Mountain States Genetics
Regional Collaborative

Message from the Project Directors

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October 2014

Greetings,

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We hope everyone is ready to light those lanterns in search of some tasty Halloween treats. May the weather be mild for those of you with children planning trick or treating adventures! Here at MSGRC, in addition to resisting the temptation of the candy bowl, are some of our latest activities as we head into Fall.

Family Health History Project: Summary of Phase I and Thoughts on Phase II

In Spring 2014, we launched the Family Health History (FHH) mini- project. In the first phase of the Family Health History project, MPH students at the University of North Texas Health Sciences Center (UNTHSC) investigated existing family health history tools and summarized important considerations in the implementation of FHH tools in health practices. Our next step is to have discussions with our Medical Home and Health Information Technology co-chairs about (1) best ways to gather additional information on the practicalities of implementing a FH tool in obstetrics practices, family practices/pediatric practice in our region; and (2) exploring piloting a FH tool in those pediatric practices where the Parent Partner program (one of the MSGRC mini-projects) is being implemented.

Here are some of the highlights of what we have learned about FHH tools. Family health history is a written document of the diseases and health conditions from past and present family members. A useful family health history tool records information on at least three generations of a person's biological relatives, including family member names, relationships in the family clan, illnesses, diseases and deaths along with when these were diagnosed. In a clinical setting, providers may use family history tools to educate patients about possible present and future health risks. The family history can inform recommendations for screening and preventive measures, including lifestyle choices for lowering risk of developing illness/disease. We know that genetic, environmental and behavioral factors can potentially contribute to a person inheriting or developing an illness/disease. Examples of family history questionnaires or checklists are the Prenatal Genetic Screening Questionnaire, the Pediatric Clinical Genetics Questionnaire, and the Adult Family History Form (American Medical Association). Examples of the pedigree or family tree tools include the *Family Healthware* (CDC), the *Heart of Diabetes Family HistoryTree* (AHA) and the *My Family Health Portrait* (US Surgeon General's Office). Some family history tools consist of questionnaires/checklists as well as the pedigree.

Because of the wide usage of the Surgeon General's tool, you might find it informative to explore the tool at http://www.hhs.gov/familyhistory/. Since 2004, the Surgeon General has declared Thanksgiving to be National Family History Day, to encourage families to discuss and record information about the health problems that seem to run in their families. The Surgeon General's *My Family Health Portrait* is an internet-based tool that makes it easy for a person to record his or her family health history. The tool is easy to access on the web and simple to complete. It assembles information and makes a pedigree family tree that can be downloaded. It is private--it does not keep anyone's information.

In 15 to 20 minutes, *My Family Health Portrait* gives a person a health history that can be shared with family members or sent to a health care practitioner. If an individual shares his or her family health history with relatives, they can use this information as a starting point to create their own family health history. This is made even easier by re-indexing. "Re-indexing" means the person's relative becomes the center of the information, with his or her own family health history, and the original individual's information is automatically readjusted to show the correct relationship to the relative.

Research studies have been conducted for effective integration of family health history tools with electronic health records (EHRs) data. The *My Family Health Portrait* (MFHP) tool developed by the Surgeon General offers interoperability with EHRs. MFHP is ready to use with both EHRs and PHRs (Personal Health Records). In order to capture patient-generated family history data across diverse patient populations, EHR's may need to offer patients a variety of data entry options, customizing for preference, convenience, computer literacy, and computer availability. In a research trial conducted by Brigham and Women's Hospital, researchers developed and implemented three innovative portals to transfer and integrate patient-generated family history data with an EHR. The three portals included:

- computer tablets in waiting rooms to complete the MFHP;
- a secure internet portal to transfer data collected by patients at home using MFHP; and
- an interactive voiceresponse (IVR) system to collect necessary data elements by phone.

Based on this review and the recommendations of our MSGRC workgroups, we will proceed with this project here in the mountain states. Look for updates as the project progresses.

Planned Activities and Announcements

- **Hemoglobinopathies Long-term Follow-up Workgroup** will be meeting in Denver on November 17, 2014, to review the NHBLI guidelines and plan how to both disseminate and support these guidelines in our region.

- **HIT Workgroup** is currently drafting a white paper on Personal Health Records (PHR). The purpose of this paper is too educate consumers on PHR and start community conversation not only in our region but the other regions as well.

- **Telegenetics Workgroup** will be having a webinar with Brad Schaeffer, Project Director, and Lori Dean, Project Manager, of Heartland Regional Genetics and Newborn Screening Collaborative on their experience developing technical assistance for telegenetics. The webinar will be on November 10, 2014 at 1:30 pm CT. Please contact Marilyn

Brown if interested in attending. Also, please spread the word to others!

- For our **Consumer Advocacy Workgroup** we would like to announce the HRSA Regional Genetics Collaboratives' Advocate Leaders Partnership Program. At least one consumer for the MSGRC region will be selected to attend the American College of Medical Genetics (ACMG) conference from March 24th-28th on scholarship. This program is meant to broaden consumer advocate leaders' perspective of medical genetics and is a great opportunity to network with leaders from many areas of genetic medicine. While we welcome all applicants, preference will be given to those that have not previously attended. The informational flyer and application are linked and please do not hesitate to contact Marilyn for additional information.

Closing Thoughts

Although the hustle and bustle of the holiday season will soon be upon us, we will also be staying very busy here at MSGRC with our many activities planned and already in progress. For those of you that will be in costume this coming Friday please share. Photos from our dear friends and colleagues are always a welcome diversion during this fast-paced season!

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.

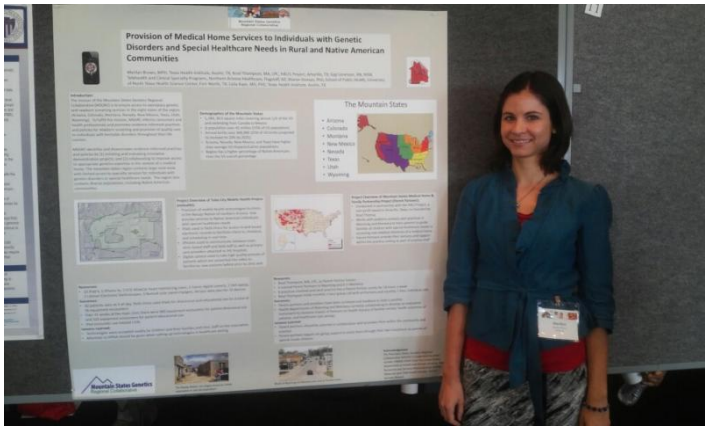
Of great interest to states in the mountain states region was the discussion on timeliness of newborn screening, led by Kellie Kelm and Susan Tanksley of the Laboratory Procedures and Standards Subcommittee. As most of you know, SDACHDNC is reviewing recommendations regarding timeliness of newborn screening 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.



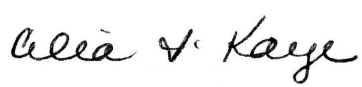
Support Grey's 5k!

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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