NCC COLLABORATOR

Working Together to Enhance Genetic Service Delivery

February 2014 Edition, Volume 8, Number 1

# New and Unique Approaches to Expanding Genetic Services

Since the inception of the NCC/RC system, each of the seven HRSA Genetic Collaboratives has consistently found unique ways to accomplish the common goals of the system as a whole. One key approach has been to support "incubator" projects that respond to particular needs of the region, but, if proved effective, can be modified for, disseminated to, and replicated by other RCs. This edition of the *NCC Collaborator* celebrates some regional projects that are just getting underway and others that are expanding to build on past successes. Through these activities and others, the RCs are continually striving toward enhancing the delivery of genetic services both in their own regions and across the country.

From NEGC you'll read about the expansion of a patient navigation project to include children with sickle cell disease (SCD), a new project that looks at parent psychosocial response to critical congenital heart disease (CCHD) screening, and the efforts by its Transition Workgroup to engage the mitochondrial disease community in its activities.

NYMAC continues to support small projects that encourage the implementation of distance strategies to improve access to care. It is exploring the use of distance strategies in urban, as well as in the more typical rural, settings.

SERC has launched its SECRET transition project. Despite its name, this project doesn't require security clearance--merely a strong desire to advocate for successful medical transition for individuals with genetic conditions. This project has its basis in the successful Atlanta Sickle Cell Disease Transition Program.

Region 4 Midwest maintains its ongoing interest in improving the collection and utilization of data on individuals with genetic conditions. In this edition, it shares its quality improvement tool for the care of children with sickle cell disease in emergency departments, which was developed in collaboration with Duke University.

Heartland recognizes that an individualized health care plan is as important in the school setting as it is in the health care setting. Its new project to address barriers to care for Hispanic families is responding to the dramatic increase in the Hispanic population in this region during the past decade.

MSGRC, through its co-Director, Dr. Kathy Hassell, recently initiated a hemoglobinopathies long-term follow up mini-project to understand who is delivering care to individuals with hemoglobin disorders across their lifespans and what the needs of these practitioners are.

Western States leads the NCC/RC system in providing a comprehensive, easy-to-use, family-friendly web resource on the Affordable Care Act. It has created an interactive platform modeled on a game board in order to simplify this complex subject and engage users in learning how the ACA will affect individuals and families with genetic conditions.

NGECN introduces a new set of online resources for consumers. Through *GenesInLife.org*, Genetic Alliance is focusing on providing accurate, relevant information for individuals on genetics, health, and family history.

One of NCC's national partners, NewSTEPs, highlights some of its first year accomplishments, including making its case definitions available on the NewSTEPS website that debuted this fall. Another partner, AAP, shares information regarding its Genetics in Primary Care Institute (GPCI) Quality Improvement Project, which focuses on enhancing the delivery of genetic medicine in primary care settings.

All of these projects provide exciting opportunities for the NCC/RC system and the larger genetics community. We will all be watching to see which projects have accomplished their goals and which are ripe for expansion.

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NCC National Coordinating Center for the Regional Genetic Service Collaboratives

THE NEW ENGLAND
GENETICS COLLABORATIVE

# NEGC's New Partnerships to Improve Patient Care and Education

Submitted by
Karen Smith,
Project Coordinator, NEGC

The New England Genetics Collaborative (NEGC) is engaging a number of new partners in order to improve care for patients with genetic conditions and to better educate them and their families. First, NEGC has partnered with the Department of Pediatrics at Boston Medical Center (BMC) to expand its existing program of patient navigation services to include children with sickle cell disease (SCD). A new patient navigator will be hired and, once trained, will act as liaison and coordinator of care for SCD patients. This will include:

1) Assisting in arranging health care appointments, genetic counseling, and emotional support for parents of infants with SCD at the time of diagnosis, as appropriate;

2) Linking patients, especially adults, with SCD to primary care providers;

3) Providing the appropriate monitoring for initiation of hydroxyurea by adult SCD patients and for adherence to the hydroxyurea regimen;

4) Assessing adherence to routine SCD clinic and specialist appointments by patients with SCD making the transition to adult care;

5) Ensuring that children with SCD who have an abnormal stroke screening test (Transcranial Doppler, or TCD) have appropriate repeat testing and further treatment, as needed; and

6) Identifying barriers to treatment for both children and adults with SCD and developing remedies for them.

Another NEGC partner, Joanna Fanos, PhD, will work to assess parental psychosocial response to Critical Congenital Heart Disease (CCHD) screening. To do so, she will interview 25 families of infants who were screened positive for CCHD, using a semi-structured interview guide. She will also develop a Knowledge and Attitudes Questionnaires for parents. Results from this study will provide information about how families respond to receiving news that their newborn has screened positive for CCHD and how their responses vary depending on whether that screen was a true positive (CCHD diagnosis) or a false positive (no CCHD, but perhaps another health issue).

Finally, the NEGC Transition Workgroup will engage new partners in the mitochondrial disease community. Planned activities include development of a one-page fact sheet for young adults with mitochondrial disease and of an Educator's Guide to Mitochondrial Disease, through a partnership with the Emerson College Health Communication Program. The Education Guide may later be adapted for the GEMSS (Genetics Education Materials for School Success) website. These efforts will incorporate an important lesson learned during a special session at the Face Forward Conference in July 2013, thanks in large part to the contributions of a young person with mitochondrial disease. The lesson? Difficult conversations with parents (on subjects such as palliative care) need to be part of transition planning.

# NEW YORK-MID-ATLANTIC CONSORTIUM FOR GENETICS AND NEWBORN SCREENING SERVICES

## NYMAC To Begin Using Distance Strategies for Improving Care

Submitted by
Katharine B. Harris, MBA,
Project Manager, NYMAC

All of us at the New York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC) are excited about our new forays into the use of distance strategies to help provide more consistent care for people with special health needs in our region. Included in our definition of "distance strategies" is, of course, traditional telemedicine using equipment that visually links a patient at a spoke site to a specialist at a hub site. One of our funded contractors, the Sickle Cell Program (directed by Dr. Lakshmanan Krishnamurti) at the Children's Hospital of Pittsburgh of the University of Pittsburgh Medical Center (UPMC), is using existing equipment at UPMC and distance sites to reach sickle cell patients living in the counties of Erie, Butler, and Cambria. This strategy will enable eligible patients to take advantage of hydroxyurea therapy, because Dr. Krishnamurti will now be able to closely monitor their health status. Once these procedures are in place, he also plans to work with primary care providers in their offices to provide specialty care using portable technology.

Closely mirroring this strategy, Skype will be used for communication via a network of camera-enabled computers. These will be located in local primary care centers of the Hudson Headwaters Health Network in New York State's Adirondack Region and will link to the Metabolic Program at Albany Medical Center, directed by Dr. Darius Adams. While it is still in the development and approval stages, it is expected that this communication tool will allow patients living in this six-million-acre preserve ready access to needed care without having to travel many miles through the sometimes wild weather of northern New York.

NYMAC readily recognizes that long distances and mountain roads might not be the only barriers facing patients needing specialty care services. Sometimes care can be enhanced by improving communication among patients, their primary care providers, and their specialists. Dr. Thomas Moulton, a pediatric hematologist in the Bronx-Lebanon Hospital Center Sickle Cell Program, is working to accomplish this using an encrypted and secure e-mail network of his patients and their pediatricians in the Bronx and beyond. He hopes that, by using this network, patients can receive frequent reminders about upcoming appointments and improve their show-rate. He will also be able to send clinic summaries to his patients' pediatricians--and get their notes promptly back to him.

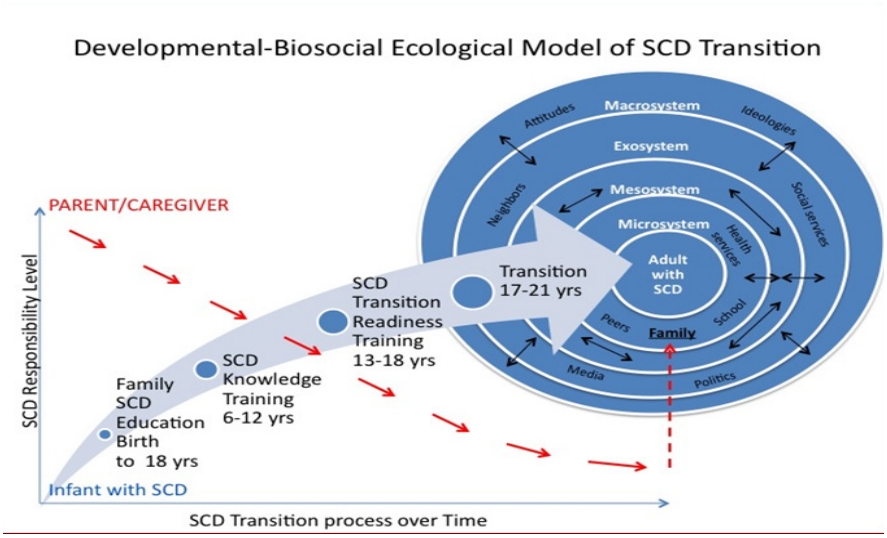
These projects are still in their developmental stages. Institutional Review Boards are reviewing the proposed technologies to ensure security of transmissions and maintenance of patient confidentiality. Stay tuned for updates.

# SOUTHEAST REGIONAL NBS & GENETICS COLLABORATIVE

## Successful Medical Transitioning:

## Addressing both Patient and Provider Needs

Submitted by
Alana Aisthorpe, BA,
Assistant Coordinator, SERC




Achieving successful medical transition is increasingly important for children with heritable disorders. With improved comprehensive care and advances in medical technology, these individuals are living significantly longer. Unfortunately, health care systems have failed to keep pace with this improved longevity, creating a chasm in care for these individuals as they transition to adulthood.


The Southeast Regional NBS & Genetics Collaborative (SERC) Transitioning Workgroup was formed to address this gap in continuity of care. Our primary goal is to educate patients, caregivers, and professionals about services to improve quality of healthcare across the lifespan. Often, the patient's priorities for transition are very different from those of his or her providers. For example, issues like obtaining and keeping a job, marriage and family life, living independently, access to quality health care, and satisfaction with the transition process may be most important to the patient, while medical indicators of health, such as medication adherence and stable markers of disease severity, may be more important to providers. The challenge of merging both providers' and patients' needs to achieve a mutually beneficial endpoint of "transition success" is the workgroup's primary goal.

The South Eastern Collaborative Regional Transition (SECRET) demonstration project addresses the need for a practical, developmentally appropriate, and socially/culturally adaptable model for transition from pediatric to adult centered medical care that meets these challenges. The project is based on the Biopsychosocial-Ecological framework of adolescent development, a theoretical model established by clinical psychologist Anya Griffin, PhD in collaboration with the Atlanta SCD Transition Program.[1] Transition planning begins at birth with the parent/caregiver and continues through childhood and adolescent development into adulthood. It involves a multidisciplinary team of providers, family, peers, staff of social services and other community agencies, and adult mentors living with SCD. The program focuses on the medical, psychosocial, educational and vocational needs of the patient, with the ultimate aim of fostering increased disease knowledge, self-efficacy and autonomy while decreasing reliance on parents/caregivers.[1]

We are now completing a regional needs assessment and putting together a cohort of evaluation tools in concert with medical and community partners from each state in SERC. During the next phase of the project, we will evaluate whether it works better to use the model in transition programs targeted at individuals with a single disease or in programs that cut across disease groups. In addition, we will develop a transition program manual to facilitate practical adaptation of this model.

Reference:
[1]Griffin et al, Journal of Clinical Pediatric Psychology (2013) 1039; RPM Article number cpp-0028 Proof# 289161 Issue 3, Vol 1 Issue date 9/1/2013 (in press)

# REGION 4 MIDWEST GENETICS COLLABORATIVE

## Region 4 Develops A Quality Improvement Tool for the Care of Children with Sickle Cell Disease in Emergency Departments

Submitted by
Kristen Hawkins, BA, Patient Coordinator, Region 4


Children who experience medical complications associated with sickle cell disease (SCD), one of the most common genetic disorders affecting African American children,[1, 2] often require evaluation and treatment in emergency department (ED) settings. However, of the 32 quality-of-care indicators recommended for children with SCD, only five are applicable to the ED setting. These indicators address the disease broadly, but not its complex management in the ED setting.

To address this issue, the Region 4 Midwest Genetics Collaborative Hemoglobinopathies Workgroup partnered with Paula Tanabe at Duke University, the developer of the Emergency Department Sickle Cell Assessment of Needs and Strengths (ED-SCANS) that is used with adult patients. This decision support tool and set of algorithms can be used as a quality improvement framework to guide the medical management of patients with SCD in the emergency department.

Under the direction of Dr. Tanabe, the Hemoglobinopathies Workgroup conducted a qualitative study to guide modifications of ED-SCANS for use with children. We conducted nine focus group and individual interviews with ED nurses, ED physicians, parents, SCD nurse practitioners, and hematologists in six states. We also partnered with the Sickle Cell Disease Association of Illinois and the Sickle Cell Disease Association of America to recruit parent participants. ED personnel and parents identified critical areas that can be used to organize and improve the assessment, management, and disposition/referral decisions in order to provide better care to children with SCD in the ED. Full descriptions of the methodology and study findings are published in the Journal of Pediatric Oncology Nursing[3].

The Family Centered Pediatric Emergency Department Sickle Cell Assessment of Needs and Strengths (FC-Peds-ED-SCANS) was the result of the regional collaborative's work with Dr. Tanabe. The FC-Peds-ED-SCANS can be used as a framework to help focus and design initiatives to improve the quality of ED care for children with SCD. It is available online using the following links: *http://region4genetics.org/docs/ED-SCANS_2013.pdf* and *http://sickleemergency.duke.edu*. Future activities will include dissemination, implementation, and evaluation of the tool.

If you have any questions about the tool, please contact Dr. Paula Tanabe, paula.tanabe@duke.edu.

Reference:
[1]Centers for Disease Control and Prevention. (2012). Sickle cell disease: Data and statistics. Retrieved from http://www. cdc.gov/ncbddd/sicklecell/data.html
[2]Hassell, K. L. (2010). Population estimates of sickle cell disease in the U.S. American Journal of Preventive Medicine, 38(4Suppl.), S512-S521.
[3]Tanabe, P., Dias, N., & Gorman, L. (2013). Care of children with sickle cell disease in the emergency department: Parent and provider perspectives inform quality improvement efforts. Journal of Pediatric Oncology Nursing, 30(4), 205-217. doi: 10.1177/1043454213493509.

# HEARTLAND GENETICS AND NEWBORN SCREENING COLLABORATIVE

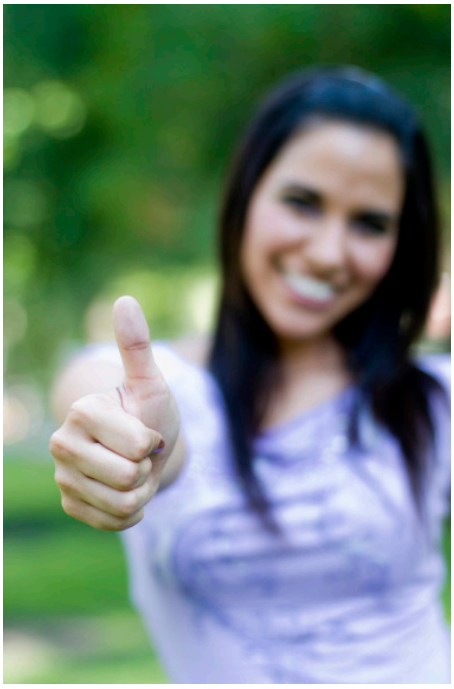
## Heartland Engages New Partners to Improve Access to High Quality, Culturally Competent Genetic Services by Hispanic Patients

Submitted by
Mary Ann Coffman, MS, CGC, Project Coordinator

According to the 2010 census, the US Hispanic population grew in every region between 2000 and 2010, but most significantly in the South and Midwest regions. The Heartland Genetics and Newborn Screening Collaborative encompasses states in both these regions. This led Heartland to ask two questions: "What are the barriers to accessing genetic services for Hispanic patients?" and "What can genetic service providers do to provide culturally competent quality care for Hispanic patients?" To answer these questions, Heartland realized that it needed to engage new partners in its work.

Since May 2013, approximately 20 individuals have been contacted in four Heartland states to begin to identify the key issues regarding access and provision of culturally competent genetic services. These individuals include genetic service providers, primary care providers serving Hispanic children, and outreach coordinators from the states' Children and Youth with Special Health Care Needs (CYSHCN) family support systems who also serve Hispanic families. The outreach coordinators, particularly those who are Hispanic themselves, responded quickly and positively, expressing a high level of interest in collaborating with Heartland in any capacity that would be helpful. Many invited the Project Coordinator (who is a bilingual Hispanic) to attend their meetings, organize focus groups, and give presentations. Based on their own experience and/or those of other Hispanic families with whom they are working, they were able to readily identify issues regarding access to genetics and other specialty services. These issues included financial barriers, such as lack of transportation and cost of services, language barriers, cultural barriers, and fear of immigration services.

The statement, "We are about relationships," was repeated by several of the Hispanic outreach coordinators. It refers to a cultural norm, but was also an invitation to Heartland to reach out in partnership (relationship) to the Hispanic families with genetic conditions. The other message we heard, loud and clear, from several of the outreach coordinators and from the service providers? "There is a need for more bilingual staff and literature at the clinics."



An *ad hoc* committee of clinical service providers, Hispanic families with genetic conditions, and outreach coordinators has been formed. The family support systems that are being invited to participate are the Oklahoma Family Network, the Family-2-Family Health Information Center of Arkansas, Parent Training and Information (PTI) Nebraska, and Families Together, Inc. of Kansas. The primary goal of this committee is to build partnerships and provide recommendations on the development of interview questions in Spanish related to access to genetic services for Hispanic families with genetic conditions. Currently the interview questions are being revised and a research proposal is being developed for submission to the Institutional Review Board.

# MOUNTAIN STATES GENETICS REGIONAL COLLABORATIVE

## MSGRC Launches Program to Improve Follow-Up for Individuals with Sickle Cell Disease

Submitted by
Celia Kaye, MD, PhD, Project Director; Kathryn Hassell, MD, Associate Project Director; Liza Creel, Former Project Manager; Joyce Hooker, Director of Regional Outreach; Donna Williams, Project Coordinator, MSGRC and Camille Miller, Project Administrator, MSGRC

The Mountain States Region (Arizona, Colorado, Montana, Nevada, New Mexico, Texas, Utah, and Wyoming) is pleased to announce the launch of the Hemoglobinopathies Long-term Follow-up (LTFU) Mini-Project under the leadership of Dr. Kathryn Hassell. The overall goal of the project is to better understand who is delivering care to children and adults with sickle cell disease in the region and what the needs of these practitioners are. During the project's initial phase, staff will identify the medical homes to which newborns in each state are referred. These sites of care will be targeted for dissemination of the National Heart Lung and Blood Institute's (NHLBI) report on sickle cell disease management, which is expected to be released in the near future.

Following preliminary information collected by Dr. Sharon Homan, MSGRC Evaluator, and her team of representatives from each of the region's state newborn screening follow-up programs, members of the Hemoglobinopathies Interest Group will obtain and collate the specific elements of information collected by state programs. This information will eventually be used as the basis for characterizing the capacity for longer-term follow-up in all the states in our region. Project staff will be reviewing the activities of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children's (SACHDNC) Treatment and Follow-up Subcommittee, specifically its use of sickle cell disease as a use case for characterizing the most important processes and outcomes for LTFU of newborn screening. In addition, they will gather basic information regarding the NewSteps database and its potential for providing an LTFU database. Project staff also plan to review the publication from the Centers for Disease Control and Prevention's Registry and Surveillance System for Hemoglobinopathies that describes approaches to surveillance in hemoglobinopathies and to discuss with project members how to identify potential collaborators on surveillance activities within their states. Finally, the project will be identifying pediatric and adult providers and community support groups who may be familiar with sites of adult care in their states.

A number of other project activities are already underway. Recently, physicians recruited by MSGRC participated in developing and reviewing the NBS case definitions for hemoglobinopathies, a key component of future data collection. Dr. Hassan Yaish (Utah) and Dr. Melissa Frei-Jones (Texas) are currently preparing sickle cell disease-specific materials for the Medical Home Portal, a national resource organized by Dr. Chuck Norlin in Utah. This material will incorporate and augment the NHLBI guidelines, once they are available. The Hemoglobinopathies Interest Group, in collaboration with regional sickle cell providers from each state, is also devising sickle cell disease care templates for MSGRC's recently funded Affordable Care Act (ACA) project.

For more information on this project contact Dr. Kathryn Hassell at the University Of Colorado School Of Medicine, Hematology Division at 303-724-4086.

# WESTERN STATES GENETIC SERVICES COLLABORATIVE

## In With the New: New Project Activities in the WSGSC

Submitted by
Lianne Hasegawa, MS, CGC, Project Coordinator; Sylvia Mann, MS, CGC, Project Director

The Affordable Care Act (ACA) is a complex piece of legislation, and many of the details of its implementation are still being worked out by the various federal agencies involved. Even now, three years post-enactment, most of the general public readily admit they do not understand how the ACA will affect them. The Western States Genetic Services Collaborative (WSGSC) has identified a pressing need to make straightforward information about the ACA available for the families we serve.

With the strong support and urging of our Collaborative partners, WSGSC has developed a website that highlights useful ACA information and resources in a format that is easy to understand and navigate. Using the Health Resources and Services Administration's (HRSA) "through the life course" approach and drawing inspiration from Milton Bradley's Game of Life, our ACA website resembles a board game with a circular path containing thirteen boxes. Each box in our online "path of life" represents a life event, such as a pregnancy or losing a job, and each box is linked to ACA information specific to that life event. Within the middle of the path of life is a town square filled with buildings such as a police station, marketplace, and library. Each of these buildings is also linked to appropriate ACA information. For example, the marketplace building is linked to information about health insurance exchanges, which are also known as health insurance marketplaces.

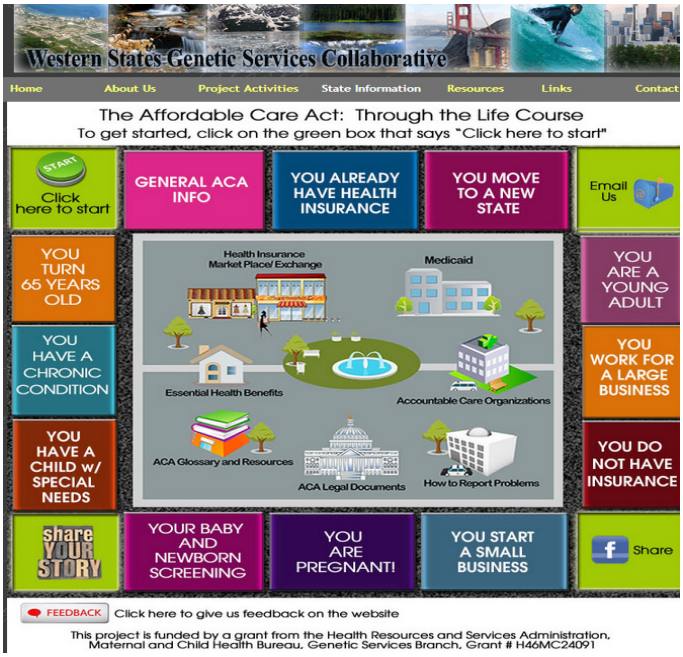
To help make the information for each life event easier to understand, we have linked important or technical terms, such as "pre-existing condition" and "grandfathered," to a glossary. Users are also able to find additional information by going to the resources listed at the end of the section.

Although some of the website content is original, we drew readily from several wonderful existing online resources, including the federal HealthCare.gov website and HRSA funded Catalyst Center, and have linked to or summarized information from these sites.

We have completed testing and launched the website. We held focus groups with stakeholders (Title V programs, family advocates, and health care providers) from our region, and improved the site based on their comments. The website became available to the public in October 2013, just in time for the opening of the health insurance exchanges. Our initial target audiences will be the state Title V programs and Family to Family Health Information Centers since they do the broadest outreach to families that may benefit from the website.

We realize the importance of public education as part of ACA implementation activities and hope our website will help with these efforts. The WSGSC will continue to update the website, and we hope that it will become a valuable resource for people in all of the Regional Genetics Collaboratives.

Visit the new Affordable Care Act Through the Life Course Website at: *http://www.westernstatesgenetics.org/ACA_home.htm.*



# NATIONAL GENETICS EDUCATION AND CONSUMER NETWORK

## Genetic Alliance Introduces New Online Resources for Consumers

Submitted by
Sharon Romelczyk, MPA, Program Manager; Rachel Koren, Programs Assistant

We live in an age of technology, a time when, even before we walk out our doors in the morning, we can know which roads have the worst traffic, whether the skies will be gray or sunny, and what important events occurred next door and halfway across the globe as we slept. In such a world, it is no surprise that, when it comes to our health, we have also come to rely more and more on technology for sharing and receiving information. And, now that there is an increasing focus nationwide on the role of genetics in health, more people are turning to the Internet to search specifically for information on genetics and genetic services.

There are thousands of websites that provide information on treatment and emotional support for individuals with genetic conditions and their family members, but important gaps remain in the information these individuals can access online.[1] We at Genetic Alliance believe that online resources must provide more information about screening and preventive care in order to effectively serve consumers, their families, and their caregivers. We say that these individuals use the Internet for health-related searches far more than the average adult, but in truth, they are average adults. These individuals are husbands and wives and parents and employees and bill payers. They are invested in their own health and the health of their loved ones, and the more information that is put in the hands of invested users, the more it will spread and the more helpful it will be.

Health information should empower individuals to take care of themselves, but also to take the initiative to address the health needs of their loved ones. In a study conducted by PEW Research Center in 2011, half of Internet users reported that their last health search was made on behalf of someone else. If we give those seeking information online the resources to protect the health of their families and loved ones as well as themselves, this information can have a much larger impact.

With this in mind, Genetic Alliance has launched an online resource for individuals and families that provides accurate, up-to-date information on the whole process of integrating genetics and health, from prevention and screening to diagnosis and treatment. *GenesInLife.org* specifically addresses issues such as heredity, how and why to collect a family health history. and the differences between various types of genetic testing and services. The website provides a comprehensive overview of genetics information and explains the importance of genetics in health. It also includes resources on facing a diagnosis and participating in research, including helpful information on which genetics professionals to consult. All of this information is designed to be easy to understand and apply. Finally, *GenesInLife.org* provides a comprehensive glossary of terms.

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*GenesinLife.org* was created by Genetic Alliance, a nonprofit organization, with funding from the Leona M. and Harry B. Helmsley Charitable Trust and the Maternal and Child Health Bureau of the Health Resources and Services Administration. *Genes in Life.org* is part of a larger initiative led by Genetic Alliance to create a network of partnerships and tools for improving consumers' access to education about genetic services. This initiative, the National Genetics Education and Consumer Network (NGECN), is a three-year program focused on consumer education and engagement in conjunction with the National Coordinating Center for the Regional Genetic Service Collaboratives.

Reference:
[1]Susannah Fox. "Health Topics." Pew Research Center, Washington, D.C. (February 1, 2011) http://pewinternet.org/Reports/2011/HealthTopics.aspx, July 29, 2013.

# NewSTEPs Makes Major Strides Forward

Submitted by
Sikha Singh, MHS, PMP,
Manager, APHL

The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs) wrapped up its first year of activities during 2013. We are excited to have gotten off to a great start in implementing quality improvement initiatives for newborn screening systems. Several major projects are underway and are described below. We look forward to continuing to offer technical assistance and educational resources to the community and to working with the NCC/RC system in the coming year.

Our first year's key activities include the following:

Stakeholder and Partner Collaborations: NewSTEPs had a strong presence at each of the Regional Collaborative meetings during the past year. Additionally, NewSTEPs coordinated small workgroups and engaged the community in its ongoing activities through surveys, discussions, focus groups, and the NewSTEPs listserv.

Case Definitions: NewSTEPs continues to work with the Health Resources and Services Administration (HRSA) and the medical sub-speciality community to develop, refine, pilot test, and implement newborn screening surveillance case definitions. The purpose of this activity is to better estimate the true prevalence and incidence of disorders, as well as to compare outcomes across states. This will establish a foundation for effective long-term follow up. Case definitions became available in the NewSTEPs data repository during the Fall 2013.

Quality Indicators: Newborn screening quality indicators designed to evaluate program quality and measure improvement were developed and extensively vetted by newborn screening stakeholders. These eight quality indicators will be used to provide longitudinal comparisons within a program and comparisons across programs in an aggregate manner. Additionally, the quality indicators will be captured within the NewSTEPs Data Repository. These indicators will contribute to the content of Annual Reports, summarizing trends in newborn screening from a national, regional, and state level. These Annual Reports will be provided to newborn screening programs to allow opportunities for benchmarking and quality improvement. States will only be identified in their own reports.

Evaluation Site Visits: NewSTEPs provides up to three evaluation site visits each year to newborn screening programs. The non-regulatory evaluation site visit is aimed at assessing various components of a newborn screening program, including the laboratory system, birth facilities, and follow-up system, for quality improvement purposes. The comprehensive evaluation site visit is facilitated by a team of experts who evaluate programs in a customizable manner, with a focus on state legislation and policy, ethics, funding models, organizational structure, point of care testing, education, and more. The site visit evaluation team utilizes an evaluation tool developed by the NewSTEPs Steering Committee and Workgroups, building from existing resources such as the National Newborn Screening and Global Resource Center Program Evaluation and Assessment Scheme (PEAS) and Clinical and Laboratory Standards Institute documents. The tool also benefited from the NewSTEPs Delphi community input process. The guided, comprehensive evaluations occur on a scheduled basis and are followed by written reports and recommendations.

Disease Specific Outreach and Technical Assistance: NewSTEPs staff works to facilitate monthly technical assistance calls on critical congenital heart disease activities. NewSTEPs continued to engage the NBS community and partners in further disease- and-topic-specific outreach activities, including short-term follow up and health information technology, during the Fall of 2013.

Data Repository: The NewSTEPs data repository debuted in Fall 2013 and has the ability to accept data in a secure and streamlined fashion. Ease of use and opportunities to integrate data with other data systems has been an important consideration during its development. During the past year, a robust set of data elements has been developed with community input; these elements include state profile information, case definitions, quality indicators, and baby level data. The Association of Public Health Laboratories will enter into Memoranda of Understanding with all state public health laboratories and newborn screening programs to ensure that data sharing activities are transparent and conducted with the highest levels of security and privacy protections in place. During 2013, NewSTEPs worked with the Colorado Multiple Institutional Review Board and the Office of Human Research Protection to determine the oversight needed for the repository. The comprehensive set of variables will be available for programs to run queries and generate reports with the end goals of quality improvement for newborn screening programs and better outcomes for newborns.

In the year ahead, we are looking forward to furthering NewSTEPs' progress toward achieving its mission by expanding upon our previous quality improvement efforts and by harmonizing newborn screening activities through innovation and technology.

# National Quality Improvement Initiative Improves the Integration of Family History and Genetic Services in Pediatric Primary Care

Submitted by
Natalie Mikat-Stevens, MPH, Genetics in Primary Care Institute, American Academy of Pediatrics

While genetics has long been viewed as a discipline focused on rare conditions, recent advances have highlighted the role of genetics in common conditions that are generally managed by primary care providers. While family history is considered the first genetic screen, less than a third of pediatricians routinely collect detailed histories from their patients. Changing this has been one focus of a nine-month quality improvement initiative more broadly aimed at integrating genetic services into primary care and strengthening medical homes for more than 130,000 children in 11 states. The initiative has been supported by the Genetics in Primary Care Institute (GPCI), which was established through an agreement between the American Academy of Pediatrics and the Health Resources and Services Administration's Maternal and Child Health Bureau.

Thirteen pediatric practices completed the GPCI Quality Improvement Project. These practices achieved significant improvements, including the establishment of processes for using a standardized family history tool and discussing follow-up plans with families of patients with family histories and/or clinical concerns suggestive of genetic conditions. By the end of the project, all participating physicians rated their knowledge of family history in assessing predisposition to disease as "very competent." Practices were able to select family history templates or modify their own tools to systematically collect multi-generational family histories for all their pediatric patients.

Preliminary results revealed that in 84 percent of patient charts reviewed, family histories were discussed with patients/families. Among 68 percent of the charts reviewed, where there were family histories and/or clinical concerns suggesting a genetic condition, there was documentation in the chart that a follow-up/plan of care was discussed with the patient/family. A follow-up evaluation will be conducted in Spring 2014 to assess continued improvements.

While practices will continue to make progress toward meeting their goals, the most significant result from this quality improvement initiative was the change in mindset of participating practices regarding the value of family history and genetic consultations. "As a result of this project, I have realized that unless we ask the question we may not find the answer. So going back to the foundation of medicine--the family history--we have learned that we can do a better job of including genetics in the family history," said Cynthia Nassim, MD, FAAP, of Nassim and Associates in New Albany, Ind. "We will continue to commit to making the changes in our practice, such as implementing an electronic family history tool and establishing an audit system with our family history to ensure all patients' histories are being reviewed as part of the health supervision visit."

Lessons learned, tools developed, and strategies for implementing family history collection and assessment as part of the pediatric primary care visit will be promoted widely in the coming months. For more information about the GPCI, visit: *www.geneticsinprimarycare.org.*

# NCC CALENDAR

## NCC and RC Meetings

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| Heartland Genetics and Newborn Screening Collaborative Annual Meeting | April 23-25 | Kansas City, MO |
| Region 4 Midwest Genetics Collaborative 2014 Regional Meeting | April 29-May 1 | Lansing, MI |
| NYMAC Advisory Council Meeting | March 5-6 | Baltimore, MD |
| NCC Transition and Medical Home Workgroup In-Person Meetings | March 24-25 | Nashville, TN |

## National Conferences

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| ACMG Annual Clincical Genetics Meeting | March 25-29 | Nashville, TN |
| Early Hearing Detection & Intervention Annual Meeting | April 13-15 | Jacksonville, FL |
| National Society of Genetic Counselor Annual Education Conference | Sept 17-20 | New Orleans, LA |
| American Academy of Pediatrics Annual Meeting and Exhibition | Oct 11-14 | San Diego, CA |

## Announcement

It is with great joy that the NCC welcomes Gloria Weissman back to the NCC/RC system as Editor-in-Chief of the *NCC Collaborator*. Ms. Weissman brings expertise and extensive knowledge of broader maternal and child health to the NCC/RC system, and NCC is pleased to have her at the helm of the *NCC Collaborator*.

NCC
National Coordinating Center
for the Regional Genetic Service Collaborative

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