



# 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

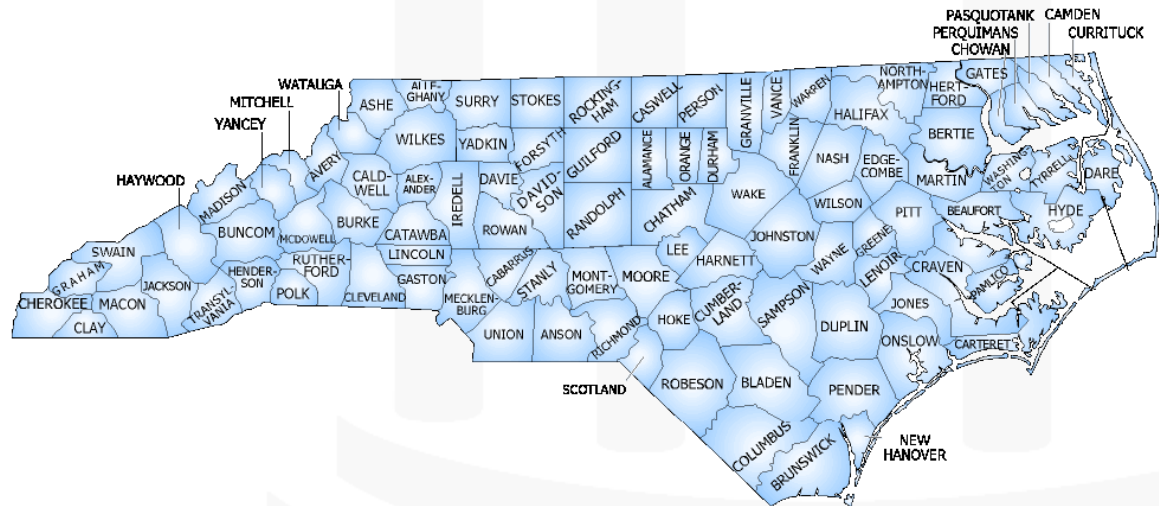


# Where is North Carolina?



# 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)

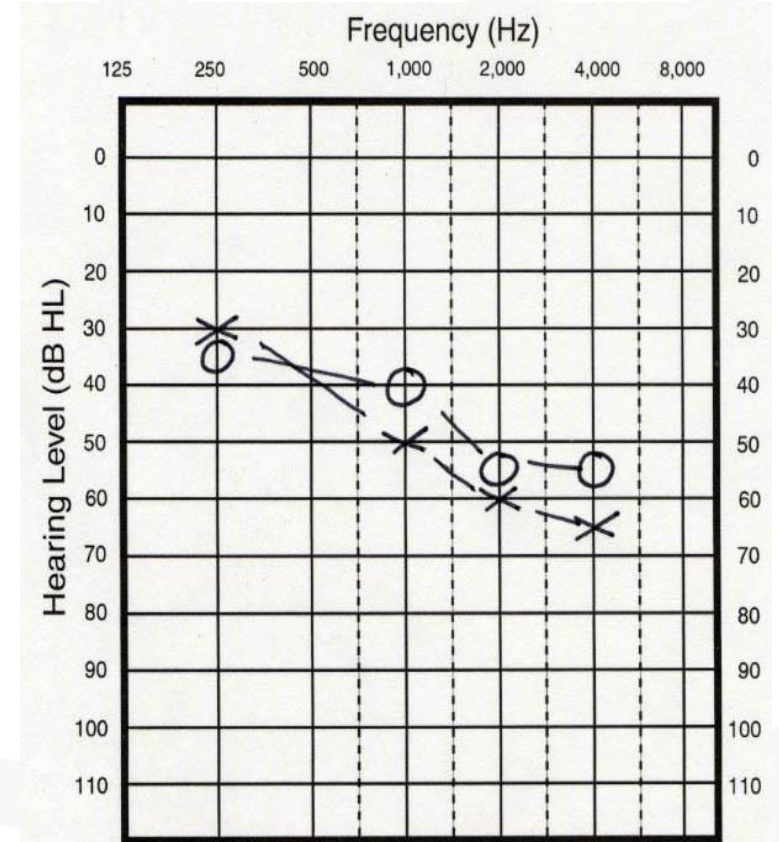
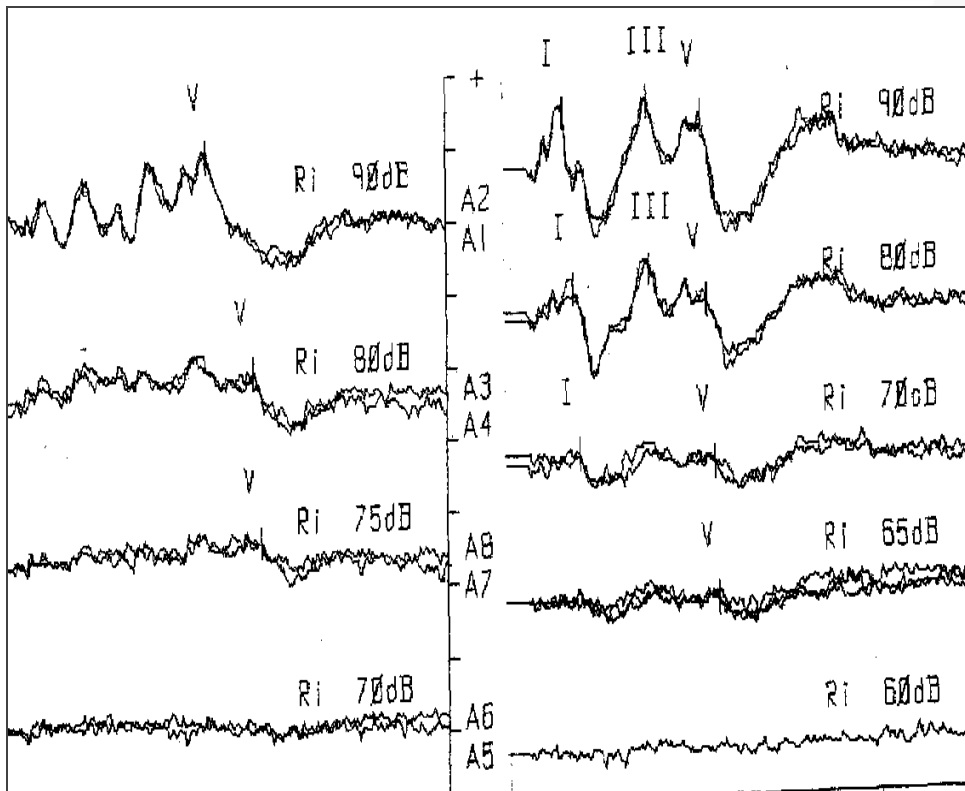


# Assessment: Electrophysiologic Measures

- **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
  - » 8<sup>th</sup> 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**

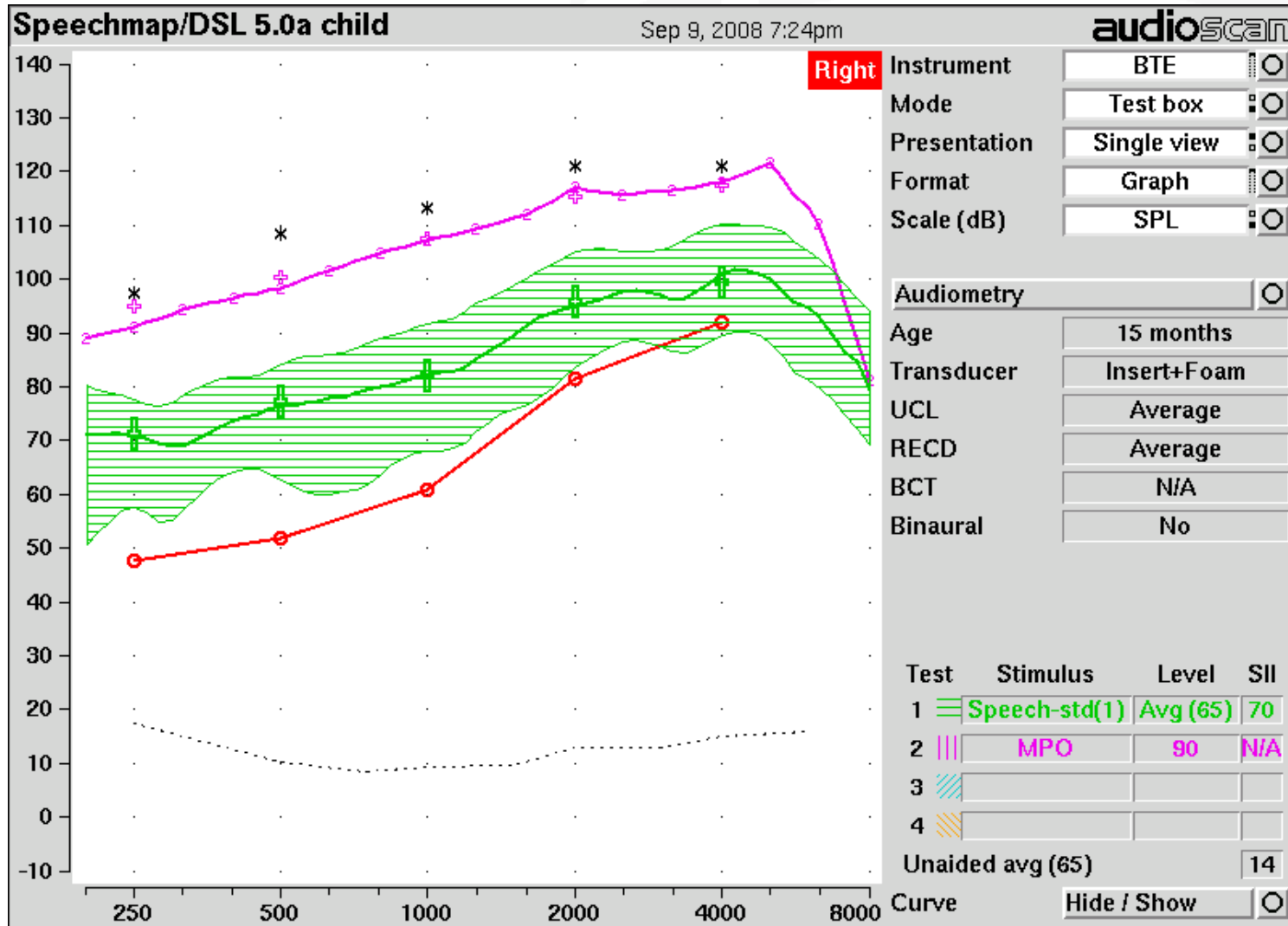
# Hearing Aid Fitting in Infants



- **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](http://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 )
  - (Ching and Hill, 2007, Zimmerman-Phillips, et al., 2000; Robbins, et al., 1991)
- 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**



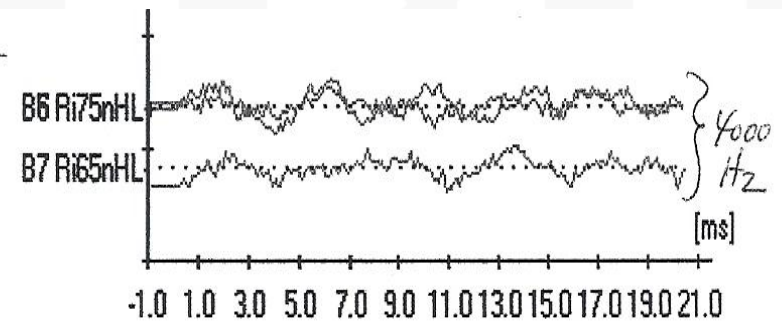
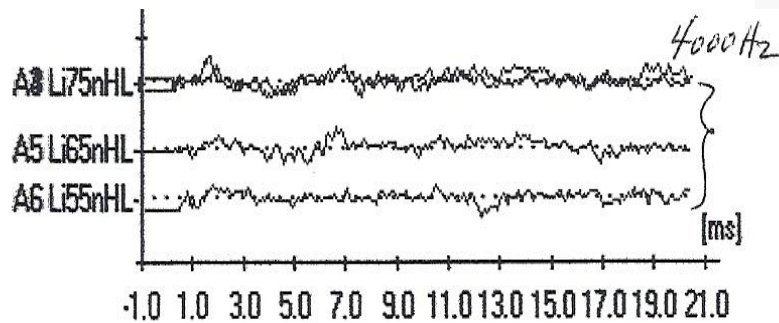
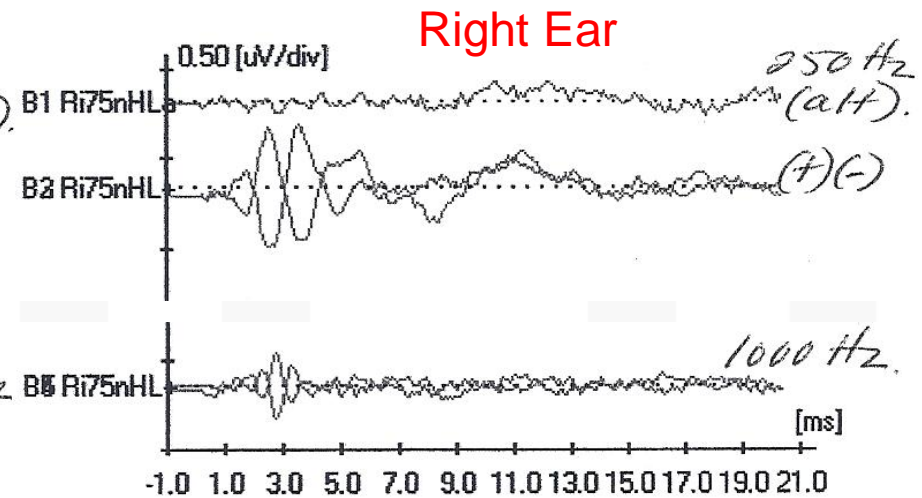
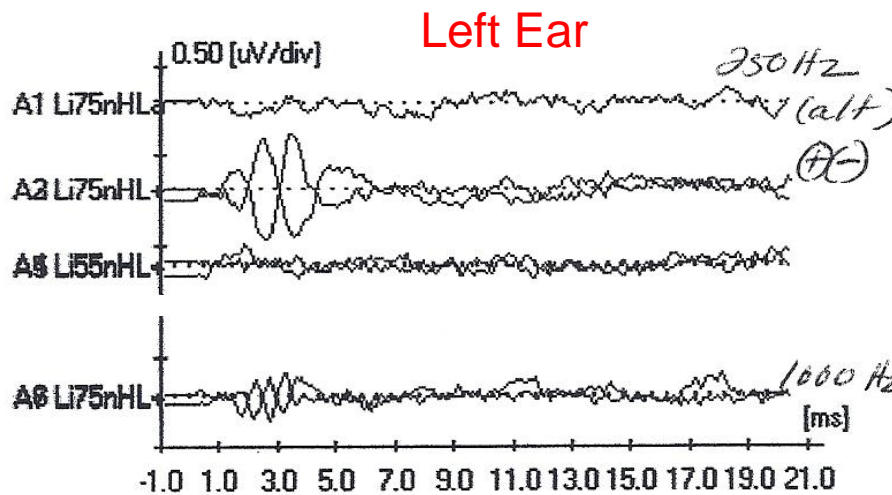
# Case #1



# Introduction

- Born at full term without complications
- Newborn hearing screening status unknown
- View the following slides and try to predict child's audiogram

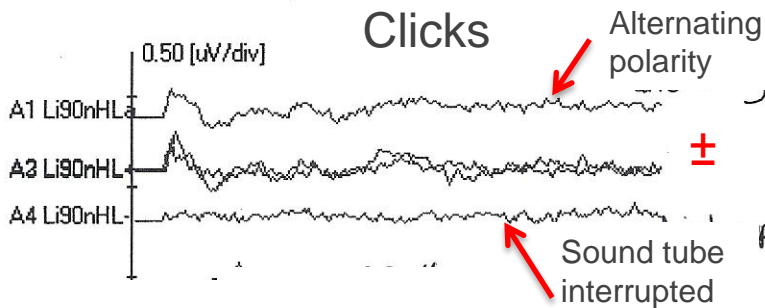
# First ABR in Natural Sleep



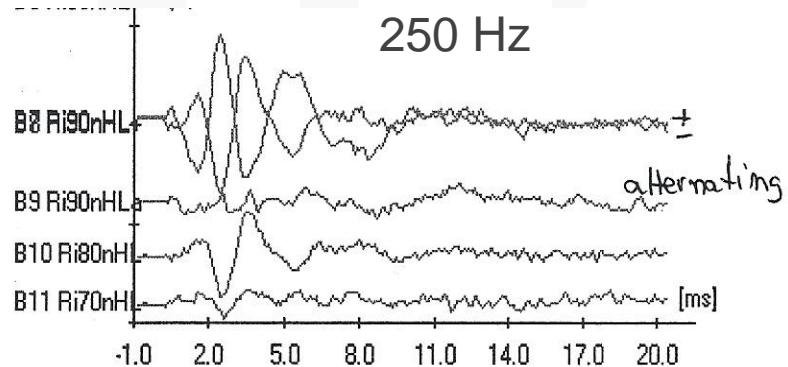
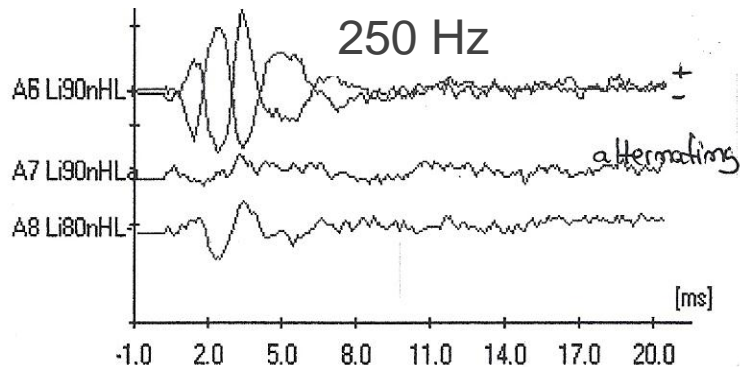
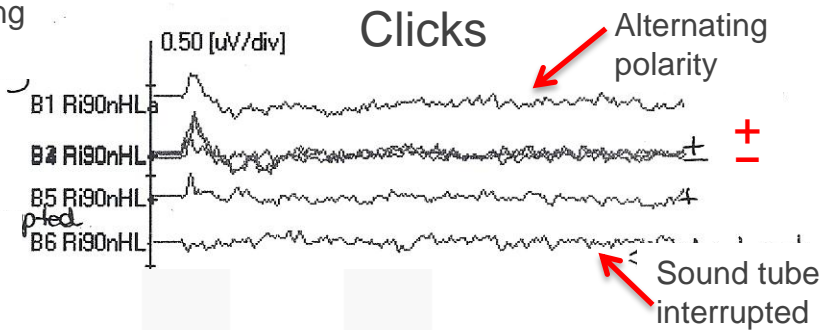
# Second ABR

## Under General Anesthesia Following MRI

Left Ear

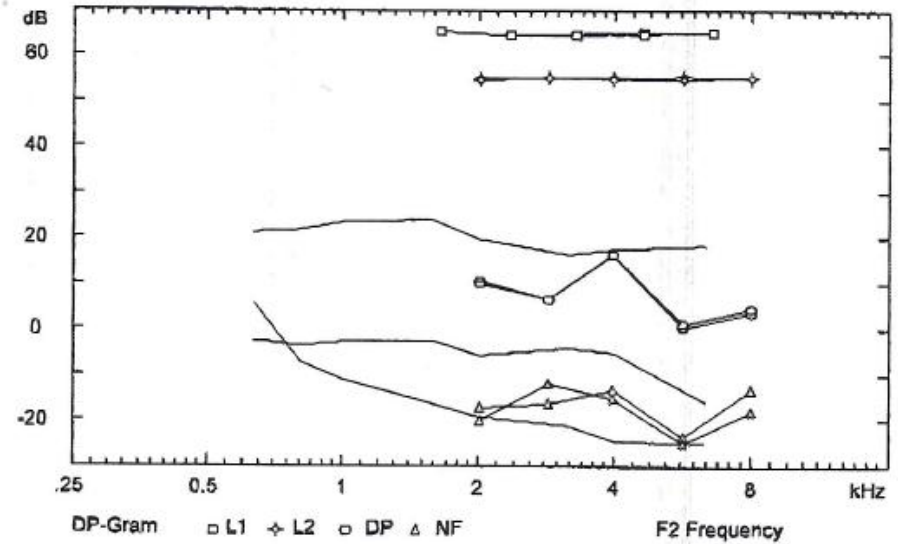
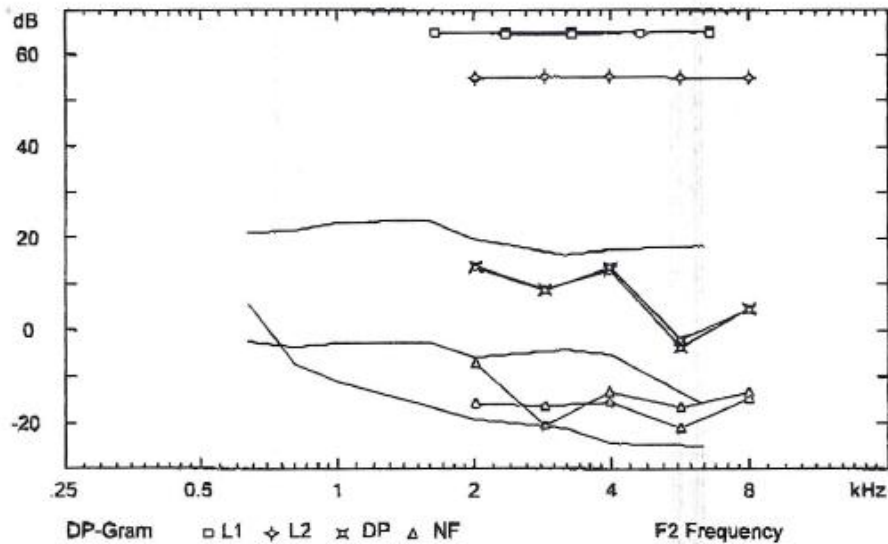


Right Ear



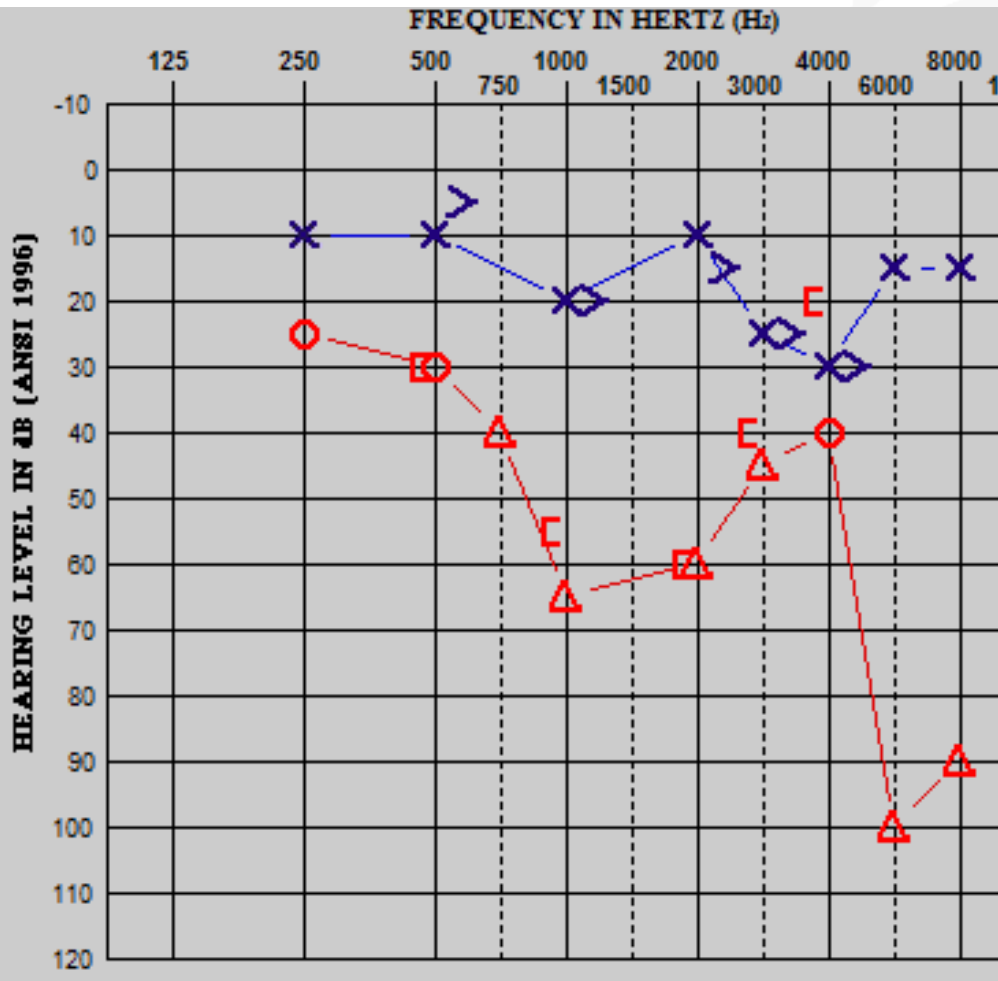


# Otoacoustic Emissions Present Bilaterally





# 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



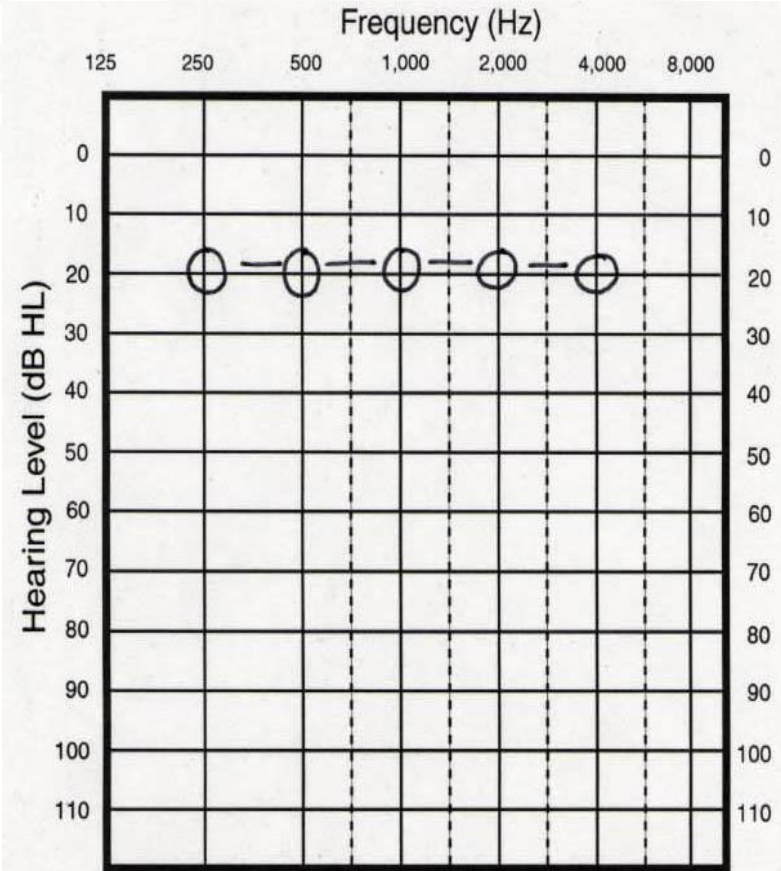
## Background:

- **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



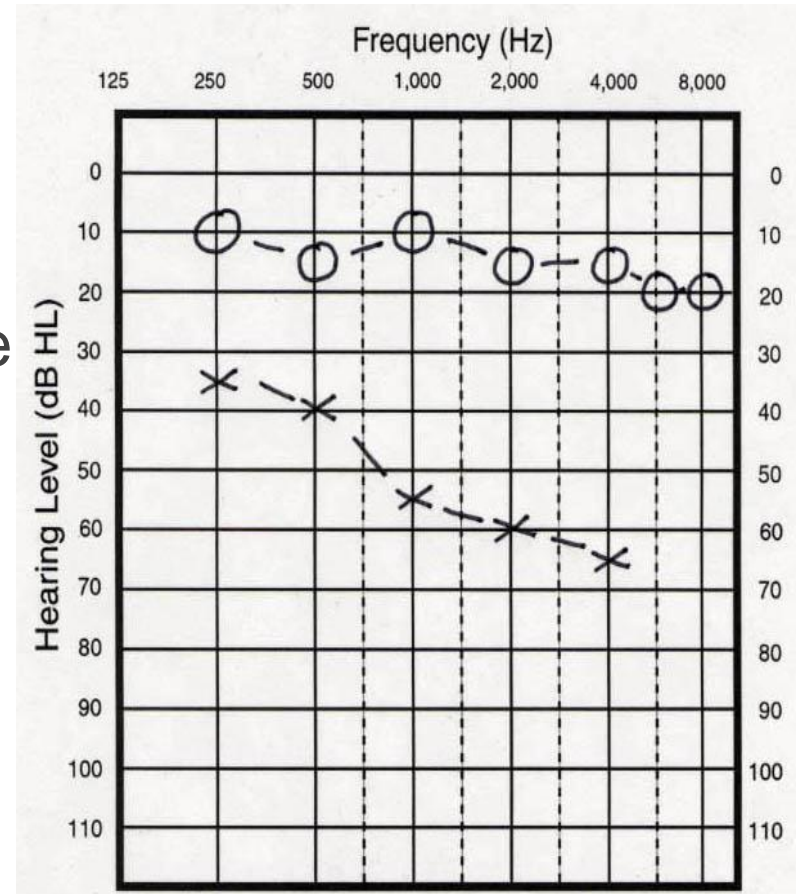
# Age: 8 months

- Tympanometry
  - » Right: normal
  - » Left: Negative middle ear pressure (-275)
- Otoacoustic Emissions
  - » Right: Absent above 2000Hz
  - » Left: Absent



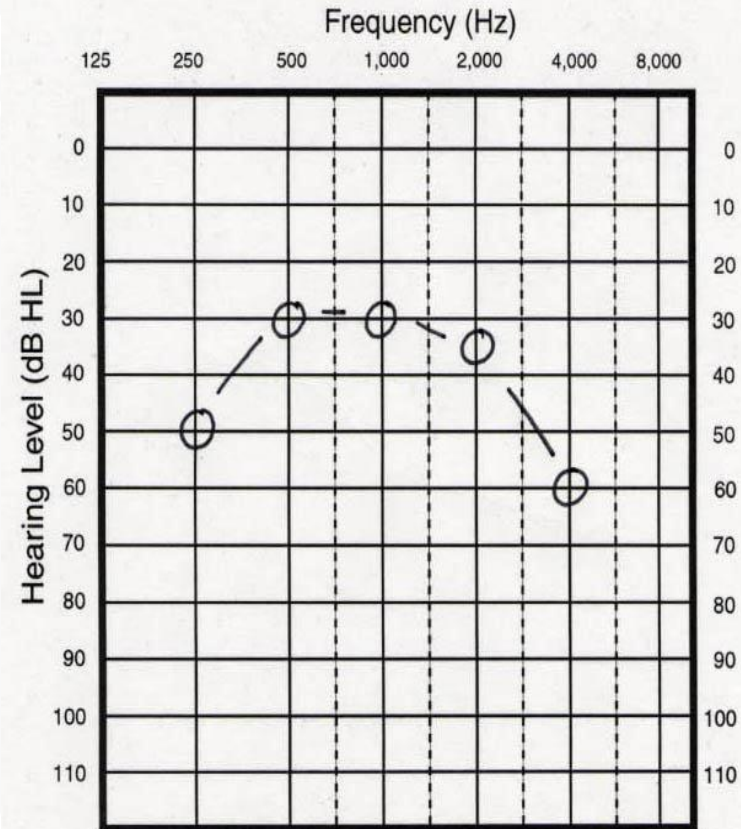
# Age: 12 months

- Right ear:
  - » Normal
- Left ear:
  - » Mild to moderate
- Tympanometry
  - » Right: normal
  - » Left: normal



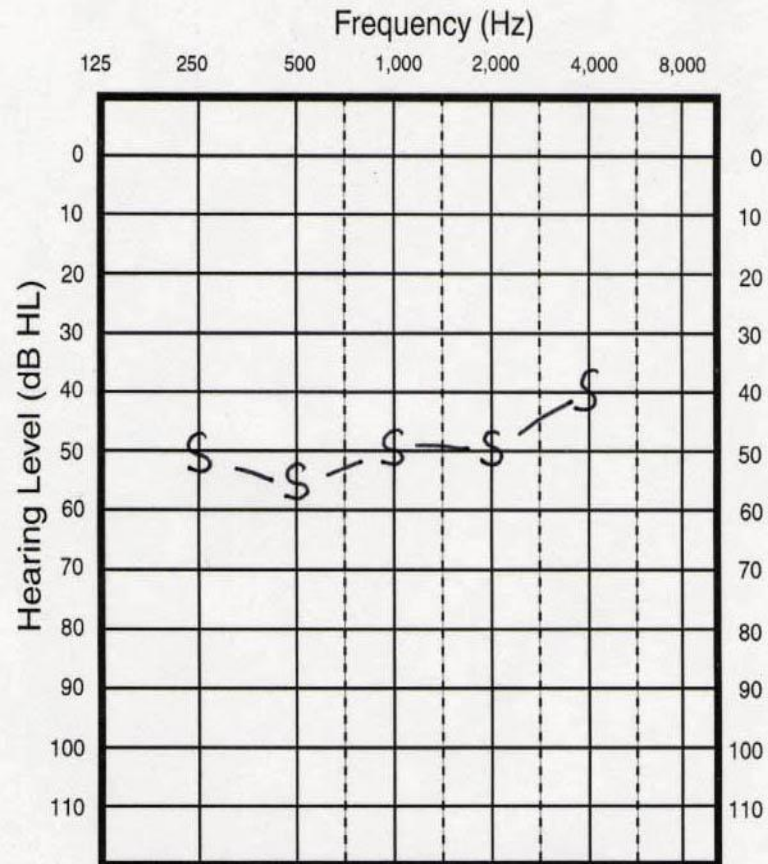
# Age: 17 months

- 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

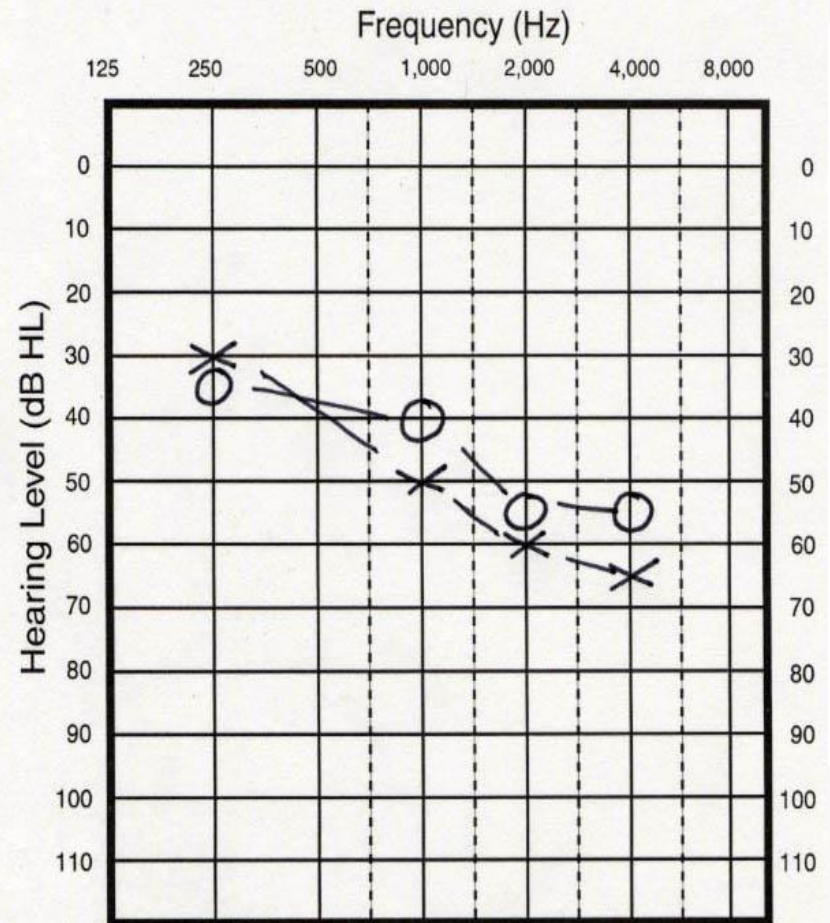
- Child will not tolerate insert earphones
- Unable to rule out hearing loss for “better ear”
- Tympanometry
  - » Right: Negative middle ear pressure (-225)
  - » Left: Negative middle ear pressure (-190)
- Repeat ABR with sedation recommended





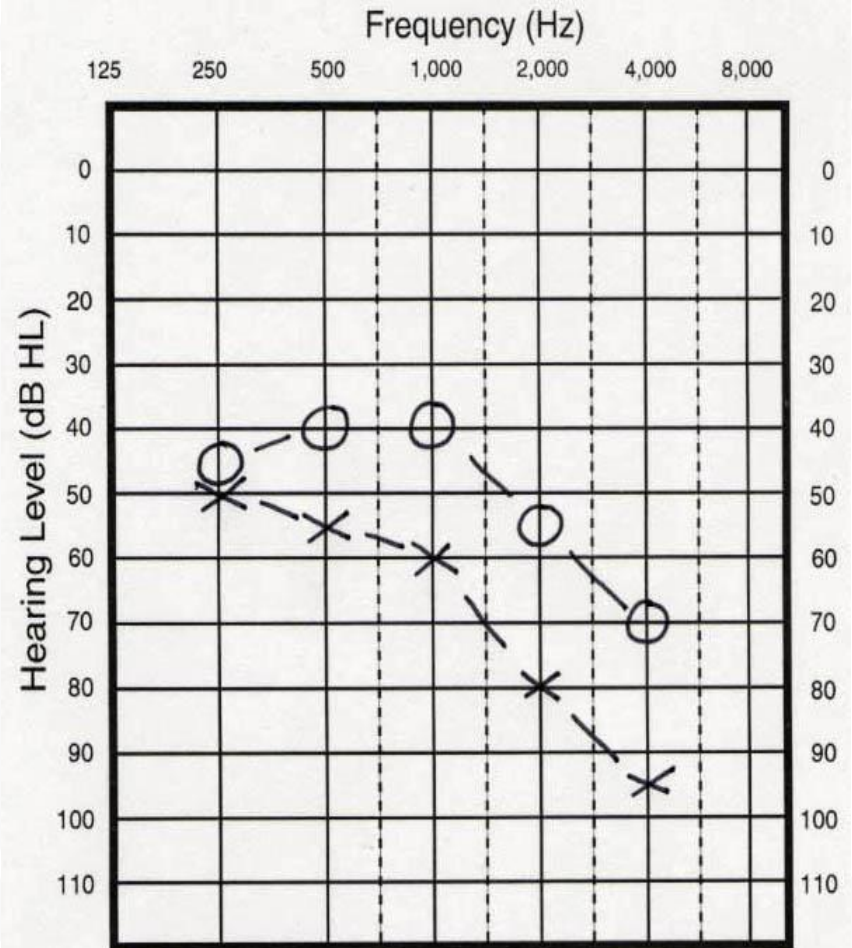
# Age 20 months: Estimated Thresholds (eHL) Based on Sedated Tone Burst ABR

- Binaural hearing aids and personal FM dispensed 2 weeks later



# Age: 22 months

- Continued progression of hearing loss noted
- Tympanometry
  - » Right: normal
  - » Left: normal

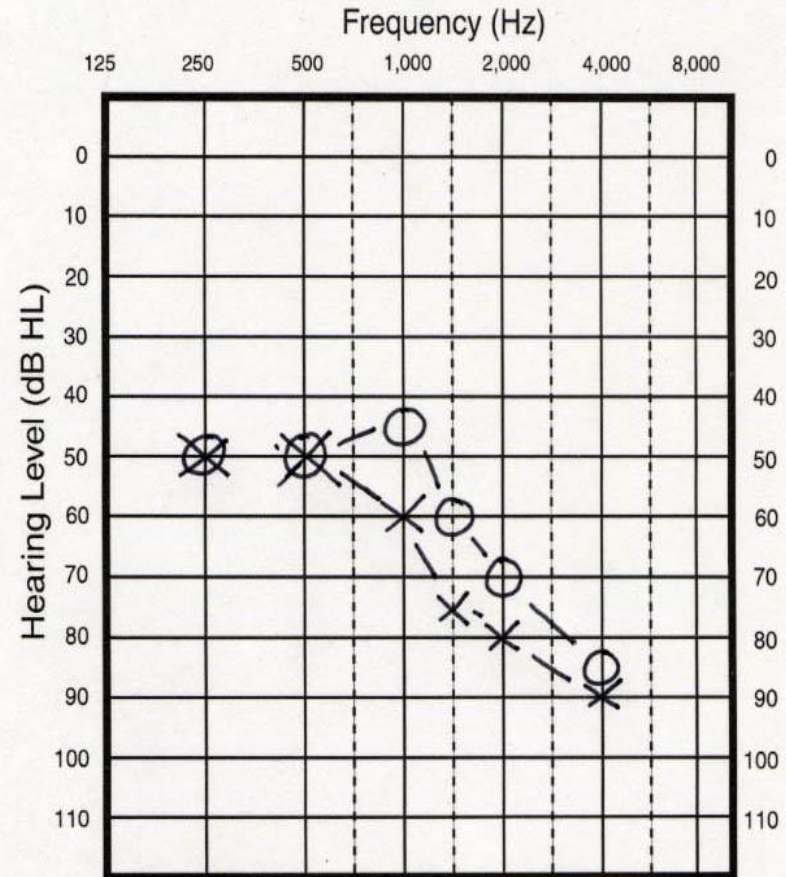






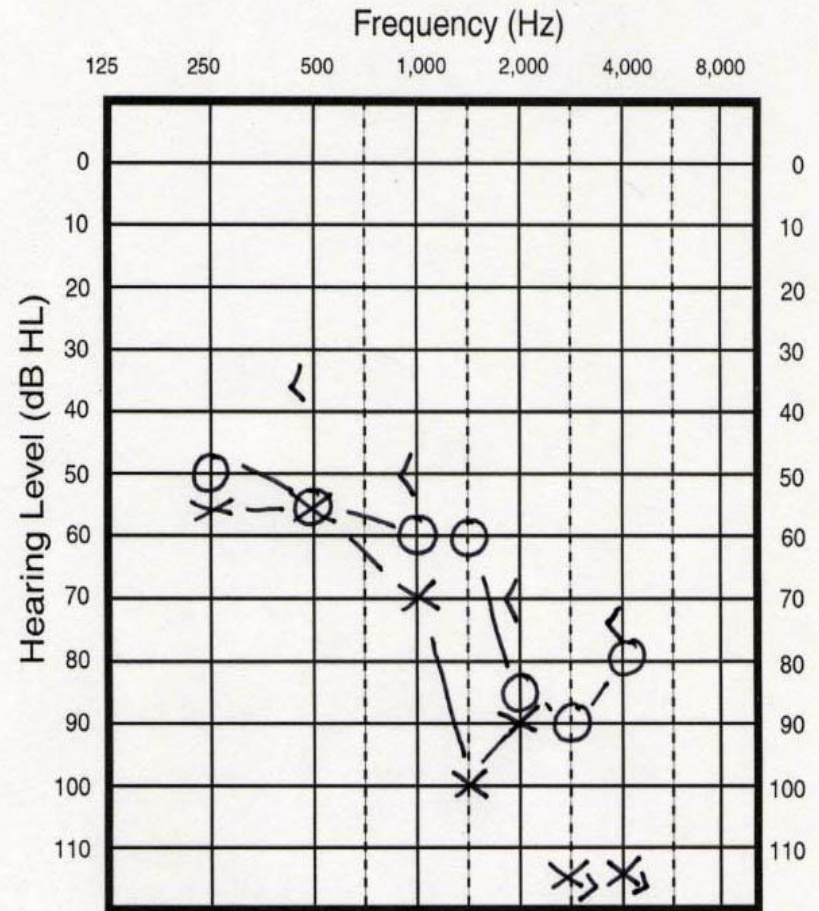
# Age: 23 months

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



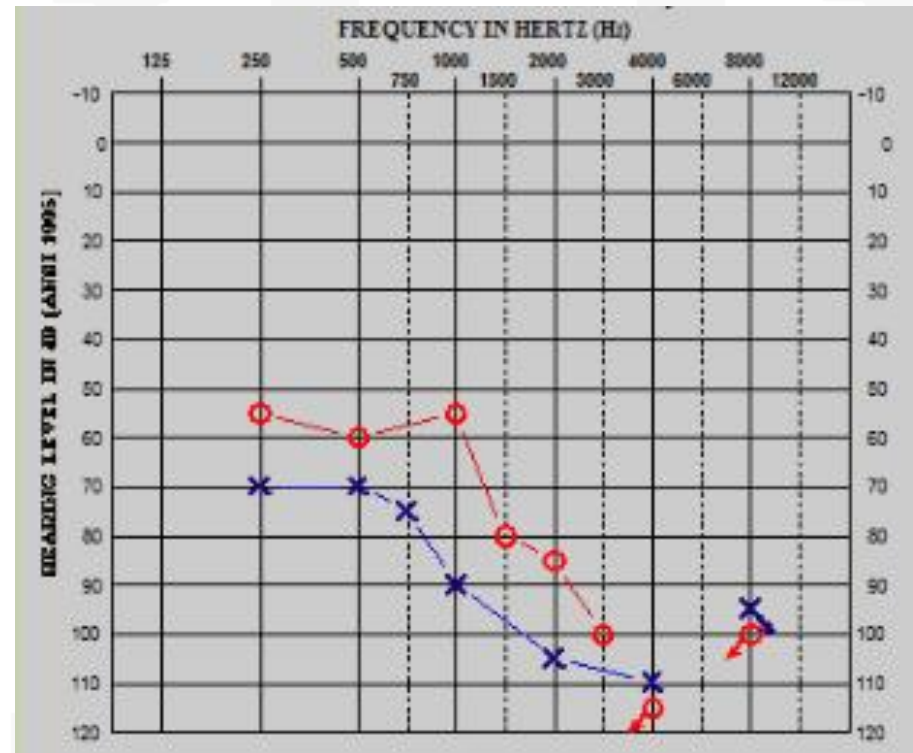
# Age: 24 months

- 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



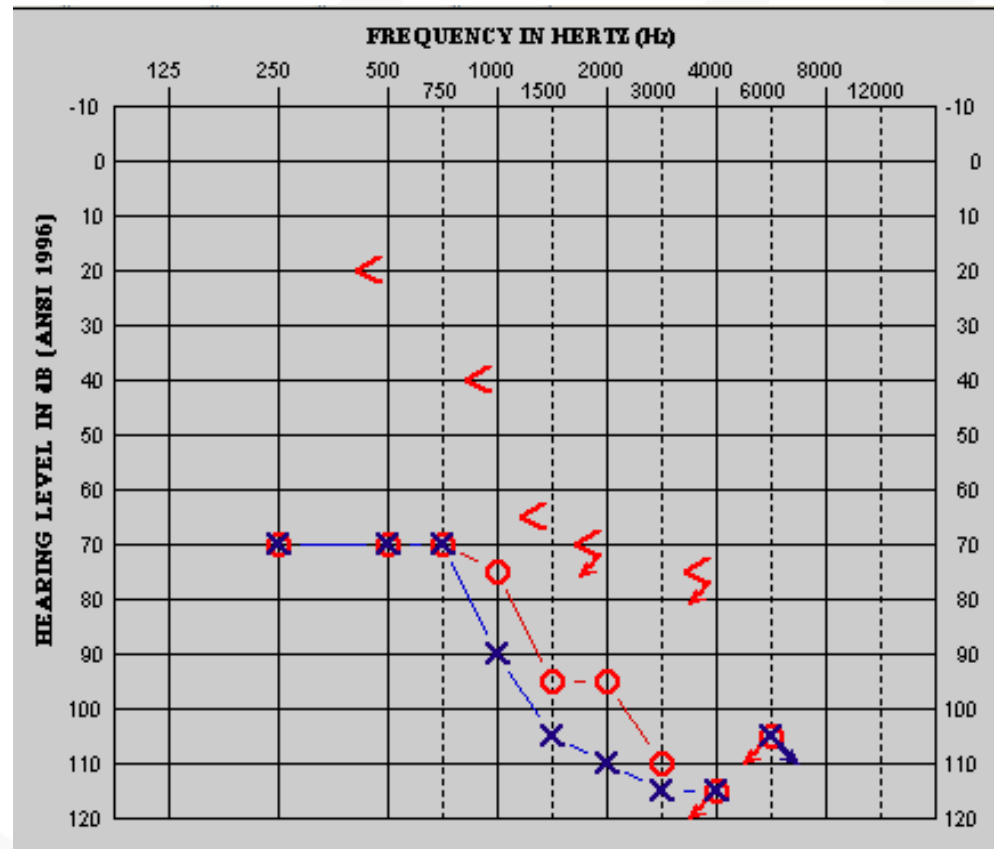
# Age: 4 years

- 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

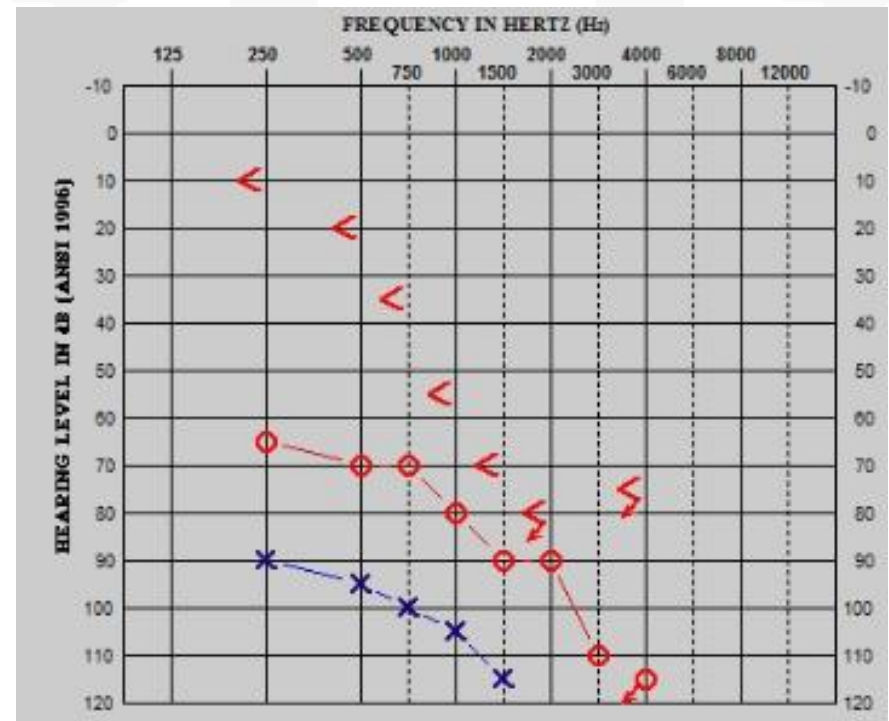
HA&CI: 15dBHL

Recorded monosyllabic words (PBKs):

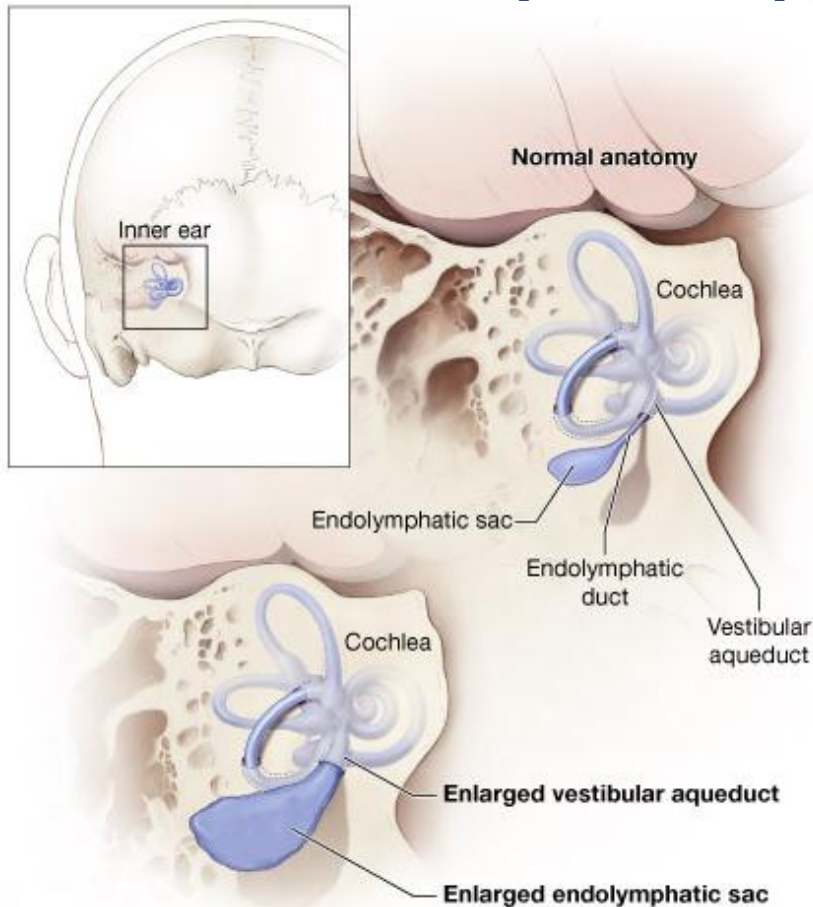
HA right: 40%

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)

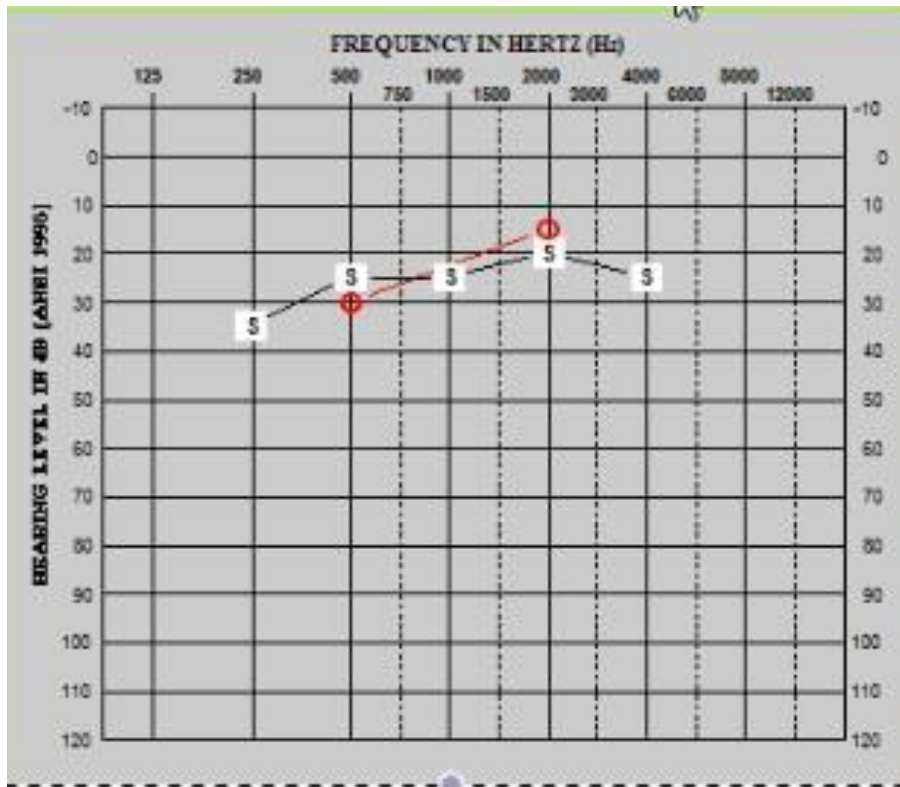


## Plan

- 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

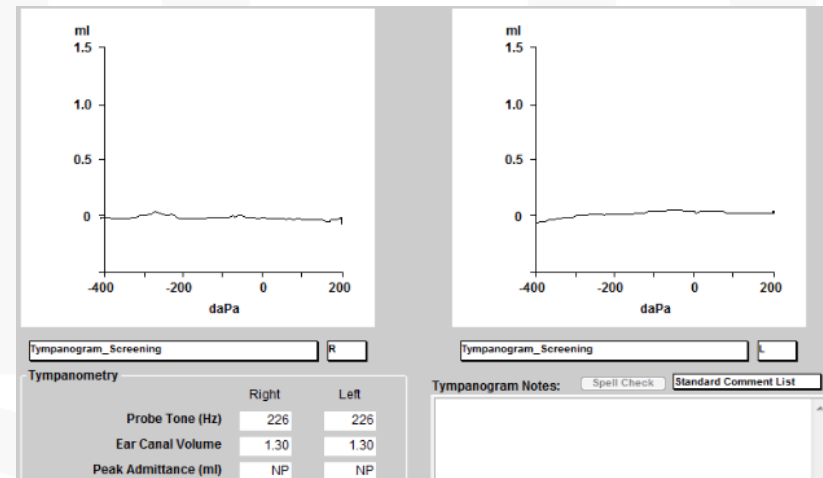
# Age 11 months

- Tympanometry:
  - » Flat with large physical volumes bilaterally



Right Ear

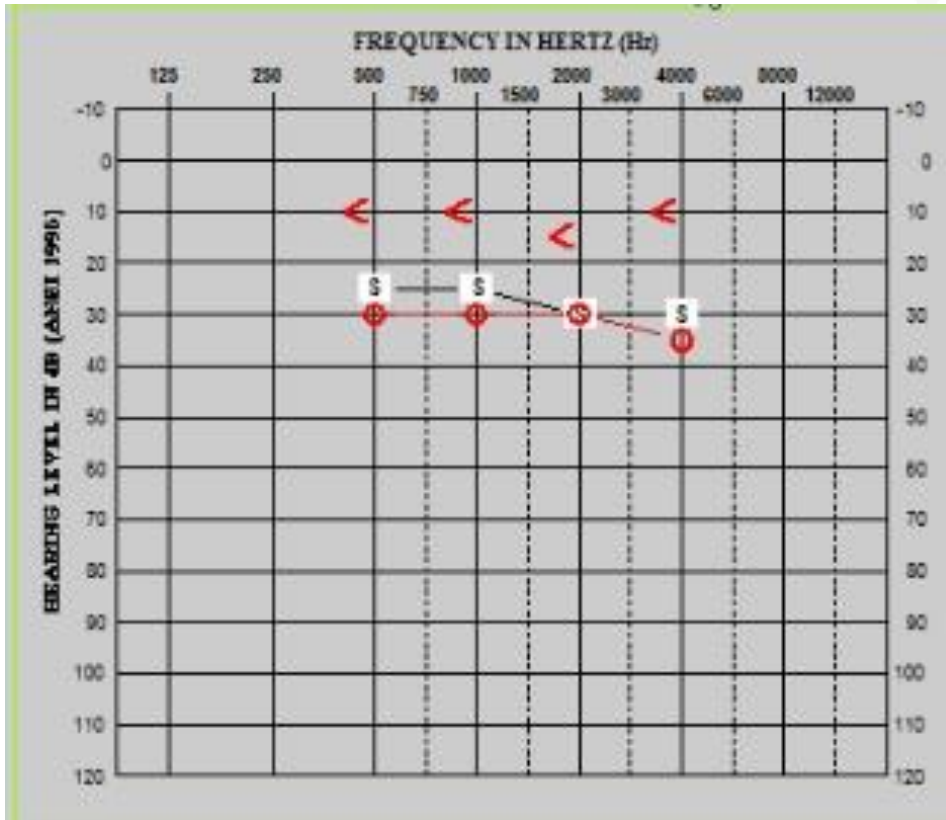
Left Ear





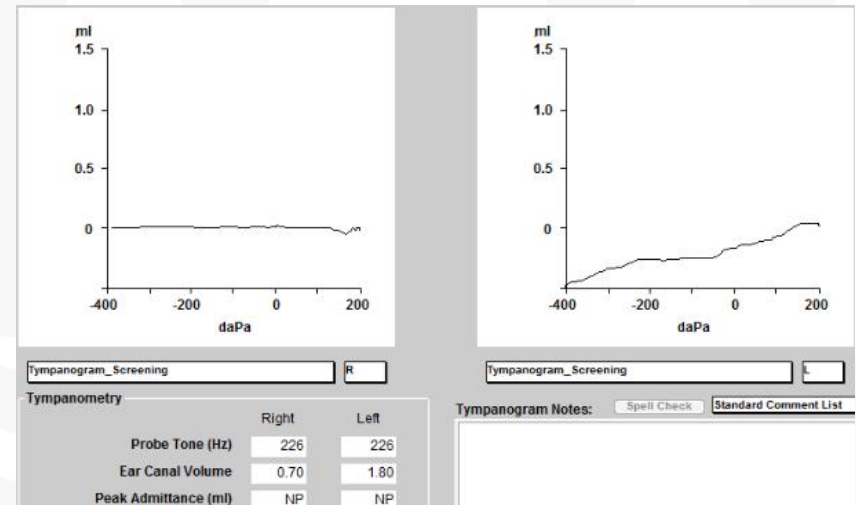
# Age 14 Months

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



Right Ear

Left Ear

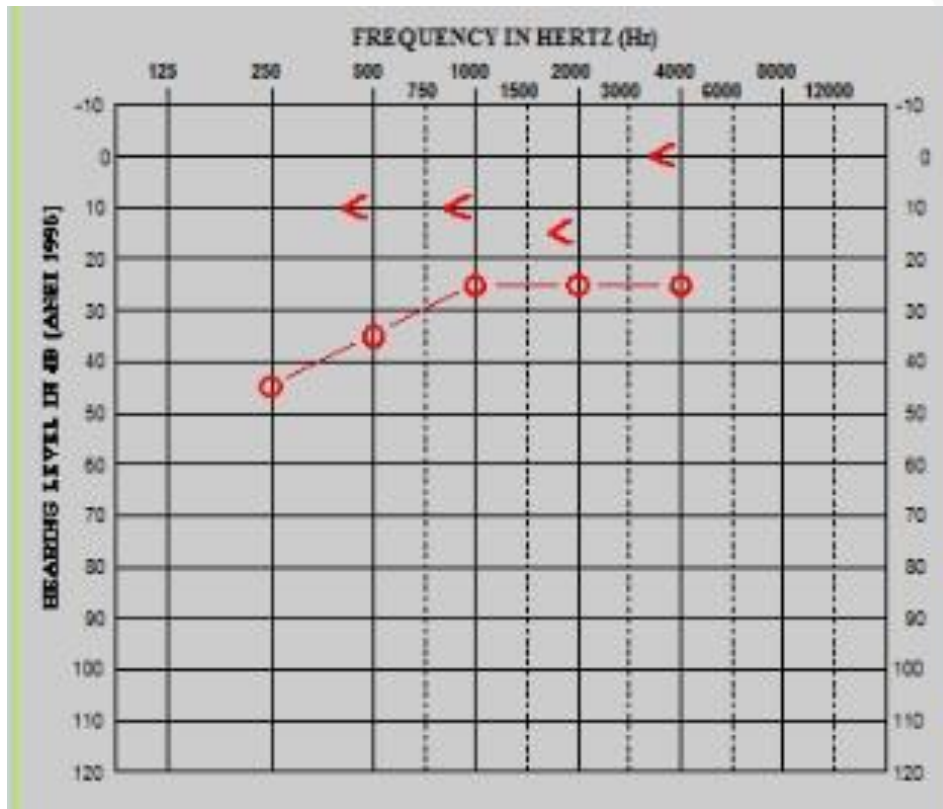


# Age 22 Months

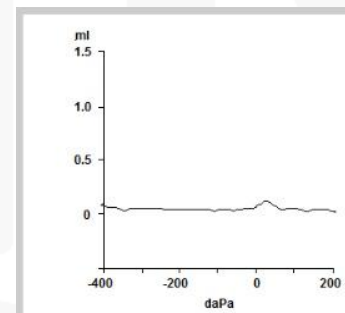
- Child making good progress with speech and language
- Mother offered but declined EI services because she feels child is doing very well

## Tympanometry:

- » Right: Flat with large ear canal volume
- » Left: Type A



## Right Ear

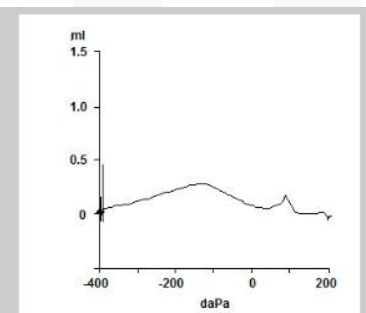


Tympanogram\_Screening R

### Tympanometry

	Right	Left
Probe Tone (Hz)	226	226
Ear Canal Volume	2.50	0.50
Peak Admittance (ml)	NP	0.40
Peak pressure (daPa)	NP	-365

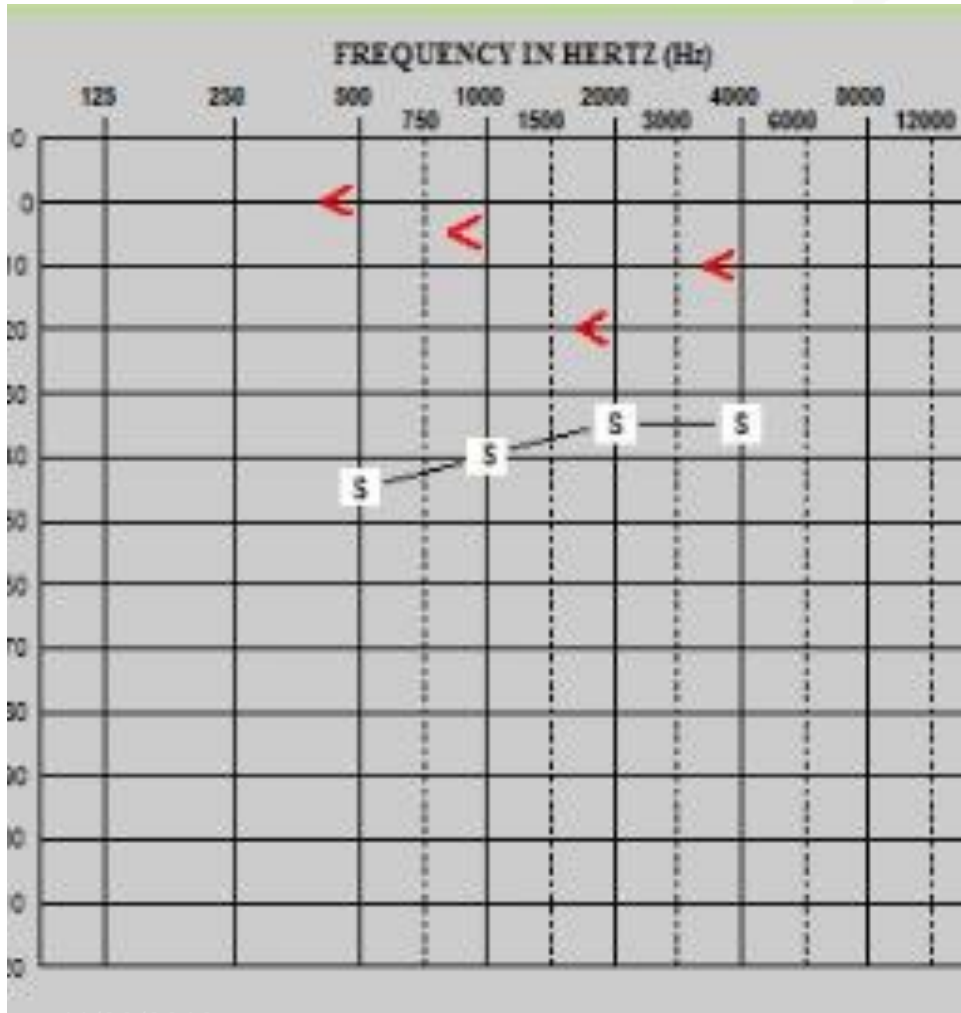
## Left Ear



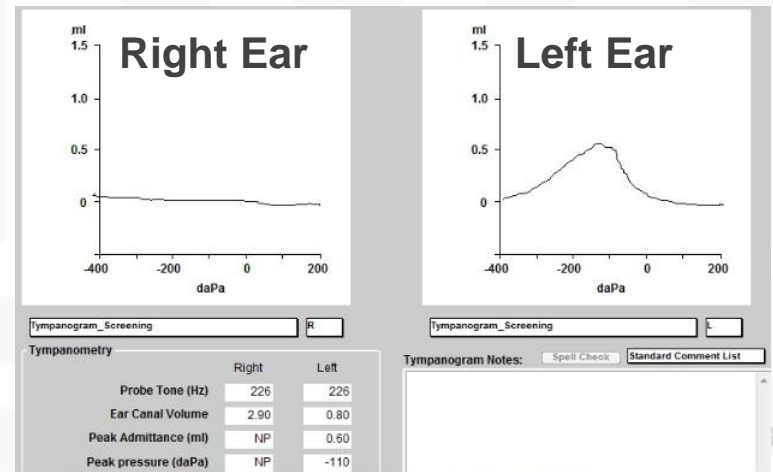
Tympanogram\_Screening L

Tympanogram Notes:

# Age 29 Months



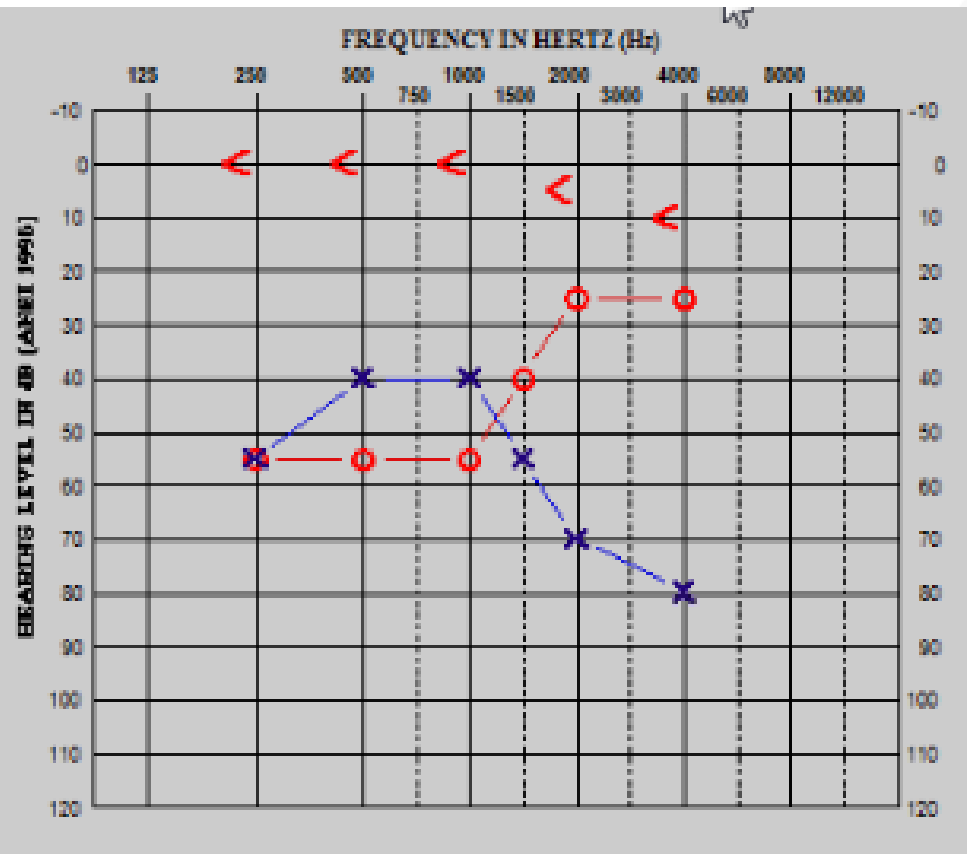
- Child more difficult to test
- Family feels child hears fine
- Tympanometry
  - » Right: Flat with large ear canal volume
  - » Type A left
- Repeat ABR recommended



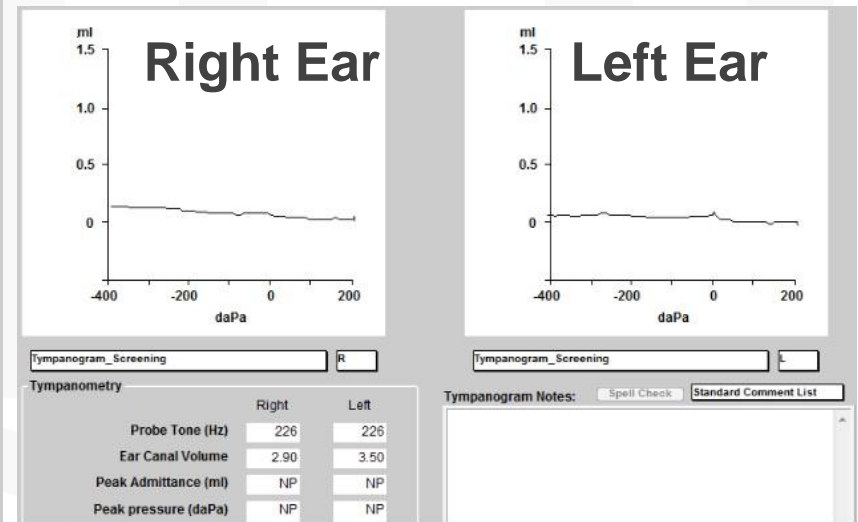
# ABR Test Results

- 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



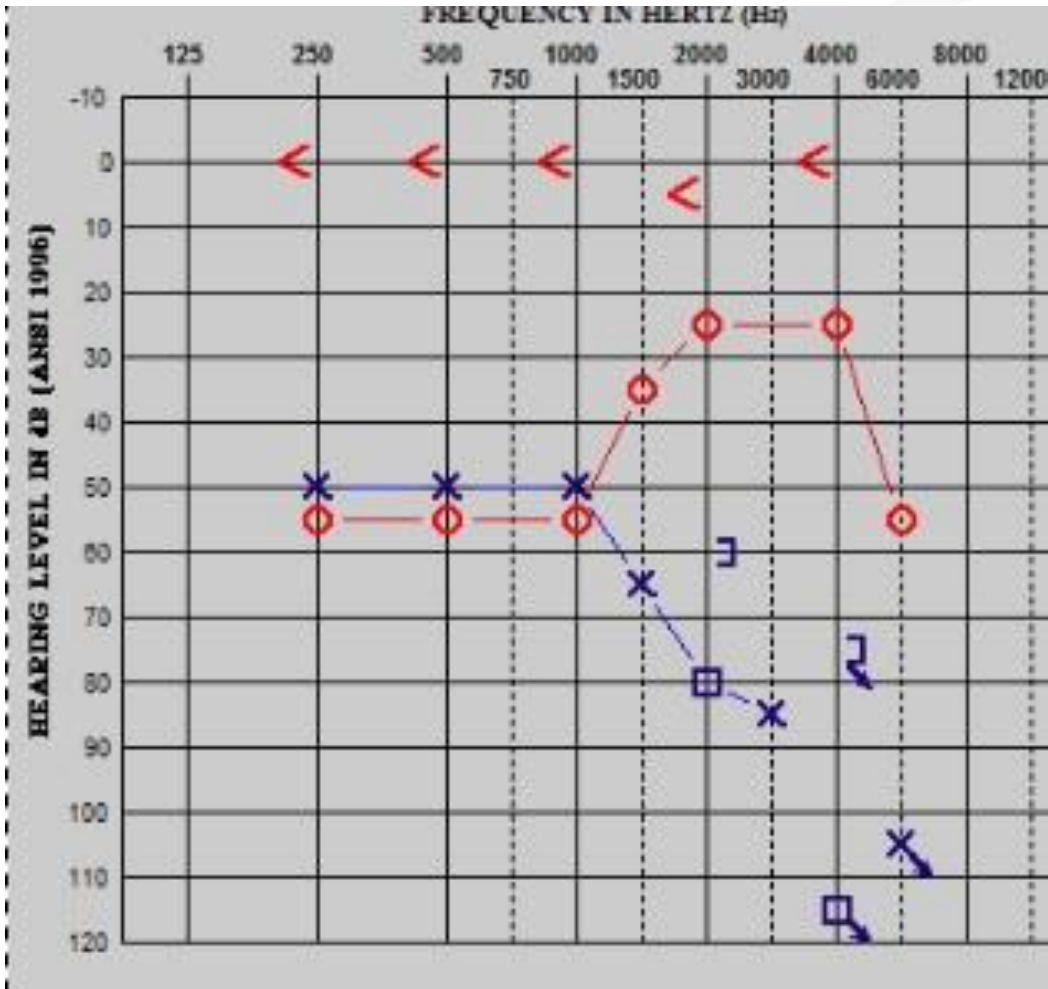
- Tympanometry:
  - » Flat with large physical volumes bilaterally





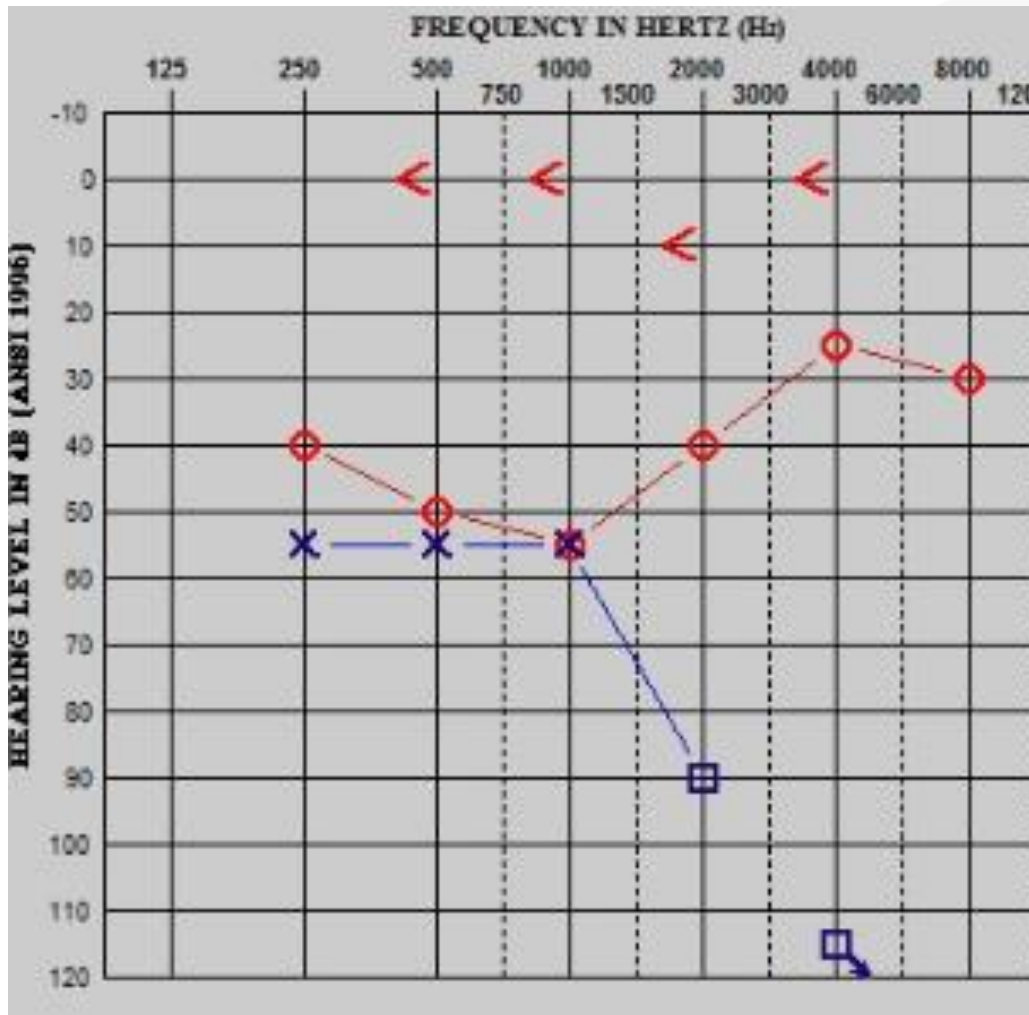


# Age 3 years, 5 months

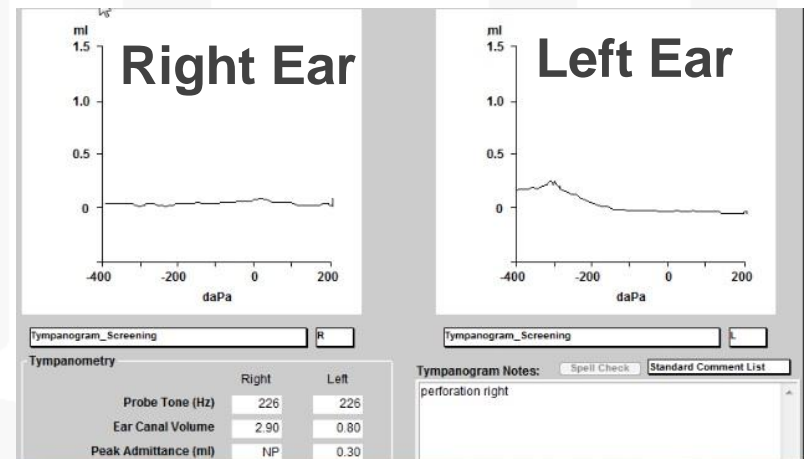


- Masked bone conduction testing and speech perception testing completed
- Score: 24/24 on ESP monosyllable test (closed set test)

# 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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