Change is the law of life. And those who look only to the past or present are certain to miss the future.”

– John F. Kennedy

In the first year of their third funding cycle, the seven Regional Genetic Service Collaboratives (RCs) and their National Coordinating Center have made significant changes in order to respond to the shifting needs of individuals with heritable conditions, the genetic specialists who care for them, and the public health infrastructure that supports both the consumers and providers of genetic services. As they have evolved to become fully functioning, well-recognized, and valued components of public health systems at the national and regional level, the RCs and the NCC continue to promote change through effective collaborations and projects.

This issue of the NCC Collaborator welcomes two new national partners to the NCC/RC system: the National Genetics Education and Consumer Network (NGECN), a partner in the NCC housed at the Genetic Alliance; and the Newborn Screening Technical Assistance and Evaluation Program (NewSTEPS), housed at the Association for Public Health Laboratories (APHL). NGECN and NewSTEPS represent important consumer and public health lab perspectives on genetic services. In their articles, they share their broad goals and plans.

Continued on page 2
NEGC leads the NCC/RC system in pursuing projects related to healthcare financing and in collaborating with the Association of University Centers on Disability (AUCD) and the Leadership Education in Neurodevelopmental and Related Disabilities (LEND) programs in the region.

NYMAC welcomes new partners and staff, continues to provide seed money to pilot projects, is focusing on distance strategy projects in both urban and rural settings, and is expanding to embrace the federally-qualified health centers through an educational outreach endeavor.

SERC remains focused on building its consumer participation base by including consumers at every level of the RC.

Region 4 Midwest looks to care coordination as a means to engage consumers. During this year the RC has offered a pilot care coordination training curriculum for parents.

Heartland is leveraging the work of other federally-funded projects to develop a template for an individualized health plan (IHP) focused on children with heritable conditions for use in school settings.
MSGRC continues to nurture telegenetics, medical home, transition, and long-term follow-up priorities through new initiatives in hemoglobinopathies, formalizing family partnerships in medical home, and improving access to care for Native American communities in Northern Arizona.

WSGSC is supporting new activities that include: engaging companies in discussions about how to determine when genetic services and testing should be authorized; making complex genetic information understandable; and involving new genetic specialists in telehealth.

NCC continues to embrace the exciting opportunities offered by the rapid changes taking place in the field of genetics and in our healthcare system. Future issues of the NCC Collaborator will showcase the ways in which the RCs and the NCC are working to positively alter both the quality and accessibility of services for individuals and families with heritable conditions.

The Genetics in Primary Care Institute (GPCI) is pleased to announce the launch of its new website, GeneticsinPrimaryCare.org. This site houses practical tools and information for primary care providers regarding genetics and genomics, genetic testing, family history, genetic counseling, and patient communication. “As we learn more about genetics and genomics, the need for integration of those informational pieces into the routine practice of medicine is inescapable. Over the next several decades we will see primary providers called upon repeatedly to provide competent and effective interpretation of genetic information for patients and their parents,” notes Barry Thompson, M.D., ACMG’s Medical Director.

GPCI is funded through a cooperative agreement between the American Academy of Pediatrics and the Maternal and Child Health Bureau in the US Department of Health and Human Services’ Health Resources & Services Administration.
NEGC Adds Two Workgroups in the New Funding Cycle

Submitted by
Karen Smith,
Project Coordinator, NEGC

The New England Genetics Collaborative (NEGC) is very excited to embark on activities with two new workgroups: the Healthcare Access and Financing (HAF); and the NEGC/Leadership Education in Neurodevelopmental and Related Disabilities Workgroups.

Healthcare Access and Financing (HAF) Workgroup

The overarching goal of the HAF Workgroup is to improve healthcare insurance coverage and reimbursement for individuals with genetic conditions. The NEGC has partnered with the Catalyst Center at the Boston University School of Public Health on this endeavor.

In preparation for a series of activities, in the spring of 2012 NEGC initiated a conference call with the LEND Program Directors to identify work to be done and to generate interest in collaboration. A follow-up planning call was held last fall and the workgroup is implementing the next steps:

- Drafting a needs assessment for LEND programs regarding the degree to which genetic services are currently included;
- Collecting AUCD (Association of University Centers on Disabilities) genetics curriculum materials;
- Reviewing NCHPEG (National Coalition for Health Professional Education in Genetics) materials to identify those appropriate for LEND training programs; and
- Further engaging LEND programs in New England.

NEGC looks forward to providing updates on the successes of both of these workgroups in expanding access to and improving the quality of genetic services within the region.

http://www.negenetics.org
NYMAC Cues Up Capable Team to Carry Out Diverse Activities

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

Meet Our Team

The New York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC) is pleased to introduce our current leadership team and staff for the new funding cycle. Dr. Michele Caggana, the Director of the NYS Newborn Screening Program, has been NYMAC’s Project Director (PD) for several years. She was joined by Co-PD, Dr. Joann Bodurtha, a medical geneticist at Johns Hopkins Medical Institution (JHMI) in November 2011. Joann brings not only expertise in genetics, but also a long history with the LEND programs. NYMAC looks forward to developing LEND/RC collaborations that will benefit both communities.

Kunal Sanghavi, MS, a genetic counselor who joined JHMI and NYMAC last August, is our new Patient and Family Project Coordinator. He is focusing on outreach to the consumer/advocate community, as well as to genetic counseling graduate programs in the NYMAC region, fostering educational alliances between the two groups.

Suzette Pennacchio is NYMAC’s new Project Aide. All of our partners will be hearing from her about meetings, pending reports, and other project-related deliverables. Katharine Harris, MBA, remains as the Project Manager, and Bonnie Fredrick, MS, as the Project Coordinator. Susanna Ginsburg continues as NYMAC’s ever-important Evaluation Consultant.

Our RC in Action

We begin the new cycle most excited about the new Distance Strategy Projects awarded to the Pediatric Hematology Program at Bronx-Lebanon Hospital, the Comprehensive Hemoglobinopathies Program at Children’s Hospital of Pittsburgh, and the Section of Genetics and Metabolism at Albany Medical Center. Each project has proposed different mechanisms to enable their patients to receive optimal medical care without constant travel to the specialty care center.

We are continuing the three Transition Navigator grants and will expand NYMAC’s involvement in Region 4 Genetics’ Inborn Errors of Metabolism Collaborative to include additional metabolic treatment centers. We continue a focus on emergency preparedness: Several of the state newborn screening programs have successfully exchanged, tested, and reported the results on dried bloodspot specimens. We are also expanding our emergency preparedness efforts into the clinical and home settings of our population.

Another new initiative is geared towards education and outreach to federally-qualified health centers (FQHCs). NYMAC will provide educational activities to FQHC staff about the benefits and the availability of comprehensive genetic services for their patients with, or at-risk for, genetic diagnoses. Lastly, in a true collaborative sense, we are working with the other HRSA Genetics Collaboratives and the National Coordinating Center to maximize the ability of the Affordable Care Act to help people with genetic disorders access the care they need. And, to further that access, the RCs are working to apply Family Health History to family healthcare and decisions.

With a lineup of new and returning team members and a wealth of exciting programs, NYMAC anticipates advancing its important work—enhancing access to genetic services—in the new funding cycle.

http://www.wadsworth.org/newborn/nymac
Creating Solutions Through Partnerships with Consumers

Submitted by
Alana Aisthorpe, BA,
Assistant Coordinator, SERC

The Southeast Genetics Consumer Alliance provides an integral link between SERC activities and consumers. Formed during the summer of 2007, the Consumer Alliance serves a host of critical functions and develops priorities that guide its work with SERC in order to ensure that patient and family voices are heard and their needs are met.

SERC annual meetings strengthen the partnership between the RC and the Consumer Alliance by providing opportunities for interaction. Professionals share presentations, clinical issues, and research opportunities with the consumers, who in turn offer keen feedback and insights into the challenges they face on a daily basis. This unique level of consumer support and the rich opportunity for in-depth exploration of needs and issues allow SERC to create mutually beneficial solutions with, and for, the patients and families seen in genetic clinics every day. To further enhance the presence of consumers in research, SERC made it a goal to have representation from the Consumer Alliance on each of its seven workgroups: Emergency Preparedness, Long-Term Follow-up, Laboratory Performance, Hemoglobinopathies, Medical Home, Telecommunication/Telemedicine, and Transitioning.

The Genetic Alliance and American College of Medical Genetics and Genomics annual meetings also provide networking opportunities for leadership from the SERC Consumer Alliance.

NBS Connect is a special example of collaboration between SERC professionals and consumers. Currently in its pilot phase, NBS Connect is a unique tool developed in the region to link patients, families, parent organizations, professionals, and industry in one forum related to inherited metabolic disorders. Through NBS Connect, all stakeholder groups are offered solutions that meet their needs. Professionals and industry representatives are able to share clinical trial information with patients; professionals have access to de-identified patient survey data; and patients and families are able to connect with one another and access useful resources, tools, and even recipes! Furthermore, interest in this project continues to grow beyond the southeast region, and NBS Connect is gaining an international presence.

The SERC Consumer Alliance is an essential part of SERC’s website infrastructure and communication strategy. Consumers have developed a strong social media network via the SERC website, a listserv, and Facebook, all of which aid in disseminating related RC news. SERC is committed to growing its partnership with consumers, which guides RC work by prioritizing and focusing research, improving clinic experiences, promoting self-advocacy, and advancing the quality of life for patients and their families.

http://southeastgenetics.org/
Empowering Parents Through Care Coordination Training

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

Region 4 believes that care coordination is a needs-driven, team-based process that values families as essential partners. Care coordination is intended to integrate care among multiple providers, enhance the caregiving ability of the family, and maximize the potential of the child. According to a 2009 study by Dr. Renee M. Turchi and colleagues, parental report of adequate care coordination was associated with favorable family-provider relations and family/child outcomes.¹ In the study, families that reported receiving adequate care coordination had increased odds of receiving family-centered care, experiencing partnerships with professionals, and being satisfied with services. In recognition of the importance of care coordination for optimal health outcomes, Region 4 formed a workgroup and developed a parent training curriculum to increase education and access to care coordination for families of children with genetic conditions.

This Care Coordination Workgroup is comprised of primary care providers, public health professionals, genetic specialists, and parent representatives. Last year, the workgroup, with the help of the Family Stakeholder Forum, developed a care coordination training curriculum for parents. The purpose of the training is to provide parents with the skills, knowledge, and resources they need to coordinate care for children with complex needs, in partnership with a medical home. Parents of children with genetic conditions will learn advocacy skills, techniques to organize information and find reliable resources, navigation of healthcare and insurance systems, skills to increase communication and coordination between multiple providers, and methods for coping with stress.

In 2013, the Collaborative piloted the training with parents of children with genetic conditions in three states within the region. Three pilot training sessions took place: in Louisville, KY (February 25); Bloomington, MN (March 4); and Columbus, OH (March 22).

The pilot training experiences and participant evaluations will be used to assess effectiveness and improve the training program. Region 4 will then revise the curriculum and identify partners interested in working together to provide the training to other parents within the region as well as nationally. For more information, please contact the Region 4 Parent Coordinator, Kristen Hawkins at khawkins@mphi.org.

Reference:
The Heartland Advocate Workgroup identified the individualized health care plan (IHP) as a valuable, yet underutilized, tool whose broader implementation they could address. The IHP has been defined by the Kansas Guidelines for Educators, School Nurses and Parents as “a plan developed by the professional school nurse in collaboration with lawful custodians and others which outlines specific health care to be given to an individual student.” Chronic physical and mental health conditions can interfere with learning and may require nursing or health services at school. Most recent data indicate that 15.5 percent of all Children and Youth with Special Health Care Needs between ages 5-17 years miss more than 11 school days annually due to illness (http://www.childhealthdata.org/). At the present time, the IHP is not mandated by law; however, the National Association of School Nurses recommends that an IHP be developed for any student requiring medical attention during the school day. The IHP is often developed when a child has a Section 504 Plan and/or an Individualized Education Plan (IEP), and it is recommended that the IHP be embedded within either of these plans, if applicable. Anticipated benefits of an IHP include:

1) Better school attendance  
2) Improved alertness  
3) Increased physical stamina  
4) Fewer medical symptoms  
5) Increased ability to participate in school activities  
6) Fewer medical emergencies. Students, families, and school staff can all benefit from the IHP.

Previous work in Kansas found vast discrepancies across schools regarding IHP processes and protocols. The school nurse was often the sole individual responsible for developing the plan, with families generally not involved in the process. Work done by Heather Smith and Dr. Wendy Parent-Johnson, with funding from Kansas Integrated Community Systems (HRSA D-70 grant), led to the development of an IHP template. Input from families, nurses, and medical providers was incorporated into the template to assist school nurses in constructing IHPs for students with health needs. This population includes children who take medications during the school day, require health monitoring, are at risk for medical crises and require a crisis care plan, use special medical equipment while at school, or require frequent health care procedures.

Heartland’s efforts continue to build on this previous work in Kansas. To date, the template has been reviewed by Heartland Advocates for use with children with genetic conditions. A webinar held December 19, 2012 promoted the project and identified stakeholders. Learning teams met April 22 to 23, 2013 for the kickoff of the Heartland IHP Learning Collaborative. The learning collaborative process will not only support local implementation of the IHP, but also inform the development of a train-the-trainer curriculum and provide data necessary for policy change.

http://www.heartlandcollaborative.org/
The Mountain States Region (Arizona, Colorado, Montana, Nevada, New Mexico, Texas, Utah and Wyoming) is challenged with delivering genetic services over large distances and to unique populations of Americans, many of whom are culturally and linguistically distinct from the majority population.

The Mountain States Genetics Regional Collaborative (MSGRC) has served the Mountain States Region since 2004 and seeks to ensure that individuals with heritable disorders and their families have access to quality care, appropriate genetic expertise, and information in the context of a medical home.

MSGRC staffs the activities of six workgroups and funds regional mini-projects to address identified barriers to access for available genetic and medical home services. The six regional workgroups are comprised of diverse stakeholders representing multiple disciplines, including public health, genetics and other subspecialties, primary care, consumer advocacy, local and state government, community-based organizations, and regional and national partners that share similar goals. The current workgroups are:

- Consumer Advocacy
- Emergency Preparedness
- Health Information Technology
- Medical Home
- Newborn Screening
- Telegenetics

MSGRC is continuing to support the following mini-projects:

- Colorado Tele-Genetic Counseling for Newborn Hearing Loss and Metabolic Disorders--to expand access to genetic services for families with children who are deaf or hard of hearing or have been identified as having a metabolic disorder.
- Improving Medical Homes and Transition Outcomes for Youth Served in IMD Clinics--to improve transition services for patients with inborn errors of metabolism (IMD).
- Metabolic Newborn Screening Long-Term Follow-Up Study and Metabolic Consortium--to improve the early identification and long-term outcomes of newborns with metabolic disorders.

MSGRC is also supporting three new projects:

- Hemoglobinopathies Long-Term Follow-Up--to expand long-term follow-up of and improve the medical home for individuals with hemoglobinopathies.
- Mountain States Medical Home and Family Partnership--to incorporate formal family partnerships into primary care practices as a way to strengthen and support the medical home.
- Improving Access to Services for Children with Special Health Care Needs in Native American Communities of Northern Arizona--to assess and then identify solutions to gaps in and barriers to services for families of children with special health care needs in Native American communities.

More information about the MSGRC is available at www.MountainStatesGenetics.org. To get involved with MSGRC, please contact Donna Williams, MSGRC Project Coordinator, at dwilliams@mountainstatesgenetics.org.
**In With the New: New Project Activities in the WSGSC**

Submitted by Lianne Hasegawa, MS, CGC, Project Coordinator; Jennifer Boomsa, MS, CGC, Project Specialist and Sylvia Au, MS, CGC, Project Director, WSGSC

The Western States Genetic Services Collaborative (WSGSC) is forging ahead with priority projects and activities identified by our collaborative partners for the new funding cycle. These initiatives focus on a wide range of important issues, such as financing and reimbursement, increasing access to genetic services (including the use of telegenetics), translation of genetic and genomic information, improving newborn screening, and education and training. Brief descriptions of some of our most exciting activities follow.

**Genetic Services Authorization Project**

Building on an earlier WSGSC project to assess the use of medical decision trees by state Medicaid programs, a workgroup is creating a pilot project to provide expert opinion to insurance companies on authorizing genetic services and/or testing. The goal is to create a centralized program that allows for more accurate, efficient, and expert review of genetic services and testing requests. The workgroup is currently designing an organizational chart and recruiting state Medicaid programs, private payers, and other stakeholders.

**Translation of Genomic Information Project**

In response to an identified need to help families, primary care providers, and public health professionals understand how the rapid and continual increase in genomic knowledge affects their lives and work, this project uses genetic experts to translate information on current genomics-related topics into practical applications and appropriate language levels. Based on the stakeholder need, articles and “genomic messages” will be developed for newsletters, websites, brochures, presentations, and more. The first topic to be covered is whole genome sequencing.

**Telehealth Training Program**

The WSGSC is partnering with the Heartland RC to develop a telegenetics training curriculum for clinical geneticist and genetic counseling trainees and recent graduates. The training will be provided via in-person and distance learning sessions. A workgroup is currently developing a curriculum that will include: an introduction to the use of telegenetics for genetic evaluations and counseling; equipment available for various settings; session logistics; reimbursement; facilitator training; clinical workflow; evaluation of services; and a business plan to justify the use of telegenetics. It is anticipated that the training can increase the number of genetics professionals using this technology and improve access to genetic services and counseling for families.

**Practice Model Activities**

We are continuing our Practice Model activities to determine the best practices for providing genetic services to families living far away from genetic specialists. Information from outreach and telegenetics services is being collected, and details about emerging best practices is being shared regionally and nationally. We invite stakeholders in our region who are currently providing or interested in providing telegenetics to participate in our workgroup activities.

**Affordable Care Act Activities**

To help WSGSC stakeholders understand how the Affordable Care Act (ACA) will affect families with or at risk for genetic disorders, periodic webinars with Meg Comeau, Project Director of the Catalyst Center, are planned to provide timely education about the ACA. (The Catalyst Center is a HRSA-funded national center dedicated to improving healthcare coverage and financing of children with special health care needs.) We will also devote a section on our website to national and state ACA-related resources. Other activities related to the implementation of the ACA are included in such RC projects as Practice Model (genetics and counseling outreach), Medicaid Authorization, and Medical Foods and Formula Coverage and Reimbursement. We are also working with the NCC and other RCs on joint ACA projects related to genetic services and testing.

The WSGSC looks forward to continuing our work with our many stakeholders to improve the health of children and families with or at risk for genetic conditions within our region. If you are part of our region and have an interest in any of our activities, please contact Lianne Hasegawa at lianne@hawaiigenetics.org.

http://www.westernstatesgenetics.org/
NGECN: A Network to Support and Empower

Submitted by
James O’Leary, Chief Innovation Officer, Genetic Alliance

I frequently struggle to pick the right word to describe my role in my own healthcare. Am I a patient, an advocate, or something else? The first term sounds passive, while the second sounds disconnected from the actual services I receive. One word I rarely choose is “consumer.” Yet, in the business community, consumers (or customers) drive whole industries with their preferences and purchases. In healthcare, where there are trillions of dollars in expenditures every year, I shy away from that term because it feels weak. Why is that?

Unfortunately, our healthcare system is fragmented in a way that makes it difficult for consumers to drive change. As individuals, we feel that we are at the mercy of the healthcare system rather than at the helm. We are trapped in an adversarial relationship with our insurers, our employers, and even our doctors. In response, a huge, diverse set of support organizations has emerged to help magnify each individual’s voice. They shore up our healthcare system, provide information and support, and advocate on the local, state, and federal levels. But, like the healthcare system itself, this network of support is fragmented and not always able to reach those who need it most.

As part of the three-year National Coordinating Center cooperative agreement, there is a new focus on consumer education and engagement through the National Genetics Education and Consumer Network (NGECN). Genetic Alliance, through a partnership with the American College of Medical Genetics and Genomics, seeks to make a tangible national impact and augment the existing HRSA Regional Genetics Collaborative (RC) infrastructure. The NGECN aims to make healthcare consumers more powerful by strengthening the network of support, collecting and disseminating advocacy resources, and funding expansion of existing consumer-focused initiatives. The project has five primary goals.

1. Collaborate with Family Voices and Parent to Parent USA to build an advocacy toolkit for RC consumer/family representatives.
2. Expand and strengthen the network of support organizations, including disease-specific advocacy, peer-to-peer connection, patient navigation, and medical assistance.
3. Create a planning guide that will help states identify and expand consumer outreach and education initiatives.
4. Fund seven implementation demonstration projects to scale existing consumer-focused initiatives that have been shown to be effective within a region or expand them to other regions.
5. Educate the public about the impact of genetics on health and help individuals and their families navigate related services through GenesInLife.org.

http://www.geneticalliance.org/ngecn
NewSTEPs Announces the First Steps in its Journey

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

NewSTEPs (Newborn Screening Technical assistance and Evaluation Program) is a national newborn screening project designed to serve as a central link for access to newborn screening information, data, and resources across the country. This comprehensive resource center for state newborn screening (NBS) programs and stakeholders is funded through a cooperative agreement to the Association of Public Health Laboratories (APHL) by the Genetic Services Branch of the Health Resources and Services Administration (HRSA).

State NBS programs will benefit from the shared resources that NewSTEPs offers, including a data repository, interactive website, technical assistance, education and training, policy guidance, and program evaluation.

NewSTEPs’ mission is to assist newborn screening systems achieve the highest quality by providing them with relevant, accurate tools and resources and by facilitating collaboration between state programs and other newborn screening partners. The program’s vision is to ensure that dynamic newborn screening systems have access to and utilize accurate, relevant information to achieve and maintain excellence through continuous quality improvement.

To facilitate these efforts, the NewSTEPs’ staff (listed below) include newborn screening professionals from APHL and faculty from the Colorado School of Public Health (CSPH) with expertise in epidemiology and evaluation. NewSTEPs is also collaborating with state NBS programs and stakeholders in the newborn screening community.

NewSTEPs Team
• APHL NewSTEPs Staff
  - Director: Jelili Ojodu, MPH
  - Senior Advisor: Jane Getchell, DrPH
• Manager: Sikha Singh, MHS, PMP
• Senior Specialist: Careema Yusuf, MPH
• Specialist: Thalia Wood, MPH
• Newborn Screening and Genetics in Public Health Liaison: Elizabeth Jones, MPH

• CSPH NewSTEPs Staff
  - Director of Epidemiology: Marci Sontag, PhD
  - Program Evaluator: Yvonne Kellar-Guenther, PhD
• NewSTEPs Steering Committee
  - Chair: Scott Shone, PhD

For additional information on NewSTEPs, please contact Sikha Singh at sikha.singh@aphl.org

NewSTEPs’ staff includes newborn screening professionals from APHL and faculty from the Colorado School of Public Health with expertise in epidemiology and evaluation.

### Data Repository and Interactive Website
- The data repository will feature a new set of quality indicators for newborn screening programs, and reports that will allow annual tracking and trend analysis for state newborn screening programs.
- The interactive website will serve as a clearinghouse for resources to support state newborn screening systems.
- Development of the repository and website began in 2012, with phased testing completed in Spring 2013.
- Debut of the features of the NewSTEPs data system took place at the Joint Meeting of the Newborn Screening and Genetic Testing Symposium and the International Society for Neonatal Screening in May 2013 in Atlanta, GA.

### Quality Indicators and Case Definitions
- Case definitions for disorders identified by newborn screening have been developed and reviewed by clinical experts. State newborn screening representatives are pilot testing the definitions in 2013.
- A new list of quality indicators developed in partnership with state NBS programs were introduced in Spring 2013, collecting data from 2012.

### Technical Assistance and Site Visits
- Technical Assistance teams will be available for site visits starting Spring 2013, pending funding.
- NewSTEPs looks forward to visiting state NBS programs and working together to improve the newborn screening systems.
Pediatric primary care providers are cordially invited to the CME Course, “Dive into the Gene Pool: Integrating Genetics and Genomics into Your Pediatric Primary Care Practice,” August 9-10, 2013, at the Swissotel in Chicago, Illinois.

For the first time ever, the American Academy of Pediatrics, in collaboration with the National Coalition for Health Professional Education in Genetics, will present a live, 1½ day course dedicated to providing pediatric primary care professionals with cutting edge research, and practical guidance for navigating new technologies and information that will support their early identification and management of patients with genetic conditions. Family health history, expert referrals, and genetic testing will be among the topics covered. Online registration is now open at www.pedialink.org/cmefinder.

Through participating in this conference, participants will be able to:

• Recognize the role of genetics, genomics, and epigenetics in health and disease.

• Identify the role of the primary care provider in the identification and management of patients with genetic conditions, using family health history, expert referrals, and genetic testing.

• Understand the importance of family history in prevention, diagnosis, and management of children and adolescents in primary care and how to utilize practical and innovative tools to construct a problem-based family history, a living history, or a pedigree in the medical chart.

• Apply effective communication strategies when conducting a genetic evaluation, interpreting test results, or conducting difficult discussions with patients and their families.

• Demonstrate appropriate coding for genetic conditions and utilize the electronic health record to capture valuable genetics information.

• Apply a wide range of available resources, technologies, and tools that promote early identification, referral, and management of patients with genetic conditions.

The American Academy of Pediatrics gratefully acknowledges support for this activity in the form of a grant from the CDC National Center for Birth Defects and Developmental Disabilities, with additional funding provided through a cooperative agreement with the Health Resources and Services Administration’s Maternal and Child Health Bureau.
NCC Transition Workgroup Statement of Transition

In late 2012, the National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives (NCC), as part of the larger NCC/RC system-wide evaluation program, asked each NCC subject matter workgroup to propose a definition of a particular subject and delineate ways in which that could be applied to individuals with heritable disorders. The NCC Transition Workgroup developed the following Transition Statement as a guide to the NCC and RCs transition-related activities.

The American Academy of Pediatrics (2002) Consensus Statement on Health Care Transitions noted that transition should take place within a “family centered, continuous, comprehensive, coordinated, compassionate, and culturally competent health care system that is as developmentally appropriate as it is technically sophisticated.” The AAP statement is focused on the role of the healthcare system and physician.

The NCC Transition Workgroup believes that a patient-focused and psychologically more realistic definition of transition is called for, one that addresses social maturation and psychological adjustment as well as optimal physical outcomes. The workgroup proposes the following definition: “Transition is an ongoing, collaborative process that involves preparing for, facilitating, and adjusting to a shift from pediatric-centered healthcare to adult-centered healthcare.”

This definition emphasizes a type of collaboration in which healthcare professionals support young people and their families in recognizing the benefits of and process for moving from pediatric-centered to adult-centered healthcare, encompassing medical care, educational/vocational adjustments, and social adjustment. Medically, healthcare transition does not always mean transfer of care, but rather a shift in how care is provided. Young people need to gradually assume more responsibility for their healthcare choices and treatment. Families need to negotiate developmental issues as the young person gains independence from parents. Assistance may still be required, but may come from roommates, friends, spouses or home health aides. Successful transition requires conversations with the young person and the family in a developmentally appropriate and culturally sensitive manner.

Transition planning must also address educational and vocational plans, taking into account the ability of individuals to exercise independence in managing finances and activities of daily living, as well as in medical decision making. Individuals should be encouraged and supported in their educational and occupational pursuits and in obtaining accommodations for their medical needs, as appropriate. Individuals who have cognitive limitations and/or difficulties meeting self-care needs should be supported in long-term planning about living situations, financial support, and legal guardianship, as needed.

Socially, healthcare transition involves coming to terms with issues related to friendships, dating, sex, reproduction, and life expectancy, among others. Moreover, parents and healthcare providers must recognize the need for young people to establish identities beyond their diagnoses. Providers need to be cognizant that young people may engage in exploration and risk taking before accepting new responsibilities and commitments. Therefore, the goal for healthcare providers is to establish a safety net and support system, as well as uninterrupted, comprehensive, culturally sensitive, and coordinated care.

References:
SERC Emergency Preparedness Workgroup Publishes Article on Reporting Mechanisms

Newborn Screening: A National Snapshot with Implications for Emergency Preparedness, by Phaidra Floyd-Browning, William Perry, and Hans Andersson, to be published in a forthcoming issue of The Journal of Pediatrics, is currently available on-line at http://www.jpeds.com/. In summarizing the findings of their study, Dr. Andersson points out that access to newborn screening results can be time-critical. For some inborn errors of metabolism, urgent diagnosis and initiation of treatment will be lifesaving. Emergencies such as Hurricanes Katrina and Sandy, ice storms, tornadoes, and other events can make connecting abnormal newborn screen results with the patient difficult or impossible. This paper summarizes reporting mechanisms for all states and territories. Presently, many states rely on reporting mechanisms that will likely be interrupted in severe emergencies.

For some inborn errors of metabolism, urgent diagnosis and initiation of treatment will be lifesaving.

The authors suggest the need for harmonization of reporting mechanisms among all states and territories. A high percentage of states/territories are not using an online mechanism for reporting results; yet, in a severe emergency, online access to newborn screening results from distant locations is essential for responding to abnormal results in a timely manner.
NCC and RC Meetings

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<td>RC Reports for the NCC Collaborator</td>
<td>Aug 5</td>
<td>NCC Office</td>
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<tr>
<td>Western States Regional Summit</td>
<td>Oct 2-3</td>
<td>Seattle, WA</td>
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<td>NCC/RC PD/PM Annual Meeting</td>
<td>Nov 14-15</td>
<td>Washington, DC</td>
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<td>NCC/RC and LEND Joint Meeting</td>
<td>Nov 16</td>
<td>Washington, DC</td>
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National Conferences

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<td>Dive into the Gene Pool: Integrating Genetics into Primary Care</td>
<td>Aug 9-10</td>
<td>Chicago, IL</td>
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<tr>
<td>DACHDNC Meeting</td>
<td>Sept 19-20</td>
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<td>5th Annual Health Care Transition Research Symposium</td>
<td>Oct 16</td>
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<td>American Society of Human Genetics Annual Meeting</td>
<td>Oct 22-26</td>
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<td>AAP National Conference</td>
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Announcement

In 2007 Judith Benkendorf, Special Assistant to the Executive Director of ACMG, envisioned a quarterly newsletter that would highlight the excellent work of the NCC/RC system and its partners. During the past seven years she has nurtured and championed this effort, and we thank her for her leadership and stewardship. Ms. Benkendorf has taken on new responsibility on ACMG publications and will devote her work for the NCC on policy matters in the future. We look forward to continuing to work together and benefit from her expertise.