



# NCC and AAP Joint Visiting Professorships: Moving Principles into Practice

**H**ow might a busy medical geneticist incorporate medical home principles into the care of patients and families? Likewise, how might a community pediatrician become savvy at caring for a family whose infant was diagnosed with a rare genetic disorder through newborn screening? Based on a belief that genetics knowledge and medical home principles are best moved into practice through active learning, the NCC and the American Academy of Pediatrics (AAP) have twice joined forces to sponsor Genetics and Medical Home Visiting Professorship Programs (VPs).

The VPs are a mechanism for the Regional Collaboratives (RCs) and AAP chapters to improve collaboration and co-management between pediatricians and sub-specialists. Visiting Professorship applications were made available to all RCs and AAP chapters; completed applications were reviewed by a committee comprised of AAP newborn screening program staff, AAP chapter affairs staff, NCC staff, the AAP liaison to the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children and a member of the National Center for Medical Home Implementation Project Advisory Committee.

In 2008-9, Medical Home Visiting Professorships (MHVPs) were awarded to Mountain States RC (MSGRCC), Region 4, and Western States RC (WSGSC). Genetics Visiting Professorships (GVPs) were awarded to the following AAP Chapters: Illinois, Georgia, Montana, New York 3, and Uniformed Services West. Brief summaries of these experiences follow below. The 2010-11 MHVP winners include two RCs—MSGRCC and

Southeast (SERC). Three AAP chapters were awarded GVPs: the District of Columbia, South Dakota and Virginia. The activities of these groups are currently in progress.

## MEDICAL HOME VISITING PROFESSORSHIPS

### Mountain States (MSGRCC)

In February 2009, the MSGRCC hosted a MHVP in Phoenix, AZ. Speakers included Laura Pickler, Assistant Professor of Family Medicine and Pediatrics at The Children's Hospital Denver; Kathy Watters, Director of the Children with Special Health Care Needs Unit at the Colorado Department of Public Health and Environment; and Brad Thompson, Director, The HALI Project. The Forum focused on connecting public health, patients and their advocates, and medical providers of primary and specialty care under a medical home model. Objectives included developing a common language and appreciation of what each stakeholder brings to the discussion, and identifying resources to inform medical home efforts specific to states represented in MSGRCC. Participants included specialty and primary care providers, public health practitioners and consumer advocates. A follow-up MHVP Program was conducted in late February 2011.

### Region 4

The Region 4 MHVP was held during the September 2008 regional meeting. Renee Turchi, Director, PA Medical Home Program (EPIC IC), served as the visiting professor. Region 4's goals for the MHVP were to educate families about the importance of medical home for children with heritable disorders and to create a learning consortium focused on identifying and resolving barriers to effective communica-

tion among families, specialists, and primary care providers. Since one of the main goals was to increase family involvement and knowledge, a session solely for parents was conducted during the VP, and was well received.

### Western States (WSGSC)

Louise Iwaishi, Assistant Professor of Pediatrics, John A. Burns School of Medicine, University of Hawai'i at Manoa, served as the MH visiting professor for the WSGSC. The RC's objectives were to: develop a tool to help healthcare providers understand the history of a child with a genetic disorder in order to improve care; and identify useful screening and genetic information for inclusion into existing guidelines already in use by the medical home. Dr. Iwaishi recommended that the RC: 1) organize a medical home workgroup to develop a genetics-specific medical summary form to facilitate transition of care from a pediatric to an adult health care provider; 2) raise awareness of regional opportunities to partner and promote medical home activities with local chapter initiatives, state Title V agencies and university MCH LEND training programs; and 3) promote the RC's website broadly through the region.

## GENETICS VISITING PROFESSORSHIPS

### Illinois

This GVP involved three events, featuring Bruce Korf, Chair, Department of Genetics, University of Alabama at Birmingham and President, ACMG: 1) a meeting at a local, urban hospital (not affiliated with an academic center) held in the evening to accommodate providers from the hospital and the local healthcare community;

2) a combined grand rounds at Rush University Medical Center (Children's Hospital) and John H. Stroger Hospital (public hospital); and 3) a case-based noon lecture for pediatric and internal medicine-pediatrics residents and medical students at Rush Children's Hospital/University Medical Center. The overall goal of the GVP was to improve the health of children in Illinois by moving genetic medicine into public health and healthcare services. A secondary objective of the GVP was to begin establishing linkages between staff of the Illinois newborn screening program and providers in an effort to further solidify the collaborative relationship between the AAP chapter and the state newborn screening program.

### **Georgia**

The Georgia Chapter's GVP was held during their annual *Pediatrics by the Sea* meeting in June 2009. John Moeschler, Director of Clinical Genetics at the Dartmouth-Hitchcock Medical Center, and Project Director of the New England RC served as the visiting professor and presented during a 3-hour genetics seminar on Thursday, June 18, as well as during the general session on Friday, June 19. The chapter's goal for the GVP was for participants to gain knowledge of genetics in the medical home in order to improve care and to create linkages between pediatric practices and the RC.

### **Montana**

Susan Berry, Professor of Pediatrics, University of Minnesota School of Medicine was invited to serve as the Montana Chapter GVP, which was conducted as an educational session during the annual chapter meeting. The goals of the GVP were to increase pediatricians' knowledge of genetics,

to provide support for practitioners addressing their patients' genetics issues, and to increase provider knowledge about new genetics tests. The chapter plans to build upon the GVP by: setting up a secure link on the chapter website for a members' genetics discussion board; working with the state to improve the time in which newborn screening results are returned; and developing a state genetics registry.

### **New York 3**

Regional hospitals were invited to host grand rounds lectures or pediatric department meetings with David Kronn, Director, Biochemical Genetics Program, Westchester Medical Center and three events were held during spring 2009. Objectives for the GVP were to increase: 1) general practitioners' knowledge and understanding of newborn screening as well as of newer genetic testing techniques; 2) the number of families with children identified through newborn screening who receive appropriate referral to genetics counseling centers; and 3) access to local genetic resources through the Lower Hudson Valley Perinatal Network webpage. Although the chapter and the RC had participated previously in joint activities with the RC, this GVP gave them an opportunity to re-establish and further their working relationships and to pave the way for future collaborations.

### **Uniformed Services West**

In June 2009, the Uniformed Services West Chapter hosted Cynthia Curry, Professor of Pediatrics and Genetics, UCSF Fresno during a GVP consisting of three events: 1) a morning lecture session during which specific genetics topics not previously covered as part of resident education were discussed; 2) an informal lunch discussion focused

on strategies for understanding newborn screens and serving as the newborn screening coordinator at small, remote locations; and 3) a review of individual dysmorphism cases with Dr. Curry. As a result of the GVP, a resource manual was developed and disseminated to all participants.

## **OUTCOMES AND LESSONS LEARNED**

The NCC and AAP are pleased with the outcomes of this program. The MHVPs succeeded in increasing participant knowledge of medical home principles and available resources, while enhancing the relationship between the RCs and local AAP chapters and initiating ongoing dialogue and information sharing between the RCs and their visiting professor. All five of the GVP recipient chapters felt that the program increased member knowledge about genetics and available genetic resources, enhanced their relationship with the local RC, and began an ongoing relationship with the visiting professor. This program demonstrates how national organizations can work together to further education and behavior change at the practice level.

# the new england **negc** genetics collaborative

*Submitted by Michelle M. Winchester, JD, University of New Hampshire School of Law, Durham, NH and  
Monica R. McClain, PhD, Project Manager, NEGC*

## Legal Review of Health Information Exchange in the New England States

Many of the current improvements in healthcare delivery depend on information technologies that allow the exchange of medical record information among and between providers, healthcare organizations, and public health programs. With these technologies becoming more ubiquitous, the genetic practitioners within the New England Genetics Collaborative (NEGC) have begun to collect and analyze clinical information in order to increase the RC's ability to look at health outcomes and use quality improvement strategies to enhance patient care. In doing this, NEGC is committed to maintaining the highest standards of health information privacy and security, and ensuring that all participants are knowledgeable about their legal protections related to health information technology and exchange. Before the NEGC expanded its efforts to integrate genetic information into new health information technologies, it recognized the need to assess the laws and regulations that impact work conducted in clinical and public health settings. At the request of the NEGC, Michelle Winchester, an attorney with expertise in healthcare, reviewed those laws and regulations in each New England state that specifically relate to the exchange of health data that may contain genetic information. Barriers, impediments, and challenges to conducting health information exchange activities within and between states



were identified. Particular attention was focused on those NEGC activities involving the formation of registries, data sharing and exchange between non-administrative or fiscally related organizations, and patient consent requirements. When applicable, national and regional legal and policy analyses in these areas were also reviewed.

The study found that it is standard for state and federal law to treat genetic health information as sensitive information, subject to enhanced protections. How this should be operationalized, however, remains a subject of considerable debate. Even the six New England states cannot reach agreement on how healthcare providers should treat genetic information. One state has no law on the subject, two states treat genetic information in the same manner as all other health information, and three states require written consent (or "informed" written consent) prior to disclosure of genetic information. In the states that require written

consent for disclosure, the common exception to the requirement is newborn screening (NBS) information obtained pursuant to state NBS requirements. The extent of the NBS exception may be limited to information use and disclosure explicitly identified in state law and may not extend to testing or disclosure performed secondary to or subsequent to the required newborn screen and initial report, or to the initial treatment referral. It should also be noted that privacy concerns are not limited to genetic information but also extend to the general medical record. The full study report may be accessed at: <http://goo.gl/BS0nB>.

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

*Submitted by Jane M. Breck, MD, PKU Program Director and Nicole Payne, RD, LDN, PKU Program Coordinator, Children's Hospital of Pittsburgh of UPMC*

## Innovative Program Fosters Independence in Teens with PKU

*NYMAC recognizes the vital role a coordinated transition program can play in successfully moving a young adult with a chronic medical condition from a pediatric practice and parental monitoring of diet and activities to medical care by adult-oriented providers and substantial self-sufficiency. In order to implement model transition programs in our region, NYMAC posted a request for applications to use the transition modules developed by the Cristine M. Trahms Program for Phenylketonuria (PKU) at the University of Washington (UW), in a Mid-Atlantic setting. The PKU Program at Children's Hospital of Pittsburgh applied and received funding. This is their story.*

Although rare, PKU is one of the more common inherited metabolic disorders. It is an ongoing challenge to obtain an optimum clinical outcome due to the demands of lifelong dietary restriction of phenylalanine—a diet even more limited than a vegan diet! Difficulty maintaining diet therapy leads to high phenylalanine (Phe) levels in patients, which can contribute to neuro-psychological, social, and intellectual difficulties. My team and I, as Pittsburgh's PKU Program Director, have implemented an adolescent transition curriculum based on The Teen Transition Project from the UW Cristine M. Trahms Program for Phenylketonuria. The information in the eight educational modules gives adolescents

with PKU the tools and knowledge they need to manage their PKU and to meet the challenges of living independently.

All members of our multi-disciplinary team, which includes a physician, two metabolic dietitians, a clinical psychologist, a social worker, a genetic counselor, and a registered nurse, take part in implementing the educational curriculum. We are using a two-pronged approach for module education—providing education in the clinic and also through off-site events in the “real world.” Innovative lessons have included: a cooking demonstration provided by the owner of a low-protein foods company; a demonstration of the effects of toxic Phe levels on the brain using a life-size brain model;

and hands-on exercises in accurately determining portion sizes. Thinking outside of the typical clinic-exam-room box, we have taken these young adults to entertainment complexes, teaching kitchens, and grocery stores.

One teen participant offered to act as our teen mentor, providing valuable information regarding program planning and future initiatives. Future plans include: visiting a college campus; collaborating with support groups to provide scholarship assistance; increasing efforts to pass legislation that

would ensure financial support for young adults with PKU; and continuing to educate the community about the value these young adults can provide. We extend a genuine thank you to NYMAC for the grant that has allowed us to work toward achieving our ultimate goal of nurturing an independent lifestyle for young adults with PKU.

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





# SOUTHEAST NBS & GENETICS COLLABORATIVE

Submitted by Rosalynn R. Borlaza, MA, Program Coordinator, Emory University/SERC

## SERC Presents Fourth Training Course for Genetic Metabolic Clinicians

The Southeast NBS and Genetics Collaborative (SERC) hosted the fourth Genetic Metabolic Nutrition Symposium from October 11-15, 2010 at the Emory Conference Center Hotel in Atlanta, GA. Course Director Rani H. Singh, PhD, RD convened experts from Emory University, Children's Hospital of Orange County (CA), Maria Fareri Children's Hospital (Valhalla, NY), Oregon Health & Science University, Tulane University, and UNC Chapel Hill to serve as faculty. The twenty-three course participants included nineteen metabolic dietitians, one metabolic nurse practitioner, and four medical fellows, from the United States\*, Australia, Mexico, and the Netherlands. SERC provided partial support for the US attendees and the Mountain States RC contributed funding for the metabolic dietitian attending from Arizona. These clinicians, with experience in inherited metabolic disorders (IMD) ranging from two months to nine years, brought their unique perspectives to the weeklong training course.

Each year, the symposium gives attendees a foundation in genetic metabolic nutrition as it relates to children with the four major categories of IMD identified through newborn screening (NBS)—amino acid disorders, organic acidemias, fatty acid oxidation disorders, and disorders of carbohydrate metabolism. Instructors also present



2010 Genetic Metabolic Nutrition Symposium Attendees, Faculty and Staff

management strategies for short-term and long-term follow-up of these patients. The lectures further address genetics principles, the biochemistry of underlying metabolic pathways, and the pathophysiology of IMD. Faculty and attendees also bring case studies to discuss challenges and best treatment practices in the clinical setting. This multidisciplinary approach not only serves to highlight the importance of nutrition intervention as a mainstay of therapy for these patients, but also outlines the roles of various team members and the importance of care coordination between them to achieve optimal outcomes for patients with IMD across the lifespan.

Since 2007, 84 people have participated in an annual Genetic Metabolic Nutrition Symposia. Pre-tests, post-tests, and course evaluations indicated: increases in knowledge; particular satisfaction with opportunities to share practical experience through case study discussions; and the benefits of presenting current and comprehensive information on NBS and IMD to both new and experienced genetic metabolic clinicians. The comments also reveal the strong need to both continue

and expand these specialized training courses beyond the region. SERC will continue efforts to secure funding in order to maintain such educational endeavors and to share this important training opportunity with more clinicians working in the field of IMD. A poster fully describing this educational event and its outcomes was presented at the annual meeting of the Society for Inherited Metabolic Disorders (SIMD), February 27 to March 2, 2011 (<http://simd.org/Meetings/SIMD2011/>).

For more information please visit <http://southeastgenetics.org/>

\*US attendees represented to following states: AZ, CA, DC, ID, MO, NC, NJ, NY, PA, OH, RI, SC, and WV.

<http://www.southeastgenetics.org>



# Region 4 Genetics Collaborative

Submitted by Cynthia Cameron, PhD, Director, Region 4 Genetics Collaborative

## Management of Congenital Hypothyroidism and the Importance of Three-Year Follow-Up

Newborn screening for congenital hypothyroidism (CH) was adopted several decades ago and is now universal in the United States.<sup>1</sup> In order to prevent permanent cognitive and physical delays, infants with CH must be quickly and properly identified and treated. In addition, the American Academy of Pediatrics guidelines<sup>2</sup> recommend follow-up of these children for the first three years of life to ensure they receive appropriate treatment and management. However, many endocrinologists are concerned that this follow-up is not occurring in a large number of cases.

In 2009, Michigan's Newborn Screening Program, in cooperation with the Michigan Pediatric Endocrine Advisory Council (PEAC) and Region 4, collected data on 45 children with CH three years after diagnosis to determine whether testing and/or treatment for CH was continuing. Many of the children were receiving active treatment by the endocrinologist or pediatrician of record. For most cases considered lost to follow-up, the Michigan Care Improvement Registry (MCIR), which includes information about providers of childhood immunizations and was readily accessible, was used to identify and contact providers. In a few cases where current care providers



could not otherwise be identified, the program made direct contact with the parents to obtain the information.

Based on Michigan's data, it appears that a surprising number of children diagnosed with CH as newborns are no longer being treated for CH at three years of age. While a re-evaluation by TSH challenge is recommended practice, only two of the thirteen children no longer being treated had been re-evaluated using the challenge. At least eight families stopped treatment on their own accord.

Region 4 recently established a CH Workgroup, chaired by Kupper Wintergerst, MD from the University of Louisville. Endocrinologists and state follow-up representatives from each Region 4 state will expand data collection to the other six Region 4 states. This study will increase understanding of when and why treatment is discontinued for children diagnosed with CH. The CH Workgroup will use the data to develop guidelines for state

follow-up systems that support the standard of care recommended by AAP. It is anticipated that these efforts will result in a decrease in the number of children diagnosed with CH whose treatment is discontinued without re-evaluation by TSH challenge.

### References:

<sup>1</sup>National Newborn Screening and Genetics Resource Center. All states screen for CH. <http://genes-r-us.uthscsa.edu/nbsdisorders.htm>. (accessed 2/4/11).

<sup>2</sup><http://www.guideline.gov/content.aspx?id=9383> (accessed 2/4/11)

<http://region4genetics.org>



# Heartland Genetics and Newborn Screening Collaborative

Submitted by Lori Williamson, MS, CGC, LGC, Program Coordinator, Heartland Collaborative

## Development and Implementation of State Genetics Plans: Technical Assistance

The Heartland Pilot Project Program has allowed our RC to test innovative solutions to regional and national needs, while building relationships among our stakeholders through the collaboration required by each project. To date, eleven projects have been completed, and two are underway. Outcomes of the completed projects include three peer-reviewed publications, seven poster presentations at national meetings, continued funding by a state agency for one of the clinical service pilots, and conversion of one project into two priority projects funded annually by the Heartland RC. The project featured in this article, **Development and Implementation of State Genetics Plans**, led to two states developing state genetics plans that have had positive long-term effects on genetic services in the region.

At the start of the project in 2005, three states in the Heartland RC (KS, ND, SD) did not have state genetics plans. State genetics plans guide state decision making with respect to genetic services for its citizens, and are the out-

growth of a process that requires leaders in the state to examine the state's capacity to meet its citizens genetics needs and identify opportunities and threats on the horizon. When these are clearly defined for decision makers, a rational course of action can be developed. The project leaders hypothesized that significant capacity and resource limitations in these three rural and frontier states may have resulted from the inability of their public health departments to develop and implement effective genetic plans.

Two project collaborators, Lisa Butterfield (KS) and Mary Riske (ND), proposed to address this priority need for infrastructure building. Kansas hosted a workshop on development and implementation of state newborn screening and genetics plans for the three states; key stakeholders from each state attended. Peers from states (HI and OK) with a completed state genetics plan that had been used successfully were the presenters. After each presentation, each state team participated in a breakout session to work on the aspect of the state plan (e.g., needs assessment, advisory council, key components of the plan) highlighted in that particular presentation. State teams left the workshop with action steps and a good deal of initial work on the plans completed. As a result of this project,

KS and ND now have state genetics plans and KS has a newborn screening advisory council.

"Laborious but worth the effort in the end! Created increased awareness of all the health department and genetic department collaborative efforts in providing services across the State."

—Mary Riske RN, MS, CNS, State Genetics Coordinator, ND

### Technical Assistance Workshop Agenda

#### State Panel Discussions: Current Status of State Genetics Plans

Kansas: Linda Kenney  
North Dakota: Jayne Brown  
South Dakota: Lucy Fossen  
*Speaker: Sylvania Au, MS, CGC, Hawai'i State Genetics Coordinator*

#### Essential Components of a State Genetics Plan: What You Want vs. What You Can Get

*Speaker: Pam King, MPA, RN, Oklahoma Director of Genetics and State Genetics Coordinator*

#### Abbreviated Strategy for Assessment, Development and Implementation of a State Genetics Plan

Afternoon sessions followed by state breakout workgroups

#### Needs Assessments: Tools for State Genetics Plans

Sylvia Au, MS, CGC

#### Developing an Advisory Council for Your State Genetics Plan

Lori Williamson, MS, CGC

#### Key Components of Your State Plan

Pam King, MPH, RN

#### Large Group Discussion of Next Steps

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



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

## MSGRCC Funds Two Innovative Projects in Telemedicine

In May 2010, the Mountain States Genetics Regional Collaborative Center (MSGRCC) issued a Request for Applications for innovative projects that would link existing telehealth and telemedicine capacity in the region to consumers in need of genetic information and access to services. After input from external reviewers and the MSGRCC Advisory Council, MSGRCC funded two regional telemedicine projects.

**The Colorado Early Hearing Detection and Intervention program** is housed at the Colorado Department of Public Health and Environment, Children with Special Health Care Needs unit. The goal of their telemedicine project is to use a medical home approach to provide community-based comprehensive systems of care for families and their children identified with hearing loss. Currently, any child identified with a permanent hearing loss is referred to their local Colorado Hearing Resource Coordinator (CO-Hear Coordinator), an early interventionist that specializes in deafness. The CO-Hear Coordinator provides resource and referral information to families, including genetic services at The Children's Hospital, which currently has only one genetic counselor specializing in the genetics of deafness. Families residing in the Den-



ver area have direct access to genetic counseling, but families living outside of Denver do not have the same access. This project is using telehealth equipment to videoconference counseling sessions between affected families and the genetic counselor. The CO-Hear Coordinators facilitate the referral and assist the family with the equipment setup. This project is projected to increase the number of families with children identified with permanent hearing loss receiving genetic counseling services from 100 to 150 by 2012.

Montana is a geographically large state with a dispersed patient population. Only one of the seven genetic counselors in the state is based in the eastern half of Montana, causing limited access to genetic counseling services for patients in that region. **The Montana Tele-genetics Project: Improving Access, Educating Providers, and Consumers** is working to increase access to services in a time- and cost-effective

manner by implementing and expanding telemedicine genetic counseling. The project builds on the existing Eastern Montana Telehealth Network to accomplish three goals. First, Montana's genetic

counselors are developing telemedicine scheduling and genetic counseling protocols to offer

to patients within four states including Montana, eastern North and South Dakota, and northern Wyoming. Additionally, they will provide genetics education to consumers and healthcare providers using the existing telemedicine infrastructure. Finally, they will develop and present two consumer-oriented educational sessions, "Everything you need to know about genetic screening in pregnancy" and "Is cancer inherited?" Each project component is being evaluated, and the evaluation data will be used to further refine counseling protocols and expand educational opportunities.

For more information on these projects, please contact Liza Creel at 512-279-3906 or [lcreel@msggcc.org](mailto:lcreel@msggcc.org).

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



# Western States Genetic Services Collaborative

Submitted by Jacquie Stock, MPH, Project Evaluator; Lianne Hasegawa, MS, CGC, Project Coordinator; Barb Chambers, Co-Collaborator Lead, Family Work Group; Kristine Green, Co-Collaborator Lead, Family Work Group and Sylvia Au, MS, CGC, Project Director, WSGSC

## Portable Health Record for People with Genetic or Metabolic Conditions

With an eye toward future use of electronic personal health records, family leaders and primary care providers in the Western States Genetic Services Collaborative (WSGSC) partnered in a work group to develop a Portable Health Record (PHR) specifically for people with genetic or metabolic conditions. The PHR is intended for use during times of transition or emergencies. The individual (or caregiver), together with healthcare providers, keep the information up to date. With up-to-date health information all in one place, providers meeting a new person with a genetic condition may more readily understand the person's needs when first working together.

Work group members began by conducting an Internet and literature search to learn about existing models of PHRs for people with chronic conditions. Next, they created a PHR form based on findings from their search and their own personal and professional experiences. The larger WSGSC Family Work Group then reviewed the form and suggested content and formatting changes. The group wanted the form to be adaptable to either a paper or electronic format. Until Internet-based electronic health records are fully functioning throughout the



country, families and providers may need a paper PHR to have information accessible when needed.

The work group determined that a PHR for people with genetic or metabolic conditions would need the following content areas:

- 1) demographic information, including language;
- 2) alerts (allergies, medication, procedures to do or avoid);
- 3) primary care and specialty physician contact information;
- 4) diagnosis (by whom, at what age, lab tests);
- 5) current physical condition;

- 6) medications, food, formulas, supplements;
- 7) surgeries;
- 8) other management information, such as prostheses, assistive devices;
- 9) common health concerns and suggested management;
- 10) behavior and communication; and
- 11) comments on patient and family.

In August 2010, the WSGSC Family Work Group received approval from the Hawai'i

Department of Health Institutional Review Board to conduct a research project to explore family and provider beliefs and attitudes toward the PHR. Young adults with a genetic or metabolic condition, or parents of a child with such a condition, will fill out the PHR and participate in a telephone interview. Physicians will be recruited to review the completed PHRs and participate in a telephone interview about their beliefs and attitudes toward use of the PHR. Currently three young adults and one mother have participated in the project. Recruitment for additional family and provider participants is now underway.

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

# NCC Noteworthy

## Join the Community Conversation at the ACMG Annual Meeting and Learn About Screening for CPT1A in First Nations Populations

The National Coordinating Center for the Genetic and Newborn Screening Service Collaboratives (NCC) is supporting its second special satellite symposium at the Annual Clinical Genetics Meeting as an opportunity to expose conference attendees to unique genetic services issues in underserved populations. This year we are delighted to partner with 'Kloshe Tillicum' British Columbia and Yukon Territories Network Environment for Aboriginal Health Research and the ACMG Foundation to discuss *Screening for Carnitine-Palmitoyl Transferase, Type 1A (CPT1A) in the First Nations Populations of Alaska and British Columbia*.

This public symposium will present evidence for both risk and protection conferred by the CPT1A variant prevalent in First Nations populations. Local experts will then discuss on-going population management and public health strategies, setting the stage for a community conversation with families of those identified with a CPT1A variant, medical geneticists, healthcare providers, and aboriginal health policymakers. ACMG conference participants are encouraged to attend, learn, and join the conversation.

The Community Conversation will be held on Wednesday evening, March 16 from 7:00-9:00 PM at the Vancouver Convention Centre. For a detailed listing of the presentations and speakers visit the ACMG meeting site

at [www.acmgmeeting.net](http://www.acmgmeeting.net), and click on Program Sessions and Events Schedule in the left-hand column.

### Genetic Counseling Cultural Competence Toolkit (GCCCT) Now Available

The October issue of the *NCC Collaborator* featured a special article by Nancy Steinberg Warren, MS, CGC about a Genetic Counseling Cultural and Linguistic Competence Online Project. The project's long-term goal is to enhance alliances between genetic services providers and families, improve knowledge skills and attitudes of health professionals, and increase client trust and satisfaction, in a breadth of cross-cultural contexts. Beta testing has recently ended and the toolkit and resources are available to all.

The GCCCT website and resource portal are designed to provide information on demand to members

of the genetic counseling profession, students, and other interested health-care providers and consumers. The website is filled with links and information on various topics in cultural diversity, and cultural and linguistic competence that can be applied to genetic service delivery, research, and teaching.

The complete contents of the website are freely accessible on the Internet. In addition, a variety of options are offered for individuals who want to register for Category 1 CEUs or a Learning Certificate for the 9 cases. The cases have been approved for 1.44 Category 1 CEUs or 14.43 contact hours by the NSGC as an approved provider of the IACET (International Association for Continuing Education and Training).

The site is directly accessible from: <http://www.geneticcounselingtoolkit.com>.





# NCC Calendar

## NCC MEETINGS

<b>CPT1 Screening in First Nations' Populations:</b> A Community Conversation	Mar 16	Vancouver, BC
<b>NCC Effective Follow-Up PIs and Partners Meeting</b>	Mar 28-29	Arlington, VA
<b>NCC Telegenetics Workgroup Meeting</b>	May 4	Tampa, FL
<b>NCC/RC/GSB PD/PM Mid-Year Meeting</b>	Jun 6	Chicago, IL

## NATIONAL CONFERENCES

<b>Genetic Diseases of Children</b> Advancing Research & Care	Mar 8-9	New York, NY
<b>American College of Medical Genetics (ACMG)</b> Annual Clinical Genetics Meeting	Mar 16-21	Vancouver, BC
<b>Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC)</b> 24th Meeting	May 5-6	Washington DC
<b>Second National Conference on Genetics, Ethics and the Law</b> Regional Summit	Jun 1-2	Charlottesville, VA
<b>Genetic Alliance</b> Annual Conference: 25 Years of Innovation	Jun 23-25	Bethesda, MD



## NCC Collaborator

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