GUIDELINES FOR GENETIC EVALUATON REFERRAL

The prevalence of permanent hearing loss in infants is estimated to be 2-3/1000 in the United States (Finitzo et al., 1998; Prieve et al., 2000). One or both ears may be affected. Until the 1970's when universal immunization was introduced, maternal rubella infection caused a significant proportion of congenital hearing loss. Today, about 50% of congenital and early onset hearing loss is attributable to genetic factors (Marazita et al, 1993), and considerable progress has been made in identifying genes for deafness over the last few years.

Genetic hearing loss is usually divided into two categories. In approximately 70% of cases present at birth, the hearing loss occurs as an isolated trait and is called *nonsyndromic*. The auditory deficit in nonsyndromic hearing loss can vary, but is most often sensorineural. It can be unilateral or bilateral, congenital or late-onset, and stable or progressive. The remaining 30% of hearing loss is classified as *syndromic*, meaning that it occurs as part of a collection of anomalies (i.e., physical features and/or medical conditions). Syndromic hearing loss varies widely and can be conductive, sensorineural or mixed; unilateral or bilateral; congenital or late-onset; and stable or progressive (Resendes et al, 2001). There are at least 75 genetic types of nonsyndromic hearing loss (Van Camp & Smith, 2002) and more than 400 genetic types of syndromic hearing loss (Gorlin, et al, 1995; Stell & Kros, 2001). Children born with hearing loss often have the same physical and cognitive development as those who are born hearing.

I. PURPOSE OF REFERRAL

The primary purpose of a genetic evaluation is to investigate the etiology of the hearing loss in order to anticipate whether the child has, or is at risk for, other medical conditions. Examination by a skilled dysmorphologist may uncover subtle indicators of a genetic syndrome. While genetic evaluation does not always pinpoint an exact etiology, it can yield important information about the hereditary nature of a hearing loss. Establishing a genetic diagnosis whenever possible is more important now than in the past. New information about the pathophysiology and natural history of the different forms of hereditary hearing loss is rapidly emerging. Therapeutic interventions to reduce the risk of complications associated with specific etiologies may someday be available.

Other important benefits of the genetic evaluation are to 1) identify other family members, particularly young siblings, who should be evaluated for possible hearing loss or associated medical conditions, and 2) provide recurrence information for family planning. A referral to a genetics center allows genetic tests to be offered in conjunction with appropriate counseling.

II. WHEN TO MAKE A REFERRAL

A. Position Statements

The Joint Committee on Infant Hearing has recommended that families be offered the option of genetic evaluation and counseling by a medical geneticist where

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thorough physical and laboratory investigations fail to define the etiology of hearing loss. (2000, p. 16). In a report produced for the Maternal and Child Health Bureau (MCHB) of the Federal Health Resources and Services Administration (HRSA), the American College of Medical Genetics recommends that all children with confirmed hearing loss be referred for genetic evaluation and counseling to a genetics team that typically includes a geneticist and genetic counselor. Accordingly, it is reasonable to refer all infants with confirmed permanent hearing loss (i.e., permanent sensorineural, conductive or mixed hearing loss) for genetic evaluation after discussion with the parent(s)/caregiver(s) about the potential benefits and limitations of the genetic evaluation and counseling process.

B. Prioritizing the Appointment

The Medical Home should facilitate referral for genetic evaluation as soon as possible once the diagnosis of permanent hearing loss has been confirmed, ideally by 3 months of age. It is important to prioritize the timing of the appointment in light of many other appointments the family members may have, and their adjustment to the diagnosis.

1. Reasons for Immediate Referral

Reasons for immediate genetic referral include, but are not limited to the following:

a. Suspected genetic diagnosis associated with additional health conditions.
b. Parent(s)/Caregiver(s) are asking for information about the possible cause and/or want to know the chances of recurrence for family planning and appropriate medical care for other family members, especially children.
c. Parental consanguinity (i.e., the parents share a common biological ancestor).
d. Relative with a syndromic cause of hearing loss or other manifestations of a syndrome known to include hearing loss.
e. Hearing loss in a child exposed to aminogylcosidic antibiotics (i.e., antibiotics from the -mycin group used to fight certain infections). Susceptibility to hearing loss induced by these antibiotics can be inherited.
f. Need for assistance with interpretation of genetic or other test results ordered by the Medical Home or pediatric specialists

2. Possible issues with families with a negative family history of hearing loss

The majority of genetic hearing loss is transmitted as an autosomal recessive trait. Autosomal refers to genes located on non-sex chromosomes; recessive means that both copies of a gene, in a given pair, must be affected for the person to have the condition. Therefore, there is often no history of similar hearing loss in a child's biological parents or other close relatives. It is estimated that 90% of children with hearing loss have parents who are hearing (Gorlin, Toriello & Cohen, 1995). In the absence of a family history, the news that their child has hearing loss can come as a shock to parent(s)/caregiver(s) and they often experience stages

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of grieving with regard to the loss of their anticipated "healthy" baby. When a diagnosis of hearing loss is made, parent(s)/caregiver(s) are often overwhelmed by the number of medical appointments needed for their child and the amount of information and advice they are receiving from a variety of professionals, friends, and family. This, in addition to an absence of family history, and the social stigma sometimes associated with genetic conditions, may make parent(s)/caregiver(s) reluctant to pursue a genetic consultation. It is important for the Medical Home to be sensitive to the family's adjustment process as they coordinate the overall medical management of the child, while communicating the importance of a genetic evaluation for assessing other potential health risks and possibly, optimal interventions.

3. Possible issues with families with a positive family history of hearing loss
Parent(s)/Caregiver(s) who are deaf may view hearing loss as a difference that is a normal variation in humans, not a disorder. Since it is a common misconception that the only purpose of a genetic evaluation is to reduce the recurrence of a condition in a family, parent(s)/caregiver(s) may be hesitant to accept a genetic referral. The Medical Home should counsel the family about the role of the genetic evaluation in determining potential medical management and intervention strategies.

III. RESPONSIBILITIES FOR THE REFERRAL PROCESS

A. The Medical Home's Responsibilities

1. Obtain a family history and past medical history to assess the urgency of genetic referral.
2. Discuss the importance of a genetic referral for investigating the etiology as a basis for decisions that might affect medical and audiologic intervention.
3. Refer parent(s)/caregiver(s) to a genetics center (listed on page 8).
4. Ensure that the Service Coordinator is aware of all medical implications.

B. Audiologist's Responsibilities

1. Complete an audiologic report describing the hearing loss (e.g., type, degree, and configuration) and audiologic tests performed (e.g., tympanometry, auditory brainstem response, otoacoustic emissions).
2. Provide the Medical Home with a copy of the audiologic report and recommend to the Medical Home the need for genetic referral as it relates to the overall implications for the medical and intervention management of the child.
3. Counsel the family regarding the role of a genetics evaluation in determining the etiology of the hearing loss and identifying other health-related issues.

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C. Service Coordinator's Responsibilities (*Early On* ® Michigan)

1. Counsel the family regarding the role of a genetics evaluation in determining the etiology of the hearing loss and identifying other health-related issues.
2. Ensure that the plan for intervention services takes into account any information available from genetic counseling, if known.

IV. WHERE TO MAKE THE GENETIC REFERRAL

Six regional pediatric genetic centers are located in southeast and mid-Michigan and also conduct outreach clinics in other geographic areas as noted on page 8 (Genetic Clinic Contact Information). The Hereditary Disorders Program at the Michigan Department of Community Health helps to coordinate a statewide network of clinics that can provide genetic evaluation and counseling. For more information contact the state genetics coordinator at (517) 335-8887 or visit http://www.mdch.state.mi.us/dch/clcf/hdp/.

V. GENETIC EVALUATION

A. Professionals Involved

A comprehensive genetic evaluation with genetic counseling requires a multi-disciplinary approach. Specific genetic professionals include:

1. Clinical Geneticist: Physicians who are certified by the American College of Medical Genetics after completing a two year fellowship in clinical genetics, which trains them to diagnose and treat genetic conditions. Clinical geneticists are usually also trained and board certified in one of the following disciplines: pediatrics, internal medicine, oncology, pathology, or obstetrics/gynecology.
2. Genetic Counselor: Professionals who have graduated from an accredited Master's level genetic counseling graduate program and are certified (or eligible for certification) through the American Board of Genetic Counseling. Genetic counselors are trained to recognize and assess genetic conditions in an individual and family, educate patients about genetic conditions and genetic testing, and help patients and families cope with the diagnosis of a genetic condition.

B. Evaluation Process

Typically, the genetic evaluation includes:

1. Reviewing pertinent medical records.
2. Taking a complete, 3-4 generation family history, with emphasis on key genetic and phenotypic features (e.g. visual anomalies, specific facial dysmorphology, endocrine abnormalities, cardiac symptoms, etc.) as well as audiometric characteristics of hearing loss in relatives.
3. Taking a comprehensive patient medical and developmental history, including assessment of possible infectious etiologies.
4. Performing a physical and dysmorphology examination.

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5. Discussing the benefits, risks, limitations, and cost of genetic testing with parent(s)/caregiver(s).
6. Ordering genetic and/or other laboratory tests as indicated.

C. Genetic Counseling and Follow-up

After a genetic diagnosis is established, genetic counseling is provided to address the family's questions and concerns, including an explanation of the natural history of the condition, pattern of inheritance, medical implications, and possibility of recurrence or associated health risks in other relatives. Sometimes a precise genetic diagnosis cannot be made, even after thorough evaluation and testing. Often genetic counseling can still provide helpful information about differential diagnoses, recurrence, medical management, and importantly, available community, state, and national resources for information, support, and services. In addition, the genetics staff arranges for referrals (through the Medical Home) to other specialists when indicated, to help further evaluate etiology, and/or follow-up evaluations through the genetics clinic.

D. Funding

The cost of the genetics consultation and testing is often covered by third-party insurers, Children's Special Health Care Services and Medicaid. The genetic counselor may be able to help identify and coordinate sources of payment for families without health insurance.

E. Reporting

A summary of the genetic evaluation, including test results, is communicated to the Medical Home via letter or clinic consultation note. Parent(s)/Caregiver(s) may also receive a copy and should request one if it is not provided. In addition, at the parents'/caregivers' request, this information can be sent to other specialists involved in their child's care. The genetics professionals also communicate verbally with the Medical Home and/or other specialists as needed.

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