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Clinical Management of ANSD in Infants and Young Children

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The term ‘auditory neuropathy spectrum disorder’ (ANSD) encompasses a group of patients with evidence of normal outer hair cell function as reflected by present otoacoustic emissions (OAE) and/or present cochlear microphonics (CM) but absent or abnormal auditory brainstem responses (ABR). Santarelli describes auditory neuropathy as “a hearing disorder characterized by disruption of temporal coding of acoustic signals in auditory nerve fibers resulting in impairment of auditory perception that rely on temporal cues.”[1] Functionally, the disorder may result in hearing thresholds that range from normal in some individuals, to profound in others.[2] Commonly cited characteristics include speech perception abilities that are poorer than expected for the degree of hearing loss and difficulty hearing in noise.[3]

Since the first reported cases of auditory neuropathy in the late 1980s and early 1990s, audiologists and other professionals working with these patients have struggled with several complex issues surrounding diagnosis and management. Early reports of auditory neuropathy by Starr and colleagues in 1996 described several patients with perplexing findings that included absent or abnormal ABR and present cochlear microphonics and otoacoustic emissions, implying normal outer hair cell function and altered neural transmission.[4] The majority of these patients had other peripheral neuropathies, and several were subsequently found to have Charcot-Marie-Tooth

disease and other progressive neurologic conditions.[4] In many cases, the hearing complaints preceded neurologic symptoms by several years.

Prevalence

Over time, as clinical protocols were developed to diagnose ANSD, more children have been identified with this condition. It is now known that ANSD is a relatively common hearing disorder affecting up to 10% of children with permanent hearing loss.[2] And while the actual prevalence is unknown, it is clear that those who work in settings with a high volume of pediatric patients that employ evidence-based protocols will encounter a substantial number of children with this condition. The prevalence of ANSD in healthy, full-term infants is low, on the order of 1:10,000.[5] There is a much higher prevalence of ANSD in premature infants and those who have required hospitalization in the neonatal intensive care nursery where histories may have included anoxia, hyperbilirubinemia, low birth weight, or exposure to ototoxic medications.6 In those infants, the prevalence may be as high as 24%.[7]

It has now been nearly 30 years since the earliest case reports and although there is still much to learn, we now have a better understanding of the heterogeneity of the disorder as well as better evidence to guide recommendations for children and families. In particular, discoveries in the areas of genetics, radiologic imaging, and environmental factors have contributed to the role that etiology and site of lesion play in determining outcomes.

Etiology

The etiology of ANSD includes structural abnormalities affecting the auditory system such as an absent or deficient VIII nerve, genetic abnormalities and conditions associated with prematurity, and/or hospitalization in the newborn critical care nursery. Rance and Starr (2015) provide an excellent review of the clinical and pathophysiological features of auditory neuropathy that distinguish site(s) of dysfunction. They emphasize that the diagnosis of auditory neuropathy relies on (1) objective neurophysiological measures of cochlear hair cell and auditory nerve functions; (2) imaging of auditory nerve/brainstem; and (3) behavioural audiological measures. They also describe the diagnostic criteria for presynaptic disorders affecting

inner hair cells and ribbon synapses; postsynaptic disorders affecting unmyelinated auditory nerve dendrites; postsynaptic disorders affecting auditory ganglion cells and their myelinated axons and dendrites; and central neural pathway disorders affecting the auditory brainstem.[8]

While it is clear that we now have a variety of radiologic, genetic, and audiologic tools to determine the site of the lesion, complex diagnostic measures are not universally available and, as pointed out by,1 etiologic factors can be identified in only about half of the patients. However, when etiology or site of the lesion can be identified, it may inform clinical decision-making. For example, when genetic evaluation identifies an Otoferlin (OTOF) mutation, a pre-synaptic disorder which disrupts the function of the ribbon synapses, this may predict positive outcomes from cochlear implantation9; whereas radiologic imaging showing an absent or deficient VIIIth nerve may signal a less optimistic prognosis for cochlear implantation and a recommendation for visual communication, with or without a cochlear implant.[10,11]

Diagnosis

Well defined, evidence-based clinical protocols are needed to diagnose ANSD. When ABRs are absent or grossly abnormal, testing using single polarity clicks (rarefaction and condensation) at a high-intensity level must be used to determine if auditory neuropathy is present. In addition to ABR, otoacoustic emissions and acoustic immittance measures including the acoustic reflex should be included in the diagnostic test battery. If clinics are using ASSR for diagnostic testing, a click ABR at a high-intensity level with single polarity clicks should be performed in addition to ASSR.

Once an accurate diagnosis is made, families need to be counseled regarding the implications of the diagnosis and the next steps. Inevitably, counseling regarding prognosis at the time of diagnosis will need to be guarded. Apart from the electrophysiologic findings supporting the ANSD diagnosis, little information is available to make predictions regarding expected outcomes until additional clinical data can be obtained. Depending on the medical history of the child and family, this may include imaging of the inner ears (e.g., MRI, CT scans) and referrals for genetics, neurology and ophthalmology.