



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

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October 2012

### Greetings!

Happy Halloween! We've had our first snow in Denver but are promised mild weather for trick or treating. We wonder if any of you will be out with your children, or even by yourselves. As the weather cools, we have more time to catch up with inside activities, and we've been very busy with MSGRC work. Here's an overview of the past month.

### MSGRC Business

As a reminder, the rescheduled MSGRC Annual Meeting will be held **March 17-19, 2013 in Phoenix, Arizona**, just prior to the American College of Medical Genetics and Genomics (ACMG) Clinical Genetics Meeting. If you are planning your travel for the ACMG meeting, please request a hotel reservation at the Sheraton Phoenix Downtown where we will also hold the MSGRC meeting. We have made arrangements with the hotel to allow a limited number of people to stay in the same room throughout both meetings. You will still need to make hotel reservations for the MSGRC meeting through our registration system, and we will work with the Sheraton to link with your ACMG reservation. We are awaiting a few final decisions before announcing the draft agenda but you should expect it very soon!

The MSGRC team continues to grow! MSGRC recently hired Kristi Wees to serve as the MSGRC Social Media Coordinator. In this role, Kristi will work with the MSGRC team (and all of you!) to strengthen the content on [www.MountainStatesGenetics.org](http://www.MountainStatesGenetics.org) and to expand our reach into social media. Prior to becoming a mom to her son and daughter, Kristi was a chemist, spending much of her time in a lab with beakers, test tubes, and instruments. She left the lab after having severe chemical sensitivities and went on to serve as a recruiting, marketing and sales manager of a scientific recruiting company.

Since the moment of her daughter's birth, Kristi and her husband have been on a roller coaster of a journey, medically speaking, for their daughter. From colic to food allergies, and from reflux to eosinophilic esophagitis, they now are walking on the mitochondrial disease path, searching for a metabolic disease that is not currently part of the NBS panels in her state (TX). In 2012, Kristi was chosen as a member of the Baby's First Test Consumer Task Force through Genetic Alliance. It was her hope that serving on the Baby's First Test task force will allow her to help even just one other parent avoid the pain and worry of wondering what is wrong with their child. Through her experiences with Baby's First Test, local awareness campaigns targeting other mothers, her blog documenting her family's journey, and through her Baby's First Test supported newborn screening awareness project, Kristi has utilized many forms of social media to make new connections with families. She envisions great opportunity through social media to spread the message that newborn screening saves lives, and to find and to offer support to others looking for genetic answers. More about Kristi:

- Read Kristi's blog ([Baby Food Steps](#)) about her family's journey.
- Read [Kristi's recent blog post for Baby's First Test here](#).
- Kristi is featured in [this video](#) produced by Baby's First Test.
- Kristi (and her son) recently completed [this video](#) for her newborn screening awareness project for Baby's First Test.
- In honor of Newborn Screening Awareness month, Kristi also completed a blog series called [Texas Born - Newborn Screening in the Lone Star State](#), which shared stories from 6 families stories who were impacted by Newborn Screening in TX.

She is excited to take on this opportunity with MSGRC as a Social Media Coordinator and continue spreading awareness and resources to all genetics stakeholders in this region. You can reach Kristi at [kwees@mountainstatesgenetics.org](mailto:kwees@mountainstatesgenetics.org).

#### 28th Meeting of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC)

The SACHDNC met in Washington, DC on September 13-14, 2012 and Celia Kaye, Kathy Hassell, Camille Miller, and Liza Creel attended. The Committee's agenda covered many topics including updates on the newborn screening case definitions and quality indicators projects, which many of you helped with last spring. Teams for both projects met in July 2012, and outcomes of these activities will inform the new Newborn Screening Technical Assistance and Evaluation Program (NewSTEPS). The Committee also reviewed a new condition review matrix that will help the Committee assess nominated conditions based on net benefit, certainty of the evidence, public health system readiness, and feasibility of screening.

The nomination and prioritization report for Adrenoleukodystrophy (ALD) was presented, and the Committee voted to not move ALD forward for evidence review until a large pilot study is completed and/or published. Other presentations offered information on the condition review for Pompe Disease; ethical issues and collaboration opportunities with the Ethical, Legal, and Social Implications Program at the National Human Genome Research

Institute (NHGRI); the Multistate Second Screen Study; the CDC Recommendations for Good Laboratory Practices in Biochemical Genetic Testing and Newborn Screening for Inherited Metabolic Disorders training module; the NCHPEG prenatal family history project; the Carrier Screening Draft Report from the Population-based Carrier Screening Work Group; and a summary of the IOM meeting Assessing the Economics of Genomic Medicine.

The three SACHDNC subcommittees - Laboratory Standards and Procedures, Education and Training, and Follow-up and Treatment - also met during the meeting. Our own Kathy Hassell was recently selected as a member of the Follow-up and Treatment Subcommittee, and this was her first meeting as an official member. The subcommittees have been prioritizing their activities for the coming year or two and spent time at the meeting further defining their projects. If you are interested in complete summaries of any SACHDNC meetings, you can visit <http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/>.

### Closing Thoughts

In November, our team will travel back to Washington, DC for the National Coordinating Center's (NCC) Annual Meeting. While there, we will focus on evaluation, learning more about expectations for our national evaluation and working with the NCC and other regional collaboratives to identify measurable quality indicators that help us understand our impact on the population. We will also be meeting with other regional collaboratives on a new family history project that we will highlight in the next Monthly Message.

Until that November message, enjoy these last beautiful days of fall, and don't forget to send your photos. We want to see you when you're not working!

Warm regards,



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