The NCC/RC System at 10 Years: A Look Back

In a 10-year history of successful activities at the local, state, regional, and national levels, the NCC/RC system has significantly altered the landscape of genetics service provision in the US. Working together, the NCC, the National Genetics Education and Consumer Network (NGECN), and the seven RCs have effectively focused on a variety of challenging areas, ranging from long-term follow-up and laboratory quality assurance to the empowerment of consumers within the context of a medical home. Equally important, the NCC/RC system has developed a comprehensive infrastructure to facilitate the improvement of care provision and health outcomes for individuals with genetic conditions. All RCs have devoted considerable time and effort to developing both regional and state infrastructure and engaging key stakeholders. This issue of the Collaborator shares some highlights from each RC’s work during the past decade.

The New England Genetics Collaborative (NEG) created the Genetics Education Materials for School Success (GEMSS) online portal, which offers tools to assist schools and other educational institutions in their work with individuals with genetic conditions. Additionally NEG has developed a regional quality improvement program to help practitioners improve diagnostic processes and clinical outcomes. Finally, NEG has provided small project funding to address specific populations and needs within the region.

Many important projects supported by the New York Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC) have achieved national prominence. The Guidelines for Diagnosis of Screen-Positive Newborn Screening Results has helped to standardize diagnostic protocols. NYMAC’s focus on transition has led to successful transition navigator projects and a recent adult care providers’ conference that focused on improving these providers’ confidence in caring for individuals with genetic conditions.

The Southeast Regional NBS & Genetics Collaborative (SERC) has enhanced genetic expertise among professionals in the region through its evidence-based nutrition management guidelines project, while engaging consumers through the development of a web-based self-report patient registry, NBS Connect. SERC has also developed and published a model for transition for individuals with genetic conditions.

Continued on Page 2
A number of major projects developed by the Region 4 Midwest Collaborative are now national or international in scope. Through the Region 4 Stork (R4S) and Inborn Error of Metabolism Collaborative (IBEM-C) projects, the RC has utilized regional expertise and engagement to improve newborn screening and follow up globally. R4S is used by 49 states and territories and 66 foreign countries.

The Heartland Genetics and Newborn Screening Collaborative has championed the cause of laboratory preparedness. Starting with pilot projects, the RC moved into broader lab back-up testing drills and is now engaged in conducting actual sample exchanges and in process refinement. Heartland has also developed the Genetic Services Assessment (GSA), which offers a set of quality metrics to assess the availability and utility of genetic service delivery at the state level.

Early on, the Mountain States Genetics Regional Collaborative (MSGRC) identified the key role consumers play in enhancing the delivery of genetics services, and the RC has continued to engage consumers as full partners in its projects. Through a Parent Partner project and engagement of native populations, the RC has focused on addressing the unique needs within its region.

The Western States Genetic Services Collaborative (WSGSC) established guiding principles in 2004 and has worked diligently to adhere to those principles. Relationship building has been the key to its work, which has included forging and nurturing strong partnerships with public health in all the diverse states and territories within its region. The RC has also been a leader in improving access to genetic services through implementation and evaluation of various practice models for families living away from urban centers.

Finally, this edition of the Collaborator shares: the work of the National Genetics Education Consumer Network (NGECN); a consumer perspective on the NCC/RC system at ten years; and the current efforts of the National Organization for Rare Disorders (NORD) to promote state-based policy through their Rare Action Network.
Highlights of the Work of the New England Genetics Collaborative

Submitted by
Monica McClain, MS, PhD, Co-Director, NEGC

The mission of the New England Genetics Collaborative (NEGC) is to promote and improve the health and social well-being of those with inherited conditions through collaborations among public health professionals, private health professionals, educators, consumers, and advocates in all New England states. During the past seven years, our stakeholders have been engaged in a broad array of activities and played a variety of roles to accomplish this mission. Below are several highlights of the work accomplished by NEGC.

Launched in 2012, Genetics Education Materials for School Success (GEMSS) is a website (www.gemssforschools.org) that offers tools for schools and other educational settings that explain genetic conditions and provide helpful strategies for those who work with individuals with these conditions. The tools include relevant information about emergencies, field trips, diet, communication, and more. GEMSS was developed by the NEGC’s Education and Outreach Workgroup, which relied on feedback from parents, teachers, and other users to help refine and improve it. Additional conditions are being added regularly, and content is continually enhanced.

The NEGC’s Quality Improvement (QI) Workgroup aims to improve quality of genetic health care services in New England. A regional quality improvement program assesses clinical practices by site in order to inform practitioners about how to improve diagnostic processes and subsequent clinical outcomes for patients with genetic diseases (e.g., the etiology of global developmental delays or intellectual disabilities, PKU, MCAD deficiency). Currently, the registry associated with this project contains data on approximately 1,000 patients with developmental delays and/or intellectual disabilities; the PKU and MCAD deficiency project began its registry this past year.

The NEGC has provided funding for a number of smaller projects related to its mission and goals. These projects have allowed the NEGC to expand its expertise to new areas and to work with new collaborators. Examples of these projects include: a conference for individuals and families with Lynch syndrome that provided medical updates and psychosocial support; an assessment of the impact of the Patient Protection and Affordable Care Act and its regulations on the availability of genetic medical services; development of a method for tracking outcomes of a community-based health management team approach for adults with sickle cell disease; and a patient-as-teacher project in which patients with metabolic disorders and/or family members present their perspectives on living with the disorder, their diagnostic journeys, and their encounters with the health care system.
NYMAC Helps Families of Children with Special Health Care Needs Access Services and Treatments

Submitted by Susanna Ginsburg, MSW, Evaluation Consultant, NYMAC

The New York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC) has significantly contributed to newborn screening (NBS) and genetics education and outreach efforts using a multi-approach strategy that has included the development, piloting, and dissemination of a number of products. These have included: informational brochures; emergency wallet cards; a childbirth education toolkit for nurses, nurse midwives, and other parent educators; postcards and magnets highlighting NBS and the importance of timely follow-up (more than 20,000 of these have been distributed); Genetics in Your Health brochures, in English and Spanish, illustrating the impact of genetics through the lifespan; and the Understanding Genetics manual, prepared in partnership with Genetic Alliance. In addition, NYMAC has supported interactive events, such as webinars, presentations and exhibitions at conferences, as well as genetics and NBS workshops.

NYMAC's Guidelines for Diagnosis of Screen-Positive Newborn Screening Results has helped to standardize diagnosis protocols (http://www.wadsworth.org/newborn/nymac/docs/Kronn.pdf). We have supported educational videos created by the Sickle Cell Thalassemia Patients Network (Let’s Talk about Sickle Cell with Spanish subtitles) and Sickle Cell Association of New Jersey (Say It Loud). NYMAC also provided funding to Save Babies Through Screening Foundation, Immune Deficiency Foundation, Hunter’s Hope Foundation, Genetic Alliance, and CARES Foundation to enhance their outreach efforts.

Our partnerships with our state NBS programs have been particularly fruitful. NYMAC has hosted seminars on lysosomal storage diseases, severe combined immune deficiency, and Pompe disease in order to disseminate information on new conditions and issues related to short- and long-term follow-up. We have also addressed NBS lab emergency preparedness through specimen exchanges and drills.

In addition, NYMAC workgroups have focused on transition, medical home, primary care linkages, public health outcomes, and distance strategies. NYMAC's focus on transition started with a trial of the Cristine Trahms transition modules at Children's Hospital of Pittsburgh. Three “transition navigator” projects transformed approaches to transition (webinar available on the NYMAC website). NYMAC hosted an Adult Care Providers’ Conference and the Affordable Care Act/Care Coordination meeting, demonstrating NYMAC’s strategic association with other regional collaboratives. NYMAC has partnered with primary care associations, the Genetics in Primary Care Institute, and Leadership Education in Neurodevelopmental and Related Disabilities programs to promote integration of genetics in primary care and public health.

Since its inception, NYMAC has included advocate/family views in its efforts. Our Consumer Collaborative Network is increasing collaboration with Parent-to-Parent, Family Voices, and other organizations, ensuring effective outreach to this vital audience. NYMAC is thankful to these consumers and to all of its stakeholders for their contribution and to HRSA and NCC for its guidance and support as we strive to fulfill the charge of helping families of children with special health needs.

http://www.wadsworth.org/newborn/nymac
SOUTHEAST REGIONAL NBS & GENETICS COLLABORATIVE

SERC’s Successful Focus on Enhancing Genetic Expertise and Infrastructure

Submitted by
Lokie Harmond, MPH, SERC Program Manager; Rani Singh, PhD, Emory University, SERC Principal Investigator; Hans Andersson, MD, SERC Co-Principal Investigator

The Southeast Newborn Screening & Genetics Regional Collaborative (SERC) has made significant progress towards improving intraregional relationships and genetic services infrastructure, as well as enhancing genetic expertise across the region. Several of our successful projects have focused on state newborn screening (NBS) programs. For example, we have implemented a peer-to-peer, de-identified sample sharing program aimed at improving regional knowledge about tandem mass spectrometry screened conditions, local data harmonization, and evaluating the degree of analytical correlation between laboratories. SERC has also developed an emergency plan for laboratory backup (using the Florida NBS Lab) and trained laboratory technicians in the event there is laboratory lost capacity. State public health NBS laboratories and follow-up programs participated in this effort and developed a memorandum of understanding and shared blood spot cards/demographic data collection forms during an emergency preparedness lab tour.

Through our Evidence Based Nutrition Management Guideline (NMG) project, we are developing open access NMGs for inherited metabolic disorders included in the recommended uniform screening panel (RUSP). The first set of guidelines, for maple syrup urine disease (MSUD), has been launched and is now available online for all metabolic clinicians nationally. PKU guidelines were published, and a related toolkit for professionals is currently being developed. Also in the area of professional training, we have developed online educational models to increase the knowledge and capacities of laboratorians and genetic workforce engaged in NBS service delivery. In addition, our in-person/publicly-broadcasted educational series, Lunch-and-Learn, is now available on SERC’s webpage. Our most recent series included a session on NBS for severe combined immune deficiency.

One of SERC’s major accomplishments is NBS Connect, a web-based self-report patient registry that serves as a resource for individuals with inborn errors of metabolism, their parents, and/or guardians. NBS-PKU Connect was launched in October 2011, and NBS-MSUD Connect in July 2013. Features include new monthly recipes and an NBS discussion board. Through Facebook and Twitter, we will share instant updates with our valued participants. This is just one of several ways that we seek to make NBS Connect interactive and engaging for both our registered families and professionals.

Helping youth with genetic conditions transition from pediatric to adult care has been another important focus of our work at SERC. Through our SouthEastern Collaborative REgional Transition (SECRET) project, we have published a manuscript describing a proposed model for transition and have developed regional transition program surveys to assess PKU and sickle cell disease transition programs in SERC.

SERC will continue to build the infrastructure needed to effectively identify, share, and, when necessary, develop the genetic resources necessary for our region. Moving forward, we will be focusing on expanding access to care, improving the health of children with heritable disorders through intraregional communication, and developing/implementing a genetic services model.

http://southeastgenetics.org/
Improving Public Health Systems through a Regional Approach

Submitted by
Gina Gembel, MSW, Project Coordinator, Region 4 Midwest Genetics Collaborative;
Piero Rinaldo, MD, PhD, Principal Investigator, Region 4 Stork (R4S) Collaborative Project; Cythnia Cameron, PhD & Susan A. Berry, MD, Co-Principal Investigators, Inborn Errors of Metabolism Collaborative; Janice Bach, MS, CGC & Lisa Gorman, PhD, Co-Directors, Region 4 Midwest Genetics Collaborative

The R4S project, led by Piero Rinaldo at the Mayo Clinic, is focused on improving the analytical performance of newborn screening by tandem mass spectrometry (MS/MS). Newborn screening (NBS) professionals contribute data through a web-based data collection system. The web-based tool is currently being used by 233 lab sites in 49 states and territories and in 66 foreign countries. To date, the database includes close to 18,000 true positive cases.

The simultaneous detection of many metabolic disorders using MS/MS represents one of the most significant advancements in the NBS field. However, this technology brought up challenges, leading to disparities in the distribution of services nationwide. With funding from HRSA beginning in 2004, R4S collaborators were able to regionalize activities to include state programs in data collection and sharing for the purpose of defining disease specific and clinically-defined cutoff ranges. The benefits are greater uniformity of testing and improved analytical performance. In May 2012, the R4S database became part of the Newborn Screening Translational Research Network, which is funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD).

The IBEM-C project, led by Sue Berry and Cindy Cameron, fosters collaboration across multiple clinics to systematically collect data on medical status, management, treatment, and long-term outcomes for children with inborn errors of metabolism (IBEM). This project has made significant strides toward improving scientific knowledge about the natural history of inborn errors of metabolism and effective management and treatment for these disorders. Initial work by the Long-term Follow-up Workgroup of Region 4 created the Inborn Errors of Metabolism Information System (IBEM-IS) and set the stage for innovation in studying IBEM.

In 2007, HRSA awarded Region 4 a grant to expand the IBEM-IS. As the IBEM-IS gained momentum, Region 4 developed an infrastructure to support the collaboration of multiple clinics in collecting and analyzing data. In 2011 the project received funding through NICHD and began collecting data nationally. Region 4 Midwest Genetics Collaborative remains actively involved in the project by supporting data entry of new clinics within the region and partnering with the Region 4 Public Health Long-term Follow-up Workgroup.

The Region 4 Midwest Genetics Collaborative (Region 4) has a history of building successful regional projects that, when replicated, have been of national significance. The Region 4 Stork (R4S) collaborative project and the Inborn Error of Metabolism Collaborative (IBEM-C) are two examples. Both built on our regional collaborative environment of broad based participation, mutual trust, and long-term commitment from stakeholders.

http://region4genetics.org
HEARTLAND GENETICS AND NEWBORN SCREENING COLLABORATIVE

Heartland Collaborative’s Major Contributions to NBS Emergency Preparedness, Quality Assessment, and Telegenetics

Submitted by
Lori Williamson Dean, MS, LCGC, Program Manager, Heartland; Barbara Jackson, PhD, Program Evaluator, Heartland

The Heartland Regional Genetics and Newborn Screening Collaborative has made major contributions to the field by piloting diverse and innovative projects in three areas: newborn screening (NBS) laboratory emergency preparedness; quality assessment; and delivery of genetics services through telehealth.

NBS laboratory back-up testing drills established by Iowa and Missouri were successfully implemented throughout the Heartland region. A comparative analysis found nearly identical results between the originating and the back-up laboratories. These drills not only demonstrated that the region could successfully take care of its own NBS samples in the event of an emergency, but proved that utilizing the Emergency Management Assistance Compact (EMAC) process was an effective strategy for addressing critical needs in this situation. This project has national significance as the laboratory drill process is being adopted by states outside of the region.

Quality measurement is common in chronic disease management, but has been critically lacking in the area of genetics services. The Genetic Services Assessment (GSA) project convened an expert panel to develop a set of quality metrics to assess the availability and utility of genetic service delivery at the state level. Data obtained from the assessment process can further needs assessment, cost-benefit analyses, or program implementation to enhance access to genetic services. Multiple steps were taken during the development process to maximize its validity and utility for states. Results from the implementation pilots were the basis for determining weights and scoring for the measures that constitute the basic building blocks (e.g., all states surveyed were able to meet those measures) of a quality program in genetics. Disparities in genetics services among states have drawn interest from clinicians and public health experts. However, many states have not been equipped to collect information in a comprehensive manner and thus cannot fully assess their own capacity. The GSA represents one of the first opportunities to develop a set of genetics-specific quality measures for both clinical and public health genetics.

In a region where there is a shortage of genetic providers, as well as large geographic distances between many patients and the providers that do exist, implementing a telegenetics system has proved to be an important way of connecting patients and families to genetics services. To date, Heartland has partnered with state and private institutions to support four new telemedicine sites in the region. Experts in the region also developed a guide for how to start a telegenetics clinic. Most recently, Heartland and the Western States Genetic Services Collaborative partnered to offer an educational program (didactic and skills-based) for medical geneticists, genetic counselors, and trainees. The purpose of this program is to demystify telegenetics and, through exposure, education, and training, increase the use of telehealth for delivering genetics services.
Consumer Leadership Plays Key Role in MSGRC Achievements

Submitted by
Celia Kaye, MD, PhD, Project Director, MSGRC; Kathryn Hassell, MD, Associate Project Director, MSGRC; Joyce Hooker, Director of Regional Outreach, MSGRC; Marilyn Brown, MPH, Project Manager, MSGRC; Kristi Wees, MS, Social Media Coordinator, MSGRC

The unique perspective and expertise of consumers have been vital to the work of the Mountain States Genetic Regional Collaborative (MSGRC). Our consumer advocates, whose families have been profoundly affected by genetic conditions, bring to MSGRC a remarkable amount of knowledge and passion. Dedication consumer members Rod Slaught, Joe Martinec, and Lori Wise have been instrumental in identifying and integrating new consumer members into MSGRC’s work. Due to their effective mentorship, consumers are actively involved in all six of our workgroups and co-chair five of them. Some of our most notable projects have been spearheaded by consumer members.

Brad Thompson, M.A., LPC, Co-Founder of the HALI Project, is both Co-Chair of MSGRC’s Telegenetics Workgroup and Project Lead for the Parent Partners Program. Parent Partners, initiated by Brad in Amarillo, Texas, works with pediatric primary care practices to train parents to help other families of children with special health care needs access the non-medical elements of a medical home. The HALI Project, a 501(c)(3) organization focused on improving quality of life for those with special needs and their families, received an impact award from HRSA. These awards have allowed the project to expand to two new practices in each of these two states. Brad’s experience caring for his daughter, who has special health needs, and his training as a Licensed Professional Counselor have been integral to the success of the Parent Partners Program.

Dorey and Yolanda Nez, members of the Navajo Nation, are long-standing advocates with MSGRC. The documentary, Sun Kissed, highlights their quest to understand why their two children were born with Xeroderma Pigmentosum (XP) and why the prevalence of this disorder in the Navajo community is considerably higher than it is among the general population. Sun Kissed premiered on PBS in 2012 and was shown at the 2013 ACMG Annual Meeting as part of a Community Conversation, “Genetics and Genetic Research: Native American Perspectives.” A panel discussion was held with the Nez’s and the film’s directors, Maya Stark and Adi Lavy. In November 2013, MSGRC met with five of the eight Leadership Education in Neurodevelopmental and Related Disabilities (LEND) programs in our region to develop a Collaboration ACTION Plan and Next Steps. One objective was the translation of Sun Kissed into a learning module for LEND programs; this project should be completed this month.

MSGRC is also fortunate to have advocate Kristi Wees on staff as Social Media Coordinator. Kristi works with our Webmaster to administer our website and manages and develops content, such as consumer stories, for the MSGRC Facebook page. The page had 10,083 visits in the last grant year. MSGRC consumer members continue to demonstrate exceptional leadership in providing information and insight to our professional staff, educating the public, and engaging our stakeholders.
WSGSC Relies on Guiding Principles and Strong Relationships to Achieve Its Vision

Submitted by Sylvia Mann, MS, CGC, Project Director.

Regional Summit 2007

Regional Summit 2013

http://www.westernstatesgenetics.org/
In 2004, the Health Resources and Services Administration (HRSA) began to support a system, made up of seven Regional Collaboratives (RCs) and a National Coordinating Center (NCC), focused on genetics and newborn screening (NBS). The NCC’s initial goal was to facilitate infrastructure and partnership development within the RCs by bringing them together with other national organizations, particularly those with established connections to local communities. The initial two years of funding allowed the NCC and RCs to establish the core elements of the system that is still in place today. It was also during this time, that ACMG, with direct support from HRSA, published a seminal supplement in *Genetics in Medicine*. This supplement laid out a recommended uniform screening panel for NBS (RUSP) and emphasized the states’ need for improved newborn screening and genetics service infrastructure in order to effectively adopt the RUSP.

A new five-year funding period, beginning in 2006, allowed the NCC to:
1. Develop subject matter workgroups comprised of RC members, national organizations (e.g., the Center for Medical Home Improvement), and national experts;
2. Engage in project specific activities, such as developing and disseminating clinical decision support tools, commonly known as *ACT Sheets*, and facilitate emergency preparedness lab exchanges;
3. Continue to support and provide technical assistance around RC infrastructure and stakeholder development.

During this time, NCC also helped RCs with funds to initiate the “scaling up” of certain successful projects that were of national significance. For example, the NCC hired a consultant to help the RCs conduct emergency preparedness exercises at their annual meetings. It also gave support to the American Academy of Pediatrics (AAP) to provide genetics and primary care visiting professorships to the RCs and local state AAP chapters. This work helped increase awareness of both the NCC/RC system and the specific needs of individuals with genetic conditions within medical homes.

In 2012, the NCC entered its current, three-year funding period and welcomed the National Genetics Education and Consumer Network (NGECN) (see article on page 11) into the NCC. The NCC also formalized the voluntary national evaluation program pursued in the 2006 to 2012 funding cycle. In addition, the NCC is continuing its work on long-term follow-up, in partnership with the Newborn Screening Translational Network (NBSTRN), by developing data sets for public health follow-up over the lifecourse.

As the NCC enters its final year of current funding, it looks forward to the continued evolution of the NCC/RC system, while pausing to appreciate the successful activities, partnerships, and accomplishments made possible by that system during the past decade.
NGECN Works with the RCs to Connect Individuals to Genetic Services and Support

Submitted by Sharon Romelczyk, MPH, Program Manager, NGECN

While Genetic Alliance (GA) has participated in various ways with the Regional Collaboratives (RCs) since their inception toward a common goal of improving access to high quality genetic services, in the past two years GA has taken on a more formal role, directing the National Genetics Education and Consumer Network (NGECN). NGECN’s activities focus on both public education and consumer engagement. We work closely with individuals with or at risk for genetic conditions and their families in each of the regions to identify any gaps that exist around genetic services and to provide resources that fit their needs. Guided by our Consumer Advisory Group, we have developed tools and trainings and shared best practices for strengthening consumer engagement within the regions.

The rich, collaborative nature of NGECN has also allowed us to disseminate educational materials that facilitate better understanding of the importance genetics plays in health and how to access genetic services. While GA has produced a number of these resources itself, we focus primarily on bringing together the best of what exists through collaboration with our valued partners. One example of a such a collaborative effort is the Guide to Successful Outreach and Education Programs, which compiles strategies from 16 model programs around planning, developing, implementing, and evaluating consumer-focused outreach and education efforts.

This past year we worked with the RCs and other network partners to compile advocacy training resources and create the Advocacy ATLAS. With the help of consumers and clinicians, we created a site called GenesInLife.org to address the needs of the public around understanding and accessing genetic services. We also distributed more than 8,000 Family Health History booklets this year, spreading the word through our national network. Our most recent activity was funding seven “Impact Awards” to help programs expand outreach and education activities already shown to improve access to genetic services and support.

These collaborative efforts have already resulted in increased knowledge about and access to genetic services and, in some cases, have encouraged individuals to make changes in their lifestyles or behaviors. We believe it is the breadth of NGECN that has allowed us to reach more than 191,000 people this past year and will allow us to continue to positively...
Consumer Corner

A Consumer’s Perspective on the Accomplishments of the NCC/RC System

Submitted by
Jana A. Monaco, BS, Member Advisory Council and Medical Home Workgroup, NYMAC

Prior to the growth of genomic medicine, obtaining timely and accurate diagnoses of many genetic disorders identified through newborn screening and navigating the process that followed were extremely difficult for families, as well as for medical professionals not well versed in genetics. It was not uncommon for a child presenting with an acute medical crisis to receive a diagnosis of an inborn error of metabolism too late to prevent lifelong disabilities. Many families embarked on diagnostic odysseys that led to road blocks and unanswered questions as their babies’ health rapidly declined. Too many babies died. Inadequate access to information and services left families and physicians unable to save them.

In response to the significant changes occurring in the field of genetics and the potential impact of genomic medicine on public health, HRSA developed seven Regional Genetic Services Collaboratives (RCs) and a National Coordinating Center (NCC) a decade ago. As a parent of two affected children, an advocate for newborn screening, and a member of the New York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC), I have seen the many ways in which the RCs and the NCC have changed the lives of children with genetic disorders and their families. The NCC and RCs have supported projects to provide both consumers and medical professionals with access to genetic information and services. They have facilitated partnerships at the state, regional, and national levels to eliminate disparities in newborn screening and improve education, treatment, and follow-up. As genomic medicine becomes better integrated into health care, research continues to progress, and new conditions are added to the recommended newborn screening panel, the NCC/RC system plays a vital role in ensuring the needed expansion of services and resources.

The diagnosis of a child with a heritable genetic disorder brings an overwhelming sense of fear and uncertainty to a family. It is critical that the family, as well as the health care providers caring for that child across his or her lifespan, have access to high quality genetic services and the other resources needed to educate, support and guide them. Through its support of the NCC/RC system, HRSA has developed mechanisms to meet those needs. Family advocates have an important role to play in ensuring that new projects are implemented, successful projects are publicized and replicated, and the work of the National Coordinating Center and the Regional Collaboratives continues to be responsive to the challenges families face.

If you are a consumer/advocate would like to submit an article for the next NCC Collaborator, contact Alisha Keehn (akeehn@acmg.net).
Promoting State-Based Policy through NORD’s Rare Action Network®

Submitted by
Paul Melmeyer, Assistant Director of Public Policy, National Organization for Rare Disorders

Since the passage of the Affordable Care Act in 2010 and the subsequent Supreme Court decision allowing states to decide whether to expand their Medicaid programs, decisions about healthcare coverage and quality standards for patients with rare diseases are increasingly being made at the state level. From public health initiatives, such as newborn screening programs, to insurance plan structure and coverage regulations, state governments play an integral role in facilitating the access of quality, affordable care for individuals with rare diseases.

For 31 years, the National Organization for Rare Disorders (NORD) has served as America’s leading patient advocacy organization for patients with rare diseases. Following the passage of the Orphan Drug Act in 1983, NORD has been involved in numerous Federal policy initiatives, including the Rare Diseases Act of 2002, the Affordable Care Act, and the Food and Drug Administration Safety and Innovation Act (FDASIA), among others.

Beyond our policy efforts, NORD represents over 220 organizations for individuals with rare diseases and provides education and coordination services for patients and their families. We work with medical and scientific researchers to advance basic and translational research and with the biopharmaceutical industry to ensure the necessary incentives for orphan drug development remain strong.

Always active at the federal level, we are now increasing our efforts around state-based public policy and advocacy. Using Rare Disease Day, February 28, 2014 as our platform, NORD launched the Rare Action Network® (RAN) to bring together the entire rare disease community around common state-based goals and initiatives. When mature, RAN will serve as a regional and state collaboration of patients, patient families and advocates, medical and scientific professionals, industry, and governmental organizations working towards a common goal of increased access to affordable, high quality, rare disease treatments.

We have organized RAN activities around identifying state-based coalitions and advocates, equipping them with tools and resources, and establishing lines of communication with decision-makers in every state. We are also creating a framework for state policy report cards that will evaluate states on their performance in areas important to the rare disease community and we are communicating with state officials on specific legislation.

We are in the early phases of building state-based coalitions of volunteer advocates, who will serve as the rare disease ambassadors to their state governments. Within these coalitions, we are seeking bold and effective leaders. On the policy side, we are joining existing state and regional policy coalitions and engaging state lawmakers on issues important to the rare disease community. We have also identified four initial focus areas for state policy report cards: Medicaid expansion, newborn screening, specialty tiers, and medical foods.

This is just a snapshot of our Rare Action Network® plan. We hope you will join us in collective advocacy for individuals with rare diseases. After all, as we say at NORD, “Alone we are rare, together we are strong.”

For more information, contact Paul Melmeyer at pmelmeyer@raredisorders.org.

www.rarediseases.org
Community Conversation: The Journey of the Undiagnosed and the Rare: How Researchers and Families Partner to Address the Challenges of Persons with Undiagnosed and Rare Conditions

March 24th, 2015
7:00 PM-9:00 PM
Salt Palace Convention Center 250 DEF

Participate in the Community Conversation to learn more about family history in Utah, undiagnosed conditions, and the views of patients, families, and researchers. The Utah Population Database (UPD) is a unique and rich source of in-depth information that supports research in the areas of genetics, epidemiology, demography and public health. UPD allows researchers to answer to families about various genetic conditions including those highlighted in the film, Undiagnosed. Undiagnosed, which will be previewed during the community conversation, shares the untold story of individuals and families whose lives have been dramatically altered by living with unknown and unsolved conditions. A panel that includes parents of children with undiagnosed and genetic conditions and researchers will offer their perspectives and engage the audience in exploring the issues. ACMG Conference participants, families, health care providers and public health professionals are invited to join the conversation.

HRSA Regional Genetics Collaboratives’ Advocate Leaders Partnership Program

March 25th to 28th, 2015
Salt Palace Convention Center 151 G

Advocate leaders from each RC, along with graduate students in genetic counseling will have full access to the ACMG Clinical Genetics meeting in order to broaden the consumer perspective on medical genetics. Participants will engage with NCC, RC, and ACMG leaders, as well as with medical genetics professionals. They will convene each day for breakfast and lunch to further discussion of topics discussed at the meeting. Additionally, presentations will be given that are tailored for the advocate leaders. Advocate leaders will share their experience with the NCC. This information will also be shared electronically with the NCC/RC consortium.
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<td>WSGSC Regional Summit</td>
<td>Oct 1-2</td>
<td>Seattle, WA</td>
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<tr>
<td>Region 4 Midwest Regional Meeting</td>
<td>Oct 19-21</td>
<td>Lansing, MI</td>
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<tr>
<td>NCC/RC PD/PM Annual Meeting (Pending Funding)</td>
<td>Nov 12-13</td>
<td>Washington, DC</td>
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<tr>
<td><strong>National Conferences</strong></td>
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<tr>
<td>ACMG Annual Clinical Genetics Meeting</td>
<td>Mar 24-28</td>
<td>Salt Lake City, UT</td>
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<tr>
<td>DACHDNC Meeting/Webinar</td>
<td>May 11-12</td>
<td>Washington, DC</td>
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<tr>
<td>DACHDNC Meeting/Webinar</td>
<td>Aug 13-14</td>
<td>Washington, DC</td>
</tr>
<tr>
<td>Southwest Conference on Disability</td>
<td>Oct 6-9</td>
<td>TBD</td>
</tr>
<tr>
<td>American Society of Human Genetics</td>
<td>Oct 6-10</td>
<td>Baltimore, MD</td>
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<tr>
<td>National Society of Genetic Counselors Annual Education Conference</td>
<td>Oct 21-24</td>
<td>Pittsburgh, PA</td>
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<tr>
<td>AAP National Conference</td>
<td>Oct 24-27</td>
<td>Washington, DC</td>
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<tr>
<td>APHA Annual Meeting</td>
<td>Oct 31-Nov 4</td>
<td>Chicago, IL</td>
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<tr>
<td>DACHDNC Meeting/Webinar</td>
<td>Nov 2-3</td>
<td>Washington, DC</td>
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<tr>
<td>AUCD</td>
<td>Nov 15-18</td>
<td>Washington, DC</td>
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</table>
Website Updates

To view the latest resources developed and/or discussed by NCC and the RC’s, go to www.nccrcg.org. Listed below are a few of the webpages that have been updated with new resources:

- News and Events
- Healthcare Access and Financing (Formerly ACA Implementation) Workgroup Resources
- Family Health History Workgroup Resources
- Transition Workgroup Resources
- For Families
- For Providers

NCC is on Social Media!

NCC Social Media is updated daily/weekly. Check Facebook and Twitter for the latest updates from NCC. To be included in a post or tweet, please contact ncc@nccrcg.org