

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

**Reflections on Project Activities 6/1/09- 5/31/10**

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**New England Genetics Collaborative**  
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## EXECUTIVE SUMMARY

This annual report covers the activities of the New England Genetics Collaborative (NEGC) from June 1, 2009 to May 31, 2010. 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 third stakeholder survey; an update on the status of core project components from Year Three; a list of objectives for each group for Year Four; 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, 2010. 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 third year of activity, core project staff have continued to focus on improving the infrastructure of the NEGC (updates to the website, grant applications, and structural improvements) 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. The Quality Improvement Workgroup completed the infrastructure for a genetics registry across participating clinics, began the process for creating a Patient Safety Organization, and launched a new learning collaborative initiative. The Transitions Workgroup continued to build on both regional and national level activities, including multiple efforts to integrate transition activities to adulthood into regional outreach programs, conducting a review of transition practices in the region, implementing the annual Teen Challenge Program, and working on a pilot for effective transition models for care. The Medical Home Workgroup finalized the care planning tool for use with family members, patients, clinic specialists, and primary care providers and started the process of piloting the tool in the field. The Dissemination, Education, and Marketing Workgroup completed an initial version of its classroom guide for children with genetic conditions and reviewed the work with three focus groups across the New England region. Additionally, the group expanded its role to include a review of materials for the NEGC website and, via the work of Dr. Leah Burke, began support for the National Newborn Screening Clearinghouse. Drawing from last year's efforts which

documented the need to establish laboratory specific cut offs for analysis work, the Laboratory Quality Assurance Workgroup began work on establishing cutoffs for the Connecticut lab, conducted analyses on congenital adrenal hyperplasia (CAH) disorders, and analyzed index data versus known follow up information from Connecticut, Wisconsin, and New York on multiple conditions and their corresponding analytes (markers). Lastly, the Long-Term Follow-up Workgroup achieved a major accomplishment by supporting passage of legislation in Maine which codifies the collection of LTFU data. Additionally, they continued educational efforts in Vermont and Rhode Island, and planned a national conference on improvement of long term outcomes for individuals with Sickle Cell Disease (held in Sept., 2010).

Concerning stakeholder satisfaction with the progress of the NEGC, findings from the recently completed stakeholder survey showed improvement since the previous year. A majority of respondents (N=40) understood the mission of the NEGC (67%) and felt that the NEGC has made clear and substantive progress in achieving its mission (56%). In reviewing the goals and objectives for Year Three, 90% of 64 objectives have either been completed or have made satisfactory progress in accordance with the long term goals of the grant. Objectives for Year Four have been shared and agreed to by project staff and chairs of the project's work groups. These objectives will be reviewed by the Advisory Council in preparation for Year Five funding requests. 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, and Peter Antal, Ph.D. as Project Evaluator. Administrative support is provided by the UNH Institute on Disability, which acts as fiscal agent.

In 2009 – 2010, 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 quarterly to facilitate coordination of Workgroup activities. The RCC and Collaborative Council are guided by an Advisory Council 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

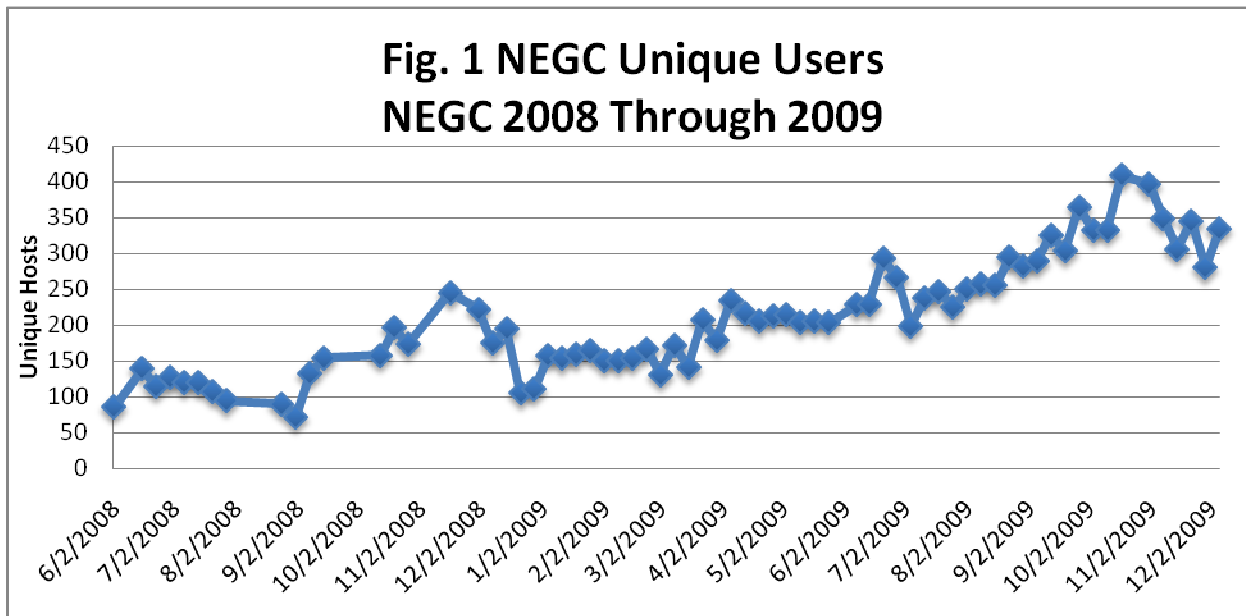
The NEGC staff built on the website capacity and functionality, sought to leverage new grant resources to support achievement of the NEGC mission, and made substantial structural changes to the work of the NEGC.

### Website Improvements

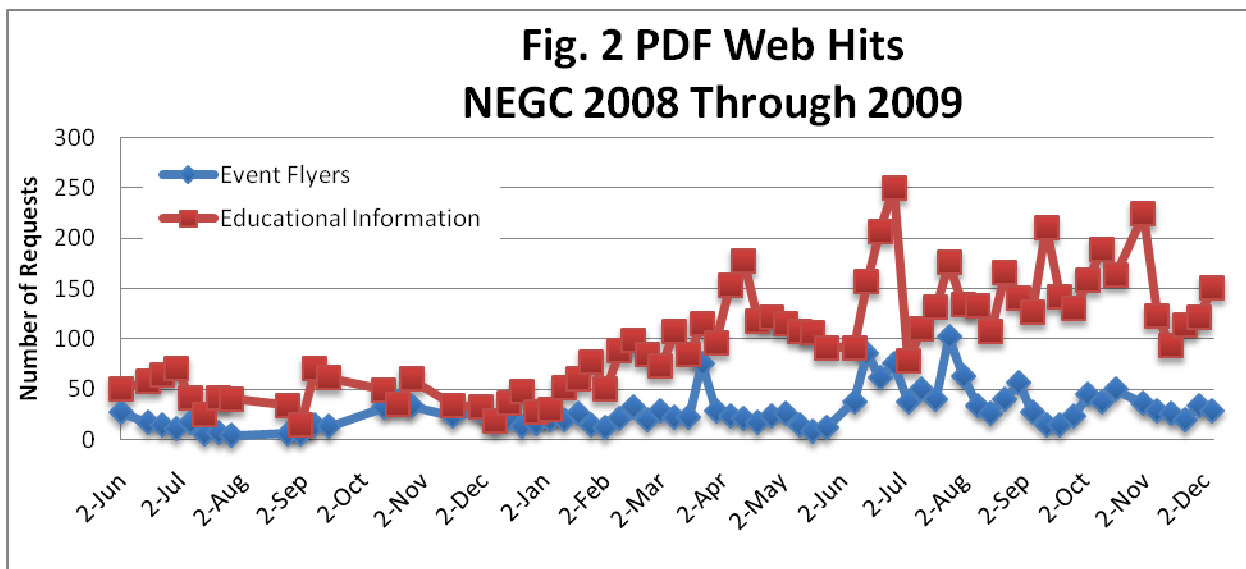
During the course of the project year, project staff continued providing updates to the project's website. Enhancements were made in the following areas: updating the review process for innovative grants, creating a section for community grant applications, providing substantive updates on the work of the Transition group, enhancing the staff page, and re-organizing the presentation of materials for families and professionals. Through December 2009<sup>1</sup>, there was a gradual increase in unique users accessing the NEGC website each week; from 85 in the first week of June, 2008 to 335 in the last week of November, 2009 (See Fig. 1).

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<sup>1</sup> Due to the proposed website restructuring, data reporting had been stopped by the website provider effective December, 2009.



There was an increase in the average number of event flyer requests (from 20 to 42 per week) while requests for informational resources on genetics issues continued to increase substantially in 2009 (averaging from 69 to 147 per week - Fig. 2).



The highest number of requests for educational information was for the week ending June 28, 2009 (250 requests). Resources with 10 or more requests during this week included: Newborn screening brochures (arabic, portugese, simplified chinese), NCC Collaborator (March edition), Genetic Deafness Booklet, Genetic Hearing Loss Booklet in Spanish, Genetics Directory, Genetic Information Nondiscrimination Act of 2008, Prenatal Guidebook, and the Understanding Genetics guide.

#### Resource Leveraging

During Year Three, NEGC staff submitted one grant application to support new or expanded work in genetics in the New England region. As of Oct 1, 2010, the application was funded for \$45,000 to support

legal analysis of cross-state data sharing and to launch the new learning collaborative. In addition, the NEGC provided 2 letters of support for projects led by the Genetic Alliance.

<b>Direct Applications</b>		
<b>Grant Name</b>	<b>Description</b>	<b>Amount</b>
Administrative Supplemental	HRSA; funds for legal analysis work and creation of the learning collaborative.	\$45,000 FUNDED June 2010
<b>Letters of Support for Partner Applications</b>		
<b>Grant Name</b>	<b>Description</b>	<b>Amount</b>
Clearinghouse of NBS Information	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. Dr. Leah Burke serves on the project Advisory Committee
Congenital Conditions Program	The NEGC supported an application by the Genetic Alliance and Family Voices.	Funded to Genetic Alliance.

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

### **Structural Changes**

Major structural changes in the NEGC include the addition of a new funding cycle to support innovations at the family and community level in the New England region (described in the next section), preparing for a complete redesign and structural change of the NEGC website (new site will be live by Dec. 1, 2010), launching a new learning collaborative effort (see QI Workgroup activities), and the addition of the new project manager and co-PI, Monica McClain, Ph.D.

### **Collaborative Activities**

Project staff have actively sought new opportunities for partnerships with both regional and national partners. During Year Three, this included: joint planning of the annual meeting with NERGG, 3 presentations and 12 publications by affiliated staff, 4 newly funded innovative projects, 7 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. 2-3, 2009

The annual meeting was well attended by forty-six partners in the initiative. During the 2 day event, staff presented on some of the major accomplishments of the past year (finalization of the medical home pilot, new tools to aid in transition activities, continued refinement of the special educator tool, multiple presentations and publications), and highlighted major activities to be undertaken for next year (expansion of the medical home pilot, implementation of a data registry for quality improvement, continued refinement of marker analysis in lab testing). Breakout sessions on how to effectively engage with and provide information to consumers were held. In addition, open workgroup meetings were held that enabled cross-group and new stakeholder participation in the activities of individual workgroups.

Concerning the overall work of the initiative, members identified a range of both national and regional challenges that need to be addressed, including: the lack of national funding focused on adults, the role of Medicare changes and their influence on genetics systems, various levels of interest and knowledge among providers in the genetics field, and the need to practice medical care more explicitly. While challenges were identified, opportunities were offered as well, including: new tools for consumers, the potential to expand core project areas (e.g. medical home pilot), and helpful resources for improving care (e.g. Mountain State Care Plan). A copy of the full meeting 'mid-year' report is available at <http://www.negenetics.org/reports.html>.

## Presentations and Publications Supported by the NEGC

During Project Year Three, NEGC coalition stakeholders conducted 3 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 presentation on Cystic Fibrosis: "Recommendations to Increase Newborn Screening Efficiency" to the 7th International Congress, Latin American Society of Inborn Errors of Metabolism and Neonatal Screening. Both were conducted by Dr. Anne Comeau. The third presentation was by Dr. Susan Waisbren during a conference on PKU 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 Three, 12 additional publications were created by NEGC collaborative council members, bringing the total publications list of NEGC stakeholders up to 22. The most recent publications include:

### ❖ Dr. Susan Waisbren

- *Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders.* Mol Genet Metab.
- *Projected costs, risks, and benefits of expanded newborn screening for MCADD.* Mol Genet Metab.
- *Impact of false-positive newborn metabolic screening results on early health care utilization.* Mol Genet Metab.
- *The psychology and neuropathology of phenylketonuria.* Mol Genet Metab.
- *Final commentary: a new chapter.* Mol Genet Metab.
- *Screening for cognitive and social-emotional problems in individuals with PKU: tools for use in the metabolic clinic.* Mol Genet Metab.
- *Psychosocial issues and outcomes in maternal PKU.* Mol Genet Metab.
- *Psychiatric symptoms and disorders in phenylketonuria.* Mol Genet Metab.

- ❖ Dr. John Moeschler, Dr. Tom Brewster, Dr. Wendy Smith, Dr. Rosemarie Smith, Dr. Leah Burke, Dr. Mary Beth Dinulos, and Dr. Steven Amato
  - *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.
- ❖ Drs. McGrath, Laflamme, Moeschler & Amy Schwartz.
  - *Access to genetic counseling for children with autism, Down syndrome, and intellectual disabilities.* Pediatrics.
- ❖ Dr. Carl Cooley
  - *Medical home 2009: what it is, where we were, and where we are today.* Pediatr Ann.
  - *Improved outcomes associated with medical home implementation in pediatric primary care.* Pediatrics.

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

### Innovative Projects

The RCC continued to build on the innovative projects program and completed its third round of grant funding. The NEGC received nine proposals and awarded three grants, with a combined total disbursement of \$90,000. The studies funded by these grants include:

- **“The Personal Transition Health Plan Project,”** submitted by Dr. Susan Waisbren of Children’s Hospital, Boston. The long-term goal of this project was to develop a model that ensures that every young adult patient seen at a genetics or metabolic clinic has thought about and documented a plan for ongoing health care that addresses the specific needs of his or her condition, focusing on issues as they appear in adulthood. Amount funded: \$30,000.

The project focused on the following four areas: conducting an environmental scan of national transition practices, finalizing the transition toolkit (comprised of a health readiness assessment, fact sheets on five conditions, a medical health summary, and a transition plan). All of the instruments were designed to be easily understood and easily used via good website design. The forms can be downloaded onto a flash drive and taken along to a doctor visit. The one-page fact sheets were developed after patients noted their frustration at having to repeatedly explain their condition; feedback from pediatricians suggested the forms would be useful to them as well. All of the instruments are free and available to anyone.

- **“Development and Initiation of a New England Birth Defects Consortium,”** submitted by Stephanie Miller of Dartmouth Medical School. The goal of this project will be to form a regional consortium to facilitate new project development 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 conditions by promoting regional collaboration in surveillance data sharing, birth defects research, prevention activities, and health care quality improvement. Amount funded: \$30,000.

As a result of funding and member efforts, the Consortium has:

- enabled members to participate in assessment of research and grant opportunities, to discuss data standardization among the states including the possibility of creating regional prevalence data, and to share ideas related to the day to day operations of birth defects programs;
- begun review and aggregation of detailed program operations and case information for all member states;

- developed collaborative partnerships with the Maine Birth Defects Program and the Maine Department of Environmental Services to pursue a regional case-control study;
  - shared its work with the National Birth Defects Prevention network to promote similar work in other state birth defects programs.
- **“Developing Parent-Provider Partnership Plans for Children Referred to Specialty Care,”** submitted by Dr. Chris Stille of the University of Massachusetts Medical School. The goal of this project was to further develop and pilot test a care planning tool, based on the Medical Home model of care to increase parents’ capability to share medical information and plan care with primary care physicians and specialists. Amount funded: \$30,000.

During the project year, staff pursued two major goals: 1) fine tune a prototype care plan and develop a brief educational intervention using frameworks adapted from adult chronic illness care; and 2) pilot test the intervention for feasibility in a diverse sample of pediatric practices.

In fine tuning the prototype care plan, staff facilitated five physician focus groups, began qualitative data analysis of these groups and fine-tuned the intervention form, presented the form and “5 A’s” tool for physicians to four parent experts, and interviewed them to gain further information on how to polish the care plan form and the educational intervention. The care plan has been converted into a web-fillable PDF form.

A pilot test was begun in two pediatric primary care practices and three pediatric specialty practices at UMass (genetics and metabolism, neurology, and endocrinology) in the Spring of 2010. While the target for the year was to have 40 patients enrolled, multiple barriers (including when the pilot started, lack of full participation by doctors and a low number of patients who qualified for the pilot) resulted in poor enrollment. Despite the slow start up of the pilot, physicians and particularly parents have found the care plan useful, though they acknowledge it adds 2-5 extra minutes to a visit. Dr. Waisbren’s group has adapted the care plan for use in their clinic setting, adding detail needed for patients with complex metabolic conditions, and found it helpful, but again difficult to complete in the context of an already busy visit. Data collection will extend into Year Four of the NEGC project as feasibility data is collected and new sites are identified to participate in the pilot of the care planning tool. Team members recommend that a process be created to begin integrating the care plan into an electronic medical record to avoid duplication of work, and to ensure that practices have a care coordinator or other coach available for brief education about how to use the plan.

- **“Thyroid Dysfunction: Long Term Follow Up of Very Low Birth Weight and Extremely Low Birthweight Infants”** submitted by Dr. Chanika Phornputkul of Rhode Island Hospital. The project sought to analyze newborn screens across multiple time periods for incidence of Congenital Hypothyroidism with a delayed TSH elevation in extremely low and very low birth weight infants. Amount funded: \$32,000 (funding provided during previous project year).

The project documented that the incidence of CH with a delayed TSH elevation is significantly higher in ELBW and VLBW infants compared to infants with weight greater than or equal to 1500 grams. It was noted that outcomes of VLBW infants with CH with a delayed TSH elevation were comparable to controls. Project staff recommend that, since outcomes of children were comparable to control when delayed TSH elevation is detected, the practice of rescreening in premature infants should continue.

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 Grants**

During its 2009 annual meeting, the NEGC Advisory Council requested that resources be developed that would support community and family level innovation and participation in genetic services. In response, the NEGC created the Community and Family Network grant initiative. During 2009, \$10,000 was set aside from grant funds for the purpose of developing small community-based projects that address the needs of people with genetic conditions and their families within the mission of the NEGC. Seven applications have been approved, supporting: attendance at regional Family Voices meetings, enhancing organizational annual meetings, and website development.

### **Special Projects**

During the course of its work, the NEGC pursues and engages in a range of opportunities to improve the field of genetics education and services. During Year Three, the NEGC supported work in the following areas: assessing genetic workforce capacity, reviewing emergency preparedness protocols in New England, and supporting additional training for nutritionists.

#### *Assessing Genetic Workforce Capacity*

Led by Dr. Robert McGrath from UNH and in partnership with the American College of Medical Genetics, this project seeks to document the potential impacts of expanded newborn screening protocols on the genetic workforce. During the project year, the NEGC and ACMG collaborated on collecting background data and refining the study's aims and methods. The analyses and final report will be completed in Year Four.

#### *Emergency Preparedness in New England*

During the project year, Dr. Roger Eaton began laying the groundwork for an emergency preparedness conference in New England (to be held in Spring of Project Year Four). As a part of this effort, Dr. Eaton attended two national level conferences and has begun consultations with key partners in preparation for the 2011 New England conference.

#### *Training for Nutritionists*

The NEGC identified a need for training of metabolic nutritionists who were new to the field, served in a back-up role, and who didn't have other support to attend educational conferences. To help fill this gap, the collaborative sponsored seven nutritionists from the region to attend GMDI training in April 2010.

### **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 Council*

The NEGC have 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.
- Provider Network: Anne Comeau, Ph.D.
- Publications: John Moeschler, MD

### *Genetic Alliance*

The NEGC supported the Genetic Alliance on two of their projects, both of which were funded in September, 2009. The first involves the creation of a Clearinghouse of NBS information with Leah Burke, MD, serving on the advisory board. Secondly, the NEGC wrote a letter of support for the Genetic Alliance focus on a Congenital Conditions project.

### *Other Groups*

- Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
  - LTFU Sub-committee: Carl Cooley, MD
  - Health Information Technology Workgroup: Roger Eaton, Ph.D.
  - Evidence Review: Anne Comeau, Ph.D.
- National Transition Interest Group: Susan Waisbren, Ph.D.
- National Clearinghouse (Genetic Alliance): Leah Burke, MD
- Newborn Screening Translational Research Network
  - Clinical Centers Workgroup: John Moeschler, MD
  - Laboratories Workgroup: Roger Eaton, Ph.D., Anne Comeau, Ph.D.
  - Joint Committee of the NBSTRN Clinical Centers Workgroup and the NCC/RC Long-Term Follow-Up Workgroup: Anne Comeau, Ph.D.

## WORKGROUP ACTIVITY IN YEAR THREE

This section provides an overview description of each workgroup's activities during Year Three. 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 seven members and is led by the NEGC's Principal Investigator, John Moeschler, MD. They met in June, October, and December of 2009 and in March of 2010. During the project's third year, workgroup members focused their efforts on three major areas. First, members worked on the creation of a data registry that can handle web-based data input and reporting from multiple clinic sites. They also pursued the establishment of a Patient Safety Organization (PSO) that would enable the cross-site sharing of medical information for improving patient health outcomes. Lastly, work group members laid the ground work for a new learning collaborative to be established among the members of the metabolic centers.

In their efforts to create a registry, members built on the data points of interest and general framework established during the past two years and worked with Global Vision Technologies (GVT) to create a data entry and reporting structure into a web-based registry. This will be ready to use pending successful creation of the PSO. The Dartmouth Committee for Protection of Human Subjects determined that this work is quality improvement and not human subject research. HIPAA Business Associate Agreements are in process among Dartmouth, GVT and UNH to protect health information and participants appropriately.

The creation of a PSO for the New England region is important as it will enable staff at centers affiliated with the PSO to share and learn from patient data at each of their clinics. Once in place, staff will be able to gain a much better understanding of how their own medical practice compares with those of their peers, both in terms of process quality and health outcomes. This will help them to take a closer look at their own practices to determine how their care can be improved. During Year Three, staff met with multiple partners to create a PSO within existing infrastructures but have been faced with multiple challenges along the way. While this part of the project is still in process, it is anticipated that a PSO will be formally created during Project Year Four.

The other major new initiative for the year concerns the creation of a Learning Collaborative among Metabolic Centers. In the context of health care, a Learning Collaborative is a group of providers that have agreed to work together to review and discuss health care practices at their respective clinics with the purpose of identifying and supporting the implementation of best practices in health care improvement (for more detail, please see:

<http://www.ihl.org/IHI/Results/WhitePapers/TheBreakthroughSeriesIHIsCollaborativeModelforAchieving+BreakthroughImprovement.htm>). During Year Three, project staff formed a planning group, created a draft charter that would guide the implementation of the collaborative, and identified the first two conditions

(PKU and MCAD) on which the Learning Collaborative would pursue further work during Project Year Four.

## The Transitions Workgroup

The Transitions Workgroup is led by Dr. Susan Waisbren, who is also the leader of the National Transitions Interest Group. The group currently has 13 members. Dr. Waisbren has been working with a subgroup of the New England Consortium of Metabolic Programs to conduct the work of the group. This subgroup met eight times during Year Three. Six members of this subgroup also participated in the monthly meetings for the National Transition to Adult Care interest group which met eight times via conference call during the year. In Year Three, workgroup members focused their efforts on four major areas: education and outreach, conducting a review of transition practices in the region, defining a best practice training protocol for metabolic patients, and piloting a model for ensuring effective transitions in care.

In the area of education and outreach, multiple steps were taken to integrate transition activities to adulthood into regional outreach programs. This included presentations at the National Urea Cycle Disorders Study and National PKU association, publication of five articles in *Molecular Genetics and Metabolism*, the posting of the transition toolkit on the New England Consortium of Metabolic Programs website, support for two students' work on galactosemia, as well as a conference for adults with galactosemia (held in conjunction with a research study on outcomes in adults with this disorder, the first prospective study of its kind ever conducted on that population). Of note, the Transition Toolkit includes four components: a health readiness assessment, basics on metabolic conditions, a medical health summary, and a transition plan. As part of the basics on metabolic conditions, five fact sheets were created with the lay reader in mind. These include documentation on: Biotinidase Deficiency, Galactosemia, Homocystinuria, Ornithine Transcarbamylase Deficiency, and Phenylketonuria. Two graduate students were supported to create three new informational resources on galactosemia: [Moving Forward: Your Guide to Galactosemia and Primary Ovarian Insufficiency \(POI\)](#), [Understanding Galactosemia](#), and [Understanding Galactosemia: Resources for Educators](#).

The Transitions Workgroup conducted a review of transition practices in the region and found one or more examples in each state, including: efforts by Brigham and Women's hospital in Boston on maternal PKU activities and the Transition network; a Healthy and Ready to Work initiative in Maine; a liaison position in public health in Vermont; a new National Transition Center being created in New Hampshire; the Parent Information network in Rhode Island; and a range of work conducted by the public health department of Connecticut.

In continuing to pursue refinement of their training models, Dr. Waisbren again led the implementation of the "Teen Challenge Program." This program seeks to enhance the training currently offered to teens by providing a range of tools for coping and managing their conditions. Eighteen youths participated in a variety of activities designed to challenge them to try new things, build self-confidence, develop relationships with other adolescents with metabolic conditions, and develop skills to manage their health. One of the weekend highlights was the "Iron Chef Low-Protein Cooking Challenge," in which teams of youth worked together to create great-tasting food that fit their dietary restrictions. In a parallel effort to support leadership and connections among youth, the NEGC provided funding for technical support to an individual who

successfully created the PKU Challenge, a website designed to connect people with PKU ([www.thepkuchallenge.com](http://www.thepkuchallenge.com)).

Lastly, in working on piloting effective transition models to care, the workgroup focused on finalizing two sets of documents that would aid in transition efforts. Part of their work supported the Medical Home workgroup's goal of developing communication tools that could be used with parents/ primary doctors/specialists. Work on this effort led to improvements on their Transition Plan efforts. Their goal was to develop a model that ensures that every young adult patient seen at a genetics or metabolic clinic has thought about and documented a plan for ongoing health care that addresses the specific needs of his or her condition as they appear in adulthood. To date, the forms and process have been developed and the group is looking forward to running a formal pilot implementing the plan during Project Year Four.

### **Medical Home Workgroup**

During Year Three, the Medical Home Workgroup was led by Dr. Carl Cooley. The diverse membership of 18 staff includes primary care physicians with medical home experience, specialists from regional metabolic disease programs, and parents of children with metabolic disorders. The group held conference calls in June, March, and May and held a face to face joint meeting with the Transition Group in December.

Primary efforts of the Workgroup focused on the final design and implementation of a care planning tool for use by families, physicians, and specialists. Creation of the tool involved a unique collaboration between UMass Medical School, the Center for Medical Home Improvement, Children's Hospital Boston, the New England Genetics Collaborative, and Mass Family Voices. The care plan is a one-page form in a fillable PDF format which requires the parent and doctor to explicitly note together what has been done and what concerns or requests should be related to the specialist. In addition to the team review of the tool and previous parent focus groups, five focus groups were held with physicians to further refine the tool.

The evaluation phase of the project began in the spring, but, unfortunately, encountered challenges due to a lack of sufficient new referrals meeting the project's eligibility criteria, as well as inconsistent follow-through by participating physicians in utilizing the new tool. Members of the workgroup are planning on continuing the pilot phase of the instrument during Project Year Four through current collaborations with Dr. Susan Waisbren, as well as invitations to new clinics to participate in the pilot (during the 2010 New England Metabolics Consortium Meeting).

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

The ten members of the Dissemination, Education, and Marketing (DEM) Workgroup, which met six times during Year Three, are led by Dr. Leah Burke. During Year Three, major activities of the group focused on supporting the creation of a Newborn Screening Clearinghouse, supporting access to effective resources on the NEGC website, and further refinement of a special educator's tool for children with genetic conditions.

Work on the Newborn Screening Clearinghouse involves a partnership between Dr. Burke, as chair of the DEM Workgroup, and the Genetic Alliance. The end result of this partnership will be a 'one-stop-shop' website for any information related to newborn screening.

New in Year Three, this workgroup has agreed to review and recommend potential resources for inclusion on the NEGC website. The group recently reviewed a resource from the March of Dimes; as a result the group determined the resource was outdated and not appropriate for the NEGC website.

Lastly, much of the group's energy has been focused on the continued refinement of a web-based guide for educational teams working with children with genetic conditions: *Children with Genetic Conditions: A Guide for the Classroom*. The guide helps educators to deal with a range of scenarios, for example: child needs supports for pain; child fatigues easily; child has dietary/medical needs; child's conditions declines/changes; and child will attend special functions. Each of these domain areas are linked to a range of useful informational items that can be used to ensure the most appropriate resources are in place for children with genetic conditions. As part of its review process for this year, this workgroup collaborated with the UNH Survey Center to conduct three focus groups with elementary school educators, special educators, para-professionals, occupational and physical therapists, speech specialists, and school nurses in Connecticut, New Hampshire, and Vermont. In looking forward to Year Four, the group will work on posting the resource to the website and expanding the number of conditions that are included.

### **Laboratory Quality Assurance Workgroup**

The Laboratory Quality Assurance (QA) Workgroup has ten members and is led by Dr. Roger Eaton. The members work regularly in collaborations between the New England Newborn Screening Lab and other state labs based in New York, Connecticut, and Wisconsin. QA members as well as members from the participating states collaborated during the year via phone, email, and webex meetings.

QA Workgroup members, following what they learned about the need for lab specific cut-offs to be created for most markers, focused on collection and analysis of 50,000 raw marker data from partners in Connecticut and 10,000 from Wisconsin. Connecticut specific cut offs were then created for VLCAD, LCHAD, and CPT2. Additionally, utilizing NENSP data, analysis was done on CAH disorders and out of range initial screens. Results of this effort were not supportive of further work in this area, so the group will reorient its energies in Year Four on developing the state specific index cut offs and continued analysis of marker concentrations. Lastly, the team analyzed index data vs. known follow-up information from Connecticut, Wisconsin, and New York on IVA, 2MBG, VLCAD, LCHAD, and CPT2 (C5, C14, C14:1, C14:2, C16OH, C18:1OH, C16, C18:1).

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

The Long Term Follow-Up (LTFU) Work Group has 8 members and is led by Dr. Anne Comeau. The full group met four times during Year Three, and the Hgb workgroup met four times.

During Year Three, LTFU members focused on educational efforts with state legal and privacy officers. As a result of their efforts to date, the workgroup reports that significant progress has been made in establishing the authority to collect LTFU data (MA, ME, RI) as well as in data collection (MA and ME). Additionally, the New England Newborn Screening Program submitted a manuscript to Genetics and Medicine documenting strategies for sustainable LTFU systems based on their work over the past 10 years and collected data on metabolic cases, CF, and hemoglobinopathies.

The Hemoglobin Workgroup began review of data on current patients, including information on hospitalizations, Transcranial Doppler (TCD) practices, and occurrences of strokes. As a result, tracked data

elements have been modified and will be included in future reporting. Lastly, the group planned for a conference in the fall of 2010 to focus on LTFU efforts. The "Surviving to Thriving: Improving Long Term Outcomes in Sickle Cell Disease" conference will bring various experts from the around the country to identify best practices for improvements in patient care.

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

As part of its focus on developing best practices in long term follow up, the NEGC has continued to support the work of Drs. Waisbren and Fanos on improving the psychosocial aspects of LTFU. Dr. Waisbren continues to take the lead on developing the genetics and metabolic network. And, Dr. Fanos has taken the lead on developing the psychosocial workgroup that now has 15 members, all from New England. As a result of their work during the year, the Genetics and Metabolism Psychology Network or GMPN ([www.gmpsynch.org](http://www.gmpsynch.org)) is firmly established, with over 25 members. The website is up and funds have been received from outside of the NEGC to revise it next year. To date, the membership is about 25% from New England. In February, 2011, members of the GMPN will discuss the establishment of a Universal Neuropsychology Follow-up Screening Method for all disorders identified by newborn screening. This discussion will take place at a meeting in conjunction with the International Neuropsychology Meetings. Psychologists from the Urea Cycle Consortium as well as the GMPN will attend these meetings which will take place in Boston.

The Universal Assessment Method, comprised of three questionnaires for parents selected by the GMPN at the inaugural meeting in 2008, was presented to the New England Consortium of Metabolic Programs. Sample copies were distributed along with information on how to obtain the questionnaires. A grant was submitted to the NEGC to pilot this method via phone interviews with all families of children identified by newborn screening in New England. This study has been funded in Project Year Four and approved by the Children's Hospital IRB and the first subjects have been scheduled for January, 2011.

Considering Galactosemia specifically, Dr. Waisbren has pursued submission of a manuscript for the Galactosemia follow-up study as well as a grant submission to NIH and to the Parents of Galactosemic Children support group to conduct a comprehensive study of outcomes in galactosemia, with one of the aims to assess psychosocial adjustment.

In addition to these activities, Dr. Joanna Fanos has continued her work on two areas: studying the impact of genomic information on childhood sibling relationships as well as psychosocial follow up of newborn screening. Published in 2009, her work on genomic information summarizes the current knowledge on family communication, sibling relationships, parental mourning, sibling guilt and shame, as it relates to several serious pediatric genetic disorders and their impact on siblings. The work outlines the challenges faced by siblings of children with known genetic conditions and recommends further review and education among medical professionals concerning the impact of pediatric illness on siblings and the need for effective models of support. Work on psychosocial follow up of newborn screening was carried out during 2009 and is expected to be published during Project Year Four. The focus of the research is on documenting the multiple methods for providers to relay news of a genetic condition and its implications to parents at the beginning of a child's life and the steps that can be taken during follow up appointments to regularly assess the well being of parents. The project documented some of the concerns that arose among providers and families and provides recommendations on needed resources and research that could be helpful in aiding families'

transition into post diagnosis life. A report was provided by Dr. Fanos on the recommendations of the psychosocial workgroup.

## **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 Three, the following areas were addressed:

### **Ethical Issues**

Much of the combined work of the Transition and Medical Home Workgroups focused on changing the practice of care such that families have direct participation in the health care that they receive. This will be accomplished through encouraging greater participation by family members in setting a common understanding with pediatricians and specialists about next steps in the care of their children.

### **Legal Issues**

Advances in this area were made in two ways: efforts by the QI workgroup on developing a registry and the potential development of a Patient Safety Organization, and LTFU efforts to work with state privacy officers and legal teams to create an appropriate environment for the ongoing collection of LTFU data for the purposes of health care improvement.

### **Social Issues**

There were three ways the NEGC impacted on the social context of healthcare for individuals with genetic conditions. 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. Funding for community grants helped 45 individuals to gain training via regional conferences. Lastly, the special educators guide, once completed, will substantially improve the quality of interactions between educators and students.

### **Policy Issues**

As a result of LTFU work in Maine and Massachusetts, the ability to share LTFU data has now been formally established in state law. This will 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.

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

## **Evaluation Activities**

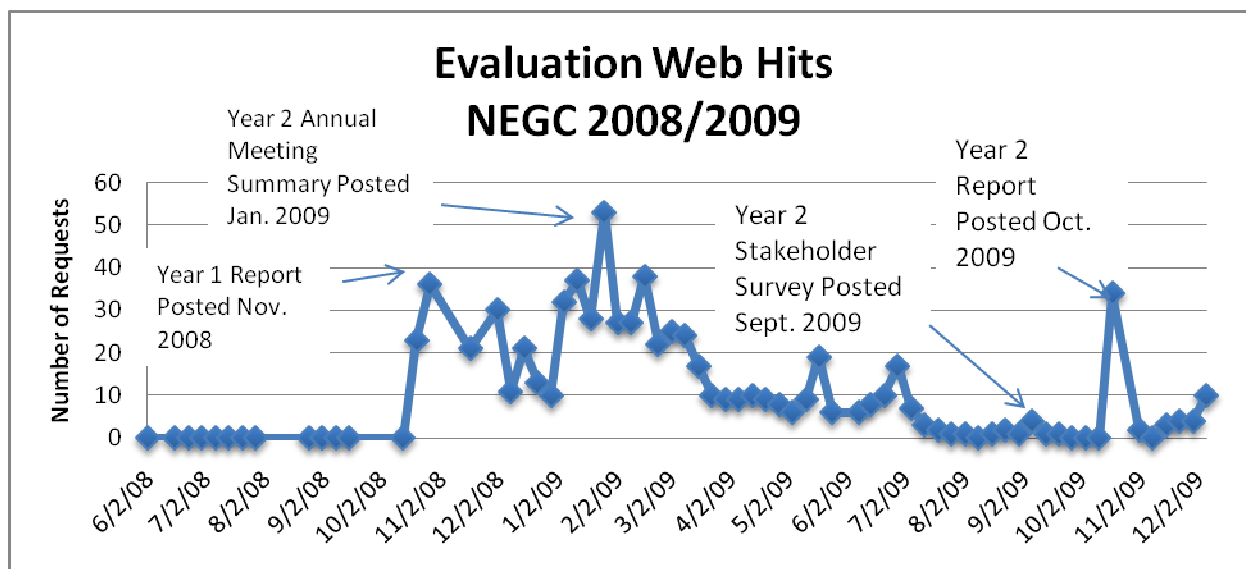
During Year Three, the evaluation of the NEGC was led by Peter Antal, Ph.D. Primary roles during the third year of the collaborative focused on: providing ongoing review of activities, summarizing project activities via evaluation reports, promoting coordination with the national evaluation initiative,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

## Ongoing Review of Activities

Peter Antal has actively participated in NEGC meetings, including ongoing planning meetings and meetings with the Collaborative 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. A new goal for the year has been to support the establishment of the new Learning Collaborative led by the QI Workgroup. Efforts in this area have focused on providing ongoing evaluation supports for the group, assisting in the design of new medical forms, and strategic planning.

## Evaluation Reporting

During the past year, the Year Two Project Report and Year Three Annual Meeting report was posted as was the Year Two Stakeholder survey. All reports were provided to staff for review and feedback prior to final publication as public documents on the project's website. As shown in Figure 3 below, requests for evaluation reports during Project Year Three peaked in October with 34 requests.



## 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 for the most recent year 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:

- NEGC reviewed the use of the Newborn Screening Toolkit and the New England Genetic Resource Directory developed during the first year of funding. These projects involved genetics education and dissemination across the region and are specifically targeted towards PCPs.
  - Stephanie Miller, via the RC's innovative projects grant program, initiated a regional 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.
  - Dr. Susan Waisbren, also with support from the innovative projects program, is leading the Personal Transition Health Plan Project at Children's Hospital, Boston. The long-term goal of this project is to develop and pilot a practice model that ensures that every adolescent and young adult patient seen at a genetics or metabolic clinic has thought about and documented a plan for on-going health care that addresses the specific needs of his or her specific condition, with a focus on symptoms that are relevant to them as adults.
  - In Vermont, NEGC provided support for Dr. Mark Korson to provide education and consultative services to the pediatricians and to Dr. Leah Burke. Through this collaboration, Dr. Korson provides monthly lectures to medical students and residents. He has also given multiple Grand Rounds in Pediatrics and Internal Medicine to help make the practitioners aware of rare metabolic conditions that may present to their offices. Dr. Korson is engaged similarly at several medical centers in New England.
  - Work carried out through Dr. Anne Comeau's Long Term Follow Up project seeks to ensure that health care providers have access to information that is critical for maintaining appropriate care plans and improving the long term health of individuals living with genetic conditions.
- ❖ 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: NA. NEGC did not provide support in this area during this period.
  - ❖ 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.
  - ❖ 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 (those diagnosed with condition(s) 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). By 2011, Maine will have systems in place for all areas except for hearing.
- ❖ 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.

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

For Year Three, the evaluator again worked with project staff and collaborative council members to update and implement the NEGC stakeholder survey. The survey was administered online between October and November 2010. A summary of the results follows.

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

*[Executive Summary excerpted from New England Genetics Collaborative, Results of the Stakeholder Survey for Project Year Three by Peter Antal, Ph.D. (December, 2010). For the full report, please download from [www.negenetics.org](http://www.negenetics.org)]*

To facilitate feedback from its partners, the NEGC conducts an annual survey to identify concerns, document how the project is doing, and solicit suggestions for improvement. Seventy-two email invitations were sent out between September and October 2010 to stakeholders of the New England Genetic Collaborative (NEGC). Of these, 42 (58%) provided responses.

Since last year's report, there was slight improvement in two important areas. When asked whether they had a clear understanding of the NEGC's mission, 67% agreed (vs. 60% in 2009). Concerning whether the NEGC had made substantive and clear progress in achieving its mission, 56% agreed (vs. 47% previously). Feedback on the project's evaluation reports were generally positive with 48% to 71% of respondents indicating that each of the reports helped them understand the progress and challenges of the initiative. Respondents suggested a wide range of information that would be useful on the NEGC's new website as well as in future updates provided by the NEGC.

Feedback from the Advisory Council was high this year, with 13 members participating. Most participants 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. The number of responses from two workgroups (Dissemination, Education, & Marketing and Transition) were high enough to include in this year's report. Fifty percent or more of these two groups agreed that there was good: collaboration and cooperation across meetings, that meetings were well run, that the RCC provided excellent support and that the workgroup was achieving its main objectives. There was less agreement in both groups around the effective use of data on the group's outcomes.

When asked about the strengths of the collaborative, several common themes emerged across survey respondents, highlighting: leadership capacity, collaborations, diversity of respondents, project focus, skills, administrative supports, commitment, and other.

Concerning some of the obstacles faced, the most frequently cited challenge was the potential for competing interests to tie up resources, better clarity around accomplishments, greater diversity among membership, need for better communication (particularly across groups), limited diversity, funds, time, the need for better staffing and collaboration building support, and other.

Respondents were also asked about what the NEGC could do to provide better support. In response, most members focused on the continued need for administrative support (managing meetings, reports, tracking objectives), identifying new collaboration opportunities and other.

## **COMPLETION OF OBJECTIVES IN YEAR THREE**

The following table 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, 2010. 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 individual efforts during Project Year Three. Review results are defined as:

- ❖ Successful (45 of 64): Definition of success for the year has been fully met or the results of the activity in question fulfill the intent of the measure.

- ❖ Partially Successful (13 of 64): The definition 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 (6 of 64): Although some work on an activity may have been done, primary components of an activity targeted for the year were not fulfilled 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 Three.

<b>ESTABLISH AND MAINTAIN NEGC</b>				
<i>No.</i>	<i>Objective</i>	<i>Project Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Continue implementation of core administrative supports to the NEGC	3	NEGC is able to successfully pursue goals and objectives and appropriately responds to changing conditions.	Review: Successful  All core staff activities completed during course of year.
2	Continue close collaboration with WG and AC	3	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.	Review: Successful  Meetings are held regularly and supports provided when requested as resources allow. A majority of survey respondents from the Advisory Council, DEM Workgroup, and Transition Workgroup indicated that the RCC provides excellent support (85%, 100%, and 67% respectively).
3	Develop and implement a communications and outreach plan for the NEGC	1	Number of stakeholders (both professional and consumer) increase annually and maintain their participation in the NEGC.	Review: Successful  Between Yrs 2 and 3, participation of stakeholders (defined by mailing list) increased from 68 to 75. Communication plan created with Kathy Beal.
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	Review: Successful  Stakeholders received 4 quarterly updates, mid-year report, and annual

			project developments.	report describing project progress.
			Website is utilized by growing numbers of individual users.	Review: Successful Between Year 2 and Year 3 December, average weekly unique users increased from 160 to 294.
5	Implement Special Projects	3	Complete research on newborn screening experiences of parents.	Review: Successful Research completed and published effects of genetic conditions among siblings of children with genetic conditions. New article in development looking at parent perspectives of the diagnosis and follow up process and recommendations for improvement.
			Genetic Workforce study	Review: Partially Successful Interviews conducted and preliminary analysis completed. Final analysis to be completed Sept. 2010.
			Emergency Preparedness Conference	Review: Partially Successful Participation in national conferences to establish a conference model for NH. Planning underway for a conference tentatively scheduled for April 1, 2011.
			Training Activity with Mayo Clinic	Review: Unsuccessful. Lack of success due to insufficient interest by members in participating in training activity, as well as members who had access to travel funds from their own institutions.
			Nutritionist Training	Review: Successful Training held on 4/15-4/16 for 7 dieticians from the New England region.

<b>QUALITY IMPROVEMENT</b>				
<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Complete quality improvement analysis for one site	1	Analysis completed	Review: Successful  Data sets refined, data dictionary developed, assessments of reporting mechanisms at each center conducted.
2	Implement QI report structure	3	Data report-format completed	Review: Successful.  Basic format and reporting structure completed.
			Data by site and region reported on provider websites	Review: Unsuccessful  Further work on this area is dependent on establishment of a PSO or establishment of an exemption letter from each site's CPHS (committee for protection of human subjects).
3	Create a Patient Safety Organization (PSO) to host data collected from clinic sites	4	Approval received.	Review: Partially Successful  Despite multiple attempts by staff to solicit creation of a PSO by major entities (Dartmouth, ACMG), the creation of a PSO is challenged by multiple hurdles. While progress has been made in this area and relevant partners have been involved, formation of the PSO is not expected until sometime in Year 4 of the grant. The group is also looking at alternatives, such as an exemption letter from each site's CPHS.
4	Implement clinical genetics pilot registry to aggregate clinical outcomes data	3	By end of Q2, all five sites will be entering data each week or month.	Review: Partially Successful  Approval to enter data has been received by Dartmouth. Additional clinics will be added in Year 4 pending creation of a PSO or exemption letters from each site's CPHS.

			By end of year, all five sites will be entering data on 100% of appropriate patients.	Review: Partially Successful  Approval to enter data has been received by Dartmouth. Additional clinics will be added in Year 4 pending creation of a PSO or exemption letters from each site's CPHS.
5	Submit for publication white paper on utilization of genetics services.	5	Publication submitted	Review: Unsuccessful  Further work on this area is dependent on establishment of a PSO or exemption letters from each site's CPHS and collection of data.
6	QI data submitted, analyzed and reported from all four current clinical genetics sites.	3	Data base in place	Review: Partially successful.  Data in place for one clinic, structure in place for all clinics. However, data entry for additional clinics is dependant on PSO creation or exemption letters from each site's CPHS.
7	Convene one "Breakthrough Learning Series" in quality improvement for NE Genetics Centers	3	Learning Collaborative is launched with diverse and sustained participation.	Review: Partially Successful  Initial groundwork created to establish a process for the Learning Collaborative, including draft charter, measure review, and schedule for Project Year 4.

**TRANSITION**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Assess willingness and ability of providers to provide adult care	3	Patients have contact with a medical home.	Review: Partially successful  As a key component of the Personal Transition Health Plan ( <i>Innovative Project</i> ), several clinicians were interviewed about their willingness/ability to provide adult care. While interest was generated in this area, changes to care practices will still need to be worked on in future

				years.
2	Hold conference on transition to adulthood	5	Conference takes place	Review: Unsuccessful  Activity postponed until Project Year 4.
3	Integrate transition activities to adulthood into regional outreach programs	3	Materials integrated into outreach efforts	Review: Successful  Presentations provided to Urea Cycle Disorder conference and national PKU association on importance of transition, distributed resources at each. Published findings in peer reviewed journals, with a focus on focus on maternal PKU, psychosocial issues and outcomes, screening for cognitive and social emotional problems. Collaborated with the national PKU association on adapting the transition toolkit to their efforts.
4	Create materials for women with galactosemia about premature ovarian insufficiency	1	Materials are published	Review: Successful  3 booklets created " <u><a href="#">Moving Forward: Your Guide to Galactosemia and Primary Ovarian Insufficiency (POI)</a></u> ", <u><a href="#">Understanding Galactosemia</a></u> and <u><a href="#">Understanding Galactosemia: Resources for Educators</a></u> . All are available at <a href="http://www.newenglandconsortium.org">www.newenglandconsortium.org</a> and linked to <a href="http://www.negenetics.org">www.negenetics.org</a> .
5	Create a range of fact sheets on various conditions for use by parents.	3	8 Fact sheets produced.	Review: Partially Successful  Five fact sheets developed and posted on website (Galactosemia, PKU, Biotinidase Deficiency, Homocystinuria, Ornithine Transcarbamylase (OTC) Deficiency). An additional 8 fact sheets are expected in Project Year 4.
6	Hold conference for adults with galactosemia	1	Conference takes place	Review: Successful  Conference and Research Opportunity for Adults with Galactosemia (Boston, August 20-23, 2009): 34 adults with

				galactosemia attended, 33 of whom were eligible to participate in the research activities that occurred in tandem with the conference.
7	Complete comprehensive review of transition practices	3	Review completed.	Review: Successful  Transition projects in the New England region identified and will be posted to the NEGC website. This list will be updated annually.
8	Create transition best practice protocol and customize for metabolic patients	3	Protocol defined and customized.	Review: Successful  Personal Transition Health Plan Project: reviewed literature on transition practices in the country, and created four health planning instruments. Available at: <a href="http://newenglandconsortium.org/for-families/transition-toolkit/">http://newenglandconsortium.org/for-families/transition-toolkit/</a> .  Supported creation of the PKU Challenge website; facilitates communication and sharing among individuals with PKU.  Counselor in Training model in use with other teen programming. Used in Teen Challenge: young adults with genetics
9.	Leadership training for teens with genetic disorders. Program at Teen Challenge Weekend	1	Leadership training takes place	Review: Successful  The Teen Challenge weekend was held in August 2009. Eighteen youths participated in a variety of activities designed to challenge them to try something new, build self-confidence, develop relationships with other adolescents with metabolic conditions, and develop skills to manage their health.
10.	Pilot transition practice(s) protocol in one metabolic clinic	4	Pilot project initiated and data analyzed.	Review: Partially Successful  New transition protocol established in Children's Hospital Boston. Due to late finalization of the data forms (as a

				result of hospital review processes), insufficient data were collected to complete the pilot study by Spring. While forms and resources are in place, a full pilot study and patient recruitment will be one of the objectives for Year 4 of the grant.
<b>MEDICAL HOME</b>				
<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Design suitable care planning tool for the care coordination pilot	3	Completion of a prototype care planning tool.	Review: Successful  Form created. The form requires the parent and doctor to explicitly note together what has been done and what concerns and requests should be related to the other doctor. Five focus groups were held with parents and physicians to improve the quality and utility of the tool.
2	Begin field test of the care coordination project in two specialty clinic catchment areas	4	At least two specialty clinics have implemented a care planning tool with at least 10 patients each.	Review: Partially Successful  The care planning tool was tested for feasibility in diverse pediatric practices both as formal research and as an informal quality improvement activity. The formal research aspect was based in two pediatric primary care practices and three pediatric specialties at UMass, and the informal pilot was based in the metabolic clinic at Children's Hospital Boston under the leadership of Dr. Susan Waisbren.  Due to delayed implementation of the tool (Spring 2010) as well as an insufficient number of patients meeting the criteria for the pilot studies, a full review of the tool could not be completed in Year Three. Efforts are underway to continue implementation of the tool in Year Four.

3	Convene at least 3 meetings of the MHWG during Year 3.	1	Two conference calls and one face to face meeting happen during Year 3.	Review: Successful Group met three times in Project Year Three.
4	Unforeseen Outcome: The work on the care planning tool influenced the improvement of the Transition Plan tool, an integral part of the Transition Plan Toolkit created by Dr. Waisbren. As a result, the Transition Plan tool now seeks <i>explicit written agreement about who does what next</i> .			

#### DISSEMINATION, EDUCATION AND MARKETING

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Continuously improve educational products and activities for providers and consumers	3	Collaborate with partners to improve access to high quality resources for providers and consumers.	Review: Successful Participating in NBS Clearinghouse project with the Genetic Alliance.
2	Assess the needs of special educators in the public sector	2	Deploy the survey to special educators	Review: Successful Due to constraints in the quality of information that would be received, the DEM group dropped pursuit of a survey and focused efforts on a more detailed focus group review with 3 groups in the New England area. Groups were completed and a report was written providing recommendations to the DEM group for further improvement of the educator tool. A poster outlining the project and focus group results was accepted for the NCHPEG meeting in Sept, 2010.
3	Develop educational resources for special educators	3	Develop continuing education modules for special educators on genetics	Review: Partially Successful The DEM group continues to use a range of resources (including the focus group results above) to improve the quality of the special educator tool.
4	The DEM group will continuously review and	3	Promote increased utilization of educational tools developed in the past years by improving their	Review: Partially Successful The DEM group has taken on role of reviewing ed materials to determine

	improve materials. They will focus on making them available via the web, and will discuss expanding the web-based chart to a broader audience (i.e. pediatricians).		electronic accessibility	applicability for disseminating to region. Recent example is a review of March of Dimes Training Modules for Genetic Issues for Perinatal Nurses.
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**DEMONSTRATE EFFECTIVE COLLABORATIONS**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
6.1	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.
6.2	Catalog WG and AC member participation in national groups	3	Members participate in key groups across the sphere of agencies providing or impacting services for individuals with genetic conditions.	Review: Successful  Participation on national groups continues to be tracked.
6.3	Link with affiliated programs (LEND and AUCD)	4	MOAs developed with participating programs identifying methods of collaboration.	Review: Unsuccessful  No major new endeavors have been established with LEND or AUCD in Year 3. In project Year 4, an innovative project has been funded with the LEND program.
			NIRS documentation kept up to date.	Review: Successful  NIRS data set is updated on an ongoing basis.
6.4	Represent genetics issues	3	Additional health care fields are educated about	Review: Successful

	to wider healthcare system		the needs of individuals living with genetic conditions.	<p>Provided letters of support to two grant applications by the Genetic Alliance, currently collaborating on one of these projects. Presentations made by staff at regional, national, and international conferences.</p> <p>Participated as featured resource in <i>Meet Your Neighbor</i> webinar series through Genetic Alliance, May 09</p>
			Public Health Genetics and Genomics is integrated into other academic course work	<p>Review: Successful</p> <p>Presentation, “Genetics and Public Health” was made by Amy Schwartz in Oct 09 as part of the Health Dialogue at UNH Manchester; also made to SNH Med School Grand Rounds in June 09. Ms. Schwartz also presented her genetics certification capstone project to Sarah Lawrence College in June 09.</p>

**INNOVATIVE PROJECTS PROGRAM**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Monitor innovative grant awardees	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.	<p>Review: Successful</p> <p>Monitoring and updating of the grant process has been continually implemented.</p>
2	Work with grantees to develop poster presentations for grant cycle 2	1	Poster presentations developed that represent and convey the spirit of the innovative projects program.	<p>Review: Successful</p> <p>Poster presentations were created and shared at the NEGC annual meeting.</p>
3	Release and award grantees for grant cycle 3	1	Appropriate grantees selected and awarded.	<p>Review: Successful</p> <p>4 Innovative grants were awarded</p>
4	Confirm award amount and	1	RFP issued.	Review: Successful

	issue RFP for grant cycle 4			Grant cycle 4 process was implemented.
<b>ETHICAL, LEGAL, AND SOCIAL ISSUES</b>				
<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Address ELSI issues within workgroups as well as through special projects	3	NEGC appropriately integrates ELSI issues within its work.	Review: Successful  Examples: Ethical (Transition and Medical Home work on changes in systems to enhance care via participation of families), Legal (analysis of cross data sharing, QI work on registry development), Social (Fanos work on parent perspectives, community grants, special educators tool, innovative project on diverse populations), Policy (LTFU data sharing agreements in Maine and Mass). Mass working on collaborative application.
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 as well as annually during the project's annual meeting.
<b>LABORATORY QUALITY ASSURANCE</b>				
<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Continue collaborations, creation of indices and assessment of variability of testing across labs.	3	Assessment report completed documenting differences (and their drivers) across lab settings.	Review: Successful  We discovered this year that lab-to-lab differences in mean marker levels are sufficiently great to require lab-specific cutoffs for most markers. To develop these we collected raw concentrations of relevant markers from 50,000 from our partner Region 1 lab of CT and 10,000 from WI. With these data we have calculated CT-specific proposed cut-offs for indices related to VLCAD, LCHAD, and CPT2. More data are required next year to extend this

				aspect to WI and NY.
2	Conduct data analysis using additional MSMS markers, expand to CAH screens.	3	<p>A table of additional (non-C3-related) cases identified in each participating region, showing the numbers of cases that fall into each of the three categories according to the pre-determined NENSP indices</p> <p>Development of detailed tables analogous to those created using New England data, for data from the collaborating laboratories, using the pre-determined NENSP indices.</p>	<p>Review: Successful</p> <p>Collected all marker concentrations on specimens from CT, WI, and NY, on cases and false positives related to IVA, 2MBG, VLCAD, LCHAD, and CPT2 (C5, C14, C14:1, C14:2, C16OH, C18:1OH, C16, C18:1). Analyzed index data vs. known follow-up from each State, and began applying and utilizing State-specific index cutoffs where possible (CT only, see obj. #1).</p>
			Table analogous to above, but for CAH disorders and out-of-range initial screens, derived from NENSP data alone.	<p>Review: Successful</p> <p>MSMS markers were analyzed for MA babies with CAH out-of-ranges. This (CAH)aspect of the project was determined to be not promising, and will not be further pursued.</p>
3	Convene collaborative conference May 2010	2	Conference Held	<p>Review: Successful</p> <p>Formal in person meeting not held due to scheduling challenges. However, group met and jointly worked on the project via other methods, including phone, email, and webex.</p>
<b>LONG TERM FOLLOW-UP</b>				
<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Complete Regional Charter Agreement	2	Draft of Charter completed	<p>Review: Successful</p> <p>Based on challenges in adopting a region wide agreement, a legal review group determined the regional charter was no longer appropriate. the group moved towards establishing state level agreements. Some states have made</p>

				significant 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).
2	Test of feasibility of data collection approach using NENSP LTF variables & Test of feasibility / census.	3	Limitations in data collection approach identified and solutions developed.	Review: Successful  Preliminary data collection was started as a feasibility test and consisted of a census of current patients (95% of cases were accounted for), hospitalizations, TCD practices and occurrence of strokes.
3	Define the quality assurance minimum dataset	1	Dataset defined.	Review: Successful  Minimum data set defined. Updated based on ongoing review of information.
4	Refine dataset variables per condition-specific needs	3	Variables reviewed at least annually and have support from Workgroups.	Review: Successful  Several changes were suggested and will be implemented going forward including reason for hospitalizations and accounting for routine TCD practices that vary by center (primarily age at first TCD).
5	Participate in Interregional and NCC activities	3	Participation yields – import or export of products.	Review: Successful  Presented LTFU activities in New England to the NCC/RC PI Annual Meeting in November 2009. Continued participation on NCC Provider Network and NSTRN clinical workgroup.
6	Convene Regional Policy and HSR conference	1	Conference yields at least one interstate commitment to data sharing policy.	Review: Successful  A full meeting was held in January 2010. Data sharing policy established with Maine.
7	New: Manuscripts developed documenting findings.	3	Manuscripts published.	Review: Successful  NENSP submitted an invited manuscript on LTFU efforts in New England that will be published in a Genetics and Medicine supplement. The manuscript (submitted in July 2009) focused on description of our strategies for sustainable LTFU, with

				<p>key notations on what we've learned after ten years of "expanded newborn screening." - They have accepted in Year 3, but not published.</p> <p>Additionally, a manuscript was drafted describing the psychosocial workgroup, and qualitative assessment of newborn screening communications.</p>
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**QUANTITATIVE AND QUALITATIVE EVALUATIONS**

<i>No.</i>	<i>Objective</i>	<i>Status</i>	<i>Yr. 3 Definition of Success</i>	<i>Yr 3 Results</i>
1	Gather data on program activities and outcomes and provide ongoing feedback to project staff and funder on project progress.	3	Management staff report evaluation support has been an effective aid in decision making and program improvement.	<p>Review: Successful</p> <p>Two of the work group chairs have self reported that the monthly focus on measures throughout the year has helped keep them on track and complemented their ability to be innovative.</p> <p>Management Staff Review: Evaluation process and results have been instrumental to the effective administration of the NEGC. Active participation by Dr. Antal at the administrative and other key meetings is key to our information for decision making.</p>
2	Conduct annual stakeholder survey	5	A majority of stakeholders participate in the survey process and provide recommendations for the project's improvement	<p>Review: Unsuccessful</p> <p>Due to creation of the NEGC website and the common interest in using the stakeholder survey as a means for gathering feedback on the website, the survey was postponed until Fall 2010.</p>
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.	<p>Review: Successful</p> <p>Yr 2 Report and Yr 3 Mid-Yr report completed and reviewed by staff.</p>

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 provide information for all national level reporting and discussions.
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## OBJECTIVES FOR YEAR FOUR

The following tables provide a list of objectives to be completed by each of the relevant workgroups and administrative teams for Year Four 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.

<b>Establish and Maintain the NEGC</b>			
<i>No.</i>	<i>Objective</i>	<i>Yr. 4 Definition of Success</i>	<i>Measurement of Success</i>
1	Continue implementation of core administrative supports to the NEGC	NEGC meets yearly objectives.	NEGC 2010/2011 Workplan.
2	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.	Satisfaction scores as measured by the annual Stakeholder Survey. Advisory Council chair participates on the Collaborative Council
3	Develop and implement a communications and outreach plan for the NEGC	Stakeholders report satisfaction with being able to voice their opinions and feel that they've been heard.	Stakeholder Survey.
		New groups and individuals are represented on the NEGC stakeholder list.	Stakeholder Survey Contact List
4	Maintain, update and enhance NEGC website	The NEGC stays current with state, regional, and national level developments.	Evaluator review
		Stakeholders have information necessary to keep informed of all project developments.	Stakeholder Survey

		Website is utilized by growing numbers of individual users.	Web logs
5	Implement Special Projects	Special Projects achieve stated goals within agreed upon timelines.	NEGC 2010/2011 Workplan.
<b>Quality Improvement</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 4 Definition of Success</b>	<b>Measurement of Success</b>
1	Registry will be implemented for all patients with developmental delays at all 5 sites.	All sites entering complete, quality data on all patients meeting criteria.	Number of patients entered by site.
			Quality of data audit by NEGC staff for every site will be designed and summary report to all members at end of 2011.
2	Create a PSO to host data collected from clinic sites / and/or obtain exemption letters for each site through CPHS	ARHQ website lists all approved PSOs / sites participate in registry under exemptions	Meeting Minutes
3	Implement QI report structure	Report format in Registry	Sample data by site will be generated by Registry for every site by year end.
4	QI data submitted, analyzed and reported from all five current clinical genetics sites.	Registry in place and utilized by all 5 clinical sites	Meeting Minutes.
5	Establish the Metabolic Quality Improvement Learning Collaborative	10 metabolic centers will send teams of 2-3 members each to QILC (3 meetings during the year).	Meeting Minutes.
		Support webinars between learning sessions will support teams	Meeting Minutes.
6	Establish quality improvement clinical process and outcomes for patients with metabolic disorders	A common set of data will be agreed upon.	An agreed upon data set forth.
		Condition-specific measures for at least 7 metabolic disorders or problems will be set forth.	Agreed-upon additional specific measures set forth for the seven selected metabolic conditions

			Document describing the internal quality assessment of data set will be written by the planning group.
7	Metabolic quality improvement registry will be established (customization of Genetics QI registry).	Registry exists and contains all the data elements defined by the QILC.	Registry in place and deployed at all sites
8	Metabolic centers will be members of the PSO / and/or will obtain CPHS exemption letters and have HIPAA BAAs in place.	PSO is in place / letters are obtained  Membership contracts are in place for all centers	PSO established / or exemptions established  Contracts established.
<b>Transition</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 4 Definition of Success</b>	<b>Measurement of Success</b>
1	Develop assessment tool for measuring successful transition to medical home	Written list of criteria identified.	Consensus from Regional and National Work Group
2	Continue to publish, present, and disseminate transition related agenda	Agenda promoted via published articles and presentations	Publications, presentations, and disseminated materials
3	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. 8 fact sheets will be produced in Year 4.	Distribution of Fact Sheets through the internet and clinics.  Web log.
4	Hold conference for adults with metabolic disorders	Conference held.	Summary report created.
5	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.
6	Assessment of best practice protocol by metabolic physicians and dieticians and other professional staff	Reviews received by at least 3 professional staff (dietician, nurse, fellow).	Reviewed comments.
7	Continue to Pilot transition practice at Children’s Hospital	10 patients participate at Children’s Hospital.	Written transition plans created for 10 patients

8	Leadership training for teens with genetic disorders. Program at Teen Challenge Weekend	Leadership training takes place.	Summary on training
9	Participate in effort to improve quality in metabolic clinics via learning collaborative methodology	Plan developed, ratified and implemented by QILC planning group and expert panel	Meeting reports
10	Continue to represent transition activities on LTFU as needed.	Improved access to assessment for all adults with genetic conditions in New England	Consensus on assessment method document at international meeting and piloted with 10 adults.
11	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

**Medical Home**

<i>No.</i>	<i>Objective</i>	<i>Yr. 4 Definition of Success</i>	<i>Measurement of Success</i>
1	Begin field test of the care coordination project in two specialty clinic catchment areas	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).	Evaluator review
		Process data are collected at the Children's Hospital site including number of plans implemented, number of visits documented.	
2	Convene at least 3 meetings of the MHWG during Year 4	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 December 2010 is a joint meeting of the two work groups.	Meeting agenda and attendees document the meeting.

**Dissemination, Education, and Marketing**

<i>No.</i>	<i>Objective</i>	<i>Yr. 4 Definition of Success</i>	<i>Measurement of Success</i>
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1	Continuously improve educational products and activities for providers and consumers	Current model finalized and dissemination plan created based on recommendations	Meeting Minutes
		Expansion of modules utilized in educational tool for special educators.	Meeting Minutes
		Revise tool for pediatricians and parents.	Meeting Minutes
2	Create web portal based on tool "Children with Genetic/Metabolic Conditions in the Educational Setting"	Tool posted on website	Evaluator Review
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

**Effective Collaborations**

<i>No.</i>	<i>Objective</i>	<i>Yr. 4 Definition of Success</i>	<i>Measurement of Success</i>
1	NEGC continues to participate in national work groups	The NEGC is actively represented on a national level by staff and NEGC constituents and contributes to the improvement and coordination of genetic services.	Meeting minutes and dissemination of information.
2	Link with affiliated programs (LEND and AUCD)	MOAs developed with participating programs identifying methods of collaboration	Meeting minutes, signed MOAs with other states
3	Represent genetics issues to wider healthcare system	Additional health care fields are educated about the needs of individuals living with genetic conditions.	Stakeholder participation on the Advisory Council, Members of the NEGC mailing list, # of publications in medical journals covering issues facing genetic services, cross-collaborative grants submitted with primary care providers.
		Public Health Genetics and Genomics is integrated into other academic course work	Evaluator review of courses actively linked to NEGC staff / resources.

**Innovative Projects**

<i>No.</i>	<i>Objective</i>	<i>Yr. 4 Definition of Success</i>	<i>Measurement of Success</i>
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1	Monitor innovative grant awardees including new micro grants to spur consumer involvement.	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	Meeting minutes.
2	Release, award and monitor grantees for 2010-11	Grant Cycle completed.	Report out during advisory, collaborative council and advisory council meetings. Release press coverage of field projects to local, regional and national audience
3	Work with grantees to develop poster presentations (regular grants) / brief summaries (micro grants)	Poster presentations / brief summaries developed that represent and convey the spirit of the innovative projects program.	Poster presentation, review by management staff and Advisory Council.
4	Confirm award amount and issue RFP for grant cycle 5	RFP issued.	Meeting minutes.

**Ethical, Legal, and Social Issues**

<i>No.</i>	<i>Objective</i>	<i>Yr. 4 Definition of Success</i>	<i>Measurement of Success</i>
1	Address ELSI issues within workgroups as well as through special projects	NEGC appropriately integrates ELSI issues within its work and actively pursues projects that improves the field of genetics in this area.	NEGC Workplan
2	Discuss ELSI issues within the RCC network.	Issues raised and discussed, NEGC lessons learned shared with the network	Quarterly email updates, NEGC Annual Meeting minutes. Review of NEGC by Advisory Council - Have we appropriately addressed ELSI issues in our work? What else can we be doing?

**Long Term Follow Up**

<i>No.</i>	<i>Objective</i>	<i>Yr. 4 Definition of Success</i>	<i>Measurement of Success</i>
1	Continued representation of LTFU workgroup in regional and national forums	Full participation in meetings	Meeting Minutes
2	Continue to Facilitate Stepwise Implementation of Activities Leading to Full Regional	Continued education of state teams (NBS Advisory committees) about Massachusetts	Meeting Minutes

	Participation in Long Term Follow Up	and Maine experience with implementation.	
		Facilitating Workgroups and reports back to state teams from work groups	Meeting Minutes
		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.	Meeting Minutes
3	Continued Data Collection and Expansion of Data Collection Activities	Subcontracts established with Maine and Rhode Island.	Meeting Minutes
		State specific data modules created and integrated.	Meeting Minutes
4	Data Analyses and Publication of Analyses	Data analysis prepared for QI at the clinic and program levels	Meeting Minutes
		Manuscripts developed documenting findings.	Publications
5	Enhancing Development of Best Practices	Hgb conference to facilitate development of best practices.	Conference evaluation
		Development of best practices by clinical workgroups	Meeting Minutes
<b>Quality Assurance</b>			
<b>No.</b>	<b>Objective</b>	<b>Yr. 4 Definition of Success</b>	<b>Measurement of Success</b>
1	Continued representation of quality control workgroup in regional and national forums	Full participation in meetings	Meeting Minutes
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.	Analysis completed.	Development of detailed tables (analogous to those created using New England data), for data from the collaborating laboratories, showing the deviation from the mean (in terms of Z-scores of log-transformed data) for each marker (even if not measured by NENSP) of each OOR specimen.

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	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.	Detailed data tables produced.
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.	Meetings held. Target web-ex meetings in Sept., Nov., Jan, and face-to-face meeting in March, possible web-ex in May	Full and regular participation by key partners.
5	Present preliminary findings to Lab Subgroup Committee of SACHDNC	Presentation	Presentation given

**Evaluation**

<i>No.</i>	<i>Objective</i>	<i>Yr. 4 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 semi-annual and 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.

## 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 (1 of 10): no substantive activities have been undertaken
- In process (2 of 10): activities are under way to address the challenge but have not yet led to substantive changes in practice
- Improving (7 of 10): activities have led to substantial improvements in the challenge area
- Addressed (1 of 10): the basic nature of the challenge has been successfully addressed by project staff

### Update on Challenges Identified to Date

#### **Status: Not Addressed**

##### *Access to Genetic Specialists*

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.

##### Recommendation:

The NEGC Advisory and Collaborative Council should determine whether this area needs to be a priority in the near term (1-2 years). If so, action steps and leaders should be defined to make improvements in this area.

#### **Status: In Process**

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

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 and Three, substantive efforts were made to educate both regional and national level stakeholders about the need for a medical home. In addition, the care coordination and transition pilot projects as well as efforts around the learning collaborative and long term follow up have great potential to have a direct impact in this area; setting the stage for documenting the importance and utility of medical home approaches in the region.

##### Recommendation:

- Continue supporting the Medical Home workgroup's efforts to implement the care planning and transition tools in a variety of settings. As this is done, a basic protocol should be put in place to gather a minimal level of evaluation information 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.

### *Many Stakeholders, Limited Funds*

During Year Three, partners of the collaborative continued to grapple 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 is not always achievable.

#### Recommendation:

Given that continued funding has a direct impact on the long term viability of the NEGC as well as its partners, and the fact that the field is strengthened by the continued presence and activity of multiple partners, it is in the interests of all involved to identify a broad range of ways to support each group's efforts. It may be helpful for an annual review to be conducted of each entity's grant seeking processes and to identify some of the major challenges to successful grant applications (e.g. were unsuccessful grants due to an inappropriate lead, too large of a scope of work, insufficient documentation, lack of consumer involvement, lack of access to more appropriate funding institutions?). Based on this review, and, to the extent that it fits within the NEGC's role of coordinator and supporter of change in the region, it may also be helpful for the NEGC to actively seek out opportunities where it could better support lead applications from partner agencies and lend some of its resources towards the successful application of these efforts.

### **Status: Improving**

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

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. The Transitions Workgroup, at both a national and regional level, have taken a number of steps towards ensuring access to continuous care among youth (creation of the Transition toolkit, leading national and regional dialog on Transition, and initiation of the pilot transition project).

#### 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*

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.

#### Recommendation:

Given the ongoing challenge of this area, it is recommended that continued efforts focus on improving and broadening communication efforts by the NEGC at all levels (not only from mgmt staff to other groups, but also within and across workgroups). Several areas of improvement have already been identified by project staff and are being worked on, including: outreach to specific groups, renovation of the project website, and initiation of newly formatted updates to the group.

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

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.

#### Recommendation:

Use the opportunity of the annual meeting, with its range of participants, to both review current partnerships and identify needed new ones. Break-out sessions offer a unique opportunity to work with a targeted group (such as the Birth Defects Consortium, genetic counselors, or consumers) to identify avenues of collaboration as well as specific steps to be taken in the year ahead.

#### *Geographic Barriers to Meeting*

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 Year Three, 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).

#### Recommendation:

Workgroup leaders continue to make good use of conference calls and technology to support their meetings, such as GoToMeeting.com and utilization of video conferencing technology through Webex. These resources continue to provide an effective means for members to conduct their work.

### *Quality Data Systems*

The QI, Transitions, 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 legislation 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.

#### Recommendation:

Continue supports to each of the workgroups. Identify steps that each of these initiatives can use to collaborate together; this is particularly important given the limited number of individuals in the field, as well as the limited availability of time each of them has to work on initiatives outside of their immediate scope of work.

### *Patient Access to Genetics Information.*

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 and Three of the project, substantive additions were made to the NEGC website to help fill this gap. Additionally, the DEM workgroup has agreed to begin reviewing resources for appropriateness and potential inclusion on the website.

#### Recommendation:

Continue to provide support to the DEM group to review potential resources for the NEGC website and lend framing and dissemination support to their efforts for the special educator tool. Additionally, there should be, at minimum, an annual review process (involving consumers and other professionals) to look at the website and identify any critical gaps of information that would be of use to NEGC stakeholders. Lastly, given the use of the 211 information system in the New England states, it would be helpful for the NEGC to facilitate links between various providers of genetic health and support services and the main host for each state's 211 system.

### *Tracking Progress of Work Groups*

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.

#### Recommendation:

As noted last year, there continues to be variety in the level of information provided in workgroup minutes. Additionally, there are often substantial delays in the approval and posting of meeting minutes from different workgroups. At a minimum, it is recommended that all workgroups should include the following information in tracking their meeting events: meeting date, participants, major discussion points, barriers encountered and solutions identified (if any), next steps and person(s) responsible. Whenever possible, 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 Year Three. 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.

## **New Challenges and Recommendations for Year Four**

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

At several points during the last few years, individuals 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. As the NEGC continues to grow and form a range of stronger partnerships with groups providing services across the region, there is the potential for the NEGC to have a natural role in this area.

Recommendation: If not already available, it would be helpful to conduct an analysis of current and proposed future health care changes at the national and state levels respective of the ability for individuals with genetic conditions to access and sustain appropriate health care. Following a review of this analysis by the advisory council, collaborative council, and management staff, a decision can be made as to what role, if any, the NEGC should take in addressing this area.

## **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

### COLLABORATIVE COUNCIL WORKING GROUP/LEADERS

#### DISSEMINATION, EDUCATION, MARKETING

Leah Burke, MD

#### ACCESS TO SERVICES

##### LINKAGES TO MEDICAL HOME

Carl Cooley, MD

##### 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>
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>
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.

## APPENDIX C: NEGC PRESENTATIONS LIST

### \* New in Year Three

#### Sharing Work on Project Activities

\* *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

\* *CF: recommendations to increase Newborn Screening efficiency.*

7th International Congress, Latin American Society of Inborn Errors of Metabolism and Neonatal Screening,  
December 7, 2009  
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

## **Poster Presentations**

*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 Three

- \* Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders Consortium Seminara J, Tuchman M, Krivitzky L, Krischer J, Lee HS, Lemons C, Baumgartner, M, Cederbaum S, Diaz GA, Feigenbaum A, Gallagher RC, Harding CO, Kerr DS, Lanpher, B, Lee B, Lichter-Konecki U, McCandless SE, Merritt JL, Oster-Granite ML, Seashore MR, Stricker T, Summar M, Waisbren S, Yudkoff M, Batshaw ML. Mol Genet Metab. 2010; 100 Suppl 1:S97-105. Epub 2010 Feb; Review. PubMed PMID: 20188616; PubMed Central PMCID: PMC2858794.
- \* Projected costs, risks, and benefits of expanded newborn screening for MCADD Prosser LA, Kong CY, Rusinak D, Waisbren SL Pediatrics. 2010, Feb; 125(2):e286-94. PubMed PMID: 20123779.
- \* Impact of false-positive newborn metabolic screening results on early health care utilization Lipstein EA, Perrin JM, Waisbren SE, Prosser LA Genet Med. 2009 Oct; 11(10):716-21. PubMed PMID: 19661808; PubMed Central PMCID: PMC2773165.
- \* The psychology and neuropathology of phenylketonuria. White DA, Waisbren S, van Spronsen FJ. Mol Genet Metab. 2010;99 Suppl 1:S1-2.
- \* Final commentary: a new chapter. White DA, Waisbren S, van Spronsen FJ. Mol Genet Metab. 2010;99 Suppl 1:S106-107.
- \* Screening for cognitive and social-emotional problems in individuals with PKU: tools for use in the metabolic clinic. Waisbren S, White DA. Mol Genet Metab. 2010;99 Suppl 1:S96-99.
- \* Psychosocial issues and outcomes in maternal PKU. Koch R, Trefz F, Waisbren S. Mol Genet Metab. 2010;99 Suppl 1:S68-74.
- \* Psychiatric symptoms and disorders in phenylketonuria. Brumm VL, Bilder D, Waisbren SE. Mol Genet Metab. 2010;99 Suppl 1:S59-63.
- \* Improving genetic health care: a Northern New England pilot project addressing the genetic evaluation of the child with developmental delays or intellectual disability. Moeschler JB, Amato RS, Brewster T, et al. Am J Med Genet C Semin Med Genet. Aug 15 2009;151C(3):241-254.
- \* Access to genetic counseling for children with autism, Down syndrome, and intellectual disabilities. McGrath RJ, Laflamme DJ, Schwartz AP, Stransky M, Moeschler JB. Pediatrics. Dec 2009;124 Suppl 4:S443-449.
- \* Medical home 2009: what it is, where we were, and where we are today. Homer CJ, Cooley WC, Strickland B. Pediatr Ann. Sep 2009;38(9):483-490.

\* Improved outcomes associated with medical home implementation in pediatric primary care. Cooley WC, McAllister JW, Sherrieb K, Kuhlthau K. *Pediatrics*. Jul 2009;124(1):358-364.

20p12.3 microdeletion predisposes to Wolff-Parkinson-White syndrome with variable neurocognitive deficits Lalani SR, Thakuria JV, Cox GF, Wang X, Bi W, Bray MS, Shaw C, Cheung SW, Chinault C, Boggs BA, Ou Z, Brundage EK, Lupski JR, Gentile J, Waisbren S, Pursley A, Ma L, Khajavi M, Zapata G, Friedman R, Kim JJ, Towbin JA, Stankiewicz P, Schnittger S, Hansmann I, Ai T, Sood S, Wehrens XH, Martin JF, Belmont JW, Potocki L. *J Med Genet*. 2009 Mar; 46(3):168-75. Epub 2008 Sep 23. PubMed PMID: 18812404; PubMed Central PMCID: PMC2680125.

Parental tolerance of false-positive newborn screening results. Susan Waisbren. *Archive of Pediatric Adolescent Medicine*, 2008, 162: 870-6

Short-chain acyl-CoA dehydrogenase (SCAD) deficiency: an examination of the medical and the neurodevelopmental. Susan Waisbren. *Molecular Genetic Metabolism*, 2008, 95: 39-45

Stability of blood phenylalanine levels and IQ in children with PKU. Susan Waisbren. *Molecular Genetic Metabolism*, 2008, 95:17-20

Oropharyngeal flora in healthy infants: observations and implications for cystic fibrosis care. Carlson D, McKeen E, Mitchell M, Torres B, Parad R, Comeau AM, O'Sullivan BP. *Pediatric Pulmonology* 2009 May;44(5):497-502.

Population-based research within a public health system: two models for common rule compliance in the Massachusetts Newborn Screening Program In M. Bailey & T. Murray (Eds.), In: *Ethics and newborn genetics screening: New technologies, new challenges*. Comeau, A., & Levin, D. Baltimore, MD: Johns Hopkins Press.2009.

Spectrum of Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency detected by newborn screening Hsu H-W, Zytovicz TH, Comeau, AM, Strauss AW, Marsden D, Shih VE, Grady GF and Eaton RB. *Pediatrics*, 2008;121:e1108-e1114

Newborn Screening Showing Decreasing Incidence of Cystic Fibrosis. Hale JE, Parad RB, Comeau, AM. *New England Journal of Medicine*, 2008, 358:9:973-974 (Correspondence)

Medical genetics diagnostic evaluation of the child with global developmental delay or intellectual disability. John Moeschler. *Current Opinion in Neurology*, 2008, 21:117-122

Expanded newborn screening: information and resources for the family. Susan Waisbren. *American Family Physician*, 2008, 77: 987-9

## **APPENDIX E. SUMMARY OF WORKGROUP MILESTONES**

	<i>June 09</i>	<i>July 09</i>	<i>Aug 09</i>	<i>Sept 09</i>	<i>Oct 09</i>	<i>Nov 09</i>	<i>Dec 09</i>	<i>Jan 10</i>	<i>Feb 10</i>	<i>Mar 10</i>	<i>April 10</i>	<i>May 10</i>
Project Staff							Annual NEGC Meeting	Support for Learning Collaborative launched	Reapp. for federal funding.			Communications plan created
Innovative Projects	LOI due for 2009-2010 projects	Proposal apps. due	Review process completed & awardees notified	2009-2010 Project start date							LOI due for 2010-2011 apps	2009-2010 projects end
Advisory Committee							Annual Meeting held.					
Collab. Council												
Evaluation Team	Yr 2 survey complete			Yr 2 survey results released	Year 2 annual report released			Mid-year update report released. National measures submitted.				

	<i>June 09</i>	<i>July 09</i>	<i>Aug 09</i>	<i>Sept 09</i>	<i>Oct 09</i>	<i>Nov 09</i>	<i>Dec 09</i>	<i>Jan 10</i>	<i>Feb 10</i>	<i>Mar 10</i>	<i>April 10</i>	<i>May 10</i>
Quality Improve.								Planning meetings for the Learning Collaborative start				Agreement reached on supporting future LC work.
Medical Home							Joint meeting of the Medical Home and Transition groups.			Care planning tool approved for use in pilot study at UMASS and Children's Boston.	Dr. Carl Cooley presented to the NH Pediatric Society on implementing AAP Developmental Screening Guidelines in the Primary Care Medical Home.	
Transition			Teen Challenge – Boston, 9 youth				Joint meeting of the Medical Home and Transition groups.			Support provided for PKU Challenge website		Adult transition toolkit, 5 fact sheets posted Biotinidase Deficiency, Galactosemia, Homocystinuria, OTC Deficiency

	<i>June 09</i>	<i>July 09</i>	<i>Aug 09</i>	<i>Sept 09</i>	<i>Oct 09</i>	<i>Nov 09</i>	<i>Dec 09</i>	<i>Jan 10</i>	<i>Feb 10</i>	<i>Mar 10</i>	<i>April 10</i>	<i>May 10</i>
												PKU
Diss. Education & Marketing												Report on focus group re: special educator tool
Lab QA												
Long-Term Follow-up*					Data report created for Hgb group			Policy Conference held		Legislation passed in Maine for LTFU		

\* CF workgroup received ongoing tracking reports. Genetics in Medicine paper data distributed to MA Metabolic Workgroup and to LTFU workgroup for them to distribute to their clinics (occurred twice during the year).

