



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

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February 2015

Greetings,

We hope everyone is staying warm as chilly temperatures continue to descend upon many of the states in our region. As Spring draws near, we look forward to warmer temperatures!

Overview of the February SDACHDNC Meeting

We recently met with other regional collaboratives and partners in newborn screening at the Secretary's Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (SDACHDNC). Last September, a condition review update of Mucopolysaccharidosis (MPS I), which was under consideration for addition to the RUSP, was presented by Alex Kemper of the Duke Clinical Research Institute. This February, Dr. Kemper presented the final condition review update which covered MPS I diagnosis, genotyping, and treatment strategies. MPS I was voted into the Recommended Uniform Screening Panel (RUSP) by SDACHDNC. This recommendation will now go to the Secretary of HHS for her review and action.

While much interest centered around the Committee decision on MPS I, there were many other topics of notable interest. Jeff Botkin, Committee Member and Chair of the Pilot Study Workgroup, presented analysis on pilot studies that have been included in previous condition nominations. There was discussion on the different mechanisms and difficulties in carrying out pilot studies. Kellie Kerm and Susan Tanksley presented again on the Timely Newborn

Screening Project and the final recommendations were voted on. The recommendations have now been revised to stress time points most important to outcome- reporting positive results to the healthcare provider for time critical conditions by 5 days of life; reporting of all presumptive positive results for all other conditions to the healthcare provider by 7 days of life; reporting all NBS results by 7 days of life. This topic has continued to be of great interest in our region, and several of our states have made positive advancements in decreasing transit times of initial specimens to the laboratory. The Condition Review Workgroup presented on conducting cost analysis when implementing screening for a new condition.

Next steps for the SDACHDNC were discussed. For all future nominated conditions, the Committee will review and vote on within 9 months. To aid in this development, the committee now has authorization to conduct meetings four times a year. Currently, all three subcommittees remain (Laboratory Standards and Procedures, Follow-up and Treatment, and Education and Training). Reprioritization was discussed, and it was agreed that future workgroups should include representatives from all three subcommittees. We expect that many of the topics addressed at the SDACHDNC meeting will continue to be of great importance to our MSGRC members and colleagues. More detailed information on the meeting will be posted in the future on the Committee website linked [here](#).

Associate Project Director Opportunity

We are pleased to announce our recruitment of an Associate Project Director for MSGRC. Details on the position can be found [here](#). Please share this opportunity with colleagues that may be interested.

Registration for MSGRC Annual Meeting

As a reminder, registration for the MSGRC Annual Meeting is now open. All registration information is now posted on our website under [Events](#). Please register early!

Closing Thoughts

We are pleased to have experienced recent regional and national activities. We look forward to seeing many of you in the upcoming months at ACMG and the MSGRC 2015 Annual Meeting. In the meantime, please stay warm!

Warm regards,



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Acknowledgement

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under Grant H46MC24095. The Mountain States Genetics Regional Collaborative for \$600,000. This information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.



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