New England Genetics Collaborative

Annual Evaluation Report for Project Year Nine

Reflections on Project Activities 6/1/15- 5/31/16

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

September 2016

New England Genetics Collaborative

Annual Evaluation Report for Project Year Nine

Contents

EXECUTIVE SUMMARY

This annual report covers the activities of the New England Genetics Collaborative (NEGC) from June 1, 2015 to May 31, 2016. 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 during the period; an update on the status of core project components from Year Nine; and recommendations to the project by the project evaluator. 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) cooperative agreement (HRSA Grant # U22MC10980) officially began June 1, 2007.

During its ninth year of activity, core project staff members have focused on 1) creation, analysis and summary of results for a new research brief by the HAF group looking at family access to care for children born with genetic conditions; 2) improved data analysis and reporting out of summary data for members of the QI group; 3) expanding the content of the Genetics Education Materials for School Success(GEMSS) website along with a thorough review of recommended improvements and creation of a new learning module for higher education facilities; 4) full launch of the Genetic Metabolic Center for Education (GMCE); 5) continuing to build advocacy supports for young adults living with genetic conditions 6) creation of a new transition policy which was adopted by Children's Hospital Boston, 7) 4 Medical Home webinars on a range of timely topics; 8) supporting alignment and integration of genetic resources among regional LEND programs; 9) supporting multiple new training sessions, presentations and publications; and 10) continuing to expand the social media reach of the NEGC.

In reviewing the goals and objectives for Year Nine, 94% of 31 objectives have either been fully successful (77%) or have made satisfactory progress (16%) in accordance with the long term goals of the grant. Of the remaining objectives, 6% are dependent on previous activities being completed or were not scheduled for this year, 0% did not make satisfactory progress, and 0% of activities were abandoned and resources reallocated. Feedback from the Annual Meeting held in Spring of 2016 (N=12-15) indicated strengths in several critical areas: the NEGC is headed in the right direction (92%), participants understood how their own work fits with the NEGC (92%), participants had a good understanding of what the NEGC will accomplish in the following year (92%), and the NEGC has had tangible outcomes that have resulted in improvements in accessibility of high quality genetic services in the region (100%). Objectives for Year Ten have been shared and agreed to by project staff and chairs of the project's work groups. In preparing to successfully meet the Collaborative's objectives, a summary review and recommendations for improvement have been identified in the final section of this report.

COALITION ACTIVITIES

Organizational Overview

The NEGC is staffed by John Moeschler, MD and Monica McClain, Ph.D., who serve as Co-Directors, 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 2015 – 2016, the NEGC carried out substantial portions of its work through six Workgroups: Education and Outreach, Health Care Access & Financing, Medical Home, Quality Improvement, Transition, and NEGC/LEND. The chair of each Workgroup is a member of the Collaborative Council which facilitates coordination of Workgroup activities. The NEGC and Collaborative Council are guided by an Advisory Committee, which meets twice annually to help set direction for the collaborative and to provide feedback / raise issues throughout the year as needed. Please see Appendix A for the current organizational chart. There were no changes in the staffing of key positions during Project Year Nine.

Collaborative Activities

Project staff continued to seek out new opportunities for partnerships with both regional and national partners. During Year Nine, this included: the NEGC Annual meeting, 14 presentations, 3 publications, a learning module for GEMSS, a new fact sheet on improving the health and well-being of people with inherited conditions, as well as 17 training and technical assistance activities. Also this year, the NEGC continued to support the Genetics and Metabolic Center for Education and finalized its work on the CCHD project. The following outlines each of these accomplishments in more detail.

Annual Meeting, April 7-8, 2016

The annual meeting was well attended by 60 people. In addition to holding a remembrance for Dr. Sondra Solomon, plenary sessions included a focus on:

- Programmatic and Ethical Implications of Integration Next Generation Genomic Sequencing into State NBS Programs by Aaron Goldenberg, Ph.D.
    - (Slides with voice over available at https://vimeo.com/172143121)
- NewSTEPS: A View of Newborn Screening Nationally and Regionally with an Eye to the Future by Thalia Wood, MPH
    - (https://vimeo.com/171819109)
- Genetic Metabolic Center for Education, Mark Korson, MD
    - (https://vimeo.com/171819111)
- Discussion of proposed future models for the NCC/RC Network by Celia Kaye, MD
    - (https://vimeo.com/172143122).

As with previous years, open workgroup meetings were held that enabled cross group and new stakeholder participation in the activities of individual workgroups. These included:

- Education and Outreach: The group shared the new learning module for higher education institutions to aid in educating future teachers about the GEMSS website, reviewed a very successful partnership with the GEMSS action group to improve the site, and held a special collaborative session with the Medical Home group.
- Medical Home: Reviewed the upcoming webinar on Strengths Based Communication, potential collaborations with FQHCs and the National Center for Care Coordination and Technical Assistance, held a substantive discussion on payment recommendations, and discussed how to adapt GEMSS for health care providers.
- Health Care Access & Finance: Discussed evolving challenges to families (including multiple changes to health care policy and network adequacy, the potential for engaging with Medicaid advisory bodies, addressing challenges with access to durable medical equipment, and navigating appeals processes
- Transition: The group reviewed everyone's goals around needed quality of life changes for youth, results from the adult health survey, updates to the adult healthcare transition policy, as well as future ideas for discussion (including potential for additional health outreach, expanding the use of the transition policy, building social connections, parent support, improving access to services and care coordination / support, creating effective transition supports and addressing the whole person.
- Quality Improvement: The group discussed current levels of interest in gathering data on metabolic activities, reviewing the current data on children with DD/ID, and identified a better understanding of the impact of whole exome sequencing as a possible focus for the next year.

Among those providing feedback on the meeting evaluation forms (N=15), 100% agreed that they had an opportunity to share and contribute and that the NEGC has produced tangible outcomes that have resulted in improvements in the accessibility of high quality genetic services in the region. Eleven of 12 felt the NEGC was headed in the right direction and 12 of 13 understood what the NEGC would accomplish in the next year and how their work fit in. For those involved in the workgroup sessions, all stated that they found the sessions somewhat or very helpful, with most in each case indicating the experience was very helpful.

Presentations and Publications Supported by the NEGC

During Project Year Nine, NEGC coalition stakeholders provided 14 presentations across a diverse mix of presenters.

Poster Presentations:

- Genetics Education Materials for School Success (GEMSS) for Neuromuscular Disorders
    - Muscular Dystrophy Regional Conference, November 7, 2015; New Hampshire by Dillon A, Fox S.
- Genetics Education Materials for School Success (GEMSS).
    - American with Disabilities Act Equity and Access Fair, University of New Hampshire; November 18, 2015; Durham, NH by Smith K.
- Genetics Education Materials for School Success Learning Module
    - NEGC Annual Education & Outreach Work Group Meeting, Portsmouth, NH; April 7,2016 by Baril, E.

Invited oral presentations:

- Genetics Education Materials for School Success: School Can Be a Great Place for Everyone.
    - The Center for START Services Annual Conference, March 14, 2016 by Dillon, A. & Smith, K.
- Using state birth defects registries to evaluate outcomes of critical congenital heart disease newborn screening.
    - American Public Health Laboratories Newborn Screening and Genetic Testing Symposium, St. Louis, MO, February-March 2016 by McClain MR.
- "A World of Uncertainty": Impact on Parents of a Diagnosis of Critical Congenital Heart Disease (CCHD).
    - Grand Rounds at UCSF Benioff Children's Hospital, September 15, 2015; Oakland, CA by Fanos J, Landon C, Bose A, McClain M.
- GEMSS for Neuromuscular Disorders.
    - Muscular Dystrophy Regional Conference November 7, 2015; New Hampshire by Dillon A.
- Adult Health Survey.
    - Brigham & Women's Hospital Fellows' Program; September 2015; Boston, MA by Rajibi F.
- Transition Policy.
    - Post Clinic at Children's Hospital Boston, September 29, 2015; Boston, MA by Waisbren S.
- Transition Policy.
    - New England Consortium of Metabolic Programs; November 13, 2015; Boylston, MA by Waisbren S.
- Genetics and Metabolic Center for Education.
    - New England Regional Genetics Group; December 3, 2015; Portsmouth, NH by Korson M.
- Genetic Counseling via Telemedicine.
    - New England Regional Genetics Group; December 3, 2015; Portsmouth, NH by Smith R and Upton S.
- Genetics and Metabolic Center for Education
    - National Coordinating Center Annual Meeting; November 12, 2015; Washington, DC by Korson M.
- Genetics and Metabolic Center for Education.
    - New England Consortium of Metabolic Programs; November 13, 2015; Boylston, MA.Returning to Therapy & the ACMG Diagnosis & Management Guidelines by Korson M.

By the end of Project Year Nine, three additional publications were supported by NEGC funds. The most recent products include:

Peer Reviewed Publications

- McClain MR, McGrath R, Stransky ML, Benkendorf JL. National survey of providers treating patients with metabolic disorders identified by newborn screening demonstrates challenges faced by clinical care systems. Clinical Pediatrics, 54:759-64. 2015.
- Waisbren, S. E., He, J., & McCarter, R. (2015). Assessing Psychological Functioning in Metabolic Disorders: Validation of the Adaptive Behavior Assessment System, (ABAS-II), and the Behavior Rating Inventory of Executive Function (BRIEF) for Identification of Individuals at Risk. JIMD Reports, 21, 35.

- Waisbren SE, Rohr F, Anastasoaie V, Brown M, Harris D, Ozonoff A, Petrides S, Wessel A, Levy HL. Maternal Phenylketonuria: Long-term Outcomes in Offspring and Post-pregnancy Maternal Characteristics. JIMD Rep. 2015;21:23-33. doi:10.1007/8904_2014_365. Epub 2015 Feb 25.

In addition to the above areas, the project was responsible for the creation of a GEMSS higher education learning module, and a new fact sheet from the Health Care Access and Financing Group on improving the health and well-being of people with inherited conditions.

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

Trainings and Technical Assistance

During Year 9, staff funded by the NEGC carried out 17 training and technical assistance activities to families, state staff, and others. Of the 357 aided in this manner, most were health providers and other professionals (279), followed by state MCH staff (30), LEND trainees (38), families (8), and other (2)[1]. Areas of support provided included: a training to LEND trainees, creating a shared plan of care for kids, documenting a pediatric family history, incorporating genetics in the management of children in the primary care office, and lessons learned from the New England CCHD project.

Special Projects

During Year 9, the NEGC completed its work with the Critical Congenital Heart Disease (CCHD) screening project, and continued its support for the Genetic Metabolic Center for Education.

The goal of the ***New England CCHD Newborn S****creening Project* is to develop processes for CCHD screening that will set the stage for improved health outcomes for newborns with CCHD, and their families. As a result of its efforts, the project established a consistent method for screening and reporting of CCHD across New England. Over 42,000 children were screened since the project started, and 3 children diagnosed with CCHD. An electronic resource was developed for families on psychosocial supports and went live in May 2016 (www.necongenitalheartresources.org). 500 resource bags were distributed to 4 pediatric cardiology practices for distribution to families that receive a CCHD diagnosis.

Also during Year 9, NEGC continued its partnership with the **Genetic Metabolic Center for Education (GMCE)**. The purpose of this collaboration is to create a consulting and educational platform able to support the diagnosis and treatment of inherited metabolic disorders throughout New England. As a result of its activities, GMCE partners developed a HIPAA compliant telemedicine system for hosting conferences between specialists and other care providers, launched a pilot effort with three clinical sites from across the New England region, and completed 19 consultations with providers.

Collaborations with Regional and National Partners

[1] Note, summaries can include more than one count per person as some individuals will have attended multiple events

This section provides documentation on the affiliations held by NEGC management and collaborative council members with the intent of highlighting the connections members have to both regional and national initiatives which have parallel missions to the work of the NEGC.

Supporting the National Coordinating Center

- Evaluation: Peter Antal, Ph.D., Monica McClain, Ph.D.,
- Care Coordination: Greg Prazar, MD
- Health Care Access and Financing: Monica McClain, Ph.D., Peter Antal, Ph.D., Meg Comeau, MHA
- Regional Support Service Model / Advisory Committee: Monica McClain, Ph.D.; Susan Waisbren, Ph.D.
- Telegenetics: Rosemarie Smith, MD

Collaboration with Other Regional and National Groups

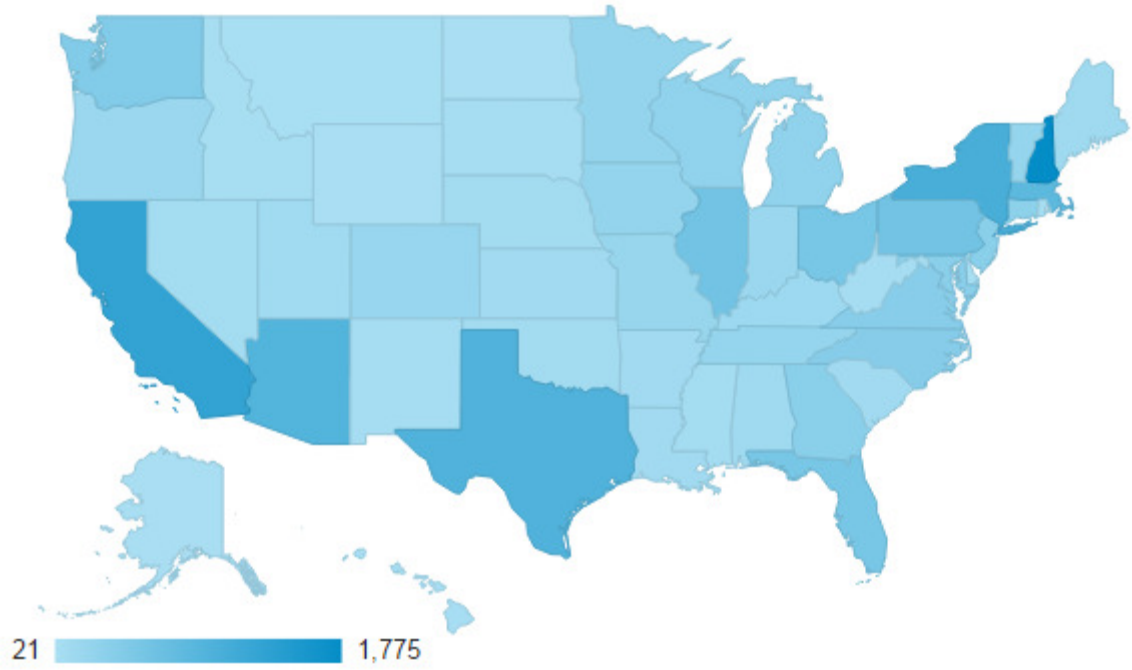
- Catalyst Center: Meg Comeau, MHA
- Jackson Laboratories for Genomic Medicine: Monica McClain, PhD
- Leadership Education in Neurodevelopmental and Related Disabilities (LEND): John Moeschler, MD; Monica McClain, PhD; Wendy Smith, MD; Leah Burke, MD; Peter Antal, Ph.D.
- New England Regional Genetics Group: Monica McClain, Ph.D., Karen Smith, Peter Antal, Ph.D., John Moeschler, MD.
- New England Consortium of Metabolic Programs: Susan Waisbren, Ph.D., Leah Burke, MD
- Next Step: Susan Waisbren, Ph.D.
- AAP - Genetics and Birth Defects: Leah Burke, MD
- PKU and Allied Disorders, Inc: Susan Waisbren, PhD
- National Urea Cycle Disorders Consortium: Susan Waisbren, PhD (Lead Psychologist/PI for New England center)
- BabySeq Project: Susan Waisbren, PhD (Lead psychologist for CHB and B&W sites)
- NIH – Genomic Sequencing and Newborn Screening Disorders research program: Susan Waisbren, PhD

A Focus on the GEMSS Online Resource

The GEMSS (Genetics Education Materials for School Success) site, initially launched in February 2012, has continued to experience rapid growth in content and high utilization (ranging from 1,137 unique users per month to 2,529 in the current year). During Year Nine, an estimated[2] 20,645 individuals visited the website (an increase of 6.5% over the previous year). These users had 25,843 sessions and 86,762 pageviews. Users represented all 50 states and 137 other countries / territories.

[2] Counts for FY2015 are estimated due to a loss of web traffic data during a 3 month period when the website was upgraded.

Figure 3: GEMSS Access by State



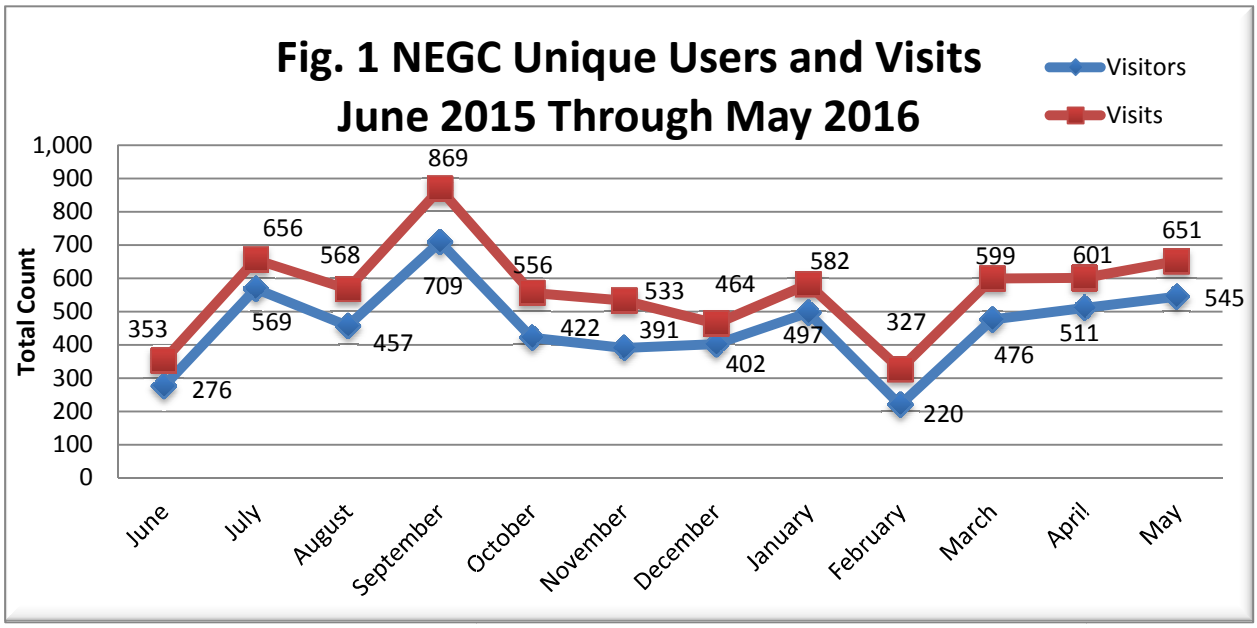
Project leaders have continued to seek out expertise and feedback from users, regional and national advocacy groups, as well as leaders in the field. By May 2016, the site added 3 new conditions (Russell Silver Syndrome, CHARGE, and Neuromuscular disorders) and content for Autism Spectrum Disorders was significantly enhanced bringing the total count of conditions covered to 36.

NEGC on the Web

In 2015-2016, the NEGC and GEMSS website continued to receive a number of new enhancements and updates, including the addition of multiple presentations and webinars on a range of genetics topics accessible to public audiences.

NEGC Visitors

During Year Nine, monthly visitors to the NEGC ranged from a low of 220 and 276 (March and April) to a high of 869 (September) - see Fig.1. Compared to the previous year, there were substantively more visitors to the NEGC (5,212 vs. 2,667) and more sessions (6,759 vs. 3,525). Over the course of these 12 months, the average time spent on the website changed throughout the year, ranging from a low of 0:53 (December) to a high of 3:10 (February) with total page views ranging from 818 (Dec) to 2,106 (Sept).



Sources of Referral to the NEGC Website

In looking across different referral sources to the website and the levels of activity generated (See Table 1), the most effective source (in terms of time spent on the site and a low bounce rate) was email referrals, with 136 sessions generated and an average time of 8:45 (minutes:seconds) per visit and a bounce rate of 35%, followed by referrals by partner organizations (time - 3:27, bounce rate 35.3%) and direct link (time - 3:59, bounce rate 57.5%). Outside of UNH, the top five organizational drivers to the NEGC website were the NCC (122), Maine Medical Partners (56), Clinica Curet (34), Genetics in Wisconsin (27), and the CDC (22). Of note, the GEMSS website generated 53 sessions to the NEGC and the IOD, 94. A total of 33 organizations (27 in Year 8, 35 in Year 7) were identified as referral sources to the NEGC. Of note, links driven by social media (primarily from Facebook in Year 9) accounted for 151 visits (38 in Year 8, 6 in Year 7), an average time of 1:13 seconds on the website and a bounce rate of 83%. 'Other' refers primarily to web market generated referrals or from other unaffiliated sites.

Table 1: Referring Sources to the NEGC Website[3]

Yr 9	Sessions	S/Mth	Pages/S	Avg Time	% New	Bounce Rate[4]
Direct Link	1,278	106.5	4.3	0:03:59	66.3%	57.5%
Partner Org	594	5.0	4.2	0:03:27	60.3%	43.6%
Email Referral	136	3.0	6.6	0:08:45	22.1%	35.3%
Search	1870	122.0	2.7	0:01:46	76.0%	58.8%
Social	151	5.1	1.5	0:01:13	78.1%	83.4%
Other	2,730	22.8	1.1	0:00:13	88.2%	95.2%

Yr 8	Sessions	Adj. Sessions	S/Mth	Pages/S	Avg Time	% New	Bounce Rate[5]
Direct Link	609	812	67.7	3.94	3:49	61.9%	56.8%
Partner Org	589	785	6.2	5.3	5:53	41.4%	36%
Email Referral	12	16	0.7	1.8	1:21	25%	33.3%
Search	1006	1341	87.5	3	2:12	77.4%	53.7%
Social	42	56	0.8	1.6	1:15	71.4%	69%
Other	476	635	7.9	1.1	0:15	95%	83.2%

In comparing results between Years 9 and 8, there were substantive increases in all categories except for referrals from partner organizations which held fairly stable. Of note for future reviews, web hits classified as coming from "Other" sources, increased by a factor of 6 from previous years indicating the continued growth and presence of web crawlers and other search engines which can be tagged as a hit on the website, making accurate assessments of website growth among intended audiences a challenge unless these sources are accounted for.

[3] Adjusted Sessions adds in an estimate based on monthly averages for the three months of data that was missing from Google Analytics reports.

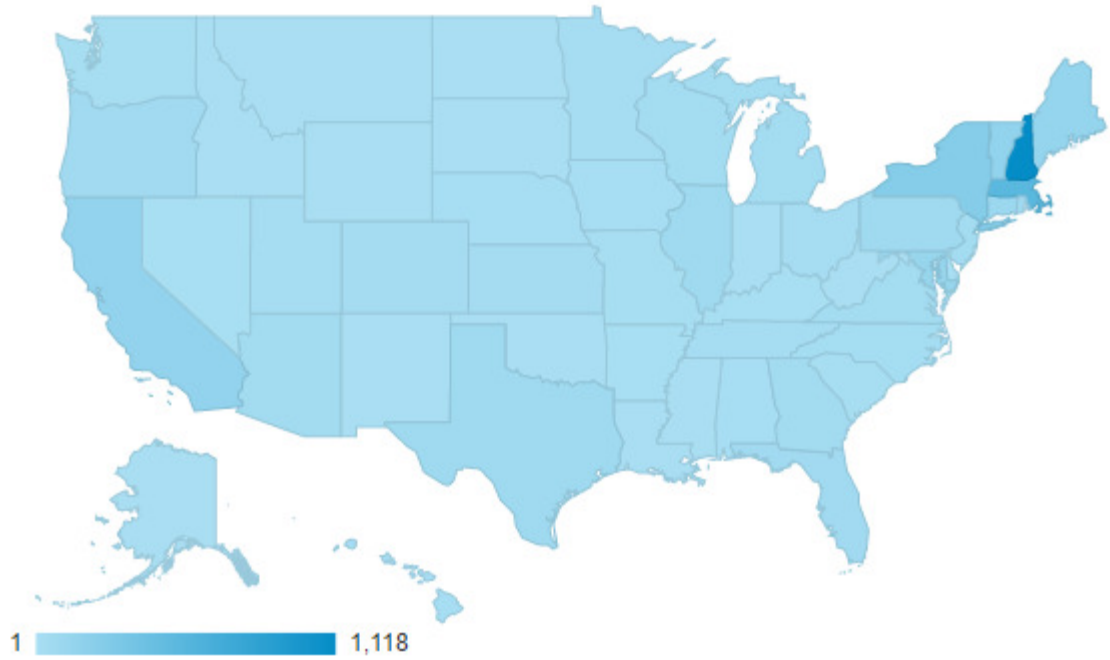
[4] Bounce rate refers to percentage of initial visitors to a site who "bounce" away to a different site, rather than continue on to other pages within the same site

[5] Bounce rate refers to percentage of initial visitors to a site who "bounce" away to a different site, rather than continue on to other pages within the same site

National Outreach of the NEGC

Of 6,759 visits to the NEGC website, 3,712 were from the US and 3,047 were from other countries (with 2,026 of these from Russia[6]). Of the US visits, about one-third were from New Hampshire (1,118)[7]. States with 50 or more visitors include: Massachusetts (537), New York (249), Vermont (165), California (158), Maine (135), Maryland (122), Connecticut (99), Illinois (75), Texas (70), Pennsylvania (66), Florida (58), Rhode Island (56), Oregon (52), Georgia (51). Please see Fig.2.

Figure 2: Distribution of NEGC Website Traffic by State



Resource Leveraging

During Year Nine, NEGC staff did not submit any new grant applications for core or expanded activities.

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

[6] Many of these appear to be bots as the bounce rate for traffic originating from Russia was 98.5% and an average session duration of .08 seconds.

[7] However, 573 (51%) of the NH traffic were generated from the city of Durham (where the administrative staff of the NEGC resides).

COMPLETION OF OBJECTIVES IN YEAR NINE (6/1/15 - 5/31/16)

This section provides an overview of each workgroup's activities during Year Nine. 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.

Table 3 provides a complete list of the objectives set forth by project staff at the beginning of the project year (with modifications based on changes in the project) as well as the status of each objective as of June, 1, 2016. Measures of objective "status" relative to implementation over the course of the 5 year project are defined by the following key: 1. Completed as planned, 2. Completed - deviated substantially from plans, 3. In progress - satisfactory, 4. In progress - unsatisfactory, 5. Initiation of activity deferred, 6. Activity abandoned, 7. Not scheduled to initiate this period, 8. Insufficient documentation available. Additionally, a review is provided on the relative success of the objectives during Project Year Nine. Review results are defined as:

- Successful (24 of 31): All definitions of success for an objective have been fully met or the results of the activity in question fulfill the intent of the measure.
- Partially Successful (5 of 31): Definitions of success for the year only partially met. Although not fully realized, substantive progress has been made in a number of core areas with fulfillment of the goal expected by the next project year.
- Unsuccessful (0 of 31): Although some work on an activity may have been done, primary components of an activity targeted for the year were not substantively addressed within the time period. Lack of success may be due to a number of factors, including lack of participation by certain groups, delays in timeline for other project components, and the need to shift project priorities such that other components could be fulfilled in Year Four.
- NA - Activity abandoned (0 of 31). Project staff determined that the objective/activity in question was no longer applicable to their work and resources have been reallocated to other work of the NEGC.
- NA - Future Activity (2 of 31). Engagement in the activity is dependent on completion of previous project objectives or not scheduled for the current year.

Table 3: Status of Goals and Objectives of the NEGC, Project Year 9

Objective 1. ESTABLISH AND MAINTAIN NEGC				
No.	***Objective***	***Project Status***	***Yr. 9 Definition of Success***	***Yr. 9 Results***
1.1	Continue implementation of core administrative supports to the NEGC	3	NEGC is able to successfully pursue goals and objectives and appropriately respond to changing	Review: Successful All core staff activities completed during course of year.

			conditions.	
1.2	Budget Management	3	Operating expenses for the fiscal year are within the budgeted amounts	Review: Successful Meetings are held regularly to review and supports provided when requested as resources allow.
1.3	Continue close collaboration with WG and AC	3	Collaborative Council and Advisory Committee members feel supported in the work they do and have access to the resources they need to accomplish their goals.	Review: Successful 100% of Advisory members agree that there is a spirit of collaboration, that they received excellent support from RC staff, and that main goals were achieved. All but one (neutral rating) agreed that meetings were constructive and that the NEGC effectively answered questions. Collaborative Council meetings continue to be constructive and aid in the work of the NEGC.
			Meetings and conference calls held	Review: Successful Management meetings were held multiple times per month, 2 meetings of the collaborative council and one face to face and one phone meeting of the Advisory Committee was held.
1.4	Annual meeting	1	Meeting held, Participants understand and are satisfied with the progress of the NEGC.	Review: Successful 60 participated in the Year 9 NEGC Annual meeting. Among those providing feedback on the meeting evaluation forms (N=12-15), 92% agreed that the NEGC was headed in the right direction. all felt that the NEGC had achieved tangible outcomes that resulted in improvements in genetic services in the region, 92% felt they understood what the NEGC would accomplish in the

				next year, and 92% understood how their own work would fit in.
1.5	Communications and outreach plan	3	Stakeholders report satisfaction with being able to voice their opinions and feel that they've been heard.	Review: Successful 100% of respondents at the Annual Meeting indicated that they had an opportunity to share perspectives and contribute to the discussion. Members of Collaborative Council and Advisory Committee meetings report overall satisfaction with meeting progress.
			Consistent increases in NEGC web site utilization.	Review: Successful Between Years 8 and 9, total unique users increased from 2,667 to 5,212. Facebook page likes increased from 320 to 418 and Twitter followers increased from 171 to 307.

Objective 2. NEGC/LEND: *Link LEND program requirements for trainee skill development in technical assistance (TA) and continuing education (CE) to NEGC needs*

No.	***Objective***	***Project Status***	***Yr. 9 Definition of Success***	***Yr. 9 Results***
2.1	Continue to participate with AUCD ad hoc national LEND working group on Genetics Education	3	Bi-directional flow of information established (NEGC and ad hoc group); integration of national with regional	Review: Successful Multiple discussions continue to be held with directors of regional LEND programs to discuss opportunities for collaboration in an effort to meet trainee educational requirements. Efforts coordinated with national level activities between the annual AUCD and NCC meeting.
2.2	Coordinate NE LEND trainees' requirements in	3	Shared understanding of	Review: Partially Successful Three Leadership in Action

	skill development for CE/TA with NEGC		TA/CE requirements and NEGC needs Implement measures of quality and success of TA/CE	activities were identified and pursued between the NH/Maine LEND program and the NEGC. Review meetings are held during and after TA activities occur to ass quality of experiences and identify recommendations for improvement. No new collaboration activities were pursued with other LEND programs in the region.
Objective 3. Education & Outreach: *Continually improve and expand the Genetics Education Materials for School Success (GEMSS) website, and disseminate/promote this resource to a broad range of audiences*				
No.	***Objective***	***Project Status***	***Yr. 9 Definition of Success***	***Yr. 9 Results***
3.1	Update GEMSS	3	Conditions and supporting material updated	Review: Successful. Website was continuously reviewed and updated. In particular, a Leadership team reviewed the site in detail and provided staff with a range of recommended improvements to the site.
3.2	Develop resources for additional conditions	3	Additional conditions content developed for GEMSS	Review: Successful. Three new conditions added during Year 9.
3.3	Promote and disseminate GEMSS via Parent Ambassadors and NH Leadership	3	Parent Ambassadors recruited, Presentations made, Training seminars given	Review: Successful NH Leadership Action Group - Parent Ambassadors - thoroughly reviewed the site and provided a range of useful recommendations, dedicated Facebook pages; publications about GEMSS in various newsletters; presentations at major events, and collaboration with partners involved in similar missions.
Objective 4. Medical Home: *Continuously improve and expand the workforce needed to care for those children with disorders identified*				

by NBS programs				
No.	***Objective***	***Project Status***	***Yr. 9 Definition of Success***	***Yr. 9 Results***
4.1	Implement genetics in primary care webinars	1	Webinars implemented with high satisfaction ratings. # webinars held, # participants, satisfaction ratings of participants.	Review: Successful The NEGC held 4 webinars (225 total participants) educating care providers around concepts of creating a shared plan of care, documenting family history, incorporating genetics in the management of children in the primary care office, and holding family centered conversations. Based on satisfaction surveys collected across the 4 webinars held, participants agreed that the educational session: enhanced their competence (97%, N=74); influenced their practice (90%, N=70); and impacted patient outcomes (90%, N=70). More details on the webinar series (including recordings) are accessible here: http://www.negenetics.org/work-groups/medical/products_pubs_mh.
Objective 5. Transition: *Continuously improve and expand the workforce needed to care for those children with disorders identified by NBS programs*				
5.1	Assess health in young adults with genetic and metabolic conditions	3	Development and implementation of health survey incorporating modified Erikson Psychosocial Stage Inventory # of surveys distributed, analysis completed, report	Review: Successful To date, survey data has been collected from 85 adults living with a condition and 52 who do not. Provides information suggesting early indications of increased onset of older adult health challenges among the young adult population living with genetic conditions.

			completed	
5.2	Develop strategy to obtain data on transition outcomes	3	Include both qualitative and quantitative data; work in sync with NCC Transition work group to obtain outcomes # of new data collection efforts initiated to track transition outcomes	Review: Partially Successful Continued support for transition activities at Children's Hospital Boston, pursuit of new grant opportunities enabling long term follow up, continued supports for IBEM, collaboration with Next Step on outcome measures for youth .
5.3	Leadership Training	1	Youth complete the program and report high satisfaction Youth are engaged to become effective advocates.	Review: Successful Face Forward Camp: 18 youth participated in the summer Face Forward event. In a pre and post comparison of participant perceptions concerning whether or not they would be willing to advocate for themselves or others living with their condition, the percentage increased from 78% to 92%. Homocystinuria group (8 members) continues to meet providing mutual group support to members.

Objective 6. Health Care Access and Financing: *Engage and educate families, stakeholders, and decision makers in each New England state using existing information about coverage, costs, and benefits related to genetic conditions / Assess family-based definitions of needed benefits and coverage related to genetic conditions and educate stakeholders on the potential implications of the research for accessing quality care within each state's policy environment*

No.	***Objective***	***Project Status***	***Yr. 9 Definition of Success***	***Yr. 9 Results***
6.1	Design and implement research strategy for 2nd research stage	1	New survey developed with input from core stakeholders.	Review: Successful The HAF group successfully completed the design,

				implementation, and analysis of a new survey of families concerning health care access and financing of health care for their children living with a genetic condition.
6.3	Creation of a policy brief and web seminar highlighting findings from the 2nd research stage.	1	Key decision makers are better informed about the coverage, costs, and benefits of quality services for those with genetic conditions	Review: Partially Successful To date, the research brief was made available on the website and shared with local partners, but a large push to share the work has not yet occurred. Plans are in place to further disseminate via the NEGC newsletter, IOD contacts, and area Family Voices organizations. Positive feedback was received from those reviewing receiving copies of the report.
6.4	Collaborate with regional Family Voices partner organizations to develop and disseminate material	3	Families are better prepared to self-advocate for needed benefits to decision makers	Review: NA - Future Activity Activity to be undertaken during Year 5.
Objective 7. Quality Improvement: *Engage all centers in continuous quality improvement of metabolic center care of patients with inborn errors of metabolism*				
No.	***Objective***	***Project Status***	***Yr. 9 Definition of Success***	***Yr. 9 Results***
7.1	Obtain human subjects review committee waivers and business associates agreement (BAAs) from all participating centers	3	IRB exemptions obtained and BAAs are executed	Review: Successful Waivers and BAAs are filed with NEGC/UNH Obtained human subjects review committee waivers and BAAs from all centers entering data for DD/ID (N=5). Ten sites currently participating in the overall process at varying levels of involvement.
7.2	All centers have web-portal access to registry	3	Data entered into registry	Review: Successful All participating centers who are entering data have access to the

				registry.
7.3	Data analysis	3	Data are analyzed	Review: Successful Available data was reviewed by NEGC staff and feedback provided to members of the collaborative. As of May 2016, the registry has records for 2,042 individuals with DD/ID.
7.4	Compare care processes to standard care guidelines.	3	Collaboration made easy and user-driven	Review: Successful Where standard care guidelines exist, these are incorporated into review processes of the group. For example, PKU guideline has been developed by the American College of Medical Genetics (includes W. Smith). Will be used by Metabolism teams and NEGC to assess care practices in NE as new ones are identified. This is ongoing.
7.5	Utilized modern social media and telecommunications to support quality improvement learning and action.	3	Workgroup participation is improved via effective use of communication technologies.	Review: Successful Teleconference / Zoom continues to be utilized for online conferences. Additional resources available online to aid in utilization of QI resources.
7.6	Update registry with CSHCN data variables and additional performance indicators.	7	Expanded regional capacity to adopt QI processes for metabolic centers	Review: Deferred Registry to be updated once initial data collection stages are completed.

7.7	Expand QI process to additional centers.	3	Additional centers are added to the learning collaborative.	Review: Partially Successful Although no new centers have joined the NEGC's efforts in this area, the NEGC has been successful in recruiting new staff from participating centers (particularly from Children's Hospital Boston) to participate in the initiative.
Objective 8. Public Health Infrastructure: *Build capacity in state public health (PH) departments to enhance and sustain the delivery of newborn screening and follow-up and treatment services*				
No.	***Objective***	***Project Status***	***Yr. 9 Definition of Success***	***Yr. 9 Results***
8.1	Pilot project: Critical congenital heart disease (CCHD) screening	1	CC meeting held (1 per year); educational materials developed and distributed; screening and follow-up and data collections protocols for each facility documented; Completion of evaluation reports, development of resource page for children and families living with CCHD.	Review: Successful Pilot project implemented for: Critical congenital heart disease (CCHD) newborn screening. The project established a consistent method for screening and reporting of CCHD across New England. Over 42,000 children screened since the project started, 3 children diagnosed with CCHD. An electronic resource was developed for families on psychosocial supports and went live in May 2016 (www.necongenitalheartresources.org). 500 resource bags were distributed to 4 pediatric cardiology practices for distribution to families that receive a CCHD diagnosis.
Objective 9. QUANTITATIVE AND QUALITATIVE EVALUATIONS				
No.	***Objective***	***Project Status***	***Yr. 9 Definition of Success***	***Yr. 9 Results***

9.1	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.	Review: Successful Evaluation and survey data are used to inform NEGC activities.
9.2	Conduct annual stakeholder survey	3	A majority of stakeholders participate in the survey process and provide recommendations for the project's improvement	Review: Partially Successful Only 28% of stakeholders invited to participate in the survey did so (20% in Year 8).
9.3	Complete annual reports which can be used by staff to improve project outcomes	3	Reports completed and utilized by staff to improve project outcomes and utilized by stakeholders to stay informed of project progress.	Review: Successful Evaluation information is used regularly to improve project outcomes.
9.4	Participate on national outcome measurement efforts	3	NEGC is actively represented on national measurement efforts.	Review: Successful NEGC was represented on all meetings and provided information for all national level reporting and discussions.

PLANS FOR NEGC YEAR 10 (6/1/16-6/31/17)

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

Table 4: Year 10 Goals and Objectives

Activity	Outcome	Outcome Indicator
NEGC/LEND: *Link LEND program requirements for trainee skill development in technical assistance (TA) and continuing education (CE) to NEGC needs*		
Continue to participate with AUCD ad hoc national LEND working group on Genetics Education	Bi-directional flow of information established (NEGC and ad hoc group); integration of national with regional	Curricular materials agreed upon and implemented by program with "implementation reports" by each LEND
Education & Outreach: *Continually improve and expand the Genetics Education Materials for School Success (GEMSS) website, and disseminate/promote this resource to a broad range of audiences*		
Develop resources for additional conditions	Additional conditions content developed for GEMSS	Conditions added to GEMSS website and new resources added.
Evaluate the effectiveness of GEMSS	Draft document of survey and demonstration results; recommendations for changes	Document of survey results concerning learning module feedback.
Promote and disseminate GEMSS	Presentations made Training seminars given	# of presentations made and to which groups # of training seminars given and to which groups
Medical Home/Care Coordination: *Continuously improve and expand the workforce needed to care for those children with disorders identified by NBS programs*		
Support integration of genetics in primary care	Webinar implemented focusing on sharing information on Family Voices organizations with participating physicians as well as best practice for writing a medical need letter for families. GEMSS website resources will serve as a resource for primary care audiences.	# webinars held, # participants, satisfaction ratings of participants.
Transition: *Improve understanding and resources available to support successful transitions among youth*		
Assess health in young adults with genetic and metabolic conditions	Develop, distribute, and analyze Adult Health Survey	# of participants completing the survey, report developed, and # of report recipients Report posted on NEGC
Develop strategies to improve leadership and connections among young adults and their service providers concerning	Youth are engaged to become effective advocates. Primary care providers adopt age appropriate transition plans	# of youth participating / completing the Face Forward program. # clinics utilizing transition plans.

successful transitions		
Health Care Access and Financing: *Engage and educate families, stakeholders, and decision makers in each New England state using existing information about coverage, costs, and benefits related to genetic conditions / Assess family-based definitions of needed benefits and coverage related to genetic conditions and educate stakeholders on the potential implications of the research for accessing quality care within each state's policy environment*		
Creation of resources to improve parent ability to be more effective advocates for their children's health.	Families receive resources and training in how become better advocates for their children's health care.	# of family members of a child with a genetic condition receiving resources, # of unique downloads, satisfaction ratings of reviewed materials
Assess the impact of the 2015 HAF policy brief	Data is collected documenting a range of impacts of the 2015 HAF policy brief.	Evaluator review of materials received.
Create a suite of support materials for physicians receiving training via the Medical Home workgroup webinar focused on physicians and family voices organizations.	Physicians receive an array of materials that will improve their ability to effectively advocate for needed health care services among children.	# of physicians attending, satisfaction ratings, downloads of website materials.
Quality Improvement – DD / ID / PKU / MCADD Project: *Engage all centers in continuous quality improvement of metabolic center care of patients with inborn errors of metabolism*		
Obtain human subjects review committee waivers and BAAs from all centers	IRB exemptions obtained and BAAs are executed	Waivers and BAAs are filed with NEGC/UNH
All centers have web-portal access to registry	Data entered into registry	# of patients for whom complete data are entered
Data analysis	Data are analyzed	Reports back to metabolic teams; # of publications
Compare care processes to standard care guidelines.	Collaboration made easy and user-driven	Best practices in care guidelines are reviewed by the group and incorporated as appropriate.
Utilized modern social media and telecommunications to support quality improvement learning and action.	Workgroup participation is improved via effective use of communication technologies.	A range of technologies is utilized to enhance communication and participation among workgroup members
Update registry with CSHCN data variables and additional performance indicators.	Expanded regional capacity to adopt QI processes for metabolic centers	Registry updated with CSHCN data variables and other performance measures as identified by the workgroup.
Expand QI process to additional	Additional centers are added to the	# of regional and national centers

centers.	learning collaborative.	added
GMCE: *Strengthen capacity of primary care providers to effectively diagnose and treat individuals with inherited metabolic disorders*		
Monitor and seek to improve collaboration activities with the GMCE	Expand successful utilization of GMCE by NEGC stakeholders	# of clinics participating in the GMCE, % expressing high satisfaction with supports received.

CONCLUSIONS AND RECOMMENDATIONS

During Project Year Nine, the NEGC had a very productive year while also putting in place critical activities for completion in Year Ten. Among its key accomplishments:

- Creation of a new body of research helping stakeholders to better understand the challenges faced by families seeking to access care for their children living with a genetic condition.
- Strong growth and utilization of the NEGC and GEMSS websites, while continuing to add new material and review for effective resources.
- Implementing a highly successful series of medical home webinars, each of which garnered strong ratings of satisfaction from participants.
- Creation of a new electronic resource concerning psychosocial supports for families with children born with CCHD
- Continued development of the QI collaborative, a nationally unique partnership of 10 clinical sites which have partnered together to form a learning community around improving the care of individuals living with DD/ID, PKU, and MCADD. To date, five of these clinics are regularly entering data into the DD/ID data registry, with 2,042 records entered.
- Launching of the GMCE telehealth and education platform with pilot tests among 3 participating clinics and 19 consultations completed
- A highly successful and informative annual meeting, where 100% of respondents felt that the NEGC had achieved tangible outcomes that resulted in improvements in genetic services in the region
- Ongoing efforts were made to provide presentations and trainings this past year along with publishing research in peer reviewed journal articles (16 presentations, 3 publications, and 17 CE/TA activities)

In looking forward to future activities of the NEGC, there continue to be a few areas identified in the previous year's report that that would be of value to review on a regular basis during project management meetings and to develop an action plan around in order to continue the NEGC's focus on improved outreach and impact:

- *Keeping the NEGC website current* - it will continue to be helpful to establish and monitor a common time frame for when minutes should be reviewed by participants, finalized, and posted to the website (for example, within one week of approval by meeting participants).
- *Continue to build on and expand organizational partnerships* by taking advantage of existing collaborations and identifying new ones in order to help drive more traffic to the NEGC websites. It would be helpful to review the sites that are and are not referring individuals to the NEGC and outreaching to those sites where the NEGC believes a natural partnership exists.

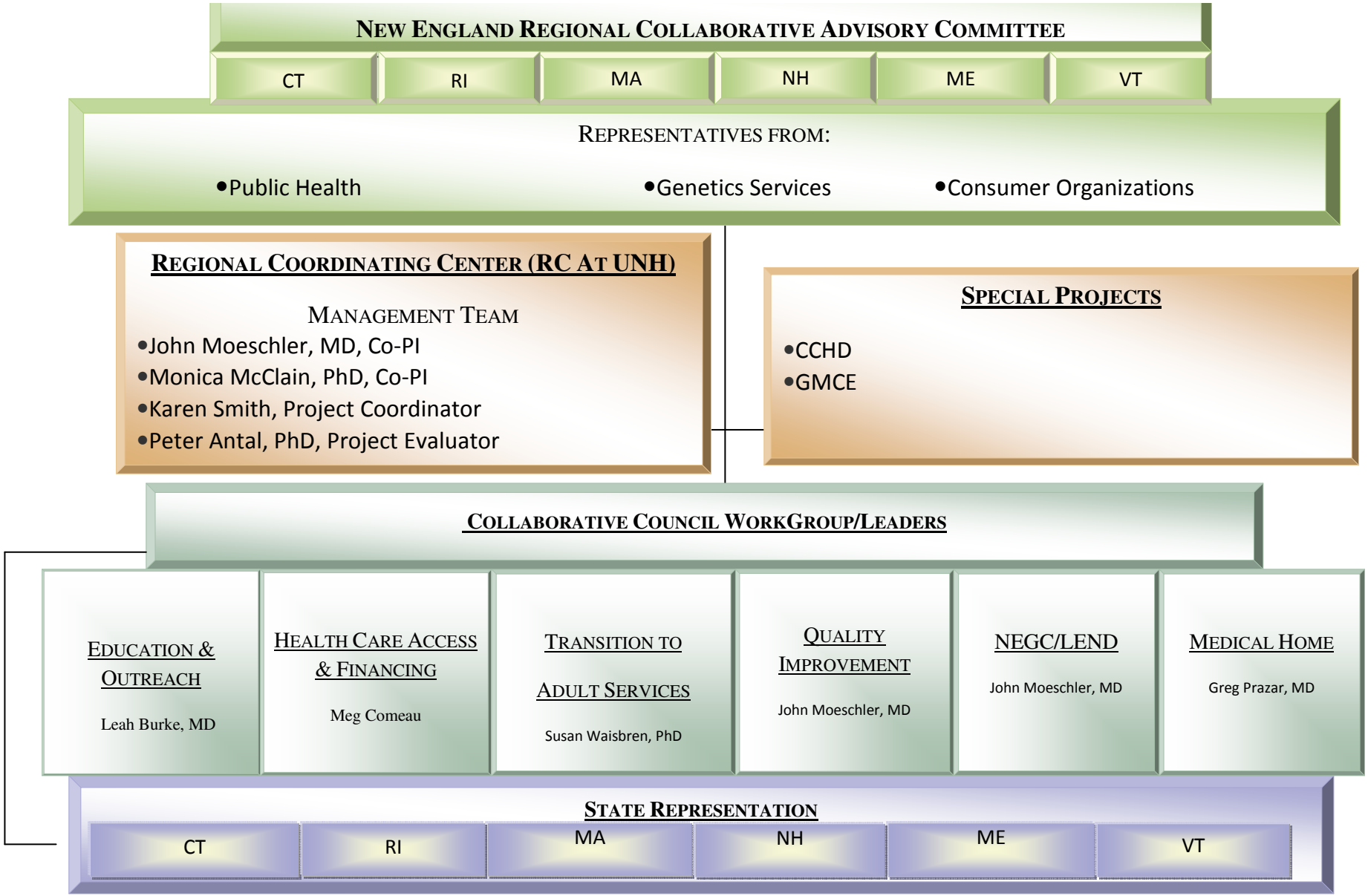
- *Identify resources that may be underutilized and develop a plan for improving access and utilization of these resources.* Since its inception, the NEGC has been able to make a broad array of resources available - transition plans, toolkits, assessment resources, educational brochures, and the GEMSS website just to name a few. It would be helpful to review these resources in light of who and how many people are accessing them and identify potential market gaps which could be filled if these resources were made more easily accessible (or more broadly marketed).
- *Address Sustainability Challenges for Core NEGC Activities.* As the NEGC is nearing completion of its current five year cycle, it will be critical that clear strategies for sustainability be outlined and implemented in collaboration with NEGC stakeholders. In the event that future cycles do not fund some of these areas that have been identified as a clear need for our region, new supports should be identified to ensure their continuation beyond the life of the current grant. Such supports may take the form of new grant applications, internalizing activities within the structures of partner organizations, business sponsorships, and targeted fund raising.

Respectfully Submitted,

Peter Antal, Ph.D.
NEGC Project Evaluator

APPENDIX A: NEGC ORGANIZATIONAL CHART

NEW ENGLAND REGIONAL COLLABORATIVE ORGANIZATIONAL CHART FOR 2014/2015



APPENDIX B: NEGC PRESENTATIONS LIST

*** New in Year Nine**

Sharing Work on Project Activities

** "Genetics Education Materials for School Success Learning Module"; NEGC Annual Education & Outreach Work Group Meeting, Portsmouth, NH; April 7,2016.*
Baril, E.

** "Genetics Education Materials for School Success: School Can Be a Great Place for Everyone." The Center for START Services Annual Conference,*
March 14, 2016 .
Dillon, A. & Smith, K.

** "Genetics and Metabolic Center for Education". New England Regional Genetics Group; Portsmouth, NH.*
December 3, 2015;
Korson M.

** "Genetic Counseling via Telemedicine". New England Regional Genetics Group; Portsmouth, NH.*
December 3, 2015;
Smith R and Upton S.

** "Genetics Education Materials for School Success (GEMSS)". American with Disabilities Act Equity and Access Fair, University of New Hampshire; Durham, NH.*
November 18, 2015;
Smith K.

** " Transition Policy. New England Consortium of Metabolic Programs; Boylston, MA.*
November 13, 2015
Waisbren S.

** "Genetics and Metabolic Center for Education." New England Consortium of Metabolic Programs; Boylston, MA.*
November 13, 2015;
Korson M.

** "Genetics and Metabolic Center for Education". National Coordinating Center Annual Meeting; Washington, DC.*
November 12, 2015;
Korson M.

** "Genetics Education Materials for School Success (GEMSS) for Neuromuscular Disorders". Muscular Dystrophy Regional Conference, New Hampshire.*
November 7, 2015;
Dillon A, Fox S.

** "GEMSS for Neuromuscular Disorders." Muscular Dystrophy Regional Conference; New Hampshire.*
November 7, 2015;
Dillon A.

** "Adult Health Survey". Brigham & Women's Hospital Fellows' Program; Boston, MA.*
September 2015;
Rajibi F.

"NEGC Update", Consortium of Metabolic Programs Annual meeting; Boylston MA; Nov. 2014.
Karen Smith

"Moving on with Mito Overview"; Consortium of Metabolic Programs Annual meeting; Boylston MA; Nov. 2014.
Rob Auffrey

"Quality Improvement in care provided by general genetics clinics and in care for patients with PKU and MCAD deficiency: project overview, methodology, and results to date"; NCC/RC/PD Meeting, Washington, DC Nov. 2014
John Moeschler

"HRSA MCHB Grants for CCHD – RC Partnership Opportunity (Dissemination of information/resources/products)";
NCC/RC/PD Meeting, Washington, DC
Nov. 2014
Monica McClain

"QI Developmental Delay Registry: What it takes to secure a diagnosis" NERGG Annual Meeting / Collaborative Session; Portsmouth, NH; Dec 4, 2014
John Moeshler

"GEMSS – Genetics Education Materials for School Success: Parent Voices." AMCHP; Jan 2015
Anne Dillon

"Public Health Approaches for Implementing or Supporting Cascade Screening for Tier 1 Genetic Conditions" Public Health Special Interest Group, American College of Medical Genetics and Genomics, Salt Lake City, UT, March 2015
Monica McClain

"QI Developmental Delay Registry: What it takes to secure a diagnosis" QI Special Interest Group, American College of Medical Genetics and Genomics, Salt Lake City, UT, March 2015
John Moeschler

"NEGC Genetics Workforce Project"; NYMAC Summit; May 28th 2015
Monica McClain

The New England metabolic centers program to improve care for patients with inherited metabolic disorders. Society for Inherited Metabolic Disorders Annual Meeting, March 31 – April 3, 2012, Charlotte, NC.
Smith W, Martin M, Greenstein RM, Korson M, Levy H, Waisbren SE, Moeschler JB, Cooley WC, McAllister JW, Antal P, McClain MR.

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

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

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

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

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

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

Poster session:
- *NEGC*
- *NEGC Work Groups*
- *Innovative Projects*

NEGC Annual Meeting
Dec 2009

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

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

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

Poster session:
- *NEGC*
- *NEGC Work Groups*
- *Innovative Projects*
- *CSHN Survey Analysis Presentation – Bob McGrath*

NEGC Annual Meeting
Dec 2008

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

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

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

Educating Students

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

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

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

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

Training Professionals

** "Using state birth defects registries to evaluate outcomes of critical congenital heart disease newborn screening." American Public Health Laboratories Newborn Screening and Genetic Testing Symposium, St. Louis, MO, February-March 2016.*
McClain MR.

** "A World of Uncertainty: Impact on Parents of a Diagnosis of Critical Congenital Heart Disease (CCHD)." Grand Rounds at UCSF Benioff Children's Hospital, Oakland, CA.*
September 15, 2015;
Fanos J, Landon C, Bose A, McClain M.

** "Transition Policy." Post Clinic at Children's Hospital Boston, Boston, MA.*
September 29, 2015;
Waisbren S.

"The Affordable Care Act and Access to Genetic Services: Opportunities and Challenges", NERGG Annual Meeting / Collaborative Session; Portsmouth, NH; Dec 4, 2014
Meg Comeau

"New England Children with Genetic Disorders and Health Reform: Information and Recommendations for State Policymakers". Webinar. Meg Comeau; Sept 2014

"Returning to Therapy & the ACMG Diagnosis & Management Guidelines, American College of Medical Genetics, Salt Lake City, Utah (BioMarin, Pharmaceuticals)
Susan Waisbren

"Welcome to Holland: The Impact on Parents of a Diagnosis of CCHD"; NEGC Annual Meeting; Portsmouth, NH; April 2015
Joanna Fanos

"Living with Distinction: the Psychosocial Correlates of Genetic Disorder Related Stigma"; NEGC Annual Meeting; Portsmouth, NH; April 2015
Sondra Solomon

"Adult Health Issues"; NEGC Annual Meeting; Portsmouth, NH; April 2015
Farrah Rajib

"Envisioning the Future – How GEMSS can Help!"; NH Family Support Conference; May 2015
Ann Dillon and three members of the GEMSS Action Group

"Why Medical Home and Care Coordination are Important for Children"; Webinar; May 2015
Jill Rinehart, Jeanne McAllister

NBS Follow Up, Uniform Assessment Method, and Transition
Presented to Genetic Counselors at Children's Hospital Boston by Susan Waisbren, PhD; May 6, 2014.

Development of Educational Brochure for Mitochondrial Disease
Presented to Health Communication Fellows & Preceptors at Emerson College by Robert Auffrey, under supervision of Susan Waisbren; May 2014.

Adults Health Concerns: Are our young people aging prematurely?
Presented to NEGC Transition Work Group at Annual Meeting by Susan Waisbren; April 16, 2014.

Uniform Assessment Method
Oral presentation by Dr. Waisbren, PhD, at NE Consortium of Metabolic Programs annual meeting; Boston, MA; November 15, 2013.

Cognitive Reserve and Establishing Identity in PKU and Galactosemia
Oral presentation by Dr. Waisbren, PhD, at Albany Medical Center; Albany, NY; Oct. 5, 2013.

Psychological Outcomes for Urea Cycle Disorders
Oral presentation by Susan Waisbren, PhD; International UCD Consortium (I-SIMD); Spain; September 2013.

The Adult Galactosemic Phenotype.
Oral presentation by Susan Waisbren, PhD, of paper published in JIMD in 2012 for which Dr. Waisbren received the Archibald Garrod award; I-SIMD; September 2013.

2013 4th Annual Northern New England Symposium on Marfan Syndrome & Related Disorders, Booth displaying GEMSS and other NEGC materials, UVM COM, September 14, 2013

Sickle cell community based collaborative research: the roles and experiences of sickle cell advocacy organizations. American College of Medical Genetics Annual Meeting, March 19-23, 2013; Phoenix, AZ.
Pertillar V, Williams-Edwards D.

Quality improvement in the clinical genetic evaluation of patients referred for global developmental delays/intellectual disabilities or autism.
American College of Medical Genetics Annual Meeting, March 19-23, 2013; Phoenix, AZ.
Moeschler JB, McClain MR, Burke LW, Dinulos MB, Smith R, Smith W, Miller P.

Newborn screening and the potential for whole genomic screening of newborns: Parental response to true positive, false positive and inconclusive results. Grand Rounds at Rhode Island Hospital. January 2013. Susan Waisbren.

"I am more than a diagnosis!" Impact of Personal Identity on Health Care Transitions for Young Adults with Special Health Care Needs. Got Transition Webinar Series, Webinar #6. November 2012. Susan Waisbren, Mallory Cyr.

Transition to Independent Living. Got Transition Webinar Series. Live from Face Forward Summer Conference for Youth, Ashland, MA. July 2012. Susan Waisbren, Mallory Cyr.

Exploring the Role of the School Nurse as a Facilitator of Health Care Transition. Maine Nurses' Association Summer Institute. July 2012. Carol Orton.

Keynote to 2012 Galactosemia Foundation Conference; topic included Transition. Dallas, TX. July 2012. Susan Waisbren.

Genetics Education Materials for School Success(GEMSS).
13th International Williams Syndrome Conference, July 2012; Boston, MA.
Giummo C.

Outcomes of clinical genetic evaluation of 186 patients referred for intellectual disability or global developmental delays of unknown cause.
International Association for the Scientific Study of Intellectual Disabilities World Congress, July 9, 2012; Halifax, Nova Scotia.
Moeschler J.

Caring for patients with metabolic disorders from positive newborn screen to year 1: provider workload, workflow, and issues for the medical genetics workforce. Association of American Medical Colleges Physician Workforce Research Conference, May 3-4, Washington, DC.
McGrath RJ, Stransky M, Benkendorf J.

Presented abstract on GEMSS as a poster at ACMG annual meeting and published in ACMG Annual Meeting Abstract book
Dr. Leah Burke

Post-Analytic Molecular Challenges: Algorithm development, Clinical interpretation, Reporting Data and Reporting Risk. CDC Molecular Training Workshop, Atlanta, GA, May 2012.
Dr. Anne Comeau (oral presentation)

Improved interpretation of newborn screening results using predictive indices. Mass General Hospital Genetics Conference, 2012, Boston, MA.
Sahai, I.

Long term outcomes in newborn screening; Pediatric Academic Society; May 1, 2012; Boston, MA; Member of the pane.
Dr. Susan Waisbren

Long Term Outcomes in Newborn Screening; Levey Symposium; Boston, MA; April 2012, Waisbren, S.

Genetics education materials for school success: a guide for the classroom for children with genetic conditions.

American College of Medical Genetics Annual Meeting, March 27-31, Charlotte, NC.
Burke LW, Burke B, Dillon AD, Giummo C, Larson F, Lavochkin M, Mulcahy E, Smith W, Tutko H, Williams-Edwards D.

The New England metabolic centers program to improve care for patients with inherited metabolic disorders. Society for Inherited Metabolic Disorders Annual Meeting, March 31 – April 3, 2012, Charlotte, NC.
Smith W, Martin M, Greenstein RM, Korson M, Levy H, Waisbren SE, Moeschler JB, Cooley WC, McAllister JW, Antal P, McClain MR.

New England birth defects consortium: 6-state folic acid/multivitamin distribution project. National Birth Defects Prevention Network Annual Meeting, February 27-29, 2012, Arlington, VA.
Miller S, Haberman D, Wall T, Mason C, Tu S, Brozicevic P, Davis K, Davis J, Viner-Brown S, Arias W, Higgins C, Anderka M.

Long-Term outcomes and Management across the Lifespan; NIH conference: PKU Scientific Review Conference: State of the Science and Future Research Needs; Feb 2012, Bethesda, MD
Dr. Susan Waisbren

Transition Issues; Urea Cycle Disorder Consortium, Boston, MA, Jan 20, 2012,
Waisbren, S.

Transition as a Psychological Rite of Passage; New England Regional Genetics Group (NERGG) Annual Conference Collaborative Session; December 1, 2011; Portsmouth, NH; Waisbren, S.

Quality Improvement. The 2011 Newborn Screening and Genetic Testing Symposium, San Diego CA, November 2011.
Dr. Roger Eaton (Panel Leader)

Multicenter validation of algorithsm to improve communications of positive NBS results to the medical home. Newborn Screening and Genetic Testing Symposium, Nov. 2011, San Diego, CA.
Sahai, I.

Factors that can Influence the Immunoreactive Trypsinogen (IRT) Concentrations on Dried Blood Spot Samples. The 2011 Newborn Screening and Genetic Testing Symposium, San Diego CA, November 2011.
Dr. Roger Eaton (poster presentation)

The Long and Short of Newborn Screening for LCHAD: The New England Experience. The 2011 Newborn Screening and Genetic Testing Symposium, San Diego CA, November 2011. Dr. Inderneel Sahai (oral presentation)

Congenital Hypothyroidism in Multiple Births. The 2011 Newborn Screening and Genetic Testing Symposium, San Diego CA, November 2011. Dr. Inderneel Sahai (poster presentation)

Outcomes of clinical genetic evaluation of 186 patients referred for intellectual disability or global developmental delays of unknown cause. American Society of Human Genetics Annual Meeting, October 11-15, 2011, Montreal, Canada.
Moeschler JB, McClain MR, Burke LW, Dinulos MB, Smith R, Smith W, Miller P.

LTFU and Transition Issues; Maine PKU Network: Family Weekend, September 2011

Laboratory aspects of IRT screening: Quality assurance activities. Joint APHL and HRSA conference on IRT as a Biomarker for Cystic Fibrosis: Technical Issues and Challenges for Newborn Screening. Bethesda, MD, May 2011.
Roger Eaton (oral presentation)

Experiences with IRT analysis in New England. Joint APHL and HRSA Conference on IRT as a Biomarker for Cystic Fibrosis: Technical Issues and Challenges for Newborn Screening. Bethesda, MD, May 2011. Roger Eaton (oral presentation)

Parents' role in specialty referrals: views from both sides of the exam table. Pediatric Academic Societies Annual Meeting, April 28-May 1, 2011, Denver, CO.

Fischer SH, Cooley WC, Mazor KM, Dworetzky B, Stille CJ.

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

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

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

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

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

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

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

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

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

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

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

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

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

Development of Collaborative Organizations.
National Coordinating Center of the Newborn Screening and Genetics Collaborative meeting.
Chicago, IL.
June 5, 2009.
John Moeschler

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

Genetic Health Care Quality Improvement.
Annual Meeting of the National Newborn Screening and Genetics Coordinating Center, Bethesda MD.
January 7, 2009.
John Moeschler

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

Keynote address
International Conference for Adults and Children with PKU, Chicago, IL
Aug 2008
Susan Waisbren

Transition: Psychosocial Considerations
(power point presentation, available on NEGC website)
Susan Waisbren

Innovative Project: Sickle Cell Disease Life Skills Training to Improve Outcomes
Multiple presentations to young adults in NE
2007-2009
Bill Kubicek, Next Step, project PI

Communication of relative risk for cystic fibrosis following a positive newborn screening result. Newborn Screening and Genetic Testing Symposium, November 3-6, 2008, San Antonio, TX
Hale JE, Parad RB, Dorkin HL, Gerstle r, Lapey A O'Sullivan BP, Spencer, T, Yee W and Comeau AM.

Quality measures enhanced by short and long-term follow up in a newborn screening program collaborating with multiple centers.
University of Massachusetts Medical School/Commonwealth Medicine Conference, October 25, 2007, Worcester, MA.
Hale JE, Parad RB, O'Sullivan BP, Quizon AI, Martin T, Yee W, Dorkin HL, Comeau AM.

Quality measures enhanced by short and long-term follow up in a newborn screening program collaborating with multiple centers.
21st 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 C: NEGC PUBLICATIONS LIST

*** New in Year Nine**

Peer-Reviewed Journal Articles

1. * McClain MR, McGrath R, Stransky ML, Benkendorf JL. National survey of providers treating patients with metabolic disorders identified by newborn screening demonstrates challenges faced by clinical care systems. Clinical Pediatrics, 54:759-64. 2015.
2. * Waisbren, S. E., He, J., & McCarter, R. (2015). Assessing Psychological Functioning in Metabolic Disorders: Validation of the Adaptive Behavior Assessment System, (ABAS-II), and the Behavior Rating Inventory of Executive Function (BRIEF) for Identification of Individuals at Risk. JIMD Reports, 21, 35.
3. * Waisbren SE, Rohr F, Anastasoaie V, Brown M, Harris D, Ozonoff A, Petrides S, Wessel A, Levy HL. Maternal Phenylketonuria: Long-term Outcomes in Offspring and Post-pregnancy Maternal Characteristics. JIMD Rep. 2015;21:23-33. doi: 10.1007/8904_2014_365. Epub 2015 Feb 25.
4. John B. Moeschler, MD, MS, FAAP, FACMG, Michael Shevell, MDCM, FRCP, COMMITTEE on GENETICS. Comprehensive Evaluation of the Child With Intellectual Disability or Global Developmental Delays. *Pediatrics,* August 25, 2014 released online.
5. Longo N, Siriwardena K, Feigenbaum A, Dimmock D, Burton BK, Stockler S, Waisbren S, Lang W, Jurecki E, Zhang C, Prasad S. Long-term developmental progression in infants and young children taking sapropterin for phenylketonuria: a two-year analysis of safety and efficacy. 2014; Epub ahead of print.
6. McClain MR, Cooley WC, Keirns T, Smith A. A survey of the preferences of primary care physicians regarding the co-management with specialists of children with rare or complex conditions. *Clinical Pediatrics,* 2014; 53:562-6.
7. Waisbren, S. "Uniform Assessment Method for Screening and Outcomes Research: Validation of the Adaptive Behavior Assessment System, 2nd edition (ABAS-2), and the Behavior Rating Inventory of Executive Function (BRIEF)"; submitted February, 2014.
8. Christopher J. Stille, MD, MPH; Shira H. Fischer, MD, PhD; Nancy La Pelle, PhD; Beth Dworetzky, MS; Kathleen M. Mazor, EdD; W. Carl Cooley, MD. Parent Partnerships in Communication and Decision Making. Academic Pediatrics. March-April 2013; 13(2):122-132.
9. Stille CJ, Fischer SH, LaPelle N, Dworetzky B, Mazor KM, Cooley WC. Parent partnerships in communication and decision making about subspecialty referrals for children with special needs. Academic Pediatrics, January, 2013.
10. Waisbren SE, Potter NL, Gordon CM, Green RC, Greenstein P, Gubbels CS, Rubio-Gozalbo E, Schomer D, Welt C, Anastasoaie V, D'Anna K, Gentile J, Guo CY, Hecht L, Jackson R, Jansma BM, Li Y, Lip V, Miller DT, Murray M, Power L, Quinn N, Rohr F, Shen Y, Skinder-Meredith A, Timmers I, Tunick R, Wessel A, Wu BL, Levy H, Elsas L, Berry GT. The adult galactosemic phenotype. J Inherit Metab Dis. 2012. Mar;35(2):279-86.
11. Fanos, J. New "first families": the psychological impact of new genetic technologies. Genetics in Medicine, 2012 Feb;14(2):189-90.
12. Cooley WC, Kemper AR, NCC Medical Home Workgroup. An Approach to Family-Centered Coordinated Co-management for Individuals with Conditions Identified through Newborn Screening. Genetics in Medicine, 2012, in press

13. Mitchell, M.L., Hsu, H.W., Sahai, I., & Massachusetts Pediatric Endocrine Work Group. (2011). The increased incidence of congenital hypothyroidism: Fact or fancy? Clinical Endocrinology, 75(6), 806-10.
14. Sahai, I., Bailey, J.C., Eaton, R.B., Zytkovicz, T., & Harris, D.J. (2011). A near-miss: very long chain acyl-CoA dehydrogenase deficiency with normal primary markers in the initial well-timed newborn screening specimen. Journal of Pediatrics, 1(1), 172.
15. Kemper AR, Kus CA, Ostrander RA, Comeau AM, Boyle CA, Dougherty D, Mann MY, Botkin JR, Green NS. Implementing Point of Care Newborn Screening. Genetics in Medicine *In press.*
16. Waisbren SE, Potter NL, Gordon CM, Green RC, Greenstein P, Gubbels CS, Rubio-Gozalbo E, Schomer D, Welt C, Anastasoaie V, D'Anna K, Gentile J, Guo CY, Hecht L, Jackson R, Jansma BM, Li Y, Lip V, Miller DT, Murray M, Power L, Quinn N, Rohr F, Shen Y, Skinder-Meredith A, Timmers I, Tunick R, Wessel A, Wu BL, Levy H, Elsas L, Berry GT. The adult galactosemic phenotype. J Inherit Metab Dis. 2012 Mar;35(2):279-86. Epub 2011 Jul 21. PubMed PMID: 21779791.
17. McGrath RJ, Stransky ML, Cooley WC, Moeschler JB. National profile of children with Down Syndrome: disease burden, access to care, and family impact. J Pediatr. 2011
18. Woo HC, Lizarda A, Tucker R, Mitchell ML, Vohr B, Oh W, Phornphutkul C. Congenital hypothyroidism with a delayed thyroid-stimulating hormone elevation in very premature infants: incidence and growth and developmental outcomes. J Pediatr. 2011;158(4):538-42.
19. Gubbels CS, Maurice-Stam H, Berry GT, Bosch AM, Waisbren S, Rubio-Gozalbo ME, Grootenhuis MA. Psychosocial developmental milestones in men with classic galactosemia. J Inherit Metab Dis. 2011 Apr;34(2):415-9. Epub 2011 Feb 25. PubMed PMID: 21350966; PubMed Central PMCID: PMC3112026.
20. Sahai I, Eaton RB, Hale JE, Mulcahy EA, Comeau AM. Long-term follow-up to ensure quality care of individuals diagnosed with newborn screening conditions: early experience in New England. Genet Med. 2010;12(12 Suppl):S220-7.
21. Hale JE, Parad RB, Dorkin HL, et al. Cystic fibrosis newborn screening: using experience to optimize the screening algorithm. J Inherit Metab Dis. 2010;33(Suppl 2):S255-61.
22. Waisbren, S. Establishing a consortium for the Study of Rare Diseases: The Urea Cycle Disorders Consortium. Mol Genet Metab., Feb 2010; 100 (Suppl 1): S97-S105
23. White DA, Waisbren S, van Spronsen FJ. The psychology and neuropathology of phenylketonuria. Mol Genet Metab. 2010;99(Suppl 1):S1-2.
24. White DA, Waisbren S, van Spronsen FJ. Final commentary: a new chapter. Mol Genet Metab. 2010;99(Suppl 1):S106-107.
25. Waisbren S, White DA. Screening for cognitive and social-emotional problems in individuals with PKU: tools for use in the metabolic clinic. Mol Genet Metab. 2010;99(Suppl 1):S96-99.
26. Koch R, Trefz F, Waisbren S. Psychosocial issues and outcomes in maternal PKU. Mol Genet Metab. 2010;99(Suppl 1):S68-74.
27. Brumm VL, Bilder D, Waisbren SE. Psychiatric symptoms and disorders in phenylketonuria. Mol Genet Metab. 2010;99(Suppl 1):S59-63.
28. Moeschler JB, Amato RS, Brewster T, et al. Improving genetic health care: a Northern New England pilot project addressing the genetic evaluation of the child with developmental delays or intellectual disability. Am J Med Genet C Semin Med Genet. Aug 15 2009;151C(3):241-254.
29. McGrath RJ, Laflamme DJ, Schwartz AP, Stransky M, Moeschler JB. Access to genetic counseling for children with autism, Down syndrome, and intellectual disabilities. Pediatrics. Dec 2009;124(Suppl 4):S443-449.

30. Homer CJ, Cooley WC, Strickland B. Medical home 2009: what it is, where we were, and where we are today. Pediatr Ann. Sep 2009;38(9):483-490.
31. Cooley WC, McAllister JW, Sherrieb K, Kuhlthau K. Improved outcomes associated with medical home implementation in pediatric primary care. Pediatrics. Jul 2009;124(1):358-364.
32. Waisbren SE, Levy HL, Noble M, et al. Short-chain acyl-CoA dehydrogenase (SCAD) deficiency: an examination of the medical and neurodevelopmental characteristics of 14 cases identified through newborn screening or clinical symptoms. Mol Genet Metab. Sep-Oct 2008;95(1-2):39-45.
33. Waisbren SE. Expanded newborn screening: information and resources for the family physician. Am Fam Physician. Apr 1 2008;77(7):987-994.
34. Prosser LA, Ladapo JA, Rusinak D, Waisbren SE. Parental tolerance of false-positive newborn screening results. Arch Pediatr Adolesc Med. Sep 2008;162(9):870-876.
35. Hsu HW, Zytkovicz TH, Comeau AM, et al. Spectrum of medium-chain acyl-CoA dehydrogenase deficiency detected by newborn screening. Pediatrics. May 2008;121(5):e1108-1114.
36. Anastasoaie V, Kurzius L, Forbes P, Waisbren S. Stability of blood phenylalanine levels and IQ in children with phenylketonuria. Mol Genet Metab. Sep-Oct 2008;95(1-2):17-20.

Chapters

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

Other Products

Web-Based

1. * Rinehart J. and McAllister, J. How to Achieve a Shared Plan of Care for Kids with Primary Care Providers, Families and Specialists. June 4, 2015.
2. * Burke L. and Donnelly C.; Obtaining and Documenting a Pediatric Family History – Understanding Why it is Important, Identifying Red Flags, and Capturing the Information in the Electronic Medical Record. July 16, 2015.
3. * Smith W. and Smith R. Sustaining the Momentum / Incorporating Genetics in the Management of Children in the Primary Care Office. September 17, 2015.
4. * McClain MR. Lessons Learned from the New England CCHD Newborn Screening Implementation Project. Association of Public Health Laboratories CCHD Technical Assistance Webinar, August 2015.
5. * McClain MR. Lessons Learned from the New England CCHD Newborn Screening Implementation Project. National Society of Genetic Counselors, Cardiovascular Special Interest Group Webinar, August 2015.
6. GEMSS Promotional Video, Project Year 8
7. Transition Guide for Mitochondrial Disorders, Project Year 8
8. Instructional videos on QI registry changes for participating Centers
9. *An Educator's Guide to Urea Cycle Disorders*, was posted in June 2013
10. Educator's Guide to PKU (2013-2014)
11. Educator's Guide to Urea Cycle Disorders (2012-2013)
12. Educator's Guide to MCAD (2011-2012)

13. Transition Toolkit (2010-2011)
14. Understanding Galactosemia (2009-2010)
15. Galactosemia – Resources for Educators (2009-2010)
16. Moving Forward – Your Guide to Galactosemia and Primary Ovarian Insufficiency (POI) (2009-2010)
17. A Guide for Prenatal Educators (2008-2009)
18. Newborn Screening Brochures in Multiple Languages (2008-2009)

Reports

1. * Hamill, A. Promoting and Improving the Health and Well-Being of People wih Inherited Conditions. Health Care Access and Financing Group, NEGC, IOD. Apri, 2016.
2. New England Children with Genetic Disorders and Health Care Reform: Information and Recommendations for State Policymakers (May, 2014)
3. Metabolic Phase II Final Report (May 2012)
4. Metabolic Workforce Phase One Report (April 2011)
5. State Laws of New England: Use and Disclosure of Genetic and Newborn Screening Information for the Purposes of Treatment, a Registry, and Research (M. Winchester, Oct 2010)

APPENDIX D: NEGC Grant Applications

Direct Applications		
Grant Name	**Description**	**Amount**
Genomic Information and Clinical Decision Tools to Prevent Colorectal Cancer	Submitted in response to RFA-HG-13-004 – Genomic Medicine Pilot Demonstration Projects. Seeks to improve the evidence base needed to support universal Lynch syndrome screening, expand and link existing Lynch syndrome screening efforts, identify barriers and harmonize best practices.	$3,739,047 for 4 years. NOT FUNDED
HRSA; *"A Regional Approach to CCHD NBS Implementation"*	Project Yr 5. The goal of this project is to develop processes for CCHD screening that will set the stage for improved health outcomes for newborns with CCHD, and their families. This collaborative project aims to enhance and expand existing networks among state public health departments and birthing facilities, and to share resources and expertise in developing critical congenital heart disease (CCHD) newborn screening protocols, educational materials and programs, and program evaluation among five New England states: Maine (ME), New Hampshire (NH), Vermont (VT), Rhode Island (RI) and Connecticut (CT).	FUNDED $900,000 over three years.
HRSA; *"Secondary data analysis to describe long-term follow-up of disorders identifiable by newborn screening"*	Project Yr 5. Newborn screening (NBS) has been described as one of the greatest public health successes; however, little is known about the clinical histories and	Amount sought: $99,352. NOT FUNDED

	subsequent health care of those infants identified with a disorder. The primary specific aim of this project is to describe the patterns of health care, health care utilization, and total paid claims for infants and children with a disorder that is identifiable by NBS.	
NHGRI; *"Universal screening for Lynch Syndrome";*	Project Yr 5. The over-arching goal of this project is to develop methods for, and evaluate the feasibility of, incorporating an individual patient's genomic findings, into his or her clinical care via the electronic health record when available, or by other acceptable methods when an electronic health record is not available. This project will collect the outcomes data necessary to provide evidence of reduced mortality and morbidity in at-risk relatives of probands identified through Lynch syndrome screening programs	Amount sought for 4 year grant: $3,743,130; NOT FUNDED
Natural History of Disorders Identifiable by NBS	Project Yr 4. NIH. Collaborate with NYMAC to assess natural history of several targeted conditions in order to create a stronger foundation for improving care.	NOT FUNDED
Administrative Supplemental	Project Yr 3. HRSA; funds for legal analysis work and creation of the learning collaborative.	$45,000 FUNDED June 2010
Administrative Supplemental	Project Yr 2. HRSA; funds for QI data registry and electronic medical record pilot	$75,000 FUNDED April 09
Assess capacity of genetic workforce	Project Yr 2. ACMG; assess genetic workforce in light of expanded NBS; Bob McGrath will	$36,000 FUNDED April 09

	collaborate	
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 Ce nter	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

APPENDIX E. SUMMARY OF WORKGROUP MILESTONES YEAR 9 (6/1/15-5/31/16)

	June 15	*July 15*	*Aug 15*	*Sept 15*	*Oct 15*	*Nov 15*	*Dec 15*	*Jan 16*	*Feb 16*	*Mar 16*	*April 16*	*May 16*
Project Staff	Supported Family/Advocate from RI to represent NEGC @ national meeting: A Dialogue: Addressing and Paying for Genetic Services in Integrated Delivery Systems	Added pediatric improvement sites to negc website	Following up on AC recommendation, created contact list for broader dissemination of NEGC news within AAP	NEGC representatives began participating in NCC work groups Supported distance learning / workforce enhancement for metabolic clinics in the region by promoting work of GMCE to 21 recipients	Provided LOS for Next Step, Inc., for Colburn-Keenan Foundation organization grant MT States RC published adaptation of HAF Policy Brief		Facilitated connection between young adult interested in genetics and 1) genetic counselor from DHMC, 2) NERGG Outreach Program			Supported family member to attend Advocate Leadership Program @ ACMG conference	Annual meeting, 60 in attendance	Supported Family/Advocate from RI to represent NEGC @ national meeting: A Dialogue: Addressing and Paying for Genetic Services in Integrated Delivery Systems

Advisory Committee							Bi-annual meeting, discussed potential NCC RSSM workgroup recommendations				In-person meeting held in conjunction with annual;	
Collab. Council	Discussion at in-person meeting included NCC direction; preparing for next funding cycle, and sustainability						Discussion at in-person meeting included planning for annual agenda; integrating work groups, and measuring impact					
Evaluation				HRSA Perf. Measure Reporting, New collab. with LEND trainee					Release of Yr 8 Evaluation Report	HRSA Progress Reporting	Present. of Year 8 Stake. Survey Findings	
Quality Improve.				In-person meeting of QI work group; enough		At annual meeting of metabolic programs consortium		Through email and calls, reached out to new			In-person meeting at annual New work	In-person meeting of QI work group; enough

				data from DD registry to provide mid-level insights; made revisions as needed		, reintroduced registries, particularly metabolic track		partners at CHB who were interested in doing metabolic registry			group member from CHB attended Established that the group would, in fact, gather data via metabolic registry	data from DD registry to provide mid-level insights; made revisions as needed
Education and Outreach	Added CHARGE and finalized Russell Silver syndrome on GEMSS		Collaborative with Global Genes on Rare Tool Kit for Special Education launched this month	Initiated systematic review of all conditions on GEMSS, by GEMSS Acton Group with NH Leadership Series		Added Neuromuscular Disorders to GEMSS; presented new material at MD conference in NH; developed MD Parent Ambassador poster for outreach		Launched Learning Module for GEMSS	Incorporated first round of edits on GEMSS by Action Group	Presented on GEMSS at Center for START Services conference, GA	In-person meeting with core team and web designer to envision gems enhancements In-person work group meeting, included presentation by parent reviewers: GEMSS Action Group,	Following with recommendation from annual meeting, met to discuss blending goal of bringing GEMSS to primary doctors, with LEND trainee outline for next year

											and by LEND trainee on learning module	
NEGC / LEND	Bi-annual Meeting; set the course for coming year, shared general updates New work group member from VT					Met in jointly held NCC-AUCD meeting					Capstone presentation for 3 NH LEND trainees working on: 1) GEMSS; 2) NEGC program eval; 3) Health Care Access and Financing	
Health Care Access and Financing	Finalized new survey of family health care access	Pilot test of new survey	Full survey launched			Survey Closes				Annual face to face meeting, participation reflected collaboration from multiple workgroups	Research brief created by LEND trainee summarizing new survey results	

Transition		Face Forward conference for young adults, Nat. UCD Foundation Meeting, Submitted R01 App for NBS Follow Up	Collaborative Meeting on PKU	Adult Health Survey presentation, Transition Policy Presentation, AHRQ consultation	Transition policy finalized, 2nd annual Adult support group for Homocystinuria and other metab. conditions, review of RSSM models	Met. Consortium meeting, IBEMC training			Transition plan posted to Met Consort. and NEGC websites		Annual workgroup meeting, Harvard Newborn Research Seminar, PKU presentation in London	Ed brochure re: recruitment of people for IBEM, Keynote speaker National PKU, Presented to post clinic re: BabySeq project
Medical Home	Webinar: Shared Plan of Care	Webinar: Family History		Webinar: Incorp. Genetics into Primary Care							2 New Members	Webinar: Family Centered Conversations

APPENDIX F. WORKGROUP MEETINGS YEAR 9 (6/1/15-5/31/16)

	June 15	*July 15*	*Aug 15*	*Sept 15*	*Oct 15*	*Nov 15*	*Dec 15*	*Jan 16*	*Feb 16*	*Mar 16*	*April 16*	*May 16*
Management	X	X	X	X	X	X	X	X	X	X	X	X
Advisory Committee								X			X	
Collaborative Council*	X						X					
Quality Improvement				X							X	
Education & Outreach	X	X	X	X	X		X	X	X	X	X	X
NEGC/ LEND	X					X						
Health Care Access & Financing		X		X				X	X	X	X	X
Transition*											X	
Medical Home	X	X		X	X	X	X	X	X	X		

*NCC Transition work group was dissolved in Year 9. Regional transition activities were overseen by Dr. Susan Waisbren and public input was sought during a dedicated group meeting at the 2016 annual meeting of the NEGC.

APPENDIX G. COMMONLY USED ACRONYMS

- AC: Advisory Committee
- ACMG: American College of Medical Genetics
- AUCD - UCEDDs: Association of University Centers on Disabilities - University Centers for Excellence in Developmental Disabilities Education
- BAA: Business Associates Agreement
- CC: Collaborative Council
- CCHD: Critical Congenital Heart Disease
- CSHCN: Children with Special Health Care Needs
- GEMSS: Genetics Education Materials for School Success
- GPCI: Genetics in Primary Care Institute
- GVT: Global Vision Technologies
- HAF: Health Care, Access, and Financing Workgroup
- HRSA: Health Resources and Services Administration
- IOD: Institute on Disability, University of New Hampshire
- LEND: Leadership in Neurodevelopmental Disabilities
- NECMP: New England Consortium of Metabolic Programs
- NEGC: New England Regional Genetics and Newborn Screening Collaborative
- NERGG: New England Regional Genetics Group
- NCC: National Coordinating Center
- QI: Quality Improvement
- UNH: University of New Hampshire
- WG: Workgroup