



**Region4**  
Genetics Collaborative

## **Region 4 Genetics Collaborative Advisory Group**

November 17, 2008

11 am CST / 12 pm EST

Call in number: 1/866/489-0573; \*4545164\*

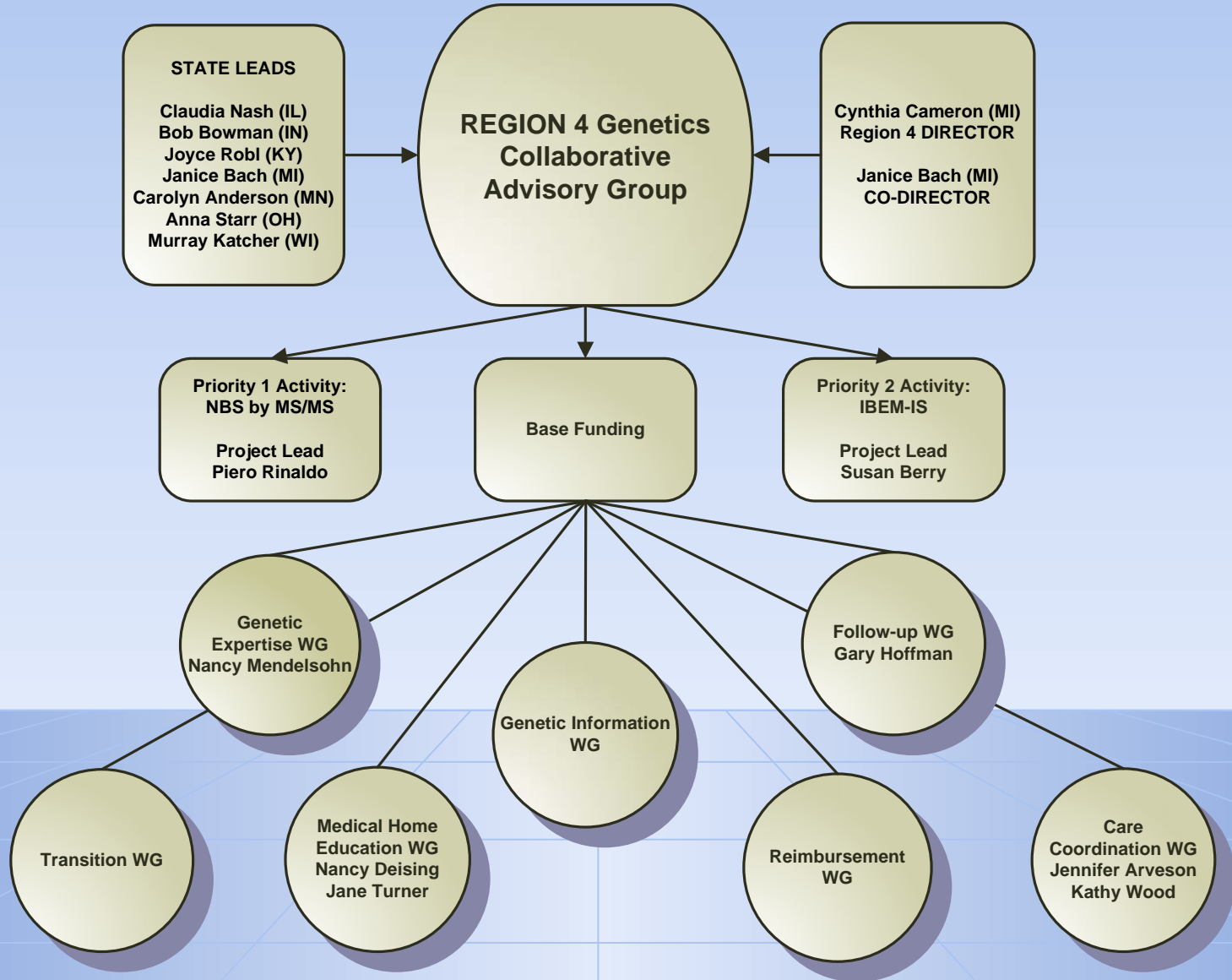
### **Agenda**

- I) Welcome and Introductions (Handout 1) Cameron  
Please give your name, state and role in the R4 Collaborative
  
- II) Updates Cameron
  - A) Region 4 Cameron
    - 1) Regional meeting feedback (Handout 2)
    - 2) Carry forward (Handout 3)
    - 3) HRSA Office of Performance Review Site Visit
  
  - B) Base Funding Hiner
    - 1) Medical Home Education Deising/Turner/Wedepohl
    - 2) Care Coordination (Handout 4 - ACTION REQUIRED) Arveson/Hiner/Wood
    - 3) Endocrine (CAH disease registry) Hiner
    - 4) Genetic Expertise Mendelsohn/Wedepohl
  
  - C) Priority 1 – NBS by MS/MS Rinaldo
  
  - D) Priority 2 – IBEM-IS Anderson/Berry
  
- III) National Connections Berry/Rinaldo
  - A) National Evaluation (Handout 5) Cameron/Wedepohl
  
  - B) National Newborn Screening Translational Research Network
  
- IV) Opportunities Bach
  - A) Collaboration with University of Michigan on regional conference
  - B) Other grant opportunities
  
- V) Reminders Cameron  
Please enter your contact information  
<http://Region4genetics.org>
  
- VI) Other news and issues Advisory Group



# Region4

Genetics Collaborative





## Regional Meeting Feedback

### Combined responses to open-ended questions 1 & 2

1. In your opinion, what were the most useful parts of the meeting (responses in blue)
2. In your opinion, what were the least useful parts of the meeting? (responses in black)

#### **Networking**

- Informal networking
- communication among primary care physicians, specialists & families
- Ability of workgroups to meet in person
- Parent face-to-face meeting
- Regional people together
- Meeting others in person
- Getting people from the states in the region together
- Networking during breaks
- Family meeting time
- general networking time with others
- Networking with others
- Regional collaboration
- Meeting other people with similar concerns
- Meeting people and experiencing the RC in action
- The opportunity to hold 2 impromptu sessions with EHDI folks from other states. The meeting included little info on EHDI, but we were still able to gather and use our time well.
- Interactive conversations
- Information/ideas/networking
- The mix of collaborators is fantastic; the parent and family advocates were fantastic

#### **Updates on Regional Activities**

- Updates on various projects in the region
- Update of MEMSCIS system and the MEMSCIS breakout group
- work group updates
- Program/project overviews
- updating everyone as to what has been going on in the region
- Follow-up on the endocrine project & presentations on CAH
- CAH project summary
- Hearing from each workgroup in the Region 4 regarding an update on what each group is currently working on and what they have done so far
- learning about collaborative
- Hearing subcommittee's reports/updates
- Because it was my first time, I'm glad Dr. Cameron explained the mission of the collaborative again
- Regional project presentations – would have been more useful to read a short abstract/description of the project, decide which interested in, and attend longer discussion section

### Exploring Regional Opportunities

- Exploring regional opportunities
- Possible future collaboration project presentations & breakouts;
- Exploring regional opportunities section was very different from the normal program. It was very very good!
- Research breakout groups
- Research results were interesting, but not as useful
- The breakout session after the research presentations; on Tuesday morning. The purpose/focus was not clear

### Medical Home

- How to establish care coordination for medical home
- The focus on medical home as well as the breakout sessions or possible next projects was useful.
- Care coordination
- Breakout sessions for the medical home – everybody restated the same comments that were already known
- Medical home topics are better addressed to primary care practitioners. This conference should be used for genetic related topics with a section for lab issues.
- Don't understand why the "genetics" collaborative is championing the medical home initiative- - feel we're losing site of the genetics thread throughout all the subcommittee work. I sit on EDHI workgroup and a genetics component seems totally absent to me. While I think it is great to see more "heterogeneity" in workgroup members and conference participants, I feel some of the genetics focus has been lost by going so broad. With increase restrictions on our time and travel at the state public health department level, I feel we're running the risk of losing buy in and involvement of the public health genetics community in the future of this grant.

Medical Home Presentation and Breakout Discussion I now have a better understanding of:	Strongly/ Somewhat Agree	Don't Know/ Neutral	Strongly/ Somewhat Disagree
The National Center for Medical Home Implementation	31	3	2
Specific medical home resources and tools that are available	29	4	3
What I learned in this medical home session is useful to my work/life	26	6	3

### Workgroups

Workgroup breakout meetings; small group discussions

Opportunity to meet with my workgroup

Workgroup sessions

Workgroup meetings; it applied more specifically to my role in the collaborative

Subcommittee meeting

Concurrent Workgroup Sessions	Strongly/ Somewhat Agree	Don't Know/ Neutral	Strongly/ Somewhat Disagree
My workgroup made a lot of progress during our meeting	31	4	1
My workgroup meeting was a good use of my time	32	3	1
I am comfortable with the decisions made in our meeting	33	2	1
I have a good understanding of my workgroup's next steps	30	5	1

**General**

- Depth of knowledge I received
- Breakout sessions
- IRB help
- IBEM training
- I learned some important info regarding metabolic disorders that will assist in making me a better member of my departmental community
- Think this is a great meeting overall
- The caliber of the presenters was generally very good, as were the topics
- General sessions were very broad and not always relevant to my area in endocrine
- I thought all parts were informative and useful
- None
- Zero
- None
- Can't think of anything

**Suggestions****Meeting logistics**

- Great facility!!
- Recommend that meeting be held in a more centrally located place such as Chicago suburbs
- Consider using a travel agent
- It was somewhat difficult to get to Lansing, although the facilities were very nice
- Please notify guests if their room assignment will be changed before arrival

**Meeting organization**

- Keep people on time
  - Need a time keeper – some presenters don't understand the concept of time and amount of information
  - Put a time limit on people- if there were only allowed 15 minutes stop them
  - The AM session on 9/16 was rushed. Microphone problems caused distractions and took away from the presenters' material. Wished there was more time in the meeting for Q/A
- Sessions too long
  - Sitting and listening for 2-2 1/2 hours- would have preferred more hands-on time
- Handouts
  - It would have been nice to get the detailed agenda before the meeting
  - Please put 4 slides to a page on handouts – text on many of slide pages too small to be useful
  - Need PowerPoint on all presenters
  - It would have been nice to have a description of the goals and objectives for each workgroup in this grant cycle included in a handout packet
  - Needed a handout with summaries of what Priority 1, Priority 2, & subcommittees names and purposes with this grant cycle
- Workgroup meetings
  - I think workgroups held Wed. afternoon should not all overlap for those individuals that are in more than 1 workgroup
  - More working group time would have been useful
  - Could have used formal workgroup time on Day 1; some had to leave early so missed designated workgroup time
- Other

- All of the breakout sessions would benefit from more specific goals
- Should have brought us back together for a wrap up to go over when we are meeting next
- You should color code the name tags according to states
- Optional planned evening social time

<b>Overall Meeting</b>	<b>Strongly/ Somewhat Agree</b>	<b>Don't Know/ Neutral</b>	<b>Strongly/ Somewhat Disagree</b>
In general, the meeting was well organized	34	1	1
The two days were a good use of my time	30	1	5
I have a clear understanding of the Collaborative's accomplishments	33	1	2
I had adequate opportunities to express my views during the meeting	31	4	1
I had adequate time to talk with other attendees at the meeting	32	1	3
I learned some new ideas or concepts at the meeting	31	4	0
I will apply some of what I experienced to my work and/or family	33	2	1

### **Quality of presentations**

- Need list of all abbreviations of words
- Too many acronyms
- Michigan endocrine screening evaluations. The speaker used too many abbreviations in slides. The disease should be identified when speaking to a general audience
- I need a cheat sheet for all the alphabet labels. Please don't assume we know them

<b>Presentations</b>	<b>Strongly/ Somewhat Agree</b>	<b>Don't Know/ Neutral</b>	<b>Strongly/ Somewhat Disagree</b>
In general, the presentations were well organized	35		1
The presentation from HRSA was informative	32	3	0
The update from the National Coordinating Center was informative	31	3	1
The presentations about Exploring Regional Opportunities to Support State and National Initiatives were informative	34	1	1
I now have a better understanding of the Collaborative's workgroups	34	1	1

### **Suggested topics for future meetings**

- Discussion of how non-newborn screening genetics fits in collaborative as well as public health genetics (non-newborn screening)
- I feel that short term follow-up of metabolic NBS needs to be reintroduced. This was the only conference where issues were discussed & work was done to standardize what was being done across the region.
- More focus on clinical genetics services in the Region.
- Genetic services access in rural areas; telemedicine
- Integration of EHDI into the general topics
- Coordination & follow-up beyond clinical services
- Medical home definitions in each state and reimbursement related to care coordination
- Regional reimbursement issues for care coordination and other components

- Can we spend any time addressing solutions for the impending clinical geneticist shortage or insurance coverage for genetic services?

### **Additional comments**

- It might have been helpful to meet with people from my state – to hear updates more specific to us
- Is this a good time to have a “state” meeting since we are all together?
- Would like to meet with co-leads and others from state working with
- EHDl workgroup needs a leader who knows better about national EHDl goals and objectives & be connected with the CDC EHDl team.
- States do not necessarily have resources to implement/develop/follow models developed in other states

Administration of the Grant	<b>Strongly/ Somewhat Agree</b>	<b>Don't Know/ Neutral</b>	<b>Strongly/ Somewhat Disagree</b>
I am satisfied with MPHI's administration of the grant so far	<b>34</b>	<b>1</b>	<b>1</b>
MPHI has provided me with the assistance that I need	<b>32</b>	<b>4</b>	<b>0</b>



## Proposed Activities to be supported with Carry Forward

### Base Funding

Carry forward funds from the Base Funding budget resulted from the following:

- Better use of telemeetings and fewer face-to-face meetings led to a reduction in travel costs and cost of honoraria for workgroup leads.
- Fewer cases than anticipated entered into the Midwest Emergency Medical Services Children's Information System
- Funds set aside for product development and distribution were not used because work group products were not yet finalized
- Not all funds were expended by the website developer

### Activities to be supported by carry forward

1. **EHDI Disease Registry** (Short Term Follow-Up page 15 of grant application) – The State of Minnesota EHDI program has contracted with MPHI to work with DocSite to create a disease registry. **Carry forward funds will be used to pay for case entry and for staff to develop contracts/letters of agreement with those entering cases, coordinate training and assist with IRB documents.**
2. **CAH Disease Registry** (Endocrine Project, pages 35-36 of the grant application) – Region 4 endocrinologists are in the process of finalizing data elements for a disease registry which will use the platform created by the Priority 2 Project. **Carry forward funds will be used to support the development of the registry, support state coordinators in recruiting additional endocrinologists, pay for case entry, and for staff time to develop contracts/letters of agreement, coordinate training, negotiate with the software provider and assist with IRB documents.**
3. **Improved Communication with Region 4 States** – (Page 10 of grant application) State leads have indicated a need for additional communication with Region 4 staff (Director, Co-Director, Project Coordinator and Parent Coordinator). In order to respond to this need, Region 4 staff will make site visits to each state. Meeting participants can include current Region 4 members and additional parties who are interested in Region 4 activities. The objectives of the meetings are to (1) provide updates on regional and national activities; (2) disseminate Region 4 products and best practices developed by the Region 4 Collaborative; and (3) recruit additional members. It is anticipated that meetings will begin in the morning and adjourn in the early afternoon. **Carry forward funds will be used to pay for travel expenses for Region 4 staff and to cover breakfast and lunch for meeting participants.**
4. **3 year follow-up of CH** (Endocrine Project, page 9 of grant application) – As part of Region 4 activities, the Michigan Newborn Screening Program has undertaken a limited pilot to determine the feasibility of locating and evaluating the CH diagnosis and treatment status of diagnosed cases after three years. Indiana, Kentucky, Michigan, Minnesota and Wisconsin, have indicated an interest in exploring the feasibility of conducting the evaluations on a

routine basis. The Michigan Department of Community Health has offered to provide 5% of Newborn Screening Manager and Newborn screening epidemiologist (in kind) during this pilot. **Carry forward funds will support up to 5 conference calls among the participating parties and a .10 FTE project coordinator.**

5. **Modified Second-Tier CAH Protocol** (Endocrine project, page 9 of grant application) – Based on previous work done in Michigan, the goal of this project is to develop and publish a two tiered screening algorithm for CAH that reduces the false positive rate by at least 80% and increases the positive predictive value by three fold. Indiana, Wisconsin, and Michigan will participate in this study. Michigan's Department of Community Health will provide project leaders in-kind (2% Newborn Screening Manager and Newborn screening epidemiologist) during this pilot. **Carry forward funds will be used to conduct up to 5 conference calls among the participating parties and a project coordinator at .15 FTE.**
6. **Midwest Emergency Medical Services for Children Information System (MEMSCIS): Sickle Cell Initiative** (Care Coordination Workgroup, pages 33-34 of grant application) – Sickle cell programs in Illinois are already working with Region 4 to adapt MEMSCIS to meet the needs of sickle cell patients. **Carry forward funds will be used to engage additional sickle cell programs in Region 4, hold a face-to-face meeting with sickle cell providers, make necessary revisions to software and support staff to coordinate training.**
7. **Genetic Services Survey** (Access to Genetic Expertise Workgroup, pages 29-32 of the grant application)– The Michigan Birth Defects Registry (MBDR) and the Children's Special Health Care Services (CSHCS) program are preparing to survey families of children with heritable disorders to assess families' knowledge of and perceived need for clinical genetic services. Minnesota and Ohio expressed an interest in joining this effort. **Carry forward funds will be used to (1) support conference calls for participating states; (2) coordinate survey development across states, (3) develop an electronic format; (4) provide participant incentives; and (5) summarize survey results from across the region.**
8. **Marketing and dissemination of Region 4 products and best practice guidelines** (Pages 15, 18 & 20 of grant application) Region 4 has developed multiple products and best practice guidelines that, if marketed and disseminated successfully, can improve practice in both the newborn screening and genetic services arenas. This includes newborn screening short-term follow-up guidelines, the on-line course in newborn screening for prenatal providers, and the soon to be completed Family is the Center of a Medical Home: A Guide for Families with Children with Heritable Conditions, and care coordination plan templates.
9. **Expanded evaluation to measure change in practice** (pages 36-68 of the grant application) – Additional evaluation activities are needed to determine the use and impact of products and best practice guidelines created by the Region 4 Genetics Collaborative. Carry forward funds will be used to complete an evaluation of the following:
  - Online course on newborn screening for prenatal providers – over 500 people have completed the Newborn Screening online course. An email survey will be used to assess how they found out about the course (to inform future marketing efforts) and how they have used that they learned.
  - NBS Short-term Follow-up Guidelines – Telephone interviews will be used to determine how the guidelines have been distributed and used within each Region 4 state.

- MS/MS Training provided by Mayo – A survey will be developed to assess how the experience has impacted practice.
- Evaluation of Priority 2 Activities – This evaluation will be modeled on the previously completed evaluation of Priority 1 and will consist of telephone surveys of workgroup members to assess impact of the project to date.

### Priority 1

Carry forward funds are available from a contract with the State of Kentucky lab for coordinating the sample exchange.

10. **MS/MS Training at Mayo** (pages 6-8 of Priority 1 application)– Week-long training is provided by Mayo at no cost to participants except travel. **Carry forward funds will be used to support at least one person from each Region 4 NBS lab to attend training.**

### Priority 2

Due to delays in finalizing the data platform and processing IRB forms, fewer cases were entered into the information system than anticipated. This resulted in carry forward of funds that had been set aside to support case entry.

11. **Chronic Disease and Family Functioning** (page 6 of the Priority 2 application) - Darin Erikson, PhD, a parent member of the Priority 2 Workgroup, will identify parents of children with inborn errors of metabolism using the IBEM-IS. Parents will be asked to participate in focus groups to identify and assess the impact of stressors on families who have a child with a heritable disorder. Ohio and Michigan have expressed interest in joining this effort. **Carry forward funds will be used to support 1) focus groups in each of the 3 states to provide qualitative data on stressors 2) analyze data obtained 3) develop a survey and 4) staff support to coordinate, schedule and facilitate focus groups. Additional funding will be sought to support implementation of the survey.**



**Region 4 Genetics Collaborative Care Coordination Workgroup  
Recommendation to the Advisory Group  
November 17<sup>th</sup>, 2008**

**RE: Midwest Emergency Medical Services for Children Information System (MEMSCIS)**

**Goal**

Promote care coordination for children with heritable disorders

**Strategies**

- Promote care plans (emergency) for children with heritable disorders
- Implement use of emergency information forms specific to the needs of children with heritable disorders

**Workgroup Activities Supporting This Recommendation**

There are an estimated 250 babies born in Region 4 each year with inborn errors of metabolism ascertained through expanded newborn screening by MS/MS and many more children are born in Region 4 each year with other heritable conditions. Both populations of children face unique concerns in an emergency and would benefit from coordinated plans of care that are created in collaboration with their medical home and specialty health care providers.

The Region 4 Care Coordination Workgroup Emergency Subcommittee engaged in a process to become familiar with best- and promising practices based on findings in relevant literature, identify existing tools, assessing the identified tools, selecting a tool for adaptation to meet the needs of children with heritable disorders in an emergency situation, and assessing the selected tool for adaptability, feasibility, and resources needed.

**A. Identifying and reviewing emergency information forms as clinical tools for improving emergency care for children with heritable disorders.**

Several emergency care plans were reviewed and evaluated using a multi-step process. Emergency care plans reviewed included: Individual care plans currently being used by specialists' with Region 4 who treat children with heritable disorders; emergency plans/protocols developed for families and for professionals and promoted by disorder specific support groups; and emergency protocols from other genetic regional collaboratives.

Initially, the committee members reviewed plans according to a basic review tool developed by the Care Coordination Workgroup that directed the reviewer to comment on plan components (information captured), format, ease of use and strengths/weaknesses. Information was summarized so that the committee could compare components found in individual plans and the format in which the emergency information was provided.

## B. MEMSCIS

After the review process was completed, the subcommittee reached consensus that the MEMSCIS program meets the essential elements of emergency care plans. MEMSCIS is a HIPAA compliant, web-based communication tool that utilizes the Emergency Information Form (EIF) developed and endorsed by the American Academy of Pediatrics and the American College of Emergency Physicians<sup>1</sup>. With permission from the patient/family; the medical home providers, specialty providers, emergency providers, and other parties involved in the child's health care all have input into the content of the EIF and can access that information in an emergency. MEMSCIS is available anywhere in the world where there is internet access, and is not tied to any one particular health care system. The data on the EIF is owned by the patient.<sup>2</sup> Reporting functions enable use by, and identification of patients by, geographic area or resource needs in event of disaster.<sup>3</sup>

The MEMSCIS plan captured components of the plans reviewed and organized them in a logical, easy-to-read format. The subcommittee also compared the components of the MEMSCIS plan to information from the Newborn Screening ACT Sheets to ascertain that the MEMSCIS format was compatible to store information from the ACT sheets.

MEMSCIS, although web-based, includes a printer-friendly option that meets the needs of families who need, or desire, to keep the information available in hard-copy as well as accessible on the web.

## C. Assessing the selected strategy for feasibility, adaptability and resources needed:

The subcommittee then assessed MEMSCIS according to adaptability, feasibility and resources needed to implement

### Feasibility

Previous experience with Region 4 partners demonstrates the feasibility of implementing MEMSCIS. MEMSCIS is fully implemented at the University of Minnesota Children's Hospital, Fairview for use with children who have inborn errors of metabolism, congenital adrenal hyperplasia, and other chronic health conditions. As of September 2008, 233 children with special health care needs requiring emergency care plans were enrolled in MEMSCIS. Of the 233 enrolled, 83 have inborn errors of metabolism, 81 have cardiac conditions, 18 have endocrine conditions (congenital adrenal hyperplasia), and 51 have other chronic health conditions.

### Adaptability

Programming of the MEMSCIS system is flexible and can accommodate different types of services and specialties by means of using specific diagnosis, medication list, and condition-specific common presenting problems and plans of care. Negotiations are in process to expand MEMSCIS availability to the University of Illinois at Chicago for use by patients with sickle cell disease.

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<sup>1</sup> American Academy of Pediatrics Committee on Pediatric Emergency Medicine. Emergency Preparedness for Children with Special Health Care Needs. *Pediatrics*. 1999; 4:104. Available at: <http://www.pediatrics.org/cgi/contents/full/104/4/53e>

<sup>2</sup> Development of a web-based database to manage American College of Emergency Physicians / American Academy of Pediatrics Emergency Information Forms. *ACAD Emergency Med.* March 25, 2005. Vol 12 No 3 Available at: [www.aemj.org](http://www.aemj.org)

<sup>3</sup> American Academy of Pediatrics Committee on Pediatric Emergency Medicine. Emergency Preparedness for Children with Special Health Care Needs. *Pediatrics*. 1999; 4:104. Available at:

Indiana is considering the use of MEMSCIS for their patients with inborn errors of metabolism. A teleconference was held in October 2008 to demonstrate MEMSCIS to researchers at Northwestern University in Illinois who are interested in improving access to vital, disorder specific information at the time of emergency transfer between medical care facilities.

MEMSCIS also is adaptable to meet family needs. Several participants do not have internet access in the home. These users can be given a copy of the EIF printed directly off the MEMSCIS system website. They can keep that copy for their record or when they visit the ED when needed. Other participants have a primary language other than English. A multi-language informational DVD has been made to teach families about MEMSCIS. Languages include: English, Spanish, and Hmong.

### **Resources**

Resources are currently available to support the expansion of MEMSCIS through Region 4 Base Funding. In 2007, the Region 4 Genetics Collaborative agreed to support the expansion of the Minnesota Emergency Medical Services for Children Information System to include all metabolic centers in all seven Region 4 states, becoming the Midwest Emergency Medical Services for Children Information System (MEMSCIS). Later in the grant cycle, MEMSCIS expansion strategies include facilitating interoperability between the web-based server (DocSite) used in the IBEM-IS (Priority 2 activity) and the web-based server (ImageTrend) used in the MEMSCIS project. This will enable the data on emergency encounters for patients with inborn errors of metabolism to be integrated with long-term follow-up data.

### **Recommendation to the Region 4 Genetics Collaborative Advisory Group**

**Support, promote and encourage the use of MEMSCIS throughout Region 4 as a clinical tool for improving emergency care to individuals with heritable disorders.**

To realize this recommendation, the following next steps will be initiated:

- Restructure the MEMSCIS Advisory Board to reflect a broader group of Region 4 stakeholders (disorder specific, role, etc.) to provide input, oversight, and access to expertise as MEMSCIS is expanded to include additional heritable disorders and a broader geographic region.
- Develop an implement a process to respond to inquires about MEMSCIS to engage interested parties in a timely fashion.
- Develop an implement a marketing plan, including recruiting clinics to enroll their patients.

## National Evaluation of the Regional Genetics Collaboratives

### Outcome Measures

Evaluation Domain	Outcome Measure	Definition
A1. Improved care coordination for people with heritable disorders	Increase the percentage of states/territories in the region with collaborations facilitated by the regional collaborative between PCPs and specialty (including genetic) providers to improve care coordination for people with heritable disorders	“Collaborations” may include, but are not limited to: learning collaboratives, work groups, practice models and consortia. A collaboration may be at the state/territory level, e.g. between PCPs and specialty providers within a state/territory, or may be at the regional level, provided the collaboration involves both PCPs and specialty providers from the state/territory being counted.
B1. Improved access to genetic services for people with heritable disorders	Increase in the number of genetic services visits provided to people with or at risk for heritable disorders through distance strategies implemented by the regional collaborative.	“Genetic services visit” is defined as “an encounter between a proband/consultand and a genetics provider.” “Distance strategies” may include, but are not limited to: outreach/satellite clinics, formalized long-distance consultation arrangements, telemedicine approaches.
C1. Development of regional/interregional emergency backup	Increase in the percentage of states/territories in the region that have received current materials or other assistance from the RC on developing back-up systems for NBS and genetic services	
D1. Implementation of expanded NBS	Increase in the percentage of states/territories in the region that have evaluated and made recommendations on implementing the ACHDGNC recommended NBS panel	In order to be counted, states/territories can have evaluated and made recommendations and/or have actually implemented the panel.
E1. Improved follow-up of children identified with heritable disorders through NBS	Increase in the number of NBS follow-up specialty visits provided to families through distance strategies implemented by the regional collaborative	“NBS follow-up specialty visit” is defined broadly. These visits can include short-term and/or long-term follow-up. “Distance strategies” may include, but are not limited to: outreach/satellite clinics, formalized long-distance consultation arrangements, telemedicine approaches
E2. Improved follow-up of children identified with heritable disorders through NBS	Increase in the percentage of states/territories in the region with systems in place to track entry into clinical management for newborns who are diagnosed with condition(s) mandated by their State-sponsored newborn blood spot screening programs	“Entry into clinical management” means that “a health care provider has accepted responsibility for treatment and/or monitoring of the child”
E3. Improved follow-up of children identified with heritable disorders through NBS	Increase in the percentage of states/territories in their region with systems in place to track entry into clinical management for newborns who are diagnosed with hearing loss through their State-sponsored newborn hearing screening programs	“Entry into clinical management” means that “a healthcare provider has accepted responsibility for treatment and/or monitoring of the child”

Evaluation Domain	Outcome Measure	Definition
E4. Improved follow-up of children identified with heritable disorders through NBS	Increase in the percentage of states/territories in the region with systems in place to track receipt of clinical services and/or health outcomes for children who are diagnosed with condition(s) mandated by their State-sponsored newborn blood spot screening program and/or with hearing loss through their State-sponsored newborn hearing screening programs	
F1. Improved/expanded education of PCPs about treatment of people with heritable disorders and about clinical genetic resources in region	Increase in the percentage of state/territories in their region whose NBS programs disseminate “just-in-time/point-of-care” information on specific heritable disorders to PCPs	“Just-in-time/point-of-care information” may include ACT sheets, modified ACT sheets or similar information
G1. Improved regional planning around delivery of genetic services to people with heritable disorders	Completion of regional genetic services plan	Plan should at a minimum, include support for optimal diagnosis, genetic counseling, follow-up, and management of people with heritable disorders
G2. Improved regional planning around delivery of genetic services to people with heritable disorders	Annual review and/or update of regional genetic services plan	Plan should at a minimum, include support for optimal diagnosis, genetic counseling, follow-up, and management of people with heritable disorders