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New England Genetics Collaborative Annual Report for Project Year Eight

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New England Genetics Collaborative

Annual Evaluation Report for Project Year Eight

Reflections on Project Activities 6/1/14- 5/31/15

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NEGC Project Evaluation Staff

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New England Genetics Collaborative

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"Absolute compassion is the only thing that works"
- From Dr. Solomon's Presentation to the NEGC 2015 Annual Meeting

This work is dedicated in memory to Sondra Solomon, Ph.D., a lifelong advocate and leader in the research, education and advocacy fields for improved acceptance and appreciation of the diversity that exists among all.

Contents

EXECUTIVE SUMMARY

This annual report covers the activities of the New England Genetics Collaborative (NEGC) from June 1, 2014 to May 31, 2015. 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 Eight; 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 eighth year of activity, core project staff members have focused on 1) dissemination of the ACA policy brief and development of a 2nd research stage; 2) expanding the number and quality of partnerships involved in the Quality Improvement initiative; 3) expanding the content and evaluation of the Genetics Education Materials for School Success(GEMSS) website; 4) completion of the CCHD project; 5) pursuing a range of best practices in effective transition supports; 6) launching a new medical home web series; 7) supporting alignment and integration of genetic resources among regional LEND programs; and 8) supporting multiple new training sessions, presentations and publications.

In reviewing the goals and objectives for Year Eight, 97% of 38 objectives have either been fully successful (76%) or have made satisfactory progress (21%) in accordance with the long term goals of the grant. Of the remaining objectives, 3% 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 2015 (N=22) indicated strengths in several critical areas: the NEGC is headed in the right direction (100%), participants understood how their own work fits with the NEGC (95%), and participants had a good understanding of what the NEGC will accomplish in the following year (94%), and the NEGC has had tangible outcomes that have resulted in improvements in accessibility of high quality genetic services in the region (89%). Objectives for Year Nine 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 2014 – 2015, 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 Eight.

Collaborative Activities

Project staff continued to seek out new opportunities for partnerships with both regional and national partners. During Year Eight, this included: the NEGC Annual meeting, 17 presentations, 3 publications, a promotional video for GEMSS, an instructional video for the QI group, a new transition guide for mitochondrial disorders, and six training and technical assistance activities. Also this year, the NEGC continued its work on the CCHD project, and launched a new initiative to address workforce shortages in providing specialty care for those with genetic conditions. The following outlines each of these accomplishments in more detail.

Annual Meeting, April 9-10, 2015

The annual meeting was well attended by 64 people. Plenary sessions included a focus on Genomic Sequencing in the General Newborn Population by Caroline Weipert and Meghan Towne, the Psychosocial Correlates of Genetic Disorder Related Stigma by Sondra Solomon, and "Welcome to Holland"; the Impact on Parents of a Diagnosis of CCHD by Joanna Fanos. In addition, participants had the opportunity to talk with Joan Scott from HRSA and David Flannery from the National Coordinating Center, American College of Medical Genetics, around the potential for new models of genetic services as HRSA prepares for a new funding cycle. 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 focused on reviewing the GEMSS website, a new promotional video, a discussion of do not resuscitate content on the website, a new partnership with Global Genes, and sustainability.
- Medical Home: Reviewed the new webinar series on care coordination, how to partner with other states in the region, and helping children with complex behavioral issues
- Health Care Access & Finance: Started off with a special speaker, Gabrielle Orbaek White, from Community Catalyst who led the group on a discussion of health policy and advocacy. Following this, the group outlined initial strategies and content areas for its second family survey.

- Transition: The group reviewed regional and national updates, a presentation on Adult Health Issues by Farrah Rajib from Children's Hospital Boston, and a new app on Emotional Intelligence from Elizabeth Donovan from Bodimojo.
- Quality Improvement: Seven clinical sites participated during this meeting, providing updates on their current efforts and planning out next steps for the future development of the group's activities.

Among those providing feedback on the meeting evaluation forms (N=22), 94% agreed that they had an opportunity to share and contribute (6% unsure). All respondents (N=18) thought that the NEGC was headed in the right direction. There was slightly less than full agreement on four areas: NEGC has had tangible outcomes that have resulted in improvements in accessibility of high quality genetic services in the region (89%, N=19), respondent understands how their own work fits with the NEGC (95%, N=19), and respondent has a good understanding of what the NEGC will accomplish next year (94%, N=18). 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 Eight, NEGC coalition stakeholders provided 17 presentations across a diverse mix of presenters.

- *New England Children with Genetic Disorders and Health Reform: Information and Recommendations for State Policymakers*
    - Webinar, Sept. 2014 by Meg Comeau
- *NEGC Update*
    - Consortium of Metabolic Programs Annual meeting; Boylston MA; Nov. 2014 by Karen Smith
- *"Moving on with Mito Overview"*
    - Consortium of Metabolic Programs Annual meeting; Boylston MA; Nov. 2014 by 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 by John Moeschler
- *HRSA MCHB Grants for CCHD – RC Partnership Opportunity (Dissemination of information/resources/products)"*
    - NCC/RC/PD Meeting, Washington, DC, Nov. 2014 by Monica McClain
- *QI Developmental Delay Registry: What it takes to secure a diagnosis*
    - NERGG Annual Meeting / Collaborative Session; Portsmouth, NH; Dec 4, 2014 by John Moeshler
- *The Affordable Care Act and Access to Genetic Services: Opportunities and Challenges*
    - NERGG Annual Meeting / Collaborative Session; Portsmouth, NH; Dec 4, 2014 by Meg Comeau
- *GEMSS – Genetics Education Materials for School Success: Parent Voices.*
    - AMCHP; Jan 2015 by 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 by Monica McClain
- *Returning to Therapy & the ACMG Diagnosis & Management Guidelines*
    - American College of Medical Genetics, Salt Lake City, Utah (BioMarin, Pharmaceuticals) by Susan Waisbren
- *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 by John Moeschler
- *Welcome to Holland: The Impact on Parents of a Diagnosis of CCHD*
    - NEGC Annual Meeting; Portsmouth, NH; April 2015 by Joanna Fanos
- *Living with Distinction: the Psychosocial Correlates of Genetic Disorder Related Stigma*
    - NEGC Annual Meeting; Portsmouth, NH; April 2015 by Sondra Solomon
- *Adult Health Issues*
    - NEGC Annual Meeting; Portsmouth, NH; April 2015 by Farrah Rajib
- *Envisioning the Future – How GEMSS can Help!*
    - NH Family Support Conference; May 2015 by Ann Dillon and three members of the GEMSS Action Group
- *Why Medical Home and Care Coordination are Important for Children"*
    - Webinar; May 2015 by Jill Rinehart, Jeanne McAllister
- *NEGC Genetics Workforce Project*
    - NYMAC Summit; May 28th 2015 by Monica McClain

By the end of Project Year Eight, three additional publications were created by NEGC collaborative council members. 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. Clin Pediatr, 2015 Jul;54(8):759-64..
- 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.
- 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.

In addition to the above areas, the project was responsible for the support of a GEMSS promotional video, a Transition Guide for Mitochondrial Disorders, as well as instructional videos on QI registry changes.

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 8, staff funded by the NEGC carried out six training and technical assistance activities to families, state staff, and others. Of the 133 aided in this manner, most were health providers and other professionals (111), followed by education providers (5), state MCH staff (5), community based organizations (5) and other (7). Areas of support provided included: a training to LEND trainees, an educational activity on cancer genetics management, a medical home webinar training, and collaboration with LEND trainees on projects related to GEMSS, the annual NEGC stakeholder survey, as well as the CCHD project.

Special Projects

During Year 8, the NEGC continued to engage in the Critical Congenital Heart Disease (CCHD) screening project. The goal of the ***New England CCHD Newborn Screening 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 CCHD newborn screening protocols, educational materials and programs, and program evaluation among five New England states: Maine, New Hampshire, Vermont, Rhode Island, and Connecticut. Support has been provided to clinical sites and public health newborn screening programs in a variety of ways, including financial, regular conference calls and in-person meetings, and sharing of local and national resources. This project received a no-cost extension to continue through May 2016., with 19 members serving on the project council across 5 New England States. As of April 2015, 48,294 babies were screened, and 3 cases of CCHD were identified.

Also during Year 8, NEGC initiated a new partnership with the Genetic Metabolic Center for Education (GMCE) that will come to fruition in Year 9. 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.

Collaborations with Regional and National Partners

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.,
- Transition: Susan Waisbren, Ph.D., Karen Smith
- Health Care Access and Financing: Monica McClain, Ph.D., Peter Antal, Ph.D., Meg Comeau, MHA

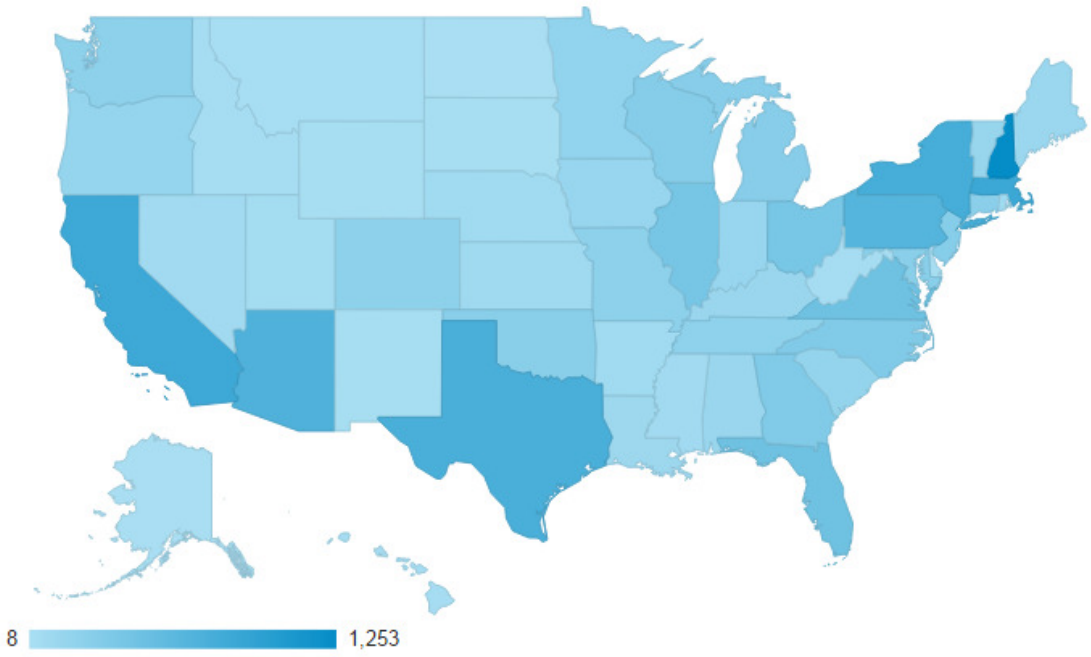
Collaboration with Other Regional and National Groups

- AAP - Genetics and Birth Defects: Leah Burke, MD
- AUCD LEND Genetics Work Group, John Moeschler, MD
- Catalyst Center, The National Center for Health Insurance Coverage and Financing For Children with Special Health Care Needs, 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
- New England Regional Genetics Group: Monica McClain, Ph.D., Karen Smith, Peter Antal, Ph.D., John Moeschler, MD, Greg Prazar MD
- New England Consortium of Metabolic Programs: Susan Waisbren, Ph.D., Leah Burke, MD
- Next Step: Susan Waisbren, Ph.D.
- NH LEND: John Moeschler, MD

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 1,103 unique users per month to 2,357). During Year Eight, an estimated[1] 19,380 individuals visited the website (an increase of 45% over the previous year). These users had over 24,000 sessions and 74,000 pageviews. Users represented all 50 states and 144 countries / territories.

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 2015, the site added 4 new conditions (Congenital Heart Defects, Rubinstein-Taybi Syndrome, Smith-Magenis Syndrome, and Sotos Syndrome) and updated many existing conditions, bringing the total count of conditions covered to 36.

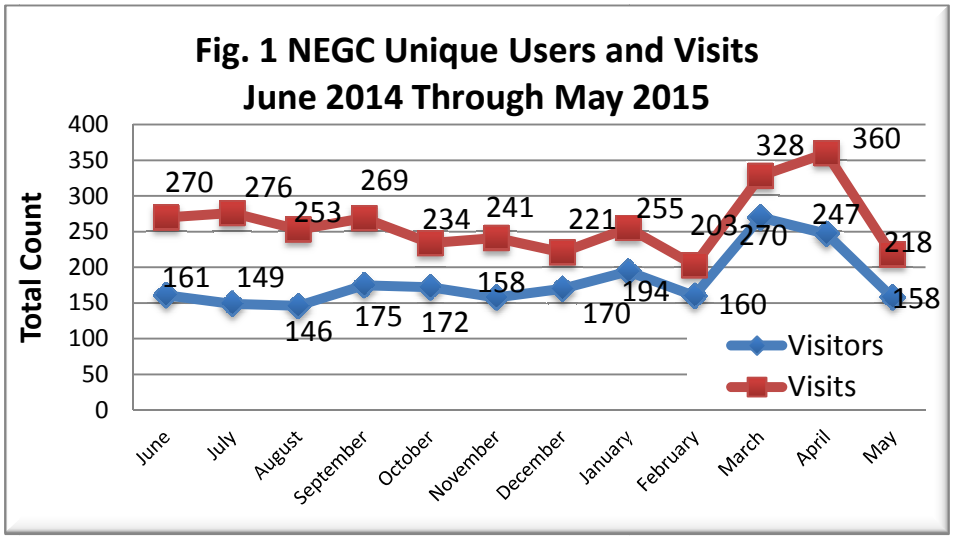
[1] Counts for FY2015 are estimated due to a loss of web traffic data during a 3 month period when the website was upgraded.

NEGC on the Web

In 2014-2015, the NEGC and GEMSS were mobile optimized, the contact list for the NEGC was improved with 19 new partners, and resources were added throughout the site.

NEGC Visitors

During Year Eight, monthly visitors to the NEGC held fairly stable from June (161) through October (172), with a slight uptick during March (270) and April (247), when the NEGC held its annual meeting) - see Fig.1.[2] Compared to the previous year, there were substantively more visitors to the NEGC (2,667 vs. 1,901) and more sessions (3,525 vs. 3,128). Over the course of these 12 months, the average time spent on the website changed throughout the year, ranging from a low of 1:27 (November) to a high of 5:50 (July) with total page views ranging from 412 (Dec) to 1,540 (March).



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 partner organization referrals, with 589 sessions generated and an average time of 5:53 (minutes:seconds) per visit, followed by Search engine results which generated more visits (1,006), but an average time of 2:12. Outside of UNH, the top five organizational drivers to the NEGC website were Genes in Life (88), the NCC (70), Maine Medical Center (55), CDC (20), and Clinica CURET (10). Of note, the GEMSS website generated 55 sessions to the NEGC. A total of 27 organizations (35 in Year 7) were identified as referral sources to the NEGC. Of note, links driven by social media (primarily from Facebook in Year 8) accounted for 38 visits (only 6 in 2007), an average time of 1:23 seconds on the website and a bounce rate of 66%. 'Other' refers primarily to web market generated referrals or from other unaffiliated sites.

[2] Note: Data for December through February are estimates based on averages for the year due to a lack of data for this time period (the site was transitioned to a new web hosting infrastructure)

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

Yr 8	Sessions	S/Mth	Adj. Sessions	Pages/S	Avg Time	% New	Bounce Rate[4]
Direct Link	609	67.7	812	3.94	3:49	61.9%	56.8%
Partner Org	589	6.2	785	5.3	5:53	41.4%	36%
Email Referral	12	0.7	16	1.8	1:21	25%	33.3%
Search	1006	87.5	1341	3	2:12	77.4%	53.7%
Social	42	0.8	56	1.6	1:15	71.4%	69%
Other	476	7.9	635	1.1	0:15	95%	83.2%

Yr 7	Sessions	S/Mth	[NA - Blank]	Pages/S	Avg Time	% New	Bounce Rate
Direct Link	697	58.1		4.21	03:28.2	60%	48.9%
Partner Org	933	3.98		5.52	06:28.1	42%	34%
Email Referral	24	0.15		3.08	00:48.4	21%	33%
Search	1,336	57.10		3.88	04:06.2	65.2%	51.1%
Social	6	0.15		1.83	00:14.3	83.3%	66.7%
Other	132	4.57		1.48	00:23.9	99%	93%

In comparing results between Years 8 and 7 using the Adjusted Sessions column, total sessions visits from direct links generated a slightly higher number of sessions and slightly improved in terms of effectiveness (more time per visit). Visitors driven by organizational partner referrals dropped by 148 sessions and had a slightly shorter visit time. The number of visits to the NEGC website that originated via organic search held stable in comparison to last year. Social driven visitors increased substantively (from 6 in the previous year to 56 in Year 8), but email referrals dropped (from 24 to 12).

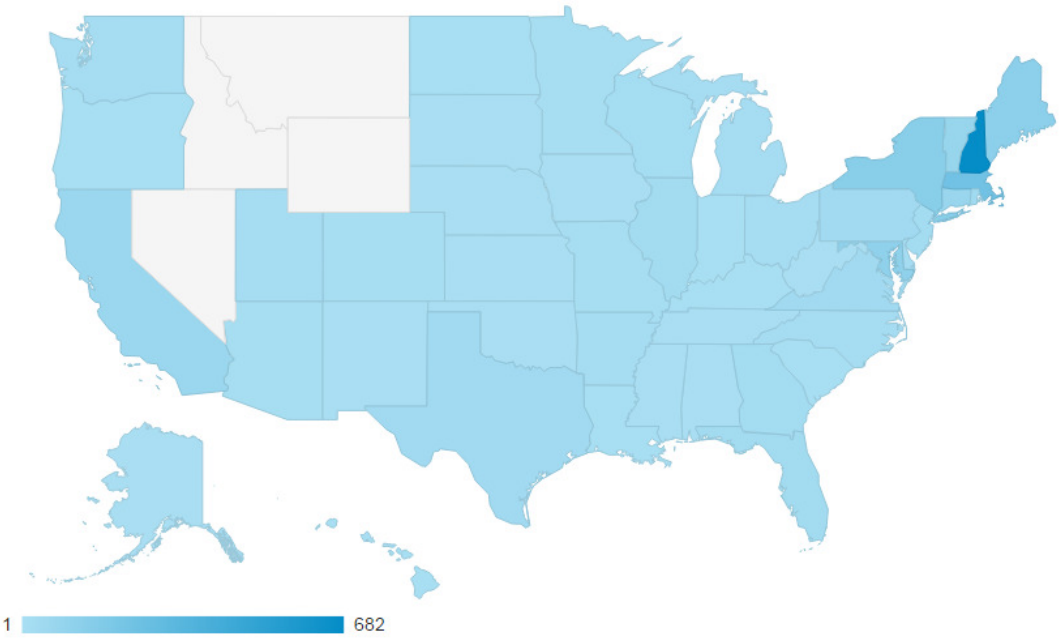
[3] Adjusted Sessions adds in an estimate based on monthly averages for the three months of data that was missing from Google Analytics reports.

[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

National Outreach of the NEGC

Of 2,730 visits to the NEGC website over 9 months, 2,123 were from the US and 367 were from other countries. Of the US visits, about one-third were from New Hampshire (682)[5]. States with 50 or more visitors include: Massachusetts (238), New York (138), Maine (124), Maryland (110), Vermont (75), Connecticut (69), and California (61). Please see Fig.2.

Figure 2: Distribution of NEGC Website Traffic by State



Resource Leveraging

During Year Eight, NEGC staff did not submit any new grant applications for core or expanded activities. However, letters of support were written for:

- ACMG's policy recommendation letter re: payment for genetic testing, to the National Government Services Medical Policy Unit; Aug 2014
- Federation for Children with Special Needs, MA, for their application for continued funding as Mass Family-to-Family Health Information Center; Dec 2014
- ACMG's application to HRSA to continue as NCC; Jan 2015
- Adelaide Murray, work study, in her application for a scholarship from the Patient Advocate Foundation; Feb 2015

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

[5] However, 432 (39%) of the NH traffic were generated from the city of Durham (where the administrative staff of the NEGC resides).

COMPLETION OF OBJECTIVES IN YEAR EIGHT (6/1/14 - 5/31/15)

This section provides an overview of each workgroup's activities during Year Eight. 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, 2015. 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 Six. Review results are defined as:

- Successful (29 of 38): 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 (8 of 38): 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 38): 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 38). 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 (1 of 38). 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 8

Objective 1. ESTABLISH AND MAINTAIN NEGC				
No.	***Objective***	***Project Status***	***Yr. 8 Definition of Success***	***Yr. 8 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 conditions.	Review: Successful All core staff activities completed during course of year.
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 workgroup participants felt satisfied with the progress their committee/workgroup made during the year. 100% of Advisory members agree that there is a spirit of collaboration, that meetings are well run and productive, that the RC provides excellent support and responds effectively, and that the Advisory Committee provides guidance and support to NEGC.
			Meetings and conference calls held	Review: Partially Successful Management meetings were held multiple times per month, 0 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 64 participated in the Year 8 NEGC Annual meeting. Among those providing feedback on the meeting evaluation forms (N=22), all agreed that the NEGC was headed in the right direction. 89% felt that the NEGC had achieved tangible outcomes that resulted in improvements in genetic services in the region, 94% felt they understood what the NEGC would accomplish in the next year, and 95% 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 94% 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 During Year 8, unique users increased primarily during the annual meeting event. Compared to the previous year, total users accessing the NEGC site had increased by 766 (2,667 vs. 1,901). Website transition to a new infrastructure for improved use by staff and consumers. Facebook page likes increased to 320 and Twitter followers increased to 171.
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. 8 Definition***	***Yr. 8 Results***

			of Success	
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	Integrate with counselor training programs in New England (Boston Univ & Brandeis)	3	Ongoing training program available to support genetics curricula within counselor training programs..	Review: Successful LEND at Children's Hospital Boston is collaborating with Boston University and Brandeis to recruit genetic counseling student in LEND program. No other LEND programs are currently pursuing this area.
2.4	Coordinate NE LEND trainees' requirements in skill development for CE/TA with NEGC	3	Shared understanding of TA/CE requirements and NEGC needs Implement measures of quality and success of TA/CE	Review: Partially Successful Three Leadership in Action activities were identified and pursued between the NH/Maine LEND program and the NEGC. 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. 8 Definition of Success***	***Yr. 8 Results***
3.1	Develop resources for additional conditions	3	Additional conditions content developed for GEMSS	Review: Successful. Four new conditions added during Year 8.

3.2	Develop a tool for families to access educational transition programs	1	Report of findings Feedback received; checklist finalized	Review: Successful Information for GEMSS page on transition reviewed and updated
3.3	Collaborate with Global Genes to develop "Rare Toolkit" for education	3	Resource page for parents developed.	Review: Partially Successful During Year 8, GEMSS and Global Genes collaborated on developing a new resource page. (completed in July of 2015 (https://globalgenes.org/toolkits/educationadvocacy/intro/).
3.4	Promote and disseminate GEMSS	3	Parent Ambassadors recruited, Presentations made, Training seminars given	Review: Successful 4 Parent Ambassadors participated during Year 8. Presentation made to AMCHP in 1/15. New promotional video developed with LEND trainee. Action group established within NH Leadership and presentation made to NH Family Support conference in May 2015. Utilized GEMSS magnets and parent ambassador posters for dissemination materials.

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. 8 Definition of Success***	***Yr. 8 Results***
4.1	Develop strategy to implement genetics in primary care	1	Develop a series of webinars to facilitate the integration of genetics into primary care pediatrics.	Review: Successful Critical partners were brought together and developed a plan for a series of 4 webinars on the integration of genetics in primary care. The first of four webinars was held in May 2015, with 60 online participants. Jeanne McAllister and Dr. Jill Reinhardt presented. Most respondents (N=18) rated the presenters as either good or excellent. In terms of the usefulness and value of the webinar, 6% felt it was average, 67% good, and 28% excellent. Fifteen of 18 felt that the

				educational session will enhance their competence, influence their practice, and impact their patient outcomes.
Objective 5. Transition: *Continuously improve and expand the workforce needed to care for those children with disorders identified by NBS programs*				
5.1	Develop strategy to establish why primary care doctors should take cases of young adults with genetic conditions	3	Develop, distribute, and analyze Adult Health Survey.	Review: Successful Adult survey administered at 3 condition-specific family conferences (summer of 2014), and continued to disseminate during Year 8.
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	Review: Successful Continued development of clinical policy to support transition activities at Children's Hospital Boston. Includes utilization of the ABAS screening tool (which was previously validated with NEGC support). Collaborating with the IBECM (Inborn Errors of Metabolism). Continued efforts to seek research funding to support this area.
5.3	Develop transitions brochure on mitochondrial diseases.	3	Brochures to be developed outlining important information for school personnel to understand in regards to supporting students with mitochondrial disease.	Review: Successful The Moving on with Mito educational resource was completed.
5.2	Leadership Training	1	Youth complete the program and report high satisfaction Youth are engaged to become effective	Review: Successful Face Forward Camp: 19 youth participated in the summer Face Forward event. Participants reported: an improvement in having the tools and skills needed

			advocates.	to be successful in school while managing their condition (53% pre to 76% post); a doubling of participants planning to see an adult care physician; and an increased perception that they are part of a community that understand their condition (63% increase). Support for a young adult to initiate a support group for adults with metabolic conditions. Homocystinuria group established to provide connections and support to participants.

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. 8 Definition of Success***	***Yr. 8 Results***
6.1	Creation of a new policy brief and webinar	1	Key decision makers are better informed about the coverage, costs, and benefits of quality services for those with genetic conditions	Review: Successful New policy brief released and webinar held in September 2014 highlighting major findings from the HAF group's research. Webinar included recommendations on approaches to potentially address gaps in state service systems.
6.3	Collaborate with regional Family Voices partner organizations to develop and disseminate new research to families, identify data points from the research speaking to state level	1	Families are better prepared to self-advocate for needed benefits to decision makers	Review: Successful The policy brief was widely disseminated to contacts from the NEGC, the Institute on Disability at UNH, as well as our partners in the region. Positive feedback was received from those participating

	issues			in the webinar, as well as those receiving copies of the report, with multiple respondents on the stakeholder survey and annual meeting evaluations reflecting on the positive impact of this work.
6.4	Development of survey questionnaire for 2nd research stage with a focus on assessing network adequacy	3	New survey developed with input from core stakeholders.	Review: Partially Successful During the course of the year, members of the HAF group developed a range of new topics to pursue and further clarified potential questions for the second survey round during the 2015 annual meeting of the NEGC.
6.5	Development of agreements with partner organizations in each state to identify potential participants to participate in the new survey	3	Research pool created	Review: Successful Although formal written agreements were not put in place, the contact list for dissemination of the survey was expanded greatly during the year.
6.6	Design and implement research strategy	3	Data collected	Review: Partially Successful Initial discussions were held on how best to collect the 2nd round of data. These discussions continued into Year 9 and were finalized during the Summer of 2015.

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. 8 Definition of Success***	***Yr. 8 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 (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 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 Year 8. The registry has records for 1,669 individuals.
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 Webex utilized for online conferences. QI Registry vendor (GVT) has asynchronous orientation videos completed. Registry workflow video completed. Moeschler has 2 videos (using Camtasia Relay) on the principles of quality improvement and value creation in Medical Genetics completed.
7.6	Test decision-support mobile app for	3	Decision support app reviewed and made	

	intellectual disability, DD QI.		available to workgroup members	Review: Successful All DDID QI sites are aware of the www.Treatable-ID.org decision support tool, and Dartmouth-Hitchcock Medical Center uses this decision support regularly.
7.7	Meet requirements of American Board of Medical Genetics Maintenance Of Certification module		Regular information on family centered care in coordinated systems is integrated into the QI process	Review: Successful ABMG has approved the NEGC QI for credit for Maintenance of Certification, Part IV. W. Smith successfully submitted this activity to ABMG. Other centers' physicians plan to do same.
7.8	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.
7.9	Expand QI process to additional centers.	3	Additional centers are added to the learning collaborative.	Review: Partially Successful Recently added Baystate Medical Center, Springfield, MA, to the QI group for those referred with developmental delays and intellectual disability bringing the number of sites to five. Other sites have expressed an interest. As participation at the regional level strengthens, the NEGC plans to invite centers from other regions to participate in the collaborative.
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. 8 Definition of Success*	*Yr. 8 Results*
8.1	Pilot project: Critical congenital heart disease (CCHD) screening	3	CC meeting held (1 per year); educational materials developed and distributed; screening and follow-up and data collections protocols for each facility documented;	Review: Successful Pilot project implemented for: Critical congenital heart disease (CCHD) newborn screening. The project has enrolled 9 birthing centers, developed a uniform screening protocol and disseminated a Tier 2 educational brochure for parents/families. As of April 2015, 48,294 babies were screened and 3 cases of CCHD were identified.
Objective 9. QUANTITATIVE AND QUALITATIVE EVALUATIONS				
No.	*Objective*	*Project Status*	*Yr. 8 Definition of Success*	*Yr. 8 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 20% of stakeholders invited to participate in the survey did so.
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	Review: Partially Successful While information included in the Year 8 report was provided to management staff throughout the year, the report was not finalized

			progress.	until January of 2015.
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 9 (6/1/15-5/31/16)

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

Table 4: Year 9 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
Collaborate with Genetic Counselor training programs in NE (Boston U., Brandeis U.)	Articulation of options between LEND programs and GC programs to co-recruit a GC student	Written summary of co-recruitment and training plans
Coordinate NE LEND trainees' requirements in skill development for CE/TA with NEGC	Shared understanding of TA/CE requirements and NEGC needs Implement measures of quality and success of TA/CE	Number of CE/TA projects engaged in with LEND trainees. Satisfaction measures on quality of CE/TA.
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*		
Update GEMSS	Conditions and supporting material updated	Review by project evaluator.
Develop resources for additional conditions	Additional conditions content developed for GEMSS	Conditions added to GEMSS website
Promote and disseminate GEMSS via Parent Ambassadors and NH	Parent Ambassadors trained and	# Parent Ambassadors trained

Leadership	enrolled. Families / Organizations receive information on GEMSS	# of families / organizations outreached to by Parent Ambassadors and GEMSS Action group.
Medical Home/Care Coordination: *Continuously improve and expand the workforce needed to care for those children with disorders identified by NBS programs*		
Implement genetics in primary care webinars	Webinars implemented with high satisfaction ratings.	# 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	Development and implementation of health survey incorporating modified Erikson Psychosocial Stage Inventory	# of surveys distributed, analysis completed, report completed
Complete transitions brochure on mitochondrial diseases		
Face Forward Camp	Youth complete the program and report high satisfaction Youth are engaged to become effective advocates.	# of youth participating / completing the Face Forward program, # youth reporting satisfaction with the program
Develop strategy to obtain data on transition outcomes	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
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*		
Design and implement research strategy for 2nd research stage	Data collected	# of responses collected; Data set created and analyzed
Creation of a policy brief and web seminar highlighting findings from the 2nd research stage.	Key decision makers are better informed about the coverage, costs, and benefits of quality services for those with genetic conditions	# of representatives from key target groups participating in presentations and/or reviewing project documents (legislators, insurance groups, professional assoc), # of unique web-based viewers
Collaborate with regional Family Voices partner organizations to develop and disseminate material	Families are better prepared to self-advocate for needed benefits to decision makers	Family feedback, stakeholder survey
Quality Improvement – DD/ID 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 centers.	Additional centers are added to the learning collaborative.	# of regional and national centers added
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*		
Pilot project: Critical congenital heart disease (CCHD) screening	Completion of evaluation reports, development of resource page for children and families living with CCHD.	Completion of evaluation report and NEGC resource page.

CONCLUSIONS AND RECOMMENDATIONS

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

- Multiple discussion forums highlighting the implications of the HAF group's policy brief on the Affordable Care Act and outlining core components for a second research stage
- Continued growth and utilization of the NEGC and GEMSS websites, including a new collaboration with Global Genes to develop a rare toolkit for education
- Launching of a new medical home webinar series as well as a new effort to understand health concerns among adult patients living with genetic conditions
- 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 1,669 records entered.
- A highly successful and informative annual meeting, where 89% of respondents felt that the NEGC had achieved tangible outcomes that resulted in improvements in genetic services in the region
- Continuation of the CCHD project to create an electronic resource with psychosocial supports for families and provide resource bags to parents at the time of diagnosis.
- A new initiative to create and support a telehealth platform for New England hospitals and clinics that serve metabolic patients,
- Substantive efforts were made to provide presentations and trainings this past year along with publishing research in peer reviewed journal articles (17 presentations, 5 publications, 6 CE/TA activities)

In looking forward to future activities of the NEGC, there are a few areas 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).
- *Improve access and utilization of evaluation reports* to help stakeholder have a more informed understanding of the work of the NEGC, its challenges and opportunities. While management staff were kept apprised of needed adjustments to the NEGC throughout the year, completion of the Year 8 report took longer than what was hoped for during the year. The goal for Year 9 activities will be to have an updated report available no later than 9/30/16.
- *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).

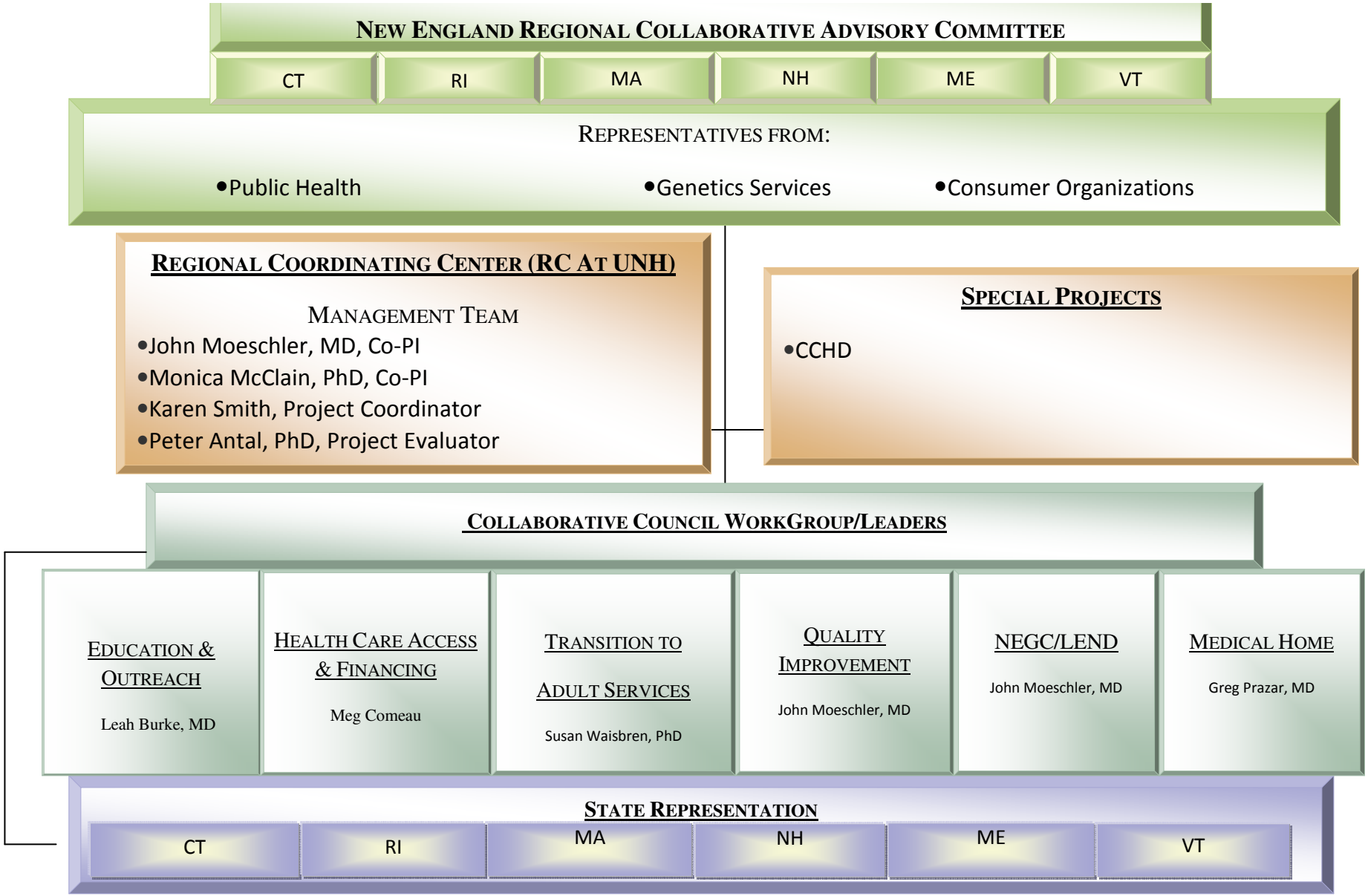
- *Address Sustainability Challenges for Core NEGC Activities.* As the NEGC is nearing completion of its current five year cycle, it will be critical that clear strategies for sustainability be outlined and implemented in collaboration with NEGC stakeholders. In the event that future cycles do not fund 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 Eight**

Sharing Work on Project Activities

* *"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

* *"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 Eight**

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. Clin Pediatr, in press.
2. * 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.
3. * 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.
4. 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.
5. 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.
6. 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.
7. 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.
8. 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.
9. Fanos, J. New "first families": the psychological impact of new genetic technologies. Genetics in Medicine, 2012 Feb;14(2):189-90.
10. 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
11. 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.
12. 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.
13. 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.*

14. 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.
15. 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
16. 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.
17. 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.
18. 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.
19. 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.
20. 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
21. White DA, Waisbren S, van Spronsen FJ. The psychology and neuropathology of phenylketonuria. Mol Genet Metab. 2010;99(Suppl 1):S1-2.
22. White DA, Waisbren S, van Spronsen FJ. Final commentary: a new chapter. Mol Genet Metab. 2010;99(Suppl 1):S106-107.
23. 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.
24. Koch R, Trefz F, Waisbren S. Psychosocial issues and outcomes in maternal PKU. Mol Genet Metab. 2010;99(Suppl 1):S68-74.
25. Brumm VL, Bilder D, Waisbren SE. Psychiatric symptoms and disorders in phenylketonuria. Mol Genet Metab. 2010;99(Suppl 1):S59-63.
26. 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.
27. 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.
28. 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.
29. 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.

30. 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.
31. Waisbren SE. Expanded newborn screening: information and resources for the family physician. Am Fam Physician. Apr 1 2008;77(7):987-994.
32. 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.
33. 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.
34. 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. * GEMSS Promotional Video, Project Year 8
2. * Transition Guide for Mitochondrial Disorders, Project Year 8
3. Instructional videos on QI registry changes for participating Centers
4. *An Educator's Guide to Urea Cycle Disorders*, was posted in June 2013
5. Educator's Guide to PKU (2013-2014)
6. Educator's Guide to Urea Cycle Disorders (2012-2013)
7. Educator's Guide to MCAD (2011-2012)
8. Transition Toolkit (2010-2011)
9. Understanding Galactosemia (2009-2010)
10. Galactosemia – Resources for Educators (2009-2010)
11. Moving Forward – Your Guide to Galactosemia and Primary Ovarian Insufficiency (POI) (2009-2010)
12. A Guide for Prenatal Educators (2008-2009)
13. Newborn Screening Brochures in Multiple Languages (2008-2009)

Reports

1. New England Children with Genetic Disorders and Health Care Reform: Information and Recommendations for State Policymakers (May, 2014)
2. Metabolic Phase II Final Report (May 2012)
3. Metabolic Workforce Phase One Report (April 2011)
4. 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 Center	Project Yr 2. Submitted by John Moeschler to supplement QI project	NOT FUNDED
Galactosemia and Premature Ovarian Insufficiency	Project Yr 2. AUCD; collaboration with Susan Waisbren; submitted Oct 08	NOT FUNDED

APPENDIX E. SUMMARY OF WORKGROUP MILESTONES YEAR 8 (6/1/14-5/31/15)

	June 14	*July 14*	*Aug 14*	*Sept 14*	*Oct 14*	*Nov 14*	*Dec 14*	*Jan 15*	*Feb 15*	*Mar 15*	*April 15*	*May 15*
Project Staff						Present. on QI and CCHD activities at NCC annual (Moeschler / McClain) Support. activity at Jackson Lab: Cancer Genetics Management in the Primary Care Setting Helped support key author of GEMSS to attend AUCD to enhance knowledge	Provided LOS for Mass Family Voices McClain participate in NERGG PH Genomics Task Force Sponsored Collaborative Session with NERGG: Affordable Care Act (Comeau), QI DD registry (Moschler)	One of 4 RCs in panel present. on family engagement at AMCHP Part. in NEGCN-led call on Emergency Prep LOS for ACMG's application to HRSA		Supported attendee at Advocate Leader Program, ACMG Provided info. and materials to Heartland Region to adapt policy brief on health reform McClain presented at ACMG: Public Health Approach. for Implementing or Supporting Cascade Screening	Annual meeting Added roughly 20 to NEGC contact list following due to engage. around annual meeting Final report by Dr. Joanna Fanos: "Developmental Impact of Growing up with a Sibling with CCHD", and Sibling Interview Guide	McClain presented at NYMAC Summit on Genetic Workforce Project

						base				for Tier 1 Genetic Conditions		
Advisory Committee							Bi-annual meeting, advised of changes in national context; discussed medical home group and annual meeting				In-person meeting held in conjunc. with annual; provided feedback to HRSA, NCC re: genetic service needs	
Collab. Council												Set the agenda for in-person meeting next month
Evaluation				HRSA Perf. Measure Reporting, New collab. with LEND trainee				Release of Yr 7 Evaluation Report		HRSA Progress Reporting	Release of Stake. Survey Report	

Quality Improve.			Moeschler pub. released: Comprehensive Evaluation of the Child with Intellectual Disability or Global Developmental Delays	In-person meeting of QI work group; enough data from DD registry to provide early insights; made revisions as needed				Facilitated using RedCap data platform at CHB for QI registry			In-person meeting at annual	
Education and Outreach	Identified this year as "Year of the Family"			NH LEND trainee identified to work on 1) autism, 2) communi-cation content	Launched GEMSS in mobile platform Parent Ambassador "Outreach Day" netted highest # of visitors to GEMSS Facebook this year			Developed Parent Ambassador posters, new method to facilitate dissemination	Began collaborating with Global Genes, CA based advocacy org, to create "Rare Toolkit" on special education		In-person meeting included present.by parent ambassador and GEMSS Action Group; and premier viewing of promo video developed by LEND trainee	GEMSS Action Group presented on their year-long leadership activity, at NH Family Support Conference GEMSS was disseminated in national webinar by START (project at UNH): Genetic

												Disorders and Associated Behavioral Phenotypes
NEGC / LEND	Bi-annual Meeting via video-conference; set the course for coming year, shared curriculum ideas and resources					Met in jointly held NCC-AUCD meeting					Capstone presentation for 3 NH LEND trainees working on: 1) GEMSS; 2) NEGC program eval; 3) CCHD data analysis	
Health Care Access and Financing	Finalized policy brief: New England Children with Genetic Disorders and Health Reform: Information and Recommendations for State Policymake	Created contact list of roughly 1600 policy makers and those who might influence them, in NE	Work group members dissem. completed policy brief to contact list; mailed or hand-delivered 250 copies	Webinar by Meg Comeau explaining the policy brief (roughly 50 attendees)	1st draft of survey was sent to group for discussion	Continued dissemination of policy brief (LEND directors)					In-person work group meeting included Advocacy training which was attended by young adults from Transition work group. Developm	

	rs										ent of core interest areas for Family Survey Phase 2	
Transition		Face Forward conference for young adults				Finalized "Moving on with Mito" educational brochure; presented at Met Consortium mtg Waisbren presented Adult Health Survey at Consortium; Smith provide NEGC update		Facilitated consumer-led support group for people with metabolic conditions in Boston Created family-friendly 1 page fact sheet for Mito, added to Metabolic Basics on Consortium website and NEGC		Waisbren led NCC Transition meeting, and NEGC supported 2 additional members to attend	In-person work group meeting Provided 100 hard copies of "Moving on with Mito" to support networks including Mito Action	
Medical Home	Kick-off in-person meeting of re-envisioned Med Home		Dr. Greg Prazar became official work group chair;		Added representation from additional NE state (Dr. Alex Geertsma,	Facilitated nomination to NCC for exemplary care coordinati		Family Voices.			In-person meeting, after which 4 additional members joined,	Launched webinar series on care coordination: It's All About

	work group		members added from 4 of 6 NE states		CT)	on (Dr. Jill Rinehart, VT)					including parent representative from Rhode Island Family Voices	Teamwork: Incorporating Genetics and Family History into the work of the Patient Centered Medical Home" Article on Webinar Series in NH AAP newsletter

APPENDIX F. WORKGROUP MEETINGS YEAR 8 (6/1/14-5/31/15)

	June 14	*July 14*	*Aug 14*	*Sept 14*	*Oct 14*	*Nov 14*	*Dec 14*	*Jan 15*	*Feb 15*	*Mar 15*	*April 15*	*May 15*
Management	X	X	X	X	X	X	X	X	X	X	X	X
Advisory Committee							X				X	
Collaborative Council*												
Quality Improvement				X							X	
Education & Outreach	X	X	X			X		X		X	X	X
NEGC/ LEND	X					X						
Health Care Access & Financing		X	X	X	X	X		X	X	X	X	
Transition**	X	X		X	X	X/X	X	X	X	X	X/X	X
Medical Home	X	X			X	X	X	X	X	X	X	X

* The Collaborative Council met in June of 2015.

**NCC Transition work group has monthly calls/NEGC wg met face to face at Met Consortium in Nov at NEGC annual in April

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