

## Newborn Screening ACT Sheet [Congenital Hearing Loss >30db] Congenital Hearing Loss

**Differential Diagnosis:** Extensive. Includes 40% environmental (mostly bacterial/viral) and 60% genetic (30% syndromal and 70% non-syndromal representing over 100 genes).

**Condition Description:** Defined as hearing loss that is permanent, bilateral or unilateral, sensorineural or conductive, and averaging loss of 30 decibels or more in the frequency range important for speech recognition.

### ***YOU SHOULD TAKE THE FOLLOWING ACTIONS:***

- Contact family to inform them of the newborn screening result.
- Ensure coordinated and comprehensive multidisciplinary hearing loss evaluation and care.
- Initiate timely diagnostic evaluation by a multidisciplinary hearing loss team, including evaluation by a genetic specialist.
- Report findings to state Early Hearing Detection and Intervention (EHDI) program.

**Diagnostic Evaluation:** Hearing loss is confirmed and followed up by a comprehensive hearing loss team evaluation and testing for an etiologic diagnosis. Testing algorithms are prioritized around family history and likelihood of a syndromal condition. If familial and/or non-syndromal, *GJB2* (Connexin 26) and *GJB6* (Connexin 30) gene testing is done. Cytomegalovirus (CMV) and mitochondrial etiologies are also possible. Confirmatory work should be completed by age 3 months and early intervention services initiated before 6 months of age.

**Clinical Considerations:** Hearing loss may indicate a genetic syndrome with involvement of other organ systems. Untreated hearing loss can result in lifelong deficits in speech and language development, so it is critical that all infants who fail newborn screening have follow-up testing.

### **Additional Information:**

[Gene Tests/Gene Clinics](#)  
[National Center for Hearing Assessment and Management](#)  
[Genetics Home Reference](#)  
[Centers for Disease Control and Prevention](#)  
[Joint Commission on Infant Hearing](#)  
[American College of Medical Genetics](#)  
[American Academy of Pediatrics](#)

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

[Testing](#)  
[Clinical Services](#)  
[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It 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. Adherence to this guideline does not necessarily ensure a successful medical outcome. 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 guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

LOCAL RESOURCES: Insert State newborn screening program web site links

State/Local Resource site (insert state/local newborn screening program website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Tests/Gene Clinics

<http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=EJ4Gy2VAan2GT&gry=&fcn=y&fw=d1eQ&filename=/profiles/deafness-overview/index.html>

<http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=EJ4Gy2VAan2GT&gry=&fcn=y&fw=Rbu7&filename=/profiles/mt-deafness/index.html>

National Center for Hearing Assessment and Management

<http://www.infanthearing.org/physicianeducation/index.html>

Genetics Home Reference

<http://ghr.nlm.nih.gov/ghr/search/template/gen%2Cbrowse%2Cglossary%2Cbrowse.vm?query=hearing+loss&x=5&y=11>

CDC

<http://www.cdc.gov/ncbddd/dd/ddhi.htm>

Joint Commission on Infant Hearing

<http://pediatrics.aappublications.org/cgi/content/extract/120/4/898>

American College of Medical Genetics

<http://www.acmg.net/resources/policies/we0302000162.pdf>

American Academy of Pediatrics

<http://pediatrics.aappublications.org/cgi/content/extract/106/4/798>

Referral (local, state, regional and national):

Testing

Clinical Services

<http://www.genetests.org/servlet/access?id=8888891&key=ycNiicOsM18KA&fcn=y&fw=z4HV&filename=/clinicsearch/searchclinic.html>

Find Genetic Services

<http://www.acmg.net/GIS/Disclaimer.aspx>

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