A PROBE INTO THE AUDIOLOGICAL FINDINGS OF SIBLINGS WITH HEARING LOSS IN THEIR FAMILIES

Reg. No. M9921

An independent project submitted as part fulfillment of the First Year M.Sc. (Speech & Hearing) to the University of Mysore.

ALL INDIA INSTITUTE OF SPEECH & HEARING
MYSORE - 570 006

MAY 2000

DEDICATED TO

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CERTIFICATE

This is to certify that the independent project entitled *"A PROBE INTO THE AUDIOLOGICAL FINDINGS OF SIBLINGS WITH HEARING LOSS IN THEIR FAMILIES"* is a bonafide work in part fulfillment of the First Year M.Sc. in Speech & Hearing of the student with Reg. No. M 9921.

Mysore

May 2000

DIRECTOR
All India Institute of Speech & Hearing
Mysore.

CERTIFICATE

This is to certify that the independent project entitled ***"A* PROBE INTO THE AUDIOLOGICAL FINDINGS OF SIBLINGS WITH HEARING LOSS IN THEIR FAMILIES"** has been prepared under my supervision and guidance.

Mysore

May 2000

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DECLARATION

I hereby declare that this independent project entitled "A **PROBE INTO THE AUDIOLOGICAL FINDINGS OF SIBLINGS WITH HEARING LOSS IN THEIR FAMILIES"** is the result of my own study under the guidance of **DR. K. RAJALAKSHMI,** Lecturer in Audiology, All India Institute of Speech & Hearing, Mysore & has not been submitted earlier to any University for any Diploma or Degree.

Mysore

May 2000 Reg. No. **M 9921**

A CKNO WLED GEMENTS

I sincerely thank my guide Dr. Rajalakshmi, Lecturer in Audiology for her constant support, valuable suggestions and patient guidance for completing this study. Thank you very much Ma'am..

I would like to thank Dr. (Miss) S. Nikam. Director. All India Institute of Speech and Hearing, for giving me opportunity to do this study.

My hearty thanks to all the subjects who participated in this study and especially to all parents of the children who participated, for spending their valuable time for my study.

Special thanks to Dr. Venkatesan, Reader in Psychology. AIISH. for guiding me to do statistical analysis

I would like to thank Anita Didi. Sapna Bhat, & Mr. Gururaj for helping me with my data collection.

Dear Appu, I don't know how to thank for the support you have give me.

Special thanks to Anshula. for balancing me whenever I went through various mood swings.

Mini.... a new bond of friendship which I made in AIISH.

Last but not the least, I would like to thank all my classmates, especially Aruna, Komal. Prachi, Lakshmi for their help in different stages of my study.

Finally, thanks to Mr. Sridhara for typing my IP and struggling with my hand writing.

CONTENTS

INTRODUCTION

The old German colloquialism says,

'the acorn doesn't fall far from the tree" - the notion that behaviour runs in families. The word gene is derived from the same root that gives us genesis and generation. It means to be derived from, or to begin with. The concept of genetics began with the observation that traits ran in families and children looked like their parents.

Human take great pride in identifying distinguishing traits from one generation to the next. Genes are found in the nuclei of the many cells that compose the body. Genes are concerned with the determination of what a person's characteristic shall be. and they form the hereditary link between one generation to the next. The characteristics of an offspring are. to a large degree, determined by the genes he or she receives from parents - from the mother through the ovum or egg and from the father through the sperm cell that fertilized that ovum at the time of conception. Genetic factors present at conception are largely unaltered throughout the life.

For three reasons, knowledge of genetics and molecular biology is becoming more important in the evaluation of children and adults with sensorineural hearing impairment and deafness. First, during the past two decades, there has been a change in the most common cause of severe and profound sensorineural hearing loss. The chances that a child will lose hearing from an infectious or traumatic cause are diminishing with time. Consequently, because the incidence of hereditary hearing impairment (HHI) is not known to be decreasing, the relative importance of HHI is increasing. Second, within the last 10 years, approximately 49 genes that cause hereditary impairment have been identified.

Third, as genes that cause hereditary hearing impariment are located and cloned, scientists gain information about the biologic mechanisms that likely will lead to new methods for treating & preventing sensorineural hearing loss.

DEFINITIONS OF HEARING LOSS :

The American National Standard Institute defined hearing loss as the difference from the normal ability to detect sound relative to its established standards. Audiometric zero are those values of hearing level that correspond to the average detection of sound at a range of signal frequencies for eg: 5OOHz, l000Hz, 2000Hz and so forth.

CLASSIFICATIONS OF MAGNITUDE OF HEARING IMPAIRMENT :

dBHL Range	Term
< 26	Normal Hearing
26 - 40	Mild Hearing Impairment
41-55	Moderate Hearing Impairment
56 - 70	Moderately Severe Hearing Impairment
71-90	Severe Hearing Impairment
> 90	Profound Hearing Impairment

* Modified from Lloyd & Kaplan (1978).

Hearing loss may be bilateral or unilateral. Individuals with hearing losses are the mild, moderate, and even severe categories are more likely to be called hard of hearing. Those with profound hearing loss are more likely to be called deaf.

Hearing loss can be congenital, progressive or acquired. Children may be born with hearing loss, either of genetic or non-genetic origin, or may experience a loss over time.

CONGENITAL VERSUS HEREDITARY :

Congenital means present at birth, where as hereditary means caused by the effect of a gene. One of the most common types of hereditary hearing impairment in autosomal dominant non syndromic delayed - onset progressive sensorineural hearing loss.

SYNDROME VERSUS GENETIC DISORDER :

Many syndromes are caused by gene defects and therefore can be inherited. Syndrome literally means running together and some syndromes are inherited where as others are caused by exogeneous factors rather than by an abnormal gene.

A patient diagnosed with one of the hereditary syndromes known to include hearing impariment does not necessarily have to have hearing impairment.

SYNDROMES AND NON SYNDROMIC HEARING LOSS :

70° e of genetically determined hearing impairment occur in non syndromal forms (Gorlin et al., 1994). Non Syndromal Sensorineural Hearing Loss (NSSNHL) represents the most common form of genetic hearing impairment (Kimberling, 1993). While phenotypic expression is very similar, a high degree of genetic hetrogeneity has been recognised.

In both the syndrome & non syndrome classification there are various models of inheritance of the genes responsible for hearing impairment, including autosomal dominant, autosomal recessive, and X-linked transmission.

The diagnosis of non syndromic genetic hearing loss and the distinction from other known causes of hearing loss is not always easy. Auditometric configuration is an important diagnostic consideration in the diagnosis of genetic hearing loss. On previous

studies the audiograms of non syndromic genetic hearing loss have shown that they could be divided into. (Xuezhong et al., 1994)

1. Sloping group (including gently sloping & sharply sloping)
2. A flat group
3. A residual group and
4. A specific group (including ascending and U shaped).

Several different forms of non syndromic genetic hearing loss have been discrimated on the basis of audiogram findings. In general, autosomal dominant (AD) deafness is said to be milder than autosomal recessive (AR).

In the present investigation the audiological results of 20 individuals with hearing loss from 10 families were analysed in order to determine the variation in audiometric shape and the degree of hearing loss between different siblings in the same family and across the families.

PURPOSE OF THE STUDY :

1. To find out the shape and the degree of the hearing loss among two siblings in the same family is same or different.
2. Any common audiological pattern specially exist for differentiating the hearing loss distributed in families with more than one Hearing Impaired child.

REVIEW OF LITERATURE

Concepts of heredity were introduced into the field of hearing disorders during the second half of the 19th century and in the last quarter of the 19th century, heredity was accepted as a cause of hearing impairment (Stephens, 1985, Ruben, 1991, Reardon 1992).

In numerous surveys throughout the world the proportion of genetic cause of hearing impairment in childhood varies. In the survey performed in 1977 on 2,988 subjects living in the member countries of the European community and born in 1967, genetic factors accounted for 9% of the hearing impairment.

Epidemiologically, atleast one thrid of all hearing losses are of genetic origin, moreover, about one half of children with profound hearing loss have an inherited type of hearing loss.

According to Gorlin et aL (1995) there are over 400 types of genetic hearing loss, amongst which 22 are recognised as being transmitted without any accompanying symptoms, the rest being syndromal.

Important factors indicating a genetic etiology are ; a family history, the existence of two or more effected children in a sibship, and anatomical symmetry of the lesions.

SOME GENERAL PRINCIPLES :

Genetic disease occurs when one or a pair of genes is abnormal.

Recessive deafness has been estimated to account for 75-88% of genetic deafness. A trait, such as deafness, is considered recessive if it appears only when both genes of a pair are defective.

The persons who are deaf with a recessive disorder is referred to as a homozygote or as being homozygous for that gene. Persons who have one gene for recessive deafness

and one normal gene are hearing are referred to as heterozygotes or heterozygous carriers of that gene. In some recessive traits, partial or clinical signs may be detected in heterozygotes.

Typically pedigree of recessive deafness show affected individuals in just one sibship (set of children). Each affected child has two genes for deafness at one locus, having received one from each parent.

It is unlikely that two parents carry the same rare recessive gene, unless they are related and have both received the same mutuant gene from a common ancestor. Therefore the hall mark of a rare recessive disorder, such as recessive deafness, is consanguinity.

In a dominant deafness, which has been estimated to comprise 12 to 24% of genetic deafness, the possession of a single abnormal gene is sufficient to produce deafness. In other words, the heterozygotes is deaf. On the average, half the children of a parent with dominant deafness will be affected, irrespective of the hearing status of the marriage partner, thus the recurrence risk is 50%.

Males have one X and one Y chromosome, whereas females have two X chromosomes. If the abnormal gene is on the X chromosome, the syndrome will be seen in males but not in females, for the additional X chromosome in females is sufficient to protect them from the disease. Thus females can transmit a sex-linked disease to about one half of their sons.

The most common form of genetic hearing impairment noted are the autosomal recessive forms accounting for about 80% of cases, autosomal dominant forms

accounting about 13% and X-linked inheritance for 2-3% cases (Fraser, 1976; Rose et al., 1977; Newton 1985; Morton, 1991).

A chinease study has indicated a proportion of 92% of recessive inherited hearing impairment, where as only 5% was caused by dominant traits.

Throughout the history of genetic hearing impairment, it has been learned that pre-lingual hearing loss affects both marriage and reproductions rates and that marriage between deaf persons are frequent (Ruben 1991; Christiansen. 1991: Dolnick. 1993).

The higher prevalence rate of 2.6/1.000 of profound hearing impairment is present in Asian children compared with the prevalence rate of 0.7/1000 in non - Asian children, and that 68% between cousins (Lumb, 1981).

A study demonstrated a prevalence of hearing impairment among consanguinous marriages of 12.9/1000 where as the prevalence in non-consanguinous marriages in the same area was 3.1/1000 (Al Shihabi, 1994).

Two thirds of hereditory hearing impairment (HHI) is non syndromic, and genetic defects resulting in non syndromic hereditary hearing impairment may be passed to affected individuals through several modes of inheritence.

The number of different genetic loci involved in non syndromic recessive hearing loss (NSRHL) has been estimated at between 20 and 150 (VanCamp

et al,1998)

Autosomal dominant modes of inheritance account for 15% of cases non-syndromic hearing loss.

MEASUREMENT OF AUDITORY SENSITIVITY :

During the first few months of life it is difficult to implement any formal behavioural test technique to obtain information regarding thresholds.

Behavioural hearing tests for assessment of hearing sensitivity at this age are therefore generally qualitative and not quantitative based on skilled observation of changes in the baby's behaviour in response to auditory stimulation.

Formal test procedures available from this age, such as distraction tests (Mc Cormik, 1993) or visual reinforced audiometry (VRA-Bomford and Mcsporran, 1993) generally give information in a sound field setting with the sound delivered by loudspeaker.

Interpretation of test results should take into account the fact that progressive improvement in behavioural response sensitivity is observed as a function of increasing age in the first year of life (Northern and Down, 1991).

Objective techniques for measuring auditory sensitivity are the auditory evoked responses, classified in terms of the latency at which the response occurs after the auditory stimuli occurs after the auditor}' stimulus, early middle or late response.

Techniques like auditory brainstem responses (ABR) and electrocochleography (EcochG) give very stable and highly repeatable ear - specific responses regardlers of a child's age (Weber, 1982) and show good agreement with psycho acoustical thresholds at the test frequencies (Hyde et al, 1990; Arstan, Conti and Prosser, 1986).

Tympanometry and middle ear measurement provide, a rapid method for obtaining objective information about the pathological state of the middle ear.

For confirming hearing levels, a battery of tests, with cross-checking of test results is important. Coroboration of results using information gained from the case history,

parental comments, observations of behaviour, otoscopy and imaging of middle and inner ear structures is important.

The audiogram configuration with respect to the positive or negative slope of the air-conduction threshold is classified as flat, gradually sloping, sharply sloping, precipitiously sloping, rising, trough or saucer.

CRITERIA FOR CLASSIFYING AUDIOMETRIC CONFIGURATIONS

TERMS	**DESCRIPTION**
Flat	< 5 dB rise or fall per octave
Gradually sloping	5-12 dB threshold increase per octave
Sharply sloping	15-20dB threshold increase per octave
Precipitously sloping	Flat or gradually sloping
Rising	5 dB or more threshold decrease per octave
Trough	20 dB or greater loss at the mid frequencies than at the extreme frequencies
Notch	Sharp dip at a single frequency with recovery at the immediately adjacent frequencies
Saucer	20 dB or greater loss at the extreme frequencies than at the mid frequencies

Adapted from Cahart (1945) and Llyod and kaplan (1978).

Zakzouk and Bafaqeeh (1996) studied the prevalence rate of hearing impairment among children with siblings and family members having hearing and speech disorders. A random survey of 6,421 Saudi, infants and children between 2 months and 12 years of age was conducted in the Riyadh area. Children with no or few siblings and those who were first born were at relatively higher risk, and there was a strong association between children in the hearing impaired group among those having living siblings with deafness. Those children who had family members with hearing and speech problems were at twice the risk of their counterpart groups regarding hearing impairment.

De-vitto, Corta, Bevilaqua, Passertotti. Richieri and Corta (1997) report on two sisters with cataracts and progressive sensorineural hearing loss, starting in infancy. They were born to consanguineous parents, and there were no similar cases in the family.

According to Zbar, Ramesh, Srisailapathy, Fukushima, Wayne and Smith (1998), hereditary hearing impairment affects approximately 0.05% of all children born in the United States. It is most commonly autosomal recessive, non syndromic and monogenetic (autosomal recessive non syndromic hearing loss (ARKSHL). Although the number of disease loci is not known, some estimates exceed 100. Using a strategy of homozygority mapping to localize ARNSHL genes by screening consanguineous families for chromosomal regions that are homozygous by descent, the anthous have mapped several genes in multiplex, nuclear consomguinous families in Tamil Nadu, India, from the mean frequency of the ARNSHL genes in this population, the total number of disease genes is estimated to be 57.

AUDIOMETRIC PATTERNS OF GENETIC HEARING LOSS :

Discriminating between the different form of hereditary non-syndromal sensorineural hearing loss usually focus on the mode of inheritance, the type and severity of hearing loss and vestibular function.

Two main issues arise from a clinical point of view. The first is whether it is possible to distinguish between a genetic and a non genetic hearing loss. Second is whether the audiometric pattern can differentiate different genotypes.

Albrecht (1922) observed that recessive forms of hearing loss are profound or total, congenital and non-progressive, whereas the dominant forms are usually less severe, post natal and variable progressive.

Langenbeck (1935) was probably the indicator of hereditary and only a few cases have been reported of unilateral SNHL due to genetic cases (Smith, 1939; Everberg 1957). It is well known that the fundamental processes involved in the mechanism of hearing are controlled by hundreds of genes (Nance. 1980).

The majority of authors report that it is impossible to subclassify autosomal recessive SNHL (ARSNHL) by audiometric criteria as there is extreme heterogeneity (and phenotypic variability) in the andiometric profile (Smith et al, 1995).

The autosomal dominant SNHL wide range of andometric patterns which the affected individuals may exhibit.

Two large and well documented multigenerational pedigrees have been reported as showing low frequency hearing loss, which led to the identification of two different genes.

Although the audiometric patterns and age of onset (about age 10 years) appear similar, the progression is different in two families.

It is very rapid in the large Kindred from Costa Rica in which SNHL progresses to severe hearing loss across the entire frequency range by the age of 30, leading to profound flat hearing loss at approximately 40 years of age (Leon et al, 1992).

Four genes involved in high frequency SNHL have been localized. DFNA2 (Couchcke et al., 1994) causes high tone hearing loss and progresses at a highly variable rate to affect all frequencies. The current results involve four large kindreds from different continents (Indonesian, American, Belgin and Dutch) (Van camp et al., 1995).

Mid frequency or flat hearing loss, the andiometric profiles of a family described by Chen (Chen et al., 1995) in which DFAA4 was localized on chromosome 19 q - 13 and of a family reported by Kirschhofer C (Kirschho fer et al., 1995) moderate to severe SNHL involving all the frequency of about 60-70 dBHL.

FAMILY ORIENTED STUDIES ON AUDIOMETRIC PATTERNS :

A study done as boys town research hospital, by Edward et al (1998) found that approximately 50% of childhood nonsyndromic recessive hearing loss is caused by mutations in the connexin-26 (CX26) gene. Hearing loss was examined in 46 individuals from 24 families who were either homozygous or compound heterozygous for CX26 mutations. The criteria for inclusion in the study of subjects required two or more siblings with non-syndromic sensorineural hearing loss with no history of similar hearing loss in their parents, aunts or uncles. Results showed although all persons had hearing impairment, noconsistant andiologic phenotype was observed.

Hearing loss varied from mild-moderate to profound even within group of families homozygous. Further more hearing loss was observed to be progressive in a number of cases.

The importance of CX 26 is underlined by the fact that mutations in it are responsible for 40% of genetic childhood hearing loss.

Reports from several families are there showing the occurrence of low frequency hearing loss. Iinuma et al.. (1967) described two siblings with low frequency sensory neural hearing loss. No other affected family members were mentioned. The authors also mentioned "10 cases of familial occurrence'". They found familial occurrence in brothers or sisters. They concluded that they had too little data to state the type of transmission.

A report from Vander Bilt University (196S) presented two kindreds, thought to be related, with a low frequency neural hearing loss in affected family members. They presented the audiometric findings in 18 cases. 30 to 40 dB hearing loss was found below 2000 Hz, with normal thresholds above this level. All of the audiograms presented were as patients under 30 years of age.

Parving (1984) studied about inherited low frequency hearing loss. 18 patients with low frequency hearing loss (LFHL), 10 males, and 8 females with an average age of 27 years were examined in order to classify their hearing loss.

Based on the test results the patients could be subdivided into two groups.

(Group A : comprising 11 patients with true SN hearing impairment caused by various well-known etiologies and Group B : comprising 7 patients in whom no classifications of LFHL could be made because of conflict results).

In group B family investigations demonstrated that hearing loss could be ascribed to inheritance in 6 of the patients.

The investigations of family members included otoscopy, pure-tone octave and audiometry with routine determination of air bone conduction thresholds impedance audiometer with determination of spatial reflex thresholds and Bekesy audiometry to reveal carries of genes of deafness (Anderson and Wadenberg 1968,1976, Parving 1978).

The criteria used to assess a genetic hearing disorder was identical configuration with more or less pronounced in one/both parents, in one/more siblings, or in offspring of the proband or cousins of the proband.

Low frequency hearing loss is relatively rare and when a hearing disorder in the relative shows an identical configurations and presents simililiar classification problems, it can conclude from this is Low frequency hearing loss is inherited.

LLu.. *and* Xu (1994) reported their study about features of the audiograms of 136 individuals, from 28 families and affected by non syndromic genetic hearing loss. There were 83 (12 families) with autosomal dominant (AD) loss, 50 (15 families) with autosomal recessive (AR) loss, and 3(1 family) with X-linked recessive loss. The main audiogram shapes found were sloping (50.3%), residual (26.5%) and flat (21%). Specific shapes (ascending and U-shaped) only occurred in 3.7% of AD cases. Audiogram shapes were found to be significantly different between Autosomal Dominant and Autosomal Recessive families and showed interfamilial and intrafamilial variability. In the Autosomal Recessive group, the main shapes were residual and sharply sloping and in the Autosomal Dominant group, sharp sloping, flat and gently sloping. There is a significant difference in the degree of hearing loss between Autosomal Dominant and

Autosomal Recessive types, with Autosomal Dominant being milder than Autosomal Recessive. It has been shown that there is more marked intrafamilial variation in the degree of hearing loss in Autosomal Dominant families than in Autosomal Recessive ones. The results suggest that the audiograms of nonsyndromic hearing loss are usually nonspecific.

Fraser (l976) found from his study a variation of severity of hearing loss within and between families. In 5 Autosomal Dominant families examined completely, the degree of hearing loss in affected individuals of 4 families was found to range from slight to profound and the audiogram shapes in these families been shown this range.

Recessive gene expressively been described as varying little within a sibship. (Connor, 1984) This is seen in the small variation in degree of hearing loss found among affected sibs in 3 of Autosomal Recessive families, and the similarity of the audiogram shapes. The results indicate that it would be necessary to undertake careful audiological tests in family members before determining which individuals are affected and predicting their degree of hearing loss.

Some studies have questioned whether a certain type of audiogram is associated with a typical cause of hereditary hearing loss. Analyzing 250 audiograms from 250 cases of congenital hearing loss Fisch, (1955) pointed that an identical audiogram did not necessarily mean the same cause of hearing loss. In nonsyndromic hearing loss the value of the audiogram in relation to not only phenotype definition but also genotype.

Reardon et al.,(l992,) having evaluated the audiograms of affected individuals in a number of families with nonsyndromic X-linked recessive hearing loss found no reliable correlation between audiogram and genotype.

Mengel et al., (1967) studied 10 affected persons in a Mennonite kindred. Family members stated that the affected children had at least some hearing at birth. The children responded to sounds and sometimes learned a few words. Progressive hearing loss occurred fairly rapidly between the ages of one and one and half and six years, with severe hearing loss in all affected persons by six years of age. One child attended public school for several years before hearing loss forced her to a school for the deaf. Audiograms on the 10 available affected family members showed severe to profound sensarineural hearing loss from 60 to 100 dB in all frequencies.

Teig (1968) studying a family with 25 affected persons, found a similar pattern of hearing loss. Follow up studies for as many as 15 years showed progressive deterioration of hearing. Special audiometric tests suggested the possibility of a degenerative lesion in the cochlea.

In the family described by Smith (1939). no cause for the hearing loss was found other than hereditary. The pedigree showed 11 affected persons in 4 generations. Involved were four of seven siblings, their mother, two maternal aunts, 4 their maternal grand father. Only one person had unilateral hearing loss and other members had other hearing impairments.

METHODOLOGY

The methodology for the present study is described under the following headings

1. Subjects
2. Instrumentation
3. Test environment
4 Test procedure

SUBJECTS :

10 families who were having more than one hearing impaired child were taken for the study. Two Hearing Impaired siblings from each family were taken for the study. The subjects were totally 20, both children and adults. Their age ranged from 2 1/2 year to 35 years.

Subjects were taken from families where non syndromic hearing loss was present. A detailed case history including age of onset, and knowledge of pathogens (infections, drugs ... etc) and family history of deafness were obtained.

INSTRUMENTATION :

Madsen OB 822 with TDH-39 earphone lodged in mx-41/AR ear cushions was used for puretone audiometry.

Children who were below 3 years and not co-operative were screened by behavioural observation audiometry conditions (6 children) by a Grason-Stadler-61 audiometer. The output of the audiometer was transmitted through GSI loud speakers.

IMPEDANCE AUDIOMETRY:

Tympanograms curves, acoustic reflex threshold data were obtained using Simens SD-30 inpedance audiometer which has fecilities for complete automatic or manual diagnostic testing of middle ear function.

BSERA : (Brain Stem Evoked Response Audiometry) :

An eletrophysiological unit, Nicolet audiotory evoked potentials was used for ABR testing.

TEST ENVIRONMENT:

The test was conducted in an air conditioned sound treated room. The enviornmental conditions like temperature (85°F) and the humidity conditions were within specified limits. The noise levels were within permissible limits as per ANSI specifications.

TEST PROCEDURE:

During puretone audiometry. air conduction threshold and bone conduction threshold were taken. PTA was calculated by taking the average of thresholds of 500Hz, lOOOHz and 2000Hz frequencies.

Administration of speech audiometry tests were done. Mostly SRT. SDS, SAT/SDT whatever is possible in particular case was obtained.

Routine impedance audiometry measures to find out the type of tymponogram (obtained with 220 Hz probe tone for both ears) and acoustic reflexes were also observed.

ABR administred at a click rate of 30.1/sec, polarity - rarefaction to measure the type and degree of hearing loss.

RESULTS

In the present investigation, audiological results of 20 individuals with hearing loss from 10 families were analysed in order to determine the variation in audiometric shape and the degree of hearing loss between different siblings in the same family and across the family.

A total of 20 siblings, two from each family (10 family) were taken for the study. The age range was from 2 1/2 years to 35 years, subjects taken were with no associated problems. There was no significant history of noise exposure, trauma, or ototoxic drugs or related abnormalities that could be detected in any of these subjects.

The degree of hearing found (PTA value) in the 20 individuals can be seen in Table-1. Among the 20 individuals 6 children were screened by behavioural observation audiometry and their test results were confirmed by BSERA, and PTA was calculated.

SDT value measured for small children *(6* of them) with using speech as a stimuli, through loudspeakers in behavioural audiometry settings. The observed value is taken as SDT for the ease of comparision with siblings SDT value.

SDT value shows that it ranges from 50 to 90 dB across siblings of the present study (Table No. 1).

Distribution of hearing loss between families, is presented in Table No.2. 55% of the studied population is with profound hearing loss, 30% as severe and 15% as moderately severe hearing loss. The destribution of degree of hearing loss is in moderately severe to profound range.

The 40 audiogram shapes have been divided into different categories shows in Table 3. Tables gives the comparision between the audiogram pattern of Elder child and

younger child in each family, (if the children were not co-operative for conditioned esting objective tests were carried out. In case of objective testing, no audiogram pattern s evident (-). Mark indicates audiogram could not be obtained).

The raw data collected from the subjects was tabulated and subjected to statistical inalysis using the software package numerical calculation for social sciences (NCSS).

TABLE - 1

AUDITORY TEST RESULTS

Family SLNo."	PTA (Elder)		PTA (Younger)		SDT (Elder)		SDT (Younger)	
	Right dB	Left dB	Right	Left	Right	Left	Right	Left
Family 1	105	106.6	83	83	80	85	75	75
Family 2	98.3	85	75.3	83	90	70	60	60
Family 3	90	90	90	90	90	90	90	90
Family 4	66	86	56.6	56.6	60	70	50	50
Family 5	86	S3	90	90	80	80	90	90
Family 6	88	88	96.6	100	80	80	80	80
Family 7	105	106.6	111.6	113.3	90	90	90	90
Family 8	88	105	90	90	80	95	80	80
Family 9	63.3	71.6	63.3	68.3	60	70	55	60
Family 10	111.6	110	90	90	85	90	90	90
Mean(X)	90.12	93.18	84.64	86.42	79.5	82	76.0	76.5
S.D	15.9793	12.94242	16.04287	15.63179	11.16791	9.48683	15.59914	14.91643

In case if there was no responses the threshold values were **marked** as **the highest** intensity levels available for testing on the instruments.

TABLE - 2

COMPARISION OF DEGREE OF HEARING LOSS BETWEEN FAMILIES

Degree of Loss (dB HL)	**Number of Ears**	**Percentage**
Mild (26-40)		.
Moderate (41-55)		
Moderately severe (56-70)	**6**	15
Severe (71-90)	**12**	30
Profound > 90	**22**	55
TOTAL	**40**	100

Modified from Llyod and Kaplan (1978).

TABLE -3

DISTRIBUTION OF VARIOUS AUDIOGRAM SHAPES

Comparing Elder Persons and Younger Persons of the Different Families

Family Number	**Elder**		**Younger**	
	Left	**Right**	**Left**	**Right**
Family 1	Flat	Flat		
Family 2	Gradually Sloping	Gradually Sloping	Fait	**Flat**
Family 3				
Family 4	Gradually Sloping	Gradually Sloping	Flat	**Flat**
Family 5	Flat	Flat		
Family 6	Flat	Flat	Gradually Sloping	Gradually **Sloping**
Family 7	Flat	Flat	Flat	**Flat**
Family 8	Gradually Sloping	Gradually Sloping	-	-
Family 9	Flat	Flat	Sharply Sloping	**Sharply Sloping**
Family 10	Flat	Flat		

Criteria for classifying audiometric configuration adapted from Carhart (1945) and Llyod and Kaplan (1978).

TABLE -4

DISTRIBUTION OF VARIOUS AUDIOGRAM SHAPES ACROSS THE FAMILIES

Audiogram shape	Number of Ears	Percentage
Flat	**19**	47.5
Gradually sloping	**7**	17.5
Sharply sloping	**2**	5
Precipitously sloping		
Risina		
Trough	—	
Notch		
Sancer		
Could not define audiogram*	**12**	30
TOTAL	**40**	100

* Could not define - indicates the results of objective hearing evaluation.

Criteria for classifying audiometric configuration adapted from Carhart (1945) and Llyod and Kaplan (1978).

Table 4 : Shows distribution of various audiogram shapes across families. Main audiogram shapes are flat (47.5%) gradually sloping (17.5%) and sharply sloping (5%) could not define properly the audiometric pattern of 30% of the subjects (*). Audiogram shape showed intrafamilial and interfamilial variability-

TABLE -5

COMPARISION OF AUDIOLOGICAL RESULTS OF ELDER AND YOUNGER CHILDREN AS A GROUP

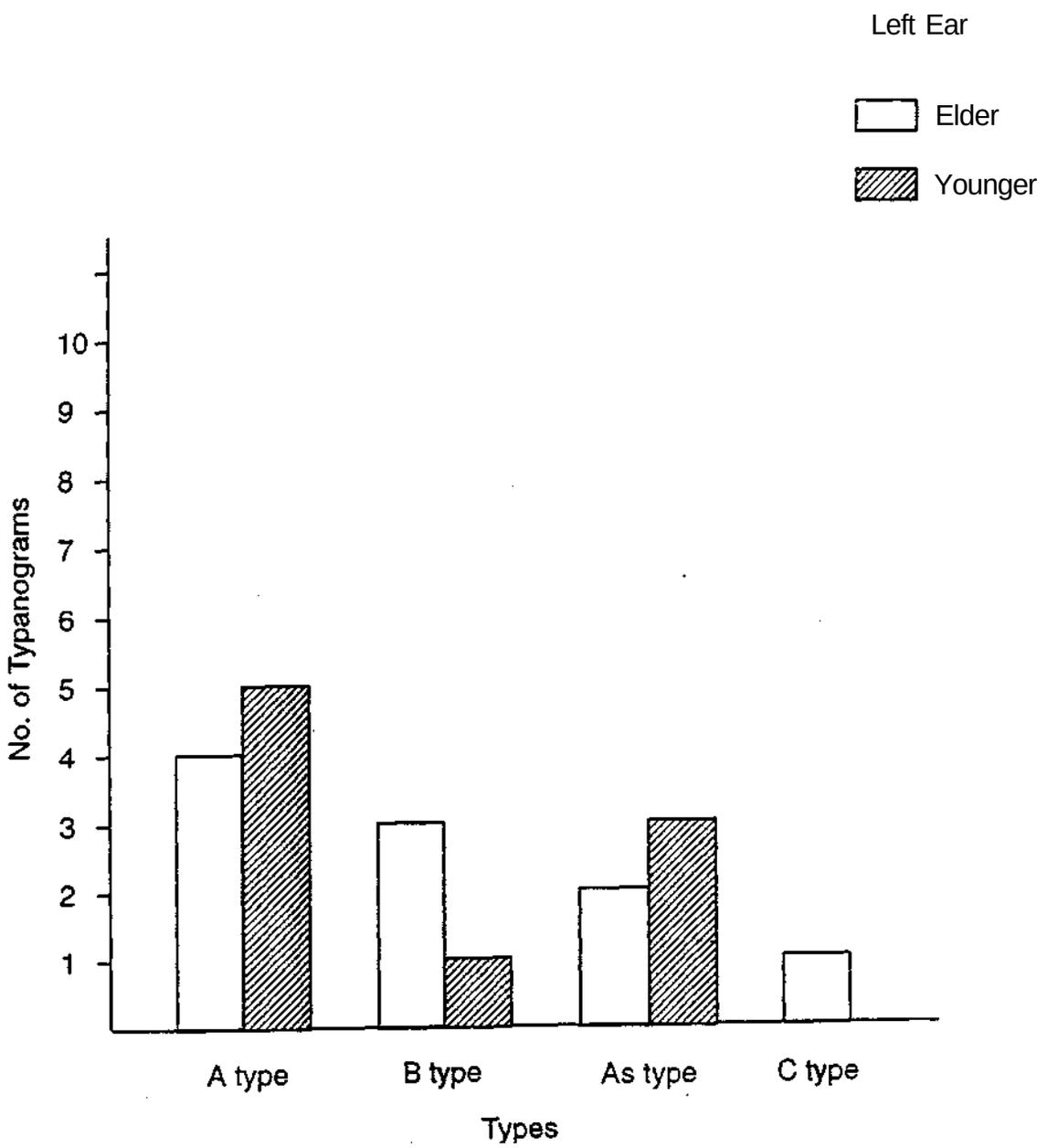
Audiological test results	Ear	Elder			Younger			TTest		
		N	X	S.D	N	X	S.D	Value	df	Sig
PTA	Left	10	93.18	12.94	10	86.42	15.63	1.07	19	p>0.05 NS
	Ridit	10	90.12	15.97	10	84.64	16.04	0.77	-	-
SDT	Left	10	82	9.48	10	76.5	14.91	0.88		
	Right	10	79.5	11.16	10	76	15.59	0.58	-	-

Bv comparing elder siblings as a group younger siblings as a group (Table 5) with using t-test found that there is no significant difference in PTA values and SDT values (P > 0.05). It can be concluded that comparing as a group no significant difference were found in the values of PTA and SDT between elder group of siblings and younger group of siblings.

In impedance audiometry typomograms were shown in the histogram and found that majority of the tymphonogram patterns present are A' type both in elder and younger sibling group (in both ears) (Graph - I &- Il)

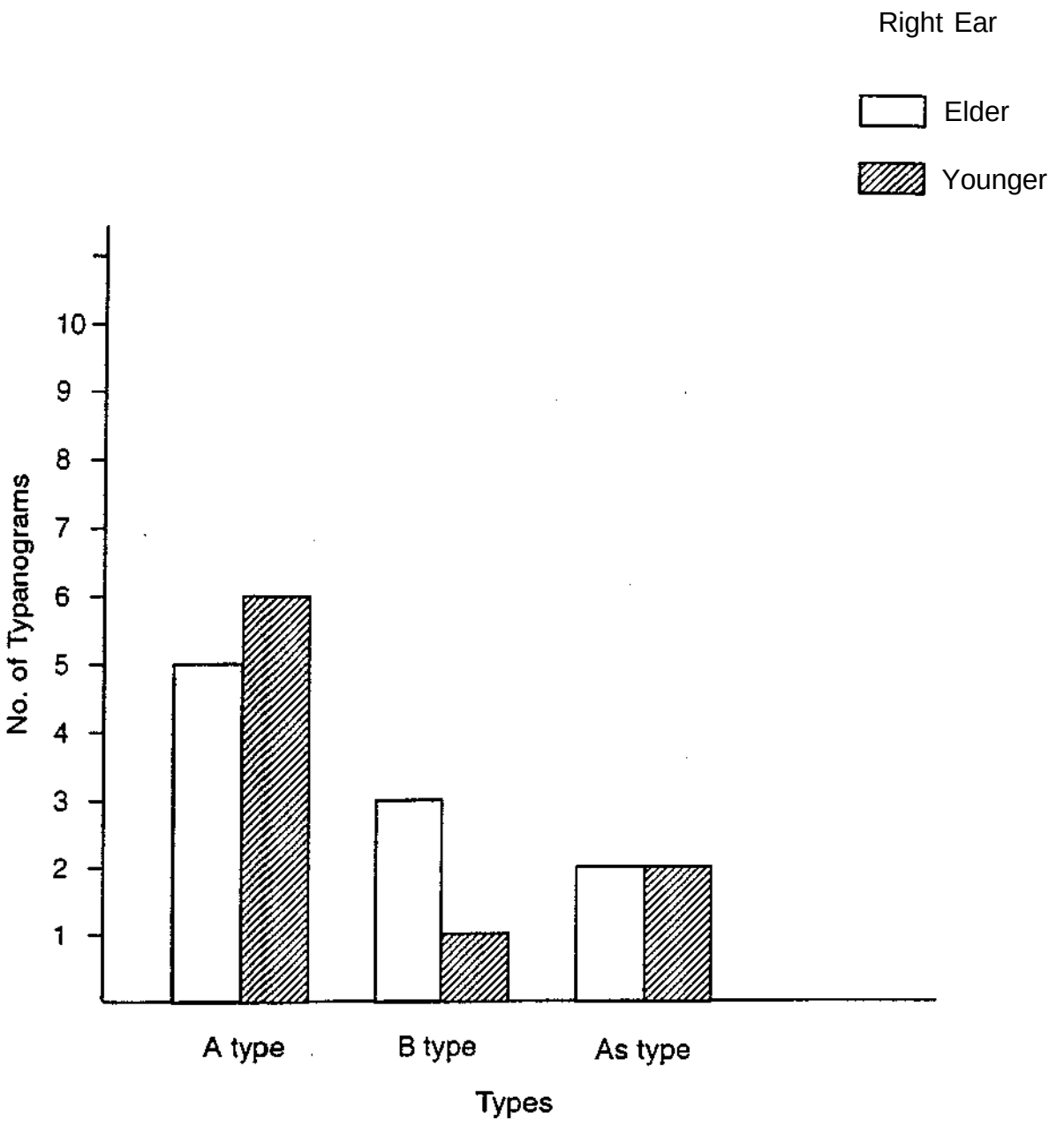
Graph -1

DISTRIBUTION OF TYMPOMOGRAMS AMONG ELDER AND YOUNGER CHILDREN OF FAMILY



Graph - II

DISTRIBUTION OF TYMPOMOGRAMS AMONG ELDER AND YOUNGER CHILDREN OF FAMILY



DISCUSSION

Genetic hearing impairment can be broadly classified into syndrome and non-syndromic hearing impairment. Audiological configuration has been considered to be one feature thai may help in indicating a hearing loss of genetic origin.

In the present investigations, audiological results of 20 individuals with hearing loss from 10 families were analysed in order to determine the variation in audiometric shape and the degree of hearing loss between different siblings in the same family and across the family.

Considering the destribution of degree of hearing loss between families, 55% of them with profound, 30% of which severe and 15% as moderately severe. It ranges from moderately severe to profound loss. This results is in accordance with the study done by Mengal et al., (1967). In his study 10 affected persons in a Mennonite Kindred showed severe to profound semorineural hearing loss from 60 to 100 dB in all frequencies.

Present study is also supported by Fraser G.R's study (1976). He found from his study a variation of severity of hearing loss within and between families. Among the 5 autosomal dominant families examined found degree of hearing loss vary from slight to profound. Audiogram shapes in these families has shown this range in degree of hearing loss. In the present study the degree of hearing loss varied from moderately severe to profound range.

Findings from Connor.J.M. (1984) study supports the present study in stating variations in degree of hearing loss found among affected sibs in the family. His study reports that a small variation in degree of hearing loss among affected sibs in 3 of autosomal recessive families and the similarity of the audiogram shapes.

In the present investigation 40 audiogram shapes were analysed and found flat type (47.5%). Gradually sloping type (17.5%) and sharply sloping (5%) and could not define properly the audiometric pattern of 30% of the subjects. These audiogram pattern are in accordance with the reports of the Chen et al., (1995). Mid frequency or flat hearing loss, the audiometric profiles of the family described by him.

Findings of Paparella et al., (1975) also strengthen the results of the present investigation i.e., a flat audiogram can be autosomal dominant or autosomal recessive inherited. He also suggests that a flat curve with fairly good descrimination was a crucial indication of genetic hearing loss.

Liu. and Xu (1994) reported after studying the audiograms of 136 individuals from 28 families, affected by non syndromic genetic hearing loss. The main audiogram shapes found were sloping (50.3%). residual (26.5%), flat (21.0%), specific shapes (ascending and U shaped in 3.7% autosomal dominant cases). From the present study the flat type found as (47.5%) and gradually sloping type (17.5%) and sharply sloping 5%. The current study audiometric patterns are in consonance with the audiometric patterns of *Liu. and* Xu.'s (1994) investigation.

A study done on Boys town Research Hospital by Edward et al., (1998) supports results of our study that intra familial and interfamilial variation exists in results. Boys town research found that although all subjects had hearing impairment, no consistent audiologic phenotype was observed. In their study also variation in the hearing loss was present. Hearing loss varied from mild -moderate to profound even within group of families.

Although there are reports available whether a certain type of audiogram is associated with a typical cause of hereditary hearing loss. In the present study the causes of hearing loss in most of the cases is unknown. Fisch (1995) analyzing 250 audiogram from 250 cases of congenital hearing loss pointed out that an identical audiogram did not necessarily mean the same cause of hearing loss. Reardon et al., (1992) also stated after evaluating the audiograms of affected individuals in a number of families with non syndromic X-linked recessive hearing loss found no reliable co-relation between audiogram and genotype.

SUMMARY AND CONCLUSION

The present study was undertaken to study the audiometric findings that exist in siblings of the same family and across the families who have hearing loss.

The present study was aimed at:

1. to find out the shape and the degree of the hearing loss among two siblings in the same family.
2. Any common audiological pattern specifically exist for differentiating the hearing loss distributed in families with more than one hearing impaired child.

In the present investigation, audiological results of 20 siblings with hearing loss from 10 families (two from each family) were analysed. The age range was from 2½ years to 35 years. Routine clinical audiological tests were administered from the results it in shows that.

—> Intrafamilial and interfamalial variably in audiological findings present.

—> The degree of the hearing loss ranged from moderately severe to profound ranse. 55% of them showed profound, 30% as severe, 15% as moderately severe, hearing loss.

The shape of the audiogram found were flat (47.5%), gradually sloping (17.5%) sharply sloping (5%). 30% of the audiogram patterns could not be defined properly.

LIMITATIONS OF THE PRESENT STUDY

1. The study was done on a small sample of 10 families, hence the results can not be generalized.
2. Gene mapping for the subjects participated in this study was not done which would have provided information about the genetic factors causing hearing loss.
3. The etiologies for hearing loss was not known properly for all the subjects. No serious investigation was done to probe into the causative factors.

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