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February 2011

### Greetings!

January was a month of wild weather and cancelled travel plans for many of us in the mountain states region. Despite the challenges of travel, the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) met for their twenty-third meeting in Washington, DC. Although I was trapped in the Denver airport due to blizzard conditions in Washington, Liza Creel and Joyce Hooker were able to make it to Washington. As always, the SACHDNC had a full agenda. Of particular interest to those of us watching the addition of new disorders to the recommended panel for newborn screening, SACHDNC heard a report on the status of screening for Severe Combined Immunodeficiency Disorder (SCID) in California, Massachusetts, New York, and Wisconsin. As of the January SACHDNC meeting, over 575,000 babies have been screened for SCID in these four states. The Immune Deficiency Foundation is working to improve education and advocacy efforts around SCID, and the Newborn Screening Translational Research Network is updating ACT Sheets for SCID. Although none of the states in the Mountain States Region has implemented population-based screening for SCID, Texas is participating in the SCID screening study taking place in Massachusetts. We will plan for an update of these screening activities at our MSGRCC annual meeting in July.

SACHDNC continues to review additional conditions where point-of-care newborn screening is possible. During this meeting, they discussed a preliminary report on hyperbilirubinemia. The report summarizes the available literature on three case definitions: neonatal hyperbilirubinemia, acute bilirubin encephalopathy (ABE), and chronic bilirubin encephalopathy (kernicterus). In the coming months, the External Evidence Review Workgroup, led by Dr. James Perrin, will continue to collect evidence for the SACHDNC to review before making a screening recommendation. The final report should be available in May, 2011, and we will share the results with you.

During the SACHDNC Education and Subcommittee Meeting, MSGRCC member Brad Thompson presented the HALI Project. Through the HALI Project, Brad trains parents of children with special health care needs to partner with community medical providers to offer some of the non-medical components of the medical home (navigation of social, educational, and other systems outside the doctor's office). These parent leaders work within the medical setting to supplement the medical services families are already receiving. Currently, The HALI Project is partnering with Boy's Town to provide the parent training in Omaha, Nebraska. Brad is a co-chair of the MSGRCC Consumer Advocacy Workgroup. The SACHDNC Education and Training Subcommittee is also working with the CDC to explore a newborn screening public awareness campaign. The first step is planning and strategy development; the group will examine potential target audiences, clarify the message about newborn screening, identify qualitative and quantitative measures for success, and outline implementation of the campaign.

The SACHDNC Long Term Follow-Up Subcommittee discussed the challenges of implementation of newborn point-of-care screening (including critical congenital cyanotic heart disease and hyperbilirubinemia). Separate workgroups are talking about the role of states in these hospital based programs, including the challenges to state public health programs for tracking these infants. There will be much more to hear as providers discuss the issues related to this type of screening, where public health and health care delivery intersect.

In other news, new NCQA standards for the patient centered medical home were released in late January and can be found at <http://www.ncqa.org/tabid/1300/Default.aspx>. Finally, a Genetic Counseling Cultural Competence Toolkit was recently launched and is directly accessible from <http://www.geneticcounselingtoolkit.com>, which is the final product of the 2009 Jane Engelberg Memorial Fellowship (JEMF) award of the National Society of Genetic Counselors.

I'm looking forward to seeing many of you at the MSGRCC mid-year workgroup meetings in Phoenix in February. We are excited to be also hosting our second Medical Home Visiting Professorship (MHVP) in conjunction with the mid-year meetings. The MHVP is funded by a grant from the American Academy of Pediatrics. Dr. Chuck Norlin, our visiting professor, will be describing the medical home portal. For those of you who are attending one or both of these meetings, please say hello and let me know how you are doing!

Warm regards,

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