



# Region 4 Genetics Collaborative

WISCONSIN STAKEHOLDER MEETING – APRIL 22, 2009

Comfort Inn Suites – Lake Country

Pewaukee, Wisconsin

## Agenda

8:00	<b>Registration, breakfast &amp; networking</b>	
8:45	<b>Introductions</b>	Cameron
9:00	<b>Overview of Region 4</b> (Handout 1) <ul style="list-style-type: none"><li>▪ Federal legislation: Heritable Disorders Program</li><li>▪ Infrastructure between Region 4 and Wisconsin</li></ul>	Cameron
10:30	<b>15 minute Break</b>	
10:45	<b>Work Group Presentations</b> <ul style="list-style-type: none"><li>▪ Priority 1 (Handout 2)<ul style="list-style-type: none"><li>• Website Demonstration</li><li>• Sample Exchange</li></ul></li><li>▪ Priority 2 (Handout 3)</li><li>▪ EHDI (Handout 4)</li><li>▪ Endocrine Project (Handout 5)</li><li>▪ Medical Home Education (Handout 6)<ul style="list-style-type: none"><li>• Medical Home Guide</li></ul></li><li>▪ Care Coordination (Handout 7)<ul style="list-style-type: none"><li>• Care Coordination Plans</li><li>• MEMSCIS (Handout 8)</li></ul></li><li>▪ Genetic Expertise (Handout 9)<ul style="list-style-type: none"><li>• Access Survey (Handout 10)</li><li>• Telemedicine</li></ul></li></ul>	Hoffman Hiner Shah/Seeliger Hiner Fleischfresser Hiner Wedepohl
12:00	<b>Lunch</b>	
1:00	<b>Beyond Region 4</b> <ul style="list-style-type: none"><li>▪ Updates from HRSA</li><li>▪ Newborn Screening Translational Research Network (Handout 11)</li><li>▪ Emergency Preparedness (Handout 12)</li></ul>	Cameron
1:30	<b>Ideas for Regional Projects</b>	Katcher
2:15	<b>Discussion with stakeholders</b> <ul style="list-style-type: none"><li>▪ Region 4 Infrastructure</li><li>▪ Access to information</li></ul>	Katcher
2:45	<b>Adjourn/Set up for Discussion Groups</b>	Cameron
3:00	<b>Parent Discussion Groups</b>	



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## Ohio Collaborative Participants

Participant	Email	Region 4 Work Group Role
Joyce Andersen	joyce.andersen@dhs.wisconsin.gov	
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Elizabeth Seeliger	elizabeth.seeliger@dhs.wi.gov	EHDI
Ravi Shah	Ravi.Shah@dhs.wisconsin.gov	EHDI
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Meg Steimle	msteimle@chw.org	
Todd Varness	tvarness@uwhealth.org	



# Region 4 Genetics Collaborative

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Holiday Inn Country Side - LaGrange

Country Side, Illinois

## Region 4 Genetics Collaborative Work Group Descriptions

### Region 4 Advisory Group

The Advisory Group supports the success of projects and activities by providing input, guidance, and assistance in addressing challenges. The Advisory Group includes representatives from each state in Region 4, family members, priority project leaders, and workgroup leads and co-leads.

### Care Coordination Workgroup

Care coordination engages families in development of a proactive care plan and links them to needed services and supports that address the full range of their needs and concerns. It is an essential component of an effective system of care designed to optimize the physical and emotional health and well-being of the child, improving the child's and family's quality of life. Workgroup activities will focus on improving care coordination for children with heritable disorders in Region 4.

### Endocrine Project Workgroup

Endocrinologists and Region 4 State newborn screening, lab and follow-up programs participate in this project to maximize the effectiveness of newborn screening for CH and CAH by improving detection and management of endocrine cases as part of existing newborn screening and follow-up programs.

### Follow-up Workgroup

Effective follow-up for children with heritable disorders provides “just in time information” to the child's family and medical home. Statewide service systems such as newborn screening, early hearing detection programs, birth defects registries and early intervention programs provide opportunities to identify children with heritable disorders and initiate the continuum of follow-up activities. The Follow-Up Workgroup will focus on protocols and practices for providing information to primary care providers and families at the time of identification (positive screen or report) and improving communication practices between those identifying children with a heritable disorder and the primary care physician.

### Genetic Expertise Workgroup

Access to genetic expertise is differentially available to individuals with rare genetic conditions and their families who: 1) are aware that such services even exist; 2) have the financial means to travel or by chance, live in close geographic proximity to the relevant specialist; 3) have a primary care provider who is able to stay abreast of new developments in testing and management for specific disorders; or 4) have the ability to find and interpret sophisticated information on their own. In addition, relatively few health care professionals in the United States are available with expertise to follow-up, diagnose and treat patients with some of these uncommon concerns. The Genetic Expertise Workgroup will focus on improving access to genetic expertise for underserved populations and maximizing time of genetic specialists and genetic counselors so expertise is ultimately available to more families.

### **IBEM-IS Workgroup**

Made up of clinical genetic-metabolic disease specialists, state Departments of Health staff, parents, epidemiologists, and care coordinators, the Priority 2 workgroup has developed and is implementing a comprehensive Inborn Errors of Metabolism Information System (IBEM-IS). This HIPAA compliant disease registry will include all diseases screened for using tandem mass spectrometry (MS/MS). To extend utility of the IBEM-IS, activities of the Priority 2 workgroup will include facilitating direct linkages with other electronic records and with data from newborn screening programs. It is anticipated that the IBEM-IS will result in the development of evidence-based treatment protocols, potentially improving the care, health and quality of life for children diagnosed with these rare conditions.

### **Medical Home Education Workgroup**

The term "medical home" describes an approach to providing comprehensive primary care that is coordinated and family centered, meeting the needs of the child and supporting the family. The goal of the Medical Home Education Workgroup is to educate families and health care providers about the importance of a medical home for children with inherited conditions. Group activities will focus on creating learning opportunities in Region 4.

### **Transition Workgroup**

Transition from youth to adulthood involves many factors: education, employment, finances, home living, community living, recreation and leisure, personal responsibility. For the individual with a heritable disorder, transition includes issues related to medical care and disease management such as identifying adult health care providers knowledgeable about specific conditions, developing new relationships with adult health care providers, adapting to new service delivery approaches, changes in health care coverage and insurance, and learning to manage one's own health care.

Transition is a process, not an event that should occur gradually, in concert with adolescent and family development. Including parents, pediatric providers, specialists and adult providers in the process is essential to smooth, coordinated, comprehensive transition. The Transition Workgroup will focus on identifying and promoting effective models addressing transition to adult services for youth with heritable disorders

### **Reimbursement Workgroup**

Children with heritable disorders often have complex needs, best addressed through services integral to the medical home model. While responsibilities associated with care coordination require a good deal of time on the part of physicians and other staff, often they are not reimbursed. Reimbursement issues may stem from billing policies of the providers, reimbursement policies and practices of third party payers, or both.

Medical foods and formulas, which can be extremely expensive, are not always covered by private insurance or programs such as Medicaid. Also, children with some rare heritable conditions require special foods and formulas in order to maintain health and avoid serious consequences including medical emergencies. The Reimbursement Workgroup will focus on addressing reimbursement issues that are barriers to quality care.

**Region 4 Advisory Group**

**Cynthia Cameron, Ph.D. DIRECTOR**  
**Janice Bach, CO-DIRECTOR**  
**Sally Hiner, Region 4 Coordinator**  
**Jodi Griffin, Project Coordinator**  
**Sarah Wedepohl, Parent Coordinator**

**Priority 1 Project: NBS by MS/MS**

**Improve Newborn Screening**

**Project Lead**  
**Piero Rinaldo, M.D. Ph.D.**

**Base Project Goal:**

Improve access to:  
**Genetic services**  
**Genetic expertise**  
**Genetic information**

**In the context of the medical home**

**Priority 2 Project: IBEM-IS**

**Gather information about treatment and outcomes**


**Project Lead**  
**Susan Berry M.D.**

**Base Project Workgroups**

**Care Coordination**  
**Endocrine Project**  
**Follow-up**  
**Genetic Expertise**

**Medical Home Education**  
**Reimbursement**  
**Transition**


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## State of the Region

Cynthia Cameron, PhD  
Director  
Region 4 Genetics Collaborative


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## Why We Are Together

- To share information about Region 4 activities and opportunities
- To strengthen relationships among stakeholder groups and Region 4
- To provide information on relevant projects at the national level and from other Regions
- To learn what regional activities you would like to initiate, expand or enhance


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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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www.thehrsa.com/region4

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## Region 4

- One of 7 regional collaboratives
- 2004 start up
- 2007 - Region 4 awarded 5-year, \$5,000,000 grant from HRSA
  - Base funding - \$500,000
  - Priority 1 - \$250,000
  - Priority 2 - \$250,000

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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
  - Region 4 website
  - Family is the Center of the Medical Home
  - On-line course

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## Our Mission

- Facilitate data collection and analysis to guide decision-making regarding screening cut-offs, diagnosis and long term treatment of heritable disorders
  - Laboratory Quality Improvement of Newborn Screening by MS/MS
  - Inborn Errors of Metabolism Information System
  - CAH disease registry

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## Our Mission

- Support state public health agencies in improving infrastructure for genetic service delivery to children with heritable disorders
  - Short term follow up protocols
  - CH follow up project
  - EHDI follow up protocols

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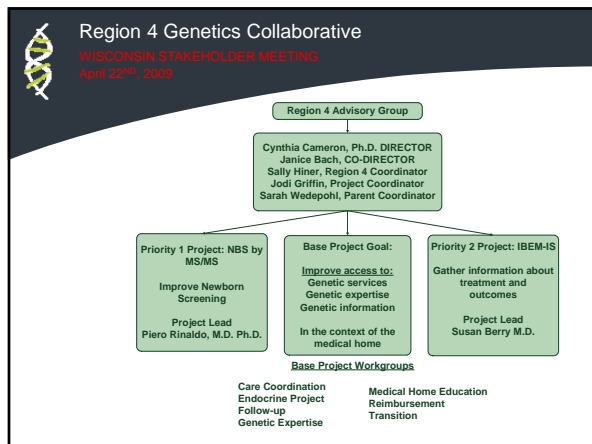
## Our Mission

- 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
  - Regional meetings
  - Workgroups
  - National Advisory Boards
    - Priority 1
    - Priority 2
    - MEMSCIS

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## Our Mission

- Link Region 4 states with regional and national initiatives for improving the quality of newborn screening and genetic service delivery
  - Region 4 and Mountain States
    - Interoperability between clinical/public health long-term follow up systems
  - Region 4 and Region 3
    - Survey on reimbursement for medical foods and formulas
  - Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
    - Family Survey on Medical Foods and Formulas
  - Newborn Screening Translational Research Network
  - National Coordinating Center
    - Emergency Preparedness
    - Data Coordination



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- ## Where We Are Going
- Increase collaboration with national partners
    - Genetic Alliance
    - American Academy of Pediatrics
  - Cultivate partnerships within Region 4 and across other regions
  - Support implementation of processes and products developed by Region 4
  - Leverage funds to expand Region 4 projects

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## Priority 2

Long-term Follow-up and Clinical Outcome Workgroup (LTFWG)

The Inborn Errors of Metabolism Information System (IBEM-IS)

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## Expanded NBS – A national priority

- Justice: all should be screened equally
- NBS should improve outcomes and save lives
- NBS is only as effective as the care it prompts
- Collaboration between screening, short-term and long-term team members is critical to improved outcomes
- Data sharing is essential

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## Advancing care for babies in Region 4

- Approximately 730,000 births per year
- Commitment to cooperative interaction
  - Clinicians
  - State Departments of Health
  - Parent representation
- All states screening by MS/MS (tandem mass spectrometry)
- Selected disorders common enough to develop protocols BUT only via multistate-multicenter collaborations
- (MCADD/yr in WI = <1/yr in Region 4 = 45-50)

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## Region 4 Collaborators Our P2 Workgroup

Metabolic clinicians and state health department NBS Specialists

Wisconsin


- Bill Rhead
- Abby Donovan
- Sandy vanCalcar



Region 4

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## Treating IBEM

- How did your mentor treat?
- What does the (text/manual/guide) say?
- What have you learned from experience?
- What does Metab-L say?

*Tend not to have uniform or organized strategy.  
Almost no clinical trial-based care, though substantial clinical experience*

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## Priority 2 Project IBEM-IS

- Develop and implement web-based inborn errors of metabolism information system (IBEM-IS)
  - Long-term management
  - Define “natural” history
  - Platform to integrate other critical data

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## Objectives

- Refine research and intellectual questions
  - Because each disease is rare, no single center can achieve statistical power to assess outcomes or treatment
- Expand spectrum and power of data by adding other disorders and other regions
- Integrate registry with other information systems
  - Web-based emergency plans, state NBS data systems, electronic medical records
- Evaluate and report results

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## MCADD: Initiation of Registry

- Review literature and existing treatment plans to define disease specific elements
  - Included elements contributed by Mountain states collaborative
- Identify *essential elements* for uniform data collection
- Identify *anecdotal elements* subject to randomization

All states contributed protocols so elements of difference could be characterized  
Parents involved with all aspects of planning

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## Critical components of IBEM-IS

- Ease of entry at point of service
  - Enrollment data, interval history
- Web-based with option of paper checklist for use during visits "visit planner"
- HIPAA compliant
- Local control of PHI (protected health information)
- Reporting functions
- All participants access
  - Varying levels
- Ease of analysis

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## Progress

- Elements defined for
  - all Fatty Acid Oxidation Disorders (FAOD)
  - MSUD (Maple Syrup Urine Disease) an aminoacidopathy
  - C3, C5OH disorders (organic acidemias) – includes Biotinidase
  - Galactosemia, Gluteric Acidemia-1
- Next – tyrosinemia

*Total number of conditions defined = 33*

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## Progress

- Training for 8 centers in 6 Region 4 states
- One R4 state still pending IRB approval
- Data sets posted:
  - <http://region4genetics.org/region4/dataelementssurveys.aspx>
- Data Entry
 

– MCADD	45
– MSUD	7
– LCFAOD	5
– SCADD	3

Total = 60 patients

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## National Expansion



- Arkansas
- Iowa
- Missouri
- Nebraska
- Oklahoma

**Region 4**

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## What next?

- Define strategy/research agenda
  - Uniform follow-up and reporting
  - Consensus on questions
- Add more disorders
- Collaboration with others
  - All clinics in Region 4
  - Clinics in other genetics collaborative regions
- Respond to data project requests
  - Impact on families
  - Assessing early MCADD complications
- Explore integration of data
  - Departments of health
  - Emergency information (MEMSCIS)

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## Acknowledgements

- Cindy Cameron, Region 4 Director
- Sally Hiner, Region 4 Coordinator
- Carolyn Anderson, Priority 2 Co-Lead
- Kristi Bentler, MN NBS Coordinator
- DocSite staff
- Anne Jurek, Epidemiologist

**All Priority 2 Workgroup Members**


- State Clinical Leads
  - Barbara Burton, IL
  - Bryan Hainline, IN
  - **Gordon Gowans, KY**
  - Gerald Feldman, MI
  - Sue Berry, MN (Also, Priority 2 Co-Lead)
  - Nancy Leslie, OH
  - Bill Rhead, WI
- Parent representatives
  - Darin Erickson
  - Kathy Stagni

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## Early Hearing Detection & Intervention Follow-up

Ravi Shah

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## From the Region 4 Mission


- Increase access to information about newborn screening and genetic resources, services and family support systems
- Support state public health agencies in improving infrastructure for genetic service delivery to children with heritable disorders

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## Current Practices

To ensure effective follow-up


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

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## Current Practices


Existing service systems

- Metabolic Screening
- Birth Defects Registry

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## Goal


Improve referral and follow-up practices for children identified through NBS, EHDI, and Birth Defect registries for genetics evaluation

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## Workgroup Focus

Develop and distribute follow-up protocols and practices for providing information to primary care providers and families at the time of diagnosis and improving communication practices between care providers along with communication to the families; resulting in increased access to genetic services for children identified through EHDI services.

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## Tasks

Determine needs in Region 4 and identify strategies to meet those needs:

- Collect and compare state follow-up protocols
- Identify & prioritize Issues
- Identify promising practices/strategies to address issues
- develop model practices and/or recommend practices for adaptation/use in Region 4


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## Accomplishments

- Review of
  - State EHDl Follow-up protocols/guidelines
  - Existing state infrastructure – how does EHDl fit with NBS? Genetics?
  - Survey of workgroup members to identify issues
    - Categorized and prioritized issues


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## Accomplishments

- Opportunity – EHDl Registry
  - May 2008, Chicago
  - Dec 2008 – consensus date elements
- Border Baby follow-up guidelines
  - Recommendation accepted by Advisory Group, Feb 2009
- Presentation at National EHDl Conference March 2009

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## Putting it all together


March 2009

- List of all EHDl partners
- Continuum of follow-up activities

April 2009

- Identified opportunities during EHDl follow-up for referral for genetic services

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## Putting it all together


May 2009

- Select strategies/tools to use to increase referrals at opportunities identified

June 2009


- Draft product – EHDl follow-up guide with identified opportunities for appropriate referral for genetic services specified. Guide will include tools and model practices to facilitate

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## Possible Tools

- Development of a Regional/State brochure for parent information kits
- Development of a power-point presentation for professionals discussing the need for and importance of a genetic referral for infants and toddlers with diagnosed follow-up
- Development of a fact sheet to be provided to the medical home at the time of notification of failed UNHS



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## Challenges

Stakeholder groups

- Appropriately and sensitively conveying the importance of Genetics testing to the DHH community.


Variation by State

- Follow-up procedures and workflow
- Structure within the state system

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## Early Hearing Detection & Intervention Follow-up

Ravi Shah

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## From the Region 4 Mission


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Determine needs in Region 4 and identify strategies to meet those needs:

- Collect and compare state follow-up protocols
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Stakeholder groups

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Variation by State

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- Structure within the state system


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## Endocrine Projects

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## History

Purpose: conduct an epidemiological evaluation of CH and CAH screening in at least four Region 4 states by:

- 1) determining case definitions used in each state;
- 2) describing current screening, follow-up and diagnostic methods;
- 3) evaluating and comparing false positive and negative rates and positive predictive values (PPV), and detection rates; and
- 4) providing consensus recommendations for improving detection rates and reducing false positive and negative rates in the region.

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## Activities to address

- Obtained data from participating states
  - Initial endocrine screening values
  - Follow-up data
- Developed state-by-state performance metrics
- Reviewed and compared Region 4 state short-term follow-up patient management algorithms

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## Activities to address

Convened a meeting in of lab representatives, follow-up staff and clinical endocrinologists within the region (Lansing, May 2007) to

- *discuss results and draft minimal guidelines for detection of CH and CAH in NICU and non-NICU populations.*
  - Presented preliminary findings
  - Identified issues
  - Formulated recommendations

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## Focus

1. Three-year follow up of CH detected by NBS
2. Modified 2<sup>nd</sup> Tier CAH Screening Protocol
3. Long-term follow-up disease Registry for CAH

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## CH 3-year Follow-up Issues

Standard of care is to follow-up CH cases until at least 3 years (AAP Guidelines)

- Diagnostic verification recommended (permanent vs. transient CH)
- Rate and outcome of diagnostic verification unknown

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## Opportunity for other Region 4 States

- Michigan 3-year follow-up pilot study
- March 2009 telemeeting
  - MI process, findings, tools for replication
    - Flow outline
    - Surveys
    - Fax forms
    - Telephone scripts

*All 5 participating states expressed interest*

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## CAH Screening Problems

- High false positive rate associated with neonatal stress and low positive predictive value
- Perceived low cost/benefit of CAH screening
- Prenatal steroid treatment
- False negative for classical simple virilizing CAH

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## Opportunity – Region 4 States

MI modified second tier screening pilot

- March 2009 Telemeeting
  - MI process, findings
  - Tools and assistance for replication available from MI team

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## CAH Long-term Follow-up

Objective

- to create CAH/DSD surveys for disease registry

Lead –


- Kyriakie Sarafoglou, M.D.; University of Minnesota

Process –

- workgroup telemeetings;
- email communications and reviews;
- Website postings; and
- face to face meetings

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## Benefits

Provide valuable information on a broader scale than can be obtained at a single institution


Catalyst to create collaborative research projects

- Examine effect of “stigma” on CAH patients
- Better methods of monitoring disease control

Improved understanding of the disease course and ways to improve outcomes

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## Accomplishments

- CAH surveys complete – ready for inclusion in DocSite platform
- DSD surveys drafted
- Urogenital surveys drafted

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## Opportunity – Region 4


NIH Grant

- University of Minnesota with Region 4 as a collaborating partner

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## Medical Home Education Workgroup



Sharon Fleischfresser, MD, MPH  
Wisconsin CYSHCN Medical Director

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## Our Goal

- Our goal is to educate primary care providers, specialists and families about the importance of providing a medical home for children with heritable conditions
- We strive to educate parents about the potential benefits available to children who receive care in the context of a medical home

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## Objective

As part of this education process, the workgroup moved forward to plan and develop an educational and resource guide for families.

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## Guide for Families

- To avoid duplicating existing materials, the workgroup examined materials focused on the importance of providing a medical home using criteria developed by the workgroup
- One resource was chosen to act as a template, "A Guide for Michigan Families: Special Care for Special Kids" and was adapted to include information and state-specific and national resources for children with heritable conditions


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## Where are we now?

- Parents and other key stakeholders have reviewed the guide
- The guide is being finalized to prepare for approval, marketing and distribution
- The guide will be marketed to families and professionals through organizations, departments and medical facilities



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**Family is the Center of the Medical Home**  
*A Guide for Families with Children who have Heritable Conditions*

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## Guide Features


- THE MEDICAL HOME, DEFINED
- HAVING A MEDICAL HOME MEANS
- BENEFITS OF HAVING A MEDICAL HOME
- WHAT IS FAMILY-CENTERED CARE?
- CORE PRINCIPLES OF FAMILY-CENTERED CARE
- WHAT IS COORDINATED CARE?
- WHAT IS MEDICAL TRANSITION?
- WHAT IS COMPASSIONATE CARE?
- WHAT IS CULTURALLY EFFECTIVE CARE?
- GENETICS OVERVIEW
- CONDITION SPECIFIC RESOURCES
- STATE RESOURCES
- NATIONAL RESOURCES

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## How to Use the Guide

- This guide aims to share with families and caregivers of children who have a heritable condition(s), the experiences of other families with similar situations.
- This guide provides a wealth of information, checklists, templates and helpful tips to make navigating the health care system easier for families and children.
- Below are two icons you will find throughout the guide:

 **Tip!** The Blue Arrow icon indicates a tip or suggestion for you and your family to consider for a variety of situations, such as working with doctors, planning for life events and other potential situations.

 The Orange Circle with Pen and Paper icon designates a page that includes a tool or list of questions for you and your family to think about, fill out and possibly share with your doctors, other family members or for you to reflect on.

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## Quotes from Families

*“Because we always have Amanda’s care plan and medical records with us the specialist was able to diagnose the problem and give us a game plan right then and there, otherwise it would have taken days! This is just one example of what having a “Medical Home” has done for Amanda and us as a Family!!”*

*Marion (Amanda’s mom)*

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## Parent-to-Parent Tips

- One thing parents always recommend to other parents of children with heritable conditions is to stay organized! A really great tool for organizing care is an individual care plan.
- A Care Plan lists: Health problems; tests or procedures; health care providers; health care services; equipment; treatment plan and expected outcomes

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It's QUESTION TIME!!



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## Care Coordination Workgroup


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## Goal

Promote care coordination for children with heritable disorders


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## Outcomes

- Increase access to care plans that are specific to heritable disorders
- Improve communication among families, specialists, and primary care providers
- Improve the quality of care for children with heritable disorders
- Increase access to emergency care plans for children with heritable disorders
- Increase the number of children with heritable disorders who have an accessible care plan


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## Membership

- Two representatives from each Region 4 state
  - One professional (genetic specialists, genetic counselors, primary care, medical home, state follow-up)
  - One parent/family member


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## Care Planning Issues

- Discussion ~ what care plans should include
- Audiences/users of plan
- Who responsible for
  - Insuring correct information
  - Updating information
  - Determining access to information
- Input from audiences/users regarding content and format

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## Process

- Literature search for existing care plans, systems of care coordination, emergency plans
- Requests to Region 4 participants – share care plans and emergency plans
- Development and use of an evaluation tool to assess existing plans
- Input from stakeholder groups – primary care, ER, genetic specialists, family members

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## Subcommittees

- Established to focus on specific areas of care coordination
  - Emergency Care Plan Subcommittee
  - Medical Care Coordination Subcommittee
  - Care Plan Subcommittee

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## Emergency Care Plan Subcommittee

- Reviewed and evaluated Emergency Care Plans identified through literature review, websites, and Region 4 sources
- Developed and implemented family member stakeholder survey (9/2008)
- Formal recommendation to Region 4 Advisory Group

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## Emergency Care Plan Recommendation

MEMSCIS (Midwest Emergency Services for Children Information System) as *the emergency care plan of choice* for children with heritable disorders *in Region 4*

**Approved!**

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## Emergency Care Plan Subcommittee

Pending activities

- ER staff input
  - Survey developed
  - implementation pending
- Online survey for other stakeholders – primary care, specialists
- Incorporating tool into Family is the Center of the Medical Home....guide

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## Medical Care Coordination Subcommittee

- Collected and reviewed information and plans – literature review, web search, Region 4 samples
- Finalized draft plan for stakeholder input
- Developed and implemented tool for reviewing and assessing draft medical care coordination plan
  - Family members
  - MHWG, CCWG

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## Medical Care Coordination Subcommittee

Pending Activities

- Stakeholder input – May 1 Genetic Experts and Primary Care meeting
- Include tool in Family is the Center of the Medical Home....guide (MHWG)


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### Care Plan Subcommittee

- Conducted literature review, web search
- Gathered and reviewed plans for broader care coordination “all life domains”
- Parent/family member input – what is important/helpful to include
- Developed and implemented tool for assessing components of care plans
- HRSA Site Visit and Performance Improvement Plan (HRSA PIP)

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### Care Plan Subcommittee

Pending Activities

- Identify and provide variety of samples for each component identified (HRSA PIP)
- Develop web-based tool to build family-specific care plan with menu of components
- Include tool in Family is the Center of the Medical Home....guide (MHWG)

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**An integrated approach to emergency care planning for patients with heritable conditions**

Sally J Hiner  
Region 4 Genetics Coordinator

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**Emergency planning: A national priority for CSHCN**

- CSHCN – unique concerns in an emergency
- Rare, complex conditions with multisystem involvement
- Paper charts – unavailable, voluminous, outdated, incomplete
- Electronic charts – not designed for emergency use
- Improved communication methods with Medical Home are needed

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**Emergency Care Planning**

- Approximately 730,000 births/year  
~ 250 new babies with special health care needs due to IBEM each year
- Opportunity for multistate-multicenter collaborations to better define, better implement coordinated plans of emergency care for patients with rare and complex conditions

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**Goal**

Expanding use of the MEMSCIS system to children with heritable conditions requiring emergency plans in a large geographic area (Region 4) will test the robustness and flexibility of the system with a large number of patients and a variety of health concerns

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**Emergency care planning**


Continuum:

- Organized strategies, widely accepted protocols for some conditions (SCD)
- No widely accepted consensus on others (many IBEM, CAH)

Most care currently based on clinical experience/ family experiences (IBEM with expansion of NBS by MS/MS)

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**R4 Care Coordination Workgroup**

- Reviewed literature and center-specific and other existing plans for emergency care planning
- Identified essential elements that should be done uniformly (all in line with MEMSCIS EIF)
- Plans made to gather input from key stakeholders regarding emergency care plan needs and implementation for patients with heritable conditions in Region 4

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## MEMSCIS in Region 4



- Available to all 7 states in Region 4
- NO COST to R4 Center or patient/family
- Appropriate for ANY patient with special health care needs who requires an emergency plan
- MEMSCIS Liaison coordinates with each R4 center
- Easily tailored to center specific needs (templates, permissions)

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## MEMSCIS implementation and planning

- **Illinois** –planning & developing protocols for Sickle cell disease emergency information form
- **Indiana** – planning IBEM implementation
- Kentucky
- Michigan
- **Minnesota** – implementation with IBEM, CAH, other
- Ohio
- Wisconsin

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## The EIF



**Created by the American Academy of Pediatrics (AAP) & the American College of Emergency Physicians (ACEP)**

**Blank form available anywhere in the world via internet access (not tied to any one medical system)**

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## MEMSCIS Components

- Data
  - Ease of entry from point of service (home, clinic, ED, traveling)
  - Real-time editing, management and access
- HIPAA compliant
  - Secure, web-based with option of paper and pdf versions
  - Varying levels of access (controlled by patient/family)
    - Patient/Parent/Guardian; Medical Home; Specialty Providers; Emergency Services (911, ED); Other (dietitian, pharmacy, inpatient teams, home care, care coordinator,...)
- Family materials – English, Spanish, Hmong
- Disaster Preparedness - Reporting functions
  - patients by geographic area or resource needs in event of disaster

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## Broad Utility

- Host uses web-based technology not tied to any one particular health care system
  - Internet access in the home isn't required
  - English as primary language not required
  - Receive specialty or emergency care across geographic borders

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## MEMSCIS Features






Patient/Family Controls Data  
Click button for "Emergency Access"

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## EIF Data Features

 Updated by Parent  
 Updated by Medical Staff  
 Updated by Administration

Date/time/user marked authentication system fully implemented

Data stored and backed up in 2 different cities

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## Accomplishments

- Total patients enrolled (9/2008) = 233
- Collaborating with
  - UIC to expand MEMSCIS access for patients with SCD in Illinois
  - Riley Children’s Hospital, IN for IBEM patients
- Large scale expansion May 31 or SOONER (pending completion of web programming)

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## Summary

- No need to reinvent the wheel: MEMSCIS utilizes the emergency information forms (EIF) have been developed after thorough study by AAP, ACEP.
- A flexible, child/disease specific detailed yet user-friendly web-based communication tool is AVAILABLE for patients and their providers (medical home, specialists, emergency care, etc)
- The price is right: Professional and IT support, coordination of MEMSCIS related research initiatives and materials available at no cost to families or Region 4 participating centers as part of our Region 4 base funding

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## Expansion communication strategies

- UMN: letter and brochure sent to all MN Emergency Departments, ED providers
- Presentations
- Family Video
- Parent User Guide and Program Information Brochures
- Letter & Brochure from NBS/IBEM Coordinator to each new Medical Home Provider when a new patient is enrolled (UMN, other P2 clinics)
- Region 4 website [www.region4genetics.org](http://www.region4genetics.org)

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## Acknowledgements

<p><b>Cynthia Cameron</b> Region 4 Collaborative Director</p> <p><b>Sally Hiner</b> Region 4 Collaborative Coordinator</p> <p><b>Sarah Wedepohl</b> Region 4 Parent Coordinator</p> <p><b>Lee Pyles and Susan Berry</b> Principal Investigators</p> <p><b>Jehad Adwan</b> Region 4 MEMSCIS Liaison</p>	<p><b>Kathy Wood</b> Riley Children’s Hospital, Indianapolis</p> <p><b>ImageTrend® staff</b></p> <p><b>Kristi Bentler</b> MN NBS Coordinator</p> <p><b>Richard Labotka</b> University of Illinois Chicago</p> <p style="color: red;"><b>Region 4 Genetics Collaborative Care Coordination Workgroup</b></p>
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## See for yourself


Take the MEMSCIS Tour

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


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Genetic Expertise Workgroup  
Region 4 Genetics Collaborative

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- The Genetic Expertise Workgroup includes two representatives from each state
- The goals and focus of the workgroup are:
  - To promote links between genetic specialists, rural health service and communication systems
  - Explore practice models to assist the limited number of genetic specialists in maximizing use of their time

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What we have worked on to meet our goals...

- Attempting to map the prevalence of genetic conditions in our region's seven states
- Locating genetic counselors and specialists in each state to determine whether they are near rural areas
- Reviewing literature and other regions' efforts to look at access to genetic services issues

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What we are working on now...

- Partnering with Michigan's Children's Special Health Care Services (CSHCS) Program and surveying families in Michigan about access to genetic services
- Actively planning with Ohio and Illinois to survey families in their states using the same methodology and survey as Michigan's
- Conducting a facilitated discussion in May between genetic experts and primary care physicians in a face-to-face meeting
- Exploring existing telemedicine efforts in the genetics community

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Access to Clinical Genetic Services:  
A View from Michigan's Children's Special  
Health Care Services' (CSHCS) Families

Michigan Department of Community Health  
Genomics and Genetic Disorders Section  
Janice Bach, Joan Ehrhardt, Carrie Langbo  
and Nancy Deising




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Genetic Services Utilization

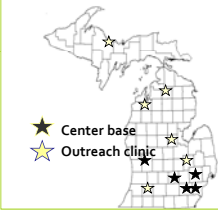
- The Michigan Department of Community Health (MDCH) sponsors clinical genetic outreach clinics throughout the state
- Clinic reports show client numbers in certain areas are dropping

Q: Why? and Do we know what the use of genetic services in Michigan should be?



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Michigan's Clinical Genetic Service Centers




★ Center base  
★ Outreach clinic

FY07-08 (first half)

- 762 (84%) seen in Clinical Genetics Centers
- 147 (16%) seen in Outreach Clinics

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Q: Are there regional differences?



County Name	MBDR Reports 2001-2003	Genetic Counselor Reports 2001-2003	Percent Seen by Genetic Counselors
Michigan	30169	694	2.3
Bay	275	5	1.8
Genesee	890	37	4.2
Ingham	1029	30	2.9
Kalamazoo	726	16	2.2
Kent	1787	72	4.0
Saginaw	689	5	0.7
Wayne	8833	143	1.6

County Level  
FY07-08 (first half)


- In 33/83 counties (~40%) 5 or more families seen
- In 17/83 counties (~20%) 10 or more families seen

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Access to Clinical Genetic Services: A View from Michigan's Children's Special Health Care Services' (CSHCS) Families

Parent survey to identify barriers such as:

- Distance and location
- Time to appointment
- Cost and coverage
- Provider (MH) practice
- Confusion about purpose
- ?




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Access to Clinical Genetic Services: A View from Michigan's Children's Special Health Care Services' (CSHCS) Families


Process:

- Identify CSHCS program enrollees with medical conditions likely to have a strong genetic component
- Invite families to take an anonymous survey
- Evaluate responses to understand needs and barriers
- Report to MDCH programs, Region 4 Genetics Collaborative and Families


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***Thank you!***

*Questions?*


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## Newborn Screening Translational Research Network

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## Background

- October 2008
- \$13.5 million dollar contract awarded to ACMG from NICHD
- Broad mandate:
  - To develop resources and infrastructure that facilitate newborn screening research

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## Goals

- Development of a comprehensive **research infrastructure** in the following areas:
  - Dried blood spot resources
  - Informatics (e.g. databases, data management systems)
  - Patient registries
  - Protocols for patient diagnosis and treatment
  - Laboratory and clinical practice guidelines

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## Goals (cont'd)

- Development of **policies** that underpin the success of such large studies
- Enhancement of **relationships** between the public health community and the diagnostic sector

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## Upcoming Meeting


- April 6-7<sup>th</sup>, Washington, D.C.
- Objective: to review existing state legislative and regulatory policies related to use of NBS dried blood spots and sharing of data related to patients identified by newborn screening.
- Attendees: individuals from state NBS programs, state public health departments, provider community



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### Disaster Preparedness Genetic/Metabolic Patients

- Difficulty accessing treatment under normal conditions
- May have little or no effective treatments available
- May have trouble finding a pharmacy that will stock the drug they need
- May have trouble finding a knowledgeable physician



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### Possible Regional Solutions

- Regionalization – sharing
- Agreements for lab help
- Pantry or stockpile of needed foods, formulas, medications or agreements with pharmaceutical providers
- Telemedicine for consultation to disaster area
- MEMSCIS