




NCC Collaborator

Expanding Genetic and NBS Services Through Multifaceted Partnerships



RCs' Cross-Cultural Programs Address Health Disparities in Targeted Populations

The late Carl Rogers described culturally and linguistically competent communication as follows: "The more authentic you become, the more genuine...the more people can relate...and the safer it makes them feel to express themselves. That expression in turn feeds back on the other person's spirit and genuine empathy takes place, producing new insights and learning and a sense of excitement and adventure that keeps the process going."¹ Although Rogers is referring to direct communication between individuals engaged in a helping relationship, his words also apply to interactions that take place in public health settings.

Rogers' words are particularly relevant to HRSA's Regional Genetics Collaboratives, whose mission is to

improve access to genetic services and which exist in a nation in which racial and ethnic minorities are expected to make up more half of the population within the next four decades. The Collaboratives are natural laboratories for developing initiatives to improve access by addressing health disparities. It is well accepted that appropriately tailored programs that include community members are the optimal way to mitigate health disparities in underrepresented and special populations. The articles in this issue of the NCC Collaborator demonstrate that the HRSA Regional Genetics Collaboratives are implementing these cross-cultural programs—and are doing them well.

The programs you will read about include: addressing the need for genetic services in etiologic evaluation of the population diagnosed with hearing loss (Region 4 and NCC); de-

veloping tools for special education teachers responsible for a student with a rare genetic/metabolic condition in the classroom (NEGC); hospital-based programs to deliver genetic services to diverse Asian communities in New York City (NYMAC); hemoglobinopathy screening follow-up in non-African American communities (MSGGCC); and newborn screening and clinical genetic services improvement in Guam (WSGSC). In its article, the SERC wonders whether barriers to widespread adoption of telegenetics use may be underpinned by a cultural mismatch, while the Heartland Regional Collaborative discusses moving parent advocates into more professional roles. Three invited articles complete this culturally rich issue of the newsletter, making the point that listening to and understanding the world of another—be it an individual or a population—can narrow gaps in the delivery of genetic services, enhance program value, and work towards decreasing health disparities.

¹ cited in Covey SR. (2004) *The Seven Habits of Highly Effective People*. New York: Free Press, p. 267.

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Genetic Counseling Cultural and Linguistic

Submitted by Nancy Steinberg Warren, MS, CGC, 2009 Jane Engelberg Memorial Fellowship (JEMF) Awardee, National Society of Genetic Counselors

Never judge someone by the way he looks or a book

by the way it's covered; for inside those tattered

pages, there's a lot to be discovered"

— Stephen Cosgrove

The goal of the Genetic Counseling Cultural and Linguistic Competence Online Project is to increase cultural and linguistic competence in the genetic counseling profession through the development of an online Genetic Counseling Cultural Competence Toolkit (GCCCT). The GCCCT is targeted at both the professional workforce and genetic counseling training programs. The project's intended long-term outcomes are to enhance counselor-client alliances, improve knowledge, skills, and attitudes of health professionals, and increase client trust and satisfaction.

In addition, this project will bring the genetic counseling profession into the national dialogue on reducing health disparities. Racial and ethnic disparities in healthcare access, quality, and outcomes abound. While poverty is associated with poor health status, health disparities persist even when adjusted by income and level of education. Disparities may be linked to lack of provider training in cultural competency, stereotyping or biases, as well as an inability of organizations to support diverse populations. Thus, promoting cultural competence among genetic counselors aligns with national public health initiatives to improve

the health of minority individuals.

Cultural competence is defined as creating a healthcare system and workforce capable of delivering the highest-quality care to every patient regardless of race, ethnicity, culture, or language proficiency. To become culturally competent, healthcare providers and professional organizations must acquire and conduct ongoing education and training in culturally and linguistically appropriate service delivery. Cultural competence includes the deep respect demonstrated by professionals who fully understand a client, a concept that resonates with the goals of genetic counseling and the delivery of medical genetics services. **Linguistic competence** includes communicating effectively with people who have limited English proficiency.

By the year 2050, racial and ethnic minorities will make up more than 50 percent of the US population. These population trends, along with



"It is never too late to give up
your prejudices"

— Henry David Thoreau

the elucidation of the genetic basis of common diseases, will increase demands on genetic counselors to deliver quality care to diverse clients. These demands will be exacerbated by the fact that common diseases are often over-represented in minority populations due to health disparities. Cultural competence toolkits have been developed by other health professions, but genetic counseling lacks targeted resources in this area.

Competence Online Project



In addition, more minority providers are needed, especially since they are more likely to serve minority and underserved communities. When advocating for workforce diversity and efforts to promote cultural competence, leaders in the National Society of Genetic Counselors (NSGC) cite cultural sensitivity as a missing link between minority recruitment and the provision of optimal services to culturally diverse populations.

The GCCCT provides didactic information and self-directed learning activities to promote cultural competence in genetic counseling profession-

“Every person is like all others, like some others, like no others.”

— Proverb

als and students. By using the GCCCT, learners will be able to:

- identify their own cultural values, assumptions, and beliefs, and recognize how those may impact patient care;
- conduct client interviews that elicit health beliefs and incorporate these beliefs into the counseling session;
- describe ways to manage discrepancies between the counselor’s and the client’s approach to health, illness, and testing;
- use techniques and resources for working with interpreters; and
- identify resources for continuing professional development in cultural competence.

Drawing from the National Standards for Culturally and Linguistically Appropriate Services in Healthcare and existing models of cultural competency, the GCCCT emphasizes the importance of self-awareness and an individual’s journey towards becoming culturally competent.

The content of the toolkit is being reviewed by experts in education, cultural competence and genetic counseling. The anticipated launch date for this online resource portal is Fall 2010.

This work is supported by the Jane Engelberg Memorial Fellowship, the 2009 grant from the Engelberg Foundation to the National Society of Genetic Counselors, Inc.

Biographical Sketch: Nancy Steinberg Warren, MS, CGC is a board certified genetic counselor and educator. She has been a graduate training program director and educator in the field of genetic counseling for 27 years. Her experience and expertise in cross-cultural healthcare has involved developing and leading local, regional and national efforts to enhance recruitment of underrepresented minorities into the field. She has also led projects with the goal of infusing strategies for improving cultural competence into genetic counseling graduate training programs and clinical practice settings. nancysteinbergwarren@gmail.com

the new england **negc** genetics collaborative

Submitted by Leah Burke, MD, Professor, Pediatrics and Medicine, University of Vermont and Fletcher Allen Healthcare, Burlington, VT (DEM Workgroup Chair); Tracy Fowler, MA, Survey Center, University of New Hampshire; Karen Smith, Project Coordinator and Monica McClain, PhD, Project Manager, NEGC

Dissemination, Education, and Marketing Workgroup Produces a Tool for Educators

Put yourself in the shoes of a newly graduated special education teacher. You love the children, but knowing that some of them may have underlying genetic conditions makes you worry that they are fragile. Or imagine you are a seasoned teacher in a regular education classroom, but this year you have a student with PKU, a rare metabolic genetic disorder. She appears to function like all your other students, but you suspect there are some issues related to her condition that you should probably know about. Finally, picture yourself as the nurse in a bustling elementary school. This year a new family moved into the area with two children affected by a genetic condition about which you are totally unfamiliar.

These kinds of scenarios were on the minds of NEGC's Dissemination, Education and Marketing (DEM) workgroup members when they conceived of a web-based tool to provide guidance to educational teams in school settings on the needs of children with genetic conditions. The DEM group debated key questions, including:

1) How much information do people need to know—basic genetics or just what is useful in the classroom?



2) What is the best way to disseminate information so that it will be used?

The group's vision solidified when members viewed a chart created at their request by Ann Dillon, Coordinator of Clinical Services and Training at the University of New Hampshire's Institute on Disability. This chart became the basis for the web-based tool, *Children with Genetic Conditions: A Guide for the Classroom* that the DEM workgroup is currently refining. What makes the tool unique is that it begins by addressing those topics likely to raise an educator's apprehension. These include things like: *child needs supports for pain; child fatigues easily; child has dietary/medical needs; child's condition declines/changes; and child will attend special functions*. Each of these headings links to bullet points that address the concerns in more detail. In addition, links to related topics are emedded along the way, allowing users easy access to further information.

This past spring the group took its prototype tool on the road. Three formal focus groups, attended by elementary school educators, special educators, para-professionals, occupational and physical therapists, speech specialists, and school nurses, were conducted in Connecticut, New Hampshire, and

Vermont. Overall, participants felt that a web-based tool for information on genetic disorders would be very useful to those in the classroom. Focus group members also suggested that the tool should be adaptable for additional populations—including patients and families—conditions, and characteristics.

The DEM workgroup's task now is to incorporate the feedback they received from the focus groups into the final product, which will be a user-friendly website. Once this website is developed, it will be piloted in a variety of school settings. The group has also begun discussing best practices for dissemination. These may include school in-services, continuing education credit modules, and special education summits or forums. "The success of any product is gauged by its use by the intended audience," says Leah Burke, chair of the DEM group. "We are hoping that this tool becomes indispensable in the classroom."

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

Submitted by Katharine Harris, MBA, Project Manager, NYMAC

Hospital-Based Programs Address Genetic Service Needs of Diverse Asian Communities

The NYMAC region includes over three million people of diverse Asian ethnicities. Language and culture present significant barriers to these populations' ability to obtain health-care services, including information about genetic risk. The availability of health insurance and location of ethnically-oriented services also play important roles in individuals' ability to access genetic information. Using supplemental funding from HRSA/MCHB, we are working with three hospital-based programs to reach out to Asian communities at high risk for alpha and beta thalassemia, as well as for other hemoglobinopathies.

Dr. Patricia Giardina, a hematologist, and Ann Carlson, a genetic counselor, at Weill-Cornell Medical Center in New York City, established an outreach program to the Chinese community at the Charles B. Wang Community Health Centers in Manhattan and Queens many years ago. Their program provides prenatal screening for thalassemias and other congenital disorders, genetic counseling and parent and professional education about genetics and genetic disease. More than 15,000 Asian women have been screened, yielding a carrier rate for alpha thalassemia of about 5 percent and for beta thalassemia of about 3 percent. Much of the patient education



is provided by Wang Center staff—as often as possible in the patient's own language. Educational pamphlets are available in English and simplified Chinese to reinforce the information provided (see *NCC Collaborator*, Vol. 2, No. 4, page 10, December 2008).

Sunset Park, in Brooklyn, New York, is home to a relatively new Chinatown. Asians constitute 25 percent of that population, with half of them Chinese or of Chinese descent. Dr. Rita Bellevue, a hematologist, and Rose Boliscar, a health educator, from New York Methodist Hospital (NYM) in Brooklyn, have identified the health centers and Asian physicians serving the Asian community in their native languages. They are acquiring educational materials in several languages from Weill-Cornell, Thalassemia International Federation, Cooley's Anemia Foundation, and Oakland Children's Hospital. They are also currently

translating a brochure into Burmese for a local population of about 2,000. These materials will be part of a public awareness campaign about thalassemias and hemoglobinopathies implemented through NYM's Public Relations Department.

Dr. Susan Brooks, a clinical geneticist and pediatrician at the UMDNJ-Robert Wood Johnson (RWJ) Medical School, will be working with a Sarah Lawrence College genetic counseling graduate student to provide outreach to the Asian Indian population in Middlesex County, New Jersey. The student expects to do her thesis on thalassemia education in this population. Dr. Brooks is also working on adding a genetics component to the RWJ Department of Pediatrics South Asian Total Healthcare Initiative (<http://tiny.cc/64jqr>) directed by Drs. Sunanda Gaur and Naveen Mehrotra. This program focuses on improving health outcomes by providing culturally competent care and empowering the Asian community.

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



SOUTHEAST NBS & GENETICS COLLABORATIVE

Submitted by Phaidra Floyd-Browning, RN, Project Coordinator (Tulane) and Hans Andersson, MD, FACMG, Project Co-Director, SERC

Barriers to Widespread Adoption of Telegenetics Services: Is the Rejection of Technology Due to a Cultural Mismatch?

During the past few years, the use of telegenetics has been explored in a variety of settings, using an array of systems. Several RCs have done telegenetics projects within their region. The Southeast region has offered telegenetics equipment and support to our states/territories since 2007. The Western States have offered telegenetics services in and between several of their states. The Heartland RC has established a telemedicine network within their genetics community. Georgia and Florida have implemented independent telegenetics systems within their states.

Most of these telegenetics projects were developed to provide improved access to specialized healthcare services (including genetic services) in rural and other remote settings. With the number of geneticists dwindling, telegenetics provides an opportunity for geneticists to serve a larger population base. This might lead one to believe that this service would be highly sought after and utilized. However, our experience in the Southeast has been that there is much less interest in the adoption of telegenetics systems than we initially anticipated. We have discovered several of the reasons for this.

In July 2009, we conducted an electronic survey of the Southeast states/territories regarding regional telemedicine practices. The purpose of the survey was to ascertain a better understanding of how telemedicine was already being used within the region as well as current needs. Our hope was to use the survey results to better tailor the efforts of our telemedicine workgroup to assist those who had a desire to use telegenetics. The survey was sent to approximately 1,075 individuals (the SERGG email population). The response window was one month. The response rate itself was an indicator of the lack of interest in telegenetics/telemedicine in our region. Of the 1,075 individuals who were invited to participate in the survey, only 30 responded.

Among those who did respond to the survey, genetic counselors and clinical geneticists represented 60 percent; the majority of those individuals were located at an academic institution. Of the 30 responders, 43 percent had used telegenetics in some fashion before, and 57 percent had never used telegenetics in any form. Of those who *had not used telegenetics*, 76 percent felt that they could benefit from some sort of telegenetics technology in their daily work. When asked why they had never used telegenetics, the majority cited cost and lack of availability as the main barriers to their implementation of a telemedicine system. Institutional barriers, such as policies and firewalls, and staffing/time constraints, were also mentioned by multiple respondents as reasons for not using telegenetics. Those who *had used telegenetics* listed various systems and methods of

delivery being utilized. Among those who had been engaged in telegenetic activities, 50 percent said they had received no compensation for these services; another 40 percent responded that clinical billing supported their efforts. Only 10 percent reported that their telegenetics activities were grant funded. Those who had experience with telegenetics reported equal use for clinical services and educational purposes (60% for both uses).

What are the real barriers to the use of telemedicine? Financial barriers appear to be overstated. Interestingly, national publications report that reimbursement for telemedicine in other areas of healthcare has been equivalent to reimbursement for in-person services. Billing codes are available that provide reasonable compensation for services. In the Southeast region, we have found that people at the receiving end of these services are reluctant to request or engage in telemedicine systems, even when it is provided to them free of charge, because it deviates from their usual method of asking for and receiving clinical services. Barriers within hospital administrations make establishing telemedicine systems difficult. Many providers are uncomfortable with the technology or reluctant to take time to initiate telemedicine connections. Solutions to these common problems will be required before telegenetics becomes a widely adopted method of providing clinical genetic services. What it may take is a cross-cultural mindset by all participants.

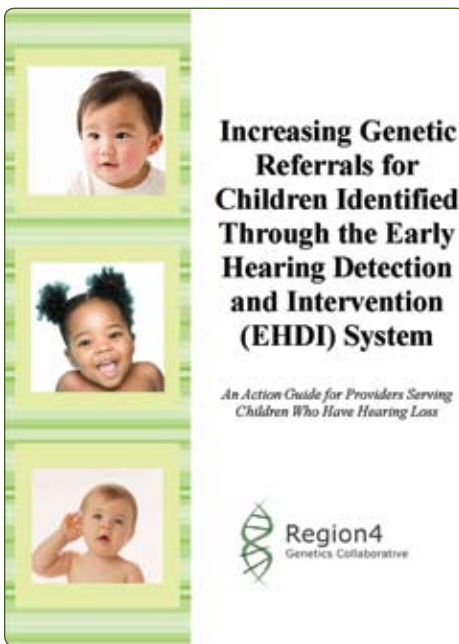
<http://www.southeastgenetics.org>



Region 4 Genetics Collaborative

Submitted by Sally J. Hiner, BS, LSW, Coordinator, Region 4

Special Populations and Cultural Sensitivity: Children with Hearing Loss and Their Families



All families of infants identified with hearing loss should receive information about genetic services as a complement to their child's overall care. Research shows that more than 50 percent of infant hearing loss has a genetic basis.¹ Among those babies, about a third have a complex medical syndrome. Identification of associated features in hearing loss syndromes may have health-saving or life-saving implications.

However, it is important to recognize that serving children with hearing loss and their families raises special chal-

lenges. In order to provide culturally sensitive genetic referrals for these children and their families, health providers need to be aware of several important issues:

- Providers must be sensitive to family needs when communicating the importance of the genetic evaluation. As professionals, it is important to be culturally sensitive to how hearing screening and hearing loss are approached. This includes cultural awareness of how the hearing screening results may be interpreted by a deaf family. Caregivers who are deaf may view hearing loss as a difference, not a disability. Members of the Deaf Community suggest there is an inherent and unwarranted bias in the medical profession that views deafness as a disability or needing medical intervention. The Deaf Community is a separate and valued culture in which members are bilingual (communicating in both American Sign Language and English). Decisions concerning evaluation and potential "treatment" of deafness are personal family matters. Families may not wish to have hearing aids, cochlear implants and/or intervention for their baby with hearing loss. While this perspective may be more common in parents who are deaf, some hearing parents hold this view as well.
- In some cultures, consanguinity (a marriage or partnership between two individuals of the same bloodline) is an accepted practice. With consanguineous partners there is a much higher incidence of autosomal recessive disease and hearing

impairment in offspring.² Taking a family history can help identify consanguineous relationships. When working with families where consanguineous relationships have been identified, it is essential to be sensitive and non-judgmental in order to effectively address the important issues of diagnosis, recurrence, and treatment

The Region 4 Genetics Collaborative has developed an action guide for providers serving children who have hearing loss. The guide, *Increasing Genetic Referrals for Children Identified through the Early Hearing Detection and Intervention System*, includes information on how to be respectful of family and cultural issues when providing genetic referrals and services.

To get the guide into the hands of as many providers as possible, Region 4 will distribute copies (electronic and hardcopy) to Early Hearing and Detection Intervention (EHDI) and early intervention programs in each Region 4 state, free of charge. The guide is available for download at <http://tiny.cc/7cj32>.

¹Canalis, RF and Lambert, PR. (2000). *The Ear: Comprehensive Otolaryngology*. Philadelphia: Lippincott Williams and Wilkins.

²Reddy and Rani (2006). Role of Consanguinity in Congenital Neurosensory Deafness. *International Journal of Human Genetics*, 6(4).

<http://region4genetics.org>



Heartland Genetics and Newborn Screening Collaborative

Submitted by Leisha Suckstorf, MBA, mother of seven children (two with MCAD) and Heartland Collaborative advocate from Nebraska

Parents: The Building Blocks for Advocacy

In October 2007, my life changed drastically when I delivered fraternal twin sons 10 weeks before their due date. One of the twins had a positive newborn screening test for medium-chain acyl-CoA dehydrogenase (MCAD) deficiency, a genetic condition that prevents the body from transforming certain fats into energy. Since then and the birth of a second child with MCAD deficiency, I have been searching for new ways to understand this disorder and to help other parents of children diagnosed with rare conditions. This is why I jumped at the opportunity to become involved in the Advocate Partnership Program at the 2010 Genetic Alliance Conference as soon as I heard about it from the Heartland Regional Collaborative (RC). What a privilege it was to be granted a scholarship by the Genetic Alliance, with matching funds from the Heartland RC, to attend this conference and meet with other advocates, speakers, and rare disease community representatives.

We started off with an eye-opening event, Genetics Day on the Hill 2010. Advocacy organizations, researchers, parents, students, healthcare providers, and industry representatives joined together to visit elected officials and their staffs and to educate them about the needs of children like mine with rare genetic conditions. The Capitol Hill journey was personally invaluable because I was able to connect with and learn from many different health



Leisha Suckstorf (far right) along with other attendees at the 2010 Genetic Alliance Conference in Washington, DC.

professionals, as well as congressional representatives. This experience has motivated me to become a more vocal and articulate advocate for not only my children, but other children and families from Nebraska.

The Genetic Alliance's President and CEO, Sharon Terry, is truly an inspiration to parents and professionals alike, especially since she began this journey as a parent. Like Sharon and me, many parents are thrust into situations that are very difficult to navigate alone. Learning about rare illnesses, metabolic conditions, genomic correlations, and clinical characteristics, especially when these are described in technical terminology, can be overwhelming. It is also important to become informed about a variety of organizations and laws that affect our children.

At the Genetic Alliance Conference, I discovered that there is so much more to learn. Initially, I was concerned that my opinions would be dismissed or judged, or that people would have stereotypes or prejudice about me because I am not a doctor—but I realized that sometimes those are my own

roadblocks. I left the conference feeling so proud of the work that I and many other parents have accomplished by educating ourselves. I also discovered there are new technologies in relation to genes and the data that they produce. I learned about the BioBank initiative, and understood that sharing information among different advocacy groups is imperative. Equally important, I realized that there is such a broad range of talent in the medical genetics community! During the Dinner Debates, I was overcome with emotion as I looked around and realized that the room was full of professionals, researchers, and doctors who weren't there to make money, but because they truly care.

Like many other parents, I didn't start this journey because of my "love for science." I was just a frightened mom with a two-pound baby in my arms and a laptop, trying to understand how to help my child. Parents are the building blocks for advocacy and I implore more parents to become involved, beginning with their Regional Collaborative. I do not have to look farther than the faces of my children for inspiration and motivation. This advocacy process is not something that I set out to do for a living, but it definitely has become something that I will do for life.

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



Submitted by Kathryn L. Hassell, MD, Professor of Medicine, University of Colorado at Denver; Celia Kaye, MD, PhD, Project Director; Joyce Hooker, Project Manager and Liza Creel, MPH, Project Coordinator, MSGRCC

MSGRCC Hemoglobinopathies Newborn Screening and Follow-Up Project

All states in the Mountain States region mandate universal newborn screening (NBS) for sickle cell disease, the condition most frequently detected by NBS programs. Screening can reveal the presence of not only sickle cell disease, but other major hemoglobin diseases, or hemoglobinopathies, as well. In the course of testing for hemoglobinopathies, many more infants are identified as carrying a hemoglobin trait than are identified with disease.

The Hemoglobinopathies Newborn Screening and Follow-up Project seeks to standardize approaches to NBS follow-up in the Mountain States RC. The project depends on input from all states and stakeholders to: train state and community personnel; develop community-specific, culturally sensitive consumer education materials; and ensure referral to medical homes with appropriate expertise in hemoglobinopathies.

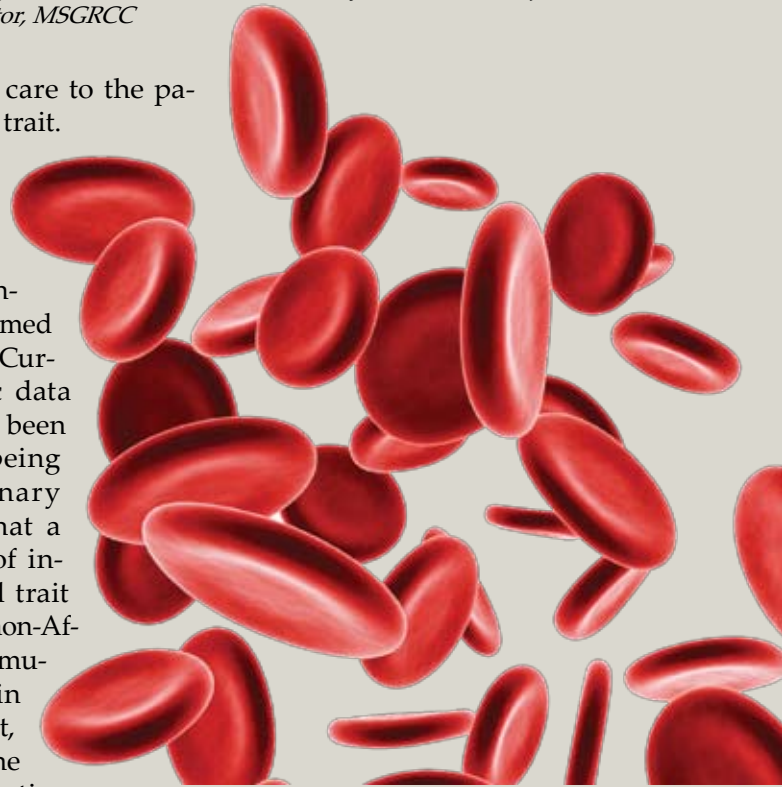
To help achieve these outcomes, project staff organize annual meetings of regional stakeholders. These stakeholders review and provide advice on the sickle cell trait education content, the sickle cell trait demographics project, and a Primary Care On-Line Education Module, which will target primary care providers who deliver

long-term follow-up care to the patients with sickle cell trait.

The sickle cell trait demographics project is collecting data to catalog the race/ethnicity of infants confirmed with sickle cell trait. Currently, demographic data from five states have been collected and are being analyzed. Preliminary analysis suggests that a significant portion of infants with sickle cell trait appear to come from non-African American communities in the Mountain States Region. In fact, projects funded by the Mountain States Genetics

Regional Collaborative Center (MSGRCC) recently demonstrated that approximately 30 percent of infants identified with sickle cell trait in Colorado are of Hispanic ethnicity.

When these sickle cell trait outreach programs, which provide free laboratory testing for parents and family members, are offered few families of African-American background choose to attend. However, there is significant interest and relatively greater attendance by those of Hispanic and other backgrounds. We believe this is due to the lack of anticipation of finding sickle cell disease or trait outside the African-American community. Culturally appropriate follow-up programs must thus reach a variety of communities, including recent African immigrant families, those of Hispanic, Caucasian, and other backgrounds, as



well as African-Americans. The diversity of backgrounds of those families identified with sickle cell trait through NBS programs and the extent to which NBS follow-up programs are culturally competent to offer effective services in the MSGRCC region have not been well documented. This lack of knowledge hampers effective planning for optimal, standardized sickle cell and other hemoglobinopathy disease and trait counseling.

To address these issues, future plans include collecting region-wide information on the effectiveness of NBS notification systems, the capacity of follow-up programs, and access to medical homes for individuals with sickle cell disease.

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



Submitted by Lianne Hasegawa, MS, CGC, Project Coordinator and Sylvia Au, MS, CGC, Project Director, WSGSC

Newborn Screening and Clinical Genetics Improvement in Guam

“Hafa adai” are often the first words heard upon arriving on the small, beautiful island of Guam. The cheerful Chamorro greeting is used by people throughout the island, which boasts a diverse population including individuals of Chamorro (the island’s native community), Filipino, Micronesian, and Chinese ancestry. Although Guam is a US territory, it is geographically closer to Southeast Asia than to Hawai’i, the westernmost US state. The relative isolation of this island territory has contributed to Guam’s unique culture, but it also presents obstacles to the development and maintenance of successful clinical and public health programs.

In 2006, the Western States Genetic Services Collaborative (WSGSC) conducted a needs assessment focused on newborn screening and clinical genetic services in Guam. The needs assessment gathered information from parents of children with special healthcare needs, public health staff, primary care providers, and allied healthcare professionals. These stakeholder groups identified several gaps in healthcare and newborn screening services, but the two primary ones were:

1. **Lack of access to clinical genetic services.** Most families reported that they had to travel to Hawai’i, the US

mainland, or Asia to receive specialty care and that the wait for Guam insurance companies to process non-emergent travel requests could take two to three years.

2. **Lack of a cohesive newborn screening follow-up program.** At the time of the assessment, each birthing center had its own method of following-up on positive newborn screening results. In addition, many primary care providers, public health department staff members, and parents did not know how Guam Memorial Hospital, which cares for 90 percent of the territory’s births, followed-up on abnormal results.

The WSGSC responded to the needs assessment by first organizing a weeklong genetics outreach clinic to Guam. Satisfaction surveys completed by families attending the clinic were overwhelmingly positive. However, when the genetics team attempted to follow-up on the testing recommendations made during the clinic, the genetics staff learned that none of the Guam insurance companies would cover any genetic testing. This included routine testing such as a chromosome analysis and Fragile X molecular testing. The following year, a WSGSC team traveled to Guam and met with representatives from five insurance companies, policymakers, public health staff, and families to help develop a policy for genetic testing. To test the success of the reimbursement activities in Guam, the WSGSC sponsored a second genetics clinic in February of this year. Twenty-four families were evaluated, recommended genetic testing has been or will be ordered, and reimbursement for these tests will be tracked. Depend-

ing on the outcome of this clinic, additional efforts to improve coverage may be implemented in the future.

To address the need for a standardized newborn screening follow-up program in Guam, the WSGSC sent representatives to meet with key public health and clinical staff. After much discussion, it was decided that a newborn screening follow-up program would be created and housed within the Guam Department of Public Health and Social Services (DPHSS). Dr. Laurie Seaver, a Hawai’i-based geneticist, provides clinical consultation to the program, and Dr. Robert Leon Guerrero, a Guam pediatrician, is contracted by the DPHSS to help manage clinical follow-up activities. Over the last year and a half, the WSGSC has worked with program staff to successfully follow-up on abnormal newborn screening results for two of Guam’s three birthing centers. The third birthing center, the Naval Hospital, does follow-up on its own positive newborn screening results.

The WSGSC is dedicated to helping isolated and underserved communities receive genetic and newborn screening services. As such, we are excited to be a part of the development and expansion of these services in Guam. The WSGSC is currently working on expanding outreach genetic services by introducing telemedicine into the existing Guam infrastructure. WSGSC staff looks forward to continued work with the leadership and people of Guam!

<http://www.westernstatesgenetics.org>

A Community Health Worker Model for Genetics Education in the Latino Community

Submitted by Rosita M. Romero, MSW, DWDC Executive Director; Claudia De la Cruz, MSW/MDiv, LCGEN Program Coordinator; Jesus Sanchez MSUP, LCGEN Local Evaluator

The Latina/o Community Genetics Education Network (LCGEN) Project was developed by the Dominican Women's Development Center (DWDC) to increase access to information, resources, and genetic services in the Washington Heights/Inwood neighborhood of New York City. It was one of four projects supported by the March of Dimes Foundation's Community Genetics Education Network (CGEN) project. CGEN was funded by HRSA's Maternal and Child Health Bureau.

LCGEN activities included a workshop composed of two modules: *Latino/a Families: How to Improve Your Health and the Health of Your Community*; and *How to Have a Healthy Pregnancy*. A Community Health Worker (CHW) delivered the workshop, using a desk-top flip chart presentation. Each participant was given a workbook with all the information provided in the flip chart, along with space to take notes. The project also developed a CHW Facilitator's Guide, which contains additional background information on genetics and guidance on how to conduct a workshop. A local community resource guide of health and genetic services is included in the participant workbook and is also available as a stand-alone document.

The LCGEN project was committed to engaging community members in the development of the project's structure and products. DWDC used a community-based participatory research approach to develop culturally and linguistically relevant genetics education materials. A Community Advisory Board (CAB) consisting of local health professionals, a genetic counselor, and university professors,



was instrumental in developing the materials. CHWs also actively participated in the development of the materials. In addition, the CHWs received training in capacity building and how to conduct community workshops.

A LCGEN Network, composed of community members, CHWs, representatives from local community-based organizations, healthcare providers, members of the CAB, DWDC's Executive Director, the local Project Evaluator, and the Project Coordinator, met monthly during the planning stages of the project. These meetings provided opportunities to conduct informational workshops and capacity building trainings for network members. Meetings also allowed the Network to collectively identify methods to be used in accomplishing the project's goals.

CHWs conducted 26 community presentations, reaching 247 community residents. A pre-/post-test evaluation method was used. Preliminary find-

ings showed statistically significant increases in participants' knowledge, an increase in their intent to talk with their family and healthcare providers about family health history, and a high level of satisfaction with the program.

The LCGEN project materials are available in Spanish online at <http://dwdc.org/HGenetic.html>. For more information, or for materials in English, please contact Rosita M. Romero, DWDC Executive Director at 212-994-6060 and/or rromero@dwdc.org.

Genetics and Genetic Research: Native American Special Satellite Meeting to the ACMG Clinical Genetics Conference

Submitted by Nanibaa' Garrison, PhD (Navajo), Postdoctoral Fellow, Stanford Center for Biomedical Ethics in the Center for Integration of Research on Genetics and Ethics at Stanford University (panel moderator)

Controversies over genetic research on Native Americans have led many tribes to institute moratoriums on genetic research studies and have raised ongoing issues around genetic testing, services, and research in the Native American community. To bring these issues to the attention of the broad community of medical geneticists, the American College of Medical Genetics (ACMG), at its annual meeting in Albuquerque, New Mexico in March 2010, held a special panel discussion with tribal leaders and community representatives of the Navajo (Diné) Nation. This important satellite session, the first of its kind at a national genetics conference, was sponsored by the HRSA Genetics Collaboratives and their National Coordinating Center. It grew out of and capitalized on the first of a series of community conversations with the Navajo people on genetics and newborn screening initiated in 2009 by the Mountain States Genetics Regional Collaborative Center (MSGRCC).

The panelists were: Mr. Johnson Dennison (*Navajo*) from the Office of Native Medicine at the Indian Health Service Chinle Comprehensive Healthcare Center in Chinle, AZ and a practicing Navajo medicine man; Ms. Patricia Thomas (*Laguna Pueblo*) from Family Voices National Center for Family/Professional Partnerships in Albuquerque, NM and an educator and advocate for children with special healthcare needs; Mrs. Yolanda Sandoval-Nez (*Navajo*) from the Native American Disability Law Center in Gallup, NM and the mother of two children with Xeroderma Pigmentosa

(XP); and Mr. Robert Yazzie, JD (*Navajo*) from Diné Policy Institute in Tsaile, AZ, where he develops policy using Navajo thinking. Cultural taboos, trust, identity, and politics were four of the major themes that emerged from the wide-ranging panel discussion.

Mr. Dennison described traditional teachings, philosophy, and **taboos** integrated into the intricate clan system to prevent consanguineous marriages. Navajo medicine men are attuned to events before or during childbirth that later affect the child's well-being. Blessingway ceremony prayers help cleanse the mother's womb so only good characteristics are passed to the next generation. Mr. Dennison emphasized that because we've (Navajos) had this knowledge for thousands of years, we don't do research and we don't need others to do it for us.

Lack of **trust** of Western scientists and doctors on the part of the Navajo Nation stems from a long history of trauma. The introduction of diseases from Europeans, forced relocation of hundreds of thousands of people from their homelands by US army soldiers, provision of sub-standard foods (e.g., canned meats, flour, and high sugar/calorie foods), and attempts to assimilate young children by placing them in boarding schools have caused tribes to suffer extreme population reduction and health disparities. In some cases, minor population expansions occurred, allowing rare genetic diseases mutation that might otherwise have remained rare



to increase in frequency. Because of this, researchers became interested in studying the genetic basis of diseases in Native Americans.

Many of these genetic studies have focused on common diseases such as Type 2 diabetes, which is prevalent in up to 50 percent of Pima Indian adults. Genetic association studies largely proved unfruitful, so scientists moved on to other projects, while still requesting additional DNA from Native American subjects. Many Native Americans feel over-researched, while very few have learned the results of the research done on them or have personally benefited from it. Tension and mistrust between Native Americans and the genetics community have ensued. In 2004, the

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Panelists (left to right): Johnson Dennison; Yolanda Sandoval-Nez; Robert Yazzie; Nanibaa' Garrison and Trish Thomas

excitedly asked to collect blood for a genetic study on the family; after all, everyone traveled together that day and it would be easy to collect blood and assemble a pedigree. Offended by the approach and feeling objectified, the family immediately declined. They now wonder what would be done with their blood if they had consented. The nurse's direct approach had been culturally inappropriate: she and the doctors involved needed to build **trust** with the family, to include the extended family in discussions, and to give family members time to think about whether they wanted to become involved in this type of research.

Reservations have been flooded with anthropologists and population geneticists who want to study Native Americans, discover relationships between tribes, and explore human origins. Questions about origins lead to other issues around **identity** that permeate genetic research at multiple levels, from issues in ancestry and migration to privacy and anonymity of a tribe.

In the 1990's, scientists from the Human Genome Diversity Project approached tribes requesting DNA to study migration and diversity. Their scientific views conflicted with Native American views of and beliefs about migration, evolution, and origin. Tribal members also feared the results of these studies could be used to challenge their land rights. Additionally, tribes were offended by the scientists' urgency about studying Native Americans before they all "disappeared."

Havasupai tribe sued Arizona State University researchers for misuse of DNA samples and lack of informed consent. Samples collected for studies on Type 2 diabetes were used for studies on schizophrenia, migration, and inbreeding. A month after the ACMG meeting, in April 2010, the university's Board of Regents settled the case with a "desire to remedy what was done wrong;" this included paying \$700,000 to the 41 tribe members, returning the blood samples, and providing other forms of assistance to the Havasupai.

At a time when little was known about the rare genetic diseases in her extended family, Ms. Thomas and her family sought treatment in Albuquerque, a few hours drive away from their rural home. During one visit, a nurse

Genetic research is very much a **political** issue. In 2002, the Navajo Nation issued a moratorium on genetic research studies that is still in place today. Mrs. Sandoval-Nez described her personal experiences as a mother raising two children with a rare genetic disease. Told by the doctors that her children wouldn't live past age 10, Mrs. Sandoval-Nez constantly struggled with her personal decision to allow her children to participate in genetic research, despite the moratorium.

Mr. Yazzie is interested in creating a policy to regulate genetic research on the Navajo Nation, if the moratorium is ever lifted. Currently, no policies are in place around such issues as intellectual property and ownership of data.

Panel discussions like this, along with work being done by MSGRCC and by other RCs with Native American tribes in their regions, are helping to foster ongoing dialogue on genetic research, as well as the delivery of genetic services to these important populations.

Special thanks to Dr. Murray Brilliant (Marshfield Clinic) for bringing this panel together and creating a valuable opportunity to share this conversation with a wider audience.

Hearing Loss, Genetics and Your Child: New Parent Brochure Available from the NCC

Submitted by Judith Benkendorf, MS, CGC, Special Assistant to the Executive Director, ACMG and Associate Project Director, NCC, with Kathleen Arnos, PhD, FACMG, Genetics Program Director, Gallaudet University

In April 2005, ACMG and MCHB/HRSA, Genetic Services Branch, jointly convened a meeting to address issues in hearing loss screening arising from the molecularization of screening technologies. Meeting attendees with broad expertise in the detection, evaluation and management of childhood hearing loss, newborn screening, molecular diagnostics, and public health were invited; they were joined by consumers of these services.

Early in the meeting it became clear that prior to policy development on role of molecular technologies as an adjunct to the evaluation of genetic contributions to the etiologic diagnosis of hearing loss in newborns and children, it was even more critical to address a bigger problem—infants detected with hearing loss through the state-based early hearing detection and intervention (EHDI) programs were not being referred to genetic services in the numbers expected when about 50% of childhood hearing loss is due to a genetic cause. Rather, follow-up typically focused on information and services related to hearing and communication. The group determined that the lack of genetics referrals could best be addressed by a two-pronged approach: 1) improved knowledge among EHDI professionals about genetic influences in early hearing loss and 2) empowering parents of children with hearing loss to ask for a genetics referral by providing appropriate education materials at the point of care.

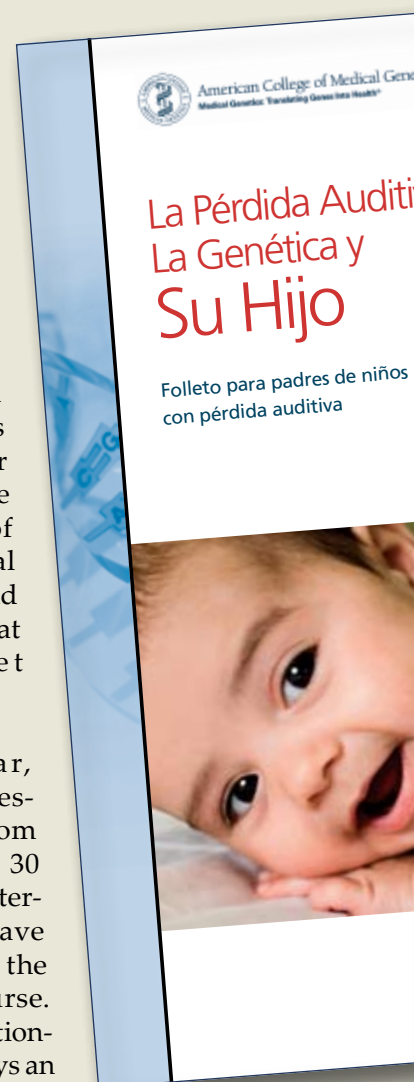
Arnos Uses the 2006 Tm BioScience/ACMG Foundation Award to Promote EHDI Genetics Education

Each year the ACMG Foundation selects a project that will promote safe and effective genetic testing and services for a \$100,000 award, generously funded by Luminex (then Tm Bioscience). Dr. Kathleen Arnos, a participant in the April 2005 meeting, was selected as the 2006 awardee for her proposal to develop of a comprehensive educational effort to disseminate information about the genetics of hearing loss to EHDI professionals, and ultimately to the parents of children with hearing loss. Project staff at Gallaudet University collaborated with geneticists at the Virginia Commonwealth University (VCU) and with the director and staff of the National Center on Hearing Assessment and Management to achieve two project goals: 1) to improve the basic knowledge of EHDI professionals regarding genetic factors in hearing loss, and 2) to facilitate interaction among EHDI professionals from different states and territories in the US to share ideas and improve their efforts to integrate genetics into EHDI programs at the local level.

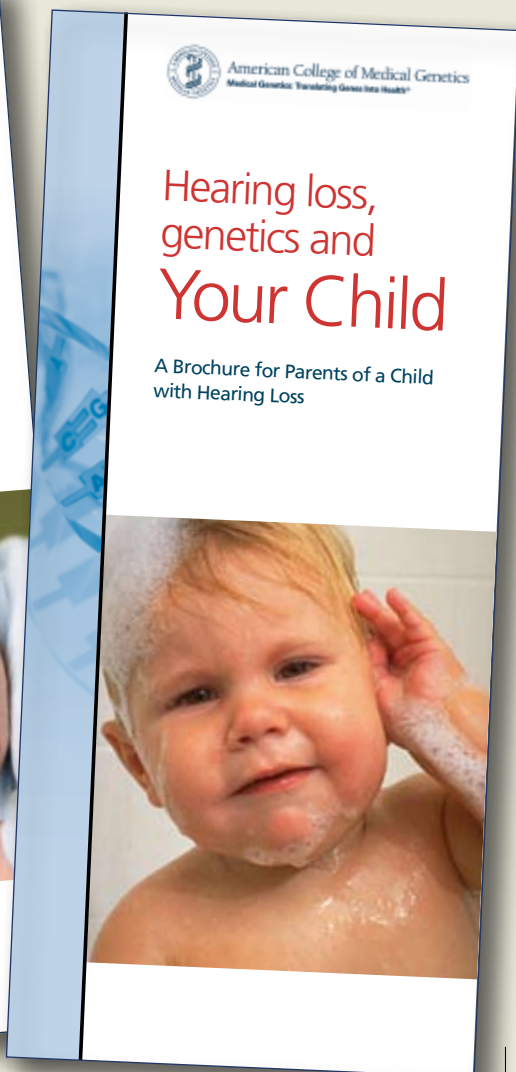
Educational workshops about genetics were conducted at the 2007 annual EHDI meeting, attended by EHDI state coordinators and program professionals, pediatricians, geneticists, and audiologists. Thereafter, a more comprehensive educational program was planned and implemented, and based on the results of a 2004 needs assessment targeted to EHDI state coordinators, a distance learning course with modules on specific topics in genetics was designed. The 10-week online course, taught by geneticists from

Gallaudet University and VCU, was first offered in 2007 and has been repeated 1-2 times per year through the College of Professional Studies and Outreach at Gallaudet University.

Thus far, EHDI professionals from more than 30 states and territories have completed the online course. This educational effort plays an important role in the safe and effective translation of science related to the ongoing discovery of genes for hearing loss into the practice of medical genetics. However, an ongoing theme discussed by participants in the online course is the need for accessible educational materials about genetic evaluation and testing in English and Spanish for the parents of deaf and hard of hearing children.



NCC and ACMG has Important History



and health science information specialists in the development of the new *Hearing Loss, Genetics and Your Child* brochure. The brochure was designed both as a tool to assist medical professionals to convey information about the purpose and process of genetic evaluation and to increase parents' comfort with genetic services by empowering them with information about the importance of genetic services in the etiologic diagnosis of hearing loss, thus complementing the referral. The goal is to have the *Hearing Loss, Genetics and Your Child* brochures available at the point of care, such as waiting rooms in primary care settings and ENT and Audiology centers, where infants and children with hearing loss are most often evaluated.

The *Hearing Loss, Genetics and Your Child* brochure, which will be disseminated by the NCC and HRSA Regional Genetics Collaboratives, covers topics that families of a child who has been diagnosed with hearing loss or is being tested for hearing loss should know, such as:

- Why should I have a genetics doctor see my child?
- How can I prepare for the genetics appointment?
- What happens at the genetics appointment?
- What should I know about genetics, genes and hearing loss?
- Will insurance pay for genetic testing?

The brochure has space for individuals to take notes and write down questions, making it especially handy for taking to medical appointments.

Hearing Loss, Genetics and Your Child is written at a low-literacy level and is available in both English and Spanish. Downloadable brochures are available online at www.nccrcg.org (under the Resources tab) at no cost and contain modifiable space for adding local resource information. Print copies may be ordered by contacting Matthew Tranter (mtranter@acmg.net). The minimum order for printed copies is 100 brochures for \$15.00, plus shipping and handling.

The development of this brochure was partially funded by U22MC03957, awarded as a cooperative agreement between the Maternal and Child Health Bureau/Health Resources and Services Administration, Genetic Services Branch, and the American College of Medical Genetics.

New Parent Brochure Now Available to Fill this Need

Most parents are surprised to learn that there is about a 50% chance that a child's hearing loss is due to a genetic cause, and a child can have a genetic-related hearing loss even if no one in his or her family has hearing loss. Therefore, it is very important that parents know to make an appointment with a clinical geneticist.

To meet this need, Arnos led a team of medical geneticists, genetic counselors, audiologists, parent advocates,



2011 ACMG Annual Clinical Genetics Meeting March 16–20, 2011

Vancouver, British Columbia, Canada



NCC Calendar

NATIONAL and NCC/RC CONFERENCES

National Society of Genetic Counselors (NSGC) Annual Education Conference	Oct 14-17	Dallas, TX
Maternal Child Health Bureau (MCHB/HRSA) Celebrating Title V at 75	Oct 20	Washington, DC
Association of Maternal and Child Health Programs (AMCHP) AMCHP/CityMatCH Meeting: Opportunities and Challenges in Health REform for MCH (Looking to the Future)	Oct 21	Washington, DC
Association of University Centers on Disabilities (AUCD) Annual Meeting	Oct 31-Nov 3	Crystal City, VA
American Society of Human Genetics (ASHG) Annual Meeting	Nov 2-6	Washington, DC
American Public Health Association (APHA) Annual Meeting	Nov 6-10	Denver, CO
Face-to-Face NCC/RC PD Meeting	Nov 18-19	Gaithersburg, MD
New England Genetics Collaborative (NECG) Annual, Advisory Council and Workgroup Meetings	Nov 30-Dec 1	Portsmouth, NH



NCC Collaborator

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Editor-in-Chief Judith Benkendorf, MS, CGC

Associate Editors Alisha Keehn, MPA, Matthew Tranter, BA and Gloria Weissman, MA

Design & Production Lori J. Oxendine, BFA AIGA

Contact Information:

NCC | c/o American College of Medical Genetics | 7220 Wisconsin Avenue, Suite 300 | Bethesda, MD 20814

Tel: 301-718-9603 | Fax: 301-718-9604

ncc@nccrcg.org | www.nccrcg.org

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