CONGENITAL HEARING LOSS

Congenital hearing loss, for the purposes of this fact sheet, is defined as permanent and is bilateral or unilateral, is sensory or conductive, and averages 30 dB or more in the frequency region important for speech recognition. Congenital hearing loss has many etiologies, with at least half associated with genetic risk factors. Congenital nonsyndromic hearing loss is usually categorized by mode of inheritance—autosomal recessive, autosomal dominant, X-linked, or mitochondrial.[33–35]

Newborn hearing screening programs became possible after the development of hearing screening technologies. Although most states have begun screening for congenital hearing loss, the integration of these programs with ongoing screening and early intervention programs remains a challenge.[36]

Prevalence

Estimates of the prevalence of moderate-to-profound bilateral hearing loss vary, depending on the criteria used to define the different degrees of hearing loss and the characteristics of the studied population.[37] The prevalence of congenital hearing loss also depends on race, birth weight, and other risk factors.[38] Profound and permanent congenital hearing loss is estimated to occur in approximately 1 in 1000 births.[39,40]

Clinical Manifestations

The spectrum of congenital hearing loss ranges from mild to profound hearing loss. In syndromic hearing loss, the auditory pathology may be conductive and/or sensorineural, unilateral or bilateral, symmetrical or asymmetrical, and progressive or stable. The auditory pathology of nonsyndromic hearing impairment is usually sensorineural.[41,42]

Pathophysiology

Approximately half of the cases of congenital hearing loss are thought to be attributable to environmental factors (acoustic trauma, ototoxic drug exposure [aminoglycosides], bacterial or viral infections such as rubella or cytomegalovirus).[39,41,42] The remaining cases are attributable to genetic mutations. Although these cases may seem to be part of a recognizable syndrome, approximately 70% are nonsyndromic (the deafness is not associated with other clinical findings that define a recognized syndrome) and, therefore, clinically undetectable at birth. In the remaining 30%, 1 of more than 400 forms of syndromic deafness can be diagnosed because of associated clinical findings.[39,42]

Inheritance

Approximately 77% of congenital nonsyndromic hearing impairment is autosomal recessive, 22% is autosomal dominant, and 1% is X-linked. As a general rule, individuals with autosomal recessive congenital nonsyndromic hearing impairment have profound prelingual deafness, and dominant mutations lead to a more variable phenotype. More than 90% of children with congenital profound autosomal recessive congenital nonsyndromic hearing impairment are born to parents with normal hearing, and the remaining 10% or less are born to deaf parents.[41]

There has been significant progress in identifying and sequencing autosomal dominant, autosomal recessive, and sex-linked genes for deafness.[41,43] However, it is clear that more genes and mutations await discovery. This knowledge may lead to mutation-specific therapies that can delay or prevent certain forms of genetic deafness, such as the avoidance of aminoglycoside therapy in those with specific mitochondrial mutations.

Benefits of Newborn Screening

The goals of newborn screening are to identify those infants with hearing loss early for prompt intervention to diminish the morbidity associated with congenital hearing loss. Left undetected and untreated, hearing impairment can affect speech and many other cognitive abilities. For children without risk factors, hearing loss frequently escapes detection until the age when hearing children normally begin to talk (9 months or older).[44–48] Current theory views auditory stimulation during the first 6 months of life as critical to development of speech and language skills. Children who are identified early as having hearing loss and receive intensive early intervention perform better on school-related measures (reading, arithmetic, vocabulary, articulation, percent of the child's communication understood by non–family members, social adjustment, and behavior) than children who do not receive such intervention.[49] Early intervention resulted in improvements in receptive language[50] and prevented developmental delays.[51] However, the efficacy of universal newborn hearing screening to improve long-term language outcomes remains uncertain.[52–54]

Screening

Newborn hearing screening is accomplished through the use of a variety of computerized equipment that uses automated auditory brainstem response (AABR), distortion product otoacoustic emissions (OAEs), or transient evoked OAEs. Screening is performed before discharge from the nursery.[55] Screening for congenital hearing loss is a simple process and in some cases may be performed by specially trained volunteers under the supervision of nurses or audiologists. Screening with AABR is accomplished by placement of soft earphones through which a series of soft clicks are introduced, usually at the 30- to 40-dB level. An

auditory brainstem response detected through electrodes attached to the infant's forehead and neck indicates that there is no significant sensorineural hearing loss. If OAE technology is selected as the screening test, a tiny microphone that detects sounds generated by the outer hair cells of the cochlea is introduced into the infant's auditory canal. Presence of those sounds indicates a functioning inner, middle, and outer ear. Each of these tests has advantages and disadvantages that should be considered carefully when selecting equipment. AABR tends to be somewhat more expensive and must be used in a quiet setting. OAE screening may result in higher false-positive rates if the infant's ear canal is blocked by fluid or debris.[56,57] Some hospitals use a combination of screening tests or repeat the OAE screening to reduce the false-positive rate and thereby minimize the need for follow-up after hospital discharge, which may reduce costs overall.[58]

Follow-up and Diagnostic Testing

Infants who do not "pass" the screening are either rescreened before discharge or given an appointment for rescreening as outpatients. Results of the screening are generally transmitted to the primary care physician of record, to the parents, and to the state health department. Failure to pass the screening results in a recommendation for referral to a qualified audiologist for confirmatory testing for congenital hearing loss.

In areas where universal newborn hearing screening is occurring, appropriate and timely diagnosis and intervention continue to be a major challenge. Attrition rates as high as 50% between initial referral and diagnostic confirmation still are not unusual.[36] Linkages between hospital-based screening programs and early intervention programs may not be well established, and data management and tracking of infants through the screening and diagnostic process also may be in the developmental stage.[49] As state programs assume more responsibility for the tracking and follow-up, these linkages will be more firmly established.[36]

Brief Overview of Disease Management

Appropriate management of all persons identified with congenital hearing loss requires a comprehensive pediatric and genetic evaluation.[33] Core personnel include individuals with expertise in the genetics of hearing loss, dysmorphology, audiology, otolaryngology, and genetic counseling. Qualified interpreting services may be needed when the parents are deaf. On the basis of the outcome of the evaluation, other types of professional expertise also may be needed, including professionals with experience with syndromal hearing loss (eg, ophthalmology, cardiology, nephrology, neurology).

After a family history, patient history, and physical examination, it may be possible to ascribe an etiology to the hearing loss. However, in approximately 30% of patients, there will be no obvious etiology.[33] An important goal of the

genetic evaluation is to attempt to distinguish isolated or simplex cases, in which the risk of deafness in subsequent offspring may be 25%, from sporadic cases, which have a low risk of recurrence.[33]

After diagnosis of hearing loss, continuity of care for the affected infant is important to reduce morbidity. The pediatrician should ensure referral to the state early intervention program and/or the state program for children with special health care needs as appropriate. Referral to these programs at hospital discharge helps to minimize loss to follow-up.

Current Controversy

The US Preventive Services Task Force did not find evidence for the benefit of (nor evidence against the benefit of) universal newborn hearing screening.[53] They argued that, among low-risk infants, the prevalence of hearing impairment was very low, and substantial numbers of infants would be misclassified. They found that evidence for the efficacy of early intervention for patients diagnosed by screening was incomplete.

Additional controversy centers on the generally inadequate integration of these programs with ongoing newborn screening and early intervention programs.[36] The Newborn Screening Task Force suggested that child health–related programs such as newborn genetic and hearing screening programs would avoid unnecessary duplication of effort if they were more closely aligned with each other.[59]

REFERENCES

33. American College of Medical Genetics, Genetic Evaluation of Congenital Hearing Loss Expert Panel. Genetics evaluation guidelines for the etiologic diagnosis of congenital hearing loss. *Genet Med.* 2002;4 :162 –171[ISI][Medline]

34. Rehm HL. A genetic approach to the child with sensorineural hearing loss. *Semin Perinatol.* 2005;29 :173 –181[CrossRef][ISI][Medline]

35. Schrijver I. Hereditary non-syndromic sensorineural hearing loss: transforming silence to sound. *J Mol Diagn.* 2004;6 :275 –284[Abstract/Free Full Text]

36. Lloyd-Puryear MA, Forsman I. Newborn screening and genetic testing. *J Obstet Gynecol Neonatal Nurs.* 2002;31 :200 –207[CrossRef][ISI][Medline]

37. Stein LK. Factors influencing the efficacy of universal newborn hearing screening. *Pediatr Clin North Am.* 1999;46 :95 –105[CrossRef][ISI][Medline]

38. Van Naarden K, Decoufle P, Caldwell K. Prevalence and characteristics of children with serious hearing impairment in metropolitan Atlanta, 1991–1993. *Pediatrics.* 1999;103 :570 –575[Abstract/Free Full Text]

39. Gorlin RJ, Toriello HV, Cohen MM. *Hereditary Hearing Loss and Its Syndromes*. New York, NY: Oxford Press; 1995

40. Morton NE. Genetic epidemiology of hearing impairment. *Ann N Y Acad Sci.* 1991;630 :16 –31[ISI][Medline]

41. Rsendes BL, Williamson RE, Morton CC. At the speed of sound: gene discovery in the auditory nervous system. *Am J Hum Genet.* 2001;69 :923 –935[CrossRef][ISI][Medline]

42. Steel KP, Kros CJ. A genetic approach to understanding auditory function. *Nat Genet.* 2001;27 :143 –149[CrossRef][ISI][Medline]

43. Van Camp G, Smith RJH. Hereditary hearing loss homepage. Available at: http://webhost.ua.ac.be/hhh. Accessed March 15, 2005

44. National Institutes of Health. *Early Identification of Hearing Impairment in Infants and Young Children*. Rockville, MD: National Institutes of Health; 1993

45. Fonseca S, Forsyth H, Grigor J, et al. Identification of permanent hearing loss in children: are the targets for outcome measures attainable? *Br J Audiol.* 1999;33 :135 –143[ISI][Medline]

46. Davis A, Bamford J, Wilson I, Ramkalawan T, Forshaw M, Wright S. A critical review of the role of neonatal hearing screening in the detection of congenital hearing impairment. *Health Technol Assess.* 1997;1 :i –iv, 1–176[Medline]

47. Meado-Orlans KP, Mertens DM, Sass-Lehrer MA, Scott-Olsen K. Support services for parents and their children who are deaf or hard of hearing: a national survey. *Am Ann Deaf.* 1997;142 :278 –288[ISI][Medline]

48. Harrion M, Roush J. Age of suspicion, identification, and intervention for infants and young children with hearing loss: a national study. *Ear Hear.* 1996;17 :55 –62[ISI][Medline]

49. Blake PE, Hall JW 3rd. The status of state-wide policies for neonatal hearing screening. *J Am Acad Audiol.* 1990;1 :67 –74[Medline]

50. Moeller MP. Early intervention and language development in children who are deaf and hard of hearing. *Pediatrics.* 2000;106(3) . Available at: www.pediatrics.org/cgi/content/full/106/3/e43

51. Yoshinaga-Itano C, Sedey AL, Coulter DK, Mehl AL. Language of early- and later-identified children with hearing loss. *Pediatrics.* 1998;102 :1161 –1171[Abstract/Free Full Text]

52. Helfand M, Thompson DC, Davis R, McPhillips H, Homer CJ, Lieu TL. *Systematic Evidence Review Number 5: Newborn Hearing Screening.* Washington, DC: Agency for Healthcare Research and Quality: 2001. AHRQ publication No. 02-S001

53. US Preventive Services Task Force. *Guide to Clinical Preventive Services*. 3rd ed. Washington, DC: Office of Disease Prevention and Health Promotion; 2000

54. Atkins D, Siegel J, Slutsky J. Making policy when the evidence is in dispute. *Health Aff (Millwood).* 2005;24 :102 –113[Abstract/Free Full Text]

55. National Center for Hearing Assessment and Management. Early hearing detection and intervention (EHDI) resources and information. Available at: www.infanthearing.org. Accessed March 15, 2005

56. Stach BA, Santilli CL. Technology in newborn hearing screening. *Semin Hear.* 1998;19 :247 –261

57. Folsom RC, Diefendorf AO. Physiologic and behavioral approaches to pediatric hearing assessment. *Pediatr Clin North Am.* 1999;46 :107 –120[CrossRef][ISI][Medline]

58. Vohr BR, Oh W, Stewart EJ, et al. Comparison of costs and referral rates of 3 universal newborn hearing screening protocols. *J Pediatr.* 2001;139 :238 –244[CrossRef][ISI][Medline]

59. American Academy of Pediatrics, Newborn Screening Task Force. Serving the family from birth to the medical home. Newborn screening: a blueprint for the future. *Pediatrics.* 2000;106 :389 –427[Free Full Text]