

Childhood Congenital Deafness and Its Association with Consanguineous Marriage: A Comparative Study on Hereditary Awareness

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ABSTRACT

Objective: To find the association between consanguineous marriages and congenital sensorineural deafness.

Study Design: Comparative cross-sectional study.

Place and Duration of Study: PAC (Pakistan Aeronautical Complex) Hospital, Kamra, Pakistan, from Feb 2022 to Mar 2023.

Methodology: A total of 120 cases were selected with an age range of 5-15 years. They were divided into two groups of 60 cases each. Group-1 had cases of congenital sensorineural hearing loss, and Group-2 had cases of normal hearing threshold. Hearing loss due to other acquired diseases was not included; the Chi-square test was applied, and an odds ratio was calculated.

Results: Among 120 cases 74(61.66%) were males and 46(38.33%) were females, 68(56.6%) children's parents were blood related and 52(43.3%) children parents had no blood relation. Out of the blood-related consanguineous marriages, 48(70.56%) were 1st cousins and 20(29.41%) were 2nd cousins. In Group-1, parents of 48(80%) cases were blood relatives and 12(20%) were not blood-related. In Group-2, parents of 20(33.3%) cases were blood relatives, and parents of 40(66.6%) cases were not blood-related. The p -value <0.001 was considered significant as the p -value is <0.05 , and an odds ratio value of 8 indicates increased occurrence of congenital hearing loss in consanguineous marriages.

Conclusion: Consanguinity and cousin marriage are strongly related to congenital childhood sensorineural deafness; further studies are required to raise public awareness about this social issue.

Keywords: Awareness, Consanguinity, Congenital Hearing Loss.

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INTRODUCTION

Hearing loss is one of the significant health issues globally. Infections, environmental factors, genetic factors, and birth defects can all contribute to hearing loss. Causes of childhood hearing loss include birth asphyxia, meningitis, suppurative otitis media, measles, mumps, head trauma, ototoxic medications, etc. In literature, consanguinity is also one of the major factors for congenital hearing loss. Consanguinity means the marriage of blood-related individuals who have a recent common ancestor, like first cousins, second cousins, and close relatives.¹ Hearing loss is one of the major causes of referral to the ENT outpatient department and can present at all ages and in both sexes. Hearing loss can be congenital, perinatal, and acquired. The prevalence of consanguineous marriage varies globally and is related to social customs, including arranged marriage within families and public unawareness of the adverse effects of such practice.² Genetic and congenital

disorders can contribute to newborn mortality, morbidity, and disability. Congenital hearing loss is one of the common chronic conditions in children and a major cause of referral to the ENT outpatient department. Congenital hearing loss in newborns has strong adverse effects on individuals' learning, mental, linguistic, educational, and psychological development.^{3,4} Congenital hearing loss is not always present at birth, but the child may develop it later in life also.⁵ Hereditary hearing loss accounts for more than fifty percent of sensorineural hearing loss, which limits exposure to environmental sounds and prevents normal auditory and speech development.⁶ Sense of hearing is a gift to mankind, and speech is closely related to hearing because those who have impaired hearing often have a speech delay.⁷ Consanguinity is associated with autosomal recessive inheritance and increases the chances of defective gene sharing in the offspring, which in turn increases the risk of occurrence of hearing loss occurring in the family.⁸ In Pakistan, consanguinity in one study has been reported to be 50-70 percent and is considered a major cause of congenital hearing loss.⁹

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Awareness of parents about consanguinity and congenital hearing loss is important for the success of early identification and rehabilitation programs. The study aims to identify and highlight the association between consanguineous marriage and congenital hearing loss.

METHODOLOGY

A comparative cross-sectional study was carried out at PAC (Pakistan Aeronautical Complex) Hospital, Kamra, Pakistan, from Feb 2022 to Mar 2023 after approval from the ethical committee, letter reference number 28 dated 22 Feb 2022. The sampling was done by a non-probability convenience method. The sample size of 118 was calculated from the World Health Organization (WHO) calculator by using a 95% confidence level and a 9% margin of error, and the prevalence of sensorineural hearing loss was 54.3%.¹⁰ Initially, 139 cases were selected; 19 cases did not report for investigations, so finally 120 cases were selected.

Inclusion Criteria: Children with normal hearing threshold and congenital hearing loss of either gender with age range of 5-15 years, presenting in the ENT outpatient department of PAC Hospital Kamra.

Exclusion Criteria: Hearing loss due to other environmental factors like Infections, ototoxic drugs, prematurity, birth asphyxia, meningitis, kernicterus, syndromic child, head injury, and other acquired diseases were excluded from the study.

The cases were divided into two groups. Group-1 consisted of 60 children with congenital sensorineural hearing loss, and Group-2 consisted of 60 children with normal hearing thresholds. Consent and willingness from parents were obtained, and they agreed to participate in the study and get their children screened for hearing thresholds. A complete medical, family, prenatal, natal, and post-natal history was obtained from the parents; details and information were gathered on whether they were blood relatives (first cousin who shared a grandparent or second cousin who shared great-grandparents). History of hearing loss in the family, number of children with congenital hearing loss (SNHL) since birth, education, economic status and income, awareness about risk factors of consanguineous marriage on congenital hearing loss in children before their marriage were also inquired. A detailed questionnaire was made to get the information from parents, available records, and for data collection. Confounding factors were catered for by matching

both groups. Complete ENT examination and audiological investigations, including tympanometry, brain-evoked response audiometry (BERA), and pure tone audiometry, were obtained in cases of both groups. Severe and profound hearing loss were noted according to the WHO classification of hearing loss. Normal hearing threshold is considered between -10 and 15 dB (decibel), Severe hearing loss is a hearing threshold between 71- 90 dB, and Profound hearing loss is a hearing threshold of 91db or more.

The data were statistically analyzed using the Statistical Package for the Social Sciences version 24. Categorical variables are presented as percentages and as frequencies for all the categories and odds ratios were calculated. The two groups were compared with respect to the relationship between congenital hearing loss and cousin marriages. A chi-square test was applied, and a p -value ≤ 0.05 was considered significant.

RESULTS

Among 120 children, 74 (61.66%) were males, and 46(38.33%) were females. Mean age of cases was 8.01+2.50 years. Out of 120 cases, 68(56.6%) children's parents were blood-related, and 52(43.3%) children's parents had no blood relation. In the blood-related consanguineous marriages, 48(70.58%) were first cousins and 20(29.41%) were second cousins. In Group-1, 14(23.3%) children had profound sensorineural hearing loss, while 46(76.6%) children had severe sensorineural hearing loss Table-I. We also observed that in Group-1, in the parents of 21(36%) children, family history of childhood hearing impairment was present; parents of 105(88%) children belonged to the low socioeconomic group; and parents of 102(85%) children did not know about the effects of consanguineous marriage on congenital hearing loss in children.

Table-I: Type of Marriage and Degree of Consanguinity and Hearing Loss (n=120)

Parameters	Sub-types	n (%)
Types of Marriages (n=120)	Consanguineous	68(56.60%)
	Non-Consanguineous	52(43.30%)
Degree of Consanguinity (n=68)	1st Cousin	48(70.58%)
	2nd Cousin	20(29.41%)
Degree of Hearing Loss (HL) in Group-1 (n=52)	Profound HL	14(23.30%)
	Severe HL	46(76.6%)

In this study, we also found that in Group-1, parents of 48(80%) children were blood relatives and 12(20%) were not blood-related. In Group-2, parents of 20(33.3%) children were blood relatives and parents of 40(66.6%) children were not blood-related Table-II.

Table-II: Blood Relationship Between Parents and Association with Congenital Hearing Loss (n=120)

Study Variables	Group-1 Congenital Hearing Loss (n=60)	Group-2 No Hearing Loss (n=60)	p-Value
Consanguineous Marriage	48 (80%)	20 (33.3%)	< 0.001
Non- Consanguineous Marriage	12 (20%)	40 (66.6%)	< 0.001

The *p*-value is <0.001, which is considered significant because it is < 0.05. An odds ratio of 8 indicates an increased occurrence of congenital hearing loss in consanguineous marriage.

DISCUSSION

The present study included 120 children, of whom 74(61.66%) were males, and 46(38.33%) were females, with a mean age of 8.01 ± 2.50 years. Overall, 68(56.6%) children had parents in a consanguineous marriage, including 48(70.58%) first-cousin and 20(29.41%) second-cousin marriages. Parental consanguinity was significantly associated with congenital sensorineural hearing loss (80% vs. 33.3%; $p < 0.001$; OR=8).

Consanguinity and its association with congenital deafness has been highlighted in previous studies and considered a major factor in congenital deafness in various communities of the world. Consanguineous marriages are usually practiced for cultural reasons and to strengthen family ties, which has been reported previously in many neighboring populations.¹¹ Parental knowledge about early identification of hearing loss and timely and appropriate intervention has lifelong consequences for a hearing-impaired child. Consanguineous marriage is not prohibited, but the health issues and hereditary disease which occur because of this type of marriage can affect the well-being of generations to come and ultimately the health care system. Consanguineous marriage is referred to as a marital union between individuals who belong to the same genealogical kin.¹²

Ghanim *et al.*, indicated that worldwide, a huge amount of the healthcare system resources is spent on patients with genetic or congenital disorders. The incidence of consanguinity is higher in the developing

world than in developed countries.¹³ In a study by Miller *et al.*, consanguinity was found to be in 56% of the population¹⁴; similarly, in another study by Khabori *et al.*, 70% of the children with congenital hearing loss were found to have consanguineous parents.¹⁵ Batool *et al.*, from Pakistan, reported that congenital hearing loss due to consanguineous marriages was found to be approx 73%.¹⁶ In comparison, in this study we found that 68(56.6%) of the children have parents with consanguineous marriages and 52(43.3%) had no blood relation. In the consanguineous marriage group, 48(70.59%) were first-cousin marriages and 20(29.41%) were second-cousin marriages. In general, first-, second-, and third-degree consanguinity have 50% or more, 25%, and 12.5% chances, respectively, to affect the offspring regarding hearing loss. In our study in Group-1, i.e children with congenital hearing loss, 48(80%) parents had consanguineous marriage and 12(20%) had non-consanguineous marriage. In Group-2, children with no hearing loss, parents of 20 (33.3%) cases had consanguineous marriage and parents of 40(66.6%) cases had non-consanguineous marriage; the *p*-value is <0.001, considered significant as *p*-value is < 0.05, which is comparable to other studies mentioned above. We also found that in Group-1, 14(23.3%) children have profound sensorineural hearing loss while 46(76.6%) children have severe sensorineural hearing loss.

History of childhood hearing impairment in parents is another important factor linked with consanguinity in causing genetic hearing impairments¹⁷; in our study, 21(36%) of the parents had a positive family history of childhood hearing impairments. It was also found that 105(88%) of the patients belonged to the low socioeconomic class, and 102(85%) of the parents were not aware of the risks of consanguineous marriage regarding hearing loss in children. The results of our study are comparable to the study by Al-Jabri *et al.*, who highlighted the significance of this social health problem and implementing preventive measures to increase public awareness regarding congenital hearing loss due to consanguineous marriages.¹⁸ The genetic pool also gets constrained by consanguineous marriages and is a compounding factor for hereditary hearing loss, which has been highlighted by Klau *et al.*¹⁹

In the literature, around 80% of non-syndromic congenital hearing loss is due to autosomal recessive transmission and 20% due to autosomal dominant

transmission.²⁰ Such health issues and diseases resulting from consanguineous marriages can negatively impact the economy and the healthcare system. As the practice of consanguineous marriage among the Pakistani population is high, special steps are required to increase public awareness in this regard. Social changes associated with urbanization have not positively affected the practices of consanguineous marriage among the Muslim population.²¹

When hearing improvement is unlikely, early use of hearing aids or cochlear implants can support language development. Otoacoustic emission enables early detection in newborns. Effective policies and education about consanguineous marriage and hereditary disease can reduce genetic disease burden and improve healthcare resource use.

LIMITATION OF STUDY

This study has a limited sample size; further research and larger population samples are needed to study public awareness about congenital hearing loss and its association with consanguinity, to highlight the role of public health professionals and campaigns in raising public awareness of this social issue.

CONCLUSION

Consanguinity is strongly related to congenital childhood sensorineural hearing loss; first cousin consanguinity is one of the major factors in this regard. Further research, a larger population sample, and more efforts are required for raising public awareness regarding this preventable social issue.

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Authors' Contribution

Following authors have made substantial contributions to the manuscript as under:

ASU: Data acquisition, data analysis, critical review, approval of the final version to be published.

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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