



Increasing Genetic Referrals for Children Identified Through the Early Hearing Detection and Intervention (EHDI) System

An Action Guide for Providers Serving Children Who Have Hearing Loss



Region4
Genetics Collaborative

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ACKNOWLEDGEMENTS

Thank you to the Region 4 Genetics Collaborative Early Hearing Detection and Intervention (EHDI) Follow-up Workgroup members who contributed their time and effort to produce this helpful guide. The information and resources are intended for educational use only and are provided solely as a service. The information provided should not be used for diagnosing or treating a health problem or disease, and is not a substitute for professional care.

The Region 4 Genetics Collaborative is funded by the Health Resources and Services Administration (HRSA) Maternal and Child Health Bureau (MCHB) Cooperative Agreement U22MC03963. The Region 4 Genetics Collaborative is a project of the Michigan Public Health Institute.



REGION 4 WELCOMES YOUR INPUT

We hope that this Action Guide proves to be a useful tool for increasing access to genetic services for families whose children have been diagnosed with hearing loss. We welcome suggestions for adding encounters, actions for providers, tools, or other feedback to improve its utility. Please complete the feedback from included at the end of this guide (page 66).

Table of Contents

INTRODUCTION	3
HOW TO USE THIS GUIDE	3
SECTION ONE: GUIDELINES & OPPORTUNITIES	4
Importance of the Genetic Referral	5
Timeliness of the Genetic Referral	6
Opportunities for Genetic Referral during the EHDI process	6
Special Consideration when Referring	7
SECTION TWO: ACTIONS FOR PROVIDERS	8
Actions for EHDI Follow-up Staff	9
Actions for Pediatric Audiologists, Otolaryngologist (ENT) & Otologists	11
Actions for Primary Care Providers, Pediatricians, Family Practitioners and the Medical Home	14
Actions for Early Intervention Service Providers	17
SECTION THREE: TOOLS FOR PROVIDERS	19
Tool A: “ <i>Hearing Loss, Genetics, and Your Child</i> ”	21
Tool B: “ <i>Learning about Hearing Loss, - A Roadmap for Families</i> ”	24
Tool C: “ <i>Region 4 State Genetic Services Lists & National Resources</i> ”	27
Tool D: “ <i>Questions You May Want to Ask Your Genetics Team</i> ”	29
Tool E: “ <i>Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics, and Hearing Loss for Families</i> ”	31
Tool F: “ <i>Promoting the Genetic Referral</i> ”	35
Tool G: “ <i>Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics, and Hearing Loss for Physicians</i> ”	36
Tool H1: “ <i>Medical Home Care Management for Infants with Confirmed Hearing Loss</i> ”	41
Tool H2: “ <i>Universal Newborn Hearing Screening, Diagnosis, and Intervention Guidelines for Pediatric Medical Home Providers</i> ”	44
Tool H3: “ <i>Hearing Assessment Algorithm within an Office Visit</i> ”	46
Tool H4: “ <i>ACMG ACT Sheet - Congenital Hearing Loss</i> ”	49
Tool H5: “ <i>1-3-6 Primary Care Provider Patient Care Plan</i> ”	62
Tool I: “ <i>Increasing Genetic Referrals for Children Identified Through EHDI - An Action Guide for Providers Serving Children Who Have Hearing Loss</i> ”	65
EHDI GUIDE FEEDBACK FORM	66



Increasing Genetic Referrals for Children Identified Through EHDI

An Action Guide for Providers Serving Children Who Have Hearing Loss

INTRODUCTION

The Region 4 Genetics Collaborative strives to improve outcomes for children who have genetic conditions. Research has shown that more than 50% of hearing loss in infants is genetically related.¹ Of those, about one-third are affected with a complex medical syndrome. In response, the Early Hearing Detection and Intervention (EHDI) Follow-up Workgroup was established and charged with improving access to genetic services for families whose children have been diagnosed with hearing loss through Universal Newborn Hearing Screening (UNHS).

The workgroup engaged in a series of activities to: 1) learn how state EHDI and genetics programs interact; 2) examine the EHDI follow-up system to identify encounters with families that provide opportunities for offering a genetic referral; and 3) explore materials for educating parents and professionals about the importance of a genetic assessment for children diagnosed with hearing loss.

The workgroup developed this guide to help providers increase genetic referrals for children with hearing loss. The guide also includes tools to facilitate appropriate referrals to genetic services.

HOW TO USE THIS ACTION GUIDE

The Action Guide is organized into three sections:

Section One: Guidelines & Opportunities for Genetic Referrals

Service providers involved in the EHDI follow-up system identified guidelines for increasing genetic referrals for children identified through the EHDI system.

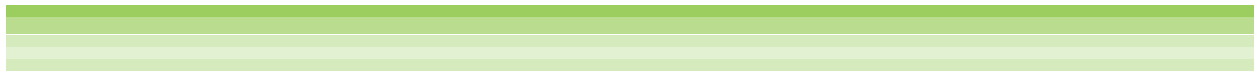
Section Two: Actions for Providers

A list of actions is suggested for each type of service provider that identifies opportunities to increase referrals to genetic services for children diagnosed with hearing loss.

Section Three: Tools for Providers

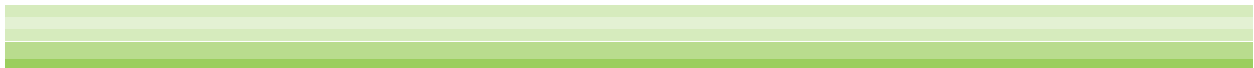
Tools that service providers may use or adapt to increase referrals to genetic services for children who have hearing loss.

¹ Canalis, R. F., & Lambert, P. R. (2000). “The ear: comprehensive otology”. Philadelphia: Lippincott Williams & Wilkins.



SECTION ONE

Guidelines & Opportunities for Genetic Referral



IMPORTANCE OF THE GENETIC REFERRAL

The American College of Medical Genetics (ACMG) recommends that all children with confirmed hearing loss be referred for genetic evaluation and counseling.² The [Joint Committee on Infant Hearing \(JCH\) 2007 Position Statement](#) declares:

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.

Making sure infants with hearing loss receive genetic evaluation and counseling can have many benefits for the baby and family. A genetic evaluation can:

- Help avoid unnecessary and often costly clinical tests to rule out conditions associated with syndromic deafness
- Have health saving or life saving implications – particularly when syndromes with complex medical problems are identified
- Be useful in developing an individualized treatment plan to ameliorate the complications of various syndromes
- In some cases, help predict whether the hearing loss will be progressive. It also may be useful in determining what kind of damage has happened to the hearing system
- Dispel misinformation and ease parental guilt
- Determine recurrence risks for future children to have hearing loss
- Provide information about the possible cause, the chances of recurrence for family planning purposes and appropriate medical care for other family members

A genetic evaluation and consultation are performed 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?

Guideline 1: Families of children diagnosed with hearing loss should receive information about genetics services as a complement to their child's overall health care.

Identification of associated features in hearing loss syndromes may have health-saving or life-saving implications. 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.

Guideline 2: Referral for genetic evaluation should be made after the diagnosis of hearing loss has been confirmed (ideally between 3 to 6 months of age).

Referral should be made by providers working with the family, and may include the medical home provider, otolaryngologist/ear nose and throat (ENT) specialist, otologist, audiologist, ophthalmologist, early interventionist, etc.

TIMELINESS OF THE GENETIC REFERRAL

There are times when making an immediate referral is of utmost importance. 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)

Guideline 3: To maximize the likelihood that the families will follow through with the referral, the message about the importance and benefit of genetic consultation should be provided at every opportunity.

There is always the chance that the parent is not ready for additional information the first time it is offered. Implementing a public health education model that systematically provides information over time and through different venues increases the chance the family will act on the suggestion. Helping the service providers understand the importance of genetic consultation for a child who has hearing loss will help increase the likelihood the family will follow through with the referral.

OPPORTUNITIES FOR GENETIC REFERRAL DURING THE EHDI PROCESS

Guideline 4: All providers should take responsibility for ensuring the genetic referral.

There are typical encounters throughout the EHDI follow-up services system which present opportunities for providers to make referrals for genetic services. The following list identifies the encounter and the most likely provider to have an opportunity to make the referral.

Encounter	Provider
Confirmation of hearing loss	Audiologist
Medical clearance	Otolaryngologist (ENT)/Otologist
Hearing aid fitting & follow-up visits	Pediatric audiologist or Educational audiologist
Routine health care visits	Primary care provider/Pediatrician/Family practitioner
Early intervention visit	System point of entry (SPOE), Part C of IDEA, Title V, and others

SPECIAL CONSIDERATIONS WHEN REFERRING

Sensitivity

Providers must be sensitive to family needs while communicating the importance of the genetic evaluation in assessing for other possible health risks and addressing implications for other family members. Parents/caregivers who are deaf may view hearing loss as a difference, not a disability. Many members of the deaf community feel that there is an inherent and unwarranted bias in the medical profession that views deafness as a disability or needing medical intervention. As professionals, it is important to be culturally sensitive concerning how hearing screening and hearing loss are approached. This includes cultural awareness of how the hearing screening

results may be interpreted by a deaf family. The deaf community is a separate and valued culture in which members are bilingual (communicating in both American Sign Language and English). Family perspective is very important.

- The decision about how to proceed with the evaluation and potential “treatment” of deafness is a personal family matter for the 90% of deaf children who are born to hearing parents, as well as those born to deaf parents.
- The deaf community may not wish to have hearing aids, cochlear implants and/or intervention for their baby who has hearing loss. While this perspective may be more common in parents who are deaf, the view is held by some hearing parents as well.

Consanguinity

Consanguinity, literally meaning common blood, describes a marriage or partnership between two individuals of the same family. Being sensitive and non-judgmental in addressing consanguinity is essential to getting at the important issues of diagnosis, recurrence, and treatment.

- Consanguinity is a cultural norm for some; taking a family history can help identify consanguineous relationships.
- When consanguinity is present, there is a much higher chance for hearing loss in children.³ The children of consanguineous relationships have a significantly higher incidence of autosomal recessive diseases, including hearing impairment.⁴

SECTION ONE RESOURCES

² ACMG statement (2002). “Genetics Evaluation Guidelines for the Etiologic Diagnosis of Congenital Hearing Loss”. *Genetics in Medicine*, 4(3).

³ Turan and Apaydin (2002). “Genetic Sensineural Hearing Loss”. *The Turkish Journal of Ear Nose and Throat*, 9(2).

⁴ Reddy and Rani (2006). “Role of Consanguinity in Congenital Neurosensory Deafness”. *International Journal of Human Genetics*, 6(4).



SECTION TWO

Actions and Strategies for Providers

This section includes specific actions and strategies for the providers serving children in the EHDI follow-up system and their families. Some of the strategies are aimed at working with families or fellow providers, while others are aimed at increasing one's own knowledge and comfort concerning the genetics referral. Each tool mentioned in this section is included in SECTION THREE: Tools for Providers of this Action Guide. Tools may be used as presented or adapted for use.

Actions for EHDI Follow-up Staff

1. At the time of confirmation of hearing loss:

- Include a brochure with genetics and hearing loss information in packets sent to families. *Hearing Loss, Genetics, and Your Child* (Tool A).
- Provide information to help the family navigate services during the first year of their child's life with hearing loss. *Learning about Hearing Loss – A Roadmap for Families* (Tool B).
- Provide the family with contact information for local genetic services. *Region 4 State Genetic Services Lists & National Resources to Identify Services Near You* (Tool C).
- Include a prompt in the client record to remind you to offer a genetic referral.

2. At the time of follow-up contacts:

- Ask the family if they have had a genetic consultation.

If the answer is “no”:

- Offer to assist the family in obtaining a genetic referral. Some third-party insurance providers have requirements about who may make the referral in order for the service to be a covered benefit. Provide the family with a referral form and information to take to their medical home or primary care provider.
- Offer the family printed information about the genetic consultation. *Questions You May Want to Ask Your Genetics Team* (Tool D).
- Provide the family with contact information for local genetic services. *Region 4 State Genetic Services Lists & National Resources to Identify Services Near You* (Tool C).
- Provide information to help the family navigate services during the first year of their child's life with hearing loss. *Learning about Hearing Loss – A Roadmap for Families* (Tool B).
- Include a prompt in the client record to remind you to follow up at a later date to see if the family has had a genetic consultation.

When discussing the genetic referral with a family, they may have questions and concerns about *why* the consultation is important, and what they should expect if they choose to see a genetic counselor. Helping the family understand why it is important will increase the chances they will obtain a genetic consultation.

3. Be prepared to respond to questions about the importance of a genetic consultation for a family who has a child diagnosed with hearing loss:

- View the frequently asked questions (FAQs) for families on the Region 4 website. *Frequently Asked Questions about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Provide the family with the link to the FAQs, or print a hard copy to share with the family *Frequently Asked Questions about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Review the brochure *Hearing Loss, Genetics, and Your Child* (Tool A).
- Offer the family written information about genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).

4. Be prepared to respond to questions about *what to expect* from the genetic consultation:

- View the frequently asked questions on the Region 4 website. *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Provide the family with the website to the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* or print a hard copy to share with the family (Tool E).
- Review the brochure, *Questions You May Want to Ask Your Genetics Team* (Tool D).
- Offer the family printed information about the genetic consultation. *Questions You May Want to Ask Your Genetics Team* (Tool D).

To maximize the likelihood that the family will follow through with the referral, the message about the importance and benefit of genetic consultation should be provided at every opportunity. There is always the chance that the parent is not ready for additional information the first time it is offered. Implementing a public health education model that systematically provides information over time and through different venues increases the chance the family will act on the suggestion. Helping the service providers understand the importance of genetic consultation for a child who has hearing loss will help increase the likelihood the family will follow through with the referral.

5. Share this Guide with members of the care team and other providers when coordinating care:

- Making a referral;
- Sharing patient records, and/or;
- Working with the medical home or primary care provider to obtain a referral for a genetic consultation.

6. When planning staff or peer training and professional development:

- Review the web-based tutorial *Promoting the Genetic Referral* on how, why, and when to refer for genetic services (Tool F).
- Use the web-based tutorial *Promoting the Genetic Referral* on how, why and when to refer for genetic services as a standalone training or incorporate into presentations, training sessions, workshops (Tool F).

Actions for Pediatric Audiologists/Otolaryngologist (ENT)/Otologists

1. At the time of confirmation of hearing loss:

- Offer the family a genetic referral. Some third-party insurance providers have requirements about who may make the referral in order for the service to be a covered benefit. If you cannot make the referral directly, provide the family with detailed case history information, results of the diagnostic evaluation and a referral form to take to their medical home or primary care provider.
- Give the family written information on genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).
- Provide information to help the family navigate services during the first year of their child's life with hearing loss. *Learning about Hearing Loss – A Roadmap for Families* (Tool B).
- Provide the family with contact information for local genetic services. *Region 4 State Genetic Services Lists & National Resources to Identify Services Near You* (Tool C).
- Include a prompt in your standard office visit planner to remind you to follow-up at a later date to see if the family has had a genetic consultation.

2. At the time of hearing aid fitting and all follow-up visits:

- Ask the family if they have had a genetic consultation.

If the answer is “no”:

- Offer the family a genetic referral. Some third-party insurance providers have requirements about who may make the referral in order for the service to be a covered benefit. If you cannot make the referral directly, provide the family with recent test results and a referral form or letter to take to their medical home or primary care provider.
- Give the family written information on genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).
- Provide information to help the family navigate services during the first year of their child's life with hearing loss. *Learning about Hearing Loss – A Roadmap for Families* (Tool B).
- Provide the family with contact information for local genetic services. *Region 4 State Genetic Services Lists & National Resources to Identify Services Near You* (Tool C).
- Include a prompt in your standard office visit planner to remind you to follow up at a later date to see if the family has had a genetic consultation.

3. Increase your knowledge of genetics and hearing loss and comfort in referring to genetic services:

- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Physicians* (Tool G).
- View the web-based module on how, why, and when to refer for genetic services. *Promoting the Genetic Referral* (Tool F).

When discussing the genetic referral with a family, they may have questions and concerns about both *why* the consultation is important and, if they choose to see a genetic counselor, *what to expect*. Helping the family understand why it is important will increase the chances they will choose to obtain a genetic consultation.

4. Be prepared to respond to questions families may have about the importance of a genetic consultation for a family who has a child with a permanent hearing loss:

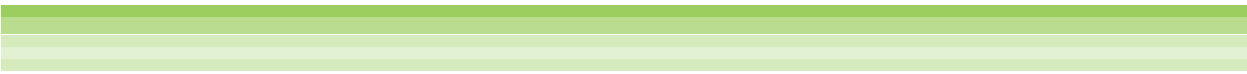
- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Provide the family with the website to the FAQs for families or print out a hard copy for the family to take with them *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Physicians* on the Region 4 website (Tool G).
- Review the web-based tutorial on how, why, and when to refer for genetic services. *Promoting the Genetic Referral* (Tool F).
- Provide the family with written information on genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).

5. Be prepared to respond to families' questions about *what to expect* from the genetic consultation:

- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Provide the family with the website to the FAQs for families or print out a hard copy for the family to take with them *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Physicians* on the Region 4 website (Tool G).
- Provide the family written information on the genetic consultation. *Questions You May Want to Ask Your Genetics Team* (Tool D).
- Provide the family with written information on genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).
- Review the web-based tutorial on how, why, and when to refer for genetic services. *Promoting the Genetic Referral* (Tool F).

6. Facilitate coordinated care:

- Provide a diagnostic report describing the hearing loss (type, degree, and configuration) and tests performed (tympanometry, auditory brain response, oto-acoustic emissions, etc.) to the medical home or primary care provider, and make a genetic referral or explain the need for a referral.
- Offer to provide the complete diagnostic report to the genetics clinic.



To maximize the likelihood that the family will follow through with the referral, the message about the importance and benefit of genetic consultation should be provided at every opportunity. There always is the chance that the parent is not ready for additional information the first time it is offered. Implementing a public health education model that systematically provides information over time and through different venues increases the chance the family will act on the suggestion. Helping the service providers understand the importance of genetic consultation for a child diagnosed with hearing loss will help increase the likelihood the family will follow through with the referral.

7. Share this Action Guide with members of the care team and other providers when

- Coordinating care
- Making a referral
- Sharing patient records
- Working with the medical home or primary care provider to obtain a referral for a genetic consultation

8. When planning staff or peer training and professional development:

- Review the web-based tutorial *Promoting the Genetic Referral* on how, why, and when to refer for genetic services (Tool F).
- Use the web-based tutorial *Promoting the Genetic Referral* on how, why and when to refer for genetic services as a standalone training or incorporate into presentations, training sessions, workshops (Tool F).

Actions for the Primary Care Provider/Pediatrician/ Family Practitioner/Medical Home

1. At the time of routine health screenings/office visits:

- Ask the family if they have had a genetic consultation.

If the answer is “no”:

- Offer the family a genetic referral.
- Give the family written information on genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).
- Provide information to help the family navigate services during the first year of their child’s life with hearing loss. *Learning about Hearing Loss – A Roadmap for Families* (Tool B).
- Provide the family with contact information for local genetic services. *Region 4 State Genetic Services Lists & National Resources to Identify Services Near You* (Tool C).
- Include a prompt in your standard office visit planner to remind you to follow up at a later date to see if the family has had a genetic consultation.

2. Increase your knowledge of genetics and hearing loss and your comfort in referring to genetic services:

- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Physicians* on the Region 4 website (Tool G).
- Review the web-based tutorial on how, why, and when to refer for genetic services *Promoting the Genetic Referral* (Tool F).

When discussing the genetic referral with a family, they may have questions and concerns about both *why* the consultation is important and, if they choose to see a genetic counselor, *what to expect*. Helping the family understand why it is important will increase the chances they will choose to obtain a genetic consultation.

3. Be prepared to respond to questions families may have about the importance of a genetic consultation for a family who has a child with a permanent hearing loss:

- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Provide the family with the website to the FAQs for families or print out a hard copy for the family to take with them *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Physicians* on the Region 4 website (Tool G).
- Review the web-based tutorial on how, why, and when to refer for genetic services. *Promoting the Genetic Referral* (Tool F).
- Provide the family with written information on genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).

4. Be prepared to respond to families' questions about *what to expect* from the genetic consultation:

- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Provide the family with the website to the FAQs for families or print out a hard copy for the family to take with them *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Physicians* on the Region 4 website (Tool G).
- Provide the family written information on the genetic consultation. *Questions You May Want to Ask Your Genetics Team* (Tool D).
- Provide the family with written information on genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).
- Review the web-based tutorial on how, why, and when to refer for genetic services *Promoting the Genetic Referral* (Tool F).

5. Facilitate coordinated care:

- When referring, provide a diagnostic report describing the hearing loss (type, degree, and configuration) and tests performed (tympanometry, auditory brain response, oto-acoustic emissions, etc.) to the specialist or other care provider, and recommend to the provider the need for offering the family a genetic referral.
- Offer to provide the complete diagnostic report to the genetics clinic.

To maximize the likelihood that the family will follow through with the referral, the message about the importance and benefit of genetic consultation should be provided at every opportunity. There always is the chance that the parent is not ready for additional information the first time it is offered. Implementing a public health education model that systematically provides information over time and through different venues increases the chance the family will act on the suggestion. Helping the service providers understand the importance of genetic consultation for a child who has hearing loss will help increase the likelihood the family will follow through with the referral.

6. Share this Action Guide with members of the care team and other providers when:

- Coordinating care
- Making a referral
- Sharing patient records
- Working with the provider to obtain a referral for a genetic consultation

7. When planning staff or peer training and professional development:

- Review the web-based tutorial *Promoting the Genetic Referral* on how, why, and when to refer for genetic services (Tool F).
- Use the web-based tutorial *Promoting the Genetic Referral* on how, why and when to refer for genetic services as a standalone training or incorporate into presentations, training sessions, workshops (Tool F).

8. Establish referral standards for your practice that include common resources:

- Adopt one of the care management tools as standard procedure for your practice:
 1. “Medical Home Care Management for Infants with Confirmed Hearing Loss” (Tool H1).
 2. “Universal Newborn Hearing Screening, Diagnosis and Intervention Guidelines for Pediatric Medical Home Providers” (Tool H2).
 3. “Hearing Assessment Algorithm within an Office Visit” (Tool H3).
 4. “ACMG ACT Sheet—Congenital Hearing Loss” (Tool H4).
 5. “1-3-6 Primary Care Provider Patient Care Plan” (Tool H5).
- Share this Action Guide, Increasing Genetic Referrals for Children Identified through EHDI - an Action Guide for Providers Serving Children who have Hearing Loss, or the website (www.region4genetics.org/region4_products/genetics_hearingloss.aspx) for electronic retrieval throughout your practice. Consider adapting the information and tools for your practice (Tool I).

Actions for Early Intervention Services Providers

1. At the time of intake and all follow-up contacts:

- Ask the family if they have had a genetic consultation.

If the answer is “no”:

- Offer the family a genetic referral. Some third-party insurance providers have requirements about who may make the referral in order for the service to be a covered benefit. Provide the family with a referral form and information to take to their medical home or primary care provider.
- Give the family written information on genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).
- Provide information to help the family navigate services during the first year of their child’s life with hearing loss. *Learning about Hearing Loss – A Roadmap for Families* (Tool B).
- Provide the family with contact information for local genetic services. *Region 4 State Genetic Services Lists & National Resources to Identify Services Near You* (Tool C).
- Include a prompt in your visit planner to remind you to follow up at a later date to see if the family has had a genetic consultation.

2. Increase your knowledge of genetics and hearing loss, and your comfort in referring to genetic services:

- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Review the web-based tutorial on how, why, and when to refer for genetic services *Promoting the Genetic Referral* (Tool F).

When discussing the genetic referral with a family, they may have questions and concerns about both *why* the consultation is important and, if they choose to see a genetic counselor, *what to expect*. Helping the family understand why it is important will increase the chances they will choose to obtain a genetic consultation.

3. Be prepared to respond to questions families may have about the importance of a genetic consultation for a family who has a child with a permanent hearing loss:

- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Provide the family with the website to the FAQs for families or print out a hard copy for the family to take with them *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Review the web-based tutorial on how, why, and when to refer for genetic services *Promoting the Genetic Referral* (Tool F).
- Provide the family with written information on genetics and hearing loss. *Hearing Loss, Genetics, and Your Child* (Tool A).

4. Be prepared to respond to families' questions about *what to expect* from the genetic consultation:

- View the *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Provide the family with the website to the FAQs for families or print out a hard copy for the family to take with them *Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics and Hearing Loss for Families* (Tool E).
- Provide the family written information on the genetic consultation. *Questions You May Want to Ask Your Genetics Team* (Tool D).
- Review the web-based tutorial on how, why, and when to refer for genetic services *Promoting the Genetic Referral* (Tool F).

To maximize the likelihood that the family will follow through with the referral, the message about the importance and benefit of genetic consultation should be provided at every opportunity. There always is the chance that the parent is not ready for additional information the first time it is offered. Implementing a public health education model that systematically provides information over time and through different venues increases the chance the family will act on the suggestion. Helping the service providers understand the importance of genetic consultation for a child who has hearing loss will help increase the likelihood the family will follow through with the referral.

5. Share this Action Guide with members of the care team and other providers when:

- Coordinating care
- Making a referral
- Sharing patient records
- Working with the medical home or primary care provider to obtain a referral for a genetic consultation.

6. When planning staff or peer training and professional development:

- Review the web-based tutorial *Promoting the Genetic Referral* on how, why, and when to refer for genetic services (Tool F).
- Use the web-based tutorial *Promoting the Genetic Referral* on how, why, and when to refer for genetic services as a stand-alone training or incorporate into presentations, training sessions, workshops (Tool F).



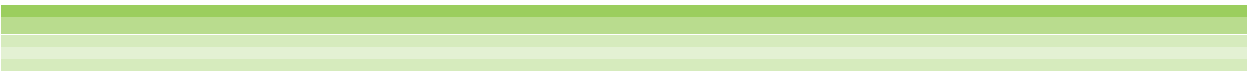
SECTION THREE

Tools for Providers

All of the tools provided in this Action Guide also are available on the Region 4 website:
http://region4genetics.org/region4_products/genetics_hearingloss.aspx

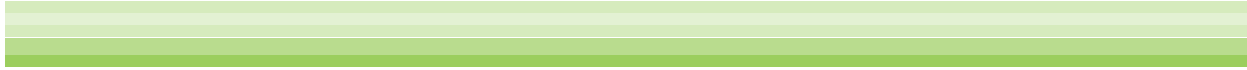
INDEX

- A. "*Hearing Loss, Genetics, and Your Child*". A brochure for families developed by the American College of Medical Geneticists (ACMG) and the National Coordinating Center for the Regional Genetic Collaboratives.
- B. "*Learning about Hearing Loss – A Roadmap for Families*". A brochure to guide families through the service system from diagnosis of permanent hearing loss through the first year; includes space to record important appoints and contacts.
- C. "*Region 4 State Genetic Services Lists & National Resources*"
- D. "*Questions You May Want to Ask Your Genetics Team*". A brochure describing what to expect at a genetics consultation for a child who has hearing loss. This brochure includes a form to help parents prepare for the consultation. Developed by the Center for Disease Control.
- E. "*Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics, and Hearing Loss for Families*". A web-based resource to provide information for families located on the Region 4 website. www.region4genetics.org/
- F. "*Promoting the Genetic Referral*". A web-based PowerPoint tutorial on how, why and when to refer for genetic services for health care providers; may be used as a stand-alone learning tool or incorporated into other training, workshop, or grand-rounds presentations. Available on the Region 4 website www.region4genetics.org/
- G. "*Frequently Asked Questions (FAQs) about Newborn Hearing Screening, Genetics, and Hearing Loss for Physicians*". A web-based resource to provide information for physicians located on the Region 4 website www.region4genetics.org/
- H. Care Management Tools:
 - H1. "*Medical Home Care Management for Infants with Confirmed Hearing Loss*". Adapted from a document developed by the American Academy of Pediatrics (AAP) and the National Center for Hearing Assessment and Management (NCHAM).
 - H2. "*Universal Newborn Hearing Screening, Diagnosis, and Intervention Guidelines for Pediatric Medical Home Providers*". Developed by the AAP.
 - H3. "*Hearing Assessment Algorithm within an Office Visit*". AAP, Copyright 2009.
 - H4. "*ACMG ACT Sheet – Congenital Hearing Loss*". Developed by the ACMG, the ACTION (ACT) sheet describes the short term actions a health professional should follow in communicating with the family and determining the appropriate steps in the follow up of the infant that has screened positive.
 - H5. "*1-3-6 Primary Care Provider Patient Care Plan*". Adapted from a document developed by the EHDI program at the Indiana State Department of Health.
- I. "*Increasing Genetic Referrals for Children Identified Through EHDI – An Action Guide for Providers Serving Children Who Have Hearing Loss*". Available at www.region4genetics.org.



Tool A

“Hearing Loss, Genetics, and Your Child”



What should I know about genetics, genes and hearing loss?

- Genetics is the study of genes. Genes are in nearly every cell of your body.
- Your child inherited, or got, his or her genes from both of his or her natural parents.
- Genes are like recipes. Each gene tells different body parts how to grow and how to work.
- If some genes are changed even a little bit, they may change how parts of your body work.
- Genetic hearing loss in a family can often be a surprise. A person can have a genetic hearing loss even if no one in his or her family has hearing loss. Therefore, it is important to make an appointment with a genetics doctor.
- Scientists have found many different genes that can affect the way ears work. Changes in some of these genes will affect other parts of the body, too.
- Scientists have not found every gene that can cause hearing loss. They are still doing research to learn about other new hearing loss genes.
- An examination by a genetics doctor and genetic testing may not be able to find the exact cause of your child's hearing loss. But it is important to check.

Will insurance pay for genetic testing?

- Medicaid will pay for genetic testing in some states. Many other insurance plans may also pay. You need to check your own plan or state Medicaid, to be sure.
- If you are in a managed care plan (HMO), you will probably need to talk to your child's primary doctor about a genetics referral first.

Local contact information:



National Coordinating Center for the
Regional Genetic and Newborn Screening
Service Collaborators

www.nccrcg.org

The development of this brochure was partially funded by U22MC03957, awarded as a cooperative agreement between the Maternal and Child Health Bureau/Health Resources and Services Administration, Genetic Services Branch, and the American College of Medical Genetics.



American College of Medical Genetics
Medical Genetics: Translating Genes into Health®

www.ACMG.net

Hearing loss, genetics and Your Child

A Brochure for Parents of a Child
with Hearing Loss



National Coordinating Center for the
Regional Genetic and Newborn Screening
Service Collaborators

Now that I know my child has a hearing loss, what should I do next?

- Ask your child's doctor for a referral to a genetics doctor to learn why your child has a hearing loss.
- The genetics doctor is one of many doctors your child may need to see.
- It is also important that your visits to your child's audiologist continue.

Why should I have a genetics doctor see my child?

- Knowing that your child has a hearing loss is different than knowing how the hearing loss happened.
- There is about a 50% chance that your child's hearing loss is due to a genetic cause.
- Learning the cause of your child's hearing loss can help doctors understand the complete picture of your child's health.
- Most children with hearing loss do not have health problems in the rest of their body, but a few do. Genetic testing may help doctors find these problems.
- The genetics doctor might be able to help you make decisions about your child's health care.
- Some children's hearing changes as they grow older. Doctors might be able to figure out if your child's hearing loss will stay the same.
- You could find out the chance that your other children will have a hearing loss.



How can I prepare for the genetics appointment?

You and your child may be seen by a medical genetics team, with a genetics doctor (also called a medical geneticist) and a genetic counselor or nurse. You will get the most out of your visit if you write down your questions and bring them with you. You should also plan to answer questions about the information listed below:

- the health of your child's family members, including brothers and sisters, parents, grandparents, aunts and uncles, and cousins.
- any family members who have problems with balance, heart, diabetes, kidneys, hair, skin, face and eyes. The geneticist will also ask about hearing loss in other family members.
- the pregnancy and birth of your child, including infections and other problems.

Genetics is not just about families. It is also about health.



What happens at the genetics appointment?

- The genetics doctor will review your child's health records with you.
- A genetics doctor will talk to you and examine your child. The doctor will review medical conditions and history of hearing loss in the family.
- The genetics doctor may test your child for previous infection by taking a blood sample.
- The genetics doctor might offer to test for common genes that cause deafness.
- You will have time to ask questions.
- You may need to come back for another visit to discuss the test results with the genetics doctor.
- The genetics doctor will look at all the information and try to figure out if your child's hearing loss is due to a genetic cause.
- This information will help you prepare for your child's future health needs.
- The genetics doctor will send a letter to your child's doctor. Your child's doctor will work with the genetics team to determine if any follow-up is needed.





Tool B

*“Learning about Hearing Loss – A Roadmap
for Families”*


Learning about Hearing Loss — A Roadmap for Families

From Diagnosis of Permanent Hearing Loss Through the First Year

Diagnosis of Hearing Loss	During the first two weeks	Within the first month
<p><input type="checkbox"/> Evaluation completed by an audiologist experienced in working with infants and children. (Babies over four months old may need sedation.) Date of diagnosis: ___/___/___</p> <p>Hearing Loss Diagnosis: Left Ear: <input type="checkbox"/> mild <input type="checkbox"/> moderate <input type="checkbox"/> severe <input type="checkbox"/> profound _____</p> <p>Right Ear: <input type="checkbox"/> mild <input type="checkbox"/> moderate <input type="checkbox"/> severe <input type="checkbox"/> profound _____</p> <p>Your audiologist will connect you to: <input type="checkbox"/> Your local early intervention services provided through your local public school district and community partners. <input type="checkbox"/> Other families of children with hearing loss for parent-to-parent support. _____ <input type="checkbox"/> Your audiologist will inform your child's primary care provider and the state Department of Health about your child's hearing loss.</p>	<p><input type="checkbox"/> Learn more about your child's hearing loss. Visit www.babyhearing.org</p> <p><input type="checkbox"/> Discuss your child's hearing loss with your health care provider at your child's next well child checkup or sooner.</p> <p><input type="checkbox"/> Your child may need to return to the audiologist to finish further testing. Place _____ Date ___/___/___ Time _____</p> <p><input type="checkbox"/> You will be contacted by the following:</p> <ul style="list-style-type: none"> • The state Department of Health's Early Hearing Detection and Intervention Program will send a packet of information. • A representative from your local Early Intervention services will contact your family. <p><input type="checkbox"/> Get to know other resources that are available for your child and family through your regional Deaf and Hard of Hearing Services Division.</p>	<p><input type="checkbox"/> Your health care provider will help you coordinate referrals to recommended specialists who have experience working with young children, including a geneticist, otolaryngologist (ear, nose and throat specialist), and ophthalmologist.</p> <p><input type="checkbox"/> Evaluation by an ENT specialist. This will include the medical clearance for hearing aids (if chosen): Place: ___/___/___ Date ___/___/___ Time _____</p> <p><input type="checkbox"/> Evaluation by a geneticist: Place: ___/___/___ Date ___/___/___ Time _____</p> <p><input type="checkbox"/> Evaluation by an ophthalmologist: Place: ___/___/___ Date ___/___/___ Time _____</p> <p><input type="checkbox"/> Return to your audiologist for hearing aid fitting (if chosen). Information about loaner hearing aids will be provided: Place: ___/___/___ Hearing aid fitting date ___/___/___ Time _____</p> <p><input type="checkbox"/> Begin to learn about different choices in hearing technology and ways to communicate.</p> <p><input type="checkbox"/> With parental consent, an eligibility evaluation will begin through your local early intervention services. Most children with permanent hearing loss will be eligible to participate in these family-focused services. Your early intervention team may include a teacher for deaf and hard of hearing, an audiologist, an early childhood special education teacher, social service providers, health care providers, and family members.</p>

Learning about Hearing Loss — A Roadmap for Families

From Diagnosis of Permanent Hearing Loss Through the First Year

Within the first two months	Within the first six months	Within the first year
<p><input type="checkbox"/> Discuss your child's hearing loss with your health care provider at your child's next well child check up or sooner.</p> <p><input type="checkbox"/> Continue audiology care. Return to your audiologist for tests of your child's hearing aids (if chosen). Discuss a plan with your audiologist in case your child's hearing aids are lost or not working.</p> <p>PLAN: _____</p> <p>_____</p> <p><input type="checkbox"/> After an evaluation is completed through early intervention services, an Individualized Family Service Plan (IFSP) will be completed for eligible children and their families. This plan will include outcomes, services and supports for your family based on identified strengths and needs.</p> <p>IFSP start date ___/___/___</p> <p><input type="checkbox"/> Continue to connect with other families of children with hearing loss and adults who are deaf or hard of hearing.</p>	<p><input type="checkbox"/> Your health care provider will help you coordinate referrals to recommended specialists with young children, including ENT, genetics, and ophthalmology (if not yet completed).</p> <p><input type="checkbox"/> Evaluation by a pediatric ophthalmologist: Place: _____ Date ___/___/___ Time _____</p> <p><input type="checkbox"/> Continue audiology care. Audiology evaluation may include behavioral testing. Date ___/___/___ Time _____</p> <p><input type="checkbox"/> Continue services provided through the early intervention services as described in your child's IFSP.</p> <p><input type="checkbox"/> Continue to learn about and discuss communication considerations and choices in hearing technology for your child.</p>	<p><input type="checkbox"/> Your health care provider will help you coordinate referrals to recommended specialists who have experience working with young children, including ENT, genetics, and ophthalmology (if not yet completed).</p> <p><input type="checkbox"/> Evaluation by a geneticist: Place: _____ Date ___/___/___ Time _____</p> <p><input type="checkbox"/> Ongoing audiology care and hearing aid evaluation at least every six months.</p> <p><input type="checkbox"/> Continue services provided through the early intervention services as described in your child's IFSP.</p> <p><input type="checkbox"/> Continue to learn about and discuss communication considerations and choices in hearing technology for your child.</p> <p><input type="checkbox"/> Continue to connect with other families of children with hearing loss and adults who are deaf or hard of hearing.</p>
<p>Contact Information</p> <p>Health Care Provider: _____</p> <p>Audiologist: _____</p> <p>Ophthalmologist: _____</p> <p>Geneticist: _____</p>		<p>Early Intervention: _____</p> <p>Service Coordinator: _____</p> <p>Teacher: _____</p> <div style="text-align: right;">  <p>Region4 Genetics Collaborative www.region4genetics.org</p> </div>



Tool C

“Region 4 State Genetic Services Lists & National Resources”

State resources are web-based only.

Please access this information at:

*[www.region4genetics.org/region4_products/
genetics_hearingloss.aspx](http://www.region4genetics.org/region4_products/genetics_hearingloss.aspx)*

NATIONAL RESOURCES

The following resources can help you locate a genetics professional in your area online or over the phone:

- GeneTests' Clinic Directory is a voluntary listing of U.S. and international genetics clinics providing genetic evaluation and genetic counseling. Clinics can be chosen by state or country, by service, and/or by clinic name; identifying for families the most conveniently located genetics clinics providing services that they need. State maps can help locate a clinic in a specific geographic area.
<http://www.ncbi.nlm.nih.gov/sites/GeneTests/clinic?db=GeneTests> or call (206) 616-4089
- The National Society of Genetic Counselors, Inc. offers a searchable directory of genetic counselors in the United States. You can search by location, name, area of practice/specialization, and/or ZIP Code. <http://www.nsgc.org/resourcelink.cfm> or call (312) 321-6834

The Utah State University houses the National Center for Hearing Assessment and Management (NCHAM). NCHAM serves as the National Resource Center for the implementation and improvement of comprehensive and effective Early Hearing Detection and Intervention (EHDI) systems. As a multidisciplinary Center, their goal is to ensure that all infants and toddlers with hearing loss are identified as early as possible and provided with timely and appropriate audiological, educational, and medical intervention.

For resources, tools and more information, please visit their website:

<http://www.infanthearing.org/>



Tool D

*“Questions You May Want to Ask
Your Genetics Team”*



Tool E

*“Frequently Asked Questions (FAQs) about
Newborn Hearing Screening, Genetics, and
Hearing Loss for Families”*

Frequently Asked Questions for Families about Newborn Hearing Screening, Hearing Loss, and Referral to Genetic Services

What is congenital hearing loss?

Congenital means that a person was born with the hearing loss.

How is congenital hearing loss identified?

Babies born with a hearing loss can be identified through a newborn hearing screening test. The test is generally done before the baby leaves the hospital. All infants who do not pass the newborn hearing screening should be referred for medical testing to rule out or confirm hearing loss.

What causes hearing loss?

Some infants are born with hearing loss. This is called congenital hearing loss. Genetic factors – changes in genes – are thought to cause more than 50% of all incidents of congenital hearing loss. Congenital hearing loss that is not hereditary may be the result of prenatal infections, illnesses, toxins consumed by the mother during pregnancy or other conditions occurring at the time of birth or shortly thereafter.

What is acquired hearing loss?

Acquired hearing loss is a hearing loss which appears after birth, at any time in one's life, as a result of a disease, a condition, or an injury. There are many hereditary diseases and syndromes that can lead to hearing loss.

What is a gene?

Genes are a packet of information that tell our bodies to do the things they are supposed to do, such as for an ear to hear.

How is hearing loss genetic?

Genetic hearing loss occurs when genes which cause hearing loss are passed through the family. There are about 20,000-25,000 genes in our bodies. Genes come in pairs. One member of each pair is inherited from the child's mother, and one from the child's father. At least 100 genes play a role in hearing. Sometimes, a small change occurs within a gene which may be harmful. A change in one of the many genes controlling the functioning of the ear may lead to hearing loss.

Is my child's hearing loss genetic?

At least half (50%) of all children with inner ear (sensorineural or "nerve-based") hearing loss have a genetic cause for their hearing loss. Sensorineural/nerve-based hearing loss is likely to be genetic if:

- Hearing loss is present at birth or in early childhood.
- Hearing loss is related with other health problems the child has.
- Hearing loss affects both ears.
- Hearing loss is severe to profound.
- There is a family history of hearing loss.

How can my child's hearing loss be genetic if no one else in the family has hearing loss?

Your child's hearing loss may be genetic, even if no other people in your family have hearing loss. A small change in even one of the many genes which control the functioning of the ear may lead to hearing loss. Ninety percent of babies with hearing loss are born to parents who can hear. This can happen when:

- Both parents are silent carriers of a hearing loss gene.
- Your child's hearing loss is the result of a new gene mutation or change that is not present in either parent.
- For some genetic conditions hearing loss may be only one of many symptoms. You may discover that some family members have other features of the diagnosed condition, but no hearing loss.

What does "carrier" or "silent carrier" of hearing loss mean?

A carrier of a genetic condition is someone who has a change in one of his or her genes but does not show symptoms of the condition. People who are a carrier of a genetic condition can pass the condition on to their children. So, it is possible to have a child with genetic hearing loss, even when parents are hearing and have no history of hearing loss.

Why is it important to find out if my child's hearing loss is genetic?

Genetic testing can determine the exact cause of your child's hearing loss. In some cases, knowing the cause of your child's hearing loss may:

- Help you and your child's doctor determine the best treatment and long-term medical management for your child.
- Provide you with information about your child's future hearing.
- Alert you to other problems you should look for.
- Tell you if other family members might develop hearing loss.
- Provide family members with information about the chances that they may have a child with the same condition.

How do I find out if my child has a genetic hearing loss?

Make an appointment to have your child seen at a genetics clinic. Your child's doctor, audiologist or an early intervention service provider can help you make the appointment and collect the medical information needed.

What is genetic testing?

Genetic testing is the process by which the more common genes related to hearing loss are analyzed in a laboratory. A small sample of blood or other tissue (skin or cheek cells) is needed for analysis.

What happens during a visit with a geneticist and genetic counselor?

As parents, you will be asked many questions about your child's medical history, hearing loss and family history. In addition, the counselor will review your child's medical records.

Next, the geneticist will examine your child for physical features related to syndromic forms of hearing loss. Sometimes, this initial evaluation will provide enough information to establish a diagnosis. More often, however, the geneticist will recommend additional tests or evaluations – such as an eye exam or lab work for genetic testing – to get more information.

Once these evaluations are completed, the geneticist and genetic counselor will schedule another meeting with you to discuss the test results and any possible diagnoses. If a specific diagnosis is established, the geneticist and genetic counselor will explain the details, including specific causes, the kind of genetic testing that is available, the prognosis (what you can expect for your child's future) and recurrence risks (chances of it occurring in future pregnancies).

What is syndromic hearing loss?

Syndromic means that the hearing loss is just one part of a condition that may have many other features. These other features can be:

- Characteristics that make a child look different, like eyes that are different colors.
- Symptoms that you can't see, like kidney or heart problems.

What happens if the genetic specialists can't determine the exact cause of my child's hearing loss?

Sometimes, a genetic evaluation and other studies can't immediately determine the exact cause of your child's hearing loss. If this is the case, doctors will continue to monitor your child and treat her for any medical issues she may develop, such as vision problems or hypothyroidism. In addition, scientists continue to develop new genetic tests for hearing loss, so it's important to maintain contact with your geneticist and genetic counselor so you can learn about new testing as it becomes available.

If a diagnosis is established for my child's hearing loss, how can I contact other parents whose children have the same diagnosis?

Your geneticist and genetic counselor can give you information about local and national parent support groups associated with your child's diagnosis. You also can find additional information on this website under Family Support Resources.

Where can I get medical consultation and genetic counseling services?

http://region4genetics.org/medical_home_resources/genetic_specialist_map.aspx



Tool F

“Promoting the Genetic Referral”

This tool is web-based only.

Please access this tutorial at

*[www.region4genetics.org/region4_products/
genetics_hearingloss.aspx](http://www.region4genetics.org/region4_products/genetics_hearingloss.aspx)*



Tool G

*“Frequently Asked Questions (FAQs) about
Newborn Hearing Screening, Genetics, and
Hearing Loss for Physicians”*

Frequently Asked Questions about Newborn Hearing Screening, Hearing Loss, and Genetics for Physicians

Preparing the physician to respond to a family's questions and concerns

Why is newborn hearing screening important?

Universal newborn hearing screening is the standard of care for hospitals and birthing centers in the United States. Currently, more than 90% of all newborns are screened before hospital discharge.

Universal Newborn Hearing Screening means that all newborns are screened at birth regardless of the presence of a risk factor. Approximately 3 out of every 1,000 infants are born with congenital hearing loss, making it the most common birth defect. Newborn hearing screening is important since it allows for early detection of hearing loss and subsequently allows appropriate intervention to occur for language and communication development.

Moderate to profound hearing loss in early infancy has been shown to be associated with delayed language and communication development. Children who are identified early and receive intensive early intervention perform as much as 20-40 percentile points higher on school related measures than children who do not receive such intervention.

(Retrieved from: <http://mchb.hrsa.gov/programs/specialneeds/unhs.htm>)

What tests are used for newborn hearing screening?

Currently, there are two tests that hospitals use for newborn hearing screening: Auditory Brainstem Response (ABR) and Otoacoustic Emissions (OAE). These tests may be used alone or in combination. Hearing screening is fully automated and requires no participation from the baby.

How does Auditory Brainstem Response (ABR) work?

Auditory brainstem response (ABR) measures the electroencephalographic waves generated in response to clicks via three electrodes placed on the baby. ABR results are averaged indicating a pass or refer response. The test is optimal when the baby is well fed and asleep, it takes only a few minutes to complete for each ear.

How does Otoacoustic Emission (OAE) work?

Otoacoustic emission (OAE) emits tones and records responses to natural sounds generated by cochlear hair cells. OAE's are recorded with a small probe that is placed in the baby's ear canal. OAE's can be affected by debris or fluid in the external and middle ear, resulting in the potential need for re-screening.

What happens if the infant does not pass the Universal Newborn Hearing Screen?

If the infant does not pass the newborn hearing screen, a second screen must be ordered. It is recommended that a second screening be conducted prior to discharge from the hospital whenever possible or in accordance with each state's legislation. If the second screening is not conducted before the infant leaves the hospital, it should occur on an outpatient basis within 1 month of age per Joint Committee on Infant Hearing (JCIH) 2007 Position Statement.

What follow-up is recommended if the newborn does not pass the hearing screening?

Generally if a newborn does not pass the hearing screening at birth, a second screening is conducted. If the newborn does not pass the second screening then outpatient follow-up testing is indicated with an audiology provider. The primary care provider may need to make a referral to the audiology provider. Most state health departments have a listing of providers that can evaluate young infants and babies. The follow-up testing includes a number of diagnostic tests that would determine if hearing is normal or if hearing loss is present. Diagnostic testing reveals individual, ear-specific information and allows the type and degree of hearing loss to be determined.

Additionally, the family may be referred to the statewide Early Hearing Detection and Intervention (EHDI) program as well as the Early Intervention (EI) program. Each of these programs may have additional assessments that are performed. This can vary by state, please check with your state for state specific information regarding EHDI and or EI.

Are risk factors important?

Universal Newborn hearing screening means that all babies are screened at birth regardless of risk factors. The Joint Committee on Infant Hearing (JCIH) has a 2007 position statement that lists the risk factors. To see the list, click this link:

<http://pediatrics.aappublications.org/cgi/reprint/120/4/898>

What is considered normal hearing?

Normal hearing thresholds are determined by a variety of diagnostic audiologic tests including Auditory Brainstem Response (ABR), Otoacoustic Emissions (OAE), pure tone air and bone conduction testing, immittance testing and/or Auditory Steady State Response (ASSR).

Hearing loss is classified by type, degree and configuration for each ear. Hearing screening only provides a pass or non-pass result and indicates the need for additional diagnostic testing. Newborn hearing screening allows a subsequent referral to rule out potential hearing loss. The follow-up diagnostic testing will reveal the type, degree and configuration. See this link for the different types, degrees, and configurations of hearing loss:

<http://www.asha.org/public/hearing/disorders/types.htm>

How often is congenital hearing loss hereditary?

Studies have shown that at least 50% of congenital early-onset hearing loss has a genetic cause. Although most genetic hearing loss is isolated, about 1/3 of individuals affected with hearing loss also have complex syndromes with other associated medical problems such as cardiac, eye, renal, metabolic musculoskeletal, neurologic or other disorders. More than 400 syndromes have been identified which can cause hearing loss.

What factors put a newborn at high risk for hearing loss?

Newborns are at a higher risk for sensorineural hearing loss if they have one of the following factors: family history of hearing loss, congenital or central nervous system infections, ototoxic drug exposure, prematurity, congenital malformations of the head and neck, trauma, and other factors that have led to an admission to an intensive care nursery.

Why is genetic testing important for infants with hearing loss?

Making sure infants with hearing loss receive genetic evaluation and counseling can have many benefits for the baby and family. A genetic evaluation can:

- Help avoid unnecessary and often costly clinical tests to rule out conditions associated with syndromic deafness.
- Lead to improved decisions about treatment and management.
- Have health saving or life saving implications – particularly when syndromes with complex medical problems are identified.
- Be useful in developing an individualized treatment plan to ameliorate the complications of various syndromes.
- In some cases, help predict whether the hearing loss will be progressive. It also may be useful in determining what kind of damage has happened to the hearing system. Knowing how the inner ear is affected can determine what type of hearing device may help a patient.
- Dispel misinformation and ease parental guilt.
- Determine recurrence risks for future children to have hearing losses.
- Provide information about the possible cause, the chances of recurrence for family planning purposes and appropriate medical care for other family members.

Who should be referred for a genetic evaluation?

- All children with bilateral moderate, severe or profound sensorineural hearing loss.
- A child with any degree of hearing loss (including unilateral or mild) who also has either a family history of hearing loss or other co-existing medical problems or birth defects.
- Families who are interested in learning more about the possible genetic causes of their child's hearing loss.

What is genetic counseling and why is this important?

Genetic counseling is a health service that provides information and support for individuals and families who have, or who may be at risk for, a genetic condition. A genetic counselor helps the individual or family understand how inheritance works and what causes genetic conditions. The genetic counselor also can help the individual understand how this genetic information may impact them and their family emotionally. The genetic counselor will ask many questions about the family's medical history. Family history is used to determine the individual's risk for developing a genetic disorder, to determine the risk of having a child with a genetic disorder, and to help make a genetic diagnosis.

How important is family history?

Reviewing the family history can help establish whether the hearing loss is genetic; identify family members who might have a future child with hearing loss, as well as those who may be at risk for later onset hearing loss; and may be the key to establishing the correct diagnosis.

What do I need to understand about the deaf culture?

Many members of the deaf community suggest there is an inherent and unwarranted bias in the medical profession that views deafness as a disability or as needing medical intervention. As professionals, we need to be culturally sensitive concerning how we approach hearing screening and hearing loss. This includes cultural awareness of how the hearing screening results may be interpreted by a deaf family. The deaf community is a separate and valued culture in which members are bilingual (communicating in both ASL and English). The deaf community may not wish to have hearing aids, cochlear implants and/or intervention for their baby who has hearing loss.

While this perspective may be more common in parents who are deaf, the view is held by some hearing parents as well. Family perspective is very important. The decision about how to proceed with the evaluation and potential “treatment” of deafness is a personal family matter for the 90% of deaf children who are born to hearing parents as well as those born to deaf parents.

What is the future of genetic tests in diagnosing hearing loss?

Genetic testing for many genes associated with hearing loss is likely to become routine within the next few years. Knowing the precise cause of the hearing loss will help to determine the most effective management and therapeutic options. The availability of genetic testing is adding a new dimension to our ability to provide the best possible care for individuals with hearing loss.



Tool H1

*“Medical Home Care Management for Infants
with Confirmed Hearing Loss”*

Medical Home Care Management for Infants with Failed Newborn Screening or Confirmed Hearing Loss

Birth	<p>Hospital-based Inpatient Screening Results (OAE/AABR) (also home births)</p> <p>Left ear: <input type="checkbox"/> Missed <input type="checkbox"/> Incomplete <input type="checkbox"/> Refer ^{(a), (b)} <input type="checkbox"/> Pass</p> <p>Right ear: <input type="checkbox"/> Missed <input type="checkbox"/> Incomplete <input type="checkbox"/> Refer ^{(a), (b)} <input type="checkbox"/> Pass</p> <p style="text-align: right;">DATE: / / </p>
Before 1 Month	<p>Outpatient Screening Results (OAE/AABR)</p> <p>Left ear: <input type="checkbox"/> Missed <input type="checkbox"/> Incomplete <input type="checkbox"/> Refer ^{(a), (b)} <input type="checkbox"/> Pass</p> <p>Right ear: <input type="checkbox"/> Missed <input type="checkbox"/> Incomplete <input type="checkbox"/> Refer ^{(a), (b)} <input type="checkbox"/> Pass</p> <p style="text-align: right;">DATE: / / </p>
Before 3 Months	<p>Pediatric Diagnostic Audiology Evaluation</p> <p><input type="checkbox"/> Degree and configuration of hearing loss confirmed</p> <p><input type="checkbox"/> Documented child and family auditory history</p> <p><input type="checkbox"/> Received copy of confirmation of Hearing Loss form from Audiologist</p> <p><input type="checkbox"/> Refer to Early Intervention (IDEA, Part C)</p> <p><input type="checkbox"/> Medical and Otolgic Evaluations to recommend treatment and provide clearance for hearing aid fitting</p> <p><input type="checkbox"/> Pediatric Audiologic hearing aid fitting and monitoring</p> <p><input type="checkbox"/> Family Received Parent Brochure (Tools A, B, D, and F)</p> <p><input type="checkbox"/> Advise Family About Assistive Listening Devices & Hearing Aids (cochlear implants, etc and communication options)</p> <p style="text-align: right;">DATE: / / </p>
Before 6 Months	<p><input type="checkbox"/> Enrollment in Early Intervention (IDEA, Part C) (transition to Part B at 3 years of age)</p> <p>Medical Evaluations to determine etiology and identify related conditions</p> <p><input type="checkbox"/> Ophthalmologic (annually)</p> <p><input type="checkbox"/> Genetic</p> <p><input type="checkbox"/> Developmental pediatric, neurology, cardiology, and nephrology (as needed)</p> <p><input type="checkbox"/> Ongoing Pediatric Audiologic Services</p> <p style="text-align: right;">DATE: / / </p>

(a) In screening programs that do not provide Outpatient Screening, infants will be referred directly from Inpatient Screening to Pediatric Audiologic Evaluation. Likewise, infants at higher risk for hearing loss, or loss to follow-up, also may be referred directly to Pediatric Audiologic Evaluation.

(b) Part C of IDEA may provide diagnostic audiologic evaluation as part of Child Find activities.

(c) Infants who fail the screening in one or both ears should be referred to further screening or Pediatric Audiologic Evaluation.

(d) Includes infants who parents refused initial or follow-up hearing screening.

Patient Name: _____

Date of Birth: / /

Ongoing Care of All Infants ^(c)

Provide parents with information about hearing, speech, and language milestones

Provide parents with information about a genetic cause for hearing loss

Identify and aggressively treat middle ear disease

Vision screening and referral as needed

Ongoing developmental surveillance / referral

Referrals to otolaryngology and genetics, as needed

Risk indicators for late onset hearing loss

(refer for audiologic monitoring)

Service Provider Contact Information

Medical Home
Pediatric Audiologist
Early Intervention
Otolaryngologist/ENT
Geneticist
Ophthalmologist
Otologist
Other

OAE = Otoacoustic Emissions
AABR = Automated Auditory Brainstem Response
ABR = Auditory Brainstem Response
IDEA = Individuals with Disabilities Education Act



Medical Home Care Management from Birth to 36 Months for Infants with a Confirmed “Hearing Loss”

History and Examination

- **Coordinate audiologist visits.** Review the audiologist's report that confirms the diagnosis of hearing loss with the parents. Encourage Follow-up with an audiologist with pediatric expertise. A list of audiologists in your state can be found at www.region4genetics.org. Refer for regular audiologic evaluation based on audiologist's/otolaryngologist's recommendations. Sometimes hearing loss is progressive; unilateral loss can become bilateral; mild can become severe.
- **Review child and family history.**
- **Evaluate for genetic or syndromic etiologies.** Assess for other physical findings. About half of newborns with hearing loss have a genetic cause, some associated with syndromes. The most common organs involved are eyes, heart, kidneys, thyroid, and bones. If you suspect a syndrome, consider referral to a geneticist and/or appropriate sub-specialist.
- **Ensure early intervention.** Refer to your local early intervention. Research shows typical or near-typical language development in children who receive intervention before 6 months of age. (See the Region 4 EHDI Resources page at: http://region4genetics.org/region4_products/genetics_hearingloss.aspx)
- **Monitor middle ear status.** This is especially critical in children with confirmed hearing loss as middle ear effusion may further compromise hearing.
- **Maintain scheduled well-child visits and immunizations.**
- **Precautions for children with cochlear implants.** Children with cochlear implants may be at higher risk for meningitis. Make sure they are up to date on their Haemophilus influenzae type b and pneumococcal immunizations. Refer to www.CDC.gov/ncbddd/ehdi/cochlear/ for recommendations.

Working with Families

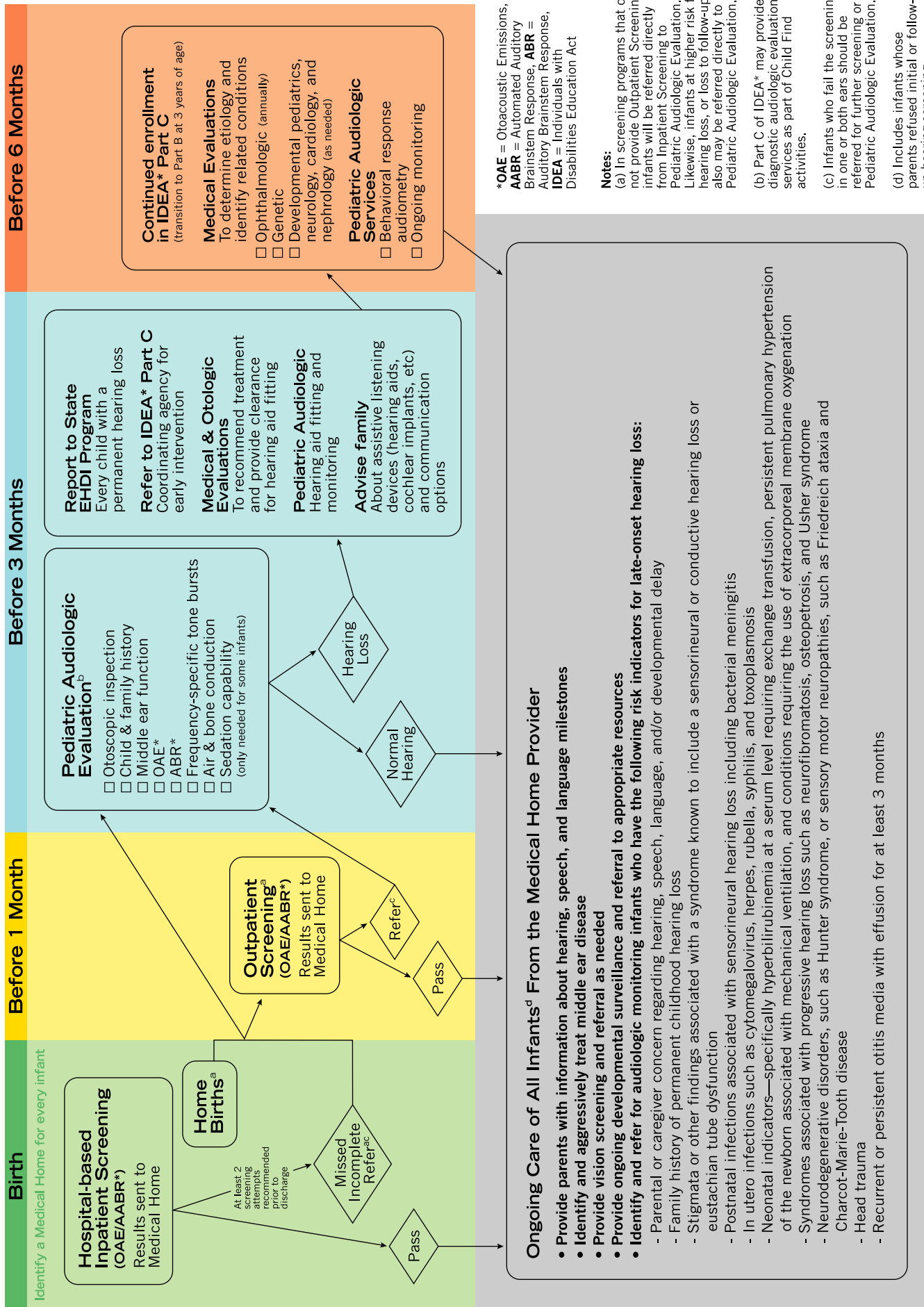
- **Family support.** Be aware that many families will experience the same grief that accompanies other significant diagnoses of the newborn. Families need the emotional support of other families. The family benefits from contact with people who are deaf or hard of hearing. Provide the family with names of state or local organizations that provide information and support. Family support resources and information on intervention options are available at www.region4genetics.org.
- **Early intervention.** Discuss the importance of early intervention. Children who receive qualified and ongoing intervention before 6 months of age may, in many cases, maintain language development commensurate with their cognitive abilities through the age of five years. Delayed intervention can result in significant delays in communication and language skills, including reading. There is no advantage in delaying intervention.
 - There are many intervention options and strategies that may be appropriate for children who are deaf or hard of hearing or their families.
 - Communication options for families include American Sign Language, Auditory/Oral approaches, as well as a blending of varied communication methods based on the child's needs and family's goals. All forms of communication may be used alone or with an amplification device.
 - Amplification devices include hearing aids, which may be fitted to infants as young as four weeks, and cochlear implants, which may be implanted at 12 months of age.
- **Parent bonding.** Parents may need support in bonding with their infant/young child; encourage parents to hug, hold, smile and even sing and talk to their baby—all attention given with love is beneficial.
- **Language and auditory skills assessment.** Assure that the child's language, communication, and auditory skills are assessed by people with the qualification and experience to do so.
- **Amplification.** If the child is using amplification devices, make sure they are worn continuously while awake. Ensure the parents know how to use the devices.



Tool H2

*“Universal Newborn Hearing Screening,
Diagnosis, and Intervention Guidelines for
Pediatric Medical Home Providers”*

Universal Newborn Hearing Screening, Diagnosis, and Intervention Guidelines for Pediatric Medical Home Providers





Tool H3

*“Hearing Assessment Algorithm within
an Office Visit”*

FIGURE: 1 Hearing-assessment algorithm within an office visit

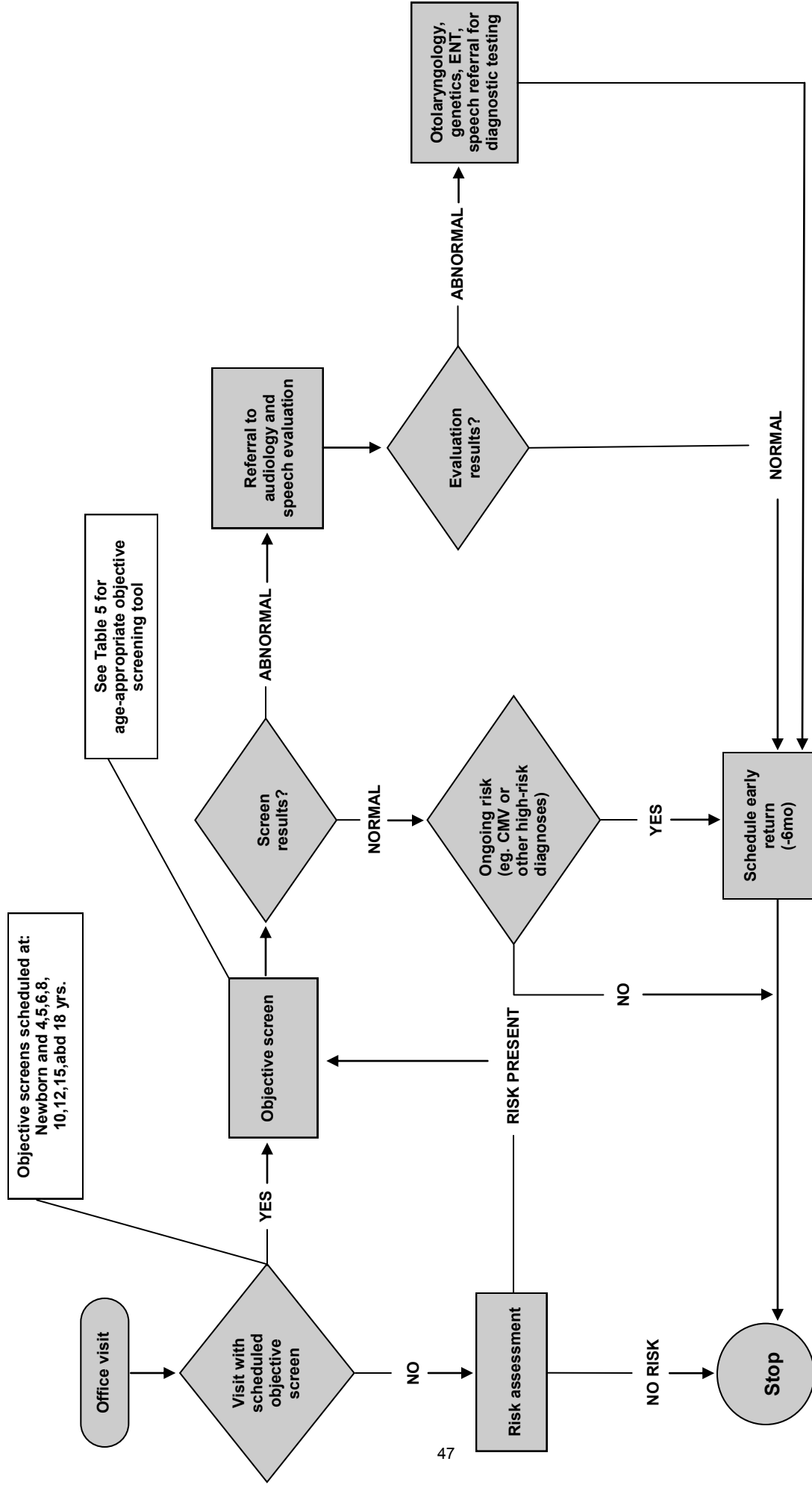


Table 5 Audiologic Tests for Infants and Children

Developmental Age of Child	Auditory Test/ Average Time	Type of Measurement	Test Procedures	Advantages	Limitations
All Ages	Evoked OAEs/ 10 min. test	Physiologic test specifically measuring cochlear (outer hair cell) response to presentation of a stimulus; stimuli may be clicks (transient evoked OAEs) or tone pairs (distortion product OAEs)	Small probe containing a sensitive microphone is placed in the ear canal for stimulus delivery and response detection	Ear-specific results; not dependent on whether patient is asleep or awake; quick test time; screening test	Infant or child must be relatively inactive during the test; not a comprehensive test of hearing, because it does not assess cortical processing of sound; OAEs are very sensitive to middle-ear effusions and cerumen or vernix in the ear canal
Birth to 9 mo	Automated ARB/ 15 min. test	Electrophysiologic measurement of activity in auditory nerve and brainstem pathways	Placement of electrodes on child's head detects neurologic response to auditory stimuli presented through earphones or ear inserts 1 ear at a time	Ear-specific results; responses not dependent on patient cooperation; screening test	Infant or child must remain quiet during the test (sedation is often required); not a comprehensive test of hearing, because it does not assess cortical processing of sound
9 mo to 2.5 yrs	VRA/15 to 30 min test	Behavioral tests measuring responses of the child to speech and frequency-specific stimuli presented through speakers or insert earphones	Technique conditions the child to associate speech or frequency-specific stimuli with a reinforcer, such as a lighted toy or video clips; VRA requires a calibrated, sound-treated room	Assesses auditory perception of child; diagnostic test	When performed with speakers, only assess hearing of the better ear; not ear specific; if VRA is performed with insert, earphones can rule out a unilateral hearing loss
2.5 to 4 yrs	Play audiometry/ 15 to 30 min	Behavioral test of auditory thresholds in response to speech and frequency – specific stimuli presented through earphones and/or bone vibrator	Child is conditioned to respond when stimulus tone is heard, such as to put a peg in a pegboard or drop a block in a box	Ear-specific results; assesses auditory perception of child; diagnostic test	Attention span of child may limit the amount of information obtained
4 yrs to adolescence	Conventional audiometry/15 to 30 min test	Behavioral test of auditory thresholds in response to speech and frequency – specific stimuli presented through earphones and/or bone vibrator	Patient is instructed to raise his or her hand when stimulus is heard	Ear-specific results; assesses auditory perception of child; screening or diagnostic test	Depends on the level of understanding and cooperation of the child
All ages	Diagnostic ABR	Electrophysiologic measurement of activity in auditory nerve and brainstem pathways	Placement of electrodes on child's head detects auditory stimuli presented through insert earphones 1 ear at a time	Ear-specific results; multiple frequencies are tested, creating a map of hearing loss similar to an audiogram; responses not dependent on patient cooperation; diagnostic test	Infant or child must remain quiet during the test (sedation is often required); not a true test of hearing, because it does not assess cortical processing of sound
All ages	Tympanometry	Relative change in middle ear compliance as air pressure is varied in the external auditory canal	Small probe placed in the ear canal and pressure varied in the ear canal	Tests for possible middle ear pathology and pressure equalization tube function	Not a test of hearing; depends on ear canal seal; high-frequency tone probe needed for infants younger than 6 mo.

Adapted with permission from Bachmann KR, Arvedson JC, Pediatr Rev. 1998; 19(5):155-165.



Tool H4

*“ACMG ACT Sheet -
Congenital Hearing Loss”*



Clinical Report—Hearing Assessment in Infants and Children: Recommendations Beyond Neonatal Screening

abstract

Congenital or acquired hearing loss in infants and children has been linked with lifelong deficits in speech and language acquisition, poor academic performance, personal-social maladjustments, and emotional difficulties. Identification of hearing loss through neonatal hearing screening, regular surveillance of developmental milestones, auditory skills, parental concerns, and middle-ear status and objective hearing screening of all infants and children at critical developmental stages can prevent or reduce many of these adverse consequences. This report promotes a proactive, consistent, and explicit process for the early identification of children with hearing loss in the medical home. An algorithm of the recommended approach has been developed to assist in the detection and documentation of, and intervention for, hearing loss. *Pediatrics* 2009;124:1252–1263

KEY POINTS

1. Every child with 1 or more risk factors on the hearing risk assessment should have ongoing developmentally appropriate hearing screening and at least 1 diagnostic audiology assessment by 24 to 30 months of age.
2. Periodic objective hearing screening of all children should be performed according to the recommendations for preventive periodic health care.¹
3. Any parental concern about hearing loss should be taken seriously and requires objective hearing screening of the patient.
4. All providers of pediatric health care should be proficient with pneumatic otoscopy and tympanometry. However, it is important to remember that these methods do not assess hearing.
5. Developmental abnormalities, level of functioning, and behavioral problems (ie, autism/developmental delay) may preclude accurate results on routine audiometric screening and testing. In this situation, referral to an otorhinolaryngologist and a pediatric audiologist who has the necessary equipment and expertise to test infants and young children should be made.
6. The results of abnormal screening should be explained carefully to parents, and the child's medical record should be flagged to facilitate tracking and follow-up.

Allen D. “Buz” Harlor, Jr, MD, Charles Bower, MD, THE COMMITTEE ON PRACTICE AND AMBULATORY MEDICINE, THE SECTION ON OTOLARYNGOLOGY—HEAD AND NECK SURGERY

KEY WORD

hearing screening, hearing loss, audiology

ABBREVIATIONS

AAP—American Academy of Pediatrics

OAE—otoacoustic emission

ABR—auditory brainstem response

VRA—visual reinforced audiometry

The guidance in this report does not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account, individual circumstances may be appropriate.

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www.pediatrics.org/cgi/doi/10.1542/peds.2009-1997

doi:10.1542/peds.2009-1997

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PEDIATRICS (ISSN Numbers: Print, 0031-4005; Online, 1098-4275).

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7. Any abnormal objective screening result requires audiology referral and definitive testing.
8. A failed infant hearing screening or a failed screening in an older child should always be confirmed by further testing.
9. Abnormal hearing test results require intervention and clinically appropriate referral, including otolaryngology, audiology, speech-language pathology, genetics, and early intervention.

INTRODUCTION

Failure to detect congenital or acquired hearing loss in children may result in lifelong deficits in speech and language acquisition, poor academic performance, personal-social maladjustments, and emotional difficulties. Early identification of hearing loss and appropriate intervention within the first 6 months of life have been demonstrated to ameliorate many of these adverse consequences and facilitate language acquisition.² Supportive evidence is outlined in the Joint Committee on Infant Hearing's "Year 2007 Position Statement: Principles and Guidelines for Early Hearing Detection and Intervention Programs," which was endorsed by the American Academy of Pediatrics (AAP).³ This evidence also is part of the rationale for the AAP statement "Newborn and Infant Hearing Loss: Detection and Intervention,"⁴ which endorses universal hearing screening and reviews the primary objectives, important components, and recommended screening methods and parameters that characterize an effective universal hearing screening program. Furthermore, the AAP statement "Recommendations for Preventive Pediatric Health Care"¹ promotes objective newborn hearing screening as well as periodic hearing screening for every child through adolescence (Table 1).

TABLE 1 Recommendations for Preventive Pediatric Health Care¹

Stage	Age	Sensory Screening: 6. Hearing
Infancy	Prenatal	
	Newborn	a
	3–5 d ¹	b
	By 1 mo	b
	2 mo	b
	4 mo	b
	6 mo	b
Early childhood	9 mo	b
	12 mo	b
	15 mo	b
	18 mo	b
	24 mo	b
	30 mo	b
	3 y	b
Middle childhood	4 y	a
	5 y	a
	6 y	a
	7 y	b
	8 y	a
	9 y	b
	10 y	a
Adolescence	11 y	b
	12 y	b
	13 y	b
	14 y	b
	15 y	b
	16 y	b
	17 y	b
	18 y	b
	19 y	b
	20 y	b
	21 y	b

^a To be performed.

^b Risk assessment, with appropriate action to follow if positive.

All providers of pediatric health care need to recognize children who are at risk of or who suffer from congenital or acquired hearing loss, be prepared to screen their hearing, and assist the family and arrange for proper referral and treatment by identifying available hearing resources within their communities. In addition, the pediatric health care professional can play an important role in communication with the child's schoolteacher and/or nurse and special education professionals to facilitate proper accommodation and education once a hearing deficit has been confirmed.

This clinical report replaces the previous 2003 clinical report and seeks to promote a proactive, consistent, and

explicit process for the early identification of children with hearing loss in the medical home. To assist in the detection and documentation of and intervention for hearing loss, an algorithm of the recommended approach with key points has been developed (Fig 1), as have several tables.

RISK INDICATORS FOR HEARING LOSS

Some degree of hearing loss (Table 2) is present in 1 to 6 per 1000 newborn infants.⁵ Most children with congenital hearing loss are potentially identifiable by newborn and infant hearing screening. However, some congenital hearing loss may not become evident until later in childhood. Hearing loss also can be acquired during infancy or childhood for various reasons. Infectious diseases, especially meningitis, are a leading cause of acquired hearing loss. Trauma to the nervous system, damaging noise levels, and ototoxic drugs can all place a child at risk of developing acquired hearing loss.^{6,7} Otitis media is a common cause of usually reversible hearing loss. Certain physical findings, historical events, and developmental conditions may indicate a potential hearing problem. These conditions include, but are not limited to, anomalies of the ear and other craniofacial structures, significant perinatal events, and global developmental or speech-language delays. All older infants and children should be screened for risk factors involving hearing problems. A summary of high-risk indicators for hearing loss and developmental milestones are included in Tables 3 and 4, respectively. All infants with a risk indicator for hearing loss, regardless of surveillance findings, should be referred for an audiologic assessment at least once by 24 to 30 months of age, even if the child passed the newborn hearing screening. Children with risk indicators that are highly associated with delayed-onset hearing loss, such as hav-

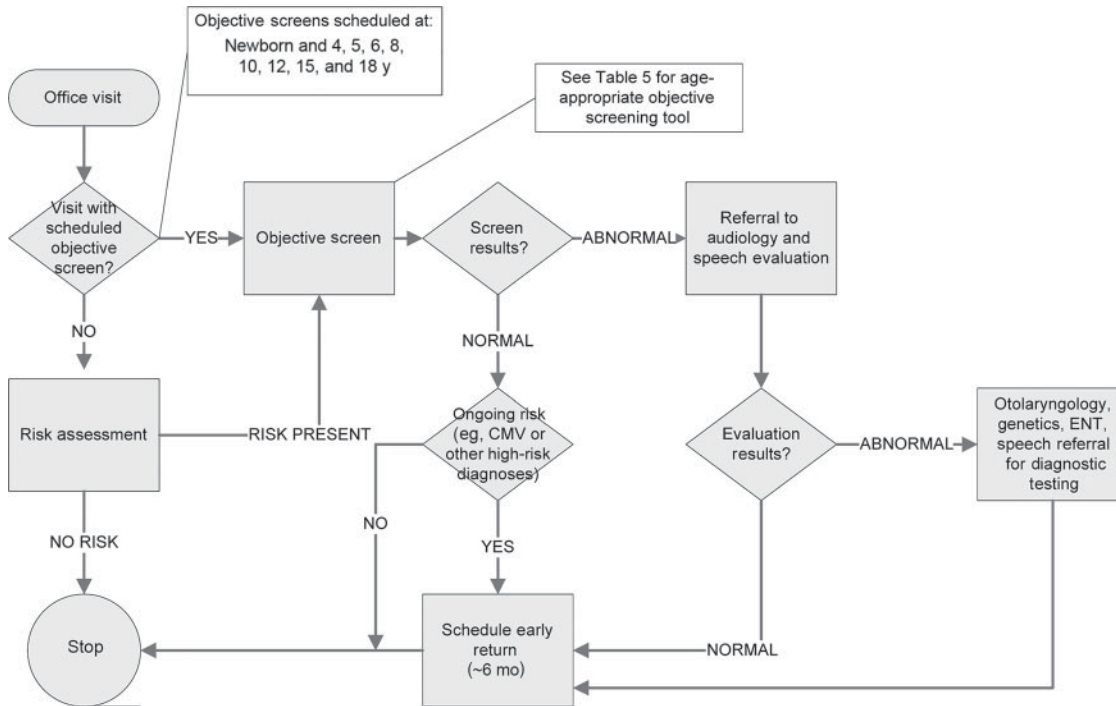


FIGURE 1 Hearing-assessment algorithm within an office visit. CMV indicates cytomegalovirus; ENT, ear, nose, and throat.

ing received extracorporeal membrane oxygenation or having cytomegalovirus infection, should have more frequent audiological assessments. Key point 1: Every child with 1 or more risk factors on the hearing risk assessment should have ongoing developmentally appropriate hearing screening and at least 1 diagnostic audiology assessment by 24 to 30 months of age (Table 1).

Although questionnaires and checklists are useful for identifying a child at risk of hearing loss, studies have

shown that only 50% of children with hearing loss are identified by the comprehensive use of such questionnaires.^{8,9} Key point 2: Periodic objective hearing screening of all children should be performed according to the recommendations for preventive periodic health care¹ (Table 1).

If a parent or caregiver is concerned that a child might have hearing loss, the pediatrician needs to assume that such is true until the child's hearing has been evaluated objectively. Paren-

tal concern is of greater predictive value than the informal behavioral examination performed in the physician's office.¹⁰ Parents often report suspicion of hearing loss, inattention, or erratic response to sound before hearing loss is confirmed.¹¹ One study showed that parents were as much as 12 months ahead of physicians in identifying their child's hearing loss.³ Key point 3: Any parental concern about hearing loss should be taken seriously and requires objective hearing screening of the patient.

PHYSICAL EXAMINATION

Thorough physical examination is an essential part of evaluating a child for hearing loss. Findings on head and neck examination associated with potential hearing loss include heterochromia of the irises, malformation of the auricle or ear canal, dimpling or skin tags around the auricle, cleft lip or palate, asymmetry or hypoplasia of the facial structures, and microcephaly.¹² Hypertelorism and abnormal pig-

TABLE 2 Definitions of Hearing Loss

Hearing Loss	Definition
Mild	On average, the most quiet sounds that people can hear with their better ear are between 20 and 40 dB. People who suffer from mild hearing loss have some difficulties keeping up with conversations, especially in noisy surroundings.
Moderate	On average, the most quiet sounds heard by people with their better ear are between 40 and 70 dB. People who suffer from moderate hearing loss have difficulty keeping up with conversations when not using a hearing aid.
Severe	On average, the most quiet sounds heard by people with their better ear are between 70 and 95 dB. People who suffer from severe hearing loss will benefit from powerful hearing aids, but often they rely heavily on lip reading, even when they are using hearing aids. Some also use sign language.

Adapted from: European Group on Genetics of Hearing Impairment. Martini A, ed. European Commission Directorate, Biomedical and Health Research Programme (HEAR) Infoletter 2, November 1996;8.

TABLE 3 American Academy of Pediatrics Joint Committee on Infant Hearing Year 2007 Position Statement³: Risk Indicators Associated With Permanent Congenital, Delayed-Onset, and/or Progressive Hearing Loss in Childhood

1	Caregiver concern ^a regarding hearing, speech, language, or developmental delay.
2	Family history ^a of permanent childhood hearing loss.
3	Neonatal intensive care of more than 5 days or any of the following regardless of length of stay: ECMO ^a , assisted ventilation, exposure to ototoxic medications (gentamicin and tobramycin) or loop diuretics (furosemide/Lasix), and hyperbilirubinemia ¹⁹ that requires exchange transfusion.
4	In utero infections such as CMV ^a , herpes, rubella, syphilis, and toxoplasmosis.
5	Craniofacial anomalies, including those that involve the pinna, ear canal, ear tags, ear pits, and temporal bone anomalies.
6	Physical findings, such as white forelock, that are associated with a syndrome known to include a sensorineural or permanent conductive hearing loss.
7	Syndromes associated with hearing loss or progressive or late-onset hearing loss ^a , such as neurofibromatosis, osteopetrosis, and Usher syndrome; other frequently identified syndromes include Waardenburg, Alport, Pendred, and Jervell and Lange-Nielson.
8	Neurodegenerative disorders ^a , such as Hunter syndrome, or sensory motor neuropathies, such as Friedreich ataxia and Charcot-Marie-Tooth syndrome.
9	Culture-positive postnatal infections associated with sensorineural hearing loss ^a , including confirmed bacterial and viral (especially herpes viruses and varicella) meningitis.
10	Head trauma, especially basal skull/temporal bone fracture ^a that requires hospitalization.
11	Chemotherapy ^a .
12	Recurrent or persistent otitis media for at least 3 months.

Risk indicators that are marked with ^a are of greater concern for delayed onset hearing loss. ECMO indicates extracorporeal membrane oxygenation; CMV, cytomegalovirus.

Reproduced with permission from: American Academy of Pediatrics, Joint Committee on Infant Hearing. *Pediatrics*. 2007; 120(4):898–921.

mentation of the skin, hair, or eyes also may be associated with hearing loss, as in Waardenburg syndrome. The presence of renal abnormalities (Alport syndrome), cardiac anomalies (prolonged QT interval in Jervell and Lange-Nielsen syndrome), and other syndromes should also prompt evaluation of hearing. Abnormalities of the eardrum should alert the physician to the possibility of hearing loss. Cerumen impactions can obscure the tympanic membrane, preventing an accurate examination, and may cause hearing loss. Dense cerumen impactions should be removed before diagnostic testing. A leading cause of acquired hearing loss is otitis media with effusion. Temporary hearing loss has been demonstrated during episodes of acute otitis media. The child with repeated or chronic otitis media with effusion is at high risk of acquired hearing loss and should undergo comprehensive hearing evaluation.^{13,14} Key point 4: All providers of pediatric health care should be pro-

ficient with pneumatic otoscopy and tympanometry. However, it is important to remember that these methods do not assess hearing.

TOOLS FOR OBJECTIVE HEARING SCREENING

In addition to universal newborn hearing screening and regular surveillance of developmental milestones, auditory skills, parental concerns, and middle-ear status, objective screenings for hearing loss should be performed periodically on all infants and children in accordance with the schedule outlined in the AAP statement “Recommendations for Preventive Pediatric Health Care”¹¹ (Table 1). The technology used for hearing screening should be age appropriate. The child also should be comfortable with the testing situation; young children may need preparation. A variety of objective tools have been developed for screening tests. The choice of tool to use in screening depends on the child’s age, degree of cooperation, and available resources.

Screenings should be conducted in a quiet area where visual and auditory distractions are minimal. For children for whom screening is not possible because of developmental level, referral to a pediatric audiologist should be initiated for appropriate physiologic and/or behavioral audiological assessment. Various tests performed by audiologists are outlined in Table 5.

Tympanograms

Conductive hearing loss may be the most common cause of infant hearing screening failures.¹⁵ Objective middle-ear assessment can best be performed by tympanometry. Tympanometry measures relative changes in tympanic membrane movement as air pressure is varied in the external auditory canal. Tympanograms (Fig 2) can most simply be classified as types A, B, and C depending on the curve shape relative to 0 as the pressure is changed (www.audiologyonline.com/askexpert/display_question.asp?question_id=451). The presence of a type A, high-peaked tympanogram significantly decreases the probability that middle-ear effusion is the cause of hearing loss. A type B, flat tympanogram has the highest probability of the presence of middle-ear effusion or tympanic membrane perforation, which are both likely to cause some degree of hearing loss. A type C tympanogram, with a peak shifted toward negative pressure, has a low probability of middle-ear fluid and associated hearing loss. Type B and C tympanograms require clinical correlation and possibly further evaluation and treatment. Traditionally, tympanograms have been obtained by using low-frequency probe tones. These tones have been historically inaccurate for infants younger than 6 months. The use of a high-frequency probe tone (1000 Hz) was recently shown to be a better measure of middle-ear status in infants and young

TABLE 4 Developmental Milestones in the First 2 Years of Life

Milestone	Average Age of Attainment, mo	Developmental Implications
Gross motor		
Head steady in sitting	2.0	Allows more visual interaction
Pull to sit, no head lag	3.0	Muscle tone
Hands together in midline	3.0	Self-discovery
Asymmetric tonic neck reflex gone	4.0	Child can inspect hands in midline
Sits without support	6.0	Increasing exploration
Rolls back to stomach	6.5	Truncal flexion, risk of falls
Walks alone	12.0	Exploration, control of proximity to parents
Runs	16.0	Supervision more difficult
Fine motor		
Grasps rattle	3.5	Object use
Reaches for objects	4.0	Visuomotor coordination
Palmar grasp gone	4.0	Voluntary release
Transfers object hand to hand	5.5	Comparison of objects
Thumb-finger grasp	8.0	Able to explore small objects
Turns pages of book	12.0	Increasing autonomy during book time
Scribbles	13.0	Visuomotor coordination
Builds tower of 2 cubes	15.0	Uses objects in combination
Builds tower of 6 cubes	22.0	Requires visual, gross, and fine motor coordination
Communication and language		
Smiles in response to face, voice	1.5	Child more active social participant
Monosyllabic babble	6.0	Experimentation with sound, tactile sense
Inhibits to "no"	7.0	Response to tone (nonverbal)
Follows 1-step command with gesture	7.0	Nonverbal communication
Follows 1-step command without gesture (eg, "Give it to me")	10.0	Verbal receptive language
Speaks first real word	12.0	Beginning of labeling
Speaks 4–6 words	15.0	Acquisition of object and personal names
Speaks 10–15 words	18.0	Acquisition of object and personal names
Speaks 2-word sentences (eg, "Mommy shoe")	19.0	Beginning grammaticization, corresponds with vocabulary of ≥ 50 words
Cognitive		
Stares momentarily at spot where object disappeared (eg, yarn ball dropped)	2.0	Lack of object permanence (out of sight, out of mind)
Stares at own hand	4.0	Self-discovery, cause and effect
Bangs 2 cubes	8.0	Active comparison of objects
Uncovers toy (after seeing it hidden)	8.0	Object permanence
Egocentric pretend play (eg, pretends to drink from cup)	12.0	Beginning symbolic thought
Uses stick to reach toy	17.0	Able to link actions to solve problems
Pretend play with doll (gives doll bottle)	17.0	Symbolic thought

Modified from: Behrman RE, Jenson HB, Kliegman R, eds. *Nelson Textbook of Pediatrics*. 16th ed. Philadelphia, PA: Saunders; 2003.

children. These tympanograms are generally not classified as A, B, or C but rather as peak or no peak.

Evoked Otoacoustic Emissions

Evoked otoacoustic emissions (OAEs) are acoustic signals generated from within the cochlea that travel in a reverse direction through the middle-ear space and tympanic membrane out to

the ear canal. These signals are generated in response to an auditory stimulus, either clicks or tone bursts. The signals may be detected with a very sensitive microphone/probe system placed in the external ear canal. The OAE test allows for individual ear assessment, can be performed quickly at any age, and does not depend on whether the child is asleep or awake.

Mild degrees of motion artifact do not interfere with test results; however, screening results are frequently influenced by the presence of middle-ear pathologic abnormalities. The OAE test is an effective screening measure for middle-ear abnormalities and for moderate or more severe degrees of hearing loss, because normal OAE responses are not obtained if hearing thresholds are approximately 30- to 40-dB hearing levels or higher. The automated OAE screener provides a pass-fail report; no test interpretation by an audiologist is required. The OAE test does not further quantify hearing loss or hearing threshold level. The OAE test also does not assess the integrity of the neural transmission of sound from the eighth nerve to the brainstem and, therefore, will miss auditory neuropathy and other neuronal abnormalities. Infants with such abnormalities will have normal OAE test results but abnormal auditory brainstem response (ABR) test results. A "failed" OAE test only implies that a hearing loss of more than 30 to 40 dB may exist or that the middle-ear status is abnormal.

Automated ABR

One objective physiologic means of screening hearing is the automated ABR. This instrument measures cochlear response in the 1- to 4-kHz range with a broadband click stimulus in each ear. Many ABR screening instruments incorporate built-in artifact rejection for myogenic, electrical, and environmental noise interference, which ensures that data collection is halted if testing conditions are unfavorable. The automated screener provides a pass-fail report; no test interpretation by an audiologist is required. A "fail" report on an automated ABR implies a hearing level of worse than 40 dB. Automated ABR can test each ear individually and can be performed on children of any age. Motion artifacts interfere with test results. For this reason, the test

TABLE 5 Audiologic Tests for Infants and Children

Developmental Age of Child	Auditory Test/Average Time	Type of Measurement	Test Procedures	Advantages	Limitations
All ages	Evoked OAEs/10-min test	Physiologic test specifically measuring cochlear (outer hair cell) response to presentation of a stimulus; stimuli may be clicks (transient evoked OAEs) or tone pairs (distortion product OAEs)	Small probe containing a sensitive microphone is placed in the ear canal for stimulus delivery and response detection	Ear-specific results; not dependent on whether patient is asleep or awake; quick test time; screening test	Infant or child must be relatively inactive during the test; not a comprehensive test of hearing, because it does not assess cortical processing of sound; OAEs are very sensitive to middle-ear effusions and cerumen or vernix in the ear canal
Birth to 9 mo	Automated ABR/15-min test	Electrophysiologic measurement of activity in auditory nerve and brainstem pathways	Placement of electrodes on child's head detects neurologic response to auditory stimuli presented through earphones or ear inserts 1 ear at a time	Ear-specific results; responses not dependent on patient cooperation; screening test	Infant or child must remain quiet during the test (sedation is often required); not a comprehensive test of hearing, because it does not assess cortical processing of sound
9 mo to 2.5 y	VRA/15- to 30-min test	Behavioral tests measuring responses of the child to speech and frequency-specific stimuli presented through speakers or insert earphones	Technique conditions the child to associate speech or frequency-specific stimuli with a reinforcer, such as a lighted toy or video clips; VRA requires a calibrated, sound-treated room	Assesses auditory perception of child; diagnostic test.	When performed with speakers, only assesses hearing of the better ear; not ear specific; if VRA is performed with insert, earphones can rule out a unilateral hearing loss
2.5 to 4 y	Play audiometry/15–30 min	Behavioral test of auditory thresholds in response to speech and frequency-specific stimuli presented through earphones and/or bone vibrator	Child is conditioned to respond when stimulus tone is heard, such as to put a peg in a pegboard or drop a block in a box	Ear-specific results; assesses auditory perception of child; screening or diagnostic test.	Attention span of child may limit the amount of information obtained
4 y to adolescence	Conventional audiometry/15- to 30-min test	Behavioral test measuring auditory thresholds in response to speech and frequency-specific stimuli presented through earphones and/or bone vibrator	Patient is instructed to raise his or her hand when stimulus is heard	Ear-specific results; assesses auditory perception of patient; screening or diagnostic test	Depends on the level of understanding and cooperation of the child
All ages	Diagnostic ABR	Electrophysiologic measurement of activity in auditory nerve and brainstem pathways	Placement of electrodes on child's head detects auditory stimuli presented through insert earphones 1 ear at a time	Ear-specific results; multiple frequencies are tested, creating a map of hearing loss similar to an audiogram; responses not dependent on patient cooperation; diagnostic test	Infant or child must remain quiet during the test (sedation is often required); not a true test of hearing, because it does not assess cortical processing of sound
All ages	Tympanometry	Relative change in middle-ear compliance as air pressure is varied in the external auditory canal	Small probe placed in the ear canal and pressure varied in the ear canal	Tests for possible middle-ear pathology and pressure-equalization tube function	Not a test of hearing; depends on ear canal seal; high-frequency tone probe needed for infants younger than 6 mo

Adapted with permission from: Bachmann KR, Arvedson JC. *Pediatr Rev*. 1998;19(5):155–165.

is performed best in infants and young children while they are sleeping. If the test cannot be performed because of

motion artifact, sedation may be necessary. The ABR is currently used in many newborn screening programs.

ABR and OAEs are tests of auditory pathway structural integrity but are not true tests of hearing. Even if ABR or OAE test

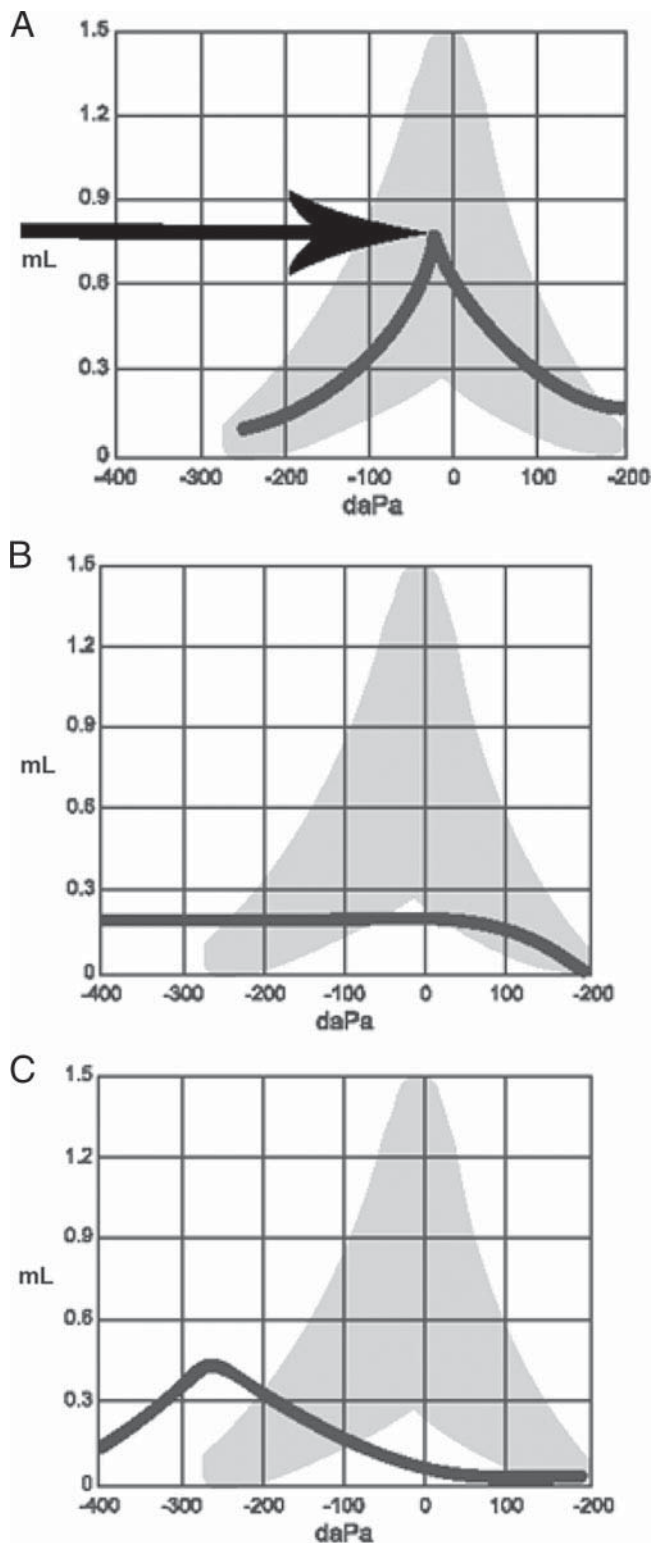


FIGURE 2 Tympanograms. Type A: normal. Type B: abnormal, needs medical attention. Type C, borderline normal; monitor; may need medical attention.

results are normal, hearing cannot be definitively considered normal until a child is mature enough for a reliable be-

havioral audiogram to be obtained. Behavioral pure-tone audiometry remains the standard for hearing evaluation.

Hearing thresholds at specific frequencies can be determined, and the degree of hearing loss can be assigned. If there are distractions or the room is not sound treated, pure-tone audiometry in the office should be considered solely a screening test.

Play Audiometry

Children 2 to 4 years of age are screened or tested more appropriately by play audiometry. These children are conditioned to respond to an auditory stimulus through play activities, such as dropping a block when a sound is heard through earphones. Air-conduction hearing threshold levels of greater than 20 dB at any of these frequencies indicate possible hearing loss, and referral to a pediatric audiologist should be made.

Conventional Screening Audiometry

For children aged 4 years and older, conventional screening audiometry can be used. The child is asked to raise his or her hand when a sound is heard. The test should be performed in a quiet environment using earphones, because ambient noise can affect test performance significantly, especially at lower frequencies (ie, 500 and 1000 Hz). Each ear should be tested at 500, 1000, 2000, and 4000 Hz. Air-conduction hearing threshold levels of greater than 20 dB at any of these frequencies indicate possible hearing loss, and referral to a pediatric audiologist should be made.

If the child does not pass the screening, earphones should be removed and instructions carefully repeated to the child to ensure proper understanding and attention to the test and then re-screened with the earphones repositioned. A child whose repeat test shows hearing thresholds of greater than 20 dB at any of these frequencies, especially if there is no pathologic abnormality of the middle ear on physical

examination, should be referred for formal hearing testing. Key point 5: Developmental abnormalities, level of functioning, and behavioral problems (ie, autism/developmental delay) may preclude accurate results on routine audiometric screening and testing. In this situation, referral to an otorhinolaryngologist and a pediatric audiologist who has the necessary equipment and expertise to test infants and young children should be made (Table 5). Key point 6: The results of abnormal screening should be explained carefully to parents, and the child's medical record should be flagged to facilitate tracking and follow-up.

It is important to remember that a "fail" report on any 1 of a combination of tests warrants additional testing. It is also important to remember that failure of speech, language, and hearing screening assessments warrants additional testing (Tables 6–9).

Comprehensive Audiological Evaluation Using Physiologic and/or Behavioral Testing

The ABR test may be used as a diagnostic tool by audiologists for more definitive diagnosis of hearing loss. Usually performed in children in natural sleep up to approximately 3 to 6 months of age and then under sedation for older infants, diagnostic ABR can provide not only a general level of hearing but also frequency-specific hearing data. Diagnostic ABR is performed with different frequency tone bursts and across varying sound levels to effectively estimate an audiogram. Diagnostic ABR can also be performed with bone conduction to separate conductive from sensorineural hearing loss. Diagnostic ABR is often the definitive test used by audiologists in children and infants who are unable to cooperate with other methods of hearing testing. Audiologic evaluation using ABR or auditory steady-state response provides

TABLE 6 Ten Ways to Recognize Hearing Loss: Adolescents (11- to 21-Year Visits)

1.	Do you have a problem hearing over the telephone?
2.	Do you have trouble following the conversation when two or more people are talking at the same time?
3.	Do people complain that you turn the TV volume up too high?
4.	Do you have to strain to understand conversation?
5.	Do you have trouble hearing in a noisy background?
6.	Do you find yourself asking people to repeat themselves?
7.	Do many people you talk to seem to mumble (or not speak clearly)?
8.	Do you misunderstand what others are saying and respond inappropriately?
9.	Do you have trouble understanding the speech of women and children?
10.	Do people get annoyed because you misunderstand what they say?

Adapted from: National Institute on Deafness and Other Communication Disorders. *Ten Ways to Recognize Hearing Loss*. Bethesda, MD: National Institute of Health; 2006. NIH publication 01-4913. Available at: www.nidcd.nih.gov/health/hearing/10ways.asp.

frequency-specific hearing thresholds by air and bone conduction in each ear separately. ABR is the gold standard for determination of hearing thresholds in infants younger than 6 months and in children who cannot be tested behaviorally.

Children as young as 6 to 24 months can be tested by means of visual reinforced audiometry (VRA). This technique conditions the child to associate speech or frequency-specific sound with a reinforcement stimulus such as a lighted toy or animated toy or video clips. VRA is performed by an audiologist with experience testing young children. This testing is not readily applied in screening programs, because infants younger than 6 months' developmental age cannot perform the task, and sound-treated rooms are needed. The results of VRA can approximate those of conventional audiometry.

Children with unilateral or mild hearing loss also should be evaluated further. Studies have shown such children to be similarly at risk of adverse communication skills as well as difficulties with social, emotional, and educational development.¹⁶

FOLLOW-UP AND DIAGNOSTIC TESTING

Key point 7: Any abnormal objective screening result requires audiology referral and definitive testing. Screen-

ing will only result in benefit for the patient if abnormal test results are confirmed and appropriate intervention is provided. Most studies that have evaluated the success rate of infant hearing screening programs have described a fairly high rate of failure to confirm a failed screen with definitive testing. A similar problem could also occur in screening older infants and children. Improving the physician's involvement not only in screening but also in arranging and confirming appropriate follow-up testing and intervention is necessary to achieve optimal speech, language, and hearing.

Key point 8: A failed infant hearing screening or a failed screening in an older child should always be confirmed by further testing. Audiologists may repeat the audiometric test as described above in a sound booth and using a variety of other tests. ABR can also be used for definitive testing of the auditory system. A diagnostic ABR is usually performed under sedation or general anesthesia in children aged approximately 3 to 6 months and older. The test is performed with frequency-specific stimuli and presentation levels to approximate hearing threshold levels. Diagnostic ABR provides information that is accurate enough to allow for therapeutic intervention. Hearing aids can be fitted with information obtained from a diagnostic ABR. Audiologic assessment and intervention is

TABLE 7 Developmental/Behavioral Screening Tools

Resource	Description	Age Range	Where to Find
General developmental screening tools			
Ages & Stages Questionnaire (ASQ)	A series of 19 questionnaires used to screen infants and young children for developmental delays during the first 5 y of life	4–60 mo	www.brookespublishing.com/tools/asq/index.htm
Ages & Stages Questionnaire: Social-Emotional (ASQ:SE)	A series of 19 questionnaires used to screen infants and young children at risk for social or emotional difficulties, to identify behaviors of concern to caregivers, and to identify any need for further assessment	6–60 mo	www.brookespublishing.com/tools/asqse/index.htm
Parents' Evaluation of Developmental Status (PEDS)	A method for detecting developmental and behavioral-emotional problems in children	Birth to 8 y	www.pedstest.com
Parents' Evaluation of Developmental Status-Developmental Milestones (PEDS:DM)	A collection of 6–8 items per age/encounter designed to replace informal milestones checklists with highly accurate items known to predict developmental status	Birth to 11 y	www.pedstest.com/dm
Autism-specific screening tools			
Checklist for Autism in Toddlers (CHAT)	A screening tool for early detection of autism	18 to \geq 24 mo	www.autismresearchcentre.com/tests/chat_test.asp
Checklist for Autism in Toddlers (CHAT), Denver Modifications	CHAT scoring modifications	18 to \geq 24 mo	
Checklist for Autism in Toddlers-23 (CHAT-23)	Combination of M-CHAT and CHAT items	16–86 mo	
Childhood Asperger Syndrome Test (CAST)	A parental questionnaire to screen for autism spectrum conditions	4–11 y	www.autismresearchcentre.com/tests/cast_test.asp
Modified Checklist for Autism in Toddlers (M-CHAT)	23-item scale pointing to express interest, responsiveness to name, interest in peers, showing behavior, response to joint attention, social imitation	16–48 mo	http://depts.washington.edu/dbped/Screening%20Tools/MCHAT.doc
Pervasive Developmental Disorders Screening Test-II, Primary Care Screener (PDDST-II PCS)	A parental questionnaire to screen for autism spectrum conditions	18–48 mo	www.pearson-uk.com/product.aspx?n=1315&skey=2960
Autism-specific or psychosocial screening tools Pediatric intake form from <i>Bright Futures</i>	Questionnaire to help gather a general understanding of the history, functioning, questions and concerns of the family	Birth to 21 y	www.brightfutures.org/mentalhealth/pdf/professionals/ped_intake_form.pdf
ADHD screening tools			
Vanderbilt rating forms	Parent- and teacher-completed forms that help a clinician diagnosis ADHD and to categorize the problem into 1 of its various subtypes	6–12 y	www.brightfutures.org/mentalhealth/pdf/professionals/bridges/adhd.pdf
AAP ADHD toolkit	A comprehensive toolkit developed from evidence-based guidelines for the diagnosis and treatment of children with ADHD; this resource toolkit contains a wide array of screening, diagnosis, treatment, and support materials for clinicians and other health care professionals	6–12 y	www.aap.org

ADHD indicates attention-deficit/hyperactivity disorder.

an ongoing process. The child requires regular audiologic reevaluations to determine if there is fluctuating or progressive hearing loss. Middle-ear monitoring is also essential. Hearing aid selection, fitting, verification, and validation require ongoing and regular visits with the audiologist. Candidacy

for cochlear implantation should be considered when there is limited residual hearing or when progress with amplification is insufficient. Recommendations to the family regarding cochlear implantation should be based on a team evaluation that includes audiology, otology, psychology, speech-

language pathology, and other intervention personnel.

Most providers of pediatric health care realize the importance of referring to an otolaryngologist, an audiologist, and a speech-language pathologist. Less recognized is the potential

TABLE 8 Guidelines for Children with Abnormal Speech Development

Age, mo	Referral Guidelines for Children With "Speech" Delay
12	No differentiated babbling or vocal imitation
18	No use of single words
24	Single-word vocabulary of ≤ 10 words
30	Fewer than 100 words; no evidence of 2-word combinations; unintelligible
36	Fewer than 200 words; no use of telegraphic sentences; clarity $< 50\%$
48	Fewer than 600 words; no use of single sentences; clarity $\leq 80\%$

Source: Matkin ND. *Pediatr Rev.* 1984;6:151.

TABLE 9 Guidelines for Children With Suspected Hearing Loss

Age, mo	Normal Development
0–4	Should startle to loud sounds, quiet to mother's voice, momentarily cease activity when sound is presented at a conversational level
5–6	Should correctly localize to sound presented in a horizontal plane, begin to imitate sounds in own speech repertoire or at least reciprocally vocalize with an adult
7–12	Should correctly localize to sound presented in any plane, should respond to name, even when spoken quietly
13–15	Should point toward an unexpected sound or to familiar objects or persons when asked
16–18	Should follow simple directions without gesture or other visual cues; can be trained to reach toward an interesting toy at midline when a sound is presented
19–24	Should point to body parts when asked; by 21 mo, can be trained to perform play audiometry

Source: Matkin ND. *Pediatr Rev.* 1984;6:151.

benefit of genetic and ophthalmologic evaluation of children and infants with sensorineural hearing loss.¹⁷

Especially in young infants and children, referral to an otolaryngologist for a complete diagnostic assessment is essential. More than 90% of the children with permanent hearing loss are born to "hearing" parents, resulting in a sense of urgency for parents of newly identified children with hearing loss to seek out answers regarding etiology, risk of progression, implications, and other questions. If children are indeed identified with sensorineural hearing loss, a variety of diagnostic tests can be recommended depending on the patient's history and physical examination. Otolaryngologists may play a role in diagnosis and treating middle-ear fluid or other middle-ear disorders as well as assisting in the definitive diagnosis of the cause of sensorineural hearing loss.¹⁸ Diagnostic testing may include imaging of the temporal bone to identify structural defects; genetic tests, such as for abnormalities of the *Connexin* gene; and,

occasionally, evaluation for other metabolic defects. Evaluation by a geneticist and genetic testing can be important for diagnosis as well as for providing the family with information for future planning purposes.

The hearing health care team (comprising the audiologist, otolaryngologist, teachers of the child with hearing impairment, speech-language pathologists, and other educational and medical personnel) should assist the family with intervention for hearing loss. Interventions may include observation with increased attention to speech and language development, hearing aids, auditory-assisted systems for the school environment, or more invasive surgical hearing devices such as cochlear implants or bone-anchored hearing aids. The goal is to provide families with appropriate options so that they may make well-informed decisions. Interventions should be driven by family desires and guided by accurate and timely information from all hearing-related health care professionals. Family goals and expectations are influenced by culture, parental

education, level of income, availability of local resources, language in the home, and more. The role of the hearing health care team is to assist families in identifying all the options available to them and to support them throughout the ongoing decision-making processes that will occur throughout the child's development. All members of the hearing health care team, in conjunction with parents and on the basis of informed choice, should recognize that no decision regarding intervention is "final," and periodic opportunities should be identified for discussion regarding progress, alternative interventions, and new developments.

Medical follow-up includes ongoing evaluation and management of the adequacy of hearing rehabilitation; observation for potential complications of hearing rehabilitation, such as otitis externa and cerumen impactions; and monitoring for appropriate speech and language development.

Speech and language evaluation by a speech-language pathologist with training in working with children with hearing loss is also important for documentation of baseline speech and language skills and implementing a program of intervention that reflects the family's choice regarding language development.

At least one third of children with hearing loss will have an additional coexisting condition.³ Because many causes of hearing loss are associated with abnormal ophthalmologic findings, formal ophthalmologic evaluation is appropriate, not only to assist with the diagnosis but also to optimize vision. A diagnosis of Usher syndrome with associated progressive hearing and vision loss may influence communication choices.

Children with hearing loss should also be monitored for developmental and behavioral problems (attention-deficit/hyperactivity disorder, autism, learning disabilities) and referred for

additional evaluation when necessary. Health care professionals can use screening tools to evaluate young children periodically for such concerns (Table 7) and refer for additional evaluation when concerns arise.

A medical professional should participate as an active member of a family's hearing health care team after diagnosis and provide input to assist in the adequacy of the rehabilitative efforts to monitor the child for progression and additional disabilities.

HEARING REFERRAL RESOURCES

Key point 9: Abnormal hearing test results require intervention and clinically appropriate referral, including otolaryngology, audiology, speech-language pathology, genetics, and early intervention. Pediatric health care professionals should maintain a list of referral re-

sources available in their community for children with hearing loss and should advocate for increasing options and choices for families. Otolaryngologists, audiologists, and speech-language pathologists with special training and experience in treating children should be consulted for specific diagnosis, counseling, and treatment. Pediatric health care professionals should collaborate to refer the child for comprehensive educational counseling and treatment services. Communication among professionals caring for a child with hearing loss is essential to ensure appropriate case management.

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Tool H5

*“1-3-6 Primary Care Provider
Patient Care Plan”*

Early Hearing Detection and Intervention (EHDI) “1-3-6” PCP Patient Care Plan

Patient Name: _____ Birth Facility: _____
 DOB: _____

Before One (1) Month:

Hearing Screening Results (OAE/AABR)

Right Ear: Pass Refer Unknown
 Left Ear: Pass Refer Unknown

Chart Documents

Hospital Screening Results: Yes No
 Audiological Evaluation Results: Yes No
 Care Management Checklist: Yes No

Before Three (3) Months:

Audiologic (Hearing) Evaluation

Date Completed: _____ Yes No
 Normal Hearing: _____ Yes No
 Permanent Childhood Hearing Loss: Yes No
 Otolaryngology Referral: Yes No
 Genetics Referral: Yes No
 Ophthalmology Referral: Yes No
 Other Referrals: _____

Before Six (6) Months:

Enrollment in Early Intervention

M.D. approval for hearing aids Yes No
 Completion of hearing aid evaluation Yes No
 Fitting of hearing aids Yes No

Risk Factors and Hearing Loss: Did or does this child have any of the following risk factors for hearing loss? Yes No

Family history of congenital childhood hearing loss _____ In-utero infection _____ Oto-toxic medications _____ Spinal Meningitis _____
 Hyperbilirubinemia _____ Craniofacial Anomalies (also note ear tags, pits or malformations) _____ Spent 5 days or longer in NICU _____
 Serious head injury _____ Parent concern _____

Diagnosis-Related Procedures and Documentation

Procedure	Specialist/Professional	Ordered	Reviewed	Results
Hearing Screening	Hospital Screening Personnel			
Audiologic (Hearing) Evaluations (confirmation of hearing loss)	Audiologist			
ENT Evaluation	Otolaryngologist/Otologist			
Medical Work-up for Sensorineural Hearing Loss	Otolaryngologist/Otologist			
Genetics Work-up	Geneticist and Genetic Counselor			
Ophthalmology	Ophthalmologist			
Individualized Family Services Plan (IFSP) from Early Intervention	Intake Coordinator			

This child’s audiologist is: _____
 This child’s otolaryngologist is: _____

PCP/Medical Home Care Management Checklist for Children with Confirmed Permanent Childhood Hearing Loss

History and Examination Following Diagnosis:

- 1-3-6!** Did you know? Children with hearing loss who are identified and in direct early services before age 6 months have an excellent opportunity to have age-level communications skills by the time they enter regular kindergarten.
- Review child and family history.**
- Ensure that chart contains diagnosis documents.** Review the audiologist's report that confirms the diagnosis of hearing loss with the parents.
- Establish that child has an otolaryngologist and that etiology studies have been conducted.** Every infant with confirmed hearing loss should be evaluated by an otolaryngologist who has knowledge of pediatric hearing loss.
- Offer genetics evaluation and counseling.** 50 % of hearing losses are genetic. Some are associated with syndromes. The most common organs involved are eyes, heart, kidneys, thyroid, and bones.
- Refer for ophthalmologic evaluation.** Every infant with confirmed hearing loss should have at least one (1) examination to assess visual acuity.
- Encourage and assist family (if needed) in continuing regular audiology follow-up.** A list of audiologists who provide services to babies and children can be found at www.region4genetics.org.
- Ask about amplification.** Amplification devices, including hearing aids, may provide assistance to infants as young as four weeks. Early and consistent daily use of amplification should allow a child to develop listening skills that will assist in hearing conversation more easily. For those children with severe to profound hearing losses, a cochlear implant may be a better solution to hearing. Cochlear implants are available for children, who are candidates, at age 12 months, or earlier in some cases. For a variety of reasons, some parents will choose to forego amplification for their child.
- Ensure early intervention.** There is no advantage in delaying early intervention services. Ask the family if they are involved with Early Intervention (privately), ask if they need assistance from you. Research shows typical or near-typical language development in children who receive intervention before 6 months of age. Tell the family of the importance of working with early intervention providers who are skilled and experienced in teaching children and families to communicate in the method(s) this family has chosen or will choose.
- Monitor middle ear status.** This is especially critical in children with confirmed hearing loss as middle ear effusion may further compromise hearing.
- Vaccinate as appropriate.** Note that children with cochlear implants may be at higher risk for meningitis. Make sure they are up to date on their Haemophilus influenzae type b and pneumococcal immunizations. Refer to www.CDC.gov/hcbddd/ehdi/cochlear for recommendations.
- Discuss family support opportunities.** Be aware that many families will experience the same grief that accompanies other significant diagnoses of the newborn. In addition, the family may benefit from contact with people who are deaf or hard of hearing. Provide the family with names of state or local organization that provide information and support (see www.region4genetics.org).
- Discuss communication between the parents and child.** There are a number of methods available to teach children who are deaf or hard of hearing to communicate and there is “no one-size fits all approach”. Encourage the family to explore all options before making decisions. Talking with professionals, other parents, and children and adults who are deaf or hard of hearing can provide a variety of perspectives. Parents will likely hear different and opposing opinions regarding communication methods. Ultimately, it is up to the child's family or primary caregivers to decide what choice(s) will work best for their child and family to meet the child's desired outcomes.



Tool I

*“Increasing Genetic Referrals for Children
Identified Through EHDI – An Action
Guide for Providers Serving Children Who
Have Hearing Loss”*

*Please access this guide online at
[www.region4genetics.org/region4_products/
genetics_hearingloss.aspx](http://www.region4genetics.org/region4_products/genetics_hearingloss.aspx)*



www.region4genetics.org

Contact us at: info@region4genetics.org