

Region 4 Genetics Collaborative 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.

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.

National Support for Genetic Referral

The Joint Commission 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 associate 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 hearing loss.

Special Considerations

It is important to consider the unique family situation when making the referral. There are times where making the referral immediately is of utmost importance; while there may be other circumstances where sensitivity to the unique family situation determines how and when a referral should be made to increase the chances of the family following through with the genetic consultation.

- 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
 - 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
 - Parents/caregivers who are deaf may view hearing loss as a difference, not a disability
 - 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 needs of the family while still communicating the importance of the genetic evaluation for assessing for other possible health risks and implications for other family members

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.

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)
- Discuss with the family the referral as a tool for decision 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 and identifying other health related issues
- 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:

- Counsel the family about the role of the genetics evaluation in determining etiology of the hearing loss and identifying other health related issues
- Ensure the intervention plan includes information that develops from the genetic evaluation

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