
(5) Mitochondrial associated mutation is?		
Less than 5 years	27 (36.49%)	47 (63.51%)
5 or more than 5 years	24 (40.68%)	35 (59.32%)
(6) The below pedigree chart shows which pattern/mode of inheritance? (X-linked dominance)		
Less than 5 year	31 (41.89%)	43 (58.11%)
5 or more than 5 years	24 (40.68%)	35 (59.32%)
(7) *Usher's syndrome* is a type of?		
Less than 5 years	46 (62.16%)	28 (37.84%)
5 or more than 5 years	33 (55.93%)	26 (44.07%)

P value- 0.712, > 0.05

be included in the curriculum and clinical practice because auditory genetics is a significant emerging discipline that has the potential to play a vital role in the practice of audiology in the future.

Practical implications of genetic education for audiologists:

- The detection of genetically associated hearing loss is achievable with a better understanding of the genetic basis of hearing loss.
- The early diagnosis of hereditary hearing loss is possible if audiologists are aware of the genetic testing used to identify these specific genetic abnormalities.
- Educating patients and families about the risks of hereditary hearing loss and providing them with resources and assistance.
- An understanding of genetic aspects assists audiologists in developing a tailored, effective therapy plan for each patient.
- Novel cutting-edge research in the field of auditory genetics can be developed. Researchers can develop new treatments and interventions that may be more effective in treating hearing loss by investigating the genetic basis of hearing loss [14].

Limitations

The current study had a mixed sample of respondents with regard to educational qualifications, work experience, work setting, and age. Also, the sample size of this study was limited. Future research expansion should gather a greater variety of responses to permit more generalisation.

Table 13 Comparison of practice based on working experience

Questions	5 or more than 5 years N (%)	Less than 5 years N (%)	*P* value
(1) In your practice, how often do you take a family history/ pedigree chart?			
Always	21 (35.59%)	30 (40.54%)	
Frequently	21 (35.59%)	17 (22.97%)	
Sometimes	8 (13.56%)	15 (20.27%)	
Seldom	5 (8.47%)	4 (5.41%)	0.128
Never	4 (6.78%)	8 (10.81%)	
(2) How often do you refer a patient with concern for genetic evaluation?			
Always	7 (11.86%)	3 (4.05%)	
Frequently	5 (8.47%)	6 (8.11%)	
Sometimes	26 (44.07%)	29 (39.19%)	
Seldom	17 (28.81%)	23 (31.08%)	
Never	4 (6.78%)	13 (17.57%)	0.097
(3) How often do you discuss the genetic basis of the disorder/condition with the client?			
Always	8 (13.56%)	8 (10.81%)	
Frequently	16 (27.12%)	20 (27.02%)	
Sometimes	24 (40.68%)	32 (43.24%)	
Seldom	9 (15.25%)	12 (16.22%)	0.218
Never	2 (3.39%)	2 (2.7%)	
(4) How often do you provide counseling or guidance to the clients in deciding whether to have genetic testing or not?			
Always	5 (8.47%)	7 (9.46%)	
Frequently	11 (18.46%)	9 (12.16%)	
Sometimes	27 (45.76%)	30 (40.54%)	
Seldom	7 (11.86%)	13 (17.57%)	0.167
Never	9 (15.25%)	15 (20.27%)	

Acknowledgements
The authors thank the management of Sri Ramachandra institute of Higher Education and Research for the facility and support for the conduct of this research.

Author contributions
HM and VDP have contributed to the design, conduct, analysis, and manuscript preparation. SH has contributed to conduct of the study.

Funding
No funding was received for the present study.

Availability of data and material
The data are available with corresponding author.

Declarations

Ethical approval and consent to participate
Has been obtained from Institutional Ethical committee 2022.

Consent for publication
Is provided by all authors.

Competing interest
All the authors declare that they have no competing interest.

Received: 7 February 2023 Accepted: 6 June 2023
Published online: 15 June 2023

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