Early Hearing Detection and Intervention
How You Matter!

- Laura Davis-Keppen, MD
  - SD AAP EHDI Champion
  - Professor of Pediatrics, USD

- Jessica J Messersmith, PhD, CCC-A, FAAA
  - Associate Professor of Audiology, USD
  - Pediatric and Cochlear Implant Audiologist
Why is early identification of hearing loss important?

• Hearing loss is the most common birth condition
  • https://www.youtube.com/watch?v=TD5E88fFnxE

• Previous methods for detecting hearing loss have been ineffective

• Undetected hearing loss can delay speech, language, social & academic development
Hearing in Children

- Hearing loss is the most common birth abnormality
  - About 2 or 3 out of every 1000 children in the United States are born deaf or hard-of-hearing (NIDCD, 2013)
    - ~ 12,000 per year
  - ~ Another 4,000 to 6,000 infants and children are identified with hearing loss by age 3 years (NIDCD, 2013)
Identification is crucial

• Hearing is necessary to grow neural connections throughout the brain
  • Crucial for development of spoken language, reading, and auditory learning [Gordon, Papsin, and Harrison, 2004]
• Even those with profound HL can achieve auditory brain access through use of current technology
  • Allows developmental model of intervention rather than re/habilitative
Early identification is critical

• Children identified before 6 months of age have better developmental outcomes than those identified later (Yoshinaga-Itano et al., 1998)
  • Expressive and receptive language skills
  • Regardless of degree of hearing loss

• Higher vocabulary and verbal reasoning skills at age 5 for those children with hearing loss who began intervention by 11 months of age compared to children who began intervention later (Moeller, 2000)
Neural projections are impacted

- Left image: NH children: bilateral activation of the auditory cortical areas (superior temporal sulcus and inferior temporal gyrus)
- Middle image: early treatment: activation of the auditory cortical areas contralateral to their CI
- Right image: late treatment: activation outside the auditory cortical areas (in the visual, insula, and parietotemporal areas)

Gilley, Sharma, & Dorman, 2008
Newborn Hearing Screening

- Considerations for implementing universal screening (World Health Organization)
  - Is the problem significant?  
    - To the individual
    - To society
  - Is there good evidence on an effective treatment once the problem is detected?
  - Has the screening test been properly evaluated and shown to be appropriate in the setting where the screening is to be performed?
  - Is there evidence that a screening program that leads to treatment is of greater benefit than waiting until symptoms develop?
  - Have cost issues been considered and developed?
  - Are there plausible strategies and sufficient resources to ensure implementation?

Yes!  
Yes!  
Yes!  
Yes!  
Yes!
Previous methods for detecting hearing loss have been ineffective

- High risk screening failed to identify ~ 50% of the infants with hearing loss
- Large retrospective cohort study5, 6: mean age of diagnosis 21.6 months
- Similar findings reported in US7,8,9
EHDI: EARLY HEARING LOSS DETECTION AND INTERVENTION

The process
Early Identification of Hearing Loss - Method

Screening
• No later than 1 month of age

Diagnostic testing with Pediatric Audiologist
• No later than 3 months of age

Early Intervention
• No later than 6 months of age

1:3:6
Screening method

AABR
- More $$$ than OAE
- Sensitive to more types of HL
- Needed if baby in NICU >5 days

OAE
- Less $$$ than AABR
- Faster than AABR
- Not sensitive to all types of HL

Can obtain invalid results from both if obstruction in middle or outer ear (e.g. fluid or vernix)
2007 JCIH Position on Screening

**NICU**
- >5 days in NICU
- ABR should be included to screen for neural loss
- Rescreen BOTH ears, even if only one ear fails
- Non pass – refer to Audiologist
- Readmission – rescreen before discharge

**Well baby nursery**
- Screen with OAE or ABR
- Repeat screen when necessary before discharge
- When using 2 step protocol test order should be OAE then ABR
- Rescreen BOTH ears, even if only one ear doesn’t pass
Does UNHS Improve the Age of Identification and Intervention?

• Prior to UNHS (Harrison and Roush, 1996)
  • Median age of identification without risk factors was 13 months for severe-to-profound SNHL
  • Median age of identification with risk factors was 12 months for severe-to-profound SNHL

• After UNHS (NY State, 2000)
  • Median age of identification was 3 months

• EHDI 10 years later- (Holte et al., 2012)
  • only 32% children included in NIH OCHL study met all of the 1-3-6 benchmarks
What is the role of the provider and nurse?

- Discuss results
- Facilitate visit for rescreen
- Like any newborn screen, the initial screen is **not** diagnostic
What is the role of the provider and nurse?

- Ensure that family understands that a “pass” does not mean that a HL will not be dx later on
- Children who pass UNHS but have risk factor should have at least one dx audiology assessment by 24-30 months of age (JCIH, 2007)
What is the role of the provider and nurse?

- Connect infants to audiologic follow-up

Initial screening @ hospital

- Pass
- No pass

No pass

Rescreen

- Pass
- No Pass
Who is an audiologist?

- Audiologist are the primary health care professional who diagnose, treat, and manage hearing loss, tinnitus, and/or balance disorders.
  - Doctoral level education
  - Licensure requirements
  - Certification
Only refer infants to a pediatric audiologist

http://www.ehdi-pals.org/
A pediatric audiologist is best equipped to diagnose hearing loss in infants/children

- Detailed history
- Diagnostic evaluation using a test battery
  - No one single test can identify the type, degree, and configuration of HL
    - Otoscopy
    - Immitance
    - Physiologic
    - Behavioral
- Counsel parents
Treatment plan

• Mode of communication is parent(s) choice
  • If spoken- pursue amplification ASAP (pediatric audiologist)
    • No age limit on fitting amplification
    • Cochlear Implant Center should be involved in those case of severe or greater HL, bacterial meningitis, ANSD, those demonstrating limited benefit from amplification
  • If sign- connect family with sign support
What is the role of the provider and nurse?

- Talk with family about results
- Review family history and examine child for craniofacial abnormalities or syndromes associated with hearing loss
- Refer to ENT
- Refer to ophthalmology
- Refer for genetic testing/consultation

No Dx HL

Audiologist

Dx HL
National EHDI Data

Universal Newborn Hearing Screening

Percent of Infants Receiving Hearing Screening: 1999-2007

1999 2001 2003 2005 2007 2009 2011
Infants Identified as Permanently Deaf or Hard Of Hearing, 2005-2012. *Total = 34,416*

National EHDI Data
Incidence of Children who are Deaf or Hard of Hearing
2012 National CDC EHDI Data

• % Screened: 96.6% (n=3,820,624)
• Prevalence of children who are deaf/hh: 1.6 per 1,000 screened (Range 0.0-4.3 per 1,000)
• % of those identified receiving Early Intervention: 61.7% (n=3,527)

• % Screened before 1 month of age: 86.0% (n=3,287,614)
• % Diagnosed before 3 months of age: 69.1% (n=20,102)
• % Receiving Intervention before 6 months of age: 67.1% (n=2,367)

• % Loss to Follow-up or Documentation: 35.9% (n=19,006)
2012 South Dakota EHDI Data

- % Screened: 97% (n=12,722), 96.4% by 1 month
- Rescreened 65%
- Audiologist by 3 months 77.7%
- Birth to 3 only 18.75%
- 33.4% of South Dakota’s newborns who should have been rescreened did not have hearing evaluated.
- Based on national data 68.6% of SD infants who were deaf/HH did not receive early diagnosis and intervention. Early intervention should be considered a developmental emergency. Children identified as deaf/hh who begin services before 6 months develop language (spoken or signed) on a par with their hearing peers.
- 86.8% loss to follow-up/loss of documentation
The Role of Medical Home

Early Hearing Detection and Intervention

- The medical home plays a key role in the success of EHDI programs.

- A medical home can help families understand the EHDI process.

- The medical home ensures that appropriate and timely steps are taken to identify children who are deaf/hh and get them into an early intervention program.

- The medical home serves as the primary coordinating entity which can help significantly reduce loss to follow-up/documentation.
Medical Home: Strategies to Promote Follow Up

- At prenatal visit, encourage families to identify you as follow-up care location
- Inform hospital to facilitate communication of results
- Provide checkbox on newborn well child form/patient chart for hearing screening results & risk factors
- AAP: The primary care provider must assume responsibility to ensure that audiological assessment is conducted on infants who do not pass screening and must initiate referral for medical specialty evaluations necessary to determine the etiology of the hearing loss. Set up tracking system for infants who do not pass hearing screening.
Counseling Parents

• Effective communication of results to families has an influence on follow up behaviors

• Balance between reassurance and importance of follow up testing

• “Your infant may or may not have a hearing loss…but let’s be sure about it. If further testing shows hearing loss, the earlier we get started helping the child, the better.”
Optimal Surveillance in the Medical Home (JCIH, 2007)²

- If hearing loss is diagnosed, refer siblings of infant for audiological evaluation
- Refer infants with any RISK indicators for audiological assessment by 24-30 months of age
- Carefully assess middle ear status at all well child visits; refer for otologic evaluation if persistent middle ear effusion lasts for > 3 months
Risk Indicators of Delayed Onset or Progressive Hearing Loss

- Caregiver concerns*
  - about hearing, speech, language, development

- Family history*
  - of permanent childhood hearing loss

- NICU stay > 5 days or any of following (regardless of length of stay):
  - ECMO assisted ventilation*
  - Ototoxic medications (gentamycin, tobramycin)
  - Loop diuretics (furosemide, Lasix)
  - Hyperbilirubinemia requiring exchange transfusion

JCIH, 2007
Risk Indicators for permanent congenital, delayed onset or progressive hearing loss

- In Utero infections
  - CMV*, herpes, rubella, syphilis, toxoplasmosis

- Craniofacial anomalies

- Physical findings (e.g. white forelock)

- Syndromes* involving hearing loss
  - Neurofibromatosis, osteopetrosis, Usher, Waardenburg, Alport, Pendred, Jervell & Lange-Nielson

* = greater risk for delayed onset HL
Risk Indicators for permanent congenital, delayed onset or progressive hearing loss

- **Neurodegenerative disorders**
  - Sensory motor neuropathies (Friedrich ataxia, Charcot-Marie-Tooth)

- **Culture positive postnatal infections associated with HL**
  - Herpes, varicella, meningitis

- **Head trauma** (basal skull, temporal bone)*
- **Chemotherapy***

* = greater risk for delayed onset HL
Medical Workup

- Complete prenatal & perinatal history
- Family Hx of onset of HL < age 30
- Physical exam: unusual facial appearance with attention to asymmetry, ear anomalies, neurologic, balance, skeletal, other unusual physical findings
- Test for CMV as soon as possible, before age 6 weeks.
- Refer to ENT
- Refer to Genetics and Ophthalmology
- As needed: developmental pediatrics, cardiology, neurology
Causes of Prelingual Hearing Loss > 40dB

Prelingual Deaf Children 1/500

Idiopathic 25%

Non-genetic 25%

Genetic 50%

Nonsyndromic 70%

Syndromic 30%

Autosomal recessive 75% - 85%

Autosomal dominant 15% - 24%

X-linked 1% - 2%

DFNB1 50%

Other DFNB 50%
Waardenburg Syndrome
Abnormal pigmentation of skin and hair
Lateral displacement of medial canthi-type I
Heterochromia iridis
Deafness in some—not always present
Branchio-Oto-Renal (BOR) Syndrome

Pre-auricular pits 82%
Branchial fistulae 49%
Cupped or mildly altered auricle 36%
Renal abnormalities 67%
Hearing loss 93%
• Mixed 52%
• Conductive 33%
• Sensorineural 29%

Autosomal Dominant: EYA1 gene (8q13.3) or SIX5 gene (19q13.32)
Treacher Collins Syndrome

Malar hypoplasia
Downslanting palpebral fissures
Defects of lower lid
Malformations of external ear
Usher syndrome
3-6% of Children Who Are Deaf or Hard of Hearing Develop retinitis pigmentosa - deterioration of vision

Left: Usher syndrome - Optic nerve - pale,
Vessels (stars) very thin,
Characteristic pigment (double arrows)
Usher Syndrome

- Usher Syndrome Type I (70%)
  - Congenital, profound SNHL
  - Childhood onset of retinitis pigmentosa
  - “Clumsiness” & developmental delay
  - Absent vestibular function

- Type II (26%)
  - Congenital SNHL, high frequency > low frequency, stable
  - Later onset of retinitis pigmentosa
  - Normal vestibular function

- Type III (4%)
  - Normal hearing and vision at birth
  - Deterioration of hearing and vision over the years
Jervell and Lange-Neilsen Syndrome

**Prolongation of QT interval** on EKG. May develop arrhythmias leading to **sudden death**
- Profound sensorineural hearing loss
- **Autosomal recessive** inheritance. Due to mutations in the *KVLQT1* gene on chromosome 11p15 and *KCNE1* gene on chromosome 21q22
- potassium channel genes
70% of Genetic Hearing Loss is Nonsyndromic

- **DFNB1** locus which includes the **GJB2 gene** encoding the gap junction protein **connexin 26** and **GJB6 gene** encoding the gap junction protein **connexin 30** is the most common cause of AR nonsyndromic HL

- **Next-generation sequencing** technologies are replacing single gene-sequencing tests for hearing loss.

- **Advantage**: ability to address **genetic heterogeneity** since many different genes results in phenotypes that cannot be distinguished clinically.

- **Disadvantage**: Large deletions and duplications and copy-number and structural variations not as efficiently detected so alternative genetic testing strategies may be necessary
Comprehensive Genetic Testing for Hereditary HL Using Massively Parallel Sequencing

- The most common cause of prelingual-onset SNHL is genetic in developed countries.

- Panels may test for all genes known to cause NSHL, Usher syndrome, and Pendred syndrome using custom-targeted sequence capture for DNA enrichment followed by massively parallel DNA sequencing. All genes sequenced at the same time.

- The panel we use currently tests for 109 genes known to cause NSHL and some syndromic HL. Total of genes currently tested 116 genes.

- **Cost effective**-$1500.

- Familial mutation testing $200.
Targeted Next-Generation Sequencing of Deafness Genes

• When 30 individuals with nonsyndromic hearing loss were tested using targeted next-gen sequencing, 52% of the probands were diagnosed with monogenic nonsyndromic HL.

• Determining the etiology of HL provides answers: recurrence risk, prognosis (will HL worsen over time), best treatment (cochlear implants or hearing aids), and whether vision will also be later affected.
Other Tests/Recommendations

- CMV recommended as an initial test if the NB hearing screen is not passed. Congenital CMV is a leading cause of hearing loss—found in 30%.
- CMV testing most diagnostic when done before 3-6 weeks. With increasing age there is a greater likelihood that a +CMV test is due to postnatal exposure.
- Consider testing for mitochondrial mutations associated with aminoglycoside ototoxicity for individuals with a history of aminoglycoside antibiotics.
- Every infant with confirmed HL should have an evaluation by an ophthalmologist.
- Temporal bone imaging should be considered if indicated.
CMV

- CMV is the leading cause of nongenetic hearing loss
- 1/3 of NSHL in young children
- At birth infant may be asymptomatic or have microcephaly, IUGR, petechiae, hepatosplenomegaly
- Hearing loss may be fluctuating and progressive
- Stabilization or improvement of hearing with antiviral tx
- Test for CMV if initial hearing screen not passed. This testing is mandated in some states.
Hearing Screening Myths
Misconception vs Clinical Fact

• Parents can tell if their child has a hearing loss by age 2-3 months

• Parents/physicians can test for hearing loss by clapping hands or banging pots near the baby

• Hearing loss risk factor assessments will identify all children with hearing loss

• Hearing loss if rare, so newborn hearing screening is not necessary

• There is no rush to identify hearing loss

• Before NB hearing screening hearing loss not recognized until 2-3 years of age; age 4 if milder

• Babies who are deaf/HH can still startle to loud noises but may not be able to hear all the sounds important for speech

• 50% of infants who are deaf/HH have no known risk factors

• Hearing loss affects 1-3 per 1000 births—the most common condition

• Identification before 6 mo. can avoid speech and language delays through evidence-based early intervention
Hearing Screening Myths
Misconception vs Clinical Fact

- Children younger than 12 months cannot be fitted with hearing aids
- Newborns can be fit with amplification

- Babies need to be sedated to complete ABR testing
- Babies younger than 3 months can typically be tested without need for sedation

- Fluid prevents completion of diagnostic ABR
- Underlying sensory loss can and should be ruled out as soon as possible through use of bone conduction ABR stimuli

- Abnormal OAEs along with a flat tympanogram (normal volume) confirms a conductive hearing loss
- Diagnostic ABR including bone conduction testing is needed in combinations with OAE and tympanograms for a complete diagnosis of type and degree of hearing loss in each ear.
The Role of Medical Home

- Do listen to parents' concerns
- Encourage prompt follow-up with rescreens and diagnostic evaluations
  - Make sure diagnostic evaluations are done by an audiologist who has experience with infants
- Set up electronic medical record (EMR) system to include results of auditory screening
- Flag all patient charts for children that require follow-up for hearing screens
- Flag all patient charts for children that are at risk for late onset hearing loss
Infants identified as deaf or hard of hearing

- Address the family’s concerns
- Ensure the family is seeing an experienced pediatric audiologist
- Refer the family to appropriate specialists: Otolaryngology, Genetics, Ophthalmology
- Help the family obtain early intervention services
- Monitor developmental milestones and ear infections
Resources:

• Early Intervention
  • SDSD (South Dakota School for the Deaf)
  • Birth to 3 Program

• Parent
  • www.handsandvoices.org
  • www.beginningssvsc.com
  • www.babyhearing.org
  • http://www.babyhearing.org/

• Physician
  • www.aap.org
  • www.medicalhomeinfo.org
  • http://www.infanthearing.org/
  • http://www.pedialink.org
  • http://www.cdc.gov/ncbddd/dd/ddhi.htm
Early Hearing Detection and Intervention (EHDI) Guidelines for Pediatric Medical Home Providers

**Newborn Screening Birth**
- Identify a Medical Home for every infant
- Hospital-based Inpatient Screening (OAE/ABR)** (only ABR or OAE if NICU 5+ days)
- All results sent to Medical Home

**Screening Completed Before 1 Month**
- Home Birth**
  - No more than screening before 3rd day of life
  - Passed
- Outpatient Re-Screening** (OAE/ABR)**
  - All results sent to Medical Home and State EHDI Program
  - Passed
- Failed Screen, or Missed, or Incomplete

**Diagnostic Evaluation Before 3 Months**
- Pediatric Audiologic Evaluation** with Capacity to Perform:
  - OAE**
  - ABR**
  - Frequency-specific tone bursts
  - Air & bone conduction
  - Sedation capability (only needed for some infants)
- Normal Hearing
- Hearing Loss
  - Unilateral/Interaural Sensomotor/Conductive/Mixed/Mild/Moderate/Severe/Profound

**Audiolist Reports to State EHDI Program**
- Every child with a permanent hearing loss, as well as all normal follow-up results
- Refer to IDEA** Part C

**Intervention Services Before 6 Months**
- Coordinating agency for early intervention
- Team Advised Family About:
  - All communication options; different communication modes; assistive listening devices (hearing aids, cochlear implants, etc.); parent support programs
  - Medical & Otologic Evaluations
  - To recommend treatment and provide clearance for hearing aid fitting
  - Pediatric Audiology
    - Hearing aid fitting and monitoring
  - Ongoing monitoring

**Ongoing Care of All Infants**: Coordinated by the Medical Home Provider
- Provide parents with information about hearing, speech, and language milestones
- Identify and aggressively treat middle ear disease
- Provide vision screening (and referral when indicated) as recommended in the AAP “Bright Futures Guidelines, 3rd Ed.”
- Provide ongoing developmental screening (and referral when indicated) per the AAP “Bright Futures Guidelines, 3rd Ed.”
- Refer promptly for audiology evaluation when there is any parental concern regarding hearing, speech, or language development
- Refer for audiology evaluation (at least once before age 30 months) infants who have any risk indicators for later-onset hearing loss:
  - Family history of permanent childhood hearing loss
  - Neonatal intensive care unit stay of more than 5 days duration, or any of the following (regardless of length of stay):
    - ECMO, mechanically-assisted ventilation, other toxic medications or loop diuretics, exchange transfusion for hyperbilirubinemia
    - In utero infections such as cytomegalovirus, herpes, rubella, syphilis, and toxoplasmosis
    - Postnatal infections associated with hearing loss, including bacterial and viral meningitis
    - Cranial anomalies, particularly those that involve the pinna, ear canal, ear tags, ear pits, and temporal bone anomalies
    - Findings suggestive of a syndrome associated with hearing loss (Waardenburg, Apert, Jervell and Lange-Nielsen, Pendred)
    - Syndromes associated with progressive or delayed-onset hearing loss (neurofibromatosis, osteopetrosis, Usher Syndrome)
    - Neurodegenerative disorders (such as Hunter Syndrome) or sensory motor neuropathies (such as Friedreich's ataxia and Charcot-Marie-Tooth disease)
- Head trauma, especially basal skull/temporal bone fracture that requires hospitalization
- Chemotherapy

**Notes**
(a) In screening programs that do not provide Outpatient Screening, Infants will be referred directly from inpatient screening to Pediatric Audiologic Evaluation. Likewise, Infants at higher risk for hearing loss for follow-up also may be referred directly to Pediatric Audiology.
(b) Part C of IDEA may provide diagnostic audiology evaluation services as part of Child Find activities.
(c) Even Infants who fail screening in only one ear should be referred for further testing of both ears.
(d) Includes Infants whose parents refused initial or follow-up hearing screening.

February 2010 - American Academy of Pediatrics Task Force for Improving Newborn Hearing Screening, Diagnosis and Intervention (www.medicalhomeinfo.org)
1. Identify the correct guidelines for hearing detection and intervention

   a. Screen hearing no later than 1 month of age, repeat hearing screen by 3 months of age, referral to ENT no later than 6 months of age

   b. Screen hearing no later than 1 month of age, referral to ENT no later than 3 months of age, enrollment in early intervention if speech delayed

   c. Screen hearing before hospital discharge, rescreen in the office since middle ear fluid likely, refer to ENT if infant doesn't pass 3 screens

   d. Screen hearing no later than 1 month of age, diagnostic evaluation for hearing loss no later than 3 months of age, enrollment in early intervention no later than 6 months of age
Correct Answer:

D. Screen hearing no later than 1 month of age, diagnosis of hearing loss no later than 3 months of age, enrollment in early intervention no later than 6 months of age
2. Which of the following statements is incorrect:

   a. Infants admitted to the NICU for more than 5 days should have an ABR for the hearing screen

   b. It is not necessary to rescreen hearing if only 1 ear did not pass the initial screening

   c. At least 1 ABR test is recommended as part of a complete audiology evaluation to confirm hearing loss

   d. Early intervention services should begin as soon as possible after diagnosis of hearing loss but at no later than 6 months of age.
Answer:

B. It is not necessary to rescreen hearing if only 1 ear did not pass the initial screening
3. If an infant receives a refer on their newborn hearing screening and does not pass an outpatient rescreen what is the next step:

   a. Refer to ENT

   b. Repeat the hearing screen again

   c. Refer to a pediatric audiologist for a full audiologic evaluation

   d. Check to see if the infant startles to loud noises or clapping.
Correct Answer:

C. Refer to a pediatric audiologist for a full audiologic evaluation
4. What is/are additional recommended evaluations for infants identified with a hearing loss?
   a. CMV testing in the first month
   b. Ophthalmology
   c. Refer to genetics-complete family history, evaluate for syndrome, genetic testing
   d. All of the above
Correct Answer:

D. All of the above.
5. Which statement is NOT correct:

- a. If the family is concerned about hearing or communication a repeat hearing test is recommended

- b. Regular audiologic monitoring is recommended when there has been an NICU stay > 5 days or there is a history of a congenital infection, ECHMO, chemotherapy, or a family history of childhood hearing loss

- c. Children younger than 12 months cannot be fitted with hearing aids

- d. It is the primary physician's responsibility to ensure that infants who do not pass the hearing screen receive a diagnostic evaluation by an audiologist, are referred for intervention, and report this to the state EHDI program
Answer:

C. Children younger than 12 months cannot be fitted with hearing aids