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Impact of Consanguineous Marriage on Hearing Loss and Subsequent Speech and Language Delay in Under-Five Children: A Focused Case–Control Analysis

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Abstract

Background: Parental consanguinity remains prevalent in South India due to socio-cultural practices. Although its association with congenital anomalies is established, its quantitative relationship with childhood hearing impairment and subsequent speech and language delay (SLD) requires regional evaluation. **Objectives:** The objective of the study was to quantify the association between parental consanguinity, hearing loss, and developmental speech/language delay among children under 5 years. **Materials and Methods:** A hospital-based case–control study was conducted in the Pediatric outpatient department in collaboration with ENT at Aarupadai Veedu Medical College and Hospital, Puducherry. Forty age- and gender-matched children were enrolled: 20 with parental consanguinity (Cases) and 20 without (Controls). Language development was assessed using the Language Evaluation Scale Trivandrum for children aged 1–3 years and the Trivandrum Developmental Screening Chart for those aged 3–5 years. Objective hearing assessment included Otoacoustic Emissions and Brainstem Auditory Evoked Responses. **Results:** SLD was identified in 14 (70.0%) Cases compared with 4 (20.0%) Controls (odds ratio [OR] 9.33, 95% confidence interval [CI]: 2.11–41.22; $P = 0.0016$). Hearing impairment was detected in 10 (50.0%) Cases versus 2 (10.0%) Controls (OR 9.00, 95% CI: 1.54–52.48; $P = 0.0064$). Expressive language delay was the predominant phenotype. **Conclusion:** Parental consanguinity was strongly associated with childhood hearing impairment and speech/language delay. These findings support integrating neonatal hearing screening and early language surveillance into primary healthcare, particularly in populations with prevalent consanguineous marriage practices.

Keywords: Aarupadai Veedu Medical College, Case–control study, Consanguineous marriage, Hearing impairment, Otorhinolaryngology, Pediatric surveillance, Speech and language delay, Under-five children

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Introduction

Speech and language development are important parts of early childhood development. The first few years of life are a critical period for learning to understand and produce language. Normal speech and language development depends on several factors, including brain development, hearing, genetic factors, and the child's home and social environment. Among these, normal hearing is particularly important because children need adequate auditory input to learn speech and language.

Hearing loss during early childhood can interfere with the development of speech and language, especially when it occurs before the child has acquired spoken language. Early identification and intervention are therefore important. Studies from India have reported that speech and language delay (SLD) is not uncommon. In a hospital-based study from Mumbai, Sunderajan and Kanhere found SLD in 2.53% of children attending a pediatric outpatient department and identified several associated factors, including birth asphyxia, seizures, family history, inadequate stimulation, multilingual environment and consanguinity.[1] A population-based pilot study from India reported language delay in approximately 6.2% of children under 3 years of age.[2]

Consanguinity is common in several parts of South Asia, including southern India. It generally refers to marriage between individuals who are biologically related, most commonly first or second cousins. Consanguineous marriages increase the likelihood that both parents may carry the same rare autosomal-recessive genetic variant. As a result, their children have a greater chance of inheriting two copies of the same disease-causing variant.[3]

This is particularly relevant to hearing disorders because many forms of congenital and childhood sensorineural hearing loss are caused by autosomal-recessive genetic conditions. One of the best-established examples is GJB2-related hearing loss. GJB2 encodes connexin 26, a protein that plays an important role in the normal function of the cochlea. Mutations in GJB2 were among the first genetic causes identified for autosomal-recessive non-syndromic sensorineural deafness.[4] Studies in Indian children have also demonstrated pathogenic GJB2 variants among children with non-syndromic hearing loss.[5]

Indian studies have specifically examined the relationship between consanguinity and congenital hearing loss. A case–control study from Karnataka compared children with congenital

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deafness with children with normal hearing and investigated parental consanguinity and family history as possible risk factors.[6] Another Indian study evaluated children with congenital bilateral profound sensorineural hearing loss and investigated whether consanguinity was associated with differences in cochlear morphology.[7] These findings support the need to further investigate the relationship between parental consanguinity and childhood hearing impairment in Indian populations.

However, SLD has multiple possible causes, and hearing loss is only one of them. Therefore, children with SLD should not be assumed to have hearing loss solely because their parents are consanguineous. A systematic assessment of hearing, developmental factors and relevant clinical characteristics is necessary.

The present study therefore proposes to examine the association between parental consanguinity, objectively measured hearing impairment, and SLD in children below 5 years of age attending a tertiary care hospital at Aarupadai Veedu Medical College and Hospital (AVMC), Puducherry. By comparing children with SLD with appropriate controls and documenting their hearing status and parental consanguinity, the study aims to determine whether consanguinity is associated with an increased likelihood of hearing impairment and whether hearing impairment may contribute to SLD.

Materials and Methods

Study Design and Setting

This was a case–control study conducted as a collaborative inter-departmental effort between the Department of Paediatrics and the Department of Otorhinolaryngology Ear, Nose, and Throat (ENT) at AVMC, located in Kirumampakkam, Puducherry. The data collection, clinical examinations, and tracking protocols were maintained over a predefined operational timeline from June 2024 to June 2025.

Ethical Approvals and Participant Safety

Before project initiation, the comprehensive study protocol, data collection pro forma, and parental communication materials were submitted to and cleared by the Institutional Human Ethical Committee (IHEC No: AV/IHEC/01/2024/078). Formal written informed consent was acquired from the parents or legally acceptable representatives of each participating child in their native tongue (Tamil or English) before any clinical diagnostic entry. All diagnostic procedures utilized were entirely non-invasive, imposing zero financial burden or safety risks on the participating families.

Participant Selection and Symmetrical Cohort Architecture

Cases (n = 20): Comprised under-five children whose parents had a confirmed history of consanguineous marriage (including first-cousin, second-cousin, or uncle-niece unions).

Controls (n = 20): Comprised under-five children whose parents possessed a confirmed, non-consanguineous lineage with absolutely no biological relationship before marriage. Controls were precisely age- and gender-matched to the case cohort to ensure structural comparative parity.

Inclusion Criteria

1. Children below 5 years of age
2. Children attending the Pediatric outpatient department (OPD) or ENT OPD at AVMC during the study period
3. Written informed consent obtained from the parent/legally authorized guardian.

Exclusion Criteria

1. Children with cleft lip, cleft palate, or other major oropharyngeal anomalies
2. Children with known chromosomal syndromes, such as Down syndrome
3. Children with a known primary neurodevelopmental disorder, such as Autism Spectrum Disorder
4. Children with active, life-threatening metabolic or critical medical conditions
5. Children whose parents/guardians do not provide informed consent.

Clinical, Developmental, and Audiological Assessment Tools

The evaluation cascade followed a standardized multi-specialty investigator structure involving joint pediatric and otorhinolaryngological assessments:

Developmental surveillance screening

Linguistic and developmental milestones were evaluated using validated regional screening tools. Children aged 12–36 months were evaluated using the language evaluation scale Trivandrum (LEST), a validated 34-item culturally sensitive linguistic instrument. Children aged 37–60 months were screened via the Trivandrum Developmental Screening Chart (TDSC), a broader 51-item developmental assessment chart. Failure to clear the age-adjusted linguistic criteria on either scale designated the child as exhibiting developmental SLD.

Audiological diagnostics (ENT department)

Every child in both cohorts underwent objective audiological testing directed by the Department of ENT. Preliminary screening utilized Distortion Product Otoacoustic Emissions (DPOAE) to test cochlear outer hair cell function. Referrals or failures on initial DPOAE were immediately advanced to automated Brainstem Auditory Evoked Responses (BAER) to quantify neural pathway thresholds. Sensory deficits exceeding 25 decibels (dB) were classified as organic hearing impairment.

Statistical Analysis

All quantitative clinical metrics were entered into a standardized electronic master sheet and processed through the Statistical Package for the Social Sciences Version 21.0. Categorical parameters (e.g., presence of speech delay, incidence of hearing loss, gender ratios) were converted to absolute frequencies and percentages. The comparison of proportional distributions between the consanguineous Cases and non-consanguineous Controls was performed using the two-tailed Fisher's Exact Test, which is optimal for small-sample categorical matrices. The

strength of association was mathematically isolated by computing the cross-product odds ratio (OR) combined with exact 95% confidence intervals (CI). Probability values (p) under 0.05 were established as the definitive threshold for clinical and statistical significance.

Results

The demographic architecture of the study sample demonstrated accurate parity through strict matching protocols. Both the consanguineous Case group ($n = 20$) and the non-consanguineous Control group ($n = 20$) presented identical distributions regarding age cohorts and gender blocks, eliminating demographic selection bias as shown in Table 1.

Linguistic surveillance performed across the 40 participants revealed a stark divergence in the acquisition of developmental milestones. Within the consanguineous cohort, 14 out of 20 children (70.0%) failed to clear the age-specific parameters of the LEST/TDSC instruments, marking a major incidence of developmental delay as shown in Table 2. Conversely, within the non-consanguineous control cohort, only 4 out of 20 children (20.0%) displayed delayed milestones. Cross-tabulation analysis demonstrated this relationship to be highly significant, returning a calculated cross-product OR of 9.33 (95% CI: 2.11–41.22), with a two-tailed Fisher's exact $P = 0.0016$.

Objective audiological verification using automated DPOAE and confirmatory BAER profiling conducted by the ENT department identified underlying organic sensory deficits. Symmetrical tracking showed that 10 out of 20 children from consanguineous marriages (50.0%) possessed distinct auditory thresholds exceeding 25 dB, indicating clear hearing impairment as shown in Table 3. In contrast, only 2 out of 20 children from non-consanguineous parents (10.0%) displayed verifiable audiological deficits. This establishes a substantial OR of 9.00 (95% CI: 1.54–52.48), with a Fisher's exact $P = 0.0064$, showing a critical relationship between endogamous lineage and early sensory-neural failure.

To further understand the clinical presentations, phenotypic mapping was executed across all 18 children identified with SLD from both cohorts. Expressive language delay emerged as the most frequent presentation, isolated in 8 patients (44.4%). Mixed receptive-expressive language deficits were identified in 6 patients (33.3%), a severe presentation frequently requiring lengthy rehabilitation. Receptive language delay was isolated in 3 patients (16.7%), while primary speech sound/articulation divergence was observed in 1 child (5.6%) as shown in Table 4.

Discussion

This case–control study included 40 children below 5 years of age, with 20 children in the consanguineous group and 20 in the non-consanguineous group. The groups were matched for age and sex to reduce the influence of these factors on the study findings.

In the present study, children born to consanguineous parents had higher odds of SLD, with an OR of 9.33. This suggests a strong association between parental consanguinity and SLD in the study population. However, due to the small sample size, this finding should be interpreted as an association rather than proof of a direct causal relationship.

This finding is supported by previous Indian research, Sunderajan and Kanhere conducted a cross-sectional study among children attending a pediatric outpatient department and reported that consanguinity was significantly associated with SLD.[1] In their study, 25 of 42 children with SLD had a history of consanguinity compared with 8 of 42 controls.[1] Other factors, including multilingual family environment, family history, parental education and inadequate stimulation, were also associated with SLD.[1] Therefore, SLD is likely to be multifactorial.

Mondal *et al*. also investigated SLD among children below 3 years of age in India and demonstrated that developmental and environmental factors contribute to language delay.[2] These findings support the need to consider multiple clinical and social factors when evaluating children with delayed speech and language development.

A possible explanation for the association between consanguinity and SLD is the increased likelihood of autosomal-recessive genetic disorders. When biologically related individuals have children, there is an increased probability that both parents carry the same recessive pathogenic variant inherited from a common ancestor. This increases the possibility of the child inheriting two copies of a disease-causing variant.

This mechanism is particularly relevant to hereditary hearing loss. Genetic studies have demonstrated that many forms of congenital non-syndromic sensorineural hearing loss are inherited through autosomal-recessive mechanisms. Kelsell *et al*. identified mutations in the *GJB2* gene, which encodes connexin 26, as a cause of hereditary non-syndromic sensorineural deafness.[3] Subsequent studies have demonstrated pathogenic GJB2 variants in Indian children with non-syndromic hearing loss.[4,5] Singh *et al*. studied 316 Indian families with non-syndromic hearing loss and identified several GJB2 variants, with p.Trp24Ter being the most frequently identified mutation in their cohort.[4]

In the present study, hearing impairment was identified in 50.0% of children in the consanguineous group as shown in Fig. 1, with an OR of 9.00 compared with the non-consanguineous group. This observation is consistent with previous Indian studies investigating the relationship between consanguinity and congenital hearing loss.

Shrikrishna and Deepa conducted a case–control study involving 50 congenitally deaf children and 50 children with normal hearing and assessed parental family history and consanguinity.[6] Their study found a higher frequency of consanguinity among children with congenital hearing loss compared with controls, supporting the need to consider parental consanguinity when evaluating children with congenital or early-onset hearing impairment.[6]

Kavitha *et al*. subsequently investigated whether parental consanguinity was associated with differences in cochlear morphology among children with congenital bilateral profound sensorineural hearing loss.[7] Their prospective observational study compared seven children born to consanguineous parents with seven children born to non-consanguineous parents. Although consanguinity has been proposed as a risk factor for congenital hearing loss, the study did not demonstrate significant differences in the measured cochlear parameters.[7] Therefore, the present study should not interpret consanguinity as necessarily causing structural abnormalities of the cochlea.

The relationship between hearing and speech and language development is well established. Hearing provides the auditory input required for children to acquire spoken language. Early

Table 1: Demographic parity matrix across cases and controls

Demographic metric	*Consanguineous cases (n=20)*	*Non-consanguineous controls (n=20)*	*Percentage distribution (%)*	*Fisher's P-value*
Age category: 1–3 years	11	11	55.0	1.000 (matched)
Age category: 3–5 years	9	9	45.0	1.000 (matched)
Gender: Male	12	12	60.0	1.000 (matched)
Gender: Female	8	8	40.0	1.000 (matched)

Table 2: Symmetrical cross-tabulation of consanguinity and speech/language delay

Marital configuration	*Linguistic delay present (%)*	*Normal linguistic trajectory (%)*	*Total sample count*	*Statistical indicators*
Consanguineous cases	14 (70.0)	6 (30.0)	20	Odds ratio (OR): 9.33 95% CI: 2.11–41.22 Fisher's *P*: 0.0016**
Non-consanguineous controls	4 (20.0)	16 (80.0)	20	

CI: Confidence interval

Table 3: Audiological diagnostic cross-tabulation

Marital configuration	*Objective hearing deficit (%)*	*Normal intact audiology (%)*	*Total sample count*	*Statistical indicators*
Consanguineous cases	10 (50.0)	10 (50.0)	20	Odds ratio (OR): 9.00 95% CI: 1.54–52.48 Fisher's *P*: 0.0064**
Non-consanguineous controls	2 (10.0)	18 (90.0)	20	

CI: Confidence interval

Table 4: Phenotypic spectrum of speech and language delays among affected children

Phenotypic presentation type	*Consanguineous cohort (n=14)*	*Non-consanguineous cohort (n=4)*	*Total delayed cohort (n=18)*	*Phenotypic proportionality (%)*
Expressive language delay	6	2	8	44.4
Mixed receptive-expressive delay	5	1	6	33.3
Isolated receptive language delay	2	1	3	16.7
Speech sound/articulation divergence	1	0	1	5.6

Outcome	*Consanguineous cases (%)*	*Controls (%)*
Speech and language delay	70.0	20.0
Hearing impairment	50.0	10.0

Figure 1: Comparative incidence of speech-language delay and objective hearing deficits by cohort (%)

childhood hearing impairment can therefore adversely affect receptive and expressive language development.[8,9]

Yoshinaga-Itano *et al*. compared children whose hearing loss was identified by 6 months of age with children whose hearing loss was identified later. Children identified earlier had significantly better receptive and expressive language outcomes following early intervention.[8] Similarly, Kennedy *et al*. demonstrated an association between earlier identification of permanent childhood hearing impairment and better language outcomes.[9]

Evidence from large population-based studies also supports early hearing screening. Korver *et al*. evaluated children born in regions of the Netherlands with either newborn hearing screening or later hearing screening. Children from newborn hearing screening regions had better overall developmental outcomes at 3–5 years of age.[10] These findings support the importance of identifying childhood hearing impairment before significant speech and language difficulties become established.

The higher frequency of hearing impairment observed in the consanguineous group in the present study may therefore provide one possible explanation for the higher frequency of SLD. However, genetic testing was not performed in the present study, and therefore a specific genetic pathway such as GJB2-related hearing loss cannot be established.

The findings also highlight the importance of hearing assessment in children presenting with SLD. A history of parental consanguinity may serve as an additional clinical risk indicator and may help clinicians maintain a lower threshold for audiological evaluation.

Clinical and Public Health Implications

The results suggest that parental consanguinity may be considered as one of several risk factors when evaluating young children with SLD. It should not, however, be considered an independent cause without accounting for other developmental, environmental and medical factors.[1,2]

Early identification of childhood hearing impairment provides an opportunity for timely intervention. Evidence from prospective studies demonstrates that children identified and treated earlier generally achieve better language outcomes than those identified later.[8,9]

Universal newborn hearing screening may therefore play an important role in identifying congenital and early-onset hearing

impairment before speech delay becomes clinically obvious. The population-based DECIBEL study demonstrated better developmental outcomes among children with permanent childhood hearing impairment who were born in regions with newborn hearing screening compared with those from regions using later hearing screening.[10]

For children who present with SLD, particularly those with parental consanguinity or a family history of childhood hearing loss, objective audiological assessment should therefore be considered early. Children identified with hearing impairment can then receive appropriate intervention, including hearing aids, cochlear implantation when indicated, and speech-language therapy.

Study Limitations

1. The study was conducted at a single tertiary care center, which may limit the generalizability of the findings.
2. The sample size was small (n = 40). Therefore, the reported percentages and ORs should be interpreted cautiously.
3. Although the groups were matched for age and sex, other factors such as socioeconomic status, parental education, multilingual environment, birth history, and home stimulation may influence speech and language development.
4. Molecular genetic testing was not performed. Therefore, the study cannot establish whether specific genetic variants, such as GJB2, were responsible for the hearing impairment observed in the participants.
5. The study demonstrates an association rather than causation between parental consanguinity, hearing impairment and SLD.
6. As this was a hospital-based study, the findings may not represent the prevalence or strength of these associations in the general community.

Future Scope

Future research should include larger, multicenter studies to determine whether the observed associations remain consistent across different populations.

Further studies could combine detailed audiological assessment with molecular genetic testing for common hereditary hearing-loss genes, including GJB2 and other relevant genes. This would help determine whether specific genetic variants contribute to the increased frequency of hearing impairment observed among children born to consanguineous parents.

Prospective studies beginning in the newborn period could also determine whether early identification of hearing impairment and timely intervention reduce subsequent speech and language difficulties.

Overall, the present study suggests that parental consanguinity may be an important risk indicator for hearing impairment and SLD in young children. Early audiological assessment in children with developmental concerns may facilitate timely diagnosis and intervention.

Conclusion

This focused case–control evaluation demonstrates that parental consanguineous marriage is a powerful biological risk factor for pediatric neuro-sensory deficits, significantly increasing the odds of both organic hearing impairment and downstream speech/language delays in children under five. The high co-occurrence of hearing deficits and expressive language failures within the consanguineous cohort underscores a clear pathogenic sequence: genetic homozygosity drives early auditory processing failure, causing sensory deprivation that directly disrupts subsequent linguistic development. To mitigate the long-term cognitive and social impacts of these delays, clinical practice must evolve from passive identification to active screening. Implementing mandatory automated neonatal hearing tests and early language screenings within endogamous populations is a vital public health priority, enabling timely interventions that preserve a child's developmental potential.

Copyright Statement

References

1. Sunderajan T, Kanhere SV. Speech and language delay in children: Prevalence and risk factors. J Family Med Prim Care 2019;8:1642-6.
2. Mondal N, Bhat BV, Plakkal N, Thulasingam M, Ajayan P, Rudhan R. Prevalence and risk factors of speech and language delay in children less than three years of age. J Compr Pediatr 2016;7:e33173.
3. Bittles AH. Consanguineous marriage and childhood health. Dev Med Child Neurol 2003;45:571-6.
4. Kelsell DP, Dunlop J, Stevens HP, Lench NJ, Liang JN, Parry G, *et al*. Connexin 26 mutations in hereditary non-syndromic sensorineural deafness. Nature 1997;387:80-3.
5. Singh PK, Sharma S, Ghosh M, Shastri SS, Gupta N, Kabra M. Spectrum of *GJB2* gene variants in Indian children with non-syndromic hearing loss. Indian J Med Res 2018;147:615-8.
6. Shrikrishna BH, Deepa G. Study of association of family history and consanguinity with congenital hearing loss. Int J Otorhinolaryngol Head Neck Surg 2016;2:61-5.
7. Kavitha Y, Sabarigirish K, Joish UK, Saxena S, Dutta A. A study of effect of consanguinity on cochlear morphology in patients with congenital bilateral profound sensorineural hearing loss. Indian J Otolaryngol Head Neck Surg 2017;69:483-7.
8. Yoshinaga-Itano C, Sedey AL, Coulter DK, Mehl AL. Language of early- and later-identified children with hearing loss. Pediatrics 1998;102:1161-71.
9. Kennedy CR, McCann DC, Campbell MJ, Law CM, Mullee M, Petrou S, *et al*. Language ability after early detection of permanent childhood hearing impairment. N Engl J Med 2006;354:2131-41.
10. Korver AM, Konings S, Dekker FW, Beers M, Wever CC, Frijns JH, *et al*. Newborn hearing screening vs later hearing screening and developmental outcomes in children with permanent childhood hearing impairment. JAMA 2010;304:1701-8.