

GENETICS AND NEWBORN SCREENING REGIONAL COLLABORATIVE (RC) GROUPS

Monthly PI Call
Thursday February 15, 2007
2:00 – 3:30 PM EST

CALL SUMMARY

Attendance: Tom Brewster (NERGG, joining late); Katharine Harris and Bonnie Frederick (NYMAC); Hans Andersson, and Rani Singh (SERGG); Janice Bach and Cynthia Cameron (Region IV); Lori Williamson (Heartland); Liza Creel and John Johnson (Mountain States); Sylvia Au and Kerry Silvey (Western States); Judith Benkendorf, Gloria Weissman and Mike Watson (NCC); Michele Lloyd-Puryear, Penny Kyler, and Jill Shuger (MCHB/HRSA).

Mike Watson moderated the call. It had been previously decided that this call would feature an overview by the NCC of activities that involve presently established workgroups or will evolve into the development of a workgroup.

I. General Information

The April 19th conference call will need to be rescheduled because both Mike and Judith will be unavailable. Group consensus was to keep the call during the 2:00-3:30 PM (Eastern time) Thursday time block. Judith will poll the group to determine participant availability on April 12th and April 26th (either a week earlier or later). NOTE: This was done immediately following the call; feedback due back to Judith no later than Thursday February 22nd.

Topics for the March 15th call to be determined at end of this call, so time will be left for this activity.

II. Summary of Emergency Preparedness for NBS and Genetic Service Meeting, held February 13th in the Washington DC area

This meeting is an outgrowth of the workgroup (WG) formed in July 2006 and co-chaired by Drs. Ken Pass and Jess Thoene. The meeting brought together representatives of the medical genetics and public health communities, State and Federal government agencies, industry, a telecommunications expert and a consumer advocate. Mike reminded call participants that many of the ideas that incubate locally are brought to the national level through RC participation on WGs. Disaster response plans and activities from NYMAC, SERGG, Heartland and California were all presented at the meeting. Meeting participants began by delineating the needs of this unique patient population. Other over-arching themes included how to better manage a disaster, and how to interface with stakeholders and work with people in the disaster area. The agenda also included the topics below, several of which were highlighted by Mike on the call:

- Lessons learned from 9/11 and Hurricane Katrina
- Existing Regional Disaster Response Plans and other models

Kathleen Velazquez presented the disaster preparedness plans and activities of the CA Department of Health, including response to earthquakes; Stan Berberich (Iowa) discussed a model program that IA's NBS lab is developing with neighboring states (MI, WI, MN) through their EMAC program. He sees this as a model other states should be able to replicate. It was noted that the participating states are in Region IV, and so there may be opportunities to expand this to the rest of that RC, as part of the model program. Heartland also has a proposed project coming in with the renewal that addresses NBS Back-Up Testing and Quality Assurance.

National organizations/agencies that participated in the meeting included EMAC, which deals between states and was represented by Angela Copple; CDC, which presented their Emergency Communication Systems, and Dr. David Canton represented the Office of the HHS Secretary and discussed the National Disaster Medical System (NDMS). The NDMS sends physicians and other health professionals into emergency areas. Most of these are primary care providers, but all in attendance agreed that the system could benefit from the additions of medical geneticists—especially metabolic specialists—who can be available as consultants to the teams on location following a disaster.

- Shelley Bowen, of the Barth Syndrome Foundation, discussed the roles and needs of patient support and advocacy organizations.
- The roles of lab and medical product manufacturers during a disaster were covered from the perspectives of formula, orphan drug and lab equipment and reagent industry representatives. Of particular interest is how the therapeutics manufacturers interact with patients.
- Needs of patients in rural and remote areas
This presentation emphasized the multiple applications of telecommunication systems. Omar Pecantte was brought in from a CHC in rural Louisiana by Bruce Bowdish, via the Collaborator System (which Hans Andersson had demonstrated at the October 2006 NCC AC Meeting). As a follow-up activity Mike has asked Bruce Bowdish to look at the variety of systems, including the Collaborator system, for a variety of communication issues such as HIPAA compliance, teleconferencing with capacity for slides, white board and participant viewing, tele-education, and telehealth. Mike will put out to the RCs information about the telecommunication systems so the RC's can help decide what is the best system for the RC's use.

Next steps: The Workgroup will write a white paper that will highlight the components of NBS system and the actions needed around disasters. There is not a national disaster plan that can fit all localities in all disasters; plans must be local and even institutional, but the white paper and concomitant tools should help all to develop emergency preparedness plans that cover each step in the NBS and subsequent genetic services processes—from getting specimens to getting treatment. There are plans to have it in a checklist format so people can use it at the local level. We will also develop some drills that each center can do for itself at the local level. NCC will also attempt to identify next sources of funding, as these programs (i.e., developing a COOP, or Continuity of Operations Plan) can become expensive. We will go to the genetic support groups, other non- profits and other federal agencies to see about additional sources of funding. Penny Kyler also suggested looking to state endowments.

III. Development of a National Collaborative Data Collection System useful for Translational Genetic Disease Research

On Monday and Tuesday February 19-20, 2007 there will be the first of 3 yearly international meetings on this topic, supported by NICHD (NIH) funding of ACMG, through the NCC grant, to look at expert-driven protocols that are disease specific. This meeting has been planned by a Steering Committee, but the activities it identifies are expected to generate a RC-based workgroup. RCs are in a very strong position to become integral to these data collection efforts; for example, a better evidence base is needed for adding conditions to NBS.

Cooperative medical study groups are often based at academic medical centers. Keeping patients close to home allows for an improved educational process and can provide a mechanism for requiring high levels of lab performance. The meeting will examine the value to research of activities such as rare disease centers, a national newborn screening translational research network and building an alliance with states with large numbers of births (25% of US births occur in NY, CA, TX FL).

NICHD has great interest in NBS and funds projects that ask specific questions about NBS cohorts. Two RFAs are presently out: one addresses new technologies and the other involves treatment for conditions

associated with NBS. We are talking with NICHD regarding the possibility of developing the NCC as a TA center, with supplemental funding beginning in October 2008.

- There will be a forthcoming meeting with SIMD to develop a national metabolic disease consortium.
- Beginning at the February 19-20 meeting, there will be an assessment of various databases that can be used for national data collection. Ken Buetow will be presenting the NCI database system, called caBIG, as a model. This system is federated and uses modularized information. It is able to deal with multi-center IRB's consent documents, etc.
- There was follow-up discussion regarding roles of NIH, ACMG, and the RC NBS network. At what level should there be involvement or the RCs? The project currently is focused on defined research topics, data from working with patients that have been lost. How can we have complementary partnerships? The goal is to build a national system, a database system that can be used across the country, realizing that there are groups doing things independently and that will continue.
- Michele Puryear reported that a subcommittee of the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC) is developing a meeting, to be held April 18, 2007, to address long-term follow-up. Those RC projects that have been actively involved in this area are going to be asked to attend. Dr. Alex Kemper (Duke University) is part of the organization of this meeting, which plans to produce a white paper.

IV. GIS Mapping System/Provider Directory

The NCC is in the process of developing a Genetic Services Provider Directory that will use a GIS mapping system (built with the Google Maps system). The final product will be expanded beyond genetics providers. Once all information is input Mike will share it (as requested by the RCs), as we would like all listed clinics or providers to confirm the accuracy of their information; additionally, the RC's will want to compare the information in this database with information they have collected, so there is compatibility. It is expected that some information exchange between the RCs and NCC will occur at this step. Discussion took place regarding who would be involved and how to share the information with RCs. This may be the place to re-engage the Provider Network Workgroup, formed in late 2005. Hans Andersson pointed out that directory will be an excellent disaster preparedness resource, so we need to make it accessible. For this to happen we will need to plot out what types of testing is occurring, and where, as well as what information patients need in case of emergency. (Providers will need to find back-up labs and patients will need to find providers if dislocated.)

V. Telegenetics Workgroup (Benkendorf)

This telegenetics workgroup, co-chaired by Hans Andersson and Becky Butler has been fairly active. A survey was drafted a survey to assess the level of telegenetics activity nationally, barriers that need addressing and to identify a cohort of experts. It will be disseminated electronically to all genetic service providers and through the RC's to the different state programs. We are incorporating the last round of services and the survey should go to the survey vendor within one week, for dissemination via the web within 6-8 weeks. The survey is brief and user friendly.

VI. NCC Newsletter

Because of several unfortunate delays, the reports submitted over the summer by the RCs are aged. These will be sent back to all authors for review, with a return date of February 22nd (NOTE: Done immediately following the call). The newsletter will be electronically disseminated to the RCs and the NCC partner organizations; it will also be posted on the NCC website. We expect this to occur prior to the mid-March ACMG Clinical Genetics Conference.

VII. ACT Sheet Projects

There are several activities occurring, enhanced by our 1-year supplemental grant: 1) Three hematologists are developing another set of ACT sheets specific to variant hemoglobinopathies. We expect these to be done in next 8 weeks. 2) ACT sheets are under discussion for active NBS pilot tests, two such conditions are SCID and Krabbe disease (pilot in NY State). Kathy Harris offered a flowchart on workup of Krabbe disease following a positive screen, which is on their website. 3) ACT sheets for positive genetic tests, to be disseminated by genetic testing labs, are being drafted by workgroup. The cystic fibrosis (CF) ACT sheet will address CF from diagnostic and carrier screening perspectives. Fragile X testing will be covered by a similar ACT sheet. A meeting in May will move this work forward. 4) As a way to increase the likelihood that genetic services are included in the etiologic evaluation of hearing loss in all babies detected through state EHDI programs, Dr. Kathy Arnos, clinical geneticist at Gallaudet University and recipient of an ACMG Foundation award to develop a genetics education module for EHDI professionals (to be delivered in conjunction with the national EHDI meeting in late March, 2007) will spearhead a project to take the ACMG practice guideline and adapt it into an appropriately tailored brochure for parents. The rationale is that if parents are informed about the components of a comprehensive workup for the etiologic basis of deafness they will request (or at least inquire about) genetic services. Dr. Karl White, Director of the National Center for Hearing Assessment and Management (NCHAM) will also be involved.

Once completed, all of these materials will be available to the RCs for distribution and use regionally and locally.

IX. Topics for Future PI Calls

The following topics were suggested for the March PI conference call. They will be sent to the PIs to decide which to place on agenda.

- SERGG Laboratory Performance Project (Singh)
- EMR
- Evaluation – This remains an important topic, both in terms of current efforts and future program development
- Region IV's planning process and web site integration of planning and work plans (Cameron)
- Mountain States – transition, QA, and LTFU projects (Johnson)
- RC small grant projects (e.g., SERGG's Ask the Geneticist project)

For the April call Western States offered to lead a discussion of defining genetic services, which would include findings of work by Celia Kaye and the NNSGRC as well as the Washington State Genetic Services and Policy Project.

Next Actions:

Revised RC reports for newsletter and preferred date for April PI Call (poll response) due to NCC by Thursday February 22nd.

PIs to vote on topics choices for March call agenda.