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# New England Genetics Collaborative Annual Evaluation Report for Project Year Four

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**New England Genetics Collaborative**  
**Annual Evaluation Report for Project Year Four**

**Reflections on Project Activities 6/1/10- 5/31/11**

*By: Peter Antal, Ph.D.*  
*NEGC Project Evaluation Staff*

*February 2012*

**New England Genetics Collaborative**  
**Annual Evaluation Report for Project Year Four**

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## EXECUTIVE SUMMARY

This annual report covers the activities of the New England Genetics Collaborative (NEGC) from June 1, 2010 to May 31, 2011. The purpose of this report is to provide the reader with additional documentation on the utilization of grant funds and what has been achieved as a result, to provide an overview of NEGC activities for both old and new partners, and to offer recommendations for the collaborative's improvement and ultimate achievement of its mission and vision.

*Mission: The mission of the NEGC is to promote and improve the health and social well-being of those with inherited conditions through collaborations among public health professionals, private health professionals, educators, consumers and advocates in Maine (ME), New Hampshire (NH), Vermont (VT), Massachusetts (MA), Rhode Island (RI) and Connecticut (CT).*

*Vision: All individuals with genetic conditions living in New England have the opportunity to achieve their fullest potential.*

This report includes: a summary of activities by the Regional Coordinating Council (RCC), Workgroups, and Evaluation Staff during the period; primary findings of the project's fourth stakeholder survey; an update on the status of core project components from Year Four; a list of objectives for each group for Year Five; and recommendations to the project by the project evaluator. The material provided in this report is based on information submitted to evaluation staff as of Oct. 1, 2011. Members of the Collaborative Council were provided an opportunity to review and comment on the enclosed material. Evaluation of the project is led by Peter Antal, Ph.D., Institute on Disability, UNH.

The current New England Regional Genetics and Newborn Screening Collaborative (NEGC) grant (HRSA Grant # U22MC10980) officially began June 1, 2007. During its fourth year of activity, core project staff have continued to focus on improving the infrastructure of the NEGC (launching the new website, improving the RFP process, and structural improvements to the organization) and increasing support to coalition members. Together with the Workgroups, they have been meeting and carrying out the work of the NEGC through a broad range of collaborative activities, including a special focus on metabolic centers workforce capacity and launching New England's first Emergency Preparedness symposium. The Quality Improvement Workgroup completed business associate agreements with centers in Maine and New Hampshire with Vermont's in process to support quality improvement activities and successfully launched a new learning collaborative with 8 participating metabolic centers representing all NE states. The Transition Workgroup continued to build on both regional and national level activities, implementation of a new Teen Challenge Program, disseminating the Transition Toolkit, and collaborating with national partners. The Medical Home Workgroup pursued the development of a new survey needs assessment to document care, coordination and communication practices among primary care providers, specialists and families. The Dissemination, Education, and Marketing Workgroup developed the framework and background material for the new GEMSS website for special educators to improve support for students with genetic conditions. The Laboratory Quality Assurance Workgroup conducted analyses and compared results to follow-up for: 3MCC, BKT, GA-I, MSUD, CIT-I, and ASA, with a presentation to the Laboratory Subcommittee of the SACHDNC. Lastly, the Long-Term Follow-up Workgroup achieved a major accomplishment by solidifying an agreement with legal

representatives from Rhode Island which allows for the collection of LTFU data. Additionally, they held a national conference on improvement of long-term outcomes for individuals with Sickle Cell Disease.

Concerning stakeholder satisfaction with the progress of the NEGIC, findings from the recently completed stakeholder survey showed multiple improvements over the previous year. A majority of respondents (N=63) understood the mission of the NEGIC (73%) and felt that the NEGIC has made clear and substantive progress in achieving its mission (72%). In reviewing the goals and objectives for Year Four, 96% of 54 objectives have either been completed (63%) or have made satisfactory progress (33%) in accordance with the long term goals of the grant. Objectives for Year Five have been shared and agreed to by project staff and chairs of the project's work groups. In preparing to successfully meet the collaborative's objectives, a range of challenges and recommendations for improvement have been identified in the final section of this report.

## COALITION CHANGES AND IMPROVEMENTS

### Organizational Overview

The Regional Coordinating Center (RCC) is staffed by John Moeschler, MD and Monica McClain, Ph.D., who serve as Principle Investigators, Ms. Karen Smith as Project Coordinator, Kit McCormick as Project Staff, and Peter Antal, Ph.D. as Project Evaluator. Administrative support is provided by the UNH Institute on Disability, which acts as fiscal agent.

In 2010 – 2011, the RCC carried out substantial portions of its work through six Workgroups: Quality Improvement, Medical Home, Transition, Laboratory Quality Assurance, Long Term Follow-Up, and Dissemination, Education and Marketing. The chair of each Workgroup is a member of the Collaborative Council; the Council meets three times a year to facilitate coordination of Workgroup activities. The RCC and Collaborative Council are guided by an Advisory Committee which meets annually to help set direction for the collaborative and to provide feedback / raise issues throughout the year as needed. Lastly, a Review Committee is formed annually to provide review and guidance on funding requests from the collaborative's innovative projects program. Please see Appendix A for the current organizational chart.

### Organizational Improvements

During Year 4, the NEGC staff focused on strengthening its communication strategies and supporting sustainability efforts for genetic services region wide.

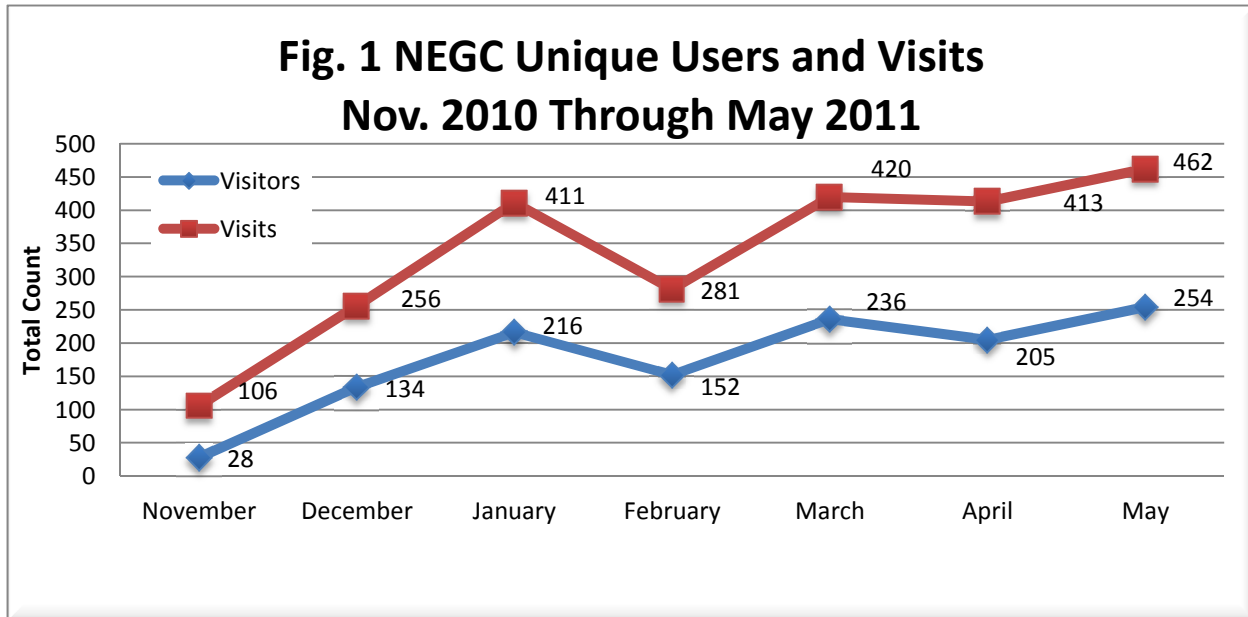
### Implementing the Communication Plan

During the course of the project year, project staff launched a new e-newsletter describing NEGC activities and important updates for partners and made a range of enhancements to the project's website ([www.negenetics.org](http://www.negenetics.org)). Enhancements were made in the following areas:

- Structural
  - New site launched in December, 2010
  - Expanded list format for presenting information
  - Improved access to information for families and professionals
  - Added join our mailing list option, twitter link
  - Improved access to products and publications
  - Enabled password protection for joint team review of grant applications for innovative projects
- Information
  - Added more state resource information
  - Provided updates to Did You Know and Featured Resource allowing for easier highlighting of key activities
  - Extracted glossary of terms and made accessible on website
- Design
  - Updated graphics throughout to reflect diversity
  - Redesigned logo

NEGC Visitors

Between November 2010 and May 2011, there were 962 unique visitors to the NEGC website and 2,349 visits. Over the course of these 7 months, the number of users and visits gradually increased, the average time spent on the website increased from 3:52 to 5:31 minutes, and the average number of page views increased from 2.4 to 6.0, with total page views increasing from 258 to 2,758.<sup>1</sup>



As the NEGC grows it will be helpful to track how the NEGC is doing relative to its outreach efforts and to identify what kinds of outreach works best. In looking across different referral sources to the website and the levels of activity generated (See Table 1), the most effective source was direct web links provided to stakeholders, with 77 visits generated and an average time of 6:58 minutes per visit, followed by referrals from partner organizations which generated 337 visits and an average time of 4:23 minutes. Of note, links driven by social media (Tumblr, Facebook, Twitter) accounted for 200 visits, an average time of 0:55 minutes on the website and a bounce rate of 89%. Other refers primarily to web market generated referrals from PRweb.com and other unaffiliated sites.

<sup>1</sup> The reader should note that these numbers are lower than previous years due to the fact that a completely different tracking system was used in conjunction with the new website. The new program, run through Google Analytics, provides a more accurate and detailed view of NEGC website users. Given the substantive changes, the numbers presented this year are not comparable with data from the previous year.



*Sources of Referral to the NEGC Website*

**Table 1: Referring Sources to the NEGC Website**

<b>Source</b>	<b>Visits</b>	<b>Pages/Visit</b>	<b>Avg Time</b>	<b>% New</b>	<b>Bounce Rate<sup>2</sup></b>
Direct Link	777	6.6	0:06:58	28.7%	36.4%
Partner Org	337	4.6	0:04:23	35.0%	38.9%
Email Referral	23	4.2	0:01:41	47.8%	52.2%
Search	871	2.7	0:01:40	66.1%	55.5%
Social	200	1.6	0:00:55	2.5%	89.0%
Other	54	1.6	0:01:25	3.7%	50.0%

Outside of the IOD, the top five organizational drivers to the NEGC website was the NCC (88), NBS Clearinghouse (22), MCH LEND (21), CDC (19), and the New England Consortium of Metabolic Programs(11). A total of 21 organizations were identified as referral sources to the NEGC.

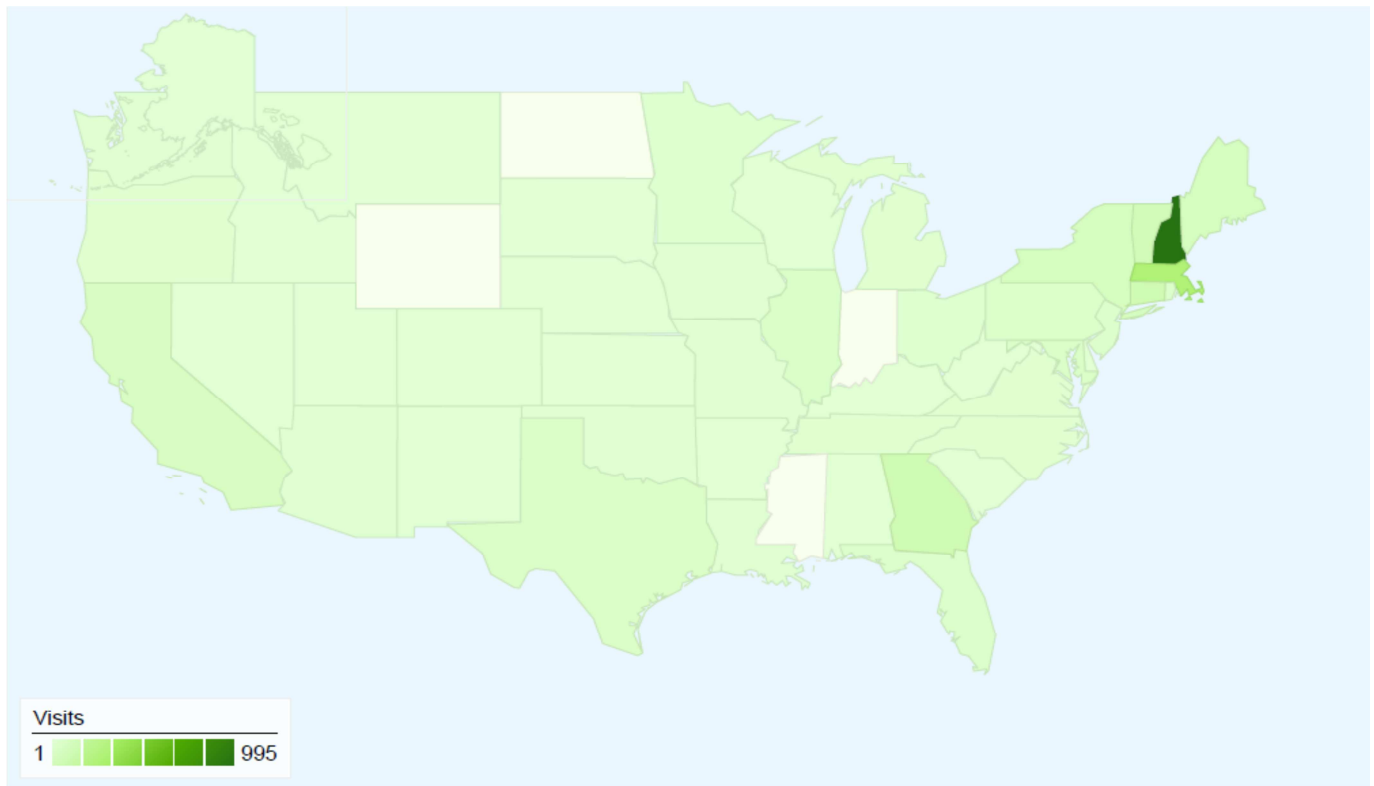
*National Outreach of the NEGC*

Of the 2,165 visitors to the NEGC website, about half were from New Hampshire (995)<sup>3</sup>. States with 50 or more visitors include: Massachusetts (277), Georgia (96), Connecticut (96), Vermont (88), New York (61), Maine (60), and California (51); please see Fig.1.

<sup>2</sup> Bounce rate refers to percentage of initial visitors to a site who "bounce" away to a different site, rather than continue on to other pages within the same site

<sup>3</sup> However, 604 visits (61%) of the NH traffic were generated from the city of Durham (where the administrative staff of the NEGC resides).

Figure 1: Distribution of NEGC Website Traffic by State



**Resource Leveraging**

During Year Four, NEGC staff submitted one grant application to support new or expanded work in genetics in the New England region. This application was not funded. The NEGC provided 5 letters of support for five projects, two of which were funded.

**Table 2: Applications and Letters of Support**

<b>Direct Applications</b>		
<b>Grant Name</b>	<b>Description</b>	<b>Amount</b>
Natural History of Disorders Identifiable by NBS	Project Yr 4. NIH.	NOT FUNDED
<b>Letters of Support for Partner Applications</b>		
<b>Grant Name</b>	<b>Description</b>	<b>Amount</b>
Genetics in Primary Care Institute	Project Yr 4. American Academy of Pediatrics. Create a community of learners to enhance primary care provider ability to provide genetic related services, address	Funded.

	systems and policy supports to accelerate provision of genetic medicine, assess residency training curriculum for genetic medicine.	
Family to Family Health Information Center	Project Year 4. Federation for Children with Special Needs.	Funded.
NBSTRN	Project Year 4. American College of Medical Genetics. Build an electronic data capture tool for long term follow up of children identified by newborn screening.	Letter written to support need for this activity.
Noonan Foundation	Project Year 4. Children's Hospital Boston. For follow up meetings of the Face Forward Program.	Not funded.
The Parent-Child Relationship and Newborn Screening: Preserving the Ties that Bind	Project Year 4. Assess whether the parent-child relationship is disrupted in parents whose infant receives an initial out-of-range newborn metabolic screening result and whether uncertainty surrounding the result is associated with reduced self-reported ratings of bonding for both mothers and fathers.	Not funded.

For a complete list of resources leveraged to date, please see Appendix B.

### **New Committee Launched - Advocacy Committee**

As a result of feedback gathered from the 2010 Annual Meeting, the NEGC brought together 13 individuals representing all New England states, as well as a range of professional and family interests. The group brainstormed a range of project ideas with the result of focusing directly on the concept of essential benefits under health care reform. Through a partnership with NH Family Voices, an application was developed and submitted to the NEGC Innovative Projects program. Although the effort was not funded, the group anticipates continuing to work on this area during Year 5 of the project as well as providing feedback to the NEGC on how it can best meet the needs of individuals and families.

### **Collaborative Activities**

Project staff continued to seek out new opportunities for partnerships with both regional and national partners. During Year Four, this included: joint planning of the annual meeting with NERGG, 11 presentations and 5 publications by affiliated staff, 10 training/technical assistance activities, 4 newly funded innovative projects, 3 applications supported through the community and family network grants, special

projects supporting the mission of the NEGC, and continuing collaborations with regional and national partners. The following outlines each of these accomplishments in more detail.

### **Annual Meeting, Dec. 1, 2010**

The annual meeting was well attended by 65 partners in the initiative, representing an increase of 19 participants over the previous year. Project staff highlighted the key accomplishments of the project over the course of the past year and identified major activities to be undertaken for next year. Breakout sessions on how to effectively engage with genetic counselors, families, and the New England Birth Defects Consortium were held. In addition, open workgroup meetings were held that enabled cross group and new stakeholder participation in the activities of individual workgroups.

Most participants found the meeting helpful, that they had opportunities to share their perspectives, that they had a good understanding of what the NEGC will accomplish in Year 5, and that the NEGC is “headed in the right direction.” More than half of responding participants felt that the work of the NEGC resulted in tangible outcomes resulting in improvements in high quality genetic services in the region. In terms of recommendations, participants highlighted a range of groups to which the NEGC staff could do additional outreach, including major medical centers, school nurses, family advocate groups, March of Dimes, Save our Babies, Medicaid leadership and other major organizations (AAP, AFP, ACM, CCPCMH). A copy of the full meeting 'mid-year' report is available at [http://www.negenetics.org/AboutUs/Evaluation\\_reports.aspx](http://www.negenetics.org/AboutUs/Evaluation_reports.aspx).

### **Presentations and Publications Supported by the NEGC**

During Project Year Four, NEGC coalition stakeholders conducted 12 presentations about project activities. This included an update on Long Term Follow Up (LTFU) activities in New England to the National Coordinating Center / Regional Center Annual Meeting as well as a short course on CF NBS and Care Quality Improvement at the the 2010 North American Cystic Fibrosis Conference. Both sessions were conducted by Dr. Anne Comeau. A third presentation was by Dr. Susan Waisbren during a conference on neurocognitive issues in PKU and transition to adult care. For a detailed listing of presentations and presenters to date, please see Appendix C.

By the end of Project Year Four, 5 additional publications were created by NEGC collaborative council members, bringing the total publications list of NEGC stakeholders up to 20<sup>4</sup>. The most recent publications include:

- ❖ McGrath RJ, Stransky ML, Cooley WC, Moeschler JB. *National profile of children with Down Syndrome: disease burden, access to care, and family impact*. J Pediatr. 2011; in press.
- ❖ Woo HC, Lizarda A, Tucker R, Mitchell ML, Vohr B, Oh W, Phornphutkul C. *Congenital hypothyroidism with a delayed thyroid-stimulating hormone elevation in very premature infants: incidence and growth and developmental outcomes*. J Pediatr. 2011;158(4):538-42.
- ❖ Sahai I, Eaton RB, Hale JE, Mulcahy EA, Comeau AM. *Long-term follow-up to ensure quality care of individuals diagnosed with newborn screening conditions: early experience in New England*. Genet Med. 2010;12(12 Suppl):S220-7.

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<sup>4</sup> The number of total publications has been reduced from last year due to updated definitions from national partners as to what constitutes an NEGC supported publication.

- ❖ Hale JE, Parad RB, Dorkin HL, Gerstie R, Lapey A, O'Sullivan BP, Spencer T, Yee W, Comeau AM. *Cystic fibrosis newborn screening: using experience to optimize the screening algorithm.* J Inherit Metab Dis. 2010;33(Suppl 2):S255-61.
- ❖ Fanos JH, Wiener L, Brennan T. *Potential impact of genomic information on childhood sibling relationships.* In: *Handbook of genomics and the family*, Issues in clinical child psychology, K.P. Tercyak (ed.), Springer Science, 141-61, 2010.

For a detailed listing of publications supported by the collaborative and its members, please see Appendix D.

### **Trainings and Technical Assistance**

Through its many collaborators and supporters, staff funded by the NEGC carried out a range of training and technical assistance activities to families, consumers of health services, health providers, education providers, state staff, community organizations, and others. Of the 3,309 aided in this manner, an estimated 3,000 individual contacts were made by the Birth Defects Consortium in its effort to improve understanding and utilization of folic acid during pregnancy. Additional areas of focus touched on: support around Down syndrome education, emergency preparedness, support for grant applications and conducting research, training professionals on assessment tools and transition practices, training staff on the use of learning collaboratives, database utilization for improving care, and the legal aspects of data use.

### **Innovative Projects**

The RCC continued to build on the innovative projects program and completed its fourth round of grant funding. The NEGC received 10 proposals and awarded four grants, with a combined total disbursement of \$86,985. The studies funded by these grants include:

- **"Exploring and identifying the knowledge level and attitudes of (selected) diverse populations toward genetics and genetic services,"** submitted by Patricia Rissmiller of Simmons College. Awarded: \$12,500.

Four focus groups were held to gain an understanding of how select diverse populations think about and discuss genetics and genetic services. One focus group involved 15 members of the Haitian Community Group and three groups were held at the Somalian development center, with 7-10 women participating in each group. Initial review indicates that, although participants had limited if any knowledge of the topic, they were eager to learn more. Next steps for this effort are to complete the analysis of the focus groups and to consult with the organizations to identify the most culturally relevant strategy to educate the community about genetics and genetic services. If successful, a long term goal will be to launch this effort on a broader scale via a community based participatory research grant.

- **"Implementation of the New England Birth Defects Consortium - Year 2"**, submitted by Stephanie Miller of Dartmouth Medical School. Awarded: \$30,000.

In addition to formalizing the Consortium via regular meetings and membership development, the Consortium collaborated with the Women, Infants, and Children departments in each of the six New England states to launch a multi-vitamin distribution campaign. As a part of this effort, the Consortium is conducting a pre and post survey study (ending 12/31/11) that will document the utility of a multi-vitamin distribution campaign to improve knowledge of folic acid use among

women. A second major effort of the group this year focused on determining the utility of combining data for 12 birth defects across participating states. Data for this effort was collected from Maine, Connecticut, Rhode Island and New Hampshire, with Vermont sending data in 2011. The NEBDC notes that it had a successful second year and plans to work together and maintain the integrity of the group in the future. It looks forward to working with the NEGC on future activities as needed.

- **"Integrative Community-Based Management for Adults with Sickle Cell Disease,"** submitted by Victoria Odesina of UConn Health Center. Awarded: \$15,000.

Two home care agencies (Masonicare Partners Home Health & Hospice and the VNA of South-Central Connecticut) were successfully recruited and became active participants in a multi-agency effort to provide integrated community based care for adults with sickle cell. Participants received educational information on integrative care models, clinical research ethics, and participatory research. During this first year of the project, partners identified a list of home health care needs for adults with SCD. Additionally, home care agencies received information on the services that would and would not be covered by insurance companies as well as the fees for other services that may be needed but not covered for this population. The group created an MOU and outlined a series of objectives for project year two which outlines the working relationship among partner organizations including their roles and responsibilities as part of their involvement in the Collaboration of Care for Adults Living with Sickle Cell Disease. The overall goal of the collaboration will be to improve the home health care and quality of life for adults with SCD with a specific focus on: appraising home services needs of adults with SCD, facilitating home care management prescriptions for clients and providers, demonstrating a seamless sharing of client health information concerning acute, chronic care and coordination needs, maintaining or increasing community social supports and providing prevention and health education services for improving functional status and health related quality of life indicators, and examining the cost effectiveness of home health care on ER visits, day treatment or inpatient care.

- **"Increasing Access to Care for Newborn-Screened Children with Fatty Acid Oxidation Disorders,"** submitted by Dr. Susan Waisbren of Children's Hospital, Boston. Awarded: \$29,485.

Dr. Waisbren focused on improving access to psychological and developmental evaluations for patients with fatty acid oxidation disorders (FAODs) and to improve education about the developmental and psychological issues in FAODs.

The project identified 41 patients with FAODs and focused on conducting 21 interviews with parents of children with MCADD. In addition to in-person interviews, parents completed questionnaires assessing overall functioning, emotional well being, and executive functioning. Four families of older children were contacted to assess school outcomes and medical records were reviewed on an additional 20 children with other FAODs to examine results from the parent questionnaires and developmental and neuropsychological testing.

Of note, the study's results suggest that children with FAODs may have emotional and learning issues that have not previously been recognized by assessments such as the Bayley Scales of Infant Development or tests of intelligence. Forty-four percent of children in the study had scores in the at-risk range on withdrawal scales and 33% had scores in the at-risk range on anxiety scales from the Behavioral Assessment System for Children. The potential implications of these factors were discussed particularly as they related to energy depletion and school and the decreased likelihood of

youth seeking out and obtaining needed nutrients throughout the day. It was also noted that teachers may not be aware of the disorder or understand the symptoms that children may experience.

In response to these findings, the project developed the MCADD Educator's Guide to help teachers better understand the disorder and prevent problems related to an FAOD from occurring. The Guide will be posted online at: <http://newenglandconsortium.org> and at [www.negenetics.org](http://www.negenetics.org). In addition, the guide will be featured on the GEMSS website, developed by the DEM workgroup, upon its completion.

For final reports submitted by each of the above grant recipients, please visit the NEGC website at: <http://www.negenetics.org/innovative.html>.

### **Community and Family Network (CFN) Grants**

During 2010, community and family level innovation and participation in genetic services was supported by three CFN grants. Five applications were submitted and three were approved. The awardees include:

- National Tay-Sachs & Allied Diseases Association (NTSAD). \$2,500 was provided to support the development of a new web site ([www.ntsad.org](http://www.ntsad.org)), increasing access to comprehensive information for families & members, which would in turn increase donations and support for the organization.
- Maine Down Syndrome Network: \$2,500 was provided to help cover costs of presenters at their annual conference in November 2010. Presenters included: Susan Shapiro (“Friendship for all Kids: What to Do and What to Undo”), D. Kelley Young (“Estate Planning: Thoughts and Concerns for Parents”), and Dr. David Stein (“Behavioral Problems and Behavioral Interventions In Children with Down Syndrome”).
- Alzheimer’s Association, CT Chapter: \$2,460 provided to support the keynote speaker (Dr. Robert Green) on the genetics of Alzheimer’s Disease, at their annual conference in November 2010.

### **Special Projects**

Throughout the year, the NEGC engaged in several unique projects to improve the field of genetics education and services. During Year Four, the NEGC supported work in the following areas: assessing genetic workforce capacity, reviewing emergency preparedness protocols in New England, and clarifying state rules for information use around newborn screening.

#### *Assessing Genetic Workforce Capacity*

Led by Robert McGrath, Ph.D. from UNH and in partnership with the American College of Medical Genetics, this project reviewed the newborn screening process in five New England states and held a series of key informant interviews to document care processes for selected patients. Much of the focus of this work was on understanding the complexities that can hinder the care process as well as the strategies adopted to address these barriers. As a result, the project documented that the newborn screening processes, while different for each state, appeared to work well. In taking a close look at the care process following newborn screening, the authors highlighted three important theme areas worthy of further consideration: 1) reimbursement for genetic services was found to be particularly burdensome and often lacking; 2) there continues to be a need for improved care coordination - the authors suggest that new models should be

explored which incorporate different roles and approaches among team members/care providers; and 3) the need for more coordinated approaches to education - not just on how to provide effective educational resources to families, but also how to ensure that all components of a care team are knowledgeable of the different roles and needs of each element of the care process. Overall, the study raises the concern that the field is ill equipped to accommodate future growth in NBS conditions, particularly given potential shortages in supply of genetic providers. They argue for considering a shift in approach that takes into account where the whole health system is moving and how best to incorporate this information within a flexible dynamic of care provision (e.g. rethinking how care teams are structured, how medical homes and care coordination function for rare conditions).

### *Emergency Preparedness in New England*

On April 1, 2011, Dr. Roger Eaton launched New England's first Emergency Preparedness Symposium. The event included 23 participants, with representatives from NBS community labs, state and federal emergency preparedness contacts and two consumers. Presenters included Stan Berberich, Hans Andersson, and Bill Perry. Participants learned about what worked in other states, identified federal resources that would be helpful, and identified a range of action steps to pursue. Recommended steps to pursue included: create a regional group to conduct further planning and implementation, identify clear cut protocols, prepare hospitals for management of serious cases, improve utilization of electronic records/communication, develop formalized agreements, create a website to facilitate communications, develop a useful checklist for families, and provide funding to family-to-family health centers to train and strengthen consumer networks.

### *Use and Disclosure of Genetic and Newborn Screening Information*

In October 2010, Michelle Winchester completed her work analyzing the many differences in state approaches for using and disclosing genetic and newborn screening information for the purposes of treatment, a registry, and research. The report provides an overview of relevant guiding policies, addresses state variations in the use and disclosure of PHI for quality improvement, registry or research activities, documents relevant state laws, and offers a range of considerations for the NEGC to pursue as it seeks to improve the coordination and use of information for improved care in the region.

## **Collaborations with Regional and National Partners**

This section provides documentation on the affiliations held by NEGC management and collaborative council members.

### *Supporting the National Coordinating Center*

The NEGC has representatives in each NCC Work Group:

- Telegenetics Work Group: Rosemarie Smith, MD
- Emergency Preparedness: Roger Eaton, Ph.D.
- Long Term Follow-Up Workgroup: Anne Comeau, Ph.D.
- Evaluation: Peter Antal, Ph.D.
- Publications: Monica McClain, Ph.D.
- National Transition Workgroup: Susan Waisbren, Ph.D.



- Medical Home Work Group: Carl Cooley, MD

*Collaboration with Other Regional and National Groups*

- Genetics and Metabolism Psychology Network: Susan Waisbren, Ph.D.
- National Coalition for Health Professional Education in Genetics (NCHPEG): Leah Burke, MD
- National Newborn Clearinghouse (Genetic Alliance): Leah Burke, MD
- National Health Care Transition Center: Susan Waisbren, Ph.D., Carl Cooley, MD
- Newborn Screening Translational Research Network
  - Clinical Centers Workgroup: John Moeschler, MD, Anne Comeau, Ph.D.
  - Bioethics Workgroup: Anne Comeau, Ph.D.
  - Laboratories Workgroup: Roger Eaton, Ph.D., Anne Comeau, Ph.D.
  - Effective Follow Up Workgroup: Roger Eaton, Ph.D., Anne Comeau, Ph.D.
  - Information Technology Workgroup: Monica McClain, Ph.D.
- New England Consortium of Metabolic Programs: Susan Waisbren, Ph.D., Leah Burke, MD
- Next Step: Susan Waisbren, Ph.D., Carl Cooley, MD
- Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
  - LTFU Sub-committee: Carl Cooley, MD, Anne Comeau, Ph.D.
  - Health Information Technology Workgroup: Roger Eaton, Ph.D.
  - Evidence Review: Anne Comeau, Ph.D.
- Vermont Children's Health Improvement Program (VCHIP): Leah Burke, MD

## WORKGROUP ACTIVITY IN YEAR FOUR

This section provides an overview description of each workgroup's activities during Year Four. Material presented is drawn from each group's year-end report to the NEGC, with minor edits to improve readability. For an across-the-board view of major highlights from each group, please see Appendix E. A record of when groups met during the course of the year is provided in Appendix F.

### The Quality Improvement Workgroup

The Quality Improvement (QI) Workgroup has nine members and is led by the NEGC's Principal Investigator, John Moeschler, MD. They met three times as a full group between September and November, 2010. During the project's fourth year, workgroup members focused their efforts on three major areas: 1) development of a legal framework that would allow for entry of protected health information into a quality improvement registry; 2) creation of web-based software that would house and facilitate development of electronic reports of patients involved in participating clinics; and 3) implementation of a quality improvement learning collaborative (QILC).

One of the group's first tasks for the year was to establish a legal framework that would enable the utilization of patient data across sites. While the group initially pursued creation of a Patient Safety Organization, substantive background research and discussions with partners resulted in the group dropping this effort in lieu of an approach based on establishing Business Associate Agreements between each participating clinic and the hosting data site. Currently 2 centers have established BAAs under HIPAA with UNH and Global Vision Technologies (GVT), with a third center pending. This arrangement allows entry of protected health information into the quality improvement registry and enables the cross-site sharing of non-identifiable aggregated information for the express purpose of improving patient healthcare processes and, eventually, group outcomes.

The second area of work focused on the creation of a web-accessible data-base. GVT developed and implemented the NEGC quality improvement data-base, and has provided modifications, as requested. Web access has been provided to seven genetics health care professionals (physicians and genetic counselors) at two clinical sites. Data for 186 patients have been entered and a first analysis has been accepted by the American Society of Human Genetics for an abstract presentation at the annual meeting in October 2011. For the third area of work, project staff implemented the first regional learning collaborative to address quality of care issues for individuals living with PKU or MCAD<sup>5</sup>. In addition to a series of planning meetings, 2 full face to face meetings were held and one support webinar was held. The first session of the QILC focused on: introducing participants to the QILC model and discussing some of its strengths and limitations; suggested revisions to the PKU and MCAD data collection forms, a review of work flow at each clinic site and how the introduction of the new data forms shaped clinic activity, as well as a brief discussion of the

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<sup>5</sup> for more detail on learning collaboratives, please see:  
<http://www.ihl.org/IHI/Results/WhitePapers/TheBreakthroughSeriesIHIsCollaborativeModelforAchieving+BreakthroughImprovement.htm>

patient registry for individuals with developmental delays. The webinar, held in March, offered participants an opportunity to begin sharing notes on the implementation process, learned more about the quality improvement registry and discussed possible linkages. Lastly, in April 2011, participants again met face to face to review some of the collected data and share what was learned. In general, participants agreed that the forms were fairly easy to implement on an ongoing basis and several reported unexpected benefits as a result of form implementation. These benefits include:

- helping a clinic to address the multiple issues that they want to address during a patient's visit
- generating information that is important for care by complementing other care protocols
- supporting quality care by having critical information in one spot and having access to this information over time
- improving standardization of care
- helping to see overlap in clinic tasks and seeing each person's role in the transition process.

## The Transition Workgroup

The Transition Workgroup is led by Dr. Susan Waisbren, who is also the leader of the National Transition Work Group. The group currently has 18 members. The primary role of the regional Transition Work Group is to improve access to transition resources, implement innovative models for transition leadership among youth, and to enhance integration of transition practices into the activities of partner organizations. The group has solidified due to working together over time in a number of ways. In Year Four they developed strong collaborations with the National Health Care Transition Center (NHCTC) in NH and Next Step in MA. These innovative partners have been planning their activities and consulting with each other in the process of creating a community with shared goals. Specifically, NHCTC, led by Dr. Carl Cooley (Chair of the NEGC Medical Home Work Group), began conducting transition Learning Collaboratives and launched the *Got Transition* website. Next Step and Children's Hospital Boston planned a summer conference with expanded outreach for young adults (see details about *Face Forward* below). Bill Kubicek, Executive Director of Next Step and member of the Transition Work Group, sits on the NHCTC Advisory Board. Ann Walls and Mallory Cyr, both with NHCTC, joined the Transition Work Group. Ms. Cyr was also a key facilitator of *Face Forward*.

### *Improving Access to Resources*

At the end of Year Three, the group had produced the Transition Toolkit, including both one page educational fact sheets (*Metabolic Basics*) and the tool itself (*Transition Plan*). Usage reports at the end of Year Four were discouraging in that the tool wasn't used as much as hoped. The *Metabolic Basics*, however, were viewed quite often and for longer periods, suggesting real and meaningful benefits. Further, a physician in the work group shared that he valued the sheets for their portability and regularly advised families to use them. The group discussed changing the target audience to providers, who could then steer families to the site.

### *Developing Young Adult Leaders*

The Transition work group continued to address the need for leadership training for teens with metabolic and genetic disorders through *Teen Challenge 2010*. This three day camp for young people, aged 13-20, was designed to build confidence, strengthen bonds, challenge comfort zones and develop some of the skills needed to manage complex health conditions. 2010's Teen Challenge was held July 7th - 9<sup>th</sup> with nine young people in the rural setting of the Friendly Crossways Youth Hostel in Harvard, MA.

Another area re-examined by the group was *how* to engage and empower youth and young adults – an essential ingredient in a successful transition. Working with Next Step and modeling their strategies, the work group identified *functional outcomes* (getting a car, going on a date, etc.) rather than *health outcomes* as meaningful motivators. Future efforts may focus more on how to subtly help young adults make the link between being healthy and reaching their other goals.

This focus became the premise for Face Forward Summer Conference for Youth, jointly sponsored by Children’s Hospital Boston and Next Step. The conference was held at the start of Year Five, with planning underway during Year Four. Other modifications made to heighten the success include: 1) creating a youth council to plan the agenda; 2) including young adult facilitators; 3) changing the age range to 16-24; 4) stretching it to four days, and 5) including participants with other conditions.

### *Collaborating with Partners*

Members of the Transition Workgroup are integrated with the New England Consortium of Metabolic Programs, regularly touching on transition issues over the course of the year, as well as meeting in person at their annual meeting in November 2010. They help drive transition activities in the Consortium by dissemination of information about the Teen Challenge and the tools on the website.

Since December 2009, the Transition Work Group has been holding joint meetings with the Medical Home Work Group chaired by Dr. Carl Cooley. They met again in person at the NEGC annual meeting in December 2010. Dr. Waisbren and Dr. Cooley took initiative by bringing in two young adult presenters who helped participants come to a better understanding of the youth perspective in engaging the health care system for genetic conditions. They also held a joint conference call in May 2011. Five new members joined the Transition Work Group in time for this call: a metabolic specialist, a genetic counselor, a parent, a young adult with a genetic condition, and the project director of the NHCTC. The thrust of the call was a robust conversation on how to tell if a person has made a successful transition. Dr. Cooley noted that, other than information from adult providers, it is difficult to find evidence of research regarding transition outcomes; sharing characteristics of successful transitions may foster resilience in individuals and families.

Dr. Waisbren also continues to co-chair the National Transition Work Group, which was recently given “Work Group” status by the National Coordinating Center in recognition of their efforts and their charge. The group holds monthly calls regularly attended by members of the Transition Work Group, including the NEGC’s Monica McClain and Karen Smith. There are generally over 12 people on the call, with 3 or 4 from New England. The primary role of this group is to provide a forum for sharing current transition activities across the nation and discussing new directions for the field. Highlights from the discussion during the past year include a better understanding of the challenges inherent in developing transition programs (especially with locating adult specialists), the need to engage the youth in planning programs, the value of including individuals with various conditions as well as separate break-out sessions when developing programs, and being honest about what doesn’t work (written transition plans posted on a website rather than introduced by a provider).

### **Medical Home Workgroup**

During Year Four, the Medical Home Workgroup was led by Dr. Carl Cooley. The 16 member group held 2 formal meetings.

In Year Four, the group was initially poised to further test a communication tool among specialists, families, and the primary care medical home. This tool, or care plan, had been conceived and developed in previous years with the help of Dr. Chris Stille, formerly of UMass Memorial, and with support from the NEGC.

The care plan itself was a one page form in a fillable PDF format. The purpose of the form was to aid communications between the parent and doctor concerning what has been done and what concerns and requests should be related. Unfortunately, the group concluded at the end of Year Three that although the tool itself was deemed useful and user-friendly, it still wasn't being *used* in practice settings for a number of reasons. One of those reasons, noted by a specialist, was that much of the information was already contained in a letter that was routinely sent from her clinic to her patients' primary physicians. This led Dr. Cooley to wonder what other communication methods were already in use.

To explore this question, Dr. Cooley collaborated with the UNH Survey Center to develop the **Survey of Primary Care Clinicians Regarding the Care of Children with Rare and/or Complex Conditions**. The goal of this survey was to assess primary care clinicians' comfort, clarity of role, and quality of communication in the co-management with specialists of children and youth with rare and/or complex chronic conditions. Dr. Cooley defined rare or complex conditions as those that occur in less than .1% of children (rare) or that involve two or more body systems (complex) and require on-going medical management of some kind. While the conditions are individually uncommon, the aggregate of conditions of this type accounts for 5 - 7% of children and youth and includes children who are the highest users of health care services.

The survey was sent to email lists of the NH Pediatrician Society (275) and NH Academy of Family Physicians (604). 115 primary care providers responded to the survey. Further analysis of the survey results will occur in Year Five. Also in Year Five, Dr. Cooley will conduct a targeted survey/interview of genetic and metabolic clinics in New England, looking at the same essential question from that point of view, and collaborate with NEGC staff on a national survey of genetic and specialty clinics. Preparation for that work was completed in Year Four.

Since December 2009, the Medical Home Work Group has been holding joint meetings with the Transition Work Group chaired by Susan Waisbren, PhD. They met again in person at the NEGC annual meeting in December 2010. Dr. Cooley and Dr. Waisbren took initiative by bringing in two young adult presenters who helped participants come to a better understanding of the youth perspective in engaging the health care system for genetic conditions.

The two work groups also held a joint conference call in May 2011. Dr. Cooley provided an update on the National Health Care Transition Center (NHCTC), a separate MCHB/HRSA-funded project he has been leading since July 2010. He also updated the group on a clinical report, *Supporting the Health Care Transition From Adolescence to Adulthood in the Medical Home*. This report, published in *Pediatrics* in July, 2011, was prepared by the AAP, ACFP and ACP and Dr. Cooley was a lead author. One major component of the NHCTC has been to conduct Learning Collaboratives on transition, using to some extent the algorithm for a smooth transition outlined in the new clinical report.

Dr. Cooley was selected to chair the National Coordinating Center's (NCC) Medical Home Work Group. This committee met in person in January 2011 in Year Four and held monthly conference calls. The primary role of this group is to develop a common set of definitions and principles across all seven regional genetics collaboratives related to the medical home and the coordination of care among specialists, primary care physicians, and families. Highlights of the discussion during the past year include arriving at a common understanding of medical home, considering models of care coordination and communications between specialists and primary care, and acknowledging the crucial role of families in the process. The workgroup intends to produce a white paper related to its deliberations.

### **Dissemination, Education, and Marketing Workgroup**

Leah Burke, MD, chairs the Dissemination, Education and Marketing (DEM) Work Group, which currently has 12 members. The group met five times during the year, one of which was in person at the NEGC annual meeting in December 2010.

In Year Four the DEM work group continued to work toward completion of an interactive website for elementary school teams, a guide for the classroom for children with genetic conditions. This work builds on the group's efforts of the previous two years in which they primarily assessed the need for education about genetic conditions among potential audiences and the best way to provide it, and conducted focus groups with special educators to vet the project. In Year Four the group focused on 1) working with a web design vendor and 2) developing content for the website for the initial five conditions: PKU, Sickle Cell disease, Fragile X, Fatty Acid Oxidation Disorders, and 22q Deletion. In Year Five they will pilot, disseminate, and expand this new resource for additional conditions.

Working with a vendor to develop the design was a multi-layered process. The goal was to present unfamiliar and complex information in a simple manner that was compelling to educators ranging from paraprofessionals to specialists. How the information was displayed was as important as the content itself, as this impacts whether or not the website will actually be used. The group's original concept of *starting* with information teachers might find most relevant in the classroom – “challenge areas” (i.e. child has pain, field trips, etc.) – and then *linking* to more in-depth information about genetics, became concrete within the web design.

An important task was to finalize the name and URL address with a simple and easy to remember URL to increase usage. The group ultimately chose the following name: ***GEMSS: Genetic Education Materials for School Success*** and [www.gemssforschools.org](http://www.gemssforschools.org). They also developed a concept for the logo which will be finalized in Year Five.

The DEM work group was fortunate to collaborate with Christine Giummo, Certified Genetic Counselor from the University of Vermont, to write content for the website. Ms. Giummo used her expertise to address each challenge area for each of the five selected conditions. Challenge areas included: 1) Dietary and Medical Needs; 2) Special Education Supports; 3) Behavior/Sensory; 4) Field Trips/Special Functions; 5) Absences/Fatigue; 6) Emergencies; 7) Additional Considerations. Ann Dillon, a special education specialist from the Institute on Disability at UNH helped to simplify the language. Ms. Giummo also compiled links to helpful resources.

Focus groups previously vetted the *need* for the information. The DEM work group also wanted to vet the information itself. Toward that end, they identified two condition-specific advocacy groups per condition that were willing and able to review the content. Some groups were larger, some smaller; some were regional and some were national. One organizational director is a member of the DEM work group, and another is on the NEGC Advisory Committee. These new partnerships will strengthen the validity of GEMSS.

In addition to the special education project the DEM group screened an additional resource for the NEGC website, the Genetics and Rare Diseases Information Center (GARD). GARD was determined to be not user friendly and therefore not recommended for inclusion in the NEGC website.

One final ongoing project, the Newborn Screening Clearinghouse, occurred in conjunction with the Genetic Alliance and their collaborative agreement with HRSA. Dr. Burke participated on this Materials Committee and the National Advisory Council to recommend resources and represent the needs of our region.

### **Laboratory Quality Assurance Workgroup**

The New England Newborn Screening Program (NENSP) has developed algorithms to categorize tandem mass spectrometry (MSMS) results to better discriminate between false positives and true cases, improve the clarity of communications to the medical home, and to better target the use of scarce specialty care resources. To be useful for application to regions outside of the NENSP, the universality of these algorithms must be proven in a robust manner by application to independent data sets. The project proposed to apply these algorithms to data sets independently derived by the newborn screening labs in Connecticut, New York, and Wisconsin. During year four, CT informed us that it was unable to continue participation in the project due to internal reasons.

In prior years of the project, concentrations of all relevant MSMS markers were collected from CT, MA, NY, and WI, for all specimens with out-of-range values for the following markers: C3, C5, C14, C14:1, C14:2, C16OH, C18:1OH, C16, C18:1. During year four, analogous data were collected from MA, NY, and WI on all remaining markers with relevance for newborn screening (C5OH, C5:1, C5DC, Leu, Cit, and Arg). Drs. Sahai and Eaton analyzed these data according to indices and cutoffs developed at UMass. We then compared the index-based categorizations to actual follow-up for the following disorders: 3MCC, BKT, GA-I, MSUD, CIT-I, ASA.

Some indices could not be applied directly for categorization since some markers utilized in the index were not tested by all laboratories. In such cases additional indices were created and cut-offs for these were established based on the site-specific population statistics. These markers were C3, Cit, and Arg. The categorizations were very effective for most markers but not as useful for one marker in particular. For example, markers that showed universal applicability included C16, C18:1, C16OH, C18:1OH, C5, C5:1, C5DC, Leu, C14:1, and C14:2. The marker that was less effective was C5:OH. An unexpected finding from analysis of the raw data from partner laboratories was that a significant number of specimens had “0” concentrations for some markers. In such cases we utilized the “multiples of the mean” (MOM) while evaluating profiles and establishing cut-offs.

Use of web-based conferences utilizing Acrobat Presenter introduced in Year 3 was utilized for sharing of excel and PowerPoint files during web conferencing with all partners.

A summary of findings to date was presented at the Laboratory Subcommittee of the SACHDNC Meeting, in May 2011. The New England Newborn Screening Program has already begun accompanying newborn screening lab reports with fact sheets to the medical home that reflect the categorizations of this work. It is anticipated that other collaborators will make similar use of these indices when the work is completed and confirmed. The collaborators plan to submit this work as a detailed publication by the end of the grant period, which will make available this approach to screening programs of all states and countries.

### **Long Term Follow Up Workgroup**

The Long Term Follow Up (LTFU) Workgroup (NENSP and New England state NBS coordinators) is led by Anne Comeau, Ph.D. and has nine members. During Year Four, they continued to focus on issues surrounding interstate data sharing and operating principles relevant to our current and future regional LTFU system. States continued to make progress in moving forward with both establishing the authority to collect LTFU data (MA, ME, RI in particular) and actual data collection (MA and ME and RI). Work on the development of IT systems for protected state-specific LTFU data that are compatible with updates to the NENSP core data system has begun. Meetings of “condition” specific NBS workgroups have also continued over the course of the year in order to engage specialists caring for infants and children diagnosed with newborn screening conditions to develop and refine data collection tools and variables.

The LTFU Workgroup held a meeting as a part of the 2010 NEGC Annual Meeting in December 2010. Dr. Inderneel Sahai presented LTFU data on children diagnosed with long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD) by NBS. LTFU (age range 2-10 years) revealed that while some cases remain asymptomatic, others had associated clinical findings such as recurrent biochemical abnormalities, mild language delays, muscle pain on exertion, and retinal abnormalities after 3 years of age. This project will be formally presented by Dr. Sahai at the upcoming 2011 NBS and Genetic Testing Symposium. In Spring 2011, under Dr. Sahai’s direction, a second project to evaluate the long term metabolic outcomes of children identified with Short chain acyl-CoA dehydrogenase (SCAD) began.

Dr. Anne Comeau presented Massachusetts data as a part of the CF NBS and Care Quality Improvement Short Course at the 2010 24th Annual North American Cystic Fibrosis Conference held in Baltimore, MD (October 21-23, 2010). The Massachusetts CF NBS Workgroup began discussions and design of a new LTFU project with data collection to begin in late 2011. This project will address outcomes of a subset of patients identified by CF NBS over the course of 10 years in order to enhance the development of follow up and best practices for this particular subset of children.

The Hemoglobin Workgroup continued to focus and build upon their LTFU activities. In September 2010, the group hosted a successful conference, “Surviving to Thriving: Improving Long-term Outcomes in Sickle Cell Disease.” The conference was attended by over 100 people and brought together experts from around the county to identify best practices for improvements to patient care. Ms. Claire Hughes of NENSP and Dr. Philippa Sprinz of Boston Medical Center also attended the International Public Health Learning Collaborative on Hemoglobinopathies meeting held in Atlanta, GA (November 3-4, 2010). This meeting focused on the integration of public health and clinical practice as related to hemoglobinopathies.



Dr. Anne Comeau continued to represent the LTFU workgroup at national forums including two LTFU subcommittee meetings of the SACHDNC (January 27-28, 2011 and May 5-6, 2011), the Clinical Centers Workgroup Meeting of the NBSTRN (October 14-15, 2010), and at the NBSTRN Effective Follow Up PP's and Partners meeting (March 28-30, 2011).

#### *Focus on Long Term Psychosocial Follow-Up of Newborn Screening*

In Year Four, Dr. Waisbren collected data on children with the Uniform Screening Method. The Uniform Screening Method consists of three instruments to assess development, each of which can be administered by non-psychologists (parents) and computer scored and interpreted:

1. Adaptive Behavior Assessment System-Second Edition (ABAS-II, for infants and adults)
2. Behavior Rating Inventory of Executive Function (BRIEF, for preschoolers to adults)
3. Behavior Assessment System for Children-Second Edition (BASC-2, for preschoolers to adults)

Dr. Waisbren gave a presentation on her work in April, 2011, to the International Neuropsychological Society. Dr. Waisbren had collected data on 30 cases (MCAD, PKU, and Galactosemia). She included data on children who would have been missed for referral, noting that the Adaptive Behavior Assessment System was shown to be 94% accurate.

In the process of conducting the work, Dr. Waisbren noted that there were challenges (including insurance company coverage for screenings) in getting metabolic clinics to participate.

During Year 5, Dr. Waisbren plans to promote the use of the Uniform Screening Method via the websites of the NEGC, the NE Consortium, and the Psychology Network. In the future she will build on her work by piloting the Method at two centers, one of which will be Children's Boston. The goal is to have a "shovel ready" vetted instrument that has buy-in from potential users.

#### *Posttraumatic Stress Disorder (PTSD) and Newborn Screening*

Dr. Joanna Fanos' work focused on understanding how best to identify the need for additional supports to parents who are notified, post newborn screening, that their child has a serious illness. The need for this project centered on the fact that several features of the disorder (re-experiencing, avoidance, numbing and hyper-arousal) could impact medical care of the child as well as cause difficulties for the family, including well siblings. Dr. Fanos' team conducted an extensive literature review and held multiple informational interviews to identify appropriate scales and to develop a recommended protocol following a negative outcome for newborn screening. The team recommended use of the Breslau scale<sup>6</sup> (a seven item, Y/N response questionnaire) as an initial screener for parents. Parents who screen positive on the scale should complete either an Adult Self Report Scale<sup>7</sup> or an Adult Interview<sup>8</sup> in order for a more rigorous assessment to be made concerning the need for additional supports.

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<sup>6</sup> Breslau, N., Peterson, E.L., Kessler, R.C., Schultz, L.R. Short screening scale for DSM-IV posttraumatic stress disorder. *American Journal of Psychiatry*. 1999 156:908-11.

<sup>7</sup> Recommended scales include: Davidson Trauma Scale (DTS), Distressing Events Questionnaire (DEQ), Impact of Events Scale-Revised (IES-R), Los Angeles Symptom Checklist (LASC), Modified PTSD Symptom Scale (MPSSSR), Penn Inventory for Posttraumatic Stress Disorder (Penn Inventory), Posttraumatic Diagnostic Scale (PDS), PTSD Checklist (PCL), Screen for Posttraumatic Stress Symptoms (SPTSS), Trauma Symptom Checklist-40, and Trauma Symptom Inventory (TSI).

## Ethical, Legal and Social Issues

Through its multiple endeavors, the NEGC seeks to address relevant public policy and ethical, legal, and social issues (ELSI) affecting individuals with genetic conditions, their families, and health care providers and educators. During Year Four, the following areas were addressed:

### Ethical Issues

Work around the creation of a central data resource to improve understanding of patient services raised many discussions around the appropriate use of data for improving health care quality. During Year Four, NEGC staff met with a range of national, regional, and state level providers in order to ensure appropriate steps were taken to safely manage and appropriately utilize patient information. Additionally, in preparing for its Face Forward Summer Conference for Youth, members of the Transition Workgroup took an important step in addressing the role of youth participation in program development by creating a youth council that became actively involved in planning the event.

### Legal Issues

Advances in this area continued with the QI workgroup's exploration of forming a patient safety organization and subsequent pursuit of BAA agreements among participating clinics as well as LTFU work to solidify an agreement with the State of Rhode Island in Year 4 to pursue long term data tracking. The Birth Defect Consortium pursued integration of data on birth defects from Maine, Connecticut, Rhode Island, New Hampshire, and Vermont. Dr. Eaton's work to launch an Emergency Preparedness Symposium explored some of the initial areas to address in developing a regional emergency preparedness plan. Lastly, Michelle Winchester completed her work analyzing differences in New England state laws concerning the use of protected health information from newborn screening for quality improvement and research purposes<sup>9</sup>.

### Social Issues

The NEGC impacted the social context of healthcare for individuals with genetic conditions in a variety of ways. Joanna Fanos published her work on the impacts of genomic information on childhood sibling relationships. The Birth Defects Consortium outreached to WIC centers in the New England States to educate groups about the importance of folic acid. Patricia Rissmiller worked to expand our understanding of genetic awareness among minority populations, and Victoria Odesina laid the groundwork for improving home health care and quality of life for adults with SCD. Dr. Waisbren created the MCADD Educator's Guide to help teachers better understand the disorder and prevent potential problems from occurring. The NEGC supported the development of the National Tay-Sachs and Allied Diseases Association website. Lastly, the work of the DEM group to create an online resource for special educators will result in a very valuable information resource that will be made available to educators across the nation.

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<sup>8</sup> Recommended interviews include: Clinician-Administered PTSD Scale (CAPS), PTSD Symptom Scale–Interview (PSS-I), Structured Clinical Interview for the DSM-IV Axis Disorders (SCID PTSD Module), and Structured Interview for PTSD (SI-PTSD)

<sup>9</sup> A copy of the report is available here:

[http://www.negenetics.org/Libraries/Ongoing\\_materials/State\\_Laws\\_of\\_NE\\_Use\\_Disclosure\\_of\\_Genetic\\_NBS\\_Information\\_Winchester\\_Oct\\_2010.sflb.ashx](http://www.negenetics.org/Libraries/Ongoing_materials/State_Laws_of_NE_Use_Disclosure_of_Genetic_NBS_Information_Winchester_Oct_2010.sflb.ashx)

## **Policy Issues**

As a result of LTFU work in Rhode Island, the ability to share LTFU data has now been formally established. Along with Massachusetts and Maine, this will continue to have multiple positive impacts on the ability of the Newborn screening program to track health outcomes, identify best practices, and ultimately improve the care of individuals with genetic conditions. NEGC's support of partner's work, either through letters of support or direct funding of innovative grants, led to a range of improvements impacting the policy arena. Support for the American Academy of Pediatrics will help to ensure better integration of genetic medicine practices among primary care providers. New research was published creating a national profile of children with Down syndrome, and Joanna Fanos completed her recommendations for brief and more in-depth mental health screens for parents receiving a medical diagnosis for their child. Dr. Sahai published some of the early experiences of long term follow up efforts to ensure quality of care of individuals. Lastly, work on Assessing Genetic Workforce Capacity continued in Year 4 and resulted in several policy recommendations.

ELSI issues will continue to be reviewed annually by the NEGC Advisory Council which will advise on potential new directions for the collaborative to pursue.

## **Evaluation Activities**

During Year Four, the evaluation of the NEGC was led by Peter Antal, Ph.D. Primary roles during the fourth year of the collaborative focused on: providing ongoing review of activities, summarizing project activities via evaluation reports, promoting coordination with the national evaluation initiative, and conducting the third annual stakeholder survey.

## **Ongoing Review of Activities**

Peter Antal has actively participated in NEGC meetings, including ongoing planning meetings and meetings with the Quality Improvement Learning Collaborative, Collaborative Council, Advisory Council, as well as monthly meetings with the principal investigator, project manager, and workgroup chairs. The focus of his participation in these meetings is to provide historical context to guide decision making, technical support in areas of research, and suggest areas of follow-up by staff.

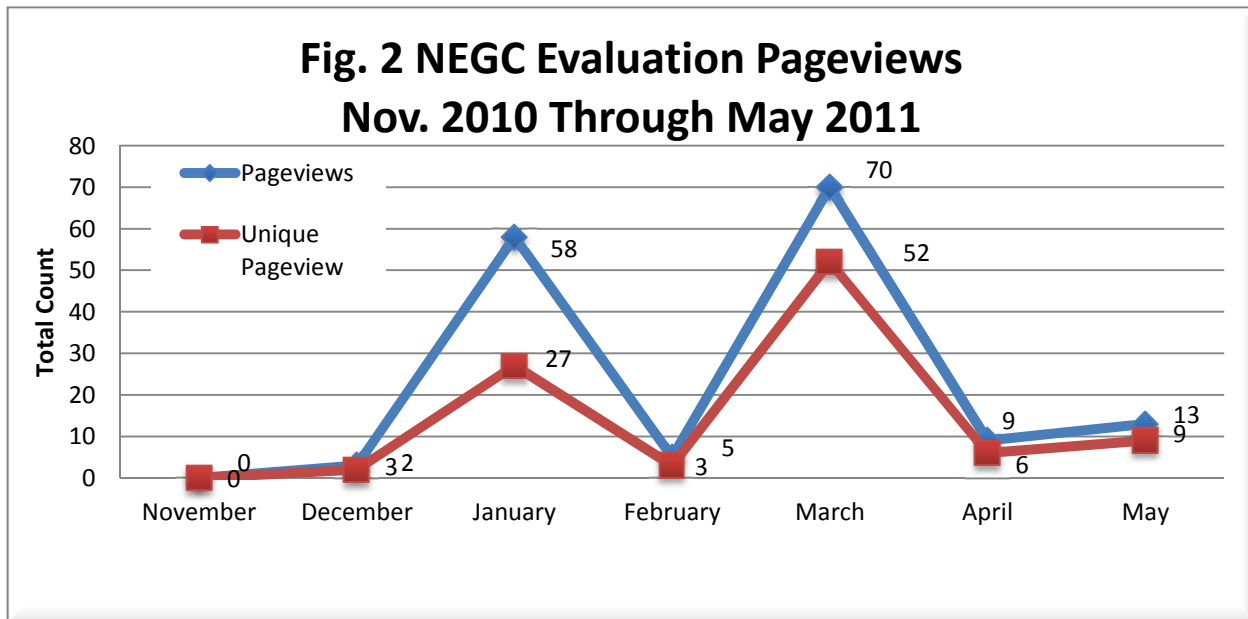
## **Evaluation Reporting**

During the past year, the Year Three Project Report and Year Four Annual Meeting report was posted as was the Year Three Stakeholder survey. All reports were provided to staff for review and feedback prior to final publication as public documents on the project's website. Reports include:

- ❖ Results of Stakeholder Survey for Project Year 3: Dec. 2010
- ❖ Annual Evaluation Report for Project Year 3: Dec. 2010
- ❖ Summary of the Year 4 Annual Meeting: Feb. 2011

In addition to the above reports, Dr. Antal also supported HRSA's strategic planning process by conducting a survey and a facilitated discussion with other regional collaboratives involved in improving genetic services. The summary of recommendations can be found in the document, Strategic Planning Recommendations for HRSA: Mar. 2011, available on the National Coordinating Center's website ([www.nccrcg.org](http://www.nccrcg.org)).

As shown in Figure 2 below, online views of evaluation material peaked in January (27 unique views) and March of 2011 (52 unique views).



### National Benchmarks

Dr. Antal has continued to represent the New England region by monitoring and updating national benchmarks for regional genetics programs. Dr. Antal has participated in conference calls and has regularly solicited input from NEGC staff on key issues leading to their creation and utilization. Reporting for the national benchmarks is based on regional activities between Dec. 1, 2009 and Nov. 30, 2010. Results are provided below:

- ❖ Outcome Measure A1: Increase in the percentage of states/territories in the region with collaborations facilitated by the Regional Collaborative between primary care providers (PCPs) and specialty (including genetic) providers to improve care coordination for people with heritable disorders.
  - Result: 100%. All New England states were involved in collaborations facilitated by the RC between PCPs and specialty providers. Examples of collaboration include:
    - Education and awareness building regarding medical home occurred through the NEGC annual meeting and the New England Consortium of Metabolic Programs annual meeting.
    - Collaboration between medical home workgroup and transition work group targeted care in both specialty and primary care settings. Dr. Susan Waisbren has used the Transition Plan documents with 10-15 patients and has posted these care plans on the New England Consortium of Metabolic Programs website. Additional material reviewed and posted to the website included: Acute Illness Protocols, Newborn Screening Guide for Prenatal Educators, Newborn Screening Prenatal Curriculum, Transition to Adult Care guide, Transition Plan, and many others.
    - In Massachusetts, Dr. Waisbren, via NEGC's innovative projects program, led the Personal Transition Health Plan Project at Children's Hospital Boston. The long-term goal of this project was to develop and pilot a practice model that ensured that every

adolescent and young adult patient seen at a genetics or metabolic clinic had thought about and documented a plan for on-going health care that addressed the specific needs of his or her specific condition, with a focus on symptoms that are relevant to adults.

- Dr. Burke attended the annual NCHPEG meeting as the American Academy of Pediatrics representative. She presented the data from our focus groups on the special educators' tool at that meeting. Dr. Burke reported back to pediatricians and pediatric clinical geneticists on the NCHPEG meeting presentations and initiatives.
  - NEGC's workgroups have begun integrating their efforts (Medical Home, Transition, and Quality Improvement). The medical home pilot project was implemented in 2010. Key staff (Dr. Carl Cooley, Dr. Chris Stille, Dr. John Moeschler, Dr. Wendy Smith) have collaborated with the annual metabolic consortium meeting and have laid the foundation for more integrated work with the NEGC.
  - Joanna Fanos' work on parent perspectives of the diagnostic and follow up process helped to identify a series of recommendations that can provide needed supports for families dealing with the challenges of a new diagnosis.
  - Dr. John Moeschler has been working with the QI workgroup on developing a standard set of quality improvement data points that will be collected by multiple clinic sites. Central to this effort is the researching and potential creation of a Patient Safety Organization (PSO) and/or data sharing business agreements that will enable the sharing of information among providers to aid in quality improvement efforts in the region.
  - Dr. Moeschler has collaborated with a planning team to create the initial framework and pilot tools for a new Quality Improvement Learning Collaborative serving the New England region.
  - Stephanie Miller of Dartmouth Medical School has formed the New England Birth Defects Consortium to facilitate project and data collection coordination among New England birth defect registry programs. The aim of the consortium is to improve services for infants and children with birth defects by promoting regional collaboration in surveillance data sharing, birth defects research, prevention activities and health care quality improvement.
- ❖ Outcome Measure B1: Increase in the number of genetic services visits and NBS follow-up specialty visits provided to individuals/families through distance strategies implemented by the regional collaborative.
- Result: NA. NEGC did not provide support for service visits through RC implemented distance strategies during this period.
- ❖ Outcome Measure C1: Increase in the percentage of states/territories in the region that have received current materials or other assistance from the RC on emergency preparedness/contingency planning for newborn screening (NBS) and genetic services.
- Result: 100%. Departments of Health / NBS programs in each of the states have received a New England Newborn Screening Program COOP Plan. Stakeholders were involved in the development of an Emergency Preparedness workshop held on April 1, 2011.
- ❖ Outcome Measure D1: Increase in the percentage of states/territories in the region that have evaluated and made recommendations on implementing the ACHDNC recommended NBS panel.
- Result: 100%. All states in the region have evaluated and made recommendations on implementing the ACHDNC recommended NBS panel. Note that this process is independent of NEGC activities for the reporting period. Only Massachusetts, Rhode Island, and Connecticut have evaluated and made recommendations on SCID.

- ❖ Outcome Measure E1: Increase in the percentage of states/territories in the region with systems in place to track entry of newborns into clinical management for newborns who are diagnosed with conditions mandated by their State-sponsored newborn blood spot screening programs.
  - Result: 100%. All states in the region have systems in place to track entry into clinical management for newborns diagnosed with conditions mandated by State-sponsored newborn blood spot screening programs. Note that this process is independent of NEGC activities for the reporting period.
- ❖ Outcome Measure E2: Increase in the percentage of states/territories in the region with systems in place to track entry into clinical management for newborns who are diagnosed with hearing loss through their State-sponsored newborn hearing screening programs.
  - Result: 100%. All states in the region have systems in place to track entry into clinical management for newborns who are diagnosed with hearing loss through their State-sponsored newborn hearing screening programs. Note that this process is independent of NEGC activities for the reporting period.
- ❖ Outcome Measure E3: Increase in the percentage (number) of states/territories in the region with systems in place to track receipt of clinical services and/or health outcomes for children who are diagnosed with condition(s) mandated by their State-sponsored newborn blood spot screening program and/or with hearing loss through their State-sponsored newborn hearing screening programs.
  - Result: 17%. Only Massachusetts meets the criteria of having a long term follow up system for all conditions in each area (metabolic, endocrine, hemoglobin, cystic fibrosis, and hearing) mandated by the State-sponsored newborn blood spot screening program. States in the region provide variable extents of long term follow up tracking for genetic conditions identified by NBS. The variety is dependent on the state and the particular condition. 5 of 6 states are working with the New England Newborn Screening Program (NENSP), following and modifying the model set in Massachusetts to ensure LTFU in a manner that allows quality assurance and quality improvements.
- ❖ Outcome Measure F1: Increase in the percentage of states/territories in the region whose NBS programs disseminate “just-in-time/point-of-care” information on specific heritable disorders to primary care providers (PCPs).
  - Result: 100%. All state NBS programs in the region disseminate information on heritable disorders to primary care providers. Note that this process is independent of NEGC activities for the reporting period.
- ❖ Outcome Measure G1: Increase in the percentage of Regional Collaboratives that have completed a regional genetic services plan.
  - Result: 100%. NEGC’s plan is outlined in its annual grant application to HRSA. The plan is tied to a series of objectives, action steps, timelines, and resources that are followed to carry out NEGC’s mission. The goals and strategies adopted by NEGC are reviewed and updated annually by the Advisory Council.
- ❖ Outcome Measure G2: Increase in the percentage of Regional Collaboratives that have reviewed and/or updated their regional genetic services plan at least every two years.
  - Result: 100%. The plan is reviewed on an annual basis by the project’s collaborative council, advisory board, and stakeholders. Initiatives proposed for 2011 were reviewed during the December, 2010 annual meeting of the collaborative. Multiple recommendations were provided on next steps for the collaborative during 2011 which the staff will review and incorporate as appropriate into grant activities.

## **NEGC Stakeholder Survey for Project Year Four**

For Year Four, the evaluator again worked with project staff to update and implement the NEGC stakeholder survey. The survey was administered online between October and November 2011. A summary of the results follows.

### *SUMMARY OF FINDINGS FROM THE YEAR FOUR STAKEHOLDER SURVEY*

*[Executive Summary excerpted from New England Genetics Collaborative, Results of the Partner Survey for Project Year Four by Peter Antal, Ph.D. (January, 2012). For the full report, please download from [http://www.negenetics.org/AboutUs/Evaluation\\_reports.aspx](http://www.negenetics.org/AboutUs/Evaluation_reports.aspx)*

To facilitate feedback from its stakeholders, the NEGC conducts an annual survey to identify concerns, document how the project is doing, and solicit suggestions for improvement. One hundred-forty-one email invitations were sent out between October and November 2011 to stakeholders of the New England Genetic Collaborative (NEGC). Of these, one opted out and 63 provided responses (45% response rate).

Since the 2009 report, there was improvement in two important areas. When asked whether they had a clear understanding of the NEGC's mission, 73% agreed (vs. 60% in 2009). Concerning whether the NEGC had made substantive and clear progress in achieving its mission, 72% agreed (vs. 47% in 2009). Feedback on the project's evaluation reports was generally positive with 67% to 70% of respondents indicating that each of the reports helped them understand the progress and challenges of the initiative (vs. 60% to 75% in 2009).

Feedback from the Advisory Council was high this year, with 13 members participating. Most participants (>75%) felt that there was a good spirit of cooperation, that meetings were well run, that the RCC provided excellent support and responded effectively to questions, and that the Advisory Council was achieving its main objectives.

Project recommendations highlight the need for continuing to strengthen communication efforts of the NEGC, identifying new collaboration opportunities for members of the Advisory Committee, making effective use of potential partner contributions, improving consumer/family representation in regional change, pursuing sustainable initiatives, addressing multiple barriers to care for families, and improving access to NEGC resources.

## **COMPLETION OF OBJECTIVES IN YEAR FOUR**

Table 3 provides a complete list of the objectives set forth by project staff at the beginning of the project year (with modifications based on changes in the project) as well as the status of each objective as of June, 2011. Measures of objective "status" relative to implementation over the course of the 5 year project are defined by the following key: 1. Completed as planned, 2. Completed - deviated substantially from plans, 3. In progress - satisfactory, 4. In progress - unsatisfactory, 5. Initiation of activity deferred, 6. Activity abandoned, 7. Not scheduled to initiate this period, 8. Insufficient documentation available. Additionally, a review is provided on the relative success of the objectives during Project Year Four. Review results are defined as:

- ❖ Successful (34 of 54): All definitions of success for an objective have been fully met or the results of the activity in question fulfill the intent of the measure.
- ❖ Partially Successful (18 of 54): Definitions of success for the year only partially met. Although not fully realized, substantive progress has been made in a number of core areas with fulfillment of the goal expected by the next project year.
- ❖ Unsuccessful (2 of 54): Although some work on an activity may have been done, primary components of an activity targeted for the year were not substantively addressed within the time period. Lack of success may be due to a number of factors, including lack of participation by certain groups, delays in timeline for other project components, and the need to shift project priorities such that other components could be fulfilled in Year Four.

**Table 3: Status of Goals and Objectives of the NEGC, Project Year 4**

<b>ESTABLISH AND MAINTAIN NEGC</b>				
<i>No.</i>	<i>Objective</i>	<i>Project Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	Continue implementation of core administrative supports to the NEGC	3	NEGC meets yearly objectives.	Review: Successful  All core staff activities completed during course of year.
2	Continue close collaboration with WG and AC	3	Work Group and Advisory Committee members feel supported in the work they do and have access to the resources they need to accomplish their goals.	Review: Successful  Meetings are held regularly and supports provided when requested as resources allow. 89% of Stakeholder Survey respondents from the Advisory Committee indicated that the RCC provides excellent support.
3	Develop and implement a communications and outreach plan for the NEGC	3	Stakeholders report satisfaction with being able to voice their opinions and feel that they've been heard.  A majority of stakeholders understand the NEGC's mission and the steps it is taking to achieve that mission	Review: Successful  A majority (74%) of participants at the 2010 annual meeting felt they had an opportunity to share their perspective.  73% of respondents of the Year 4 Stakeholder survey felt that they had a clear understanding of NEGC's mission and steps it is taking to achieve that mission.



			New groups and individuals are represented on the NEGC stakeholder list.	Review: Successful Between Yrs 3 and 4, participation of stakeholders (defined by mailing list) increased from 75 to 140.
4	Maintain, update and enhance NEGC website	3	The NEGC stays current with state, regional, and national level developments.	Review: Successful Website is maintained and updated continuously.
			Stakeholders have information necessary to keep informed of all project developments.	Review: Successful Stakeholders received 3 quarterly updates, mid-year report, and an annual report describing project progress.
			Website is utilized by growing numbers of individual users.	Review: Successful Starting Nov.2011 with the new website, unique users increased from 28 to 254.
5	Implement Special Projects	3	Genetic Workforce study	Review: Successful Research and analysis completed during Year 4 for the New England region and recommendations provided for improvement of the workforce.
			Emergency Preparedness Conference	Review: Successful Event held April 1, 2011 with 23 participants.
			Launch Advocacy Committee	Review: Successful Group met for the first time on May 9, 2011 to identify core issues and outline next steps.

<b>QUALITY IMPROVEMENT</b>				
<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	Registry will be implemented for all patients with developmental delays at all 5 sites.	3	All sites entering complete, quality data on all patients meeting criteria.	<p>Review: Partially Successful</p> <p>2 sites and four medical geneticists entering data, 1 site in IRB review, 340 total patients entered as of Dec. 2011.</p> <p>Data have been analyzed and poster will be presented at ASHG/ISHG annual meeting in Oct 2011 in Montreal</p>
2	Create a PSO to host data collected from clinic sites and/or obtain exemption letters for each site through CPHS	3	<p>Updated Definition: Legal framework in place enabling hosting and utilization of data from participating sites.</p> <p>Was: ARHQ website lists all approved PSOs / sites participate in registry under exemptions</p>	<p>Review: Successful.</p> <p>PSO efforts were dropped after multiple discussions with national and regional partners as it was determined that the developments of BAA agreements would be a better fit for the NEG's work.</p> <p>BAA agreements in place for 2 sites (and 4 medical geneticists) and templates created for IRB waivers as not human subjects research but quality improvement activity.</p>
3	Implement QI report structure	1	Report format in Registry	<p>Review: Successful</p> <p>The vendor hosting the database, GVT, has created and implemented the database, with revisions added, as needed.</p>

4	QI data submitted, analyzed and reported from all five current clinical genetics sites.	3	Updated Definition: Registry in place and utilized by all 3 clinical sites  Was: Registry in place and utilized by all 5 clinical sites	Review: Partially Successful  2 sites are submitting data to this project; a third is in IRB review. No additional sites are being considered at this time.  Data have been analyzed and an abstract presentation has been accepted by ASHG for Oct. 2011. Additional analyses/reports will be presented at the QI work group meeting in Nov. 2011.
5	Establish the Metabolic Quality Improvement Learning Collaborative	1	10 metabolic centers will send teams of 2-3 members each to QILC (3 meetings during the year).	Review: Partially Successful  9 centers initially agreed to participate in the QILC, 5 centers have sent teams to full meetings of the QILC. 7 centers have provided summary data. Two face to face sessions were held during the project year: Feb., 2011 and April, 2011; a third was held October 2011.
			Support webinars between learning sessions will support teams	Review: Successful  1 webinar held between sessions one and two of the QILC. A second was held in June 2011.
6	Establish quality improvement clinical process and outcomes for patients with metabolic disorders	3	A common set of data will be agreed upon.	Review: Successful  Data set agreed upon.
			Condition-specific measures for at least 7 metabolic disorders or problems will be set forth.	Review: Partially Successful  Agreed-upon additional specific measures set forth for 2 conditions.
7	Metabolic quality improvement registry will be established (customization of Genetics QI registry).	3	Registry exists and contains all the data elements defined by the QILC.	Review: Partially Successful  Substantive progress made during Year 4. Data sheets for PKU and MCAD were developed. These collection sheets have been submitted to GVT for implementation into electronically

				accessible data base in Year 5.
8	Metabolic centers will be members of the PSO and/or will obtain CPHS exemption letters and have HIPAA BAAs in place.	3	PSO is in place / letters are obtained or  Membership contracts are in place for all participating centers	Review: Partially Successful  Group decided that no linkage would be established at this time. Two Centers will be involved in QI activities for both Metabolic and GDD/ID. Those providers will access the Registry by QI activity.
<b>TRANSITION</b>				
<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	Develop assessment tool for measuring successful transition to medical home	3	Written list of criteria identified.	Review: Partially Successful  Collaborative work with Dr. Cooley continued through Year 4 with resolution achieved concerning whether a medical home was able to handle a transition. The next phase of the work will be to determine indicators of success that a transition has "successfully" occurred.
2	Continue to publish, present, and disseminate transition related agenda	3	Agenda promoted via published articles and presentations	Review: Successful  Publications, presentations, and materials related to Transition were shared in a range of venues (website, conferences, regional and national teleconferences).
3	Create materials for youth and adults on metabolic disorders	5	Creation of Fact Sheets that list issues for adults with these disorders written for a lay audience. 8 fact sheets will be produced in Year 4.	Review: Partially Successful  Dr. Waisbren continued development of 4 fact sheets in Year 4. The new worksheets are due to be released once the new ACMG forms are completed and information has updated and adapted for a lay audience.
4	Hold conference for adults with	6	Conference held.	Review: Unsuccessful

	metabolic disorders			A separate conference for adults with metabolic disorders was not held in Year 4. Efforts transitioned to try and support additional learning opportunities for youth/young adults. A separate grant was secured for a conference in January 2012 for adults with PKU.
5	Continue to monitor new advances in transition programs – especially through special education initiatives	4	Transition practices are summarized for genetics and metabolism	Review: Partially Successful  Although staff remain well informed of current transition practices and actively support their implementation, a formal summary was not updated in Year 4.
6	Assessment of best practice protocol by metabolic physicians and dieticians and other professional staff	1	Reviews received by at least 3 professional staff (dietician, nurse, fellow).	Review: Successful  Protocol reviewed by Dr. Levy, Fran Rohr, and Leah Hecht. It was determined that the protocol required too many resources to implement at this time.
7	Continue to Pilot transition practice at Children’s Hospital	1	10 patients participate at Children’s Hospital.	Review: Partially Successful  As a result of piloting the transition tool, it was determined that the transition tool would be better implemented in a different setting as there were too many barriers to implementation in the hospital setting (fewer than 10 patients had participated). The Metabolic Basics resource received hundreds of hits on the New England Consortium website indicating that the resource was being accessed in alternative ways.
8	Leadership training for teens with genetic disorders. Program	1	Leadership training takes place.	Review: Successful  The Teen Challenge weekend was held in July 2010. Nine youths

	at Teen Challenge Weekend			worked to build confidence, strengthen bonds, challenge comfort zones and develop some of the skills needed to manage complex health conditions.
9.	Participate in effort to improve quality in metabolic clinics via learning collaborative methodology	3	Plan developed, ratified and implemented by QILC planning group and expert panel	Review: Successful Dr. Waisbren supported the QILC throughout Year 4 by providing feedback on project material.
10.	Continue to represent transition activities on LTFU as needed.	3	Improved access to assessment for all adults with genetic conditions in New England	Review: Successful Dr. Waisbren continued work on the Uniform Screening Method by collecting data on 30 cases, analyzing and presenting the results.
11.	Collaborate with the National Transition Resource Center being developed at the Center for Medical Home Improvement	3	Seek out new opportunities and collaboration	Review: Successful Integration with the NHCTC is ongoing. Of note, NHCTC staff have been directly integrated into the Transition Workgroup.
<b>MEDICAL HOME</b>				
<b>No.</b>	<b>Objective</b>	<b>Status</b>	<b>Yr. 4 Definition of Success</b>	<b>Yr 4 Results</b>
1	Begin field test of the care coordination project in two specialty clinic catchment areas	6	Patients and families are recruited into trials of the care planning tool at Children's Hospital Boston (10 patients / families) and one other metabolic / genetics clinic (at least 5 patient / families).	Review: Unsuccessful After review of some of the barriers to implementation of the pilot (the previous project lead transitioned to a new appointment in another region, and some of information for the care planning tool was already being captured by some components of the care model), the workgroup decided to abandon this effort and pursue a new objective to improve care coordination in the

				region (Obj. 2).
			Process data are collected at the Children's Hospital site including number of plans implemented, number of visits documented.	Review: Unsuccessful Activity dropped.
2	New Objective: Assessment of communication between primary care providers, families, and specialists	3	Survey tool designed and fielded.	Review: Successful  In order to further work in the care coordination area the group decided to seek clarification of the care processes that were in place and how they were communicated between providers and families as well as between providers and specialists. A tool was developed and fielded for the first survey, with analysis and completion of the second survey expected in Year 5.
3	Convene at least 3 meetings of the MHWG during Year 4.	1	Two conference calls and one face-to-face meeting occur.	Review: Partially Successful  Group met two times in Project Year Four (December 2010, May 2011).
4	Continue to integrate meetings and work with the Transition Workgroup	3	Annual face-to-face meeting in December 2010 is a joint meeting of the two work groups.	Review: Successful  A joint meeting was held December, 2010 and May, 2011

**DISSEMINATION, EDUCATION AND MARKETING**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	Continuously improve educational products and activities for providers and consumers	3	Current model finalized and dissemination plan created based on recommendations	Review: Successful  Substantive work was carried out during Year 4 to improve the resources and capacity of the new GEMSS tool.

			Expansion of modules utilized in educational tool for special educators.	Review: Successful Multiple modules were researched and collaboratively reviewed for incorporation.
			Revise tool for pediatricians and parents.	Review: Unsuccessful Activity to be reviewed during Year 5.
2	Create web portal based on tool "Children with Genetic/Metabolic Conditions in the Educational Setting"	3	Tool posted on website	Review: Partially Successful During Year 4 the workgroup developed the infrastructure and basic schematics for the new website. The tool will go live in Year 5.
3	Improve utilization of genetic education materials	3	Identification of new resources / tools to be linked to the NEGC website and distributed to stakeholders	Review: Partially Successful During Year 4, the group reviewed the Genetics and Rare Diseases Information Center (GARD), determined that it was not user friendly and therefore not appropriate for inclusion on the NEGC website.

**DEMONSTRATE EFFECTIVE COLLABORATIONS**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	NEGC continues to participate in national work groups	3	The NEGC is actively represented on a national level by staff and NEGC constituents and contributes to the improvement and coordination of genetic services.	Review: Successful Project directors and workgroup chairs are involved in one or more national groups engaged in transforming genetic services.



2	Link with affiliated programs (LEND and AUCD)	3	MOAs developed with participating programs identifying methods of collaboration.	Review: Partially Successful  In project Year 4, an innovative project has been funded with the LEND program. Staff actively sought to integrate NEGC activities into the NH LEND program, with new collaborations to take place during Year 5.
3	Represent genetics issues to wider healthcare system	3	Additional health care fields are educated about the needs of individuals living with genetic conditions.	Review: Successful  Presentations made to AMCHP, NCHPG, and participation in a Genetics Blog. Work initiated to better integrate genetic services into each New England state's 211 system.
			Public Health Genetics and Genomics is integrated into other academic course work	Review: Successful.  Areas of involvement include: administering a course on public health genetics, providing support to UNH's MPH program, and a presentation to AMCHP.

**INNOVATIVE PROJECTS PROGRAM**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	Monitor innovative grant awardees including new micro grants to spur consumer involvement.	3	A common process is established and continuously improved for the review, selection and monitoring of awardees that is agreed to by all members of the review committee	Review: Successful  Monitoring and updating of the grant process has been continually implemented.
2	Release, award and monitor grantees	1	Grant Cycle completed.	Review: Successful

	for 2010-11			Grant cycle successfully completed.
3	Work with grantees to develop poster presentations (regular grants) / brief summaries (micro grants)	1	Poster presentations / brief summaries developed that represent and convey the spirit of the innovative projects program.	Review: Successful 4 Innovative grants were awarded Poster presentations created, reviewed by management staff and Advisory Council.
4	Confirm award amount and issue RFP for grant cycle 5	1	RFP issued.	Review: Successful Grant cycle 5 process was implemented.

**ETHICAL, LEGAL, AND SOCIAL ISSUES**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	Address ELSI issues within workgroups as well as through special projects	3	NEGC appropriately integrates ELSI issues within its work and actively pursues projects that improve the field of genetics in this area.	Review: Successful Examples: Ethical (review of patient data utilization to improve service quality, involvement of youth in planning activities), Legal (establishment of BAA agreements to enable the work of the QILC), Social (education of groups across the New England region on the importance of folic acid), Policy (addition of Rhode Island to the LTFU network).
2	Discuss ELSI issues within the RCC network.	3	Issues raised and discussed, NEGC lessons learned shared with the network	Review: Successful ELSI issues are reviewed on an ongoing basis.

**LABORATORY QUALITY ASSURANCE**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	Continued representation of quality control	3	Full participation in meetings	Review: Successful Dr. Eaton continues to participate on a number of regional and

	workgroup in regional and national forums			national initiatives (eg. NBSTRN, SACHDNC).
2	Request and analyze lab-specific data on marker descriptive statistics (means, standard deviations, etc.) on ~ total of ~ 50,000 newborns from WI and NY. Determine adjustments to category index cut-offs as appropriate.	3	Data received and reviewed.	Review: Successful  Substantive analysis work undertaken, with wrap up of analysis work to be completed in Year 5.
3	Analyze the raw data submitted, using lab specific cutoffs as appropriate. Add C4, C5DC, C5OH, C%:1, multiple acylcarnitine elevations in same sample, Cit, ASA	3	Additional analysis tables created, new indices possibly identified. Evaluation of such tables may suggest additional index possibilities beyond the indices currently used by the NENSP.	Review: Successful  New detailed data tables were produced, analysis completed on 3MCC, BKT, GA-I, MSUD, CIT-I, ASA.
4	Hold regular conference calls and face-to-face meetings, as appropriate, to review the data submitted with the partners, and compare index categorizations with follow-up data on final diagnoses.	1	Meetings held. Target web-ex meetings in Sept., Nov., Jan, and face-to-face meeting in March, possible web-ex in May	Review: Partially Successful  Full meeting of the group held in May.

5	Present preliminary findings to Lab Subgroup Committee of SACHDNC	1	Presentation	Review: Successful Presentation made in May, 2011.
<b>LONG TERM FOLLOW-UP</b>				
<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	Continued representation of LTFU workgroup in regional and national forums	3	Full participation in meetings	Review: Successful Dr. Comeau continues to participate on a number of regional and national initiatives (eg. NBSTRN, Institute of Medicine, CF/SCID conferences).
2	Continue to facilitate stepwise implementation of activities leading to full regional participation in long term follow-up	3	Continued education of state teams (NBS Advisory committees) about Massachusetts and Maine experience with implementation.	Review: Successful Primary focus during Year 4 was on collaborations with Rhode Island.
			Facilitating Workgroups and reports back to state teams from work groups	Review: Successful Primary focus during Year 4 was on collaborations with Rhode Island.
			Continue to legal counsel from each state in the discussion of method for implementation, which may be by Charter or by other agreements between and among states.	Review: Successful Rhode Island agreed to participate in the LTFU process.
3	Continued Data Collection and Expansion of Data Collection Activities	3	Subcontracts established with Maine and Rhode Island.	Review: Successful Minimum data set defined. Updated based on ongoing review of information. Maine and Rhode Island contracts in place.
			State specific data modules created and integrated.	Review: Partially Successful These modules are being created in conjunction with a very large data system replacement by the NENSP (otherwise internally funded). This project will not be complete during this year, and so full implementation

				of the LTFU accessibility aspects will occur after the grant year is completed
4	Data Analyses and Publication of Analyses	3	Data analysis prepared for QI at the clinic and program levels	Review: Successful  Analysis reports provided back to Mass. and Maine clinics. Reports run on MCAD and VLCAD. CF clinics received reports and inquiries. Hemoglobin clinics received reports and inquiries. Metabolic clinics received reports and inquiries. A special report on LCHADD has also been presented to Metabolic Clinics in preparation for publication.
			Manuscripts developed documenting findings.	Review: Successful  Genetics in Medicine paper published.
5	Enhancing Development of Best Practices	3	Hgb conference to facilitate development of best practices.	Review: Successful  Hgb conference held in September, 2010 focused on identifying best practices for improvements to patient care; attended by over 100 people from across the country.
			Development of best practices by clinical workgroups	Review: Partially Successful.  In process. None of the workgroups have established best practices. Hgb would like an organization similar to the CF foundation. Data collection is based on the CDC's RUSH program. Each group (Hgb, Metabolic, CF) is structured similarly in that they each have clinician advisors. They differ in the questions they are trying to address or document.

**QUANTITATIVE AND QUALITATIVE EVALUATIONS**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 4 Definition of Success</i>	<i>Yr 4 Results</i>
1	Gather data on program activities and outcomes and provide ongoing feedback to project staff and	3	Management staff report evaluation support has been an effective aid in decision making and program improvement.	Review: Successful  Management Staff Review:  Evaluation and survey data are used to inform NEGC activities.

	funder on project progress.			
2	Conduct annual stakeholder survey	1	A majority of stakeholders participate in the survey process and provide recommendations for the project's improvement	Review: Partially Successful  Although there was a substantive increase in the number of respondents (42 to 63), the participation rate of NEGC stakeholders for the Year 4 Survey was 45%. Substantive feedback received on potential improvements and future directions for genetic services in the New England region.
3	Complete semi-annual and annual reports which can be used by staff to improve project outcomes	3	Reports completed and utilized by staff to improve project outcomes and utilized by stakeholders to stay informed of project progress.	Review: Successful  Yr 3 Report and Yr 4 Mid-Yr report completed and reviewed by staff.
4	Participate on national outcome measurement efforts	3	NEGC is actively represented on national measurement efforts.	Review: Successful  NEGC was represented on all meetings and provided information for all national level reporting and discussions.

## OBJECTIVES FOR YEAR FIVE

Table 4 provides a list of objectives to be completed by each of the relevant workgroups and administrative teams for Year Five of the NEGC project. The status of each objective will be updated by the Project Manager on a monthly basis during meetings with the various Workgroup chairs using the following key: 1. Completed as planned, 2. Completed - deviated substantially from plans, 3. In progress - satisfactory, 4. In progress - unsatisfactory, 5. Initiation of activity deferred, 6. Activity abandoned, 7. Not scheduled to initiate in period. Workgroup chairs have established a series of performance measures to document successful achievement of each of their objectives.

**Table 4: Year 5 Goals and Objectives**

<b>Establish and Maintain the NEGC</b>			
<i>No.</i>	<i>Objective</i>	<i>Yr. 5 Definition of Success</i>	<i>Measurement of Success</i>
1	Continue implementation of core administrative supports to the NEGC	NEGC is able to successfully pursue goals and objectives and appropriately respond to changing conditions.	NEGC 2011/2012 Workplan.
2	Budget Management	Operating expenses for the fiscal year are within the budgeted amounts	Budget analysis of UNH records
3	Continue close collaboration with WG and AC	Work Group and Advisory Council members feel supported in the work they do and have access to the resources they need to accomplish their goals. Meetings and conference calls held	Work Group and Advisory Council members feel supported in the work they do and have access to the resources they need to accomplish their goals. Meeting and call minutes
4	Annual meeting	Meeting held	Meeting report
5	Communications and outreach plan	Stakeholders report satisfaction with being able to voice their opinions and feel that they've been heard. Consistent increases in NEGC web site utilization.	Stakeholder survey responses, quarterly updates sent, web site analytics
6	Implement Special Projects	Special Projects achieve stated goals within agreed upon timelines.	NEGC 2010/2011 Workplan.
<b>Quality Improvement</b>			
<i>No.</i>	<i>Objective</i>	<i>Yr. 5 Definition of Success</i>	<i>Measurement of Success</i>
1	Registry will be implemented for all patients with developmental delays at all 5	100% of appropriate data sheets entered at each of the 5 sites;	# sheets entered/# eligible patients by site;

	sites.	100% of data sheet 100% completed (i.e., no missing data elements)	# data sheets completed correctly/# total sheets by site;  Data sources are: 1) the NEGC registry; and, 2) data compilation for those sites not yet on Registry.
2	Obtain exemption letters for each site through CPHS	HIPAA BAAs in place at all participating centers	NEGC offices have copies of each HIPAA BAA.
3	Implement QI report structure	Standard report form in place and functional.	The standard report generated aggregate data for all sites in Registry.
4	Submit for publication white paper on process of quality improvement in clinical genetics services.	Paper completed	Paper completed and posted on NEGC site.  Paper submitted and published in Am J Med Genet (Part C) in 2009.  White paper on the Metabolic QI LC is in process and will be completed in year 5.  Abstract of the MET QI LC to be submitted to annual SIMD meeting late Fall 2011.
5	QI data submitted, analyzed and reported from five clinical genetics sites.	Registry reports all centers entering data in Registry.  Data is complete for each site.	Registry reports total numbers of records and data entry points by site.  Registry reports on data quality for completeness by site  Five sites participating
6	Convene one “Breakthrough Learning Series” in quality improvement for NE Metabolic Centers, based on the Institute of Healthcare Improvement using the existing four sites and with the	Series completed.  Measures in place to assess implementation of quality improvement activity in 8 metabolic centers  100% of <i>participating centers</i> will utilize care	a) X=participating centers; Y=centers using checklists; Y/X = % participating centers active with checklists  b) # Children with MCAD,



<p>purpose of rapid dissemination to other NE Centers. One face-to-face meeting with 3 webinars will be completed. We will target urban academic or private clinical genetics practices from Boston, Worcester, Providence, New Haven, etc.</p>	<p>checklists</p> <p>100% of <i>eligible patients</i> will have <i>completed</i> checklists by the end of Collaborative.</p> <p>Information is complete for 90% of patients identified and enrolled in the clinic registry (by end of the collaborative).</p> <p>100% checklist completion (number of items on the checklist complete/total number expected to be completed)</p> <p>Checklist activity leads to predicted and/or unanticipated “tests of change” in the practice.</p> <p>[“tests of change” are when an improvement area is identified, an aim written, and change ideas are tried out with a few patients, refined, implemented and measured (plan, do, study, act (PDSA)] cycle.]</p> <p>Optimal time interval between PKU screen positive and PKU diagnostic confirmation.</p> <p>Optimal time interval between MCAD screen positive and diagnostic confirmation.</p> <p>Optimal time from confirmation of dx to “metabolic control” of PKU.</p> <p>Optimal time from MCAD diagnosis and</p>	<p>PKU, = X;</p> <p># Children with checklists completed = Y</p> <p><math>Y/X = \% \text{ of identified with a checklist}</math></p> <p># Children with MCAD, PKU, others conditions identified and enrolled in registry = X</p> <p>Registry information complete = Y</p> <p><math>Y/X = \% \text{ registry information is complete}</math></p> <p>Score self on checklist for % complete (e.g. 25%, 50%, 75%, 100%) X=number complete; Y=total number; <math>X/Y = \% \text{ complete}</math>.</p> <p># of predicted or not predicted tests of change (defined by site; require PDSA cycle; # cycles) .</p> <p># days from birth to diagnosis (both PKU, MCAD). Same as above</p> <p># days from PKU dx to Phe level <math>\leq 6 \text{ mg/dL}</math></p> <p># days from MCAD</p>
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		patient diet counseling completed	diagnosis to all diet/medication counseling complete
<b>Transition</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 5 Definition of Success</b>	<b>Measurement of Success</b>
1	Continue to publish, present, and disseminate transition related agenda	Agenda promoted via published articles and presentations	Publications, presentations, and disseminated materials
2	Create materials for youth and adults on metabolic disorders	Creation of Fact Sheets that list issues for adults with these disorders written for a lay audience. 4 fact sheets will be produced in Year 5.	Distribution of Fact Sheets through the internet and clinics.
3	Continue to monitor new advances in transition programs – especially through special education initiatives	Transition practices are summarized for genetics and metabolism	Publication of review article and/or posting of summary to NEGC website.
4	Leadership training for teens (Face Forward) with genetic disorders in collaboration with Children’s Hospital and Next Step	Leadership training takes place.	Summary on training
5	Participate in effort to improve quality in metabolic clinics via learning collaborative methodology	Continued participation in QILC, incorporation of transition elements into QILC activities and recommendations.	Meeting reports
6	Continue to represent transition activities on LTFU as needed.	Completion of study to determine validity of the Uniform Assessment Method (using PKU, UCD’s and Galactosemia as models)	Publication in a peer reviewed journal
7	Collaborate with the National Transition Resource Center being developed at the Center for Medical Home Improvement	Seek out new opportunities and collaboration	List of opportunities identified and 'next steps' for collaboration defined
<b>Medical Home</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 5 Definition of Success</b>	<b>Measurement of Success</b>

1	Assessment of PCPs regarding care provision for children with complex conditions.	Survey implemented with response rate of 15%. Results reviewed and summary report generated.	Evaluator review
	Assessment of current methods among genetic and metabolic clinics regarding communication and coordination of care with primary care physicians and families.	Telephone interviews conducted with 75% of clinic settings. Results reviewed and summary report generated.	Evaluator review
2	Convene at least 3 meetings of the MHWG during Year 5	Two conference calls and one face-to-face meeting occur.	Meeting agenda and attendees document the meetings.
3	Continue to integrate meetings and work with the Transition Workgroup	Annual face-to-face meeting in November 2011 is a joint meeting of the two work groups.	Meeting agenda and attendees document the meeting.
4	Assessment of PCPs regarding care provision for children with complex conditions.	Survey implemented with response rate of 15%. Results reviewed and summary report generated.	Evaluator review

**Dissemination, Education, and Marketing**

<i>No.</i>	<i>Objective</i>	<i>Yr.5 Definition of Success</i>	<i>Measurement of Success</i>
1	Launch GEMSS website	Website launched	Evaluator review.
	Continuously improve GEMSS resource.	Expansion of modules utilized in GEMSS for special educators. Targeted conditions for Year 5 include: Down syndrome, Williams syndrome, Achondroplasia and other dwarfing conditions and possibly NF1.	Meeting Minutes
		Gather feedback and recommendations from website visitors	Survey created, implemented, results analyzed, and followed up on.
2	Disseminate GEMSS website	Increasing web hits throughout the year.	Google Analytics.
3	Improve utilization of genetic education materials	Identification of new resources / tools to be linked to the NEGC website and distributed to stakeholders	NEGC Website, Weblogs, Evaluator Review.
4	Collaborate with core staff to	Increased number of genetic services	Number of genetic services

	enhance 211 linkages for genetic services	posted to each state's 211 system	posted to each state's 211 system
<b>Effective Collaborations</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 5 Definition of Success</b>	<b>Measurement of Success</b>
1	Core staff and collaborative council members participate in national and regional groups	Each staff and CC member participate in at least one regional or national work group	Work group rosters
2	Engage LEND students and/or students at genetic counseling programs in research activities	Student participation results in poster or abstract development	Product (poster/abstract)
3	Presentations/publications at regional/national venues	Additional health care fields are educated about the needs of individuals living with genetic conditions. Presentations given / publications issued	NEGC Publications / Presentations list. # of publications in medical journals covering issues facing genetic services, cross-collaborative grants submitted with primary care providers.
<b>Innovative Projects</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 5 Definition of Success</b>	<b>Measurement of Success</b>
1	Quarterly reports from PI of each project	Reports received	Quarterly and year end reports
2	Release RFPs, select reviewers, review applications, notify applicants	Grants awarded	# of applications received, # of applications funded
3	Present posters at annual meeting	Posters created and displayed	# of posters displayed at annual meeting
<b>Ethical, Legal, and Social Issues</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 5 Definition of Success</b>	<b>Measurement of Success</b>
1	Integrates ELSI issues within projects	ELSI projects completed	Publications, activities identified in Year End report
<b>Long Term Follow Up</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 5 Definition of Success</b>	<b>Measurement of Success</b>

1	Document formal authority for LTFU on state-by-state basis.	Legislation, regulations or interpretations of state rules	NBS coordinators
2	Develop State Agreements for extending centralized database to include LTFU variables	Contract amendments	Contracts in place
3	Data collection and analyses of minimum data sets.	Updates on 70% of patients.	Summary data analysis
4	Refine dataset variables per condition-specific needs	Variable list updated	Meeting Minutes
5	Participate in Interregional and NCC activities	egular participation in activities.	Meeting Minutes, presentations
<b>Psychosocial Follow Up</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 5 Definition of Success</b>	<b>Measurement of Success</b>
1	Validate method using historical data to compare results of the Uniform Assessment System to psychological testing results	Method identifies at least 90% of children who are at risk for developmental delay, learning disabilities, or emotional problems	Medical records at Children's Hospital Boston
2	Finalize method	Agreement is reached on a method	Members of work group
3	Develop computerized form	A system is up and running by 6 months into the 5 <sup>th</sup> year	A website
4	Pilot the method in 2 metabolic centers	Parents of 10 patients (from a range of ages, 6 months to 10 years) will complete the Uniform Assessment Method	Completed forms from 10 parents
5	Create a website for the Uniform Assessment Battery	Includes description of the method, instructions for completing questionnaires and secure access to questionnaires and results	Website
<b>Quality Assurance</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 5 Definition of Success</b>	<b>Measurement of Success</b>
1	Continued representation of quality control workgroup in regional and national forums	Participation at appropriate meetings	Documentation of participation at meetings
2	Analyze applicability of lab-	Completion of the task as stated	Tables that accurately

	specific index categorizations (developed over past 4 years) to follow-up data of the remaining disorders (those not yet analyzed in first 4 years) detectable by MSMS		represent the positive predictive values of all categories with all disorders
3	Analyze applicability of lab-specific index categorizations to follow-up data for those new babies detected after analyses for those disorders which were done during earlier phases of the study	Completion of the task as stated	Tables that accurately represent the positive predictive values of all categories with all disorders
4	Hold regular conference calls and face-to-face meetings, as appropriate	Holding of said meetings	Documentation of meetings
5	Publish final findings in peer-reviewed journal	Publication	Submission and publication

**Evaluation**

<i>No.</i>	<i>Objective</i>	<i>Yr. 5 Definition of Success</i>	<i>Measurement of Success</i>
1	Gather data on program activities and outcomes and provide ongoing feedback to project staff on project progress	Management staff report evaluation support has been an effective aid in decision making and program improvement.	Annual review, Meeting minutes of review.
2	Conduct annual stakeholder survey	A majority of stakeholders participate in the survey process and provide recommendations for the project's improvement	Data collected, More than 50% of known stakeholders participate in the survey (documented by Survey Monkey), Stakeholder Survey report generated and published to website.
3	Complete annual reports which can be used by staff to improve project outcomes	Reports completed and utilized by staff to improve project outcomes and utilized by stakeholders to stay informed of project progress.	Meeting minutes affirming utilization of material, Stakeholder Survey report documenting majority agreement that the report is a useful resource for stakeholders.

4	Participate on national outcome measurement efforts	NEGC is actively represented on national measurement efforts.	Performance Measure reports are fully completed and delivered on time.
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## PROJECT CHALLENGES AND RECOMMENDATIONS

This section provides an overview of both project-wide and Workgroup level issues identified by Dr. Antal along with recommended next steps. Challenges included in this section are drawn from issues raised by stakeholders during the course of the project, findings from stakeholder surveys and annual meetings, and/or staff review during project meetings. Status updates for each are defined as:

- ❖ Not addressed (0 of 12): no substantive activities have been undertaken
- ❖ In process (3 of 12): activities are under way to address the challenge but have not yet led to substantive changes in practice
- ❖ Improving (8 of 12): activities have led to substantial improvements in the challenge area
- ❖ Addressed (1 of 12): the basic nature of the challenge has been successfully addressed by project staff

Since the previous year's report:

- ❖ one item (Implications of Insurance Reform) has been moved from 'new challenge' to 'in process.'
- ❖ one item (Access to Genetic Specialists) has been moved from 'not addressed' to 'in process'
- ❖ one item (Many Stakeholders, Limited Funds) has been moved from 'in process' to 'improving'

### Update on Challenges Identified to Date

#### Status: In Process

##### *Implications of Insurance Reform for Individuals with Genetic Conditions*

Background: At several points during the last few years, several NEGC partners have noted the significant challenges that are created by the lack of coverage for certain services by insurance policies. As health care reform continues to be implemented, clarity will be needed as to the implications for the health and well-being of individuals living with genetic conditions. With greater clarity should come a better sense of what actions can be taken to address some of the gaps in the health care system.

Recent Activity: Starting in Year 5, the NEGC took several steps to begin addressing this area. One, forming partnerships with Kay Johnson (a national expert on MCH policy and funding challenges and a speaker in health reform conversations) as well as the Catalyst Center (a national center dedicated to improving health care coverage and financing for Children and Youth with Special Health Care Needs). Additionally, through its innovative project, the NEGC is supporting the University of Connecticut Health Center's work to assess implications of the Affordable Care Act for access to genetic medical services in New England. Lastly, the NEGC has recently launched two new groups, the Advocacy group which has taken on this issue broadly, as

well as a subgroup focused specifically on ensuring access to medical foods for families and individuals in the region.

**Recommendation:** Review analysis work conducted by the University of Connecticut Health Center as well as discussions and findings from the Advocacy and Medical Foods group. Consult with Kay Johnson and the Catalyst Center to help the Advisory Committee and NEGC staff to identify best next steps.

#### *Access to Genetic Specialists*

**Background:** One of the challenges identified by the Medical Home workgroup during the first project year is the scarcity of physicians with specialty training in genetics. More genetics doctors are leaving the field than are entering it. Without other substantive changes in the field, this trend will threaten the NEGC goal of improving patient access to quality care.

**Recent Activity:** The Medical Home workgroup has begun looking at this issue through an assessment of communication lines between families, PCPs, and genetic specialists. The purpose of this work is to assess the comfort level and communication preferences of primary care physicians related to caring for children with rare conditions including genetic disorders. In combination with their survey work of genetic specialists, the workgroup will have the potential to identify strategies for improving lines of communication, thus making better use and enhanced dissemination of specialist knowledge across PCPs. Additionally, recent developments in Year 5 have led to new partnerships with the NH LEND (Leadership Education in Neurodevelopmental and Related Disabilities) program. The improved collaborations with both New Hampshire's and Maine's activities in this area has the potential of maintaining and even improving PCP access to specialists with genetic research knowledge and training.

**Recommendation:** Continue to support activities in these areas. Concerning the NH LEND partnerships, outline a set of strategic goals that should be accomplished each year that meets the needs of both organizations. Consider integration of recommendations developed by the Medical Home workgroup concerning improvements in collaboration between PCPs, families, and specialists into the NH LEND curriculum.

#### *Availability of Care Management Information for Individuals with Genetic Disorders*

**Background:** Another challenge for Medical Home practice is that little case management information for genetic disorders has been published. If this information was more accessible, it is possible that PCPs could perform more elements of patient care (and so help to address the lack of physicians trained in a genetic specialty). During Years Two through Four, substantive efforts were made to educate both regional and national level stakeholders about the need for a medical home.

**Recent Activity:** The care coordination and transition pilot projects helped to raise awareness of these issues and created a set of online materials for use by a variety of stakeholders. The Long Term Follow Up workgroup had major successes with the inclusion of Maine and Rhode Island as partners in tracking long term follow up data. Lastly, the launching of the new Quality Improvement Learning Collaborative as resulted in a long term learning partnership being formed across 8 metabolic centers in the New England region.

**Recommendation:**



- Continue supporting the Medical Home and Transition workgroup's efforts to implement the care planning and transition tools in a variety of settings. The NEGC should consider gathering a minimal level of evaluation information (beyond web entries) to aid in future improvement and expansion of the tools.
- Continue supporting the QI and LTFU efforts to integrate data from a variety of settings so that an accurate picture can be created on what does and does not produce successful outcomes among individuals with a range of genetic and other health conditions. Re: LTFU, projects such as the Hemoglobinopathy conference should be supported as they have great potential to bring together LTFU data, leaders from across the nation, and area clinics to develop their thinking around best practices and to set the stage for improvements in knowledge for gaining successful health outcomes.

### **Status: Improving**

#### *Many Stakeholders, Limited Funds*

Background: Partners of the collaborative grappled with the challenge of multiple partners planning to submit grant applications in response to the same RFA/PAs. Some of the issues encountered included: how to balance sometimes competing interests, when the NEGC (and its fiscal agent, UNH) should take a leadership vs. supporting role in a grant application, how to determine what is best for the region, and how partner organizations can better balance working toward the NEGC mission while fulfilling their own organizational mission. In August of 2009, the collaborative council met and, in the process of discussing the above issues, developed a protocol for handling future grant opportunities. While the protocol is helpful for laying out a process for initial discussion when an RFP notice is sent out, finding agreement to everyone's satisfaction as to which entity should lead may not always be achievable.

Recent Activity: The protocol has been used consistently over the past year to inform partners of emerging grant opportunities, hold collaborative discussions around potential projects to pursue and to identify most appropriate leads. During Year 4, the NEGC had considered applying for a grant to the Genetic Alliance to integrate Family Health History Patient Education Toolkits into a health care setting. Based on discussion with partners from Vermont, it was determined that it was most appropriate for a community health center in that state to take the lead on applying for the initiative. However, this group ultimately decided not to apply. A review of how this process worked during the past year indicated that this was the most feasible solution.

Recommendation: Continue to seek out and take advantage of opportunities to collaboratively improve grant resources in the region. Review annually with partners the NEGC's approach to this area to determine how well it is working and to identify any recommendations for improvement in the process.

#### *Lack of Specialty Care Providers for Adults*

Background: During Year One, concerns were raised about the ability for youth with genetic conditions who were transitioning to adult care to have regular access to a PCP in their adult life.

Recent Activity: The Transition Workgroup, at both a national and regional level, has continued to support access to continuous care among youth. This includes dissemination of the Transition toolkit, leading national and regional dialog on Transition, and partnering with the Face Forward program to implement youth

directed programs geared towards giving youth the skills sets they need to manage a successful health care transition.

**Recommendation:** Efforts in this area should continue to be supported to ensure that as many youth as possible find a seamless transition in their care provision from youth to adult health care systems. Parallel to this, it may be helpful to conduct a region wide survey every few years to gain an accurate scope of the problem (e.g. % of youth ages 19-29 with genetic conditions without access to a PCP) as well as a better understanding of the primary barriers for effective care among the members of this group.

#### *Common Conceptions of People, Roles, and Decision Making Processes*

**Background:** During Years Two and Three, substantive efforts were made to revise the NEGC website with information on project structure, major events and membership, increase email communications and updates, provide more accessible meetings, as well as organize monthly calls with workgroup chairs. Despite these endeavors (and some improvement since then), results from the Stakeholder Survey and the NEGC annual 2009 meeting continue to indicate a need for better dissemination of information around the work of the NEGC and the roles of each of the workgroups and projects.

**Recent Activity:** In Year Four, the NEGC launched a major redesign of its website to make information more accessible and regularly implemented quarterly email updates to partners in order to inform them of major activities of the NEGC. Additionally, the NEGC focused outreach efforts to genetic counselors as well as advocates and family members (through the creation of a new Advocacy Committee).

**Recommendation:** As documented by respondents to the participant survey, efforts to communicate the NEGC's mission to stakeholders has resulted in improvements in understanding. It is recommended that the NEGC continue efforts to update the website on a regular basis, inform partners of evolving national priorities, continue with the provision of quarterly updates via email, and facilitating communication at the annual meeting. In preparation for any future annual meetings, it would be helpful to allocate 15 to 30 min to briefly highlight the year's accomplishments to all meeting participants to better ensure that everyone is on the same page in moving forward

#### *Cross-Fertilization of Ideas, Resources*

**Background:** Findings from the 2009 annual meeting as well as several individuals from the stakeholder survey noted the continued need to reach out to like-minded groups at the national, regional, and state levels. During Year Three, new partnerships were formed with the Birth Defects Consortium, Genetic Alliance, and area hospitals. As the NEGC continues to grow and promote the health and social well-being of those with inherited conditions through collaborations of its partners, it will be critical to sustain existing partnerships and identify new ones.

**Recent Activity:** Additional outreach during Year Four focused on the Birth Defects Consortium, genetic counselors, and advocates. Notably, concerted efforts were made to begin strategic integration of NEGC and NH LEND activities.

Recommendation: Continue to use the opportunity of the annual meeting, with its range of participants, to both review current partnerships and identify needed new ones. Further solidify partnership arrangements with NH LEND, and pursue potential partnership ideas identified in the 2011 NEGC Stakeholder survey.

#### *Geographic Barriers to Meeting*

Background: Continued limitations in use of state funds for travel, as well as multiple national and regional meetings pose substantive challenges to holding collaborative meetings. During Years Three and Four, the NEGC has increased its use of Webex technology for meetings and has sought to combine meeting events with other initiatives whenever possible (e.g. combination of NERGG and NEGC annual meeting).

Recent Activity: Workgroup leaders continue to make good use of conference calls and technology to support their meetings when face to face meetings are not feasible. These resources continue to provide an effective means for members to conduct their work.

Recommendation: Explore more dedicated spaces and/or equipment among partners to further improve web-based (e.g. Webex or similar) technologies. While Webex has been useful in the past, video and audio capabilities are sometimes limited for full group meetings and can limit potential participation.

#### *Quality Data Systems*

The QI, Transition, Medical Home, and LTFU Workgroups have all expressed a need for quality patient data systems to inform their work and improve outcomes for individuals with genetic conditions. During Year Three, substantive progress was made in laying the foundation for data improvement. This was achieved through work by the LTFU Workgroup in Maine supporting integration of LTFU systems, QI initiatives to start a learning collaborative, and Medical Home and Transition Workgroup efforts to improve on information collected (and how it was used) between patients, PCPs, and specialists.

Recent Activity: In Year Four, substantive progress was made through the addition of Rhode Island to the LTFU partnership, establishment of BAAs with Maine (Vermont is pending) and Dartmouth clinics to track data on children with developmental delay, and the launching of the quality improvement learning collaborative which will look at quality improvement initiatives for PKU and MCAD.

Recommendation: As data collection activities get more fully underway and used for quality improvement work, there will be a natural collective interest in sharing findings with broader audiences to ensure broad dissemination of useful findings. Given the distinction that IRBs can place on research for the sake of evaluation, quality improvement, vs. improving knowledge, it will be important to clarify what the potential implications are for IRB reviews and the most appropriate role for each group to take concerning the handling and use of protected health information for vulnerable populations.

#### *Patient Access to Genetics Information.*

Background: Concerns have been raised during the course of the project relative to the ability for patients with a genetic condition to access relevant information. During Years Two through Four of the project, substantive additions were made to the NEGC website to help fill this gap. Additionally, the DEM workgroup began reviewing resources for appropriateness and potential inclusion on the website.

Recent Activity: During Year Four, the NEGC launched a new Advocacy workgroup which has established access to care and understanding implications of the Affordable Care Act as one of its primary areas of focus. In addition, this group will serve as a reviewer of the NEGC website and provide suggestions concerning additional material to include that would benefit patients and their families.

Recommendation: Continue supports for the Advocacy workgroup and the new Medical Foods sub group that begins in Year Five. Additionally, make resources available to complete NEGC efforts to support the 211 system in New England. Currently, there is a substantive lack of information on genetic services in this system. It would be helpful to request relevant service providers to link their information in to this system in order for individuals and families to have an additional route of access to critical care and support information.

#### *Tracking Progress of Work Groups*

Background: In Year One, an issue was raised by evaluation staff concerning the flow of information and timeliness of material / feedback provided. There have been continued improvements in communication as observed via monthly meetings, more timely responses to federal report requests, and openness in discussion during collaborative council meetings. The addition of an objective and activity tracking plan in Year Three aided oversight and planning of project activities substantially.

Recent Activity: Monthly chair calls, posting of workgroup minutes to the website, and regular use of the NEGC workplan have kept staff well informed of the progress of the NEGC and helped to identify needed areas for action.

Recommendation: To support efforts in this area, it would be helpful to establish a standard set of items to include in workgroup minutes. At a minimum, it is recommended that all workgroups should include the following information in tracking their meetings: meeting date, participants, major discussion points, barriers encountered and solutions identified (if any), next steps and person(s) responsible. Though not always possible (given the need for formal approval of minutes), minutes should be posted on the NEGC website within two weeks following a meeting.

#### **Status: Addressed**

#### *Development of Logic Models and Performance Measures for Workgroups*

During Year One, evaluation staff sought to develop a series of additional logic models and measures with each of the Workgroups. However, given the status of the project and the need for chairs to focus on the start up of the program it was decided by both project management and evaluation staff that such reporting went beyond the immediate needs of the project. While information flow improved in Year Two, workgroup chairs agreed to an initial set of performance measures for their activities during Years Three and Four. These measures were then tied to goals, objectives, and individual activities and used throughout the course of the year for program oversight. While there will continue to be refinement of the process in the years ahead, the necessary infrastructure and culture is in place that will enable effective use of the work carried out by the NEGC.

## **APPENDIX A: NEGC ORGANIZATIONAL CHART**

# NEW ENGLAND REGIONAL COLLABORATIVE ORGANIZATIONAL CHART

## NEW ENGLAND REGIONAL COLLABORATIVE ADVISORY COMMITTEE

CT

RI

MA

NH

ME

VT

### REPRESENTATIVES FROM:

•Public Health

•Genetics Services

•Consumer Organizations

### REGIONAL COORDINATING CENTER (RCC AT UNH)

#### MANAGEMENT TEAM

- John Moeschler, MD, Co-PI
- Monica McClain, PhD, Co-PI, Project Manager
- Karen Smith, Project Coordinator
- Peter Antal, PhD, Project Evaluator

### SPECIAL PROJECTS

- Workforce
- Advocacy and Education
- Emergency Preparedness
- Innovative Grants

### COLLABORATIVE COUNCIL WORKGROUP/LEADERS

#### DISSEMINATION, EDUCATION, MARKETING

Leah Burke, MD

#### LINKAGES TO MEDICAL HOME

Carl Cooley, MD

#### ACCESS TO SERVICES

#### TRANSITION TO ADULT SERVICES

Susan Waisbren, PhD

#### QUALITY IMPROVEMENT

John Moeschler, MD

#### FOLLOW-UP

Anne Comeau, PhD

#### LABORATORY QUALITY ASSURANCE

Roger Eaton, PhD

### STATE REPRESENTATION

CT

RI

MA

NH

ME

VT

## APPENDIX B: NEGC Grant Applications

<b>Direct Applications</b>		
<b>Grant Name</b>	<b>Description</b>	<b>Amount</b>
Natural History of Disorders Identifiable by NBS	Project Yr 4. NIH. Collaborate with NYMAC to assess natural history of several targeted conditions in order to create a stronger foundation for improving care.	NOT FUNDED
Administrative Supplemental	Project Yr 3. HRSA; funds for legal analysis work and creation of the learning collaborative.	\$45,000 FUNDED June 2010
Administrative Supplemental	Project Yr 2. HRSA; funds for QI data registry and electronic medical record pilot	\$75,000 FUNDED April 09
Assess capacity of genetic workforce	Project Yr 2. ACMG; assess genetic workforce in light of expanded nbs; Bob McGrath will collaborate	\$36,000 FUNDED April 09
Down Syndrome Surveillance	Project Yr 2. CDC; 4 yr grant for \$400,000 to study prevalence of DS at birth and older ages; overview of health across lifespan; Bob McGrath, David LaFlamme, IOD will collaborate	NOT FUNDED
Genetics Health Care Quality Improvement Project: A Multi-State Pilot Collaboration	Project Yr 2. AHRQ; \$300,000 for 2 yrs  QI activities	NOT FUNDED
Dartmouth Translational Research Center	Project Yr 2. Submitted by John Moeschler to supplement QI project	NOT FUNDED

Galactosemia and Premature Ovarian Insufficiency	Project Yr 2. AUCD; collaboration with Susan Waisbren; submitted Oct 08	NOT FUNDED
<b>Letters of Support for Partner Applications</b>		
<b>Grant Name</b>	<b>Description</b>	<b>Amount</b>
Genetics in Primary Care Institute	Project Yr 4. American Academy of Pediatrics. Create a community of learners to enhance primary care provider ability to provide genetic related services, address systems and policy supports to accelerate provision of genetic medicine, assess residency training curriculum for genetic medicine.	Funded.
Family to Family Health Information Center	Project Year 4. Federation for Children with Special Needs.	Funded.
NBSTRN	Project Year 4. American College of Medical Genetics. Build an electronic data capture tool for long term follow up of children identified by newborn screening.	Letter written supporting need for activity.
Noonan Foundation	Project Year 4. Children's Hospital Boston. For follow up meetings of the Face Forward Program.	Not funded.
The Parent-Child Relationship and Newborn Screening: Preserving the Ties that Bind	Project Year 4. Assess whether the parent-child relationship is disrupted in parents whose infant receives an initial out-of-range newborn metabolic screening result and whether uncertainty surrounding the result is associated with reduced self-reported ratings of bonding for both mothers and fathers.	Not funded.
Clearinghouse of NBS Information	Project Yr 3. The NEGC supported an application by the Genetic Alliance and NNSGRC.	Funded. The NEGC received a subcontract of \$10,000 per year to support further collaboration. Leah Burke serves on the project Advisory Committee



Congenital Conditions Program	Project Yr 3. The NEGC supported an application by the Genetic Alliance and Family Voices.	Funded to Genetic Alliance.
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## APPENDIX C: NEGC PRESENTATIONS LIST

### \* New in Year Four

#### Sharing Work on Project Activities

\* *Region 1 Quality Control Project: Multicenter Validation of Algorithms to Improve Communications of Positive Newborn Screening Results to the Medical Home.* Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, Laboratory Standards and Procedures Subcommittee Meeting, May 2011, Washington, D.C.  
Sahai I, Caggana M, Morrissey M, Rodriguez D, Baker, M, Hoffman G, Sommers P, Manning A, Eaton R.

\* *Joint presentation by five Regional Genetics Collaboratives*  
Association of Maternal and Child Health Programs, Washington DC  
February 2011  
Karen Smith

\* *LTFU data on children diagnosed with long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD) by NBS.* December, 2010, Portsmouth NH.  
Dr. Inderneel Sahai

\* *Presentation of Massachusetts data as a part of the CF NBS and Care Quality Improvement Short Course.* 2010 24<sup>th</sup> Annual North American Cystic Fibrosis Conference. October 21-23, 2010. Baltimore, MD.  
Dr. Anne Comeau

\* *A guide for the classroom for children with genetic conditions: preliminary needs assessment and development.* National Coalition for Health Professional Education in Genetics Annual Meeting, Sept. 23-24, 2010; Bethesda, MD.  
Dr. Leah Burke

*Update on LTFU activities in New England.*  
NCC/RC PU Annual Meeting,  
November 17, 2009, Bethesda, MD.  
Dr. Anne Comeau

#### Poster session:

- NEGC
  - NEGC Work Groups
  - Innovative Projects
- NEGC Annual Meeting
- 
- Dec 2009

*Meet Your Neighbor: NEGC*  
Genetic Alliance webinar  
May 2009  
Amy Schwartz

*Poster Session: NEGC*  
ACMG Meeting, Tampa, FL  
March 2009  
John Moeschler

*Poster session: NEGC*  
NCC/RC Meeting, Bethesda, MD  
January 2009  
John Moeschler & Amy Schwartz

#### Poster session:

- NEGC
  - NEGC Work Groups
  - Innovative Projects
  - CSHN Survey Analysis Presentation – Bob McGrath
- NEGC Annual Meeting
- 
- Dec 2008

*Long Term Follow up of Newborn Screening Conditions in New England ~ New Hampshire NBS Advisory Committee*  
October 2008  
Anne Comeau

*Long Term Follow up of Newborn Screening Conditions in New England ~ Rhode Island NBS Advisory Committee*  
September 2008  
Anne Comeau

*Long Term Follow up of Newborn Screening Conditions in New England ~ Maine NBS Advisory Committee*  
September 2008  
Anne Comeau

## **Educating Students**

*Public Health and Genetics*  
Rivier College and Nursing School, Nashua, NH  
March 2009  
Amy Schwartz

*Class at UNH Graduate Program: Fundamentals of Public Health*  
Fall 2008  
Amy Schwartz (co-faculty)

*Innovative Project: Patients as Teachers*  
Multiple presentations to medical school students  
2007-2009 (2 funding cycles)  
Mark Korson, Tufts University, project PI

*Innovative Project: Nurse Educators Incorporate ANA Guidelines on Genetics*  
Videotaped training module presentations, now available online  
2007-2008  
Susan Capasso, St. Vincent's Academy, project PI

## **Training Professionals**

*\* Parents' role in specialty referrals: views from both sides of the exam table.* Pediatric Academic Societies Annual Meeting, April 28-May 1, 2011, Denver, CO.  
Fischer SH, Cooley WC, Mazor KM, Dworetzky B, Stille CJ.

*\* Poster Session: Parents' role in specialty referrals: views from both sides of the exam table.*  
Pediatric Academic Societies Annual Meeting, April 28-May 1, 2011, Denver, CO.  
Fischer SH, Cooley WC, Mazor KM, Dworetzky B, Stille CJ.

*\* Poster Session: Notes from the front lines: psychosocial follow-up of newborn screening.*  
ELSI Congress: Exploring the ELSI Universe, April 12-14, 2011, Chapel Hill, NC.

Fanos JH.

*\* Neurocognitive Outcomes in PKU.*  
South East Regional Genetics Group (SERGG), March 31, 2011  
New Orleans, LA (presented via webinar)  
Waisbren, S.

*\* Poster Session: The adult galactosemic phenotype.*  
Society for Inherited Metabolic Disorders Annual Meeting, Feb 27-March 2, 2011; Pacific Grove, CA.  
Waisbren S.

*\* "Surviving to Thriving: Improving Long-term Outcomes in Sickle Cell Disease."* New England Conference sponsored by the Hemoglobin Workgroup.  
September 16 2010, Boston, MA.

*\* Poster Session: A guide for the classroom for children with genetic conditions: preliminary needs assessment and development.*  
National Coalition for Health Professional Education in Genetics Annual Meeting, Sept. 23-24, 2010; Bethesda, MD.  
Burke L.

*CF: recommendations to increase Newborn Screening efficiency.*  
7th International Congress, Latin American Society of Inborn Errors of Metabolism and Neonatal Screening,  
December 7, 2009, Cancun, Mexico  
Anne Comeau

*Neurocognitive issues in PKU and Transition to Adult Care*  
National PKU Alliance Mtg  
Texas  
January, 2010  
Susan Waisbren

*Implementing AAP Developmental Screening Guidelines in the Primary Care Medical Home*  
NH Pediatric Society  
April 2009  
Carl Cooley

*DEM work group project: Family Health History Awareness*

Multiple presentations during pilot phase to health care community in NE, now available online 2007-2009

Meagan Krasner

*Incorporating Genetics Into the Medical Home*

NEGC/NERGG Collaborative Session at annual meeting

December 2008

Carl Cooley

Genetics presentation at NERGG annual meeting December 2008

Leah Burke

*The Primary Care Medical Home and the Care of Children with Metabolic Disorders*

New England Metabolic Program Consortium

November 2008

Carl Cooley

*Newborn Screening Molecular Training Workshop*

November 18-24, 2008

Anne Comeau

*Newborn Screening and Genetic Testing Symposium*

November 3-6, 2008

Anne Comeau

*Genetic Health Care Quality Improvement.*

Annual Meeting of the National Newborn Screening and Genetics Coordinating Center, Bethesda MD.

January 7, 2009.

John Moeschler

*Development of Collaborative Organizations.*

National Coordinating Center of the Newborn Screening and Genetics Collaborative meeting.

Chicago, IL.

June 5, 2009.

John Moeschler

*Lectures given: Office-Based Evaluation of Children with Suspected Genetic or Metabolic Disorders.*

American Academy of Pediatrics Visiting Professor to the Georgia Academy of Pediatrics. The Diagnostic Evaluation of Children with Autism & Related Diagnoses.

Amelia Island, FL. Host Paul Fernhoff, M.D. and Frank Bawyer, M.D., FAAP.

June 18-19, 2009.

John Moeschler

*Translating clinical guidelines into quality improvement: the New England Genetics Cooperative experience.*

American College of Medical Genetics, Annual Meeting. Quality Improvement Special Interest Group.

Marc Williams, M.D., host. Albuquerque, N.M.

March 24, 2010.

John Moeschler

*Workshop: Genotype-first or phenotype-first? How to balance laboratory testing with genetic evaluations. Plenary Presentation: "Clinical evaluation of patients with developmental delays, birth defects and other potential genetic disorders—why complete evaluation should precede genetic testing.*

American College of Medical Genetics, Annual Meeting. Ballroom C, Albuquerque Convention Center.

Robert Saal MD and Yves Lacassie MD, hosts.

March 25, 2010.

John Moeschler

*Keynote address*

International Conference for Adults and Children with PKU, Chicago, IL

Aug 2008

Susan Waisbren

*Transition: Psychosocial Considerations*

(power point presentation, available on NEGC website)

Susan Waisbren

*Innovative Project: Sickle Cell Disease Life Skills Training to Improve Outcomes*

Multiple presentations to young adults in NE 2007-2009

Bill Kubicek, Next Step, project PI

*Communication of relative risk for cystic fibrosis following a positive newborn screening result.* Newborn Screening and Genetic Testing Symposium, November 3-6, 2008, San Antonio, TX

Hale JE, Parad RB, Dorkin HL, Gerstle r, Lapey A O'Sullivan BP, Spencer, T, Yee W and Comeau AM.

*Quality measures enhanced by short and long-term follow up in a newborn screening program collaborating with multiple centers.*

University of Massachusetts Medical  
School/Commonwealth Medicine Conference,  
October 25, 2007, Worcester, MA.  
Hale JE, Parad RB, O'Sullivan BP, Quizon AI,  
Martin T, Yee W, Dorkin HL, Comeau AM.

*Quality measures enhanced by short and long-term follow up in a newborn screening program collaborating with multiple centers.*

21<sup>st</sup> Annual North American CF Conference  
October 3-5, 2007, Anaheim, CA.  
Hale JE, Parad RB, O'Sullivan BP, Quizon AI,  
Martin T, Yee W, Dorkin HL, Comeau AM.

## APPENDIX D: NEGC PUBLICATIONS LIST

### \* New in Year Four

#### *Peer-Reviewed Journal Articles*

1. \* McGrath RJ, Stransky ML, Cooley WC, Moeschler JB. National profile of children with Down Syndrome: disease burden, access to care, and family impact. *J Pediatr*. 2011; in press.
2. \* Woo HC, Lizarda A, Tucker R, Mitchell ML, Vohr B, Oh W, Phornphutkul C. Congenital hypothyroidism with a delayed thyroid-stimulating hormone elevation in very premature infants: incidence and growth and developmental outcomes. *J Pediatr*. 2011;158(4):538-42.
3. \* Sahai I, Eaton RB, Hale JE, Mulcahy EA, Comeau AM. Long-term follow-up to ensure quality care of individuals diagnosed with newborn screening conditions: early experience in New England. *Genet Med*. 2010;12(12 Suppl):S220-7.
4. \* Hale JE, Parad RB, Dorkin HL, et al. Cystic fibrosis newborn screening: using experience to optimize the screening algorithm. *J Inherit Metab Dis*. 2010;33(Suppl 2):S255-61.
5. Waisbren, S. Establishing a consortium for the Study of Rare Diseases: The Urea Cycle Disorders Consortium. *Mol Genet Metab.*, Feb 2010; 100 (Suppl 1): S97-S105
6. White DA, Waisbren S, van Spronsen FJ. The psychology and neuropathology of phenylketonuria. *Mol Genet Metab*. 2010;99(Suppl 1):S1-2.
7. White DA, Waisbren S, van Spronsen FJ. Final commentary: a new chapter. *Mol Genet Metab*. 2010;99(Suppl 1):S106-107.
8. Waisbren S, White DA. Screening for cognitive and social-emotional problems in individuals with PKU: tools for use in the metabolic clinic. *Mol Genet Metab*. 2010;99(Suppl 1):S96-99.
9. Koch R, Trefz F, Waisbren S. Psychosocial issues and outcomes in maternal PKU. *Mol Genet Metab*. 2010;99(Suppl 1):S68-74.
10. Brumm VL, Bilder D, Waisbren SE. Psychiatric symptoms and disorders in phenylketonuria. *Mol Genet Metab*. 2010;99(Suppl 1):S59-63.
11. Moeschler JB, Amato RS, Brewster T, et al. Improving genetic health care: a Northern New England pilot project addressing the genetic evaluation of the child with developmental delays or intellectual disability. *Am J Med Genet C Semin Med Genet*. Aug 15 2009;151C(3):241-254.
12. McGrath RJ, Laflamme DJ, Schwartz AP, Stransky M, Moeschler JB. Access to genetic counseling for children with autism, Down syndrome, and intellectual disabilities. *Pediatrics*. Dec 2009;124(Suppl 4):S443-449.
13. Homer CJ, Cooley WC, Strickland B. Medical home 2009: what it is, where we were, and where we are today. *Pediatr Ann*. Sep 2009;38(9):483-490.
14. Cooley WC, McAllister JW, Sherrieb K, Kuhlthau K. Improved outcomes associated with medical home implementation in pediatric primary care. *Pediatrics*. Jul 2009;124(1):358-364.

15. Waisbren SE, Levy HL, Noble M, et al. Short-chain acyl-CoA dehydrogenase (SCAD) deficiency: an examination of the medical and neurodevelopmental characteristics of 14 cases identified through newborn screening or clinical symptoms. *Mol Genet Metab.* Sep-Oct 2008;95(1-2):39-45.
16. Waisbren SE. Expanded newborn screening: information and resources for the family physician. *Am Fam Physician.* Apr 1 2008;77(7):987-994.
17. Prosser LA, Ladapo JA, Rusinak D, Waisbren SE. Parental tolerance of false-positive newborn screening results. *Arch Pediatr Adolesc Med.* Sep 2008;162(9):870-876.
18. Hsu HW, Zytovicz TH, Comeau AM, et al. Spectrum of medium-chain acyl-CoA dehydrogenase deficiency detected by newborn screening. *Pediatrics.* May 2008;121(5):e1108-1114.
19. Anastasoie V, Kurzius L, Forbes P, Waisbren S. Stability of blood phenylalanine levels and IQ in children with phenylketonuria. *Mol Genet Metab.* Sep-Oct 2008;95(1-2):17-20.

## Chapters

- \* Fanos JH, Wiener L, Brennan T. Potential impact of genomic information on childhood sibling relationships. In: *Handbook of genomics and the family*, Issues in clinical child psychology, K.P. Tercyak (ed.), Springer Science, 141-61,2010.

## APPENDIX E. SUMMARY OF WORKGROUP MILESTONES YEAR 4

	<i>June 10</i>	<i>July 10</i>	<i>Aug 10</i>	<i>Sept 10</i>	<i>Oct 10</i>	<i>Nov 10</i>	<i>Dec 10</i>	<i>Jan 11</i>	<i>Feb 11</i>	<i>Mar 11</i>	<i>April 11</i>	<i>May 11</i>
Project Staff					Legal analysis completed of state approaches to PHI utilization		Annual NEGC Meeting. NEGC website launched. Redesigned logo.	NEGC Newsletter launched.	.	Reapp. for federal funding	Emerg. Prepared. Conf.	Launch of Advocacy Committee
Innovative Projects	Proposals due for 2010-2011 projects. Review process completed & awardees notified; CFN grants made available	2010-2011 Project start date							Special Advisory Mtg to revise policies (90 – 60)		LOI due for 2011-2012 apps	2010-2011 projects end
Advisory Committee	Part in review process for inno. Projects.						Annual Meeting held.		Meeting to review Innovative Projects			



Collab. Council					Began planning with reapplicati on in mind							
Evaluation						Yr 3 Stakeholde r Survey complete	National measures submitted. Yr 3 Stakeholde r Survey results released		Year 4 Annual Mtg. report released.			
Quality Improve.				BAA signed with Dartmouth.					BAA signed with Maine.			
Medical Home							Joint meeting of the Medical Home and Transition groups.			Survey of primary care providers		Joint meeting of the MH TR groups, Survey completed, Presentatio n of pilot care tool at Pediatric Acad. Societies meeting

Transition			Teen Challenge – Boston, 9 youth				Joint meeting of the Medical Home and Transition groups.					Joint meeting of the Medical Home and Transition groups. 5 new members  Report on usage of toolkits
Diss. Education & Marketing	Focus group report completed	NCC Collab. special ed project article		Poster present at NCHPEG			First Draft version of online educator's resource	Review and feedback NEGEC website	Formalization of content development for GEMSS	Meetings with Silvertech to design website		SilverTech completes website which is now ready for content
Lab QA											Chair facilitates Emer Prep meeting with other labs in attendance	Present to SACHDN C
Long-Term Follow-up*				Hemoglobinopathy conference on best practices	Presented LTFU data to 24th Annual CF Conf.		Presented LTFU data on LCHAD at NEGEC Annual Mtg.				Contract signed with Rhode Island to participate in LTFU.	

## APPENDIX F. WORKGROUP MEETINGS YEAR 4

	<i>June 10</i>	<i>July 10</i>	<i>Aug 10</i>	<i>Sept 10</i>	<i>Oct 10</i>	<i>Nov 10</i>	<i>Dec 10</i>	<i>Jan 11</i>	<i>Feb 11</i>	<i>Mar 11</i>	<i>April 11</i>	<i>May 11</i>
Management	Mtg (4)	Mtg (2)		Mtg (3)	Mtg (4)	Mtg (2)	Mtg (2)	Mtg (4)	Mtg (3)	Mtg (3)	Mtg (3)	Mtg (1)
Advisory Committee							Mtg		Mtg			
Collaborative Council	Mtg				Mtg		Mtg (annual)				Mtg	
Quality Improvement				Mtg		Mtg	Mtg (annual)					
Learning Collaborative (QILC)	Mtg			Mtg	Mtg	Mtg	Mtg		Mtg	Mtg	Mtg	
Medical Home							Mtg					Mtg
Lab QA											Mtg	Mtg
Dissemination, Education, & Marketing	Mtg		Mtg		Mtg	Mtg	Mtg	Mtg	Mtg	Mtg	Mtg	Mtg
Transition: Regional	Mtg	Mtg					Mtg	Mtg	Mtg	Mtg	Mtg	
Transition: National	Mtg			Mtg	Mtg		Mtg		Mtg			Mtg
Transition & Medical Home							Mtg					Mtg
Long-Term Follow-Up: Full							Mtg					
LTFU: Metabolic Workgroup												
LTFU: Hgb				Mtg				Mtg				Mtg

University of New Hampshire, Institute on Disability

New England Genetics Collaborative

Workgroup												
LTFU: CF Workgroup						Mtg						