AN EPIDEMIOLOGICAL STUDY ON CHILDREN WITH SYNDROMIC HEARING LOSS

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ABSTRACT : *Objectives : To study the epidemiological factors in children with syndromic hearing loss.*

Study design : Interview based prospective study.

Settings : Govt. ENT Hospital, AYJNIHH, NIMH-SEC, and Schools for the Deaf-in Hyderabad and Secunderabad

Patients : Children aged below 14 years, with hearing loss, their parents/guardians.

Intervention(s) : The study revealed type and degree of hearing impairment. In high risk groups genetic counseling was offered.

Results : Epidemiological studies were carried out in 743 children below 14 years with hearing impairment and 138 (18.57%) were found to have syndromic deafness. Majority of the children with hearing loss have an association of ocular abnormality (22.46%, n=31) followed by skeletal anomalies 14.49% (n=20) and dental anomalies (10.86%). We observed 24 cases (3.21%) with genetically well recognized syndromes.

Conclusion : Data is generated on epidemiological and etiology of Hearing Impairment. Hearing Impairment is due to both environmental and genetic causes. Environmental factors in 17 (13.77%), genetically inherited 21 cases (15.22%) and the cause is not known in the remaining cases. Such a data is required in order to offer genetic counseling to reduce the genetic burden.

Key Words : *Epidemiology, Syndrome, Consanguinity.*

INTRODUCTION

Hearing loss is found to be one of the most common disabilities of man and over 700 million individuals world wide are hearing impaired with pure tone averages greater than 55 dB. Epidemiological data for pre-lingual hearing loss show that 1 in 1000 neonates are born with severe to profound hearing loss of which 50% of the loss is inherited[2]. Hearing loss is categorized as syndromic or non-syndromic hearing loss. Approximately 70% of congenital cases in association with genetic factors are categorized as non-syndromic and the remaining as syndromic deafness because of clinical findings[3,4].

Walch et al in their study to determine the etiology of bilateral sensorineural hearing disorder, found that age of first diagnosis was 4 months to 11 years with a mean of 42 months. The sex ratio of male-female was found to be approximately 1:1. The cause of hearing impairment was found to be acquired in 38% of the cases and genetically inherited in 18%. However, in 44% of the cases, the etiology of hearing loss could not be determined[5].

Bafaqeeh et al revealed that parental education was an important demographic factor in causation of deafness[6]. Sutton and Rowe in a study to find the risk factor for childhood sensorineural hearing loss found that significantly increased number of cases with hearing loss were observed in families with low socio-economic status. They also found high proportion of cases (24%) with cranio-facial abnormalities that include non-aural abnormalities and dysmorphic features[7].

The epidemiological data for syndromic hearing loss from Andhra Pradesh are very meager and hence the present study was taken up to understand various epidemiological parameters relevant to syndromic hearing loss in children from twin cities of Hyderabad and Secunderabad.

MATERIALS AND METHODS

743 children with hearing loss, below 14 years of age

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Table-1 : Type of deafness in children

Sl. No	Type	No. of Subjects	Percentage
1.	Syndromes	138	18.57
2.	Non-syndromes	605	81.43
3.	Total	743	100

Table-II : Age-wise distribution of children with syndromic hearing loss

Sl. No.	Age group (in years)	No. of cases	percentage
1.	0-3	19	13.77
2.	3-6	29	21.01
3.	6-9	30	21.74
4.	09-12	23	16.67
5.	12-14	37	26.80
Total		138	100

Table- III : Sex-wise distribution of children with syndromic hearing loss

Sl. No.	Sex of the child	No. of cases	Percentage
1.	Male	75	54.35
2.	Female	63	45.65
Total		138	100

attending Govt. ENT Hospital, Koti and various schools for the deaf in and around the twin cities formed the subjects. Information on age, sex, religion, parents' occupation and literacy along with birth history, developmental and family history were recorded from the parents or guardians of the child using a standard questionnaire. Audiological evaluations (Pure Tone Audiometry, Brain stem Evoked Response Audiometry, and Oto-acoustic Emission) were done to determine the type and degree of hearing loss besides physical and clinical examination. Psychologist and Ophthalmologist were consulted wherever necessary. More emphasis was given to children with syndromic deafness.

RESULTS

The results on the type of the hearing impairment are presented in table-1. Out of 743 children with hearing loss, aged below 14 years, 138 (18.57%) were found with syndromic hearing impairment and 81.73% (n=605) constituted for only isolated (non-syndromic) deafness.

Table-IV : Literacy of parents of children with syndromic hearing loss

Sl. No.	Literacy	No. of Cases	Percentage
1.	Literate	72	52.17
2.	Illiterate	66	47.83
Total		138	100

Table V : Distribution based on the Socio-economic status of the children with deafness

Sl. No.	Socio-economic status	No. of cases	Percentage
1.	Lower	30	21.74
2.	Lower-Middle	58	42.03
3.	Middle	44	31.88
4.	Higher	6	4.35
Total		138	100

Table-VI : Distribution based on the religion of the children with syndromic deafness

Sl. No.	Religion	No. of Cases	Percentage
1.	Hindus	121	87.68
2.	Muslims	14	10.15
3.	Christians	3	2.17
Total		138	100

The results of various epidemiological data that include the age of the child, sex, religion, parents' literacy, socio-economic status and family history are presented in tables 2-7. The table-2 lists the distribution of children with deafness based on their age. Majority of children belonged to 12-14 years of age (n=37, 26.8%) followed by 6-9 (n=30, 21.74%), 3-6 (n=29, 21.01%), 9-12 (n=23, 16.67%) and 0-3 (n=19, 13.77%) years of age. The sex wise distribution of cases (Table-III) shows that majority of them are boys (n=75, 54.35%). Only 45.65% (n=63) were girls. The distribution of subjects based on the parent's literacy and socio-economic status is given in tables 4 and 5 respectively. The percentage of literate (n=72, 52.17%) were greater as compared to illiterate parents (n=58, 42.03%) and lowwer middle class (n=58, 42.03%) was greater than middle (n=44, 31,88%) and low (n=30, 21.74%) socio-economic status. Only 4.35% (n=6) parents belonged to higher socio-economic status. Table –VI represents the distribution of patients based on

Table-VII : Distribution of the subjects based on the nature of marriage in their parents

Sl.No.	Nature of marriage	No. of cases	Percentage
1.	Consanguineous	78	57.52
2.	Non-consanguineous	60	43.48
Total		138	100

Table VIII : Etiology of hearing loss in children with syndromic hearing impairment

Sl. No.	Etiology	No. of cases	Percentage
1.	Geneticaly inherited	21	15.77
2.	Acquired	17	13.77
3.	Unknown etiology	100	71.01
Total		138	100

the religion of the child. The percentage of Hindus (n=121, 87.68%) is far greater than Muslims (n=14, 10.15%) and Christians(n=3, 2.17%). An analysis on the nature of marriage of parents of children with hearing loss (Table VII) shows that 57.52% (n=78) of children were the products of consanguineous parents. 43.48% (n=60) were born to non-consanguineous parents.

The results on etiology of hearing loss in children with deafness are given in table-VIII. It shows that in 15.22% (n=21) of children, deafness was inherited, in 13.77% (n=17) it was acquired and in 71.01% (n=100) the etiology was unknown.

The classification of syndromic hearing loss, as given table-IX, based on the organ/system involved gives the prevalence of hearing loss under each category. It is seen that in most of the children deafness is associated with ocular abnormality (n=31, 22.46%) followed by skeletal (n=20, 14.49%) dental (n=15, 10.86%), dermal (n=9, 6.52%), metabolic (n=4, 2.89%), craniofacial and neurologic (n=3, 2.17%) and renal abnormality (n=2, 1.5). Cryptogenic deafness as observed in 17 (12.31%) cases and deafness with other abnormalities accounted for 24.63% (n=34).

Out of 743, 24 (3.21%) cases were observed with genetically well-recognized syndromes. It includes one(0.13%) with Vcerveld and Lange-Nielsen syndrome (JLNS), 2 (0.27%) cases each with Usher and Renal

Table-IX : Incidence of Syndromes in children based on the organ/system involved.

Sl. No.	Organ involved	No. of cases	Percentage
1.	Ocular	31	22.46
2.	Skeletal	20	14.49
3.	Dental	15	10.86
4.	Dermal	9	6.52
5.	Metabolic	4	2.89
6.	Craniofacial	3	2.17
7.	Neurologic	3	2.17
8.	Renal	2	1.50
9.	Cryptogenic	51	36.9
Total		138	100

Table- X : Incidence of well-recognized syndromes in children with hearing loss.

Sl. No.	Syndrome	No. of Cases	Incidence in deaf Children (%)
1.	JLNS (Jervel and Lange-Nielsen Syndrome)	1	0.13
2.	Renal	2	0.27
3.	Usher	2	0.27
4.	Earpits	3	0.40
5.	Pendred	3	0.40
6.	Mental retardation	3	0.40
7.	Severe-Myopia	5	0.67
8.	Waardenberg	5	0.67
Total		24	3.21

syndromes, 3 (0.4%) cases each with Earpits syndrome, Pendred syndrome and Mental Retardation, and 5(0.67%) cases each with Waardenberg Syndrome and severe-Myopia.

DISCUSSION

Deafness is defined as the disability of using hearing as a primary channel for receiving speech even with amplification[3]. Hearing loss hence affects various related aspects of child's overall development as the development of speech and language is impaired. It may also hinder and impair the child's social and emotional relationship[9].

Deafness is found to occur in about 1 per thousand live births, and atleast 50% of congenital deafness is

hereditary[10]. Over 70% of congenital sensorineural hearing impairment are non-syndromic while about 30% are associated with other defects[8].

In the present study, out of 743 children below 14 years with hearing loss, 138 (18.57%) were found with syndromic deafness which is comparatively lower when compared with the earlier study[8]. 81.43% (n=605) constituted for isolated deafness.

Age-wise analysis of the children in the present study showed that majority of children belonged to the age group of 12 to 14 years (26.8%) followed by 6-9 (21.74%), 3-6 (21.01%), 9-12 (16367%) and 0.3 (13.77%) age groups. However, the hearing loss in all these children was diagnosed before 5 years of age. Elango et al in their study found that hearing loss was diagnosed only in 19% of children by the age of 2 years[11]. Tschopp revealed that hearing handicap was recognized at an average age of two years[12].

Clifton and Swart in a study with 169 children found the hearing-disability in 95% of them before the age of 3 years and majority of them were boys[14]. In the present study, prevalence of deafness in boys accounted for 54.35% and 45.65% in girls. The sex ratio in childhood deafness analyzed by Cremers et al also showed unexplained male predominance[15]. The population of deaf people consisted of an average of 54% men and 46% women[15], which was similar to that of our findings. Zazouk correlated an increased risk of hearing impairment with male children, consanguinity, lower socio- economic level and family history for deafness[16].

In the present study, it is found that majority of parents of children with syndromic deafness were from lower-middle class (42.03%). The middle-class people constituted 31.88% followed by low socio economic status (21.74%). Only 4.35% belonged to higher socio-economic group. Bafaqeeh et al in their study to find the relevant demographic factors in hearing loss found that low socio-income and parental education were also important risk factors in deafness[6]. However, in the present study, the percentage of literate parents (52.11%) was greater that that of illiterates (47.89%).

Distribution based on the religion of the child represent that majority of them belonged to the Hindu community (87.68%) followed by Muslims (10.15%) and Christians (2.17%). This only reflects the percentage of patients attending the clinic. There is no evidence to support that a particular religion is at higher risk for deafness.

In the current study, it was found that 78 (57.52%) out of 138 children were products of consanguineously married couples while 60 (43.48%) were from non-consanguineous parents. Zlotogora and Barges in their study found that children born to consanguineously married couples were at a higher risk for deafness[17]. Since the consanguineous marriages are very common in the Hindus and Muslims, the risk of having deafness is greater in them. Zazouk also observed that consanguinity and positive family history were risk parameters for hearing loss[16].

In the present study, it is found that 15.22% (n=21) of children with hearing loss had a familial history for deafness and 13.77% (n=19) acquired hearing loss due to prenatal, perinatal and postnatal problems and /or infections. In 71.01% (n=98) the etiology for hearing loss is not known. Studies by Clifton and Swart reported one-tenth of children with a family history had deafness. They further observed in 30% of cases no apparent cause could be established[14]. Elango et al observed acquired deafness in 35% of cases. They could not establish the cause in 28.4% of the cases with hearing loss[11]. In the present study , the deafness with unknown etiology is greater than the earlier studies[11,14].

Sutton and Rowe observed high proportion of craniofacial abnormality (24%) in children with hearing impairment and counted it to be a risk factor for deafness[7]. However, in the study carried out by Leguire et al, 48.7% of children with hearing loss were found with ocular abnormality.

Beighton et al in their study observed that only 10% of syndromes were well recognized genetically and 20% had familial undifferentiated deafness. Acquired deafness constitured for 40% while cryptogenic for 30%. In the present study, genetically well-recognized syndromes were observed in 24 (17.39%) out of 138 cases and cryptogenic deafness in 51 (36.9%).

The incidence of Jervell and Lange-Nielsen Syndrome (JLNS), a rare cardio-auditory disorder is found to vary from 0.1% and 1% of children with congenital deafness. In the present study JLNS was observed in only one case (0.13%).

Usher syndrome, an autosomal recessive sensory defect associated with deafness and visual loss, affects 1 in 25,000 and is a most frequent cause of hereditary deaf-blindness in humans[22]. In the present study, 2 (0.27%) cases each out of 743 children with hearing loss were reported with Usher syndrome and renal dysfunction.

Three (0.40%) cases each out of 743 children with hearing loss were found with Earpits syndrome, mental retardation and Pendred syndrome Pendred syndrome accounts for 5% of the congenitally deaf population.

5 (0.67%) cases each out of 743 children with deafness were reported with Waardenberg syndrome and severe myopia. Waardenberg syndrome is a commonly inherited autosomal dominant syndrome. It is estimated to occur in 1.44 to 2.05 per 100,000 in general population and more than 1% in congenitally deaf.

The overall study has shown the epidemiological factors involved in the causation of deafness. Besides, the study threw light on different syndromes associated with deafness of which some are acquired and the remaining are inherited. Thus there is a need to carry out extensive studies on epidemiology of deafness and also to identify the genetically inherited syndromes so that necessary genetic counseling can be offered to families at high risk in order to prevent the birth of such children.

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