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Case Report

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A Case Report on Familial origin of Auditory Neuropathy Spectrum Disorder

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ABSTRACT

Auditory neuropathy Spectrum Disorder (ANSD) is a unique hearing disorder where outer hair cell status is found to be normal, but inner hair cell and/or the synaptic connections to the auditory nerve is disrupted. It is a diverse group of hearing disorder which can be manifested either congenitally or can be late in onset. However, there are various etiologies of auditory neuropathy which is represented in the literature, which includes birth related risk factors for hearing loss like prematurity, bilirubin synthesis issues, lack of oxygen supply to the baby during or after delivery, and genetic actors. It is estimated that approximately 40% of cases have an underlying causal factor as genetic origin, which can be inherited in either syndromic or non-syndromic way. The below case report provides an extra support for the fundamental genetic trait in ANSD. The study presents two congenital ANSD cases where, both children were diagnosed as congenital ANSD.

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Introduction

Auditory neuropathy spectrum disorder (ANSD) is a retro cochlear disorder in which patient shows normal outer hair cells functioning (described by normal otoacoustic emissions /cochlear microphonics) and deviant or absent peaks in auditory brainstem response (ABR) evaluation. The site of lesion in ANSD is often unidentified still, but a possible site reported in literature includes sensory cells of cochlea especially inner hair cells, cochlear neural ganglia, and the auditory nerve [1]. Multiple causal risk factors like Prenatal/neonatal infections, Hyperbilirubinaemia, Anoxia/Hypoxia, certain Immune disorders, syndromal disorders and Genetic have been revealed in the literature of ANSD [2]. Researches revealed that prevalence of ANSD vary across the subjects being studied. The prevalence data in ANSD is reported to be 40 % in neonates who are at high risk for hearing loss, predominantly babies admitted in intensive care unit [3]. Berlin et al. projected the incidence of ANSD in children with severe hearing impairment to be 4% [4]. Colm et al. study reported the prevalence of ANSD in patients with SNHL range from 0.5% to 15% [5]. Similarly Kirkim et al. determined in infants with hearing impairment at 15.4% [6]. Sininger estimated ANSD occurrence at 10%, while Rance et al. described a prevalence rate of 11% in a child population with hearing loss, and 0.23% within the at-risk population [7, 8]. In the earlier years, a number of genes associated with ANSD were documented, these studies were noted that the major pathological mechanism of ANSD lies at the molecular level (10). The OTOF gene was the first gene identified for autosomal recessive non-syndromic ANSD (6). It is located on chromosome 2p23.1, considered as an key role in afferent synaptogenesis of the auditory ribbon synapse. In this case report we have made an effort to highlight the detailed audiological findings and casual factor for ANSD with familial origin of 2 cases.

Method

Case Presentation

Case 1

Child aged 3.6 years, female with a history of 2nd degree consanguineous marriage came to the department of audiology and speech language pathology with the main complaint of reduced hearing sensitivity and limited speech output. Detailed case history was noted which revealed presence of congenital hearing loss running in the family where, her elder sister and cousin brother also had similar complaints and was diagnosed as having hearing impairment. There was no other significant birth related risk factor for hearing loss. Child's developmental milestones showed normal motor development and delayed speech language milestones.

Case 2

Child, Aged 22 month male child born for a non consanguineous marriage came to the department of audiology and speech language pathology with the main complaint of reduced hearing sensitivity and limited speech output. Detailed case history was noted which revealed the child was related to case 1 described above (cousin sister). There was no other significant birth related risk factor for hearing loss. Child's developmental milestones showed normal motor development and delayed speech language milestones.

Audiological Evaluation

Before initiating the routine audiological evaluation, otoscopic evaluation was performed to rule out any presence of external or middle ear pathology and the ear canal was noted to be clear with the tympanic membrane visible in both cases.

Detailed audiological assessment started with Immitance evaluation to see for the presence of any middle ear related issues.

In case 1 result obtained were, right ear 'As' type and left ear 'Cs' tympanogram, indicating a (?) middle ear pathology. In case 2 results obtained were bilateral 'As' type tympanogram suggestive of normal middle ear functioning. Bilateral absence of acoustic reflex in both cases indicated an abnormal auditory neural pathway.

Transient Evoked Oto Acoustic Emission (TEOAE) examination was carried out to get the information about the performance of the outer hair cells present in the inner ear. In both the cases TEOAEs were present bilaterally indicating a normal outer hair cell functioning.

Auditory Brainstem response (ABR) testing was carried out to assess the integrity of neural pathway up to brainstem level and also to know the hearing sensitivity objectively. The ABR results of both the children showed absence of peak V at 90dBnHL indicating retro cochlear pathology. Presence of Cochlear Microphonics (CM) was checked to confirm the intactness of the inner ear. Cochlear microphonics is obtained by repeating the testing at the same intensity at two different polarities, Rarefaction and condensation respectively. As the polarity changes the direction of movement of the basilar membrane in the inner ear varies resulting in an opposite and ringing waveforms at the initial time window of the ABR waveform. Ringing CM was present up to 2 ms in both the case which further indicated a normal cochlear functioning. Absence of ringing CM when the polarity changed to alternate and also when the insert tube was blocked confirmed the presence of CM and ruled out the chance of it being a stimulus artifact. Figure 1 and 2 below are the ABR waveforms of the 2 cases where ringing cochlear mechanics can be observed in the time window from 0-2 msec and absence of V in later time window suggesting a neural pathology.

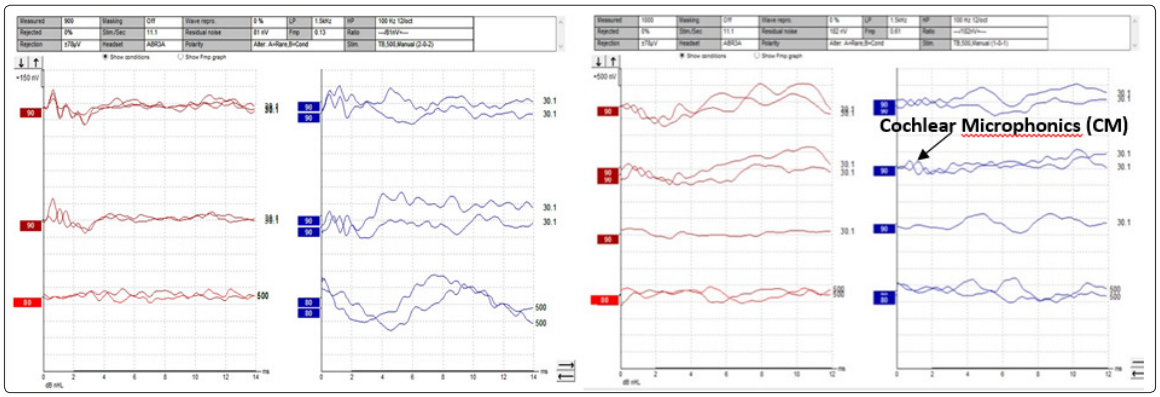

Figure 1 and 2: Auditory brainstem response of Case 1 and Case 2 respectively

Discussion

Manchaiah, Zhao, Danesh, & Duprey study reported non syndromic hereditary ANSD variety in Chinese families and their pedigree analysis suggested transmission is mainly through X-linked or autosomal recessive traits [9]. Similarly studies have discovered that mutation with some of the genes like OTOF and/or loci to be the cause for auditory neuropathy spectrum disorders (ANSDs) [5, 10]. The above studies are in compliance with the findings of the current study which reveals a familial transmission of ANSD.

Conclusion

Familial transmitted ANSD cases need to be focused more and hence it will be help to study their genetic transmission in detail with necessary genetic testing. It is also very important to account such cases details so that the genetic factor ensuing in ANSD is profoundly understood and researched [11, 12].

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