

# **Audiologic Diagnosis of Infants: Two Cases of Auditory Neuropathy Spectrum Disorder**

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## **Introduction**

Over the past 20 years, remarkable progress has been made in the United States toward the goal of screening all newborns for hearing loss by 1 month of age. According to the National Center for Hearing Assessment and Management, nearly all States in the US now provide newborn hearing screening prior to discharge from the birthing hospital (NCHAM 2011). This contrasts sharply to the early 1990s when only a few States screened all infants for hearing loss at birth. As a result of newborn hearing screening, many infants receive diagnosis of hearing loss by 3 months of age; are fitted with hearing aids within one month of diagnosis; and are enrolled in early intervention by 6 months of age, as recommended in 2007 by the Joint Committee on Infant Hearing (JCIH 2007). Early identification and intervention have made it possible for many infants to attain communication milestones on par with their hearing peers. Despite these advancements, some families report frustration that their child's hearing loss was confirmed well beyond the first year of life, even though the infant did not pass a newborn hearing screen. In some cases, inaccurate results were given at the time of initial screening whereas in others referrals were made to audiologists who were not qualified to perform accurate and comprehensive pediatric assessment of a young infant. Because the initial diagnostic evaluation creates a foundation for later habilitative intervention, inaccuracies at this stage can result in significantly delayed hearing aid fitting or cochlear implantation.

In 2001, Dr. Judith Gravel, in an article entitled: "Potential Pitfalls in the Audiological Assessment of Infants and Young Children" described three types of audiologic misdiagnoses: 1) confirmation of hearing loss but with incorrect conclusions regarding type or degree of hearing loss; 2) diagnosis of hearing loss in a child with normal hearing, that is, false positive findings; and 3) diagnosis of normal hearing in a child with hearing loss, that is, false negative findings (Gravel 2001). She noted that these errors can result in: delayed confirmation of the child's true hearing status; delayed referral for a treatable medical condition; delayed referral for early intervention services; inappropriate audiological, medical, surgical, prosthetic, educational, or communicative intervention; parental anxiety, confusion, or loss of confidence; unnecessary expenditure of family or health care resources; and reduced confidence in audiologists by others in the healthcare system (policy makers, legislators, physicians, educators). Dr. Gravel was a passionate and tireless advocate for children with hearing loss and devoted much of her professional life to promoting evidenced-based procedures for the audiologic management of infants and young children. Her words, which remain true today, continue to inspire us to pursue the highest level of care for the families we serve.

The advancement of clinical practice requires well-controlled, cohort studies involving groups of children followed prospectively. Case presentations, although they are a lower level of evidence, provide an important medium for sharing outcomes that occur with individual patients and families. The cases reported here involve auditory neuropathy spectrum disorder (ANS), a hearing impairment characterized by absent or abnormal auditory brainstem responses (ABR) with evidence of normal outer hair cell function as indicated by the presence of cochlear microphonics and/or otoacoustic emis-

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sions (Rance and Starr 2010). Unfortunately, diagnostic errors can occur in patients with ANSD unless appropriate diagnostic protocols are carefully followed (Roush 2010). The two cases reported here illustrate misdiagnoses resulting in delayed or inappropriate treatment that could have been avoided by using established, evidenced-based methods of pediatric assessment.

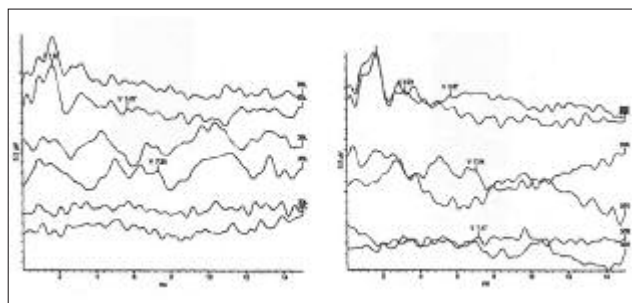
## Case Examples

### Case 1

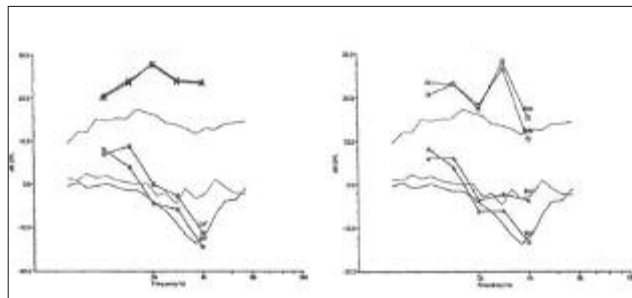
#### *Background and Initial Diagnosis*

Case 1 is a child born full term, without complications, to two deaf parents. He did not pass the initial newborn screen with automated ABR (AABR) in the right ear and passed in the left. He was seen for a re-screen and did not pass AABR in either ear. At 1 month of age, the parents were seen at a local ENT clinic where an audiologist performed a diagnostic ABR using 500 Hz tone burst and click stimuli, tympanometry using a 1000 Hz probe tone and otoacoustic emissions. Tympanometry was reported as normal. The results of the ABR and otoacoustic emissions testing were interpreted as normal (Figures 1a and b). The written report from this evaluation stated: “The results of ABR testing rule out a significant hearing loss for each ear at 500 Hz and within the 1000–4000 Hz region. Recommend return in six months for behavioral assessment.” At 7 months of age the parents, accompanied by the maternal grandmother who had normal hearing, returned to the clinic as recommended. Behavioral audiometry was not completed, however; another ABR using click stimuli was completed in natural sleep (Figure 2). The written report from this second diagnostic ABR stated, “ABR to click stimuli yielded a reliable response for intensity levels down to at least 30 dB HL for each ear, tympanometry was consistent with normal middle ear function and robust DPOAEs were present bilaterally (1000–4000 Hz). The results of ABR testing again rule out a significant hearing loss in the 1000–4000 Hz region.” The family was told that because the parents were deaf, it was important that they provide language stimulation to the child by talking to him during daily care activities, reading to him, etc. It was also recommended that they return for behavioral audiometry in three months. Over the next few months, the parents became concerned that their child, whom they assumed had normal hearing, was not developing speech, so they quit their jobs in

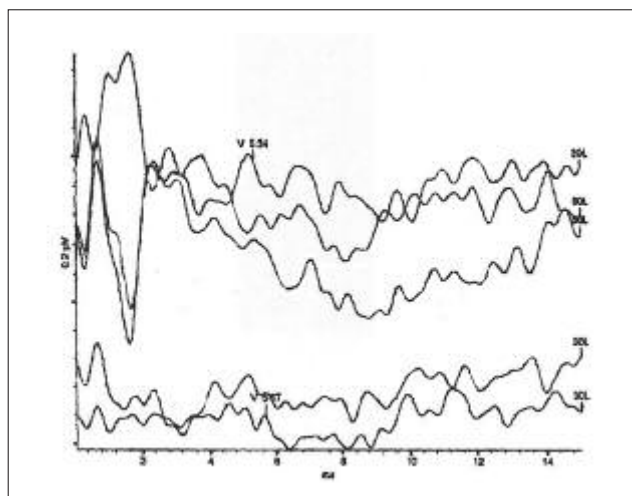
the community where they were living and moved 200 miles away to live with the maternal grandparents so the child would have more exposure to spoken language. At age 19 months the grandparents noticed the child did not seem to be responding to sounds or developing speech, and they arranged for another evaluation at an



**Figure 1a.** First ABR obtained from ENT clinic for Case 1. Note the incorrect identification of waveforms.



**Figure 1b.** Present otoacoustic emissions for Case 1.



**Figure 2.** Second ABR obtained from ENT clinic for Case 1. Note the incorrect identification of waveforms.

ENT clinic near their home. Behavioral audiometry was performed and results were consistent with profound bilateral sensorineural hearing loss. Because the family had received conflicting information with two diagnostic ABRs indicating normal hearing and a behavioral test indicating profound hearing loss, the family decided to obtain another opinion and arranged to have the child evaluated in a large academic medical center that sees a large volume of infants and young children with hearing loss.

### Diagnosis Following Second Opinion

After a telephone interview with the grandmother who reported the child's history of multiple diagnostic evaluations with conflicting diagnoses, the child was scheduled for a two-day evaluation. On the first day, behavioral audiometry using VRA with insert earphones, acoustic immittance and otoacoustic emissions testing were completed. The results of the behavioral test confirmed profound bilateral hearing loss (Figure 3a). Tympanometry was consistent with normal middle ear function, acoustic reflexes were absent, and robust otoacoustic emissions were present bilaterally (Figure 3b). On the second day, a sedated ABR was completed and showed no neural responses at maximum intensity levels (90 dB nHL) for both rarefaction and condensation clicks; however, a cochlear microphonic was present consistent with auditory neuropathy spectrum disorder (ANS; see Figure 3c). Following the audiologic diagnosis, the otologist who examined the child ordered an MRI, EKG and genetic testing. Results of the MRI showed normal inner ear morphology, and the EKG was normal. Genetic testing was negative for connexin, but the otoferlin test was positive, indicating a genetic basis for this child's auditory neuropathy. The implications of severe to profound hearing loss and the auditory neuropathy diagnosis were discussed with the family who indicated they were interested in their child learning both sign language and spoken language. They were provided with information about hearing aids, cochlear implants and early intervention and contact information for other deaf parents who had chosen cochlear implantation for their children. The child was initially fitted with hearing aids and enrolled in an early intervention program that would facilitate the development of audition and spoken language as well as sign language communication, and at age 26 months the family decided to proceed with cochlear implantation for their son.

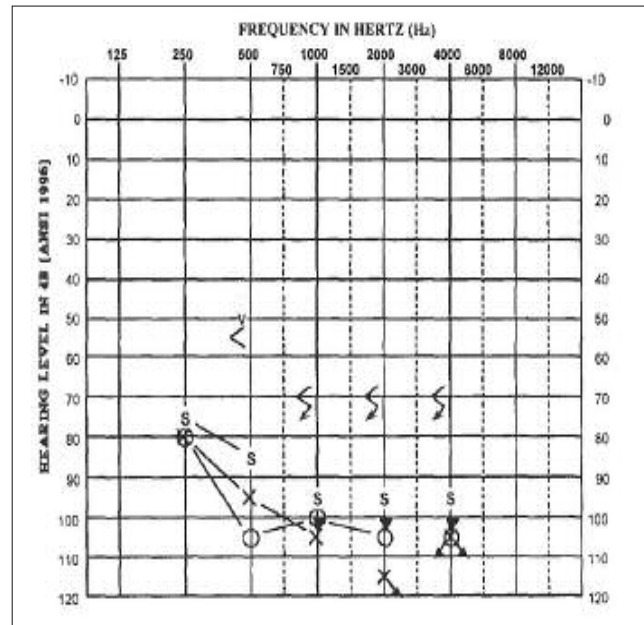


Figure 3a. Behavioral audiometry for Case 1 showing a severe-to-profound bilateral sensorineural hearing loss.

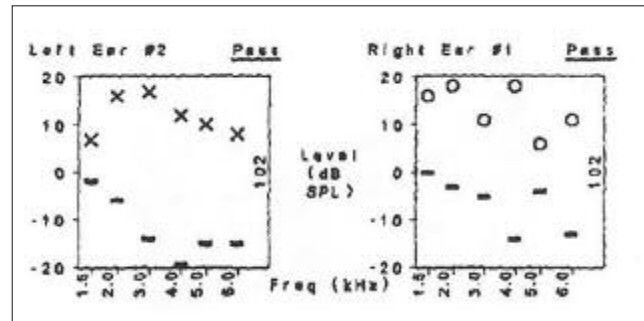


Figure 3b. Present distortion product otoacoustic emissions for Case 1.

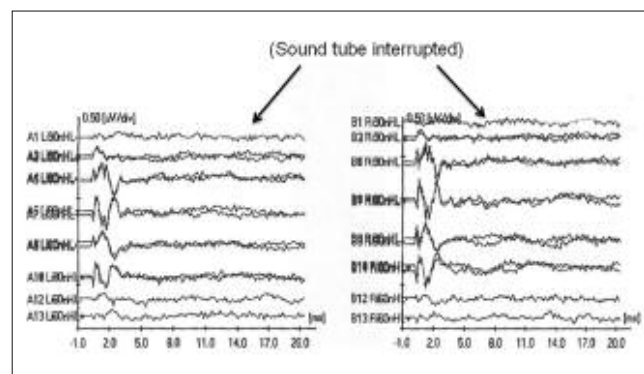


Figure 3c. ABR for Case 1 showing absent neural responses with only a cochlear microphonic present at high intensity levels (90 dB nHL) consistent with ANSD.

## Comment

Examination of the case history and test results from the child's initial evaluations reveal several problems that could have been avoided: 1) The audiologist performing the test likely assumed that hearing was normal based on present otoacoustic emissions at the time of the initial diagnostic test and interpreted the ABR as showing present waveforms at normal intensity levels; however, a review of the initial diagnostic ABR at 1 month of age reveals poor quality recordings with waveforms that were not reproducible, and latencies that did not increase with decreased intensity. Further, only rarefaction clicks were used at high intensity levels rather than both rarefaction and condensation. If both polarities had been used, it would have been possible to identify the presence of a cochlear microphonic. 2) At the time of the child's second visit at age 7 months, behavioral audiometry using VRA should have been completed to confirm the initial impression of normal hearing. The child in this case was born at full term and meeting normal developmental milestones. Behavioral audiometric testing using VRA is an essential component of any comprehensive diagnostic hearing evaluation and can easily be accomplished for most infants between 6 and 8 months of age. 3) Review of the second ABR did show evaluation of clicks using both rarefaction and condensation polarities and a present cochlear microphonic; however, no mention of a cochlear microphonic was made in the report. The ABR again showed noisy recordings and a poor quality study. 4) The parent's history of deafness and their expressed concern about their child's lack of responsiveness should have raised red flags in this case. An essential component of any pediatric audiology assessment is for the audiologist to take a careful case history and to listen to the parents' reports regarding their assessment of how their child hears. If results of our evaluation conflicts with their assessment, it is our role to understand why.

Fortunately, because the parents used sign language as a primary mode of communication in the home, this child had an extensive sign vocabulary and a way to communicate with his family despite the inaccurate initial diagnosis. However, the misdiagnosis in this case caused significant delays in intervention. Furthermore, the family made unnecessary changes in employment and living arrangements while experiencing considerable anxiety in the first two years of their child's life.

## Case 2

### *Background and Initial Diagnosis*

Case 2 is a 2 year old child born at 26 weeks gestation. He was hospitalized in the newborn intensive care nursery for several weeks where he received a brief period of artificial ventilation, treatment with bili lights for jaundice and two blood transfusions. He did not pass his newborn hearing screen with AABR in either ear. This child had four diagnostic ABR evaluations in the first six months of life, which are summarized below.

#### ABR #1 (age 1–2 months)

Summary: "Inconclusive due to movement and poor ear-phone fit."

ABR #2 (age 3 months) was completed using clicks with maximum intensity level of 80 dB nHL for the right ear and 70 dB nHL left ear (see Figure 4).

#### Summary:

- Right Ear: auditory neuropathy
- Left Ear: Borderline-normal to normal hearing
- "Hearing is adequate for speech and language acquisition."

ABR #3 (age 6 months) was completed using clicks with maximum intensity level of 70 dB nHL for the right ear and 85 dB nHL left ear (see Figure 5)

#### Summary:

- Bilateral auditory neuropathy
- "These results indicate change from previous study."

ABR #4 (age 6 ½ months) was completed using click stimuli with maximum intensity level of 80 dB nHL for each ear. Testing was also completed using tone bursts (see Figure 6).

#### Summary:

- Testing with clicks show only a cochlear microphonic for right and left ears
- Tone bursts are absent for the right ear and show mild to moderate hearing loss in the left ear
- Trial with amplification and evaluate for CI candidacy

At 7 months of age, the child was seen in the same clinic for behavioral audiometry. The report stated that the child was not responsive to pure tones, but a speech

awareness level was obtained at 35 dB HL and a startle was obtained at 90 dB HL. Otoacoustic emissions were absent for the right ear and present for the left. The results were discussed with the family and hearing aids were recommended. At the age of 9 months, binaural hearing aids were dispensed. At 18 months of age behavioral audiometry was repeated and responses were obtained to warbled pure tones in sound field at the

following levels: 500 Hz: 30 dB HL; 1000 Hz: 20 dB HL; 2000 Hz: 20 dB HL; and 4000 Hz: 30 dB HL. The family was advised that another ABR should be completed. The results of this ABR (the child's fifth ABR in the first 1½ years of life) were again interpreted as bilateral auditory neuropathy.

### Diagnosis Following Second Opinion

At age 20 months, the family moved to another state because of a work transfer. The parents scheduled an appointment with a pediatric audiologist at an academic medical center. The child was initially evaluated using behavioral audiometry with VRA and insert earphones. The results obtained were consistent with normal hearing sensitivity, bilaterally (Figure 7a). Tympanometry was consistent with normal middle ear function, acoustic reflexes were absent bilaterally and otoacoustic emissions were present, bilaterally (Figure 7b). The results were discussed with the family, and it was recommended that they discontinue use of the hearing aids, enroll in an early intervention program to monitor communication development and return for repeat behavioral testing in conjunction with an otologic exam in two months. Although the child had several ABRs in another facility, it was also recommended that he have another sedated ABR due to the conflicting information they had received in the past. At 28 months of age, the ABR was repeated showing an abnormal ABR consistent

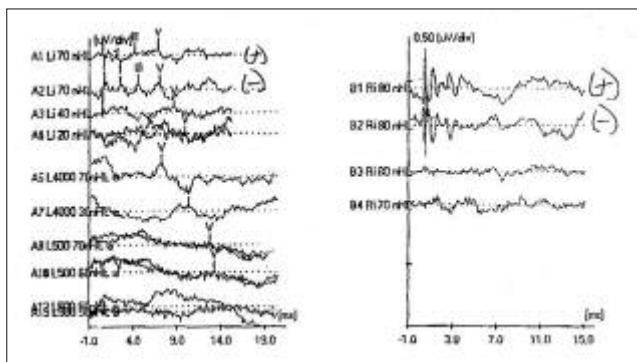


Figure 4. Second ABR for Case 2.

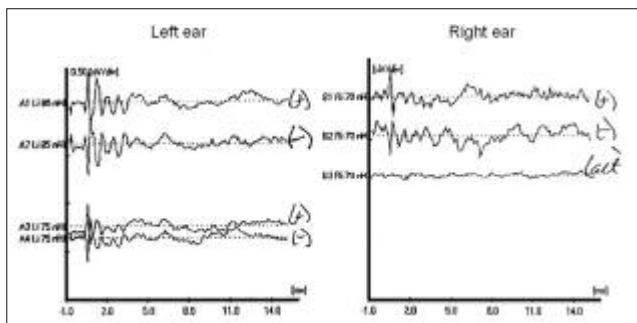


Figure 5. Third ABR for Case 2 showing maximum intensity levels of 85 dB nHL for the left ear and 70 dB nHL for the right ear.

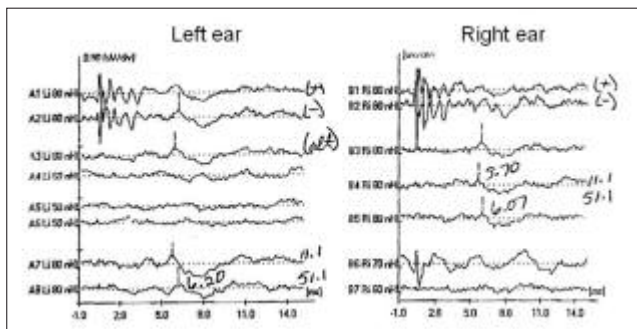


Figure 6. Fourth ABR obtained for Case 2. Note the present cochlear microphonics at high intensity levels (80 dB nHL for each ear).

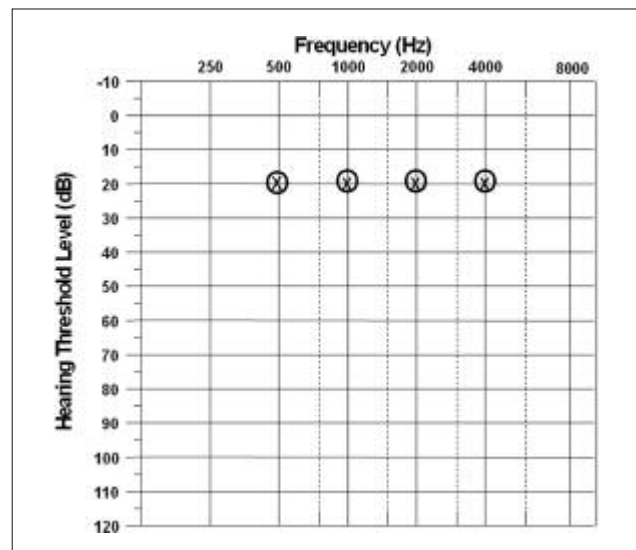
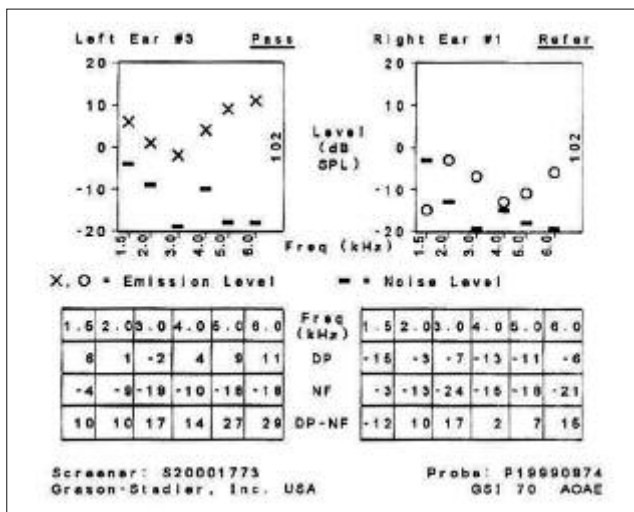
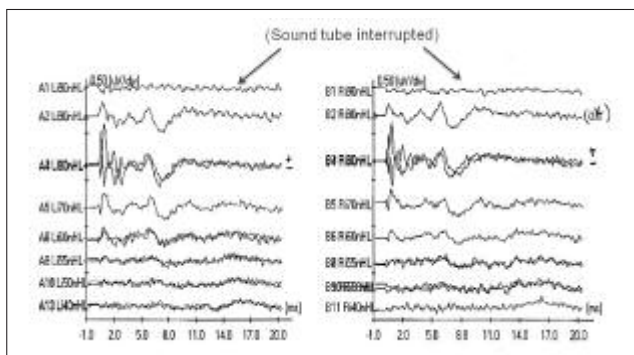


Figure 7a. Behavioral audiometry for Case 2 showing normal hearing sensitivity bilaterally.



**Figure 7b.** Distortion product otoacoustic emissions for Case 2.



**Figure 8.** ABR obtained for Case 2. Note the present cochlear microphonics at high intensity levels (90 dB nHL) with present distal waveforms.

with auditory neuropathy (Figure 8). A cochlear microphonic was present in the ABR completed with single polarity clicks at high intensity levels (90 dB nHL) with some distal waveforms present; however, repeatable waveforms were only observed down to a level of 40–50 dB HL for the left ear and 55 dB HL for the right ear.

A tone burst series was also completed, which also showed present cochlear microphonics with distal waveforms; however, as with the clicks, the waveforms were not observed at normal intensity levels. The child is continuing to receive early intervention services and is making good progress with spoken language communication. He continues to return for behavioral audiometry every six months and at the present time continues to show normal hearing sensitivity despite having an ABR that shows the ANSD pattern.

### Comment

This case illustrates the importance of following an established, evidenced-based protocol when evaluating infants suspected of having ANSD (Guidelines Development Conference on the Identification and Management of Infants with Auditory Neuropathy 2008; Buchman, Roush and Teagle 2008). The following steps should be completed to avoid an incorrect diagnosis: 1) whenever an ABR is absent or grossly abnormal at high intensity levels, evaluation should be completed using high intensity clicks (80-90 dB nHL) with both rarefaction and condensation polarities to determine if a cochlear microphonic is present. With surface recording electrodes, the cochlear microphonic can be identified in most cases at 80-90 dB nHL, but may not be present at lower levels. 2) A high intensity run should also be completed with the sound tube interrupted to insure that the responses obtained are a true physiologic response rather than stimulus artifact. 3) The ABR should not be used to estimate behavioral thresholds when waveform morphology at high intensity levels is clearly abnormal, showing absent or abnormal neural responses with present cochlear microphonic even when distal waveforms are present. 4) The use of behavioral audiometry with VRA to obtain individual ear and frequency specific measures is essential when attempting to determine a child's behavioral audiometric thresholds in all cases, especially in children suspected of having ANSD.

In this case, the use of different maximum intensity levels for click stimuli for each ear likely resulted in the audiologist reaching different conclusions regarding the diagnosis between the first and second tests. The ABR at 3 months of age (see Figure 4) was completed using click stimuli presented at a maximum intensity level of 70 dB nHL for the left ear and 80 dB nHL for the right ear. When the child returned at 6 months of age, the ABR was completed using click stimuli presented at a maximum intensity level of 85 dB nHL for the left ear and 70 dB nHL for the right ear. Furthermore, the use of the abnormal ABR to estimate behavioral thresholds resulted in estimates of behavioral thresholds that were elevated as compared to the child's actual thresholds and a recommendation for "trial of hearing aids and evaluation for a cochlear implant." In this case, multiple diagnostic ABRs were completed that served to confirm the diagnosis of ANSD; however, limited behavioral audiometric testing with VRA was performed, which would have demonstrated that the child had normal hearing sensitivity.

## Summary and Conclusions

Assessment of hearing in infants and toddlers requires experienced pediatric audiologists using protocols supported by high levels of evidence. Two case examples have been reviewed in this chapter: an infant born deaf but not accurately diagnosed with ANSD and profound hearing loss until 19 months of age; the other, a premature infant with ANSD and normal hearing sensitivity initially fitted with hearing aids and referred for cochlear implant evaluation, but not accurately diagnosed until 20 months of age. Although the audiological management of infants and toddlers is often challenging even for experienced clinicians, the use of age appropriate, evidence-based test procedures will result in fewer cases of incorrect or late diagnosis of hearing loss. The two cases reported here illustrate the importance of appropriate electrophysiologic measures applied in combination with age appropriate and accurate behavioral audiometry.

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