Mountain States Genetics
Regional Collaborative

Message from the Project Directors

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Greetings,

Perhaps you noticed that we usually begin these messages with a comment on the garden, the holidays, the change of seasons, or the weather. Today we want to call your attention to sports. We don't think the Olympics are going on right now, and the Super Bowl happened a couple of months ago. It's too early for ....

ACMG Meeting - General Overview

Moving on to genetics and newborn screening, the American College of Medical Genetics and Genomics (ACMG) held its annual meeting March 25-29 in Nashville, Tennessee. We presented a poster on our telegenetics activities, "Provision of Genetic Services through Telegenetics in the Mountain States: Experience with 4 Programs". Our poster highlighted the impressive activities of Cook Children's Hospital in Fort Worth, TX; the Marion Downs Hearing Center Foundation in Aurora, CO; the Billings Clinic in Billings, MT; and the Flagstaff Medical Center in Flagstaff, AZ. As the individual who got to stand next to the poster for two hours, I was truly gratified to see the interest in what we are doing here in the mountain states. The examples provided by these four programs will be useful to geneticists and their patients throughout the United States.

The ACMG meeting provided a venue for face to face meetings of the Medical Home Workgroup and the Transition Workgroup of the National Coordinating Center for the Regional Genetics Collaboratives (NCC). The Medical Home Workgroup, chaired by our own Chuck Norlin, is narrowing in on two projects. The first project is

seeking to identify best practices for care coordination in the medical home. The Workgroup hopes to engage the regions in nominating specific practice groups that provide excellent care coordination for children with heritable disorders. Once identified, members of these practices will be interviewed to identify barriers to as well as enablers of excellent care coordination. The second project will ascertain how families identify services and support. Using the general pediatric practices involved in the American Academy of Pediatrics Genetics in Primary Care Institute, practices will be interviewed to identify barriers and mechanisms that support coordination.

The Transition Workgroup is working to define successful transition and the methods for measuring this success at the level of the individual. Definitions of success may differ within pediatric and adult care settings, and it is important to clarify these differences. The Transition Workgroup is also designing a study centered around index cases that will explore quality of life, including but not limited to health concerns. At a joint breakfast of the two workgroups, it was clear that there is overlap between their goals, and the two workgroups will work together closely to avoid duplication and to share resources where possible. For example, both groups hope to identify high performing practice groups. They will coordinate their efforts in the search for these groups, rather than looking independently. They will also identify and share validated measures of quality of life, and they will coordinate any surveys sent to consumers and providers.

ACMG Meeting - Consumer Leader Program

Two MSGRC consumers, Joe Martinec and Rita Aitken, attended the consumer leader program at ACMG.

Feedback from Joe:

"It is critical to my effort to serve well that I have the fullest possible exposure to the efforts of other people, their successes and failures, the means by which hurdles were encountered and overcome. The ACMG meetings I have attended have been refreshing and energizing...The frequent meetings with other family advocates from other states and regions invariably produce an exchange of ideas. Starting with breakfast, following through at lunch, and often continuing with an evening event, the regional representatives can not only initiate conversations but can sustain those conversations with each other. The benefit I receive from those extended conversations allow me to get to know and appreciate the daily lives of people who are carrying on the struggle in different states and different regions, often dealing with issues with which I am not familiar or which I had not previously recognized... It is especially gratifying when I am able to suggest a method, a solution or a process that is helpful to someone else.

If there was one revelation to me at the AMG meeting, it is the plight and frustration of parents, patients, consumers and family members who are on a "diagnostic odyssey", a term...which conjured up childhood tales of brave Ulysses and the monsters and travails he had to overcome. I am amazed by the persistence, realistic optimism and good cheer of parents who get no more encouragement than learning that genetics plays some unknown role in their child's medical difficulties. In some way, those parents are forced to focus their efforts on and encourage a broad success in the entire developing science of the genome, rather than hoping for a solution to one known and treatable condition. Theirs is a harder journey to be on, and more like the much-chronicled ancient odyssey into an utterly unknown world filled with real and imagined existential threats. They are my heroes.

Feedback from Rita:
My conference experience began with the Community Conversation...it was really refreshing to hear the perspective of transition as a movement into all facets of adulthood, not just healthcare transfer. I know those experiencing transition are the best source of information on this topic, and it was good to hear it from them.

Since I've been back I've been talking constantly about what I learned...One of the topics was so timely, it alone would have made the trip a success for me, it was the session on Krabbe screening in NY. New York state screens for Krabbe and the presenter reported findings of their screening and treatment. This was very important to me because my state, which requires legislation to amend the NBS panel, is considering legislation this spring to add Krabbe. As the bill came up in committee, I and others brought it to a Newborn Screening Partners meeting and to the attention of the geneticists, whose input had not been sought. Shortly thereafter a rewrite left the committee, recommending Krabbe be added, dependent upon director approval, rather than simply requiring Krabbe to be added as it was originally written.

Another session that spoke to me was the undiagnosed conditions, held Friday evening. I was not sure what to expect when we arrived to wine and snacks. But I must tell you that the session made me proud to be even remotely associated with the professionals who attended. I have always understood the importance of phenotype, but had not realized that this meeting provided a forum for input by hundreds of professionals in an almost face-to-face manner. The respect shown for the patients as well as the families' preferences was, yet another time, reaffirming. As

a parent, whose now adult children have been and continue to be similar subjects, I felt extremely grateful for the generosity of spirit and knowledge displayed here.

Other MSGRC Priorities and Projects

Now that we're home from Nashville, the other work of MSGRC continues. There is great interest in the impact of expansion of the recommended panel for newborn screening on departments of Public Health in our region. Kathy Hassell (Colorado), Susan Tanksley (Texas), Ned Calonge (Colorado), and Erica Wright (Colorado) served on an Impact Panel convened by the Secretary's Discretionary Advisory Committee on Heritable Disorders in Newborns and Children on April 10th and 11th. There was lively discussion about the key elements of implementation and potential impact on State NBS programs by the adoption of new testing added to the Recommended Uniform Screening Panel. This led to recommendations for specific questions that should be asked of all States in areas of resources, training, and follow-up before a final decision by the Secretary's Advisory Committee to add a new test.

We also continue to work quickly and hard on our various projects related to implementation of the Patient Protection and Affordable Care Act (ACA). The Metabolic Consortium and the Hemoglobinopathies Interest Group are nearing completion of templates for review of covered services for PKU and Sickle Cell Disease in state benchmark and Medicaid plans. This project, which is being done in coordination with the other regions and the NCC, is designed to improve coverage under ACA for genetic disorders throughout the life course. In addition to developing templates to review specific conditions, this project will develop a white paper describing services that should be provided to individuals with genetic disorders in Accountable Care Organizations and integrated health systems.

Next Steps!

Also related to the ACA, here is an ACTION ALERT for you. Please help us share our new Facebook campaign about the Affordable Care Act called ACA Café: One Cup at a Time!

We intend to provide small cups of info in an attempt to simplify ACA for Children and Youth with Special Health Care Needs (CYSHCN) families and consumers.

Please log on to the MSGRC Facebook Page and Share our "Cups" by posting on your FB Pages!
facebook.com/mountainstatesgenetics.org

Closing Thoughts

I hope it's clear that we know more about caring for individuals with heritable disorders than we know about sports. Now that its spring (at last, a mention of the season!) we hope you're organizing croquet tournaments in your back yards and looking forward to neighborhood swim competitions in just a few more weeks.

Warm regards,

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