




NCC Collaborator

Expanding Genetic and NBS Services Through Multifaceted Partnerships



RC Tools Move from Concepts to Communities

The authoritative Merriam-Webster dictionary defines a tool as an implementation aid, such as would be used to accomplish a set task or as a means to an end. In this issue of the *NCC Collaborator*, the HRSA Genetics Collaboratives (RCs) report on the many ways the successful tools they have developed have helped them leverage existing resources, grow their programs, and expand services to their stakeholders.

Region 4 and one of the states in the Heartland RC are tackling issues related to public attitudes about the storage and use of residual newborn screening bloodspots in research. Meanwhile, state newborn screening labs in the Heartland RC have been recognized by the Association of Public Health Laboratories for demonstrating that

less than optimally collected bloodspots can provide clinically meaningful results, thus giving newborn screening a new tool in the area of disaster preparedness.

On one coast, the NYMAC Genetic and Newborn Screening Services Consumer Leadership Project is growing a pipeline of consumer leaders to assist the RC in developing and assessing projects in the region, while on the other, the Western States Genetic Services Collaborative is using the results of their stakeholder assessment to improve RC programs, communication, and overall effectiveness.

When current information is the missing tool, all RCs can look to the National Human Genome Research

Institute's Genetics/Genomics Competency Center, just launched as a free, web-based repository of curricular materials on genetics and genomics. Others can turn to the Southeast RC, where hemoglobinopathy experts have revamped The Sickle Cell Information Center website as a robust resource for the sickle cell community and an informational database and tool to engage experts, patients and families. And, nothing ties together consumers and information like the Advocates Partnership Program at the ACMG Annual Meeting, also featured in this issue.

Finally, the Mountain States and New England RCs continue to utilize small grants to support projects in their regions that might not be funded from other sources. Startup funds from Mountain States RC have been leveraged into several larger national grants, while New England RC has developed micro-enterprise grants to help local support groups and organizations sustain their programs and services.

With the Regional Collaborative system just completing its sixth year, it is exciting to see the RCs continue to develop new tools and adapt old ones to meet both their unique needs and their shared commitment to improving genetic and newborn screening services.

June 2010
Volume 4
Number 2

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Determining Knowledge and Attitudes of and Parents-to-be on Newborn Screening

Submitted by Sharmini V. Rogers, MPH, MBBS, CPHG, State Genetics Coordinator, Missouri Department of Health and Senior Services and Heartland Regional Collaborative

"To Save or Not to Save is the Question"

The question of whether to save newborn screening bloodspots is causing turbulence in some states and has become a battlefield in others. Two states, Texas and Minnesota, have been dealing with litigation from parents who are unhappy with the idea of storing the bloodspots or with the fact that they were not made aware that their infant's bloodspots were being stored and given the option to decline. The Minnesota case was ultimately dismissed, but the Texas case resulted in a requirement that the Texas Health Department destroy more than 5 million residual dried bloodspots and release the list of all research projects involving those specimens. The negative publicity surrounding bloodspot retention has prompted states to take another look at their bloodspot retention policies and protocols. This is important both to regain the public's trust in states' abilities to maintain personal privacy and to avoid further disruption to the current newborn screening (NBS) system.

Missouri passed legislation in 2007 allowing the State Public Health Laboratory to store the NBS bloodspot cards for five years. The law also allows the bloodspots to be used for anonymous research. As a requirement of the Public Health Genomics Certificate Program at Sarah Lawrence College, for which I was competitively selected and sponsored by the Heartland RC in 2009, I undertook this study in Missouri to:

- Assess the public's awareness of and attitudes toward newborn screening;
- Examine attitudes around bloodspot storage;

- Determine the best way to let parents know their options regarding the state's storage of their child's blood and its use for research;
- Document parent feedback regarding the issue of bloodspot retention; and
- Develop policies and procedures for bloodspot storage and research.

Methodology

To learn more about parents' and other interested parties' knowledge of and attitudes toward NBS and bloodspot retention for research, six focus groups were held around the state of Missouri, supported by funds set aside for the Maternal and Child Health Block Grant's five-year needs assessment. These meetings were held in the western, eastern, central, and southern regions of the state, with a total of 83 individuals participating. The focus group questions were taken from the Michigan Newborn Screening and Blood Trust Initiative Project, although our Facilitator Guide modified some questions to meet the needs of the Missouri Newborn Screening Program. Focus group participants also completed a survey with the same questions, once the group discussion was completed.

In addition, surveys were sent to a random sample of parents of children who had newborn screening to determine their knowledge and attitudes about NBS screening and the blood storage and research process. A total of 750 surveys were sent out: Approximately 50 were returned due to incorrect addresses and 94 completed surveys were returned and analyzed (response rate =13%). A higher response rate had

been expected but fiscal constraints prevented a planned second mailing of surveys.

Results

Of the 83 focus group participants, 51% percent were between the ages of 18-20 years; 35% percent were 21-29 years; 8% were 30-39 years; 2% percent were > 40 years and 4% did not respond. As for racial differences, the majority of the participants were African Americans (52%); the second largest group was Caucasian (31%); followed by Native Americans (10%); Asians (2.4%); and "Other" (2.4%). 2.4% of participants did not list a race.

Overall, the majority of focus group participants were not aware of the NBS program or blood storage law. While they favored using bloodspots for research they felt that the research had to be done in academic settings rather than private sector (Table 1), with a preference for their use in research of childhood conditions (Table 2). Most importantly, the majority of the participants did not care how their permission was obtained, but felt they should be allowed to make the decision whether their infants' bloodspots would be used for research. They wanted to be given options. When

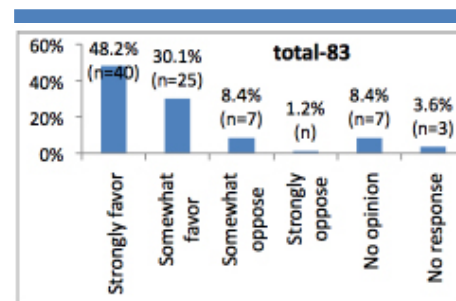


Table 1: "How strongly would you favor or oppose the use of leftover de-identified bloodspots for research?"

Parents and Bloodspot Storage

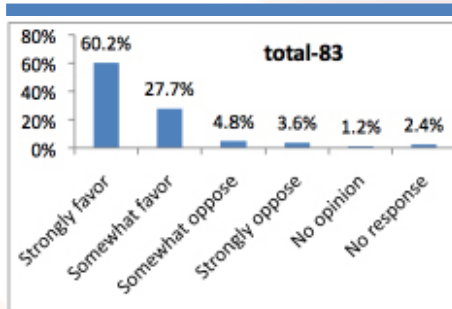


Table 2: "How strongly would you favor or oppose the use of leftover de-identified bloodspots for research of childhood conditions?"

asked what they were going to do with the knowledge they now possessed about newborn screening and blood storage, all participants said that they were going to tell their families and friends about it. Many also took the Missouri Newborn Screening brochures that were brought to the focus group meetings. The comments below illustrate the range of sentiments held about this issue:

"My primary concern... is that there's a lot of fear in the hospital about particularly if you're young or if you're on Medicaid, or you're a person of color, that they're taking baby's stools and they're doing drug tests on them—there's a lot of fear about that right now, that if they think that you're off, somehow, that they're going to be doing that. And so there's a lot of distrust about the system from people, and I don't know if that's true or not, or if it's just one of those rumors that's flying around. I mean, they're scared, and I think that if they heard that stuff was being stored for five years, there would be a lot of fear about like genetic profiling, and coming back."

"I like research. I'm a doctoral student, and so I understand the concerns that people have about the profiling and everything, but at some point you just have to trust that if they say it's going to be anonymous, it's going to be anonymous."

"I think that's great, and I don't think you should have an option. I mean to be honest, I think that would be selfish if you were like, 'No, I don't want that,' because you're already getting your child's foot pricked, and why not if it's gonna help another child."

As described previously, the response rate for the mail-in survey was lower than we expected and additional resources to send a second round of surveys were not available. However, the information gathered from this survey will still be helpful because unlike many focus group participants, those who sent in the survey by mail had recently given birth. While 66% of these survey respondents indicated that they knew about newborn screening, 94% were not aware of the bloodspot storage law, yet 79% of this latter group were in favor of using the leftover specimens for research. Overall, it appears that parents of new-

borns understood the importance of newborn screening and were in favor of using the specimens for research, irrespective of the types of research or the populations who will benefit.

"I think that's great, and I don't think you should have an option. I mean to be honest, I think that would be selfish if you were like, 'No, I don't want that,' because you're already getting your child's foot pricked, and why not if it's gonna help another child."

However, parents overwhelmingly wanted to be asked permission to have the bloodspots used in research, though the methods for obtaining permission were of less importance to them.

FOCUS GROUP DISCUSSION QUESTIONS

- What have your experiences been with newborn screening (heel prick)?
- What are your thoughts about storing the bloodspot for five years to use for 7 medical research if all personal information is removed?
- What types of research using dried bloodspots do you feel would be acceptable?
- How should consent to use the bloodspot sample for research be obtained?
- Now that you have heard this information, what would you tell your friends?

the new england **negc** genetics collaborative

Submitted by Monica R. McClain, PhD, Project Manager and Katherine McCormick, MBA, NEGC

NEGC Community and Family Network Grants

In 2009, the New England Genetics Collaborative (NEGC) created Community and Family Network (CFN) Grants to encourage the development of small community-based projects that address the needs of people with genetic conditions and their families. The CFN Grant program was conceived as a way for NEGC to reach out to family-driven non-profit groups for whom the typical grant process can be difficult. Members of the Collaborative pooled their ideas and developed the concept for the program, along with supporting policies and procedures. These were all enthusiastically endorsed by the NEGC Advisory Committee. For the first round of grants in 2010, a total of \$10,000 was made available, but each award was capped at \$2,500.

In reviewing grant applications, the NEGC review team looked for:

- The likeliness the monies will make a real impact and have lasting benefit;
- How the grant funds may help in the continuance or sustainability of a long-term project; and
- The overall needs of the organization or group that would be receiving the funds.

The intent of these grant projects is to contribute to the overall mission of the NEGC, which is to promote and improve the health and social well-being of those with inherited condi-



tions through collaborations among public and private health professionals, educators, consumers and advocates in the New England states. It is our expectation that recipients of the grants will give back to the RC by completing a brief form describing their experiences; we will also invite them to participate in our annual meeting, if possible.

This year's round of grants has resulted in some exciting projects and activities:

- The Rhode Island Parent Information Network utilized its grant funds to support the attendance of 36 families at their annual Family/Professional Partnerships Conference. This enabled them to provide these families with access to extraordinary expertise and information about best practices for children with special health care needs. Families attending the

conference also had opportunities to build relationships with others dealing with similar issues.

- Nine family leaders representing three Family Voices chapters—Family Voices at Parent-to-Parent of Vermont, Massachusetts Family Voices, and Family Voices of Maine—used grant funds to attend the Region 1 Family Voices/F2F family leadership conference.

The NEGC views the CFN Grant program as a great step forward in building collaborations to improve the care and well-being of individuals with genetic conditions, their families, and their communities.

<http://www.negenetics.org/>



Submitted by Kate Tullis, PhD, Patient and Family Coordinator and Katharine B. Harris, MBA, Project Director, NYMAC

Building a Consumer Leadership Pipeline

NYMAC is very excited to report that our Consumer Leadership Pipeline project, which grew out of ideas from the October 2009 Strategic Planning Meeting, and is being championed by Diana Autin, co-director of the New Jersey Statewide Parent Advocacy Network, is well underway. This project allows us to build on the strong consumer training programs of Family Voices and Genetic Alliance.

In April 2010, 19 scholars attended the first training session for this program. The session was held in conjunction with the Family Voices Region 1 bi-annual meeting. NYMAC invited parents of children and youth with special healthcare needs (CYSHCN) from throughout our region to apply for scholarships for this new leadership-training program by distributing information directly to consumers as well as to specialty care providers and to the Family Voices Chapters in our region.

At the April Family Voices meeting, the scholars learned about HRSA/MCHB's six core outcomes for CSHCN (mchb.hrsa.gov/cshcn05/MI/cokmp.pdf) and the roles played by MCHB-funded National Centers, Family Voices organizations, and the HRSA Genetics Collaboratives to accomplish these outcomes. More importantly, they explored the ways in which family leaders can and should get involved in building a comprehensive system of care for these children and youth. Sessions on leadership

skill development focused on listening, working with people with varied conflict styles, and creative problem solving.

The NYMAC scholars heard presentations from representatives of the Catalyst Center (a national organization dedicated to improving healthcare insurance and financing for CYSHCN; www.hdwg.org/catalyst/) and the Co-Directors of the Family Voices Policy Team on the implications of the new federal healthcare law for CYSHCN. Presenters stressed the importance of parent leaders sharing their experiences and needs with policy makers as regulations are developed and this law implemented. Other presentations focused on the legislative process.

Attendees were inspired by the stories told by family leaders who have successfully influenced public policy to ensure that healthcare reform helps CYSHCN and their families.

NYMAC is currently planning to hold several conference calls with the new consumer leaders who participated in the April training. These calls will introduce them to NYMAC and to current and potential projects in the region. The consumer leaders will get together again in July to attend the Genetic Alliance Annual Meeting and to

take part in the Genetics Day on the Hill program. Most important, the consumer leaders participating in this project will form a Consumer Caucus to ensure

that their issues and personal perspectives are considered in all NYMAC activities. By instituting a vigorous formal consumer-training program in conjunction with Family Voices and Genetic Alliance, NYMAC hopes to better address the needs of consumers already involved in our projects, support new consumer-focused projects, and establish permanent consumer/professional partnerships that will enhance genetic services and inform public policy.

<http://www.wadsworth.org/newborn/nymac>



SOUTHEAST NBS & GENETICS COLLABORATIVE

Submitted by Alan Platt PA-C, MMSc, Web Master, Sickle Cell Information Center and Elizabeth Otwell, MSPH, Project Coordinator, SERC

Improving Online Resources for the Sickle Cell Community

There is a great need for better understanding of optimal symptom and pain management for people with sickle cell disease. To help meet this need, in 1991 the clinical guidelines developed in the Georgia Comprehensive Sickle Cell Center were published in a handbook titled *Problem Oriented Management of Sickle Syndromes*. The handbook was funded by HRSA's Maternal and Child Health Bureau and distributed worldwide. Because of the Internet's potential for more widespread, cost-effective education, in 1998 these guidelines were included on a website called The Sickle Cell Information Center (www.SCInfo.org).

Since that time, this website has been online continuously and has attracted visitors from around the globe. Content is maintained by Allan Platt, PA-C, with input from James Eckman, MD, SERC Hemoglobinopathies Workgroup Chair, and Lewis Hsu, MD, PhD. Current content areas include:

- a sickle cell disease overview for the lay audience;
- several online guidebooks for health care providers;
- pain management guidelines;
- research and news updates;
- a list of US sickle cell clinics and centers;
- web links;
- streaming video presentations; and



- a means of submitting e-mail questions to be reviewed and answered by appropriate medical staff.

The website currently receives 500,000 hits a month. The monthly e-mail news update, which provides readers with the latest news in research, education, and clinical care, has grown from 1,500 participants in 2001 to 4,500 in 2010. The site is on the first page of results for the Google search engine for the term "sickle cell" and has "Health on the Net" certification.

The website is currently being updated and modernized, using a web-based content management system to provide better content, links and functionality. In addition to these improvements, a new website template was created to give the Sickle Cell Information

Center a clean, updated look. The site overhaul is being led by Adrya Stembridge, an application developer responsible for the SERC website (www.southeastgenetics.org), as well as the State of Georgia Newborn Screening Follow-up program database.

Although an official launch date for the redesigned website has not been set, updates will appear in the monthly Sickle Cell Center newsletter. Interested readers should join the free monthly e-mail listserv at www.SCInfo.org by clicking on the "Join the Newsletter Listserv" link.

<http://www.southeastgenetics.org>



Region 4 Genetics Collaborative

Submitted by Jodi Griffin, MPA, Project Coordinator, Region 4

Panelists Discuss the Changing Moral Focus of Newborn Screening at 2010 Regional Meeting

With every state mandating newborn screening (NBS) and retention of the bloodspots used for screening for some period of time, a rich supply of residual dried bloodspots has accumulated in the US. Although collecting dried bloodspots (DBS) always has been and will continue to be about NBS, there is a new focus on actively promoting the use of these specimens for research. This has raised complex ethical questions, including those related to indefinite retention and the need for parental consent.

The Region 4 mission includes increasing access to information about newborn screening and genetic resources, services, and family support systems. The Regional Collaborative thus has an important role to play in informing and educating parents as existing policies and processes around dried bloodspots are re-examined and new guidelines emerge to address the expanded use of these bloodspots for research. We believe there needs to be continuing discussion with parents so that they can make informed decisions about NBS, as well as determine whether to consent to use their infants' bloodspots for research. To this end, at our 4th Regional Meeting in April 2010, Region 4 included a panelist session on the challenges and ethical issues surrounding the use

of bloodspots gathered from the NBS process.

Panelists Alissa Johnson (Principal Consultant, Johnson Policy Consultants), Aaron Goldenberg (Assistant Professor, Case Western Reserve University), Amy Gaviglio (Genetic Counselor, Minnesota Department of Health, Newborn Screening Program), and Denise Chrysler (Public Health Legal Director, Michigan Department of Community Health) provided a range of perspectives including:

- Overview of state perspectives and policies concerning retention and secondary use of bloodspots for research (Johnson);
- Implications of the views of the public health, research and practitioner communities on the ethical challenges of bloodspot use; review of consent materials; and studies on parental concerns (Goldenberg);
- Education and engagement of parents and the public through transparent, informational, assurance-based, and actionable materials that give parents authority over the use of their infants' bloodspots (Gaviglio); and
- Legal issues around the structuring of dried bloodspot research programs (Chrysler).

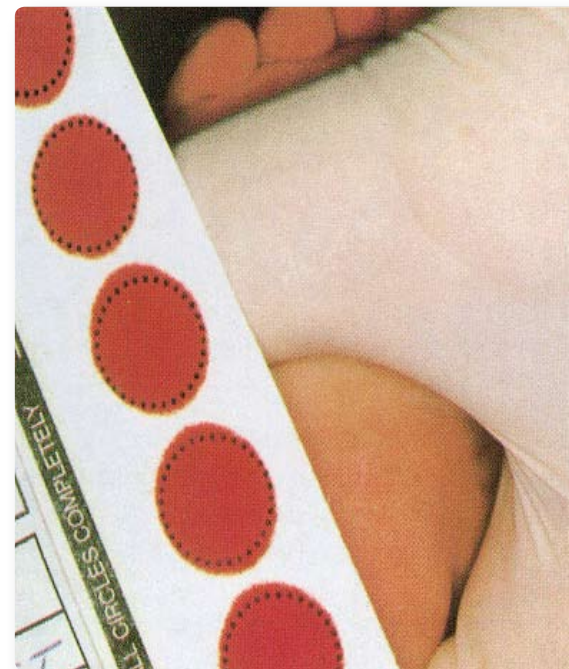
Meeting participants responded with great enthusiasm, contributing questions and ideas for addressing some of the challenges. Issues raised included:

- Costs associated with banking/storage and retrieval processes and potential funding sources;

- Utilization of social media and other outlets to educate parents on the consent process;
- Increased dialogue to explore promising practices and lessons learned;
- Ensuring the utility of NBS is not confused with the issues of storage, use and research; and
- Strategies for educating providers and parents during the prenatal period.

Region 4 has a diverse community of stakeholders with the expertise to examine the myriad of practical and ethical issues raised by the use of NBS bloodspots for research. We plan to support continued discussion to facilitate the development of sound policies and promising practices around this research.

<http://region4genetics.org>





Heartland Genetics and Newborn Screening Collaborative

Submitted by John J. Mulvihill, MD, Program Director, Heartland RC

The Heartland Good Spot/ Bad Spot Project

Each spring, the Heartland Newborn Screening Workshop convenes the region's newborn screening (NBS) professionals to share problems and to propose solutions and novel procedures to be piloted. The group is gaining national recognition for its interstate exchanges of specimens in preparation for the possible disruption of any of the state newborn screening programs due to a disaster.

Now, the workgroup's assessment of so-called "unsatisfactory" (UNSAT) bloodspots has earned the best poster award at the recent Association of Public Health Laboratories symposium. If results from UNSAT specimens could be validated rapidly, this would eliminate the need to wait for a perfect second specimen, which is particularly problematic in a disaster situation. And, recognizing affected babies sooner can save their lives! CLIA inspectors in two Heartland states currently prohibit laboratory analysis of an UNSAT spot (e.g., insufficient, over-saturated, layered, or clotted blood), making the findings of the workgroup's study of critical importance to public health.

We conducted a two-part study. In the first part, an under-saturated sample



was simulated by purposefully punching a spot with 50% white filter paper showing, and an over-saturated spot was simulated by putting two punches in the testing well. Of under-saturated samples from 135 true and confirmed positive cases, 117 (87%) remained positive, 9 (7%) became borderline, and 9 became normal. None of the over-saturated spots became borderline, and 4 (3%) converted to normal: 3 were carnitine uptake deficiencies and 1 was D/G galactosemia. Part two of the study looked at 1,996 assays of normal samples (an average of 218 specimens for each of 9 analytes); the findings are illustrated in Figure 1. No under-saturated normal spot converted to abnormal, and 43 (2%) of the over-saturated normal specimens became abnormal.

The group concluded that analyzing UNSAT specimens to look for the most serious disorders could be accom-

plished without undue numbers of falsely positive or negative results. In fact, doing so may be the only chance to provide life-saving intervention for a child with a true disorder. As a side benefit, the project independently illustrates the merit of long-term storage of dried blood spots for use in research to improve test performance.

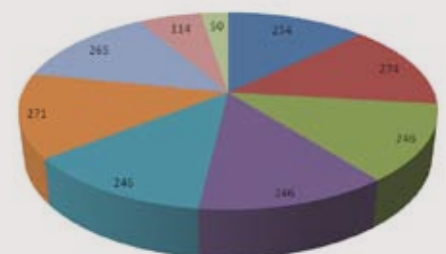
Adapted from an award-winning poster by McCallister, Himstedt, Ross, Freer, Valbracht, Berberich, Ramirez, Kline, Eiken, and Hopkins presented at the 2010 Newborn Screening and Genetic Testing Symposium of the Association of Public Health Laboratories, Orlando, FL, May 3-6, 2010.

<http://www.heartlandcollaborative.org/>

Figure 1: Frequency of analytes tested

TSH	Hypothyroidism
GALT	Galactosemia
Phe	Phenylalanine
C8	MCADD marker
C3	PA and MMA marker
17-OHP	Congenital adrenal hyperplasia
IRT	Immunoreactive trypsinogen
Hb	Hemoglobinopathies
BIO	Biotinidase deficiency

■ TSH ■ GALT ■ Phe ■ C8 ■ C3 ■ 17-OHP ■ IRT ■ Hb ■ BIO





Submitted by Celia Kaye, MD, PhD, Project Director; Joyce Hooker, Project Manager and Liza Creel, MPH, Project Coordinator, MSGRCC

Leveraging MSGRCC Funding and Infrastructure Leads to Success for the Entire Region

Since 2004, The Mountain States Genetics Regional Collaborative Center (MSGRCC) has funded small projects that contribute to its mission of improving access to genetic services in the region. Several current projects in the region started, at least in part, with MSGRCC funding and have leveraged our support to obtain additional grants and resources. These projects exemplify the many ways in which MSGRCC funding and capacity have been employed to expand access to genetic services, information, and expertise in the region, fulfilling a central RC belief that successful work should be sustained.

The Colorado Sickle Cell Care Network (PI: Karen Chacko, MD, with Kathryn Hassell, MD), funded by the Centers for Disease Control and Prevention (CDC), seeks to coordinate sickle cell disease services and programs statewide, as well as to identify and integrate new resources and partnerships as new areas of need are recognized. The Network complements a current MSGRCC-funded sickle cell disease project, also led by Dr. Hassell, which focuses on identification of sickle cell carrier populations and provision of education to families in the region through the Medical Home.

Evaluation and Implementation of Second-Tier Testing for Disorders Identified by MS/MS in Newborn Blood Spots in the Mountain States Region (PI: Marzia Pasquali, PhD) is funded by CDC and builds on a Laboratory Quality Assurance Project that MSGRCC funded between 2007 and 2009 (see *NCC Collaborator* Vol. 3, No.3, page 9, December 2009). Dr. Pasquali's new project provides a resource for second-tier testing to all states in the region and is attempting to determine if this process reduces out-of-range results.

The Colorado Effective Follow-up in Newborn Screening Project (PI: Chris Wells, MS, PhD) is funded by the Genetic Services Branch in HRSA's Maternal and Child Health Bureau. It builds upon the "Metabolic Newborn Screening Long-Term Follow-Up Study" funded by MSGRCC since 2005 and led by Janet Thomas, MD. Dr. Thomas's work on developing the disease-specific care plans and shared datasets needed to establish uniform data collection and outcome measurement of long-term follow-up (LTFU), along with the Colorado Department of Public Health and Environment's commitment to developing a comprehensive health information exchange system, have both provided the foundation for LTFU in Colorado, Wyoming and other states in the region.

As featured in the April issue of the *NCC Collaborator* (Vol. 4, No. 1, page 9) Laura Pickler, MD, MPH, is leading the

MSGRCC-funded project **Improving Medical Homes and Transition Outcomes for Youth Served in IMD Clinics**, which is implementing Medical Home standards within a specialty clinic that serves youth with inherited metabolic diseases who will soon transition into adult care. Several national collaborators, including the Transition to Adult Care National Interest Group and the Center for Medical Home Improvement, have contributed to the success and growth of this work.



Other projects in the region, while not originally funded by the MSGRCC, build on existing collaborations initiated and sustained by the MSGRCC. **The Texas Newborn Screening Performance Measures Project** (PI: Susan Tanksley, PhD), **Promoting Public Dialogue on the Use of Residual Newborn Screening Samples** (PI: Jeffrey Botkin, MD), and the **Utah Newborn Screening Clinical Health Information Exchange** (PI: Richard S. Harward, AuD) all depend on and are strengthened by established MSGRCC partnerships.

<http://www.msgrcc.org/>



Submitted by Jacquie Stock, MPH, Project Evaluator; Kerry Silvey, MS, CGC, Project Co-Director; Lianne Hasegawa, MS, CGC, Project Coordinator; Arthur Yu, MS, CGC, Genetics Project Specialist and Sylvia Au, MS, CGC, Project Co-Director, WSGSC

Stakeholder Perceptions of Regional Collaborative Activities

The Western States Genetic Services Collaborative (WSGSC) includes stakeholders from each of its state and territory’s public health genetics and newborn screening programs, family advocates, medical geneticists, genetic counselors, primary care providers, and others working in genetic services or newborn screening. A Regional Genetics Plan, developed by these stakeholders, includes ongoing evaluation of stakeholder opinions regarding the functioning and effectiveness of the Collaborative. To this end, 51 stakeholders who attended either the

2008 or 2009 annual WSGSC Regional Summit were e-mailed a request to complete an online, self-administered questionnaire. The survey was written by the WSGSC Co-Directors and staff, and was designed to measure stakeholder opinions on the communication of the Collaborative, as well as activities related to financing, quality improvement and access to genetic services. The questionnaire also included an open-ended response option seeking qualitative input on WSGSC activities. The survey had 25 respondents (response rate=57%).

Washington State had the largest percent of stakeholders completing the survey (N=6), while public health genetics and newborn screening representatives together comprised the largest cohort of respondent types, followed by clinicians.

Knowledge and Information Sharing in the Collaborative

The WSGSC considers knowledge and information sharing as a key goal. As shown in Table 1, 6 out of 17 of the survey statements relating to knowledge

Knowledge and Information	No.	Mean	Standard Deviation
My opportunities to receive genetics information have increased because of WSGSC activities.	25	4.08	1.18
My opportunities to share genetics information have increased because of WSGSC activities.	25	3.84	1.14
WSGSC activities have helped me to routinely exchange genetics information with public health genetics representatives.	25	3.46	1.18
WSGSC activities have helped me to routinely exchange genetics information with family representatives.	25	3.12	1.33
WSGSC activities have helped me to routinely exchange genetics information with medical geneticists.	25	3.44	1.26
WSGSC activities have helped me to routinely exchange genetics information with genetic counselors.	25	3.20	1.12
I often read WSGSC WIKI e-mail notices.	25	3.80	1.26
I often read WSGSC e-mail updates.	24	4.08	.97
I often participate in WSGSC sharing calls.	24	3.42	1.38
I often participate in WSGSC work group telephone conferences.	24	3.17	1.37
WSGSC WIKI e-mail updates are useful.	24	3.86	.79
WSGSC e-mail notices are useful.	25	4.00	.78
WSGSC sharing calls are useful.	25	3.81	.98
WSGSC work group telephone conferences are useful.	24	3.60	1.14
WSGSC communications help me understand successful ways to collaborate within the western region.	25	4.00	.82
WSGSC communications help me understand challenges to collaboration within the western region.	25	4.16	.94
WSGSC activities help increase my awareness of various people or groups who have a stake or interest in genetics in the western region.	24	4.30	.81

Table 1: Mean responses to WSGSC stakeholder on-line survey statements about knowledge and information sharing in the regional collaborative (5=agree, 1=disagree)

Financing Genetic Services and Newborn Screening	No.	Mean	Standard Deviation
Information provided at the 2009 WSGSC Summit increased my understanding of barriers to reimbursement for cognitive genetic services.	22	4.04	1.30
Information provided at the 2009 WSGSC Summit increased my understanding of solutions to reimbursement for cognitive genetic services.	21	3.80	.87
WSGSC activities increased my understanding of financing of genetic services.	22	4.00	.8729
WSGSC activities increased my understanding of financing for newborn screening.	22	3.60	1.22
Health insurance reimbursement for genetic services has improved in my state as a result of WSGSC activities.	12	3.59	1.62
Financing for newborn screening in my state has improved as a result of WSGSC activities.	15	3.13	1.68

Table 2: Mean responses to WSGSC stakeholder on-line survey statements about financing genetic services and newborn screening in the regional collaborative (5=agree, 1=disagree)

and information sharing in the Collaborative had a mean response of at least 4.0 or higher (5=agree, 1=disagree). The remaining 11 statements had a mean response between 3.17 and 3.86.

Financing Genetic Services

Fewer respondents completed the survey statements about WSGSC's activities to increase understanding of the financing of genetic services in the region. As shown in Table 2, the WSGSC had the highest positive response related to information about barriers to reimbursement for cognitive genetic services and understanding of the financing of genetic services.

Quality Improvement in Genetic Services

An article describing the *Defining Genetic Services Framework and Outcomes for Genetic Services Menu* was published in the *American Journal of Medical Genetics*.¹ Among the respondents who rated the usefulness of information in this article, 63% assigned a ranking of 4 or 5 to a statement indicating the article was useful to them. 35% of 20 respondents indicated that they had used the *Defining Genetic Services Framework*, while 28% of respondents assigned a rank of 4 or 5 to a statement indicating that they had used the *Outcomes of Genetic Services Menu*.

Access to Clinical Genetic Services

Testing use of a regional model to enhance access to genetic services is also a goal of the WSGSC. 53% of respondents agreed with the statement that, "Access to clinical genetic services has improved in my state as a result of WSGSC activities and projects." 73% gave a ranking of 4 or 5 to a statement indicating WSGSC activities helped them understand barriers to providing clinical genetic services to people who live far from tertiary centers.

Discussion

Information dissemination within the WSGSC seems to be the most appreciated function among survey respondents, with informational e-mail notices being the most highly rated. Information dissemination about financing and increasing access to clinical genetic services was ranked high among most respondents, while Collaborative impact on actual financing for genetic services and newborn screening was ranked lower. The same relationship was true for responses related to access to clinical genetic services. Fewer respondents reported finding WSGSC products designed to measure quality improvement useful. Survey responses may reflect the fact that it takes more time to implement effective regional solutions to access and financing than it does to lay the groundwork for understanding the challenges facing the Collaborative in doing so. Strong family participation

in both the survey and the Collaborative may indicate family commitment to and comfort level working with the WSGSC.

<http://www.westernstatesgenetics.org/>

Reference

¹Silvey K, Stock J, Hasegawa LE, Au SM. Outcomes of genetics services: creating an inclusive definition and outcomes menu for public health and clinical genetics services. *Am J Med Genet C Semin Med Genet*. 2009 Aug 15;151C(3):207-13.

The Advocates Partnership Program: Connecting Consumer Advocates with the Regional Collaboratives, Clinical Geneticists, and ACMG Leaders

Submitted by Andria M. Cornell, Advocacy and Health Policy Coordinator, Genetic Alliance



In March 2010, for the fifth consecutive year, the American College of Medical Genetics (ACMG) teamed up with Genetic Alliance to sponsor the Advocates Partnership Program at ACMG's Clinical Genetics Conference. The Advocates Partnership Program is specifically designed to provide leaders of genetic advocacy organizations with an opportunity to connect with genetics professionals and the ACMG leadership. It offers participants an unparalleled opportunity to network with other advocates and with genetics professionals from around the world. Funding for the program is provided by the National Coordinating Center (NCC) for the Regional Genetic and Newborn Screening Service Collaboratives—housed at ACMG and supported by the Genetic Services Branch in HRSA's Maternal and Child Health Bureau—and by Genetic Alliance. The NCC provides registration and travel support and Genetic Alliance is responsible for all program logistics and offers a stipend for participants. Several Regional Collaboratives matched the stipends provided by Genetic Alliance for the advocates from their regions.

Eight representatives of advocacy organizations participated in the Advocates Partnership Program at the ACMG Clinical Genetics Conference, held this year in Albuquerque, New Mexico. The diversity of the participants—including their conditions of interest, the communities they represented, and their engagement in local, state, national, and international initiatives—contributed to the richness of the discussions and the shared learning throughout the program. Additionally, four family members (including a 17-year-old daughter with interest in the field of genetics, a parent, and two spouses) accompanied advocates to share in the significance of the experience and offer support.

The first day of the Advocates Partnership Program began with introductions, an overview of the ACMG conference highlights, and strategies for successfully getting the most out of the sessions. Genetic Alliance staff and ACMG faculty shared what they were most excited about for the conference and, guided by the interests of the advocates, discussed sessions and workshops in detail and provid-

ed insider tips for networking at the meeting. Later in the evening, over an informal dinner, participants shared brochures from their organizations, their personal stories, and what they hoped to come away with after the five-day conference.

One advocate wrote: *"The question I had hoped to answer in respect to participating in the program was, 'will the current study of genetics make a difference in my consumer advocacy work and the cause I serve?' In particular for people with intellectual and/or other developmental disabilities and their families there is, perhaps, an even more emotionally charged underlying question: 'do you finally understand why me or my child is challenged?' I found the answer to the first question to be a resounding YES, and the answer to the second question to be essentially the reason we were all in the program."*

Throughout the conference, the Advocates Partnership Program offered exclusive daily briefings with ACMG faculty, the HRSA Genetics Collaboratives' leadership, and a wide range of clinical genetics professionals. Discussion in these one-hour meetings was often prompted by questions from the advocates in response to presentations they attended or conversations they joined or overheard. Guest speakers answered questions, provided insights from their personal experiences, and facilitated discussion about how advocates' work can impact genetics professionals—particularly in the Regional Collaboratives. The advocates found these lively and thought-provoking discussions energizing and empowering and reported that their perspectives and input were highly valued by the guest speakers. They described the speakers as "caring," "warm," and "really wanting to make a difference for

families." One advocate wrote: *"I was impressed with the speakers...in addition to being knowledgeable about his/her particular study or field, it seemed as if our dialogues were genuine and open. Additionally, I didn't have the impression that the speakers were guarded in communicating."*

Themes that arose in the discussions included:

- The importance of case management for families;
- What it feels like to know more than some professionals do about the health condition your child has been diagnosed with; and
- The relationship between the disability community and the genetics and newborn screening communities.

One discussion session focused on the importance of cultural competency training for professionals, with a Navajo participant sharing his experiences with a provider around his daughter's diagnosis. Participants noted on numerous occasions their surprise at the commonalities in gaps and needs that were identified across conditions.

Many advocates stated that one of the beneficial outcomes of participating in the Advocates Partnership Program was networking with meeting attendees. Advocates met professionals working on their condition of interest or representatives from industry developing a treatment for their condition of interest, and they shared information with them about their organization's initiatives. Having a forum to share and explore together, however, proved to be most valuable. In response to a question about preparing for participating in the Advocates Partnership Program, distributed at the end of the conference, one participant wrote, "I

don't think I could have prepared for just how helpful the informal sessions would be."

At the conclusion of the conference, the advocates came away with ideas for new collaborations and outlets for their energy. One advocate said, *"I was quite charged about changes we need to make at my nonprofit to incorporate what I learned!"* In response to learning about advancements in the genetics for her particular condition of interest, another advocate shared, *"I will go home to encourage our sickle cell families to become better advocates. We will work with them to know more about their disease and get involved with all the new changes."* All advocates felt that attending the ACMG Clinical Genetics Conference and participating in the Advocates Partnership Program made them want to keep learning more.

The Advocates Partnership Program at the ACMG Clinical Genetics Conference is made possible by the commitment of the HRSA Genetics Collaboratives and the NCC to engage consumers as partners in the advancement of genetics and newborn screening. The collaborations and ideas generated by the program enhance family and provider knowledge and awareness of key issues and challenges. At the same time, Genetic Alliance continues to engage the disease-specific organizations and the general public in the development of education and communication strategies and tools on a national level, while the HRSA Genetics Collaboratives provide an ideal setting for bringing ideas, information and services to local communities.

2010 ACMG Advocates Partnership Program

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Susan Atkins

State Coordinator and Parent
Washington State Parent to Parent
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Carol Barton

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The Genetics/Genomics Competency Center

Submitted by Jean Jenkins, PhD, RN, FAAN, Senior Clinical Advisor to the Director, National Human Genome Research Institute, NIH

Increasingly, professional groups and state, federal, and academic organizations are recognizing the need to provide genomic education to health professionals that is appropriate to all levels of learners. As noted by Dr. Greg Feero in the April 2010 *NCC Collaborator* newsletter, "Genomic medicine's full potential to improve healthcare in the United States will take considerable continued investment in biomedical research and changes in how we educate health professionals." In many ways the Internet is the most logical venue for the development, storage, and dissemination of educational resources for health professionals and their teachers. Recognizing this, the National Human Genome Research Institute (NHGRI), part of the National Institutes of Health (NIH), has launched the Genetics/Genomics Competency Center, a free, web-based repository of curricular materials on genetics and genomics designed for nurses and physician assistants.

The Genetics/Genomics Competency

To encourage sharing and avoid duplicative efforts across healthcare disciplines, [G2C2] helps match existing educational resources with educational competencies for health professionals.

Center (G2C2) was developed by the University of Virginia in Charlottesville with NHGRI funding and the guidance of an advisory group representing a wide range of research and



professional organizations. These organizations included the American Academy of Physician Assistants, the American Association Colleges of Nursing, National Cancer Institute (NIH), National Coalition for Health Professional Education in Genetics, National League for Nursing, National Society of Genetic Counselors, Physician Assistant Education Association, and Sigma Theta Tau International.

This new resource was first announced at the 2010 American Association of Colleges of Nursing Master's Education Conference in New Orleans in late February of this year. In the future, the developers hope to expand this tool to include other healthcare professions, such as genetic counselors, pharmacists, and physicians. Immediate next steps for G2C2 include creation of a plan by genetic counselors to add their competencies and resources on this site.

To encourage sharing and avoid duplicative efforts across healthcare disciplines, the Genetics/Genomics Competency Center helps match existing educational resources with educational competencies for health professionals. The resource accomplishes this through sophisticated, cross mapping of learning activities and assessments, outcome indicators and professional competencies, such as *Genomics Nursing: Competencies, Curricula Guidelines and Outcome Indicators*, <http://tiny.cc/1ikoe>, and similar guidelines for physician assistant education, <http://tiny.cc/mqzff>.

Educators can use G2C2 to find and download materials. They also can share their favorite genomic and genetic teaching resources with other educators by uploading material. The center's editorial board reviews uploaded material regularly to ensure quality. A rating system will soon be implemented to help the busy educator quickly identify the most valuable resources. Visit the rapidly evolving G2C2 website at <http://www.g-2-c-2.org> to learn more and provide feedback about the site's look and value by clicking on the survey link (under the G2C2 survey box) posted to the right side of the home page.

G2C2 will be an invaluable tool for the Regional Genetics Collaboratives in their ongoing efforts to provide genetics education to healthcare professionals in their regions.

The views of the author are her own and are not necessarily those of the NHGRI, NIH or the US Department of Health and Human Services.

NCC MEETINGS

ACT Sheet Workgroup Meeting	July 28-29	San Francisco, CA
Face-to-Face PD Meeting	Nov 18-19	Bethesda, MD

RC MEETINGS

Mountain States Genetics Regional Collaborative Center (MSGRCC) Annual Meeting	Jul 13-15	Denver, CO
Southeast Newborn Screening and Genetics Collaborative (SERC/SERGG) Annual Meetings	Jul 22-24	Ponte Vedra Beach, FL
New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services (NYMAC) Emergency Preparedness Seminar/Table-Top Drill	Aug 6	Baltimore, MD
Heartland Regional Genetics and Newborn Screening Collaborative (Heartland RC) Annual Meeting	Sep 22-24	Des Moines, IA
Western States Genetic Services Collaborative (WSGCC) Regional Summit	Sep 27-29	Seattle, WA
New England Genetics Collaborative (NEGC) Annual Meeting	Dec 1	Portsmouth, NH

ADVISORY COMMITTEE MEETINGS

Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) Meeting	Sep 16-17	Washington, DC
Secretary's Advisory Committee on Genetics, Health and Society (SACGHS) Meeting	Oct 5-6	Washington, DC

NATIONAL CONFERENCES

Training Institute: New Horizons for Systems of Care, Effective Practice and Performance for Children and Youth with Mental Health Challenges and their Families	Jul 14-18	National Harbor, MD
Genetic Alliance Annual Conference	Jul 16-18	Bethesda, MD
National Conference of State Legislatures (NCSL) Legislative Summit	Jul 25-28	Louisville, KY

NATIONAL CONFERENCES

National Coalition for Health Professional Education in Genetics (NCHPEG) Annual Meeting: Genetics and Public Health	Sep 23-24	Bethesda, MD
American Academy of Pediatrics (AAP) National Conference and Exhibition	Oct 2-5	San Francisco, CA
Southwest Conference on Disability	Oct 6-8	Albuquerque, NM
National Society of Genetic Counselors (NSGC) Annual Education Conference	Oct 14-17	Dallas, TX
Maternal Child Health Bureau (MCHB/HRSA) Celebrating Title V at 75	Oct 20	Washington, DC
Association of University Centers on Disabilities (AUCD) Annual Meeting	Oct 31-Nov 3	Crystal City, VA
American Society of Human Genetics Annual Meeting	Nov 2-6	Washington, DC
American Public Health Association Annual Meeting	Nov 6-10	Denver, CO
4th National Conference on Genomics and Public Health	Dec 8-10	Bethesda, MD



June 2010 Edition Volume 4, Number 2

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The NCC Collaborator is the official newsletter of the National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives (NCC). The newsletter is published quarterly and is available in a portable document file (PDF) via email and at www.nccrcg.org. Copyright © 2010 American College of Medical Genetics. The National Coordinating Center is funded by cooperative agreement No. U22MC03957 between the American College of Medical Genetics and the Genetic Services Branch/Maternal and Child Health Bureau/Health Resources and Services Administration.

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