

MSGRCC Quarterly Newsletter
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Exciting and fast. That partially describes the first three months of the new Mountain States Genetics Regional Collaborative Center (MSGRCC) grant. The 2007 Annual Conference was very successful. It was a pleasure to see longtime members and get to know so many new people. The educational topics were excellent, informative and encouraging.

We are pleased to announce that we have a new logo and a new web site is under construction. We will keep you abreast as the web site evolves and will want your input and contributions as we expand.

We all know the importance of evaluation to a project or program. The National Coordinating Center (NCC) is supporting an evaluation workgroup. MSGRCC is responding for the region and has an RFP out to hire an evaluator. This evaluation will enhance sustainability to the MSGRCC.

Thank you to everyone that participated in the challenging process of defending your committee for continuation support. Thoughtful but difficult decisions have been made on the organization and structure of the MSGRCC. Critical to the decisions is the mission of MSGRCC - ***to collaborate for the purpose of ensuring that individuals with heritable disorders and their families have access to quality care and appropriate genetic expertise and information in the context of a medical home.***

Below are the workgroups that will have fiscal and staff support from the MSGRCC:

Consumer Advocacy Workgroup - The consumer workgroup will examine and advise the MSGRCC to ensure consumer focus. At least one consumer representative will represent this workgroup on all other workgroups.

Public Health Workgroup - The public health workgroup will examine and advise the MSGRCC to ensure the use of public health practices. This workgroup is where issues such as emergency preparedness, medical home, telemedicine and family history will be addressed. We will create ad hoc sub-groups within this workgroup to address these topics. At least one public health representative will represent this workgroup on all other workgroups.

Standard and Emergent Technologies Workgroup (previously the Laboratory Practices Committee) - This workgroup will collaborate on initiatives for continuous quality improvement for regional genetics laboratories and education related to clinical laboratory testing of heritable disorders to primary care physicians and families by direct collaboration among participants in the region.

Quality Improvement Workgroup (previously the Clinical Services Committee)
- This workgroups mission is to improve the quality and accessibility of clinical genetics services in the mountain states.

Newborn Screening Committee - The NBS workgroup will continue. Newborn Screening Committee members share expertise and resources between laboratory and follow-up aspects of newborn screening in order to assure quality in biochemical genetics-screening and to promote state-of-the-art follow-up of affected newborns in this region.

All consumer and family members' travel and expenses will be covered. We will ask those who can afford to pay their own way, to do so, thus freeing funds for others who need support.

You will be receiving more information on the workgroups such as meeting schedule and membership. The workgroups, as were the committees, are the heart of the MSGRCC and the site where the work is prepared and completed.

Watch for e-mails about the mid-year meeting. It will be in Austin, Texas on November 3rd and 4th. This is an important planning meeting for the next year of the MSGRCC. Unfortunately, the MSGRCC only has limited funds for members to attend. To compensate, for not being able to fund as many attendees, we will have regular conference calls of the workgroups. We cannot stress how important it is to have everyone's thoughts, ideas and participation.

We are off to a great start. We are recruiting membership from all states and recruiting additional Hispanic, African American and Native American participation.

Thank you to everyone who is supporting MSGRCC. We look forward to working and sharing more with you soon.

Sincerely,

Joyce Hooker
Associate Director
MSGRCC

Camille D. Miller
Director
MSGRCC

A Note from John Johnson

After attending a Washington, DC meeting of the committee described below, I think it would be helpful to present their mandate for existence and their focus, which is very relevant to our Mountain States Regional Genetics Collaborative Center. From the HRSA website, MCH section, the following is stated:

“The Secretary of Health and Human Services (HHS) is directed under section 1111 of the PHS Act to establish an Advisory Committee on Heritable Disorders in Newborns and Children (Committee). HHS has expanded the Committee to include genetic diseases and has renamed the Committee accordingly. The Committee's purpose is to provide to the Secretary advice and recommendations concerning the grants and projects authorized under section 1109 and technical information to develop policies and priorities for this program that will enhance the ability of the State and local health agencies to provide for newborn and child screening, counseling and health care services for newborns and children having or at risk for heritable disorders. Specifically, the Committee shall advise and guide the Secretary regarding the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and programs for effectively reducing morbidity and mortality in newborns and children having or at risk for heritable disorders.”

Our project is funded under section 1109. It is apparent from the above that newborn screening (NBS) is the priority for the funding and the committee. However, they were originally named “ACHDNC”, and now, Genetic Diseases has indeed been added, to create the rather cumbersome ACHDGDNC” acronym. This opens the door for some work on genetic services, which has always been a Mountain States focus. At the same time, we must realize that the government is most interested that this funding results in improved NBS. Implicit in this is that there is universal screening, and that all states screen for the same basic recommended panel, which should currently include the ACMG list of 29 primary conditions detected by tandem mass spectrometry. Accordingly, a major bloc of the recent DC meeting was to develop a process for submission and review for inclusion of conditions not currently on the panel. We have all heard early presentations on NBS for Fragile X syndrome, and there are many other groups advocating for NBS for other disorders.

Given all this, what are we doing and where are we going? Some of the direction we were given encourages us to expand our membership with more diverse groups. Please be on the “lookout” for Hispanic, African American, or Native American/American Indian individuals, professional or lay, who might be interested in membership. We are also encouraged to engage states which are not strongly represented by projects currently funded. These include Montana, New Mexico, Wyoming, Texas, and Nevada. Our funded NBS projects are predominantly in Utah and Colorado. In Utah, Dr. Marzia Pasquale is collecting samples from confirmed patients with disorders detected by MS/MS, Drs. Nicola Longo and Dr. Jeff Botkin are exploring clinical and database issues in development of a long term registry/follow up system for such patients. In Colorado, Dr. Janet Thomas is the project lead and along with Montana, New Mexico, Utah and Texas is working on implementation of standard long term outcome measures and shared data sets. Each of the eight states in the region had metabolic experts involved in the development of the disease specific care plans. All of the projects have done wonderful groundwork for

these projects in our first 3 years. We now have 5 years of funding to bring these projects to fruition.

We also have cultural diversity projects led by Dr. Murray Brilliant, working with the Navajo and Hopi nations in Arizona, and by Dr. Kathy Hassell, Donna Holstein and Shannon Gillette in Colorado for Hispanics and African Americans. Both focus on genetic education and literacy and obstacles to those goals in those populations. We also have developing projects in Emergency Preparedness, a federal goal following the lessons from Katrina with regard to NBS, and in Medical Home, a major sponsored project of HRSA and others. Finally, HRSA and others have also funded many telehealth projects in the nation, and would love to see more activity in genetic services and education provided by this medium.

There is room for other projects, but those are the current priorities for our Collaborative. And by using this latter term, I hope to encourage and promote more interstate activities, and eventually, interregional ones. Please become active in your Committees, to be renamed Workgroups to emphasize the first “four letter” word, and in your states.

John Johnson, MD
Principle Investigator
MSGRCC

Annual Meeting Recap

On July 12-14, 2007, the Mountain States Genetics Regional Collaborative Center (MSGRCC), in collaboration with the Mountain States Genetics Foundation, held its annual conference in Denver, Colorado. This conference was intended to encourage education, planning and implementation preparation. Conference presenters included:

- **John Johnson, M.D.**
- **Laura Pickler, M.D.**
- **Murray Brilliant, Ph.D.**
- **Mike Watson, Ph.D.**
- **Vivian Ota-Wang, Ph.D.**
- **Piero Rinaldo, M.D., Ph.D.**
- **Jeff Botkin, M.D., M.P.H.**
- **Nicola Longo, M.D., Ph.D.**
- **Janet Thomas, M.D.**
- **Kathryn Hassell, M.D.**
- **Marci Sontag, Ph.D.**
- **Jan Lowrey, Ph.D.**
- **Sarah Hartway**

- Marzia Pasquali, Ph.D.
- Robin Temple-Smolkin, Ph.D.
- Marc Williams, M.D.
- Sharon Milligan, R.N.
- Henry Clamen, M.D.
- Marilyn Coors, Ph.D.
- Randi Hagerman, M.D.
- Paul Hagerman, M.D., Ph.D.

The conference evaluations reveal a very successful meeting. There were approximately 150 attendees representing all eight states in the region and at least fifteen disciplines.

During the conference, the MSGRCC convened its committees for strategic planning meetings. Breakout sessions were also designed around topical areas the region will be exploring and addressing. The outcomes of these meetings will provide direction and focus for the current grant cycle. To see the complete agenda and PowerPoint presentations from the conference, go to <http://www.mostgene.org>.

Current Grant

New Purpose

The new purpose of the MSGRCC is to collaborate to ensure that individuals with heritable disorders and their families have access to quality care and appropriate genetic expertise and information in the context of a medical home. Additionally, the MSGRCC also has a new logo and a new website will follow. Details will come soon!

There are several projects funded under the MSGRCC:

New Projects

Laboratory Quality Assurance Project

Project Lead: Dr. Marzia Pasquali

This project will address the increased rate of false positives/false negatives due to failure to recognize patterns of abnormal metabolites and failure to adopt 2nd tier tests. Specifically, **Dr. Pasquali** will:

1. Collect and exchange newborn blood spots from patients with confirmed metabolic disorders and from patients with abnormal metabolites due to iatrogenic effects;
2. Provide an evaluation of results that will include information about the disease, why certain metabolites are abnormally elevated, urgency of follow-up; and

3. Promote the use of second tier testing to reduce the number of false positive (and, in some cases, false negative) results.

Newborn Screening Outcomes Project

Project Lead: Dr. Janet Thomas

Nearly all states throughout the country are moving toward expanded newborn screening. Doing so will result in the diagnosis of many inborn errors of metabolism in asymptomatic newborns that were previously diagnosed in symptomatic infants and children. Early diagnosis is naturally felt to be beneficial and result in better medical and developmental outcomes for the children, however, for many of the disorders detected by expanded newborn screening, this is speculative as there is a current lack of long term follow-up.

The lack of outcome data for presymptomatically or very early diagnosed infants with many inborn errors of metabolism signals the need for this information to be gathered. This requires systematic, consistent, and reliable data collection of as many patients as possible. Given the rarity of these conditions, patient information needs to be gathered from across the region.

The biochemical genetic care providers throughout the Mountain States Region have a long history of strong collaboration and cooperation. By bringing together the regional care providers, systematically reviewing the disorders to be diagnosed via expanded newborn screening, determining appropriate clinical parameters and outcome measures to be collected, and subsequently systematically gathering the determined information, we will be able to prospectively collect the necessary information that will begin to inform on the long term outcome of such disorders and do so with appropriate patient volume.

Socio-Cultural Diversity Project

African American and Hispanic Populations

Project Lead: Dr. Kathryn Hassell

In all states in the Mountain States Region, infants identified with sickle cell disease by newborn screening are contacted by newborn screening follow-up personnel and referred to a regional sickle cell disease expert. However, the extent of life-long care for sickle cell patients in other areas of the region by providers with relevant knowledge about sickle cell disease, especially into adulthood, is unknown.

The target population in the region extends beyond the African American community. According to results of a Colorado Sickle Cell Center project (“Identification of Possible Communities at Risk for Sickle Cell Trait”), funded by the Mountain States Genetics Regional Collaborative Center, 29% of infants born in Colorado with sickle cell trait in 2005 were born to mothers who self-identified to be of Hispanic descent on their birth certificate. Unfortunately, only a limited

amount of patient educational material is available in Spanish, and most Spanish-speaking providers lack even basic knowledge about sickle cell disease. Other cultural groups, including native Africans, seek care in the region. An increasingly diverse population of families potentially at-risk for infants with sickle cell disease may not be aware of the risk due to community assumptions about the distribution of sickle cell trait. Improving the awareness of providers who provide such services that identify a broader group of patients at-risk is a critical step in sickle cell disease prevention. Significant cultural and language barriers can impede access to a competent medical home for these groups as well. Serving each of these target populations by identifying and supporting medical homes and communities in areas where sickle cell patients and their families choose to live and work is of critical importance in the region.

Native American Populations

Project Lead: Dr. Murray Brilliant

At the crossroads of medicine, molecular biology, biochemistry, and all other life sciences, genetics is wide-ranging and inherently interdisciplinary. Genetics provides the fundamentals for the understanding of life, and it has profound implications for human health and economics. Although the general public is aware of the importance of Genetics, there are misunderstandings leading to unjustified fears about genetics research, as well as a few publicized breaches of trust, leading to justified concerns regarding Genetics research. This is especially an issue among Native American populations in the mountain states of Arizona, New Mexico, Montana and Utah.

A general lack of respect and sensitivity to the religious and cultural beliefs of the Native American peoples by some researchers and medical personnel has given the Native American population cause to not trust or believe information given to them by the American health system. Genetics raises its own set of questions and fears as it deals with DNA, a very sensitive issue for Native Americans.

The goal of the project is to increase knowledge about genetics and genetic services with the ultimate aim of improving access to genetics information and services in the region. This will be accomplished through the development and implementation of consumer education and dialogue programs created specifically for and offered in locations accessible to the Native American population.

Genetics Policy and Education

Project Lead: Liza Creel

In the Genetics Policy and Education project, the MSGRCC will focus on systemic issues impacting or affecting the genetic services delivery system. Initial focus areas will be:

- The medical home

- Telemedicine
- Emergency Preparedness
- Family History

The project will educate local and state policy decision makers and other stakeholders (especially primary care providers) on implementation, models of care and relevant solutions affecting the genetic services delivery system in the Mountain States Region.

Policy research, policy development and education provide an opportunity to identify solutions to address individual needs. This process will include policy research to identify models of care, treatment effectiveness and cost-effectiveness of these models and inform policy makers and stakeholders of evidence-based policy solutions as well as best and promising practices. Policy education forums (expected in year two), to have the most impact, must be provided not only to state policy makers but to all stakeholders and decision-makers in local jurisdictions and organizations.

Resources and Interesting Topics

In June 2007, **Camille Miller** and **Joyce Hooker** attended the release of the Commonwealth Fund's report "Closing the Divide: How Medical Homes Promote Equity in Health Care: Results from The Commonwealth Fund 2006 Health Care Quality Survey" at the National Press Club in Washington, DC. For a copy of the report and more information, please visit

http://www.commonwealthfund.org/publications/publications_show.htm?doc_id=506814.

In September, Genetic Alliance launched the new website WikiGenetics, which provides genetics information to consumers and other stakeholders. All genetics professionals are invited to visit the site at: www.wikigenetics.org.

Other Useful Websites:

National Center of Medical Home Initiatives for Children with Special Needs
<http://www.medicalhomeinfo.org/index.html>

National Coordinating Center for the Genetics and Newborn Screening Regional Collaborative Groups
<http://www.nccrcg.org/>

About the MSGRCC

Senior Editor: Camille D Miller
Editors: Delia Presley and Liza Creel

The purpose of the MSGRCC is to collaborate for the purpose of ensuring that individuals with heritable disorders and their families have access to quality care and appropriate genetic expertise and information in the context of a medical home. The Texas Health Institute serves as the grant administrator for the Mountain State Genetics Regional Collaborative Center.

The MSGRCC is one of seven regional collaborative centers covering the nation. It is federally funded by the US Department of Health and Human Services, Health Resources and Services Administration (HRSA) Genetic Services Branch. The Mountain States Genetics Regional Collaborative Center includes the states of Arizona, Colorado, Montana, New Mexico, Nevada, Texas, Utah and Wyoming.

The MSGRCC newsletter is a quarterly publication of the Mountain States Genetics Regional Collaborative Center written to update its readers on grant projects.

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