Perspectives in Genetic Counseling
Volume 31, Number 4
Winter 2009

Deepti Babu, MS, CGC
Editor
deepti.babu@albertahealthservices.ca

Kirsty McWalter, MS, CGC
Associate Editor
kirsty@hawaiigenetics.org

President's Beat

Strategically Advancing the Mission and Vision of the NSGC

I am writing this column after returning from this year's Annual Educational Conference (AEC) in Atlanta. For those of you who were not able to attend, you unfortunately missed a great meeting. However, you can still take advantage of the conference presentations by ordering the online recordings. I want to say a special thank you to **Sarah Jane Noblin** and **Shannan DeLany Dixon**, whose tremendous efforts as AEC Chair and Vice-Chair really helped make this a great meeting. For me, it is the networking opportunities and interesting discussions with new colleagues that really make our annual meeting an outstanding professional opportunity that I would not want to miss.

The year may be coming to a close, but our work is just beginning. It has been an honor representing our society in 2009. We have had many great accomplishments as we continue to advance the mission and vision of our organization. It started earlier in the year with our new Strategic Plan for 2010-2012. It continued throughout the year as I represented the NSGC at many professional meetings targeting key strategic initiatives. Review the President's Blog for a more detailed account of the NSGC liaison activities this year. As an organization, we need to stay focused and committed to the activities that will help us accomplish the goals laid out in our Strategic Plan, and this has been a major focus in all of our efforts.

One of our most important issues is to increase the access of genetic counseling services for patients. In order to accomplish this, we will be introducing federal legislation in 2010 that will recognize genetic counselors as approved providers under Medicare. Most third party payors will follow the lead of Medicare. Of course, we are not just relying on this approach and we are also working directly with several third party payors to recognize genetic counselors at the same time. We are also working with a number of our members to accumulate research to demonstrate the value of genetic counseling and genetic counselors. All of these efforts require a large grassroots effort that involves our entire membership. Please be alert for any communications that will be coming out in the next several months asking for your support and action, and remember that we cannot accomplish anything without an involved membership.

I am also extremely excited about the development of our new logo, website and branding plan, which will all be put into action during 2010. You can read more about our branding efforts in this issue's article by our 2010 President, **Liz Kearney**. I urge everyone who wants to know what is happening in the NSGC to continue following all of our communications, including weekly updates, the President's Blog and our twitter account at http://twitter.com/NSGC_Org. Finally, I want to say a very special thank you

to Meghan Carey, our Executive Director, who has provided tremendous support to me during my presidency and continues to support and help lead our organization.

Steven Keiles, MS, CGC
NSGC President 2009

Leaders Among Us

By Deepti Babu, MS, CGC and Kirsty McWalter, MS, CGC

Perspectives in Genetic Counseling continues the **new series** highlighting genetic counselors who are exemplary contributors to the field and the NSGC. We hope this series will facilitate communication, show our appreciation for the countless hours of volunteer work donated by our members, and also illustrate that true leadership can be found everywhere within the organization. If you feel that a genetic counselor you know (including yourself) deserves mention in this series, please contact Deepti at deepti.babu@albertahealthservices.ca or Kirsty at kirsty@hawaiigenetics.org. We look forward to learning more about the different contributions made by genetic counselors throughout the NSGC!

We are pleased to announce the leader chosen for the Winter 2009 issue of *PGC*: **Dawn Jacob Laney, MS, CGC, CCRC**. Dawn was nominated by one of her work colleagues and she answers our questions, below.

1. Please list your current position and include a short description of your role(s).

   I am privileged to be the Program Leader at Emory University's Lysosomal Storage Disease Center in Atlanta, Georgia. The Center provides diagnostic, evaluation, management and treatment services for individuals with lysosomal storage diseases. In my current role, I manage a genetic infusion center, coordinate several clinical research trials, and am the clinical care coordinator/genetic counselor for Fabry disease patients. In addition, I supervise a fabulous and experienced team of four genetic counselors, one infusion nurse, and one administrative assistant.

2. What was your first volunteer activity with the NSGC? How and why did you get involved?

   My very first volunteer activity with the NSGC was coordinating an Educational Breakout Session (EBS) at the Annual Education Conference (AEC) in 2000. Previously, I had attended the AEC but I started taking a more active volunteer role when a colleague/friend asked if I would be interested in volunteering. Generally, I have difficulty saying no to any request from a friend, but in this case I said yes because I was also interested in learning more about the "behind the scenes" planning of the AEC.

3. What is your current involvement/role with the NSGC?

   I was a local member of the Planning Committee for the 2009 AEC. In addition, I coordinated and moderated a Pre-Conference Symposium on lysosomal storage diseases.

4. What has been your best role or experience with the NSGC?

   I have had some very interesting and exciting roles with the NSGC, and actually have two favorites. The first was serving as a Co-Chair for the Metabolism and Lysosomal Storage Disease Special Interest Group (SIG). This was a great opportunity to meet genetic counselors in similar positions across the NSGC membership and work on some wonderful projects (like the Metabolic series in PGC*) and several great EBS opportunities. My other favorite role was co-planning the NSGC Region III Meetings in 2006 and 2007. It was a great chance to learn more about meeting planning, and very rewarding to have it all come together in a well attended educational event.*

5. How can volunteer experiences with the NSGC be improved? How can volunteer involvement be encouraged in general within the NSGC, and for traditional leadership roles?

   I feel that a brief "job description" for each volunteer option would be incredibly helpful. Sometimes you volunteer for something, but you may not necessarily have a good idea of what is expected in that role. For example what are the various responsibilities and expected time commitment/amount of participation? I think if members understand exactly what they are getting into at the outset, the NSGC will get more first-time volunteers and keep them happy and participating. I also think showing applicability to daily life would foster more of a sense of community for "specialists" or "niche" genetic counselors and encourage them to volunteer. Some folks I know with great leadership potential feel that they are more of researchers or general clinicians than genetic counselors.

6. How has your NSGC leadership experience enhanced your career?

My NSGC leadership experience has given me experience in meeting organization, and connected me with a great network of genetic counselors who serve as cross-country colleagues/resources.

The University of Texas Genetic Counseling Program Celebrates Its Twentieth Anniversary

By Sarah Jane Noblin, MS, CGC & Claire N. Singletary, MS, CGC

October 3, 2009 marked an important date in the University of Texas Genetic Counseling Program's rich history: we celebrated the twentieth entering class with the 20th Anniversary Celebration at the Hotel Icon in Houston. We imagine that Founding Director, **Dr. Jacqueline T. Hecht**, had no idea the impact the program would have on the lives of so many when the program started in 1989 with a single student. In twenty years, the program has certainly grown and evolved. We now admit six students each year and have graduated 74 genetic counselors thus far.

To recognize the accomplishments of the University of Texas Genetic Counseling Program (UT GCP), a celebratory gala and silent auction were planned by **Marianna Raia** (Chair of the 20th Anniversary Celebration Planning Committee) **Jennifer Hoskovec** (Vice-Chair of the 20th Anniversary Celebration Planning Committee), and **Claire Singletary** (Program Director of the UT GCP), along with the entire Planning Committee of UT GCP supervisors: **Amanda Brandt, Rebecca Carter, Jennifer Czerwinski, Andrea Harbison, Michelle Jackson, Sarah Jane Noblin, Kaylene Ready, Sarah Seth, Blair Stevens, Cathy Sullivan,** and **Kate Wilson**. Enough cannot be said about the tireless efforts of Marianna and Jen as they oversaw all of the details of this remarkable evening and actualized a celebratory event that will not soon be forgotten by those in attendance. A huge thank you must also be said to Dr. Giuseppe Colasurdo, Chairman of the Department of Pediatrics and the Dean of the UT Medical School, for supporting the UT GCP and the Celebration and to the UT Graduate School of Biomedical Science (GSBS), Dr. George Stancel, Dean of the Graduate School, and his fantastic administrative staff for their continuous support of the UT GCP.

University of Texas Genetic Counseling Program students, faculty, and alumni at the 20th Anniversary Celebration

The idea to host a gala in honor of the twentieth anniversary started small: How can we create another scholarship opportunity for our genetic counseling students? As we all know, there is very little financial support in the form of scholarships to help support Master's-level genetic counseling students. The UT GSBS supports its PhD students with stipends. In recognition that stipends are not awarded to M.S. students, the UT GSBS has generously allocated several scholarships for UT GCP students. Recipients of any competitive academic scholarship in the amount of $1,000 or more through the University of Texas are also eligible for in-state tuition, regardless of their residency status. Therefore, another scholarship goes a long way to make the dream of becoming a genetic counselor without a substantial financial burden a reality for another student. When the resources to host a celebration for our twentieth anniversary were made available, the program leadership saw an opportunity to reach out to those touched by the UT GCP and create a scholarship fund. What the leadership did not share until the night of the event was the plan to honor founding director, Jacqui Hecht.

For those of you not familiar with Jacqui, let us provide you with a brief introduction. She is a woman of great character who is intelligent, loyal, and exacting. She sets high standards and expects her students to rise to them. As she has told many a genetic counseling student, "there is no crying in genetic counseling." Jacqui's passion for the field is evident. After completing her undergraduate degree at New York University, she spent some time abroad until she returned to the states and settled in Colorado. She was persistent in her discussions with the University of Colorado geneticists about the importance of the new field of genetic counseling. This persistence paid off when she became a part of the first genetic counseling class at the Colorado program.

When her family obligations brought her to Houston, she built a circle of support for the establishment of a genetic counseling program in Houston. After multiple discussions with the Board of Regents, Jacqui again persevered and **Deborah Durand** became the first UT GCP student in 1989. In her clinical life, Jacqui developed a passion for skeletal dysplasias. This led to the pursuit of a PhD and certification in Medical Genetics in addition to Genetic Counseling. In 1995, she discovered the *COMP* gene for pseudoachondroplasia. As her focus turned to complexly inherited conditions such as cleft lip and clubfoot, her stronghold in research was cemented. In 2006, Jacqui became the Vice Chair for Research for the Department of Pediatrics at the University of Texas Medical School at Houston. The demands of the position required that she shift her focus away from directing the UT GCP. Claire Singletary came on board as Program Director and Jacqui retained the title of Founding Director.

Dr. Jacqui Hecht and her husband, Dr. Wayne Dorris,

at the 20th Anniversary Celebration

Another of Jacqui's qualities that allowed the UT GCP to thrive is the ability to surround herself with quality individuals who are as passionate about genetic counseling as she. Throughout the years, there have been innumerable supervisors who have contributed to the education of UT GCP students. Several have dedicated tremendous parts of their careers to the program, including medical director Dr. Hope Northrup, genetics nurse Barbara Dominguez, genetic counselor **Patti Furman**, former program Co-Director **Catherine Wicklund**, and former Assistant Program Director **Aimee Tucker Williams**. The gala provided an opportunity for all of these key individuals to reunite and celebrate the program they love.

The evening commenced with a silent auction and raffle. The 20th Anniversary Celebration Committee had scoured Houston for donations, particularly difficult during this economic time, and had collected a wide assortment of items for bidding. All proceeds of the silent auction, ticket and table sales went to the establishment of an endowed scholarship. During dinner, several key presentations were made. The annual Catherine L. Wicklund Outstanding Alumni Award was presented. Established in 2005 and named after the inaugural recipient, Catherine Wicklund (class of 1993), the award honors a UT alumni who has given back to the profession and embodies what it means to be an excellent genetic counselor. Sarah Jane Noblin (class of 1999) was the second recipient in 2006, followed by **Michelle Strecker** (class of 1999) in 2007, and Jennifer Hoskovec (class of 2003) in 2008. This year, class of 2006 graduate and 20th Anniversary Celebration Chair Marianna Raia was honored with the award. She was lauded as "a highly observant, dependable, and outstanding counselor, invaluable, gracious, a wonderful colleague, an exceptional supervisor, and a champion of the UT GCP" by those that nominated her. It was a wonderful way to cap off a magical evening for Marianna.

One thing not often seen from Jacqui Hecht is a display of emotion. However, when Claire Singletary began summarizing Jacqui's career and the impact that she has had on the Program, the surprise on her face was evident. She was truly touched by the announcement that the Genetic Counseling Scholarship Excellence Fund created by the proceeds of the 20th Anniversary Celebration would be named the Jacqueline T. Hecht Founding Director Scholarship to honor her everlasting contributions to genetic counseling in the state of Texas. The UT GCP owes a heartfelt thank you to all of the alumni, current and past faculty, students, friends and family that supported the 20th Anniversary Celebration with both their presence and generous donations.

The 20th Anniversary Celebration was a success in many ways. We created an endowed scholarship. We honored our Founding Director. We brought together our family tree. We celebrated our beloved field. We learned how to promote our program in the community and acquired new outreach skills. We hope to do all of these things again….for our fiftieth anniversary!

NSGC Branding: What's In It For You?

By Liz Kearney, MS, MBA, CGC
2009 NSGC President-Elect

Branding the NSGC

This past summer, **Steve Keiles**, 2009 NSGC President, and I sat in a conference room in Speaker of the House Nancy Pelosi's office in the Capitol Building. We were prepared to speak to a member of the Speaker's staff about the important role genetic counselors play in the genomic era. If we were successful in piquing the Staffer's interest during our ten-minute discussion, she would likely return to her desk and search for "NSGC" or "genetic counselors" on the Internet to enhance her understanding of how significant an influence genetic counselors have over the future of genomic medicine. After reviewing the current NSGC website, the Staffer would probably move on to her next task, because the NSGC website does not clearly communicate the NSGC's critical messages.

Fortunately, the NSGC will be kicking off a branding campaign in 2010. As a guest blogger on the NSGC President's Blog at http://nsgcpresident.blogspot.com, I described what branding is and why it is important for an association like the NSGC. A strong brand provides a clear definition in the customer's mind of the benefits of using that particular product or service when compared to other available options. Branding the NSGC will bring increased visibility and credibility to our profession and will provide other benefits, outlined below.

An Example: Branding the American Medical Association

The NSGC is certainly not the first professional association to build a brand for itself. A few years ago, the American Medical Association (AMA) faced plummeting membership. Many physicians elected to join specialized associations that were more closely associated with their areas of practice because they did not see how the AMA related to their day-to-day patient care. To better demonstrate the value of AMA membership, the AMA hired an advertising agency to create a brand. With the agency's guidance, the AMA identified its target customer and the key benefits that the AMA can deliver better than any other association.

The agency then created the brand experience, which included key messaging, a new logo, a color palette, and graphical elements that were incorporated into all of the AMA's communications. However, a visit to www.ama-assn.org demonstrates that branding is much more than simply a new website. The AMA appears to have selected the practicing physician as its primary customer and communicates through consistent images and messages that the AMA will provide support to its members throughout their medical careers. The overall impact is to create an image in physicians' minds about what the AMA can deliver; an image of ongoing support that likely did not exist prior to the branding effort. For example, the AMA's brand promise was fulfilled in 2008 through successful lobbying efforts to prevent cuts in Medicare and in 2009 through its active voice in the debate about healthcare reform.

How Will Branding Impact NSGC Members?

The NSGC's mission is to advance the various roles of genetic counselors in health care by fostering education, research, and public policy to ensure the availability of quality genetic services. Building a strong brand for the NSGC is a critical step to achieving this mission and helping others understand who we are and what we do. Branding will bring tremendous benefits to us as members, including:

- Increased credibility and visibility for the genetic counseling profession
- Improved understanding of the value genetic counseling delivers
- Higher volume of referrals
- More appropriate referrals
- Attraction of dollars to the NSGC to improve and expand member services
- Stronger influence over public policy issues that affect the practice of genetic counseling.

Steps in the Branding of the NSGC

The NSGC Leadership will continue to communicate its branding plans to the membership. The next step in this exciting process is to develop the specific creative elements of the brand, such as the logo and color palette that we will use to establish a consistent presence in all of our communications. These elements are the first step in moving the brand forward and will set the stage for developing a new, redesigned website and other communication pieces in 2010. Members can obtain information about branding progress through NSGC Weekly Updates, www.nsgc.org, the NSGC President's Blog, and future issues of *Perspectives in Genetic Counseling*.

SIG Speak

Genetic Counselors in Research: Expanding Opportunities and How to Get Started

By Courtney Nichols, MSc, ScM, CGC, Research SIG Co-Chair; Kara L. Simpson, MS, and Heather Zierhut, MS, CGC, Research SIG Co-Chair

As is clear from the National Society of Genetic Counselor's (NSGC) Professional Status Survey (PSS) and from the number of articles published over the past decade, a substantial proportion of the genetic counseling community is involved in research. The 2008 PSS indicated that Research/Study Coordination was the second largest primary role for both clinical and non-clinical genetic counselors (1). In earlier PSSs in which respondents were allowed to choose more than one primary role, Research/Study Coordinator was reported as a primary role for 27-32% of respondents (2,3,4).

The opportunities are out there and genetic counselors want to conduct and be involved in research, so it's time to look at how someone can get started. This article will look at the unique set of skills that make genetic counselors particularly suitable to growing research needs. We'll also hear from genetic counselors who work in various research roles about the work they do and what skills they feel are important. Finally, these experienced research genetic counselors will provide tips on how a genetic counselor interested in research can get started.

The roles and types of research in which genetic counselors are involved has been suggested to be incredibly diverse, ranging from Study Coordinator to Principal Investigator, from natural history and epidemiology studies to ethical, legal, and social issues (ELSI), and behavioral research (5,6). In 2003, the Jane Engelberg Memorial Fellowship was granted to **Susan Hahn**, **Elizabeth Melvin Heise**, and **Emily Burkett** to study the roles of genetic counselors who report spending 50% or more of their time in a research capacity. Responses to a questionnaire revealed varied roles, but the majority of respondents acted as a study coordinator and/or provided genetic counseling to study participants. The most common

types of studies research genetic counselors reported involvement in included gene/mutation identification, psychosocial issues, clinical/medical epidemiology, natural history, and genetic epidemiology. This questionnaire revealed that research genetic counselors clearly had a diverse range of skills, many of which were not part of their genetic counseling training. However, there is a paucity of data available about the role of genetic counselors in research before and after this project was completed. The next phase of the research will aim to repeat this survey to determine what, if any, changes have occurred regarding the roles and the skills and knowledge required to succeed in genetic counseling research roles.

There is evidence to suggest increased interest in research by genetic counselors and an increase in research training. A slight trend toward more genetic counselors publishing peer-reviewed original research was seen from 2002-2008. In 2008, 21% of genetic counselors had published papers, up from 18% reporting this activity in 2002 (1,4). A possible contributing factor to this increase was the addition of research to every accredited genetic counseling program. In 2003, the American Board of Genetic Counseling (ABGC) began requiring a core research component in all accredited training programs. Survey results subsequent to the implementation of the research requirement report a statistically significant greater emphasis and degree of preparation for students within training programs as compared to practicing genetic counselors surveyed previously (5,7). Furthermore, results showed that recent graduates of genetic counseling programs were more likely to have taken a research design course, completed a thesis, and written a grant or research proposal than genetic counselors who had graduated more than five years prior to the study (5).

Even with the variety of research roles available and trends toward increasing involvement and greater training, it appears that some genetic counselors want more. In 2008, only 59% of PSS respondents said they were satisfied with research opportunities in their current positions (1). A 2003 survey of the NSGC membership reported that 69.4% indicated a plan to perform research in the future and that the ideal job for 83% of respondents involved spending 25% or more of their effort on research. This is a much higher percentage of genetic counselors than those who reported a primary role of research in the PSSs. This gap between the number of counselors who conduct research and the number who express an interest in research involvement may be related to reported barriers to conducting research, including a lack of time, opportunities, and knowledge on how to perform research, write grants, and write manuscripts (5).

The time may be ripe to overcome those barriers and find research opportunities. Research funding has increased significantly with the beginning of the Obama administration and the establishment of the American Recovery and Reinvestment Act (ARRA) (8). Funding for genetics and specific genetic conditions has been slowly increasing since 2007 and the total National Institutes of Health funding requests for the fiscal year 2010 is $30,838 million (an increase of $443 million), or 1.4 percent more than 2009 (9,10). More funding opportunities will undoubtedly increase the need for clinicians interested in conducting research. This is a great opportunity for genetic counselors to get involved in research of all types as they are trained to have the skills to conduct and help in research, particularly with regards to genetic conditions. Since genetic counselors are used to working in a quickly evolving field, they also have the ability to learn and adapt quickly to new and additional skills that may be needed.

Transferable Skills

Genetic counselors are trained to have a broad knowledge of genetics. In addition to the direct application of clinical counseling skills, such as collecting pedigrees and providing analysis/counseling in genetic studies, there are other skills that genetic counselors possess that can be used to perform research.

An NSGC Task Force report on the core professional skills of genetic counselors identified several important skills that can be applied to research roles (11). One of the key professional skills that genetic counselors possess is, "the ability to tailor, translate and communicate complex information in a simple, relevant way for a broad range of audiences" (11). Many patients may feel intimidated when presented with complex research protocols. A simple explanation of what will happen during a study, including risks versus benefits of participation, is essential to ensuring informed consent. Additionally, an ABGC practice-based competency required during genetic counselor training, promoting client decision making in an unbiased, non-coercive manner, is also valuable to informed consent discussions (12). Recognizing and responding to ethno-cultural issues and ethical dilemmas are also competencies that are valuable in both informed consent and research design. Another core skill that the NSGC Task Force report identified is the ability to dissect and analyze a complex problem (11). Genetic counselors are trained to deal with problems as they arise during a counseling session. Since research is fraught with obstacles during protocol development, research implementation and data analysis, our ability to identify, synthesize and organize pertinent information can be key in working through problems that arise in research processes (12). Furthermore, the interpersonal skills required for genetic counselors to work with both patients and a healthcare team (11) are also an asset in solving problems and working as part of a study team.

Genetic counselors see patient concerns firsthand and work to find solutions. We are well aware of how research findings can translate into changes in medical care and counseling. The following are examples of how some genetic counselors have put these genetic counseling core skills to work performing research and a discussion of additional skills they have found useful.

Research Study Coordination

Responsibilities as a study coordinator can include study design, recruitment, data collection and management, Institutional Review Board (IRB) submissions, providing genetic counseling and psychosocial support to study participants, collecting pedigrees and providing analysis, coordinating participant visits, and data analysis.

Meg Bradbury, MS, CGC, is a genetic counselor who works in both the clinical and research settings at Children's Hospital in Washington, DC. She has worked in this dual role for five years. Clinically, she works in a neuromuscular clinic and her research is often related to the neuromuscular field. As a research genetic counselor, she has a multitude of different responsibilities that include providing consultation to investigators on incorporating genetic analysis into their studies, serving on the IRB and Ethics Board, and acting as key personnel in writing grants for which the center has applied with various Primary Investigators (PIs) in the institution. She has also written funded studies about informed consent of exon skipping studies and bone health. In the studies where she is the PI, Meg manages study personnel and executes all aspects of the study development.

Meg feels that most of what she has learned as a genetic counselor has been transferable to her roles in research. She says, "I often utilize the fact finding and critical thinking skills learned in genetic counseling. I often find that the counseling psychosocial insight can be helpful when working with researchers as well as the interviewing techniques." She also draws on known genetic counseling resources to develop research projects or to expand upon existing studies. Patient autonomy is another thing Meg feels she has learned from genetic counseling that applies directly to research, and she says, "I continue to ensure that [patient autonomy] is respected in all research I am involved in."

In her role as PI on several studies, Meg felt that there were some skills, such as statistics, that she was not adequately prepared for by her genetic counseling background. She also feels that without a PhD, there is a limit to funding opportunities that are available to genetic counselors. As a result, she is planning on returning to school for a PhD in Public Health with a minor in Epidemiology, which would provide her with much of the fundamental background for research in public health genetics.

What do genetic counselors need to be successful in research coordination? Meg feels there are several important traits that contribute to success, including flexibility, perseverance, ingenuity, and organization. She states, "This job is constantly changing and you need to be willing to adapt to these variations. I often find myself working long hours to get the job or task at hand completed; it is often the case that you get out what you put into your career. You have to love what you do."

Genetic Counseling Professional Research

Major technological advancements in genetic testing and increased public awareness of genetics and disease have put the field of genetic counseling in the spotlight. The explosion of information has also intensified the need to understand the clinical and psychosocial implications to those at genetic risk.

Bonnie LeRoy, MS, CGC, the Associate Professor and Program Director at the University of Minnesota Genetic Counseling Program, has been researching genetic counseling since 1996. Bonnie's research focuses on the ethical and social challenges associated with the genetic counseling profession. In addition to the advising, administrative tasks, and research associated with a faculty position, Bonnie serves as Editor-In-Chief of the Journal of Genetic Counseling (JGC). Bonnie's job is always challenging and never boring. There is always something new to learn and more to do.

Bonnie sees research as a team sport. She advises, "Don't stay isolated in your own area." Given the limitations in our training regarding research methods (due mostly to the length of the Master's training programs), collaboration is vital to supplementing studies. There is a gold mine of data in the field of genetic counseling but it may require people collaborating to best assess the questions that need to be addressed. If someone is just starting research, most centers have a research center that can help provide guidance on conducting research and publishing. Finding someone that has performed research and successfully published data can be an especially helpful resource.

Bonnie feels that the skills to perform genetic counseling research are transferable. She points out that students naturally learn how to analyze research papers; they critically assess journal articles to learn how to design and control information in their own research projects. Writing and communicating skills are essential for genetic counselors and easily translate into grant applications and article publication. Genetic counselors that can communicate and justify why they are conducting research and demonstrate how the findings will be influential will be successful. Bonnie also notes, however, that additional skills may be needed to perform robust research studies. There are a myriad of methods and statistics to examine genetic counseling studies. Determining which research method is best may require additional training.

Before taking on the task of Editor-In-Chief of JGC, Bonnie thought she had to undertake vast research projects to make significant contributions. Now that she sees hundreds of research papers pass over her desk, her view has changed. Bonnie states, "Every publication chips away at the questions and improves our understanding. We all have something to contribute to the literature."

Social and Behavioral Research

Social and behavioral research is another area in which genetic counselors work. Research in this field involves the study of individuals, groups and societies and the psychological, social, and behavioral processes in which they engage in various situations. Genetic counselors may be involved in all aspects of this research, through developing research ideas and protocols to coordinating studies.

Christina Palmer, PhD, CGC, Associate Professor, UCLA and **Erin Baldwin, MS, CGC** are UCLA genetic counselors who have worked on the Deaf Genetics Project, an NIH-funded, collaborative project assessing the impact, utility and appropriateness of genetic testing for deafness within the deaf/hard of hearing population. Christina and Erin are the PI and Study Coordinator, respectively, on the project.

Christina and Erin point out that genetic counselors bring, "an understanding of the types of social issues that arise and behaviors clients engage in" to research, which can be valuable for developing studies. An understanding is developed through clinical interactions with clients and discussions with colleagues during training and practice. Christina and Erin maintain that cultural competencies and a strong ethical perspective are helpful skills and attributes. They also emphasize the need for researchers who interact with and enroll participants to, "present complex research protocols to study participants in a way that is understandable and accessible."

Other skills that Erin points out as important for Study Coordinators are familiarity with IRB regulations and submissions and data management. Christina notes that PIs need skills for developing protocols, writing grants, analyzing and interpreting data, and managing funding. Even genetic counselors who are not PIs on research projects may find themselves involved in these aspects of projects in some manner. For many genetic counselors, they may have to learn these skills outside their formal training.

So, how should a genetic counselor interested in conducting social and behavioral research get started? Christina and Erin have several suggestions: "Mentoring has proven to be one of the greatest teaching tools," they say, and suggest that those interested in research seek a network of support from seasoned researchers and collaborate with others who share similar interests. Networking can be accomplished at meetings and workshops or through involvement in NSGC Special Interest Groups (SIGs). They also suggest taking courses in Social/Behavioral studies and research practices, particularly if this was not an emphasis in one's training program. Additional training classes may be available through IRBs or academic institutions. Genetic counselors can also look for jobs that have some level of involvement in research where they can further develop skills. Finally, Christina and Erin suggest that once someone has identified an area of interest, they should delve into the literature: "A researcher will need to have the skills necessary to understand what information and protocols are important for their type of study and to identify what new information is needed within the field of interest."

How to Get Started: Advice from the Experts

The previously discussed barriers to involvement in research, including a lack of time, opportunity, and knowledge to complete various steps of research, can be difficult to overcome. For those who want to do research, however, the genetic counselors featured in this article have some tips for getting started:

- Find a question or topic that really interests you; it's important to love what you do
- Once you find a topic, delve into the literature
- Look for a job that includes a research role or time for research so you don't have to add it on top of full-time clinical duties

- Seek additional training or guidance from research centers or IRBs at your institution (e.g. additional courses in research design or statistics)
- Find a mentor who is a seasoned researcher
- Collaborate with others who share your research interests, and be open to integrating their ideas
- Be flexible and adaptable to the changing knowledge in the field
- Remember, even small research studies contribute to our knowledge and are important to the profession

The Research SIG

Due to the expanding role of genetic counselors, research interests have infiltrated the NSGC. Many individual members, SIGs, and organizational publications already serve as wonderful resources for members. There is great promise of a valuable role for the Research SIG in this expanding atmosphere.

In an informal survey of the NSGC membership listserv, members expressed interest in a Research SIG. The membership was most interested in a comprehensive list of research/research genetic counselors, grant opportunities and a multitude of ideas that included networking opportunities, education and resources on study coordination, conducting research, IRB issues, grant writing and publishing.

The goal of the Research SIG is to serve the needs of members performing research-related tasks and to promote the advancement of research in genetics and genetic counseling. We work hard not to duplicate, but to enhance and unify the already strong research presence within the organization. The Research SIG may be an avenue for those interested in, or accomplished in, research design, methods, and implementation.

References

1. Smith M, Freivogel ME and Parrot S. 2008 NSGC Professional Status Survey. 2009.
2. Parrott S and Del Vecchio M. 2006 NSGC Professional Status Survey 2006. Boston Information Solutions. 2007.
3. Parrott S and Clark C. 2004 NSGC Professional Status Survey. Boston Information Solutions. 2004.
4. Parrott S, Clark C and Shannon KM. 2002 NSGC Professional Status Survey 2002. Boston Information Solutions. 2002.
5. Clark HM, Gamm J, Huether CA, Buncher CR, Blough Pfau RI and Steinberg Warren N. Genetic Counselors and Research: Current Practices and Future Directions. *American Journal of Medical Genetics* Part C. 142C:276-283. 2006.
6. Research SIG Informal Survey. 2009.
7. Bedard AC, Huether CA, Shooner K, Buncher CR and Steinberg Warren N. Career Research Interests and Training of Genetic Counseling Students. *Journal of Genetic Counseling*. 16:645-653. 2007.
8. "NIH Announces American Recovery and Reinvestment Act Funding Opportunities," National Institutes of Health. 2009. http://www.nih.gov/news/health/mar2009/ncrr-11.htm
9. "Estimates of Funding for Various Research, Condition, and Disease Categories (RCDC). Research Portfolio Online Reporting Tool," National Institutes of Health. 2009. http://report.nih.gov/rcdc/categories/
10. "Summary of the President's Budget," National Institutes of Health, Office of Budget. http://officeofbudget.od.nih.gov/br.html
11. "Core Skills of Genetic Counselors," NSGC Task Force. 2009. http://www.nsgc.org/career/Skills.cfm

12. American Board of Genetic Counseling Practice-Based Competencies. August 2006. http://www.abgc.net/CMFiles/Practice_Based_Competencies_Aug_2006_10-29-0951KFH-10292008-6844.pdf

---

NSGC News

NSGC Leadership Awards

By the NSGC Award Committee Members: Adam Buchanan, MPH, MS, CGC; Renee Chard, MSc, CGC; Sarah Coombes, MS, CGC; Mary Freivogel, MS, CGC; Joy Larsen Haidle, MS, CGC; Sheetal Parmar, MS, CGC; Deborah Wham, MS, CGC

Natalie Weissberger Paul National Achievement Award

Maureen Smith, MS, CGC

To quote one of her nominators, "Maureen Smith has been at the cutting edge of advances in genetic counseling throughout her career." She was a founding member of the Online Teratogen Information Service (OTIS). She was one of the first cancer genetic counselors and the chair of the very first cancer genetics short course. She was one of the first genetic counselors to promote having high quality genetic information on the internet in her position as the clinical director of Gene Sage. In her current position, she is again on the cutting edge of genetic counseling practice as Clinical Director of a genomic medicine initiative called the NUgene Project. Maureen's CV boasts a long list of publications and invited presentations, and she has been awarded numerous research grants and contracts to acknowledge her tireless efforts.

As is evident from her work experience, Maureen has been pivotal in expanding roles for genetic counselors. Not only has Maureen personally embraced new opportunities throughout her career, but along the way she has mentored students, colleagues, and other healthcare professionals. She has a strong interest in education and has served as an American Board of Genetic Counseling (ABGC) site visitor

leader since 2003. Maureen holds numerous faculty appointments at Northwestern University, where she developed a course that teaches genetic counseling students about non-clinical skills, managing their workplace, and alternative roles for genetic counselors.

Maureen seems to have endless energy and, like the "Energizer Bunny," she keeps going, and going, and going. The number of hours she has spent working on activities for the National Society of Genetic Counselors (NSGC) throughout the past two decades is amazing. She began to emerge as an NSGC leader in 1990 when she worked as part of the Speakers Bureau project and the Annual Education Conference (AEC) Planning Committee. Maureen has since dedicated time to various educational programs, the Education Committee, a Presidential rotation, a Web Editor position, and various other Committees and Task Forces. She was a founding member of the "Familal Cancer Risk Counseling Alliance", which eventually became the Cancer Special Interest Group (SIG), and has also been involved in both the Industry SIG and Research SIG. Most recently, Maureen played a key role in the Membership Committee and tirelessly worked to adapt the Professional Status Survey to the changing paradigm of genetic counseling. Her valuable experience, endless energy, and unique talents have enabled her colleagues to learn so much from Maureen and many feel lucky to call her a mentor and friend.

Maureen has been described by others as strategic, articulate, intelligent, and focused. Maureen's contributions to the NSGC and the genetic counseling profession are too many to count and her efforts are extremely deserving of this most prestigious NSGC award. Congratulations to **Maureen Smith**, winner of the NSGC's 2009 Natalie Weissberger Paul National Achievement Award.

Strategic Leader Award

Kelly Ormond, MS, CGC

There are few who work as hard at advancing the profession of genetic counseling as Kelly Ormond. Her dedication to the NSGC has not only increased the visibility of genetic counselors within the health care arena, but it has also increased our value.

Kelly helped the NSGC spearhead the identification of a new management company during a difficult and emotional era of NSGC. Throughout it all, she maintained the NSGC's clear messages to the genetics community at-large, policy makers, and the public. She provided direction and consistent communication not only to the Directors on the Board but also to the membership at-large, which enabled the NSGC to stay on course and continue to tackle the goals and objectives of our strategic plan. Her broad vision helped lead efforts to assess and change the NSGC's governance structure.

Kelly graduated from the Graduate Program in Genetic Counseling at Northwestern University in 1994 and hit the ground running. Very early on, she worked on numerous AEC Planning Committees, including the Workshop Committee and Abstract Restructuring Committee, eventually co-chairing the Savannah AEC in 2000. She has served on the NSGC Board of Directors as Education Committee Chair from 2001-2003 and President from 2004-2005. All the while, Kelly expertly maintained her responsibilities as Program Director of the Northwestern University Graduate Program in Genetic Counseling. More recently, Kelly became the inaugural Program Director of a new genetic counseling program at Stanford University. She has been a member of the NSGC Ethics subcommittee, the Finance Committee and the Billing and Reimbursement Task Force. She is also an active member of the Association of Genetic Counseling Program Directors, the American Society of Human Genetics, and the American College of Medical Geneics.

Kelly's passion for our profession is evident in all the activities in which she is involved. Her research addresses not only the ethical issues we as professionals face, but has opened the door to thought-provoking discussions regarding our profession's impact on client-decision making and the disability community. Kelly is a role model not only for her many students, but to several of her colleagues. Congratulations to **Kelly Ormond**, winner of the NSGC's 2009 Strategic Leader Award.

International Leader Award

Elizabeth Gettig, MS, CGC

It's probably safe to say that Elizabeth (Betsy) Gettig is a well known member of the NSGC. Since matriculating with a Master of Science degree in genetic counseling from Sarah Lawrence College in 1981, Betsy has forged a path for genetic counselors in the United States. She has served as President of the NSGC and was a founding member of both the ABGC and of the American College of Medical Genetics.

Betsy consistently strives to move the NSGC and the genetic counseling profession forward. She tackled the World Wide Web by launching an online version of the NSGC's Board Review Course in Medical Genetics and Genetic Counseling, along with other interactive, web-based educational opportunities. Over the past twenty-eight years, Betsy has directed programs, taught courses, chaired committees, received awards, presented lectures, facilitated discussions, tutored students, mentored fledgling

counselors, represented our organization and our profession, supervised residents, edited manuscripts, reviewed grants, and volunteered her time, talent and considerable knowledge with enthusiasm and boundless energy in a multitude of ways. In 2008, Betsy received the Marjorie Guthrie Award, the most prestigious award presented by Huntington Disease (HD) Society of America for outstanding service to the HD community.

Now, she's gone international. Between July 2004 and May 2009, Betsy has made several trips to India to conduct workshops and present lectures on subjects including family history taking and evaluation, dysmorphology, risk assessment, psychosocial issues in genetic counseling, principles of research, and ethics, and she continues to supervise fellows in India. As part of two National Institutes of Health Fogarty awards, she has served as a mentor to trainees of these grants. In 2007, Betsy set foot on another continent to deliver a talk on the subject of genetic counseling for psychiatric illness in Melbourne, Australia. More recently, she served as a consultant to the Genetic Counselor Consortium of South Africa. Her hope is that her legacy will be the establishment of a genetic counseling training program in India.

In 1999, Betsy was honored for her outstanding contributions to the NSGC when she was awarded the Natalie Weissberger Paul National Achievement Award. Ten years later, it is time to recognize a genetic counselor whose vision knows no borders. Congratulations to **Betsy Gettig**, winner of the NSGC's 2009 International Leader Award.

New Leader Award

Sara Pirzadeh, MS, CGC

Sara Pirzadeh received her Bachelor of Science from Texas A&M University in 2003 and her Master of Science from the University of Minnesota in 2005. Even before she graduated from the University of Minnesota, she was very involved with the NSGC. As a student, she presented interview techniques at an AEC Educational Breakout Session (EBS). In the year she graduated, she was selected for an AEC platform presentation, became Co-Chair of the Legislative Subcommittee, and created the Legislative News Bulletin. The Legislative News Bulletin is a bi-monthly publication addressing current legislative issues and pertinent articles in the general media of concern to the membership. This bulletin was a significant contribution to the genetic counseling community as it keeps members informed and motivates others to get involved in legislative issues pertaining to genetic counselors.

In the four short years since her graduation, Sara has consistently demostrated her enthusiasm for, and dedication to, the genetic counseling profession. She has worked to expand the outreach services for her institution and was successful in this endeavor. She has already published at least three articles in peer-reviewed journals, given many presentations to professional and lay audiences, created tools for genetic counselors to use, and written multiple newsletter and magazine articles about genetic counseling. In 2008, Sara was awarded a grant from the Cancer SIG to develop quick tools for physicians to recognize syndromes and refer patients to genetic counseling.

Sara played a major role in educating health care providers and patients in response to the recent Public Awareness/Direct-to-Consumer campaign in Texas. She has become an active public speaker and author as a result of this experience. At the recent AEC in Atlanta, she shared her lessons and experiences in a workshop entitled: How to Create a Statewide Genetics Education Campaign.

Sara Pirzadeh has shown strong leadership skills and initative to keep the profession moving with the pace of healthcare. She has made a positive impact on those around her. In the words of a colleague, "Sara's dedication to the genetic counseling profession is impressive… She has been a mentor to others and active in so many areas of the genetic counseling community… She consistently makes significant contributions to NSGC, the genetic counseling profession, and the medical community, and she is clearly a very strong new leader in the organization." Congratulations to **Sara Pirzadeh**, winner of the NSGC's 2009 New Leader Award.

New Leader Award

Heather Zierhut, MS, CGC

Heather Zierhut received her Bachelor of Science in Cellular and Molecular Biology from the University of Wisconsin-La Crosse in 2004. She received her Master of Science in Genetic Counseling from the University of Minnesota in 2006 and is currently working on a Ph.D. in Molecular, Cellular, Developmental Biology and Genetics. With Heather, it is difficult to find the place to begin describing her contributions in such a short time frame since graduation. As a graduate student, Heather was interested in learning about the knowledge base of genetics and genetic counseling amongst legislators. She reached out to the legislators in Minnesota to assess their knowledge of genetics and became a resource to them. She has testified on The Hill in Minnesota and is active in public policy both on the state and national levels.

Heather has been quite active in the NSGC. She is the Co-Chair of the Research SIG and has been working to help this SIG contribute to the NSGC and add additional benefits for the SIG members. She created surveys to elucidate member interests and developed a directory of the members, interests, and areas of expertise to foster communication and networking. Using this information, she was able to develop a set of SIG goals, projects and a timeline to execute the agenda.

Heather is also an active member of the Public Policy Committee. She helped to create and implement the "Public Policy and Genetic Counseling" EBS in 2007, writes for the NSGC Public Policy blog, and educates the NSGC members and policy makers on imporant NSGC initiatives. She also spoke at the AEC as part of the "Job Hunt" EBS in 2007 regarding experiences applying and interviewing for genetic counseling positions. She served on the Planning Committee for the 2006 Region IV annual meeting and coordinated a social event titled "Genetic Counseling BINGO" to promote membership communication and networking

At the state level, Heather helped organize the efforts to create the Minnesota Genetic Counselor Association (MNGCA) and devoted countless hours to the development of the website, by-laws, and foundation of the new NSGC chapter. She currently serves as the Vice Chair of the MNGCA and Chair of the Public Policy Subcommittee. She has also served as a judge for the DNA Day essay contest from 2006-2009 and is actively involved in the University of Minnesota Genetic Counseling Program via running journal clubs, teaching classes, and mentoring students.

In the short time since graduation, Heather has worked tirelessly to promote the NSGC and the genetic counseling profession on both a state and national level. Congratulations to **Heather Zierhut**, winner of the NSGC's 2009 New Leader Award.

Outstanding Volunteer

Sarah Jane Noblin, MS, CGC

The Outstanding Volunteer Award is presented to those who have made exemplary contributions to the NSGC over an extended period of time. This includes volunteering on Committees and special projects on a national level and making the kind of contributions that go "above and beyond the expected duty" of the position. In a field full of individuals for whom "above and beyond the expected duty" is the norm, this is truly a special recognition.

Sarah Jane Noblin's current title reads like a list of several jobs, with multiple clinical and academic roles, including serving as a genetic counselor at LBJ General Hospital in Houston, Texas and as the Assistant Director of the genetic counseling program at the University of Texas Health Science Center in

Houston. Since joining the NSGC in 1997, Sarah has served the organization in many significant capacities. She has been on several AEC Planning Committees and was chair of the 2009 AEC planning committee. Other NSGC Committee membership has included the Genetic Services Committee, Professional Issues Committee, and Billing and Reimbursement Subcommittee. Sarah is also a Continuing Education Unit reviewer and is serving a term on the Ethics Advisory Board. She has also been active in the Prenatal SIG, serving as Co-Chair from 2003-2005.

Sarah's volunteering is not limited to the NSGC. One of her nominators stated that, at her community hospital, Sarah "co-chairs the Perinatal Compassionate Care Initiative, is a member of the Breastfeeding Advocacy Committee and serves on the Ethics Committee. In recent years, she has given educational addresses to nurses, midwives and other medical professionals, participated in annual area Health Family Wellness Days, facilitated pregnancy loss support groups and led teams for the March of Dimes". That is just a sample of the work she's done within the NSGC and her community. To top it off, her multiple nominators noted that Sarah is "a pleasure to work with," "very gracious to [other] volunteers," and "an outstanding mentor to newer NSGC members." Congratulations to **Sarah Jane Noblin**, winner of the NSGC's 2009 Outstanding Volunteer Award.

Outstanding Volunteer

Wendy Uhlmann, MS, CGC

Wendy Uhlmann's career in genetics began as a cytogenetics technologist at the University of Michigan, where she later received her Masters degree in Human Genetics in 1987. She first worked as a prenatal genetic counselor at Wayne State University. Since 1993, Wendy has been the genetic counselor/clinic coordinator of the Medical Genetics Clinic at the University of Michigan, where she also helped launch the Cancer Genetics Clinic. Since 2007, she has also served as study clinician for the REVEAL study. In 2008, Wendy was appointed Clinical Assistant Professor of Internal Medicine and Human Genetics.

Wendy is a member of the executive faculty for the University of Michigan genetic counseling training program and has served as an instructor and research mentor for countless lucky genetic counseling graduate students and medical students. She is the author of many peer-reviewed articles. Notably, she is one of three co-editors of the book A Guide to Genetic Counseling, the Second Edition of which was just published this past summer. Many of us know the First Edition simply as "the purple book".

Wendy has been, and is still, an outstanding role model as a volunteer. At the state level, Wendy has been a member of several work groups tackling issues such as informed consent for genetic testing, licensure, and genomics policy. At the national level, the NSGC has been the beneficiary of Wendy's tireless

efforts. Since joining the NSGC in 1987, she has served as Chair of our Professional Issues Committee, Co-Chair of a national meeting, served as secretary for the Jane Engelberg Memorial Fellowship, and has been an active member of several other NSGC committees. Wendy previously served on the NSGC Board of Directors and was our President from 1999-2000. 2000, as many of you may remember, was a busy year. Wendy was our society's representative for President Clinton's signing of the Executive Order on Genetic Nondiscrimination. She was at the White House for the ceremony marking the completion of the working draft of the human genome, and she testified three times to the Secretary's Advisory Committee on Genetic Testing on behalf of the NSGC. She served as the NSGC Liaison to the National Advisory Council for Human Genome Research for five years, making thirteen trips to Washington DC on our behalf! She also served on the Board of Directors of the Genetic Alliance for five years.

Wendy is outstanding in many roles: an educator, mentor, researcher, clinician, and leader. Indeed, it is no surprise that she received an NSGC Regional Leadership award in 1996. Congratulations to **Wendy Uhlmann**, winner of the NSGC's 2009 Outstanding Volunteer Award.

NSGC News

NSGC-Sponsored Internship Offers Exposure in a Unique Setting

By Mary Freivogel, MS, CGC, Membership Committee Chair and
Sheetal Parmar, MS, CGC, Membership Committee Vice Chair

In an effort to provide experiences in expanded roles for genetic counselors, the NSGC and Myriad Genetic Laboratories, Inc. partnered to offer three, seven-day student rotations during the summer of 2009. The rotations took place at Myriad's headquarters in Salt Lake City, Utah and offered in-depth experience in hereditary cancer and laboratory processes, including exposure to the multitude of ways genetic counselors contribute in a diagnostic laboratory setting.

The three students selected for this rotation were **Kelly East** (University of North Carolina Greensboro), **Amanda Knoth** (Stanford University), and **Sara Rhode** (University of South Carolina).

At the end of their rotation, each student was asked for their feedback about the internship experience. Below are excerpts from their comments:

Kelly East

"I learned that genetic counselors play a vital role in the laboratory/industry setting and can contribute their skills in a variety of ways – from providing medical support to patients and health care providers, to helping market and sell the product in the field, to working with insurance companies in an effort to increase reimbursement for genetic testing. The internship really helped diversify my view of the genetic counseling profession…." – Kelly East

Amanda Knoth

"During my time at Myriad Genetic Laboratories, I have had the opportunity to explore the different ways in which I can utilize my genetic counseling degree, training, and skill set. One of my goals in my future career is to serve the cancer patient population, and Myriad offers positions in numerous departments that directly benefit both the individual patients and the cancer community on a broader policy level." – Amanda Knoth

Sara Rhode

"As a genetic counseling intern, this experience has given me increased confidence to discuss the many intricacies of genetic testing with my patients." – Sara Rhode

This joint internship between the NSGC and Myriad Genetic Laboratories will be offered again during the summer of 2010. The NSGC is working to expand this program to include internship experiences at other organizations. Please watch the NSGC website, www.nsgc.org, and the NSGC listserv in early 2010 for additional details and application information.

ABGC Update

How Does The ABGC Score Certification Examinations?

By the ABGC Board of Directors

The ABGC has made several significant changes in the examination development and administration process, so it is natural that there are questions concerning how the examination is scored. We would like to take this opportunity to let you know exactly how we score every examination.

The ABGC uses a criterion-referenced methodology in scoring examinations. With this type of scoring methodology, there is no "curve," and candidates do not compete against one another. In constructing criterion-referenced examinations, the score that is most important is the passing score, that is, the minimum score needed to pass the exam. A great deal of time is spent ensuring that passing scores are derived fairly. The Angoff scoring method is used to set the passing score for the examinations, and a statistical process called equating is used to adjust for the slight variations in difficulty that can occur among multiple test forms (versions of the exam). These steps help ensure that all candidates are held to the same standard.

The examination development process involves multiple layers of review. The utmost care is taken to verify that examination items are clear, correct, and assess the proper content. When new items are introduced on an examination, an item analysis is performed as a final check of the validity of the items: After the examination administration is complete, a statistical analysis is performed on the examinees' responses to the items. The ABGC Certification Examination Committee (CEC) then reviews any items for which there are examinee comments as well as those that show unusual statistics, such as an item for which an unusually low percentage of examinees selected the correct answer. Based on this review, items that did not perform well are removed or otherwise adjusted so that candidates are not penalized. The entire examination development, administration, and scoring process are overseen by a measurement professional known as a psychometrician.

A general outline of the examination administration and scoring process is shown below.

The Exam Overview

- An examinee is randomly assigned to one of the two examination forms. Multiple examination forms are necessary to enhance security due to the examination window.
- The exam is administered through computer-based administration software.
- The candidate receives a provisional completion of examination report: a piece of paper from the proctor saying, "You have completed the exam, and you will be notified of your results of your test by [specified date]."
- The CEC reviews the statistical analysis of item responses and reviews items that had multiple candidate comments.

- Any scoring adjustments are applied, and final scores are determined.
- The final passing score for the exam is determined by means of the Angoff method (see below).
- Score reports are printed and mailed to candidates.

The Passing Score Study

The passing point for the examination is determined using a criterion-referenced process known as the Angoff method. This is the most common methodology for establishing passing scores for credentialing exams. Note that the following process is overseen by a psychometrician and is based on a combination of statistical analysis and the expert judgment of the CEC. The process begins after the examination forms are finalized and before the examination is administered.

- The CEC discusses characteristics of the examinee pool and what constitutes a competent genetic counselor, taking into account the tasks on the content outline. This discussion culminates in agreement on the meaning of the "minimally competent practitioner" – meaning a genetic counselor who *should* pass the exam, but just barely.
- The committee reviews each item on the examination, one at a time, and rates the item with a number that represents their answer to the question, "What percent of minimally competent examinees will get this item correct?" Widely divergent ratings are discussed to ensure that the CEC members' conceptions of the minimally competent practitioner remain consistent.
- The CEC members' ratings for each item are averaged. Then, those averages are averaged over the entire examination, yielding a grand mean rating. This grand mean, as a percentage, is then multiplied by the number of scored items on the test form, 170, to determine the number of correct responses needed for the initial estimated passing score on that exam form.
- The final passing score is determined on the basis of the initial estimated passing score, the variability of CEC members' ratings, and statistical information about the scores and potential passing scores. The final passing score is not determined until after the examination is administered. As a result of equating, the final passing score may differ on different forms of an examination because of small differences in the overall difficulty of each form.

Final determination of the passing score is a policy decision that is informed by the psychometric analysis. Careful adherence to this process yields a rationally determined score that can be used with confidence to make pass/fail decisions on the basis of candidates' scores. Note that with this method of scoring, it is theoretically possible for all examinees to pass the examination or for all examinees to fail the examination. Since candidates are compared to the criterion of competent practice, there is no minimum percentage of examinees that must fail the exam.

Each examination form contains 170 scored items and 30 pre-test items. Pre-test items do not contribute to candidates' scores, but information is collected on how well these items perform. This information will be used in the statistical analysis of future exams. In addition, by collecting data on scored and pre-test items, the ABGC may ultimately be able to offer instant scoring – meaning the candidate would receive a score report from the testing center proctor prior to leaving the center. It is not clear at this time when this may be available.

The examination development process and scoring determination for the ABGC certification examination are extremely detailed. This elaborate process is designed to ensure that *all* candidates are treated fairly and that all pass/fail decisions are valid. This detailed process provides distinction and value to genetic counselors in the field and ensures that passing candidates have the knowledge, skills, and abilities necessary for competent practice.

Student Forum

Expanding Our Borders: A Summer Rotation In Jordan And Many Lessons In Cultural Competency

By Jasmine Wong, MS, Sarah Lawrence College Class of 2009

Jasmine Wong (L) and a Bedouin woman (R)

Two students from Jordan, **Nadeen Jaradat** and **Rifaat Rawadesh**, were accepted into the Sarah Lawrence College (SLC) Genetic Counseling Program in 2007. In the first term they made a presentation on their country with the hope that two fellow students would assist in assessing the possibility of introducing a genetic service in Jordan. My classmate, **Monique Simard,** and I quickly jumped aboard since we were avid travelers and interested in participating in this research and clinical opportunity.

Such a great opportunity came with a great deal of work; from late winter to early summer, we found ourselves very busy planning and coordinating the project. First we formed a task group that met every month – we called ourselves Team Jordan. It was composed of **Caroline Lieber** (SLC Genetic Counseling Program Director), **Dr. Siobhan Dolan** (a public health specialist and instructor at SLC), Nadeen, Rifaat, Monique and me. Next, we began formulating our graduate theses which, after many revisions, we decided to combine. Monique focused on surveying the population of Jordan looking at attitudes, knowledge and resources regarding three common genetic diseases: beta-thalassemia, cystic fibrosis and Down syndrome. Complementing Monique's thesis, I surveyed Jordanian physicians along similar measures. We hoped that combining our projects would allow for a well-rounded needs assessment and a guideline for future genetic counselors in Jordan.

To begin, we researched the Jordanian medical system, culture, and law. Jordan is a developing country, strong in Muslim religion and Arabic family tradition, and borders the politically restless countries of Syria, Israel and Iraq. It is well known for its seventh new wonder of the world, Petra, an ancient city carved into sheer dusty pink rock, which has been featured in movies such as *Indiana Jones* and *Transformers*.

Jordan currently does not have medical geneticists or genetic counselors. Genetic counseling is provided by non-genetic physicians. **Dr. Saied Jaradat**, Director of Molecular Genetics at the King Abdullah Hospital in Irbid and our supervisor in Jordan, noted that Jordan may benefit from genetic services. High

consanguinity rates, traditional large families and anti-abortion laws naturally result in a higher prevalence of genetic disorders to the country. Also, the country is quickly developing its health care system. In 2004, Jordan started pre-marital screening for beta-thalassemia, and this year Northern Jordan will launch newborn screening. We aimed to study these considerations as well during our time in Jordan.

On July 27, 2008, I took a plane from New York City to Amman, while Monique arrived ten days later. The culture shock was immediate: stray camels roamed the streets and Bedouins and Gypsies sold their wares. By the end of the third week, I had begun to adjust to some of the cultural differences, such as sexual segregation and the practice of polygamy. I learned that I could shake a man's hand only if he put his out first and found that introducing myself as a Canadian studying at an American school was best received.

Once we felt more comfortable with Jordanian customs, social etiquette, and medical practices, we took part in genetic counseling sessions. Along the way we learned that most Jordanians believe that a greater power gives them trying situations, which they can endure. For some, genetic explanations for these trials were thought of as being discrepant with their beliefs. For others, it did not present a challenge to their faith. Because of this, we learned to ask each family if they were interested in hearing about the inheritance of their family's disease and respected their choices. We found that this approach was well received and appreciated.

Another adjustment to counseling style was necessary since abortion is illegal in Jordan and, therefore, not available and not discussed medically. Instead we focused on exploring the options that were available such as resources for a child with a disability and planning future pregnancies. We found it challenging, yet interesting, to alter our counseling styles to accommodate this and other different cultural, political and legal norms.

I found that modifying our counseling techniques was made easier by understanding Jordanian culture. To illustrate, traditionally it is the parents who choose their children's spouses. Therefore, if it were a public goal to lower the number of consanguineous marriages, parents would be the proper individuals to target for public education. In the clinic, most of our affected patients had autosomal recessive disorders and were from consanguineous marriages. When counseling the parents, I attempted to present information in a balanced manner by respecting and discussing the pros of consanguinity – financial stability, decreased risk of assault on women, and maintaining tradition – while reviewing how consanguinity can increase the risk of inherited disorders. The parents may choose to integrate the information about inherited disorders in their decision-making regarding a spouse for their child.

To finish, I would like to share the inspiring story of a Bedouin woman I met while in Jordan. Dr. Saied found a way for us to speak with a Bedouin community in the county of Mafraq. The little I knew about Bedouin people was gained from a few "National Geographic" images of weathered, bronzed women in colorful wool costumes. They are a nomadic group who live symbiotically with their camels and other animals. With simple expectations, we went to Mafraq.

The county was poor, with a lone intersection of stores and tents scattered every twenty miles. In a narrow corridor outside one of these intersections was a small women's community center. We met the founder of the center and marveled at her story. She married at the age of 14 (a typical age for Bedouin girls) and had seven children. In her twenties, she had a pivotal moment and went on to complete her Ph.D., while having four more children. But she still was not satisfied; after a man donated 100 Jordanian Dinars (about 140 U.S. dollars) to her, she started a women's center, which is now filled with exercise

machines, a beauty parlor, an arts and crafts center, computers and a kitchen. Workshops run weekly on topics such as blanket making, basket weaving, interviewing, internet surfing, nutrition, typing and hairstyling. It is available to many families in the surrounding area, some which she told us had a variety of rare genetic diseases because of a high consanguinity rate. She explained that the consanguinity rate was perpetuated by the family stigma of genetic disease, which inhibited members from marrying outside their family. Likewise, children with these diseases were hidden.

She shared that she is currently working with a social worker to aid these families. Together, we brainstormed ideas for other ways to help the families in this community and decided that we would provide workshops on consanguinity.

Overall, we never could have prepared for this experience through research or readings. Much was learned from firsthand experience in a culture so different from our own. I have always been an individual who seeks a good understanding of my own attitude, values and biases regarding society and culture. This experience shook my understanding of myself, and I am forever changed because of it; I imagine that this change has trickled into how I practice genetic counseling.

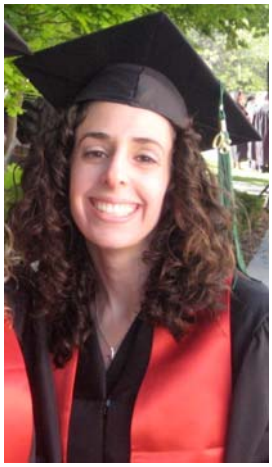
Generally, I have learned to be more patient, non-judgmental and empathic toward others, of my own culture and of other cultures. For example, before this experience, I generally felt uneasy when a man dominated a prenatal session because I imagined that this may be a relationship filled with control and not love. Of course, this scenario can certainly exist, but I no longer feel uncomfortable during a prenatal session when a husband/wife asks me to speak specifically to the husband. The Qur'an teaches that the woman has to do the most difficult and important task of all, to give birth, and a man must do all else for it to be more equal. Some men of Muslim faith or another faith/culture may believe that love and compassion is taking it upon himself to make a difficult decision for the family. It is not my own culture, but I see beauty in it and am happy to respect a couple's relationship, as long as it is legally appropriate. Lastly, I have learned that there is a want and a need for medical genetic services in countries around the world that are just awaiting a 'first step.'

I encourage other genetic counselors and genetic counseling students to seek out ways to become more internationally involved. Many developing countries lack genetic resources and Monique Simard, Caroline Lieber and I are currently asking for donations of old-edition textbooks and reference books. If you are interested in donating books, please contact me at Jasmine.Wong@albertahealthservices.ca. Thank you.

The New Graduate Life

Becoming A Genetic Counselor, Finding My Passion and A New Identity

By Sara Goldstein, MS, CGC, University of Utah, Class of 2008



Sara Goldstein at her graduation

Perspectives in Genetic Counseling *is happy to launch a* ***new series*** *about recent training program graduates. We hope this series will highlight the experiences from this critical and unique time in a new genetic counselor's career. If you feel that a recent graduate you know (including yourself) deserves mention in this series, please contact Deepti at deepti.babu@albertahealthservices.ca or Kirsty at kirsty@hawaiigenetics.org.*

There is a woman in my step class at my gym who is tall and lean, with short, straight blond hair that is delicately graying at the roots. Every time I see her, I think of a patient of mine, "C". They could be clones of one another, right down to the same style of thin-rimmed eyeglasses they wear and the way they smile, wide and bright. The difference between them is that C can't walk more than five minutes along a flat, paved road because she has end-stage congestive heart failure and a diagnosis of Fabry disease, whereas I watch her clone at the gym rhythmically bounce up and down on her step three times a week to a digitally enhanced aerobic version of Gloria Gaynor's "I will Survive." When I see the gym clone, I can only think of my patient. I know that all she wants is to be active again and live her life, but she is stuck in her house, stuck in the hospital armchair for her infusions, and stuck living a life she does not deserve. She has taught me the valuable lesson of perseverance and clinging on to the will to live, even in the worst of times.

Since my senior year in high school, I knew that I would be a genetic counselor. Even as I was applying to colleges I would tell people, "I am going to major in Biology so I can become a genetic counselor." After college, I spent three years living in Boston, working in a fruit fly lab. As my former colleagues will tell you, the most enjoyable part of that job for me was rewriting lyrics to classic Madonna songs to include accurate information about *Drosophila* mating. Suffice it to say, my job bored me. I longed for interaction with people and for the fulfillment of helping someone in crisis, rather than counting the

number of hairs on the “shoulders” of fruit flies. Not much time passed before I was ready to move on to graduate school, and there was no question that it would be to study genetic counseling.

I will never forget what it was like arriving in Salt Lake City for my interview at the University of Utah. Driving away from the airport, I could barely pay attention to the road. In front of me, behind me, and on every side there were jagged, snow-capped mountains spread against the purple horizon. Those majestic mountains have become a symbol of my greatest accomplishments and reflect a period of my life that truly changed me.

Most days I don’t recognize even an ounce of that girl who sat for hours in the fruit fly lab, filling vials with gooey pink fly food. No longer do I find my job boring. In fact, when it comes to anything related to genetics, I just can’t get enough. Undoubtedly, my affinity towards television shows such as “Mystery Diagnosis” and “House” is so strong because they are chock-full of genetic quandaries. I love genetics so much that a small part of me *enjoyed* studying for the Board exam. This does not mean I would want to study for it again, but my fellow study buddies and I definitely found the fun in the hours upon hours of review. When else could you create “Gene,” the white-board sketched, scruffy and buff mountain man, who was a fine model for displaying the body areas affected by various cancer syndromes?

My love for science has always been strong, but it is the counseling aspect of my job that has begun to shape me. It occurred to me that the way I identify myself is strongly based on the fact that I am a genetic counselor. Being a genetic counselor has changed my relationships with people. I have truly learned the value of listening closer, speaking softer and carrying around no big sticks. These are simple yet powerful skills. Being a genetic counselor has made me want to be an even stronger support for my friends and family. From my patients, I have learned some of the different ways that people express joy, pain, fear, anger and relief. I have used this to better understand people I have known for more than ten years, or less than ten minutes.

My training and career has not only changed my relationships with others, but it has affected my relationship with myself. We all carry around expectations for other people, but many of us have the highest of expectations for ourselves. I see it in my patients, and I see it in me. We help others struggle through difficult decisions or circumstances by giving them time, information, and unconditional support. I have learned to do the same for myself: give it time, let it move through you, and do what you need to do.

There will always be a warm spot in my heart for that rugged, beautiful and inexplicably spiritual land out west in Utah, and all the people there. It is the place where I took my first steps towards the person who I am today. My life is henceforth divided between pre- and post- genetic counseling. Pre-genetic counseling dates back to my years yawning through my days in the fruit fly lab, and post-genetic counseling is my present and future, wrapped up in inspiration from patients like C and a newfound connection with myself and others. I crossed over the bridge that divides my life, and it tastes so sweet on the other side.

AEC Update

Save The Date For The 2010 Annual Education Conference In Dallas, Texas

By Shannan DeLany Dixon, 2010 AEC Chair, and
Elizabeth Wood Denne, 2010 AEC Vice-Chair

Planning has already begun for the 29th NSGC Annual Education Conference (AEC) in Dallas, Texas on **October 14-17, 2010.**

The AEC will be held at the Hyatt Regency Dallas, which is about 25 minutes from the Dallas/Fort Worth International Airport (DFW). NSGC attendees will receive complimentary access at the hotel's 24-Hour StayFit Center, and will enjoy the hotel's Grand Bed™ and in-room iHome stereos with iPod docks. The Hyatt Regency Dallas, famous for its landmark tower in the Dallas skyline, is adjacent to the city's historical district and West End, featuring ample dining, shopping, and entertainment. The hotel is also within walking distance to The John F. Kennedy Memorial Plaza and a short taxi or train ride from the Dallas Arts District with museums, galleries, and grand performance venues.

Revised AEC format continues in 2010

The AEC debuted a new format for the 2009 meeting in Atlanta, Georgia. In response to the membership's desire to shorten the overall length of the AEC without cutting the number of CEU opportunities, the AEC will again begin with the Student and First-time Attendee Orientation followed by the Welcome Reception 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 with the conference concluding in mid-afternoon.

Call for Speakers opens December 2, 2009!

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 opens on December 2, 2009 and the deadline for submission is January 15, 2010.** Applicants will be notified of their acceptance in February. Watch for 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 on the electronic submission form. A Plenary session is typically one hour, while an EBS is typically two hours in length. Presentations may include more than one speaker and encompass two or more integrally related topics. A Pre-Conference Symposium is a minimum of four hours but can last as long as six hours in length. 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 2010 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 the proposal but do not plan to speak, list yourself as the "Primary Submitter/Session Coordinator." You will be considered the main contact for the proposal. You will also need to identify the speakers. *It is important that the AEC 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 form. Instructions and the submission form can be found at the following link: www.nsgc.org/conferences/speakers.cfm. Any questions can be directed to the AEC Chair at sdelany@som.umaryland.edu and Vice-Chair at ewdenne@jhmi.edu or 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 2010 AEC Subcommittee

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

AEC Chair
Shannan DeLany Dixon, sdelany@som.umaryland.edu

AEC Vice-Chair
Elizabeth Wood Denne, ewdenne@jhmi.edu

AEC Planning Subcommittee
Becky Althaus, beckyalthaus@texashealth.org
Carrie Atzinger, carrie.atzinger@cchmc.org
Erynn Gordon, egordon@coriell.org
Karen Heller, karenheller@swbell.net
Rebecca Kern, kern@kennedykrieger.org
Katrina Lowstuter, klowstut@myriad.com
Kirsty McWalter, kirsty@hawaiigenetics.org
Krista Redlinger-Grosse, kredlin1@fairview.org
Claire Singletary, Claire.n.singletary@uth.tmc.edu
Quinn Stein, steinq@sanfordhealth.org
Meredith Weaver, mweaver@acmg.net

Resources / Book Review

Reviewed by Nancie Petrucelli, MS, CGC

To Test or Not to Test: A Guide to Genetic Screening and Risk

Author: Doris Teichler Zallen
Publisher: Rutgers University Press, 2008
Pages: 198
Retail price: $18.95

Paper IBSN-978-0-8135-4378-9
Cloth ISBN-978-0-8135-4377-2

To Test or Not to Test provides a decision-making template to help consumers navigate the far reaching implications and considerations of genetic screening and offers additional resources where they can gain more information. This very readable book, which also provides a brief introduction to genetics, was based on over 150 interviews conducted with genetic specialists, as well as people who have faced decisions about genetic testing. The author, Doris Teichler Zallen, a professor at Virginia Tech, focuses her research on the ethical, social, and policy issues raised by advances in genetic science, especially genetic testing. She is also the author of Does It Run in the Family?: A Consumer's Guide to DNA Testing for Genetic Disorders.

Tests are a standard part of modern medicine, with genetic tests being the newest addition to the medical test toolbox. But, unlike other types of medical tests, genetic tests can offer glimpses into, or predictions about, the future and can impact other family members. We are, after all, defined by our genes. The range of available genetic tests has expanded over the last several decades, culminating in a whole new generation of genetic tests that look for variants associated with common disorders such as cancer or Alzheimer's disease. This suggests that in the years ahead most individuals will have an opportunity to consider genetic testing that can indicate susceptibility to a wide range of health problems.

Given the innumerable magazine, television, and Internet advertisements aggressively marketed to the public touting the importance of genetic testing, as well as the limited genetics knowledge of most medical providers, how does one decide to have a genetic test? In order to begin the decision-making journey about genetic testing, Doris Teichler Zallen arms readers with four questions that should be considered as one embarks on this very personal and often challenging voyage, and uses each chapter to thoroughly explore these questions.

1. Am I at higher risk for this disease?
2. Will the genetic test give me useful information?
3. Is the timing right for genetic testing?
4. Do the advantages of having the genetic information outweigh the disadvantages that testing can bring?

Doris Teichler Zallen teaches us about genetic screening and the complexities involved when deciding about whether to be tested by using breast/ovarian cancer (*BRCA1*/*BRCA2*), colon cancer (Lynch syndrome), late-onset Alzheimer's disease, and hereditary hemochromatosis as examples and focal points throughout the book.

The inclusion of nearly one hundred personal stories of those who have already faced genetic testing decisions (only half of whom had any form of genetic counseling prior to testing) provides a very poignant look into the very unique process that occurs when one is faced with such issues. Ultimately, the decision-making process involves a melting pot of the realities of an individual's life, personal values, attitudes toward risk, as well as his/her commitments, strengths, and fears.

It is clear that when it comes to genetic screening, 'one size does not fit all.' The genetic testing decision-making process is unique and there is no guarantee that the final decision will be free of complications. All one can do is make the best decision he or she can based on the available information and To Test or Not to Test could be an important resource in that process. Although written for the consumer, To Test or Not to Test also gives genetic counselors insight into patients' struggles when faced with genetic testing decisions and can help enable them to become even more thoughtful and compassionate health care professionals.

Media Watch

By Claire Noll, MS, CGC and Roxanne Maas, MS, CGC

(names of genetic counselors appear in bold)

July 2009 – *Journey* Volume 1, Issue 3 (http://www.renown.org/workfiles/pdf/cancer/Journey3.pdf)
"Knowledge is power – three generations find strength and answers through genetic testing"
This article from a publication of the Renown Institute for Cancer chronicled the experience of two sisters with *BRCA*2 mutations who developed breast cancer at young ages and the daughter of one of the sisters, who also tested positive. **Brandy Smolnik** counseled the sisters and provided information for the extended family. The daughter who tested positive is a reporter who posted her televised story online (http://www.youtube.com/watch?v=ufww__CC6o8). In the video, **Lynn Holt** provided support by explaining screening and treatment options that were not available for her mother and aunt because they did not know of their *BRCA* status before they developed cancer.

August 5, 2009 – Esquire.com
(www.esquire.com/features/abortion-doctor-warren-hern-0909)
"The Last Abortion Doctor"
This lengthy article described the clinical practice and thoughts of Dr. Warren Hern, one of the only remaining providers of late-term abortions since the murder of Dr. George Tiller. The article was written as a real-time interview with Dr. Hern. During the interview, Dr. Hern received a phone call from **Steve Keiles,** identified in the article as President of the NSGC. Dr. Hern asked Steve to issue a statement denouncing Dr. Tiller's murder.

August 6, 2009 – WDBJ7 Roanoke News (http://www.wdbj7.com/Global/story.asp?S=10861493)
"Are you at increased risk for cancer? Genetic testing is an option to find out"
A woman and her daughter, who were both found to be carriers of a *BRCA1* mutation, discussed their options with **Kara Bui**. The protections offered by GINA legislation were mentioned.

August 25, 2009 – *The Plain Dealer*
(http://www.cleveland.com/healthfit/index.ssf/2009/08/using_athome_genetic_testing_d.html)

"Using at-home genetic testing doesn't mean you can skip a visit to a specialist"
This column highlighted the difficulty in obtaining interpretation of direct-to-consumer genetic tests. **Judith Benkendorf** was quoted, "Just because a test may show that a woman doesn't carry the mutated *BRCA1* or *BRCA2* gene doesn't mean she has no risk for breast or ovarian cancer." Dr. Charis Eng, director of the Genomic Medicine Institute in Cleveland, stated that she has counseled "about a dozen" people who had difficulty understanding the results of their direct-to-consumer tests. Other medical providers interviewed for the article agreed that ordering physicians may not have the necessary background in genetics to effectively interpret the results.

August 31, 2009 – HealthNewsDigest.com
(http://www.healthnewsdigest.com/news/Family_Health_210/Genetic_Tests_More_Available_But_No_Less_Complex.shtml)
"Genetic tests more available, but no less complex"
and
September 1, 2009 – Media-Newswire.com
(http://media-newswire.com/release_1098011.html)
"Counseling is a crucial aspect of genetic testing"
These two articles described the experiences of a patient counseled by **Amy Sturm** for positive *BRCA* mutation test results. The first article covered the complexity of interpreting test results and the second article portrayed the patient's emotional distress upon receiving her results from the doctor's office via a cell phone call while driving home with her children. Both articles stressed the importance of pre- and post-test genetic counseling.

September 14, 2009 – Telegram.com
(http://www.telegram.com/article/20090914/BUSINESS01/909140331/1002/rss01&source=rss)
"On the job: Valerie Loik Ramey, Licensed genetic counselor, Dept. of Pediatrics, UMass Memorial Health Care"
This article profiled the career of a licensed genetic counselor, with **Valerie Ramey** explaining what types of information she obtains from her patients, the benefits of genetic counseling, and what she likes about the job.

September 25, 2009 – Star-Telegram.com
"Your family's medical history could affect your care, lifestyle"
This article discussed the importance of knowing more details about one's family history than people typically have at their fingertips. **Steve Keiles** presented eight tips for collecting a useful family history.

September 29, 2009 – The News-Gazette.com
(http://www.news-gazette.com/news/local/2009/09/29/genetic_testing_helps_sisters_in_fight_against_cancer)
"Genetic testing helps sisters in fight against cancer"
Three sisters, one of whom had breast cancer in her twenties, sought genetic counseling after their brother died of liver cancer, and all three were found to carry a *BRCA* mutation. Their genetic counselor, **Jennifer Burton**, explained that "genetic testing isn't for everyone and patients considering it should always consult their doctors first."

Research Network

By Suzanna Schott, MS, CGC

Ambivalence and Decision-Making in Prenatal Testing

The National Human Genome Research Institute is sponsoring a study that seeks to learn more about how women make decisions about prenatal testing, and how to best help them when they are uncertain. Prenatal patients who are ambivalent or uncertain about prenatal testing and have not yet had testing are eligible to participate in an anonymous online survey. See http://research.nhgri.nih.gov/SBRB/Prenatal for more information and to access the survey.

Contact: Barbara Biesecker at **301-496-3979**, or email barbarab@mail.nih.gov

DHREAMS: Diaphragmatic Hernia Research & Exploration, Advancing Molecular Science

The goal of the DHREAMS study is to develop a better understanding of the causes of congenital diaphragmatic hernia (CDH). This National Institutes of Health-funded research team is composed of health care providers and researchers across the country, coordinated at Columbia University Medical Center in New York City. DHREAMS is designed to identify specific genetic alterations that contribute to CDH through analysis of biological samples from fetuses and children with CDH and their family members. Visit the website http://www.cdhgenetics.com or http://www.clinicaltrials.gov posting NCT00950118 for more information.

Contact: Julia Wynn at **212-305-6987**, or email jw2500@columbia.edu

Familial Gastroschisis Project

The Utah Familial Gastroschisis Project is studying the potential genetic contribution to gastroschisis. This study involves collecting blood in families who have more than one member with gastroschisis. The affected family members do not need to be first-degree relatives. The University of Utah Clinical Research Program's Phenotyping Core has University Institutional Review Board approval to contact interested families, obtain consent, obtain medical records, and collect blood. Once data are collected, the best method to study the potential genetic contribution involved in gastroschisis will be determined.

Contact: Marcia Feldkamp, PhD, PA at **801-584-8490**, or email Marcia.feldkamp@hsc.utah.edu

Fragile X-associated Tremor/Ataxia Syndrome

Participants are sought for two studies of Fragile X-associated Tremor/Ataxia Syndrome (FXTAS) conducted by Dr. Maureen Leehey of the Department of Neurology at The University of Colorado Denver in collaboration with Dr. Randi Hagerman of the M.I.N.D. Institute at University of California Davis. The first study involves cognitive testing, review of family history, blood draw, neurological exam and Magnetic Resonance Imaging. The second study is a clinical drug trial evaluating the effect of a medication (memantine) on neurological symptoms of FXTAS. Carriers of an *FMR1* premutation who

are at least thirty years old may be able to participate. All testing and evaluations are free of charge and travel support may be available.

Contact: Wendi Legg at **303-724-8305**, or email wendi.legg@ucdenver.edu

Isolated Congenital Heart Defects Cell Repository

The National Institute for General Medical Science (NIGMS) supports a DNA biobank and cell repository at the Coriell Institute for Medical Research in Camden, New Jersey. This biobank supplies scientists worldwide with materials facilitating research on the diagnosis, treatment and prevention of disease. The NIGMS is collecting samples from individuals with *isolated* congenital heart defects. Our focus is atrial septal defects, ventricular septal defects, and pulmonic stenosis; however, others are welcomed. Interested donors may contact Coriell to receive a blood collection kit, patient/parental informed consent, submission form and a clinical information summary. Coriell covers the cost of shipment.

Contact: Tara Schmidlen at **856-757-4822**, or email tschmidl@coriell.org

MOMS - The Management of Myelomeningocele Study

MOMS is actively recruiting pregnant women for a randomized clinical trial designed to compare prenatal surgery versus standard postnatal surgery for spina bifida. Screening begins by telephone and a review of medical records. Interested candidates who qualify are assigned to one of three MOMS Centers for a comprehensive evaluation: The Children's Hospital of Philadelphia, the Vanderbilt University Medical Center in Nashville, or the University of California San Francisco. Eligible candidates are randomized to the prenatal surgery group or the postnatal surgery group. Participants must complete enrollment by 25 weeks gestation. See http://www.spinabifidamoms.com for more information.

Contact: Jessica Ratay toll-free at **866-275-6667**, or email moms@bsc.gwu.edu

Molecular and Family-based Studies of Gastrointestinal Neoplasia

Dr. Nicholas Davidson is seeking individuals with familial adenomatous polyposis (FAP) to participate in a study at Washington University School of Medicine's Siteman Cancer Center. The study aims to better understand inter- and intra-familial phenotypic variability in FAP. Participants will be asked to submit a sample of blood, provide medical and family history, and contact other family members about the study. In addition, participants are asked to allow samples of polyps, other abnormal lesions, and/or tumors (detected during standard clinical cancer screening) to be stored, analyzed, and used in research studies. Participants will be contacted on an annual basis for follow-up for at least five years.

Contact: Melanie Baxter at **314-454-5122**, or email baxterm@siteman.wustl.edu

Parental Response to Undiagnosed Pediatric Medical Conditions

The National Human Genome Research Institute is sponsoring a study that seeks to learn more about how parents of children with an undiagnosed medical condition think and feel about their child's condition. Eligible participants are men and women who are 18 years or older and have at least one child with a medical condition that has remained undiagnosed for more than two years. Participation involves one survey that takes about 45 minutes to complete. The survey can be completed online or a paper copy can be mailed. For additional information please see http://www.surveymonkey.com/StudyNoticeUncertainty.

Contact: Anne Madeo at **301-443-2635**, or email anne.madeo@nih.gov

The Williams Syndrome Patient and Clinical Research Registry

Do you have a patient with Williams syndrome? Are you interested in studying individuals with Williams syndrome? The Williams Syndrome Patient and Clinical Research (WSPCR) Registry was developed for the Williams Syndrome Association to help connect individuals with Williams syndrome (WS) and their families with researchers. The Registry also collects annual health and development information to learn how individuals with WS do over time. Patients can join the more than 400 families who are already members. Researchers with approved accounts can search the database, submit study proposals, and recruit study participants. Visit www.registry.williams-syndrome.org to learn more.

Contact: Jessica Waxler at **617-726-5318**, or email WSRegistry@partners.org

Young Women's Breast Cancer Research Program

Women diagnosed with invasive breast cancer before forty years of age are invited to participate in this study, which aims to identify genetic factors that distinguish breast cancer in young women. Women who have already had clinical genetic testing may also participate, regardless of the results. Participants are asked to sign a consent form, submit a sample of blood (obtained at a follow-up appointment), release a copy of cancer-related records, ask parents or sisters to participate, and be contacted to collect family history. Study kits are mailed directly to the participants. All phlebotomy and shipping charges are covered by the study. Please see http://www.ywbcp.wustl.edu for more information.

***Contact*:** Jennifer Ivanovich at **314-454-5076**, or email ivanovichj@siteman.wustl.edu

Please send Research Network items to sschott@cpdhealth.com