Chapter 32. Managing infants and children with auditory neuropathy spectrum disorder (ANSD)

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Chapter Outline

Learning objectives

The reader will be able to 1) describe how cortical auditory evoked potentials and assessments of functional auditory behaviour can assist with amplification decision-making and assessment, (2) explain the importance of performing medical imaging in cases of ANSD, and (3) explain why some children with ANSD perform better with cochlear implants than others.

Key Points

1. ANSD is a heterogeneous disorder and has a number of potential sites-of-lesion depending on the underlying cause. Hence, the hearing configuration, speech perception, and functional and speech language outcomes are highly variable.

2. Specific assessment techniques need to be used to correctly diagnose ANSD using auditory brainstem response (ABR) testing.

3. Clinicians need to be aware of undiagnosed and late-onset ANSD

4. Cortical auditory evoked potentials (CAEPs) and assessments of functional auditory behaviour can assist with the decision to fit hearing aids and the evaluation of the effectiveness of amplification.

5. Medical imaging and electrical ABR testing through the cochlear implant can help explain variations in outcomes for children with ANSD using cochlear implants.

6. The early intervention team needs to be multi-disciplinary to take into consideration both the hearing and non-hearing related needs of each child, particularly for those with additional disabilities.

7. Parental involvement and consultation is vital during the assessment and management process, and particular care needs to be taken to minimize parental stress relating to conflicting information and advice.

INTRODUCTION

Overview of auditory neuropathy spectrum disorder

Auditory Neuropathy Spectrum Disorder (ANSD), is now a widely used term to describe a specific hearing disorder characterized by abnormal function of the auditory pathway in the presence of cochlear outer hair function. The audiological characteristics are demonstrated by the presence of evoked otoacoustic emissions (OAEs) and/or cochlear microphonic (CM) with absent or abnormal morphology of auditory brainstem response (ABR) waveforms.[1] Up until recently there has been conflicting information about appropriate management and expected outcomes for children diagnosed with ANSD. Chapter 27 has provided more detailed population-based information about spoken language outcomes in children with ANSD wearing hearing aids or cochlear implants. This chapter will describe the new techniques being used to assist in the early fitting and verification of amplification in infants with ANSD, together with information from medical imaging and electrical ABR testing that help to explain why variations in outcomes may be observed.

Definition and terminology

The condition of ANSD has been referred to in the literature as auditory neuropathy,[1] auditory dys-synchrony,[2] and Type I afferent neuron dysfunction.[3] The reason behind the use of different terminology relates to a number of possible sites of lesion, which are not limited to the auditory nerve and/or neural pathways[4] such as the inner hair cells and synapses.[5] The term ANSD has now been adopted to acknowledge the heterogeneous and multifaceted nature of this disorder.[6]

Prevalence and aetiology

The prevalence of ANSD has been estimated to range from 7% to 10% of all children with permanent hearing loss.[7,8] Within the neonatal intensive care (NICU) population 25% of those diagnosed with a permanent hearing loss have been reported to have ANSD.[9] Medically-related risk factors in the NICU population include prematurity, low birth weight, mechanical ventilation, hyperbilirubinemia, respiratory distress, meningitis and ototoxic medication such as vancomycin.[10]

Approximately 40% of cases of ANSD are believed to have a genetic basis.[11] Genetic mutations associated with congenital ANSD include non-syndromic genes such as Otoferlin[12] and Connexin 26,[13] and syndromic conditions such as some cases of CHARGE[14] and Waardenburg syndrome.[15] Genetic mutations associated with late-onset ANSD include those associated with Friedreich's Ataxia,[16] Leber's Hereditary Optic Neuropathy,[17] Charcot-Marie Tooth disease,[18] riboflavin transporter deficiency,[19] and Mohr Tranebjærg syndrome.[20]

ANSD can also develop during infancy as a result of dietary thiamine deficiency,[21] and temperature-sensitive ANSD can present transiently.[22] For these reasons clinicians need to keep in mind the possibility of ANSD when a child who has passed their newborn hearing screening, or other hearing assessments during infancy and early childhood, later presents with a hearing loss.

Site-of-lesion

Variations in outcomes in patients with ANSD are likely to be related to differences in the underlying pathophysiology. For example, the inner hair cell (IHC) is responsible for the synchronous release of neurotransmitter and triggering of the auditory nerve to fire. A history of hypoxia during the newborn period, or a mutation of the Otoferlin gene can cause the impairment of inner hair cell function. Accurate transfer of temporal information for speech understanding is also dependent on good neural conduction. Late-onset ANSD is frequently associated with neurological conditions that can affect the peripheral dendrites, spiral ganglion cells, axons and/or myelin. These include conditions such as Mohr-Tranebjaerg syndrome,[20] Friedreich's Ataxia,[23] mutations of the OPA1 gene,[24] Brown-Vialetto-Van Laere syndrome[19] and Charcot-Marie Tooth.[25] It is important to note that some of these conditions have associated problems such as vision loss, balance disturbance, and other peripheral neuropathies. Therefore, referral for vision and neurological assessment is particularly important if late-onset ANSD has been identified.

Audiological characteristics

The degree of hearing loss can range anywhere from normal to profound, and the audiometric configuration is variable.[26] Fluctuations in hearing and perceptual abilities have also been

reported in some individuals.[27] Figure 32.1 shows the breakdown of degree of hearing loss

for a population of children with ANSD identified through newborn hearing screening

participating in the Longitudinal Outcomes of Children with Hearing Impairment (LOCHI)

study.[8] At the age of 3 years approximately 50% have pure-tone thresholds in the mild to

moderate range, and 50% in the severe to profound range.

<Figure 32.1 >

Speech perception characteristics

Like behavioral pure tone thresholds, speech perception ability can be varied. Speech

discrimination scores have been reported to be worse than the lowest score expected for the

behavioral audiogram in almost 50% of children with ANSD.[28] The most consistent

abnormalities found in subjects with ANSD have been in the processing of timing

information such as pitch discrimination at low frequencies, gap detection, temporal

integration, signal detection in noise and sound localization using interaural time

differences.[29] The severity of the temporal processing deficits is correlated to speech

perception performance with better temporal resolution relating to better performance for

speech discrimination in quiet. In some cases speech discrimination ability may be similar to

children with SNHL in quiet listening conditions but worse in the presence of background

noise.[30]

Functional auditory behaviour and language outcomes

The majority of studies to date have focused on sound detection and speech perception, with

very limited information on functional ability.[31] Parent questionnaires such as Parent's

Evaluation of Aural/Oral Performance of Children (PEACH) questionnaire, LittlEARS

auditory questionnaire and the Infant-Toddler: Meaningful Auditory Integration Scale (IT-

MAIS) provide a valuable way of assessing infants' auditory behaviour, monitoring progress over time, and engaging parents in the observation process.

More recently Ching et al (2013) reported that there were no significant group difference in the speech and language outcomes of a population-based sample of 3 year old children with ANSD wearing hearing aids compared to those with a SNHL, after controlling for the degree of hearing loss and other factors known to affect speech and language outcomes.[8] Systematic reviews on children with ANSD following cochlear implant generally show an improvement in sound detection, speech discrimination and speech recognition and appeared similar to children with SNHL.[32,33]

DIAGNOSING ANSD

Clinical test battery

Diagnosing ANSD is accomplished on the basis of a combination of electrophysiological assessments. The key features identified include evidence of surviving outer hair cell function due to the presence of otoacoustic emission (OAEs) and/or a cochlear microphonic (CM) on auditory brainstem response (ABR) testing, together with absent or abnormal ABR waveforms. The absence of OAEs therefore does not preclude a diagnosis of ANSD, as OAEs have been reported to be absent in around 50% of children with ANSD.[27] Tympanometry or high frequency tympanometry (for infants ≤ 6 months) should also be included to rule out any middle ear involvement as the OAEs and CMs can be diminished by a conductive component. Auditory steady state response (ASSR) results can be misleading in cases of ANSD with reports of both significant over and underestimation of hearing

thresholds.[27] For this reason ASSR testing should not be used in isolation when assessing infants and children with ANSD.

<Box 32.1>

Table 32.1 describes the key components to ABR testing, and the potential pitfalls if these steps are not taken. The presence of a CM is determined by performing clicked-evoked auditory brainstem response testing using a high intensity click stimulus ($\geq$ 70 dB HL). The CM follows the polarity of the stimulus, and therefore reverses in polarity when changing from a condensation to rarefaction click stimulus (see Figure 32.2). Insert earphones rather than supra-aural headphones must be used. Also, a control run with clamped insert earphone tubing (blocked stimulus) should be conducted to eliminate the possibility of stimulus or transducer artefacts.[34] ANSD can be diagnosed if the ABR assessment conducted in this way shows an absent or abnormal morphology of ABR waveforms following the presence of CM with normal middle ear function.

<Table 32.1 >

<Figure 32.2 >

<Figure 32.3 >

<Box 32.2>

Middle-ear muscle reflexes (MEMRs) are elevated or absent in the majority of children with ANSD.[35] Normative data on MEMR thresholds for infants $\leq$ 6 months using a high-frequency probe-tone is now available.[36] However, the application of MEMR in the clinical test battery of infants with ANSD remains to be investigated.

Recovery

Another challenge clinician's face when managing infants with ANSD is the possibility that the ANSD may recover or improve over time. Improvements in the ABR can be seen over the first few months of life in premature infants where myelinisation may not have reached an equivalent level to that of a full-term baby.[37] Improvements in behavioral thresholds have also been reported in some infants with a history of hyperbilirubinemia.[38] These highlight the importance of repeating ABR and behavioral threshold assessments over time to monitor for any changes. It is important to differentiate between improvements in hearing thresholds together with normalization of the ABR, versus an improvement in hearing thresholds with persisting abnormality of the ABR. An improvement in auditory thresholds does not necessarily translate to an improvement in functional auditory performance if the ABR is still abnormal.

Late diagnosis

Given the onset of ANSD can occur later in childhood, and newborns with ANSD screened using OAEs can pass their screen, clinicians need to be alert to the possibility of undiagnosed ANSD in the paediatric population. Concerns are raised if children demonstrate significantly poorer speech discrimination than expected given their pure tone audiogram, and/or inconsistent auditory behaviour that is not explained by intermittent middle-ear pathology. If OAEs are detected in the presence of a significant pure tone loss, and/or MEMRs are absent for degrees of loss where they are expected to be present, referral for ABR testing should be initiated to check for ANSD.

<div align="center">**<Box 32.3>**</div>

POST-DIAGNOSTIC MANAGEMENT

It has been generally accepted that children diagnosed with ANSD should be fitted with appropriate hearing aids when behavioral thresholds show a significant hearing loss,[6] and cochlear implant when poor progress in speech and language development is found in spite of appropriate amplification.[7] When infants with ANSD are considered it is more controversial to fit hearing aids before behavioral thresholds can be obtained, for fear of over amplification causing damage to surviving cochlear structures.

Unfortunately infants are not typically able to perform visual reinforcement audiometry (VRA) until approximately 8 months developmental age,[39] and the abnormal or absent brainstem activity to sound means that ABR and ASSR testing cannot be used to estimate hearing thresholds in infants with ANSD in the same way that they can for infants with a SNHL. This delay in obtaining behavioral thresholds means that an infant identified from newborn hearing screening may not be fitted with amplification until the child is one year old. The delay in treatment can be even longer for infants born prematurely or with additional disabilities.

Current evidence from the LOCHI study indicates that positive advantages for language development are associated with early intervention for both children with SNHL and ANSD (see chapter 27). For this reason, cortical auditory evoked potentials (CAEPs) is being used as an alternate measurement of auditory function, which together with the Parent's Evaluation of Aural/oral performance of CHildren (PEACH) questionnaire, can help guide the management of infants with ANSD over the first year of life.[40]

Objective and functional approach

Cortical auditory evoked potentials (CAEPs) are a series of waves recorded on the scalp generated by the auditory cortex. In infants, the CAEP is dominated by a positive-polarity peak with a latency of around 200 ms after stimulus onset, and can be recorded from infants within the first few months of life.[41] The peak amplitude of P1 is relatively large in infants, and extends into hundreds of milliseconds compared to the small peaks occurring every 1-2 ms for the ABR. This means that whilst small disruptions in neural synchrony can result in abnormalities of the ABR, they have less impact on the CAEP. Whereas ABR may be absent in infants with ANSD, CAEPs are often detectable. This makes it particularly relevant to use when assessing individuals with ANSD.

Accurately estimating hearing thresholds using CAEPs in babies has been challenging to achieve, which is likely to be due to the high myogenic noise levels relative to the small amplitude CAEP at threshold. Nevertheless, testing at intensities of 55-75 dB SPL in the free field can help to narrow down the range of the degree of hearing loss, which can assist in the decision of whether or not to fit amplification.

Research has shown a correlation between stimulus sensation level and CAEP response detection in infants.[42] When unaided CAEPs are present at 55 dB SPL in the free field, it is likely the behavioral hearing thresholds lie within the normal to mild hearing loss range at the frequency band tested. Similarly, when CAEPs are present at 65 dB SPL but absent at 55 dB SPL; present at 75 dB SPL but absent at 65dBSPL; or absent at 75 dB SPL, the hearing

thresholds are likely to be in the mild to moderate, moderate to moderately severe, and severe to profound range respectively. It is important to keep in mind that up to 32% of infants show no CAEP even when the sound is audible to them.[43] For this reason an absent CAEP should be interpreted together with measures of functional auditory behaviour and behavioral response audiometry (BOA) to establish whether there is consistency between behavioral and objective measures.

A number of studies have used the speech sounds /m/, /g/ and /t/, which have spectral emphasis at around 250, 1500 and 3000 Hz respectively, as speech stimuli for CAEP testing. Using a combination of these sounds presented at different intensities, it is possible to establish the general degree and configuration of the hearing loss. If unaided CAEPs are absent at 55 dB HL, and baseline PEACH scores are not age-appropriate, aiding should be considered in consultation with the family and early intervention therapist working with the child.

<Box 32.4>

Tests of functional auditory behaviour such as the PEACH questionnaire have a number of benefits. Firstly, there is validated normative data that can be used as a reference point for functional auditory development.[44,45] Secondly, the PEACH can be repeated over time to ensure the infant is progressing as expected or showing signs of plateauing or deterioration. Thirdly, it engages parents in the observation and assessment process. A positive correlation has been shown between aided CAEP results and PEACH score in infants with SNHL and ANSD whereby infants with CAEPs present to a wider range of stimuli showing higher PEACH scores.[46,47] In a similar vein, abnormal CAEP latencies and morphology have been linked to lower IT-MAIS scores in infants, and lower auditory skills and speech

discrimination scores in children.[48,49] The relationship between cortical detection and perception demonstrates the value of combining CAEP testing and formal measures of functional auditory behaviour to complement the audiological assessment battery in these cases.

Hearing aids

Once a decision to aid is reached, the initial estimated audiogram based on CAEPs and/or BOA results can be used to derive prescriptive targets according to a prescriptive procedure for children, such as the NAL or the DSL procedure. Real-ear measurements are required to verify that the prescriptive targets are met in hearing devices. After verification, the effectiveness of the amplification in providing audibility should be evaluated using aided CAEP and PEACH assessments, both in infants with SNHL[43,50] and ANSD.[42] CAEPs typically show a monotonic pattern to speech stimuli whereby an increase in the stimulus sensation level results in an increase in the detection rate of the CAEP. The same monotonic pattern has been seen in some[42] but not all[46] infants with ANSD.

Evaluation Procedure

CAEP testing for infants is typically carried out in an acoustically treated room. Speech stimuli can be presented from a free-field loudspeaker at a calibrated level. An infant is positioned in a high-chair or a parent's lap at 0° azimuth, at a distance of about one metre from the loudspeaker. Three electrodes are placed on the scalp: a recording electrode on the vertex of the head, a reference electrode on the mastoid, and a ground electrode on the forehead. In aided testing, the infant wears his/her hearing aids that have been verified to match prescriptive targets.

Figure 32.4 shows the possible scenarios that can occur. The first scenario shows absent unaided but present aided CAEP responses. This demonstrates aided benefit in the frequency band tested and the results can be used to assure the parents that their child is 'detecting' the sound at conversational level. Whilst this is not a test of thresholds or speech discrimination, parents can find this early management very reassuring. As the child grows it will then be possible to perform VRA to assist in fine-tuning of the hearing aid gain settings, and eventually formal measures of speech discrimination ability, to monitor development.

<Figure 32.4 >

The second scenario shows that no response is seen at 65 dB SPL in both the unaided and aided conditions. This can mean one of four things. First, the infant is under-fitted so the clinician can consider re-estimating the hearing levels and adjusting the hearing aids accordingly before repeating the test. Secondly, the infant has a severe-profound hearing loss, in which case amplification is only likely to provide limited access to the speech range in the same way as it would for a case with SNHL. Thirdly, the infant may be one of those with an absent CAEP even when the sound is audible. Finally, based on previous studies on children with ANSD showing an association between absence of CAEPs and poor speech discrimination ability,[49] it may be an indication that the infant has poor speech discrimination ability even though the sound is audible.

<Box 32.5>

Case study

Case AN1 presented in chapter 27 shows how CAEPs and the PEACH diary were used to assist in the decision to refer for cochlear implant candidacy. The infant referred bilaterally

on their newborn hearing screen, and was diagnosed with ANSD. At 2 months corrected age, CAEPs were measured in the unaided condition to estimate hearing sensitivity and the need for amplification. Results revealed no detectable responses to speech sound stimuli /t/ /m/ /g/ at 75 dB SPL. Hearing aids were then fitted to a severe degree of hearing loss in both ears. Aided CAEPs and PEACH were used to evaluate the amplification, and the hearing aids were then adjusted accordingly. Subsequent evaluation results showed limited progress in the child's functional and language development. The family was advised to consider referral for cochlear implant candidacy evaluations.

In addition to assessing speech detection, the child's progress with the development of speech perception, production and language should be monitored regularly to determine whether the progress is adequate and age appropriate or whether an alternate mode of communication or alternate technology such as cochlear implant are indicated. As many children with ANSD experience difficulties when listening in noisy situations, such as the classroom, clinicians should also consider frequency modulation (FM) or remote microphone (RM) systems. These can be used in isolation (e.g. in the case of a child with normal hearing sensitivity), or in addition to hearing aids or cochlear implants.

Cochlear Implant

Studies investigating outcomes with cochlear implants in subjects with ANSD have on the whole been positive[51,52] although not all have had successful outcomes.[53] The reasons why outcomes have varied can be explained by differences in the underlying pathology. Individuals with peripheral sites-of-lesion (IHC, ribbon synapse and terminal dendrites) are

likely to have more successful outcomes than those with pathology involving spiral ganglion cells, axons or hypoplasia of the auditory nerve.[54]

Implant evoked electrical ABR

Implant evoked Electrical Auditory Brainstem Response (ImpEABR) testing has helped to provide objective evidence of pre vs. post-synaptic sites-of-lesions in children with ANSD, and to predict children who are more likely to progress well with their cochlear implant. The testing technique is similar to standard ABR testing except that electrical pulses are delivered from the implant to elicit the response. Figure 32.5 shows examples of ImpEABR results for three different children with ANSD. The first set of results is from a child who suffered significant hypoxia at birth and shows present wave V responses on all electrodes. The second set of traces on Figure 32.5 is from a child with late-onset ANSD due to riboflavin transporter deficiency. The morphology and latency of the response is inconsistent across the array, and is in keeping with the neural pathology underlying this condition. The third set of results is from a child with hypoplastic auditory nerves. In this case responses are only seen on two electrodes at the basal end of the cochlea suggesting a small patch of auditory nerve fibres are present and capable of responding to electrical stimuli. Recent research showed that children who demonstrated clear ImpEABR waveforms demonstrated speech perception, receptive and expressive language scores that were similar to implanted SNHL children.[55]

<Figure 32.5 >

Medical imaging

Radiological imaging of the temporal bones prior to cochlear implantation is important in determining whether there are any structural abnormalities that may impact on the insertion of the CI array, and to identify whether there is a reduction in the size of the auditory nerve

that may prevent a complete signal from the cochlear implant being transferred along the auditory brainstem pathways. Magnetic resonance imaging (MRI) using a cross-sectional/parasagittal view of the internal auditory meatus is recommended. This allows the radiologist to compare the diameter of the auditory nerve in relation to the facial nerve and two branches of the vestibular nerve. Figure 32.6 shows the parasagittal view on MRI for a child with a full complement of nerves in the bundle (left); a child with only two nerves (centre); and a child with only one nerve (right). Birman et al. (2016) analysed the results of 50 children with varying degrees of abnormality of the auditory nerve and reported higher scores on tests of auditory perception for those with small but identifiable nerves compared to children where the nerve was either absent or fused with other nerves in the bundle.[53] Other researchers have reported a similar pattern of results whereby the greater the degree of the abnormality the more limited the outcomes with a cochlear implant.[56] This information is useful when counselling families about realistic expectations following cochlear implantation, and the likely need to integrate visual forms of communication such as sign language into the child's early intervention program.

<Figure 32.6 >

<Box 32.6>

Mild-moderate hearing loss

It has been reported that some children with ANSD who have a mild to moderate pure-tone loss demonstrated speech discrimination ability below that expected for their degree of hearing loss, even when wearing hearing aids that were optimally fitted.[49] These children could potentially benefit from cochlear implantation. It is important for clinicians to include measures of speech discrimination ability when establishing an audiological management

plan, and not exclude the option of cochlear implantation based on the pure-tone thresholds alone if discrimination is significantly affected.

Family centred multi-disciplinary assessment and rehabilitation

Family-centred early intervention (FCEI) enhances parental engagement in the management and monitoring process, and can help to optimise outcomes for children with hearing loss. Clinicians are referred to the consensus statement describing the ten principles of FCEI published by Moeller et al. (2013) which describes the validated, evidence-base behind these principles, and clinician behaviours that support them.[57]

As mentioned previously, the majority of infants diagnosed with ANSD have experienced a very stormy start to life. As a result parents can feel overwhelmed with perinatal health issues, even before the diagnosis of ANSD is made.[58] Parents have identified additional stressors associated with the diagnosis of ANSD including conflicting information from the professionals working with them, and uncertainty around the prognosis for their child.[59] Parents will inevitably search on social media for further information, which can further exacerbate the situation if they are not able to critically evaluate the information presented. It is particularly important for clinicians to spend time with the family to gain an understanding of their current level of knowledge, and establish the parent's goals and preferences for their child. In addition, families need to be provided with comprehensive information on all the options available, including any advantages, disadvantages and uncertainties around each one.

<div align="center"><Box 32.7></div>

Ching et al. (2013) reported that 30% of the ANSD population in their study were found to have at least one other disability in addition to hearing loss, which included developmental delay, physical disabilities such as cerebral palsy, and autism.[8] For this reason it is not unusual for a child to need to access input from a number of other professionals including physiotherapy, occupational therapy and ear, nose and throat specialists. Audiologists need to work closely with speech and language therapists in particular to obtain formal information about each child's progress with speech and language development.

CONCLUSION

The prevalence of ANSD is estimated to be around 10% of all children diagnosed with permanent hearing loss. Appropriate test procedures need to be used for the accurate diagnosis of ANSD using ABR, and repeat testing performed to check for improvement in ABR morphology over time. Clinicians should refer older children for ABR testing if they demonstrated poorer than expected speech discrimination ability, or presented with OAEs despite significant hearing loss, so that cases of late-onset ANSD can be identified.

The majority of children with ANSD have spent time in the NICU and have medically-related risk factors, however genetic causes have been identified in approximately 40% of cases. The different underlying causes, and associated sites-of-lesion, are likely to explain the variations seen in pure-tone sensitivity, speech discrimination ability, and outcomes following amplification or cochlear implantation.

The use of CAEPs together with functional auditory assessments such as the PEACH enable us to reach decisions about the need to provide amplification, and to proceed with the early

fitting of hearing aids in infants with ANSD. In addition, these methods enable us to evaluate the effectiveness of amplification, and identify infants who would benefit from a signing mode of communication and/or referral for cochlear implant candidacy evaluations. Where available, speech discrimination ability with amplification should also be considered. In the work up for cochlear implantation, an MRI scan using a para-sagittal view should be arranged to check for hypoplasia of the auditory nerve. By using a combination of objective testing and parent ratings of auditory behaviour in real life, clinicians are now in a better position than before to provide early intervention to infants diagnosed with ANSD.

CHAPTER SUMMARY

With the implementation of newborn hearing screening, infants with ANSD are being identified within the first months of life. Current evidence shows that the provision of early amplification and/or cochlear implants is associated with better speech language outcomes in infants with hearing loss. However, clinicians are unable to use ABR testing to estimate the audiogram for infants with ANSD so determining whether amplification is required, and how much amplification to apply, has been problematic.

Over recent years new testing methods have become available to assist clinicians in the early management of infants with ANSD. This chapter describes how the combination of CAEPs, and measures of functional auditory behaviour such as the PEACH, can provide valuable information to help guide the management of these infants shortly after diagnosis. This includes their use in the decision of whether to fit amplification; assessing the effectiveness of amplification; discussions around the need for signed communication; and considerations for referral for cochlear implant candidacy evaluation. These tools, together with results

from medical imaging and input from a multi-disciplinary team, place clinicians in a better position to advise families on the most appropriate way forward for each individual child.

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Chapter review questions

1. What are some of the medical risk factors associated with ANSD?

2. What are some of the genetic mutations associated with late-onset ANSD?

3. What pattern of results are you likely to see on CAEP and PEACH assessments for an infant who has inadequate amplification?

4. What are some of the reasons why some children with ANSD perform better with a cochlear implant than others?

5. Which multi-disciplinary team members are likely to need to be involved for a child with ANSD and cerebral palsy?

6. Where do parents potentially source information on ANSD that can provide conflicting information and management recommendations?

Text boxes:

Box 32.1 ABR and ASSR testing do not provide accurate estimates of auditory thresholds in infants with ANSD.

Box 32.2. Performing ABR testing using separate polarity 'click' stimuli via insert earphones is important in correctly diagnosing ANSD.

Box 32.3 Clinicians need to be aware of the possibility for children to develop late-onset ANSD, and children with ANSD who passed their newborn hearing screen using OAE's. An ABR should be arranged if auditory performance is poorer than expected given the degree of the hearing loss.

Box 32.4 CAEPs and tests of functional auditory behaviour can be used to assist in the early fitting of amplification before VRA results can be obtained.

Box 32.5 Absent CAEPs do not necessarily mean the infant cannot hear the sound.

Box 32.6 MRI's need to be performed with a para-sagittal view to check for hypoplasia of the auditory nerve in ANSD.

Box 32.7 Clinicians need to provide parents with good quality, evidence-based information to reduce the parental stress associated with the provision of conflicting messages.

Figure legends:

Figure 32.1 shows the better ear 4FA results for 3 year old children with ANSD involved in the LOCHI study. Half of the children have pure tone thresholds in the mild to moderate range, and half in the severe to profound range.

Figure 32.2 shows ABR waveforms for a child with a) Normal hearing: The top traces show the condensation, rarefaction and alternating ABR waveforms for a child with normal hearing. A CM is seen prior to wave I when the condensation and rarefaction traces are superimposed, and ABR waveforms are present. B) SNHL: The middle traces show ABRs to different polarity stimuli for a child with a moderate SNHL. No CM is seen but reproducible ABR waveforms are present. C) ANSD: The bottom traces show ABRs for a child with ANSD. A CM is seen when the response to condensation and rarefaction stimuli are superimposed, however no ABR waveforms are present.

Figure 32.3 shows a recording to a single-polarity 'click' stimulus (top - "Rarefaction trace a). The response appears similar to a typical ABR waveform, however when the same response is superimposed with a condensation 'click' recording (middle traces) the responses are a mirror image of one another. Had only one polarity recording been used the waveforms may have been misinterpreted as a present ABR rather than a CM.

Figure 32.4 shows the potential scenarios that can occur during CAEP testing. Scenario 1 shows absent unaided CAEPs at conversational levels, which are present when aided – demonstrating aided benefit. Scenario 2 shows absent unaided and aided responses including the four possible explanations for this result.

Figure 32.5 shows the ImpEABR responses elicited from individual electrodes for a 22 channel Cochlear® Nucleus® device. The responses to electrodes positioned towards the

apical end of the cochlea are at the top starting with electrode 22 (e.22) and the basal

electrodes at the bottom (e.1). The set of traces on the left are for a child who suffered

significant hypoxia who is likely to have ANSD due to damage of the IHC's; in the centre for

a child with a neurological condition associated with riboflavin transporter deficiency; and on

the right a child with a hypoplastic auditory nerve on MRI.

Figure 32.6 shows a para-sagittal MRI views of the nerve bundle within the internal auditory

meatus. On the left all four nerves are present in the bundle including the cochlear, facial,

and two branches of the vestibular nerve. In the centre only two nerves are present, and on

the right only one nerve.

Tables:

Table 32.1 describes the key ABR test procedures for diagnosing ANSD and potential

pitfalls if these are not followed.

Table 32.1 Key ABR procedures required for the correct diagnosis of ANSD

	Description & diagnostic risk	ABR test procedure for correct interpretation
1. Stimulus artefact and response interpretation	The electrical stimulus artefact generated from the earphone transducer changes in polarity with the stimulus and can be mistaken for the CM.	• Use insert earphones rather than supra-aural headphones to separate the stimulus artefact from the CM. The stimulus artefact occurs immediately following the onset of the stimulus, whereas the CM (if present) occurs approximately 0.8 ms later when insert earphones are used (see Figure 32.2) • Maintain the transducer of the insert earphone away from the electrode sites to reduce electrical artefact interference. • Perform an additional recording with the insert tubing clamped to prevent sound reaching the ear. The stimulus artefact will remain but the CM (if present) will disappear.
2. Alternating 'click' and missed ANSD	When an alternating 'click' stimulus is used the CM is cancelled out and the diagnosis of ANSD can be missed (see Figure32.2).	Do not use an alternating 'click' stimulus alone unless your recording system is capable of separating rarefaction and condensation 'clicks' into separate buffers for viewing and interpretation.

3. Single polarity 'click' and incorrect ABR interpretation	If only one polarity 'click' is used the CM can be mistakenly interpreted as an ABR (see Figure 32.3) and ANSD can be missed.	Perform both rarefaction and condensation 'clicks' in separate traces. If the response to one polarity is a mirror image of the response to the opposite polarity it is a CM rather than an ABR waveform (see Figure 32.2 & 3).