Original Article

Congenital sensorineural hearing loss in consanguineous marriages – Does the cochlear length vary

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Abstract

Objective: There is an increased prevalence of congenital sensorineural hearing loss (SNHL) among children born out of consanguineous wedlocks, and congenital deafness is associated with increased prevalence of structural inner-ear malformations. This study is done to evaluate whether consanguinity affects the cochlear length, which in turn will influence the type of cochlear implant and depth of electrode insertion during surgery in these patients.

Methods: Children presenting with congenital SNHL and born out of consanguineous marriages (Group A) were compared with children presenting with SNHL and born out of nonconsanguineous marriages (Group B). Patients in both groups were evaluated with magnetic resonance imaging as a routine pretreatment workup. A high-resolution three-dimensional T2-weighted sampling perfection with application-optimized contrasts using different flip-angle evolution imaging of the inner ears was performed. Curved multiplanar reconstruction module was used to deconvolute the membranous cochlea and measure its length. The cochlear lengths among both the groups were compared using analysis of variance test.

Results: A total of seven patients were included in both Groups A and B each. The mean length of membranous cochlea in Group A was 22.6 mm and Group B was 22.5 mm. There was no statistically significant variation in the cochlear lengths of both the groups.

Conclusion: Consanguinity is unlikely to produce any significant variation in the length of the cochlea.

Keywords: Cochlea, congenital sensorineural hearing loss, consanguinity

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INTRODUCTION

Imaging plays a vital role in the evaluation of children with congenital sensorineural hearing loss (SNHL). High-resolution computed tomography (HRCT) helps in assessing the bony cochlea comprehensively.[1] Magnetic resonance imaging (MRI) is done to evaluate cochlear nerve and cerebellopontine angle and also to detect any abnormalities of the membranous cochlea which may be elusive on HRCT.[2]

Cross-sectional imaging has become indispensable with the advent of cochlear implantation, as it guides the selection

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of candidates for cochlear implantation, type of cochlear implant, and depth of electrode insertion.[3]

There is a near consensus among the ear surgeons that children born out of consanguineous marriages have higher incidence of congenital SNHL.[4-6] There is also higher incidence of underlying inner-ear abnormalities among such children.[7-10] With the increasing utility of cochlear implants among children with profound SNHL, it becomes imperative to study whether consanguinity will affect the length of the cochlea, which in turn will influence the type of implant and depth of insertion of the electrode.

METHODS

A prospective study was done in a tertiary care hospital including children presenting with congenital SNHL. Prior approval of the institutional ethics review committee was taken.

All the patients who presented with congenital SNHL during the study period and were considered eligible candidates for cochlear implants based on existing norms as described by Sampaio *et al*. were included in the study.[11] Of these, children born out of consanguineous marriages were placed in Group A and those born out of nonconsanguineous marriages were placed in Group B. As a routine workup for cochlear implant, all the patients underwent a HRCT temporal bone and MRI for inner ear on a 1.5T scanner (Siemens, Magnetom Avanto, Erlangen, Germany) after obtaining informed consent of the parents/guardians. The MRI protocol used is as mentioned in Table 1. Volume dataset acquired with heavily T2-weighted, high-resolution three-dimensional sampling perfection with application-optimized contrasts using different flip-angle evolution sequence was loaded for postprocessing in a multiplanar reconstruction (MPR) module on workstation. The membranous cochlea was selectively traced out manually in serial MPR images from apex to vestibule. The resultant image in the coronal plane was modified into a 5-mm thick maximum intensity projection (MIP).

Table 1: Magnetic resonance imaging protocol for sensorineural hearing loss

MPRAGE sequence	TE (ms)	TR (ms)	Slice thickness (mm)
FLAIR axial	121	8400	5.0
T2W TSE coronal	113	3890	3.0
3D T1 MPRAGE	4.4	1160	0.9
3D SPACE	264	1100	1

MPRAGE – Magnetization prepared rapid action gradient echo; SPACE – Sampling perfection with application-optimized contrasts using different flip-angle evolution; T2W – T2 weighted; TSE – Turbo spin echo; 3D – Three dimensional; TR – Repitition time; TE – Echo time; FLAIR – Fluid attenuated inversion recovery

The cochlea in this MIP image was then uncoiled using a curved freehand function, and a virtually uncoiled image of membranous cochlea was obtained as shown in Figure 1. Using a digitized ruler, on the workstation, cochlear lengths were measured from apex of the cochlea to its junction with vestibule, which was identified as abrupt transition in diameter.

A comparative analysis of cochlear lengths so measured in both the groups was done using analysis of variance (ANOVA) test.

RESULTS

A total of 17 children reported with congenital SNHL during the study period. All these patients were considered eligible for cochlear implant. Among these, seven children were born out of consanguineous marriage and were placed in Group A and seven were a result of nonconsanguinity and hence were placed in Group B. The rest of three patients had cochlear morphological abnormalities and hence were excluded from the study. There were three male and four female children in each group. The mean age of children in Group A was 4.3 years with an age range from 2 to 9 years. In Group B, the mean age was 4.7 years with age range from 3 to 8 years.

The mean length of membranous cochlea in Group A was 22.6 mm and Group B was 22.5 mm. The shortest membranous cochlea measured 19.7 mm in Group A and 19 mm in Group B. The longest cochlea measured 25.6 mm in Group A and 25.4 mm in Group B. The mean length of right-sided cochlea among children in Group A was 22.5 mm and left-sided cochlea was 22.6 mm. The mean length of right-sided cochlea among children in Group B was 22 mm and left-sided cochlea was 23 mm.

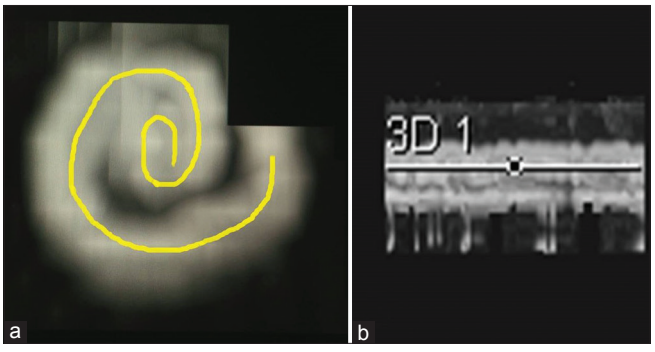

Figure 1: Curved multiplanar reconstruction of membranous cochlea. (a) Thin maximum intensity projection image in coronal plane depicting the cochlea with yellow line tracing the membranous cochlea from apex to vestibule. (b) Uncoiled cochlea with white line showing linear measurement of cochlear length

There was no statistically significant variation in the cochlear lengths of both the groups as suggested by *P* value of 0.90 obtained by ANOVA test.

DISCUSSION

Several studies have found that profound SNHL is commonly seen in children born out of consanguineous marriages.[4,12-14] Studies have focused on the genetic link between consanguinity and SNHL highlighting the presence of mutation of Gap Junction beta 2 gene (GJB2), which encodes for protein connexin 26, among many other genetic abnormalities, in children born out consanguineous wedlocks. It has also been established that the mutated gene GJB2 is associated with molecular level changes in the basement membrane of hair cells and thus causing SNHL.[15-18] There is very limited literature on macroscopic morphological changes in cochlea in such patients. Alsmadi *et al*., Tekin *et al*., and Samsi *et al*. observed that population with increased prevalence of consanguineous marriages in different parts of the world were more prone to produce children with autosomal recessive medical conditions.[6-8] Inner-ear abnormalities were found to be more common among such children. Such abnormalities included labyrinthine aplasia, cochlear aplasia and hypoplasias, common cavity, and deficient cochlear nerve.

Moreover, Agha *et al*., in their study, found 46.7% of patients with congenital SNHL with smaller cochlea.[16] Nair *et al*. also found high prevalence of cochlear malformations in children with congenital profound SNHL, which included hypoplasia and less number of cochlear turns.[10]

With increasing utility of cochlear implants across spectrum of cases, and with the availability of numerous types of such implants, it is imperative to analyze and decide which implant is better suited for a particular child. In this context, cochlear measurement is of great utility.[17]

As consanguinity is associated with increased incidence of profound congenital SNHL and congenital SNHL can be associated with altered morphology of cochlea including alterations in size and number of turns, it was found apt to study whether consanguinity has an independent effect on the length of otherwise normal appearing cochlea, which in turn will influence the type of cochlear implant/depth of electrode insertion in such cases. No other similar study could be found in this context. The human inner ear is completely developed and attains final size and morphology in utero before birth.[18] Hence, the need for age-matched controls is alleviated in this study. In the present study, no significant variation was observed in the cochlear lengths between children in Group A and Group B. The mean length of cochlea among Group A children was 22.6 mm and Group B children was 22.5 mm. This is similar to values obtained in the study by Pochini Sobrinho *et al*., wherein the authors measured the length of 6 cadaveric cochlea by MRI, by summing up serially measured multiple short segments of cochlea (up to 14 segments) in coronal images, and found a cochlear length range of 17–26.5 mm.[19] This method is cumbersome due to the spiral shape of cochlea and its three-dimensional orientation. In the present study, 28 cochleae were measured and the spiral shape of cochlea was virtually uncoiled using curved MPR algorithm and length measurements were done in the resultant two-dimensional image.

Connor *et al*. measured the "cochlear distance" on both CT and MRI, which is the distance between the round window to the opposite wall of the cochlea and applied adjusted estimates to the spiral function to estimate the length of electrode for a 360° insertion.[17] The limitation of that study was that the measurements could be used only for 360° insertions. Moreover, cochlear dimensions are not uniform in all individuals.[20] This study has shown that no significant change in the cochlear lengths exists between the two subgroups of patients born out of consanguineous and nonconsanguineous marriages.

The prevalence of congenital SNHL is 1–2/1000 live births,[21-24] of which consanguinity/genetic factors contribute to 50% of cases.[24-27] Taking a birth rate of 19/1000 population (India), the incidence of congenital SNHL due to consanguinity is 0.002%.[28] In an Indian setting, actual numbers presenting to the tertiary care facilities are even lower.

This study focuses on a very unique finding with scant literature and high applicability which has potential to influence current management practices.

Moreover, incidence/prevalence has not been calculated in this study in which context sample size would have been an issue. This study compared cochlear lengths objectively. Since we are comparing means of the two groups only based on a single factor of consanguinity and we make no assumptions on equality of variances of the two groups, we employ one-factor ANOVA (F-test). Since its a comparison between only two groups, *t*-test and F-test yield the same *P* value, and hence, there is no preference of one over the other. Hence, to improve accuracy and applicability, ANOVA test was applied to find statistical significance.

CONCLUSION

Although consanguinity is associated with increased prevalence of congenital SNHL, it is unlikely to produce any significant variation in the length of otherwise normal appearing cochlea.

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Conflicts of interest
There are no conflicts of interest.

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