Hearing Loss: Newborn Screening

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Key Points

- Identification of hearing loss in the newborn period, followed by intervention, is considered critical for language development in hearing-impaired children.
- Approximately half of congenital hearing loss is believed to have a genetic cause; 30% is associated with a hereditary syndrome while 70% is nonsyndromic (isolated).
- Newborn hearing screening, unlike other testing, is often not performed in conjunction with an informed consent process.

Learning Objectives

Participants will be able to:

- Explain the rationale for implementing newborn hearing screening programs and describe how such screening is conducted;
- Determine the recurrence risk for a couple that has had a child with isolated, nonsyndromic deafness and understand how genetic testing may further clarify the risk;
- Appreciate the range of different hereditary deafness syndromes, including their inheritance patterns and their associated findings.

Family History Issues

Approximately 90% of children with deafness are born to hearing parents. Seventy-five to eighty-five percent of hereditary nonsyndromic deafness is inherited in an autosomal recessive manner.

Red Flags 

Due to universal newborn hearing screening, most babies with congenital hearing loss are identified shortly after birth. However, symptoms of possible deafness in the first year of life include: a 0- to 3-month-old baby who does

not jump at a sudden, loud noise; a 3- to 6-month-old baby who does not turn its head in response to voice or make sounds; a 6- to 10-month-old baby who does not react to hearing its name or does not recognize easy words like "no;" a 10- to 15-month-old baby who does not repeat sounds or words.

Case 10. A Newborn Boy with a Failed Newborn Hearing Screen

The resident is seeing a newborn, Scott, and his parents, Mr. and Mrs. W. Their three-year-old daughter has received well-child care in your office since she was born. The W's tell the resident that Scott was referred for a follow-up hearing screen and a consultation with an audiologist after his hearing test indicated hearing loss. They want to know what the test was, and "who ordered it in the first place?" They also want to know what will happen as a result of the referral. Mr. and Mrs. W report no family history of hearing loss.

Clinical Care Issues

Rationale for newborn hearing screening

Scott's hearing test was performed as part of routine newborn hearing screening. Such screening is done to detect hearing loss in early infancy, in order to ensure appropriate interventions for language development. Moderate to profound hearing loss in early infancy has been shown to be associated with impaired language development, as auditory stimuli during this period are critical to development of speech and language skills. If newborn hearing loss detected on screening is confirmed by definitive diagnosis, then both general therapy and specific treatment based on the etiology of the hearing loss (conductive, sensorineural, or mixed) can be instituted.

Newborn hearing programs are implemented at the state level with funding from federal EHDI (Early Hearing Detection and Intervention) programs as well as state and private funding. These programs identify two to three per 1000 babies with moderate to profound bilateral hearing loss. By 2004, approximately 90% of newborns in the U.S. were being screened for hearing loss, with some states performing at higher levels than others, according to

the National Center for Hearing Assessment and Management.

The American Academy of Pediatrics (AAP), the American Academy of Audiology, and Directors of Speech and Hearing Programs in State Health and Welfare Agencies recommend universal newborn hearing screening. A 1993 National Institutes of Health consensus conference recommending that all infants be screened before hospital discharge and a 2000 Joint Committee on Infant Hearing report also endorsed EDHI programs in newborn screening. The rationale for screening is based on the availability of low-cost and effective screening tests, and evidence that undetected hearing loss is associated with delay in speech, language, and cognitive development. The Joint Committee on Infant Hearing notes that early intervention for hearing-impaired infants will maximize their communication skills and avoid delays in language, cognition, and social development that might otherwise impair adult educational attainment and employment [Joint Committee on Infant Hearing Year 2000 Position Statement]. The AAP specifies that screening programs should detect hearing loss in affected infants by three months of age, and institute appropriate interventions by six months of age. Appropriate follow-up requires coordination between the birthing hospital where screening occurs, the primary care physician, appropriate specialists, and the family (AAP Policy Statement).

The US Preventive Services Task Force found insufficient evidence to recommend for or against routine screening of newborns for hearing loss (USPSTF Guideline: Newborn hearing screening). This recommendation was based on the Task Force's conclusion that it could not determine from current studies whether the benefits of earlier identification and intervention outweighed the potential harms of the high rate of false-positive test results expected in a universal screening program. The Task Force noted that both the yield and the proportion of true-positive test results would be higher if screening were targeted to high-risk infants, including those with neonatal intensive care unit admission for two days or more, syndromes known to include hearing loss, family history of childhood sensorineural hearing loss, congenital infections, and craniofacial abnormalities, especially those involving the ear. A comparison of the guidelines issued by the AAP, the United States Preventive Services Task Force, and the Joint Committee on Infant Hearing is available from the National Guideline Clearinghouse.

Evaluation of the infant

Two types of tests are commonly used for newborn hearing screening: otoacoustic emissions (OAEs) and auditory brainstem response (ABR). More

information about these test methods is available at AAP and Agency for Healthcare Research and Quality.

Infants who are identified as having possible hearing loss on initial newborn screening tests are referred for repeat testing; positive second-stage results are usually validated by a combination of otolaryngologic and audiologic consultation, diagnostic ABR testing, or other electrophysiologic testing that can be performed as early as age three months. Visual reinforcement audiometry cannot be performed reliably before age eight to nine months.

If hearing loss is confirmed, experts recommend that the infant be referred for an evaluation by a multidisciplinary team experienced in neonatal deafness. Hearing is considered normal if an individual's thresholds are within 15 decibels (dB) of normal thresholds. Hearing loss is graded as mild (26-40 dB), moderate (41-55 dB), moderately severe (56-70 dB), severe (71-90dB), and profound (90dB). Hearing loss is categorized by the portion of the hearing system affected (conductive, sensorineural, or mixed); the time period of speech development at which it occurred (prelingual or postlingual); whether it is primarily due to a genetic cause or an environmental one (hereditary or acquired); and if genetic, whether it is a component of a genetic syndrome or nonsyndromic.

Physical examination focuses on findings that may be associated with syndromic forms of hearing loss, including the presence of pigmentary abnormalities (Waardenburg syndrome), structural ear changes such as ear tags or pits (branchiootorenal syndrome), retinitis pigmentosa (Usher syndrome), thyroid enlargement (Pendred syndrome), cardiac conduction defects (Jervell and Lange-Nielsen syndrome), and pigmentary abnormalities (Waardenburg syndrome). See *GeneReview*: Hereditary Hearing Loss and Deafness for information about genetic syndromes associated with hearing loss.

Risk Assessment

Neonates are in a higher-risk group for sensorineural hearing impairment if they have one of the following factors: family history of hearing impairment; congenital infections such as CMV (cytomegalovirus, the most common cause of congenital, non-hereditary hearing loss); ototoxic drug exposure; prematurity; congenital malformations of the head and neck; trauma; or other factors that have led to admission to an intensive care nursery.

Genetic Counseling and Testing

Figure 1. Causes of Prelingual Deafness in Children

Seventy-five to eighty-five percent of hereditary nonsyndromic deafness is inherited in an autosomal recessive manner. Of these, 50% are due to DFNB1. DFNB1 is characterized by congenital, nonprogressive mild-to-profound sensorineural hearing impairment. No other associated medical findings are present (see Figure 1). DFNB1 is diagnosed by molecular genetic testing of the *GJB2* gene (which codes for the protein connexin 26) and the *GJB6* gene (which encodes the protein connexin 30). DFNB1 related hearing loss has an approximate prevalence in the general population of 14/100,000 (~1/7,000).

Full sequencing of the *GJB2* gene detects two identifiable mutations in about 98% of individuals with DFNB1. The most common mutation, 35delG, is found in over two-thirds of persons with DFNB1, but at least 80 other disease-causing mutations have been identified. The remaining 2% of individuals with DFNB1 have one identifiable *GJB2* mutation and a large deletion which includes a portion of *GJB6* — i.e., they are double heterozygotes. (See *GeneReview*: DFNB1 for further information.)

For parents of a child with isolated, nonsyndromic hearing loss, empiric estimates are available to determine their risk of having another deaf child. If Scott is deaf, Mr. and Mrs. W have an 18% empiric probability that future children will be deaf. If Scott is tested and does not have DFNB1, the recurrence risk for deafness is 14%. If Scott does have DFNB1, the risk that Scott's sibs will be deaf is 25%. If the couple is consanguineous or comes from an inbred community, their subsequent offspring have close to a 25% probability of deafness due to the high likelihood of an autosomal recessive disorder.

Interventions

Clinical management. For all types of hearing loss, early intervention with speech therapy and amplification are considered key components. The most common type of hearing loss found in neonates is sensorineural. Treatment depends on the severity of the loss. Amplification through hearing aids is used in the majority of cases; cochlear implantation is a possibility for severe to profoundly deaf children. Nonrandomized, prospective studies have demonstrated superior communication performance in children with prelingual deafness who received cochlear implants as compared to children

with similar hearing impairment using more traditional tactile or acoustic hearing aids. Language development can also be fostered in profoundly deaf children through American Sign Language (ASL).

Ethical/Legal/Social/Cultural Issues

Factors influencing the effectiveness of newborn screening. There has been insufficient study of factors that may influence outcomes of testing such as patient characteristics (e.g., socioeconomic status, level and laterality of hearing loss, the presence of co-morbidity, or developmental delay), family characteristics, and the presence and nature of other therapeutic interventions. Additionally, there may not be provisions such as financial support to guarantee access to the suggested interventions for all newborns identified with hearing loss; the interventions can be effective only if they can be implemented.

Deaf culture. Many members of the Deaf community suggest there is a bias in the medical profession that views deafness as a disability or as needing medical intervention. Rather, the Deaf community views deafness as a separate and valued culture in which members are bilingual (communicating in both ASL and English). While this perspective may be more common in parents who are deaf, the view may be held by hearing parents as well. As a result, the decision about how to proceed with the evaluation and potential "treatment" of deafness is a personal family matter that may be influenced by values and perceptions about deafness.

Informed decision making. Since Scott's parents were not aware that Scott would undergo hearing screening, they will now need a more detailed discussion of the process and what the next steps will be in his workup. Some helpful brochures available from the National Institute on Deafness and Other Communication Disorders include Has Your Baby's Hearing Been Screened? and What to Do if Your Baby's Screening Reveals a Possible Hearing Problem.

Resources

- **American Academy of Pediatrics**

- **Boystown National Research Hospital: Resources for Parents**

- **American Society for Deaf Children**
  PO Box 3355
  Gettysburg, PA 17325
  Phone: 717-334-7922 (business V/TTY); 800-942-ASDC (parent hotline)
  Fax: 717-334-8808
  Email: ASDC1@aol.com

- **National Association of the Deaf**
  814 Thayer
  Silver Spring, MD 20910
  Phone: 301-587-1788 (voice); 301-587-1789 (TTY)
  Fax: 301-587-1791
  Email: NADinfo@nad.org

- **The Morton Hearing Research Group**

- **NCBI Genes and Disease Webpage: Deafness**

- **Hereditary Hearing Loss Homepage**

- **World Council on Hearing Health**

- **CDC Early Hearing Detection and Intervention Program (EHDI)**

- **National Center for Hearing Assessment and Management**

- **American Speech-Language-Hearing Association**

- **National Institute On Deafness and Other Communication Disorders**

- **Harvard Medical School Center for Hereditary Deafness brochure:**
  Understanding the Genetics of Deafness: A Guide for Patients and Families

- **National Library of Medicine Genetics Home Reference**

Nonsyndromic Deafness

- **GeneTests Resources for DFNB1**
- **GeneTests Online Medical Genetics Information Resource**

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