Mountain States Genetics
Regional Collaborative

Message from the Project Directors

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September 2014

Greetings,

Summer is over, and we in the mountains are beginning to enjoy some cool days and nights and some beautiful colors. Fall holidays will soon be here!

SDACHDNC Meeting - General Overview

September found us meeting together with other regional collaboratives and newborn screeners at the Secretary's Discretionary Adivsory Committee on Heritable Disorders in Newborns and Children (SDACHDNC). Sue Berry from the University of Minnesota reported on the Inborn Errors of Metabolism Collaborative; we're happy to note that Jan Thomas and her group at the University of Colorado School of Medicine are now members of the collaborative and are including data from patients in the mountain states in this important national long-term follow-up database. Piero Rinaldo from the Mayo Clinic reported on the other data from newborn screening laboratories; many laboratories from the mountain states region participate in this project and take advantage of this powerful dataset to reduce false positives and improve the diagnosis of newborn screening disorders. Alex Kemper from Duke Clinical Research Institute provided a condition review update on MPS I, which is in the process of evidence review as it is considered for addition to the RUSP. Walter Suarez from Kaiser Permanente presented a report on recommendations to the Secretary of HHS from the National Committee on Vital and Health Statistics. Finally, SDACHDNC approved a recommendation to the Secretary regarding the need for a national dialogue among federal and state stakeholders on the benefits of measuring succinylacetone as a primary marker for tyrosinemia type I.

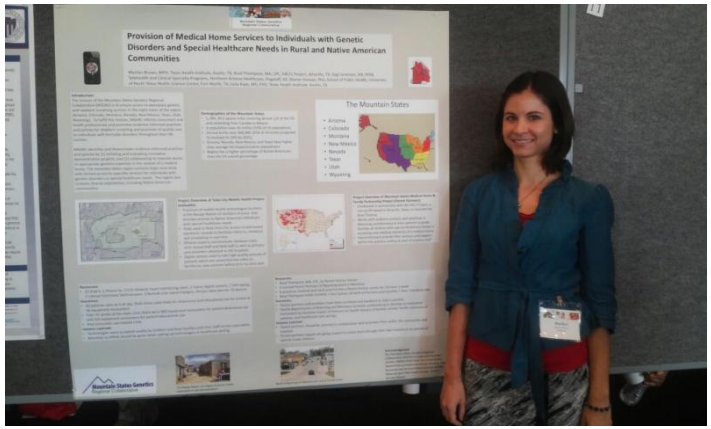
Of great interest to states in the mountain states region was the discussion on timeliness of newborn screening, led by Kellie Kelm and Susan Tanksley of the Laboratory Procedures and Standards Subcommittee. As most of you know, SDACHDNC is reviewing recommendations regarding timeliness of newborn screening that include collection of the initial dried blood spot (DBS) sample at 24-48 hours, receipt of the dbs sample in the newborn screening (NBS) lab within 24 hours of collection, reporting of time critical conditions by 5 days of life, and reporting of all results by 5 days after collection. The Laboratory Procedures and Standards Subcommittee was tasked with reviewing these recommendations and suggesting new recommendations if needed. To fulfill this task, the subcommittee held focus groups with two regional collaboratives and based on the focus group results, developed a survey with APHL that was sent to all states. Responses were received from all 50 states, although all states did not respond to all survey questions. The subcommittee will be summarizing all of these results and will use them to develop new recommendations regarding timeliness for SDACHDNC to consider. Preliminary review of some of the results showed that none of the recommendations are currently being met for all babies screened in the United States. All states are making efforts to meet these recommendations, within the constraints of geography, population, and funding that they encounter. After extensive discussion, the subcommittee will consider revising the recommendations to emphasize time points that are most important to outcome (reporting of presumptive positive results to the healthcare provider for time critical conditions by 5 days of life; reporting of 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). Recommendations that are helpful in achieving these goals will also be made: that initial specimens should be collected in the appropriate time frame for the baby's condition but no later than 48 hours after birth; and that NBS specimens should be received in the lab within 24 hours of collection. Based on discussion at the meeting, these recommendations will be refined and edited, and then presented for discussion again at the next SDACHDNC meeting in February 2015.

The Society for Inherited Metabolic Disorders (SIMD) convened a workgroup to determine time critical conditions in the RUSP that require earlier reporting. This list, which is currently still in review, was also utilized by the Laboratory Procedures and Standards Subcommittee. Hemoglobinopathy experts identified no disorders that require reporting by 5 days of life; endocrinologists indicated that congenital adrenal hyperplasia (CAH) results should be reported by 5-7 days of life, and that congenital hypothyroidism is time sensitive and should be reported by 7-14 days. Input from cystic fibrosis experts is still pending.

Altogether, the SDACHDNC meeting was filled with substantive discussion on issues that are of great importance to states, providers, and consumers in our region. For more detailed information on the meeting, please consult the committe website linked here.

Conference to Eliminate Health Disparities in Genomic Medicine

Marilyn represented MSGRC at the Conference to Eliminate Health Disparities in Genomic Medicine held in Washington, D.C. At the conference, she presented a poster on two of our current projects, Parent Partners and Mobile Health (mHealth). She was also joined by MSGRC colleague, Donna Holstein, who presented a poster on follow-up phone calls to ensure sickle cell trait diagnosis reaches the family. The conference brought together many stakeholders for discussion on the current health disparities that exist in genomic medicine and approaches to solving these disparities. We would like to express our appreciation for Brad Thompson, Parent Partners Project Director, and Gigi Sorenson, mHealth Project Director, for their valuable leadership and continued support.



Support Grey's 5k!

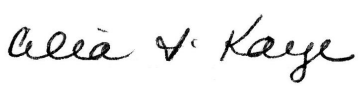
For all of our partners in Texas get ready to lace up those sneakers! MSGRC would like to support consumer advocate and Health Information Technology (HIT) co-chair Bill Morris, as his Grey's Gift Memorial Foundation (GGMF) holds its third annual "Grey's 5k - For Little Texans" on October 18, 2014, in New Braunfels, Texas. The mission of GGMF is to promote newborn screening education of the

public. Registration is available online through the GGMF website. Let's support Bill and have a record turnout!

Closing Thoughts

And now, as the days begin to grow shorter and the nights colder, I hope you're considering your Halloween costume. Please send us a picture to include in our next PD message.

Warm regards,



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