Cochlear anomaly with preserved hearing - A Clinical Paradox: Our Experience with Two cases

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May 6, 2020

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Introduction

Bilateral profound hearing loss, whether congenital or acquired, is one of the most disabling anomalies a child can have. As the child grows up, there is associated delay in development of speech leading to social and functional stigmata for the child as well as the caregivers. Here comes the importance of early diagnosis and intervention. Out of the well-studied causes, Inner Ear Malformations (IEMs) comprise 20% of causes of congenital hearing loss (1,2). IEMs have been classified exhaustively by various authors depending on the site of involvement, each category displaying a variable degree and type of hearing loss. A normal cochlea has 2 $\frac{1}{2}$ or 2 $\frac{3}{4}$ turns with modiolus as the central axis and interscalar septum as a partition between cochlear inner wall and modiolus (2,3). The cochlea is said to be hypoplastic when there are less than 2.5 turns. There are four types of cochlear hypoplasia (I to IV) with hearing status ranging from normal to profound loss; it being very rare to have normal hearing (2). When the cochlea has 1.5 turns then it is termed as Mondini dysplasia. Before proceeding to any intervention in case of IEMs, it is imperative to look for the presence of cochlear nerve and various audiologic parameters (4). Here we are reporting two cases, one with cochlear hypoplasia type IV and one patient with Mondini dysplasia. The point of interest in these cases was a preserved audiologic status inspite of severe malformations of the inner ear. The clinico-radiological discrepancies piqued our interest and thus, we would like to share these cases owing to their rarity.

Case 1

A 3 years old male child was brought to our Outpatient Department by his parents with the complaints of less responsiveness to sounds and delayed speech development. He was born to healthy, normal hearing parents by normal vaginal delivery with a birth weight of with no perinatal risk factors. At the time of presentation, he could speak only limited monosyllables. There was no history of ear discharge, recurrent meningitis or dizziness. A delay in milestones was reported. On examination, the child was active and playful with lack of eye contact. He had microcephaly. The rest of the clinical examination was non-contributory. Patient was subjected to a complete audiological evaluation. Oto Acoustic Emissions (OAEs) test showed pass in both ears. Tympanometry showed bilateral 'A' type tympanogram with normal reflex. Brainstem Evoked Response Audiometry (BERA) showed a normal wave V peak till 30 dBnHL in both ears suggestive of bilateral normal hearing sensitivity (Figure.1). In view of normal hearing as per audiological investigations, a detailed psychological evaluation was sought. On Development Screening Test, he obtained a Developmental Age of 18 months (Development Quotient of 50), indicative of mild developmental delay. On ISAA (Indian Scale for Identification of Autism), he scored 84, indicative of mild Autism. Main problem areas were identified

as social relationships, speech-language and communication. As part of further work up, Magnetic Resonance Imaging (MRI) of brain, inner ear and internal auditory canal (IAC) was done. Multiplanar images were acquired using T1, T2, T2- FLAIR, FIESTA sequences by using GE discovery 750W 3T MRI. Surprisingly, MRI revealed markedly hypoplastic middle and apical turns of bilateral cochlea with normal basal turns with non-visualisation of modiolus and interscalar septa in addition to bilaterally dilated IACs (Figure 2 A-D). High Resolution Computed Tomography (HRCT) temporal bone was done for correlation. In view of bilateral cochlear hypoplasia type IV, audiological investigations were repeated from a different reliable audiology clinic with congruent results.

Case 2

A 15 years old male who had apparently normal hearing till the age of 14 years with normal speech and language development presented to us with bilateral fluctuating progressive hearing loss in both ears (left>right). There was no history suggestive of any vestibular symptoms apart from intermittent tinnitus. On clinical examination bilateral tympanic membrane was intact. Otoacoustic Emissions test showed refer in both ears suggestive of poor functioning of outer hair cells. Pure tone audiometry showed moderately severe sensorineural hearing loss in right ear and moderately severe mixed hearing loss in left ear (Figure. 3). Multiple pure tone audiograms were performed revealing progressive hearing loss in left ear. HRCT Temporal bone and MRI brain and IAC showed an abnormal cochlea with one and a half turns suggestive of Mondini malformation on the left side (Figure. 4 A-C).

DISCUSSION

Synopsis of key/new findings

- Inner ear malformations may range from mild to severe with variable degrees of hearing loss. Severely malformed cochlea with a normal hearing is a rarity.
- Our first case was a three years old boy whose hearing was normal as per various audiological tests and he was diagnosed to have autism. Hence, unresponsiveness to sounds and poor speech development could have been attributed to autism. However, further evaluation was carried out and radiology revealed cochlear hypoplasia type IV. Thus, radiology as a diagnostic tool must not be overlooked.
- The second patient had an apparently normal hearing till adolescence which is hard to explain in the presence of Mondini dysplasia. A recent onset, progressive fluctuating bilateral hearing loss led to the patient seeking medical attention and the radiologic diagnosis of left sided Mondini dysplasia.
- The number of turns and morphology of the cochlea may not be the only determinants of the final hearing status of individuals with cochlear malformations.
- Radiology and psychological evaluation may be invaluable adjuncts aiding in the final diagnosis and should be resorted to in such cases as described above.

Current literature

Developmental anomalies of inner ear depend on timing of insult exposure during embryonic development. Degree of hearing loss will depend on the severity of the malformation. A number of classifications have been proposed by various authors over the years (5,6,7).

Our first case is a 3 years old autistic child with bilateral cochlear hypoplasia type IV with normal audiological parameters. Minimal anomalies can be associated with normal hearing or may even be missed on routine radiological imaging. In our case, the apical and middle turns were severely hypoplastic which doesn't correlate with the audiology. As per current evidence, the number of turns of the cochlea plays a limited role in causing hearing loss (3). Polvogt et al performed an interesting study to correlate structural changes of cochlea and hearing. Temporal bone dissection of seventeen subjects who had normal hearing in their life was performed and a variety of cochlear anomalies were found. The authors stated that several other features such as length of basal turn, length and height of upper turn need to be measured in cases of cochlear hypoplasia. The arrest of cochlear development prematurely will cause a decrease in height from base to apex of cochlea, which can lead to hearing loss (8). The mechanism of decrease in hearing is by reduction

of displacement of basement membrane vibration (8,9). It is claimed that this decrease in displacement is maximum when the cochlea is the shortest. In our case, the cochlea was severely malformed which should have caused measurable hearing loss. On the contrary, we observed all audiological parameters to be normal. Additionally, the cochlear nerve and auditory tract were normal. In Autistic Spectrum Disorder, the auditory abnormalities can be either peripheral or central. The most common auditory deficit is the inability to filter auditory input due to abnormalities in auditory brainstem. However, a normal waveform pattern on BERA merits some explanation. Mohammed AS et al in 2017 reported a case of a two- year old with incidentally detected cochlear hypoplasia on one side and cochlear nerve aplasia on the other (10). The authors in their description of cochlear hypoplasia mention that hearing in such ears is variable and maybe remarkably good. The variability maybe attributed to the degree of membranous labyrinthine development within the truncated cochlear lumen.

Mondini dysplasia presents with specific features such as flat cochlea, immature semicircular canals and vestibule and severe to profound hearing loss (5,7). Limited evidence exists regarding cases with Mondini dysplasia and a normal hearing. (7). In our scenario, the point of interest is presence of normal hearing till adulthood followed by a progressive, fluctuating hearing loss.

It may thus be re-emphasized that apart from number of turns of cochlea and presence/absence of cochlear nerve there are certain parameters which are yet to be studied in detail which influence the hearing status of malformed ears.

Clinical Applicability

Inner ear malformations have been studied and classified in great detail by various authors in the past. The consensus is that the timing and severity of insult determine the morphology of the malformation and the hearing. However, cases with severe IEMs as in our patients with normal hearing are rare to be found in literature pointing to the fact that certain other factors may play a more important role than the dimensions of the cochlea alone on the ultimate hearing status of the patient. Also, one must keep in mind that patients with a radiologically severely abnormal cochlea may have serviceable or even a remarkably good hearing.

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FIGURE LEGENDS