

REGION 4 GENETIC COLLABORATIVE
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Purpose of Project

The provision of optimal genetic services to children with heritable disorders and their families relies on a partnership between public health programs, screening/diagnostic laboratories, clinical providers and families. The Region 4 Genetics Collaborative; which includes Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio and Wisconsin has developed an infrastructure that provides the bridge between public health, primary care providers, genetic specialists and families of children with heritable disorders. These stakeholders work together to identify issues and plan, select and implement strategies that will improve the quality of both newborn screening and genetic services and access to those services.

Over the next three years, Region 4 will build on the existing infrastructure to support regional collaborative activities that improve access to genetic services, expertise and information within the context of the medical home.

Region 4 Genetic Collaborative Mission and Vision

In 2008, the following mission and vision were developed with input from the members of the Region 4 Advisory Group to help guide collaborative activities.

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.

Mission

The mission of the Region 4 Genetics Collaborative is to:

- 1) increase access to information about newborn screening and genetic resources, services and family support systems
- 2) facilitate data collection and analysis to guide decision-making regarding screening cut-offs, diagnosis and long term treatment of heritable disorders
- 3) support state public health agencies in improving infrastructure for genetic service delivery to children with heritable disorders
- 4) 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
- 5) link Region 4 states with regional and national initiatives for improving the quality of newborn screening and genetic service delivery

Needs Assessment and Rationale

Identification of Regional Priorities

The Region 4 infrastructure developed in the first two years of the project was used to facilitate a collaborative process for the identification of priorities for the current grant period (2007-2012).

In October 2006, 85+ members of the Region 4 Genetics Collaborative and invited guests met to review accomplishments to-date and begin the planning process for this proposal. Three concurrent facilitated planning sessions were held in which members representing each

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stakeholder group participated. Participants of each session were guided in the process of identifying the important goals, ideas, and creative strategies necessary to improve access to genetic services, expertise and information within the context of a medical home. Each session resulted in a Mindmap®, a large visual summary of the facilitated brainstorming process.

Collaborative participants from 6 states participated in conversations that resulted in development of a logic model including assumptions, strategies and outcomes (see Attachment 3) and a draft work plan (see Attachment 4) including the following Regional Priorities:

- development and distribution of best practice follow-up protocols/practice models for population-based identification of children with heritable disorders (Goal 2);
- education of primary care providers, specialists and families regarding the importance of providing a medical home for children with heritable disorders (Goal 3);
- promotion of care coordination for children with heritable disorders (Goal 4);
- addressing reimbursement issues that create barriers to quality care for children with heritable disorders in the context of a medical home (Goal 5);
- providing access to genetic information, resources and disease management guidelines in the context of a medical home (Goal 6);
- facilitating access to genetic expertise for underserved populations (Goal 2 and 6)
- identifying and promoting effective models addressing transition to adult services for youth with heritable disorders (Goal 7);
- continuation of efforts to maximize the effectiveness of newborn screening for congenital hypothyroidism (CH) and congenital adrenal hyperplasia (CAH) by improving detection and management of endocrine cases as part of existing newborn screening and follow-up programs (Endocrine Project);
- exploration of MEMSCIS (Minnesota Emergency Medical Services for Children Information System) for expansion into Region 4; becoming *Midwest* EMSCIS utilizing modern information management technology to make Emergency Information Forms (EIF) available via the internet in a secure, private fashion, improving emergency and disaster preparedness for children with genetic disorders; (Pilot project)
- continuation of the Region 4 NBS by MS/MS project and transition from a regional to a national and international project (Priority Activity 1); and
- implementation of a web-based Inborn Errors of Metabolism Information System (Priority Activity 2)

In September 2008, 100+ members of the Region 4 Genetics Collaborative met in Lansing, Michigan to review progress and propose future activities. Region 4 members were asked to present their ideas for projects that would expand current regional impact on newborn screening and access to genetic services. Eleven projects were identified. Carry forward funds to support these activities were requested and approved. The following activities were added to the workplan for FY ending 05/31/2009.

- development and implementation of a long term follow-up disease registry for conditions identified through EHDI in partnership with the State of Minnesota
- development and implementation of a disease registry using the platform created by the Priority 2 project, for long-term follow-up of Congenital Adrenal Hyperplasia (CAH)

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- improvement of communication with Region 4 states through hosting and facilitating stakeholder meetings in each of the seven Region 4 states
- expansion of the Michigan 3-year congenital hypothyroidism (CH) follow-up pilot to additional Region 4 states. This project works to determine the feasibility of locating and evaluating the diagnosis and treatment status of diagnosed cases after three years
- development and publication of a two-tiered screening algorithm for Congenital Adrenal Hyperplasia (CAH, based on work done in Michigan, Indiana and Wisconsin)
- expansion of the Midwest Emergency Medical Services for Children Information System (MEMSCIS) to meet the needs of sickle cell patients
- implementation of a genetic services survey to assess families' knowledge of and perceived need for clinical genetic services in partnership with the Michigan Birth Defects Registry, the Children's Special Health Care Services program; and other interested Region 4 states
- marketing and dissemination of Region 4 products and best practice guidelines
- expansion of the evaluation to measure change in practice by determining the use and impact of products and guidelines created by the Region 4 Genetics Collaborative, including:
 - assessment to learn how participants in the Newborn Screening Online Course found out about the course and how they have used what they learned
 - telephone interviews determining how the NBS Short-term Follow-up Guidelines have been distributed and used within each Region 4 state
 - survey of participants assessing how the Mayo provided MS/MS Training experience has impacted practice
 - survey of workgroup members assessing the impact of the project to date
- support for one person from each Region 4 NBS lab to attend training at Mayo
- implementation of discussion groups with parents of children with heritable disorders to identify and assess the impact of stressors on families who have children with heritable disorders

Project Goals and Objectives

The initial workgroup structure provided the opportunity for the development of diverse projects and products. For the current grant cycle, the Advisory Group agreed that each workgroup should address one piece of an overall issue in order to have greater impact in the Region. It was determined that the overall focus for the five year grant cycle will be on ***improving access to genetic services, expertise and information within the context of the medical home***. Project goals and objectives are delineated in the Region 4 Base Funding Logic Model (Attachment 3) and the updated Work Plan (Attachment 4).

Methodology

A detailed discussion of goals and strategies follows. Updates and progress to date on specific goals and strategies are highlighted in bold font. Region 4 methods for accomplishing work are discussed in further detail in the section titled: Collaboration and Coordination.

Goal 1: Facilitate collaboration within Region 4, with national partners and across regional collaboratives

Region 4 has established an effective infrastructure to facilitate collaboration that has resulted in:

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- strengthened communication and collaboration among public health, individuals, families, primary care providers, and genetic medicine and other subspecialty providers
- improved sharing of information across geographic locations and service systems
- diversity of input into Region 4 Genetics goals, activities, products, planning and direction;
- improved sharing of genetic resources
- improved supported participation of all stakeholders, with a special emphasis on adequately supported and effectively prepared parent participants
- improved collaboration across Regions and with national partners

During the past year, Region 4 focused on increasing collaboration with two partner groups: primary care providers and sickle cell programs.

Region 4 applied for, and was awarded, an AAP visiting professorship. Renee Turchi, visiting professor, presented on the Medical Home in two separate sessions at the Regional meeting – one targeted to parents and the other targeted to health care providers. To maximize the impact of this education opportunity, Region 4 states identified primary care providers and recruited them for participation on the September 2008 Regional meeting. Following Dr. Turchi's presentations, Regional Meeting participants took part in facilitated discussion groups to address increasing communication between primary care, genetic specialists and families. Information collected was analyzed and used to inform activities of the following workgroups: care coordination, medical home education, and genetic expertise (Goals 3, 4 & 6). Subsequent to the Regional meeting, the primary care providers were added to current regional activities as workgroup members and enlisted in review of products and best practice guidelines.

Region 4 also engaged in outreach to sickle cell programs. Representatives were invited to participate in a half-day meeting in Chicago to explore expanding the Midwest Emergency Medical Services for Children Information System (MEMSCIS) to meet the needs of sickle cell patients. Data elements specific to sickle cell disease were developed and included in MEMSCIS. Paula Tannabe presented at the Regional Meeting on opportunities for regional partnerships to improve access to genetic services. We continue to work to engage sickle cell providers in Region 4 activities.

The Region 4 staff team is currently finalizing plans to host and facilitate stakeholder meetings in each of the seven Region 4 states. Region 4 partners in each state are assisting with agenda development and participant recruitment. Stakeholder meeting objectives include: providing updates on regional and national activities, disseminating products and best practices developed by the Region 4 Collaborative, providing state updates relevant to improving NBS and access to genetic services, recruiting additional members and identifying opportunities to extend partnership and develop champions for Region 4 efforts within the state.

Strategy 1.1 Solicit input, advice and direction from regional stakeholders on grant implementation

The Region 4 Advisory Group is instrumental to the success of the Region 4 Genetics Collaborative. Through facilitated Advisory Group meetings conflicts have been resolved, barriers addressed, and support and direction provided to workgroups. The Advisory Group includes workgroup leads and co-leads, the Priority 1 and Priority 2 PIs, and State Leads. The

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Advisory Group meets quarterly to provide input, advice and guidance on barriers to grant implementation, workgroup products and overall grant direction.

Strategy 1.2 Establish or expand workgroups

The following workgroups have been established and are expected to continue their work in grant Year 3:

- Medical Home Education
- Care Coordination
- Access to Genetic Expertise
- MEMSCIS
- Follow-up
- Endocrine

In establishing new workgroups, we are mindful of including multiple stakeholder groups within each workgroup. For example, the Medical Home Education Workgroup includes family members, primary care providers, genetic counselors and genetic specialists.

Strategy 1.3 Facilitate ongoing communication among regional partners

Ongoing communication will occur through regularly scheduled telemeetings of the Advisory Group and each workgroup. The Region 4 website has been adapted to align with the new workgroup structure and Region 4 Genetics Collaborative goals. Postings to the Region 4 website include: meeting summaries, notes, updates, member lists, working group products, presentations, resources and publications. The member directory function of the Region 4 website allows for identifying targeted groups for increased communication between collaborative members via email. The sender selects a group or group of stakeholders and composes an email. The email is sent to the Region 4 administrator who reviews and releases the email. This measure is to protect members from spam or inappropriate emails. A newsletter for parent partners will be developed quarterly. The first issue was released in March 2008. It was sent to Region 4 parent partners and state leads and posted to the Region 4 website.

A regional face-to-face meeting will be convened April 12 – 13, 2010. The Regional Meeting provides an opportunity for interactions across workgroups including sharing updates, products, and resources; sharing expertise and state-of-the-art developments in Region 4 partner states, updating the Region 4 needs assessment and revising workgroup action plans.

Strategy 1.4 Support parent participation in collaborative activities

Although Region 4 provided stipends to parents to participate in the Advisory Group, workgroup activities and regional meetings, the level of parent participation in the previous funding cycle was not optimal. The following activities are now being implemented to support active participation of parents/family members.

The parent perspective is particularly important to the success of the Medical Home Education and Care Coordination workgroups. Parent co-chairs provide leadership for these working groups along with genetics professional. Both the parent and professional co-chairs are provided honorariums on a per-meeting basis. Parents also participate as active members of the Priority 2

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and MEMSCIS workgroups. Parent partners participating as workgroup members and are offered honoraria on a per-meeting basis.

The parent coordinator will continue to assist State Leads to identify family members to participate in Region 4 Workgroups and to provide ongoing support to the family members to help them prepare for meetings; facilitate parent input on products, processes and recommendations resulting from workgroup activities; assist parents in bringing issues to the attention of the Region 4 administration, Advisory Group and workgroups; and work with parents to plan portions of the Regional meetings.

Region 4 parent representatives will be invited to participate in a face-to-face meeting one day prior to the Regional Meeting to be held in April, 2010. Parents will have an opportunity to network with other parents, to identify family needs that can be addressed by Region 4, and to review Region 4 products that would benefit from parent input.

Strategy 1.5 Facilitate collaboration across regions.

As all regions continue to participate in structured events hosted by the NCC, communication between Regions is flourishing. Collaboration across RCs has continued to grow as we learn about each others strengths and needs. An email network provides Project Managers with the opportunity to request information and advice. Most recently, Region 4 requested information about surveys to identify the needs of primary care providers for access to genetic information and expertise and NYMAC requested a telephone conference with Region 4 to learn more about the format and content of our regional meetings.

Region 4 staff will continue work with the NCC to share information with other regions; invite other regions to participate in Region 4 activities, share information and expertise; and collaborate with other regions on national projects. The Region 4 website will be promoted as a communication tool and resource regarding Region 4 activities.

Evidence of collaboration between Region 4 and other Regional Collaboratives (RCs) includes:

- active participation of 44 states in the Region 4 Priority 1 Project
- financial support of 5 of the 6 RCs in the Region 4 Priority 1 Project
- participation of clinicians from Heartland Region in the Region 4 Priority 2 Project
- interest of Region 3 in the Midwest Emergency Medical System for Children Information System (MEMSCIS)
- Mountain States and Region 4 are currently working on bringing together two of their software providers to address interoperability of the public health-focused and the clinician-focused long term follow-up systems for inborn errors of metabolism.

Goal 2: Develop and distribute best practice follow-up protocols/practice models

A Follow-up Workgroup (FWG) has been established that expands the work of the Region 4 Short Term Follow-Up Workgroup established in the first funding cycle of the project (primarily representing state health departments and labs to address NBS follow-up).

Early in the second year of the current grant cycle, membership was expanded to include representatives from Early Hearing Detection and Intervention (EHDI) programs. The focus of

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the Follow-up Workgroup will change as the Early Hearing Detection members finish their work and Region 4 examines models to identify children for short-term follow-up through birth defects registries. Over the remaining three years, membership will include representatives from Birth Defects Registries, Early Intervention (Part C) Programs and Children with Special Health Care Needs (CSHCN) programs. The FWG will solicit input from primary care providers and specialists on revision and dissemination of materials.

The Follow-Up Workgroup will address:

- improving access to genetic services for children with heritable disorders identified by services systems including EHDI, Birth Defects Registries, Early Intervention and CSHCN programs
- development of follow-up protocols and practices for providing information to primary care providers and families at the time of identification (positive screen or report)
- improving communication practices between those identifying children with a heritable disorder and the primary care physician
- sharing follow-up protocols with Region 4 partners, other Regional Collaboratives and national audiences through presentations, web postings and publications

The following strategies will be employed to accomplish Goal 2:

Strategy 2.1 Conclude efforts of the Short Term NBS Workgroup

This strategy was completed. The following products developed by the NBS Short Term Follow-up Work Group were distributed to Regional PIs and are available on the Region 4 website:

- position statement on hospital-based NBS coordinator
- cover letter to accompany position statement on hospital-based NBS coordinator
- NICU protocols
- compiled specimen storage protocols/practices in Region 4 states
- compiled strategies for follow-up with/identifying the PCP

Strategy 2.3 Expand Follow-up Workgroup to include representatives of EHDI, Birth Defect Registries Early Intervention, and CSHCN

Currently the Follow-up Workgroup is addressing increasing access to services for genetic services for children identified through EHDI programs. Workgroup members represent EHDI programs and state NBS follow-up. It is anticipated the workgroup activities will be completed by December 2009. The Birth Defects Registries (BDR) Follow-up Workgroup is scheduled to begin in January 2010.

Strategy 2.4 Develop and disseminate follow-up protocols/practice models

The FWG will identify protocols and practice models for providing information at the time of identification by positive screen or report. The FWG will adapt and disseminate follow-up models, protocols and templates throughout Region 4. Dissemination materials and processes will include recommendations for using the protocols. All products will be posted on the Region 4 Website and will be made available to our national partners. A more detailed description of workgroup activities is included in Objective 2, Attachment 4: Work plan table.

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The EHDI Follow-up Workgroup has developed data elements for regional long-term follow-up; identified and prioritized EHDI issues; identified promising practice strategies to impact priority issues; assessed strategies for recommendation for implementation; reviewed state EHDI follow-up protocols for genetics referral processes; and developed a Border Baby Protocol for follow-up. The group is working to develop model guidelines for EHDI follow-up that include processes and strategies to improve EHDI program success in identifying and referring children and their families for Genetic Services.

The Endocrine Project has collected follow-up models, protocols and practices. The compiled summary comparing and contrasting protocols received across the region is being used to assess and formulate recommendations for improving short term follow-up of endocrine disorders. Tools and processes for locating and evaluating the diagnosis and treatment status of diagnosed CH cases after three years have been developed through a pilot project in Michigan. The tools and processes are being disseminated for implementation in the other six Region 4 states.

Goal 3: Educate primary care providers, specialists, and families about the importance of providing a medical home for children with heritable disorders

The Medical Home Education Workgroup (MHWG) began meeting in December 2007. The group is co-chaired by a Primary Care Provider and a family member representative. Members representing all seven states in the region include primary care providers, genetic specialists, genetic counselors, and family members. The group convenes by telemeeting monthly.

The Medical Home Education Workgroup will address:

- adaptation of medical home materials to address the unique needs of children with heritable disorders
- sharing of materials addressing the medical home for children with heritable disorders with Region 4 partners, other regional collaboratives and national audiences through web postings, presentations, and publications
- providing consultation and education opportunities to primary care providers, family members and care coordinators and specialists
- sharing information about pre-service initiatives in Region 4 to educate medical residents and nursing students about the medical home model

Strategy 3.1: Revise medical home educational materials to address the needs of children with heritable disorders

The Medical Home Education Work Group has held monthly telemeetings since October of 2007. Educational materials developed for health professionals and families were collected; and medical home education activities summarized and reviewed by state. Since few activities were identified to educate families, the group selected educating families about the medical home model its priority. A checklist for reviewing the quality, user-friendliness and content of materials was developed, based on the Toolkit available on the Genetic Alliance website and a review of effectiveness data.

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Based on review of the materials to educate families about the medical home identified by the group, A Guide for Michigan Families: Special Care for Special Kids was selected to be adapted as a Region 4 resource. The workgroup focused on modifying the content to develop a guide to assist families of children with heritable disorders in advocating for their child to be served in the context of a Medical Home. National and state specific resources for each of Region 4's seven states were added. The adapted, comprehensive guide for families is titled "Family is the Center of the Medical Home: A Guide for Families with Children who have Heritable Conditions". The guide has been reviewed by key stakeholders, including parent and family members, genetic specialists, genetic counselors and primary care physicians.

Strategy 3.2: Provide educational opportunities in each Region 4 state

During the fall of 2008, Region 4 participated in a HRSA site review and development of a performance improvement plan. The need for a marketing plan was identified as a high priority to assist Region 4 in reaching a broader audience with Region 4 developed products, information and resources.

Included in this strategy initially was education of two primary stakeholder groups about the medical home – primary care providers and family members. Because the American Academy of Pediatrics has a national effort focused on education primary care providers about the importance of, and strategies for implementing, the Medical Home in their practices; Region 4 will focus on education of families because no other education efforts for families were identified in any of the Region 4 states. The group believes that if parents become more knowledgeable about the medical home concept, they can advocate for services to be provided to their children and family in the context of the medical home. The Guide includes a section on "How to Advocate."

A marketing expert, Kathy Beal from ACMG, has been contacted and agreed to provide her expertise to development of the Region 4 marketing plan. Ms. Beal's services will be supported with resources available through the HRSA Site Visit Performance Improvement Plan.

The Region 4 marketing plan for the medical home guide includes identifying contacts in each relevant organization – national or serving one or more of the seven Region 4 states national organization to assist Region 4 in identifying distribution strategies and opportunities. The guide will be provided to the contact of each identified organization. A follow-up web-cast will be scheduled to "roll out" the guide and identify opportunities for making this educational tool available throughout the Region. Region 4 also will solicit input from the other regional collaboratives to learn how they market materials to families.

Region 4 will establish connections with the state AAP Chapters and service systems (NBS, CSHCN, EHDI, Part C) and professional organizations for purposes of making the family guide available.

Strategy 3.3 Explore educating medical residents and nursing students about the medical home model

Region 4 staff will review current initiatives aimed at educating residents specializing in genetics about medical home, using effectiveness data and criteria developed by the group. Summarized

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findings will be shared with Region 4 partners in order to promote understanding regarding the value of educating medical residents and nursing students about medical home, and the potential for spreading best practice models throughout the region will be evaluated.

Goal 4: Promote care coordination for children with heritable disorders

The first meeting of the Care Coordination Workgroup (CCWG) was held in January 2008. It is co-chaired by a professional and a family member. The membership of the group includes representatives of each state in Region 4 including a pediatric metabolic nurse, a pediatric nurse practitioner, public health representatives, seven family members, and staff from the MEMSCIS project.

The Care Coordination Workgroup addresses:

- communications issues among specialists, primary care providers and families;
- implementation of the Emergency Information Forms in all Region 4 states; and
- integration of care plan elements into the Emergency Information Forms to expand the EIF as the basis for electronic care plans

The workgroup members established three sub-committees to tackle the three components of care plans – emergency, coordination of medical care, and the broader care plan including all life domains. The workgroup convened by monthly telemeeting, and each subcommittee met 1 -3 times between monthly workgroup meetings, as needed. Activities of two of the subcommittees, emergency care plan and medical care coordination, are anticipated to be completed during the current fiscal year.

Strategy 4.1 Promote the use of care plans specific to the needs of children with heritable disorders.

- The Emergency Care Plan subcommittee collected and assessed existing Emergency plans. A recommendation to promote MEMSCIS as the Emergency Care Plan of choice was made to, and adopted by, the Region 4 Advisory Group.
- The Medical Care Coordination (MCC) subcommittee collected and assessed existing medical care plans. A model care plan was drafted and a structured review tool developed. Input was solicited from parents of children with heritable disorders and the Region 4 medical home education workgroup. Review by genetic specialists and primary care providers is scheduled to occur during a face-to-face meeting of these stakeholders in May, 2009.
- The Care Plan subcommittee collected sample care plans. To conduct a thorough review and assessment of the collected plans, the subcommittee developed a care plan assessment tool. This evolved into a tool to help families assess care plans for criteria important and unique to the family. The family can use the tool to help them think about components to include in care plans that are important to the family. Based on feedback from development of the Performance Improvement Plan during the HRSA site visit, the workgroup will use the tool to identify and develop a menu of components that a family could choose from to develop a personalized care plan. The menu options will be both paper-based and web-based.

Once the elements of care plans and care coordination that are unique to children with heritable disorders are established, they will be integrated into existing materials and disseminated along

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with recommendations for use to genetic specialists, primary care providers, and families of children with heritable disorders. As a result of the HRSA site visit, Region 4 established as a performance improvement goal to market and disseminate products and tools. National organizations concerned with genetic conditions will be contacted and invited to participate in a webcast to learn about Region 4 products. Participants will be asked to provide ideas for dissemination through existing networks. Partner sites will be invited to link to the products posted on the Region 4 website.

Strategy 4.2 Create Learning Consortia to identify and resolve barriers to effective communication among families, specialists and primary care providers.

Several states in Region 4 already have primary care providers and other health care providers participating in learning consortia directed at implementing the medical home model. Region 4 will explore partnering with these consortia to serve as a resource for consortia interested in exploring newborn screening and access to genetic services in the context of the learning consortia focus. Representatives from the Minnesota Medical Home Learning Consortia will be participating in the Minnesota Region 4 Stakeholder meeting in May, 2009. They will provide a brief presentation on potential opportunities for Region 4 and genetics in their project.

During the September 2008 Regional Meeting, AAP Visiting Professor Renee Turchi provided an educational session and materials on Medical Home. This session was followed by concurrent breakouts to discuss strategies for improving communication between primary care, genetic specialists and family members. The data were collected and summarized. The CCWG and MHWG are incorporating feedback from these breakouts into tools and guidelines to improve care coordination. For example, the Medical Care Coordination plan includes prompts to include “responsible provider” for specific medical care activities and the medical home guide includes tools to help families think about information they would like their primary care provider to know that may not be prompted for during the medical visit.

Strategy 4.3 Implement the use of web-based Emergency Information Forms (EIF) specific to the needs of children with heritable disorders by expanding the implementation of Minnesota Emergency Medical Services for Children Information System (MEMSCIS) in Region 4

In order to implement region-wide use of emergency information forms for children with heritable disorders, Region 4 has partnered with the Minnesota-based MEMSCIS to become the Midwest MEMSCIS. In addition to including the remaining six Region 4 states, states in other genetics regions have indicated interest. Disorder specific information has been added to allow MEMSCIS to be used effectively with Sickle Cell patients in addition to children with inborn errors of metabolism and congenital adrenal hyperplasia.

MEMSCIS, originating as a research project at the University of Minnesota has transitioned to a clinical tool under the expanded use prompted by the partnership with Region 4. To facilitate utility as a clinical tool, revisions have been identified that include web-based consent and authorization with built in prompts for annual re-consent. Additional software updates pending include enhancing the “break the glass” security option which allows for emergency access without entering a password, revising the immunization record to reflect the recently revised recommendations from the CDC and requiring physician review for data entry into certain fields.

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MEMSCIS, previously under the Care Coordination Workgroup, now has its own workgroup which meets monthly. It is anticipated that the utility and application of MEMSCIS will continue to expand. The workgroup consists of the project PIs, coordinator, Region 4 staff, care coordination workgroup co-lead, parent partners and clinicians. Membership is fluid and time-limited, allowing for the temporary inclusion of key experts in the development of disorder specific data elements. The workgroup is addressing issues including: adapting MEMSCIS to include additional heritable conditions, marketing, development and implementation of a center-based user training plan, interoperability between MEMSCIS and the Inborn Errors of Metabolism Information System (IBEM-IS) and expanding applicability as a tool for emergency and disaster preparedness.

Revisions to MEMSCIS are pending and as soon as they are completed by the vendor, Region 4 will actively promote the availability of MEMSCIS both within and beyond Region 4. Because of interest in states beyond Region 4 in using this tool, Region 4 will establish a national advisory board. The national advisory board will provide oversight, make recommendations to address implementation and marketing issues, identify conditions to be added and key stakeholders to provide input into development of data elements for additional disorders, and respond to requests for use of the data.

For MEMSCIS to be successful as a national project additional resources are needed. Additional staff will be required to perform project coordination functions including: obtaining and disseminating: informational materials for patients enrolled, enrollee cards, and marketing materials; adding clinics to the platform to allow enrollment of clinic patients; providing basic training for new users; triaging and responding to requests for access to MEMSCIS, developing and implementing a center-based user training process and facilitating meetings of the MEMSCIS workgroup and the MEMSCIS national advisory group.

Strategy 4.4 Facilitate interoperability between the web-based server used in the IBEM-IS (Priority 2 activity) and the web-based server used in the MEMSCIS project

In Years 2-5 we will facilitate interoperability between the web-based server used in the IBEM-IS priority project (DocSite®) and the web-based server used in the MEMSCIS project (ImageTrend, Inc®).

Region 4 will bring together the software vendors DocSite (IBEM-IS) and ImageTrend (MEMSCIS) early in the next year to begin work on establishing interoperability between these two data banks.

Strategy 4.5 Revise Emergency Information Forms to facilitate electronic sharing of care plans among primary care specialists, emergency care providers, families

In Year 3, one task of the CCWG at the Regional Meeting will be to identify the data elements that need to be added to the EIF to enable it to include a child's care plan. This will be done in conjunction with the Priority 2 IBEM-IS project to ensure interoperability among all of the Region 4 electronic records projects.

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Region 4 will work with DocSite and ImageTrend to explore options for adding components of care plans which currently fall outside of the parameters of these two platforms (clinical follow-up and emergency medical information). Where possible, care plans, which include information related to all life domains, or care plan components will be available electronically. Establishing interoperability between these tools and the EIF will facilitate coordination of care by making information across life domains easily accessible to the entire care provider team.

Goal 5: Address reimbursement issues that create barriers to quality care for children with heritable disorders

Because collaboration between insurers and professionals on the local and state levels is one of the components of successful care coordination models,¹ the RWG will aim to foster such collaborations. Workgroup membership will reflect Region 4's expanded partnerships and will include representatives of the National Conference of State Legislatures and the American Academy of Pediatrics, along with primary care providers, specialists, genetic counselors, family members, and state CSHCN programs. Health policy advocacy organizations (e.g., ABC for Health Wisconsin), policy makers (e.g., legislative staff, Medicaid directors), and representatives of health plans also will be invited to participate in the RWG. It is expected that the AAP will provide support through the provision of technical assistance provided by a reimbursement coding expert. The NCSL has also agreed to assume a number of critical roles.

The Reimbursement Workgroup will address:

- identification of real and perceived barriers to reimbursement for medical home services;
- education to address *perceived* barriers
- identification of strategies to resolve reimbursement issues
- sharing of information about successful strategies to remove barriers

Although the Reimbursement Work Group was scheduled to have its first meeting in March of 2008, this activity was delayed due to cross-region collaborative activities which we anticipate will provide data to inform the group. These activities include a survey of state programs and clinicians about reimbursement for medical foods and formulas and a parent survey on medical foods and formulas. They will hold their first meeting in March 2008. Once these activities are completed and the results available, the workgroup will be convened.

Strategy 5.1: Identify barriers to reimbursement of services integral to the medical home (e.g. care coordination)

Strategy 5.2: Identify and implement strategies to resolve barriers to reimbursement of care coordination

At the time of the initial proposal, the National Council of State Legislatures (NCSL) agreed to support the Region 4 Genetics Collaborative by identifying and analyzing state and national policies around coverage and reimbursement issues that create barriers to quality care for children with heritable disorders including services provided through the medical home. These activities were to be supported by a contract from the National Coordinating Center to NCSL. Because of staff changes at NCSL, this resource is no longer available. Until this issue is resolved, activities to support these strategies are on hold.

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Strategy 5.3: Identify barriers to reimbursement for medical foods and formulas; identify and distribute successful strategies to resolve barriers

Region 4 has participated in two surveys to collect information on barriers to reimbursement of medical foods and formulas. The first was in collaboration with Region 3 and included telephone interviews one clinician and one follow-up person in each Region 4 state. The second is a survey of two Region 4 metabolic clinics developed by the Long-term Follow-up Subcommittee of the Secretary's Advisory Group. Data from these two surveys will be used to identify barriers within the Region.

Goal 6: Provide access to genetic information, resources and disease management guidelines in the context of a medical home

Goal 7: Facilitate access to genetic expertise for underserved populations

Goals 6 and 7 have been combined: Revised Goal 6: Facilitate access to genetic expertise, resources and disease management guidelines in the context of a medical home with a focus on underserved (rural) populations.

The Genetic Expertise Workgroup convened their first meeting in September 2008 and continues to convene monthly by telemeeting. Membership includes two representatives from each Region 4 state. Dr. Nancy Mendelsohn, MN, serves as the workgroup lead.

Under the new, revised goal, Region 4 has expanded the Genetic Expertise workgroup membership to bring together genetic specialists with primary care providers to determine (1) needs for information and access to genetic expertise of primary care providers; (2) how primary care providers would like to receive the information and be connected to genetic expertise; (3) identify needs of families for access to genetic expertise; (4) develop a workplan to meet these needs.

The expanded Genetic Expertise Workgroup will be convening for face-to-face meeting on May 1, 2009 in Chicago to further develop priorities and next steps. This will include learning about other regional genetic collaboratives' efforts to expand telemedicine to serve children with heritable disorders; reviewing the family survey about access to care; and holding a conversation with pediatricians/primary care providers about their need for genetic information and expertise when providing a medical home for children with heritable conditions. At least two primary care providers with patients in rural areas are being recruited from each of the Region 4 states for the discussion on May 1st. The information gathered at this meeting and through other efforts such as the access survey for families and existing efforts in the area of telemedicine and genetics will be used to fine-tune the activities connected to each of the strategies related to this goal.

In order to better understand the family perspective on access to genetic services, Region 4 is collaborating with Michigan's Children's Special Health Care Services (CSHCS) Program/ Birth Defects Referral and Follow-up Program (BDRFP) to conduct a survey of families of children with heritable conditions. The main goal of the survey is to determine any difficulties in accessing genetic services, especially those families located in rural areas. The survey will be

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distributed to Michigan families first, and then offered for distribution and partnership with each of the other states in Region 4 depending on their interest and available resources.

Strategy 6.1: Promote the use of accurate and up-to-date, disorder-specific resources for primary care providers and families

The GEWG will identify existing disorder-specific resources targeted to both the primary care provider and the family, including an inventory of relevant disease management guidelines. Materials will be collected and criteria identified for assessing the quality of existing materials. Review criteria for materials targeted to families will include “Quality Assessment Criteria for Families” developed by the Community Collaboration and Family Support Working Group in the first funding cycle (2005-07). These criteria include recommendations on how materials should be developed in order to meet family needs.

When available, data will be reviewed on how existing materials have been used by physicians (e.g. NCC survey on use of ACT Sheets) and the effectiveness of the materials. Selected materials will be packaged and marketed to primary care providers serving children with heritable disorders in the context of the medical home. During the second Regional meeting, the GEWG will be brought together with the Follow-up Workgroup to develop strategies for making materials available “just in time” through state partners engaged in population-based identification of children with heritable disorders.

The GEWG will stay abreast of releases of new disorder-specific ACT sheets and replicate the review, adaptation and marketing process as this new information is released by the ACMG. Region 4 will explore partnering with Mountain States RC to utilize the “minimum standard of care” materials they are developing. Marketing and dissemination will include publications, web posting, and presentations and will be included in the marketing plan we will be developing in response to the HRSA Site Visit Performance Improvement Plan with the expert assistance of Kathy Beal.

Strategy 6.2: Develop disorder-specific electronic grand rounds/web casts to address gaps in information (targeted at primary care providers).

Primary care providers treating children with a heritable disorder in the context of the medical home also may benefit from more detailed disorder-specific information focused on disease management and provided by a genetic specialist. This is particularly true for rural providers, who have limited access to genetic specialists. The GEWG will gather input from primary care providers to ascertain interest in receiving genetic information through electronic grand rounds/web-casts. Project activities that will continue based on ascertainment of interest include: review of existing disorder-specific electronic grand rounds/web casts, identification of gaps in specific disorders and disease management; selection of topics for developing electronic grand rounds/web casts, identification of content experts, development of a content outline and selection of a genetic specialist to create and deliver the each presentation. Efforts will be made to collaborate with rural health initiatives in each Region 4 state to provide genetic information to their membership. Continuing medical education units will be pursued.

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Strategy 6.3: Partner with the South East Regional Genetics Group (SERGG) to adapt “Ask the Geneticist” website feature for use in Region 4

SERGG continues to develop this resource, which allows for individualized questions, issues and concerns to be addressed by an individual or panel with heritable disorder(s) expertise. The primary care provider, family or interested individual can submit a question online and an expert response will be provided. This resource also fits the GEWG goal of identifying resources which are specific, immediate and easily accessible. Once this” resource is available for expansion, we stand ready to adapt Region 3’s “Ask the Geneticist” website feature for use in Region 4. “Ask the Geneticist” will be marketed to physicians and families and linked to the Region 4 website,

Strategy 6.4: Promote links between genetic specialists, existing rural initiatives and telemedicine networks

Geneticists and primary care providers representing rural areas have been recruited to participate in the GEWG. The GEWG will work to establish collaborative partnerships between genetic specialists and existing rural health service delivery and communication systems. These formalized links will improve the quality of health care for rural residents and strengthen the local health care infrastructure by making genetic expertise available. The GEWG will examine the existing communication systems, including telemedicine networks and identify opportunities for geneticists to provide expertise to rural providers and patients. Protocols for providing genetic expertise using web-based resources will be developed and distributed through the rural health partners. Legal expertise will be accessed to address legal aspects of telemedicine.

Strategy 6.5: Explore practice models to assist the limited number of genetic specialists in maximizing the effective use of their time

In order to better prepare physicians, the Workgroup will identify, adapt and develop educational and other resources to broaden physician knowledge and understanding of heritable disorders, disease management and best practices regarding the diagnosis and treatment of the child with a heritable disorder in the context of the medical home.

According to Dr. Helga Toriello, Director of Spectrum Health Genetic Services, genetic experts are frequently contacted to address disease management issues that could be handled by the primary care physician. Developing relationships between the PCPs and specialists, increasing PCP access to the specialist, and improving both information available and dissemination of the information will facilitate the transition of primary disease management to the PCP in situations where it is in the best interest of the child. This will lead to more effective use of the genetic specialist resources available in Region 4. Based on feedback obtained during facilitated discussions at the September 2008 Regional Meeting, the CCWG already has begun to include clear delineation of responsibilities in tools for medical care coordination and the care plan. Since this issue crosses the concerns and activities of several workgroups, it will be further explored at the next Regional Meeting (April 2010).

Goal 7 (previously Goal 8): Identify and promote effective models addressing transition to adult services for youth with heritable disorders

In order to help address issues related to improving medical care and disease management for youth with heritable disorders, a Transition Workgroup (TWG) will be established that includes

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specialists, primary care providers, genetic counselors, CSHCN representatives and at least three family members. The TWG will be led by co-chairs, one of whom is a family member.

The Transition Workgroup will address:

- development of modified transition plans that address the unique needs of individuals with heritable disorders
- sharing of transition plans with Region 4 partners, other regional collaboratives and national audiences through presentations, web postings and publications

The Transition Workgroup will hold its first meeting in September 2010.

Strategy 7.1 Promote the use of individual transition plans specific to the needs of children with heritable disorders

Activities of the Transition Workgroup (TWG) will build on work completed during the first funding cycle. The Region 4 Genetics Collaborative has identified and compiled transition models, resources and protocols which have been disseminated to Region 4 collaborative partners and posted on the Region 4 website. The TWG will review these resources for effectiveness data, cost and feasibility of implementation. These resources, tools and protocols will be reviewed for applicability to young adults with heritable disorders. Selected models will be modified to address the transition needs of young adults with specific heritable disorders. The products resulting from this work will be transition protocols for specific heritable disorders that describe best practices for transitioning young adults to adult health care services.

The Transition Workgroup will develop a care plan template to be used with young adults with heritable disorders. The TWG will build on the work of the Care Coordination Workgroup by adding elements specific to young adults to the care plan template recommended by the Care Coordination Workgroup. The product will be a Care Plan Template to be used by the adult-focused physician to create a medical home for young adults with heritable disorders. A marketing plan will be developed that builds on lessons learned from marketing materials developed by the Care Coordination Workgroup

The TWG will disseminate the adapted transition models, processes, protocols and templates throughout Region 4. Dissemination materials and processes will include recommendations for implementation and will target systems who serve children with heritable disorders including: Children's Special Health Care programs, primary care, genetic and other specialty clinic providers. All products will be posted on the Region 4 Website. We will make them available to our national partners such as the AAP, AAFP, ACMG and family advocacy groups for posting if appropriate.

Strategy 7.2 Participate in the National Ad Hoc Transition Workgroup

Sally Hiner, Region 4 Coordinator participates in monthly telemeetings of the national Transition Group. Kathy Stagni (Minnesota), a parent of a 19 year-old daughter who has a heritable disorder was recommended by the Advisory Group to represent Region 4 on this national workgroup.

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Strategy 7.3: Identify strategies to develop a systems approach to promoting transition and distribute to Region 4 partners

Information will be gathered and reviewed by the TWG for applicability in Region 4 transition issues and activities. The TWG will examine systems issues, such as the role of state programs like CSHCN and Genetics, in promoting appropriate transitions to adult care. This will include identifying existing activities in each state with respect to transition and exploring needs specific to heritable disorders. The product will be identified processes for implementing a systemic approach to assuring successful transitions for youth with heritable disorders.

Endocrine Project

This project has brought together, NBS laboratorians, follow-up staff and clinicians to address screening, follow-up and treatment issues of CAH and CH. The group developed two areas of focus: (1) screening and short-term follow-up; and (2) long-term follow-up and treatment.

Screening and short-term follow-up

The goal of this project is 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. Objectives for the project are to: (1) determine case definitions used in each state within the region; (2) describe current screening, follow-up and diagnostic methods; (3) evaluate and compare false positive and negative rates and positive predictive values (PPV), and detection rates; and (4) provide consensus recommendations for improving detection rates and reducing false positive and negative rates in the region.

The Endocrine Project has been successful in obtaining CH and CAH newborn screening data from 5 of the 7 Region 4 states. A database has been developed and analysis by demographic variables has occurred.

1. Since 7/06 (185,000 births) Michigan has sent Mayo blood spots on all positive CAH (.6% of newborns) to determine if there are any false negatives on their 2nd tier steroid profile. Michigan has developed a modified second tier testing algorithm based on the results of this study. The pilot project results and tools will be disseminated to the remaining 6 Region 4 states via telemeeting in March 2009.

Several abstracts have been developed with plans to submit articles for publication:

- Month to Month Variations of TSH and 17-OHP Initial Newborn Screening Test Results from Five States 2005-06
- TSH and 17-OHP Initial Newborn Screening Test Results from Five States 2005-2006 by Demographic and Specimen Card Information
- A ROC curve analysis of the initial TSH newborn screening results from five states to determine more effective cut-off TSH values for referral to specialists' follow-up
- A Project Report of the Michigan Newborn Screening Program: A pilot project to explore additional data sources for identification of false-negative cases of CH and CAH

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2. All newborns on or after 1/1/2003 and treated for CH and in the lowest 25 percentile of serum TSH at time of treatment, regardless of initial DBS TSH, will be followed up at 3-years of age to determine if the child has been re-evaluated for permanent hypothyroidism and if not why not. The plan is to make the 3-year follow-up (serum TSH below a certain percentile with no positive scan at time of treatment) a part of our confirmatory protocol for the diagnosis of CH. A flag for the 3-year follow-up will be entered in the database at the time treatment is started.

The impetus for this project was the Region 4 meeting last May. Pediatric endocrinologists who agreed that CH is probably over diagnosed by most screening programs in the U.S. and it is unknown if children treated for CH but without evidence of permanent hypothyroidism are routinely evaluated at 3-years of age. It was generally agreed that many cases probably are not re-evaluated especially if they are not being followed by a pediatric endocrinologist. Michigan agreed to pilot the project and share findings.

Findings from the Michigan study, including tools for replication will be disseminated during a telemeeting scheduled for March 2009.

Several abstracts have been developed with plans to submit articles for publication:

- A diagnosed Congenital Hypothyroidism cases and their initial screening test values and characteristics
- A Project Report of the Michigan Newborn Screening Program: A pilot project to explore additional data sources for identification of false-negative cases of CH and CAH

Long-term follow-up and treatment

Kyriakie Sarafoglou, MD served as lead Region 4 clinicians in developing a disease registry for CAH. Data elements for both CAH intake and interval clinical visits have been drafted and reviewed by endocrinologists throughout Region 4. Data elements for urogenital disorders and disorders of sex development (DSD) also have been drafted, but are not yet finalized. Additional resources are necessary to continue development of the urogenital and DSD surveys. Carry forward funds have been set aside to have the CAH data elements entered into DocSite. It is anticipated that Dr. Sarafoglou will submit a proposal to NIH to support research using the CAH disease registry. Region 4 has offered to provide ongoing support by contracting with DocSite, scheduling and assisting with trainings on case submission, and providing assistance with IRB applications.

The endocrinologists brought together to work on CH and CAH long-term follow-up issues also identified two additional priorities: follow-up algorithms for children diagnosed with CAH and follow-up algorithms for children diagnosed with Down Syndrome concerning appropriate diagnosis and treatment of CH. Activities during the current fiscal year were supported with carry-forward funding. Additional funding will be needed to support these activities.

Collaboration and Coordination

When the Region 4 Genetics Collaborative Project was first initiated, there was little interchange of information and expertise among the seven states in the region as no forum existed to support these activities.

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Since the initiation of the project, a great deal of progress has been made to promote sharing among states and the maximization of available newborn screening and genetic resources. A collaborative infrastructure to support and sustain activities in Region 4 was developed in 2003 which facilitated the implementation of a regional plan to address needs identified by stakeholders.

Workgroups were developed at the outset of the Collaborative. The process is designed to make good use of the valuable time of workgroup members by providing them with the opportunity to use their expertise and participate in creative problem solving, while MPHI staff provides support. Participants found the workgroup structure to be effective and efficient and the process will continue to be used to support grant activity.

The groups are designed to (1) share information about current practices; (2) identify barriers to improving practices; and (3) develop materials and products to reduce the maldistribution of genetic resources across the Region.

Workgroup members are volunteers. Parent workgroup member participation is supported by a stipend allocated an hourly rate. Workgroup leads and co-leads work with MPHI staff to set the agenda and ensure that the group remains on task. Workgroup leads and co-leads also are offered stipends at a per-workgroup rate. MPHI staff facilitate meetings, prepare and disseminate meeting notes, research issues important to workgroup activities and help prepare workgroup products.

Ongoing Collaboration

Advisory group discussions and collaborative problem solving efforts resulted in the development of several strategies to further strengthen the infrastructure and address limitations (Goal 1). These strategies focus on increasing parent participation, including primary care providers, and increasing collaboration with national and regional partners.

National

- Region 4 Director Cindy Cameron and Co-Director Janice Bach participate in monthly NCC telemeetings.
- Region 4 Director Cindy Cameron and Co-Director Janice Bach participate in quarterly NCC calls
- Region 4 Director Cindy Cameron participates in national RC Evaluation workgroup telemeetings.
- Region 4 Director Cindy Cameron attends meetings of the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children.
- Co-Director Janice Bach represents Region 4 on the NCC Planning Group.
- Region 4 Coordinator, Sally Hiner participates on the national Transition Group.
- Two metabolic clinics in Region 4 are piloting a parent survey on medical foods and formulas developed by the Long-Term Follow-up Subcommittee of the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children.
- Nina Antoniotti, WI continues to represent Region 4 on the Telegenetics workgroup
- Region 4 provides articles as requested to the NCC Collaborator as requested by the NCC

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- Region 4 Project Coordinator, Jodi Griffin, serves as a communications liaison with the National Coordinating Center

Across Regions

- Region 4 Director clinicians and state follow-up staff from each Region 4 participated in the Region 3 Reimbursement for Foods and Formula's Survey
- Clinicians from Region 3 and Heartland have asked for information on how to participate in MEMSCIS

Within Region 4

- Workgroups have been expanded to include two members per state, as it is often difficult for one person working alone within a state to accomplish change.
- Workgroups now include representatives from multiple stakeholder groups. State identification and follow-up system stakeholders have been joined by primary care providers and genetic specialists to promote creativity and encourage dialogue.
- Family member co-leads and family member workgroup participants were identified and a parent coordinator is on staff to encourage active participation.
- Co-leads and participants representing primary care are actively participating in teleconferences.
- The Illinois State Lead, Claudia Nash, has initiated discussions between the Region 4 Director and the Disease Treatment Demonstration Project; Illinois Sickle Cell Action Project, and Sickle Cell Disease and Newborn Screening Program, both funded by HRSA. The Directors of each projects indicated an interest in participating in MEMSCIS.
- Representatives from Region 4 state labs and follow-up programs are telemeeting to learn about replicating the two pilot projects spearheaded in Michigan – CH 3-year follow-up and CAH 2nd tier testing algorithm.
- Region 4 staff are preparing to host and facilitate stakeholder meetings in each of the Region 4 states. Region 4 partners from each state have been instrumental in developing each state stakeholder meeting agenda and in identifying participants.

Other Partners

- Region 4 applied for and received the AAP Visiting Professorship. The visiting professor provided presentations and educational materials on the medical home at the Regional meeting.
- Although not yet defined, we hope that there will be linkages between the Region 4 Priority 1 and Priority 2 projects and the Newborn Translational Research Coordinating Center.
- We are examining Genetic Alliance tools and products for use and/or adaptation in Region 4. Genetic Alliance has offered the opportunity for Region 4 to present during their monthly telemeetings.
- Region 4 will be contacting national organizations concerned with genetic issues to share products and tools developed in the Region.

Administration and Organization (See Attachment 6: Region 4 Organizational Chart)

The master organizational chart for the Region 4 Genetics Collaborative illustrates the infrastructure that will support the work of the Collaborative over the next five years. The

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Michigan Public Health Institute (MPHI) will function as the administrative and procedural hub for activities described in this proposal and serve as a resource as needs are identified through project implementation. MPHI will partner with all seven state public health agencies, geneticists, primary care providers, care coordinators, genetic counselors, families, epidemiologists from within Region 4, and national partners.

Advisory Group

An advisory group including representatives from each of the Region 4 states (State Leads), Chairs and Co-chairs of each newly established Workgroup, 3 family members (these parents will be the co-chairs of the Education, Care Coordination, and Transition Workgroups), and the PI's of the Priority 1 and Priority 2 projects provides program oversight.

Staffing Plan

The roles and responsibilities of each staff member are described below.

Project director (.50 FTE) Cynthia Cameron, Ph.D., provides administrative and collaborative leadership for the project. She is the main contact for the State Leads and the Advisory Group; reviews quarterly progress with project coordinators and works with them to identify and resolve barriers; and identifies and establishes relationships with national partners who can be of assistance in implementing the regional plan. Dr. Cameron initiates and maintains linkages and relationships with the NCC, HRSA, other Regional Collaboratives and national partners.

Co-Director (.10 FTE in-kind) Janice Bach, MS, CGC, State Genetics Coordinator at the Michigan Department of Community Health. Janice draws on her extensive background and experience with genetic counseling, public health genetics and NBS service delivery to assist the project director with content development as well as general program oversight.

Coordinator of Regional Activities (.70 FTE) Sally Hiner staffs monthly teleconferences and scheduled face-to-face meetings for the workgroups and Advisory group, collects and prepares meeting materials, prepares quarterly reports; facilitates communications within, among and across the workgroups, assists in the development of workgroup products. She monitors participation and prompts member recruitment and identification; and monitors compliance with timely submission of quarterly reports by project leads.

Administrative Assistant (.40 FTE) Patricia Losey, administrative assistant for the Systems Reform Program at MPHI processes and audits project reports, records, schedules, budgets and contracts; reconciles expenses to budgets; creates purchase orders, audit invoices and voucher payments; and oversees payroll time entry. For face-to-face meetings, she is responsible for meeting logistics and reimbursing participants for travel expenses.

Project Coordinator (.75 FTE) Jodi Griffin, researches topics of interest for workgroups and assists in development of products, and works with the Region 4 Genetics Collaborative website contractor to develop an up-to-date, user friendly website. She provides technical assistance to MEMSCIS and CAH Registry participants by responding to requests about IRB issues, assisting with passwords and scheduling training. She is also Region 4's communication liaison to the American College of Medical Genetics.

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Parent Coordinator (.50 FTE) Sarah Wedepohl, the parent coordinator has leadership experience and is also a parent of a child with a heritable disorder. She is responsible for recruiting parent participants; supporting parent participation in meetings and other activities; coordinating parent leadership training opportunities; working to ensure the perspective of parents is considered in all decisions that have the potential to affect families; working to ensure parent partners are informed; assisting parent partners in preparing for participation in Region 4 meetings and activities; and coordinating stipends for parent participation and ensuring timely reimbursement of parent partners.

¹ Ibid.