

Quick Reference Guide:

Genetic Counseling Training Guide for Genetic Hearing Loss

Pre-test Genetic Counseling

- Genetic counseling is important to provide patients and families with the information and support they need to make informed decisions about undergoing genetic testing for hearing loss
- Review the patient's personal and family history, provide education about hearing loss genetics, address psychological considerations, and discuss the elements of informed consent, including genetic discrimination and legal protections (i.e. Genetic Information Nondiscrimination Act - GINA), the potential results, and implications

Critical Talking Points:

- "Genetic testing may identify a genetic cause for [your/your child's] hearing loss. This comprehensive gene panel is designed to look for genetic changes in genes associated with hearing loss. It does not look at all your genes."
- "This test requires a [blood/saliva/cheek swab] sample."
- "There are 3 types of results we may receive from the test:
 - A positive result means...
 - A negative result means...
 - An uncertain result means..."
- "These results may impact [your/your child's] prognosis and medical management because..."
- "Regulations may protect [you/your child] against some forms of genetic discrimination..."

Post-test Genetic Counseling

- Review result disclosure and implications, explain the impact on prognosis and management, address psychological considerations, identify and provide support resources, share the test report with the patient, and offer referral to genetics if further genetic counseling is needed

Positive Genetic Test Results

- Indicates the patient has a pathogenic or likely pathogenic variant(s) in a gene associated with hearing loss
- Implications for receiving a positive result might include:
 - Clarify diagnosis (syndromic or isolated) and prognosis (stable or progressive)
 - Inform decisions about referral to a specialist or surveillance
 - Provide information about recurrence risks (inheritance pattern)
 - Indicate preventive or therapeutic interventions
 - Allow an individual to participate in research opportunities like registries and clinical trials

Negative Genetic Test Results

- Indicates the patient does not have a pathogenic or likely pathogenic variant in one of the genes associated with hearing loss
- This result significantly decreases the likelihood of a genetic cause for hearing loss, which can be reassuring. However, additional testing may be recommended if a small panel of genes was tested

Uncertain Genetic Test Results

- The lab identified a variant of uncertain significance (VUS) in a gene for hearing loss; there is no definitive evidence the variant is associated with hearing loss
- This result classification may change in the future if more data become available



Pre-test
Counseling



Interpreting Genetic
Test Reports



Post-test
Counseling

