

# Genetic Knowledge among Audiologist Related to Hearing Loss

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## ABSTRACT

Hearing loss is the loss of hearing which may be due to tumors, trauma, genetic factors, aging etc. The main aim and objective of the study is experimental studies conducted regarding knowledge about hearing loss related to genetics, involving 100 participants out of which 31 responded. 25 questions were validated and send through e-mails, responses from all the active participants were analysed. Findings have implicated that the audiologist have sufficient knowledge but practical skills are insufficient and there is a need to know about genetics of hearing loss. Knowledge about this increases self-confidence, better planning and treatment, better understanding of genetic disorders related to audiology.

**Keywords:** Audiologists; Genetics; Hearing loss; Knowledge

## INTRODUCTION

Hearing loss is relatively common in the human population. Profound congenital hearing loss is estimated to occur in about 1 in 1000 births [1,2]. Early intervention has been shown to be effective in facilitating speech and language development in deaf and hard-of-hearing children [3,4]. Ninety-five percent of newborns with hearing loss identified by newborn hearing screening programs are born to hearing parents, obscuring the fact that the majority of newborns have a hereditary cause for their hearing loss. The majority of genetic hearing loss is inherited in an autosomal recessive pattern and often presents in the absence of a positive family history for hearing loss [5]. One gene, GJB2, which encodes the gap junction protein connexin 26, accounts for the largest proportion of autosomal recessive early childhood hearing loss in many populations [6-9]. If a specific etiology is known, descriptions of hearing loss may also include the etiologic diagnosis, such as Usher syndrome type 1-related hearing loss or GJB2-related hearing loss.

## Epidemiology

Approximately 50% of cases are through environmental factor which include bacterial or viral infections, acoustic trauma, ototoxic drug exposure. Remaining 50% of cases through genetic factor which include non-syndromic and syndromic hearing loss. In that genetic factor Approximately 70% of cases are associated with

non-syndromic hearing loss and 30% associated with syndromic hearing loss in that more than 400 forms of syndromic deafness can be diagnosed with symptom of hearing loss [10].

## Genetic and non-genetic cause hearing loss

Nonsyndromic hearing loss may be transmitted as an autosomal recessive (~80%), autosomal dominant (~15%), or X-linked trait (~1%) [11]. More than 150 deafness-causing variants have been identified in GJB2, but a few common mutations account for a large percentage of alleles in several populations. GJB2-related hearing loss is sensorineural, usually present at birth, typically bilateral and nonprogressive, and can range from mild to profound in severity. Age-related hearing loss, or presbycusis, is a common neurosensory deficit. Presbycusis generally affects higher frequencies of sound disproportionately, making it difficult for those with presbycusis to understand speech. Presbycusis is a complex condition influenced by genetic and environmental factors. Certain environmental (nongenetic) factors play a major etiologic role in hearing loss. Congenital CMV infection is the most common nongenetic cause of hearing loss among children.

## NEED OF THE STUDY

Audiologist's in the Early Intervention may play an important role in discussing the complex relationships between developmental and communication disorders and genetics with other members of the Individualized Educational Program team and ultimately

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influence clinical decisions for students and their families. In general, the literature on the knowledge among audiologist about genetic disorders/syndromes clearly needs more definitive data before any solid conclusions can be reached.

Therefore, the purpose of this study was to survey knowledge among audiologist about genetics beyond anecdotal reports and fill research gaps regarding Audiologists' pre service and professional development needs.

## AIM OF THE STUDY

To find the awareness among the audiologist about the impact of genetic related hearing loss.

## OBJECTIVE

- To investigate about genetic knowledge related to hearing among audiologist.
- To find the knowledge about genetic evaluation and genetic counseling in the field of audiology.
- To find the knowledge among audiologist about gene therapy, this is needed in hearing related genetic problems.

## METHODOLOGY

Research design: Experimental study

Research sampling: Convenient samplings

Sample size: 100 Participants.

### Selection criteria

**Inclusion criteria:** The criteria for subject selection were that respondents had to be practising at the time of the study and literate in English.

### Method

From the 50 question distributed to senior audiologists practising in Telangana, based on the importance, the questions were reduced to 25 number and validated. Questionnaire is distributed in the form of Google Forms through email and through social media and made sure that subjects can only once enter their responses using their email Id.

### Statistical analysis

Descriptive statistics formed the basis of the data analysis procedure. The data collected from the questionnaires were encoded and then tabulated and analyzed using the IBM SPSS Software, Version 20. Averages, means and statistical significance were used to analyse the data.

## RESULTS

One hundred professionals were e-mailed of which 31 responded (response rate of 31%). A majority of the respondents were females 67.7% (n=21) while the remaining 32.3% were males (n=10). Out of 31 responds 44.4% were Students, 56.6% professionals who are practicing at hospitals, clinics and private practice (Figure 1).

The above results shows Audiologists' percentages of correct and incorrect responses on the 22-item. The mean number of questions answered correctly was 13.5 (S.D.=3.2; median 14) with a range

from 7 to 18. These scores converted to a range of 31.8%–81.8%, with a mean summary score of 66.2% correct responses and a median score of 63.6% correct responses. A review of result shows the majority of respondents (>50%) provided correct responses to 15 items. Eleven of the 22 items were answered correctly by more than 75% of Audiologists. Of the total of 682 responses (31 participants \*22 questions), 52.1% (n=356) were correct, 21.9% (n=150) were incorrect and 25.8% (n=176) were unsure.

Respondents were asked that is it necessary for audiologist to know about Genetics and its related effect in hearing and related disorders most of responds accept that its is mandatory to have the knowledge related to genetics regarding Hearing and its disorders with the percentage of 77.8% only 22% tells that they don't know whether knowledge about genetics is related with Audiology. All I.e., 100% responds agree that there are chance that hereditary plays a role in hearing impairments (Figure 2).

Respondents were more confused in the question related to transfer of affected gene equal percentage (43.7%)of population have no idea that even a healthy parents can have child with hereditary disease/disorder.but most of respondents (93.5%) can able to say that gene for the child is equally comes from paternal and mother.

Eighty Six percent of the response can able to correctly say that equal amount of hearing impairments were caused due to Genetic reasons. Only 4.8% were don't know that Connexin 26 gene is the most common affected gene in Genetic related hearing impaired populations.

General questions regarding types and carrier of affected gene we can able to see almost equal response 56% can answer correctly 44% were unable to answer correctly but it surprising to see that more amount of correct response if the cue were provided with the famous syndrome. Most of our respondents were able to correctly say either syndrome comes under which type of genetic category i.e., Autosomal recessive, autosomal dominant, mitochondrial or X linked (Table 1).

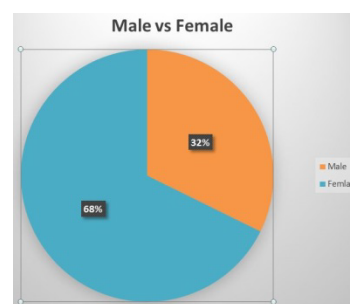


Figure 1: Male vs. Female.

### NEED FOR AUDIOLOGIST TO KNOW ABOUT GENETICS

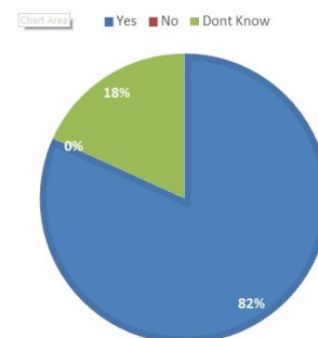


Figure 2: Need for audiologist to know about genetics.

Table 1: Responses from the audiologists.

Educational qualification	UG		PG
	-54.30%		-45.70%
What is your current primary work setting?	Clinical/hospital (25.6%)	Private practice (46.3%)	Others (28.1%)
Is it necessary to have knowledge on genetics in Audiology?	Yes (43.2%)	No (26.8%)	Don't know (30%)
Is hearing loss, a hereditary disorder?	Yes (81.8%)	No (18.2%)	Don't know (0%)
Is Pedigree Chart necessary during History taking?	Yes (100%)	No (0%)	Don't know (0%)
Genetic diseases can be categorized into single-gene, chromosomal, and multifactorial.	Yes (72.7%)	No (9.1%)	Don't know (18.2%)
Healthy parents can have a child with a hereditary disease.	Yes (63.6%)	No (9.1%)	Don't know (27.3%)
Half your genes come from your mother and half from your father.	Yes (63.3%)	No (27.3%)	Don't know (9.1%)
More than 100 genes are responsible for hearing loss?	Yes (18.2%)	No (36.4%)	Don't know (45.5%)
70 genes are identified with hearing loss?	Yes (36.4%)	No (9.1%)	Don't know (54.5%)
Hearing loss is divided into syndromic vs. non-syndromic hearing loss?	Yes (81.8%)	No (18.2%)	Don't know (0%)
50% of hearing loss is due to Genetics?	Yes (72.7%)	No (27.3%)	Don't know (0%)
Connexin 26 on the GJB2 is a most common gene of non-syndromic hearing loss?	Yes (45.1%)	No (13%)	Don't know (41.9%)
More than 400 Syndromes are identified with Hearing loss	Yes (54.5%)	No (9.1%)	Don't know (36.4%)
An Autosomal recessive disorder means two copies of an abnormal gene must be present in order for the disease or trait to develop	Yes (36.4%)	no (36.4%)	Don't know (27.3%)
If a person is the carrier of a disease gene it means that they have the disease.	Yes (9.1%)	No (63.6%)	Don't know (27.3%)
Waardenburg syndrome is a one of the syndromic autosomal dominant syndrome?	Yes (100%)	No (0%)	Don't know (0%)
Usher syndrome is a autosomal recessive syndrome?	Yes (54.5%)	No (36.4%)	Don't know (9.1%)
Alpert syndrome is a mitochondrial syndrome?	Yes (36.4%)	No (45.5%)	Don't know (18.2%)
Kearns-sayre syndrome (KSS) is X-Linked syndrome?	True (36.4%)	No (27.4%)	Not sure (36.4)
Helping the people to understand how Genetic affect their health and lives is termed as	Genetic evaluation (0%)	Genetic counseling (100%)	Genetic therapy 0%
How many generations are needed to evaluate genetic related disorders	1-2 (0%)	2-4(100%)	5-6 (0%)
Can Audiologist do Genetic Counseling	Yes (63.6%)	No (0%)	May be (36.4%)
GF 166 IS	Gene therapy (54.5%)	Genetic counseling (0%)	Gene evaluation (45.5%)

Nearly 89.6% of respondents were able to give the correct response that GF166 is the gene therapy used for Genetic related hearing impairment, but less than 25% are agreeing that an audiologist cannot able to do Genetic counselling may be others were misunderstood that audiologist can refer the clients for genetic counseling and we can't provide genetic counseling.

## DISCUSSION

One of the important findings of this study was that audiologist's who provide services to the largest number of children with genetic disorders/syndromes. It appears that Audiologist's who were more likely to provide services to a larger number of children with genetic disorders/syndromes also acquired more learning and knowledge. This enhanced knowledge may lead to the likelihood of having greater confidence in working with students, parents and caregivers, while also assisting in the development of positive attitudes toward genetics assessment, treatment, and prevention for children with communication and genetic disorders on their caseloads.

Telangana audiologist's on average regard their knowledge is sufficient and practical genetic skills as insufficient. As a result, only some of their patients receive information on the hereditary nature of syndromic hearing loss, the majority of audiologist never refers patients to a clinical geneticist, and DNA tests in patients or family members are seldom initiated. In a recent systematic review of 68 peer reviewed articles about the delivery of genetic health care services for adult-onset conditions, the authors concluded that

the healthcare field is unprepared to meet the informational and clinical needs of consumers with genetic disorders. They remarked that continuing education for all healthcare professionals should be a top educational priority [12].

The survey items in this study were comprised of basic knowledge and were not specific to any one genetic-based syndrome. The lack of mastery of even basic genetic knowledge by many audiologists may become more problematic as a result of rapid information exploration resulting from the Human Genome Project. Early identification of genetic disorders and syndromes could result in increased numbers of clients receiving services by Audiologist's as early as possible. It is concerning that at a "basic level of knowledge" Audiologist's scored a mean and median score of 66.2% correct responses. The results suggest current needs of Audiologists' education in genetics are not being addressed or met.

Vural et al. [13] reported on a survey of 162 nursing students' knowledge of medical genetics. The majority of students reported only "some" to "minimal" knowledge of Mendelian genetics. The authors supported the need for changes in the current education and training for nursing students in the area of genetics. The GKI score results in this study support other studies suggesting the need for changes in curriculum for more healthcare and educational professionals in genetics education [14-16]. One could speculate that when an audiologist learns a child with a genetic disorder/syndrome is scheduled for an assessment, the audiologist may immediately obtain new knowledge in preparation for the evaluation

and treatment Audiologists with greater knowledge about genetic disorders/syndromes may be more alert for students displaying certain characteristics, thereby increasing the likelihood for a greater number of assessments and treatment of specific children. The Audiologists' experiences of working with children with genetic disorders/syndromes may serve to increase their knowledge bases, resulting in more positive outcomes. Audiologists need to find multiple ways to acquire new knowledge and experiences with children with genetic syndromes/disorders. It would appear that curricular innovations for Audiologists' preparedness in working with children with genetic disorders/syndromes and their families are warranted. This might include use of NHPEG modules, enhanced educational opportunities and courses in undergraduate and graduate training programs, as well as in-service workshops and web-based courses for current audiologists working in the hospitals and private practice [17-19].

## SUMMARY AND CONCLUSION

The results of our study showed that the knowledge acquisition about genetics through regular educational system is very important for Audiologists, in conditions when we can notice the increase in the number of children with genetic and verbal communication disorders. Level of general knowledge influences the increase in self-confidence of Audiologists in working with children because the increased level of knowledge provides better planning of treatment in accordance with child's potentials. Level of general knowledge also leads to a better understanding of influence of genetic disorders on Audiology and thus to the more positive attitudes to the genetic influence on Audiology. The findings of the present study are in tune with those reported on Audiologists from other geographic locations. Most of the professionals reported that they learnt about genetics at school, this warrants the need to provide changes in the professional studies curriculum and provide more sources of learning to update their knowledge in genetics. There are several opportunities for additional research in the area of genetics that is clinically related to speech and hearing related disorders.

## LIMITATIONS

Sample size is very less for generalization since it's sent through e-mail the response have a chance of biased. The response obtained was completely based on honesty of the respondents. Results can be analyzed educational qualification and with the work experience so that it can give much more clear idea.

## FUTURE RESEARCH

More amounts of samples can be taken. This can be done with Students and can be done across various work setups (Hospital vs. Private). Questions can be much deeper about how it has an effect.

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