

**EARN CATEGORY I CME CREDIT** by reading this article and the article beginning on page 48 and successfully completing the posttest on page 54. Successful completion is defined as a cumulative score of at least 70% correct. This material has been reviewed and is approved for 1 hour of clinical Category I (Preapproved) CME credit by the AAPA. The term of approval is for 1 year from the publication date of June 2008.

- Define the audiologic terms used to describe congenital hearing loss (CHL) in infants
- Discuss the incidence, etiology, and medical evaluation of CHL
- Describe the components of a language acquisition program
- Understand the role of assistive technology for children with CHL

## Come hear, baby! Identifying and managing congenital hearing loss

Thanks to universal hearing screening for newborns and improved assistive technology, hearing-impaired children born today have a vastly better prognosis than did those in previous generations.

**Genevieve DelRosario, MHS, PA-C**

**H**elen Keller is believed to have said that whereas blindness cuts you off from things, deafness cuts you off from people. She later explained that of the two disabilities, she would prefer to be blind.<sup>1</sup> Indeed, the impact of congenital hearing loss (CHL) may profoundly affect almost all aspects of the lives of deaf and hard-of-hearing persons. In 2003, the *Journal of Pediatrics* summarized existing research with the following statement: “Congenital or acquired hearing loss in infants and children has been linked with lifelong deficits in speech and language acquisition, poor academic performance, personal-social maladjustments, and emotional difficulties.”<sup>2</sup>

With the advent of mandatory newborn hearing screening in 1999, far superior outcomes can be expected for hearing-impaired children born today as compared with those seen in past decades. This article reviews the rationale for developing a national newborn screening program; current practice, incidence, and etiology of CHL; the history and current status of the national newborn hearing screening program; and the most advanced treatment and communication options for children with diagnosed hearing loss.

### DEFINITION OF TERMS

As a first step, a review of audiologic terms may be useful. First, hearing is measured by the use of an *audiogram*. This test measures both the *degree* of hearing loss (how loud something is) as well as the *audible frequency* (how high or low the pitch). The unit of loudness is *decibels* (dB). Zero dB represents a very soft sound, such as the rustle of leaves on a tree. At the other end of the spectrum, 120 dB is a very loud sound, such as the sound of an airplane passing directly



Otoacoustic emissions testing on a newborn

© James King-Holmes / Photo Researchers, Inc.

overhead. *Pitch* is measured in cycles per second (hertz; Hz), with fewer cycles (eg, 250 Hz) producing a deeper pitch and more cycles (eg, 4,000 Hz) producing a higher pitch. See Figure 1 (page 24) for an example of an audiogram.

Clinicians should also be aware of the different types of hearing loss: *conductive*, *sensorineural*, *mixed*, and *central*. *Conductive* hearing loss, caused by pathology of the tympanic membrane or middle ear, is most commonly due to fluid but may also be caused by pathology affecting the ossicles. *Sensorineural* hearing loss typically refers to hearing loss caused by damage to the hair cells of the inner ear. *Sensorineural* hearing loss may also refer to auditory neuropathy, a relatively rare form of hearing loss caused by an abnormality of the 8th (auditory) cranial nerve. *Mixed* hearing loss refers to combined sensorineural and conductive deficits. Finally, *central* hearing loss denotes damage to the upper auditory pathways. This type is a relatively rare form of hearing loss. This review will focus primarily on permanent sensorineural hearing loss.

#### NEWBORN SCREENING: HISTORY AND RATIONALE

The rationale behind the newborn hearing screening program was very simple: children with hearing loss were being identified very late. In 1988, the average age at diagnosis for children with severe to profound hearing loss was 2½ years,<sup>3</sup> and children with milder forms of hearing loss were often not identified until formal audiologic testing was done in their school years. This late diagnosis was a source of frustration and guilt for families, but more important, it meant that many children had already passed through the so-called “critical period” for language development before their hearing loss was diagnosed. The neuroplasticity of the brain in the first 3 years of life allows for optimal language development during that time. This plasticity is greatest at younger ages and slowly decreases as we age. If auditory pathways are not utilized, the brain will “rewire” itself to receive greater input from the other senses, primarily vision.<sup>4</sup>

Despite recommendations as early as 1965 to screen for CHL, major movement did not occur until the 1990s. In 1994, the Joint Commission on Infant Hearing (JCIH) recommended identification of infants with CHL before 3 months of age and early intervention for those infants iden-

“The most common cause of congenital hearing loss in infants and children is genetic, causing more than 50% of cases.”

tified.<sup>5</sup> This guideline was endorsed by the American Academy of Pediatrics (AAP) in 1999.<sup>6</sup>

Today, newborn hearing screening is tracked and aided by the Early Hearing Loss Detection and Intervention Program, a program funded by the CDC. The primary program goals, based on the JCIH Year 2000 position statement, are as follows:<sup>7</sup>

- All newborns will be screened for hearing loss before 1 month of age, preferably before hospital discharge.
- All infants whose screening result is positive will have a diagnostic audiologic evaluation before 3 months of age.
- All infants identified with a hearing loss will receive appropriate early intervention services before 6 months of age.<sup>8</sup>

These three goals comprise the national “one–three–six” campaign that is used by programs nationwide. Overall, more than 90% of all infants receive newborn hearing screening.<sup>9</sup>

#### INCIDENCE OF CONGENITAL HEARING LOSS

Hearing loss is one of the most common congenital anomalies. Approximately 1 to 3 per 1,000 infants in the well-baby population have permanent hearing loss, and this figure is as high as 2 to 4 infants per 100 in the neonatal intensive care unit population.<sup>6</sup> In fact, severe to profound hearing loss is more prevalent than birth defects such as phenylketonuria, sickle cell anemia, and congenital hypothyroidism.<sup>10</sup>

The severity and laterality of CHL varies dramatically. Data available for 2004 show that 31% of infants with classified CHL had a unilateral loss.<sup>11</sup> For persons with either bilateral or unilateral loss, the degree of hearing loss (mild, moderate, severe, or profound) varies dramatically.<sup>11,12</sup> PAs must realize, however, that a child born with mild to moderate hearing loss is at great risk for progressive hearing loss as well.<sup>13</sup>

Continued on page 24

#### KEY POINTS

- Hearing loss in children is associated with deficits in speech and language acquisition, poor academic performance, personal-social maladjustments, and emotional difficulties.
- The advent of mandatory newborn hearing screening in 1999 has greatly improved outcomes for hearing-impaired children.
- Hearing loss is one of the most common congenital anomalies. Approximately 1 to 3 per 1,000 infants in the well-baby population have permanent hearing loss, and the figure is as high as 2 to 4 infants per 100 in the neonatal intensive care unit population.
- Once the diagnosis of hearing loss has been made, the child should begin a language acquisition program as soon as possible.
- Cochlear implants offer greater access to speech and language. In the past decade, they have been refined and used more widely.

#### COMPETENCIES

●●● Medical knowledge

● Interpersonal & communication skills

●●● Patient care

●●● Professionalism

● Practice-based learning and improvement

●● Systems-based practice

### ETIOLOGY

Causes of CHL in children may be grouped into three categories: *genetic*, *environmental*, and *idiopathic*.<sup>14</sup> The most common etiology is genetic, causing more than 50% of cases. Of the cases of hearing loss with a genetic cause, approximately 30% are known to be part of a syndrome, and more than 400 syndromes are known to be associated with hearing loss.<sup>15</sup>

Nonsyndromic genetic hearing loss is most often caused by the disorder DFNB1, which is caused by mutations in the *GJB2* and *GJB6* genes. These encode the proteins connexin 26 and connexin 30, respectively, and together cause approximately 15% of all cases of congenital hearing loss. A significantly smaller percentage is caused by an autosomal dominant or x-linked mutation.<sup>14</sup>

Environmental causes of hearing loss most commonly include prematurity, hypoxia, neonatal icterus, and medications (particularly aminoglycosides). TORCH infections (toxoplasmosis, rubella, cytomegalovirus, herpes, and other infections) are known to cause hearing loss as well.<sup>14</sup>

### SCREENING, EVALUATION, AND REFERRAL

Typically, two techniques are used to provide hearing screening in newborns. The first is *otoacoustic emissions* (OAE) testing. In this noninvasive test, a series of clicks is made into the baby's ear. In children with a normal middle ear and normal hair cell function, the hair cells send back an echo response that is measured by the equipment. The second routine form of newborn hearing screening is *automated auditory brainstem response* (AABR) testing. This test measures brain-

wave response to sound. It offers greater specificity than OAEs in most circumstances but has the disadvantage of being somewhat more expensive.<sup>16</sup>

Many centers use a protocol that includes both techniques. OAE testing is performed first, and if that test indicates a hearing problem, the infant is tested again before discharge using AABR testing. This approach has significantly reduced the number of infants who have false-positive results at discharge.<sup>17</sup> When screening tests indicate that a newborn has a hearing problem, the testing should be repeated on an outpatient basis.

If both the original screening and the repeated outpatient testing indicate a problem, the infant should be referred to a pediatric audiologist, who will do a comprehensive evaluation. This will include nonautomated ABR testing (typically performed when the infant is sleeping or sedated), as well as an evaluation of middle ear function.

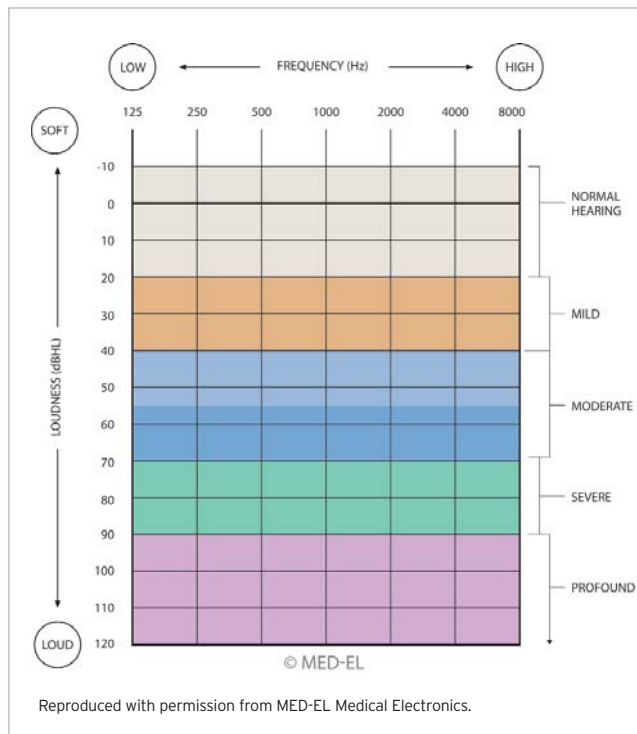
If hearing impairment is confirmed, the infant's primary care physician assistant is well placed to perform a careful history and physical examination on the infant. A careful family history is essential, not only reviewing hearing loss but also eliciting any family history of craniofacial, renal, cardiac, endocrine, or ophthalmologic dysfunction.<sup>18</sup> The infant's medical history should be reviewed with careful attention given to possible infections during pregnancy, trauma in the neonatal period, severe jaundice, medicines taken by the infant, and the use of mechanical ventilation. During the physical examination, the PA should look for signs of any possible syndromes. Subtle signs of possible syndromic involvement include abnormally tall or small stature, ear tags or pits, ophthalmologic abnormalities, and delayed disappearance of newborn reflexes.<sup>18</sup>

The PA should follow current JCIH and AAP recommendations and offer several medical referrals,<sup>7</sup> including to an otolaryngologist (ENT), a geneticist, and an ophthalmologist. Whereas an ENT referral might seem quite obvious, the need for referrals to a geneticist and ophthalmologist is very often missed. In fact, only 8.9% of clinicians in a recent study referred patients with CHL to a geneticist, and only 0.9% referred these children to an ophthalmologist.<sup>19</sup> Given the higher than typical rate of eye disease that may accompany hearing loss,<sup>20</sup> it is critical to confirm normal vision. Referrals to the local early intervention program should also be made in a timely manner. A list of resources for parents is given in Table 1.

Continued careful monitoring of the child with CHL is needed because many children, particularly those with a genetic syndrome or known viral cause of hearing loss, have other medical or developmental problems. Among the most common concomitant medical problems is vestibular dysfunction, which occurs in the majority of children with hearing loss.<sup>21</sup>

### LANGUAGE ACQUISITION AND COMMUNICATION OPTIONS

Once the diagnosis of hearing loss has been made, the child should begin a language acquisition program as soon as pos-



**FIGURE 1.** An audiogram used to record hearing test results

sible. Traditionally, sign language has been the primary mode of communication for many profoundly deaf children. However, this is no longer universally true for families of deaf infants. There are several basic communication philosophies. They include the following:

- **The use of sign language as the primary method of communication** This methodology is used especially by families who have parents or other members who are deaf or hard of hearing.
- **Auditory oral philosophy** This encourages oral communication among deaf children. This methodology supports children using their residual hearing but allows for visual cues, such as lip reading, to assist in the process.
- **Auditory verbal philosophy** Like the auditory oral philosophy, the auditory verbal philosophy helps infants and children to learn to listen and speak using their residual hearing and technological aids. It differs from the auditory oral philosophy in that it discourages the use of visual aids such as lip reading.
- **Total communication philosophy** This philosophy encourages the use of any type of communication, including the use of sign language, natural gestures, listening, lip reading, and so on.

The decision of which communication method to use is fiercely debated in the hearing loss community. To date, no one philosophy has clearly proven advantageous in all circumstances. The compassionate clinician can help parents be aware of their options and offer support in what can be a very challenging decision.

### ASSISTIVE TECHNOLOGY

Along with newborn screening, advances in supportive technology are of particular interest. Hearing aids have been used with success for decades, and infants with hearing loss are now routinely fitted with them as young as 2 months of age. Improvements in cochlear implant technology offer greater access to speech and language. Although cochlear implants have been in existence for more than 30 years, in the past decade they have been refined and used more widely.

**Cochlear implants** As sensorineural hearing loss is typically caused by damage to the hair cells of the inner ear, a cochlear implant inserts an electrode into the cochlea that bypasses the nonfunctional hair cells and directly stimulates the auditory nerve. The electrical current is then understood by the brain as sound. The electrode is connected to a magnet, which is also surgically implanted. The internal device then communicates via an external magnet with an external, programmable processor that is worn much like a behind-the-ear hearing aid. The newest generation of processors may be programmed for quiet or noisy situations or for directional listening.

Cochlear implants are now used by many deaf persons worldwide. In the United States, they are approved by the FDA for implantation as young as 12 months of age. However, centers are beginning to provide implants to children at even younger ages to obtain maximum impact and listening capability when neuroplasticity is greatest. These children

**TABLE 1. Internet resources for families of children who are deaf/hard of hearing**

• Alexander Graham Bell Association for the Deaf and Hard of Hearing <a href="http://www.agbell.org">www.agbell.org</a>
• Early Hearing Detection & Intervention (EHDI) Program <a href="http://www.cdc.gov/ncbddd/ehdi/">www.cdc.gov/ncbddd/ehdi/</a>
• Hands and Voices <a href="http://www.handsandvoices.org/">www.handsandvoices.org/</a>
• My Baby's Hearing <a href="http://www.babyhearing.org/">www.babyhearing.org/</a>
• National Association of the Deaf <a href="http://www.nad.org">www.nad.org</a>

often develop listening and speech communication skills that are on par with their hearing peers.<sup>22</sup>

**Controversy** Cochlear implants are not without controversy. When they first were introduced, many in the Deaf community argued that they would deprive deaf persons of their culture.<sup>23</sup> Although much of the opposition to cochlear implants has since subsided, there are still concerns and cautions surrounding implantation. Risks include facial nerve paralysis, vestibular dysfunction, and possibly an increased risk of meningitis. Cost and insurance coverage may also be a limiting factor.

PAs should understand that cochlear implants are unable to offer normal hearing and that at best, hearing remains consistent with mild loss. Finally, even in optimal circumstances, children (or adults) with implants will need years of speech therapy to learn to hear, listen, and speak clearly. Nonetheless, the overall results are very encouraging: more than 90% of children who receive implants by the age of 2 years attain intelligible speech, as compared to only 20% of profoundly deaf children who wear hearing aids alone.<sup>4</sup>

### FUTURE DIRECTIONS

Many changes are occurring in the field of early hearing loss detection and intervention. At present, the concept of bilateral cochlear implantation is beginning to gain global acceptance. In 2005, an international consensus statement formally recommended bilateral cochlear implantation for all children with permanent profound bilateral sensorineural hearing loss. Benefits cited by the consensus group include better understanding of speech, sound localization, more natural hearing, and an improved quality of life.<sup>24</sup>

Hair cell regeneration is another exciting avenue of research. Although scientists have been working on this concept for decades, promising new research is emerging. Both gene therapy and pluripotent stem cell therapy are active avenues of research that may eventually offer normal hearing to those with congenital or acquired hearing loss.<sup>25</sup>

The possibilities for a child born with hearing loss in 2008 are significantly improved from even 10 years ago. By stay-

ing current with advances in this quickly changing field and working together with a team of medical professionals, the PA can make a tremendous difference in the lives of children with CHL. **JAAPA**

**Genevieve DeRosario** is an assistant professor in the Department of Physician Assistant Education, Saint Louis University, St. Louis, Missouri. She practiced in the Department of Pediatrics, University of Kansas Medical Center, Kansas City, when she wrote this article. The author has indicated no relationships to disclose relating to the content of this article.

#### REFERENCES

1. Taking childhood hearing loss seriously [editorial]. *Lancet*. 2007;369(9569):1234.
2. Cunningham MC, Cox EO; Committee on Practice and Ambulatory Medicine and Section on Otolaryngology and Bronchoesophagology. Hearing assessment in infants and children: recommendations beyond neonatal screening. *Pediatrics*. 2003;111(2):436-440.
3. Commission on Education of the Deaf. *Toward Equality: Education of the Deaf. A Report to the President and the Congress of the United States*. Washington, DC: US Government Printing Office; 1988.
4. Flexer C. The listening brain: research to practice. National Early Hearing Detection and Intervention Conference. February 1, 2006. Video available from <http://www.infantheating.org/meeting/ehdi2006/presentations/index.html>. Accessed May 13, 2008.
5. Joint Committee on Infant Hearing 1994 Position Statement. *Pediatrics*. 1995;95(1):152-156.
6. Task Force on Newborn and Infant Hearing, American Academy of Pediatrics. Newborn and infant hearing loss: detection and intervention. *Pediatrics*. 1999;103(2):527-530.
7. Joint Committee on Infant Hearing, American Academy of Audiology, American Academy of Pediatrics, et al. Year 2000 position statement: principles and guidelines for early hearing detection and intervention programs. *Pediatrics*. 2000;106(4):798-817.
8. Early Hearing Detection & Intervention (EHDI) Program. National EHDI goals. Centers for Disease Control and Prevention Web site. <http://www.cdc.gov/ncbddd/ehdi/nationalgoals.htm>. Accessed May 13, 2008.
9. Preliminary summary of 2005 national EHDI data (version 5). Centers for Disease Control and Prevention Web site. [http://www.cdc.gov/ncbddd/ehdi/documents/Nat\\_Summ\\_2005\\_Web\\_V5.pdf](http://www.cdc.gov/ncbddd/ehdi/documents/Nat_Summ_2005_Web_V5.pdf). Updated August 10, 2007. Accessed May 13, 2008.
10. Jacobson J, Jacobson C. Evaluation of hearing loss in infants and young children. *Pediatr Ann*. 2004;33(12):811-822.
11. Estimated type and degree for cases of unilateral hearing loss 2004. Version B. Centers for Disease Control and Prevention Web site. [http://www.cdc.gov/ncbddd/ehdi/2004/Type\\_2004\\_Unilat\\_A\\_web.pdf](http://www.cdc.gov/ncbddd/ehdi/2004/Type_2004_Unilat_A_web.pdf). Accessed May 13, 2008.
12. Estimated type and degree for cases of bilateral hearing loss 2004. Version B. Centers for Disease Control and Prevention Web site. [http://www.cdc.gov/ncbddd/ehdi/2004/Type\\_2004\\_Bilat\\_A\\_web.pdf](http://www.cdc.gov/ncbddd/ehdi/2004/Type_2004_Bilat_A_web.pdf). Accessed May 13, 2008.
13. Berrettini S, Ravecca F, Sellari-Franceschini S, et al. Progressive sensorineural hearing loss in childhood. *Pediatr Neurol*. 1999;20(2):130-136.
14. Sanford B, Weber PC. Etiology of hearing impairment in children. UpToDate Web site. <http://www.uptodateonline.com/utd/content/topic.do?topicKey=ped Iryn9030&view=text>. Updated August 11, 2005. Accessed May 29, 2006.
15. Smith RJH, Van Camp G. Deafness and hereditary hearing loss overview. Gene Clinics Web site. <http://www.geneclinics.org/profiles/deafness-overview/details.html>. Updated January 30, 2007. Accessed May 13, 2008.
16. Early identification of hearing impairment in infants and young children. National Institutes of Health Consensus Development Conference Statement. March 1-3, 1993. *NIH Consens Statement*. 1993;11(1):1-24.
17. Gravel JS, White KR, Johnson JL, et al. A multisite study to examine the efficacy of the otoacoustic emission/automated auditory brainstem response newborn hearing screening protocol: recommendations for policy, practice, and research. *Am J Audiol*. December 2005;14:S217-S228.
18. Doyle KJ, Ray RM. The otolaryngologist's role in management of hearing loss in infancy and childhood. *Ment Retard Dev Disabil Res Rev*. 2003;9(2):94-102.
19. Moeller MP, White KR, Shisler L. Primary care physicians' knowledge, attitudes, and practices related to newborn hearing screening. *Pediatrics*. 2006;118(4):1357-1370.
20. Nikolopoulos TP, Lioumi D, Stamatakis S, et al. Evidence-based overview of ophthalmic disorders in deaf children: a literature update. *Otol Neurotol*. 2006;27(suppl 1):S1-S24.
21. Horak FB, Shumway-Cook A, Crowe TK, Black FO. Vestibular function and motor proficiency of children with impaired hearing, or with learning disability and motor impairments. *Dev Med Child Neurol*. 1988;30(1):64-79.
22. Waltzman SB, Roland JT. Cochlear implantation in children younger than 12 months. *Pediatrics*. 2005;116(4):e487-e493.
23. Copeland BJ, Pillsbury HC III. Cochlear implantation for the treatment of deafness. *Annu Rev Med*. 2004;55:157-167.
24. Offeciers E, Morera C, Muller J, et al. International consensus on bilateral cochlear implants and bimodal stimulation. *Acta Otolaryngol*. 2005;125(9):918-919.
25. Murugasu E. Recent advances in the treatment of sensorineural deafness. *Ann Acad Med Singapore*. 2005;34(4):313-321.