



## Message from the Project Directors

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### In This Issue

[MSGRC 2012-2013 Annual Meeting](#)

[MSGRC Co-Sponsors  
ACMG Community  
Conversation](#)

[Closing Thoughts](#)

### Join Our List

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### Greetings!

Greeting from the mountains, where the snow has continued to fall on some of us, while others are enjoying the warmth of spring - or even summer. It was wonderful to see so many of you in Phoenix, and we'll use this message to go over some of the exciting activities we enjoyed with you there.

### MSGRC 2012-2013 Annual Meeting

Thank you to many of our readers and members for participating in the 2012-2013 MSGRC Annual Meeting in Phoenix, AZ on March 17-19, 2013. We had a very busy and productive meeting! The agenda included an update on the State of the Region from Dr. Celia Kaye, MSGRC Project Director, and Dr. Kathryn Hassell, MSGRC Associate Project Director. Many of MSGRC's national partners also presented on their work:

- **Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC)**, presented by Joseph A. Bocchini, Jr. MD, SACHDNC Chair & Professor and Chairman, Department of Pediatrics, Louisiana State University Health - Shreveport
- **National Coordinating Center for the Regional Genetic and Newborn Screening Collaboratives (NCC)**, presented by Alisha Keehn, MPA, Project Manager, NCC and Debbie Maiese, MPA, Evaluator, NCC
- **National Genetics Education and Consumer Network (NGECN) and Genetic Alliance,**

presented by Sharon Romelczyk, MPA, Project Manager, NGEEN

- **Newborn Screening Technical Assistance and Evaluation Program (NewSTEPS)**, presented by Marci Sontag, PhD, Director of Epidemiology, NewSTEPS
- **Newborn Screening Translational Research Network (NBSTRN)**, presented by Amy J. Hoffman, MPH, Project Manager, NBSTRN

We also learned about regional activities addressing newborn screening for Critical Congenital Heart Disease (CCHD) during a panel of presentations, including:

- **Colorado CCHD Screening Pilot Study**, presented by Jason Wright, MD, Neonatology Fellow, University of Colorado School of Medicine
- **Utah CCHD Demonstration Grant**, presented by Harper Randall, MD, FAAP, Medical Director, Division of Family Health and Preparedness, Utah Department of Health
- **Texas Pulse Oximetry Project (TxPOP)**, presented by Alice Gong, MD, Professor of Pediatrics, University of Texas Health Science Center at San Antonio and Charleta Guillory, MD, Associate Professor of Pediatrics, Neonatology Section, Baylor College of Medicine
- **Use of Telehealth in CCHD Screening**, presented by Robert Puntel, MD, FAAP, FACC, Staff Cardiologist, Phoenix Children's Hospital

We've posted many of the PowerPoint presentations from the meeting on our [website](#).

The Annual Meeting also included a number of workgroup and project team meetings, including the following:

- MSGRC Advisory Council
- Consumer Advocacy Workgroup
- Newborn Screening Workgroup with three breakouts (Laboratory, Follow-up, and Point-of-Care Screening)
- Medical Home Workgroup
- Emergency Preparedness Workgroup
- Telegenetics Workgroup
- MSGRC Metabolic Consortium
- MSGRC Hemoglobinopathies Advisory Group

The meeting marked the launch of the MSGRC Facebook page - be sure to "Like" MSGRC [here](#). You can view pictures from the Annual Meeting on the Facebook page.

There are many activities "in the works" based on these meetings and our discussions. Stay tuned for workgroup meeting minutes, draft workgroup logic models, and more information about our next meeting July 11-13, 2013. The July meeting will have more time dedicated to our workgroups - a draft agenda will be out soon!

### **MSGRC Co-Sponsors ACMG Community Conversation**

We were honored to co-sponsor the Community Conversation at the American College of Medical Genetics and Genomics (ACMG) Annual Clinical Genetics Meeting, which immediately followed the MSGRC Meeting. The Community Conversation, titled Emerging from Darkness to Shed Light on Genetics and Cultural Taboos in Native American Populations: A Community Conversation was held on the evening of March 19th and was open to public attendees as well as ACMG meeting participants. The tension between the potential knowledge that can be gained through genetics research and medicine, and cultural beliefs within the Native American populations of the United States, was explored at the inaugural Community Conversation held at the 2009 ACMG Annual Meeting ("Genetics and Genetic Research: Native American Perspectives".) With the release of the documentary, "Sun Kissed," (<http://sunkissedthefilm.com/>) these tensions are further highlighted as the Nez family works to understand why their children were born with xeroderma pigmentosum (XP) and why the prevalence within the Navajo community is 1 in 30,000 versus 1 in one million in the general population. "Sun Kissed" follows the Nez's as they negotiate between the cultural norms of their community and their information and support needs as parents of children with an incurable genetic condition.

The Community Conversation gave attendees an opportunity to view the documentary "Sun Kissed" and participate in a panel discussion with the Nez's and Maya Stark, one of the filmmakers. Dorey and Yolanda Nez are close friends and partners of MSGRC - be sure to check out the trailer for Sun Kissed!

### **Closing Thoughts**

This wonderful annual meeting reminded all of us at MSGRC of the critical importance of each one of our members - providers, laboratory specialists, public health experts, individuals with heritable disorders, and families. As MSGRC and the other regional collaboratives enhance our focus on access to services, we need all of you to advise us and help us ensure the availability and accessibility of

excellent services within the medical home for everyone in our region.

Don't forget we want to hear about what you do outside of your work in genetics and public health.

Here are two pictures from our colleague and friend, Dr. Mary Kukulich, on one of her trips to Angel Fire, New Mexico this winter.



Visit from a neighbor!



Walking my dogs in the snow!

Please send us your pictures and stories too!

Warm regards,

*Celia I. Kaye* *Kathryn Hassell*

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