Perspectives in Genetic Counseling - Volume 31, Number 2

President's Beat
Leadership in the NSGC

In talking with many members over the past few months, I know that there are questions about the responsibilities of our organization's leaders. I also believe there is a view that there are limited opportunities for leadership positions within the NSGC. In this column I will attempt to address these and other perceptions to help take some of the mystery out of how the NSGC functions. In the process, I hope to inspire many of you to become not only the future leaders of your organization, but today's leaders as well.

Most people would identify the Board of Directors and Committee chairs as the leaders of the NSGC. However, those are just the obvious ones. There are also the SIG chairs, organizational liaisons, Task Force chairs and members, members of ad hoc committees, Committee members, Editors of our newsletter and journal, advisory group members, chapter leaders, and the list goes on. Each one of these individuals helps to lead some aspect of this organization. Without all of these working parts, the NSGC would not be the success it is today.

With the continued involvement of these individuals and many more like them, the NSGC can continue to grow and thrive in this rapidly changing healthcare environment. The NSGC's 2010-2012 Strategic Plan was approved and released last month. This was the result of the hard work and involvement of many people throughout the last nine months. If you have not had a chance to review it, I would encourage everyone to take this opportunity to see what your organization intends to accomplish over the next several years. Additional information about the plan is available on the President's Blog, including a link to the copy posted on the NSGC website.

How is strategic planning tied to the leadership of the NSGC? Development of the Strategic Plan was the main focus of your Board of Directors during the first several months of this year. This was completed with significant input from the NSGC's leaders, key NSGC stakeholders, and many outside organizations representatives as well. In addition, we received direct input from about 25% of our membership through our vision focus groups and vision survey. These members who shared their thoughts are an additional 500 people I would include as leaders of our organization.

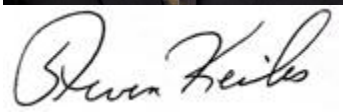
Many of you may not be sure of the precise focus or activities that take place at the Board level. The recent work in developing and approving the Strategic Plan is representative of the work of our Board, as it focuses on strategic thinking to go along with our strategic planning. The role of your Board members is to take a high level look at the organization to determine the direction(s) it should take. The Board accomplishes this by bringing together their diverse experiences, unique ideas and visions for the future to come to a consensus about what is in the best interests of genetic counselors and the genetic counseling profession, and what the NSGC can achieve on behalf of all of us.

It is not the role of the Board to discuss details of how we will accomplish our plans. That is the role the Board entrusts to our many Committee, Task Force and Workgroup leaders, and volunteer members in partnership with our competent staff. Together, these individuals provide the power to move our organization into the future, and emerge as the leaders of today and tomorrow. One of the things I love about this organization is that we all work together for the greater good. As volunteers

become leaders, many leaders continue to volunteer and help us accomplish the tasks that move us forward.

We have a lot of work to do in order to accomplish the goals we have set forth in our new and very ambitious Strategic Plan. However, I know there are more than enough leaders among us who will step up to drive our efforts and I have faith that, together, our membership can make the plan a reality. As we watch our profession grow into an integral part of the healthcare system and as genetic counselors become fully integrated across it, we can all be proud knowing that we all have taken leadership roles in our achievements.

I look forward to working along side many of you as volunteers and leaders throughout the coming years.



Steven Keiles, MS, CGC
NSGC President 2009

Cure CMD: A Non-Profit Agency Bringing the Congenital Muscular Dystrophies into Focus

By Ann Rutkowski, MD

I am an emergency medicine physician in Los Angeles. Last summer, I partnered with two parents to launch Cure CMD, a non-profit agency dedicated to helping those with congenital muscular dystrophy (CMD), their families, and those who care for them.

We have made exciting progress since Cure CMD's inception. The CMDs have historically been a largely ignored group of congenital muscle diseases. Cure CMD's mission is to promote research, treatments and in the future a cure for CMD. We will be targeting research funding towards translational medical therapies to slow disease progression in the CMDs. Cure CMD supports education and disease awareness for patients, families, medical providers, genetic counselors, and scientists.

The CMDs were first described in 1909 by Batten. They are characterized by infant and childhood onset of hypotonia and weakness, followed by progressive muscular decline over decades, often complicated by scoliosis, contractures and respiratory failure requiring ventilatory support. Each CMD subtype displays a phenotypic spectrum that spans early to late-onset with variable prognosis and disease severity.

The identification of ten new CMD genes in the last decade has helped drive new, more accurate classification schemes and understanding of underlying disease mechanisms. It has become possible to identify affected individuals based upon phenotypic presentation, and, through careful correlation of history with phenotypic clues, proceed through a logical algorithm to allow molecular confirmation of disease. Prior to gene discovery, CMD was classified as "merosin negative" or "merosin positive" based upon muscle biopsy immunostaining for merosin (or laminin). While "merosin negative" CMD is an accurate diagnosis; those with a label of "merosin positive" CMD need to be re-examined for phenotypic clues to allow for molecular confirmation and a precise diagnosis. "Merosin positive" CMD no longer is a complete or accurate diagnosis.

The last five years have seen an explosion in understanding of the underlying pathophysiology with the identification of new CMD genes. Cure CMD has been able to capitalize upon forward momentum with several initiatives, such as:

- Working with TREAT-NMD, the European neuromuscular initiative, to build international consensus around the CMD International Patient Registry (CMDIR), scheduled to be launched June 2009.
    - The CMDIR will be accessed through a Cure CMD portal, curated by a genetic counselor at Emory University. Each CMD patient with genetic confirmation of disease will be directed to update his or her profile on an international locus-specific database that is gene specific, with an expanded question set to track natural disease

progression and outcome. The purpose of the CMDIR is to connect people with confirmed CMD genetic mutations to future clinical trials and maintain the truly "undiagnosed" CMD cohort in a consolidated group for future genetic testing options as they become available.

- Developing a CMD diagnostic algorithm with international consensus
- Supporting CMD high-throughput genetic screening using microarray technology, currently being validated by Dr. Madhuri Hegde at Emory University
- Organizing the first "CMD Affected Person and Family Conference" highlighting education, awareness and networking for families.
    - The next conference is August 15-16, 2009 at Children's Hospital of Philadelphia (see www.curecmd.org for details)
- Organizing the first scientific conference, "Therapeutic Targets in CMD", July 9-11, 2009 at Emory University in Atlanta (see www.curecmd.org for details)
- Updating CMD online genetic resources, such as GeneTests (www.genetests.org), WikiGenetics (www.wikigenetics.org), National Organization for Rare Disorders (www.rarediseases.org), and Orphanet (www.orpha.net)

For more information, please visit our website: www.curecmd.org

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Natalie Weissberger Paul Award Acceptance Speech

Editors' Note: *Nancy Callanan was the 2008 winner of the Natalie Weissberger Paul award for lifetime achievement. Although she was unable to attend the 2008 Annual Education Conference to accept the award in person, Nancy's acceptance speech was delivered by a colleague and lauded by many as one of the conference highlights. Nancy has graciously allowed her speech to be printed in this issue of Perspectives, and we hope readers will take a moment to reflect upon these enlightening words of wisdom from an exceptional genetic counselor.*

By Nancy Callanan, MS, CGC

If you have never read Audrey Heimler's article about the history of the National Society of Genetic Counselors, published in the *Journal of Genetic Counseling* in 1997, you should, as this will give you some perspective on how a group of **visionary leaders** formed this Society in 1979, just 8 years after the first group of Masters-level genetic counselors graduated, and thus established genetic counselors as a distinct and autonomous professional group. I recently re-read this article, and once again recognize the huge debt of gratitude that all of us here today owe the many incredible leaders who worked diligently, thoughtfully, and strategically to accomplish this goal. Several of these early leaders, along with others who provided incredible leadership for NSGC in the years that followed have been recognized and honored by receiving the Natalie Weissberger Paul award. To know that I have been honored in the same way is truly overwhelming and humbling. I am grateful to my colleagues who nominated me for this award. It is a tremendous honor. Thank you.

When I entered the Sarah Lawrence College program in 1975, I truly had no idea where genetic counseling would take me. Intrigued by a profession that would combine my background in biology with my innate interest in helping people through difficult times, I jumped in with both feet. In those days, I did not perceive myself as a risk-taker, but here I was, choosing a very young and un-tested profession, taking a gamble on one of the most important life decisions – career choice. I can vividly recall driving up to Connecticut two years later to start my first job, not sure what to expect, or what would be expected of me, and thinking that I really did not know much of anything important, and it was just a matter of time before everyone found out! And here I am, **a mere 31 years later**, being recognized for my contributions to the profession and our professional society. What a journey this has been: a journey with many challenges, lessons, negotiations, and celebrations - a journey during which I had the privilege to serve patients, to collaborate with colleagues, and to train and mentor students; and a journey during which, by working as a volunteer and in leadership positions with NSGC, ABGC, and other organizations, I was able to develop the perspective and skills to make contributions to the growth of our profession. Along the way, I have

worked with and formed lasting friendships with many wonderful and accomplished genetic counselors and my life has been enriched by each of these friendships.

I must say, and this is important, that in my early years as a genetic counselor, the newly formed NSGC provided many opportunities for people that wanted to be involved. I still tell my students that all you need to do is show up, roll up your sleeves and start working, and soon you will find yourself engaged in useful, important and fulfilling work for our profession and our professional society. I hope that this is still true. It really needs to be. It is crucial for NSGC members to **stay informed** – this is really the first step. This means taking the time to read the membership updates, articles in Perspectives and The Journal of Genetic Counseling, and being aware of and responding to emerging issues and initiatives. NSGC is our professional voice and it is important for every member to be a part of forming and delivering our messages. When I was President of NSGC, I challenged all NSGC members to actively seek out ways, both large and small, to contribute to the important initiatives and activities of NSGC. Genetic counselors should view this not only as an opportunity, but also as a responsibility. Leadership at all levels is crucial for the growth and sustainability of our profession. As a volunteer-leader, you can have an important role in setting, and driving, the vision for the profession. As I have experienced personally, this can be a source of tremendous personal and professional growth.

Recently, I was asked to write a chapter about leadership for an advanced genetic counseling text that will be published in the near future. This provided me with an opportunity to reflect on some of the lessons I've been lucky to learn over the course of this challenging and rewarding career. At the end of the chapter, I list 10 things that I have learned about leadership, and I'd like to share a few of these with you now.

1. **Leadership is not about position.** You do not need to be granted a specific position or title in order to exercise leadership. In fact, demonstrating leadership in your daily work environment will result in opportunities for growth in terms of both influence and position.
2. **Genetic counselors have many of the skills that are commonly associated with leadership.** There is a good fit between the qualities and skills often associated with effective leadership and those associated with genetic counseling. Some of these include: critical thinking skills, intelligence, ability to perform complex tasks and commitment to life-long learning; communication skills; empathy and emotional intelligence; and problem-solving and decision-making skills.
3. **You are responsible for your own career satisfaction and success.** Just showing up each day and completing all the tasks assigned to you does not automatically put you in a position to advance. If you want increased professional recognition, advancement, reward, or satisfaction, you must seek opportunities to demonstrate your leadership skills. This is the difference between viewing genetic counseling as a job or as a career. If you are offered an opportunity, take it, even if it is not associated with a new title or increase in salary. A common mistake is to stick too close to your job description. If you do this, it is likely that you will miss great opportunities for professional and personal growth as well as career advancement.
4. **One important aspect of leadership is developing leadership in others.** Good leaders facilitate, promote and celebrate the success of others. By modeling leadership skills, sharing what you know, inspiring and collaborating with others you can accomplish great things. Leadership involves *incremental influence*. The more people that you engage in the process, the greater your influence.

Of course, no achievement happens in a vacuum, and what would an award acceptance speech be without a long list of recognizing and thanking people that have contributed to your success? So, here goes. I am grateful to:

- My parents, for encouraging and supporting me through college, graduate school and well beyond. As immigrants to this country, they poured their heart and soul into ensuring that their children would have opportunities that they never had, selflessly making daily sacrifices so that my siblings and I could get the education necessary to be successful. They clearly communicated their values of hard work, integrity and commitment to their community.
- My husband, Roger, and my wonderful children Angela and Brandy. They make me whole. Roger is the best husband and partner on earth. I used to tell people that my children made me smile everyday, and they still do.
- Joan Marks, for believing in me and admitting me to the Sarah Lawrence Program, and who inspired me by her example to actively contribute to the profession.
- My patients, from whom I learned the real purpose for this profession, which has provided the context for all of my professional activities and accomplishments.
- My work colleagues, past and present. I have been fortunate to work closely with many wonderful people over the years.
- The NSGC and ABGC for providing so many opportunities to contribute in meaningful ways to the growth of our profession.
- All of the incredible genetic counselors with whom I have shared this journey. Knowing you and working with you has enriched my life in many ways.
- My students, who, by their energy, enthusiasm and dedication to the cooperative learning environment we strive to achieve in our program, keep me open, and optimistic about the future of this profession that I love.

It seems ironic that, for the first time ever, I was not able to attend the NSGC meeting this year to accept this award in person. As those of you who know me well understand, I have placed a high priority over the years in balancing my professional interests and activities with my devotion to my husband and family. I believe that it is possible to have both a fulfilling personal life as well as a fulfilling professional life. But sometimes, you just need to choose, and this was one of those times. Some of you know that I have spent the past several months focused on supporting my daughter, Angela, as she simultaneously plans her wedding while undergoing treatment for cancer. Talk about a challenge that is at the same time an opportunity for tremendous growth. I am daily impressed by her strength and optimism and by the incredible support she has received from her fiancé, James and his entire family, her brother, Brandy, her family, friends, and co-workers. I am also grateful for the support that I have received from my friends, students, and colleagues. I dedicate this award to her future health and happiness.

Personal Accolades and SIG Funding Awards

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

Editors' Note: *Perspectives strives to be a method of communication for all types of news between members of the NSGC. As such, we felt it appropriate to include noteworthy personal professional achievements in this issue. Please feel free to contact the Editors about any submissions you may wish us to consider in the future.*

Congratulations to the following NSGC members for their recent personal achievements:

On April 7, 2009 **Robin Bennett**, Senior Genetic Counselor and Co-Director of the University of Washington Medical Genetics Clinic was awarded an honorary Doctor of Science from Kenyon College in Gambier, Ohio, her alma mater, for her work as an author, educator and geneticist. She was saluted as a "pioneer in the field of genetic counseling and as a scientist motivated by the desire to serve all humanity."

In March 2009, The *Reno Gazette-Journal* recognized regional Entrepreneurs of the Year for 2008. **Robbin Palmer** was recognized as a runner-up in this contest for her business, Northern Nevada Genetic Counseling.

Congratulations to the following NSGC members who received SIG funding over the last year:

Julia Barone: "Postnatal outcome of mild isolated ventriculomegaly detected on prenatal ultrasound" - $725 from the Prenatal SIG

Erin Carney: "Pediatricians' Knowledge of and Attitudes towards the New York State Newborn Screening Program" - $200 from the Public Health SIG

Martha Dudek: "Retrospective review of individuals prenatally diagnosed with mosaic trisomy 16 with a focus on the natural history of the disorder and ascertained through support group members" - $725 from the Prenatal SIG

Pinchia Huang: "Implications of false-positive trisomy 18 or 21 screening results in predicting adverse pregnancy outcomes" - $385 from the Prenatal SIG

Angela Inglis: "Views of parents of individuals with Down syndrome: media reporting, research directions, and prenatal testing" - $657 from the Prenatal SIG

Denise Lautenbach: "Perceptions of severe mental illness compared to other common complex disorders among family members of affected individuals" - $200 from the Psychiatric SIG

Brandie Leach *et al*.: "Evaluation of Efficiency and Reimbursement for Genetic Counseling Services in Traditional and Novel Service Delivery Models" - $2,000 from the Cancer SIG

Amber M. Mathiesen: "Parents of Children with Structural Birth Defects in Utah: Their Needs, Recommendations, and Preferences for Services and Support" - $200 from the Public Health SIG

Candace Peterson *et al*.: "Patient's Perspectives Regarding Written Communication Tool Following Genetic Test Results" - $1,889 from the Cancer SIG

Sarah Pirzadeh *et al*.: “Quick Cancer Genetics Education for Healthcare Providers” - $471 from the Cancer SIG

Deborah Wham *et al*.: “Assessment of Current Clinical Practices and Time Allocation Amongst Cancer Genetic Counselors” - $140 from the Cancer SIG

Licensure / Billing and Reimbursement Issues

The Impact of Genetic Counselor Licensure in Illinois

By Judith Miller, MS, CGC

Licensure for genetic counselors became available in Illinois (IL) in January 2008. Since many genetic counselors working towards licensure in other states are interested in knowing how licensure will impact their practices, this article describes the experience of IL genetic counselors since licensure became mandatory.

Provisions of the Illinois Licensure Act

In IL, licensure bills that will regulate a medical field are scrutinized by two very powerful groups: the Illinois Medical Society (IMS) and the Division of Professional Regulation (DPR). Both groups made changes to our proposed bill.

Our Act provides both title and practice protection. It contains two major practice restrictions:

1. Genetic counselors cannot provide genetic counseling services without a referral from a physician, advanced practice nurse, or physician assistant and the referring individual shall be provided a written report of the services
2. Genetic testing shall be ordered by a physician

These two issues are often contentious when passing licensure legislation for non-physician providers. Most licensure bills will only be passed with the support of the state medical society, and most state medical societies will require a referral. Many mid-level providers and specialists require a referral. The NSGC believes that inclusion of a referral requirement is one area in which we may need to compromise.

A temporary license is available for those who are qualified except for certification. Temporary licensees are required to practice under the supervision of a qualified supervisor (a licensed genetic counselor or a physician); a bi-monthly meeting with the supervisor for case review is required. The Rules for temporary licensure are very strict: a holder must take the first available certification exam, and the temporary license shall automatically expire upon notification that the licensee failed the exam, has been issued a license, or 24 months have passed.

The Act does not provide for a Genetic Counselor Board; the DPR functions as a Board.

Tracking the Number of Genetic Counselors in Illinois

Before licensure (December 2007), there were an estimated 94 genetic counselors in IL; our current list has about 100 names, but not all are employed in IL or see patients.

As of April 2009, 15 months after implementation, 91 licenses have been issued (15 are temporary). At least six licensees are not currently practicing in IL. Two licensees work primarily in Missouri and occasionally in IL. At least five licensees work for commercial laboratories. Three licensees are employed by a company that provides genetic counseling on the phone; these genetic counselors reside in another state but are making sure that they can legally provide genetic counseling to

residents of IL. In addition, at least three recently hired genetic counselors are now in the process of obtaining a license.

Survey on the Impact of Licensure

In April 2009, the Licensure Committee of the Genetic Task Force of IL sent an email survey to all genetic counselors licensed in IL to determine the impact of licensure. Thirty-five of the estimated 79 (44%) licensed individuals who see (or have seen) patients responded. Therefore, the following information is based on a limited number of respondents and may not reflect the opinions and experiences of all genetic counselors in the state.

Employment

Prior to licensure, we believed that the availability of licensure and billing ability might encourage employers to create more positions. Survey respondents reported four recently created positions; at least two were created because the employer knew about (pending) licensure and the ability to bill. We are aware of only one genetic counselor in the state with an independent practice (as a consultant for a physician group) and at least one survey respondent employed by a hospital does not have a physician supervisor.

Respect from the medical community

We theorized that genetic counselors would have "increased professionalism" and increased respect from medical colleagues. Unfortunately, no difference was reported.

Billing and reimbursement issues

Of the respondents now seeing patients, more than one third (12/33) bill for their services; seven of these did not bill prior to licensure. However, the 96040 billing code became available in 2008, so we are unsure if the increased billing is due to licensure, the new billing code, or both. Several reported plans to bill in the future.

It appears that billing is associated with the employment setting – specifically, if the genetic counselor works closely with a physician. Genetic counselors who do not bill typically work with physicians whose reimbursement is much greater than the genetic counselor's would be. Genetic counselors that bill reported using the 96040 billing code or a facility fee. At least one genetic counselor is now credentialed by her hospital and most of the insurance companies with which her practice deals. Others reported that they have requested credentialing or a provider ID from their institution, but the requests have not (yet) been granted.

Requirement for referral

The majority of respondents (70%) did not think this requirement caused major problems for themselves or their patients. Groups in which a physician bills under a consultation code already obtain referrals. Other genetic counselors comply by arranging for a referral, but noted this caused appointment delays and extra work. Occasionally, patients who wished to self-refer ended up not being seen. Several respondents felt that the referral restriction has limited their ability to expand their practices; when potential patients are associated with physicians practicing in a variety of areas and locales, obtaining referrals is difficult, time-consuming and problematic. Several genetic counselors felt strongly that the referral requirement has reduced access to genetic services. One respondent acknowledged that the referral and written report requirements have provided

opportunities to connect patients to a primary care physician and keep the referring physicians informed.

Prohibition on ordering genetic tests

Most respondents (87%) have always had a physician co-sign for tests and therefore have not changed their practice since licensure. The vast majority, however, stated that this prohibition is an inconvenience and causes difficulty and delay. Many felt strongly that the prohibition makes no sense for a multitude of reasons, including that professionals less knowledgeable about genetics can order tests and that patients are often referred specifically for the purpose of testing. Again, this restriction was more of a problem for genetic counselors not working closely with a physician. One genetic counselor who works independently reported cases in which referring physicians, knowing they would have to order genetic tests, found it easier to simply order testing themselves and not refer the patient for genetic counseling.

Temporary licenses

In order to apply for a license, new graduates need to obtain proof of education plus verify Active Candidate Status (ACS) for the certification exam, which has meant that some counselors are employed for a period before obtaining their license. Surprisingly, several reported that they practiced anyway. None of those with a temporary license reported a problem with the requirement for a qualified supervisor and case review.

All, however, reported a concern with the strict Rules for a temporary license described earlier. The rules for the certification exam have recently changed. However, these changes will not mitigate the problems with the high standards in the IL Licensure Rules.

Violations

The majority of genetic counselors were not aware of any violations of the Act. One person, not a genetic counselor, is known to have stopped using that title. Several respondents reported knowing about probable violations of the title or practice restrictions, but they did not wish to report this to the DPR. The Genetic Task Force of IL wishes to support genetic counselors, and in this role is willing to write a friendly, educational letter to violators. No one reported awareness of a genetic counselor acting in a way that would be grounds for discipline.

Conclusions

Surprisingly, many genetic counselors reported that licensure has had little or no impact on their practices.

One positive impact of licensure is that several jobs have been created because of the increased ability to bill. At least seven genetic counselors that were not previously billing are now doing so. It is difficult to determine the impact of licensure on reimbursement rates.

Survey respondents did report concerns. Several recently-hired genetic counselors stated they were unaware of many provisions of the Licensure Act, and think their facilities also do not understand the provisions. In addition, genetic counselors have asked for help with billing and increasing reimbursement. Clearly, continuing education to genetic counselors and their employers is needed.

A major concern regards the strict Rules for temporary licensure that were written by the DPR, in spite of our strenuous objections. The concern is that new graduates may not be willing to take a job in IL due to these rigorous Rules.

For genetic counselors not working with a physician, the Act's practice restrictions may result in reduced access to genetic services, and may make it much more difficult for a genetic counselor to practice in some situations.

Looking to the future, the survey results suggest that we need to think about the settings in which genetic counselors should be able to practice. The number of geneticists may be decreasing at the same time that the number of patients and physicians aware of the need for genetic services is increasing. The NSGC and those working on Licensure Acts should strategize how to best promote and support jobs in which genetic counselors work fairly independently. This is essential if we wish to increase access to quality genetic services.

The results of our survey of licensed genetic counselors in IL have illuminated several areas of concern in our Act and Rules. We may need to amend provisions of our Act, and it clearly is important to try to change the temporary licensure Rules.

The IL Genetic Counselor Licensure Act and Rules can be viewed on the Illinois Department of Finance and Professional Regulation website, http://www.idfpr.com/DPR/. Click on "genetic counselor" in the drop down box.

SIG Speak

A "Perspective" on Thrombophilia

By Kristin Paulyson Nuñez, MS, CGC and Elizabeth Varga, MS, CGC, Co-Chairs, Hematology SIG

Testing for thrombophilia has increased significantly over the years. The primary indications for testing include a history of clotting in the veins, known family history of thrombophilia, poor pregnancy outcome, or recurrent pregnancy loss. While many clinicians understand the risks, benefits and limitations of testing and attempt to convey these to the patient, others do not. Many clinicians may be unfamiliar with the significance of a patient's result(s) or, more importantly, how to integrate the information into the patient's health care management. As a result, patients are being referred "after the fact" to a genetic counselor to assist the clinician in the individual's counseling or management. NSGC listserv postings, as well as personal communications with colleagues, support an increasing need for information on the relevance of certain tests and their significance to the patient. In this article we, as two genetic counselors experienced in the area of thrombophilia, will share our perspectives on testing and counseling for thrombophilia in various contexts.

Overview

Thrombophilias are conditions that increase an individual's risk to clot. They may be acquired or inherited. Most inherited thrombophilias result in venous-side clotting as opposed to the arterial side. Most clotting will occur in the presence of several risk factors. These can include, but are not limited to, inherited thrombophilia, surgery, immobilization, malignancy, diabetes mellitus, obesity, smoking, estrogen-containing oral contraceptives (possibly hormone replacement therapy), and pregnancy. Individuals with a known inherited thrombophilia have a higher risk to clot as compared to the general population. For example, the presence of one copy of the factor V Leiden mutation, which occurs in about 1/20 Caucasians, results in approximately a seven-fold risk to clot. Those who are homozygous for the factor V Leiden mutation have approximately an eighty-fold risk to clot.

Thrombophilia Testing by Patient Request

In the primary care setting, patients may request testing because a family member has been diagnosed with an inherited thrombophilia. In the majority of these situations, the patient has no personal history of thrombosis but is concerned about his or her chance to have the same condition as a family member. The clinician may indulge this request without fully considering the clinical indication or informing the patient about benefits and limitations of testing. Furthermore, patients may not understand or appreciate whether changes will occur in their medical management once results are known. In the event of a positive test result, the patient may be referred for genetic counseling.

How is the genetic counselor to approach this situation? We advise the genetic counselor to begin by obtaining a detailed pedigree and medical history of the patient. It is helpful to know if the patient has had a complete thrombophilia workup, a partial workup or was only tested for the condition identified within the family. Medical records are essential as laboratories values and methodologies vary from center to center. The genetic counselor should be particularly interested in asking questions about health behaviors, including known risk factors like smoking, obesity, and oral contraceptive use.

Along with discussing the inheritance and risks associated with being a carrier for an inherited thrombophilia, the genetic counselor can also discuss identified risk factors and educate the patient about healthy lifestyle changes. For example, an individual with an inherited thrombophilia who smokes may be more inclined to be referred to a smoking cessation program. Similarly, a young woman on oral contraceptives with an inherited thrombophilia may benefit from a referral to her OB/GYN for an alternate form of birth control. Ideally, the patient should be provided with a referral to a clinician who specializes in thrombophilia or who can assist with a particular lifestyle intervention.

Many families may have questions about testing young children (minors). In our experience, testing young and asymptomatic children is not indicated as it would not typically affect medical management for this age group. We encourage parents to be educated about their child's risk and any risk factors they may encounter during their lifetime. In situations of trauma or surgery, we recommend that the child's clinician be informed of the family history. As with any condition occurring within a family, we encourage the parent to identify the right time in their child's life when they feel he or she is emotionally capable of understanding the information and the impact on his or her life. Appropriately timed genetic counseling is always encouraged prior to consideration of genetic testing.

Thrombophilia Testing in the Context of Obstetrics-Gynecology (OB/GYN)

Many patients in this context present with a history of recurrent pregnancy loss, poor pregnancy outcome (fetal growth restriction, stillbirth, pre-eclampsia) or post-partum clotting (specifically deep vein thrombosis or pulmonary embolism). Depending upon the patient's concerns, a genetic counseling session in this setting can assist the patient with the understanding of past events, provide medical management for future pregnancies, or both. Often, a family history will include extended family members with histories of pregnancy loss, poor pregnancy outcome or spontaneous clotting. In the case of an acquired thrombophilia, there may be no relevant family history beyond the immediate couple's history of loss or poor pregnancy outcome.

In addition to traditional testing options, couples with the above indications are typically offered screening for the factor V Leiden (FVL) mutation, antithrombin III deficiency, functional protein C deficiency, functional protein S deficiency, the Prothrombin G20210A gene mutation, and antiphospholipid antibodies (including anticardiolipin antibodies, lupus anticoagulant and, more recently, anti-beta 2-glycoprotein-I).

Testing for thrombophilia during pregnancy can be tricky because pregnancy typically naturally decreases protein S and protein C function overall. It is important for a clinician to be cautious in his/her interpretation when testing during pregnancy or in the post-partum period for this reason. As well, the laboratory may benefit from knowing whether a patient is pregnant or was recently pregnant. It is also important to be familiar with one's laboratory and normative values. Often, repeating the protein C or protein S testing after the 12-week postpartum period or following a loss will help to determine whether the patient truly has protein C or protein S deficiency.

Patients identified as FVL or Prothrombin G20210A mutation heterozygotes, compound heterozygotes or homozygotes are generally offered prophylactic anticoagulation during pregnancy, serial ultrasounds for fetal growth assessment, and non-stress testing as early as 32 weeks gestation. After pregnancy, they may be offered non-estrogen containing birth control options such as Micronor, Depo-Provera, IUD, condoms, and diaphragms.

Couples with a history of recurrent pregnancy loss or poor pregnancy outcomes may, naturally, be concerned about preventing a recurrence. They may become so focused on their pregnancy that they

may overlook the genetic risk for their child to also have a thrombophilia. Our experience indicates that these patients will often present to their child's pediatrician later, anxiously requesting testing. In order to reduce this anxiety and the number of unnecessary tests, we encourage genetic counselors to discuss risks to offspring within the antenatal period.

The Best Situation of All: The Opportunity to Provide Pre-Test Genetic Counseling

If a patient has not yet been tested for thrombophilia but presents with a known diagnosis of an inherited thrombophilia in the family, the genetic counselor has a unique and valuable opportunity to properly inform the patient about the risks, benefits, and limitations of testing. In many situations, patients are looking for guidance and have come because a concerned family member has encouraged them to seek testing. Many patients want to know what they can do to reduce their risk to develop a clot. Ultimately, recommended lifestyle changes are often in the best interests of the patient's overall well-being, not just in the context of thrombophilia.

Sometimes, educating the patient and not pursuing testing may be his or her best option. Individuals who are otherwise healthy and have no identified risk factors should be educated regarding the signs and symptoms of a clot, but should not expect much to change in their health management. However, if the patient does pursue testing and is found to have a thrombotic condition, a referral to a clinician who specializes in thrombophilia is critical.

Some Final Comments: MTHFR Polymorphisms, Arterial Thrombosis and More

Methylenetetrahydrofolate reductase (MTHFR) polymorphisms have been linked to several genetic conditions. The incidence of carrying one of the two most common polymorphisms is, in some populations, as high as about 1 in 3. Some MTHFR polymorphisms (particularly the MTHFR C677T polymorphism) have been associated with thrombophilia because of their involvement in the homocysteine pathway. As such, testing for MTHFR polymorphisms was previously considered relevant/significant in the evaluation of thrombophilia.

However, data now suggest that MTHFR polymorphisms should <u>not</u> be included in a thrombophilia workup. Regardless of personal medical history, the presence of other inherited or acquired thrombophilias, or pregnancy complications, genetic testing for MTHFR polymorphisms is not clinically warranted. Individuals identified with MTHFR polymorphisms should be reassured that their risk to clot is <u>not</u> significantly increased above the general population. Furthermore, anticoagulation should not be offered in these situations. Likewise, for the patient who presents with a personal history of clotting and has a known inherited or acquired thrombophilia <u>and</u> a MTHFR polymorphism, the risk to develop a future clot is <u>not</u> thought to be further increased based on the presence of the MTHFR polymorphism. In our anecdotal clinical experience, there no longer appears to be any benefit in testing for MTHFR polymorphisms.

Previously, fasting homocysteine levels were recommended as a preferred method to assess thrombotic risk. While significant elevations (e.g. > 50 μM) in homocysteine are a strong risk factor for aterial and venous blood clots, recent data suggest moderate elevations (12-50 μM) pose only a moderate risk and treatment (with folate, vitamins B6 and B12) is not of proven benefit. Therefore, many clinicians with expertise in thrombophilia are moving away from routine screening of fasting homocysteine levels.

Random screening of the general population for the factor V Leiden mutation is not recommended. In 2006, The American College of Medical Genetics revised its Practice Guideline to state, "Routine testing [for the factor V Leiden mutation] is not recommended for patients with a personal or family history of arterial thrombotic disorders (e.g., acute coronary syndromes or stroke) except

for the special situation of myocardial infarction in young female smokers. Testing may be worthwhile for young patients (<50 years of age) who develop acute arterial thrombosis in the absence of other risk factors for atherosclerotic arterial occlusive disease." In reality, an eager-to-test patient may seek out whomever they can who will order the test, despite a clinical indication or medical recommendation. Unfortunately, many primary care clinicians do not know that the ACMG's Practice Guideline exists. Therefore, we suggest that genetic counselors provide education and always explore a patient's reasons or motivation for testing.

To Learn More

In 2008, the NSGC's Hematology SIG reviewed published medical literature and developed a spreadsheet containing a list of references that would provide genetic counselors with a solid foundation in the area of the thrombophilias and hemoglobinopathies. We encourage you to review this document (found at www.nsgc.org under the Hematology SIG). We have also found the following websites to be useful for patients and clinicians:

http://www.fvleiden.org
http://www.stoptheclot.org/
http://www.marchofdimes.com/professionals/14332_9264.asp
http://www.hemophilia.org/bdi/Thrombo_article.pdf
http://www.cdc.gov/ncbddd/hbd/clotting.htm
http://www.acmg.net/StaticContent/StaticPages/Factor_V.pdf

In addition to these resources, the Hematology SIG members are always available to take any questions you may have about your clinical situation.

NSGC News

NSGC Governance Evaluation Task Force: Report to NSGC Membership on 2008 Evaluation Activity and Results

By the NSGC Executive Office

The NSGC Board of Directors appointed a Governance Evaluation Task Force in January 2008 to (1) develop a framework for ongoing evaluation of NSGC's governance, and (2) conduct an initial evaluation of the changes during the first full year under the new governance structure. The Governance Evaluation Task Force was comprised of **Cathy Wicklund** (Chair) **Luba Djurdjinovic, Tene Franklin Hamilton, Steven Keiles, Beth Leeth, Peter Levonian, Rob Pilarski, Angela Trepanier**, and **Vickie Venne**. Evaluation of the governance changes and the transition from the previous structure to the new structure is an ongoing process. Evaluation using the framework developed in this initial year will need to continue over several years to assess the full effect of the changes.

Preliminary evaluation focused on the full implementation of the changes and member participation in the new structure. Additionally, Board members, Committee Chairs and Committee members were surveyed about their perception of the effectiveness and value of their volunteer experience.

Longer term evaluation will include membership surveys to assess:

- the effect of the governance changes on the membership
- the success of the governance structure in providing the appropriate Committee and Board structure to support the NSGC's strategic initiatives, and
- the development of a robust leadership pipeline to maintain the NSGC's many initiatives, member services, volunteer opportunities and projects

Summary of Recommendations

The Governance Evaluation Task Force has developed and implemented a framework for evaluation of the NSGC governance changes approved in 2007 and implemented in 2008. This framework will be replicated for use on an annual basis to monitor the success of the governance changes and to assess when adjustments are recommended. With this structure in place, responsibility for the ongoing evaluation activity will transfer to the Nominating/Governance Committee for 2009 and moving forward. This will ensure that this activity is an annual charge of a permanent Committee of the NSGC.

Following are the details about the evaluation mechanism developed for each component of the governance changes and the results of the evaluation for the 2008 year. The activities included in the governance evaluation process are summarized in Table 1.

<u>Evaluation Communication Plan</u>

Communication of the evaluation plan to the membership began on an ongoing basis in early 2008, and occurred through various NSGC communication vehicles, including:

- a governance article in Perspectives to provide ongoing reporting on the implementation process and present available quantitative data from the evaluation process
- ongoing communication via the "President's Beat"
- eblasts specific to the nominations process

In addition, this final report from the Governance Task Force, developed at the close of the 2008 process, shares with the membership the success of the process (including information from the Nominating Committee Report) and the challenges identified through our evaluation efforts.

Evaluation Process

Although not limited, the evaluation process focused primarily on those areas that were most affected by the governance changes. The primary areas of evaluation and items considered during the evaluation process are detailed below.

Committee Chair and Committee Member Evaluation

Committees' progress toward their annual tasks and charges should be monitored. The following questions were considered:

- Was the Committee able to complete their specified tasks within a given year? If not, why?
- Did the Committee follow its particular charges? If not, why?
- If Committee charges were not able to be implemented as envisioned, how could charges be reframed in 2009 to make them more appropriate or feasible?
- Were the expectations/tasks/charges from the Board of the Directors clear?
- Were the Committee Chairs clear on the structure of the Committee and how to utilize their Committee members and the volunteer database?
- Is communication happening within and outside of the Committee?
- Did the Committee utilize the volunteer database to identify additional members or members of work groups?
- What has been the role of the Board Liaison? Of staff? Are the responsibilities of everyone clear?
- Are Committee Chairs engaging their Committee members and building a leadership pipeline?
- Is communication effective and reciprocal between the Committees and the Board?

Anonymous web surveys were distributed to the NSGC Committee Chairs and Committee members in late December 2008. While there was some overlap between surveys, there were questions specific for each group.

The Committee Chair evaluation identified two major areas for improvement. First, Chairs indicated a lack of clarity regarding how to use Committee members to accomplish their Committee charges. Recommendations for the future include:

- clearer communications regarding how to staff Committees to ensure charges can be accomplished
- periodic check-ins by the Board Liaison to provide any support or assistance
- more education on use of Committee members and additional volunteers to ensure adequate volunteer involvement

In addition, annual and ongoing leadership training will continue to address this topic.

Second, Committee Chairs reported varying effectiveness among the Board Liaisons in facilitating communication with the NSGC Board. This is another area where expectations and roles need to be further clarified, communicated and reinforced. Ongoing training and skill development regarding the expectations of the Board/Board Liaison/Committee Chair relationship will continue to enhance the effectiveness of this communication channel.

Based on the Committee member evaluation results, clear communication of expectations by Committee Chairs to Committee members was identified as an area for improvement. It should be noted that this may be related to the lack of clarity by Committee Chairs regarding how to best utilize Committee members. Another area for improvement identified in the Committee member evaluation was staff support for organizing Committee activities. It is recommended that staff and Committee roles be clarified at the outset of the year and that communication about expectations take place on an ongoing basis, to ensure that the needs of all involved are met.

Nominations Process Evaluation

The success of the Call for Nominations and Nominations Process was monitored and evaluated by the Governance Evaluation Task Force. The following questions were considered during this process:

- Was there significant member participation in the Call for Nominations process?
- How many people participated in the Call for Nominations process?
- Who nominated the candidates (such as general members, Nominating Committee, Board)?
- Were potential new leaders identified during the process (i.e. people who were not "on the radar" previously)?
- Were the nominated individuals willing to undergo further consideration by the Nominating Committee for either an elected leadership position or an alternate volunteer position?
- Was the Nominating Committee able to identify and slate candidates for the Board of Directors who met the current leadership qualities as desired by the Board?
- How did the candidates evaluate the process?
- Did the candidates have any outstanding questions regarding the nominations process?

The 2008 Nominating Committee submitted a full report to the Governance Evaluation Task Force. After review of the report, the Governance Evaluation Task Force concurred with all recommendations of the Nominating Committee and made the following recommendations for the 2009 nominations process:

- The nominations process should be outlined in detail for nominees at the outset to clarify the full process and set expectations. This outline should include all steps in the process and a detailed timeline.
- The interview questions asked of candidates should be modified, based on 2008 candidate feedback. The questions should be reviewed by NSGC members who are not on the Board or Nominating Committee to ensure that they are clear. The Membership Committee is a potential source for this review.
- Nominating Committee members should discuss interviewing techniques and receive interviewing tips prior to beginning candidate interviews. This will help ensure productive interviews and increase consistency among the interviewers.
- The Board should consider changing the terminology used in communications about the election to indicate that we are asking for "ratification" of the slate rather than "election" of the slate.
- Voting rates should be monitored over the next three to five years. If voting rates are determined to be low relative to rates in other professional organizations, this trend should

be assessed. If the uncontested slate system is determined to be a factor, adjustments to the slate election process should be considered.

Liaison Evaluation

Although the liaison process will be officially monitored through the liaison relationship assessment form, the Governance Evaluation Task Force also considered the following:

- Are the Liaison relationships meeting their desired values?
- Are the Liaisons reporting their activity adequately?
- Are the Liaison relationships tied to the NSGC's Strategic and/or Branding Plan(s)?
- Is the Board of Directors considering relationships that should be developed in the future, as well as those necessary for the present?
- Is adequate direction being given to the Liaisons by the Board to ensure the desired outcomes from the relationships?
- Which Liaison relationships do we want to strengthen, and which may no longer tie in to the NSGC's strategic initiatives?

The Board prioritized relationships with liaison organizations in 2008 and for 2009 based on how they support or enhance work being done toward the NSGC's strategic initiatives, increasing access to genetic counseling services, branding, provider education, and future vision efforts. Relationships were prioritized in order to strengthen the NSGC's relationship with each organization and to ensure that the interaction is taking place at the appropriate level. The Board reviewed all current and desired liaison relationships and prioritized these relationships into one of three levels to help ensure appropriate allocation of financial, volunteer and staff resources. These levels are outlined below:

- **Priority liaisons:** The primary relationship will occur at the Presidential level (President, President-Elect or Immediate Past President, with the exception of the Institute of Medicine as Past President Cathy Wicklund is the ongoing liaison), with continuity provided by the Executive Director and Government Relations Director and additional supporting relationships by volunteers who provide project-specific expertise.
- **Presidential liaisons:** Relationships important to maintain or enhance collaboration regarding promotion of genetics within healthcare and access to genetic counseling services. While usually primarily a President-to-President relationship, ongoing interaction and relationship development may also be performed by staff and volunteers working on specific projects.
- **Public Policy liaisons:** Liaison relationships that impact the NSGC's involvement in public policy efforts. Ongoing, consistent relationships can be maintained by official representation from a member of the Public Policy Committee, or by the current liaison with oversight from the Public Policy Committee.

SIG Evaluation

An additional goal of the evaluation process is to achieve better collaboration with the NSGC's Special Interest Groups (SIGs) in support of the NSGC's strategic initiatives. Questions considered by the Task Force are as follows:

- Are the activities of the SIGs aligned with the NSGC's Strategic Plan?
- Do the SIG leaders know the role of the SIG and how it fits within the NSGC?
- Are there clear and open lines of communication between the SIGs, the NSGC Board of Directors and the Executive Office Staff?

While no formal evaluation of the SIGs was conducted, informal feedback was monitored by the SIG Board Liaison throughout the year. Two conference calls were conducted with SIG Chairs to enhance communication, provide support and outline expectations. In addition, the President and staff communicated with SIGs regularly to involve them in the NSGC activities whenever applicable and tie their activities to the NSGC strategic initiatives. These efforts will be ongoing to further increase the level of communication between the SIGs and the NSGC Board and Committees. The Governance Evaluation Task Force recommends implementing an evaluation process with the SIG Chairs. This process should be defined further by the Nominating Committee during 2009 and utilized at the end of the year.

NSGC Board of Directors and Society as a Whole

The Task Force also recommended that the Board and the Society be monitored, especially for the following:

- success towards strategic objectives
- volunteers' satisfaction with their experience(s) (e.g., did volunteers feel that they were utilized appropriately?)

The above questions were not formally evaluated during 2008, as the changes needed to be fully implemented for one year prior to conducting this evaluation. Questions addressing the above topics will be included in the 2009 Membership Survey to provide all members the opportunity to share their feedback. The survey should be distributed to members in June.

Board of Directors Governance Evaluation

In October 2007, the NSGC Board of Directors began evaluating the Board's functioning during each in-person meeting. Overall, the results remain consistent between the October 2007 and February 2008 meetings. The results of the October 2008 meeting evaluation showed improvement over previous meetings in all areas evaluated. As the Board looks to strengthen its governance skills, it will work to develop the areas in which it scored lower. Realizing that governance is an important process that needs to be monitored, the Board will also seek to maintain those areas in which it scored high.

It is the recommendation of the Governance Evaluation Task Force that the Board add a second component to the Board evaluation process based on the new NSGC Board Culture Statement. By adding this component, the Board will maintain the current process to allow for ongoing assessment of progress, but will also be able to evaluate the Board culture and how this is mapping to the desired culture outlined in the Board Culture Statement. A Board Culture Evaluation has been developed by the Governance Evaluation Task Force and use of this evaluation was initiated at the February 2009 in-person NSGC Board meeting.

Table 1: 2008 Governance Evaluation Activities

- GETF = Governance Evaluation Task Force

Evaluation Task	**Responsible Group**	**Status**
Overall Governance		
Governance evaluation article in	Evaluation	Published Summer 2008

Perspectives	Task Force	
Board Culture Article in *Perspectives*	Board	Published Fall 2008
Use of Leader/Volunteer Database for Committees, Task Forces and projects	Evaluation Task Force/Staff	Accessed for 13 distinct projects/appointments in 2008; Available and in use by Committee Chairs for 2009 committee appointments
Committees		
Committee reports	Committee Chairs	Reports completed for 2008 in-person NSGC Board meetings
Evaluation of Board Liaison function	Board and GETF	Conference call with the Committee Chairs and Board Liaisons to discuss feedback; Need to evaluate further in 2009, and clarify the role of Board Liaison
Committee Chair Evaluations	Board and GETF	Evaluations distributed in late Fall 2008; results available for GETF evaluation in January 2009
Committee Member Evaluations	GETF	Evaluations distributed in late Fall 2008; results available for GETF evaluation in January 2009
Nominations		
Nominating Committee Report/Recommendations	Nominating Committee	Completed in December 2008 and submitted to GETF
Survey of candidates regarding process/experience	Nominating Committee	Completed in December 2008 and submitted to GETF
Liaisons		
Implement discussion with Liaisons to review agendas and prepare them for meetings	Board	Underway and ongoing - this has been taking place formally and informally in 2008
Assess the value of all current liaison relationships annually	Board	Assessment completed at October 2008 Board meeting
Discuss priority liaison relationships for 2009, including any new relationships	Board	2009 liaison priorities approved by the Board in October 2008
SIGs		
Bi-annual reporting for SIGs, to include relationship of SIG activities to the NSGC strategic plan	SIGs	Received end-of-year reports from SIGs
Ongoing communication with SIGs, Board Liaison, and Staff	Board/Staff	Two SIG Co-Chair calls held; One-on-one communication between SIG leaders, Board and Staff regarding specific issues and projects is ongoing

ABGC Update

New ABGC Mission Statement and Survey Results

By the ABGC Board of Directors

Recently, the American Board of Genetic Counseling (ABGC) surveyed genetic counselors for feedback on a variety of professional issues, such as how you feel about our proposed new mission statement, whether you are an ABGC diplomat, and, if not, why? We would like to present our new mission statement, which was fine-tuned based on your comments, and address some of your very helpful and enlightening survey feedback here.

The ABGC Board of Directors appreciates that the proposed new mission statement, as presented in the survey, was too lengthy. As such, we took your advice and significantly shortened it. The new and improved mission statement, which was unanimously approved by the Board, is as follows:

The American Board of Genetic Counseling establishes standards of competence through accreditation of graduate training programs and certification and recertification of genetic counselors to advance the profession and protect the public.

Many of you indicated that you were not happy with or did not understand why we included the phrase "protect the public" in the mission statement. As the ABGC certifies (and recertifies) genetic counselors, when a genetic counselor displays his/her Board certification to the public, it demonstrates that his/her qualifications represent a certain standard. This notifies the public (including employers) that this individual has met this standard and attests to a certain level of competence. This is what sets the certified genetic counselor apart from another, non-certified health professional that might otherwise choose to call him or herself a genetic counselor. The National Organization for Competency Assurance (NOCA), which provides accreditation to credentialing organizations, stresses that credentialing programs serve to (among other things):

- Protect the public
- Establish standards for professional knowledge, skills, and practice
- Assure consumers that professionals have met standards of practice
- Meet the requirements of governmental regulators

Protecting the public is the number one priority of NOCA, a nationally known and well-respected organization. Likewise, with licensure becoming increasingly important, the ABGC credential becomes critical because this is typically used by states in their legislation. "Protecting the public" is a pivotal issue to states when considering a piece of legislation such as licensure for health care professionals.

Licensure is the mandatory process by which a governmental agency grants time-limited permission to an individual to engage in a certain profession after verifying that he or she has met predetermined and standardized criteria, and offers title protection for those who meet the criteria. Certification is the voluntary process by which a non-governmental agency grants (usually) time-limited recognition and use of a credential to an individual after verifying that he or she has met predetermined and standardized criteria. So, while the ABGC grants certification and states grant licensure, both are concerned with protection of the public.

Finally, many of you asked what it means to be an ABGC "diplomate." It means that you have been Board certified by the ABGC (or by the American Board of Medical Genetics prior to the creation of ABGC, and you subsequently joined ABGC). Credentialing organizations like the ABGC are comprised of diplomates who have achieved and continue to maintain certification. Professional organizations like the National Society of Genetic Counselors are comprised of dues paying members. To remain an ABGC diplomate, you must recertify if you were certified after 1993; to remain in good standing, you must pay the Certification Maintenance Fee each year. ABGC diplomates who are in good standing are entitled to participate in committee activities, run for Board office, and collect Professional Activity Credits, which can be used for recertification.

As a Board, we remain available to our diplomates, all genetic counselors, and the public at large to answer any questions. For those of you who noted that the ABGC website could use some improvement, please know that we are actively updating it and will continually strive to provide our constituents with timely information in a user-friendly format. You can access the website at www.abgc.net.

We are always open to suggestions and happy to receive feedback. Please feel free to contact the ABGC Executive Office at 913-895-4617 or by e-mail at info@abgc.net.

Student Forum

When Learning is Uncomfortable: The Best lesson I Learned as a Student

By Jessica Cass, BA, University of Colorado

As genetic counseling graduate students, our training consists of challenging classes, diverse clinical settings and attempting to find a balance between the two. Beyond the integration of information and the opportunity to practice these new skills, there is a tight rope we must walk. This is the fine line between compassion and professionalism with which all of us in the helping profession are very familiar. I share the following story because it illustrates some of the unique challenges we face as genetic counseling graduate students. We are learning to be effective counselors in situations that, until our training, many of us have never faced.

The first patient I saw on my own, an 8-month-old little girl, died suddenly just two months after I met her in clinic. I was a first-year student about to start my second semester of graduate school when I received a voice message from my patient's mother, telling me of her daughter's unexpected death. As I listened to her message, I froze; my heart was pounding, my palms sweaty. All I could think was, "How could this be happening right now? I'm not ready to deal with a mother whose infant just died. Not yet." Here she was, reaching out to me for support, and I was afraid that I was going to let her down; that I wouldn't know what to say to her.

When I returned her phone call, somehow I found the words to say. I showed her compassion and empathy as she retold the story of finding her baby dead. Her daughter had suffocated in the middle of the night, wedged between the side of her crib and mattress. I offered her support and resources, and didn't stumble over my words or break into tears, which were my fears. After we hung up, however, the tears came. After reliving this unbelievably traumatic story with this distraught mother, I was unprepared for the process that happened inside of me. I was so concerned about what to say to her, I didn't stop to think about what I should have said to myself. At home that evening, I cried as I imagined what that morning must have been like for my patient's mother. My imagination, I'm sure, couldn't come close.

A week later, as I retold the story to my classmates and my psychosocial professor, the tears were still fresh. I wondered to myself, "Why is this affecting me so much?" I thought I was supposed to be professional and leave the bad news at the office. I wondered, "Will I ever be able to do this? Am I just too empathetic, too sensitive, and too emotional to be a genetic counselor?"

Fast-forward a year and a half. I now know that if it ever becomes easy for me to see my patients experience extreme grief, it will be time to take a break from my career. To aid in managing these tough situations, I have gathered some tools that will help me to be an effective genetic counselor and also take care of myself at the end of the day. Specifically, I now know how important it is to have people in my life to talk to about anything but genetics. Additionally, I focus on the positive aspects of my life and spend some extra time playing with my dog, treating myself to a latte and a good book or, I will admit it, watching mindless television.

In retrospect, I am thankful that I was faced with something so difficult early in my training. I learned an invaluable lesson that didn't come from any lectures or textbooks, but rather from experiencing this important moment in counseling. As I retell this story now, my eyes fill with tears and I get a lump in my throat, but I am able to take a deep breath and see the bigger picture – that I hope I never lose my sensitivity and ability to empathize with my patients.

These and other difficult situations may occur during the course of one's genetic counseling training. I have spoken both with students who have experienced similar difficulties and students who have worried about the opposite, that they wouldn't be able to show emotion when it was perhaps appropriate to do so. It can be particularly difficult to learn how to be a genetic counselor when attempting to master so many aspects of the role at once.

I have no doubt that I will face many more tough situations in my career, and that this is not the last patient's story I will take home with me. The difficult situations that terrified me in the beginning of my training are now the reasons why I love this profession. The constant challenge, the unexpected – these are the things that drive me. I hope that ten years from now I can remember what it was like to be a student, first negotiating that walk on the tight rope. I recognize that I will continue to maneuver this walk for many years to come. As students, we may think we are inexperienced or unable to handle emotionally challenging situations, but we often surprise ourselves.

The most important lesson I have learned during my graduate training is to trust my instincts. As students we may find ourselves preoccupied with following a textbook or imitating a supervisor's style in counseling. Too often, we ignore our own instincts and risk denying ourselves the personal growth that is necessary for us to become confident, effective genetic counselors. The lesson I learned and want to share with fellow trainees is this: don't doubt your capabilities to provide empathy and support to a person in need. Just remember to give yourself that same support after a challenging case as well.

Genetic Counselor Publications

By Deborah McDermott, MS, CGC

Featured Article

Bernhardt BA, Rushton CH, Carrese J, Pyeritz RE, Kolodner K, Geller G. Distress and burnout among genetic service providers. *Genetics in Medicine*. July 2009 (in press).

Barbara Bernhardt is a Clinical Associate Professor of Medicine in the Division of Medical Genetics at the Hospital of the University of Pennsylvania. She is involved in clinical genetic counseling for adults with Marfan syndrome, Hereditary Hemorrhagic Telangiectasia, and other disorders. She also serves as Co-Director of the Pennsylvania Center for the Integration of Genetic Healthcare Technologies, which is one of the six NIH-funded Centers of Excellence in ELSI research.

Thankfully, Barbara has a touch of Andy Hardy in her. For those of you not familiar with these Mickey Rooney and Judy Garland films, they're worth a Netflix rental. Not one to let her own feelings of burnout simply get the best of her, Barbara and her colleague Dr. Gail Geller, a bioethicist, saw a research opportunity. They applied for and were awarded an NHGRI grant focused on workforce issues in genetics in 2005. This funding has led to a number of publications focused on issues pertaining to genetic service providers including nurses, physicians and genetic counselors. One of these publications by Geller et al. is also listed below.

Barbara's publication raises incredibly thought-provoking issues related to the professional well-being of genetics health care providers, and will hopefully lead to more open discussions about how we can tackle some of these issues. For genetic counselors in particular, Barbara suggests that we can begin to overcome feelings of burnout and distress by more effectively educating other members of the healthcare team about what we as professionals can do for our patients. She envisions our ultimate professional goal as attaining the authority to apply those self-defined skills.

She also suggests that many of us complete graduate training with a very limited understanding of how we might fit into the healthcare team or be helpful to patients. Often, we measure our own success based upon our perception of a patient's increase in knowledge. In fact, she suggests that we may better serve our patients, and ourselves, by spending more time with them to work through their emotions.

From a professional perspective, Barbara suggests that feelings of collegial distrust that some genetic counselors experience relate to our discomfort with the different approaches to patient care among members of a medical team, particularly when those approaches are discordant with our own values. Additionally, she learned from ongoing interviews related to this work that it is often uncomfortable for genetic counselors to discuss their personal views if they differ from the mainstream views of the genetics community.

Barbara also notes that we as genetic counselors take our jobs very seriously and often take on the burden of care for our patients, feeling a sense of ownership and responsibility for their entire experience with the genetics team. Hopefully, recognizing this and trying to define our own roles more effectively, rather than having them defined for us, will allow us to improve the genetic counseling experience for patient and provider. Overall, Barbara has definitely given us a great springboard for further discussion and research into these issues.

Articles co-authored by genetic counselors from March 2009 to July 2009
(names of genetic counselors appear in bold)

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Please send references of published articles by genetic counselors to Deb McDermott at mcermottdeborah@yahoo.com

AEC Update

By Sarah Noblin, MS, CGC, 2009 AEC Chair and Shannan DeLany Dixon, MS, CGC, 2009 AEC Vice-Chair

Something for Everyone!

Check your mailboxes soon for your official conference booklet detailing the 28th NSGC Annual Education Conference (AEC) in Atlanta, Georgia. The NSGC membership has grown significantly in the number of members and the variety of professional environments in which we practice. As such, the AEC will feature a wide variety of content in the hopes of addressing the diverse educational and professional needs of our group.

A New Look to the AEC

As you have heard, the AEC has a new look that will debut in 2009. In response to the membership's desire to shorten the overall length of the AEC without cutting the number of Continuing Education Unit (CEU) opportunities, the overall conference format has been modified. First, the conference has been shortened to a total of three days. There will be six hours of plenary sessions, including the Janus series, two Beverly Rollnick lectures, and other sessions appropriate for a wide audience. There will be three Educational Breakout Sessions (EBS) with four offerings at each session. In the future, we hope to clearly identify an EBS as a beginner, intermediate or advanced level so there is something that appeals to everyone. As in 2008, we plan to have two concurrent paper sessions, for a total of three contact hours. This will allow you to attend more platform presentations by your colleagues. We are planning to close the conference with a late-breaking session.

Pre-Conference Symposia

Another exciting change for 2009 is the introduction of Pre-Conference Symposia, held on the day before the main conference. This year's session topics include genomic medicine, clinical teratology, mitochondrial disorders, and more. Each session will last 4 to 6 hours, allowing for a deeper review and discussion of the topics. We anticipate the attendance at each symposium will be smaller than at an EBS, which will allow for a more interactive experience. Each symposium will require registration separate from the AEC and will have limited space available, so sign up early!

Outreach in Atlanta

In an effort to reach out to the community of our local host city, the NSGC conducts an annual Outreach Event during the AEC. This year's event will involve reaching out to the conference hotel's employees to share with them the importance of knowing one's family's medical history. Stay tuned to www.nsgc.org for more details as they become available.

Prepare for Fun!

Atlanta – "City Lights, Southern Nights"
The NSGC is excited to host the AEC in Atlanta. Considered one of the most popular cities in the US, Atlanta boasts 8,000+ restaurants, is highly recommended for shopping and makes you feel right at home by giving the best in southern hospitality. Popular attractions include New World of Coca- Cola, Stone Mountain, Inside CNN Atlanta, and the Martin Luther King Jr. National Historic Site. For more information on exciting things to do while you are in Atlanta, please visit www.nsgc.org/conferences/aec.cfm.

Many Thanks

We would like to thank our conference subcommittee members – **Kimberly Banks**, **Erynn Gordon**, **Dawn Laney**, **Elizabeth McCracken**, **Kirsty McWalter**, **Courtney Sebold**, **Claire Singletary**, **Lucy Talbott Andrews** and **Kelly Taylor**. We owe them all a huge debt of gratitude. This conference is the result of the tireless efforts of this outstanding committee. Without each and every one of these individuals, this conference would not be possible. We would also like to thank **Ellen Schlenker** and **Jennifer Hoskovec**, Education Committee Chair and Vice-Chair, for their constant guidance during this transitional year. We look forward to seeing you in Atlanta!

Sarah Jane Noblin, 2009 AEC Chair (Sarah.J.Noblin@uth.tmc.edu)
Shannan DeLany Dixon, 2009 AEC Vice-Chair (SDelany@som.umaryland.edu)

Resources / Book Review

Reviewed by Ruth Abramson, PhD, FACMG

Psychiatric Genetics: Applications in Clinical Practice

Editors: Smoller, JW; Rosen Sheidley, B; and Tsuang, MT
Publisher: American Psychiatric Publishing, Inc., Washington, 2008
Pages: 323 $60.00 (paperback)

Psychiatric Genetics should be a welcome addition to the libraries of mental health, primary care, and genetic professionals. With increasing interest in genetics, in part as a result of the Human Genome Project, there has also been an increasing awareness of genetics in the popular press. This has led patients to ask questions about their personal and family genetic risks. They may have browsed the internet or seen the CDC and the Surgeon General campaigns for the "Family Health History Initiative". Curious patients are not only asking about family histories of Mendelian inherited medical problems, but also about common multifactorial conditions such as neuropsychiatric illnesses.

Psychiatric Genetics is divided into three sections. Chapter One introduces the reader to the range of neuropsychiatric disorders. Chapters Two and Three in the first section, entitled "Principles of Genetic Counseling and Risk Communication: Simple Tools to Foster Understanding," prove to be among the most helpful. Many health professionals may feel that all they need are the facts and, in turn, they relay these to their patients. Genetic counselors, however, are uniquely trained to provide more than just facts to patients. These two chapters provide a wealth of practical information including common indications for genetic counseling, components of a typical session, integrating genetic counseling into clinical practice, and risk communication. They also provide informative clinical cases to guide the process and demonstrate when to refer to genetics professionals.

Chapter Three highlights different risk communicating methods and identifies those that are most effective, which may be useful even for the experienced genetic counselor.

The second section provides practical information on the genetics and familial risk, clinical features, epidemiology, and implications of genetic counseling for neuropsychiatric disorders. The authors address childhood disorders, schizophrenia and psychotic disorders (such as schizophrenia, 22q deletion syndrome), mood and anxiety disorders, Alzheimer disease, and the neuropsychiatric symptoms associated with certain genetic disorders.

Chapter Four in the second section focuses on childhood disorders and reviews attention deficit hyperactivity disorder (ADHD), autistic disorder, obsessive compulsive disorder (OCD) and Tourette syndrome. It provides a starting point for the consideration of inherited behavioral disorders with childhood onset.

Chapter Eight in the second section deals with the neuropyschiatric symptoms of classic genetic disorders and highlights the behavioral symptoms of many most commonly referred for behavioral management. This chapter provides an excellent review of diagnostic clues and behavioral symptoms that, if found in a child in the absence of a medical genetic diagnosis, might raise suspicion that he/she should be referred for genetic evaluation.

The third section addresses perinatal psychiatry and teratogen issues, the ethical, legal and social implications of genetic research and counseling, and future issues that the practitioner may have to address.

Chapter Nine in the third section covers the management of psychiatric illness during pregnancy and postpartum, medication, teratogenic risks, neonatal symptoms and potential long-term neurobehavioral concerns. It provides information regarding pregnancy planning, management and postpartum risk for the patient with psychiatric illness and for the neonate.

Chapter Ten in the third section, focusing on ethical and social issues, raises questions about the complex issues of genetic testing for adults, premarital couples, prenatal patients, and children, pharmacogenetic profiling, screening potential adoptees, and discrimination. In particular, the topic of a physician's duty to warn regarding hereditary diseases is well written.

Finally, Chapter Eleven in the third section leads the reader to think about future issues such as the potential for more specific treatments once genes are identified, pharmacogenetics, personalized medicine, and gene-environment interaction. The burden to and the role of the clinician in providing genetic testing and counseling as risk evaluation becomes more complex is thoughtfully addressed.

Genetics is now more commonly taught in medical schools. However, the number of medical schools that thoroughly address the genetics of psychiatric disorders, syndromic neuropsychiatric behavioral features, pregnancy issues, how to communicate and when to refer is lacking. Psychiatric Genetics is a well written, easily read and valuable resource for both the mental health and general medical professional, as both groups commonly encounter neuropsychiatric disorders. I highly recommend Psychiatric Genetics to all clinicians as it fills a void with a much needed resource.

Media Watch

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

(names of genetic counselors or relevant organizations appear in bold)

February 27, 2009 – *MedlinePlus* (medlineplus.gov)
"Men with BRCA Gene Mutations Unaware of Cancer Risks"
This article reported the findings of a study published in the *Journal of Genetic Counseling* that men who are part of families with *BRCA1* or *BRCA2* mutations typically avoid genetic counseling or testing. Men who do receive counseling or testing are less likely than women to pass on the information or to discuss it thoroughly with their family members. **Steve Keiles** said that some reasons men should consider genetic counseling include "having relatives who have had breast or ovarian cancer before age 40 and/or who tested positive for genetic mutations."

March 12, 2009 – *MSNBC (msnbc.com)*
"Tempted by an at-home gene test?"
This article warned consumers about potential risks of at-home genetic susceptibility testing because the market has developed far more quickly than have rules to govern the tests or the science to guide the results. Consumers were encouraged to ask whether their privacy would be protected, if they could trust the results, and what the results would mean for their health before embarking on this kind of testing. For help understanding the findings of a genetic test, the article recommended locating a medical geneticist through the ACMG or **NSGC**. Websites were provided.

March 15, 2009 – *Voices of San Diego (voiceofsandiego.org*)
"Me and My Genome"
In this article about the benefits and limitations of at-home genetic susceptibility testing, **Elissa Levin** was quoted about how to look at one's genetic risks in a different context. Instead of worrying about a 10% risk for colon cancer, "This is saying there's a 90 percent risk that you won't ever have colon cancer in your life… Some people might walk away and say that's pretty good."

March 16, 2009 – *Southtown Star (southtownstar.com)*
"Gehrig patient hopeful about genetic research"
This article concerned a woman with two first degree relatives with amyotrophic lateral sclerosis (ALS, or Lou Gehrig disease), who just received her own diagnosis of ALS. **Sandra Donkervoort,** a genetic counselor involved with the research that identified a second gene for ALS, was quoted about the importance of this research for future treatment by saying, "We hope that by learning more about the underlying disease mechanisms that we will come up with targets that will provide measures for intervention."

April 9, 2009 – *MSNBC (msnbc.com)*
"Do-it-yourself DNA testing: Helpful or harmful?"
This article explored the pros and cons of DNA analysis, "once the exclusive domain of doctors and genetic counselors, now a do-it-yourself proposition, with several dozen companies marketing tests directly to consumers." The article recommended ordering tests through companies that offer genetic counseling and avoiding tests from companies without strict privacy policies. **Caroline Lieber** is quoted as saying, "Using your information without your consent is unethical."

April 9, 2009 – *BBC News (bbcnews.com)*
"Living with a 'faulty gene'"
This article tells the story of various family members deciding to undergo genetic testing for arrhythmogenic right ventricular cardiomyopathy. Members of the family were aided in considering the issues surrounding testing by **Colleen Brown**.

April 15, 2009 – *American Academy of Physician Assistant News* (aapa.org)
"The genetic solution"
This article helped physician assistants (PAs) learn how to handle the challenge they will face when patients start to ask for help interpreting their at-home genetic test reports. While discussing her limited ability to understand a sample report, one PA commented "I would refer [the patient] to a genetic counselor." **Erynn Gordon** was quoted extensively, including, "To counsel your patient, you have to know how to incorporate multiple factors such as what symptoms the patient's having, what their lab results look like, what does the genetic DNA test say, what does the family history show… Knowing when to refer a patient to a genetic counselor really depends on the patient, how comfortable you are with genetics, and how well you think the genetic counselor can answer the patient's questions."

Research Network

By Suzanna Schott, MS, CGC

Accuracy and Reproducibility of an Ashkenazi Jewish Panel Study
The Victor Center for Jewish Genetic Diseases is recruiting subjects for a study of accuracy and reproducibility of an Ashkenazi Jewish genetic testing panel. Participants must be over eighteen years old and a known carrier for Tay-Sachs disease, Canavan disease, Familial dysautonomia, Fanconi Anemia -Type C, Gaucher disease, Niemann-Pick disease type A, Mucolipidosis type IV, or Bloom syndrome. Participants must provide a blood sample with pre-paid mail kits provided by the researchers. All participants will receive a $25 Best Buy gift card as a token of appreciation.

Contact: Faye Shapiro at **215-456-3565**, or email shapirof@einstein.edu

Clinical and Molecular Characterization of RET Proto-Oncogene Sequence Variation
ARUP Molecular Genetics Laboratory is conducting a study to determine the clinical significance of novel or uncertain RET sequence variations. Eligible participants include individuals with poorly-characterized RET mutations, an unusual MEN2A phenotype, or earlier than previously reported age of onset of medullary thyroid cancer, pheochromocytoma or hyperparathyroidism. Participation involves DNA samples and phenotypic information from affected and non-affected family members.

Contact: Patti Krautscheid at **800-242-2787 Ext. 3439**, or email patti.krautscheid@aruplab.com

Clinical and Molecular Evaluations in Glycogen Storage Disease Type IX
Duke University Medical Center, Division of Pediatric Medical Genetics, under the direction of Priya Kishnani, MD, and Deeksha Bali, PhD, is investigating the symptoms and clinical course of glycogen storage disease Type IX (GSD IX). The study includes collecting clinical information for natural history studies, testing for genetic changes, evaluating the utility of biomarkers to track disease severity, and determining muscle, liver, heart, and other organ involvement in specific subtypes of GSD IX.

Contact: Jennifer Goldstein at **919-684-0626**, or email golds018@mc.duke.edu, or Stephanie Austin at **919-668-1347**, or email Stephanie.Austin@duke.edu

Genetic Basis of Inherited Reproductive Disorders
Massachusetts General Hospital is conducting a study to learn about the hereditary basis of disorders including early puberty, late puberty, complete absence of puberty (Kallmann syndrome, hypogonadotropic hypogonadism), and normal puberty that is accompanied by abnormalities of the reproductive system later in life. Participation requires a blood sample, family history review, medical questionnaire, and a scratch & sniff test to examine sense of smell. Study participation does not require a visit to Massachusetts General Hospital and a study package can be mailed to eligible participants.

Contact: Margaret Au at **617-726-5526**, or email ReproEndoGenetics@partners.org

Genetic Epidemiology of Lung Cancer
A consortium of six centers is collecting familial lung cancer cases for both traditional linkage analysis and for genome-wide association studies. Participants must have three to four lung cancer

cases on one side of the family, with at least one lung cancer case being alive and two to three cases of lung cancer on one side of the family, with at least one living affected for a blood sample collection.

Contact: Colette Glynn Gaba at **419-251-8086**, or email colette.gaba@utoledo.edu

Management of Myelomeningocele Study (MOMS)
The Management of Myelomeningocele Study (MOMS) compares prenatal surgery to standard postnatal surgery for spina bifida. Participants must be age eighteen years or older, residents of the U.S., enrolled by twenty-five weeks gestation, have a Body Mass Index of less than 35, and a fetus with myelomeningocele defect starting between T1 and S1, Chiari II malformation, and normal chromosomes. Participants are assigned to one of three MOMS Centers: The Children's Hospital of Philadelphia, the Vanderbilt University Medical Center in Nashville, and the University of California San Francisco.

Contact: Jessica Ratay at **866-275-6667,** or email moms@bsc.gwu.edu, www.spinabifidamoms.com

Prader-Willi Syndrome and Early-onset Morbid Obesity Natural History Study
University of California - Irvine Medical Center is conducting a study on the natural history of Prader-Willi syndrome (PWS) from birth to adulthood. Individuals from birth to sixty years of age with a confirmed diagnosis of PWS are eligible, regardless of race/ethnic background, sex, or growth hormone status. Participation includes an interview, physical exam, blood draw, bone scan, intelligence and behavior tests. The study may continue for up to five years with yearly appointments prior to sixteen years of age, and biannual appointments after sixteen years of age.

Contact: Virginia Kimonis at **714-456-5791**, or email vkimonis@uci.edu

Risk Factors for Psychosis in 22q11.2 Deletion Syndrome
Researchers at Duke University are studying the links between learning disabilities, behavior and emotional problems, brain structure, and hereditary variants in children (ages nine to fourteen) with Velocardiofacial syndrome or 22q11 deletion syndrome. Participation in this study involves a three-part assessment consisting of psychological testing (including neurocognitive and IQ/achievement testing), magnetic resonance imaging (MRI), and genotyping of genes in the 22q11 region. A fee of $100.00 is paid to families participating in the study.

Contact: Kelly Schoch at **919-681-2772**, or email orkelly.schoch@duke.edu

Trial of Losartan in Adults with Marfan Syndrome
Brigham and Women's Hospital and Children's Hospital in Boston is researching the effects of Losartan and Atenolol on the stiffness of the heart and aorta of people with Marfan syndrome. Participants must be over age eighteen years with no history of aortic dissection or aortic surgery. The trial involves a non-invasive ultrasound of the heart and the aorta before and after six months of treatment with Losartan or Atenolol.

Contact: Ami B. Bhatt, MD at **617-732-6320**

Variability in Gene Expression in Neurofibromatosis Type 1
The National Human Genome Research Institute is recruiting subjects with Neurofibromatosis type 1 (NF1) for a study examining the variability in phenotype between families and among family members. Eligible participants must have a diagnosis of NF1 and be at least sixteen years old. Study participants will undergo a two-day evaluation which will include a genetics evaluation, MRI

of the spine, echocardiography, dental evaluation, photography, and a blood draw. Family members will also be invited to participate. All testing and evaluations are free of charge and travel support is available.

Contact: Sarah Coombes at **301-451-9145**, or email coombessl@mail.nih.gov

Please send Research Network items to Suzanna Schott at sschott@cpdhealth.com

In Memoriam

By Elizabeth (Betsy) Gettig, MS, CGC

Editors' Note: *While this section is typically reserved for NSGC members, Betsy Gettig's heartfelt tribute following the untimely passing of Dr. George Tiller certainly merits an exception. Dr. Tiller was a steadfast supporter of the NSGC and a continual presence at our Annual Education Conferences. For years, he even helped cover printing costs for Perspectives when it was in paper format.*

Dr. George Richard Tiller
August 8, 1941 – May 31, 2009

> "Whatever you do, you need courage. Whatever course you decide upon, there is always someone to tell you you are wrong. There are always difficulties arising, which tempt you to believe that your critics are right. To map out a course of action and follow it to the end requires some of the same courage which a soldier needs."
>
> Ralph Waldo Emerson

By now you've heard the tragic news: Dr. George Tiller, a Wichita, Kansas-based physician who provided late-term abortions, was shot and killed at his church on the morning of Sunday, May 31, 2009. Dr. Tiller was a long-time target of anti-abortion activists; in previous attacks he was shot and his clinic, Women's Health Care Services, was bombed. Yet, at age 67, he was still providing services to women.

In a statement, Dr. Tiller's family said, "George dedicated his life to providing women with high-quality heath care despite frequent threats and violence. We ask that he be remembered as a good husband, father and grandfather and a dedicated servant on behalf of the rights of women everywhere." Indeed, that sums up Dr. Tiller eloquently.

Dr. Tiller leaves his beautiful wife of forty-five years, Jeanne, four children and ten grandchildren. Our thoughts and prayers remain with his family and friends. We remember him for his courage and service to women's health. Today and every day, genetic counselors, doctors, nurses and other caregivers go to work to provide health care services to women in clinics and hospitals, around the country and the world. Let us pledge that they will have our continued support, and that we will continue to speak out as advocates and citizens for womens' safe and secure access to reproductive health care.

One of Dr. Tiller's many mottos was, "Attitude is everything" and words could not more fittingly apply to this terrible event. The fight to keep abortion services available to women must go on. Dr. Tiller lived his life dedicated to women and families who unexpectedly found their pregnancies involving devastating health problems. He wished to assist these families and comfort them in the most critical moments of their lives.

Another motto he had was, "Trust women." Decision-making is not an easy process, and Dr. Tiller never sought to influence reproductive decisions. He worked to protect a woman's right to choose. He was the healthcare provider on the scene after a couple made the difficult decision about their pregnancy. If they wanted or needed his services, they were available to them. These included counseling, support groups targeted to spouses and patients, discussing sibling care, and keeping accurate data on the safety of treatment and management at his clinic.

Dr. Tiller was a former Navy flight surgeon who planned to become a dermatologist. His parents, sister and brother-in-law were killed in a plane crash, after which he adopted his nephew, the only child of his sister. He then took over his father's medical practice in conservative Wichita. Later, he found that his father began secretly performing then-illegal abortions, after a patient on whom he refused to perform one died.

Dr. Tiller began performing legal abortions, specializing in the one percent of them performed in the late second and third trimesters. These were often for women who discovered their fetus had anomalies or a confirmed genetic disorder. He also provided terminations to women with healthy pregnancies, where the woman had been previously raped or whose health was in danger.

Most doctors will not terminate a fetus after 21 weeks, so women from all over the world came to Wichita to access Dr. Tiller's services after learning their babies would be born with abnormalities. The clinic provided private transportation to patients who were often harassed by anti-abortion protesters at the airport or their hotels. The clinic offered grief counseling, funeral viewings, cremations and even postmortem baptisms. Dr. Tiller wanted the experience of losing a wanted, yet abnormal, pregnancy to be handled with respect and dignity.

Dr. Tiller spent thousands of dollars on his own legal defense, and also in support of legislation throughout the nation to protect a woman's right to choose. He refused to be intimidated even after being shot in 1993. He often stated, "What I am doing is legal, what I am doing is moral, what I am doing is ethical." He remained committed to the provision of services to women and persistent about doing his work.

I remember calling Dr. Tiller's office in 1993 after learning he had been shot. I was told, "Doctor is in surgery." I panicked until the staff clarified – Dr. Tiller was in his surgery performing procedures. He provided services to women and he was not going to stop, even if he was shot.

Dr. Tiller always believed in the court system and that justice would prevail. Throughout his life, he was investigated by two separate grand juries. He was investigated by the District Attorney's office at the behest of protesters, and even faced criminal charges this past year. In the end, both grand juries found he had done absolutely nothing wrong, as did the local District Attorney's office.

In March of 2009, a group of citizens, by petition, organized a grand jury to investigate Dr. Tiller. The grand jury subsequently subpoenaed the medical records of thousands of his patients. When that happened, patients came forward wanting to help Dr. Tiller. However, they not want their confidentiality breached and wished to preserve the confidentiality of their medical records. Dr. Tiller was incredibly committed to protecting the confidentiality, safety, health and well-being of

his patients. Several attorneys represented the women who were trying to block the turning over of their medical records to the grand jury.

Ultimately, justice was served. The Kansas Supreme Court quashed the subpoenas that were in place, substantially limited them, and allowed only very limited information (none of the patients' names or identifying information) to be turned over to the grand jury. It similarly found that he was not guilty of any of the charges laid against him.

Dr. Tiller never wavered in his support of women and was a remarkable man, husband and father. He will be missed. Women now – today – have fewer options if they are told the devastating news that their baby is seriously ill.

I will leave you with Dr. Tiller's words, which should resonate with the genetic counseling community: "Women are spiritually, morally and intellectually capable of struggling with complex ethical decisions, and arriving at the correct decision for themselves and their family." How true, and how tragic that one with such a generous philosophy has been taken from us. He will be missed.