



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

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February 2014

We hope you had a wonderful Valentine's Day, and for those of you who have been snowed in, we hope the icicles are beginning to melt. It was absolutely wonderful to see so many of you at our MSGRC annual meeting in Phoenix. The flowers were blooming and the sun was shining, which was a welcome relief for so many of us. The food was outstanding, and the chocolates were good, too! We'll defer reporting on the details of the annual meeting until next month. For this message, we want to focus on the January meeting of the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC).

Members of MSGRC participated in a two day webinar and subcommittee meetings (January 16-17, 2014) of DACHDNC. The agenda for this meeting, as well as materials from many of the presentations, are available on the [committee website](#). We'll provide a brief summary here of activities that are of most direct concern to MSGRC members.

The Newborn Screening Saves Lives Act is before Congress for re-authorization this year. At the time of the DACHDNC webinar, the Senate Committee responsible for this law gave it unanimous approval, meaning that it can now move to the full Senate for discussion. The House of Representatives has not yet begun work on this bill.

Kathleen Sebelius, Secretary of Health and Human Services, reviewed the DACHDNC recommendation on addition of Pompe disease to the Recommended Uniform Screening Panel (RUSP). The Secretary referred the recommendation to the Interagency Coordinating Committee on Newborn and Child Screening (ICC) for additional input, and you can review her letter [here](#). Secretary Sebelius also reviewed the DACHDNC recommendation on retention and use of dried blood spots. The Secretary accepted four of the eight

recommendations, and [you can review her response here.](#)

Alexis Thompson presented an update from the Sickle Cell Disease and Screening for Trait in Athletes Ad Hoc Workgroup. The prior recommendations of DACHDNC were re-affirmed and [can be reviewed here.](#)

Marci Sontag and Jelili Ojodu presented an update on the NewSTEPS program, which is a cooperative agreement between the Genetic Services Branch, HRSA and APHL and the Colorado School of Public Health. The major focus of NewSTEPS is to function as a comprehensive resource center for state newborn screening programs through development of a data repository, website, and technical assistance program. You can [learn more about NewSTEPS here.](#)

A current high priority of NewSTEPS is the development of memoranda of understanding with each state regarding data reporting and sharing. MSGRC will be working with NewSTEPS to facilitate the signing of these MOUs with each of our mountain states.

Dieter Matern presented a report from the Nomination and Prioritization Workgroup on X-linked Adrenoleukodystrophy (ALD). This condition has been nominated for addition to the RUSP, and the workgroup has done a careful review of the nomination. Of particular interest was their review of data from the Mayo pilot of newborn screening for this disorder, as well as several lysosomal storage disorders. Based on review of information on the clinical findings, natural history, and response to treatment of the disorder, as well as preliminary data from the pilot, the workgroup recommended that ALD undergo an evidence review by the Condition Review Workgroup, chaired by Alex Kemper. DACHDNC accepted this recommendation, and the evidence review will now commence.

Susan Tanksley, co-chair of the Laboratory Standards and Procedures Committee, presented an update on Newborn Screening Specimen Transport. The impetus for this update was concern raised by families and consumer advocates that delay in transport of dried blood spot specimens, as well as delayed newborn screening laboratory results, may have adverse consequences for a newborn.

The Laboratory Standards and Procedures Committee sent a web-based survey to all newborn screening laboratory directors, as well as directors of newborn screening follow-up programs. This report was based on the response of 32 states. It was noted that many states do not currently record the time when specimens are received in the laboratory and therefore cannot track this information. Most states do not have regulatory authority to sanction birthing centers for delayed sending of specimens. Many laboratories do not have

Saturday or Sunday working hours, and many follow-up programs are also closed on Saturdays and Sundays. All reporting states indicated that they recommend specimen collection 24-48 hours after birth. Of those states able to report the time from specimen collection to receipt of specimen by the laboratory, 41% reported 2-3 days, and 47% reported 3 days.

The Committee reviewed recommendations published in Newborn Screening: Towards a Uniform Screening Panel and System (2005). This report, prepared with the leadership of ACMG, recommended that most results should be reported within 2 days of receipt of the specimen by the laboratory, and specimens should be received in the laboratory within 24 hours or 3 days of collection (report is inconsistent). Following discussion of this report by the DACHDNC, it was agreed that the Subcommittee will bring a proposal on timing of specimen transport and laboratory results to DACHDNC, and the parent committee will consider a recommendation to the Secretary.

Alex Kemper, Chair of the Condition Review Workgroup, presented an update on Mucopolysaccharidosis type 1 (MPS-1). The evidence review provided information on clinical findings, laboratory findings, incidence, life expectancy, and response to various treatments, including hematopoietic stem cell transplantation and enzyme replacement therapy. A newborn screening population pilot study is underway in Missouri, but full results are not yet available. The Condition Review Workgroup plans to complete its evidence review, speak with experts, and begin a modeling study to assess population benefit. APHL will work on a survey to assess feasibility and readiness of the public health system to begin screening. Additional information should be ready for the May, 2014 meeting of DACHDNC.

Many of the topics reviewed on the DACHDNC webinar were discussed at the MSGRC annual meeting. Newborn screening programs in our region have both interest and concerns about the addition of new disorders to the RUSP, and we'll be working with our workgroups and members over the next several months in an effort to address these concerns. But for now, let's look forward to March (the snowiest month in Colorado!) and the eventual arrival of those robins and little green shoots in the garden. For those of us where it's already warm, please send a picture of your park or garden. We'd love to share in your good fortune.

Warm regards,

*Alisa V. Kaye*



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