



# National EHDI Conference

March 2, 2010

## Promoting the Genetic Referral through the EHDI Process

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## History of the Regional Collaboratives

- Established under Title XXVI of the Children's Health Care Act of 2000, "Screening for Heritable Disorders"
- "...The Secretary shall award grants to eligible entities to enhance, improve or expand the ability of State and local public health agencies to provide screening, counseling or health care services to newborns and children having or at risk for heritable disorders..."





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## Role of the Regional Collaboratives

- Ensure that children with heritable disorders and their families have access to quality care and appropriate genetic expertise and information in the context of a medical home that provides accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally effective care.
  - To strengthen communication and collaboration among public health, individuals, families, primary care providers, and genetic medicine and other subspecialty providers.
  - To quantitatively and qualitatively evaluate outcomes of projects undertaken to accomplish their goals.



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## Our Vision

- All newborns will receive state-of-the-art newborn screening and follow-up; children and youth with heritable disorders will have access to genetic expertise and coordinated care in the context of a medical home.





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## Our Mission

- Increase access to information about newborn screening and genetic resources, services and family support systems
- Facilitate data collection and analysis to guide decision-making regarding screening cut-offs, diagnosis and long term treatment of heritable disorders
- Support state public health agencies in improving infrastructure for genetic service delivery to children with heritable disorders
- Provide a forum for families, public health, and clinical providers to share best practices and models for improving newborn screening, follow-up and genetic care coordination
- Link Region 4 states with regional and national initiatives for improving the quality of newborn screening and genetic service delivery





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## Assumptions

**Effective follow-up is most successful when:**

- **“Just in time” information**
  - **Family**
  - **Health care provider /medical home**

**Existing service systems**

- **Provide an opportunity to identify children who could benefit from genetic services**



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## Region 4 Outcome

Improve follow-up practices for children with heritable disorders who are identified through NBS, EHDI, Birth Defect registries and early intervention





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## Workgroup Members

**Representatives from all 7 Region 4 state  
EHDI programs**

**Representatives from State Departments  
of Health**



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## Workgroup Focus

- **Develop and distribute follow-up protocols for providing information to primary care providers and families at the time of identification of hearing loss**
- **Improve communication practices between those identifying children with heritable disorders and the primary care physician; resulting in...**



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## Outcome

**....increased access to genetic services  
for children identified through the EHDI  
process.**



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## Putting it all together

### June 2009

- **Draft product – EHDI Follow-up Guide**
  - Opportunities for appropriate referral for genetic services
  - Tools and model practices to facilitate referrals



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## Challenges

### **Stakeholder groups**

- **Respecting the culture of the deaf and hard of hearing community re:genetics**

### **Variation by State**

- **Follow-up**
- **Structure within the state system**



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## Workgroup Assumptions

- **There are opportunities for providing information about genetic consultation throughout the EHDI process**
- **Families should have the opportunity for genetic consultation**
- **Families need to be informed of the opportunity for genetic referral/consultation**
- **Providers need to understand the value of genetic referral/consultation for families who have a child with permanent hearing loss**



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## Workgroup Assumptions

- **Providers need to educate families about the opportunity for genetic referral/consultation**
- **Parents who understand the benefits of genetic consultation are more likely to obtain genetic services**
- **Providers who understand the importance of genetic consultation for families who have a child with hearing loss are more likely to refer or offer genetic referrals to those families**



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## Tool Kit

- **Develop materials for professionals**
- **Develop materials for parents**
- **Located on one website for easy access**





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## Today

- **PowerPoint Tool- Generic Version**
- **Review the Genetic Evaluation process**
  - Why, When, Who, Where
- **Discuss ways to facilitate the referral**
- **Available for use by providers**



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## Promoting the Genetic Referral through the EHDI Process



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## Did You Know?

- **3% of all babies are born with a birth defect**
- **1 in 9 children admitted to the hospital has a genetic disorder**
- **3-5 % of the population is born with a birth defect**
- **20-30% of all infant deaths are due to genetic disorders**



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## Introduction

- **Hearing loss is the most common congenital disorder**
- **Nearly half of the babies with hearing loss are due to genetic causes**
- **Connexin-26 (GJB2) accounts for more than one-half of the genetic causes of profound deafness**



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## Genetics and Hearing Loss

- **More than 400 syndromes are associated with deafness (ex. Usher, Waardenburg, Brachio-oto-renal, etc)**
- **More than 100 genes for non-syndromic deafness have been localized (ex. *GJB2* and *GJB6*)**



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## Hearing Loss Categories

- **Type, Degree and Configuration**
  - **Portion of the hearing system affected:**
    - Sensorineural, conductive, mixed, neural
    - Mild, moderate, severe, profound
    - Flat, rising, sloping
- **Genetic or Environmental**
  - Hereditary or acquired



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## If Genetic

- **Nonsyndromic:**
  - **70% of permanent childhood hearing loss**
    - Only obvious problem is hearing loss
    - Almost half are Connexin 26 defects
    - Most often sensorineural
- **Syndromic:**
  - **30% of permanent childhood hearing loss**
    - Other medical problems can be present
    - Varies widely, can be conductive, mixed or sensorineural



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## Support for Genetic Referral

- **The Joint Commission on Infant Hearing (JCIH) 2007 Position Statement states:**

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





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## Support for Genetic Referral

- **The American College of Medical Genetics (ACMG) recommends that all children with confirmed hearing loss be referred for genetic evaluation and counseling**
- **American Academy of Pediatrics**
- **Centers for Disease Control and Prevention**
- **National Institutes for Health**
- **Maternal and Child Health Bureau**



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## Purpose of the Referral

- **To determine the cause of the hearing loss**
  - **Why does our child have a hearing loss?**
- **To understand the cause and provide answers to many questions**
- **To identify other medical issues that are associated with hearing loss**



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## Purpose of the Referral

- **To coordinate available genetic testing**
- **To develop long term medical management plans based on associated conditions**
- **To determine recurrence risk**
  - **What is the chance that we might have a second child with hearing loss?**



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## Special Considerations

### Unique family situations

- There are times where making the referral immediately is of utmost importance
- There may be other circumstances where sensitivity to the family situation determines how and when a referral should be made



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## Immediate Referral

- **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 aminoglycosides**
  - **(there is an inherited susceptibility to hearing loss induced by these antibiotics)**



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## Families with a history of hearing loss

**The Medical Home/ENT should discuss the role of the genetic evaluation in determining possible medical management and intervention strategies**

**Parents/caregivers who are deaf may view hearing loss as a difference, not a disability**



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- **The majority of families (> 90 %) have no history of hearing loss**
- **With diagnosis, caregivers may be overwhelmed**





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## The Genetic Team

### Professionals include:

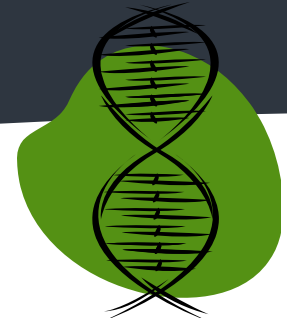
- **Clinical Geneticist: Physician (certified by ACMG) who diagnose and treat genetic conditions (usually trained in one or more other disciplines i.e. pediatrics, internal medicine, oncology, pathology)**
- **Genetic Counselor: (certified by ABGC) Master's level professional trained to:**
  - Recognize and assess genetic conditions
  - Educate families about genetic conditions and available testing or treatment options
  - Help families deal with the diagnosis of a medical condition





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## The Genetic Evaluation Process

- **Family case history (3-4 generations)**
- **Medical records review**
- **Comprehensive medical and developmental patient history**
- **Physical examination**
- **Discussing benefits, risks, limitations and costs of genetic testing**
- **Informed consent and confidentiality**
- **Obtain appropriate genetic testing as indicated**



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## Tests May Include

- **Chromosomal analysis**
- **Skeletal survey if short stature/disproportional growth**
- **Evaluation of other systems (renal, cardiac, skin, vision)**
- **CT or MRI of the brain**
- **Specialized genetic studies**



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## Genetic Counseling and Follow-Up

- **After testing, address the family's questions**
  - natural history of the condition
  - pattern of inheritance
  - medical implications
  - recurrence risks
  - associated health risks in others
- **If results are positive, meeting quickly with the family is important**



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## Genetic Counseling and Follow-Up

- **Provide information about available state and national resources**
- **Present the options available to families and discuss the benefits/risks of each option**
- **Arrange for referrals to other specialists when needed**
- **Assist with funding, if needed**



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## Funding



- **Often covered by third-party insurers, Children's Special Health Care Services, Medicaid**
- **May or may not be covered by private insurance**
- **Privacy issues may be addressed by self-pay when possible**



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## Benefits of Genetic Testing

- **Assists with establishing etiology of the hearing loss**
- **Can alleviate parental anxiety**
- **Sometimes identify risk for progression**
- **Helps to predict complications or other health conditions**
  - **Identifies potential risk of aminoglycoside ototoxicity**
- **Helps with planning treatment and follow-up**
- **Helps families make educated choices**



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## Limitations of Genetic Testing

- **May not always give clear answers**
- **Forces the family to confront difficult issues**
- **Ethical dilemmas in reproduction**
- **May generate concern over who has access to this information (self-pay rather than insurance)**
- **Can sometimes increase parental anxiety**



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## Who should be offered a referral?

- **All children with bilateral moderate, severe or profound sensorineural hearing loss**
- **A child with any degree of loss (unilateral or mild) where there is a family history of hearing loss or other medical problems**
- **Families who want to learn about the possible genetic cause of their child's loss**





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## When, By Whom and How

- Referral should be made as soon as possible once the diagnosis of hearing loss has been confirmed, ideally by 3-6 months of age
- Referral should be made by providers working with the family and may include the medical home (PCP), ENT, Audiologist, Early Interventionist, etc.
- The Region 4 Genetics Collaborative EHDI Follow-up Workgroup has developed a toolkit to assist providers in offering/making genetics referrals



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## Provider Responsibilities in the Referral Process

- **All providers should take responsibility for the genetics referral as soon as possible after confirmation of hearing loss**
- **This responsibility may include making the referral directly (usually a physician), or it may mean working with the medical home or primary care physician to encourage the referral**



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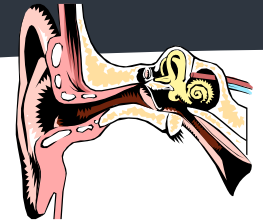
## Medical Provider's Responsibilities:

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



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## Otolaryngologist's Responsibilities

- **Recommend advanced diagnostic testing**
- **Be familiar with the indication and implications of genetic testing**
- **Complete History and Physical should be helpful in directing further testing**
  - **Acquired (meningitis)**
  - **Environmental (NOISE)**
  - **Family history**
  - **Obvious syndrome**



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## Audiologist's Responsibilities

- Provide complete test battery describing the hearing loss and tests performed (tympanometry, auditory brain response, otoacoustic emissions)
- Provide report to the medical home/primary care and recommend the need to offer genetic referral





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## Audiologist's Responsibilities

- **Counsel the family about how the genetic evaluation can determine etiology of the hearing loss and identify other health-related issues**
- **Ask the family if the physician has discussed this referral with them**
- **Provide materials about genetic evaluation for children with confirmed hearing loss and local genetics services**
- **Encourage the family to discuss genetic referral with the medical home**
- **Answer any questions the family may have**



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## Service Coordinator's Responsibilities

- **Counsel the family about the role of the genetic evaluation in determining etiology of the hearing loss and identifying other health-related issues**
- **Ensure the intervention plan includes information that develops from the genetic evaluation**
- **Know where to refer for a genetic evaluation**



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## Actions for All Providers

**At the initial visit with a child with confirmed hearing loss:**

- Ask if they have had a genetic consultation
- Give a brochure on genetics and hearing loss
- Provide contact information for local genetic services
- Offer information to assist them in asking their physician for a genetic referral
- Include a reminder in your records to check whether or not the genetic referral or services have been offered to the family
- Physician- order the evaluation





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## Actions for All Providers

- **Increase your knowledge of genetics and hearing loss**
- **Increase your comfort level in referring to genetic services**
- **View the web-based information on how, why, when to refer for genetic services ([Region4genetics.org](http://Region4genetics.org))**
- **Be prepared to respond to questions families may have**



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## Promoting the Referral

- To maximize the likelihood that a referral is both *made and followed up on by the family*, the offer 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



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## Promoting the Referral

***Implementing a public health model of systematically providing the information over time and through different venues increases the chance the suggestion will be acted on***



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## Tools to Use

- **Genetics and Hearing Loss (Brochure for Families)**
- **Learning about Hearing Loss – A Roadmap for Families (Brochure for Families)**
- **Region 4 State Genetic Services (Lists & National Resources to Identify Services Near You)**



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## Tools to Use

- **Questions You May Want to Ask Your Genetics Team (Brochure for Families, specific to hearing loss)**
- **Frequently Asked Questions about Genetics and Hearing Loss for Families (Web based resource)**
- **Frequently Asked Questions (FAQS) about Genetics and Hearing Loss for Providers (Web based resource)**



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## Tools to Use

- **Promoting the Genetic Referral through the EHDI Process (PowerPoint)**
- **Increasing Genetic Referrals for Children Identified through EHDI (Action Guide for Providers)**



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Available on.....

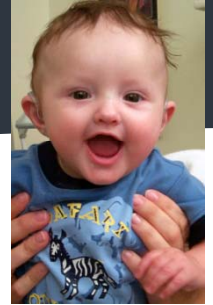
**Region 4 Website**

[www.region4genetics.org](http://www.region4genetics.org)



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## Future Directions

- **Currently only single genes can be screened**
- **New genes being identified all the time**
- **Development of effective gene therapies based on gene identification is still in infancy**
- **In the future, specific genes may be an accurate predictor of HL severity and prognosis to allow for appropriate habilitation during critical period for speech/language development**





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## References & Thanks to:

- **Region 4 EHDI Workgroup**
- **Minnesota's Physician Guidelines: Genetic Evaluation and Counseling for Children with a Confirmed Hearing Loss**
- **Michigan's EHDI Guidelines for Genetic Evaluation Referral**
- **Genetics of Hearing Loss: Presentation from Building Bridges Conference May 2009 Cincinnati Children's-Luis F Escobar, MD**
- **Epidemiology and Etiology of Pediatric SNHL; Presentation from Building Bridges Conference May 2009 Cincinnati – John H Greenwalk, Jr, MD**
- **Genetic Evaluation Guidelines for the Etiologic Diagnosis of Congenital Hearing Loss: American College of Medical Genetics 2007**