Perspectives in Genetic Counseling - Volume 30, Number 4

## President's Beat

### Approaching Genetics Education Through a Primary Care Lens

*A 25-year old woman schedules an appointment with her primary care provider for her annual physical examination. During the appointment, the patient reveals that her 30-year old sister was just diagnosed with breast cancer. The physician knows that her mother was diagnosed with the disease over 20 years ago at the age of 35.*

I asked **Dr. Frederick Chen, MD, MPH, FAAFP and Chair of the American Academy of Family Physicians' Commission on Science Subcommittee on Genomics (AAFP-SOG)**, to describe how a typical family physician might manage this case.

"Given the amount of interest and media attention on BRCA and other genetic factors in breast cancer, the average family physician will clearly be alert to the possibility of 'something genetic' happening in this young woman's family. The first reaction of many physicians would be to increase her usual breast cancer surveillance – perhaps ordering a mammogram right away and reviewing the importance of breast self-exam. The next questions, in order, would be whether this patient needs to be tested for BRCA, how to obtain testing and what to do with the results.

Most family physicians will not be familiar with specific risk criteria for women who should be tested for BRCA. They may either find this information on their own or refer this patient to a genetics clinic or genetic counselor, if there is one available. If the patient is referred to a genetic counselor, the physician would certainly appreciate a copy of the consultation with specific recommendations for how the patient and physician should proceed. A more challenging situation exists when no genetic counselor is available. In these areas, physicians may or may not offer BRCA testing, and it is not at all clear who interprets the test results and how follow-up care is arranged.

Clearly, all of these decisions need to be made with the patient, who is well aware of her family history. Discussions of risk, whether to consider prophylactic mastectomy and familial and societal implications can all ensue from the initial visit. Family physicians, in general, are well-equipped to have these kinds of difficult discussions, as they are faced with similar situations in everything from prostate cancer screening to end-of-life decision-making. However, many may not feel comfortable with the specifics of genetic testing "and risk assessments and will likely rely upon genetic counselors to help patients understand the results of their genetic tests." -- *Frederick Chen, MD, MPH*

The passage above provides us with a view of genetic counseling through the primary care lens. This year, at AAFP-SOG, National Coalition for Health Professional Education in Genetics (NCHPEG) and National Human Genome Research Institute (NHGRI)/Physicians Assistant meetings, I had several opportunities to learn more about what it is like to look through this lens.

#### Physician Preferences

I learned that family practice physicians looking for information about genetics generally do not want to hear hour-long talks. However, many family physicians have expressed that it would be helpful to have relevant genetic information incorporated into talks on healthcare issues (e.g., diabetes, obesity, cancer) that they encounter in every day practice. If a genetics curriculum is going to be developed, they would like to be involved from the very beginning rather than be handed a finished product that has no primary care input, and they favor a case-based approach. I also learned that family practice physicians value shared decision-making over non-directiveness, except in those situations where nondirective counseling is warranted (e.g., reproductive decision-making). Some family physicians view this as a philosophical difference between primary care practitioners and genetic counselors whereas, in reality, our approaches may be more similar than they realize.

#### Looking Beyond Ourselves

This information may not be new to some of you. In fact, there is an excellent article entitled, "Genetics in primary care: A USA faculty development initiative," that discusses these points and others.[1] However, as we increasingly are being asked, individually and as a professional society, to educate primary care providers in order to facilitate the integration of genomics into clinical practice, it is essential for all of us to recognize the factors that can increase our effectiveness as educators. If we do not take these factors into consideration, tools like our new Health Care Providers section of the NSGC website and the Speakers' Bureau will not be fully

utilized. More importantly, we may not be seen as the “go to” providers for genetic information and genetic counseling.

As delineated in initiative No. 3 of our strategic plan, the NSGC is committed to “position genetic counselors as key players in the integration of genetics across the healthcare spectrum.” We believe that enhancing our members’ abilities to provide targeted educational outreach to other health care providers and generating opportunities to do so is a vital component of this process. We already are beginning to work with AAFP, NHGRI and NCHPEG in pursuit of this goal. We hope that you will participate in these efforts as opportunities develop over the next few years.

Angela Trepanier, NSGC President 2008

1. Burke W, Acheson L, Botkin J et al. Genetics in primary care: A USA faculty development initiative. *Community Genetics*. 5:128-146. 2002.

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## For Your Practice

### The Impact of a Career Ladder in Genetic Counseling on Professional Development

*By Erin Miller, MS, CGC and Nancy Steinberg Warren, MS, CGC* [a,b]

[a] Division of Human Genetics, Cincinnati Children’s Hospital Medical Center (CCHMC), Cincinnati, OH
[b] Genetic Counseling Program, University of Cincinnati, Cincinnati, OH

Over ten years ago, several reports[1,2] noted the emergence of genetic counseling career ladders, especially in large genetic centers like ours at Cincinnati Children’s Hospital Medical Center (CCHMC). Optimism abounded about the anticipated utility of the career ladder to formalize professional development pathways and tangibly recognize accomplishments of genetic counselors. The 2006 Professional Status Survey (PSS)[3] reports 12 genetic counselor job classifications, suggesting a growing multi-level workforce. Yet the “opportunity for professional advancement” was reported by only 34.5% of PSS respondents, and just 26.7% of PSS respondents were “satisfied with their earning potential.”

#### The CCHMC Model

We believe it is time to reflect on the impact of a career ladder. The CCHMC Human Resources department recognizes a three-tier career ladder for genetic counselors, where all three levels require a Master’s degree from an ABGC accredited training program.

*Genetic Counselor I:* Board eligible; previous work experience not required. Duties and responsibilities include aspects of clinical genetic counseling and maintaining active membership in national professional organizations.

*Genetic Counselor II:* Board certified; minimum of three years of experience (or full time equivalent) in genetic counseling. Position description includes duties and responsibilities of a GCI in addition to an active role in clinical genetic research, creation of national educational materials, development and coordination of clinical and/or lab programs, supervision of other staff and development of educational activities.

*Genetic Counselor III:* Board certified; minimum of six years of experience in genetic counseling. Expectations include all duties and responsibilities of GCI and GCII, in addition to taking an active leadership role in local, regional or national professional organizations.

**Assessing Pros and Cons**

The career ladder at CCHMC has facilitated recognition and advancement of genetic counselors. However, no model exists without limitations. The benefits and limitations of a career ladder are outlined below.

| Benefits | Limitations |
|---|---|
| Establishes baseline expectations | Could hold back counselors who exceed expectations |
| Outlines broad duties and responsibilities | Reduces incentive to focus duties and responsibilities in targeted areas |
| Eliminates need to define individual job positions | May be a proscriptive influence on job descriptions |
| Provides professional pathway | Creates hierarchy which could limit counselors' creativity and/or exploration of alternative professional pathways |
| Recognizes achieving board certification | Does not adequately address recertification |
| Provides structure to help counselors envision future opportunities | Structure can limit some counselors from acting on alternative or personal professional goals |
| Focuses on clinical roles | Not readily applicable to non-clinical positions |
| Ladder structure acknowledges years of experience | Does not explicitly acknowledge skill level or autonomy |
| Encourages productivity in new counselors | May not influence more experienced counselors' productivity |
| Provides a formal salary structure | Limits counselors' ability to negotiate for themselves on salary |
| Useful in a larger center with regular position turnover and/or growth | Requires a core number of counselors to be worth the effort |
| Designed for full time positions | Assessment of part time years of experience must be specified |
| Facilitates hiring of genetic counselors into non-genetics departments at the institution such as cardiology, neurology, internal medicine | May not apply to non-clinical positions |
| Grounded in institution's hiring structure to be applicable over time | Must be regularly re-evaluated |

**Ladder Evolution**

At CCHMC, we recognize that our career ladder is not static and needs to change with the times. As such, we have some work ahead of us. We intend to:

- reassess the ladder structure in light of the new annual ABGC board exam schedule
- suggest that recertification activities be clearly delineated
- adapt the current structure to accommodate a growing number of positions requiring unique skill sets (clinical trials, administrative, research or genetic laboratory settings, etc).

We share our perspectives on this career ladder to promote further discourse among genetic counselors, although institution-specific issues may apply. Developing a career ladder can raise awareness among administrators of the roles and contributions of genetic counselors. By engaging administrators, counselors will gain insight into "labor grades" or "band levels" and other aspects of the institution's administrative structure.

When striving to develop a career ladder, be patient but persistent. It could take a year or two to develop a model that is agreeable to counselors and administrators and to gain institutional approval. The outcome, though, is worth it. Career ladders can enhance opportunities for professional development and satisfaction with earning potential needed to retain genetic counselors in the profession.

References:

1. Warren NS and Hoechstetter L. Creating a career ladder. *Perspectives in Genetic Counseling*. 21:2;5. Summer 1999.
2. Hoechstetter Land Warren NS. Genetic counseling career ladders: Maturation of a profession. *Journal of Genetic Counseling*. 7:6;507-8. December 1998.
3. Parrott S and DelVecchio M. Professional Status Survey 2006. National Society of Genetic Counselors, February 2007.

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## SIG Speak

# Fetal Intervention and Therapy: New Opportunities for Genetic Counselors

*By Betsy Peach MS, CGC (2007-2008 FIT SIG Co-Chair), Lori Dobson MS, CGC (2007- 2009 FIT SIG Co-Chair) and Martha Dudek MS, CGC (2009-2011 FIT SIG Co–Chair)*

Fetal intervention and therapy options are becoming increasingly available. The types of conditions for which there are treatment, and the various treatment options for each condition, continues to evolve at a rapid pace. Interventions now include ultrasound guided therapies, fetoscopic procedures, open procedures and Ex Utero Intrapartum Treatment (EXIT) deliveries.

As awareness of treatment grows among families and providers, so does demand. Many conditions applicable for fetal intervention provide new opportunities for genetic counselors to specialize. The standard information expected from general prenatal counselors also has changed to include fetal MRI, 3D/4D imaging and specific fetal echocardiography techniques. Genetic counselors may need to facilitate patients' referrals for services, present options regarding fetal intervention, explain imaging findings and possible etiologies, obtain information regarding outcomes in the prenatal and postnatal periods after fetal intervention and provide psychosocial support throughout the process.

### Help for Previously Untreatable Conditions

Many fetal interventions are now available for conditions that were once considered untreatable. For example, the most common fetal treatment is currently for Twin-to-Twin Transfusion Syndrome. Previously, families with this diagnosis were provided a plan for expectant management. Now families have the options of amniocentesis reduction, laser ablation or radio frequency ablation. Options must be weighed with the clinical factors of each case, including the psychological impacts and potential risks and benefits.

Another interesting example is lower urinary tract obstruction, or bladder outlet obstruction. In this condition, the most common cause is posterior urethral valves (PUV) in males. PUV results from a piece of tissue that does not resolve naturally in development, causing a blockage at the posterior end of the urethra and leading to a backup of fluid in the bladder. The typical result is a huge bladder, enlarged cortical kidneys and anhydramnios threatening lung development. Treatment traditionally involves a shunt to redirect the fluid from the bladder into the amniotic cavity. Even with treatment, approximately 50% of babies need renal transplants in their lifetime.

Today, PUV options include fetoscopic valve ablation and open fetal surgery to create a vesicostomy that is then repaired postnatally. Both procedures have only been performed a handful of times, but the expectation is a better preservation of renal function and better long-term outcome. In light of the newness of the procedure, counseling families can be difficult, and knowing the experience of the centers offering the procedure is extremely important. The need for collaboration of research and outcome studies also is essential.

### When No Genetic Testing Exists

Another complexity of fetal intervention occurs when an underlying genetic syndrome may be present, but no prenatal testing exists for that condition. For example, in Congenital Diaphragmatic Hernia (CDH) with complex congenital heart disease, the karyotype may be normal, but a number of genetic syndromes may still be the cause, some possibly fatal and with a 25% recurrence risk. Another example is a fetus suspected of having Congenital High Airway Obstruction Syndrome (CHAOS). Although typically isolated and sporadic, CHAOS can be associated with Fraser syndrome, a rare recessive genetic condition with potentially lethal outcomes.

Both CDH and CHAOS have fetal intervention options that can improve the chances of survival. However, due to the underlying syndrome, the child would not be expected to live, regardless of the success of the fetal intervention. Thus, families are faced with very difficult decisions. Should they proceed with fetal intervention, accepting maternal and fetal risk in an attempt to save a baby who is not expected to survive long-term? The role of a genetic counselor and other specialists is crucial in assisting families understand the potential outcomes of the possible genetic syndromes.

### Ultrafast Fetal MRI

The genetic counselor also plays a critical role in helping patients understand the utility and results of new imaging technology. Ultrafast fetal MRI is a frequent adjunct to standard screening in the presence of multiple congenital anomalies, low or absent amniotic fluid and brain abnormalities. It also has become a necessity when considering fetal intervention. For example, in cases of CDH, lung volumes determined on MRI at 32 weeks gestation are important in assessing whether a baby may need an EXIT procedure or may do well with conventional delivery. Other diagnoses, such as Congenital Cystic Adenomatoid Malformation/ Bronchopulmonary Sequestration, teratomas, airway obstruction and bladder outlet obstruction, benefit from fetal MRI in determining the degree of the condition and planning appropriate treatment.

MRI can help clarify birth defects and underlying diagnoses that may not have been available without an examination or autopsy post delivery. Babies with oligohydramnios or anhydramnios used to need an infusion or post delivery exam. Now, fetal MRI can look at the internal structures in the paucity of amniotic fluid, which helps the family obtain information about recurrence risks in cases where the outcome is known.

We cannot overlook, though, the other side of fetal MRI. It is still a relatively new diagnostic tool, and there are many things that are not yet understood. Some prenatal findings may be well delineated in the pediatric or adult population as normal variations or having specific outcomes. However, in the presence of brain findings such as heterotopias, vermian hypoplasia and other non-specific findings, fetal MRI can complicate counseling, as there are limited prenatal/postnatal correlation studies to clarify the outcomes.

**New Technology Brings New Ethical Issues**

Fetal interventions have created many new psychosocial and ethical issues, such as how much risk is acceptable and for what conditions. How does one counsel a mother regarding placing her body at risk for the sake of saving an unborn child without knowing the long-term consequences? How does one counsel about potential outcomes based on prenatal imaging that has yet to be equated to postnatal outcome? When should one offer fetal intervention -- when there is a 5% chance of an underlying genetic syndrome? 10%? 20%? What about when there is a confirmed diagnosis -- should these patients be offered the same fetal intervention options as pregnancies that have normal karyotypes? These are only some of the questions that have been considered when dealing with cases involving fetal intervention.

**Availability of FIT**

As with any new technology, knowledge of its availability and usefulness determines the uptake. In smaller practices (i.e. private prenatal groups) or rural areas, fetal imaging and fetal intervention techniques may not be available, and prenatal specialists may have limited exposure to the technological advances. Additionally, centers for fetal intervention are being established at a rapid pace, and individual centers have different areas of expertise. Prenatal genetic counselors must stay on top of the current options for fetal intervention to provide their patients and medical colleagues the information needed to make appropriate referrals and decisions.

**Welcome to the Fetal Intervention and Therapy SIG!**

So many questions remain to be answered and so many challenging counseling issues to be explored. Genetic counselors are on the front line of this field and need a forum for discussing the issues, a place to go with questions and a reference point from which to start when asking about available options.

In 2007, the Fetal Intervention and Therapy (FIT) SIG was founded by genetic counselors specializing in these new technologies. We review cases, share resources and make contact with other genetic counselors in this unique field. Each of us comes from a different center where there are different opportunities in fetal intervention and different areas of specialization. We at the FIT SIG want to be a resource for genetic counselors who may want to learn about this field and who may need support.

The goals of the FIT SIG include:

- maintain a list of current fetal intervention and diagnostic options
- act as a resource for questions about fetal interventions and diagnostic options
- provide educational materials and opportunities to allow all genetic counselors equal access to knowledge about fetal interventions
- create opportunities for research across multiple centers and jobs.

**FIT SIG Activities**

In our first year we wanted to get a better feel for what information genetic counselors needed and determine the number of genetic counselors involved in fetal intervention and therapy. This past fall, the FIT SIG surveyed the

NSGC Prenatal SIG and FIT SIG members about their involvement in fetal intervention and therapy. The FIT SIG plans to publish this data and use it to guide future projects.

The FIT SIG also created handouts specific to fetal intervention and therapy for an Educational Breakout Session at the 2007 NSGC Annual Education Conference. These documents are available to the membership on the NSGC webpage:

- Fetal Therapy/Intervention Glossary
- Fetal Therapy/Intervention Guidelines

The FIT SIG is currently planning for 2009. Visit the NSGC website for contact information for the SIG Co-Chairs, **Lori Dobson** and **Martha Dudek**, as well as updated information regarding future activities.

Fetal intervention and therapy is an amazing opportunity for genetic counselors to expand their role with families and increase visibility within the medical community. Think about becoming a part of the FIT SIG in 2009!

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## Editor's Farewell

*By Jessica Mandell, MS, CGC, PGC Editor*

*Hot off the presses.* That is how I summarize my past five years as Editor for *Perspectives in Genetic Counseling*. I recall reading *PGC* in graduate school in 1996. Every month I looked forward to seeing the gray, stapled cardstock newsletter in my mailbox and reading the stories and quotes to connect with the vast genetic counseling community. I felt "up-to-date" and "in-the loop." I thought of the counselors behind *PGC* who got to deliver the news and, in turn, spark ideas and bring people together. Now my turn delivering the news has come and gone, and I have enjoyed every minute.

Aside from delivering the news, I also call these last years *hot off the presses* because I have felt the heat fueling the profession of genetic counseling with every idea, e-mail and article that has crossed my desk. I have been privileged to watch news develop and to help NSGC leaders and genetic counseling colleagues formalize and finesse their prose to make the greatest impact.

I also have witnessed great changes within NSGC and *PGC*. Back in 1996, NSGC was run by **Bea Leopold**, our one-woman-show executive director, conference organizer and PR maven. Cancer was the "nontraditional" specialty of choice and prenatal screening consisted of only three markers. There was no licensure, no HIPAA (or GINA), no genetic Internet companies, no e-blasts from our Board of Directors. *PGC* itself had no color, had a set number of pages and had limited column space. As our scientific field has matured, so have our professional foundations and our means of communication.

I credit **Janice Berliner** with the foresight and tenacity to bring *PGC* into the computer age. Just as I was starting my editorship, the change to an online format brought graphics and photos as well as immediate access to information, added efficiency via a streamlined contents page and linked articles, and the option of a printable PDF for those who still like the feel of paper between their fingers.

In addition, within *PGC* I sought content to reflect the breadth of our profession, the depth of our passions and the variety of our skills and interests. E-mail replaced the need for *PGC* to communicate general NSCG business, leaving room for more pinpointed features: an in-depth series on metabolic diseases in response to new newborn screening requirements; an exploration of the feats of counselors pioneering genetic services in countries like Saudi Arabia; recognition of colleague heroes who helped to house as well as counsel families devastated by Hurricane Katrina; and minute by minute accounts of efforts to lobby for GINA on Capital Hill. New departments, like GC Publications, SIG Speak and Licensure Update, also reflect on our expanded activities and allow us to appreciate the efforts of hardworking individuals.

I am proud to leave *PGC* with these accomplishments, and I am grateful to the team of individuals who have helped make *PGC* happen: Bea Leopold and Janice Berliner, my *PGC* mentors; **Anne Greb**, desktop publisher during the online transition; the staff at Smith Bucklin, especially **Emily DiTommaso** who juggles every ball behind the scenes; my staff of committee members who keep me on my toes; and the endless NSGC members I have approached (and even bombarded) to write articles and who have almost always said yes!

In January of 2009, two NSGC members will take over the helm of *PGC*, and I am very excited by the fresh ideas and experiences they will bring. **Deepti Babu**, from the University of Alberta in Canada, has been appointed Editor, and **Kirsty McWalter**, from the Hawaii Department of Health in Honolulu, will be the Associate Editor, a new position at *PGC*. They are already busy collecting stories for issues to come, and they await your correspondence. Please contact them at **Deepti.Babu@capitalhealth.ca** and **kirsty@hawaiigenetics.org**.

*Hot off the presses*. I can hardly imagine where the next five years will take our profession. But I do know that I will continue to look forward to reading about it in *PGC*. I guess some things do not change.

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## NSGC News

### Meet Your 2009 NSGC Board of Directors

Below are the newly elected (*) and returning leaders for NSGC in 2009. New members of the Board of Directors take office on January 1. Your Board members have been elected to represent YOU – the membership – and they want to hear from you. Keep this list handy in the coming year to email your leadership with your questions, ideas and accomplishments.

**Executive Committee**
President: Steven Keiles, MS, CGC
President-elect: Elizabeth Kearney, MS, CGC*
Past President: Angela Trepanier, MS, CGC
Secretary/Treasurer: Peter J. Levonian, MS, CGC
Secretary/Treasurer-elect: Elizabeth A. Leeth, MS, CGC*
Executive Director (non-voting): Meghan Carey

**Directors-At-Large**
Fiona M. Field, MS, CGC
Brenda Finucane, MS, CGC
Karen Heller, MS, CGC
Joy Larsen Haidle, MS, CGC*
Caroline Lieber, MS, CGC*
Jennifer Sullivan, MS, CGC*
Deborah Wham, MS, CGC*

**Congratulations to the 2008 Leadership Award Recipients**

The NSGC Awards Workgroup would like to thank all members that nominated their colleagues during the 2008 Call for Award Nominations. We are pleased to announce the winners of the 2008 Natalie Weissberger Paul and Leadership Awards. Please congratulate the following individuals on their accomplishments!

Natalie Weissberger Paul Award: Nancy Callanan, MS, CGC
International Award: Clara Gaff, MS, CGC
New Leader: Jennifer Hoskovec, MS, CGC
Outstanding Volunteer: Leslie Cohen, MS, CGC
Outstanding Volunteer: Jennifer Farmer, MS, CGC
Strategic Leader: Dawn Allain, MS, CGC
Strategic Leader: Cheryl Scacheri, MS, CGC

Stay tuned for announcements regarding the 2009 Call for Award Nominations this Spring.

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## ABGC Updates

### A Change in the Recertification Period

*By the ABGC Board of Directors*

The American Board of Genetic Counseling (ABGC) recertification shows a commitment to lifelong learning and helps ensure that knowledge and skills are maintained in a rapidly evolving field. Recertification is becoming even more significant for licensing, professional advancement, employment opportunities, hospital credentialing and insurance reimbursement. After reviewing the standard recertification practices in other healthcare fields, it was determined that a recertification period of ten years was too long, so the ABGC Board of Directors decided to move to a five-year recertification period beginning in 2010.

**The New Recertification Schedule**

- For an ABGC diplomate with a time-limited certificate issued from 1996 through 2009, this certificate will expire on December 31 ten years from the year the most recent certification was awarded.
- For time-limited certificates issued in 2010 or later, the certificate will expire on December 31 five years from the year the most recent certification was awarded.
- If their recertification efforts are successful, diplomates will be issued a new time-limited certificate dated January 1 of the year following expiration of the current certificate.

For instance, genetic counselors that passed the 1999 exam were awarded ABGC certification on July 1, 1999. This ten-year certificate will expire on December 31, 2009. If their recertification is successful, a new five-year certificate will be issued January 1, 2010 and will expire December 31, 2014.

**Two Pathways Remain**

There are still two pathways for recertification: the Re-examination Pathway and the Continuing Education Pathway.

- Diplomates recertifying by examination will need to pass the ABGC certification examination prior to expiration of the current certificate and will be awarded a five-year certificate if successful.
- For the Continuing Education Pathway, diplomates will need an average of 2.5 continuing education credits per year. Diplomates certified or recertified in 1999 through 2009 hold ten-year certificates and are required to earn 25 continuing education credits during the ten-year period of their most recent certification to achieve recertification. Diplomates certified or recertified in 2010 or later will hold five-year certificates and are required to earn 12.5 continuing education credits during the five-year period of their most recent certification to achieve recertification.

See the table below for specifics regarding Continuing Education Unit (CEU) and Professional Activity Credit (PAC) requirements.

**Summary Table of CEU and PAC Requirements**

| Year of Most Recent Certification / Recertification | Certification Period | No. of CEUs Required Over Certification Period | Minimum No. Category 1 CEUs | Maximum No. PACs | Maximum No. Category 2 CEUs |
|---|---|---|---|---|---|
| **2010 or later** | 5 years | 12.5 | 7.5 | 2.5 | 5 |
| **1999-2009** | 10 years | 25 | 15 | 5 | 10 |

Voluntary recertification is strongly encouraged for genetic counselors certified by ABGC before 1996 or by the American Board of Medical Genetics (ABMG). Although these diplomates do not have a time-limited certification, recertification demonstrates an ongoing commitment to lifelong learning.

**Still Time to Gain Credits**

Currently, there are numerous opportunities to earn continuing education credits. For those who have already earned some CEUs and PACs, there may still be time to accumulate the remaining credits needed (25 total) to recertify by the 2009 recertification cycle deadline, December 31, 2009. Diplomates without time limited certification who will not have the required continuing education credits needed to recertify by December 31, 2009 can still participate in the ABGC Recertification Program in an upcoming year. Beginning January 1, 2010, a

diplomate seeking voluntary recertification will be required to submit 12.5 CEUs accrued within the previous five year period to apply for voluntary recertification.

Please remember to consult the ABGC website (**www.abgc.net**) on a regular basis for detailed and updated information. All instructions, deadlines and online forms are located at the website. If you have questions about any aspect of the recertification process, feel free to contact the ABGC Executive Office at 913-895-4617 or **info@abgc.net.**

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## Licensure Update

### The Tennessee Genetic Counselors Association – Four Years Going Strong

*By the 2008 TGCA Executive Committee*

*Top row: Martha Dudek (President 2008), Janet Ulm, Eric Fowler, Carrie Heuer, Peggy Kerper, Jill Pouncey, Diana Chambers; Bottom row: Debbie Pencarinha, Melinda Cohen, Tene Franklin, Kami Schneider, Courtney Rowe-Teeter (President-elect 2008)*

The Tennessee Genetic Counselors Association (TGCA) met at the 2008 NSGC Annual Education Conference in Los Angeles for the fourth year. Since the group formed in 2005, we have been successful in getting legislation passed in Tennessee to license genetic counselors. Tennessee is one of only four states in the country that recognize genetic counselors as licensed providers.

“It has been an amazing journey, and we are fortunate to have had support of our institutions and colleagues in the state to move the legislation through on the first session it was introduced,” said **Martha Dudek**, 2008 President for the TGCA.

**Genetic Counselors Needed**

The TGCA has 28 members. The group hopes that licensure will increase awareness of genetic counseling services and improve billing opportunities in Tennessee. This may allow the profession to grow to better suit the needs of Tennessee residents. “With over six million people residing in Tennessee, there is only one genetic counselor per 20,000 residents. We could use a few more!” said Martha.

**Executive Committee Changes**

The TGCA met in Los Angeles to review the licensure process and discuss the summer survey results of its members. The Executive Committee and regional volunteers conducted the informal survey to gather the opinions of the state genetic counselors about the mission and structure of the TGCA. It was decided that in 2009 the state would be divided into three regions (East, Middle and West) to form an Executive Committee of three members. Each representative will serve as president of the TGCA in his/her third year in office.

“Rotating the president position will allow more shared responsibility and recognition across the state,” said President-elect and East TN representative, **Courtney Rowe-Teeter**, who will roll on as President in 2009. **Carrie Heuer** was elected to serve a three-year term as the West TN representative. An election for the Middle TN representative between **Misti Williams** and **Julie Kaalberg** will be held in December. Both Misti and Julie served on the 2008 TGCA Executive Committee during this transition year.

Currently the TGCA membership is reviewing the bylaws proposed by the 2008 Executive Committee. This month the membership will hold a vote to ratify these bylaws. The bylaws outline the leadership and responsibilities of the TGCA and it members.

**2009 Tasks**

The main mission for the TGCA in 2009 will be to monitor the Tennessee licensure legislation and ensure that it is implemented by the Tennessee Board of Medical Examiners in the way it was intended.

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## Student Forum

### NSGC-Sponsored Internship Offers Exposure in a Unique Setting

*By Martha Dudek, MS, CGC*

*Tomoko Bessho, student, and Eric Rosenthal, PhD, ScM, Myriad Genetics*

In an effort to provide experiences in expanded roles for genetic counselors, the NSGC and Myriad Genetic Laboratories, Inc. partnered to offer three, seven-day student rotations during the summer of 2008. The rotations took place at Myriad's headquarters in Salt Lake City, Utah and offered in-depth experience in hereditary cancer and laboratory processes, including exposure to the multitude of ways genetic counselors contribute in a diagnostic laboratory setting.

The three students selected for this rotation were:

- Marjan Eshraghi from Brandeis University
- Tomoko Bessho from Ochanomizu University (Tokyo, Japan)
- Kelli Swan from Northwestern University.

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

*I have really enjoyed my time here and have learned a great deal about the different roles genetic counselors can play within a company and how genetic counseling skills can be used in a variety of settings. [I learned the most from] observing genetic counselors in the various roles here at Myriad - sales, marketing, regional support team – and having the opportunity to talk to them about their jobs and what led them to this company; also, observing the differences between an academic and commercial lab.*

*I was exposed to a variety of "non-traditional" genetic counseling roles, exactly as was the goal of the rotation. [The best part of my internship] was being able to spend time with people in different departments and learn their roles and how everything is connected in the company.*

*[It was great meeting] with a lot of genetic counselors, learning the genetic testing process at Myriad, observing how counselors [work] (chart reviewing, phone calls, emails) and learning about insurance issues.*

This joint internship between NSGC and Myriad will be offered again during the summer of 2009. NSGC is working to expand this program to include internship experiences at other organizations during 2009 and 2010. Please watch the NSGC website, **www.nsgc.org**, and the NSGC listserv in early 2009 for additional details and an application.

## Lobbying 101: Wayne State University Students Take a Stab at Grassroots Advocacy

*By Mary Nyhuis, BS and Kate Zellmer, BS, Wayne State University Genetic Counseling Program*

*Wayne State GC Class of 2009 Kelly Kennelly, Tiara Johnson, Abbey Putnam, Mary Nyhuis, Kate Zellmer, Preethi Premkumar, with Angela Trepanier (center) Program Director*

As a student, it is easy to get lost in all the work that has to be done. There are papers to write, tests to study for, and on top of all that, case preparation and research! However, as budding genetic counselors, we cannot forget that we have something to offer the community, and during our training we can begin to expand our knowledge and skills to reach above and beyond traditional clinical roles. Even in our first year of graduate education, we have a unique knowledge base that enables us to be advocates for our patients, our profession and ourselves.

This past June, our genetic counseling class was invited to participate in the March of Dimes Lobby Day 2008. We were unsure of what to expect, but we knew that we could bring a voice to the Michigan legislature and advocate for our patient population.

We ventured to the state capitol and were grouped with other volunteers from throughout Michigan. Each student had a different experience, colored by our fellow group members. Two of our personal stories are shared below.

***Mary's Story***

*The members of my group included men and women who had been involved with the March of Dimes for many years, and most were well placed in the community. I was intimidated at first, as I was at least ten years younger than the other four members of my group. I thought that I lacked experience and that I was not going to be a worthwhile contributor for the team.*

*We were given an introduction about the issues for which the March of Dimes was advocating: preventing premature birth and ensuring funding for metabolic formula. At that point in my training I had not counseled patients regarding either issue, but I had some knowledge from observing counseling sessions and from my course work. However, I decided that I would just observe and speak up if necessary. As we walked to the Senate offices, our group started to get to know each other, and I was asked about my background. As soon as I explained that I knew what metabolic formula was and why it was important, the group decided that I was going to do the majority of the talking. This was not what I expected, and I felt uncomfortable being appointed to such an important role. However, I pushed aside my fears and put on a confident façade. If they believed in and respected my view, then I knew that I had to step up to the plate; after all, that was the reason I was there.*

*In the first office we visited, we were able to sit down with the Senator and speak about the issues. I ended up doing the majority of the talking and felt more confident as the discussion progressed. I realized during that moment that I did not have to have ten years of experience volunteering or thousands of dollars to donate in order to make a difference; I just needed to utilize my expertise. My other group members were so impressed that I took the lead role at each of the other visits. They encouraged me to be confident in what I was saying and to believe that I could make a difference. As the day continued, I found myself more and more excited to be an advocate for the March of Dimes.*

*Later in the day, I also had the chance to pull Representatives off the floor of the House in order to speak with them about prematurity and metabolic formula. I had only a few minutes to make an impact and assert myself and the issues at hand. Both Representatives I spoke with were receptive to what I explained and were open to exploring the issues further. Lobbying was exhilarating, and I am excited to pursue similar activities in the future. This experience helped me to discover one of my strengths and passions, and now I look forward to joining the communications and public policy committees in the NSGC. I cannot wait to engage in more advocacy opportunities.*

***Kate's Story***

*I had a group that was less willing to trust and respect my perspectives and input. When we broke off into the groups, I felt intimidated by the age, gender and professions of those I was placed with. I realized that if I wanted to contribute, I would have to step outside of my comfort zone.*

*My first strategy to assert myself was to take the lead in the hallways when finding the offices of our representative and to introduce our mission to the office aides when we arrived. After the introductions, the rest of the group members barely said a word while our group leader talked about the geographical area the representative covered, the legislative process and taxes for higher-income residents. As our group leader was speaking solely about issues unrelated to the goal and mission of the March of Dimes, I decided that someone needed to redirect the conversation. I respectfully interrupted him and spoke about the relevant issues: Medicaid coverage for metabolic formula and reducing the risk of premature birth. The group members seemed impressed with my ability to focus our discussion to the issues at hand and looked to me to lead them back to the elevator and take us to the next office.*

*Once we arrived at our next location, the group leader again began speaking about irrelevant issues, such as taxes and hunting. For the second time, I redirected the conversation and explained our reason for being there. I felt frustrated with our group leader for having a personal agenda and with the other group members for not helping to keep us on task. However, taking a stance in our group paid off because when I began speaking about the March of Dimes, the legislative aide looked directly at me and listened. At times it was an extremely difficult situation to navigate, but it taught me valuable lessons in how to respectfully communicate and take initiative within a group and how to advocate for particular important issues.*

While our group situations were different, both helped us to overcome our anxieties about our limited experience in grassroots advocacy and to develop communication skills that may transfer into other aspects of our careers. Helping the March of Dimes was an excellent opportunity in learning how to work with legislators, advocate for our patients and educate others about the profession of genetic counseling. We encourage all students to gain experience in advocating for pertinent issues and to have the confidence to bring our unique skill set and knowledge to the public eye.

## Perspectives in Genetic Counseling - Volume 30, Number 4

## Genetic Counselor Publications

*By Deborah McDermott, MS, CGC*

### Featured Papers:

**Hadley DW**, Jenkins JF, Steinberg SM, Liewehr D, Moller S, Martin JC, Calzone KA, Soballe PW, Kirsch I. Perceptions of cancer risks and predictors of colon and endometrial cancer screening in women undergoing genetic testing for Lynch syndrome. *J Clin Oncol*. 26(6): 948-954. 2008.

Shiloh S, Koehly L, Jenkins JF, Martin JC, **Hadley DW**. Monitoring coping style moderates emotional reactions to genetic testing for hereditary non-polyposis colorectal cancer: A longitudinal study. *Psychooncology*. 17(8):746-55. 2008.

Ashida S, Hadley DW, Vaughn BK, Kuhn NR, Jenkins JF, Koehly LM. The impact of familial environment on depression scores after genetic testing for cancer susceptibility. *Clin Genet*. Oct 2008.

Henry David Thoreau wrote, “None are so old as those who have outlived enthusiasm.” If this is true, **Don Hadley** is a proverbial fountain of youth. Don is a clinical researcher in the Social Networks Methods Section of the Social and Behavioral Research Branch and a genetic counselor in the Office of the Clinical Director at the National Human Genome Research Institute (NHGRI).

After graduating from the UC Berkeley genetic counseling program in 1981, Don worked for 12 years as a clinical genetic counselor at the Wisconsin Clinical Genetics Services (WCGS) program at the University of Wisconsin in Madison. After a short time in prenatal genetics, he worked for nearly a decade in the pediatric and adult clinical genetics programs. Don developed a particular interest in neurogenetics, most notably Huntington disease (HD). He was especially drawn in by visits from Marjorie Guthrie, wife of folk singer Woody Guthrie who died of HD, when she came to offer inspiration to patients and physicians and promote HD research.

Additionally, Don was actively involved in genetics outreach programs throughout the state of Wisconsin, including programs for Native Americans. He considers his colleagues, Drs. Renata Laxova, Raymond Kessel and Rich Pauli, as truly being ahead of their time in developing genetics education programs that reached out to teachers, special educators, occupational and physical therapists, audiologists and faith-based communities supporting those with hereditary diseases or children born with genetic conditions. Among his many experiences in Wisconsin, Don recalls receiving numerous “pink slips” warning of potential layoffs. Pink slips resulted from uncertain funding through a patchwork of state and federal block grants, hospital and targeted disease-funding sources. Fortunately, Don’s and other genetic counselors’ jobs were somehow always salvaged due to the tremendous efforts of the leadership at WCGS.

In 1993, Don was recruited to the NIH to what was then the National Center for Human Genome Research. He speaks of his move to the NIH as “incredible good fortune.” He worked with **Barb Biesecker** and **Barbara Bernhardt** for the first five years to develop the graduate program in genetic counseling, a joint venture between NHGRI and Johns Hopkins University. Simultaneous to the program’s development, he began creating his own research niche, aimed at identifying the psychological and behavioral outcomes associated with genetic counseling and the option of genetic testing. He hoped this focus would eventually allow for the development of more effective approaches to providing genetic counseling services. His work has most notably involved families with Lynch syndrome, formerly known as HNPCC.

His current research examines the significant role of the family in influencing an individual’s knowledge, attitudes, psychological wellbeing and behaviors related to genetic testing. The ultimate goal of his work is to develop family-based interventions that facilitate the adaptation of individuals/families to genetic risk information and

maximize their use of preventative behaviors. He currently is working on several manuscripts examining the impact of cascade genetic testing in relatives that present for genetic services over time, as the genetic risk information works its way through families.

His work has been published in many professional journals, which he hopes will stimulate practitioners to think more about the myriad issues involved in the genetic counseling and testing process.

Don continues to provide clinical services through the Office of the Clinical Director of NHGRI. He provides education and counseling to people participating in NHGRI clinical protocols for an array of diseases including Holoprosencephaly, Muenke syndrome, Familial Presenile Dementia with Neuronal Inclusion Bodies and other familial neurogenetic disorders. In addition, he participates with other genetic counselors (**Ann C.M. Smith, Flavia Facio** and **Barb Biesecker**), geneticists and students to provide medical genetics consult services to individuals and families participating in NIH protocols.

Don stresses the importance of genetic counselors being "at the table" during the development of public policy, public health initiatives, clinical services and medical and nursing training programs, to provide clinical insight that other specialists may not have. He makes it a point to share clinical experiences with his non-clinician colleagues (psychologists, anthropologists, communication specialists and epidemiologists) who are conducting research in the burgeoning field of genomics. He stresses how our unique training and insight as genetic counselors can shape and appropriately focus the research that our colleagues are doing, even if we are not doing the research ourselves.

**Articles Co-Authored by Genetic Counselors September-November, 2008**

(names of genetic counselors appear in bold)

Aina-Mumuney A, **Wood ED, Corson VL**, Stetten G, Jari S, Boehm CD, Blakemore KJ. Clinical consequences of an increasing trend of preferential use of cultured villi for molecular diagnosis by CVS. *Prenat Diagn.* 28:332-334. 2008.

Geifman-Holtzman O, **Ober Berman J**. Prenatal diagnosis: Update on invasive versus noninvasive fetal diagnostic testing from maternal blood. *Expert Rev Mol Diagn*. 8:727-51. 2008.

**Lee K, Williams B**, Roza K, **Ferguson H**, David K, Eddleman K, Stone J, Edelmann L, Richard G, Gelb B, Kornreich R. PTPN11 analysis for the prenatal diagnosis of Noonan syndrome in fetuses with abnormal ultrasound findings. *Clin Genet.* 2008 Aug 26. [Epub ahead of print]

**McIlvried DE, Prucka SK, Herbst M, Barger C**, Robin NH. The use of role-play to enhance medical student understanding of genetic counseling. *Genet Med.* 10:739-44. 2008.

Moore AF, Jablonski KA, McAteer JB, Saxena R, **Pollin TI**, Franks PW, Hanson RL, Shuldiner AR, Knowler WC, Altshuler D, Florez JC; Diabetes Prevention Program Research Group. Extension of type 2 diabetes genome-wide association scan results in the diabetes prevention program. *Diabetes.* 57:2503-10. 2008.

**Prucka SK, McIlvried DE, Korf BR**. Cancer risk assessment and the genetic counseling process: Using hereditary breast and ovarian cancer as an example. *Med Princ Pract*. 17:173-89. 2008.

**Quinn D**, Voyer Lavigne S, Chambers C, Wolfe L, Chipman H, Cragan J, Rasmussen S. Addressing concerns of pregnant and lactating women after the 2005 hurricanes. MCN, *Amer J of Mat/Child Nurs.* 33:235-41. 2008.

Rampersaud E, Mitchell BD, **Pollin TI**, Fu M, Shen H, O'Connell JR, Ducharme JL, Hines S, Sack P, Naglieri R, Shuldiner AR, Snitker S. Physical activity and the association of common FTO gene variants with body mass index and obesity. *Arch Intern Med.* 168:1791-7. 2008.

Rampersaud E, Mitchell BD, Naj AC, **Pollin TI**. Investigating parent of origin effects in studies of type 2 diabetes and obesity. *Curr Diabetes Rev.* 4:329-39. 2008.

**Reis LM**, Tyler RC, Abdul-Rahman O, Trapane P, Wallerstein R, Broome D, Hoffman J, Khan A, **Paradiso C**, Ron N, **Bergner A**, Semina EV. Mutation analysis of B3GALTL in Peters Plus syndrome. *Am J Med Genet A*. 146A:2603-10. 2008.

Streeten EA, McBride D, Puffenberger E, Hoffman ME, **Pollin TI**, Donnelly P, Sack P, Morton H. Osteoporosis-pseudoglioma syndrome: Description of 9 new cases and beneficial response to bisphosphonates. *Bone.* 43:584-90. 2008.

Streeten EA, Beck TJ, O'Connell JR, Rampersand E, McBride DJ, Takala SL, **Pollin TI**, Uusi-Rasi K, Mitchell BD, Shuldiner AR. Autosome-wide linkage analysis of hip structural phenotypes in the Old Order Amish. *Bone*. 43:607-12. 2008.

**Wood E**, Dowey S, Saul D, Cain C, Rossiter J, Blakemore K, Stetten G. Prenatal diagnosis of mosaic trisomy 8q studied by ultrasound, cytogenetics and array-CGH. *Am J Med Genet*. 146A:764-769. 2008.

*Please send references of published articles written by genetic counselors to Deb McDermott at **dam2001@med.cornell.edu**.*

## Perspectives in Genetic Counseling - Volume 30, Number 4

## AEC Update

### Save the Date for the 2009 AEC in Pittsburgh, PA

*By Sarah Noblin, 2009 AEC Chair and Shannan Delaney Dixon, 2009 AEC Vice-Chair*


We are already thinking about the 28th NSGC Annual Education Conference (AEC) in Pittsburgh, Pennsylvania. Save the dates: **November 12-15, 2009**.

The AEC will be held at the Hilton Pittsburgh, which is about 25 minutes from the Pittsburgh International Airport. NSGC attendees will receive complimentary Fitness Center access at the hotel. The Hilton Pittsburgh is adjacent to Point State Park which features a fountain, a riverfront promenade and a bike trail. The hotel is also within walking distance to PNC Park (home of the Pittsburgh Pirates), Heinz Field (home of the Pittsburgh Steelers), Majestic Star Casino, theatres, museums and shopping.

**A New Look to the AEC**

The AEC has a new look, which will debut in 2009. In response to the membership's desire to shorten the overall length of the AEC without cutting the number of CEU opportunities, the AEC will now begin with the Student and New Attendee Orientation followed by the Welcome Reception on Thursday evening. There will be two full days of outstanding educational opportunities within the plenary and educational breakout sessions (EBS) on Friday and Saturday, followed by a shorter day on Sunday with the conference concluding in mid-afternoon.

**Call for Speakers – Submit Today!**

NSGC is actively inviting members to submit presentation proposals for **Plenary Sessions, Educational Breakout Sessions and the Pre-Conference Symposia.**

We are seeking informative and stimulating presentations by genetic counselors, physicians, researchers and other industry leaders that will help advance our knowledge within the profession of genetic counseling. **The deadline for submission is January 14, 2009**. We will notify all applicants of their acceptance in February.

**Submission Guidelines**

Submissions need to contain a brief descriptive paragraph outlining the presentation, as well as three learning objectives written to the continuing education standards, outlined on the electronic submission form. A Plenary Session typically is one hour, while an EBS typically is two hours. Presentations may be provided by more than one speaker and encompass two or more integrally related topics. A Pre-Conference Symposia will be a minimum

of four hours but can last as long as six hours. All presentations must be educational in nature and not include any sales, product or marketing information. Speakers are encouraged to prepare and present original material. Members of the 2009 AEC Planning Subcommittee will carefully review all submissions.

***Please note:*** If you are submitting the proposal but do not plan to speak, list yourself as the "Primary Author/Speaker" and note in the Program Description who the speaker(s) will be. You still will be considered the main contact for the proposal.

Submit your proposal by completing the online submission form. Instructions on how to submit your proposal and the submission form can be found at the following link: **www.nsgc.org/conferences/speakers.cfm**. The online process was very successful in 2008, so we are using it again for 2009. Any questions can be directed to the AEC Chair and Vice-Chair, or **nsgcadministration@nsgc.org**.

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

**The 2009 AEC Committee**

Please contact your committee members with ideas, comments and suggestions.

AEC Chair
Sarah Jane Noblin, **sarah.j.noblin@uth.tmc.edu**

AEC Vice-Chair
Shannan Delany-Dixon, **sdelany@som.umaryland.edu**

AEC Planning Subcommittee
Kimberly Banks, **Kimberly.banks@stjoe.org**
Erynn Gordon, **egordon@coriell.org**
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Lucy Talbott Andrews, **lucy.andrews@perkinelmer.com**
Kelly Taylor, **ktaylor@chgr.mc.vanderbilt.edu**

## AEC Evolution – New Changes to Better Meet Your Needs

*By Ellen Schlenker, MS, Education Committee Chair and Jennifer Hoskovec, MS, Education Committee Vice-Chair*

As our profession continues to grow, it is imperative that we continue to evaluate our educational offerings. To that end, the Board of Directors appointed an Education Task Force that met in the summer of 2007. This task force evaluated many of our activities, including the AEC.

**An Improved Format**

Based on recommendations from this task force, we are making several changes to the format for next year's AEC. First, the overall conference length will be reduced to a total of three days. There will be six hours of plenary sessions, including the Janus series, the Beverly Rollnick lecture and other sessions appropriate for a wide audience. There will be three Educational Breakout sessions with four offerings at each session. In the future, we hope to be able to clearly identify breakout sessions as beginner, intermediate or advanced 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 also are planning to have a late breaking research session to close the conference.

**Pre-Conference Symposia**

Another exciting change for 2009 is the introduction of pre-conference symposia on the day before the main conference. These will be high level or in-depth sessions for specific specialty practice areas, new issues in genetics and genomics or professional development topics. Each session will last four to six hours, allowing for a deeper dive into a particular topic for counselors with specific interests. We anticipate the attendance at each

symposium will be smaller than at the Educational Breakout sessions, which will allow for a more interactive experience. Each symposium will require separate registration from the AEC and will have limited space available. Sign up early!

**CEU Changes**

NSGC became an approved provider for CEUs earlier this year through the International Association for Continuing Education and Training (IACET), which is different from the organization used previously to obtain CEUs. Therefore, some of the requirements NSGC and its members must meet have changed. One new requirement is that we must document that people actually attended the sessions for which they are requesting CEUs. If you take a remote learning course, like the online course or the *JGC* CEU program, participation is documented by the administration of a quiz, which you must pass. For the AEC this year, the scantron forms that you filled out for each session are documentation of your attendance. Based on feedback from the 2008 conference, we may look into other ways to document attendance for future meetings.

Thank you again to everyone who worked so hard on the 2008 AEC, Education Committee and the Education Task Force. We can't wait to see you next year in Pittsburgh!

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## Book Review

### *Overtreated: Why Too Much Medicine Is Making Us Sicker and Poorer*

*Author:* Shannon Brownlee

*Publisher:* Bloomsbury USA; 1st Edition (September 18, 2007); 352 pages, $25.95 hardcover

*Reviewed by:* Vickie Venne, MS, CGC

I like to listen to mysteries as books on tape because of my bad habit of peeking at the last few pages to see who-dun-it. If you're like me and like to skip to the end, or if you're just too busy to read a whole book, at least read chapter ten of Shannon Brownlee's, *Overtreated: Why Too Much Medicine is Making us Sicker and Poorer*. The book, and this chapter in particular, provide concrete suggestions regarding how our health care system can be improved. It will take sacrifice and a new way of thinking, but it can be done. (Yes, it can!)

**Questions and Answers**

Brownlee was a medical journalist before becoming a senior fellow at the New American Foundation. She wrote this book to explore the question of why we can't seem to fix our U.S. health care system. She provides a readable and fascinating response that involves case studies supported by interviews and opinions from clinicians and health service researchers.

In the first nine chapters, she outlines issues that those of us in clinical service see regularly. Why is it that various regions of the country and different hospitals offer such diverse health care recommendations, even in the face of practice guidelines? Why are hospitals some of the most dangerous places to be, especially if you are sick? Genetics is cutting edge and some of our patients are desperate for cures – and at what cost?

In chapter ten, most of Brownlee's suggestions to fix the system involve streamlining the documentation – and encouraging a single electronic record for each person that all appropriate health care providers can access. After years of working to protect genetic information, I wonder how health care would improve if genetic test results were available to all the clinicians involved in providing care for our patients?

**Generalist vs. Specialist**

Brownlee doesn't tackle the rare conditions we see. She speaks of the value of the generalist over the specialist, and I immediately think of the patients who have a long and frustration medical journey before coming to the geneticist for a relatively straightforward diagnosis. She uses bone marrow transplant for breast cancer as an example of patient – and advocacy group – driven medicine that turned out not to work. That effort was just underway when I started in cancer genetics 15 years ago, and it was poignant to read that chapter and reflect on

friends who died after a miserable treatment regimen that cost huge amounts of money, as well as time that might have better been spent with family and friends. When does the science undermine the compassion?

**Direct-to-Market Effects**

Much of the information in this book is probably vaguely familiar. Brownlee references most of the content from news stories as well as peer reviewed medical literature. When all of these sources are pulled together, the book offers an amazing history of policies that haven't improved the system and an incredible indictment of how market-driven practices clearly harm the public. Her description of the process of the direct-to-market pharmaceutical campaigns kept me awake at night – but knowing that I wasn't going to ask my doctor for a pill to make me sleep!

I am writing this review the week that the *New England Journal of Medicine* article regarding statins for cardiovascular prevention is publicized, and I wonder if insurance companies will readily pay $1200 per year for this drug or approve it only after a year of unsuccessful smoking cessation or weight loss programs. There are many parallels in direct-to-consumer DNA testing efforts, with seemingly similar marketing forces at work. Genomic SNPs are an example. They may better define the efficacy of drug prescriptions and identify appropriate surgical candidates, but with genetic services often being the loss leader in our hospitals, what is our role as genetic healthcare providers to monitor the use of this testing in our broken system?

**The Next Challenge**

This is a book for all clinicians as well as your well-informed clients. Brownlee supports the concept of truly informed consent and would encourage our patients to ask the hard questions regarding the advantages of a particular new imaging technology or drug. You will not think of our medical "service" the same way after reading this book. The challenge now is to consider the role genetics – and genetic information – will have in the new health care system.

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## Public Eye

### Media Watch

**October 2008 – *Continental* in-flight magazine, "An Ounce of Prevention: DNA Testing Could Lead to Improved Health Down the Road"**
This short article profiled a woman who opted for BRCA testing after meeting with a genetic counselor, based on the family history of early breast cancer in the woman's mother and maternal grandmother. **Caroline Lieber** discussed possible outcomes of genetic testing and the reasons why testing should occur in the context of genetic counseling.

**October 1, 2008 – *Reno Gazette Journal*, Your Turn editorial column, "Genetic Testing Can Assess Cancer Risk"**
In honor of Breast Cancer Awareness Month, **Robbin Palmer** wrote this article about genetic testing for hereditary breast cancer. She noted Christina Applegate's decision to have bilateral mastectomies upon learning she has a BRCA mutation and acknowledged the PBS special, "In the Family," a personal documentary by filmmaker Joanna Rudnick about the decisions she faced as a BRCA mutation carrier.

**October 1, 2008 – MSN.com, Health and Fitness column, "The Easiest Hard Decision"**
A woman with a strong family history of breast and ovarian cancer underwent BRCA testing and, after being found to have a mutation, elected prophylactic mastectomies. Her insurance company initially denied coverage for the surgery but, with the help of a genetic counselor, the woman successfully obtained coverage. **Rachael Brandt** clarified what is meant when we say that a person has tested positive for BRCA1 or BRCA2.

**October 3, 2008 – Newsweek.com, "Why We Need to Talk Openly about Death: The Ultimate Homework Assignment"**
Following her father's death from leukemia, **Wendy Uhlmann** wrote this touching article about the need for our culture to become more knowledgeable about death and dying. She suggested that our society should learn words of comfort to acknowledge death, the types of words she and other genetic counselors use to offer condolences to clients experiencing various losses.

**October 23, 2008 – Yahoo news, "Adapting to Life with the Risk or Reality of Genetic Disease: Genetic Counselors Suggest Ways to Help Patients Cope"**

This article highlighted research presented at the recent NSGC AEC in Los Angeles by **Jaclyn Douyard, Katie Voss** and **Jessica Young Adcock**. Topics included how and when people with cystic fibrosis inform dating partners of their diagnosis, coping strategies of caregivers when providing care for a person with an autism spectrum disorder and whether and when to disclose a diagnosis of Huntington disease.

**October 25-26, 2008 – *The Wall Street Journal*, "The Toughest Test"**
A front-page article highlighted the experience of a couple whose pregnancy was diagnosed with autosomal recessive polycystic kidney disease (ARPKD). The wife and husband reached divergent decisions about termination vs. continuation of the pregnancy because they had little information about the disease. After the child was born, **Helga Toriello** established that ARPKD was not the correct diagnosis. A sidebar entitled "More Prenatal Testing Brings New Worries" mentioned several labs offering DNA microarray testing, as well as ACOG's 2007 recommendation that all women be offered maternal serum screening.

**November 17, 2008 – *Houston Chronicle*, "Family History Can Trump Breast Cancer Gene Test"**
Research presented at the American Association for Cancer Research meeting this week revealed a fourfold increase in the risk for breast cancer in women with a family history of breast cancer who tested negative for BRCA mutations. In other words, family history may be a stronger predictor of breast cancer risk than are BRCA test results. **Beth Peshkin** pointed out that "this is contrary to what I think the common perception is," as many women who test negative may be falsely reassured.

*Note: We welcome Claire Noll as our new co-editor of Media Watch along with Roxanne Ruzicka. Please send media items to **Claire.Noll@uth.tmc.edu** or **rruzicka@gmail.com.***

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## Research Network

### Discontinuation of Antidepressants in Pregnancy

The Organization of Teratology Information Specialists (OTIS) is investigating the effect of discontinuing antidepressants during pregnancy on maternal behavior and cognitive-behavioral development of infants. Eligible women must be at least 18 years of age and prior to 15 weeks gestation. Women with or without antidepressant use may participate. The study involves pre- and post-partum telephone interviews and cognitive developmental evaluation of the infant at 12 months of age.

***Contact:*** 1-877-875-7333 (toll free in Canada and the US), **medications.pregnancy@hotmail.com**

### Autoimmune Diseases in Pregnancy

OTIS is conducting a study on medications used to treat autoimmune diseases and their effect in pregnancy. Women who are less than 20 weeks gestation and have psoriasis, rheumatoid arthritis, ankylosing spondylitis or psoriatic arthritis are eligible. Participation involves phone interviews during pregnancy and post-partum pediatric exam at no cost.

***Contact:*** 1-877-311-8972 (toll free in Canada and the US), **raandpregnancy@ucsd.edu**

### DCIS and BRCA1/BRCA2 Cohort Study

Researchers at Yale University are following outcomes of ductal carcinoma in-situ (DCIS) in women with BRCA1 or BRCA2 gene mutations. Women over the age of 20 years whose first breast diagnosis was pure DCIS and have a mutation in BRCA1 or BRCA2 are eligible. Participation includes a telephone questionnaire, a saliva or blood sample used to look at changes in DNA and in some cases tissue specimen and mammogram review. This study is funded by the Susan G. Komen Breast Cancer Foundation.

***Contact:*** Collect call 203-764-9084, **charmila.fernandes@yale.edu**

### Families with Meningioma

Yale University, Brigham and Women's Hospital, Duke University, MD Anderson Cancer Center and the University of California San Francisco are conducting a study to identify genes associated with the development of

meningioma. Persons who are over the age of 20 years, have been diagnosed with meningioma and have at least one other relative who also has had meningioma are eligible. Participation involves a telephone questionnaire, a blood or mouthwash sample used to look at changes in DNA, review of tissue specimens and consent to contact family members.

***Contact:*** Collect call 203-764-8422 or 617-732-6826, **lisa.calvocoressi@yale.edu**

## Genetics of Autism Spectrum Disorders

The Developmental Medicine Center, Program in Genomics, Division of Genetics and Neurology Department at Children's Hospital–Boston are seeking children and adults diagnosed with an autism spectrum disorder (ASD) to be a part of a research study to understand the genetic and environmental factors that influence the development of ASD. Individuals 18 months of age or older with an ASD diagnosis are eligible to participate along with their parent(s) and other family members.

Participation involves completion of a medical/family history questionnaire via mail or telephone and medical records review. To investigate genes that may be important in ASD, a small blood and/or saliva sample is requested. The blood draw is arranged through the participant's physician or a certified clinical laboratory, and saliva samples are obtained through the mail. Travel to Boston is not required. There is no cost to participate.

***Contact:*** Carly Grant, Research Genetic Counselor, 617-355-9152, **grant@childrens.harvard.edu**

## Genetic Epidemiology of Pancreatic Cancer (PACGENE)

Researchers at Wayne State University, the Mayo Clinic, Johns Hopkins University, MD Anderson Cancer Center, Dana Farber Cancer Institute and the University of Toronto aim to map one or more pancreatic cancer susceptibility genes. They are currently enrolling families with at least two cases of pancreatic adenocarcinoma. Participation includes a phone interview, a mailed questionnaire, medical record review and analysis of a DNA sample. Families will not receive individual results. For more information visit **www.karmanos.org/cancer.asp?id=927&cid=19**.

***Contact:*** Kate Sargent, MS, CGC, Study Coordinator, 313-578-4240, **sargentk@med.wayne.edu**

## MOMS - Management of Myelomeningocele Study

The Management of Myelomeningocele Study (MOMS) is actively recruiting pregnant women to compare prenatal surgery vs. standard postnatal surgery for spina bifida. Screening begins by telephone and a review of medical records. Interested candidates who qualify are assigned to one of three MOMS Centers for a comprehensive evaluation: Children's Hospital of Philadelphia, Vanderbilt University Medical Center in Nashville and the University of California at San Francisco. Participants are randomized to the prenatal surgery or the postnatal surgery groups. Follow-up evaluations are performed on infants in both groups through two and a half years of age.

Minimum inclusion criteria for participants includes:

- age 18 or older
- a resident of the United States
- complete enrollment by 25 weeks of gestation
- a body mass index of less than 35

In addition, the fetus must have the following:

- myelomeningocele defect that starts between T1 and S1 (may extend below S1)
- hindbrain herniation (Chiari II malformation)
- normal chromosomes.

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## Letter to the Editor

### Personal Concept of Genetic Counseling Anagram

This creative and insightful anagram was created by **Ruth Turnbull**, BS, 2nd year genetic counseling graduate student at Sarah Lawrence College, as part of the class "Issues in Genetic Counseling."

**G** Getting detailed family and medical histories, and eliciting significant information

**E** Evolving, progressive field of human genetics

**N** Non-directive counseling and support

**E** Educating patients about the implications of and adjustments to their genetic disorders for their health and for their families

**T** Treating patients with respect, patience and compassion

**I** Including different health care professionals, when appropriate, such as geneticists, speech therapists, physiotherapists, psychologists

**C** Continuing education through seminars, conferences and talks

**C** Challenging ethical issues and psychosocial concerns

**O** Offering appropriate diagnostic testing to provide patients with as much information as they need to make choices about their health and future

**U** Unconditional positive regard and empathy

**N** Never judging or imposing your own belief or values on patients

**S** Supporting patients for differences in values, beliefs and family situations

**E** Explaining genetics, risk recurrence and implications of testing and results to patients in language they can understand

**L** Liaison between patient and the health care system

**I** Inclusive environment that encourages patients to ask questions

**N** Never the same situation twice; treating patients as people who are managing and coping with their conditions as individuals

**G** Genetic counseling for a wide variety of situations including prenatal, pediatric, cancer, adult, infertility, cardiac and metabolic diseases