



## A Study of Congenital and Early Acquired Impairment of Hearing

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### Abstract

The present study included 261 patients (M:F=1.72:1) suffering from congenital and early acquired hearing loss. The aetiological breakup of the hearing loss was : genetic factors 47.5%, non-genetic factors 16.8%, congenital ear malformations 8.5% and cryptogenic factors in 27.2% cases. Autosomal recessive mode of inheritance was seen most commonly (62%) followed by autosomal dominant (20%) in the genetic group of hearing loss. Maternal Rubella was most common cause of prenatal group of hearing loss while perinataly birth anoxia and prematurity were common. Postnatally meningitis was most common aetiology of hearing loss. Linkage analysis on SLINK 2 point autosomal data yielded LOD score of more than 3 in an autosomal dominant family.

### Key Words

Congenital, Genetic, Non-genetic, Hearing loss.

### Introduction

Impairment of hearing is one of the major disabilities occurring in India, affecting about 1.8% of the population. According to the National Sample Survey (1), the prevalence of hearing disability per 1000 population were 5.53 in rural and 3.90 in urban areas. These rates are higher than those for disability due to loss of vision and locomotor problems.

There is virtually no data regarding prevalence of congenital and early acquired hearing loss in India except few regional surveys. Kameswaran and Rajendra Kumar (ICMR) (2) reported 0.1% of the population to be suffering from congenital sensorineural

hearing loss and 0.02% of urban and rural population suffering from congenital conductive hearing loss in Tamil Nadu. Pat *et. al.* (3) reported 2 per 1000 children under 5 years and 4.2 per 1000 children under 15 years to have bilateral sensorineural hearing loss of worse than 60 dB. This study was undertaken to find out the prevalence of congenital and acquired hearing loss with analysis of various causes for the same.

### Material and Methods

The present study was undertaken at ENT OPD, All India Institute of Medical Sciences, New Delhi to study the main aetiological factors causing congenital and early acquired hearing loss.

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## 1. Patient Selection

- (a) Inclusion criteria : All those patients with prelingually impaired hearing who have no history of environmental factors to explain the hearing loss during the prelingual period were subjected to further detailed work up.
- (b) Exclusion criteria : The following cases were excluded from the study.
  - (i) History of ear infections and trauma.
  - (ii) History of ototoxic drug intake, mumps, measles or meningitis after 6 months of age.
  - (iii) Family history of otospongiosis.

## 2. Work-up of an Individual Case

Detailed work-up of an individual case was carried out including prenatal, perinatal and postnatal history to find out various exogenous congenital causes.

Pedigree chart of the affected child was drawn at least upto three generations and a detailed systemic examination of ophthalmology, cardiovascular system and abdomen was performed to look for associated anomalies. ENT examination with special attention to branchial arch system was performed. Mode of inheritance was ascertained whenever possible as per the criteria used by Rijn and Cremers (4) for acceptance of hereditary and acquired causes.

Special investigations were performed wherever indicated and possible to establish the diagnosis. These included :

1. Antibody titres for toxoplasmosis, rubella, CMV, measles, mumps in cases when suspected and feasible especially for Rubella within 4 years of age (6) and CMV within 3 weeks of life (7).
2. EKG for suspected Jervell and Lange-Neilson syndrome.
3. ERG for suspected Usher's syndrome.

4. Thyroid function test for suspected Pendred's syndrome.
5. Mucopolysaccharide and amino acids screening in the urine in suspected metabolic aetiology.
6. Renal function test for Alport's and other otorenal syndromes.
7. Serum uric acid, pyrophosphates etc to exclude metabolic causes.
8. Chromosomal studies : In the presence of multiple malformations in a case with hearing loss not suspected to be due to a single gene disorder were subjects for chromosomal studies.

## 3. Hearing Evaluation

Included the pure tone audiometry, free field testing (FFT) impedance and acoustic reflex and brainstem evoked response audiometry (BERA).

## 4. Radiology

High resolution CT Scan (1.5 mm sections through temporal bone in 30 degrees axial plane) was done in selected cases particularly where cochlear dysgenesis was suspected.

## Results

The present study group included 261 cases of congenital and early acquired hearing impairment attending ENT outpatients department. All patients with a hearing loss conductive, mixed or sensori-neural, averaging 25 dB HL or more in the better ear by air conduction, within the frequency range of 500-2000 Hz, were included in the study.

## Aetiology

The aetiology of 261 children studied, was classified into four main groups as shown in Table 1. Congenital ear malformations were considered separately because they did not fall into any definitive group (5).

**Table No. 1**  
**Aetiology of Hearing Loss**

Aetiology	No. of Cases	Percentage
<b>1. Genetic</b>	124	47.5
Autosomal Recessive	77	29.5
Autosomal Dominant	26	10.0
X-Recessive	1	0.4
Multifactorial	15	5.7
Chromosomal	2	0.8
Unclassified association	3	1.1
<b>2. Non-Genetic</b>	44	16.8
Prenatal	6	2.3
Perinatal	12	4.6
Neonatal jaundice	3	1.1
Birth anoxia & prematurity	9	3.5
Postnatal	26	9.9
Meningitis	18	6.9
Ototoxicity	5	1.8
Typhoid	2	0.8
Infantile Measles	1	0.4
<b>3. Cryptogenic</b>	71	27.2
<b>4. Congenital Ear Malformation</b>	22	8.5
External ear malformation	19	7.3
Middle ear malformation	3	1.2
<b>Total</b>	<b>261</b>	<b>100.00</b>

1. **Non-genetic group (n=44)** was further sub-classified into 3 main categories depending upon time of exogenous insult to auditory apparatus which may be antenatal, perinatal or immediate post natal period extending till 6 months of age.

(a) **Prenatal group (n=6)** : This included 5 cases of congenital rubella and in one case, the mother had uncontrolled tuberculosis during pregnancy. Serology was possible in only two cases, while in rest of three cases, no serology was advised due to late presentation. No case of congenital CMV could be diagnosed because of later presentation of cases as serology in diagnostic only in first 3 weeks of life (7).

(b) **Perinatal group (n=12)** : It included three cases of neonatal jaundice, nine cases of birth anoxia and prematurity (Birth weight < 1500 gm.) Both the factors i.e. birth anoxia and prematurity, were grouped together because of small sample and both are known to supplement each other in causation of hearing impairment (8).

(c) **Immediate postnatal causes (n=26)** : This group included cases of postnatal causes which were presumed to have acted upon during first 6 months of life. These included one case of infantile measles, two cases of typhoid fever, five cases of ototoxic drug (aminoglycosides) intake and remaining 18 cases with history of neonatal meningitis. The causative organism was tubercular bacilli in 1 patient, pneumococcal in 3 patients, while in remaining 14 patients no definite causative organism was found.

2. **Genetic factors (n=24)** were further subclassified as shown in Table 1.

(a) **Autosomal dominant (n=26)** : It included 4 cases of non-syndromal inheritance and 22 cases with specific syndromes. It included 7 cases of Waardenburg's syndrome, 6 cases of Treacher-collins syndrome, 4 of Alport's syndrome, 2 of Crouzon's syndrome and one with Apert's syndrome. Two patients had associated malformations not fitting into any known syndrome. It included one female patient with associated upper limb malformation and bilateral conductive hearing loss, while in another patient, bilateral conductive hearing impairment was associated with optic atrophy and velopharyngeal incompetence, both showing autosomal dominant pattern of inheritance.

(b) **Autosomal recessive (n=77)** : It included two cases of Usher's Type I syndrome and rest 75 of non-syndrome hearing loss. No case of Pendred's



syndrome or Jervell and Lange Nielson syndrome was detected.

(c) **X-linked inheritance (n=1)** : The present study included one case of Hunter's syndrome with moderately severe mixed hearing impairment.

(d) **Multi Factorial inheritance (Polygenic group) (n=15)** : It included 11 cases of craniofacial microsomia, one of Charge association and 3 patients of Klippel-Feil syndrome.

(e) **Chromosomal group (n=2)** : Included one case of Turner's syndrome (moderately severe conductive hearing impairment) and one case of Down's syndrome with severe sensory hearing loss.

(f) **Unclassified association (n=3)** : It included two female patients with associated congenital heart defects, one had atrial septal defect and pulmonary stenosis with moderate conductive bilateral hearing impairment. The other had ventricular septal defect with severe SNHL and an epileptic focus on EEG. The third case had associated microcephaly, syndactyly and mental retardation with severe hearing impairment.

### 3. Congenital Ear Malformations (n=22) :

It included 19 cases of external ear malformation and 3 cases of isolated middle ear malformations. External ear anomalies included 9 cases with bilateral and 10 with unilateral involvement (7 with right side and 3 with left side involvement). The 3 cases of isolated middle ear malformation were taken up for middle ear exploration and the findings were :

- Body ankylosis of Malleus and Incus in first case.
- Absence of long process of Incus with absent stapes suprastructure and narrow oval window niche in the second case.

- In third case, stapedius tendon was absent with malformed stapedial crura.

### 4. Unknown aetiology (cryptogenic group) (n=71) :

There were 71 cases where the history, laboratory and other evidence could not identify the precise cause of the hearing impairment to any of the preceding aetiological factors. In this group, 5 cases which had more than two possible causes of hearing impairment, were included as per Rijn and Cremer's (4) guidelines, while in rest 66, possible aetiology could be ascribed.

### Sex Ratio

The present study of 261 cases consisted of 165 males (63.2%) and 96 females (36.8%) with male preponderance of 1.72 : 1. Table 2 shows a marked preponderance of males in the postnatal, Mendelian single gene inheritance group and cryptogenic group while preponderance of females in multifactorial and chromosome abnormalities group.

### Audiological Assessment of the Study Group

Two hundred and ten children cooperated for pure tone audiometry. Results from free-field tests supplemented by BERA were available in 44 children. In remaining 7 children, results were inconsistent and till the termination of our study, no consistent response was found even on BERA testing. Four patients had blank audiograms (no response to any frequency). It included 3 cases of genetic deafness (Waardenberg's syndrome = 1 and craniofacial microsomia = 2) and one case of cryptogenic deafness.

Air conduction threshold of these patients were categorised according to severity of hearing impairment

into 5 groups as per Kemker's (9) criteria. The results were as per Table 3. There was an overall male preponderance in all categories of hearing loss. The degree of hearing impairment in each category relative to the aetiology of hearing impairment was observed as shown in Fig. 1. With maximum number of cryptogenic and genetic cases with severe to profound hearing loss.

#### Age of Diagnosis of Hearing impairment :

In the present study, the age of presentation and subsequent diagnosis was variable, the range being from less than 1 month to 22 years (mean age = 6.73 years) as shown in Fig. 2. 40.5% of cases presented before 4 years of age with peak at fourth year (16%). The range of age group when the parents suspected the hearing impairment was right from birth due to presence of congenital external ear

malformations to as late as 16 years (mainly in females of marriageable age desirous of early treatment). In 242 cases the mean was 2.54 years (insufficient data in 19 cases).

#### Linkage Analysis :

18 family pedigrees were studied out of which four were selected for linkage analysis (3 of autosomal recessive and one of autosomal dominant inheritance). Only the pedigree of autosomal dominant inheritance generated LOD (log of odds) score of 3 or greater 72.8% of the times (Fig. 3) while 3 autosomal recessive families were not good enough to generate significant linkage data. This analysis was done at National Institute of Deafness and other Communication Disorders (NIH), USA on SLINK programme for Linkage Analysis (10, 11).

**Table No. 2**  
**Sex distribution in relation to aetiology of hearing impairment**

Sex	Genetic	Non genetic	Cryptogenic	Congenital Ear malformations
Male	73	29	45	18
Female	51	15	26	4
MF Ratio	1.43 : 1	1.93 : 1	1.73 : 1	4.5 : 1

#### In Genetic Group :

Sex	Autosomal Dominant	Autosomal Recessive	Multifactorial	X-linked	Chromosomal	Unknown association
Male	19	48	4	1	-	1
Female	7	29	11	-	2	2
MF Ratio	2.71 : 1	2.52 : 1	0.36 : 1	-	-	0.5 : 1

#### In non-genetic group :

	Prenatal	Perinatal	Postnatal
Male	3	6	20
Female	3	6	6
MF Ratio	1 : 1	1 : 1	3.3 : 1

Table No. 3

Audiological assessment (Kemker's Criteria)

		Number	Male : Female	
Mild	25-40 dB	Nil	-	-
Moderate	41-55 dB	15	9	6
Moderately-severe	56-70 dB	30	19	11
Severe	71-90 dB	84	50	34
Profound	> 90 dB	125	84	41
Inconsistent	-	7	3	4

Number of children of each sex in hearing impairment category

Sex	< 70 db HL	70 db HL
Male	28	134
Female	17	75
MF Ratio	1.65 : 1	1.78 : 1

(\* Results in 3 Males and 4 Females were inconsistent)

Fig. 1 : Degree of Hearing Loss (n = 261)

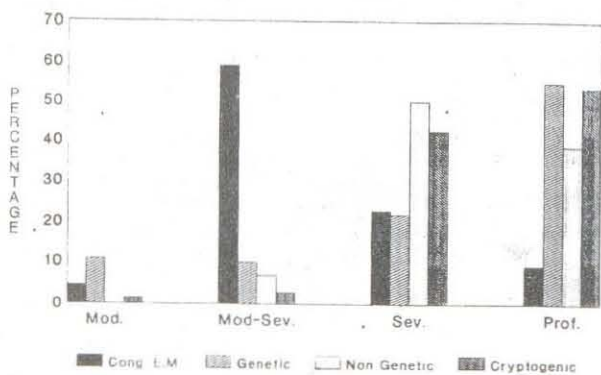


Fig 2 : Age of Diagnosis of Hearing Loss (n = 261)

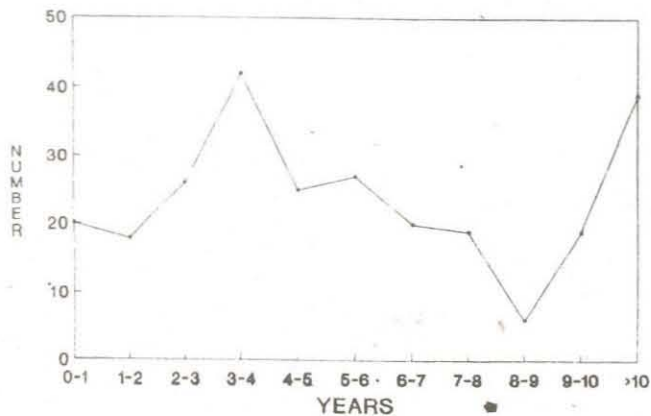
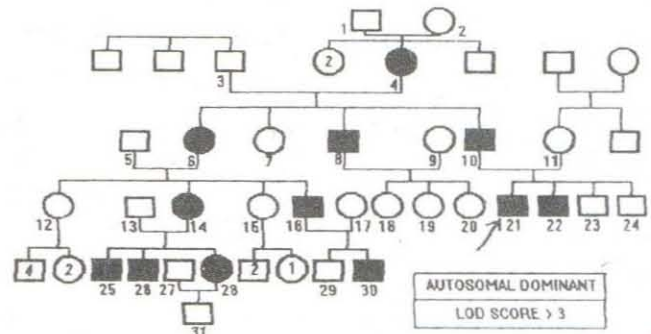


Fig. 3



Discussion

Male Preponderance in Sex Ratio

There is an overall preponderance of males in the present sample which is consistent with the literature (12-14). Male children appear to be more susceptible to adverse factors acting in the prenatal, natal and post-natal life, but the reason for this has not been proved satisfactorily. The present study had female preponderance in chromosomal and multifactorial inheritance which has been well documented by Newton (13) and Das (14).

Age of Diagnosis of Deafness

In the present study, the age of presentation and subsequent diagnosis is quiet late (6.73 year) as compared to western literature (13-15). The part of delayed diagnosis rests upon community practices for delayed speech and the social awareness and partly due to absence of any active health surveillance in this aspect in many places and absence of any high risk registry.

Consanguinity

In the present study, 10 families were picked up, who had consanguineous marriage within three generations. Eighteen children born to these patients were included in the study accounting for 6.9% of all cases. In the literature, the incidence reported ranges from 5% to 9% (13, 16). Consanguinity in India is seen mainly in South India, 60 to 70% as reported by Ganga et. al. (17)

and Rajinder Kumar (18) and in Muslims. Our study sample mainly had cases from North and West India thereby it had relatively small sample of consanguinity.

## Aetiology

### (a) Genetic Group

The modes of inherited deafness in the present study are comparable to the various studies in the literature like Rijn and Cremers (4), Elango (7), Feinmesser (16) and Fraser (19), all revealing predominance of autosomal recessive inheritance except studies by Parving (20) and Ruben and Ruzycski (21) with significant autosomal dominant inheritance.

### (b) Non-Genetic Group (n=44)

**1. Prenatal group (n=6) :** There were five cases of congenital rubella in the present study. In the Western literature, it varies from < 7% in non-epidemic years (19) to about 60% in epidemic years (22).

**2. Perinatal causes (n=12) :** (a) Neonatal jaundice (n=3, 1.15%) : The proportion of neonatal jaundice in the present study was very small as compared to the western literature which varies from 4.5% to 15% (13, 14, 16, 23, 24).

(b) Birth anoxia and prematurity (n=9) : There were nine cases (3.5%) of birth anoxia and prematurity group. In the literature, various studies have shown different figures varying from 1% to 15% (5-7, 16, 25).

**3. Postnatal causes (n=26) :** The present study revealed meningitis as most common cause of postnatal hearing loss which is well documented in the literature by various authors (4, 7, 12, 13, 15).

In the literature the percentage of non-genetic group varies from 17% to 75% (maximum studies have quoted 30% to 40%) (5, 13, 16, 26). In this study the relatively few cases of nongenetic congenital and early acquired group (16.8%) as compared to genetic and cryptogenic group is not in agreement with the literature. This may be due to inclusion of the congenital and early acquired group (within 6 months postnatal) excluding

all other acquired causes in this study. Also other possible explanation in the referral system where multiple handicapped deaf children are referred directly to rehabilitation departments and special clinics for mentally retarded. Also due to lack of "High Risk Registry", the cases are not referred for subsequent screening and management. Due to lack of good infrastructure at peripheral level, increased mortality in acquired group results in less number of survivals to present subsequently seeking advice for deafness. Also large number of studies in the literature have included mainly the postlingual deafness and therefore the subsequent increase in the percentage of nongenetic acquired causes as compared to the present study.

### Cryptogenic Group (unknown cause) (n=71) (27.2%)

In various studies in literature, the cryptogenic group has been reported from as low as 10% to about 60% (average 40%) which is well in accordance with the present study (5, 13, 15, 26, 27).

### Congenital Ear Malformations

The present study included 22 (8.5%) cases of external ear malformations with 19 cases of external auditory canal atresia and three cases of isolated congenital middle ear pathology which is different from the figures reported in literature varying from 2% to 4% (19, 23, 28, 29).

### Conclusion

In this study, the mean age of presentation and diagnosis of hearing loss was 6.73 years. The genetic aetiology was found to be most common cause (47.5%) followed by cryptogenic (27.2%) and non-genetic (16.8%) of the congenital and early acquired deafness. Autosomal recessive mode of inheritance was seen most commonly (62%) followed by autosomal dominant (20%) in the genetic group. Maternal rubella infection was the most common cause of prenatal deafness while perinatally birth anoxia and prematurity were common. Post natively the meningitis and ototoxicity were more common.



The present study being a preliminary study conducted at a tertiary centre, has its own bias, merits and demerits but does reflect the need of 'High risk' registry maintenance at all the peripheral health centres with active surveillance and subsequent screening for early detection of hearing loss.

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