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President's Beat

NSGC Brand Platform: Demonstrating the Value of Genetic Counselors

You may be asking yourself, what is a brand? Contrary to my initial beliefs, it does not involve burning symbols into cattle, and it is not just about logos. A brand is defined as "… a promise that is kept again and again because of what the brand does and what the brand believes in." For an association, a brand is synonymous to a strong organizational identity. The key questions NSGC needs to answer in defining our brand are, "What is the value of NSGC as an organization, and to whom do we want to demonstrate this value?"

To help answer these questions, the NSGC Board of Directors took part in a facilitated session on branding at our June 2007 meeting. **Cindy Kuhn**, Vice President of Marketing and Communications at SmithBucklin, led the session. Other participants included the NSGC staff and representatives from our public relations firm, PCI. During the session, Cindy led the Board through a series of brainstorming activities that helped us identify NSGC's various audiences and the current relationships to each.

For professional associations, it is essential to demonstrate the value of the organization to the membership. After all, without members, there is no professional society. NSGC will continue to focus on ensuring that we maintain our value to our members. However, the membership may not be the primary audience for whom the brand is developed. During the branding session, we considered various external audiences to whom we might want to demonstrate value including physicians, physician organizations, other healthcare providers and/or their organizations, health insurers and industry. Ultimately, the Board determined that NSGC would most benefit if it were able to develop a brand that demonstrated the value of NSGC and its genetic counselor members to physicians.

There were several reasons for this decision.

> 1) Physicians are critical to genetic counselors' access to patients (and, potentially, to jobs). Time/cost efficiencies and concerns about liability are real issues that physicians face in trying to integrate genetics into their practices, and these are issues that genetic counselors can help solve.

2) Many physicians still are not fully aware of the value of genetic counseling.

3) Some physicians who are aware have misconceptions about which patients may benefit and the scope of what genetic counselors can do to enhance the care they provide.

With these reasons in mind, the Board developed a positioning statement that defines the critical elements of our brand. Our statement aims to position NSGC's genetic counselor members as the "go-to" healthcare providers for physicians who seek to provide their patients with comprehensive care incorporating the latest practices in genetics. The position identifies our value – our expertise and our ability to provide services in a time and cost-efficient manner that reduces liability – and strives to differentiate us from other healthcare providers who do not possess our specialized training. The characteristics we want this brand to portray include efficiency, caring, communication, rapport, scientific knowledge, versatility, collaboration and support.

How does this translate into member value? Actively promoting the genetic counseling profession will lead to increased recognition of genetic counselors as integral members of the healthcare delivery team. As such, this will lead to increased opportunities for our members. In addition, NSGC will ensure that its members are equipped to deliver high quality services and to leverage the increasing demand for services through efforts such as continuing education, tools to enhance practice and networking opportunities to augment professional advancement. Notably, these outcomes are in line with our strategic initiatives to position genetic counselors as key players in the integration of genetics into healthcare and to promote an organizational culture that will meet the evolving needs of genetic counselors.

The NSGC Board approved the brand platform summarized briefly above at the October Board of Directors meeting in Kansas City. The next step will be for the SmithBucklin Marketing and Communications team to develop a two-to-three year brand marketing strategic and tactical plan. This plan will include concrete items such as changes in our logo, web appearance and published materials. We also will explore ways to demonstrate our stated value through evidence-based research. Branding is an exciting project that will support and help inform all of our other strategic initiatives, including our educational, strategic, planning and billing and reimbursement efforts. Stay tuned for more updates!

Angela Trepanier

Angela Trepanier, MS, CGC
NSGC President-Elect

Contributed Feature

An Immersion Course in the Democratic Process

By Lori Williamson, MS, CGC, LGC, University of Oklahoma Health Sciences Center

For twelve years, GINA, the Genetic Information Nondiscrimination Act (H.R.493, S.358), has attempted to pass Congress. This bill would "prohibit discrimination on the basis of genetic information with respect to health insurance and employment" (http://www.geneticalliance.org/ws_display.asp?filter=policy.leg.nondiscrim). On April 25, 2007, GINA passed the House of Representatives by a vote of 420 to three. This was the **first time** that GINA had ever passed the House. The bill also had many co-sponsors in the Senate and was predicted to pass. However, in late July **Senator Tom Coburn** (R-Oklahoma) placed a hold on GINA. A hold is an "informal practice by which a Senator informs his or her floor leader that he or she does not wish a particular bill or other measure to reach the floor for consideration" (http://www.senate.gov/reference/glossary_term/hold.htm). This hold prevents the full Senate from expeditiously considering the bill.

The hold on GINA happened to coincide with the Genetic Alliance's "Genetics Day on the Hill" event on April 26, 2007. Given the presence of genetics advocates in Washington, DC that week and the bipartisan support for the bill, hopes were high that

the hold could be reversed, especially if constituents from Oklahoma became involved. The series of events that follow trace my immersion into this political arena.

Wednesday, July 25

7:40 a.m. I slog into work, tired and ready for Friday. I open up e-mail. There's a message from the NSGC listserv, "NSGC Action Alert: Tell Senator Coburn to Release His Hold on GINA!" The next email is from Genetic Alliance, "YOUR HELP IS NEEDED." My stomach flips. They request that I come to Washington, DC as a genetic counselor practicing in Oklahoma to meet with Coburn. Personally, I think this is overkill. How would my going to DC have any effect? But I write back that I'm willing to go, if needed. I note that **John Mulvihill** – our section chief – is in Delaware on vacation and would probably be a better candidate… you know, doctor to doctor (Coburn is also a physician).

10:30 a.m. I get a response from Genetic Alliance. "We need both of you, and we are contacting a patient in Oklahoma, too. Can you come?" By now, I clearly sense the urgency.

12:30 p.m. I am in the car en route to my home 45 minutes away to pack, then another hour-plus to the airport. I cannot believe this! What am I going to say? I'm nervous. I'm packed in 30 minutes.

5:00 p.m. My plane lands in Dallas for a connecting flight, and I meet **Dennis**, the patient from Oklahoma. We both confess we are a bit nervous. Dennis has alpha-1-antitrypsin deficiency but is able to respond to this invitation due to his new set of transplanted lungs. We share our backgrounds and our efforts at recruiting other Oklahomans to contact Coburn's office.

Thursday, July 26

12:30 a.m. Dennis and I arrive at the hotel. (John is due to arrive in the morning by car.)

10:00 a.m. John, Dennis and I meet with Genetic Alliance and their advisors for a briefing. (Hmm, this is a big deal). The plan is first to meet with **Senator McConnell's** office because he is the Minority Leader. Maybe he can enlighten Coburn. McConnell's staffer doubts that pressure from senior members of Congress will have any influence.

12:00 p.m. Dennis, John and I place separate calls to Coburn's office requesting a meeting with him and not his staffer. We are denied.

3:00 p.m. We arrive at Coburn's office. The contingent includes representatives from Genetic Alliance, the American Society of Human Genetics, employee rights, industry and us Oklahoma folks. In walks the senior staffer who works on this bill; she has delayed a flight to her best friend's wedding. (Okay, now I get that visiting a Congressman in DC is a very big deal). Dennis takes the lead and shares his story. I tell a story about the 20-year-old from Shawnee, Oklahoma who has to decide about *BRCA1/2* testing now or postpone until she is fully employed and has her own insurance, meanwhile implementing a high-risk surveillance protocol. Most of the conversation is "in the weeds" as they say in DC, meaning that the conversation quickly moves to the technical minutia that legal experts wrangle over. I'm not sure our personal stories matter. Our 20-minute time allowance expands to 40. We are assured Coburn is in favor of the bill's intent but has two technical issues with the bill. They say these issues will get worked out before August recess.

5:00 p.m. Sharon Terry, President/CEO of Genetic Alliance, receives a call. **Senator Burr** from North Carolina has now placed a hold on the bill. She receives another call from an unidentified Senate staffer, "What is going on in Coburn's office?" WOW! Word of our meeting is already rippling across the Hill.

Back in Oklahoma, Friday, July 27

1:30 p.m. I request a meeting with Senator Coburn over August recess regarding the bill. The only option that I'm given is to meet in his Tulsa office (two hours away) at 3:30 p.m. on August 7. I take it.

Tulsa, Sunday, August 7

3:30 p.m. I'm REALLY nervous. I'm meeting with THE man… I've never met with a U.S. Senator before. (Breathe.) I'm seated, and I acknowledge my nervousness to him. "Just say it," he orders. "Okay, I respectfully request that you release your hold on GINA." He erupts. (Just breathe.) I let him talk. My time slot is slipping by, so I interrupt, "Senator, I cannot debate the technical aspects of this bill. I can only tell you that this is a very important issue for patients. And, could you please explain why you voted for the bill in the last Congress if you have a hold on it now?" "I have never voted for this bill! You genetics people are disseminating misinformation!" (Oh, good grief! Do I have my facts incorrect? Swallow hard). "You need to check your facts and think for yourself," he adds. I agree and explain that I am doing just that by coming to him to learn firsthand about the issues preventing him from supporting this bill. We begin to repeat our positions. I pull out a book, *The Language of God*, by **Francis Collins** and give it to him. Forty minutes later, I leave the office feeling frustrated and yet empowered. I immediately seek information regarding Coburn's voting history on this bill. He DID vote for this bill in the 109th Congress!

Monday. August 22

9:00 a.m. I email my follow-up letter to Coburn and include the information about him actually voting for the bill and my request for clarification. Ten minutes later, my telephone rings. It is Coburn's senior staffer calling to explain what Coburn meant by, "I've never voted for GINA." She explains that the bill in the previous Congress was a little bit different from the current bill. She cannot explain any more than to provide the general statement.

Looking Back

My experience with the legislative process has taught me a few lessons:

1) legislators are approachable
2) postal mail takes about two months to reach a legislator because it first goes to a clearinghouse (for security) in Ohio before being sent to Washington, DC
3) visits to a legislator IN DC send a big message about the importance of the issue to you
4) the democratic process is not what I learned in school.

Despite the butterflies in my stomach, the time taken from my schedule, my disappointment in learning the real life cycle of a bill and failure to change Coburn's actions, I have no regrets in my legislative adventure. The experience was energizing, grounding and humbling. The vitality and advocacy that I felt as a new counselor 15 years ago merged with my perspective and maturity as an experienced counselor. Am I now destined to become a legislative guru? Not likely. However, I pay more attention to legislation, and I have no hesitation to contact a state or U.S.

legislator. I encourage the "young" and "seasoned" counselors alike to raise your voices. We have something to say.

For Your Practice Special Series: Cases in Expanded Metabolic Screening

This is the fourth article in a series presented by the Metabolism/Lysosomal Storage Disease SIG in response to the expanded newborn screening panel developed in 2005 by the American College of Medical Genetics' Newborn Screening Expert Group. The expanded panel comprises 29 conditions to be tested by all state newborn screening programs. Perspectives *is highlighting several lesser-known genetic conditions now included in newborn screening to help genetic counselors as they come face-to-face with these diseases.*

CASE 4: Cobalamin C (CblC) Deficiency: Combined Methymalonic Acidemia and Homocystinuria

By Dawn Peck, MS, CGC

DISEASE REVIEW

Biochemistry: Deficiency in cobalamin C results in the body's inability to convert cobalamin (vitamin B12) into the active coenzymes adenosylcobalamin and methylcobalamin. Deficiency of these coenzymes leads to decreased or absent activity of the enzymes methylmalonyl-CoA mutase and methionine synthetase, resulting in elevated levels of methylmalonic acid and homocysteine. Classified as an organic acid disorder, elevated levels of propionylcarnitine (C3 acylcarnitine) are commonly seen on newborn screening.

Genetics: Autosomal recessive; caused by mutations in the MMACHC gene located at 1p34.1. The most common mutation, 271dupA, accounts for up to 40% of disease-causing alleles. Ethnic-specific mutations have been reported, and limited genotype-phenotype correlations exist. Clinical testing is available at a few laboratories. Seven other cobalamin defects exist and are classified based on elevated analytes and cell complementation studies.

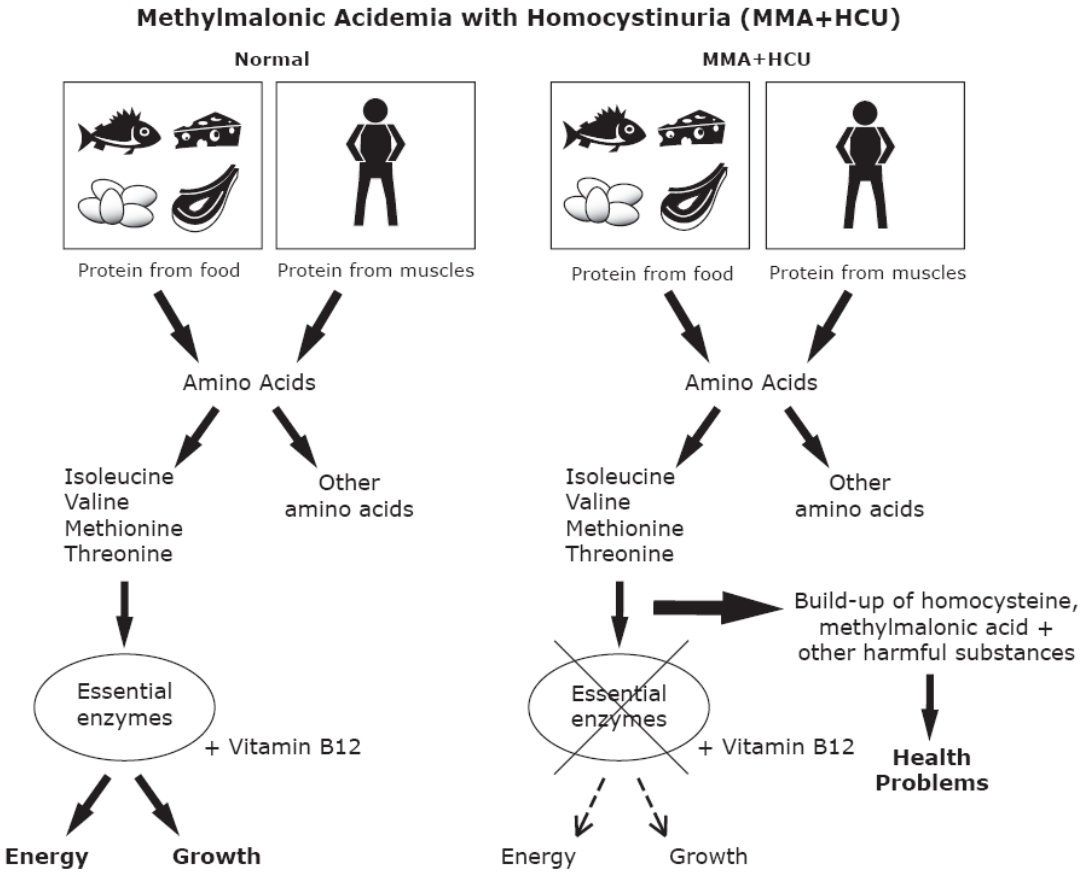
Incidence: Approximately 1 in 48,000 for all cobalamin metabolism defects. CblC deficiency is the most common defect, with over 300 patients reported.

Natural History: First reported in 1969 by **Mudd et al.**, the majority of individuals with CblC deficiency typically present with symptoms in the first few months of life.

Characteristic features of early onset disease include poor feeding, failure to thrive, hypotonia and metabolic acidosis and may also include seizures, retinopathy, developmental delay, megaloblastic anemia or thrombosis. Individuals with adolescent or adult onset typically present with acute neuropsychiatric symptoms such as confusion, dementia, tremors and weakness. Clinical presentation and outcome is variable, but early onset patients typically have a worse clinical course.

A report published in 2006 by **Morel et al.** looked at genotype-phenotype correlations in 37 published case reports. This study identified some ethnic specific origins of mutations, including an increased incidence of the 331C>T mutation in Cajun and French-Canadian patients, and the 394C>T mutation in Asiatic-Indian/Pakistani/Middle Eastern patients. Common mutations in Hispanic patients also have been reported.

Limited information has been published on outcomes of patients with CblC detected by newborn screening. Early diagnosis and treatment with hydroxycobalamin injections, a protein restricted diet and carnitine, folate and betaine supplementation may prevent complications of this disease.



Genetic Counseling – Positive Newborn Screening in Hispanic Siblings

A premature, 28-week Hispanic female infant was seen for consultation following an elevated level of propionylcarnitine on her initial newborn screen. The differential diagnosis included propionic acidemia, methylmalonic acidemia, vitamin B12/cobalamin defects or severe maternal B12 deficiency (often associated with a strict vegetarian diet). The initial confirmatory testing included serum short chain fatty acids, which revealed a mild elevation of propionate of 38 mmol/l (normal is less than 10), elevated serum methylmalonic acid of 15.97 umol/L (normal is less than 0.4) and elevated homocysteine of 42 mmol/L (normal is less than 15). Acylcarnitine analysis revealed an elevated propionylcarnitine level of 27.5 (normal is less than 1.12). Serum methionine was low. Urine organic acids demonstrated elevated levels of methylmalonic and methylcitric acids. A defect in cobalamin metabolism was suspected because of the above abnormalities. Intramuscular hydroxycobalamin injections were started.

Confirmatory testing for cobalamin disorders requires cell complementation studies on cultured skin fibroblasts. Turnaround time for results is up to eight weeks. This baby remained in our NICU for seven weeks, with a suspected diagnosis of CblC or D deficiency. Accordingly, counseling about a specific diagnosis was impossible at our initial visit.

Clinically the baby exhibited no symptoms other than mild acidosis that corrected with treatment. Although the family had lived in the United States for five years, they spoke no English. Our translator was unable to adequately translate many of the relevant biochemical terms into Spanish, and the mother, having only an eighth grade education, had a hard time grasping many of the genetic concepts. At the time of the initial visit, she reported that the father of the baby was unknown. Her diet during pregnancy also was in question, as she was unable to confirm if she was a vegetarian. The family was counseled about the variable nature of the diagnosis and that data on long term outcome was lacking.

Cell complementation studies were consistent with CblC deficiency, and the diagnosis was confirmed. However, mutation testing was performed and demonstrated only one copy of the 482G>A mutation. Clinically the baby was doing well and her methylmalonic acid and homocysteine levels dropped into the normal range with only hydroxycobalamin injections as treatment. She was discharged to go home, and her injections and follow-up lab work were to be provided by a local physician.

At the baby's follow-up visit with us at six months of age, she was thriving and doing well. The mother asked if the baby would need to be on injections and treatment her entire life. She stated that her family in Mexico had never heard of something like this, so it couldn't be true. She also reported that it was difficult to see her baby have shots so frequently. We counseled her about the diagnosis and treatment again but could not be sure how well she understood because of the language barrier. In addition, the mother was pregnant again and had an estimated due date of six months from the visit. Since the

mother could not confirm the identity of the affected baby's father, we were unable to provide her with an accurate recurrence risk for the current pregnancy.

About six months later, we received a call from the newborn screening lab regarding a positive screen with a familiar name. The call concerned the new sibling of our patient. The new baby boy had a propionylcarnitine level that was slightly above our cutoff. A call was made to the office of the physician of record with recommendations for diagnostic studies. This physician, who was not the family's primary pediatrician, chose to do a repeat screen rather than the recommended diagnostic studies. The repeat screen had a normal propionylcarnitine level, and no further tests were performed. At the new baby's two-week check-up with the family's usual pediatrician, he had begun having trouble with feeding and vomiting. The family pediatrician obtained methylmalonic acid and homocysteine levels, which were both extremely elevated. The levels confirmed that the new baby was affected with CblC deficiency. Treatment was started, and his clinical symptoms improved.

The remainder of this case proved difficult because these siblings had inconsistent responses to treatment. The first child's treatment levels became unstable at about one year of age, correlating with increased food intake. She ended up on a regimen including injections three times weekly, daily supplements with betaine, carnitine and folate and a low-protein diet. The younger child's levels were not as responsive to the medication regimen, and he was started on a low-protein diet by three months of age. As the daughter's diet wasn't changed until one year of life, the family questioned the lack of consistency between the siblings. In addition, while the treatment levels of methylmalonic acid and homocysteine were above the normal range in both patients, neither had any severe clinical symptoms, and symptoms prior to diagnosis were different. With the lack of knowledge about the outcome in patients diagnosed by newborn screening, we were unable to give the family an accurate prediction for outcome with age.

Teaching Lessons

1. Limited Knowledge of Natural History

Although there are a few reports of long-term outcome in patients with CblC deficiency, data is lacking on the outcomes of patients diagnosed by newborn screening. One could assume that early diagnosis and treatment should prevent complications of the disease, yet there is limited data to back up this hypothesis. This uncertainty proves frustrating for the families of children with these diagnoses who watch their children undergo weekly injections without the certainty that it will ultimately prevent any complications, such as psychiatric disease.

2. Limitations of Newborn Screening

Many of us rely on newborn screening tests to predict a specific diagnosis. However, with some of the disorders detected on newborn screening, second screening is clearly

not recommended, as certain analytes, including propionylcarnitine, characteristically elevate and fall at certain time points. The importance of confirmatory testing is quite clear, and with the rarity of many of these conditions, primary care physicians need education about the benefits and limits associated with newborn screening. With this case, the second child's repeat screen had a "normal" propionylcarnitine level, which may have proved confusing for the family as well as the ordering physician. If we had not known the history of the already affected sibling, the results of the second screen would have delayed the diagnosis until the onset of symptoms.

3. Language/Cultural Barriers

When there is a language difference between the medical staff and the patient, it is difficult to assess how well the patient is grasping the information that you are providing. The fact that the mother only had an eighth grade education and had not been introduced to many medical concepts until her children were diagnosed made it even more difficult to adequately counsel her. The lack of certified medical translators and written information in languages other than English also makes counseling a challenge. In addition, having relatives in another country who had never heard of this disorder made it difficult for this family to understand why it happened to them. Enhanced awareness of cultural differences and medical beliefs relevant to this family's ethnic background may have helped this family understand and accept the diagnosis made in their children.

Career Watch

On-Line Clinical Supervision Course for Genetic Counselors

By: Becky Butler, MSSW, LCSW, University of Arkansas for Medical Sciences and Lori Williamson, MS, CGC, LGC, University of Oklahoma Health Sciences Center

"Supervision is an essential component of genetic counseling education, as it is in the preparation for any health care profession. It serves two primary purposes: promoting the professional development of student supervisees and ensuring the continued provision of quality client services." (Baker et al., 1998)

Training clinical supervisors in the health professions has emerged as an important issue in the last decade (Haynes et al., 2003). The literature on genetic counseling student supervision, however, indicates a lack of standards or guidelines, limited training for supervisors and great variation in supervision methods (Hendrickson et al., 2002; Lindh, et al., 2003). Over the last decade, the medical, nursing and mental health training programs have begun to address these same issues in their long-established professions.

The Need to Set Standards

The American Board of Genetic Counseling (ABGC) requires all logbook cases to be obtained under the supervision of a certified genetic counselor or geneticist (http://www/abgc.net). Training programs use professional competencies for genetic counselors as a core guide for educating genetic counseling students (ABGC, 1996). To date, however, no competencies, guidelines or formal educational programs have been established for clinical supervisors of genetic counseling students. In contrast, in the mental health professions, accrediting bodies require supervision training in some of the graduate curricula and endorse supervision as a professional competency. Some professional boards even require a special license for supervisors (Bernard and Goodyear, 2004) and require a minimum number of years in practice before supervising (Council of Social Work Education, 2007).

Training Course for Supervisor Instructors

To help fill the lack in supervisory education for instructors of genetic counseling students, a pilot clinical supervision course was generated by two academic medical institutions in the Heartland, the University of Oklahoma Health Sciences Center (OUHSC) and the University of Arkansas for Medical Sciences (UAMS). UAMS houses a Genetic Counseling Department that administers the Mid-America Genetic Education Consortium (MAGEC). Faculty members at UAMS teach courses using distant education (DE) technologies, lending to the use of this technology for the pilot study. DE also was beneficial to the pilot study because the faculty participants were scattered over several states.

The eight-week course started the first week of March 2007 and was posted in WebCT on the UAMS server. Participants were given security clearance by the IT Department at UAMS. Several supervisors were walked through WebCT and parts of the course in order to facilitate use and comfort with the online format.

Results and Evaluation

Twenty-three genetic counselors from nine institutions in six states signed up for the course, and of these, twelve started the program. Half of these participants completed the course. The biggest barrier to finishing the course was personal workload. Several institutions had vacancies in genetic counseling positions, and existing staff was pulling extra clinic duty. The course was scheduled to end the last week of April but was open until the last week of May to accommodate the course directors and supervisors with heavy schedules. Aside from clinical work, many of the supervisors also had other commitments, making them late in posting content or discussions on several occasions.

Another barrier to completing the course was the participants' lack of experience in DE. In addition, genetic counselors supervising students for the first time were more motivated to finish the course than those who already had some experience. Lastly, because the course was a pilot, we had not applied for continuing education credits, which would have enticed more supervisors to participate.

Participants rated their overall satisfaction with the course at 4.0 on a Likert scale of 1-5, with 5 being "strongly agree." The strength of the course was described by one of the participants as a "good overview of supervision process." Those participants who had experience in supervision, either of staff or students, did not find the course as helpful as those with no supervision experience. Satisfaction with the web site received the lowest score of 3.50, due to course content posted in numerous documents and subsequent problems with downloading.

An Improved Program

The lessons learned from this pilot study have lead to major changes in the supervisors' training course. Content has been moved from attachments to the web-based platform. The handbook and forms are better integrated into the weekly sessions. Activities that were viewed as "busy work" are being replaced with videotaped case scenarios. The focus of the course is refined to genetic counselors who have recently received their certification or who are new supervisors. More experienced genetic counselors are welcome to take the course, but we plan to have a two-hour web-based presentation on a variety of supervision topics annually to meet the needs of those with more experience.

Registration Open Soon

Our Clinical Supervisor's Training Course will be offered again in Spring 2008. The eight-hour, web-based course will start the first of February, with two orientation sessions in January for those who are new to DE. We are applying for CEUs as an added attraction. Supervisors for the Oklahoma and Arkansas programs have priority, but enrollment is open to supervisors from other programs on a first come, first served basis. A nominal fee will be charged and is due by the registration deadline on February 1.

For more information, contact: Lori-Williamson@ouhsc.edu or bbbutler@uams.edu.

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NSGC News

Meet Your 2008 NSGC Board of Directors

Below are the new (*) and returning leaders for NSGC in 2008. New members of the Board of Directors take office on January 1. All of these people have been selected by YOU – the membership – and they want to hear from you. Keep this list handy in the coming year to email your leadership with your questions, ideas and accomplishments.

Executive Committee
President: **Angela Trepanier, MS, CGC**
President-elect: **Steven Keiles, MS, CGC**
Past President: **Catherine A. Wicklund, MS, CGC**
Secretary/Treasurer: **Susan Manley, MS, CGC**
Secretary/Treasurer-elect: **Peter J. Levonian, MS, CGC**
Executive Director (non-voting): **Kristen Smith, CAE**

Directors-At-Large
Renee Chard, MS, CGC
Karen Copeland, MS, MBA, CGC
Fiona M. Field, MS, CGC
Brenda Finucane, MS, CGC
Tene Hamilton Franklin, MS
Karen Heller, MS, CGC
Elizabeth A. Leeth, MS, CGC

Congratulations to the 2007 Leadership Award Recipients

The NSGC Awards Committee would like to thank all members that nominated their colleagues during the 2007 Call for Nominations. We are pleased to announce the

winners of the 2007 Natalie Weissberger Paul and Regional Leadership Awards. Please congratulate the following individuals on their accomplishments!

Natalie Weissberger Paul Award: **Robin Bennett, MS, CGC**
Region I Leadership Award: **Stephanie Brewster, MS, CGC**
Region II Leadership Award: **Kathleen Valverde, MS, CGC**
Region III Leadership Award: **Courtney Rowell, MS**
Region IV Leadership Award: **Becky Butler, MS**
Region V Leadership Award: **Karen Heller, MS, CGC**
Region VI Leadership Award: **Michelle Fox, MS, CGC**

Stay tuned for announcements regarding the 2008 Call for Award Nominations this Spring.

Get Involved in NSGC - Online Leadership and Volunteer Development Resources for Members

NSGC recently launched a new Leadership/Volunteer Development program at the Annual Education Conference in Kansas City. We are pleased to announce that these resources also are available in the Members Only section of the NSGC website. This gives all members, regardless of their ability to attend the AEC, the opportunity to learn about NSGC leadership and volunteer opportunities.

This new Leadership/Development resource page will provide a year-round opportunity for personal leadership development, resources for volunteers and information on how to submit your name, skills and interests to NSGC's new Volunteer Database.

NSGC Volunteer Database

NSGC is developing a comprehensive database of volunteers who are interested in participating on NSGC committees, being contacted for special initiatives or working on time-limited projects in specific interest areas. NSGC members interested in volunteering will be asked to complete a Willingness to Serve Interest Inventory, indicating your skills, areas of interest or expertise and your desired time commitment.

This form is available on the Leadership/Volunteer Development web page at http://www.nsgc.org/members_only/leader_volunteer_program/index.cfm. You can submit or change your information at any time by completing this form and returning it to the Executive Office.

Getting Involved in Committees, Task Forces & Special Projects

The self-identification of skills and interests by members is extremely important because committee Chairs will now be appointing members of their committees based on the projects the committee will address during the year. By limiting Chair terms to one or

two years and appointing new committee members each year based on the work to be done, we will be calling on greater numbers of members over time and ensuring that if you are asked to join a committee it will be a meaningful experience.

Outside of committee appointments, there is plenty of work to be done on behalf of NSGC. The Board and all committee Chairs will be enlisting the help and expertise of members for the Task Forces and numerous special projects that come up throughout the year. The easiest way for members to identify themselves as a potential volunteer or resource will be to ensure that you have submitted your name to the Volunteer Database by completing the NSGC Willingness to Serve Interest Inventory. While NSGC leaders will always reach out to members whom they know have expertise or time for special projects, the Volunteer Database will be a primary resource for identification of members who may be interested in special projects as they arise.

NSGC has a strong tradition of volunteer involvement by members at all experience levels. The involvement of our members has made NSGC the vibrant and exciting organization it is today, and we are looking forward to ensuring that this tradition continues by providing volunteer and leadership experiences that are both personally and professionally rewarding.

Legislation Report

Update on Increasing Access to Care for Genetic Counseling Services

By Flavia Facio, MS, CGC

During the recent AEC in Kansas City, the Billing and Reimbursement Task Force presented an annual update regarding our profession's top priority: to improve access to genetic counselors while allowing direct reimbursement of services.

The Three-Prong Approach

Leslie Cohen and **Cheryl Harper** presented an update on NSGC's efforts to date regarding the Three-Prong Approach:

> **PRONG 1**: **Pursue Federal legislation to include genetic counselors as providers under Medicare.** Legislation has been drafted. In addition, we have developed supporting documents that we can offer to legislators when we speak with them. These resources include a handout explaining who genetic counselors are and what we do and a paper delineating our value. Congressional champions, co-sponsors and stakeholders of this bill are being identified, and members of NSGC have visited with over 40 legislators. The plan is to introduce this bill in

2008. Although the bill specifically is aimed at gaining recognition for genetic counselors under Medicare, third party payers (private insurers) often follow the guidelines put in place for Medicare.

Legislation related to health care is politically charged and often contentious. NSGC recognizes that some compromises may need to be made as we proceed through this process. For more details about the legislative process and these issues, please read the FAQ that can be found at http://www.nsgc.org/publicpolicy/breducation.cfm.

PRONG 2: **State Licensure.** In parallel to the proposed federal approach, NSGC continues to support organized efforts to obtain licensure at the state level. Six states now have licensure laws, with Utah and Oklahoma being the only states so far in which licenses are available. The Licensure Subcommittee has created two documents to assist members pursuing licensure: *Guiding Principles* and *Model Legislative Language*. Both documents can be found on the NSGC website under State Licensing and Federal Advocacy. As a reminder, licensure grants are available to assist state groups in their pursuits. Award cycles occur in the Spring and Fall. In addition, rolling grants are awarded throughout the year. In order for grant monies to be awarded, bill language needs to be reviewed and approved by the Licensure Subcommittee to ensure that the proposed legislation is in line with NSGC's Guiding Principles. What happens at the state level can have ramifications for the entire country by setting precedent. For this reason, we encourage members to utilize the expertise of the Licensure Subcommittee as well as that of **John Richardson**, NSGC's Director of Government Relations. John is available to answer questions about licensure and to help you navigate the legislative process.

PRONG 3: **Engage Third Party Payers (TPPs).** After the federal bill gains momentum, efforts will focus on TPPs. Until then, we encourage genetic counselors to continue meeting with their local payers to determine the best way to bill and gain reimbursement for their services.

Governance Changes

As NSGC has reconfigured its governance structure, the Professional Issues Committee is getting a new name. Starting in 2008, the Genetic Counseling Access and Service Delivery Committee (GCASD) will oversee issues related to billing, reimbursement, licensure and access. In addition, this committee will be responsible for creating and updating practice guidelines, which were part of the Genetic Services Delivery Committee. Leslie Cohen will chair the GCASD committee.

96040 CPT Code

It has been one year since the genetic counseling CPT code was created. The CPT working group has designed a survey to assess the use of this code. They are interested

in finding out who has tried to use it, who has been successful, who has not been successful and why. Please look for this survey in the near future. Your participation is very important, whether or not you have used this code.

Interested in learning more about the legislative process? Log on to NSGC.org and click on the link labeled State Licensing and Federal Advocacy on the NSGC home page.

Stay tuned for articles addressing B&R and Licensure topics in upcoming 2008 issues of *Perspectives*.

Student Forum

Experiences in the Down Syndrome Clinic at Children's Hospital

By Nancy G. Slate, BA, BFA

On acceptance to the Brandeis Genetic Counseling Program two years ago, I received a summer reading list. One of our books, *No Pity*, by **Joseph Shapiro**, chronicles the history of the disability movement. The idea behind the summer readings was to help students understand the legal struggles and also the daily challenges of living with a disability. Even before our formal curriculum education began, an emphasis on disability awareness would frame our training as genetic counselors.

Disability Awareness

The Brandeis program is dedicated to the memory of **Andreas Tsipis** (the son of program founder **Judith Tsipis**) who had Canavan disease. The environment of the Brandeis Genetic Counseling Program focuses on disabilities and the importance attached to interaction with individuals and families in addition to knowledge of their genetic diseases. The day-to-day interaction with people who have a disability gives students a first-hand experience of what is important to these families and also how the families cope. In the first semester, each student is assigned to a placement where learning focuses on a specific condition.

My placement was at the Down Syndrome Clinic at Children's Hospital in Boston, Massachusetts. The program is coordinated by **Angela Lombardo**, and the director is **Dr. Allen Crocker**. Families who have a child with Down syndrome may enroll their child in the program at birth and come in to the clinic every six months until the child reaches the age of three. The program offers access to many specialists including a nutritionist, an occupational therapist, a physical therapist, a speech therapist, an ophthalmologist, a dentist and a pediatrician.

I came weekly and each time was matched with a different family. I spent the morning and part of the afternoon shadowing them as they received their services. Because the program sees children for an extended period of their early lives, I had a chance to observe children at every age: a little after birth, six months, one year, 18 months, 24 months, 30 months and three years. In spending time with the families, I learned how variable the Down syndrome spectrum can be.

The Power of Early Intervention

I was privileged to see how early intervention and knowledge helped the children progress while giving their parents a forum to process concerns, ask questions and learn new specialized approaches to care for their children.

I saw the speech therapist suggest innovative ways to use signing to communicate, helping to alleviate frustration in a child who in the past would have been unable to interact with family members.

I saw a nutritionist recommend a cup with a cutout for the nose to help make drinking easier.

A physical therapist showed parents of younger children how to strengthen hypotonic muscles and how to help older children walk down stairs and learn to navigate uneven surfaces.

The occupational therapist worked to enhance fine motor skills with infants and older children. She started with the grasping of objects for infants and continued with utensil usage for older children.

The dentist, at the first appointment, would check both the mother's bacterial level and the child's, explaining that the mom's bacterial count would affect her child's dental health. The dentist had a very quiet demeanor and calmly was able to clean the children's teeth, setting the stage for future successful checkups. This is very important, as children with Down syndrome have small mouths, causing the tongue to protrude and making a mouth exam challenging. The eye and medical exams also enabled the parents to have a proactive approach to their child's ongoing healthcare by providing additional medical personnel.

Another advantage of the program was that the six-month cycle often had families returning together on the same appointment days. Friendships soon evolved, enabling families to socialize together outside the clinic.

Intensified Learning

By allowing students to be a part of a clinic that focuses on a specific syndrome, students can feel confident when asked about that syndrome in a counseling session. I know that I have a deeper outlook on Down syndrome. If I had only used my textbook knowledge, I

would have failed to understand how variable the syndrome can be and the complexity of daily living. Now when I talk about Down syndrome in a session, I see the many faces of these unique children and their families who allowed me to spend time with them, and I remember with fondness watching the children achieve new milestones.

Myriad Internship Offers Unique Experiences in Genetic Counseling

By NSGC Executive Office

In an effort to provide experiences in expanded genetic counseling roles and exposure to different models for the delivery of genetic services, the National Society of Genetic Counselors (NSGC) and Myriad Genetic Laboratories, Inc. partnered to offer three six-day student rotations during the summer of 2007. The rotations took place at Myriad's headquarters in Salt Lake City, Utah and offered in-depth experience in hereditary cancer and laboratory processes, including exposure to the multitude of ways genetic counselors contribute in a diagnostic laboratory setting.

The three students selected for this rotation were:

- **Kristen Lipscomb Sund**, University of Cincinnati
- **Amanda Eppolito**, Mount Sinai in New York
- **Tara Sousa**, Boston University.

Each student was asked, "What significantly did you learn from your internship with Myriad?"

Kristen Lipscomb Sund: "The internship at Myriad was an excellent opportunity to see genetic counseling from a perspective outside of the traditional clinic setting. For me, it was surprising to see the number of roles filled by genetic counselors at Myriad. I was expecting one or two different positions, and I came across at least six within the company, ranging from a more typical role (the Professional Support Specialists and Regional Medical Specialists), to Account Executives, marketing, management and research. Talking to genetic counselors in each of these areas made me realize that there really is something very unique about our training that makes us valuable to a variety of fields. In each of these conversations, there seemed to be a common thread that brought them to their alternative careers. Several of the counselors saw patients before starting at Myriad but felt that they could have a broader impact on patients' lives by working with and advocating for a population of cancer patients instead of one patient at a time."

Amanda Eppolito: "Entering into my time at Myriad Genetic Laboratories, I expected to learn about hereditary cancer, the testing process and how a commercial laboratory runs. But after six days, in addition to learning about hereditary melanoma, variants of uncertain significance and double blinded review, I had learned even more about genetic counseling in general and the various career paths that a genetic counselor can take. I

realized that many of the skills necessary for conventional genetic counseling can be applied to careers in medical services, sales and marketing (i.e. educating, listening, asking appropriate questions) and that a genetic counselor's specialized knowledge and training can be especially valuable in such positions. Although some genetic counselors in such non-traditional roles may miss one-on-one contact with patients, many feel fulfilled in knowing that they are reaching more patients in their alternate roles. My greatest realization is perhaps that non-traditional roles for genetic counselors abound and that trainees should keep their eyes open for positions in health care or industry that best utilize their knowledge, skills and training."

Tara Sousa: "My experience at Myriad Genetic Laboratories exposed me to numerous non-traditional roles for genetic counselors. Since Myriad is one of the largest employers of genetic counselors, I was able to meet and learn about genetic counselors in research, sales, marketing and medical support, both internally and regionally. Furthermore, through my interactions with genetic counselors and other employees at Myriad, I was able to enhance my knowledge of hereditary cancer syndromes, learn more about laboratory processes and gain a better understanding about billing and reimbursement for genetic testing. I also learned about TheraGuide 5-FU™, which is a genetic test that gauges toxicity to 5-FU/capecitabine-based chemotherapeutic therapies. After seeing first hand the steps involved from receiving a sample to reporting of a result, I realized how committed Myriad is to ensuring that the patient receives the most appropriate test. I was also able to strengthen my telephone counseling skills via my interaction with patients and providers at Myriad. This internship truly provided me with a wealth of knowledge for my future as a genetic counselor and exposed me to a variety of opportunities for a genetic counselor in a diagnostic industry setting. I feel very fortunate to have been chosen as one of the students to receive a NSGC/Myriad Student Scholarship."

This joint internship between NSGC and Myriad will be offered again during the summer of 2008. Please watch the NSGC website, www.nsgc.org, and the NSGC listserv in December 2007 for additional details and an application.

Editor's Note: *For the past several years,* ***Stephanie Herbert*** *has been the Student Forum coordinator for* Perspectives. *She worked diligently, first as a student herself and then a practicing counselor, to transform this section into a recurrent and important feature. Due to a career change, Stephanie is leaving her post at* Perspectives. *Stephanie, we thank you for all of your hard work and dedication, and we will miss you! Anyone who may be interested in coordinating the Student Forum or submitting an article, please contact the* Perspectives *editor at jmandell@slc.edu.*

Genetic Counselor Publications

By Deborah McDermott, MS, CGC

Featured Paper

Clyman JC, Nazir F, Tarolli S, Black E, Lombardi RQ, Higgins JJ. The impact of a genetics education program on physicians' knowledge and genetic counseling referral patterns. *Med Teach*. 29(6):143-50. 2007.

Even before the completion of the Human Genome Project, the need for integration of genetics education into the training of new physicians and for continued education of established physicians has been an ongoing topic of discussion. Part of this discussion has focused on the importance of educating primary care providers (PCPs) who are on the front lines of patient health care. In response to surveys suggesting that patients would discuss their desire for genetic testing with their PCP first, the AMA and other organizations have developed both patient- and provider-focused campaigns stressing the importance of the family history in medical practice.

As our language becomes peppered with terms like pharmacogenomics and personalized medicine, and with the growth of an industry that is offering personalized genomic assessments, the genetics-savvy PCP seems all the more important. **Jonathan Clyman's** recent article highlights an impressive program aimed at educating PCPs in genetics, in part to provide them with the tools to recognize patients who might benefit from in-depth genetic counseling.

Until about a year ago, Jonathan was a genetic counselor in a large family practice setting in a medically underserved region (MUR) in New York State. His position was in part funded through a federal grant aimed at increasing genetic services in such MURs.

Jonathan developed and implemented a two-year genetics educational program for the general practitioners and residents in training at the practice, all of whom had little background in human genetics. While the training project was not developed as a research study, he began to notice some interesting trends while tracking data about the effectiveness of his educational modules. It was clear that the program was increasing physicians' knowledge about genetics based on pre- and post-test scores for the various modules. However, the training was not associated with an increase in the number of referrals for genetic counseling.

Jonathan noted that while the PCPs attending his presentations found the material fascinating, they were not able to translate that information to their patients. The PCPs were struck by the calculation that about 1/130 of their patients have one of about ten "common" (which he defined as < 1/10,000 incidence) genetic disorders, almost all of which can be easily unrecognized or misdiagnosed.

Jonathan speculates that a combination of factors might explain why these PCPs did not increase referrals. For example, on some days, the physicians had only seven minutes to spend with a patient, seeing up to 40 patients a day. Patient visits were often patient-driven and focused on the immediate issues, leaving little time to incorporate genetics into the discussion or to update a family history.

Jonathan found his time in the family practice setting to be an incredible learning experience, especially for gaining an understanding about how certain genetic conditions can manifest in unexpected ways. He witnessed how PCPs have a clear understanding of familial risk and that certain conditions run in families, but the integration of genetics into their day-to-day practice is just not on their radar screens yet. Residents training at this practice now are required to learn how to take a careful family history and develop a pedigree for patients.

In Jonathan's opinion, the incorporation of genetics into patient care needs a greater emphasis in medical school and in the training of PCPs in order to get physicians thinking along these lines in practice. Hopefully, genetic counselors can be an integral part of such education moving forward.

Jonathan is currently a genetic counselor and clinical cancer trial program coordinator in the Cancer Center at St. Francis Hospital in Poughkeepsie, New York.

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(names of genetic counselors appear in bold)

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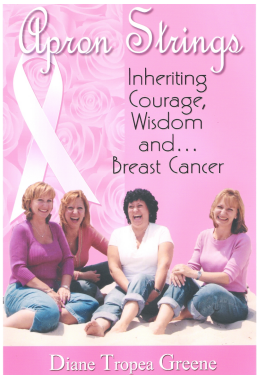
Resources

Book Review



Title: ***Apron Strings: Inheriting Courage, Wisdom and…Breast Cancer***
Author: **Diane Tropea Greene**
Publisher: Rainbow Books Inc., Highland City, Florida, 2007, pp.133

Reviewed by Shelly Cummings, MS

Apron Strings: Inheriting Courage, Wisdom and…Breast Cancer is an inspirational book for breast cancer survivors, their families and all medical professionals involved in caring, counseling and helping families cope with a diagnosis of cancer. This autobiographical book recounts **Diane Greene's** decades-long battle with cancer, initially from an outsider perspective and more movingly from a personal experience. She narrates her relatives' unsuccessful battle with various forms of cancer and her successful battle with breast cancer, the familial inheritance of cancer, her and other at-risk family members' decision-making process and the knowledge she has gained throughout these experiences.

An Emotional Legacy

The beginning of the book chronicles the diagnoses of multiple cancers in Diane's mother's generation, among them a male breast cancer in her Uncle Bob and her mother's breast and lung cancers. As Diane painfully details the diagnoses and ultimate deaths of her relatives, the reader cannot help but wonder, "What could possibly happen next?" The story then leads to Diane's generation, where the family learns the legacy of a *BRCA2* mutation.

While Diane does not share the exact mutation present in the family, she does discuss the pre- and post-test counseling process conducted by a genetic counselor. She emphasizes the "extensive counseling process" and all of the components of the session. She accurately states the estimated lifetime risks for cancer associated with having an altered gene, the population risks and the medical options following testing. One of the most impressive aspects of this book is how courageously and graphically she explains the emotional burden this disease plays on her, her husband, her two children and her three unaffected sisters.

The Courage to Fight Back

The strong bond between Diane and her husband, who was luckily away from his office in the Twin Towers during the 9/11 terrorist attack, highlights the important role that caregivers and significant others have in one's experience with cancer. Diane's personal adventure with cancer propelled her to address what was "really important in life." Her journal entries share how she sought out as much information, support and empowerment as possible during her fight with breast cancer. Diane and her family now spend their time as advocates fighting this disease.

Pass it On

This book is rich with valuable references and resources, including a listing for the National Society of Genetic Counselors website. The underlying theme is very hopeful and provides an insightful perspective for families and friends who face a diagnosis of cancer, regardless of the presence of an altered gene. The humanitarian and compassionate nature of this book, along with the knowledge that the reader gains, is relevant to anyone touched by cancer. I have never felt more strongly about recommending a resource for genetic counselors and patients as I have with this book, and I plan to mention it to every woman I counsel for cancer.

AEC Update

Save the Date for the 2008 AEC in Los Angeles, California

We are already thinking about the 27th NSGC Annual Education Conference (AEC) in Los Angeles, California. Save the dates **October 24-28, 2008.** The 2008 Short Course, "Taking Heredity to Heart: Cardiovascular Genetics, An Overview," is scheduled for **October 23-24.**

Both the AEC and Short Course will be held at the **Hyatt Regency Century Plaza**, located about 10 miles from LAX airport and across the street from the Westfield Shopping Center with over 180 stores, a movie theater, six full service sit-down restaurants and a "fine dining" food court that includes outdoor seating.

New Amenities and Events

After a successful AEC in Los Angeles in 2005, we are looking forward to heading back again in 2008. There have been many exciting changes in the hotel over the past few years. In January 2007, the Hyatt completed a $40 million renovation of the property, including all guest rooms and meeting spaces. Additionally, new amenities have been added such as an upscale Equinox Fitness Center, a full-service Starbuck's in the hotel lobby and the elite X-Bar, an indoor/outdoor lounge open daily from 4pm to 2am.

With the ease of travel in and out of Los Angeles International Airport, the array of extra-curricular activities in LA and the surrounding areas, the ever-growing Exhibitor Suite and the exciting educational program we are currently developing, we are sure your 2008 AEC experience at the Hyatt Regency Century Plaza will be unmatched. We look forward to seeing you next year in Los Angeles!

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Janice Berliner, jberliner@sbhcs.com
Stephanie Brewster, stephanie.brewster@childrens.harvard.edu

Program Co-Chairs
Lynn Holt, lynnholt@genetics.uab.edu
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Short Course Co-Chairs
Amy Sturm, amy.sturm@osumc.edu
Heather MacLeod, hmacleod22000@yahoo.com

AEC Call for Speakers – Submit Today

Are you interested in being recognized as an industry leader and expert? NSGC is actively inviting members to submit presentation submissions for **Plenary Sessions** and **Educational Breakout Sessions** for the 27th Annual Educational Conference in Los Angeles, October 23-28, 2008.

Why Should I Present at the AEC?

As a presenter, you will interact with an appreciative audience of genetic counseling professionals who will apply your insights to meet their daily challenges. Many other professionals have found this speaking experience to be gratifying and helpful for their

career development. Also, you will receive ample benefits from the educational and networking opportunities – the hallmark of the NSGC Annual Education Conference. Submit your presentation for consideration for the NSGC 27th Annual Education Conference by completing the online submission form today. Instructions on how to submit your abstract can be found by visiting the NSGC website at http://www.nsgc.org/conferences/aec.cfm.

Don't miss this exciting opportunity -- the submission deadline is January 4, 2008. All speakers will receive email notification of the status of their submissions in early February.

For More Information

Visit the NSGC website at http://www.nsgc.org/conferences/aec.cfm or contact the Plenary Co-Chairs, Lynn Holt (lholt@genetics.uab.edu) and Brooke Smith (bsmith@ggc.org) or the EBS Co-Chairs, Beth Wood (ewood@jhmi.edu) and Christine Stanislaw (cstanislaw@genetics.emory.edu).

Research Network

GenTAC Cardiovascular Health Registry

GenTAC is a national registry to promote cardiovascular health and help prevent aortic aneurysm, a disorder that weakens the main artery from the heart. GenTAC is sponsored by the National Heart, Lung and Blood Institute and the National Institute of Arthritis and Musculoskeletal and Skin Diseases. The goal of GenTAC is to establish a registry of patients with genetic conditions that may be related to thoracic aortic aneurysms and to collect medical data and biologic samples. The samples and data will be made available to qualified investigators to determine best medical practices and to advance the clinical management of genetic thoracic aortic aneurysms and other cardiovascular complications.

We are seeking people of all ages and ethnicities to join this important registry. Individuals with the following conditions are eligible:

- Marfan syndrome
- Turner syndrome
- Ehlers-Danlos syndrome
- Loeys-Dietz syndrome
- Shprintzen-Goldberg syndrome

- Biscupid Aortic Valve with personal and/or family history of aortic enlargement/dissection
- a family history of aneurysms and dissections of the aorta.

Other less common conditions may qualify; for certain genetic conditions, family history and other clinical criteria may be required.

Please contact the nearest medical center below to find out if you qualify. If you don't live near any of the clinics, we may be able to make special arrangements to enroll you. Call the Data Coordinating Center (RTI) at 1-800-334-8571 ext. 24640, or e-mail gentac-registry@rti.org. Further information also is available at the GenTAC website http://gentac.rti.org.

Johns Hopkins University: Kira Lurman, RN, 410-502-5903, kmarant1@jhmi.edu

Weill Cornell Medical College: Deborah A. McDermott, MS, CGC, 212-746-2054, dam2001@med.cornell.edu

University of Pennsylvania: Megan Morales, 215-662-4740, Megan.Morales@uphs.upenn.edu

Oregon Health & Science University: Jessica D. Kushner, MS, CGC, 503-346-0023, kushnerj@ohsu.edu

University of Texas at Houston and Baylor College of Medicine: Kristi L. Baysinger-Morin, RN, 713-500-5774, Kristi.L.Baysinger@uth.tmc.edu

Registry for Sporadic and Familial ALS

The Neuromuscular Disorders Research Laboratory at Northwestern University Feinberg School of Medicine is recruiting sporadic and familial ALS patients and their families for our neurologic diseases registry. Our research is focused on identifying genes associated with ALS, both sporadic and inherited. The study requires a blood sample from the patient, and from siblings and parents if possible. Blood tubes are provided, and shipping costs are paid.

Contact: Sandra Donkervoort, MS, 312-503-0154, s-donkervoort@northwestern.edu; Nailah Siddique, RN, MSN, 312-503-2712, nsiddique@northwestern.edu, www.neurogenetics.northwestern.edu

NEI/NIH Repository on Inherited Eye Disease

The National Ophthalmic Disease Genotyping Network, eyeGENE™, sponsored by the National Eye Institute (NEI)/National Institutes of Health (NIH), is creating a national tissue repository to further advance genetic research on inherited eye disease (ClinicalTrials.gov identifier NCT00378742; NIH study number 06-ei-0236). Patients may enroll through a certified eye or genetic care medical professional who has registered online as an eyeGENE™ provider. Patients will be asked for a blood sample and results from a standard eye examination. The blood sample and clinical information will be sent to the NEI for testing and storing in the tissue repository. Stored information will be coded by a unique identification number and will be available to researchers without patient identifiers. Patients may choose to receive results back and/or be re-contacted in the event of future clinical studies.

Contact: Ms. Ajaina Nezhuvingal, Project Coordinator, 301-435-3032, eyeGENEinfo@nei.nih.gov

Whole Genome SNP Array Analysis for the Detection of Subtle Abnormalities

The University of Chicago Genetic Services Laboratory is offering a whole genome quantitative SNP array on a research basis. The project is seeking individuals who have had chromosome analysis or are in the process of having chromosome analysis and have moderate to severe mental retardation (MR) plus one or more of the following:

- family history of MR
- major congenital anomalies
- multiple minor dysmorphic features
- pattern of minor dysmorphic features suggestive of a syndrome or growth failure.

Each patient enrolled in the study would have a whole genome SNP array analysis. There is no charge for any research testing in our lab.

Contact: Chris Tan, MS, Study Coordinator and Genetic Counselor, 773-834-9110, ctan@bsd.uchicago.edu; Dr. Stuart Schwartz, Principal Investigator, sschwart@bsd.uchicago.edu

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Public Eye

Media Watch

By Roxanne Maas, MS, CGC

August 8, 2007 – CDC's Genomics and Health Weekly Update, "The Importance of Genomics in Public Health"
A video interview featured **Kristin Oehlke** discussing appropriate integration of genomics into public health. She mentioned examples of the expansion of newborn screening programs and the development of genetic testing for cancer and other chronic diseases. She emphasized how public health can and should advocate for people with limited access to genetic testing, monitor the use of genetic testing technologies and participate in public policy decisions.

Fall, 2007 – *Women & Cancer* magazine, "Genetic Testing: What Every Woman Should Know"
Heather Shappell wrote this article about genetic testing for hereditary cancer, including the signs of hereditary cancer, the way medical management may change based on genetic test results and/or a thorough cancer risk assessment by a genetic counselor, how to find a genetic counselor (via www.nsgc.org) and why it is important to consult with a genetics expert before and after genetic testing.

September 11, 2007 – *The Wall Street Journal* online, "Ad Campaign Fuels Debate On Breast-Cancer Gene Test," and *The New York Times*, "A Genetic Test That Very Few Need, Marketed to the Masses"
These articles reviewed Myriad Genetic Laboratories' new direct-to-consumer ad campaign for breast cancer gene testing, reigniting debates about who really needs the test and whether ads will induce low-risk women to take drastic measures to prevent the disease. Some experts worried that a campaign calling attention to a rare condition could create fear and lead to unnecessary tests or a false sense of security about their results. According to the *WSJ* article, "Some genetic counselors — who prepare patients for the possible results, emotional fallout and preventive steps involved with such tests — are concerned that the Myriad ads oversimplify the benefits of gene tests." **Ellen Matloff** was interviewed for both articles and expressed her concerns about this test being marketed directly to consumers.

September 16, 2007 – Yale Cancer Center radio show, "Cancer Answers"
Ellen Matloff was featured on this radio show discussing the hallmarks of hereditary forms of cancer, the importance of knowing one's family history and how genetic counseling and testing can help individuals at risk for hereditary cancer. Ellen provided contact information for NSGC and promoted the importance of genetic counseling in the context of genetic testing.

September, 2007 – *Vanity Fair* magazine, "Arthur Miller's Missing Act"
For all the public drama of writer **Arthur Miller's** career, one character was absent: his child, **Daniel**, who has Down syndrome. Miller, referred to by some as "the moralist of the past American century," institutionalized Daniel shortly after his birth, largely refusing to see him or speak about him. However, upon Miller's death, the state of Connecticut made Miller's estate pay Daniel a full quarter of his father's assets. Per the article, "Born with an extra 21st chromosome, children with Down syndrome are often

recognized by their upward-slanted eyes and flattened facial features. They suffer from hypotonia—decreased muscle tone—and mild to moderate retardation." A disability rights advocate stated that Daniel has "made a life for himself; he is deeply valued and very, very loved. What a loss for Arthur Miller that he couldn't see how extraordinary his son is."

September 16, 2007 – *The New York Times*, "Cancer Free at 33, but Weighing a Mastectomy"
Through the story of a 33 year-old BRCA1 mutation carrier trying to decide whether to have a mastectomy, this article explored how women facing hereditary cancer can manage their family's strong and divergent opinions on cancer risk management options. Family was noted as both a source of support and stress when it comes to decisions such as elective surgery. The article mentioned that one source of support for the patient was Bright Pink, a group of young BRCA carriers that the patient was referred to by a genetic counselor. **Deborah McDermott** wrote a letter to the editor about this article, published September 23, regarding her concerns with the way patients receive genetic test results. She stated, "People undergoing genetic tests should receive qualified counseling," and "People should seek out a certified genetic counselor or geneticist, or a doctor with significant experience in ordering and interpreting these tests."

September 18, 2007 – *The New York Times*, "Where Risk and Choice and Hope Converge, A Guiding Voice"
Prenatal genetic counselor, **Daniela Iacoboni**, was featured in this article about the challenges she faces helping her under-educated patient population understand genetic risks and navigate the choices in prenatal diagnosis. In explaining and interpreting complex risk information, Daniela stated, "I try not to be patronizing but to use the simplest language I can. Numbers are very hard for most people. I try to let the patient guide me."

October, 2007 – *Nature*, "How Geneticists Can Help Reporters to Get Their Story Right"
This article aimed to help geneticists improve the coverage of genetics in the media by explaining the forces that shape science news. The article outlined specific options for reducing hype and preventing the use of genetics to reinforce discriminatory messages.

October, 2007 – PBS stations, "The Key of G"
This TV show told the story of **Gannet** (also called "G"), a charismatic 22-year-old man with severe disabilities resulting from Mowat-Wilson syndrome. The story chronicled G as he moved out of his mother's home and into an apartment with three musicians and artists as primary caregivers. Together they created a uniquely successful model of supported living and a compelling alternative to institutionalized care.

October, 2007 – *Women's Health* magazine, "Breaking the Code"
The new frontier of genetic testing for disease susceptibility was examined through the story of **Kendra**, a woman found to carry a BRCA1 mutation. **Judith Benkendorf** was quoted explaining the function of DNA in our cells. The article mentioned that **Lori**

Ballinger was the genetic counselor who "could help Kendra deal with her results and evaluate her options." Kendra recommended that people with a concern about their family history visit a genetic counselor to "help you consider your options and provide support if you're feeling overwhelmed" and "encourage you to ask yourself tough questions."

October 16, 2007 – *NPR, Talk of the Nation,* "Predicting the Likelihood of Disease"
Prompted by the recent development of a blood test that could potentially help doctors diagnose Alzheimer's disease, this story reviewed the ins and outs of predictive testing for genetic conditions. A woman with a family history of early-onset Alzheimer disease was interviewed. **Barbara Biesecker** fielded questions from callers about predictive genetic testing and prenatal testing and screening and explained how genetic counselors can be integral to patients' decision process about testing.

October 18, 2007 – *Nature*, Special Report, "DNA Masters"
The burgeoning field of genetic counseling was examined in this special article. The various roles genetic counselors can play, the training and personality characteristics necessary for the job and the different types of workplace settings where counselors are employed were all discussed. Several genetic counselors were quoted in the article.

November 5, 2007 – *NPR*, "Voices in the Family" talk show
Barbara Bernhardt was interviewed for this show about genetic predisposition testing, primarily for the breast cancer genes, and how genetic counselors can help people understand and deal with testing results. She fielded questions regarding the stigma people may feel when testing positive for a genetic mutation, genetic discrimination and the influence of non-genetic factors in mitigating risk for disease development.

November 10, 2007 – *Star Tribune*, "Five Drops of Blood: Invasion of Privacy?"
Various professionals argued whether newborn screening for genetic conditions constitutes an involuntary genetic test that should be opted-in after informed consent or an important public health mandate that should be opted-out only if a parent is strongly against it. One of Minnesota's most outspoken privacy advocates claimed that the testing has unknown future implications for employment and insurance. Doctors and public health officials countered that testing is a small intrusion that can save about 140 babies a year from death or serious disability. The article mentioned one family whose daughter was diagnosed with CF by newborn screening, noting that, "the first day, they spent eight hours meeting with doctors, genetic counselors and nurses."

November, 2007 – *Reno* magazine, "Shaking the Family Tree"
Robbin Palmer was a featured interviewee in this article about the role of the genetic counselors in investigating the hereditary nature of health issues, assessing an individual's hereditary risks and educating patients about testing and medical care management options. On a TV show called, "Join the Race," on public access station SNCAT, Robbin also participated on a panel to promote the Race for the Cure and talked about hereditary breast cancer and hereditary cancer risk assessment.

December, 2007 – *Scientific American*, "Diet Advice from DNA?"
Laura Hercher authored this article on the concerns regarding direct-to-consumer marketing of nutrigenetic tests. She commented on how the commercial applications of personalized genetic testing for optimizing diet are getting ahead of science and that giving out inaccurate or oversold information may leave people unnecessarily worried or inappropriately overconfident.

Public Relations Update: Common Breast Cancer Myths Dispelled

As part of NSGC's Breast Cancer Awareness Month efforts, our PR firm, Public Communications Inc. (PCI), devised a fact sheet to dispel common myths associated with breast and ovarian cancer and genetic testing. Cancer SIG Co-Chairs, **Nancie Petrucelli** and **Joy Larsen Haidle**, assisted PCI in developing this fact sheet. During October, the fact sheet was "pitched" to the media nation-wide. Several publications ran the news release, including *The Dallas Morning News*, *Belleville News-Democrat* and *Lansing State Journal*.

We encourage you to contact your local media about genetic counseling and testing, too. The breast and ovarian cancer myths are posted to www.nsgc.org/consumer and are available for *your* reference.

Be a PR Representative for NSGC

Mail or email the cancer myth fact sheet to the health or medical reporters at the following media outlets: hospital newsletters, local newspapers, local radio stations, local television stations, local/regional magazines and community group newsletters. Call each local media outlet to ensure that you are forwarding the information to the correct person.

Include a brief, personalized cover note with the reporter's name and title and a sentence or two explaining why you feel this information will be of interest to the reporter's viewers/readers. Follow-up with a phone call to ensure that the information has been received and to ask if the reporter has additional questions. If an article appears, contact NSGC and let us know – working together, the more we can achieve.

Bulletin Board

ABGC Certification Maintenance Fee

By Robin E. Grubs, PhD, CGC, ABGC President

As was announced at the 2006 and 2007 American Board of Genetic Counseling (ABGC) business meetings, a yearly Certification Maintenance Fee (CMF) has been instituted. The collection of an annual fee was approved when ABGC was incorporated in 1993. However, the decision was made to not collect this fee until the first recertification cycle in 2006 was complete. Most certification boards collect an annual fee to help support the work of the organization. For smaller boards like ABGC, the profit margin from administering certification exams is only three percent, and other sources of revenue, like an annual fee, are essential.

Certification Support

As the profession has grown, so have the administrative responsibilities of the ABGC. The number of ABGC diplomates and training programs has increased. Additionally, to better meet the needs of our constituents, we increased the examination cycle from three to two years and moved to a computer-based testing format. These changes are expensive but important for strengthening the certification program.

Paying the annual CMF will allow certified genetic counselors to recertify with no additional fees and receive benefits such as participation in the PAC (professional activity credit) program. Promoting the CGC® is critical so that your credential continues to be recognized by the health care industry as the gold standard for genetic counseling services – particularly in areas such as billing, licensure and reimbursement.

Check Your Mailbox

For additional information please see the ABGC website at www.abgc.net. The fee in 2007 is $75. An invoice was sent at the end of October to all certified genetic counselors. If you did not receive an invoice, please contact the ABGC office at 913-895-4617 or info@abgc.net. Genetic counselors certified in 2007 will receive their invoices after the first of the year.