



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% nonsyndromal 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.
- Consult audiologist or ENT to confirm hearing loss and refer, if needed.
- Evaluate infant (see clinical expectations below).
- Initiate timely diagnostic and comprehensive genetics evaluation as recommended by a multidisciplinary hearing loss team.
- Initiate treatment as recommended by team.
- Educate family that hearing intervention improves developmental outcomes.
- Report findings to newborn screening 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 nonsyndromal, GJB2 testing is done. Cytomegalovirus (CMV) and mitochondrial etiologies are also possible. Confirmatory work should be completed by age 3 months.

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:

(Click on the name to take you to the website. Complete URLs are listed in the Appendix)

Gene Tests/Gene Clinics

National Center for Hearing Assessment and Management

Genetics Home Reference

Joint Commission on Infant Hearing

American College of Medical Genetics

American Academy of Pediatrics

Referral (local, state, regional and national):

Genetic Etiology

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality clinical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines 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 health care provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines.

State Resource Site *(insert state newborn screening program website information)*

Name
URL
Comments

Local Resource Sites *(insert local and regional newborn screening website information)*

Name
URL
Comments

Name
URL
Comments

Name
URL
Comments

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>

Joint Commission on Infant Hearing <http://www.jcih.org/jcih2000.pdf>

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):

Genetic Etiology

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

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