Perspectives in Genetic Counseling
Volume 34, Number 1
Spring 2012

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President's Beat

How a Bill Became a Law in Pennsylvania

By Brenda Finucane, MS, CGC

In January 2012, I was among a small but hearty group who attended a signing ceremony for a Pennsylvania law creating licensure for the practice of genetic counseling. This was a project that took almost six years, with numerous starts, stops, and changes in government. With almost two hundred practicing genetic counselors and two accredited programs that train ten percent of all new graduates in the field, Pennsylvania is a key state in the NSGC's national licensure initiatives. The passage of the Pennsylvania licensure bills reflects the collective efforts of many genetic counselors across the state, notably **Virginia Speare**, **Kathy Valverde**, **Laura Conway**, **Mary Delany**, **Betsy Gettig**, **Maria Baker**, **Robin Grubs**, **Gail Martino**, **Kelly Donahue**, and **Susan Sell**, of whom some joined me for the signing ceremony. Behind the scenes, dozens of genetic counselors willingly contacted their congress members and identified families who testified on behalf of the legislation. Without their support, the bills might never have passed.

Members of the Pennsylvania licensure committee at the signing ceremony (Standing left to right: Susan Sell, Maria Baker, Laura Conway, Brenda Finucane, and Kathy Valverde; Seated left to right: Representative Thomas Killion, Governor Tom Corbett, and Licensure Committee Chair, Virginia Speare)

The car trip from my office near Philadelphia to the state capitol in Harrisburg took almost two hours, giving me ample time to reflect on the meaning of this accomplishment for our state and the genetic counseling profession. I thought back to our Licensure Committee's first tentative efforts to form a group; we were obvious "newbies" as we began educating ourselves about the legislative process. In truth, prior to my work on the licensure bill, I had skillfully avoided volunteering for anything related to governmental or legislative activities. Like many genetic counselors, my education and interests rarely intersected with the world of government affairs, with its strange rules and protocols, legal terms, and political posturing. My committee colleagues likely felt the same way, and yet we knew that efforts to license genetic counselors in our state would have to start with us.

Pennsylvania's path to licensure was made easier by those states that went before us. We were able to learn from their experiences and shamelessly borrow from the field-tested language in their licensure bills. Our committee applied for and received an NSGC licensure grant, for money spent on tangibles like conference calls and postage, which kept the process moving. We sought advice from the NSGC's Licensure Subcommittee and from its Director of Policy and Government Relations, John Richardson. From the start, we had the unwavering support of our congressional sponsor, Representative Thomas Killion, who championed the bills straight to the governor's desk.

From a personal political standpoint, I marveled to find myself rolling up sleeves and collaborating harmoniously with both a Republican sponsor and a Republican governor. As the saying goes, "politics makes strange bedfellows." In Pennsylvania, at least, the need to protect the public by licensing qualified genetic counselors crossed all political

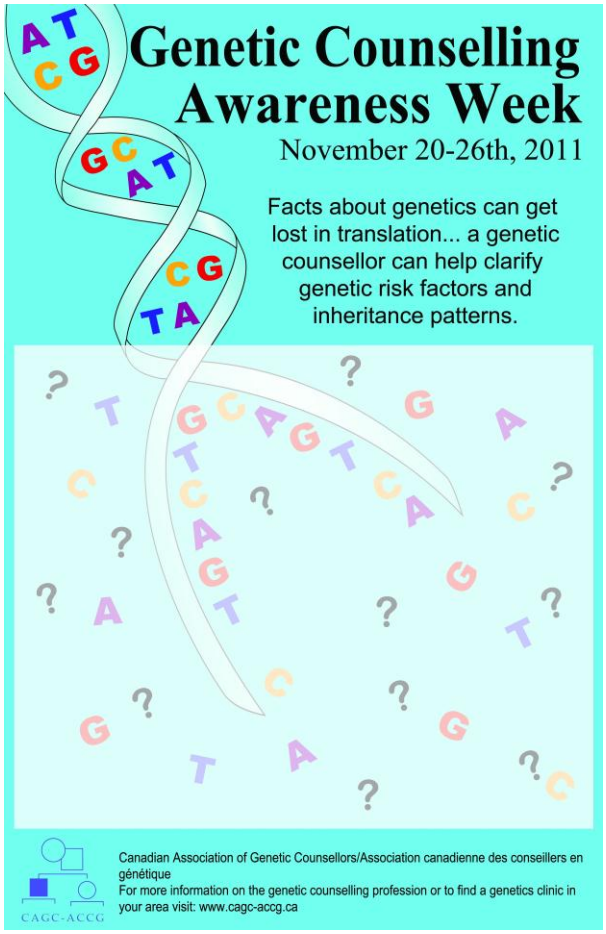
boundaries and received strong bipartisan support. Our Pennsylvania committee still has work to do, as we consult with the state this year on the specifics of licensure rules and regulations, but we're confident that we'll be able to cross these final bridges toward implementation of the new law.

One might consider Pennsylvania's achievement a local victory with little relevance to genetic counseling practice outside the state. My leadership roles with the NSGC have allowed me to view licensure in terms of the bigger legislative picture. Just as a small but dedicated band of genetic counselors was able to change Pennsylvania law, similar efforts are being replicated in many other states, building on the experiences of those who went before them.

The NSGC is making steady progress toward its goal of passing a federal bill that would recognize genetic counselors as providers under Centers for Medicare and Medicaid Services (CMS), the agency charged with implementing these national health care programs. Simultaneously, at local, regional, and national levels, individual genetic counselors and the NSGC are successfully working with payers on issues of billing and reimbursement. Each of these separate successes contributes to the larger effort reflected in the NSGC's mission: *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.* Efforts toward achieving this goal now have solid momentum after slowly building over several decades. With this in mind, let's celebrate the licensure achievements in Pennsylvania and other states as they collectively move us all forward.

Brenda Finucane, MS, CGC
2012 NSGC President

Genetic Counselling Awareness Week 2011:
Canadian Genetic Counselors Bust Genetic Myths!



As previously reported in *Perspectives*, in 2010 the Canadian Association of Genetic Counsellors (CAGC) Media and Communications Committee launched *Genetic Counselling Awareness Week* (GCAW), an initiative to increase the profile of our profession in Canada. Given our achievements in 2010, the CAGC continued this initiative in 2011.

By building on the accomplishments of last year, we were able to surpass our expectations for our second annual GCAW, held from November 20-26, 2011. Perhaps the most important piece in the success of GCAW 2011 was the chosen theme: ***Genetics Myth Busters***. This theme provided a strong focus for the development of community and institution-based events, and also resonated quite strongly with the media and the general public.

Rocking the Airwaves

Certainly, one of the most notable successes of this year was the increase in media coverage, with the airing of three different television spots. Three genetic counselors in three different provinces were showcased in the news. **Jessica Hartley** was interviewed by *CTV Morning* in **Winnipeg**, **Manitoba** about genetic myths commonly portrayed on primetime television shows and in popular literature. **Susan Christian** appeared on Global News' *Health Matters* in **Edmonton**, **Alberta** to discuss how she provided genetic counseling to the family of a young boy with hemophilia; his mother was also interviewed. **Allison Janson Hazell** was interviewed in **Toronto**, **Ontario** on the national morning show, *Canada AM*. She explained what genetic counselors do and shed some light on common myths about the field.

From Airwaves to E-waves: Who knew we had so much musical talent?

For the second year in a row, genetic counselors used social media to showcase their musical and comedic talent in the name of genetic counseling awareness. Genetic counselors in **North York**, **Ontario** and **Calgary**, **Alberta** each separately produced entertaining and hilarious videos that were posted on YouTube. Links to these videos were distributed through Facebook, Twitter and several blogs, which not only resulted in an impressive number of YouTube views, but also helped to spread the reach of our awareness campaign beyond Canada's borders. If you haven't had a chance to see the videos yet, you can find them posted on the GCAW website (www.gcawareness.wordpress.com) or by searching the following on YouTube: *"DNADiva Productions"*and *"Calgary GCs"*

A Not So "Trivial Pursuit"

Genetic counselors in **Ottawa** and **Kingston, Ontario** and **Labrador** busted myths (literally!) by holding "Genetics Trivia Night" at local establishments. The group in Ottawa even enlisted the help of some of the writers for the *Trivial Pursuit* board game to help host their event. Snacks and prizes were provided, and these events had a great turnout.

Busting myths at Genetics Trivia Night in Ottawa

Genetic counselors in **Winnipeg**, **Manitoba** reiterated the theme by cleverly hosting events that used genetic myths commonly portrayed on primetime television shows and in popular literature to help educate the public.

Speaking Out

Genetic counselors provided educational seminars to a variety of audiences across the country. For example, genetic counselors in **Calgary**, **Alberta** gave a talk to 150 people from the Alberta Family Histories Society about the importance of knowing your family history. Genetic counseling students from the University of Toronto gave a talk to a high school careers class about why they chose such an exciting career path. In addition, several interactive information booths were set up at universities and hospitals in **Victoria**, **British Columbia**; **Edmonton**, **Alberta**; **Saskatoon**, **Saskatchewan**; **Toronto**, **Ontario**; **Sudbury**, **Ontario**; **Montréal**, **Québec**; and the **Maritime provinces**. Those passing by were welcomed to peruse the displays and chat with a genetic counselor to learn more about our profession.

University of Toronto genetic counseling students, Amanda and Laurence, presenting to high school students about genetic counseling

Kirsten Bartels at an information booth in Victoria, British Columbia

These are just some highlights from events that took place across Canada. For more detailed information about the events that took place, please check out the Genetic Counselling Awareness Week website at www.gcawareness.wordpress.com.

As an incentive to participate and plan events, the CAGC once again offered $200 and $50 restaurant gift certificates to the groups who showed outstanding commitment to GCAW and to the designated theme. This year the $200 prize was given to the genetic counselors from **Toronto/North York** (Regional Coordinator: **Jeanna McCuaig**) who planned an impressive variety of events, including a film screening and a genetic counseling information night. The $50 restaurant gift certificate second place went to the **Calgary** group (Regional Coordinator: **Sajid Merchant**), who showed a lot of creativity with their initiatives this year.

Thank You

We would like to acknowledge the hard-working group of genetic counselors from the *Media and Communications Committee*, including the Chair of this committee **Mireille Cloutier**, for all the time and effort put into ensuring this year's GCAW was a success. We'd also like to personally thank our dedicated regional coordinators: **Kirsten Bartels**, **Janet Lucas**, **Jessica Hartley**, **Jeanna McCuaig**, **Rachel Vanneste**, **Aidan Thomas**, **Mary Connolly-Wilson** and **Sajid Merchant**. Finally, a huge thank you to genetic counselors across Canada who took time out of their busy lives to plan, advertise and participate in GCAW 2011.

Lauren Higgins and **Allison Janson Hazell**

Co-Chairs, Genetic Counselling Awareness Week 2011

Licensure / Billing & Reimbursement

Are You Prepared for ICD-10?

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

The Coding Corner is supported by the Coding Subcommittee of the NSGC's Access and Service Delivery Committee. It aims to assist NSGC members with the application and understanding of governmental regulations and guidelines regarding terminology and Current Procedural Terminology (CPT) / International Classification of Diseases (ICD) coding in genetic services, as well as keep the membership educated regarding billing and reimbursement issues.

As many of you may already be aware, International Classification of Diseases, Ninth Revision (ICD-9) codes have been the standard coding system to report diagnoses in patients for billing and epidemiologic disease tracking purposes for several years. Recently, you may have heard that October 1, 2013 was designated by the Centers for Medicare and Medicaid Services (CMS) as the day that all healthcare providers covered by the Health Insurance Portability and Accountability Act (HIPAA) would be required to switch to the new ICD-10 diagnosis codes. ICD-10 codes are alphanumeric, with three to seven digits, and are much more descriptive than the previous ICD-9 codes. There are approximately 14,000 ICD-9 codes, whereas there will be 68,000 ICD-10 codes. The updated codes reflect advances in technology, medicine and medical terminology. It is also hoped that these codes will reduce claims rejections, improve tracking of diseases, and assist with public health reporting.

Much debate has centered on the switch to ICD-10 codes. The American Medical Association recently opposed the adoption of ICD-10 due to concerns about the costs that may be associated with such a transition. This resulted in a recent announcement by CMS to extend the timeline on ICD-10 implementation.

It is now not clear exactly when ICD-10 implementation will occur. However, standards for electronic administrative transactions were required to be updated by January 1, 2012. Many of the third party billing services and practice management software providers are well aware of these changes and have already been in the process of implementation.

The next natural question is, "How will this affect me as a genetic counselor?" This change may bring about a great opportunity to be involved in the discussion and planning to assess the internal impact of these coding changes. It may be possible to use this transition to identify areas of improvement in care, as well as improved utilization of your unique skill set as a genetic counselor.

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

SIG Speak

From the Cystic Fibrosis and *CFTR* Spectrum Special Interest Group

CFTR2: Update on a new tool for your practice

By Karen Siklosi, MGC, CGC

Many of you reading this – especially those involved with cystic fibrosis (CF) – may have heard about the upcoming release of CFTR2, a resource intended to provide clinical information about cystic fibrosis transmembrane conductance regulator (*CFTR)* mutations. But how much do you really know about it, and how can you use it? Below you will find a quick rundown of CFTR2 basics in question-and-answer form. Even if you don't counsel about CF regularly, you might want to take a peek. We think this type of project could be a model for other conditions.

Since the discovery of the *CFTR* gene, the number of reported mutations has climbed to over 1,800. A greater number of patients now know their specific mutations, and the variety of clinical features associated with *CFTR* mutations is increasing rapidly. The expansion of newborn screening to include CF has further broadened the group of patients, families, and clinicians looking for *clinical* information about particular mutations. A greater need emerged to distinguish which *CFTR* mutations actually cause CF, and which cause related disorders or nothing at all; thus, the idea for CFTR2 was born.

What is CFTR2?

CFTR2 is both a database and website aimed to provide information about specific *CFTR* mutations to a wide audience. The name CFTR2 stands for the **C**linical and **F**unctional **Tr**anslation of **CFTR**. The CFTR2 database is a large collection of patient data in searchable form via the CFTR2 website. This website will soon be publicly available.

With a grant from the Cystic Fibrosis Foundation, partnership with the CFTR1 database team, and support and input from CF researchers and clinicians across North America and Europe, the CFTR2 database was created at Johns Hopkins University.

The database contains clinical data from over 35,000 patients from across the globe. These patients hail from the United States and Canada, as well as approximately twenty European countries. We have just begun gathering data from countries in South America, and we hope to expand the database to eventually encompass patients from every continent.

How is CFTR2 different from what is currently available?

Prior to the development of CFTR2, the only publicly available comprehensive resource for information about *CFTR* mutations was the Cystic Fibrosis Mutation Database maintained through the Hospital for Sick Children in Toronto, Ontario. This database, sometimes called CFTR1, was developed for scientists to share information and increase communication regarding the discovery of *CFTR* mutations. Although CFTR1 has been a wonderful asset to the CF research community, it was not intended to provide clinical information or be viewed by the general public.

What type of information was collected and how was it analyzed?

Patient data were collected through national CF registries, making the database a robust source of information. Most important to collect was genotype; beyond that, nearly any data available were accepted. From sweat chloride levels to the number of exacerbations requiring hospitalization, the goal was to create as wide-ranging and deep a database as possible.

From these data, the 160 most common *CFTR* mutations were selected for more comprehensive analysis. Each mutation was (or is currently being) evaluated using three methods: 1) clinical characteristics review; 2) functional analysis; and 3) penetrance

analysis – to determine whether or not it causes CF. Using these methods, the CFTR2 team has essentially assigned a "disease liability" to each mutation.

What can I learn from the website?

Perhaps the most important piece of information that the website provides is the outcome of the analysis – the "disease liability" of the 160 most common mutations in the CFTR2 database. The CFTR2 team has characterized each mutation as one of the following:

- CF-causing
- Causing varying clinical consequences
- Non CF-causing
- Unknown clinical significance (analysis not yet complete)

Each mutation can be paired with F508del ("deltaF508") or a specific category of mutations to provide clinical information from patients with various types of mutation combinations. This information includes average sweat chloride, percentage of patients who have pancreatic insufficiency, and other measures.

There is additional information for some mutations, such as those involved in complex alleles (R117H and 5T, for example). As more patients are added and analysis continues, we intend to update the list of characterized mutations and expand it beyond the current 160.

Who should use CFTR2?

The website is geared toward a broad audience, including people with a scientific background (CF team members, genetic counselors, researchers) and general users (patients, friends and family, CF carriers, parents of babies newly diagnosed by newborn screening).

To accommodate these differing backgrounds and needs, the website has two sides – one for members of the scientific community and one for the general public. Although the information on both sides is similar, the general user side offers explanations in patient-friendly language. Accordingly, the scientific side offers more details regarding mutation analysis.

We expect that genetic counselors involved with CF will find the information on the CFTR2 website useful, and we hope that you will also feel comfortable providing CFTR2 as a resource to patients and families.

When can I start using CFTR2?

Soon, we hope! We are in the final stages of website development and testing. We hope to go live on http://cftr2.org this spring. At that time, the website will no longer be password-protected and you will be able to use it freely. There will likely be an announcement to the CF and genetic counseling community when this happens.

How can I find out more information about CFTR2 or the CF and CFTR Spectrum SIG?

If you have further questions regarding CFTR2, please contact **Karen Siklosi**, **MGC**, **CGC** at ksiklos1@jhmi.edu or email cftr2@jhmi.edu.

*If you are interested in joining the CF and CFTR Spectrum SIG, please contact the Chair, **Jessica Chavey**, **MS**, **CGC** at chavey.jessica@mayo.edu or Vice-Chair, **Kristen Hanson**, **MS**, **CGC** at hansokrl@trinity-health.org.*

NSGC News

Call for Abstracts for the Audrey Heimler Special Projects Award

By Emily Edelman, MS, CGC

The deadline for the 2012 Audrey Heimler Special Projects Award (AHSPA) is **May 15, 2012**, so start thinking about your proposals today! Awards up to $5,000 are available to support projects that focus on the future of genetic counseling or the provision of genetic counseling services.

Some types of projects that might be appropriate for the AHSPA include:

- A pilot study to collect preliminary data for a larger future project
- Development of patient education materials
- Creation of tools for genetic counselors
- Development of novel ways to encourage leadership among new counselors

Additional details regarding the application process can be found here in the Member Center, Grants & Awards section of the NSGC website. All proposals must be submitted to the Executive Office by May 15, 2012 for consideration. Submit proposals to nsgc@nsgc.org with the subject heading "Audrey Heimler Special Projects Fund Proposal."

If you have questions about a proposal, please contact the Chair of the Audrey Heimler Special Projects Award committee for 2012, **Emily Edelman**, at eedelman@nchpeg.org or 410-583-0600.

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New NSGC Position Statements

The NSGC Board of Directors met in Chicago, Illinois from February 17-18, 2012 and approved three new position statements. These and all NSGC position statements are available on the NSGC website under the Advocacy section.

- NSGC Position Statement on Noninvasive Prenatal Testing/Diagnosis (NIPT/NIPD)
- NSGC Position Statement on Newborn Screening
- NSGC Position Statement on Blood Spot Storage and Use

Please feel free to refer to these statements for the NSGC's official position on various genetics and public policy issues. NSGC Position Statements reflect our official organizational position and are used in our ongoing advocacy and media efforts.

ABGC Update

A New Accrediting Body Emerges

By the ABGC Board of Directors

As genetic counseling has grown as a profession, our accrediting and credentialing body needs to evolve. At the American Board of Genetic Counseling (ABGC) Annual Business meeting held at the 2011 NSGC Annual Education Conference, the ABGC announced the formation of a separate accrediting body. This article will discuss this change and provide information about why the board has decided to separate, the current status of the separation, and the goals for the future.

Why have two separate boards?

The Board of Directors (BOD) was advised several years ago by our legal counsel that it is a conflict of interest for the certifying body to <u>also</u> accredit training programs. The conflict exists because the board requires that examinees must graduate from an ABGC accredited training program in order to qualify for the examination, and this *same* board develops the examination and sets the standards of training. The currently operating ABGC board took several measures to help ameliorate this conflict while we planned long-term for a more formal separation.

First, in 1996 when the board began to accredit training programs, the BOD split. Five members address, in detail, matters pertaining to accrediting training programs, while five members address matters pertaining to credentialing. In subsequent years, the board formed a separate Examination Committee to manage the development of the examination items and scoring procedures, and to draft test forms. Finally, the board formed a separate Accreditation Review Committee that is responsible for reviewing the program applications for accreditation and advising the BOD, and a Task Force to give recommendations on a revision to the standards of accreditation. The board formed these committees not only to help lessen any effects of the conflict, but also to allow the five-member Accreditation Committee members to begin planning both operationally and fiscally for a separate accrediting body. These actions helped to defer the conflict, but do not completely eliminate it. Thus, the BOD is actively engaged in planning and implementing a separation.

Why is it important to have certifying and accrediting bodies?

As a healthcare profession evolves, recognition of a standard of practice also evolves. In order to legally assure that the genetic counseling profession is a recognized profession within the healthcare field, there has to be a measure of achievement that defines a "competent" professional. In defining a "competent genetic counselor," the profession has to have an objective measure of practitioners (i.e., the certifying examination) and minimum training standards (i.e., accreditation of programs). Establishing these components allows us to define and protect our scope of practice through state licensure, and federal and payer recognition. It also protects healthcare consumers and healthcare facilities from unscrupulous practitioners.

Where is the ABGC in the separation process?

The BOD approved the separation in October 2010, and moved to establish a Transition Task Force (TTF) charged with developing the rules and processes of the new accrediting board. The goal is to separate completely by the first quarter of 2013.

The TTF includes two current ABGC board members, one former board member, and three non-board members who are accredited genetic counselors (two of whom are Program Directors). These six individuals have been meeting monthly since the end of 2010 to achieve a goal of separation by the first quarter of 2013. Towards that end, the TTF has established a name for the new board and drafted bylaws that define the new organization's purpose, total number of board members, nomination and election processes, and committee structure. The TTF has revised the charges for the accrediting committees, and has also looked very closely and diligently at the financial structure for the new agency. Financial stability is of the utmost importance in setting the foundation for the new agency. The board appreciates that the current size of the profession and the total number of training programs require the new board to remain highly dedicated to fiscal responsibility.

Further, the TTF has consulted with legal counsel and will begin working on the paperwork to incorporate this new organization. Filing the federal and state paperwork for incorporation and application for federal tax status may take six months or longer. Because of this delay, the ABGC BOD has determined changes to its own bylaws that are needed in order to allow the Accreditation Committee – or, as the new organization will be called, the Accreditation Commission for Genetic Counseling (ACGC) – to work independently as soon as March 2012.

What will this mean for genetic counselors and training programs?

The split itself will not cause any changes to the credentialing board (which will still be named the American Board of Genetic Counseling), credentialing processes, or the

examination. The membership of this board will continue to be the diplomates. All decisions about credentialing of genetic counselors will rest with the ABGC.

The ACGC will have a membership that includes the genetic counseling training programs. All decisions about accreditation of genetic counseling training programs will rest with the ACGC.

The Board of Directors is excited about this step as an evolution of our profession. We look forward to the launch of the new organization and will continue to keep you informed along the way.

Global Genetics

A Dream Fulfilled: The Philippines' Master of Science in Genetic Counseling Program

By Mercy Laurino, MS, CGC, LGC

Editors' Note: *"Global Genetics" is a new* Perspectives *column, highlighting the experiences of genetic counselors who have worked internationally. We kick off the column with* ***Mercy Laurino,*** *who works at the Fred Hutchinson Cancer Research Center and the Seattle Cancer Care Alliance in Seattle, Washington. She is also a Ph.D. student in the Institute for Public Health Genetics at the University of Washington School of Public Health.*

If you are or know of a genetic counselor with an interesting international story, please contact ***Janice Berliner****, column editor, at berlinej@mskcc.org to discuss submission of an article.*

I vividly remember when Dr. Carmencita Padilla, a medical geneticist from the Philippines, asked me to collaborate with her in creating the first Master of Science in Genetic Counseling training program in the country. We initially met in San Diego at the American Society of Human Genetics meeting in 2007. Over a cup of coffee, Dr. Padilla told me that the Philippines urgently needed a training program in genetic counseling, given the increasing application of genetic and genomic technologies. She told me about the expansion of the country's National Newborn Screening Program, the Philippines Birth Defects Study Surveillance Study Group, and the proposed implementation of the Telegenetics Program.

Honestly, I was initially apprehensive to accept the offer, because I couldn't foresee being able to take on such great responsibility so early in my career. In 2007, I had only been a genetic counselor for about four years, and I did not have any experience

implementing training programs. However, just as W.H. Murray's passage in *The Scottish Himalayan Expedition* says –

> Until one is committed
> there is hesitancy, the chance to draw back,
> always ineffectiveness.
> Concerning all acts of initiative (and creation),
> there is one elementary truth,
> the ignorance of which kills countless ideas
> and splendid plans:
> that the moment one definitely commits oneself, then Providence moves too.

As a Filipino immigrant, I feel I was offered the chance to make a remarkable contribution to my home country. I was invited to collaborate with the Philippines' highly motivated medical genetics team to promote health in the Filipino population. The care of the Philippines' 92 million people, in terms of clinical and research genetics, was in the hands of only seven medical geneticists. It was evident that more support from health care providers with specialized training in genetics and genetic counseling was needed. I realized that I could really make an extraordinary difference – I didn't have to wait.

Then the moment came when I fully committed myself – the time I did say "yes." I shifted my conversation with supporters from "I don't know if I can do this," to "how I am going to make this happen?" I gave my word to Dr. Padilla in 2008 that I would take part in this partnership, and I gave myself one year to prepare. I ended my work at the University of Washington's Genetic Medicine Clinic in May 2009, and went to the Philippines to start curriculum development in a formal way.

Given the current needs of the country, we committed to developing a curriculum not only to train Filipino students in didactics pertinent to genetic counseling, but also to train them how to best integrate this field into the existing public health care infrastructure. My work began by reviewing several existing genetic counseling curricula from various programs around the world.

Early on, we realized that one of our challenges was finding faculty members who had the expertise to teach specific courses. We ultimately ended up requesting a U.S.-based expert in genetic epidemiology to teach some of our modules via web-based communication. In the end, the curriculum became a compilation of both genetic counseling and public health didactic courses, as well as clinical training rotations.

In January 2011, the Board of Regents at the University of the Philippines (UP), Manila formally approved the curriculum of the M.S. in Genetic Counseling Program. It was certainly a historic moment to welcome and teach the Philippines' first cohort of genetic counseling students six months later. The students came from diverse backgrounds: nursing, cell and molecular biology, medical technology, social science, and medicine. Some of the students had previously worked at the country's Institute of Human Genetics Department as newborn screening program managers and nurse coordinators. As part of

their training, the medical fellows are also now required to obtain their genetic counseling degrees – a major accomplishment.

I am proud to share that one of these pioneering students, Mr. Peter James Abad, has an accepted publication in the *Journal of Genetic Counseling* to reflect how the issue of cultural awareness in genetic counseling impacted him. He states, "In the Philippines, culture plays a powerful role in shaping the people's understanding of what is normal and deviations from normal, the latter often attributed to supernatural and mystical retributions. Acknowledging the already existing folk understanding and perceptions about the cause of their illness is important prior to explaining its probable genetic etiology."[1]

Furthermore, the UP Manila and Stanford University's M.S. in Human Genetics and Genetic Counseling departments have established a "sister program" partnership. The intention is to promote cross-cultural awareness and clinical exchange through bi-monthly video-conferencing. We had our first collaboration in November 2011, which proved successful. Stanford made the connection to UP Manila's secure network, while I was on Skype, since I had been back in Seattle since September. I crossed my fingers the whole time for technology to cooperate, and thank goodness it did! The UP Manila and Stanford genetic counseling students met virtually, and a second-year-student from Stanford presented the first patient case for international discussion.

If left to my own timeline, the fulfillment of this dream would have not happened until 35 years from now – as my retirement work. Indeed, it was the ongoing gracious support and valuable guidance from my family, friends, and esteemed medical genetics and genetic counseling colleagues that fueled my commitment and determination. I am especially grateful to receive insightful words of wisdom from my own program director, **Carol Walton, MS, CGC,** in the midst of this work.

And no matter how much I pinched myself (and had teary eyes) during my recent flight back to Seattle from Manila, this dream – not just a dream anymore – had manifested into reality.

Reference

1. Abad PJ. Explanatory models of illness may facilitate cultural competence in genetic counseling. *J Genet Couns*. 2012 Jan 28. [Epublication ahead of print.]

For details on the Philippines' M.S. in Genetic Counseling Program, please go to http://ihg.upm.edu.ph or email mercy@uw.edu

Student Forum

A New "Sister Genetic Counseling Program" Partnership between Stanford University and the University of the Philippines, Manila

By Kelly Ormond, MS, LGC and Louanne Hudgins, MD (Stanford University); Mercy Laurino, MS, CGC, LGC, Maria Melanie Alcausin, MD, DPPDS, and Carmencita Padilla, MD MAHPS (University of the Philippines, Manila)

In October 2011, a "sister program" relationship began between the genetic counseling program at Stanford University and the newly initiated genetic counseling program at the University of the Philippines (UP), Manila. This collaboration is meant to enhance genetic counseling training by allowing mentorship between an established and a new program, and to promote cross-cultural awareness for all involved. A bi-monthly video case conference is scheduled for students and faculty members to share challenging genetic counseling cases, with a focus on both the medical management and psychosocial issues. We hope that in the future we can also initiate joint research projects and, potentially, an exchange of clinical rotation sites.

In this article, we share our genetic counseling students' reflections on our international partnership thus far. For each program, selected student representatives summarized the insights and perceptions shared by their fellow graduate students.

Reflections from the Stanford University genetic counseling students:

Stanford's sister program with the UP Manila genetic counseling program provides a unique opportunity for students to gain insight into healthcare systems in other parts of the world. Exploring complex clinical cases together is extremely valuable, as it allows us to discuss similarities and differences in how we think about the issues that arise in genetic counseling sessions. Through collaboration, we can develop a more informed view of how health professionals from other cultures view and respond to clinical genetics cases. This helps us better understand the differing cultural norms and values to which our patients may adhere. Additionally, the interchange between our sister programs provides us with an excellent opportunity to learn about the challenges that genetic counselors face in other countries. We learn how their culture influences their counseling styles, as well as their outlooks of the field. Being able to share cases and learn about differences in culture and health care in the Philippines provides a new perspective on genetic counseling in the U.S., and knowledge that we can apply when counseling Filipino families.

As the field of genetics continues to grow rapidly, the need for genetic counseling will continue to expand globally. Our relationship with UP Manila will allow us to watch the development of their program, and demonstrates the genetic counseling community's effort to develop services in countries where the field is beginning to emerge. Along with

broadening our professional network, we will create relationships that we can utilize as we enter the field. Lastly, our interactions with the UP Manila program will expose us to the nuances of integrating genetic counseling services into foreign healthcare systems; this is particularly exciting considering that some of us are international students or bilingual ourselves. In the future, we hope that this exchange between the UP Manila and Stanford will pave the way for future international collaboration between the two programs. As graduate students, we look forward to getting to know the students in Manila better in the upcoming year, both as peers and future colleagues.

2011-2012 Stanford University Genetic Counseling Students

Front Row: Megan Grove, Layla Sharmirzadi, Greg Kellogg, Nadine Reyes, Emily Hurford
Middle Row: Bita Nehoray, Adrienne Miller, Jessica Profato, Kristen Dilzell,
Emily Hendricks, Rachel Farrell, Sahil Kejriwal
Back Row: Aarin Ables, Amirah Khouzam

Reflections from the University of the Philippines, Manila genetic counseling students:

This school year, UP Manila responded to the increasing demand of health professionals formally trained in genetics and genetic counseling by offering the very first genetic counseling program in the Philippines. Being the pioneer batch, the challenge for us is immense. We are currently in the midst of defining and embodying the ideals of what a genetic counselor in the Philippines should be. We are expected, in two years, to respond to the genetic counseling needs of our patients and members of their families. In addition, we face the challenge to sustain and expand the presently available national genetic services in the country, such as the Newborn Screening Program and the National Telegenetics Referral System. We are likewise expected to be proactive in the provision of other genetics services in the future.

Taking on this challenge may not be an easy task, but thanks to the commitment and creativeness of our mentors, we are partnering with Stanford University as our sister program. This is a great opportunity for us budding genetic counselors to learn from an

established genetic counseling program. We started this collaboration by conducting bi-monthly videoconferences where, alternately, students from the two programs select and present a case that is special in each of the program's clinical practice settings. The initial videoconferences have opened us to the possibility of acquiring knowledge and skills outside the confines of our classroom and highlight the importance of utilizing technology to the advantage of both parties. We deem that this regular videoconference will serve as an excellent venue for the exchange and discussion of issues that surround the provision of genetic counseling services to a wide array of patients and their families. Perhaps an important outcome of this collaboration is the development of an awareness and sensitivity to cultural differences that exist in our uniquely different clinical practice settings. As such, this will be an occasion for us to learn the various social, economic and cultural contexts of genetic counseling. This collaboration moves us closer to achieving cultural competence – an essential element in bridging the cultural differences that initially exist between us, our patients and their families.

For the future of our genetic counseling profession, we are profoundly committed to continue our current "sister program" collaboration to enhance the training and development of our students and ourselves. Knowing about the global provision of genetic counseling will provide us awareness of how to better understand our patients from other cultures, and will positively impact care in both countries.

We intend our international partnership to be long-lasting and meaningful, and hope that it will strengthen our shared genetic counseling passion. We encourage other established genetic counseling training programs to consider such partnerships for the future.

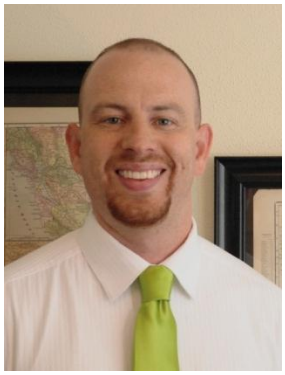
2011-2012 University of the Philippines, Manila genetic counseling students

Front Row: Margrette Patena, Gay Luz Talapian, Melissa Baluyot, Maria Elouisa Reyes, Vanessa Aldemita, Peter James Abad
Back Row: Riza Suarez, Angela Villa, Aster Lynn Sur

The New Graduate Life

How to ~~Make Millions~~ *Have Success* Launching a New Genetics Clinic

By Ian Wallace, MS, CGC



The first thing I learned when establishing a new genetics clinic is to never, ever skip an opportunity to promote my services. That said, if you know of anyone in need of genetic consultation in eastern Washington or northern Idaho, have them call (509) 332-5106 to set up an appointment at their earliest convenience. Okay, now that we have that out of the way, let's proceed.

After spending my entire two-year genetics career in Boston, my dear wife decided that we should head west to her hometown to establish our new lives and careers in rural Washington State. Note that Boston is a city where you can go years without seeing a pickup truck outfitted with a gun rack, but you can't go five minutes without seeing another genetic counselor. Now I live in a place where wearing hunter orange is actually stylish and never displayed with any sense of hipster irony.

Before I arrived, the nearest genetic counselor in our new location was a ninety-minute drive away, with some of my distant patients needing to drive three hours one-way. I've learned many lessons since I started my new position last October, but this article will focus on three: how to get a job, how to promote your services, and how to deal with the personal challenges and triumphs of establishing a new practice, especially as a fairly recent genetic counseling graduate.

Since I'm not smart, talented, or good-looking enough to start a private practice, I focused on creating a new clinic for an existing hospital. For example, I can barely spell the word "insurance," so I need to rely upon billing experts to help me get through the hurdles of reimbursement. I'm very thankful to work with a highly competent, endlessly patient, and intellectually curious staff that provides me with all the support I could ever need. I lucked out by having a great working environment, but it's something job seekers

should deduce early on in the process. The willingness of the hospital staff and leadership to support you through the long slog of building a new practice is crucial to your long-term success and happiness. Here are a few steps to creating a new position in a rural location:

1. **Find a place where you want to work.**
2. **Contact the hospital's Chief Executive Officer (CEO) to arrange a meeting.** Don't go through the Human Resources staff unless you want to fall into a pit of hiring regulations and despair. Scheduling meetings with the CEO of a large hospital is difficult, but at a smaller hospital there is far less bureaucracy.
3. **Always lead with the hospital's needs when making your proposal to the CEO.** He/she has likely never heard of genetic counseling before, and since the CEO's primary responsibility is running a successful business, you have to think about your proposal from a financial viability standpoint first.
4. **Don't expect to get a full-time position right away. (If you do, that's a bonus).** Communicate that you're ready to drum up referrals and that you're willing to work on an as-needed/per diem basis with no benefits to start. This will excite most business leaders, as there's little to no risk for them.
5. **Get ready.** You can start by getting a new mattress, because with all the money you'll be making, you're going to need a place to store all those $100 bills!! Not really. However, the pay and benefits provided by rural hospitals are often better than their urban counterparts to assist in the recruitment of medical staff.

Now that you have a job, it's time for self-promotion, because most of the area's physicians, nurses, and residents haven't a clue what genetic counselors actually do. If they don't know about you or the benefits your service provides, you won't be seeing any patients anytime soon. Here are a few tips:

1. **Geography is important**. Always think about drawing patients from across borders and competing for the attention of residents from neighboring towns, counties, and even states.
2. **Develop a press release for the hospital's Public Relations staff to send to all the regional newspapers.** Add a bio page with a description of you and your services to the hospital website. Create a LinkedIn account and add your clinic to Google Maps. These will often be high on search results, which are critical in the promotion of any modern business. I personally question the value of Facebook and Twitter to drive patients to my door, but some counselors may find these to be valuable platforms.
3. **Contact all regional physician groups and clinics and ask to present at their staff meetings.**
4. **Send a letter introducing yourself to all area physicians and nurse practitioners.** In rural areas, the "specialist" concept so common in big cities can be foreign, as a family practitioner will often cover obstetrics, pediatrics, geriatrics, and everything in between. In any given area there may be a

different medical landscape, so stay open to other practitioners who may provide referrals.

5. **Present to any group, club, or health fair that will take you.** Don't worry about targeting only the patient populations that will be seen in your clinic, as seemingly irrelevant people and organizations can still lead to referrals via word-of-mouth.

Establishing my own clinic has affected me in many personal ways, some of which were predictable and some that were wholly unexpected. I expected that I would work harder than I did in previous positions, as I now see patients for any genetic indication and I alone am responsible for cultivating relationships with providers to gain a steady stream of referrals. In my last job, I would come to work and nonchalantly look at the schedule, not really caring how many people were scheduled for that day and not really appreciating what went into getting them through the door. And zero patients? That used to simply mean a boring day, but now it brings an amalgamation of dread, shame, and tedium fueled by fears of looming budget cuts, thoughts that I've disappointed the hospital leadership who have put so much faith in me, and uncertainty over when the next patient will come. Now every patient sparks an internal celebration, much as the vintner rejoices at the culminating cork, having once borne witness to the barren vine.

I'm amazed at how often genetic counselors and their skills are taken for granted in places where their presence has never been questioned. I would have never realized this until I got to a region where genetic services were non-existent. Literally every day, someone tells me how excited they are to have my services available, and people have an insatiable intellectual thirst for even the most basic of genetic information. My pay, benefits, and office space all dramatically improved when I started this new position, but it's this intangible feeling of veneration and value that has been the greatest benefit of all. This is not a tribute to me, but rather to the genetic counselors that developed the profession and those who taught and mentored me; if I have seen it further, it's by standing on the shoulders of giants.

I have long held a professional interest in providing genetic services to traditionally underserved patient populations, and it can be argued that those with the least access to our specialty are our rural residents. According to the 2010 Census, for the first time in our nation's history less than twenty percent of the U.S. population lives in rural areas. This continues a two century-long exodus from rural farms to urban cities and suburbs, with projections that ninety percent of Americans will be urbanized by 2050. So, while it makes sense for most genetic practitioners to be in the cities where most patients are and will continue to be, so long as there are still a few rural neighbors amongst us, someone should provide care for them. And if we don't, who will?

Ian Wallace established the Center for Genetics at Pullman Regional Hospital in Pullman, Washington. He can be reached at Genetics@pullmanregional.org *to answer*

questions, provide you with a job proposal template, or give you questionable advice on any number of provocative topics.

Genetic Counselor Publications

Editors' Note: *Going forward, the Research Special Interest Group (SIG) will be coordinating this listing of publications by genetic counselors. The listing will be housed at the Research SIG's page on the NSGC website (*http://www.nsgc.org/MemberCenter/SIGs/Research/tabid/262/Default.aspx*). One publication will still be highlighted in Perspectives as part of a Feature Article. Please contact* ***Rebecca Hulinsky, MS, CGC*** *at* Rebecca.Hulinksy@va.gov *to have a citation included.*

Feature Article

By Sara Spencer, MS, CGC

Bernhardt BA, Zayac C, Trerotola SO, Asch, DA, Pyeritz, RE. Cost savings through molecular diagnosis for hereditary hemorrhagic telangiectasia. *Genet Med*. 2012 Jan 26. [Epub ahead of print].

"Genetic testing is so expensive!" As genetic counselors, you have surely heard this said before. According to research performed by **Barbara Bernhardt** and her colleagues at the University of Pennsylvania, however, genetic testing *saves* healthcare dollars in some families. In fact, appropriate genetic testing for hereditary hemorrhagic telangiectasia (HHT) in the United States versus a repeated clinical screening protocol for four at-risk family members saves $22,000 in reimbursable healthcare dollars. To get this data into the hands of federal, state and local elected leaders, insurance and hospital executives, and industry and consumer representatives, Bernhardt and her team published a policy brief that is now circulated to over 6,000 policy makers. She highlights in her article that such cost savings may likely be the case with other genetic syndromes that have sufficient negative predictive value, like Lynch syndrome and the long QT syndromes. She also points out in her article that, "the cost of DNA analysis continues to fall,

eventually to be replaced by whole exome or whole genome screening with a target cost of $1,000 or less in the near future."

Bernhardt and her colleagues began conducting research on the economics of genetic services back in the 1980s, when they published studies documenting the time and labor intensity of delivering genetic services. They also showed how genetics clinics bring downstream revenue to medical centers.[1,2,3,4,5] To this date, genetic counselors could make use of this data to support the value of genetics services!

"I've always been excited by research," said Bernhardt, who devotes up to eighty percent of her current position to research. As a genetic counselor, she enjoys turning clinical observations into researchable questions. Throughout her thirty-plus year genetic counseling career, she has always had her hands in research in one way or another. In 1991, she was fortunate enough to have been chosen for a pure research position with the Genetics and Public Policy Unit at Johns Hopkins University. There, she had the privilege of working with a tremendous team of researchers that helped her to expand her research skills, including manuscript and grant writing, and data analysis and interpretation. Bernhardt receives grant funding to perform research and has been a recipient of a number of National Institutes of Health (NIH) grants. The research reviewed in the current featured article was funded through the National Human Genome Research Institute (NHGRI).

Bernhardt has some words of wisdom for genetic counselors interested in conducting research. She says, "Always be on the lookout for an interesting project, and start small by connecting with other people doing research; they can become your research mentors. Or, take the step to be principal investigator on a small project which can lead to something much bigger." Bernhardt states, "These past few years, with all the technological advances in genetics, there have been incredible opportunities for genetic counselors to be involved in research." She also suggests taking advantage of opportunities to enroll in classes in research methodology and grant writing, and volunteer to review grant proposals or manuscripts because it will help you to think like a researcher and improve your writing skills.

Bernhardt's work is very inspiring and her contributions have played – and will continue to play – an important part in transforming the uses of genetics in medicine.

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AEC Update

NSGC 31st Annual Education Conference

By Claire N. Singletary, MS, CGC, 2012 AEC Chair and
Quinn Stein, MS, CGC, 2012 AEC Vice-Chair

We are excited to invite you to join us in Boston, Massachusetts for the NSGC 31st Annual Education Conference (AEC). You will soon receive your program brochure with all of the dates and deadlines for the AEC, which will be held **October 24-27, 2012**. We look forward to expanding the spectrum of our educational offerings at the Hynes Convention Center.

Schedule shifts to Wednesday to Saturday format

In response to the membership's desire to not lose an entire weekend to AEC attendance, the schedule has changed from a Thursday to Sunday format to a Wednesday to Saturday format. However, to continue to provide the maximum number Continuing Education Unit (CEU) opportunities, the content is expected to extend through the afternoon on Saturday. We encourage you to plan to stay in Boston on Saturday evening so that you may appreciate all of the educational offerings of the AEC and get to enjoy historic Boston after the conclusion of the meeting.

The 2012 AEC will again begin with the "Welcome to the AEC" orientation, followed by the opening plenary Janus Series and Best Abstract Awards on Wednesday afternoon. Concluding this kickoff will be the Welcome Reception in the Exhibitor Suite on Wednesday evening. There will be three full days of outstanding educational opportunities on Thursday, Friday, and Saturday. Attendees wanting to maximize their learning and CEU opportunities should consider attending the Pre-Conference Symposia Wednesday morning.

Pre-Conference Symposia

Based on the positive feedback from the past several years, we will again have six Pre-Conference Symposia on the opening day, Wednesday. The Pre-Conference Symposia are high level, in-depth sessions for specific specialty practice areas, new issues in genetics and genomics, or professional development topics. Each session will last five

hours, allowing for a deeper review and discussion of a particular topic. The attendance at each symposium will be smaller than at the Educational Breakout Sessions, which will allow for a more interactive experience. Pre-Conference symposia will require separate registration from the AEC and will have limited space available. Sign up early!

Continuing Education Units

The NSGC is approved as an Authorized Provider for CEUs through the International Association for Continuing Education and Training (IACET). IACET requires that the NSGC document attendance for the sessions for which individuals are requesting CEUs. The 2012 AEC is pleased to announce a transition to electronic evaluations instead of Scantron® forms for our conference in Boston. The details are still being worked out, so please watch for future *Perspectives* articles with additional information.

Program book

A link to the conference handouts will be sent to registered attendees prior to the conference; attendees may review the conference materials prior to arriving in Boston and print the ones that they wish to have on paper. Another option is to download handouts for viewing on your tablet, netbook, or laptop device for use during the presentations. In addition, many of the onsite materials will feature Quick Response (QR) barcodes that may be scanned to gain session information. Attendees who have smartphones are encouraged to download a QR scanning application prior to arriving in Boston. For example, RedLaser is a free QR scanning application available for iPhones in the App Store, Android phones in the Android Market, and the Windows Marketplace. Similar free apps are available for iPhone, Android, BlackBerry, and other smartphones.

Accommodations

The AEC will be held at the Hynes Convention Center in the heart of Boston's historic Back Bay neighborhood. Two nearby hotels, the Boston Marriott Copley Place and the Sheraton Boston Hotel, will have room blocks available for NSGC AEC attendees. Surrounding the convention center area, attendees will find world-class shopping, dining, and entertainment. Visit http://www.cityofboston.gov/visitors/thingstodo.asp or http://advantageboston.com/Hynes/Advantages.aspx for more information.

Dates to remember

Early bird registration is expected to launch in mid-May and run through the summer. Watch for future e-blasts and discussion forum posts announcing the launch to take

advantage of this discount! Abstracts for platform or poster presentations will be accepted from **March 15 to May 14, 2012.** Please see the NSGC website for more information.

Join us as we embark upon a new path for expanded genetic counselor education by utilizing a conference center location (instead of a hotel) for the NSGC's 31st AEC. We look forward to seeing you in Boston.

If you have questions, please contact Claire N. Singletary at Claire.n.singletary@uth.tmc.edu or Quinn Stein at quinn.stein@sanfordhealth.org.

Resources / Book Review

Reviewed by Deepti Babu, MS, CGC

Blink: The Power of Thinking Without Thinking

Author: Malcolm Gladwell
Publisher: Little, Brown and Company (1st Edition, 2005)
Pages: 288
Retail price: $25.95
ISBN-10: 9780316172325
ISBN-13: 978-0316172325

I was introduced to Malcolm Gladwell's book, *Blink,* when a fellow genetic counselor suggested it for our Genetics Book Club that we started at work. In fact, it was the first book we read together. I've never been part of a book club, so I was motivated to read and discuss it with my colleagues. Given that I don't find much time to read for pleasure these days, I was relieved to find that the size of the book was not intimidating.

I first learned of Gladwell while watching the 2010 Public Broadcasting Service's television series "Faces of America," which featured famous people discussing, of all things, their ancestry and family lineages. They traced their family roots using what they referred to as "tools of genealogy" and "DNA testing." Gladwell has been a staff writer at *The New Yorker* since 1996 and during that time, he has also been a fairly prolific book author.

Blink introduces the idea of "thin slicing," which is essentially what we do when forming conclusions in the blink of an eye. Many of us think that a conclusion is only valid after careful thought. However, Gladwell challenges that a "blink" conclusion may have as much validity as one that arises following analysis – in fact, it may even be a stronger one. He suggests that we could potentially be more efficient if we took more "blink" moments seriously and acted on them.

Some of us naturally think this way and act on thin slicing already. Others, like me and many other genetic counselors, have learned that most – if not all – conclusions require prior consideration. Certainly, in science, this is usually true. There are clear correct and incorrect answers, and going with your first reaction isn't always going to lead you down the right path.

After the introduction, Gladwell gives several illustrative examples of thin slicing in action with each chapter. It begins with European museum curators analyzing a famous statue prior to potentially adding it to a collection. One particularly astute curator thin-slices the statue and realizes it is a fake, saving the museum significant amounts of money and embarrassment.

This engaged me, and at this point I couldn't easily put the book down. Pages flew by without my noticing as I read more examples, such as the inner-city African-American young man that police officers assume has a gun when they spot him from behind one dark evening. He does not. This was an example of thin slicing gone wrong.

A few chapters in, and I began to get bored. Gladwell kept giving more stories of thin slicing without offering explanations as to whether they were effective or made sense. The theme was repetitive and tedious. I kept thinking, "Okay, I get it… so what about it?" It would have been more interesting if Gladwell stepped in and offered insight about the examples, rather than try to convince me about thin slicing by simply offering more examples. More is not always better.

The book doesn't end wrapped with a neat little bow. It left me hanging with no obvious conclusion. Was I supposed to thin slice it?

Fortunately, my colleagues and I found insights from *Blink*, even if Gladwell didn't spell them out for us. We figured that everyone thin slices, even if we're not always aware it's happening. Not everyone acts on his or her "blink" reactions, but some do. For those who do, thin slicing may really be a faster and more effective way to get to the same conclusion they would have reached after giving it significant thought.

To tie that in to genetic counseling, we discussed examples of how we may thin slice to form conclusions about a patient or colleague, and how they may be doing the same with us. We felt it was good to be aware that thin slicing occurs – and that sometimes it can be useful or, conversely, detrimental in our interactions with others. *Blink* was not a genetic counseling text, per se, but reading it allowed us to have introspective moments to reflect on how thin slicing impacts our personal and professional relationships. This is something we, as busy professionals, don't often make time to do.

Although I think it could have been written better, *Blink* discusses an intriguing topic that can actually be applied to some genetic counseling scenarios. In that sense, it worked well for our book club because it was an effective catalyst for what turned out to be a thought-provoking evening.

Research Network

By Emily Place, MS, CGC

Editors' Note: *Going forward, the Research Special Interest Group (SIG) will be coordinating this listing of clinical research opportunities. The listing will be housed at the Research SIG's page on the NSGC website (http://www.nsgc.org/MemberCenter/SIGs/Research/tabid/262/Default.aspx). Please contact* ***Meadow Heiman, MS, LCGC*** *at mheiman@ihtc.org to have a study listing included.*

Simons VIP Connect

Simons VIP Connect **(**www.SimonsVIPConnect.org**)** has launched a new research study. The Simons Variation in Individuals Project (VIP) is characterizing individuals with 16p11.2 deletions and duplications. Both of the child's biological parents are strongly encouraged, but not required, to take part in this study. One parent must be willing to travel for a minimum of two days to one of the study sites which include Baylor College of Medicine, Houston; Children's Hospital of Boston; and University of Washington, Seattle. The visit will include medical, neurological, and psychometric assessments and MRI. Research findings will be shared with the families. All expenses will be paid. A web-based community for 16p11.2 families has also been developed to facilitate communication among these families.

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

Myelin Disorders Bioregistry Project

Children's National Medical Center in Washington, D.C. is recruiting patients for the Myelin Disorders Bioregistry Project. Eligible patients are those with a leukodystrophy, both with known and unknown genetic etiologies. Cases are reviewed by a pediatric neurologist with special expertise in biochemical genetics and leukodystrophy. To enroll, patients must provide medical records, CD-ROMs of neuroimaging studies, and a biological sample. Next generation sequencing approaches for diagnosis and discovery of novel genes may be used when appropriate.

Contact: Johanna Schmidt, CGC at 202-476-4975 or jlschmid@childrensnational.org

Study of novel chromosome rearrangements

Families with previously identified chromosome rearrangements are encouraged to enroll in this study at Emory University in the Department of Human Genetics. Dr. Katie Rudd is investigating the causes of chromosome rearrangements by analyzing the DNA sequences underlying chromosomal breakpoints. Participants are asked to provide a blood sample and previous cytogenetic results.

Contact: Katie Rudd, PhD, FACMG at katie.rudd@emory.edu