PERSPECTIVES in genetic counseling

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national society of genetic counselors, inc.

the leading voice, authority and advocate for the genetic counseling profession

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Table of Contents

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SmithBucklin Announces New Executive Director for NSGC

NSGC is pleased to announce the appointment of ***Kristen Smith, CAE,*** *as our permanent Executive Director at SmithBucklin. Kristen replaces* ***April Snyder,*** *who has served as our interim Executive Director since November 2005 and who has helped facilitate NSGC's recent transition. Thank you, April, and welcome, Kristen!*

Jessica Mandell, PGC Editor

Kristen Smith

I am very excited to join NSGC as the Executive Director. I currently work with the American Society of Hand Therapists and Special Care Dentistry Association, both SmithBucklin clients. Prior to joining SmithBucklin, I worked with Special Care Dentistry when it was a stand-alone organization, and I was the Director of Management Services for the American Osteopathic Association, where I served as Executive Director for multiple organizations.

I am a graduate of Purdue University and earned my Certified Association Executive degree in 2004.

I have experience in strategic planning, financial management and advocacy efforts. I hope to apply my areas of expertise to NSGC's strategic efforts. I plan to immerse myself in NSGC and learn as much as I can. I believe it is very important to focus on meeting your needs as members, and I look forward to working with you.

Kristen Smith, CAE

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Genetic Counseling and Health Disparities

Kelly Ormond, MS

The issue of health disparities in genetics is enormous. It involves access to genetic counseling for individuals of all socioeconomic levels and cultural and ethnic backgrounds, and a need for genetic counselors to be culturally competent and able to serve clients with whom they do not share a common language.

In the past, our profession has tried to address health disparities by recruiting minority genetic counselors. However, it is not sufficient just to attract students from underrepresented populations. We must provide a welcoming academic and professional environment, opportunities for professional development and employment opportunities that allow our colleagues to shape the profession, serve as role models and give back to various communities.

Retreat Allows Free Expression

During 2005, NSGC began to make progress on this front in two ...to page 3

President's Beat

The past few months have been a busy time for NSGC. While our transition to SmithBucklin continues to hold our attention, NSGC remains focused on the three initiatives of the 2004-2006 Strategic Plan: Genetic Counseling Service Delivery Models, Increased NSGC Visibility and Improving Outlook for Billing and Reimbursement. I am pleased to announce that many goals have been met and work on remaining goals continues to progress.

Getting our Message Out

The NSGC Speakers Bureau is up and running. Over 100 members have signed up online at our website. If you have not done so, I encourage you to join this effort.

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The opinions expressed herein are those of the authors and do not necessarily reflect those of the editorial staff or NSGC.

Next issue ***June 15***
Submission deadline ***May 11***

Our public relations consultants at StarRosen developed opportunities for promoting NSGC's message for several members including **Jill Allen, Robin Bennett, Rene Chard, Jill Fischer, Nancy Kramer, Kelly Ormond, Dan Riconda, Jill Stopfer, Scott Weissman** and myself. SmithBucklin will continue to identify topics for future public relations efforts.


Nancy Callanan

NSGC has committed resources to develop a marketing strategy. I appointed a Marketing Resource Group to work with **Linda Schwartz,** Director of Marketing and Communications at SmithBucklin.

Three NSGC Practice Guidelines (Cystic Fibrosis, Fragile X Syndrome and Recurrent Miscarriage) also have been submitted to the National Guideline Clearinghouse (www.guideline.gov).

Speaking Up and Speaking Out

NSGC liaisons continue to play a key role in representing our issues and values externally. **Dawn Allain** will Chair a workgroup that includes **Wendy Uhlmann, Beth Balkite** and **Teri Creeden** to evaluate our current liaison relationships, review their goals and make recommendations on the responsibilities and training of liaisons.

In the past few months, several liaisons including **Ken Loud** (Institute for Quality in Laboratory Medicine), **Wendy Uhlmann** (NHGRI), **Jennifer Sullivan** (Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children) and **Heather Ferguson** (Professional Guidelines for Genetic Testing hosted by the Genetics and Public Policy Center) represented NSGC at meetings.

NSGC was invited to participate in the Banbury II Summit hosted by the American College of Medical Genetics. Board Member **Angela Trepanier** represented NSGC.

Seeking Opportunities

NSGC was invited to present at "Genetics Programs and Services: Predictive, Preventive & Personalized Patient Care Through Genetics," scheduled for May 2006 in Chicago. This conference, coordinated by Active Communications International, targets healthcare professionals and hospital administrators. **Dawn Allain** will present. NSGC will be a media partner, helping us further inform conference attendees about NSGC.

Planning for the Future

NSGC will begin developing our 2007-09 Strategic Plan this year. The process will include a membership survey, a great opportunity for members to weigh in on the directions of NSGC. Related to this, **Kelly Ormond** will lead a work group on Long Range Executive Office Planning Issues, seeking input from NSGC membership (via the Strategic Planning Survey and other methods) and from external stakeholders.

I hear frequently from our staff at SmithBucklin how impressed they are by the level of involvement of NSGC members in our professional organization. Knowing how many are engaged in NSGC activities energizes me. On behalf of the BOD and staff, I thank each and every one of you who supports NSGC with your contributions of time, creativity and expertise. ❖

Nancy Callanan

Nancy P. Callanan, MS
2005-2006 President

areas. First, on August 25-26, NSGC facilitated a retreat in Nashville to discuss internal NSGC culture. The agenda was developed by **Tene Franklin, Barbara Harrison** and myself and was facilitated by **Vivian Ota Wang.** Invitees represented various ethnic backgrounds and included individuals who demonstrated interest in this issue for genetic counseling. Attendees included: **Anne Greb** (representing ABGC), **Linwood Lewis** (a psychologist from Sarah Lawrence College who has published in this area) and genetic counselors **Damini Dasai, Maria Della Rocca, Kisha Johnson, Kelly Taylor** and **Patrick Wilson.**

Left to right, bottom row: Kisha Johnson, Kelly Ormond, Tene Hamilton, Damini Dasai, Barbara Harrison; top row: Vivian Ota Wang, Patrick Wilson, Linwood Lewis, Maria Della Rocca, Kelly Taylor; Anne Greb (taking picture)

The unique meeting offered an opportunity to talk confidentially about how people of any background feel welcome or unwelcome within NSGC and which experiences play a role in feeling welcome into our professional community. While many comments reflected the feelings of "new" members to any organization, other ideas were specific to individuals of minority backgrounds (including but not limited to racial/ethnic backgrounds). We hope to provide a complete report in upcoming months.

TASK FORCE ESTABLISHED

NSGC's second accomplishment was to establish the Health Disparities Initiative Task Force, working in conjunction with the Diversity Subcommittee and outside groups. Task force members include **Sylvia Au, Katy Downs** (co-chair, Diversity subcommittee), **Tene Franklin, Barbara Harrison, Ari Martinez, Ilana Mittman, Kelly Ormond, Nancy Warren** and **Vince Bonham** (representing NHGRI). The group agreed that their work must address many different issues, include groups external to NSGC and focus beyond ethnic background in considering diversity and disparities.

The Task Force first reviewed the work currently done within NSGC and related groups. For example, the Diversity Subcommittee is compiling a history of the Diversity SIG and Subcommittee, as there is no comprehensive reference that summarizes its evolution, mission and activities; updating the language resources on the NSGC website (starting with Spanish); and continuing to award two graduate student scholarships, based on essays addressing diversity and genetic counseling, to help defray the cost of attending NSGC's AEC. NHGRI is developing a DVD to address overall recruitment for professions related to genetics, including genetic counseling, in a culturally competent way.

THE CHALLENGE IS ACCEPTED

It will be a challenge for NSGC to determine the most efficient ways to approach the vast issues of diversity in conjunction with our work groups, our Diversity Subcommittee and other organizations. In 2006, the Task Force plans to investigate what other groups have achieved (e.g., the American Psychological Association). We will learn about bridge programs that mentor high school students from underserved communities in science and medical fields and develop a coordinated plan to infuse cultural issues into NSGC at an organizational level.

☞To learn more:

- Health Professionals for Diversity Coalition; www.hpd-coalition.org/about.htm
- IOM report, "In the Nation's Compelling Interest: Ensuring Diversity in the Health Care Workforce;" www.iom.edu
- Sullivan Commission report, "Missing People: Minorities in the Health Professions;" http://admissions.duhs.duke.edu/SULLIVANCOMMISSION/index.cfm
- *Am J Public Health* v. 95(12), Dec 2005 – volume on genetics and health disparities
- "Closing the Gap: Solutions to Race-Based Health Disparities" offers a review of the latest literature related to health disparities, in-depth field research and comprehensive case studies; www.arc.org/Pages/pubs/closinggap.html
- AAMC report on Minorities in Medical Education; www.aamc.org/diversity/initiatives.html

Special Report

The True Meaning of Counseling

A Story Inspired by Hurricane Katrina

Claire Knoll, MS

This is the story of **Annette Patterson** and **Laura Hercher,** two genetic counselors who know how to rise to an occasion.

Annette and Laura met as students at Sarah Lawrence College in 1998. Ever since, they have been "inseparable," despite Annette moving back to her home in Dallas and Laura remaining in NY. When Annette first counseled a woman in Texas for hereditary breast cancer and then began counseling her extended family in an adjacent state, naturally Laura heard about it.

Bi-State Counseling

Annette's original patient was from Louisiana, one of the few members of a 200-strong extended family to move out of state. When discussing the patient's large number of at-risk relatives and their restricted access to genetic services, the patient pleaded with Annette to go to Louisiana and talk with the family. Fortunately, Annette's job included funds for studying hereditary cancer families. In the fall of 2000, Annette and her team took the first of many flights to St. Bernard Parish, an area just outside New Orleans, to counsel a "very close, very warm" family about breast cancer. Testing proved that their breast cancer was linked to the BRCA2 gene.

Many branches of the family lived in the same neighborhood, so counseling sessions were held in members' homes or at church after Sunday services. The sessions involved up to 20 women at a time, included lots of home cooking and addressed everything from breast self exams to screening guidelines for mutation carriers. Annette felt privileged to be accepted as "one of the gang" despite being an outsider.

Annette Patterson and Laura Hercher

A Family Exodus

When Hurricane Katrina hit New Orleans, every member of this family was affected. Their neighborhood was under water for weeks. Annette and Laura shared the same first thoughts: "Did the family survive? What will they do?" Annette called her original patient and learned that the whole family had fled to Dallas, and Annette's patient was trying to find living space for all of her 200 relatives.

A Haven in Dallas

Just by coincidence, Annette and her husband had recently purchased a new house. Because they wanted to make some structural changes, the house was standing empty. Annette and her husband hesitated before offering the use of this house to her patient, wondering how the neighbors might respond. She quickly found out. The neighbors helped her mobilize a work force, and in three days, the house was cleaned. Annette's friends and neighbors, and **Linda Robinson,** her fellow genetic counselor at UT Southwestern Medical Center, arranged donations of furniture and furnishings, "right down to the appropriate hair care items in the bathrooms," Annette recalled.

Multi-State Support

Back in NY, Laura was finding clothing for the family, including items like prosthetic-compatible bras. Again, friends and neighbors helped. Laura paid out of pocket to ship 1,000 pounds of clothing to Dallas. "The FedEx guys got to know me really well," Laura said.

Annette's husband and his co-workers also collected money so each of the 25 nuclear families received a $250 gift card to supply extra items. How did the extended family react when they had a place to shelter together? "They had a barbecue for the neighborhood – of course!" said Laura.

Filling a Need

Recently, the two families that lived in Annette's house returned to St. Bernard Parish. Both received FEMA trailers and have begun putting their lives back together. Many other family members remain in the Dallas area. Having helped fill their survival needs, Annette is again helping them obtain their "genetic" needs of counseling, mammograms and testing. Laura continues to provide support, now mostly moral, from NY. "You see a need and want to fill it," she said. "That's what we're trained to do. Plus, people in NY remembered how everybody pulled together for us in 2001, and we wanted to reciprocate."

Thank you, Annette and Laura, for reminding us that counseling doesn't stop at the office door. ❖

On the Road

The First Genetic Counseling Program in Saudi Arabia

Shelley Kennedy, MS

If you asked me in 2003 where I envisioned myself in 2006, the Kingdom of Saudi Arabia would not have entered my mind. Yet here I am, entering my third year in the Kingdom. How did this happen?

For six years, I had worked at the Hospital for Sick Children in Toronto. Our geneticist asked if I'd like to help set up the Clinical Genetics Department at King Faisal Specialist Hospital and Research Center (KFSHRC) in the Kingdom of Saudi Arabia. It seemed interesting, and I reasoned, "It's only for a year." So that October, I packed my bags and began the adventure of a lifetime.

First Impressions

The first thing I thought on my arrival was, "It's really hot!" Women in the Kingdom wear a black cloak over their clothes and cover their hair in public. I'm not required to cover my clothes or hair in the hospital, but I respect this norm outside. Another Saudi behavior dictates that in a business meeting you inquire about the other person's health and family. How rude they must have found me launching into my agenda. While I initially struggled with the culture shock, I soon settled into life as an expatriate.

In Dire Need

The Department of Medical Genetics at KFSHRC was formed in 2001. There are five geneticists, specialized in metabolic and clinical dysmorphology, all certified by the American Board of Medical Genetics. I was the first genetic counselor in the department – and in the Kingdom.

When I began attending genetics clinics, I was shocked at the volume. The dietitian follows over 1,200 patients per year, 80 with maple syrup urine disease alone. Genetic disorders are common, due to consanguinity, the relative homogeneity of the gene pool and large family size. The need for properly trained genetic counselors was evident.

Setting Up the Program

In planning the genetic counseling program, I worked with **Dr. Moeen Al-Sayed,** clinical geneticist in the Department of Medical Genetics. We encountered reams of red tape trying to secure an academic affiliation, locally and internationally, so we opted to begin the training program within the hospital. We hope to secure an academic affiliation in the future and grant our trainees a Masters degree.

At the end of my first year, we were so close to starting the program that I signed on for year two. This decision was rewarded when the KFSHRC genetic counseling program opened in March 2005. The main goal is to train Saudi Nationals to work as genetic counselors within the Kingdom. Our program is modeled after my training at the University of Cincinnati and my work experience at the University of Toronto. It is designed to meet the needs of genetic counselors within the Kingdom, such as including a course on Genetic Counseling and Islam. It also incorporates the practice based competencies established by the American Board of Genetic Counseling.

The KFSHRC Diploma

We currently have three trainees enrolled. Two entered after undergraduate school, and one worked as a science teacher. Most universities in the Kingdom do not offer genetics. As a result, our first semester is spent bringing trainees up to speed.

The Program consists of four 18-week semesters and a six-month clinical internship. I teach most of the genetic counseling courses. We have guest lectures by Obstetrics and Gynecology, Social Work and Neurosciences, and we have hands-on laboratory exposure.

For clinical training, students complete rotations in Medical Genetics, Prenatal, Preimplantation Genetic Diagnosis and Specialty Clinics (cystic fibrosis, spina bifida, hemoglobinopathies, hemophilia and cleft lip and palate). We hope to add a cancer rotation when a counselor with this expertise joins our center.

International Expansion

A variety of challenges awaits our trainees when they graduate in 2007. As a profession, genetic counseling must encourage and facilitate the training of our international colleagues, including accrediting international programs. **Janice Edward's** conference, "Genetic Counseling Education: Connecting the Global Community," to be held May 2006 in Manchester, England will engage dialogue between genetic counseling program directors from around the world (see p11).

The Future is Open

How long will I stay in Saudi Arabia? That's hard to answer. I certainly want to see our first students graduate and begin their careers. After that, the door is open – and I'm excited to see where the profession of genetic counseling will take me next. ❖

For Your Practice

Genetic Testing for Cardiovascular Disease

Heather MacLeod, MS

Clinical genetic testing for cardiovascular disease is rapidly increasing. Information obtained from genetic testing, in conjunction with personal and family history, modifies screening and treatment, including medications, imaging approaches and implanted devices like pacemakers or cardiac defibrillators (ICDs).

Information for Families

Genetic tests for cardiovascular conditions also have implications for family members. A considerable investment is made in the medical surveillance of at-risk relatives. For those shown not to carry a familial mutation, genetic testing can reduce the need for costly echo-cardiograms, electrocardiograms and MRIs. In addition, several cardiovascular diseases are associated with sudden death; early identification of those at risk may be life saving.

Dilated Cardiomyopathy (DCM)

An example of how genetic testing impacts diagnosis and risk determination is illustrated by the gene encoding Lamin A/C (LMNA). LMNA mutations cause dilated cardiomyopathy and/or limb girdle muscular dystrophy and are associated with conduction system disease and atrial fibrillation. Mutation carriers often receive pacemakers, yet this does not reduce the high rate of sudden cardiac death. This suggests that implantable defibrillators may be necessary to prevent sudden death.

Genetic testing for LMNA began in 2004. Mutations account for 5% – 8% of individuals with familial dilated cardiomyopathy. Genetic testing for desmin (DES), a gene that accounts for less than 1% of familial dilated cardiomyopathy, is now available.

Hypertrophic Cardiomyopathy

Genetic testing is available for eight genes that cause 55% – 70% of hypertrophic cardiomyopathy (HCM). Genetic testing can determine if an individual is at high risk for sudden cardiac death, by identifying a mutation in beta-myosin heavy chain (MYH7) or troponin T (TNNT2), or is at lower risk due to a mutation in myosin binding protein C (MYBPC3). Clinical genetic testing also exists for glycogen storage diseases that cause hypertrophy, such as Fabry and Danon Disease and/or a variant due to mutations in PRKAG. The eight genes implicated in hypertrophic cardiomyopathy also account for 10% – 15% of dilated cardiomyopathy, making this testing applicable for these families as well.

Long QT Syndrome

Genetic testing for Long QT syndrome became available for five of the six responsible genes in 2004. Testing identifies a mutation in 75% of cases. Mutations in specific genes influence whether an individual receives an anti-arrhythmic drug or an ICD. Providing an anti-arrhythmic drug to individuals with mutations in the SCN5A gene may increase the risk of sudden death, as SCN5A mutations account for 25% of Brugada syndrome, an arrhythmia syndrome allelic to Long QT.

ARVD

The next anticipated genetic test is for ARVD, arrhythmogenic right ventricular dysplasia. Mutations in the gene plakophilin 2 (PKP2) was found to account for 25% of cases in one paper. Untreated ARVD can cause a fatal heart rhythm irregularity. Johns Hopkins is expected to begin offering clinical PKP2 testing this spring.

Third Party Coverage

Insurance coverage for cardiovascular disease genetic testing is increasing with patient demand. Familion, the company performing clinical Long QT testing, found that most insurance companies cover the test to the limits of patients' plans (i.e., based on the deductible and co-pay). Aetna and Cigna have added Long QT syndrome as a diagnosis that should be covered under their genetic testing policies. The University of Chicago found 32% of its Lamin A/C genetic tests were covered in 2004 vs. 62% in 2005. ❖

Genetic Testing for Cardiovascular Disease

Disease	Genes Tested	Company/ Institution	2004 tests ordered	2005 tests ordered
HCM	MYH7, MYBPC3, TNNT2, TNNI3, TPM1, ACTC, MYL2, MYL3	Harvard Partners	1X	5X
DCM	Lamin A/C	University of Chicago	22	37
Long QT	KvLQT1,HERG, SCN5A, KCNE1, KCNE2	Familion	46	412

Student Corner

The Impact of Learning Abroad

Jeanie Schaller, BS


Jeanie Schaller

From June to August 2005, I participated in a multi-center clinical rotation at the Royal Children's Hospital in Melbourne, Australia. My experience was self-directed, as I worked in several clinics based on my interests. I attended prenatal, pediatric, bowel, metabolic and cancer clinics. I participated in lecture series, journal clubs, case presentations and pediatric grand rounds. My internship concluded with the Human Genetic Society of Australasia's annual education conference in Newcastle.

Public vs. Private Healthcare

One of the most novel aspects I encountered in Australia was socialized health care. Ironically, people with private insurance do not have access to more experienced doctors because there is not enough demand for doctors in the private sector; therefore, private doctors are not as well paid. Why then have private insurance? The waitlist for appointments in national health care extends for months. Those with private insurance can be seen sooner.

Genetic Testing Uptake

The health care system also affects genetic testing. Most genetic testing is paid for by the government as long as a doctor deems it necessary. While results tend to take a long time (e.g., BRCA1 and BRCA2 testing can take a year or longer), the relative ease of just "ordering" genetic testing has the potential of patients not being fully informed of the risks and benefits.

The Travel Advantage

During my internship, I engaged in thought-provoking discussions with the Australian genetic counselors. Understanding how genetic counseling in Australia is similar to and different from the US has been invaluable. I believe that international student exchanges are important for the development of well-rounded genetic counselors. ❖

Jeanie is a second year genetic counseling student at the University of South Carolina.

Building Cultural Competence through Community Involvement

Beth Dudley, BS


Beth Dudley

Working in culturally diverse communities is a great tool for promoting cultural competence in genetic counseling. I work at the Center for Minority Health (CMH), an organization designed to reduce minority health disparities in Pittsburgh. I coordinate the Family Health History Initiative, a program to educate participants, primarily African American, about the importance of family history as a risk factor for multifactorial conditions like cancer, heart disease and diabetes.

Genetic counseling students from the University of Pittsburgh meet with individuals for a family health history session that includes creating a pedigree and providing risk analysis for common diseases. After the session, we send participants a computerized copy of the pedigree and personalized health information.

Public Health Awareness

My work at CMH has helped me to explore my interest in public health genetics. A pedigree is an ideal tool to assess risk because it combines the factors of genetics, behavior and environment. As genetics becomes more integrated into public health, acknowledgement is growing for the use of family history. By identifying genetic risk factors, we can focus on lifestyle changes, like improved diet and increased physical activity, that can help reduce risk.

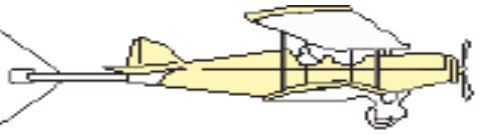
Building New Relationships

I also appreciate CMH for its ability to help build cultural competence. By going out to the community, the genetic counseling students gain insight into culturally appropriate ways to engage our participants so they will accept and act on the information we share with them. Because our participants are the experts on their family histories, they gain a sense of empowerment. These interactions create trust and allow the students and participants to better appreciate each others' role in the health care relationship. ❖

☞ www.cmh.pitt.edu.

Beth is second year genetic counseling student at the University of Pittsburgh.

MEDIA WATCH



Angela Geist, MS and Roxanne Ruzicka, MS

Winter, 2006 – Canadian Museum of Nature, "Genome"

A Canadian-made, traveling museum exhibit called, "Genome," is being featured at science museums. This spectacle examines the basics of genes and genomics, highlighting recent scientific discoveries and the impact of genomic research on society. The show includes an extensive section on genetic counseling, complete with an interactive video with **Amanda Bergner**.

January, 2006 – *Les Debrouillards*

This French-language children's magazine featured cytogeneticist **Irene Uchida** tracing her origins as a Canadian-born individual of Japanese descent who was sent to the internment camps during World War II. The article highlighted Dr. Uchida as a pioneer in the field.

January, 2006 – MTV, "True Life"

New to reality TV, this show featured the trials and tribulations of a pregnant 17 year-old girl. When her triple screen came back positive, she received genetic counseling from **Liesl Mestre.** The show filmed the genetic counseling and the couple's discussion after leaving the session.

February 1, 2006 – PBS, "African American Lives"

A four-hour documentary hosted by **Dr. Henry Louis Gates, Jr.,** the Chair of African and African-American studies at Harvard, used a combination of genealogy and genetic testing to trace some prominent African-Americans' ancestries through American history and back to Africa.

February 6, 2006 – *Newsweek*, "In Our Blood"

This article highlighted the increasing popularity of genetic genealogy – the use of DNA testing to determine from which region of the world an individual's ancestors originated. The author reviewed how some genetic diseases are more prevalent in certain ancestries and cited *National Geographic's* Genographic Project, which hopes to collect DNA samples from indigenous populations around the world to trace the origin of our species and global migration.

February 9, 2006 – Shaw Cable TV, Canada

This Edmonton-based station aired an interview with **Deepti Babu** and a patient she counseled for hereditary breast and ovarian cancer. The interview focused on the genetic counseling process, the impact of genetic test results and the importance of documenting medical family history by constructing a family tree. Deepti highlighted the NSGC website and family history tool.

February 10, 2006 – Public radio, "High Desert Forum"

Robbin Palmer was the guest on a one-hour, local call-in public radio program. She spoke about genetic counseling and genetics in general. Robbin also wrote an article called, "'Tis the season – to document family health histories," that appeared in the *Sparks Tribune* on Dec. 22, 2005.

February 12, 2006 – CNN

A little girl with Angelman syndrome and her family were featured on TV. The story mentioned the girl's symptoms, her initial lack of a diagnosis and the cause of Angelman syndrome (a deletion in her case). The family relayed their experiences with their daughter's school and their need to educate her doctors who weren't familiar with Angelman syndrome. The girl's mother is now the director of the Angelman Syndrome Foundation.

February 14, 2006 – CNN.com Health section, "Children with Down Syndrome Sought for Adoption"

According to this article, more families than ever before are seeking to adopt children with Down syndrome, especially families that already have children with Down syndrome. Down syndrome is described as "a type of retardation caused by a genetic malfunction." The common medical problems were mentioned.

February 20, 2006 – *People* magazine, "A Most Fragile Boy"

This story traced the life of a six year-old boy with epidermolysis bullosa (EB). The article discussed the boy's symptoms and special care needs. EB was described as an "incurable genetic disease," but the inheritance pattern was not mentioned.

February/March 2006 – *Homemakers* magazine, "Should You Have a Genetic Test?"

A family was followed undergoing genetic counseling and testing for hereditary breast and ovarian cancer. The article presented the process well. **Stephanie Kieffer** was the counselor who helped the family consider the implications of test results, and **Joanne Honeyford** was quoted. ❖

2006 Annual Education Conference Update

Join Us In Music City USA – Nashville

WHEN:

November 10-14: The NSGC 25th Annual Education Conference

November 9-10: Short Course, "Beyond the Genome: Health Promotion and Disease Prevention through Genetic Counseling"

WHERE:

The Gaylord Opryland Resort and Convention Center, Nashville TN
www.gaylordopryland.com

The Meetings

The 25th annual NSGC conferences will provide several exceptional educational opportunities.

The Short Course Planning Committee, chaired by **Pamela Clouser McCann, Amy Lemke, Melanie Myers, Kristin Peterson Oehlke** and **Debra Duquette,** has organized a slate of experts to discuss how innovations in genetics and genomic research are influencing health risk assessment, treatment options and disease prevention strategies.

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The AEC Plenary Committee, chaired by **Nathalie McIntosh** and **Janice Berliner,** has arranged a notable slate of presentations ranging from genomic methods for cancer risk assessment to portrayals of genetics in the media.

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Our Educational Breakout Sessions, chaired by **Brooke Smith** and **Lynn Holt,** will highlight specific topics of interest, such as current issues in reproductive genetics and ancestry-based carrier screening.

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As always, new and exciting information will come from presented abstracts and posters. Please consider submitting your work for possible presentation at the AEC (see application guidelines on p11).

The Hotel

The Gaylord Resort has hosted presidents, heads of state and visitors from around the world, embracing one and all with hospitality that reflects the true heart of the South. In soaring atriums under glass, nine sprawling acres of lush gardens and cascading waterfalls have been enhanced with an $85 million renovation. Come aboard for a lazy Delta river flatboat ride along an indoor winding river. Savor gourmet cuisine in a glorious antebellum-style mansion or one of four other restaurants inside the resort. Stroll through exclusive shops, or travel right next door to a huge shopping mall. Relax in two outdoor swimming pools, or workout at the fitness center. If you are looking for a roommate, **Carrie Guy** is organizing the roommate match.

☞ carrie.guy@kp.org

The Activities

Nashville is Music City and is full of wonderful and exciting opportunities to shop and explore. Our Logistics Committee, chaired by **Martha Dudek,** is coordinating activities for your free time. Information on hosted dinners will be available at the conference. Wellness classes and walking/running groups will be organized by NSGC members. Or you can take advantage of Relache Spa at the Gaylord Resort. Childcare programs are offered for hotel guests. There also will be opportunities for a lunch or dinner cruise on the General Jackson showboat, Nashville sightseeing tours and trips to the Grand Ole Opry and Ryman auditorium. Stay tuned for details from our Communications Committee, chaired by **Courtney Rowell.**

☞ martha.dudek@vanderbilt.edu; crowell@myriad.com

The 25th Anniversary

It is a landmark for NSGC to have offered its educational conference for 25 years, and we are planning some special events to commemorate this milestone. AEC Co-Chairs **Daragh Conrad** and **Cheryl Dickerson,** and the entire Conference Committee, look forward to bringing you this wonderful educational opportunity as well as time to reconnect with friends, classmates and colleagues. Please mark your calendars, and join us in Nashville.

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Research Network

✓ Recruiting Patients with Marker Chromosomes or Telomere Imbalances

Drs. David Ledbetter and **Christa Lese Martin** in the Department of Human Genetics at Emory University are collecting samples from individuals with marker chromosomes or telomere rearrangements of known origin for analysis in an NIH funded research project. Fine mapping studies to delineate the size of the imbalance will be carried out to develop genotype/phenotype correlations. There is no cost to participants. Results will be reported back to the referring provider.

Marker Chromosomes

Individuals with a supernumerary marker chromosome of known origin (excluding those derived from chromosome 15) and a normal or abnormal phenotype are being recruited. Identifying the origin and euchromatic content of supernumerary marker chromosomes helps determine their significance to a clinical phenotype. For cases of familial marker chromosomes, the G-banded chromosomes may look identical between child and parent. However, FISH analysis with pericentromeric probes can reveal that the child has an unbalanced composition, while the parent has a balanced composition for this region.

Telomeres

Two categories of telomere rearrangements (that were not discovered by standard chromosome analysis) are being recruited: causative of an individual's phenotype OR apparently benign familial variants that were identified in an affected individual but subsequently found to be carried by an unaffected family member. ❖

☞ ✆ 404-727-7098;
research@molecular-rulers.org;
www.molecular-rulers.org

✓ NCI and Avon Partner for Chemoprevention Studies

NCI and Avon Foundation are sponsoring two chemoprevention studies at the City of Hope Cancer Center in Duarte CA.

Biomarkers, Breast Density and Risk Reduction Perspectives

This study is seeking premenopausal women, age 21-48, at high risk for breast cancer (e.g., BRCA carriers). Subjects self-administer nasal spray containing deslorelin, a gonadotropin-releasing hormone agonist (GnRHA), along with low-dose add-back estrogen and testosterone. Endpoints include mammographic density, breast MRI and breast tissue changes pre- and post-study. We previously established that this regimen reduces mammographic density in BRCA carriers while maintaining quality of life, bone density and reproductive options.

Aromatase Inhibitor in Postmenopausal Women at Risk for Breast Cancer

This project aims to determine whether grape seed extract (GSE) lowers circulating estrogen levels in postmenopausal women, age 40-75, at any risk level. GSE, currently marketed as an antioxidant, has been shown to inhibit aromatase, suppress estrogen synthesis and inhibit mammary tumor growth in preclinical studies. ❖

☞ Katrina Lowstuter, MS,
✆ 626-256-4673 ext. 64324;
klowstuter@coh.org

☞ www.cityofhope.org/ccgp or
www.clinicaltrials.gov/ and
search for "deslorelin" and/or
"grape seed extract"

✓ Paraganglioma/ Pheochromocytoma Research

Dr. Charis Eng, Director of the Cleveland Clinic's Genomic Medicine Institute, is currently enrolling patients in the following research study: testing of the SDHB, SDHC and/or SDHD genes for people with paraganglioma(s) and/or pheochromocytoma(s). The appropriate genes are sequenced, and if testing is negative, large deletion studies are done. This study involves a blood draw and may require participant medical records and/or pathology specimens.

This testing is being done through a DNA/RNA banking protocol. As such, samples are stored for 20 years and could be used in future research, anonymously or with identifiers if the patient would like results. MEN 2 and VHL testing should be negative before enrolling.

☞ Amy G. Shealy, MS,
✆ 216-445-1251;
shealya@ccf.org

Bulletin Board

Important Deadlines

April 3rd

2006 NSGC AEC Call For Abstracts

Starting April 3 to June 2, abstracts of interest to the genetic counseling profession are being accepted for consideration as platform or poster presentations during the 2006 AEC. Students, non-members and full members of NSGC are encouraged to submit abstracts. Monetary awards will be presented for best full member and student abstracts. Instructions can be found on the NSGC website using a link to the Abstract Submission Form. ❖

☞ Abstract Committee Co-Chairs:

Stephanie Brewster, ✆ 617-355-2499; stephanie.brewster @childrens.harvard.edu

Sara Cooper, ✆ 404-778-8536; scooper@genetics.emory.edu

May 8th

2007 NSGC AEC Short Course Proposals

The Educational Subcommittee is seeking Short Course Proposals for the 2007 NSGC AEC in Kansas City MO on October 11-12. All NSGC members and SIG Chairs are urged to submit a proposal. Send submissions by May 8, 2006 to the AEC Subcommittee Chair, **Juliann Stevens.** Contact her directly for a submission form or with questions.

☞ Jmsharvey@aol.com; ✆ 412-692-5969.

May 15th

Audrey Heimler Special Projects Award Proposals

The deadline for the 2006 Audrey Heimler Special Projects Award is approaching. In the past, awardees have used funds for projects such as creating educational materials for patients, developing practice guidelines and exploring ways to involve new genetic counselors in the NSGC leadership structure. Proposals should focus on the future of the genetic counseling profession and/or the provision of genetics services. The application can be found in the members area of the NSGC website, following the "funding opportunities" link. All proposals must be submitted by May 15, 2006.

☞ Matthew Bower, AHSPA Chair; MBOWER1@fairview.org

December 31st

ABGC Recertification

Genetic counselors certified by ABGC in 1996 must recertify by December 31, 2006 to remain certified genetic counselors (CGC). Diplomates who do not have time-limited certificates are encouraged to recertify. The ABGC Administrative Office will begin accepting recertification applications July 1, 2006.

ABGC has developed several fact sheets related to recertification, which are currently posted at www.abgc.net (click on RECERTIFICATION). ❖

Pregnancy Loss and Infant Death Alliance 15th National Perinatal Bereavement Conference

October 12-15, 2006
Chicago IL

This comprehensive conference for professionals and bereaved parents focuses on parents' experiences of the death of their babies during pregnancy, birth or infancy. Hear from renowned speakers about current research, clinical guidelines and innovative programs related to compassionate care for bereaved families.

☞ www.perinatalbereavement conference.org; sponsored by PLIDA; www.plida.org

Genetic Counseling Education: Connecting the Global Community

May 15-17, 2006
Manchester, England

There are 30 genetic counseling programs in the US and 30 more in 16 other countries. Sponsored by the JEMF, this international conference will be the first time genetic counseling program directors and organizations that represent genetic counselors will meet to foster the transnational development of the genetic counseling profession. Look for a conference summary in the next issue of *Perspectives* and a copy of the proceedings in a special issue of the *Journal of Genetic Counseling*.

☞ Janice Edwards, Principal Investigator and Conference Chair; igce.med.sc.edu

Letter to the Editor

Assuming the Role of Genomic Counselor

The era of genomic medicine is upon us. Genomics was a term coined over 18 years ago[1] and describes a shift in the focus from the study of one gene and its function (genetics) to the study of the function and interaction of all genes in the human genome[2]. Has our profession considered the downstream effect of genomic medicine? Will we be calling ourselves genomic counselors instead of genetic counselors? We may not need to wonder long, as many of us are performing genomic counseling each day in our practices.

The Cancer Example

Considered a forbearer of adult onset conditions within genetic counseling, cancer genetics is also a leader in the field of genomic medicine. Those working in cancer genetics are not surprised when patients ask why they have been referred, given that their oncologists said their cancers were "not genetic." These patients are referring to testing of their tumors for genetic markers, not for a genetic predisposition. We find ourselves explaining somatic vs. germ line mutations and why genetic testing was performed on the tumor, thus embarking on genomic counseling.

A recent Cancer-SIG listserv discussion questioned our role in explaining a new "genetic test" called Oncotype DX. This test quantifies the gene expression of a specific breast tumor and, through a mathematical model, predicts the risk for recurrence of the tumor. This is genomic medicine at work, utilizing the genome to treat and manage disease. The listserv respondents were mixed in their enthusiasm to embark on counseling for such indications. Understandably, this information is not traditional in our focus on hereditary cancer syndromes. However, I agree with one respondent who wrote that most providers may not understand the technology well enough to explain it to their patients or do not have the time.

The (Near) Future

Other examples of genomic counseling include genetic testing for the cytochrome P450 genes, frequently ordered to assist in the treatment and management of patients with depression and other psychiatric illnesses. I predict, as genetic testing for cytochrome P450 genes becomes more clinically applicable, more physicians will refer their patients to genetic counselors.

We also cannot ignore that genetic markers for multifactorial conditions soon will become clinically applicable for prediction testing. With approximately 60% of the population affected by multifactorial conditions[2], genetic services will be a routine component of most patients' care.

I believe the direction of our profession into genomic counseling is both exciting and inevitable. While we likely will not need to change our job title, I think our approach to these cases will need to shift to address the complexities of genomic medicine. ❖

Katherine Hunt, MS
Mayo Clinic, Scottsdale AZ

[1] McKusick VA, Ruddle FH. A new discipline, a new name, a new journal. *Genomics*. 1987; 1:1-2.

[2] Nussbaum RL, McInnes RR, Willard HF. Thompson and Thompson *Genetics in Medicine*. Philadelphia, W.B. Saunders Company; 2001:2.

Nominations for ABGC Board of Directors

The American Board of Genetic Counseling (ABGC) is looking for genetic counselors board certified for five or more years to serve on its Board of Directors (BOD). Nominations are being accepted through April 24th from which a slate of four candidates will be chosen. Elections will take place in August for two positions starting January 1, 2007.

To make a nomination, contact **Sharon Robinson** (ABGC@genetics.faseb.org). Contact **Leslie Cohen** with questions about serving on the board (Leslie.Cohen@uhhs.com).

ABGC 2006 Nominating Committee: **Leslie Cohen,** Board Representative, **Beverly Yasher,** Chair, **Elsa Reich, Stephanie Cohen** and **Richard Dineen.** ❖

Nominations for NSGC Board of Directors

The nomination process will open soon for the NSGC Board of Director positions of President-Elect, Secretary and regional representatives for Region IV and Region V. Positions will be for a three year term (four years for the Presidential Term) starting January 1, 2007. Nominate yourself or a colleague at the Member News Section of the NSGC website (www.nsgc.org). The nomination process will close on June 16, 2006. We appreciate your help in designing a high-quality slate of candidates.

Contact **Kelly Ormond,** Chair NSGC Nominating Committee, with questions (k-ormond@northwestern.edu). ❖