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Echoes of Uncertainty: Investigating Adult Auditory Neuropathy Spectrum Disorder- A Diagnostic Odyssey

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Abstract

Auditory Neuropathy Spectrum Disorder (ANSD) presents a formidable diagnostic challenge in audiology due to its intricate interplay between peripheral and central auditory processing. ANSD is a rare audiological condition characterized by a discrepancy between otoacoustic emissions (OAEs) and auditory brainstem responses (ABRs). While OAEs are often preserved, ABRs may show minimal or absent responses, signifying impaired neural transmission despite intact cochlear function. This disorder spans a broad spectrum of presentations, contributing to the diagnostic complexity.

Keywords: Auditory; ANSD; Neuropathy; Hearing

Abbreviations

ANSD: Auditory Neuropathy Spectrum Disorder; OAEs: Otoacoustic Emissions; ABRs: Auditory Brainstem Responses; HRCT: High Resolution Computed Tomography; CE-MRI: Contrast Enhanced Magnetic Resonance Imaging.

Introduction

The terms "Auditory Dyssynchrony" and "Auditory Neuropathy" were coined in the late 1990s to describe a unique hearing disorder where there is a mismatch between the function of the cochlea's sensory cells and the auditory nerve. The term "Auditory Neuropathy" was introduced by Dr. Douglas Starr and colleagues in 1996 to describe patients with abnormal auditory brainstem responses (ABRs) and intact otoacoustic emissions (OAEs), suggesting a problem in the auditory nerve. "Auditory Dyssynchrony" emerged around the same time to indicate disrupted neural firing timing along the auditory pathway. The terms have evolved, with a preference for "Auditory Neuropathy Spectrum Disorder" (ANSD) to encompass the varying degrees and causes of the condition. These terms have been vital for classifying and understanding this distinct hearing impairment and have led to further research and tailored management approaches [1,2]. Our case report delves into the diagnostic journey of a twenty-six years old male exhibiting atypical audiological responses and speech perception difficulties, ultimately highlighting the complexity of ANSD assessment, etiology, and management.

Case Report

A 26-year-old male presented with complaints of bilateral HL for eight years which was gradual onset, progressive and non-fluctuating. Previously he was diagnosed as bilateral sensorineural (SNHL)-noise induced, but the individual exhibited no improvement with hearing aid usage which was prescribed to him at other centre. The patient's HL

had a profound impact on his daily communication abilities and was concomitant with notable deficiencies in speech discrimination. Noteworthy is the patient's history of recurring exposure to high-intensity noise, particularly during small arms firing exercises. Detailed evaluation ruled out any additional ENT complaints. On audiological evaluation, bilateral tympanic membrane was intact. Tuning fork test was positive (256Hz) bilateral. Weber's was central and absolute bone conduction was reduced bilateral. Pure tone audiometry revealed severe SNHL on right and moderate SNHL on left (Figure 1). Tympanometry was 'A' type and Acoustic reflex was absent bilateral. Caloric test revealed hypoactive labyrinth on right and normal response on left. Screening and diagnostic OAE was 'pass' bilateral. ABR showed no identifiable peak up to 100dBHL on right, while identifiable wave was obtained on left side suggesting of threshold sensitivity with 85dBHL (Figure 2). ASSR (auditory steady state response) revealed no response at highest threshold in right ear and single response at 500Hz at 55dB (Figure 3).

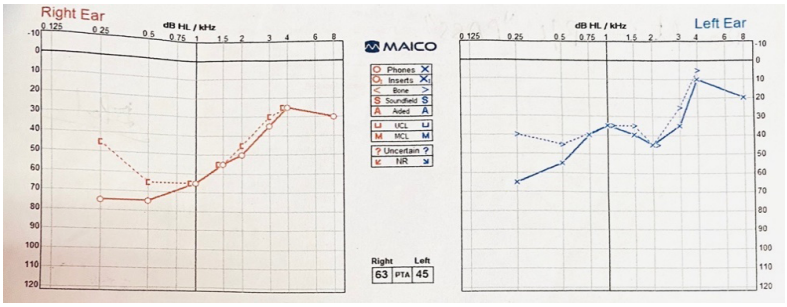

Figure 1: Right ear: AC: 61.25; BC: 57.5. Left Ear: AC: 43.75; BC: 40 dBHL.

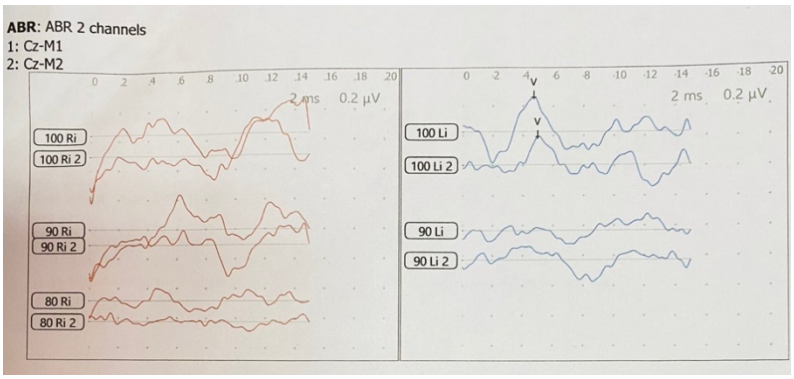

Figure 2: Right Ear: No identifiable peak V obtained till 100dBHL suggestive of threshold sensitivity beyond 90dBHL. Left Ear: identifiable peak V obtained till 100dBHL, suggestive of threshold sensitivity within 85dBHL.

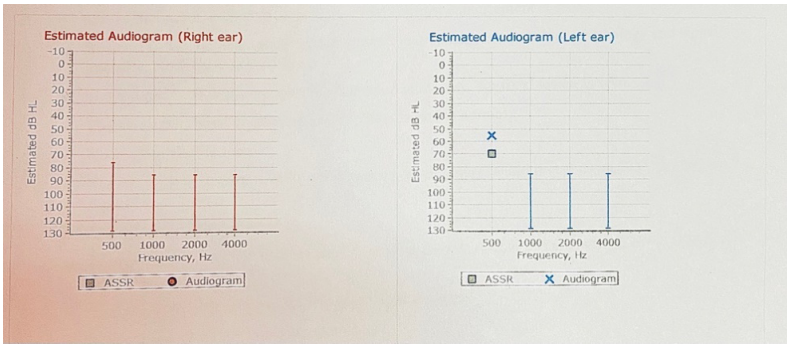

Figure 3: ASSR: no response at highest threshold in right ear and single response at 500Hz at 55dB.

CE-MRI (Contrast enhanced magnetic resonance imaging) Brain and high resolution computed tomography (HRCT) Temporal bone showed normal study. The audiometric evaluation was repeated and similar results were obtained. Based on above results, patient was diagnosed as case of ANSD. Patient is presently under follow -up and planned for cochlear implantation in right ear.

Discussion

The prevalence of ANSD ranges between 0.5 to 15% among all cases of HL [3]. The Guide- lines Development Conference (2008) defined an abnormal ABR as 1) a "flat" ABR with no evidence of any peaks or 2) presence of early peaks (waves up to III) with absence of later waves or 3) some poorly synchronized but evident later peaks (wave V) that appear only to stimuli at elevated stimulus levels [2]. The diagnostic process for ANSD demands meticulous scrutiny of audiological findings, patient history, and potential etiologies [1]. In the current case, the patient's reported speech comprehension difficulties were corroborated by audiological evaluations revealing preserved OAEs but irregular ABR waveforms, also supported with absent acoustic reflex. Notably, ABR provides objective measurement of neural activation and neural synchrony [2]. This aligns with the classical ANSD pattern, emphasizing the importance of comprehensive assessment of both cochlear and neural pathways.

The foundation of ANSD diagnosis lies in ABR assessment, yet deviations from the norm can confound the diagnostic process, for example patients with poor neural synchrony. ANSD can manifest as absent, delayed, or atypical ABR waves, complicating differentiation from auditory processing disorders or retrocochlear pathologies [2,3]. These subtleties require nuanced scrutiny, necessitating audiologists' acumen to distinguish genuine ANSD from imitating conditions.

The etiology of ANSD is multifaceted. Genetic causes can be syndromic (example- Charcoat Marie Tooth or Friedreich's ataxia) or non-syndromic. It plays a pivotal role, with mutations in DFNB9 (otoferlin), DFNB59 (pejvakin), OTOF, DIAPH3, CACNA1D and other genes identified in ANSD cases [2]. Additionally, acquired factors such as neonatal jaundice can contribute to ANSD development, as evidenced by the patient's history in this case. Hyperbilirubinemia-induced neural damage disrupts auditory nerve function and may lead to ANSD [4].

The spectrum of clinical manifestations in ANSD engenders diagnostic intricacies. Ranging from subtle speech perception challenges to profound hearing impairment, this variability underscores the need for judicious diagnostic precision. While objective tests such as OAE and ABR provide vital data, deciphering the patterns and translating them into diagnostic clarity remains an intricate endeavour, especially in adult cases. The primary mechanism underlying these challenges appears to be impairments in the coding of temporal features of acoustic stimuli. It is these deficiencies in temporal encoding that likely contribute to the often restricted advantages observed with acoustic hearing aids [3,5].

Navigating the complex landscape of ANSD requires collaboration across various disciplines. Audiologists, otolaryngologists, speech therapists, neurologists, and geneticists work together to encompass the multidimensional facets of the disorder. Genetic profiling and advanced imaging techniques enhance the diagnostic armamentarium, unraveling the cryptic underpinnings of ANSD [6]. Management of ANSD requires a personalized approach. Cochlear implantation has demonstrated success in improving speech perception and auditory outcomes in ANSD patients.4 However, candidacy evaluation requires meticulous consideration of residual hearing, auditory potential, and communication needs. Auditory training programs can also aid in optimizing speech perception and auditory skill development [3]. The intricate interplay between peripheral and central auditory processing in ANSD underscores the necessity of interdisciplinary collaboration. On-going research in auditory neuroscience promises to illuminate the underlying mechanisms and guide tailored interventions for ANSD patients.

Conclusion

This case report exemplifies the intricacies of ANSD diagnosis and management, underscoring the value of multidimensional assessment in audiology. The diagnostic journey in this case highlights the importance of a holistic perspective, encompassing audiological assessments, medical history, and evidence-based interventions. Cochlear implantation and auditory training hold promise in addressing the communication challenges posed by ANSD. By embracing a comprehensive approach that bridges audiology, genetics, and neuroscience, we can navigate the intricate landscape of ANSD, enriching our understanding and enhancing patient outcomes.

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