Mountain States Genetics
Regional Collaborative

Message from the Project Directors

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May 2014

Greetings,

May is a month of transition in the mountains - from cool weather to warm, from snow to rain (or sun, if we're lucky!)

It is also a time when children are finishing school for the year and looking forward to summer, and adults are beginning to think about summer vacation, too. For MSGRC, May is a time to say good-bye to Donna Williams, who will be leaving us as coordinator for MSGRC. Donna has provided wonderful support to MSGRC, and we will miss her. In June, we will welcome Marilyn Brown as the new project manager for MSGRC, and our June message will introduce her to you.

A major event during the month of May was a face-to-face meeting of the Secretary's Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (SDACHDNC). Three of us from MSGRC attended this meeting (Celia, Kathy and Joyce). What follows is a brief summary of a very full two day meeting.

Public Health Impact of Addition of New Disorders to the Recommended Panel for Newborn Screening

SDACHDNC has been concerned about the public health impact of adding new disorders to the recommended panel for newborn screening for several years. This concern was heightened when Health and Human Services Secretary Sebelius emphasized this issue in a letter to SDACHDNC in 2011, following the committee's recommendation to include critical congenital heart disease (CCHD) on the panel. Following the letter from Secretary Sebelius,

SDACHDNC changed its method of evaluating conditions nominated for addition to the panel. The new evaluation method allowed the impact on state public health systems to change the determination of net benefit of screening for a disorder. In keeping with this new evaluation method, states were surveyed by APHL regarding public health impact prior to final review of Pompe disease. SDACHDNC then voted to include Pompe disease on the recommended panel, and Secretary Sebelius referred this recommendation to the Interagency Council prior to making a final decision. Meanwhile, after discussion with state public health leaders, SDACHDNC determined that the public health impact analysis is still too weak. Discussions with public health leaders and HRSA leadership occurred, and it was decided to strengthen the public health impact analysis even further before proceeding with the evidence reviews for MPS 1 and adrenoleukodystrophy (ALD). An expert advisory panel on public health impact was convened April 10-11, 2014 at APHL to develop a systematic approach for collecting and evaluating the necessary information on public health impact. Key public health impact elements were determined at the meeting, including the need to consider incidental findings, detection of carriers, genotypes of uncertain significance, and late-onset disease. A major component of the final impact analysis will be the determination of how to weight or prioritize the various elements identified by the states. The next step in this whole process is circulation of a written report of the April 10-11 meeting to SDACHDNC members, participants of the expert panel meeting, and the regional collaboratives for input and feedback. We at MSGRC will share the draft document with state public health leaders in our region for comments and suggestions when we receive it. This revised methodology for review of nominated conditions will be used to review MPS-1.

Conducting Research on Population-Based Screening

Jeff Botkin from the University of Utah led a discussion on how state newborn screening programs could become even more active participants in pilot studies of disorders proposed for the newborn screening panel. Jeff noted that newborn screening programs do not generally have a research mission and lack the infrastructure to design and carry out large pilot studies. However, SDACHDNC requires pilot studies prior to adoption of new disorders to the panel. Jeff proposed development of a multi-state network to support large scale population-based pilot studies; infrastructure such as IRB support, shared data

agreements, and project coordination would be part of such a system. It was noted during discussion that the Newborn Screening Translational Research Network (NBSTRN), supported by the NIH and administered by the American College of Medical Genetics (ACMG), was designed to meet this need and played a role in the recent Severe Combined Immunodeficiency (SCID) pilot.

To move this discussion forward, a workgroup will be established with Jeff as chair. Goals of the workgroup will be to recognize and support what is already being done by NBSTRN regarding pilots; to identify other resources to support pilots; and to define more clearly what pilot studies need to be done prior to approval of a disorder for the newborn screening panel.

Lab Standards and Procedures Subcommittee Report

A set of slides about SCID is under development for use by newborn screening programs as they work toward the addition of this disorder to their state newborn screening panels. The intent of the slide set is to provide information for state labs as they consider implementation of SCID. We at MSGRC will let you know when this slide set is available for distribution.

This subcommittee is also working on the issue of timeliness of newborn screening. SDACHDNC has re-affirmed earlier recommendations on timeliness, including: initial dried blood spot (dbs) specimens should be collected at 24-48 hr after birth; dbs specimens should be received in the newborn screening lab 24 hr after collection; critical results should be available by 5 days of life; and all results should be available 5 days after collection. The subcommittee is working with the Society for Inherited Metabolic Disorders (SIMD) to assess the metabolic disorders with the most urgent need for early screening results; they will use the ACMG newborn screening ACT sheets and work with other specialists to identify the other disorders on the recommended newborn screening panel that require early results. They are also working with APHL to generate a survey with accompanying webinar to gather information on gaps, barriers, and best practices for timely screening.
This subcommittee presented a report on the use of succinylacetone (SUAC) as the primary marker for detection of tyrosinemia type 1. Most but not all states are using this analyte to detect the disorder. The subcommittee concluded that tyrosinemia type I needs to

stay on the recommended panel, and that SUAC is the best marker, independent of the analytical method. Based on the subcommittee recommendation, SDACHDNC will focus on education of newborn screening labs not currently using SUAC as a marker.

Follow-Up/Treatment Subcommittee

This subcommittee is nearing completion of two manuscripts that will be submitted for publication. The first focuses on lessons learned from newborn hearing screening programs that are applicable to screening for critical cyanotic congenital heart disease (CCHD). The second manuscript proposes a framework for assessing outcomes of newborn screening, using sickle cell disease (SCD) and PKU as example conditions. The goal of this second manuscript is to present a framework that would allow the newborn screening community to assess gaps in data that are necessary for determination of whether or not the goals of newborn screening are being achieved. We at MSGRC will do our best to let you know when these two manuscripts are actually in print.

This subcommittee is considering new projects. After discussion with the full SDACHDNC, it was decided that a description of the public health/clinical interface might be useful. The initial goal would be to determine how the public health components of the newborn screening system currently interface with hospitals and clinicians to provide the full range of newborn screening services in some example states. This proposal will be discussed more fully at the next SDACHDNC meeting.

Education and Training Subcommittee

This subcommittee has completed a project to assess what information might be helpful in deciding if a later onset condition should be considered for mandatory childhood screening. Information was collected on 3 paradigm conditions: fragile X, long QT syndrome, and Wilson's Disease. Categories of information that were considered included typical patterns of presentation; feasibility of population screening outside of the newborn period; information on the best scenario for early identification in the absence of population screening; and identification of important stakeholders that would need to be engaged in discussion about moving to population screening. Please note that there is no proposal to begin population screening for these or any other later onset disorders. This exercise was undertaken as partial fulfillment of the SDACHDNC mandate to consider heritable disorders in newborns and *children.*

As we move past this very informative SDACHDNC meeting, we want to ask your help as we work on some MSGRC priorities. First, as a reminder to our providers and families, the MSGRC Emergency Preparedness Workgroup has developed surveys for providers and consumers on how families of children with metabolic disorders are helped to prepare for emergency situations. These surveys can be found on our MSGRC website or by clicking here. The EP Workgroup would appreciate your help - please complete the survey if you or your family member is affected with a metabolic disorder, or if you provide care to individuals with metabolic disorders. MSGRC will be developing tools to help providers and families cope with emergency situations in our region, based on the results of these surveys.

Recently, we began to assemble consumer stories as was discussed at our consumer workgroup meeting in Phoenix earlier this year. These stories highlight the consumers in our region and their successes, challenges and insights into navigating the world of genetics. April was Fabry Disease Awareness month and highlighted Lori Wise's journey
and May was Neurofibromatosis month and highlighted Rod Slaght's journey. Please click these links to read their stories and keep checking our Facebook page
for more consumer stories to come. And a challenge- please share one or both of these stories on your facebook page to get the word out about genetic conditions and the amazing consumer advocates we have in our region.

Closing Thoughts

And, here is another request. Many of you completed the "working together" survey last year, which provided us with invaluable information on how we are doing in supporting workgroup and project activities. Would you take a few minutes to complete the survey again, so that we can see if we have improved based on your responses last time? You can find the survey at this link.

Thank-you, as always, for all you do for our families and each other in our mountain states region!

Warm regards,

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