Challenges in Audiologic Diagnosis
Illustrative Case Examples

Sound for a Young Generation Conference

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Pediatric Audiology and CI Teams
CASTLE pre-school (SLPs, teachers of the Deaf, AVTs)
Total 1800 infants and children
  » 1000 using amplification
  » 600 with cochlear implants
  » 200+ with ANSD diagnosis
Where is North Carolina?
Early Hearing Detection and Intervention (EHDI) in North Carolina

- Passed legislation in 1999
- Started screening in 2000
- 130,000 births per year
- Screening approximately 98% in 89 hospitals
Newborn Hearing Screening…
The First of Many Steps
Audiologic Management of Infants and Young Children: Essential Components

- **Diagnostic Evaluation**
  - Auditory Brainstem Response (ABR)
  - Acoustic Immittance
  - Otoacoustic Emissions

- **Hearing Aid Selection and Fitting**
  - Appropriate selection of device (size, features)
  - Hearing aid programming
  - Hearing aid verification
  - Hearing aid validation

- **Behavioral Audiometry**
  - Visual reinforcement audiometry (VRA)
  - Conditioned play audiometry (CPA)
Assessment: Electrophysiologic Measures

- **ABR**
  - Tone burst stimuli used to estimate thresholds for low, mid and high frequencies
  - When ABR shows no response, must use single polarity clicks to rule out auditory neuropathy
  - Air conduction and bone conduction

- **Otoacoustic Emissions**

- **Acoustic immittance measures**
  - (1000Hz probe tone <4 months)
Audiogram estimated based on electrophysiologic tests
Otologic Evaluation

- Electrocardiogram (Jervell and Lang-Neilson)
- Imaging of the ear
  - Malformations
  - Labyrinthine Ossification
  - 8th nerve aplasia
  - Tumors
  - Associated Brain problems
- Lab Studies as needed
  - VDRL, Thyroid function, lipid profile, ESR
  - Renal ultrasound
- Eye examination/Electro-retinography (Usher’s)
- Genetic studies
  - GJB2 and GJB6 testing +/- others as indicated
  - Able to obtain genetic and CMV tests from newborn blood spot stored in state database
- Other Medical Referrals
Hearing Aid Fitting in Infants

- Prescriptive formula selected
  - e.g. DSL, NAL-NL1
- Program Hearing instrument
  - Manufacturer’s software used
- Verification of Fitting
  - An alternative procedure to traditional probe microphone measures for use with infants and children is Real Ear to Coupler Difference measurement (RECD)
Goal: Audible Speech Signal for Average Speech Inputs...
Behavioral Audiologic Assessment

• Begin VRA at 6-7 months

• Goal: Complete audiogram for each ear (air and bone) by 8-9 months of age.

• Hearing aids readjusted as new threshold information is obtained
Referral for Early Intervention

- Referral to “Beginnings” on day hearing loss diagnosed (www.ncbegin.org)
- Family contacted within one week of diagnosis and home visit from early childhood specialist scheduled
- Weekly home visits with teacher of the HI scheduled as soon as family decides on initial educational approach
Evaluation of Speech Perception

- Parent Questionnaires (e.g. PEACH, IT-MAIS or MAIS)
- Early Speech Perception Test battery (ESP)
  (Moog and Geers, 1990)
- Low Verbal
- Standard
- MLNT/LNT words and phonemes
  (Kirk, et al, 1995)
- PB-K words and phonemes (Haskins, 1949)
- HINT sentences in quiet and noise conditions
Sounds Easy But…

Challenges Remain
Case #1
Introduction

- Born at full term without complications
- Newborn hearing screening status unknown
- View the following slides and try to predict child’s audiogram
First ABR in Natural Sleep

Left Ear

Right Ear
Second ABR
Under General Anesthesia Following MRI

Left Ear
- Clicks
- Alternating polarity
- Sound tube interrupted

Right Ear
- Clicks
- Alternating polarity
- Sound tube interrupted

250 Hz

- A6 Li90nHL
- A7 Li80nHL
- A8 Li80nHL

- B8 Ri90nHL
- B9 Ri80nHL
- B10 Ri80nHL
- B11 Ri70nHL
Otoacoustic Emissions
Present Bilaterally
Audiogram at Age 10 years

- **Speech Recognition Testing:**
  - Left Ear: 96%
  - Right Ear: 12%

- **Tympanometry:**
  - Right: Normal
  - Left: Normal

- **Acoustic Reflexes:**
  - Right: Absent
  - Left: Absent

- **DPOAEs:**
  - Right: Present
  - Left: Present
Radiologist’s Report of MRI using N.VIII Protocol:

- The right cochlear nerve is not visualized;
- The left cochlear nerve appears significantly atrophied versus possibly aplastic

**Impressions:**
- Findings concerning for right cochlear nerve aplasia and left cochlear nerve aplasia versus hypoplasia
Additional Information

- Child was 10 years old at time of referral to our program
- Referred from school audiologist who questioned possibility of ANSD after testing showed present OAEs and absent acoustic reflexes
- Following audiologic and otologic evaluations child referred to pediatric neurologist
- Important to consider what management recommendations might have been made in infancy with only ABR test findings and imaging available
Key Points

- A test battery approach is needed for accurate audiologic diagnosis.
  - No single test available provides all of the diagnostic information necessary to make management decisions

- ABR useful in estimating behavioral thresholds but…
  - ABR is not a test of hearing
  - Confirmation with behavioral audiometry remains essential

- Radiologic imaging provides useful information in search for etiology of hearing loss but results obtained don’t always tell the whole story

- Otoacoustic emissions useful indicator of outer hair cell function but should be used as a component in a test battery not in isolation
CASE #2
Background:

- **Newborn Screen with AABR:**
  - Referred on left
  - Passed on right

- **Age 2 months:**
  - Diagnostic ABR: moderate HL left, normal right

- **Age 2 ½ months:**
  - Otologic evaluation: MRI, EKG, connexin 26 and CMV testing ordered
Background:

- **Age 3 months:**
  - Referred to Beginnings for information and referral to early intervention
- **Age 4 months:**
  - MRI: Bilateral enlarged vestibular aqueducts and enlargement of endolymphatic sacs (EVAS or LVAS)
  - Otologist advises of risk for progressive hearing loss and avoiding head trauma and refers to neurology and genetics for evaluation
Background:

- **6 months: Genetics consult completed**
  - Most common cause of EVAS is alteration of Pendred gene
  - Several other syndromes can be associated with EVA including branchio-oto-renal syndrome
  - Will test for Pendred’s and if negative will order renal ultrasound
  - Lab results shows child is connexin 26 negative but has two copies of gene for Pendred’s
  - Recommendation made for pediatrician to periodically monitor thyroid levels
Age: 8 months

- **Tympanometry**
  - Right: normal
  - Left: Negative middle ear pressure (-275)

- **Otoacoustic Emissions**
  - Right: Absent above 2000Hz
  - Left: Absent
Age: 12 months

- **Right ear:**
  - Normal
- **Left ear:**
  - Mild to moderate
- **Tympanometry**
  - Right: normal
  - Left: normal
Age: 17 months

- Difficult to test but right ear responses poorer than expected
- Tympanometry
  - Right: normal
  - Left: normal
- Family advised of our concern re progression of HL
Age: 18 months

- Child will not tolerate insert earphones
- Unable to rule out hearing loss for "better ear"
- Tympanometry
  - Right: Negative middle ear pressure (-225)
  - Left: Negative middle ear pressure (-190)
- Repeat ABR with sedation recommended
Age 20 months: Estimated Thresholds (eHL) Based on Sedated Tone Burst ABR

- Binaural hearing aids and personal FM dispensed 2 weeks later
Age: 22 months

- Continued progression of hearing loss noted
- Tympanometry
  - Right: normal
  - Left: normal
Age: 23 months

- Play audiometry
- Hearing aids exchanged for model with more power
- Hearing aids programmed for best match to DSL targets
Age: 24 months

- Hearing aids readjusted to better match DSL targets
- Recently fitted with new hearing aids with frequency compression
- Speech and language evaluation scheduled with SLP from CI team to obtain baseline and review current services
- Child will be monitored regularly and referred for CI evaluation if indicated
Age: 4 years

- **Aided Testing**
  - SRT=25dBHL
- **Aided PBK score:**
  - 80% at 55dBHL
Age: 4 years, 11 months

- Limited HA benefit even with HA with FC
- Aided speech recognition:
  - 36% at 55dBHL (PBKs)
- Struggling in pre-school
- After extensive discussion with family, referred to CI team for evaluation
- Note air/bone gap:
  - Tympanometry normal bilaterally
Age 7 years: HA right, CI left

Tympanometry:
Normal bilaterally

Speech Perception Testing:
SRT:
- HA right: 25dBHL
- CI left: 25dBHL
- HA&CI: 15dBHL

Recorded monosyllabic words (PBKs):
- HA right: 40%
- CI left: 76%
- HA&CI: 94%
Large Vestibular Aqueduct Syndrome (LVAS) or (EVAS)

- 5-15 % of children with permanent HL have EVAS
- Vestibular aqueduct considered enlarged if >1.5 mm
- Most well known cause is mutations in the SLC26A4 formerly known as PDS gene
- May present with conductive or mixed HL
Key Points

• Comprehensive team evaluation useful when working with infant with newly diagnosed HL
  » Audiology, ENT, Genetics, Early Intervention Specialists, Pediatrics all played role
• ABR used to determine initial thresholds for first hearing aid fitting and to help when results are ambiguous but…
• Behavioral audiometry with VRA to obtain accurate unaided thresholds most useful tool after six months of age in this case
• Evaluation of unaided hearing thresholds combined with use of hearing aid verification measures allowed child to continue to make progress even with progressive changes to hearing
CASE #3
Background

- Born at full term without complications
- Newborn Screen with AABR:
  - Failed bilaterally
- Age 5 months:
  - Diagnostic ABR following tube placement:
    - Borderline normal to mild HL right
    - Mild to moderate HL left
  - Otologic evaluation:
    - Connexin 26: Negative
    - MRI consistent with Large Vestibular Aqueduct Syndrome (LVAS)
Plan

• Results discussed with family
• Referral made for early intervention services
• Parents chose not to proceed with amplification for left ear
• Recommended return appointments to obtain ear and frequency specific measures for each ear
Age 11 months

- **Tympanometry:**
  - Flat with large physical volumes bilaterally

Right Ear

Left Ear

11/23/2012
Age 14 Months

- **Tympanometry:**
  - Right: Flat with normal ear canal volume
  - Left: Flat with large ear canal volume

**Right Ear**

**Left Ear**
Age 22 Months

- Child making good progress with speech and language
- Mother offered but declined EI services because she feels child is doing very well
- Tympanometry:
  - Right: Flat with large ear canal volume
  - Left: Type A

**Right Ear**

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**Left Ear**

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**Tympanometry Notes:**

- Probe Tone (Hz): 226
- Ear Canal Volume: 2.50
- Peak Admittance (ml): 1.59
- Peak pressure (daPa): -350

11/23/2012
Age 29 Months

- Child more difficult to test
- Family feels child hears fine
- Tympanometry
  - Right: Flat with large ear canal volume
  - Type A left
- Repeat ABR recommended
ABR Test Results

- Tone burst ABR results consistent with bilateral hearing loss
- Family agrees to proceed with amplification
- Child fitted with binaural hearing aids
Age 3 years, 2 months

- **Tympanometry:**
  - Flat with large physical volumes bilaterally
Age 3 years, 5 months

- Masked bone conduction testing and speech perception testing completed
- Score: 24/24 on ESP monosyllable test (closed set test)
Age 4 years

- **Tympanometry:**
  - Right: Flat with large ear canal volume
  - Left: Negative pressure
Key Points

• In this case, conductive hearing loss (CHL) due to middle ear fluid added additional confusion to already difficult diagnosis.

• CHL in presence of normal tympanometry or continued presence of CHL following tube placement should raise suspicion for LVAS or other “inner ear conductive HL”
Key Points

- Inner ear conductive hearing loss is common finding in individuals with EVAS as well as other conditions:
  - superior, posterior and lateral canal dehiscence
  - X-linked stapes gusher

- Imaging studies such as MRI and CT are often helpful in determining etiology of childhood HL

- When imaging studies are not available, the presence of air/bone gap with normal tympanometry or open tubes may alert audiologist to possibility of EVAS or other inner ear malformations
Gracias!

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