

Dear NEGC partners,

I am pleased to be writing my first update on the activities of the NEGC during the most recently completed quarter. Below are a summary of activities, by work group/topic area.

Dissemination, Education and Marketing

The DEM group has completed a regional focus group review of our new education tool for special education teachers (3 groups held: VT, NH, CT). Three versions of the tool (PKU, Sickle Cell Disease, and 22q deletion) were presented. The results of these meetings have been reported, and are being summarized; this summary will be used to improve teacher access to, and utilization of, the tool.

Medical Home and Transition

The Medical Home and Transition Pilot Study is designed to facilitate health care follow through between specialists, primary care providers, and families. The study protocols are complete, and care protocols are being implemented with new patients.

Assessment of Transition Practices in the Region is an inventory of transition practices. The assessment has been completed, and Susan Waisbren is developing a white paper documenting next steps to improve transition practices in the region.

The Transition group is working on fact sheets for parents with a child with a genetic disorder. The goal is to complete four new fact sheets (galactosemia and PKU are complete).

Quality Improvement

A vendor has been reviewed, selected, and hired to implement the data registry for the clinics participating in the QI project around children referred for developmental delays. This project is focused on understanding patient needs, care processes, and needed areas of improvement. (See press release at: <http://www.iod.unh.edu/6-14-10.html>)

The Metabolic Clinic Quality Improvement Learning Collaborative is underway. The Planning Group and Expert Panel have met three times in the last two months, and met again in June. These groups have scoped out an initial set of data collection tools (using work by NYMAC and MSGRCC), performance measures for the initiative, and a timeline to carry out the work of the collaborative during FY11. During the summer we will be piloting the data collection tools for PKU and MCAD.

Quality Assurance

The work of the Quality Assurance group continues to improve accuracy of lab tests as members expand their analysis work to include new markers and their utility in establishing better identification methods for children with genetic conditions. To date, analysis has been completed on the C3 and C5 markers. The group is currently working on expanding its analysis with additional markers.

LTFU

Regional LTFU initiatives enable better identification and follow-up of children born with genetic conditions in the New England region. During the last quarter, Maine saw passage of legislation enabling tracking of LTFU data. Partnership building is continuing to develop with Vermont and Rhode Island. This group is continuing to work with clinical workgroups on QI initiatives, and on developing a psychosocial data set, potentially starting with MA data.

Other Projects

The NEGC is working with our partners to complete the following special projects:

- Newborn Screening Clearinghouse: working with a committee to identify where some of the gaps are in knowledge for families and health care providers, and to determine how the Clearinghouse can best address these gaps.
- Genetic Workforce study: interviews have been conducted with metabolic clinicians, genetic counselors, nutritionists, neurodevelopmental psychologists, nurses, and NBS coordinators to document workflow processes for children who are NBS positive. These data will be analyzed to provide an assessment of resources needed to provide quality genetic health care to a population of infants, due to a potentially expanded NBS panel.
- Review of Legal Issues in HIT/HIE: review laws and regulations in all New England states specifically related to the exchange of health data which may contain genetic information. Barriers, impediments and challenges to conducting HIT/HIE activities within and between states will be identified.

Providing Training Opportunities

NEGC, along with the Genetic Alliance, sent a lay person from New England to attend the Genetic Alliance Advocates Partnership Program at the ACMG annual meeting in Albuquerque, NH. The participant reported back on discussions around the need for genetic expertise across health care, particularly about DNA Direct - a company launched in 2005 to address the growing need for genetics expertise in health care.

NEGC sent seven nutritionists representing every NE state to the Genetic Metabolic Dieticians International (GMDI) Educational Conference in Baltimore, MD, in April. Feedback from one of the participants: "It was an amazing experience and one that has proven to me that metabolics is the direction I want to pursue in my career."

Community and Family Network Grants

Four family support organization members attended a leadership development conference. A recipient questionnaire to assess how this experience relates to NEGC mission and its impact on the individual was developed, and responses are being received detailing some of the benefits of this resource for consumers.

Innovative Project Awards

NEGC initiated the fourth round of *Innovative Project Awards* this spring. The review panel met on Friday, June 18, and selected the following projects for funding:

1. Exploring and identifying the knowledge level and attitudes of (selected) diverse populations toward genetics and genetic services – *Patricia Rissmiller, Simmons College*
2. Development & Initiation of a New England Birth Defects Consortium - Yr 2, *Stephanie Miller, Dartmouth College*
3. Increasing Access to Care for Newborn-Screened Children with Fatty Acid Oxidation Disorders, *Susan Waisbren, Children's Hospital Boston*
4. **Partial Funding for** Integrative Community-Based Management for Adults with Sickle Cell Disease, *Victoria Odesina, UConn Health Center*

Communications

In April, the NEGC held a very successful communications planning day with the aid of Kathy Beal. As a result of our work, the NEGC has undertaken a substantial re-vamping of its website, reviewed our communications materials with a focus on improving access to the information, and identified new and innovative ways to reach out to our partners. (Did you notice our revised logo on the first page?) These new tools should be implemented in July.

In addition to these regional activities, John and I participated in the National Coordinating Center Project Director/Project Manager meeting on June 7. Topics that were discussed with all seven regional collaboratives included: emergency preparedness, IRB issues, and advisory committee governance.

As always, we welcome your thoughts and suggestions on how the NEGC can better fulfill its mission to promote and improve health and social well-being of those with inherited conditions through collaborations among public and private health professionals, educators, consumers and advocates in New England.

Best Regards,

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