PREVALENCE AND CHARACTERISTICS OF CONGENITAL HEARING LOSS IN CHILDREN – A REGISTER BASED STUDY

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September 2021

CERTIFICATE

This is to certify that this dissertation entitled **'Prevalence and Characteristics of Congenital Hearing Loss in Children – A Register Based Study'** is the bonafide work submitted as part for the fulfilment for the degree of Master of Science (Audiology) of the student Registration Number: 19AUD016. This has been carried out under the guidance of the faculty of this institute and has not been submitted earlier to any other University for the award of any other Diploma or Degree.

Mysuru

September 2021

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DECLARATION

This is to certify that this dissertation entitled **'Prevalence and Characteristics of Congenital Hearing Loss in Children – A Register Based Study'** is the result of my own study under the guidance of Dr. Mamatha N.M., Assistant Professor, Department of Audiology, All India Institute of Speech and Hearing, Mysore and has not been submitted earlier to any other University for the award of any other Diploma or Degree.

Mysuru **Registration Number: 19AUD016**

September 2021

Dedicated

To

God

My beloved family

And

Teachers

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Abstract

Congenital hearing loss is present at birth or soon after birth due to various reasons such as genetic or environmental factors during the prenatal or perinatal period. Congenital hearing loss can be of any type or degree based on the etiology affecting the outer, middle, and/or inner ear. Due to limited prevalence studies showing the distribution of various degrees of hearing loss in children having congenital hearing loss, the current study is planned to provide information about the recent prevalence and characteristics of congenital hearing loss reported to All India Institute of Speech and Hearing (AIISH) in 2019. A retrospective study was done in a total of 1217 pediatric cases with age less than 15 years were reported to AIISH audiology OPD for audiological evaluation. Out of 1217 children reported to AIISH in 2019, 823(67.2%) children were reported to have congenital hearing loss. The degree of hearing loss in these 823 children ranged from mild to profound. The congenital hearing loss was more prevalent in male children (54.4%, n=448) compared to females (46.6%, n=375) in the study period. It was found that profound hearing loss was most prevalent in children with congenital hearing loss in both the ears followed by severe, moderately severe, moderate and mild hearing loss. More children with congenital hearing loss were in the age range of 5-10 and 3-5 years. Consanguinity was one of the most reported risk factors. There was a significant association between gender of children and birth complications and risk factors in occurrence of congenital hearing loss. Associated co-morbid conditions with congenital hearing loss were intellectual disability, chronic heart disease, cerebral palsy, downs syndrome, seizure disorder, microcephaly, visual impairment, and autism. Delayed speech and language development was the main complaint followed by misarticulation and poorer academic performance in children with congenital hearing loss.

Keywords: Children, congenital, hearing loss, prevalence, birth complications

Chapter 1

INTRODUCTION

Hearing loss refers to the loss or reduced ability of an individual in perceiving sounds that is caused due to abnormalities in the anatomy or physiology of the outer ear, middle ear, or inner ear (Lasak et al., 2014). The hearing loss can be of a congenital or acquired type and congenital hearing loss is present at or acquired soon after birth. Congenital hearing loss can be caused by hereditary and non-hereditary genetic factors or certain complications during pregnancy and childbirth (WHO, 2020). The acquired causes of hearing loss include middle ear infections, head trauma, viral infections, noise exposures, and ototoxic medications (WHO, 2020). It has been found that the cause of hearing loss in 50% of infants who are suffering from congenital hearing loss was associated with environmental factors and the other 50% were due to genetic abnormalities (Ahmed et al., 2018).

The genetic causes of congenital hearing loss include mutations or hereditary, including autosomal recessive, autosomal dominant, or X-linked. Most of those with genetic factors are non-syndromic of about 70% and syndromic of about 30% (Gorlin et al., 1995). The environmental factors, including prenatal TORCH (toxoplasmosis, rubella, cytomegalovirus, and herpes) infections, perinatal birth asphyxia, hypoxia, birth complications, postnatal meningitis and hyperbilirubinemia are found to be the manifesting factors of congenital hearing loss (Shearer et al., 1993). Other environmental factors include ototoxic agents, prematurity, sepsis, craniofacial anomalies, low birth weight, Rh incompatibility, extracorporeal membrane oxygenation, and noise exposure (Kral & O'Donoghue, 2010).

The attempt to diagnose the hearing impairment in infants and children goes back to 1964 where Marion Downs reported the incidence of hearing loss as 1 in 1000 infants using Behavioral Observation Audiometry (Downs & Sterritt, 1964). Marion, in 1969 led efforts to form the Joint Committee on Infant Hearing (JCIH) to provide multi-disciplinary efforts and guidance for addressing newborn and infant hearing issues (Finitzo et al., 2000). JCIH was established in 1969, emphasized the mass screening of infants to detect and identify childhood hearing loss as early as possible, and introduced the high-risk register in 1972 and published (JCIH, 1973). North America in 1994 started newborn hearing screening programs, including programs from the Centres for Disease Control and Prevention (CDC) and Early Hearing Detection and Intervention (EHDI) (JCIH, 1995). European countries followed the EDHI program and started screening newborns after the National Institute of Health consensus statement (NIH, 1973). Universal Newborn hearing screening (UNHS) in 1994 has been launched by CDC and EDHI using automated auditory brainstem evoked response and otoacoustic emissions for newborns (JCIH, 1995). World health assembly in 1995 highlighted the need for hearing screening and prevention of hearing impairment in member countries (WHO, 2009). The Government of India launched the National Programme for Prevention and Control of Deafness (NPPCD) in 2007 to decrease the burden of deafness and to work in preventing future hearing loss in India (NPPCD, 2016).

The incidence of congenital hearing loss is reported to be 1 to 3 per 1,000 live births, where a minimum of 3 children out of 1000 was found to have a permanent hearing loss of greater than 20 dB, and 1 in 1000 infants has severe to profound hearing loss bilaterally (greater than 70 dB HL) (Gettelfinger & Dahl, 2018). Also, hearing loss is noted to be 10-times higher in infants having one or more risk factors

associated with hearing loss compared to those without having any risk factors (Gettelfinger & Dahl, 2018).

1.1 Need of the study

World Health Organization (WHO) has reported that out of 466 million estimated with disabling hearing loss, 7% were children (WHO, 2018). The prevalence of hearing loss in children reported by WHO in 2018 is mentioned in Table 1.1.

Table 1.1

Prevalence of disabling hearing loss in children across the world

Selected Regions	**Disabling hearing loss in children(hearing loss>30dBHL in better ear)**	
	Millions	**Prevalence %**
High Income	0.8	0.5
Central/Eastern Europe and Central Asia	1.2	1.5
Sub Saharan Africa	8.9	1.9
The Middle East and North Africa	1.4	0.9
South Asia	12.2	2.4
Asia Pacific	3.6	2.0
Latin America and the Caribbean	2.6	1.6
East Asia	3.3	1.3
World	**34.1**	**1.7**

Permanent hearing loss (PHL) is one of the most prevalent congenital diseases, with an estimated prevalence rate of 2-4 per 1000 infants evaluated. The prevalence of permanent congenital hearing loss is higher in those admitted to NICU

compared to healthy newborns in very highly developed countries (Butcher et al., 2019; UNDP, 2015). The prevalence of sensorineural hearing loss (SNHL) is reported to be 2-3 per 1000 live births in India, and out of 1000 babies, one is found to have profound hearing loss at birth or in the pre-lingual age group (Shrivastava & Gupta, 2018). It has been reported that the prevalence of overall hearing loss among infants was 2.1 per 1000, and for early-onset, at least moderate SNHL was 1.2 per 1000 live births (Dietz et al., 2009). Four in every 1000 children suffer from severe to profound hearing loss (Varshney, 2016), whereas the prevalence of childhood-onset hearing loss is 2% (Garg et al., 2009). Department of Prevention of Communication disorders (POCD) All India Institute of Speech & Hearing (AIISH), Mysuru screened 22,450 newborns in 2014-2015, out of those reported for detailed evaluation from refereed cases 32.9% were identified with permanent congenital hearing loss (AIISH, 2015). Late identification of hearing loss is found to cause delayed speech-language development and has a detrimental impact on literacy, affecting social and academic performance (Wood et al., 2015).

Congenital hearing loss affects a child's ability to acquire spoken communication skills and impair speech-language, cognitive and psychological development (Mace et al., 1991). Early identification of hearing loss, degree, and type of hearing loss highlights the probable effects on speech-language acquisition and appropriate early intervention to minimize the effects and maximize the speech-language, cognitive and psychological development (Mace et al., 1991). Many studies show the prevalence of hearing loss in children, while the number of studies showing various degrees of hearing loss is limited. The Government of India launched the National Programme for Prevention and Control of Deafness (NPPCD) in 2007 to reduce the burden of deafness and prevention of future occurring hearing loss in India

(NPPCD, 2018). The several awaressness programs conducted from AIISH would have increased the number of cases who have visited AIISH for assessment of hearing abilities. The department of POCD, AIISH has been conducting several awareness activities such as public education regarding awareness among public, community health workers, parents, teachers on early signs and symptoms of hearing loss and other communication disorders to facilitate early identification. The routine activities of department of POCD includes newborn screening services, preschool screening services and camps for awareness, prevention and identification of hearing loss on prevention and identification of hearing loss at AIISH as well as outside AIISH, which has been increased tremendously. Hence the current study is planned to provide information about the recent prevalence of congenital hearing loss reported to AIISH in 2019. The study will also help to bridge the research gap that exists due to limited prevalence studies showing the distribution of various degrees of hearing loss in children. The results obtained from the current study will help to better plan the awareness and intervention programs for hearing impaired children.

1.2 Aim of the study

The primary aim of the study is to study the prevalence and characteristics of congenital hearing loss in children.

1.3 Objectives of the study

- To determine the total number of pediatric cases reported at All India Institute of Speech and Hearing (AIISH) for a hearing assessment in the year 2019.
- To estimate the prevalence of congenital hearing loss in children.
- To estimate the gender predominance in congenital hearing loss in children.

- To find the prevalence of different degrees of congenital hearing loss in children.
- To report the risk factors and associated conditions in children having congenital hearing loss reported to AIISH in the year 2019.

Chapter 2

REVIEW OF LITERATURE

Congenital hearing loss is defined as the hearing loss present at birth or soon after birth due to various reasons such as genetic or environmental factors during the prenatal or perinatal period (WHO, 2020). Congenital and early-onset hearing loss is defined by Joint Committee on Infant Hearing (JCIH) in 2007 as the presence of hearing loss during birth or not more than three years of life as some of the causative factors for congenital hearing loss cause delayed onset like congenital rubella (Busa et al., 2007). Congenital hearing loss can be of any type or degree based on the etiology and factors and how these factors affect the outer, middle, and/or inner ear. The impact of childhood hearing loss is found to be very profound not only for the acquisition of speech and language development but also found to affect psychological, social, and quality of life (Yoshinaga-Itano et al., 1998).

2.1 Causes and Risk factors for hearing loss

Environmental and genetic factors are found to share 50-50% as a cause for congenital hearing loss (Ahmed et al., 2018). Genetic hearing loss comprises of non-syndromic of about 70% and syndromic of about 30% causes (Gorlin et al., 1995). Autosomal recessive hearing loss is typical of the congenital type and accounts for the most frequent cause for congenital severe hearing loss among non-syndromic hearing losses, while autosomal-dominant can be manifested as late-onset hearing loss. X-linked and mitochondrial hearing loss is found to be rare (Smith et al., 2005). Autosomal recessive, autosomal dominant, and X-linked are inherited as 75%, 22%, and 3%, respectively, for non-syndromic hearing loss cases (Hegarty, 2005).

The associated “Deafness” genes are DFN A, DFN B, and DFN for autosomal dominant, autosomal recessive, and X-linked genes, respectively (Hegarty, 2005). The mutation is observed in the gap junction protein beta 2 gene (GJB2), which is reported to be the leading cause of non-syndromic autosomal recessive hearing loss. However, still, there is heterogeneity in causative genes causing severe to profound hearing loss (Zelante et al., 1997). Pathological variants in Gap junction beta-2 protein (GJB2) and otoferlin (OTOF) have been reported to be present in 50% of cases of profound hearing loss with an autosomal recessive trait of non-syndromic type (Shearer & Smith, 1993; Snoeckx et al., 2005).

While syndromic hearing loss also can be categorized with the mode of inheritance as non-syndromic, including autosomal recessive (A.R.), autosomal dominant (A.D.), or X-linked (Gettelfinger & Dahl, 2018). The primary syndromes that have been reported to be associated with congenital syndromic hearing loss are Usher syndrome, Pendred syndrome, and Jervell and Lange-Nielsen syndrome. Usher syndrome is the most common form of autosomal recessive sensorineural hearing loss (SNHL), affecting both vision and hearing (Allen & Goldman, 2020). Primary features of Pendered syndrome include sensorineural hearing loss, retinitis pigmentosa. Pendered syndrome is associated with iodine abnormality in the body due to mutation in the PDS gene.

The clinical features of pendered syndrome are euthryoid goiter, high-frequency slopping hearing loss, and affected vestibular aqueduct (Allen & Goldman, 2020). Jervell and Lange-Nielsen syndrome is associated with SNHL along with cardiac abnormalities. Waardenburg syndrome, branchio-oto-renal syndrome, Stickler syndrome (Melnick-Fraser syndrome), Treacher Collins syndrome, neurofibromatosis type 2, and osteogenesis imperfecta have also been reported to be associated with

hearing loss (Allen & Goldman, 2020). Craniofacial anomalies including cleft lip and palate, submucous cleft, pre auricle tags and pits, branchial cysts, widened nasal bridge, low set ears, dystopia canthorum, heterochromia iridis indicate the association of syndromes associated with congenital hearing loss (Cone-Wesson et al., 2000). There are various factors genetic, hereditary, and pre natal, perinatial factors associated with congenital hearing loss.

A family history of hearing loss, visual abnormalities, and congenital heart disease are all important factors to consider as risk for hearing loss. Other important information that needs to be obtained includes a prenatal history including TORCH infections (toxoplasmosis, rubella, cytomegalovirus, and herpes), gestational diabetes, hypothyroidism, and maternal drug, alcohol, and tobacco use during pregnancy. The various risk factors associated with congenital hearing loss include a family history of hereditary sensorineural hearing loss, craniofacial abnormalities, and bacterial meningitis (Ahmed et al., 2018).

Prenatal TORCH infections, perinatal birth asphyxia, hypoxia, birth complications, and postnatal meningitis, hyperbilirubinemia are the environmental factors that are found to be manifested for the cause of congenital hearing loss (Shearer et al., 1993). Other environmental factors include ototoxic agents, prematurity, sepsis, craniofacial anomalies, low birth weight, Rh incompatibility, extracorporeal membrane oxygenation, and noise exposure (Kral & O'Donoghue, 2010). The above mentioned complications during birth can increase the risk of hearing loss.

A newborn baby is tested for "Appearance, Pulse, Grimace, Activity, and Respiration," also known as APGAR, soon after birth to check the baby's health

status, and each is scored on a scale of 0 to 2, with 2 being the best score. Low APGAR score of 0-4 at 1 minute or 0-6 at 5 minutes, or five or more days of mechanical ventilation and hyperbilirubinemia are also among the risk factors for hearing loss. Other risk factors include significant head trauma and chemotherapy, ototoxic medications, TORCH infections, and other viral infections (JCIH, 2019). It has also been found that there is a significantly increased number of cases with low socio-economic status as the risk factors for childhood sensorineural hearing loss (Sutton & Rowe, 1997).

A retrospective study was done in Brazil, including 70 infants in 2016, reported that the congenital Zika Virus infection can also cause hearing loss and highlighted that these populations might pass during screening tests because the onset of hearing loss may be delayed (Leal et al., 2016). A case series study in 340 newborns in Romania has highlighted low birth weight, head circumference, gestational age, ototoxic medication, NICU stay perinatal infections, and hypoxia, including mechanical ventilation, are risk factors for causing hearing loss. Even though the APGAR score is not directly associated, authors have highlighted that newborns with low APGAR scores should be considered for newborn screening for hearing loss (Balázs & Neagoș, 2017).

2.2 Prevalence of Congenital Hearing Loss

The prevalence of permanent congenital hearing loss is higher in those admitted to NICU than healthy newborns in very highly developed countries (Butcher et al., 2019; UNDP, 2015). It has been reported that the prevalence of overall hearing loss among infants was 2.1 per 1000, and for early-onset, at least moderate sensorineural hearing loss was 1.2 per 1000 live births (Dietz et al., 2009).

It has been stated that hearing loss is the second most prevalent cause of impairment and a common cause of sensory deficit (National Sample Survey Organization, 2003). The report on status of disability in India in 2002 reported that in 0-4 year age group highest prevalence is recorded in West Bengal (l1.28 per 1000) followed by Karnataka (6.66 per 1000) and Bihar (4.06 per 1000). A crude estimate of the population suffering from hearing impairment in India is 0.3 million in the 0-4 year age group and 1.5 million in the 5-12 year age group as per India: Human Development Report 1999 (Planning Department, 1999). In India, the prevalence of sensorineural hearing loss is about 2-3 per 1000 live births. Among them, one has a profound degree of hearing loss at birth or in the prelingual age group (Shrivastava & Gupta, 2018). It has been reported that 4 in every 1000 children have severe to profound hearing loss, and every year more than 100,000 babies are found to have been born with hearing loss (Suneela et al., 2009) which was similar to the study by Varshney (2016).

In contrast, the prevalence of childhood-onset hearing loss is 2% (Garg et al., 2009). It has also been reported that when unilateral permanent hearing loss is included, the incidence increases to 8 per 1000 live births (Jewel et al., 2013). Studies in India have estimated neonatal hearing loss to varying between 1 and 8 per 1000 babies screened (John et al., 2009). A prospective one-year study by James et al. (2018) in Govt. T.D. Medical College, Alappuzha showed prevalence of 1.3 per 1000 population where the risk factors reported were prematurity, perinatal asphyxia, hyperbilirubinemia requiring phototherapy, congenital infection, family history of deafness, NICU admission more than five days, culture-positive sepsis, and newborns receiving ototoxic medications.

In the Cochin , at least 1% of the screened high-risk newborns are detected to have hearing loss, which correlates with other studies in the country, highlighting the need for Universal Newborn Hearing Screening to be made as national practice (Paul, 2011). The study by Ahmed et al. in 2018 in neonates reported 1.3% cases of bilateral congenital hearing loss in neonates, which is very high compared to previous studies around the world (Ahmed et al., 2018). Regarding the sex distribution of permanent congenital hearing loss, it was found that about 44% of males and 57% of females were affected. (Berninger & Westling, 2011; Uus & Bamford, 2006). Verma (1992) reports that the incidence of hearing impairment in non-consanguineous marriages is 3.1/1000, whereas the incidence of hearing impairment in consanguineous marriage is 12.9/1000 (4 times higher) (Verma et al., 1992).

Vignesh et al. (2015) reported the prevalence of 1.42 per 1000 babies for congenital bilateral hearing losses in Chennai using a two-step hearing screening protocol with Distortion Product Otoacoustic Emissions (DPOAE) and Automated Auditory Brainstem Response (AABR) for well-born and high-risk newborns, which is comparable to the previous studies in India (Jewel et al., 2013). The prevalence of hearing loss has been reported to vary depending on the protocol utilised in various studies (Dhawan & Mathur, 2007; Suppiej et al., 2007). The major classifications are degree and laterality (White & Muñoz, 2008) and types like conductive and sensory. Some authors also used to classify as degree, type, and neural, including Auditory Neuropathy Spectrum Disorder (Dhawan & Mathur, 2007; Suppiej et al., 2007) and even late-onset hearing loss of progressive type (White & Muñoz, 2008).

The incidence of congenital hearing loss is as low as 1.43% in neonates with a family history of hearing loss. However, a positive family history of hearing loss is considered one of the risk factors (Driscoll et al., 2015). Van Dommelen and

colleagues in 2015 reported prevalence of hearing loss increases as 1.2-7.5% in premature babies born at 24 -32 weeks. Birth weight at birth is also one of the conditions where hearing loss has been reported, and prevalence is higher as and 1.4%–4.8% in babies weighing 750–1500g) in the same study. Admission to the Neonatal Intensive Care Unit for preterm infants is also considered the risk factor of hearing loss (Van Dommelen et al., 2015).

The association between consanguinity and autosomal recessive disorders has been highlighted in previous studies in Saudi Arabia (Bayoumi & Yardumian, 2006; S. Zakzouk et al., 1993). Consanguineous parents are more likely to be homozygous for the same trait, and therefore, increase the chance of having a child affected by congenital deafness. The risk of having more than one child with SNHL was 3.5 times higher in consanguineous marriages (Almazroua et al., 2020; Sanyelbhaa et al., 2017). In a study to explore the relationship between consanguinity and neurosensory deafness in that was carried out in Hyderabaad (Reddy et al., 2006) reported significantly higher percentage of consanguinity among the parents of children with hearing impairment.

Additionally authors have discussed the high percentage of children with amino acid disorders, congenital anomalies and genetic diseases which indicates consanguinity as one of the high risk factors for congenital hearing loss (Reddy et al., 2006). In children with congenital cytomegalovirus (CMV) prevalence of hearing impairment is found to be between 2% and 18% (Das, 1996; Fowler et al., 2017; Goderis et al., 2014). Fowler, in 2017 reported that congenital CMV infection causes approximately 10–21 % of all congenital hearing loss.

2.3 Audiological findings in congenital hearing loss

Hearing impairment associated with syndromes can be highly varied, ranging from surgically and medically treatable conductive deficits to sensory hearing loss requiring amplification devices to treat central auditory dysfunction with or without a peripheral component. Middle ear disease, and conductive hearing impairment, appear to be especially prevalent (Hall III et al., 1995).

Among the autosomal dominant syndromic hearing loss, Wardenburg syndrome is most common, consisting of various degrees of non-progressive pre-lingual SNHL (Kochhar et al., 2007). Likewise, Branchio-Oto-Renal (BOR) syndrome is reported to result in conductive, sensorineural, or mixed hearing loss along with malformations of the external ear, branchial cleft cysts, or fistulae and preauricular pits (Fraser et al., 1978). Similarly, Stickler syndrome is an autosomal dominant type 2 collagen condition that causes sensorineural hearing loss, cleft palate, and congenital myopia (Kochhar et al., 2007). The clinical features of Pendered syndrome are found to be euthryoid goiter, high-frequency slopping hearing loss, and affected vestibular aqueduct (Allen & Goldman, 2020). Even the Pendered syndrome is associated with abnormal bony labyrinth (Mondini dysplasia or dilated vestibular aqueduct) and hearing loss (Kochhar et al., 2007; Reardon et al., 2000).

Congenital severe-to-profound sensorineural hearing loss and vestibular dysfunction is main feature seen in individuals with Usher Syndrome type I. Individuals having Usher syndrome who have hearing loss find typical sound amplification inefficient and usually communicate manually. Because of concomitant vestibular dysfunction, developmental motor milestones for sitting and walking are achieved later than expected. Type II Usher syndrome is distinguished by congenital

mild-to-severe sensorineural hearing loss and intact vestibular function. For these people, hearing aids provide appropriate sound amplification, and use aural oral communication. The rarest form of Usher syndrome is type III, which is characterized by gradual hearing loss and degradation of vestibular function (Rosenberg et al., 1997). Jervell and Lange-Nielsen syndrome is reported to be the third most common type of autosomal syndromic hearing loss consisting of congenital sensorineural hearing loss, with cardiac problems (Kochhar et al., 2007).

Among the genetic loss, the non-syndromic type comprises around 70% but may not be detected at birth or soon after as they may not have apparent symptoms, and some are late-onset and progressive. Hearing impairment is reported to be of various types and degrees ranging from surgically and medically treatable conductive deficits to sensory hearing loss requiring amplification to central auditory dysfunction with or without a peripheral component (Hall III et al., 1995).

2.4 Complications and effects of congenital hearing loss

The Joint committee on Infant hearing (JCIH) recommended that every child should undergo hearing testing and evaluations by three months of age, and intervention should be started by six months of age (JCIH, 2019) . Rout et al (2008) reported a mean age of hearing loss detection as 3.03 years and commencement of habilitation by a mean age of 7.38 years in eastern India.

Delayed speech-language development due to delay in detecting hearing loss and intervention is found to impact literacy, social life, and academic performance (Wood et al., 2015). Hearing impairment of any degree is found to have a profound effect on child's speech language development and education, leading to stigmatization (Rout & Singh, 2010). It has also been noted that most parents choose

to wait for the child to speak until the age of two or three then only seek medical consultation, which delays hearing loss detection. It has been reported that about 34% of the children with hearing loss are identified after five years of age (Rout & Singh, 2010).

Hearing loss is noted to impact literacy, self-esteem, and social skills, though language development is found to be the most apparent effect of congenital hearing loss (Figueras et al., 2008; Northern & Downs, 2002; Tellevik, 1981; Yoshinaga-Itano et al., 1998). Hearing loss of any degree and type is reported to impact academic performance negatively, thus leading to reduced employment opportunities later in life (Karchmer & Allen, 1999; Olusanya et al., 2014). Apart from affecting language development and academic performance, psychosocial consequences are equally highlighted, such as a feeling of isolation, depression, and loneliness due to difficulty in communication as a consequence of hearing loss (Fellinger et al., 2012; Mason & Mason, 2007; Stevenson et al., 2010; Theunissen et al., 2014).

It has also been reported that in persons with congenital hearing loss are found to have high percentage of children being associated with other conditions including intellectual disability, vision impairment motor milestone delay, other metabolic disorders (Morava et al., 2011). In addition to individuals being affected, hearing loss is equally found to have a profound impact on family and parents as they have to deal with emotional, financial, and social challenges and are at greater risk of stress and depression (Stacey et al., 2006). It has been reported that parents of children with hearing impairment have been financially suffered, and at the same time, some of them even left their job to availing treatment for their children. (Mckellin, 1995; Wood Jackson & Turnbull, 2004; Zaidman-Zait et al., 2016). It has been highlighted

that untreated hearing is an impacting factor for family, as well as social and economic development in society and the country (Mohr et al., 2000).

From the above review, the causes of congenital hearing loss, the prevalence of hearing loss worldwide and in India, the related syndromes, risk factors, comorbid disorders, and the effect of congenital hearing loss in children and their families can be noted. It is also evident that congenital hearing loss is one of the major sensory disorder which can be caused by the different environmental factors before and during pregnancy or soon after birth. Hereditary and genetic factors are also equally involved as cause of congenital hearing loss. All these factors are found to affect either development of auditory structures or causes complications in functioning of the ear affecting outer, middle or inner as well as auditory neural structures.

Various genes have been identified to be associated with hearing loss, some has prenatal effect while some cause late onset of hearing impairment and other sensory deficits. Along with the prenatal, genetic and hereditary factors environmental factors including birth complications, viral infections are also highly associated with the etiology of hearing loss. The prevalence of congenital hearing loss is much higher in developing countries ranging from 2-8 per 1000 while the overall prevalence of congenital hearing loss is approximately 1 per 1000 individuals. Hearing impairment caused by congenital factors ranges from mild to profound and conductive, sensory mixed as well as neural types as lot of factors have been reported to cause abnormalities in different parts of auditory system.

The congenital hearing loss affects the overall development of child including physical, mental and social wellbeing, results in poorer academic performance and personal achievements. Due to a lack of communication, there are fewer educational

and job availability for individuals with hearing impairment. Emotional problems caused by a reduction in self-esteem and confidence, social withdrawal owing to reduced access to services and difficulties communicating with people. When one of the family members has congenital hearing loss, it affects the entire family financially, emotionally, and socially.

Chapter 3

METHODS

The aim of the study was to find the prevalence and characteristics of congenital hearing loss in children aged from 0-15 years. The register-based study was carried out by reviewing the case files of children who visited All India Institute of Speech and Hearing (AIISH), presenting the complaints of hearing loss from January 2019 to December 2019. The following method was adopted to meet the aim of the study.

3.1. Participants

The retrospective case review consisted of 1217 case files of (647 males & 570 females) children who visited AIISH between 1st January 2019 to 31st December 2019 to estimate the prevalence and characteristics of congenital hearing loss in children. The Clinical Database Management Application (CDMA) and excel based application register were used to obtain the OPD numbers of the pediatric cases. The case files of the pediatric clients who have visited AIISH for hearing evaluation were retrieved from the department of clinical services' registration counter.

The details regarding case history included information about demographic data (age, gender, & socioeconomic status), medical history including prenatal, perinatal, and postnatal history, family history, otologic complaints, results of the otolaryngologic evaluation, speech-language, and developmental history. Neurological and psychological reports were collected for associated disorders.

3.2. Inclusion Criteria

The following inclusion criteria were used for the study:

- Children less aged than 15 years who were reported to AIISH for hearing assessment.
- Children who have undergone a battery of audiological tests that consisted of both behavioral tests (Behavioral Observation audiometry (BOA)/Visual Reinforcement Audiometry (VRA)/Conditioned Play Audiometry (CPA)/Pure Tone Audiometry(PTA), and objective tests such as Auditory Brainstem evoked response (ABR), Otoacoustic Emissions (Transient Evoked Oto-acoustic Emission (TEOAE)/Distortion Product Oto-acoustic Emissions (DPOAE)) and Immittance evaluation (Tympanometry & Acoustic reflexes).
- Children identified as having unilateral or bilateral hearing loss with pure tone audiometry having pure tone thresholds poorer than 25dBHL. The degree of hearing loss that was classified according to Goodman's (1965) classification was considered.
- Children identified as having unilateral or bilateral hearing loss with air conduction Auditory Brainstem Evoked Response, thresholds poorer than 30dBnHL (Stapells & Oates, 1997).
- Absence or presence of Transient Evoked Otoacoustic Emissions or Distortion Product Otoacoustic Emissions, considering 6dB or above SNR levels according to the degree of hearing loss (Norton et al., 2000).
- Presence of comorbid conditions such as craniofacial anomalies.
- Children having congenital malformations of the external ear like anotia, microtia or atresia, or canal stenosis.

- Children with congenital hearing loss associated with comorbid conditions such as intellectual disability and cerebral palsy were considered and analyzed.

3.2.1 Behavioral Assessment

The case files referred for the study followed the battery of assessment starting from detailed otorhinolaryngological evaluation to the detailed audiological, speech-language evaluation, and psychological and neurological evaluation as per the child's need determined by the professionals. The behavioral assessment consisted of an otoscopic evaluation of the external ear and tympanic membrane, followed by BOA/VRA/CPA/PTA) with a calibrated dual-channel audiometer in a sound-treated room standard ANSI guidelines. Selection of the test for audiological evaluation was based on the age and the global development of the child, such as BOA used for infants lesser than six months of age (Sabo, 1999), VRA for children of 5-6 months to 36 months of age (Suzuki & Ogiba, 1961), CPA for children of 3-6 years (Madell, 1998) and pure tone audiometry used for children older than six years of age.

3.2.2 Objective Assessment

All the cases considered for the study had done the objective test battery in AIISH, including immittance evaluation, otoacoustic emission, and auditory evoked brainstem response tests. Tympanometry was performed bilaterally, using a 226 Hz probe tone. Acoustic reflex thresholds were measured at octave frequencies from 500Hz, 1000Hz, 2000Hz, and 4000Hz ipsilateral and contralateral in both ears. TEOAE testing was done in a sound proof room for click stimulus at 80- 85dBPeSPL (Kemp, 1978), and DPOAE testing was done with pure tone stimulus of intensity L1 (65dBSPL) and L2 (55 dB SPL) with a stimulus frequency ratio of 1.22. Overall SNR

level of 6 dB or above was considered the presence of OAEs at least at the consecutive three frequencies tested (Norton et al., 2000). Auditory Brainstem Evoked response carried out for the children was using click and tone burst stimulus. The recording parameters that were used for ABR are tabulated below.

Table 3.1

Protocol used for auditory brainstem response (ABR) testing

Stimulus Parameters	**Acquisition Parameters**
Stimulus: Clicks, Tone burst -500Hz	Filter setting 100-3000 Hz
Duration Clicks- 100us. Tone burst (5us 2-1-2)	Montage Cz-A1 and C-A2
Polarity: Rarefaction	Rate: 11.1/s
Presentation level: 70-90dBnHL Decrease the stimulus level until no identifiable wave V	Analysis window Clicks- 10 ms Tone burst-15-20 ms
Transducer (ER-3A)	Artifact rejection 50uV
Number of sweeps: 2000	Electrode montage: Non-Inverting-Cz, Inverting-M1, Reference-M2

The interpretation and diagnosis of both behavioral and objective tests for the children considered for the study were made by the experienced clinical audiologist at AIISH. All the test battery results were noted to identify the degree of hearing loss. Comorbid conditions such as craniofacial anomalies, associated syndromes, and associated conditions like intellectual disability and cerebral palsy were noted (Chilosi et al., 2010). Risk factors associated with congenital hearing loss including a positive family history of hearing loss, consanguinity, TORCH infections, preterm delivery (<34 weeks of gestational age), birth complications (Birth asphyxia, hypoxia), low

birth weight (< 1.5 kg), hyperbilirubinemia, Meningitis, neonatal intensive care stay more than a week and mechanical ventilation of more than five days were noted.

Those cases with sensorineural type, conductive, and mixed hearing loss that may be acquired due to middle ear infections, viral or bacterial infections, or any otologic trauma were excluded from the study.

3.3 Statistical analysis

The collected case details from the case files were systematically segregated and tabulated in Microsoft Excel and Statistical Package for the Social Sciences (SPSS Version 25), and descriptive analysis was carried out. The data were analyzed further to find the prevalence and characteristics of congenital hearing loss in children across various degrees. The prevalence of congenital hearing loss across gender was analyzed and the various conditions associated with congenital hearing loss were also analyzed separately. For inferential statistics, Pearson Chi-square test was done to see if there was an association between gender and the children with consanguinity as a risk factor for congenital hearing loss and as well as association between gender and birth complication in congenital hearing loss children.

Chapter 4

RESULTS

The aim of the present study was to determine the prevalence and characteristics of congenital hearing loss in children (age less than 15 years) who reported to All India Institute of Speech and Hearing (AIISH), Mysuru, from January 2019 to December 2019 for audiological evaluation. A register-based retrospective study was carried out by reviewing the case files of 1217 children from the Audiology outpatient department (OPD) as revealed by Clinical Database Management Application. The Statistical Package for the Social Sciences (SPSS Version 25) was used to analyse the data, and Pearson Chi-square test was used to determine whether there was an association between gender and children with consanguinity, as well as between gender and birth complications in children with congenital hearing loss.

4.1. Distribution of Congenital Hearing loss in Children reported to AIISH Audiology OPD

In the year 2019 from January to December, a total of 1217 pediatric cases were reported to AIISH audiology OPD for audiological evaluation. All the case files were reviewed, and their demographic details and audiological findings were documented. From the 1217 case files, it was noted that there were 117 children with normal hearing, 213 children with acquired conductive hearing loss, and 64 children with acquired sensorineural hearing loss (total: 394), which were excluded from the study. Out of 1217 children reported to AIISH in 2019, 823 children were reported to have congenital hearing loss and prevalence of congenital hearing loss in children is 67.62%. The degree of hearing loss in these 823 children ranged from mild to

profound. Among the total reported congenital hearing loss (n=823, 67.62%), 375 (45.6%) were females, and 448 (54.4%) were males, as depicted in figure 4.1.

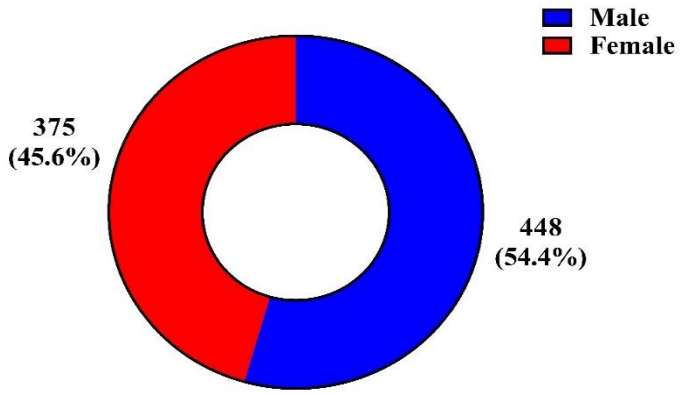

Figure 4.3 Number of children with congenital hearing loss across gender

Among the pediatric cases reported to AIISH in 2019, congenital hearing loss was found to be more compared to acquired hearing loss and more male children were reported than females in this period. As noted from Figure 4.1, the number of male children was more than females among the congenital hearing loss group.

Out of total 1217 children reported to AIISH in 2019 for audiological evaluation the overall prevalence of congenital hearing loss in males and females is 69.24% and 65.78 % respectively as shown in figure 4.2.

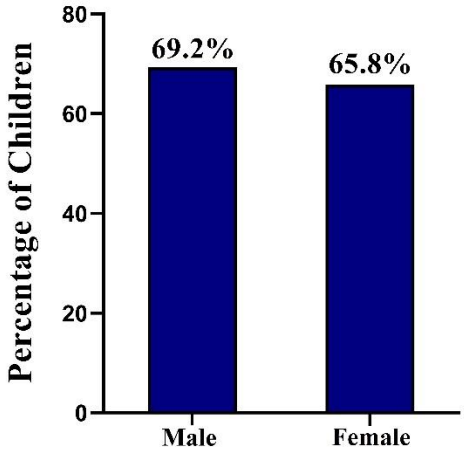

Figure 4.4 Percentage of congenital hearing loss in males and females

4.2. Prevalence of different degrees of congenital hearing loss.

Out of 823 children diagnosed with congenital hearing loss, 2.9% (n=24) children had unilateral hearing loss (mild; n=2, moderate; n=7, severe; n=4, moderately severe; n=4 and profound; n=7) , and 97.1% (n=799) children had bilateral hearing loss with the degree of hearing loss ranging from mild to profound in both ears. The bilateral congenital hearing loss was found to be more than unilateral hearing loss. The degree of hearing loss in individual children's ears was noted, and the degrees of hearing loss in the right and left ear are represented in figure 4.3.

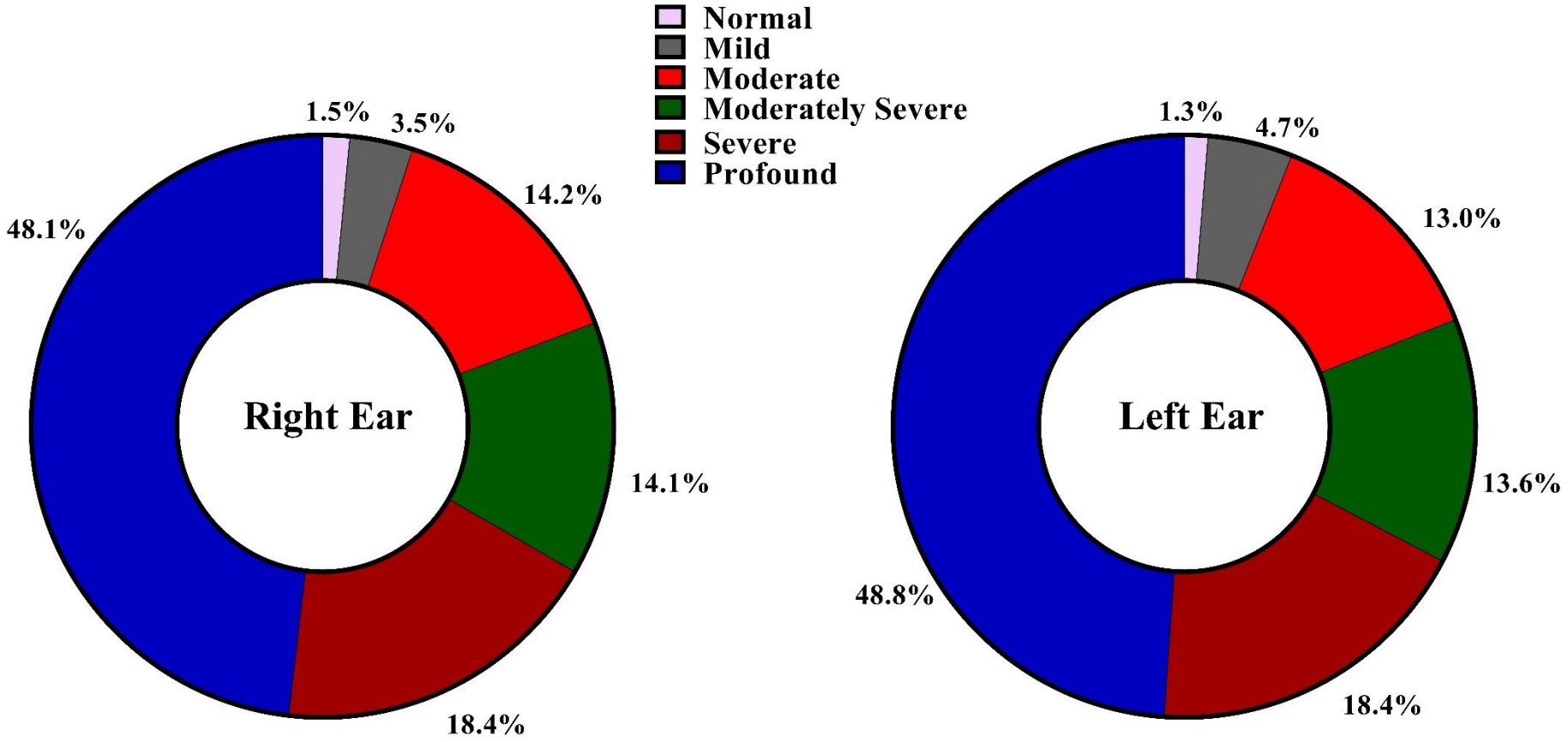

Figure 4.5 Percentage of ears with different degrees of hearing loss in children with congenital hearing loss

From the figure 4.3, it is evident that profound hearing loss was most prevalent in children with congenital hearing loss in both the ears. In right ear profound hearing loss was present in 48.1% (n=396) ears, followed by severe in 18.5% ears (n=152), moderately severe in 14.1% ears (n=116), moderate in 14.2% ears (n=117) and mild hearing loss in 3.5% ears (n=29) and normal hearing (normal ears of unilateral hearing loss cases) in 1.5% ears (n=13). Similarly, in left ear, profound hearing loss was present in 48.8% ears (n=402), followed by severe in 18.4% ears (n=152), moderately severe in 13.6% ears (n=112), moderate in 13% ears (n=107) and mild hearing loss in 4.7% ears (n=39) and normal hearing (normal ears of unilateral hearing loss cases) in 1.3% ears (n=11).

The children's age range was classified as 0-1, 1-3, 3-5, 5-10, and 10-15 years of age for finding the occurrence of congenital hearing loss across gender. The percentage of congenital hearing loss across gender in each age range is represented in figure 4.4.

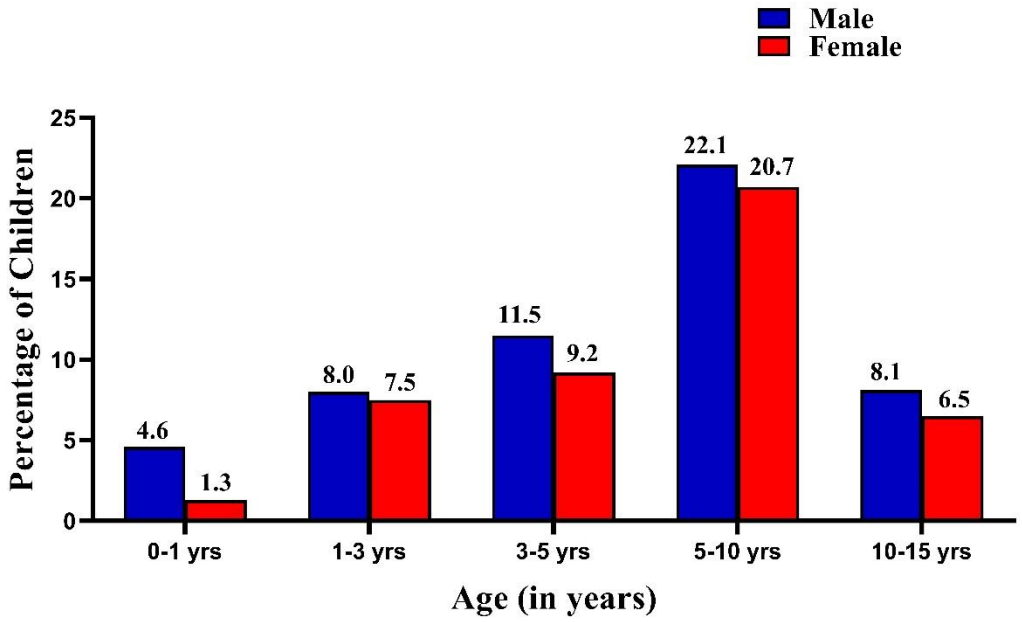

Figure 4.6 Percentage of congenital hearing loss in male and female children across age groups

From the figure 4.4, it was noted that 6% (n=50) of children with hearing loss were in the age range of 0-1 years, 15.6% (n=128) in 1-3 years, 20.8% (n=171) in 3-5 years, 42.8% (353) in 5-10 years, and 14.8% (121) in 10-15 years age range. From the figure 4.4 it can be observed that, percentage of congenital hearing loss in children was 4.6% (n=38) in males and 1.3% (n=12) in female children aged 0-1 year; 8.0 % (n=66) in males and 7.5 % (n=62) in female in 1-3 year age group; Similarly 11.5 % (n=95) in males and 9.2 % (n= 76) in females in 3-5 year group; 22.1% (n=182) in males and 20.7% (n=171) in female in 5-10 year group; 8.5 % (n=67) in males and 6.5 (n=54) in females aged 10-15 year. As represented in figure 4.4, more number of children with congenital hearing loss were in the age range of 5-10 years who had reported to AIISH in 2019 followed by 3-5 years, 1-3 years, 10-15 years and 0-1 years respectively.

The children with various degrees of congenital hearing loss in all age range considered were represented in table 4.1. From table 4.1 it can be noted that profound hearing loss was most prevalent in children (48.5%, n=798) followed by severe (18.5%, n=304), moderately severe (13.9%, n=228), moderate (13.6%, n=224) and mild hearing loss (4%, n=68) and normal hearing (normal ears of unilateral hearing loss cases) in 1.5% (n=24) children in overall all age groups combined.

Table 4.1

Number of children with different degrees of congenital hearing loss across the age range

		Degree of hearing loss in the right ear						Total
		Normal	**Mild**	**Moderate**	**Moderately Severe**	**Severe**	**Profound**	
Age Range (in years)	0 to 1	3(0.4%)	3(0.4%)	5(0.6%)	2(0.2%)	8(1.0%)	29(3.5%)	50(6.1%)
	1 to 3	3(0.4%)	1(0.1%)	11(1.3%)	6(0.7%)	19(2.3%)	88(10.7%)	128(15.6%)
	3 to 5	0(0.0%)	7(0.9%)	18(2.2%)	17(2.1%0	31(3.8%)	98(11.9%)	171(20.8%)
	5 to 10	2(0.2%)	9(1.1%)	56(6.8%)	69(8.4%)	75(9.1%)	142(17.3%)	353(42.9%)
	10 to 15	5(0.6%)	9(1.1%)	27(3.3%)	22(2.7%)	19(2.3%)	39(4.7%)	121(14.7%)
Total		**13(1.6%)**	**29(3.5%)**	**117(14.2%)**	**116(14.1%)**	**152(18.5%)**	**396(48.1%)**	**823(100.0%)**

		Degree of hearing loss in the left ear						Total
		Normal	**Mild**	**Moderate**	**Moderately Severe**	**Severe**	**Profound**	
Age Range (in years)	0 to1	1(0.1%)	4(0.5%)	7(0.9%)	2(0.2%)	4(0.5%)	32(3.9%)	50(6.1%)
	1 to 3	3(0.4%)	1(0.1%)	10(1.2%)	6(0.7%)	21(2.6%)	87(10.6%)	128(15.6%)
	3 to 5	0(0.0%)	8(1.0%)	17(2.1%)	15(1.8%)	28(3.4%)	103(12.5%)	171(20.8%)
	5 to 10	4(0.5%)	8(2.1%)	53(6.4%)	61(7.4%)	80(9.7%)	138(16.8%)	353(42.9%)
	10 to 15	3(0.4%)	9(1.1%)	20(2.4%)	28(3.4%)	19(2.3%)	42(5.1%)	121(14.7%)
Total		**11(1.3%)**	**39(4.7%)**	**107(13.0%)**	**112(13.6%)**	**152(18.5%)**	**402(48.8%)**	**823(100.0%)**

4.3. Etiology of congenital hearing loss in children.

In order to identify the various etiological factors of congenital hearing loss in children, case files of the children reported to the Audiology outpatient department (OPD) at AIISH in the year 2019 were reviewed. The demographic details and detailed case history were noted, information was analyzed, and the results are shown in figure 4.5.

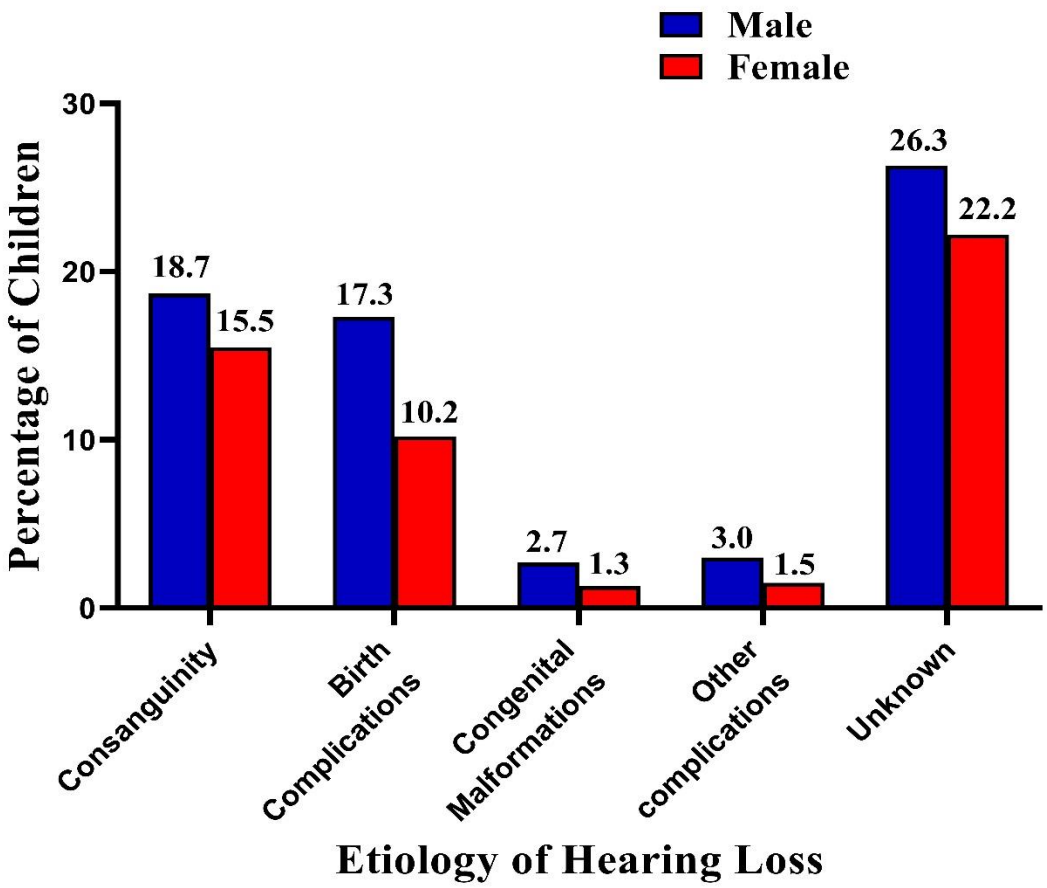

Figure 4.5 Percentage of children having different etiologies of congenital hearing loss

From figure 4.5, it can be noted that, the consanguinity was highly prevalent in the children (n=282), followed by birth complications (n=165), congenital malformations (n=32), other complications such as TORCH infection, chronic heart disease, seizure disorder (n=38) and unknown causes (n=400) where hearing loss was

the primary complaint, but the etiology was unknown. It can be noted that 18.7% male children and 15.5 % female children had parents with a history of consanguineous marriage; 17.3% of male children, 10.2% female children had etiology as birth complications; 2.7% male children and 1.3% female children as congenital malformations; 3 % male children and 1.5 % female children as other complications; 26.3 % male children and 22.2% female children with unknown etiology. Birth complications and risk factors were birth asphyxia, Neonatal Intensive care Unit (NICU) stay of 5 or more days, Neonatal jaundice, premature delivery, and low birth weight TORCH infections. Congenital malformation included microtia (n=10), anotia (n=15) with atresia (n=24), and (n=6) children with cleft lip and palate.

Pearson Chi-square test was done to see an association between gender and children with consanguinity as a risk factor for congenital hearing loss. The results indicated that there was no association between consanguineous marriage and gender of children with congenital hearing loss [χ^2 (1) =0.005, p =0.94].

4.4 Birth complications and risk factors associated with congenital hearing loss.

From the detailed history of reviewed case files, the associated birth complications and risk factors were documented. Birth complications and risk factors reported were birth asphyxia, neonatal intensive care unit (NICU) stay of 5 or more days, neonatal jaundice, premature delivery, and low birth weight, chronic heart disease, TORCH infections and seizure disorders. The number of cases reported with birth complications and risk factors associated with congenital hearing loss are given in figure 4.6.

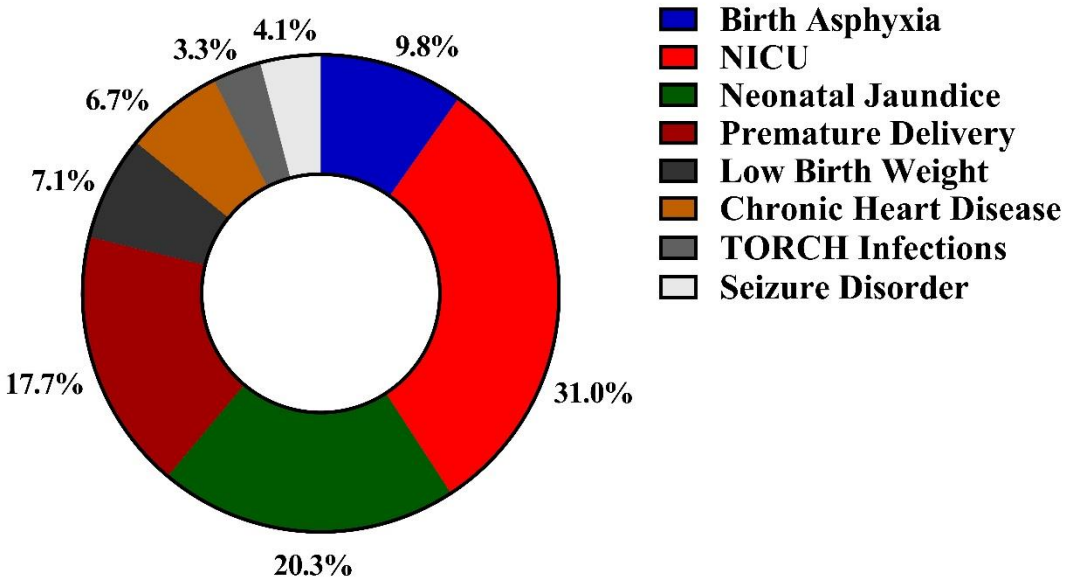

Figure 4.7 Percentage of male and female children with different birth complications and risk factors reported for congenital hearing loss

From the figure 4.6 it was noted that Neonatal Intensive Care Unit stay for more than 5 days was more prevalent (31.0%, n=87) with those who reported birth complications followed by neonatal jaundice (20.3%, n=54), premature delivery (17.7%, n=47), birth asphyxia (9.8%, n=26), and low birth weight (7.1%, n=19), chronic heart disease (6.7 %, n=18) TORCH Infection (3.3%, n=9) and seizure disorder (4.1 %, n=11).

The distribution of children having congenital hearing loss with and without birth complication across gender in depicted in table 4.2. From table 4.2 it can be noted that more number of male children were reported to have birth complications compared to females.

Pearson Chi-square test was done to see an association between gender and children with birth complications and risk factors for occurrence of congenital hearing loss. The results indicated that there was a significant association between gender of

children and birth complications and risk factors for occurrence of congenital hearing loss [$\chi 2$ (1) =8.34, p =0.004].

Table 4.2

Distribution of children with and without birth complication across gender

	With birth complications and risk factors	Without birth complications and risk factors	Total
Male	97 (11.8%)	351(88.2%)	448
Female	52 (6.3%)	323(92.7%	375
Total	149	674	823

4.5 Associated conditions in children with congenital hearing loss

Among the reported children with congenital hearing loss, various associated co-morbid conditions were documented. The number and percentage of children with associated co-morbid conditions (intellectual disability, chronic heart disease, cerebral palsy, downs syndrome, seizure disorder, microcephaly, visual impairment, and autism) with congenital hearing loss across gender is shown in table 4.2.

Table 4.3

Number of children with associated co-morbid conditions reported with congenital hearing loss across gender

Co-morbid conditions	Male	Female	Total
Intellectual Disability	30(48.4 %)	30(71.4 %)	60(57.6%)
Chronic Heart Disease	15(24.2 %)	3(7.1 %)	18(17.3%)
Cerebral Palsy	6(9.7 %)	4(9.5 %)	10(9.6%)
Downs Syndrome	2(3.2 %)	1(2.4 %)	3(2.8%)
Seizure Disorder	7(11.3 %)	4(9.5 %)	11(10.5%)
Visual Impairment	1(1.6 %)	0	1(0.9%)
Autism	1(1.6 %)	0	1(0.9)
Total	**62**	**42**	**104**

It was noted that intellectual disability was the most prevalent associated condition among the co-morbid conditions, with 57.5% (n=60). Followed by chronic heart disease 18.2% (n=18), seizure disorder 10.5 (n=11), cerebral palsy 9.6% (n=10), downs syndrome 2.8% (n=3), and 0.9% (n=1) each for autism and visual impairment among those reported with comorbid conditions with congenital hearing loss as shown in table 4.3.

4.6 Effect of congenital hearing loss in children.

The consequences of hearing loss in the reported cases at the time of assessment process were recorded during case file documentation and analysis. Among the children who reported as having congenital hearing loss (67.6%; n=823), it was observed that in children with unilateral hearing loss (2.9%, n=24), 18 of them had congenital ear malformations, and 6 of them had cleft lip and palate, which were reported to have resulted in delayed speech and language and misarticulation. Out of 18 children with congenital malformation, 4 of them were infants, parents did not report any consequences of hearing loss, although they were worried about their children's hearing abilities due to deformed external ear. Delayed speech and language development was the main complaint in severe to profound hearing loss group followed by misarticulation and poorer academic performance in mild loss. Some of the parents have complained of reduced academic performance as the child progressed to higher classes in mild to moderate hearing loss group. In children with bilateral hearing loss (97.1%; n=799), the degree of hearing loss ranged from mild to profound degrees and the main consequence was delayed speech and language.

Those children with mild to moderate hearing loss (9.5%; n=78) were reported to have misarticulation and reduced academic performance, while those with higher

degrees of hearing loss (80%; n= 659) complained about delayed speech and language. The remaining children (10.5%; n= 86) who had a mild degree of hearing loss had no significant complaints on speech-language development, but some of them had complaints related to academic and classroom performance. Some parents of children with mild degree of hearing loss reported that children needed repetition and used high volume while watching television.

Chapter 5

DISCUSSION

The objective of this study was to retrospectively analyze the pediatric cases in the age range of 0 to 15 years reported to the All India Institute of Speech and Hearing from January 2019 to December 2019 for audiological evaluation. The presence of a congenital hearing loss in those children and the risk factors associated with the hearing loss was documented. The study also reported prevalence of different degrees of congenital hearing loss present in children across the age range.

5.1 Prevalence of congenital hearing loss in children

Out of 1217 children assessed for hearing loss, 67.62% (n=823) were found to have congenital hearing loss while remaining 32.8% (n=394) were excluded as they were diagnosed with normal hearing (n=117), acquired conductive (n=313) and acquired sensorineural (n=64) hearing loss. Since the current study is a register based study carried out for duration of one year, higher prevalence of congenital hearing loss of 67.62 % obtained in current study does not match with the other prevalent studies are performed in live births in the pediatric hospital setup or during newborn screening. The prevalence of sensorineural hearing loss (SNHL) is reported to be 2-3 per 1000 live births in India, and out of 1000 babies, 1 child is found to have profound hearing loss at birth or in the pre-lingual age group (Shrivastava & Gupta, 2018). In another study by Varshney (2016), it was reported that 4 in every 1000 children suffer from severe to profound hearing loss. In a study in south India, the estimated prevalence of hearing loss among newborns was 4.1 per 1000 babies tested (Augustine et al., 2014). Similarly, Dietz (2009) reported that the prevalence of overall hearing loss among infants was 2.1 per 1000 in Finland.

A higher number of congenital hearing loss cases reported at the institute could be attributed to AIISH being the premier institute for speech and hearing disorders, with many referrals coming from outreach service centers and other primary hospitals. Another reason could be that people are visiting AIISH for both hearing assessment and rehabilitation due to initiation of various programs such as newborn hearing screening, school screening and screening camps etc. at AIISH. Also people from all over India visit AIISH for hearing evaluation.

Among the 823 children more males (54.4 %, n=448) were found to have congenital hearing loss compared to females (45.6 %, n=375). Bamiou et al (2000) found a male predominance in children with sensorineural hearing loss (59.8 %), and no statistically significant gender difference in terms of degree of hearing loss. This was consistent with a study by Kumar et al (2011) which found similar distributions in men (54%) and women 46%. Similarly, another study reported a 1.72: 1 male predisposition among 261 cases of congenital and acquired hearing impairments, with 165 males (63.2 % and 96 females (36.8 %) (Kalsotra et al., 2002). However, in contrary to current findings, a study consisting of 50 children, Shrivastava and Gupta (2018) observed that percentage of female is more than female 27 (54%), and 23 (46%) respectively with female: male a ratio of 1.76:1.

5.2 Occurrence of different degrees of congenital hearing loss

In 823 children diagnosed with congenital hearing loss, bilateral hearing loss (n=799) was found to be more than the unilateral hearing loss (n=24) in the current study with degrees of hearing loss ranging from mild to profound in both ears. Similar to current study, a total of 80.1 percent of children reported bilateral hearing loss (Billings & Kenna, 1999). Likewise, the results of current study were in consonance

with the prevalence study in neonates where 62.5% had bilateral hearing loss and 37.5% unilateral hearing loss.

Profound hearing loss was most prevalent in children with congenital hearing loss 48.5% (n=798) ears, followed by severe 18.4% (n=304), moderately severe 13.9% (n=228), moderate 13.6% (n=224) and mild hearing loss 4% (n=68) and normal hearing (normal ears of unilateral hearing loss cases) in 1.5% (n=24). Bilateral profound hearing loss was more evident in all age groups. Similar to the current study findings, Wiranadha and Hartayanti (2020) reported that 71% of the individuals were found to have severe to profound hearing loss, 8% severe, 5% in moderate to severe, 10%in moderate and 6% mild hearing loss in a study consisting of 125 children.

In the present study, it was found that 6% (n=50) of the total children with hearing loss were in the age range of 0-1 years, 15.6% (n=128) in 1-3 years, 20.8% (n=171) in 3-5 years, 42.8% (353) in 5-10 years, and 14.8% (121) in 10-15 years age range. A lower number of children in 0 to 1 year of age may be owing to non-syndromic hearing loss with no visible symptoms. In addition, parents were unable to notice hearing loss in younger age group who did not have a newborn hearing screening at birth. In developing countries like India, newborn hearing screening is still not available in every part of the country (Garg et al., 2015). Lack of Health education among the general public about the risk factors for hearing loss in children, as well as awareness of the availability of screening tests in neonates, might have delayed early suspicion and identification of hearing loss (Kumar et al., 2018). During the study period, most children reported to the institution for evaluation were between the ages of 5 and 10, followed by those between the ages 3 and 5. This could be because parents wait for the development of speech and language until preschool, or they may have gone to other institutes and been recommended to AIISH for further

evaluation later. Lack of knowledge about new born screening in the rural areas and even in primary health care centers may delay the suspicion and identification of hearing loss (Ramkumar, 2017). Lack of awareness and poor socioeconomic status of people, particularly in rural India, may be one of the reasons why people do not seek hearing screening for newborns and babies.

5.3 Etiology of congenital hearing loss in children

Among the various etiological factors of congenital hearing loss that were noted, consanguinity (n=282) were highly prevalent in the children reported to AIISH in the year 2019, followed by birth complications (n=165), congenital malformations (n=31). The children with unknown causes (n=400) which includes hearing loss as main complain but etiology was unknown.

The results of the current study are in consonance with that reported by (Panakhian, 2005), who has indicated that consanguineous marriages increase homozygosis of pathogenic recessive genes, increasing the probability of disabled babies being born. Consanguineous parents are more likely to be homozygous for the same trait, and therefore, increase the chance of having a child affected by congenital deafness. The risk of having more than one child with SNHL was 3.5 times higher in individuals with consanguineous marriages (Almazroua et al., 2020; Sanyelbhaa et al., 2017). Also, Zakzouk (2002) reported 50 % consanguinity in hearing loss in 6421 participants from Riyadh and 9540 from other parts of Saudi Arabia. It was also reported that consanguinity is common even in south India (Gray, 1989).The high occurrence of consanguinity is attributed to social traditions, the practice of arranged marriage within families, and a lack of awareness of the negative genetic influences of such practices.

Congenital malformation that were found in present study included microtia (10), anotia (15) with atresia (24), and (6) children with cleft lip and palate. Previous studies have also found craniofacial abnormalities and syndromes linked with congenital hearing loss(Cone-Wesson et al., 2000). Waardenburg syndrome, branchio-oto-renal syndrome, Stickler syndrome, Melnick-Fraser syndrome, Treacher Collins syndrome, neurofibromatosis type 2, and osteogenesis imperfecta have also been reported to be associated with hearing loss (Allen & Goldman, 2020).

5.4 Birth complications and risk factors associated with congenital hearing loss

In this study, the reported risk factors that were birth asphyxia, neonatal intensive care unit (NICU) stay of 5 or more days, neonatal jaundice, premature delivery, and low birth weight. Among these factors, NICU stay was found to be more prevalent with 31% (n=87) of those who reported birth complications, followed by neonatal jaundice 20.3% (n=54), premature delivery 17.7% (n=47), birth asphyxia 9.8% (n=26), and low birth weight 7.1% (n=19), chronic heart disease 6.7% (n=18), seizure disorder 4.1%(11),TORCH infections 3.3% (n=9). The risk factors noted in the current study are supported by Joint Committee on Infant Hearing who has listed all above as risk factors for hearing loss(JCIH, 2019).In this study, children admitted to the NICU had a higher rate of hearing loss. In another study, it is reported that neonates, those who have received NICU care account for 10% to 15% of the newborn population and have a higher prevalence of hearing loss than healthy newborns (Robertson et al., 2009).

Children reported with hyperbilirubinemia and who received phototherapy are also at risk of developing hearing loss (Boskabadi et al., 2018). Studies supported that increased blood indirect bilirubin can pass the blood-brain barrier and accumulate in

the auditory ventricular nucleus cells, which is one reason for early sensorineural hearing loss in developing countries (Öğün et al., 2003).

The reported findings in this study were supported by previous studies on risk factors for hearing loss. Birth hypoxia, asphyxia, and ischemia have been identified as primary causes of early hearing loss or deafness(Borg, 1997). Perinatal asphyxia, also known as hypoxia ischemic encephalopathy, is associated with a significant increase in permanently increased hearing thresholds, especially if hypothermia treatment is required (Shankaran et al., 2012).Similarly, birth asphyxia was also one of the major risk factors reported in children with congenital hearing loss, similar to the study by Pawar et al.(2019).Low APGAR scores and the presence of hyperbilirubinemia in premature babies are risk factors, and the NICU stay after these issues increase the likelihood of hearing loss (De Vries et al., 1987; JCIH, 2019).

In this study, there was significant association found between gender and birth complications in children with congenital hearing loss, where more number of male children with congenital hearing loss had birth complications than females. In coherence to this study, the high prevalence of males and preterm birth has been reported by multicenter cross sectional study in Japan (Funaki et al., 2020) . Similar result has also been reported by European Program of Occupational Risks and Pregnancy Outcome (EUROPOP) where male children has higher risk for preterm birth (Zeitlin et al., 2002). In support to the finding in this study, significant association between male child and preterm birth has been reported in a national cohort study from Netherlands (Peelen et al., 2016).

5.5 Associated conditions with congenital hearing loss in children

The total number of children with congenital hearing loss having comorbid conditions in current study is 104. Among the reported comorbid conditions, intellectual disability was present in 57% (n=60) of children followed by chronic heart disease 17.3% (n=18), cerebral palsy 9.6% (n=10), seizure disorder 10.5% (n=11), Downs syndrome 2.8% (n=3), 0.9% (n=1) autism and 0.9% (n=1) visual impairment. It has been shown that more number of children with congenital hearing had intellectual disability, visual impairment, motor milestone delay, and other metabolic disorders as associated condition (Morava et al., 2011). A higher prevalence of chronic heart disease in consanguineous marriages has been reported in South Indian studies (Ramegowda & Ramachandra, 2006). Similarly, children with congenital heart disease (CHD) are more likely to have neurodevelopmental abnormalities and sensorineural hearing loss (SNHL) (Gopineti et al., 2020). According to a review study, children with cerebral palsy were reported to have varying degrees of hearing loss (Reid et al., 2011).

5.6 Effects of congenital hearing loss in children

In present study, delayed speech and language development was the main complaint followed by misarticulation and poorer academic performance. Some of the parents have complained of reduced academic performance as the child progressed to higher classes. Many children with mild to moderate hearing loss without other complications demonstrated misarticulation, which might be due to attenuation of speech and the missing acoustic characteristics of speech. Reduced academic performance due to hearing loss may be because of inattention and unable to follow the classroom activities during teaching and learning. Similar to the findings of the

current study, low attention and reduced communication performance in school children have been reported (Elbeltagy, 2020).

Chapter 6

SUMMARY AND CONCLUSIONS

The present study aimed to find the prevalence and characteristics of congenital hearing loss in children aged from 0-15 years reported to the All India Institute of Speech and Hearing (AIISH) for audiological evaluation from January to December 2019. A retrospective register-based study was carried out by assessing the case files of the children reported to the AIISH Outpatient Department (OPD). All the retrieved information was segregated and analyzed for different degrees of congenital hearing loss, the primary etiology of hearing loss reported by parents, risk factors and birth complications associated with the hearing loss, comorbid conditions, and the present effect of congenital hearing loss at the time of investigation that was documented. From the findings of the study, it can be concluded that the majority of the children, 823 out of 1217(67.6%), reported to the institute had congenital hearing loss and bilateral profound hearing loss was more prevalent among them.

Consanguinity was one of the most reported risk factors. The congenital hearing loss was more prevalent in male children (54.4%, n=448) compared to females (46.6%, n=375) in the study period. More children with congenital hearing loss were in the age range of 5-10 and 3-5 years. There was a significant association between gender of children and birth complications and risk factors for occurrence of congenital hearing. Associated co-morbid conditions seen in children with congenital hearing loss were intellectual disability, chronic heart disease, cerebral palsy, downs syndrome, seizure disorder, microcephaly, visual impairment, and autism. Delayed speech and language development was the main complaint followed by misarticulation and poorer academic performance in children with congenital hearing loss.

Implications of the study:

- The study provides data regarding the prevalence, associated risk factors and etiology of congenital hearing loss in children who visited AIISH, which can be used for various awareness programs for general public and other professionals regarding prevention, early identification and rehabilitation of children with hearing impairment conducted by AIISH in future.
- Knowing the prevalence of various degrees of hearing loss will help to highlight severity of hearing loss and its effect on the child's speech-language development, academic performance, psycho social and overall achievement.
- Knowing the occurrence of higher number of consanguineous marriages and its association with congenital hearing loss, helps in counseling parents to educate them about the consequences of getting married in blood relation and its effect on infants hearing and those planning for a second child.
- Knowing the effect of different degrees of hearing loss and congenital hearing loss in children helps family members during counseling parents and planning for effective rehabilitation programs.

Future directions:

- Prevalence study can be carried out in newborn hearing screening programs to obtain more accurate prevalence data of congenital hearing loss.
- Risk factors and the complications for congenital hearing loss can be thoroughly studied in individuals who visit AIISH to further plan effective awareness programs for prevention of hearing loss.

- Study can be carried out regarding consequences and impact of hearing loss in parents and family having children with congenital hearing loss and also to assess the quality of life of family due to hearing loss.

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