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September 2010

Greetings !

Summer is nearly over, and fall activities have begun to find their ways onto our calendars. I hope some of you are hiking, apple picking, harvesting pumpkins from your gardens, and getting your vehicles ready to brave the snowstorms that will be coming soon to some of our states.

This month I'd like to highlight one of the projects that has been funded by MSGRCC since 2005. **Janet Thomas**, Associate Professor of Pediatrics and Director of the Inherited Metabolic Diseases Clinic at The Children's Hospital in Aurora, Colorado, directs a project entitled the MSGRCC Metabolic Newborn Screening Long-term Follow-up Study. In 2005, when this project began, only about half of the state and territorial newborn screening programs in the United States conducted long-term follow-up. Of those states, only about half had a standard protocol in place, and there was great variation in what was followed. The primary objective of this study was to develop a systematic, long-term follow-up program over a large enough population so that factors that affect long-term outcome of patients with inborn errors of metabolism diagnosed through newborn screening could be identified. Through this project, the MSGRCC Metabolic Consortium was established, which represents metabolic physicians, nurses, genetic counselors and dieticians from AZ, CO, MT, NM, TX, UT and WY. Disease-specific care plans and outcome measures have been finalized and implemented for 28 metabolic disorders detected by newborn screening. The care plans include baseline and follow-up data sets for each disorder, including specific developmental assessments. The Colorado Clinical Health Information Records of Patients (CHIRP) database, developed by the Colorado Department of Public Health and Environment (CDPHE), has been modified for this project, and pilot data for PKU and MCADD patients are now being entered. A manuscript describing the project is in press (MSGRCC Metabolic Newborn Screening Long-Term Follow-up Study: A collaborative multi-site approach to newborn screening outcomes research. **Erica Lynn Wright, Johan L.K. Van Hove, Janet Thomas**, Genetics in Medicine

(in press)). We are particularly proud of this project because the care plans developed by Dr. Thomas and her group have been adopted with modifications by other regions; the format is now also being used by expert groups caring for children with endocrine disorders and hemoglobinopathies so that uniform data sets for these disorders can also be collected. **Janet Thomas** and **Chris Wells** have now been funded by HRSA to enhance data collection through the CHIRP database, integrating newborn screening follow-up data with other state databases and developing systems to provide these data to primary care providers. Most important, data from this project will allow us to identify important variables, including specific treatments, that improve long term outcomes for these children.

MSGRCC participated in the September meeting of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC), during which we provided an update on RCC activities to the Subcommittee on Education and Training. Specifically, **Katharine Harris**, Project Manager for the New York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC), highlighted their recent emergency preparedness tabletop exercise. The Genetic Services Branch at HRSA is hosting a strategic planning retreat in October, and we are happy to be participating in that meeting as well. I have had the privilege of serving on the Steering Committee for the retreat, and I know all of the regions will contribute their unique perspectives as GSB looks toward the future. We will be reporting on the outcomes of both of these meetings in future messages.

Look for reports on other MSGRCC projects in future messages as well. We're privileged to work with a remarkable group of dedicated and energetic professionals and consumers in our region, and we want all of you to know what they are doing.

Warm regards,



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