Children With Auditory Neuropathy Spectrum Disorder (ANSD)

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Introduction

In 1991, Arnold Starr and colleagues described an 11-year-old child with the paradoxic findings of absent auditory brainstem responses in the presence of normal cochlear microphonics (CMs) and otoacoustic emissions (OAEs). The child demonstrated severely impaired speech understanding despite a mild loss of hearing sensitivity. Further evaluation showed impaired temporal processing as exhibited by abnormal findings on a battery of psychophysical tests that included gap detection and binaural masking level differences. In 1996, Starr and colleagues reported on a group of 10 children and adults with similar findings (Starr, Picton, Sinner, Hood, & Berlin, 1996). Although the patients presented with no apparent neurologic involvement when their hearing impairments were first identified, eight of the ten showed later evidence of other peripheral neuropathies. Starr and colleagues coined the term “auditory neuropathy” (AN) to describe patients whose hearing impairment was attributed to “neuropathy of the auditory nerve.”

Since the initial report by Starr and colleagues it has become clear that individuals diagnosed with AN are a heterogeneous group even though they may exhibit common audiologic findings. Some individuals who exhibit the AN pattern may have a congenital form of the disorder resulting from a genetic mutation or pre/perinatal causes, whereas others may have a later onset form of the disorder associated with other peripheral neuropathies. Speculation regarding the underlying mechanisms of AN includes selective inner hair cell loss, a synaptic or myelinization disorder, or an auditory nerve disorder with other peripheral neuropathies (Starr, Sinner, & Pratt, 2000). Over the past decade, as universal newborn hearing screening has expanded and as audiologists have become more familiar with AN and its diagnosis, a growing number of infants and young children have been identified with this disorder. Characteristics of AN have been reported in children with histories of low birth weight, prematurity, neonatal insult, hyperbilirubinemia, perinatal asphyxia, artificial ventilation, and various infectious processes (Dowley et al., 2009; Mason, De Michele, Stevens, Ruth, & Hashisaki, 2003; Xoinis, Weirather, Mavoori, Shaha, & Iwamoto, 2007). Genetic abnormalities have also been identified, including those associated with the genes OTOF, PMP22, MPZ, and NDRG1 (Kovach et al., 1999; Starr et al., 2003; Varga et al., 2006; Yasunaga et al., 1999). In 2006, Buchman and colleagues described a group of children who presented with physiologic test results typical of AN who were subsequently diagnosed by magnetic resonance imaging (MRI) as having cochlear nerve deficiency (CND), that is, absent or small cochlear nerves (Buchman et al., 2006).

Auditory neuropathy was initially thought to be quite rare; however, current estimates of prevalence range from 7 to 10% of children with permanent hearing loss (Madden, Rutter, Hilbert, Greinwald, & Choo, 2002; Rance, 2005). Considerable controversy exists regarding almost every aspect of the disorder including its etiology, site of lesion, treatment, and even the terminology used to describe it. In an effort to provide recommendations to clinicians working with these children a panel was convened in June, 2008, at an international conference in Como, Italy (Guidelines Development Conference on the Identification and Management of Infants with Auditory Neuropathy, 2008). The panel was charged with developing guidelines for identification and management of infants with AN. After extensive review and discussion, the term
“auditory neuropathy spectrum disorder” (ANSD) was adopted as a way of describing the heterogeneous and multifaceted nature of the disorder.\(^1\) Even so, current terminology remains inadequate since the level of dysfunction for many of the children with this condition may be central to the auditory nerve (Rapin & Gravel, 2003, 2006).

Further examination of this disorder, including information about etiology, possible mechanisms, and clinical characteristics is provided in Chapter 13 in this volume. The present chapter addresses clinical management guidelines for pediatric audiologists and other professionals who work with these children.

**Audiologic Evaluation**

The Joint Committee on Infant Hearing (JCIH; American Academy of Pediatrics, JCIH, 2007) recommends that a comprehensive audiological evaluation be performed for all infants referred from newborn screening by audiologists experienced in pediatric assessment. The recommended test battery includes physiologic measures and, when developmentally appropriate, behavioral measures. The goal of the diagnostic assessment is to determine the type of hearing loss and to estimate hearing sensitivity across a range of frequencies for each ear. The same comprehensive approach is needed to identify ANSD, and care must be taken to select and apply an appropriate electrophysiological test protocol. The initial battery should include: oto-scopic examination, immittance measures, auditory brainstem response (ABR) testing, and assessment of otoacoustic emissions (OAEs). When developmentally feasible, behavioral audiometry and speech perception measures should be included.

**Diagnostic Criteria**

In ANSD there is evidence of hair cell function while afferent neural conduction in the auditory pathway is disordered. Typical audiologic findings include absent or markedly abnormal auditory brainstem response (ABR) in combination with a present cochlear microphonic (CM) and/or otoacoustic emissions (Figure 37–1).

Some earlier definitions of auditory neuropathy included present OAEs as a requirement for diagnosis; however, it is now recognized that OAEs may be present initially and disappear over time (Deltenre et al., 1999; Rance et al., 1999; Starr et al., 2001) and many infants with the disorder have present CM with absent emissions at the time of diagnosis, even when middle ear status is normal. Furthermore, both OAEs and CM provide evidence of hair cell function, albeit by different mechanisms. Thus, more recent definitions of ANSD include either present OAEs or a present CM in combination with markedly abnormal or absent ABR.

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\(^1\) The term “auditory neuropathy spectrum disorder” was suggested by Judy Gravel who noted the varied etiologies, presentations and outcomes for children with similar electrophysiologic test findings.
Physiologic Measures

When diagnostic ABR testing using either click or tone burst stimuli shows no evidence of a neural response at high levels of stimulation, the clinician must determine if a CM is present. To accomplish this, ABR testing should be performed with insert earphones and high-intensity click stimuli (80–90 dB nHL) with separate response averages obtained for both rarefaction and condensation click polarities (Berlin et al., 1998; Starr et al., 2001). The CM occurs in the first few milliseconds of the response and, unlike neural responses, will show a reversal of the waveform when the stimulus polarity is inverted. If testing is limited to the evaluation of click stimuli with alternating polarity, the CM will not be evident because of cancellation and this may result in an incorrect diagnosis of profound sensory hearing loss in a child who actually has ANSD.

To eliminate the possibility of incorrectly identifying stimulus artifact as the CM, a “no sound/control run” should be completed by clamping or disconnecting the sound tube from the transducer without altering the spatial relationship between the transducer and the electrodes/leads (Rance et al., 1999). Insert earphones are required instead of standard headphones because inserts introduce a delay between the electrical signal at the transducer and the acoustic signal in the ear canal, further eliminating the possibility of electromagnetic stimulus artifact being incorrectly identified as the CM. Figure 37–2 shows an example of stimulus artifact incorrectly identified as CM.

As in all diagnostic ABR procedures, it is important to optimize recording conditions and to complete the testing with the infant sleeping quietly in either natural or sedated sleep. If an infant tested in natural sleep is too active for accurate, artifact-free assessment, it is better to reschedule the procedure and complete a sedated test than to risk an incorrect diagnosis due to poor recording conditions.

Both the amplitude and the latency of the CM vary in ANSD, so clinicians need to be familiar with the full range of possible test results (Starr et al., 2001). Although the characteristic pattern associated with the diagnosis of ANSD includes a “flat” ABR with no evidence of a neural response but a present CM (see Figure 37–1), it also is possible to have a CM in combination with a markedly abnormal ABR morphology, for example, an ABR with early waves absent at high intensity levels but with a present wave V that can be tracked to lower intensity levels (Figure 37–3).

With abnormal patterns like these, caution must be exercised in using the ABR to estimate behavioral thresholds since the minimal response levels obtained from the ABR may not correlate with the child’s behavioral thresholds. Once the ANSD pattern has been identified, it is no longer possible to use the ABR to estimate the infant’s behavioral thresholds, even when there is evidence of distal waveforms. At the time of this writing, controversy exists regarding whether these patients should be diagnosed as having ANSD. However, this electrophysiologic pattern is clearly abnormal, and these children should be closely monitored until behavioral audiograms are obtained and functional abilities can be determined.

Caution is also needed when interpreting abnormal ABR findings obtained in infants prior to 34 to 36 weeks gestation as maturation of the auditory pathway may be incomplete. Furthermore, because cases of transient ANSD have been reported when the initial ABR was obtained in the first few months of life, the ABR should be repeated at a later point in time to confirm the diagnosis (Madden et al., 2002; Psarommatis et al., 2006).

FIGURE 37–2. Example of stimulus artifact that may be misinterpreted as CM.
Acoustic immittance measures (tympanometry and acoustic reflex testing) are also included in the comprehensive evaluation of ANSD. Middle ear muscle reflexes (MEMR; both ipsilateral and contralateral) are usually absent or elevated in cases of ANSD (Berlin et al., 2005). For infants under 6 months of age it is important to include a high-frequency (e.g., 1000 Hz) probe tone frequency (Margolis, Bass-Ringdahl, Hanks, Holte, & Zapala, 2003).

**Behavioral Audiometry**

Physiologic measures such as ABR and ASSR are not true tests of hearing in the perceptual sense, yet for many years these tests have enabled pediatric audiologists to make predictions regarding behavioral hearing thresholds. It is important to emphasize that in children with ANSD, thresholds cannot be predicted using ABR or ASSR. Thus, accurate behavioral audiometric procedures must be completed as soon as the child is developmentally capable of providing reliable responses. Behavioral audiometry using a conditioned response procedure such as visual reinforcement audiometry (VRA) can be initiated at a developmental level of 6 to 7 months. Using insert earphones attached to either foam tips or the infant’s custom earmolds, it is often possible to obtain ear and frequency-specific measures by 7 to 9 months of age for typically developing infants (Widen et al., 2000). However, many infants identified with ANSD are born prematurely or have complex medical conditions putting them at risk for delayed motor and/or cognitive development (Teagle et al., 2010). These children are less likely to perform conditioned response procedures at the same age as healthy, typically developing infants, and testing at multiple intervals over a period of weeks or months may be required to obtain a complete audiogram. Furthermore, some infants with complex medical conditions or developmental delays may never be able to perform behavioral audiometry well enough to provide reliable estimates of hearing thresholds. In these cases behavioral observation audiometry (BOA) may be used; however, it is important to recognize the inter- and intra-subject variability inherent in this procedure (Widen, 1993) and the limitations of BOA for purposes of hearing aid fitting. In cases where it is unlikely the infant will perform VRA reliably in a reasonable timeframe for decisions regarding intervention, cortical evoked potentials may be a useful supplement to BOA for determining the child’s auditory capacity (Pearce et al., 2007; Wunderlich & Cone-Wesson, 2006).
Medical Assessment

A comprehensive examination by an otolaryngologist is recommended for all children suspected of having ANSD. The otolaryngologist will obtain a medical history, perform a head and neck examination, and order laboratory studies needed to determine the etiology of the hearing loss or to identify coexisting conditions. Radiologic assessment using MRI is essential in ANSD since the auditory neuropathy phenotype is often present in children who have cochlear nerve deficiency (small or absent VIII nerves; Buchman et al., 2006). Furthermore, children with ANSD also have a higher incidence of other abnormal MRI findings. In a group of 118 children with ANSD who had available imaging studies at the University of North Carolina, nearly 65% had at least one abnormal finding on MRI (Roche et al., 2010). The abnormalities identified in addition to cochlear nerve deficiency included: prominent temporal horns; abnormalities of the brainstem, cerebellum, midbrain or cerebrum; cerebrospinal fluid (CSF) and ventricular abnormalities; white matter changes; Dandy Walker malformation; and Arnold Chiari Type I malformation. In contrast, only approximately 30% of children with non-ANSD hearing loss have been reported to have abnormal MRI findings (Mafong, Shin, & Lalwani, 2002; Simons, Mandell, & Arjmand, 2006).

Other medical consultations include evaluation by an ophthalmologist to assess visual acuity and to rule out concomitant visual disorders, and referral for medical genetics to determine if there is a genetic basis for the disorder. Although not routinely recommended for children with sensory hearing loss, referral to a pediatric neurologist is recommended for children diagnosed with ANSD since some may have neurologic disease or other conditions requiring medical treatment. It is important to inform the child’s primary care physician of the ANSD diagnosis and related findings, and to provide information regarding the disorder and how it will be treated.

Evidence to Guide Management Decisions

Studies of children with ANSD show considerable variability in auditory capacity. Among the clinical characteristics reported are pure tone thresholds that range from normal to profound; disproportionately poor speech recognition abilities for the degree of hearing loss; difficulty hearing in noise; and impaired temporal processing (Rance et al., 2002; Rance, McKay, & Grayden, 2004; Starr et al., 1996; Zeng & Liu, 2006; Zeng, Oba, Garde, Sininger, & Starr, 1999). It is important to recognize that the characteristics of ANSD vary, and not every individual diagnosed with the condition will present with the same symptoms or level of severity. Some children with ANSD have disproportionately poor speech recognition ability for their degree of hearing loss, while others perform at a level similar to peers with non-ANSD hearing loss. For example, Rance and colleagues in Australia compared unaided and aided speech perception abilities for a group of 15 children with ANSD to a group with typical sensory hearing loss matched for age and hearing level (Rance et al., 2002). Their results showed that approximately 50% of the children with ANSD had speech recognition scores that were similar to the children with sensory hearing loss; the other 50% showed essentially no open-set speech perception ability. Interestingly, the children showing no open-set speech perception had absent cortical evoked potentials, whereas the children who had measureable speech recognition scores had present cortical evoked potentials.

Similarly, although it has been reported that children with ANSD have particular difficulty hearing in the presence of background noise (Gravel & Stapells, 1993; Kraus et al., 2000), a recent study by Rance and colleagues showed that children with typical sensorineural hearing loss and those with ANSD had more difficulty in noise than children with normal hearing. However, the effects were not consistent across subjects and some children with ANSD showed relatively good speech perception abilities even at low signal to noise ratios (Rance et al., 2007).

Following the initial report by Starr and colleagues (1991) describing patients with what appeared to be a “neural” hearing loss, several journal articles and book chapters have included recommendations for clinical management. Recommendations include: low gain hearing aids or FM systems; low gain hearing aids in one ear only; or the avoidance of hearing aid use altogether (Berlin, 1996, 1999; Berlin, Morlet, & Hood, 2003). Furthermore, because early reports described what appeared to be pathology of the auditory nerve, it was initially thought that cochlear implantation would not be beneficial (Cone-Wesson, Rance, & Sininger, 2001; Miyamoto, Kirk, Renshaw, & Hussain, 1999). Over time, as more young children diagnosed with ANSD have been evaluated, investigators have shown that both hearing aid use and cochlear implantation can be of benefit to some children with ANSD (Buss et al., 2002; Rance et al., 1999; Rance et al., 2002;
Rance & Barker, 2008; Rance, Barker, Sarant, & Ching, 2007). The clinician’s challenge is to determine, as soon as possible, the hearing technology that will provide the most benefit for a particular child. Further research is needed to guide the clinician in predicting, at an early age, the technology and communication strategies that will be most beneficial for each child.

### Hearing Aids and FM Systems

Currently available clinical tests provide limited information regarding site of lesion and, in many cases, it is difficult to predict whether a given child will benefit from amplification. In cases where there is residual hearing, and once reliable threshold estimates have been obtained, a trial period with amplification using an evidence-based hearing aid fitting protocol should be completed (American Academy of Audiology Pediatric Amplification Guidelines 2003; Bagatto, Scollie, Hyde, & Seewald, 2010). This recommendation was also endorsed by the panel convened for the Guidelines Development Conference on the Identification and Management of Infants with Auditory Neuropathy (2008) in Como, Italy.

Established protocols for children include real ear measures or simulated real ear measures based on real-ear-to-coupler differences (RECD) and use of a prescriptive hearing aid fitting method (e.g., desired sensation level or National Acoustics Laboratory Approaches), to ensure that speech at conversational levels is audible and comfortable. When managing amplification in these children the clinician must keep in mind that ANSD is thought to cause a disruption in temporal rather than spectral processing (Rance et al., 2004; Zeng et al., 1999). As such, improving the audibility of the signal may not be sufficient to allow a child to make adequate progress with spoken language. Studies also have shown that in children with ANSD it is possible to have varying degrees of temporal disruption (Rance et al., 2004). Thus, one might expect better performance with acoustic amplification in individuals who have milder forms of the disorder, although at the present time there is limited peer-reviewed literature regarding the benefits of amplification in children with ANSD.

Finally, considering the likelihood of difficulty hearing in the presence of background noise, use of a personal FM system by parents and other caregivers may be beneficial. As with children who have non-ANSD hearing loss, the use of an FM system in the classroom is especially important to reduce problems related to distance, reverberation, and background noise.

### Evaluating Outcomes

Once hearing aids have been provided it is important to evaluate the child’s speech perception ability. Although evaluation of speech perception abilities in young children is challenging, a battery of age-appropriate tests such as those used by cochlear implant teams allow the pediatric audiologist to evaluate a child’s unaided and aided performance. Parent questionnaires such as the Infant-Toddler: Meaningful Auditory Integration Scale (IT-MAIS; Zimmerman-Phillips, Robbins, & Osberger, 1997), informal tests such as identification of body parts, as well as closed set speech perception tests such as the Early Speech Perception test (ESP; Moog & Geers, 2003) may be useful in assessing progress when children are too young for open-set testing. Once the child is able to perform open-set speech recognition testing, measures such as the Multisyllabic Lexical Neighborhood Test (MLNT) and the Lexical Neighborhood Test (LNT; Kirk, Pisoni, Sommers, Young, & Evanson, 1995) may be used. These tests use vocabulary in the lexicon of children under 5 years of age.

For a review of speech recognition testing in children less than 3 years of age, see Eisenberg, Johnson, and Martinez (2005).

When children reach 5 years of age, the phonetically balanced kindergarten words (PBKs; Haskins, 1949) may be used. It is important to use recorded speech materials whenever possible. In addition to evaluation of speech perception abilities, the child’s speech and language development must be carefully evaluated to monitor communication milestones. As with young children who have sensory hearing loss, the pediatric audiologist is advised to partner with speech-language pathologists and early intervention specialists. Experienced clinicians who know what to expect from children with varying degrees of sensory loss are essential to the process of monitoring communication development. Changes to the intervention strategy may be needed based on the child’s progress and the preferences of the family.

### Considerations for Cochlear Implantation

Children diagnosed with ANSD who exhibit severe to profound detection levels with stable thresholds over a period of several months, and whose families desire a spoken language approach, are often good candidates for cochlear implantation. For children without additional disabilities, decisions regarding cochlear implantation usually are uncomplicated once MRI has confirmed auditory nerve sufficiency (Buchman et al.,
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Decisions regarding the advisability of cochlear implantation for children with lesser degrees of hearing sensitivity loss are more challenging, due to the range of functional abilities seen in this population and the difficulty determining the degree of impairment in very young children with ANSD. Because the behavioral pure-tone audiogram is of limited prognostic value in the prediction of aided benefit in children with ANSD, cochlear implantation should be considered even if audiometric thresholds are better than what typically would be considered when progress with conventional amplification is inadequate. For children with ANSD who have substantial residual hearing, an adequate trial with amplification, appropriate early intervention services, and comprehensive evaluation by a team of professionals experienced in the evaluation and management of young children with hearing loss, are all prerequisites to cochlear implantation. For any child with a hearing disorder a variety of factors may influence developmental outcomes. These include age at diagnosis and hearing aid fitting, consistency of hearing aid use, quality and intensity of early intervention services, and presence of additional developmental and/or medical conditions. The amount of time needed to determine benefit from hearing aid use will vary depending on each of these factors. Rather than identify an arbitrary time period for determining whether the child is receiving sufficient benefit from amplification, it is the role of the pediatric audiologist, in partnership with the family and other team members, to identify the possible influence of each factor and its impact on developmental outcomes. Optimal management requires careful observation and a comprehensive team approach.

In the United States, candidacy for cochlear implantation is based on criteria developed by the Food and Drug Administration (FDA) using information obtained during clinical trials; the criteria specified on the device labeling varies by manufacturer. At the time of this writing, children between 12 and 18 to 24 months may be considered for cochlear implantation if they have a profound bilateral sensorineural hearing loss. Children older than 18 months with severe to profound hearing loss may be considered for cochlear implantation if there is a “failure to meet auditory milestones” or if performance in the best aided condition is less than 20 to 30% for MLNT or LNT at 70 dB SPL. The criteria for one manufacturer states that in children older than age 4 years, cochlear implantation may be considered if the score is less than 12% on PBK words or 30% on open-set sentences presented at 70 dB SPL. Although no specific FDA guidelines for cochlear implant candidacy currently exist for children with ANSD, professionals attempting to determine CI candidacy for children with ANSD who have less than a profound hearing loss should consider these general guidelines.

Counseling Families

Professionals who work with families of infants and young children with newly diagnosed hearing loss understand the critical role of counseling in helping a family understand the nature of the child’s hearing loss and the implications of the diagnosis. Families need assistance with the emotional aspects of the new diagnosis and information they will need to make the best decisions for their child. This is a challenging process for the family and for the clinician. Families receiving a diagnosis of ANSD face additional challenges due to the uncertainties inherent at the time of the diagnosis and the complexities associated with management decisions for their child. When delivering a diagnosis of ANSD it is important to share information based on the best available scientific evidence while providing the family with hope. For the young infant diagnosed with ANSD through electrophysiologic testing, the audiologist knows little more at the time of diagnosis than that the infant has an electrophysiologic pattern that is abnormal. Considering the heterogeneous nature of the disorder and range of functional outcomes, it makes little sense to make definitive predictions to the family regarding “expected” auditory behaviors until additional diagnostic information has been obtained.

For an infant only a few weeks of age, it may be appropriate to simply advise the family that the results of the ABR are not normal and that testing should be repeated in a few weeks. If the infant returns for a repeat study that yields similar results, additional information regarding ANSD should be provided to the family. For some families this may include showing them the ABR waveforms and contrasting them with expected results for children with non-ANSD hearing loss, as well as for a child with normal hearing sensitivity. Other families will prefer a more basic explanation. It is important for families to understand
that an MRI will be needed to rule out cochlear nerve deficiency. Once cochlear nerve status has been determined, families will benefit from knowing the heterogeneous nature of ANSD and the variable outcomes associated with the disorder. It is important for families to understand that, unlike with non-ANSD hearing loss, the ABR does not assist in predicting the degree of hearing loss or in establishing thresholds for purposes of hearing aid fitting.

It is helpful for families to have a timeline for management during the first year including the need for medical evaluations, enrollment in intervention and for behavioral audiology to establish thresholds beginning at 6 to 7 months of age. They will need to understand that decisions regarding amplification may need to be deferred until behavioral audiometric thresholds can be established.

If the child is determined to be a good candidate for amplification, the family must understand the importance of full time hearing aid use and the need to monitor communication milestones. Cochlear implantation may be discussed as one of several interventions that might be considered, but it should be explained that decisions regarding implantation need to be deferred until behavioral thresholds are established and benefit from hearing aid use has been determined. As discussed earlier, decisions regarding continuation of hearing aid use versus cochlear implantation must be made on an individual basis and determined by the needs of the child and the preferences of the family. Further research is needed to understand ANSD, its diagnosis, and its optimal management, but families should leave the clinic knowing that much can be done to facilitate their child’s acquisition of functional communication ability.

Case Studies

As discussed above, the electrophysiologic test results that are characteristic of ANSD show a wide range of functional outcomes. The following case illustrations will demonstrate the variable results obtained from children who exhibit the audiological profile of ANSD. Key points are made to highlight the different habilitative recommendations made and/or outcomes obtained for each case.

Case 1

This child was born at 25 weeks gestation, with a history of hyperbilirubinemia that required an exchange transfusion. He was ventilated for six weeks and was on oxygen for 3? months. He did not pass his newborn hearing screen with automated ABR and was referred for diagnostic ABR testing. Following the ABR testing with tone bursts and clicks, the parents were told that the infant had a profound hearing loss, and he was fitted with high gain hearing aids. The father had a job transfer and the family moved into our state. They were told that another ABR would be needed prior to acceptance into our program. On the day of the sedated ABR evaluation, the family reported that in spite of the diagnosis of profound hearing loss, they observed the baby respond to a variety of sounds at home and had even observed him startle to a loud sound.

Subsequent ABR testing showed an absent ABR with only a large CM present consistent with a diagnosis of ANSD. Otoacoustic emissions were absent bilaterally (Figure 37–4).

The diagnosis was explained to the family, and it was recommended that they discontinue use of the high gain hearing aids until behavioral audiometry could be completed. The child was enrolled in an early intervention program, and the family was informed of various communication options. They chose spoken language as their initial communication approach and were assigned an auditory verbal therapist who provided weekly visits to the family’s home. The child was seen by an otolaryngologist who recommended an MRI. The MRI showed normal inner ear anatomy with present auditory nerves bilaterally. Subsequent behavioral audiometric testing at 10 months of age (7 months adjusted age) using VRA showed a moderate bilateral sensorineural hearing loss (Figure 37–5).

Once behavioral thresholds were obtained the family decided to move forward with amplification, and the child was fitted with hearing aids appropriate for a moderate hearing loss. In addition to hearing aids, the family used a personal FM system at home once the child began to walk, and there was increasing distance between the child and the speaker. Aided speech perception measures showed aided benefit, and subsequent speech and language evaluations showed the child was making excellent progress in meeting communication milestones. At 3½ years of age, the child had recurrent middle ear problems and was scheduled for placement of tympanostomy tubes. ABR testing was repeated and again showed no neural responses to click stimuli at high intensity levels with only a CM present. The child is now 5½ years old and has successfully completed kindergarten in a mainstream classroom. At the time of his most recent audiologic evaluation his speech reception thresholds were 60 dB
HL unaided and 35 dB HL aided and he scored 0% on a monosyllabic word test (LNT) at 55 dB HL unaided and 100% aided. His most recent speech and language evaluation showed age-appropriate language skills with some mild speech production problems. He continues to receive support from a teacher of the deaf and a speech and language pathologist and uses FM in the classroom.

**Key points:** In this case, the first ABR was done at a clinic that used alternating polarity rather than single polarity clicks. This resulted in an incorrect diagnosis of profound bilateral sensory hearing loss. Fortunately, the family’s move to another state resulted in the child having a repeat ABR study. The case illustrates the importance of asking the family about their observations of the child’s auditory behaviors. Although the child was initially diagnosed with profound hearing loss, it was obvious to the family that their baby was responding to moderately loud sounds at home even without amplification, an unexpected finding with profound bilateral hearing loss. It also illustrates the importance of accurate initial diagnosis, as habilitative recommendations are often based on the initial diagnostic ABR evaluation. Finally, this is an example

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**FIGURE 37-4.** A. ABR for child in Case 1. Note large CM with reversal of waveform when stimulus polarity is inverted and lack of neural response. B. OAEs for Case 1.
of a child who shows electrophysiologic findings that are characteristic of auditory neuropathy, yet receives significant benefit from acoustic amplification.

**Case 2**

The second case is a child who was born at full term in a hospital that used only otoacoustic emissions for their infant hearing screening program. The child passed his newborn OAE screen bilaterally. At age 3½ years, he subsequently developed recurrent middle ear problems and was scheduled for myringotomy and tube placement. His postoperative hearing evaluation showed normal hearing sensitivity for the right ear and a profound bilateral sensorineural hearing loss for the left ear (Figure 37–6A). Unexpectedly with a profound hearing loss in one ear, otoacoustic emissions were present bilaterally (Figure 37–6B).

The child was referred to our center for further evaluation. Diagnostic ABR testing showed, in the right ear, normal waveform morphology with responses consistent with normal hearing sensitivity; in the left ear, the ABR indicated absent neural responses with only a prolonged CM present at high intensity levels (Figure 37–7). MRI testing was completed and revealed a normal study for the right ear and an absent auditory nerve for the left ear.

**Key points:** As previously noted in this chapter, individuals who have cochlear nerve deficiency (small or absent VIII nerves) often present with the phenotype of ANSD (absent ABR, present CM and present OAEs; Buchman et al., 2006). MRI is useful in identifying the site of lesion in these cases and will assist the audiologist and intervention specialist in making appropriate management recommendations. Use of radiologic imaging is particularly important when determining cochlear implant candidacy in cases of profound bilateral hearing loss when there is no evidence of residual hearing. In this child’s case, availability of MRI findings allowed the pediatric audiologist to make recommendations for management that were the same as those recommended for a child with profound, unilateral, sensory hearing loss.

Another key point is that it is possible to pass OAE-based newborn hearing screening and yet have a profound hearing loss in one or both ears. Professionals must be aware that passing newborn hearing screening does not ensure normal hearing and further diagnostic study is warranted when there is concern about hearing status even when an individual has passed a newborn hearing screen.
FIGURE 37–6. **A.** Audiogram for child in Case 2. **B.** OAEs for Case 2.
Case 3

This is a child who was born prematurely at 24 weeks gestation. He was hospitalized in the newborn intensive care nursery (NICU) where he received ventilation. He did not pass his initial newborn hearing screen or a repeat screen using automated ABR. Subsequent diagnostic ABR testing at 4 months of age showed absent neural responses with only a CM present for single polarity clicks at a high intensity level (Figure 37–8A). Otoacoustic emissions were present bilaterally (Figure 37–8B).

Behavioral audiometry was attempted at regular intervals beginning at 7 months adjusted age. At 12 months of age (8 months adjusted age), a sound-field audiogram showed normal hearing sensitivity for 250 to 4000 Hz. At 13 months of age (9 months adjusted age), individual ear measures were completed with insert earphones and confirmed normal hearing sensitivity for each ear (Figure 37–9).

The results were discussed with the family and the importance of monitoring the child’s communication status in view of the abnormal ABR pattern was discussed. The child was enrolled in an early intervention program, and a teacher visited the home on a monthly basis. Because of reports of “recovery” in cases of ANSD, ABR testing was repeated at 17 months of age and test results again showed an abnormal ABR with no neural responses at maximum intensity levels for click stimuli with only a CM present.

It was recommended that the child return for audiologic evaluation every six months to monitor the stability of his thresholds and to obtain speech perception measures once he was developmentally able to perform this testing. At age 3, the child scored 100% on a closed-set monosyllabic word test (Early Speech Perception Test; ESP). The child developed spoken language and at 3½ years of age he was able to comprehend speech and use multiple word sentences expressively. He was also able to repeat monosyllabic words in quiet without difficulty in an auditory only condition.

Key points: In this case, although the child’s ABR was grossly abnormal showing absent neural responses, the child’s audiogram demonstrates normal hearing sensitivity. While the child is not currently exhibiting any functional difficulty and is developing speech and language appropriately, it will be important to monitor performance at regular intervals and continue early intervention services. It also will be important to monitor the child’s ability to hear in the presence of background noise and consider use of FM in the classroom if the child experiences difficulty hearing in noise.
FIGURE 37–8. A. ABR showing no neural response with only a CM for child in Case 3. B. OAEs for child in Case 3.
outside clinic had recognized the importance of not re-screening with otoacoustic emissions when the child failed the ABR screen. Fortunately, the family obtained a second opinion in sufficient time for her to receive appropriate management. If a child does not pass a hearing screen with ABR, the child either should have a second level screen with ABR or a diagnostic ABR as a follow up. In many clinics ABR has replaced OAE screening in the intensive care nursery due to the high prevalence of ANSD in this population; however, screening with OAEs is still common in many well baby nurseries. Although the majority of cases of ANSD will be found in the NICU, it is possible to have a full term birth without complications yet present with ANSD. Clinicians must be mindful of the possibility of ANSD when there is suspicion of hearing loss in a full term infant who has passed a newborn hearing screen with OAEs. In some of these cases, a genetic basis for the ANSD may be the etiology of the disorder.

**Case 4**

This child was born at full term without any significant complications. There was no family history of hearing loss. This child did not pass an automated ABR hearing screen at birth but passed a re-screen with otoacoustic emissions at an outside ENT office, and her parents were told she had normal hearing. At 8 months of age, her parents became suspicious that she was not hearing and brought her to our university hospital clinic for a diagnostic ABR. The results of the diagnostic ABR evaluation showed absent neural responses to clicks with only a CM present at high intensity levels (Figure 37–10A). Otoacoustic emissions were present bilaterally (Figure 37–10B).

MRI evaluation was normal and indicated the presence of auditory nerves bilaterally. Behavioral audiometry at 9 months of age using VRA was consistent with a profound bilateral sensorineural hearing loss (Figure 37–11).

She was fitted with high gain hearing aids; however, her parents did not observe any improvement in her responses to sound with amplification, and she was subsequently referred to the cochlear implant team for evaluation. She received a right cochlear implant at 21 months of age.

**Key points:** In this case, the child’s profound hearing loss would have been identified earlier if the outside clinic had recognized the importance of not re-screening with otoacoustic emissions when the child failed the ABR screen. Fortunately, the family obtained a second opinion in sufficient time for her to receive appropriate management. If a child does not pass a hearing screen with ABR, the child either should have a second level screen with ABR or a diagnostic ABR as a follow up. In many clinics ABR has replaced OAE screening in the intensive care nursery due to the high prevalence of ANSD in this population; however, screening with OAEs is still common in many well baby nurseries. Although the majority of cases of ANSD will be found in the NICU, it is possible to have a full term birth without complications yet present with ANSD. Clinicians must be mindful of the possibility of ANSD when there is suspicion of hearing loss in a full term infant who has passed a newborn hearing screen with OAEs. In some of these cases, a genetic basis for the ANSD may be the etiology of the disorder.

**Case 5**

This is a child who had normal development for the first two years of life and then developed peripheral neuropathies including optic neuropathy. She was hospitalized at 3 years of age and underwent several diagnostic studies including electromyography and muscle biopsies. She has had numerous specialty med-
ical consultations including: otolaryngology, genetics, neurology, ophthalmology, and infectious disease. The etiology for her medical problems was never determined; Guillain-Barré, Charcot-Marie-Tooth and mitochondrial disease were all ruled out. An audiogram obtained at age 6 years showed a bilateral low-frequency hearing loss with unaided speech recognition scores of 100% for the right ear and 84% for the left (Figure 37–12).

An audiogram at age 10 years showed a bilateral rising audiogram; mild on the right and moderate on the left with speech recognition scores using monosyllabic words of 40% for the right at 65 dB HL and 24% for the left at 95 dB HL (Figure 37–13).
FIGURE 37–12. Audiogram showing rising low frequency hearing loss for child in Case 5.

FIGURE 37–13. Audiogram showing left ear worse than right for child in Case 5.
Subsequent hearing evaluations showed fluctuating speech recognition scores and a subsequent diagnostic ABR evaluation showed a pattern consistent with ANSD. Hearing aids were tried; however, the child and family reported they were of minimal benefit. By age 11 years, despite an audiogram showing significant residual hearing (Figure 37–14A), the child was unable to repeat any monosyllabic words on a speech recognition test and successful communication could only be accomplished at close range with lip reading. Robust otoacoustic emissions were present bilaterally (Figure 37–14B).

The family was counseled extensively regarding potential benefits and limitations of cochlear implantation, particularly in view of her history of multiple peripheral neuropathies. After careful consideration, the

![Figure 37–14](image-url)

**FIGURE 37–14.** A. Audiogram showing severe hearing loss for child in Case 5. B. Present OAEs for child in Case 5.
family decided to proceed with a left cochlear implant. After one year of device use, the child’s monosyllabic word score with her cochlear implant was 32% on words and 66% on phonemes. Her parents reported that while she continued to have significant communication difficulty, they felt that the need for repetitions was reduced with the device on. After four years of device use, her speech recognition score while wearing her CI was only 20% and she continued to have deterioration in her motor abilities. The parents reported that she had significant difficulty understanding anyone other than her family members with her implant. Since the child still had significant residual hearing in her right ear, a decision was made to attempt hearing aid use again in the right ear. At the age of 17 and after six years of implant use and with a hearing aid in the contralateral ear, this child only achieves a score of 20% for monosyllabic words.

Key points: This child’s case is complex and is similar to cases described by Dr. Arnold Starr and his colleagues in 1996. It likely represents a true case of “auditory neuropathy.” Despite the use of hearing aids, a cochlear implant, supplemental visual input, and the best effort of her parents and the professionals who work with her, this child continued to have significant communication difficulty.

Conclusion

The disorder described as AN is more complicated than originally thought and the patient population is more heterogeneous. Early recommendations were often based on findings in adults with other peripheral neuropathies. Hearing aids, cochlear implants and other management strategies were both promoted and discouraged based on minimal evidence. There is now a considerable body of clinical evidence that indicates some children with ANSD are good candidates for amplification while others obtain greater benefit from cochlear implantation. With either technology a child’s performance may differ from that expected in children with cochlear hearing loss, and in some cases neither strategy provides sufficient benefit. There is a growing body of literature but the available evidence to guide clinical management of ANSD remains limited and more research is needed, especially with infants and young children. Considering the likelihood of varied etiologies, sites of lesion, age of identification, and risks of cognitive/developmental delays, it is unlikely that a single management strategy will apply to all infants and young children who present with this problem of audiologic findings. As with other sensorineural hearing loss, a continuum of multidisciplinary care is needed to provide optimal management of infants and children with ANSD.

References


