



**Region4**  
Genetics Collaborative



**Region 4 EHDl Follow-up Workgroup**  
**Telemeeting Agenda - Thursday, December 18, 2008**  
1/866/489-0573; \*4545164\*

- |  |                    |
|--|--------------------|
| I. Welcome & Introductions by State Roll Call  | Hoffman            |
| III. EHDl Registry   | Hiner/MN reps      |
| A. Update  |                    |
| B. Piloting in each state  |                    |
| III. EHDl Priorities & Genetic Issues  |                    |
| 1. Children with heritable disorders identified through EHDl in Region 4 – statement (Handout) | Kothari/OH<br>Reps |
| 2. Workgroup Charge, Action Plan (Handout)   | Hiner              |
| 3. Assessing workgroup priority issues for fit with genetic focus (Handout)                    | Pope               |
| IV. Products & Activities  |                    |
| 1. Border Baby Protocol (Handout)  | Pope               |
| 2. Genetics and Hearing Loss Brochure - Adapting MI tool (Handout)                             | Garcia             |
| 3. Form for collecting information on Risk Factors (Handout)                                   | Ehrhardt           |
| V. Announcements   | Group              |
| VI. Agenda additions   | Hoffman            |
| VII. Adjourn   | Hoffman            |



## Region 4 EHDI Follow-up Workgroup Telemeeting Notes - Thursday, November 20, 2008

**Participating:** Gary Hoffman, WI - Lead; Ginger Mullin, IL; Molly Pope, IN; Karen Mercer, KY; Barb Dalbec, Alison LaPointe, Yaoli Li, MN; Reena Kothari, Shelley Nottingham, OH; Ravi Shah, WI. Sally Hiner, Region 4 Genetics Collaborative Coordinator.

### **Announcements**

**I. EHDI Conference Abstract** – was developed by Molly, Ravi, Reena and Sally with an opportunity for input from all workgroup members. Our abstract was accepted. Our acceptance explained that many high quality abstracts which fit well with the conference theme were received, so the conference presentation sessions have been modified. We will be partnered with a compatible presentation for a one hour session. Sally will be pulling together the presentation team in the next week or so to respond to our presenter requirements and begin working on the presentation materials. The submitted abstract was included with the meeting materials.

**II. Carryforward Funds** – Region 4 submitted a request for use of unspent funds from last fiscal year's budget. We have received verbal notification that the request has been approved. Once we have official notification, plans for use of the carryforward funds will be publicized to the workgroups. Of interest to this workgroup are funds to support enrollment of patients into the EHDI Follow-up Registry and funds to support Region 4 staff to assist with the IRB process. Carryforward funds must be spent by 05/31/09.

### **III. EHDI Registry Proposed Elements**

The workgroup engaged in a facilitated review and discussion of the excel spreadsheet of proposed data elements for the EHDI Registry.

Next steps:

1. Sally will make changes discussed and, as realistic, highlight for easy review by the workgroup.
2. Sally will highlight "free text" options for Ravi to draft menu/check box options
3. Ravi will draft menu/check box options where reasonable to replace free-text.
4. Revised elements will be emailed for review and comment by workgroup members. Members will be asked to give particular attention to new menu's and re-ordering into sections (Risk Factor Monitoring, Diagnostic Monitoring, Follow-up and Tracking, Demographics, Birth Information, etc.)
5. Workgroup members are asked to consider other people who could potentially enter data into the registry and solicit review and input from them. Ravi agreed to request a review from WI Birth – 3. It was suggested that representatives from the CDC review, as well.

Questions and answers were discussed as follows:

Q1. Are all data fields required?

A1. No. We recognize that not everyone collects all the same information. Fill in what is relevant to your site.

Q2. Where does the data come from?

A2. The Registry is designed so that visit planners – both enrollment and interval – can be printed prior to the patient visit. Notes can be made on the hard copy forms and entered into the web-based registry later.

Q3. Who will be entering the data?

A3. This is yet to be determined for 6 of the 7 states. MN intends to “pilot” with the state Department of Health and a Multi-Disciplinary Hearing Clinic. Individual states will be asked to recruit sites to participate in “piloting” the registry with the enrollment and IRB support from the carryforward funds.

Q4. MN anticipates that several sources will enter the data for any one patient. Do certain fields the need to be protected so that information is not inadvertently changed?

A4. Yes, this is something that should be addressed after the registry is up.

Q5. How long does data entry take?

A5. From a completed visit planner, on average 25 minutes for enrollment survey and 15 minutes for interval survey.

Q6. Where is the registry located?

A6. It is web-based, HIPAA compliant, accessible by username and password from anywhere internet access is available.

Q7. How many cases do we expect each state to enroll?

A7. A number of cases enrolled for the region was included in the carryforward request. The actual number will be made available when we have official approval of our carryforward request.

#### **IV. December Meeting**

There are lots of items for our December meeting. Supporting materials already have been sent, so please prepare for discussion prior to the meeting. In addition to the items proposed, we will need to carry over from this meeting, discussion about piloting the EHDI Registry in each state. Proposed agenda follows:

- A. EHDI Priorities & Genetic Issues
  - 1. Children with heritable disorders identified through EHDI in Region 4 – statement
  - 2. Assessing workgroup priority issues for fit with genetic focus
- B. Products & Activities
  - 1. Border Baby Protocol
  - 2. Genetics and Hearing Loss Brochure (Adapting MI tool)
  - 3. Form for collecting information on Risk Factors
- C. EHDI Registry
  - 1. Update
  - 2. Piloting in each Region 4 State

Notes by Hiner



## EHDI Follow Up Workgroup: WorkPlan

Charge: Develop and Distribute best practice follow-up protocols/practice models for population-based identification of children with heritable disorders.			
Objective	Who	What	When
1. Expand Follow up WG to include representatives of EHDI	MPHI, AG	Identify and recruit members to participate in FWG	February 2008
2. Develop and disseminate follow-up protocols/practice models	FWG	Identify protocols/practice models for providing information at time of identification (positive screen, report, etc.)	March 2008
	MPHI	Compile and distribute list of protocols/practice models	
	FWG	Review, discuss and add samples identified to list of protocols/practice models	
	MPHI	Summarize compare and contrast identified protocols; disseminate summary	April 2008
	FWG	Review and discuss summary	May 2008
	FWG	Develop criteria for determining quality, cost and feasibility of replicating protocols/practice models	
	MPHI	Develop/modify assessment tool	
	FWG	Review and finalize assessment tool	
	FWG	Assess protocols/practice models using assessment tool	June 2008
	MPHI	Compile/summarize reviews	
FWG	Formulate recommendations based on reviews (e.g. adapt protocols/practices, recommend protocols/practice models)	July 2008	

<b>A. Promote the use of care plans specific to the needs of children with heritable disorders</b>			
Objective	Who	What	When
2. Develop and disseminate follow-up protocols/practice models (continued)	MPHI	Summarize recommendations	August 2008
	MPHI	Poll states to determine which practices models they would be willing to pilot	
	TBD	Implement selected practice models	August 2008-Y5
	Evaluate or	Evaluate implementation	Y2-Y5
	FWG	Adapt, refine practice models/protocols based on evaluation findings	Y3-5
	R4 Partners	Disseminate protocols and models	Y2-5

Region 4 Genetics Collaborative  
Proposed Priority Issue for EHDl Workgroup  
10/20/08

**Increase Access to Genetics Services for Children with Hearing Loss**

- 50% of children diagnosed with sensorineural hearing loss are due to genetics causes.
  - Of those, about 1/3 are affected with a complex medical syndrome.
  - Since the majority of genetic hearing loss is caused by recessive genes, family history is usually negative.
- Identification of associated features in hearing loss syndromes may have health saving or life saving implications.
- Audiologists working in EHDl programs need to be knowledgeable about genetics services in their state.
- Genetics programs in states need to be knowledgeable about the EHDl program in their state.
- Families of children diagnosed with hearing loss should receive information about genetics services as a complement to their child's health care.

**Can this group look at the diagnosed children in each state and determine if, if at all, these children are referred for genetics risk assessment, counseling, etc? Of those that are referred and receive genetics counseling, what is the breakdown of syndromic vs. non-syndromic losses, age of identification, etc? From this, we can formulate how to better educated families, professionals and others in EI about genetics and hearing loss, etc.**

**EHDI Follow up Workgroup  
Objective, Charge and Priority Issues**

***Proposed* EHDI/Genetics Objective: Increase access to genetic services for children with hearing loss.**

***Workgroup Charge: Develop and distribute best practice follow-up protocols/practice models for population based identification (i.e. children identified through EHDI) of children with heritable disorders***

Workgroup Identified Priority Issues	
A	(Follow-up) Getting data back from Early Intervention/Part C programs
B	(Referrals) Getting babies into diagnostic testing
C	(Border Babies) Don't know about border baby births/NBS
D	(Diagnosis) Following up with the PCP for older children – what is in the scope of the EHDI program
E	(Education) Educating practitioners about the importance of diagnostic testing
F	(Education) Education practitioners about the urgency of repeat testing
G	(Follow-up) Getting PCPs to support re-screen and/or follow-up by encouraging the family
H	(Follow-up) Providing information to primary care providers at the time of identification ~ positive screen or reports
I	(Follow-up) Providing information to families at the time of identification ~ positive screen or report

## GENETIC CENTERS IN MICHIGAN

### **Detroit Medical Center, Detroit**

Children's Hospital: Div. of Genetics &  
Metabolism (313) 745-4513

### **Henry Ford Hospital, Detroit**

Outreach clinics: Saginaw & Midland  
Dept. of Medical Genetics (313) 916-3188

### **Michigan State University, E. Lansing**

Outreach clinics: Flint & Kalamazoo  
Genetic Counseling Clinic (517) 353-2030

### **Spectrum Health, Grand Rapids**

Genetic Services (616) 391-2700

### **University of Michigan, Ann Arbor**

Outreach clinics: Marquette, Traverse City &  
Gaylord  
Pediatrics Genetics Clinic (734) 764-0579

### **William Beaumont Hospital, Royal Oak**

Pediatric Genetics Clinic (248) 551-0487

# DRAFT

## STATE OF MICHIGAN RESOURCES

### **Genetics Program**

E-mail: [genetics@michigan.gov](mailto:genetics@michigan.gov)  
Phone: (866) 852-1247 (toll-free)  
(517) 335-8887

Web Site: [www.MIGeneticsConnection.org](http://www.MIGeneticsConnection.org)

### **Early Hearing Detection and Intervention Program**

Phone: (517) 335-8884  
TTY: (517) 335-8246  
FAX: (517) 335-8036  
Web Site: [www.michigan.gov/ehdi](http://www.michigan.gov/ehdi)

### **Children's Special Health Care Services**

E-mail: [cshsfc@michigan.gov](mailto:cshsfc@michigan.gov)  
Phone: (800) 359-3722 (toll-free)  
Web Site: [www.michigan.gov/cshs](http://www.michigan.gov/cshs)

### **Early On<sup>®</sup> Michigan**

Phone: (800) Early-On or (800) 327-5966  
TTY: 517-668-2505  
Web Site: [www.1800earlyon.org](http://www.1800earlyon.org)

## Genetics

&

## Young Children with Hearing Loss



Early Hearing, Detection  
and Intervention Program



### Resources on the Web

**MI Genetics Connection**, Support Directory - [www.migeneticsconnection.org/supportdirectory.shtml](http://www.migeneticsconnection.org/supportdirectory.shtml)

Boys Town National Research Hospital - [www.boystownhospital.org/home.asp](http://www.boystownhospital.org/home.asp)

**National Institute on Deafness and Other Disorders** - [www.nidcd.nih.gov/index.asp](http://www.nidcd.nih.gov/index.asp)

Gene Reviews - [www.geneclinics.org/profiles/deafness-overview/details.html](http://www.geneclinics.org/profiles/deafness-overview/details.html)

**Raising Deaf Kids** (Children's Hospital of Philadelphia) - [www.raisingdeafkids.org](http://www.raisingdeafkids.org)

Centers for Disease Control and Prevention - [www.cdc.gov/NCBDDD/EHDI/genetics.htm](http://www.cdc.gov/NCBDDD/EHDI/genetics.htm)

**Newborn Screening ACT Sheets and Algorithms** -

<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>

*The Michigan EHDI Program recommends a genetic evaluation for babies and young children with hearing loss. Families may wonder why this is important. This pamphlet provides some information about genes and hearing loss. It also explains what to expect at a genetics clinic.*

## What are genes?

Genes are like a recipe for the human body. They tell all the parts of the body how to grow and function. A *genetic condition* occurs when a person has a change in a gene that prevents it from working correctly.

## How are genes related to hearing loss?

More than *one-half* of all cases of hearing loss are caused by genetic factors. This is true even when no other family members are known to have hearing loss. Genetic hearing loss may be linked to health or learning problems.

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

It is important to find out if your child is at risk for other health conditions. You might also want to know the chance for your other children to

have hearing loss or other problems. By knowing more about your child's hearing loss, you can best decide what to do for your child and family.

## How is hearing loss inherited?

Genetic hearing loss can be passed through a family in different ways. Sometimes hearing loss occurs in every generation. More often, only one child, or brothers and sisters, may be affected. Even if your child has no family history of hearing loss, the cause could still be genetic.

## 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 primary health provider, audiologist, and/or an *Early On*<sup>®</sup> provider can help you make the appointment and collect the medical information needed. You can also call one of the clinics listed on the back of this brochure.

## What happens at the genetics appointment?

Your child's health history and records will be reviewed. Your child's development will be discussed. You will also be asked questions about your family's medical history. A health exam will be done to look for signs of genetic hearing loss.

Based on the history and exam, the doctor will discuss with you whether any tests (such as blood or urine) are needed and what might be learned from them.

The doctor will then look at all the information and try to figure out if the hearing loss is caused by genetic or other factors. You will have time to discuss what has been learned with the genetics clinic staff. This may include suggestions for your child's future medical care. A report will also be sent to your child's doctor.

## What happens if the cause is not found?

More is being learned about genetics every day. If the cause of your child's hearing loss is not found the first time, your family may be asked to return in a year or two to see if there is any new information. If your child develops new medical or developmental problems, it may be time to revisit the genetic specialist.

## Will insurance cover the cost of the appointment?

The clinic visit and testing are covered by many insurance plans, including Medicaid and Children's Special Health Care Services. Check with your plan to be sure. If you are in a managed care plan, you will need a referral from your child's primary doctor. If you need help finding coverage, please e-mail [genetics@michigan.gov](mailto:genetics@michigan.gov) or call (866) 852-1247 (toll-free).

Hi to all!

I took our main priorities and tried to think of some concrete ideas that we could develop as a team to incorporate the genetics component into the EHDI process. I'm sure there are many other good ideas that I didn't think about, but perhaps we can use this as a starting point to move us in the right direction. Many of the ideas are repeated through out the document as the education priorities really seemed to overlap (at least to me) so it isn't quite as overwhelming as it first appears.

Thanks,

Molly Pope  
Indiana Lead Regional Consultant

### **Ideas for Region 4 Priorities**

- A. **(Follow-up) Getting data back from Early Intervention/Part C Programs**
- **Develop a general brochure for Part C that discusses the importance of and need for genetic referrals for infants with diagnosed hearing loss**
  - **Develop a universal FAX back form to gather information on children with diagnosed hearing loss**
- B. **(Referrals) Getting babies into diagnostic testing**
- **Regional training for audiologists regarding appropriate test protocols, including a section that directly addresses the genetic referral issues**
  - **Provide audiologists with a list of genetic contacts for each state or Region 4**
  - **Develop a brief power point on genetic issues as it relates to children with diagnosed hearing loss for the EHDI programs in Region 4 to incorporate into the audiology trainings and college training programs**
- C. **(Border Babies) Don't know about border baby births/NBS**
- **Adopt Region 4 State to state Newborn Screening Guidelines for EHDI**
  - **Continue to address the "sharing" of information between states in Region 4**
- D. **(Diagnosis) Following up with the PCP for older children – what is the scope of the EHDI program (after 1 Year of age)**
- **Review spreadsheet developed by Joan**
  - **Develop Region 4 guidelines for ways to obtain needed risk factor information**
- E. **(Education) Educating practitioners about the importance of diagnostic testing**
- **Develop Region 4 guidelines for physician follow-up of children with diagnosed hearing loss**
  - **Develop a system for how this information could/should be disseminated to physicians in Region 4**
- F. **(Education) Educating Practitioners about the urgency of repeat testing**

- Develop Region 4 Guidelines for babies who referred from UNHS for physician
  - Develop a bullet point brochure for physicians that includes genetic referral information
  - Develop a system for how this information could/should be disseminated to physicians in the Region
  - Power point (see above) could also be incorporated into any physician training
- G. (Follow up) Getting PCPs to support re-screen and/or follow-up by encouraging the family
- Develop Region 4 Guidelines for babies who referred from UNHS for physicians
  - Develop a bullet point brochure for physicians that includes genetic referral information
  - Develop a system for how this information could/should be disseminated to physicians in the Region
  - Power point (see above) could also be incorporated into any physician training
- H. (Follow-up) Providing information to primary care providers at the time of identification – positive screen or reports
- Develop Region 4 Guidelines for babies who referred from UNHS for physician
  - Develop a bullet point brochure for physicians that includes genetic referral information
  - Develop a system for how this information could/should be disseminated to physicians in the Region
  - Power point (see above) could also be incorporated into any physician training
- I. (Follow-up) Providing information to family at the time of identification~ positive screen or report
- Develop a general brochure for parents that discusses the importance of and need for genetic referrals for infants with diagnosed hearing loss
  - Develop state specific brochures with genetic contact information

**Early Hearing Detection and Intervention (EHDI)  
Region IV Programs  
Risk Factor Monitoring Survey**

Primary Reference: Early Hearing Detection and Intervention  
Program Guidance Manual  
<http://www.cdc.gov/ncbddd/ehdi/documents/EDHI%20Guidelines.pdf>

Appendix III  
Surveillance Data Items

**CDC Recommended:**

M = minimum (recommended for all state data systems; required for follow-up on universal NBHS and for full r  
C = core (recommended for complete state data systems  
E = enhanced (additional data useful for clinicians, enhanced tracking, or research)

Item	Need
Family History	M
In utero / Congenital infections	M
" CMV	M
" Rubella	M
" Syphilis	M
" Herpes	M
" Toxoplasmosis	M
" Other?	M
Low birth weight	C
" <1500 g	C
APGAR score 0-4 @ 1min or 0-6 @ 5 min	C
Birth Defects	
" Craniofacial (including those with morphologic	M
" Head and neck (e.g., cleft palate, ear tags)	C
" Findings associated with a syndrome known to	M
NICU admission	M
" # days (> 48 h)	M
" < 48 h	C
" 48 h to 28 d	C
Neonatal Indicators	C
" hyperbilirubinemia requiring exchange transfus	M
" Mechanical ventilation (5+ d)	C
" sistent pulmonary hypertension assoc w/mech v	C
" Conditions requiring ECMO	M
Neurodegenerative disorder	C
Ototoxic drugs (specify/list)	C
Bronchio-pulmonary dysplasia	C
Other risk factors at discharge	C
Parent or caregiver concern regarding hearing, speech, language , d	M
Stigmata or other findings associated with SNHL or conductive HL or	M
Postnatal infection	
" Bacterial meningitis	M
" Other associated w/HL	M
Syndromes associate (e.g., Neurofibromatosis (NF) 1 and 2, Usher S	M

Neurodegenerative di (e.g., Hunter sx, Charcot-Marie-Tooth)	M
Head trauma	M
Recurrent or persistent otitis media with effusion (OME) for at least 3	M
Other risk factors ider (describe)	E
Description	
" of syndromes and stigmata	C
" of neurologic disorders	C
" of infections	C
" of parental concerns	C
" of other risk factors	C

Reporting on national EMDI goals)

## EHDI State to State UNHS Follow-up Guidelines

1. When the newborn is a resident of one state but is born in a hospital in a bordering state:
  - a. The UNHS screen should be completed and recorded on the state-of-birth blood collection card
  - b. The screening results should be shared with the child's state-of-residence
  - c. Follow-up should occur in accordance with the UNHS screening policies of the state in which the child resides
  
2. When the newborn is born in one state but is transferred for critical care to a tertiary level NICU in a bordering state:
  - a. Same as 1.a The newborn UNHS screen should be completed and recorded on the state-of-birth blood collection card
  - b. If the UNHS screen is not obtained prior to transfer, the UNHS screen should be completed and recorded on the blood collection card of the state where the tertiary care NICU is located (border state)
  - c. The screening results should be shared with the child's state-of-residence
  - d. Follow-up should occur in accordance with the UNHS screening policies of the state in which the child resides
  
3. When the newborn's family permanently relocates to another state prior to the completion of re-screening or diagnostic testing
  - a. The EHDI Program in the child's state-of-birth should notify the EHDI Program in the family's new state-of-residence with results to date and the EHDI state-of-residence program should assume responsibility for any further follow-up that is needed
  - b. In addition, the EHDI Program in the child's state-of-birth should notify the parents and/or primary care provider of the need for further follow-up in the family's new state-of-residence
  - c. The EHDI contacts in the state-of-birth and the state-of-residence should collaborate on the follow-up process
  - d. When follow-up is performed by the EHDI Program in the state-of-residence, the final screen results or diagnostic results should be forwarded to the state-of-birth EHDI Program where the screening was initiated, so the case may be closed

The HIPAA Privacy Rule recognizes the need for public health programs to access protected health information (PHI) to conduct public health activities to prevent or control disease, injury or disability. The Privacy Rule\* expressly permits release of PHI relating to newborn screening, without individual authorization, from a covered entity to state public health departments or agencies contracted, by public health departments, to provide newborn screening follow-up.

\*<http://www.cdc.gov/mmwr/preview/mmwrhtml/m2e411a1.htm>