

Region 4 Genetics Collaborative Early Hearing Detection and Intervention (EHDI) Follow-up Workgroup Genetic Referral Guidelines

Introduction:

In children diagnosed with sensorineural hearing loss, 50% are found to be due to genetic causes. Of those, about one third are affected with a complex medical syndrome. Since the majority of genetic hearing loss is caused by recessive genes, family history is usually negative. However, when family history is positive for permanent hearing loss, the chance for hearing loss in a child ranges from minimal, <1%, to approaching 100%.

Identification of associated features in hearing loss syndromes may have health saving or life saving implications. Thus, families of children diagnosed with hearing loss should receive information about genetics services as a complement to their child's overall health care. In certain circumstances it is appropriate to refer a hearing sibling of a child with permanent hearing loss for genetic evaluation. For example, the onset and/or progression of inherited hearing loss may vary among family members. Also, genetic syndromes can present differently in different family members. Genetic evaluation may allow for targeted investigations to identify health conditions early. It is most appropriate to defer carrier testing in siblings until an informed decision about testing can be made by the individual to be tested.

National Support for Genetic Referral

The Joint Committee on Infant Hearing (JCH) 2007 Position Statement states:

"All families of children with confirmed hearing loss should be offered, and may benefit from, a genetics evaluation and counseling. This evaluation can provide families with information on etiology of hearing loss, prognosis for progression, associated disorders (e.g., renal, vision, cardiac), and likelihood of recurrence in future offspring. This information may influence parents' decision making regarding intervention options for their child".

The American College of Medical Genetics (ACMG) recommends that all children with confirmed hearing loss be referred for genetic evaluation and counseling.

Purpose of the Genetic Referral

A genetic evaluation and consultation is done to try to determine the cause of the hearing loss. Understanding the cause can provide answers to many questions and provide families with information to make important decisions.

- Does the child have, or is he at risk for, any other health conditions?

- Are there any associated medical complications?
- Will the hearing loss progress?
- What are the chances another family member might be born with or develop hearing loss?
- Are there other family members that could be affected?

Facilitating the Referral

Referral for genetic evaluation should be made as soon as possible after the diagnosis of hearing loss has been confirmed, ideally by 3-6 months of age. Referral should be made by providers working with the family and may include the Medical Home, ENT, Audiologist, Early Interventionist, etc. There are several opportunities for providers to facilitate referral for genetic services along the continuum of follow-up activities that occur throughout the Early Hearing Detection and Intervention (EHDI) service systems. The Region 4 Genetics Collaborative EHDI Follow-up Workgroup has developed a toolkit to assist providers in making genetic referrals for families who have a child with confirmed permanent hearing loss.

Special Considerations

It is important to consider the unique family situation when making the referral. There are times when making an immediate referral is of utmost importance; there are others when sensitivity to the family situation determines the timing of a referral to increase the chances of follow through.

- 1) Reasons for immediate referral include, but are not limited to:
 - Suspected genetic diagnosis with additional health concerns
 - Parent/caregiver concern
 - Parental consanguinity
 - Relative who has a syndrome associated with hearing loss (do we need to clarify how close a relative? and biologic/blood relative?)
 - Child with diagnosed hearing loss who had exposure to aminoglycosidic antibiotics (susceptibility to hearing loss induced by these antibiotics can be inherited)
- 2) Issues when families have a history of hearing loss (occurs about 10% of the time)
 - Parents/caregivers who are deaf may view hearing loss as a difference, not a disability
 - The cause of the relative's hearing loss may not be genetic. The relatives' biologic relationship to the child with hearing loss should be confirmed. (I'm not positive this needs to be said, or that this is the best place for it. Maybe it would be better with recs for provider to take the family history?)

- The Medical Home/ENT should discuss the genetic evaluation role in determining potential medical management and intervention strategies
- 3) Issues when families have no history of hearing loss (occurs about 90% of the time)
- The Medical Home must be sensitive to the needs of the family while communicating the importance of the genetic evaluation for assessing for other possible health risks and **discussing? addressing?** implications for other family members
 - Consanguinity is a cultural norm for some. **(Does anything more need to be said here? – Families can be given accurate information concerning recurrence in a manner that recognizes the importance of committed, engaged parents. I don't want this to sound preachy, but people need to be approached in a nonjudgemental manner to be able to get at the important issues of diagnosis, recurrence and treatment at home)**

Provider Responsibilities in the Referral Process

All providers should take responsibility for the genetic referral as soon as possible after confirmation of hearing loss. This responsibility may include making the referral directly, or depending on the family's insurance it may mean working with the Medical Home or primary care physician to have them make the referral. (To foster coordinated care, specialty providers should make every effort to communicate recommendations and results to the Medical Home)

The Medical Home/Primary Care/ENT Responsibilities:

- Obtain family history to assess timing (urgency) of the referral (other medical conditions that put the child's health at risk) **Should this be family and medical history?**
- Discuss with the family the referral as a tool for decision-making? that might affect medical and audiologic intervention
- Refer the family to a genetics center and facilitate scheduling of the appointment
- Ensure that all professionals working with the family are aware of any medical implications

The Audiologist's Responsibilities:

- Complete report describing the hearing loss (type, degree and configuration) and tests performed (tympanometry, auditory brain response, oto-acoustic emissions, etc.)
- Provide report to the Medical Home/primary care and recommend to the Medical Home/primary care the need for offering the family a genetic referral
- Counsel the family about the role of the genetics evaluation in determining etiology of the hearing loss, identifying other health related issues, and addressing implications for other family members

- Ask the family if the physician has discussed this referral with them and provide materials about genetic evaluation for children with confirmed hearing loss and local genetic services
- Encourage the family to discuss genetic referral with the Medical Home The Service Coordinator's Responsibilities (is this the same as Early Intervention?):
- Counsel the family about the role of the genetics evaluation in determining etiology of the hearing loss , identifying other health related issues, and addressing implications for other family members
- Ensure the intervention plan includes information that develops from the genetic evaluation? e.g., monitor for x due to being a frequent symptom of y syndrome?

Where to Refer for Genetic Evaluation

Each Region 4 state (IL, IN, KY, MI, MN, OH, & WI) has a list of available centers for genetic referral. Lists are included in the Tools Section of the Provider Action Guide.