



Message from the Project Directors Celia I. Kaye, MD, PhD & Kathryn Hassell, MD

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April 2012

Greetings!

April in Colorado this year has been a wonder - sunny, cool and delicious! Trees are blooming, spring flowers are in blossom, and the blue mountain sky is filled with little white clouds and birds returning for the summer. How fortunate we all are to live in these "mountain states"!

You may have noticed that we've dropped a "C" from our name. In line with what other regions have already done, the Mountain States Genetics Regional Collaborative will no longer use the word Center in our name, as to not confuse our programmatic work with the work of other HRSA-funded centers.

ACMG Annual Clinical Genetics Meeting

March ended with the annual meeting of the American College of Medical Genetics (ACMG), held this year in beautiful Charlotte, North Carolina. This meeting marked an important name change for ACMG, now the American College of Medical Genetics and Genomics. While this change may seem of limited importance to some, it is of great importance to those of us who are dedicated to public health applications of genetic and genomic discoveries. The new name emphasizes the interaction of the entire personal genome with environmental factors to cause disease, as well as opportunities to apply preventive strategies to populations. We're pleased to see ACMG rising to the challenge of this new world of genomic medicine.

Liza attended the ACMG meeting to moderate the community conversation on telemedicine, a special satellite session designed to introduce telemedicine to genetics

providers. Clinicians from several regions presented on their experiences using telemedicine to deliver clinical services, and a family from the Southeastern Region talked about how telemedicine improved care for their daughter. MSGRC also collaborated on a poster presented at the ACMG meeting. The poster, titled "Telegenetics Clinics in the United States: Success, Challenges and Future Directions," summarized how telemedicine has been used in the Western States, Heartland, and Mountain States Regions. There were several sessions on applications of next generation sequencing as the cost of personal whole genome sequencing continues to drop. New, less invasive approaches to prenatal diagnosis also received attention. There were interesting presentations on various aspects of newborn screening, emphasizing new laboratory techniques as well as new treatments for disorders already included on the recommended panel. There was also plenty of opportunity to review the hundreds of posters on genetic diagnosis and treatment, view new books and technologies introduced in the past year, and network with old and new friends from all over the world.

MSGRC was fortunate to send two consumer representatives to the ACMG meeting through the HRSA Genetics Collaboratives' Advocate Leaders Partnership Program at the National Coordinating Center. Melanie and Marc Wiseman, members of the MSGRC Consumer Advocacy Workgroup, attended the meeting and participated in the Advocates Program, which engages advocates with NCC and RC leaders, ACMG leadership, and medical genetics professionals. Throughout the conference, the advocates enjoyed mentoring and the personal ambassadorship of genetic counseling graduate students from the University of Arkansas for the Medical Sciences.

"I just want to say that I am very honored that I was able to attend the ACMG conference. As a mother, I have been able to keep up on the research, and most importantly, keep updated on the best care available for my daughter. As a nurse, I have been able to take back the knowledge that I learn and use it in my everyday practice. With all of this information at hand, I have been put in a situation to where I can give back to a community that has given so much to my family. I hope that the Advocate Leaders Partnership Program will continue to grow. With physicians, clinicians, researchers, genetic counselors, nurses and parents involved together, I am certain that great things can come out of it for each and every patient."

~Melanie Wiseman, MSGRC Consumer Advocate

On April 13, Kathy and Celia introduced MSGRC at the 2012 Bi-Regional Hemophilia Conference in Colorado Springs. There are networks of hemophilia treatment centers around the country, and our genetics regional collaborative currently overlaps with three of these hemophilia regions (VII, VIII, IX). The Health Resources and Services Administration (HRSA), which funds both the regional genetics collaboratives and the hemophilia treatment centers, has asked us to work more closely together in the future to assure that individuals with these genetic disorders are receiving the full range of treatment services they require. It was a pleasure to meet and interact with providers who care for children and adults with bleeding disorders, and to share strategies for further cooperation in the future.

Other Project Activities

NBSTRN Information Technology Workgroup

Kathy attended the Information Technology Workgroup meeting of the Newborn Screening Translational Research Network (NBSTRN) this month. She and others, including Marci Sontag from the Mountain States region, provided input gathered from the Clinical Centers and Data Access Workgroups regarding clinical information to be incorporated into the database, which is under development.

Case Definitions Project

April also saw a flurry of activity as metabolic, endocrine, and hematology specialists in our region reviewed a series of draft definitions of the disorders detected by newborn screening. This project, spearheaded by the Genetic Services Branch at HRSA, is meant to come up with definitions that are accepted throughout the country, to standardize reporting at the state level and allow for meaningful comparisons between states. We appreciate the assistance of our outstanding specialists in the mountain states; this project should result in better reporting and therefore even better quality of newborn screening services for the families in our region.

2012 MSGRC Annual Meeting

Don't forget that our Annual Meeting is scheduled for June 28-30, 2012 in Denver, Colorado. The meeting will include plenary sessions, project meetings, workgroup meetings, and a special screening of SUN KISSED, a documentary following the journey of Yolanda and Dorey Nez as they navigate their Navajo culture and the medical care systems that cared for their children. Registration for the

Annual Meeting will be opening soon, but please mark your calendars now!

As mild and beautiful April draws to a close, we're looking forward to working with all of you on workgroup projects and other activities to improve genetic services in our region. Meanwhile, we wish you cool nights and sunny days, and a few precious moments to enjoy this beautiful spring.

Warm regards,

 

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