Diagnostic Evaluation of Children with Sensorineural Hearing Loss

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KEYWORDS

- Pediatric hearing loss • Sensorineural hearing loss
- Diagnostic evaluation of hearing loss • Genetic • Cytomegalovirus
- Enlarged vestibular aqueduct • Syndromes

KEY POINTS

- Sensorineural hearing loss (SNHL) occurs in approximately 2 to 4 per 1000 live births.
- The cause of pediatric SNHL may be genetic, acquired, or idiopathic.
- Approximately 50% of cases have a genetic cause.
- Of all genetic cases, 15% to 30% occur as part of a syndrome and 80% are transmitted in an autosomal-recessive pattern.
- Approximately 35% of SNHL patients have an acquired cause resulting from intrauterine infections (eg, cytomegalovirus), environmental exposures, meningitis, or prematurity.

INTRODUCTION

Sensorineural hearing loss (SNHL) is the most common sensory deficit in humans, occurring in 2 to 4 per 1000 live births and affecting an estimated 40,000 children in the United States annually.[1,2] This deficit can be unilateral or bilateral and is graded as mild to profound, based on the degree of hearing loss. Although the cause of SNHL can be broadly classified as genetic, acquired, or idiopathic, approximately 50% of children with moderate to profound congenital SNHL have a genetic cause.[3,4]

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Abbreviations

CMV	Cytomegalovirus
CT	Computed tomography
EKG	Electrocardiogram
PCR	Polymerase chain reaction
PDS	Pendred syndrome
SNHL	Sensorineural hearing loss

Most of these genetic phenotypes are not associated with a named syndrome or other anomaly (nonsyndromic), with only 15% to 30% occurring as part of a recognized syndrome.[5] Of all hereditary hearing loss cases, 80% are transmitted in an autosomal-recessive pattern (designated DFNB_), whereas the remaining are autosomal-dominant (DFNA_), mitochondrial, or sex-linked (DFNX_).[6]

Approximately 35% of children with SNHL have an identifiable acquired cause, which may be attributed to a wide array of causes, including intrauterine infections such as cytomegalovirus (CMV), environmental exposures, meningitis, ototoxic medications, maternal drug or alcohol use, prematurity, or low Apgar scores.[7] Hearing loss without an identified cause is classified as idiopathic.

Widespread adoption of universal newborn hearing screening has improved the ability to identify SNHL; however, the best approach to establishing the cause of the hearing loss has been controversial, and a wide variety of diagnostic tests have been used. Historically, patients underwent a comprehensive simultaneous "shotgun" test battery, which included temporal bone imaging, laboratory tests, and an electrocardiogram (EKG). Patients were also concurrently referred for consultation with specialists. Over the past decade, however, research pertaining to individual test yields and the optimization of genetic testing has shown that a stepwise sequential diagnostic approach is more prudent, because it reduces costs as well as utilization of resources.[8,9] This paradigm is now widely accepted and is discussed in this article.

IDENTIFYING PATIENTS WITH SENSORINEURAL HEARING LOSS

In 2007, the American Academy of Pediatrics Joint Committee on Infant Hearing issued a position statement that outlined recommendations for newborn hearing screening.[10] The report recommended that all infants be screened for hearing loss within the first month of life. More specifically, those at high risk should be screened with auditory brainstem response testing, whereas those at lower risk may be screened with otoacoustic emission testing. Infants who fail the initial screening should have repeat testing, and those who fail this testing should receive a comprehensive audiometric evaluation by 3 months of age. Infants who pass the initial screening should undergo surveillance of communication milestones, starting at the 2-month well-child checkup. If concerns arise, a referral for an audiometric evaluation should be made. Intervention should be initiated by 6 months of age.[10]

The degree or severity of hearing loss must also be documented. A loss between 20 and 40 dB is considered mild, a loss between 41 and 54 dB is moderate, a loss between 55 and 70 dB is moderately severe, and a loss between 71 and 90 dB or greater is considered profound.

CLINICAL HISTORY AND PHYSICAL EXAMINATION

Once the diagnosis of SNHL is established, a thorough medical history should be taken to identify possible risk factors for the hearing loss. The clinician should