

- The diagnosing pediatric audiologist should work with the Birth to Three early intervention coordinator to provide the parent/responsible party with the information they need to make informed decisions regarding early intervention options. This transition period should include ongoing audiologic assessment as the families select intervention options.
- The audiologist should notify the infant's pediatric healthcare provider and the DPH EHDI Program of all testing results, including inconclusive results, failure to show for scheduled appointments and any subsequent referrals.
- The audiologist is responsible for faxing the initial diagnostic and any subsequent audiological evaluation results to the CT Early Hearing Detection & Intervention Program **within 2 days** following the appointment. Results should be faxed to the EHDI Program (860 509-8132) for **any child born after July 1, 2000 (see Appendix G, Diagnostic Testing Reporting Form)**.

Audiological Evaluation from 6 to 36 Months of Age

For subsequent testing of infants and toddlers at developmental ages of 6 to 36 months, the confirmatory audiological test battery includes:

- Child and family history.
- Parental report of auditory and visual behaviors and communication milestones.
- Behavioral audiometry (either visual reinforcement or conditioned-play audiometry, depending on the child's developmental level), including pure-tone audiometry across the frequency range for each ear and speech-detection and -recognition measures.
- OAE testing.
- Acoustic immittance measures (tympanometry and acoustic reflex thresholds).
- ABR testing if responses to behavioral audiometry are not reliable or if ABR testing has not been performed in the past.

MEDICAL EVALUATION

Every infant with confirmed hearing loss and/or middle ear dysfunction should be referred for otologic and other medical evaluation. The purpose of these evaluations is to determine the etiology of hearing loss, to identify related physical conditions, and to provide recommendations for medical/surgical treatment as well as referral for other services. Essential components of the medical evaluation include:

- Clinical history,
- Family history of childhood-onset permanent hearing loss,
- Identification of syndromes associated with early or late-onset permanent hearing loss.
- A physical examination.
- Indicated radiologic and laboratory studies (including genetic testing).

Portions of the medical evaluation, such as urine culture for CMV, a leading cause of hearing loss, might even begin in the birth hospital, particularly for infants who spend time in the NICU.

PRIMARY HEALTH CARE PROFESSIONAL

The infant's pediatrician or other primary health care professional (PCP) is responsible for monitoring the general health, development, and well-being of the infant. The PCP should verify that the newborn's hearing screen was conducted during the child's first office visit. If the hearing screen was not done prior to discharge, the PCP should refer the family back to the birth facility, or to a diagnostic center where the hearing screen can be conducted. The following outlines additional responsibilities of the infant/child's PCP:

- Must assume responsibility to ensure that the diagnostic testing/audiological assessment is conducted in a timely fashion on all infants who do not pass the hearing screening.
- Initiate referrals for any medical specialty evaluations necessary to determine the etiology of the hearing loss.
- Monitor the child's middle-ear status because the presence of middle-ear effusion can further compromise hearing.
- Partner with other specialists, including the otolaryngologist, and audiologist to facilitate coordinated care for the infant and family.
- Because 30% to 40% of children with confirmed hearing loss will demonstrate developmental delays or other disabilities, the primary care physician should closely monitor developmental milestones and initiate referrals related to suspected disabilities.
- Review every infant's medical and family history for the presence of risk indicators that require monitoring for delayed-onset or progressive hearing loss and ensure that an audiological evaluation is completed for children at risk of hearing loss, at least once by 24 to 30 months of age according to the Joint Committee on Infant Hearing, regardless of their newborn screening results. The CT EHDI Advisory Board recommends that infants with one or more risk factors have an audiological evaluation every six months, up until age three. This includes infants who passed the hearing screening at birth.
- Infants with specific risk factors, marked with an asterisk on page 7 – such as CMV infection or a family history, are at increased risk of delayed-onset or progressive hearing loss and should be monitored closely.
- Conduct ongoing surveillance of parent concerns about language and hearing, auditory skills, and developmental milestones of all infants and children regardless of risk status, as outlined in the pediatric periodicity schedule published by the AAP.
- Children with cochlear implants may be at increased risk of acquiring bacterial meningitis compared with children in the general US population. The PCP must assure that all children with, and all potential recipients of, cochlear implants receive the CDC recommended pneumococcal immunization that apply to cochlear implant users and that they receive age-appropriate *Haemophilus Influenzae* Type B vaccines.

OTOLARYNGOLOGIST

Otolaryngologists are medical doctors who diagnose, treat, and manage a wide range of diseases of the head and neck and specialize in treating hearing and vestibular disorders. They perform a full medical diagnostic evaluation of the head and neck, ears, and related structures, including a comprehensive history and physical examination, leading to a medical diagnosis and appropriate medical and surgical management. Often, a hearing or balance disorder is an indicator of, or related to, a medically treatable condition or an underlying systemic disease. The otolaryngologists work closely with other healthcare professionals, including other physicians, audiologists, speech-language pathologists, educators, and others, in caring for patients with hearing, balance, voice, speech, developmental, and related disorders. It is important for a family to select an otolaryngologist that specializes in working with infants and children.

- The otolaryngologist's evaluation includes a comprehensive history to identify the presence of risk factors for early-onset childhood permanent hearing loss, such as family history of hearing loss, having been admitted to the NICU for more than 5 days, and having received ECMO.
- A complete head and neck examination for craniofacial anomalies should document defects of the auricles, patency of the external ear canals, and status of the eardrum and middle-ear structures. Atypical findings on eye examination, including irises of 2 different colors or abnormal positioning of the eyes, may signal a syndrome that includes hearing loss. Congenital permanent conductive hearing loss may be associated with craniofacial anomalies that are seen in disorders such as Crouzon disease, Klippel-Feil syndrome, and Goldenhar syndrome. The assessment of infants with these congenital anomalies should be coordinated with a clinical geneticist.
- In large population studies, at least 50% of congenital hearing loss has been designated as hereditary, and nearly 600 syndromes and 125 genes associated with hearing loss have already been identified. The evaluation, therefore, should include a review of family history of specific genetic disorders or syndromes, including genetic testing for gene mutations such as *GJB2* (connexin-26), and syndromes commonly associated with early-onset childhood sensorineural hearing loss. As the widespread use of newly developed conjugate vaccines decreases the prevalence of infectious etiologies such as measles, mumps, rubella, *Haemophilus Influenzae* Type B, and childhood meningitis, the percentage of each successive cohort of early-onset hearing loss attributable to genetic etiologies can be expected to increase, prompting recommendations for early genetic evaluations.
- Approximately 30% to 40% of children with hearing loss have associated disabilities, which can be of importance in patient management. The decision to obtain genetic testing depends on informed family choice, in conjunction with standard confidentiality guidelines.
- In the absence of a genetic or established medical cause, a computed tomography scan of the temporal bones may be performed to identify cochlear abnormalities, such as Mondini deformity with an enlarged vestibular aqueduct, which have been associated with progressive hearing loss.
- Temporal bone imaging studies may also be used to assess potential candidacy for surgical intervention, including reconstruction, bone-anchored hearing aid, and cochlear implantation. According to the JCIH, recent data have shown that some children with electrophysiologic evidence suggesting auditory neuropathy/dyssynchrony may have an absent or abnormal cochlear nerve that may be detected with MRI.

Historically, an extensive battery of laboratory and radiographic studies was routinely recommended for newborn infants and children with newly diagnosed sensorineural hearing loss. However, emerging technologies for the diagnosis of genetic and infectious disorders have simplified the search for a definitive diagnosis, which obviates the need for costly diagnostic evaluations in some instances. If, after an initial evaluation, the etiology remains uncertain, an expanded multidisciplinary evaluation protocol including electrocardiography, urinalysis, testing for CMV, and further radiographic studies is indicated. The etiology of neonatal hearing loss may remain uncertain in as many as 30% to 40% of children.

- Once hearing loss is confirmed, medical clearance for hearing aids and enrollment into Birth to Three should be initiated as soon as possible. Amplification and early intervention should not be delayed pending the outcome of the diagnostic process.
- The otolaryngologist should conduct careful longitudinal monitoring to detect and promptly treat any coexisting middle-ear effusions.

MEDICAL GENETICIST

A referral to the Medical Geneticist can provide families with information on the etiology of the hearing loss, prognosis for progression, associated disorders (e.g., renal, vision, cardiac), and the likelihood of recurrence in future offspring. This information may influence the parents' decision-making regarding intervention options for their child. All families of children with confirmed hearing loss should be offered a genetics evaluation and counseling. The medical geneticist is responsible for the following:

- Interpreting family history data.
- Clinically evaluating and diagnosing inherited disorders, if present.
- Performing and assessing genetic tests.
- Providing genetic counseling to the family.

OTHER MEDICAL SPECIALISTS

Every infant with a confirmed hearing loss should have an evaluation by an ophthalmologist to document visual acuity and rule out concomitant or late-onset vision disorders such as Usher syndrome. Indicated referrals to other medical subspecialists, including developmental pediatricians, neurologists, cardiologists, and nephrologists, should be facilitated and coordinated by the primary health care professional.

EARLY INTERVENTION

Before newborn hearing screening was instituted universally, children with severe-to-profound hearing loss, on average, completed the 12th grade with a 3rd- to 4th-grade reading level and language levels of a 9- to 10-year-old hearing child. In contrast, infants and children with mild-to-profound hearing loss who are identified in the first 6 months of life and provided with immediate and appropriate early intervention services have significantly better outcomes than later-identified infants and children in vocabulary development, receptive and expressive language, syntax, speech production, and social-emotional development.

According to federal guidelines and CT State law (C.G.S. 19a-59), once any degree of hearing loss is confirmed in a child, a referral should be initiated to the CT Birth to Three System, Connecticut's early intervention program, within 2 days of confirmation of hearing loss. Referrals are made to Birth to Three by calling the Child Development Infoline at 1-800-505-7000. More information about the CT Birth to Three System can be obtained by visiting their website at: <http://www.birth23.org>

- The family should be referred to Birth to Three by the diagnosing audiologist at the time of diagnosis and EI services should be initiated as soon as possible, and no later than 6 months of age.
- CT has three EI centers that specialize in infants and children who are deaf or hard of hearing: American School for the Deaf, CREC/Soundbridge and New England Center for Hearing Rehabilitation (NECHEAR). **See Appendix H, Birth to Three Centers.** Upon referral to Early Intervention the family will receive information about each of the three programs. Although the audiologist and PCP should be available to answer any questions that the family may have about the three programs, the choice as to which program is selected, should be made by the family.
- Children with other medical conditions in which hearing loss is not the primary disability, should have access to intervention with a provider who is knowledgeable about hearing loss.