

# New England Genetics Collaborative

## Annual Meeting, December 2008

### Meeting Summary

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### Introduction

This report provides a summary of the New England Genetics Collaborative (NEGC) annual meeting held December 4, 2008 in Portsmouth, New Hampshire. The summary includes highlights of new research supported by the collaborative (pg.2), an overview of workgroup activities (pg. 2), results of the small group (pg. 4) and large group (pg. 5) discussions, an update from the National Coordinating Council (pg.6) as well as our federal representative (pg. 6), feedback provided on the evaluation form (pg.7), and conclusions / recommended next steps for the NEGC (pg. 11).

Thirty-six individuals participated in the meeting, including representatives from physicians, state planners, and family members of consumers. Project staff highlighted the key accomplishments of the project over the course of the past year including newly sponsored research, multiple conferences aiding in knowledge development and training by partners, improvement in testing of quality controls for labs, and toolkits developed for consumers. Major activities for next year were identified as well, including: greater collaboration between partners, better outreach to consumers and other service providers, and refining the innovative projects process. Lastly, staff facilitated discussions among partners on a range of issues shaping the future of the project including: consumer outreach, communication, innovative projects, and development of state teams.

While most participants agreed that the project was headed in the right direction, several individuals were uncertain about their understanding of project activities and accomplishments as well as how their own activities fit in with the objectives of the NEGC. Although the project staff did seek to address some of these areas during the day's proceedings, it may be helpful to find additional methods to ensure that all members of the collaborative have a clear understanding of the current status and future of the collaborative. This speaks to a broader challenge for the project which will be to determine how best to continue building on the project's information infrastructure and to shape it in such a way as to: keep all stakeholders clearly informed about their roles; disseminate information about the mission, activities, and achievements of the NEGC; and regularly bring in and appropriately process new information coming in from new partners, state teams, and national initiatives.

## Overview of the Day

Following general introductions for the day, recent research undertaken by UNH staff was presented using the 2005/2006 Children With Special Health Care Needs data set. Afterwards, chairs of each workgroup provided an overview of their group's activities during the past year as well as their next steps for 2009. This was followed by small group discussions on issues of innovative projects, consumer participation, and enhancing communication, a large group discussion on the development of state teams, an update from the National Coordinating Center, and an update from our federal partner, Jill Shuger.

Thirty-six people participated, including a mix of physicians, specialty care providers, state coordinators, family members of consumers, and other service providers. Attendees of the 4.5 hour meeting included:

Peter Antal, Judith Benkendorf, Tom Brewster, Bev Burke, Leah Burke, Kathy Higgins Cahill, Anne Comeau, Carl Cooley, Lou DiNicola, Roger Eaton, Janet Farrell, David Helm, Cindy Ingham, Audrey Knight, Fay Larson, Marcia Lavochkin, Barbara Lerner, Martha-Jean Madison, Stephanie Miller, John Moeschler, Ellie Mulcahy, Debra Nelson, Marinell Newton, Denise Queally, Heidi Rehm, Valerie Ricker, Vine Samuels, Amy Schwartz, Judith Shaw, Vivian Shih, Karen Smith, Rosemarie Smith, Wendy Smith, Chris Stille, Paul Vetter, Susan Waisbren.

### New Research on Children with Special Health Care Needs

John Moeschler presented new research by Bob McGrath looking at access to genetic counseling services for children with Down's Syndrome, Autism, and Mental Retardation. Dr. McGrath found that the presence of a medical home, with the support of insurance, were the most important factors (out of the variables studied) in facilitating access to Genetic Counseling Services.

### Workgroup Summaries

The following are the highlights presented from each of the workgroup chairs concerning their activities in 2008 as well as group plans for 2009. Items below include additional information included in the 2008/2009 grant application not covered at the meeting.

#### Medical Home (Chair: Carl Cooley)

- Activities in 2007-2008:
  - Work group kick off meeting in June, 2008; Working with Transition Work Group;
  - Addressed Metabolic Consortium and NERGG/NEGC annual meeting
- Focus in 2009:
  - Develop pilot for implementation in September, 2009 (pilot will focus first on specialty care, then lead back to primary care physicians)

*Discussion Point: Tools for connecting to a medical home may need to start from the specialty care clinics rather than primary care physicians.*

Transition (Chair: Susan Waisbren)

- Activities in 2007-2008:
  - PKU Toolkit; Adult Conference for PKU; monthly conference calls; collaboration with Medical Home Work Group; NE Consortium meeting; survey development; coordination with other HRSA projects
- Focus in 2009:
  - Publish report on adult conference; conduct survey & publish results; focus on premature ovarian insufficiency; hold adult conference on galactosemia; track adults with PKU in NE; establish adult division of New England Connection for PKU and Allied Disorders (NECPAD); continue collaboration with Medical Home Work Group, continued work with HRSA sponsored projects

*Discussion Point: Dr. Waisbren noted that there is a focus on individuals with metabolic conditions but not necessarily those having physical disabilities; also identified the challenge for adults seeking care when services were primarily still offered through pediatric centers.*

Dissemination, Education and Marketing (Chair: Leah Burke)

- Activities in 2007-2008:
  - Newborn screening toolkit; Family Health History Awareness project; Community Health Centers Genomics Outreach project; and NE Resource Directory
- Focus in 2009:
  - Design a “Children with Genetic/Metabolic Conditions in Educational Settings” chart; create a needs assessment system for special educators using an online tool for common conditions; evaluate the utilization and impact of the genetics outreach project for community health centers

*Discussion Point: Can the online tool developed also be made to apply for students at the college level?*

Quality Improvement (Chair: John Moeschler)

- Activities in 2007-2008:
  - Submitted two invited papers: “Barriers to Genetics Services” to *Pediatrics* – McGrath et al. as well as paper submitted to AJMG special issue on quality improvement; brought on Patrick Miller, MPH, for data and registry development work; developing clinical genetics registry for 4 New England practices
- Focus in 2009:
  - Obtain IRB approvals and implementation for site-by-site descriptive evaluation of clinical genetics practices using claims data; improve level of metabolic disease awareness at medical centers through the Metabolic Outreach Service; develop mechanisms for long-distance consultation regarding diagnosis and management of metabolic patients; provide educational opportunities in metabolism for medical students and residents and encourage formal training in biochemical genetics; partner with federally qualified Health Centers to reach the vulnerable, underserved, and hard to reach populations

Quality Assurance (Chair: Roger Eaton)

- Activities in 2007-2008:

- New England Newborn Screening Program (NENSP) collaborated with other state labs based in New York, Connecticut and Wisconsin; initiated project to obtain objective evidence about the applicability of NENSP algorithms to other newborn screening labs in our region and across the country; began analyzing raw data from partner labs on babies diagnosed with disorders associated with propionylcarnitine (C3) elevations.
- Focus in 2009:
  - Conduct data analysis using additional MSMS markers; convene collaborative conference May 2009

#### Long-Term Follow-Up (Chair: Anne Comeau)

- Activities in 2007-2008:
  - Enhancing core data base and developing data-sharing agreements with participating states (Data base will be used to track outcomes and identify best practices and lead to newborn screening quality improvements that are *sustainable*); developing LTFU variables with the CF, Hemoglobin, and Metabolic Workgroups
- Focus in 2009:
  - Develop qualitative study to assess impact of genetic disorders on siblings; developing psychosocial measures for inclusion in outcomes tracking; participate in interregional and NCC workgroups, convene Regional Policy and HSR conference, complete regional charter agreement.

#### *Summary Discussion Point*

*A concern was raised about the different levels (local, state, regional, national) of activities underway and the extent to which there may be some duplication of effort (e.g. multiple attempts to collect data for follow-up, a range of outreach and education efforts, etc.). It was noted that it was precisely because of these multiple efforts from different levels of service providers that the NEGC is so critical for the region. The NEGC will continue to seek out ways to help the various partners effectively collaborate and to stay abreast of major changes at the state, regional and national level.*

#### Small Group Discussions

Following the workgroup summaries, the participants self-selected to participate in one of three mini-discussion groups: innovative project programs, enhancing consumer participation, and communicating with constituents. Each was provided with an outline of the issues at hand for each topic and asked to report out to the full group.

#### Innovative Project Program

The group discussed issues around the timeline of the awards, the range of types of funded projects, length of funding, and sustainability issues. The group recommended that:

- The timeline for the process be moved up so that staff will be done with reviewing proposals at the point they expect to receive the Notice of Grant Award from HRSA (June 1);
- the NEGC provide a maximum of 2 years of funding;
- requestors should identify additional funding sources;

- requestors should have a sustainability plan in place;
- carry over money should be included as part of a subsequent budget unless specifically stipulated.

### Enhancing Consumer Participation

Participants focused on figuring out new ways that the NEGC could have meaningful consumer participation in its activities. The group recommended that:

- The NEGC create a consumer advisory pool with specific tasks and goals, one of which is to help evaluate which work groups do in fact need to have a consumer voice and which do not. The group should include affected people and adults, not just parents of affected children. Consumers should be mentored so that they can more fully participate.
- The NEGC should contact other regions about how they work with consumers

### Communication with Constituents

Discussion members primarily focused on the quality of communications with current stakeholders and whether it was time to make a substantial shift into consumer outreach. The group recommended that:

- While communications with current stakeholders (recognized primarily as service providers and state public health officials) was seen to be effective, the group noted that it was time to add substantial communication efforts to consumer groups (possibly through Kristi Zonno) as well as better outreach to providers not involved with the NEGC (e.g. education of barriers to services, needs of populations served).
- As a part of this process, the NEGC should continue to focus on building a stronger information infrastructure that enables access to core sources of information. Updating the website to reflect multiple participant interests was seen as one key strategy.
- In undertaking these efforts, attention should be paid to having a clear message for each group about what they should know about the NEGC, our mission, and/or what it is they should be able to do with NEGC resources.

### Large Group Discussion: Developing State Teams

During the lunch session, all participants talked about how best to integrate the activities of the NEGC with the needs of individual states. Members noted that there should be an assessment of what groups already exist in each state to address state issues, and that key decision makers in each state as well as consumers (including a range of ages) should be involved at some level. Members also noted a significant barrier in that the people most likely to participate in the teams have already been tapped repeatedly. Recommended next steps fell under themes of group dynamics, what the groups would do, potential NEGC roles, and how to implement the groups:

- Group Dynamics
  - More likely to have participation if there is a unique brainstorming session – “reality check” – (once a year?) rather than asking people to be in a new ongoing group
  - Meetings should be well-planned and agenda specific (avoid using more of people’s time than is necessary)
  - Identify common issues with other groups and use existing groups – don’t duplicate
  - Priorities should be clear
- Group To Dos
  - Ask the state advisory groups for their yearly work plan and NEGC review to see what is in line with NEGC priorities / goals.
  - State Teams of NEGC would identify something relevant to that state that NEGC can impact – not necessarily a “state plan” or even “the” issue of the state but rather an issue that the NEGC can address for each state.
- NEGC’s Role in Supporting State Groups
  - Possibly provide incentive to set up a group, i.e. grant
  - Help organizations write grants and/or accept money on their behalf in order to distribute funds
- Implementing
  - Implement a survey (Survey Monkey) to determine who will serve as the points of contact (specific to agencies)
  - Look at examples of similar state informants from other regions
  - Approach HRSA for money by incorporating state teams (if appropriate) under one of the work groups

National Update by Judith Benkendorf, Project Manager for the National Coordinating Center (NCC)

Judith provided a summary to participants of the NCC structure and their relationship to the regional collaborative centers and an overview of national working groups. Key projects for 2009 of the NCC include:

- ACT sheets for expanded NBS
- Patient based data collection activities
- Newborn Screening Translational Research Network
- Quality improvement: integrating with RCCs
- Integrating LTFU data collection and the NBSTRN

Judith noted that the NCC will continue to work closely with HRSA and RCCs to bring forward new priorities and regional projects appropriate for expansion to national efforts.

Federal Update by Jill Shuger, Genetic Services Branch, HRSA/MCHB

Via phone conference, Jill updated participants about major initiatives underway at the federal level which are likely to have an impact on our regional efforts for improving access and utilization of genetic services. Topics addressed included:

- An overview of the Newborn Screening Saves Lives Act which adds new activities to the Children's Health Care Act of 2000 for Screening for Heritable Disorders. Primary efforts will be focused on assisting states in improving and expanding NBS programs, federal guidelines on screening for newborns, improving education, outreach and follow up care, and improving laboratory quality and surveillance.
- HRSA Grant Program: Eligible applicants will adopt and implement guidelines and recommendations from the Advisory Committee on Heritable Disorders in Newborns and Children.
  - The role of the advisory committee will be to provide recommendations on diagnostic / screening activities, develop a model for newborn screening expansion, and consider ways to ensure that all states have the capacity to screen for the recommended conditions.
- An internet-based Information Clearinghouse will increase awareness of NBS across a range of stakeholders, provide information on diseases and services to individuals and families, and provide data on performance measures for NBS.
- An Interagency Coordinating Committee on Newborn and Child Screening will collect, analyze, and make data available on heritable disorders, conduct research on effective interventions, promote prevention of poor outcomes, and provide education and information to the public.
- Laboratory Quality and Surveillance will ensure quality of laboratories involved in newborn screening activities
- A National Contingency Plan for NBS (including the collection, shipment, processing, and reporting out for screening as well as education of families in case of emergencies)
- NIH Hunter Kelly Research Authority: will focus on conditions that can be detected but not treated; focusing on carrying out research on new screening technologies as well as new disease management strategies.

## Meeting Evaluation

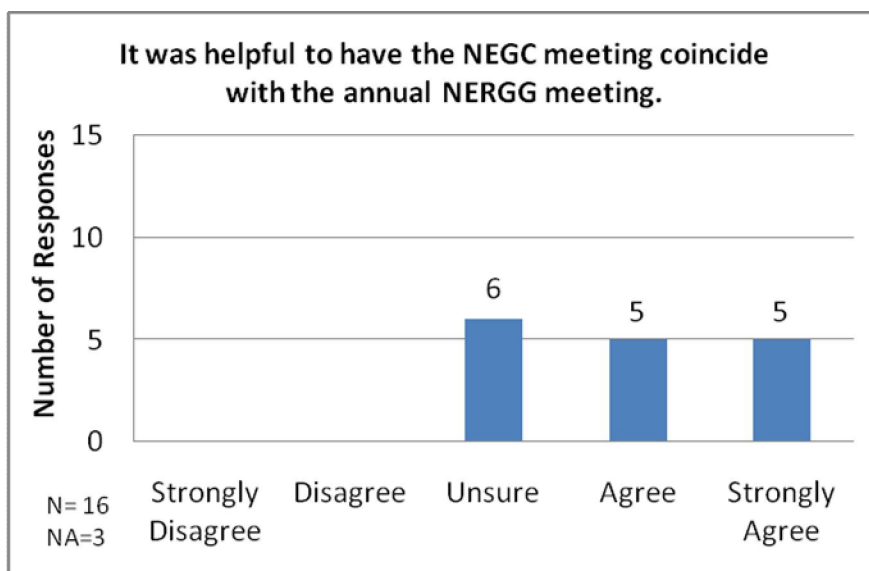
At the end of the meeting, participants were asked to complete a one page evaluation of the day's activities. Participants who completed their evaluation forms were entered into a drawing for a \$50 gift certificate to Amazon.com. Out of the 36 participants, 19 (53%) completed an evaluation form. The following pages provide a summary of the responses provided.

*Was it helpful to combine the NEGC and NERGG annual meetings?*

Out of 16 responses, 10 thought it was helpful while 6 were unsure.

Comments:

- ❖ Not clear at point in completing evaluation
- ❖ Reduces travel times and funds, increases knowledge - gives a broader perspective; foster increased collaboration
- ❖ Not necessarily for me - but clearly for others

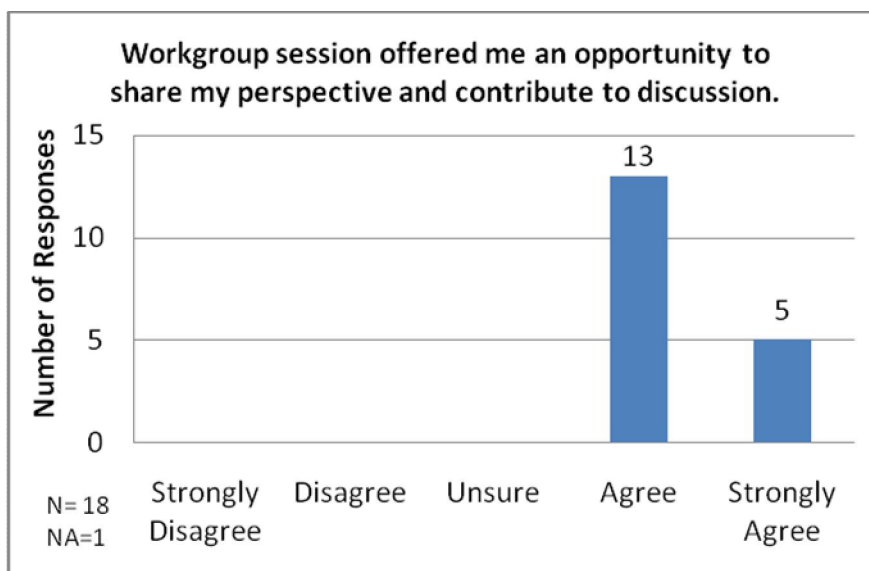


*Did participants have an opportunity to contribute to the discussion?*

Out of 18 responses, everyone agreed that the workgroup session was helpful to their ability to contribute to the broader dialogue.

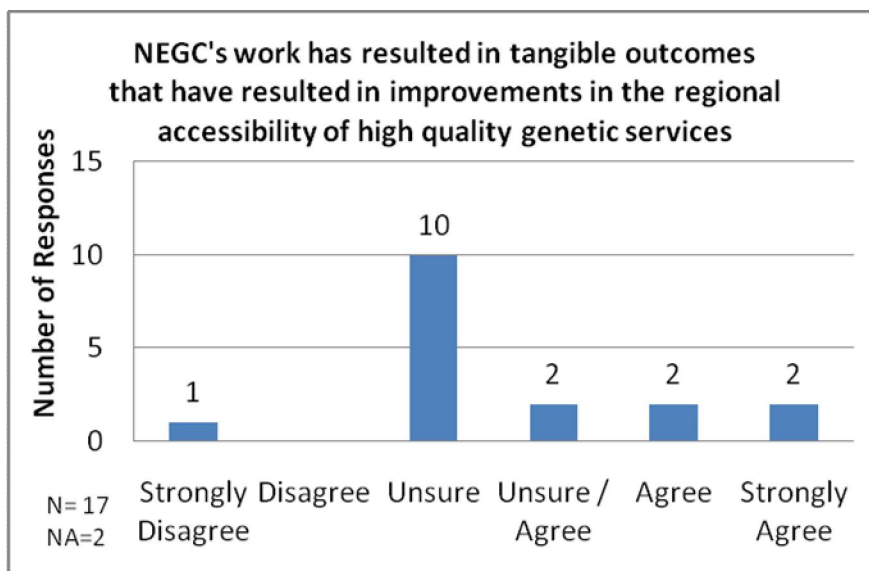
Comments:

- ❖ Need more time (3)
- ❖ The work group was small enough for everyone to participate (2)
- ❖ Very good discussion and ideas shared
- ❖ Having more like this would be good; perhaps breakouts with each working group (e.g. med home) to give more detail about updates



*To what extent has the NEGC been able to improve regional accessibility of high quality genetic services?*

Among the 17 responses, most (12) indicated some level of uncertainty with this topic, 4 agreed, and 1 strongly disagreed.



Comments:

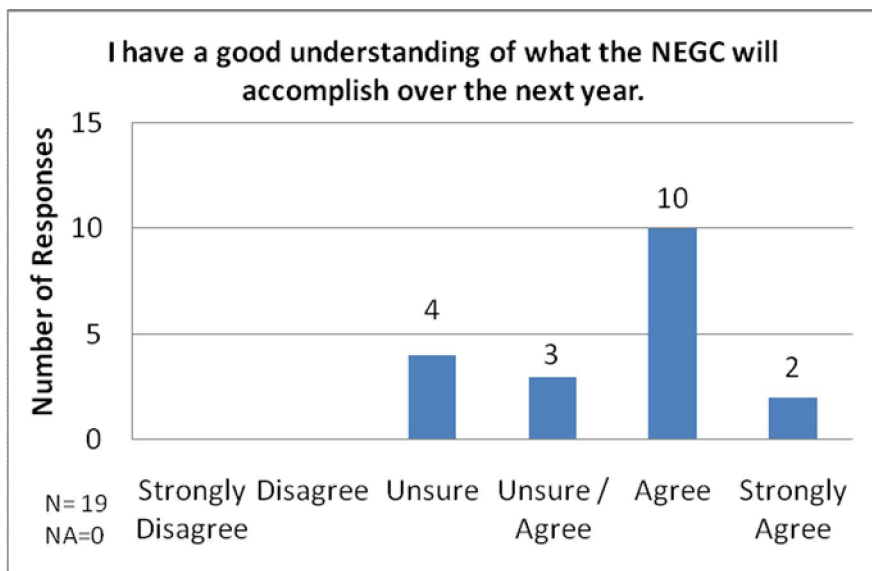
- ❖ It is difficult to extract the exact tangible outcomes of the NEGC; the outcomes need to be more visible
- ❖ Getting there...
- ❖ Haven't seen it yet but much looks promising (e.g. ACT sheets) in practice

*Did participants come away with a good understanding of what the NEGC would do over the next year?*

While 12 of 19 believed they did have a good understanding of the NEGC's plans, over a third indicated some level of uncertainty about the NEGC's next steps.

Comments:

- ❖ Seemed a bit fuzzy to me. Certain tasks will be done by LUG's and others from RCC. Unclear to me how "research" component fits in.

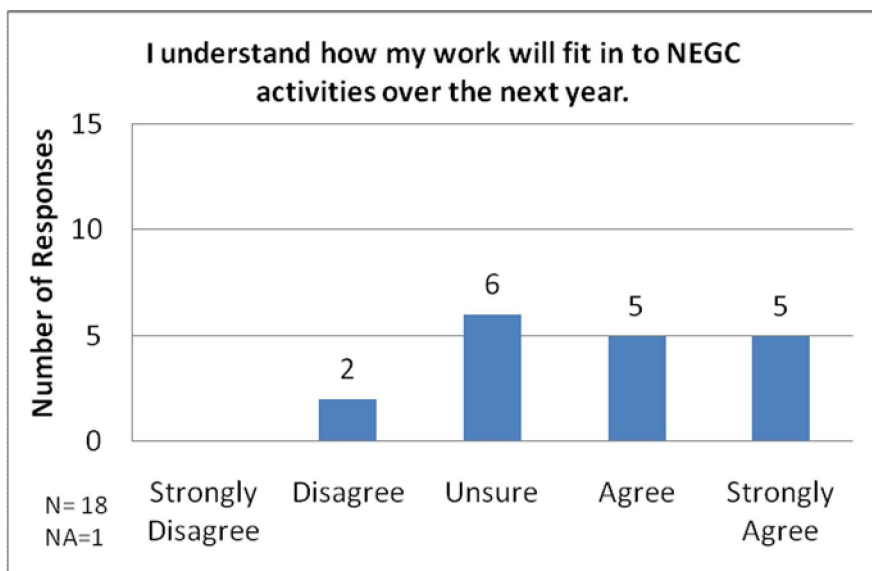


*Did participants have a good understanding of how their work will fit into the NEGC's activities over the next year?*

Of 18 responses, 8 indicated that they were either unsure or did not know how their work would fit in. 10 participants felt that they did know how their work would fit in.

Comments:

- ❖ This is my first meeting. My time is limited and not sure how involved I can be over the next year
- ❖ Consumer role

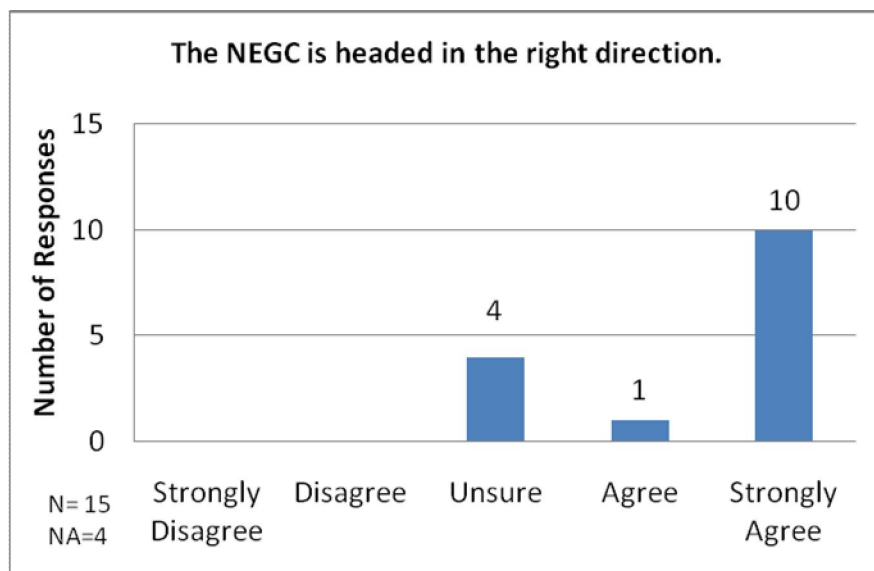


*Overall, did participants believe that the NEGC is headed in the right direction?*

Out of 15 responses, most (11) agreed, while 4 members were unsure.

*Comments:*

- ❖ Headed in direction predetermined - whether that's right...
- ❖ Yes - w/ bumps to work out; one seems to be role of consumers, would be happy to help; discuss further



*What other groups / organizations should we outreach to in order to ensure a successful collaborative effort? How should each be involved?*

Groups recommended include:

- ❖ AAP State chapters (2) to synergize with practice - based activities and/or national efforts
- ❖ Schools of public health; incorporation of genetics education; specialized epi's,etc.
- ❖ State Family Practice MD Chapters
- ❖ Other HRSA funded programs
- ❖ Consumers
- ❖ Specific consumer organizations, not just individual consumers.
- ❖ NERGG

*What recommendations did participants have for improving next year's meeting?*

Recommendations centered around the facility, improving meeting process of the meeting, and general recommendations for the NEGC.

- ❖ About the Facility
  - Larger room
  - Location was great and facility was great.
- ❖ Improving the Process of the Meeting
  - Fewer big group activities, more small group activities. Have some networking time.
  - Written summaries of what the work groups have done and plan for coming year.
  - Ask people not to wear perfume / cologne; Individual's perfume was strong. For me strong perfumes trigger headache, itching and throat irritation
- ❖ Improving the Collaborative

- Seems like there is a lot of overlap and duplication of state / regional / national efforts that could be better utilized instead of reinventing the wheel
- I wonder if HRSA or each region should contact some of the larger consumer support organizations (ie. Nat. Fragile X, NDSS / NDSC) to inform them of opportunities to become involved in the regional collaborative / Genetic Alliance or NORD can disseminate info too.... this might pull in consumers other than newborn screening.

## Conclusions and Recommendations

The annual meeting of the NEGC offered an important opportunity for the project's stakeholders to learn about the year's accomplishments, next year's goals, and the barriers and facilitators along the way that will shape the achievement of these goals. While most felt that the NEGC was headed in the right direction, and everyone responding felt like they were able to participate in the small group discussion, there remains some uncertainty about the activities of the project, people's individual roles, and the impact of the project at this stage.

In discussing some of the core areas of the NEGC's activities, participants touched on the major activities of the project, including the improvement of services, education and outreach, improving transition supports, data integration and service integration. As a result of these discussions, participants identified new resources, as well as challenges and opportunities for new directions in the project.

Concerning the utilization of new resources, several items were identified that would be useful for the NEGC to pursue. These include: the development of the new national level internet clearinghouse, new grants being made available from HRSA, information resources available via the 211 social services information system, and state level resources that have not been yet utilized by the project.

On the topic of challenges and next steps, multiple areas were identified to which the NEGC should develop a response strategy that will best meet changing needs. Challenges include:

- Multiple stakeholders and service providers at the local, state, regional and national levels providing sometimes similar, sometimes complementary services.
  - As initiatives continue to grow and move forward, it will be important to keep abreast of changes on at least a quarterly basis to ensure appropriate utilization of efforts and allocation of resources.
- Among current stakeholders, a lack of clarity about the NEGC's activities for next year, both in terms of what it will accomplish as well as what each member's involvement will be.
  - Given the number of individuals who attended the meeting indicating a level of uncertainty about the NEGC's activities in 2009, it may be assumed that a number of those not attending are likely to have similar concerns as well. It will

be important to ensure that all participants have a clear understanding of what the project will do as well as what their role will be in helping the collaborative as a whole to achieve its mission.

- The need to establish a clear message of involvement for those not involved in the project.
  - As we seek to expand involvement in the collaborative as well as develop our outreach and education efforts, there should be a clear message about what we want to see changed, what the role of individual groups would be, and a clear sense of conveying “what’s in it for them” to help promote buy-in to the project.
- Addressing additional challenges without new resources and time-stretched staff. Several issues were identified throughout the day which will be important for the NEGC to consider, including:
  - To what extent does the 211 information system in New Hampshire include information on genetic counseling / services? What information would be most helpful for them to share with residents who call in?
  - If we are relying heavily on the web as a primary source for information sharing, how do we outreach to people who are not connected to the internet?
  - At what point should the NEGC undertake efforts to address the lack of insurance coverage for essential medical supports?
  - How can the NEGC best support the creation of education modules for training physicians on how to navigate the system for medical services for children with special health care needs?

A key theme across these challenges is the need to continue improving on the NEGC’s information infrastructure as it shapes most all of the project’s components. In this regard, a number of next steps were identified in the small and large group discussions (e.g. improvement in the use of information posted on the project’s website, formation of and outreach to state teams which will have a direct line of communication into the collaborative’s structure, and regular updating of all key groups).