Perspectives in Genetic Counseling - Volume 30, Number 3

## President's Beat

### Envisioning the Future of Genetic Counseling Services

*By Angela Trepanier, MS, CGC, NSGC President, **atrepani@wayne.edu***

The sequencing of James Watson's DNA, the availability of direct-to-consumer genome scans, the FDA's updated product labeling of Coumadin to include a statement about the importance of genetic make-up, and the promise of the $1000 genome in the not-so-distant future, have brought us a real glimpse of what life in the personalized medicine era will be like. These events, along with advancing technology, have stimulated a tremendous amount of discussion regarding what healthcare professionals need to do to be prepared to integrate genomics into primary care practice. NSGC's involvement in meetings such as the NHGRI Population Carrier Screening conference, the American Academy of Family Practice Physicians' Subcommittee on Genomics, the Secretary's Advisory Committee on Health, Genetics and Society, the HHT Health Initiatives for the 21st Century, the HHS's Understanding the Interest and Needs of Consumers in the Use of Genomic-Based Health Information Services, and the Institute of Medicine's Genomics Roundtable have positioned us and our members to be active participants in these conversations.

What is clear from all of these discussions is that the time is now for us to determine what personalized medicine is going to mean for the future of genetic counselors and genetic counseling services. It is critical for NSGC, with the help of our members and external stakeholders, to determine the impact of personalized medicine on the services we deliver and how we deliver them. We are the ones best equipped to decide what we want our role to be and then to advocate for this role.

#### Questions to Address

As an example of how personalized medicine may impact our roles, consider the following issues related to genetic testing to assess risk for common chronic disease:

- If such testing becomes part of primary care, to whom will we provide services? Will we provide pre-test counseling (to everyone) or will that be the role of the primary care provider (nurse, physician assistant or physician)? If we only provide the post-test counseling, which clients will we see? Just those with the highest risk (possible single gene etiologies) or those at moderate risk as well?

- What will our role be? Will we still act primarily as consultants (one to two visits to assess risks, provide consent and explain results/management), or will we be involved with our clients throughout their lifespan, helping them understand and manage genetic information about their risk for childhood disease, adult onset disease and reproductive risks over time?

- Given the dynamic nature of genetic information and its implications, will we need to meet with clients regularly to update them about the clinical significance of their genetic test results or does this fall to the primary care healthcare professional? If primary care healthcare professionals will be providing more genetic services to patients, what will our role be in helping these healthcare providers stay up to date on genetic information?

- What, if any, will our role be in genetic testing and counseling for drug response (pharmacogenomics)?

- For personalized medicine to make a difference in a person's health, a person has to be willing to comply with recommendations for behavioral change. Will genetic counselors have any role in facilitating and reinforcing change outside of our current role (education and short-term client-centered counseling)?

- Where will we practice? Will a majority of those providing clinical care still practice in academic medical centers with other genetics professionals or will we increasingly be employed by primary practice groups in both large centers and rural areas?

- How will we accommodate the potential increase in demand for our services? Will the size of our profession hinder us from taking on what we see as the ideal role for genetic counselors in personalized medicine? If so, what can we do to increase the size of our workforce? And realizing that there are limits to how quickly the size of the workforce can change, how can we work with other healthcare

professionals to ensure that they are adequately educated to provide the level of genetic services needed for the timely integration of personalized medicine into healthcare?

**Developing Our Vision**

In anticipating the changes that personalized medicine will bring to genetic counseling practice, and recognizing that 2009 will mark our 30th anniversary, NSGC is developing our vision for the profession in the next 30 years. The process, which will be data driven, will begin by convening focus groups of external stakeholders and asking them questions such as those posed above. We will then use their answers and information from the discussions we have had with various groups regarding the future of genomics to develop additional questions for member focus groups to be held at the AEC. The focus group responses will then be used to develop a web-based, quantitative survey that will be distributed to the membership at large in the winter. Finally, the Board will analyze all the data collected and use it to develop the vision statement.

To ensure that the vision we develop is the shared vision of the membership, we need the input of each and every member! The upcoming advances in genetics and genomics are both exciting and challenging. This vision process is your opportunity to help NSGC direct our long-term efforts and ensure that we are appropriately planning for changes to come. I hope you will contribute to the vision process by taking part in a focus group and/or completing the survey. We need your collective experience, expertise and input to shape and prepare for the future of the genetic counseling profession.

## Professional Development

### ABGC Certification Examination Update: Implementing a One-Examination Format

*By the ABGC Board of Directors*

The genetic counseling profession has grown from a handful of training programs in the 1970s to 32 programs today, from a few hundred genetic counselors to over 2,000 and from a position of counselors creating jobs to employers actively recruiting counselors. Through these years, genetic counselors have continuously demonstrated their competencies and professionalism. Changes in the American Board of Genetic Counseling (ABGC) certification examination format reflect one more critical step in the evolution of our field and will lead to improvements in the ABGC certification examination program.

Over the past year, the ABGC Board of Directors has carefully considered different models for its certification examination program. In particular, the Board has evaluated the pros and cons of the two-examination format (general genetics and genetic counseling specialty) versus a one-examination format. After long and careful deliberation, the Board has decided to proceed with a one-examination format.

Beginning in 2009, the ABGC Certification Examination in Genetic Counseling will be:

- offered annually
- offered over a 30 day time period at numerous sites in North America
- constructed based on a Practice Analysis (completed in 2008).

These changes are being made in response to National credentialing standards, licensure requirements, issues raised by diplomats and training programs and the need for fiscal responsibility.

**Decision Rationale**

***Practice Analysis***

Recently, ABGC completed a practice analysis (PA), a systematic process of collecting, analyzing and documenting the information related to the nature of a professional group's practice. The PA was performed according to the guidelines of the National Organization for Competency Assurance (NOCA) and its accrediting arm, the National Commission for Certifying Agencies (NCCA), and conducted by a survey of practicing genetic counselors.

A PA provides the basis for identifying and validating elements of current practice, to define the necessary knowledge and skills associated with a profession. This information can then be used to develop a relevant and

valid certification examination supported by evidence-based data. A PA is typically conducted every three to five years so that the examination content reflects current practice. Basing a certification examination on a PA is considered best practice in the credentialing industry because the resulting examination is considered professionally sound.

The PA allows ABGC to develop examination questions that uphold the integrity of the examination while ensuring that appropriate knowledge of genetic principles and relevant genetic counseling skills are tested. If genetic counselors continue to sit for the general examination, which is wholly developed by the American Board of Medical Genetics (ABMG), it is not possible to ensure that the content of that examination is tied directly to genetic counseling practice. The results of the ABGC PA confirm that one test would be a psychometrically sound examination for the purpose of certifying genetic counselors. Because ABGC will be able to construct one certification examination based on a comprehensive content outline developed from the PA data, any redundancy that has historically existed between the independently developed general genetics examination and the genetic counseling specialty examination will be eliminated.

***Frequency***

Moving to a one-examination format will allow qualified genetic counseling professionals to sit for the ABGC certification examination on an annual basis. A more frequent examination administration has become increasingly important as states continue to implement licensure for genetic counselors. Some licensing bodies have expressed concern about a certification examination that is only offered every other year. Given that most licensing bills use passage of the ABGC certification examination as the basis for licensure, it is imperative that ABGC acknowledge the importance of administering the examination more frequently. The ABMG currently develops and administers the general genetics examination and has not expressed plans to offer that examination on an annual basis. By moving to a one-examination format, ABGC will have the flexibility to offer a number of new options:

- new graduates will no longer have to wait up to two years to attempt the examination
- examinees who do not pass initially will be able to reattempt the examination on a more frequent basis
- examinees successfully achieving ABGC certification will be able to serve as clinical supervisors for training programs in an expedited fashion.

***Testing Administration***

A one-examination format also allows ABGC to implement additional innovations now and in the future. Historically, the general genetics and genetic counseling specialty examinations each have been administered on a single day only. This created difficulties for candidates who could not take the examinations due to a conflict or unanticipated emergency. A one-examination format will allow ABGC to offer the examination during a 30 day window. This gives candidates more flexibility in scheduling. Should an unexpected event occur, a candidate would be able to reschedule his or her examination within that 30 day time period.

***Financial Considerations***

Moving to one examination helps ABGC contain the escalating costs that were occurring with the two-examination format. As a result, the fees for the 2009 ABGC certification examination program will not be increased. This would not have been possible with the two-examination format.

For more information, please visit the ABGC website at **www.abgc.net**.

## Career Watch

### Thinking Outside the Box and Inside Genome Screening

***By Elissa Levin, MS, CGC, Director, Genetic Counseling Program, Navigenics***


I remember being in graduate school as the Internet was coming into its own and imagining the vast potential that such a tool could afford the field of genetic counseling… global access to information, tools to help people learn about inherited diseases and decision-making strategies, and an outlet for sharing experiences. Above all, I saw what an amazing opportunity it would be for genetic counselors to reach more people. I even toyed with the idea of investigating such a service as my thesis project, but the consensus seemed to be, "You just can't do that. We need to see people face-to-face."

That was ten years ago, and a lot has happened since. I spent the first half of my career in academic genetic medicine – first as a cardiology research coordinator at Children's Hospital of Philadelphia, and then in general clinical genetics at UCSF. It wasn't until I came to Northern California that limited access to genetics services re-entered my consciousness. Funding cuts and financial pressures had forced a number of genetics clinics in the region to close. At UCSF people could wait over four months for an appointment. Some were traveling up to eight hours to see us. Others never came because of limited time off, childcare, travel costs and more. Many patients didn't understand why they had to come in to get tested for a familial mutation. I heard people recount stories like, "I've been living with this in my family for my entire life, I know more about it than you probably do, and I know that I want to get tested. Can't we just do this over the phone?" That's where we were stuck.

By coincidence or good fortune, a company called DNA Direct posted a job search for a genetic counselor to head their "virtual" genetics services. I thought long and hard about the job, met with them several times to better understand the company's approach and goals, and then I made the leap into online genetic services. Looking back, I have no regrets.

**Converging Trends**

At DNA Direct I worked to establish their clinical services through online genetic testing and counseling. I soon realized, however, that we were facing a new challenge. As genetics professionals, most of us are familiar with patients asking, "Can't I get a test that tells me what overall health conditions I am at risk for? I want to know what to expect." For years we sighed and said, "Unfortunately, we just aren't there yet." But genetic research started gaining momentum as new technologies enabled researchers to identify genetic markers that impart smaller effects than typical Mendelian factors.

Consumer awareness about genetics also intensified through a variety of multimedia channels. With the Internet, a forum emerged for offering alternative access to genetic testing services. Dozens of companies offering genetic testing directly to consumers have entered the field – some using validated tests by qualified labs and genetics professionals, others providing recreational or unproven genetic testing in the absence of professional services. In the last year this trend has fostered the emergence of several companies that are striving to bring the promise of genetic association studies for common health conditions directly to individuals.

As I watched the movement toward testing for common variants, I was interested to see how this new industry would take shape. I learned about one of the companies at the confluence of these trends: Navigenics. Since its conception, the company has strived to establish a responsible model for this new type of testing and help to set industry standards, including genetic counseling as a key component. When they offered me the opportunity to build their web-based genetic counseling program, I couldn't pass it by.

**Introduction to Navigenics**

Navigenics was founded by a clinical oncologist, Dr. David Agus, and a human geneticist, Dr. Dietrich Stephan. Their hope was to bring the promise of the ever-increasing number of genetic research discoveries to the individual, shifting the healthcare paradigm from our current model of diagnosis and treatment to one of early detection and prevention, thus improving health outcomes across the population.

Based in Redwood Shores, CA, the company began offering its first service, called the Navigenics Health Compass, in April of 2008. This testing service utilizes genome-wide microarray technology to identify common genetic variants associated with specific health conditions in order to generate personalized risk assessment for a variety of conditions.

**What Does Navigenics Do?**

Behind the scenes, the process begins with a staff of human geneticists and epidemiologists who review top tier medical and scientific journals to identify genetic association studies. They assess whether each study meets specific criteria, developed by Navigenics and its advisors, to ensure that the genetic associations are replicated, statistically significant and clinically relevant. Less than 5% of reviewed studies meet these standards.

The clinical staff of physicians and genetic counselors scrutinizes health conditions to be included in the service, looking at parameters such as: how common the condition is, if it is socially responsible and if it is preventable, treatable or could be lessened in impact by increased knowledge. A website is then developed to support pre-test decision-making, informed consent and post-test understanding of results for the designated condition. Results are framed as a comprehensive online report so individuals can go beyond their genotype and risk estimates to learning about the health condition, genetic versus environmental contributions, what medical research and guidelines say about mitigating risks and how to talk to their healthcare providers about this information.

Navigenics also has an advisory board of leading genetic counselors, scientists, clinicians and bioethicists to help inform strategies, goals and approaches, and support the company's responsible approach.

**Who is Appropriate for Testing?**

Unlike more traditional, indication-specific genetic testing, genetic screening for common variants can be considered by any adult who is able to provide informed consent. For adults who are adopted or have uncertain or incomplete family medical histories, this service can provide insight they previously would never have known. The information gained is most valuable for individuals motivated to learn about their potential genetic risks in order to make lifestyle and healthcare modifications. For individuals who know their family history or already live a healthy lifestyle, test results can help personalize relevant screening regimens, clarify early intervention strategies and provide insight into diagnostic dilemmas.

This type of testing does not replace traditional genetic assessment for individuals who have a personal or family history of a known disease. For example, a 40 year-old male with a family history of early-onset Alzheimer's disease would not benefit from this screening test with regard to AD. However, he could benefit from information gained about the other two-dozen common diseases included in the test, such as type 2 diabetes, colon cancer and heart disease (for a current list of conditions included, visit **www.navigenics.com**).

Ultimately, this type of service can be thought of as a clinical tool beyond family history, environmental risk factors and traditional medical screening. It is not a diagnostic test but rather a level of medical insight that we have not yet been able to provide patients clinically.

**How Does the Process Work?**

Once someone is ready to test, the process is quite simple:

1. An order is placed online – either by a physician or by the individual (in compliance with state and federal regulations).
2. A saliva sample is collected and sent to a CLIA-certified lab.
3. The lab uses microarray and Taqman technologies to screen almost two million points of variation throughout the genome and securely transfers the results to Navigenics.
4. The genotypes for the selected SNPs are extracted from the data file and combined into condition-specific risk estimates using odds ratios, prevalence data and established epidemiologic algorithms for combining risk factors.
5. When results are ready, the doctor or individual can login to review the personalized online report.

Prior to seeing any results, a person is required to opt-in to each condition for which results will be provided. This step ensures that if someone does not want to know their risk for a specific condition, that information will not be shown. Should the individual choose to view that information later, they can revisit the report and opt-in at any point.

Over time people also will receive ongoing updates as new scientific and clinical discoveries are made. Updates can include new markers for tested conditions, risk estimates for new health concerns (again, which someone needs to opt into) and additional information such as novel intervention strategies or treatments.

**What Do the Genetic Counselors Do?**

For individuals seeking testing, genetic counseling is available by phone at any point during the process. The genetic counseling staff (currently three full-time counselors) have received specific training in genome-wide association research and technologies as well as risk assessment for common, complex diseases. We interact with people before testing, discussing the benefits and limitations of the service and deciding whether testing is appropriate for them. When someone chooses to test, they are assigned a specific counselor, complete with a bio and picture to ensure that the person knows there is a qualified professional available to them over time.

Once results are received, people can schedule a phone consultation. We typically begin the session by eliciting questions, concerns and general comprehension. Together, we review their results, focusing on specific conditions as appropriate. We then delve deeper into personal and family history, discuss lifestyle behaviors, consider strategies to integrate this information into their life and address additional psychosocial considerations.

Although this type of testing platform looks at a large amount of genetic information, it certainly does not detect everything, especially more rare, Mendelian traits that require targeted analysis. Therefore, a key adjunct includes personal and family history risk assessment, to ensure that referrals for additional testing and services (not included in the test) are recommended as appropriate.

Working with Navigenics for over a year, aside from patient interaction I have had the opportunity to help develop company policies, practice guidelines, website content and technology to support the phone and web-based genetic counseling service. The genetic counselors also assist in product development, marketing and physician outreach and education. We provide education to co-workers so others can understand the intricacies of genetic testing, risk assessment and the role of the genetic counselor.

**Considerations**

Navigenics was founded to clarify and expand the field of genome screening and deliver this information in a responsible, accurate, easy to understand fashion - clearly a challenging task. Any effort toward a major paradigm shift, particularly when it relates to medicine and public health, should be met with healthy skepticism, so we encourage colleagues to ask questions and get involved in related efforts.

The bottom line is that the technology is here, the information is here and patients are demanding it. Now when people ask us, “What am I at risk for?" we can provide them with some insight into their genetic predispositions for common conditions. As professionals, we should be at the forefront to help guide them through the experience.

## NSGC News

### The NSGC Board Culture Statement: Articulating the Values that Guide Governance

***By the 2008 Board of Directors***

A well-defined sense of culture is part of the foundation of governance for an organization’s Board of Directors. This culture is defined by the principles of the organization, how communication takes place within the Board and between the Board and membership, and how decisions are made. Board culture develops over time, and understanding the culture is often a learning process for new Board members.

To that end, in the spring of 2008 the NSGC Board of Directors developed and approved a Board Culture Statement to provide transparency around how the Board conducts its business and to ensure that all Board members are operating under the same set of expectations. In developing the culture statement, the Board carefully considered the values of NSGC, evaluated the nature of Board discussion and decision-making and outlined general expectations for all Board members.

The NSGC Board Culture Statement is being actively used in the Board evaluation process that has been implemented as part of overall governance evaluation. We are incorporating the Board Culture Statement into our Leader/Volunteer Training at the AEC to ensure that all NSGC volunteers are aware of the standards and

expectations set for members volunteering in any capacity. The Board Culture Statement was also used during the nominations process for the 2009 Board of Directors. All nominees were provided with a copy of the culture statement to provide additional information as they considered whether they were interested in service on the Board and to let them know what to expect from their potential Board experience.

The Board takes our stewardship of NSGC very seriously, and we are proud to present the Board Culture Statement to the membership. We want this statement to be a "living" document, setting standards for the actions of the Board and volunteers as we guide the organization and ensuring that we uphold the NSGC culture as we do our work. Please review this statement and feel free to contact any member of the Board if you have questions or comments.

**NSGC Board Culture Statement**

*Approved March 2008*

1. A Foundation of Holism: The Board strives to build a foundation of "holism," recognizing that our Society serves many constituencies with sometimes different and even conflicting requirements and needs, and puts aside individual agendas for the good of the whole.

2. Diversity: The Board recognizes that diversity exists in many forms and seeks to maintain a Board that encourages respect for and inclusion of diversity at all levels of the Society.

3. Openness, Transparency and Accountability: The Board is committed to creating and nurturing an atmosphere of openness, transparency and accountability.

4. Conflict Resolution: The Board is committed to a swift, direct, honest approach to resolving conflict with one overriding objective: to work it out and move on.

5. Board/Staff Partnership: There is a strong partnership between volunteer leaders and staff based upon common expectations, trust, collaborative planning, joint evaluation, strong communication and mutual respect. Information flows directly between volunteer leaders and staff at all times.

6. Speaking with One Voice: The Board values respectful dissent and differences of opinion and practices a philosophy of debate and constructive conflict inside the Board room but unity and support for our common purpose outside. We value the "confidential" protection of our Board meeting discussions.

7. Position Seeks the Person*: The Board values the concept that the "position seeks the person, not the person the position" in developing current and future leaders of NSGC.

8. Involvement in Association Affairs: Members of the Board are expected to be engaged – reading communications when they are received, responding with input when requested and initiating discussions on provocative topics inside and outside of Board meetings. Being a Board member is more than just reading a packet of information before a meeting.

9. Consensus Decision-Making: Though the Board votes on motions, the Board favors a consensus decision-making process followed by voting on a motion.

10. Informed Decision-Making: The Board values informed decisions – seldom will it act on a matter without first having another group or individual study the issue and make a recommendation for the Board to consider.

11. Governance vs. Management: The Board believes its primary responsibility is to govern the Society by determining direction, stating desired outcomes, understanding why the direction and outcomes are desirable and determining when things will be accomplished while, at the same time, refraining from managing the Society.

12. Openness to Feedback: The Board consistently evaluates its own performance, striving not for perfection but for openness to feedback and a willingness to improve the performance of the Board as a whole and of individual Board members.

*This means that the positions are filled based on the skill sets needed in relation to the collective skills of the Board, Committee or Task Force. Members who feel they possess the appropriate skills would be encouraged to nominate themselves.

## AEC Update

### See You at the 2008 AEC!

***By Janice Berliner, MS, CGC and Stephanie Brewster, MS, CGC, 2008 AEC Co-Chairs***

There are exciting changes in the air! Join us for the 27th NSGC Annual Education Conference (AEC) in Los Angeles, CA, October 24–28 and the Short Course, "Taking Heredity to Heart: Cardiovascular Genetics, An Overview," October 23–24. Before you head to Los Angeles, make sure you check out these new offerings.

**Program Book Changes: Print Your Notes Before the Meeting**

Does your back ache after carrying around the AEC program book for four days? New this year, to save some trees as well as your back, our conference booklet has changed. Speakers' notes and PowerPoint presentations will be available online before the conference instead of in the program book. Decide which talks you cannot miss and print out the presentations you want to have on hand at the conference ahead of time. Read over the presentations on the plane and spend your first night catching up with friends instead of flipping through a big program book. There will be computer kiosks and printers available at the conference hotel for printing presentations for a fee. A smaller program book will be given out at registration with the AEC sessions schedule and hotel information so you will have something in hand to find your way around.

**Sessions Available Online**

Never able to get to all the talks you would like to see? Wish you could see some sessions again? Available for the first time this year, the sessions from the 2008 AEC will be available online after the conference. In early 2009, a recording of the conference sessions along with synchronized PowerPoint presentations will be available. Just make sure you register for access to the online sessions before the conference. Can't make the AEC this year? These recordings will be available for purchase to all members after the AEC and can be used for CEUs.

**Networking, Networking, Networking**

The AEC is a prime networking opportunity. The Welcome Reception is a good place to see colleagues and should not be missed. A list of attendees will be sent to all conference goers before the meeting. You can see who will be at the AEC and set up some of your networking activities before you go.

**Movie Screening of "In the Family"**

Join us for a special screening of the film, "In the Family," at the AEC. The film follows four women and their family members as they deal with various issues surrounding BRCA predictive testing. Watch the film and learn how you can arrange for a screening in your community to increase awareness of predictive genetic testing and the roles genetic counselors play in this process. For more information about the film, which will be broadcast on PBS on October 1, please visit **http://inthefamily.kartemquin.com/**.

**Casino Night with Local Students**

This year's Outreach Event will be a Casino Night with local college students on Saturday, October 25. Students will spend the evening playing blackjack with NSGC members as dealers. As they win fabulous prizes, the students will get to learn about genetic counseling career options. The students also will have the opportunity to visit the AEC poster session before hitting the tables to see the research being performed by genetic counselors. So when you are checking out the posters, be on the lookout for students interested in learning more about what you do.

**Best Poster Award**

Also for the first time this year, a "Best Poster" Award will be given out at the end of the Posters with Authors session. The winner will be announced later in the conference.

It's not too late to register for the AEC. Join us in the recently renovated Hyatt Regency Century Plaza for a great educational opportunity. Catch up with friends and meet new colleagues in sunny California. For more information, please visit the AEC webpage at **www.nsgc.org**.

## Student Forum

### Summer in Alaska: A Unique Internship of Genetics and Geography

***By Jennifer Dick, BS, Boston University Genetic Counseling Program, Class of 2009***

Shortly following the 2007 NSGC Annual Education Conference, my first-year classmates and I received an email from our program director, **Maryann Whalen Campion**, indicating that a counselor she had met at the conference, **Maggie Dewhurst Miller**, expressed interest in hosting a student for a summer rotation. Maggie is one of two practicing genetic counselors in the state of Alaska and works as a cancer counselor for Providence Cancer Center in Anchorage. MaryAnn extended the rotation invitation to any of us who might be interested.

At first I brushed off the idea, convincing myself that Alaska would be too far away from my family and friends, and certainly a much different living and working environment than what I was used to. But after a few days of ruminating over Alaska in the back of my mind, the same reasons that first discouraged me were quite intriguing, and I decided to pursue a summer rotation in Alaska!

**I'm Going to Alaska! Now What?**

After the initial excitement wore off, more practical concerns arose such as determining the dates of my rotation, finding housing, and addressing transportation issues. I expressed these concerns to Maggie, and she surprised me with an opportunity that would seemingly solve all of those problems. As a former University of Alaska Anchorage student, Maggie served as a mentor for high school students during a six-week summer program at the university. The Della Keats/U-DOC summer enrichment program was created for underrepresented populations of high school juniors and seniors interested in pursuing careers in medicine and health care. Students enrolled in the program live in the dorms, take classes at the university, perform community service, and participate in other various activities. Because the students are in class during the week from 9am to 5pm, mentor responsibilities are primarily during evenings and weekends. Thus, most mentors have full time jobs in addition to the mentor position. Maggie put me in touch with the Della Keats/U-DOC program coordinators and encouraged me to apply as a mentor for the summer 2008 session.

I eagerly applied for the position and after two phone interviews and three anxious months of waiting, I finally received the good news: I was offered the job! The mentor position would provide me with free dorm housing (conveniently located only several blocks from Providence Cancer Center), a group of peers to interact with on a daily basis, weekend trips and activities allowing me to explore and experience the area, and even a stipend!

With the dates set for my rotation and housing and transportation issues addressed, I was ready to head to Alaska, but the decision to go to Alaska was met with mixed reactions from others. Most friends and family were quite enthusiastic, assuring me that the trip would be a once in a lifetime opportunity! But a few were perplexed, asking, "Alaska? Why Alaska? What's up there?" To these individuals I responded, "Why not Alaska?" The truth was, I had never been to the 49th state, but I felt certain that at the very least, spending six weeks there would generate some new experiences and stories to share. And so I boarded the Anchorage-bound plane at the end of June, not knowing what to expect but excited nonetheless.

**Genetic Counseling in Alaska**

I spent most of my time working with Maggie in cancer genetic counseling, where we primarily saw patients referred for personal and/or family histories suggestive of Hereditary Breast and Ovarian Cancer syndrome. In addition, we conducted several colon cancer sessions in which we evaluated patients' risk for Lynch syndrome, APC-related conditions, and Li Fraumeni syndrome. My rotation also afforded me the benefits of attending various cancer-related rounds, seminars, and meetings, which helped me to become familiar with current cancer treatments, disease pathology, and ethical issues related to patient care and treatment.

The patient population at Providence Cancer Center is mostly comprised of middle class Caucasian women; however, I had the opportunity to counsel Native Alaskan patients as well. There are many reasons why so few native patients are seen for genetic counseling, one of which is a transportation issue. Commuting to Anchorage from many of the tribal villages and rural communities across the state is an expensive and time-consuming venture. Therefore, many natives prefer to seek medical services at community or regional native hospitals.

In addition to cancer counseling, I had the privilege of also working with **Audrey Burke**, a prenatal genetic counselor (and consequently the only other practicing genetic counselor in Alaska) affiliated with Providence Hospital. With Audrey, I participated in a diverse sampling of prenatal sessions, ranging from straightforward AMA scenarios, to balanced chromosome translocations, to a couple experiencing recurrent anencephaly. The content and flow of the prenatal sessions provided a contrast to the cancer sessions, and taking part in both during the same rotation helped me to sharpen my counseling skills.

**My Mentor Position: The Della Keats/U-DOC Program**

My mentor position was an extremely rewarding and positive experience, mostly due to the friendly and open personalities of the students. A handful of the students enrolled in the program were Alaskan natives, growing up in small villages scattered throughout the state. As a group, they educated me about their cultural beliefs, values, and practices, and what it means to be an Alaskan native in 2008. They explained and demonstrated traditions of "Eskimo throat singing" and native dances, allowed me to sample dried halibut and smoked salmon prepared by their families, and shared with me personal stories of what life is like in a village of 200 people, subsisting off the land and relying on all members of their tight-knit communities.

Through mentoring in the Della Keats/U-DOC program, I not only gained a deeper understanding of Alaskan culture, but I was able to experience much more of what Alaska has to offer. From camping trips, to hikes, to wild animal encounters (moose, bears, and even a bald eagle), I had ample opportunities to truly appreciate the natural beauty of Alaska. And of course, evenings and weekends with the students also included more typical "teen-preferred" activities, such as going to the movies, bowling trips, and pizza parties.

Overall, I feel that the combination of my genetic counseling and mentor positions made my Alaskan experience complete. I consider myself extremely lucky to have had the opportunities to live and work in a unique environment, learn firsthand aspects of Native Alaskan culture, and make lasting friendships with fellow colleagues and students. The memories made and lessons learned during my summer rotation in Alaska will not be forgotten.

**Advice to First-Year Students**

When it comes to choosing your summer rotation, think big and don't limit yourself! If there is a particular location or practice area of genetic counseling that you have always wanted to pursue, now is the time to go for it! Your first rotation can be a great opportunity for personal and professional growth, so have faith that a little determination and persistence can go a long way in making your ideal summer internship a reality.

*Please direct any student-related story ideas and inquiries to the new Student Forum editor, Denise Lautenbach, at **dmlaut@bu.edu**.*

## Research Network

# National Cancer Institute Familial Testicular Cancer (FTC) Study (CA-SIG REGISTRY ID 70)

The clinical arm of the Familial Testicular Cancer (FTC) study is being resumed to evaluate more thoroughly men with a history of testicular cancer and their relatives. This is a multidisciplinary study of families prone to the development of TGCT (testicular germ cell tumors). Eligibility criteria include families with two or more cases of

testicular cancer OR a family in which one man is affected with bilateral testicular cancer OR families in which one or more of a set of identical siblings is affected. Non-testicular germ cell tumors not testicular in origin (e.g. mediastinal, ovarian, pineal or retroperitoneal) also confer eligibility.

Data is collected regarding clinical, epidemiologic, genetic, behavioral and molecular aspects of this familial syndrome. The goal is to acquire a comprehensive understanding of both the genetic and non-genetic factors which contribute to the risk of familial TGCT. Participants contribute research specimens to the International Testicular Cancer Linkage Consortium (ITCLC) efforts to map genes for TC susceptibility. Study PI is Dr. Mark H. Greene, lead investigator is Dr. Larissa Korde and genetic counselors are June Peters and Ann Carr.

***Contact:*** Stephanie Steinbart, intake research nurse, 1-800-518-8474, **StephanieSteinbart@westat.com**, or visit the study website at: **http://familial-testicular-cancer.cancer.gov**/

## Congenital Sucrase-Isomaltase Deficiency (CSID) Study

The Biochemical Genetics Program at the University of Washington in Seattle is seeking volunteers greater than six months of age who have already been diagnosed with Congenital Sucrase-Isomaltase Deficiency (CSID) to donate a blood/saliva sample for a genotype/phenotype correlation study.

***Contact*:** Stefanie Uhrich, MS, CGC, Study Coordinator/Genetic Counselor, 1-866-577-1187, **stef@u.washington.edu.**

### Genetic Counselor Publications

## Featured Paper:

**Scacheri C, Redman J, Pike-Buchanan L, Steenblock K.** Molecular testing: Improving patient care through partnering with laboratory genetic counselors. *Genet Med.* 10:337-342. 2008.

Cheryl Scacheri, MS, CGC, is currently the Director of Genomics Education and Policy at the Cleveland Clinic Genomic Medicine Institute in Ohio where she is spearheading a campaign aimed at educating medical professionals, other employees and patients on the many ways that genetic counselors (GCs) can be used as resources. This initiative targets all individuals entering the doors of the Cleveland Clinic, including over 1,000 physicians and 38,000 employees.

This ambitious endeavor, combined with her array of past career experiences, complement her recent publication in *Genetics & Medicine* on the value of the laboratory GC. A graduate of the University of Pittsburgh genetic counseling program, Cheryl worked in prenatal for two years at The Genetics Center on Long Island and then in research, clinical and molecular diagnostics for five years in the Pittsburgh, PA laboratory of **Eric Hoffman, PhD**, focussing on muscular dystrophies, ion channel disorders and other neurological conditions. She subsequently accepted a job with GeneDx DNA Diagnostic Experts, Inc., in Gaithersburg, MD. Most recently in Ohio, she served as the Director of the Genetic Counseling Program at the Cleveland Clinic Lerner Research Institute.

Cheryl's current article highlights the advantages of using laboratory GCs, with their extensive training in relaying genetic information, as liaisons between laboratory personnel and clinical professionals. Her motivation to write the article largely was due to a miscommunication early in her career, which caused her to realize that the provision of subpar genetic services could frequently be avoided by contextual and timely dialogue between all relevant parties. Her article also serves as a didactic tool for genetic counseling students and will be used to bolster her current educational campaign.

Cheryl and her co-authors, **Joy Redman**, **Lisa Pike-Buchanan** and **Kelle Steenblock**, with the support of several contributing GCs, **Erin O'Rourke**, **Cynthia Frye**, **Karen Giger** and **Andrew Faucett**, demonstrate the benefits of clinicians working with laboratory GCs in molecular diagnostic labs in the pre-analytic, analytic and post-analytic stages of genetic testing. Benefits in the pre-analytic stage include the ability of the laboratory GC to discuss the advantages and limitations of genetic testing for a particular patient, as well as to describe the laboratory's previous experiences with specific tests. In the analytic stage, a GC can ensure that proper tests are ordered and samples prioritized that require expedited screening. In the post-analytic stage, a GC can assist a clinician in interpreting specific test results in a context relevant to a specific patient.

Through several case histories, the authors illustrate how better communication between the clinician and the laboratory can avert problems and consequently improve patient care. In a practical and thoughtful critique, the authors emphasize the role of the GC as a key resource for the referring clinician as well as an advocate for clinicians and patients within the laboratory setting.

Cheryl is an accomplished instructor, clinical and research genetic counselor and health advocate who has co-authored several publications and contributed to various medical texts. She is dedicated to public policy issues in genetics and is a member of the Genetic Alliance. Based on the breadth of her experiences and achievements, Cheryl's latest educational campaign surely will constitute another successful effort for the genetic counseling profession.

**Articles Co-Authored by Genetic Counselors June –August, 2008**
(names of genetic counselors appear in bold)

Boe NM, Towner DT, **Rhee-Morris L**, Moon-Grady AJ. Prenatal diagnosis of omphalocele and left atrial isomerism (polysplenia) including complex congenital heart disease with ventricular noncompaction cardiomyopathy: A unique case. *J Ultrasound Med.* 27(7):1117-21. 2008.

**Cowan J, Morales** A, Dagua J, et al. Genetic testing and genetic counseling in cardiovascular genetic medicine: Overview and preliminary recommendations. *Congest Heart Fail.* 14(2): 97-105. 2008.**

Gillan T, **Davies C**, Innes M, Howard J, Graham L, Chernos J, Bridge P, Parboosingh J. An undiagnosed cytogenetic abnormality results in the misidentification of a Duchenne muscular dystrophy carrier. *Am J Med Gen*. 146A:1067-1071. 2008.

Gripp K, Innes M, Axelrad M, Gillan T, Parboosingh J, **Davies C**, Leonard N, **Lapointe M**, Doyle D, Catalano S, **Nicholson L**, Stabley D, Sol-Church K. Costello syndrome associated with novel germline HRAS mutations: An attenuated phenotype? *Am J Med Gen.* 146A:683-690. 2008.

**Hercher L, Bruenner G**. Living with a child at risk for psychotic illness: The experience of parents coping with 22q11 deletion syndrome: An exploratory study. *Am J Med Gen A.* 2008 Aug 12. [Epub ahead of print]

**Hippman C**, Oberlander T, Honer W, Misri S, **Austin J**. Depression during pregnancy: The potential impact of increased risk for fetal aneuploidy on maternal mood. *Clin Genet.* 2008 Jul 14. [Epub ahead of print]

Lynch HT, **Ferrara K**, Barlogie B. Coleman EA, Lynch JF, Weisenburger D, Sanger W, Watson P, Nipper H, Witt V, Thomé S. Familial myeloma. *N Engl J Med.* 359(2):152-7. 2008.

Lindor NM, McMaster ML, Lindor CJ, **Greene MH**. Concise handbook of familial cancer susceptibility syndromes - Second Edition. *JNCI Monographs*. (38):3-93. 2008.

**Matloff E**, Caplan A. Direct to confusion. Lessons learned from marketing BRCA testing. *Am J Bioethics.* 8(6):5-8. 2008.

**Sturm AC, Sweet K**, Schwirian PM, Koenig C, Westman J, Kelly KM. Lessons learned while developing a cancer family history campaign in the Columbus, Ohio metropolitan area. *Community Genet*. 11:304-10. 2008.

Lin AE, Basson CT, Goldmuntz E, **Magoulas PL, McDermott DA, McDonald-McGinn DM**, McPherson E, Morris CA, Noonan J, Nowak C, Pierpont ME, Pyeritz RE, Rope AF, Zackai E, Pober BR. Adults with genetic syndromes and cardiovascular abnormalities: Clinical history and management. *Genet Med.* 10(7):469-94. 2008.

***Correction: This reference was listed in the Summer 2008 Perspectives at which time the first author's name was inadvertently left out. We apologize to J. Cowan for the error.*

## Public Eye

### Media Watch

**May 15, 2008 – *Nature*, "A Case of Genetic Counselling for Dr. Watson"**
Considering the recent publication of James Watson's DNA sequence and whether the sequence means much to the general public, **Myra Roche** wrote this letter to the editor asking "how a [genetic counseling] session with Watson would stack up against one with a non-scientific member of the general population." She included a humorous table suggesting the differences in genetics knowledge and savvy between Dr. Watson and the lay patient.

**June 8, 2008 – *New York Magazine*, "Mind Bomb - If you had a fifty-fifty chance of carrying a mutant gene that causes a fatal brain disease, would you want to know?"**
A man wrote this moving, personal account of Huntington disease (HD) in his family and the difficult decision to undergo testing. He described the testing protocol, including how the genetic counselor warned him, "Once you know, you can't not know." He explained that although his doctor seemed to be against him taking the test, "the genetic counselor had insisted that there was no right or wrong reason for wanting to take the gene test." He also revealed his emotions and thoughts upon learning of his positive results.

**June 15, 2008 – *U.S. News and World Report*, "A Health Gift for Men on Father's Day"**
For Father's Day, NSGC suggested that as a gift to themselves, all men consider their risk for prostate cancer. Men should look at their family history as well as other risk factors and speak with a genetic counselor about whether genetic testing might be appropriate for them. **Angela Trepanier** was quoted: "Genetic tests for prostate cancer can help save lives, but they can also raise more questions than answers. It is important that someone who has expertise in genetics helps you understand and interpret your risks, options and the impact that test results could have on you and your family."

**June 16, 2008 – *Denver Post*, "Genetic Testing Concerns Consumer Experts"**
This article explored how the numerous genetic tests currently available to consumers may be of limited utility and how consumers should be wary of whether the tests are analytically or clinically valid. The article suggested that, "With any test, you should get a clear explanation, preferably from a genetic counselor, about how the data correlate with any suggested condition."

**July 5, 2008 – *The Arizona Republic*, "Shining a Light on Genetic Testing"**
Problems inherent in the rapid development of the largely unregulated market of direct-to-consumer genetic testing were reviewed in this article. "People think these tests will provide them good information that will modify their health," said **Angela Trepanier**. "I am worried they are making health decisions based on tests that may not be valid."

**July 14, 2008 – 49 News (ktka.com), "DNA Tests Available at Local Stores"**
**Molly Lund** was interviewed about the availability of over-the-counter paternity tests. "My concern is that patients may use this testing in a situation where it really isn't appropriate for them, or they don't understand fully the kind of information they will be getting," said Lund. "I always think it's best for a person to ask their physician before you get a test off the shelf that you are not really familiar with."

**August 1, 2008 – ABC News, "What Effect Does Gene-Screening Have?"**
A TV segment reported that researchers are studying how people who undergo the growing array of genetic screening interpret the information they receive and what kind of medical advice they seek once they have that information. "As a genetic counselor, I feel this type of research is really critical," said **Jill Stopfer**. The article stated that "counselors like Stopfer worry about how people will interpret the results they get from such tests, since they typically do not include the counseling that would accompany it in a clinical setting."

**August 16, 2008 – Sun-Sentinel.com, "Reliability of Home Screenings Questioned"**
The value was questioned of the many new genetic tests now available as a part of the rising trend toward consumer-driven, personalized health care. "These are not tests that doctors use to make medical decisions," said **Talia Donenberg.** "There's no regulation behind it, and there's no hard data behind it. It's very hard for patients to tell their risks."

**September 3, 2008 – LaCroseeTribune.com, "She had cancer and I didn't: Family history of disease leads to genetic testing"**
Two sisters shared their story of choosing prophylactic mastectomy after learning that a BRCA mutation was carried in the family. One sister was diagnosed with breast cancer during the preventive surgery. **Peter Levonian**, the genetic counselor for these ladies, was quoted throughout the article, explaining the role of BRCA genes in familial breast and ovarian cancer, the value in family history assessment and screening and prevention options.

**October, 2008 – *SHAPE Magazine*, "Beating Breast Cancer"**
Within a special "Breast Health Handbook" in this month's issue of the magazine, six women shared their stories, including a 25 year old who inherited a BRCA1 mutation and started the organization Bright Pink (**bebrightpink.org)** for young women with a family history or genetic predisposition for breast and ovarian cancer. A side bar, titled "The Facts on Genetic Testing," stated "it's recommended you get screened through a genetic counseling program rather than your family doctor or gynecologist, who may not be equipped to deal with the ramifications." Readers were directed to NSGC to find a genetic counselor.

*** Note: Roxanne Ruzicka is leaving her post as Media Watch editor after several years of faithful service. Thank you, Roxanne! If you are interested in coordinating Media Watch for Perspectives, please contact Jessica Mandell at **jmandell@slc.edu**.*

# In Memoriam

## Liz Stierman

*By Steven Keiles, MS, CGC*

We are saddened to announce that after an 11 year battle with breast cancer our dear friend, colleague, teacher and mentor, Liz Stierman, passed away on Wednesday, August 13, 2008. Those who had the privilege to know her will always remember Liz as brilliant, incredibly witty, generous and completely devoted to her family and friends. Those who did not know her will still reap the benefits of her numerous contributions to the genetic counseling profession.

After graduating from the genetic counseling program at UC Berkeley, Liz began her genetic counseling career at the University of Utah in Salt Lake City. Later Liz moved to California and took a position at the Cedars-Sinai Medical Center in Los Angeles. Her interests and talent led her to a position with the California Birth Defects Monitoring Program, where she had an opportunity to influence genetics research and public policy.

One of Liz's lifelong passions was her commitment to the genetic counseling profession and to the education of genetic counseling students. She served as the assistant director of the genetic counseling program at Cal State

Northridge and was an educator and mentor to numerous students throughout the years. She was the co-founder of the SIG for Clinical Supervisors and served as the editor of *Perspectives* during the late 90s. She was instrumental in the development of the NSGC communications committee and was the founding chair of that committee. She was awarded the NSGC Region VI leadership award in 1999.

Liz's indomitable spirit will live on through her two beautiful daughters, Claire and Miranda, and in the hearts of all those who loved her.

The following is a note from Liz's daughter:

*I would like to start out by thanking everyone for their letters and kind words. I shared your messages and remembrances with my mom and the rest of our family, and it really meant a great deal to us over these past weeks. Sadly, I am writing now to let you know that my mom passed away the evening of Wednesday, August 13. It was a very peaceful passing--I, my sister Miranda, and my Aunt Karen were all with her in her last few moments, and were able to tell her how much we loved her and always would.*

*My mother did not want to have any sort of funeral services or memorial. If you wish to honor her memory, please feel free to make contributions in her name to the Tumor Vaccine Group at the University of Washington, or to the Genetic Counseling Foundation, Special Projects Fund.*

*The Tumor Vaccine Group, headed by Dr. Nora Disis, is exploring innovative forms of immune-based treatments for cancer. My mother participated in the group's clinical trials, and felt their work was the future of cancer research. The best way to donate is to make a check payable to the University of Washington Foundation. Indicate "Tumor Vaccine Group" in the memo section of the check, or via an enclosed note, along with my mother's name. Checks can be sent to:*

*Jennifer Goforth Stead*
*Development Office*
*University of Washington*
*Box 358045*
*Seattle, WA 98195-8045*

*The Genetic Counseling Foundation was created to support the integration of genetic counseling services into healthcare through research, education and public policy, and provides funding for research projects. My mother had a passion for teaching and mentoring young genetic counselors and spent her career promoting the field of genetic counseling. The contact information for the GCF is:*

*The Genetic Counseling Foundation--Special Projects Fund*
*401 N. Michigan Avenue, Suite 2200*
*Chicago, IL 60611*

*Again, thank you all for your kind words and support. My mother was a very special woman, and I know that she will not be forgotten as long as all those whose lives she touched continue to live as she would have, with compassion and kindness, humor and grace, and generosity of spirit.*

*Sincerely,*

*Claire*