

## Newborn Screening ACT Sheet

# [Congenital Hearing Loss]

## Congenital Hearing Loss

**Differential Diagnosis:** Non-syndromic (hearing loss as the only clinical finding), including non-genetic and genetic causes; syndromic (associated with/caused by various genetic syndromes); anatomic abnormalities of the cochleovestibular system; congenital CMV; environmental exposures; ototoxic medications; prematurity.

**Condition Description:** Hearing loss can be congenital, acquired, and/or progressive. Congenital hearing loss can be classified as syndromic or non-syndromic. Acquired hearing loss is due to environmental factors such as ototoxicity or prenatal or congenital infections. Types of permanent hearing loss are characterized as sensorineural, conductive, or mixed. Most non-syndromic hearing loss is sensorineural, caused by alterations in the cochlea, auditory nerve, or auditory processing pathways, and may be unilateral or bilateral. Conductive hearing loss, due to abnormal mechanical transmission of sound from the external ear to the cochlea, is most commonly due to otitis media with middle ear fluid. Permanent conductive hearing loss is less common at birth but may occur due to anatomic changes to the external ear and auditory canal. The severity of hearing loss depends on the etiology of the disorder.

### You Should Take the Following Actions:

- Inform family of the newborn screening result and review the high false positive rate for initial hearing screening.
- Ascertain clinical status (with special attention to complications during pregnancy/delivery, NICU stay, family history, and genetic syndromes).
- Evaluate the newborn for associated clinical findings.
- Consult with pediatric audiologist and otolaryngologist.
- Initiate confirmatory testing and management, as recommended by the specialist.
- Follow Early Hearing Detection and Intervention (EHDI) 1-3-6 Guidelines for the timeline for screening, diagnostic audiologic testing, and habilitation (see link below). **Refer to Part C of the IDEA Act/Early Intervention if permanent hearing loss is suspected or confirmed.**
- Consider referral to pediatric geneticist or genetic counselor after hearing loss is confirmed.
- Provide family basic information about hearing loss.
- Report the final diagnostic outcome to the EHDI newborn screening program.

**Diagnostic Evaluation:** **CMV testing:** should be performed by PCR on a saliva sample within the first 3 weeks of life. **Diagnostic Auditory Evaluation by ABR, OAEs, high-frequency tympanometry:** may be used to confirm the diagnosis. **Molecular genetic testing:** may identify non-syndromic or syndromic causes of hearing loss and may identify other testing or monitoring that is indicated.

**Clinical Considerations:** Development of communication is integral to social, emotional, language, and developmental outcomes; there is no association between hearing loss and cognition. Current screening programs detect hearing loss to greater than 30 decibels. While genetic causes account for about half of congenital hearing loss, the remainder is most often due to anatomic changes or environmental exposures. Newborn screening may also identify neonates who may develop later onset hearing loss and it is important to establish regular monitoring for these infants. The majority of newborns with hearing loss have hearing parents. Management depends upon the etiology and the severity of hearing loss as well as on family preferences.

### Additional Information:

[How to Communicate Newborn Screening Results](#)

[Gene Reviews](#)

[Medline Plus](#)

[Condition Information for Families- HRSA Newborn Screening Clearinghouse](#)

[Clinical evaluation and etiologic diagnosis of hearing loss: A clinical practice resource of the ACMG](#)

[American Academy Of Audiology](#)

[ASHA EHDI Resources](#)

[Refer to Part C of the IDEA Act/Early Intervention](#)

[EHDI 1-3-6 Plan](#)

[EHDI by State](#)

[Clinicaltrials.gov](#)

### Referral (local, state, regional, and national):

[Find a Genetics Clinic Directory](#)

[Genetic Testing Registry](#)

[Find An Audiologist](#)

Disclaimer: This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.