President's Beat

To Infinity and Beyond

As I sat down to write my last *President's Beat*, I felt an unusual mix of emotion. I smiled as I thought about the many accomplishments from the year and the great strides forward we took as an association and as a profession: another state with licensure, a new Vision Statement, a new logo and website, an outstanding Annual Educational Conference, and more collaboration with other stakeholders in genetics. This list is incomplete, as there were too many advances to list everything.

Of course, there are things I wish we had been able to accomplish. We were unable to introduce our federal bill, which aims to amend the Medicare regulations to recognize genetic counselors as providers. Few legislators wanted to touch a healthcare topic after the rollercoaster ride of healthcare reform. It's disappointing, yet under the leadership of our Policy and Government Relations team, we will continue forward. Our Special Interest Groups have reached out to numerous genetic disease support groups and a few organizations have already written or offered letters of support.

I am truly excited for 2011. The momentum is growing. I attended several of our Committee meetings in Dallas and witnessed their enthusiasm for next year's activities. A few highlights include expanding the content for physicians on the NSGC website, designing local marketing kits for our members to use, developing new Position Statements and Practice Guidelines, researching different ways to deliver genetic counseling services, and increasing efforts behind the federal bill and state licensure. (Eight states currently have bills introduced, and nine more are organizing!)

Since our meeting in Dallas, we haven't slowed our efforts. The Policy and Government Relations Team and I met with the Federal Drug Administration (FDA) on November 9 to provide detailed input into the FDA's oversight of genetic testing. We met with about ten representatives from the FDA including Dr. Jeff Shuren, Director of the Center for Devices and Radiologic Health, Dr. Elizabeth Mansfield, Director of Personalized Medicine, and Dr. Alberto Gutierrez, Director of the Office of In Vitro Devices. The FDA indicated they are very aware of the importance of access to genetic testing for rare diseases due to the benefits that families have experienced because of the advances in testing. We informed them of the NSGC's support for overall increased oversight of genetic testing using a risk-based approach that the FDA uses with medical devices in general. (Medical devices are classified at three different levels of risk based on the potential for harm to a patient. For example, a tongue depressor would not be in the same risk category as an artificial knee.)

Some of the suggestions for risk classification we made based on the NSGC's Public Policy Committee's discussions include: 1) Considering the likelihood that a Board-certified genetics professional will be involved in applying or interpreting genetic testing and 2) Evaluating how a test will be applied instead of only the disease being tested. The reasoning behind these recommendations is that Board-certified genetics professionals serve as effective gatekeepers for genetic testing, so the risk to the public is greatly reduced when they are involved. The population being tested is important because, for example, a carrier test for a very rare disease carries different risks when applied to a family with a known affected individual as compared to the general population. We also stressed the importance of inter-agency collaboration, as many healthcare professionals don't feel comfortable with genetic testing and FDA oversight alone can't address all aspects of delivery of genetic information to patients. The FDA representatives expressed interest in

this input and also asked questions about other areas, such as whole genome sequencing and pharmacogenetics. The NSGC will continue to communicate with the FDA, and many of you attended a webinar given by Dr. Elizabeth Mansfield on November 10 on this same topic.

Also on November 9, we met with officials from the Center for Medicare and Medicaid Services (CMS) to inform them of our intent to introduce legislation to amend the CMS regulations to recognize genetic counselors as providers. We provided an overview of the value genetic counselors bring to patients and other healthcare professionals, as well as the current billing methods genetic counselors use for Medicare patients. The overall message was that patients need to have access to the most qualified providers in a specialty, and genetic counselors are clearly experts in translating genetic information.

All of these efforts will continue under the leadership of 2011 President **Karin Dent**, the 2011 Board of Directors, and of course our dedicated Executive Office Team. I admit I feel a lump in my throat as I realize I'm almost done with my year as NSGC President. I've enjoyed every minute, even those that stretched my skills or challenged my beliefs as a genetic counselor. There is little else for which I have felt such passion, and I've had the honor of serving for a cause that will broadly impact people's health. I will never forget 2010 and the gifts I received from the NSGC.

Having watched a few children's movies recently with my young nephews, I'm reminded of Buzz Lightyear's famous phrase "To Infinity and Beyond!" as I look forward for the NSGC. We don't know exactly what the future holds for the genetic counseling profession, yet we leap forward with the confidence that, wherever we land, we will find our role as stewards of genomic medicine.

Elizabeth Kearney

Elizabeth Kearney, MS, CGC, MBA
2010 NSGC President

Advocates Partnership Program at the 2010 AEC

By Andria Cornell, Advocacy and Health Policy Manager, Genetic Alliance

For the fifth consecutive year, Genetic Alliance partnered with the National Society of Genetic Counselors (NSGC) to sponsor the Advocates Partnership Program at the 2010 Annual Education Conference (AEC) in Dallas, Texas in October. The Advocates Partnership Program is specifically designed to connect family advocates and students with genetics professionals and the NSGC leadership. It offers an unparalleled opportunity for participants to gain unique insights into the genetic counseling profession, while expanding their understanding of cutting edge issues in genetics and technology. Funding for the program is provided by Genetic Alliance, which coordinates the logistics of the program and offers a stipend for participants, and the NSGC, which provides support for registration and travel.

This year, seven individuals participated in the Advocates Partnership Program at the NSGC AEC. Our most diverse group yet, the participants included parents of children affected by genetic conditions, staff members from disease-specific advocacy organizations, leaders of community-based organizations, a physician assistant, and two young women who recently finished their undergraduate education and wish to pursue careers in genetic counseling. The structure of the Advocates Partnership Program complements the annual conference agenda; each day of the conference includes an exclusive daily briefing for the advocates with the NSGC leadership and a wide range of professionals in clinical genetics. Advocates' interests guide discussions in these one-hour meetings. Guest speakers answered questions and provided insights from their personal experiences.

This year's Advocates Partnership Program launched with the advocates sharing personal stories of what brought them to the program and what they hoped to come away with after the four-day conference. The first scheduled briefing engaged the guest speakers in explorations of key tensions families experience in the care of their children. Advocates spoke directly to the value of the communication partnership that occurs in a genetic counseling session, including the encouragement to ask questions. In the medical setting, when patients or parents ask "Why?" of a physician, it can be interpreted as acting suspicious or pushy. Advocates also acknowledged the sheer volume of conditions about which a health professional must be knowledgeable, and how making an early diagnosis, though desired, is understandably challenging. The topic of care coordination inspired a lively discussion from the group, and in response to a question around reasonable expectations for the medical home, an advocate said, *"If you ask me, 'Who is your primary care provider?' I say ME."*

These conversations continued in the second scheduled briefing, where members of disease-specific advocacy organizations shared the importance of managing expectations in a family's experience with disease. Some parents can be given false hope, both in terms of outcomes for their child and in the possibility of finding a cure, while other

parents are told what could be perceived as catastrophic news that is later proven to be false. When it comes to communicating the severity of a condition and treatment options, parents must be engaged to develop strategies that respect the family's values and how they choose to cope with the diagnosis.

By far the most energetic conversation of the Advocates Partnership Program took place on the afternoon of Saturday, October 16 during the third daily briefing that followed the Dr. Beverly Rollnick Memorial Lecture, featuring author and broadcaster Ian Brown. The impact of Mr. Brown's presentation can be summarized by one advocate's impressions that she shared in her post-program evaluation:

Ian's lecture reminded me that grief is a process and it changes, but never completely resolves for families. I have always speculated about the true feelings of my clients, but have not always heard them out loud. Ian's willingness to say the ugly and uncomfortable things was shocking, at times, but definitely refreshing. Because genetic counselors have a limited amount of time with families, they may never hear these perspectives. I would encourage NSGC to have more of these lectures and/or panels of parents at the AEC in the future. Life experiences are not something that can be taught in a textbook.

This particular advocate serves in a community-based family support network. In fact, each post-program evaluation submitted by the advocates specifically mentioned Ian Brown's presentation as a highlight of the conference. Discussion in the third briefing centered on the topic of "best outcomes." Participants agreed that the best outcome for a child, from the provider perspective, might be vastly different from the best outcome as defined from the family perspective. How can best outcomes from the family perspective be measured and promoted? One advocate shared that Ian Brown's presentation put health in perspective, saying, *"First and foremost, you want your kid to be safe and happy."* The group discussed quality of life, finding the hope, and finding the joy in life. Together we asked, how could we use stories like these to share the value of taking the time to listen to families?

The final briefing focused on actionable steps going forward. Ideas included genetic counselors leading trainings for medical school students on the importance of partnering with the family in care. Another was for more genetic counselors to become involved in the Health Resources and Services Administration genetics collaboratives, as they are forums for idea exchange on a broader level. In reference to the disease-specific lectures, which many of the advocates found valuable and relevant to the communities they serve, an advocate shared that more parents could be included in panels to share their perspectives of the diagnostic process and provide first-hand commentary of their experiences, in order to better capitalize on the value of parents being involved in the forum.

The advocates enjoyed working with the speakers throughout the program, but were particularly appreciative of being joined by 2010 NSGC President **Elizabeth Kearney** and NSGC Executive Director **Meghan Carey** at the final briefing. One of the advocates who recently obtained her undergraduate degree, shared, *"I feel that they really*

appreciated our comments about the conference, how they can reach more people, and some suggestions for improvements for next year. I loved telling them about how this experience has affected me as a recent graduate looking to gain an M.S. in genetic counseling in the future." The other recent graduate said, *"These meetings gave me the opportunity to see how parents and advocates view the profession I am trying to join. This was an invaluable experience."*

Reflecting on her participation in the program, one advocate offered in her evaluation, *"Before the conference, I did not have a clear understanding of the role that genetic counselors play in the life of their patients. I always assumed that they were there to merely guide families through the difficult time of diagnosis. Now I see that the job is so much more."* Another advocate went on to say of the Advocates Partnership Program, *"I consider the Advocates Partnership Program the best part of the conference, because it gave me the vocabulary to discuss complex topics with individuals from every facet of the genetics field."*

The value of our discussions was further articulated in the following excerpt from a participant's evaluation; this is from a physician assistant in a family practice setting:

"I was reminded that though most in the medical field are trained in a constructed paradigm of disease and prevention, there are so many other factors that contribute to wellness. Good communication between patients, families and providers is possibly the best trajectory for wellness. I return to my practice in general medicine prioritizing patient-centered questions and certainly even more of an effort to participate in active listening."

The Advocates Partnership Program has far-reaching effects outside the AEC. The experience shapes advocate understanding of the genetic counseling profession, and advocates bring this new knowledge back to their communities to inform future partnerships. The speakers that participated in the daily briefings expressed feelings of being recharged after brainstorming with the advocates, having committed to perpetuate the shared learning that occurs in the forum.

Genetic Alliance and the NSGC will work together in the months following the program to implement many of the ideas for collaborations generated by the program and to continue to expand the accessibility and diversity of the program. We want to hear your ideas for doing so! Please contact Andria Cornell, Advocacy and Health Policy Manager at Genetic Alliance (acornell@geneticalliance.org) or Meghan Carey, NSGC Executive Director (mcarey@nsgc.org) with questions or ideas. We look forward to hosting another dynamic forum next year in San Diego, California!

2010 NSGC Advocates Partnership Program Participants

Hilary Andreff
B.A., 2008

Mount Holyoke College
Englewood, Colorado

Rebecca Bassett
Physician Assistant
Family Care Medical Center
Goshen, Massachusetts

Brian Denger
Collaborative Programs Administrator
Parent Project Muscular Dystrophy
Biddeford, Maine

Brittany Jenkins
B.S., 2010
Clemson University
Summerville, South Carolina

Sheilah Michaels
Advocate
The Arc of Aurora
Aurora, Colorado

Yolanda Sandoval-Nez
Parent and Senior Advocate
Native American Disability Law Center
Brimwall, New Mexico

Tina Sasser
Public Outreach Specialist
LaunchAbility
Dallas, Texas

2010 NSGC Advocates Partnership Program Speakers

Mary Freivogel, 2011 NSGC Board Member
Katie Dunn, Student/New Member SIG Chair
Patricia Devers, Prenatal SIG Chair
Pamela Callum, ART/Infertility SIG Chair
Christine Spaeth, Fetal Intervention and Therapy SIG Chair
Angela Trepanier, NSGC Past President
Elizabeth Kearney, 2010 NSGC President
Linda Robinson, 2010 NSGC Board Member
Meghan Carey, NSGC Executive Director

Do One Thing a Day That Scares You

By Deepti Babu, MS, CGC

I recently did something I've rarely done before in my career as a genetic counselor. I went to a conference put on by a support group, which are typically geared for families and those who care for them in the healthcare community. It's not that I've intentionally avoided these conferences, but I think my focus has been on attending those that gain me the most Continuing Education Units (CEUs), in order to avoid taking another certification exam. But when I learned that the Huntington Society of Canada (HSC) was having their annual national conference in Edmonton, where I live, I saw a unique opportunity. Although I'm a generalist, I do see quite a few families for Huntington disease (HD) counseling. The HSC has a Chapter in Edmonton that I always mention to families, but I've never attended a support group meeting to check them out for myself.

I will admit, I wasn't sure what to expect. Would the meeting be scientific enough? Would I learn something? Would it be hard to see a bunch of people clearly affected by HD and their families all in one room? And would it be worth attending without getting CEUs?

Going into the conference, I now suspect I was feeling early signs of compassion fatigue… or, at the very least, *fatigue*. I'd had a few trying sessions, after which I would come home to my young family with their own needs. I felt torn, like I wasn't able to offer my usual amount of understanding and patience to either group. As the conference approached, I wondered whether I'd have the energy or attention span to get through 2½ days of topics all about HD.

This year's HSC conference theme was "The Early Years," so many talks pointed the lens at a group often overlooked in the HD community: young people. Younger members of the group, some at risk to develop HD, spoke eloquently about the experiences of witnessing their loved ones "wear the mask of HD" as their symptoms progressed over time. Underneath it all, their parent with HD was still their parent. They talked about caring for their family members and growing up too quickly because of their circumstances. There were few dry eyes after listening to these brave young people openly talk about their lives. But what surprised me was that some of their relatives with HD were actually in the audience, and got up to hug them after they were done speaking. I wasn't expecting to be a witness to that kind of openness, tenderness, and love.

Many talks were scientific, from a workshop on clinical trials and studies, to one mapping out the intricate start-to-finish process of pre-implantation genetic diagnosis, to another on the clinical implications of CAG repeat sizes in the intermediate range. The topics didn't surprise me, but the questions did. This audience knew their stuff. This information *mattered* to them. Their questions were complex, and they weren't afraid to ask them. I had underestimated their knowledge base, and came away with several

scientific learning points of my own. In fact, I may have learned more than at some meetings held by scientific groups.

My favorite part of the conference was the humor and fun that were present throughout. Let's face it, I was prepared to be brought down. But quite the opposite happened, which was a wonderful surprise. I learned that many families with HD cultivate a wicked ability to laugh, perhaps in part to cope with the genetics they are handed. And with this, judgment falls away. The conversations I had with many families and the laughter I heard throughout the conference illustrated this vividly for me.

I think I saw this statement on a lululemon athletica™ shopping bag the other day, and I now know that it's more than a marketing ploy: "Do one thing a day that scares you." When I registered for the HSC conference, a part of me was trepidatious. I've been practicing genetic counseling for awhile, so this was embarrassing to admit – even to myself. But attending kept me engaged in life. And what I gained from the experience is far more than can be encapsulated in a CEU. I now feel mentally refreshed, and I return to my "normal life" with a new perspective that I hope to cultivate and maintain.

For any of you in the scientific community who have gone to these types of meetings before, I applaud you. For those who have not, I urge you to consider it. What you learn may surprise you.

Licensure / Billing & Reimbursement

Coding Corner

Grassroots advocacy for yourself and your profession: A call to arms to the NSGC membership!

By Shanna Gustafson, MS, MPH, CGC and
John Richardson, NSGC Government Relations Director

The Coding Corner is supported by the Coding Subcommittee of the NSGC and aims to assist NSGC members with the application and understanding of governmental regulations and guidelines regarding terminology and CPT/ICD coding in genetic services.

For those of you who were in Dallas this year, it was an eventful and exciting time. One of the most interesting and critical discussions occurred in the late-breaking professional issues panel, consisting of: **John Richardson**, NSGC Policy and Government Relations Director, **Amber Trivedi**, MS, CGC**,** Senior Vice President of Provider and Client Services with Informed Medical Decisions, and **Karen Lewis**, MS, MM, CGC, Medical Policy and Technology Administrator for Priority Health. This panel provided a call to arms for our members to work together and take charge of educating our colleagues, providers, institutions, and third party payers about our availability and the benefits gained by including genetic counselors as unique members of their health care teams.

One of the most important initial steps in moving forward with this grassroots effort is for counselors to have a solid understanding of how their institution is currently delivering, billing for, and being reimbursed for genetic counseling services. Although the Service Delivery Task Force is well on the way to defining some of the current models of service delivery, a disturbing statistic from the recent Service Delivery survey showed that 50% of responding genetic counselors were either not billing for their services or did not know if or how their institution was billing for genetic counseling services.

This statistic is astonishing! Genetic counselors are known for being detail-oriented and skilled at investigating the tiniest details relevant to patients and their diagnoses. Yet when it comes to the details of how we practice and how we are valued, it seems to have become less of a priority. It is time to start asking questions and understanding your services from first referral to final bill collection! Is your institution credentialing non-physician providers? Can genetic counselors be credentialed? Is your time being billed for, and with what model? From where does the financial support for your services come? How was the value of your services determined? No question is a bad question if it helps you to understand where you fit into your health care system.

Feeling like you don't know where to start? You can begin by educating yourself about billing issues now by taking the NSGC's online coding course, entitled "*Learn the 3 C's*

to Maximize your Service Delivery Model: Coding, Credentialing and Compliance." Also, look forward to the upcoming NSGC online course "*Take Control of the Revenue You Generate: How to Become a Credentialed Provider"* that will take you through a step-by-step guide to credentialing and contracting. Start contacting your medical staff offices and billing departments; perhaps they don't really understand what a genetic counselor does, or the benefits of these services to patients and the institution. It may seem small, but this is an amazing opportunity to start being a local leader and to begin the education and advocacy that the NSGC is calling for.

"The Coding Corner" is your resource for questions about coding. If you have questions you wish to be considered for this section, please send them to ***Shanna Gustafson*** *at shannagustafson@gmail.com or* ***John Richardson*** *at jrichardson@nsgc.org.*

NSGC News

NSGC Leadership Awards – As Announced at the 2010 AEC

By the NSGC Award Committee Members: Adam Buchanan, MPH, MS, CGC; Samantha Baxter, MS, CGC; Jennifer Eichmeyer, MS, CGC; Caroline Lieber, MS, CGC; Sheetal Parmar, MS, CGC; Julie Rousseau, MS, CGC; Jennifer Sullivan, MS, CGC

Natalie Weissberger Paul National Achievement Award

Bonnie Jeanne Baty, MS, CGC

During the awards selection process, a Committee member observed that selecting the recipient of the Natalie Weissberger Paul Award is like choosing between diamonds. Each one is special, a rare and valuable individual in the field of genetic counseling, making the selection process a challenging one. The 2010 Natalie Weissberger Paul recipient, **Bonnie Jeanne Baty**, is an especially bright diamond, whose lifetime of accomplishments and contributions to the field of genetic counseling and to the NSGC clearly stand out among the rest.

Bonnie is a pioneer in the field of genetic counseling. She received her genetic counseling degree in 1973 from Rutgers University and shortly thereafter began an almost 37 year-long career at the University of Utah, first as a research associate and genetic counselor, and then as the Program Director for the University of Utah Graduate Program in Genetic Counseling. She is a Professor of Pediatrics and an Adjunct Professor in the College of Nursing.

Bonnie's professional achievements and contributions are vast in all arenas including clinical, academics, and research. She established genetic counseling services at the University of Utah for perinatal, clinical, and metabolic genetics; for families with hemophilia, retinoblastoma and Huntington disease; and for cancer genetic counseling in

a research setting. She has authored 28 peer-reviewed journal articles, written seven book chapters (two published this year alone!), and edited two seminar editions of the American Journal of Medical Genetics. Her areas of research include genetic counseling practice, the natural history of genetic conditions, and cancer genetics research.

Of course, for someone as passionate about genetic counseling as Bonnie, being a counselor, professor, program director, and author is certainly not enough. In addition to these responsibilities, she has been an active leader within the NSGC. She has been a member of the NSGC since it was formed in 1979, and since then has served as Chair of the Logistics Committee, a member of the Nominating Committee, a Region V representative, on the Board of Directors for the Jane Engelberg Memorial Foundation, a member of the Scope of Practice Task Force, and a Region V Education Conference co-organizer. She has presented at both regional and national meetings. Her contributions were recognized in 1996 when she received the NSGC Region V Leadership Award.

Bonnie believes in leading by example, and her leadership within the NSGC speaks for itself. She also believes in the mission and vision of the NSGC, and is an advocate for membership. Within the first few days of coursework, her counseling students are introduced to the NSGC as their professional organization. They are taught the history and application of the Code of Ethics and the importance of professional growth and development as a genetic counselor within the NSGC.

In addition to work with the NSGC, Bonnie is also a member of the American Society of Human Genetics, Transnational Alliance for Genetic Counseling and the Association of Genetic Counseling Program Directors, for which she is the current Recruitment Workgroup Chair. Her passion for education of genetic counselors, other health professionals and the community at large, is evident from her CV (which reads like a university course manual!). As a professor and program director, she has developed and directed nine different clinical genetics and genetic counseling courses. She has served on many professional and scientific committees and given lectures at local schools, and for parent and community groups. She is a Charter Member of the American Board of Genetic Counseling and has served as an ABGC Accreditation Chair.

An individual who nominated Bonnie summarized it best: “Bonnie is a true advocate for our organization. She upholds the mission and visions of the society in all of her professional interactions and volunteer work, as well as in her teaching of future genetic counselors… I believe that recognizing lifetime contributions to the society and profession are due.”

Congratulations to **Bonnie Baty**, winner of the NSGC’s 2010 Natalie Weissberger Paul National Achievement Award.

Strategic Leader Award

Angela Trepanier, MS, CGC

Angela Trepanier wears many hats.

She is the director of the Wayne State University Genetic Counseling Graduate Program.

She is a board member for the National Coalition for Health Professional Education in Genetics and a founding board member of the Genetic Counseling Foundation.

She is currently the NSGC appointed liaison to the National Human Genome Research Institute's "G2C2" project.

She has been a liaison to the Coalition for accessible family history tools, and a liaison for the task force on the evolving role of the medical geneticist.

She has served on many NSGC Committees, including Communications and AEC planning.

She helped found the Michigan Association of Genetic Counselors, took part in a workgroup on newborn screening, and helped plan a conference for Genetics in Primary Care.

She has a longstanding record of doing community outreach, speaking at career fairs, in schools and at national conferences.

She has given many interviews on the role of genetics in health to a wide variety of media sources including *Self*, Parents.com, *The Advocate*, *The Los Angeles Times*, and the *Pharmacogenomics Reporter*.

She probably holds the record for longest serving on the NSGC's Board of Directors – seven years!

The ways in which Angie has contributed to the promotion of the NSGC are almost too numerous to count. The foundation of her work has been her tireless service on the NSGC's Board of Directors, especially during the transition to the new leadership model. She has been a clear, articulate voice for genetics and genetic counseling, both within the medical community (her work with the "G2C2" project), policy makers (all of her work as a past NSGC President), and the lay public (as a former presidential face of the NSGC). Without her vision and energy, the NSGC's growth in the late 2000's would not have been the same.

It goes without saying that Angie has contributed a tremendous amount to NSGC's strategic planning for more than a decade. Her contributions in virtually all areas of the NSGC's strategic plan have enabled the organization to move forward in becoming recognized as a thought leader in genetics/genomics. She is very deserving of this award and recognition. Congratulations to **Angela Trepanier**, winner of the NSGC's 2010 Strategic Leader Award.

International Leader Award

Rawan I. Awwad, MS, CGC

Rawan Awwad was born and raised in the cities of Jerusalem and Ramallah. After finishing high school, she moved to the United States for higher education. She graduated in 1998 from Earlham College in Indiana with a Bachelor's degree in Biology and Minors in Psychology and Chemistry. In 2001, she received a Master's degree in Genome Science and Technology from The University of Tennessee. She then went on to complete her Master's degree in Genetic Counseling at the University of Minnesota in 2006, followed by a publication in the *Journal of Genetic Counseling* entitled "Culture and Acculturation Influences on Palestinian Perceptions of Prenatal Genetic Counseling" relating to her M.S. thesis work.

After completing her education, Rawan moved to San Francisco to provide prenatal genetic counseling services, and after two years she returned to the Palestinian Territories and Jerusalem. She has been a member of the NSGC since 2005 and recently started working on the Communications Committee. She has served as a reviewer for the *Journal of Genetic Counseling* since 2008.

What is most impressive about Rawan is what she has accomplished in a region with very difficult political and cultural challenges in such a short time. Upon returning to Jerusalem in 2008, Rawan volunteered at a main clinical genetics center in the city, which serves both Palestinian Arabs and Israeli Jews. She was the first Arabic-speaking genetic counselor, and managed several genetic counseling cases with geneticists who previously resorted to translators to communicate with patients. After achieving licensure by the Israeli Board of Clinical Genetics, she was granted a position.

Since early 2009, Rawan has expanded genetic counseling services to include Palestinians living in the West Bank and Gaza who, due to the current political situation, cannot enter Jerusalem without special permits that are often hard to obtain. Rawan made genetic counseling services available to them remotely and/or in person by traveling to their cities of residence. She also coordinated their genetic testing process by transporting their blood samples back to the hospital.

At the end of 2009, Rawan volunteered with a newly formed Palestinian genetics center, which aims to provide genetic and metabolic testing services to Palestinians in the West Bank. She created an educational brochure called “Genetic counseling in prenatal care” that was distributed widely among obstetricians and gynecologists to raise awareness and increase referrals. She also prepared lectures and training material on genetic counseling that aimed to raise awareness among nurses, physicians, and social workers. In June of 2010, she participated as a lecturer in the first workshop on genetic counseling at Al-Quds University, Abu Dies, Palestine.

During Rawan’s work in the past two years, she identified several ethical and social issues involving Arab patients that were shared with other professionals through conferences and meetings at various cities such as Haifa, Ramallah, and Geneva. In May 2010, she was invited to share her experience with counseling consanguineous Arab families at the International Workshop on Consanguinity, Geneva. The workshop concluded with several working papers that aim to address issues of genetic counseling regarding consanguinity.

Outside of work, Rawan enjoys running, swimming, and traveling to different countries to learn about other cultures and cuisines. She has been active in dialogue workshops between Israelis and Palestinians, which aim to increase awareness on the thoughts, struggles, and lives of individuals from “the other side.” She believes that the two populations have completely different narratives on the history and reality of the current political situation. Although these workshops will most likely not bring a solution to the complex and painful struggle between Palestinians and Israelis, Rawan believes that

knowledge and understanding of the other side is a power that will eventually bring change.

It goes without saying that Rawan has been an incredible advocate for her patients and continues to strive to bring genetic services to populations otherwise without access. She demonstrates the very best of genetic counseling with her dedication and compassion for the people, particularly women, of Palestine. She continues to be an active member of the genetic counseling community. Congratulations to **Rawan Awwad**, winner of the NSGC's 2010 International Leader Award.

International Leader Award

Janice G. Edwards MS, CGC

The International Leader Award is bestowed upon an individual whose contributions to the profession have reached an international scope. This includes such activities as expanding the reach of genetic counseling services outside the United States, significant contribution to educating international lay and healthcare communities on genetic counseling, and significant contribution to research involving genetic counseling in an international venue.

Our next recipient of this award has performed all of these activities many times over. In fact, as the Awards Committee was reviewing nominations, a Committee member said, "Seriously, she hasn't received this award? With everything she's done, I just figured she had." Please join us in recognizing **Janice Edwards** for her international leadership and congratulating her for this well-deserved award.

Janice's record of accomplishments within the global genetic counseling community is distinguished and influential. Many of you are familiar with the Transnational Alliance for Genetic Counseling (TAGC), which fosters communication and collaboration among genetic counselor educators throughout the world. As Founding President of the TAGC, Janice incorporated the alliance and chaired TAGC meetings in Manchester, Barcelona, and more recently, Gothenburg, Sweden. This is no small feat, considering the inaugural meeting gathered genetic counseling educators from eighteen countries and included the fifteen professional organizations in the world that represent genetic counselors. Janice continues to lead the Board of Directors and manage the TAGC website, which links

international genetic counseling programs and professional societies; this website (http://tagc.med.sc.edu) is a great resource for our now global profession.

Janice's work in advancing international collaboration includes other service as well, including being a former Chair of the American Board of Genetic Counseling's Ad Hoc Committee on International Genetic Counseling Education and as a member of the NSGC's International Service Providers Committee. She has given professional talks in China and Australia. And, as the Director of the Genetic Counseling Program at the University of South Carolina, Janice has educated international students and coordinated clinical rotations abroad for several graduate students – her students, other North American students, and students from programs on other continents as they seek international experiences.

It is clear that the potential in transnational collaboration is enormous. Congratulations to **Janice Edwards**, winner of the NSGC's 2010 International Leader Award.

New Leader Award

Bronson Riley, MS, CGC

A few years after graduating from the University of Nebraska with a dual major in English and Education, Bronson began his work as a registry coordinator and genetic research associate for Henry Lynch, M.D. at the Hereditary Cancer Institute at Creighton University. It was through his work at the institute that Bronson became interested in the field of genetic counseling. He received his Master of Science in Genetic Counseling from Case Western Reserve University in 2006. After graduating, he started the genetic counseling program at the Southeast Nebraska Cancer Center in Lincoln, Nebraska. He currently sees patients for hereditary risk assessment, genetic counseling, and genetic testing. He is also a Research Assistant in the Center's Department of Clinical Research.

Bronson's contributions to the NSGC in the last five years are countless. Currently, Bronson serves as the as the Practice Issues Subcommittee Co-Chair for the Familial Cancer Risk Counseling Special Interest Group, as well as being an active member of the Communications Committee, Membership Committee and Genetic Counseling Access and Service Delivery Committee. He has previously participated in the NSGC Core Skills Task Force. If his contributions to the NSGC weren't enough, Bronson also serves as the chair of the Genetic Counseling State Licensure Committee for Nebraska and was just elected as the upcoming Co-Chair of the American Board of Genetic Counseling's Genetic Counseling Certification Exam Committee.

One would assume that with this number of responsibilities, Bronson would be limited in the amount of time and energy he could afford to give to each project, but that is not the case. Instead, Bronson takes on more and more tasks and brings endless amounts of energy and enthusiasm that inspire others around him to do more. In Bronson's own words, "NSGC has given so much to me I feel a need to give back." His tireless service and eagerness to volunteer is why he is so deserving of the New Leader award. Congratulations to **Bronson Riley**, winner of the NSGC's 2010 New Leader Award.

Outstanding Volunteer

Susan Hahn, MS, CGC

When writing about **Susan Hahn**, one of her nominators stated a common theme for those who have had the pleasure to work with her, " I was amazed by her energy, enthusiasm, and dedication to the NSGC and its projects. "

To say that Susan is a tireless worker on behalf of the NSGC is an understatement. Just in 2010, she has been the Vice-Chair of the Public Policy Committee, Co-Chair of the Public Health Special Interest Group, and a member of the Practice Guidelines Committee of the American College of Medical Genetics.

And her work on behalf of the NSGC in 2010 is by no means a deviation from the norm for Susan. In the past, she has also chaired the Practice Guideline Subcommittee and the

Research Sub-Committee. She has contributed to a variety of continuing education programs for the NSGC including co-chairing the NSGC Annual Education Conference (AEC) in 2005 and the Research Short Course in 2004. Related to this, she also either chaired or co-chaired the NSGC AEC Communications Committee (2002), Educational Breakout Session (EBS) Committee (2004), and Abstract Committee (2001), and participated in an EBS on the Genetics of Cardiovascular Disease (2001).

Over the years, Susan has served in many professional roles during her "spare time." After completing her graduate degree at Brandeis University in 1996, Susan began her genetic counseling career in a private clinical genetics laboratory with both clinical and administrative responsibilities. In 1998, she became a Study Coordinator at the Center for Human Genetics at Duke University for GENECARD, an international study aimed at identifying the genetic factors that contribute to early-onset coronary artery disease.

After almost ten years at Duke University, she was recruited to a Faculty position at the John P. Hussman Institute for Human Genomics at the University of Miami Miller School of Medicine in 2007, where she is currently the Assistant Director of Communications, Compliance and Ethics.

Needless to say, as an employee and a volunteer Susan seems to always be on the cutting edge of the genetic counseling profession. Susan has a gift of getting to the heart of an issue, identifying what needs to happen to move it along... and most of all getting it done (and motivating others to do the same). She always has ideas to do things better and has incredible insight to offer to many difficult, complex issues that come before the various committees and SIGs she is involved with.

It would be hard to say enough about Susan's generous spirit to friends, colleagues, the profession of genetic counseling and the NSGC. Congratulations to **Susan Hahn**, winner of the NSGC's 2010 Outstanding Volunteer Award.

Outstanding Volunteer

Shannan DeLany Dixon, MS, CGC

Shannan DeLany Dixon has a long volunteer history with the NSGC. She has been involved in all facets of the Annual Education Conference, including being a member of the Program Committee in 2000-2001, a member of the Mini-Course Committee in 2001-2002, Co-Chair of the Abstract Committee in 2001-2003, Co-Chair of the Educational Breakout Session Committee in 2002-2004, Vice-Chair for the 2009 meeting, and Chair for the 2010 AEC. Shannan was the NSGC state representative from Georgia for a number of years and currently is a CEU evaluator. She has also been a member of a number of advisory boards and committees outside of the NSGC, including her current membership on the New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services (NYMAC) Committee for Genetic Education of Primary Care Physicians.

Shannan has been Director of the Genetic Counseling Training Program at the University of Maryland School of Medicine since 2004. In addition to being an instructor for many courses, she has mentored students as a clinical rotation supervisor and has been a member of a number of students' thesis committees. She also instructs medical, nursing, physician assistant, physical therapy, and occupational therapy students.

Shannan has extensive clinical experience in many areas of genetic counseling and medicine, which has translated into numerous poster presentations at society meetings and platform presentations at the American College of Medical Genetics and American Society of Human Genetics meetings. She has been an invited speaker for many organizations' meetings, including the Susan G. Komen Maryland Annual Symposium.

Shannan has continuously dedicated a significant amount of time and energy to volunteer work for the NSGC. When she was informed that she received this award, her first question was, "Is there anything I can do to help?" As one of her nominators stated, "Shannan DeLany Dixon exemplifies an outstanding leader of the National Society of Genetic Counselors and is deserving of this recognition" as Outstanding Volunteer.

Congratulations to **Shannan DeLany Dixon**, winner of the NSGC's 2010 Outstanding Volunteer Award.

ABGC Update

2010 ABGC Certification Examination Update

By the ABGC Board of Directors

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The 2010 American Board of Genetic Counseling (ABGC) Certification Examination was the second exam administered using the single exam format introduced in 2009. The scoring method was previously reviewed in detail (see http://www.abgc.net/CMFiles/FINAL_Scoring_methodology_for_NOV09_Perspectives_11090951NSD-1192009-7883.pdf). Therefore, rather than repeating that information, this update will summarize the examination development process and present data on the outcome of the 2010 certification exam.

The ABGC certification exam used exam items written by more than forty contributors, including genetic counselors, MD geneticists, and PhD geneticists. Item writers contributed items in their respective areas of expertise that fit the ABGC detailed content outline constructed in 2008 based on the Practice Analysis (Hampel *et al.*, 2009). The Practice Analysis surveyed over 2,000 genetic counselors to determine and define the skills and activities associated with the practice of genetic counseling. The ABGC Certification Exam Committee (CEC), a group of nine genetic counselors and one clinical geneticist with diverse clinical and demographic backgrounds, reviewed each item to make certain the items were clear, correct, supported by a reference, and that they assessed the proper content. Ultimately, the CEC reviewed all items several times before they were used on the exam. Contracted psychometricians helped guide and facilitate the exam development process, but Board-certified genetic counselors and geneticists finalize all exam items. Jointly, this expertise ensures the validity of the exam.

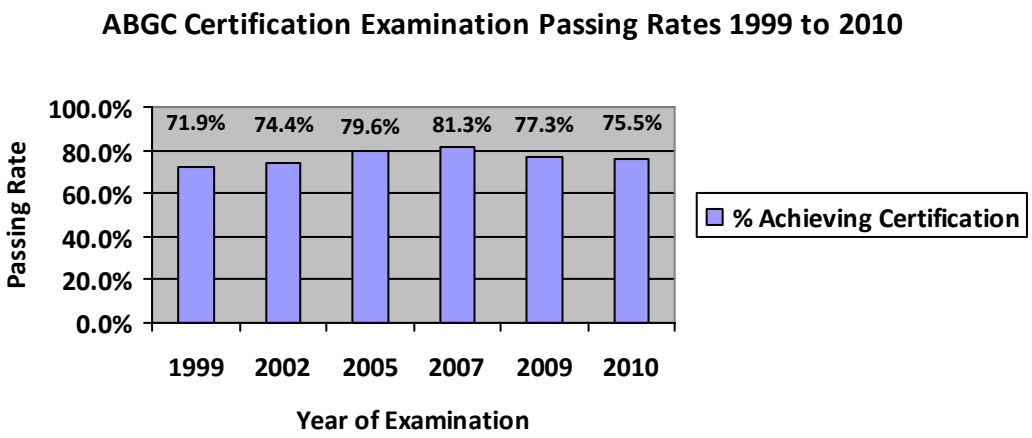
The 2010 ABGC certification exam window occurred between August 16th and September 18th, 2010. During this period 314 candidates took the exam; only one reported a test administration problem. This mirrors the 2009 test administration experience (with only four reported problems), and represents a significant improvement over the difficulties experienced in 2002 and 2007 with our previous exam vendor.

As a reminder, following the examination administration, a statistical analysis of the examinees' responses to the items is performed. The ABGC CEC reviews all exam items

that examinees have commented on, as well as those that show unusual statistics such as a high percentage of high-performing examinees choosing the incorrect answer. Poor-performing items are adjusted or eliminated so candidates are not penalized. This process guarantees fair treatment for *all* candidates, as well as valid pass/fail decisions.

We're happy to announce that, for the 2010 certification exam, 237 out of the 314 candidates earned a passing score, for an overall passing rate of 75.5%. As shown in Table 1, this rate is consistent with previous years, including the single-format examination of 2009 and the two-part ABGC/ABMG combined exam (pre-2009). Although passing rates across the years are similar, there is no set passing rate for the certification exam. The final passing score is determined using a process called the Angoff method, which is based on multiple factors and is not determined until after the examination is administered.

Table 1. ABGC Certification Examination Passing Rates 1999-2010



People passing the 2010 certification exam include 2009 graduates, 2010 graduates, graduates prior to 2009, repeat test takers, international candidates who achieved active candidate status prior to 2010, and genetic counselors seeking re-certification.

We understand that candidates taking the certification exam spend a significant amount of time preparing for the exam, and it can be hard to gauge the right time in one's life or

career to take or re-take the exam. Table 2 shows the detailed passing statistics for different groups of individuals who took the 2010 examination.

Table 2. 2010 ABGC Certification Examination Passing Statistics – Detailed

Group	Number Tested	Passing rate	Number Passed
All candidates	314	75.5%	237
2009 graduates	162	80.8%	131
2010 graduates	52	78.8%	41
1st time examinees	241	78.0%	188
Repeat examinees (all)	72	66.7%	48

Although this was the first time in many years that new graduates were allowed to sit for the exam, there does not appear to be a clear advantage or disadvantage to taking the exam directly after graduation because the passing rates appear to be relatively unchanged.

Finally, the ABGC Board of Directors would like to commend all of the candidates for the 2010 certification exam for their efforts to obtain certification. In addition, we'd like to encourage all genetic counselors not currently certified to work towards certification in the near future.

Reference

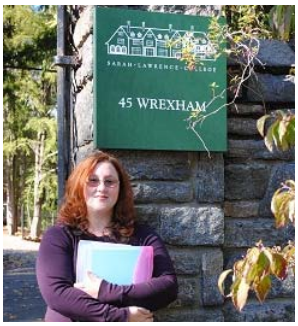
Hampel H, Grubs RE, Walton CS, Nguyen E, Breidenbach DH, Nettles S. American Board of Genetic Counseling 2008 Practice Analysis Advisory Committee, Callanan N, Corliss M, Fox S, Hiraki S, Ku L, Neufeld-Kaiser W, Riley B, Taylor J, Weik L. "Genetic Counseling Practice Analysis." *J Genet Couns.* 18(3):205-16. June 2009.

Student Forum

Part-Time Student, Full-Time Life

By Christine Colón, The Joan H. Marks Graduate Program in Human Genetics at Sarah Lawrence College, Class of 2011

Editors' Note: *We are pleased to welcome Christine (Chris) Colón to the* Perspectives *team as our new Editor for "Student Forum," and thought it fitting to have her introduce herself to all of us through her column.*



I entered the Joan H. Marks Graduate Program in Human Genetics at Sarah Lawrence College in Bronxville, New York in Fall 2008. Being a native New Yorker, I was excited to find a program so close to home. Alongside my classmates, I spent countless hours over the next two years studying the complex genetic material, completing clinical rotations, and laboring over an original thesis project. As a class of twenty-one students we celebrated holidays, worried over deadlines, and shared life's ups and downs together. So when graduation rolled around in May 2010, there were very few dry eyes to be found. After two years of very hard work, graduation day was finally here – at least, for my classmates. As a part-time student, I still had another year left to go.

One of the great aspects about Sarah Lawrence's program is the opportunity for students to complete the curriculum on a part-time basis, if need be. While most people may not want to extend their time in school (at least, that is the way I first felt), sometimes things happen beyond the individual's control. For example, two of my classmates who had started on a full-time schedule gave birth during their studies (one to twins!). Instead of having to leave school altogether, accommodations were made for their special circumstances. Now they are graduating with me in 2011, also as part-time students. When unexpected circumstances arise, having the option for part-time study can make the difference between finishing or dropping out of the program.

After initially meeting my classmates in 2008, I noticed some stark differences in our personal lives right away. Most of them were single, without children and had spent some time after completing their undergraduate program gaining unique experiences. For me, having two children by age twenty-four left no time for college. I started my

undergraduate program at age twenty-five, when my daughter was five months old. I graduated with a Bachelor of Science in Psychology in December 2007 and immediately applied to the Sarah Lawrence program. My life was filled to the brim – husband, two school-aged children, and a full-time job. Still, I entered my training full of enthusiasm. I was hoping to maintain a delicate balance of work, school and family and was successful for a short time. Soon however, it all came tumbling down after a series of unexpected family complications.

Life became too hectic; I was being pulled in too many different directions at once. The pressure and stress of my numerous responsibilities was overwhelming. Completing such an intense program full-time seemed impossible to me. I was convinced that my goal of becoming a genetic counselor would have to be put on hold indefinitely, until I found I was able to go part-time.

I was a bit apprehensive in the beginning – I of course wanted to graduate with my friends, and was eager to get out into the field and practice on my own. However, I knew I could not succeed under such stressful conditions; it was just too much. So I put my initial disappointment aside, chose to continue part-time, and vowed to make the best of things. It was quite a relief to have this option.

Almost immediately, I saw and felt a difference. I was able to spend more time with my family and help work out some difficult situations. I was able to space out my course load, which allowed me to focus on absorbing the material in a way I could understand in the present, and apply in the future. I was able to complete my capstone/thesis project during my second year, which was a tremendous burden off my shoulders. While it was an endeavor I thoroughly enjoyed, it was a huge amount of work. I was happy to be able to complete it, and to be one step closer to graduation.

Having an extra year of school also meant extra clinical rotations. I feel that I gained some very valuable and unique experiences during that additional time. As I get closer to completing my required hours, I feel more confident with not only the material, but with patients as well. Finally, now that I have a little more time than the average student finishing up his or her program, I can focus on some interesting projects outside of school, such as being the new “Student Forum” editor for *Perspectives*, being the Student Coordinator for the New York chapter of the March of Dimes, and giving genetics presentations to my son’s junior high school science class.

Most people have a plan when thinking about how they want the course of their lives to unfold – whether its school, family, career, or other events. I did as well. When things did not go according to plan, it was frustrating and frightening. For me, it was also an important learning experience. I discovered one size does not fit all in life. You have to roll with the punches, adapt, and make the most of what you have in the moment. I have learned that although I was forced to make many twists and turns along my journey, for right now, I am exactly where I need to be.

The New Graduate Life

Exploring the "ART" of Genetic Counseling for PGD

By Andria Besser, BEd, MS

I never expected to specialize in pre-implantation genetic diagnosis (PGD). In fact, during the first half of my graduate studies at the Mount Sinai School of Medicine in New York City, I was barely aware of the existence of PGD. Throughout my clinical rotations, I would mention PGD to patients as a reproductive option during discussions of carrier screening and smiled at their shocked reactions ("Wow, this sci-fi stuff really exists?"). It wasn't until my class volunteered at Camp Sunshine, a Maine retreat for children with life-threatening illnesses and their relatives, that I realized PGD truly is a viable option for many families. It was incredible to see firsthand how the use of this technology changed the lives of so many people, and it quickly grabbed my attention. Upon returning to Sinai for my second year and beginning the dreaded job search, I began to think about which area of genetic counseling would best suit my varied interests. I always enjoyed working with children and was intrigued by the diagnostic dilemmas that arose in a pediatric genetics clinic. I was also very interested in reproductive medicine, so I became fixated on finding a position that would allow me to work as both a prenatal and pediatric genetic counselor.

When I saw a job posting for a PGD genetic counselor, a kink suddenly formed in my focused life plan. Something about the position was calling to me, and although I wasn't quite sure what it was, I knew that it was just *right*. As I wrote my cover letter to apply for the position, I started to figure out why this job seemed so intriguing. While pursuing my undergraduate degree, I took a class in Reproductive Ethics (which even included a subset of classes that specifically focused on PGD). In my physiology class, I chose to participate in a laboratory project to perform *in vitro* fertilization (IVF) on cows and made sure to visit the university's IVF lab. My interests in fertility, pregnancy, and genetics were what drew me to the field of genetic counseling in the first place, and I couldn't think of a better place to put those interests to work than in a laboratory that

provides genetic testing with the aim of offering a new reproductive option to people in need.

After my interview with Reproductive Genetics Institute (RGI), I was over the moon with excitement that I may have found my perfect job. The more I considered it, however, the more I began to have some second thoughts. I debated whether working for a private company rather than a hospital or clinic was "selling out." I knew that, in addition to genetic counseling, part of the job would be marketing RGI as a company, and like many fellow genetic counselors, sales is *not* my forte. In the end, however, I couldn't resist the idea of working in a field that is so fascinating, where I knew I could become an expert in a specialized area that is not well known among genetic counselors.

And I was right – working as a PGD counselor has been a tremendously interesting and engaging experience. I admit that the learning curve has been exceptionally steep and sometimes I feel as though I can never learn all of the ins and outs of the PGD industry, but it is extremely rewarding to have such specialized knowledge in one of the most up-and-coming fields in genetics.

From the beginning, I knew that I wanted to work in an environment where I would have substantial patient contact. Although I knew this desire could change in the future, I wasn't ready to begin working in a research coordinator or non-clinical laboratory role. When I interviewed with RGI, it was explained that, since most patients are not in close proximity to our Chicago location, the majority of my counseling would be performed over the phone, with only sporadic in-person consults. It was also discussed that a portion of my job would be spent coordinating patient care instead of directly providing it. I was concerned that the amount of patient contact might not compare to what I was looking for, but I soon learned that seeing patients all of the time is not the be-all, end-all of genetic counseling. The more I thought about it, I realized that seeing patient after patient after patient would be exhausting! I can imagine that type of position taking an emotional toll on me, and actually appreciate being able to do paperwork while recuperating from a difficult patient encounter.

On a similar note, however, I was concerned that telephone counseling might not provide as much exposure to the psychosocial aspects of PGD, and I admit that I do still prefer to meet with patients in person. However, I have found that since phone counseling allows for some degree of anonymity, patients often tend to open up more than they might if we were meeting in person.

I love being one of a few in a unique field that is rapidly expanding. It has opened up a whole new world to me within the realm of genetic counseling: the world of assisted reproductive technologies (ART). This year, in addition to the NSGC Annual Education Conference in Dallas, I was able to represent RGI at the American Society of Reproductive Medicine (ASRM) meeting in Denver. ASRM was a very different experience (as can be expected of a conference of 8,000 attendees!), but attending the conference really enabled me to feel like a part of the ART community.

I agree with other genetic counselors in the field that say, with PGD counseling, the highs are very high and the lows are very low. We deal with a high proportion of couples who have lost a child or who have a son or daughter with a severe disability. IVF with or without PGD is not an easy method through which to achieve pregnancy, and repeated failed cycles are common. Each negative pregnancy test is devastating, while a positive pregnancy test brings tremendous joy. Although it is often emotionally difficult, it is extremely satisfying to feel that we are truly making a difference in these patients' lives.

Although I'm only in the fifth month of my career, working as a PGD counselor has been enormously challenging and rewarding. I enjoy playing a variety of roles, including genetic counselor, IVF/PGD coordinator, and educator. Even the sales part of my job has been easier to deal with than I initially expected. I guess when you believe so much in a product or service, it just comes naturally to want others to feel the same way!

Genetic Counselor Publications

By Jamie C. Fong, MS, CGC

Standing (left to right): Danielle Campfield Bonadies, Ellen T. Matloff
Seated: Karina L. Brierley

Featured article

(Names of genetic counselors appear in bold)

Brierley KL, **Campfield D**, **Ducaine W**, **Dohany L**, **Donenberg T**, **Shannon K**, **Schwartz RC**, **Matloff ET**. Errors in Delivery of Cancer Genetics Services: Implications for Practice. *Connecticut Medicine*. 74(7):413-423. 2010.

Forget about reading a horror novel for a good scare. For those immersed in human genetics, look no further for hair-raising stories than the article, “Errors in Delivery of Cancer Genetics Services: Implications for Practice,” in the August issue of *Connecticut Medicine*. Rather than featuring horror archetypes like zombies or other worldly monsters, however, the article by **Karina L. Brierley**, **Danielle Campfield Bonadies** (published under **Campfield**), and **Ellen T. Matloff** of Yale Cancer Center at Yale School of Medicine in New Haven, Connecticut, along with colleagues **Whitney Ducaine**, **Lindsay Dohany**, **Talia Donenberg**, **Kristen Shannon**, and **Robin C. Schwartz**, presents a nationwide series of cases that illustrate negative outcomes of cancer genetic testing performed without appropriate genetic counseling by a qualified provider.

Co-authors Karina, Danielle, and Ellen chuckle at the suggestion that their article could stand in for a horror novel, but the current state of genetic testing for cancer predisposition syndromes – as facilitated by uninformed or under-informed providers – is for them no laughing matter. They are upset by reports of negative outcomes and are motivated to do something about it.

They became aware of the matter a number of years ago when anecdotes first appeared on the National Society of Genetic Counselors (NSGC) Cancer Special Interest Group (SIG) listserv. According to Karina, Danielle, and Ellen, these anecdotes described

patients with a variety of experiences, including undergoing a genetic test for which a patient's personal and family histories of cancer were not suggestive, undergoing a genetic test without receiving appropriate genetic counseling, and receiving a misinterpreted genetic test result. The frequency of anecdotes on the listserv gradually increased over the years, and as Karina, Danielle, and Ellen also encountered patients with previous negative experiences of genetic testing, they decided to collect these stories systematically and report them.

"We kept hearing it all over the country, but no one had documented it," said Ellen. "At the same time, in parallel, all these other publications were saying that everyone can do their own genetic testing. You [providers] don't need to offer genetic counseling. You can just do testing in your own office with the [test company's] brochure. So it really seemed like it was time for a group of people to document these cases."

In the spring of 2009, Karina, Danielle, and Ellen invited members of the NSGC Cancer SIG listserv to submit cases of interest. They required each submission to have a multi-generational pedigree and a detailed case description that included: information about a patient's clinical presentation, the delivery of genetic counseling (if it occurred at all) throughout the testing process, the provider who facilitated the testing and/or offered genetic counseling, and the occurrence of adverse events that followed disclosure of genetic test results.

Karina, Danielle, and Ellen recall receiving many responses, including one from a patient who contacted them directly with her story after learning about the case series through a cancer advocacy website. The co-authors then had the difficult task of narrowing down the number of cases for their manuscript. They settled on twenty-one vignettes.

"We chose cases that were most striking or most representative of the themes we were trying to convey," explained Karina. "We also wanted cases that reflected some variety."

One story is so unnerving that it sends chills down the spine. The co-authors described three healthy sisters with a striking family history of early-onset breast and ovarian cancer who were told by their family's gynecologist that the cancers in the family were hereditary. The sisters were also told that *BRCA1/2* genetic testing, though clinically available at the time, was unnecessary because their cancer risks were unequivocal in the context of the family history. At the advice of the gynecologist, the three sisters underwent risk-reducing total hysterectomies at ages 33, 34, and 36 years, respectively. All three experienced surgical menopause without the potential relief of hormone replacement therapy (HRT) because they were told they were poor candidates for HRT, given their family history of breast cancer. A few years later, the eldest sister participated in a research study on menopausal therapy and was referred to a genetic counselor, who suggested the sisters' mother should also be referred. The mother, the closest living relative with a history of early-onset breast and ovarian cancer, eventually sought genetic counseling and testing, and learned she carried a *BRCA1* gene mutation. The woman's daughters, the three sisters, subsequently underwent testing for the known familial

mutation. All three received the same news: each had a true negative test result, and each had not inherited her mother's mutation.

The sisters were devastated. The untimely, menopause-inducing surgery each had experienced had been unwarranted. At least one of the sisters, who had wanted more children, underwent the surgery because she had been convinced that her chance of dying from cancer was too great. She could not burden her children with her death, let alone entertain a desire to have more children.

That the gynecologist was neither callous nor intentionally negligent poignantly underscored the sisters' experiences. "[The gynecologist] really cared about them. He was just uninformed. There was no malice. He really thought he was giving them the right information," said Ellen. But following receipt of their true negative test results, the sisters felt angry about having had unnecessary surgery. "It was sad. It was also a sharp lesson for the physician," Ellen warned.

Unmistakably an adverse event that resulted from inadequate genetic counseling, the unwarranted risk-reducing surgery of the sisters' story represented only one end of the spectrum of outcomes that emerged from the case series. The co-authors also identified negative outcomes such as psychosocial distress, false reassurance about cancer risk resulting in inappropriate medical management, and unnecessary genetic testing. The co-authors went so far as to quantify the impact of unnecessary testing, calculating the approximate amount of wasted healthcare dollars that occurred in some cases. For example, $2,865 and $2,765 were unnecessarily spent in cases in which complete sequencing of *BRCA1/2* was inappropriately performed rather than familial mutation testing or common Jewish *BRCA1/2* mutation testing, respectively.

According to Karina, Danielle, and Ellen, one way to combat the growing number of cases with negative outcomes is to improve provider education and to establish a framework in which providers could identify patients who would benefit from a referral to an experienced cancer genetics professional, like a genetic counselor. To be clear, the co-authors do not contend that non-genetics professionals are completely ill-suited to provide genetic counseling. However, they firmly believe that inadequate genetics education, insufficient experience with cancer predisposition syndromes, time constraints, provider fatigue, and genetic complexity of patient cases interfere with the ability of non-genetics professionals to provide comprehensive cancer genetic services.

"[Provider education] is something that our office is very dedicated to," said Danielle. "We're trying to educate physicians, nurses, physician assistants on the risk factors [of a cancer predisposition syndrome] to look for in families so that the providers can try to identify patients who are candidates for genetic counseling. We also try to educate them on what to do with these patients once a hereditary syndrome has been identified. We try to point out the complexities of genetic counseling and encourage them to use us as a referral service."

The biggest obstacle to minimizing negative outcomes, the co-authors maintain, is the largest biopharmaceutical company that markets directly to non-genetics professionals. They claim that the company promotes the use of its genetic tests by emphasizing the potential benefits of having genetic information, a position that the co-authors do not decry, but they do criticize the company for promoting its products while overlooking or minimizing the complexities of genetic counseling and testing. Karina, Danielle, and Ellen assert that the company's take-home message to non-genetics professionals is this: genetic testing is straightforward; any provider can easily order a test and interpret the results.

Not lost on Karina, Danielle, and Ellen is the observation that the biopharmaceutical company is a significant player in the cancer genetics community. The co-authors claim it employs the largest number of genetic counselors in the country. They contend that the company's large presence, with its stifling entrepreneurial might, has instilled undue fear among genetics providers who wish to raise awareness about the incidence of negative outcomes that result from genetic tests performed without genetic counseling by a qualified provider but who are too afraid to speak publicly. These genetics providers prefer anonymity, or they remain altogether silent, fearful that any negative comments about the company will have negative personal repercussions. For all they know, they might one day in the future be placed in a position to seek employment with the company and would not want to sabotage any potential job opportunities because they had been associated with a publication, or other activity, that portrayed the company negatively.

Karina, Danielle, and Ellen, however, are not easily daunted. They are proud of the efforts that they and colleagues have made to collect and document cases with negative outcomes. They plan to continue to report stories like that of the three sisters. If it seems like the co-authors are trying to scare people into re-evaluating the widely marketed notion that cancer genetic testing is a simple process and that any provider could tackle it, at least they are doing so with purpose. They are doing so for their patients and families.

"This meant a lot to me personally," said Ellen, about the importance of this case series. "It was our responsibility to get this published."

Genetic Counselor Publications

By Jamie C. Fong, MS, CGC

Articles co-authored by genetic counselors
February 2010 – October 2010
(Names of genetic counselors appear in bold)

Ambhekar SK, Carroll JL, **Keiles S.** Report of two patients with associated conditions in addition to cystic fibrosis. *J Cyst Fibrosis*. 9(4):269-71. 2010.

Annes JA, **Giovanni MA**, Murray MF. Risks of presymptomatic direct-to-consumer genetic testing. *NEJM*. 362(12):1100-1. 2010.

Bradbury AR, Patrick-Miller L, Egleston B, Sands CB, Li T, Schmidheiser H, Feigon M, Ibe CN, Hlubocky FJ, Hope K, **Jackson S**, Corbman M, Olopade OI, Daly M, Daugherty CK. Parent opinions regarding the genetic testing of minors for *BRCA1/2*. *J Clin Oncol*. 28(21):3498-505. 2010.

Brown SB, **Culver JO**, Osann KE, Macdonald DJ, Sand S, Thornton AA, Grant M, Bowen DJ, Metcalfe KA, Burke HB, Robson ME, Friedman S, **Weitzel JN**. Health literacy, numeracy, and interpretation of graphical breast cancer risk estimates. *Patient Educ Couns*. 2010 May 27. [Epub ahead of print]

Campfield Bonadies D, Moyer A, **Matloff ET**. What I wish I'd known before surgery: *BRCA* carriers' perspectives after bilateral salpingo-oophorectomy. *Fam Cancer*. 2010 Sep 18. [Epub ahead of print]

Churpek JE, Garcia JS, Madzo J, **Jackson SA**, Onel K, Godley LA. Identification and molecular characterization of a novel 3' mutation in *RUNX1* in a family with familial platelet disorder. *Leuk Lymphoma*. 51(10):1931-5. 2010.

Eisenbraun A, Wenstrup R, Hellerstedt B, Patton J, Reid J, Sailer L, **Copeland K**, **Brunger J**, Hamilton S, **Frye C**, Sandbach J. Hereditary breast and ovarian cancer testing: Integration and outcomes within community oncology practices. *Commun Onco*l. 7:75-81. 2010.

Elden AC, Kim HC, Hart MP, Chen-Plotkin AS, Johnson BS, Fang X, Armakola M, Geser F, Greene R, Lu M, Padmanabhan A, **Clay-Falcone D**, McCluskey L, Elman L, Juhr D, Gruber PJ, Rüb U, Auburger G, Trojanowski JQ, Lee VM, Van Deerlin VM, Bonini NM, Gitler AD. Ataxin-2 intermediate-length polyglutamine expansions are associated with increased risk for ALS. *Nature.* 466(7310):1069-75. 2010.

Ersig AL, Ayres L, **Hadley DW**, Koehly LM. Explanations of risk in families without identified mutations for hereditary nonpolyposis colorectal cancer. *J Nurs Scholarsh*. 42(2):139-46. 2010.

Fowler E. Hereditary cancer: The challenges are pedigree interpretation, risk assessment, and management-not genetic testing. *Commun Oncol*. 7:82-83. 2010.

Giovanni MA*, Fickie MR*, Lehmann LS, Green RC, Meckley LM, Veenstra D, Murray, MF. Health care referrals from direct-to-consumer genetic testing. *Genet Test Mol Biomarkers*. 2010 Oct 28. [Epub ahead of print]

González-Hormazábal P, Gutierrez-Enriquez S, Gaete D, Reyes JM, Peralta O, Waugh E, Gómez F, **Margarit S**, Bravo T, Blanco R, Diez O, Jara L. Spectrum of BRCA1/2 point mutations and genomic rearrangements in high-risk breast/ovarian cancer Chilean families. *Breast Cancer Res Treat*. 2010 Sep 22. [Epub ahead of print]

Jamal SM, Basran RK, **Newton S**, Wang Z, Milunsky JM. Novel de novo *PCDH19* mutations in three unrelated females with epilepsy female restricted mental retardation syndrome. *Am J Med Genet A*. 152A(10):2475-81. 2010.

Jara L, Dubois K, Gaete D, de Mayo T, Ratkevicius N, Bravo T, **Margarit S**, Blanco R, Gómez F, Waugh E, Peralta O, Reyes JM, Ibáñez G, González-Hormazábal P. Variants in DNA double-strand break repair genes and risk of familial breast cancer in a South American population. *Breast Cancer Res Treat*. 122(3):813-22. 2010.

Kemper AR, Knapp AA, Green NS, Comeau AM, **Metterville DR**, Perrin JM. Weighing the evidence for newborn screening for early-infantile Krabbe disease. *Genet Med*. 12(9):539-43. 2010.

Kirkpatrick BE, **El-Khechen D**. A unique presentation of 22q13 deletion syndrome: Multicystic kidney, orofacial clefting, and Wilms' tumor. *Clin Dysmorphol*. 2010 Sep 6. [Epub ahead of print]

Lea DH, Kaphingst KA, Bowen D, Lipkus I, **Hadley DW**. Communicating genetic and genomic information: Health literacy and numeracy considerations. *Public Health Genomics*. 2010 Apr 20. [Epub ahead of print]

Matloff ET, **Brierley KL**. The double-helix derailed: The story of the *BRCA* patent. *The Lancet*. 376(9738):314-15. 2010.

Nowaczyk MJM, **Huggins MJ**, Fleming A, Mohide PT. Femoral-facial syndrome: Prenatal diagnosis and clinical features. Report of three cases. *Am J Med Genet A*. 152A(8):2029-33. 2010.

Perrin JM, Knapp AA, Browning MF, Comeau AM, Green NS, Lipstein EA, **Metterville DR**, Prosser L, Queally D, Kemper AR. An evidence development process for newborn screening. *Genet Med*. 2010 Feb 11. [Epub ahead of print]

Roberts JS, Karlawish JH, **Uhlmann WR**, Petersen RC, Green RC. Mild cognitive impairment in clinical care: A survey of American Academy of Neurology members. *Neurology*. 75(5):425-31. 2010.

Wax JR, Pinette MG, Mallory B, **Carpenter M**, Winn S, Cartin A. Prenatal sonographic diagnosis of a perineal lipoma. *J Ultrasound Med*. 29(8):1257-9. 2010.

Book chapters co-authored by genetic counselors
(Names of genetic counselors appear in bold)

Edwards JG. (2010). Contemporary Genetic Counseling. In W.F. Rayburn (Consulting Ed.), A.R. Gregg & J.L. Simpson (Guest Eds.). *Obstetrics and Gynecology Clinics of North America*: *Vol. 37. Genetics Screening and Counseling* (pp. 1-9). Philadelphia: W.B. Saunders Co.

Giovanni MA, Murray MF. (2010). The application of computer-based tools in obtaining the genetic family history. *Curr Protoc Hum Genet*. 66:9.21.1-9.21.9.

Walker P, Gregg AR. (2010). Screening, Testing, or Personalized Medicine: Where do Inherited Thrombophilias Fit Best? In W.F. Rayburn (Consulting Ed.), A.R. Gregg & J.L. Simpson (Guest Eds.). *Obstetrics and Gynecology Clinics of North America*: *Vol. 37. Genetics Screening and Counseling* (pp. 87-107). Philadelphia: W.B. Saunders Co.

* The authors contributed equally to this work.

Please send "Genetic Counselor Publications" submissions to Jamie Fong at jaf2025@med.cornell.edu

AEC Update

Save the Date for the 2011 AEC in San Diego, California

By Elizabeth Wood Denne, MS, CGC, 2011 AEC Chair, and
Claire N. Singletary, MS, CGC, 2011 AEC Vice-Chair

In 1981, the first NSGC Annual Education Conference was held in San Diego, California. Planning is already underway for the NSGC's return to San Diego for the 30th Annual Education Conference (AEC) from **October 27-30, 2011.**

The AEC will be held at the waterfront San Diego Marriott Hotel and Marina, which is less than five miles from the San Diego International Airport (SAN). The Marriott provides beautiful views of the San Diego Marina and is adjacent to Seaport Village, which features ample waterfront dining and shopping. The Gaslamp Quarter, also within walking distance, is famous for its great restaurants and shopping. The world famous San Diego Zoo is less than five miles from the hotel, as is historic Old Town San Diego.

AEC Format Continues in 2011

Pre-Conference Symposia will take place on Thursday, October 27, 2011 and the AEC will again begin with the "Welcome to the AEC" orientation, followed by the opening plenary Janus Series and Best Abstract Awards. Concluding this kickoff will be the Welcome Reception in the Exhibitor Suite on Thursday evening. There will be two full days of outstanding educational opportunities within the Plenary and Educational Breakout Sessions (EBS) on Friday and Saturday, followed by a shorter day on Sunday and the conference's conclusion in the afternoon.

Call for Speakers – Opened December 1, 2010!

The NSGC is actively inviting members to submit presentation proposals for **Plenary Sessions**, **Educational Breakout Sessions and Pre-Conference Symposia**. We are seeking informative and stimulating presentations by genetic counselors, physicians, researchers and other industry leaders that will help advance our knowledge within the profession of genetic counseling. **The Call for Speakers opened on December 1, 2010 and the deadline for submission is January 14, 2011.** Applicants will be notified of their acceptances in February. Watch for additional Submission Guidelines and Information in upcoming NSGC e-mails and on the NSGC Web site.

Submission Guidelines

AEC presentation submissions need to contain a brief descriptive paragraph outlining the presentation, as well as three learning objectives written to the continuing education standards in the electronic submission. A Plenary session is typically one hour in length, while an EBS is typically two hours. A Pre-Conference Symposium is five hours of content. Presentations may include more than one speaker and encompass two or more integrally related topics. All presentations must be educational in nature and not include any sales, product or marketing information. Speakers are encouraged to prepare and present original material. Members of the 2011 AEC Planning Subcommittee will carefully review all submissions. Proposals may be considered for other session formats, in addition to the format requested.

Please note: If you are submitting/coordinating the proposal but do not plan to speak, you will have the opportunity to list yourself as such. You will be considered the main contact for the proposal. You will also need to identify the speakers. *It is important that the Subcommittee knows ALL speakers for each proposal during their review, as this will impact the sessions selected.*

Submit your proposal by completing the online submission. Instructions and the submission form can be found at the following link: http://www.nsgc.org/Education/2011AnnualEducationConference/NSGCSpeakerCenter/CallforSpeakers/tabid/331/Default.aspx Any questions can be directed to the AEC Chair, **Beth Wood Denne**, at ewdenne@jhmi.edu and Vice-Chair, **Claire Singletary** at claire.n.singletary@uth.tmc.edu or to nsgc@nsgc.org.

Submissions become the property of the NSGC and will not be returned. The NSGC has the right to publish each selected submission in promotional materials, such as the AEC Preliminary Program.

The 2011 AEC Subcommittee

Please contact the Subcommittee members with ideas, comments and suggestions:

AEC Chair
Elizabeth Wood Denne, ewdenne@jhmi.edu

AEC Vice-Chair
Claire N. Singletary, Claire.n.singletary@uth.tmc.edu

AEC Planning Subcommittee
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Quinn Stein, quinn.stein@sanfordhealth.org
Meredith Weaver, mweaver@acmg.net

Emily Windsor, emily.windsor@gmail.com

Call For Abstracts

Abstracts that are of interest to the genetic counseling profession and related fields are being accepted for consideration as platform or poster presentations from **March 21, 2011 to May 16, 2011**. Student members, non-members and Full members are encouraged to submit abstracts. Monetary awards will be presented for best Full member and Student member abstracts. Starting on March 21, 2011, guidelines and instructions for submission of abstracts can be found on the National Society of Genetic Counselors website at www.nsgc.org.

Questions may be addressed to the Executive Office at nsgc@nsgc.org or the Abstract Workgroup Chair, **Carrie Atzinger** (Carrie.Atzinger@cchmc.org) or Abstract Workgroup Vice-Chair, **Brandie Leach** (leachb@ccf.org).

Resources / Book Review

Reviewed by Shivani Nazareth, MS, CGC

The Story of Forgetting

Author: Stefan Merrill Block
Publisher: Random House, 2008
Pages: 310
Retail price: $25.00
ISBN-10: 0812979826
ISBN-13: 978-0812979824

The Story of Forgetting creatively weaves together three stories about the psychological impact of hereditary Alzheimer's disease on various residents of a small English town named Iddylwahl. It is a skillfully crafted novel and, as one gets absorbed in the plot, it becomes clear that the author — a 28-year-old Brooklyn resident — did his share of homework about genetics and the far-reaching implications of memory loss. Stefan Merrill Block manages to inject humor into a serious topic by exemplifying the founder effect in a provocative way: the main character conducts love affairs with nearly one hundred women in the town and promptly forgets what he has done. The women enjoy the anonymity of their affairs, but the next generation of Iddylwahl children carries on the legacy of their father's disease. The author artfully describes that the mutation, which he refers to as the EOA-23 alteration, "began to call upon his loose phosphates to clog the microtubule tracks that held the neurons of his brain in place, causing them to fray and recoil like hair over a flame."

Alongside this plot is the story of a man named Abel, who is in love with his brother's wife, Mae. When Mae conceives a child, the author poetically illustrates the human tendency to selectively forget painful moments of life. The choice to forget is juxtaposed against the disease of forgetting in a way that speaks to the heart of the reader.

The third plotline is narrated by a teenager named Seth, who witnesses his mother's mental decline and articulates his father's inability to cope, as well as his own curiosity about the origins of the disease. His search for answers leads him to a group of researchers who unknowingly help him understand his own fate. Interwoven throughout the novel are glimpses of an imaginary world called Isadora, where memory does not exist and life is consequently free from pain.

The Story of Forgetting provides the reader with an honest depiction of the complexity of the human mind and the relationships we forge based on our perceptions of the world. For genetic counselors, it is not only a memorable read, but an informative one as we attempt to navigate our patients' emotional turmoil. We are privileged to participate in our patients' lives at what are often very vulnerable times, and this book reminds us of

the heavy burden that travels alongside inherited diseases. To the credit of this young debut author, The Story of Forgetting is definitely not a story worth forgetting.

Media Watch

By Claire Noll, MS, CGC

(Names of genetic counselors appear in bold)

Editors' Note: *After much consideration and review of member feedback, we have decided that this will be the final "Media Watch" column to appear in* Perspectives. *While the column was initiated as a way to help grow awareness about our profession and colleagues' activities, the natural evolution of our field makes these needs less applicable today. We thank Claire Noll, the column's Editor, and those before her for their dedication to* Perspectives *and to "Media Watch" in particular.*

July 26, 2010 – *KUSI.com* (www.youtube.com/watch?v=HljAuCoAk9c)
"Tracing your Family Cancer History"
A local morning news show highlighted the importance of taking one's own family history of cancer. **Lisa Madlensky** explained what types of cancer are most important to record, the difference between primary and metastatic cancers, how to obtain medical records, and what will be done if someone is found to carry a *BRCA* mutation.

August 29, 2010 – *lancasteronline.com*
(http://articles.lancasteronline.com/local/4/282553)
"Expanding genetic testing center helps high-risk patients plan their treatment"
Two local patients, one female and one male, discussed the ways in which positive *BRCA* test results affected their lives and families. **Rachelle Gehr** described the associated cancer risks and treatment options. The volume of referrals at the center has increased such that a second genetic counselor, **Erin Sutcliffe**, was recently added to the staff.

August 31, 2010 – *WCIU.com*
(www.wciu.com/youandme.php?section=home&assets=videos&assetID=10003361)
"DNA Tests for Kids?"
The hosts of a morning TV show discussed talent camps in China that are reported to use genetic testing to make predictions about a child's future personality and skills. **Sara Cherny** explained that such tests may not be helpful, because gene-environment interactions may affect children in ways that cannot be predicted.

September 2, 2010 – *Reno Gazette-Journal*
(www.rgj.com/apps/pbcs.dll/article?AID=2010100902036)
"People you should get to know: Robbin Palmer"
A column to introduce notable local people described **Robbin Palmer's** background, interests, and motivations, as well as how she participates in her local community and why she likes where she works.

September 16, 2010 – *The Jewish Chronicle*
(www.thejewishchronicle.net/view/full_story/9541630/article-Knowledge-is-crucial?instance=home_news_1st_left_title)
"Knowledge is crucial"
A new community program to screen Ashkenazi Jews for carrier status held an information session recently at which **Katie Long** answered questions.

September 24, 2010 – *nursezone.com* (http://www.nursezone.com/Nursing-News-Events/more-features/What-Nurses-Need-to-Know-About-Geneticsand-Where-to-Find-It_35254.aspx)
"What nurses need to know about genetics… and where to find it"
Alyson Krokosky was one of several experts who described the challenges nurses face when learning about genetic medicine, as they do not usually take a genetics course during their training. "With over 6,000 genetic conditions out there, it's hard to be the expert on any one condition," she noted, adding "But having some background knowledge in terms of inheritance and the fundamentals of biology, understanding such things as the importance of taking a family history will be important for everyone, nurses included."

September 29, 2010 – *WSPA.com* (www2.wspa.com/news/2010/sep/29/last-week-september-dedicated-hereditary-cancer-aw-ar-896597/)
"Last week in September dedicated to hereditary cancer awareness"
In conjunction with the designation of the last week of September as "National Hereditary Breast and Ovarian Cancer Week," **Karen Brooks** stressed the importance of knowing one's medical family history. "This information, that really isn't too tough to find, can save your life," she said.

September 30, 2010 – *WICD.com*
(http://www.wicd15.com/newsroom/top_stories/videos/vid_3128.shtml)
"Top Stories"
Jennifer Burton explained that the findings of a recent Swedish study indicate that if women are routinely screened for breast cancer beginning at age 40 years, many cases of cancer are caught early enough to reduce mortality. This is in contradiction to a recent American advisory that routine mammography should start at 50 years of age.

October 2, 2010 – *WABC.com*
(http://abclocal.go.com/wabc/video?id=7700980&syndicate=syndicate&section)
"Breast cancer: survival and health"
In a series of interviews of previvors and survivors of breast cancer, **Shivani Nazareth** described how paternal family history can make it appear as if a *BRCA* mutation is skipping generations. "The counseling really helps to clarify what the risks are and what the options are," she continued, before discussing the Genetic Information Nondiscrimination Act (GINA).

October 10, 2010 – *The Marietta Daily Journal*
(http://mdjonline.com/bookmark/9860768)

"Genetic testing may help treatment"
An interview with **Kimberly King-Spohn** addressed the questions of who should consider cancer genetic counseling and genetic testing, and the cost and turn-around time for *BRCA* testing, as well as the existence of other genes involved in breast cancer risk besides the *BRCA* genes.

October 14, 2010 – *wickedlocal.com* (www.wickedlocal.com/salem/news/lifestyle/health/x1389361878/Local-talk-on-role-of-genetics-lifestyle-in-breast-cancer)
"Local talk on role of genetics, lifestyle in breast cancer"
An announcement of a community workshop on the role of genetics and lifestyle in breast cancer mentioned **Kristen Shannon**, who provides counseling within that community.

October 20, 2010 – *EurekAlert.com* (www.eurekalert.org/pub_releases/2010-11/asoh-a2n102010.php)
"ASHG 2010: New research on implications of direct-to-consumer and clinical genetic testing"
This article reviewed some research results in advance of this year's annual meeting of the American Society of Human Genetics. One study mentioned was **Barbara Bernhardt's** project entitled "Public Understanding of and Reactions to Personalized Genetic Risk Information: Results from the Coriell Personalized Medicine Collaborative." She said, "Through our interviews with individuals receiving information about their genetic risk for common complex health conditions, we found that most of the study participants had a good understanding of personalized test results predicting their disease risk." Also highlighted was **Andy Faucett's** study entitled "The Use of Current Genetic Testing Oversight to Select the Best Test for Each Patient." He stated, "It is important for both healthcare providers ordering clinical genetic tests for their patients and individuals who are considering purchasing direct-to-consumer genetic testing services to evaluate these tests carefully before making decisions."

October 25, 2010 – *KVOA.com* (www.kvoa.com/news/breast-cancer-prevention-through-genetic-testing/)
"Breast cancer prevention through genetic testing"
A pair of sisters described their experiences with hereditary breast cancer associated with a *BRCA1* mutation. Their genetic counselor, **Jessica Ray**, described her job as helping patients decide what to do next, by "[h]elping them reduce their risk as much as possible."

October 28, 2010 – *ivanhoe.com* (www.ivanhoe.com/channels/p_channelstory.cfm?storyid=25548)
"Your paternal family history can save your life" and
October 29, 2010 – *businessweek.com* (www.businessweek.com/lifestyle/content/healthday/644952.html)
"Dad's family history of breast, ovarian cancer matters, too"

Jeanna McCuaig noticed that many of the patients referred to her center for breast and ovarian cancer counseling commented that they had not thought a paternal family history of cancer was relevant. After reviewing two years' worth of patient charts, she determined that patients were five times more likely to be referred if the family history of cancer was on the maternal side, adding "The lack of awareness that women may inherit a mutated gene from their fathers is also present among many healthcare providers."

November 7, 2010 – *ScienceDaily.com*
(www.sciencedaily.com/releases/2010/11/101107214516.htm)
"The use of current genetic testing oversight to select the best test for each patient"
This article summarized **Andy Faucett's** presentation at the annual meeting of the American Society of Human Genetics, in which he provided points for clinicians to consider before ordering genetic tests, as well as questions for consumers to ask before ordering direct-to-consumer genetic testing. "Most U.S. laboratories performing genetic testing do an excellent job of analysis -- however, that said, sometimes the test results may not provide the appropriate information to address the questions that the healthcare providers or patients are asking," he said.

Research Network

By Emily Place, MS, CGC

Autoimmune Diseases in Pregnancy Project

The Organization of Teratology Information Specialists (OTIS) is researching the effects of autoimmune diseases such as Crohn's disease, rheumatoid arthritis, psoriatic arthritis, ankylosing spondylitis, and psoriasis, as well as the medications used to treat these conditions during pregnancy. Participants will not be asked to take any medication as part of this study. Controls (women who do not have an autoimmune disease, but who are pregnant) are also being enrolled for this study. Visit the website to learn more about this study: www.otispregnancy.org/autoimmune-studies-s13049

Contact: Dee Quinn at (520)626-3547 or dquinn@email.arizona.edu

Vaccines and Medications in Pregnancy Surveillance System (VAMPSS)

The Organization of Teratology Information Specialists (OTIS) is researching vaccines and medications in pregnancy such as the H1N1 vaccine, seasonal flu vaccine, and antiviral medications. Participants will not be asked to take any medication or vaccines as part of this study. Eligible participants will be pregnant women who have already received the vaccines or taken antiviral medications. Pregnant women who have not received the vaccines or antiviral medications are also eligible to participate as controls. Visit the website to learn more about this study:

www.otispregnancy.org/vaccines-and-medications-in-pregnancy-surveillance-system-s13053

Contact: Dee Quinn at (520) 626-3547 or dquinn@email.arizona.edu

NTD Research at the University of Miami

The Hussman Institute for Human Genomics is looking for families to participate in research to identify the genetic and environmental factors that contribute to neural tube defects (NTDs). Any individual with a diagnosis of an NTD and his/her selected family members can participate, if willing. Participation is free and travel is not required. Participation involves reading and signing a consent form, providing a blood sample, a family and medical history interview, and granting the research staff permission to review the medical records of the individual(s) with the NTD. The highest standards of confidentiality are maintained for all families. Visit the website to learn more about this study: www.hihg.org

Contact: Maria Ciliberti at 1-877-686-6444 (toll free) or 305-243-4360; Mciliberti@med.miami.edu

Genetic Epidemiology of Pancreatic Cancer (PACGENE) Study

Researchers at Wayne State University, Mayo Clinic, Johns Hopkins University, MD Anderson Cancer Center, Dana Farber Cancer Institute, and the University of Toronto are aiming to map one or more pancreatic cancer susceptibility genes. The study is currently enrolling families with at least two cases of pancreatic adenocarcinoma. Participation includes phone interview or mailed questionnaire, medical record review, contacting family members and donation of a blood, tissue or saliva sample. Families will not receive individual test results. Affected individuals need not be living, however a DNA sample, such as tissue, must be available on at least one affected. Travel is not necessary; participants are compensated financially for their time. For more information, visit the website: www.karmanos.org/cancer.asp?id=927&cid=19

Contact: Kate Sargent, MS, CGC at 313-578-4240

Where's my answer? Understanding counseling issues surrounding VUS in BRCA1/2 genes

Individuals seek *BRCA1/2* testing for many reasons, including gaining certainty. Approximately, 5-15% of patients seeking *BRCA* testing receive a result of a variant of uncertain significance (VUS), a change that has an unknown application to cancer risk. The purpose of this study is to explore what patients are told about their uncertain variants and what methods are used to tell them. Patients will be asked to participate in a short online, anonymous survey. Any patient with a VUS is encouraged to participate, regardless of whether or not that VUS has been reclassified. Participants may be entered in to a drawing for a free iPod Shuffle. The survey can be found at www.surveymonkey.com/uncertainvariant

Contact: Megan Judkins at (801) 541-0753 or Megan.Blanksma@hsc.utah.edu

Simons VIP Connect

Simons VIP Connect (www.SimonsVIPConnect.org) has launched a new research study. The Simons Variation in Individuals Project (VIP) is characterizing individuals with 16p11.2 deletions and duplications. Both biological parents must participate and be willing to travel for a minimum of two days to one of the study sites which include Baylor University in Houston, Children's Hospital of Boston, and University of Washington in Seattle. The visit will include medical, neurological, and psychometric assessments and MRI. Research findings will be shared with the families. All expenses will be paid. A web-based community for 16p11.2 families has also been developed to

facilitate communication among these families. Please contact us with questions or to request brochures for patients.

Contact: Andrea Paal, M.S. or Audrey Bibb, M.S. at 1-888-493-6682 (toll free) or Coordinator@SimonsVIPConnect.com

The Risk Factor Analysis of Hereditary Breast and Ovarian Cancer

This study at Women's College Research Institute in Toronto, Canada is the largest long-term study of women who carry a mutation in *BRCA1* or *BRCA2*. Upwards of 11,000 participants have been enrolled from across Canada, the United States, Europe, and Asia. Its purpose is to better understand the prevention and treatment of hereditary breast and ovarian cancers. Participation entails completion of a baseline questionnaire and follow-up research questionnaires every two years. For more information please visit the website: www.womensresearch.ca/carrierstudy. If your center might be interested in collaborating in this study please, contact Marcia Llacuachaqui.

Contact: Marcia Llacuachaqui at marcia.llacuachaqui@wchospital.ca

The Risk Factor Analysis for Familial Breast Cancer

This study at Women's College Research Institute in Toronto, Canada is the only long-term study of women with a strong family history of breast cancer that do not carry a mutation in *BRCA1* or *BRCA2*. Its purpose is to better understand the interactions between various factors that may be associated with breast cancer development in women from high-risk families. Participants must provide a blood, urine, and toenail sample. For more information please visit the website: www.womensresearch.ca/noncarrierstudy. If your center might be interested in collaborating in this study, please contact Dr. Joanne Kotsopoulos.

Contact: Dr. Joanne Kotsopoulos at joanne.kotsopoulos@wchospital.ca

Genetic Basis of Bilateral Breast Cancers

This study at Women's College Research Institute in Toronto, Canada aims to identify genetic and non-genetic factors that may cause bilateral breast cancer. Participation is voluntary and is open to women who have been diagnosed with bilateral breast cancer, with the first diagnosis under the age of 65 years. Participants must provide a blood or saliva sample. Women who do not live close to the research center can still participate in this study. For more information please visit the website: www.womensresearch.ca/bilateralbreastcancer

Oxidative Stress Markers in Inherited Homocystinuria and the Impact of Taurine

The Inherited Metabolic Diseases Clinic at The Children's Hospital/University of Colorado Denver is recruiting individuals with homocystinuria due to cystathionine β-synthase deficiency to participate in a study that examines biochemical markers of oxidative stress and inflammation, platelet function and endothelial function. Additionally, the study examines the response to short-term taurine supplementation. This study has been approved by the Colorado Multiple Institution Review Board. Please contact us for more information if interested.

Contact: Cindy Freehauf, RN, CGC, at 303-724-2342

Clinical Trial of Coenzyme Q10 and Lisinopril in Muscular Dystrophies

The study is enrolling individuals age 8 and older with a diagnosis of Duchenne muscular dystrophy, Becker muscular dystrophy, or autosomal recessive limb-girdle muscular dystrophy (specifically: LGMD 2C-2F and 2I) who have no clinical cardiac symptoms. The study's enrollment goal is 120 participants. Participants will be randomized to one of four study arms: CoQ10 alone, Lisinopril alone, CoQ10 and Lisinopril or no study medication. Randomization will be stratified by ambulatory status and corticosteroid use. The study will last 24 months with visits at Months 1, 2, 3, 6, 9, 12, 18 and 24.

Contact: Lauren Hache, MS, CGC at 412-383-7207 or lpm6@pitt.edu

A Randomized Placebo-Controlled Study of Lovastatin™ in Children with Neurofibromatosis Type 1

The University of Alabama at Birmingham is conducting a study to determine whether Lovastatin™ can improve cognitive function in children with Nuerofibromatosis Type 1 (NF1). Participants will be randomized into one of two groups: Lovastatin™ or a placebo control (inactive substance). Cognitive function of the two groups will be compared to determine if treatment with Lovastatin™ can improve cognitive function. The study will last for 24 weeks and a screening assessment will be performed to determine a child's eligibility. For more information please visit the website, www.nfconsortium.org.

Contact: Study Coordinator, Ravin Winfrey, MS at 205-996-2916 or rwinfrey@uab.edu

Please send "Research Network" submissions to Emily Place at emily.place@gmail.com